Diaphragmatic Anomalies

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Incidence
Congenital diaphragmatic hernia (CDH) refers to a congenital defect in the posterolateral diaphragm at the “foramen of Bochdalek.” It is a relatively common cause of neonatal respiratory distress with an overall incidence between 1:2000 and 1:5000 live births. CDH accounts for about 90% of congenital diaphragmatic defects. Eighty to ninety percent of congenital diaphragmatic hernias occur on the left side. A hernia sac is only present 20% of the time. Retrosternal hernias (Morgagni) are much less common and only account for 2-6% of congenital diaphragmatic defects. Diaphragmatic eventration is even rarer but is a postoperative complication in 1-2% of children undergoing surgery to repair congenital heart defects.

Etiology
The specific etiology of CDH is unknown but it is believed to result from a defective formation of the pleuroperitoneal membrane. In the early weeks of development, the pleural and peritoneal cavities communicate via the paired pleuroperitoneal canals. During the 8th week, the pleural cavity becomes separated from the peritoneal cavity by the developing pleuroperitoneal membrane. If the pleuroperitoneal membrane fails to develop, closure of the pleuroperitoneal canal is incomplete and a posterolateral diaphragmatic defect results. A newer hypothesis has arisen from the nitrofen rat model of CDH. Electron microscopy of these nitrofen exposed rat embryos suggests that CDH results from a defective development of the “posthepatic mesenchymal plate” which also contributes to closure of the pleuroperitoneal canal. Although familial cases are reported, most cases of CDH are sporadic. CDH is associated with trisomies 18, 21, and 23 but a specific genetic etiology has yet to be identified.

Morgagni hernias result from failure of the sternal and crural portions of the diaphragm to fuse at the site where the superior epigastric artery traverses the diaphragm. Morgagni hernias are associated with congenital heart disease and trisomy 21. A variant of the retrosternal hernia is associated with the pentalogy of Cantrell which includes: omphalocele, inferior sternal cleft, severe cardiac defects (including ectopia cordis), diaphragmatic hernia and pericardial defects. The diaphragmatic defect results when the septum transversum fails to develop in the embryo.

Eventration of the diaphragm may be either a congenital or acquired lesion. Neonatal eventration may be due to defective central development or enervation of
the diaphragm. It may also result from a traction injury to the nerve roots of the phrenic nerve during traumatic delivery. Eventration most often results from iatrogenic phrenic nerve injury complicating cardiac or mediastinal surgery.

**Clinical Presentation**

Thirty percent of fetuses with CDH will be stillborn. If born alive, neonates with CDH usually present with respiratory distress. The onset of respiratory distress can be immediate at the time of delivery or may be delayed for 24-48 hours. Only 10% of patients with CDH present beyond the neonatal period. These children (or adults) may present with vague gastrointestinal symptoms or may be completely asymptomatic discovered only as an incidental finding. Rarely, an older child with CDH may present with life-threatening respiratory and cardiopulmonary distress. Hemodynamic instability may result from severe mediastinal shift caused by a massively distended, intrathoracic stomach. Volvulus and intestinal obstruction are exceedingly uncommon, but reported, presentations of CDH beyond the neonatal period.

The initial signs of CDH in the neonate include tachypnea, grunting respirations, chest retractions, cyanosis, and pallor. Physical exam may reveal a scaphoid abdomen, shifting of the heart sounds to the right (i.e., left hernia), and bowel sounds within the chest. Breath sounds are decreased bilaterally, but are often more diminished on the side of the hernia. Disparity between preductal and postductal pulse oximetry may confirm the presence of right-to-left shunting and persistent fetal circulation. The differential diagnosis of CDH includes cystic adenomatoid malformation, cystic teratoma, pulmonary sequestration, bronchogenic cyst, neurogenic tumors and primary lung sarcoma.

The majority of children with Morgagni (retrosternal) hernias are asymptomatic. Diagnosis is often not made until adulthood. Children with this lesion may present with recurrent respiratory infections, coughing, vomiting or epigastric pain/dyscomfort. Intestinal obstruction and bowel ischemia/necrosis may result from incarceration of bowel within the hernia sac.

Eventration in the neonate can be asymptomatic but most present with tachypnea, respiratory distress and pallor. Chest physical signs include ipsilateral dullness to percussion and unilateral or bilateral diminished breath sounds. The point of maximal cardiac impulse is shifted away from the side of the lesion. Neonates with diaphragmatic eventration have difficulty sucking and tire easily with feedings. This combination often causes inadequate weight gain. Older children may present with recurrent pneumonia or upper gastrointestinal symptoms. The differential diagnosis of eventration includes tumors, bronchogenic cysts, pulmonary sequestration, pulmonary consolidation, and pleural effusion.

**Diagnosis**

Prenatal diagnosis of CDH can be made by fetal ultrasonography as early as 25 weeks gestation. CDH suspected in a newborn infant with respiratory distress is confirmed by “babygram” performed simultaneously with resuscitation. The common radiographic findings of left-sided CDH include air/fluid filled loops of bowel in the left chest, mediastinal shift, and the stomach gas bubble within the chest (Fig. 72.1). A nasogastric tube may appear to coil in the chest if the stomach lies
within the thorax. Right-sided CDH (Fig. 72.2) is often more difficult to discern and on x-ray examination may resemble lobar consolidation, fluid within chest, or diaphragmatic eventration.

For Morgagni hernias, posteroanterior and lateral chest radiographs often demonstrate an air-fluid filled structure located immediately posterior to the sternum (Fig. 72.3). This diagnosis is frequently made when gastrointestinal symptoms lead to a contrast study that demonstrates herniated stomach, small bowel or colon within the chest.

Chest radiographs in patients with eventration demonstrate an elevated hemidiaphragm although this finding can be obscured by intubation and positive pressure ventilation. The diagnosis is confirmed by ultrasonographic or fluoroscopic demonstration of paradoxic diaphragmatic motion. Occasionally, computed tomography may be necessary to distinguish eventration from other mass lesions.

**Pathology/Pathophysiology**

Congenital diaphragmatic hernia has a complex pathophysiology. Lung hypoplasia occurs as a direct consequence of progressive compression of the developing lungs by herniated viscera. The severity or degree of pulmonary hypoplasia depends upon both the duration and timing of visceral herniation into the chest. Hypoplasia is most severe on the ipsilateral side but occurs on both sides. Gas exchange within these grossly small lungs is limited by a reduced functional area, decreased number of bronchial divisions, a reduced number of mature alveoli, and surfactant deficiency. Alveoli of CDH lungs are immature and have thickened intra-alveolar septa.
Fig. 72.2. Neonatal chest x-ray demonstrating the much rarer right-sided diaphragmatic hernia with bowel entering the right hemithorax around the liver. Small arrows indicate bowel loops, large arrows demonstrate nasogastric tube and endotracheal tube, while medium arrows indicate the cannulae of an extracorporeal life support system.
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The pulmonary vasculature is best characterized by the presence of increased muscularization of the pulmonary arterioles. The abnormally muscularized and reactive pulmonary vasculature bed contributes to persistent fetal circulation, pulmonary hypertension and acute respiratory failure. Left ventricular hypoplasia is also present in CDH and may adversely affect cardiopulmonary function.

The hypoplastic lungs in patients with CDH are functionally immature and have limited capability for gas exchange. In many cases, alveolar function is inadequate. Hypoxemia, hypercarbia and acidosis can quickly develop causing further deterioration in pulmonary function. The overly muscularized pulmonary artery tree quickly vasoconstricts in response to reduced oxygen tension and acidosis. This vasoconstrictive response is both exaggerated and sustained causing pulmonary hypertension. Pulmonary hypertension in the newborn with CDH may effect a return

Fig. 72.3. Lateral chest x-ray demonstrating the central, anterior diaphragmatic herniation generally referred to as a hernia of Morgagni.
to the fetal pattern of circulation with right-to-left shunting across both the ductus arteriosus and foramen ovale. Intrapulmonary shunting also occurs. Right-to-left shunting further limits gas exchange exacerbating hypoxia, hypercarbia and acidosis. A viscous cycle ensues which can rapidly progress to hypotension, shock and cardiorespiratory failure/arrest.

Morgagni hernias do not typically produce the pathophysiologic problems encountered with the posterolateral diaphragmatic defect. Gastrointestinal obstruction or ischemia and their associated pathophysiologic changes are the presenting features of this lesion when symptomatic.

Unilateral diaphragmatic eventration results in abnormal respiratory mechanics. Ventilation may be ineffective due to a paradoxical motion of the ipsilateral diaphragm during inspiration. Contralateral lung ventilation is also impaired. During inspiration, the mediastinum shifts toward the contralateral side reducing the effective tidal volume on that side.

**Treatment**

The treatment of CDH depends upon the time of diagnosis and the clinical presentation. Institutional expertise and the availability of advanced life support techniques (i.e., extracorporeal membrane oxygenation (ECMO)) also influence the management strategy of these infants. Once considered a surgical emergency, CDH is now managed by a delayed surgical approach. Preoperative stabilization and control of pulmonary hypertension is advised. Mechanical ventilation techniques which avoid barotrauma are helpful. High frequency and oscillatory ventilation, nitric oxide administration, surfactant replacement, and ECMO are interventions and therapies readily employed to manage these infants. Fetal interventions (i.e., fetal repair, tracheal ligation), liquid ventilation and lung transplantation are currently experimental therapies investigated at only a few specialized centers.

Neonates with CDH may present with severe respiratory distress that requires aggressive resuscitation to include endotracheal intubation, neuromuscular blockade, and positive pressure ventilation. Initial ventilation should attempt to maintain preductal saturation at or above 90% using the lowest airway pressures capable of providing oxygenation. Barotrauma to the hypoplastic lungs must be minimized. Orogastric or nasogastric decompression is used to minimize bowel distension which can further compromise respiratory function. Echocardiography is performed to evaluate for associated cardiac anomalies and assess the severity of pulmonary hypertension and shunting. Inotropic agents are used to augment left ventricular function and to raise systemic pressure minimizing right-to-left ductal shunting. Hypervolemia and hypovolemia must be avoided. Hypoxemia, hypercarbia and acidosis must be identified and promptly corrected when present. Bicarbonate may be used to treat acidosis.

Several pharmacologic interventions may be useful in the perioperative management of neonates with CDH. Inhaled nitric oxide, a potent pulmonary vasodilator, may successfully control refractory pulmonary hypertension. Surfactant replacement therapy for CDH is controversial but may be beneficial in improving gas exchange. ECMO is indicated for infants with CDH and respiratory failure that cannot be managed with conventional therapy. Candidates must have a reasonable chance for survival with no major, nonreversible anomalies. ECMO can be used as a preoperative
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or postoperative therapy. Some centers perform diaphragmatic repair while on ECMO.

Surgical repair of congenital diaphragmatic hernia is delayed until after preoperative stabilization and resolution of pulmonary hypertension. The repair should be performed efficiently and expeditiously to minimize operative stress. The abdominal viscera are reduced from the chest via a transabdominal approach. The diaphragmatic defect is closed primarily if possible, or a prosthetic patch may be inserted for larger defects. Tube thoracostomy is optional. The abdominal wall is stretched prior to closure to increase the capacity of the abdominal cavity. Rarely, abdominal closure is achieved with a prosthetic silo or by creating skin flaps and a ventral hernia.

Postoperative ventilator management can be difficult. Chest compliance is decreased after repair and surgical stress can precipitate intense pulmonary vasoconstriction and pulmonary hypertension with recurrent fetal circulatory pattern. Respiratory failure can occur abruptly and ECMO may be required as a rescue therapy. Postoperative ventilation strategies should attempt to minimize barotrauma while maintaining normal \( PO_2 \), \( PCO_2 \), and \( pH \).

Morgagni hernias are surgically repaired via a transabdominal approach. Primary closure of small defects is preferred, but larger defects may require prosthetic patch closure. The treatment of symptomatic eventration is also surgical. The diaphragm on the affected side is plicated via either a transthoracic or transabdominal approach. Plication effectively immobilizes the flaccid diaphragm, reducing the paradoxical movement and mediastinal shift that occurs with respiration.

**Outcomes**

Despite the many therapeutic options available to manage patients with CDH, the overall survival remains about 60%. Institutional variation in survival is great and ranges from 25-80%. CDH accounts for 4-10% of neonatal deaths occurring as a result of congenital anomalies. ECMO improves survival by 15-20% at institutions employing this therapy.

Most survivors of CDH are generally healthy and are without respiratory problems. Long-term respiratory status is dependent on the severity of pulmonary hypoplasia at birth and the degree of lung injury sustained during the perinatal period. Gastroesophageal reflux is common in survivors of CDH repair; surgical intervention may be required in 10-15%. Survivors of CDH are at increased risk for neurodevelopmental delays. The overall incidence of neurologic abnormalities is 10-45%.

Surgical results from repair of Morgagni hernias are in general excellent. Complication rates are low. Morbidity and mortality is usually due to associated cardiac anomalies which are frequently found in these children.

The perioperative morbidity and mortality of diaphragmatic plication for eventration is low. Complications are mostly secondary to prolonged mechanical ventilation and/or cardiac dysfunction associated with an underlying cardiac pathology. Plication results in immediate improvement in pulmonary mechanics but long-term respiratory function depends on lung damage prior to plication surgery.
Selected Readings

