MICHIGAN MEDICINE
UNIVERSITY OF MICHIGAN

FETAL DIAGNOSIS AND TREATMENT CENTER

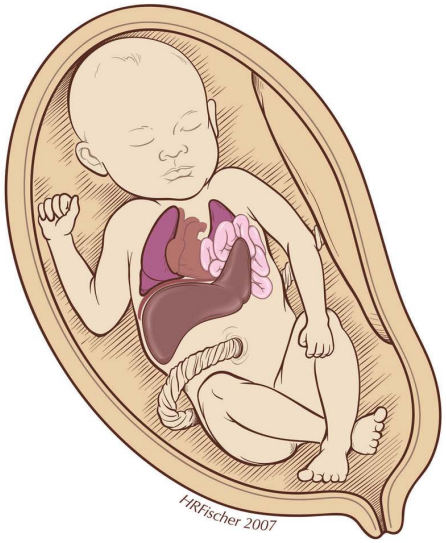
Congenital Diaphragmatic Hernia (CDH)

The diaphragm is the muscle that separates the chest and abdominal contents. A congenital diaphragmatic hernia (CDH) means there is a hole in the diaphragm. The hole permits the abdominal organs to move into the chest area before birth.

C.S. Mott Children's Hospital is home to one of the country's most experienced CDH centers. Our comprehensive experience with CDH includes fetal diagnosis and counseling, individualized delivery planning, postnatal surgical management, and long-term follow-up care. Patients are transferred to Michigan Medicine from throughout the state and from around the country, with support from ECMO-equipped Survival Flight air and ambulance service.

Congenital diaphragmatic hernia overview

In a child with CDH, the lung on the side of the hole is prevented from growing normally due to the abdominal organs having shifted into the chest area.



The lung on the opposite side is also smaller than expected. Small lungs can also be called pulmonary hypoplasia.

For children with CDH, breathing problems are common at birth. Also, the infant's small lungs have blood vessels that are not developed normally. These vessels are more likely to constrict, making it difficult to get oxygen to the lungs at birth. This causes high blood pressure in the lungs which is called pulmonary hypertension.

There is another problem with the diaphragm that can be similar to CDH called eventration. With an eventration, the diaphragm structure has more fibrous and elastic tissue and less muscle. This makes the diaphragm less effective separating the chest and abdominal contents. As a result, the abdominal organs can move the diaphragm allowing the organs into the chest area. Lung growth is limited on the affected side.

CDH can be isolated, meaning it is the baby's only major health problem. However, babies with CDH may have problems with other developing organs besides the lungs. The most commonly affected organs are the heart, the kidneys, the intestines, and the brain. Sometimes, CDH is seen with other major and minor findings. Together, these findings may be associated with a genetic syndrome which can impact the baby's ability to survive.

CDH occurs in about one in every 2,200 births. About 85% of these defects occur on the left side. CDH can also affect the right side of the diaphragm and, in rare situations, both sides. Overall survival of CDH babies in the United States is approximately 65 - 70%. At Michigan Medicine, however, survival rate is over 80%.

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Diagnosis of congenital diaphragmatic hernia

Ultrasound is first used to evaluate the defect in the diaphragm and to look for other associated anomalies. A measurement is taken with the ultrasound called a lung-to-head ratio (LHR). This considers the baby's lung size compared to the gestational age. This value is one factor that helps the team anticipate the degree of breathing problems the baby may have at birth.

A fetal echocardiogram (heart ultrasound) is used to assess the heart structures and its function.

Amniocentesis is recommended given the chance there could be a genetic problem. Fetal MRI is used to calculate lung volumes and reliably assess the degree of liver herniation. The lung volumes are used with other findings to assess the severity of breathing problems at birth.

Fetal treatment of congenital diaphragmatic hernia

Fetal Endoscopic Tracheal Occlusion (FETO) is a treatment reserved for a few babies with very severe CDH. Using a tiny fetoscope, a small detachable balloon is placed in the baby's trachea during the mid-second trimester. Since the fetal lung normally produces fluid, the balloon prevents this fluid from escaping the lungs. In turn, the gentle pressure from the fluid build-up causes the small lung to expand and grow. This treatment option is currently available at few centers in the United States. Michigan Medicine is one of these select FDA-approved centers offering the FETO procedure to fetuses with severe right- or left-sided CDH. The multicenter trial is looking at the effectiveness of this therapy.

Management of pregnancy

Ultrasounds are scheduled regularly to monitor the baby's growth and well-being. They are also used to check the amount of amniotic fluid around the baby. Extra fluid (polyhydramnios) can develop if the esophagus is compressed by the abdominal organs. If this happens, there is a concern for an early delivery.

Other antenatal testing may be used as the pregnancy continues.

Delivery of a baby with congenital diaphragmatic hernia

In general, delivery is planned as close to term as possible. A vaginal delivery is recommended for the majority of babies with CDH. A cesarean section delivery is reserved for obstetrical reasons.

Delivery of a baby with CDH should be planned at a hospital that is prepared for high-risk births, including immediate access to a newborn intensive care unit (NICU), pediatric surgical services and ECMO. This will simplify communication between obstetrical, neonatal and pediatric surgical teams as well as limit the separation between mother and baby.

Michigan Medicine is one of only a few centers nationwide at which the birth center is co-located within a comprehensive children's hospital. This unique setting allows for seamless integration between our private-room birth center, state-of-the-art NICU, and access to around-the-clock pediatric surgical services in one convenient location.

Delivery should be planned at a hospital with experience using ECMO (extra corporeal membrane oxygenation), a heart-lung bypass machine used to treat severe pulmonary hypoplasia and pulmonary hypertension. The machine provides oxygenated blood directly to the baby. The constricted blood vessels in the lungs are given time to relax while the lungs rest. This allows the baby to maintain normal oxygen levels until it can be transitioned to the ventilator. ECMO was developed at the University of Michigan, and C.S. Mott Children's Hospital remains an international leader in the application of ECMO on neonatal, pediatric, and adult patients.

In rare cases, for the most severely affected CDH babies, a special birth plan involving a procedure known as "EXIT-to-ECMO" may be performed. EXIT-to-ECMO is a procedure that transitions the baby from placental oxygenation to ECMO support. The potential advantage of this procedure is avoiding lung injury from high ventilator settings with a smooth transition from the womb to extrauterine life. Our ECMO team is on stand-by for birth of every severe CDH patient.

Postnatal treatment of congenital diaphragmatic hernia

All babies with CDH are at high risk of severe breathing problems due to the underdevelopment of the lungs. Managing any degree of breathing distress is the first and most important step in caring for these babies. A breathing tube is immediately placed after birth to assist the baby with breathing. A small tube is then inserted into the stomach to prevent air build up that could cause pressure on the lungs. Repair of the CDH is delayed until the baby is stable from a heart and lung standpoint.

Ventilation

A method called "gentle ventilation" is used to provide oxygen to the baby. The breathing machine (ventilator) is carefully set to prevent damage to the baby's delicate lungs. There are several different types of ventilators that may be used based on the baby's response to treatment. A gas called nitric oxide may be used if pulmonary hypertension is a big problem. This gas is used to relax the constricted lung vessels.

ECMO is offered if "gentle ventilation" fails to provide the oxygen the baby needs. As the baby improves, the rate on the ECMO machine is slowly turned down. The surgical repair may be performed during or after weaning from ECMO.

Surgery

In some stable patients, a minimally invasive approach may be possible (using tiny incisions and instruments). These smaller defects are repaired primarily with sutures. However, many babies require a standard abdominal incision to repair the diaphragm. The larger defects often require a special patch to complete the closure.

CDH babies have a smaller abdomen because the organs moved into the chest during fetal life. This sometimes makes it difficult to safely return all the organs to their proper place. If this occurs, a temporary silo (elevated sterile bag) is created on the abdominal wall. It allows for gradual reduction of the organs over the next several days.

Recovery

The C.S. Mott Children's Hospital NICU exemplifies patient and family centered care. You will be able to visit your baby at any time day or night. The staff will be available to support both you and your baby during this time. It is difficult to determine how long your baby will be in the hospital. The baby needs to learn all the normal things that were interrupted during medical treatment. Daily goals will be made for the baby in an effort to help him transition towards discharge. Targeted issues that will be addressed include breathing, nutrition (eating and weight gain), and motor skills.

Long-term care

Many babies with CDH have long-term health issues of varying degree. The severity of these problems is usually related to the severity of the CDH and neonatal illness. Some patients may have long-term breathing and lung problems. Patients with severe CDH may require oxygen at discharge. Patients with persistent pulmonary hypertension are followed by pediatric cardiology.

Feeding difficulties are common. Many babies need supplemental feeding through a tiny tube in their nose for a period of time. Those who continue to struggle with weight gain may need a second operation to place a gastrostomy (feeding tube placed into the stomach). Even with additional feedings, some babies still have trouble gaining weight. This is especially common during the first year of life. Gastroesphogeal reflux or "heartburn" is common in babies with CDH. It is treated successfully with medicine for most babies. Some babies do not respond to medicine and need a separate operation (Nissen fundoplication) to treat the reflux.

There are possible neurological problems that may occur. Hearing loss, developmental delay, and sometimes more serious neurological problems can be seen with CDH. These risks are greater in children requiring ECMO support. The surgical team will be following the baby with these concerns in mind.

Scoliosis and chest wall abnormalities may develop in some patients.

Close follow-up by a multidisciplinary team of specialists is essential to manage the long-term health issues in patients with CDH. Michigan Medicine's congenital diaphragmatic hernia clinic provides multidisciplinary follow-up care. The team includes a pediatric surgeon, pulmonologist, cardiologist, dietician, nursing, and physical and occupational therapists. Our comprehensive team of specialists works together to identify any long-term problems and facilitate early intervention.

Future pregnancy risk

The risk of CDH occurring in another pregnancy is rare. Most cases of CDH are random events. Since the cause is unknown, the chance of it happening again is thought to be about 2% with each following pregnancy. If the CDH is part of a genetic syndrome or chromosome anomaly, recurrence is dependent on that underlying cause.

Prenatal diagnostic testing may be available in future pregnancies if a genetic cause is identified. Your doctor and a genetic counselor will review the risk in your family.

Congenital diaphragmatic hernia research

The underlying cause of isolated CDH remains unknown. Molecular research has been ongoing to look for answers. The Michigan Medicine C.S. Mott Children's Hospital is participating in the DHREAMS study (Diaphragmatic Hernia Research & Exploration, Advancing Molecular Science) in an effort to identify specific alterations in genes that cause CDH and therefore be able to better diagnosis, prevent, and treat children with this disease.

Schedule an appointment

For more information or to schedule an appointment call 734-763-6295.