### **Aerodigestive Disorders**

**ALTERNATIVE NAMES**

* Aerodigestive tract disorders
* Aerodigestive diseases
* Aerodigestive conditions
* Upper aerodigestive tract disorders
* Aerodigestive dysfunction
* Aerodigestive syndrome
* Aerodigestive pathology
* Oropharyngoesophageal disorders
* Pharyngolaryngeal disorders (when focusing on throat and voice box)
* Swallowing and airway disorders (emphasizing functional aspects)
* Pediatric aerodigestive disorders (when referring to children)

**DEFINITION AND DESCRIPTION**

**Aerodigestive disorders** are conditions or diseases of the aerodigestive tract—including the **airway** (pharynx and larynx), **pulmonary tract** (trachea, bronchi, and lungs), and **upper digestive tract** (esophagus)—that may affect respiratory and swallowing functions. Aerodigestive disorders, or the management of them (e.g. surgery, intubation), may result in voice, feeding, and/or swallowing problems as well as **laryngeal airway problems**—the term used in this Practice Portal page to refer to paradoxical vocal fold movement (PVFM) and chronic cough.

Aerodigestive disorders may be congenital, developmental, or acquired. They are not mutually exclusive individuals may have more than one disorder. In children, some aerodigestive disorders may resolve with maturity or with behavioral management, but others may require medical and/or surgical intervention.

Examples of common aerodigestive disorders (grouped by anatomical location) include, but are not limited to, the following:

**Airway (Pharynx and Larynx)**

* chronic cough
* dystussia
* epiglottitis
* fungal infections of the pharynx or the larynx (e.g., blastomycosis [rare], histoplasmosis, candidiasis)
* irritable larynx
* laryngeal clefts
* laryngeal or pharyngeal paralysis/paresis (unilateral or bilateral)
* laryngeal stenosis (supraglottic, glottic, or subglottic)
* laryngeal webs
* laryngomalacia (moderate to severe)
* laryngopharyngeal reflux
* laryngospasm
* muscle tension dysphagia
* muscle tension dysphonia
* PVFM
* breathing–swallowing incoordination (secondary to medical conditions such as chronic obstructive pulmonary disease (COPD), neurological insult, or head and neck cancer)
* structural or physiologic changes secondary to injury, neoplasms, radiation therapy, or surgery (e.g., pharyngeal or laryngeal resections, radiation for head and neck cancer treatment, prolonged intubation)
* velopharyngeal dysfunction

**Pulmonary Tract (Trachea, Bronchi, and Lungs)**

* bronchomalacia
* chronic lung disease (e.g., asthma, emphysema, COPD)
* infectious diseases affecting pulmonary function (e.g., upper respiratory infection, pertussis [whooping cough], pneumonia, tuberculosis)
* neoplasms
* structural or physiologic changes affecting pulmonary function secondary to injury or surgery (e.g., pneumothorax [collapsed lung], pneumonectomy, lung transplantation)
* tracheal stenosis
* tracheoesophageal fistula
* tracheomalacia

**Upper Digestive Tract (Esophagus)**

* esophageal motility disorder, including spasm and achalasia
* esophageal structural disorder, including stricture, web, diverticulum, and ring
* gastroesophageal reflux
* inflammatory forms of esophagitis (e.g., eosinophilic, viral, reflux-related)

### **Speech-Language Pathologist (SLP) Involvement in Aerodigestive Disorders**

Aerodigestive disorders can cause secondary problems in feeding, swallowing, voice, and/or laryngeal airway function. SLPs play a role in the screening, assessment, diagnosis, and treatment of these secondary problems and often work collaboratively with other professionals in providing services to individuals with aerodigestive disorders.

#### **Paradoxical Vocal Fold Movement (PVFM)**

**Paradoxical vocal fold movement, or PVFM,** is the intermittent, episodic adduction of the vocal folds during inspiration. During episodes, the vocal folds adduct partially or fully and restrict the passage of air to the lungs. PVFM can occur in isolation, or it can co-occur with other conditions, including pulmonary disorders (e.g., asthma), laryngeal abnormalities, and cardiac pathology

#### **Chronic Cough**

**Chronic cough** is most commonly defined as a cough lasting more than 8 weeks in adults and more than 4 weeks in children. Chronic cough may be termed “somatic cough syndrome” in the absence of a known cause or “tic cough” when it is accompanied by core clinical features of tics (e.g., suppressibility, distractibility, suggestibility, variability, and presence of a warning sensation; . SLP intervention is an effective treatment for chronic cough and addresses the management of cough regardless of the initiating cause.

#### [**Incidence and Prevalence**](https://www.asha.org/Practice-Portal/Clinical-Topics/Aerodigestive-Disorders/#collapse_1)

The **incidence** of a disorder or condition refers to the number of new cases identified in a specified time period. **Prevalence** refers to the number of individuals who are living with the disorder or condition in a given time period.

The following aerodigestive disorders provide a sample of conditions that can cause feeding, swallowing, voice, and/or laryngeal airway problems that may involve treatment by a speech-language pathologist (SLP). Some problems may be symptomatic or indicative of an aerodigestive disorder but do not involve treatment by an SLP. For example, dysphagia symptoms in individuals with esophageal dysmotility may resolve following medical and/or surgical intervention.

**Airway Disorders**

* **Laryngeal cleft** occurs in 1 out of 10,000–20,000 infants, with slightly higher rates noted in males with a ratio ranging from 1.2:1.0 to 1.8:1.0 . Approximately 41% of patients with laryngeal cleft were found to silently aspirate
* **Laryngomalacia** was estimated to affect approximately 1 in 2,600–3,100 newborns, and stridor was a reported symptom in the majority of children in the study. Estimates reported dysphagia to be present in 41%–50% of patients with laryngomalacia.

**Pulmonary Tract Disorders**

* **Chronic obstructive pulmonary disease (COPD)** was self-reported by 15.5 million adults in the United States (40.3 cases per 100,000 individuals). Higher prevalence was reported in rural areas An increase in compromised swallowing function was reported for adults with COPD over 60 years of age.
* **Paradoxical vocal fold movement (PVFM)** has been associated with chronic cough and asthma. True prevalence of PVFM is unknown due to inconsistent diagnostic criteria and a lack of awareness of the disorder. However, studies have reported PVFM as a subset of the investigated clinical population. For example, PVFM was estimated to occur in 2.5%–22% of emergency room patients presenting with shortness of breath or asthma.
  + Across the life span, reports estimated that PVFM is more common in females than in males, with a ratio ranging from 2:1 to 3:1.
  + Increased rates of PVFM were reported in elite athletes), in individuals with elevated stress, and in individuals exposed to irritants (e.g., reflux, allergens;

**Upper Digestive Tract Disorders**

* **Gastroesophageal reflux disease (GERD)** is associated with many aerodigestive disorders, including, but not limited to, laryngeal cleft, esophageal atresia, and eosinophilic esophagitis (EoE). Estimates of GERD in individuals with esophageal disorders ranged from 40.2% to 65.3% of patients. Voice disorders and dysphagia may also be associated with GERD. Voice disorders are reported to be 1.8 times higher in patients with GERD
* **Esophageal atresia/tracheoesophageal fistula** was estimated to affect 2.3 out of 100,000 live births in 2017. Prevalence rates of long-term dysphagia in individuals with esophageal atresia ranged from 18.2% to 84.2%. Prevalence of dysphagia decreases as children get older, but dysphagia has been commonly indicated in adults who had esophageal atresia repair as children
* **Eosinophilic Esophagitis (EoE)** was estimated to affect 71.1 out of 100,000 children and 25.9–55.5 out of 100,000 adults, with a prevalence rate significantly higher in adult men than in adult women (35.8 vs. 17.8 out of 100,000 persons, respectively). Relative to other races and ethnicities, EoE was highest in Caucasians. EoE was estimated to affect 18.6 out of 100,000 elderly persons .
  + Patients with EoE reported dysphagia as a common symptom. Dysphagia was reported in 46.2%–94.5% of adults. Estimates reported that 4.8%–60.9% of children with EoE experience dysphagia and globus sensation, but estimates may be as high as 88% .

Voice and swallowing problems commonly occur as a result of structural or physiologic changes to the aerodigestive tract secondary to surgery or radiation therapy. For example, the majority of individuals (70.5%) who received a lung transplant and subsequent swallowing evaluation showed laryngeal penetration or aspiration .

**Signs and Symptoms**

**Signs** are observations made by a third party (e.g., clinician or family member). For example, observations of coughing when someone swallows may be a sign of aspiration, and observations of changes in someone's vocal pitch may be a sign that the vocal folds are swollen or inflamed.

**Symptoms** are a person's own perception of changes in their swallowing, voice, breathing, or desire to eat or drink. Symptoms are usually described in terms of severity, location, frequency, and duration. For example, a person may notice that, recently, they have been coughing a great deal following exercise.

Signs and symptoms of aerodigestive disorders can vary depending on the specific disorder and the severity of the condition causing the disorder.

The following signs and symptoms are grouped by the function that can be affected:

**Feeding and Swallowing**

* aversion, disinterest, or refusal behaviors surrounding eating or drinking
* avoidance of certain foods and/or food characteristics
* avoidance of eating and drinking in public
* coughing or choking during or after eating
* globus sensation (feeling of something stuck in the pharynx)
* increased duration of feeding and/or mealtimes
* increased swallowing effort
* odynophagia (painful swallowing)
* pharyngonasal backflow (often referred to as “nasopharyngeal reflux”)
* poor weight gain in infants and children
* recurrent pulmonary infections, such as pneumonia
* regurgitation of swallowed food back into the pharynx or into the oral or nasal cavity
* slow or uncoordinated feeding in infants and children
* throat clearing during or after eating
* unexplained weight loss in adults or children
* wet breath sounds or wet vocal quality during or after eating

**Voice**

* aphonia (no voicing)
* breathiness
* increased vocal effort
* pain while voicing (odynophonia)
* rough vocal quality
* strained vocal quality
* vocal fatigue
* vocal pitch changes (e.g., in response to inflammation and edema)
* weak or inadequate vocal volume

**Respiration**

* chronic cough
* discoordinated or weak voluntary cough
* excessive mucous secretion
* excessive sputum production
* inability to manage oral and/or pharyngeal secretions independently
* increased effort of breathing
* overwhelming need to want to “take a breath”
* pneumonia
* rapid respiratory rate
* recurrent respiratory infections
* stridor (secondary to vocal fold paralysis or other airway obstruction; inspiratory or biphasic)
* weak reflexive cough
* wheezing

**Paradoxical Vocal Fold Movement (PVFM)**

* cough and rough vocal quality before or during an episode of vocal fold adduction
* difficulty inhaling, exhaling, or both (sole report of difficulty exhaling suggests asthma)
* lightheadedness that resolves quickly when trigger is removed
* stridor on inhalation (stridor for both inhalation and exhalation suggests laryngeal obstruction)
* sudden adduction of the vocal folds induced by triggering stimuli, such as activity, stress, or environmental irritants
* sudden and total loss of voice
* tightness in the throat

**Chronic Cough**

* productive (wet) or nonproductive (dry) cough
* cough lasting more than 8 weeks in adults and more than 4 weeks in children

#### **Causes**

There are many ways to categorize the causes of aerodigestive disorders, given the overlap of structures and functions involved.

This Practice Portal page uses the following categories:

**Congenital**

* embryologic origins, including incomplete or atypical development, innervation, structure, and function of the aerodigestive tract structures

**Structural/Anatomical**

* injury or surgery affecting airway, pulmonary, or digestive structure and function
* weakness or dysfunction of the upper esophageal sphincter, allowing for regurgitation of acidic content into the pharynx, larynx, or nasal airway
* weakness or dysfunction of the lower esophageal sphincter, allowing acidic stomach contents to reenter the esophagus

**Functional**

* emotional stressors, fear, and/or anxiety that contribute to increased muscle tension
* environmental irritants or exercise
* laryngeal hyperreactivity
* laryngotracheal hypo reactivity

**Other Medical Conditions**

* autonomic dysfunction (e.g., diabetic neuropathy, vasovagal syncope)
* cardiovascular, pulmonary, or neurological diseases, or cancer, leading to breathing–swallowing incoordination (e.g., congenital heart defects, meconium aspiration syndrome, chronic lung disease, cystic fibrosis, head and neck cancer, motor neuron disease)
* irregular, unsynchronized, inappropriate, or absent esophageal contractions causing motility problems
* neurological diseases affecting aerodigestive sensorimotor function (e.g., stroke, Parkinson's disease, prematurity, hypoxic ischemic encephalopathy, cerebral palsy, muscular dystrophy, myopathies)
* neurological problems that trigger coughing, laryngospasm, bronchial constriction, or long-term bronchial changes affecting lung function
* recovery from respiratory failure or aerodigestive disuse during periods of critical care due to the use of artificial airways

**Paradoxical Vocal Fold Movement (PVFM)**

The exact cause of PVFM is not known, although PVFM may be related to laryngeal hyperresponsiveness.

PVFM may be triggered by

* organic factors, such as gastroesophageal reflux or environmental irritants, or
* nonorganic factors, such as exercise or psychological stress.

**Chronic Cough**

* asthma syndrome
* esophageal diseases, such as gastroesophageal reflux
* idiopathic heightened cough response, particularly in females
* rhinitis and sinusitis
* postnasal drip
* use of angiotensin-converting enzyme inhibitors (medications for the treatment of high blood pressure and heart failure)

Assessment and treatment of aerodigestive disorders may require use of appropriate personal protective equipment.

Most aerodigestive disorders are identified by a physician on the basis of physical examination and one or more of the following:

* gastrointestinal evaluation (e.g., esophageal motility study; gastric emptying test; esophagogastroduodenoscopy; esophagram; esophageal manometry; 24-hour pH or impedance test; Raman spectroscopy)
* instrumental examinations (e.g., endoscopy; videofluoroscopy; airway fluoroscopy; flexible bronchoscopy; bronchoalveolar lavage; direct microlaryngoscopy; high-resolution pharyngeal manometry)
* pulmonary function tests
* X-ray and other imaging studies (e.g., chest X-ray; chest computed tomography scan; magnetic resonance imaging; electromyography; ultrasound)

Assessment of impairments caused by aerodigestive disorders often requires a multidisciplinary approach involving the speech-language pathologist (SLP) and other medical, surgical, and rehabilitation specialists. In collaboration with other health care specialists, the SLP provides expertise on feeding, swallowing, voice, and laryngeal airway problems related to aerodigestive disorders.

A multidisciplinary approach may include

* a team of medical and other professionals,
* team meetings,
* combined assessment procedures,
* care coordination, and
* follow-up clinic visits.

A core multidisciplinary team may include one or more of the following professionals:

* allergist
* anesthesiologist
* gastroenterologist
* nurse
* nurse practitioner
* oncologist
* otolaryngologist
* physician assistant
* primary care physician (pediatrician in the case of a child, geriatrician in the case of elderly patients)
* pulmonologist
* registered dietitian
* SLP

Depending on the age of the individual and the specific concerns, other team members may include the following:

* cardiologist
* coach/athletic trainer
* medical geneticist
* neurologist
* occupational therapist
* physical therapist
* psychologist
* radiologist
* respiratory therapist
* sleep specialist
* social worker or case manager
* sports medicine physician
* surgeon

### **Screening by an SLP**

An SLP may be the first to see an individual who is experiencing voice or swallowing problems. These individuals may or may not have an underlying aerodigestive disorder. The purpose of screening is to identify individuals who require further assessment by an SLP or referral for other professional services. Screening may uncover findings that suggest underlying medical problems.

It is important for SLPs to

* be familiar with anatomical structures affected by various aerodigestive disorders;
* be familiar with changes in feeding, swallowing, voice, and respiration problems that can be caused by aerodigestive disorders;
* recognize deviations in structure and function that warrant an aerodigestive evaluation by a physician; and
* make appropriate referrals, as needed.

SLPs screen for the following observed and reported changes:

* **Voice and respiration**
  + vocal quality (e.g., rough voice, strained voice)
  + vocal effort (e.g., vocal fatigue, report of pain while voicing)
  + presence of stridor or labored breathing that affects breath support for voicing
  + rapid respiratory rate
  + chronic cough
* **Swallowing and dietary changes**
  + clinical signs of feeding and swallowing problems (e.g., coughing, throat clearing, discomfort or globus sensation when swallowing
  + other indicators such as poor weight gain in infants and unintentional weight loss in adults, or purposeful avoidance of previously enjoyed liquids or foods

SLPs also look for signs of neurologic conditions (e.g., abnormal sensorimotor function) that can affect voice, swallowing, or respiration, or that signal an underlying medical condition.

If screening results indicate feeding, swallowing, or respiratory difficulties that suggest an underlying disease process, referral is made to an appropriate medical professional.

#### **Treatment**

Decisions about goals and treatment options are made in partnership with the person, their family/caregiver, and other caregiving professionals. As part of a multidisciplinary team (see the Assessment section above), the speech-language pathologist (SLP) may be involved in assessing the individual's response to medical treatment and in implementing both indirect and direct strategies during or following medical treatment.

Comprehensive multidisciplinary treatment of aerodigestive disorders may include

* medical management (including pharmacotherapy and/or surgery) of underlying causes;
* indirect or compensatory treatment via environmental, dietary, and lifestyle modification; and
* direct or restorative intervention via voice, swallowing, and/or laryngeal airway treatment by an SLP.

### **Medical Management**

**Medical management** decisions in aerodigestive disorders balance airway needs for breathing with optimal preservation of vocal quality and swallowing integrity (Dinwiddie, 2004). Approaches vary from “wait and watch” to complex surgical interventions.

Examples of medical approaches by appropriate medical professionals include, but are not limited to, the following:

* endoscopic treatment of structural abnormalities (e.g., dilation)
* medical or surgical management of the underlying disease/condition leading to the aerodigestive disorder
* surgical repair of structural abnormalities affecting aerodigestive function (e.g., arytenoidopexy, fundoplication, laryngeal cleft repair, supraglottoplasty, arytenoidectomy)

### **Dietary and Environmental Management**

**Dietary, compensatory, and environmental management** may include the following:

* Dietary changes, such as
  + implementing elimination diets,
  + conducting food challenges (systematic introduction of new foods or textures),
  + reducing acid-producing foods, and
  + increasing water intake to hydrate the vocal folds and to support healthy phonation.
* Compensatory changes, such as
  + using positional strategies while eating or drinking (e.g., elevating the head of the bed, turning the head) and
  + implementing maneuver-based strategies when eating and drinking (e.g., supraglottic swallow).
* Environmental management, such as
  + avoiding triggers (e.g., environmental pollutants, strenuous exercise).

### **Direct or Restorative Intervention**

SLPs provide **direct or restorative treatment** to address functional voice problems (including respiratory support for voicing) and feeding and swallowing problems. SLPs also provide direct treatment for laryngeal airway problems, including paradoxical vocal fold movement (PVFM), and chronic cough.

The nature, scope, and duration of SLP management depend on

* the underlying aerodigestive disorder, structures and functions affected, severity, and relevant history;
* the type and course of medications to treat underlying and co-occurring diseases; and
* the type and extent of surgical management required (e.g., surgical intervention and healing time, need for a temporary feeding tube).

### **Treatment Considerations for Pediatrics**

Treatment selection depends on the child's age, cognitive and physical abilities, and specific swallowing and feeding problems.

Infants and young children with aerodigestive disorders may benefit from alterations of liquid viscosity to improve airway protection during swallowing and/or to reduce the impact of reflux when tube feeding. This may include the use of natural foods or commercial dietary thickening agents to increase liquid viscosity. When making such recommendations, SLPs should consult with the medical team and be aware of the possible impact of thickening agents on nutritional status and overall health. For example, the addition of a thickener may alter the nutritional composition of the formula or breast milk. This may require the child to ingest more volume in order to obtain the necessary nutrients, or it may provide more than the recommended calories or the amount of certain nutrients (e.g., more than the recommended iron, if rice cereal is the thickening agent).

In addition, children with a history of necrotizing enterocolitis are advised to avoid gel-based thickeners containing the agent xanthan gum. Food allergies must also be considered when thickening agents are being considered.

**Precaution**

The U.S. Food and Drug Administration (FDA) has cautioned consumers about using commercial, gum-based thickeners for infants from birth to 1 year of age, especially when using the product to thicken breast milk. SLPs should be aware of these cautions and consult, as appropriate, with their facility to develop guidelines for using thickened liquids with infants. See FDA consumer cautions

### **Intervention for PVFM**

The goal of treatment is to establish consistent vocal fold abduction during the breathing cycle to maintain a patent airway. This reduces anxiety and affirms that breathing is consistently achievable, even in the presence of environmental or activity-related triggers.

Behavioral management by an SLP is the preferred treatment approach to PVFM Other disciplines may also be involved in treatment (e.g., medical intervention to treat reflux or allergy triggers, when present).

SLPs may implement the following procedures with most individuals with PVFM. Procedures are individualized based on triggers or other factors and include the following:

* **Relaxed throat breathing**—trains the vocal folds to abduct and remain abducted throughout the breathing cycle. Techniques include
  + sniffing in through the nose with the tongue relaxed on the floor of the mouth and the lips gently touching, followed by exhalation through pursed lips or the production of a strident sound such as /s/, an
  + sipping air in through pursed lips, followed by an exhalation through pursed lips or the production of a strident sound such as /s/.
* **Diaphragmatic/abdominal breathing**—trains attention to expansion of the lower rib cage and abdomen during inhalation to avoid clavicular breathing patterns and shoulder/neck tension.

Once the individual has identified their most effective breathing technique, the SLP may introduce challenges (triggers) while using the technique. These include the following:

* **Sports or exercise-specific training**—implementing breathing techniques during a routine exercise activity or competitive sports training
* **Training in the presence of environmental triggers** (if applicable)—implementing breathing techniques during exposure to odors or other environmental triggers, beginning with non-noxious stimuli and progressing through noxious stimuli

### **Intervention for Chronic Cough**

The goal of treatment is to help the individual manage their cough by identifying triggers, using strategies to suppress the cough, reducing laryngeal irritation, and using healthy vocal hygiene behaviors. Speech-language services should be coordinated with medical management of the underlying cause; services should be implemented after ruling out or addressing other contributing factors

Treatment activities include the following:

* **Educating the individual about chronic cough and its treatment,** including
  + discussing the difference between acute cough and chronic cough, emphasizing that chronic cough does not have physiological benefits;
  + establishing cough suppression as a safe and achievable goal;
  + defining the cough trigger threshold and desensitization of the cough response; and
  + emphasizing the importance of adhering to medications prescribed by physicians to manage cough.
* **Implementing healthy vocal hygiene practices** to maximize hydration and reduce irritation of the vocal folds, including helping the individual
  + identify behaviors that are contributing to the cough (e.g., poor hydration, mouth breathing) and
  + practice healthy vocal hygiene behaviors (e.g., drinking plenty of water and talking at moderate volume).
* **Teaching cough suppression strategies** (as appropriate), including
  + monitoring the cough precursor or trigger;
  + using relaxed throat breathing or prolonged, slow exhalation (see PVFM above);
  + using pursed-lip breathing; and
  + substituting coughing with other behaviors or distractions such as
    - sucking on ice or non-medicated candy and
    - swallowing dry or with sips of water.

The SLP typically introduces strategies without the presence of triggers to establish functional behaviors and to determine the person's most consistent response. The SLP may then introduce stimulants such as strong odors, increased activity levels, or other identified triggers to help the individual use the strategies before the “need” to cough. Treatment ends when the person can manage cough across a variety of contexts and in the presence of triggers.

## Aerodigestive Disorder Differential Diagnosis List

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| --- | --- |
| Category | Disorders / Conditions |
| Airway (Pharynx, Larynx) | Laryngomalacia, laryngeal cleft, laryngeal web, laryngeal stenosis, vocal cord paralysis/dysfunction, paradoxical vocal fold movement (PVFM), airway obstruction, respiratory papillomatosis, chronic croup, airway stenosis |
| Pulmonary Disorders | Aspiration pneumonia, bronchomalacia, bronchiectasis, chronic cough, asthma (including reflux-related), recurrent respiratory infections, lung collapse, lung abscess |
| Esophageal / Upper Digestive | Gastroesophageal reflux disease (GERD), extraesophageal reflux, eosinophilic esophagitis (EoE), esophageal achalasia, esophageal atresia, esophageal strictures, esophageal webs, esophageal diverticula, tracheoesophageal fistula (TEF), caustic ingestion injury, esophageal dysmotility, colonic interposition |
| Feeding and Swallowing | Dysphagia, cricopharyngeal dysfunction, feeding tube dependence, aspiration during swallowing, oral aversions |
| Other Related Conditions | Chronic otitis media, sinusitis, chronic laryngitis, hiatal hernia, caustic ingestion, foreign body aspiration (FBA) |

**Epidemiology of aerodigestive disorders**

Aerodigestive foreign bodies are a notable subset of aerodigestive disorders. In a six-year study at Rwanda Military Hospital, aerodigestive foreign bodies accounted for 0.74% of ENT consultations (290 cases out of 39,240 patients), predominantly affecting toddlers aged 1–3 years (50.34%). Males were more commonly affected (57.24%) than females (42.76%).

* Another study from Nigeria reported a hospital prevalence of 0.61% for aerodigestive foreign bodies, with the mean patient age around 17 years and a male-to-female ratio of approximately 2.24:1. The youngest patient was 3 months old, and the oldest was 80 years.
* Neonatal aerodigestive disorders are significant due to their impact on feeding and respiration, with a high potential for lifelong morbidity and healthcare costs. These disorders involve complex coordination of swallowing and breathing, critical for infant development.
* Aerodigestive cancers, particularly of the upper aerodigestive tract (larynx, pharynx, oral cavity), represent a significant epidemiological burden worldwide. Approximately 500,000 new cases of upper aerodigestive tract cancers occur annually worldwide, with a strong male predominance (up to 85%) and peak incidence between 40 and 60 years of age.
* Pediatric aerodigestive foreign bodies are most common in children aged 1 to 5 years, consistent with the toddler and preschool age groups being at highest risk for foreign body aspiration or ingestion.
* Aerodigestive disorders may present across all age groups but show distinct epidemiological patterns:
  + Children (especially toddlers and preschoolers) are most affected by foreign bodies and congenital/developmental aerodigestive anomalies.
  + Adults and older adults are more commonly affected by aerodigestive cancers and chronic inflammatory conditions.
  + Males tend to have a higher prevalence across many aerodigestive conditions, including foreign bodies and cancer

## **Doctor-Patient Conversation: Aerodigestive Disorder**

Doctor: Good morning! I understand you've been having some trouble with coughing and difficulty swallowing. Can you tell me more about your symptoms?

Patient: Yes, doctor. For the past few months, I've had a persistent cough, especially after eating or drinking. Sometimes, I feel like food gets stuck in my throat, and I even choke a little.

Doctor: I see. Have you noticed any changes in your voice or any wheezing or noisy breathing?

Patient: Actually, yes. My voice sounds a bit hoarse sometimes, and I occasionally hear a wheezing sound when I breathe.

Doctor: Thank you for sharing that. These symptoms suggest that there might be an issue involving both your airway and your swallowing function. Have you had any episodes of pneumonia or chest infections recently?

Patient: I had pneumonia about six months ago, and before that, I had a few chest infections over the last year.

Doctor: That information is important. Sometimes, problems with swallowing can cause food or liquids to accidentally enter the airway, leading to infections. We call this aspiration. To better understand what's going on, I recommend we perform some tests, including a swallowing study and possibly a flexible endoscopy to look at your throat and airway.

Patient: Will these tests be uncomfortable?

Doctor: The swallowing study is non-invasive and involves eating or drinking substances with a contrast material while we take X-rays. The flexible endoscopy involves passing a thin, flexible camera through your nose to look at your throat and voice box; it might cause some mild discomfort but is generally well tolerated.

Patient: Okay, that sounds reasonable. What treatments are available if something is found?

Doctor: Depending on the cause, treatments can range from swallowing therapy with a speech-language pathologist to medications for reflux or inflammation. In some cases, surgery might be necessary if there are structural problems.

Patient: Thank you, doctor. I’m glad we’re looking into this.

Doctor: You're welcome. We’ll work together to find the cause and the best way to help you feel better.

REFERENCES

<https://applications.emro.who.int/imemrf/Sudan_Med_Monit/Sudan_Med_Monit_2014_9_1_39_43.pdf>

<https://www.asha.org/practice-portal/clinical-topics/aerodigestive-disorders/>

**PULMONARY HYPERTENSION**

Alternative names and related terms for Pulmonary Hypertension (PH) include:

* Pulmonary arterial hypertension (PAH) — refers specifically to Group 1 PH involving the pulmonary arteries
* Pulmonary vascular hypertension
* Pulmonary artery hypertension
* Pulmonary pressure elevation
* Pulmonary arterial pressure increase
* Pulmonary hypertensive disease
* Secondary pulmonary hypertension — PH due to underlying conditions (Groups 2–5)
* Idiopathic pulmonary hypertension — when no known cause is identified
* Chronic thromboembolic pulmonary hypertension (CTEPH) — a distinct subtype caused by chronic clots
* Pulmonary hypertensive syndrome
* Pulmonary hypertension syndrome

**DEFINITION AND DSCRIPTION**

Pulmonary hypertension is a type of high blood pressure that affects the arteries in the lungs and the right side of the heart.

In one form of pulmonary hypertension, called pulmonary arterial hypertension (PAH), blood vessels in the lungs are narrowed, blocked or destroyed. The damage makes it hard for blood to move through the lungs. Blood pressure in the lung arteries goes up. The heart must work harder to pump blood through the lungs. The extra effort eventually causes the heart muscle to become weak and fail.

In some people, pulmonary hypertension slowly gets worse. It can be life-threatening. There's no cure for pulmonary hypertension. But treatments are available to help you feel better, live longer and improve your quality of life.

Pulmonary arterial hypertension strains the right side of your heart, which pumps oxygen-poor blood to your lungs. This strain can lead to right-sided heart failure.

Plus, PAH slows down blood flow between your heart and lungs. This means less blood can enter your lungs to gain fresh oxygen. As a result, blood flow to the rest of your body also slows down. So, your organs and tissues can’t get enough oxygen.

Without treatment, PAH can be fatal.

**Causes**

Pulmonary hypertension is caused by changes in the cells that line the lung arteries. The changes can make the artery walls narrow, stiff, swollen and thick. It gets harder for blood to flow through the lungs.

Pulmonary hypertension is sorted into five groups, depending on the cause.

### **Group 1: Pulmonary arterial hypertension (PAH)**

Causes include:

* Unknown cause, called idiopathic pulmonary arterial hypertension.
* Changes in a gene passed down through families, called heritable pulmonary arterial hypertension.
* Use of some medicines or illicit drugs, including methamphetamine.
* Heart condition present at birth, called a congenital heart defect.
* Other health conditions, including scleroderma, lupus and cirrhosis.

### **Group 2: Pulmonary hypertension caused by left-sided heart disease**

This is the most common form of pulmonary hypertension. Causes include:

* Left heart failure.
* Left-sided heart valve disease, including mitral valve or aortic valve disease.

### **Group 3: Pulmonary hypertension caused by lung disease**

Causes include:

* Scarring of the lungs, called pulmonary fibrosis.
* Chronic obstructive pulmonary disease, also called COPD.
* A sleep disorder in which breathing repeatedly stops and starts, called sleep apnea.
* Being at high altitudes for extended periods of time, if you are at high risk of pulmonary hypertension.

### **Group 4: Pulmonary hypertension caused by blockages in the pulmonary artery**

Causes include:

* Blood clots in the lungs that don't go away.
* Tumors that block the pulmonary artery.

### **Group 5: Pulmonary hypertension triggered by other health conditions**

Causes include:

* Blood disorders, including polycythemia vera and essential thrombocythemia.
* Inflammatory disorders such as sarcoidosis.
* Conditions that affect the body's ability to break down certain sugars, including glycogen storage disease.
* Kidney disease.

### **Eisenmenger syndrome and pulmonary hypertension**

Eisenmenger syndrome can lead to pulmonary hypertension.

Eisenmenger syndrome is a long-term complication of an unrepaired heart condition present at birth. An example is a large hole in the heart between the two lower heart chambers called a ventricular septal defect.

The unrepaired hole in the heart causes oxygen-rich blood to mix with oxygen-poor blood. The blood then goes to the lungs instead of going to the rest of the body. This increases pressure in the pulmonary arteries.

**Risk factors**

Pulmonary hypertension is usually seen in people ages 30 to 60. Growing older can increase the risk of developing Group 1 pulmonary hypertension, called pulmonary arterial hypertension (PAH). PAH from an unknown cause is more common in younger adults.

Other things that can raise the risk of pulmonary hypertension are:

* A family history of the condition.
* Being overweight.
* Smoking.
* Blood-clotting disorders or a family history of blood clots in the lungs.
* A history of being around asbestos.
* A heart condition present at birth, called a congenital heart defect.
* Living at an altitude of 8,000 feet (2,438 meters) or higher.
* Use of some medicines, including those used for weight loss.
* Illicit drugs such as cocaine or methamphetamine.

**Symptoms**

The symptoms of pulmonary hypertension develop slowly. You may not notice them for months or even years. Symptoms get worse as the disease continues.

Pulmonary hypertension symptoms include:

* Shortness of breath. It may first start during exercise and eventually happen at rest.
* Blue or gray skin. Depending on skin color, these changes may be harder or easier to see.
* Chest pressure or pain.
* Dizziness or fainting.
* Fast pulse or pounding heartbeat.
* Fatigue.
* Swelling in the ankles, legs and belly area.

These symptoms may be caused by many other health conditions. See a healthcare professional for an accurate diagnosis.

## **Diagnosis**

Pulmonary hypertension is hard to diagnose early. It's not often found during a routine physical exam. Even when pulmonary hypertension is more advanced, its symptoms are similar to those of other heart and lung conditions.

To diagnose pulmonary hypertension, a healthcare professional examines you and asks about your symptoms. You are usually asked questions about your medical and family history.

### **Tests**

Tests to diagnose pulmonary hypertension may include:

* **Blood tests.** Blood tests can help find the cause of pulmonary hypertension. The test also may help find complications of the disease.
* **Chest X-ray.** A chest X-ray is a picture of the heart, lungs and chest. It may be used to check for other lung conditions that can cause pulmonary hypertension.
* **Electrocardiogram (ECG or EKG).** This simple test records the electrical activity of the heart. It shows how the heart is beating.
* **Echocardiogram.** Sound waves create pictures of the beating heart. An echocardiogram shows how blood flows through the heart and heart valves. This test may be done to help diagnose pulmonary hypertension or to learn how treatments are working.

Sometimes, an echocardiogram is done while exercising on a stationary bike or treadmill to learn how activity affects the heart. If you have this test, you may be asked to wear a mask that checks how well the heart and lungs use oxygen and carbon dioxide.

* **Right heart catheterization.** If an echocardiogram shows pulmonary hypertension, this test may be done to confirm the diagnosis.

During this procedure, a doctor places a thin, flexible tube called a catheter into a blood vessel, usually in the neck. The tube is gently guided into the lower right heart chamber and the pulmonary artery. The doctor can then measure blood pressure in the main pulmonary arteries and the right ventricle.

Other tests may be done to check the condition of the lungs and pulmonary arteries. The following tests may give more information about the cause of pulmonary hypertension:

* **Exercise stress tests.** These tests often involve walking on a treadmill or riding a stationary bike while the heartbeat is watched. They can show how the heart reacts to exercise.
* **Computerized tomography (CT) scan.** This test uses X-rays to make pictures of specific parts of the body. Dye called contrast may be given into a vein to help the blood vessels show up more clearly on the images.

A heart CT scan, called a cardiac CT scan, can show the size of the heart and any blockages in the pulmonary arteries. It can help diagnose lung diseases that might lead to pulmonary hypertension. Examples are COPD or pulmonary fibrosis.

* **Magnetic resonance imaging (MRI).** This test uses magnetic fields and radio waves to make detailed pictures of the heart. It can show blood flow in the pulmonary arteries. The test may be done to learn how well the right lower heart chamber is working.
* **Lung function test.** For this test, you blow into a special device. The device measures how much air the lungs can hold. It shows how air flows in and out of the lungs.
* **Sleep study.** A sleep study measures brain activity, heart rate, blood pressure, oxygen levels and other things as you sleep. The test can help diagnose sleep apnea, which can cause pulmonary hypertension.
* **Ventilation/perfusion (V/Q) scan.** In this test, a radioactive tracer is given through a vein (IV). The tracer shows how blood flows. You also may breathe in a tracer that shows airflow to the lungs. A V/Q scan can tell whether blood clots are causing symptoms of pulmonary hypertension.
* **Lung biopsy.** Rarely, a sample of tissue may be taken from the lung to check for a possible cause of pulmonary hypertension.

### **Genetic testing**

Screening for gene changes that cause pulmonary hypertension may be recommended. If you have these gene changes, other family members may need to be screened too.

### **Pulmonary hypertension functional classification**

Once a diagnosis of pulmonary hypertension is confirmed, the condition is classified according to how the symptoms affect you and your ability to do everyday tasks.

Pulmonary hypertension may fall into one of the following groups:

* **Class I.** Pulmonary hypertension is diagnosed, but there are no symptoms during rest or exercise.
* **Class II.** There are no symptoms at rest. Everyday chores or activities such as going to work or the grocery store may cause some shortness of breath or mild chest pain. There's a slight limitation of physical activity.
* **Class III.** It's comfortable at rest, but doing simple tasks such as bathing, dressing or preparing meals causes fatigue, shortness of breath and chest pain. The ability to do physical activity becomes very limited.
* **Class IV.** Symptoms occur at rest and during physical activity. Any type of activity causes increasing discomfort.

Your healthcare team may use a risk calculator that looks at symptoms and test results to understand what type of treatment is needed. This is called pulmonary hypertension risk stratification.

**Treatment**

There's no cure for pulmonary hypertension. But treatments can improve symptoms and help you live longer. Treatment also can help keep the disease from getting worse.

It often takes some time to find the best pulmonary hypertension treatment. The treatments are often complex. You usually need a lot of health checkups.

### **Medications**

If you have pulmonary hypertension, you may get medicines to treat your symptoms and help you feel better. Medicines also may be used to treat or prevent complications. Treatment may include:

* **Medicines to relax blood vessels, called vasodilators.** These medicines open narrowed blood vessels and improve blood flow. The medicine may be breathed in, taken by mouth or given through a vein. Sometimes, it's given continuously through a small pump attached to the body.

Examples of vasodilators to treat pulmonary hypertension include epoprostenol (Flolan, Veletri), treprostinil (Remodulin, Tyvaso, others), iloprost and selexipag (Uptravi).

* **Soluble guanylate cyclase (sGC) stimulators.** This type of medicine relaxes the pulmonary arteries and lowers pressure in the lungs. An example is riociguat (Adempas). Do not take these medicines if you're pregnant.
* **Medicines to widen blood vessels.** Medicines called endothelin receptor antagonists reverse the effect of a substance in the walls of blood vessels that causes them to narrow. Such medicines include bosentan (Tracleer), macitentan (Opsumit) and ambrisentan (Letairis). They may improve energy level and symptoms. Do not take these medicines if you're pregnant.
* **Medicines to increase blood flow.** Medicines called phosphodiesterase 5 (PDE5) inhibitors may be used to increase blood flow through the lungs. These medicines also are used to treat erectile dysfunction. They include sildenafil (Revatio, Viagra) and tadalafil (Adcirca, Alyq, Cialis).
* **High-dose calcium channel blockers.** These medicines help relax the muscles in the walls of blood vessels. They include amlodipine (Norvasc), diltiazem (Cardizem, Tiazac, others) and nifedipine (Procardia). Although calcium channel blockers can be effective, only a small number of people with pulmonary hypertension improve while taking them.
* **Blood thinners.** Also called anticoagulants, these medicines help prevent blood clots. One example is warfarin (Jantoven). The medicines can increase the risk of bleeding. This is especially true if you're having surgery or a treatment that enters the body or creates an opening in the skin. Talk to your healthcare team about your risk.
* **Digoxin (Lanoxin).** This medicine helps the heart beat stronger and pump more blood. It can help control irregular heartbeats.
* **Water pills, also called diuretics.** These medicines help the kidneys remove excess fluid from the body. This reduces the amount of work the heart has to do. Diuretics also may be used to reduce fluid buildup in the lungs, legs and belly area.
* **Oxygen therapy.** Breathing pure oxygen may be suggested if you live at a high altitude or have sleep apnea. Some people with pulmonary hypertension need oxygen therapy all the time.

### **Surgery or other procedures**

If medicines do not help control the symptoms of pulmonary hypertension, surgery may be recommended. Surgeries and procedures to treat pulmonary hypertension may include:

* **Atrial septostomy.** This treatment may be done if medicines don't control pulmonary hypertension symptoms. In an atrial septostomy, a doctor creates an opening between the upper left and right chambers of the heart. The opening reduces the pressure on the right side of the heart. Potential complications include irregular heartbeats called arrhythmias.
* **Lung or heart-lung transplant.** Sometimes, a lung or heart-lung transplant may be needed, especially for younger people who have idiopathic pulmonary arterial hypertension. After a transplant, medicine must be taken for life to prevent the body from rejecting the new organ.

**pulmonary hypertension (PH) treatment drugs and their common side effects**

1. Phosphodiesterase-5 (PDE-5) Inhibitors

* Examples: Sildenafil (Revatio), Tadalafil (Adcirca)
* Mechanism: Promote vasodilation by increasing nitric oxide signaling in pulmonary arteries.
* Common Side Effects:
  + Headache
  + Flushing
  + Nasal congestion
  + Dyspepsia
  + Visual disturbances (more with sildenafil)
  + Hypotension (especially if combined with nitrates)

## 2. Endothelin Receptor Antagonists (ERAs)

* Examples: Bosentan (Tracleer), Ambrisentan (Letairis), Macitentan (Opsumit)
* Mechanism: Block endothelin-1 receptors causing vasodilation and reduced vascular remodeling.
* Common Side Effects:
  + Liver enzyme elevation (requires regular liver function monitoring)
  + Anemia
  + Peripheral edema
  + Nasal congestion
  + Teratogenicity (contraindicated in pregnancy)

## 3. Prostacyclin Analogues and Prostacyclin Receptor Agonists

* Examples:
  + Epoprostenol (Flolan, Veletri)
  + Treprostinil (Remodulin, Tyvaso, Orenitram)
  + Iloprost (Ventavis)
  + Selexipag (Uptravi) – a selective prostacyclin receptor agonist
* Mechanism: Vasodilation, inhibition of platelet aggregation, and antiproliferative effects.
* Common Side Effects:
  + Jaw pain
  + Headache
  + Flushing
  + Diarrhea, nausea
  + Hypotension
  + Infusion site pain (especially with subcutaneous treprostinil)
  + Risk of catheter-related infections (for intravenous forms)
  + Jaw pain and musculoskeletal pain

## 4. Soluble Guanylate Cyclase (sGC) Stimulators

* Example: Riociguat (Adempas)
* Mechanism: Stimulates sGC to increase cyclic GMP leading to vasodilation.
* Common Side Effects:
  + Hypotension
  + Headache
  + Dizziness
  + Gastrointestinal discomfort
  + Bleeding risk

## 5. Calcium Channel Blockers (CCBs)

* Examples: Amlodipine, Diltiazem, Nifedipine
* Use: Only in a small subset of patients who respond positively to vasoreactivity testing.
* Common Side Effects:
  + Peripheral edema
  + Hypotension
  + Headache
  + Flushing
  + Constipation (especially with verapamil)

**Lifestyle and home remedies**

Lifestyle changes may help improve pulmonary hypertension symptoms. Try these tips:

* **Eat healthy.** Eat a healthy diet rich in whole grains, fruits and vegetables, lean meats, and low-fat dairy products. Try to stay away from saturated fat, trans fat and cholesterol. Use less salt.
* **Stay as active as possible and manage weight.** Even mild forms of activity might be too exhausting for some people who have pulmonary hypertension. For others, moderate exercise, such as walking, might be helpful — especially when done during oxygen therapy. Your healthcare team can help you plan an appropriate exercise program.
* **Don't smoke.** If you smoke, quit. If you need help, ask your healthcare team for treatment that can help. Avoid secondhand smoke too, if possible.
* **Get plenty of rest.** Resting can reduce tiredness related to pulmonary hypertension.
* **Avoid high altitudes.** High altitudes can make pulmonary hypertension worse. If you live at an altitude of 8,000 feet (2,438 meters) or higher, you might be told to consider moving to a lower altitude.
* **Avoid activities that can lower blood pressure a lot.** These include sitting in a hot tub or sauna or taking long hot baths or showers. Such activities lower blood pressure and can cause fainting. Also, do not do activities that cause a lot of straining, such as lifting heavy objects or weights.
* **Tell your healthcare team about the medicines you take.** Some medicines can make pulmonary hypertension worse or affect its treatment.
* **Get regular health checkups.** Tell your healthcare team about any new or worsening symptoms or medicine side effects. If pulmonary hypertension affects your quality of life, ask about treatments that could help.
* **Get recommended vaccines.** Respiratory infections can cause serious health concerns for people with pulmonary hypertension. Ask your healthcare team which vaccines you need to prevent common viral infections.
* **Talk to a healthcare professional before becoming pregnant.** Pulmonary hypertension can cause serious complications for the pregnant person and unborn baby, also called a fetus. Birth control pills can increase the risk of blood clots. Talk to your healthcare team about other birth control options.

**Complications**

Potential complications of pulmonary hypertension are:

* **Right-sided heart enlargement and heart failure.** Also called cor pulmonale, this condition causes the heart's right lower chamber to get larger. The chamber has to pump harder than usual to move blood through narrowed or blocked lung arteries.

As a result, the heart walls get thick. The right lower heart chamber stretches to increase the amount of blood it can hold. These changes create more strain on the heart. Eventually the right lower heart chamber fails.

* **Blood clots.** Pulmonary hypertension increases the risk of blood clots in the small arteries in the lungs.
* **Irregular heartbeats, also called arrhythmias.** Pulmonary hypertension can cause changes in the heartbeat, which can be life-threatening.
* **Bleeding in the lungs.** Pulmonary hypertension can lead to life-threatening bleeding in the lungs and coughing up blood.
* **Pregnancy complications.** Pulmonary hypertension can be life-threatening for the mother and the developing baby.

## **Outlook / Prognosis**

The outlook for people with pulmonary hypertension depends on:

* The cause of PH.
* How early it’s diagnosed.
* The severity of symptoms.
* Associated medical conditions.

The outlook for each person is different. Talk with your provider to learn more about your prognosis and how to manage your condition.

### **Can pulmonary hypertension be cured?**

Most cases of pulmonary hypertension can’t be cured. Your provider may prescribe medications to:

* Ease your symptoms.
* Improve your quality of life.
* Slow down the progression of the disease.

Your provider may also recommend lifestyle changes.

However, surgery can cure some people with chronic thromboembolic pulmonary hypertension (CTEPH).

The life expectancy varies from person to person. It depends how quickly you’re diagnosed and what other medical conditions you have. Talk with your provider about what you can expect in your individual situation.

Pulmonary hypertension is a progressive disease. That means it gets worse over time. It progresses more quickly in some people than in others. Treatment can improve your chances of surviving pulmonary hypertension for many years.

### **Is pulmonary hypertension fatal?**

Without treatment, pulmonary hypertension leads to right-sided heart failure and, ultimately, death. Treatment can help you live longer and give you a better quality of life.

**Prevention**

Risk factors for developing pulmonary hypertension include:

* Exposure to asbestos.
* Family history of blood clots.
* Family history of pulmonary hypertension.
* Living at high altitudes.
* Smoking and using tobacco products.
* Use of diet medications such as “fen-phen” (dexfenfluramine and phentermine).
* Use of some prescription medications that treat cancer and depression.
* Use of recreational drugs.

Certain medical conditions also raise your risk. These include:

* Blood clots in your pulmonary arteries.
* Connective tissue disease.
* Down syndrome.
* Gaucher disease.
* Heart disease.
* HIV.
* Liver disease.
* Lung disease.

Talk with your provider about your risk factors and what you can do to lower your risk.

### **How can I prevent pulmonary hypertension?**

It’s not always possible to prevent pulmonary hypertension. Some risk factors are out of your control. If you have risk factors, your provider may recommend preventive screenings to check your heart and lung function.

Doing whatever you can to prevent or manage other medical conditions can help lower your risk of pulmonary hypertension. Steps you can take include:

* **Create an exercise plan**. Ask your provider what exercises are safe for you.
* **Follow a heart-healthy diet**. Avoid processed foods, fast food and other foods high in salt and saturated fat.
* **Quit smoking and stop using tobacco**. Smoking and tobacco use are top risk factors for heart and lung problems. Quitting isn’t easy, especially if you’ve been smoking or using tobacco for a long time. But your provider can help provide resources. Support groups may also help.
* **Take medications** for blood pressure and other conditions as prescribed.

## **Living With**

Talk with your provider about how to manage and monitor your condition. Your provider will tell you when you need to come in for checkups and tests. Be sure to tell your provider about any new or changing symptoms.

Also, talk with your provider about:

* **Birth control options and whether it’s safe to become pregnant**. Pregnancy is risky for people with pulmonary hypertension.
* **Creating an emergency kit**. People with pulmonary hypertension need certain supplies and information with them at all times. Ask your provider what you should include in your emergency kit.
* **Exercise**. Ask what’s safe for you and how best to add exercise into your daily life. Ask what exercises to avoid.
* **Seasonal vaccines**, including for flu and pneumonia. Your provider will let you know which vaccines you should receive.
* **Support groups or counseling** as you adjust to the “new normal” of life with pulmonary hypertension. A medical diagnosis brings many emotions. You don’t have to carry them alone.
* **Travel plans.** You may need to take certain precautions if flying on an airplane. You may also need to be careful when traveling to high altitudes. Talk with your provider about how to prepare for travel and what to bring with you.

### **What dietary changes should I make?**

Your provider will give you specific recommendations. One key step involves reducing your sodium intake. This means:

* Avoid adding salt at the table or using “seasoning salt.”
* Avoid smoked, cured, salted and canned meat products.
* Buy foods that are “low sodium” or “low salt.”
* Limit fast foods and prepared foods.

Other dietary changes include:

* Eat foods high in fiber (like whole grains, bran, fruits and vegetables).
* Eat foods high in potassium (like dried fruits, bananas and oranges).
* Eat foods high in magnesium (like peanuts, tofu and broccoli).
* Limit foods that contain refined sugar, saturated fat and cholesterol.

### **When should I call my healthcare provider?**

Call your provider if you’re having problems with:

* A fast heart rate (120 beats per minute).
* A respiratory infection or cough that’s getting worse.
* Constantly feeling dizzy or lightheaded.
* Episodes of chest pain or discomfort with physical activity.
* Extreme fatigue or decreased ability to do your normal activities.
* Nausea or lack of appetite.
* Restlessness or confusion.
* Shortness of breath that’s gotten worse, especially if you wake up feeling short of breath.
* Swelling in your ankles, legs or tummy that’s gotten worse.
* Trouble breathing with regular activities or at rest.
* Weight gain (2 pounds in one day or 5 pounds in one week).

Go to the emergency department or call your local emergency number if you have:

* A fast heart rate (120-150 beats per minute) that won’t go down.
* Fainting spells with loss of consciousness.
* Hickman catheter complications with intravenous prostacyclins. These include infection, catheter displacement, solution leak, bleeding and IV pump malfunction.
* Shortness of breath that doesn’t go away when you rest.
* Sudden and severe chest pain.
* Sudden and severe headache.
* Sudden weakness or paralysis in your arms or legs.

**PREDEFINED QUESTIONS AND ANSWERS**

## 1. What is the likely cause of my symptoms or condition?

Pulmonary hypertension is caused by increased blood pressure in the pulmonary arteries. The causes vary and include:

* Pulmonary arterial hypertension (PAH): idiopathic, inherited, or associated with connective tissue diseases, HIV, or drugs.
* Left heart disease: such as left ventricular dysfunction or valvular disease.
* Lung diseases: COPD, interstitial lung disease, or sleep apnea.
* Chronic blood clots: chronic thromboembolic pulmonary hypertension (CTEPH).
* Other rare causes: metabolic or systemic disorders.

Your symptoms like breathlessness, fatigue, and swelling may be due to increased strain on the right side of your heart from elevated pulmonary artery pressures.

## 2. What are other possible causes?

Other conditions with similar symptoms include:

* Chronic obstructive pulmonary disease (COPD)
* Heart failure
* Interstitial lung disease
* Asthma
* Pulmonary embolism
* Anemia or other systemic illnesses

Because symptoms overlap, thorough testing is needed to differentiate.

## 3. What tests do I need?

Common tests to diagnose and evaluate PH include:

* Echocardiogram: estimates pulmonary artery pressure and heart function.
* Right heart catheterization: gold standard to measure pulmonary artery pressures directly and confirm diagnosis.
* Electrocardiogram (ECG): assesses heart rhythm and right heart strain.
* Chest X-ray and CT scan: evaluate lung structure and possible causes.
* Pulmonary function tests: assess lung capacity and function.
* Blood tests: rule out other diseases like thyroid, liver disease.
* Exercise tests: 6-minute walk test or cardiopulmonary exercise testing to assess functional capacity.
* Ventilation-perfusion scan: to detect chronic blood clots (CTEPH).

## 4. What treatment do you recommend?

Treatment depends on the type and cause of PH:

* For pulmonary arterial hypertension (PAH): targeted therapies such as endothelin receptor antagonists, phosphodiesterase-5 inhibitors, prostacyclin analogues, or soluble guanylate cyclase stimulators.
* For CTEPH: surgical removal of clots (pulmonary thromboendarterectomy) or targeted medications if surgery is not possible.
* For PH caused by left heart or lung disease: managing the underlying condition (e.g., heart failure treatment, oxygen therapy).
* Supportive care: diuretics for fluid retention, anticoagulation if indicated, and oxygen supplementation.

## 5. What are the other treatment options?

* Combination therapy: using two or more PH-specific drugs.
* Lung transplantation: considered in severe, refractory cases.
* Lifestyle modifications: including supervised exercise programs.
* Clinical trials: for novel therapies.
* Treatment of contributing factors like sleep apnea or anemia.

## 6. Is there a generic form of the medicine you're prescribing?

* Some PH medications like sildenafil (a PDE-5 inhibitor) are available in generic forms.
* Many newer drugs, such as endothelin receptor antagonists and prostacyclin analogues, may still be brand-name only, depending on your country and insurance coverage.
* Your doctor or pharmacist can provide specific information on generics available to you.

## 7. What's an appropriate level of physical activity?

* Physical activity should be tailored to your functional class:
  + Mild to moderate exercise is encouraged in stable patients.
  + Avoid strenuous or high-intensity activities if you experience symptoms.
  + Pulmonary rehabilitation or supervised exercise programs are beneficial.
* Your doctor will advise based on your symptoms and test results.

## 8. Are there any restrictions that I need to follow?

* Avoid high altitudes or situations causing low oxygen levels.
* Limit strenuous physical activity if symptomatic.
* Avoid pregnancy due to high risks.
* Avoid medications that can worsen PH (e.g., certain appetite suppressants, stimulants).
* Follow dietary recommendations, including sodium restriction if fluid retention occurs.

## 9. How often do I need health checkups?

* Regular follow-up every 3 to 6 months is typical.
* Monitoring includes symptom assessment, echocardiograms, blood tests, and exercise capacity.
* Frequency may increase if your condition worsens or treatment changes.

## 10. I have other health conditions. How can I best manage them together?

* Coordinated care with your cardiologist, pulmonologist, and primary care provider is essential.
* Manage comorbidities such as heart disease, lung disease, or sleep apnea aggressively.
* Inform all your doctors about your PH diagnosis and medications to avoid drug interactions.
* Lifestyle modifications like smoking cessation, weight management, and controlling blood pressure are important.

## 11. Should I see a specialist?

* Yes, PH requires evaluation and management by specialists in pulmonary hypertension or cardiology.
* Referral to a pulmonary hypertension center is recommended for diagnosis confirmation and treatment planning

## **Genomic Data on Pulmonary Hypertension:**

* BMPR2 gene mutations are the most common genetic cause of PAH. BMPR2 encodes a receptor in the transforming growth factor-beta (TGF-β) superfamily that regulates cell growth in pulmonary arteries. Mutations in BMPR2 lead to excessive cell proliferation and narrowing of small pulmonary arteries, increasing pulmonary vascular resistance and pressure.
* Inheritance pattern: BMPR2 mutations are inherited in an autosomal dominant manner but with *incomplete penetrance* (~10–30%). This means not all carriers develop PAH. Women have a higher penetrance than men. Each child of a carrier has a 50% chance of inheriting the mutation, but only about 20% of those with the mutation develop the disease during their lifetime.
* Other genes implicated include:
  + ALK1 (ACVRL1) and ENG (endoglin), associated with hereditary hemorrhagic telangiectasia and PAH.
  + EIF2AK4, linked to pulmonary veno-occlusive disease (PVOD) and pulmonary capillary hemangiomatosis (PCH), inherited recessively.
  + CAV1 and KCNK3, among others, identified in some familial and idiopathic cases.
* Genetic mutations explain over 70% of familial PAH cases and about 10–40% of idiopathic PAH cases.
* The presence of a mutation often requires additional environmental or biological "second hits" such as hypoxia, inflammation, or drug exposure to trigger disease manifestation.
* Genetic testing panels mainly focus on BMPR2 and related genes but may not cover all rare mutations

## **Diagnostic Considerations**

Other WHO groups of pulmonary hypertensions should be considered as coincident or alternative diagnoses. These include pulmonary arterial hypertension (PAH) (WHO group 1), pulmonary hypertension (WHO groups 2, 3, and 5), and other pulmonary arterial obstruction (e.g., tumors of the pulmonary artery, pulmonary artery stenoses, arteritis, and mediastinal fibrosis).

## **Differential Diagnoses**

* Pulmonary Embolism CT Imaging and Diagnosis
* Atrial Septal Defect
* Chronic Obstructive Pulmonary Disease (COPD)
* CREST Syndrome
* Drug-Induced Pulmonary Toxicity
* Emphysema
* High Altitude Pulmonary Hypertension
* HIV Infection and AIDS
* Hypertrophic Cardiomyopathy
* Connective Tissue Disease-Associated Interstitial Lung Disease (CTD-ILD)
* Mitral Regurgitation
* Mitral Stenosis
* Obstructive Sleep Apnea (OSA)
* Pediatric Obesity-Hypoventilation Syndrome
* Portopulmonary syndrome
* Pulmonary Veno-Occlusive Disease
* Restrictive Cardiomyopathy
* Restrictive Lung Disease
* Schistosomiasis (Bilharzia)
* Scleroderma
* Systemic Lupus Erythematosus (SLE)
* Vascular Surgery for Arteriovenous Malformations
* Ventricular Septal Defects

## **Epidemiology**

According to registry data, there are no sex differences in the prevalence of CTEPH. The average age onset is usually in the 60s. Nonresolution of PE is the most common cause of CTEPH.In a European registry, 75% of patients with CTEPH reported previous PE.

The incidence of CTEPH after acute PE is not clear. A review estimates that the full incidence of CTEPH in the United States is 4,886 cases per year,based on an incidence of CTEPH after PE of 0.57% reported by Klok et al.Possibly the most quoted study related to PE incidence was published in the *New England Journal of Medicine* in 2004.In this study, 223 patients with acute PE and without a previous venous thromboembolic event were followed for 10 years. Seven (3.8%) of 223 patients developed symptomatic CTEPH 2 years after the initial thromboembolic event. Other studies corroborated the fact that the majority of CTEPH cases developed 2 years after the initial event of acute PE.Historically, between 0.1% and 0.5% of patients with PE were thought to develop CTEPH. [3] In 2006, another study showed that CTEPH developed in 1% of patients with acute PE, but this study excluded patients with known venous thromboembolic risk development. Since then, other studies have revealed a prevalence of 0.4-4.8%.These rate differences are likely due to the different populations studied, different methods of screening, and follow-up duration. In summary, irrespective of the rate of CTEPH from PE used in estimates, it is clear that a large proportion of CTEPH cases are underdiagnosed each year. Factors contributing to this underdiagnosis and treatment include poor specificity of initial signs and symptoms and unknown PE history at presentation.

The epidemiological analysis suggests that the full incidence of CTEPH in the United States and Europe ranges from 3-5 cases per 100,000 population per year. The average diagnosis rate was 14.2% for the United States and Europe in 2013, which equates to a diagnosed incidence of 4-7 cases per million. Although the incidence of PE in Japan is substantially lower than the incidence observed in United States and Europe, the rate of CTEPH is much higher. This disparity might be related to genetics, population demographics, or lifestyle, among other factors. An alternative explanation is that only severe cases of PE might be diagnosed, while mild cases might not be recognized or reported.

Not only diagnosis, but also proper referral to an expert center, seem to be delayed in CTEPH. In one study, the mean delay from symptom onset to diagnosis of CTEPH was 18 ±26 months. This also transpires in the severity of disease, since most of the patients present with WHO functional class III and IV.Late diagnosis can negatively impact treatment options and overall quality of life.

Epidemiological projections in seven countries indicate that the incidence of CTEPH, especially late-stage disease, will increase over time from 32,636 cases (16%) diagnosed in 2015 to 37,009 cases (28%) in 2025.

Pulmonary hypertension can affect people of any age. Present estimates suggest a prevalence of approximately 1% in the global population. Left heart disease (LHD) is the leading cause, followed by lung disease, particularly chronic obstructive pulmonary disease (COPD). In developing countries, CHD, infectious diseases like schistosomiasis and HIV, and high altitude are significant pulmonary hypertension causes. PAH incidence is approximately 6 cases per million adults, and the prevalence is about 49 to 55 cases per million adults. PAH was initially thought to affect predominantly young women. However, recent data show that the condition is also prevalent in patients aged 65 years and older with cardiovascular comorbidities, leveling the sex distribution.

At least 50% of patients with heart failure with preserved ejection fraction either have IpcPH or CpcPH.The prevalence of pulmonary hypertension increases with disease severity in these patients, with 60 to 70% of patients with severe and symptomatic mitral valve disease and 50% with symptomatic aortic stenosis affected by pulmonary hypertension. Mild pulmonary hypertension is common in patients with advanced COPD and interstitial lung disease (ILD). Only 1% to 5% of patients with advanced COPD were found to have severe pulmonary hypertension. The prevalence increases with increasing severity in patients with idiopathic pulmonary fibrosis and is as high as 60% in patients with end-stage disease.

Registry data indicate that CTEPH’s incidence and prevalence are 2 to 6 and 26 to 39 cases per million adults, respectively. Pulmonary hypertension in individuals with sarcoidosis is frequent and often associated with increased mortality and morbidity

## **Guidelines Summary**

The guidelines have been endorsed by the International Society for Heart and Lung Transplantation (ISHLT) and the European Reference Network on rare respiratory diseases (ERN-LUNG)

Among the key changes were a lowered cutoff from mean pulmonary arterial pressure (mPAP) >25 mm Hg. Pulmonary hypertension (PH) is now defined by a mPAP >20 mm Hg at rest. The definition of pulmonary arterial hypertension (PAH) also implies a pulmonary vascular resistance (PVR) >2 Wood Units and pulmonary arterial wedge pressure ≤15 mm Hg.

### Diagnosis

The diagnostic algorithm was also restructured so that the diagnosis is made earlier. The following three steps are now included in the algorithm:

* Step 1: Suspicion by a general practitioner or other primary care provider.
* Step 2: Detection by echocardiography.
* Step 3: Confirmation with right heart catheterization in a pulmonary hypertension center. If the patient probably has intermediate or high-risk pulmonary hypertension, then he or she should be referred to a pulmonary hypertension center for a comprehensive diagnostic workup and invasive assessment, as needed.

In patients with acute PE, and the following clinical conditions, ventilation-perfusion lung scanning, echocardiography and cardiopulmonary exercise testing should be performed to assess for CTEPH:

* Radiological signs suggest CTEPH on the computed tomography pulmonary angiography (CTPA) performed to diagnose PE and/or estimated systolic pulmonary arterial pressure (sPAP) is >60 mmHg on echocardiogram
* Persistent or new-onset dyspnea or exercise limitation post-PE
* Asymptomatic patients with risk factors for CTEPH or a high CTEPH prediction score

High risk factors for CTEPH include :

* Permanent intravascular devices (pacemaker, long-term central lines, ventriculoatrial shunts)
* Inflammatory bowel disease
* Essential thrombocythemia or polycythemia vera
* Splenectomy
* Antiphospholipid syndrome
* High-dose thyroid hormone replacement

### Treatment

Key recommendations specific to management of group CTEPH and CTEPD without PH are summarized below.

All patients with CTEPH should receive lifelong anticoagulation therapy and be tested for antiphospholipid syndrome; patients positive for antiphospholipid syndrome should receive a vitamin K antagonist (VKA) for anticoagulation therapy.

The treatment of choice is pulmonary thromboendarterectomy (PTE) for patients with fibrotic obstructions within pulmonary arteries accessible by surgery. Balloon pulmonary angioplasty (BPA) is recommended for the following patients with distal obstructions amenable to BPA :

* PTE cannot be performed due to technical inoperability
* Persistent/residual PH after PTE

Consider medical therapy prior to BPA. Long-term follow-up is required for patients following PEA and BPA, as well as for patients with CTEPH on medical therapy.

Riociguat is recommended for symptomatic patients with inoperable CTEPH or persistent/recurrent PH after PTE. The following medications may be considered for management of inoperable CTEPH in select patients:

* Subcutaneous treprostinil
* Off-label use of PAH medications
* Combination of soluble guanylate cyclase (sGC) stimulator/phosphodiesterase 5 inhibitor (PDE5i), endothelin receptor antagonist (ERA), or parenteral prostacyclin analogues

**DOCTOR PATIENT CONVERSATION**

Doctor: Hello, how have you been feeling since our last visit?

Patient: I’ve been more short of breath lately, especially when I walk or climb stairs. Sometimes I feel tired and dizzy.

Doctor: I’m sorry to hear that. These symptoms can be related to your pulmonary hypertension. Let’s talk about what might be causing this change and how we can help you feel better.

Patient: What exactly is causing my symptoms?

Doctor: Pulmonary hypertension means the blood pressure in your lungs’ arteries is higher than normal. This puts extra strain on your heart. The causes vary, but in your case, it’s related to the narrowing of the small blood vessels in your lungs, which makes it harder for blood to flow through.

Patient: Are there other things that could cause these symptoms?

Doctor: Yes, symptoms like breathlessness and fatigue can also be caused by heart failure, lung diseases like COPD, or anemia. That’s why we do tests to be sure.

Patient: What tests do I need now?

Doctor: We’ll start with an echocardiogram to check your heart function and estimate the pressure in your lungs. We may also do a right heart catheterization, which is the best way to measure lung pressures directly. Other tests like lung function tests and blood work will help us understand your overall health.

Patient: What treatments do you recommend?

Doctor: Based on your diagnosis, we’ll use medications that help open the blood vessels in your lungs and reduce the pressure. These include drugs like endothelin receptor antagonists or phosphodiesterase-5 inhibitors. We’ll also manage symptoms with diuretics if you have fluid retention.

Patient: Are there other treatment options?

Doctor: Yes, sometimes combination therapy with more than one medication is needed. In severe cases, procedures like lung transplantation or surgery for chronic clots might be considered. Lifestyle changes and supervised exercise can also improve your quality of life.

Patient: Is there a generic version of the medicine you’re prescribing?

Doctor: Some medications, like sildenafil, are available as generics. Others may only be available as brand-name drugs. We’ll consider cost and availability when choosing your treatment.

Patient: What about physical activity? How much can I do?

Doctor: Moderate, supervised exercise is encouraged to maintain your strength, but avoid strenuous activities that worsen your symptoms. We can refer you to a pulmonary rehab program for guidance.

Patient: Are there any restrictions I should follow?

Doctor: Avoid high altitudes and situations that lower your oxygen levels. Also, avoid pregnancy as it poses high risks. It’s important to avoid medications that can worsen PH, and we’ll review all your medicines carefully.

Patient: How often do I need checkups?

Doctor: Usually every 3 to 6 months, but if your symptoms change, we might see you sooner to adjust treatment.

Patient: I have other health problems. How do I manage them with PH?

Doctor: Coordinating care is key. We’ll work with your other doctors to manage all your conditions safely and effectively.

Patient: Should I see a specialist?

Doctor: Yes, managing PH is complex, and care at a specialized center improves outcomes. I’ll refer you to a pulmonary hypertension specialist.

Patient: Is there information I can take home? Any websites you recommend?

Doctor: Absolutely. I’ll give you some printed materials, and you can visit trusted sites like the Pulmonary Hypertension Association (phassociation.org) and the American Lung Association (lung.org). They offer excellent patient resources and support groups.

Patient: Thank you, doctor. I appreciate you explaining all this.

Doctor: You’re welcome. Remember, we’re here to support you every step of the way. Don’t hesitate to contact us if you have questions or concerns.

REFERENCES

[Pulmonary hypertension - Diagnosis and treatment - Mayo Clinic](https://www.mayoclinic.org/diseases-conditions/pulmonary-hypertension/diagnosis-treatment/drc-20350702)

[Pulmonary Hypertension: Symptoms, Treatment](https://my.clevelandclinic.org/health/diseases/6530-pulmonary-hypertension-ph#outlook-prognosis)

<https://www.ncbi.nlm.nih.gov/books/NBK482463/#article-28033.s4>

<https://emedicine.medscape.com/article/2500057-guidelines>

<https://www.nhs.uk/conditions/pulmonary-hypertension/diagnosis/>

### **Pulmonary arterial hypertension**

**ALTERNATIVE NAME**S

* Primary Pulmonary Hypertension (PPH) — used historically for idiopathic cases before updated classification
* Idiopathic Pulmonary Arterial Hypertension (IPAH) — when no underlying cause is identified
* Heritable Pulmonary Arterial Hypertension (HPAH) — when there is a known genetic cause
* Secondary Pulmonary Arterial Hypertension — when PAH occurs due to other diseases (though this term is less used as PAH specifically refers to Group 1 pulmonary hypertension)
* Pulmonary Hypertension, Group 1 (per WHO classification)
* Plexogenic Pulmonary Arteriopathy — referring to characteristic vascular changes seen in PAH
* Eisenmenger syndrome (a subset involving PAH in congenital heart disease)

**DEFINITION AND DESCRIPTION**

Pulmonary arterial hypertension (PAH) is a condition in which the small blood vessels in your lungs become narrow. As a result, blood can’t flow through your lungs as well as it should. This leads to high blood pressure in your pulmonary arteries. These are the arteries that carry oxygen-poor blood from your heart to your lungs.

Eventually, your heart gets weaker because it has to work harder to pump blood to your lungs. Without treatment, PAH can cause a cascade of problems in your heart, lungs and the rest of your body.

PAH is one form of pulmonary hypertension. This is a general diagnosis for high blood pressure in your pulmonary arteries from any cause.

#### **How does pulmonary arterial hypertension affect my body?**

Pulmonary arterial hypertension strains the right side of your heart, which pumps oxygen-poor blood to your lungs. This strain can lead to right-sided heart failure.

Plus, PAH slows down blood flow between your heart and lungs. This means less blood can enter your lungs to gain fresh oxygen. As a result, blood flow to the rest of your body also slows down. So, your organs and tissues can’t get enough oxygen.

Without treatment, PAH can be fatal.

#### **How serious is pulmonary arterial hypertension?**

Pulmonary arterial hypertension is a serious condition that can be life-threatening. An early diagnosis and swift treatment can help you live longer and have a better quality of life.

### **Who does pulmonary arterial hypertension affect?**

Pulmonary arterial hypertension can affect adults at any age. It’s more common among females, who are usually diagnosed between the ages of 30 and 60. Males over age 65 who develop PAH are more likely to have severe cases.

PAH can also affect infants. This condition is known as persistent pulmonary hypertension in the neonate (PPHN).

PAH isn’t as common as other forms of pulmonary hypertension, including those caused by underlying heart or lung disease. Each year, about 500 to 1,000 people are diagnosed with PAH in the U.S.

In Western countries, about 25 per 1 million people are living with PAH.

## **Symptoms**

You may not feel any symptoms early on. Most people with PAH begin to notice symptoms as the condition progresses. These symptoms can include:

* Blue fingers or lips.
* Chest pain or pressure.
* Dizziness or fainting.
* Fatigue.
* Racing or pounding heartbeat.
* Shortness of breath that gets worse over time.
* Swelling (edema) in your feet and legs, later progressing to your belly and neck.

Without treatment, PAH symptoms continue to get worse over time. You may find it harder to do your usual activities without needing to catch your breath or rest.

### **CAUSES**

Damage to the lining of the blood vessels in your lungs leads to pulmonary arterial hypertension. It’s not always clear what causes this damage. When there’s no clear cause, you have what’s called “idiopathic” pulmonary arterial hypertension.

Other times, there is a clear cause. Known causes of PAH include associated medical conditions, genetic mutations and certain drugs.

Medical conditions that may lead to the development of PAH include:

* Congenital heart disease.
* Glycogen storage diseases.
* HIV.
* Liver disease.
* Lupus.
* Portal hypertension.
* Pulmonary capillary hemangiomatosis.
* Pulmonary veno-occlusive disease.
* Schistosomiasis.
* Scleroderma.

**Genetic mutations can also cause pulmonary arterial hypertension. Here’s what we know about genetics and PAH:**

* Mutations in your *BMPR2* gene are usually responsible. This gene manages how many cells you have in specific tissues. When there’s a mutation in this gene, you can end up with too many cells in the small arteries of your lungs. This overcrowding of cells narrows the opening of your arteries.
* PAH can run in families. This is called heritable PAH. About 80% of people with heritable PAH have mutations to their *BMPR2* gene.
* Some people carry an altered gene without ever developing PAH.
* Some people develop PAH caused by genetic mutations even though they have no family history of the condition. This is called sporadic PAH.

Certain drugs can cause PAH, including:

* **Diet pills** like “fen-phen.” This drug isn’t available anymore, since research has shown it’s dangerous. But it may cause PAH many years after you’ve stopped taking it.
* **Recreational drugs** like cocaine and methamphetamine.

## **Diagnosis and Tests**

PAH can be hard to diagnose because its symptoms could be caused by many other conditions. Your provider will perform a physical exam and talk with you about your symptoms and medical history. You’ll likely need many tests (including imaging tests and blood tests) to find out if you have pulmonary hypertension and if so, which form.

Your provider may also refer you to a pulmonologist or cardiologist. These specialists will run specific tests to check your heart and lung function. They’ll determine what form of pulmonary hypertension you have (PAH or another form). They’ll also evaluate how far your condition has progressed.

PAH requires specific types of treatment that can’t be used for other forms of pulmonary hypertension. So, your care team must learn as much as possible about what’s happening in your lungs and your heart. This information helps them tailor treatment to your specific diagnosis and needs.

Your care team will use a combination of different tests to rule out other conditions and diagnose PAH.

If your provider suspects you have PAH based on your physical exam, the first test they’ll order for you is a transthoracic echocardiogram. This test evaluates the overall structure and function of your heart.

Other tests may include:

* **Blood tests**: To check organ function and hormone levels and identify underlying disorders. Specific blood tests include a complete metabolic panel and complete blood count.
* **Chest CT scan**: To check for (and rule out) kidney disease.
* **Chest X-ray**: To see if your heart or pulmonary arteries are bigger than they should be.
* **Heart MRI**: To evaluate your right ventricle (the chamber of your heart that pumps blood to your pulmonary arteries).
* **Polysomnogram (PSG)**: To check if you have sleep apnea, which can make PAH worse. A PSG is one type of overnight sleep test.
* **Pulmonary function tests**: To check your lung function.
* **Pulmonary ventilation/perfusion (VQ) scan**: To check for blood clots in your lungs. This test can rule out chronic thromboembolic pulmonary hypertension.
* **Right heart catheterization**: To measure your pulmonary artery pressures. This test is essential for diagnosing PAH.
* **Six-minute walk test**: To see how much exercise you can handle and how much oxygen circulates in your blood while you move.

Talk with your provider about the tests you need and how to prepare for them.

### **What are the criteria for diagnosing pulmonary arterial hypertension?**

Providers diagnose pulmonary hypertension based on the blood pressure in your pulmonary arteries. The criterion for diagnosis is pulmonary arterial pressure higher than 20 mmHg while you’re at rest. A right heart catheterization measures this number.

**Management and Treatment**

Treatment for PAH focuses on slowing down its progression and giving you a better quality of life. There’s no one-size-fits-all approach to PAH treatment. Your provider will work with you to decide the best treatment for your specific needs.

Your individualized treatment plan for PAH may include:

* **Balloon atrial septostomy (BAS)**. This procedure typically treats babies with congenital heart defects, but it’s also sometimes used for adults with PAH. A septostomy helps take pressure off the right side of your heart and also allows more oxygen to circulate in your blood. It may serve as a bridge to a lung transplant.
* **Calcium channel blockers**. These medications help lower the blood pressure in your pulmonary arteries and throughout your body.
* **Diuretics**. These “water pills” get rid of extra fluid in your body and reduce swelling.
* **Oxygen therapy**. You may need this treatment if you don’t have enough oxygen in your blood. Supplemental oxygen can help people during rest, sleep or exercise.
* **Pulmonary vasodilators**. These medications help your pulmonary arteries relax and open up better. This relieves the strain on your heart and helps ease your symptoms.

A last resort option for some people with severe PAH is a lung transplant. This surgery may give you one or two new lungs. A heart-lung transplant gives you a new heart as well.

### **What medications treat pulmonary arterial hypertension?**

Medications that treat PAH come in several different forms:

* **Oral**. To help the blood vessels in your lungs relax, and to prevent them from becoming narrow. These medications may help you be more physically active.
* **Inhaled**: To treat shortness of breath.
* **Portable infusion pump**: To open up your blood vessels so more blood can flow through. This can help improve your symptoms.
* **Intravenous (IV)**: To open up your blood vessels and relieve symptoms like chest pain and trouble breathing.

The U.S. Food and Drug Administration (FDA) has approved the following drugs for people with PAH:

* Ambrisentan (oral).
* Bosentan (oral).
* Epoprostenol (IV).
* Iloprost (inhaled).
* Macitentan (oral).
* Riociguat (oral).
* Selexipag (oral).
* Sildenafil (oral).
* Tadalafil (oral).
* Treprostinil (oral, inhaled, infusion pump, IV).

#### **Side effects of PAH medication**

Common side effects of drugs that treat PAH include:

* Feeling lightheaded or like you might faint.
* Flushing (feeling warm in your head, neck or arms).
* Gastrointestinal symptoms like bloating, nausea, vomiting and diarrhea.
* Headache.
* Hypotension (low blood pressure).
* Pedal edema (swelling in your feet and ankles).
* Rash.
* Upper respiratory congestion.

Specific drugs may have additional side effects. Talk with your provider about how to manage any side effects. In some cases, your provider may need to adjust your dosage.

### **Can pulmonary arterial hypertension be reversed?**

Currently, medications can slow down PAH progression but not reverse the damage already done. However, researchers are working on promising new medications that could help reverse PAH. Such medications would repair damage to the endothelial cells that line your pulmonary arteries.

Talk with your provider to learn more about the latest research and clinical trials for PAH therapies.

## **Outlook / Prognosis**

Thanks to advances in treatment, people with PAH can live longer than ever before. Your life expectancy depends on many factors, including the severity of your condition and how early you’re diagnosed. Talk with your provider to learn your specific prognosis.

It’s important to keep up with your treatment and also closely follow your provider’s guidance. Steps you can take to improve your outlook include:

* Create an emergency kit. You need to have certain supplies and information with you all the time. Ask your provider what you should include in your kit, and never leave home without it.
* Get your seasonal vaccines as your provider recommends. It’s important to protect yourself against the flu and pneumonia.
* Keep all your follow-up appointments. Regular testing is crucial to check your lung and heart function and measure treatment progress.
* Take your medications as your provider prescribes, and at the same time every day. Don’t make any changes to your medication routine unless your provider tells you to do so.

## **Prevention**

Risk factors for developing PAH include:

* Connective tissue disease.
* Down syndrome.
* Family history of pulmonary hypertension.
* HIV.
* Use of diet medications such as “fen-phen” (dexfenfluramine and phentermine).
* Use of street drugs.

If someone in your biological family has PAH, ask your provider about genetic testing. If you’re diagnosed with PAH, you may want to tell your family members so they can consider genetic testing.

Some risk factors for PAH (like genetic mutations) are beyond your control. However, avoiding street drugs can help lower your risk of PAH and many other health problems. Also, talk with your provider before taking any diet pills.

Depending on your risk factors, your provider may recommend preventive screenings for PAH.

**Living With**

Follow your provider’s guidance for any lifestyle changes you should make. In general, some tips include:

* Avoid hot tubs, saunas and traveling to areas at a high altitude.
* Consider birth control options. Pregnancy can be dangerous for people with PAH. Talk with your provider if you’re planning a pregnancy or could become pregnant.
* Exercise and stay active as much as you can. Talk with your provider about which exercises are safe for you and how much activity you should aim to get each day. Check with your provider before starting any new exercise routine.
* Follow a heart-healthy diet. This includes eating foods low in saturated fat, trans fat and sodium.
* Quit smoking and using tobacco products. Also, avoid secondhand smoke.
* Seek support, and don’t try to handle your PAH journey alone. Ask your provider to recommend support groups and resources.

### **WHEN TO CALL A DOCTOR**

Call your provider if you’re having problems with:

* A fast heart rate (120 beats per minute).
* A respiratory infection or cough that’s getting worse.
* Constantly feeling dizzy or lightheaded.
* Episodes of chest pain or discomfort with physical activity.
* Extreme fatigue or decreased ability to do your normal activities.
* Nausea or lack of appetite.
* Restlessness or confusion.
* Shortness of breath that’s gotten worse, especially if you wake up feeling short of breath.
* Swelling in your ankles, legs or stomach that’s gotten worse.
* Trouble breathing with regular activities or at rest.
* Weight gain (2 pounds in one day or 5 pounds in one week).

Go to the emergency department or call your local emergency number if you have:

* A fast heart rate (120-150 beats per minute) that won’t go down.
* Fainting spells with loss of consciousness.
* Complications with your IV or infusion pump. These include infection, catheter displacement, solution leak, bleeding and IV pump malfunction.
* Shortness of breath that doesn’t go away when you rest.
* Sudden and severe chest pain.
* Sudden and severe headache.
* Sudden weakness or paralysis in your arms or legs.

## **Differential Diagnoses**

* Aortic Stenosis
* Atrial Myxoma
* Atrial Septal Defect
* Chronic Obstructive Pulmonary Disease (COPD)
* Chronic Pulmonary Embolism
* Dilated Cardiomyopathy (DCM)
* Emphysema
* Hepatopulmonary Syndrome
* Hypertrophic Cardiomyopathy
* Interstitial Lung Disease
* Mitral Regurgitation
* Mitral Stenosis
* Obesity-Hypoventilation Syndrome
* Obstructive Sleep Apnea (OSA)
* Restrictive Cardiomyopathy
* Restrictive Lung Disease
* Ventricular Septal Defects

## **Epidemiology**

The overall prevalence of pulmonary arterial hypertension (PAH) is difficult to determine given the disease’s heterogeneity and likely underdiagnosis.

Worldwide, schistosomiasis is likely the most prevalent cause of PAH,with studies suggesting that over 7% of patients with hepatosplenic schistosomiasis have pulmonary hypertension.However, data registries in countries most burdened by schistosomiasis-related PAH are limited.

Data registries in areas without endemic schistosomiasis such as the United States and Europe report a PAH prevalence ranging from 6.6-26 cases per million adults.The majority of these cases are idiopathic. While approximately 10% are classified as heritable, it is likely that this number will increase with time as genetic testing becomes more widespread.

Studies have also estimated the prevalence of specific subgroups of PAH. An observational study of 277 patients with HIV infection found that 0.46% of patients had pulmonary hypertension.In comparison with prior studies, no change in prevalence rate was seen with modern highly active antiretroviral treatment (HAART). In scleroderma patients, the incidence has been estimated to be 6-60% of all patients, with the variance based on the extent of disease.

Women are more likely to have PAH, with registries reporting a 65-80% female predominance of the disease.Interestingly, while prior studies suggested a mean age of diagnosis in the thirties, current registries suggest a mean age of diagnosis in the fifties.Although PAH can affect all races, data from the US REVEAL registry suggest a white predominance (73% white vs 12% African American, 9% Latino, and 3% Asian)

## **Staging**

pulmonary hypertension may be divided into the following functional classes:

* Class I: These are patients with pulmonary hypertension but without resulting limitation of physical activity. Ordinary physical activity does not cause undue dyspnea or fatigue, chest pain, or near-syncope in patients.
* Class II: These are patients with pulmonary hypertension resulting in slight limitation of physical activity. The patients are comfortable at rest, but ordinary physical activity causes undue dyspnea or fatigue, chest pain, or near-syncope.
* Class III: These are patients with pulmonary hypertension resulting in marked limitation of physical activity. Patients are comfortable at rest, but even less-than-ordinary activity causes undue dyspnea or fatigue, chest pain, or near-syncope.
* Class IV: These are patients with pulmonary hypertension who are unable to perform any physical activity without symptoms. These patients manifest signs of right-sided heart failure, dyspnea or fatigue may even be present at rest, and discomfort is increased by any physical activity.

## **Guidelines Summary**

Among the key changes were a lowered cutoff from mean pulmonary arterial pressure (mPAP) >25 mm Hg. Pulmonary hypertension (PH) is now defined by a mPAP >20 mm Hg at rest. The definition of pulmonary arterial hypertension (PAH) also implies a pulmonary vascular resistance (PVR) >2 Wood Units and pulmonary arterial wedge pressure ≤15 mm Hg.

### Diagnosis

The diagnostic algorithm was also restructured so that the diagnosis is made earlier. The following three steps are now included in the algorithm:

* Step 1: Suspicion by a general practitioner or other primary care provider.
* Step 2: Detection by echocardiography.
* Step 3: Confirmation with right heart catheterization in a pulmonary hypertension center. If the patient probably has intermediate or high-risk pulmonary hypertension, then he or she should be referred to a pulmonary hypertension center for a comprehensive diagnostic workup and invasive assessment, as needed.

### Risk Assessment

Risk assessment based on a multiparametric approach using a three-strata model to classify patients at low, intermediate, or high risk of death has been updated with 1 year mortality rates in the intermediate- and high-risk groups increased to reflect new data. The estimated 1-year mortality rates are as follows:

* Low Risk: < 5%
* Intermediate-risk: 5%-20%
* High risk: >20%.

Key recommendations for evaluating PAH disease severity and risk of death include:

* PAH severity should be evaluated using data derived from clinical assessment, exercise tests, biochemical markers, echocardiography and hemodynamic evaluation
* At diagnosis, the three-strata model taking into account all available data including hemodynamics should be used to classify patients at low, intermediate, or high risk of death; during follow-up, a four-strata model (low, intermediate-low, intermediate-high, high) should be used based on World Health Organization functional class (WHO-FC), 6-minute walking distance (6MWD) and brain natriuretic peptide/N-terminal pro-brain natriuretic peptide (BNP/NT-proBNP) along with any needed additional variables as needed.
* Obtaining and maintaining a low-risk profile is a treatment goal

### Treatment

Although PAH may be diagnosed in patients with mPAP >20 mmHg and PVR >2 WU, there is no data are available for the efficacy of drugs approved for PAH in patients whose mPAP is < 25 mmHg and whose PVR is < 3 WU. Thus, the guidelines recommend treatment be initiated in patients with PAH and mPAP >25 mm Hg.

General PAH treatment recommendations include the following [[22](javascript:void(0);)] :

* Patients receiving medical therapy should receive supervised exercise training
* Psychosocial support should be provided
* Patients should receive vaccinations against influenza, *Streptococcus pneumoniae*, and SARS-CoV-2
* Patients with signs of RV failure and fluid retention should receive diuretic therapy
* Patients with arterial blood oxygen pressure < 8 kPa (60 mmHg) should receive long-term oxygen therapy
* Iron-deficiency anemia should be treated
* Anticoagulation is not routinely recommended but may be considered in select patients
* The use of ACEis, ARBs, ARNIs, SGLT-2is, beta-blockers, or ivabradine is not recommended in the absence of comorbidities (*i.e.* high blood pressure, coronary artery disease, left HF, or arrhythmias)
* Patients with arterial blood oxygen pressure < 8 kPa (60 mmHg) at sea level or those using oxygen should receive oxygen administration while in-flight

Criteria lung transplantation evaluation referral in patients with PAH:

* PAH intermediate–high or high risk of 1 year mortality or REVEAL risk score >7 while on appropriate PAH medication
* Progressive disease or recent hospitalization for worsening PAH
* Need for IV or subcutaneous prostacyclin
* Known or suspected high-risk variant, systemic sclerosis or large and progressive pulmonary artery aneurysms
* Signs of secondary liver or kidney disfunction or other life-threatening complications

Guidelines criteria lung transplantation listing in patients with PAH are as follows:

* Patient has received full evaluation and is prepared for transplantation
* PAH high risk of 1 year mortality or REVEAL risk score >10 while on appropriate PAH medication, including IV or subcutaneous prostacyclin
* Progressive hypoxemia, especially in patients with pulmonary veno-occlusive disease (POVD) or pulmonary capillary hemangiomatosis (PCH)
* Progressive but not yet end-stage liver or kidney dysfunction or life-threatening hemoptysis

## **PREDEFINED QUESTIONS AND ANWERS**

## 1. What is pulmonary arterial hypertension (PAH)?

Answer:  
PAH is a type of high blood pressure that affects the arteries in your lungs and the right side of your heart. It occurs when the small arteries in the lungs become narrowed, blocked, or destroyed, making it harder for blood to flow through. This increases pressure in the pulmonary arteries and forces the heart to work harder.

## 2. What causes PAH?

Answer:  
PAH can be idiopathic (no known cause), hereditary (genetic mutations such as BMPR2), or associated with other conditions like connective tissue diseases (e.g., scleroderma), HIV infection, congenital heart disease, or certain drugs and toxins.

## 3. What are the common symptoms of PAH?

Answer:  
Symptoms often include shortness of breath, fatigue, chest pain, palpitations, dizziness, and swelling in the legs or abdomen. Symptoms usually worsen gradually and may initially occur only during physical activity.

## 4. How is PAH diagnosed?

Answer:  
Diagnosis involves multiple tests:

* Echocardiogram: to estimate pulmonary artery pressure and evaluate heart function.
* Right heart catheterization: the gold standard to measure pulmonary artery pressures directly.
* Pulmonary function tests, blood tests, chest imaging, and exercise tests help identify causes and assess severity.

## 5. What treatments are available for PAH?

Answer:  
Treatment depends on severity and cause but may include:

* Medications: endothelin receptor antagonists, phosphodiesterase-5 inhibitors, prostacyclin analogues, and soluble guanylate cyclase stimulators.
* Supportive care: oxygen therapy, diuretics for fluid retention, and anticoagulants if indicated.
* Advanced options: lung transplantation or surgical procedures for specific cases like chronic thromboembolic disease.

## 6. Are there side effects of PAH medications?

Answer:  
Yes, common side effects include headache, flushing, nausea, jaw pain, swelling, and low blood pressure. Your healthcare team will monitor and manage side effects carefully.

## 7. Can I exercise with PAH?

Answer:  
Moderate, supervised exercise is generally encouraged to maintain strength and quality of life. However, strenuous activity or overexertion should be avoided. Pulmonary rehabilitation programs can help tailor safe exercise plans.

## 8. How often will I need follow-up and testing?

Answer:  
Regular follow-up every 3 to 6 months is typical, including repeat echocardiograms and clinical assessments. Right heart catheterization may be repeated if symptoms worsen or treatment changes.

## 9. Is PAH hereditary? Should my family be tested?

Answer:  
Some forms of PAH are hereditary, especially those involving BMPR2 gene mutations. Genetic counseling and testing may be recommended for family members if a hereditary form is suspected

**DOCTOR PATIENT CONVERSATION**

Doctor: Hello! How have you been feeling since your last visit?

Patient: I’ve been feeling more tired and short of breath lately, especially when I walk or do simple chores.

Doctor: I’m sorry to hear that. These symptoms are common in PAH, and it’s important we address them. Can you tell me more about how your daily activities have been affected?

Patient: It’s been harder to keep up with my usual routine. Sometimes I feel dizzy, and I worry about fainting.

Doctor: That’s understandable. PAH causes increased pressure in the blood vessels of your lungs, making your heart work harder. This can cause symptoms like breathlessness, fatigue, and dizziness.

Patient: Are there treatments that can help me feel better?

Doctor: Yes. We have several medications that can reduce the pressure in your lungs and improve your heart function. For example, drugs like endothelin receptor antagonists or phosphodiesterase-5 inhibitors. Some patients also use pumps that deliver medication continuously.

Patient: Will I need to take these medicines for life?

Doctor: PAH is a chronic condition, so treatment is ongoing. Our goal is to control your symptoms, slow disease progression, and improve your quality of life. We will monitor you closely and adjust treatment as needed.

Patient: Are there side effects I should watch for?

Doctor: Each medication has potential side effects, such as headache, flushing, or swelling. We will discuss these in detail and monitor you regularly to manage any issues.

Patient: What about exercise? Can I still be active?

Doctor: Moderate, supervised exercise is beneficial and encouraged. We’ll tailor an activity plan that suits your abilities and helps maintain your strength without overexertion.

Patient: Should I see a specialist for this?

Doctor: Yes, managing PAH is complex and is best done with a team experienced in this condition. I’ll refer you to a pulmonary hypertension center where you can get comprehensive care.

Patient: Is there anything I can read or websites you recommend?

Doctor: Absolutely. The Pulmonary Hypertension Association website (phassociation.org) and the American Lung Association (lung.org) have excellent resources. I’ll also give you printed materials to take home.

Patient: Thank you. It helps to know there are options and support.

Doctor: You’re welcome. Remember, we’re here to support you. Please reach out anytime you have questions or concerns.

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**HEREDITARY HEMORRHAGIC TELANGIECTASIA**

**DEFINITION AND DESCRIPTION**

Hereditary hemorrhagic telangiectasia (tuh-lan-jee-uk-TAY-zhuh) is a condition that's passed through families, called inherited. It causes atypical links between arteries and veins called arteriovenous malformations (AVMs). The most common sites AVMs affect are the skin, nose, digestive system, lungs, brain and liver.

AVMs may get larger over time. They can bleed or burst. This can result in serious complications, including death.

Nosebleeds that happen for no known reason are the most common symptom. Nosebleeds can happen every day. Ongoing bleeding from the nose and the intestinal tract can result in serious iron deficiency anemia and poor quality of life.

Also called Osler-Weber-Rendu disease and HHT, hereditary hemorrhagic telangiectasia passes from parents to children. How bad it is can vary greatly from person to person, even within the same family.

If you have HHT and have children, you may want to have them checked for the condition. HHT can affect them even if they don't have symptoms.

### **Causes hereditary hemorrhagic telangiectasia (HHT)**

HHT is genetic, meaning it’s passed down from parents to children. It’s a dominant disorder, caused by one copy of one abnormal gene from one parent. Hundreds of possible mutations in six different genes have been linked to HHT, but the vast majority of cases are due to mutations in two genes, *ENG* and *ACVRL1*. Scientists are still studying the mutations and genes involved.

### **Symptoms of hereditary hemorrhagic telangiectasia (HHT)**

The symptoms of HHT vary from person to person, depending on where abnormal blood vessels develop in the body. Some people may have no significant signs, but others may develop very serious symptoms.

The most common symptom is frequent nosebleeds (epistaxis).

Some people with HHT also might have delicate red spots on certain parts of the body. They may get lighter when you press on them and are common on the:

* Face.
* Fingers or fingertips.
* Hands.
* Lining of the mouth.
* Lips.
* Nose.

Some people with HHT may also have:

* Anemia (not enough red blood cells).
* Bleeding in the stomach or intestines.

People with HHT may develop abnormalities in larger blood vessels, called arteriovenous malformations (AVMs). AVMs can form in the lungs, brain, spinal cord and liver, and they can cause:

* Blue discoloration of the skin.
* Coughing up blood (hemoptysis).
* Fatigue.
* Headaches.
* Trouble breathing (dyspnea).

Rare but serious complications can occur when an AVM hemorrhages, such as:

* Dizziness, double vision, seizures and strokes, if the condition affects blood vessels in the brain.
* Heart failure, as the heart works harder to provide blood throughout the body, if HHT affects the liver.
* Back pain or numbness in the arms or legs, if HHT affects the spine.

If you have HHT, there is a 50% chance for each child to inherit it.

**Risk factors**

The major risk factor for hereditary hemorrhagic telangiectasia is having a parent with the condition.

## **Diagnosis**

Your healthcare professional may diagnose HHT based on a physical exam, results of imaging tests and a family history. But some symptoms may not show up in children or young adults. Having genetic testing for HHT may confirm the diagnosis.

### **Imaging tests**

In HHT, atypical links called arteriovenous malformations, also called AVMs, happen between arteries and veins. HHT AVMs can be present in internal organs such as the lungs, brain and liver. One or more of the following imaging tests can help find AVMs:

* **Ultrasound.** This test can show whether the AVMs affect the liver.
* **MRI.** This scan can check for AVMs in the brain as well as the liver and other organs in the belly.
* **Echocardiogram bubble study.** During this echocardiogram test, a healthcare professional puts a line in a vein, called an IV. A small amount of air bubbles put into the IV lets the healthcare professional find and assess any lung AVMs.
* **CT scan.** These can confirm AVMs in the lungs, the liver and other organs in the belly.

**Treatment**

If you or your child has HHT, if you can, seek treatment at an HHT Center of Excellence. HHT is a rare condition that is best managed at centers that treat all aspects of this condition at every age. So it can be hard to find a specialist to treat it.

In the United States, Cure HHT names HHT Centers of Excellence for being able to diagnose and treat all aspects of the condition. Mayo Clinic is an HHT Center of Excellence and cares for many people and their family members diagnosed with HHT.

### **Medications**

Medicines that help stop the bleeding linked with HHT can be divided into three broad groups:

* **Hormone-related drugs.** Medicines that have estrogen can be helpful. But side effects are common with the high doses needed. Anti-estrogens such as tamoxifen (Soltamox) and raloxifene (Evista) also can control HHT.
* **Medicines that block blood vessel growth.** One treatment for HHT is bevacizumab (Avastin). Avastin goes through a tube in a vein, called intravenous. Other medicines healthcare professionals use to block blood vessel growth include pazopanib (Votrient), pomalidomide (Pomalyst) and tacrolimus (Prograf, Protopic, others).
* **Medicines that slow clot dissolving.** Tranexamic acid (Cyklokapron, Lysteda) can help stop serious bleeding in emergencies. If taken regularly, it may help prevent bleeding.

If you get iron deficiency anemia, you might get an iron replacement through a vein. This most often works better than taking iron pills.

### **Surgical and other procedures for the nose**

Serious nosebleeds are one of the most common signs of HHT. These sometimes happen daily. They can cause so much blood loss that you become anemic. You might need to receive blood, called a transfusion, and iron through an arm vein.

Procedures to lower the number of nosebleeds and lessen how bad they are may include:

* **Ablation.** This procedure uses energy from lasers or other devices to seal the vessels that cause the nosebleeds. But this most often is short-lived. The nosebleeds come back over time.
* **Skin graft.** Skin from another part of the body can be put inside the nose. The skin most often comes from the thigh. Healthcare professionals rarely do this procedure anymore because of how well newer medicines work.
* **Surgically closing the nostrils.** If nothing else works, joining flaps of skin within the nose to close the nostrils often is a success. This is done only when other treatments have failed. Healthcare professionals rarely do this procedure anymore because of how well newer medicines work.

### **Surgical and other procedures for the lungs, brain and liver**

HHT most often affects the lungs, brain and liver. Procedures to treat AVMs in these organs may include:

* **Embolization.** In this procedure, a healthcare professional threads a long, slender tube through the blood vessels to the AVM. Then the health professional puts in a plug or a metal coil to block blood from entering the AVM. This shrinks and heals the AVM over time. Embolization treats lung and brain AVMs, but not liver AVMs.
* **Surgical removal.** Rarely, the best way to treat certain AVMs in the brain or the lungs is to remove them with surgery.
* **Stereotactic radiotherapy.** This procedure treats AVMs in the brain. It uses beams of radiation that come from different directions. They meet at the AVM to treat it.
* **Liver transplant.** Rarely, treatment for AVMs in the liver is a liver transplant.

**Drug treatments for Hereditary Hemorrhagic Telangiectasia (HHT) and their common side effects**

## 1. Antifibrinolytic Drugs

* Examples: Tranexamic acid, Aminocaproic acid
* Use: Help reduce bleeding by preventing clot breakdown, often used for nosebleeds and gastrointestinal bleeding.
* Side Effects:
  + Risk of thrombosis (blood clots)
  + Gastrointestinal upset (nausea, diarrhea)
  + Rare allergic reactions

## 2. Antiangiogenic Drugs

* Examples: Bevacizumab (Avastin), Pomalidomide, Pazopanib
* Use: Inhibit abnormal blood vessel growth and reduce bleeding severity. Bevacizumab is given intravenously; pomalidomide is oral.
* Side Effects:
  + Bevacizumab: Hypertension, proteinuria, impaired wound healing, increased risk of bleeding or thrombosis
  + Pomalidomide: Fatigue, constipation, risk of blood clots, neutropenia (low white blood cells)
  + Pazopanib: Hypertension, liver toxicity, diarrhea, fatigue

## 3. Hormone-Related Drugs

* Examples: Estrogen-based therapies, Tamoxifen, Raloxifene
* Use: Used to reduce epistaxis frequency and severity by stabilizing blood vessels.
* Side Effects:
  + Estrogens: Risk of thromboembolism, breast tenderness, nausea
  + Tamoxifen/Raloxifene: Hot flashes, risk of blood clots, uterine changes (rare)

## 4. Immunosuppressants / Immunomodulators

* Examples: Sirolimus, Tacrolimus
* Use: Emerging therapies that may reduce bleeding by modulating vascular inflammation and repair.
* Side Effects:
  + Sirolimus: Mouth ulcers, increased infection risk, hyperlipidemia
  + Tacrolimus: Kidney toxicity, hypertension, tremors

## 5. Antibiotics

* Example: Doxycycline
* Use: Sometimes used for anti-inflammatory effects or infection prevention in HHT patients.
* Side Effects:
  + Photosensitivity
  + Gastrointestinal upset
  + Rare allergic reactions

**Lifestyle and home remedies**

To help prevent HHT nosebleeds, you may want to:

* **Not use certain medicines.** Your risk of bleeding can be higher from using certain medicines and drugs you get without a prescription. These include aspirin, ibuprofen (Advil, Motrin IB, others), fish oil supplements, ginkgo and St. John's wort.
* **Not eat certain foods.** In some people, having blueberries, red wine, dark chocolate or spicy foods can cause HHT nosebleeds. Try keeping a food diary to see if there's any link between what you eat and how bad your nosebleeds are.
* **Keep your nose moist.** Use saline sprays, lotions or gels that add moisture to help lower the risk of bleeding. Using a bedside humidifier overnight also is helpful.
* **Not do heavy lifting.** Bending over and lifting heavy objects can cause nosebleeds.

## **Outlook / Prognosis**

People with HHT have an almost average life expectancy. But AVMs in the lungs and brain and chronic bleeding are serious and should be treated.

**Prevention**

There is no way to prevent HHT or reduce your risk of getting it. But tell your healthcare provider if your parent, sibling or child has it. That may help you catch it early and prevent complications.

**PREDEFINED QUESTIONS AND ANSWERS**

## 1. Should anyone in my family get tested?

Yes. Because HHT is an autosomal dominant genetic disorder, all first-degree relatives (parents, siblings, children) of a person diagnosed with HHT should be offered genetic testing or clinical screening, even if they have no symptoms. Genetic testing helps identify family members who carry the mutation early, allowing timely monitoring and treatment to prevent complications.

## 2. Can I play sports?

Generally, you can participate in most sports and physical activities, but it is important to avoid activities with a high risk of trauma or injury that could cause bleeding, especially if you have fragile blood vessels or a history of nosebleeds or arteriovenous malformations (AVMs). Discuss your specific situation with your healthcare provider to tailor recommendations.

## 3. Should I avoid any particular activities?

* Avoid activities that increase the risk of head trauma or severe nosebleeds, such as contact sports or heavy weightlifting.
* Avoid high-altitude exposure without medical advice, as low oxygen levels can worsen AVMs.
* Avoid activities that increase bleeding risk if you have known AVMs or bleeding tendencies.

## 4. Should I avoid alcohol, certain foods or any medications?

* Alcohol can dilate blood vessels and may worsen nosebleeds, so moderation or avoidance is advised if you have frequent epistaxis.
* Avoid medications that increase bleeding risk such as aspirin, nonsteroidal anti-inflammatory drugs (NSAIDs), and anticoagulants unless prescribed and closely monitored by your doctor.
* No specific food restrictions are generally required, but maintaining good nutrition and hydration supports vascular health.

## 5. What can I do to prevent or stop nosebleeds?

* Keep nasal mucosa moist using saline sprays, gels, or humidifiers.
* Avoid nasal trauma (e.g., nose picking).
* Use gentle nasal hygiene.
* In recurrent or severe cases, treatments such as laser therapy, cauterization, or topical medications may be recommended by an ENT specialist.
* Some patients benefit from systemic therapies like antifibrinolytics or antiangiogenic drugs under specialist care.

## 6. Is it safe for me to get pregnant?

Pregnancy in women with HHT carries increased risks due to potential complications from AVMs (especially in the lungs and brain) and bleeding. It requires specialized multidisciplinary care with close monitoring by obstetricians experienced in high-risk pregnancies and HHT specialists. Pre-pregnancy screening for AVMs is strongly recommended.

## 7. When should I seek medical attention for bleeding?

Seek urgent medical care if you experience:

* Severe or uncontrollable nosebleeds lasting more than 20 minutes despite first aid.
* Signs of significant blood loss such as weakness, dizziness, fainting, or rapid heartbeat.
* Bleeding from the gastrointestinal tract (vomiting blood or black stools).
* Symptoms suggestive of AVM complications like sudden severe headache, chest pain, shortness of breath, or neurological symptoms

## **Diagnostic Considerations**

The diagnosis of Osler-Weber-Rendu disease (OWRD; i.e., hereditary hemorrhagic telangiectasia [HHT]) for the purposes of improving patient care and standardizing research.These criteria are as follows:

* Epistaxis - Spontaneous and recurrent
* Telangiectases - Multiple characteristic sites (e.g., lips, oral cavity, fingers, or nose)
* Visceral lesions - Gastrointestinal (GI) telangiectasia (with or without bleeding), pulmonary arteriovenous malformation (AVM), hepatic AVM, cerebral AVM, and spinal AVM
* Family history - A first-degree relative who has HHT (according to these same criteria)

The HHT diagnosis is classified as definite if three criteria are present, possible or suspected if two criteria are present, and unlikely if fewer than two criteria are present. There is no firm consensus on the number of episodes or degree of epistaxis necessary for diagnosis; according to the Curaçao criteria, nosebleeds should occur spontaneously on more than one occasion, and night-time bleeding should be considered especially suspicious.

Early diagnosis of family members or confirmation with genetic testing of patients who meet the Curaçao criteria may assist in the identification of those patients who are most at risk for specific sequelae.

In addition to the conditions listed in the differential diagnosis, other problems to be considered include the following:

* Cockayne syndrome
* Louis-Bar syndrome
* Essential telangiectasia
* Actinically damaged skin (actinic keratosis)
* Scleroderma

## **Differential Diagnoses**

* Ataxia-Telangiectasia
* CREST Syndrome
* Pediatric Syphilis
* Rosacea
* Rothmund-Thomson Syndrome

## **Epidemiology**

### United States statistics

OWRD (ie, HHT) is rare in North America. The reported incidence is 1-2 cases per 100,000 population annually. The overall prevalence is estimated to be approximately 1-2 cases per 10,000 population. However, the prevalence may be underestimated because many cases may be asymptomatic.In Vermont, the frequency has been estimated at 1 case per 16,500 population.The disease has a clinical penetrance of 97%.

### International statistics

The worldwide prevalence is 1 case per 5000-10,000 population In Europe and Japan; the frequency is estimated to be between 1 in 5000 to 8000 people. The prevalence of HHT in a Danish population increased from 13.8 cases per 100,000 population in 1974 to 15.6 cases per 100,000 population in 1995.

The frequency may vary considerably between populations. The highest rates are seen in parts of the Dutch Antilles among the Afro-Caribbean population, with a prevalence of between 1 case per 200 persons and 1 per 1331 persons in the Curaçao and Bonaire regions.In the French department of Ain, the prevalence is 1 case per 2351 persons; in France overall, it is 1 per 8345.Other examples include the Danish island of Funen (1 per 3500) and northern England (1 per 39,216).

### Age-, sex-, and race-related demographics

HHT may occur in children, in whom a tendency to bleed may be the first symptom. However, it is far more common during puberty or adulthood. The syndrome most often manifests by the second or third decade of life, though it may also be clinically silent. Pulmonary AVMs may be congenital and therefore may present within the first year of life. The risk of GI tract bleeding increases in patients older than 50 years.

HHT occurs with equal frequency and severity in males and females.Although it most commonly occurs in whites, it has a wide geographic distribution and has been described in people of Asian, African, and Arabic descent.

## **Genetic Testing**

The sensitivity of molecular diagnosis is highest in probands with a clinically confirmed diagnosis of HHT.However, a substantial fraction of probands do not carry an identifiable mutation in the coding exons of either of the two responsible genes, *ENG* and *ALK1*. Targeted family-specific mutation analysis for *ENG* exon deletions could lead to misdiagnosis of some affected family members with HHT, as was illustrated by the findings of a study in which two distinct *ENG* deletions were found in a single family

## **Hereditary Hemorrhagic Telangiectasia Guidelines**

Guidelines for the management and prevention of HHT-related symptoms and complications, were published.The guidelines included the following recommendations.

### **Management of epistaxis**

To reduce HHT-related epistaxis, use moisturizing topical therapies that humidify the nasal mucosa.

Consider oral tranexamic acid (TXA) for epistaxis that does not respond to moisturizing topical therapies.

Consider ablative therapies (eg, laser treatment, radiofrequency ablation [RFA], electrosurgery, and sclerotherapy) for nasal telangiectasias that have not responded to moisturizing topical therapies.

Consider systemic antiangiogenic agents for epistaxis that has not responded to moisturizing topical therapies, ablative therapies, or TXA.

Consider septodermoplasty for epistaxis that has not responded sufficiently to moisturizing topical therapies, ablative therapies, or TXA.

Consider nasal closure for epistaxis that has not responded sufficiently to moisturizing topical therapies, ablative therapies, or TXA.

### **Management of gastrointestinal bleeding**

Esophagogastroduodenoscopy (EGD) is recommended as the first-line diagnostic test for suspected HHT-related bleeding. Patients who meet colorectal cancer screening criteria and patients with SMAD4-HHT (genetically proven or suspected) should also undergo colonoscopy.

Consider capsule endoscopy for suspected HHT-related bleeding when EGD does not reveal significant HHT-related telangiectasia.

Grade the severity of HHT-related gastrointestinal (GI) bleeding according to the following proposed framework: *mild* (hemoglobin [Hb] goals [reflective of age, gender, symptoms, and comorbidities] met with oral iron replacement); *moderate* (Hb goals met with intravenous [IV] iron treatment); or *severe* (Hb goals not met despite adequate iron replacement, or blood transfusions needed).

Use endoscopic argon plasma coagulation only sparingly during endoscopy.

Consider treating mild HHT-related GI bleeding with oral antifibrinolytics.

Consider treating moderate-to-severe HHT-related GI bleeding with IV bevacizumab or other systemic antiangiogenic therapy.

### **Anemia and anticoagulation**

Test for iron deficiency and anemia in all adult HHT patients, regardless of symptoms, and in all pediatric HHT patients with recurrent bleeding and/or symptoms of anemia.

Provide iron replacement for treatment of iron deficiency and anemia as follows: initial therapy with oral iron; IV iron replacement for patients in whom oral iron is not effective, not absorbed, or not tolerated or who are presenting with severe anemia.

Provide red blood cell (RBC) transfusions in the following settings: hemodynamic instability/shock; comorbidities requiring a higher Hb target; need to increase Hb acutely (eg, before surgery or during pregnancy); and inability to maintain adequate Hb despite frequent iron infusions.

Consider evaluation for additional causes of anemia in the setting of an inadequate response to iron replacement.

Provide HHT patients with anticoagulation (prophylactic or therapeutic) or antiplatelet therapy when there is an indication, with individualized bleeding risks taken into consideration; bleeding in HHT is not an absolute contraindication for these therapies.

Where possible, avoid the use of dual antiplatelet therapy (DAPT) and/or a combination of antiplatelet therapy and anticoagulation.

### **Liver vascular malformations**

Offer screening for liver vascular malformations (VMs) to adults with definite or suspected HHT.

Perform diagnostic testing for liver VMs in HHT patients with symptoms and/or signs suggestive of complicated liver VMs, using Doppler ultrasonography (US), multiphase contrast computed tomography (CT), or contrast abdominal magnetic resonance imaging (MRI).

Provide intensive first-line management only for patients with complicated and/or symptomatic liver VMs, and tailored such management to the type of liver VM complication(s).

It is recommended that HHT patients with high-output cardiac failure and pulmonary hypertension be comanaged by the HHT Center of Excellence and an HHT cardiologist or a pulmonary hypertension specialty clinic.

Estimate the prognosis of liver VMs using available predictors so as to identify patients in need of closer monitoring.

Consider IV bevacizumab for patients with symptomatic high-output cardiac failure due to liver VMs who have not responded sufficiently to first-line management.

Refer patients with symptomatic complications of liver VMs (eg, refractory high-output cardiac failure, biliary ischemia, or complicated portal hypertension) for consideration of liver transplantation.

### **Pediatric care**

Offer diagnostic genetic testing for asymptomatic children of a parent with HHT.

Screen for pulmonary arteriovenous malformations (AVMs) in asymptomatic children with HHT or at risk for HHT at the time of presentation/diagnosis.

Treat large pulmonary AVMs and pulmonary AVMs associated with reduced oxygen saturation in children.

Repeat pulmonary AVM screening in asymptomatic children with or at risk for HHT, typically at 5-year intervals.

Screen for brain VMs in asymptomatic children with HHT or at risk for HHT at presentation/diagnosis.

Treat brain VMs with high-risk features.

### **Pregnancy and delivery**

Discuss preconception and prenatal diagnostic options, including preimplantation genetic diagnosis, with HHT-affected individuals.

Perform testing with unenhanced MRI in pregnant women with symptoms suggestive of brain VMs.

For pregnant women with HHT without recent screening and/or treatment for pulmonary AVM:

* Asymptomatic – Perform initial pulmonary AVM screening with either agitated saline transthoracic contrast echocardiography (TTCE) or low-dose non contrast chest CT, depending on local expertise; chest CT, if performed, should be done early in the second trimester
* Symptoms suggestive of pulmonary AVM – Perform diagnostic testing with low-dose non contrast chest CT; this may be done at any gestational age, as clinically indicated

Treatment of pulmonary AVMs should start in the second trimester unless otherwise indicated.

It is recommended that pregnant women with HHT be managed at a tertiary care center by a multidisciplinary team if they have untreated pulmonary AVMs and/or brain VMs or have not been recently screened for pulmonary AVMs.

Do not withhold an epidural because of a diagnosis of HHT; screening for spinal VMs is not required.

Allow women with known non-high-risk brain VMs to labor and proceed with vaginal delivery; an assisted second stage may be required on a case-by-case basis.

**DOCTOR PATIENT CONVERSATION**

Doctor: Hello! I see from your records and symptoms that you may have Hereditary Hemorrhagic Telangiectasia, or HHT. Have you experienced frequent nosebleeds or unusual bleeding?

Patient: Yes, I’ve had nosebleeds since I was a teenager, and sometimes they seem to come out of nowhere. I also noticed some small red spots on my skin.

Doctor: Those red spots are called telangiectasias, which are small abnormal blood vessels common in HHT. The nosebleeds are also a classic symptom. HHT is a genetic condition that affects blood vessel formation in various parts of the body.

Patient: Is this something I was born with? Can it run in families?

Doctor: Yes, HHT is inherited in an autosomal dominant pattern, meaning if a parent has it, there is a 50% chance of passing it on. We recommend that your close family members get screened, even if they don’t have symptoms yet.

Patient: What kind of problems can HHT cause besides nosebleeds?

Doctor: HHT can cause abnormal connections between arteries and veins called arteriovenous malformations (AVMs) in organs like the lungs, liver, and brain. These can lead to serious complications such as bleeding, anemia, or even stroke if untreated.

Patient: How do you find out if I have these AVMs?

Doctor: We perform screening tests such as contrast echocardiography for lung AVMs, MRI for brain AVMs, and imaging for liver involvement. Early detection helps us manage and prevent complications.

Patient: What treatments are available?

Doctor: Treatment depends on your symptoms and which organs are affected. For nosebleeds, we use local therapies like humidification, nasal gels, or laser treatment. For more severe bleeding, medications such as antifibrinolytics or antiangiogenic drugs like bevacizumab may help reduce bleeding. AVMs may require embolization or surgery.

Patient: Are there risks with these treatments?

Doctor: Like all treatments, there are potential side effects. For example, antiangiogenic drugs can cause high blood pressure or impaired wound healing. We monitor you closely and tailor treatment to your needs.

Patient: Can I still live a normal life? Are there things I should avoid?

Doctor: Many people with HHT live normal lives with proper management. Avoiding nasal trauma, keeping nasal passages moist, and regular follow-up are important. Also, inform your healthcare providers about your condition before surgeries or dental work.

Patient: What about pregnancy? Is it safe?

Doctor: Pregnancy can increase risks, especially if you have untreated AVMs. If you plan to become pregnant, we recommend thorough evaluation and close monitoring by a multidisciplinary team.

Patient: Should my family get tested?

Doctor: Yes, we advise genetic counseling and testing for first-degree relatives. Early diagnosis allows preventive care to reduce complications.

Patient: Where can I learn more or get support?

Doctor: The Pulmonary Hypertension Association and Cure HHT websites offer excellent patient resources and support groups. I’ll also provide printed materials for you.

Patient: Thank you, doctor. It’s a relief to have a plan.

Doctor: You’re welcome. We’ll work together to manage your condition and maintain your quality of life. Please reach out anytime with questions or concerns.

REFERENCES

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### **Primary ciliary dyskinesia**

**DEFINITION AND DESCRIPTION**

Primary ciliary dyskinesia (PCD) affects your respiratory system and is a rare disorder. It’s due to issues with cilia, microscopic hair-like organs. Healthy cilia use wave-like motions to move cells and other substances within your body.

With primary ciliary dyskinesia, cilia may:

* Be the wrong size.
* Have an abnormal shape.
* Be missing.
* Move in an uncoordinated manner.
* Not move.

### **What do cilia do?**

Cilia line tissue in the respiratory system and other areas. Here, they help eliminate germs, waste and substances, like dust. They also play a role in the placement of developing organs in a fetis.

### **How can nonfunctioning cilia make me sick?**

They can cause health issues beginning at birth or later in life, such as:

* **Abnormal organ placement:** The heart, lungs or spleen may be facing the wrong direction. They may also be on the wrong side of your body.
* **Chronic, severe respiratory disease:** Nonfunctioning cilia make it challenging to clear mucus. Instead, it builds up in your lungs and other passageways, causing infections.
* **Fertility issues:** Men with PCD are infertile. Women may experience serious pregnancy complications, such as an ectopic pregnancy.

### **causes PCD**

Primary ciliary dyskinesia is passed down in families through gene abnormalities (mutations). There are dozens of mutations that can cause the disease.

### **Symptoms of primary ciliary dyskinesia**

Symptoms are often present at birth and worsen over time.

### **PCD symptoms at birth**

* Congenital heart disease.
* Cyst-like growths on organs, including the kidneys and pancreas.
* Difficulty breathing (respiratory distress).
* Heterotaxia, organs that are out of place, missing or not fully developed.
* Humoral (antibody) deficiency, which can contribute to lung and sinus infections.
* Lungs that cannot inflate properly (atelectasis).
* Nasal congestion.
* Situs inversus, organs that are a mirror image of where they should be.
* Wet cough that does not go away.

### **Ongoing primary ciliary dyskinesia symptoms**

* Chronic cough.
* Chronic sinusitis.
* Ear infections.
* Excess mucus and phlegm.
* Infertility.
* Fluid buildup in the brain (hydrocephalus).
* Nasal polyps.
* Severe pneumonia.
* Upper respiratory infections.

## **Diagnosis and Tests**

There is no single test that can confirm a PCD diagnosis. Evaluations include:

* **Physical exam** to determine whether medical history and symptoms are consistent with primary ciliary dyskinesia.
* **Biopsy** of tissue that contains cilia. Healthcare providers take a tissue sample from the nose or lungs and examine it under a microscope.
* **Genetic testing** to check for mutations associated with primary ciliary dyskinesia. The majority but not all people with PCD have one of these mutations.

Other tests that may indicate a primary ciliary dyskinesia diagnosis include:

* **Exhaled nasal nitric oxide:** A special device measures nitric oxide levels, a gas that’s present when you exhale. People with PCD have abnormally low levels.
* **Pulmonary function tests:** These tests evaluate how well your lungs work.
* **Video microscopy:** A healthcare provider views a sample of cilia through a microscope equipped with a high-powered video camera. Viewing video output in slow-motion enables healthcare providers to test cilia movement.

## **Management and Treatment**

There is no cure for PCD. But treatment can slow disease progression.

Therapies that help you eliminate mucus and fluids include:

* **Airway clearance:** A special machine loosens mucus. There are also special coughing techniques.
* **Chest physical therapy:** Some people wear a vest-like device that taps on their chest.
* **Ear tubes:** Surgeons implant small tubes in the eardrums. They make it easier to clear fluid from the middle ear, lowering the likelihood of buildups.

Care may also include medications for infections and inflammation:

* **Antibiotics:** These medications help your body fight infections. In severe cases, you may receive antibiotics through a vein in your arm (intravenously).
* **Azithromycin:** This medication helps treat the inflammation in your lung. You take it on a daily basis.
* **Bronchodilators:** Medications like albuterol make it easier to breathe.
* **Corticosteroids:** These medications lessen inflammation by quieting chemical reactions in your body.
* **Mucus thinners:** Medications you inhale to thin mucus in the airways.

Surgery may be necessary to treat abnormal organ placement. Care may include repairing abnormal structures shortly after birth. Additional procedures may be needed to prevent or treat complications later in life.

**Outlook / Prognosis**

Many people live a long life, even into older adulthood.

### **What is the prognosis for people with primary ciliary dyskinesia?**

The prognosis varies. Some people are sick at birth and remain so throughout their lives. Other people feel healthy between periods of illness that may sometimes be severe.

### **What are the potential complications of PCD?**

Ongoing infections from primary ciliary dyskinesia can scar organ tissue, leading to complications. These include:

* Hearing loss.
* Bronchiectasis.
* Respiratory failure.

**Prevention**

The condition is inherited, so there is nothing you can do to prevent it. If you are thinking of starting a family and a close relative has PCD, you may wish to consider genetic testing and counseling. These services help you learn the likelihood of having a child with the disease.

## **Living With**

Steps you can take to feel your best include:

* **Exercise:** Moderate physical activity makes you breathe harder, loosening mucus and improving physical strength.
* **Emotional support:** Living with a chronic condition such as PCD can bring challenges that are difficult to cope with. A mental health professional can provide you with the support that makes life a little less stressful.
* **Monitoring:** Healthcare providers assess whether treatments are working or if symptoms are worsening. Visits may include pulmonary function testing or a chest X-ray.

**DIFFERENTIAL DIAGNOSIS**

**Chronic sinopulmonary disease and bronchiectasis.** Like primary ciliary dyskinesia (PCD), the following disorders are associated with chronic sinopulmonary disease and bronchiectasis. Unlike PCD, these disorders are not associated with situs abnormalities:

* Cystic fibrosis
* Inborn errors of immunity. Overlap is likely if associated with respiratory manifestations. Immunodeficiencies such as immunoglobulin G (IgG) subclass deficiency, activated PI3K delta syndrome, and GATA2 deficiency may have low nasal nitric oxide levels in addition to overlapping clinical features with PCD. See the following Phenotypic Series for genes associated with primary immunodeficiency in OMIM:
  + Immunodeficiency (select examples)
  + Immunodeficiency with hyper-IgM
  + Immunodeficiency, common variable
  + Severe combined immunodeficiency (select examples)
* Bronchiectasis and nasal polyposis (OMIM 620984) may be seen in those with an autosomal recessive airway disease with respiratory symptoms and low nasal nitric oxide levels in individuals with biallelic *WFDC2* pathogenic variants .
* Mucin deficiency due to biallelic loss of *MUC5B*
* Asthma and/or allergic rhinitis
* Gastroesophageal reflux disease
* Wegener granulomatosis (upper- and lower-airway disease)

**Sinopulmonary disease.** Cri-du-chat syndrome (OMIM 123450) is caused by a segmental chromosome 5p deletion that usually includes *DNAH5* (a gene known to be associated with PCD). PCD co-occurs in cri-du-chat syndrome when a PCD-causing pathogenic variant is detected in the remaining allele of *DNAH5*. Cri-du-chat syndrome is associated with chronic sinopulmonary disease but not situs abnormalities.

**Situs abnormalities.** Multiple genes are associated with visceral heterotaxy for genes associated with this phenotype in OMIM). PCD is not likely if the affected individual has visceral heterotaxy but no associated respiratory manifestations.

**GENOMIC DATA**

a heterozygous pathogenic (or likely pathogenic) variant in *FOXJ1* or *TUBB4B*, or a hemizygous pathogenic (or likely pathogenic) variant in a male in *DNAAF6* (*PIH1D3*), *RPGR*, or *OFD1.*

Note: (1) Per ACMG/AMP variant interpretation guidelines, the terms "pathogenic variants" and "likely pathogenic variants" are synonymous in a clinical setting, meaning that both are considered diagnostic and can be used for clinical decision making to "pathogenic variants" in this *Gene Review* is understood to include likely pathogenic variants. (2) Identification of variant(s) of uncertain significance cannot be used to confirm or rule out the diagnosis.

### **Molecular Genetic Testing**

Molecular genetic testing approaches can include a combination of [**gene**](https://www.ncbi.nlm.nih.gov/books/n/gene/glossary/def-item/gene/)**-targeted testing** (multigene panel, targeted analysis) and **comprehensive** **genomic testing** (exome sequencing, genome sequencing). Gene-targeted testing requires that the clinician determine which gene(s) are likely involved, whereas comprehensive genomic testing does not require this.

## **Epidemiology**

### United States statistics

The prevalence of PCD is approximately 1:16,000 live births. Geographic area and consanguinity may affect the prevalence. Specific types of defects are consistent within individual families and appear to be genetically determined. Based on the autosomal recessive mode of inheritance, the probability of having subsequent children with PCD is 1:4.

### International statistics

The reported frequency is 1 per 26,000-40,000 live births. However, this is likely to be an underestimate because misdiagnosis is common.

A genetic database analysis by Hannah et al estimated that the overall minimum global prevalence of PCD is at least 1 in 7554 persons. The expected frequency of PCD is higher in persons of African ancestry than in most other populations.

### Race-, sex-, and age-related demographics

No racial predilection is reported.

No sex predilection is reported.

No particular age predilection is recognized; infants are born with this genetic disorder. Cases associated with dextrocardia and with respiratory symptoms are more likely to be diagnosed in early infancy.

## **Procedures**

Bronchoscopy reveals mucosal inflammation and mucopurulent secretions. It can also be used to confirm the reversal of bronchial anatomy in those patients with situs inversus.

Examination of the ciliary ultrastructure by electron microscopy in a nasal or bronchial ciliary biopsy sample can be used as a diagnostic test.

Nasal biopsy (brush or curettage) samples are obtained from inferior surface of turbinates. Electron microscopy reveals the abnormalities in the cilia.

Bronchial brush biopsy demonstrates ciliary ultrastructure abnormalities using an electron microscope. Due to the varying orientation of the cilia in a biopsy specimen and resultant technical difficulties in the full analysis of cilia, a quantitative method includes assessing axonemal defects in less than perfectly oriented cilia, with dynein arms being assessed only in those cilia in which these small structures can be discerned.

A review of quantitative transmission electron microscopy in 1182 patients referred for ciliary structure analysis reported confirmation of diagnosis of PCD in 242 (20%) cases.In addition to describing an algorithm including screening tests such as exhaled nasal nitric oxide, saccharine test, light microscopy, and electron microscopy, the authors describe the use of transmission electron microscopy using a rapid quantitative method. However, electron microscopy does not always exclude the diagnosis of PCD.

**PREDEFINED Q AND A**

## 1. What is Primary Ciliary Dyskinesia (PCD)?

Answer:  
PCD is a rare inherited disorder caused by defects in the structure or function of cilia—tiny hair-like structures that move mucus and debris out of the lungs, sinuses, and ears. When cilia don’t work properly, mucus builds up, leading to recurrent infections and inflammation in the respiratory tract.

## 2. What are the common symptoms of PCD?

Answer:  
Symptoms often start early in life and include:

* Chronic wet cough
* Persistent nasal congestion or runny nose
* Recurrent ear infections and hearing problems
* Recurrent sinus infections
* Respiratory distress in newborns
* Chronic bronchitis or pneumonia
* About half of patients have situs inversus (organs reversed in the chest/abdomen).

## 3. How is PCD diagnosed?

Answer:  
Diagnosis involves a combination of:

* Measuring nasal nitric oxide (usually very low in PCD)
* High-speed video microscopy to assess ciliary motion
* Electron microscopy to examine ciliary ultrastructure
* Genetic testing for known mutations
* Clinical history and symptoms are also important.

## 4. Is there a cure for PCD?

Answer:  
Currently, there is no cure for PCD. Treatment focuses on managing symptoms, preventing infections, and maintaining lung function.

## 5. How is PCD treated?

Answer:  
Treatment includes:

* Airway clearance techniques (chest physiotherapy, breathing exercises)
* Antibiotics to treat lung and sinus infections
* Nasal saline washes and anti-inflammatory nasal sprays
* Monitoring lung health with sputum cultures, imaging, and pulmonary function tests
* In severe cases, oxygen therapy or lung transplantation may be considered.

## 6. Can children with PCD live normal lives?

Answer:  
With proper care and management, many children with PCD can lead active lives. Early diagnosis and multidisciplinary care improve outcomes. Support from specialists including pulmonologists, ENT doctors, and physiotherapists is important.

## 7. Is PCD inherited? Should family members be tested?

Answer:  
Yes, PCD is inherited in an autosomal recessive pattern. Genetic counseling and testing may be recommended for family members if a diagnosis is confirmed.

## 8. What should I do if my child with PCD gets worse?

Answer:  
Contact your healthcare provider if your child has increased coughing, fever, difficulty breathing, or other signs of infection. Prompt treatment can prevent complications.

## 9. Are there support resources for families affected by PCD?

Answer:  
Yes. Organizations like the PCD Foundation and national lung health charities provide education, support groups, and resources for patients and families

**DOCTOR PATIENT CONVERSATION**

Doctor: Hello! I’d like to talk with you about your symptoms and the possibility of Primary Ciliary Dyskinesia, or PCD. Have you noticed chronic cough, frequent sinus infections, or ear problems?

Patient: Yes, I’ve had a persistent wet cough since I was a child, and I often get sinus infections. My ears also seem to get infected a lot.

Doctor: Those are common symptoms of PCD, which is a rare inherited disorder where tiny hair-like structures called cilia don’t work properly. Normally, cilia help clear mucus and bacteria from your lungs and sinuses, but when they don’t function well, infections can become frequent.

Patient: So, this is something I was born with?

Doctor: Yes, PCD is genetic and usually starts early in life. Some people also have a condition called situs inversus, where their internal organs are mirrored, but not everyone has this.

Patient: How do you confirm if I have PCD?

Doctor: We use several tests. One is measuring nasal nitric oxide, which is typically low in PCD. We also look at the movement and structure of your cilia under a microscope, and genetic testing can identify mutations linked to PCD.

Patient: Is there a cure?

Doctor: There’s no cure yet, but we focus on managing symptoms and preventing lung damage. This includes airway clearance techniques, treating infections promptly with antibiotics, and regular monitoring of your lung health.

Patient: Can I still live a normal life?

Doctor: Many people with PCD lead active lives, especially with early diagnosis and proper care. We’ll work with a multidisciplinary team including ENT specialists, pulmonologists, and physiotherapists to support you.

Patient: What should I watch out for?

Doctor: Be alert for worsening cough, fever, or difficulty breathing, which may signal infections needing treatment. Also, regular follow-up is important to monitor lung function.

Patient: Is this something my family should be concerned about?

Doctor: Since PCD is inherited, family members might consider genetic counseling and testing, especially if they have symptoms.

Patient: Thank you, doctor. It helps to understand what’s going on and what to expect.

Doctor: You’re welcome. We’ll provide you with resources and support to manage your condition effectively. Please feel free to ask any questions as we go along.

REFERENCES

[Primary Ciliary Dyskinesia: Causes, Symptoms & Prognosis](https://my.clevelandclinic.org/health/diseases/22585-primary-ciliary-dyskinesia#overview)

<https://www.ncbi.nlm.nih.gov/books/NBK1122/#pcd.Differential_Diagnosis_of_Primary_Ci>

<https://emedicine.medscape.com/article/1002319-workup#c7>

**TRACHEOSTOMY**

**DEFINITION AND DESCRIPTION**

A tracheostomy (tray-key-OS-tuh-me) is a hole that surgeons make through the front of the neck and into the windpipe, also known as the trachea. Surgeons place a tracheostomy tube into the hole to keep it open for breathing. The term for the surgical procedure to create this opening is tracheotomy.

A tracheostomy allows air to pass into the windpipe to help with breathing. Tracheotomy is done when the usual way of breathing is blocked or reduced. A tracheostomy is often needed when health problems require long-term use of a machine called a ventilator to help with breathing. A tracheostomy also may be needed when surgery would require breathing to be rerouted for a short time because of swelling or airway blockage in the neck or face. Rarely, an emergency tracheotomy is done when the airway is suddenly blocked, such as after a major injury to the face or neck.

When a tracheostomy is no longer needed, it's allowed to heal shut, or a surgeon can close it. For some people, tracheostomies stay in place for the rest of their lives.

**Why it's done**

A tracheostomy may be needed when:

* Medical conditions make the use of a breathing machine, also known as a ventilator, necessary for an extended period, usually more than one or two weeks.
* Medical conditions, such as vocal cord paralysis, throat cancer or mouth cancer, block or narrow the airway.
* Paralysis, conditions that affect the brain and nerves, or other conditions make it hard to cough up mucus from your throat and make direct suctioning of the windpipe, also known as your trachea, necessary to clear your airway.
* Major head or neck surgery is planned. A tracheostomy helps with breathing during recovery.
* Severe injury to the head or neck blocks the usual way of breathing.
* Other emergency situations occur that block your ability to breathe and emergency personnel can't put a breathing tube through your mouth and into your windpipe.

### **Emergency care**

Most tracheotomies are done in a hospital setting. But in an emergency, emergency personnel may need to create a hole in a person's throat when outside of a hospital, such as at an accident scene.

Emergency tracheotomies are hard to do and have a greater risk of complications than those that are planned. A related and somewhat less risky — and more straightforward — procedure used in emergency care is a cricothyrotomy (kry-koe-thie-ROT-uh-me). This procedure creates a hole slightly higher up in the neck right below the voice box, also known as the larynx. The hole is placed right below the Adam's apple, which usually looks like a bump on the throat and is made up of thyroid cartilage that covers the front of the voice box.

Once a person transfers to a hospital and is stable, a tracheotomy replaces a cricothyrotomy if that person needs help breathing long term.

**Risks**

Tracheostomies are generally safe, but they have risks. Some complications are more likely during or shortly after surgery. The risk of complications is greater when a tracheotomy is done as an emergency procedure.

Complications that can occur right away include:

* Bleeding.
* Damage to the windpipe, thyroid gland or nerves in the neck.
* Movement of the tracheostomy tube or placement of the tube that isn't correct.
* The trapping of air in tissue under the skin of the neck. This is known as a subcutaneous emphysema. This issue can cause breathing problems and damage to the windpipe or the food pipe, also known as the esophagus.
* Buildup of air between the chest wall and lungs that causes pain, breathing problems or lung collapse. This is known as pneumothorax.
* A collection of blood, also known as a hematoma, that may form in the neck and squeeze the windpipe, causing breathing problems.

Long-term complications are more likely the longer a tracheostomy is in place. These problems include:

* Blockage of the tracheostomy tube.
* Movement of the tracheostomy tube from the windpipe.
* Damage, scarring or narrowing of the windpipe.
* Development of an unusual passage between the windpipe and the esophagus. This makes it more likely that fluids or food could enter the lungs.
* Development of a passage between the windpipe and the large artery that supplies blood to the right arm and right side of the head and neck. This can result in life-threatening bleeding.
* Infection around the tracheostomy or infection in the windpipe and bronchial tubes or lungs. An infection in the windpipe and bronchial tubes is known as tracheobronchitis. An infection in the lungs is known as pneumonia.

If you still need a tracheostomy after you've left the hospital, you'll likely need to keep regularly scheduled appointments to watch for possible complications. You'll also likely get instructions about when you should call your healthcare professional about problems, such as:

* Bleeding at the tracheostomy site or from the windpipe.
* Having a hard time breathing through the tube.
* Pain or a change in comfort level.
* A change in skin color or swelling around the tracheostomy.
* A change in the position of the tracheostomy tube.

**How you prepare**

How you prepare for a tracheostomy depends on the type of procedure you'll have. If you'll be having general anesthesia, your healthcare professional may ask that you not eat or drink for several hours before your procedure. You also may be asked to stop taking certain medicines.

### **Plan for your hospital stay**

After the tracheostomy procedure, you'll likely stay in the hospital for several days as your body heals. If your tracheostomy is a planned procedure, you can prepare for your hospital stay by bringing:

* Comfortable clothing, such as pajamas, a robe and slippers.
* Personal care items, such as your toothbrush and shaving supplies.
* Entertainment to help you pass the time, such as books, magazines or games.
* A communication method, such as a pencil and a pad of paper, a smartphone, or a computer, as you won't be able to talk at first. Your healthcare team may give you a writing whiteboard with a marker after your tracheostomy to help you communicate during the early part of your recovery.

**What you can expect**

### **During the procedure**

A tracheotomy is most commonly done in an operating room with general anesthetic, which is medicine that puts you to sleep. Occasionally, the procedure needs to be done when you're awake or lightly sedated rather than fully asleep. To do this, a surgeon uses a local anesthetic to numb the neck and throat to complete the procedure comfortably. Once the tracheostomy is completed and a tube is in place, you can then be safely put under general anesthesia to complete other parts of surgery if needed.

The type of procedure you have depends on why you need a tracheostomy and whether the procedure was planned. There are basically two options:

* **Surgical tracheotomy.** A surgeon can do this procedure in an operating room or in a hospital room. The surgeon usually makes a horizontal cut through the skin at the lower part of the front of your neck. The surgeon pulls back the surrounding muscles, and a small portion of the thyroid gland is cut. This exposes the windpipe, also known as the trachea. At a specific spot on your windpipe near the base of your neck, the surgeon creates a tracheostomy hole.
* **Minimally invasive tracheotomy.** Also called a percutaneous tracheotomy, a surgeon usually does this procedure in a hospital room. The surgeon makes a small cut near the base of the front of the neck. A special lens is fed through the mouth so that the surgeon can view the inside of the throat. Using this view of the throat, the surgeon guides a needle into the windpipe to create the tracheostomy hole. Then the surgeon expands the hole to the right size for the tube.

For both procedures, the surgeon inserts a tracheostomy tube into the hole. A neck strap attached to the faceplate of the tube keeps it from slipping out of the hole. Temporary sutures also can secure the faceplate to your neck.

### **After the procedure**

You'll likely spend several days in the hospital as your body heals. During that time, you learn the skills you need to maintain and cope with your tracheostomy, including how to:

* **Care for your tracheostomy tube.** A nurse teaches you how to clean and change your tracheostomy tube to help prevent infection and lower the risk of complications. You continue to do this as long as you have a tracheostomy.
* **Speak.** Generally, a tracheostomy prevents speaking because air goes out the tracheostomy rather than up through your voice box. But there are devices and techniques to redirect airflow enough to allow you to speak. Depending on the type of tube, width of your windpipe and condition of your voice box, you may be able to speak with the tube in place. If needed, a speech therapist or a nurse trained in tracheostomy care can help you learn to use your voice again.
* **Eat.** While you're healing, swallowing will likely be hard. You'll get nutrients through an IV, a feeding tube that passes through your mouth or nose, or a tube inserted directly into your stomach. When you're ready to eat again, you may need to work with a speech therapist. The speech therapist can help you regain the muscle strength and coordination you need to swallow.
* **Cope with dry air.** The air you breathe will be much drier because it no longer passes through your moist nose, mouth and throat before reaching your lungs. This can cause irritation, coughing and too much mucus coming out of the tracheostomy. Putting small amounts of saline directly into the tracheostomy tube, as directed, may help loosen mucus. Or a saline nebulizer treatment may help. A device called a heat and moisture exchanger captures moisture from the air you exhale and humidifies the air you inhale. A humidifier or vaporizer adds moisture to the air in a room.
* **Manage other effects.** Your healthcare team shows you ways to care for other common effects of a tracheostomy. For example, you may learn to use a suction machine to help you clear mucus from your throat or airway.

**Results**

In most cases, a tracheostomy is needed for a short time as a breathing route until other medical issues resolve. If you don't know how long you may need to be connected to a ventilator, the tracheostomy is often the best permanent solution.

Your healthcare team talks with you to help decide when it's the right time to take out the tracheostomy tube. The hole may close and heal on its own, or a surgeon can close it.

[Tracheostomy - Mayo Clinic](https://www.mayoclinic.org/tests-procedures/tracheostomy/about/pac-20384673)

### **Subglottic stenosis**

Alternative names and related terms for Subglottic Stenosis include:

* Subglottic narrowing
* Subglottic airway stenosis
* Subglottic obstruction
* Subglottic stricture
* Laryngotracheal stenosis (when involving both the larynx and subglottic area)
* Subglottic laryngeal stenosis
* Subglottic constriction
* Congenital subglottic stenosis (if present from birth)
* Acquired subglottic stenosis (due to trauma, intubation, infection, etc.)

**DEFINITION AND DESCRIPTION**

Subglottic stenosis is when the upper section of your trachea (windpipe) is narrower than usual so that there’s a whistling noise when you breathe, or you feel short of breath. “Subglottic” means the part of your trachea just below your vocal cords and “stenosis” means there’s a narrowing.

Anyone can have subglottic stenosis. Your newborn baby may be born with an unusually narrow airway (congenital subglottic stenosis). Or you could develop it after you’re born (acquired subglottic stenosis). Several things may cause subglottic stenosis in adults, but it can happen for no known reason (idiopathic subglottic stenosis).

Without treatment, severe subglottic stenosis can be life-threatening. But treatment to widen the narrow area in your windpipe can effectively treat the condition.

Subglottic stenosis is a rare condition, affecting 1 in every 400,000 people.

**Symptoms**

Subglottic stenosis symptoms can be mild, moderate or severe, depending on the narrowness of your airway. Think of sipping water through a flexible straw. You only get a sip of water when the straw is wide open. Squeeze it a little, and there’s less to drink. The more you squeeze, the less water flows through the straw.

In mild or moderate subglottic stenosis, part of your airway is narrower than usual. In severe subglottic stenosis, that part of your airway is nearly closed shut so that very little air that comes in through your nose makes it through your windpipe and into your lungs.

If your baby has this condition, they may have:

* Stridor, which is a high-pitched whistling noise when your baby breathes in or out.
* Dyspnea (difficulty breathing).
* Recurring croup.

If you or your older child has subglottic stenosis, symptoms may include:

* A cough that doesn’t go away or you bring up more mucus than usual when you cough.
* Shortness of breath (dyspnea) that doesn’t improve with inhalers or other treatments
* Stridor.

### **What causes subglottic stenosis?**

The condition can happen if your baby’s airway is narrower than usual (congenital subglottic stenosis). A child or an adult may develop it if they:

* Need intubation on a ventilator. People who need intubation for more than two weeks can sometimes develop subglottic stenosis.
* Have an injury that damages their airway.
* Have a rare autoimmune disorder or vasculitis that causes scar tissue to build up in their airway.

Some of the time, however, there’s no known reason why people develop subglottic stenosis. These cases tend to affect females between the ages of 30 and 60.

## **Diagnosis and Tests**

Your healthcare provider will perform a physical examination. They’ll ask about your medical history and your symptoms. If they suspect subglottic stenosis, they may refer you to an ear, nose and throat specialist (ENT or otolaryngologist). The specialist may perform tests, including:

* **Flexible laryngoscopy:** Your provider inserts a flexible tube down your throat. There’s a tiny camera on the tube that allows your provider to look at your larynx (voice box) and windpipe**.**
* **Pulmonary function test:** This test measures the amount of air that you can breathe in or out. Reduced airflow is a sign of subglottic stenosis.
* **Computed tomography (CT) scan:** Your provider may order a CT scan so they can look at the structure of your airway.

## **Management and Treatment**

Subglottic stenosis is rare, so there’s no standard or common treatment for it. Treatments may include checking on your condition, steroid injections and surgery.

#### **Monitoring**

If tests show subglottic stenosis is causing mild narrowing of your airway, your provider may schedule regular appointments so they can check your airway. They may do more tests to see if your airway is narrowing.

#### **Steroid injections**

Your provider may do steroid injections as initial or first-line treatment. The treatment involves injecting a steroid through your neck into your airway. (Your provider will use local anesthesia to numb your neck, airway and nose so you won’t have pain during treatment.) Typically, people who have this treatment have a series of injections every four to six weeks.

#### **Surgery for subglottic stenosis**

There are a few different surgical procedures to treat subglottic stenosis:

* **Endoscopic dilation:** In this treatment, your provider uses an endoscope to place a tiny balloon in the narrow area of your windpipe. The balloon dilates (stretches) the section. An endoscopic dilation isn’t a permanent solution: at some point, you’ll need to have the procedure again to keep your airway open.
* **Cricotracheal resection:** This surgery involves removing the narrowed part of your airway just below your voice box and then reconnecting your airway and voice box.
* **Endoscopic laryngotracheoplasty**: In this surgery, providers widen your airway by placing pieces of cartilage in your airway. Surgeons may call this surgery laryngotracheal reconstruction.

## **Outlook / Prognosis**

Often, prompt treatment can effectively treat the condition. But subglottic stenosis can come back, which means you or your child will need more treatment.

### **When to see a doctor**

If you or your child has treatment for subglottic stenosis, contact your provider as soon as you notice changes or symptoms, like stridor or breathing issues, that may mean the condition is coming back. You or your child may need more treatment to prevent symptoms from getting worse.

## **Epidemiology**

### Frequency

The frequency of congenital subglottic stenosis (SGS) is unknown.

The incidence of acquired subglottic stenosis (SGS) has greatly decreased since the late 20th century. In the late 1960s, when endotracheal intubation and long-term ventilation for premature infants began, the incidence of acquired subglottic stenosis (SGS) was as high as 24% in patients requiring such care. In the 1970s and 1980s, estimates of the incidence of subglottic stenosis (SGS) were 1-8%.

In 1998, Choi and Zalzal reported that the incidence of subglottic stenosis (SGS) had remained constant at the Children's National Medical Center in Washington, DC; it was approximately 1-2% in children who had been treated in the neonatal intensive care unit (ICU).Walner reported that, among 504 neonates who were admitted to the level III ICU at the University of Chicago in 1997, 281 were intubated for an average of 11 days, with no patients developing subglottic stenosis (SGS) over a 3-year period. Moreover, in a systematic review published in 2001, Walner et al reported a decreasing incidence of neonatal subglottic stenosis in the literature, with studies published after 1983 finding an incidence of less than 4% and studies after 1990 having an incidence of under 0.63%. In 1996, a report from France described no incidence of subglottic stenosis (SGS) in the neonatal population who underwent intubation with very small endotracheal tubes (ie, 2.5-mm internal diameter) in attempts to prevent trauma to the airway.

### Morbidity

Using the Kids’ Inpatient Database (KID), Arianpour et al found that in addition to gastroesophageal reflux (GER), comorbidities more likely to be diagnosed in inpatients aged 20 years or younger with acquired subglottic stenosis (SGS) include trisomy 21, asthma, and additional upper airway anomalies. However, the chance of prematurity and dehydration was indicated to be lower in pediatric acquired SGS.

## **Diagnostic Procedures**

In a child with mild or moderate airway obstruction, perform flexible fiberoptic nasopharyngoscopy and laryngoscopy in the clinic or the emergency department (ED). If extreme airway obstruction exists or if an active supraglottic infectious process is suspected in a young child, flexible endoscopy may be deferred in favor of formal rigid bronchoscopy in the operating room (OR). However, flexible fiberoptic nasopharyngoscopy may be performed in a controlled setting in the OR, because determination of the nature of the supraglottis and glottis in awake, unsedated patients is crucial. The procedures are described as follows.

### Flexible fiberoptic nasopharyngoscopy and laryngoscopy

During flexible fiberoptic nasopharyngoscopy and laryngoscopy, topical anesthesia and decongestion can be accomplished in older infants and children with topical Afrin and lidocaine. A 3-mm endoscope can be used, even in an infant. Pass the endoscope into both nasal cavities to access pyriform aperture stenosis, midnasal stenosis, choanal atresia or stenosis, lesions of the nose and nasopharynx, and the adenoid pad.

Pass the endoscope into the superior oropharynx and hypopharynx. The hypopharynx and larynx can be assessed. Identify the structure and position of the supraglottis. Evaluate the epiglottis and arytenoids for malacia or stenosis. Evaluate the position and movement of the true vocal cords. Evaluate edema or erythema of the true vocal cords, epiglottis, and arytenoids.

### Flexible endoscopy

This can be performed with the patient in the supine or sitting position. The supine position often results in the obstruction of certain supraglottic processes. If the goal is to obtain the best visualization of the true vocal cords and supraglottis, place a child (even an infant) in the sitting position with his or her neck extended.

If the child is older, the voice can be evaluated, and videostroboscopy can be performed to assess the vocal cord waveform and vocal cord mobility.

Occasionally, the subglottis can be visualized with flexible endoscopy; however, rigid laryngoscopy and bronchoscopy are the safest procedures and offer the best visualization for the subglottis and tracheobronchial tree.

### Rigid laryngoscopy and bronchoscopy

Rigid laryngoscopy and bronchoscopy is the best single test for evaluating airway obstruction in children. The otolaryngologist must have knowledge of the pediatric airway, and the OR must have adequate bronchoscopes and telescopes of various sizes. Prepare all equipment for bronchoscopy, including laryngoscopes, light sources, video documentation equipment, telescopes, and bronchoscopes prior to the child's arrival in the OR. Throughout the procedure, maintain good communication between anesthesiologists, surgical nursing staff, and physicians, so that any potential airway obstruction can be quickly assessed and addressed.

Do not further injure the pediatric airway—this point is of paramount importance. Use the smallest bronchoscope or telescope alone for evaluation of the subglottis in a child who does not require ventilation throughout the procedure. This practice allows good visualization without iatrogenic injury to the area. If ventilation is required throughout the evaluation, use a bronchoscope-telescope combination.

If a child has a tracheotomy or is not in extreme distress, the child can breathe spontaneously and inhale oxygen and anesthetics through an endotracheal tube in the pharynx while the airways are visualized with a laryngoscope and large telescope. Frequently, the true vocal cords are anesthetized with lidocaine prior to evaluation to help prevent laryngospasm.

Determine the size of the child's airway by using endotracheal tubes. As previously mentioned, Myer and Cotton established a scale for subglottic stenosis (SGS) severity that is based on the child's age and the size of the endotracheal tube that can be placed in the airway with an air leak pressure of less than 20 cm of water.

Evaluate the subglottis and glottis for fixation, scarring, granulation, edema, paralysis or paresis, and other abnormalities. Evaluate the distance and caliber of the stenosis. Apply the Myer and Cotton staging system only to circumferential subglottic stenosis (SGS). [[27](javascript:void(0);)] Glottic stenosis and SGS often coexist and must be considered when reconstruction is planned.

Evaluate the maturity of the stenosis. If a firm white scar is present, the stenosis is mature. If the stenosis has a granular or erythematous appearance, GERD, viral infection, allergic esophagitis, or another inflammatory process may be present.

Examine the area below the subglottis into the trachea and bronchi for secondary lesions. The suprastomal area is important because pathologic stenosis or malacia can influence the choice of surgical procedure. In severe subglottic stenosis (SGS), viewing the suprastomal area requires the passage of a tiny telescope through a narrow subglottis or a telescope or bronchoscope through a tracheotomy site, if available.

**pediatric differential diagnosis (DDx) of subglottic stenosis**

* Congenital subglottic stenosis: Narrowing present at birth due to thickened soft tissue or cartilage in the subglottic airway.
* Acquired subglottic stenosis: Often due to prolonged or repeated endotracheal intubation causing scarring and fibrosis.
* Laryngomalacia: Most common cause of stridor in infants, caused by floppy supraglottic structures collapsing during inspiration.
* Subglottic hemangioma: Vascular tumor in the subglottic region causing airway obstruction and biphasic stridor.
* Vocal cord paralysis: Unilateral or bilateral immobility leading to airway compromise and voice changes.
* Laryngotracheobronchitis (Croup): Viral infection causing inflammation and narrowing of the subglottic airway.
* Epiglottitis: Acute bacterial infection causing supraglottic swelling and airway obstruction.
* Bacterial tracheitis: Severe bacterial infection causing airway inflammation and narrowing.
* Foreign body aspiration: Sudden onset airway obstruction with history of choking.
* Laryngeal webs or cysts: Congenital or acquired membranes causing partial airway obstruction.
* Granulomatosis with polyangiitis: Autoimmune vasculitis causing subglottic inflammation and stenosis.
* Extrinsic airway compression: From vascular rings or masses compressing the airway externally.
* Gastroesophageal reflux disease (GERD): Can exacerbate airway inflammation and contribute to stenosis or edema.

**PREDEFINED Q AND A**

## 1. How severe is my child’s condition?

The severity of subglottic stenosis (SGS) is usually graded based on how much the airway is narrowed:

* Mild (Grade 1-2): Less than 50% narrowing; many children may have few or no symptoms and sometimes improve as they grow.
* Moderate (Grade 3): 50–70% narrowing; symptoms like stridor and breathing difficulty may be more noticeable.
* Severe (Grade 4): More than 70% narrowing; significant breathing problems often require urgent intervention.

Your doctor will determine severity through endoscopic airway evaluation and imaging.

## 2. Will my child need treatment right away?

* Mild cases: Often monitored closely without immediate intervention. Many improve with growth and airway maturation.
* Moderate to severe cases: Usually require treatment to prevent breathing difficulties. Timing depends on symptoms and overall health.
* If your child has significant breathing problems or recurrent infections, more urgent treatment may be needed.

## 3. What treatment do you recommend for my child?

Treatment depends on severity and symptoms:

* Observation: For mild stenosis without symptoms, regular monitoring is key.
* Endoscopic surgery: Minimally invasive procedures like balloon dilation or laser removal of scar tissue are often used for mild to moderate stenosis.
* Open surgery: For moderate to severe cases, procedures like laryngotracheal reconstruction (LTR) or partial cricotracheal resection (PCTR) are performed to widen the airway.
* Tracheostomy: In some cases, a temporary tracheostomy may be needed to secure the airway before or during treatment.

Your child’s care team will tailor treatment based on the grade of stenosis, airway anatomy, and your child’s overall condition.

**GENOMIC DATA**

* Congenital SGS and Genetic Syndromes:  
  Congenital subglottic stenosis is rare and can be associated with genetic syndromes such as Trisomy 21 (Down syndrome) and CHARGE syndrome, indicating a developmental genetic component affecting airway cartilage formation.
* Idiopathic Subglottic Stenosis (iSGS) and Genetics:  
  Idiopathic SGS primarily affects middle-aged Caucasian women, suggesting a possible hormonal influence rather than a classic inherited genetic disorder. Studies investigating HLA genotyping found no significant genetic association within the major histocompatibility complex, indicating that if genetic susceptibility exists, it likely lies outside the HLA locus. Familial cases are rare but reported, supporting a possible genetic or familial predisposition in some patients.
* Molecular Mechanisms:  
  Emerging research points to a complex interplay of genetic susceptibility, growth factors, cytokines, fibroblast activity, hypoxia, and biomechanical stress contributing to the fibrotic scarring and airway narrowing seen in SGS

**DOCTOR PATIENT CONVERSATION**

Doctor: Hello! I want to discuss the results of your child’s airway evaluation. The findings show that your child has subglottic stenosis, which means there is narrowing just below the vocal cords.

Parent/Patient: What does that mean exactly? How serious is it?

Doctor: The subglottic area is the narrowest part of the airway in children. When it’s narrowed, it can make breathing more difficult and cause symptoms like noisy breathing or stridor. The severity can vary—from mild narrowing that may improve over time, to more significant narrowing that needs treatment.

Parent/Patient: What caused this? Was it something we did?

Doctor: Subglottic stenosis can be congenital, meaning present from birth, or acquired, often from prolonged intubation or injury to the airway. Sometimes, we don’t find a clear cause. It’s not your fault, and we have effective treatments available.

Parent/Patient: What kind of treatments are there? Will my child need surgery?

Doctor: Treatment depends on how severe the narrowing is and how much it affects breathing. Mild cases might just need monitoring. Moderate cases can sometimes be treated with minimally invasive procedures like balloon dilation. More severe cases may require surgery to widen the airway. We’ll tailor the plan to your child’s needs.

Parent/Patient: How soon do we need to start treatment?

Doctor: If your child is having significant breathing difficulties, we’ll want to start treatment soon to prevent complications. If symptoms are mild, we can monitor closely and plan treatment accordingly.

Parent/Patient: Are there risks with the treatments?

Doctor: Like any procedure, there are risks such as bleeding, infection, or need for repeat treatments. But these are relatively uncommon, and we take every precaution to keep your child safe.

Parent/Patient: What should I watch for at home?

Doctor: Watch for worsening noisy breathing, difficulty feeding, fatigue, or bluish color around the lips. If you notice these, seek medical attention promptly.

Parent/Patient: Will this affect my child long-term?

Doctor: Many children do very well after treatment and can breathe normally. Some may need ongoing follow-up, but with proper care, the outlook is good.

Parent/Patient: Thank you for explaining everything. What’s the next step?

Doctor: We’ll schedule the appropriate treatment or monitoring and provide you with information and support. Please feel free to call anytime with questions or concerns.

REFERENCES

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**LARYNGEAL WEB**

**DEFINITION AND DESCRIPTION**

Laryngeal web is a rare condition where the windpipe is partially constricted, or narrowed, making it difficult to breathe normally. The larynx (voice box) contains web-like tissue that reduces the amount of air flowing in and out of the windpipe. The web may be very thin or thick, which will determine how much the breathing is affected and may also impact the treatment method.

### **Causes**

Laryngeal web is often a congenital defect, which means it is present from birth. In some cases, laryngeal webs can form later, often after long-term intubation.

### **Signs and Symptoms**

The most common symptom is frequent shortness of breath and stridor, which includes a vibrating sound as if something is partially blocking the windpipe. Poor feeding and weak cry have been associated with these webs.

Other symptoms that may occur include:

* Wheezing
* Coughing
* Frequent chest infections

Some children also tend to raise their head or stretch their neck to open the airway as much as possible.

### **Diagnosis**

Laryngeal web often has symptoms that can look like asthma, and it can be initially misdiagnosed.

When asthma medications or other treatments do not improve your child’s condition (often called refractory asthma), it is important to inform your doctor.

To properly diagnose tracheal web, the following diagnostic procedures may be used:

* Micro laryngoscopy: A micro laryngoscopy, or microscopic laryngoscopy, uses a small, flexible telescope to look in the airway for signs of a tracheal web or partial blockage.
* CT scan: In some cases, a picture of the chest may be needed to look for airway abnormalities.

### **Treatment**

The goal of treatment is to remove the blockage by breaking the web, allowing the airway to open up to its full size. Depending on the severity of the web, the surgeon may do this by dilating (widening) the trachea and using a laser or cutting instrument to break the web. In some cases of a thicker web, the surgeon may recommend an open surgical approach to expand the airway.

#### **Dilation**

A small balloon is placed in the airway to widen or break the web. This may need to be repeated to prevent the web from coming back because of scar tissue.

#### **Surgery**

For some webs, the correct treatment is an open surgical procedure called a larynogofissure, opening into the larynx, generally through a midline cut through the neck.

#### **Recovery**

If dilation is needed, your child will spend one to two nights in the hospital until their breathing is safe. If an open neck procedure is needed, then one to two weeks is usually needed in the hospital following the operation.

### **Long-Term Outlook**

Prognosis is good with a normal life span. The only noticeable long-term symptoms may be a hoarse or rough voice.

**Differential diagnoses (DDx) for laryngeal web**

* Congenital laryngeal complex stenosis
* Laryngeal papillomatosis
* Subglottic cysts
* Laryngeal cleft
* Congenital subglottic stenosis
* Post-intubation laryngeal injury (acquired webs or stenosis)
* Tracheal webs
* Tracheomalacia
* Tracheal stenosis
* Laryngeal tumors (benign or malignant)
* Functional laryngeal disorders (e.g., vocal cord dysfunction)
* 22q11.2 deletion syndrome-associated anomalies
* Gastroesophageal reflux-related laryngeal inflammation

## **Epidemiology**

### Frequency

Approximately 15% of patients who are intubated for more than 10 days develop some degree of glottic stenosis. Ninety percent of acquired subglottic stenoses in infants and children are due to endotracheal intubation. The incidence of subglottic stenosis after intubation is reported to be 1-10%.

Congenital glottic stenosis is a rare disorder and may exist as a thin membranous stenosis, as a thick anterior or posterior web, or as a complete fusion of the vocal cords. Congenital laryngeal webs are rare; one report identified 51 children with webs during a 32-year period.

### Sex and Race

In a retrospective, multi-institutional study, Gelbard et al reported an interesting degree of homogeneity between patients with idiopathic subglottic stenosis, finding that the vast majority of the 479 patients, from 10 participating centers, were female (98%) and Caucasian (95%)

## **Diagnostic Procedures**

See the list below:

* Flexible fiberoptic endoscopy in the awake patient is performed to assess for supraglottic and glottic pathology such as vocal cord paralysis and laryngomalacia.
* Direct laryngoscopy and bronchoscopy with the patient under general anesthesia remain the criterion standard of diagnosis by allowing careful evaluation of each segment of the airway: supraglottis, glottis, subglottis, and trachea. The outer diameter of the largest bronchoscope that can pass through the stenosis should be noted as well as the length of the stenotic segment, its location, thickness, and composition. Use of the telescope without the bronchoscope allows dynamic assessment. Rule out secondary sites of stenosis and palpate arytenoids for cricoarytenoid joint fixation. Laryngeal electromyography may be useful.
* Determine the size of the airway visually and objectively by using endotracheal tubes. An individual’s endotracheal tube size is the largest tube that permits an air leak at less than 30 cm of water pressure.
* Evaluate for gastroesophageal reflux (pH probe) in all surgical candidates.
* Assess vocal status (with the assistance of speech pathologists) preoperatively in older children and adults.

## **Staging**

The classification of glottic webs as described by Cohen is as follows:

* Type I is an anterior web involving 35% or less of the glottis with visible true cords and no subglottic extension. Airway and voice symptoms are mild.
* Type II is an anterior web involving 35-50% of the glottis. Minimal involvement of subglottis. Mild airway and voice symptoms.
* Type III is an anterior web involving 50-75% of the glottis with associated cricoid abnormalities. True vocal cords may not be visualized. Severe airways symptoms with marked vocal dysfunction. Airway intervention may be necessary.
* Type IV is a web occluding 75-90% of the glottis. True vocal cords are not identifiable. Subglottis is narrowed. The patient is aphonic. Immediate airway management is required.
* Posterior glottic stenosis may be defined as one of the following 4 types: (Bogdasarian and Olson).
  + Type I is vocal process adhesion from an interarytenoid scar with a mucosally lined posterior sinus tract.
  + Type II is a posterior commissure stenosis involving the submucosa of the posterior commissure, interarytenoid region, and internal surface of the posterior cricoid lamina.
  + Type II and IV stenosis are posterior commissure scars involving the cricoarytenoid joint unilaterally or bilaterally, respectively.
* The Myer-Cotton Grading system for subglottic stenosis is the most frequently used and is based on the leak pressure, tube size, and age of the patient, as follows:
  + Grade I - Less than 50% laryngeal lumen obstruction
  + Grade II - 50-70% obstruction
  + Grade III - 71-99% obstruction with an identifiable lumen present
  + Grade IV - Complete obstruction; no lumen present

**DOCTOR PATIENT CONVERSATION**

Doctor: Hello! I want to talk with you about the diagnosis we found during your child’s airway examination. Your child has a condition called a laryngeal web, which is a thin membrane of tissue partially blocking the voice box.

Parent/Patient: What does that mean? How serious is it?

Doctor: The laryngeal web can vary in thickness and size. Sometimes it causes mild symptoms like a hoarse or weak voice, but if it’s thicker or covers more of the airway, it can cause breathing difficulties or noisy breathing called stridor. We’ll assess how much it’s affecting your child’s breathing and voice.

Parent/Patient: How did this happen? Is it something we caused?

Doctor: Laryngeal webs are usually congenital, meaning they develop before birth. Sometimes they can form after injury or prolonged intubation, but in most cases, it’s not anything caused by parents or caregivers.

Parent/Patient: What treatment does my child need? Will surgery be necessary?

Doctor: Treatment depends on the severity. For thin webs with mild symptoms, sometimes we just monitor closely. For thicker webs causing breathing or voice problems, surgery to remove or open the web is often recommended. We use minimally invasive techniques like laser surgery or endoscopic procedures, which are effective and safe.

Parent/Patient: What can we expect after treatment?

Doctor: Most children improve significantly after surgery, with better breathing and voice. We’ll follow your child closely after treatment to watch for any recurrence or complications.

Parent/Patient: Are there any risks with surgery?

Doctor: As with any procedure, there are some risks like bleeding, infection, or scarring, but these are uncommon. We take every precaution to minimize risks and ensure your child’s safety.

Parent/Patient: What should we watch for at home?

Doctor: Watch for worsening breathing difficulty, persistent hoarseness, or feeding problems. If you notice any of these, please contact us promptly.

Parent/Patient: Thank you for explaining everything clearly.

Doctor: You’re very welcome. We’re here to support you and your child every step of the way. Please feel free to ask any questions anytime.

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**ACUTE RESPIRATORY DISTRESS SYNDROME**

**DEFINITION AND DESCRIPTION**

Acute respiratory distress syndrome (ARDS) occurs when lung swelling causes fluid to build up in the tiny elastic air sacs in the lungs. These air sacs, called alveoli, have a protective membrane, but lung swelling damages that membrane. The fluid leaking into the air sacs keeps the lungs from filling with enough air. This means less oxygen reaches the bloodstream, so the body's organs don't get the oxygen they need to work properly.

ARDS usually occurs in people who are already critically ill or have major injuries. People usually are severely short of breath — the main symptom of ARDS — within a few hours to a few days after the injury or infection that caused ARDS.

Many people who get ARDS don't survive. The risk of death gets higher with age and how severe the illness is. Of the people who survive ARDS, some fully recover. But others have lasting lung damage.

**CAUSES**

Causes of ARDS include:

* **Sepsis.** The most common cause of ARDS is sepsis, a serious and widespread infection of the bloodstream.
* **Severe pneumonia.** Severe cases of pneumonia usually affect all five lobes of the lungs.
* **Coronavirus disease 2019 (COVID-19).** People who have severe COVID-19 may get ARDS. Because COVID-19 mainly affects the respiratory system, it can cause lung injury and swelling that can lead to COVID-19-related ARDS.
* **Head, chest or other major injury.** Accidents, such as falls or car crashes, can damage the lungs or the portion of the brain that controls breathing.
* **Breathing in harmful substances.** Breathing in a lot of smoke or chemical fumes can lead to ARDS, as can breathing in vomit. Breathing in water in cases of near-drownings also can cause ARDS.
* **Other conditions and treatments.** Swelling of the pancreas (pancreatitis), massive blood transfusions and severe burns can lead to ARDS.

**Risk factors**

Most people who get ARDS already are in a hospital for another condition. Many are critically ill. People are especially at risk if they have an infection, such as sepsis or pneumonia. They're also at higher risk if they have COVID-19, especially if they also have metabolic syndrome.

People who have alcohol use disorder or who use recreational drugs or smoke ― lifestyle habits that can harm the lungs ― are at higher risk of getting ARDS. Having a history of alcohol, drug or tobacco use also raises the risk of ARDS.

**Symptoms**

The seriousness of ARDS symptoms can vary depending on what's causing them and whether there is underlying heart or lung disease. Symptoms include:

* Severe shortness of breath.
* Labored and rapid breathing that is not usual.
* Cough.
* Chest discomfort.
* Fast heart rate.
* Confusion and extreme tiredness.

### **When to see a doctor**

ARDS usually follows a major illness or injury, and most people who have ARDS are already in a hospital. But if you have symptoms of ARDS and are not in a medical facility, go to the nearest emergency department right away or call 911 or your local emergency number for help.

## **Diagnosis**

There's no specific test for ARDS. Healthcare professionals base the diagnosis on physical exams, chest X-rays and oxygen levels. It's also important to rule out other diseases and conditions, such as certain heart problems that can lead to similar symptoms.

### **Imaging**

A chest X-ray can show which parts of your lungs, and how much of the lungs, have fluid in them and whether your heart has gotten bigger. Another test called a CT scan combines X-ray images taken from many directions and creates cross-sectional views of internal organs. CT scans can give detailed information about the structures within the heart and lungs.

### **Lab tests**

A test using blood from an artery can measure your oxygen level. Other types of blood tests can check for symptoms of infection or other medical conditions. If your healthcare professional thinks that you have a lung infection, secretions from your airway may be tested to find the cause of the infection.

### **Heart tests**

Because the symptoms of ARDS are like those of certain heart problems, your healthcare professional may recommend heart tests such as:

* **Electrocardiogram.** This painless test, which also is known as an ECG, tracks the electrical activity in your heart. During the test, a healthcare professional attaches several wired sensors to your body.
* **Echocardiogram.** This test uses sound waves to create pictures of the heart. It shows how blood moves through the heart chambers and heart valves, and whether there are changes in the structures of your heart.

**Treatment**

The first goal in treating ARDS is to improve the levels of oxygen in your blood. Without oxygen, your organs can't work properly.

### **Oxygen**

To get more oxygen into your bloodstream, your healthcare professional likely will use:

* **Extra oxygen.** For milder symptoms or as a short-term treatment, oxygen may be delivered through a mask that fits tightly over your nose and mouth.
* **Mechanical ventilation.** Most people with ARDS need the help of a machine to breathe. A mechanical ventilator pushes air into your lungs and forces some of the fluid out of the air sacs.

### **Extracorporeal membrane oxygenation (ECMO)**

ECMO may be an option for severe ARDS when other treatment options, such as mechanical ventilation, don't work. ECMO takes over for the heart, lungs or both for a limited time while the lungs rest and heal. This treatment can help when the body can't provide the tissues with enough oxygen.

The ECMO machine is an artificial heart and lung, removing blood from the body through tubes and pumping the blood through the artificial lung. This process removes carbon dioxide and adds oxygen. Then the machine pumps the blood back into the body. Because of the risks involved, it's important to discuss the pros and cons of ECMO with your healthcare team.

### **Prone positioning**

For some people with ARDS, positioning on the stomach — what's known as a prone position — during mechanical ventilation may make more oxygen available to the lungs.

### **Fluids**

Carefully managing the amount of IV fluids given to people with ARDS is very important. Giving too much fluid can make more fluid build up in the lungs. Giving too little fluid can strain the heart and other organs, leading to shock.

### **Medication**

People with ARDS usually get medicine to:

* Prevent and treat infections.
* Ease pain and discomfort.
* Prevent blood clots in the legs and lungs.
* Reduce gastric acid reflux as much as possible.
* Help them feel calm or less anxious.

### **Lung transplant**

When other treatments don't help, lung transplant may be an option for some carefully chosen people who have ARDS. Usually, these are people who were healthy before they developed severe ARDS. Because lung transplant is such a hard process, it should be done at a center that has highly skilled, experienced surgeons and transplant teams.

**Lifestyle and home remedies**

If you're recovering from ARDS, these suggestions can help protect your lungs:

* **Quit smoking.** If you smoke, seek help to quit. Also, stay away from secondhand smoke whenever you can.
* **Get vaccinated.** Getting the flu, also called influenza, shot every year, as well as the pneumonia vaccine as often as recommended, can lower your risk of lung infections.
* **Attend pulmonary rehabilitation.** Many medical centers now offer pulmonary rehabilitation programs that include exercise training, education and counseling to help you learn how to get back to your usual activities and get to your ideal weight.

**Complications**

ARDS can cause other medical problems while in the hospital, including:

* **Blood clots.** Lying still in the hospital while you're on a ventilator can make it more likely that you'll get blood clots, particularly in the deep veins in your legs. If a clot forms in your leg, a portion of it can break off and travel to one or both of your lungs, where it can block blood flow. This is called a pulmonary embolism.
* **Collapsed lung, also called pneumothorax.** In most people with ARDS, a breathing machine called a ventilator brings more oxygen into the body and forces fluid out of the lungs. But the pressure and air volume of the ventilator can force gas to go through a small hole in the very outside of a lung and cause that lung to collapse.
* **Infections.** A ventilator attaches to a tube inserted in your windpipe. This makes it much easier for germs to infect and injure your lungs.
* **Scarred and damaged lungs, known as pulmonary fibrosis.** Scarring and thickening of the tissue between the air sacs in the lungs can occur within a few weeks of the start of ARDS. This makes your lungs stiffer, and it's even harder for oxygen to flow from the air sacs into your bloodstream.
* **Stress ulcers.** Extra acid that your stomach makes because of serious illness or injury can irritate the stomach lining and lead to ulcers.

Thanks to better treatments, more people are surviving ARDS. But many survivors end up with potentially serious and sometimes lasting effects:

* **Breathing problems.** After having ARDS, many people get most of their lung function back within several months to several years, but others may have breathing problems for the rest of their lives. Even people who do well usually have shortness of breath and fatigue and may need extra oxygen at home for a few months.
* **Depression.** Most ARDS survivors also report going through a period of depression, which can be treated.
* **Problems with memory and thinking clearly.** Sedatives and low levels of oxygen in the blood can lead to memory loss and learning problems after ARDS. In some people, the effects may get better over time. But in others, the damage may last for the rest of their lives.
* **Tiredness and muscle weakness.** Being in the hospital and on a ventilator can cause your muscles to weaken. You also may feel very tired after treatment.

## **Outlook / Prognosis**

ARDS can be life-threatening and scary. But improved care and ventilator treatments — including having people lay face down (prone) to improve oxygen flow — are helping more people survive and reduce ARDS complications. The outlook is typically better in people younger than 65 and when trauma or a blood transfusion causes ARDS.

Recovery from ARDS may take a long time. Most people who are taken off a ventilator can breathe freely. Some recover completely, but others may develop chronic lung problems that require care by lung specialists (pulmonologists).

### **Can your lungs recover from ARDS?**

Yes, your lungs can recover from ARDS. The exact amount of time varies depending on how much lung damage you have. Most people regain their lung function within two years, although several factors go into that estimate.

## **Prevention**

There’s no way to prevent acute respiratory distress syndrome (ARDS). But you may be able to prevent it from being severe by seeking immediate medical attention if you have symptoms of respiratory distress, or if you have a lung injury or other disease. You can lower your risk of getting severe ARDS by quitting smoking and avoiding alcohol.

## **Living With**

Life after ARDS can be challenging. Have patience with yourself as you navigate recovery and lean on friends and family members for support. You may need help performing everyday tasks while your lungs regain function. Talk to your healthcare professional

## **Common Questions**

### **Are ARDS and COVID-19 the same?**

No, they aren’t the same. People with COVID-19 can develop ARDS when the virus gets into their lungs.

#### **What is the survival rate for ARDS in people with COVID-19?**

Researchers are always learning more about the COVID-19 virus. Most studies conclude that the survival rate for a person with both COVID-19 and ARDS is about 50% to 60%. But many factors go determining if a person survives ARDS and COVID-19, including health history and other medical conditions.

## **Diagnostic Considerations**

Other conditions to be considered include the following:

* Oxygen toxicity
* Ventilator-induced lung injury
* Decreased capillary oncotic pressure
* Neurogenic pulmonary edema
* Postobstructive pulmonary edema (due to increased negative interstitial pressure)
* Fat embolism

## **Differential Diagnoses**

* Acute Poststreptococcal Glomerulonephritis
* Afebrile Pneumonia Syndrome
* Airway Foreign Body Imaging
* Aspiration Pneumonitis and Pneumonia
* Asthma Imaging and Diagnosis
* Bacterial Pneumonia
* Constrictive Pericarditis
* Drowning
* Empyema and Abscess Pneumonia
* Gastroesophageal Reflux Disease (GERD) Imaging
* Goodpasture Syndrome
* Heart Failure
* Hemosiderosis
* High Altitude Pulmonary Hypertension
* Hydrocarbon Inhalation Injury
* Hypersensitivity Pneumonitis
* Pulmonary Alveolar Proteinosis Imaging
* Pulmonary Interstitial Emphysema Imaging
* Lymphangitis
* Meningococcal Infections
* Myocardial Infarction in Childhood
* Nephrotic Syndrome
* Pediatric Malignant Pericardial Effusion
* Pediatric Nephritis
* Pediatric Nonviral Myocarditis
* Pediatric Pneumococcal Bacteremia
* Pediatric Pneumonia
* Pediatric Idiopathic Pulmonary Artery Hypertension
* Pediatric Respiratory Failure
* Pediatric Viral Myocarditis
* Pediatrics, Bronchiolitis
* Pneumococcal Infections (Streptococcus pneumoniae)
* Pneumomediastinum
* Pneumonia in Immunocompromised Patients
* Pneumonia, Mycoplasma
* Pneumothorax
* Pseudomonas Infection
* Pulmonary Atelectasis
* Pulmonary Embolism (PE)
* Pulmonary Infarction
* Viral Pneumonia

## **Epidemiology**

The incidence of ARDS is certainly lower in the pediatric population as compared to adults. The adult studies have reported a very wide range of incidence: from 17.9-86.2 per 100,000 person-years. For the population aged 15 years and older, age-adjusted incidence was 86.2 per 100,000 person-years, 38.5% hospital mortality; accounting for an estimated 190,600 cases of acute lung injury, 74,500 deaths, and 3.6 million hospital days each year in the United States.

The incidence in the pediatric population is reported between 2.2 and 12.8 per 100,000 person-years. From the critical care perspective, ALI/ARDS accounts for 2.2% to 2.6% of pediatric intensive care unit (PICU) admissions, 8.3% of those receiving mechanical ventilation for more than 24 hours, and PICU and hospital mortality ranging between 18% and 32.8%.

The Pediatric Respiratory Distress Incidence and Epidemiology (PARDIE) study, which involved 27 countries, found that pediatric ARDS occurs in about 3% of patients in PICUs and in about 6% of those who are receiving mechanical ventilation. In addition, among mechanically ventilated patients, the greatest number of new cases of pediatric ARDS occurred in North America, in high-income countries, and during non-summer months.

The age-related statistics of ARDS can be obtained by comparing the results of two different studies from King County, Washington, USA, that were conducted around the same time between 1999 and 2000.

|  |  |  |  |
| --- | --- | --- | --- |
| Study | Zimmerman JJ et al | Rubenfield GD et al | |
| Age in years | 0.5 to 15 | 15 through 19 | 75 through 84 |
| Incidence per 100,000 person-years | 12.8 | 16 | 306 |
| Mortality | 18% | 24% | 60% |

Incidence and severity of ARDS are somewhat similar at different geographical locations. The study from Australia and New Zealand reported an incidence of 2.95 per 100,000 person-years, 2.2% of PICU admissions, and 30% of PICU mortality. A Dutch study reported an incidence of 2.2 per 100,000 person-years and 20.4% mortality. Investigators in Spain found an incidence of 3.9 per 100,000 patients-years and a PICU mortality of 26%. A German study showed incidence of 3.2 per 100,000 person-years. The incidence in the US-based study was a little higher at 12.8 per 100,000 person-years; however, mortality was slightly lower at 18%. Search of the Chinese literature revealed 2.6% of PICU admissions were for ARDS, with a mortality of 32.8%.

Of note, the above reported epidemiologic data are from studies prior to the Berlin definition, a study that eliminated the category of ALI and classified ARDS as mild, moderate, and severe. Thus, the epidemiology of both ALI and ARDS has been included here.

### Environmental and genetic influences

ARDS develops after the insult from diverse etiologies discussed above. However, the heterogeneity of susceptibility and outcomes is intriguing. This could partially be explained by environmental and genetic influences. Research is still growing in this area.

From an environmental standpoint, literature from adult populations has shown increased risk of ARDS with alcohol abuse and smoking (active and passive) after blunt trauma. The association of passive smoking could be applied to the pediatric population.

From a genetic standpoint, a total of 34 genes has been reported to impact ARDS susceptibility. The majority of them are linked to the currently described pathophysiological pathways of ARDS. These include inflammation, epithelial cell function, endothelial cell function, coagulation, oxidative injuries, apoptosis, and platelet cellular process.The other reported genetic mutations associated with ARDS were linked to surfactant dysfunctionand to the epidermal growth factor gene polymorphism in males.

There is not enough literature suggesting ethnic differences for ARDS incidence and outcomes. The vast majority of initial genetic studies were in European populations. The literature is scant for other ethnic backgrounds. Thus far, approximately nine genes in African population and three genes in Asian population have been reported to be linked with ARDS. Studies have reported poor outcomes in African Americans with ARDS as compared to patients of other ethnicities. However, in one study, higher mortality was associated with greater severity of illness on presentation in Black patients. Higher mortality in Hispanic patients was not explainable by severity of illness on presentation in the same study.

Some of the epidemiologic studies have reported a slightly higher incidence of ARDS among male children (54-63%); however, the mortality (31% in male children) was not significantly different. One adult study reported higher mortality among males.

There is also not enough literature in the area of genetics pertinent to pediatric ARDS in the context of growing lungs and developing immunity.

**practice guidelines on pediatric acute respiratory distress syndrome (PARDS).**

The overarching goal is to support the patient's respiratory status, while placing an utmost focus on lung protection. Deviation from these guidelines can lead to increased mortality.

### Ventilation strategies

(1) Tidal volume (Vt)

The use of physiologic tidal volumes (6-8 mL/kg) is recommended to prevent ventilator-induced lung injury (VILI). If plateau and driving pressure exceed the upper limit, the use of 4-6 mL/kg might be necessary.

(2) Inspiratory pressures

An inspiratory plateau pressure of less than or equal to 28 cm H2O is recommended. If the chest wall compliance is reduced, it can be maintained at or below 32 cm of H2O. The driving pressure should be limited to 15 cm H2O.

(3) Permissive hypercapnia (to a lower limit pH of 7.20) may be allowed in pediatric patients with ARDS in order to remain within recommended pressure and tidal volume ranges as outlined above.

It is suggested that the routine use of bicarbonate supplementation be avoided. However, bicarbonate supplementation can be considered for patients in whom severe metabolic acidosis or pulmonary hypertension is adversely affecting cardiac function or hemodynamic stability.

### Oxygenation strategies

(1) Positive end-expiratory pressure (PEEP)

Titration of PEEP to oxygenation/oxygen delivery, hemodynamics, and compliance measured under static or quasi-static conditions is suggested.

It is recommended that PEEP levels be maintained at or above the lower PEEP/higher fraction of inspired oxygen (FiO2) table from the ARDS Network protocol.

ARDS Network PEEP/FiO2 table

|  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
| FiO2 | 0.3 | 0.4 | 0.5 | 0.6 | 0.7 | 0.8 | 0.9 | 1 |
| PEEP | 5 | 6-8 | 8-10 | 10 | 10-14 | 14 | 14-18 | 18-24 |

(2) Oxygen saturation (SpO2)

For pediatric patients with mild to moderate ARDS, it is suggested to maintain SpO2 between 92% and 97%.

In patients with severe PARDS, an SpO2 of less than 92% can be accepted after PEEP optimization. The goal is to reduce exposure to FiO2.

It is also suggested to avoid SpO2 of < 88% and > 97%.

### Other recommendations and good practice guidelines

(1) In pediatric patients with acute respiratory failure, NIV can be tried for a limited time. HFNC or CPAP can be used in resource-limited settings.

(2) A utilization of cuffed endotracheal tube (ETT) is always a good practice consideration.

(3) A nonroutine use of instilled saline for ETT suction can be considered.

(4) Daily assessment of extubation readiness with a spontaneous breathing trial can be considered.

(5) Continuous monitoring of respiratory rate, heart rate, SpO2, intermittent noninvasive blood pressure monitoring, spontaneous breathing efforts, end-tidal CO2, tidal volume, peak inspiratory pressure, plateau pressure, driving pressure, intrinsic PEEP, and flow-pressure-time curves should be considered.

(6) The committee was not able to recommend in favor of or against the use of prone positioning and alveolar recruitment maneuvers. Further research is ongoing in this area.

(7) The committee recommended against the use of surfactant.

(8) A meticulous consideration of inhaled nitric oxide (INO) therapy is recommended in severe ARDS cases and in cases bridging to extracorporeal life support (ECLS). If INO is being utilized, a careful assessment should be made for the benefits within the first 4 hours and throughout its utilization in the course of treatment. The goal is to discontinue the treatment in the lack of benefits to minimize the toxicity.

(9) Corticosteroids can be recommended in a limited population only.

(10) High-frequency oscillatory ventilation (HFOV)

The committee was not able to recommend a routine use of HFOV in all the cases. However, it can be considered when oxygenation and ventilation are unable to be achieved on a conventional ventilator with lung protection. While the patient is on HFOV, alveolar recruitment should be achieved by stepwise increase and decrease in mean airway pressure, with continuous monitoring of oxygenation, ventilation (CO2 assessment), and its impact on hemodynamics.

(11) Airway pressure release ventilation (APRV)

The committee was not able to recommend on any specific mode while on mechanical ventilation including controlled or assisted, APRV, or neurally adjusted ventilatory assist.

(12) Airway clearance regimen

The committee did not recommend the routine use of any specific mode of chest physiotherapy and mucolytic agents in patients with PARDS.

(13) Extracorporeal life support (ECLS) and extracorporeal membrane oxygenation (ECMO)

ECLS can be considered in severe PARDS cases when lung protective strategies failed to achieve adequate oxygenation and ventilation. Potentially reversible etiologies are an important part of this consideration. A final decision should be based on a thorough evaluation by the expert team. Serial evaluations are preferable over single evaluations. Venovenous ECMO is preferred over venoarterial in patients with adequate cardiac function. Avoidance of hyperoxia and slow correction of hypercapnia are recommended. The committee was not able to provide indications and guidelines on the use of extracorporeal CO2 removal.

(14) Sedation, withdrawal, and delirium

A careful adjustment of minimal yet effective sedation is recommended based on serial pain, sedation, and delirium scales. The goal is to optimize oxygen delivery, oxygen consumption, work of breathing, and spontaneous breathing efforts in conjunction with the aforementioned ventilator strategies.

Patients with PARDS who require 5 or more days of sedation should be carefully analyzed and possibly treated for iatrogenic withdrawal syndrome.

Daily assessment for delirium utilizing standardized delirium tools is recommended. Minimization of delirium using nonpharmacologic interventions, maintaining sleep hygiene, and family involvement should be carefully considered. The utilization of early mobilization and activity is suggested. The committee was not able to provide guidance for or against the use of melatonin or antipsychotics for the prevention or treatment of delirium.

(14) Neuromuscular blockade (NMB)

A minimal but effective NMB is recommended in addition to sedation in selected cases when protective and effective lung protective strategies can not be maintained. While utilizing NMB, train-of-four and other clinically validated tools, in addition to movements and mechanical ventilation parameters, should be monitored.

(15) Early initiation (< 72 hrs) of enteral nutrition is recommended with guided protocol in conjunction with an interprofessional team approach, with a daily protein intake of at least 1.5 g/kg/day.

(16) Fluid management should be adjusted to achieve adequate nutrition, oxygen delivery, and end-organ function preservation while avoiding fluid overload.

(17) Transfusion threshold for packed red blood cells was reduced to a hemoglobin level of 5 g/dL in the absence of hemolytic anemia.

### Follow-up and long-term management

(1) Patients with PARDS should be screened by a primary care pediatrician (PCP) by 3 months after discharge to assess for post-PICU morbidities, including but not limited to overall life quality, emotional and social functioning, pulmonary function tests, physical function, and cognitive and neurodevelopmental assessment, especially in patients requiring ECLS.

(2) A referral to a specialist can be made in case of any deficits identified in the initial screening by the PCP.

**predefined Q&A about Pediatric Acute Respiratory Distress Syndrome (ARDS)**

1. What is acute respiratory distress syndrome (ARDS) in children?

Answer: ARDS in children is a serious condition where the lungs become severely inflamed, leading to the inability to provide sufficient oxygen to the bloodstream . It is defined by acute onset hypoxemia that cannot be explained by cardiac failure . This syndrome involves a significant disruption of the alveolar-capillary membrane, leading to increased permeability and pulmonary edema, which reduces gas exchange efficiency .

## 2. What are the primary causes of ARDS in children?

Answer: ARDS in children is most commonly caused by pneumonia, sepsis, and aspiration . Other potential causes include trauma, burns, pancreatitis, inhalational injury, transfusion, and cardiopulmonary bypass .

## 3. What are the key features and symptoms of pediatric ARDS?

Answer: Symptoms typically include rapid breathing and low oxygen levels . In severe cases, children may require mechanical ventilation . Diagnosis is often made through chest X-rays, blood gas analysis, and clinical evaluation of the child’s symptoms and medical history .

## 4. How is ARDS treated in children?

Answer: Treatment focuses on several key areas:

* Addressing the underlying cause: This includes treating infections (e.g., early antibiotics) or controlling inflammatory sources .
* Lung protective ventilation strategies: This involves using low tidal volumes, limited peak pressures, high PEEP (Positive End-Expiratory Pressure), and permissive hypoxemia and hypercapnea to minimize further lung injury .
* Goal-directed fluid therapy .
* Supportive care: Includes oxygen therapy and mechanical ventilation if necessary .
* Rescue therapies: For severe cases not achieving adequate oxygenation, options like proning (lying face down), neuromuscular blockade, High-Frequency Oscillatory Ventilation (HFOV), and Extracorporeal Membrane Oxygenation (ECMO) may be considered on a case-by-case basis .
* Steroid therapy is generally advised against in all children with ARDS .

## 5. What is the prognosis for children with ARDS, and are there long-term effects?

Answer: The prognosis largely depends on factors such as the severity of ARDS and the child’s overall health status . Many children can recover with appropriate treatment . However, some children may experience lingering respiratory issues, decreased exercise tolerance, or psychological impacts such as anxiety or PTSD following hospitalization .

## 6. What happens to the lungs at a cellular level in ARDS?

Answer: ARDS involves the destruction of Type II pneumocytes, which are responsible for producing surfactant. Surfactant reduces surface tension in the alveoli and prevents their collapse. Their destruction leads to alveolar collapse and atelectasis. Lung remodeling occurs as these cells regenerate . The forces involved in mechanical ventilation can also promote inflammation and further lung injury (volutrauma, barotrauma, atelectrauma

**DOCTOR PATIENT CONVERSATION**

Doctor: Hello. I want to discuss your child’s condition called Acute Respiratory Distress Syndrome, or ARDS. It means that the lungs are inflamed and not working well to get enough oxygen into the blood.

Parent: What causes this? Is it serious?

Doctor: ARDS can happen after infections like pneumonia or sepsis, aspiration of food or liquids, trauma, or other serious illnesses. It is a serious condition but with modern treatments many children recover well.

Parent: What symptoms should I expect?

Doctor: Your child may breathe very fast, have difficulty breathing, or need help with oxygen or a ventilator. We often see changes on chest X-rays showing lung inflammation.

Parent: What treatments will my child need?

Doctor: Treatment focuses on supporting breathing and treating the underlying cause. We use gentle mechanical ventilation strategies to protect the lungs, carefully manage fluids to avoid overload, and sometimes use special therapies like prone positioning or advanced ventilation if needed.

Parent: Are there risks with the treatments?

Doctor: Yes, but we carefully balance the benefits and risks. For example, mechanical ventilation can sometimes cause lung injury if not managed carefully, so we use lung-protective strategies. We also monitor closely for infections or other complications.

Parent: How long will my child be in the hospital?

Doctor: It varies depending on severity and response to treatment. Some children improve in days, others may need weeks of support. We’ll keep you informed every step of the way.

Parent: What can I do to help?

Doctor: Being with your child, providing comfort, and communicating with the care team are very important. We also encourage you to ask questions anytime.

Parent: What should I watch for after discharge?

Doctor: Some children have lingering breathing difficulties or need follow-up for lung function. We’ll arrange appropriate outpatient care and support.

REFERENCES

[Acute Respiratory Distress Syndrome (ARDS)](https://my.clevelandclinic.org/health/diseases/15283-acute-respiratory-distress-syndrome-ards#outlook-prognosis)

[ARDS - Diagnosis and treatment - Mayo Clinic](https://www.mayoclinic.org/diseases-conditions/ards/diagnosis-treatment/drc-20355581)

<https://emedicine.medscape.com/article/803573-medication#2>

### **Esophageal atresia (EA)**

**DEFINITION AND DESCRIPTION**

Esophageal atresia is a birth defect (congenital malformation) that affects the way your baby’s esophagus develops. The esophagus is the swallowing tube that connects their mouth to their stomach. “Atresia” means that a passageway in the body is missing or closed. In esophageal atresia, the esophagus is closed at the bottom where it’s supposed to connect to your baby’s stomach. This makes it impossible for your baby to feed normally.

Esophageal atresia comes in several different forms, some of which can cause additional complications for your baby. Up to 90% of babies with esophageal atresia also have another birth defect called a tracheoesophageal fistula. This means their esophagus connects to their trachea — their windpipe — instead of their stomach. This can cause them to inhale or choke on what they swallow.

### **Types of esophageal atresia**

The different types of esophageal atresia (EA), with or without tracheoesophageal fistula (TEF), are defined by where the esophagus is closed and where it connects to the trachea, if it does.

* **Type A:** While it isn’t the most common, Type A is the most classical version of esophageal atresia. This version doesn’t include a tracheoesophageal fistula. Instead, the esophagus is simply closed at the bottom. It ends some distance above the stomach as a closed pouch.
* **Type B:** In Type B, the esophagus is closed at the bottom, and a tracheoesophageal fistula branches off from the upper part of the esophagus, connecting it to the trachea.
* **Type C:** This is the most common type of esophageal atresia. In Type C, the esophagus is in two separate pieces. The upper part that connects to the mouth ends in a closed pouch. The lower part connects to the stomach at the bottom and the trachea at the top.
* **Type D:** This is the rarest type, and also the most severe. In Type D, the esophagus is in two unconnected segments, and both segments have separate tracheoesophageal fistulas.

As congenital malformations go, esophageal atresia is relatively common, occurring in approximately 1 in 3,500 babies. About 85% of these are Type C. Other variations account for about 2% to 7% each. About half of babies with EA have one or more additional birth defects. Only 1% are associated with a genetic syndrome. EA occurs in 25% of babies with Edwards syndrome and 20% with VATER syndrome.

## **Symptoms**

Healthcare providers learn to recognize esophageal atresia by the “three Cs”:

* Coughing.
* Choking.
* Cyanosis (a bluish tint to the skin, a sign of low oxygen).

Additional signs and symptoms can include:

* Foamy mucus in your baby’s mouth.
* Excess saliva, spitting up or drooling.
* Gagging when attempting to feed.
* Respiratory distress.

While many different esophageal disorders can cause swallowing difficulties for babies, most don’t cause breathing difficulties the way esophageal atresia can. Both difficulties showing up together usually indicates EA with TEF.

### **CAUSES**

Esophageal atresia is a congenital malformation, which means something forms differently during fetal development, leaving a defect that’s present at birth. In typical fetal development, the esophagus and trachea begin as one tube, which later separates. Esophageal atresia (and often, tracheoesophageal fistula with it) occurs when this tube doesn’t finish developing and separating. This is the main cause.

What causes this developmental process to stall is another question. Researchers aren’t sure what the specific causes are, but they suspect genetic and environmental factors are involved. Genetic mutations are changes to your baby’s DNA that affect the way your baby develops. Mutations can be random or partly inherited. Environmental factors, like substances and stress, can make mutations more likely.

### **Risk factors associated with esophageal atresia**

Researchers haven’t identified the specific environmental factors that may be involved in triggering esophageal atresia. But they’ve observed certain commonalities among babies born with EA. These common factors aren’t direct causes, but they may indirectly raise the risk of EA. They include:

* Advanced maternal age (over 35) and/or paternal age (over 40).
* Assisted reproduction technologies, such as IVF (in vitro fertilization) or IUI (intrauterine insemination).
* Multiple births.

EA also commonly occurs with other congenital malformations and genetic syndromes. If a fetus was diagnosed with one of these conditions before birth, there’s a higher chance it may be born with esophageal atresia, as well. Some of the other birth defects and syndromes commonly associated with EA include:

* Trisomy (13, 18 or 21).
* VACTERL association.
* CHARGE syndrome.
* Congenital heart disease, especially ventricular septal defects, patent ductus arteriosus and tetralogy of Fallot.
* Other gastrointestinal (GI) atresias, including duodenal atresia, intestinal malrotation and imperforate anus.
* Kidney and genitourinary defects, such as horseshoe kidney or hypospadias.
* Spinal malformations, such as tethered spinal cord.
* Limb malformations.

## **Diagnosis and Tests**

Only a minority of fetuses are diagnosed with EA before birth. Prenatal diagnosis is more likely when the fetus has other abnormalities that are detectable before birth, which may lead to further tests. But early signs of EA can appear on your prenatal ultrasound even if no other conditions are evident. These signs typically appear on your standard 20-week ultrasound, which is also called an anatomy scan.

The most common prenatal sign of EA is that there’s more amniotic fluid around the fetus than there should be (polyhydramnios). Since the fetus usually swallows some of this fluid, too much left over suggests the fetus may not be able to swallow it. Your provider might also notice that the fetus’s stomach isn’t filled (small or missing stomach bubble). They might suggest following up with a fetal MRI (magnetic resonance imaging).

If your healthcare provider suspects EA before your baby is born, they’ll be able to test for it immediately after. If they don’t suspect it before birth, they’ll likely notice the signs soon after. The procedure to check for EA is to attempt to pass a tube from your baby’s mouth or nose through their esophagus to their stomach. If this passageway is closed (atresia), the tube won’t reach their stomach.

Your healthcare provider can confirm the diagnosis and determine the type of EA by taking X-rays. This will tell them the nature of the defect and whether your baby has fluid in their lungs or air in their stomach. After diagnosing esophageal atresia, your healthcare provider will check for other congenital malformations that commonly occur with it. Some of these may need treatment first, before your provider can treat EA.

**Management and Treatment**

Most of the time, surgery can fix the abnormality soon after your baby is born. Some babies may need to stay a little longer in the hospital, receiving nutrition and breathing support, before they’re ready for the operation. They may need more time to grow if they were born prematurely or if their esophagus is too short to repair. Some may need treatment for other life-threatening conditions first, like heart malformations.

**How can a baby eat with esophageal atresia?**

Babies with esophageal atresia will need medical assistance to eat until the abnormality can be fixed. Their healthcare providers will schedule surgery to correct it as soon as possible. In the meantime, they’ll receive nutrition through a tube (enteral nutrition) or a vein (parenteral nutrition). This will continue during their surgery and recovery, until they can safely transition to mouth feeding.

**TREATMENT**

Treatment for esophageal atresia includes stabilizing your baby’s breathing, providing safe nutrition and, ultimately, repairing the malformation through surgery. Some babies may be in the hospital longer than others.

#### **Initial management**

Immediate interventions for EA include:

* Suctioning of fluids from your baby’s esophagus.
* Installing a breathing tube to protect their airway.
* Installing a feeding tube or IV to deliver nutrition and fluids.
* IV antibiotics to prevent or treat pneumonia.

#### **Extended neonatal care**

Some babies may need to spend more time in the neonatal intensive care unit (NICU) before they’re ready for esophageal atresia surgery. This includes babies born prematurely, babies with multiple congenital malformations and babies with long-gap esophageal atresia (LGEA). Long-gap EA means that the two segments of the esophagus are too far apart to connect in surgery. They need more time to grow and close the gap.

##### **Traction process for LGEA**

Sometimes, healthcare providers can speed up the growth process for babies with long-gap EA using a type of traction. In this process, a surgeon places stitches on the two ends of your baby’s esophagus and threads them through a small incision in your baby’s back. Here, they can access the stitches and control their tension. Applying gentle traction encourages the esophagus segments to grow together faster.

#### **Surgical repair**

Your baby’s healthcare team will determine when they’re ready for surgery. The goals of esophageal atresia surgery are to:

* Connect separate segments of the esophagus together (anastomosis).
* Close off any connections between your baby’s esophagus and airway.

Depending on your baby’s condition, they may have one surgery for all of this, or they may have surgery in stages, addressing the fistulas first.

Surgery on your baby’s esophagus is called thoracic surgery. Thoracic means in their chest. When possible, surgeons use minimally invasive surgery methods to repair esophageal atresia. This means they access your baby’s chest through small incisions instead of opening it up. A surgeon inserts a small video camera (thoracoscope) through one micro-incision and operates through another, guided by the video.

#### **Recovery and follow-up**

After surgery, your baby will return to the NICU to recover. After several days, they’ll have an imaging test to look inside their esophagus and see how it’s healed. The test is called an esophagram. It’s a type of video X-ray (fluoroscopy) that looks at what happens when fluids pass through their esophagus. When fluids pass through without leaking, your baby will be ready to transition to oral feeding.

Your baby will need a little practice to learn to feed by mouth, and their healthcare team will monitor the process closely. They may continue to have some difficulties swallowing. Babies’ esophageal muscles might not work quite as they should. This can be a result of the original congenital malformation, a side effect of surgery or both. Their healthcare team will want to make sure they can get enough nutrition orally.

##### **Surgery complications**

Rarely, the repaired esophagus may continue to leak even after it’s had ample time to heal. This means the anastomosis failed for some reason. An anastomotic leak would require a second surgery to fix it.

Another possible complication is that the esophagus develops excessive scar tissue at the site of the anastomosis, which makes it too narrow (esophageal stricture). It might need stretching after surgery.

**Outlook / Prognosis**

Esophageal atresia alone is rarely fatal. Babies who are born prematurely, or who have other life-threatening conditions — like a heart abnormality — are more at risk. The survival rate for babies who receive treatment and don’t have other compromising conditions is near 100%. The survival rate for babies with either major heart abnormalities or birth weights below 3.5 lbs (1.6 kg) is 80%. For babies with both factors, it’s 50%.

While most children recover well and grow to adulthood, some may have lingering side effects from the esophageal atresia and the surgery to fix it. These conditions often improve over time, but they’re likely to require some additional treatment for at least a few years, if not longer.

Common long-term side effects include:

* **Tracheomalacia**. This means that the cartilage in their trachea (windpipe) is weak, causing the windpipe to partially collapse. It can cause wheezing or noisy breathing, sleep apnea and shortness of breath. Shallow breathing makes children more susceptible to airway infections, like pneumonia and bronchitis.
* **Swallowing difficulties**. Your child may continue to have some esophageal dysmotility — difficulties activating or coordinating their esophageal muscles. This can make eating difficult, especially when children transition to solid foods. You may need to pay special attention to what you feed them, make solid pieces smaller and make sure to serve them with ample liquids.
* **Gastroesophageal reflux disease (GERD)**. Esophageal dysmotility can also make chronic acid reflux more likely. The lower esophagus muscles may not be able to effectively keep stomach acid from rising into your child’s esophagus. This can cause uncomfortable symptoms, and over time, it can damage their esophagus tissues. Children may also inhale (aspirate) acid particles and damage their trachea. GERD occurs in up to half of children treated for esophageal atresia.

## **Living With**

Every child is different, and some children have more challenges than others following treatment for esophageal atresia. You can continue to consult their healthcare team for personal advice and support.

Some things they might advise include:

* **Delayed weaning**. For children with swallowing difficulties, the International Dysphagia Diet Standardization Initiative (IDDSI) has written specific guidelines for introducing solid foods in stages, beginning at age 3.
* **Consulting a specialist**. A speech-language pathologist (SLP) can help train children to use their swallowing muscles more effectively.
* **Vaccination**. Since children treated for EA are at a greater risk of chest infections, their healthcare team may recommend vaccines against COVID, flu, pneumonia and RSV.
* **GERD reevaluation**. Your child may have a prescription to treat GERD, but medication isn’t always enough. It’s worth checking as they get older to see if GERD is damaging their tissues. If it is, a minor procedure can help stop it.

**DIFFERENTIAL DIAGNOSIS**

* Tracheoesophageal fistula (TEF) (often occurs with EA but can also present alone)
* Laryngotracheoesophageal clefts — abnormal communication between the larynx, trachea, and esophagus
* Esophageal webs or rings — thin membranes causing partial esophageal obstruction
* Esophageal strictures — narrowing due to scarring or inflammation
* Tubular esophageal duplications — congenital cystic or tubular duplications of the esophagus
* Pharyngeal pseudodiverticulum — can mimic fistula on imaging, usually post-traumatic
* Congenital esophageal stenosis — intrinsic narrowing of the esophagus
* Gastrointestinal atresias or malformations — such as intestinal atresia, duodenal atresia, or imperforate anus (may coexist in syndromes like VACTERL)
* Neurologic or functional swallowing disorders — causing feeding difficulties without anatomical discontinuity
* Other congenital syndromes associated with EA — e.g., VACTERL association, Trisomy 13, 18, 21, CHARGE syndrome

**EPIDEMIOLOGY**

EA is a congenital malformation of the upper gastrointestinal tract with an estimated prevalence worldwide varying from 1 in 2500 to 1 in 4500 births. In the United States, the prevalence is estimated to be 2.3 per 10,000 live births.The relative incidence of EA/TEF increases with maternal age

**GENOMIC DATA**

* Genetic Syndromes and Associations:  
  EA/TEF can occur as part of several genetic syndromes, including:
  + VACTERL association: A multifactorial condition involving vertebral, anal, cardiac, tracheoesophageal, renal, and limb anomalies.
  + CHARGE syndrome: Caused by mutations in the *CHD7* gene, with about 10% of affected individuals having EA/TEF.
  + Feingold syndrome: Caused by mutations in *MYCN*, characterized by microcephaly, limb anomalies, and EA/TEF in 30–40% of cases.
  + Opitz G/BBB syndrome: Linked to mutations in *MID1* gene on X chromosome.
  + Other syndromes include Pallister-Hall syndrome, Fanconi anemia, and anophthalmia-esophageal-genital syndrome.
* Key Genes Identified:
  + *MYCN* (Feingold syndrome) — transcription factor important in development.
  + *CHD7* (CHARGE syndrome) — chromatin remodeling gene.
  + *MID1* (Opitz syndrome) — involved in microtubule stabilization.
  + Genes in the Sonic Hedgehog (SHH) signaling pathway, including *FOXF1*, *FOXC2*, and *FOXL1*, implicated in VACTERL-like phenotypes with gastrointestinal atresias.
* Chromosomal Aberrations and Copy Number Variations (CNVs):
  + Several rare CNVs have been identified in patients with EA/TEF, including deletions at 16q24.1 (FOXF1 cluster) and 22q11.2, which overlap with known syndromes and developmental delay loci.
  + De novo CNVs contribute to a minority of cases but are important in genetic diagnosis.

## **Approach Considerations**

The treatment plan for each baby must be individualized. The prognostic classifications can provide guidance in patients with multiple problems, but early and decisive identification of the most life-threatening anomaly is essential.

Management plans for a delayed repair of the esophageal atresia may include placing a 10-French Replogle double-lumen tube through the mouth or nose well into the upper pouch to provide continuous suction of pooled secretions from the proximal portion of the atretic esophagus. The baby may be positioned in the 45° sitting position. Prophylactic broad-spectrum antibiotics (eg, ampicillin and gentamicin) may be used. General supportive care and total parenteral nutrition (TPN) are needed.

With careful bedside attendance, these measures may permit a delay of days to perhaps weeks. Some have described cases in which the baby was discharged home with a Replogle tube in situ while waiting for staged repair of an esophageal atresia. However, deaths have been reported in infants in whom the tube did not maintain an empty upper pouch. A gastrostomy, distal tracheoesophageal fistula (TEF) ligation, or cervical esophagostomy may permit longer delays in the esophageal atresia repair. However, each intrusion carries a price.

If no distal TEF is present, a gastrostomy may be created. In such cases, the stomach is small, and laparotomy is required. In all cases of esophageal atresia in which a gastrostomy is created, care should be taken to place it near the lesser curvature to avoid damaging the greater curvature, which can be used in the formation of an esophageal substitute. When a baby is ventilated with high pressures, the gastrostomy may offer a route of decreased resistance, causing the ventilation gases to flow through the distal fistula and out the gastrostomy site. This condition may complicate the use of ventilation.

In cases such as those above or in cases in which a distal fistula continues to cause lung soiling, distal TEF ligation should be considered. This ligation is performed by means of a right-side thoracotomy, ideally via an extrapleural approach. The fistula may be clipped or simply ligated. If it is ligated and divided, subsequent staged repair of the esophageal atresia may be difficult because the distal esophageal segment tends to retract inferiorly to a substantial degree when it is detached from its tracheal mooring. However, simple fistula ligation may allow subsequent reopening of the fistula. Division of the fistula and attempts to anchor it at the midchest with sutures are usually unsuccessful.

A cervical esophagostomy or spit fistula may be constructed in the right or left side of the neck, depending on the choice for subsequent esophageal substitution. It allows drainage of the upper pouch and precludes aspiration from the upper pouch. Sham feeding may be commenced in cases in which a long delay to repair is anticipated. This feeding may prevent subsequent oral aversion, which is a real problem in babies who have not been fed by mouth in their early weeks to months of life. However, cervical esophagostomy usually dooms the child to some form of esophageal substitution.

In May 2017, the US Food and Drug Administration (FDA) approved the Flourish Pediatric Esophageal Atresia Anastomosis (Cook Medical) for management of esophageal atresia in infants up to 1 year old who do not have teeth and do not have a TEF (or have had a TEF repaired).The device closes the gap in the esophagus by using magnets to pull together the upper and lower portions of the esophagus. It is not indicated for use in patients in whom the distance between the esophageal segments is 4 cm or greater.

### Future and controversies

In the future, more accurate antenatal diagnosis and antenatal treatment may be possible. Minimally invasive techniques for repair with thoracoscopic surgery are now used in some centers, with good results.A better understanding of the pathoembryologic processes of this condition may reveal its causative agents or genetic factors. This knowledge, in turn, may lead to specific antenatal treatments or preventive techniques. In recent years, the incidence of this disorder has decreased, perhaps because of increased usage of antenatal folic acid supplements.

Debates continue about the best operative technique (eg, right-side or left-side thoracotomy) for patients with right-side aortic arches, suture type and technique, esophageal lengthening strategies, and procedures for mobilizing the distal esophagus.Other discussions include when to use cervical esophagostomy and the choice of esophageal replacement.The advent of esophageal atresia repairs that combine both minimally invasive and radiologic interventional techniques may be near.

**GUIDELINES**

The management of gastroesophageal reflux (GER) in esophageal atresia is particularly challenging; some advocate aggressive fundoplication, and others prefer more conservative medical treatment. In addition, the true incidence and treatment of tracheomalacia continues to be the subject of debate. Finally, the proper evidence-based guidelines for long-term follow-up remain to be established.

Tissue engineering of the esophagus may offer solutions for replacement of lost esophageal tissue.Experimental studies have shown promising results in the culture of esophageal epithelial cells and esophageal smooth-muscle cells. Viability of these cells on biodegradable scaffolds in vitro may provide the necessary replacement esophageal tissue in future.Studies in large animal models have shown promising results in the generation of rudimentary esophageal tissue using tissue engineering and regenerative medicine technology

**PREDEFINED Q AND A**

## 1. What is esophageal atresia?

Answer:  
Esophageal atresia is a congenital birth defect where the esophagus (the tube connecting the mouth to the stomach) does not form properly and ends in a blind pouch instead of connecting to the stomach. This prevents food and liquids from passing normally to the stomach.

## 2. How common is esophageal atresia?

Answer:  
Esophageal atresia occurs in about 1 in 3,500 live births. About 85% of cases are the most common type (Type C), where the upper esophagus ends blindly and the lower esophagus connects abnormally to the trachea (tracheoesophageal fistula).

## 3. What causes esophageal atresia?

Answer:  
It occurs during fetal development when the esophagus and trachea fail to separate properly from a single tube. The exact cause is unknown but may involve genetic and environmental factors. It can also be associated with syndromes like VACTERL or Edwards syndrome.

## 4. What are the symptoms of esophageal atresia?

Answer:  
Newborns with EA often have difficulty swallowing, excessive drooling, coughing, choking, and may have breathing problems. They may also have frothy bubbles in the mouth and cyanosis (bluish skin) during feeding.

## 5. How is esophageal atresia diagnosed?

Answer:  
Diagnosis is suspected if a feeding tube cannot be passed into the stomach after birth. Chest and abdominal X-rays confirm the diagnosis by showing the tube coiled in the upper esophageal pouch and absence of air in the stomach (unless a fistula is present).

## 6. What is the treatment for esophageal atresia?

Answer:  
Surgery is required to reconnect the two ends of the esophagus and to close any abnormal connection to the trachea (fistula). Surgery is usually done soon after birth. Feeding is initially done intravenously or via a feeding tube until healing occurs.

## 7. What are the possible complications after surgery?

Answer:  
Complications may include leakage at the repair site, esophageal strictures (narrowing), gastroesophageal reflux, and feeding difficulties. Some children may require additional procedures like esophageal dilation or fundoplication to manage reflux.

## 8. What is the long-term outlook for children with esophageal atresia?

Answer:  
With appropriate surgical repair and follow-up care, most children grow and develop normally. Some may have ongoing issues with swallowing, reflux, or respiratory infections, requiring multidisciplinary care.

## 9. Can esophageal atresia be detected before birth?

Answer:  
Sometimes prenatal ultrasound shows signs such as polyhydramnios (excess amniotic fluid) or absence of a visible stomach bubble, which may suggest EA. However, definitive diagnosis is usually made after birth.

## 10. What support is available for families?

Answer:  
Specialized centers offer multidisciplinary care including surgeons, pulmonologists, nutritionists, and therapists to support children and families throughout treatment and recovery

**DOCTOR PATIENT CONVERSATION**

Doctor: Hello, I want to talk with you about your baby’s diagnosis. Your child has a condition called esophageal atresia, which means the esophagus—the tube that connects the mouth to the stomach—did not form properly. Instead, it ends in a blind pouch and does not connect to the stomach.

Parent: What does that mean for my baby? How serious is it?

Doctor: This condition affects feeding and breathing because food and saliva cannot pass normally into the stomach, and sometimes there is an abnormal connection between the esophagus and the windpipe, called a tracheoesophageal fistula. It is serious but treatable, and we have specialized teams experienced in managing this.

Parent: How is it treated?

Doctor: Your baby will need surgery to connect the two ends of the esophagus and close any abnormal connection to the windpipe. This is usually done within the first day or two of life. In some cases, if the gap between the esophageal ends is large, we may use specialized techniques to gradually lengthen the esophagus or consider other surgical options.

Parent: What happens after surgery? Will my baby be able to eat normally?

Doctor: After surgery, your baby will be closely monitored in the neonatal intensive care unit. Initially, feeding may be through a tube placed directly into the stomach. Over time, as healing occurs, your baby will start feeding by mouth. Some children may have issues like reflux or narrowing at the repair site, which we manage with medications or additional procedures if needed.

Parent: Are there risks or complications?

Doctor: Like any surgery, there are risks such as infection, leakage at the repair site, or narrowing of the esophagus. Some babies may also have breathing issues or lung problems, especially if born prematurely. Our multidisciplinary team will monitor and support your baby throughout recovery.

Parent: How long will my baby stay in the hospital?

Doctor: It depends on how your baby recovers, but typically several weeks. We will keep you informed and involve you in daily care decisions. Many babies go on to live healthy lives with appropriate follow-up.

Parent: Can this be detected before birth?

Doctor: Sometimes prenatal ultrasounds show signs like excess amniotic fluid or an absent stomach bubble, which can raise suspicion. However, the definitive diagnosis is usually made after birth.

Parent: Thank you for explaining everything. What should I do now?

Doctor: We will prepare for surgery and ensure your baby is stable. Please feel free to ask any questions at any time. We are here to support you and your family.

REFERNCES

[Esophageal Atresia: Causes, Symptoms, Diagnosis & Treatment](https://my.clevelandclinic.org/health/diseases/21178-esophageal-atresia#overview)

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### **Tracheoesophageal fistula**

**DEFINITION AND DESCRIPTION**

Typically, your esophagus (the hollow tube that connects your throat to your stomach) and trachea (windpipe) are separate. People with tracheoesophageal fistula (TEF) have an abnormal connection between these two structures. This results in aspiration (inhalation) of food and liquids into your lungs. Tracheoesophageal fistula commonly occurs with esophageal atresia (EA) — a condition that’s characterized by an abnormal connection between your esophagus and stomach.

### **Who does tracheoesophageal fistula affect?**

Tracheoesophageal fistula is a congenital condition. In other words, people are born with it. Most cases of TEF are diagnosed and treated in infancy.

It’s also possible for adults to develop tracheoesophageal fistula as the result of esophageal cancer or lung cancer, infections (such as tuberculosis) or trauma from a medical procedure. This is called acquired tracheoesophageal fistula.

It’s estimated that tracheoesophageal fistula affects 1 in every 3,000 to 5,000 births in the United States. Approximately 50% of babies with TEF or EA have another congenital condition.

Acquired tracheoesophageal fistula, which occurs in adulthood, isn’t common. Very rarely, adults can have congenital tracheoesophageal fistula. This means that they were born with the condition but didn’t develop symptoms until later in life.

### **Types of tracheoesophageal fistula**

Tracheoesophageal fistula is generally categorized into five different types:

* **Type A:** There’s no TEF, only EA. Your esophagus is divided into two parts, with both portions ending in blind pouches (cavities that are closed at one end). This is also commonly referred to as pure esophageal atresia. It makes up about 8% of all cases.
* **Type B:** This rare form of TEF affects about 2% of all cases. The lower portion of your esophagus ends in a blind pouch, and the upper portion is connected to your windpipe by a tracheoesophageal fistula.
* **Type C:** The most common form of TEF, type C is when the upper portion of your esophagus ends in a blind pouch, and the lower portion is connected to your trachea by a tracheoesophageal fistula. About 85% of babies born with TEF have this type.
* **Type D:** In this rarest form of TEF, a tracheoesophageal fistula connects both the upper and lower portions of your esophagus to your trachea. Less than 1% of babies born with TEF have this type.
* **Type E:** Your esophagus connects to your stomach normally and is fully intact. However, a tracheoesophageal fistula connects your esophagus and trachea. Type E affects about 4% of TEF cases.

## **Symptoms**

Tracheoesophageal symptoms depend on whether EA is present, as well as TEF. Babies who are born with TEF but not EA usually don’t show symptoms at birth. Over time, however, they may:

* Have frequent lung infections.
* Cough when feeding.

Babies who have esophageal atresia with tracheoesophageal fistula usually exhibit symptoms immediately after delivery. The most common symptoms include:

* Coughing.
* Choking when trying to swallow.
* Breathing problems.

### **CAUSES**

During fetal development, your esophagus and trachea form as one single tube. Typically, about four to eight weeks after conception, a wall develops between these structures, making them two distinct tubes. If this wall doesn’t form properly, it can result in tracheoesophageal fistula.

## **Diagnosis and Tests**

In rare instances, your healthcare provider may suspect a tracheoesophageal fistula before your baby is born due to abnormalities on an ultrasound. Most of the time, however, your medical team may suspect TEF within a few hours of delivery if your baby has excessive mucous, breathing difficulties or is unable to swallow.

To confirm a TEF or EA diagnosis, your healthcare provider will likely take X-rays of their chest and abdomen. In addition, they may recommend an endoscopy or bronchoscopy. These tests allow them to look inside of the airways using a thin tube and a lighted camera. If your baby is diagnosed with TEF or EA, your healthcare provider will probably order additional tests to identify or rule out other congenital conditions.

## **Management and Treatment**

Tracheoesophageal fistula treatment involves corrective surgery. During this procedure, the connection between your baby’s esophagus and trachea is repaired. This may be done using traditional or minimally invasive techniques.

#### **Are there risks involved with tracheoesophageal fistula repair?**

As with any surgical procedure, tracheoesophageal fistula repair may be accompanied by complications. Possible risks include anastomotic leaks (when fluid leaks out from the area where the esophagus and trachea were joined), esophageal strictures (abnormal tightening of the esophagus) and damage to the laryngeal nerve.

There’s also a chance that TEF could come back later. Approximately 3% to 14% of TEF repairs result in fistula recurrence (return).

### **Recovery from tracheoesophageal fistula treatment**

Recovery time depends on several factors, including the severity of your baby’s condition and how well they respond to treatment. In most cases, full healing takes up to 12 weeks.

## **Outlook / Prognosis**

If your baby has TEF, your healthcare provider will recommend surgery to address the problem. The extent of surgery depends on the type of TEF. If your baby has postoperative complications, your medical team will keep your baby in the hospital for a few days to monitor their progress.

Babies who have complications after their initial surgery are more likely to experience recurrence. Therefore, if your baby has postoperative complications, your healthcare provider will perform periodic follow-ups.

#### **Is tracheoesophageal fistula curable?**

Yes. Tracheoesophageal fistula is curable with surgical intervention. Because it’s a life-threatening condition, it should be treated immediately.

**Prevention**

Because people are born with congenital TEF, there’s no way to prevent it from happening. And since most cases of acquired TEF are caused by cancer and infections, there’s no known way to reduce your risk for the condition.

**Living With**

If your baby has TEF, your healthcare provider will recommend surgery to address the problem. The extent of surgery depends on the type of TEF. If your baby has postoperative complications, your medical team will keep your baby in the hospital for a few days to monitor their progress.

Babies who have complications after their initial surgery are more likely to experience recurrence. Therefore, if your baby has postoperative complications, your healthcare provider will perform periodic follow-ups.

## **Diagnostic Considerations**

Consider adult presentation of congenital tracheoesophageal fistula (TEF) (ie, recurrent TEF) in adults who present with cough and recurrent aspiration pneumonia. [[11](javascript:void(0);)] Recurrent TEF in adults may be a late complication of surgical repair performed when they were children.

Pharyngeal pseudodiverticulum should be considered in the differential diagnosis of TEFs. This may occur secondary to traumatic perforation of the posterior pharynx from finger insertion into the oropharynx during labor or following vigorous efforts at tube insertion during resuscitation of the newborn. These patients develop pneumomediastinum.

A very rare cause of neonatal respiratory distress is tracheal agenesis, which is always fatal within hours of birth. In tracheal agenesis, a nasogastric tube can be inserted easily.

Zenker diverticulum is also known as posterior hypopharyngeal pouch and pharyngoesophageal diverticulum. This condition involves herniation of mucosa and submucosa through the oblique and transverse fibers of the cricopharyngeus muscle. The blind pouch develops at the pharyngoesophageal junction at the level of the C5-C6 disc space. The pouch is the result of hyperdynamic cricopharyngeal sphincter contraction associated with an abnormality of cricopharyngeal relaxation.

## Other considerations

The International Network on Oesophageal Atresia (INoEA) consensus statements on the transition of patients with EA-TEF notes that asthma symptoms in adults with EA-TEF can result from aspiration and/or tracheomalacia rather than classic asthma.

Vocal cord paralysis is a differential diagnosis in adolescents and adults with EA-TEF who have persistent swallowing dysfunction and chronic respiratory symptoms.Consider laryngoscopy as a key diagnostic modality in these patients.

In the setting of persistent respiratory and dysphagia symptoms, particularly if a previous history exists for long-gap EA or congenital cardiac disease, maintain an index of suspicion for a vascular anomaly.If there is a clinical concern for this diagnosis, evaluate further.

## **Differential Diagnoses**

* Aspiration Pneumonitis and Pneumonia
* Esophageal Cancer
* Esophageal Diverticula
* Esophageal Rupture
* Esophageal Stricture
* Esophagitis
* Gastroesophageal Reflux Disease
* Respiratory Failure
* Tracheal Tumors
* Tracheomalacia
* Zenker Diverticulum

## **Epidemiology**

### United States data

TEFs are a common congenital anomaly with an incidence of 1 case in 2000-4000 live births. Acquired TEFs are quite rare, and incidence rates have not been well documented.

Acquired nonmalignant TEFs occur in approximately 0.5% of patients undergoing tracheostomy.The incidence of malignant TEFs was reported at 4.5% for primary malignant esophageal tumors, and 0.3% for primary malignant lung tumors.Other investigators have reported the incidence of TEFs secondary to esophageal carcinoma to be 4.3-8.1%.

### Race- and age-related demographics

No racial predilection is apparent.

Congenital TEFs are primarily observed in neonates and during the first year of life. Adults rarely present with congenital TEFs that were undiagnosed during their early years of life.

Acquired TEFs may occur in individuals of any age, and elderly individuals are at increased risk if they become ventilator dependent because of respiratory failure.

## **Procedures**

Flexible esophagoscopy or flexible bronchoscopy may be useful in the diagnosis of acquired TEFs. Either or both of these procedures may be required to evaluate the anatomy of these structures and to exclude an unsuspecting mucosal lesion. The role of endoscopic procedures is especially important in localizing the acquired nonmalignant or malignant TEF.

## **Treatment Recommendations**:

* Systematic Evaluation:  
  Children with EA/TEF require ongoing multidisciplinary assessment for GI complications such as gastroesophageal reflux (GER), dysphagia, feeding difficulties, anastomotic strictures, and respiratory symptoms. Regular surveillance including endoscopy, pH monitoring, and imaging is recommended based on symptoms.
* Acid Suppression Therapy:
  + It is recommended to treat GER with proton pump inhibitors (PPIs) starting in the neonatal period to prevent peptic complications and anastomotic strictures.
  + Treatment duration is generally at least 12 months post-surgery, but may be extended depending on persistence of reflux symptoms.
  + Acid suppression should be used cautiously in patients with extra-esophageal reflux manifestations.
* Management of Anastomotic Strictures:
  + Strictures should be dilated only when associated with significant symptoms such as feeding difficulties or recurrent respiratory infections.
  + Endoscopic evaluation and biopsies are advised to exclude eosinophilic esophagitis (EoE) before dilation or surgery.
* Fundoplication (Anti-Reflux Surgery):
  + Indicated in patients with severe GERD refractory to medical management, recurrent strictures (especially in long-gap EA), cyanotic spells, or long-term feeding tube dependency.
  + Preoperative workup should include barium contrast studies, endoscopy with biopsies, and pH or pH-impedance monitoring.
  + Post-fundoplication dysphagia warrants thorough evaluation including contrast studies and manometry.
* Investigation of Life-Threatening Events:
  + Multidisciplinary evaluation is essential before surgical intervention in patients with acute life-threatening events (ALTE), cyanotic spells, or brief resolved unexplained events (BRUE).
* Exclusion of Other Causes:
  + In children with persistent symptoms, other anatomical abnormalities such as vascular rings, recurrent or missed fistulae, congenital esophageal stenosis, and laryngeal clefts must be excluded.
* Long-Term Follow-Up:
  + Regular follow-up by a multidisciplinary team including pediatric gastroenterologists, surgeons, pulmonologists, and nutritionists is recommended to monitor growth, nutrition, respiratory health, and quality of life.

**PREDEFINED Q AND A**

## 1. What is tracheoesophageal fistula (TEF)?

Answer:  
TEF is a congenital or acquired abnormal connection between the trachea (windpipe) and the esophagus (food pipe). It often occurs with esophageal atresia (EA), where the esophagus ends in a blind pouch. This abnormal connection can cause feeding difficulties and respiratory problems.

## 2. How is TEF diagnosed?

Answer:  
Diagnosis is suspected when a newborn has feeding difficulties, choking, coughing, or respiratory distress. Passing a nasogastric tube may be impossible or abnormal. Confirmation is by chest and abdominal X-rays showing the tube coiled in the upper esophagus and air in the stomach if a fistula is present. Contrast studies, endoscopy, and bronchoscopy can further delineate anatomy.

## 3. What are the common complications associated with TEF?

Answer:  
Complications include aspiration pneumonia, recurrent respiratory infections, gastroesophageal reflux disease (GERD), anastomotic strictures after repair, esophageal dysmotility, and feeding difficulties. Life-threatening events like cyanotic spells may also occur.

## 4. What is the standard treatment for TEF?

Answer:  
Surgical repair is required, typically soon after birth, to close the fistula and restore esophageal continuity. Postoperative care includes management of GERD, nutritional support, and monitoring for complications.

## 5. How is gastroesophageal reflux (GER) managed in children with TEF?

Answer:  
Routine use of proton pump inhibitors (PPIs) is recommended for at least 12 months post-surgery to reduce reflux and prevent complications like strictures. Multidisciplinary follow-up is important even if symptoms are absent, as silent reflux and esophagitis are common.

## 6. When is anti-reflux surgery (fundoplication) indicated in TEF patients?

Answer:  
Fundoplication is considered for patients with recurrent anastomotic strictures, poorly controlled GERD despite maximal medical therapy, long-term dependence on transpyloric feeding, or cyanotic spells. Before surgery, thorough evaluation including endoscopy, pH monitoring, and imaging is essential.

## 7. How are anastomotic strictures diagnosed and treated?

Answer:  
Strictures are diagnosed by symptoms such as feeding difficulties, regurgitation, cough, or recurrent respiratory infections, and confirmed by endoscopy or contrast studies. Dilatation is recommended only for clinically significant strictures causing symptoms.

## 8. What other conditions should be excluded in symptomatic TEF patients?

Answer:  
Other causes of symptoms such as eosinophilic esophagitis (EoE), laryngeal clefts, vocal cord paralysis, missed or recurrent fistula, congenital esophageal stenosis, and vascular rings should be ruled out before further interventions.

## 9. What is the recommended follow-up for children with repaired TEF?

Answer:  
Regular multidisciplinary follow-up including pediatric gastroenterology, pulmonology, and otolaryngology is recommended. Surveillance endoscopy with biopsies is advised even in asymptomatic patients to detect esophagitis or Barrett’s changes early.

## 10. What is the prognosis for children with TEF?

Answer:  
With timely surgical repair and appropriate multidisciplinary care, most children survive and have good long-term outcomes. However, many face chronic issues like reflux, feeding difficulties, and respiratory problems requiring ongoing management.

**DOCTOR PATIENT CONVERSATION**

Doctor: Hello. I want to talk with you about your child’s diagnosis. Your child has a condition called tracheoesophageal fistula, or TEF. This means there is an abnormal connection between the windpipe (trachea) and the food pipe (esophagus).

Parent: What does that mean? How does it affect my child?

Doctor: Normally, the trachea and esophagus are separate tubes. In TEF, there’s a small passage connecting them, which can cause food or saliva to enter the lungs, leading to coughing, choking, and infections. It also often occurs with esophageal atresia, where the esophagus is not properly connected to the stomach.

Parent: How did this happen? Is it something we caused?

Doctor: TEF is a congenital condition, meaning it developed before birth. It’s not caused by anything you did. Sometimes it can also be acquired later due to injury or illness, but in your child’s case, it’s congenital.

Parent: How do you fix it?

Doctor: The main treatment is surgery. The surgeon will separate the trachea and esophagus, close the abnormal connection, and repair the esophagus so your child can swallow safely. The exact approach depends on the type and location of the fistula.

Parent: Will my child need surgery right away?

Doctor: Usually, surgery is done soon after birth once your child is stable. Sometimes, if the esophageal ends are far apart, we may need to delay or use special techniques to connect them safely.

Parent: What happens after surgery? Will my child be able to feed normally?

Doctor: After surgery, your child will be monitored closely. Feeding might start through a tube initially, then gradually by mouth as healing occurs. Some children may experience reflux or narrowing at the repair site, which we manage with medications or additional procedures if needed.

Parent: Are there risks or complications?

Doctor: Like any surgery, there are risks such as infection, leakage at the repair site, or narrowing of the esophagus. We also watch for breathing problems and infections. Our team will support you and your child throughout recovery.

Parent: How long will my child stay in the hospital?

Doctor: It varies, but typically several weeks. We will keep you informed and involve you in care decisions.

Parent: What should I watch for at home?

Doctor: Watch for coughing, choking during feeding, difficulty breathing, or fever. If you notice these, contact us immediately.

Parent: Thank you for explaining everything.

Doctor: You’re welcome. Please feel free to ask any questions anytime. We’re here to support you and your child.

**GENOMIC DATA**

* Genetic Syndromes and Associations:  
  TEF is commonly associated with several genetic syndromes, including:
  + VACTERL association (Vertebral anomalies, Anal atresia, Cardiac defects, Tracheoesophageal fistula, Renal anomalies, Limb abnormalities). This is a multifactorial condition with no single genetic cause but often involves microdeletions in the FOXF1 gene cluster (16q24.1).
  + CHARGE syndrome, caused by mutations in the *CHD7* gene, with ~10% of affected individuals having TEF.
  + Feingold syndrome (mutations in *MYCN*).
  + Other syndromes include Pallister-Hall syndrome, Fanconi anemia, and chromosomal abnormalities such as trisomy 18, 13, and 21.
* Key Genes and Pathways:
  + The FOXF1 gene cluster at 16q24.1 is implicated in a VACTERL-like phenotype including TEF.
  + Genes involved in the Sonic Hedgehog (SHH) signaling pathway play a role in foregut development; mutations in these genes have been shown in animal models to cause EA/TEF-like malformations.
  + Mutations in *CHD7* (CHARGE syndrome) and *MYCN* (Feingold syndrome) have confirmed roles in human cases.

REFERNCES

[Tracheoesophageal Fistula: Types, Symptoms & Treatment](https://my.clevelandclinic.org/health/diseases/23395-tracheoesophageal-fistula#overview)

### [**https://emedicine.medscape.com/article/186735-overview**](https://emedicine.medscape.com/article/186735-overview)

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### **Alpha-1 antitrypsin deficiency**

**DEFINITION AND DESCRIPTION**

Alpha-1 antitrypsin deficiency (sometimes just called “Alpha-1”) is an inherited genetic disorder that causes low levels of a protein (AAT) that protects your lungs. Alpha-1 increases your risk of developing certain diseases, including emphysema (damaged air sacs in your lungs), cirrhosis (liver scarring) and panniculitis (an uncommon skin condition). Some of these conditions can be life-threatening.

**Alpha-1 is sometimes referred to as “genetic COPD” or “genetic emphysema.”**

Alpha-1 antitrypsin deficiency affects people who have two copies of the *SERPINA1* gene that makes an abnormal type of the Alpha-1 protein. Genes are the instructions for how your body should function.

These gene changes can cause your body to have low levels of AAT or no AAT in your lungs, and, depending on the changes, a buildup of AAT in your liver. Any of these changes can cause health issues. People with one abnormal copy of the gene and one normal copy (called Alpha-1 carriers) can also have symptoms and are at an increased risk of lung damage, especially if they smoke.

Alpha-1 is one of the most common genetic disorders among those with European ancestry, but it’s uncommon in people of non-European descent. One in 25 people of European descent have at least one abnormal copy of the gene for Alpha-1. In 1 in about 3,500 people in the U.S., both of their genes for AAT are abnormal, putting them at risk for severe organ damage. About 75% of people with two malfunctioning genes will eventually develop lung function issues.

#### **How does Alpha-1 deficiency affect my lungs and liver?**

Alpha-1 antitrypsin (AAT) is a protein that forms in your liver and moves through your bloodstream to your lungs. It’s the “off switch” for an enzyme called neutrophil elastase. Neutrophil elastase is important for fighting infections in your lungs, but it can also destroy your healthy lung tissue. After elastase has had time to help fight an infection, AAT shuts it off (inhibits) so it won’t damage your lungs.

If a gene mutation causes low levels of AAT or creates incorrectly formed AAT, you won’t have enough in your lungs to stop elastase, which will start breaking down the protein elastin in your lungs. Elastin gives strength to the small air sacs of your lungs (alveoli) and allows them to stretch and contract, like a rubber band. Without it, your alveoli lose their shape and become floppy. This makes it so you can’t breathe or get oxygen properly. This is a condition called emphysema.

Gene mutations that change the shape of AAT keep it from moving out of your liver. It builds up there and can cause scarring. Since it can’t get out of your liver, it’s not able to move to your bloodstream and your lungs.

Alpha-1 isn’t necessarily a terminal illness. Many people with Alpha-1, especially if they don’t smoke, can live a normal life span.

**Symptoms**

Lung diseases caused by Alpha-1 have symptoms similar to chronic obstructive pulmonary disease (COPD). Lung symptoms usually start between the ages of 30 and 50 and include:

* Shortness of breath (dyspnea), especially with exercise or exertion.
* A whistling sound when you breathe (wheezing).
* Chronic cough, often with mucus.
* Extreme tiredness.
* Frequent chest colds.

About 10% of infants and 15% of adults with Alpha-1 develop liver disease. Signs and symptoms of liver disease may include:

* Yellowing of the skin and eyes (jaundice).
* Itchy skin.
* Swelling in your legs or abdomen (ascites).
* Throwing up blood.

Rarely, your first symptoms of Alpha-1 are painful, red bumps on your skin (panniculitis). These can move around on your body and may break open, leaking fluid or pus.

**CAUSES**

Everyone has two sets of genes (the instructions for how your body functions), one from each parent. Just like differences in these instructions can determine what color eyes or hair you have, they can also change how your body functions. Differences (mutations) in a specific gene cause Alpha-1 antitrypsin deficiency.

The *SERPINA1* gene makes a protein called Alpha-1 antitrypsin (AAT) that protects your lungs from damage caused by another protein in your body (neutrophil elastase, which attacks infections in your lungs). There are many *SERPINA1* mutations that can change how your body makes AAT. Some tell your body to make less AAT, some tell your body not to make any at all, and some cause AAT to form incorrectly so it can’t get to your lungs. Any of these can cause you to have too little AAT in your lungs to protect them.

If both copies of your *SERPINA1* gene have mutations, you have Alpha-1 antitrypsin deficiency. Depending on the types of abnormal genes, you have a 75% chance of developing lung symptoms. If you have a mutation in just one of your copies of the gene (carrier), your body can usually make enough functioning AAT to protect your lungs. You’re still at an increased risk for lung damage and could eventually develop symptoms, especially if you smoke.

#### **Alpha-1 inheritance**

Carriers of abnormal Alpha-1 genes can pass the mutation on to their children. If both parents are carriers, their children have a 25% chance of having two abnormal genes and a 50% chance of being carriers (one abnormal copy and one normal copy). Since both genes contribute to how you make AAT rather than one being dominant, Alpha-1 antitrypsin deficiency is said to have a codominant inheritance.

**Diagnosis and Tests**

A provider diagnoses Alpha-1 with blood tests. Because it has symptoms of other illnesses, sometimes it can take a long time to diagnose Alpha-1. You might be tested for Alpha-1 if you have liver symptoms or if you’ve received a COPD diagnosis. Tests and procedures your provider might perform include:

* **Blood tests.** A provider takes a sample of your blood to measure your levels of AAT and understand how well your liver is working. If you have low levels of AAT, they’ll do genetic testing to identify gene differences associated with Alpha-1.
* **Imaging.** X-rays and CT scans can show signs of Alpha-1 in your lungs and rule out other conditions. These tests can show the location of any damage and how severe it is.
* **Pulmonary function tests.** These tests can’t diagnose Alpha-1, but they can tell your provider how well your lungs are working. They often involve breathing into a machine that measures your lung function.
* **Liver ultrasound or elastography.** If your provider suspects issues with your liver, they may get a liver ultrasound or elastography ultrasound (FibroScan®) to see if there’s any scarring.
* **Liver biopsy.** If you have liver damage, your provider may take a small sample of tissue (biopsy) from your liver to determine how severe the damage is.

## **Management and Treatment**

For those with lung conditions from Alpha-1, your provider can treat you with COPD medications and therapies, like bronchodilators and pulmonary rehabilitation. If you have emphysema due to very low levels of Alpha-1 in your blood, they may recommend augmentation therapy. Augmentation therapy delivers normal Alpha-1, collected and purified from blood donors, through an IV. Augmentation therapy can slow the progression of emphysema.

If Alpha-1 affects your liver, your provider may be able to treat some of the symptoms, but only a liver transplant can cure Alpha-1 by restoring normal AAT production.

Not smoking or drinking reduces your risk of lung and liver damage from Alpha-1. It’s recommended that you get vaccinated to help prevent viral hepatitis and pneumonia.

### **Medications/treatments are used**

Depending on where Alpha-1 affects you, treatment options may include:

* **Augmentation therapy.** Your provider can increase your AAT levels by giving you supplemental normal AAT (collected and purified from blood donors) directly into a vein (IV infusion). This can’t reverse lung damage but can prevent future damage. It doesn’t prevent liver damage from Alpha-1.
* **Medication**. Inhaled corticosteroids and bronchodilators can make it easier to breathe by reducing inflammation and opening your airways.
* **Oxygen therapy.** If your oxygen levels are low, your provider may prescribe supplemental oxygen, which a machine delivers through a mask on your face or through small tubes in your nose.
* **Pulmonary rehabilitation.** Breathing exercises and physical therapy can make breathing easier.
* **Smoking cessation therapy.** If you smoke, your provider can recommend therapies to help you quit.
* **Lung transplant.** If your lungs are severely damaged, getting a healthy lung through a transplant can help improve your quality of life.
* **Liver transplant.** If your liver is badly scarred, your provider may recommend a liver transplant. A healthy liver should make normal AAT.

You should avoid alcohol if you have certain types of Alpha-1 or are a carrier. Alcohol can increase your chances of liver damage. You should also avoid medications that can cause liver damage. A provider can help you identify which medications to avoid.

## **Outlook / Prognosis**

Your provider can tell you what to expect in your specific situation. Some people with Alpha-1 never have symptoms or related organ damage, especially if they never smoke. Others can have life-threatening complications. Follow your provider’s recommendations to get the best outcome possible.

#### **Complications of Alpha-1**

Complications of Alpha-1 can affect your lungs, liver or other organs and include:

* Progressive lung conditions (like COPD).
* Permanent damage to your airways (emphysema, bronchiectasis).
* High blood pressure in the arteries leading from your heart to your lungs (pulmonary hypertension).
* Liver scarring (cirrhosis).
* Liver cancer (hepatocellular carcinoma).
* Heart, liver or respiratory failure.
* Inflammation of the fat under your skin (panniculitis).

The life expectancy of someone with Alpha-1 varies widely from person to person. Some people live a normal life span and some have life-threatening complications. Your prognosis will depend on:

* How quickly you’re diagnosed.
* The type of Alpha-1 you have and how it affects your body.
* The amount of organ damage you have at diagnosis.
* How well your lungs are working.
* How quickly lung or liver disease is getting worse.
* If you smoke, whether or not you continue to smoke after diagnosis. Smoking reduces your life expectancy with Alpha-1.

## **Prevention**

Because it’s inherited (you get it from your parents and are born with it), you can’t prevent Alpha-1. But that doesn’t mean you’ll develop the diseases it can cause. Even with an Alpha-1 diagnosis, there are several things you can do to reduce your risk of organ damage, including:

* Don’t smoke or vape. Avoid secondhand exposure to tobacco smoke.
* Avoid lung irritants. Use safety equipment (like face masks) if you work with chemicals or dust.
* Avoid alcohol use. You should limit or completely avoid alcohol use if you have certain types of Alpha-1 that can cause liver damage. You shouldn’t drink alcohol if you have liver damage.
* Ask your provider before taking medications or supplements that can affect your liver, like acetaminophen (Tylenol®). Read labels on prescription and over-the-counter medications, vitamins and supplements for warnings about liver damage.
* Get vaccinated against infectious diseases. This includes respiratory illnesses like the flu, pneumonia and COVID-19 and liver infections like hepatitis A and B. Wash your hands and take other precautions to avoid getting sick with illnesses that can cause lung inflammation.
* If a family member has Alpha-1, talk to your provider about getting tested. Having a family member with Alpha-1 increases your risk of having it or being a carrier.
* If you have Alpha-1 or are a carrier and want to have children, you may want to speak with a genetic counselor. They can help you understand the risk of passing genetic changes to your child.

## **Living With**

The best way to take care of yourself with an Alpha-1 diagnosis is to avoid things that can damage your lungs or liver. This includes smoking, lung irritants, alcohol and certain medications. Follow your provider’s recommendations for other ways to stay healthy and manage your symptoms.

### **When to see a doctor**

See your provider if you have symptoms of Alpha-1 or if a family member has Alpha-1. Early diagnosis is important, so if you have COPD or asthma, ask your provider if you should get an Alpha-1 test.

If you’ve been diagnosed with Alpha-1, see your provider if you have any new symptoms or questions about your care, or if you’re having trouble managing your symptoms.

## **PREDEFINED Q AND A**

## Based on my child’s symptoms, should I get tested for Alpha-1?

If your child has unexplained lung problems (such as wheezing, chronic cough, or difficulty breathing), liver issues (like jaundice or elevated liver enzymes), or a family history of Alpha-1 or early lung/liver disease, testing is recommended. A simple blood test measuring alpha-1 antitrypsin levels or genetic testing can confirm the diagnosis.

## 2. How can I protect my child’s lungs and liver from damage?

* Avoid exposure to tobacco smoke, air pollution, and respiratory infections as these increase lung damage risk.
* Ensure your child receives all recommended vaccinations, including flu and pneumococcal vaccines, to prevent lung infections.
* Maintain a healthy diet and regular check-ups to monitor liver health.
* Avoid unnecessary medications or substances that can stress the liver.

## 3. What foods or medications should I avoid?

* There are no specific foods to avoid, but a balanced, nutritious diet supports liver health.
* Avoid medications that can harm the liver unless prescribed and monitored by your doctor (e.g., certain antibiotics, acetaminophen in high doses). Always consult your healthcare provider before giving new medications.

## 4. Should my family members be tested for Alpha-1?

Yes. Since Alpha-1 is a genetic condition, family members—especially siblings and parents—should be offered testing to identify if they carry the gene or have the deficiency. Early diagnosis helps with monitoring and prevention of complications.

## 5. What other tests might my child need?

* Lung function tests to assess breathing capacity.
* Liver function tests and imaging (ultrasound) to monitor liver health.
* In some cases, genetic testing to identify specific mutations.
* Regular clinical evaluations to track symptoms and organ function.

## 6. What’s the prognosis?

* Prognosis varies. Many children with Alpha-1 live normal lives with proper care.
* Lung disease risk increases with exposure to irritants.
* Liver disease can range from mild to severe; some children may develop liver fibrosis or cirrhosis over time.
* Early diagnosis and preventive care improve outcomes significantly.

## 7. How do I manage my child’s symptoms at home?

* Follow your doctor’s advice on medications and therapies.
* Avoid smoke and pollutants.
* Encourage physical activity appropriate for your child’s ability.
* Monitor for respiratory symptoms like cough or shortness of breath.
* Keep vaccinations up to date.

## 8. What new or worsening symptoms should I look out for?

* Increased coughing, wheezing, or difficulty breathing.
* Yellowing of the skin or eyes (jaundice).
* Unexplained fatigue, poor appetite, or abdominal swelling.
* Frequent lung infections or prolonged recovery from colds.

## 9. When should I follow up with or contact my doctor?

* Regular follow-ups as recommended (usually every 6–12 months).
* Contact your doctor if your child develops new respiratory symptoms, jaundice, or signs of infection.
* If symptoms worsen or you have concerns about medication side effects.

## 10. When should I go to the Emergency Room (ER)?

* If your child has severe difficulty breathing, persistent chest pain, high fever, confusion, or bluish lips/face, seek emergency care immediately.
* Also go to the ER for sudden worsening of liver-related symptoms like severe abdominal pain or swelling.

## **Epidemiology**

### Frequency

*United States*

Alpha1-antitrypsin deficiency (AATD) is 1 of the 3 most common lethal genetic diseases among adult white persons, affecting 1 per 3000-5000 individuals. Severe AATD affects an estimated 70,000-100,000 individuals, and approximately 25 million people carry of at least 1 deficient gene. However, less than 10% of severely deficient individuals are currently identified.

*International*

AATD has been identified in all populations, but it is most common in individuals of Northern European (1 in 1600) and Iberian descent. Similar rates are found among white persons worldwide, with an estimated 117 million carriers and 3.4 million affected individuals.

### Race

White persons constitute an estimated 117 million carriers and 3.4 million affected individuals. Racial groups other than whites are affected less frequently.

### Sex

Women and men are affected in equal numbers.

### Age

The enzyme deficiency is congenital and has a bimodal distribution with respect to symptoms. It can be seen in neonates as a cause of neonatal jaundice and hepatitis. It can present in infants as cholestatic jaundice and in children as hepatic cirrhosis or liver failure. AATD is also the leading underlying condition requiring liver transplantation in children.

In adults, AATD leads to chronic liver disease in the fifth decade of life. As a cause of emphysema, it is seen in nonsmokers most commonly in the fifth decade of life and during the fourth decade of life in smokers.

## **Differential Diagnoses**

* Autoimmune Hepatitis
* Bronchiectasis
* Bronchitis
* Chronic Obstructive Pulmonary Disease (COPD)
* Cystic Fibrosis
* Emphysema
* Primary Ciliary Dyskinesia (Kartagener Syndrome)
* Viral Hepatitis

## **Staging**

No specific grading system exists for alpha1-antitrypsin deficiency (AATD), but the severity of the emphysema that it creates can be staged using the body mass index, airflow obstruction, dyspnea, and exercise capacity (BODE) index.This 4-step evaluation of patients with chronic obstructive lung disease appears to identify a population with limited survival who might benefit from intensified therapy. The index has not been evaluated in a population of individuals with AATD.

**Recommendation**

### Screening and Diagnostic Testing

The GOLD guidelines recommend all patients diagnosed with COPD be screened for AATD. Family members should also be screened. The patient and family members should be referred to specialist centers.

The Alpha-1 Foundation guidelines recommend the following individuals be tested for AATD :

* All patients with COPD regardless of age or ethnicity
* All patients with unexplained chronic liver disease
* All patients with necrotizing panniculitis, granulomatosis with polyangiitis, or unexplained bronchiectasis
* Parents, siblings, and children, as well as extended family of individuals identified with an abnormal gene for AAT, should be provided genetic counseling and offered testing for AATD; AAT level testing alone is not recommended because disease risk from AATD is not fully denoted.

Testing of symptomatic patients should include *SERPINA1* mutations genotyping for at least the S and Z alleles. Advanced or confirmatory testing should include Pi-typing, AAT level testing, and/or expanded genotyping.

The initial evaluation of patients diagnosed with AATD should include the following:

* Complete lung function testing
* Baseline CT scan of the chest in symptomatic patients or those with abnormal pulmonary function testing results

At a minimum, annual follow-up should include a spirometry test and monitoring for liver disease with a focused physical exam for signs of liver disease, liver ultrasound, and laboratory monitoring of AST, ALT, GGT, albumin, bilirubin, INR, and platelets. Repeated chest CT scanning to monitor progression of disease is not recommended.

Testing for AATD should be considered in all patients with chronic airflow obstruction, asthma with persistent airflow limitation, emphysema disproportionate to the smoking history or in the presence of liver or skin disease. Initial testing should include spirometry, lung volumes, gas transfer and CT chest scan.

**Management of pulmonary disease**

Intravenous augmentation therapy is recommended by the Alpha-1 Foundation guidelines for patients with necrotizing panniculitis or an FEV1 ≤ 65% predicted. Intravenous augmentation therapy is not recommended for the following:

* Individuals with the MZ genotype
* Individuals who continue to smoke
* Individuals with emphysema or bronchiectasis who do not have airflow obstruction.
* The treatment of liver disease
* Individuals who have undergone liver transplantation.

Additionally, weekly doses higher than the current FDA-approved dose are monitoring of trough AAT blood levels to evaluate the adequacy of AAT augmentation dosing are not recommended. Lung volume reduction surgery is also not recommended for the treatment of AATD.

The GOLD guidelines note that ex- or nonsmokers with FEV1 between 35% and 60% are the most likely to benefit from intravenous augmentation therapy. consideration of intravenous augmentation therapy in non‐smoking patients with AATD

**Genomic Data on Alpha-1 Antitrypsin Deficiency (AATD):**

* Gene Involved:  
  Alpha-1 antitrypsin deficiency is caused by mutations in the SERPINA1 gene, located on chromosome 14q32. This gene encodes the alpha-1 antitrypsin (AAT) protein, which protects tissues from enzymes like neutrophil elastase.
* Inheritance Pattern:  
  AATD is inherited in an autosomal codominant manner, meaning both gene copies (alleles) contribute to the trait. Individuals can have different combinations of normal and mutant alleles, influencing disease severity.
* Common Alleles and Mutations:
  + M allele: The normal allele producing normal levels of AAT.
  + Z allele (p.Glu342Lys): The most common pathogenic variant causing severe deficiency; leads to abnormal protein folding, polymerization, and retention in liver cells, reducing circulating AAT to about 10–15% of normal. Homozygous ZZ individuals are at high risk for lung and liver disease.
  + S allele (p.Glu264Val): Causes moderate deficiency with plasma levels about 50–60% of normal; individuals with SZ genotype have intermediate risk.
  + Over 500 rare SERPINA1 variants have been identified, including null alleles (no protein production) and dysfunctional alleles affecting protein activity or secretion.

**DOCTOR PATIENT CONVERSATION**

Doctor: Hello! I want to talk with you about your child’s diagnosis of Alpha-1 Antitrypsin Deficiency, or Alpha-1 for short. This is a genetic condition that can affect the lungs and liver.

Parent: What does this mean for my child? How serious is it?

Doctor: Alpha-1 means your child’s body doesn’t make enough of a protein called alpha-1 antitrypsin, which normally protects the lungs and liver from damage. Some children develop liver problems early on, like jaundice or poor growth, while lung symptoms usually appear later. With careful monitoring and care, many children do well.

Parent: How do we protect my child’s lungs and liver?

Doctor: It’s very important to avoid exposure to smoke, secondhand smoke, and other lung irritants. Keeping up with vaccinations, especially for flu and pneumonia, helps prevent infections. For the liver, we monitor function regularly and may recommend special diets or vitamins if needed. Your child’s growth and development will be closely followed.

Parent: Are there special foods or medicines to avoid?

Doctor: There’s no specific food to avoid, but children with liver involvement may need special formulas or extra calories to help with growth. We also avoid medicines that can stress the liver, like ibuprofen or aspirin, unless your doctor says it’s safe. Always check before giving any new medicine.

Parent: Should other family members be tested?

Doctor: Yes, since Alpha-1 is inherited, testing parents and siblings can help identify who else might have the condition. Early diagnosis helps with monitoring and prevention.

Parent: What tests will my child need?

Doctor: We’ll do blood tests to check liver function and alpha-1 levels, ultrasound scans of the liver, and lung function tests as your child grows. Genetic testing confirms the diagnosis and helps guide care.

Parent: What symptoms should I watch for at home?

Doctor: Look for yellowing of the skin or eyes, poor weight gain, tiredness, swelling of the belly, or breathing problems like cough or wheezing. If you notice these, please contact us.

Parent: When should we come back for check-ups?

Doctor: We usually see children every few months at first, then yearly if stable. But if you notice any new or worsening symptoms, call us sooner.

Parent: When should we go to the emergency room?

Doctor: If your child has severe difficulty breathing, persistent vomiting of blood, black or bloody stools, or becomes very lethargic or confused, go to the ER immediately.

Parent: Thank you for explaining everything.

Doctor: You’re welcome. We’re here to support your family and answer any questions anytime.

REFERENCES

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**ALVEOLAR CAPILLARY DYSPLASIA**

**DEFINITION AND DESCRIPTION**

Alveolar capillary dysplasia is a rare lung disorder that affects newborns. It is characterized by abnormal development of the blood vessels in the lungs, specifically the small blood vessels surrounding the air sacs called alveoli. This condition hinders the proper exchange of oxygen and carbon dioxide in the lungs, leading to respiratory issues.

The exact cause of alveolar capillary dysplasia is not fully understood, but it is believed to be a result of abnormal development during fetal growth. Genetic factors may also play a role in some cases. Understanding this condition is crucial for early detection and management, as it can significantly impact a newborn's breathing and overall health.

## **Symptoms of Alveolar Capillary Dysplasia**

Alveolar capillary dysplasia is a rare lung condition that can present with symptoms such as difficulty breathing, rapid breathing, and low oxygen levels in newborns. Babies may also have feeding problems, poor weight gain, and blue discoloration of the skin due to inadequate oxygenation.

These symptoms typically manifest soon after birth and can be severe, requiring immediate medical attention. If your baby is showing these signs, it is crucial to seek prompt evaluation and care from a healthcare provider familiar with this condition to ensure proper management and treatment.

Infants with alveolar capillary dysplasia may experience severe respiratory distress shortly after birth.

Difficulty feeding and poor weight gain are common symptoms in infants with alveolar capillary dysplasia.

Persistent pulmonary hypertension can occur in individuals with alveolar capillary dysplasia.

Failure to thrive, despite appropriate nutritional intake, is often observed in patients with alveolar capillary dysplasia.

Recurrent respiratory infections are a hallmark symptom of alveolar capillary dysplasia in affected individuals.

## **Causes of Alveolar Capillary Dysplasia**

The exact cause of this condition is not fully understood, but it is believed to involve genetic factors. Mutations in certain genes have been associated with alveolar capillary dysplasia, impacting the formation of the tiny blood vessels in the alveoli.

These abnormalities can lead to severe respiratory difficulties in affected individuals, often presenting in infancy. Further research is needed to fully elucidate the complex genetic mechanisms underlying this condition.

Genetic mutations affecting the FOXF1 gene Abnormal development of lung blood vessels during fetal growth Environmental factors disrupting normal vascular formation Inherited genetic predisposition from parents Disruption of signaling pathways crucial for lung vascular development

## **Types of Alveolar Capillary Dysplasia**

Alveolar capillary dysplasia (ACD) encompasses different types that affect the development of the lung's blood vessels. While specific subtypes may not be clearly defined, variations in the severity and presentation of ACD have been observed. These may include forms with distinct genetic underpinnings or variations in clinical manifestations.

Researchers continue to investigate the complexities of ACD to improve diagnosis and treatment strategies. Understanding the nuances within the spectrum of ACD is crucial for providing tailored care to individuals affected by this rare lung disorder.

**Classic type:** Characterized by severe alveolar capillary dysplasia with misalignment of the pulmonary veins.

**Variants:** Includes milder forms such as hypoplasia, absence of pulmonary veins, or a mixed form.

**Associated conditions:** Often linked to congenital heart defects like atrial septal defects or ventricular septal defects.

**Genetic factors:** Some cases are associated with mutations in the FOXF1 gene.

**Prognosis:** Generally poor, with most cases leading to severe respiratory failure in infancy.

**Treatment:** Lung transplantation is the only curative option for severe cases.

## **Risk Factors**

While the exact cause is not fully understood, several risk factors have been identified. These include genetic mutations, particularly in the FOXF1 gene, which plays a crucial role in lung development. Maternal conditions such as diabetes, hypertension, and substance abuse during pregnancy have also been linked to an increased risk of alveolar capillary dysplasia.

Additionally, certain environmental factors and exposures may contribute to the development of this condition. Early recognition of these risk factors is essential for prompt diagnosis and management of alveolar capillary dysplasia.

Genetic mutations, especially in the FOXF1 gene, are a significant risk factor for alveolar capillary dysplasia.

Maternal diabetes during pregnancy has been associated with an increased likelihood of a child developing alveolar capillary dysplasia.

Exposure to certain environmental toxins or medications while in the womb can raise the risk of alveolar capillary dysplasia.

Family history of alveolar capillary dysplasia or other lung disorders can predispose individuals to the condition.

Premature birth and low birth weight are risk factors that may contribute to the development of alveolar capillary dysplasia.

## **Diagnosis of Alveolar Capillary Dysplasia**

Clinicians may first suspect ACD based on symptoms like severe respiratory distress in newborns. Imaging studies can reveal characteristic findings in the lungs that suggest ACD. Genetic testing plays a crucial role in confirming the diagnosis by identifying specific genetic mutations associated with ACD.

Additionally, a lung biopsy may be performed to further confirm the presence of abnormal lung architecture typical of ACD. This comprehensive approach helps healthcare providers accurately diagnose ACD and develop appropriate management strategies.

Diagnosis of alveolar capillary dysplasia typically involves genetic testing to identify mutations associated with the condition.

Imaging studies such as chest X-rays and CT scans can reveal characteristic findings suggestive of alveolar capillary dysplasia.

Lung biopsy may be performed to confirm the diagnosis by examining the lung tissue for specific abnormalities.

Echocardiography is often done to assess the heart's structure and function, as heart defects are commonly associated with this condition.

## **Treatment for Alveolar Capillary Dysplasia**

Treatment options for alveolar capillary dysplasia are limited and challenging due to the complex nature of the condition. Currently, there is no definitive cure for this condition, and management focuses on supportive care to address symptoms and complications. This may involve ventilator support, supplemental oxygen therapy, and medications to manage pulmonary hypertension.

In some cases, lung transplantation may be considered as a last resort for patients with severe and progressive respiratory failure. Close monitoring and multidisciplinary care are essential in the management of alveolar capillary dysplasia to optimize outcomes and provide the best possible quality of life for affected individuals.

**Differential diagnosis (DDx) list for Alveolar Capillary Dysplasia (ACD)**

1. Idiopathic Persistent Pulmonary Hypertension of the Newborn (PPHN)
   1. Usually reversible with pulmonary vasodilators and respiratory support, unlike ACD which is refractory.
2. Sepsis, Pneumonia, and Other Infectious Etiologies
   1. Infectious causes of respiratory failure with systemic signs and treatable with antibiotics.
3. Primary Respiratory Disorders:
   1. Surfactant Protein Deficiencies (e.g., surfactant protein B deficiency)
   2. Hyaline Membrane Disease (Neonatal Respiratory Distress Syndrome)
   3. Pulmonary Hypoplasia (often secondary to congenital diaphragmatic hernia or oligohydramnios)
   4. Congenital Diaphragmatic Hernia (CDH)
   5. Rare Diffuse Interstitial Lung Disorders (e.g., acinar dysplasia, congenital alveolar dysplasia)
4. Congenital Cardiopulmonary Diseases:
   1. Pulmonary venous stenosis
   2. Total anomalous pulmonary venous return (TAPVR)
   3. Other cardiac defects causing cyanosis and pulmonary hypertension
5. Neurologic Disorders:
   1. Perinatal asphyxia
   2. Congenital neuromuscular disorders causing respiratory failure without primary lung pathology
6. Pulmonary Arteriopathy Secondary to Chronic Lung or Cardiac Disease
   1. Mimics vascular abnormalities seen in ACD but with different clinical course
7. Primary and Secondary Pulmonary Lymphangiectasia
   1. Dilated lymphatic vessels causing respiratory distress

**Epidemiology of Alveolar Capillary Dysplasia (ACD/MPV):**

* Incidence and Prevalence:  
  ACD with misalignment of the pulmonary veins (ACD/MPV) is a very rare, lethal congenital lung disorder. Its exact incidence and prevalence are unknown but estimated to be approximately 1 in 100,000 newborns based on limited data. Fewer than 200 cases have been formally reported worldwide, but many cases likely go undiagnosed or misclassified, especially as idiopathic persistent pulmonary hypertension of the newborn (PPHN).
* Demographics:
  + More than 90% of affected infants are born at term.
  + There is a slight male predominance (~60%) among reported cases.
  + No specific geographic or ethnic predilection has been identified; cases are reported worldwide.
* Age of Onset:
  + Clinical symptoms typically present within the first 48 hours of life, with cyanosis and respiratory failure.
  + Atypical or milder forms with delayed onset and longer survival have been described but are rare.
* Mortality:
  + The condition is almost universally fatal without lung transplantation, with most infants dying within the first few weeks to months of life.
  + Survival beyond the neonatal period is rare and may be related to less severe disruption of capillary development.
* Associated Anomalies:
  + Many infants with ACD/MPV have additional congenital malformations, especially of the gastrointestinal tract (e.g., intestinal malrotation), cardiovascular system, and genitourinary tract.

**Genomic Data on Alveolar Capillary Dysplasia with Misalignment of Pulmonary Veins (ACD/MPV):**

* Key Gene Involved:  
  The primary genetic cause of ACD/MPV is mutations or deletions affecting the FOXF1 gene located on chromosome 16q24.1. FOXF1 encodes a transcription factor critical for pulmonary vascular development.
* Types of Genetic Alterations:
  + Point mutations (single nucleotide variants) in FOXF1, often de novo (not inherited), are common causes.
  + Genomic deletions involving FOXF1 or its lung-specific enhancer regions upstream of the gene also cause the disease.
  + Mutations can be maternally inherited due to genomic imprinting effects, where the maternal allele is primarily expressed.
  + Some cases involve deletions of the FOXF1 gene cluster including neighboring genes like FENDRR.
* Molecular Impact:  
  Mutations result in loss of function or haploinsufficiency of FOXF1, disrupting the transcriptional regulation of genes essential for alveolar and vascular development in the lung. This leads to abnormal formation and misalignment of pulmonary veins and deficient alveolar capillary networks.
* Genomic Imprinting:  
  Studies show parent-of-origin effects, with the maternal allele of FOXF1 being predominantly expressed in lung and intestinal tissues, explaining why mutations on the maternal allele cause disease.
* Associated Malformations:  
  Besides pulmonary defects, FOXF1 mutations can cause gastrointestinal malformations such as intestinal malrotation and annular pancreas, consistent with its role in mesenchymal development.
* Recent Research:
  + Novel FOXF1 upstream enhancers have been identified that regulate gene expression in endothelial and mesenchymal lung cells; deletions here also cause ACD/MPV.
  + Mouse models with FOXF1 mutations recapitulate human disease features and show disrupted signaling pathways (e.g., STAT3).

**PREDEFINED Q AND A**

## 1. What is Alveolar Capillary Dysplasia with Misalignment of Pulmonary Veins (ACD/MPV)?

ACD/MPV is a rare congenital lung disorder where the tiny blood vessels (capillaries) in the lungs fail to develop properly and are abnormally positioned. This disrupts oxygen exchange and causes severe pulmonary hypertension and respiratory failure shortly after birth.

## 2. What causes ACD/MPV?

The condition is caused by abnormal development of lung blood vessels, often linked to mutations or deletions in the FOXF1 gene on chromosome 16q24.1. Most cases occur sporadically, but some familial cases suggest a genetic basis.

## 3. What are the symptoms of ACD/MPV?

Symptoms typically appear within minutes to hours after birth and include:

* Severe respiratory distress
* Cyanosis (bluish skin and lips due to low oxygen)
* Persistent pulmonary hypertension that does not respond to usual treatments
* Feeding difficulties and poor growth may also occur

## 4. How is ACD/MPV diagnosed?

Diagnosis is challenging and often requires:

* Clinical suspicion in newborns with severe hypoxemia and pulmonary hypertension unresponsive to therapy
* Lung biopsy showing characteristic vascular abnormalities
* Genetic testing for FOXF1 mutations or deletions
* Imaging and other tests help exclude other causes

## 5. Is there a treatment for ACD/MPV?

There is currently no cure for ACD/MPV. Treatment is supportive and includes respiratory support and management of pulmonary hypertension. Lung transplantation may be considered but is rarely feasible due to rapid disease progression.

## 6. What is the prognosis for infants with ACD/MPV?

The prognosis is poor; most infants do not survive beyond the first few weeks or months of life without lung transplantation. Some rare cases with milder or patchy disease have survived longer.

## 7. Are there associated abnormalities with ACD/MPV?

Yes, many infants have other congenital anomalies, including gastrointestinal malformations (e.g., intestinal malrotation), cardiovascular defects, and genitourinary abnormalities.

## 8. Can ACD/MPV be prevented?

Currently, there is no known way to prevent ACD/MPV due to its genetic and developmental nature.

## 9. Is genetic counseling recommended?

Yes, genetic counseling is advised for families affected by ACD/MPV, especially if a FOXF1 mutation or familial cases are identified.

## 10. Where can I find support or more information?

Support groups such as the Alveolar Capillary Dysplasia Association and resources from rare disease organizations can provide assistance and information.

**DOCTOR PATIENT CONVERSATION**

Doctor: Hello. I want to discuss your baby’s diagnosis. Your child has a rare lung condition called Alveolar Capillary Dysplasia with Misalignment of Pulmonary Veins, or ACD/MPV.

Parent: What exactly is that? It sounds serious.

Doctor: It is a very rare and serious condition where the small blood vessels in the lungs don’t develop properly. This causes very high blood pressure in the lungs and makes it hard for oxygen to get into the blood. Babies with ACD usually have trouble breathing soon after birth.

Parent: How did this happen? Is it something we did?

Doctor: This is a congenital condition, meaning it happens during lung development before birth. In most cases, it’s caused by changes in a gene called *FOXF1*. It’s not caused by anything you did during pregnancy.

Parent: How do you diagnose this?

Doctor: The diagnosis is often suspected when a newborn has severe breathing problems and high lung blood pressure that doesn’t get better with usual treatments. We can confirm it by looking at lung tissue under a microscope, usually from a biopsy or sometimes after unfortunately the baby passes away. Genetic testing for *FOXF1* mutations can also support the diagnosis without invasive procedures.

Parent: Is there any treatment?

Doctor: Currently, there is no cure. Treatments like oxygen, ventilators, and medications to lower lung pressure can help temporarily, but the condition usually progresses quickly. For some babies with milder forms, these treatments may work longer. The only definitive treatment is a lung transplant, but that’s very complex and not always possible.

Parent: What does this mean for my baby’s future?

Doctor: The condition is very serious and often fatal in the newborn period. However, there are rare cases where babies survive longer, especially if the disease is less severe. We will support your baby with the best care possible and discuss all options with you.

Parent: Are there other problems my baby might have?

Doctor: Sometimes babies with ACD also have other congenital problems, like issues with their intestines or heart. We will check carefully for these.

Parent: What should I watch for?

Doctor: Watch for worsening breathing difficulty, blue or pale skin, or poor feeding. If your baby has trouble breathing or becomes very tired, seek medical help immediately.

Parent: Is there any way to prevent this in future pregnancies?

Doctor: Since this is usually caused by a new genetic mutation, the risk of recurrence is low, but genetic counseling can help assess your family’s specific risk.

Parent: Thank you for explaining. It’s a lot to take in.

Doctor: I understand. We are here to support you and your family every step of the way. Please ask any questions anytime.

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<https://child-foundation.org/what-is-child/child-disorders/alveolar-capillary-dysplasia/>

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### **Asbestosis**

**DEFINITION AND DESCRIPTION**

Asbestosis is a lung disease that occurs in people who inhale asbestos fibers and dust over a long period of time. Asbestos is a mineral that forms tiny and long-lasting fibers when it’s in the air. People in certain industries and occupations like construction, manufacturing, mining, milling, mechanics and electricians (especially those who worked before the 1970s) are most at risk for asbestosis. People who remove asbestos products are also at risk, as are people who served in the Navy on ships where asbestos was in use.

When asbestos fibers and dust get into your lungs, they can cause fibrosis (thickening and scarring of your lungs). Asbestos can also cause the membranes surrounding your lungs (pleura) to thicken. This scarring and thickening of your lung tissue can make breathing difficult.

In some cases, asbestosis can lead to life-threatening health complications. In severe cases, asbestosis can be fatal. Asbestos exposure can also increase your risk of lung cancer.

Asbestos is a group of six natural mineral fibers. These fibers are known for their strength and fire- and chemical-resistant properties. Because of these qualities, the manufacturing and building industries use asbestos to:

* Strengthen cement and plastics.
* Provide insulation.
* Fireproof buildings, textiles and military vehicles.
* Absorb sound.

Asbestos fibers may be white, blue, brown, gray or green. White asbestos fibers (called chrysotile) are the most widely used worldwide.

Everyone is exposed to asbestos at some point in their life. Low levels of asbestos are virtually everywhere. Most people don’t get an asbestos-related disease. People who get an asbestos-related disease tend to have had exposure on a regular basis and over a long period of time.

The risk of asbestos exposure on the job was highest before the 1970s, though asbestos exposure can still occur today. Because it takes a long time to develop symptoms, providers are still diagnosing many new cases of asbestosis. Regulation of on-the-job exposure to asbestos has reduced the risk of asbestosis, but even with these regulations, workers can be exposed by accident.

According to the World Health Organization (WHO), millions of people face exposure to asbestos, mostly in the workplace. It’s hard to estimate how many people are living with asbestosis and other asbestos-related diseases because signs of the disease may not show up for 30 or 40 years. Some studies show that up to 20% of all workers who breathe in asbestos will develop a disease from exposure to asbestos. Not all workers who develop asbestos related disease will have asbestosis. There are other diseases that asbestos can cause, including:

* Interstitial lung disease (ILD).
* Pleural effusion.
* Certain kinds of lung cancer, including pleural mesothelioma.

## **Symptoms and Causes**

Symptoms of asbestosis vary depending on the severity of the disease and may not appear for 20 to 30 years or more after asbestos exposure. The scarring on your lungs from asbestosis typically gets worse slowly. Because of that, you may not notice your symptoms right away.

The first symptom of asbestosis is having trouble breathing (being short of breath), especially when you’re working hard or exercising. Other signs and symptoms may include:

* Chest pain and tightness.
* Clubbing of your nails.
* Cough.
* Crackling sound when inhaling.
* Fatigue (extreme tiredness) and trouble exercising.
* Unexplained weight loss.

### **CAUSES**

The main cause of asbestosis is inhaling tiny asbestos fibers or dust. Asbestos fibers are not harmful unless they release into the air. When they are released, the fibers break down into tiny particles. The particles become airborne, and we inhale them. Then they collect in the lungs, causing scarring and inflammation. Scarred lung tissue is stiff and unable to expand, which makes it hard to breathe.

The longer you had exposure to asbestos and the more intense the exposure, the higher your chances of developing asbestosis.

Building and manufacturing industries used asbestos widely in the past. People who work in these industries and repeatedly inhale asbestos particles are the most at risk for developing asbestosis.

Breathing in asbestos particles in the air causes asbestosis. Undisturbed asbestos — such as in insulation or tile — doesn’t increase your risk of the disease.

Materials and products that may contain asbestos include:

* Car clutch pads and brake linings.
* Construction cement, putties and plaster.
* Insulation.
* Pipe wrapping.
* Siding and roof shingles.
* Vinyl floor tiles.
* Casings for electrical wires.
* Millboard.
* Patching and joint compound.
* Floor tile and adhesives.
* Soundproofing material.

Many household products and substances also contain asbestos, including:

* Artificial ashes and embers used in gas-fired fireplaces.
* Automobile brake pads and linings, clutch facings and gaskets.
* Fireproof gloves, stovetop pads, table pads and fire-resistant fabrics (including blankets and curtains).
* Some plastics, paints, coatings and adhesives.
* Vermiculite-containing attic insulation and consumer garden products.

### **How long can it take for asbestosis to develop?**

Healthcare providers sometimes diagnose asbestosis in people who haven’t worked or been around asbestos for decades. It can take up to 30 years to develop symptoms.

### **Risk factors for asbestosis**

There are regulations in place now that reduce your asbestos exposure while on the job. Still, certain occupations face asbestos exposure, particularly if you demolish or renovate buildings built before the 1970s. Homes built before 1977 may also contain asbestos in materials like pipes, ceilings and floor tiles. Just because you live in a home built before 1977 doesn’t mean you’re at risk. Risk only occurs when asbestos is in an inhalable form like dust. When it’s in a solid state, you’re not at risk.

You’re more at risk of getting asbestosis if you have long-term exposure to asbestos. This is true if your job involves handling materials containing asbestos. These types of jobs include:

* Asbestos miners, installers or removers.
* Auto and aircraft mechanics.
* Construction crews.
* Electrical workers.
* Railroad and shipyard workers.

Studies also show that people who were involved in the rescue and cleanup at the site of the attacks on the World Trade Center (WTC) in New York City are at risk for asbestos-related diseases like asbestosis.

##### **Factors that affect your risk of developing asbestosis**

The following factors play a role in your risk:

* **Duration:** How long you were exposed to asbestos. In general, the longer your exposure, the higher your risk.
* **Intensity:** How much asbestos you were exposed to.
* **Type of industry:** Your risk is lower if asbestos is bonded into a product (such as walls or tiles). Your risk is higher if asbestos is released into the air, such as during sawing or demolition.
* **Personal risk factors:** Smoking or preexisting lung disease.
* **Genetics:** Having a genetic mutation to the *BAP1* gene**.**

People with the disease tend to have had exposure for many years through an occupation. You’re not likely to get asbestosis if you disrupt asbestos during a home renovation, for example.

### **Complications that may be associated with asbestosis**

Many people who have asbestosis have breathing trouble and a cough that doesn’t go away. In more severe cases, complications can be life-threatening.

Complications of asbestosis may include:

* **Lung cancer:** People who have asbestosis and smoke cigarettes have an even higher risk of lung cancer.
* **Mesothelioma:** Cancer that forms in the lining of your abdomen, chest or lungs.
* **Respiratory failure:** Your lungs can’t put enough oxygen into your blood, and carbon dioxide builds up in your tissues.
* **Right-sided heart failure:** The right side of your heart stops working correctly.

## **Diagnosis and Tests**

Your healthcare provider will examine you and ask about your medical history. Remember to tell them about your exposure to any harmful substances like asbestos.

Your healthcare provider may also order tests to complete the diagnosis. These might include:

* A chest X-ray.
* A computed tomography (CT) scan.
* Lung function tests like spirometry.
* Bronchoscopy.

## **Management and Treatment**

Treating asbestosis aims to manage symptoms and preserve function in your lungs.

Treatment can’t reverse lung damage from asbestos. Treatment for asbestos-related diseases aims to relieve symptoms, treat complications related to the disease and slow its progress.

Your treatment depends on the severity of the disease. Your options might include:

* **Oxygen therapy:** Receiving extra oxygen through a mask or tube in your nostrils helps you breathe more comfortably.
* **Pulmonary rehabilitation:** Exercises and behavioral changes can improve your quality of life.
* **Lung transplant surgery:** In rare cases, a new, healthy lung from a lung transplant can relieve symptoms and prolong life.
* **Medication:** Medicines called “anti-fibrotics” can slow down the rate at which scarring gets worse but can’t heal existing scarring. Your provider can discuss the risks and benefits of these medicines and help you decide what’s best for your health.

### **What can I do at home to manage the symptoms of asbestosis?**

To make things easier on yourself if you have asbestosis, you can follow a healthy lifestyle by:

* Not smoking. If you do smoke, get help quitting. Smoking speeds up the progression of the disease and makes it worse.
* Avoiding breathing air that's contaminated with allergens, particles, chemicals or secondhand smoke.
* Avoiding sick people and practicing good handwashing hygiene.
* Drinking lots of water.
* Eating nutritious foods.
* Exercising regularly after discussing an exercise plan or routine with your healthcare provider.

## **Outlook / Prognosis**

There isn’t a cure for asbestosis, and you can’t reverse the damage from the disease. Once you breathe in asbestos fibers, they stay in your body. Your prognosis varies depending on how long and how much exposure you had to the particles.

Many people with mild asbestosis live fulfilling lives for many years after being diagnosed. Others get worse and need medical treatment for the rest of their lives.

Your healthcare provider is likely to order chest X-rays and lung function tests every few years to look for changes in the scarring in your lungs. The findings on your imaging tests will change as the stages of your condition progress.

The average life expectancy is about 10 years once you receive a diagnosis. It depends on how severe the disease is and how fast it progresses.

## **Prevention**

You can reduce your risk of asbestosis by avoiding long-term exposure to asbestos. If your job involves exposure to the mineral, you should wear a respirator (a mask that filters particles from the air). This protective mask keeps you from inhaling asbestos fibers or dust.

If you know you’ve had asbestos exposure, you should have regular exams and chest X-rays. These tests don’t prevent asbestosis but can help catch it early.

If you smoke and have exposure to asbestos, quitting smoking is your best way to reduce your risk of getting cancer.

#### **Should I avoid all asbestos products?**

Asbestos fibers are only harmful when they get in the air. Today, building materials and many other products use bonded asbestos. This process keeps them from being released into the air. There’s little to no risk of harmful health effects from these products. However, take care not to sand, tear or otherwise damage or crumble the material. This can release the fibers into the air.

#### **Do I need to remove asbestos materials from my home?**

If you have asbestos materials in your home that are in good condition, it’s best to leave them alone. If you touch or disturb the material, you risk releasing the fibers into the air. Have these materials inspected from time to time for signs of damage or deterioration.

**When to see a doctor**

Contact your healthcare provider if you’ve had exposure to asbestos and have trouble breathing, chest discomfort or a cough that doesn’t go away. Be sure to tell your provider about your asbestos exposure, even if it’s secondhand.

**PREDEFINED Q AND A**

#### **If I was exposed to asbestos through my job, does that increase health risks for my family members?**

It’s possible to have “secondhand” asbestos exposure. When a person works with asbestos materials, they can bring home particles on their shoes, clothing, skin and hair.

To decrease this risk, most jobs that use asbestos materials make sure that the workers change when they arrive and leave work. Most companies also have showers available for employees to clean particles from hair and skin. These precautions lower the risk of family members developing any diseases.

### **What is the difference between asbestosis and mesothelioma?**

The main difference between asbestosis and mesothelioma is that mesothelioma is cancer and asbestosis isn’t cancer. The disease of asbestosis remains in your lungs and pleura (the covering of your lungs). Mesothelioma begins in the tissue of your lungs and abdomen. It can spread throughout your body.

## **Diagnostic Considerations**

Determining the cause of asbestosis depends on the clinician's assessment of the levels and duration of exposure and on knowledge of occupational epidemiologic studies. Assessment of impairment, which is a key ingredient in determining disability, is based mainly on pulmonary function studies. No evidence exists to confirm that small-airway disease, which is detected by flow volume curves, progresses to asbestosis.

Clinicians should be aware of the variety of diseases that may coexist with asbestosis. Additionally, clinicians should keep in mind that the risk for bronchogenic carcinoma is increased with asbestos exposure and load, even without asbestosis.

Conditions to consider in the differential diagnosis of asbestosis include collagen-vascular diseases and other interstitial pulmonary disorders.

## **Differential Diagnoses**

* Coal Workers' Pneumoconiosis (Black Lung Disease)
* Dermatomyositis
* Hypersensitivity Pneumonitis
* Idiopathic Pulmonary Fibrosis (IPF)
* Sarcoidosis
* Silicosis

## **Epidemiology**

In 2014, the World Health Organization (WHO) estimated that 125 million people worldwide are exposed to asbestos in the workplace, that more than 100,000 people die each year from asbestos-related lung cancer, mesothelioma, and asbestosis, and that nearly 400 deaths are attributable to nonoccupational exposure to asbestos.

According to mortality data from the US National Center for Health Statistics (NCHS), 6290 deaths were attributed to asbestosis in the period 1999-2010, of which the majority (95%) were in White males (median age, 79 y). A 2024 study using data from the Global Burden of Disease (GBD) study reported that during the period 1990-2019, the overall number of deaths due to occupational exposure to asbestos increased by 20.2% in the United States, but the age-standardized mortality rate (ASMR) and the age-standardized disability-adjusted life years (DALYs) rate (ASDR) declined.

A 2024 study reported that in 2019, occupational asbestos exposure was responsible for 239,330 deaths and 4,189,000 disability-adjusted life years (DALYs) globally.Over the period 1990-2019, deaths attributed to occupational asbestos exposure increased by 65.65% globally, and DALYs increased by 43.66%.

According to World Trade Center Health Registry estimates, about 410,000 people were exposed to asbestos when as much as 400 tons of it was released following the collapse of the the Twin Towers on September 11, 2001. Those at highest risk for developing 9/11-related illnesses were workers who participated in the rescue, recovery, and cleanup efforts at the sites of the towers, along with those living and working in lower Manhattan during the cleanup.

A substantial amount of asbestos remains in buildings and eventually will be removed, either during remediation or renovations or demolition. It has been estimated that approximately 1.3 million workers in construction and general industry may be exposed to asbestos during maintenance activities or remediation of buildings containing asbestos. In the United States, vermiculite mined in Libby, Montana, was found to be contaminated with asbestos; this vermiculite was used in 70% of vermiculite insulation in the United States between 1919 and 1990. In a study of 128 Libby miners, 119 had asbestos-related findings on high-resolution computed tomography (HRCT).

Asbestos has not been mined in the United States since 2002, but in 2016, approximately 340 metric tons of asbestos was imported for use in the chloralkali industry to manufacture semipermeable diaphragms in electrolytic cells; in addition, an unknown quantity of asbestos was imported within manufactured products, possibly including brake linings and pads, building materials, gaskets, millboards, and yarn and thread, among others. In 2024, however, no asbestos was imported.

Globally, bans on asbestos use are in place in several countries, including Australia, Japan, South Africa, and the nations of the European Union; asbestos use is restricted in the United States and Canada. However, persons who have been previously exposed to asbestos continue to be at risk for asbestosis and other asbestos-related diseases as a consequence of the long latency periods following exposure. In addition, trends in developing countries and countries that are emerging as economic powers indicate an increasing problem with asbestos-related diseases

## **Procedures**

### Bronchoalveolar lavage

BAL has only limited application in the diagnosis and management of asbestosis. It is helpful in diagnosing infections that may present with diffuse infiltrates and simulate asbestosis, and it may aid in the diagnosis of a coexisting bronchogenic carcinoma. In workers who are exposed to asbestos, BAL can provide quantitative information through asbestos fiber counts. The presence of more than one asbestos body (i.e., coated asbestos fiber) per 1 mL of lavage effluent suggests significant exposure.

### Bronchoscopy

Fiberoptic bronchoscopy is performed to facilitate BAL. In addition, bronchoscopy is indicated for airway examination when findings from radiologic studies are suggestive of bronchogenic carcinoma.

Transbronchoscopic lung biopsy is not recommended for diagnosis of asbestosis. This procedure yields inadequate tissue and may cause crush alterations to the tissue.

### Open lung biopsy

Open lung biopsy is not indicated in most cases of asbestosis. However, this procedure provides sufficient tissue for the pathologist to make a definitive diagnosis.

A**sbestosis Treatment, Drug Information, and Their Side Effects**

## 1. Medications

## Corticosteroids (e.g., Prednisolone)

* Purpose: Reduce lung inflammation and suppress the immune response.
* Typical Use: May be prescribed during acute exacerbations or to reduce chronic inflammation.
* Dosage: Often starts at 0.5 to 1 mg/kg/day orally, tapered over weeks to months based on response.
* Side Effects:
  + Weight gain
  + Increased blood sugar (risk of diabetes)
  + Osteoporosis
  + Increased infection risk
  + Mood changes
  + Hypertension
  + Cataracts and glaucoma (long-term use)

## Immunosuppressive Agents (e.g., Azathioprine)

* Purpose: Sometimes used with corticosteroids to reduce inflammation.
* Side Effects:
  + Bone marrow suppression (leading to anemia, infection risk)
  + Liver toxicity
  + Nausea and vomiting
  + Increased risk of infections

## Other Agents

* Colchicine: Mild antifibrotic effects; limited evidence.
* Bronchodilators/Inhalers: For patients with coexisting COPD or airway obstruction (e.g., long-acting beta-agonists, inhaled corticosteroids).
* Antibiotics: Prompt treatment of respiratory infections.

## 2. Oxygen Therapy

* Purpose: For patients with low blood oxygen levels (hypoxemia) at rest or exertion.
* Benefits: Improves oxygenation, reduces breathlessness, and may improve survival.
* Side Effects: Generally safe; prolonged high-flow oxygen may cause dryness or nasal irritation.

## 3. Pulmonary Rehabilitation

* Structured programs including exercise training, breathing techniques, nutritional counseling, and education.
* Improves lung function, exercise tolerance, and quality of life.

## 4. Surgical Options

* Lung Transplantation: Considered in severe, end-stage disease when other treatments fail.
* Pleural Procedures: Decortication or pleurectomy may relieve symptoms related to pleural fibrosis or effusions but are not curative.

## 5. Lifestyle and Supportive Care

* Smoking Cessation: Critical to slow disease progression.
* Vaccinations: Influenza and pneumococcal vaccines to prevent infections.
* Diet: Balanced nutrition supports overall health.
* Avoidance: Minimize exposure to lung irritants and infections.

**DOCTOR PATIENT CONVERSATION**

Doctor: Hello, I want to discuss your diagnosis of asbestosis, which is a lung condition caused by exposure to asbestos fibers.

Patient: What exactly is asbestosis? How serious is it?

Doctor: Asbestosis is a chronic lung disease caused by inhaling asbestos fibers, which leads to scarring of the lung tissue. This scarring makes it harder for your lungs to expand and for oxygen to pass into your bloodstream. It can cause symptoms like shortness of breath and a persistent cough. The disease can progress over time, so it’s important we manage it carefully.

Patient: How did I get this? Is it from my job?

Doctor: Yes, asbestos exposure most commonly happens in workplaces where asbestos was used, such as construction, shipbuilding, or manufacturing. Sometimes exposure can also occur in older buildings during renovations. It’s important to avoid any further exposure.

Patient: What can be done to treat it?

Doctor: While there’s no cure to reverse the lung scarring, we focus on managing symptoms and slowing progression. This includes quitting smoking if you smoke, avoiding further asbestos exposure, getting vaccinated against flu and pneumonia, and using oxygen therapy if needed. Pulmonary rehabilitation can help improve your breathing and quality of life.

Patient: Are there medicines I can take?

Doctor: Sometimes corticosteroids or other medications are used to reduce inflammation, but their benefit in asbestosis is limited. We mainly treat complications like infections promptly. Your care will be tailored based on your symptoms and lung function.

Patient: What should I watch out for?

Doctor: Watch for worsening shortness of breath, increased coughing, chest pain, or signs of infection like fever. If you notice these, please contact us promptly.

Patient: Will I need regular check-ups?

Doctor: Yes, regular monitoring is important to track your lung function and overall health. We’ll schedule periodic visits and tests to manage your condition proactively.

Patient: Is there anything I can do to protect others around me?

Doctor: Yes, it’s important to avoid bringing asbestos fibers home on your clothes or belongings. Follow workplace safety guidelines strictly and inform family members if they might be at risk.

Patient: Thank you for explaining. It’s reassuring to know what to expect.

Doctor: You’re welcome. If you have any questions or concerns, don’t hesitate to reach out. We’re here to support you.

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## Atrial septal defect in children

**DEFINITION AND DESCRIPTION**

The atrial septum is the wall between the 2 upper chambers of the heart (right and left atria). An atrial septal defect (ASD) is an abnormal hole in this wall. ASD is a heart problem that is present at birth (congenital).

ASDs can happen on their own. Or they can happen in children born with other congenital heart defects. Girls have ASDs twice as often as boys. Doctors don't know why this is.

ASDs are classified by their different location and development:

* Secundum ASD occurs in the middle part of the atrial septum.
* Primum ASD occurs in the lower part of the atrial septum close to the tricuspid and mitral valves.
* Sinus venosis occurs in the upper part of the atrial septum near the veins that drain into the right and left atrium.
* Coronary sinus ASD occurs when there is a defect in the wall between the coronary sinus and the left atrium.

Patent foramen ovale (PFO) is an opening between the right and left atria that is normally present. It typically closes shortly after birth. But a PFO is not considered an ASD because no septal tissue is missing.

## **causes of atrial septal defect in a child**

The heart forms during the first 8 weeks of pregnancy. It starts as a hollow tube and divides into 4 chambers. These chambers are separated by walls (septa). It's normal for the walls to have openings as the fetus grows. The openings usually close shortly before or just after birth. If they don't all close, the atrial septum will have a hole in it. This is called an ASD.

Some congenital heart defects may be passed down in certain families. Most atrial septal defects occur by chance. Doctors can't find a clear reason why they happen.

## **symptoms of an atrial septal defect in a child**

Many children have no symptoms and seem healthy. If the ASD is large, your child may have symptoms. Your child may:

* Tire easily
* Have fast breathing
* Have shortness of breath
* Grow slowly
* Have respiratory infections often
* Have abnormal heart rhythm (arrhythmias)

Older children and adults with ASDs may have migraine headaches. But it's not clear if the ASD is the cause. A small blood clot that forms in the bloodstream that may cause a stroke can be linked to ASD in older children and adults. But it does not appear that closing the defect or taking blood thinners (anticoagulants) decreases risk.

The symptoms of ASD can seem like other health conditions. Have your child see his or her healthcare provider for a diagnosis.

**Diagnosis**

Your child's healthcare provider may have heard a heart murmur when listening to your child's heart with a stethoscope. The heart murmur is from the abnormal flow of blood through the heart.

Your child may need to see a pediatric cardiologist for a diagnosis. This is a doctor with special training in treating heart problems in children. The doctor will examine your child and listen to your child's heart and lungs. The doctor will find out where the murmur is best heard and how loud it is. Your child may have some tests, such as:

* **Chest X-ray.** This test may show an enlarged heart. Or it may show changes in your child's lungs because of the blood flow changes caused by an ASD.
* **Electrocardiogram (ECG).** This test records the electrical activity of the heart. It shows abnormal rhythms (arrhythmias) that may be caused by an ASD. It can also find heart muscle stress caused by an ASD.
* **Echocardiogram (echo).** This test uses sound waves to make a moving picture of the heart and heart valves. An echo can show the blood flow through the atrial septal opening and find out how big the opening is.
* **Cardiac catheterization.** This test uses a thin, flexible tube (catheter) put near the heart. Contrast dye is used to get even clearer pictures. In some children, this procedure may be used to close the ASD.

**TREATMENT**

Treatment will depend on your child’s symptoms, age, and general health. It will also depend on how severe the condition is. The most common type of ASD may close on its own as your child grows.

Once an ASD is diagnosed, your child's cardiologist will check your child to see if the defect is closing on its own. An ASD will usually be fixed if it has not closed by the time a child starts school. The decision to close the ASD may also depend on the size of the defect or the symptoms of the defect.

Treatment may include:

* **Medicine.** Many children have no symptoms and don't need medicine. But medicine can help some children's hearts work better. For example, water pills (diuretics) help the kidneys get rid of extra fluid from the body.
* **Surgery. Your** child's ASD may be repaired by surgery. The surgery is done under general anesthesia. The defect may be closed with stitches or a special patch.
* **Device closure.** Some children are helped with this procedure. The doctor uses cardiac catheterization to put a special device (septal occluder) in the open ASD. The device stops blood from flowing through the ASD.

## **possible complications**

## Large ASDs may cause lung problems over time if not treated. This is because the extra blood passing through the defect and then into the lungs may harm the vessels in the lungs.

## **How can I help my child live with an atrial septal defect?**

All children with an ASD need to be cared for by a pediatric cardiologist. Most children who have had an ASD repair will live healthy lives. After the repair, your child's doctor may want your child to take antibiotics. This will prevent an infection of the heart lining (bacterial endocarditis).

With early diagnosis and repair of an ASD, children usually do very well. They don't need much follow-up care. Children are more likely to have problems if an ASD is diagnosed later in life and never repaired. Or they may have problems if complications occur after closing the defect.

Some children develop high blood pressure in the lungs (pulmonary hypertension). These children should have follow-up care at a center that specializes in congenital heart disease.

Talk with your child's healthcare provider about the outlook for your child.

## **When to see a doctor**

Call your child's healthcare provider if your child has new symptoms or symptoms that get worse. Symptoms may include:

* Tiredness that gets worse
* Troubled breathing
* Fast breathing
* Racing or fluttering heartbeat (palpitations)
* Poor feeding

**DIFFERENTAIL DIAGNOSIS**

* Atrioventricular Septal Defect (AVSD)
* Ventricular Septal Defect (VSD)
* Innocent (Physiologic) Murmur
* Pulmonary (Pulmonic) Stenosis
* Total Anomalous Pulmonary Venous Return (TAPVR)
* Coronary Sinus Defect (Unroofed Coronary Sinus)
* Ostium Primum ASD (a type of partial AVSD)
* Sinus Venosus ASD (superior and inferior types)
* Patent Foramen Ovale (PFO)

**Pediatric Atrial Septal Defect (ASD) Epidemiology:**

* Incidence and Prevalence:  
  ASD occurs in approximately 1.6 to 3.9 per 1000 live births in children. It accounts for about 5-10% of all congenital heart defects (CHD) and is the third most common CHD after ventricular septal defects and patent ductus arteriosus.
* Types of ASD:  
  The most common type is ostium secundum ASD, comprising around 80% of cases. Other types like ostium primum, sinus venosus, and coronary sinus defects are less common in children.
* Age at Diagnosis:  
  Many ASDs are diagnosed in infancy or early childhood. For example, in a study from Ethiopia, 50% of children were diagnosed within the first year of life, and 29% during early childhood (1–5 years). However, some children remain asymptomatic and are diagnosed later.
* Sex Distribution:  
  ASD is more common in females, with female-to-male ratios reported between 1.3:1 to 2:1.
* Risk Factors:
  + Preterm birth is associated with a higher incidence of ASD, with preterm infants having 3 to 4 times increased risk compared to term infants.
  + Other risk factors include genetic predisposition and associated congenital anomalies.
* Natural History and Outcomes:  
  Many small ASDs close spontaneously in early childhood, while larger defects may require intervention. Untreated ASDs can lead to complications such as arrhythmias and heart failure in adulthood

**Genomic Data on Pediatric Atrial Septal Defect (ASD):**

* Genetic Heterogeneity:  
  ASD is a genetically heterogeneous congenital heart defect. Multiple genes and mutations contribute to its development, often involving cardiac transcription factors and structural proteins essential for heart septation.
* Key Genes Implicated:

|  |  |  |
| --- | --- | --- |
| Gene | Role & Mutation Impact | Notes |
| GATA4 | Zinc-finger transcription factor critical for cardiac morphogenesis. Mutations (e.g., p.P36S, p.H190R) cause ASD by disrupting septum formation. | Over 20 germline mutations identified; mutations co-segregate with familial ASD cases. |
| NKX2-5 | Homeobox gene involved in cardiac development. Mutations linked to ASD with atrioventricular conduction defects and arrhythmias. | Associated with familial ASD and progressive AV block; autosomal dominant inheritance. |
| TBX5 | Transcription factor mutated in Holt-Oram syndrome; causes ASD with limb abnormalities. | Nearly 100% penetrance in Holt-Oram; ~58% have ASD. |
| MYH6 | Cardiac-specific alpha-myosin heavy chain; mutations reduce sarcomeric function affecting atrial septation. | Familial ASD linked to MYH6 mutations; experimental models show disrupted septation. |
| ACTC1 | Cardiac actin gene; mutations reduce actin filament function, causing familial ASD. | Mutations identified in sporadic and familial ASD cases. |
| TBX20 | Transcription factor involved in cardiac development; mutations linked to septal defects. | Emerging evidence for role in ASD and other CHDs. |

* Genetic Syndromes Associated with ASD:
  + Holt-Oram Syndrome: TBX5 mutations; autosomal dominant; limb and cardiac defects.
  + Ellis-van Creveld Syndrome: Autosomal recessive; skeletal dysplasia and common atrium.
  + Other syndromes with chromosomal abnormalities may include ASD.
* Molecular Pathogenesis:  
  Mutations in these genes disrupt transcriptional regulation, cardiomyocyte proliferation, and sarcomeric protein interactions during atrial septum formation, leading to septal defects.
* Inheritance Patterns:  
  Many ASD cases are sporadic, but familial cases show autosomal dominant inheritance with variable penetrance.

**PREDEFINED Q AND A**

## 1. What is an atrial septal defect (ASD)?

An ASD is a hole in the wall (septum) between the two upper chambers of the heart (atria). This allows blood to flow between the left and right atria, which can affect normal blood circulation.

## 2. What causes ASD in children?

ASDs occur due to abnormal development of the atrial septum during fetal life. The exact cause is often unknown, but some cases are linked to genetic factors or syndromes. It is a congenital heart defect present at birth.

## 3. What are the types of ASD?

The main types are:

* Ostium secundum ASD (most common)
* Ostium primum ASD
* Sinus venosus ASD
* Coronary sinus ASD
* Patent foramen ovale (PFO) is not considered a true ASD but a flap-like opening that normally closes after birth.

## 4. What symptoms do children with ASD have?

Many children with small ASDs have no symptoms and grow normally. Larger ASDs may cause:

* Tiring easily or fatigue
* Shortness of breath, especially during activity
* Poor growth or feeding difficulties
* Frequent respiratory infections
* Heart palpitations or arrhythmias (less common in young children)

## 5. How is ASD diagnosed?

* A heart murmur is often the first sign detected by a doctor.
* Echocardiogram (ultrasound of the heart) confirms the diagnosis and shows the size and location of the defect.
* Other tests may include ECG, chest X-ray, or cardiac MRI if needed.

## 6. Can ASD close on its own?

Yes, small ASDs, especially ostium secundum types, may close spontaneously during infancy or early childhood. Larger defects usually require monitoring and sometimes intervention.

## 7. How is ASD treated?

* Small, asymptomatic ASDs may just need regular follow-up.
* Larger or symptomatic ASDs often require closure, either by catheter-based device or surgery, to prevent complications.
* Treatment decisions depend on the size of the defect, symptoms, and impact on heart function.

## 8. What complications can occur if ASD is untreated?

Untreated large ASDs can lead to:

* Enlargement and strain of the right side of the heart
* Pulmonary hypertension (high blood pressure in the lungs)
* Arrhythmias (irregular heartbeats)
* Increased risk of stroke in adulthood
* Heart failure in severe cases

## 9. What is the prognosis for children with ASD?

With appropriate diagnosis and treatment, most children with ASD do very well and lead normal lives. Early intervention prevents long-term complications.

## 10. When should I seek medical attention for my child?

Seek prompt care if your child has:

* Difficulty breathing or rapid breathing
* Poor feeding or failure to thrive
* Excessive tiredness or fatigue
* Signs of heart failure like swelling of legs or abdomen
* Palpitations or fainting episodes

**DOCTOR PATIENT CONVERSATION**

Doctor: Hello! I want to talk with you about your child’s heart condition called an atrial septal defect, or ASD. It’s sometimes called a “hole in the heart.”

Parent: What exactly is an ASD?

Doctor: An ASD is a small opening in the wall between the two upper chambers of the heart, called the atria. This hole lets some oxygen-rich blood from the left side mix with oxygen-poor blood on the right side, which means more blood than usual goes to the lungs.

Parent: How did this happen? Is it something we caused?

Doctor: No, ASDs happen during your child’s heart development before birth. Sometimes it can run in families, but often we don’t know the exact cause. It’s not due to anything you did during pregnancy.

Parent: What symptoms should we look for?

Doctor: Many children with ASD have no symptoms and grow normally. Larger defects can sometimes cause tiredness, poor growth, shortness of breath, or frequent lung infections. But most kids do very well.

Parent: How do you diagnose it?

Doctor: We usually hear a heart murmur during a checkup, which prompts us to do an echocardiogram — an ultrasound of the heart — to see the size and location of the hole.

Parent: Will the hole close on its own?

Doctor: Small ASDs often close by themselves during infancy or early childhood. If it doesn’t close or is large, we usually recommend closing it with a minimally invasive procedure or surgery, often before the child starts school.

Parent: What does the treatment involve?

Doctor: Many ASDs can be closed using a catheter-based procedure, where a device is placed through a vein in the leg to seal the hole. It usually requires just an overnight hospital stay and has a quick recovery. If that’s not possible, surgery is very effective too.

Parent: What happens if we don’t treat it?

Doctor: Untreated large ASDs can cause heart strain, irregular heartbeats, lung high blood pressure, and increased risk of stroke later in life. That’s why we recommend repair when needed.

Parent: Can my child live a normal life?

Doctor: Yes, with proper treatment and follow-up, most children with ASD live healthy, active lives without restrictions.

Parent: Is there anything special we should do at home?

Doctor: No special restrictions are usually needed. Just keep regular checkups and watch for any breathing problems or unusual tiredness.

Parent: Thank you for explaining everything.

Doctor: You’re welcome! We’re here to support you and your child every step of the way. Feel free to ask any questions anytime.

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**ATRIOVENTRICULAR SEPTIAL DEFECTS**

**DEFINITION AND DESCRIPTION**

An AVSD occurs when there are holes between the chambers of the right and left sides of the heart. This condition is also called atrioventricular canal (AV canal) defect or endocardial cushion defect. In people with AVSD, the valves that control blood flow between these chambers may not form correctly.

In AVSD, blood flows where it normally should not go. The blood may also have a lower-than-normal amount of oxygen, and extra blood can flow to the lungs. This extra blood being pumped into the lungs forces the heart and lungs to work harder than usual. This may lead to heart failure.

### Occurrence

About 1 in 1,712 (about 2,154) babies in the United States are born with an AVSD each year.

## Types

## There are two general types of AVSD that can occur. The types depend on which structures are not formed correctly.

## Complete AVSD

## A complete AVSD occurs when there is a large hole in the center of the heart. This allows blood to flow between all four chambers of the heart. This hole occurs where the septa (walls) separating the two top chambers *(atria)* and two bottom chambers *(ventricles)* normally meet. There is also one common valve in the center of the heart instead of two separate valves. This common valve often has leaflets (flaps) that may not be formed correctly or do not close tightly. A complete AVSD arises during pregnancy when:

* The common valve fails to separate into the two distinct valves, the tricuspid valve on the right side of the heart and the mitral valve on the left side, and
* The walls that split the upper and lower chambers of the heart do not grow all they way to meet in the center of the heart.

### Partial or incomplete AVSD

A partial or incomplete AVSD occurs when the heart has some, but not all of the defects of a complete AVSD. There is usually a hole in the atrial wall or in the ventricular wall near the center of the heart. A partial AVSD usually has both mitral and tricuspid valves, but one of the valves (usually mitral) may not close completely. This allows blood to leak backward from the left ventricle into the left atrium.

## **Signs and symptoms**

Babies with a complete AVSD usually have symptoms within the first few weeks after birth. When symptoms occur, they may include:

* Breathing problems
* Weak pulse
* Ashen or bluish skin color
* Poor feeding, slow weight gain
* Tiring easily
* Swelling of the legs or belly

Certain symptoms may indicate that a baby's complete AVSD or partial AVSD is getting worse. These symptoms include:

* **Arrhythmia (abnormal heart rhythm).** An arrhythmia can cause the heart to beat too fast, too slow, or irregularly. When the heart does not beat properly, it can't pump blood effectively.
* **Heart failure.** When the heart cannot pump enough blood and oxygen to meet the needs of the body.
* **Pulmonary hypertension**, a type of high blood pressure that affects the arteries in the lungs and the right side of the heart.

For partial AVSDs, the holes between the chambers of the heart may not be large. Therefore, signs and symptoms may not occur in the newborn or infancy periods. In these cases, people with a partial AVSD might not be diagnosed for years.

## **Complications**

## Infants who have surgical repairs for AVSD are not cured and may have lifelong complications. The most common complication is a leaky mitral valve. This is when the mitral valve does not close fully, allowing blood to flow backwards through the valve. A leaky mitral valve can cause the heart to work harder to get enough blood to the rest of the body. A leaky mitral valve might have to be surgically repaired.

## **Risk factors**

## The causes of AVSDs among most babies are unknown. Some babies have heart defects because of changes in their genes or chromosomes. A combination of genes and other risk factors may increase the risk for AVSD. These factors can include things in a mother's environment, what she eats or drinks, or the medications she uses during pregnancy.

## **Diagnosis**

## AVSD may be diagnosed during pregnancy or soon after the baby is born.

## During pregnancy

## During pregnancy, screening tests (prenatal tests) check for birth defects and other conditions. An ultrasound, a tool that creates pictures of the baby, may detect an AVSD. However, it usually depends on the size or type (partial or complete) of the AVSD.

## The healthcare provider can request a fetal echocardiogram to confirm the diagnosis if AVSD is suspected. A fetal echocardiogram is an ultrasound of the unborn baby's heart and shows more detail than the routine prenatal ultrasound test. The fetal echocardiogram can show problems with the structure of the heart and how well the heart is working.

## After the baby is born

## During a physical exam of an infant, a complete AVSD may be suspected. Using a stethoscope, a doctor may hear a heart murmur (a "whooshing" sound caused by irregular blood flow through the heart). However, not all heart murmurs are present at birth.

## A healthcare provider may request additional tests to confirm the diagnosis of AVSD. These tests include:

* Echocardiogram (ultrasound of the heart)
* Electrocardiogram (EKG) (measures electrical activity of the heart)
* Chest X-ray
* Other medical tests

## **Treatments**

All AVSD types usually require surgery. During surgery, any holes in the chambers are closed using patches. If the mitral valve does not close completely, it is repaired or replaced. For complete AVSD, the common valve is separated into two valves—one on the right side and one on the left.

The age for surgical repair depends on the child’s health and the specific structure of the AVSD. If possible, surgery should occur before there is permanent damage to the lungs from too much blood pumping to the lungs. Medication may be used to treat heart failure. However, this is only a short-term measure until the infant can grow large enough for surgery.

Even if their AVSD is surgically repaired, a child or adult with an AVSD needs regular visits with a cardiologist to:

* Monitor his or her progress
* Avoid complications
* Check for other health conditions that might develop as the child ages

With proper treatment, most babies with AVSD grow up to lead healthy, productive lives.

**Outlook / Prognosis**

Without surgery, children with an AV canal defect may have a life expectancy of two or three years. Some live to be young adults.

About 90% of children who have repair surgery have a 10-year survival rate. This means they live for at least another 10 years on average after treatment. About 65% are alive 20 years after surgery.

But even after surgery, someone with an atrioventricular canal defect won’t have a typical heart. They’ll need periodic echocardiograms to monitor their heart’s function and catch complications early.

The patch over the hole can usually stay in place for the rest of a person’s life. But over time, one of the repaired heart valves may begin to leak. About 10% to 20% of people need a second surgery.

After surgery, many people don’t need medications or more operations for their hearts. But cardiac arrhythmias may develop later in life. A provider may recommend minimally invasive procedures like ablation to treat an arrhythmia.

**Prevention**

There’s no way to prevent an atrioventricular septal defect. But if you’re pregnant, you can reduce the risk of your baby having a congenital heart defect by:

* Avoiding recreational drugs, alcohol and tobacco products
* Getting all necessary vaccinations to prevent illness
* Maintaining a weight that’s healthy for you
* Managing chronic health conditions
* Taking prenatal vitamins, including folic acid, as your healthcare provider recommends

**EPIDEMIOLOGY**

AVSD accounts for approximately 3% to 7% of all congenital cardiac malformations, with an estimated incidence of 0.24 to 0.31 per 1000 live births. While both sexes are affected, some study results suggest a slight female predominance, particularly in patients with Down syndrome, with a female-to-male ratio of 1.3 to 1.0.The association with trisomy 21 is significant, with 40% to 50% of children with Down syndrome having an AVSD, making it the most common congenital heart defect in this population.

Data from the Society of Thoracic Surgeons congenital database reported 4138 cases of complete AVSD between 2013 and 2017 when treated with or without valvuloplasty. However, cases requiring valve replacement demonstrated significantly higher perioperative mortality (16.7%).

Notably, intraoperative conversions from failed valvuloplasty to valve replacement had a mortality rate as high as 50%, underscoring the critical importance of accurate preoperative imaging and surgical planning to optimize outcomes. The widespread use of prenatal ultrasound and advances in congenital heart disease screening continue to influence reported incidence rates. Still, AVSD remains a common and clinically significant congenital defect, especially in populations with chromosomal abnormalities.

**DIFFERENTIAL DIAGNOSIS are:**

* Isolated Atrial Septal Defect (ASD)
  + Typically an ostium secundum or primum ASD without ventricular involvement.
  + Left-to-right shunt at atrial level only.
* Isolated Ventricular Septal Defect (VSD)
  + Left-to-right shunt at ventricular level; may cause similar heart failure symptoms.
* Patent Ductus Arteriosus (PDA)
  + Continuous “machine-like” murmur; left-to-right shunt from aorta to pulmonary artery.
* Tetralogy of Fallot (TOF)
  + Cyanotic congenital heart disease with VSD, pulmonary stenosis, overriding aorta, and right ventricular hypertrophy.
* Pulmonic Stenosis
  + Obstruction at pulmonary valve causing right ventricular hypertrophy and murmur.
* Total Anomalous Pulmonary Venous Return (TAPVR)
  + Pulmonary veins drain abnormally; presents with cyanosis and heart failure.
* Congestive Heart Failure from Other Causes
  + Such as cardiomyopathies or myocarditis.
* Sepsis or Other Non-Cardiac Causes of Tachypnea and Failure to Thrive
  + Important to rule out in infants presenting with respiratory distress.

**Genetic Associations:**  
AVSD is strongly associated with chromosomal abnormalities and genetic syndromes, most notably Down syndrome (trisomy 21). Key genes on chromosome 21 implicated in AVSD include DSCAM, COL6A1, COL6A2, KCNJ6, and RCAN1, which are important for cardiac development.

* Syndromic and Nonsyndromic Cases:  
  While AVSD frequently occurs in syndromic contexts (e.g., Down syndrome, heterotaxy syndrome), it can also present as an isolated defect. Familial cases are rare but usually show autosomal dominant inheritance with variable expression.
* Candidate Genes and Mutations:
  + CRELD1: Mutations in this gene are found in about 9% of partial AVSD cases and are associated with abnormal atrioventricular cushion development.
  + Cilia-related genes: Mutations affecting cilia function and left-right patterning (e.g., Polaris) disrupt cardiac development and cause AVSD, sometimes independent of situs abnormalities.
  + Other genes: Over 100 mutations identified in mouse models suggest a broad range of genetic contributors affecting cardiac septation and valve formation

**PREDEFINED Q AND A**

## . What is an atrioventricular septal defect (AVSD)?

AVSD, also called an atrioventricular canal defect or endocardial cushion defect, is a congenital heart defect where there are holes between the heart’s upper chambers (atria) and lower chambers (ventricles), along with abnormalities of the valves that control blood flow between these chambers.

## 2. What causes AVSD?

The exact cause is unknown, but AVSD occurs due to abnormal development of the endocardial cushions during fetal heart formation. It is more common in children with Down syndrome (trisomy 21), occurring in about 25% of these children.

## 3. What are the types of AVSD?

* Complete AVSD: Large holes between all four heart chambers and one common atrioventricular valve.
* Partial AVSD (primum ASD): Smaller defects, usually involving only the atrial septum and two separate but abnormal valves.
* Transitional AVSD: Features between complete and partial forms.

## 4. What symptoms does AVSD cause?

Symptoms usually appear between 4 and 8 weeks of age and include:

* Rapid breathing
* Feeding difficulties
* Poor weight gain
* Fatigue and low energy
* Sweating, especially during feeding  
  If untreated, it can lead to congestive heart failure and pulmonary hypertension.

## 5. How is AVSD diagnosed?

* Physical exam may reveal a heart murmur.
* Echocardiography (heart ultrasound) is the key test to visualize the septal defects and valve abnormalities.
* Chest X-ray and ECG may support diagnosis.

## 6. What treatments are available for AVSD?

Most children require surgical repair, usually before 6 months of age, especially those with complete AVSD. Surgery involves closing the holes and reconstructing the valves. Some partial AVSDs may be monitored if asymptomatic.

## 7. What is the prognosis after surgery?

Surgical outcomes are generally very good, with most children doing well long-term. However, some may develop valve leakage requiring further intervention.

## 8. Can children with AVSD participate in normal activities?

After repair and recovery, children usually have no restrictions and can participate in normal physical activities. Children with Down syndrome may have additional considerations.

## 9. Are there any complications to watch for?

Possible complications include valve leakage, arrhythmias, pulmonary hypertension, and the need for additional surgeries.

## 10. Is AVSD hereditary?

Most cases are sporadic, but there is an increased risk in families with genetic syndromes like Down syndrome. Genetic counseling may be recommended.

**DOCTOR PATIENT CONVERSATION**

Doctor: Hello, I want to talk with you about your child’s heart condition called an atrioventricular septal defect, or AVSD.

Parent: What exactly is that?

Doctor: AVSD is a congenital heart defect where there are holes between the heart’s upper chambers (atria) and lower chambers (ventricles). Also, the valves that control blood flow between these chambers don’t form properly and instead form one large valve. This causes extra blood to flow to the lungs, which can strain the heart.

Parent: How does this affect my child?

Doctor: Because of the holes and valve problems, blood flows abnormally inside the heart, leading to too much blood going to the lungs. This can cause symptoms like rapid breathing, poor feeding, fatigue, and eventually heart failure if untreated.

Parent: How do you diagnose AVSD?

Doctor: We usually hear a heart murmur during a checkup. To confirm, we perform an echocardiogram, which is an ultrasound of the heart. It shows the size and location of the holes and the valve abnormalities.

Parent: Is this common in any particular group?

Doctor: AVSD is more common in children with Down syndrome, but it can occur in any child.

Parent: What treatment options are there?

Doctor: Most children with AVSD need surgery, usually within the first 6 months of life. The surgery closes the holes and repairs the valve. Early surgery helps prevent complications like lung damage and heart failure.

Parent: What happens if we don’t treat it?

Doctor: Without treatment, the extra blood flow can cause lung damage, heart failure, and other serious problems.

Parent: What is the outlook after surgery?

Doctor: The prognosis is generally good. Most children recover well and lead healthy lives, though some may need follow-up care for valve function or rhythm problems.

Parent: Is there anything we can do at home?

Doctor: Keep up with regular checkups, watch for symptoms like rapid breathing or poor feeding, and follow the treatment plan. We’ll support you throughout.

Parent: Thank you for explaining everything.

Doctor: You’re welcome. Please ask any questions anytime—we’re here to help.

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**BRONCHOPULMONARY SEQUESTRATION**

**DEFINTION AND DESCRIPTION**

Bronchopulmonary sequestration (BPS) is a rare congenital disease that affects the respiratory system, presenting unique challenges for both patients and healthcare providers. This congenital malformation involves abnormal lung tissue development, leading to a mass or cyst that does not communicate with the normal bronchial tree and receives its blood supply from abnormal vessels. This article briefly explains bronchopulmonary sequestration, its causes, types, symptoms, diagnosis, and treatment.

Bronchopulmonary sequestration is a rare congenital anomaly characterized by the presence of non-functional lung tissue that lacks a connection to the normal airway. Essentially, it is a mass or cyst that receives its blood supply from systemic vessels rather than the pulmonary arteries. This abnormality typically manifests in the lower lobes of the lungs.

## **Causes Bronchopulmonary Sequestration**

Bronchopulmonary sequestration occurs during fetal development when a portion of the lung tissue becomes isolated from the normal bronchial tree. This non-functional lung tissue then forms a mass that receives its blood supply from systemic vessels rather than the pulmonary artery.

## **Types of Bronchopulmonary Sequestration (BPS)**

BPS is classified into two main types: intralobar and extralobar.

**Intralobar Bronchopulmonary Sequestration**: This type is more common and has a reported incidence of 75 percent to 86 percent among all sequestration cases. It involves a mass of abnormal lung tissue within a normal lung lobe. Intralobar BPS shares a common pleural covering with the surrounding lung tissue.

**Extralobar Bronchopulmonary Sequestration**: Extralobar BPS is rarer and involves a separate mass of lung tissue with its pleural covering. The mass is often located outside the normal lung tissue, usually in the chest cavity.

## **Symptoms of Bronchopulmonary Sequestration (BPS)**

The symptoms of BPS may vary depending on the type of bronchopulmonary sequestration.

1. **Intralobar Sequestration**:

**Frequently Asymptomatic**: Many individuals do not experience any symptoms and are diagnosed incidentally during chest CT (computed tomography) scans.

**Recurrent Pneumonia**: The most common presentation, often affecting a specific area of the lung.

**Persistent Cough**: This can be a chronic symptom, separate from or accompanying pneumonia.

**Back Pain**: This may be associated with the location of the sequestrated tissue.

**Persistent Exertional Shortness of Breath**: Difficulty breathing during physical activity.

**Hemoptysis**: Coughing up blood occurs more frequently than with extralobar sequestration.

2. **Extralobar Sequestration**: Extralobar sequestration is usually manifested clinically in early infancy with

**Respiratory Distress**: Difficulty breathing, often noticeable soon after birth.

**High Output Congestive Heart Failure**: Caused by a blood flow abnormality.

**Occasional Spontaneous Pulmonary or Pleural Hemorrhage**: Bleeding in the lungs or surrounding space.

**Rarely Infected**: Due to separation from the airway system by surrounding tissue.

## **Bronchopulmonary Sequestration Diagnosis**

Diagnosing bronchopulmonary sequestration often involves the following:

**Chest X-ray**: Provides initial clues, especially for intralobar sequestration in young patients with recurrent infections.

**Computed Tomography (CT) Scan**: Highly accurate (90 %) for both types of sequestration, especially with 3D reconstruction and contrast. The current gold standard for non-invasive diagnosis.

**Magnetic Resonance Imaging (MRI) and MR Angiography (MRA)**: Similar information to CT scans but less widely used.

**Ultrasonography**: Ideal for prenatal and postnatal settings, especially with color flow and duplex Doppler for identifying abnormal blood supply.

**Radionuclide Angiography**: Demonstrates systemic arterial blood supply to the sequestration, confirming diagnosis.

**Angiography**: This invasive procedure involves injecting a contrast dye into the blood vessels to visualize the blood supply to the anomalous lung tissue. The definitive diagnosis is made by using angiography. Angiography helps differentiate pulmonary sequestration from other abnormalities of the lung. Bleeding in the lungs must be interpreted along with clinical and chest radiographic findings.

**Doppler Ultrasound**: Can detect sequestration as early as 18 to 19 weeks' gestation.

## **Possible Complications of Bronchopulmonary Sequestration**

Bronchopulmonary sequestration can lead to the following complications:

Growth abnormalities.

Hemoptysis.

Hemorrhagic pleural effusion.

Recurrent infection is common in intralobar sequestration.

Rare cases of extra lobar sequestrations communicating with the GI (gastrointestinal) tract have been reported. They are known as congenital bronchopulmonary foregut malformations (CBPFM).

Development of malignant tumors within intralobar sequestration.

## **Treatment of Bronchopulmonary Sequestration**

The management of bronchopulmonary sequestration depends on various factors, including the patient's age, the presence of symptoms, and the size and location of the sequestration.

1. **Early Intervention**:

**Hospitalization and Intensive Care**: Specialized treatment is crucial due to potential complications like pulmonary hypoplasia (underdeveloped lungs).

**Thoracoamniotic Shunting**: For fetuses with hydrops (fluid accumulation) before 30 weeks' gestation, this procedure helps drain excess fluid.

**Postnatal Support**: Depending on severity, newborns may require ventilator support, high-frequency oscillatory ventilation, or even extracorporeal membrane oxygenation (ECMO).

**Tube Thoracostomy**: This procedure drains large pleural effusions (fluid around the lungs) in severe cases.

**Surgical Resection**: Ultimately, surgery to remove the sequestered tissue is recommended, even for asymptomatic infants, to prevent future infections and inflammation. However, in certain cases with pulmonary hypoplasia and hypertension, surgery may be delayed until stabilization occurs.

2. **Pulmonary lobectomy**: The preferred treatment for symptomatic patients, even in some asymptomatic cases, to avoid complications.

3. **Surgery Options**:

**Open Thoracotomy**: Traditional open chest surgery approach.

**Video-Assisted Thoracoscopic Surgery (VATS)**: Minimally invasive approach with smaller incisions, leading to potentially faster recovery times and fewer complications. Recent advancements even include uniportal VATS, requiring only one small incision.

4. **Endovascular Embolization**: This minimally invasive technique involves blocking blood flow to the sequestered tissue using coils or other embolic agents like particles, glue, plugs, or alcohol. This leads to shrinkage and eventual disappearance of the tissue.

## **Differential Diagnosis of Bronchopulmonary Sequestration**

Symptoms of bronchopulmonary sequestration (BPS) can mimic many other lung conditions, for example:

Cystic adenomatoid malformation (non-cancerous overgrowth of abnormal lung tissue).

Bronchogenic cyst (congenital abnormality arising from an abnormal budding of the tracheobronchial tree).

Focal bronchiectasis (usually occurs when the bronchi (large airway) become dilated).

Congenital lobar emphysema (developmental abnormality of lungs characterized by overinflation of a lung lobe due to a blockage of the airways).

Retroperitoneal tumors in extra lobar abdominal sequestrations.

* Pneumonia  
  Infectious consolidation can mimic sequestration, especially if recurrent or localized.
* Lung Abscess  
  Cavitary lesion with infection, often with systemic signs.
* Empyema  
  Pleural fluid collection with infection, may appear as a mass on imaging.
* Bronchiectasis  
  Dilated bronchi with chronic infection, can appear cystic or mass-like.
* Congenital Pulmonary Airway Malformation (CPAM) (formerly Congenital Cystic Adenomatoid Malformation - CCAM)  
  Cystic lung lesion that can resemble sequestration; may coexist as hybrid lesions.
* Diaphragmatic Hernia  
  Herniation of abdominal contents into thorax, may mimic mass on chest imaging.
* Tuberculosis  
  Chronic infection causing cavitary or mass-like lesions.
* Bronchogenic Cyst  
  Congenital cystic lesion, well-circumscribed, usually mediastinal or intrapulmonary.
* Pulmonary Arteriovenous Malformation (AVM)  
  Abnormal vascular connections, can be confused with sequestration vascular supply.
* Lung Tumors  
  Rare in children but includes inflammatory myofibroblastic tumor or congenital lung tumors.
* Pneumatocele  
  Air-filled cystic space often post-infectious.
* Foregut Duplication Cyst  
  Congenital cystic lesion adjacent to the esophagus or bronchus.
* Pulmonary Infarction or Round Atelectasis  
  Can appear as mass-like opacity on imaging.
* Neuroblastoma or Other Mediastinal Masses (for infradiaphragmatic or thoracoabdominal lesions)  
  May mimic sequestration on imaging.

**Epidemiology of Bronchopulmonary Sequestration (BPS):**

* Prevalence and Incidence:  
  BPS is a rare congenital lung malformation, estimated to occur in approximately 0.1% to 0.42 per 10,000 live births based on recent large population studies, such as data from the Chinese Birth Defects Monitoring Network. Other estimates report an incidence ranging from 0.15% to 1.8% of all congenital lung malformations. Some sources cite an incidence of about 1 in 60,000 children.
* Types and Presentation:
  + Extralobar sequestration (ELS) accounts for most cases diagnosed prenatally and shows a male predominance (historically up to 4:1), though recent data suggest similar prevalence between sexes.
  + Intralobar sequestration (ILS) tends to present later in childhood or adolescence with recurrent infections and is more common in the left lower lobe (about 70% of cases).
  + BPS most commonly involves the left lower lobe, particularly the posterior basal segment.
* Sex Distribution:  
  Slight male predominance has been reported, especially for extralobar sequestration, with male-to-female ratios around 1.2 to 1.6:1 in some series.
* Age at Diagnosis:
  + ELS is often diagnosed prenatally or in neonates due to respiratory distress or infection.
  + ILS is frequently diagnosed in late childhood or adulthood after recurrent pulmonary infections

**PREDEFINED Q AND A**

## 1. What is bronchopulmonary sequestration (BPS)?

Bronchopulmonary sequestration is a rare congenital lung malformation where a mass of nonfunctioning lung tissue develops without a normal connection to the airways. This tissue receives blood supply abnormally from a systemic artery, usually from the aorta, rather than from the pulmonary arteries.

## 2. What are the types of bronchopulmonary sequestration?

There are two main types:

* Extralobar sequestration (ELS): The abnormal lung tissue is outside the normal lung and has its own pleural covering. It is often diagnosed in infants and may be associated with other congenital abnormalities.
* Intralobar sequestration (ILS): The abnormal tissue is located within a normal lung lobe and shares the same pleural covering. It usually presents later in childhood or adulthood with recurrent infections.

## 3. How common is bronchopulmonary sequestration?

BPS is a rare condition, occurring in approximately 0.1% of live births. It most commonly affects the lower lobes of the lungs, especially the left lower lobe.

## 4. How is bronchopulmonary sequestration diagnosed?

BPS is often detected prenatally via ultrasound, which shows a bright mass near the lung with a distinct artery coming from the aorta feeding the lesion. Postnatal diagnosis is confirmed with imaging studies such as CT angiography or MRI to visualize the abnormal lung tissue and its blood supply.

## 5. What symptoms does bronchopulmonary sequestration cause?

Many cases are asymptomatic at birth. Symptoms, when present, may include:

* Recurrent pneumonia or lung infections
* Cough
* Breathing difficulties
* Chest pain
* In severe cases, heart failure due to abnormal blood flow

## 6. What complications can arise from BPS?

* Infection of the sequestered lung tissue
* Torsion (twisting) of the extralobar sequestration, which can cause pain and compromise blood flow
* Compression of normal lung tissue or heart displacement in large lesions
* Rarely, fetal hydrops (fluid accumulation) and heart failure before birth

## 7. How is bronchopulmonary sequestration treated?

* Surgical removal of the sequestered lung tissue is the standard treatment, especially for symptomatic cases or large lesions.
* Small, asymptomatic extralobar sequestrations may sometimes be monitored without immediate surgery.
* In rare fetal cases with complications, prenatal interventions or specialized delivery procedures may be necessary.

## 8. What is the prognosis for children with BPS?

With appropriate surgical treatment, most children have an excellent prognosis and normal lung function. Early diagnosis and management help prevent complications such as infections and heart failure.

## 9. Can bronchopulmonary sequestration affect pregnancy?

Large lesions can sometimes impair lung development or cause fluid buildup around the fetus (polyhydramnios or hydrops), which may require close monitoring or fetal intervention.

## 10. Is bronchopulmonary sequestration hereditary?

BPS is a congenital malformation but is generally considered sporadic with no clear hereditary pattern.

**DOCTOR PATIENT CONVERSATION**

Doctor: Hello, I want to discuss the diagnosis we made for your child. The imaging shows a condition called bronchopulmonary sequestration, or BPS.

Parent: What is bronchopulmonary sequestration?

Doctor: It’s a rare congenital condition where a small part of lung tissue develops separately from the normal lung. This tissue doesn’t connect to the normal airways, so it doesn’t help with breathing. Instead, it gets its blood supply abnormally from a large artery coming directly from the aorta.

Parent: How does this affect my child?

Doctor: Often, this extra lung tissue doesn’t cause symptoms at first. But it can sometimes lead to repeated lung infections, cough, or breathing difficulty. In some cases, the mass can press on nearby lung tissue or the heart.

Parent: How did you find out about it?

Doctor: In many cases, like yours, it’s detected on prenatal ultrasound or after birth with imaging studies such as CT angiography or MRI. These tests help us see the abnormal lung tissue and its blood supply.

Parent: What treatment does my child need?

Doctor: The standard treatment is surgical removal of the sequestered lung tissue, especially if your child has symptoms or if the mass is large. Surgery usually has excellent outcomes. If the lesion is small and not causing problems, sometimes we monitor it closely.

Parent: Are there any risks if we don’t treat it?

Doctor: Without treatment, there’s a risk of recurrent infections, inflammation, or complications like torsion, where the blood supply twists and causes pain or damage. Large lesions can also cause problems with lung development or heart function.

Parent: What is the outlook after surgery?

Doctor: Most children recover well and have normal lung function afterward. Early diagnosis and treatment help prevent complications.

Parent: Is this condition hereditary? Could future children have it?

Doctor: BPS is generally considered a sporadic congenital condition, meaning it usually happens by chance and is not inherited. The risk for future children is very low.

Parent: What should we watch for at home?

Doctor: Watch for symptoms like persistent cough, fever, difficulty breathing, or poor feeding. If any of these occur, seek medical attention promptly.

Parent: Thank you for explaining. It’s a relief to know more.

Doctor: You’re welcome. We’ll support you and your child every step of the way. Please feel free to ask any questions anytime.

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### **Chylothorax**

**DEFINITION AND DESCRIPTION**

Chylothorax (pronounced “kai-low-THOR-aks”) is a condition where fluid (chyle) from your lymphatic system leaks into the space around your lungs. Your lymphatic system — a series of vessels similar to your blood vessels — drains extra fluid from your tissues. White blood cells patrol the fluid (lymph) to look for and destroy germs like bacteria and viruses.

Chyle is a kind of lymph that comes from your digestive tract. It contains a mix of white blood cells and fats that make it look milky. It moves through your body through your thoracic duct. Your thoracic duct is a tube that moves chyle from your abdomen, up through your chest, to your neck. There, it empties back into your bloodstream.

Damage to your thoracic duct can cause it to leak. If it leaks into the space around your lungs, it’s called chylothorax.

#### **Types of chylothorax**

Chylothorax is either traumatic, non-traumatic or idiopathic:

* **Traumatic chylothorax** happens when the lymph vessels in your abdomen or chest are damaged by force. This could be during surgery or after an injury.
* **Non-traumatic (spontaneous) chylothorax** happens when something blocks your lymph vessels, weakens their walls or causes a buildup of chyle that makes the vessels burst. People who have anatomical differences in their chest can be born with non-traumatic chylothorax (congenital chylothorax). They can also develop it a few days after birth.
* **Idiopathic chylothorax** is any kind of chylothorax where your provider can’t find the cause.

## **Symptoms**

The main symptom of chylothorax is shortness of breath. Some people also experience:

* Chest pressure, tightness or heaviness.
* Tiredness (fatigue).
* Unintended weight loss.

### **Common cause of chylothorax**

Surgery is the most common cause of chylothorax (postoperative chylothorax). But anything that can damage your thoracic duct, which carries chyle, can cause chylothorax. Damage can cause chyle to leak from your lymphatic system into your chest. Other causes include:

* **Injury to your chest or back**. This includes blunt trauma injuries (something hitting your body hard) and injuries that break through your skin (like a stab wound).
* **Present at birth**. Differences in anatomy and certain illnesses you’re born with can cause chylothorax. Babies either have it at birth (congenital) or develop it in the first few days after birth.
* Many **cancers** can block or damage structures in your chest, allowing chyle to leak. This includes lymphoma, chronic lymphocytic leukemia, lung cancer, esophageal cancer, Kaposi sarcoma and others.
* Sometimes, **infections** can damage the ducts that carry chyle, allowing it to leak. Tuberculosis is the most common cause of this kind of chylothorax.
* **Blockage**. Anything that can cause conditions that block your thoracic duct can lead to chylothorax. This includes sarcoidosis, blood clots and goiter.

#### **Risk factors for chylothorax**

You might be at higher risk for chylothorax if you:

* Had recent surgery to your chest, neck or abdomen. This includes lung and cardiovascular (heart or blood vessel) surgery.
* Had an injury to your chest or back.
* Have cancer or a condition that can cause blockages in your chest or buildup in your lymphatic system.

**Diagnosis and Tests**

After listening to your symptoms and health history, a healthcare provider diagnoses chylothorax with imaging tests and by testing a sample of fluid from your chest.

A provider may perform or order these tests to diagnose chylothorax:

* **Imaging**. Your provider needs to see pictures of the inside of your chest to know if excess fluid is causing your symptoms. They might use X-rays, ultrasounds or CT scans.
* **Thoracentesis**. Your provider might drain fluid from your chest and test it. This tells them more about what the fluid is and what might be causing it to leak.
* **Lymphangiography** or **lymphoscintigraphy**. These are special imaging tests to look at your lymphatic system.

## **Management and Treatment**

How your provider treats chylothorax depends on the cause. They may suggest a special diet or drain the fluid first. If those don’t work, they may use a surgical procedure to seal the leak (lymphangiogram and embolization) or to keep the fluid from leaking again. Treating the underlying cause of chylothorax is the best way to keep it from coming back.

#### **Specific therapies that treat chylothorax**

Your provider may recommend the following treatments:

* **Bowel rest**. Your lymph vessels that transport chyle may need a break so they can heal. To do this, your provider will give you all of your nutrition through an IV. This gives your body a rest from breaking down food through your digestive system.
* **Dietary changes**. To help give your body a break from digesting fats, a dietitian can guide you through a very low-fat or modified-fat diet. This is only temporary. Limiting healthy fats in your diet for too long can lead to malnutrition.
* **Draining fluid**. A provider may drain the fluid from your chest using thoracentesis. They might drain it once or over a short period of time with a chest tube. While it can’t repair damage on its own, this can relieve symptoms while your body heals.
* **Somatostatin or octreotide**. These are IV medications that change how your body absorbs fats and nutrients. Providers sometimes use them in combination with other treatments, like diet changes, to help your body heal.
* **Thoracic duct ligation or embolization**. If your thoracic duct is damaged or doesn’t work properly, a provider may surgically tie it off (ligation) or purposefully block it (embolization). This can repair damage or redirect the flow of fluid so it doesn’t leak.
* **Pleurodesis**. Pleurodesis is a surgical procedure that sticks your lungs to the lining on the inside of your chest wall. This gets rid of the space inside your chest where the fluid is collecting.

## **Outlook / Prognosis**

What you can expect with chylothorax depends on what’s causing it and how well you respond to treatment. Mild leaks can sometimes heal on their own or with temporary dietary changes. But you might need surgery to fix the damage. Ask your provider what to expect in your specific situation.

Chylothorax can be life-threatening if left untreated. It can also be caused by serious illnesses. You may need ongoing treatments for underlying illnesses.

**Living With**

Your provider may suggest you go on a special diet if you have chylothorax. This includes eating and drinking foods and beverages with very little fat or specific kinds of fats.

They may also recommend that you not eat at all. In this case, you’ll get all of your nutrition in an IV, giving your body time to heal. In either case, you should be able to return to your normal diet within a week or two.

### **When to see a doctor**

See your healthcare provider if you have unexplained shortness of breath, especially if you:

* Had surgery recently.
* Had a recent injury to your chest or back.
* Have a condition that can cause blockages in your chest or neck.
* Have a condition that affects your lymphatic system.

Go to the nearest emergency room if you have trouble breathing, especially if you also have chest pain. This could be a sign of a life-threatening illness.

## **Main Differential Diagnoses for Pediatric Chylothorax:**

1. Traumatic Causes:
   1. Iatrogenic injury during cardiothoracic surgery (most common cause in children)
   2. Chest trauma or thoracic duct injury
2. Congenital/Idiopathic Causes:
   1. Congenital lymphatic malformations (e.g., lymphangiomatosis, lymphangiectasia)
   2. Syndromic associations:
      1. Down syndrome
      2. Noonan syndrome
      3. Turner syndrome
      4. Yellow nail syndrome
      5. Hydrops fetalis
   3. Idiopathic neonatal chylothorax (most common pleural effusion in newborns)
3. Malignancy-Associated Chylothorax:
   1. Lymphoma
   2. Leukemia
   3. Other mediastinal tumors causing thoracic duct obstruction
4. Infectious Causes:
   1. Tuberculosis
   2. AIDS-related complex (rare in pediatrics)
5. Cardiac Causes:
   1. Congestive heart failure causing pleural effusions that may mimic chylothorax
   2. Post-cardiac surgery complications (e.g., after congenital heart defect repair)
6. Other Causes:
   1. Gorham-Stout disease (massive osteolysis with associated chylous effusions)
   2. Lymphatic obstruction due to thoracic duct thrombosis or malformations
   3. Exudative pleural effusions (non-chylous) that can be confused with chylothorax
   4. Pseudochylothorax (chylous-like effusion with different biochemical profile)

## **Procedures**

Thoracentesis and pleural fluid analysis are the criterion standards to establish a diagnosis of chylothorax. Alternatively, in a postsurgical patient, tube thoracostomy output can be analyzed.

Pleural fluid analysis for triglyceride content helps to confirm the diagnosis of chylothorax. Note the following:

* A level greater than 110 mg/dL reflects a 99% chance that the fluid is chyle.
* A level less than 50 mg/dL reflects only a 5% chance that the fluid is chyle.
* If the level is 50-110 mg/dL, use lipoprotein analysis or inspect the pleural fluid for chylomicrons or cholesterol crystals.
* A ratio of pleural fluid cholesterol to triglyceride of less than 1 is also diagnostic.

The concentration of cholesterol in a chylothorax will often be less than 200 mg/dL.

A fasting patient may have serous-appearing pleural fluid. To confirm the diagnosis, administer cream through a Naso enteric tube prior to fluid collection. The cream will change the chylous production from serous to the characteristic milky white fluid. This change is diagnostic for a chyle leak.

The gold standard for chylothorax diagnosis is detecting chylomicrons within the pleural fluid via lipoprotein electrophoresis.

Chylothorax can be distinguished from pseudo chylothorax by fluid analysis. In pseudo chylothorax, the cholesterol level is greater than 200 mg/dL, no chylomicrons are present, and cholesterol crystals are seen on microscopy. There is often a neutrophil-rich exudate with triglyceride levels less than 50 mg/dL

**Pediatric Chylothorax Epidemiology:**

* Prevalence:  
  Chylothorax is a rare condition in children, with an estimated prevalence of about 1 in 15,000 live births.
* Postoperative Incidence:  
  The most common cause in pediatrics is postoperative chylothorax, especially following cardiothoracic surgery. The incidence after pediatric cardiac surgery ranges from 0.2% to 6.6%, with many studies reporting around 2–4.5%. Certain surgeries like cavopulmonary bypass have higher rates (up to 11.9%).
* Etiology Distribution:
  + Approximately 80–90% of pediatric chylothorax cases are post-surgical, mainly after cardiac operations.
  + Primary (congenital or idiopathic) chylothorax accounts for a smaller proportion, including neonatal or congenital cases.
  + Other causes include trauma, malignancy, and lymphatic anomalies.
* Gender Distribution:  
  Pediatric cases show a nearly equal male-to-female ratio (about 56% male, 44% female), with no significant gender predilection.
* Age at Presentation:
  + Neonates and infants often present with congenital or idiopathic chylothorax.
  + Postoperative chylothorax typically develops within days after surgery.
* Morbidity:  
  Chylothorax is an important cause of morbidity and prolonged hospitalization in critically ill children.

### **PREDEFINED Q AND A**

### **What’s the difference between chylothorax and pleural effusion?**

Pleural effusion is any fluid in the pleural space (space between your lungs and your chest wall). Chylothorax is a specific kind of pleural effusion where chyle leaks into the pleural space.

## What caused this?

Pediatric chylothorax is caused by leakage of lymphatic fluid (chyle) into the chest cavity. The most common causes in children are:

* Injury or disruption of the thoracic duct during cardiac surgery (postoperative chylothorax).
* Congenital lymphatic malformations or abnormalities.
* Trauma, infections, or rarely tumors that block or damage lymphatic drainage.

## What are my treatment options?

Treatment usually starts with conservative measures, including:

* Chest tube drainage to remove fluid.
* Dietary modifications such as a low-fat diet enriched with medium-chain triglycerides (MCT) or complete bowel rest with total parenteral nutrition (TPN) to reduce chyle production.
* Medications like octreotide (a somatostatin analogue) which helps reduce lymphatic fluid production.
* If conservative treatment fails (usually after 2 weeks or high output persists), surgical options such as thoracic duct ligation or minimally invasive procedures like video-assisted thoracoscopic surgery (VATS) are considered.
* Newer interventional treatments include selective lymphatic duct embolization, which blocks leaking lymphatic vessels with coils or glue.

## What should/shouldn’t I eat or drink?

* Your child will likely be started on a low-fat diet enriched with medium-chain triglycerides (MCT), which are absorbed directly into the bloodstream and reduce lymph flow.
* In some cases, complete bowel rest with intravenous nutrition (TPN) is necessary to allow the thoracic duct to heal.
* Avoid long-chain fatty acids and high-fat foods until your doctor advises otherwise.

## Is there any way to prevent this from happening again?

* Prevention depends on the cause:
  + After surgery, careful surgical technique reduces risk.
  + For congenital or idiopathic cases, close monitoring and early treatment help prevent complications.
  + Avoiding trauma and managing underlying conditions is important.
* If your child had surgery, your care team will monitor closely to catch any recurrence early.

## How long will it take to feel better?

* The median time to resolution of chylothorax in children is about 3 weeks (around 21 days) with conservative treatment.
* Some children recover faster, while others may require longer treatment, especially if surgery or embolization is needed.
* Recovery also depends on your child’s overall health and the underlying cause.

**Pediatric Chylothorax Treatment Drugs and Their Side Effects**

## 1. Octreotide

* Mechanism: Synthetic somatostatin analogue that reduces lymphatic fluid production by causing vasoconstriction in splanchnic circulation and reducing intestinal blood flow.
* Usage: Administered as subcutaneous injections (20–70 µg/kg/day divided into 3 doses) or intravenous infusion (starting 1–4 µg/kg/hr, titrated up to 10 µg/kg/hr). Typical duration ranges from 3 to 29 days depending on response.
* Effectiveness: Most pediatric chylothorax cases (about 89%) respond well to octreotide combined with dietary management.
* Side Effects:
  + Hyperglycemia
  + Hypothyroidism
  + Gastrointestinal symptoms (nausea, diarrhea, cramps)
  + Necrotizing enterocolitis (rare, especially in neonates)
  + Liver dysfunction
  + Renal impairment (rare)

## 2. Total Parenteral Nutrition (TPN)

* Purpose: Provides bowel rest by eliminating enteral fat intake, reducing chyle production.
* Usage: Used especially in infants and children with high chyle output or when dietary modification alone is insufficient.
* Side Effects:
  + Infection risk due to central venous catheter
  + Electrolyte imbalances
  + Liver dysfunction with prolonged use
  + Metabolic bone disease

## 3. Albumin Replacement

* Purpose: To replace protein losses from chylous drainage, especially when losses exceed 50 mL/kg/day.
* Side Effects: Allergic reactions (rare), volume overload if not carefully monitored.

## 4. Immunoglobulin Transfusion

* Purpose: To manage hypogammaglobulinemia resulting from loss of immunoglobulins in chyle.
* Side Effects: Allergic reactions, fever, headache (generally well tolerated).

## 5. Inotropes (if cardiac dysfunction present)

* Used selectively if diastolic dysfunction or heart failure complicates the clinical picture. Side effects depend on the specific agent used.

**DOCTOR PATIENT CONVERSATION**

Doctor: Hello, I want to discuss your child’s diagnosis of chylothorax, which means there is lymphatic fluid, called chyle, accumulating in the chest cavity.

Parent: What causes this to happen?

Doctor: In children, chylothorax most commonly occurs after heart surgery because the lymphatic vessels can get injured during the procedure. It can also happen due to congenital lymphatic abnormalities or rarely from trauma or infections.

Parent: What are the treatment options?

Doctor: We usually start with conservative treatment, which includes draining the fluid with a chest tube and modifying the diet to reduce fat intake, often using special formulas with medium-chain triglycerides. Sometimes, we give total parenteral nutrition (feeding through an IV) to rest the gut. We also use medications like octreotide to reduce lymph flow. If these measures don’t work after a few weeks, surgery or other interventions may be needed.

Parent: Should my child avoid certain foods or drinks?

Doctor: Yes, we recommend a low-fat diet with medium-chain triglycerides because these are absorbed differently and reduce the production of chyle. In some cases, we may temporarily stop oral feeding and provide nutrition intravenously.

Parent: Can this happen again? How can we prevent it?

Doctor: Recurrence is uncommon if the underlying cause is treated effectively. After surgery, careful monitoring helps catch any problems early. For congenital cases, ongoing follow-up is important. Preventing trauma and managing infections also help.

Parent: How long will it take for my child to get better?

Doctor: Most children improve within 3 to 4 weeks with conservative treatment. Some may take longer, especially if surgery is required. We will monitor your child closely to ensure the best outcome.

Parent: Thank you for explaining everything.

Doctor: You’re welcome. Please let us know if you have any questions or if your child’s symptoms change.

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**CONGENITAL DIAPHRAGMATIC HERNIA**

**DEFINITION AND DESCRIPTION**

Congenital diaphragmatic hernia (CDH) is a rare condition that happens in a baby before birth. It occurs early in pregnancy when a baby's diaphragm — the muscle that separates the chest from the abdomen — fails to close as it should. This leaves a hole in the diaphragm. The hole is called a hernia.

This hernia in the muscle of the diaphragm creates an opening between the abdomen and the chest. The intestines, stomach, liver and other abdominal organs may move through the hole into the baby's chest. If the intestines are in the chest, they don't develop the typical connections that hold them in place in the abdomen (malrotation). They may twist on themselves, cutting off their blood supply (volvulus).

In addition, the lung is small on the side of the diaphragm with the hernia, but the development of both lungs is affected. The air sacs (alveoli) inside the lungs don't develop as they should. This results in problems with blood flow and increased pressure inside the lung's blood vessels. The blood pressure in the lungs is higher than it should be, which can make it hard for the baby to breathe after birth. Some infants may also have problems with heart development.

Treatment of CDH depends on when the condition is found, how serious it is and whether there are problems with the heart.

**Causes**

In most cases, the cause of congenital diaphragmatic hernia is not known. In some cases, CDH can be linked to a genetic disorder or random gene changes called mutations. In these cases, the baby may have more issues at birth, such as problems with the heart, eyes, arms and legs, or stomach and intestines.

**Complications**

Complications that can occur with CDH include:

* Lung problems.
* Stomach, intestine and liver problems.
* Heart disease.
* Recurrent infections.
* Hearing loss.
* Changes in the shape of the chest and curve of the spine.
* Gastroesophageal reflux — stomach acid flowing back into the tube called the esophagus, which connects the mouth and stomach.
* Problems with growth and weight gain.
* Developmental delays and learning disabilities.
* Other problems present from birth.

**Symptoms**

Congenital diaphragmatic hernia ranges in severity. It may be mild and have few or no effects on the baby, or it can be more serious and affect the ability to bring oxygen to the rest of the body.

Babies born with CDH may have:

* Severe trouble breathing due to small lungs that don't work well (pulmonary hypoplasia).
* A type of high blood pressure that affects the arteries in the lungs and the right side of the heart (pulmonary hypertension).
* Problems with development of the heart.
* Damage to the intestines, stomach, liver and other abdominal organs if they move through the hernia into the chest.

### **When to see a doctor**

CDH may be found during a routine fetal ultrasound. Your health care provider can discuss treatment options with you.

## **Diagnosis**

Congenital diaphragmatic hernia is most often found during a routine fetal ultrasound exam that's done before your baby is born. A prenatal ultrasound exam uses sound waves to make images of your uterus and baby.

Occasionally, the diagnosis may not be made until after birth. Rarely, CDH may not be diagnosed until childhood or later. This may be because there are no signs or symptoms or because signs and symptoms such as respiratory and intestinal problems are mild.

Your health care provider uses prenatal ultrasound and other tests to track the growth and function of your baby's lungs, heart and other organs during your pregnancy.

### **Prenatal ultrasound**

Usually, you have your first fetal ultrasound during the first few months (first trimester) of your pregnancy. It confirms that you are pregnant and shows the number and size of your baby or babies.

Most often, you have another ultrasound during months four to six (second trimester) of your pregnancy. Your health care provider checks the growth and development of your baby. Your provider looks at the size and location of your baby's lungs, heart and other organs.

If your baby shows signs of CDH, your provider may have you get ultrasound exams more often. This can show how severe CDH is and whether it's getting worse.

### **Other tests**

More tests may be done to assess the function of your baby's organs. These may include:

* **Fetal magnetic resonance imaging (MRI).** This is a medical imaging technique that uses a magnetic field and computer-generated radio waves to create detailed images of the organs and tissues in the baby's body.
* **Fetal echocardiogram.** An echocardiogram uses sound waves to produce images of the baby's heart beating and pumping blood. The images from an echocardiogram can identify problems with the developing heart.
* **Genetic tests.** Genetic testing can identify genetic syndromes or other gene changes that are sometimes associated with CDH. Genetic counseling can help you understand these test results and give you more information about your baby's condition.

**Treatment**

Treatment of congenital diaphragmatic hernia depends on when the condition is found and how serious it is. Your health care team helps you decide what's best for you and your baby.

### **Care before delivery**

Your health care team watches you closely before your baby is born. You typically have ultrasounds and other tests often to check your baby's health and development.

An emerging treatment for severe CDH now being studied is called fetoscopic endoluminal tracheal occlusion (FETO). This surgery is done on your baby while you're still pregnant. The goal is to help the baby's lungs grow as much as possible before birth.

FETO is done in two procedures:

* **First procedure.** The first procedure happens early in the last few months (third trimester) of your pregnancy. Your surgeon makes a small incision in your abdomen and uterus. The surgeon inserts a special tube with a camera at the end, called a fetal endoscope, through your baby's mouth and into the windpipe (trachea). A small balloon is placed in your baby's trachea and inflated.

The natural uterine fluid during pregnancy, called amniotic fluid, flows in and out of your baby's lungs through the mouth. Inflating the balloon keeps amniotic fluid in your baby's lungs. The fluid expands the lungs to help them develop.

* **Second procedure.** After about 4 to 6 weeks, you have a second procedure. The balloon is removed so that your baby is ready to take air into the lungs after birth.

A special delivery method may be used if labor starts before the balloon has been removed and removal of the balloon with an endoscope is not possible. This method is called an ex utero intrapartum treatment (EXIT) procedure. The delivery is done by C-section with placental support. This means that your baby continues to get oxygen through the placenta before the umbilical cord is cut. Placental support continues until the balloon is out and a breathing tube is in place, allowing a machine to take over breathing.

FETO may not be the right choice for everyone. And there's no guarantee about the results of surgery. Your health care team evaluates you and your baby to see whether you may be candidates for this surgery. Talk to your team about the benefits and possible complications for you and your baby.

### **Care during delivery**

Usually, you can deliver your baby either vaginally or by C-section. You and your health care provider decide which method is best for you.

### **Care after delivery**

After birth, the health care team helps you plan treatment that meets your baby's needs. Your baby will likely be cared for in the newborn intensive care unit (NICU).

Your baby may need to have a breathing tube. The tube is attached to a machine that helps your baby breathe. This gives the lungs and heart time to grow and develop.

Babies who have life-threatening lung problems may need a treatment called extracorporeal membrane oxygenation (ECMO). This is also known as extracorporal life support (ECLS). The ECMO machine does the work of your baby's heart and lungs, allowing these organs to rest and heal.

How long your baby needs support to breathe depends on the response to treatment and other factors.

Most babies who have CDH have surgery to close the hole in the diaphragm. When this surgery takes place depends on your baby's health and other factors. Follow-up care to ensure the repair remains in place usually includes chest X-rays.

After leaving the hospital, your baby may need extra support. This can include supplemental oxygen. Oxygen is delivered by thin plastic tubing with prongs that fit into the nostrils or thin tubing connected to a mask worn over the nose and mouth. Feeding support may also be needed to help with growth and development. Medicine may be given for conditions associated with CDH, such as acid reflux or pulmonary hypertension.

Regular follow-up appointments with your child's health care provider can address any problems early.

## **Outlook / Prognosis**

The reported survival rate for babies born with CDH has improved. Between 7 and 9 out of every 10 babies survive. These babies are born critically ill, but for those who make it through the tense early days of their condition, the outlook gets better. Some children may have long-term complications, but they’ll still live long and full lives. Continuing advances in medicine improve the odds of both short-term survival and long-term health.

The longer-term prognosis depends on several factors, including:

* If your baby was born prematurely
* How severe your baby’s condition is at birth
* The size of the hernia
* What organs are involved
* Other health conditions

Babies who require breathing or feeding support for longer have a higher risk of ongoing complications, including chronic lung disease, growth failure, hearing loss and developmental delays. Your child’s provider will monitor them closely throughout their early life.

## **Diagnostic Considerations**

Special concerns

Using ultrasonography, congenital diaphragmatic hernia (CDH) may be prenatally diagnosed as early as the second trimester. [[15](javascript:void(0);)] Suggestive findings include polyhydramnios, an absent or intrathoracic stomach bubble, and mediastinal and cardiac shift. A detailed examination (level II ultrasonography) is typically necessary.

Prenatal diagnosis allows for chromosomal analysis and screening for other anomalies prior to the infant's birth. In addition, it allows the mother time to make important decisions about the pregnancy, including delivery in a facility with a neonatal ICU (NICU) that offers advanced respiratory support for the newborn infant.

Developing meaningful prognostic information before birth continues to be difficult. Some advocate for assessment of lung hypoplasia using ultrasound measurements of liver herniation into the thorax, lung to head ratios (LHR), or pulmonary artery to aorta ratios (modified McGoon index). MRI of the fetus is a promising technique that allows more precise measurement of the lung volume indexed to the body volume.

## **Differential Diagnoses**

* Bronchopulmonary Sequestration
* Cystic Adenomatoid Malformation
* Disorders of the Thoracic Cavity and Pleura
* Persistent Pulmonary Hypertension of the Newborn (PPHN)
* Pneumothorax Imaging

## **Epidemiology**

### International data

Congenital diaphragmatic hernia occurs in 1 of every 2000-3000 live births and accounts for 8% of all major congenital anomalies. The risk of recurrence of isolated (i.e., non-syndromic) congenital diaphragmatic hernia in future siblings is approximately 2%. Familial congenital diaphragmatic hernia is rare (< 2% of all cases), and both autosomal recessive and autosomal dominant patterns of inheritance have been reported. Congenital diaphragmatic hernia is a recognized finding in Cornelia de Lange syndrome and also occurs as a prominent feature of Fryns syndrome, an autosomal re

### Sex- and age-related demographics

Most studies report that congenital diaphragmatic hernia occurs equally in males and females.

Although congenital diaphragmatic hernia is usually a disorder of the newborn period, as many as 10% of patients may present after the newborn period and even during adulthood. A 2020 retrospective analysis of 2015-2018 data from the American College of Surgeons National Surgical Quality Improvement Program (ACS NSQIP) database found 110 adult patients with congenital diaphragmatic hernia who underwent surgical correction.

Outcome in patients with late presentation of congenital diaphragmatic hernia is extremely good, with low or no mortality. Recessive disorder with variable features, including diaphragmatic hernia, cleft lip or palate, and distal digital hypoplasia.

## **Procedures**

Endotracheal intubation and mechanical ventilation are required in all infants with severe congenital diaphragmatic hernia who present in the first hours of life. If the diagnosis is known at the time of delivery, avoid bag-and-mask ventilation in the delivery room because the stomach and intestines become distended with air and further compromise pulmonary function. A nasogastric tube should be placed as soon as possible to provide intestinal decompression.

As discussed in Treatment, the goal is to adequately expand the lung but to avoid overdistension; therefore, inspiratory pressures should be kept as low as possible. Consider the use of high-frequency ventilation (HFV) if high inspiratory pressures are required.

Place an indwelling catheter in the umbilical artery or in a peripheral artery (radial, posterior tibial) for continuous blood pressure and frequent ABG monitoring.

Place a venous catheter via the umbilical vein to allow for administration of inotropic agents and hypertonic solutions such as calcium gluconate. If the liver is in the chest, the catheter will likely not pass through the ductus venosus, and another route must be considered for central venous access.

The use of HFV in congenital diaphragmatic hernia remains controversial, and no randomized studies indicate a clear benefit. However, HFV may allow for use of lower ventilator pressures and may help normalize PaCO2. Mean airway pressures should be carefully adjusted to avoid lung overdistension. Frequent radiograph (with a goal of 8-9 rib expansion of the contralateral lung) may help in the ongoing assessment and optimization of lung expansion.

Venoarterial or venovenous ECMO support is an adaptation of cardiopulmonary bypass and involves a surgical team; insertion of catheters into the internal jugular vein, internal carotid artery, or both; systemic heparinization; and oxygenation through the use of an artificial membrane lung. Because of its complexity and resource expense, ECMO is available at fewer than 100 centers in the United States. The overall survival rate for infants with congenital diaphragmatic hernia reported to the international Extracorporeal Life Support Organization (ELSO) registry is approximately 52%, which is the lowest rate in all the neonatal conditions treated with ECMO. Although no conclusive evidence shows that ECMO improves survival or outcome for infants with congenital diaphragmatic hernia, it remains a commonly used therapy for severely affected infants.

**Pediatric Congenital Diaphragmatic Hernia (CDH) Treatment: Drug Information and Side Effects**

## 1. Pulmonary Hypertension Management

* Inhaled Nitric Oxide (iNO)
  + *Purpose:* Selective pulmonary vasodilator to reduce pulmonary artery pressure and improve oxygenation.
  + *Side Effects:* Hypotension, methemoglobinemia, rebound pulmonary hypertension on withdrawal.
* Prostaglandin E1 (PGE1)
  + *Purpose:* Maintains ductus arteriosus patency to improve systemic oxygen delivery in severe pulmonary hypertension.
  + *Side Effects:* Apnea, hypotension, fever, flushing.
* Phosphodiesterase Inhibitors (e.g., Sildenafil)
  + *Purpose:* Oral or intravenous pulmonary vasodilators used adjunctively.
  + *Side Effects:* Hypotension, headache, flushing, gastrointestinal upset.
* Milrinone
  + *Purpose:* Inodilator improving cardiac contractility and pulmonary vasodilation.
  + *Side Effects:* Hypotension, arrhythmias, thrombocytopenia.

## 2. Ventilation and Sedation

* Muscle Relaxants (e.g., Vecuronium, Cisatracurium)
  + *Purpose:* Facilitate mechanical ventilation by reducing patient-ventilator asynchrony and preventing high airway pressures.
  + *Side Effects:* Prolonged paralysis, hypotension, risk of critical illness myopathy.
* Sedatives and Analgesics
  + *Opioids (e.g., Morphine, Fentanyl)*
    - For pain control and sedation.
    - Side effects: Respiratory depression, hypotension, constipation.
  + *Acetaminophen*
    - Used adjunctively for analgesia with fewer side effects.
  + *Benzodiazepines (e.g., Midazolam)*
    - For sedation; side effects include respiratory depression and hypotension.
* Recent protocols emphasize opioid-sparing strategies with standing IV acetaminophen to reduce opioid exposure and related side effects.

## 3. Cardiovascular Support

* Inotropes and Vasopressors (e.g., Dopamine, Dobutamine, Norepinephrine)
  + Used to maintain adequate systemic blood pressure and cardiac output.
  + Side effects: Tachycardia, arrhythmias, hypertension, peripheral ischemia.

## 4. Other Supportive Medications

* Diuretics (e.g., Furosemide)
  + To manage fluid overload and pulmonary edema.
  + Side effects: Electrolyte imbalance, dehydration, nephrotoxicity.
* Antibiotics
  + Used prophylactically or to treat infections; choice depends on clinical scenario.

**Types of Genetic Causes**

* + Chromosomal abnormalities: Aneuploidies and structural chromosomal variants are common. For example, deletions or duplications at regions such as 15q26, 8p23.1, 8q23, 5p15.2, 16p11.2, 17q12, and 1q41-42 have been associated with CDH.
  + Single-gene mutations: Mutations in genes involved in diaphragm development and related pathways have been identified. Examples include LONP1, MYRF, and genes affecting retinoid (vitamin A) signaling pathways.
  + Syndromic associations: CDH occurs as part of several genetic syndromes, including:
    - Pallister-Killian syndrome
    - Fryns syndrome
    - Wolf-Hirschhorn syndrome
    - Cornelia de Lange syndrome
    - Donnai-Barrow syndrome
    - Simpson-Golabi-Behmel syndrome
    - CHARGE syndrome
    - Denys-Drash syndrome
    - Apert syndrome
    - Beckwith-Wiedemann syndrome
    - 22q11.2 deletion syndrome
* Genetic Mechanisms:  
  CDH results from disruptions in embryonic diaphragm formation, involving multiple genes that regulate cell migration, proliferation, and differentiation. Many mutations affect transcription factors, cell structural proteins, or signaling pathways critical for diaphragm and lung development.
* Inheritance Patterns:  
  Most isolated CDH cases are not inherited and occur sporadically. When CDH is part of a syndrome, inheritance follows the pattern of that syndrome (autosomal dominant, recessive, or chromosomal). Familial recurrence is rare but possible.

## **PREDEFINED QUESTIONS AND ANSWERS**

## 1. What is congenital diaphragmatic hernia (CDH)?

CDH is a birth defect where there is a hole in the diaphragm, allowing abdominal organs like the stomach, intestines, or liver to move into the chest. This can prevent the lungs from developing properly, leading to breathing difficulties after birth.

## 2. How is CDH diagnosed?

CDH is often diagnosed before birth during a routine prenatal ultrasound. After birth, chest X-rays and other imaging tests confirm the diagnosis. Sometimes it is diagnosed after birth if the baby has breathing problems.

## 3. What causes CDH?

The exact cause is unknown but it occurs during fetal development when the diaphragm does not close completely. Some cases are associated with genetic syndromes or chromosomal abnormalities.

## 4. What are the treatment options for CDH?

Treatment usually involves:

* Stabilizing the baby after birth with respiratory support, including oxygen or mechanical ventilation.
* In severe cases, extracorporeal membrane oxygenation (ECMO) may be used to support the lungs and heart.
* Surgical repair to move the abdominal organs back into the abdomen and close the diaphragm defect, usually after the baby is stable.

## 5. What is the outlook for babies with CDH?

Outcomes vary depending on the severity of lung underdevelopment and other complications. Many babies survive and go on to lead healthy lives, especially with specialized care and follow-up.

## 6. What kind of follow-up care is needed?

Long-term follow-up is important to monitor lung function, growth, development, and potential complications like asthma, feeding difficulties, or scoliosis. Specialized multidisciplinary programs provide ongoing care.

## 7. Can CDH be detected or treated before birth?

Yes, prenatal ultrasounds can detect CDH. In some severe cases, fetal interventions like fetoscopic endoluminal tracheal occlusion (FETO) may be offered to help the lungs grow before birth.

## 8. Will my child have developmental delays?

Some children with CDH may experience developmental delays or learning difficulties, but many develop normally. Early intervention and therapies can help support development.

## 9. How can families prepare for the birth of a baby with CDH?

Families are encouraged to work closely with a specialized fetal care team to plan delivery at a center experienced in CDH care. Support services and counseling are important throughout.

**DOCTOR PATIENT CONVERSATION**

Doctor: Hello, I’d like to talk with you about your baby’s diagnosis of congenital diaphragmatic hernia, or CDH. Have you heard about this condition before?

Parent: Not really. What exactly is it?

Doctor: CDH is a birth defect where there is a hole in the diaphragm, the muscle that separates the chest from the abdomen. Because of this hole, some abdominal organs like the stomach or intestines can move into the chest and affect lung development. This can make breathing difficult after birth.

Parent: How did this happen? Did we do something wrong?

Doctor: No, this happens very early during fetal development, and it’s not caused by anything you did. Sometimes it’s related to genetic factors, but often the exact cause is unknown.

Parent: What will happen now? How do you treat it?

Doctor: After birth, we will support your baby’s breathing with oxygen or a ventilator if needed. In severe cases, we might use a machine called ECMO to support the heart and lungs temporarily. Once your baby is stable, we plan to do surgery to move the organs back into the abdomen and close the hole in the diaphragm.

Parent: Is surgery risky? Will my baby be okay?

Doctor: Surgery does carry risks, but it’s necessary to help your baby breathe better. The timing depends on how stable your baby is. Many babies do very well with surgery and specialized care, but some may have ongoing challenges. We’ll monitor your baby closely and provide all the support needed.

Parent: How long will my baby stay in the hospital?

Doctor: It varies depending on the severity, but babies with CDH often need several weeks to months in the hospital, including time for breathing support and recovery after surgery.

Parent: Will my child have problems later in life?

Doctor: Some children may have issues like asthma, feeding difficulties, or developmental delays, but many grow up healthy and active. We’ll arrange follow-up care to monitor and support your child’s growth and development.

Parent: Is there anything I should do to prepare?

Doctor: We encourage you to ask questions and share any concerns. We’ll help you understand each step and connect you with support services. Planning delivery at a specialized center with experienced teams is important.

Parent: Thank you for explaining. It helps to know what to expect.

Doctor: You’re welcome. We’re here to support you and your baby throughout this journey. Please feel free to reach out anytime with questions or concerns.

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**CONGENITAL LOBAR EMPHYSEMA**

**DEFINITION AND DESCRIPTION**

Congenital lobar emphysema (CLE) is a rare lung malformation that occurs when air become trapped in the lungs. Infants with CLE have one or more lobes of the lung that are hyperinflated.

An obstruction of one of the main airways of the lungs — called the bronchi — can cause CLE in some newborns. However, experts suggest the condition may have a genetic link. The severity of CLE can vary among people, and some may not experience symptoms until adulthood.

Doctors typically diagnose the condition in the first 6 months of an infant’s life. However, CLE is very rare, occurring in about 1 in 20,000 to 30,000 births. About a third of newborns with CLE have symptoms at birth.

**CAUSES**

The exact cause of CLE is not always clear — around 50% of cases occur without doctors knowing the cause.

Some infants with CLE have issues with cartilage formation in the bronchi, the passageways into the lungs. This can lead to issues with lung development — they may either under develop or develop atypically — which may cause air to stay trapped in the lungs.

Other possible causes of CLE include:

* diseases of the parenchyma, the cells responsible for lung function
* a blockage from inside the bronchi
* a blockage from outside the bronchi

**Signs and symptoms**

Around 50% of newborns with CLE have symptoms at birth or within the first 6 months of life.

Symptoms may include:

* blue or gray tinge to the skin due to a lack of oxygen
* the chest sinking below the neck or breastbone during breathing
* wheezing
* difficulty feeding or breathing
* frequent respiratory tract infections
* chronic cough

## **What other conditions have similar symptoms?**

Some conditions with similar symptoms to CLE include:

* bronchial asthma
* pneumonia
* respiratory distress syndrome in infants
* cor pulmonale, or altered function in the right side of the heart that can occur following respiratory disease
* alpha-1 antitrypsin deficiency, a genetic condition that affects the liver and lungs
* tension pneumothorax, when air becomes trapped between the lung and chest wall
* other congenital diseases, such as congenital diaphragmatic hernia or congenital cystic adenomatoid malformation
* pneumatocele when air pockets or cysts develop within the lung parenchyma following injury or trauma
* lung hypoplasia, another rare, congenital condition of the lungs

Considering the above when diagnosing CLE helps doctors determine the best treatment plan.

## **Diagnosis**

Diagnosing CLE involves a physical exam of the lungs and medical imaging.

Part of the physical exam involves the doctor tapping the chest to hear sounds within the lungs. The doctor may notice hyper-resonance, which involves a low-pitched, booming sound upon examination. This typically indicates that a particular area of the lung is masking respiratory sounds.

Doctors regard a lung CT as the best method for imaging and diagnosing CLE. Another imaging technique that can provide valuable information is an MRI. In infants, MRI can provide detailed images that could potentially evaluate and identify abnormalities.

An ultrasound during pregnancy cannot diagnose CLE. In some cases, ultrasound experts may still notice lung areas with an exaggerated response to sound waves with typical blood flow in the area.

Doctors could also confirm a diagnosis before birth with MRI since it can show more detail in the fetal lung.

### **Differential diagnosis**

Before treating CLE, doctors need to rule out other conditions that may be causing similar symptoms. Many different respiratory diseases may resemble CLE, as mentioned in the section above.

A chest X-ray showing hyperinflated lung lobes can suggest several diagnoses:

* pneumothorax, or collapsed lung
* pneumatocele
* lung hypoplasia

Performing a differential diagnosis is the process doctors use to make a final diagnosis. They collect data, consider the most likely cause, and eliminate other less likely options.

**Treatment**

Once a doctor establishes a diagnosis of CLE, they will choose the most appropriate treatment. These strategies can depend on the severity of the disease.

### **Conservative treatment**

For mild to moderate disease, doctors prescribe conservative treatments, such as oxygen and bronchodilators, to help open the airways and improve breathing.

Follow-up appointments with specialists, such as a respirologist, may be beneficial. Overall conservative treatment can provide favorable results for some.

### **Surgery**

In severe CLE, newborns may require surgery to remove the lung lobe with CLE. Doctors refer to the removal of a lung lobe as a lobectomy, which typically provides positive results for newborns with severe CLE.

**Outlook**

With appropriate treatment, the outlook for CLE is generally favorable

Sometimes, the condition is so severe it can affect the cardiovascular system, which may affect the prognosis. However, treatments may improve both respiratory and cardiac problems.

**Epidemiology of CLE:**

* Incidence and Prevalence:  
  CLE is a rare congenital lung malformation, estimated to occur in approximately 1 in 20,000 to 1 in 30,000 live births. It accounts for about 5% of all congenital cystic lung lesions.
* Age at Presentation:  
  Most cases present in early infancy, often within the first six months of life, with respiratory distress or recurrent respiratory infections. Some mild cases may be diagnosed later or incidentally.
* Sex Distribution:  
  CLE shows a male predominance, with a male-to-female ratio reported around 3:1 in some series.
* Lobar Involvement:  
  The left upper lobe is most commonly affected, followed by the right middle lobe and right upper lobe.
* Associated Anomalies:  
  CLE may occasionally be associated with congenital heart defects or other congenital anomalies, but most cases are isolated.
* Prognosis:  
  With early diagnosis and appropriate management, prognosis is generally good. Surgical lobectomy is curative in symptomatic cases.

## **PREDEFINED QUESTIONS AND ANSWERS**

## What is congenital lobar emphysema (CLE)?

CLE is a rare lung condition present at birth where one or more lobes of the lung become overinflated with air. This causes the affected lobe to expand abnormally, compressing the remaining lung tissue and making breathing difficult.

## 2. How common is CLE and when does it usually present?

CLE is uncommon, occurring in about 1 in 20,000 to 30,000 live births. Most infants show symptoms within the first 6 months of life, often presenting with respiratory distress.

## 3. What causes CLE?

The exact cause is often unknown. It is thought to result from partial obstruction of the lobar bronchus due to intrinsic factors like bronchomalacia or extrinsic compression by blood vessels or cysts. About 50% of cases have no identifiable cause.

## 4. What symptoms does CLE cause?

Common symptoms include:

* Rapid breathing or respiratory distress
* Wheezing and coughing
* Cyanosis (bluish skin)
* Difficulty feeding

## 5. How is CLE diagnosed?

Diagnosis is made through:

* Chest X-ray showing hyperinflation of the affected lobe and mediastinal shift
* Chest CT scan to confirm the diagnosis and rule out other conditions
* Sometimes prenatal ultrasound detects lung abnormalities, but diagnosis is usually postnatal

## 6. What are the treatment options?

* Mild or moderate cases: May be managed conservatively with close observation, oxygen, and bronchodilators if needed. Some asymptomatic cases may improve without surgery.
* Severe or progressive cases: Surgical removal of the affected lobe (lobectomy) is the standard treatment and usually results in excellent outcomes.

## 7. What are the risks of misdiagnosis?

CLE can be mistaken for pneumothorax or other lung conditions, leading to inappropriate treatments like chest tube insertion, which can worsen the condition.

## 8. What is the prognosis after treatment?

With appropriate management, especially surgical lobectomy in symptomatic patients, the prognosis is generally very good. Most children recover fully without long-term lung problems.

## 9. Can CLE affect other parts of the body?

In about 10–15% of cases, CLE may be associated with cardiovascular abnormalities, so cardiac evaluation is often recommended.

## 10. Is CLE hereditary or preventable?

CLE is usually sporadic with no known hereditary pattern and cannot be prevented.

**DOCTOR PATIENT CONVERSATION**

Doctor: Hello, I want to talk with you about your baby’s lung condition called congenital lobar emphysema, or CLE.

Parent: What exactly is that?

Doctor: CLE is a rare condition where one of the lobes of the lung becomes overinflated with air. This happens because the airway to that lobe is partially blocked or narrowed, causing air to get trapped. This overinflation can press on the rest of the lung and make it harder for your baby to breathe.

Parent: How did this happen? Is it something we caused?

Doctor: No, CLE is a congenital condition, meaning it develops before birth. It’s usually due to abnormal development of the airways or surrounding tissues. It’s not caused by anything you did during pregnancy.

Parent: What symptoms should we expect?

Doctor: Babies with CLE often have fast or difficult breathing, wheezing, or coughing. Sometimes they have bluish skin from lack of oxygen or trouble feeding. Symptoms usually appear in the first few weeks or months of life.

Parent: How do you diagnose it?

Doctor: We use chest X-rays and sometimes CT scans to see the overinflated lobe and how it’s affecting the rest of the lungs. These images help us confirm the diagnosis and plan treatment.

Parent: What treatment does my baby need?

Doctor: If your baby has mild symptoms, we may watch closely and provide supportive care like oxygen. For babies with more severe breathing problems, surgery to remove the affected lobe, called a lobectomy, is usually recommended. Surgery often leads to excellent improvement.

Parent: Is surgery risky?

Doctor: Like any surgery, there are risks, but lobectomy for CLE is generally safe and effective. It can be lifesaving if the overinflated lobe is causing serious breathing difficulties.

Parent: What is the outlook after treatment?

Doctor: Most babies do very well after surgery and have normal lung function as they grow. Even babies managed without surgery often improve over time.

Parent: Can this happen again or affect other parts of the body?

Doctor: CLE usually affects only one lobe and does not recur after surgery. Sometimes it’s associated with heart defects, so we may do heart evaluations as well.

Parent: What should we watch for at home?

Doctor: Watch for signs of breathing difficulty like rapid breathing, grunting, blue lips, or poor feeding. If you notice these, seek medical attention promptly.

Parent: Thank you for explaining everything so clearly.

Doctor: You’re welcome. We’re here to support you and your baby. Please feel free to ask any questions anytime.

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## **Diaphragmatic Paralysis**

**DEFINITION AND DESCRIPTION**

The **diaphragm** is a dome-shaped muscle separating the thoracic and abdominal cavities. It plays a crucial role in the respiratory system, in particular with **breathing**. During inhalation, it contracts and flattens, increasing the volume of the chest cavity and allowing the lungs to expand and fill with air. During exhalation, the diaphragm relaxes and returns to its dome shape, resulting in the expulsion of air from the lungs. It accounts for approximately 70-80% of the effort required for breathing under normal conditions.

Diaphragmatic paralysis is a condition characterized by the impairment or loss of function in the diaphragm, the primary muscle responsible for respiration. It can be bilateral or unilateral, with **unilateral diaphragmatic paralysis** being more common. It can lead to a significant reduction in lung capacity and respiratory efficiency. Resulting symptoms include shortness of breath, especially when lying down, reduced exercise tolerance, and, in severe cases, respiratory challenges.

## **Causes of Diaphragmatic Paralysis**

Understanding the causes of diaphragmatic paralysis is essential for accurate diagnosis and effective management of affected individuals.

### **Neurological Injuries**

* Diaphragmatic paralysis can arise from various neurological causes, with nerve damage being a primary factor. The **phrenic nerve**, which originates from the cervical spinal cord (C3-C5), plays a crucial role in innervating the diaphragm. Damage to this nerve can result in impaired diaphragmatic function or paralysis.
* Common **causes of phrenic nerve injury** include surgical complications, particularly from procedures involving the neck or chest, such as cardiac surgery or neck dissections.
* Trauma, including blunt or penetrating injuries to the neck or upper chest, can also lead to phrenic nerve damage.

### **Neurological Conditions**

* Certain medical conditions can also affect the **phrenic nerve**.
* Tumors or masses in the neck or mediastinum can compress the nerve, leading to dysfunction.
* Neurological disorders such as **amyotrophic lateral sclerosis** (ALS) or **multiple sclerosis** (MS) can also result in diaphragmatic paralysis.

### **Viral Infections**

* **Infections**, such as viral neuropathies, have also been reported as possible causes.
* Herpes zoster, Zika, poliovirus, and other viral infections have all been linked to diaphragmatic paralysis, particularly unilateral diaphragmatic paralysis.

### **Autoimmune Conditions**

* **Guillain-Barré syndrome** is an autoimmune attack on the nervous system, which has been linked to diaphragmatic paralysis

### **Muscular Disorders**

* Diseases that affect muscle function, such as muscular dystrophy, can lead to diaphragmatic weakness or paralysis.
* These genetic conditions, which include Duchenne and **Becker muscular dystrophies**, cause progressive muscle degeneration, including the diaphragm.
* Over time, respiratory function may be significantly compromised, necessitating ventilatory support.

In approximately **20% of cases**, the cause of diaphragmatic paralysis is unknown (**idiopathic diaphragm paralysis**). In these instances, the underlying reason for the paralysis remains unknown despite extensive investigation. Idiopathic diaphragmatic paralysis can pose diagnostic and management challenges, as the absence of a clear etiology makes targeted treatment difficult.

## **Symptoms and Diagnosis**

Diaphragmatic paralysis can present with a range of symptoms, primarily affecting respiratory function. **Common symptoms** include:

* Shortness of Breath (dyspnea): This is often more pronounced when lying flat due to the upward pressure of abdominal contents on the paralyzed diaphragm, reducing lung capacity.
* Difficulty Breathing: Patients may struggle to breathe, especially during physical activity.
* **Fatigue**: Reduced oxygenation due to impaired lung function can lead to a feeling of tiredness and decreased exercise tolerance.
* Sleep Disturbances: Breathing difficulties may worsen during sleep, leading to conditions such as **sleep apnea** or **insomnia**.
* Elevated Hemidiaphragm: This may be observed on physical examination or imaging, as the affected side of the diaphragm moves abnormally or not at all.
* **Recurrent Pneumonia**

**DIAGNOSIS**

The diagnostic process for diaphragmatic paralysis involves a combination of clinical evaluation and imaging studies. **Chest X-rays** and ultrasound are commonly used to visualize the position and movement of the diaphragm. These imaging tests can reveal elevation of the affected hemidiaphragm and reduced or paradoxical movement during respiration.

**Pulmonary function tests** (PFTs) are also crucial in assessing the impact of diaphragmatic paralysis on respiratory capacity and function. PFTs measure the overall performance of the lungs and can indicate reduced lung capacity due to diaphragmatic dysfunction.

Specialized tests like the sniff test may be done. A **sniff test** is a fluoroscopic examination that observes the movement of the diaphragm during a forced sniff. Paralysis is indicated by paradoxical upward movement during a sharp inhalation.

**Phrenic nerve stimulation studies** may be done. These assess the integrity and function of the phrenic nerve and can provide further diagnostic information. In some cases, **electromyography (EMG) of the diaphragm** may be performed to evaluate muscle and nerve function.

## **Treatment Options for Diaphragmatic Paralysis**

The treatment for diaphragmatic paralysis depends on the severity of the condition and the underlying cause. In asymptomatic or mild cases, observation and regular monitoring may be sufficient, especially if the paralysis is expected to be temporary or if it does not significantly impact respiratory function. Patients should undergo periodic evaluations, including pulmonary function tests and imaging studies, to monitor the progression of the condition.

**Physical therapy** plays a crucial role in the management of diaphragmatic paralysis. Respiratory therapists can guide patients through exercises designed to strengthen the accessory muscles of respiration, thereby compensating for diaphragmatic weakness. **Breathing techniques**, such as diaphragmatic breathing and **pursed-lip breathing**, can also help improve ventilation and oxygenation.

For more severe cases of diaphragmatic paralysis, surgical treatments may be considered. **Diaphragm plication** is a procedure that involves folding and suturing the weakened portion of the diaphragm to reduce its volume and increase its tension. This can improve lung expansion and overall respiratory function. In cases where the phrenic nerve is damaged, phrenic nerve repair or stimulation may be attempted to restore diaphragmatic function.

Respiratory support is often necessary for patients with significant breathing difficulties. **Mechanical ventilation** may be required in acute settings or for those with severe respiratory challenges. Non-invasive ventilation, such as **continuous positive airway pressure** (CPAP) or **bilevel positive airway pressure** (BiPAP), can be used to support breathing in less severe cases or as a long-term management strategy.

## **Living with Diaphragmatic Paralysis**

Living with diaphragmatic paralysis requires adjustments and careful management to maintain quality of life and respiratory health. Patients should engage in **regular physical activity** tailored to their capacity, as exercise can improve cardiovascular health and strengthen respiratory muscles.

**Breathing exercises**, such as diaphragmatic breathing and pursed-lip breathing, are essential for enhancing lung function and ensuring efficient ventilation. These exercises can be practiced daily and incorporated into routine activities.

Lifestyle adjustments, such as avoiding heavy meals that can press against the diaphragm and **maintaining a healthy weight** to reduce respiratory strain, are beneficial. Sleeping positions may also need modification, with some individuals finding relief in sleeping with the **head elevated** to ease breathing.

Regular **medical follow-ups** are crucial for monitoring respiratory function and adjusting treatment as necessary. PFTs, imaging studies, and consultations with respiratory therapists can provide valuable insights into the management of diaphragmatic paralysis. Patients should also be vigilant for signs of **respiratory infections** or complications and seek medical attention as needed.

## **Emerging Research and Therapies**

Emerging research and therapies for diaphragmatic paralysis are focused on innovative approaches to restore diaphragm function and enhance nerve regeneration. One area of interest is the use of **diaphragmatic pacing**, a technique that involves electrical stimulation of the phrenic nerve to induce diaphragm contractions. This method has shown promise in improving respiratory function in patients with diaphragmatic paralysis, especially in those with spinal cord injuries or neurodegenerative diseases.

Another promising avenue is the field of regenerative medicine, where researchers are exploring the potential of **stem cell therapy** and growth factors to repair or regenerate damaged phrenic nerves. Animal studies have demonstrated the ability of certain stem cells to promote nerve regeneration and improve diaphragmatic function. While still in the experimental stage, these findings offer hope for clinical applications in humans.

Advancements in surgical techniques, such as the use of **minimally invasive techniques** for diaphragm plication, and new procedures, such as **nerve transfer surgery**, are also being used more commonly. Nerve transfer surgery involves rerouting healthy nerves to the phrenic nerve to restore diaphragm function.

Early results have been encouraging, with some patients experiencing significant improvements in respiratory function. Additionally, research is being conducted on the use of **biomaterials and scaffolds** to support nerve regeneration and diaphragm repair. These materials aim to provide a conducive environment for nerve growth and tissue healing, potentially enhancing the recovery of diaphragmatic function.

## **Diagnostic Considerations**

The following diagnoses may be difficult to differentiate from bilateral diaphragmatic paralysis (BDP):

* Diaphragmatic relaxation can occur in which the muscles are thin but no injury is seen to the nerves
* Alveolar hypoventilation is caused by brainstem or high cervical spine disease; patients have normal respiratory muscle strength and can voluntarily hyperventilate to lower partial pressure of carbon dioxide in arterial blood (PaCO2)
* Anterior horn cells and neuromuscular junction diseases may be difficult to differentiate from phrenic nerve dysfunction

## **Epidemiology**

The exact incidence of diaphragmatic paralysis has not been defined. However, like diaphragm eventration, it is known to be more common among males

## **Prognosis**

### Unilateral diaphragmatic paralysis

Depending on the etiology of the diaphragmatic paralysis, the prognosis for patients with UDP is usually excellent unless significant underlying pulmonary disease is present. Patients develop compensatory mechanisms, and those with phrenic injuries may recover fully or partially.At times, patients may spontaneously recover from idiopathic disease. Patients who do not recover from UDP generally lead relatively normal lives. In this group, dyspnea may develop with exertion, leading to increased ventilatory demands.

The morbidity of UDP is mainly based on the underlying pulmonary functional status and the etiology of the paralysis.Because most cases of UDP are found incidentally during imaging studies, many patients have no symptoms. Diaphragmatic paralysis is more likely to affect the left hemidiaphragm. The patients with UDP who symptoms and decreased quality of life do have are those who have concurrent underlying lung diseases.

### Bilateral diaphragmatic paralysis

The prognosis for patients with BDP depends on the nature of the underlying disease. These patients are usually symptomatic. When symptoms are severe or underlying lung pathology is present, patients may develop ventilatory failure without medical intervention. Some patients may recover diaphragmatic function if nerve injury is not permanent. Other patients, however, may require long-term treatment. If recovery occurs, it usually takes considerable time (>1 y).

**PREDEFINED QUESTION AND ANSWER**

## 1. What is diaphragmatic paralysis?

Diaphragmatic paralysis is a condition where one or both sides of the diaphragm lose their ability to contract effectively, leading to impaired breathing. It can be unilateral (one side) or bilateral (both sides).

## 2. What causes diaphragmatic paralysis?

Common causes include:

* Injury or trauma to the phrenic nerve (e.g., during surgery or chest trauma)
* Neurological diseases affecting the diaphragm muscle or its nerve supply
* Infections, tumors, or inflammatory conditions
* Congenital abnormalities (rare)
* Idiopathic causes

## 3. What are the symptoms of diaphragmatic paralysis?

* Unilateral paralysis: Often asymptomatic or mild symptoms such as shortness of breath on exertion or when lying down.
* Bilateral paralysis: More severe symptoms including difficulty breathing at rest, orthopnea (difficulty breathing when lying flat), fatigue, disturbed sleep, recurrent pneumonia, and respiratory failure in severe cases.

## 4. How is diaphragmatic paralysis diagnosed?

* Imaging studies such as chest X-ray, ultrasound, fluoroscopy, CT, or MRI can show elevated hemidiaphragm or reduced diaphragm movement.
* Pulmonary function tests show decreased lung volumes, especially when lying down.
* Electromyography (EMG) and phrenic nerve stimulation tests assess nerve and muscle function.
* Arterial blood gas analysis may show low oxygen or high carbon dioxide levels in advanced cases.

## 5. What treatment options are available?

* Conservative management: Includes respiratory support, physical therapy, and monitoring if symptoms are mild.
* Mechanical ventilation: May be needed in severe bilateral paralysis.
* Surgical options:
  + Diaphragm plication: Tightening the paralyzed diaphragm to improve lung expansion.
  + Diaphragmatic pacing: Electrical stimulation of the phrenic nerve if it is intact.
* Treatment choice depends on severity, underlying cause, and patient condition.

## 6. What is the prognosis?

* Prognosis varies; unilateral paralysis often has a good outcome with minimal symptoms.
* Bilateral paralysis can be life-threatening without intervention but may improve with treatment.
* Recovery depends on the cause; nerve injury may recover over time, while muscle damage may be permanent.

## 7. Can diaphragmatic paralysis be prevented?

* Prevention focuses on avoiding phrenic nerve injury during surgery or trauma.
* Early diagnosis and management of underlying conditions may reduce complications.

**DOCTOR PATIENT CONVERSATION**

Doctor: Hello, I want to discuss the results of your recent tests. They show that you have diaphragmatic paralysis, which means one side of your diaphragm muscle is not moving properly.

Patient: What causes this? How did it happen?

Doctor: Diaphragmatic paralysis often happens because of injury or damage to the phrenic nerve, which controls the diaphragm muscle. This can occur after surgery, trauma, infections, or sometimes from neurological conditions. In your case, it may be related to your previous injury/surgery.

Patient: What symptoms should I expect? I’ve been feeling short of breath.

Doctor: Yes, shortness of breath, especially when lying flat (called orthopnea), is common. You may also notice fatigue, chest discomfort, or difficulty with exercise. If both sides are affected, symptoms can be more severe and may lead to respiratory failure.

Patient: How do you confirm this diagnosis?

Doctor: We use several tests. Chest X-rays and ultrasound can show an elevated or immobile diaphragm. A special test called the sniff test under fluoroscopy or ultrasound shows diaphragm movement. The gold standard is electromyography (EMG) of the diaphragm and phrenic nerve.

Patient: What treatments are available?

Doctor: Treatment depends on severity. For mild cases, we monitor and support breathing. If symptoms are severe, we may recommend non-invasive ventilation at night to help you breathe better during sleep. If paralysis persists beyond 6 to 12 months and symptoms remain significant, surgery called diaphragm plication can be done to tighten the diaphragm and improve lung function. In some cases, phrenic nerve pacing may be an option.

Patient: Can this get better on its own?

Doctor: Sometimes, especially if caused by nerve injury, partial or full recovery can occur over months to a year or more. We will monitor your progress closely.

Patient: What should I watch for?

Doctor: Watch for worsening shortness of breath, especially when lying down, increased fatigue, or difficulty sleeping. If you develop any new symptoms, let us know promptly.

Patient: Thank you for explaining. What’s the next step?

Doctor: We’ll continue monitoring your breathing and may start you on nighttime ventilation if needed. We’ll also schedule follow-ups to assess recovery and discuss surgery if symptoms don’t improve.

REFERENCES

[Diaphragmatic Paralysis Treatment & Management: Approach Considerations, Unilateral Diaphragmatic Paralysis, Bilateral Diaphragmatic Paralysis](https://emedicine.medscape.com/article/298200-treatment)

### **Coughing up blood (hemoptysis)**

**DEFINITION AND DESCRIPTION**

Coughing up blood involves coughing or spitting up blood or bloody mucus from your lower respiratory tract (lungs and throat). Also called hemoptysis (pronounced “he-MOP-tih-sis”), coughing up blood is common and can have many causes. Most causes aren’t serious. However, you may need to visit an ER immediately if you’re coughing up large amounts of blood.

Blood that you cough up often looks bubbly or frothy and is mixed with mucus or spit. It can appear pink, red or rust-colored and is usually in small amounts.

#### **Coughing up blood and vomiting blood**

Coughing up blood (hemoptysis) isn’t the same as vomiting blood (hematemesis). Blood that’s coughed up usually looks like blood-stained spit mixed with mucus. The blood comes from your throat or mouth. Vomiting blood involves spewing large quantities of blood. It usually involves internal bleeding in your upper gastrointestinal (GI) tract.

See a healthcare provider immediately if you’re vomiting blood.

It can be. It all depends on what’s causing your blood loss and the extent of your blood loss. Most causes aren’t serious and are treatable. Still, coughing up blood can be a sign of serious conditions, like a severe infection or lung cancer. Losing too much blood at once can be life-threatening and require emergency medical attention.

Only a healthcare provider can determine how serious your condition is. If you’re coughing up large quantities of blood, or if your condition doesn’t improve, see a provider.

## **Possible Causes**

Causes range from mild (most common) to serious and potentially life-threatening. Usually, coughing up blood is related to an infection. The most common causes include:

* Bronchitis.
* Pneumonia.
* Tuberculosis.

Other causes include:

* Bronchiectasis.
* Cystic fibrosis.
* Vasculitis.
* Drug use (crack/cocaine).
* Complications from lupus.
* Congestive heart failure.
* Injury to the arteries in your lungs.
* Irritation from a bronchoscopy biopsy.
* Using blood thinners (anticoagulants).
* Excessive coughing that irritates your throat.
* Blood clot in your lung (pulmonary embolism).
* Chronic obstructive pulmonary disease (COPD).
* Pulmonary aspiration (breathing blood into your lungs).
* Lung cancer (especially in people over 40 who smoke).
* Blood entering your throat from your nose (nosebleed), tonsils or teeth.
* Foreign body or obstruction in your airway (more common among children).

## **Care and Treatment**

Your healthcare provider will review your medical history, perform a physical exam and ask questions to determine what’s causing you to cough up blood. They may ask:

* How much blood you’ve been coughing up.
* How often you’ve been coughing up blood.
* How long you’ve been coughing up blood.
* How much blood is mixed with mucus or spit.

Your healthcare provider may also ask about behaviors that put you at risk, like drug use or smoking. They may try to identify potential causes by asking about other symptoms you’ve been experiencing.

They may perform any of the following tests to diagnose the underlying cause:

* A chest X-ray to look for tumors or fluid in your lungs.
* A computed tomography (CT) scan of your chest to investigate findings from the X-ray or accompany an additional procedure, like a bronchoscopy.
* A bronchoscopy to check if your airways are clear and locate the site where the bleeding’s occurring.
* A complete blood count (CBC) to assess how much blood you’ve lost and look for signs of infection.
* A sputum culture of your lung excretions to look for infectious causes that may be causing you to cough up blood, like bacteria or viruses.
* A blood clotting test (coagulation test) to see if a bleeding disorder is related to your symptoms.
* Pulmonary arteriography to see how blood flows through your lungs.
* A urinalysis and/or kidney function test to rule out autoimmune conditions that affect your lungs and kidneys.

Your provider may perform additional procedures or order other tests depending on what they suspect is causing you to cough up blood.

### **How is coughing up blood treated?**

Treatment depends on how serious your blood loss is and what’s causing you to cough up blood.

If you’re experiencing severe blood loss, you’ll receive care in the intensive care unit (ICU). Your care team will work to stabilize you and stop the bleeding before proceeding to diagnose what’s causing your blood loss.

Treatments for severe blood loss related to coughing up blood may include:

* A bronchoscopy to remove clots in your airways that may be causing the bleeding.
* Bronchial artery angiography and embolization to stop blood flow in blood vessels that are causing your bleeding.
* Medicine that’s used to stop bleeding related to severe blood loss (tranexamic acid).

Once they determine what’s causing you to cough up blood, your healthcare provider will discuss the best treatment plan to address your symptoms and underlying condition.

Treatments to address conditions that may cause you to cough up blood include:

* **Antibiotics**: If pneumonia or tuberculosis is causing your condition.
* **Steroids**: If inflammation is causing your condition.
* **Surgery and cancer treatment**: If a malignant (cancerous) tumor is causing your condition.

## **When To Call the Doctor**

Coughing up blood can be a sign of a serious medical condition. Seek emergency care if you’re coughing up large quantities of blood.

If you're coughing up small amounts of blood for longer than a week, make an appointment with a healthcare provider. They’ll determine what’s causing your hemoptysis and get you the necessary treatment.

Seek immediate medical attention if you’re coughing up more than a few teaspoons of blood, if you’ve been coughing up blood longer than a week or if your cough is accompanied by other symptoms, including:

* Fever.
* Chest pain.
* Night sweats.
* Shortness of breath.
* Rapid or severe weight loss.
* Dizziness or light-headedness.
* Blood in your urine (hematuria) or stools.

**Differential Diagnosis (DDx) of Coughing Up Blood (Hemoptysis) in Pediatrics**

## Common Causes:

* Lower respiratory tract infections: Pneumonia, tracheobronchitis, bronchitis are the most frequent causes.
* Foreign body aspiration: Causes mechanical trauma and inflammation leading to bleeding; often presents with coughing, wheezing, or choking history.
* Tracheostomy-related complications: In children with tracheostomy, local trauma or granulation tissue can cause bleeding.
* Bronchiectasis: Chronic airway inflammation and infection causing dilated bronchi and bleeding.
* Cystic fibrosis: Associated bronchiectasis and infection can cause hemoptysis.
* Congenital heart disease: Especially with pulmonary vascular obstruction or pulmonary hypertension.
* Pulmonary arteriovenous malformations (AVMs): Abnormal blood vessel connections can rupture and bleed.
* Alveolar hemorrhage syndromes: Including diffuse alveolar hemorrhage, Goodpasture syndrome, vasculitis (e.g., Wegener’s granulomatosis).
* Primary pulmonary hemosiderosis: Rare cause of recurrent alveolar bleeding.
* Neoplasms: Bronchial adenoma, metastatic tumors (rare in children).
* Trauma: Chest injury causing lung or airway bleeding.
* Tuberculosis: Especially in endemic areas, can cause hemoptysis.
* Parasitic infections: Such as echinococcosis (hydatid cyst).
* Idiopathic or cryptogenic hemoptysis: When no cause is found after thorough evaluation.
* Factitious hemoptysis: Self-inflicted or psychogenic bleeding, rare but important to consider.

**Epidemiology and Characteristics:**

* Hemoptysis is rare in children under two years old presenting to emergency departments. When it occurs, it is often linked to injury or serious illness.
* In one study of children under two, all cases of hemoptysis were associated with significant coughing and respiratory infections.
* Children, especially those under six, tend to swallow sputum, so hemoptysis may go unnoticed unless bleeding is substantial.
* Differentiating hemoptysis from other sources of bleeding, such as hematemesis (vomiting blood), is crucial for diagnosis and treatment. Hemoptysis typically presents as bright red or rust-colored, frothy blood mixed with sputum and has an alkaline pH. Hematemesis is usually dark red or brown, may be mixed with food particles, is often preceded by vomiting, and has an acidic pH.

Causes:

* The most common cause of hemoptysis in children is acute lower respiratory tract infection, accounting for about 40% of cases.
* Cystic fibrosis (CF) frequently causes hemoptysis, ranging from blood-tinged sputum to massive bleeding. Massive hemoptysis occurs in about 5% of CF patients, with mild hemoptysis in approximately 1% annually, more common in older patients .
* Other causes include:
  + Bronchiectasis (non-CF related): 10-15% incidence of hemoptysis.
  + Foreign body aspiration: A leading cause, especially in children under four. An abrupt onset of coughing in a healthy child can signal foreign body inhalation.
  + Pulmonary hemorrhage: Extravasation of blood into airways or lung tissue, uncommon in children.
  + Less common causes include primary pulmonary tuberculosis (less than 1% of cases), and in older children, vasculitis, bronchial tumors, and bronchiectasis

## **PREDEFINED QUESTION AND ANSWERS**

## What is hemoptysis in children?

Hemoptysis is the expectoration of blood or blood-tinged sputum from the lower respiratory tract. It is rare in children and often not life-threatening but can be serious if massive.

## What are the most common causes of pediatric hemoptysis?

* Infections: Lower respiratory tract infections such as pneumonia and tracheobronchitis are the leading causes (~40%).
* Foreign body aspiration: Especially in children under 4 years, causing local trauma and inflammation.
* Cystic fibrosis (CF): Hemoptysis ranges from mild to massive; about 5% of CF patients experience massive hemoptysis.
* Bronchiectasis (non-CF related) and tracheostomy-related complications also contribute.
* Less common causes include tuberculosis, aberrant bronchial circulation, pulmonary hemorrhage, and tumors.

## How is massive hemoptysis defined and managed?

Massive hemoptysis is generally defined as expectoration of more than 8 mL/kg in 24 hours or more than 240 mL total blood loss. Management includes airway stabilization, selective lung intubation, mechanical ventilation with high PEEP to tamponade bleeding, and possibly bronchial artery embolization or surgery.

## How is hemoptysis diagnosed in children?

Diagnosis relies on:

* Detailed history and physical exam to identify cause and severity.
* Chest X-ray and CT scan to localize bleeding and identify underlying pathology.
* Bronchoscopy for direct visualization, diagnosis, and sometimes treatment.
* Laboratory tests including CBC, coagulation profile, and arterial blood gases.

## What treatments are used for pediatric hemoptysis?

* Treat underlying infection with antibiotics or antivirals.
* Supportive care with oxygen and airway management.
* Bronchoscopic interventions for bleeding control (tamponade, vasoconstriction).
* Bronchial artery embolization or surgery if bleeding persists or is massive.

## What should be considered in the differential diagnosis?

* Differentiate hemoptysis from hematemesis or bleeding from the upper airway or gastrointestinal tract.
* Consider non-respiratory causes such as bleeding disorders or vascular malformations

**DOCTOR PATIENT CONVERSATION**

Doctor: Hello, I understand your child has been coughing up some blood. Can you tell me more about what you have noticed?

Parent: Yes, over the last two days, my child has coughed up some red-tinged mucus a few times. It’s not a lot, but it’s definitely blood.

Doctor: I see. Has your child had any recent illnesses, like a cold, cough, or fever?

Parent: Yes, they had a bad cough and runny nose about a week ago, but it seemed to be getting better.

Doctor: Has your child had any choking episodes or sudden coughing fits recently?

Parent: Actually, a few days ago, they started coughing suddenly during dinner, but it stopped quickly.

Doctor: That’s helpful to know. Sometimes, coughing up blood in children can be caused by infections or if something was inhaled into the lungs accidentally. Has your child ever been diagnosed with cystic fibrosis or any lung conditions?

Parent: No, no known lung problems.

Doctor: Okay. Has your child had any other symptoms like difficulty breathing, chest pain, or weight loss?

Parent: No, none of those.

Doctor: Good. We will do a physical exam and some tests, including a chest X-ray, to find out what’s causing the bleeding. Most of the time, it’s due to infections or irritation and can be treated effectively. If the bleeding gets worse or your child has trouble breathing, please bring them to the emergency room immediately.

Parent: Thank you, doctor. I’m worried but glad you’re checking it out.

Doctor: It’s understandable to be concerned. We’ll work together to find the cause and make sure your child gets the right treatment.

REFERENCES

[Coughing Up Blood: Causes and When To Seek Care](https://my.clevelandclinic.org/health/symptoms/17696-coughing-up-blood#overview)

**HYPOPLASTIC LEFT HEART SYNDROME**

**DEFINITION AND DESCRIPTION**

Hypoplastic left heart syndrome (HLHS) is a rare heart condition that a child is born with. That means it's a congenital heart defect. In this condition, the left side of the heart doesn't develop fully and is too small. So it can't pump blood well. Instead, the right side of the heart must pump blood to the lungs and to the rest of the body.

Treatment for hypoplastic left heart syndrome may include medicines, heart surgery or a heart transplant. Advances in care have improved the outlook for babies born with HLHS.

**CAUSES**

Hypoplastic left heart syndrome (HLHS) happens in the womb when a baby's heart develops. The cause isn't known. Gene changes may play a role.

In hypoplastic left heart syndrome, the left side of the heart hasn't grown enough so it does not develop fully. It can't properly send blood to the body. In HLHS, the following areas of the heart are too small:

* The lower left heart chamber, called the left ventricle.
* The body's main artery, called the aorta.
* The heart valves on the left side of the heart, called the aortic and mitral valves.

After birth, the right side of a baby's heart usually pumps blood both to the lungs and to the rest of the body. The blood passes through an opening called the ductus arteriosus. This opening, also called a vessel, connects the pulmonary artery directly to the aorta. The oxygen-rich blood goes back to the right side of the heart through a natural opening between the upper chambers of the heart. The opening is called the foramen ovale.

The ductus arteriosus usually closes after the first day or two of life. When that happens, the right side of the heart has no way to pump blood to the body. The left side of the heart takes over this job.

But in babies with hypoplastic left heart syndrome, the left side can't pump blood well. So they need medicine to keep these connections open and keep blood flowing to the body until they have heart surgery.

**Risk factors**

People who have a child with hypoplastic left heart syndrome (HLHS) have a higher risk of having another baby with this or a similar condition.

There is no other clear risk factors for hypoplastic left heart syndrome.

**Symptoms**

Babies born with hypoplastic left heart syndrome (HLHS) usually are very sick soon after birth. Symptoms of HLHS include:

* Blue or gray skin, lips or fingernails. Depending on skin color, these changes may be harder or easier to see.
* Rapid, difficult breathing.
* Poor feeding.
* Cold hands and feet.
* Weak pulse.
* Being more drowsy or less active than is typical for most babies.

Without treatment, a baby with this condition may go into shock. Symptoms of shock include:

* Cool, clammy skin that can be pale or lips that can be blue or gray.
* A weak and rapid pulse.
* Breathing that may be slow and shallow or very rapid.
* Dull eyes that seem to stare.

### **When to see a doctor**

Get emergency medical help if your baby has:

* Changes in skin or nail color.
* Trouble breathing or fast breathing.
* Weak pulse or rapid pulse.
* Cool clammy skin.

## **Diagnosis**

To diagnose hypoplastic left heart syndrome (HLHS), a healthcare professional examines the baby and listens to the baby's heart. The healthcare professional may hear a sound called a heart murmur. Rushing blood flow causes this sound.

### **Tests**

Tests used to find hypoplastic left heart syndrome (HLHS) in the baby before or after birth may include:

* **Pregnancy ultrasound.** A routine ultrasound exam during the second trimester of pregnancy usually can tell if the baby has HLHS.
* **Echocardiogram.** This test uses sound waves to make pictures of the heart. It shows how blood flows through the heart. It can be used after a baby is born to diagnose hypoplastic left heart syndrome. If a baby has HLHS, the test might find that the lower left heart chamber and heart valves are small. The body's main artery, called the aorta, also may be small.

**Treatment**

A baby born with hypoplastic left heart syndrome (HLHS) needs urgent treatment. Treatment can include many surgeries or a heart transplant. Medicines and other therapies are used to manage symptoms before heart surgery.

Talk with your child's healthcare professional about treatment options for your child.

If hypoplastic left heart syndrome is found before birth, healthcare professionals usually recommend giving birth at a hospital with a cardiac surgery center.

### **Medications**

The medicine alprostadil (Prostin VR Pediatric) is used to keep the ductus arteriosus open. It typically closes in all babies soon after birth. But in babies with hypoplastic left heart syndrome, the ductus needs to stay open so that blood can go to the rest of the body.

### **Therapies**

While waiting for surgery or a heart transplant, a baby with hypoplastic left heart syndrome may be given medicine and have these treatments:

* **Breathing help.** Babies who have trouble breathing may need help from a breathing machine called a ventilator.
* **Fluids through a vein.** A baby might receive fluids through a tube inserted into a vein. These are called intravenous (IV) fluids.
* **Feeding tube.** Babies who have trouble feeding or who tire while feeding can be fed through a feeding tube.

### **Surgery or other procedures**

Most children with hypoplastic left heart syndrome need several surgeries.

* **Atrial septostomy.** This treatment uses tubes called catheters and a balloon to make or widen an opening between the heart's upper chambers. It lets more blood flow between the upper two chambers of the heart. This treatment is done if the foramen ovale closes or is too small. Babies born with a hole in the heart, called an atrial septal defect, might not need atrial septostomy.

Other surgeries can make separate pathways to get the correct blood flow to the body and lungs. The surgeries are done in three stages.

* **Norwood procedure.** This surgery is often done within the first two weeks of life. There are various ways to do this treatment.

Surgeons rebuild the aorta and connect it to the heart's lower right chamber. Then they add a tube called a shunt to provide the lungs with blood. The surgeons may use one of two types of tubes. One type of tube connects the body's main artery to the arteries leading to the lungs. Those are the pulmonary arteries. The other type of tube goes from the right lower heart chamber to the pulmonary arteries. This treatment lets the right lower heart chamber pump blood to both the lungs and the body.

Sometimes, a mixed, also called hybrid, procedure is done. Heart surgeons place a stent in the ductus arteriosus to maintain the opening between the pulmonary artery and the aorta. Then they place bands around the pulmonary arteries to reduce blood flow to the lungs. They also make an opening between the upper chambers of the heart.

After the Norwood procedure, a baby's skin often is still slightly still slightly blue or gray. This is because oxygen-rich and oxygen-poor blood continue to mix within the heart. The blue or gray color may be easier or harder to see in some babies. Once a baby has this treatment, the odds of survival can go up.

* **Bidirectional Glenn procedure.** This tends to be the second surgery. It's usually done when a child is between 4 and 6 months of age. It involves removing the first shunt and connecting the large vein that drains blood from the head and arms to the pulmonary artery. Now the lungs receive blood from the vein instead of the shunt. The large vein is called the superior vena cava.

This surgery lessens the work of the right lower heart chamber by letting it pump blood mainly to the aorta. It also lets most of the oxygen-poor blood returning from the body flow directly into the lungs. After this treatment, all the blood returning from the upper body flows to the lungs. So blood with more oxygen is pumped to the aorta to supply organs and tissues throughout the body.

* **Fontan procedure.** This surgery usually is done when a child is between 3 and 4 years of age. The surgeon creates a path for the blood from the lower legs to flow directly into the pulmonary arteries. The vessel involved is called the inferior vena cava. The pulmonary arteries then send the blood into the lungs.

The Fontan procedure lets the rest of the oxygen-poor blood returning from the body flow to the lungs. After this surgery, there's little mixing of oxygen-rich and oxygen-poor blood in the heart. So the skin should no longer look blue or gray.

* **Heart transplant.** Many babies with hypoplastic left heart syndrome need a heart transplant. (8) Children who have heart transplants need to take medicines for life to so that their bodies don't reject the donor heart.

### **Follow-up care**

After surgery or a transplant, a baby needs lifelong care with a heart doctor trained in congenital heart disease to watch for complications. Your child may need further treatment or other medicines for these complications.

Some children may need to take antibiotics before certain dental or other procedures to help prevent infections. Ask your child's healthcare professional if your child needs preventive antibiotics. Some children also may need to limit physical activity.

### **Follow-up care for adults**

Adults who were born with hypoplastic left heart syndrome (HLHS) need to see a heart doctor trained in congenital heart disease in adults. Recent advances in surgical care have helped children with HLHS grow into adulthood. So it's not yet clear what challenges an adult with the heart condition might have. Adults need regular, lifelong follow-up care to watch for changes in the condition.

People thinking about becoming pregnant should talk with their healthcare professionals about pregnancy risks and birth control options. Having hypoplastic left heart syndrome raises the risk of:

* Heart and blood vessel problems during pregnancy.
* Miscarriage.
* A baby being born with a congenital heart defect.

**Complications**

With proper treatment, many babies with hypoplastic left heart syndrome (HLHS) survive. But they do need many surgeries and can have less energy and other challenges. Complications of HLHS may include:

* Getting tired more easily during sports or other exercises.
* Irregular heartbeats, called arrhythmias.
* Fluid buildup, called edema, in the lungs, stomach area, legs and feet.
* Not growing well.
* Developmental conditions related to the brain and nervous system.
* Need for more heart surgery or a heart transplant.

**Prevention**

There's no way to prevent hypoplastic left heart syndrome. If you were born with a heart condition, talk with a heart doctor and genetic counselor before getting pregnant.

**Outlook / Prognosis**

After treatment for hypoplastic left heart syndrome, your child will need lifelong follow-up visits at least once a year with a cardiologist (heart doctor). These visits ensure that their heart, lungs and other organs continue to work properly. When your child becomes an adult, they’ll transition to care with a specialist in adult congenital heart disease.

Most children with hypoplastic left heart syndrome will need heart medications. They’ll also need to take antibiotics before any other surgeries, including dental surgeries. These medications reduce the risk of endocarditis (a heart infection).

#### **Outlook for hypoplastic left heart syndrome**

Without treatment, hypoplastic left heart syndrome is fatal days or weeks after birth.

With treatment, the prognosis depends on the complexity of your child’s heart deformity. Ask your baby’s healthcare provider about the risks associated with each surgery.

Some children may have decreased physical endurance for the rest of their lives. Usually, healthcare providers recommend limiting intensive physical activities, such as competitive sports.

### **Survival rate of hypoplastic left heart syndrome**

About 20% to 60% of babies with hypoplastic left heart syndrome survive their first year of life. After that, the survival rate for the next five, 10 and 15 years is about 40%. Babies who have a normal birth weight and aren’t born prematurely do better than babies with lower birth weights. One study found that most babies who survived their first year were still alive at age 18.

## **Prevention**

Many HLHS cases don’t have an obvious cause. However, providers always encourage healthful habits during pregnancy, including:

* Avoiding alcohol and smoking.
* Managing any medical conditions, such as diabetes.
* Eating a healthy diet.
* Taking a daily prenatal vitamin with at least 400 micrograms (mcg) of folic acid.

If you or your partner has hypoplastic left heart syndrome in their family, you may want to talk with a genetic counselor before getting pregnant.

**Living With**

Children born with this condition can live a healthy life with long-term monitoring from a cardiologist.

You can care for your child in these ways:

* Make sure your child gets vaccinated against the flu every year and against COVID-19.
* Bring your child to cardiologist appointments every six months or every year.
* Make sure your child takes their medications.
* Limit your child’s intense physical activity based on their provider’s recommendations.
* Work with them if they have learning difficulties.

### **COMMON QUESTIONS AND ANSWERS**

* What are the risks of surgery?  
  The initial surgery (Norwood procedure) carries the highest risk, with mortality rates around 20-25%, though some centers report better outcomes. Risks include complications such as aortic arch obstruction, heart rhythm problems, and organ dysfunction. Later surgeries (Glenn and Fontan procedures) have progressively lower risks but still carry potential complications.
* Are there potential surgery complications to watch for?  
  Yes. Complications can include heart failure, arrhythmias, thrombosis, infections, stroke, nerve damage, and issues related to the shunts or conduits placed during surgery. Long-term risks include heart rhythm abnormalities, protein-losing enteropathy, and decreased exercise tolerance.
* What medications does my child need?  
  Most children require lifelong heart medications, including those to support heart function and prevent infections. Antibiotics are often needed before surgeries or dental work to reduce the risk of endocarditis (heart infection).
* Are there any medication side effects?  
  Side effects vary depending on the medications but may include impacts on kidney or liver function, blood clotting, or immune system effects. Your healthcare provider will monitor for side effects closely during follow-up visits.
* How will HLHS affect my child’s life?  
  Children with HLHS often have reduced physical endurance and may need to limit strenuous activities. They may experience developmental delays and require ongoing medical care. Lifelong cardiology follow-up is essential. Some may eventually need a heart transplant.
* What follow-up care does my child need after surgery?  
  Lifelong follow-up with a cardiologist specializing in congenital heart disease is necessary, at least annually. Monitoring includes heart function, growth, development, and screening for complications. Transition to adult congenital heart disease specialists occurs as the child matures.
* If I have another baby, does that child have a higher risk of HLHS?  
  HLHS is a congenital condition with some genetic and environmental risk factors. While the exact recurrence risk varies, having one child with HLHS slightly increases the risk for subsequent children. Genetic counseling can provide personalized risk assessment

**DIFFERENTIAL DIAGNOSIS**

Several diseases or conditions can mimic the presentation of HLHS, particularly in neonates with cyanosis, signs of poor perfusion, or congestive heart failure. Accurate differentiation is critical, as management strategies for these conditions differ significantly from HLHS. Below is a discussion of diseases or conditions that can be mistaken for HLHS:

* Critical aortic stenosis
  + Severe aortic stenosis can cause LV hypertrophy and poor systemic perfusion, resembling HLHS. However, the LV is typically present in critical aortic stenosis and may be dilated rather than hypoplastic. Echocardiography can differentiate this condition by visualizing the LV size and function and assessing the degree of aortic valve obstruction.
* Coarctation of the aorta
  + Coarctation of the aorta, especially with a large ventricular septal defect, can lead to cyanosis and shock as the ductus arteriosus begins to close. Because of systemic hypoperfusion, this condition may be confused with HLHS, but echocardiography will show a normally sized or hypertrophied LV and an obstruction localized to the aortic isthmus.
* Shone complex
  + Shone complex is a rare congenital heart condition characterized by multiple levels of left-sided obstructive lesions, including a supravalvular mitral membrane, a parachute mitral valve with all chordae attached to a single papillary muscle, subaortic stenosis caused by a fibromuscular ridge below the aortic valve, and coarctation of the aorta impairing systemic blood flow. This condition may be mistaken for HLHS because both conditions involve reduced left-sided cardiac output and systemic hypoperfusion. Symptoms such as cyanosis, poor perfusion, and heart failure, as the ductus arteriosus closes, are common to both, but echocardiography can differentiate them by identifying the discrete levels of obstruction in the Shone complex versus the global underdevelopment of the left heart in HLHS.
* Interrupted aortic arch
  + This condition can present with systemic hypoperfusion and shock, similar to HLHS. This condition involves complete discontinuity of the aortic arch, but echocardiography or advanced imaging will reveal a normal or mildly hypoplastic LV and the specific site of aortic interruption.[[8]](https://www.ncbi.nlm.nih.gov/books/NBK554576/#)
* Total anomalous pulmonary venous return (TAPVR)
  + In TAPVR with obstruction, neonates may present with cyanosis and signs of low cardiac output that mimic HLHS. However, TAPVR is characterized by abnormal pulmonary venous drainage into the RA or systemic veins, which can be identified on echocardiography. The left-sided structures are typically normal in size and morphology.
* Pulmonary atresia with intact ventricular septum (PA/IVS)
  + PA/IVS may resemble HLHS due to ductal-dependent systemic perfusion and cyanosis. However, in PA/IVS, the LV is usually normal in size, and the obstruction is at the pulmonary valve rather than the aortic or mitral valves.
* Ebstein anomaly of the tricuspid valve
  + Severe forms of Ebstein anomaly can cause cyanosis and right-sided heart failure, potentially mimicking HLHS. However, echocardiography will show significant apical displacement of the tricuspid valve with atrialization of the RV, distinguishing this condition from HLHS.

**EPIDEMIOLOGY**

HLHS is a rare congenital heart defect, accounting for approximately 2% to 3% of all congenital heart defects and about 6% to 9% of critical congenital heart disease requiring surgery or catheter-based intervention in the neonatal period. The incidence of HLHS is estimated to be 0.16 to 0.36 per 1000 live births and accounts for 1.4% to 3.8% of all congenital heart defects. Males are affected more frequently than females, with a male-to-female ratio of approximately 1.5:1. HLHS is responsible for 23% of cardiac deaths during the first week of life, highlighting its significant contribution to early neonatal mortality.

Several study results have suggested a multifactorial etiology involving genetic and environmental factors. Although most cases of HLHS are sporadic, up to 20% may occur in association with chromosomal abnormalities or syndromes, such as Turner syndrome, trisomy 13, or trisomy 18. Familial clustering has been observed, and mutations in genes like *NOTCH1* and *HAND1* have been implicated, suggesting a genetic predisposition in some cases.

HLHS is often diagnosed prenatally through routine fetal ultrasonography, with advances in imaging techniques enabling earlier and more accurate detection. Early diagnosis has improved perinatal management and outcomes. Despite its rarity, HLHS remains a leading cause of neonatal mortality due to congenital heart disease if left untreated, underscoring the importance of early intervention and specialized care.

## **Procedures**

### Cardiac catheterization (pre–Norwood procedure)

Routine diagnostic catheterization is not necessary because 2-dimensional and Doppler echocardiography can provide the necessary anatomic and hemodynamic data. However, a focused catheterization may become necessary to resolve any echocardiographic discrepancies that may be deemed important in surgical management.

If catheterization is performed, the features reflect the pathophysiology described above.

Oxygen saturation data indicate moderate-to-severe systemic venous desaturation, with a step-up at the level of right atrium secondary to left to right shunt across the atrial septum. The oxygen saturations in the right ventricle, pulmonary artery and aorta are similar reflecting common mixing in the right atrium. Mild systemic arterial desaturation is usually present unless severe pulmonary edema is noted.

The right atrial pressure is mildly elevated, and the left atrial pressure is moderate to severely elevated, unless a large atrial septal defect is present. The right ventricular and pulmonary arterial systolic pressures are at systemic level. If the ductus arteriosus is constricted, these pressures are higher than those in the aorta.

Angiography, although not necessary in all cases, reveals hypoplasia of the mitral valve, left ventricle and aorta. The ascending aorta is perfused in a retrograde fashion and serves as a common coronary artery, supplying both the right and left coronary arteries.

Perform interventional catheterization with blade/balloon atrial septostomy or static dilatation of the atrial septum to relieve pulmonary venous hypertension if blood flow from left atrium to right atrium is severely restricted at the atrial septum.Because of marked hypoplasia of the left atrium, conventional Rashkind balloon or Park blade atrial septostomy may not be possible. In such situations, static dilatation of the atrial septum may be performed.If the atrial septum is extremely thick with a markedly restrictive atrial septum, stent implantation to keep the atrial septum open may become necessary.

When hybrid procedures are contemplated, stenting of the ductus, atrial septum, or both may become necessary.

### Cardiac catheterization (pre–bidirectional Glenn [stage II] procedure)

Perform routine catheterization before the operation to obtain hemodynamic data and several important angiograms.

Calculate pulmonary vascular resistance to ensure the patient's suitability for the stage II procedure.

Perform angiography in the right ventricle to show ventricular function and tricuspid regurgitation.

Perform angiography in the transverse aortic arch near the shunt to show pulmonary artery size and distribution and to rule out recurrent aortic coarctation or significant aortopulmonary collateral vessels.

If collateral vessels are found, they may be occluded with coils at the same catheterization.

### Pre-Fontan (stage III) procedure

Routine catheterization before completing the operation is generally recommended.

Measure pulmonary artery pressure and calculate pulmonary vascular resistance and perform right ventricular angiography.

Delineate pulmonary artery anatomy by performing angiography at the superior vena cava–pulmonary artery anastomosis via an internal jugular approach.

Recurrent coarctation of the aorta and significant collateral vessels are excluded again.

Descending aortography and selective right and left subclavian artery angiography to identify any collateral vessels to lungs is recommended.

Postcatheterization complications include hemorrhage, vascular disruption after balloon dilation, pain, nausea and vomiting, and arterial or venous obstruction from thrombosis or spasm but are rare.

### Catheter intervention

In the neonate, obstruction at the level of the patent foramen ovale (atrial septum) may be treated with conventional Rashkind balloon atrial septostomy. However, because the left atrium is small, Rashkind septostomymay not be feasible. In addition, the septum may be too thick to be torn by balloon septostomy; therefore, Park blade septostomy may be necessary and should precede the Rashkind procedure.However, hypoplastic left atrium may preclude blade septostomy. Static dilatation of the atrial septum with a balloon angioplasty catheter may be used and may not only relieve the obstruction but also keep some restriction, such that no rapid fall in the pulmonary vascular resistance occurs. Static balloon dilatation is preferred by the senior author.

In some patients, the atrial septum may be intact or have a tight patent foramen ovale that may not even allow passage of a catheter. In such situations, puncture of the atrial septum by a Brockenbrough technique or radiofrequency perforation of the atrial septum followed by static balloon atrial septal dilatation or, preferably, stent implantation may become necessary.

If progressive cyanosis develops after a previous Blalock-Taussig shunt, and if the hypoxemia is due to a stenotic shunt, balloon dilatation may be used to improve oxygen saturation.However, if the patient is of sufficient size and age to undergo a bidirectional Glenn procedure, this procedure should be performed instead of balloon angioplasty of a narrowed Blalock-Taussig shunt.

If severe aortic coarctation is present, particularly after Norwood procedure, balloon angioplasty may be useful in relieving aortic obstruction and may help achieve reduce right ventricular afterload.

If significant branch pulmonary artery stenosis is present before a bidirectional Glenn or Fontan conversion or after a Fontan procedure, balloon angioplasty or placement of intravascular stents is recommended.

Development of aortopulmonary collateral vessels has been increasingly observed in recent studies. Before the final Fontan conversion, occlusion of these vessels in the catheterization laboratory, usually by means of coil embolization,is recommended to reduce left ventricular volume overloading and, probably, the duration of chest tube drainage following surgery.

Following completion of Fontan procedure, some patients may develop recurrent pleural effusion, liver dysfunction, plastic bronchitis or protein-losing enteropathy.In these patients, following exclusion of obstructive lesion in the Fontan circuit, puncture of the atrial septum by a Brockenbrough technique followed by static balloon atrial septal dilatation or stent implantation may be beneficial.

Patients who have undergone a fenestrated Fontan operation or have a residual atrial defect, despite correction, may have significant right-to-left shunting causing severe hypoxemia. These residual atrial defects may be closed using transcatheter techniques.

**Here are some commonly used drug classes and their considerations in pediatric HLHS:**

* Prostaglandin E1 (PGE1)
  + Purpose: Keeps the ductus arteriosus open, which is critical for blood flow to the body in newborns with HLHS until surgical repair can be performed.
  + Side Effects: Apnea (temporary cessation of breathing) is a common side effect, especially in neonates, requiring careful monitoring.
* Diuretics (e.g., Loop diuretics like Furosemide, Thiazide diuretics)
  + Purpose: Used to manage fluid balance and control volume status, which is important for improving cardiac function and alleviating symptoms of heart failure.
  + Side Effects: Can include electrolyte imbalances (e.g., low potassium), dehydration, and kidney dysfunction.
* Inotropic Drugs (e.g., Milrinone, Dopamine, Dobutamine)
  + Purpose: These medications strengthen the heart's contractions and can help improve cardiac function, particularly in severely ill neonates with cardiogenic shock or to prevent post-operative low cardiac output syndrome. Milrinone, a phosphodiesterase-3 inhibitor (PDE3i), has shown benefits in improving heart failure symptoms and survival in infants with single ventricle physiology, often used as a bridge to transplant.
  + Side Effects: Can include arrhythmias, changes in blood pressure, and in adults, chronic use of PDE3 inhibitors has been associated with increased mortality, though this class is commonly used in children.
* Digoxin
  + Purpose: A cardiac glycoside that was historically used for heart failure and arrhythmias. While its use has decreased due to narrow therapeutic windows and drug interactions in adults, it is still occasionally used in pediatric patients with fewer comorbidities. Some retrospective studies suggest improved survival in infants with HLHS discharged on digoxin after the Norwood procedure.
  + Side Effects: Requires close monitoring due to a narrow margin between therapeutic and toxic concentrations. Potential side effects include arrhythmias and gastrointestinal issues.
* Beta-Blockers (e.g., Carvedilol, Metoprolol)
  + Purpose: Fundamental in adult heart failure treatment, but their efficacy in pediatric single ventricle heart failure, including HLHS, has been limited. Some studies have even shown worse outcomes with carvedilol in patients with a systemic right ventricle.
  + Side Effects: Can include bradycardia (slow heart rate) and hypotension.
* ACE Inhibitors (e.g., Captopril, Enalapril)
  + Purpose: Used to manage heart failure, though their specific efficacy in pediatric HLHS is not as well-established as in biventricular heart failure.
  + Side Effects: Can include hypotension and kidney dysfunction.
* Antibiotics
  + Purpose: Children with HLHS often need prophylactic antibiotics before surgeries or dental work to reduce the risk of endocarditis (heart infection).
  + Side Effects: Vary depending on the specific antibiotic but can include gastrointestinal upset, allergic reactions, and antibiotic resistance with overuse.

**PREDEFINED QUESTIONS AND ANSWERS**

## What is Hypoplastic Left Heart Syndrome (HLHS)?

HLHS is a congenital heart defect where the left side of the heart is underdeveloped and cannot pump blood effectively to the body. This includes a small or missing left ventricle, mitral valve, aortic valve, and aorta.

## What are the signs and symptoms of HLHS in a baby?

Common signs include:

* Fast or difficult breathing
* Blue or grayish skin and nails (cyanosis)
* Trouble feeding
* Low energy or lethargy
* Fewer wet diapers than normal.

## How is HLHS diagnosed?

HLHS can sometimes be detected before birth via prenatal ultrasound. After birth, diagnosis involves:

* Chest X-ray
* Electrocardiogram (ECG/EKG)
* Echocardiogram (heart ultrasound)
* Pulse oximetry to measure oxygen levels
* Blood tests.

## How is HLHS treated?

Treatment involves:

* Prostaglandin medication to keep the ductus arteriosus open, allowing blood flow to the body.
* A series of three staged surgeries:
  + Norwood procedure (within the first two weeks of life)
  + Glenn procedure (around 4-6 months)
  + Fontan procedure (between 18 months and 3 years).
* Some babies may need feeding tubes or additional catheter procedures to improve blood flow.

## What is the outlook for children with HLHS?

Without surgery, HLHS is fatal. With staged surgical treatment, many children survive into childhood and beyond, though they require lifelong cardiac care and monitoring.

## What follow-up care will my child need?

Children with HLHS need lifelong follow-up with pediatric cardiologists experienced in congenital heart disease. This includes regular imaging, monitoring for complications, and managing growth and development.

## Can HLHS be prevented?

The exact cause is unknown, and there are no known ways to prevent HLHS. It may involve genetic and environmental factors.

## If I have one child with HLHS, is my next baby at higher risk?

Having one child with HLHS slightly increases the risk for another child to have congenital heart defects. Genetic counseling is recommended for personalized risk assessment

**DOCTOR PATIENT CONVERSATION**

Doctor: Hello, I understand your baby was diagnosed with hypoplastic left heart syndrome. I want to explain what this means and what we can do to help.

Parent: Yes, we’re still trying to understand it. What exactly is HLHS?

Doctor: HLHS is a condition where the left side of the heart — including the left ventricle, mitral valve, aortic valve, and the ascending aorta — is very small or underdeveloped. Because of this, the heart cannot pump blood effectively to the body on its own. Your baby’s oxygen-rich blood mixes with oxygen-poor blood in the right side of the heart and relies on a special blood vessel called the ductus arteriosus to supply blood to the body.

Parent: Is this why the doctors started a medication right away?

Doctor: Exactly. We give a medication called prostaglandin E1 to keep the ductus arteriosus open. This is critical because if it closes, blood flow to the body will stop, which can be life-threatening.

Parent: What happens next? Is surgery needed?

Doctor: Yes, HLHS requires a series of three surgeries staged over the first few years of life. The first, called the Norwood procedure, usually happens within the first two weeks after birth. This surgery reconstructs the heart’s pathways to improve blood flow. Later, the Glenn and Fontan procedures further help the heart work more efficiently.

Parent: Will my child need other treatments?

Doctor: Besides surgery, your baby will need close monitoring and medications to support heart function and prevent complications. Some babies also need special feeding support because they can tire easily during feeding.

Parent: What is the outlook for children with HLHS?

Doctor: With modern surgical techniques and care, many children survive and grow, but they will need lifelong follow-up with a cardiologist experienced in congenital heart disease. They may have some limitations in activity and require ongoing medical care.

Parent: If we have another baby, is there a chance they could have HLHS too?

Doctor: Having one child with HLHS slightly increases the chance of congenital heart defects in future children. We can refer you for genetic counseling to better understand the risks.

Parent: Thank you for explaining everything. It’s a lot to take in, but I feel better knowing the plan.

Doctor: I understand this is overwhelming. We’re here to support you and your baby every step of the way. Please don’t hesitate to ask questions anytime.

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# **Parapneumonic pleural effusion**

**DEFINITION AND DESCRIPTION**

Pleural effusion is a buildup of fluid in the pleural space. The pleural space is the area between the layers of the tissue lining the lung and the chest cavity.

In a person with parapneumonic pleural effusion, the fluid buildup is caused by pneumonia.

## **Causes**

Pneumonia, most commonly from bacteria, causes parapneumonic pleural effusion.

## **Symptoms**

Symptoms can include any of the following:

* Chest pain, usually a sharp pain that is worse with cough or deep breaths
* Cough with sputum
* Fever
* Rapid breathing
* Shortness of breath

## **Exams and Tests**

Your health care provider will examine you and ask about your symptoms. Your provider will also listen to your lungs with a stethoscope and tap (percuss) your chest and upper back.

The following tests may help to confirm a diagnosis:

* Complete blood count (CBC)
* Chest CT scan
* Chest x-ray
* Thoracentesis (a sample of fluid is removed with a needle inserted between the ribs)
* Ultrasound of the chest and heart

## **Treatment**

Antibiotics are prescribed to treat the pneumonia.

If the person has shortness of breath, thoracentesis might be used to drain the fluid. If better drainage of the fluid is needed due to more severe infection, a drain tube can be inserted.

## **Outlook (Prognosis)**

This condition improves when the pneumonia improves.

## **Possible Complications**

Complications may include:

* Lung damage
* Infection that turns into an abscess, called an empyema, which will need to be drained with a chest tube
* Collapsed lung (pneumothorax) after thoracentesis
* Scarring of the pleural space (lining of the lung)

## **When to Contact a Medical Professional**

Contact your provider if you have symptoms of pleural effusion.

Contact your provider or go to the emergency room if shortness of breath or difficulty breathing occurs right after thoracentesis.

## **Alternative Names**

Pleural effusion - pneumonia

## **Epidemiology**

### US and international statistics

Hospital discharge data indicate that in the United States, approximately 1.3 million patients are hospitalized with pneumonia every year. The prevalence of parapneumonic effusions is dependent, in part, on the organism involved.

Overall, pleural effusions are seen in about 35-40% of patients with bacterial pneumonia or anaerobic pneumonia, with a prevalence approaching 60% in pneumococcal pneumonia. Complicated pleural effusions are more common with anaerobic pleuropulmonary infections. This results in an estimated 500,000-750,000 patients with parapneumonic effusions annually. No good estimates are available regarding how many of these patients proceed to complicated effusions or empyema, but in small series, approximately 5-10% have been found require drainage or a surgical procedure.

A study of US hospitalization data found that in 1996, the national hospitalization rate for parapneumonic empyema–related diagnoses was 3.04 per 100,000; by 2008, it had increased to 5.98 per 100,000, and by 2016, it had increased to 11.1 per 100,000. The empyema hospitalizations had a high cost per case ($38,591) and required a prolonged stay (13.8 d). Over the study period, a downward trend in length of stay and a small reduction in associated mortality were not, possibly due to increasing use of innovative treatments (e.g., surgery or intrapleural enzymatic therapy).

No good estimates are available on the international incidence of pneumonia. The World Health Organization (WHO) cited a figure of 4.2 million for cases involving death from lower respiratory tract infections in 2004. From this figure, it is possible to extrapolate the incidence of pleural effusions and empyema using a US estimate, but caution is advised because lack of treatment and delayed treatment in developing countries may skew the international incidence upward.

### Age-, sex-, and race-related demographics

In a 2021 report from the American College of Chest Physicians (ACCP), 60% of the hospitalizations related to pleural infections were found to occur in the 18- to 64-year-old age group.In the United Kingdom and Europe, this age group accounted for 40% of adult pleural infections. Advancing age leads to an increase in associated comorbidities, including a higher risk of pneumonia and, subsequently, pleural effusions and empyema.

In the 2021 ACCP report, a male predominance in adult pleural infections was noted.Plausible hypotheses for this difference include the following:

* Gender differences in health-seeking behaviors, with a delayed presentation in males
* Worse dental hygiene trends
* Greater frequency of preexisting comorbidities in males

No specific ethnic or racial predisposition is recognized for empyema; however, a larger number of ethnic minorities have limited financial resources, less access to healthcare, and more comorbidities, which, in turn, may increase their risk of pneumonia, pleural effusions, and empyema.

## **Differential Diagnoses**

* Aspiration Pneumonitis and Pneumonia
* Bacterial Pneumonia
* Boerhaave Syndrome
* Community-Acquired Pneumonia (CAP)
* Fungal Pneumonia
* Hemothorax
* Lung Abscess
* Non-Small Cell Lung Cancer (NSCLC)
* Peritonitis and Abdominal Sepsis
* Pleural Effusion
* Pleurodynia
* Pneumococcal Infections (Streptococcus pneumoniae)
* Secondary Lung Tumors
* Small Cell Lung Cancer (SCLC)
* Tuberculosis (TB)

## **Procedures**

### Thoracocentesis and evaluation of pleural fluid

A diagnostic thoracentesis is recommended for pleural fluid sampling when the effusion is suspected to be parapneumonic in nature. With the routine bedside use of US guidance for localization, there is no minimum safe size of effusion that should deter the procedure if it is clinically indicated. This determination, however, should be made on the basis of the clinician's skill level. Generally, pleural effusion greater than or equal to 10 mm thick on a lateral decubitus chest radiograph is considered safe.For large-volume pleural effusion, the procedure may also serve a therapeutic purpose in relieving the dyspnea related to the effusion.

Right lateral decubitus chest radiograph shows free-flowing pleural effusion, which should be sampled with thoracentesis for pH determination, Gram stain, and culture.

*Assessment of gross appearance*

Pleural fluid may range in appearance from a clear yellow liquid to an opaque turbid fluid to grossly purulent thick, viscous, foul-smelling pus. Foul-smelling fluid indicates an anaerobic infection.

*Pleural fluid testing*

Studies used in the evaluation of pleural fluid include the following:

* Cell count and differential - Results generally are not diagnostic, but most transudates are associated with a white blood cell (WBC) count lower than 1000/µL; in contrast, elevated cell counts with neutrophil predominance are typically noted in the early inflammatory phase of complicated parapneumic effusions,

**PREDEFINED Q AND A**

## What are parapneumonic effusions and empyema?

* Parapneumonic effusions are pleural fluid collections that occur as a complication of pneumonia. They range from simple, sterile fluid accumulation to infected fluid.
* Empyema is the presence of pus in the pleural space, representing an advanced, infected parapneumonic effusion.

## How common are parapneumonic effusions and empyema in children with pneumonia?

* More than 40% of bacterial pneumonias develop parapneumonic effusions, and a smaller percentage progress to empyema . Empyema complicates about 0.6-3% of pediatric pneumonia hospitalizations .

## What are the stages of parapneumonic effusion and empyema development?

1. Exudative (Uncomplicated) Stage: Sterile, clear or slightly cloudy pleural fluid with normal glucose and pH. Usually resolves with antibiotics alone.
2. Fibropurulent (Complicated) Stage: Bacterial invasion causes pus formation, loculations, low pleural fluid pH (<7.2), low glucose (<60 mg/dL), and high LDH (>1000 IU/L). Requires drainage in addition to antibiotics.
3. Organizational Stage: Fibroblast proliferation forms thick pleural peel restricting lung expansion. Fluid is thick and acidic (pH <7.0), with very low glucose and high LDH. Surgical intervention may be necessary.

## What symptoms suggest parapneumonic effusion or empyema in children?

* Fever and persistent or worsening cough
* Chest pain, often pleuritic
* Difficulty breathing or increased work of breathing
* Reduced breath sounds and dullness to percussion on the affected side
* Signs of systemic infection such as tachycardia and malaise

## How is the diagnosis confirmed?

* Chest X-ray showing pleural fluid
* Ultrasound or CT scan to assess fluid characteristics and loculations
* Thoracentesis with pleural fluid analysis (cell count, Gram stain, culture, pH, glucose, LDH) to differentiate uncomplicated vs. complicated effusions or empyema

## What is the treatment approach?

* Uncomplicated effusions: Antibiotics alone, often managed outpatient if respiratory status is stable.
* Complicated effusions and empyema: Require antibiotics plus drainage via chest tube thoracostomy.
* If drainage fails or loculations persist, options include intrapleural fibrinolytic therapy or video-assisted thoracoscopic surgery (VATS).
* Supportive care includes oxygen, pain management, and monitoring for complications.

## What are the potential complications if untreated?

* Chronic empyema with thick pleural peel causing lung restriction
* Bronchopleural fistula
* Septicemia and systemic infection
* Permanent loss of lung function

## What is the prognosis?

* With prompt antibiotic therapy and appropriate drainage, most children recover fully.
* Mortality is low but can reach 15-20% in severe cases or delayed treatment.

**DOCTOR PATIENT CONVERSATION**

Doctor: Hello, I understand your child has been diagnosed with a pleural effusion related to pneumonia. I want to explain what this means and how we will manage it.

Parent: Yes, can you tell me what a pleural effusion is?

Doctor: Certainly. A pleural effusion is when fluid collects in the space around the lungs, called the pleural space. When this happens as a complication of pneumonia, we call it a parapneumonic effusion. If the fluid becomes infected and turns into pus, that is called empyema.

Parent: How serious is this? What symptoms should I watch for?

Doctor: It can range from mild to more serious. Your child may have persistent fever, cough, chest pain, and difficulty breathing. If the fever continues despite antibiotics, or breathing worsens, it suggests the effusion may be complicated or empyema has developed.

Parent: What tests will you do?

Doctor: We will do a chest X-ray and an ultrasound to see how much fluid is present and whether it is thick or loculated. We may also take a sample of the fluid with a needle to check for infection.

Parent: How is it treated?

Doctor: For small, uncomplicated effusions, antibiotics alone may be enough. For larger or infected effusions, your child may need a chest tube to drain the fluid. Sometimes, if the fluid is thick or loculated, we use medications called fibrinolytics or perform a minimally invasive surgery called VATS to remove the infected material.

Parent: Will my child recover fully?

Doctor: Most children recover completely with appropriate treatment. We will monitor your child closely to ensure the infection clears and lung function returns to normal.

Parent: Is this common?

Doctor: Parapneumonic effusions and empyema are uncommon but increasing complications of pneumonia in children. Early diagnosis and treatment are important for the best outcome.

Parent: Thank you, doctor. That helps me understand what to expect.

Doctor: You’re welcome. Please contact us immediately if your child’s breathing worsens, fever persists, or if you have any concerns.

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### **Pectus carinatum(PIGEON CHEST)**

**DEFINITION AND DESCRIPTION**

Pectus carinatum is a condition in which your sternum (breastbone) sticks out more than usual. Some people call it “pigeon chest” or “keel chest” because of how it makes your chest look. It’s the opposite of pectus excavatum, in which your breastbone is depressed inward and gives your chest a sunken appearance.

#### **Types of pectus carinatum**

Healthcare providers recognize two main pectus carinatum types:

* **Chondrogladiolar**. This is the more common type. The main part of your breastbone (farther down) sticks out.
* **Chondromanubrial**. The upper part of your breastbone sticks out.

Providers may also talk about whether the condition affects both sides of your chest (symmetric) and if your sternum rotates to one side.

* Type I is chondrogladiolar and affects both sides.
* Type II is chondromanubrial and affects both sides.
* Type III only affects one side and you can have a depression on the other side.

Pectus carinatum affects about 1 in 1,000 people. But it may be even more common. Some estimate that 1 in 300 people have it.

It’s more common in men than women. Although the condition is present at birth, people often notice it during their teen years.

**Symptoms and Causes**

Most people with pectus carinatum have no symptoms. They may have some chest pain associated with certain activities and positions.

Pectus carinatum isn’t dangerous and doesn’t impact life expectancy.

**What causes pectus carinatum?**

Researchers don’t know the exact cause of pectus carinatum. But they believe it’s a disorder of the cartilage that joins your ribs to your breastbone.

Researchers haven’t found a genetic issue yet, but they may in the future. Up to 33% of people with pectus carinatum have a family history of a chest wall issue.

Pectus carinatum has an association with:

* Marfan syndrome, a connective tissue disorder.
* Noonan syndrome, a genetic disorder.
* Scoliosis, abnormal curvature of the spine.
* Asthma, a lung disease.
* Bronchitis, inflamed airways.
* Mitral valve prolapse, a floppy heart valve.

### **Complications of pectus carinatum**

Complications of pectus carinatum may include:

* Self-esteem issues.
* Bad posture.
* Back pain.

**Diagnosis and Tests**

A healthcare provider will take your medical history and perform a physical examination. They may also check to see if you have scoliosis. Then they’ll order tests if they need to.

The main test for diagnosing pectus carinatum is a chest X-ray from the front and side. Other tests may include:

* **Computed tomography (CT) scan**, a type of X-ray.
* **Magnetic resonance imaging (MRI) scan**, a type of imaging that doesn’t use X-rays.

## **Management and Treatment**

Bracing and surgery are the pectus carinatum treatment options for people who want correction. Bracing is a nonoperative intervention that’s generally more effective in children. It requires compliance with the use of a brace, which can be challenging for children.

#### **Pectus carinatum brace**

This works like the way braces work on teeth. You wear the brace around your chest under or on top of your clothes. The brace provides pressure from both the front and back to move the breastbone back to its usual position. You wear it for up to 24 hours a day, for a period of six months to years. You can remove it for showering, sports and other activities.A brace works best before a growth spurt because your chest wall is still flexible.

#### **Pectus carinatum surgery**

This involves a technique called the Ravitch procedure. A surgeon makes an incision (cut) in the mid-chest area to remove cartilage in the front of your chest. They make a small cut in the front of your sternum and depress your sternum to a normal position. The surgeon anchors it into position with various techniques including sutures, plates or a bar.

A surgeon can use a minimally invasive surgery that’s similar to a Nuss procedure. They use a metal bar to depress your sternum. They leave the bar in for one or more years and remove it in a separate surgical procedure.

Surgeons have modified these procedures in different ways, but they usually secure the breastbone in some way.

#### **Complications/side effects of the treatment**

Bracing treatment for pectus carinatum is very safe. A small number of people may have irritation or breakdown of the skin where the brace touches it. If this happens, you can stop using the brace at the first sign of any irritation. Then you can ask your provider to adjust the brace.

The surgical repair of pectus carinatum, like other extensive surgeries, carries certain risks. Both the Ravitch and Nuss procedures are safe and effective. But complications can happen, including:

* Pneumothorax (collapsed lung).
* Bleeding.
* Pleural effusion (fluid around your lung).
* Pericarditis (inflammation around your heart).
* Infection.
* Breastbone supports moving out of place.
* Recurrence (return) of pectus carinatum.

### **How long does it take to recover from this treatment?**

After surgery, you may be in the hospital for one to five days. You’ll have a follow-up appointment with your surgeon after a few weeks.

**Outlook / Prognosis**

Pectus carinatum doesn’t adversely affect life expectancy. For most people, it’s primarily a cosmetic issue (although symptoms can occur).

The outlook for people who have pectus carinatum is generally very good. People who wear a brace or have the surgery are usually very satisfied with the results and their appearance.

**Prevention**

No. Until researchers can clearly identify what causes pectus carinatum, you can’t prevent it. Some of the possible causes involve the genes you inherit from your parents, which you can’t change.

**Living With**

People who have pectus carinatum generally live a normal life and shouldn’t limit their activities unless they experience discomfort.

### **When to see a doctor**

You should see a healthcare provider if your pectus carinatum is getting in the way of your daily life. And don’t neglect your mental health. If you’re self-conscious about your chest and it’s affecting your self-esteem, correction can be beneficial.

**DIFFERENTIAL DIAGNOSIS**

* Pectus Excavatum (sunken chest; opposite deformity with inward sternum)
* Jeune Syndrome (congenital dwarfism with small, bell-shaped chest and abnormal ribs)
* Poland Syndrome (unilateral chest wall muscle and rib underdevelopment, sometimes with limb anomalies)
* Congenital Rib Anomalies (extra, absent, fused, or malformed ribs)
* Sternal Cleft and Other Sternum Malformations (partial or complete sternal defects)
* Slipped Ribs Syndrome (rib cartilage displacement causing pain and deformity)
* Jarcho-Levin Syndrome (crab-like chest with multiple vertebral and rib anomalies)
* Scoliosis and Kyphosis (spinal deformities causing chest wall asymmetry)
* Connective Tissue Disorders (e.g., Marfan syndrome, Ehlers-Danlos syndrome, Noonan syndrome, associated with chest wall deformities)
* Trauma or Previous Chest Surgery (acquired chest wall deformities)
* Chest Wall Tumors or Infections (rare causes of localized protrusions or deformities)

**EPIDEMIOLOGY**

* The estimated incidence is approximately 1 per 2,500 live births, with some reports suggesting a prevalence ranging from 0.06% to 0.1% in the general population of children. Other studies using imaging suggest prevalence rates of 2-5% when including mild or asymmetrical cases detected by CT scans.
* It is more common in males, with a reported male-to-female ratio of about 4:1.
* The condition is most frequently diagnosed in mid-childhood to adolescence, often becoming more noticeable during the adolescent growth spurt.
* Racial distribution shows higher frequency in White populations, and it is less common among Black and Asian children.
* There is a family history of chest wall deformities in about 25% to 65% of cases, indicating a genetic predisposition.
* Pectus carinatum may be associated with connective tissue disorders such as Marfan syndrome and Noonan syndrome, as well as congenital heart disease.
* Studies in different regions show some variation: for example, higher proportions of chest wall deformities including pectus carinatum have been reported in Brazil and Argentina compared to other countries.
* The deformity is rarely present at birth but tends to develop or worsen with growth, especially during puberty.

**PREDEFINED QUESTIONS AND ANSWERS**

## What is pectus carinatum?

Pectus carinatum, also called "pigeon chest," is a chest wall deformity where the breastbone (sternum) protrudes outward. It is less common than pectus excavatum and often noticed during growth spurts in adolescence. It may cause cosmetic concerns, chest pain, and sometimes affect heart and lung function. It is often associated with scoliosis.

## What causes pectus carinatum?

The exact cause is unknown, but it is believed to be due to abnormal growth of the rib cartilage. Up to one-third of patients have a family history of chest wall deformities, suggesting a genetic component. It can also be associated with connective tissue disorders.

## How is pectus carinatum diagnosed?

Diagnosis is primarily clinical through physical examination. Imaging such as chest X-rays may be done to assess the chest wall and check for associated scoliosis. Genetic testing may be considered if syndromic features are present.

## What treatments are available for children with pectus carinatum?

* Bracing: A custom-fitted chest-wall brace applies steady pressure to reshape the sternum over time, similar to orthodontic braces. It is most effective in children and teenagers with flexible chest walls and requires wearing the brace 14–23 hours per day for several months to a year.
* Surgery: For severe cases or when bracing fails, surgery such as the Ravitch procedure is performed. This involves removing abnormal cartilage and repositioning the sternum, often with a temporary implanted bar to hold the chest in place for 1–2 years.

## What is recovery like after surgery?

Children typically stay in the hospital 3–5 days post-surgery. Pain management is important during recovery, which usually takes 1–2 months. Activity restrictions apply initially to allow healing. The implanted bar is removed in a later procedure.

## Does pectus carinatum affect life expectancy or cause serious health problems?

Pectus carinatum is generally not dangerous and does not affect life expectancy. Most children have no or mild symptoms. However, it can cause chest pain and psychological distress due to appearance. Treatment can improve symptoms and self-esteem.

## When should treatment be started?

Bracing works best during childhood or early adolescence when the chest wall is still flexible. Doctors often recommend starting treatment after age 10. Surgery is reserved for more severe deformities or when bracing is ineffective.

## Can pectus carinatum improve without treatment?

No, the deformity does not typically resolve on its own and may worsen during growth spurts. Treatment is needed to correct or improve the chest shape

**DOCTOR PATIENT CONVERSATION**

Doctor: Hello! I understand you’re here because you noticed your child’s chest is protruding more than usual. Can you tell me when you first noticed this?

Parent: Yes, it started becoming more obvious over the last year, especially during their growth spurt. The chest kind of sticks out like a bird’s chest, and we’re worried.

Doctor: What you’re describing sounds like pectus carinatum, sometimes called "pigeon chest." It’s a condition where the breastbone and ribs push outward due to overgrowth of the cartilage.

Parent: Is this dangerous? Will it affect my child’s health?

Doctor: In most cases, pectus carinatum doesn’t cause serious health problems. Some children may have mild chest discomfort or breathing difficulties, but often the main concern is cosmetic. It’s also sometimes associated with conditions like scoliosis or connective tissue disorders, so we’ll check for those.

Parent: How do you diagnose it?

Doctor: Usually, we can diagnose it by physical exam. We may take a chest X-ray to see the structure better and check for any spine curvature. If needed, we can also assess lung and heart function.

Parent: What treatment options are there?

Doctor: For children and teens with flexible chest walls, bracing is the first treatment choice. A custom brace applies gentle pressure to reshape the chest over several months to a year. It needs to be worn consistently, often 14 to 23 hours a day.

Parent: What if the brace doesn’t work?

Doctor: If bracing isn’t effective or the deformity is severe, surgery might be recommended. The most common surgery is the Ravitch procedure, which involves removing abnormal cartilage and repositioning the sternum. Recovery usually takes a few weeks to months.

Parent: Will my child need to limit activities?

Doctor: During bracing, children can usually continue normal activities. After surgery, there will be some activity restrictions initially, but most children return to full activity after healing.

Parent: Does the condition get worse over time?

Doctor: It often becomes more noticeable during growth spurts but tends to stabilize once growth is complete. Early treatment during childhood or adolescence gives the best chance for correction.

Parent: Thank you, doctor. This helps me understand what to expect and what we can do.

Doctor: You’re welcome. We’ll work together to develop the best plan for your child and support you throughout treatment. Feel free to reach out with any questions.

REFERENCES

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**TRUNCUS ARTERIOSUS**

**DEFINITION AND DESCRIPTION**

Truncus arteriosus (TRUNG-kus ahr-teer-e-O-sus) is a rare heart condition present at birth. That means it's a congenital heart defect. In this condition, one large blood vessel leads out of the heart, instead of two.

Having only one large blood vessel means that oxygen-poor and oxygen-rich blood mix. This mixing reduces the amount of oxygen delivered to the body. It usually increases the amount of blood flow into the lungs too. The heart has to work harder to adjust for the changes in blood flow.

A baby, also called a fetus, with truncus arteriosus also usually has a hole between the two lower heart chambers, called the ventricles. The hole is called a ventricular septal defect.

**Another name for truncus arteriosus is common arterial trunk.**

**Causes**

Truncus arteriosus occurs as a baby's heart forms during pregnancy. There's often no clear cause. Genetics and environmental factors may play a role.

### **How the heart works**

To understand more about truncus arteriosus, it may be helpful to know how the heart typically works.

The typical heart has four chambers. They are:

* **The right upper chamber, also called the right atrium.** This heart chamber receives oxygen-poor blood from the body.
* **The right lower chamber, also called the right ventricle.** This heart chamber pumps blood into the lungs through a large vessel called the pulmonary artery. The blood flows through the pulmonary artery into smaller vessels in the lungs where blood picks up oxygen.
* **The left upper chamber, also called the left atrium.** This heart chamber receives oxygen-rich blood from the lungs through vessels called pulmonary veins.
* **The left lower chamber, also called the left ventricle.** This chamber pumps the oxygen-rich blood to the body through the body's largest blood vessel, called the aorta.

### **A baby's heart before birth**

The way the unborn baby's heart forms during pregnancy is complex. At some point, there is a single large blood vessel leading out of the heart. The vessel is called the truncus arteriosus. It usually splits in two as the unborn baby grows in the womb. One part becomes the lower end of the body's main artery, called the aorta. The other part becomes the lower part of the pulmonary artery.

But in some babies, the truncus arteriosus never splits. The wall separating the two lower heart chambers hasn't closed all the way. This results in a large hole between those chambers, called a ventricular septal defect.

Babies with truncus arteriosus also often have a problem with the heart valve that controls blood flow from the lower heart chambers to the single vessel. This valve may not close all the way when the heart relaxes. Blood can move the wrong way, back into the heart. This is called truncal valve regurgitation.

**Risk factors**

The exact cause of truncus arteriosus is unknown. But some things might increase the risk of a heart problem at birth. Risk factors include:

* **Viral illness during pregnancy.** Some infections can hurt a developing baby. For example, having German measles during pregnancy can cause changes in a baby's heart development. German measles also is called rubella.
* **Poorly controlled diabetes during pregnancy.** Careful control of your blood sugar before and during pregnancy can reduce the risk of heart problems in your baby. If you have diabetes, work with your healthcare professional to be sure blood sugar is well controlled before getting pregnant.
* **Some medicines taken during pregnancy.** Some medicines can cause heart problems and other health conditions in a baby. Tell your healthcare professional about all the medicines you take, including those bought without a prescription.
* **Some chromosomal disorders.** An extra or irregular chromosome increases the risk of truncus arteriosus. Examples are DiGeorge syndrome, also called 22q11.2 deletion syndrome, and velocardiofacial syndrome.
* **Smoking during pregnancy.** If you smoke, quit. Smoking during pregnancy increases the risk of heart conditions in your baby.
* **Alcohol use.** Drinking alcohol during pregnancy increases the risk of heart conditions and other health issues in a baby.
* **Obesity.** Obesity increases the risk of giving birth to a baby with a heart condition.

**Symptoms**

Symptoms of truncus arteriosus most often occur in the first few days of life. They include:

* Blue or gray skin due to low oxygen levels.
* Extreme sleepiness.
* Poor feeding.
* Poor growth.
* Pounding heartbeat.
* Fast breathing.
* Shortness of breath.

### **When to see a doctor**

If you are worried about your baby's feedings, sleep patterns or growth, contact a healthcare professional for an appointment.

Always seek emergency medical care if a baby has any of the following:

* Blue or gray skin.
* Fast breathing.
* Shallow breathing.
* Any breathing trouble.

## **Diagnosis**

Truncus arteriosus is usually diagnosed soon after a child is born. The baby may look blue or gray and have trouble breathing.

When a baby is born, a healthcare professional always listens to the baby's lungs to check breathing. If a baby has truncus arteriosus, the healthcare professional may hear fluid in the lungs during this exam. The healthcare professional also listens to the baby's heart to check for irregular heartbeats or a whooshing sound, called a murmur.

### **Tests**

Tests to diagnose truncus arteriosus include:

* **Pulse oximetry.** A sensor placed on the fingertip records the amount of oxygen in the blood. Too little oxygen may be a sign of a heart or lung problem.
* **Chest X-ray.** This test shows the condition of the heart and lungs. It can show the size of the heart. A chest X-ray also can tell if the lungs have extra fluid.
* **Echocardiogram.** An echocardiogram uses sound waves to create pictures of the beating heart. This is the main test to diagnose truncus arteriosus. It shows blood flow through the heart and heart valves. In a baby with truncus arteriosus, the test shows one single large vessel leading from the heart. There's typically a hole in the wall between the lower heart chambers.

**Treatment**

Infants with truncus arteriosus need surgery to improve blood flow and oxygen levels. Many procedures or surgeries might be needed, especially as a child grows. Medicines might be given before surgery to help improve heart health.

Children and adults with surgically repaired truncus arteriosus need regular health checkups for life.

### **Medications**

Some of the medicines that might be given before truncus arteriosus surgery include:

* **Water pills.** Also called diuretics, these medicines help the kidneys remove extra fluid from the body. Fluid buildup is a common symptom of heart failure.
* **Positive inotropes.** These medicines help the heart pump stronger, which improves blood flow. They also help control blood pressure. Positive inotropes may be given by IV to treat severe heart failure symptoms.

### **Surgery or other procedures**

Most infants with truncus arteriosus have surgery within the first few weeks after birth. The specific type of surgery depends on the baby's condition. Usually, the baby's surgeon:

* Rebuilds the single large vessel and aorta to create a new, complete aorta.
* Separates the upper part of the pulmonary artery from the single large vessel.
* Uses a patch to close the hole between the two lower heart chambers.
* Places a tube and valve to connect the right lower heart chamber with the upper pulmonary artery. This creates a new, complete pulmonary artery.

The tube used to create the new pulmonary artery doesn't grow with a child. Follow-up surgeries are needed to replace the tube as the child grows.

Future surgeries may be done with a flexible tube called a catheter. This avoids the need for open-heart surgery. The healthcare professional inserts the catheter into a blood vessel in the groin and guides it to the heart. A new valve can be delivered through the catheter to the proper area.

Sometimes a small balloon at the tip of the catheter is inflated at the site of a blockage, making a blocked artery wider. This procedure is called balloon angioplasty.

After surgery for truncus arteriosus, a person needs lifelong follow-up care with a heart doctor specializing in congenital disease. This type of healthcare professional is called a congenital cardiologist.

**Lifestyle and home remedies**

If you or your child had truncus arteriosus, your healthcare professional may recommend taking a few steps to protect the heart.

* **Exercise limits.** Some people with heart conditions need to limit exercise and sports activities, especially competitive sports. Ask your healthcare professional which sports and types of exercise are safe for you or your child. People with Eisenmenger syndrome should avoid strenuous physical activity.
* **Antibiotics.** Sometimes, heart conditions can increase the risk of infection in the lining of the heart or heart valves. This infection is called infective endocarditis. Antibiotics may be recommended to prevent infections before dental procedures and other surgeries. It's also important to have good oral hygiene and regular dental checkups
* **Pregnancy.** If you've had truncus arteriosus repair surgery and want to become pregnant, talk to your healthcare professional first. Ask about the possible risks and complications. It's best to be checked by healthcare professionals with training in adult congenital heart disease and high-risk pregnancies. Together, you and your care team can discuss and plan for any special care needed during pregnancy.

Depending on the level of lung damage that occurred before truncus arteriosus surgery, pregnancy might or might not be recommended. Pregnancy is considered very high risk for those with Eisenmenger syndrome and is not recommended.

**Complications**

Truncus arteriosus causes severe problems with how blood flows through the lungs, heart and rest of the body.

Complications of truncus arteriosus in babies include:

* **Breathing problems.** Extra fluid and blood in the lungs can make it difficult to breathe.
* **High blood pressure in the lungs, also called pulmonary hypertension.** This condition causes the blood vessels in the lungs to narrow. It becomes hard for the heart to pump blood into the lungs.
* **Enlargement of the heart.** Pulmonary hypertension and increased blood flow strain the heart. The heart must work harder to pump blood. This causes the heart muscle to grow larger. The enlarged heart gradually weakens.
* **Heart failure.** In this condition, the heart cannot supply the body with enough blood. Too little oxygen and too much strain on the heart can lead to heart failure.

Infants who had their hearts successfully fixed with surgery may still have complications later in life. Possible complications are:

* Pulmonary hypertension that gets worse.
* Backward flow of blood through a heart valve, called regurgitation.
* Irregular heartbeats, called arrhythmias.

Common symptoms of these complications include:

* Dizziness.
* Feeling very fast, fluttering heartbeats.
* Feeling very tired.
* Shortness of breath when exercising.
* Swelling of the belly, legs or feet.

### **Truncus arteriosus in adults**

In rare cases, some people born with truncus arteriosus can survive without heart surgery. They may live into adulthood. But those with the condition will almost certainly have heart failure and develop a complication called Eisenmenger syndrome. This syndrome is caused by permanent lung vessel damage. It results in a significant lack of blood flow to the lungs.

**Prevention**

Because the cause is unclear, it may not be possible to prevent truncus arteriosus. Getting good prenatal care is important. If you or someone in your family had a heart condition present at birth, talk to your healthcare professional before getting pregnant. You might need to see a genetic counselor and a heart doctor, called a cardiologist.

If you decide to get pregnant, taking these steps can help keep your baby healthy:

* **Get recommended vaccinations.** Some infections can be harmful to a developing baby. For example, having German measles — also called rubella —during pregnancy can cause changes in a baby's heart development. A blood test done before pregnancy can show if you're immune to rubella. A vaccine is available for those who aren't immune.
* **Talk to your healthcare professional about your medicines.** Check with your healthcare professional before taking any medicines if you're pregnant or thinking about getting pregnant. Many drugs aren't recommended for use during pregnancy because they can harm a developing baby.
* **Take a folic acid supplement.** Take a multivitamin with folic acid. Taking 400 micrograms of folic acid daily has been shown to reduce brain and spinal cord conditions in babies. It may help reduce the risk of heart conditions present at birth too.
* **Control diabetes.** If you have diabetes, ask your healthcare professional how to best manage the disease during pregnancy.

## **Outlook / Prognosis**

### **Can a baby survive with truncus arteriosus?**

Yes, but they need surgical repair to survive. The survival rate for truncus arteriosus surgery is 80% to 97%, according to the latest research. Survival depends on many factors, including the complexity of a baby’s heart anatomy.

About 75% of babies who have surgical repair are alive 20 years later. Most deaths occur within one year of repair. About 92.5% of babies who survive to one year after surgery are alive at 20 years.

Your child’s care team can tell you more about the factors that might affect your child’s life expectancy.

### **Can you live a normal life with truncus arteriosus?**

There’s limited research on how truncus arteriosus (after surgical repair) affects a person’s quality of life. One study showed children have reduced exercise tolerance and a lower health-related quality of life compared to their peers. Another study showed adults have reduced physical functioning but a similar quality of life as their peers overall.

What it means to have a “normal life” can vary from person to person. Quality of life is a subjective experience. It’s important to talk to your child about how they’re feeling — physically and emotionally. As they get older, encourage them to ask their doctors questions. Even when you don’t have all the answers or know the “right” thing to say, simply being there for your child will help them feel loved and supported.

## **Diagnostic Considerations**

## Important considerations

Note the following:

* It is important for clinicians to properly diagnose truncus arteriosus.
* Consider various abnormalities that may be associated with truncus arteriosus, some of which may have an impact on management and outcome.
* Also consider potential issues that are of particular concern in patients with truncus arteriosus, including pulmonary hypertensive crisis and volume overload in patients with persistent truncal valve regurgitation.
* Obtain routine laboratory and imaging studies (to include a full complement of echocardiographic views) in the neonate with truncus arteriosus to aid in determining therapeutic strategy and assist diagnosis.
* Consult with a cardiologist before beginning, changing, or discontinuing cardiac medications in these patients.

## Postnatal diagnosis

For postnatal diagnosis, truncus arteriosus is suggested by the history and physical findings, along with abnormal results on the critical congenital heart disease screening test in the first few days after birth, showing preductal and postductal oxygen saturation less than 95% with mild or unnoticeable cyanosis. Unscreened infants present within the first 2 weeks for evaluation of a heart murmur, or with symptoms of congestive heart failure resulting from increased blood flow to the lungs. These infants exhibit poor feeding, lethargy, tachypnea, costal-sternal retractions, grunting, nasal flaring, tachycardia, or hepatomegaly.

## Other conditions to consider

Consider the following in the differential diagnosis:

* Aortopulmonary septal defect
* Double outlet right ventricle with normally related great arteries
* Shock
* Tetralogy of Fallot with pulmonary atresia
* Total anomalous pulmonary venous connection

## **Differential Diagnoses**

* Bacterial Sepsis
* Double Outlet Right Ventricle with Transposition
* Pediatric Hypoplastic Left Heart Syndrome
* Pediatric Metabolic Acidosis
* Shock and Hypotension in the Newborn
* Tetralogy of Fallot with Absent Pulmonary Valve
* Transposition of the Great Arteries

## **Epidemiology**

An estimated incidence of 7 cases of truncus arteriosus occurs per 100,000 live births each year, comprising less than 1% of all congenital heart diseases but about 4% of critical congenital heart defects.

### United States data

Truncus arteriosus represents 1-2% of congenital heart defects in liveborn infants. Based on an estimated incidence of congenital heart disease of 6-8 per 1,000 liveborn children, truncus arteriosus occurs in approximately 5-15 of 100,000 live births. Among aborted fetuses and stillborn infants with cardiovascular anomalies, truncus arteriosus represents almost 5% of defects.

A population-based review of all cases of live births from 1999 to 2008 identified as having severe congenital heart disease from the Nationwide Inpatient Sample (NIS) database indicated a decrease of the conditions over the study period.There was a significant decreased incidence of truncus arteriosus as well as tetralogy of Fallot, pulmonary atresia, and hypoplastic left heart syndrome; however, these trends varied with sociodemographic factors. The investigators suggested a possible reason for the decreasing prevalence trend was the increased numbers of terminated fetuses with prenatally diagnosed congenital heart disease.

### International data

No significant difference in the incidence of truncus arteriosus is noted among those born in the United States compared with other countries (1-4% of congenital heart conditions.

However, a Canadian longitudinal study (1983-2010) of all individuals with congenital heart disease noted a more than 50% increase of severe and other congenital heart disease after the year 2000, with adults comprising two thirds of the cases by 2010.The prevalence of congenital heart disease in the first year of life between 1998 and 2005 was 8.21 per 1000 live births; in 2010, the overall prevalence was 13.11 per 1000 in children and 6.12 per 1000 in adults. A temporal increase in prevalence of congenital heart disease and severe congenital heart disease was noted for children and adults.

In a German retrospective study (2019-2024) of truncus arteriosus diagnosed prenatally at a local tertial referral center, 11 of 14 neonates survived to birth, of whom 7 had postnatal confirmation and underwent surgery, with an overall 55% survival. When the investigators performed a literature review, they noted there were 576 prenatal diagnoses of truncus arteriosus and 247 postnatal diagnoses, as well as 27% of tested cases demonstrated a 22q11 deletion.

### Race-, sex-, and age-related demographics

Based on limited data, no racial predilection is apparent.

Although many series report a slight male predominance, no significant predilection based on sex is apparent.

Truncus arteriosus is a congenital anomaly that is present from early in embryonic gestation. Currently, truncus arteriosus is diagnosed using prenatal ultrasonography in a small percentage of patients. Among patients diagnosed after birth, the median age at presentation is generally a few days, which is significantly earlier than was the case 20 or more years ago. Occasionally, patients are not diagnosed until later in infancy, childhood, or even adulthood, although such cases are exceedingly rare in the United States and Europe.

## Procedures

### Cardiac catheterization

For individuals with a late presentation, the ESC recommends cardiac catherization for evaluation of the pulmonary vascular resistance and for testing of the pulmonary vascular reactivity (class I).

Standard angiographic images from the truncal root can aid in the assessment of the coronary arterial anatomy and complex pulmonary artery anatomy arising from the thoracic aorta, if echocardiography is inadequate, and in the assessment of regurgitation through the truncal valve. Cardiac catheterization is important in decision-making regarding the time and type of surgery to be performed (palliative vs corrective).

Cardiac catheterization is generally not required prior to repair in neonates and young infants with truncus arteriosus.

Catheterization is an important tool for evaluating some of the most common anatomic problems in patients with repaired truncus arteriosus, such as obstruction of the surgical reconstructed right ventricular outflow tract, branch pulmonary arterial stenosis, truncal valve regurgitation, and, in patients repaired later in life, pulmonary vascular obstructive disease.

### Transcatheter balloon dilation

Treatment of pulmonary outflow tract or pulmonary arterial obstruction with transcatheter balloon dilation or stenting is an effective therapy for these problems in many patients who have undergone complex surgical repair.

### Biopsy

Histologic examination is not generally indicated in the evaluation of patients with truncus arteriosus. In the rare older patient with evidence of elevated pulmonary vascular resistance, pulmonary biopsy is occasionally performed as a means of assessing the extent of pulmonary vascular obstructive disease.

**TREATMENT DRUG AND THEIR SIDE EFFECTS**

## 1. Diuretics (e.g., Furosemide)

* Purpose: Reduce fluid overload and pulmonary congestion by promoting urine output, helping to relieve symptoms of congestive heart failure.
* Side Effects: Electrolyte imbalances (low potassium, sodium), dehydration, kidney dysfunction, increased urination.

## 2. Inotropic Agents (e.g., Digoxin, Milrinone)

* Purpose: Improve heart muscle contractility to enhance cardiac output and support circulation in heart failure.
* Side Effects:
  + *Digoxin:* Narrow therapeutic window; risk of arrhythmias, nausea, vomiting, visual disturbances. Requires monitoring of drug levels.
  + *Milrinone:* May cause low blood pressure, arrhythmias, headache.

## 3. Afterload-Reducing Agents (e.g., ACE inhibitors like Captopril or Enalapril)

* Purpose: Decrease resistance against which the heart pumps, improving cardiac output and reducing heart failure symptoms.
* Side Effects: Hypotension, kidney impairment, cough, elevated potassium levels.

## 4. Prostaglandin E1 (PGE1)

* Purpose: Maintains ductus arteriosus patency in cases where systemic or pulmonary blood flow depends on ductal circulation (e.g., associated arch obstruction).
* Side Effects: Apnea (especially in neonates), flushing, low blood pressure.

## Surgical Treatment

* Definitive treatment is early surgical repair, usually within the first few weeks of life, involving:
  + Closure of the ventricular septal defect (VSD)
  + Separation of the pulmonary arteries from the common arterial trunk
  + Reconstruction of the aorta
  + Placement of a conduit with a valve between the right ventricle and pulmonary arteries.
* Lifelong follow-up is needed, and multiple surgeries or catheter-based interventions may be required as the child grows because conduits do not grow with the patient.

**PREDEFINED QUESTIONS AND ANSWERS**

## What is truncus arteriosus?

Truncus arteriosus is a rare congenital heart defect where a baby is born with a single large artery (truncus) coming out of the heart instead of two separate arteries—the aorta and the pulmonary artery. This causes oxygen-rich and oxygen-poor blood to mix and flow to the body and lungs together.

## What causes truncus arteriosus?

It happens when the fetal heart’s large vessel fails to divide properly during development. Some cases are linked to genetic conditions like 22q11 deletion syndrome (DiGeorge syndrome), but often the exact cause is unknown.

## What symptoms does truncus arteriosus cause?

Symptoms usually appear in the first days or weeks of life and include:

* Blue or purple tint to lips, skin, and nails (cyanosis)
* Rapid or difficult breathing
* Poor feeding and poor weight gain
* Sweating, especially during feeding
* Fatigue and sleepiness
* Heart murmur heard by a doctor.

## How is truncus arteriosus diagnosed?

Diagnosis involves:

* Physical exam and pulse oximetry to check oxygen levels
* Echocardiogram (heart ultrasound) to visualize the heart’s structure
* Chest X-ray and sometimes cardiac MRI or catheterization for detailed assessment.

## How is truncus arteriosus treated?

* Surgery is required, usually within the first few weeks of life. It involves closing the ventricular septal defect (VSD), separating the pulmonary arteries from the truncus, and connecting them to the right ventricle using a conduit with a valve.
* Before surgery, medical management includes medications to support heart function and breathing support if needed. Nutritional support may be necessary for infants with feeding difficulties.

## What is the outlook for children with truncus arteriosus?

Surgical repair improves survival significantly, but some children may require additional surgeries or interventions as they grow. Lifelong follow-up with a cardiologist experienced in congenital heart disease is essential.

## Is truncus arteriosus associated with other conditions?

Yes, about one-third of children with truncus arteriosus have 22q11 deletion syndrome (DiGeorge syndrome), which may cause other health issues and requires multidisciplinary care

**DOCTOR PATIENT CONVERSATION**

Doctor: Hello, I want to talk with you about your baby’s heart condition called truncus arteriosus. Do you have any questions so far?

Parent: Yes, we are trying to understand what it means. What exactly is truncus arteriosus?

Doctor: Normally, the heart has two separate arteries coming out: the aorta, which carries oxygen-rich blood to the body, and the pulmonary artery, which carries oxygen-poor blood to the lungs. In truncus arteriosus, there is only one large artery coming out of the heart instead of two. This means oxygen-rich and oxygen-poor blood mix together and go to both the lungs and the body.

Parent: What symptoms should we expect in our baby?

Doctor: Babies with this condition often appear blue or have a bluish tint to their lips and skin because their blood doesn’t carry enough oxygen. They may breathe rapidly or have difficulty feeding and gaining weight. The heart has to work harder, which can cause sweating and fatigue.

Parent: How is this diagnosed?

Doctor: We use an echocardiogram, which is an ultrasound of the heart, to see the structure and function. It shows the single artery and the hole between the heart’s lower chambers, called a ventricular septal defect. Chest X-rays and other tests may also be done to assess the heart size and lung circulation.

Parent: What treatment does our baby need?

Doctor: Surgery is necessary, usually within the first few weeks of life. The surgery closes the hole between the ventricles, separates the pulmonary arteries from the common artery, and connects them to the right ventricle using a tube graft. Before surgery, we may use medications to support the heart and keep blood flowing properly.

Parent: What is the outlook after surgery?

Doctor: Many children do well after surgery, especially if they survive the early post-operative period. However, they need lifelong follow-up with a cardiologist because additional surgeries or interventions may be needed as they grow.

Parent: Is this condition related to any genetic problems?

Doctor: Sometimes, truncus arteriosus is associated with a genetic condition called 22q11 deletion syndrome, which can cause other health issues. We can arrange genetic counseling and testing if needed.

Parent: Thank you for explaining. What should we watch for at home?

Doctor: Watch for worsening blue color, rapid breathing, poor feeding, or lethargy. If you notice any of these, seek medical attention promptly.

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### **Pleural effusion**

**DEFINITION AND DESCRIPTION**

Pleural effusion, which some people call “water on the lungs,” is the buildup of excess fluid between the layers of the pleura outside your lungs. The pleura are thin membranes that line your lungs and the inside of your chest cavity.

Normally, everyone has a small amount of fluid in their pleura. This fluid acts as a natural lubricant and makes it easier for your lungs to move when you breathe. But with pleural effusion, you have too much fluid around your lungs. This means your body is producing too much of the fluid or not absorbing enough of the fluid it makes.

#### **Types of pleural effusion**

Healthcare providers split pleural effusion into two types, depending on the kind of fluid around your lungs.

Excess fluid may be:

* **Protein-poor and watery (transudative)**. Fluid of this kind comes from cirrhosis or heart failure, for example. This type of pleural effusion happens when there’s an increase in pressure from the fluid.
* **Protein-rich (exudative)**. Fluid of this kind comes from cancer or an infection, for example. This type of pleural effusion happens because too much fluid is getting through your smallest blood vessels, or your lymphatic system isn’t draining enough.

Treatment varies by type and cause.

Pleural effusions are very common. Healthcare providers find pleural effusions in about 1.5 million people in the United States each year.

**Symptoms and Causes**

You may have unrelated symptoms due to the disease or condition that caused pleural effusion. Pleural effusion symptoms include:

* Chest pain. Coughing or deep breathing makes it worse.
* Dyspnea (shortness of breath, or difficult, labored breathing).
* Orthopnea (the inability to breathe easily unless you’re sitting up straight or standing up straight).

Some people with pleural effusion have no symptoms. They find out they have pleural effusion when they have a chest X-ray for another reason.

### **What causes pleural effusion?**

There are many causes of pleural effusion, and some people have more than one.

Depending on the cause, the excess fluid may be either protein-poor (transudative) or protein-rich (exudative). These two categories help providers determine the cause of the pleural effusion. A pulmonary embolism (PE) can fall under either category.

**The most common causes of transudative (watery fluid) pleural effusions include:**

* Heart failure.
* Cirrhosis.
* Nephrotic syndrome (a kidney issue).

**Common causes of exudative (protein-rich fluid) pleural effusions include:**

* Pneumonia.
* Cancer (lung cancer, breast cancer or lymphoma).
* Kidney disease.
* Inflammatory disease.
* Post open-heart surgery.

**Less common causes of pleural effusion include:**

* Tuberculosis.
* Autoimmune disease.
* Bleeding from chest trauma.
* Chylothorax (chyle from your lymphatic system after trauma).
* Rare chest and abdominal infections.
* Exposure to asbestos.
* Esophageal rupture.
* Pancreatitis.
* Meig’s syndrome (from a benign ovarian tumor).
* Ovarian hyperstimulation syndrome.
* Certain medications.
* Abdominal surgery.
* Radiation therapy.

In some cases, the fluid itself may have malignant (cancerous) cells or may be a direct result of chemotherapy.

#### **Risk factors for pleural effusion**

Risk factors for pleural effusion include:

* Medical conditions that cause it.
* Tobacco products.
* Exposure to asbestos.

### **Complications of pleural effusion**

Pleural effusion can lead to:

* An infection that becomes an abscess.
* Scarring around the lungs.
* Damage to your lungs.

## **Diagnosis and Tests**

A provider will ask you about your medical history. They’ll ask what other illnesses you have and when your symptoms started. They’ll do a physical exam, which includes listening to your lungs when you breathe. Next, they’ll order tests.

Healthcare providers use these tests to diagnose and evaluate pleural effusion:

* Chest X-ray.
* Computed tomography (CT) scan of your chest.
* Ultrasound of your chest.
* Thoracentesis or biopsy (inserting a needle between your ribs to remove a fluid sample).
* Pleural fluid analysis (examining the fluid from the pleural space).

If less invasive tests don’t diagnose pleural effusion, you may need a thoracoscopy. Thoracoscopy is a minimally invasive technique, also known as video-assisted thoracic surgery, or VATS. A provider performs this while you’re under general anesthesia. Thoracoscopy allows them to see and evaluate your pleura. Often, they’ll treat your effusion during the thoracoscopy.

**Management and Treatment**

Treatment of pleural effusion focuses on getting the excess fluid out and keeping it from collecting again. Providers choose pleural effusion treatment based on the underlying condition and whether the effusion is making it hard for you to breathe.

#### **Medicines**

Providers use diuretics and other heart failure medications to treat pleural effusion from congestive heart failure or other medical causes. Some people need antibiotics. For a malignant effusion, you may also need treatment with chemotherapy, radiation therapy or a medication infusion within your chest.

#### **Procedures**

A provider can use therapeutic thoracentesis or a chest tube to drain a pleural effusion that’s causing respiratory symptoms.

Even with drainage, you may have pleural effusions that are hard to control or that come back due to a malignancy. In this case, a provider puts a sclerosing agent (a type of drug that purposely creates scarring) into your pleural cavity through a chest tube. This causes fibrosis (excessive fibrous tissue) of the pleura (pleural sclerosis). This is 50% successful in preventing pleural effusions from happening again.

#### **Surgery**

You may need surgery if drainage or pleural sclerosis don’t work. Your surgeon will carefully evaluate you and discuss the possible risks and benefits of surgical pleural effusion treatments.

The two types of surgery include:

* **Video-assisted thoracoscopic surgery (VATS).** This minimally invasive approach uses one to three small (half-inch) incisions (cuts) in your chest. VATS can manage pleural effusions that are difficult to drain or that come back because of a tumor. A provider can insert sterile talc or an antibiotic during surgery to prevent fluid from building up again.
* **Thoracotomy (traditional, “open” thoracic surgery)**. A surgeon performs a thoracotomy through a 6-inch to 8-inch incision in your chest. They use this approach when you have an infection. A thoracotomy removes all of the fibrous tissue and helps clear out the infection from the pleural space. You’ll need chest tubes for two days to two weeks after surgery to keep draining fluid.

#### **Complications/side effects of the treatment**

Complications of treatment may include:

* Pulmonary edema.
* Blood clots.
* Abnormal heart rhythms.
* Pneumothorax (collapsed lung).

### **How long does it take to recover from this treatment?**

You may need to take it easy for two days after a thoracentesis. If you have VATS, you may need to spend a few days in the hospital afterward. After a thoracotomy (open surgery), you’ll probably be in the hospital for a week.

**Outlook / Prognosis**

You may have follow-up X-rays after you receive treatment. Your provider will want to make sure the treatment worked well. You’ll also need treatment for the medical condition that caused pleural effusion.

The seriousness of your condition depends on:

* The main cause of pleural effusion.
* If it’s affecting your breathing.
* If it responds well to treatment.

### **Outlook for pleural effusion**

The outlook or prognosis varies depending on what caused your pleural effusion and what other conditions you have. For some people, pleural effusion treatment is successful. But pleural effusions can happen again. The outlook isn’t good when you have a pleural effusion from cancer. It’s also not good if you don’t get treatment for pleural effusion.

**Prevention**

You may not be able to avoid the many causes of pleural effusion. But you can lower your risk of pleural effusions in these ways:

* Avoiding tobacco products.
* Avoiding asbestos.
* Sticking to low-salt meals if your provider instructs you to.
* Taking prescribed diuretics (water pills).
* Limiting fluid intake if your provider tells you to.

## **Living With**

Some medical conditions that cause pleural effusion require long-term treatment. Be sure to talk with your provider about the best treatment for your chronic (long-term) condition. Then, stick to the treatment plan.

### **When should I see my healthcare provider?**

Go to all your follow-up appointments to manage your chronic condition. Get immediate help if you’re having trouble breathing.

## **Diagnostic Considerations**

Tests may need to be ordered to rule out immune dysfunction or other underlying systemic or local pulmonary disorders that cause empyema. Other conditions to consider in the differential diagnosis of pleural effusion include the following:

* Chest mass
* Pneumonia with pleurisy
* Pleural thickening

## **Differential Diagnoses**

* Atelectasis
* Lung Abscess
* Pediatric Pneumonia

## Infectious Causes

* Parapneumonic effusion and empyema (most common; associated with bacterial pneumonia)
* Tuberculous pleuritis (especially in endemic areas; lymphocyte-predominant pleural fluid, elevated ADA)
* Viral infections (adenovirus, influenza)
* Fungal infections (rare, e.g., histoplasmosis)
* Parasitic infections (e.g., hydatid cyst disease)

## Non-Infectious Causes

* Congestive heart failure (transudative effusion due to increased hydrostatic pressure)
* Nephrotic syndrome (hypoalbuminemia causing transudative effusion)
* Liver cirrhosis (hepatic hydrothorax)
* Malignancies (primary pleural tumors or metastatic disease causing exudative effusion)
* Collagen vascular diseases (e.g., systemic lupus erythematosus)
* Trauma or post-surgical effusions
* Chylothorax (lymphatic leakage causing milky pleural fluid)
* Pancreatitis (rare cause of pleural effusion)

## Other Considerations

* Chest wall masses or tumors causing secondary effusion
* Pulmonary embolism (rare in children but possible)
* Foreign body aspiration with secondary infection

## **Epidemiology**

Parapneumonic effusions and empyema are more common in boys than girls.In addition, parapneumonic effusions and mpyema are more commonly encountered in infants and young children than in older children. In a Spanish study, children younger than 5 years had a higher incidence of empyema than did children aged 5-17 years.

### Occurrence in the United States

Pleural effusion in children is usually a manifestation of an underlying disorder, and its prevalence mirrors that of the underlying disease. Empyema was reported in about 0.6-2% of children with bacterial pneumonia.The prevalence of pleural infections appears to be increasing in some industrialized countries. In the United States, the empyema-associated hospitalization rate increased 70% between 1997 (2.2 per 100,000) and 2006 (3.7 per 100,000 children).

Byington et al reported that a significant increase in the incidence of empyema in children, from 1 case per 100,000 children to 14 cases per 100,000 children, occurred in Utah between 1993 and 2003.

### International occurrence

Information on the incidence of pleural effusion in children is limited. As in the United States, infectious agents are the most common cause of pediatric pleural effusion internationally. The distribution of pleural effusion depends on the population studied.

In Spain, the incidence of parapneumonic effusion in children younger than age 5 years increased from 1.7 per 100,000 (in 1999) to 8.5 per 100,000 (in 2004).In France, the incidence of empyema increased from 0.5 per 100,000 (in 1995) to 13 per 100,000 (in 2003).

## A**ntibiotic Therapy**

In the earlier stages of parapneumonic effusion formation (mild symptoms, short duration), institution of appropriate empiric antibiotics, based on the patient's age and the organisms and sensitivities commonly present in the community, may discourage a small effusion from developing into a complicated parapneumonic effusion. Whenever possible, a pleural fluid sampling should be performed prior to the initiation of antibiotics.

If a causative organism is identified, antibiotic choice should be guided by the sensitivity pattern of the organism.

Some groups of antibiotics (e.g., penicillins, cephalosporins, aztreonam, clindamycin, and ciprofloxacin) exhibit more satisfactory pleural fluid penetration than others (e.g., aminoglycosides).

In a hospitalized patient with complicated parapneumonic effusion, antibiotics are commonly administered intravenously while a thoracostomy tube is present and the patient is febrile. No data from randomized trials on an appropriate length of treatment are available, and no data on whether different organisms require different durations are noted.Many centers continue with intravenous antibiotics at least 48 hours after the patient is afebrile and the chest drain is removed. Thereafter, oral antibiotics are commonly continued for 2-4 weeks.

A study by Tagarro et al that included 60 randomized children with community-acquired pneumonia and pleural effusion reported that patients receiving dexamethasone along with antibiotics had a shorter time to recovery than the placebo group

**PREDEFINED QUESTIONS AND ANSWERS**

## What is pleural effusion in children?

Pleural effusion is when excess fluid builds up in the pleural space—the thin area between the lungs and chest wall. This fluid can cause chest pressure, difficulty breathing, and low oxygen levels.

## What causes pleural effusion in children?

The most common cause is lung infection or pneumonia. Other causes include heart failure, kidney or liver problems, chest injury, inflammation (like pancreatitis), surgery, and some rare diseases.

## What symptoms might my child have?

Symptoms can include:

* Shortness of breath or rapid breathing
* Cough
* Sharp chest pain that worsens with coughing or deep breaths
* Fever and chills
* Fatigue
* Sometimes hiccups or abdominal pain.

## How is pleural effusion diagnosed?

Doctors listen to the lungs and may notice decreased breath sounds. Diagnosis is confirmed with:

* Chest X-rays (sometimes in different positions)
* Ultrasound or CT scan for detailed imaging
* Pleural fluid analysis obtained by needle drainage to identify infection or other causes.

## How is pleural effusion treated?

* Small effusions often improve with treatment of the underlying cause, such as antibiotics for pneumonia.
* Large or infected effusions (empyema) may require drainage with a chest tube.
* In some cases, surgery or minimally invasive procedures (like VATS) may be needed to remove thick fluid or pus.
* Supportive care includes oxygen and pain management.

## What is empyema?

Empyema is when the pleural fluid becomes infected and thickens into pus. It usually requires more intensive treatment like drainage and longer antibiotic courses.

## What is the outlook for children with pleural effusion?

Most children recover well with prompt treatment. Early drainage of infected fluid reduces complications and improves outcomes. The prognosis depends on the underlying cause and treatment.

**DOCTOR PATIENT CONVERSATION**

Doctor: Hello, I want to discuss your child’s recent diagnosis of pleural effusion. Do you have any questions so far?

Parent: Yes, can you explain what pleural effusion means?

Doctor: Certainly. Pleural effusion is when excess fluid builds up in the space between the lungs and the chest wall. This fluid can make it harder for your child to breathe because it restricts lung expansion.

Parent: What causes this fluid to build up?

Doctor: In children, the most common cause is infection, like pneumonia. Other causes include heart problems, kidney or liver diseases, or sometimes inflammation or injury. We’ll do tests to find out the exact cause.

Parent: What symptoms should we watch for?

Doctor: Your child might have difficulty breathing, cough, chest pain that worsens with deep breaths, or fever if infection is involved. Sometimes they may feel tired or have less appetite.

Parent: How do you confirm the diagnosis?

Doctor: We use chest X-rays and ultrasound to see the fluid. Sometimes we take a sample of the fluid with a needle to test for infection or other causes.

Parent: How is it treated?

Doctor: Treatment depends on the cause. If it’s due to pneumonia, antibiotics are given. If the fluid is large or infected, we may need to drain it using a small tube. Small effusions might just be monitored.

Parent: Is the procedure painful?

Doctor: We use local anesthesia to minimize discomfort. Your child might feel some pressure but it’s generally well tolerated.

Parent: What is the outlook?

Doctor: Most children recover well with treatment. Early drainage of infected fluid helps prevent complications and improves breathing.

Parent: Thank you, doctor. That helps me understand what’s going on.

Doctor: You’re welcome. Please call us if your child’s breathing worsens, fever persists, or if you have any concerns.

REFERENCES

[Pleural Effusion: Symptoms, Causes & Treatment](https://my.clevelandclinic.org/health/diseases/17373-pleural-effusion#overview)

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## Pneumococcal Pneumonia

**DEFINITION AND DESCRIPTION**

Pneumococcal pneumonia is a potentially serious bacterial lung disease you shouldn't ignore. It can disrupt your life for weeks and even land you in the hospital.

Cause: Pneumococcal pneumonia is a bacterial lung infection caused by *Streptococcus pneumoniae*, a common cause of pneumonia in children worldwide.

Symptoms: Children may present with cough, fever, rapid or difficult breathing, chest pain (especially with coughing or deep breaths), lethargy, and sometimes bluish discoloration of lips or nails due to low oxygen levels.

* Diagnosis: Clinical evaluation includes physical exam and listening to the lungs. Chest X-ray helps confirm pneumonia and assess severity. Laboratory tests and cultures may be done in severe cases.
* Treatment:
  + The first-line antibiotic for treating pediatric pneumonia, including pneumococcal pneumonia, is amoxicillin (often as dispersible tablets), given orally in mild to moderate cases.
  + Severe cases may require hospitalization with intravenous antibiotics and supportive care such as oxygen.
  + Treatment duration typically lasts several days, and improvement is usually seen within 24-48 hours of starting antibiotics.
* Prevention:
  + Vaccination with pneumococcal conjugate vaccines (PCV15 or PCV20) starting at 2 months of age is highly effective in preventing pneumococcal disease.
  + Other vaccines like Hib, measles, and influenza vaccines also reduce pneumonia risk.
  + Exclusive breastfeeding for the first 6 months and reducing indoor air pollution improve natural defenses.
  + Good hygiene and nutrition further reduce risk.
* Prognosis: Most children recover fully with timely antibiotic treatment. Severe infections can lead to complications requiring hospitalization.

## **Epidemiology**

### United States statistics

Pneumonia can occur at any age, although it is more common in younger children. Pneumonia accounts for 13% of all infectious illnesses in infants younger than 2 years of age. In a large community-based study conducted by Denny and Clyde, the annual incidence rate of pneumonia was 4 cases per 100 children in the preschool-aged group, 2 cases per 100 children aged 5-9 years, and 1 case per 100 children aged 9-15 years.

Thompson et al reported that, after elderly persons, the second highest rates of influenza-associated hospitalizations in the United States were in children younger than 5 years of age.These investigators evaluated annual influenza-associated hospitalizations by hospital discharge category, discharge type, and age group.

In a randomized double-blind trial, the heptavalent pneumococcal vaccine reduced the incidence of clinically diagnosed and radiographically diagnosed pneumonia among children younger than 5 years of age by 4% and 20%, respectively.Although the overall rate of pneumonia has decreased in the United States with the use of the 7-valent vaccine, the rate of empyema and complicated pneumonia has increased.Following the use of the 13-valent conjugated pneumococcal polysaccharide vaccine, the overall rates of pneumonia are anticipated to drop further. The new vaccine includes serotypes that have become associated with complicated or antibiotic-resistant disease (19A and 6A, for example).

Among school-aged children and adolescents, bronchopneumonia occurs in 0.8-2% of all pertussis cases and 16-20% of hospitalized cases. *M pneumoniae* accounts for 14-35% of pneumonia hospitalizations in this age group,and mycobacterial pneumonia has recently been noted with increasing frequency in some inner-city areas, particularly among children in homeless shelters and group homes and those with household contacts.

### International statistics

Pneumonia and other lower respiratory tract infections are the leading cause of death worldwide. The WHO Child Health Epidemiology Reference Group estimated the median global incidence of clinical pneumonia to be 0.28 episodes per child-year.This corresponds to an annual incidence of 150.7 million new cases, of which 11-20 million (7-13%) are severe enough to require hospital admission. Ninety-five percent of all episodes of clinical pneumonia in young children worldwide occur in developing countries.

Approximately 150 million new cases of pneumonia occur annually among children younger than 5 years of age worldwide. This accounts for approximately 10-20 million hospitalizations.A WHO Child Health Epidemiology Reference Group publication cited the incidence of community-acquired pneumonia among children younger than 5 years of age in developed countries as approximately 0.026 episodes per child-year. In addition, a study conducted in the United Kingdom showed that 59% of deaths from pertussis are associated with pneumonia.

## **Diagnostic Considerations**

Pneumonia can occur at any age, although it is more common in younger children. Different age groups tend to be infected by different pathogens, which affects diagnostic and therapeutic decisions.

Many patients referred for evaluation for recurrent pneumonia are diagnosed with asthma. In emergency department studies, 35% of children with an asthma exacerbation have abnormalities visible on chest radiographs. In a child not yet diagnosed with asthma, these abnormalities are frequently interpreted simply as pneumonia. Inflammation, often triggered by viral infection, is part of the asthmatic response. Wheezing responsive to bronchodilators, a history of atopy, a family history of asthma, and a history of cough or wheeze with exercise may be helpful in identifying such patients.

Consider any other diseases that may present with respiratory dysfunction in the first 24 hours of life. Keep in mind that any of the conditions listed below may also be superimposed by pneumonia:

* Alveolar-capillary dysplasia
* Arrhythmia
* Asphyxia
* Bronchial duplication
* Chest wall injury or anomaly
* Choanal atresia
* Chylothorax
* Diaphragmatic eventration
* Heart block
* Intracranial hemorrhage
* Laryngeal cleft
* Laryngeal nerve injury
* Mutation of *ABCA3* gene (for surfactant phospholipid transport)
* Neuromuscular disorders
* Phrenic nerve injury
* Pulmonary hemorrhage
* Pulmonary hypoplasia
* Pulmonary lymphangiectasia
* Spinal injury
* Surfactant-related protein B deficiency
* Tachycardia syndromes
* Tracheoesophageal fistula
* Transplacental medications
* Vascular catheter accident
* Other causes of airway obstruction
* Other congenital heart diseases
* Other inborn errors of metabolism
* Other neuromuscular diseases

A careful history and examination in patients with recurrent pneumonia are both very helpful to further narrow the differential diagnosis. However, additional testing is often needed to confirm most of these diagnoses and is generally outside the scope of a primary care provider.

## **Differential Diagnoses**

* Acidosis, Metabolic
* Acute Anemia
* Acute Hypoglycemia
* Afebrile Pneumonia Syndrome
* Agammaglobulinemia
* Alveolar Proteinosis
* Aortic Stenosis
* Aortic Stenosis, Subaortic
* Aortic Stenosis, Valvar
* Aseptic Meningitis
* Asphyxiating Thoracic Dystrophy (Jeune Syndrome)
* Aspiration Syndromes
* Asthma
* Atelectasis, Pulmonary
* Atrioventricular Septal Defect, Complete
* Atrioventricular Septal Defect, Unbalanced
* Bacteremia
* Birth Trauma
* Bowel Obstruction in the Newborn
* Bronchiectasis
* Bronchiolitis
* Bronchitis
* Bronchitis, Acute and Chronic
* Bronchogenic Cyst
* Cardiomyopathy, Hypertrophic
* Chronic Anemia
* Chronic Granulomatous Disease
* Coarctation of the Aorta
* Coccidioidomycosis and Valley Fever
* Severe Combined Immunodeficiency (SCID)
* Common Variable Immunodeficiency
* Complement Deficiency
* Complement Receptor Deficiency
* Congenital Diaphragmatic Hernia
* Congenital Pneumonia
* Congenital Stridor
* Cystic Adenomatoid Malformation
* Cystic Fibrosis
* Double Outlet Right Ventricle, Normally Related Great Arteries
* Double Outlet Right Ventricle, With Transposition
* Ebstein Anomaly
* Emergent Management of Atrial Flutter
* Empyema
* Esophageal Atresia With or Without Tracheoesophageal Fistula
* Foreign Body Aspiration
* Gastroesophageal Reflux
* Goodpasture Syndrome
* Head Trauma
* Hemosiderosis
* Hemothorax
* Human Immunodeficiency Virus Infection
* Hypersensitivity Pneumonitis
* Hypocalcemia
* Hypoplastic Left Heart Syndrome
* IgA and IgG Subclass Deficiencies
* Inhalation Injury
* Interrupted Aortic Arch
* Legionella Infection
* Meningitis, Bacterial
* Neural Tube Defects
* Patent Ductus Arteriosus (PDA)
* Pediatric Acute Respiratory Distress Syndrome
* Pediatric Airway Foreign Body
* Pediatric Histoplasmosis
* Pediatric Pleural Effusion
* Pediatric Pneumococcal Infections
* Pediatric Pulmonary Hypoplasia
* Pertussis
* Pneumococcal Infections
* Pneumonia, Aspiration
* Pneumonia, Bacterial
* Pneumonia, Empyema and Abscess
* Pneumonia, Immunocompromised
* Pneumonia, Mycoplasma
* Pneumothorax
* Pulmonary Atresia with Intact Ventricular Septum
* Pulmonary Atresia with Ventricular Septal Defect
* Pulmonary Hypertension, Persistent-Newborn
* Pulmonary Sequestration
* Q Fever
* Respiratory Distress Syndrome
* Respiratory Distress Syndrome
* Smoke Inhalation Injury
* Total Anomalous Pulmonary Venous Connection
* Transient Tachypnea of the Newborn
* Transposition of the Great Arteries
* Tricuspid Atresia
* Truncus Arteriosus
* Vascular Ring, Double Aortic Arch
* Vascular Ring, Right Aortic Arch

**commonly used drugs and their side effects**

## 1. Amoxicillin (First-line oral antibiotic)

* Use: Preferred for outpatient treatment of mild to moderate pneumococcal pneumonia in children under 5 years and school-aged children.
* Dosage: Typically given twice daily for 3–7 days depending on severity.
* Side Effects:
  + Gastrointestinal upset (nausea, vomiting, diarrhea)
  + Allergic reactions (rash, rarely anaphylaxis)
  + Possible alteration of gut flora leading to yeast infections.

## 2. Amoxicillin-Clavulanate (Augmentin)

* Use: Alternative when beta-lactamase producing organisms or mixed infections are suspected or in treatment failure.
* Side Effects: Similar to amoxicillin but higher risk of diarrhea and gastrointestinal discomfort.

## 3. Macrolides (Azithromycin, Clarithromycin)

* Use: Added or alternative therapy in school-aged children to cover atypical pathogens (e.g., *Mycoplasma pneumoniae*) and some pneumococcal strains.
* Side Effects:
  + Gastrointestinal upset (nausea, abdominal pain)
  + Rare QT prolongation and arrhythmias
  + Possible drug interactions.

## 4. Cephalosporins (Cefuroxime, Cefdinir, Ceftriaxone)

* Use: For hospitalized or severe cases, intravenous or oral cephalosporins are used. Ceftriaxone is common for inpatient therapy.
* Side Effects:
  + Allergic reactions (rash, anaphylaxis in penicillin-allergic patients)
  + Diarrhea, including risk of *Clostridioides difficile* infection
  + Injection site reactions (for IV forms).

## 5. Vancomycin or Clindamycin

* Use: Reserved for suspected or confirmed methicillin-resistant *S. pneumoniae* or *S. aureus* infections in severe cases.
* Side Effects:
  + Vancomycin: Nephrotoxicity, ototoxicity, “red man syndrome” (infusion reaction)
  + Clindamycin: Diarrhea, risk of *C. difficile* colitis.

## **PREDEFINED QUESTIONS AND ANSWERS**

## What is pneumococcal pneumonia?

Pneumococcal pneumonia is a lung infection caused by the bacteria *Streptococcus pneumoniae*. It is a common cause of bacterial pneumonia in children and can sometimes lead to serious illness like bloodstream infections or meningitis.

## What are the symptoms of pneumococcal pneumonia in children?

Symptoms include fever, cough, rapid or difficult breathing, chest pain, lethargy, poor appetite, and sometimes bluish discoloration of lips or nails due to low oxygen levels.

## How is pneumococcal pneumonia diagnosed?

Diagnosis is usually based on clinical evaluation and chest X-ray. Blood tests and cultures may be done in severe cases to identify the bacteria and guide treatment.

## How is pneumococcal pneumonia treated?

Treatment involves antibiotics, with amoxicillin being the first-line choice for most children. Severe cases may require hospitalization for intravenous antibiotics and supportive care like oxygen.

## Can pneumococcal pneumonia be prevented?

Yes. Vaccination is the most effective prevention. Pneumococcal conjugate vaccines (PCV15 or PCV20) are given starting at 2 months of age in a series of doses. Keeping up with flu vaccines and good hygiene also helps reduce risk.

## Are pneumococcal vaccines safe?

Yes, pneumococcal vaccines are safe and effective. Common side effects include pain, redness, or swelling at the injection site, mild fever, or irritability. Serious allergic reactions are very rare.

## Can a child get pneumococcal pneumonia more than once?

Yes. There are over 100 different types of pneumococcus bacteria, so infection with one type does not provide immunity against others.

## Who is at higher risk for pneumococcal pneumonia?

Young children under 5 years, the elderly, and those with certain medical conditions like immune deficiencies or chronic illnesses are at higher risk.

## What should I do if I suspect my child has pneumococcal pneumonia?

Seek medical care promptly if your child has difficulty breathing, persistent fever, lethargy, or bluish lips. Early diagnosis and treatment improve outcomes

**DOCTOR PATIENT CONVERSATION**

Doctor: Hello, I understand your child has been diagnosed with pneumonia. I want to explain what that means and how we will treat it.

Parent: Thank you, doctor. What exactly is pneumonia?

Doctor: Pneumonia is an infection in the lungs that causes inflammation and fluid buildup, making it harder for your child to breathe. In children, pneumonia can be caused by viruses or bacteria. One common bacterial cause is *Streptococcus pneumoniae*, which we call pneumococcal pneumonia.

Parent: How do you know it’s caused by these bacteria?

Doctor: Sometimes it’s hard to tell the exact cause right away. We look at symptoms, physical exam, and chest X-rays. If your child is very sick, we might do blood tests or cultures. Because pneumococcus is a common cause, we often start antibiotics that work well against it.

Parent: What treatment will my child need?

Doctor: For most children with pneumococcal pneumonia, we prescribe an antibiotic called amoxicillin, which your child will take by mouth. It usually helps improve symptoms within a couple of days. If your child has trouble breathing or is very sick, hospitalization and intravenous antibiotics may be needed.

Parent: Are there any side effects from the antibiotics?

Doctor: Some children may have mild stomach upset, diarrhea, or a rash. Serious allergic reactions are rare. It’s important to give the full course of antibiotics even if your child feels better sooner.

Parent: How can we prevent this from happening again?

Doctor: Vaccination is the best prevention. The pneumococcal conjugate vaccine is given starting at 2 months of age and protects against many types of pneumococcal bacteria. Also, good hand hygiene and avoiding exposure to sick people help reduce risk.

Parent: What should I watch for while my child is sick?

Doctor: Watch for worsening breathing difficulty, persistent high fever, lethargy, or if your child is not drinking fluids. If any of these happen, seek medical care promptly.

Parent: Thank you, doctor. This helps me understand the illness and how to care for my child.

Doctor: You’re welcome. Please call us if you have any questions or concerns during your child’s recovery.

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### **Pulmonary contusion**

**DEFINITION AND DESCRIPTION**

A pulmonary contusion is a bruise on your lungs from a chest injury. It causes fluid and blood to collect around your lungs. Its symptoms can sometimes be vague, like coughing or wheezing, until the injury is severe.

A pulmonary contusion is a serious condition that can cause life-threatening complications. Healthcare providers will need to monitor you in a hospital’s intensive care unit (ICU).

**Symptoms and Causes**

## Symptoms of a pulmonary contusion include:

* Trouble breathing or shortness of breath.
* Coughing.
* Wheezing.
* Fast heart rate (tachycardia).
* Coughing up blood.
* Chest pain.
* Bruises on your chest.

Symptoms can appear right after an injury or several hours to days later. You might notice slight changes, like a cough or breathlessness, before your symptoms become serious.

### **What causes a pulmonary contusion?**

Blunt force trauma (impact with something dull) or a penetrating injury (one that goes through your skin and deeper tissues) causes a pulmonary contusion. The most common cause is injury from a car accident. Other causes can be explosions and stab wounds. These injuries damage the blood vessels around your lungs, so blood and other fluids collect in your lung tissues.

Anyone who’s had an injury that impacts their chest is at risk for a pulmonary contusion. Children are at higher risk for serious complications.

### **Complications of a pulmonary contusion**

Pulmonary contusions can lead to serious complications, including:

* Pneumonia.
* Acute respiratory distress syndrome (ARDS).
* Hypoxia, a condition where not enough oxygen is getting to your tissues.
* Hypercapnia and respiratory acidosis, conditions where you have too much carbon dioxide in your blood.

**Diagnosis and Tests**

Healthcare providers diagnose a pulmonary contusion by getting images of your lungs. Imaging methods they might use include:

* Chest X-rays.
* Thoracic (chest) ultrasound.
* CT scan (computed tomography scan).

## **Management and Treatment**

There’s no specific treatment for pulmonary contusion. Healthcare providers will monitor you in the hospital and work to keep your condition stable while you heal. Some medications and procedures they might use to prevent complications and keep you comfortable include:

* Oxygen therapy.
* Pain medications.
* Pulmonary hygiene.
* Fluid drainage.
* Surgery to repair damage.
* Mechanical ventilation.

## **Outlook / Prognosis**

Pulmonary contusions take about a week to heal. Depending on the severity of your condition, you may need to stay in the hospital for longer. Some people need surgery to repair damage to their lungs or chest.

Up to 20% of people with a pulmonary contusion develop pneumonia. It’s important to practice good pulmonary hygiene, including coughing up mucous and deep breathing.

People who’ve had a pulmonary contusion sometimes have reduced lung function for years after the contusion heals. This can make it hard to breathe. You might also have long-lasting scarring (fibrosis).

It’s hard to know the exact mortality rate from a pulmonary contusion. But studies estimate it to be between 14% and 40%.

## **Prevention**

The only way to prevent a contusion is to avoid injury to your chest. Always wear your seatbelt when you’re driving or riding in a vehicle.

## **WHEN TO SEE THE DOCTOR**

## Go to an emergency room right away if you’ve had an injury to your chest. Even if you feel fine, having a healthcare provider check you out is always best. Call 911 if you’re having any of these symptoms:

* Chest pain.
* Difficulty breathing.
* Bluish skin, lips or nails.
* Dizziness.

**DIFFERENTIAL DIAGNOSIS**

* Pulmonary contusion (bruise of lung tissue after blunt chest trauma)
* Pulmonary laceration (tear in lung parenchyma, may cause pneumatoceles or air leaks)
* Pneumothorax (air in pleural space causing lung collapse)
* Hemothorax (blood in pleural space following trauma)
* Atelectasis (collapse of lung segments due to mucus plugging or compression)
* Aspiration pneumonia (infection from inhaled material, often with patchy infiltrates)
* Pulmonary edema (fluid accumulation in lungs from cardiac or non-cardiac causes)
* Infectious pneumonia (bacterial or viral lung infection causing consolidation)
* Pulmonary embolism (rare in children, but can cause respiratory distress and infarcts)
* Acute Respiratory Distress Syndrome (ARDS) (severe inflammatory lung injury that can follow trauma or infection)

**EPIDEMIOLOGY**

Epidemiology of Pediatric Pulmonary Contusions:

* Pulmonary contusions are the most common chest injury in children following blunt thoracic trauma, with an incidence ranging from 27% to over 50% of pediatric chest trauma cases in various studies.
* In a large Dutch trauma registry study, among children admitted with chest injuries, lung contusions accounted for 40.5% of injuries, making them the most prevalent thoracic injury in pediatric trauma.
* Chest trauma in children is relatively infrequent but remains a significant cause of morbidity and mortality. The overall incidence of pediatric chest injuries is about 4.9 per 100,000 person-years.
* The median age of affected children is around 10 to 11 years, with a male predominance (~60%).
* The leading causes of pulmonary contusions in children are blunt trauma mechanisms, especially motor vehicle accidents and pedestrian injuries, with pedestrian trauma accounting for approximately 26% to 72% of cases in some studies.
* Unlike adults, children can sustain pulmonary contusions without rib fractures due to the greater flexibility of their chest wall.
* Mortality rates related to pediatric chest trauma, including pulmonary contusions, range from 6% to 7%, with higher mortality associated with multiple trauma and severe injuries.
* Pulmonary contusions can progress to complications like acute respiratory distress syndrome (ARDS), which carries a mortality of approximately 10–25% in severe cases.
* Most pediatric chest trauma cases (80–90%) are due to closed (blunt) trauma, with pulmonary contusions being a common injury pattern within this group

**PREDEFINED QUESTIONS AND ANSWERS**

## What is a pulmonary contusion in children?

A pulmonary contusion is a bruise of the lung tissue caused by blunt chest trauma. It leads to bleeding and fluid leakage into the lung’s alveoli, impairing oxygen exchange and causing respiratory difficulties.

## How common are pulmonary contusions in pediatric trauma?

Pulmonary contusions are the most common intrathoracic injury in children after blunt chest trauma. They occur in about 27–50% of pediatric chest trauma cases and are a significant cause of morbidity.

## Why are children more prone to pulmonary contusions without rib fractures?

Children’s chest walls are more flexible and compliant, so their ribs often do not fracture during trauma. However, this flexibility allows more force to be transmitted to the lung tissue, increasing the risk of contusions.

## What symptoms do children with pulmonary contusions show?

Symptoms can include chest pain, rapid or difficult breathing, coughing, low oxygen levels (hypoxia), and respiratory distress. Symptoms may worsen over 24–48 hours after injury.

## How is pulmonary contusion diagnosed?

Initial diagnosis is usually by chest X-ray, but contusions may not appear immediately. CT scans detect contusions earlier and more precisely but are not always necessary. Clinical assessment and oxygen levels guide management.

## How is pulmonary contusion treated in children?

There is no specific cure; treatment is supportive:

* Oxygen therapy as needed
* Pain control to allow effective breathing and coughing
* Pulmonary hygiene (incentive spirometry, physiotherapy)
* Non-invasive ventilation (e.g., BiPAP) may help reduce work of breathing
* Mechanical ventilation if respiratory failure occurs
* Fluid management to avoid overload
* Antibiotics are not routinely given unless infection develops.

## What complications can occur?

Complications include pneumonia, acute respiratory distress syndrome (ARDS), and respiratory failure. Close monitoring is essential to detect and treat these promptly.

## How long does it take for a pulmonary contusion to heal?

Pulmonary contusions generally improve over about one week, but recovery depends on severity and complications.

## When should a child with chest trauma be evaluated for pulmonary contusion?

Any child with blunt chest trauma and respiratory symptoms should be evaluated promptly, especially if breathing is difficult or oxygen levels are low.

**DOCTOR PATIENT CONVERSATION**

Doctor: Hello, I want to talk with you about your child’s lung injury called a pulmonary contusion. Do you have any questions before we start?

Parent: Yes, doctor. What exactly is a pulmonary contusion?

Doctor: A pulmonary contusion is essentially a bruise of the lung tissue caused by a blunt injury to the chest, like from a fall or car accident. This injury causes bleeding and swelling inside the lungs, which can make it harder for your child to breathe properly.

Parent: How serious is this? Will my child have trouble breathing?

Doctor: The severity depends on how much of the lung is affected. Some children have mild symptoms and just need oxygen and monitoring, while others may have more difficulty breathing and need extra support. Because the lung tissue becomes stiff and filled with fluid, it can reduce oxygen exchange.

Parent: How do you know how bad it is?

Doctor: We use chest X-rays to look for signs of contusion, but sometimes the injury doesn’t show up right away. A CT scan can detect it earlier and more precisely, but we usually base treatment on how your child is doing clinically—how well they are breathing and their oxygen levels.

Parent: What treatment will my child need?

Doctor: Treatment is mostly supportive. We give oxygen if needed, manage pain so your child can breathe deeply and cough, and encourage pulmonary hygiene like breathing exercises. In some cases, non-invasive ventilation like BiPAP can help reduce the work of breathing. We try to avoid mechanical ventilation if possible, but if breathing worsens, it might be necessary.

Parent: Will my child need antibiotics?

Doctor: Not routinely. Antibiotics are only used if there is a confirmed infection like pneumonia. Pulmonary contusions themselves are not infections.

Parent: How long will it take for my child to get better?

Doctor: Most pulmonary contusions improve over about a week, but it depends on the injury’s extent and whether complications develop. We will monitor closely for any signs of worsening.

Parent: Is there anything we should watch for at home?

Doctor: Yes. If your child develops increased difficulty breathing, persistent low oxygen levels, fever, or becomes unusually sleepy or irritable, please seek medical attention promptly.

Parent: Thank you for explaining everything clearly.

Doctor: You’re welcome. We’ll keep monitoring your child carefully and adjust treatment as needed. Please don’t hesitate to ask if you have any concerns.

REFERENCES

[Pulmonary Contusion: Causes, Symptoms & Treatment](https://my.clevelandclinic.org/health/diseases/pulmonary-contusion#overview)

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**PULMONARY VALVE STENOSIS**

**DEFINITION AND DESCRIPTION**

Pulmonary valve stenosis is a narrowing of the valve between the lower right heart chamber and the lung arteries. In a narrowed heart valve, the valve flaps may become thick or stiff. This reduces blood flow through the valve.

Usually, pulmonary valve disease is caused by a heart problem that develops before birth. A heart problem present at birth is called a congenital heart defect. In adults, pulmonary valve stenosis may be a complication of another illness.

Pulmonary valve stenosis ranges from mild to severe. Some people with mild pulmonary valve stenosis don't have symptoms. They may need only occasional health checkups. Moderate and severe pulmonary valve stenosis may need a procedure to repair or replace the valve

## **Causes**

Pulmonary valve stenosis usually results from a heart problem present at birth. The exact cause is unclear. The pulmonary valve doesn't develop properly as the baby is growing in the womb.

The pulmonary valve is made of three thin pieces of tissue called flaps, also called cusps. The cusps open and close with each heartbeat. They make sure blood moves in the right direction.

In pulmonary valve stenosis, one or more of the cusps may be stiff or thick. Sometimes the cusps may be joined together. That means they are fused. So the valve doesn't open fully. The smaller opening makes it harder for blood to leave the lower right heart chamber. Pressure increases inside the chamber. The increased pressure strains the heart. Eventually the lower right heart chamber wall gets thicker.

## **Risk factors**

Things that may increase the risk of pulmonary valve stenosis include:

* **German measles, also called rubella.** Having German measles during pregnancy increases the risk of pulmonary valve stenosis in the baby.
* **Noonan syndrome.** This condition is caused by altered deoxyribonucleic acid (DNA). It can lead to many problems with the heart's structure and function.
* **Rheumatic fever.** This complication of strep throat can cause permanent damage to the heart and heart valves. It increases the risk of developing pulmonary valve stenosis later in life.
* **Carcinoid syndrome.** This condition occurs when a rare cancerous tumor releases certain chemicals into the bloodstream. It causes shortness of breath, flushing and other symptoms. Some people with this syndrome develop carcinoid heart disease, which damages heart valves.

## **Symptoms**

Pulmonary valve stenosis symptoms depend on how much blood flow is blocked. Some people with mild pulmonary stenosis do not have symptoms. Those with more-severe pulmonary stenosis may first notice symptoms while exercising.

Pulmonary valve stenosis symptoms may include:

* A whooshing sound called a heart murmur that can be heard with a stethoscope.
* Fatigue.
* Shortness of breath, especially during activity.
* Chest pain.
* Fainting.

Babies with pulmonary valve stenosis may have blue or gray skin due to low oxygen levels.

## **When to see a doctor**

Talk to your health care provider if you or your child has:

* Shortness of breath.
* Chest pain.
* Fainting.

Prompt diagnosis and treatment of pulmonary valve stenosis can help reduce the risk of complications.

## **Diagnosis**

Pulmonary valve stenosis is often diagnosed in childhood. But it may not be detected until later in life.

A health care provider uses a stethoscope to listen to the heart. A whooshing sound, called a heart murmur, may be heard. The sound is caused by choppy blood flow across the narrowed valve.

Tests to diagnose pulmonary valve stenosis include:

* **Electrocardiogram (ECG or EKG).** This quick and painless test records the electrical signals in the heart. Sticky patches, called electrodes, are placed on the chest and sometimes the arms and legs. Wires connect the electrodes to a computer, which displays the test results. An ECG can show how the heart is beating and may reveal signs of heart muscle thickening.
* **Echocardiogram.** An echocardiogram uses sound waves to produce images of the heart. This common test shows how the heart beats and pumps blood. An echocardiogram can show the shape of the pulmonary valve. The test can show how much of the valve is narrowed.
* **Cardiac catheterization.** A thin tube called a catheter is inserted into the groin and threaded through the blood vessels to the heart. Dye flows through the catheter into the blood vessels to make them show up more clearly on X-rays. This part of the test is called a coronary angiogram.

During the test, pressures within the heart can be measured to see how forcefully blood pumps through the heart. A provider can determine the severity of pulmonary stenosis by checking the difference in pressure between the right lower heart chamber and the lung artery.

* **Other imaging tests.** Magnetic resonance imaging (MRI) and computed tomography (CT) scans are sometimes used to confirm the diagnosis of pulmonary valve stenosis.

## **Treatment**

If you have mild pulmonary valve stenosis without symptoms, you may only need occasional health checkups.

If you have moderate or severe pulmonary valve stenosis, you may need a heart procedure or heart surgery. The type of procedure or surgery done depends on your overall health and the appearance of your pulmonary valve.

### **Surgeries or other procedures**

Pulmonary valve stenosis treatment may include:

* **Balloon valvuloplasty.** The provider inserts a flexible tube with a balloon on the tip into an artery, usually in the groin. X-rays help guide the tube, called a catheter, to the narrowed valve in the heart. The balloon inflates, making the valve opening larger. The balloon is deflated. The catheter and balloon are removed.

Valvuloplasty may improve blood flow through the heart and reduce pulmonary valve stenosis symptoms. But the valve may narrow again. Some people need valve repair or replacement in the future.

* **Pulmonary valve replacement.** If balloon valvuloplasty isn't an option, open-heart surgery or a catheter procedure may be done to replace the pulmonary valve. If there are other heart problems, the surgeon may repair those during the same surgery.

People who have had pulmonary valve replacement need to take antibiotics before certain dental procedures or surgeries to prevent endocarditis.

## **Self-care**

If you have valve disease, it's important to take steps to keep your heart healthy. Certain lifestyle changes can decrease your risk of developing other types of heart disease or having a heart attack.

Lifestyle changes to talk about with your health care provider include:

* Quitting smoking.
* Eating a heart-healthy diet that includes fruits and vegetables, low-fat dairy products, whole grains, and lean meat.
* Maintaining a healthy weight.
* Getting regular exercise.

## **Complications**

Possible complications of pulmonary stenosis include:

* **Infection of the lining of the heart, called infective endocarditis.** People with heart valve problems, such as pulmonary stenosis, have an increased risk of developing bacterial infections that affect the inner lining of the heart.
* **Irregular heartbeats, called arrhythmias.** People with pulmonary stenosis are more likely to have irregular heartbeats. Unless the stenosis is severe, irregular heartbeats due to pulmonary stenosis usually aren't life-threatening.
* **Thickening of the heart muscle.** In severe pulmonary stenosis, the lower right heart chamber must pump harder to force blood into the pulmonary artery. The strain on the heart causes the muscular wall of the ventricle to thicken. The condition is called right ventricular hypertrophy.
* **Heart failure.** If the right ventricle can't pump properly, heart failure eventually develops. Symptoms of heart failure include fatigue, shortness of breath, and swelling of the legs and belly area.
* **Pregnancy complications.** The risks of complications during labor and delivery are higher for those with severe pulmonary valve stenosis than for those without it

## **Diagnostic Considerations**

Other conditions to be considered in patients with suspected pulmonic stenosis (pulmonary stenosis) (PS) include the following:

* Associated or different congenital heart abnormalities
* Rheumatic valvular heart disease
* Carcinoid heart disease

## Pulmonic stenosis in pregnancy

Valvular heart disease, including PS, warrants follow-up care by a high-risk obstetrics team. The hemodynamic changes in pregnancy are significant and include relative anemia due to increased plasma volume proportionally greater than red blood cell volume, increase in cardiac stroke volume, decrease in systemic vascular resistance, decrease in pulmonary vascular resistance with a drop in pulmonary pressures and, finally, decrease in venous return that is more marked in the third trimester. Pregnancy, in the already symptomatic woman with severe PS, can worsen symptoms. However, pregnancy is generally well tolerated in asymptomatic women with PS, even if the degree of stenosis is severe.

When symptoms are referable to PS, they are similar to those of individuals who are not pregnant and symptomatic. The symptoms of healthy pregnancy can resemble those of PS, including exertional fatigue, dyspnea, orthopnea, presyncope, and, rarely, frank syncope. Palpitations due to arrhythmias have been noted to be more common in those with PS.

Mild PS produces a murmur similar to that of the benign flow murmur of pregnancy, which typically increases in intensity as the stroke volume is augmented. During the physical examination, this murmur can be distinguished from the flow murmur of pregnancy by noting a prominent jugular venous *a* wave, an RV lift, a systolic thrill over the pulmonic area, a pulmonic ejection sound (in doming PS), and a diminished or absent P2. Electrocardiographic (ECG) and echocardiographic evaluation are essential in confirming clinical suspicion. Fetal echocardiography is indicated in patients with PS or tetralogy of Fallot.

## **Treatment of PS during pregnancy**

Balloon valvuloplasty is recommended in nonpregnant, asymptomatic patients with a peak instantaneous gradient >60 mmHg or mean Doppler gradient >40 mmHg (provided there is less than moderate pulmonary regurgitation). In the presence of symptoms, balloon valvuloplasty is indicated for a peak instantaneous gradinet of >50 mmHg or mean gradient >30 mmHg (with the same caveat of less than moderate pulmonary regurgitation).

If severe symptomatic PS is detected during pregnancy, percutaneous balloon valvuloplasty to relieve the obstruction usually can be accomplished safely, obviating the need to terminate the pregnancy.

Arrhythmias are treated according to the severity and etiology.

## **Considerations for labor and delivery**

Patients who are asymptomatic during pregnancy generally tolerate labor and delivery well.

For more severe valvular disease, a high-risk obstetrics team along with a cardiology consultation may be required to manage deliveries.

## **Differential Diagnoses**

* Benign Cardiac Tumors
* Cardiac Sarcoma
* Primary Cardiac Neoplasms
* Sinus of Valsalva Aneurysm

## **Epidemiology**

### United States data

Pulmonic stenosis (pulmonary stenosis) (PS) is a common form of congenital heart disease that occasionally is diagnosed for the first time in adulthood. Isolated valvular PS comprises approximately 10% of all congenital heart disease in the United States. About 2% of familial occurrences are without a genetic cause.

### Sex-related demographics

A slight female predominance exists

**GUIDELINES**

Select class I and III recommendations are outlined.

### RV Outflow Tract Obstruction (OTO)

In valvular pulmonary stenosis (PS), balloon valvuloplasty is the intervention of choice, if anatomically suitable.

As long as no valve replacement is required, RVOTO intervention at any level is recommended regardless of symptoms when the stenosis is severe (Doppler peak gradient >64 mmHg).

If surgical valve replacement is the only option, it is indicated in (1) symptomatic patients with severe stenosis; or (2) asymptomatic patients with severe stenosis in the presence of ≥1 of the following:

* Objective decrease in exercise capacity
* Falling RV function and/or progression of tricuspid regurgitation (TR) to at least moderate
* RV systolic pressure (SP) >80 mmHg
* Right-to-left (RL) shunting via an ASD or VSD

### CHD-Associated Pulmonary Arterial Hypertension (PAH)

Counsel patients with congenital heart disease (CHD) and confirmed precapillary pulmonary hypertension (PH) against pregnancy.

All patients with PAH-CHD should undergo risk assessment.

In low- and intermediate-risk patients with repaired simple lesions and precapillary PH, initial oral combination therapy or sequential combination therapy is recommended; treat high-risk patients with initial combination therapy including parenteral proteinoids.

### After Repair of Tetralogy of Fallot

Pulmonary valve replacement (PVRep) is recommended in symptomatic patients with severe pulmonary regurgitation (PR) and/or at least moderate RVOTO.

In those without a native outflow tract, catheter intervention (transcatheter pulmonary valve implantation [TPVI]) is preferred if anatomically feasible.

**PREDEFINED QUESTIONS AND ANSWERS**

## What is pulmonary stenosis in children?

Pulmonary stenosis is a congenital heart defect where the pulmonary valve or the area around it is narrowed, making it harder for blood to flow from the right ventricle to the lungs. This causes the heart to work harder to pump blood.

## What causes pulmonary stenosis?

Most cases are present at birth due to abnormal development of the pulmonary valve or surrounding structures during fetal growth. The exact cause is usually unknown, and it is not related to anything the mother did during pregnancy.

## What are the symptoms of pulmonary stenosis in children?

Many children with mild pulmonary stenosis have no symptoms. When symptoms occur, they may include:

* A heart murmur heard by a doctor
* Fatigue or tiredness, especially during activity
* Shortness of breath
* Bluish tint to lips or skin (cyanosis) in severe cases
* Chest pain
* Fainting
* Swelling in legs, abdomen, or face in severe cases
* Poor weight gain or feeding difficulties in infants

## How is pulmonary stenosis diagnosed?

Diagnosis is often made by hearing a heart murmur during a physical exam. Confirmatory tests include:

* Echocardiogram (ultrasound of the heart) to visualize the valve and measure the narrowing
* Electrocardiogram (ECG) to assess heart muscle strain
* Chest X-ray to evaluate heart size and lung blood flow
* Sometimes fetal echocardiogram before birth
* Cardiac catheterization in rare cases for detailed assessment or treatment

## How is pulmonary stenosis treated?

* Mild cases may only require regular monitoring without treatment.
* Moderate to severe cases often need intervention, such as balloon valvuloplasty, a catheter-based procedure to widen the valve.
* Surgery is less common but may be needed if catheter treatment is not successful.
* Treatment improves symptoms and prevents complications like heart failure.

## What is the outlook for children with pulmonary stenosis?

With appropriate treatment, most children lead normal, active lives. Regular follow-up with a cardiologist is important to monitor heart function and valve status.

## Can pulmonary stenosis be prevented?

Since it is usually a congenital condition with unknown cause, it cannot be prevented. Early diagnosis and treatment improve outcomes.

**DOCTOR PATIENT CONVERSATION**

Doctor: Hello, I’d like to talk with you about your child’s heart condition called pulmonary stenosis. Do you have any questions before we begin?

Parent: Yes, doctor. What exactly is pulmonary stenosis?

Doctor: Pulmonary stenosis is a congenital heart defect where the valve that controls blood flow from the right side of the heart to the lungs is narrower than normal. This narrowing makes it harder for blood to flow through, so the heart has to work harder to pump blood to the lungs.

Parent: How do you diagnose this condition?

Doctor: We start with a physical exam and listen for a heart murmur, which is often the first clue. Then, we perform an echocardiogram, which is an ultrasound of the heart that lets us see the valve and measure how narrow it is. Sometimes, a chest X-ray or other imaging tests may be done to assess the heart and lungs.

Parent: What symptoms should we expect?

Doctor: Many children with mild pulmonary stenosis have no symptoms. In more severe cases, children might get tired easily, have shortness of breath, or even a bluish tint to their lips or skin. Infants might have feeding difficulties or poor growth.

Parent: How is pulmonary stenosis treated?

Doctor: Mild cases usually just need regular monitoring. For moderate to severe narrowing, we often perform a procedure called balloon valvuloplasty. This is done in the catheterization lab, where a small balloon is inserted and inflated to widen the valve. Surgery is less common but may be necessary if the catheter procedure isn’t successful.

Parent: Is the balloon procedure safe?

Doctor: Yes, it is generally safe and effective. It usually helps improve blood flow and symptoms with a relatively quick recovery time.

Parent: What is the outlook for my child?

Doctor: With appropriate treatment, most children do very well and can lead normal, active lives. We will continue to follow your child regularly to monitor heart function and valve status.

Parent: Thank you, doctor. This helps me understand the condition and what to expect.

Doctor: You’re welcome. Please feel free to ask any questions anytime, and we’ll support you and your child throughout the treatment.

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[Pulmonary valve stenosis - Diagnosis & treatment - Mayo Clinic](https://www.mayoclinic.org/diseases-conditions/pulmonary-valve-stenosis/diagnosis-treatment/drc-20377039)

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**PULMONARY ATRRESIA**

**DEFINITION AND DESCRIPTION**

Pulmonary atresia (uh-TREE-zhuh) is a heart problem present at birth. That means it is a congenital heart defect. In this condition, the valve that helps move blood from the heart to the lungs doesn't form correctly. The valve is called the pulmonary valve.

Instead of a valve that opens and closes, a solid sheet of tissue forms. So, blood can't travel its usual path to get oxygen from the lungs. Instead, some blood travels to the lungs through other natural passages within the heart and its arteries.

A baby in the womb needs these other passages. But they usually close soon after birth.

Pulmonary atresia is a life-threatening condition that needs emergency treatment. Treatment includes surgery to repair the heart and medicines to help the heart work better.

In addition to not having a normal pulmonary valve, a baby with pulmonary atresia may have:

* Lips, fingers, and toes that look blue because of a lack of oxygen in their blood.
* An underdeveloped right ventricle and tricuspid valve (the valve connecting the right atrium and right ventricle).
* An opening (the foramen ovale) between their heart’s right atrium and left atrium that may stay open instead of closing like it should after birth. This allows oxygen-poor blood to pass from the right atrium to the left atrium, where it flows through the left ventricle to the aorta.

In babies with pulmonary valve atresia, there also may be a problem with a dividing wall in their heart. This wall normally helps blood go where it’s supposed to go.

Your heart has four chambers, with a solid wall called the septum between your heart’s two sides. The right side sends blood to your lungs to get oxygen. The left side of your heart moves oxygen-rich blood to the rest of your body through your aorta (the main artery in your heart).

#### **Pulmonary atresia types**

* **Pulmonary atresia with a ventricular septal defect**. This opening in the wall (septum) between the right and left ventricles allows oxygen-rich blood to mix with oxygen-poor blood.
* **Pulmonary atresia with an intact ventricular septum**. The wall between the left and right sides of your heart is whole (intact).

### **Who does pulmonary atresia affect?**

Genetic factors, such as an abnormal gene or chromosomal defect, may increase the chances of heart defects in certain families. Some children with genetic disorders like DiGeorge’s syndrome or velocardiofacial syndrome may be at greater risk for pulmonary atresia.

Yes. Pulmonary atresia occurs in about one out of 10,000 live births

**CAUSES**

The cause of pulmonary atresia is not clear. During the first six weeks of pregnancy, the baby's heart begins to form and starts beating. The major blood vessels that run to and from the heart also begin to develop during this critical time. It's at this point in a baby's development that a congenital heart defect such as pulmonary atresia may begin to develop.

To understand how pulmonary atresia occurs, it may be helpful to know how the heart works.

### **How the heart works**

The typical heart is made of four chambers. There are two upper chambers, called atria, and two lower chambers, called ventricles.

The right side of the heart moves blood to the lungs. In the lungs, blood picks up oxygen and then returns it to the heart's left side. The left side of the heart then pumps the blood through the body's main artery, called the aorta. The blood goes to the rest of the body.

In pulmonary atresia, the pulmonary valve doesn't form as usual so it can't open. Blood can't flow from the right ventricle to the lungs.

Before birth, not having a pulmonary valve opening doesn't affect the baby's oxygen. That's because the baby gets oxygen from the tissue that connects the baby to the womb, called the placenta. The oxygen-rich blood from the placenta goes to the baby's right upper heart chamber.

The blood going into the right side of the baby's heart then passes through a hole between the top chambers of the baby's heart. The hole is called the foramen ovale. It lets oxygen-rich blood move to the rest of the baby's body through the aorta.

After birth, the lungs are needed for oxygen. In pulmonary atresia, without a working pulmonary valve, blood must find another way to reach the baby's lungs.

Blood from the right side of the heart can cross over the foramen ovale to the left heart. From there it can be pumped to the aorta. Newborn babies have a temporary opening called the ductus arteriosus between the aorta and the pulmonary artery. This opening lets some blood travel to the lungs. There the blood picks up oxygen to send to the rest of the baby's body.

The ductus arteriosus most often closes soon after birth. But medicines can keep it open.

Sometimes there's a second hole in the tissue between the main pumping chambers of the baby's heart. This hole is a ventricular septal defect (VSD).

The VSD lets blood flow from the right lower heart chamber to the left lower heart chamber. People with pulmonary atresia and a VSD often have other changes with the lungs and the arteries that bring blood to the lungs.

If there's no VSD, the right lower heart chamber gets little blood flow before birth. The chamber often doesn't form fully. This is a condition called pulmonary atresia with intact ventricular septum (PA/IVS).

**Risk factors**

Pulmonary atresia happens as the baby's heart is forming during pregnancy. Certain health conditions or illicit drug use during pregnancy may increase a baby's risk of pulmonary atresia or other congenital heart defects. Risk factors include:

* Obesity.
* Alcohol or tobacco use.
* Diabetes.
* Use of some types of medicines during pregnancy, such as certain acne and blood pressure medicines.

Some types of congenital heart defects occur in families. This means they are inherited. If you or someone in your family was born with a heart problem, including pulmonary atresia, ask your care team is genetic screening is right for you. Screening can help show the risk of certain congenital heart defects in future children.

**Symptoms**

Symptoms of pulmonary atresia may be seen soon after birth. They can include:

* Blue or gray skin, lips or fingernails due to low oxygen levels. Depending on the skin color, these changes may be harder or easier to see.
* Fast breathing or shortness of breath.
* Tiring easily.
* Not feeding well.

### **When to see a doctor**

Pulmonary atresia is most often found soon after birth. If your baby has symptoms of pulmonary atresia after you've left the hospital, get medical help right away.

## **Diagnosis**

Pulmonary atresia is usually diagnosed soon after birth. Tests are done to check the baby's heart health.

### **Tests**

Tests to diagnose pulmonary atresia may include:

* **Pulse oximetry.** A sensor placed on the fingertip records the amount of oxygen in the blood. Too little oxygen may be a sign of a heart or lung problem.
* **Chest X-ray.** A chest X-ray shows the size and shape of the heart and lungs.
* **Electrocardiogram (ECG or EKG).** This quick and painless test records the electrical activity of the heart. It shows how the heart is beating. Sticky patches called electrodes are put on the chest and sometimes the arms and legs. Wires connect the patches to a computer, which prints or displays the results.
* **Echocardiogram.** This test uses sound waves to create pictures of the beating heart. An echocardiogram is usually the main test to diagnose pulmonary atresia. It shows how blood moves through the heart and heart valves. If an echocardiogram is done on a baby before birth, it's called a fetal echocardiogram.
* **Cardiac catheterization.** A doctor threads a thin tube called a catheter through a blood vessel in the arm or groin to an artery in the heart. Dye is sent through the catheter. This makes the heart arteries show up more clearly on an X-ray. The test can provide detailed information on blood flow and how the heart works. Certain heart treatments can be done during cardiac catheterization.

**Treatment**

Babies need emergency medical care for pulmonary atresia symptoms. The choice of surgeries or procedures depends on how severe the condition is.

### **Medications**

Medicine may be given through an IV to keep the ductus arteriosus open. This is not a long-term treatment for pulmonary atresia. But it gives healthcare professionals more time to decide what type of surgery or procedure might be best.

### **Surgery or other procedures**

Sometimes, pulmonary atresia treatment can be done using a long, thin tube called a catheter. A doctor places the tube into a large blood vessel in a baby's groin and guides it to the heart. Catheter-based procedures for pulmonary atresia include:

* **Balloon atrial septostomy.** A balloon is used to enlarge the natural hole in the wall between the upper chambers of the heart. This hole, called the foramen ovale, most often closes soon after birth. Making the hole larger lets blood move easily from the right side of the heart to the left side.
* **Stent placement.** A doctor may put a rigid tube called a stent in the ductus arteriosus to prevent it from closing. This keeps blood flowing to the lungs.

Babies with pulmonary atresia often need many heart surgeries over time. The type of heart surgery depends on the size of the child's lower right heart chamber and pulmonary artery.

Types of surgery for pulmonary atresia include:

* **Shunting.** This involves making a new route for blood to flow, called a bypass shunt. The shunt goes from the main blood vessel leading out of the heart, called the aorta, to the pulmonary arteries. This lets enough blood flow to the lungs. But most babies outgrow this shunt within a few months.
* **Glenn procedure.** In this surgery, one of the large veins that returns blood to the heart is joined to the pulmonary artery. Another large vein keeps blood flowing to the right side of the heart. The heart then pumps it through the pulmonary valve that's been repaired. This can help the right ventricle grow.
* **Fontan procedure.** If the right lower heart chamber stays too small to do its work, surgeons may use this procedure to make a pathway. The pathway lets most, if not all, of the blood coming to the heart to flow into the pulmonary artery.
* **Heart transplant.** In some cases, the heart is too damaged to fix. Then a heart transplant may be needed.

If the baby also has a ventricular septal defect (VSD), surgery is done to patch the hole. Then the surgeon makes a connection from the right pumping chamber to the pulmonary artery. This repair may use an artificial valve.

**Lifestyle and home remedies**

Here are some tips for caring for someone with pulmonary atresia after coming home from the hospital:

* **Go to scheduled health checkups.** A person born with pulmonary atresia needs regular checkups, even as an adult. A doctor trained in congenital heart diseases, called a congenital cardiologist, often provides care. Get recommended vaccines, including yearly flu vaccines.
* **Ask about exercise and activity.** Some children with a congenital heart defect may need to limit exercise or sports activities. However, many others with a congenital heart defect can participate in such activities. Your child's care team can tell you which sports and types of exercise are safe for your child.
* **Practice good oral hygiene.** Brushing and flossing teeth and getting regular dental checkups can help prevent infection.
* **Ask about preventive antibiotics.** Sometimes, a congenital heart defect can increase the risk of infection in the lining of the heart or heart valves. This infection is called infective endocarditis. Antibiotics may be recommended before dental procedures to prevent infection, especially for people who have a mechanical heart valve.

## **Complications**

Without treatment, pulmonary atresia most often leads to death. After surgery for pulmonary atresia, babies need regular health checkups throughout their lives to watch for complications.

Complications of pulmonary atresia may include:

* Bacterial infection of the inner lining of the heart and valves, called infectious endocarditis.
* Irregular heartbeats, called arrhythmias.
* Weakening of heart function.

**Prevention**

It might not be possible to prevent pulmonary atresia. But getting good prenatal care is important. Some things you can do before or during pregnancy might help lower your baby's risk of congenital heart defects. They include:

* **Control other health conditions.** If you have diabetes, keep your blood sugar in check. For other conditions that need medicines, talk to your healthcare professional about taking these medicines while pregnant.
* **Don't smoke and don't be around others who smoke.** If you smoke, quit. Smoking during pregnancy increases the risk of a congenital heart defect in the baby.
* **Aim for a healthy weight.** Obesity increases the risk of having a baby with a congenital heart defect.
* **Get recommended vaccinations.** Having rubella, also called German measles, during pregnancy can cause problems in a baby's heart development. A blood test done before pregnancy can determine if you're immune to rubella. A vaccine is available for those who aren't immune.

## **Outlook / Prognosis**

Complications of pulmonary atresia may include:

* Delays in growth.
* Abnormal heart rhythms.
* Heart failure.
* Liver disease.

It is also very important to monitor for complications after surgical repair. Fontan circulation is not normal, so your child will need close lifelong follow up. Over time, people with Fontan circulation may develop signs of heart failure (difficulty breathing, fatigue, swelling in the belly and legs, poor exercise tolerance), liver dysfunction, and abnormal heart rhythms. Some people need a heart transplant.

#### **Is pulmonary atresia fatal?**

Without treatment, pulmonary atresia is fatal because it makes your oxygen level low. However, when your healthcare provider makes a diagnosis before or shortly after your baby’s birth, they can treat your newborn to improve their oxygen circulation. Your baby may need several surgeries at different ages to keep improving their situation.

#### **Outlook for pulmonary atresia**

Pulmonary atresia life expectancy varies depending on how severe your child’s condition is and other individual factors. Survival rates are better today than they were in previous decades.

About 60% to 85% of people who have a Fontan procedure (the last in the surgical series) are alive 20 years later. However, many have long-term complications. Some say a person who had a Fontan procedure and is now 35 years old has as many years of life left as someone who is already 72.

### **Survival rate of pulmonary atresia**

Without having surgery to fix pulmonary atresia with a ventricular septal defect, the survival rate is 50% at age 1 and 8% at 10 years of age. Most people don’t live into their 30s without surgery.

### **Can pulmonary atresia be cured?**

No. Medical procedures and surgeries can improve your child’s condition, but they aren’t cures.

**DIFFERENTIAL DIAGNOSIS**

* Pulmonary atresia with intact ventricular septum (PA-IVS)
* Pulmonary atresia with ventricular septal defect (PA-VSD)
* Tetralogy of Fallot with pulmonary atresia
* Tricuspid atresia
* Critical pulmonary stenosis (severe narrowing of pulmonary valve, may mimic atresia)
* Severe pulmonary valve dysplasia or hypoplasia
* Ebstein anomaly (malformation of tricuspid valve causing cyanosis)
* Total anomalous pulmonary venous return (TAPVR)
* Other complex cyanotic congenital heart diseases causing right ventricular outflow obstruction or cyanosis

**Epidemiology of Pediatric Pulmonary Atresia:**

* Pulmonary atresia is a rare congenital heart defect characterized by the absence or complete closure of the pulmonary valve, preventing blood flow from the right ventricle to the lungs.
* The overall birth prevalence of pulmonary atresia is approximately 1 in 6,700 to 1 in 10,000 live births in the United States and other developed countries.
* Specifically, pulmonary atresia with ventricular septal defect (PA-VSD) occurs in about 0.042 per 1,000 live births (roughly 1 in 24,000), making it an unusual but recognized cardiac malformation.
* Pulmonary atresia with intact ventricular septum (PA-IVS) is even rarer, accounting for less than 1% of all congenital heart defects, with an estimated incidence of about 1 in 15,000 live births.
* Pulmonary atresia is classified into two main forms based on the presence or absence of a ventricular septal defect, which differ in clinical presentation, associated anomalies, and epidemiology.
* It is considered a critical congenital heart defect (critical CHD) because infants typically present with cyanosis shortly after birth due to lack of pulmonary blood flow.
* Some forms of pulmonary atresia, especially PA-VSD, are associated with genetic syndromes such as 22q11 deletion syndrome (DiGeorge syndrome).
* Pulmonary atresia accounts for a significant proportion of cyanotic congenital heart diseases and often requires early diagnosis and intervention.
* The condition can sometimes be detected prenatally via fetal echocardiography during the second trimester.

**GENOMIC DATA**

## Genomic Research:

* Rare Candidate Genes: Studies have identified novel candidate genes likely involved in PA pathogenesis, including *DNAH10, DST, FAT1, HMCN1, HNRNPC, TEP1,* and *TYK2* . These genes are expressed in human pulmonary artery and embryonic heart tissues.
* Copy Number Variants (CNVs): Several rare CNVs have been implicated in PA-VSD, such as deletions at 16p11.2 (affecting *PPP4C* and *TBX6*), 5q35.3 (*FLT4*), and 5p13.1 (*RICTOR*), as well as duplications at 6p21.33 (*TNXB*), 7p15.2 (*HNRNPA2B1*), and 19p13.3 (*FGF22*) . These genes interact with known cardiac development genes and are expressed in embryonic hearts.
* Novel Rare Variants: A recent study identified 176 risk genes with rare variants in PA patients, including 18 rare variants in 11 new candidate genes potentially involved in PA pathogenesis . These findings expand the genetic landscape of PA.
* Single-Gene Mutations: Mutations in genes such as *GJA5, GDF1,* and *MTHFR* have been reported in PA patients, suggesting a cumulative effect of multiple genetic factors .
* Family Studies: Whole exome sequencing in familial cases has revealed mutations in the *BMPR2* gene, which is involved in vascular development and signaling pathways related to heart formation. A novel heterozygous mutation c.2804C>T (p.A935V) in *BMPR2* was identified in affected family members, linking it to PA.
* Genetic Syndromes: PA can be associated with genetic syndromes such as 22q11 deletion syndrome (DiGeorge syndrome), which involves multiple congenital anomalies including cardiac defects.

## **PREDEFINED QUESTION AND ANSWERS**

## What is pulmonary atresia in children?

Pulmonary atresia is a rare congenital heart defect where the pulmonary valve, which controls blood flow from the right ventricle to the lungs, is completely closed or absent. This prevents blood from reaching the lungs to get oxygen.

## What causes pulmonary atresia?

Pulmonary atresia occurs because the fetal heart does not develop normally during the first 8 weeks of pregnancy. Most cases have no known cause, though some congenital heart defects can run in families. Environmental and genetic factors may play a role.

## What are the symptoms of pulmonary atresia?

Symptoms usually appear shortly after birth or when the ductus arteriosus (a fetal blood vessel) closes. Common symptoms include:

* Bluish skin or lips (cyanosis) due to low oxygen
* Rapid or troubled breathing
* Trouble feeding or poor weight gain
* Fatigue or tiredness
* Heart murmur detected by a doctor

## How is pulmonary atresia diagnosed?

Diagnosis is made using:

* Pulse oximetry to measure oxygen levels
* Chest X-ray to assess heart and lung size
* Electrocardiogram (ECG) to check heart rhythm
* Echocardiogram (ultrasound of the heart) to visualize the valve and blood flow
* Cardiac catheterization for detailed imaging and sometimes treatment
* Fetal echocardiogram can diagnose the condition before birth

## How is pulmonary atresia treated?

* Babies need emergency care after birth.
* Medications like prostaglandins keep the ductus arteriosus open to allow blood flow to the lungs temporarily.
* Procedures such as balloon atrial septostomy or stent placement in the ductus arteriosus may be done via catheter.
* Surgery is usually required, which may include shunts to direct blood flow to the lungs or more complex repairs depending on heart anatomy.
* Some children require multiple surgeries over time.
* In severe cases, heart transplantation may be considered.

## What is the outlook for children with pulmonary atresia?

With timely treatment and surgery, many children survive and can lead active lives, though ongoing cardiac care is essential. The prognosis depends on the severity of the defect and associated heart abnormalities.

## Can pulmonary atresia be prevented?

Since the exact cause is unknown and it is a congenital defect, it cannot be reliably prevented. However, good prenatal care, avoiding harmful substances during pregnancy, and managing maternal health conditions may reduce risk.

**DOCTOR PATIENT CONVERSATION**

Doctor: Hello, I want to talk with you about your child’s diagnosis of pulmonary atresia. Do you have any questions before we start?

Parent: Yes, doctor. What exactly is pulmonary atresia?

Doctor: Pulmonary atresia is a serious congenital heart defect where the valve that allows blood to flow from the right side of the heart to the lungs didn’t form properly. The valve is completely closed, so blood can’t get to the lungs to pick up oxygen.

Parent: That sounds very serious. How does this affect my child?

Doctor: Because blood can’t flow normally to the lungs, your child’s body isn’t getting enough oxygen, which causes the bluish skin color we call cyanosis. Babies with this condition often have trouble breathing and feeding, and they can get very tired quickly.

Parent: How is this treated?

Doctor: Right after birth, we give medication called prostaglandin to keep a blood vessel called the ductus arteriosus open. This vessel helps blood reach the lungs temporarily. Then, your child will need surgery or catheter-based procedures to create or improve blood flow to the lungs. The exact treatment depends on how well the right ventricle and pulmonary arteries have developed.

Parent: What kind of surgeries will my child need?

Doctor: There are a few surgical options. Sometimes, a shunt is placed to connect the aorta to the pulmonary arteries to increase blood flow to the lungs. In other cases, more complex repairs or staged surgeries like the Fontan procedure may be needed if the right ventricle is too small to pump effectively. Your child may need multiple surgeries as they grow.

Parent: Will my child need lifelong care?

Doctor: Yes, pulmonary atresia requires lifelong follow-up with a pediatric cardiologist and later an adult congenital heart disease specialist. Your child will need monitoring, possible additional interventions, and support for a healthy life.

Parent: What can we expect in terms of recovery and quality of life?

Doctor: With timely treatment, many children do well and can lead active lives. Recovery from surgery takes time, and your child will need special care after procedures, but advances in treatment have greatly improved outcomes.

Parent: Is there anything I should watch for at home?

Doctor: Yes. Watch for worsening breathing difficulty, poor feeding, increased fatigue, or changes in skin color. If you notice any of these, seek medical attention promptly.

Parent: Thank you for explaining everything. It helps to understand what’s ahead.

Doctor: You’re welcome. We’ll support you and your child every step of the way. Please don’t hesitate to ask any questions as we go forward.

REFERENCES

[Pulmonary Atresia: Symptoms, Causes and Treatment](https://my.clevelandclinic.org/health/diseases/14779-pulmonary-atresia#outlook-prognosis)

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### 

### **Pneumomediastinum**

**DEFINITION AND DESCRIPTION**

Pneumomediastinum (pronounced “noo-mow-mee-dee-A-stuh-num”) is a condition where you have air in the space in the middle of your chest between your lungs (mediastinum) and around your heart. It’s usually caused by an injury, illness or surgery. While the condition itself is usually harmless, underlying causes can be serious.

Pneumomediastinum is also called mediastinal emphysema.

#### **Types of pneumomediastinum**

Pneumomediastinum can either be spontaneous or secondary. Spontaneous pneumomediastinum (SPM) isn’t caused by injury or illness, or the cause is unknown. Secondary pneumomediastinum develops because of an injury or illness.

Pneumomediastinum is uncommon. Experts estimate that it affects 1 in 25,000 people between the ages of 5 and 34 (the group most affected by SPM).

Spontaneous pneumomediastinum isn’t an emergency. But if you have air in your mediastinum due to an injury or illness, you should be treated right away. Go to the nearest emergency room or seek medical care immediately if you have chest pain and shortness of breath. These could be signs of life-threatening illnesses or conditions.

#### **Pneumomediastinum and pneumothorax**

Pneumothorax is a collapsed lung. It happens when there’s air between your lung and chest wall. The air pushes on your lung until it collapses under the pressure. Pneumomediastinum is air in the space between your lungs, in the center of your chest. This area is called your mediastinum.

## **Symptoms and Causes**

Symptoms of pneumomediastinum include:

* Severe pain in the middle of your chest or breastbone. It may radiate to your arms or neck. The pain may be worse with breathing or swallowing.
* Shortness of breath.
* Air pockets under your skin on your face, neck or chest (subcutaneous emphysema).
* Neck or face swelling.
* High-pitched voice or other voice distortion.

Some people don’t have any symptoms.

### **What causes pneumomediastinum?**

Pneumomediastinum is usually caused by air moving into your mediastinum from an injury to an internal organ or structure. This includes your airways (trachea or bronchi), air sacs in your lungs (alveoli), tube from your throat to your stomach (esophagus) or intestines.

Anything that increases pressure in your chest can cause spontaneous pneumomediastinum, though this is rare. Sometimes, your provider can’t find the cause.

#### **Examples of pneumomediastinum causes**

Examples of specific causes of pneumomediastinum include:

* Severe injury (trauma) to your chest.
* Surgery.
* Rupture of the small sacs of your lungs (alveoli).
* Tear in your airways or gastrointestinal (GI) tract.
* Bacterial, fungal or viral infections.
* Excessive coughing, sneezing, vomiting and other body functions that increase pressure in your chest suddenly.
* Bearing down while going to the bathroom.
* Pushing during childbirth.
* Inhaling cocaine or marijuana.
* Scuba diving.
* Mechanical ventilation.

#### **Risk factors for pneumomediastinum**

You might be at higher risk for pneumomediastinum if you:

* Smoke.
* Have asthma, COPD or other lung conditions.
* Use inhaled recreational drugs.
* Recently had surgery on your abdomen, neck or chest.
* Recently gave birth.

### **Complications of pneumomediastinum**

Some rare complications of pneumomediastinum are life-threatening and need to be treated right away. They include:

* Buildup of air might push on the area around your lungs, causing one to collapse (pneumothorax).
* Air may move from the mediastinum to the area between your heart and the sac around it (pneumopericardium).
* If too much air builds up, it can put pressure on structures in your chest, including your heart and blood vessels. They may not work properly if this happens.

## **Diagnosis and Tests**

A healthcare provider diagnoses pneumomediastinum with a chest X-ray or CT scan. These are tests that take pictures of the inside of your chest. They’ll order them after listening to your heart and lungs. Hearing a crunching sound in time with your heartbeat (Hamman’s sign) is a sign that you might have air in your mediastinum.

## **Management and Treatment**

There’s no specific treatment for pneumomediastinum. It’s usually not serious and your body will absorb the air on its own. Oxygen therapy can speed this up. If you have an underlying health condition, like an infection or injury to an internal organ, your provider will treat you for that condition.

#### **Treatments**

Your provider may admit you to the hospital for 24 hours or longer for observation or treatment. While there, they might treat you with:

* **Oxygen therapy.** Breathing in additional oxygen can help the air absorb into your body faster.
* **Medication.** Medication like pain relievers or cough suppressants can ease your symptoms until your body absorbs the extra air.
* **Needle aspiration.** If you have a lot of air that’s pressing on structures inside of your body, a provider may remove some with a needle or put a drain in. This is rare.

### **Can pneumomediastinum be fatal?**

Pneumomediastinum isn’t fatal on its own. But sometimes, serious, life-threatening health conditions can cause it. Your provider will look for causes of the air and treat them if necessary.

## **Outlook / Prognosis**

People with spontaneous pneumomediastinum (SPM) spend an average of three days in the hospital but are sometimes there for a week or more. People with underlying causes of pneumomediastinum can be hospitalized for several weeks.

Spontaneous pneumomediastinum usually goes away on its own without treatment. If it wasn’t caused by an underlying health condition, it shouldn’t come back. The health conditions that cause it can sometimes be cured.

Your outlook will depend on what’s causing the air in your mediastinum. People with spontaneous pneumomediastinum can make a full recovery and rarely have it happen again. If you have secondary pneumomediastinum, recovery will depend on the severity of your underlying condition. Some conditions that cause pneumomediastinum can be fatal.

## **Prevention**

Pneumomediastinum is rare and researchers don’t understand all the causes or risk factors. There aren’t any recommended ways to prevent it. Not smoking and not using inhaled recreational drugs might lower your risk.

**Living With**

See a healthcare provider if you’re experiencing discomfort in your chest, face swelling or unexpected changes in your voice, especially if you’ve recently had surgery or given birth.

Seek medical attention or go to the nearest emergency room immediately if you’re experiencing:

* Chest pain.
* Shortness of breath.
* Severe face or neck swelling.
* Lightheadedness.

## **Differential Diagnoses**

* Acute Respiratory Distress Syndrome (ARDS)
* Anxiety Disorders
* Aspiration Syndromes
* Blunt Abdominal Trauma
* Blunt Chest Trauma
* Bronchiolitis
* Croup
* Diabetic Ketoacidosis (DKA)
* Hypersensitivity Pneumonitis
* Mycoplasma Infections (Mycoplasma pneumoniae)
* Myocardial Infarction in Childhood
* Myocarditis
* Pediatric Asthma
* Pediatric Bronchitis
* Pediatric Infective Pericarditis
* Pediatric Metabolic Acidosis
* Pediatric Pneumonia
* Pediatric Pneumothorax
* Pediatric Viral Myocarditis
* Penetrating Abdominal Trauma
* Penetrating Chest Trauma
* Post pericardiotomy Syndrome
* Sinonasal Manifestations of Cystic Fibrosis
* Status Asthmaticus

## **Epidemiology**

### United States statistics

The epidemiology of pneumomediastinum reflects that of the associated disease states, when present.

Spontaneous pneumomediastinum is uncommon. Based on previous studies, a review by Chalumeau et al determined a prevalence of spontaneous pneumomediastinum ranging from 1 per 800 to 1 per 42,000 pediatric patients presenting to a hospital emergency department.

Stack et al reported a 0.3% incidence of pneumomediastinum in association with asthma presenting to their institution over a 10-year period.The mean age of affected patients was 11 years. No sex differences were observed in this cohort. Another study by Vianello et al that included 45 patients with severe acute asthma exacerbation found that 11.1% (5 patients) were diagnosed with pneumomediastinum.

A study reported the frequency of extra-abdominal gas in a series of patients undergoing laparoscopic esophageal surgery. Forty-seven percent of patients (N = 45) had evidence of extra-abdominal air on chest radiography. Of these, 86% had a pneumomediastinum. Pneumomediastinum persisted at least 1 postoperative day in two thirds of these cases. However, no mortality or morbidity was attributable to the presence of pneumomediastinum.

In a series of patients with sepsis-induced acute respiratory distress syndrome (ARDS), air leaks of any type, excluding pneumothorax, occurred in 3.7% of patients.Ventilator pressures and volumes delivered were not correlated with the development of air leak.

In a series of adult patients presenting with blunt chest trauma, as many as 10% had evidence of pneumomediastinum. A study by Muckart et al that included 389 patients with blunt thoracic trauma reported a minimal incidence (5.9% for pneumomediastinum) of pneumomediastinum or pneumopericardium following blunt thoracic trauma.

### International statistics

A study by Briassoulis et al from Greece, evaluating the frequency of air leaks in children receiving mechanical ventilation, reported a prevalence of 27%.However, they did not report the prevalence of specific types of air leak.

Esayag et al reported an Israeli series of 13 patients with spontaneous pneumomediastinum.This group represented 1 in 41,600 referrals to the emergency room and 1 in 15,500 hospitalizations. The median age of the patients was 19 years (range 2-72 y). Males comprised 77% of this group.

A case series from Taiwan reported by Lee et al defined an incidence of spontaneous pneumomediastinum in children of 1:8,302 patient visits to the pediatric emergency department.They observed a bimodal distribution, with cases occurring in children younger than 4 years old and in adolescents aged 15-18 years. Males outnumbered females by a ratio of 4:1.

### Sex- and age-related demographics

In a series of pneumomediastinum occurring in persons with asthma, there was a very slight male predominance in the prevalence of spontaneous pneumomediastinum. Other series confirm this excess of male cases. Damore and Dayan reported 29 cases of pneumomediastinum over a 10-year period unrelated to trauma, intubation, or surgical procedures; 69% of patients were male.Traumatic pneumomediastinum is more common in males, reflecting the male predominance among those who experience trauma and accidents.

Some have suggested that a body habitus favoring a tall thin build is an additional risk factor for the development of spontaneous pneumomediastinum. The mechanisms underlying this association are unclear.

The peak prevalence of spontaneous pneumomediastinum is seen in the second to fourth decades of life. This presumably reflects involvement in activities that increase the risk for developing pneumomediastinum, such as diving or marked physical exertion (eg, athletic activities, weight lifting). Moreover, the force of an individual's cough, vomit, and Valsalva maneuvers (all of which may lead to pneumomediastinum) attenuates with age, accounting for the decline in the prevalence of pneumomediastinum with age.

The age distribution for pneumomediastinum occurring in conjunction with specific disease processes reflects the age profile of the particular disease.

## **Procedures**

Diagnostic or therapeutic procedures are generally not necessary. Placement of a chest tube should not be attempted unless an accompanying symptomatic pneumothorax is present.

Bronchoscopy is indicated if a tracheobronchial perforation is suspected, which may occasionally be observed following blunt chest trauma. Neal et al observed that pneumomediastinum after blunt trauma in clinically stable children is rarely associated with significant underlying injury to the tracheobronchial tree.In addition to trauma indications, bronchoscopy may assist with localization and removal of foreign bodies as well as the evaluation of endobronchial lesions that may occur in association with pneumomediastinum.

Haam et al reported a series in which all patients with spontaneous pneumomediastinum undergoing contrast studies of the esophagus or esophagoscopy had normal studies.These authors suggest that these studies are indicated only if an esophageal perforation is suspected or signs of mediastinitis are present.

**PREDEFINED QUESTIONS AND ANSWERS**

## What is pneumomediastinum?

Pneumomediastinum is a condition where air is present in the mediastinum, the central space in the chest between the lungs. It can occur spontaneously or due to trauma, medical procedures, or underlying lung or esophageal problems.

## What causes pneumomediastinum?

Common causes include blunt or penetrating chest trauma, asthma exacerbations, severe coughing or vomiting, mechanical ventilation, esophageal rupture, and certain infections. Spontaneous pneumomediastinum occurs without an obvious cause, often in young adults or children after activities that increase intrathoracic pressure.

## What are the symptoms of pneumomediastinum?

Symptoms may include sudden severe chest pain beneath the breastbone that can radiate to the neck or arms, shortness of breath, difficulty swallowing, throat or jaw pain, voice changes, neck swelling, and sometimes subcutaneous emphysema (air under the skin causing swelling or crackling sensation).

## How is pneumomediastinum diagnosed?

Diagnosis is primarily made by chest X-ray or CT scan showing air in the mediastinum. Physical exam may reveal subcutaneous emphysema or a characteristic crunching sound over the chest called Hamman’s sign. Additional tests may be done to rule out esophageal perforation or other causes.

## How is pneumomediastinum treated?

Most cases resolve on their own without treatment. Supportive care includes rest, oxygen therapy to help absorb the air faster, and pain management. If pneumomediastinum is caused by trauma or esophageal rupture, more urgent interventions like surgery or chest tube placement may be necessary.

## What is the prognosis?

The outlook depends on the underlying cause. Spontaneous pneumomediastinum usually resolves completely without complications. Secondary pneumomediastinum related to trauma or esophageal injury may have more serious outcomes and require prompt treatment.

## When should I seek medical care?

Seek immediate medical attention if you experience sudden chest pain, difficulty breathing, or swelling in the neck or chest, especially after trauma or invasive procedures

**DOCTOR PATIENT CONVERSATION**

Doctor: Hello, I want to discuss your recent diagnosis of pneumomediastinum. Do you have any questions before we begin?

Patient: Yes, doctor. What exactly is pneumomediastinum?

Doctor: Pneumomediastinum means there is air trapped in the mediastinum, which is the central space in your chest between the lungs. This can happen spontaneously or due to injury or other causes.

Patient: How did this happen to me?

Doctor: In your case, it appears to be spontaneous, meaning it occurred without trauma or medical procedures. It can happen after activities that increase pressure in the chest, like severe coughing, vomiting, or asthma attacks.

Patient: What symptoms should I expect?

Doctor: Common symptoms include sudden chest pain, shortness of breath, neck pain or swelling, and sometimes a crackling sensation under the skin called subcutaneous emphysema. Some people also notice a crunching sound in sync with their heartbeat, called Hamman’s sign.

Patient: Is this dangerous? What treatment do I need?

Doctor: Most spontaneous cases are not serious and tend to resolve on their own. Treatment mainly involves rest, oxygen therapy to help absorb the air faster, and pain relief. We will monitor you closely to ensure no complications develop.

Patient: How long will it take to get better?

Doctor: Usually, symptoms improve within a few days to a week. We’ll do follow-up imaging to confirm the air has been absorbed.

Patient: When should I seek urgent care?

Doctor: If you experience worsening chest pain, difficulty breathing, swelling in your neck or face, or fever, please seek medical attention immediately, as these could indicate complications or other serious conditions.

Patient: Thank you, doctor. This helps me understand what’s going on.

Doctor: You’re welcome. Feel free to ask any questions anytime, and we’ll support you throughout your recovery.

REFERENCES

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**SUDDEN INFANT DEATH SYNDROME**

**DEFINITION AND DESCRIPTION**

Sudden infant death syndrome is the unexplained death of a baby. The baby is usually less than a year old and seems to be healthy. It often happens during sleep. Sudden infant death syndrome also is known as SIDS. It is sometimes called crib death because infants often die in their cribs.

The cause of SIDS is unknown. But it may be caused by problems in the area of an infant's brain that controls breathing and waking up from sleep.

Researchers have found some things that might put babies at higher risk. They've also found some things you can do to help protect your child from SIDS. The most important action may be to place a baby on the back to sleep.

**Causes**

Both physical and sleep factors put an infant at risk of SIDS. These factors vary from child to child.

### **Physical factors**

Physical factors associated with SIDS include:

* **Brain defects.** Some infants are born with problems that make them more likely to die of SIDS. In many of these babies, the part of the brain that controls breathing and waking up from sleep hasn't developed enough to work properly.
* **Low birth weight.** Being born early or being part of a multiple birth increases the chances that a baby's brain hasn't fully developed at birth. The baby may have less control over such automatic processes as breathing and heart rate.
* **Respiratory infection.** Many infants who died of SIDS had recently had a cold. A cold may lead to breathing problems.

### **Sleep factors**

A baby's sleeping position, items in the crib and other conditions may increase the risk of SIDS. Examples include:

* **Sleeping on the stomach or side.** Babies placed in these positions to sleep might have more trouble breathing than those placed on their backs.
* **Sleeping on a soft surface.** Lying face down on a fluffy comforter, a soft mattress or a waterbed can block an infant's airway.
* **Sharing a bed.** The risk of SIDS rises if a baby sleeps in the same bed with parents, siblings or pets. But it may help if the infant sleeps in a separate bed in the same room with parents. That seems to lower the risk of SIDS.
* **Overheating.** Being too warm while sleeping can increase a baby's risk of SIDS.

**Risk factors**

SIDS can happen to any infant. But researchers have found several factors that might raise the risk. They include:

* **Sex.** Boys are slightly more likely than girls to die of SIDS.
* **Age.** Infants are at higher risk between the second and fourth months of life.
* **Race.** For reasons that aren't well understood, SIDS occurs more often in Black, Native American and Alaska Native infants.
* **Family history.** Babies with siblings who died of SIDS are at higher risk of SIDS.
* **Secondhand smoke.** Babies who live with smokers have a higher risk of SIDS.
* **Premature birth.** Being born early and having a low birth weight increase a baby's chances of SIDS.

### **Risk factors in mothers**

During pregnancy, mothers also affect their babies' risk of SIDS, especially if they:

* Are younger than 20.
* Smoke cigarettes.
* Use drugs or alcohol.
* Do not receive good medical care while pregnant.

**Prevention**

There's no definite way to prevent SIDS. But you can help your baby sleep more safely by following these tips:

* **Back to sleep.** Place your baby to sleep in the correct position — on the back. Be sure to use the back position every time you or anyone else puts your baby to sleep for the first year of life. Don't trust that others will place your baby to sleep in the correct position: Insist on it. This won't be needed once your baby can roll over both ways without help.

Don't put your baby on the stomach or side to sleep. Advise a caregiver to only use the stomach position when the baby and caregiver are both in the same room and both are awake. Short periods of "tummy time" help a baby build muscle strength. But the baby should never be left alone during tummy time.

* **Keep the crib as bare as possible.** Use a firm, flat mattress. Make sure the mattress does not sit at an angle higher than 10 degrees. Avoid placing your baby on thick, fluffy padding, such as lambskin or a thick quilt. Don't leave pillows, fluffy toys or stuffed animals in the crib. They can cause problems with breathing if your baby's face presses against them.
* **Don't overheat your baby.** To keep your baby warm, try a sleep sack. Or dress your baby in layers instead of using blankets. Don't cover your baby's head.
* **Have your baby sleep in your room.** If possible, your baby should sleep in your room with you, but not in the same bed. Have your baby sleep alone in a crib or bassinet with a mattress designed for infant bedding. Your baby should sleep in the same room with you for at least six months.

Adult beds aren't safe for infants. A baby can become trapped and suffocate between the headboard slats. Those are the spaces between the mattress and the bed frame. A baby also can get trapped in the space between the mattress and the wall. And a baby can suffocate if a sleeping parent accidentally rolls over and covers the baby's nose and mouth.

* **Breastfeed your baby, if possible.** Breastfeeding for at least six months to a year lowers the risk of SIDS.
* **Don't use baby monitors and other commercial devices that claim to reduce the risk of SIDS.** The American Academy of Pediatrics discourages the use of monitors and other devices. These devices do not prevent SIDS. And they cannot be used instead of safe sleep practices.
* **Offer a pacifier.** Sucking on a pacifier at nap time or bedtime may reduce the risk of SIDS. Make sure the pacifier does not have a strap or cord. If you're breastfeeding, wait to offer a pacifier until you and your baby have settled into a nursing routine. It usually takes 3 to 4 weeks to set up a nursing routine.

If your baby is not interested in the pacifier, don't force it. Try again another day. If the pacifier falls out while your baby is sleeping, don't put it back in.

* **Vaccinate your baby.** There's no evidence that recommended shots to protect against diseases increase the risk of SIDS. Some evidence shows that such shots may help prevent SIDS.

## **Treatment**

There's no treatment for SIDS. But your baby's pediatrician or other health care professional can talk with you about any risks your baby may have. And there are ways to help your baby sleep safely.

For the first year, always place your baby to sleep on the back. Use a firm, flat mattress and avoid fluffy pads and blankets. Remove all toys and stuffed animals from the crib. Try using a pacifier. Don't cover your baby's head, and make sure your baby doesn't get too hot. Your baby can sleep in your room, but not in your bed. Breastfeeding for at least six months to a year lowers the risk of SIDS. Vaccine shots to protect your baby from diseases also may help prevent SIDS.

## **Diagnostic Considerations**

A clinician untrained in forensic medicine may inadvertently overlook or destroy gross or trace evidence. Furthermore, misinterpretation of physical injuries or other objective evidence may lead to an inaccurate opinion that, if documented on the chart, may pose considerable problems when used in future court proceedings.

Tragic consequences can follow the misattribution of an infant’s death. One example is that of a young African American couple who were criminally charged after a medical examiner indicated that their baby had died of abandonment—even though autopsy findings were consistent with SIDS and there were no signs of abuse or neglect. The couple spent 6 months in jail because they were unable to post bond before the charges were dismissed.

Similarly egregious examples may be found at the other end of the spectrum, as illustrated by the case of Mary Beth Tining. Only when Tining was charged with the smothering death of her adopted daughter was it discovered that 8 of her biologic children had died, their deaths having been attributed to SIDS or other natural causes. A similar case is that of Waneta Hoyt, who was convicted in 1995 of murdering her 5 children between 1965 and 1971, all of whom were described as having succumbed to SIDS.

In addition to the conditions listed in the differential diagnosis, other problems to be considered include the following:

* Aberrant thermoregulation
* Brain stem tumor
* Cardiac dysrhythmias
* Chiari malformation type I
* Choanal atresia/stenosis
* Central nervous system immaturity
* Congenital central hypoventilation syndrome
* Congenital heart disease
* Craniofacial abnormalities
* Disorders of cardiorespiratory control
* Drowning
* Drug exposure
* Fluid and electrolyte imbalance
* Gastroesophageal reflux
* Heat injury
* Hemangioma
* Lymphangioma
* Laryngomalacia
* Mast cell activation
* Neuromuscular disorders
* Occult trauma
* Pharyngeal/retropharyngeal mass
* Poisoning
* Respiratory syncytial virus infection
* Seizures
* Shaken infant impact syndrome or nonaccidental trauma
* Suffocation
* Toxin exposure
* Tracheobronchial or esophageal foreign bodies
* Tracheoesophageal fistula
* Tracheomalacia
* Upper airway obstruction
* Vascular malformation
* Vascular ring
* Vocal cord paralysis

## **Differential Diagnoses**

* Abdominal Trauma, Blunt
* Acute Anemia
* Alcohol Toxicity
* Anomalous Left Coronary Artery from the Pulmonary Artery
* Aspiration Syndromes
* Bronchiolitis
* CBRNE - Botulism
* Carbon Monoxide Toxicity
* Cardiomyopathy, Hypertrophic
* Child Abuse
* Child Abuse and Neglect
* Chronic Anemia
* Coarctation of the Aorta
* Coronary Artery Anomalies
* Emergent Management of Pediatric Patients with Fever
* Epiglottitis
* Head Trauma
* Hemorrhagic Shock Management in the ED
* Hypocalcemia
* Hyponatremia in Emergency Medicine
* Hypoplastic Left Heart Syndrome
* Hypothermia
* Hypovolemic Shock
* Intestinal Volvulus
* Long QT Syndrome
* Long-Chain Acyl CoA Dehydrogenase Deficiency
* Medium-Chain Acyl-CoA Dehydrogenase Deficiency
* Meningitis, Bacterial
* Meningococcal Infections
* Myocardial Infarction in Childhood
* Myocarditis
* Myocarditis, Viral
* Pediatric Dehydration
* Pediatric Pneumonia
* Pediatric Status Epilepticus
* Pediatrics, Hypoglycemia
* Pediatrics, Inborn Errors of Metabolism
* Pediatrics, Meningitis and Encephalitis
* Pediatrics, Pertussis
* Septic Shock
* Shock, Cardiogenic
* Sleep Apnea
* Ventricular Fibrillation

## **Epidemiology**

### United States statistics

In spring 1992, a multiagency statement directed by officials at the National Institutes of Child Health and Human Development (NICHD) was issued, informing health care providers and the general public that a supine sleep position could significantly reduce SIDS. In spring 1994, this was followed by the federal “Back to Sleep” campaign (see the image below),sponsored by the NICHD, the American Academy of Pediatrics (AAP), the Association of SIDS and Infant Mortality Programs (ASIP), the SIDS Alliance, and the United States Public Health Service.

After the “Back to Sleep” campaign, federal SIDS researchers carried out annual surveys to examine how infant sleep practices and SIDS rates have changed. These studies, conducted by NICHD, demonstrated that the rate of prone sleeping for infants decreased from approximately 75% in 1992 to a low of 11.3% in 2002. The observation that the rate of prone sleeping increased to 14.5% in 2008 is of some concern.

Since 1992, SIDS rates have fallen by approximately 58% in the United States (see the image below). In 1992, the incidence of SIDS was 1.2 cases per 1000 live births; in 2004, the incidence had dropped to 0.51.In 2004, 2246 deaths were certified as SIDS, accounting for 8% of infant deaths.In 2006, the National Center for Health Statistics reported a total of 2323 SIDS deaths nationwide, for an incidence of 0.54 per 1000 live births

Simultaneously with this decrease in SIDS, post neonatal mortalities associated with several other causes of sudden unexpected death have increased significantly, particularly since 1999.It is postulated that some deaths previously classified as SIDS are now being more correctly categorizedand that the true SIDS rate since 1999 may be static. A survey of medical examiners and coroners in 6 jurisdictions found that most used to certify many more deaths as SIDS than they do now.

Although cases of true SIDS are decreasing, concern exists that the proportion of unexplained infant deaths resulting from child abuse may be increasing. Notably, the AAP estimates that the incidence of infanticide among cases designated as SIDS ranges from less than 1% to 5%.

### International statistics

Significant reductions in the prevalence of SIDS have been observed worldwide, though rates of decline have leveled off in the past few years.These changes followed public health campaigns that emphasized the use of the supine sleep position as a simple means of lowering the risk of SIDS. These campaigns began in overseas centers in the late 1980s.

In many Asian countries, the current incidence of SIDS is 0.04 per 1000 live births. Japan has a rate of 0.09/1000,and Hong Kong has a rate of approximately 0.2/1000. Some Scandinavian countries have rates in the range of 0.1-0.06/1000.In Italy, the incidence is 0.7/1000. Before the recommendation of the supine sleeping position, the incidence of SIDS United Kingdom was 3.5/1000; this figure is now 0.41/1000. The incidence in New Zealand was once approximately 4.5/1000 but is now 0.8/1000.

In many of these countries, rates of prone sleeping have fallen to 2-5%. As an illustration of the impact of this single factor, Dwyer et al estimated that 70% of the overall decline in SIDS rates could be attributed to a change to the supine sleep position.They further noted that of 38 additional infant care variables studied, no other individual factor explained more than 7% of the overall decline in SIDS.

With a change to supine sleep for infants, cigarette smoke exposure has emerged as one of the most important potentially modifiable risk factors for SIDS. Infants of mothers who smoke have a 2-fold to 5-fold higher risk of SIDS,and postnatal smoking by one or both parents has been identified as an independent SIDS risk factor.Despite emphasis within “Back to Sleep” campaigns on avoidance of cigarette smoke exposure (prenatal and postnatal), rates of maternal smoking during pregnancy have changed little in most countries.

Other infant care practices have been found capable of modifying the risk of SIDS, as follows:

* Bed-sharing may lead to compromise of an infant’s airway, especially before the age of 3-4 months; sharing a couch or sofa is associated with an unusually high risk for SIDS and should be avoided
* Room-sharing reduces SIDS risk by 50%
* The effect of breastfeeding on SIDS risk varies; in a German infant cohort, breastfeeding reduced the risk of SIDS by approximately 50% at all ages throughout infancy
* Pacifier (dummy) use may reduce the risk of SIDS
* Over bundling of infants during sleep should be avoided; this advice appears to be more important for prone-sleeping infants than for supine-sleeping infants
* Covering the infant’s head with bedding or bed clothing is a risk factor for SID; it is estimated that avoidance of head covering could reduce SIDS deaths by more than one quarter

After "Back to Sleep" initiatives in Germany, Vennemann et al reexamined SIDS risk factors and noted that although only 4.1% of infants slept prone, those infants were at high risk for SIDS.Infants unaccustomed to sleeping prone were at very high risk, as were those who turned to prone. Bed-sharing, duvets, sleeping prone on a sheepskin, sleeping in the house of a friend or a relative, and sleeping in the living room increased the risk of SIDS. Pacifier use during sleep continued to be associated with a significantly reduced risk of SIDS.

### Age-related demographics

About two-thirds of SIDS deaths occur in infants aged 2-4 months. Ninety percent of deaths occur in children younger than 6 months, and 95% of deaths occur in children younger than 8 months; few occur in children younger than 1 month or older than 8 months. This age-at-death profile suggests a relation to neurobiological components of infant development (see Pathophysiology). Slightly higher proportions of SIDS-certified deaths occurring in the neonatal period and after 6 months were reported in 2001 than were reported in 1992.

### Sex-related demographics

Approximately 60-70% of SIDS deaths occur in males. Despite other notable changes in SIDS epidemiology, the male-to-female ratio has remained relatively unchanged in most population studies.

### Race-related demographics

Risk among racial and ethnic groups in the United States varies substantially. In 2003, SIDS rates were highest for American Indian/Alaskan Native and non-Hispanic Black mothers—2.5 and 2.2 times higher, respectively, than the rate for non-Hispanic white mothers. African Americans are twice as likely to place infants prone for sleep, and they are also twice as likely to bed-share than other racial groups are.

In contrasted, the SIDS rate for Mexican mothers was 51% lower than the rate for non-Hispanic white mothers, and the SIDS rate for Central American and South American mothers was 62% lower.The following data have been reported for the incidence of SIDS in various racial and ethnic groups (as number of cases per 1000 live births):

* Central Americans and South Americans - 0.20
* Asian/Pacific Islanders - 0.28
* Mexicans - 0.24
* Puerto Ricans - 0.53
* Whites - 0.51
* African Americans - 1.08
* American Indian - 1.24

These variations remain unexplained but appear to be independent of other risk factors, such as low birth weight, young maternal age, or high parity. They appear to mirror those observed for infant mortality in general.

**GUIDLINES**

* AAP recommends a safe sleep environment that can reduce the risk of all sleep-related infant deaths. Recommendations for a safe sleep environment include supine positioning, the use of a firm sleep surface, room-sharing without bed-sharing, and the avoidance of soft bedding and overheating.
* Additional recommendations for SIDS reduction include the avoidance of exposure to smoke, alcohol, and illicit drugs; breastfeeding; routine immunization; and use of a pacifier.
* New evidence is presented for skin-to-skin care for newborn infants, use of bedside and in-bed sleepers, sleeping on couches/armchairs and in sitting devices, and use of soft bedding after 4 months of age.
* Offer a pacifier at nap time and at bedtime. Studies show these can reduce the risk for SIDS.
* Infants should be immunized in accordance with AAP and CDC recommendations.
* Provide supervised, awake tummy time daily to facilitate development.
* Remove infants from car seats, strollers, swings, infant carriers, and infant slings, if they fall asleep in them, to reduce the risk for gastroesophageal reflux and positional plagiocephaly.
* Do not use home cardiorespiratory monitors as a strategy to reduce the risk of SIDS.

unexplained sudden death occurring in a child younger than 1 year with negative pathological and toxicological assessment be called “sudden unexplained death in infancy” (SUDI). Additional recommendations are summarized below.

Evaluation

* Personal/family history and circumstances of the sudden death, along with blood and/or tissue for molecular autopsy should be collected in all cases of SUDI (Class I)
* An arrhythmia syndrome focused molecular autopsy/postmortem genetic testing can be useful (Class IIa)
* Considered assessment by an expert cardiac pathologist to rule out the presence of microscopic indicators of structural heart when a diagnosis of SUDI is made at autopsy. (Class IIb)

Follow-up Screening of First-degree relatives

* First-degree relatives should undergo genetic testing whenever a pathogenic mutation in a gene associated with increased risk of sudden death is identified by molecular autopsy. Obligate carriers should be prioritized.
* First-degree relatives with a family history of inherited heart disease should be evaluated with resting ECG and exercise stress testing. Additional tests as indicated can be useful. Those with a history of arrhythmias or syncope should be prioritized. (Class IIa)
* Follow-up clinical assessment in young family members with a family history of inherited heart disease or other sudden unexplained death syndrome (SUDS) or SUDI death who may manifest symptoms and/or signs of the disease at an older age and in all family members whenever additional SUDS or SUDI events occur. (Class IIa)
* Consider evaluation of first-degree relatives with resting ECG and exercise stress testing. (Class IIb)

The death of an infant may be attributed to sudden infant death syndrome (SIDS) when all of the following are true:

* A complete autopsy is performed, including cranium and cranial contents, and autopsy findings are compatible with SIDS
* No gross or microscopic evidence of trauma or significant disease process is present
* No evidence of trauma exists on skeletal survey
* Other causes of death are adequately ruled out, including meningitis, sepsis, aspiration, pneumonia, myocarditis, abdominal trauma, dehydration, fluid and electrolyte imbalance, significant congenital lesions, inborn metabolic disorders, carbon monoxide asphyxia, drowning, and burns
* No evidence of current alcohol, drug, or toxic exposure is present
* Thorough death scene investigation and review of the clinical history are negative

Circumstances could indicate the possibility of intentional suffocation include:

* Recurrent cyanosis, apnea, or ALTEs occurring only while in the care of the same person;
* Age at death older than 6 months;
* Previous unexpected or unexplained deaths of 1 or more siblings;
* Simultaneous or nearly simultaneous death of twins
* Previous death of infants under the care of the same unrelated person
* Evidence of previous pulmonary hemorrhage (such as marked siderophages in the lung).

marked change in muscle tone (hyper- or hypotonia); or altered responsiveness. The guidelines also add that a BRUE is diagnosed only when there is no explanation for a qualifying event after completion of a thorough history and physical examination. Infants younger than 1 year who present with a BRUE are categorized either as (1) a lower-risk patient on the basis of history and physical examination for whom evidence-based recommendations for evaluation and management are offered or, (2) a higher-risk patient whose history and physical examination suggest the need for further investigation and treatment but for whom recommendations are not offered. This clinical practice guideline is structured to promote a patient- and family-centered approach to care, reduce unnecessary and potentially costly medical interventions, improve patient outcomes, and enhance efforts at future research.

The form contains 25 questions that medical examiners and coroners should ask before beginning an autopsy.

The following information is collected on the SUIDIRF:

* Investigation data.
* Witness interview.
* Infant’s medical history.
* Infant’s dietary history.
* Pregnancy history.
* Incident scene investigation.
* Investigation summary.
* Investigation diagrams.
* Summary for pathologist.

### **Prevention**

Since 1996, the American Academy of Pediatrics (AAP) has recommended infants be placed in the supine position for sleep. In 2011, the AAP issued a policy statement expanding its recommendations to ensure a safe sleeping environment for infants and to further reduce the risk for sudden infant death syndrome (SIDS).

Other key recommendations are:

* Breastfeeding is recommended
* Always use a firm sleep surface. Car seats and other sitting devices are not recommended for routine sleep.
* The baby should sleep in the same room as the parents, but not in the same bed (room sharing without bed sharing).
* Keep soft objects or loose bedding out of the crib.
* Wedges and positioners should not be used.
* Offer a pacifier at nap time and bedtime.
* Avoid covering the infant's head or overheating.
* Do not use home monitors or commercial devices marketed to reduce the risk for SIDS.
* Supervised, awake tummy time is recommended daily to facilitate development and minimize the occurrence of positional plagiocephaly (flat heads).
* Avoid alcohol, illicit drug use and smoke exposure during pregnancy and after birth
* Infants should be immunized in accordance with recommendations of the AAP and Centers for Disease Control and Prevention

**PREDEFINED QUESTIONS AND ANSWERS**

## What is SIDS?

Sudden Infant Death Syndrome (SIDS) is the sudden, unexpected death of an infant under 1 year of age that remains unexplained after a thorough investigation, including autopsy, examination of the death scene, and review of the clinical history.

## What causes SIDS?

The exact cause of SIDS is unknown. It likely results from multiple factors including brain abnormalities affecting breathing and arousal, genetic predisposition, and environmental factors such as unsafe sleep environments.

## Who is at risk for SIDS?

Risk factors include:

* Infants aged 1 to 4 months (peak risk)
* Male infants slightly more than females
* Premature or low birth weight babies
* Exposure to tobacco smoke during pregnancy or after birth
* Sleeping on stomach or side instead of back
* Sleeping on soft surfaces or with loose bedding
* Overheating during sleep
* Bed sharing with adults or siblings

## How can SIDS be prevented?

Prevention strategies include:

* Always placing babies to sleep on their backs
* Using a firm sleep surface without soft bedding or toys
* Avoiding bed-sharing and overheating
* Keeping the sleep environment smoke-free
* Offering a pacifier at nap time and bedtime (once breastfeeding is established)
* Ensuring regular prenatal care and avoiding smoking during pregnancy

## What are the signs or symptoms before SIDS?

Most infants do not show any warning signs before SIDS. It is usually a sudden, unexpected event during sleep.

## What should parents do if they find their baby unresponsive?

Call emergency services immediately and start infant CPR if trained. Early intervention may be life-saving in other critical conditions mimicking SIDS.

## Is SIDS hereditary?

SIDS is generally not considered hereditary, but some genetic factors may contribute to susceptibility.

## What happens after a SIDS diagnosis?

A thorough medical and forensic investigation is done. Support and counseling are offered to grieving families. Families may be referred to support groups and mental health resources.

**DOCTOR PATIENT CONVERSATION**

Doctor: I’m very sorry for your loss. I want to talk with you about what happened with your baby and explain what Sudden Infant Death Syndrome, or SIDS, means. Do you have any questions before we start?

Parent: Thank you, doctor. I don’t really understand what SIDS is. Why did this happen?

Doctor: SIDS is the sudden and unexpected death of an otherwise healthy baby under one year of age, and even after a thorough examination, including an autopsy and investigation of the death scene, we cannot find a clear cause. It is very distressing because it happens without warning.

Parent: Is there something I did wrong? Could it have been prevented?

Doctor: It’s important to know that SIDS is not caused by anything you did or didn’t do. However, certain factors increase the risk, like babies sleeping on their stomachs or soft bedding, exposure to smoke, or overheating. That’s why safe sleep practices are strongly recommended—to reduce the risk as much as possible.

Parent: What do you mean by safe sleep?

Doctor: Safe sleep means putting your baby on their back to sleep, using a firm mattress without pillows or toys in the crib, keeping the room at a comfortable temperature, and avoiding smoking around your baby. Breastfeeding and using a pacifier during sleep can also reduce the risk.

Parent: Will this happen again if I have another baby?

Doctor: The risk for SIDS is low in future babies, but it’s natural to feel anxious. We encourage following safe sleep guidelines and regular pediatric care. There are also support groups for families who have experienced this loss.

Parent: What should I look out for? Are there any signs before this happens?

Doctor: Unfortunately, most babies who die from SIDS don’t show warning signs. That’s why prevention through safe sleeping environments is so important.

Parent: Is there anything more we can do medically?

Doctor: At this time, there is no treatment to prevent SIDS because its exact cause is unknown. Our focus is on education, prevention, and supporting families through this difficult time.

Parent: Thank you for explaining. This is very hard to understand, but I appreciate your help.

Doctor: I know this is an incredibly tough time. Please don’t hesitate to reach out if you have questions or need support. We’re here for you.

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## Transient tachypnea of the newborn

**DEFINITION AND DESCRIPTION**

Transient tachypnea of the newborn, or TTN, is a respiratory disorder usually seen shortly after delivery in babies who are born near or at term. Transient means it is short lived (usually less than 24 hours) and tachypnea means rapid breathing.

## **CAUSES**

Before babies are born, they have fluid in their lungs. Babies reabsorb some of that fluid because of hormone changes that happen before birth. More fluid gets reabsorbed as they pass through the birth canal during delivery. The rest of the fluid is absorbed into the lungs after they are born and start breathing on their own. If the fluid isn't absorbed fast enough or if they have too much fluid in the lungs, they can't take in oxygen very well. Babies with this problem have to breathe faster and harder to get enough oxygen into the lungs.

## **RISK FACTOR**

Only a small number of all newborn babies get this breathing problem. Although premature babies can have it, most babies with this problem are full-term. Babies delivered by C-section (without labor) are more likely to have this condition. This is because without the hormone changes of labor the fluid in the lungs is still there. The baby has to work to reabsorb it after birth. Babies of moms with asthma and diabetes may also be more likely to have this condition.

## **Symptoms**

Newborns with TTN have respiratory problems soon after birth (within one to two hours). These usually consist of some combination of rapid, noisy breathing (grunting) and/or the use of extra muscles to breathe (flaring nostrils or movements between the ribs or breastbone known as retractions).

## **Diagnosis**

The mother’s pregnancy and labor history are important to make the diagnosis. A chest X-ray may be taken to eliminate other causes of respiratory problems. A blood count and blood culture may be drawn to try to rule out infection. TTN is usually diagnosed after monitoring your baby for one to two days.

## **Treatment**

Your baby will be given oxygen as needed to maintain an adequate blood oxygen level. Pulse oximeter and/or blood gases may be used. Your baby’s oxygen requirement will usually be highest within a few hours after birth and then begin to decrease. Most infants with TTN improve in 12 to 24 hours.

If your baby is breathing very rapidly, feedings may be withheld, and intravenous fluids may be given for nutrition until he or she improves. Your baby may also receive antibiotics during this time until infection is ruled out. Rarely, babies with TTN may have persistent lung problems for as long as one week.

## **Prognosis**

TTN usually resolves completely within 24 hours after delivery. Babies who have had TTN usually have no further problems from it and require no special care or follow-up other than their routine pediatrician visits.

## **Diagnostic Considerations**

Transient tachypnea of the newborn (TTN) is a diagnosis of exclusion. It is important not to assume that respiratory distress is solely transient tachypnea of the newborn and to consider other severe etiologies such as congenital cyanotic heart disease. Quickly assuming a diagnosis of transient tachypnea of the newborn limits the evaluation of the infant, potentially resulting in misdiagnosis and inappropriate therapy.

Further investigation should be performed with infants whose conditions are not improving or who are having worsening respiratory distress, particularly when incubator support is needed.

## Other conditions to consider

In addition to the differentials list below, other conditions to consider in the differential diagnosis include the following:

* Hyperventilation
* Congenital heart disease
* Congenital malformations
* Metabolic disorders
* Pulmonary edema
* Pneumonia
* Aspiration
* Air leaks

## **Differential Diagnoses**

* Congenital Pneumonia
* Meconium Aspiration Syndrome
* Metabolic Acidosis
* Neonatal Sepsis
* Pediatric Pneumothorax
* Pediatric Pulmonary Hypoplasia
* Persistent Pulmonary Hypertension of the Newborn (PPHN)
* Pneumomediastinum
* Polycythemia of the Newborn
* Respiratory Distress Syndrome

## **Epidemiology**

### United States data

Approximately 1% of infants have some form of respiratory distress that is not associated with infection. Respiratory distress includes both respiratory distress syndrome (RDS) (ie, hyaline membrane disease) and transient tachypnea of the newborn (TTN). Of this 1%, approximately 33-50% have transient tachypnea of the newborn (TTN).

### Race-, sex-, and age-related demographics

No racial predilection has been reported. Male neonates are more affected than females.

Clinically, transient tachypnea of the newborn presents as respiratory distress in full-term or near-term infants.

**Treatment of Transient Tachypnea of the Newborn (TTN) — Drugs and Side Effects**

## 1. Supportive Care (Primary Treatment)

* Oxygen therapy: To maintain adequate oxygen saturation; delivered via nasal cannula, oxygen hood, or CPAP if needed.
* Monitoring: Close observation and respiratory support as required. Most infants recover within 2–3 days.

## 2. Antibiotics

* Common regimen: Combination of an aminoglycoside (e.g., *gentamicin*) plus a beta-lactam antibiotic (e.g., *ampicillin* or *penicillin*) may be used if infection cannot be ruled out initially, as symptoms may overlap with neonatal infection.
* Potential side effects:
  + *Gentamicin*: Ototoxicity (hearing loss), nephrotoxicity (kidney damage), requires blood level monitoring.
  + *Ampicillin*: Allergic reactions, rash, gastrointestinal upset.
* Cautions: Use of antibiotics can alter gut flora and possibly increase risk of necrotizing enterocolitis (NEC); therefore, antibiotics are used judiciously only when infection is suspected or cannot be excluded.

## 3. Inhaled Beta-2 Agonists (e.g., Salbutamol/Albuterol)

* Some studies suggest inhaled salbutamol may decrease the severity and shorten the duration of TTN by promoting fluid clearance from the lungs.
* Side effects: Possible tachycardia, jitteriness, hypokalemia, but generally well tolerated in neonates when used carefully.

## 4. Diuretics

* Occasionally used to promote removal of lung fluid, though no strong evidence supports routine use

**GENOMIC DATA**

* Beta-adrenergic receptor gene polymorphisms:  
  A 2008 study found that loss-of-function variations in the beta1-adrenergic receptor gene (ADRB1), specifically the beta1Gly49 homozygosity, significantly increase susceptibility to TTN. Similarly, a particular TACC haplotype in the beta2-adrenergic receptor gene (ADRB2) is associated with higher TTN risk. These receptor genes modulate the catecholamine-driven process responsible for lung fluid absorption after delivery.
* Mechanism:  
  Catecholamines released during labor stimulate lung epithelial sodium channels and inhibit chloride pumps, promoting rapid clearance of fetal lung fluid. Genetic variations that reduce receptor function may impair this process, leading to fluid retention in the lungs and resulting in TTN.
* Epidemiological associations:  
  TTN occurs more frequently in male infants, those born by cesarean section without labor, and infants with lower gestational age and birthweight. Genetic predisposition acts alongside these environmental and clinical factors.

**PREDEFINED QUESTIONS AND ANSWERS**

## What is Transient Tachypnea of the Newborn (TTN)?

TTN is a common cause of respiratory distress in newborns caused by delayed clearance of fetal lung fluid after birth. It usually presents with rapid breathing shortly after delivery and typically resolves within 48–72 hours.

## What causes TTN?

TTN occurs when the newborn’s lungs retain extra fluid that normally clears quickly after birth. This can happen due to cesarean delivery without labor, premature birth, or other factors that reduce the baby’s ability to absorb lung fluid.

## What are the symptoms of TTN?

* Rapid breathing (tachypnea), usually over 60 breaths per minute
* Mild grunting or nasal flaring
* Mild chest retractions
* Possible mild cyanosis (bluish skin)
* Generally normal blood oxygen levels or mild hypoxia

## How is TTN diagnosed?

Diagnosis is primarily clinical, supported by chest X-ray showing fluid in the lung fissures, prominent pulmonary vasculature, and hyperinflation. Other causes of respiratory distress must be excluded, such as respiratory distress syndrome or pneumonia.

## How is TTN treated?

Treatment is supportive:

* Oxygen therapy if needed
* Monitoring of breathing and oxygen saturation
* Rarely, nasal continuous positive airway pressure (CPAP) for assistance
* Most infants improve within 2–3 days without specific medication

## What is the prognosis for TTN?

The prognosis is excellent. TTN is transient and resolves spontaneously typically within a few days without long-term effects.

## How can TTN be prevented?

Cesarean delivery without labor and preterm birth are risk factors, but there is no guaranteed prevention. Encouraging labor before cesarean when possible may reduce risk.

**DOCTOR PATIENT CONVERSATION**

Doctor: Hello, I want to talk with you about your baby’s breathing issue called transient tachypnea of the newborn, or TTN. Do you have any questions before we begin?

Parent: Yes, doctor. What exactly is TTN?

Doctor: TTN is a mild breathing problem that usually happens within the first few hours after birth. Basically, babies are born with fluid in their lungs, and normally this fluid clears quickly once they start breathing air. Sometimes, though, extra lung fluid sticks around for a bit longer, making it harder for the baby to breathe comfortably. That’s what we call TTN.

Parent: What causes this fluid to stay in the lungs?

Doctor: When a baby passes through the birth canal during labor, the pressure and certain hormone changes help push fluid out of the lungs. Babies born by cesarean section without labor don’t get that benefit, so they’re more likely to have extra lung fluid at birth. Other factors like maternal asthma or diabetes can also increase the risk.

Parent: What symptoms should I watch for?

Doctor: You might notice your baby is breathing faster than normal — more than 60 breaths per minute — along with grunting, flaring of the nostrils, or pulling in around the ribs with each breath. Sometimes the skin around their mouth may look a little bluish because they’re working harder to get enough oxygen.

Parent: How do you know it’s TTN and not something more serious?

Doctor: We do a chest X-ray which usually shows extra fluid in the lungs and rules out other problems like pneumonia or respiratory distress syndrome. We also monitor oxygen levels and may do blood tests to check for infection. TTN tends to get better within a few days on its own, which is a key sign.

Parent: How is TTN treated?

Doctor: Most babies just need supportive care. We give oxygen if needed, sometimes through a mask or nasal prongs. If their breathing is very fast or they’re having trouble feeding, they might get extra fluids through an IV or a feeding tube until they’re stronger. Rarely, we might use a breathing support machine called CPAP. Antibiotics may be started if there’s concern for infection, but usually discontinued if tests are negative.

Parent: Will my baby recover fully?

Doctor: Yes. TTN usually resolves within 2 to 3 days without long-term problems. Your baby should improve steadily once the lung fluid clears.

Parent: What can we do to prevent TTN with future babies?

Doctor: If possible, allowing labor to start before a cesarean section can help, but many cases are unpredictable. Overall, TTN is uncommon and usually temporary.

Parent: Thank you, doctor. This helps me understand what’s going on.

Doctor: You’re very welcome. We’ll keep monitoring your baby closely and keep you informed every step of the way. Please let us know if you notice any worsening breathing or feeding problems.

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**TOTAL ANOMALOUS PULMONARY VENOUS RETURN**

**DEFINITION AND DESCRIPTION**

Total anomalous pulmonary venous return (TAPVR) happens when veins that bring blood from your baby’s lungs connect to the wrong place in their heart. As a result, their heart can’t put enough oxygen into the blood it sends to the rest of their body. TAPVR is a life-threatening heart condition. It’s congenital, which means it’s present at birth.

Typically, your lungs send oxygen-rich blood to your heart’s left atrium (the chamber in your heart’s upper left side). In babies with TAPVR, the oxygen-rich blood flows through pulmonary veins to their heart’s right atrium instead.

In the right atrium, the oxygen-rich blood mixes with blood that doesn’t have as much oxygen. This low-oxygen blood travels out of the heart to the rest of your baby’s body.

Babies with total anomalous pulmonary venous return may have trouble breathing soon after birth. Their skin may appear blue (or gray on darker skin).

All children with TAPVR also have an atrial septal defect. This is a hole between the heart’s right and left atria.

Total anomalous pulmonary venous return is a rare condition. It affects 1 in about 7,800 newborns in the U.S. each year. You may hear a provider call this condition total anomalous pulmonary venous connection (TAPVC).

### **Types of TAPVR**

TAPVR types differ by how blood reaches the wrong place — your baby’s right atrium.

The TAPVR types are:

* **Supracardiac TAPVR**: Blood flows via an ascending vertical vein into the brachiocephalic vein. Then, it flows through their superior vena cava (a large vein in their body).
* **Cardiac TAPVR**:Blood moves through their coronary sinus. This vein normally drains blood that contains low levels of oxygen.
* **Infracardiac TAPVR**:The connecting vertical vein comes from their liver and inferior vena cava.
* **Mixed TAPVR**:A combination of veins from your baby’s lungs drain to more than one of the types mentioned above. This is the rarest and most difficult form to repair successfully.

## **Symptoms and Causes**

Symptoms of total anomalous pulmonary venous return may include:

* Cyanosis (bluish or grayish skin, nails and lips)
* Difficulty feeding
* Fatigue and lethargy (lack of energy)
* Heart murmur
* Shortness of breath (dyspnea) and trouble breathing
* Lack of growth or weight gain

Symptoms of TAPVR usually appear very soon after birth. Babies have symptoms at birth if they have any narrowing where their lung veins connect to their hearts. But some babies don’t have symptoms for several weeks.

### **TAPVR causes**

Healthcare providers aren’t sure what causes total anomalous pulmonary venous return. It happens when your baby’s heart and blood vessels are forming in the uterus during fetal development.

This doesn’t seem to be an inherited condition (passed down through families). But there have been similar cases among siblings. TAPVR has an association with certain syndromes and exposure to paint removers, pesticides and lead.

**Complications of this condition**

A baby with TAPVR can have heart failure and pulmonary hypertension. Even after the initial repair, TAPVR can sometimes lead to abnormal heart rhythms or blockages in your child’s pulmonary veins.

Children with TAPVR may have trouble with typical development, like fine motor function. You can help your child by getting screenings for developmental milestones. If a screening shows a developmental delay, you can arrange for someone to work with them to improve their skills.

Talk to your child’s healthcare provider about their ability to take part in sports and physical activities. Your child may need to limit vigorous exercise.

## **Diagnosis and Tests**

Cardiologists can sometimes diagnose this condition with an echocardiogram before birth. Some babies don’t get a diagnosis until they’re several weeks or months old.

After your baby is born, their provider will do a physical exam and listen to your baby’s heart. A pulse oximeter (pulse ox) can measure the amount of oxygen in your baby’s blood. It fits over your baby’s big toe and sends information through a wire.

To diagnose total anomalous pulmonary venous return, a provider may use:

* Chest X-ray
* CT (computed tomography) scan
* Heart MRI (magnetic resonance imaging)
* Echocardiogram
* Electrocardiogram (EKG or ECG)

Some of these tests (MRI) may require anesthesia, but they’re all noninvasive. They allow your baby’s healthcare provider to see images of your baby’s heart and veins. They also help them evaluate blood flow and look for abnormalities.

A provider can use cardiac catheterization to make a diagnosis. But they can usually get the information they need from imaging. This spares your baby from an invasive procedure.

## **Management and Treatment**

Healthcare providers treat TAPVR with open-heart surgery. Most often, providers perform this TAPVR repair as soon as they can after diagnosing the condition. Nearly every baby with total anomalous pulmonary venous return needs surgery to survive.

Though it’s extremely rare, some people don’t have surgery as babies and get a TAPVR diagnosis as adults. But they usually have high pressures within their lungs (pulmonary hypertension). This can make surgical repair very challenging.

While waiting for surgery, your child may receive extra oxygen or a ventilator to help them breathe. They may receive an inotrope, which is a medicine that helps their heartbeat more forcefully.

While your baby is asleep under general anesthesia, a surgeon:

1. Makes cuts (incisions) in your baby’s chest and heart
2. Connects the pulmonary veins to the correct place (the left atrium) in your baby’s heart
3. Closes abnormal connections
4. Removes any blockage in your child’s pulmonary vein
5. Most often, closes the hole between their left and right atria (atrial septal defect)

### **When to see a doctor**

After TAPVR surgery, your baby will have checkups every six to 12 months. They’ll need regular follow-up visits through adulthood. Your child’s provider may want to order tests like an electrocardiogram, exercise stress test or echocardiogram.

Get medical help right away if your baby:

* Has bluish or grayish skin, nails or lips
* Has trouble breathing or difficulty eating
* Seems lethargic

Questions to consider asking your child’s healthcare provider may include:

* What type of TAPVR does my child have?
* Do you think my child will need a second operation?
* How many times have you performed surgery to repair TAPVR?

## **Outlook / Prognosis**

Babies who have TAPVR need an operation. With early surgery, most children with TAPVR survive into adulthood. But some will need repeat surgery or procedures to treat narrowing in their veins later in life. Because of this, people with TAPVR need to see a cardiologist (a heart expert) regularly to monitor their health following surgery.

Without surgery, some forms of total anomalous pulmonary venous return are typically fatal a few weeks after birth. With early diagnosis and surgical treatment, the outlook for babies with TAPVR is very good. The survival rate after surgery is around 97%.

Your child will need regular visits with their cardiologist as they grow into adulthood. Lifelong follow-up visits can help cardiologists find problems like an irregular heartbeat or blockages (obstructions) in their blood vessels. An obstruction requires another surgery and may be hard to treat.

Your child may need to take medicine or have a procedure like a cardiac catheterization.

**Prevention**

There’s nothing you could’ve done to prevent this congenital heart condition. But if you’re thinking of starting a family and have TAPVR or a family history of congenital heart conditions, you might want to speak with a healthcare provider.

**DIFFERENTIAL DIAGNOSIS OD TAPVR**

**TAPVC**

The differential diagnoses for TAPVC includes the following:

1. Obstructed TAPVC

* Respiratory Distress Syndrome: Signs typically begin after birth, unlike TAPVC, where the presentation is slightly delayed (typically occurring after 12 hours of life).
* Persistent pulmonary hypertension of the newborn

2. Unobstructed TAPVC:

* Large ASD, VSD
* Truncus arteriosus
* AV canal defects
* Single ventricle lesions
* Respiratory Distress Syndrome (RDS) — typically in preterm infants, surfactant deficiency, early onset respiratory distress
* Persistent Pulmonary Hypertension of the Newborn (PPHN) — hypoxia with normal heart anatomy and elevated pulmonary pressures
* Other cyanotic congenital heart diseases (CHDs):
  + Tetralogy of Fallot (TOF)
  + Transposition of the Great Arteries (TGA)
  + Hypoplastic Left Heart Syndrome (HLHS)
* Partial Anomalous Pulmonary Venous Connection (PAPVC) — some pulmonary veins drain abnormally, often milder symptoms
* Atrial Septal Defect (ASD) — may coexist, needs to be distinguished as a primary diagnosis
* Congestive heart failure from other causes — structural or functional cardiac issues
* Neonatal pneumonia or sepsis — infectious causes of respiratory distress
* Congenital diaphragmatic hernia (CDH) — pulmonary compromise due to herniated abdominal contents
* Airway anomalies or obstruction — rare but should be considered
* Pulmonary venous obstruction conditions mimicking TAPVR physiology

**Epidemiology of Total Anomalous Pulmonary Venous Return (TAPVR):**

* TAPVR is a rare congenital heart defect, occurring in approximately 0.6 to 1.2 per 10,000 live births (about 1 in 8,000 to 1 in 16,000 births) .
* It accounts for roughly 0.7% to 1.5% of all congenital heart diseases .
* In the United States, around 1 in 7,800 babies is born with TAPVR, equating to approximately 500 cases annually .
* There is a marked male predominance, with a male-to-female ratio of about 4:1—particularly notable in the infracardiac type of TAPVR .
* TAPVR typically presents in early infancy, often within the first few months of life, as survival without surgical correction is rare; median survival without repair is about 2 months, with 50% mortality in the first 3 months.
* TAPVR is classified into types (supracardiac, cardiac, infracardiac, and mixed), with supracardiac being the most common subtype (~47%).
* The mixed type TAPVR is rarer (~3-20% depending on series) and associated with higher surgical mortality rates compared to other types.
* Survival into adulthood without repair is exceedingly rare but can occur, especially in cases with large atrial septal defects allowing adequate systemic circulation .
* Advances in surgical techniques have improved survival rates, but mortality remains significant, particularly in complex or obstructed variant

**PREDEFINED QUESTIONS AND ANSWERS**

## What is Total Anomalous Pulmonary Venous Return (TAPVR)?

TAPVR is a rare congenital heart defect where the pulmonary veins, which normally carry oxygen-rich blood from the lungs to the left atrium, instead connect abnormally to the right atrium or systemic veins. This causes oxygen-rich blood to mix with oxygen-poor blood, affecting oxygen delivery to the body.

## What causes TAPVR?

The exact cause is unknown, but it occurs during fetal development when the pulmonary veins fail to connect properly to the left atrium. TAPVR can occur alone or with other heart or body anomalies.

## What are the types of TAPVR?

There are four main types based on where the pulmonary veins drain:

* Supracardiac (most common) — veins drain above the heart to systemic veins like the superior vena cava
* Cardiac — drainage directly to the heart’s right atrium or coronary sinus
* Infracardiac — drainage below the heart, often through the liver veins or portal system (usually obstructed)
* Mixed — combination of two or more types

## What symptoms does TAPVR cause?

Symptoms depend on whether there is obstruction of pulmonary venous return:

* Without obstruction: mild cyanosis, signs of heart failure, fatigue, poor feeding
* With obstruction: severe cyanosis, respiratory distress, pulmonary edema, and rapid deterioration shortly after birth

## How is TAPVR diagnosed?

* Echocardiography is the main tool to visualize abnormal pulmonary venous connections.
* Chest X-ray may show cardiomegaly, pulmonary edema, or a characteristic “snowman” shape in supracardiac type.
* Sometimes, cardiac MRI or catheterization is performed.

## How is TAPVR treated?

* TAPVR requires surgical correction to reroute pulmonary veins to the left atrium.
* Medical management before surgery includes treating heart failure with diuretics and supporting oxygenation.
* Obstructed TAPVR is a surgical emergency.

## What is the prognosis for TAPVR?

* Without surgery, TAPVR is usually fatal in infancy.
* Surgical repair has improved survival significantly, but long-term follow-up is needed for potential complications like pulmonary hypertension or arrhythmias.

## Can TAPVR be prevented?

* There is no known prevention since it arises from developmental abnormalities during pregnancy.

## What should families expect after surgery?

* Most children improve markedly, but require lifelong cardiology care.
* Endocarditis prophylaxis is recommended for six months post-surgery or longer if residual defects exist.

**DOCTOR PATIENT CONVERSATION**

Doctor: Hello, I want to talk about your baby’s heart condition called Total Anomalous Pulmonary Venous Return, or TAPVR. Do you have any questions before we start?

Parent: I’ve never heard of TAPVR before. What exactly is it?

Doctor: TAPVR is a rare congenital heart defect where the veins that bring oxygen-rich blood from the lungs don’t connect to the left side of the heart as they should. Instead, they connect abnormally to the right side or other large veins. This means oxygen-rich blood mixes with oxygen-poor blood, which can reduce the amount of oxygen delivered to the body.

Parent: That sounds serious. What causes this?

Doctor: The exact cause isn’t known—it happens during your baby’s heart development before birth. It’s not caused by anything you did or didn’t do during pregnancy.

Parent: What kind of problems does TAPVR cause in my baby?

Doctor: The problem depends on whether the abnormal veins are obstructed or not. If the blood flow returning from the lungs is blocked, your baby may have severe breathing difficulties and very low oxygen levels soon after birth. Without obstruction, symptoms may be milder but can still include tiredness, poor feeding, and signs of heart failure.

Parent: How do you diagnose TAPVR?

Doctor: We use an echocardiogram, which is an ultrasound of the heart, to see exactly how the veins connect. Chest X-rays can also show signs that support the diagnosis. Sometimes more advanced imaging is used to plan treatment.

Parent: Can TAPVR be fixed?

Doctor: Yes. TAPVR requires surgery to reconnect the pulmonary veins to the left atrium, which is the normal heart chamber they should drain into. Before surgery, we may use medications to manage heart failure symptoms or support breathing if needed.

Parent: Is the surgery risky?

Doctor: Surgery always carries some risk, but it has greatly improved over the years. Most babies recover well, especially if the surgery is done early. Some forms of TAPVR and cases with obstruction have a higher risk, so urgent surgery might be necessary.

Parent: What happens after surgery?

Doctor: Your baby will be closely monitored in the hospital for several days to weeks. Long-term follow-up with a pediatric cardiologist is essential to watch for any complications, like narrowing of the veins or heart rhythm problems.

Parent: Is there anything I need to do now?

Doctor: Right now, we want to stabilize your baby and prepare for surgery. We will provide all the support your baby needs and keep you informed every step of the way.

Parent: Thank you for explaining all this. It’s a lot to take in, but I appreciate your help.

Doctor: I know it’s overwhelming. Please don’t hesitate to ask questions at any time. We’re here to support you and your family

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**THORACIC TRAUMA**

**DEFINITION AND DESCRIPTION**

Thoracic trauma refers to injuries sustained to the chest region, including the thoracic cage, lungs, heart, great vessels, and other structures. It can result from various causes, such as motor vehicle accidents, falls, penetrating injuries, or blunt trauma. This comprehensive article aims to explore the different types and causes of thoracic trauma, its clinical presentation, diagnostic approaches, and management strategies. By understanding the complexities of thoracic trauma, healthcare professionals can provide timely and effective care to improve patient outcomes.

## **Causes and Types of Thoracic Trauma:**

Thoracic trauma can occur due to a range of causes, including:

* Blunt trauma: This is the most common cause of thoracic trauma and typically results from motor vehicle accidents, falls, or physical assaults. Blunt trauma can cause rib fractures, lung contusions, pneumothorax, hemothorax, or cardiac injuries.
* Penetrating trauma: Stab wounds or gunshot wounds can directly injure the thoracic structures, including the lungs, heart, or major blood vessels. Penetrating trauma may lead to pneumothorax, hemothorax, cardiac tamponade, or vascular injuries.
* Deceleration injuries: Sudden deceleration forces, such as those experienced in motor vehicle accidents, can cause significant thoracic trauma. This may result in aortic tears, pulmonary contusions, or rib fractures.
* Crush injuries: Crush injuries, often associated with industrial accidents or building collapses, can cause severe thoracic trauma, including rib fractures, lung contusions, and traumatic asphyxia.
* Blast injuries: Explosions or blast events can cause primary, secondary, or tertiary blast injuries, leading to a wide range of thoracic trauma, including lung contusions, pneumothorax, and pulmonary barotrauma.

## **Diagnosis:**

The clinical presentation of thoracic trauma can vary depending on the type and severity of the injury. Common signs and symptoms include:

* Chest pain and tenderness: Patients may experience localized pain and tenderness over the chest area, particularly in the region of rib fractures or soft tissue injuries.
* Difficulty breathing: Thoracic trauma can lead to respiratory distress, shortness of breath, or shallow breathing due to lung contusions, pneumothorax, or hemothorax.
* Cyanosis: In severe cases, inadequate oxygenation may result in cyanosis, characterized by a bluish discoloration of the skin and mucous membranes.
* Hemoptysis: Blood-tinged sputum or coughing up of blood may occur in cases of lung contusions or major vascular injuries.
* Abnormal breath sounds: Auscultation of the chest may reveal decreased breath sounds on the affected side in the presence of pneumothorax or lung collapse.

Diagnostic approaches for thoracic trauma may include:

* Chest X-ray: A chest X-ray is typically the initial imaging modality used to assess for rib fractures, lung contusions, pneumothorax, hemothorax, or mediastinal abnormalities.
* Computed tomography (CT) scan: CT scans provide detailed imaging of the thoracic structures and are useful in identifying specific injuries, such as aortic tears, pulmonary contusions, or cardiac injuries.
* Ultrasound: Ultrasound can assist in the detection of pleural effusions, pneumothorax, or pericardial effusions, aiding in prompt diagnosis and management decisions.
* Diagnostic peritoneal lavage (DPL) or focused assessment with sonography for trauma (FAST): These techniques may be employed in cases of suspected diaphragmatic or abdominal injuries associated with thoracic trauma.

## **Management of Thoracic Trauma:**

The management of thoracic trauma depends on the specific injuries and their severity. It may involve:

* Oxygen therapy: Supplemental oxygen is provided to ensure adequate oxygenation and maintain oxygen saturation levels.
* Pain management: Analgesics are administered to alleviate pain associated with rib fractures, chest wall injuries, or other thoracic trauma.
* Chest tube placement: In cases of pneumothorax or hemothorax, a chest tube is inserted to evacuate air or blood from the pleural space and restore normal lung function.
* Surgical interventions: Severe thoracic trauma, such as cardiac injuries, major vascular injuries, or diaphragmatic injuries, may require surgical repair or intervention.
* Close monitoring: Patients with thoracic trauma often require close monitoring in an intensive care unit (ICU) setting to ensure stability and prompt management of any complications.

## **Common Pediatric Thoracic Trauma Diagnoses:**

* Pulmonary contusion:  
  Bruising of the lung parenchyma causing respiratory distress, hypoxemia, and abnormal breath sounds. Diagnosed primarily clinically and with chest X-ray showing patchy, nonanatomic opacities near the trauma site. May require oxygen or mechanical ventilation if extensive.
* Pneumothorax (including tension pneumothorax):  
  Air in the pleural space caused by trauma leading to lung collapse. Tension pneumothorax is a life-threatening emergency with respiratory distress, tracheal deviation, distended neck veins, and hypotension. Requires immediate needle decompression or chest tube.
* Hemothorax / Hemopneumothorax:  
  Accumulation of blood (and air) in pleural space causing respiratory distress and hypovolemia. Presents with decreased breath sounds and dullness to percussion. May need chest tube drainage.
* Rib fractures and flail chest:  
  Though less common in younger children due to thoracic compliance, rib fractures cause localized pain and can lead to flail segments causing paradoxical chest wall movement and respiratory failure.
* Pulmonary laceration:  
  Tear in lung tissue causing bleeding and air leak; may present with hemoptysis.
* Cardiac injury and tamponade:  
  Blunt or penetrating trauma may cause myocardial contusion or pericardial tamponade, causing hypotension, muffled heart sounds, distended veins (Beck’s triad), and shock.
* Tracheobronchial injuries:  
  Lacerations or disruption causing persistent air leak, subcutaneous emphysema, respiratory distress.
* Traumatic aortic injury:  
  Usually from high-energy deceleration; suspicion raised by widened mediastinum on X-ray, hypotension, and chest/back pain.
* Diaphragmatic rupture:  
  Presentation includes respiratory distress and abdominal organ herniation into the chest cavity; bowel sounds may be heard in chest.
* Open pneumothorax:  
  Chest wall wound allowing air ingress during respiration (“sucking chest wound”); needs surgical coverage and chest drainage.

**Pediatric Thoracic Trauma Epidemiology:**

* Thoracic injuries in children are relatively infrequent, occurring in approximately 4% to 13% of pediatric trauma patients depending on the study and setting.
* A large national Dutch cohort study (2015–2019) reported that among 66,751 hospitalized pediatric trauma patients, 1.1% sustained chest injuries, corresponding to an incidence of about 4.9 per 100,000 person-years.
* The median age of children sustaining thoracic trauma in this study was approximately 11 years, with a male predominance (about 63%).
* Most common thoracic injuries included lung contusions (about 40%) and rib fractures (about 28%), although rib fractures are less common in very young children due to chest wall compliance.
* The mortality rate in pediatric thoracic trauma ranges considerably but can be as high as 6.8% to 30%, especially with severe injuries or associated multi-system trauma.
* Age influences injury patterns and outcomes; incidence and severity tend to increase with age, with older children more likely to sustain rib fractures and severe chest injuries.
* Common mechanisms of injury include:
  + Motor vehicle accidents (cars, motorcycles)
  + Bicycle accidents
  + Falls (low and high energy)
  + Pedestrian injuries
  + Assaults and burns also contribute in smaller proportions.
* Approximately 43% of children with chest injuries in the Dutch study required intensive care admission, with a median hospital stay of 3 days.
* Pediatric thoracic trauma has one of the highest fatality rates among injury types: one US national trauma database reported a fatality rate of 7.7% among thoracic injury admissions in children.

**PREDEFINED QUESTIONS AND ANSWERS**

## What is pediatric thoracic trauma?

Pediatric thoracic trauma refers to injury to the chest structures—including lungs, heart, blood vessels, ribs, and airways—in children following blunt or penetrating trauma.

## How common is thoracic trauma in children?

Thoracic injuries occur in less than 10% of pediatric trauma admissions. Pulmonary contusions are the most common thoracic injury in children, often occurring without rib fractures due to their more flexible chest wall.

## What are the main causes of pediatric thoracic trauma?

The leading causes include motor vehicle accidents, falls, bicycle accidents, and less commonly penetrating injuries such as stab wounds or gunshots.

## What types of injuries occur in pediatric thoracic trauma?

* Pulmonary contusions (lung bruising)
* Pneumothorax (air in pleural space) and tension pneumothorax
* Hemothorax (blood in pleural space)
* Rib fractures (less common than in adults)
* Flail chest
* Cardiac contusion or tamponade
* Tracheobronchial injuries
* Diaphragmatic rupture

## How do thoracic injuries in children differ from adults?

Children have more compliant chest walls, making rib fractures rarer but allowing more force transmission to internal organs, leading to serious pulmonary contusions or internal injuries without obvious external signs.

## What are the signs and symptoms of pediatric thoracic trauma?

* Rapid breathing, respiratory distress
* Chest pain or tenderness
* Cyanosis (bluish skin)
* Decreased breath sounds or abnormal chest movement
* Subcutaneous emphysema (air under the skin)
* Circulatory instability if associated with cardiac injury

## How is pediatric thoracic trauma diagnosed?

* Clinical evaluation with airway, breathing, and circulation assessment following ATLS principles
* Chest X-ray is the primary imaging modality
* Ultrasound (eFAST exam) for detection of pneumothorax or hemothorax
* CT scans selectively used for detailed evaluation if needed

## How is pediatric thoracic trauma treated?

* Immediate airway management and oxygenation support
* Needle decompression and chest tube placement for tension pneumothorax or large hemothorax
* Pain control and supportive care for contusions and rib fractures
* Surgery in cases of cardiac tamponade, diaphragmatic rupture, or major vascular injuries

## What is the prognosis for children with thoracic trauma?

Prognosis depends on injury severity. Pulmonary contusions often improve with supportive care; however, severe injuries, especially with vascular or cardiac involvement, carry significant morbidity and mortality risk.

**DOCTOR PATIENT CONVERSATION**

Doctor: Hello, I want to talk with you about your child’s recent chest injury and explain what we’re doing to help. Do you have any questions before we start?

Parent: Thank you, doctor. I’m very worried. What exactly does a thoracic trauma mean for my child?

Doctor: Thoracic trauma means that your child has sustained an injury to the chest area, which includes the lungs, ribs, and sometimes the heart and major blood vessels. In children, this can affect breathing and circulation depending on the severity and type of injury.

Parent: What kind of injuries might happen with this kind of trauma?

Doctor: Common injuries include bruising of the lungs called pulmonary contusions, air leaking into the chest cavity known as pneumothorax, bleeding into the chest or hemothorax, fractured ribs, or very rarely injury to the heart or major vessels. Because children’s chest walls are more flexible than adults’, sometimes internal injuries can be serious even if there’s no obvious fracture.

Parent: How do you know what injuries my child has?

Doctor: We start with a thorough clinical examination, monitoring breathing and circulation carefully. Chest X-rays give us a good initial view. We also use ultrasound to look quickly for things like fluid or air around the lungs. Occasionally, a CT scan may be recommended for more detailed images.

Parent: What treatments will my child need?

Doctor: Treatment depends on the injuries found. Many lung bruises get better with oxygen and supportive care. If there is air or blood in the chest, we might place a chest tube to remove it and help your child breathe easier. In severe cases with injured blood vessels or heart, surgery may be necessary. Throughout, we provide pain relief and monitor closely.

Parent: Is this life-threatening? What’s the outlook?

Doctor: Serious chest injuries can be life-threatening, but with prompt diagnosis and treatment, most children recover well. We’re monitoring your child closely in the hospital and providing all necessary support. The key is rapid intervention to prevent complications.

Parent: Will my child need surgery?

Doctor: At this stage, we are assessing carefully. Most children with lung bruises or small pneumothoraces don’t require surgery and improve with supportive care. If a chest tube or surgery becomes necessary, we will discuss it with you immediately.

Parent: What should I watch for in case things get worse?

Doctor: Watch for signs like increased difficulty breathing, persistent chest pain, unusual paleness or blueness around lips, or decreased activity and responsiveness. Please alert any staff immediately if you notice these.

Parent: Thank you for explaining everything. It’s a relief to know how you’re managing my child.

Doctor: I understand this is very stressful. We’ll keep you informed every step of the way and answer all your questions.

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**SARCOIDOSIS**

**DEFINITION AND DESCRIPTION**

Sarcoidosis is a condition that causes tiny collections of immune system cells in any part of the body. These tiny collections form red, swollen lumps called granulomas. Granulomas most commonly occur in the lungs and the lymph nodes of the chest. They also can occur in the eyes, skin, heart and other organs.

Experts don't know the exact cause of sarcoidosis, but it's likely a mix of genetic and environmental factors. Some people appear to have gene changes that make them more likely to develop sarcoidosis. The condition may then be triggered by bacteria, viruses, dust or chemicals. Their immune system overreacts to the trigger, causing inflammation that forms granulomas.

There is no cure for sarcoidosis, but most people do not need treatment. Sarcoidosis may go away on its own. Other people need treatment to lessen their body's immune system response. Sometimes sarcoidosis can last for years and may cause organ damage.

**Causes**

The cause of sarcoidosis is not known. Experts think it results from a mix of genetic and environmental factors that cause the body's immune system to overreact to a substance it doesn't know.

Some people have gene changes that make their immune system more likely to overreact to triggers. Triggers could be bacteria, viruses, chemicals or dust. This causes immune cells to group into tiny collections of inflamed lumps called granulomas. As granulomas build up in an organ, the function of that organ can be affected.

**Risk factors**

While anyone can develop sarcoidosis, factors that may raise your risk include:

* **Age and sex.** Sarcoidosis can happen at any age, but often occurs between the ages of 20 and 60 years. Women are slightly more likely to develop the condition than are men.
* **Race.** Sarcoidosis occurs more often in people of African descent and those of Northern European descent. African Americans are more likely to have sarcoidosis in other organs along with the lungs.
* **Job or hobbies.** Working around chemicals and dust can raise your risk.
* **Family history.** If someone in your family has had sarcoidosis, you're more likely to get the condition.

**Symptoms**

Symptoms of sarcoidosis vary, depending on which organs are affected and how badly they're affected. Sarcoidosis sometimes develops slowly over time and causes symptoms that last for years. Other times, symptoms appear suddenly and then disappear just as quickly. Many people with sarcoidosis have no symptoms, so the condition is found only when a chest X-ray is done for another reason.

### **General symptoms**

Sarcoidosis can begin with these symptoms:

* Extreme tiredness.
* Slight fever.
* Swollen lymph nodes, such as in the chest, neck, armpits or groin.
* Weight loss.
* Pain and swelling in joints, such as the ankles.

### **Lung symptoms**

Sarcoidosis most often affects the lungs and may cause lung problems, such as:

* Ongoing dry cough.
* Shortness of breath.
* A squeaking sound when breathing out, called wheezing.
* Chest pain.

### **Skin symptoms**

Sarcoidosis may cause skin problems, which may include:

* A rash of small, itchy bumps, usually on the head, neck or legs. The rash may be painful.
* Open sores on the nose, lips, cheeks and ears, called skin lesions.
* Areas of skin that are darker or lighter in color.
* Growths under the skin, especially around scars or tattoos.

### **Eye symptoms**

Sarcoidosis can affect the eyes without causing any symptoms, so it's important to have your eyes checked regularly. When eye symptoms do occur, they may include:

* Blurred vision.
* Eye pain.
* Watery eyes.
* Burning, itching or dry eyes.
* Severe redness.
* Sensitivity to light.

### **Heart symptoms**

Symptoms related to cardiac sarcoidosis may include:

* Chest pain.
* Shortness of breath.
* Fainting.
* Heartbeats that aren't regular, called arrhythmias.
* Rapid or fluttering heartbeats, called palpitations.
* Swelling caused by extra fluid in the body.

### **Nervous system symptoms**

Sarcoidosis can cause symptoms related to the brain and nerves, such as:

* Headaches.
* Dizziness.
* Changes in vision.
* Confusion.
* Weakness.
* Nerve pain, numbness or tingling.
* Seizures.

### **Other symptoms**

Other symptoms are usually linked with the organs affected. Sarcoidosis can cause granulomas in any part of the body, such as the liver, spleen, muscles, bones and joints, kidneys, and lymph nodes.

### **When to see a doctor**

See a doctor or other healthcare professional if you have symptoms that may be sarcoidosis.

## **Diagnosis**

Sarcoidosis can be hard to diagnose because there may not be many symptoms in the early stages. When symptoms do occur, they may be much like those of other conditions.

Your healthcare professional does a physical exam and talks with you about your symptoms and possible risk factors. The healthcare professional also listens to your heart and lungs, checks your lymph nodes for swelling, and looks at any skin lesions.

No tests can specifically diagnose sarcoidosis. Tests can help rule out other conditions and show what body systems may be affected by sarcoidosis. For example, you may have:

* Blood and urine tests to check your overall health and how well your kidneys and liver are working.
* Chest X-ray to look at your lungs and heart.
* Computerized tomography (CT) scan of the chest to look at your lungs.
* Lung function tests, also called pulmonary function tests, to measure how much air you can breathe in and out and how much oxygen your lungs send to your blood.
* Electrocardiogram (ECG or EKG) and cardiac ultrasound to check for heart problems and look at your heart's health.
* Eye exam to check for vision problems that may be caused by sarcoidosis.
* Positron emission tomography (PET) scan or magnetic resonance imaging (MRI) if sarcoidosis may be affecting your heart or central nervous system.

Other tests may be added, if needed.

### **Biopsy**

Your healthcare professional may take a small sample of tissue called a biopsy. The sample is taken from any part of your body that may be affected by sarcoidosis. The sample is used to look for the granulomas commonly seen with the condition. For example, biopsies can be taken from your skin, lungs and lymph nodes.

**Treatment**

There's no cure for sarcoidosis, but often sarcoidosis goes away on its own. You may not need treatment if you have no symptoms or only mild symptoms. If you need treatment, the type of treatment you have depends on how severe your symptoms are and what organs are affected. Treatment can lessen the body's immune system response and inflammation.

### **Medicines**

If your symptoms are severe or affected organs aren't working properly, you may need medicines. These can include:

* **Corticosteroids.** These powerful medicines lessen inflammation. They're usually the first treatment tried for sarcoidosis. The medicine can be given as pills or shots. In some cases, corticosteroids can be put directly on the affected area. Examples of these types of medicines include cream put on skin lesions or rash, medicine breathed into the lungs using an inhaler, and drops put into the eyes.
* **Medicines that lower the immune system's response.** Medicines such as methotrexate (Trexall) and azathioprine (Azasan, Imuran) lessen inflammation by lowering the immune system's response to a trigger.
* **Hydroxychloroquine.** Hydroxychloroquine (Plaquenil) may be helpful for skin lesions and high blood-calcium levels.
* **Tumor necrosis factor-alpha (TNF-alpha) inhibitors.** These medicines are commonly used to treat inflammation from rheumatoid arthritis. They also can be helpful in treating sarcoidosis that hasn't responded to other treatments.

Other medicines may be used to treat specific symptoms or complications.

### **Other treatments**

Depending on your symptoms or complications, you may need other treatments. For example, you may have:

* **Physical therapy** to lessen tiredness and strengthen muscles.
* **Pulmonary rehabilitation** to help ease breathing and do more activities.
* **Implanted cardiac pacemaker or defibrillator** for heart rhythm problems.

An organ transplant may be an option for some people if sarcoidosis has severely damaged the lungs, heart or liver.

### **Ongoing monitoring**

How often you see your healthcare professional varies based on your symptoms and treatment. Seeing your healthcare professional regularly is important ― even if you don't need treatment.

Your healthcare professional monitors your symptoms and checks to see if you need treatment, how treatments are working and if you have complications. Ongoing monitoring may include tests based on your condition. For example, you may have regular chest X-rays, blood and urine tests, EKGs, and exams of your lungs, eyes, skin and other organs. Follow-up care is likely lifelong.

**Lifestyle and home remedies**

Along with your treatment, these self-care tips can help:

* **Follow your treatment plan.** Even if you start to feel better, don't stop taking your medicine without talking with your healthcare professional. Keep all follow-up appointments. Let your healthcare professional know if you have new or worsening symptoms.
* **Make healthy lifestyle choices.** These can include eating a healthy diet, working toward or keeping a healthy weight, managing stress, and getting enough sleep.
* **Get regular physical activity.** Regular physical activity or exercise can raise your mood, strengthen muscles and help lessen tiredness that can get in the way of your daily activities.
* **Avoid lung irritants.** As much as possible, stay away from smoke, dust, chemicals and other substances that irritate your lungs. If you smoke, talk with your healthcare professional about ways to quit.

## **Outlook / Prognosis**

The severity of sarcoidosis is different from person to person. For most people, it’s a temporary condition that goes away on its own or with treatment. For some, it’s a chronic illness that causes permanent damage.

About two-thirds of people diagnosed with sarcoidosis will eventually be disease-free (sarcoidosis will go into remission) in two to three years. Löfgren syndrome has a particularly good prognosis, with most cases resolving in six months to two years.

Most people who still have sarcoidosis after three years are likely to have chronic disease. Only about 10% to 20% of those with chronic sarcoidosis will have permanent organ damage. The most common complication of sarcoidosis in the U.S. is lung scarring.

### **What is the life expectancy of a person with sarcoidosis?**

Most people with sarcoidosis can expect to live as long as someone without sarcoidosis. A small percentage (1% to 5%) of sarcoidosis cases are fatal.

## **Prevention**

Since we don’t know for sure what causes sarcoidosis, there’s no way to prevent it or reduce your risk of getting it. Taking medications as prescribed by your healthcare provider will reduce your risk of organ damage that granulomas can cause.

## **Living With**

The best way to take care of yourself while living with sarcoidosis is to follow the recommendations of your healthcare provider. Monitor your symptoms for changes and take any medications as directed.

**Complications**

Sometimes sarcoidosis causes long-term problems.

* **Lungs.** Pulmonary sarcoidosis that isn't treated can lead to scarring in the lungs that lasts forever. This scarring is called pulmonary fibrosis. Pulmonary fibrosis makes it hard to breathe and sometimes causes pulmonary hypertension.
* **Eyes.** Inflammation can affect almost any part of the eye. It may cause damage to the retina, which over time can affect vision. Sarcoidosis can cause cataracts and glaucoma.
* **Kidneys.** Sarcoidosis can affect how your body handles calcium. Sarcoidosis can result in too much calcium in the bloodstream, a condition called hypercalcemia. This can lead to kidney stones and affect how well the kidneys work. Rarely, long-term kidney disease can occur.
* **Heart.** Cardiac sarcoidosis results in granulomas in the heart. These can cause problems with heart rhythm, blood flow and heart function. Rarely, sarcoidosis results in heart problems that may cause death.
* **Nervous system.** Some people with sarcoidosis develop problems related to the central nervous system. This occurs when granulomas form in the nerves, brain and spinal cord. For example, granulomas in the facial nerves can cause paralyzed facial muscles.

### **PREDEFINED QUESTIONS AND ANSWERS**

What are the benefits of treatment vs. monitoring symptoms?  
Many cases of sarcoidosis improve or even resolve without treatment, especially if symptoms are mild. Observation with regular follow-up may be appropriate in these cases. Treatment is usually recommended to control bothersome symptoms, prevent organ damage (especially in critical organs like heart, brain, or eyes), and improve quality of life. Your doctor will evaluate the extent of organ involvement and severity to decide if treatment is needed.

How long will it be before I can expect symptom improvement?  
Improvement timelines vary from a few weeks up to several months after starting treatment. Corticosteroids often show symptom control within weeks, but full remission may take longer. Your provider will monitor your progress closely to adjust therapy as needed.

How do I know if my treatment’s working?  
Your healthcare team will track symptoms, physical exam findings, and use tests such as chest X-rays, lung function tests, blood markers, and organ-specific assessments to monitor disease activity. Improvement or stability on these is a good sign treatment is effective. Relapses or progression may prompt changes in therapy.

Are there any lifestyle changes I should make?  
Following your treatment plan and attending regular check-ups are essential. Avoiding smoking and environmental exposures that can worsen lung health is advised. Maintaining a healthy diet, managing stress, and staying physically active as tolerated can support your overall well-being. Inform your provider about any side effects or new symptoms promptly.

When should I follow up with you?  
Follow-up timing depends on your individual case—whether you are being observed or treated, and the organs involved. Regular monitoring is important since sarcoidosis can fluctuate or affect new organs. Your doctor may schedule visits every few months initially, then space them out if stable, but lifelong follow-up is often recommended

## **Differential Diagnoses**

* Cat Scratch Disease (Cat Scratch Fever)
* Diffuse Large B-Cell Lymphoma (DLBCL)
* Eosinophilic Granuloma (Pulmonary Histiocytosis X)
* Follicular Lymphoma
* Fungal Infection
* Hilar Infiltrates
* Hypersensitivity Pneumonitis
* Leprosy
* Lymphoblastic Lymphoma
* Lymphomatoid Granulomatosis
* Mediastinal Lymphoma
* Non-Hodgkin Lymphoma (NHL)
* Non-Small Cell Lung Cancer (NSCLC)
* Noncaseating Granuloma (NCG) on Biopsy
* Primary Biliary Cholangitis (Primary Biliary Cirrhosis)
* Small Cell Lung Cancer (SCLC)
* Tuberculosis (TB)

## **Epidemiology**

### United States statistics

Incidence ranges from 5-40 cases per 100,000 population. The age-adjusted incidence for Whites is 11 cases per 100,000 population. The incidence is considerably higher in African Americans, at 34 cases per 100,000 population. The prevalence is 10 times greater for African Americans than for Whites.

Approximately 20% of patients who are African American reported an affected family member, while only 5% of Whites in the United States who have sarcoidosis said they have family members also diagnosed with sarcoidosis. In African Americans, siblings and parents of sarcoidosis cases have about a 2.5-fold increased risk for developing the disease.

Working on the World Trade Center (WTC) debris pile after the September 11, 2001, terrorist attacks was found to be associated with sarcoidosis(odds ratio, 9.1; 95% confidence interval, 1.1-74.0), but WTC dust cloud exposure was not (odds ratio, 1.0; 95% confidence interval, 0.4-2.8).

### International statistics

Incidence is 20 cases per 100,000 population in Sweden and 1.3 cases per 100,000 population in Japan. Sarcoidosis occurs in China, Africa, India, and other developing countries. Although its incidence may be low, the disease remains hidden and often is misdiagnosed as tuberculosis.

### Race-, sex-, and age-related demographics

African Americans seem to experience more severe and chronic disease.

Male-to-female ratio is approximately 1:2. Morbidity, mortality, and extrapulmonary involvement are higher in affected females.

Incidence peaks in persons aged 25-35 years. A second peak occurs for women aged 45-65 years.

## **Procedures**

Diagnosis requires biopsy in most cases. If therapy is to be given for sarcoidosis, tissue confirmation is essential. Watchful waiting is indicated only for patients who exhibit a classic presentation, are asymptomatic, and will reliably follow up.

The hallmark histologic finding is the presence of noncaseating granulomas (NCGs) with special stains negative for fungus and mycobacteria. Transbronchial biopsy via fiberoptic bronchoscopy has a high diagnostic yield. Results may be positive, even in the setting of normal chest radiography findings.

Standard transbronchial needle aspiration allows successful lymph node sampling in nearly all patients with sarcoidosis and is associated with high diagnostic yield regardless of disease stage.

Endobronchial biopsy is performed during bronchoscopy and increases the yield of the procedure. In a study of 34 subjects, endobronchial biopsy findings were positive in 61.8% of patients with a yield comparable to transbronchial biopsy, which showed non necrotizing granulomas in 58.8% of subjects. The addition of endobronchial biopsy increased the yield of fiberoptic bronchoscopy by 20.6%.

At least one study has shown that the diagnostic yield of endobronchial ultrasound-guided transbronchial needle aspiration for stage I and II sarcoidosis is even higher than standard transbronchial lung biopsy.

In at least one study, the CD4/CD8 ratio and tumor necrosis factor (TNF)–α levels in induced sputum correlated with those in bronchoalveolar lavage fluid (BALF) and paralleled changes with treatment. Induced sputum may therefore be a surrogate for BALF for certain markers in patients with sarcoidosis.

In another study, the differential cell count in BALF demonstrated a significantly lower percentage of neutrophils and a significantly higher percentage of macrophages than in induced sputum. The profiles of T-cell subsets, however, showed the same pattern in both groups. A CD4/CD8 ratio of 2.5 or greater had a sensitivity of 100% and a specificity of 81.2%, with a positive predictive value of 81.2%, distinguishing sarcoidosis from non granulomatous interstitial lung diseases.

**Sarcoidosis Treatment: Drugs and Their Side Effects**

## 1. Corticosteroids (First-Line Therapy)

* Examples: Prednisone, Dexamethasone, Triamcinolone
* Mechanism: Reduce inflammation by suppressing immune response.
* Common Dosage: Prednisone typically 5–40 mg daily; dosage individualized based on severity.
* Side Effects:
  + Osteoporosis
  + Weight gain and fluid retention ("moon face")
  + Diabetes and elevated blood sugar
  + Hypertension
  + Cataracts and glaucoma
  + Mood changes including depression and psychosis
  + Nausea, vomiting, headaches, acne
* Notes: Long-term use requires close monitoring due to significant side effects.

## 2. Immunosuppressants / Steroid-Sparing Agents

Used to reduce steroid dose or when steroids are not tolerated or insufficient.

* Methotrexate: 10-15 mg once weekly
  + Side effects: Nausea, bone marrow suppression, liver toxicity, pulmonary toxicity
* Azathioprine: 50-250 mg daily
  + Side effects: Nausea, bone marrow suppression, liver toxicity, increased malignancy risk
* Leflunomide: 10-20 mg daily
  + Side effects: Nausea, peripheral neuropathy, interstitial pneumonia
* Mycophenolate mofetil: 500-1500 mg twice daily
  + Side effects: Diarrhea, possible blood cell count changes

## 3. Antimalarial Drugs

* Hydroxychloroquine (Plaquenil®), Chloroquine
  + Used especially for cutaneous sarcoidosis and hypercalcemia
  + Side effects: Retinopathy (requires ophthalmologic monitoring), gastrointestinal upset
  + Less potent than methotrexate but safer for some patients

## 4. Biologic Agents (TNF-alpha Inhibitors)

Used for refractory cases or neurosarcoidosis.

* Infliximab: 3-5 mg/kg initially, then repeated doses every 4–6 weeks
  + Side effects: Risk of infections including tuberculosis reactivation, infusion reactions, contraindicated in severe heart failure
* Adalimumab: 40 mg every 1–2 weeks
  + Side effects: Similar infectious risks, injection site reactions
* Rituximab: 500-1000 mg every 1-6 months for select cases
  + Side effects: Increased infection risk, screened for hepatitis and tuberculosis prior to use

## 5. Other Agents (Less Common)

* Colchicine, Cyclophosphamide, Corticotropin (Acthar Gel) may be considered in specific situations or refractory disease but are less commonly used.

**DOCTOR PATIENT CONVERSATION**

Doctor: Hello! I understand you've recently been diagnosed with sarcoidosis. I’d like to talk with you about what this means and answer any questions you have. How are you feeling about the diagnosis?

Patient: Thanks, doctor. I’m a bit overwhelmed. I don't really know much about sarcoidosis. What exactly is it?

Doctor: That’s completely understandable. Sarcoidosis is a condition where small clusters of inflammatory cells, called granulomas, form in various organs—most commonly the lungs and lymph nodes. We don’t know exactly what causes it, but it’s related to an overactive immune response.

Patient: Will it get worse? Is it serious?

Doctor: The course of sarcoidosis varies quite a bit. For many people, it’s mild and even resolves on its own without treatment. However, in some cases it can affect how organs work, and treatment may be needed to control symptoms or prevent organ damage. We’ll monitor you closely to decide the best approach.

Patient: What treatments are there?

Doctor: The first-line treatment is usually corticosteroids, like prednisone, which reduce inflammation. For those who can’t take steroids or need long-term treatment, we might use other immune-suppressing medications. If your sarcoidosis is mild and not causing problems, sometimes we just monitor symptoms without medicines.

Patient: How long will I need treatment?

Doctor: That depends on how well you respond. Some people take steroids for several months, gradually tapering off, while others might need longer treatment if symptoms persist or come back. Regular follow-ups and tests help us tailor the plan.

Patient: Are there side effects I should worry about?

Doctor: Steroids can have side effects like weight gain, mood changes, high blood sugar, and bone thinning, especially if used long-term. That’s why we use the lowest effective dose and consider other medications if treatment lasts a long time. We’ll also monitor you carefully to manage any side effects early.

Patient: Is there anything I can do myself?

Doctor: Maintaining a healthy lifestyle helps—eating well, staying active as you can, avoiding smoking, and managing stress. Also, keep all your follow-up appointments, and let us know if you notice new symptoms like increased fatigue, shortness of breath, or new pain.

Patient: How often will I need check-ups?

Doctor: Initially, every few months to monitor your symptoms and organ function. Once stable, we might space them out. Sarcoidosis can change over time, so lifelong monitoring is important.

Patient: Thank you for explaining. It helps to know what to expect.

Doctor: You’re very welcome. It’s a lot to take in, but we are here to support you. Please call or come in if anything changes or if you have questions between visits.

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**ASPIRIN EXACERBATED RESPIRATORY DISEASE**

**DEFINITION AND DESCRIPTION**

Aspirin-exacerbated respiratory disease (AERD), also called Samter's triad, has three features:

* **Asthma,** although only a small number of people with asthma will develop AERD.
* **Nasal polyps that often come back,** even after taken out by surgery.
* **Problems with taking aspirin and nonsteroidal anti-inflammatory drugs (NSAIDs),** such as ibuprofen (Advil, Motrin IB, others) and naproxen. Keep in mind that aspirin or NSAIDs may be in cold medicines and other medicines.

Usually, warning signs of AERD don't show up until people have reached their 30s or 40s, but AERD can sometimes happen in children.

### **What happens when people with AERD take aspirin or NSAIDs?**

Problems usually start suddenly and can be serious. Symptoms may include trouble breathing, which could be an asthma flare-up, wheezing, coughing, sneezing, and a stuffy or runny nose. Some people with AERD also have these types of problems if they drink alcohol, such as beer or wine.

### **What causes AERD?**

The exact cause of AERD is not known but it is not an allergic response. There is no proof to show that it's genetic or inherited. The disease is not caused by taking aspirin or NSAIDs, but AERD sinus or asthma symptoms get worse when taking these medicines.

### **DIAGNOSIS**

There is no special test to find AERD. There are lab tests that can help in finding the cause of your illness. There is a blood test to look for higher than usual levels of white blood cells called eosinophils. And there is a urine test to look for raised leukotrienes, which are chemicals that can cause tightening of the airways. AERD also may affect your sense of smell. Finding AERD is possible if you have all three of these things: asthma, nasal polyps, and respiratory problems when taking aspirin or NSAIDs.

When it's not clear whether you have a problem taking aspirin or NSAIDs, your health care provider may do an aspirin challenge called desensitization. This is done to check if you have AERD. Your provider and medical team give you aspirin in a safe medical surrounding and follow special safety rules.

### **TREATMENT**

There is no cure for AERD, but several treatments can be given, depending on your illness. A blend of treatments often work best. Choices are:

* Stay away from aspirin and NSAIDs, unless your health care provider prescribes desensitization to aspirin.
* Taking medicines to treat asthma, such as corticosteroids that you breathe in.
* Surgery to remove nasal polyps, although they can come back.
* Taking medicines such as montelukast (Singulair, zafirlukast (Accolate) or zileuton (Zyflo) to block leukotrienes.
* Injecting man-made proteins into your body that connect to certain targets. These proteins, called monoclonal antibodies, try to affect cells that are causing your problem.
* Having desensitization to aspirin. Aspirin is given in the health care provider's office in slowly increasing doses over two days. After that, it is taken daily at high doses, which may help lessen the need for oral steroids. It also may stop nasal polyps from coming back.

**DIFFERENTIAL DIAGNOSIS**

* Allergic (atopic) asthma: Common asthma with typical allergic triggers; differs in that aspirin/NSAID reactions are absent.
* Nonallergic rhinitis: Nasal symptoms without aspirin sensitivity or asthma.
* Chronic rhinosinusitis with nasal polyps (CRSwNP): May occur without aspirin sensitivity or asthma; distinguishes from AERD due to lack of NSAID reactions.
* NSAID-induced urticaria/angioedema: Skin manifestations without respiratory symptoms.
* NSAID-exacerbated cutaneous disease: Similar to above, without respiratory involvement.
* Idiopathic (nonsteroidal) drug hypersensitivity: Respiratory symptoms triggered by drugs but with different mechanisms and no nasal polyps or asthma.
* Other causes of nasal polyps: Cystic fibrosis, allergic fungal rhinosinusitis, Churg-Strauss syndrome (eosinophilic granulomatosis with polyangiitis).
* Eosinophilic granulomatosis with polyangiitis (EGPA): Has asthma and eosinophilia but with vasculitis and systemic features absent in typical AERD.
* Infectious or nonallergic causes of asthma exacerbation: Viral infections, irritants.
* Other NSAID hypersensitivity syndromes: Some patients react to NSAIDs without asthma or nasal polyps.

**EPIDEMIOLOGY**

Epidemiology of Aspirin-Exacerbated Respiratory Disease (AERD):

* The estimated prevalence of AERD among adult asthmatic patients is approximately 7%, with estimates ranging from about 5.5% to 12.4% depending on study design and population.
* Among individuals with severe asthma, prevalence rises to as high as 15% (about 14.9%).
* In patients with nasal polyps and chronic rhinosinusitis, prevalence estimates are around 9–10%.
* The prevalence of AERD in the general population is much lower, estimated at about 0.3% to 2.5%.
* Studies report that 10% to 20% of all persons with asthma experience exacerbations triggered by aspirin or NSAIDs, and up to 30% in asthmatic patients with nasal polyposis may have AERD features.
* AERD typically presents in adulthood, with the average onset of symptoms (nasal polyps, asthma, aspirin sensitivity) between 30 and 34 years of age.
* There is a female predominance in AERD cases, with about 57% to 66% being women in study cohorts.
* Genetic predisposition is suggested by familial clustering in about 6% of cases.
* AERD is rare in children, though some pediatric cases exist.
* The disease is associated with increased morbidity and healthcare costs due to exacerbations and treatments, highlighting the importance of early recognition

**PREDEFINED QUESTION AND ANSWER**

## What is AERD?

Aspirin-Exacerbated Respiratory Disease (AERD), also called Samter’s Triad, is a chronic condition characterized by three key features: asthma, nasal polyps with chronic sinus inflammation, and sensitivity to aspirin and other nonsteroidal anti-inflammatory drugs (NSAIDs), which can trigger respiratory symptoms.

## What causes AERD?

The exact cause is unknown, but it involves a chronic inflammatory process in the respiratory tract. People with AERD have an abnormal reaction that causes increased leukotriene production and inflammation when exposed to aspirin or NSAIDs.

## What are the symptoms?

* Nasal congestion and sinus pressure
* Recurrent nasal polyps
* Asthma symptoms like wheezing and shortness of breath
* Respiratory reactions after taking aspirin or NSAIDs, including nasal blockage, asthma attacks, and sometimes, skin flushing or gastrointestinal symptoms

## How is AERD diagnosed?

Diagnosis is clinical, based on the triad of asthma, nasal polyps, and NSAID sensitivity. Medical history and physical exam are essential. Sometimes aspirin challenge testing in a controlled setting is used to confirm diagnosis.

## How is AERD treated?

* Avoidance of aspirin and NSAIDs unless desensitized. Acetaminophen is usually safe at low doses.
* Daily inhaled corticosteroids for asthma, nasal steroid sprays or rinses for sinus symptoms.
* Oral or injected corticosteroids may be used for severe symptoms or nasal polyps.
* Leukotriene modifiers like montelukast or zileuton can help reduce inflammation.
* Aspirin desensitization: a supervised process where aspirin doses are gradually increased to induce tolerance, allowing patients to take aspirin daily, which can reduce polyp regrowth and improve symptoms.
* Biologic therapies targeting type 2 inflammation (e.g., dupilumab, mepolizumab) may be effective for severe cases.

## Is there a cure for AERD?

There is no cure, but effective treatments can control symptoms, reduce polyp recurrence, improve asthma control, and enhance quality of life.

## What should I avoid?

Aspirin and most NSAIDs unless you have undergone aspirin desensitization. Also avoid respiratory irritants like smoke.

## Can I take other pain relievers?

Acetaminophen (Tylenol) is generally safe at low doses (up to 500 mg), but it’s best to discuss alternatives with your doctor.

## What is aspirin desensitization and who should have it?

It’s a supervised medical procedure to gradually increase aspirin tolerance. Nearly 9 out of 10 patients can subsequently take aspirin daily without reactions. It is recommended for patients with severe AERD or frequent nasal polyp regrowth.

## How often should I follow up with my doctor?

Regular monitoring is needed to assess asthma control, sinus health, and medication side effects. Your doctor will individualize follow-up based on severity.

**DOCTOR PATIENT CONVERSATION**

Doctor: Hello, I’d like to discuss your recent diagnosis of Aspirin-Exacerbated Respiratory Disease, or AERD. Do you have any questions before we start?

Patient: I’ve never heard of AERD before. What exactly is it?

Doctor: AERD, sometimes called Samter’s triad, is a chronic condition that includes three main problems: asthma, chronic sinus inflammation with recurrent nasal polyps, and sensitivity to aspirin and other NSAID medications. When someone with AERD takes aspirin or certain other pain relievers, it can trigger asthma attacks, nasal congestion, or other respiratory symptoms.

Patient: That sounds serious. What causes it?

Doctor: The exact cause isn’t fully understood, but it involves an abnormal inflammatory response in your airways. When you take aspirin or related drugs, it affects certain pathways in your body that leads to increased inflammation and respiratory symptoms. It’s not a typical allergy, and it’s not inherited.

Patient: What kind of symptoms should I expect?

Doctor: Most patients experience nasal congestion, sinus pressure, and recurring nasal polyps that can block the sinuses. The asthma symptoms can include wheezing, shortness of breath, and coughing. If you take aspirin or many NSAIDs, these symptoms can worsen rapidly.

Patient: Can I still take painkillers?

Doctor: If you haven't undergone aspirin desensitization, you should avoid aspirin and most NSAIDs like ibuprofen or naproxen, as they can trigger reactions. Acetaminophen is usually safe at low doses, but it’s best to check with your provider first.

Patient: How is AERD treated?

Doctor: Treatment involves managing your asthma and sinus symptoms with inhaled steroids, nasal sprays, and sometimes courses of oral steroids. Many patients also take medications that block inflammatory substances called leukotrienes. In addition, aspirin desensitization is a procedure where we carefully give increasing doses of aspirin under medical supervision to build your tolerance. After that, daily aspirin can improve your symptoms and reduce polyp regrowth. There are also newer biologic medicines that help some patients.

Patient: Is there a cure?

Doctor: There’s no cure, but these treatments can help you manage symptoms well and improve your quality of life. Regular follow-up is important to monitor your condition and adjust treatments as needed.

Patient: What about surgery for nasal polyps?

Doctor: Many patients do need sinus surgery to remove polyps, but unfortunately, polyps can regrow, which is why ongoing medical management including possibly aspirin therapy is key.

Patient: Are there side effects or risks with aspirin desensitization?

Doctor: Some patients experience mild gastrointestinal symptoms during desensitization or with daily aspirin, like stomach discomfort, but serious complications are uncommon when done in the right setting. We monitor you closely throughout.

Patient: What should I do if I have a reaction?

Doctor: If you take a medication and start wheezing, have severe nasal blockage, or other breathing trouble, seek medical attention immediately. Always inform healthcare providers about your AERD.

Patient: Thank you for the explanation. I feel more informed now.

Doctor: You’re welcome. Please don’t hesitate to ask questions anytime. We’ll work together to manage this condition effectively.

REFERENCES

[What is aspirin-exacerbated respiratory disease (AERD)? - Mayo Clinic](https://www.mayoclinic.org/diseases-conditions/asthma/in-depth/aerd/art-20482797)

<https://www.aaaai.org/tools-for-the-public/conditions-library/asthma/aspirin-exacerbated-respiratory-disease-(aerd>)

**COMMUNITY AQUIRED PNEUMONIA**

**DEFINITION AND DESCRIPTION**

Pneumonia is an infection in one or both lungs. Bacteria and viruses typically cause pneumonia. Fungi can also cause some types of pneumonia.

Pneumonia affects millions of people around the world each year. In the United States, about 25 of every 10,000 people get pneumonia each year. That rate gets higher as age increases.

Some people develop a serious case of pneumonia while in the hospital. But it’s more common to develop pneumonia outside a hospital setting. When this happens, doctors refer to it as community-acquired pneumonia (CAP).

CAP is the most common type of pneumonia people experience. Although it’s not usually as serious as hospital-acquired pneumonia (HAP), it can still have serious complications.

This article will look at CAP, how it compares with other types of pneumonia, and what you can do to protect yourself.

### **Types of pneumonia**

* **Community-acquired pneumonia (CAP):** CAP refers to pneumonia you develop outside a hospital setting.
* **Hospital-acquired pneumonia (HAP):** Also called nosocomial pneumonia HAP occurs if you fall ill with pneumonia 48 hours or more after your admission to the hospital. HAP is usually more serious than CAP because it can involve bacteria that are resistant to antibiotics
* **Ventilator-associated pneumonia (VAP):** A subgroup of people with HAP may develop VAP. It occurs in hospital patients who are receiving mechanical ventilation for respiratory failure.

## **What causes community-acquired pneumonia?**

Pneumonia has many causes. Research from 2017 references 26 common causes of CAP, mainly bacteria and viruses. Some fungi can also cause CAP.

The most common causes of CAP in the United States are:

* human rhinovirus (common cold)
* influenza virus (flu)
* *Streptococcus pneumoniae*

Other common causes include:

* the coronavirus SARS-CoV-2 (COVID-19)
* *Haemophilus influenzae*
* *Legionella* bacteria
* *Mycoplasma pneumonia*
* hard-to-detect bacteria, such as *Chlamydia pneumoniae*

So, how do humans come in contact with these germs?

Some of them, like *Streptococcus pneumoniae*, already live in your nose, sinuses, or mouth. They can eventually spread to your lungs, where they cause disease. It’s unclear why they invade the lungs.

You might also breathe in these germs if they are in the air around you. Certain activities or environments may increase your risk of coming in contact with them.

## **Who is at risk of community-acquired pneumonia?**

CAP and its complications are most likely to affect:

* adults older than age 65
* people with existing health conditions
* people with a weakened immune system
* people who smoke
* people who are malnourished

Children younger than 5 years also have a higher risk of CAP, as well as those who may interact with them in day care settings.

### **Health conditions**

Certain conditions may increase your risk of developing CAP. According to a 2017 review of 29 studies, these include:

* chronic bronchitis or COPD
* asthma
* diseases of the mouth, teeth, or gums
* conditions that cause impaired function

While the review was inconclusive on whether heart disease was a risk factor, other research from 2015 suggests it is.

Other conditions, such as diabetes and liver disease, may affect your outlook with CAP.

Researchers in the 2017 review also identified certain medications that may increase your risk:

* immunosuppressants
* oral steroids
* proton pump inhibitors and H2 blockers (both used to treat gastric reflux)

### **Environmental exposure**

According to the World Health Organization environmental factors may also affect your risk of CAP. These include:

* indoor air pollution from biomass fuels
* crowded homes
* secondhand smoke
* living on or near a farm with livestock
* living near centers of industrial food animal production

According to 2015 research, people who smoke cigarettes also have a higher risk of CAP.

### **Zoonotic (animal) exposure**

Exposure to certain animals that may carry the germs that cause CAP could put you at risk. According to research from 2016 and 2017, examples include:

* birds, including poultry
* rabbits
* goats
* sheep
* pigs
* cattle
* sick dogs
* cats in labor

### **Travel exposure**

The germs that cause CAP exist everywhere in the world. But some regions are more likely to have certain germs.

Experts know that travel to large gatherings abroad contributes to the spread of germs that cause CAP. Traveling, spending time on a cruise ship, or staying in a hotel can increase your risk of exposure to these germs.

If you have symptoms of pneumonia, be sure to tell a doctor of any recent travel. It may help them make a diagnosis.

## **symptoms of community-acquired pneumonia**

Pneumonia is a serious, sometimes fatal disease. The main symptoms of CAPT include:

* cough
* production of sputum
* fever
* difficulty breathing
* headache
* chest pain
* muscle aches
* irritability and restlessness (in infants)
* confusion (in older adults)

In some people, breathing issues and fluid buildup in the lungs may become severe. This may lead to respiratory failure and the need for oxygen therapy or mechanical ventilation.

## **Diagnosis community-acquired pneumonia**

If a doctor notices you have symptoms of CAP, they will likely perform or order the following:

* **Medical history:** A doctor will ask about your medical history to better understand how you may have become ill.
* **Physical exam:** A doctor will perform a physical exam to check for fever and lung sounds consistent with pneumonia.
* **Chest X-ray:** A chest X-ray can show any fluid buildup or inflammation in the lungs.
* **CT scan:** A doctor might use a CT scan to confirm a diagnosis or rule out other conditions. CT scans may be more effective at detecting pneumonia than chest X-rays, but they are more expensive and take longer.
* **Complete blood count (CBC):** A CBC can help doctors see whether your immune system is fighting the infection.
* **Electrolyte panel:** An electrolyte panel can check your electrolyte levels as well as your kidney and liver function.
* **Blood, sputum, or urine tests:** Blood and sputum cultures, as well as urine antigen tests, may identify the exact cause of the infection, influencing treatment.
* **Molecular testing:** This is the standard way to test for viral pneumonia, such as cases caused by COVID-19. Research from 2020 has also shown that molecular tests, like polymerase chain reaction (PCR) tests, may also be better than cultures at detecting bacterial pneumonia.

A doctor will also consider other potential diagnoses. Some illnesses with symptoms similar to CAP include:

* acute bronchitis or other respiratory infection
* chronic obstructive pulmonary disease (COPD)
* heart failure or heart attack
* pulmonary embolism (blood clot in your lung)
* tuberculosis

## **Treatment for community-acquired pneumonia**

If you have CAP, your treatment plan will depend on your symptoms and the specific cause of your infection. A doctor will also consider how severe your symptoms are.

According to 2017 research, 80% of people in the United States can treat CAP at home. If your doctor determines you have a milder case of CAP from bacteria, they may prescribe antibiotics for you to take at home. A doctor will likely prescribe one of the following:

* amoxicillin
* azithromycin
* clarithromycin
* doxycycline

It’s important to take antibiotics as prescribed. If you don’t follow the proper regimen, the bacteria could become antibiotic resistant.

If you have a more severe case of CAP, you may need to get treatment in a hospital. Healthcare professionals can provide a variety of intravenous (IV) antibiotic treatments based on your specific situation at a hospital.

But antibiotics do not work for cases of viral pneumonia. You may need to let the virus run its course. In some cases, you may be able to use antiviral medication. Examples include:

* remdesivir (Veklury) for COVID-19
* oseltamivir (Tamiflu) for influenza
* inhaled ribavirin for respiratory syncytial virus (RSV)

There are other actions you can take to manage symptoms of viral CAP:

* Take over-the-counter pain relievers, like acetaminophen or ibuprofen, to reduce fever.
* Drink plenty of fluids to stay hydrated.
* Get enough rest to allow your body to recover.
* Quit smoking if you regularly smoke.

If your symptoms are severe, you may need to go to the hospital. Additional treatments in the hospital may include:

* oxygen therapy
* intravenous (IV) fluids
* respiratory support with a ventilator

## **Potential complications of community-acquired pneumonia**

With CAP comes the risk of possible complications. These are more likely if doctors don’t make a timely diagnosis or if initial treatments don’t work.

Possible complications include:

* **Pleural effusion:** Pleural effusion is the buildup of fluid in your pleura, the space between your lungs and chest wall.
* **Empyema:** Empyema is the buildup of pus in the pleural space.
* **Lung abscesses:** An abscess is a pus-filled cavity that, in this case, forms in your lung.
* **Sepsis:** Sepsis is an extreme immune response to an infection.
* **Acute respiratory distress syndrome (ARDS):** ARDS occurs when the lungs become severely inflamed and fill with fluid, preventing oxygen from getting into the blood.

Complications like sepsis and ARDS can cause organ failure and death.

## **Outlook for people with community-acquired pneumonia**

With rapid and appropriate treatment, many people fully recover from CAP without complications. Young people tend to recover fully more quickly.

If CAP is due to bacteria, you may start to feel better within 5 to 7 days of starting antibiotics. Still, it can take months for all your symptoms to resolve.

If you have a mild case of CAP, full recovery is possible with rest, antibiotics or antivirals, and sleep.

More severe cases of CAP may require lengthy hospital stays to help keep you stable and aid recovery. For people admitted to intensive care, the death rate can be as high as 23%, according to 2021 research.

## **Prevent community-acquired pneumonia**

Taking precautions against illness, such as getting an annual flu shot, can reduce your risk of CAP. The Centers for Disease Control and Prevention (CDC) recommends that people ages 65 and older get a high dose flu vaccine.

People over 65 can also consider getting the pneumococcal vaccine to prevent CAP. This is especially important if you have other health conditions or if you smoke.

You may need a booster shot if you received your vaccine before age 65, or if you have a weakened immune system.

Other everyday actions you can do to help prevent disease include the following:

* Wash your hands frequently, especially after using the bathroom or being in public.
* Cough into your elbow when you need to cough.
* Keep a physical distance from people who are ill.
* If you smoke or vape, consider quitting to reduce damage to your lungs. Such damage can make you more vulnerable to pneumonia infection.

## **Epidemiology of Community-Acquired Pneumonia**

The incidence of CAP varies greatly from country to country. In the United States, CAP is estimated to occur in 649 of 100,000 adults annually, whereas a study from the Veterans Health Administration noted 472.2 cases per 100,000 person-years in 2017. Other countries report different frequencies, such as 8.1 per 10,000 in Vietnam and 31.2 per 10,000 in the United Kingdom. Rates also vary based on the population studied. In adults older than 65 years, 130.5 per 10,000 develop CAP in Malaysia, whereas 76.5 per 10,000 develop CAP in Germany. In individuals aged 85 years or older in Norway, the incidence of CAP is 172.4 per 10,000.

Influenza and pneumonia combined remain in the top 10 leading causes of death in the United States, ranking 9 of 10 in 2020. The combination was responsible for 1.6% (53,495) of deaths in 2020, an absolute increase of 7.5% from 2019 but a relative decrease due to the separately accounted deaths due to COVID-19, which ranked 3 of 10 at 10.3% (345,323) of all deaths.In addition, survivors of CAP have an increased incidence of cardiovascular events and mortality risk for months to years after CAP. As many as 20% of CAP survivors have been shown to have cardiovascular events, and mortality is up to 30% 2 to 5 years after CAP.

The cost of CAP has been examined in numerous studies. In a 2018 retrospective analysis of claims data for adults aged 65 years or older in a Medicare insurance plan, the rate of CAP was 846.7 per 100,000 person-years, which was greater than rates for myocardial infarction (405), stroke (278.9), and osteoporotic fractures (343.9). This study noted vaccinations against influenza and *Pneumococcus* infection cost $40 million; however, prevention for stroke and myocardial infarction cost more than $661 million.

### Age

Advanced age is associated not only with a higher incidence of CAP but also with more severe disease, greater need for hospitalization, and higher mortality. A recent US study estimated 967,470 adults aged 65 and over are hospitalized annually from CAP with a 38% one-year mortality.

CAP encountered in the ambulatory setting is more common among young adults, and is usually due to so-called atypical CAP pathogens (eg, *Mycoplasma pneumoniae*).

## **Differential Diagnoses of Community-Acquired Pneumonia**

Aside from those mentioned above, the differential diagnoses to consider in the diagnosis of community-acquired pneumonia (CAP) include the following:

* Acute bronchitis
* Acute exacerbation of chronic bronchitis
* Aspiration pneumonitis
* Tracheobronchitis
* Myocardial infarction
* CHF and pulmonary edema
* Pulmonary fibrosis
* Sarcoidosis
* SLE pneumonitis
* Drug hypersensitivity reactions (nitrofurantoin, daptomycin)
* Drug-induced pulmonary disease (bleomycin, checkpoint inhibitors)
* Cryptogenic organizing pneumonia
* Pulmonary embolus or infarction
* Bronchogenic carcinomas
* Radiation pneumonitis
* Granulomatosis with polyangiitis (Wegener granulomatosis)
* Lymphoma

## **Vaccination**

Pneumococcal vaccines have been shown to have some efficacy in preventing vaccine-strain pneumococcal pneumonia and invasive disease. They have not been shown to prevent community-acquired pneumonia (CAP) of all kinds.Annual influenza vaccination has been shown to decrease pneumonia diagnoses, hospitalizations, and cardiac events in certain populations.

Annual influenza vaccination is recommended in all persons older than 6 months. There are several options for vaccination, including standard-dose trivalent inactivated vaccine, high-dose inactivated trivalent vaccine, three different formulations of quadrivalent inactivated vaccine (egg-based, cell culture–based, and recombinant-derived), and a live attenuated influenza vaccine. Per current CDC recommendations, people with a history of egg allergy of any severity may receive any licensed, recommended, and age-appropriate influenza vaccine.Individuals with a history of severe egg allergy should receive the vaccine under the supervision of a physician experienced in the management of allergic conditions. People with a personal history of severe allergy to the flu vaccine should not receive it, and persons with history of Guillain-Barré syndrome should consult with their physician prior to being vaccinated.

Four pneumococcal vaccines are approved in the United States:

* 15-valent conjugate vaccine (PCV15; Vaxneuvance) is approved for adults 18 years and older.
* 20-valent conjugate vaccine (PCV20; Prevnar 20) is approved for adults 18 years and older.
* 21-valent conjugate vaccine (PCV21; Capvaxive)
* 23-valent pneumococcal polysaccharide vaccine (PPSV23; Pneumovax 23) is approved for adults aged 65 years or older and persons aged 2 years or older who are at an increased risk for pneumococcal disease.

In October 2024, the Advisory Committee on Immunization Practices (ACIP) approved the Recommended Immunization Schedule for Adults Ages 19 Years or Older, United States, 2025.PCV21 now is listed as a vaccination option across all relevant sections of the notes. PCV21 includes 8 pneumococcal serotypes that are not present in PCV15, PCV20, or PPSV23. However, it lacks some serotypes, such as serotype 4, that are found in other pneumococcal vaccines.

For routine vaccination, universal vaccination now is recommended for adults aged ≥50 years. For “special situations,” a risk-based vaccination recommendation is provided for adults aged 19–49 years. New details have been added about the use of pneumococcal vaccines during pregnancy and guidance for situations where PPSV23 is not available.

The administration of pneumococcal vaccines is tailored based on an individual's age, vaccination history, and specific health conditions.

The Advisory Committee on Immunization Practices (ACIP) recommends the following:

Routine vaccination

* Adults aged 50 years and older should receive a pneumococcal conjugate vaccine—either PCV15, PCV20, or PCV21. This recommendation applies to individuals who have not previously received any pneumococcal conjugate vaccine or whose vaccination history is unknown.
  + For those administered PCV15, an additional dose of PPSV23 is required 1 year later to complete the pneumococcal vaccination series. This subsequent dose may be considered at a minimum interval of 8 weeks in specific clinical scenarios, such as patients with an immunocompromising condition, those with a cochlear implant, or individuals with a cerebrospinal fluid leak.
  + If PCV20 or PCV21 is administered, no further vaccination with PPSV23 is indicated.
* For adults aged 65 years or older, shared clinical decision-making is recommended regarding pneumococcal vaccination. These patients may choose to receive PCV20 or PCV21, or opt out of additional pneumococcal vaccines if they have previously received PCV13 (but not PCV15, PCV20, or PCV21) at any age and PPSV23 at or after the age of 65.

Adults under age 50 with a risk condition

* The CDC defines a risk condition as presence of cerebrospinal fluid (CSF) leak, chronic liver disease, cochlear implant, an immunocompromising condition (congenital or acquired asplenia/splenic dysfunction, immunodeficiency, conditions associated with immunosuppressive drugs or radiation therapy, HIV infection, hemoglobinopathy), or diabetes mellitus.
* Individuals who have never received a pneumococcal vaccine are administered 1 dose of either PCV15, PCV20, or PCV21. If PCV15 is selected, it should be followed by a dose of PPSV23 at least 1 year later, although this interval can be shortened to 8 weeks for adults with an immunocompromising condition, a cochlear implant, or a cerebrospinal fluid leak. Once this regimen is completed, the pneumococcal vaccination series is considered complete.
* For those who receive PCV20 or PCV21, no subsequent dose of PPSV23 is required, and the vaccination series is complete upon administration of either vaccine.
* Individuals who have previously received only PPSV23 should be given a dose of PCV15, PCV20, or PCV21 at least 1 year after their most recent PPSV23 vaccination. Following this, no additional dose of PPSV23 is recommended regardless of the vaccine used, completing their pneumococcal vaccination series.
* Those who have only received PCV13 should receive a dose of PCV20 or PCV21 at least 1 year after their most recent PCV13 vaccination, which completes their pneumococcal vaccination series.
* For patients who have received both PCV13 and 1 dose of PPSV23, the subsequent recommendations are contingent upon the individual's risk condition.
  + Patients with high-risk conditions such as an immunocompromising condition, a cerebrospinal fluid leak, or a cochlear implant should receive a dose of PCV20 or PCV21 at least 5 years after their most recent pneumococcal vaccination. This completes their pneumococcal vaccination series.
  + For those with other risk conditions, no additional pneumococcal vaccines are recommended until the individual reaches at least age 50 years, at which time it is advisable to reassess the pneumococcal vaccine recommendations to ensure continued protection against pneumococcal disease.

**Pediatric Community-Acquired Pneumonia (CAP) Treatment: Drugs and Side Effects**

## 1. Amoxicillin (First-line antibiotic)

* Dosage:
  + 80–90 mg/kg/day divided twice daily for 5 days (WHO recommendation)
  + Typical max dose ~4,000 mg/day
* Use: Effective against *Streptococcus pneumoniae*, the most common bacterial cause in children.
* Side Effects:
  + Diarrhea
  + Rash (including mild hypersensitivity)
  + Rarely allergic reactions (anaphylaxis very rare)
  + Nausea or vomiting

## 2. Amoxicillin-Clavulanate

* Use: For children who received amoxicillin in past 30 days or suspected beta-lactamase producing organisms.
* Side Effects: Similar to amoxicillin but with higher risk of gastrointestinal upset (diarrhea, nausea).

## 3. Penicillin or Ampicillin (Parenteral)

* Use: Hospitalized children with severe pneumonia requiring IV antibiotics; often paired with gentamicin.
* Side Effects:
  + Allergic reactions
  + Injection site pain
  + Gastrointestinal symptoms

## 4. Gentamicin (Parenteral)

* Use: Combined with penicillin for severe pneumonia.
* Side Effects:
  + Nephrotoxicity (kidney damage)
  + Ototoxicity (hearing loss) especially with prolonged use or high doses
  + Rare allergic reactions

## 5. Ceftriaxone (Second-line parenteral antibiotic)

* Use: For severe pneumonia not responding to first-line IV antibiotics.
* Side Effects:
  + Diarrhea
  + Allergic reactions
  + Risk of biliary sludge (rare)

## 6. Macrolides (Azithromycin or Clarithromycin)

* Use: Target atypical pathogens like *Mycoplasma pneumoniae*, especially in school-age children and adolescents.
* Dosage: Azithromycin 10 mg/kg on day 1, then 5 mg/kg daily for 4 days (max 500 mg day 1, 250 mg days 2-5).
* Side Effects:
  + Gastrointestinal upset (nausea, vomiting, diarrhea)
  + Risk of QT prolongation (heart rhythm irregularities)
  + Potential drug interactions

## 7. Supportive Treatments

* Oxygen therapy for hypoxia.
* Antipyretics (acetaminophen or ibuprofen) for fever.
* Fluids to prevent dehydration.

## Important Considerations:

* Duration of therapy: Typically 5 days for otherwise healthy children responding well to treatment; can be longer (7–10 days) in complicated or severe cases.
* Antibiotic choice depends on:
  + Age
  + Immunization status
  + Severity of illness
  + Local resistance patterns
  + Recent antibiotic exposure
* MRSA coverage is reserved for complicated pneumonia or if risk factors present (e.g., clindamycin, vancomycin).

**PREDEFINED QUESTION AND ANSWER**

## What is pediatric community-acquired pneumonia (CAP)?

It is a lung infection acquired outside of healthcare settings, caused by bacteria, viruses, or fungi, affecting the lungs of children.

## What causes CAP in children?

Viruses are the most common cause in infants and young children (e.g., respiratory syncytial virus, influenza). In older children, bacteria like *Streptococcus pneumoniae*, *Mycoplasma pneumoniae*, and *Chlamydia pneumoniae* become more frequent causes.

## What are the typical symptoms of CAP in children?

Symptoms include cough, fever, rapid breathing (tachypnea), difficulty breathing, chest retractions, grunting, lethargy, and decreased appetite. Infants may present with nonspecific signs like poor feeding and irritability.

## How is CAP diagnosed?

Diagnosis is usually clinical, based on symptoms and physical exam findings such as crackles, decreased breath sounds, and retractions. Chest X-rays are mainly used for hospitalized or complicated cases. Tachypnea is a sensitive sign; WHO defines it differently by age group, e.g., >50 breaths/min in infants 2-12 months.

## How is CAP treated?

Most children with mild to moderate CAP are treated outpatient with oral antibiotics; high-dose amoxicillin is the first-line choice for bacterial pneumonia. Supportive care includes fever control, hydration, and monitoring. Hospitalized children may require oxygen and intravenous antibiotics.

## When should a child be hospitalized?

Hospitalization is considered for infants less than 3-6 months old, those with severe respiratory distress, hypoxia, dehydration, inability to take oral medication, or if social circumstances interfere with proper care.

## Can CAP be viral, and do children always need antibiotics?

Many cases, especially in young children, are viral and do not benefit from antibiotics. However, in practice, antibiotics are often started empirically due to diagnostic uncertainty and the risk of bacterial infection.

## What are possible complications of CAP?

Complications include pleural effusion, empyema (pus in the pleural space), abscess formation, respiratory failure, and sepsis, which are more common in severe or untreated cases.

## How soon should symptoms improve with treatment?

Improvement is typically expected within 48 to 72 hours of starting appropriate antibiotics. If symptoms worsen or persist, re-evaluation is necessary.

## How can CAP be prevented in children?

Vaccination (e.g., pneumococcal conjugate vaccine, Haemophilus influenzae type B vaccine, influenza vaccine), good hygiene, breastfeeding, and avoiding tobacco smoke exposure reduce risk.

**DOCTOR PATIENT CONVERSATION**

Doctor: Hello, I want to talk about your child’s diagnosis of pneumonia. Do you have any questions so far?

Parent: Thanks, doctor. I’m worried. What exactly is pneumonia?

Doctor: Pneumonia is an infection of the lungs that causes inflammation and fluid buildup, making it harder for your child to breathe. It’s quite common in children and can be caused by viruses or bacteria.

Parent: What caused my child’s pneumonia?

Doctor: In young children, viruses often cause pneumonia, especially during cold or flu season. In older children, bacteria like *Streptococcus pneumoniae* or *Mycoplasma pneumoniae* can also cause pneumonia. We usually treat based on the likely cause.

Parent: What symptoms should I watch for?

Doctor: Common symptoms include cough, fever, fast breathing or difficulty breathing, chest pain, and sometimes less energy or poor appetite. In infants, signs can be subtle, like poor feeding or irritability.

Parent: How is pneumonia treated?

Doctor: Mild cases, especially viral ones, often improve with rest, fluids, and fever control at home. If bacteria are suspected, antibiotics like amoxicillin are used. We monitor closely and may adjust treatment if symptoms don’t improve.

Parent: When would my child need to be hospitalized?

Doctor: Hospital care is needed if your child is very young (under 3–6 months), has trouble breathing, low oxygen levels, dehydration, or other medical problems. Otherwise, many children get better comfortably at home.

Parent: How long will recovery take?

Doctor: Fever usually improves within a few days, but cough and tiredness may last 1 to 2 weeks or longer. It can take time for your child to fully recover energy and appetite.

Parent: What can I do to help at home?

Doctor: Make sure your child gets plenty of fluids and rest. You can use a cool-mist humidifier or gently tap their chest to help loosen mucus. Avoid smoke exposure and keep up with hand hygiene to prevent infections.

Parent: Should my child get vaccines?

Doctor: Yes, vaccines against pneumonia-causing bacteria and flu are very important to prevent future infections. We’ll check that your child’s vaccinations are up to date.

Parent: When should I call you or seek emergency care?

Doctor: If your child’s breathing worsens, they become very sleepy or irritable, have bluish lips or face, or can’t drink fluids, seek emergency care immediately. Also call if fever persists more than a few days despite treatment.

Parent: Thank you, doctor. That helps me understand what to expect.

Doctor: You’re welcome. We’re here to support you and your child through recovery. Please reach out with any questions.

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**BRONCHIAL ATRESIA**

**DEFINITION AND DESCRIPTION**

Bronchial atresia is a rare congenital condition that affects the bronchi in the lungs. It is characterized by the abnormal closure or narrowing of one or more bronchi, which can cause respiratory symptoms. In this article, we will discuss the causes, symptoms, and treatment of bronchial atresia.

## **Causes:**

The exact cause of bronchial atresia is unknown. However, it is believed to be a congenital condition, meaning it occurs during fetal development. The abnormal closure or narrowing of the bronchi is thought to be caused by a failure of the bronchi to develop properly.

## **Symptoms:**

Symptoms of bronchial atresia may vary depending on the severity of the condition. Some common symptoms may include:

* Shortness of breath
* Wheezing
* Recurrent respiratory infections
* Coughing
* Chest pain

In some cases, bronchial atresia may be asymptomatic, and the condition may only be discovered incidentally on imaging studies.

## **Treatment:**

Treatment for bronchial atresia depends on the severity of the condition and the symptoms present. In many cases, observation is sufficient, especially if the condition is asymptomatic.

If symptoms are present, treatment may involve:

* Bronchodilator therapy: Medications that relax the muscles in the airways and improve breathing.
* Surgery: In severe cases, surgery may be necessary to remove the affected bronchi or to create an opening in the affected bronchi to improve airflow.
* Pulmonary rehabilitation: A program of exercise

**DIFFERENTIAL DIAGNOSIS**

* Congenital bronchial atresia (CBA): Characterized by a proximal bronchial interruption with mucus impaction (mucocele) distal to the atretic segment and hyperinflation of the affected lung parenchyma.
* Congenital lobar emphysema (Congenital lobar overinflation): A congenital overexpansion of a pulmonary lobe causing hyperlucency but without mucus plugging or a blind-ending bronchus.
* Bronchogenic cyst: A congenital cystic lesion of the bronchial tree that may cause localized mass effects without typical mucocele features.
* Pulmonary sequestration: Nonfunctioning lung tissue with anomalous systemic arterial supply, often intralobar or extralobar, which may mimic mucoid impaction and hyperinflation.
* Congenital cystic adenomatoid malformation (CCAM / Congenital pulmonary airway malformation): A cystic lung lesion often detected prenatally or in infancy, distinct from bronchial atresia but sometimes coexisting.
* Mucoid impaction due to allergic bronchopulmonary aspergillosis (ABPA) or cystic fibrosis: Both conditions lead to mucus plugging but usually accompanied by bronchiectasis and systemic symptoms.
* Acquired bronchial obstruction: Caused by foreign bodies, inflammatory strictures, tumors, or vascular anomalies compressing bronchi, which can mimic the appearance of atresia.
* Pulmonary embolism/infarction: Rare in children but may cause localized opacities and mimic segmental lung abnormalities.
* Lobar pulmonary hyperinflation secondary to airway obstruction (e.g., polyp, mucus plug): Collateral ventilation causes overinflation of lung segments like in bronchial atresia but usually lacks a blind-ending bronchus.

**EPIDEMIOLOGY**

Pediatric Bronchial Atresia Epidemiology:

Congenital bronchial atresia (CBA) is a rare congenital abnormality of the tracheobronchial tree. Its estimated prevalence is 1 in 100,000 births.

Key epidemiological features include:

* Rarity: CBA is considered uncommon. Studies often involve small numbers of patients, such as a review of 12 cases over 28 years in one hospital. In a study of congenital lung malformations in children, bronchial atresia accounted for 6.7% of cases, representing 2 out of 30 patients.
* Age at Diagnosis: While a congenital condition, CBA is infrequently diagnosed during childhood. Many cases, approximately two-thirds, are asymptomatic before diagnosis and are discovered incidentally on radiological imaging. When symptomatic, children may present with recurrent pneumonia or respiratory distress. A study showed children with CBA were aged 9, 10, and 11 years, while adults ranged from 25-74 years with a mean age of 37 years.
* Sex Predominance: There is a male predominance, with an estimated prevalence of 1.2 cases per 100,000 males and 0.6 cases per 100,000 females, leading to a male:female ratio of 2:1.
* Etiology: The exact cause is unknown, but it is hypothesized to be a focal bronchial interruption occurring before birth, possibly due to intrauterine ischemia after the 16th week of gestation.
* Prognosis and Recurrence: The condition has a high rate of perinatal mortality when detected prenatally. However, there is no increased risk of recurrence in subsequent pregnancies.

**PREDEFINED QUESTION AND ANSWERS**

## What is bronchial atresia in children?

Bronchial atresia is a rare congenital condition where a segment of a bronchus (airway) is absent or blocked, causing mucus to accumulate distally (forming a mucocele or bronchocele) and the affected lung segment to become overinflated (hyperlucent) due to trapped air.

## What causes bronchial atresia?

The exact cause is unknown. It is thought to result from a focal interruption in bronchial development during fetal life, possibly due to ischemic injury or a developmental defect occurring between weeks 4 and 16 of gestation.

## How common is bronchial atresia in children?

It is very rare, with an estimated occurrence of about 1 in 100,000 births. Many children are asymptomatic and the condition may be discovered incidentally on imaging done for another reason.

## What symptoms do children with bronchial atresia have?

Most children are asymptomatic, especially in early life. Some may have recurrent respiratory infections, cough, wheezing, or respiratory distress if complications occur.

## How is bronchial atresia diagnosed?

Diagnosis typically involves chest X-rays and high-resolution CT scans, which show a characteristic triad of findings: a blind-ending bronchus with a mucus-filled dilated segment (mucocele), surrounding hyperinflated lung tissue, and absence of normal bronchial continuity. Bronchoscopy may help confirm absence or atresia of the bronchus.

## How is bronchial atresia treated?

If asymptomatic, no treatment may be needed and observation is appropriate. Surgery, such as segmentectomy or lobectomy, is considered if there are recurrent infections, significant symptoms, or complications like lung destruction.

## What is the prognosis for children with bronchial atresia?

Prognosis is generally good for asymptomatic cases with appropriate monitoring. Surgical outcomes are typically favorable when intervention is needed.

## Can bronchial atresia be detected before birth?

Sometimes it is identified prenatally on ultrasound or fetal MRI as a cystic lung lesion or hyperinflation, but many cases are diagnosed postnatally during evaluation for respiratory symptoms or incidentally.

## Are there other lung conditions similar to bronchial atresia?

Yes, conditions such as congenital lobar emphysema, bronchogenic cysts, pulmonary sequestration, and congenital pulmonary airway malformation can present with overlapping imaging features and need differentiation.

**DOCTOR PATIENT CONVERSATION**

Doctor: Hello, I’d like to discuss the findings from your child’s recent chest imaging. We’ve identified a condition called bronchial atresia. Have you heard of this before?

Parent: No, I haven’t. What is bronchial atresia?

Doctor: Bronchial atresia is a rare congenital condition where part of one of the airways in the lung is blocked or ends abruptly. This causes mucus to build up in that segment, and the affected area of the lung beyond the blockage becomes overinflated because air can still get in through small collateral channels, but it can’t exit normally.

Parent: That sounds serious. What symptoms does it cause?

Doctor: Many children with bronchial atresia don’t have symptoms and the condition may be found incidentally on imaging done for other reasons. However, some children may have recurrent respiratory infections, cough, or difficulty breathing. Symptoms vary widely.

Parent: What causes this to happen?

Doctor: We don’t know the exact cause, but it likely occurs early during lung development before birth. Sometimes a part of the airway doesn’t form correctly or becomes blocked, which leads to the changes we see.

Parent: How do you confirm the diagnosis?

Doctor: We use chest CT scans to see the characteristic findings—the mucus-filled blind-ending airway and overinflated lung segment. Sometimes a bronchoscopy (looking into the airways with a camera) is done to further evaluate.

Parent: What is the treatment?

Doctor: If your child isn’t having significant symptoms or infections, we often manage it conservatively with close monitoring through periodic imaging and check-ups. If there are frequent infections, worsening breathing problems, or damaged lung tissue, surgery to remove the affected segment may be necessary.

Parent: What is the outlook for my child?

Doctor: The prognosis is generally good, especially if symptoms are mild or absent. Children who require surgery usually do well afterward. We will monitor your child regularly and adjust the treatment plan as needed.

Parent: Should I watch out for anything specific?

Doctor: Yes. If your child develops increased cough, fever, difficulty breathing, or other signs of infection, please seek medical attention promptly. Also, keep regular follow-up appointments to monitor lung function.

Parent: Is this something that will improve over time or get worse?

Doctor: It varies. Some children remain stable without symptoms for years. Others may experience complications that require intervention. Our goal is to support your child’s breathing and health to minimize problems.

Parent: Thank you for explaining. I feel more confident knowing what to expect.

Doctor: You’re welcome. Pl

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**IDIOPATHIC PULMONARY HEMOSIDEROSIS**

**DEFINITION AND DESCRIPTION**

Idiopathic pulmonary hemosiderosis (IPH) is a rare lung disease, mostly affecting children under 10 years old. “Idiopathic” means the cause is not yet known, “pulmonary” refers to involvement of the lungs, and “hemosiderosis” refers to the deposition of ‘hemosiderin’, a storage form of Iron in the human body. Patients with the disease experience bleeding into the lungs, resulting in recurrent episodes of shortness of breath and coughing, often with blood. The ongoing bleeding often leads to iron deficiency anemia, which causes fatigue and lack of growth in children. The disease injures the lungs, leading to inflammation and, in some people, irreversible scarring. Doctors use a flexible scope to access the lungs (bronchoscopy) and perform washing (bronchoalveolar lavage) to make the diagnosis. Sometimes, a lung biopsy could be necessary for a definite diagnosis. There is no known cure for IPH, and the prognosis is highly variable. The survival rate has improved over the past few decades as research and therapies improve. In the past, the average survival had ranged from 2.5 to 5 years after diagnosis. Standard therapies include steroids and immunosuppressants to help control the bleeding into the lungs, blood transfusions to correct the anemia and other supportive measures.

### **Synonyms**

* IPH
* immune-mediated pulmonary hemosiderosis (ImPH)
* diffuse alveolar hemorrhage (DAH)
* diffuse pulmonary hemorrhage (DPH)

### **Signs & Symptoms**

Some patients may not show any symptoms early in the disease, but the most common symptoms include:

– Coughing with blood (hemoptysis)  
– Shortness of breath (dyspnea)  
– Long, unrelenting cough  
– Anemia (due to bleeding in the lungs and Iron deficiency)  
– Fatigue  
– Fever  
– Chest pain  
– Failure to thrive (deficient growth)  
– Enlarged liver and spleen (hepatosplenomegaly)

### **Causes**

The cause of IPH is still not known but it is considered to be an autoimmune disease. Many patients also have celiac disease, another autoimmune disease, and the combination of celiac disease and IPH is known as Lane-Hamilton syndrome. In most patients with Lane-Hamilton syndrome, eliminating gluten from the diet also improved the symptoms of IPH. The autoimmune hypothesis theorizes that the cells responsible for providing immunity are somehow responsible for causing bleeding into the lungs and irreversible scarring over time. Although the exact mechanism is unknown, scientists have hypothesized that the offending agents could be bioactive proteins (such as histamine, ECP or VEGF). When triggered by a yet to be discovered antigen(s), the immune cells are inappropriately activated, causing the release of these factors and prompting bleeding in the lungs.

Some studies also suggest that IPH may have a genetic component because it has been reported in siblings and children of IPH patients. Moreover, patients with Down syndrome could be at a higher risk of developing IPH. Other research suggests there may be an environmental component to the disease, including secondhand smoking and mold exposure. More research is needed to evaluate these hypotheses. Due to the rarity of the disease, research can be difficult and take longer than other, more common diseases.

### **Affected populations**

Approximately 80% of cases occur in children, mostly under 10 years old and 20% of cases occur in adults (majority under 30 years old). IPH may affect more girls than boys and more adult men than women, according to some studies.

### **Disorders with Similar Symptoms**

The following conditions have signs and symptoms that are similar to IPH: Goodpasture syndrome, acute respiratory distress syndrome (ARDS), lung infections, systemic lupus erythematous, Henoch-Schoenlein purpura, ANCA vasculitis, and mixed connective tissue disease.

### **Diagnosis**

Since it is a rare disease, diagnosing IPH involves ruling out all other possible causes of bleeding or scarring in the lungs. This may include various blood tests (iron studies, blood cell counts and antibody levels), sputum tests, imaging (x-rays, CT scans), bronchoscopy and biopsies. A multidisciplinary team is often involved and may include pulmonologists, hospitalists, rheumatologists, respiratory therapists, intensivists and thoracic surgeons. Early recognition and treatment can help avoid serious complications and disease progression. IPH is typically diagnosed using a camera (bronchoscope) that takes samples of the fluid in the lungs, known as a bronchoalveolar lavage. The pulmonologist will also take biopsies of the lungs to confirm the diagnosis under the microscope.

### **Standard Therapies**

**Treatment**

The goal of treatment is to suppress this immune response and decrease the damage by preventing repeated bleeding episodes. There is no gold standard treatment for IPH yet. Physicians determine the best therapies based on their experience and available research.

Some commonly accepted therapies include:

Steroids—These medications may control bleeding into the lungs and scarring that happens afterward.

Immunosuppressants– In addition to steroids, these medications subdue the immune system further and assist in preventing bleeding in the lungs. These drugs may include 6-mercaptopurine/azathioprine, hydroxychloroquine, cyclophosphamide, mycophenolate mofetil or rituximab.

Blood transfusions– Replacing the blood lost due to lung bleeding may help the symptoms of anemia.

Stem cell transplant—This experimental therapy uses the body’s own stem cells to modulate the immune system and prevent bleeding.

Extracorporeal membrane oxygenation (ECMO)—This is a temporary life support system during acute severe bleeding or end-stage lung disease until a lung transplant can be accomplished for appropriate patients.

Lung transplant—This is a final resort for severe IPH. IPH may recur in the transplanted lung, but few lung transplant cases have been reported.

## **Differential Diagnosis of Pediatric IPH:**

1. Pulmonary Vasculitis / Capillaritis Syndromes
   1. Examples: Microscopic polyangiitis, granulomatosis with polyangiitis (Wegener’s)
   2. Characterized by immune-mediated alveolar capillary inflammation causing diffuse alveolar hemorrhage (DAH).
   3. Usually accompanied by systemic symptoms and positive serologic markers (ANCA, anti-GBM antibodies).
2. Autoimmune and Connective Tissue Diseases
   1. Systemic lupus erythematosus, juvenile idiopathic arthritis, systemic sclerosis can manifest with DAH.
   2. Often accompanied by other systemic signs and positive autoantibodies.
3. Cardiac Causes
   1. Left-sided heart failure, mitral valve disease leading to pulmonary venous hypertension and hemorrhage.
4. Infections
   1. Bacterial, viral, fungal, or parasitic infections causing pulmonary hemorrhage or secondary inflammation.
   2. Diagnosis aided by microbiological tests.
5. Coagulopathies and Hematological Disorders
   1. Bleeding disorders (e.g., thrombocytopenia, hemophilia), bone marrow failure, or malignancies.
   2. Lab tests include coagulation studies, bone marrow biopsy if indicated.
6. Environmental / Toxic Exposures
   1. Inhalation of toxic agents, drugs causing lung bleeding.
7. Foreign Body Aspiration or Trauma
   1. Especially focal hemorrhage, history of trauma or aspiration event.
8. Pulmonary Hemosiderosis Secondary to Other Causes
   1. Secondary hemosiderosis due to recurrent bleeding from infections, bronchiectasis, cystic fibrosis.
9. Idiopathic Pulmonary Hemosiderosis (IPH)
   1. Diagnosis of exclusion when no cause is found; characterized by repeated episodes of diffuse alveolar hemorrhage leading to iron deficiency anemia and pulmonary infiltrates.

**Pediatric Idiopathic Pulmonary Hemosiderosis (IPH) Epidemiology:**

* IPH is a rare but serious disease in children, with an estimated incidence of approximately 0.24 to 1.23 cases per million children per year.
* The condition typically presents in the pediatric age group more frequently than in adults but can occur at any age.
* IPH has a high mortality rate, reported around 50%, largely due to delayed diagnosis and progressive lung damage if untreated.
* The classical triad of presentations—hemoptysis, iron-deficiency anemia, and diffuse lung infiltrates—is often incomplete or absent in children early on, leading to diagnostic delays.
* There is some geographic variation reported but no clear gender predilection is consistently described.
* Early recognition and treatment with immunosuppression can improve outcomes, although prognosis remains guarded in severe cases.
* IPH may be associated with immune-mediated processes, and some pediatric patients also show coexisting conditions such as celiac disease (Lane–Hamilton syndrome)

**PREDEFINED QUESTIONS AND ANSWERS**

## What is Idiopathic Pulmonary Hemosiderosis (IPH) in children?

IPH is a rare lung disease characterized by recurrent episodes of bleeding into the lungs (alveolar hemorrhage) without an identifiable underlying cause, leading to iron deficiency anemia and lung infiltrates.

## What causes IPH?

The exact cause is unknown. Theories include autoimmune injury to the alveolar capillaries, allergic factors, environmental exposures, and possible genetic predisposition. It is considered idiopathic because no specific trigger is identified.

## What are the common symptoms of pediatric IPH?

Symptoms often include:

* Iron deficiency anemia (may be the initial or only sign)
* Recurrent cough and difficulty breathing (dyspnea)
* Repeated lung infiltrates visible on chest imaging
* Hemoptysis (coughing up blood) occurs in about half of cases but may be absent or unnoticed in young children who swallow sputum.

## How is pediatric IPH diagnosed?

Diagnosis involves:

* Clinical evaluation showing anemia and respiratory symptoms
* Chest X-rays or CT showing diffuse lung infiltrates
* Bronchoalveolar lavage demonstrating hemosiderin-laden macrophages (siderophages)
* Exclusion of other causes of alveolar hemorrhage such as vasculitis or infections
* Lung biopsy may be required if diagnosis is unclear

## How common is IPH in children?

IPH is very rare, with an incidence estimated at about 0.24 to 1.3 cases per million children annually.

## What is the typical age of onset?

Most children are diagnosed before the age of 10, often between 1 and 7 years old.

## What is the prognosis of pediatric IPH?

The disease course is variable; some children respond to treatment with corticosteroids and immunosuppressants, while others have recurrent hemorrhages leading to lung fibrosis. Early diagnosis and treatment improve outcomes, but mortality can be as high as 30-50% without intervention.

## How is pediatric IPH treated?

* Systemic corticosteroids are the first-line therapy to control bleeding episodes.
* Immunosuppressive agents may be added if steroids alone are insufficient.
* Supportive care includes treating anemia and preventing infections.
* If celiac disease is present, a gluten-free diet may help.

## What causes delays in diagnosis?

IPH often presents insidiously, with anemia sometimes being the only sign initially. The classic triad of hemoptysis, anemia, and infiltrates is incomplete or absent early, especially in young children, leading to diagnostic delay averaging 1–3 years.

## Are there any associated conditions?

Some children with IPH have auto-immune markers or conditions such as celiac disease, suggesting autoimmune involvement.

**DOCTOR PATIENT CONVERSATION**

Doctor: Hello. I wanted to discuss your child’s recent diagnosis. After several tests and hospital admissions, we have identified that your child has idiopathic pulmonary hemosiderosis, or IPH.

Parent: I’m not familiar with that. What exactly is IPH?

Doctor: IPH is a rare lung disease where there are repeated episodes of bleeding into the lungs. This bleeding leads to inflammation and causes symptoms like cough, breathing difficulties, and anemia because the blood loss reduces healthy red blood cells.

Parent: What causes this bleeding? Is it our child’s fault or something serious?

Doctor: The term ‘idiopathic’ means we don’t know the exact cause—it’s not related to infections, heart disease, or other conditions. It likely involves the immune system causing injury to the small blood vessels in the lungs.

Parent: What symptoms should we expect, and what did we notice in our child?

Doctor: Children with IPH often develop cough and shortness of breath. Sometimes they cough up blood, but younger children may swallow it, so it may go unnoticed. Your child has shown signs of anemia, which explains why they have been feeling tired and appearing pale.

Parent: How do you diagnose it?

Doctor: We use chest X-rays and CT scans showing abnormal lung infiltrates. A key test is bronchoalveolar lavage during bronchoscopy, where we sample lung fluid to look for special cells called hemosiderin-laden macrophages that indicate bleeding inside the lungs. We also rule out other causes like infections or autoimmune diseases before calling it idiopathic.

Parent: What kind of treatment will my child need?

Doctor: The main treatment is medications called corticosteroids, which reduce inflammation and stop the bleeding. In severe cases, high doses are given initially, followed by a maintenance dose. Some children may require other immunosuppressive medicines if steroids alone aren’t enough. Supportive care, such as oxygen and blood transfusions, may be needed during flare-ups.

Parent: Is this a lifelong condition? Can it be cured?

Doctor: IPH is chronic, so ongoing treatment and monitoring are necessary. While there’s no permanent cure, most children respond well to treatment, especially if started early. The goal is to prevent bleeding episodes and protect lung function.

Parent: What should we watch for at home? When should we seek urgent care?

Doctor: Watch for worsening cough, difficulty breathing, pale or blue lips, excessive tiredness, or coughing up blood. These signs need immediate medical attention. Also, keep all follow-up appointments and ensure your child gets routine vaccinations to prevent infections.

Parent: Can infections make this worse?

Doctor: Yes, respiratory infections can trigger flare-ups. Protecting your child with vaccines and good hand hygiene is very important.

Parent: What is the usual outlook for children with IPH?

Doctor: It varies, but with proper treatment, many children live healthy lives with few complications. Some might have recurrent flare-ups, so we need to be vigilant. We’ll guide you through managing this condition step by step.

Parent: Thank you for explaining this. It’s a lot to take in, but I’m glad we know what’s going on.

Doctor: You’re welcome. We’re here to support you and your child. Please don’t hesitate to ask any questions or reach out if you’re concerned.

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