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OLGU SUNUMU
CASE REPORT

A rare disease; congenital pulmonary airway malformation in an adult

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SUMMARY

A rare disease; congenital pulmonary airway malformation in an adult

Congenital pulmonary airway malformation (CPAM), is a rare disease known as a developmental abnormality of the lower respiratory tract. It may occur in the neonatal period due to respiratory distress and patients usually die in the first few months of life. Rarely, it may remain asymptomatic until adulthood. In this study we describe an adult case and present a review of the literature. A 19-year-old male with no relevant medical history was admitted to our clinic with cough and wheeze. The patient had a history of frequent lower respiratory tract infection during childhood. Chest radiograph revealed a diffuse opacity and volume loss in the right hemithorax. High resolution tomography showed hypoplasia of the right hemithorax, multiple cysts in all of the lobes and segments on the right side, ground glass opacity and interlobular septal thickening of the whole right lung parenchyma. Right pneumonectomy was performed with the pre-diagnosis of congenital pulmonary airway malformation and the pathological examination was compatible with CPAM. CPAM is a rare disease in adulthood. We should consider CPAM in the differential diagnosis of patients with frequent recurrent pulmonary infection and cystic lung lesions. In order to prevent infections and to eliminate the risk of malignancy, surgical treatment should be applied for definite diagnosis and treatment.

Key words: *Congenital disease; lung; cyst*

ÖZET

Nadir bir hastalık; erişkin dönemde rastlanan konjenital pulmoner hava yolu malformasyonu

Konjenital pulmoner hava yolu malformasyonu (KPHM), alt solunum yollarında gelişimsel bir anormallik olarak bilinen nadir bir hastalıktır. Yenidoğan döneminde solunum sıkıntısı nedeniyle ortaya çıkabilir ve hastalar genellikle yaşamın ilk birkaç ayında ölür. Nadiren yetişkinliğe kadar asemptomatik kalabilir. Bu çalışmada tanımlanan erişkin bir olgu eşliğinde literatürün gözden geçirilmesini sunuyoruz. Bilinen kronik hastalığı olmayan 19 yaşında erkek öksürük ve hışıltı ile kliniğimize başvurdu. Hastanın çocukluk döneminde sık alt solunum yolu infeksiyonu öyküsü vardı. Akciğer grafisinde sağ hemitoraksta difüz opasite ve hacim kaybı izlendi. Yüksek çözünürlüklü akciğer tomografisinde sağ hemitoraksta hipoplazi, sağda tüm loblarda ve segmentlerde çok sayıda kist, buzlu cam opasitesi ve sağ akciğer parankiminin tamamında interlobüler septal kalınlaşmalar görüldü. KPHM ön tanısı ile sağ pnömonektomi yapıldı ve patoloji sonucu KPHM ile uyumlu bulundu. KPHM erişkin yaşta nadir görülen bir hastalıktır. Sık sık tekrarlayan akciğer infeksiyonu ve kistik akciğer lezyonu olan hastaların ayırıcı tanısında KPHM'yi düşünmeliyiz. İnfeksiyonları önlemek ve malignite riskini ortadan kaldırmak, kesin tanı ve tedavi için cerrahi tedavi uygulanmalıdır.

Anahtar kelimeler: *Konjenital hastalık; akciğer; kist*

INTRODUCTION

Congenital pulmonary airway malformation (CPAM), previously called congenital cystic adenomatoid malformation (CCAM) is a rare disease known as developmental abnormality of the lower respiratory tract (1,2). It may occur in the neonatal period due to respiratory distress and patients usually die in the first few months of life. Rarely, it may remain asymptomatic until adulthood. Many cases are detected by routine prenatal ultrasound imaging. In asymptomatic cases with localized lesions, follow-up is recommended. However, surgical resection is the definite treatment.

CASE REPORT

A 19-year-old male with no relevant medical, family or psychosocial history was admitted to our outpatient clinic with cough and wheeze. The patient's complaint had begun 3 months ago with non productive cough without fever with shortness of breath, right sided chest pain, fatigue and generalised myalgia. His vitals were normal. The patient received nonspecific antibiotic therapy due to respiratory tract infection but his symptoms did not resolve. The patient had a history of frequent lower respiratory tract infections during childhood. He has never smoked. On oscultation; decreased breath sounds were noted over the right hemithorax. There was a restrictive patern on the pulmonary function test. Chest radiograph revealed a right-sided diffuse opacity with volume loss (Figure 1). High resolution tomography showed hypoplasia of the right lung, multiple cysts in all of the lobes and segments of the right side, ground glass opacity with interlobular septal thickening in the whole right lung parenchyma in

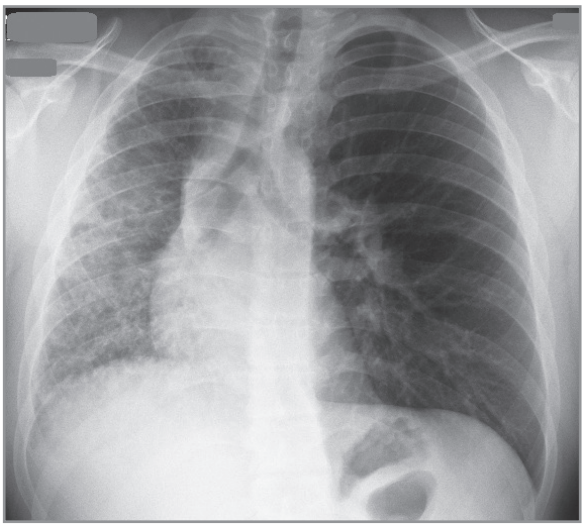

Figure 1. Chest Radiograph; a diffuse opacity and volume loss in the right hemithorax.

axial view of the ct images. (Figures 2,3). Bronchial washings cytology and cultures for bacterial, fungal, and mycobacterial organisms were negative. Bronchoscopy showed no pathological finding. Biochemical parameters were normal. The patient was consulted with the thoracic surgery department and right pneumonectomy was performed with the pre-diagnosis of CPAM. Pathological examination revealed emphysematous changes, diffuse interstitial fibrosis, cystic areas with occasionally microscopic honeycomb pattern were interpreted as findings consistent with cystic adenomatoid malformation (CPAM). The patient signed an informed consent form.

DISCUSSION

There are many congenital anomalies affecting the respiratory system. Some of these anomalies can

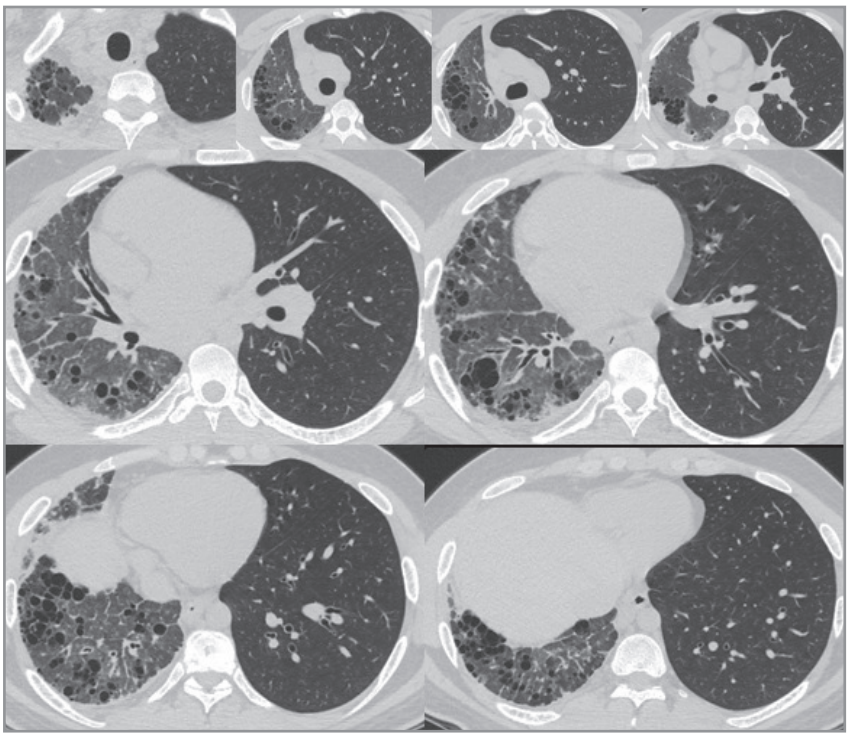

Figure 2. High resolution tomography; hypoplasia of the right lung, multiple cysts in all of the lobes and segments of the right side, ground glass opacity with interlobular septal thickening in the whole right lung parenchyma.

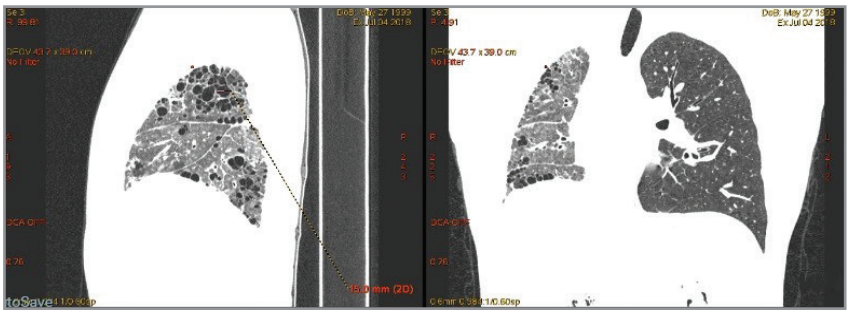

Figure 3. Sagittal view of the ct images.

cause death, some may cause severe respiratory distress during the newborn period, while others may remain asymptomatic for a long time. CPAM is the most common congenital anomaly after congenital lobar emphysema. It constitutes 25% of congenital lung malformations and 95% of congenital cystic lung diseases (3). The incidence is 1 in 8300 to 35.000 live births (4,5).

CPAM cases are sporadic and do not correlate with maternal factors such as race, age or exposure. The gender distribution shows a slight male predominance, as it has been reported in some series (6,7). Except for type 4 malformations which is associated with familial pleuropulmonary blastoma syndrome, there is no known genetic predisposition. There was no family history in our case.

Molecular mechanisms resulting in the formation of CPAM are not known, but an imbalance has been implicated in cell proliferation and apoptosis during organogenesis (8-11). The affected lung contains hamartomatous lesions including cystic and adenomatous elements from trachea, bronchus, bronchiol or alveolar tissue itself. Large lesions may distort alveolar growth and development by compressing adja-

cent normal tissues. It may be equally seen in both lungs and may occur in all lobes. Lesions are usually limited to a lobe, but rarely they can be seen in multiple lobes (12). CPAMs that are diagnosed in adulthood tend to be in the lower lobes (13). In our case, all lobes were affected in the right lung.

The disease may be associated with various congenital anomalies, including 18% of primary renal and cardiac anomalies (14). In our case, extrapulmonary involvement was not found.

Different types of CPAM are thought to occur at different levels of the tracheobronchial tree and possibly at different stages of lung development affected by acute obstruction and/or atresia (4,15,16).

CPAMs are divided into five main classes based on the size and cellular characteristics of the cysts (predominantly bronchi, bronchioles, or bronchioles/alveolar canal cells) (17-19). According to this classification, more than 65 percent of CPAMs are type 1, 20 percent to 25 percent in type 2, and 8 percent to type 3 (17,18). Each type of CPAM has distinct pathological properties.

Type 0 is the rarest form of tracheal or bronchial tissue origin. The cysts are small, with a maximum diameter of 0.5 cm and are covered with ciliary pseudostratified epithelium (20). Mucous cells and cartilage are present, but there is no skeletal muscle. As it encompasses the entire lung, gas exchange is severely impaired and affected babies die at birth (4).

Type 1 is the most common form of CPAM (60-70%) (4). This type is thought to originate from distal bronchi or proximal bronchioles. The lesions consist of thin-walled cysts that range from 2 to 10 cm in diameter. Cysts usually stand unique and can rarely be multiple. The clinical appearance depends mainly on the size of the cysts. Alveoli in the neighborhood are relatively normal (4). If the adjacent distorted parts compresses the normal lung tissue, respiratory distress of the newborn can occur, with mediastinal shift to the contralateral side and flattening of the ipsilateral diaphragm. Small cysts can be detected months or years after birth, usually as a result of infection. This type of CPAM has the potential for malignancy.

Type 2 constitues of 15 to 20 percent of CPAMs. They consist of multiple cysts with a diameter of 0.5 to 2 cm and solid areas adjacent to normal tissue. This type can be confused with bronchopulmonary sequestration. However, feeding of bronchopulmonary sequestration with systemic artery is important in the differential diagnosis. Hybrid forms with features of type 2 CPAM and bronchopulmonary sequestration have been described (2).

In addition, coexistence with other congenital anomalies is more common in the type 2 of CPAM (4,21). These malformations include esophageal atresia, bilateral renal agenesis or dysgenesis, intestinal atresia, other pulmonary malformations, and diaphragmatic, cardiac, central nervous system, and skeletal anomalies (22,23). There is no risk for malignancy.

Type 3 CPAMs account for 5 to 10 percent of cases. They are usually very large and may in volve all lobes or various lobes. They have an acinar origin and consist of adenomatoid proliferation of distal airways. They may be in cystic/solid nature or may be completely solid. There are many small cysts with a diameter of less than 0.5 cm. There may be severe respiratory distress during neonatal period (4). This type of CPAM is not associated with malignancy.

Type 4 lesions are seen in 5 to 10% (4,20). The maximum diameter of the cysts is 7 cm, which can often be associated with tension pneumothorax or infection, either at birth or in childhood. They may also be found accidentally in asymptomatic patients. Importantly, this type of CPAM is strongly associated with malignancy, particularly with pleuropulmonary blastoma.

Diagnostic Method and Clinical Presentation

CPAM is a clinically diverse disease. Developments in prenatal ultrasound technology have increased early prenatal diagnosis rates and are also useful for differentiating CPAM from other pulmonary anomalies including bronchial pulmonary sequestration, congenital diaphragmatic hernia and congenital lobar emphysema with additional evaluation by prenatal magnetic resonance imaging (MRI). These disorders are usually differentiated from CPAM based on radiological appearance and clinical history. If not diagnosed prenatally and is initially noticed in the newborn or childhood period, the diagnosis can usually be made by chest radiography; however, thorax computed tomography or magnetic resonance imaging is recommended (24,25).

25% of patients who are diagnosed with prenatal CPAM are symptomatic at birth (26). Respiratory distress probability and severity is directly proportional to the extent of disease (26).

Older children may present with recurrent pneumonia, cough, dyspnea and/or cyanosis (27,28). CPAM cases may also present with spontaneous pneumothorax. Since pneumothorax and malignancy are most commonly associated with type 4 CPAM, malignancy should be investigated in patients presenting with pneumothorax (4).

Close observation is important in infants who are asymptomatic in the neonatal period because they may become symptomatic during the latter stages of lung development (28). The decision between surgical treatment and observation is controversial for babies and children who remain completely asymptomatic.

In symptomatic patients, CPAM is treated by surgical resection (2,7). Resection in newborns with significant respiratory distress is often necessary, but is usually performed electively in older children who present with less acute symptoms. In older children, resection is performed to prevent recurrent infections and to eliminate concerns about malignancy. It also has advantages for early surgical compensatory lung growth. Lobectomy is superior to wedge resection (2).

CPAM is a rare disease in adulthood. We should consider CPAM in the differential diagnosis of patients with frequent recurrent pulmonary infections and cystic lung lesions. In order to prevent infections, and avoid malignancy, surgical treatment should be applied for definitive diagnosis and treatment.

CONFLICT of INTEREST

The authors reported no conflict of interest related to this article.

AUTHORSHIP CONTRIBUTIONS

Concept/Design: NÖ, SÖ

Analysis/Interpretation: NÖ, SÖ

Data Acquisition: NÖ, SÖ

Writting: NÖ, SÖ, BYK

Critical Revision: SÖ, BYK, ÜŞ

Final Approval: SÖ, ÜŞ

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