Congenital Diaphragmatic Hernia

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Key Points
1) Defect in the development of the diaphragm.
2) Main concern is the pulmonary hypertension and hypoplasia which affects patient outcome and survival.
3) Repair is not emergent and is institutional dependent.

Congenital Diaphragmatic Hernia

This is a condition that is characterized by a defect in the diaphragm, allowing intestinal contents to travel into the thoracic cavity and interfering with normal development of the lung. This leads to pulmonary hypoplasia and pulmonary hypertension. Through advanced neonatal intensive care and select application of extracorporeal membrane oxygenation, children can survive to correction.

1. Epidemiology
    a. 0.8–5 per 10,000 births
    b. May be higher in stillborns (1/3 of CDH)
    c. Higher in males
    d. Two percent in first-degree relative
    e. Eighty percent left-sided
    f. Linked to phenmetrazine, thalidomide, quinine, nitrofen, vitamin A deficiency

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2. Associated anomalies are present in 28–50% of infants with CDH and in 100% of stillborn infants with CDH. Common defects are neural tube, cardiac, and midline defects, such as Pentalogy of Cantrell. CDH is more common in Trisomy 13, 18, and 21.
3. Pathophysiology
    a. Diaphragm develops at 4 weeks of gestation.
    b. Midgut returns to abdomen week 9–10.
    c. Defect thought to be caused by impaired development of the post hepatic mesenchymal plate.
    d. If diaphragm open, herniates to thoracic cavity.
    e. Occurs by week 12.
    f. Malrotation occurs.
    g. Hernia sac present in 10–15%.
    h. Left more common than right.
    i. Bochdalek (posterior lateral) hernia is more common (70–75%) than Morgagni (anterior) hernia (13%) (Fig. 1).
    j. Can contain liver, spleen, GI tract, and kidney.
    k. Associated pulmonary hypoplasia
        i. Total pulmonary vascular bed reduced
        ii. Decreased bronchial branching
        iii. Decreased alveoli
        iv. Decreased total vascular diameter and number of vessels
        v. Alveolar vessels remodel and develop medial hyperplasia
        vi. Smooth muscle in alveolar vessels
        vii. Bilateral lungs affected

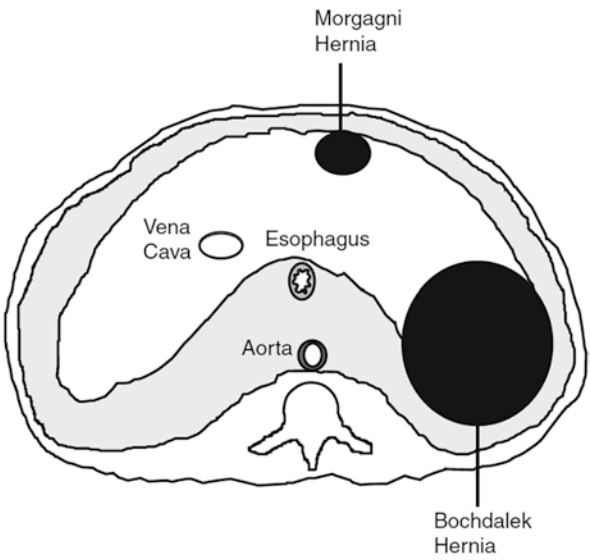

Fig. 1 Types of congenital diaphragm hernias. Morgagni hernias are defects in the anterior diaphragm, while the more common Bochdalek hernias are in the posterolateral diaphragm. (Source: Christopher Coppola)

l. Factors resulting in persistent fetal circulation in CDH

   i. Pulmonary hypoplasia
   ii. High pulmonary vascular resistance
   iii. Hypoxia
   iv. Acidosis
   v. Stress

m. Diagnosis

   i. Prenatal U/S (CDH, polyhydramnios)
   ii. Fetal MRI—calculate lung-to-head ratio (LHR) and observed to expected LHR
   iii. Respiratory distress after delivery
   iv. Scaphoid abdomen/funnel chest
   v. X-ray with NGT in chest
   vi. Ten to twenty percent with delayed presentation

n. Predicting severity of CDH

   i. Prenatal diagnosed CDH as a group has worse prognosis than CDH diagnosed after birth.
   ii. Presence of cardiac defects worsens mortality.
   iii. Smaller pulmonary artery size (which can be quantified by the McGoon Index) carries worse prognosis.
   iv. Defects large enough to allow herniation of the stomach/liver into the chest have worse prognosis.
   v. Right-sided hernia represents more severe disease occurring earlier in gestation.
   vi. Prenatal LHR calculated:

      1. LHR < 0.6 carries poor prognosis.
      2. LHR 0.6–1.0 is a gray zone and careful management.
      3. LHR > 1.35 suggests survival.

o. Prenatal care

   i. When a patient is prenatally diagnosed with CDH, parents should receive genetic counseling, and karyotyping should be performed on the fetus to detect any associated anomalies or syndromes.
   ii. Prenatal care should be provided to allow delivery to occur as close as possible to term to minimize the effect of premature delivery on an already compromised pulmonary function.
   iii. When possible, delivery should occur in a center with access to high-level NICU care and ability to perform ECMO, if it becomes necessary.

p. Treatment

   i. Initial resuscitation

1. Patient should be assessed by neonatologist and surgeon after delivery.
2. Consider intubation immediately after delivery to prevent air filling the GI tract and impairing lung function further.
3. IV fluid should be started and used judiciously, 20 ml/kg as a goal to begin, if unresponsive, consider vasopressors early to prevent pulmonary edema.
4. Nasogastric tube placed to decompress the stomach, which may be in the chest, and to reduce the volume of gas and fluid in the GI tract.
5. Pulmonary function should be monitored for signs of pulmonary hypoplasia and failure. The patient may worsen over the course of the initial 24–48 h "honeymoon" period.
6. Obtain chest X-ray to detect abdominal organs in the chest and degree of hypoplasia.
7. When intubation and mechanical ventilation is needed, use a strategy of "gentle ventilation" to avoid barotrauma to the alveoli.

    a. Conventional ventilation with peak pressures less than 25 cm H_2O.
    b. High-frequency/low-pressure ventilation, with use of an oscillating/jet ventilator when needed.
    c. Goal of preductal saturation 85–95%, post-ductal saturations >70%.
    d. Permissive hypercapnia with PCO_2 45–60 mmHg.
    e. Medications to reduce pulmonary hypertension.

        i. Inhaled nitric oxide
        ii. Sildenafil
        iii. Bosentan (Tracleer)
        iv. Epoprostenol (Flolan)
        v. Tolazoline
        vi. Inhaled prostacyclin

    f. When gentle ventilation and all adjuncts fail to provide adequate oxygenation, evaluate patient for suitability of ECMO therapy.

ii. Surgical repair of CDH

1. Timing of operation: multiple options to timing.

    a. In stable and uncomplicated case—Repair when weaned on FiO_2 and minimal ventilator settings.
    b. In complicated case—requiring ECMO.

        i. "Early" repair—Once stable on ECMO, repair within first 24–48 h before develops edema and risk of bleeding increases.
        ii. "Delayed" repair—While on ECMO and beginning to wean, once close to coming off ECMO, repair prior to decannulation in case develops pulmonary hypertensive crisis from stress of surgery.
        iii. "Late" repair—Recovered and off ECMO, then repair.

2. Technique

   a. Subcostal incision or thoracotomy: On the left, the approach via abdomen allows for convenient reduction of viscera into abdominal cavity. For a right-sided CDH, thoracotomy avoids the difficulty of working around the liver that is encountered with an abdominal approach.

      i. Reduce hernia contents back into abdomen.
      ii. Excise hernia sac when present as it can interfere with visualization of lungs or viscera while closing defect.
      iii. Identify muscular rim of diaphragm circumferentially. Some areas along posterolateral ribs and mediastinal structures may have no diaphragm and require careful placement of sutures.
      iv. Determine if primary closure is possible or if a patch of exogenous material (artificial or biologic mesh) or muscle flap will be needed (approximately one-third of cases).
      v. Consider addressing malrotation/non-fixation of bowel, if present, by performing a Ladd's procedure.
      vi. Close abdomen if possible. If there is insufficient domain in abdomen after reducing viscera from chest to abdomen, a silo or mesh closure of abdomen may be needed on a temporary basis.

q. Outcome

   i. Through the twentieth century, CDH has historically had 50% survival; however advancements in neonatal intensive care unit techniques and select use of ECMO have produced a survival range of 39–95% (mean, 69%), depending on setting of care.
   ii. There is a risk of both early and late recurrence. Early recurrence can be due to technical error and increased abdominal pressure. Placement of mesh reduces tension across the repair, but approximately one-third of patients will need a late revision of repair due to recurrence of hernia or due to tension on the thoracic wall and deformation of the ribcage.
   iii. Chronic lung disease may result from the accompanying pulmonary hypoplasia.
   iv. Gastroesophageal reflux is common after CDH repair, and some patients will require an anti-reflux procedure.
   v. Development/cognitive deficits occur and can be due to associated anomalies or as sequela from ECMO.

Additional Notes

Congenital diaphragmatic hernia is a life-threatening condition of the newborn in which a gap in the diaphragm allows abdominal contents to enter the chest. There are varying degrees of severity, and often the accompanying pulmonary hypoplasia

and pulmonary hypertension are the main determinants of long-term outcome. Historically, there has been a 50% mortality rate, but the recent reports show a survival rate that approaches 90% [1]. High-volume centers treating larger numbers of patients achieve the highest survival and will have greater resources for more intense therapy like ECMO. The majority of advances have been in neonatal intensive care and pharmaceutical therapy, but some operative alternatives are thoracoscopic approach and creation of a dome shape to the repair [2, 3]. Elevated intraabdominal pressure measured via the bladder after repair is predictive of length of mechanical ventilation and delay before enteral feeding [4]. Duration of mechanical ventilation and duration of nitric oxide treatment predict the 11% of survivors who will require pulmonary hypertension medication after discharge [5].

Study Questions

1. Question 1: What is the most common type of CDH?

   a. Morgagni
   b. Bilateral
   c. Bochdalek
   d. Anterior

Answer: (c.) Bochdalek. The most common type of CDH is Bochdalek (80%), the most common type of defect is posterior lateral. Anterior defects or Morgagni are more likely delayed in presentation and less common. Bilateral CDH are rare and usually fatal.

2. Question 2: What are the goals of initial management in a newborn with CDH?

   a. Bag ventilation, monitor to see how baby does, feed when ready.
   b. Immediate intubation, gastric decompression, gentle ventilation.
   c. Bag ventilation, fluid resuscitation as need, up to 60 ml/kg.
   d. Gastric decompression and high-frequency ventilation with high pressures.

Answer: (b.) Immediate intubation, gastric decompression, gentle ventilation. Goal in CDH is to intubate early to avoid GI filling with air, gastric decompression, gentle ventilation with low-pressure ventilation and high frequency. Do not want high-pressure ventilation to prevent barotrauma in already compromised lungs.

3. Question 3: What can predict poor outcome or difficult management CDH?

   a. LHR = 1.0, spleen in chest, $SpO_2 = 90\%$.
   b. LHR 1.35, stomach in abdomen, $PCO_2 = 40$.
   c. LHR = 1.0, stomach in chest, peak vent pressure 20 mmHg.
   d. LHR <0.6, liver in chest, stomach in chest, $PCO_2 > 70$.

Answer: (d.) LHR < 0.6, liver in chest, stomach in chest, PCO_2 > 70. Lung-to-head ratio <0.6 portends a bad prognosis, liver and stomach in the chest can be risk for poor outcome, high PCO_2 can suggest pulmonary atresia is more severe and poor prognosis.

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