Fact Sheet

Division of Child and Adolescent Health
Pediatric Screening and Genetic Services
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VDH VIRGINIA DEPARTMENT OF HEALTH
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www.vdh.virginia.gov

Congenital Diaphragmatic Hernia

What is congenital diaphragmatic hernia?
Congenital diaphragmatic hernia is an opening in the diaphragm. The diaphragm is the muscle that usually separates the lungs and chest cavity from the abdominal cavity. A diaphragmatic hernia allows the abdominal organs (stomach, intestines, spleen, liver, kidneys) to move into the chest cavity and compress the lungs.

What types of problems occur with congenital diaphragmatic hernia?
Even before a baby is born, the pressure from the abdominal organs can compress the lungs. This pressure prevents the lungs from growing well. After birth, a baby needs to use his or her lungs to breathe. If the diaphragm does not work well, the lungs can collapse, resulting in respiratory distress (severe breathing difficulties), a bluish tint to the skin, fast breathing, and a fast heart rate.

Diaphragmatic hernia is a very serious condition. While surgery can help babies, unfortunately some will die from having underdeveloped lungs. Even after surgery, children may have long-term breathing or digestive problems. Some children with diaphragmatic hernia will have other birth defects as well.

How common is congenital diaphragmatic hernia?
Diaphragmatic hernia occurs in about 1800 births each year in the United States. In Virginia, approximately 25 children are born yearly with diaphragmatic hernia.

What causes congenital diaphragmatic hernia?
Diaphragmatic hernia can occur as the only birth defect a child has. It can also occur in combination with other features as part of a syndrome (combination of findings). The way in which a syndrome is passed through the family is specific to the given syndrome. A genetic counselor or geneticist can help you determine the risks for your family and situation. If the diaphragmatic hernia is not part of a syndrome, it is likely due to a combination of genetic and environmental factors. Parents of a child with a diaphragmatic hernia have about a 2% chance of having another child with a diaphragmatic hernia.

How is congenital diaphragmatic hernia treated?
Surgery is needed soon after birth to repair the diaphragm and return the abdominal organs to the abdominal cavity. If infants need help breathing, machines like ECMO (extracorporeal membrane oxygenation) or a ventilator may be used. If infants have breathing tubes in place, they will also need a tube inserted into their stomachs to provide food.

Where can I go for more information about congenital diaphragmatic hernia?
CHERUBS, The Association of Congenital Diaphragmatic Hernia Research, Advocacy and Support
www.cherubs-cdh.org 1-866-603-1944

March of Dimes Birth Defects Foundation
www.modimes.org 1-888-MODIMES (1-888-663-4637)

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