Congenital diaphragmatic hernia

Description

Congenital diaphragmatic hernia is a defect in the diaphragm. The diaphragm, which is composed of muscle and other fibrous tissue, separates the organs in the abdomen from those in the chest. Abnormal development of the diaphragm before birth leads to defects ranging from a thinned area in the diaphragm to its complete absence. An absent or partially formed diaphragm results in an abnormal opening (hernia) that allows the stomach and intestines to move into the chest cavity and crowd the heart and lungs. This crowding can lead to underdevelopment of the lungs (pulmonary hypoplasia), potentially resulting in life-threatening breathing difficulties that are apparent from birth.

In 5 to 10 percent of affected individuals, signs and symptoms of congenital diaphragmatic hernia appear later in life and may include breathing problems or abdominal pain from protrusion of the intestine into the chest cavity. In about 1 percent of cases, congenital diaphragmatic hernia has no symptoms; it may be detected incidentally when medical imaging is done for other reasons.

Congenital diaphragmatic hernias are often classified by their position. A Bochdalek hernia is a defect in the side or back of the diaphragm. Between 80 and 90 percent of congenital diaphragmatic hernias are of this type. A Morgnani hernia is a defect involving the front part of the diaphragm. This type of congenital diaphragmatic hernia, which accounts for approximately 2 percent of cases, is less likely to cause severe symptoms at birth. Other types of congenital diaphragmatic hernia, such as those affecting the central region of the diaphragm, or those in which the diaphragm muscle is absent with only a thin membrane in its place, are rare.

Frequency

Congenital diaphragmatic hernia affects approximately 1 in 2,500 newborns.

Causes

Congenital diaphragmatic hernia has many different causes. In 10 to 15 percent of affected individuals, the condition appears as a feature of a disorder that affects many body systems, called a syndrome. Donnai-Barrow syndrome, Fryns syndrome, and Pallister-Killian mosaic syndrome are among several syndromes in which congenital diaphragmatic hernia may occur. Some of these syndromes are caused by changes in

single genes, and others are caused by chromosomal abnormalities that affect several genes.

About 25 percent of individuals with congenital diaphragmatic hernia that is not associated with a known syndrome also have abnormalities of one or more major body systems. Affected body systems can include the heart, brain, skeleton, intestines, genitals, kidneys, or eyes. In these individuals, the multiple abnormalities likely result from a common underlying disruption in development that affects more than one area of the body, but the specific mechanism responsible for this disruption is not clear.

Approximately 50 to 60 percent of congenital diaphragmatic hernia cases are isolated, which means that affected individuals have no other major malformations.

More than 80 percent of individuals with congenital diaphragmatic hernia have no known genetic syndrome or chromosomal abnormality. In these cases, the cause of the condition is unknown. Researchers are studying changes in several genes involved in the development of the diaphragm as possible causes of congenital diaphragmatic hernia. Some of these genes are transcription factors, which provide instructions for making proteins that help control the activity of particular genes (gene expression). Others provide instructions for making proteins involved in cell structure or the movement (migration) of cells in the embryo. Environmental factors that influence development before birth may also increase the risk of congenital diaphragmatic hernia, but these environmental factors have not been identified.

Inheritance

Isolated congenital diaphragmatic hernia is rarely inherited. In almost all cases, there is only one affected individual in a family.

When congenital diaphragmatic hernia occurs as a feature of a genetic syndrome or chromosomal abnormality, it may cluster in families according to the inheritance pattern for that condition.

Other Names for This Condition

- Congenital diaphragmatic defect

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Diaphragmatic hernia 2 (https://www.ncbi.nlm.nih.gov/gtr/conditions/C1857284/)
- Genetic Testing Registry: Diaphragmatic hernia 3 (https://www.ncbi.nlm.nih.gov/gtr/conditions/C1857781/)

Genetic and Rare Diseases Information Center

- Congenital diaphragmatic hernia (https://rarediseases.info.nih.gov/diseases/1481/index)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%22Congenital diaphragmatic hernia%22)

Catalog of Genes and Diseases from OMIM

- DIAPHRAGMATIC HERNIA, CONGENITAL (https://omim.org/entry/142340)
- DIAPHRAGMATIC HERNIA 2; DIH2 (https://omim.org/entry/222400)
- DIAPHRAGMATIC HERNIA 3; DIH3 (https://omim.org/entry/610187)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28Hernia,+Diaphragmatic%5BMAJR%5D%29+AND+%28congenital+diaphragmatic+hernia%5BTI%5D%29+AND+review%5Bpt%5D+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D)

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