Congenital Diaphragmatic Hernia: The Perinatalist’s Perspective

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OBJECTIVES

After completing this article, readers should be able to:

1. Describe the incidence of congenital diaphragmatic hernia (CDH).
2. Delineate what percent of all CDHs occur on the left hemithorax and what percent are associated with the presence of other malformations.
3. Describe the mortality rate for prenatally diagnosed CDH and what factors affect the presence of associated malformations and chromosomal defects and are the major determinants of mortality.
4. Describe optimal prenatal counseling for a woman carrying a fetus who has CDH.

Introduction
The incidence of congenital diaphragmatic hernia (CDH) is reported to be approximately 1 in 2,000 to 4,000 births. It is believed to result from incomplete fusion of the pleuroperitoneal membrane and passage of abdominal contents into the chest. The small bowel, stomach, spleen, and colon are the most frequently herniated organs, although pancreas, liver, adrenal glands, and kidneys also have been found. Approximately 90% of all CDHs occur on the left hemithorax, and up to 50% of cases are associated with the presence of other malformations.

CDH may be one component of recognized multiple malformation complexes, including chromosomal disorders (trisomy 13, trisomy 18, trisomy 21, tetraploidy, Pallister Killian syndrome [tetrasomy 12 p mosaicism], and Turner syndrome), single gene disorders (Fryns syndrome), and others for which the genetic etiology is uncertain (laterality sequence). Abnormalities in numerous organs other than the lungs have been described, including the heart, kidneys, gastrointestinal tract, abdominal wall, and central nervous system. In addition to more common forms of congenital heart disease, such as tetralogy of Fallot and truncus arteriosus, diminished left ventricular mass has been noted in a number of patients who have left-sided CDH and may have an impact on mortality.

The prognosis of CDH is poor, especially for infants in whom the diagnosis is made in utero. The especially poor prognosis for infants diagnosed in utero may reflect a selection bias because smaller hernias may not be detected by ultrasonography. The mortality rate for prenatally diagnosed CDH ranges from 40% to 90% and is affected by the presence of associated malformations and chromosomal defects. The incidence of stillbirths has been reported to be as high as 35%, with associated anomalies being responsible for most antenatal deaths. In neonates who have isolated CDH, the presence of pulmonary hypoplasia and pulmonary hypertension are the major determinants of survival.

Prenatal Diagnosis
CDH can be diagnosed in utero, although it often is missed on routine ultrasonography that assesses fetal size. The ultrasonographic differential diagnosis includes congenital cystic adenomatoid malformation, mediastinal cystic teratoma, bronchogenic cysts, and neurogenic tumors. CDH can be diagnosed if abdominal organs are seen occupying the thorax. Visualization of fluid-filled bowel with peristalsis at the level of the four-chamber view of the heart also is diagnostic. Associated findings include absence of an intra-abdominal stomach bubble and a shift of the heart and mediastinum away from the side of herniation.

Polyhydramnios frequently is seen and presumably is due to bowel obstruction. Its value as a predictor of adverse outcome has been inconsistent across studies. Pleural effusions and ascites can be associated with CDH and may be due to obstruction of venous return. Fetal thoracentesis and pleuroamniotic shunting have been described in this setting.

The echogenicity of the liver and lung can make the diagnosis of right diaphragmatic hernia very difficult. In one case of right-sided CDH, the interlobar fissure of the herniated fetal liver was misinterpreted as the right diaphragm, and the anomaly was missed. Identification of the gall bladder in the chest can be a helpful hint in the diagnosis of right-sided CDH.

Obstetric management in cases of in utero diagnosis of CDH requires a team approach. The option of pregnancy termination should be offered to the parents before the fetus becomes viable. Prenatal diagnosis plays a key role in preparing both the parents and the perinatal team for the birth of a neonate who has CDH. Intensive fetal surveillance and delivery at a tertiary care center may have a positive impact on the substantial morbidity and mortality associated with this condition. A careful anatomic survey for other malformations should be performed, as should karyotype determination and fetal echocardiography. Serial ultrasonographic evaluation of the fetus should continue throughout gestation to provide assurance of normal growth and to detect the development of polyhydramnios or hydrops. Decreased surfactant components in amniotic fluid have been reported in pregnancies complicated by CDH and may reflect reduced surfactant production by a hypoplastic lung. Delivery before fetal matu-
Prenatal Counseling

MULTIDISCIPLINARY TEAM

Thorough prenatal counseling of a woman carrying a fetus who has CDH requires the coordinated efforts of a multidisciplinary team. This usually begins with the patient’s obstetrician, who is responsible not only for the continuing prenatal care but who also must initiate appropriate referrals to specialists who will play a role both prenatally and postnatally in caring for mother and infant. Affected fetuses should be delivered in facilities that offer both personnel (medical and surgical) who have extensive experience and the necessary resources and technologies that often are employed in treatment. A specialist in maternal-fetal medicine (perinatologist) should be involved. Often, care for the pregnant patient is shared between the obstetrician and the perinatologist, with most of the routine obstetric care carried out in the patient’s local community.

It is not sufficient simply to address the issues arising during pregnancy; the pregnant woman and her partner must have as clear an understanding as possible of potential fetal and neonatal therapies. In addition to providing care to the pregnant woman, the obstetrician and perinatologist are in the unique position to initiate the necessary consultations with the pediatric subspecialists who care for neonates who have CDH.

Consultation with a geneticist and genetic counselor is an important element in the prenatal counseling and a good starting point. Parents have many questions when they learn the fetus has an abnormality, some of which may result from a sense of guilt about their perceived role in “causing” this problem. This meeting provides an opportunity to establish a therapeutic alliance that is likely to continue for a substantial period of time (whether or not the pregnancy results in a viable baby). Genetic counselors often are a tremendous source of ongoing support for the families of children who have congenital anomalies. Discussion of the genetics and embryology of CDH also provides an opportunity to address the risk of recurrence.

Similarly, it is vital for the pregnant woman to meet with a social worker. These professionals can provide emotional support during stressful times, coordinate financial issues, and find housing for the mother after her discharge from the hospital that will allow her to remain close to her baby during his or her stay in the intensive care nursery. These valuable members of the perinatal team often are the single most important source of support during the initial hospitalization.

OVERVIEW OF CDH

The woman carrying a fetus who has CDH may be faced with difficult decisions at many points in time before, during, and after delivery. The neonatologist often can provide the broadest overview of the short- and long-term implications of the condition. Unfortunately there is no proven and universally accepted method of identifying patients who will not be able to support gas exchange independent of extracorporeal membrane oxygenation (ECMO) or mechanical ventilation, although much work is ongoing in this area. This is an important point for parents to understand, so they do not place too much emphasis on early successes, however significant they may appear. The lowest reported mortality remains approximately 40%, and this figure fails to account for all deaths in utero, in the immediate newborn period, and beyond.

Discussion with the parents should start with a description of the physical appearance of the baby after delivery and of what procedures may be undertaken within the delivery room. It is important for the mother to understand that she may not be able to hear her baby cry after delivery or see or hold her baby if vigorous resuscitation is required, and her baby may need to be moved quickly to the intensive care nursery for further care. The immediate issues faced by the neonate who has CDH center on the physiologic effects of pulmonary hypoplasia and pulmonary hypertension. The neonatologist must explain the indications for and sequelae of positive pressure ventilation (both conventional and high-frequency), surfactant therapy, permissive hypercapnia, inhaled nitric oxide, and ECMO.

POTENTIAL THERAPIES

The potential benefits and side effects of both established and experimental therapies should be described in detail, and it is wise to provide the pregnant woman with copies of pertinent consent forms to take with her for review. This will allow ample time for her to read and begin to understand the issues involved and to develop questions that can be answered prior to delivery. This is a far better method of achieving “informed” consent than presenting technically complex information during the emotionally and psychologically challenging hours after birth. Experience has indicated that this approach is extremely helpful both in providing families with a clear understanding of what will be happening to their baby and in allowing the medical staff to balance efforts at bedside care and parent communication. As new therapies (such as liquid ventilation) become available on an investigational basis, they should be added to the list of issues covered during the prenatal consultation.

Meeting with a pediatric surgeon provides the pregnant woman with the opportunity to discuss the details of the surgical approach to CDH and allows the surgeon to provide his or her opinions on controversial issues such as timing of repair. Whereas the neonatologist is unlikely to see the infant who has CDH on a regular basis after discharge from the hospital, the pediatric surgeon probably will play a major role in follow-up. The option of a referral to a center offering fetal therapy also should be addressed. Extensive discussion of potential risks and benefits of all other experimental procedures is mandatory.
ONGOING PROBLEMS
The problems associated with CDH are not limited to the immediate neonatal period. Some neonates have hypoplasia of the pulmonary parenchyma and vasculature that makes it impossible to wean them from ECMO; others have less severe forms that are just as lethal and prevent them from weaning from positive pressure ventilation or otherwise result in late death. Pulmonary hypertension may persist, even after repair of the hernia and successful weaning from ECMO, which may complicate recovery. Gastroesophageal reflux is common in this patient population and may be severe enough to require both medical and surgical therapy. Other types of gastrointestinal problems, such as inability to tolerate enteral nutrition due to functional or anatomic obstruction, may present at any time postoperatively. Affected patients also are at risk for scoliosis, which tends to develop slowly over time and present beyond the neonatal period. Physicians and families should be aware of the possibility of late death from recurrent pulmonary hypertension, intestinal ischemia, and other problems.

The impact of a preterm delivery on a neonate who has CDH is another issue that should be discussed when the diagnosis is made early in pregnancy. The pregnant woman and her partner need to understand that the common problems of prematurity that manifest early in the neonate’s life (surfactant deficiency, high risk of sepsis, intraventricular hemorrhage) greatly complicate care of these infants and substantially reduce their chances for survival. Preterm delivery actually may preclude the use of some potentially beneficial therapies such as ECMO.

PLAN OF CARE
The pregnant woman should be reassured that she and her partner will be provided with all information regarding their baby’s condition and prognosis as it becomes available and that frequent discussions with the attending neonatologist will be necessary to insure that the physicians and parents together determine the best plan of care. Interventions that comprise intensive care should be distinguished from routine newborn care, which includes hydration, thermal regulation, and alleviation of pain and discomfort. The neonatologist should have a frank discussion with the pregnant woman and her partner regarding the indications for initiation, continuation, and termination of intensive care. This preparation may help the parents better understand and cope if the prognosis becomes dismal at any time after birth. Parents should be encouraged to verbalize to the staff any needs that they may have during their baby’s stay in the intensive care nursery.

Many neonates who have CDH are unable to feed for extended periods of time. Because breastfeeding may not be initiated for some time after birth, it is important that any woman interested in eventually breastfeeding be encouraged to pump her breasts starting shortly after birth and store her milk until gavage or nipple feedings can be initiated. Given the potential need for postnatal blood product transfusions, the risks and benefits of transfusion as well as the potential for use of directed donor blood should be discussed. Directed blood donations typically require a minimum of 24 hours for processing and, thus, are not immediately available in the event of an emergency. Coordination with a local blood bank may allow for directed donation prior to delivery, especially when the delivery is planned (induction or scheduled cesarean section).

Parents should be encouraged to identify a pediatrician close to their home who is both willing and able to care for patients who have complex medical conditions. Infants who have CDH will require follow-up with pediatric subspecialists, but they also will require standard pediatric health care maintenance, such as immunizations. It is important that families have a local, readily accessible pediatrician on whom they can rely to handle routine problems. Many pediatricians are willing to meet with prospective parents. Discussion of the need for follow-up in a pediatric developmental clinic also should be undertaken prior to delivery. Many patients who have CDH and survive to be discharged from the hospital require long-term follow-up that involves psychology, nutrition, audiology, speech and language, and physical and occupational therapy.

Although the list of issues for discussion during prenatal consultation may seem lengthy, it is vital that the pregnant woman carrying a fetus who has CDH be advised of the seriousness of the condition and the ramifications of the choices made with each decision. No amount of counseling can prepare a parent fully for the stress of bearing a child who has complex medical problems, but prenatal diagnosis provides an opportunity to build an alliance with the parents and allows them to make informed choices for the care of their fetus/neonate based on the most accurate medical information available.

Delivery Room Management
Because of the relative infrequency of CDH and the potential complexity of the care required by these patients, affected infants should be delivered in a center that has experienced personnel and available therapies. Critically ill neonates can be transported safely, but there is no reason for an infant who has CDH that is diagnosed prenatally to be delivered in a facility that cannot offer the most advanced therapies. This only results in delay of definitive care, increased morbidity and mortality, and at least temporary separation of parents from their baby.

Neonates who have CDH may be very ill from the moment their umbilical cord is clamped and they become dependent on their own lungs and pulmonary vasculature for gas exchange. In cases where the presence of CDH is known prenatally, it is imperative that the team in the delivery room consist of personnel experienced in the immediate resuscitation and stabilization of critically ill neonates. Many affected patients will require positive pressure ventilation in the delivery room. To prevent distension of the gastrointestinal tract and further...
compression of the pulmonary parenchyma, a double-lumen nasogastric or orogastric tube of large caliber is placed to act as a vent. Early intubation is preferable to bag-mask ventilation or continuous positive airway pressure via mask or nasal prongs, although this approach may eliminate any beneficial effect of endogenous nitric oxide produced in the pharynx. Positive pressure ventilation must be applied with great skill because the presence of pulmonary hypoplasia is associated with an increased risk of pneumothorax.

One of the potential pitfalls in delivery room resuscitation of the neonate who has CDH involves interpretation of the presence of diminished (or absent) breath sounds ipsilateral to the hernia. The presence of intestine in the chest will produce this finding, but it also may be indicative of a pneumothorax. Needle thoracostomy may traumatize both the lung, creating an air leak if one did not exist already, and the gastrointestinal tract. When time allows, anteroposterior and lateral radiographs of the chest should be obtained prior to instrumentation of the chest either in the delivery room or nursery. If the neonate does not respond with improved color and heart rate, the decision to continue attempts at stabilization in the delivery room or to move a baby who has marginal oxygen delivery to the intensive care nursery (where chest radiographs and arterial blood gas measurements usually are more readily obtained) is complex and requires experience and expertise.

Summary

Despite the many advances in prenatal, perinatal, and postnatal care, CDH remains a disease associated with substantial morbidity and mortality. Prenatal diagnosis provides the opportunity to provide information and support to the parents and to plan for care of the neonate from the moment of birth to eventual discharge from the hospital.

SUGGESTED READING


Congenital Diaphragmatic Hernia: The Perinatalogist's Perspective
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