Let's Talk About...

Prenatal Counseling: Congenital diaphragmatic hernia (CDH)

What is a congenital diaphragmatic hernia (CDH)?

A **congenital diaphragmatic** [DY-uh-fruh-MAT-ick] **hernia (CDH)** is an abnormality in the diaphragm (breathing muscle) that causes an opening between the abdomen (belly) and chest. This allows the belly organs (stomach, small intestine, spleen, liver, and kidneys) to move into the chest.

A CDH usually occurs on the left side but can happen on the right or, rarely, both sides. It is rare and affects about 1 in 3,000 babies each year. CDHs range from mild to life-threatening.

The **Utah Fetal Center team** will help you make the best possible decisions about your baby's CDH. The team includes maternal-fetal medicine specialists for the pregnant mother, neonatologists who are specially trained to care for newborns, and pediatric specialists to help with your baby's needs before and after delivery.

What causes a CDH?

Doctors don't know what causes a CDH, but it occurs during early fetal development. Nothing the mother does or has done during pregnancy causes this problem.

How is a CDH diagnosed?

A healthcare provider usually finds a CDH during a pregnancy ultrasound around 20 weeks into the pregnancy.

After they see the defect, they'll recommend more tests, including:

- **Fetal MRI.** This imaging test shows doctors where the liver is located in the abdomen or chest and helps determine how the baby's lungs are affected by the CDH.
- **Fetal echocardiogram** [ek-oh-CAR-dee-oh-gram] **(ECG).** This heart ultrasound lets providers see the heart's structure and function. Babies with CDH have an increased risk of heart problems.
- **Comprehensive ultrasound.** This imaging test provides more detail about your baby's other body structures that may be affected.
- **Genetic testing.** 1 in 10 babies with CDH have another genetic (passed down through families) problem, often congenital heart disease. A genetic counselor will tell you more about screening tests.

These tests help the fetal team determine the CDH's severity and how much it may affect your baby.

How does a CDH affect my baby?

A CDH can cause mild to severe heart and lung problems because the belly organs are in the chest, pressing on the heart and lungs. These organs prevent the lungs from developing properly, which can cause:

- **Pulmonary** [pul-moh-nehr-ee] **hypoplasia** [HI-po-PLAY-zee-uh]. Small, underdeveloped lungs
- **Pulmonary** [pul-moh-nehr-ee] **hypertension** [HI-per-TEN-shun]. Increased blood pressure in the blood vessels of the lungs

When your baby has small lungs and increased pulmonary blood pressure, it's hard for them to get enough oxygen while breathing. This puts a strain on your baby's heart.

These findings can only be diagnosed after your baby is born.

How severe is my baby's CDH?

A CDH can cause mild to very serious heart and lung problems depending on the CDH's location and severity. Healthcare providers use several tests to predict how the CDH will affect your baby, including:

- **Liver position.** Fetal ultrasounds and MRIs show the liver's position.
  - **Intrathoracic** [IN-tra-tho-RASS-ick], or liver up. Babies have a poor outcome and lower survival rate (about 4 in 10 babies survive). 8 in 10 babies need ECMO, or extracorporeal [EX-truh-cor-POR-ee-ul] membrane oxygenation [OX-ih-jen-AY-shun]. ECMO is heart and lung life support.
  - **Intraabdominal** [IN-tra-ab-DOM-in-ull], or liver down. Babies have a better outcome, and only 1 in 4 babies need ECMO. They have a higher survival rate (9 in 10 babies survive).
- **Ultrasound lung-to-head ratio (LHR).** During the fetal ultrasound, the radiologist takes measurements of the lungs and head. Specifically, they measure the lung area opposite the CDH at the heart level and then divide by the head circumference [sir-CUM-fer-ence] (length around the head).
  - **LHR less than 1.** Babies have poor outcomes, higher ECMO need (10 of 10 babies need it), and a lower survival rate (6 in 10 babies survive).
  - **LHR greater than 1.4.** Babies have better outcomes, lower ECMO need (4 in 10 babies need it), and a higher survival rate (8 in 10 babies survive).
- **Observed-to-expected total fetal lung volume.** After the fetal MRI, the radiologist measures your baby's fetal lung volume (observed). Then the radiologist divides that number by the fetal lung volume of a baby the same age who doesn't have CDH (expected). This leads to a percentage.
  - **Greater than 25%.** Highest risk, higher ECMO need (7 in 10 babies need it), and lower survival rate (2 in 10 babies survive).
  - **25 to 35%.** Intermediate risk, lower need for ECMO (4 in 10 babies need it), and overall higher survival (7 in 10 babies survive).
  - **Greater than 35%.** Lowest risk, lowest ECMO need (2 in 10 babies need it), and highest survival rate (8 in 10 babies survive).

How is CDH managed during pregnancy?

You should have early evaluation at the fetal center to determine the CDH's location and severity. Fetal ultrasounds allow healthcare providers to see liver position and LHR and decide if your baby qualifies for fetal therapy. The fetal team will tell you more about the CDH, recommend ways to manage your pregnancy, and help you meet the specialists who will care for your baby after delivery.

While the Utah Fetal Center does not currently offer it, a study for a fetal CDH surgery called fetoscopic endoluminal [en-do-LOO-min-ul] tracheal [TRAY-key-ul] occlusion (FETO) is currently underway at some fetal centers across the country. Your healthcare provider can discuss the trial requirements and decide whether to refer you to a center that offers the surgery.

How is CDH managed during and after delivery?

You should deliver your baby at the University of Utah Hospital. A pediatric surgeon can evaluate your child in a neonatal intensive care unit (NICU) that specializes in CDH care.

A neonatologist [NEE-oh-nay-TAHL-oh-jist], a specially trained pediatrician, will manage your baby's medicine, feeding, and daily needs in the NICU. They will make sure your baby's heart and lungs are working right.

What care does my baby receive in the NICU?

Your baby will receive care in the NICU until the doctor feels they are healthy enough to have the CDH repaired. The first priority is supporting the heart and lung function. Your baby may need oxygen to help them breathe or may even need a breathing machine (ventilator). They may also need:

- **An OG tube.** A nurse inserts an orogastric [or-oh-GAS-trick], or OG, tube in your baby's mouth all the way to the stomach. This tube sucks out any fluid in the stomach to prevent your baby from choking or breathing stomach contents into the lungs. It also prevents the intestines from becoming too dilated (enlarged) with air.
- **A PICC line.** A peripherally [per-IF-er-uh-lee] inserted central catheter (PICC line) is a tiny tube inserted through the umbilical cord and then an arm or leg. Your baby will receive fluids, antibiotics, other medicines, and nutrition through the PICC line.
- **TPN:** Your baby won't be allowed to eat at first, so they'll receive total parenteral [pah-REN-ter-ull] nutrition, or TPN, through the PICC line. TPN contains protein, fat, sugar, vitamins, and minerals and will meet all your baby's nutritional needs.
- **ECMO:** About 3 in 10 babies with a CDH will need ECMO. This is a machine that works for your baby's heart and lungs so they can rest, heal, and develop. VV-ECMO supports the lungs, and VA-ECMO supports the lungs and heart.

How is the CDH repaired?

CDH repair options may include:

- **Primary repair.** Sewing the hole in the diaphragm closed (if the opening is small enough)
- **Patch repair.** Using a piece of tissue or material to create a patch over a larger hole in the diaphragm
- **Muscle flap repair.** Closing the diaphragm hole with a piece of muscle from your baby's back
- **Thoracoscopic** [thor-AS-oh-skah-pik] **surgery.** Using a camera and small openings to repair the diaphragm

The pediatric surgeon will discuss all of these surgical options with you and help determine which repair option is best for your child.

How long will my baby stay in the NICU?

You should anticipate a long hospital stay. Even a low-risk baby spends an average of 4 to 8 weeks in the NICU. A high-risk baby's average NICU stay is 4 to 6 months.

After the CDH is repaired, it takes time to slowly turn down the breathing machine and medicines, start feedings, and help your baby get enough nutrition to go home. About 1 in 10 babies will need another procedure before discharge.

What outcome can I expect for my baby after surgery?

The outcomes for babies with CDH can vary. Some issues that may complicate your baby's health include:

- **Abnormal chromosomes** [CRO-mo-zomes] (the baby's DNA or genetic blueprint is not normal)
- **Severe genetic problems** (health problems passed down through families)
- **Right-sided defect** (CDH is on the right side of the diaphragm)
- **Liver herniation** [her-nee-AY-shun] (liver is coming up through the diaphragm into the chest cavity)
- **Lower fetal lung volume**

If your baby needs a ventilator or ECMO support for a long time, they have a higher risk of neurologic (brain and nerve) problems, including motor (movement), sensory (feeling), learning, and hearing issues.

Your baby will need long-term follow-up with a pediatrician to watch their growth, development, lung function, and nutrition. They may also need long-term care from lung, brain, heart, and digestive tract specialists.

Some children need home health nursing and physical, occupational, speech, and nutritional therapy. The discharge planners and NICU social workers will help arrange this follow-up care with you before you take your baby home.

For additional information

Utah Fetal Center
intermountainhealthcare.org/locations/primary-childrens-hospital/medical-services/utah-fetal-center

Centers for Disease Control and Prevention (CDC)
cdc.gov/ncbddd/birthdefects/diaphragmatichernia.html

American Pediatric Surgical Association (APSA)
eapsa.org/parents/conditions/a-e/congenital-diaphragmatic-hernia

CHERUBS
cherubs-cdh.org

Questions for my doctor

Intermountain
Primary Children's Hospital
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