www.jpnim.com Open Access eISSN: 2281-0692
Journal of Pediatric and Neonatal Individualized Medicine 2023;12(1):e120128
doi: 10.7363/120128
Received: 2022 Nov 08; revised: 2023 Jan 28; accepted: 2023 Mar 16; published online: 2023 Apr 29

Case report

Congenital cystic adenomatoid malformation of the lung: diagnosis in a preadolescent

Margarida S. Abreu[1], Ines Braga[1], Joana Sobreiro Silva[2], Jorge Correia Pinto[3], Augusta Gonçalves[4]

[1]Braga Hospital, Braga, Portugal
[2]Department of Anatomopathology, Hospital de Braga, Braga, Portugal
[3]Department of Pediatric Surgery, Hospital de Braga, Braga, Portugal
[4]Department of Pediatrics, Hospital de Braga, Braga, Portugal

Abstract

Congenital pulmonary airway malformation (CPAM) is a rare developmental lung malformation. Nowadays, most CPAMs are diagnosed prenatally, allowing optimal prenatal and immediate postnatal care. Rarely, however, CPAM may go undetected in asymptomatic or paucisymptomatic patients, until a complication, most commonly of infectious nature, uncovers the underlying lesion.

In this case report, a young teenage boy with a history of recurrent pneumonia presented with clinical worsening following a respiratory infection and subsequent adequate antibiotic intake. Chest radiograph revealed a left heterogeneous hypotransparency, and chest computed tomography scan disclosed exuberant cystic bronchiectasis occupying almost the entire left lower lobe. Further investigation excluded primary immunodeficiency, cystic fibrosis, and mechanical obstruction of the airway. Following the acute infectious phase, the patient underwent a programmed thoracoscopic resection of the lower lobe, that was uneventful. Macroscopic and histological examination confirmed a congenital cystic adenomatoid malformation Stocker type 1. On long-term follow-up, the patient remained asymptomatic, with normal respiratory function tests.

Recurrent pneumonia and chronic productive cough should be actively sought in routine visits, and should prompt further investigation, including imaging targeting adequate characterization of the lung parenchyma. Despite the overall good surgical prognosis, whatever the age, early intervention, especially in asymptomatic patients at birth, may decrease surgical morbidity and other late complications related to severe and recurrent infections or even malignancy.

Keywords

Lung malformation, congenital cystic adenomatoid malformation, bacterial pneumonia, bronchiectasis, cough, prenatal ultrasonography.

Corresponding author

Margarida S. Abreu, Braga Hospital, Braga, Portugal; email: amargaridasabreu@gmail.com.

How to cite

S. Abreu M, Braga I, Sobreiro Silva J, Correia Pinto J, Gonçalves A. Congenital cystic adenomatoid malformation of the lung: diagnosis in a preadolescent. J Pediatr Neonat Individual Med. 2023;12(1):e120128. doi: 10.7363/120128.

Introduction

Congenital pulmonary airway malformation (CPAM) is a rare developmental malformation of the lower respiratory tract due to abnormal lung branching during morphogenesis. The resulting hamartomatous lesion suppresses alveolar growth leading to the replacement of normal lung structure by a multicystic mass [1].

Nowadays, most CPAM are diagnosed antenatally due to sonographic improvements and managed surgically in early life. The estimated prevalence in fetuses is 0.81/10,000, according to a European registry, and in 2016, according to an 8-year retrospective review, the antenatal diagnosis of cystic lung lesions was estimated at 85.7% [2]. Postnatal diagnosis usually occurs in the neonatal period and early infancy, following cyst infection. However, clinical manifestations may comprise a wide variety of underlying complications, that occur in about 3.2% of non-operated patients, such as hemorrhage, pneumothorax, nutritional difficulties, sudden respiratory compromise and malignant changes [3]. Chest radiograph may reveal a hyperlucent mass with a radio-opaque component and display other complication-related features. Computed tomography (CT) angiogram is the preferred imaging modality for characterization and surgical planning, but does not replace specimen pathologic analysis [2-4]. Although the magnetic resonance imaging may be an alternative as to avoid radiation, it is less widely available, interpretation is more expertise-dependent, and thin-wall cysts and emphysematous changes may go undetected [3]. Regarding the cases with antenatal diagnosis, chest radiograph is performed shortly after birth in many centers, although it is often normal. CT should, therefore, complement the investigation within the first months of life [3].

Surgery remains the cornerstone treatment of symptomatic lesions, with most studies advocating early-on intervention, that also serves diagnostic purposes. However, physicians should bear in mind that symptomatic patients are more likely to experience surgical complications [2].

Although most patients present with symptoms at a very young age, around 6-7 months average, they are often unspecific [1]. In rare situations, the underlying malformation can go undetected for many years, defying clinical and epidemiological benchmarks at diagnosis.

Case report

An 11-year-old boy presented with *de novo* acute left chest pain, low grade fever and persistence of cough with mucopurulent sputum after having completed 10 days of amoxicillin clavulanate in a weight-appropriate dosage a few days earlier, following a diagnosis of a respiratory infection. Epidemiological context was irrelevant. In the medical history, the mother mentioned an adenovirus pneumonia as an infant and two other left lobar pneumonias requiring outpatient treatment as a child and a persistent productive cough, ongoing for several years. Immunizations included complete 7-valent pneumococcal conjugate and *Haemophilus influenzae* type b vaccines. Prenatal and perinatal history was unremarkable. Growth pattern (weight and height) was regular and within reference values.

Physical examination revealed mild tachypnoea and diminished breath sounds in the left lower thoracic area. There was no clubbing. Blood tests favored a bacterial etiology. Chest X-ray confirmed a lower left lung heterogeneous hypotransparency (**Fig. 1**). Finally, chest CT scan (**Fig. 2**) revealed exuberant

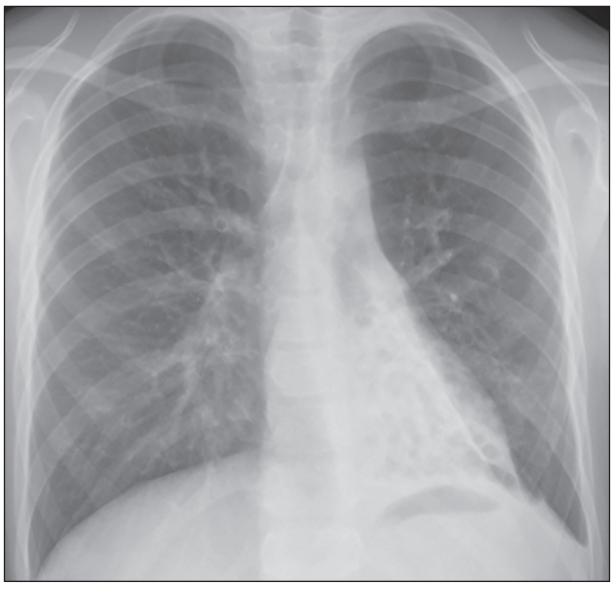

Figure 1. Chest X-ray of the patient upon admission, displaying a heterogeneous retrocardiac hypotransparency.

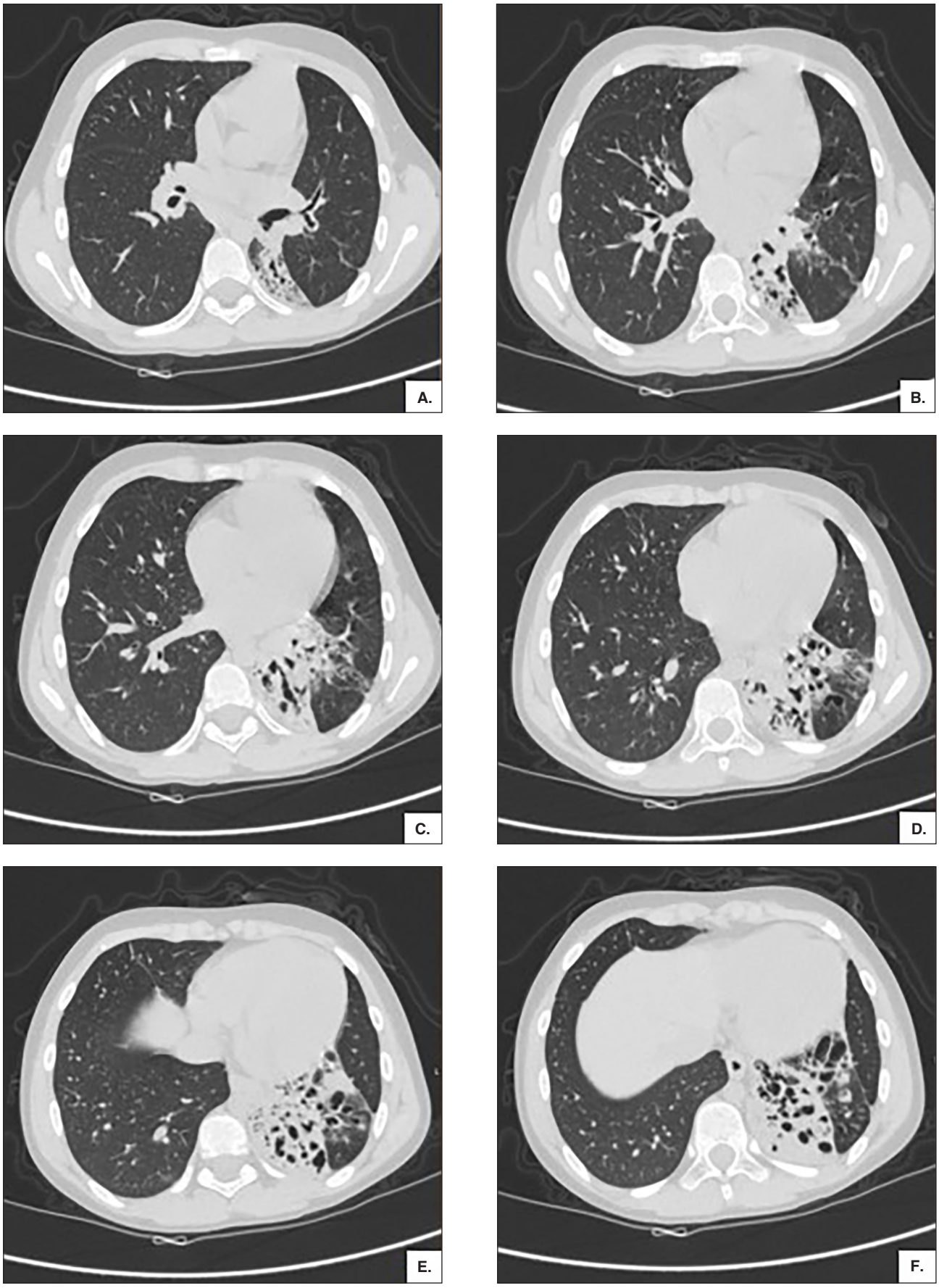

Figure 2. Chest computed tomography (CT) scan presented in image series (**A** to **F**) revealed exuberant cystic bronchiectasis occupying almost the entire left lower lobe, with foci of airy parenchymal consolidation suggesting an infectious process.

cystic bronchiectasis occupying almost the entire left lower lobe, with foci of airy parenchymal consolidation suggesting an infectious process. Cefuroxime was started (10 days total) with clinical improvement. Further etiological investigation included spirometry, flow cytometry, serum levels of IgM, G, A and E, serum C3, C4 and CH50 levels, measurement of alpha-1 antitrypsin and two sweat chloride tests that were within normal range. Bronchoscopy showed easily remade purulent secretions from the left inferior lobe, with no evidence of foreign bodies.

After discharge he continued to have bronchorrhea requiring respiratory kinesiotherapy.

The patient later underwent an elective thoracoscopic lobar resection with no postsurgical complications. The surgical specimen presented macroscopically as a multiple cystic lesion. Histological examination confirmed a congenital cystic adenomatoid malformation Stocker type 1 (**Fig. 3**). There was no evidence of malignancy. On long-term follow-up, the patient remained asymptomatic, with normal respiratory function tests.

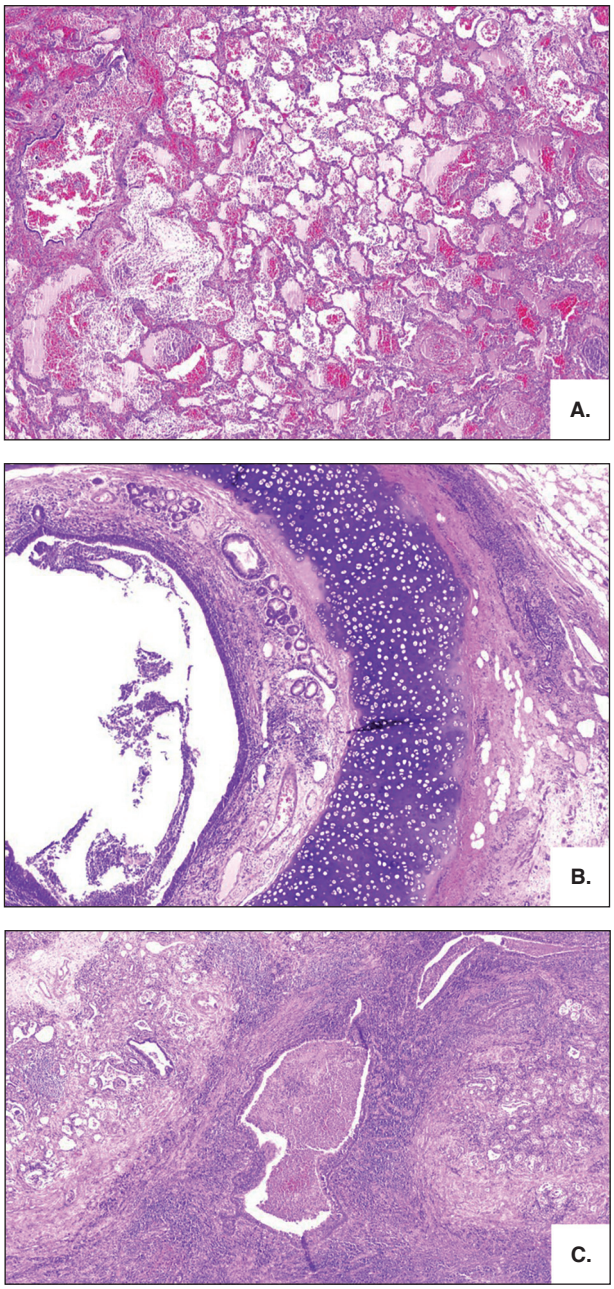

Figure 3. Anatomopathological image collection. **A.** Bronchiectasis. **B.** Intra-alveolar hemorrhage. **C.** Dilated cystic structures lined by ciliated columnar epithelium, within which a polymorphic inflammatory infiltrate can be seen with the participation of neutrophils.

Discussion

CPAM is rare, despite accounting for the majority of congenital cystic lung diseases and being the most commonly diagnosed by prenatal ultrasound screening [5]. However, the echogenicity of the lesions becomes increasingly similar to normal lung during the latter stages of pregnancy [4], a phenomenon that might camouflage previously undetected lesions. Considering asymptomatic cases at birth, the fraction that will eventually develop symptoms will tend to do so within the first year of life [5], and this case supports this tendency. However, as seen in this report, when the initial manifestation is of an infectious nature, the non-specific radiological findings may mask the underlying problem.

In the presence of post-natal symptoms, a surgical approach is somewhat consensual, but clinical stabilization is advised before any therapeutic decisions [3]. Lobectomy is recommended for most parenchymal malformations to prevent postoperative air leaks, residual disease, and perhaps reduce the risk of some later malignancies [3]. Our postoperative outcome, despite the extension of the lesion, probably exacerbated by repeated infectious events, supports the already described good surgical prognosis whatever the age at surgery [5]. Nevertheless, the undue presence of this lesion may lead to severe and late complications in paucisymptomatic patients, or in those whose symptoms are overlooked, and may compromise the success of an impending surgery.

In patients diagnosed later in life, the natural history of untreated CPAM can be inferred. The severe complications that may arise add value to the benefits of early surgical intervention in asymptomatic cases at birth, especially in macrocystic lesions [6]. The optimal timing remains controversial, and is highly dependent on the delicate balancing of many aspects, the most pointed out being the possibility of a spontaneous

regression, radiation burden, assuring patient follow-up and the increased surgical morbidity in the event of a severe infection. A more conservative approach in asymptomatic patients may favor postponing resection until the second birthday (versus intervention in the first months), ensuring at least one more intermediate CT reassessment, usually between 12-18 months (apart from the initial one, at 1-6 months) [3].

Conclusion

Although CPAMs are most commonly detected prenatally or during the neonatal period, in some rare cases diagnosis can occur much later in life. Therefore, recurrent pneumonia and chronic symptoms such as productive cough should be actively sought and evaluated in routine visits, and should prompt further investigation, including imaging targeting adequate characterization of the parenchyma. Despite the overall good surgical prognosis, even in symptomatic patients and whatever the age, the potential complications of the untreated CPAM favor early surgical intervention in patients asymptomatic at birth, especially in the presence of a macrocystic lesion, although the exact timing is still controversial.

Informed consent

Patient's informed consent was obtained.

Declaration of interest

The Authors declare that there is no conflict of interest. Funding, financial support, and material support: none.

References

1. Kantor N, Wayne C, Nasr A. Symptom development in originally asymptomatic CPAM diagnosed prenatally: a systematic review. Pediatr Surg Int. 2018;34(6):613-20.
2. Leblanc C, Baron M, Desselas E, Phan MH, Rybak A, Thouvenin G, Lauby C, Irta S. Congenital pulmonary airway malformations: state-of-the-art review for pediatrician's use. Eur J Pediatr. 2017;176(12):1559-71.
3. Annunziata F, Bush A, Borgia F, Raimondi F, Montella S, Poeta M, Borrelli M, Santamaria F. Congenital Lung Malformations: Unresolved Issues and Unanswered Questions. Front Pediatr. 2019;13;7:239.
4. Seear M, Townsend J, Hoepker A, Jamieson D, McFadden D, Daigneault P, Glomb W. A review of congenital lung malformations with a simplified classification system for clinical and research use. Pediatr Surg Int. 2017;33(6):657-64.
5. Kantor N, Wayne C, Nasr A. Symptom development in originally asymptomatic CPAM diagnosed prenatally: a systematic review. Pediatr Surg Int. 2018;34(6):613-20.
6. Swarr DT, Peranteau WH, Pogoriler J, Frank DB, Adzick NS, Hedrick HL, Morley M, Zhou S, Morrisey EE. Novel Molecular and Phenotypic Insights into Congenital Lung Malformations. Am J Respir Crit Care Med. 2018;15;197(10):1328-39.