Diaphragmatic Hernia

BIRTH DEFECT RESEARCH FOR CHILDREN

What is a Congenital Diaphragmatic Hernia?

The diaphragm is a broad muscle that separates the abdominal and chest cavities. It's a major muscle involved with breathing. The diaphragm forms in babies at eight weeks of gestation. During development, two membranes merge to close the open area between the chest and the abdomen. When this membrane does not close completely, a Congenital Diaphragmatic Hernia (CDH) occurs, leaving a hole in the muscle of the diaphragm through which abdominal organs, such as the stomach and bowels, protrude into the chest.

Congenital Diaphragmatic Hernia

What are the different types of Congenital Diaphragmatic Hernia?

The most common type of diaphragmatic hernia is called a Bochdalek hernia. In this condition, the hole in the diaphragm may be as large as the diaphragm itself. Sometimes this type of diaphragmatic hernia is called a Posterolateral CDH. This hernia tends to allow many of the abdominal organs into the chest.

When the diaphragm is completely absent, the condition is called Complete Agenesis of the Diaphragm. Agenesis of the Hemidiaphragm means that one of the membranes fails to form and there is only half of the diaphragm.

A less common hernia, a Morgagni hernia, occurs near the front of the body near the breastbone.

How often does a Congenital Diaphragmatic Hernia occur?

Diaphragmatic hernia occurs in about 1 in every 2500 babies. The condition is more common in males than females.

What causes Congenital Diaphragmatic Hernias?

The cause isn't yet known. Sometimes the condition runs in families with other genetic problems and CDH is common with such syndromes as Fryn's Syndrome and Cornelia de Lange Syndrome. Studies also suggest that environmental factors like exposure to certain chemicals such as pesticides are associated with CDHs.

Can a Congenital Diaphragmatic Hernia be prevented?

Currently no means of prevention are known. Genetic counseling is recommended for syndromes involving CDH.

What are the chances that siblings will also have a Congenital Diaphragmatic Hernia?

If there is no family history of diaphragmatic hernia or other genetic abnormality in the baby, the chances of recurrence in siblings is approximately 2%.

When are Congenital Diaphragmatic Hernias usually detected?

CDHs can be diagnosed by ultrasound as early as the 12th week of pregnancy. If a CDH is detected, the pregnancy will be closely monitored and delivery can be planned at a specialized center for high-risk pregnancies. These facilities typically have a pediatric surgeon and ECMO (extra corporeal membrane oxygenation) both of which may be critical for treatment of the diaphragmatic hernia. ECMO is a heart and lung bypass machine that allows the lungs to rest and the baby to gain strength for surgery and recuperation.

In 50% of cases, CDH is not discovered until the baby is born. Children with severely restricted lung capacity will need immediate breathing support. In children with better lung capacity, the condition may not be detected for several weeks, months, or even years, perhaps by a chest x-ray taken for other reasons.

How is a Congenital Diaphragmatic Hernia treated?

The biggest problem caused by the herniation of the bowel is that the lung on the affected side and even the lung on the opposite side cannot develop to capacity. The extent of the lung deficiency becomes apparent at birth when the umbilical cord is clamped and the infant must oxygenate its blood by breathing.

The severity of the problem depends on the size of the hole in the diaphragm, how much of the abdominal contents have entered the chest, and at what point in gestation the herniation occurred. These factors determine lung capacity.

Congenital Diaphragmatic Hernia

Shortly after birth, the baby swallows air while crying; consequently, the bowel in the chest inflates with air and further compromises the insufficient lung. Children with severe lung deficiency require intubation and ventilation in the delivery room to survive.

Surgery is required to return the organs to the abdomen and to repair the diaphragm. There is also Nitric Oxide treatment to help the lungs expand and surfactant treatment to coat the lungs and give them more elasticity. In utero treament with Betamethazone Steroids can also enhance lung growth.

Timing of the repair depends on the patient's stability and the patient's need for ECMO or other treatments such as Nitric Oxide or Oscillating Ventilation.

Sometimes the defect is large enough to require synthetic material to close the diaphragm. This will keep the bowel out of the chest, but will not grow with the child and may lead to detachment of the graft in children with a large opening. Since graft material is a foreign body, there is a greater risk of infection.

Children with severe lung deficiency will be placed on ECMO to allow circulation of the blood through an external oxygenator pump. Timing of surgery for a baby on ECMO is at the discretion of the surgeon.

Some surgeons are now repairing CDHs in utero with a procedure call Tracheal Ligation that forces the lungs to grow and moves the organs back down into place. Complete repair of the hernia can then be done after birth.

What is the prognosis?

The most important problem the child has after surgery is attaining adequate lung capacity by gradual expansion of the deficient lung.

50% of children diagnosed with CDH do not survive. About 85% of treated children have feeding problems; 50% have asthma and 75% have reflux. Some have complications such as cerebral palsy, developmental delay, and a few have other severe problems caused by not receiving enough oxygen after birth.

Fact Sheet by:

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