Congenital pulmonary lymphangiectasis mimicking congenital pulmonary interstitial emphysema in a patient with congenital diaphragmatic hernia

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ABSTRACT

Congenital pulmonary lymphangiectasis is a rare abnormality of the lung characterized by dilated pulmonary lymphatic vessels. The diagnosis of congenital pulmonary lymphangiectasis is challenging due to its unspecific presentation, as well as possible radiological imitation of interstitial emphysema. We present a patient where diagnosis was hampered by the extremely rare combination of pulmonary lymphangiectasis and congenital diaphragmatic hernia.

1. Introduction

Congenital pulmonary lymphangiectasis (CPL) – or congenital pulmonary lymphangiectasia – is a rare disorder characterized by dilatation of the pulmonary lymphatic vessels. It usually presents as respiratory distress in the newborn and is associated with a poor prognosis [1]. Due to its unspecific clinical presentation and possible radiological imitation of interstitial emphysema, CPL is difficult to diagnose.

Here we present a case of CPL, where diagnosis was obscured by the extremely rare combination with a congenital diaphragmatic hernia (CDH). We discuss the reasons of possible misdiagnosis and the etiology of CPL in this patient with CDH, as well as a possible connection between these two congenital disorders.

2. Case report

A male patient with prenatally diagnosed left CDH was delivered in a tertiary medical center at 38 2/7 weeks of gestation after an uncomplicated pregnancy. Following immediate intubation, the patient was stabilized on low settings. Associated pathologies were excluded by sonography, except for a small persisting foramen ovale and an atrial septum aneurysm. Diaphragmatic hernia repair with direct closure of the defect (no sac) was performed on the 3rd day of life. Extubation was achieved on the 9th postoperative day after an uneventful course. While inserting a new central line, the left thoracic cavity with the CDH-related hypoplastic lung was unintentionally punctured and the resulting aspirate proved to be chyle. Despite switching to a lipid-free diet, respiratory deterioration occurred due to increasing chylothorax. Drainage transiently improved respiration, but in his fourth week of life, the patient presented again with respiratory insufficiency, requiring non-invasive ventilation. X-ray showed a hyperinflated left lung with interstitial changes. In the course of the next two weeks, hyperinflation worsened, and an increasing mediastinal shift towards the healthy right side was noted (Fig. 1). CT showed a dysplastic left lung with a thickened interstitium and air filled spaces as seen in interstitial emphysema (Fig. 2). A one-sided ventilation of the healthy right lung through blockage of the left main bronchus with an endobronchial blocker resolved mediastinal shift and improved respiration, however unblocking was followed by rapid

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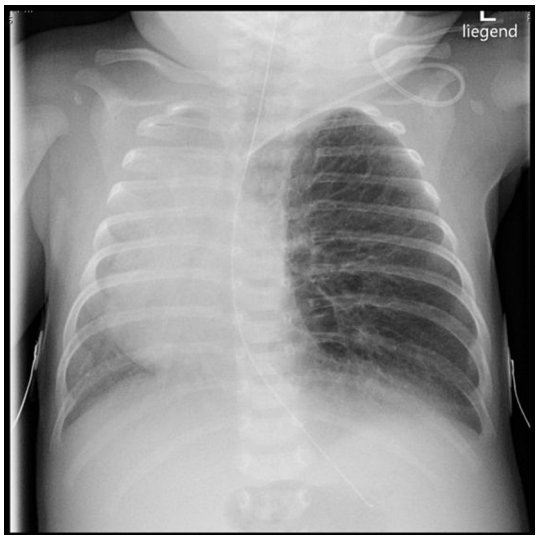

Fig. 1. Chest x-ray showing hyperinflation of the left lung and mediastinal shift towards the right.

deterioration. Therefore, pneumonectomy of what seemed to be an emphysematous and most likely nonfunctional lung that compressed the right side was decided as a last resort. Left pneumonectomy was performed at 6 weeks of life. Pathologic examination of the resected lung specimen showed diffuse small cystic lesions measuring up to 1 cm (Fig. 3). Histologically and immunohistochemically, these cystic spaces were lined with flat endothelial cells expressing CD31 and D2-40, i.e. markers for lymphatic vessels. Based on these findings, diagnosis of CPL was made.

The postoperative course was uneventful and the patient recovered well. He was discharged from the hospital at 12 weeks of life. Now at 14 months of life he is doing well and shows complete respiratory compensation.

3. Discussion

Our case reveals two noteworthy points. Firstly, the combination of CDH and unilateral CPL is extremely rare. Secondly, CPL was mistaken for interstitial emphysema.

Noonan et al. [2] divided CPL into three subgroups. In primary CPL, lymphangiectasis is confined to the lung and thought to be due to a developmental defect of the lymphatic system. This condition can be sporadic or associated with a number of congenital and genetic diseases, e.g. Noonan, Ullrich-Turner, and Down syndromes [3]. Secondary CPL is due to pulmonary venous or lymphatic obstruction, mainly attributable to cardiovascular malformations such as hypoplastic left heart syndrome, pulmonary vein atresia, congenital mitral valve stenosis, cor triatum, and thoracic duct agenesis [1]. Some have also suggested infectious agents as a possible etiology of secondary CPL [4]. The third group is represented by patients with generalized lymphangiectasis wherein the pulmonary involvement is just one of many affected systems. Independent of its etiology, localized forms of CPL with involvement of one or two lobes are rare [5].

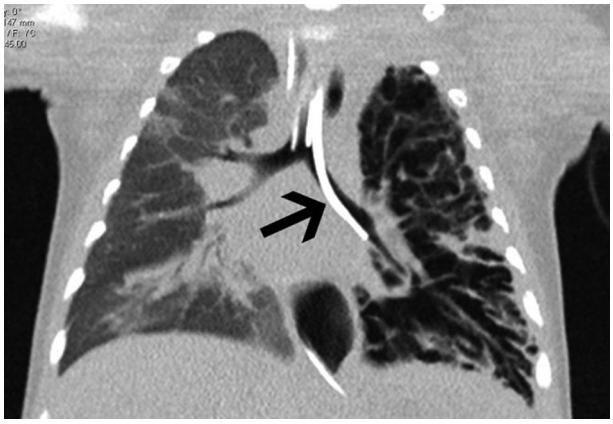

Fig. 2. CT Thorax showing the dysplastic left lung with a thickened interstitium and air filled spaces, as well as a decreasing mediastinal shift due to obstruction of the left main bronchus by a bronchial blocker (arrow).

CPL usually presents at birth with severe respiratory problems. Often, pleural effusions are evident. Diagnosis is primarily clinical, though radiological evidence of increased interstitial markings, pleural effusions, and hyperinflation may be helpful [6]. Prenatal signs for CPL may be a hydrops fetalis with hydrothorax [7].

The combination of CPL and CDH has, to the best of our knowledge, only been reported twice [8,9].

One patient [8] was found to have hydrothoraces bilaterally on prenatal scans and a possible congenital cystadenomatoid malformation in the left lower lobe. However, postnatal diagnosis of a left-sided CDH was made and the hernia was repaired. The postoperative course was complicated by generalized edema and respiratory distress. A chest CT showed changes consistent with CPL. Treatment was withdrawn on 38 days of life due to increasingly difficult management. Postmortem studies confirmed bilateral CPL. The other patient [9] was postnatally diagnosed with CDH due to perinatal respiratory distress. After hernia repair, the postoperative course was complicated by bilateral pneumothoraces. The placement of chest-tubes did not improve the patient's condition and he died. Postmortem studies showed bilateral CPL.

In contrast, our patient's CDH was known prenatally, CPL was only unilateral and the respiratory distress due to CPL presented only after an initially unremarkable postoperative course. As previously described by others, localized CPL can mimic congenital lobar emphysema (CLE) radiologically, as well as clinically [5,10,11]. The dilated lymphatics are filled with air, making a radiological differentiation very difficult. Most likely, this phenomenon is due to tears of the lymphatic walls allowing air to enter the lymphatic system [5]. The final differentiation between CPL and CLE can only be made histologically. In our case, the diagnostic process was complicated by the concomitant CDH. Some radiological features were disguised by the CDH: the interstitial changes were attributed to the CDH-related hypoplastic lung, the chylothorax was interpreted as an often seen complication after CDH repair. Prenatal MRI had not given any hints that an additional pulmonary pathology may be present, thus alertness was not increased. Due to the radiological evidence, CLE seemed the most likely diagnosis. The pathology report however revealed the final and conclusive diagnosis of CPL.

Although pneumonectomy in pediatric patients is associated with high morbidity [12], it was unavoidable in our case due to exhaustion of conservative therapy. This remains true even if the correct diagnosis of CPL instead of CLE would have been made preoperatively. The unilateral involvement in our case made a curative approach by pneumonectomy possible. This is in line with the few localized cases of CPL in the literature, where a good outcome was reported after surgical removal of the affected lobe [10,11,13–15]. In contrast, generalized forms have a poor prognosis.

The etiology of the unilateral lymphangiectasis in this case remains unclear. A unilateral primary CPL is a possibility. It is also conceivable that CDH-induced displacement of the lung leads to

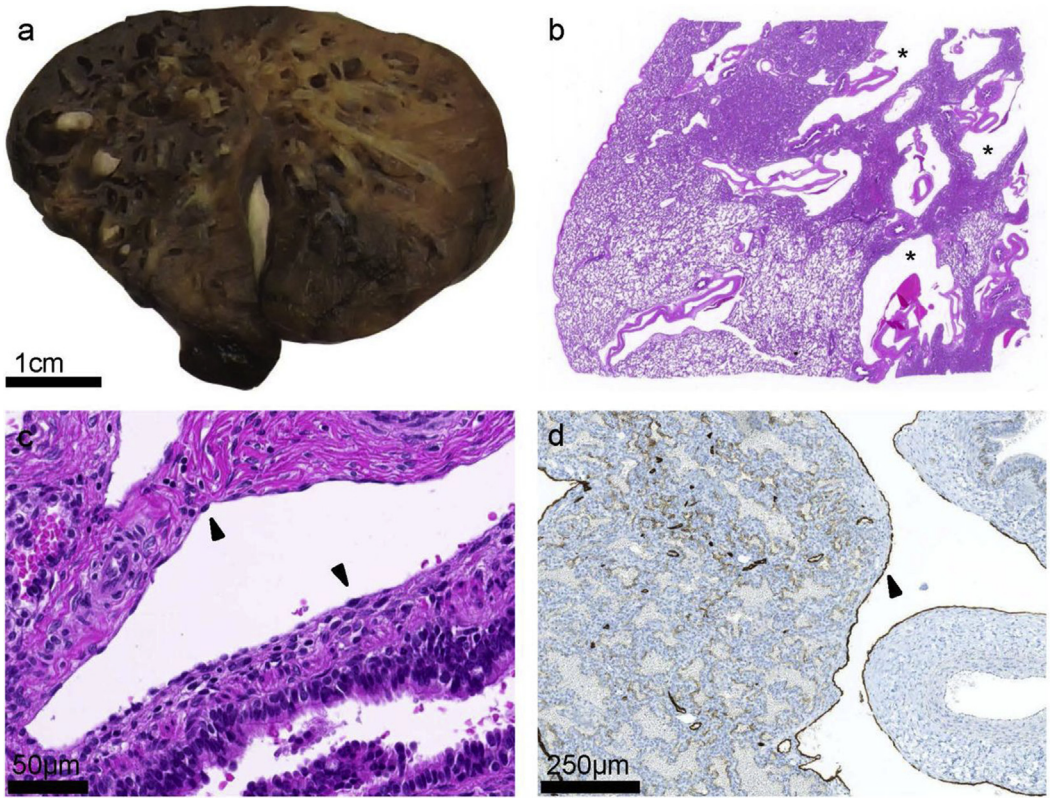

Fig. 3. Pathological work-up. a: macroscopically visible diffuse small cystic lesions measuring up to 1 cm. b: microscopically visible diffuse small cystic lesions (*) (H&E stain, overview). c: the air-filled cystic lesions are lined by flat endothelial cells (arrow) (H&E stain, 400× magnification). d: Immunohistochemistry for D2-40 (arrow) highlights lymphatic vessels (100× magnification).

hilar flow changes which may contribute to lymphatic flow obstruction and thus to the formation of secondary lymphangiectasis. Yet, the combination of CPL and CDH would then not be that rare. Another tenable hypothesis is that there is a direct relation between CDH and CPL. Chylothorax has in fact been linked to CPL [16]. But chylothorax is also a well-known complication after CDH surgery with unclear etiology. Surgical injuries seem the most likely cause, but explanations remain unsatisfactory. However, a recent mouse model [17] showed that chylothorax in CDH may be due to dysfunctional lymphatic development. It seems, therefore, plausible that the lymphangiectasis in our case could be an extreme form of lymphatic maldevelopment that can be present in CDH.

4. Conclusion

This case report highlights the rare combination of CPL and CDH, and the difficulty to diagnose CPL, which can be misdiagnosed as CLE. Localized forms of CPL have a good prognosis due to potential curative surgical treatment. Possible connections between CPL and CDH remain speculative and are in need of further research.

Conflict of interest

The authors declare that they have no conflict of interest.

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Informed consent

Informed consent was obtained from the patient's legal guardians.

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