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Case Report

Pulmonary congenital cystic adenomatoid malformation: a rare congenital abnormality in adults and review of literature

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Abstract

Congenital cystic adenomatoid malformation of the lung (CCAM) is characterized by an adenomatoid proliferation of bronchiole-like structures and cysts formation. The condition is most commonly found in newborns and children and may be associated with other malformations; rarely, the presentation is delayed until adulthood. We herein report two cases of CCAM in adult patients. 22 years old healthy female with pre-employment health screening chest X-ray showed a lesion in the upper lobe of the right lung. In another case, a computed tomographic scan of the thorax (CT) confirmed a mass in the upper right lung. A 28-year-old male presented with recurrent respiratory tract infection resistant to antimicrobial therapy. CT scan of the thorax showed a mass in the left lung upper zone. Surgical resection was performed in both cases, and histopathology of the resected specimen showed both cases were consistent with the CCAM.

More Information

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Introduction

Congenital cystic adenomatoid lung malformation (CCAM) is a rare developmental anomaly known as unilateral dysplasia of the lung. It was described in 1787 as the absence of the lung. Ch'in and Tang first reported it in 1949 [1,2]. The development of the respiratory system begins at three weeks of gestation. There is a hypothesis that CCAM occurs due to the arrest of the development of the bronchial tree or due to alteration in bronchial development. Stocker, in 1977 classified CCAM into three types, widely used until 2002, then new reclassification of CCAM was reported with five subtypes [3,4]. 85% of the cases are diagnosed within the first two years of life and are rarely encountered in adults. Primarily it is interpreted as an incidental finding on radiological examination of chest X-Ray and CT scan of the chest. In adults, this usually involves one lobe, and the patient presents with a persistent cough, hemoptysis, recurrent respiratory tract infection, and lung abscess. Up to 26% of the cases of CCAM were associated with anomalies [5]. This case series is reported in line with PROCESS criteria [6].

Case series

Case 1

Twenty-four years old female was referred to us from the employee health clinic with opacity in the upper zone right lung. She had no medical illness in the past. Clinical examination of the neck, chest and abdomen was normal. There was no palpable lymphadenopathy. Basic blood investigations CBC, Liver and renal panels were normal. CT scan of the chest showed a mass in the right upper lobe. Mass was approached during the Right posterolateral thoracotomy and we found a hard mass in the apical segment of the upper lobe of the right lung. A segmentectomy was performed, and the patient was extubated on the table and had an uneventful recovery. The histopathology report showed a typical alveolar structure with multiple cysts and fibrovascular connective tissues. A Follow-up CT scan after four years was normal.

Case 2

A 26-year-old male presented to us with a history of cough and recurrent chest infection for the last six months, which

did not resolve with antibiotics. He had no medical illness in the past, and basic blood investigations were normal. A chest X-Ray and CT scan of the chest showed a 5 cm × 3.5 cm mass in the upper lobe of the left lung. This was approached through a left posterolateral thoracotomy, and we found a large cystic mass in the left upper lobe with dense adhesions to the chest wall and pericardium. A left upper lobectomy was performed. The patient was extubated on the table and transferred to the ward. His post-operative recovery was uneventful, and a follow-up chest CT scan was normal. Histopathology report showed numerous small cystic structures lined by ciliated columnar cuboidal and columnar epithelium and fibromuscular layer consistent with CCAM.

Discussion

The exact etiology of CCAM remains unknown. There is a hypothesis that hamartomata's change in the terminal bronchioles or an arrest in their embryological development between 7 and 15 weeks of gestation is the suspected cause [7,8]. Another theory is that decreased apoptosis also plays a role; genetic studies showed that the related genes in the pathogenesis are HOXB5, Fgf7, and PDGFB [9].

Although congenital cyst adenomatoid malformation, also known as congenital airway malformation, is a rare disease but still, it is the most common malformation of the lower respiratory tract. In CCAM, the normal alveoli are replaced by a cystic component composed of adenomatous hyperplastic bronchioles. Due to recent advances in prenatal care and ultrasound, cystic lung lesions are more often detected early, and accordingly, peri Partum and neonatal care are planned. Stocker classified CCAM into five types from 0 to IV, as shown in the figure. Among all, Type III has the worst prognosis. CCAM is currently organized into five main types (0-4) based on the embryologic level of origin and the histologic features [10,11] (Figures 1-3, Table 1).

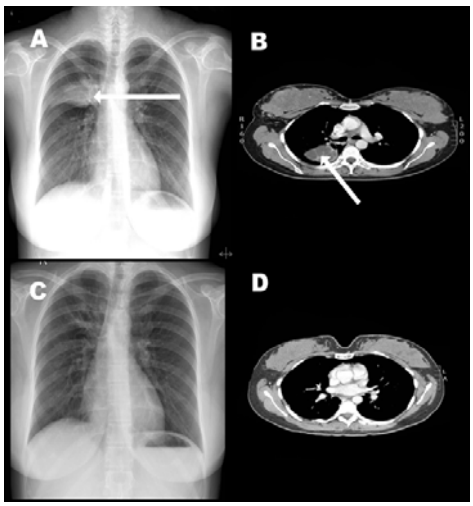

Figure 1: (A) Chest X-Ray showing a opacity in the right lung upper zone. (B) CT scan of the chest showing heterogenic mass in the right lung. (C) A normal post-operative chest X-Ray (D) Follow up CT scan of the chest revealed no abnormality.

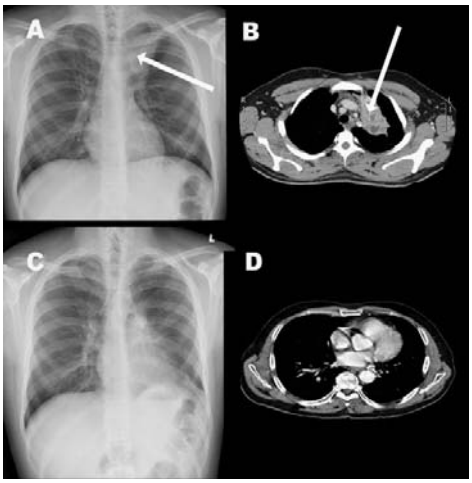

Figure 2: (A) Chest X-Ray showing a lesion in the upper lobe of left lung. (B) CT Scan of the chest showing complex cystic mass in the left upper lung. (C) A post-operative chest X-Ray. (D) Follow up CT scan of the chest.

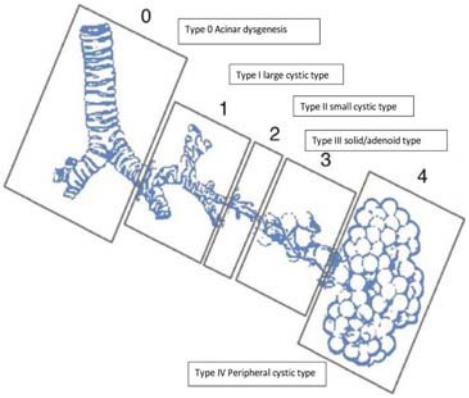

Figure 3: JT Stocker, et al. Pediatric pathology. Stoker classification with expanded form.

Table 1: Classification of congenital Cyst adenoid malformation (CCAM).

Type 0	Type 0: CCAM is the rarest form and arises from the trachea or bronchus; there is acinar dysgenesis, and cysts are small
Type 1	CCAM is the most common form, representing 50% to 70% of cases, and it arises from the distal bronchus or proximal bronchiole. Because these CCAMs may be large, they may have a significant mass effect, which can lead to hydrops.
Type II	Type II CCAMs account for 15% to 30% of cases and arise from terminal bronchioles. They are composed of smaller cysts, measuring 0.5 cm to 2 cm, and solid areas that may be difficult to distinguish from surrounding tissue.
Type III	CCAMs account for 5% to 10% of cases, are thought to arise from acinar-like tissue, and are composed of cysts so small the mass appears to be solid and highly echogenic on ultrasound. The tissue is acinar and shows adenomatoid elements consistent with the distal airway
Type IV	Type IV: CCAMs arise from alveoli accounting for 5% to 15% of cases. These CCAMs contain large peripheral cysts that may be as large as 10 cm and have been associated with malignancy, specifically pleuropulmonary blastoma.

Type 0: CCAM is the rarest form and arises from the trachea or bronchus; there is acinar dysgenesis, and cysts are small.

Antenatally, CCAMs have been classified as microcystic (5 mm) versus macrocytic (.5 mm). Microcystic lesions are usually significantly larger. Laberge, et al. l and stoker, et al. l; reported Incidence of CCAM is 1 in 11,000 to I in 35000, respectively7. Usually, 25% of cases of CCAM are diagnosed

during prenatal ultrasound check-ups. 85% are diagnosed in the first two years of life. At the same time, 71% are asymptomatic, which undergoes spontaneous regression [12].

CCAM in adults is very rare, and only fifty cases have been reported in the medical literature. CCAM is detected incidentally during the radiological examination, and patients are asymptomatic. Sometimes they present with chronic cough, recurrent respiratory tract infection, hemoptysis, pneumothorax, pneumonia, and lung abscess [13]. Medical literature shows that CCAM types I, II, and III are seen at 70%, 40% and 3% of the cases, respectively. Up to 26% of the patients were associated with other anomalies, more with type II and III, including abnormalities in chromosome 18 [14]. CCAM is sometimes misdiagnosed as sequestration as they share some histological patterns, but the criteria can differentiate them [15]. As given in Table 1. CCAM is usually diagnosed in the first two years of life, and around 17% of cases are present in the later years of life. It is commonly presented as a unilateral lesion causing respiratory distress in neonates and infants. If the fetus doesn't develop hydrops, then postnatal survival is 100%. In adults, either this is usually diagnosed incidentally, rarely patients can present with a history of cough, shortness of breath, hemoptysis, pneumonia, and bacterial and fungal lung abscess. The best treatment for CCAM is surgical resection, lobectomy, or segmentectomy because there is a risk of malignant transformation if left untreated. CCAM is commonly isolated, but this has been reported in conjunction with cardiac and renal anomalies of 15% and 17%, respectively. Particularly type 2 is associated with gastrointestinal atresia and cardiac and renal skeletal anomalies [16,17]. CAM and extrapulmonary sequestration can occur together. Transitions into bronchioloalveolar carcinoma, pleuropulmonary blastoma and rhabdomyosarcoma have been reported [18-20].

CT scan chest is the diagnostic imaging tool for CCAM, but histological confirmation is mandatory; therefore, diagnosis is reached only from the histopathology post-surgical resection of the lesion. The standard treatment of CCAM is lobectomy because of poor sensitivity and low negative predictive value of the preoperative CT, which decreases the chance of recurrence [20]. There are conditions when CCAM with extensive involvement and difficult surgical technique are treated conservatively. The operative surgical versus nonoperative treatment decision depends on the extent of the disease, patient symptoms, and risk for surgery. We recommend early surgical intervention in an asymptomatic patient who is fit for surgery because it will be easier than when it becomes symptomatic and infected. There is always a risk of complications in CCAM like pneumothorax, infection, and malignancy transformation [21-24].

Conclusion

In conclusion, Due to its rarity, physicians are unlikely to suspect cystic adenomatoid malformation (CCAM). Even if CCAM is a rare disorder, it should be suspected in adult patients with clinical manifestations of a recurrent chest infection or persistent cough. CT scan is the best radiological imaging modality to reach a preoperative diagnosis of CCAM, which can help in assisting the internal structure of the lesion and the extent of distribution of the disease. Histopathology of the resected lesion is needed to confirm the diagnosis. Malignancy risk of transformation and all other mentioned complications, in our opinion, does not justify conservative management. We recommend surgical intervention, such as undergoing segmentectomy, lobectomy, or pneumonectomy, for all patients with acceptable risk for surgery. There should be a long clinical and radiological follow-up.

Ethical approval: IRB Approval.

Patient consent: Written informed consent was obtained from the patients for publication of this and accompanying images. A copy of the written consent is available for Review by the Editor in chief of this journal on request.

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