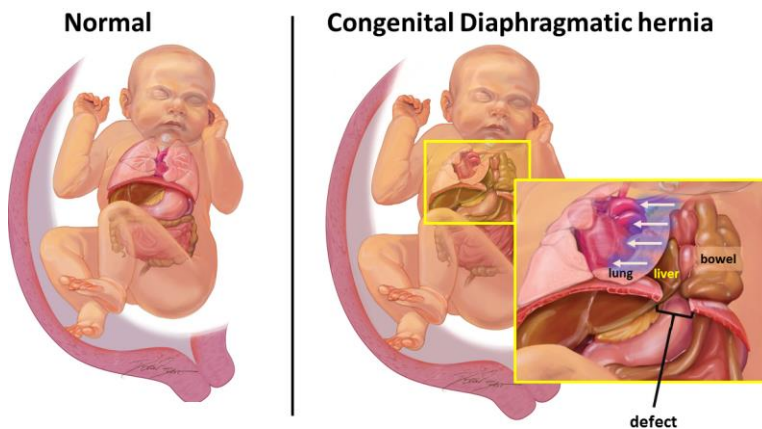


Congenital Diaphragmatic Hernia

Important things to know

What is congenital diaphragmatic hernia?

The diaphragm is a thin sheet of muscle that separates the chest from the abdomen. In the growing embryo the diaphragm is completely formed by 10 weeks gestation. Approximately 1 in 2500 fetuses have a birth defect where the diaphragm is not completely formed. The opening in the diaphragm may allow contents from the abdomen including bowel, stomach or even the liver to enter the chest. This is called congenital diaphragmatic

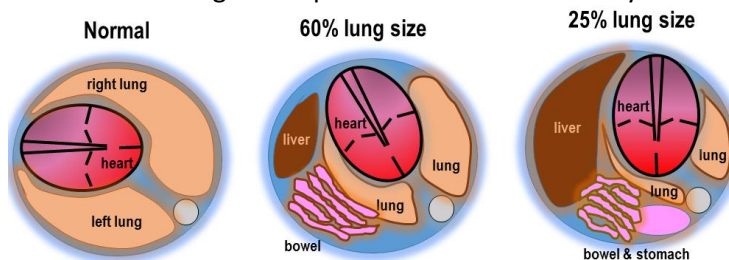


hernia or CDH and it can occur on the left side, the right side or rarely on both sides of the diaphragm. After birth the hole in the diaphragm can be closed without difficulty. However, in fetal life the abdominal contents that enter the chest compress the lungs. This interferes with normal lung development and can result in underdevelopment which is called pulmonary hypoplasia. Before birth the fetus does not need to use the lungs because the placenta provides oxygen and nutrients. It is after delivery that the baby relies on its own lungs to breathe. If lung development is severely

affected, a baby with CDH may not be able to survive without intensive care and specialized treatments, such as extracorporeal membranous oxygenation (ECMO). However, if the pulmonary hypoplasia is too severe the lungs may never be able to provide enough oxygen for the baby to survive. This is called lethal pulmonary hypoplasia. One of the major complications of CDH is the risk for lethal pulmonary hypoplasia. Babies with CDH may also have other structural or genetic problems. A detailed ultrasound and genetic testing can help determine if this is your baby's only problem.

What are the chances that my baby will survive?

The survival chances depend on lung development and if there are any additional structural or genetic abnormalities. When CDH is identified by prenatal ultrasound a detailed examination of the fetus and genetic studies are required to determine if the defect is isolated or if additional abnormalities are present. The impact of the hernia on lung development can be estimated by measuring fetal lung size using ultrasound or magnetic



resonance imaging. The ratio of lung and the fetal head circumference (**lung-to-head ratio = LHR**) or the comparison of the observed to the expected size of the lung at each gestational age (observed/expected lung-to-head ratio = o/e LHR) are effective methods to predict the survival in CDH.

Based on the LHR or o/e LHR and position of the liver, CDH can be divided into severe, moderate and mild categories evaluating the residual lung size. The survival rates are better in the mild and moderate categories especially if managed at a center that is experienced in the care of babies with CDH before and after birth. At Johns Hopkins Hospital, the overall survival of babies with CDH is well above the national average of 68%. The assessment of severity before birth is also used to decide who may benefit from fetal treatment to improve survival event more. All families that are planning to deliver a baby with CDH at Johns Hopkins Hospital meet with both the Neonatology and Pediatric Surgery teams during pregnancy.

Can my baby be treated?

Under normal circumstances the lungs produce fluid that can escape through the windpipe (**trachea**). Occlusion or blocking the trachea prevents escape of this fluid and stretches the lung tissue. This increase can stimulate lung growth. Extensive research has shown that fetal **tracheal occlusion** improves lung development in CDH. A technique has been developed where a small instrument with a miniature camera is used to look inside the uterus to insert a balloon into the trachea of the fetus. This procedure is called fetoscopic tracheal occlusion or **FETO**. This treatment is typically performed between 26-29 weeks gestation. A second procedure after 32 weeks is required to remove the balloon. After successful completion of these procedures the baby can deliver vaginally. Because it is an invasive procedure, FETO is only performed for babies with severe CDH and no other structural or genetic problems.

How is FETO performed?

This minimally invasive fetoscopic procedure is performed under epidural or local anesthesia. Prior to FETO, medication will be given to your baby for pain relief and also to prevent fetal movement. Then the fetus is gently moved into the best position. The fetoscope is introduced through the skin into the uterus and guided into the mouth and trachea of your fetus. When the fetoscope has been directed into the trachea, a small catheter is used to guide the balloon to its proper placement site. Once this is achieved, the balloon is inflated with fluid and left in the trachea. The balloon remains in the trachea of the fetus until 32-34 weeks gestation. During this time your fetus will be monitored with ultrasound. Since the placenta provides the oxygen for the fetus the balloon does not interfere with fetal breathing. Prior to birth however the balloon must be removed in order for the baby to be able to breathe. The balloon can be removed in several ways. While pregnant a second fetoscopy may be performed. Alternatively, the balloon may be punctured under ultrasound guidance using a needle. Once the balloon is removed you can deliver normally. If the baby is born before the balloon is removed, puncture can be performed after/at birth using a needle or by direct visualization of the balloon (tracheoscopy). Because of the special expertise needed, delivery in a tertiary care center with experience in the care of babies with CDH is important.

What are the risks for my baby and me?

Since FETO is a minimally invasive procedure, the risks for the mother are small. Ultrasound is used to help avoid blood vessels and the placenta. The main risks are rupture of membranes or going into labor while the balloon is still in place. Under these circumstances, our dedicated FETO team performs emergent removal. Because this must be done quickly, patients receiving FETO must stay close to Johns Hopkins Hospital while the balloon is in place. The balloon causes some bulging of the trachea, but this has not been shown to have any adverse effects and generally resolves after birth.

What are the alternatives?

You may choose to have no intervention or if your baby is not eligible for FETO, the pregnancy needs to be monitored closely with regular visits and ultrasound examinations. Your baby will need to be delivered at a tertiary care hospital experienced in the care of babies with CDH.