CDH: Prenatal

APSA American Pediatric Surgical Association Saving Lifetimes

CDH: Post Delivery

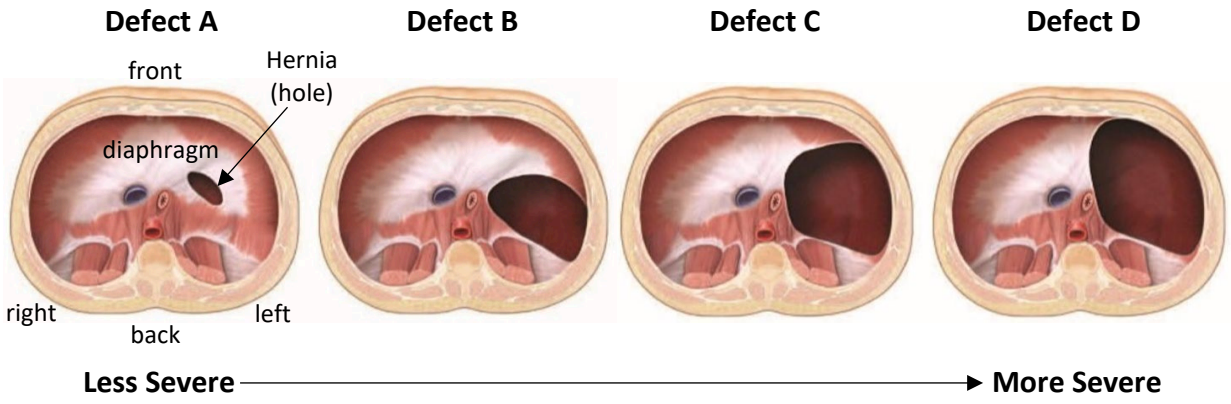
Prenatal Diagnosis of Congenital Diaphragmatic Hernia

Patient and family information, brought to you by the
Fetal Diagnosis and Treatment Committee & Education Committee of APSA

Overview – "What is it?"

Congenital Diaphragmatic Hernia (CDH) is a condition where a baby is born with a hole in the diaphragm. The diaphragm is the muscle that separates the chest from the abdomen. Normally, the heart and lungs are in the chest and the intestines and other organs are in the abdomen. When there is a hole in the diaphragm, organs from the abdomen can go through the hole and into the chest cavity. As a result, the lungs are smaller because they lack the space to grow. In addition, the blood vessels of the lungs are thicker, making it sometimes difficult for oxygen in the blood to get to the lungs, which is a condition called pulmonary hypertension.

The cause of this defect is not fully known. CDH occurs in about 1.9 in every 10,000 births. The size of the hole can vary. Larger holes are associated with more problems because more abdominal organs get into the chest and take up more space, which causes the lungs to be smaller. Sometimes, the lungs are so small that the babies are not able to breathe adequately after they are born, and they don't survive. Most diaphragmatic hernias occur on the left side (80%) and will often contain the stomach, spleen, small and large intestine. Those on the right side will often contain portions of the liver.



This picture illustrates the diaphragm looking from the abdomen upwards toward the chest. The image shows a left sided CDH with the diaphragm hernia (hole) in the most common location. However, CDH can also occur on the right side. In general, larger defects are more severe and are associated with a higher risk of breathing problems. *Image source: Pediatric Surgery NaT, Chapter on Congenital Diaphragmatic Hernia.*

Diagnosis and Evaluation – "What tests are done to find out what my child has?"

Obstetrical Ultrasound

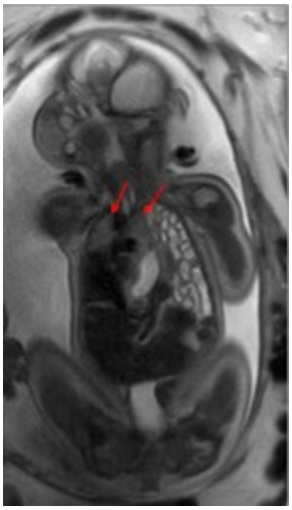
CDH may be diagnosed on a routine prenatal ultrasound at around 20 weeks gestational age by identifying abdominal organs (stomach, spleen, small and large intestines, or liver) located in the chest that should be located in the abdomen. Once identified, a more comprehensive ultrasound will be used to evaluate your child's growth, amniotic fluid, lung development, and to identify any other anatomical abnormalities.

Fetal Echocardiogram

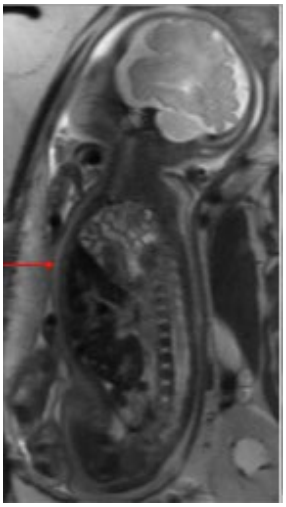
Once diagnosed with CDH, an ultrasound of your child's heart will be done to evaluate the position of the heart and to identify any major structural abnormalities. This study will also evaluate the blood vessels going to your child's lungs in order to determine the risk of developing pulmonary hypertension due to the CDH.

Fetal Magnetic Resonance Imaging (MRI)

After CDH is diagnosed, an MRI is often obtained at 24-28 weeks gestational age to better assess your child's anatomy. It will help determine the severity of CDH and can exclude other potential alternative or coexisting conditions. The MRI will also be used to evaluate your child's lung volumes to best prepare for your child's respiratory needs after delivery. In some instances, a late gestational (32-34 weeks) MRI will be obtained for delivery planning.



Fetal MRI showing stomach and small intestine in the left chest. As a result, both right and left lungs are pushed upward and smaller than normal.



Fetal MRI showing small intestine and part of the liver in the left chest.

Images courtesy of Kimberly Dannull, MD, Colorado Fetal Care Center

Amniocentesis

Amniocentesis is a procedure in which a sample of the fluid that is surrounding your child inside the uterus is removed for testing. Genetic testing is often recommended after the diagnosis of CDH has been confirmed because children with CDH may have genetic abnormalities that will affect their prognosis.

Prognosis – "What information is helpful to understanding my child's diagnosis?"

Understanding the severity of the CDH prior to delivery of your child is crucial for proper planning of the transition from fetal to postnatal life. It allows for team preparation and readiness. Moreover, it allows for access to some clinical trials.

Lungs

The lungs can be affected in two major ways as a result of CDH: 1) smaller size and 2) development of pulmonary hypertension. Because organs that should be in the abdomen are taking up space in the chest, there is less space for the lungs to develop to their normal size. Pulmonary hypertension also makes the delivery of oxygen more difficult. Both smaller lung size and more severe pulmonary hypertension are associated with worse outcomes in children with CDH. Lung size and severity of pulmonary hypertension can be assessed via obstetrical ultrasound, fetal MRI and fetal echocardiogram. However, while prenatal evaluation of pulmonary hypertension can help predict your child's breathing support needs after birth, its accuracy is still being researched. Therefore, children will need to be reassessed after they are born for pulmonary hypertension severity and breathing support needs.

One of the most important measurements to assess your child's lung development is the observed (what is seen) to expected (what it should be) Lung-to-Head ratio (O/E LHR). This measurement provides the relationship between your child's lung volume (on the opposite side of the CDH) and your child's head size, to what would be expected at the gestational age of your child at the time of measurement. O/E LHR is categorized into three levels of severity: mild (O/E > 35%), moderate (25-35%), or severe (O/E < 25%).

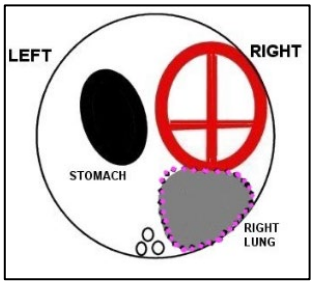


Diagram of how Lung-to-Head ratio is calculated. Red = Heart, Black = Stomach, Grey = Lung.

Image Source: Perinatology.com

Liver

In either left or right-sided CDH, the liver may be located in the chest instead of the abdomen where it normally is. How much the liver shifts into the chest is related to overall prognosis – more liver in the chest is related to worse outcomes. Liver location can be assessed via ultrasound and fetal MRI. Fetal MRI allows for evaluation of the degree of liver herniation.

Heart

Approximately 10% of children with CDH also have a structural abnormality of the heart. Congenital heart disease is associated with worse outcomes in children with CDH. Your child's heart will be assessed via fetal echocardiogram.

Other Congenital Abnormalities

Approximately 10-35% of children with CDH have genetic abnormalities. The implications of these abnormalities are highly variable. Certain genetic conditions are associated with worse outcomes in children with CDH. Additionally, genetic conditions may be associated with other anatomical abnormalities that will be important to identify for family planning and labor/delivery preparation.

Pregnancy and Delivery – "What happens now that we have a diagnosis?"

Prenatal Monitoring

Once diagnosed with CDH, more frequent obstetric ultrasounds will be performed to monitor your child. Ultrasounds will be performed approximately every four weeks until 30 weeks gestational age and then every other week until delivery.

Delivery

Early term delivery (37-38 weeks gestational age) is recommended when your child has a diagnosis of CDH. The goal is to have a planned, daytime delivery that will allow for optimal coordination of a multidisciplinary team including your pediatric surgeon, neonatologist, and any other specialists that may be needed. Delivery can be planned via elective Cesarean or an induction of labor followed by vaginal delivery depending on the severity of your child's CDH and what is best for the mother.

After Delivery – "What will happen once my child is born?"

What to expect after delivery largely depends on the severity of your child's CDH. Immediately after birth, your child will be evaluated by neonatologists and pediatric surgeons to determine the best course of action.

Hospitalization

The length of time your child will remain in the hospital depends on the severity of the CDH. Hospitalization typically ranges from one week to several months, with longer hospital stays (even up to a year or more) being necessary for more severe CDH. During this time, your child may be in the neonatal intensive care unit, may require breathing support (including a heart-lung machine), and will undergo an operation. Your multidisciplinary medical team will develop an individualized treatment plan for your child based on his/her needs.

Pulmonary (Lung) Support

Breathing support may be necessary and can range from a small amount of oxygen to mechanical ventilation depending on the size of your child's lungs and the severity of pulmonary

hypertension. Prenatal studies, such as the obstetrical ultrasound and fetal MRI, will help predict your child's support needs. It is important to know that surgical correction of CDH will help, but not cure your child's breathing problems. The amount of pulmonary support required may change throughout your child's hospital stay.

Mechanical Ventilation

A machine (ventilator) may be needed to help give oxygen and remove carbon dioxide to/from your child's body. A breathing tube will be placed to connect your child's lungs to the ventilator. There are several types of machines and modes of mechanical ventilation depending on the amount of support that your child requires.

Extra-Corporeal Life Support (ECLS, also known as ECMO)

In severe cases of CDH or if other measures of breathing support are not effective, your child may require ECLS. ECLS is a type of bypass for the heart and lungs. A procedure is required to connect the blood vessels in the neck to the heart-lung bypass machine. Once connected, blood will circulate through the machine, which will help move oxygen to other organs when the lungs are too underdeveloped to do so.

Preparation for Surgery

After delivery, your child's multidisciplinary medical team will focus on preparing your child for surgery. This may involve breathing support (as discussed above), nutritional support, as well as optimization of any other medical conditions your child has. Depending on the severity of your child's CDH, optimization for surgery ranges from twenty-four hours to several weeks.

Feeding

When and how your child can be fed depends on the severity of the CDH. In less severe cases, feeding can begin right away and continue while your child is being optimized for surgery. However, depending on the level of breathing support your child requires, direct feeding may not be possible and a tube may be placed from your child's mouth to their stomach in order to administer feeds. In more severe cases, your child may not be able to receive feeding the typical way (into the stomach), and may therefore receive nutrition via their veins – this is called total parenteral nutrition (TPN). In either situation, if you are planning to breastfeed, breast milk can be saved and given to your child once they are ready.

Treatment – "What will be done to make my child better?"

Prenatal Treatment – "Is this an option?"

Over the years, there has been significant progress in the development of procedures to promote lung growth while your child is still inside the uterus. Fetoscopic Endoluminal Tracheal

Occlusion (FETO) is a procedure that promotes stretch-induced lung growth by temporarily plugging the lung to minimize the negative effects of CDH on lung development. At this time, FETO is available only at highly experienced facilities. Determining if FETO is an option for your child is an individualized decision that should be discussed with your physician.

Newborn Management

Please refer to this Congenital Diaphragmatic Hernia resource for additional information about medical management and surgical treatment options of CDH.

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