Congenital Diaphragmatic Hernia

By Dr. N. Robert Payne

Congenital diaphragmatic hernia (CDH) is one of the most difficult, and commonly lethal, pulmonary problems seen in newborns. Many factors, from therapy options, additional anomalies to the actual cause of the problem, make CDH difficult to treat.

What is it?
CDH consists of the triad of lung hypoplasia, incomplete formation of the diaphragm and herniation of the abdominal contents into the thorax. In about 90 percent of CDH patients, the hernia occurs on the left side, with the remainder occurring on the right side. Very few of these hernias occur bilaterally. Originally, surgeons thought that the lung hypoplasia resulted from compression of the developing lung by the herniated abdominal contents. This line of thinking suggested that treatment should focus on relieving the lung compression by immediate surgical correction of the diaphragmatic hernia. Subsequent studies in the late 1990s cast doubt on this pathogenetic model.

It now seems likely that the primary problem may be lung hypoplasia caused by abnormal embryological development. This abnormal lung growth then leads to the failure of the diaphragm to form completely, with subsequent herniation of the abdominal contents into the thorax. Realization that pulmonary hypoplasia is the cause, not the result, of the diaphragmatic hernia has opened new possibilities for therapy.

Why is CDH a serious problem?
The severity and frequency of CDH make it a significant neonatal problem. CDH continues to carry a mortality risk of 30-40 percent and to occur in about 1:2,500 births. Although that may sound like a relatively rare event, there are roughly 4 million births in the U.S. each year. That means there are approximately 1,600 affected fetuses and 500 newborn deaths annually due to CDH. About two-thirds of those cases are detected prenatally.

Despite many advances in neonatal treatment, two factors make CDH a difficult problem to treat: the high frequency of associated anomalies and the lack of good therapies for pulmonary hypoplasia. About 20 percent of infants with CDH have other anomalies. These associated anomalies include trisomies, congenital heart disease, and myriad of other problems. Many of these associated anomalies are also life-threatening and increase the mortality risk associated with CDH. For example, CDH patients with congenital heart disease have about a 50 percent mortality risk, compared to about 30 per-
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mechanical ventilation, high-frequency ventilation, nitric oxide and sometimes extracorporeal membrane oxygenation (ECMO). About 30 percent of infants with CDH have such severe pulmonary hypertension that only extended artificial lung support can support these infants until the pulmonary hypertension improves. Sometimes even with artificial lung support, the pulmonary hypertension never resolves. About 50-60 percent of CDH patients treated with ECMO survive.

Prenatal treatment
Correcting the pulmonary hypoplasia before birth should offer the best hope for survival and requires prenatal treatment. Innovative surgical teams have been developing fetal surgery to correct the pulmonary hypoplasia. Despite many setbacks and frustrations, a current, randomized trial of tracheal plugging to reverse pulmonary hypoplasia seems to hold the most promise. Fetuses with CDH and liver herniation into the thorax are more likely to die than are those CDH patients without liver herniation. Harrison, et al. have organized a randomized clinical trial of video-endoscopic plugging of the trachea with an inflatable balloon at approximately 25-28 weeks gestation. Plugging the trachea causes back pressure on the lung as normally secreted fluids cannot egress out of the trachea. This back pressure causes the hypoplastic lung to grow, reversing the primary problem of pulmonary hypoplasia. At delivery, the occlusion of the trachea must be removed to allow air movement into and out of the lungs. The final results of this trial have not yet been published.

What are the outcomes?
Overall, 60-70 percent of CDH patients survive with current therapy. Survival depends on the presence of associated anomalies and the degree of pulmonary hypoplasia. Among survivors chronic pulmonary problems, gastro-esophageal reflux, oral aversion, and developmental delays occur with an increased frequency.

How can one predict which patients are at high risk?
Several prenatal tests are recommended for evaluating the severity of pulmonary hypoplasia. For example the lung/head ratio, ratio of pulmonary arteries to the aorta, the left ventricular mass, fetal lung volume from maternal MRI, and the presence of the liver in the chest are all reported to estimate survival. Most of these methods were derived from relatively small numbers of patients and have not been compared in a large, multi-institutional study. The two most generally accepted methods are the lung/head ratio and the presence of the liver in the thorax. Both of these factors can be assessed by prenatal ultrasound.

Implications for case managers
Case managers can help families sort through the often bewildering array of information and decisions that they may confront when told that their unborn baby has CDH. Often, the following questions will arise:

Should we continue the pregnancy?
Even with current prediction methods, such as the lung/head ratio, unpredictable outcomes can occur.
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Some high-risk CDH infants do well and some low-risk CDH infants die despite appropriate therapy. The fetal karyotype, the presence or absence of other anomalies and some estimate of the severity of pulmonary hypoplasia should be given to the parents when they are trying to make this decision.

Where should the mother deliver?
Some survivors have required ECMO and other high-tech support within the first few hours after delivery. Although there is controversy on this point, the best chance for survival would seem to be with delivery at an ECMO center. Such centers are familiar with the meticulous care that must be provided from the moment of birth.

What will the treatment be like?
Families are rarely prepared for the many wires, tubes, medications and procedures that will be required to treat their infant with CDH. These infants often require numerous technologies, surgery and a hospital stay that can extend over weeks or months. Parents should be prepared for this extended hospital stay, even if their infant survives and does well. A prenatal tour of the NICU helps many parents know what to expect following delivery.

Will my baby survive? The presence of associated anomalies and the degree of pulmonary hypoplasia generally determine survival. Is there a chromosomal problem? Is congenital heart disease present? What was the lung/head ratio? (A ratio <1.2 is associated with increased mortality risk.) Is the liver herniated into the thorax? On the basis of these questions it is almost always possible to assign the infant to a high- or low-risk category. Infants born prematurely do much worse than those born at 38 weeks or more gestation.

What is the long-term outcome?
Infants with CDH have a significant risk of gastro-esophageal reflux, oral aversion, short stature, scoliosis and developmental delays. Parents and case managers should be aware of these risks so that optimal therapy can be provided to the infant at the earliest possible time.

Summary
CDH remains one of the most difficult and most commonly lethal pulmonary problems seen in newborns. Although great strides have been made in treatment, 30-40 percent of CDH patients still do not survive the neonatal period. Contrary to most people’s intuition, the primary problem is not the herniated abdominal contents, but the pulmonary hypoplasia. Future treatments will likely focus on antenatal correction of the pulmonary hypoplasia.

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References


Mark your calendar for the Nineteenth Annual ROSE® Seminar July 20–22, 2003 at the Minneapolis Marriott

No registration fee. Watch for brochures and registration materials in May!

Here is a hint of some of the topics to be presented this year:

- Spinal cord injury research
- Pain management and use of botulinum toxin injections
- Quality improvement and tools that work to improve data
- Morbid obesity and treatment options
- Long term outcomes of NICU babies
Every year, 35,000 babies are born with congenital heart disease (CHD). Statistics currently show that there are more than an estimated one million Americans who suffer from cardiovascular defects. Congenital heart disease is considered one of the most common types of birth defect and is the leading cause of birth defect-related deaths.

CHD can be detected at birth and may require immediate care or the condition may be so slight that the child appears healthy for many years. Sometimes CHD will be detected when the child is older or when he or she has grown into adulthood. In the past 40 years, advances in diagnosis and surgical treatment have led to dramatic increases in the survival of children suffering from CHD.

What is congenital heart disease?
A diagnosis of congenital heart disease is used for a range of heart defects affecting different parts or functions of the heart. These defects occur during the early part of pregnancy when the heart is forming and are thought to be caused by genetic and environmental factors. Alcohol consumption during pregnancy as well as viruses such as maternal rubella or maternal diabetes mellitus have been linked to CHD, but many other contributors are still unknown.

Although most cases of CHD involve only the heart, some forms are part of genetic and chromosomal syndromes that affect many organs. Among these syndromes are Down syndrome, trisomy 13, Turner’s syndrome, Marfan syndrome, Noonan syndrome, and Ellis-van Creveld syndrome.

Prenatal echocardiography and ultrasound can accurately detect heart defects in 93 percent of cases. Some heart problems that are detected in utero can be treated with medications prior to birth. In other cases, a prenatal diagnosis of CHD provides parents and doctors with time to consider prenatal treatment options as well as the opportunity to make arrangements for delivery of the child in a center that specializes in neonatal and pediatric cardiology. As a result, there are a handful of pediatric CHD programs that make it possible for newborns suffering from CHD to receive the diagnosis and care that they need for the best possible outcomes.

Congenital Heart defects generally fall in one of two categories:

Cyanotic heart disease – a group of defects in the structure or function of the heart or great vessels preventing normal blood flow from the right to left part of the circulatory system. This abnormal blood flow results in cyanosis, poor oxygenation of the body that causes a bluish coloration in the lips, fingers and toes during exercise. The seriousness of cyanotic heart defects vary from ones that can cause enormous distress to a newborn to others that cause few, if any, problems until adulthood.

Among the congenital heart defects that cause cyanosis are:
• Tetralogy of Fallot
• Transposition of the great vessels
• Ebstein’s anomaly
• Tricuspid atresia (a deformity of the tricuspid heart valve)
• Total anomalous pulmonary venous return
• Pulmonic stenosis
• Truncus arteriosus
• Hypoplastic left heart syndrome

Acyanotic heart disease – a broad term for any congenital heart defect in which there is a defect in one of the walls separating the chambers of the heart, or an obstruction to one valve or artery. This problem occurs right before or after birth and causes problems for newborns. Some infants with severe acyanotic heart defects have difficulty breathing and sometimes fail to thrive.

Among the congenital heart defects that cause acyanotic heart disease are:
• Patent Ductus Arteriosus (PDA)
• Coarctation of the Aorta
• Atrial Septal Defect (ASD)
• Ventricular Septal Defect (VSD)
• Atrioventricular Septal Defect (AVSD)
• Aortic Stenosis (AS)
• Pulmonary Stenosis (PS)
• Tetralogy of Fallot
• Transposition of the Great Vessels (TGA)
• Total Anomalous Pulmonary Venous Return (TAPVR)
• Truncus Arteriosus
• Tricuspid Atresia
• Hypoplastic Left Heart Syndrome (HLHS)

Common CHD Defects
Many children with CHD suffer from a combination of two or more CHD defects. These defects, or combination of defects, affect newborns and children differently. Generally, there is no set course that this disease follows, as it manifests itself differently from child to child.
Below are several descriptions of cyanotic and acyanotic defects including possible signs and symptoms, treatment options and long-term concerns.

**CYANOTIC**

**Ebstein's Anomaly**

Ebstein's anomaly occurs when the heart's right-sided tricuspid valve forms in an abnormally low position in the right ventricle. This condition leads to a leaky tricuspid valve and an enlarged right atrium. In some cases, blood is pushed through a small hole in the wall between the right and left atrium and deoxygenated blood flows into the body, which can cause cyanosis. In its most extreme form, a baby born with Ebstein's anomaly may have a cardiac murmur, congestive heart failure, difficulty feeding, excessive sweating and poor growth. Infants with significant cyanosis and heart failure may need surgery, while those with milder symptoms can have the surgical intervention carried out later in life. Arrhythmias are common among people with Ebstein's anomaly and an electrophysiologist should evaluate complaints of palpitations.

**Transposition of the Great Vessels**

Here two major arteries leaving the heart are transposed resulting in abnormal blood flow. Normally, blood flows from the right side of the heart to the lungs, to the left side of the heart, throughout the body and back to the right side of the heart. In infants with TGV, blood flows from the right side of the heart to the body and back to the right side of the heart. Simultaneously, blood flows from the left side of the heart to the lungs and back to the left side of the heart. Infants born with TGV are cyanotic and need immediate treatment for this lethal defect. The surgical procedure is almost always done right after birth. No restrictions apply after successful corrective repair.

**ACYANOTIC**

**Atrial Septal Defect (ASD)**

An ASD is a hole in the wall dividing the two upper chambers of the heart. Although this hole can appear in different areas of the wall, one of the most common, atrial septal defects, occurs near the center and is called a secundum ASD. Defects that occur high on the wall between the two atria are called Sinus venousus ASD and are often associated with anomalous pulmonary venous drainage. A hole on the lower wall between the two atria is called ostium primum ASD. Children with ASD usually show no symptoms. Although some ASD's close spontaneously during the first two years of life, the hole should be closed surgically if it is still present in a child by the age of three. Very tiny ASD's generally don't require closure. Occasionally, children born with ASD suffer from palpitations and abnormal heart rhythms later in life on either the right or left atrium. This condition is usually treated with medication or therapy in the electrophysiology laboratory.

**Coarctation of the Aorta**

This defect involves the narrowing of the aorta, the large artery that sends blood from the heart to the rest of the body, and can cause the blood to flow unevenly. Coarctation of the aorta can be associated with other defects such as bicuspid aortic valve, VSD and mitral valve disease. In some cases, where the narrowing of the aorta is very tight, an infant can suffer from heart failure. Symptoms include rapid breathing and failure to thrive. In less severe cases, many children have no symptoms. Severe coarctation must be corrected immediately during the first days of a newborn's life. If the narrowing is not critical, surgery can be postponed until early childhood and will relieve the persistent high blood pressure often associated with this condition. Because some coarctation may recur, children need yearly examinations which include blood pressure measurements in the arms and legs.

**Endocardial Cushion Defect/AV Canal Defect**

Endocardial Cushion Defect/AV Canal Defect occurs when a hole forms in the area where all four heart chambers meet. This hole can range in size from a small opening between the two atria to a large hole in the center of the heart, which causes an opening between the ventricles as well as the atria. In come cases, the mitral and tricuspid valves become joined into a common valve. This defect is often associated with other congenital syndromes such as Down syndrome. Large AV canal defects should be repaired in newborns before they reach six months to guard against the permanent lung damage that can result from prolonged exposure to high blood flow in the pulmonary vessels. In less severe cases, such as primum ASD or mild mitral valve disease, surgery is not usually required until a child reaches school age. Early intervention guards against artery and pulmonary diseases. Patients with repaired Endocardial Cushion Defect/AV Canal Defect will need prophylactic antibiotics prior to dental procedures and will require careful monitoring by a pediatric cardiologist as for occasional leaking of the left sided mitral valve or subaortic stenosis.
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Finding the Best Center for a CHD Child
Early diagnosis of CHD can make an extraordinary impact on clinical outcomes. Such cases can be identified via a chromosomal abnormality, prenatal tests or through an examination of family history that reveals a previous child was born with CHD. Early identification makes it possible for parents to make informed decisions about their child’s care as well as have their child undergo a thorough medical evaluation. Early diagnosis allows for planning for delivery of the baby at a center that provides care in the area of pediatric CHD.

It is important to note that certain defects are so rare that most surgeons won’t have experience in their treatment. When selecting an institution or a surgeon for the treatment of congenital heart disease, it is important to know the number and types of CHD defects the institution or surgeon treats each year and what the outcomes are regarding survival and complications. Families should choose a center or surgeon with the most extensive expertise and best outcomes. Although large institutions generally perform most or all of the pediatric cardiac surgeries, the amount of experience of these institutions varies. Although some may perform many pediatric cardiac surgical procedures yearly the center still may not have specific experience with the complex reconstruction required for certain CHD conditions, while another may perform hundreds per year.

Long Term Prognosis for CHD Patients
The prognosis for children with congenital heart disease has improved remarkably over the past two decades as a result of increasingly successful reparative cardiac surgery. Although, some patients who have residual defects require periodic follow-up and some may eventually require another operation during adulthood, the advances in surgical treatment has led to dramatic increase in the survival of children with serious heart defects. Many patients that have had successful cardiac surgery during childhood continue to do well as adults. Patients who have undergone uncomplicated repairs live full lives and engage in activities unaffected by their cardiac condition. 

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2002 ROSE Client Satisfaction Survey Results

In September and October of 2002, we conducted a survey of our clients, the following is a summary of this survey:

The ROSE Program Commitment
The ING Re ROSE (Reinsurance Outcomes and Service Experts) Program assists clients in controlling risk and reducing costs with catastrophic claims. Our focus includes early intervention and adding value to our clients’ individual health plans.

About the Survey
A satisfaction survey was developed and administered by the ING Re ROSE Program Health Services Consultants and Program Director to assess the following areas:
- Physician Consultants
- Claims/Repricing
- Case Management/Disease Management
- Research and Education
- Overall satisfaction and Suggestions

The feedback we receive from our clients is important to our current and future operation. These survey results are essential in providing direction for a program of excellence in the reinsurance industry.

More than one-half of the survey participants were Managers or Directors of their individual health plan departments. Thirty-nine of 109 participating health plans responded to the survey within the timeframe.

Survey Results
- One hundred percent of those accessing the ROSE Program physicians for consultations stated they were satisfied, or received services near the level to which they had expected.
- Transplant specialty physicians were the most requested specialty for consultations services; closely followed by Cardiology and Oncology.

General Market Insight
- Renal dialysis and IV drugs were the two areas most often causing difficulties in negotiating vendor discounts.
- Diabetes, Asthma, and Perinatal/Neonatal programs were most commonly provided through internal disease management programs.
- Thirty-seven percent of participating health plans would consider using an outside disease management (DM) vendor for comprehensive services; twenty-four percent are doing all DM initiatives internally.
- Requests for assistance with DM programs focused on providing resources to develop programs internally.

Research and Education
- Ninety-seven percent of the participating health plans found the ROSE Program literature review services helpful in accessing relevant information. The majority of respondents felt the timeliness of response met their expectations.
  - Ninety percent of the health plans are interested in participating in future inservice/educational programs offered through ING Re.
  - Two-thirds of the participating health plans have referenced the ROSE Manual at least once in the last year. Overall, 26 percent stated they would use it more often if it were available in an electronic version.

Overall satisfaction and Suggestions
- Ninety-six percent of the participating health plans rated the ROSE Program as Good, Very Good, or Excellent in terms of assistance with reducing costs and controlling risk.

The following programs are in development for the ROSE Program as a result of our clients’ feedback
- Enhanced Case Management and Disease Management Resources
- CEU’s via teleconference offerings
- Availability of additional ROSE Program physician consultants
- On-line ROSE Manual

The purpose of the ROSE Resource newsletter is to provide clients of ING Re with information on a wide variety of topics related to catastrophic medical case management. Case histories, facility highlights and similar articles are intended to serve general information purpose and do not constitute endorsements of facilities, programs or persons by ING Re. The information contained in the articles represents the opinion of the authors and does not necessarily imply or represent the position of the editors or ING Re. Articles are not intended to provide legal, consulting or any other form of advice. Any legal or other questions you have regarding your business should be referred to your attorney or other appropriate advisor.

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