

Questions, and Answers About:

Congenital Diaphragmatic Hernia

What Is a Diaphragmatic Hernia?

Congenital diaphragmatic hernia (CDH) is a hole in the diaphragm muscle that separates the chest from the abdomen (Figures 1, 2). This hole develops early in pregnancy, usually around 6-8 weeks after conception. CDH allows the intestines to move up into the chest cavity and is associated with undergrowth of the lungs.

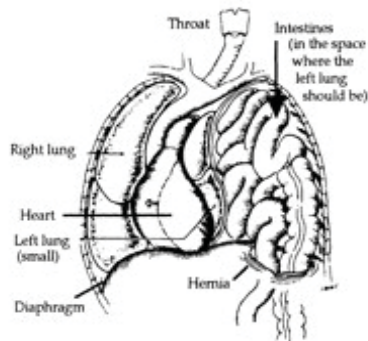


Figure 1. Diagram of the intestines pushing (herniating) up into the chest cavity. Diagram courtesy of Children's Hospitals and Clinics of Minnesota.

Why Are Babies with CDH So Sick?

Undergrown lungs cannot take up oxygen or get rid of carbon dioxide fast enough to support a baby's needs. Patients with undergrown lungs and CDH also have high blood pressure in the lungs (pulmonary hypertension). This pulmonary hypertension occurs because the blood vessels in the lung are abnormally formed and/or too small. Because the blood vessels in the lung are small, the heart must pump against high resistance to get blood through the lungs. If the pulmonary hypertension gets too high, the heart fails and cannot pump enough blood through the lungs. When this happens, the body does not receive adequate oxygen.

How Often Does CDH occur?

About 1 of every 2500 infants born in Minnesota is born with CDH. Most (78%) occur on the

left side of the chest, 20% on the right, and in 2% the defect is on both sides.

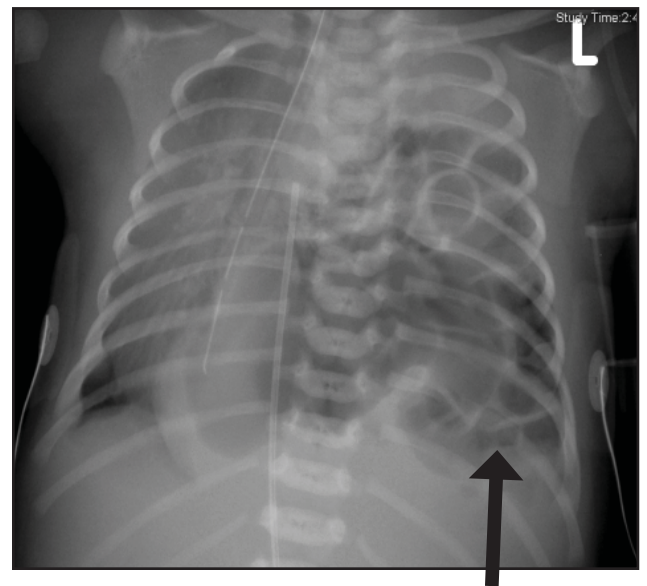


Figure 2. Chest X-ray of patient with left-sided CDH. Arrow points to the stomach and intestine in the left chest cavity.

What Are My Baby's Chances of Survival?

Overall survival at Children's Hospitals and Clinics of Minnesota- Minneapolis Campus is 65% (100/154, data from January 1995 to December 2005). These data include early deaths in the delivery room and those with other major birth defects. Survival depends on four factors:

1. The degree of lung undergrowth

The more lung tissue that is present, the more likely the baby is to survive. It is very difficult to measure the size of the lungs before birth, even with prenatal ultrasound and MRI imaging. However the following have proved somewhat helpful.

a. Absence of the liver in the chest.

Infants who do not have part of the liver in the chest usually survive. This

CONGENITAL DIAPHRAGMATIC HERNIA continued...

seems to indicate a smaller amount of intestine in the chest and less compression of the lung. This has not been 100% reliable in that some infants without liver in the chest have still died.

b. Lung-to-Head ratio.

The size of the lung opposite the hernia determines survival. The Lung-to-Head Ratio (LHR) is one way to estimate the lung's size relative to the rest of the baby (Table). However, in general, the larger the lungs, the better the chance of survival.

Table
LHR and Survival with CDH

<u>LHR</u>	<u>Survival Rate</u>
<0.6	0% (0 of 5)
>0.6 and <1.35	57% (16 of 28)
>1.35	100% (5 of 5)

The area of the lung (opposite the side of the hernia) at the level of the atria of the heart can be estimated by fetal ultrasound and compared to the head circumference (See Metkus et al, J. Ped Surg,

31, 1996:148-152). This method of estimating lung size is based on a small number of patients and we have not always found the same results as the original study reported. In the past, if the liver had moved into the chest and the LHR was < 1.4 at about 25 weeks gestation, referral for fetal surgery (tracheal plug) was considered. However, a recent study showed that fetal surgery doesn't increase survival. At this time, fetal surgery remains an unproven therapy and is offered only on research protocol.

2. Gestational age at delivery.

The second factor influencing survival is gestational age at delivery (Figure 3). Patients with CDH survive more often if they are born after 36 weeks gestation. In our experience those born at 39-41 weeks gestation have the best survival compared to those born at other gestational ages.

3. Presence of other major birth defect(s).

The third major factor determining survival is the presence of other birth defects, which are seen in about 20% of CDH patients. Patients with other major birth defects have a survival rate of only about 14%, compared to a survival rate of 73% for patients without these

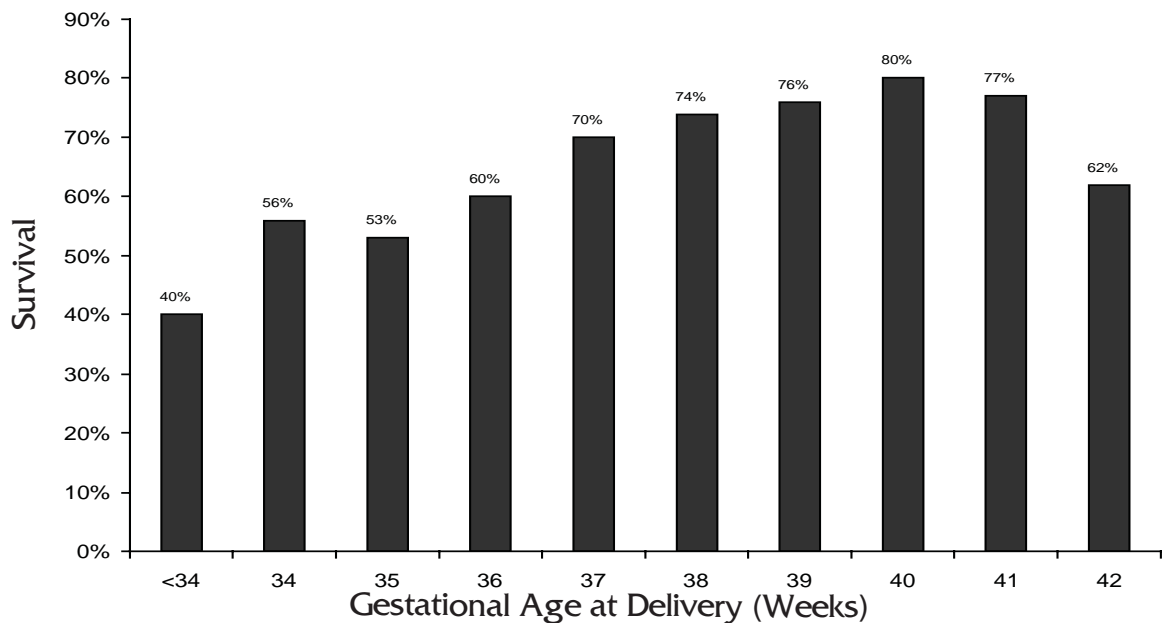


Figure 3. Gestational age and survival. Data from Children's Hospitals and Clinics of Minnesota-- Minneapolis Campus.

CONGENITAL DIAPHRAGMATIC HERNIA continued...

additional birth defects. About 10% of CDH patients will have a chromosome problem, which reduces the chance of survival and increases the risk of mental retardation. We recommend a chromosome analysis before delivery for all fetuses with CDH. Fetal ultrasound can identify most major birth defects before delivery.

4. Pulmonary hypertension

The fourth factor influencing survival is pulmonary hypertension. High blood pressure in the blood vessels of the lung reduces the amount of blood that the heart can pump through the lungs and increases the strain on the heart. Both of these factors reduce the amount of oxygen that the lungs can supply to the body. Pulmonary hypertension usually improves over the first few days to weeks after birth, but can be very severe and even require artificial heart/lung support (extracorporeal membrane oxygenation or ECMO, see below). Much of our treatment is aimed at reducing the pulmonary hypertension. All CDH patients have at least some pulmonary hypertension at birth. Unfortunately, in some babies, the pulmonary hypertension never improves enough to allow survival. It is difficult to predict the severity of pulmonary hypertension prior to birth in CDH patients. Some CDH babies with seemingly small lung

volumes will do well and others with apparently large lung volumes will not do well.

How is CDH Treated?

In the delivery room, the NICU staff will insert a breathing tube into your baby's lungs to assist breathing. We use various kinds of ventilators (conventional and high frequency) to deliver concentrated oxygen to the lungs and assist natural lung function. The ventilator helps your baby's lungs work more effectively by pushing air into the lungs and helping the air sacs to stay open. This results in more oxygen getting into the blood and better carbon dioxide elimination. The goal of artificial ventilation is to get adequate oxygen into the babies bloodstream and carbon dioxide out as gently as possible.

How Long Will My Child be on a Ventilator?

As the lungs improve, the ventilator assistance can be reduced and eventually stopped. In our experience, babies with CDH require a ventilator anywhere from 1 to 73 days. Although the median time on a ventilator is 11 days (Figure 4) we see great variability. Having a median of 11 days means that about half of all babies with CDH will no longer need the ventilator by their 11th day after birth (Figure 4). However, some babies even require a tracheostomy and ventilator support at home for a few months. Fortunately, this situation is very rare.

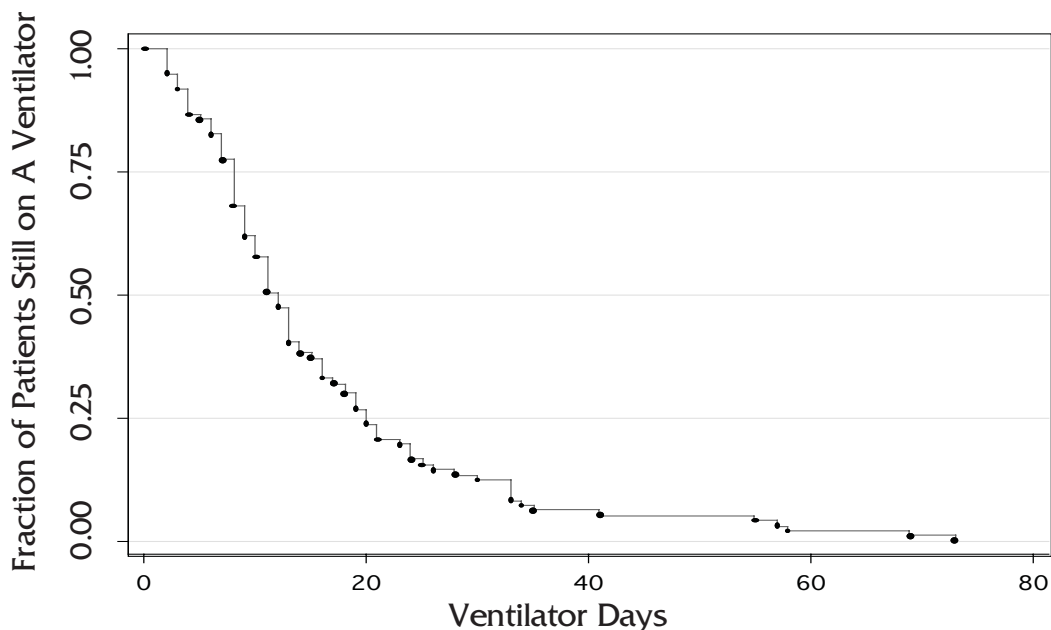


Figure 4. Length of time on a ventilator. Data from Children's Hospitals and Clinics--Minneapolis Campus.

CONGENITAL DIAPHRAGMATIC HERNIA continued...

Nitric Oxide and Other Treatments

To treat pulmonary hypertension, a special gas, called inhaled Nitric Oxide (iNO), is pumped into the lungs through the ventilator. iNO reduces pulmonary hypertension. It works by relaxing the blood vessels in the lung, which allows the blood to flow through the lungs more easily and at lower pressures. Unfortunately, the effect of iNO in CDH is often incomplete. We also use other medications such as dopamine, dobutamine, or milrinone to help the heart pump blood through the lungs.

How Long Will My Baby Stay In the Hospital?

The median length of stay is 27 days, but there is wide variation (Figure 5). In other words, half of the CDH survivors have gone home by their 27th hospital day while half still remain in the hospital.

What is ECMO and What Does It Do?

ECMO (extracorporeal membrane oxygenation) provides artificial lung and heart support that can sustain babies by adding oxygen and removing carbon dioxide from the blood until the baby's own lungs are functioning adequately. ECMO has proven extremely valuable in supporting babies with pulmonary hypertension.

The duration of ECMO support varies for CDH patients from a few days to a few weeks, but averages 9 days.

About 20% (our experience) to 34% (national experience) of CDH infants receive ECMO support as part of their treatment (Figure 6). Of those who receive ECMO, about 50% survive (Appendix 1). In general, infants with gestational ages less than 33 weeks are not offered ECMO because their survival rate is very low and they often sustain serious complications from the ECMO treatment. Some patients do not receive ECMO because this therapy would be futile. Those with multiple, severe birth defects or with extremely small lungs may not benefit from ECMO. Your baby's doctor will discuss this situation with you, if it arises.

When Will the Diaphragm Be Repaired?

Surgery to repair the diaphragm can temporarily worsen pulmonary hypertension. Therefore, we wait for the pulmonary hypertension to improve before repairing the diaphragm. This waiting period may last a few days or a few weeks. Occasionally, infants who require ECMO will be repaired while receiving ECMO therapy. If the surgeons cannot stitch the hole closed,

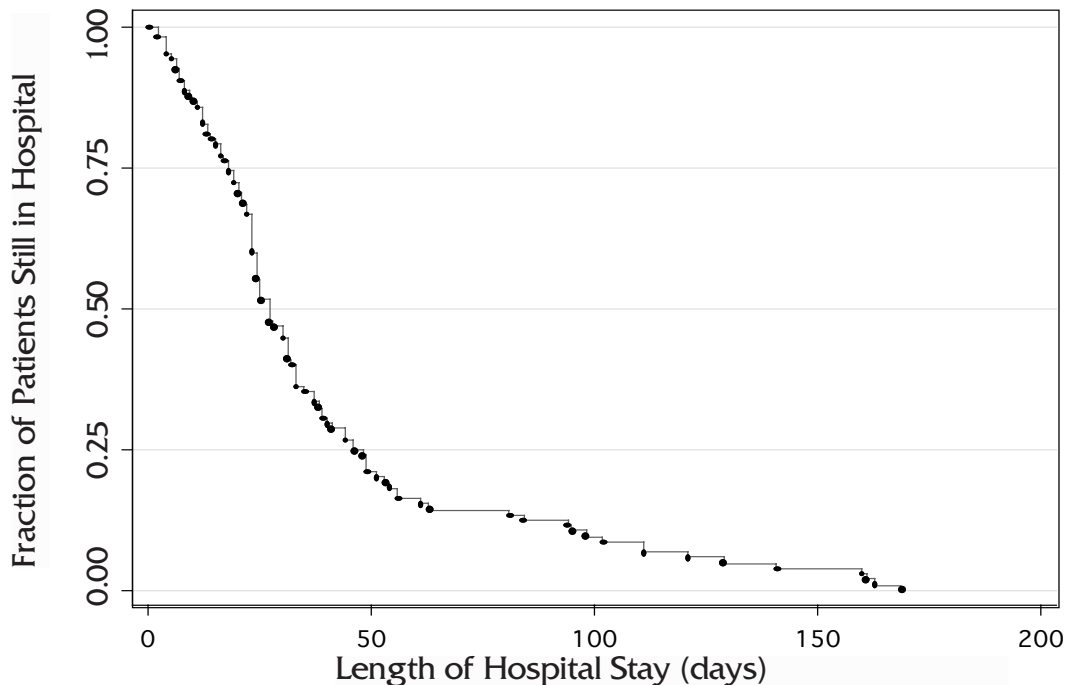


Figure 5. Length of hospital stay for patients with CDH. Data from Children's Hospitals and Clinics of Minnesota--Minneapolis Campus.

CONGENITAL DIAPHRAGMATIC HERNIA continued...

they will use a patch of synthetic material to close the diaphragm.

Why Did This Happen?

No one knows for sure why babies develop CDH. Occasionally there is a history of other family members having a CDH, but most of the time this occurs as a new problem in a family. Patients with CDH and a chromosome abnormality have more complicated conditions and a much lower survival rate than those without chromosomal problems. The most commonly associated chromosomal problems are trisomy 13, trisomy 18, and a deletion on chromosome number 15 [del (15) (q 24-26)]. Chromosome analysis is performed on all CDH babies.

Did the Mother or Father Cause the CDH?

We know of nothing that the mother or father could have done to cause or prevent CDH. No one knows why CDH occurs.

What Is My Risk of Having Another CDH Baby?

We generally quote the overall risk of having

another baby with CDH as being about 2%. However, each situation is different, and you should see a genetic counselor to review your specific risk. Your specific risk could be anywhere between about 1 in 50 to 1 in 4. We have treated several infants with CDH in the same family.

Why All the Tubes in My Baby?

Babies with CDH are often extremely ill. They require many types of support (Figure 6). Babies with CDH and severe pulmonary hypertension require breathing tubes, stomach tubes, and IV lines. Although these tubes look painful, babies receive sedation and/or narcotics to keep them comfortable. The baby in Figure 6 is sleeping and will be kept comfortable for as long as the tubes are needed. Figure 7 shows a baby who has recovered and no longer needs all of the tubes shown in Figure 6.

What Is the Long-Term Prognosis?

Most children have good lung function and participate fully with their peers in the usual activi-

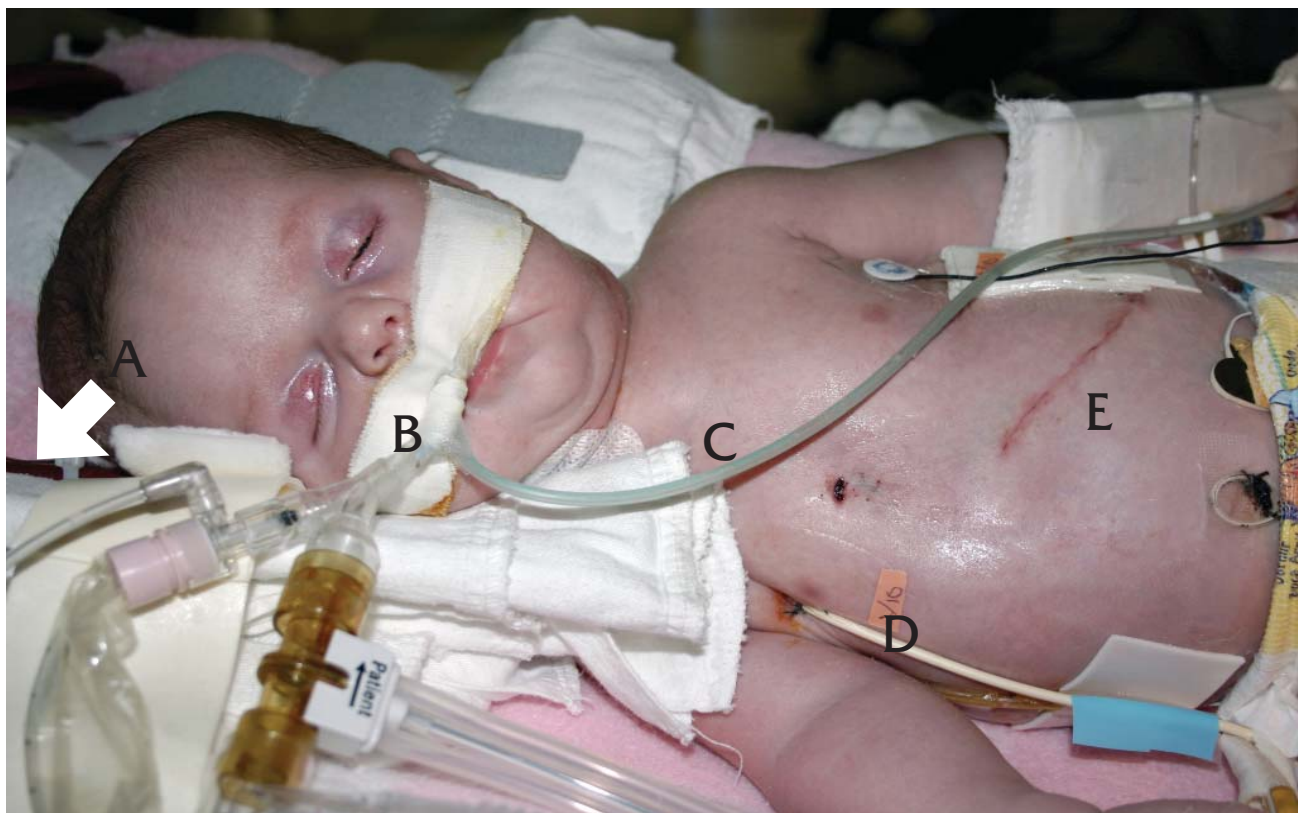


Figure 6. Photograph of infant on ECMO following surgical repair of CDH. A= ECMO catheter, B= Endotracheal (breathing) tube, C= orogastric tube (to keep the stomach empty), D=Chest tube to drain fluid or air from chest cavity, E= Incision site from repair of diaphragmatic hole. Photo courtesy of the NICU at Children's Hospitals and Clinics of Minnesota, Minneapolis Campus.

CONGENITAL DIAPHRAGMATIC HERNIA continued...



Figure 7. Baby who was treated with ECMO for CDH, shown here in the step-down unit no longer requiring the types of support that he once did. Photo courtesy of Children's Hospitals and Clinics of Minnesota, Minneapolis Campus.

ties of childhood. However, asthma occurs more commonly among infants who experience lung problems in infancy. During the first year, CDH infants are more likely to develop pneumonia with the usual viral infections of infancy. A few will have chronic breathing problems that may last months or years and require medications, supplemental oxygen, and rarely, a home ventilator. These are the exception to the rule.

Most children with CDH have normal intelligence and coordination. However, the sicker your baby is in the newborn period the more likely he/she will have some developmental problems. If your child has other birth defects, a chromosomal problem, or is severely ill, he/she has a higher risk of developmental and life-long problems, such as slow development or learning disabilities. We offer developmental screening to all CDH patients in our NICU follow-up clinic.

Hearing loss

This occurs in about 5% of CDH patients and may not be obvious at the time your baby is discharged from the hospital. Children who had CDH should have their hearing tested every 6 months until three years of age, even if the initial hearing test was normal.

Gastroesophageal reflux

This is the tendency to vomit easily and frequently, and it is a common problem in CDH patients. Treatment of reflux, may require frequent small meals, upright positioning, medication, a feeding tube, or rarely, surgery.

Having a patient with CDH is stressful and can be overwhelming. We will gladly meet with you to answer your questions and address your concerns at any time during your pregnancy or during your baby's hospital stay.

**If you have questions please contact us
(612) 813-6288, office or (612) 813-6295,
NICU.**

Internet References

We invite you to discuss information that you find on the Internet with your baby's doctor.

1. This Emedicine article provides good information and is reliable. However, it is a bit technical. <<http://www.emedicine.com/ped/topic2603.htm>>
2. CHERUBS site. This organization is the Association for Diaphragmatic Hernia Research, Advocacy and Support. They provide reliable information, a chat room, bulletin boards, and parent to parent opportunities. <<http://www.cherubs-cdh.org/>>.