

American Pediatric Surgical Association

Prenatal Counseling Series

Congenital Diaphragmatic Hernia



APSA

American Pediatric
Surgical Association

Saving Lifetimes™

from the

Fetal Diagnosis and Treatment Committee

of the American Pediatric Surgical Association

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and Erin Perrone, MD

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Congenital Diaphragmatic Hernia

Congenital Diaphragmatic Hernia (CDH) refers to congenital absence of at least one diaphragm, which results in abnormal displacement of typically abdominal structures into the fetal chest. Morbidity and mortality of infants born with this congenital anomaly is dependent on many factors including possible coexistence of congenital heart disease, other congenital or genetic anomalies, degree of herniation of structures into the fetal chest, in particular the liver, degree of pulmonary hypoplasia and pulmonary hypertension. Sac-type congenital diaphragmatic hernia (CDH) is a variant in which there is some residual diaphragm membrane with improved prognosis.



Source: Colorado Fetal Care Center

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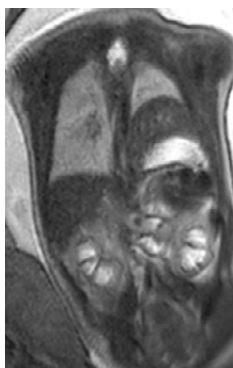
Differential Diagnosis

Differential diagnosis of congenital diaphragmatic hernia includes: Morgagni hernia, congenital hiatal hernia, pulmonary agenesis, pulmonary aplasia, pulmonary hypoplasia, pericardial teratoma, congenital pulmonary airway malformation (CPAM), bronchopulmonary sequestration (BPS), mediastinal lymphatic malformation, pericardial teratoma, bronchogenic cyst, neurenteric cyst and pleuropulmonary blastoma.

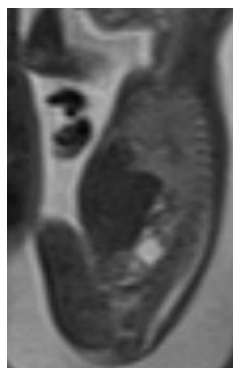
Representative MRI images of some of the differential diagnosis



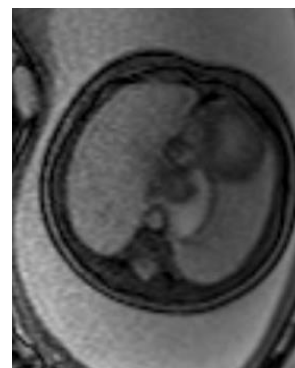
Bochdalek hernia



Sac-type hernia



Morgagni hernia



Hiatal hernia

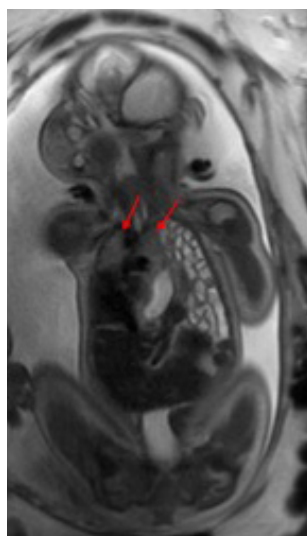


Figure 1

Fig.1: Fetal MRI with Single Shot Fast Spin Echo coronal image demonstrating fluid filled stomach and bowel loops in the left chest. Both the left and right lung are displaced superior and to the right and are small in size (arrows).

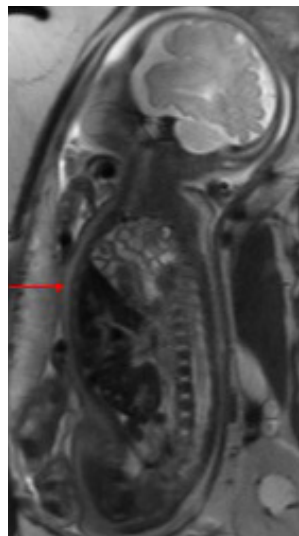


Figure 2

Fig. 2: Fetal MRI with SSFSE parasagittal image demonstrating bowel loops and left lobe of the liver (arrow) in the left thorax.

Images courtesy of Kimberly Dannull, MD, Colorado Fetal Care Center

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Initial Evaluation

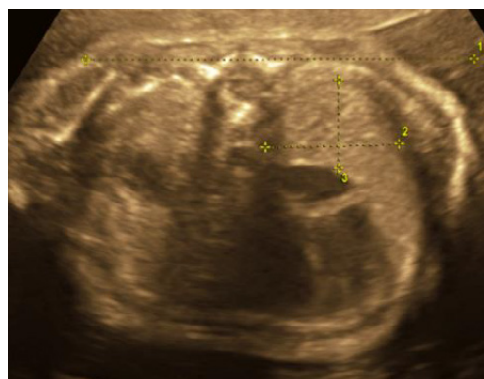
Obstetrical Ultrasound
Fetal echocardiography
Fetal magnetic resonance imaging

Prenatal Imaging

The goal of prenatal imaging is to exclude alternative diagnoses, evaluate for additional congenital anomalies, identify location and contents of the hernia, predict outcome and guide perinatal and postnatal management.

Obstetrical Ultrasound

- Evaluate fetal growth
- Evaluate the amount of amniotic fluid. The herniated stomach can result in polyhydramnios, which can predispose to preterm labor and delivery
- Evaluate anatomy for the presence of other potential congenital anomalies
- Evaluate for hydrops: pleural fluid, ascites, pericardial fluid, skin edema, placentomegaly
- Measure the Lung-to-Head Ratio (LHR) (Length x width of the lung opposite the side of the hernia, divided by the head circumference with formula $L \times W/HC$)
 - Mild pulmonary hypoplasia: LHR >1.4
 - Moderate pulmonary hypoplasia: LHR 1-1.4
 - Severe pulmonary hypoplasia: LHR <1
- Observed-to-expected LHR should be calculated
 - Mild CDH: O/E LHR $>35\%$
 - Moderate: O/E LHR 25-35%
 - Severe: O/E LHR $<25\%$
- Right- vs left-sided CDH: Most experts feel that right-sided diaphragmatic hernia has a same or worse prognosis compared to left-sided defect with similar parameters, with the exception of liver position (the liver is always up with right-sided defect as it is the only organ immediately beneath the right diaphragm.)



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Fetal Echocardiogram

- Evaluate cardiac position, structure and function (the combination of congenital heart disease and CDH drastically increases mortality, particularly if univentricular anatomy)
- **Modified McGoon Index (MMI) may be obtained to evaluate risk for pulmonary hypertension: (diameter of the left pulmonary artery plus the right pulmonary artery divided by the aorta at the level of the expected diaphragm (LPA + RPA/Aorta). A MMI <1 indicates high risk for pulmonary hypertension.**
- Perform maternal hyperoxygenation response

Fetal Magnetic Resonance Imaging (MRI)

- Fetal MRI is typically obtained at 24 weeks or around the time of diagnosis to fully evaluate the anatomy and exclude potential alternative or coexistent diagnosis. Some fetal centers will advocate for an optional 34 weeks gestation MRI, which gives a more accurate assessment of lung volumes and better visualization of the pulmonary arteries to predict the risk for pulmonary hypoplasia and pulmonary hypertension respectively.
- Obtain lung volumes utilizing volumetric software:
 - Calculate Total Lung Volume (TLV). The TLV is the simple summation of the volume of the left and right lung (left lung volume + right lung volume). A TLV < 20 ml at 34 gestational weeks indicates a poor prognosis.
 - Calculate Percent Predicted Lung Volume (PPLV). The expected lung volume is calculated by subtracting mediastinal volume (MV) from the thoracic volume (TV). PPLV is the total lung volume divided by the expected lung volume (PPLV= TLV/TV-MV). A PPLV of < 15% indicates a poor prognosis.
 - Similar to prenatal ultrasound and echocardiogram, a modified McGoon index (MMI) may be quantitated

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Prenatal Counseling and Postnatal Considerations

- After a comprehensive review of the fetus with obstetrical ultrasound, fetal MRI, fetal echocardiography and amniocentesis for karyotyping, then comprehensive counseling can be performed, typically including MFM, pediatric surgeon, neonatologist, pediatric cardiologist and pediatric radiologist.
- All cases of CDH should be delivered in a tertiary care center near a neonatal intensive care unit (NICU)
- Depending on the degree of pulmonary hypoplasia, delivery with ECMO standby should be considered

Management of Pregnancy and Delivery Planning

- Biweekly follow up beginning at 30 weeks' gestation: there is a 10% risk of intrauterine fetal demise during the third trimester even in cases with no other abnormalities except the CDH.
- All CDH babies should be considered high-risk, regardless of prenatal imaging parameters.
- Cesarean section is only indicated for obstetrical considerations.
- Scheduled delivery when full-term (37-38 weeks) should be considered. Many, but certainly not all, specialists believe scheduled delivery is necessary, but true readiness is important - and few centers truly are ready 24/7.
- Delivery should occur close to a Level 3 NICU with access to pediatric surgery and ECMO.

Consideration for Fetoscopic Endoluminal Tracheal Occlusion (FETO)

Currently, FETO is offered through a handful of centers within the U.S. who participate in the North American Fetal Therapy Network (NAFTNET) FETO consortium. FETO is offered to severe left-sided CDH (identified as O/E LHR<25%) via participation in the TOTAL trial.

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Postnatal Course

The postnatal course can be variable depending on the degree of pulmonary hypoplasia and pulmonary hypertension.

Risks to discuss with families during prenatal consultation:

- The following long-term outcomes should definitely be discussed during prenatal consultation for a fetus with CDH:
 - Recurrence of hernia
 - Death
 - Long-term O₂ supplementation
 - Ventilator dependency
 - Neurological problems
 - GERD
 - Asthma, reactive airway disease
 - Hearing loss
 - G tube dependence
 - Failure to thrive
 - Chest wall deformities
 - Intestinal obstruction
 - Diuretic dependence

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Complication rates for CDH requiring ECMO vs not requiring ECMO

OUTCOME	Rate for ECMO CDH	Rate for non-ECMO CDH
Prolonged ventilator dependence (> 1 month)	25-75%	Up to 10%
Long-term supplemental oxygen need (> 6 months)	25-75%	Up to 10%
Asthma or reactive airway disease as a school-aged child	25-75%	10-50%
Neurologic impairment: Learning difficulties	50%	5-20%
Neurologic impairment: Unable to attend regular school	10-20%	1-5%
Long-term hearing loss: Mild loss without need for medical assistance	Up to 50%	1-5%
Long-term hearing loss: Requiring hearing aid	5-10%	1%
Long-term hearing loss: Hearing loss impacting speech development	5-10%	1%
GERD: Managed with medication	75%	Up to 75%
GERD: Managed with fundoplication	20%	Up to 10%
Failure to thrive resulting in need for gastrostomy placement	Up to 50%	Up to 10%
Intestinal obstruction	5-10%	5-10%
Chest wall deformities: Mild	10-20%	10-20%
Chest wall deformities: Requiring surgery	Up to 10%	Up to 5%
Scoliosis	Up to 20%	Up to 20%
CDH recurrence after repair	10-50%	5-20%
Death	20-50%	5-20%

Courtesy of Loren Berman, MD

Berman L, Jackson J, Miller K, Kowalski R, Kolm P, Luks F. Expert surgical consensus for prenatal counseling using the Delphi method. J Pediatr Surg. 2017 Nov 28. pii: S0022-3468(17)30805-9.

Follow-up and Long-term Issues

It is strongly recommended that CDH babies follow up in a multidisciplinary clinic including pediatric surgery, pulmonary, nutrition and developmental pediatrics due to the abundance of potential long-term sequelae as detailed above.