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Birth Defects

What are birth defects?

A birth defect is a problem that happens while a baby is developing [<https://medlineplus.gov/fetalhealthanddevelopment.html>] in the mother's body. Most birth defects happen during the first 3 months of pregnancy. One out of every 33 babies in the United States is born with a birth defect.

A birth defect may affect how the body looks, works, or both. Some birth defects like cleft lip [<https://medlineplus.gov/cleftlipandpalate.html>] or neural tube defects [<https://medlineplus.gov/neuraltubedefects.html>] are structural problems that can be easy to see. Others, like heart disease [<https://medlineplus.gov/heartdiseases.html>] , are found using special tests. Birth defects can range from mild to severe. How a birth defect affects a child's life depends mostly on which organ or body part is involved and how severe the defect is.

What causes birth defects?

For some birth defects, researchers know the cause. But for many birth defects, the exact cause is unknown. Researchers think that most birth defects are caused by a complex mix of factors, which can include:

- **Genetics.** One or more genes might have a change or mutation that prevents them from working properly. For example, this happens in Fragile X syndrome [<https://medlineplus.gov/fragilexsyndrome.html>] . With some defects, a gene or part of the gene might be missing.
- **Chromosomal problems.** In some cases, a chromosome or part of a chromosome might be missing. This is what happens in Turner syndrome [<https://medlineplus.gov/turnersyndrome.html>] . In other cases, such as with Down syndrome [<https://medlineplus.gov/downsyndrome.html>] , the child has an extra chromosome.
- **Exposures to medicines** [<https://medlineplus.gov/pregnancyandmedicines.html>] , **chemicals**, or other toxic substances [<https://medlineplus.gov/reproductivehazards.html>] . For example, alcohol misuse can cause fetal alcohol spectrum disorders [<https://medlineplus.gov/fetalalcoholspectrumdisorders.html>] .
- Infections during pregnancy [<https://medlineplus.gov/infectionsandpregnancy.html>] . For example, infection with Zika virus [<https://medlineplus.gov/zikavirus.html>] during pregnancy can cause a serious defect in the brain [<https://medlineplus.gov/brainmalformations.html>] .
- **Lack of certain nutrients.** Not getting enough folic acid [<https://medlineplus.gov/folicacid.html>] before and during pregnancy is a key factor in causing neural tube defects.

Who is at risk of having a baby with birth defects?

Certain factors may increase the chances of having a baby with a birth defect, such as:

- Smoking, drinking alcohol, or taking certain "street" drugs during pregnancy [<https://medlineplus.gov/pregnancyandsubstanceuse.html>]
- Having certain medical conditions, such as obesity [<https://medlineplus.gov/obesity.html>] or uncontrolled diabetes [<https://medlineplus.gov/diabetes.html>] , before and during pregnancy
- Taking certain medicines
- Having someone in your family with a birth defect. To learn more about your risk of having a baby with a birth defect, you can talk with a genetic counselor [<https://medlineplus.gov/geneticcounseling.html>] ,
- Being an older mother, typically over the age of 34 years

How are birth defects diagnosed?

Health care providers can diagnose some birth defects during pregnancy, using prenatal testing [https://medlineplus.gov/prenataltesting.html] . That's why it important to get regular prenatal care [https://medlineplus.gov/prenatalcare.html] .

Other birth defects may not be found until after the baby is born. Providers may find them through newborn screening [https://medlineplus.gov/newbornscreening.html] . Some defects, such as club foot, are obvious right away. Other times, the health care provider may not discover a defect until later in life, when the child has symptoms.

What are the treatments for birth defects?

Children with birth defects often need special care and treatments. Because the symptoms and problems caused by birth defects vary, the treatments also vary. Possible treatments may include surgery, medicines, assistive devices [https://medlineplus.gov/assistivedevices.html] , physical therapy, and speech therapy.

Often, children with birth defects need a variety of services and may need to see several specialists. The primary health care provider can coordinate the special care that the child needs.

Can birth defects be prevented?

Not all birth defects can be prevented. But there are things you can do before and during pregnancy to increase your chance of having a healthy baby:


- Start prenatal care as soon as you think you might be pregnant, and see your health care provider regularly during pregnancy
- Get 400 micrograms (mcg) of folic acid every day. If possible, you should start taking it at least one month before you get pregnant.
- Don't drink alcohol, smoke, or use "street" drugs
- Talk to your health care provider about any medicines you are taking or thinking about taking. This includes prescription and over-the-counter medicines, as well as dietary or herbal supplements.
- Learn how to prevent infections during pregnancy
- If you have any medical conditions, try to get them under control before you get pregnant

Centers for Disease Control and Prevention




Start Here

- About Birth Defects [https://www.cdc.gov/birth-defects/about/] (Centers for Disease Control and Prevention)
Also in Spanish [https://www.cdc.gov/birth-defects/es/about/sobre-los-defectos-de-nacimiento.html]
- About Congenital Anomalies [https://www.nichd.nih.gov/health/topics/congenital-anomalies/about]  (Eunice Kennedy Shriver National Institute of Child Health and Human Development)
Also in Spanish [https://espanol.nichd.nih.gov/salud/temas/birthdefects/informacion]
- Birth Defects (For Parents) [https://kidshealth.org/en/parents/birth-defects.html] (Nemours Foundation)
Also in Spanish [https://kidshealth.org/es/parents/birth-defects.html]
- Congenital Abnormalities [https://www.healthychildren.org/English/health-issues/conditions/developmental-disabilities/Pages/Congenital-Abnormalities.aspx] (American Academy of Pediatrics)
Also in Spanish [https://www.healthychildren.org/spanish/health-issues/conditions/developmental-disabilities/paginas/congenital-abnormalities.aspx]

Diagnosis and Tests

- Diagnosis of Birth Defects [https://www.cdc.gov/birth-defects/screening/diagnosis.html] (Centers for Disease Control and Prevention)
- How Do Health Care Providers Diagnose Congenital Anomalies? [https://www.nichd.nih.gov/health/topics/congenital-anomalies/conditioninfo/diagnosed]  **Easy-to-Read** (Eunice Kennedy Shriver National Institute of Child Health and Human Development)

Also in Spanish [<https://espanol.nichd.nih.gov/salud/temas/birthdefects/informacion/diagnostican>]

- **Karyotype Genetic Test** [<https://medlineplus.gov/lab-tests/karyotype-genetic-test/>]  (National Library of Medicine)
Also in Spanish [<https://medlineplus.gov/spanish/pruebas-de-laboratorio/prueba-de-cariotipo/>]
- **Newborn Screening: MedlinePlus Health Topic** [<https://medlineplus.gov/newbornscreening.html>]  (National Library of Medicine)
Also in Spanish [<https://medlineplus.gov/spanish/newbornscreening.html>]
- **Prenatal Testing: MedlinePlus Health Topic** [<https://medlineplus.gov/prenataltesting.html>]  (National Library of Medicine)
Also in Spanish [<https://medlineplus.gov/spanish/prenataltesting.html>]


Prevention and Risk Factors

- **Good Health Habits and Birth Defects Prevention** [<https://www.marchofdimes.org/find-support/blog/good-health-habits-and-birth-defects-prevention>] (March of Dimes Foundation)
Also in Spanish [<https://nacersano.marchofdimes.org/complicaciones/defectos-de-nacimiento.aspx>]
- **Preventing Birth Defects** [<https://www.cdc.gov/birth-defects/prevention/>] (Centers for Disease Control and Prevention)
- **Reducing Risks of Birth Defects** [<https://www.acog.org/womens-health/faqs/reducing-risks-of-birth-defects>] (American College of Obstetricians and Gynecologists)

Living With

- **Assistive Devices: Positioning Aids (Slideshow)** [<https://kidshealth.org/en/parents/assistive-devices-positioners.html>] *Easy-to-Read* (Nemours Foundation)
Also in Spanish [<https://kidshealth.org/es/parents/assistive-devices-positioners.html>]
- **If Your Baby Has a Birth Defect** [<https://kidshealth.org/en/parents/baby-has-birth-defect.html>] (Nemours Foundation)
Also in Spanish [<https://kidshealth.org/es/parents/baby-has-birth-defect.html>]

Related Issues





- **Isotretinoin and Other Retinoids during Pregnancy** [<https://www.marchofdimes.org/find-support/topics/pregnancy/isotretinoin-and-other-retinoids-during-pregnancy>]  (March of Dimes Foundation)
Also in Spanish [<https://nacersano.marchofdimes.org/embarazo/accutane-y-otros-retinoides.aspx>]
- **When Your Baby's in the NICU (Neonatal Intensive Care Unit)** [<https://kidshealth.org/en/parents/nicu-caring.html>] (Nemours Foundation)
Also in Spanish [<https://kidshealth.org/es/parents/nicu-caring.html>]

Specifics

- **Anophthalmia and Microphthalmia** [<https://www.nei.nih.gov/learn-about-eye-health/eye-conditions-and-diseases/anophthalmia-and-microphthalmia>]  (National Eye Institute)
Also in Spanish [<https://www.nei.nih.gov/espanol/aprenda-sobre-la-salud-ocular/enfermedades-y-afecciones-de-los-ojos/anoftalmia-y-microftalmia>]
- **Atypical Genitalia** [<https://www.mayoclinic.org/diseases-conditions/atypical-genitalia/symptoms-causes/syc-20369273?p=1>] (Mayo Foundation for Medical Education and Research)
Also in Spanish [<https://www.mayoclinic.org/es/diseases-conditions/ambiguous-genitalia/symptoms-causes/syc-20369273?p=1>]
- **Bardet-Biedl Syndrome** [<https://www.fightingblindness.org/diseases/bardet-biedl-syndrome-bbs>] (Foundation Fighting Blindness)
- **Branchio-oto-renal Syndrome** [<https://rarediseases.info.nih.gov/diseases/10147/bor-syndrome>]  (Genetic and Rare Diseases Information Center)
- **Campomelic dysplasia** [<https://rarediseases.info.nih.gov/diseases/10027/campomelic-dysplasia/>]  (Genetic and Rare Diseases Information Center)
Also in Spanish [<https://rarediseases.info.nih.gov/espanol/11882/displasia-campomelica/>]
- **Caring for Your Child with Clubfoot** [<https://kidshealth.org/en/parents/center/clubfoot-center.html>] (Nemours Foundation)
Also in Spanish [<https://kidshealth.org/es/parents/center/clubfoot-center.html>]

Clubfoot [<https://www.mayoclinic.org/diseases-conditions/clubfoot/symptoms-causes/syc-20350860?p=1>]


(Mayo Foundation for Medical Education and Research)

- Esophageal Atresia and Tracheoesophageal Fistula [<https://familydoctor.org/condition/esophageal-atresia-and-tracheoesophageal-fistula/?adfree=true>] (American Academy of Family Physicians)
Also in Spanish [<https://es.familydoctor.org/condicion/atresia-esofagica-y-fistula-traqueoesofagica-es/?adfree=true>]
- Fetal Abdominal Cyst [<https://kidshealth.org/en/parents/fetal-ab-cyst.html>] (Nemours Foundation)
Also in Spanish [<https://kidshealth.org/es/parents/fetal-ab-cyst.html>]
- Fetal Lung Mass [<https://kidshealth.org/en/parents/fetal-lung-mass.html>] (Nemours Foundation)
Also in Spanish [<https://kidshealth.org/es/parents/fetal-lung-mass.html>]
- Fibular Hemimelia [<https://kidshealth.org/en/parents/fibular-hemimelia.html>] (Nemours Foundation)
Also in Spanish [<https://kidshealth.org/es/parents/fibular-hemimelia.html>]
- Gastroschisis [<https://www.cdc.gov/birth-defects/about/gastroschisis.html>] (Centers for Disease Control and Prevention)
Also in Spanish [<https://www.cdc.gov/birth-defects/es/about/gastrosquisis.html>]
- Genital and Urinary Tract Defects [<https://www.marchofdimes.org/find-support/topics/planning-baby/genital-and-urinary-tract-defects>] (March of Dimes Foundation)
Also in Spanish [<https://nacersano.marchofdimes.org/complicaciones/defectos-del-tracto-genitourinario.aspx>]
- Hirschsprung Disease [<https://kidshealth.org/en/parents/hirschsprung.html>] (Nemours Foundation)
Also in Spanish [<https://kidshealth.org/es/parents/hirschsprung.html>]
- Hirschsprung Disease [<https://www.niddk.nih.gov/health-information/digestive-diseases/hirschsprung-disease>]
 (National Institute of Diabetes and Digestive and Kidney Diseases)
- Hypospadias [<https://www.cdc.gov/birth-defects/about/hypospadias.html>] (Centers for Disease Control and Prevention)
Also in Spanish [<https://www.cdc.gov/birth-defects/es/about/hipospadias.html>]
- Limb Reduction Defects [<https://www.cdc.gov/birth-defects/about/limb-reduction-defects.html>]
(Centers for Disease Control and Prevention)
Also in Spanish [<https://www.cdc.gov/birth-defects/es/about/defectos-por-reduccion-de-extremidades.html>]
- Mobius Syndrome [<https://rarediseases.info.nih.gov/diseases/8549/mobius-syndrome>]
 (Genetic and Rare Diseases Information Center)
- Omphalocele [<https://www.cdc.gov/birth-defects/about/omphalocele.html>] (Centers for Disease Control and Prevention)
Also in Spanish [<https://www.cdc.gov/birth-defects/es/about/onfalocele.html>]
- Pierre Robin Syndrome [<https://kidshealth.org/en/parents/pierre-robin-syndrome.html>] (Nemours Foundation)
Also in Spanish [<https://kidshealth.org/es/parents/pierre-robin-syndrome.html>]
- Treating Gastroschisis (Gastroschisis Silo) [https://kidshealth.org/en/parents/gastroschisis_silo.html]
 (Nemours Foundation)
- Undescended Testicles (Cryptorchidism) (For Parents) [<https://kidshealth.org/en/parents/cryptorchidism.html>]
(Nemours Foundation)
Also in Spanish [<https://kidshealth.org/es/parents/cryptorchidism.html>]
- What Are the Types of Congenital Anomalies? [<https://www.nichd.nih.gov/health/topics/congenital-anomalies/conditioninfo/types>]  (Eunice Kennedy Shriver National Institute of Child Health and Human Development)
Also in Spanish [<https://espanol.nichd.nih.gov/salud/temas/birthdefects/informacion/tipos>]
- Williams Syndrome (For Parents) [<https://kidshealth.org/en/parents/williams-syndrome.html>] (Nemours Foundation)
Also in Spanish [<https://kidshealth.org/es/parents/williams-syndrome.html>]

Statistics and Research

- Birth Defects Tracking [<https://www.cdc.gov/birth-defects/tracking/>] (Centers for Disease Control and Prevention)
- Data and Statistics on Birth Defects [<https://www.cdc.gov/birth-defects/data-research/facts-stats/>]
(Centers for Disease Control and Prevention)
- FastStats: Birth Defects or Congenital Anomalies [<https://www.cdc.gov/nchs/fastats/birth-defects.htm>]
(National Center for Health Statistics)

Clinical Trials


- ClinicalTrials.gov: Congenital Abnormalities [<https://clinicaltrials.gov/search?cond=%22Congenital+Abnormalities%22&aggFilters=status:not%20rec>]  (National Institutes of Health)

Journal Articles



References and abstracts from MEDLINE/PubMed (National Library of Medicine)

- Article: Association Between Human Embryo Culture Conditions, Cryopreservation, and the Potential Risk... [<https://www.ncbi.nlm.nih.gov/pubmed/40731824>]
- Article: Advances in Fetal Surgery: A Narrative Review of Therapeutic Interventions and... [<https://www.ncbi.nlm.nih.gov/pubmed/40731766>]
- Article: Does extended follow-up period after birth improve precision of diagnosis of... [<https://www.ncbi.nlm.nih.gov/pubmed/40696248>]
- Birth Defects -- see more articles [<https://pubmed.ncbi.nlm.nih.gov/?term=%22Congenital+Abnormalities%22%5Bmajr%3Anoexp%5D+AND+humans%5Bmh%5D+AND+english%5Bla%5D+AND+%22last+1+Year%22+%5Bedat%5D+NOT+%28letter%5Bpt%5D+OR+case+reports%5Bpt%5D+OR+editorial%5Bpt%5D+OR+comment%5Bpt%5D%29+AND+free+full+text%5Bsb%5D+>]

Reference Desk

- Talking Glossary of Genomic and Genetic Terms [<https://www.genome.gov/genetics-glossary>]  (National Human Genome Research Institute)
Also in Spanish [<https://www.genome.gov/es/genetics-glossary>]

Find an Expert

- Eunice Kennedy Shriver National Institute of Child Health and Human Development [<https://www.nichd.nih.gov/>] 
Also in Spanish [<https://espanol.nichd.nih.gov/>]
- March of Dimes Foundation [<https://www.marchofdimes.org/>]
Also in Spanish [<https://nacersano.marchofdimes.org/>]
- National Institute of Neurological Disorders and Stroke [<https://www.ninds.nih.gov/>] 

Children

- Anotia (For Parents) [<https://kidshealth.org/en/parents/anotia.html>] (Nemours Foundation)
- Campomelic Dysplasia (For Parents) [<https://kidshealth.org/en/parents/campomelic-dysplasia.html>] (Nemours Foundation)
Also in Spanish [<https://kidshealth.org/es/parents/campomelic-dysplasia.html>]
- Constricted Ears [<https://kidshealth.org/en/parents/constricted-ears.html>] (Nemours Foundation)
Also in Spanish [<https://kidshealth.org/es/parents/constricted-ears.html>]
- Cryptotia [<https://kidshealth.org/en/parents/cryptotia.html>] (Nemours Foundation)
Also in Spanish [<https://kidshealth.org/es/parents/cryptotia.html>]
- Ear Canal Stenosis [<https://kidshealth.org/en/parents/ear-canal-stenosis.html>] (Nemours Foundation)
Also in Spanish [<https://kidshealth.org/es/parents/ear-canal-stenosis.html>]
- Microtia (For Parents) [<https://kidshealth.org/en/parents/microtia.html>] (Nemours Foundation)
Also in Spanish [<https://kidshealth.org/es/parents/microtia.html>]

Patient Handouts

- Differences of sex development [<https://medlineplus.gov/ency/article/001669.htm>] (Medical Encyclopedia)
Also in Spanish [<https://medlineplus.gov/spanish/ency/article/001669.htm>]



MEDICAL ENCYCLOPEDIA

Acrodysostosis [<https://medlineplus.gov/ency/article/001248.htm>]

Alpha fetoprotein [<https://medlineplus.gov/ency/article/003573.htm>]

Ambiguous genitalia [<https://medlineplus.gov/ency/article/003269.htm>]

Amniotic constriction bands [<https://medlineplus.gov/ency/article/001579.htm>]

Annular pancreas [<https://medlineplus.gov/ency/article/001142.htm>]

Clubfoot [<https://medlineplus.gov/ency/article/001228.htm>]

Clubfoot repair [<https://medlineplus.gov/ency/article/002964.htm>]

Clubfoot repair - series [https://medlineplus.gov/ency/presentations/100056_1.htm]

Congenital diaphragmatic hernia repair [<https://medlineplus.gov/ency/article/002936.htm>]

Developmental differences of the female genital tract [<https://medlineplus.gov/ency/article/001497.htm>]

Diaphragmatic hernia [<https://medlineplus.gov/ency/article/001135.htm>]

Diaphragmatic hernia repair - series [https://medlineplus.gov/ency/presentations/100014_1.htm]

Differences of sex development [<https://medlineplus.gov/ency/article/001669.htm>]

Duodenal atresia [<https://medlineplus.gov/ency/article/001131.htm>]

Fusion of the ear bones [<https://medlineplus.gov/ency/article/001633.htm>]

Gastroschisis repair [<https://medlineplus.gov/ency/article/002924.htm>]

Gastroschisis repair - series [https://medlineplus.gov/ency/presentations/100023_1.htm]

Hirschsprung disease [<https://medlineplus.gov/ency/article/001140.htm>]

Hypospadias [<https://medlineplus.gov/ency/article/001286.htm>]

Hypospadias repair [<https://medlineplus.gov/ency/article/003000.htm>]

Hypospadias repair - discharge [<https://medlineplus.gov/ency/patientinstructions/000158.htm>]

Hypospadias repair - series [https://medlineplus.gov/ency/presentations/100126_1.htm]

Imperforate anus [<https://medlineplus.gov/ency/article/001147.htm>]

Imperforate anus repair [<https://medlineplus.gov/ency/article/002926.htm>]

Imperforate anus repair - series [https://medlineplus.gov/ency/presentations/100030_1.htm]

Low-set ears and pinna abnormalities [<https://medlineplus.gov/ency/article/003303.htm>]

Meckel diverticulectomy [<https://medlineplus.gov/ency/article/002929.htm>]

Meckel diverticulum [<https://medlineplus.gov/ency/article/000234.htm>]

Meckel's diverticulectomy - series [https://medlineplus.gov/ency/presentations/100092_1.htm]

Omphalocele [<https://medlineplus.gov/ency/article/000994.htm>]

Omphalocele repair [<https://medlineplus.gov/ency/article/002938.htm>]

Omphalocele repair - series [https://medlineplus.gov/ency/presentations/100033_1.htm]

Polyhydramnios [<https://medlineplus.gov/ency/article/003267.htm>]

Prune belly syndrome [<https://medlineplus.gov/ency/article/001269.htm>]

Quadruple screen test [<https://medlineplus.gov/ency/article/007311.htm>]

Trisomy 13 [<https://medlineplus.gov/ency/article/001660.htm>]

Trisomy 18 [<https://medlineplus.gov/ency/article/001661.htm>]

Related Health Topics

Brain Malformations [<https://medlineplus.gov/brainmalformations.html>]

Chiari Malformation [<https://medlineplus.gov/chiarimalformation.html>]

Cleft Lip and Palate [<https://medlineplus.gov/cleftlipandpalate.html>]

Congenital Heart Defects [<https://medlineplus.gov/congenitalheartdefects.html>]

Craniofacial Abnormalities [<https://medlineplus.gov/craniofacialabnormalities.html>]

Fetal Alcohol Spectrum Disorders [<https://medlineplus.gov/fetalalcoholspectrumdisorders.html>]

Fetal Health and Development [<https://medlineplus.gov/fetalhealthanddevelopment.html>]

Genetic Brain Disorders [<https://medlineplus.gov/geneticbraindisorders.html>]

Genetic Disorders [<https://medlineplus.gov/geneticdisorders.html>]

Genetic Testing [<https://medlineplus.gov/genetictesting.html>]

Neural Tube Defects [<https://medlineplus.gov/neuraltubedefects.html>]

Prenatal Testing [<https://medlineplus.gov/prenataltesting.html>]

National Institutes of Health

The primary NIH organization for research on *Birth Defects* is the Eunice Kennedy Shriver National Institute of Child Health and Human Development [<https://www.nichd.nih.gov/Pages/index.aspx>]

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