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ORIGINAL ARTICLE

Congenital lobar emphysema: a 10-year case series review

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Abstract

Introduction and Objectives: Congenital lobar emphysema (CLE) is a rare congenital lung malformation that causes one or more pulmonary lobes to overinflate as a result of an air trapping-like mechanism and it may cause progressive respiratory distress. It is diagnosed in one in every 20,000 - 30,000 neonates. Diagnosis is usually made postnatally, with most cases identified in the first six months of life, but advances in ultrasound technology have allowed for some prenatal diagnosis. We report on the presentation, diagnostic methods, and management of a series of cases of CLE. **Methods:** Review of electronic files of patients diagnosed with CLE, over the past 10 years. **Results:** We identified four patients diagnosed with CLE: three female and one male. Two of these patients were diagnosed with CLE prenatally and remained asymptomatic for the entire recorded period, and the other two developed symptoms soon after birth. Chest radiographic imaging was performed in all four patients and a CT scan was carried out in three cases. The symptomatic cases required lobectomy and follow-up showed a slight decrease in residual volumes, total lung capacity, and forced expiratory volume, with no long-term pulmonary dysfunction. **Discussion:** Prenatal diagnosis of CLE is becoming more feasible with advances in imaging technology. In our population, we observed an inverted male-to-female ratio. Patients may remain asymptomatic and conservative management is advocated for such cases. When patients start to develop symptoms, there are progressive signs of increasing respiratory distress and, for these cases, surgery is recommended, with open thoracotomy being the preferred approach. Post-surgical complications are minimal and long-term pulmonary function remains stable.

Keywords: Congenital lobar emphysema. Congenital lung disease. Lung overinflation. Pulmonary emphysema.

Enfisema pulmonar congénito: uma revisão de casos de 10 anos

Resumo

Introduçã e Objetivos: O Enfisema Lobular Congénito (CLE) é uma malformação pulmonar congénita rara com consequente hiperinsuflação de um ou mais lobos pulmonares, devido à retenção de ar no seu interior e que pode conduzir a progressiva dificuldade respiratória. Ocorre em 1 em cada 20.000-30.000 recém-nascidos. Frequentemente, diagnostica-se no pós-natal, em particular nos primeiros seis meses de vida, embora o desenvolvimento da ecografia tenha permitido diagnósticos pré-natais. Apresentamos a clínica, os métodos diagnósticos e a orientação numa série de casos de CLE. **Métodos:** Revisão de todos os processos de doentes com o diagnóstico de CLE no nosso centro, nos últimos 10 anos.

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Resultados: Identificamos 4 doentes diagnosticados com CLE: 3 do sexo feminino e 1 masculino. Dois tiveram diagnóstico pré-natal e mantiveram-se assintomáticos durante o seguimento, enquanto que os outros dois desenvolveram sintomas pouco após o nascimento. Todos realizaram radiografias de tórax e três realizaram tomografia computadorizada. Os casos sintomáticos necessitaram de lobectomia, tendo-se verificado, durante o seguimento, ligeira diminuição dos volumes residuais, capacidade pulmonar total e volume expiratório forçado, mas sem disfunção pulmonar a longo prazo. **Discussão:** O diagnóstico pré-natal de CLE tem sido mais frequente, particularmente com o desenvolvimento da ecografia. Constatámos uma inversão da relação entre sexos no nosso grupo de doentes. Para os doentes assintomáticos preconiza-se atitude expectante e vigilância. Para os doentes que desenvolvem sintomas, como aumento gradual de dificuldade respiratória, recomenda-se a abordagem cirúrgica por toracotomia aberta; as complicações pós-cirúrgicas são mínimas, e a função pulmonar permanece estável a longo prazo.

Palavras-chave: Enfisema pulmonar congénito. Doença pulmonar congénita. Sobredistensão pulmonar. Enfisema pulmonar.

Keypoints

What is known
- CLE is a congenital malformation with overinflation of pulmonary lobes and progressive respiratory distress.
- Symptoms usually develop in the first six months of life.

What is added
- Asymptomatic patients can be managed conservatively.
- Follow-up studies have shown no long-term pulmonary dysfunction.

Introduction

Congenital lobar emphysema (CLE) is a rare type of congenital lung malformation (CLM). It was first described in 1932 by Nelson[1,2] and its incidence is estimated to be about one in every 20,000 to 30,000 live births. CLE is more common among Caucasians, with a male-to-female ratio of 1:3[3-6].

CLE is characterized by the overexpansion of one or more pulmonary lobes[6] due to partial bronchial obstruction that creates a “ball-valve” mechanism, leading to air trapping in the affected lobe[2,4,5]. Symptoms are present at birth in up to 33% of cases and almost all infants will develop symptoms within the first six months of life if there is continued lobar overdistension and depending on how fast it develops[2-5,7,8]. Infants typically present with tachypnea, shortness of breath, wheezing, cyanosis, and difficulty in feeding. An important aspect to take into consideration is the possible association of CLE with other congenital malformations: cardiac (the most common types, e.g. patent ductus arteriosus, pulmonary stenosis or valve atresia, and aortic coarctation), vascular (e.g. double superior vena cava), renal (aplastic kidney), gastrointestinal (e.g. omphalocele), and syndromes (e.g. Williams-Beuren syndrome and Niemann-Pick disease)[2,5,9-11].

Methods

We identified all patients with a diagnosis of CLE in our center, since 2010, and reviewed the electronic files to describe clinical presentation, diagnostic methods, imaging techniques used, management options, and development during follow-up.

Results

The patients were all term neonates and Caucasian, one male and three female; three were inborn and one was outborn. One patient presented to the emergency department and another, at two months old, was transferred from another hospital following diagnosis. The demographics and clinical characteristics are presented in table 1.

Neonates (patients 2 and 3 in Table 1) had a prenatal diagnosis and were asymptomatic at birth. Another neonate (patient 1, in Table 1) developed symptoms on day 14. The fourth patient displayed mild symptoms from birth, but was mistakenly diagnosed with transient tachypnea of the newborn and correct diagnosis was not made until two months of age. Presenting features included sleepiness, refusal to feed, inadequate weight gain, and shortness of breath.

Management and follow-up details are provided in table 2. All patients had chest x-rays (Figs. 1, 3, 4 and 5) compatible with CLE in the left upper lobe and one had a (CT) scan (Figs. 1, 3 and 4) for diagnostic purposes. The two patients with a prenatal diagnosis are closely followed up at the outpatient department. The two cases with moderate-to-severe symptoms underwent left upper lobectomy via thoracotomy and, in both cases, the histopathology study (Fig. 2 and 5) revealed diffused emphysematous lesions with no parenchymal destruction.

Table 1. Demographics and clinical characteristics

Patient	Gender	Caucasian	Prenatal diagnosis	Age at diagnosis	Start of symptoms	Symptoms
1	Female	Yes	No	14 days	14 days	Sleepiness, refusal to feed, perioral cyanosis, and subsequent shortness of breath
2	Female	Yes	Yes. Ultrasound at 22 weeks of gestation and MRI at 25 weeks	NA	NA	Asymptomatic
3	Female	Yes	Yes. Ultrasound at 22 weeks of gestation, MRI at 24 weeks	NA	NA	Asymptomatic
4	Male	Yes	No	2 months	Day 1	Poor weight gain, irritability, wheezing, costal retraction, groaning

NA: not applicable; MRI: magnetic resonance imaging.

In all four patients, whether during their initial admission or follow-up, an ultrasound examination (including abdominal and renal) and echocardiogram were performed to identify potential associated congenital malformations, but none were found.

Discussion

This series of four clinical cases of CLE recorded over a 10-year period, at a level III hospital with about 2,500 deliveries per year, highlights the rarity of this pathology. CLE is a rare disease, with an unknown cause in 50% of cases. It is believed that a partial obstruction of the lobar bronchi leads to air trapping and the consequent overinflation and compression of adjacent structures as the affected lobe continues to grow and herniates leading to a deviation of the mediastinum and compression of the structures on the opposite lung. This lobar involvement is most commonly found in the left upper lobe (43%), followed by the right middle lobe (32%) and the right upper lobe (21%), while lower lobe involvement is less common and the rarest form is polylobar involvement[5,6].

Diagnosis can be made using chest X-rays showing a hyperlucent lobe, that may herniate and cause ipsi- and even contralateral atelectasis and mediastinal shift[3-5,9]. Although CT scans are considered the gold standard for diagnosing CLE, they may not be always necessary, especially when symptoms and a chest X-ray clearly point to the condition. CT scans are valuable in confirming a suspected diagnosis and provide a more detailed characterization of lung involvement[2,5]. Bronchoscopy was once commonly performed to exclude intraluminal causes for obstruction but it is no longer routinely performed[5,9].

Histologically, there are overgrown alveoli and the acinar structure is preserved with no lung tissue destruction or other parenchymal abnormalities[2,5,6]. While the etiology is unknown in about half of cases, a quarter have absent, hypoplasic, or dysplasic bronchial cartilage and, in rare cases, there may be intrinsic or extrinsic causes for the airway obstruction[5,6]. In this series, the two patients that underwent lobectomy had histopathology reports that described emphysematous-like lesions, with multifocal ruptured septa, which was associated with the surgery itself rather than the pathology. Dysplasic or absent cartilage was not observed, which is in line with the literature[5].

Some studies have recently described the prenatal diagnosis of CLE using ultrasound and confirming it with magnetic resonance imaging (MRI)[2,6,8,10]. In some cases, a regression of the lesion is described, avoiding the need for fetal or postnatal intervention[2,6,8,10].

Contrary to previous literature, our series presents a male-to-female inverted ratio, but all our patients were Caucasian, as usually reported. Traditionally, diagnosis is made postnatally, with most cases identified in the first six months of life. However, with advances in ultrasound technology and awareness, prenatal diagnosis has been described by some authors[2,6,10]. In our series, two cases were diagnosed prenatally and were asymptomatic, one presented with symptoms at 14 days of life, and the fourth was only diagnosed at two months of age, despite symptom progression from birth.

All patients had a chest X-ray, which enabled diagnosis; three had a CT scan, but only in one case

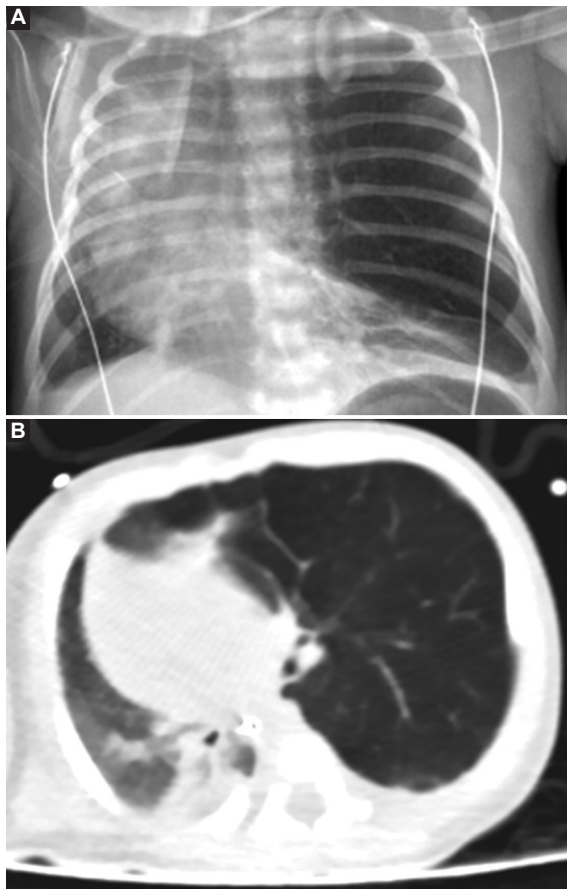

Figure 1. A: chest X-ray: hyperlucent LUL, mediastinum shift and atelectasis. **B:** CT scan: large-volume CLE, pushing the mediastinum to the opposite side.

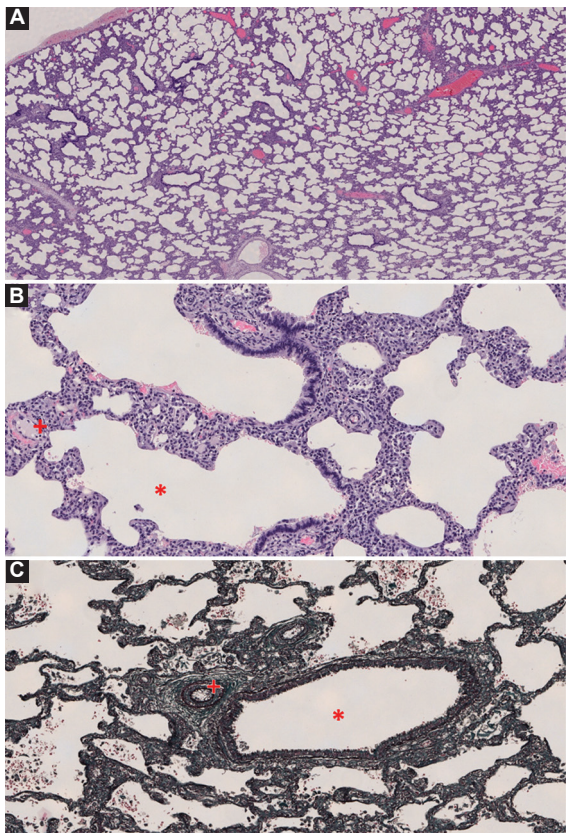

Figure 2. Histopathology images. **A:** H&E, x1.25: alveolar spaces are enlarged, but the global architecture is structurally normal; there is also lung immaturity (there were no periodic acid-Schiff positive cells or exudates). **B:** and **C:** H&E, x100; elastic fiber stain, x100 respectively: not only are alveolar spaces markedly enlarged, but also the bronchioles (*) are wider than matching arterioles (+).

(patient 4) was it necessary for diagnosis, since the image on the chest X-ray was not specific enough. The left upper lobe was affected in each case, which is in line with the literature. None of our patients underwent a bronchoscopy, as this exam is no longer routinely performed since the advent of CT[9]. Most patients who underwent a CT scan did so in order to better characterize the extension of lobar involvement, the number of lobes affected, and to evaluate the compromise of the surrounding structures[2,6,7,10]. An echocardiographic evaluation was also performed to rule out a congenital heart defect associated with CLE and no major abnormalities were found.

According to recent reports, patients with few or no symptoms can be managed conservatively, which involves close follow-up[2,5-7,10]. However, once there is any deterioration of the clinical condition with development or worsening of the symptoms, surgery is the best option. This approach has been described more frequently and shows no different outcomes compared to the normal population[2,8,10].

There are two surgical approaches available: open thoracotomy and thoracoscopy surgery. In our series, the two patients who were operated upon underwent an open thoracotomy as it is currently the preferred method. Typically, the surgery is well tolerated and recovery is fast, with a good prognosis. In our series, one patient (number 1, in Table 2) developed post-operative anemia and a red blood cell transfusion was needed, and the same patient developed an opioid withdrawal syndrome. To date, no other complications have been recorded. No other post-operative complications were reported, and no respiratory symptoms have been observed in the follow-up appointments. Without

Table 2. Study, management, and outcome

Patient	Imaging study	Affected lobe	Histopathology	Cardiology evaluation	Lobectomy	Compli-cations	Outcome
1	Chest X-Ray (D14) –hyperlucent LUL, mediastinum shift, and atelectasis	LUL	Global structure preserved, but there are enlarged alveolar spaces; bronchioles are wider than matching arterioles	PFO at 1 month	Yes (17 days)	Anemia; Opioid withdrawal syndrome	Asymptomatic
2	Chest X-Ray (D2) –hyperlucent LUL CT scan (2 months): single CLE with small parathymic atelectasis; no mediastinal shift	LUL	NA	Mesocardia; minimal VSD	No	NA	Asymptomatic
3	Chest X-Ray (D1) –hyperlucent LUL CT scan (6 months): minor CLE; no atelectasis or mediastinum shift	LUL	NA	Normal	No	NA	Asymptomatic
4	Chest X-Ray (2 months) – hyperlucent LUL and mediastinum shift; no atelectasis CT scan (2 months) – left CLE; right lobe atelectasis and right mediastinum shift	LUL	Global structure preserved, but there are enlarged alveolar spaces	Normal	Yes (2 months)	No	Asymptomatic

CLE: congenital lobar emphysema; CT: computed tomography; VSD: ventricular septal defect; NA: not applicable; PFO: patent foramen ovale; LUL: left upper lobe.

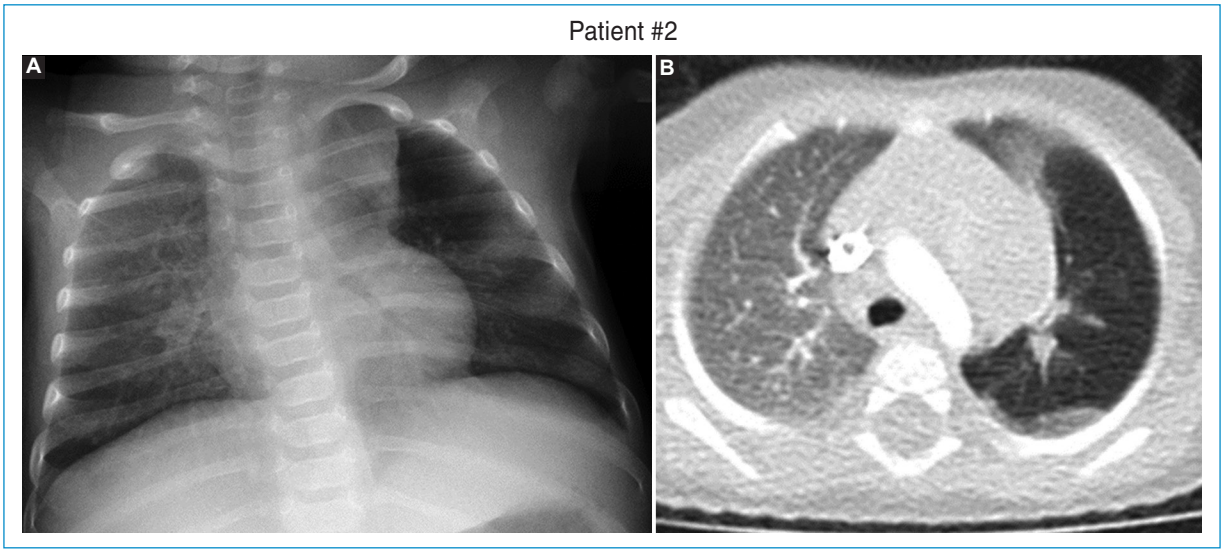

Figure 3. **A:** chest X-ray: hyperlucent LUL, with no mediastinum shift. **B:** CT scan: single CLE with small parathymic atelectasis; no mediastinal shift.

Patient #3

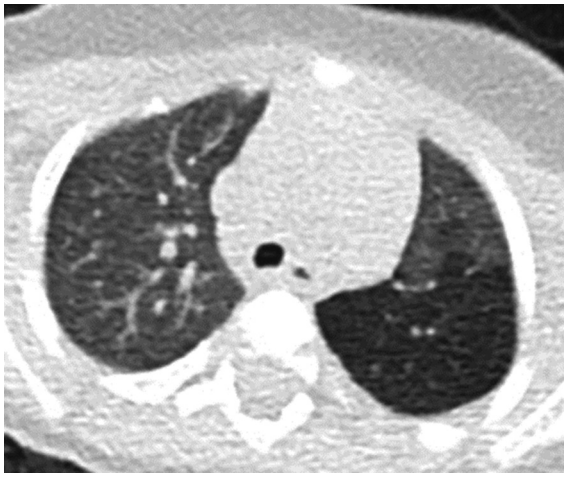

Figure 4. CT scan at 6 months of age: minor right posterior CLE; no atelectasis or mediastinum shift.

Patient #4

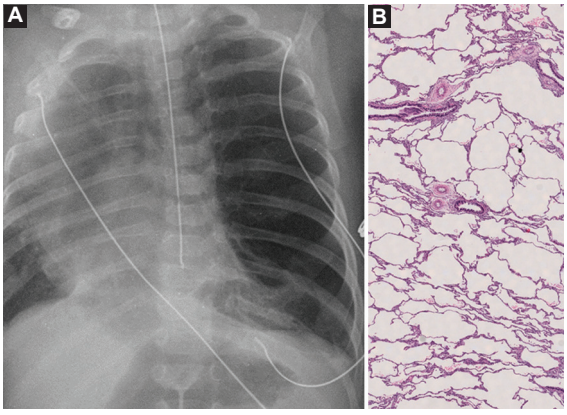

Figure 5. A: chest X-ray at 2 months of age: hyperlucent LUL and mediastinum shift; no atelectasis.
B: histopathology image (H&E, x1.25): alveolar spaces are enlarged, in this case markedly, but the global architecture is structurally normal.

surgery, the natural history of CLE is eventual progressive cardiorespiratory failure due to the compression effect over ipsi- and contralateral lung tissue as well as over the venous drainage from the pulmonary and systemic bed leading to heart failure[2,5,10].

Follow-up studies on patients who underwent surgery as well as on those who were managed conservatively showed slight decreases in residual volumes, total lung capacity, and forced expiratory volume, but no long-term pulmonary dysfunction was noted.

More recently, some authors have claimed a probable genetic predisposition, with an apparent autosomal dominant transmission[4,5], but more studies are needed in order to confirm this hypothesis.

In conclusion, CLE is a rare disease with unknown etiology in 50% of cases. The diagnosis of CLE can be made prenatally or postnatally. Imaging techniques, such as chest X-ray, in some specific cases, and CT scans are useful for diagnosis, and echocardiographic evaluation is recommended to identify any associated malformations. Conservative treatment of CLE should be preferred in mild and moderate cases, while lobectomy should be considered in severe disease cases. As more cases are diagnosed prenatally, conservative treatment seems to be increasing when intrauterine regression of CLE is observed. Histologically, broncho-alveolar involvement with over-distension of the affected lobe and no destruction of surrounding pulmonary parenchyma, is typically described. Follow-up studies have shown that there is no long-term pulmonary dysfunction and the prognosis is generally good with surgery or conservative management.

Although CLE is rare, a high index of clinical and radiological suspicion is required to diagnose this rare anomaly which may mimic other causes of respiratory distress. Therefore, in order to provide the best care for affected patients, it is important that clinicians are aware of this entity.

Authors' contribution

A. Assunção, P. Miragaia, F. Flôr-de-Lima, G. Rocha, C. Ferraz and I. Azevedo: Conception and design of the study, report, review or other type of work or paper; Acquisition of data either from patients, research studies, or literature; Anayisis or interpretation of data either from patients, research studies, or literature; Drafting the article; Critical review of the article for important intellectual content; Final approval of the version to be published; Agreement to be accountable for the accuracy or integrity of the work. Susana Guimarães: Conception and design of the study, report, review or other type of work or paper; Acquisition of data either from patients, research studies, or literature.

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Conflicts of interest

None.

Ethical disclosures

Protection of human and animal subjects. The authors declare that no experiments were performed on humans or animals for this study.

Confidentiality of data. The authors declare that they have followed the protocols of their work center on the publication of patient data.

Right to privacy and informed consent. The authors have obtained approval from the Ethics Committee for analysis and publication of routinely acquired clinical data and informed consent was not required for this retrospective observational study.

Use of artificial intelligence for generating text. The authors declare that they have not used any type of generative artificial intelligence for the writing of this manuscript, nor for the creation of images, graphics, tables, or their corresponding captions.

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