

# UNDERSTANDING CONGENITAL DIAPHRAGMATIC HERNIA

CARMEN AND JOHN THAIN CENTER FOR PRENATAL PEDIATRICS

## What is congenital diaphragmatic hernia?

Congenital diaphragmatic hernia (CDH) is a birth defect caused by the absence of, or a hole in, the diaphragm, the muscle that separates the chest cavity and the abdominal cavity. The organs normally located in the abdomen (including the stomach, intestines, liver, and spleen) can move up through that hole and enter the chest. These organs then press against the lungs and prevent them from developing properly, so a baby with CDH has a lot of difficulty breathing after birth. CDH is most common on the left side of the body (in over 90% of cases), but can sometimes be on the right side or on both sides as well.

## How common is CDH and what causes it?

CDH is seen in 1 in 3000 to 1 in 5000 live births and makes up 8% of all major birth defects. The muscles of the diaphragm normally develop between the 7th and 10th week of pregnancy, and for some reasons the muscle does not close completely in babies with CDH. The cause of this abnormality is largely unknown, though up to 40% of CDH babies have other birth defects, most commonly affecting the developing heart, kidneys, and intestinal systems. Up to 20% of CDH cases also have a chromosomal abnormality such as Down syndrome.

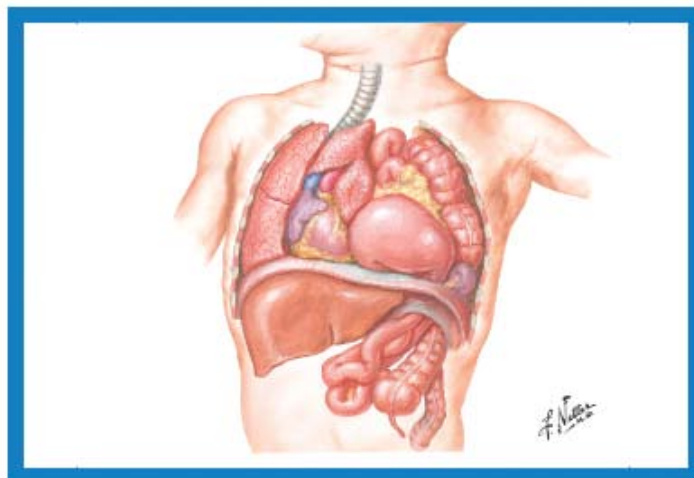
## How is CDH detected during pregnancy?

CDH is generally found through ultrasound performed in the second trimester. CDH is suspected when the stomach is not seen in its normal location and bowel appears next to the heart. Right-sided hernias may be harder to detect on ultrasound, since often the liver is the only organ that moves into the chest and appears very similar to the lung on ultrasound.

Other testing can be helpful to confirm the diagnosis of CDH. These include fetal echocardiogram (ultrasound targeted to the developing heart) and fetal MRI, which can better visualize the developing organs and help predict the severity of the hernia. An MRI image of a baby with CDH is included above.

## How will my pregnancy be managed now that CDH has been detected?

Prenatal care should be managed by a Maternal-Fetal Medicine specialist (MFM), an obstetrician with special training and expertise in high-risk pregnancy. A fetal echocardiogram will be performed by a Pediatric Cardiologist to check the heart and how the CDH has impacted its development. Furthermore, a procedure called amniocentesis may be offered to you to examine the baby's chromosomes, since babies with CDH have a higher chance to have a chromosomal abnormality.



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Several ultrasounds will likely be performed over the course of your pregnancy to closely monitor the baby, evaluate the severity of the hernia, and check the amniotic fluid so that polyhydramnios, an excess of amniotic fluid, does not develop. Since the severity of the CDH is determined by the damage to the developing lungs, the ultrasound will carefully examine the lungs and calculate a lung:head ratio (LHR). LHR is a good predictor of how severely affected the lungs are, and can be used to determine how serious the CDH may be; the lower the LHR value, the more severe the CDH. The ultrasound and the MRI will also look to see if the liver has moved into the chest, because this also helps predict the severity of the condition.

Before your baby is born, you will also meet with other specialists who will help inform and guide you in anticipation for the baby's arrival. These include a consultation with one of our Pediatric Surgeons, a consult with a Neonatologist (a pediatrician specially trained to take care of babies in the intensive care unit after birth) and a tour of the Neonatal Intensive Care Unit (NICU), and a Genetics consult. If the prenatal testing shows the baby may have other birth defects, your Clinical Care Coordinator will discuss how they will be treated after the baby's birth and potentially arrange for you to meet with the other specialists.

As your delivery date approaches, induction of labor or a cesarean section will likely be planned to optimize the delivery and ensure that the necessary specialists are available to care for the baby immediately after birth. However, labor management does not need to be changed because your baby has CDH.

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### How will the CDH be treated after birth?

After the baby is born, the diagnosis of CDH will be confirmed and the baby will be stabilized. Since babies with CDH are unable to breathe on their own because their lungs are underdeveloped, they are placed on a mechanical ventilator to help them do so. In the more severe cases of CDH, a bypass machine known as ECMO may be necessary to do the work of the heart and lungs until the baby is stable and can tolerate surgery. Only certain specialized centers in the United States have ECMO.

Once the baby is stable and can handle surgery (often days to weeks after birth), the abdominal organs that have moved into the upper chest are moved back in the abdomen into their normal location. The hole in the diaphragm is closed as well. The baby will remain on the ventilator to help breathing, and certain medications, called surfactants, may help the baby to eventually be able to breathe on his or her own. Most babies with CDH will be in the NICU for days after the operation. Once the baby is more capable of breathing without a ventilator, he or she can usually go home. However, babies with CDH will still need regular follow-up and several checkups after leaving the hospital.

### What is the long-term outlook for babies with CDH?

The long-term outcome for babies with isolated CDH is related to how severely the developing lungs were impacted by the hernia during the prenatal period and how long ventilation was necessary to help breathing in the newborn period. Infants with CDH and additional birth defects or a chromosomal abnormality have a much lower chance for survival than babies with only CDH.

There are some long-term problems that can be associated with CDH. Up to 60% of infants with CDH experience gastroesophageal reflux (when stomach acid leaks back into the esophagus), which can be treated with medications or surgery. Between 20-30% of babies with CDH will experience some neurologic issues, such as seizures and developmental delay. More severely affected babies may have difficulty getting the calories they need to grow, so a feeding tube may be required in order to provide their necessary nutrition.

The severity and therefore the long-term outlook for babies with CDH are variable, so the specific expectations for your baby can only be determined by the medical team caring for him or her. Research is ongoing to improve treatment and outcomes for infants with CDH, and fetal surgery remains on the horizon.

### What are the chances I could have another baby with CDH?

The chances of having another baby with isolated CDH are around 2%. If the CDH is part of a genetic condition the chances for another child with that same condition vary. Speak with your Genetic Counselor and Geneticist about your specific chances to have another baby with CDH.

### What can I expect from the specialists at Morgan Stanley Children's Hospital?

The specialists at NewYork-Presbyterian Morgan Stanley Children's Hospital/Columbia University Medical Center are among the most experienced with CDH in the United States. Morgan Stanley Children's Hospital was the third in the world to successfully support a newborn on ECMO, and thousands of infants have been treated by our Pediatric Surgeons as potential ECMO patients. Most of these infants are doing well and living at home.

The Neonatal Intensive Care Unit (NICU) at Morgan Stanley Children's Hospital has an international reputation for excellence in respiratory care, especially important to babies born with CDH. The frequency of chronic lung disease in the premature infants treated in our NICU is the lowest in the United States.

Lastly, the MFM team dedicated to optimizing your prenatal care has extensive experience in managing pregnancy with CDH. Over 50% of the deliveries at Morgan Stanley Children's Hospital are high-risk, and since the Carmen and John Thain Center for Prenatal Pediatrics was founded we have successfully managed cases of CDH through the prenatal period, delivery, and treatment.

The combined efforts of our medical team have boosted the survival rate for babies with CDH considerably. While the general mortality rate for babies with CDH is believed to be around 50%, Center patients diagnosed with CDH boast a survival rate of 85%.

The well-being of you and your baby are extremely important to everyone involved in your care. Together we are all dedicated to giving you the best pregnancy and healthiest outlook for your child.

### About the Carmen and John Thain Center for Prenatal Pediatrics

Complex pregnancies receive better care when specialists collaborate. The Carmen and John Thain Center for Prenatal Pediatrics is dedicated to helping pregnant women and their families when a birth defect or genetic syndrome is detected before the baby is born. The Center offers sensitive, complete, up-to-date information and testing, and an integrated approach to care that begins in the prenatal period and continues after birth with pediatric follow-up. A collaborative, coordinated program of care is created among specialists in perinatology, neonatology, genetics, pediatric cardiology, pediatric surgery and all pediatric subspecialties.