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

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Short running title: Congenital Lobar Emphysema with Congenital Heart Defect

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The data that support the findings of this study are openly available in[Authorea] at, reference number [reference number].

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Abstract

Congenital Lobar Emphysema (CLE) is a rare and life-threatening congenital lung anomaly that often poses a diagnostic dilemma. The problem can be further augmented with co-existent Congenital Heart Defect (CHD) with the left to right shunts. The clinical presentation of congenital heart defects in infantile life is quite similar to the CLE, and CHD being more common gets detected early. The presence of underlying CLE may only be suspected after the repair of the CHD. In our case, a 2-month-old patient presented with respiratory distress, and on evaluation large Ventricular Septal Defect (VSD) and Patent Ductus Arteriosus (PDA) were detected. After the successful repair of the cardiac defects, the infant continued to have cough, respiratory distress, and failure to thrive. Then on radiological re-analysis CLE was suspected on X-ray, and confirmed by Computed Tomography (CT). CLE can get detected on an X-ray chest, but it is often confusing and misleading at an early stage.

Key Words:

Lobectomy, Left to Right shunt, CT scan lung, Right middle lung lobe, Ventricular Septal Defect, Patent Ductus Arteriosus, Respiratory distress, Failure to thrive

Introduction

Congenital Lobar Emphysema (CLE) is a rare and life-threatening anomaly of the lung characterized by hyperinflation of usually one or sometimes more of the lung lobes. It sincidence rate is around 1 in the 20,000 to 30,000 live births with 3:1 male preponderance[1,2]. The hyperinflated lobe compresses the surrounding normal lung tissue causing atelectasis leading to ventilation-perfusion mismatch and hypoxia. It is usually manifested 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 right upper lobe (21%).Congenital Heart Defects (CHD) can present in around 14 – 20% of cases of CLE, and Ventricular Septal Defect (VSD) is the most commonly associated anomaly[1,4]. The clinical presentation of CLE and most CHDs are similar and overlapping in the infantile age, and CHD being far more common gets detected early.Often the diagnosis of CLE may get overlooked in presence of the CHD[5,6]. We encountered a case where the presence of CLE was established after the corrective repair of the large perimembranous VSD with moderate Patent Ductus Arteriosus (PDA).

Case Presentation

A 1 month 24 days old male child presented with cough, tachypnea, and minimal chest retraction for a short duration. The child was afebrile with fair 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 $< 3^{rd}$ centile. His birth weight was 2.6 kgs., suggesting poor weight gain. The child was on mixed feed (breastfeeding + cow's milk) from 1 month of age. The X-ray chest was reported as right upper lobe pneumonia. (Figure 1)

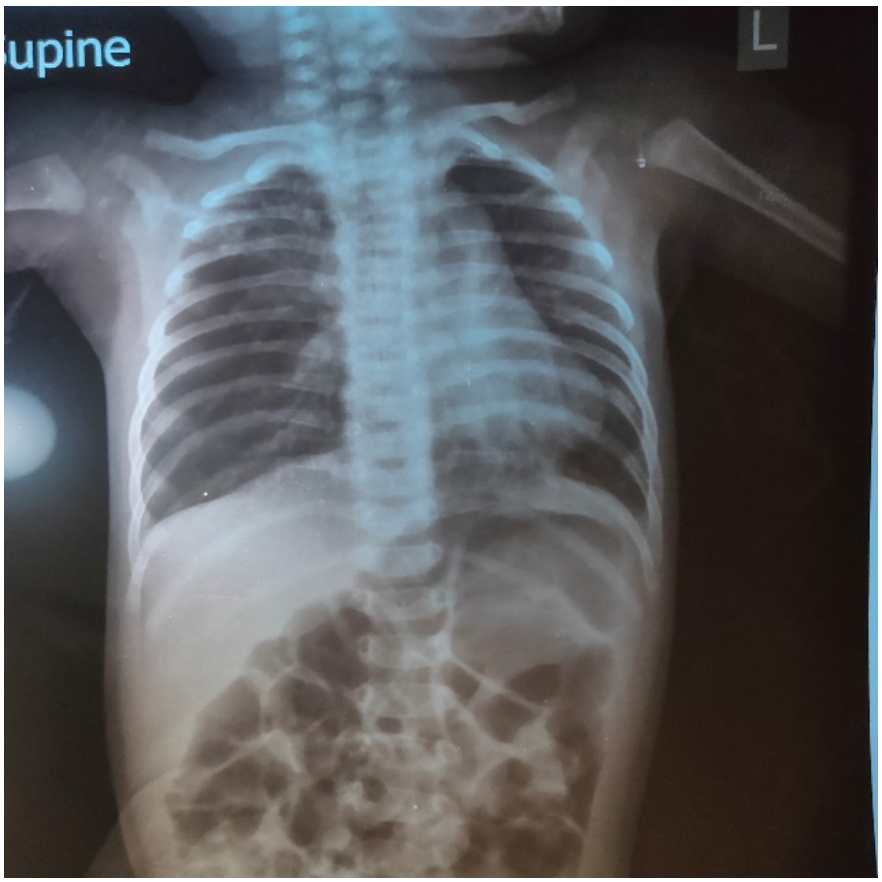


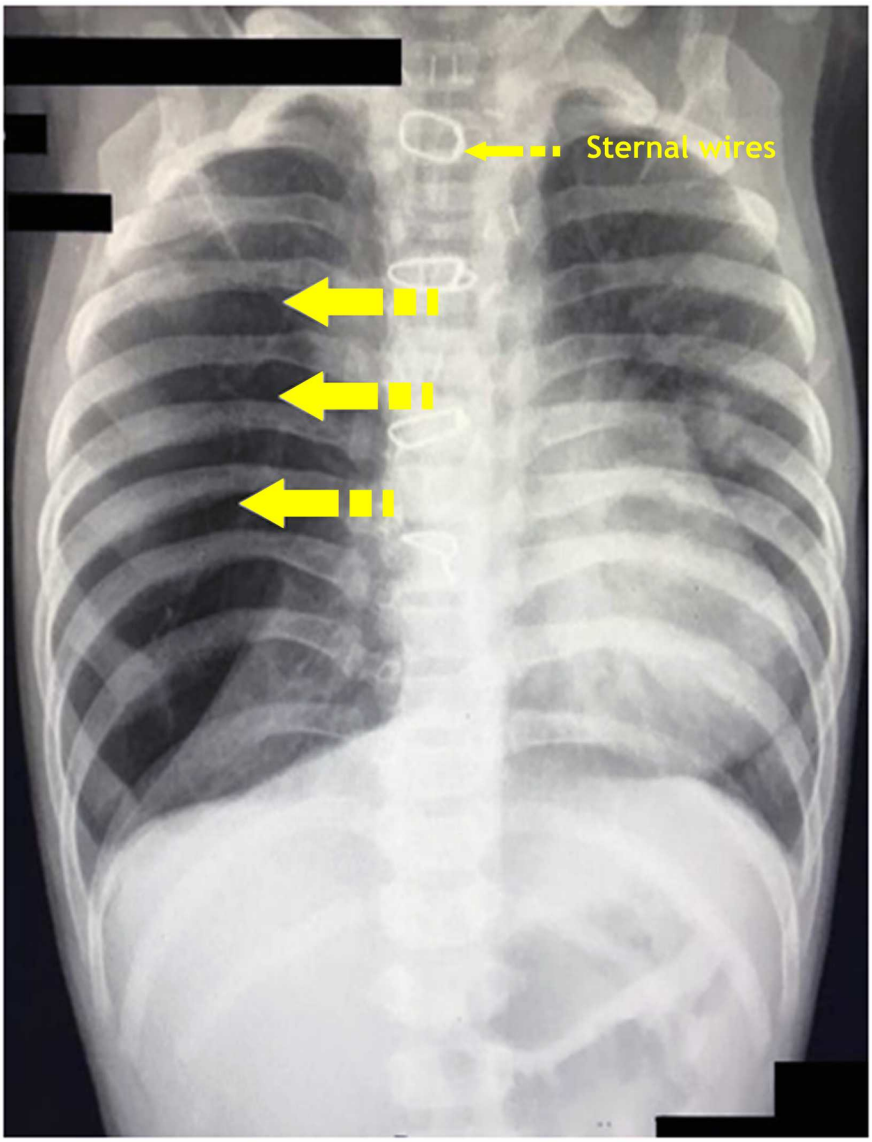
Figure 1 X-ray chest Postero-anterior (PA) view at 2 months of age reported as right upper zone pneumonia.

The child was kept on low-flow oxygen and an empirical antibiotic was started. The septic screen was

unremarkable and the blood culture was sterile. A faint murmur was appreciated at the left parasternal region with loud S2.2D Echocardiography revealed large perimembranous VSD with moderate PDA and severe Pulmonary Artery Hypertension (PAH). Anti-failure medications (Furosemide and spironolactone) were started, and definitive repair was planned after sufficient weight gain. Oxygen was weaned off and child was discharged after 7 days.

On regular subsequent follow-ups, the child persisted to have cough and tachypnea, as well growth failure was also reported. Thus, definite cardiac surgery was performed at 3 months and 7 days of age.The baby underwent Glutaraldehyde treated Pericardial Patch Closure of Ventricular Septal Defect with Patent Ductus Arteriosus Ligation. The surgical procedure was uneventful.

Even after the definitive cardiac surgery, the child continued to have cough, tachypnea, occasional retraction, and growth failure. Repeated evaluations ruled out the presence of infection or congestive cardiac failure (CCF). Repeated Echocardiography affirms the success of the cardiac repair. Then after, the child had lost 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. He had several admissions at another 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 evaluation, hyperlucency of the right middle lobe and crowding of both the right upper and lower lobe was 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)

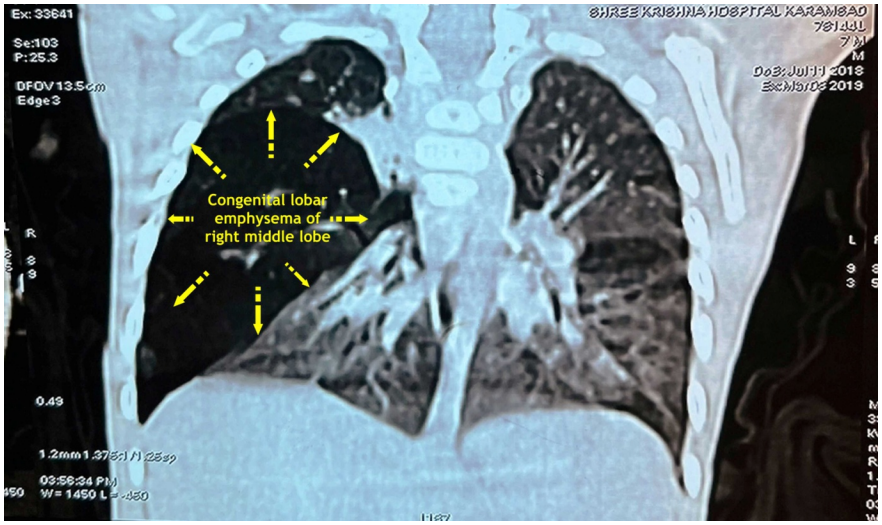


Figure 3 Pre-operative Coronal reformatted High-resolution Computed Tomography (HRCT) scan shows congenital lobar emphysema of right middle lobe.

(Image Credits: Dr. Viral B. Patel)

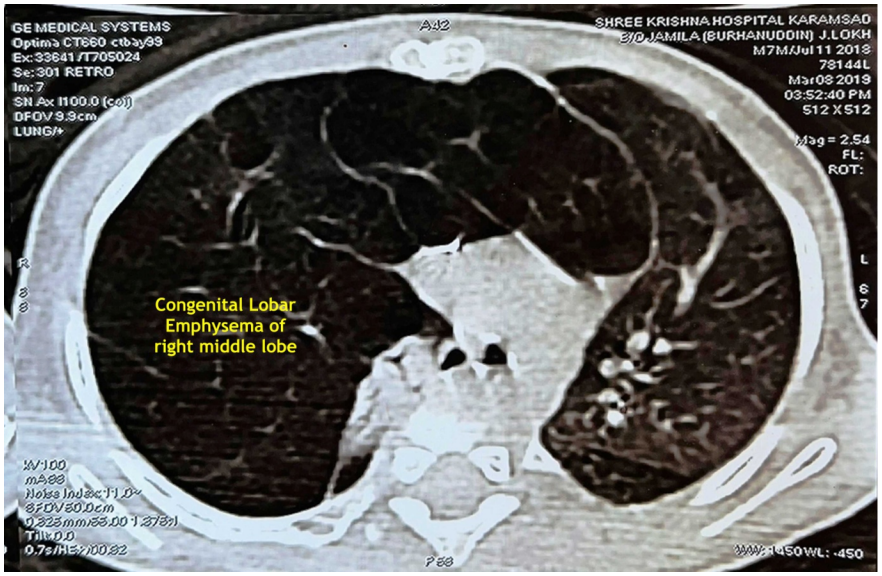
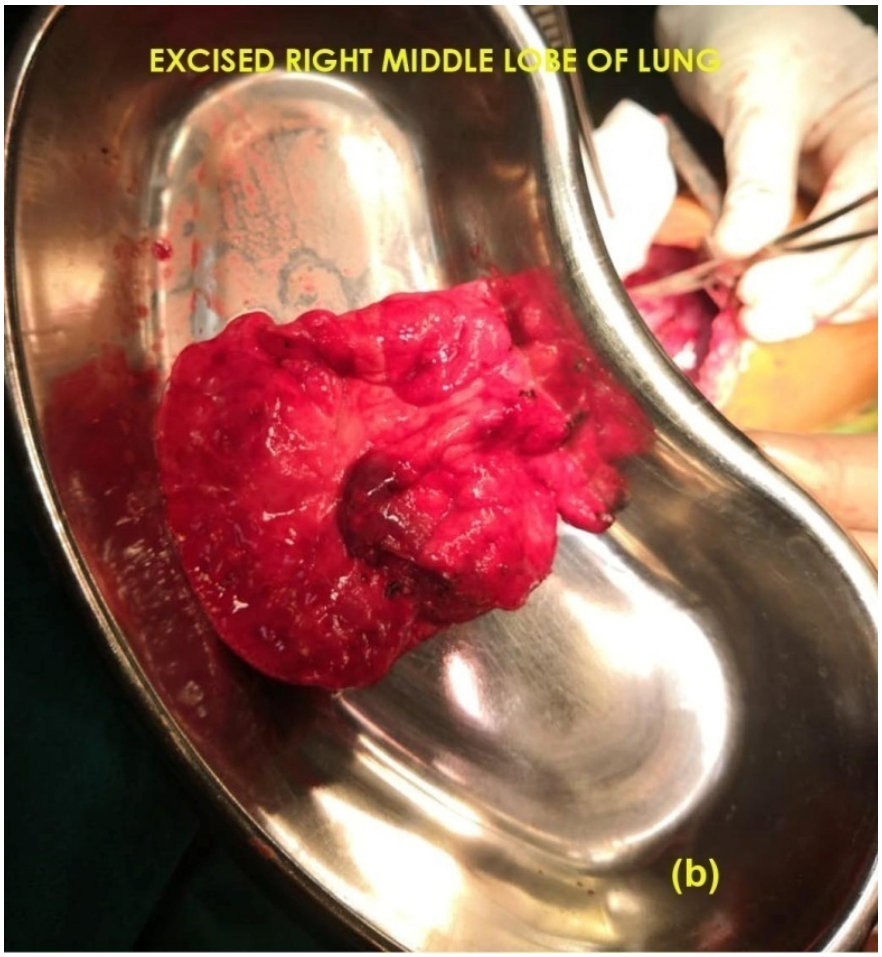
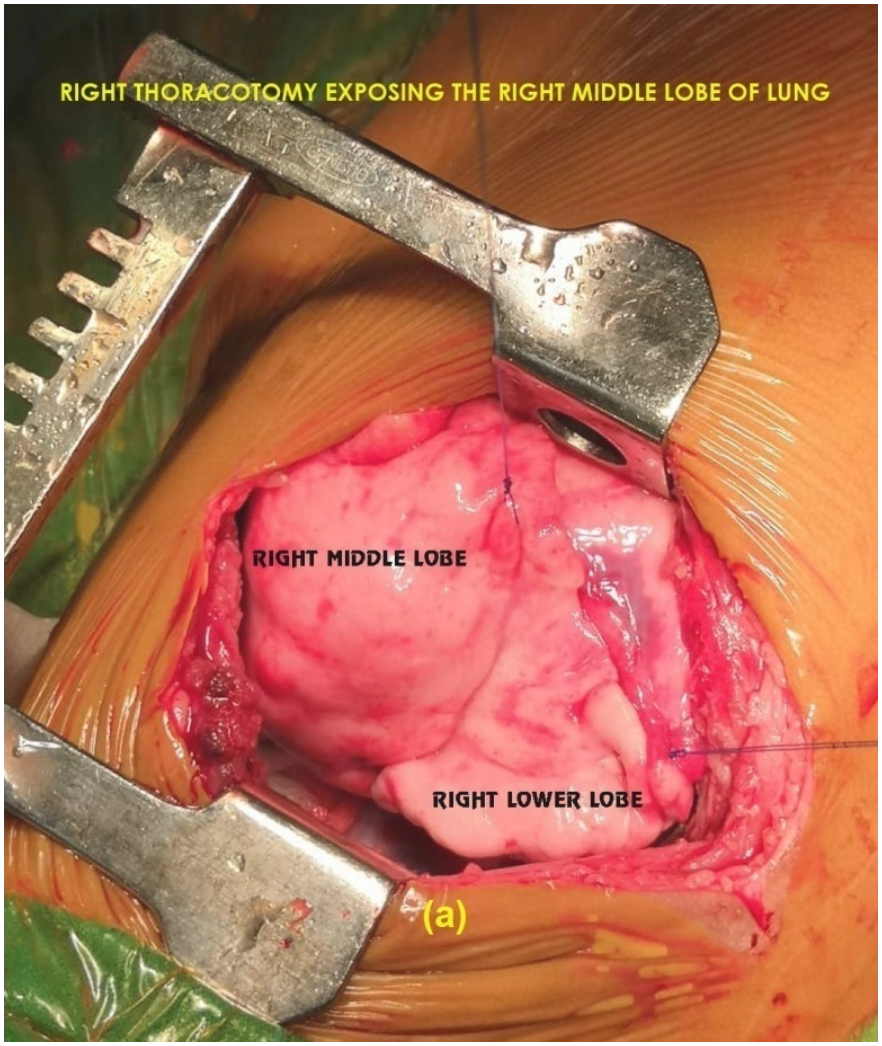


Figure 4 Axial view HRCT lung image conforming the presence of congenital lobar emphysema of right middle lobe.

(Image Credits: Dr. Viral B. Patel)

The lobectomy was planned after weighing the benefit-risk of surgical resection with a thorough literature review. Right middle lobe resection was performed at 8 months and 9 days of age. The baby was placed in the left lateral position, chest cavity entered through the right 4th intercostal space. Dissection was done by Reverse Chamberlain Method from the periphery towards the hilum all around the fissure.The right middle lobe vein was identified and ligated proximally and branches were individually tied. The right middle lobe artery entering the right middle lobe was identified and branches were separated. The brisk nature of bleeding was controlled by hemostasis using electro-cautery. A bronchial clamp was applied after the right middle lobe bronchus was slinged by the rubber tubes. Right middle lobe bronchus divided proximal to the bronchial clamp. Single lung anesthesia was commenced. The divided bronchial stump is closed with multiple interrupted vertical mattress 5/0 polypropylene sutures. Stump integrity is 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 adhered together with two interrupted 5/0





polypropylene pledgetted sutures. The specimen was sent for histopathological examination (Figure 5, a & b)

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

which was consistent with the radiological diagnosis.The clinical course was stable after the surgery. The child recovered fast during the postoperative period. The child remained asymptomatic and the tachypnea and retraction disappeared. X-ray & High-resolution CT lungs showed good expansion of the right sided lung with both the upper and lower lobe expanded well and filling the gap of middle lobe resection at 5 years of age. (Figure 6,7)

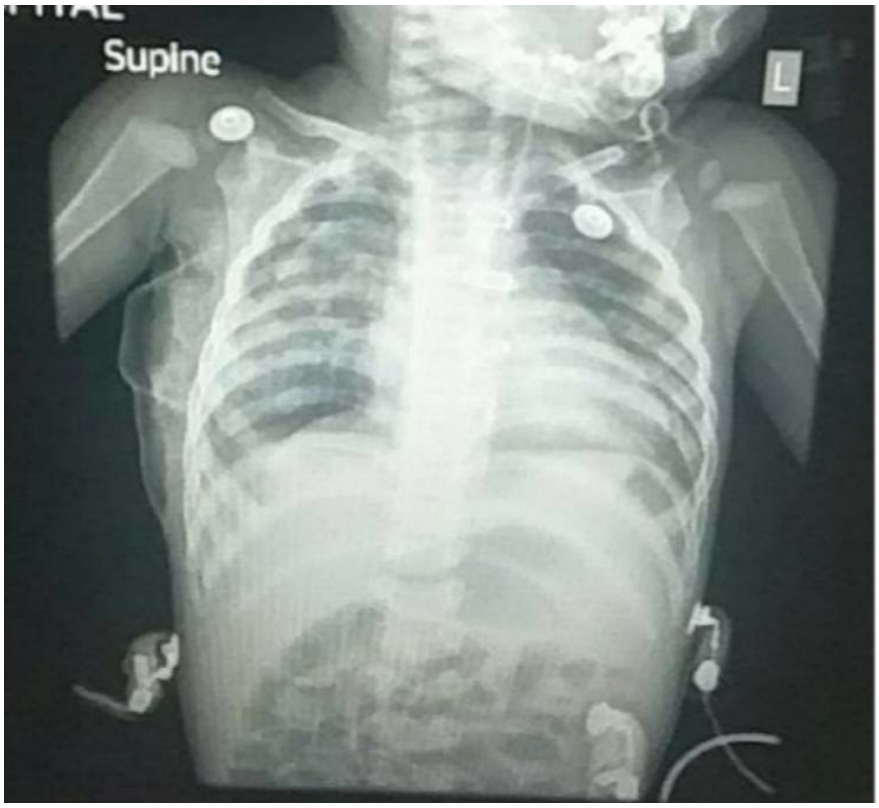


Figure 6 Post-operative X-ray chest showing good expansion of right sided lung with filling of the gap of the right middle lobe resection with right upper and lower lobes in the immediate post-operative period.

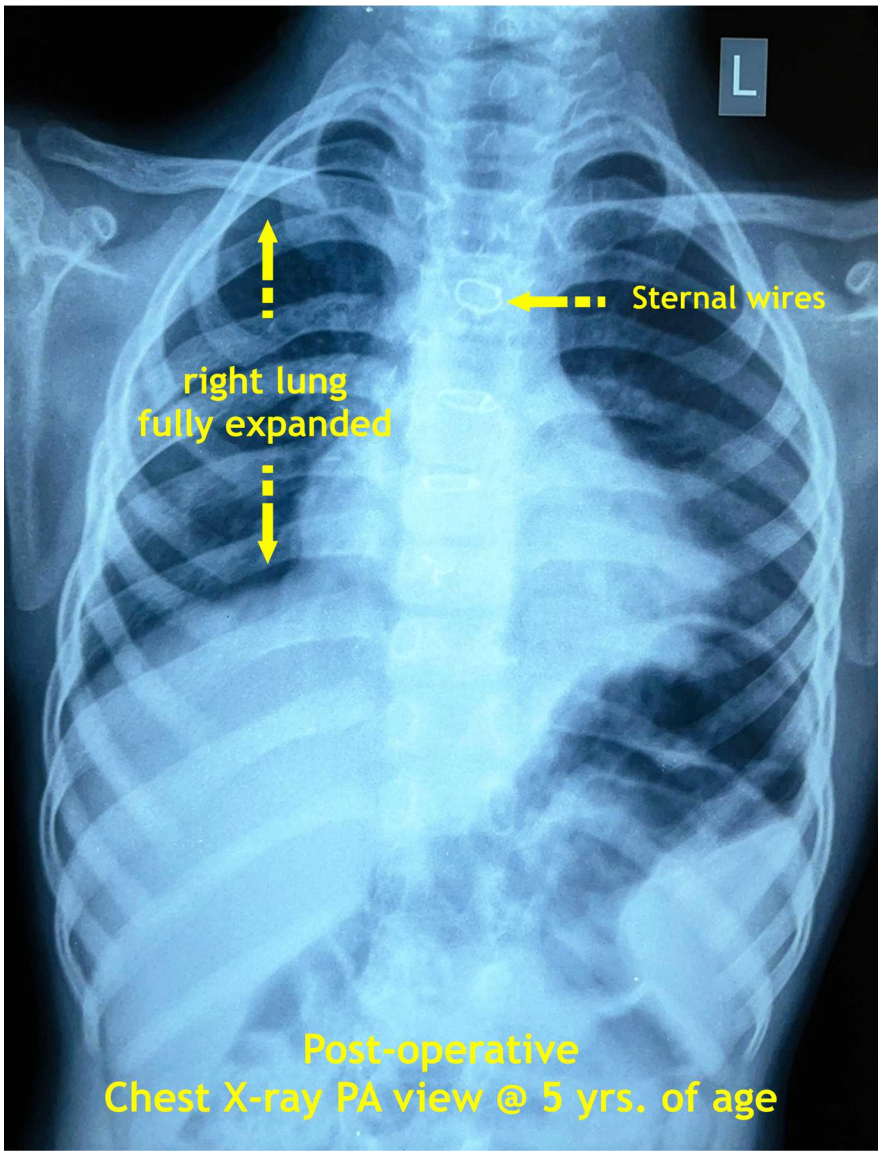


Figure 7 Post-operative X-ray chest showing good expansion of right sided lung with filling of the gap of the right middle lobe resection with right upper and lower lobes demonstrated @ 5 years of age on follow-up.

Weight gain was observed during the subsequent follow-up. HRCT done on follow-up at 5 years of age showed normally aerated lungs (Figure 8,9)

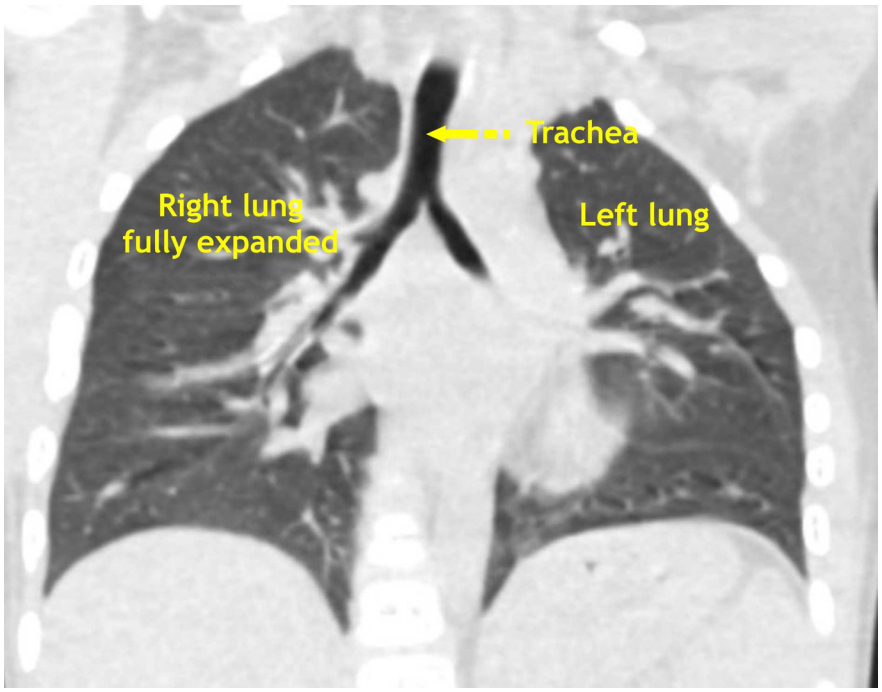


Figure 8 Post-operative Coronal CT Image show normal lung parenchyma

(Image Credits: Dr. Viral B. Patel)

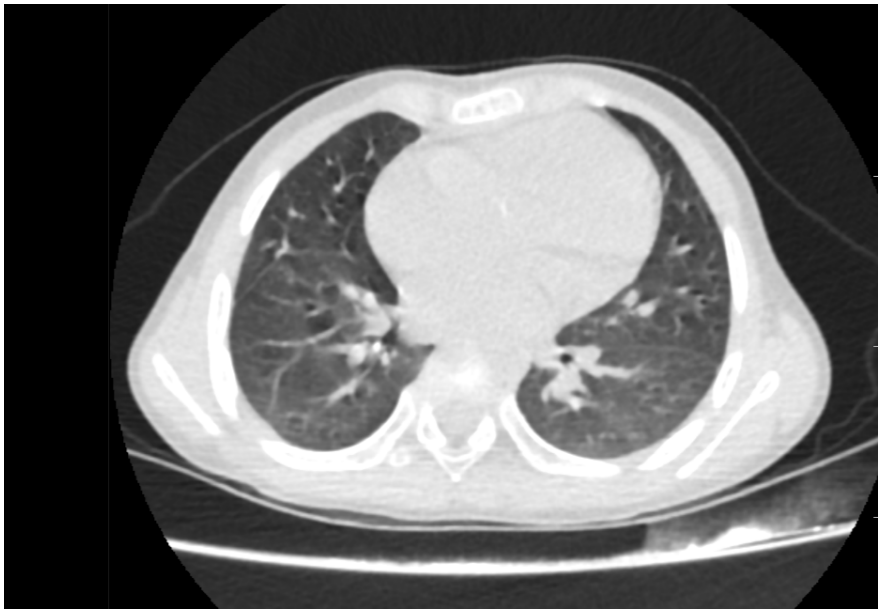


Figure 9 Post-operative HRCT Axial Image showing normally aerated lungs

(Image Credits: Dr. Viral B. Patel)

Discussion

The association of congenital heart defects and congenital lobar emphysema is well known, and the large left to right shunts are commonly associated with CHD. Though, Tetralogy of Fallot (TOF) and other right to left shunts are seen sometimes[6]. Cough, tachypnea, and respiratory distress in early infancy are the usual clinical presentation in both conditions; CLE and left to right shunts. It is quite possible in clinical practice, that CHD being much more commoner is suspected and diagnosed first. Then, the entire clinical constellation is attributed to the CHD and 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 ball valve effect and air trapping during the expiration with progressive hyperinflation of the affected lobe. The hyperinflated lobe may compress the surrounding normal lobes. In extreme cases, the hyperinflated 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 ventilation/perfusion mismatch with resultant hypoxia[1,3].

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

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

Treatment of CLE is the surgical resection of the affected lobe. Conservative management is proposed for older children having no or minimal symptoms.But the infants with persistent distress are the ideal candidates for the surgery. Surgery is by far well tolerated with low mortality and favorable outcomes[2,8]. In the case of concomitant CLE and CHD, there are diverging views in the management as to what to repair first, or a combined lung and cardiac repair would be the ideal approach. An individualized approach has been suggested. Many a time, CLE gets corrected with the relaxation of the vascular compression on the repair of the large left to right shunt and reduction of pulmonary hypertension[5,9].

In our case, the child presented with respiratory distress. During the stay, clinical suspicion leads us to perform 2D Echocardiography which revealed a large VSD with moderate PDA with PAH. The presence of a congenital defect of the lung was overshadowed by the presence of a large heart defect. The symptoms were obviously contributed to the heart defect.The X-ray finding was interpreted as pneumonia initially. We may see in Figure 1 (two months of age) that there is no obvious hyperlucency of the right middle lobe, rather haziness is evident in the right upper lobe. In contrast, later X-ray**,** Figure 2 (seven 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 make out the faint hyperlucency in Figure 1, which was initially missed out.We believe that the X-ray chest being a basic investigation is often given less attention, particularly when the other advent diagnostic modality has suggested alternative diagnosis.

Congenital lobar emphysema is a diagnostic challenge, and the confusion can be further augmented in the presence of an obvious large congenital heart defect. A close look over the chest X-ray may give the lead, but the CT scan is a confirmatory investigation. Regular follow-up and a high index of suspicion are necessary for early diagnosis. Lobectomy can lead to dramatic improvement and is well tolerated.

Conclusion

Congenital lobar emphysema is a diagnostic challenge, and the confusion can be further augmented in the presence of an obvious large congenital heart defect. A close look over the chest X-ray may give the lead, but the CT scan is a confirmatory investigation. 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

A clinician should think beyond pneumonia and left-to-right shunts when young children persisted to have respiratory distress after their correction, and congenital lung anomalies including congenital lobar emphysema can be the possibility. X-ray chest at an early age though not confirmatory but can give a clue and should be assessed carefully, and the CT scan can confirm the diagnosis.

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