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Congenital cystic diseases of the lung

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ABSTRACT

Congenital cystic diseases of the lung are rare but their incidence is increasing, especially with the widespread use of antenatal ultrasound. Close relationships between airway and respiratory segments and vascular structures during embryonic development can lead to malformations. It should be remembered that congenital cystic lung diseases have the potential to be life-threatening and the possibility of missed diagnosis is high. Surgical resection yields good results in symptomatic lesions, with lobectomy being the most commonly performed surgical approach. Some centers recommend surgical resection even in asymptomatic patients to prevent infection and exclude malignancy.

Keywords: Lung, congenital cystic diseases, antenatal ultrasound

INTRODUCTION

Although congenital cystic lesions of the lung are rare in children, their incidence is increasing with the widespread use of antenatal ultrasound.[1,2] Intrapulmonary structures consist of closely related airways and respiratory segments, which are associated with arterial, venous, and lymphatic vessels. These are complex structures that can undergo changes during embryonic development and lead to pulmonary malformations.[3] These diseases often share common etiological mechanisms and histopathological features, thought to arise from disrupted interaction between embryological mesodermal and ectodermal lung components during development.[1,2] The terminology and classification of cystic lung malformations have long been debated. Descriptions used for clinical diagnosis are generally based solely on imaging studies.[4,5] As understanding of the developmental and genetic origins of these disorders evolves, terminology and classifications are also revised.[2]

The likelihood of missing the diagnosis of congenital cystic lung diseases is high unless kept in mind and investigated with advanced imaging studies.[6] Congenital cystic lung diseases carry the potential to be life-threatening and require urgent diagnostic evaluation.[7] While the size of most lesions tends to decrease towards the end of pregnancy and may be asymptomatic at birth, the majority of these patients still exhibit abnormal postnatal CT findings.[8] Furthermore, these lesions have been reported to be responsible for fetal demise, severe respiratory distress in the neonatal period, and recurrent lung infections, and may even serve as precursors to malignant lesions.[3] However, the risk of malignancy in this population has not been fully defined.[8]

There is consensus on the necessity of surgical resection in symptomatic lesions; however, opinions are divided between surgical and more conservative approaches in clinically silent cases.[9] Some centers advocate for a watchful waiting non-surgical management, while others recommend surgical resection of the involved lung lobe to prevent infection and exclude malignancy.[8,10]

It has been suggested that early surgery (<6 months) may facilitate easier surgical intervention, shorter recovery time, potential compensatory lung growth, and potentially fewer complications.[11] Lobectomy is currently the most commonly performed surgical approach.[12]

BRONCHOGENIC CYST

Bronchogenic cysts are typically single, unilocular cysts filled with fluid or mucus and devoid of septations. In the presence of recurrent infection or bleeding, the content may become purulent or bloody.[2,13]

Bronchogenic cysts arise from abnormal budding of the endoderm of the foregut where the respiratory tract and upper gastrointestinal system develop. They are most commonly found in the mediastinum around the tracheal bifurcation, but can also occur in various locations within or outside the thorax (Figure 1). Unless complicated by secondary infection, which is more commonly seen in intraparenchymal cysts, they do not communicate with the normal tracheobronchial treer.[2,14]

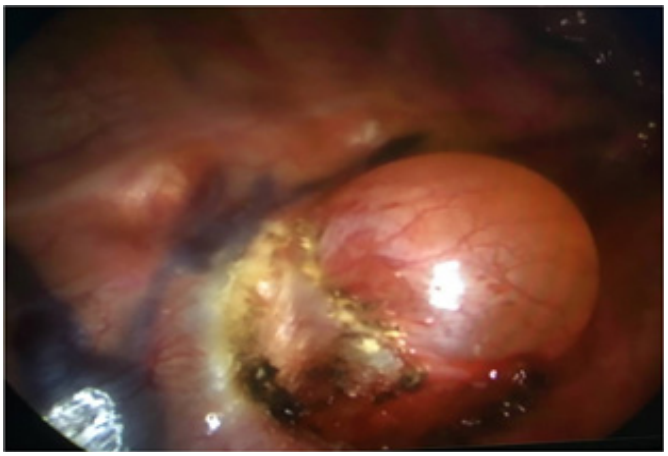

Figure 1. Bronchogenic cyst located in the right posterior mediastinum, within the superior vena cava (filled with purulent fluid)

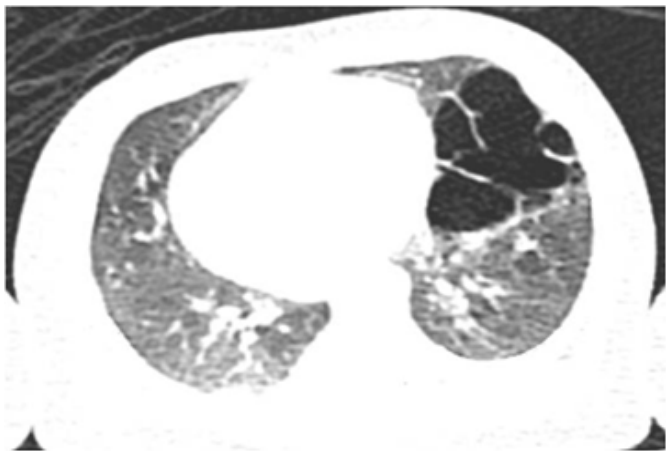

Figure 2. Congenital pulmonary airway malformation characterized by multiple cystic areas in the left lung in a newborn

In children, respiratory distress, dyspnea, recurrent pneumonia, lobar emphysema, or bleeding may occur due to infection of the cyst or mass effect caused by the cyst. The differential diagnosis list is extensive and includes pathologies such as pericardial cyst, cystic hygroma/lymphangioma, neuroenteric cyst, esophageal duplication cyst, and thymic cyst. When a bronchogenic cyst becomes complicated by infection or bleeding, it may be misdiagnosed as a simple abscess, pulmonary mass, pulmonary sequestration, or metastasis, making the diagnosis more challenging.[13]

Rarely, reports have been found of rhabdomyosarcoma, pulmonary blastoma, and malignant mesenchymoma in resected bronchogenic cysts in both children and adults.[15] Therefore, accurate diagnosis of bronchogenic cysts is important because missing the diagnosis can lead to serious complications.[13]

The treatment of bronchogenic cysts consists of complete surgical resection, enucleation, or lobectomy due to the risk of complications and malignant transformation. The use of video-assisted thoracoscopic surgery (VATS) is the preferred method for the treatment of bronchogenic cysts.[16]

CONGENITAL PULMONARY AIRWAY MALFORMATION

Congenital pulmonary airway malformation (CPAM), previously referred to as congenital cystic adenomatoid malformation, is the most common malformation of the lower respiratory tract.[2] CPAM is characterized by multiple cystic areas formed by excessive proliferation and dilation of terminal respiratory bronchioles, in the absence of normal alveoli (Figure 2). CPAMs are intrapulmonary lesions lined with various types of epithelial structures, maintaining communication with the normal tracheobronchial tree and preserving normal blood flow.[13] They are typically solitary and unilateral, with involvement of the lower lobe being common.[6,13] Nearly all CPAMs are detected on antenatal ultrasound.[17]

CPAM arises from an embryonic influence leading to abnormal development of terminal bronchioles. Typically, in most cases, a non-functional cystic portion of abnormal lung tissue replaces an entire lobe of the lung.[6]

The Stocker classification categorizes resected cysts into 5 categories based on the size and epithelial structure of the cyst: Type 0: involving all lung lobes and incompatible with life, Type 1: containing pseudostratified columnar epithelium, single or multiple cysts larger than 2 cm, Type 2: containing cuboidal or columnar epithelium, single or multiple cysts smaller than 2 cm, Type 3: primarily solid lesions containing cuboidal epithelium, approximately 0.5 cm cysts, Type 4: large air-filled cysts containing flattened epithelial cells.[13,18]

The prognosis of fetuses with CPAM is generally good; however, in rare cases, a cystic mass may compress normal lung and heart, posing a threat to the fetus's life.[6] Close monitoring with serial prenatal ultrasounds is necessary to determine the size, location, volume, blood flow, and fetal harm caused by the lesion.[13] Postnatally, the sensitivity of chest X-ray to detect CPAM is low, and usually computed tomography (CT) or magnetic resonance imaging (MRI) is required.[13,19]

The management of asymptomatic infants with CPAM remains controversial, although classical indications for resection are associated with respiratory tract infections and the risk of malignant transformation.[20] Delaying excision until the child becomes symptomatic has been shown to increase morbidity.[21] Additionally, follow-up X-rays or CT scans will increase the risk of radiation-related malignancy.[13] Early surgery is technically easier and reduces the risk of infection and the need for respiratory support. Therefore, the recommended timing for surgery is between 3 and 6 months, as the procedure is well tolerated, the risk of infection is lower, and it allows more time for compensatory lung growth.[19] With technological advancements in minimally invasive surgery, congenital lung lesions are now commonly removed with video-assisted thoracoscopic surgery (VATS).[22]

PULMONARY SEQUESTRATION

Pulmonary sequestrations (PS) are isolated portions of the lung separated from adjacent lung tissues. They have no connection with the bronchial tree and receive their blood supply from a systemic artery, with venous drainage to either the pulmonary or systemic vein.[13] PS is the second most common lung lesion in the spectrum of congenital lung malformations. PS is classified as intralobar (75%) and extralobar (25%) and tends to occur in the lower lobes of the lungs.[20]

PS likely occurs during very early embryonic development, possibly before the separation of the pulmonary and aortic circulations.[23]

Extralobar sequestration is often asymptomatic and incidentally detected posterior to the costophrenic angle.[1] It resembles an accessory lobe, located outside the visceral pleura of the lung and surrounded by its own pleura, regardless of whether it has a connection with the gastrointestinal system.[2] Hence, there is an anatomical boundary between it and the surrounding lung tissue.[13] While they more commonly occur in the thoracic cavity, infradiaphragmatic locations are not uncommon. They contain a vascular pedicle comprising systemic artery and drainage vein.[2] In postnatal period, children with PS may present with chronic cough, recurrent chest infections, feeding difficulties, or abdominal pain. Sixty percent of cases have another developmental anomaly, with congenital diaphragmatic hernia being the most common accompanying anomaly, along with others such as lung hypoplasia, congenital cystic adenomatoid malformation, congenital lobar emphysema, and bronchogenic cysts. There are also numerous associated cardiac malformations including dextrocardia, truncus arteriosus, and total anomalous pulmonary venous drainage. In 75% of cases, arterial blood supply is provided from the thoracic or abdominal aorta, but arterial supply can also arise from subclavian, intercostal, phrenic, internal thoracic, celiac trunk, or gastric arteries.[13,24]

Intralobar sequestration is typically localized in the posterobasal region of the lower lobe.[25] It may resemble a small cystic CPAM containing mucus-filled dilated airways and an atretic bronchus in the proximal portion of the lesion. Distinguishing intralobar sequestration from small cystic CPAM is achieved by identifying systemic arterial blood supply. When infected, intralobar sequestration can be mistaken for chronic pneumonia. Suspect intralobar sequestration in children or young adults with recurrent lower lobe pneumonia. Unlike sequestration, pneumonia has a bronchial connection with the involved lung.[2] Rarely, in the neonatal period, intralobar sequestration may present with respiratory distress secondary to congestive heart failure.[1]

Identifying abnormal systemic blood flow radiographically and surgically is a defining feature for both types of sequestration.[2] Vascular mapping during investigation of pulmonary sequestration is crucial. Doppler ultrasound, CT, and MRI are the main methods used; however, each has its advantages and disadvantages.[13]

Surgical resection involving segmentectomy or lobectomy is performed in children presenting with chronic complaints related to pulmonary sequestration. However, the treatment of asymptomatic patients with pulmonary sequestration is controversial; some prefer resection of the lesion due to the risk of bleeding and infection instead of conservative treatment.[13]

CONGENITAL LOBAR EMPHYSEMA

Congenital lobar emphysema is defined as hyperinflation of one or more lung lobes leading to compression of surrounding structures. This compression can cause mediastinal shift. The most commonly affected lobes are the left upper and right middle lobes (Figure 3).[13] While the etiology of congenital lobar emphysema is uncertain in half of cases, it can be attributed to intrinsic factors such as bronchial stenosis, bronchomalacia, mucosal proliferation, and vascular, lymph node, and adjacent lung compression as extrinsic factors.[20,26] A possible etiology could be a cartilage defect weakening the bronchus, leading to collapse on expiration and subsequent trapping of air causing hyperinflation.[27]

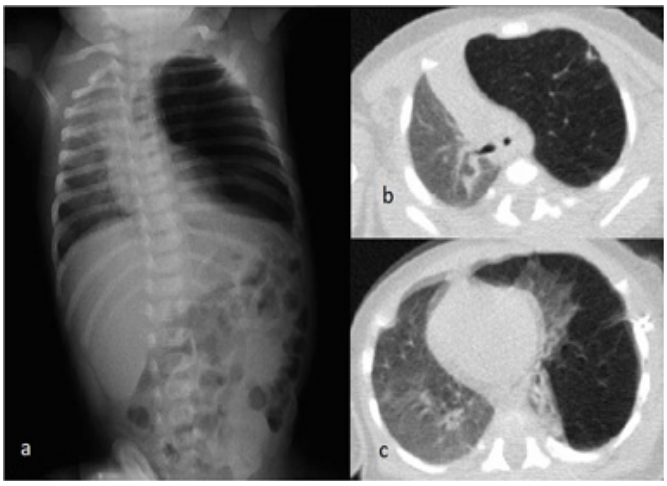

Figure 3. Mediastinal shift due to congenital lobar emphysema in the left upper lobe (a, b) and atelectasis in the left lower lobe (c)

Evaluation of associated anomalies is necessary due to the presence of associated congenital heart disease in 14% of cases of congenital lobar emphysema.[1] Additionally, distinguishing acute reversible lobar emphysema from serious irreversible lesions is important.[27]

Children may present with neonatal respiratory distress, tension pneumothorax, wheezing, or atelectasis due to lung compression, or may be asymptomatic. Chest X-ray is often diagnostic, but confirmation is usually done with CT.[6,13] Echocardiography or MRI may be needed to determine the cause of respiratory distress. Care should be taken not to misinterpret chest X-rays as pneumothorax.[13]

Treatment of children with congenital lobar emphysema consists of treating the underlying mechanism. Symptomatic cases are treated with lobectomy, while asymptomatic or mildly symptomatic patients can be managed conservatively.[6,13]

CONCLUSION

Congenital cystic diseases of the lung are rare, but their incidence is increasing, especially with the widespread use of antenatal ultrasound. Some centers recommend surgical resection even in asymptomatic patients to prevent infection and exclude malignancy. Surgical resection gives good results in symptomatic lesions and lobectomy is the most common surgical approach. It should be kept in mind that congenital cystic lung diseases have the potential to be life-threatening and the diagnosis is highly likely to be missed. Early diagnosis and treatment are important.

ETHICAL DECLARATIONS

Referee Evaluation Process

Externally peer-reviewed.

Conflict of Interest Statement

The authors have no conflicts of interest to declare.

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Author Contributions

All of the authors declare that they have all participated in the design, execution, and analysis of the paper, and that they have approved the final version.

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