Congenital Chest Malformations: A Multimodality Approach with Emphasis on Fetal MR Imaging

Pedro Daltro, MD • Heron Werner, MD • Taisa Davaus Gasparetto, MD • Romeu Cortes Domingues, MD • Leise Rodrigues, MD • Edson Marchiori, MD, PhD • Emerson Leandro Gasparetto, MD, PhD

Congenital chest malformations can range from small and asymptomatic entities to large space-occupying masses that require immediate surgical treatment. They may affect the foregut, pulmonary airway, and vasculature. Hybrid conditions are commonly seen, with interrelated chest malformations having various radiologic and pathologic features. An understanding of the in utero complications associated with fetal chest masses is essential for appropriate monitoring during pregnancy, treatment recommendations, and delivery management. Technologic advances have greatly improved the diagnosis of fetal anomalies. Congenital chest malformations are usually evaluated in the prenatal period with fetal sonography, but fetal magnetic resonance (MR) imaging is a well-established modality that is used as an adjunct technique in difficult diagnostic situations. MR imaging can provide excellent tissue contrast with more accurate analysis of the fetal anatomy and superior differentiation between the abnormalities and adjacent structures, thereby allowing early planning of prenatal management.
Introduction
Congenital chest malformations are rare, and they may involve the lung parenchyma, bronchi, arterial supply, and venous drainage (1). The most common congenital chest anomalies include congenital cystic adenomatoid malformation (CCAM), congenital diaphragmatic hernia (CDH), bronchopulmonary sequestration (BPS), congenital hydrothorax, and congenital lobar emphysema. Less common entities include congenital high airway obstruction syndrome (CHAOS), congenital bronchogenic cyst, bronchial atresia, pulmonary arteriovenous malformation (PAVM), congenital pulmonary lymphangiectasia, pulmonary hypoplasia-aplasia, mediastinal teratoma, and mediastinal lymphangioma (2). In addition, some chest malformations occupy the thorax and alter normal pulmonary development, causing hypoplastic lung formation. These pulmonary abnormalities are not mutually exclusive, since abnormalities frequently occur together as hybrid conditions (3–6).

To evaluate congenital chest malformations, all thoracic structures must be systematically analyzed to define the origin of the malformation. Prenatal sonography remains the primary imaging modality for evaluating the fetus, because it is safe, largely accessible, and inexpensive. However, in cases of inconclusive sonographic findings, the use of fetal magnetic resonance (MR) imaging helps identify potentially lethal pulmonary abnormalities (3–5). In this article, we briefly review fetal MR imaging technique and the normal lung anatomy, and we discuss and illustrate the most common congenital chest abnormalities seen at fetal MR imaging.

Fetal MR Imaging Technique and Normal Lung Anatomy
The MR imaging sequences used to evaluate the fetal chest include fast sequences such as half-Fourier single-shot turbo spin-echo, fast single-shot echo, and free induction steady-state precession sequences. T2-weighted images are the most useful for evaluating the lung anatomy. The lungs typically contain a significant amount of alveolar fluid, which is homogeneously hyperintense relative to the chest wall muscle on T2-weighted images. In cases of lung compression, the amount of alveolar fluid is decreased, resulting in a more hypointense signal. Other structures that can be evaluated with T2-weighted MR imaging of the fetal chest include the thymus, which usually has intermediate signal intensity, and the trachea and bronchi, which can be visualized due to their amniotic fluid content. T1-weighted sequences can be performed to evaluate the liver and bowel in patients with CDH (2,3,7,8).

Types of Congenital Chest Malformations

Congenital Cystic Adenomatoid Malformation
CCAM is the most commonly diagnosed lung malformation in prenatal patients and accounts for 30%–40% of all congenital diseases (3,9,10). These malformations are characterized by an abnormal branching of the immature bronchioles, with a lack of normal alveolar development (1,2). Because CCAM usually communicates with the normal tracheobronchial tree, the malformation may consist of both cystic and solid areas, resulting in various radiologic manifestations (2). Fetal MR imaging can aid in both the assessment of CCAM and the evaluation of the development of the remaining lung (3).

On the basis of histologic features and size, three predominant histopathologic subtypes of CCAM can be identified (11). Type I is composed of single or multiple large cysts (3–10 cm in diameter) surrounded by smaller cysts and a compressed normal parenchyma. Type II is composed of various smaller cysts (0.5–2 cm) lined by cuboidal or columnar epithelial cells, with a thin underlying fibromuscular layer. Type III consists of small cystic lesions that are rarely larger than 0.2 cm in diameter. The imaging find-
ings of CCAM vary depending on the subtype. Type I lesions usually manifest as hyperintense uni- or multilocular regions with discrete walls on T2-weighted images (Fig 1). Type II lesions have variable appearances depending on the composition of the malformation. Type III lesions manifest as homogeneously hyperintense solid masses with normal adjacent parenchyma.

The differential diagnosis for CCAM includes bronchogenic cyst (CCAM type I) and BPS (CCAM type III) (2,3,6). Although bronchogenic cysts are typically isolated within the carinal region, in about 15% of cases the cyst is intraparenchymal (2,3). In these unusual cases, a specific diagnosis cannot be made prenatally. Differentiation between CCAM type I and BPS is challenging and is based on vascularization (3,10). Unlike BPS, CCAM communicates with the tracheobronchial tree and has an arterial blood supply and venous drainage from the pulmonary circulation. Hybrid conditions, which have histologic and imaging features similar to those of both CCAM and BPS, are commonly diagnosed at pathologic analysis, suggesting that these two entities have a common embryologic origin (Fig 2) (2,10).
The natural history of and prognosis for a CCAM diagnosed in utero are variable and depend on the size of the malformation, presence of pulmonary hypoplasia, mediastinal shift, and fetal hemodynamic alterations. The presence of fetal hydrops is the main indicator of a poor outcome, and it is generally an indication for fetal intervention (2,3,6,10). Infants who present with a significantly compromised pulmonary system should be treated with surgical removal of the mass as soon as the patient is stabilized (9). In many cases of CCAM, however, the malformation progressively decreases in size during the third trimester and mediastinal shift is resolved, such that the infant is asymptomatic at birth (3).

Bronchopulmonary Sequestration
BPS involves abnormal pulmonary tissue that does not connect with the normal tracheobronchial tree and receives its vascular supply from the systemic circulation. These sequestrations are caused by formation of an accessory lung bud ventral to the primitive foregut during gestation. Sequestrations are classified as either extralobar or intralobar. The intralobar subtype is more common (75% of cases) and is enveloped by the visceral lung pleura. The extralobar subtype can occur in a supra- or infra-diaphragmatic location and is enveloped by its own pleura (2,3). Both types of sequestration are most frequently encountered in the lower lobes, with the left lower lobe being the most common location.

Fetal MR imaging may aid in the characterization of BPS and its associated pulmonary malformations (3). MR imaging findings of BPS usually include a solid, well-defined, uniform, hyperintense mass on T2-weighted images. In cases of intralobar sequestration, the differential diagnosis also includes CCAM type III. To differentiate between these entities, the systemic arterial supply (present only in BPS and hybrid conditions) should be identified. In cases of extralobar sequestration, the differential diagnosis includes neuroblastoma and adrenal hemorrhage. Extralobar pulmonary sequestrations typically manifest in the second trimester as solid lesions, most commonly on the left side, with an anomalous systemic arterial...
Figure 3. Left-sided CDH in a fetus at 26 weeks gestation. (a, b) Color Doppler sonogram (a) and axial T2-weighted MR image (b) of the chest show herniated content (straight arrow) displacing the heart and compressing the left lung (curved arrow). (c, d) Sagittal (c) and coronal (d) T2-weighted MR images show portions of the bowel (arrow) and stomach (S) occupying the left hemithorax.

Congenital Diaphragmatic Hernia
CDH is caused by a defect in the diaphragmatic musculature, with herniation of the abdominal viscera into the thorax leading to deficiencies in lung development (3,9). Ninety-two percent of CDHs are posterior lateral defects, with 97% of these defects being unilateral and 80% occurring on the left side (9). There are two types of CDH: an early form in which the abdominal organs develop inside the thorax, and a late form associated with secondary migration of the abdominal organs into the thorax (12–14). The herniated contents may be a combination of bowel, stomach, or liver (Figs 3, 4) (2,9).

supply (1–3,6). Neuroblastomas occur more frequently on the right side and are more commonly diagnosed in the third trimester (10).

Both types of BPS may be associated with polyhydramnios and in utero hydrops fetalis, due to compression of the esophagus and thoracic venous structures by a large lesion (5,9). However, BPS may also involute during gestation. Prenatal management of BPS consists of careful observation. Hydrops development is an indication for fetal intervention or early delivery (2). After birth, BPS may be either asymptomatic or symptomatic. Surgical excision of the lesion is advisable in all symptomatic newborns because of the late risk of pulmonary infection or malignant transformation (3,5,9).
If the viscera dislocates during early lung development, a reduced number of bronchial branches, alveoli, and vessels are formed, leading to impaired lung development. Hypoplastic pulmonary changes are found most frequently in the ipsilateral lung, but the contralateral lung can also be affected by mediastinal shift and compression (2,9). The grade of pulmonary hypoplasia, the presence of mediastinal shift, early diagnosis, and liver migration to the thorax strongly influence prognosis, with cardiac and associated systemic abnormalities being associated with a worse prognosis (2,15).

Multiple imaging modalities have been used to predict pulmonary hypoplasia by means of lung volume calculations. Fetal MR imaging is useful for determining the degree of pulmonary hypoplasia, through calculation of relative fetal lung volume and lung-to-head ratio (3,15). These calculations are made by expressing the observed value as a percentage of the expected value with use of biometric parameters (15). Both MR imaging and sonography provide valuable parameters for predicting neonatal survival and the need for extracorporeal membrane oxygenation. Kilian et al (15) observed that fetuses with a relative fetal lung volume of less than 14.3% at MR imaging typically died, whereas those with a volume greater than 32.8% survived. No neonate with a volume greater than 44% required extracorporeal membrane oxygenation (15). Similar results for relative lung-to-head ratio were obtained at MR imaging, and all MR imaging measurements correlated well with the relative lung-to-head ratios as determined with sonography. Differences in the prognostic parameter values between the two imaging techniques might have been caused by the use of entirely different measurement methods (15).

**Congenital Hydrothorax**

Uni- and bilateral pleural fluid collections, which are considered abnormal in any gestational phase, result from disequilibrium across the pleural membranes (2,3). The prevalence of congenital hydrothorax is estimated to be approximately one in 15,000 pregnancies. The primary cause of congenital hydrothorax is chylous leak (chylothorax), caused by a lymphatic duct abnormality that results in defective chest drainage. Secondary causes include generalized fluid retention related to immune or nonimmune fetal hydrops.

Fetal MR imaging of congenital hydrothorax shows a fluid collection surrounding the lungs that is hypointense on T1-weighted images and hyperintense on T2-weighted images (Fig 5). The T1 and T2 signals can differ depending on the pleural effusion content, a fact that may help in making the diagnosis (2,3,9). Fetal MR imaging also allows investigation of associated abnormalities, which are present in about 40% of fetuses with pleural effusion and include CCAM, BPS, CDH, cardiac anomalies, Turner syndrome, Down syndrome, cystic hygroma, and infection. A primary hydrothorax may resolve or may

---

**Figure 4.** Right-sided CDH in a fetus at 30 weeks gestation. Sagittal (a) and axial (b) T2-weighted MR images show the liver (arrow) occupying the right hemithorax. Note the ascites around the liver (*).
progress to fetal hydrops. Pulmonary hypoplasia caused by large pleural effusions may be incompatible with postnatal survival (9). Fetal intervention (thoracentesis or thoracoamniotic shunting) is reserved for hydrothorax with a marked mass effect leading to a mediastinal shift (9).

**Congenital High Airway Obstruction Syndrome**

CHAOS is a rare abnormality with a poor prognosis and is caused by complete or near-complete obstruction of the fetal airway (16,17), leading to the trapping of lung fluids, hyperplasia of pulmonary alveoli, and tracheal dilatation (9). Fetuses with CHAOS may develop hydrops due to cardiac compression and obstructed venous return (7). Fetal T2-weighted MR imaging of CHAOS typically shows enlarged hyperintense lungs, a flattened or everted hemidiaphragm, and massive ascites (Fig 6). MR imaging may also show a dilated airway, which helps localize the obstruction (16,18) and aids in planning for surgical intervention at delivery (19).
Figure 7. Bronchogenic cyst in a fetus at 28 weeks gestation. (a) Transverse color Doppler sonogram shows a bronchogenic cyst (cursors) in the left hemithorax, adjacent to the heart. (b–d) Axial (b), coronal (c), and sagittal (d) T2-weighted MR images demonstrate a markedly hyperintense fluid-filled cyst within the left lung parenchyma (⋆).

Congenital Bronchogenic Cysts
Congenital bronchogenic cysts are caused by abnormal budding from the ventral foregut along the tracheobronchial tree, which subsequently differentiates into a fluid-filled, blind-ended pouch that is typically located in the mediastinum near the tracheal carina. Less commonly, cysts can occur within the lung parenchyma, pleura, or diaphragm (20). The bronchogenic cyst walls are thin, are covered by respiratory epithelium, and contain mucinous material (1). Fetal MR imaging aids in determining the precise location of the suspected lesion. The cyst is markedly hyperintense at T2-weighted MR imaging (Fig 7) (2,3).

Bronchial Atresia
Bronchial atresia, caused by airway obstruction with secondary dysplastic changes in the distal lung parenchyma, is one of a spectrum of anomalies linked to obstruction of the developing fetal airway. Thus, hybrid conditions are a common finding in affected patients (5). At prenatal imag-
Figure 8. Bronchial atresia. (a) Axial color Doppler sonogram obtained in a fetus at 28 weeks gestation shows a hyperechoic lesion in the lower lobe of the left lung. (b, c) Axial (b) and coronal (c) T2-weighted MR images obtained the same day show the lesion with high signal intensity (arrow). Postnatal chest radiography showed no significant abnormalities. (d, e) Axial chest computed tomographic scan (d) and coronal maximum intensity projection image (e) obtained in the neonate at 15 days show a hypoattenuating mass in the left lower lobe, with dilatation of the segmental bronchus inside the hypoattenuating area.

ing, the developing fetal lung is still filled with fluid, and it is difficult to distinguish between these anomalies, which can manifest as congenital masses (21). Fetal MR imaging can assist in the exclusion of other congenital chest malformations, thereby helping to narrow the differential diagnosis. Bronchial atresia most commonly manifests as a focal thoracic mass with homogeneously high signal intensity at T2-weighted MR imaging (Fig 8) (6). The areas most frequently involved are the apical and posterior segments of the left upper lobe (22). The differential diagnosis includes CCAM and BPS (6,21). Bronchial atresia usually occurs in the segmental or lobar bronchi (23). Patients are usually asymptomatic, with the abnormality being discovered incidentally (22). Elective surgical management during infancy is associated with an excellent prognosis (21).

Pulmonary Arteriovenous Malformation
Almost 60% of PAVMs, which are caused by abnormal communication between the pulmonary arteries and veins, occur in the lower lobes, and approximately two-thirds of PAVM patients have
Figure 9. PAVM in a fetus at 34 weeks gestation. (a) Color Doppler sonogram shows a vascular lesion (arrow) in the right hemithorax, adjacent to the heart. (b) Coronal T2-weighted MR image reveals a hypointense lesion in the middle of the right lung (arrow).

Congenital Pulmonary Lymphangiectasia

Congenital pulmonary lymphangiectasia, which may be either congenital or secondary, is a generalized dilatation of the lymphatic vessels, including pulmonary, subpleural, interlobar, perivascular, and peribronchial dilatation (25,26). Secondary lymphangiectasia may be due to surgery, infection, tumor genesis, or, in cases of total anomalous pulmonary venous return or hypoplastic left heart syndrome, venous obstruction (25,27,28). The congenital form is usually fatal in early life, and it probably results from pulmonary interstitial connective tissue failure, leading to dilatation of the pulmonary lymphatic vessels (28). Some of the genetic diseases associated with congenital pulmonary lymphangiectasia are Noonan syndrome, Turner syndrome, Ehlers-Danlos syndrome, and Down syndrome (25). Fetal MR imaging, which assists in morphologic characterization and the identification of anatomic abnormalities, can show bilateral inhomogeneity of the lung parenchyma on T2-weighted images (Fig 10). High signal intensity is
seen in the pulmonary interstitium, a finding that is very often associated with pleural effusion (26).

**Conclusions**

Congenital chest malformations are usually evaluated in the prenatal period with sonography, but MR imaging is an important adjunct technique. They can range from small and asymptomatic abnormalities to large space-occupying masses that require immediate surgical treatment. The information obtained with fetal MR imaging can be helpful in providing a detailed assessment of the thorax, allowing early planning of prenatal management.

**References**

some chest malformations occupy the thorax and alter normal pulmonary development, causing hypoplastic lung formation.

Prenatal sonography remains the primary imaging modality for evaluating the fetus, because it is safe, largely accessible, and inexpensive. However, in cases of inconclusive sonographic findings, the use of fetal magnetic resonance (MR) imaging helps identify potentially lethal pulmonary abnormalities.

The lungs typically contain a significant amount of alveolar fluid, which is homogeneously hyperintense relative to the chest wall muscle on T2-weighted images.

In cases of lung compression, the amount of alveolar fluid is decreased, resulting in a more hypointense signal.

Hybrid conditions, which have histologic and imaging features similar to those of both CCAM and BPS, are commonly diagnosed at pathologic analysis, suggesting that these two entities have a common embryologic origin.