CASE REPORT

Aplasia of the lung in a neonate—the myth is explored

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SUMMARY

A term appropriate for gestational age baby boy was delivered by lower segment caesarean section. The baby was antenatally diagnosed with resolving cystic adenomatoid malformation. At birth, he developed respiratory distress requiring active resuscitation and later was diagnosed to have right lung aplasia, managed with respiratory support. He gradually improved and was discharged home on room air.

BACKGROUND

A newborn delivered with respiratory distress warrants a chest roentgenogram. Unresolved opaque lungs need bronchoscopy and chest CT to delineate any lung malformation, and for prognostication.

CASE PRESENTATION

A male infant boy weighing 2500 g, with an antenatal diagnosis of resolving cystic adenomatoid malformation, was born by elective lower segment caesarean section. He required bag and mask ventilation at birth and was shifted to neonatal intensive care unit with an endotracheal tube in situ, and later connected to a conventional ventilator. On examination, he was eumorphic with anthropometry appropriate for gestational age. His chest roentgenogram showed a right homogenous opaque lung with mediastinum shifting to the right ([figure 1](#)) and pneumothorax on the left ([figure 2](#)). Ultrasound of the chest demonstrated a non-aerated right lung ([figure 3](#)). Echocardiography demonstrated bidirectional patent ductus arteriosus with severe pulmonary hypertension (70 mm Hg) with dilated right atrium and ventricle. CT of the chest showed a right hyperechoic lung with right bronchus ending blindly, with probable aplasia of the right lung ([figure 4](#)). Bronchoscopy revealed a blindly ending bronchus.

The baby was managed with judicious ventilator care and gradually weaned to continuous positive airway pressure, and then to room air. The patient developed recurrent pneumothoraces requiring intercostal drainage. He was given intravenous sildenafil for pulmonary hypertension, which he was gradually weaned off. Presently, he is stable on full feeds without any respiratory support. His follow-up clinics were unremarkable.

INVESTIGATIONS

- ▶ Blood gas analysis
- ▶ Chest X-ray
- ▶ Echocardiogram
- ▶ CT scan
- ▶ Bronchoscopy

Figure 1 Chest X-ray demonstrating homogenous opacity of right lung with mediastinal shift to right.

Figure 2 Chest X-ray demonstrating left-sided pneumothorax.

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Figure 3 Chest ultrasound revealing non-aerated right lung.

DIFFERENTIAL DIAGNOSIS

- ▶ Total lung collapse
- ▶ Haemothorax

TREATMENT

- ▶ Judicious management of respiratory care
- ▶ Infection control
- ▶ Nutrition
- ▶ Immunisations
- ▶ Developmental care

OUTCOME AND FOLLOW-UP

At writing, the baby is stable on full feeds and anthropometry is unremarkable, with milestones adequate for age.

DISCUSSION

In 1909, Schneider proposed a classification for the degree of underdevelopment of the lung. Class I agenesis, characterised by total absence of bronchus and lung; class II aplasia by rudimentary bronchus without lung tissue and class III hypoplasia suggestive of bronchial hypoplasia and variable but reduced amount of lung tissue.

Pulmonary aplasia is a rare congenital pathology characterised by unilateral or bilateral absence of lung tissue. The main

difference is a short-blind ending bronchus in aplasia. Primary pulmonary hypoplasia is rare. It may be caused by an embryological defect of the lung or vascular tissues, or an in utero vascular accident. Hypothesis for pulmonary aplasia pathogenesis is explained by abnormal blood flow in the dorsal aortic arch during the fourth week of gestation. The contralateral lung may develop as much as twice the amount of alveoli in response to pulmonary aplasia. The condition is associated with other congenital abnormalities, mainly cardiovascular, and has been reported to occur with VACTERL syndrome. On prenatal ultrasound screening, it presents as hyperechoic hemithorax.^{1 2}

Infants with unilateral pulmonary aplasia may have variable clinical presentations, depending on the extent of lung involvement and comorbidities. Some infants present with severe respiratory distress in the first few hours of life, whereas some may be completely asymptomatic. Some patients may present months to years later with repeated pulmonary infections and wheezing. In rare cases, the condition goes unnoticed until later in childhood and adolescence.^{3 4} In the index case, the infant presented with respiratory distress at birth, requiring intubation and ventilation. Chest X-ray showed a completely opaque hemithorax and contralateral lung hyperinflation. CT of the chest demonstrated absence of lung parenchyma and shift of mediastinum to the same side, with cardiac congenital malformations and ipsilateral bronchus remnants. Wu *et al*⁵ described a case of unilateral lung hypoplasia diagnosed by three-dimensional reconstruction of helical chest CT. Argent and Cremin⁶ used contrast medium-enhanced CT to elucidate the anatomy of two patients with right lung agenesis.

In patients with pulmonary aplasia, a number of associated anomalies have been described, which include hydrops fetalis, renal anomalies, diaphragmatic hernia, skeletal anomalies, oligohydramnios and polyhydramnios, heart defects, facial abnormalities, and lung and tracheo-bronchial tree defects.⁷⁻¹³

The diagnosis of unilateral pulmonary aplasia requires a high index of clinical suspicion. An important aspect in management of congenital pulmonary aplasia is judicious respiratory care including intermittent mandatory ventilation, high-frequency oscillatory ventilation and even extracorporeal membrane oxygenation. Other aspects of management include nutrition, infection control and probable surgical intervention at a later age. The prognosis depends on the size of the lungs and the underlying cause.

Figure 4 Chest CT showing right hyperechoic lung with right bronchus ending blindly with probable aplasia of right lung.

Learning points

- ▶ Aplasia of the lung is a rare lung malformation.
- ▶ Antenatal scan may not pick up the anomaly.
- ▶ Antenatally diagnosed congenital cystic adenomatoid malformation may accompany aplasia of lung.
- ▶ A newborn with a completely opaque lung warrants bronchoscopy.
- ▶ CT scan confirms the diagnosis.
- ▶ Constant counselling and communication are essential.
- ▶ Regular follow-up, prevention of chest infection, immunisation and adequate nutrition, are the key factors in management.

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