Chapter

22

Congenital Diaphragmatic Hernia

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An eight-day-old, ex-35-week gestation female presents for repair of a prenatally diagnosed left-sided congenital diaphragmatic hernia (CDH). The percent predicted lung volume (PPLV) was reported to be ~15–20%. Fetal echocardiogram showed no other abnormalities. She was delivered via emergency cesarean section due to nonreassuring fetal heart tracings. APGAR scores were 1 and 6, at one and five minutes respectively. CPR was initiated in the delivery room and she was transferred to the intensive care unit, intubated, and ventilated.

What Is a Congenital Diaphragmatic Hernia (CDH)?

A congenital diaphragmatic hernia (CDH) is a defect in the diaphragm occurring during embryologic development that results in herniation of the abdominal contents into the chest. This leads to the compression of the lungs and abnormal pulmonary vascular development. The diaphragm normally develops in the first trimester, with the right diaphragm closing before the left. The organs that are typically found herniated into the thoracic cavity are the stomach, intestines, spleen, and/or liver (Figure 22.1). There are two common types of diaphragmatic hernias: Bochdalek and Morgagni. The Bochdalek hernia occurs on the posterior-lateral portion of the diaphragm, and the Morgagni hernia on the anterior aspect. These hernias can be right-sided, left-sided (most common), or bilateral.

The incidence of CDH has been reported to be between 1-in-2000 and 1-in-5000 live births.

Describe the Pathophysiology of CDH

CDH results in pulmonary hypoplasia from compression of the fetal lungs during development. Pulmonary vascular hyper-reactivity, or persistent pulmonary hypertension may also occur. Hypoxia, hypercapnia, respiratory acidosis, and hypothermia contribute to worsening pulmonary hypertension and persistence of the fetal circulation. This will lead to right-to-left shunting across a patent ductus arteriosus.

How Is the Diagnosis of CDH Made?

CDH is most commonly diagnosed with prenatal ultrasound, usually in the second or third trimester. However, fetal magnetic resonance imaging may also be used. The abdominal contents can be observed in the fetal chest on imaging. Ultrasound Doppler examination of the hepatic and umbilical vessels can aid in diagnosis. Echocardiography and genetic testing may be performed to exclude co-existing anatomic anomalies and syndromic associations.

Early diagnosis allows for counseling, coordination of delivery at a specialized tertiary center, and consideration of elective termination if desired by the family.

What Is the Differential Diagnosis for CDH?

Other diagnoses to be considered in suspected CDH include diaphragm eventration, teratoma, bronchogenic or enteric cysts, and congenital cystic adenomatoid malformation.

How Is the Prognosis Determined?

Prognosis in CDH is determined by prenatal lung volume-based prognostication. The lung-to-head (LHR) ratio is most commonly used. Another method used is percent predicted lung volume (PPLV).

The presence and extent of liver herniation, or concomitant major anomalies, is indicative of poor prognosis.

Presence of liver herniation correlates with the need for extracorporeal membrane oxygenation (ECMO) support. Greater than 80% of patients with liver herniation into the thorax require ECMO

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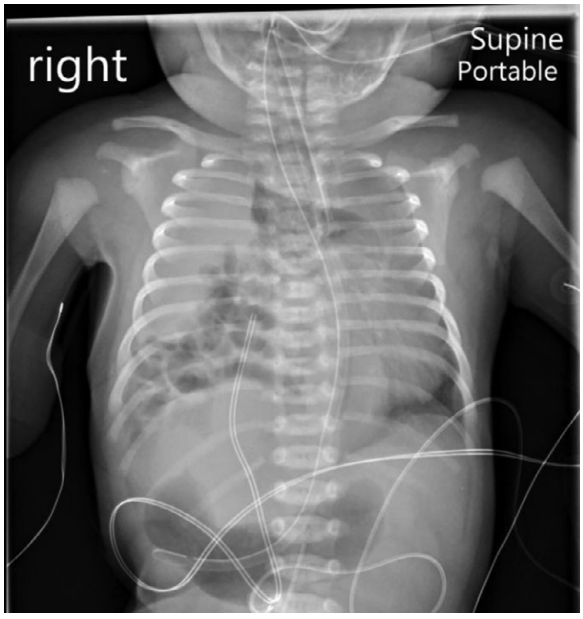

Figure 22.1 Chest X-ray demonstrating a large right-sided congenital diaphragmatic hernia with herniation of the bowel into the right hemithorax. The right lung markings are absent and the trachea is deviated leftwards. Courtesy of Adam C. Adler MD

support compared with 25% of patients without liver herniation.

What Is the Lung-to-Head Ratio (LHR)?

The LHR is calculated by measuring the lung area (on the contralateral side of the hernia) using ultrasound, and then dividing the number by the head circumference. A value less than 0.9 denotes a low rate of survival, whereas above 1.4 favors survival.

What Is Percent Predicted Lung Volume (PPLV)?

The PPLV is calculated by taking the total measured thoracic volume, and subtracting the measured mediastinal volume from this value. These measurements are based on fetal magnetic resonance imaging. A value of 15 or less denotes high risk of pulmonary morbidity and/or death.

What Is the Morbidity/Mortality Rate for CDH?

A survival rate of approximately 70% and higher has been reported in specialized tertiary care centers.

The extent of abdominal viscera herniation and presence of associated anomalies also contributes to the overall morbidity and mortality. Infants with single ventricle physiology in combination with CHD have a near 100% mortality. Patients with familial, bilateral, or syndromic CDH and those in which CDH is associated with specific genetic abnormalities are all associated with poor outcomes.

What Prenatal Interventions Are Available for CDH?

Fetal intervention has evolved in the treatment of CDH. Previously, anatomical repair of the diaphragm defect was performed in-utero; however, due to lack of improvement in overall survival, this is no longer performed. Currently, endoscopic tracheal occlusion is the most common fetal intervention for CDH. This procedure is performed between 23 and 27 weeks of gestation. A balloon in the trachea blocks the exit of lung fluid and results in large fluid-filled lungs. An Ex-Utero Intrapartum Treatment (EXIT) procedure is performed at term to remove the balloon from the trachea prior to delivery.

This approach may still result in impaired pulmonary function and is generally reserved for severe cases of CDH. Antenatal maternal steroids may be administered to improve lung maturity; however improved outcomes have not been demonstrated.

What Is the Plan for Delivery of a Fetus with CDH?

Ideally, vaginal delivery can be accomplished with induction of labor around 38 weeks of gestation. Vaginal delivery is beneficial for lung function with extrusion of lung amniotic fluid during descent. A cesarean delivery may be performed when the clinical context dictates. Regardless of the type of delivery, all infants should undergo tracheal intubation and placement of an orogastric tube at the time of birth to prevent distension of abdominal contents within the chest cavity.

What Are the Ventilator Management Options?

Gentle ventilation and permissive hypercapnia are most often used to minimize lung trauma to the

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hypo-plastic lung. Occasionally high-frequency oscillation and inhaled nitric oxide are required. Surgical repair is usually delayed until after the pulmonary vascular reactivity is improved. Early institution of extracorporeal membrane oxygenation (ECMO) prior to surgical repair may be necessary.

What Are Gentle Ventilation Strategies for CDH?

- Low peak inspiratory pressures, less than 25 cm H_2O
- Minimal positive end expiratory pressures, less than 5 cm H_2O
- Permissive hypercapnia
- Transitioning to high-frequency oscillation ventilation to prevent severe hypercapnia

What Type of ECMO Is Used in CDH?

Veno-arterial (VA) ECMO provides full cardiopulmonary support, with the right common carotid artery and right internal jugular vein being cannulated. Veno-venous (VV) ECMO provides only pulmonary support. In VV ECMO, a double-lumen cannula is inserted into the internal jugular vein. Both VV- and VA-ECMO are equally effective in managing CDH patients, however VA-ECMO is the most common form used.

When Should ECMO Be Instituted?

ECMO should be instituted when conventional therapy has been exhausted. The exact criteria depend on the institution. The EURO consensus criteria are:

- Inability to maintain pre-ductal saturations >85% or post-ductal saturations >70%.
- Increased $PaCO_2$ and respiratory acidosis with pH <7.15 despite optimal ventilator management.
- Peak inspiratory pressure >28 cm H_2O or mean airway pressure >17 cm H_2O is required to achieve saturation >85%.
- Inadequate oxygen delivery with metabolic acidosis as measured by elevated lactate > or = to 5 mmol/L and pH <7.15.
- Systemic hypotension, resistant to fluid or inotropic therapy, resulting in urine output <0.5 mL/kg/h for 12–24 hours.
- Oxygenation index (mean airway pressure × FiO_2 × 100/PaO_2) > or = 40 for at least 3 hours.

How Is CDH Repaired Surgically?

Laparoscopic, thoracoscopic, or open repair closure techniques have all been reported. With smaller diaphragmatic defects, primary closure can be accomplished; for larger defects, a Gore-Tex patch is placed.

When Is a Patient Considered Ready for Surgery?

The EURO consensus surgical criteria are:

- Mean arterial blood pressure is normal for gestation age.
- Pre-ductal saturation levels of 8–95% on FiO_2 below 50%.
- Lactate below 3 mmol/L.
- Urine output more than 1 mL/kg/h.

How Can You Manage the Associated Pulmonary Hypertension?

Inhaled nitric oxide can be used in the treatment of pulmonary hypertension. Other medical therapies, such as sildenafil, have been used but have not shown consistent benefit in CDH.

What Are the Long-Term Complications of CDH?

There are a number of long-term sequelae that have been observed in patients with CDH.

These include:

- recurrent diaphragmatic hernias,
- patch infection,
- intestinal obstruction (10% of patients),
- neurological impairment (intraventricular hemorrhage/post ECMO sequelae),
- gastroesophageal reflux disease (from alterations in normal anatomic relationships),
- chronic lung disease,
- pulmonary hypertension,
- failure-to-thrive,
- sensorineural hearing loss,
- chest asymmetry (pectus excavatum or carinatum), and
- scoliosis.

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Suggested Reading

Barnewolt CE, Kunisaki SM, Fauza DO, et al. Percent predicted lung volumes as measured on fetal magnetic resonance imaging: a useful biometric parameter for risk stratification in congenital diaphragmatic hernia. *J Pediatr Surg*. 2007;42(1):193–7. PMID: 17208564.

Guner YS, Khemani RG, Qureshi FG, et al. Outcome analysis of neonates with congenital diaphragmatic hernia treated with venovenous vs venoarterial extracorporeal membrane oxygenation. *J Pediatr Surg*. 2009;44:1691–1701. PMID: 19735810.

Hedrick H. Management of prenatally diagnosed congenital diaphragmatic hernia. *Semin Pediatr Surg*. 2013; 22(1):37–43. PMID: 23395144.

Haroon J, Chamberlain RS. An evidence-based review of the current treatment of congenital diaphragmatic hernia. *Clin Pediatr* 2013;52(2):115–24. PMID: 23378478.

Harrison MR, Keller RL, Hawgood SB, et al. A randomized trial of fetal endoscopic tracheal occlusion for severe fetal congenital diaphragmatic hernia. *N Engl J Med*. 2003;349(20):1916–24. PMID: 14614166.

Keijzer R, Wilschut DE, Jan Houmes R, et al. Congenital diaphragmatic hernia: to repair on or off extracorporeal membrane oxygenation? *J Pediatr Surg*. 2012; 47(4):631–6. PMID: 22498373.

Morini F, Goldman A, Pierro A. Extracorporeal membrane oxygenation in infants with congenital diaphragmatic hernia: a systematic review of the evidence. *Eur J Pediatr Surg*. 2006;16:385–91. PMID: 17211783.

Snoek KG, Reiss IKM, Greenough A, et al. Standardized postnatal management of infants with congenital diaphragmatic hernia in Europe: The CDH EURO Consortium Consensus – 2015 Update. *Neonatology*. 2016;110: 66–74. PMID: 29283134.

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