

Questions, and Answers About:

Congenital Diaphragmatic Hernia

Introduction

Learning that your unborn baby has congenital diaphragmatic hernia (CDH) can be a shock and cause much worry. The purpose of this handout is to give you the facts so that you know what to expect and what questions you should ask. As you can see from the picture of one on our first CDH patients (Figure 1) treated with a special therapy, extracorporeal membrane oxygenation (ECMO), the outcomes can be very good. Each baby is unique and our goal is to give you basic information that will help you to participate more fully in your child's care.



Figure 1. Picture of one of our first CDH patients treated with ECMO. He is now in college. Picture courtesy of the patient.

What Is a Diaphragmatic Hernia?

Congenital diaphragmatic hernia (CDH) is the abnormal migration of the intestines or other abdominal organs into the chest through a hole in the diaphragm. The diaphragm is a muscle that separates the chest from the abdomen (Figures 2, 3). This hole develops early in pregnancy,

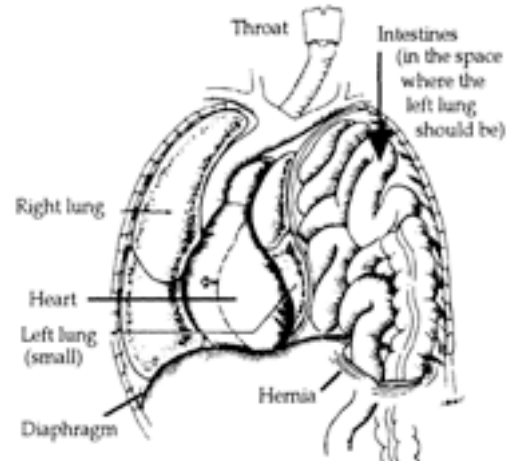


Figure 2. Diagram of the intestines pushing (herniating) up into the chest cavity. Diagram courtesy of Children's Hospitals and Clinics of Minnesota.

usually around 6-8 weeks after conception. CDH allows the stomach, intestines, spleen, and sometimes, the liver to move up into the chest cavity. Most importantly, undergrowth of the lungs usually accom-

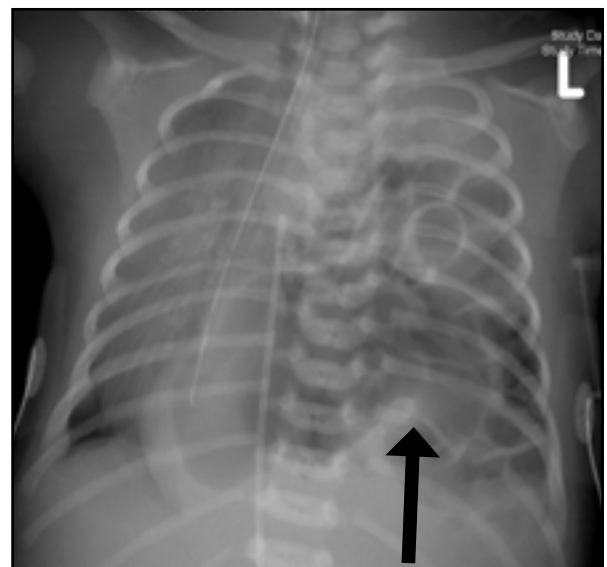


Figure 3. Chest X-ray of patient with left-sided CDH. Arrow points to the stomach and intestine in the left chest cavity. Picture courtesy of neonatologists.

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panies CDH. A diaphragmatic hernia can occur on either the left or the right side.

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). Pulmonary hypertension occurs because the blood vessels in the lungs are abnormally formed and/or too small. When the blood vessels in the lungs are small, the heart must pump against the high resistance of the narrowed blood vessels 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. All organs in the body require oxygen to survive and function properly. Pulmonary hypertension is the primary reason why CDH babies have such severe problems after birth.

How Often Does CDH occur?

CDH occurs in about 1 of every 2500 infants born in Minnesota. Most (78%) CDH occur on the left side of the chest, but about 20% occur on the right, and 2% on both sides.

Will My Baby Survive?

Overall survival at the National CDH Registry is 69%. The Registry includes over 50 centers and has data on over 3,500 babies. The survival at Children's Hospitals and Clinics of Minnesota- Minneapolis Campus is 87% (59/68, data from January 2005 to December 2009). These data include early deaths in the delivery room and those with major birth defects in addition to CDH. There are four factors which influence survival. The most important of these is the degree of lung undergrowth, which is largely determined by the size of the hole in the diaphragm. We recommend that CDH patients deliver at a perinatal center that has experience with these patients' special needs, such as ECMO (see below). We usually recommend that patients locate close to the hospital after about 36 weeks gestation, to reduce the likelihood of a delivery

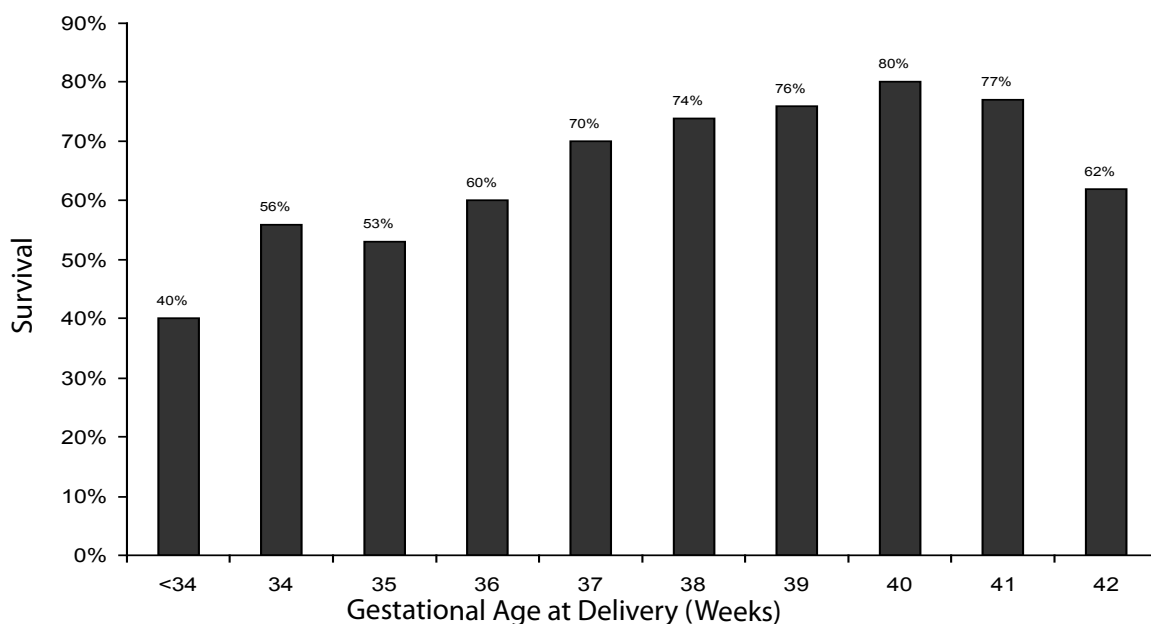


Figure 4. Survival by gestational age among CDH patients treated at Children's Hospitals and Clinics of Minnesota-- Minneapolis Campus.

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at a facility unprepared to address a CDH patient's unique needs. A vaginal delivery is often possible. Delivery by C-section does not improve the baby's chances of survival.

1. 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

With left-sided CDH, infants who do not have part of their liver in the chest have a 90% survival, as compared to 45-50% survival if part of the liver is in the chest. Absence of the liver up in the chest in left-sided CDH seems to indicate less lung compression, so the lungs can grow to a larger size than if there is liver in the chest. This has not been 100% reliable in that some infants without the liver in the chest still have severe lung undergrowth and may not survive.

b. Lung-to-Head ratio

The size of the lung on the opposite side of the hernia determines survival. The lung on the side of the hernia is always small. The Lung-to-Head Ratio (LHR) is one way to estimate the size of the lung on the opposite side of the CDH (Table). In general, the larger the lungs, the better the chance of survival.

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). 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. A somewhat more accurate meth-

od is to estimate the LHR and compare it to what would be expected at a specific gestational age (Deprest, et al., Semin Fetal Neonatal Med, 2009;14:11)

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)

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

Patients with CDH survive more often if they are born after 36 weeks gestation (Figure 4). In our experience those born at 39-41 weeks gestation have the best survival compared to those born at other gestational ages.

3. Other Major Birth Defect(s).

The third major factor determining survival is the presence of other birth defects. The CDH Registry indicates that up to 30% of CDH patients will have an additional birth defect. In our data, the rate is lower, 21%. Additional birth defects reduce survival by about 10 percentage points to 79% compared to 87% for all CDH babies. In about 10% of CDH patients a chromosome problem or a group of birth defects will occur together with the CDH. We recommend chromosome studies on all CDH patients and sometimes their parents either prior to or after birth. Fetal ultrasound can often identify other major birth defects before delivery.

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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. This reduces the amount of oxygen that the lungs can supply to the body. The pulmonary hypertension must improve for the baby to improve. It is difficult to predict the severity of pulmonary hypertension prior to birth in CDH patients. Some CDH babies with seemingly small lung volumes do well and others with apparently large lung volumes do not do well.

Much of our treatment is aimed at reducing pulmonary hypertension. All CDH patients have some pulmonary hypertension at birth. It usually improves over the first few days

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 to remove carbon dioxide as gently as possible.

How Long Will a Ventilator be Needed?

As the lungs improve, ventilator assistance can be reduced and eventually stopped. In

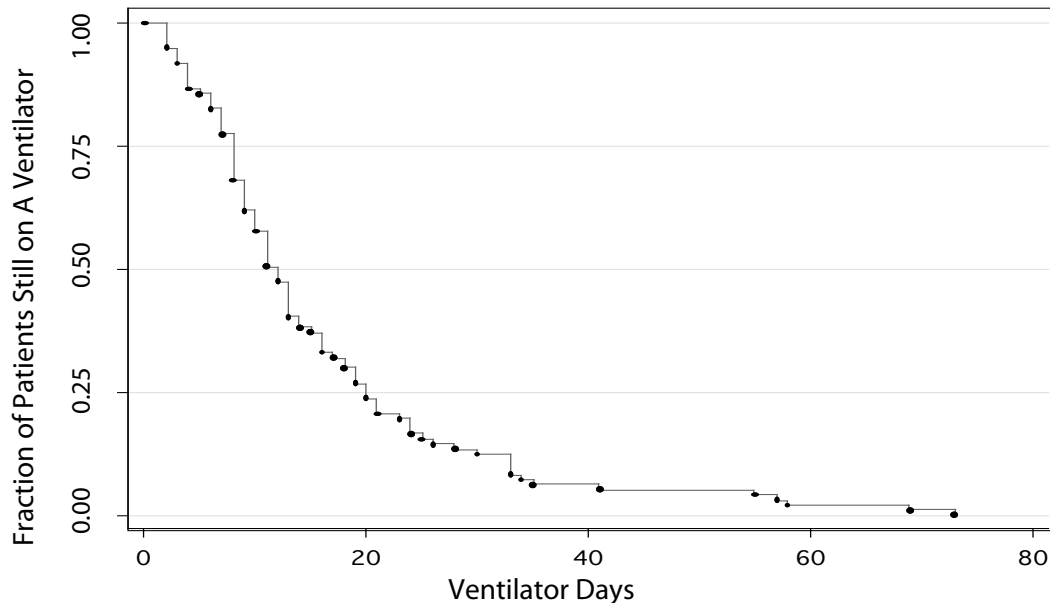


Figure 5. Total number of days spent on a ventilator from birth to hospital discharge. Data from CDH patients treated at Children's Hospitals and Clinics--Minneapolis Campus from 1998 to 2008.

to weeks after birth, but can be severe and even require artificial heart/lung support (extracorporeal membrane oxygenation or ECMO, see below).

our experience, babies with CDH require a ventilator anywhere from 1 to 73 days. Although the median time on a ventilator is

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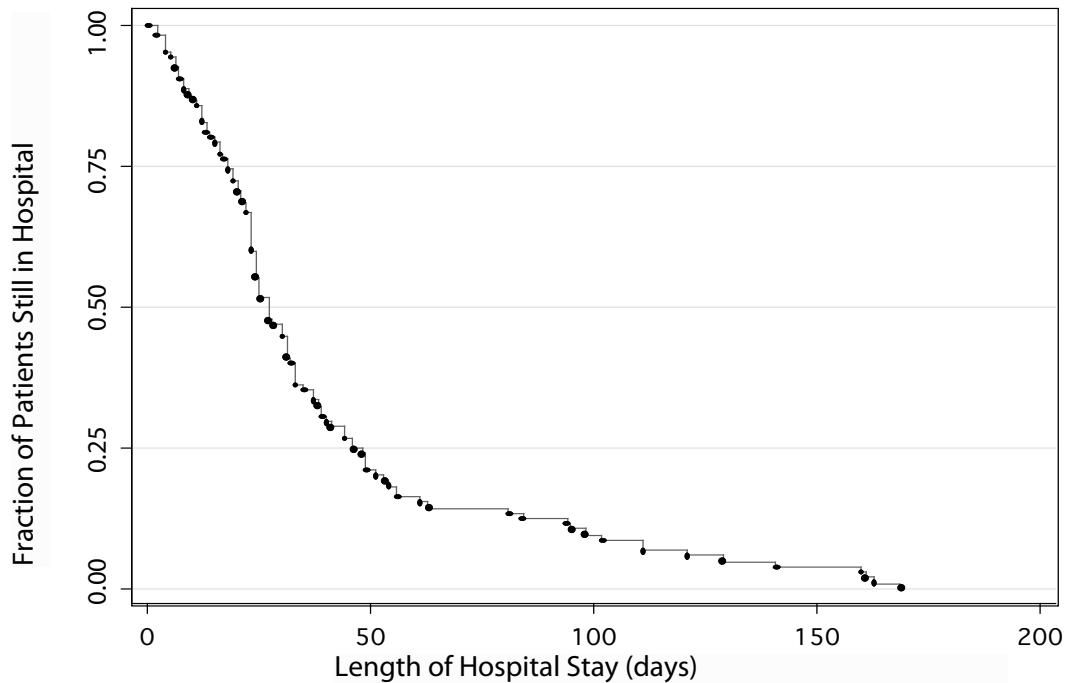


Figure 6. Total number of days from birth to hospital discharge. Data from CDH patients treated at Children's Hospitals and Clinics--Minneapolis Campus from 1998 to 2008

11 days (Figure 5) 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. Fortunately, this situation is rare.

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 hydrocortisone to help the heart pump blood through the lungs and to the rest of the body.

Feedings and Nutrition

Before repair of the diaphragm, CDH patients are fed by vein with a nutritional fluid called hyperalimentation or total parenteral nutrition (TPN). This fluid contains sugar, protein, fat, and vitamins. Initially, TPN may be given through a tube in the umbilicus. Later special IVs may be placed in other areas (arms, legs, head, or neck). These IVs may be placed by a needle inserted under the skin or may require surgery to locate them properly. As soon as possible after the diaphragm is repaired and normal intestinal function develops, we start feedings through a stomach tube. When your baby is off of the ventilator, oral feedings are started, including breast feeding, if the parents so desire. A baby's ability to take all feedings by mouth is one of the key determinants of discharge readiness.

What is ECMO and What Does It Do?

ECMO (extracorporeal membrane oxygenation) provides artificial lung and heart

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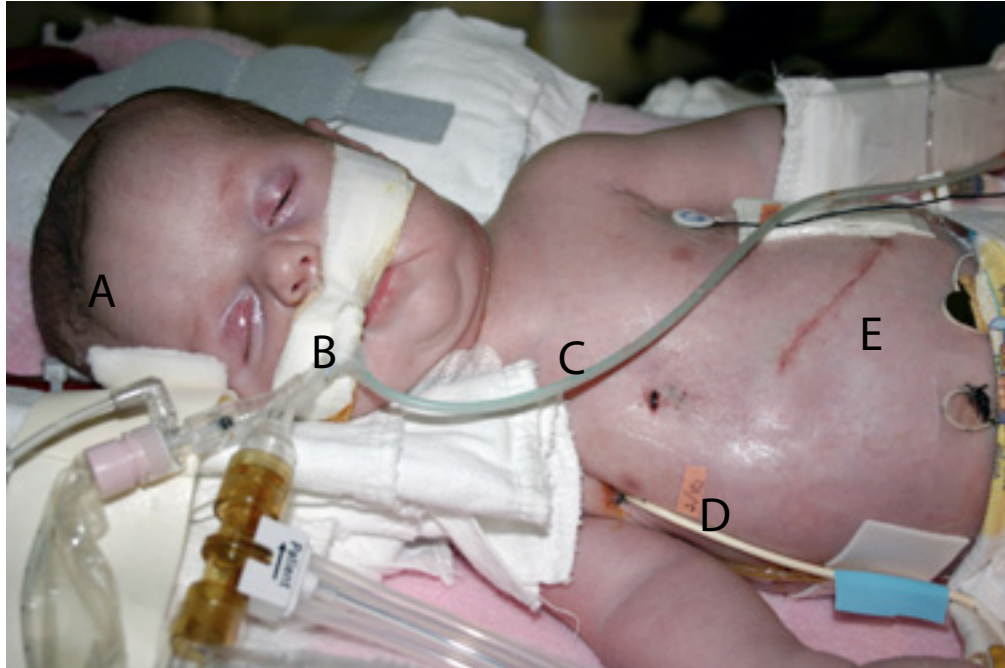


Figure 7. Photograph of infant on ECMO following surgical repair of CDH. A= ECMO catheter, B= Endotracheal (breathing) tube, C= orogastric tube (to keep the stomach empty), D=Chest tube to drain fluid or air from chest cavity, E= Incision site from repair of diaphragmatic hole. Photo courtesy of the NICU at Children's Hospitals and Clinics of Minnesota, Minneapolis Campus.

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 14 days. If ECMO is required, it is usually started on the first or

second day after birth, prior to the repair of the diaphragm.

About 40% of CDH infants receive ECMO support as part of their treatment (Figure 6). Of those who have received ECMO at Children's Hospitals and Clinics from 2005-2009, 78% (21/27) have survived to hospital discharge. With improvements we have made to



Figure 8. 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.

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our ECMO protocol in the last several years our most recent survival for CDH treated with ECMO has risen to 88% (14/16). In general, infants with gestational ages less than 34 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. There is a separate parent information sheet on ECMO.

Diaphragm Repair

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 to a few weeks. Often, infants who require ECMO will be repaired while receiving ECMO therapy. If the surgeons cannot stitch the hole closed, they will use a patch of synthetic material to close the diaphragm.

Length of Hospital Stay?

The median length of stay is 27 days, but there is wide variation (Figure 6). In other words, half of the CDH survivors have gone home by their 27th hospital day while half still remain in the hospital.

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)].

Did the Parents 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.

Could I Have 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 from the same family.

Why All the Tubes in My Baby?

Babies with CDH are often extremely ill. They require many types of support (Figure 7). Babies with CDH and severe pulmonary hypertension require breathing tubes, stomach tubes, and IV lines. Although these tubes look painful, most are not. Additionally, babies receive sedation and/or narcotics to keep them comfortable. The baby in Figure 7 is sleeping and will be kept comfortable for as long as the tubes are needed. Figure 8 shows a baby who has recovered and no longer needs all of the tubes shown in Figure 7.

Long-Term Prognosis

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Lungs. Most children have good lung function and participate fully with their peers in the usual activities of childhood. However, babies with CDH have increased risk for pulmonary problems after discharge and throughout their lives. In the first year, about 25% of babies with CDH return to the hospital for pneumonia or other respiratory problems. Routine vaccinations and for some infants, a special immunoglobulin preparation against respiratory syncytial virus (RSV) helps to reduce these rehospitalizations. All patients with any ongoing respiratory problems at discharge from the hospital should have this special immunoglobulin to prevent severe RSV. We strongly encourage that all infants be vaccinated according to current guidelines.

About 25-50% of CDH babies will develop scarring in their lungs called bronchopulmonary dysplasia (BPD). This results from damage to lung tissue as a consequence of the CDH and to some extent, damage from the treatments used to help the undergrown lungs work satisfactorily. Babies with BPD may require supplemental oxygen and inhaled or oral medications to treat the BPD both in the hospital and at home. Rarely, CDH babies may even go home on a ventilator with a tracheostomy. This occurs in about 10% of CDH patients. About 15% of CDH patients will go home requiring supplemental oxygen to help them breathe. BPD improves over time even for the most severely affected babies.

Asthma or asthma-like symptoms occur in about 25% of CDH patients by the time that they are 5 years of age. Asthma symptoms can be treated with medications, but may recur for many years.

A few babies have persistent pulmonary

hypertension that lasts for months or even years after the infant goes home. If pulmonary hypertension is present it requires careful monitoring and increases the risk for early mortality.

Growth Problems

Infants with CDH often have problems taking their feedings by mouth. This is probably due to both the CDH and treatment with oral ventilator tubes and other treatments. As many as a third of CDH patients require a feeding tube at discharge and up to about 50% will grow slowly in the first year after birth. Although the best treatment to prevent slow growth is not generally agreed upon, all clinicians agree that adequate calories are critical to lung and brain development. If your baby is not taking adequate calories by mouth, a gastrostomy (surgically placed feeding tube) may be required for the first months after discharge.

Gastroesophageal reflux (GER)

During pregnancy, the fetal esophagus and stomach are often abnormally stretched and pulled up into the chest cavity. This abnormal development allows the acid in the stomach to wash up into the esophagus. GER can cause vomiting, poor feeding, and damage the tissue lining of the esophagus. This damage to the esophagus is sometimes called Barrett's esophagus). Most patients require medicine to reduce the acid content of stomach. Upright positioning during and immediately after feedings often helps. Few infants require surgery, but the risk of Barrett's esophagus persists for many years.

Hearing Loss

Babies with CDH have a high risk of

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sensorineural hearing loss in childhood. Some studies report up to half of CDH children develop some hearing loss in infancy. The reason for this is not known, but probably relates to the severity of their illness, genetic factors, and the extensive use of multiple medications required to treat CDH. Even those infants with a normal hearing screen at hospital discharge are at risk to develop hearing problems later in childhood. We recommend formal hearing tests to screen for hearing loss every 6 months until age 3 years and then yearly until age 5 years.

Development

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 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.

Hernia recurrence

Up to 50% of patients, who require a patch to close the hernia, will have recurrence of the hernia in childhood. In other words, the patch will tear loose as the child grows.

A patch is used when the hole in the diaphragm is so large that the muscle cannot be sewn together to repair the hernia. Recurrences do not usually cause serious problems. Signs of recurrence are frequent vomiting or persistent cough. Recurrent CDH must be repaired surgically.

Orthopedic problems

From 25% to 50% of CDH patients may have either curvature of the spine (scoliosis) or a concave breast bone (pectus excavatum). This does not usually require intervention, but should be monitored carefully through childhood and adolescence until the child reaches adulthood.

Internet information

We encourage you to learn as much as you can about CDH on the Internet. However, not all of the information on the Internet is accurate and not all of it will apply to your baby. We invite you to bring any information from the Internet to our attention so that we can help you understand how that information applies to your baby.

We welcome your questions.

For questions please contact us at (612) 813-6288, office or (612) 813-6295, NICU.

Internet References

1. This E-medicine article provides good information and is reliable. However, it is a bit technical. <<http://www.emedicine.com/>



Figure 9. Photograph of former CDH patient treated with ECMO Photo courtesy of the patient.