



## CONGENITAL DIAPHRAGMATIC HERNIA\*

\*This information is intended to supplement your consultation with members of the Fetal Treatment Program regarding your unborn child with a suspected diaphragmatic hernia. The same information is also available on our web site: [www.fetal-treatment.org](http://www.fetal-treatment.org). Technical terms are explained in a glossary at the end.

### WHAT IS CONGENITAL DIAPHRAGMATIC HERNIA (CDH)?

Congenital diaphragmatic hernia (CDH) refers to a defect in the diaphragm that allows the abdominal organs (stomach, intestine, liver, spleen) to move into the chest cavity. Congenital diaphragmatic hernia can occur on either the left or the right side but is most common on the left.

### WHY WORRY ABOUT DIAPHRAGMATIC HERNIA?

Babies who have CDH suffer from small and underdeveloped lungs. This is termed pulmonary hypoplasia. CDH allows the intestines to move up into the chest cavity. It is believed that this prevents the heart and the lungs from developing properly (figures 1 and 2). CDH is a life-threatening illness. When the lungs of a baby do not develop properly during pregnancy it can be difficult for the baby to breathe after delivery. The lungs of babies born with CDH are hypoplastic. The alveoli that are present are much more fragile and are missing surfactant. In comparison, normal lungs at birth have millions of small alveoli, which can best be thought of as many, many clusters of grapes. When babies are born with CDH and hypoplastic lungs, there are not enough alveoli to get oxygen in the blood, or to get rid of carbon dioxide from the body. When this occurs, the lack of oxygen leads to severe illness requiring very aggressive supportive measures.

**We are currently conducting a pilot program, thanks to an Investigational Device Exemption from the Food and Drug Administration (FDA), whereby fetal tracheal occlusion using minimally invasive techniques will be offered to patients with the most severe form of diaphragmatic hernia. For more information, contact our coordinator at [coordinator@fetal-treatment.org](mailto:coordinator@fetal-treatment.org).**

### WHY DOES CDH OCCUR?

CDH occurs approximately in 1 of 2,500 births and can be usually detected before birth. The specific cause of CDH is unknown but is most likely multifactorial in origin. While certain drugs or environmental exposures have been questioned, no specific agent has ever been identified. Many parents are concerned that it was something genetic that they passed on to their baby; however, fewer than 2 percent of CDH cases are believed to be familial. Most cases of congenital diaphragmatic hernia and pulmonary hypoplasia are sporadic and isolated events.

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Research into the cause and treatment of diaphragmatic hernia is being performed in a number of centers around the country and around the world, including our own. Our laboratory is actively involved in trying to understand the mechanisms involved in normal and abnormal lung growth. All these efforts may ultimately help find a cure for this condition. Additionally, our Fetal Treatment Program is actively involved in counseling parents about prenatal options and helping to plan delivery and treatment of the baby after birth.

The most common type of CDH is a hole in the back of the diaphragm, also called Bochdalek hernia. The left side is affected approximately 85 percent of the time. This type of hernia is the gravest type, and the left chest usually contains the stomach, intestines, part of the liver and the spleen. This causes severe pulmonary hypoplasia of the left lung and significant hypoplasia of the right lung as well.

When it occurs on the right side of the diaphragm, a significant amount of liver may end up in the right chest, causing right lung hypoplasia and affecting the function of the heart.

A second type of diaphragmatic hernia is called the Morgagni hernia. This is a less common defect that involves a hole in the front portion of the diaphragm, just behind the sternum. Sometimes the intestines or a piece of liver may move up through this defect in between the heart and the sternum, but this usually does not cause pulmonary hypoplasia.

## I AM TOLD MY FETUS HAS DIAPHRAGMATIC HERNIA - NOW WHAT?

Prenatal diagnosis is important in that it allows for patient/parent education, identification of those babies at risk for the worst outcome and the opportunity to provide prenatal intervention if possible.

During routine prenatal care, around 15-20 weeks gestation, an ultrasound may demonstrate the existence of a congenital diaphragmatic hernia. At that point, the family may be referred to our Fetal Treatment Center for workup of a more detailed diagnosis. Often, a level II ultrasound will be performed to confirm that it is a CDH. That very detailed sonogram will look for the abdominal contents in the chest, identify the position of the liver in the abdomen or in the chest and try to assess the size of the lungs. Many times magnetic resonance imaging (MRI) is needed (figure 3). Prenatal MRI may have many benefits over conventional ultrasound imaging and can provide additional information regarding the prognosis. MRI is painless and does not affect the developing baby, since no radiation is used.

When the diagnosis of CDH is suspected and a work up is arranged, a consultation with maternal-fetal medicine specialists, pediatric surgeons and neonatologists is arranged. Prenatal counseling involves a multidisciplinary team that also includes geneticists and genetic counselors, radiologists and other specialists. The counseling will include details about the nature and the outcome of the diagnosis and the possibility of prenatal intervention. Additionally, a plan for the remainder of your pregnancy and delivery of your baby will be discussed.

## FETAL INTERVENTION AT THE FETAL TREATMENT PROGRAM

It is important to know that the treatment of babies of with diaphragmatic hernia after delivery has improved significantly over the past 15 years. Most babies can be treated after delivery at a CDH center that can provide specific care for your baby with diaphragmatic hernia. The survival rate for most babies is currently approximately 70 percent. Because the survival rate historically has been much lower, pediatric surgeons lead a search to prevent or correct the pulmonary hypoplasia before birth.

The first attempts at fetal surgery for CDH date back to the 1980's, when large animal experiments of open fetal surgery were developed. In the 1990's the first human cases of open fetal surgery were attempted and reported in the New England Journal of Medicine. The initial results of the prenatal therapy to repair the underdeveloped lungs were not as good as expected, because of the extreme

aggressiveness of this type of operation on the mother and the baby. Since, in the mean time, treatment after birth became more successful, "open" fetal surgery for CDH was abandoned.

Techniques of correcting these defects inside the uterus have evolved as well, however. Open fetal surgery for CDH has been replaced by fetal tracheal occlusion. Experimentally, this has been shown to result in accelerated growth of the hypoplastic lungs. Our laboratory, as well as others, have been very active in the last decade to further understanding the mechanisms involved in this accelerated lung growth following fetal tracheal occlusion.

Fetal intervention for CDH continues to evolve. Performance of occlusion can be performed through a single entry port technique. This technique (developed at our institution) allows occlusion of the fetal trachea with an inflatable and detachable balloon. Most recently, a multicentric study performed in Europe (Eurofoetus) has compared treatment for CDH with the single port technique to standard treatment after delivery. These results were very informative and encouraging.

It may be possible to identify a specific subgroup of babies with CDH in whom the survival can be predicted to be less than 10-40 percent despite all the current methods of postnatal therapy (including ECMO, gentle ventilation strategies, permissive hypercapnia, nitric oxide). In this group of severe CDH, fetal endoscopic tracheal occlusion increased this survival to 50-80 percent (Eurofoetus study). Fetal tracheal occlusion resulted in an increase in the lung growth based on follow up prenatal ultrasound measurements of the lung-to-head ratio (LHR)

## CURRENT RATIONALE FOR PRENATAL TREATMENT OF SEVERE CDH

Currently the majority of babies with CDH are best treated after birth. The choice of therapy is in part dictated by the presence or absence of known predictive factors. These include the gestational age at initial diagnosis (less than 25 weeks is a poor prognostic indicator), the presence of part of the liver in the chest (poor indicator), the presence of polyhydramnios (too much amniotic fluid), presence of the stomach into the chest and the lung-to-head ratio (LHR), which is one of the more reliable indicators of prognosis. Another indicator is the observed/expected lung volume (O/E), also expressed as the percentage predicted lung volume (PPLV): based on MRI images of the fetal chest cavity, it is possible to predict how large the lungs should be, and how large they actually are. The difference between these two values is the O/E, or PPLV.

For the most severe cases of CDH, extracorporeal membrane oxygenation (ECMO) is likely to be required. ECMO is a lung bypass machine that allows for gas exchange while the baby's lungs are resting and maturing. In the babies with the worst cases of pulmonary hypoplasia, however, ECMO can only be used for a few weeks and does not allow enough time for the lungs to grow or mature to live. Additionally, ECMO carries significant risks including long term damage to the lung, brain and other organs. Based on all the available literature, including reports on experimental pulmonary hypoplasia and tracheal occlusion as well as our own experience with experimental and clinical fetal surgery, the Fetal Treatment Program at Brown/ Hasbro/Women & Infants has developed a management plan for severe CDH.

Currently most cases of prenatally diagnosed CDH are best managed after birth. Cases with intermediate severity, where there is still a significant possibility that the baby's lungs won't function well enough at birth and may require the use of ECMO, are delivered in the presence of the experienced members of the CDH team. In rare cases with the worst prognosis, intervention before birth to include single port endoscopic fetal tracheal occlusion to allow accelerated lung growth will be offered.

This treatment option is offered on a case by case basis, and only after extensive discussion with parents, referring physician, local experts and the multidisciplinary antenatal diagnosis and management (MADAM) board of our Fetal Treatment Program.

It is our belief that the babies with highest risk diaphragmatic hernia, where the chances of survival is estimated at less than 10-40 percent, endoscopic fetal tracheal occlusion will allow accelerated lung growth to occur enough that it will convert the severe condition into a condition with intermediate or good prognosis, and a predicted survival greater than 50-65 percent.

## TREATMENT AFTER BIRTH

If the diagnosis is not known prenatally, the baby will develop signs suggesting a diaphragmatic hernia. This usually includes difficulty breathing with a very fast breathing rate, a very high heart rate and hypoxia (not enough oxygen in the blood) and cyanosis (a blueish color of the skin, caused by a lack of oxygen). The baby will be treated by our expert team of CDH specialists, which includes a pediatric surgeon and a neonatologist in the neonatal intensive care unit (NICU).

Babies with CDH are typically unable to breathe alone and do not get enough oxygen - they will require the help of a respirator (also called ventilator). Some babies who can not be managed successfully on the ventilator may require a temporary lung bypass machine (ECMO). Once the baby's condition has improved (this may take up to a week or longer), the operation (to bring intestines down into the abdomen and close the hole in the diaphragm) will be performed.

It may seem strange to wait that long to perform the operation (since, after all, it is the hole in the diaphragm that caused the problem). However, we now know that pulmonary hypoplasia is the true problem and that lung function needs to be corrected first. We also know that operating too soon may upset the very delicate balance that the newborn is in, causing a seemingly stable baby to get much worse very fast during or after the operation.

The operation requires an incision in the skin and the muscles of the abdomen. All of the abdominal contents (including small and large bowel, stomach, spleen and sometimes liver) are removed from the chest and placed back into the abdominal cavity, and the hole in the diaphragm is repaired. Sometimes, the hole in the diaphragm is very large and may require an artificial patch to be placed.

After surgery the baby will be kept in the neonatal intensive care unit until full recovery. Many babies will require ventilator assistance for a number of days to weeks after surgery, until the lung has time to recover and improve its function.

## OUTCOMES

The future for babies with diaphragmatic hernia is improving. The outcome for these babies is primarily based on the degree of lung underdevelopment and prevention of injury to those fragile lungs at birth. All babies will require a strict follow-up with the pediatric surgical service, to assist the long-term management of problems that may be part of the condition. Many babies may have problems requiring medications for lung therapy. Other babies have feeding problems from the malpositioned stomach in the chest during development that leads to difficulty eating and with weight gain. Gastroesophageal reflux is one such condition. Many times babies require additional feedings through the placement of a feeding tube to give more calories in order to grow and become healthier. This detailed follow up also provides the opportunity to ensure proper development. Early problems identified during follow up with feeding, growth, or failure to meet normal milestones, are treated early. Early intervention with speech therapy and occupational therapy often help these babies improve muscle strength and coordination. Although this requires a lot of work, with this established teamwork, the overall outlook for babies born with CDH and pulmonary hypoplasia is often bright.

## GLOSSARY

**Alveole:** an air-filled "bubble" lined with thin lung cells (epithelial cells) that is the smallest unit of lung tissue. Our lungs are composed of millions of such alveoli

**Bochdalek:** a type of diaphragmatic hernia named after Vincent Alexander Bochdalek, the Czech anatomist (1801-1883) who first described the condition. It is the most common type of diaphragmatic hernia, whereby the hole in the diaphragm is in the back ("posterolateral"). It is more common on the left than on the right. The other type of diaphragmatic hernia is the hernia of Morgagni

**Carbon dioxide:** Carbon dioxide (CO<sub>2</sub>) is a gas that is made as a byproduct of breathing (when oxygen is used by the body). Just as oxygen is breathed into the lungs, carbon dioxide is breathed out. One of the main functions of blood is to transport oxygen from the lungs (or the placenta, in the fetus) to the tissues in the body, and to transport carbon dioxide from the tissues back to the lung (or placenta), so that the body can get rid of it

**Chromosomal:** related to chromosomes. A disease or condition is called chromosomal when it is known to occur because of the presence of an abnormal chromosome. Normally, there are 26 pairs of chromosomes in each human cell. If there is one copy of a chromosome too many, the condition is called trisomy. The most common trisomies are Trisomy 13, Trisomy 18 and Trisomy 21 (Down syndrome). Because of the thousands of genes on each chromosome, chromosomal anomalies are generally much more serious than genetic anomalies

**Congenital:** condition that is present at or before birth. A congenital diaphragmatic hernia is a hole in the diaphragm that is present in the fetus

**Congenital diaphragmatic hernia (CDH):** a hole in the diaphragm found in the fetus; it causes the intestines and other abdominal organs to move into the chest and compress the lungs, and often causes severe breathing problems at birth

**Diaphragm:** the thin muscle layer that separates the chest from the abdominal cavity

**Endoscopic fetal surgery:** The technique of operating on a fetus or the placenta through tiny telescopes and surgical instruments, without having to open the uterus

**Esophagus:** the food-pipe. In the embryo, the trachea (wind-pipe) and bronchi (the smaller airways) form from an outpouching of the upper portion of the esophagus

**Extracorporeal Membrane Oxygenation (ECMO):** An artificial lung system that allows the baby's lungs to rest, while blood is directed through a circuit with a "membrane oxygenator" that acts like a lung. Often used for newborns with pulmonary hypoplasia or other temporary lung problems, such as meconium aspiration syndrome

**Feeding tube:** Infants who are not able to feed by mouth (either because they cannot suck and swallow, or because they get easily tired doing it), it may be necessary to thread a small plastic tube into their stomach to feed them that way. Sometimes (especially if it appears that a feeding tube will have to be used for a long time), it may be better to surgically place a small tube directly into the stomach (gastrostomy tube, or "G-tube"). More information on G-tubes is available on our pediatric surgery website

**Fetal:** related to the fetus

**Fetal intervention:** any treatment applied directly to the fetus, before birth. More specifically, it means an operation or other invasive procedure on the fetus

**Fetal surgery:** the most aggressive form of fetal intervention, whereby an operation is performed on the fetus. This can be done by "open" fetal surgery, where the fetus is partially removed from the uterus through a large incision in the mother's abdominal wall and uterus, or endoscopically, whereby tiny telescopes and surgical instruments are introduced into the uterus

**Familial:** a condition or disorder that is seen in several members of a family. Such a condition is suspected to be a genetic one: predetermined by a gene defect (even if the exact gene or gene defect is not yet known). The opposite of a familial disorder is a sporadic disorder: one that occurs randomly, not one that "runs in families"