

Congenital Diaphragmatic Hernia

What is congenital diaphragmatic hernia?

Congenital diaphragmatic hernia (CDH) is a condition in which all or part of the abdominal organs (stomach, intestine, liver and spleen) move into the chest cavity through a hole in the diaphragm. As a result, the lungs do not have room to grow or develop properly, causing the baby to have breathing problems after birth (a condition called pulmonary hypoplasia).

How common is it?

CDH is seen in approximately one in 2,500 births and usually can be detected during pregnancy. It can occur on either side of the chest cavity but is most common on the left.

How is it diagnosed?

Often, the presence of CDH is discovered through routine ultrasound between 15 and 20 weeks of gestation. A level II ultrasound is performed to confirm the CDH diagnosis and measure its severity, by assessing the size of the lungs and identifying the position of the liver—if the liver has moved into the chest, the condition is more severe. Often prenatal magnetic resonance imaging (MRI) is used to help clarify the position and size of the organs. MRI is painless and free of radiation, and so it does not affect the developing fetus.

What can happen before birth?

The biggest danger of CDH is that the lungs may be so underdeveloped that the baby is not able to breathe after birth.

What can be done before birth?

Most babies with CDH are best treated after birth; however, fetal intervention is possible for the most severe cases. The type of intervention depends on many factors, including the gestational age at initial diagnosis, the fetus' lung-to-head ratio (a measure of the size of the fetal lungs based on the amount of visible lung, also called LHR), the presence of part of the liver or stomach in the chest, and whether there is an excess of amniotic fluid. One option is fetal tracheal occlusion, a minimally invasive operation performed on the fetus that treats CDH by promoting natural lung growth. Here's how it works:

- **Fetal tracheal occlusion:** The fetal lung normally produces fluid that escapes through the mouth. When the trachea is blocked (or occluded) fluid builds up in the lungs, expanding them and pushing the other organs out of the chest and into the abdomen. In fetal tracheal occlusion, an inflatable and detachable balloon is inserted through a single entry port and filled with saline water to block the trachea. It is typically placed around 28 to 30 weeks of gestation and is removed around 34 to 35 weeks. In one large study (the Eurofoetus consortium), this procedure increased survival rates from 10 to 40 percent to 40 to 80 percent.

What are my delivery options?

Unless there are signs that the fetus is in trouble, pre-term delivery or Cesarean section is not necessary. Cesarean section may be necessary for obstetrical reasons, however.

The outcome of mild cases of CDH—when the lungs are nearly the proper size and the liver is completely in the abdomen—is often very good at birth. In severe cases, the baby may require aggressive resuscitation immediately. No matter the prognosis, it is recommended that mothers deliver in a tertiary center that has immediate access to a specialized neonatal intensive care unit (NICU) and to advanced technology, such as extracorporeal membrane oxygenation (ECMO), with a pediatric surgical specialist present.

What will happen at birth?

Babies with CDH often require the help of a respirator to breathe after birth. Serious cases may need to be treated with extracorporeal membrane oxygenation (ECMO), a lung bypass machine that allows gas exchange (taking in oxygen and giving off carbon dioxide) while the baby's lungs are resting. ECMO cannot permanently replace the lungs and can only be used for a few weeks—after that, there are significant risks such as long-term damage to the lungs, brain and other organs.

Once the baby's condition has improved (this may take up to a week or longer), he or she will undergo surgery to move the organs into the abdomen and repair the hole in the diaphragm. If the hole is large it may require an artificial patch. (It may seem strange to wait to close the hole that caused the condition; however, pulmonary hypoplasia is the true danger to the baby, making it essential to correct lung function first.) After surgery the baby will stay in the neonatal intensive care unit until full recovery. Many babies require respirator assistance for days to weeks after surgery, until the lungs have time to recover and improve their function.

What is the long-term outcome?

The long-term outcome depends primarily on how developed the lungs are at birth. The baby may need additional oxygen through a respirator for a while, even at home, and may need medication to help with breathing (not unlike children who have asthma). The baby may also require occupational therapy to improve muscle strength and coordination, especially if the baby has been very sick for several weeks and particularly if the baby has been treated with ECMO. The gastrointestinal tract (the esophagus, stomach and intestines) may not function perfectly because the organs developed in the chest, leading to feeding problems, difficulty gaining weight and gastroesophageal reflux. To get enough nutrients, babies sometimes need additional feedings through the assistance of a feeding tube.

CDH is also associated with birth defects, including those that affect the heart, kidney and central nervous system. Chromosomal defects (most commonly trisomy 21 [Down syndrome], 18 and 13) are found in approximately 10 percent of babies with CDH. 

