

Challenges in Diagnosis and Management: A Case Series of Paediatric Lung Aplasia and Agenesis

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ABSTRACT

Lung agenesis and aplasia are uncommon congenital disorders that often manifest with breathing difficulty immediately after birth. The outlook depends on which lung is impacted and whether there are accompanying anomalies. Early detection is largely reliant on prenatal imaging. In this case series, we describe three patients—two with right lung agenesis and one with an underdeveloped left lung—detailing their clinical features, disease course, and outcomes.

Key Words: Breathing difficulty, Congenital abnormality, Lung aplasia, Unilateral lung agenesis.

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INTRODUCTION

Lung agenesis is a rare congenital malformation characterised by the complete absence of the lung parenchyma, bronchial tree, and associated vasculature on one or both sides of the thorax. This condition is classified as either unilateral or bilateral. Bilateral lung agenesis is incompatible, whereas unilateral agenesis may be compatible with life depending on compensatory mechanisms and the presence of associated anomalies. Right-sided agenesis typically carries a worse prognosis because the right lung normally provides a greater proportion of respiratory function.¹ The reported incidence of pulmonary agenesis ranges from 1 to 3.1 per 100,000 live births.² In unilateral cases, the contralateral lung often undergoes compensatory hyperplasia to adapt functionally, and in rare situations, affected individuals may remain asymptomatic well into adulthood.³ Some cases are identified prenatally through ultrasound or foetal magnetic resonance imaging (MRI), while others present postnatally with respiratory distress or failure.

CASE 1:

A term neonate was reported to the Neonatal Intensive Care Unit (NICU) of a local health facility with fever and respiratory distress. He received intravenous antibiotics but did not respond and was referred further. Chest x-ray (Figure 1) displayed opacity in the right lung region.

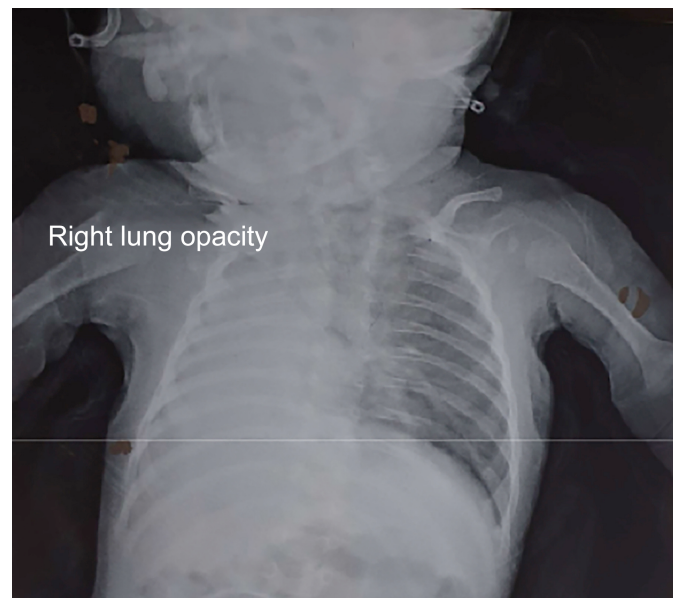


Figure 1: Right-sided lung opacity, cardiac shadow shifted to the right, only the left lung visible.

The NICU course was complicated by marked respiratory distress needing invasive ventilation initially, followed by non-invasive positive pressure ventilation, high flow nasal cannula and low flow oxygen. Upon stabilisation, he had a CT scan of the chest (Figure 2), which demonstrated a notable right-sided cardio-mediastinal shift with hyperinflation of the left lung herniating into the right hemithorax. Trachea and left bronchus were appreciated; however, right bronchus could not be traced. Findings indicated right lung aplasia. Bronchoscopy displayed difficulty in visualising the right main bronchus and no tracheal rings or fistula. His 2D echocardiography demonstrated an atrial septal defect (ASD). He was discharged home after five days of hospital stay. The parents were counselled to ensure good lung health and follow up with a cardiologist and a pulmonologist.

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Figure 2: Right-sided cardio-mediastinal shift and hyperinflation of the left lung.

CASE 2:

A four and a half month baby boy was reported to the local hospital with cough and low-grade fever. Past history revealed a term delivery and two past episodes of similar illness, and the patient was treated for pneumonia. He was prescribed oral antibiotics for seven days on his last visit, along with a chest x-ray (Figure 3), which displayed right-sided opacity. Follow-up, a week later, showed the same very high opacity, and he was admitted. At this time, chest CT scan (Figure 4) was ordered, and it displayed herniation of the left lung to the right with obvious mediastinal shift and the right main bronchus was not visualised. Bronchoscopy displayed a membrane-like structure covering the right main bronchus. 2D echocardiography displayed a ventricular septal defect (VSD), and CT angiography revealed right pulmonary artery agenesis. Comprehensive counselling of the parents was done, but his condition deteriorated, and he needed mechanical ventilation. He remained on ventilatory support for four days and finally died of respiratory failure.

CASE 3:

A 5-month baby boy was referred from a primary care hospital with complaints of cough and breathing difficulty. He was in severe respiratory distress with acidotic breathing and had air entry on the right side only. His chest x-ray (Figure 5) displayed homogenous opacity on the left side of the lung. His CT scan of the chest after initial stabilisation displayed reduced left lung volume with a smaller bronchus, while heart and mediastinum were pulled towards the left side.

Bronchoscopy displayed narrowing of the left bronchus. No heart defect was seen on 2D echocardiography, and parents were counselled in detail regarding the illness.

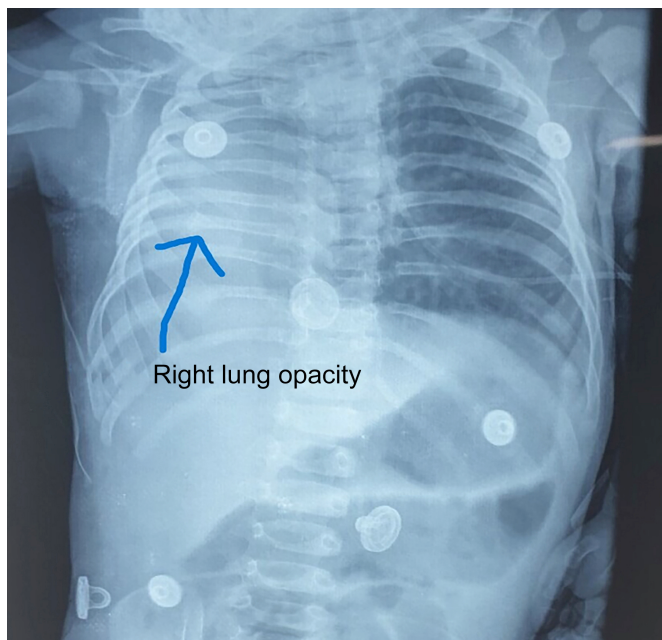


Figure 3: Right sided lung opacity with left lung visible only.

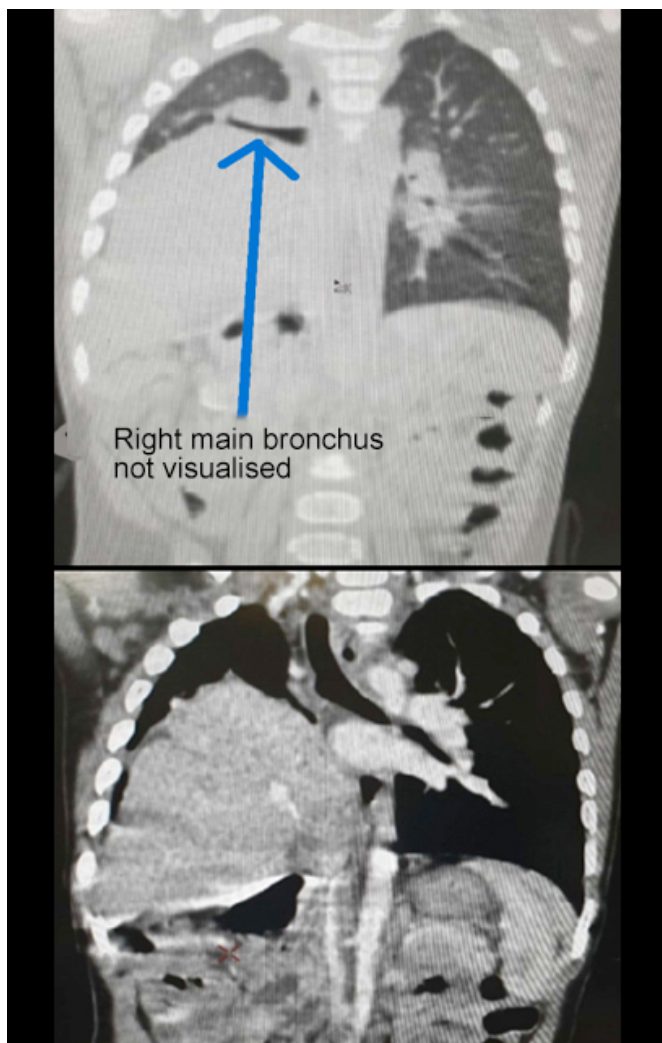


Figure 4: Failure to visualise right main bronchus due to the right lung aplasia.

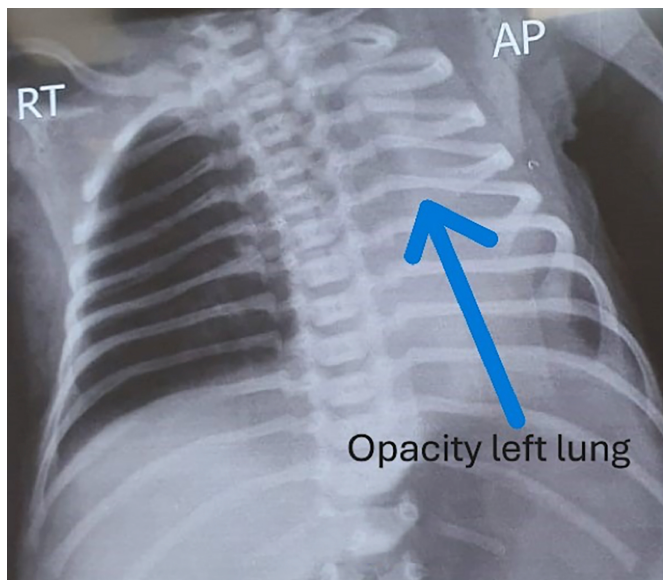


Figure 5: Left-sided lung opacity and hyperinflation of the right lung.

DISCUSSION

The first documented case was reported by De Pozze in 1673 when he accidentally found pulmonary agenesis in a female cadaver. The condition has an incidence of 1 in 15,000 autopsies. The classification system initially introduced by Schwalbe *et al.*⁴ in 1912 and later updated by Boyden⁵ in 1955, outlines three forms of pulmonary underdevelopment; pulmonary agenesis: A condition marked by a complete absence of the lung, including the bronchus and pulmonary vessels. Pulmonary aplasia: In this type, a rudimentary bronchus is present, but no functional lung tissue is developed. Pulmonary hypoplasia: This refers to an incompletely developed lung that retains both a bronchial structure and some degree of pulmonary vasculature. Bilateral pulmonary agenesis, incompatible with life outside the uterus, was first described by a paediatric pathologist, Battista Morgagni.⁶

Clinical presentation varies and may include respiratory distress, recurrent infections, or chest asymmetry. Aetiologies include embryological defects or in utero-vascular events. Diagnosis relies on imaging modalities such as x-ray, CT, or MRI. Early diagnosis and multidisciplinary management are key to improved outcomes.

Advancements in imaging technology have made prenatal diagnosis more feasible. Baiyyam *et al.* described how high-resolution prenatal ultrasonography is able to detect antenatal lung malformations.⁷ Although ultrasound is effective in identifying thoracic anomalies, MRI offers greater accuracy and shows stronger correlation with postnatal diagnostic outcomes. Cardiomeastinal shift may be a clue, but it is also seen in other thoracic anomalies. Orellana-Donoso *et al.* published a systematic review highlighting the associations of unilateral lung agenesis with Cardiovascular, Gastrointestinal and Skeletal malformations.⁸ None of our three cases underwent prenatal imaging.

Unilateral pulmonary agenesis is often diagnosed postnatally. A review conducted in a 2022 by Fukuoka *et al.* included 259 cases. One hundred and thirty-seven cases were identified during the neonatal period, with 40 suspected prenatally.⁹ Mukherjee *et al.* present a case with unilateral lung agenesis with multisystem involvement.¹⁰ Associated anomalies were common, especially cardiovascular (40%), skeletal (30%), gastrointestinal (20%), tracheal (20%), and genitourinary anomalies (14%).¹⁰

Diagnostic imaging is critical. Chest x-ray may reveal total opacity of one lung field with compensatory contralateral hyperinflation. CT with 3D reconstruction confirms the diagnosis and subtype. Kane *et al.* presented a 2-month child with unilateral pulmonary agenesis, which was incidentally found after the patient was admitted post-viral infection.¹¹

Antenatal diagnosis plays a very important role in early detection and quick management of such patients. Miyano *et al.* reported a case where antenatally, at 22 weeks of gestation, the diagnosis of unilateral lung agenesis with oesophageal atresia and tracheoesophageal fistula was made, and the patient was managed immediately after birth with successful postoperative recovery.¹²

Bronchoscopy is a valuable diagnostic tool, but it was not universally performed in all reported cases of pulmonary aplasia/agenesis—its use varied depending on patient stability, age, and the clarity of imaging findings. As compared to our patients, bronchoscopy was performed in a study published by Singh *et al.*, where a 15-year child with scoliosis was diagnosed with pulmonary agenesis.¹³ Another study published by Astuti *et al.* mentioned a 48-year lady diagnosed with hypoplastic left lung and hyperinflation of the right lung, who presented with haemoptysis.¹⁴ In older children, persistent or recurrent localised pneumonia may be the first clue. Misdiagnosis as foreign body aspiration has also been reported.¹⁵

Management is supportive, focusing on the correction of life-threatening or disabling anomalies. Prognosis has improved with advancements in early detection and neonatal care. Despite this, unilateral cases still carry a mortality rate. In the present case series, one patient with right lung agenesis did not survive.

This case series emphasises the importance of considering lung agenesis/aplasia in infants/children with persistent unilateral chest opacity and mediastinal shift. While a chest x-ray can raise suspicion, a definitive diagnosis requires a CT or MRI scan.¹⁶ Associated anomalies are common, as also noted in this case series. Early diagnosis allows appropriate follow-up and parental counselling.

PATIENTS' CONSENT:

Informed consent was obtained from the attendants of the patients to publish the data concerning these cases.

COMPETING INTEREST:

The authors declared no conflict of interest.

AUTHORS' CONTRIBUTION:

KJ: Concept and data collection.

SAM: Article writing.

AQ: Referencing and editing.

AAA, BH: Review of the article.

All authors approved the final version of the manuscript to be published.

REFERENCES

1. Parry AH, Raheem MAI, Ismail HH, Sharaf O. Late presentation of unilateral lung agenesis in adulthood. *Egypt J Radiol Nucl Med* 2021; **52**:153. doi: 10.1186/s43055-021-00533-x.
2. Robertson N, Miller N, Rankin J, McKean M, Brodlie M, Thomas M. Congenital lung agenesis: Incidence and outcome in the North of England. *Birth Defects Res* 2017; **109**(12):857-9. doi: 10.1002/bdr2.1011.
3. Kayemba-Kay's S, Couvrat-Carcauzon V, Goua V, Podevin G, Marteau M, Sapin E, et al. Unilateral pulmonary agenesis: A report of four cases, two diagnosed antenatally and literature review. *Pediatr Pulmonol* 2014; **49**(3): E96-102. doi: 10.1002/ppul.22920.
4. Schwalbe E, Gruber GB. *Die Morphologie der Missbildungen des Menschen und der Tiere*. University of Leeds Library. Available from: <https://wellcomecollection.org/works/gwu88749>.
5. Boyden EA. Developmental anomalies of the lungs. *Am J Surg* 1955; **89**(1):79-89. doi: 10.1016/0002-9610(55)90510-9.
6. Abramowsky CR, Berkowitz FE. Giovanni Battista Morgagni (1682-1771), the first pediatric pathologist. *Pediatr Dev Pathol* 2015; **18**(6):458-65. doi: 10.2350/15-05-1640-OA.1.
7. Biyyam DR, Chapman T, Ferguson MR, Deutsch G, Dighe MK. Congenital lung abnormalities: Embryologic features, prenatal diagnosis, and postnatal radiologic-pathologic correlation. *Radiographics* 2010; **30**(6):1721-38. doi: 10.1148/rg.306105508.
8. Orellana-Donoso M, Barrenechea-Salvador M, Caro-Navarro J, Cervela-Diaz M, Chacon-Ortiz C, Claudet-Cordoba N, et al. Unilateral lung agenesis: A systematic review of prevalence, anatomical variants, and clinical implications. *Diagnostics (Basel)* 2025; **15**(17):2272. doi: 10.3390/diagnostics15172272.
9. Fukuoka S, Yamamura K, Nagata H, Toyomura D, Nagatomo Y, Eguchi Y, et al. Clinical outcomes of pulmonary agenesis: A systematic review of the literature. *Pediatr Pulmonol* 2022; **57**(12):3060-8. doi: 10.1002/ppul.26135.
10. Mukherjee TG, Thurmann KE, Richardson R. Unilateral lung agenesis and left isomerism in a neonate: A case report of multi-system anomalies. *Cureus* 2025; **17**(7):e88241. doi: 10.7759/cureus.88241.
11. Kane B, Camara MA, Togo S, Traore MM, Toure BM, Ouattara M. Isolated right pulmonary agenesis in a 2-month-old infant in the pediatric ward of hospital of Mali. *Open J Pediatr* 2020; **10**(2):274-9. doi: 10.4236/ojped.2020.102028.
12. Miyano G, Morita K, Kaneshiro M, Miyake H, Koyama M, Nouse H, et al. Unilateral pulmonary agenesis associated with oesophageal atresia and tracheoesophageal fistula: A case report with prenatal diagnosis. *Afr J Paediatr Surg* 2015; **12**(1):86-8. doi: 10.4103/0189-6725.151000.
13. Singh U, Jhim D, Kumar S, Mittal V, Singh N, Gour H, et al. Unilateral agenesis of the lung: A rare entity. *Am J Case Rep* 2015; **16**:69-72. doi: 10.12659/AJCR.892385.
14. Astuti TW, Listiandoko RDW, Duta GA, Indah Sari GAF, Erawati DR. Case report: Unilateral pulmonary hypoplasia presenting as recurrent pulmonary infection. *Int J Sci Res Pub* 2021; **11**(12):143-7. doi: 10.29322/IJSRP.11.12.2021.p12021.
15. Parmeswaran K, Lim R, Lynch T. Case 2: The foreign body that wasn't. *Paediatr Child Health* 2013; **18**(3):137-9. doi: 10.1093/pch/18.3.137a.
16. Lee EY, Boiselle PM, Cleveland RH. Multidetector CT evaluation of congenital lung anomalies. *Radiology* 2008; **247**(3):632-48. doi: 10.1148/radiol.2473062124.

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