Congenital Lobar Emphysema: A diagnostic dilemma with co-existent Congenital Heart Defects

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Statement of patient informed consent for all the participants involved in the study was waived off as the data collected was retrospective and based on observational inputs. After perusal, Institutional Ethics Committee -2 (IEC-2), approved the project vide letter IEC/BU/2022/Cr.04/125/2023 dated 26.04.2023.

Abstract

Congenital lobar emphysema (CLE) is a rare and life-threatening lung anomaly that often poses a diagnostic dilemma. Co-existent congenital heart defects (CHD) with left-to-right shunts can further complicate the diagnosis of CLE. The presence of underlying CLE may only be suspected after the repair of CHD. Here, we discuss a 2-month-old patient who presented with respiratory distress and was identified to have a large ventricular septal defect (VSD) and patent ductus arteriosus (PDA). After the successful repair of the cardiac defects, the infant continued to experience respiratory distress and failure to thrive. CLE was subsequently diagnosed on imaging.

Key Words:

Left-to-Right shunt, CT scan lung, Right middle lung lobe, Ventricular Septal Defect, Patent Ductus Arteriosus, Respiratory distress, Failure to thrive, Congenital lobar emphysema.

Introduction

Congenital lobar emphysema (CLE) is a rare and life-threatening anomaly of the lung characterized by hyperinflation of at least one lung lobe. The incidence of CLE is approximately 1 in every 20,000 to 30,000 live births, with a 3:1 male preponderance[1,2]. The hyperinflated lobe compresses the surrounding normal lung tissue, causing atelectasis and leading to ventilation-perfusion mismatch and hypoxia. It usually presents in early infancy with respiratory distress[3,4]. The most common site of lobe involvement is the left upper lobe (43%), followed by the right middle lobe(32%), and the right upper lobe (21%).

Congenital heart defects (CHD) can present in around 14–20% of CLE cases, with ventricular septal defect (VSD) being the most commonly associated anomaly[1,4]. The clinical presentation of CLE and most CHDs is similar and manifests in infancy.As CHD is more common, it is usually detected first.Often, the diagnosis of CLE may get overlooked in the presence of CHD[5,6]. Here, we present a case in which the presence of CLE was established after the corrective repair of a large perimembranous VSD with a moderate patent ductus arteriosus (PDA).

Case Presentation

A one – month – and – twenty – four – day - old male presented with cough, tachypnea, and minimal chest retraction of short duration. The child was afebrile and exhibited moderate activity and sucking. The child's weight and height on admission were 3.5 kgs. and 52 cms., respectively, and both of the parameters were $< 3^{rd}$ percentile. His birth weight was 2.6 kgs., suggesting poor weight gain. The child was on mixed feed (breastfeeding and cow's milk) from one month of age. The X-ray chest demonstrated right upper lobe pneumonia (Figure 1).

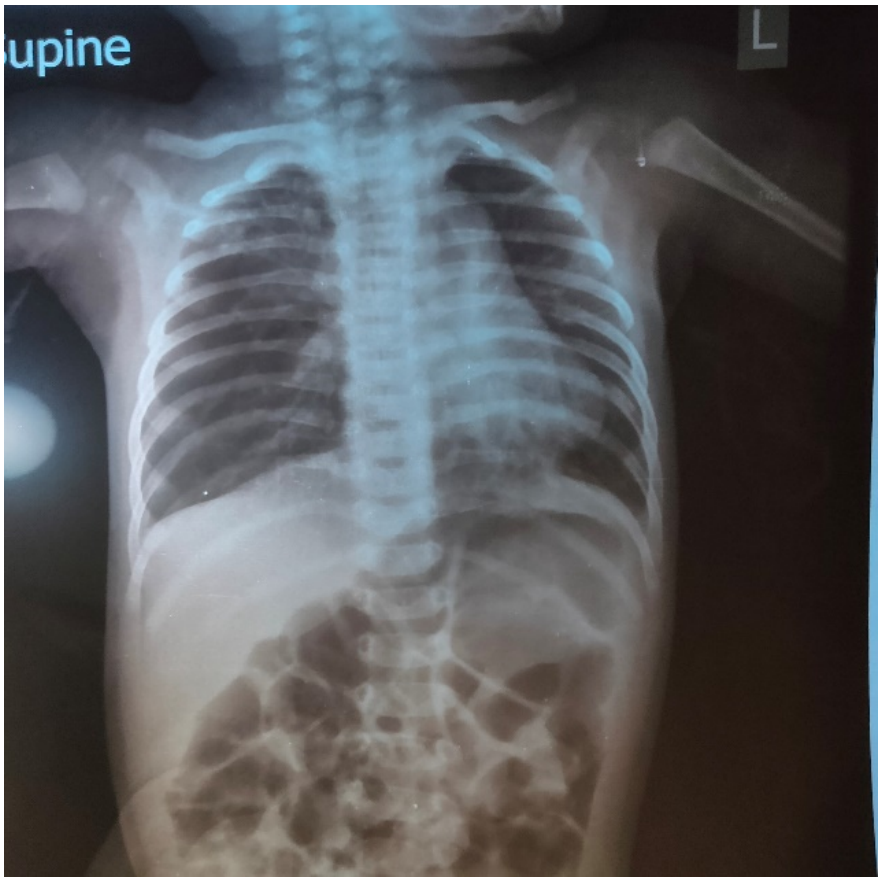


Figure 1 X-ray chest Postero-anterior (PA) view at 2 months of age reported as right upper zone pneumonia.

The child was administered low-flow oxygen and empiric antibiotic therapy. The septic screen and blood culture results were negative. A faint murmur was appreciated at the left parasternal region with a loud S_2. 2D echocardiography revealed a large perimembranous VSD with moderate PDA and severe pulmonary arterialhy pertension (PAH). Furosemide and spironolactone administration was initiated, and it was determined that definitive repair could be planned after the infant exhibited sufficient weight gain. The child was weaned off oxygen and discharged after 7 days.

On regular subsequent follow-ups, however, the child persisted to experience cough and tachypnea, as well as growth failure. Hence, definitive cardiac surgery was performed at the age of 3 months and 7 days.The infant underwent glutaraldehyde-treated pericardial patch closure of the ventricular septal defect along with patent ductus arteriosus ligation. The surgical procedure was uneventful.

The child continued to exhibit cough, tachypnea, occasional retraction, and growth failure after undergoing definitive cardiac surgery. Repeated evaluations ruled out the presence of infection or congestive cardiac failure. Serial echocardiography confirmed the success of the cardiac repair. The child was subsequently lost to follow-up for almost 3 months.

The follow-up was resumed at around 7 months and 15 days of age, with the persistence of the above complaints. The infant was admitted several times to a different healthcare facility. He continued to grow poorly, with a weight of 5.1 kgs. and a length of 62 cms. On repeat X-ray chest evaluations, hyperlucency of the right middle lobe and crowding of both the right upper and lower lobes were noted. (Figure 2)

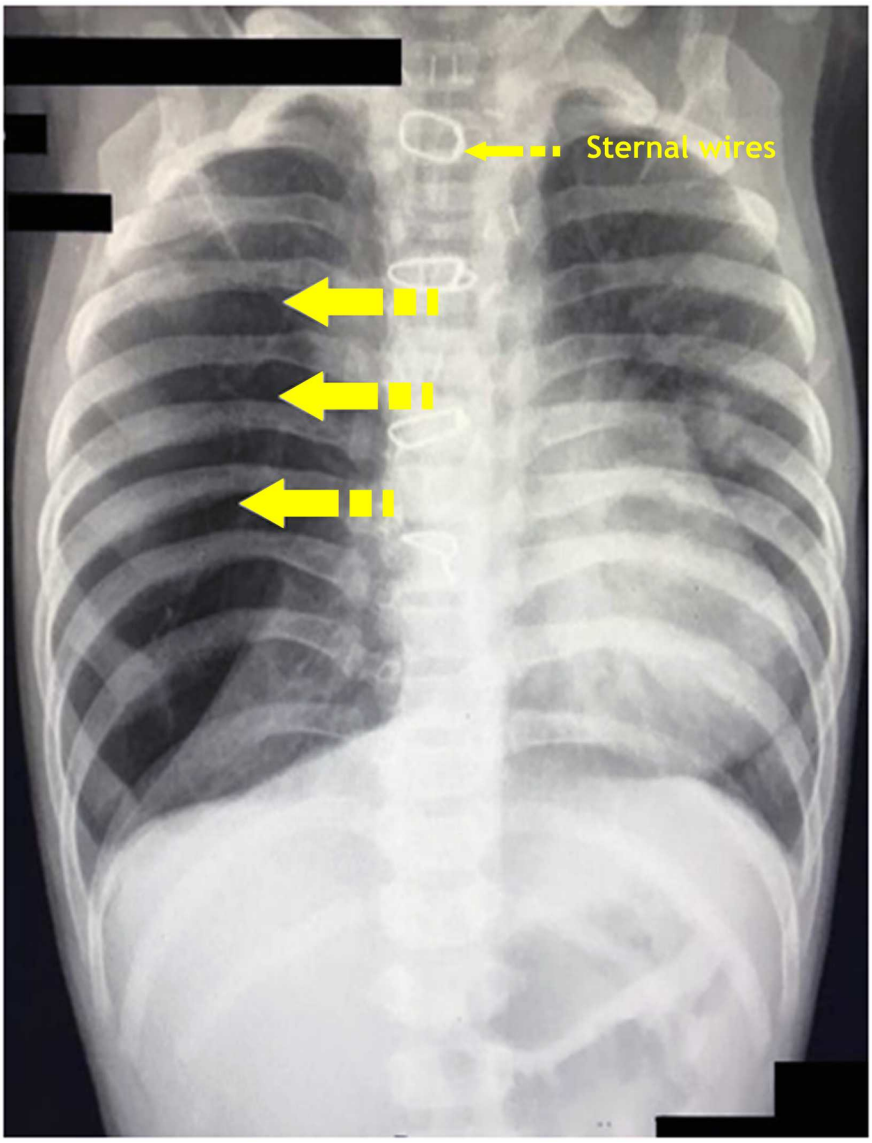


Figure 2 X-ray chest Postero-anterior (PA) view showing clear hyperluceny of the right middle lobe with crowding of right upper and lower lobe with mediastinal shift. Sternal wires from the previous heart surgery can be made out.

CT scan confirmed the presence of Congenital Lobar Emphysema involving the right middle lobe. (Figure 3,4)

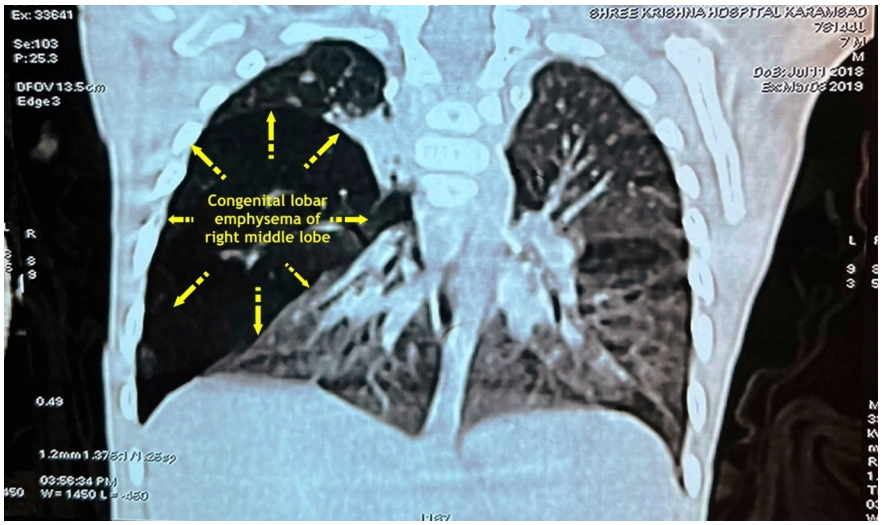


Figure 3 Pre-operative Coronal reformatted High-resolution Computed Tomography (HRCT) scan shows congenital lobar emphysema of right middle lobe.

(Image Credits: Dr. Viral B. Patel)

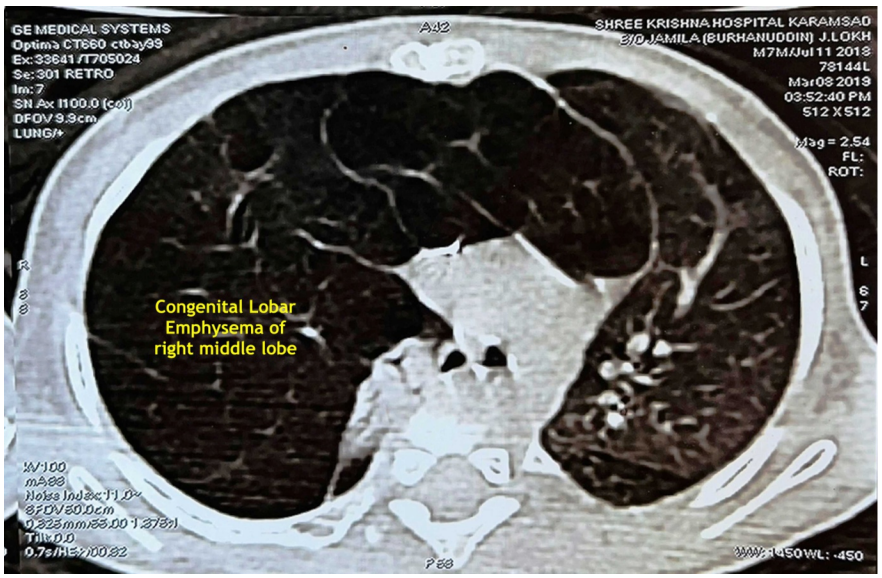
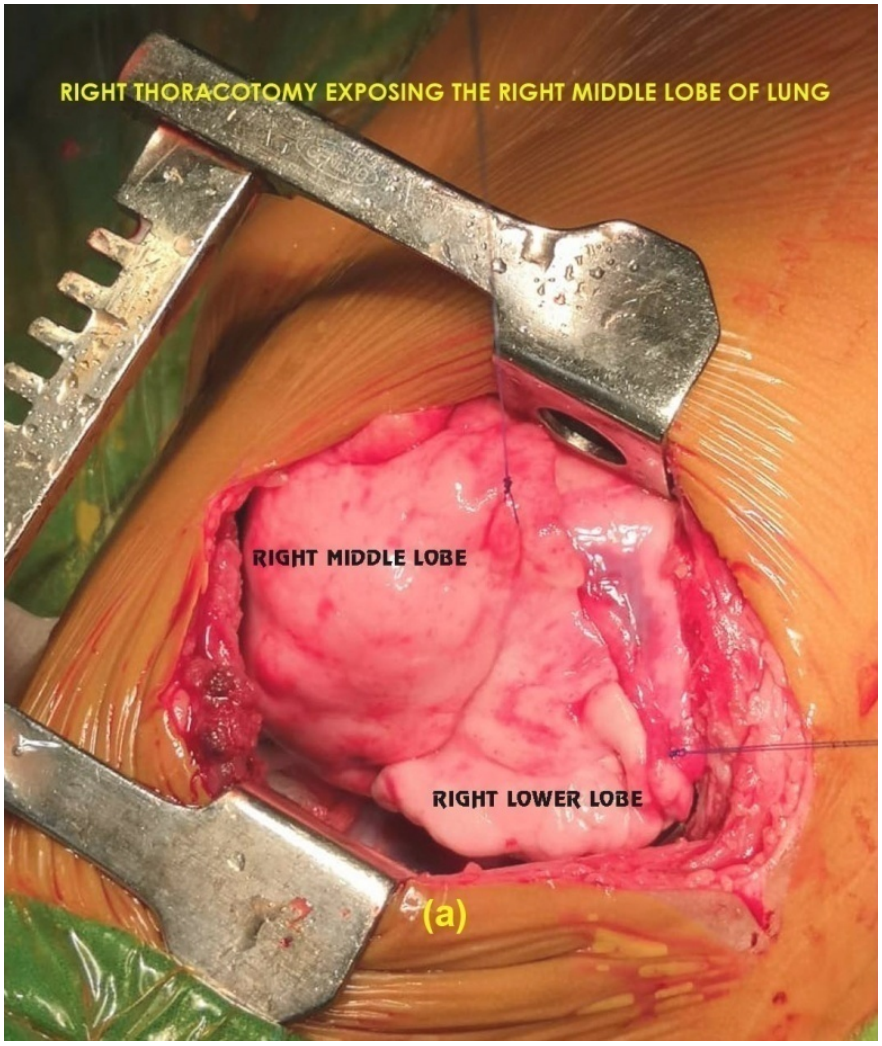


Figure 4 Axial view HRCT lung image conforming the presence of congenital lobar emphysema of right middle lobe.

(Image Credits: Dr. Viral B. Patel)

Right middle lobe resection was performed at 8 months and 9 days of age. The infant was placed in the left lateral position, and the chest cavity was entered through the right 4th intercostal space. Dissection was done using the Reverse Chamberlain Method from the periphery toward the hilum all around the fissure.The right middle lobe vein was identified and ligated proximally, and its branches were tied individually. The right middle lobe artery entering the right middle lobe was identified, and its branches were isolated. Hemostasis was achieved by controlling the brisk nature of bleeding using electro-cautery. A bronchial

clamp was applied after the right middle lobe bronchus was slung by the rubber tubes. The right middle lobe bronchus was divided proximal to the bronchial clamp. Single-lung anesthesia was commenced. The divided bronchial stump was closed with multiple interrupted vertical mattress 5/0 polypropylene sutures. The stump's integrity was checked underwater with sustained ventilation. A minor leak was noted from the lung parenchymal surface, and no leak was noted from the bronchial stump. The right upper lobe and right lower lobe surfaces were sutured together with two interrupted polypropylene pledgetted sutures. The specimen was sent for histopathological examination (Figure 5, a and b),



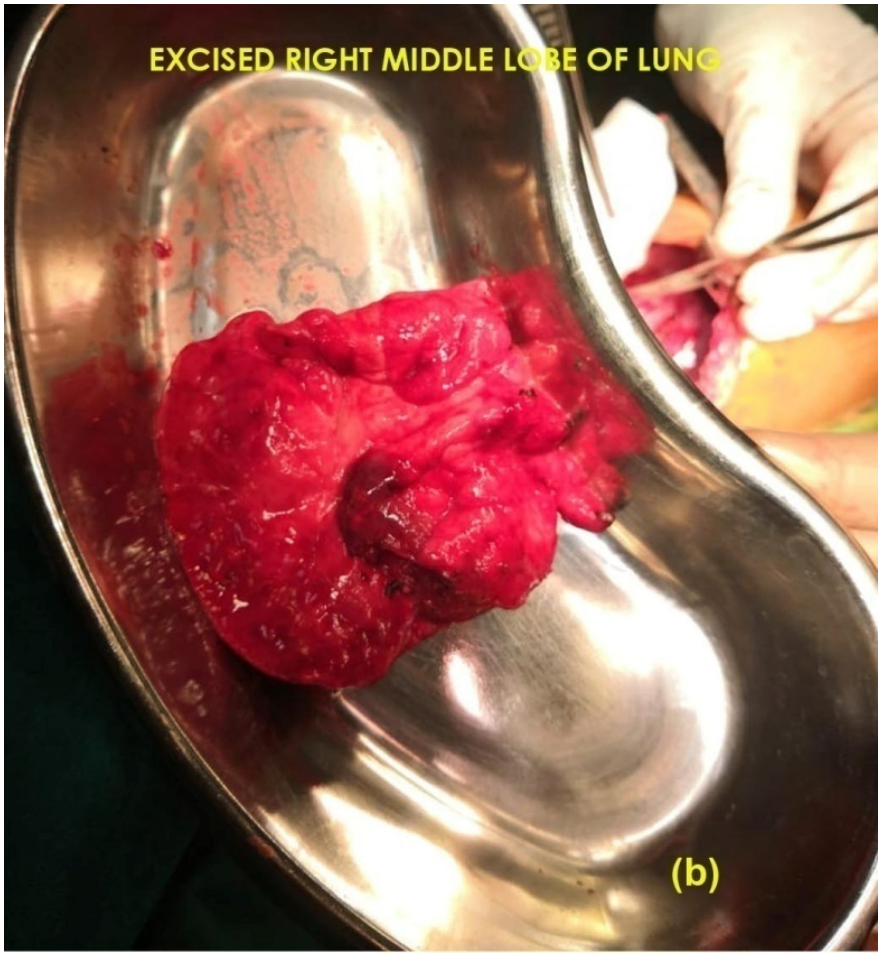


Figure 5 – (a) Intra-operative image showing expanded right middle lobe, (b) Excised right middle lobe of lung

(Image Credits: Dr. Vishal V. Bhende)

which was consistent with the radiological diagnosis.The infant remained asymptomatic,with neither tachypnea nor retraction. X-ray and high-resolution CT scans of the lungs revealed adequate expansion of the right lung, with both the upper and lower lobes expanding effectively and filling the space left by middle lobe resection at 5 years of age (Figures 6,7). The post-operative clinical course was unremarkable.

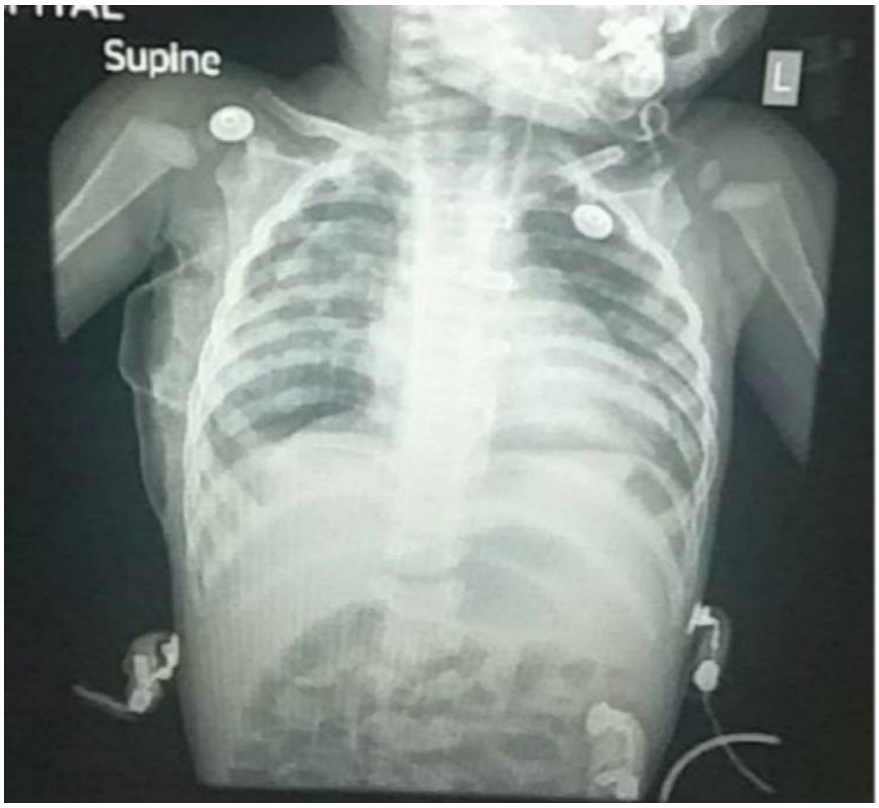


Figure 6 Post-operative X-ray chest showing good expansion of right sided lung with filling of the gap of the right middle lobe resection with right upper and lower lobes in the immediate post-operative period.

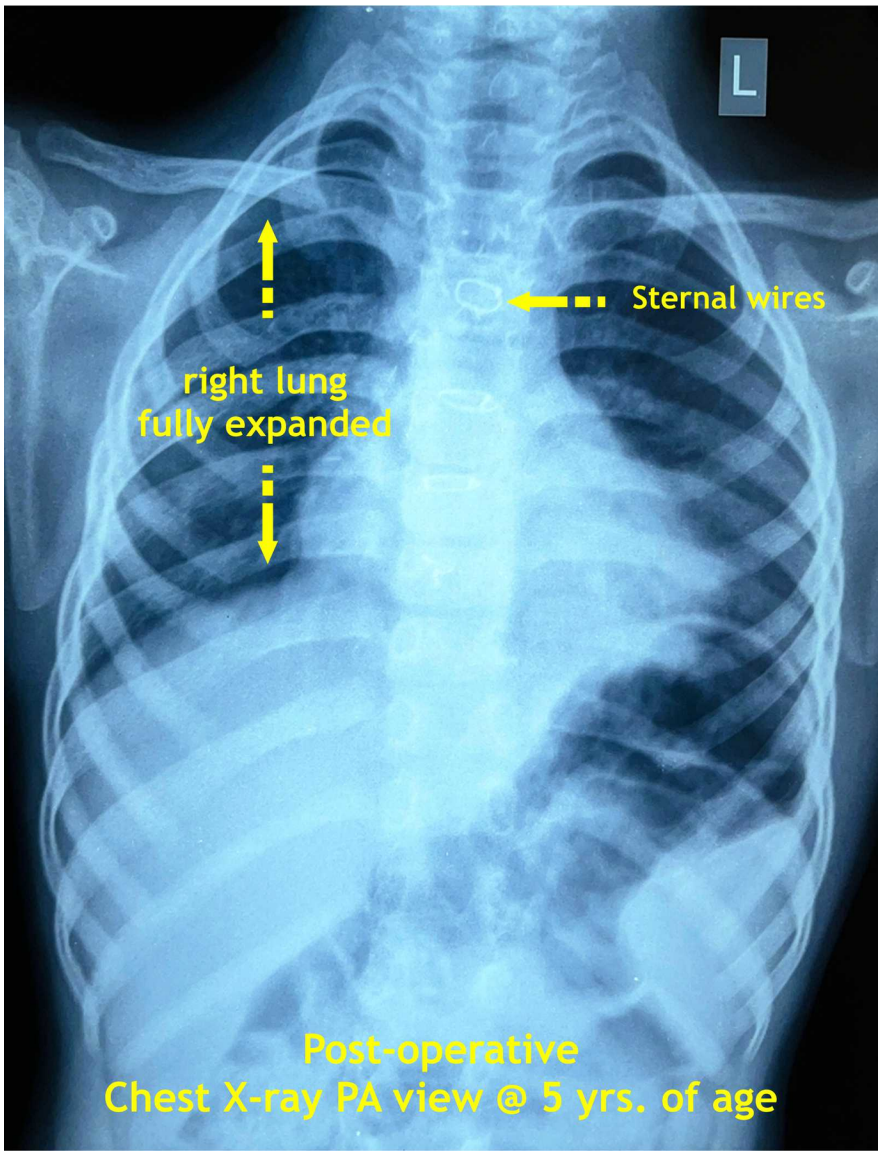


Figure 7 Post-operative X-ray chest showing good expansion of right sided lung with filling of the gap of the right middle lobe resection with right upper and lower lobes demonstrated @ 5 years of age on follow-up.

Weight gain was observed during the subsequent follow-up. high-resolution computed tomography performed on follow-up at 5 years of age showed normally ventilated lungs (Figures 8,9).

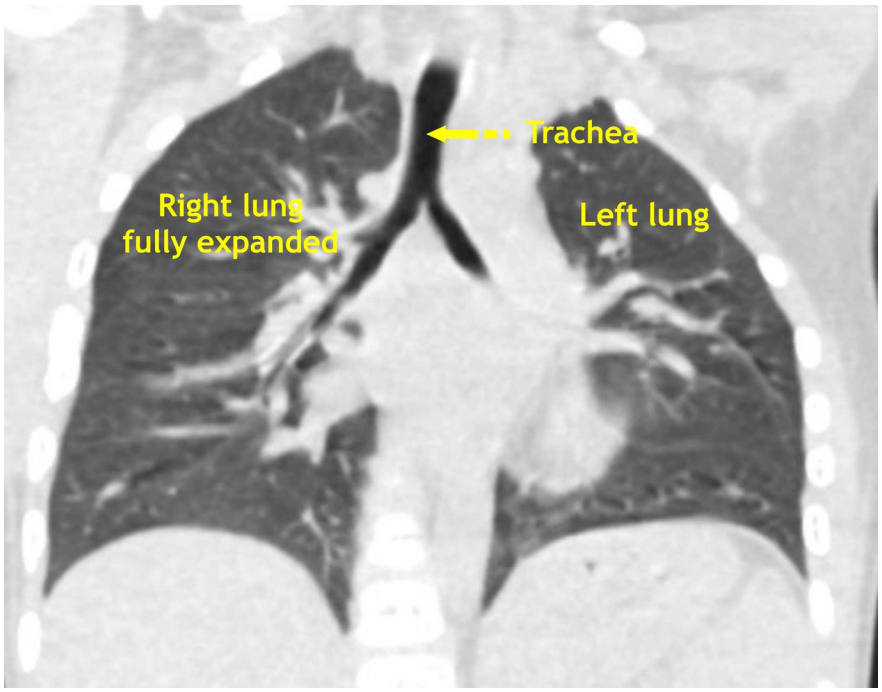


Figure 8 Post-operative Coronal CT Image show normal lung parenchyma

(Image Credits: Dr. Viral B. Patel)

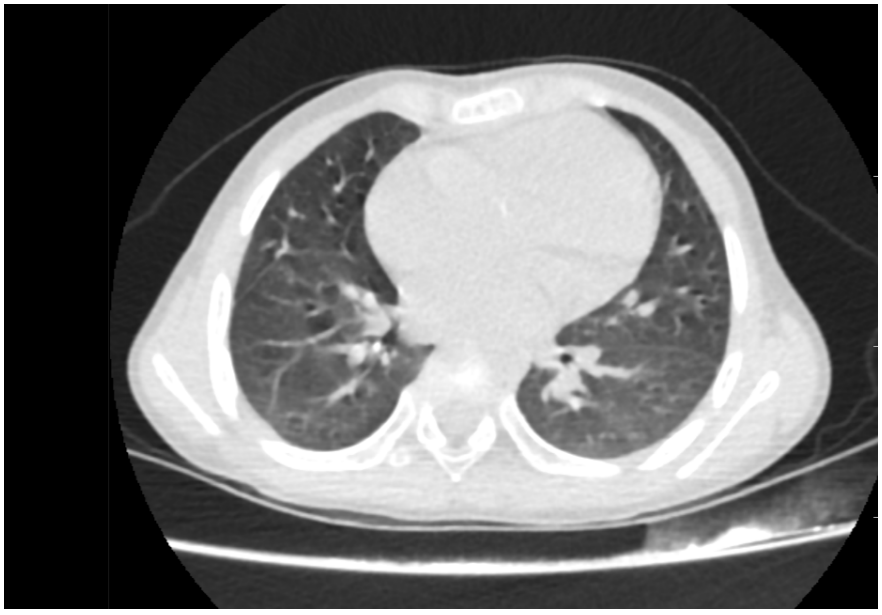


Figure 9 Post-operative HRCT Axial Image showing normally aerated lungs

(Image Credits: Dr. Viral B. Patel)

Discussion

The association between congenital heart defects and CLE is well known, and large left-to-right shunts are commonly associated with CHD. Cough, tachypnea, and respiratory distress in early infancy are the usual clinical presentations in both conditions. As CHD is more prevalent, it is often suspected and diagnosed first. In cases where the entire clinical constellation is attributed to CHD, the presence of CLE is only detected after correction of the CHD. Similar cases have been reported in the past[6,7].

The etiology of CLE remains unknown in almost half of the cases. Absent or dysplastic bronchial cartilage is evident in one-fourth of the cases. Internal obstruction or external compression of the lobar bronchus due to various causes is postulated in the remaining cases. These defects cause a ball valve effect and air trapping during expiration with progressive hyperinflation of the affected lung lobe. The hyperinflated lobe may compress the surrounding normal lobes. In patients with severe disease, the hyperinflated lung lobe can herniate to the surrounding lobes as well as to the opposite thoracic cavity with tracheal and mediastinal shifts. This reduces the respiratory reserve and causes a ventilation/perfusion mismatch with resultant hypoxia[1,3].

In the embryo, the development of the bronchial cartilage occurs during the 4^{th} to 6^{th} intrauterine weeks. This time coincides with the developmental stage of cardiac chamber septation. Thus, a defect during this stage can affect both the heart and lungs[1]. CLE is usually secondary to bronchial compression due to aberrant or dilated pulmonary vessels owing to pulmonary hypertension in the presence of a large left-to-right shunt. In this case, CLE may get resolved with the correction of the underlying CHD[5,6].

CLE continues to pose a diagnostic dilemma. The initial diagnostic modality is usually a chest X-ray. The affected lung lobe is hyperlucent with atelectasis of the adjacent lung lobes due to compression, and a mediastinal shift can be seen. Though all the features are not evident at an early age, X-ray findings are often misreported as pneumonia owing to the crowding of the surrounding lobes, and the hyperlucency of the affected lobe may get reported as a pneumothorax. A CTscan of the lung is confirmatory and delineates anatomical details. Bronchoscopy may aid the diagnosis when an internal bronchial obstruction is suspected[1-4].

Treatment of CLE involves the surgical resection of the affected lobe. Conservative management is proposed for older children with no or minimal symptoms. Infants with persistent distress are ideal candidates for surgery. The surgery is generally well tolerated, with low mortality and favorable outcomes[2,8]. In the case of concomitant CLE and CHD, there are diverging views regarding the defect to be repaired first, or whether a combined lung and cardiac repair is the ideal approach. An individualized approach has been suggested. CLE often gets corrected with the relaxation of the vascular compression, the repair of the large left-to-right shunt, and the reduction of pulmonary hypertension[5,9].

In the present case, the child presented with respiratory distress. During the assessment, clinical suspicion led us to perform 2D echocardiography, which revealed a large VSD with moderate PDA and PAH. The presence of a congenital lung defect was overshadowed by the presence of a large cardiac defect. The symptoms were obviously contributed to the heart defect.The X-ray findings were initially interpreted as being due to pneumonia. As seen in Figure 1 (2 months of age), there is no obvious hyperlucency of the right middle lobe; rather, haziness is evident in the right upper lobe. In contrast, a later X-ray(Figure 2), performed at 7 months of age, clearly demonstrated a large hyperlucent area in the right middle lobe with surrounding lobe crowding and mediastinal shift.In retrospect, we could discern the faint hyperlucency in Figure 1, which was initially missed. We believe that the X-ray chest, being a basic investigation, is often given less attention, particularly when another advanced diagnostic modality has suggested an alternate diagnosis.

CLE is a diagnostic challenge, and the confusion can be further augmented in the presence of an obvious large congenital heart defect. Regular follow-up and a high index of suspicion are necessary for early diagnosis. Lobectomy can lead to dramatic improvement and is generally well tolerated.

Conclusion

Congenital lobar emphysema is a diagnostic challenge, especially in the setting of an obvious large congenital heart defect. Regular follow-up and a high index of suspicion are necessary for early diagnosis. Lobectomy can lead to dramatic improvement and is well tolerated.

Key Clinical Message

Congenital lung anomalies such as congenital lobar emphysema can be missed in young children presenting with respiratory distress in the setting of left-to-right shunts. Advanced imaging such as CT scans are important to confirm the diagnosis.

I am the submitting author and corresponding author. I have provided ORCID IDs of all the co-authors including me.

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