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Case Report

Left Pulmonary Agenesis with Congenital Diaphragmatic Hernia: A Rare Congenital Anomaly

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Abstract

Complete absence or severe hypoplasia of one or both lungs is termed as pulmonary agenesis. It is a congenital disorder with a prenatal autopsy incidence of 1 in 15000, though true incidence is still unknown. Unilateral pulmonary involvement is more common than bilateral pulmonary agenesis. The anomaly may affect either lung, with more predilections for the right side. We observed a case of the left lung agenesis with congenital left sided diaphragmatic hernia and to the best of our knowledge very few cases have been reported in the previous literature.

Key words: Diaphragmatic hernia, Lung agenesis, Multidetector CT angiogram

INTRODUCTION

Lung agenesis is an extremely rare congenital anomaly of the lung. Proper knowledge and early diagnosis of the condition are a must to avoid management as effusion, collapse, or consolidation. Contrast-enhanced computed tomography (CECT) is the most effective diagnostic tool to differentiate various mimics and assess airways and vascular structures.

CASE REPORT

An 8-year-old boy from Sitapur presented to Pediatric outpatient clinic of King George Medical University with complaints of recurrent chest infections. He was born to a 36-year-old second gravida mother with non-consanguineous marriage at term gestation with no perinatal complications. Developmental milestones were normal for age.

On physical examination, patient was active, alert with normal anthropometric parameters. He had tachycardia (heart rate-110/min), tachypnea (respiratory rate-60/min) with mild subcostal recession. His chest was normal shaped, with slightly decreased movements on the left side. Trachea was central in position. There was dull percussion note on the whole of the left side of chest. Breath sounds were reduced on the left side. Per abdominally, liver was palpated on the right side, 1 cm below costal margin. On echocardiography cardiologist could not be able to localize pulmonary arteries properly so he suggested CT pulmonary angiography.

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Investigations

A CT chest in pulmonary angio protocol [Figures 1-5] was performed for better evaluation. The CT showed complete opacification of the left hemithorax on the topogram with no mediastinal shift. These findings were confirmed on lung and soft-tissue window. Reconstructed and raw images confirmed complete absence of the left lung. Small left bronchial stump was present with abrupt cutoff. The pulmonary artery (PA) and veins were completely absent on the left side. Left upper abdominal viscera including stomach, spleen, and omentum were herniated into the left to occupy entire left hemithorax space. There was mild compensatory hyperinflation of the right lung. There was also dextroscoliosis with segmentation anomaly of the upper dorsal vertebrae. These findings confirmed a diagnosis of the left pulmonary agenesis type I with the left-sided diaphragmatic hernia.

Treatment

No treatment is required in asymptomatic cases.[1] Treatment is necessary for lower respiratory tract infections.

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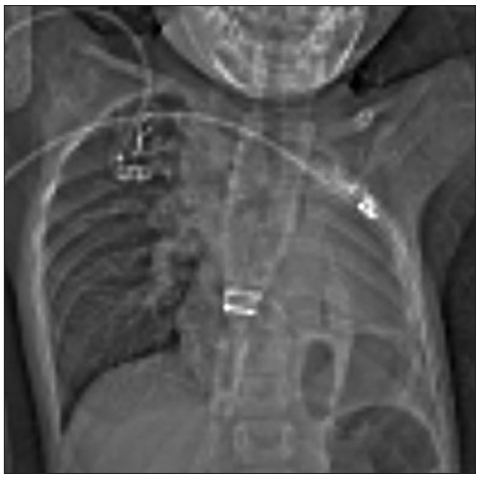

Figure 1: Computed tomography scanogram of the chest showing complete opacification of the left hemithorax with cranial migration of abdominal contents into the thorax the left airway is not present

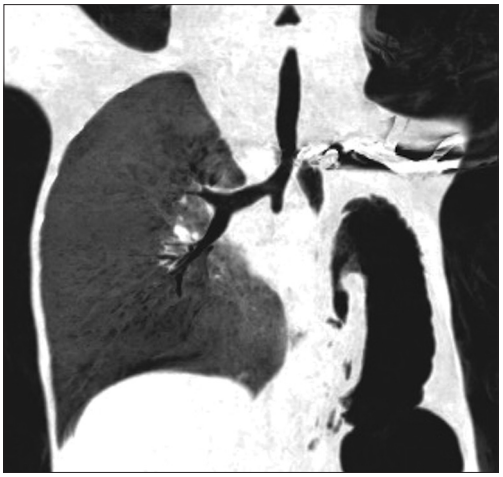

Figure 2: Coronal MnIP image - shows short left bronchus stump with complete cut-off and absent left lung, trachea, and Rt bronchus normal

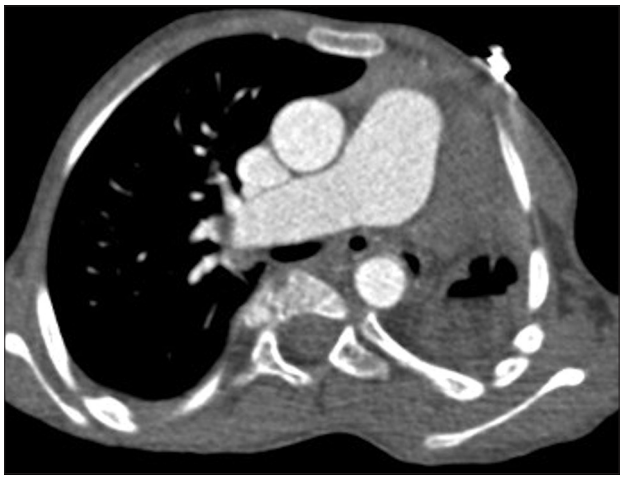

Figure 3: Axial thin images of computed tomography chest at the level of the pulmonary artery (PA) and heart show absent left PA with dilated right and main PA

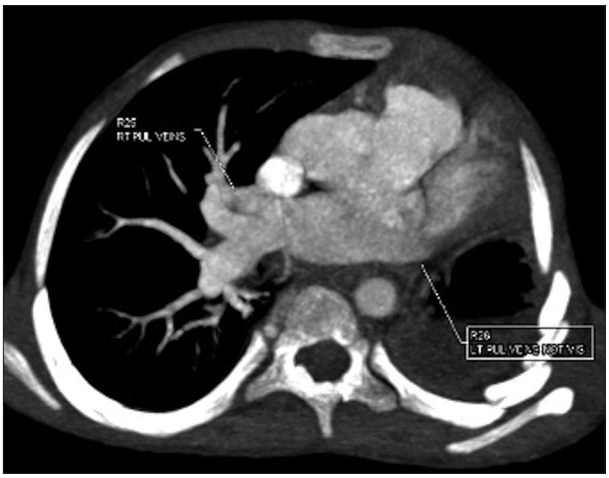

Figure 4: The axial image at the level of the heart shows cardiac chambers and absent left pulmonary veins. The left lung is absent with abdominal content of the left side herniated into the thorax

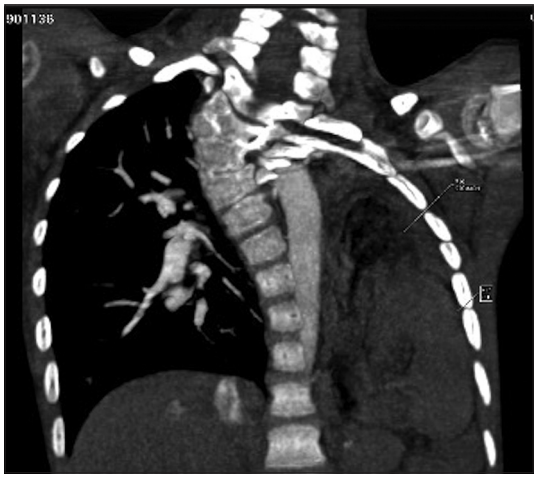

Figure 5: Coronal thin image of computed tomography chest posterior to heart shows absent left lung. The stomach and spleen are occupying the space in the left hemithorax. There is also dextroscoliosis

Patients having stumps may require surgical removal of the stump if postural drainage and antibiotics fail to resolve the infection.[2] Corrective surgery of associated congenital anomalies, wherever feasible, may be undertaken.[3]

Outcome and Follow-up

Prognosis depends on two factors. First, the severity of associated congenital anomalies and second, involvement of the normal lung in any disease process.[3] Patients with the right lung agenesis have a higher mortality than those with the left lung agenesis because of compression of the tracheobronchial tree by the shifting of normally midthoracic structures into the right chest.[4] If patient survives the first 5 years without major infection, an almost normal life span can be expected.[5] The patient was managed conservatively. The clinical symptoms were improved and the right lung was normal with no other associated congenital anomalies;

therefore, no surgical intervention was performed. The patient was followed at hospital out-patient department for a year and was clinically well.

DISCUSSION

Pulmonary agenesis and aplasia are rare abnormalities with reported incidence between 0.0034% and 0.0097%.[6] Many cases of pulmonary agenesis, aplasia, and hypoplasia have been reported at different ages including prenatally in new-born, infants, children, adults, and even at 90 years of age.[4,7-9] Bilateral pulmonary agenesis is a rare congenital anomaly that may occur in anencephalic babies.[1] Unilateral agenesis, aplasia, and hypoplasia are comparatively more common. These may have fewer symptoms and non-specific findings, that is, why only one-third cases are diagnosed during the lifetime. Functionally, unilateral agenesis and aplasia are similar. The sole lung is larger than normal, and this enlargement is true hypertrophy and not emphysema.[1]

Failure of development from the foregut can lead to these types of congenital pulmonary malformations. Bilateral pulmonary agenesis is caused by developmental arrest at the stage of the primitive lung bud. The respiratory anlage at a later stage may develop only unilaterally and leads to unilateral lung agenesis. Lobar agenesis results when developmental arrest on one side occurs in an older embryo; however, pulmonary hypoplasia may occur during the last trimester of pregnancy with failure of final alveolar differentiation.[1]

Genetic, teratogenic, and mechanical factors may have a bearing on etiology.[4,10] These are generally sporadic, with nearly similar occurrence in both the sexes and involve both lungs equally.[4,7] Only few cases are reported in siblings in an autosomal recessive pattern.[1] There is high incidence (>50.0%) of associated cardiac, gastrointestinal, genitourinary, skeletal, central nervous system malformations, and VACTERL sequence.[1,4,10,11]

Schneider and Schawatbe initially classified pulmonary agenesis which was later modified by Boyden into three types according to stages of development of lung bud:[12]

1. Type I: Absence of lung parenchyma, bronchus, and blood supply to affected side (Agenesis).
2. Type II: Absence of lung parenchyma with the presence of rudimentary bronchus only (Aplasia).
3. Type III: Variable amount of lung parenchyma, bronchial tree, and vasculature (Hypoplasia).

As this anomaly can occur at any age, the possibility of lung agenesis should be in differential diagnosis of patients having decrease to absent breath sounds with less or no movement of unilateral chest wall and opaque hemithorax in plain film. For confirmation, diagnostic imaging such as chest computed tomography scan, magnetic resource imaging, bronchoscopy, and chest angiography can be done. The early detection of the pulmonary agenesis is essential to reduce the development of fibrosis in patient's unilateral lung which can occur as result of recurrent chest infection. The surgical procedures should also be in consideration in the presence of other congenital anomalies or complications.[13]

LEARNING POINTS/TAKE HOME MESSAGES/CONCLUSION

Lung agenesis is an extremely rare congenital broncho-pulmonary anomaly.

- CECT is the most effective diagnostic tool to differentiate various radiographic mimics (like effusion, collapse, or consolidation) and assess airways and vascular structures.
- Proper knowledge and early diagnosis of the condition are a must for early diagnosis and timely definitive management.

REFERENCES

1. Krummel TM. Congenital malformations in the lower respiratory tract. In: Chernick V, Boat TF, Kendig EL, editors. Kendig's Disorders of the Respiratory Tract in Children. 6th ed. Philadelphia, PA: WB Saunders Company; 1998. p. 307-8.
2. Rajshekhar B, Gomber S, Krishna A. Pulmonary agenesis. Indian Pediatr 1997;34:534-8.
3. Chopra K, Sethi GR, Kumar A, Kapoor R, Saha MM, Mital M, *et al*. Pulmonary agenesis. Indian Pediatr 1988;25:678-82.
4. Maltz DL, Nadas AS. Agenesis of the lung. Presentation of eight new cases and review of literature. Pediatrics 1968;2:175-88.
5. Sbokos CG, McMillan IK. Agenesis of the lung. Br J Dis Chest 1977;71:183-97.
6. Mardini MK, Nyhan WL. Agenesis of lung. Report of four patients with unusual anomalies. Chest 1985;87:52.
7. Lee P, Westra S, Baba T, McCauley R. Pediatr Radiol 2006;36:449-52.
8. Holstein A, Weber M. An extraordinary finding-accidental diagnosis of complete pulmonary aplasia in a 90 year old lady. Age Aging 2009;38:487.
9. Gabarre JA, Izquierdo AG, Ponferrada MR, Gallardo CO, Agueda JM, Perez PF. Isolateral unilateral pulmonary agenesis: Early prenatal diagnosis and long-term follow-up. J Ultrasound Med 2005;24:865-8.
10. Finda JD, Michelson PH. Congenital disorders of the lung. In: Behrman RE, Kliegman RM, Jenson HB, editors. Nelson Textbook of Pediatrics. 18th ed. Philadelphia, PA: Saunders; 2008. p. 1783.
11. Fitoz S, Ucar T, Erden A, Gunlemez A. DiGeorge syndrome associated with left lung aplasia. Br J Radiol 2001;74:764-6.
12. Ta RK, Banerjee SN, Chatterjee K. Agenesis of left lung in an adult female: A rare case report. Muller J Med Sci Res 2015;6:172-4.
13. Sadiqi J, Hamidi H. CT features of lung agenesis-a case series (6 cases). BMC Med Imaging 2018;18:37.

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