# Overcoming odds: neonatal survival in absent right main pulmonary artery with unilateral right pulmonary hypoplasia

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## SUMMARY

A unilateral absent pulmonary artery with unilateral pulmonary hypoplasia is an exceptionally rare congenital anomaly. This case report details the management of a neonate diagnosed antenatally with absent right main pulmonary artery and right pulmonary hypoplasia. The neonate developed respiratory failure within 24 hours of birth and was successfully managed with invasive ventilation and conservative treatment. Despite episodes of bronchiolitis during infancy, the child demonstrated normal growth and development at 1 year. This case highlights the importance of early diagnosis, multidisciplinary care and vigilant follow-up in achieving favourable outcomes in rare congenital conditions.

## BACKGROUND

Lung hypoplasia is an unusual congenital defect characterised by a reduction in the number or size of airways, arteries and alveoli but otherwise relatively normal overall morphology. An undersized, fibrotic, non-functioning lung is the result of underdeveloped alveolar tissue.[1] Majority of pulmonary hypoplasias are caused by congenital defects or problems during pregnancy that prevent the development of the lungs.[2 3] In most situations, perinatal mortality is significant and can occur in about 70% of cases.[2 4] Numerous conditions, including hydrops fetalis, congenital diaphragmatic hernia and renal anomalies, show interference with lung growth during the early developmental stage, primarily before 16 weeks of gestation. After 16 weeks, pulmonary development may be affected by oligohydramnios, a condition caused by prolonged rupture of the membranes.[2 5 6] Congenital lung abnormalities can be detected accidentally during regular imaging and investigations, but they can also be evident in the neonatal stage.[7] Babies diagnosed to have pulmonary hypoplasia in earlier scans usually have poor prognosis. The unilateral absence of pulmonary artery is an even rarer anomaly, and reports of the same in the neonatal period are scarce. This report discusses a rare case of unilateral right lung hypoplasia with an absent right main pulmonary artery detected in the second trimester and highlights the management and survival of the neonate beyond the critical neonatal period.

## CASE PRESENTATION

A primigravida mother was detected to have fetal right lung hypoplasia with right kidney pyelectasis and cardiac dextroversion on second-trimester antenatal scan. Fetal MRI revealed right lung hypoplasia with a left lung volume of 39.94 $cm^3$. Guarded prognosis was explained to parents prior to delivery. The baby was delivered at full term via vaginal delivery with a good APGAR score and had a birth weight of 2.5 kg. The baby did not have any features of trisomy on examination. As the baby had respiratory distress in the form of tachypnoea and chest indrawing, he was admitted in the neonatal intensive care unit (NICU) and kept on continuous positive airway pressure support (5/30%). Full orogastric (OG) feeds started on day 1 of admission. The baby had respiratory failure within 24 hours after birth and was managed with invasive mechanical ventilation by synchronized intermittent mandatory ventilation mode (SIMV mode). He required maximum inspiratory pressures of 22 cm $H_20$, positive end-expiratory pressure of 5 cm $H_20$ and $fiO_2$ 40%. Intravenous milrinone was added for persistent pulmonary hypertension (PPHN) as evidenced by pulmonary pressure of 52 mm Hg measured by tricuspid regurgitation (TR) jet on echocardiography (ECHO).

## INVESTIGATIONS

Chest X-ray revealed the right lung hypoplasia with the heart position on the right side (figure 1). 2D echocardiography revealed dextroversion, right ventricular hypertrophy, patent foramen ovalis, small subaortic ventricular septal defect (VSD) and small patent ductus arteriosus (PDA). CT pulmonary angiography confirmed the finding of right lung hypoplasia with dextroversion along with absent right main pulmonary artery (figures 2 and 3). Genetic workup was not done in our case due to financial constraints.

## DIFFERENTIAL DIAGNOSIS

Pulmonary aplasia was considered along with pulmonary hypoplasia. Pulmonary hypoplasia was confirmed over aplasia based on CT pulmonary angiography findings, which showed an absent right main bronchus and pulmonary artery but the presence of underdeveloped lung tissue. Differentiation was based on the presence of compensatory lung growth and imaging characteristics.

## TREATMENT

Persistent pulmonary hypertension in the newborn was managed conservatively with milrinone and sildenafil. The baby was weaned off from invasive mechanical ventilation after 6 days of birth, and



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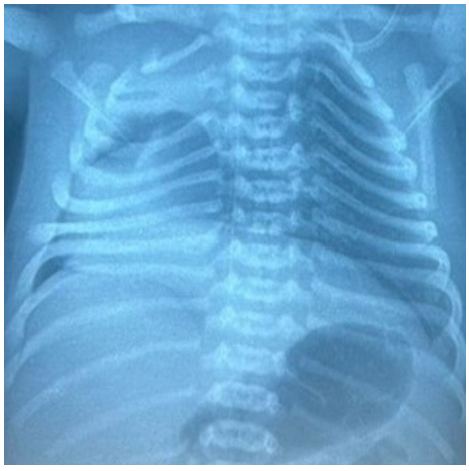

**Figure 1** Chest X-ray revealing right lung hypoplasia.

oxygen support was provided through a heated humidified high-flow nasal cannula. The oxygen support was gradually weaned off by a week of intensive care stay. Phototherapy was provided for 48 hours for raised bilirubin due to ABO incompatibility. Repeat bilirubin was normal. As the baby had congenital malformation and had a skin tag over the right cheek, malformation scans were done. Neurosonogram was normal while ultrasound of the abdomen revealed right hydronephrosis with a prominent right lower ureter. He had a multidrug-resistant Klebsiella urinary tract infection (UTI) during the neonatal period which was managed with intravenous colistin. Micturating cystourethrogram done after clearance of UTI revealed grade IIvesicoureteral reflux (VUR) on the right side. Otoacoustic emissions and brain stem evoked response audiometry were normal. Peculiarly, all the abnormalities in this baby were observed only on the right side. Parents were initially apprehensive of the condition of the baby and treatment. Repeated counselling sessions allayed the fears. Family participatory care facilitated effective neonatal

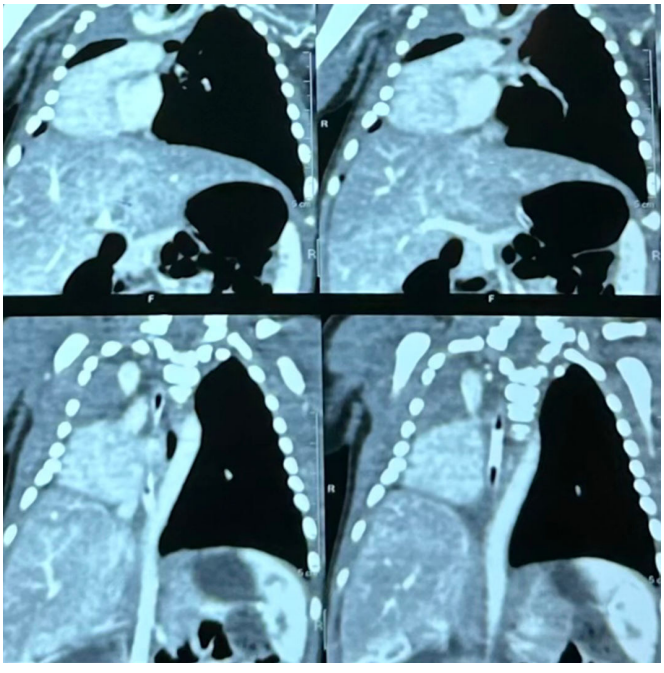

**Figure 2** Coronal section of the CT chest showing right lung hypoplasia.

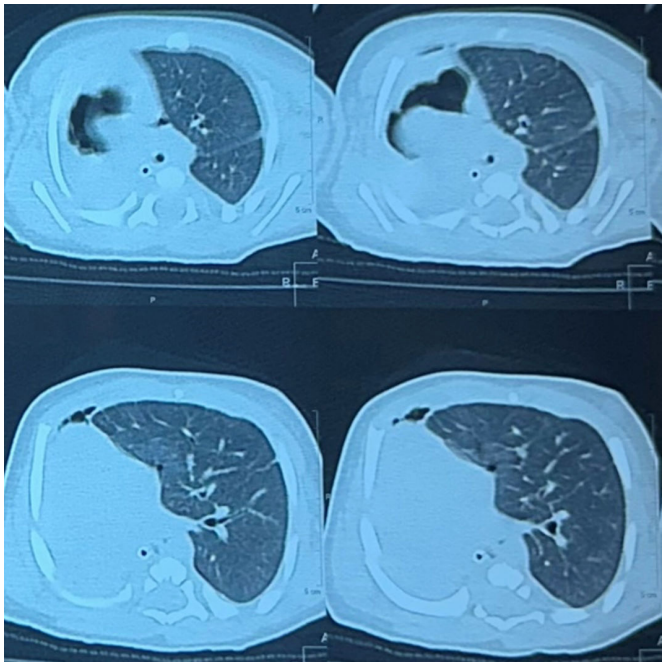

**Figure 3** Axial section of CT pulmonary angiography revealing right lung hypoplasia with absent right main bronchus and right branch of the pulmonary artery along with compensatory enlargement of left lung parenchyma.

care. He was discharged after 4 weeks of therapy on exclusive breastfeeding along with uroprophylaxis with cephalexin.

## OUTCOME AND FOLLOW-UP

Later, the baby presented to paediatric emergency, once at 4 months of age and again at 6 months of age with respiratory distress. On both occasions, the baby was diagnosed with bronchiolitis and was discharged with conservative management. He did not require any further respiratory support during infancy and his last chest X-ray available at 6 months of age. It revealed right lung hypoplasia with a total of 13 ribs on the right side including an extra floating rib and cervical rib on the left side with no obvious fractures (figure 4). A repeat 2D echo at 4 months of age was suggestive of dextroversion with small subaortic VSD with left to right shunt and left ventricular ejection fraction of 70% with no PDA. VUR was managed conservatively with only uroprophylaxis, and there were no episodes of breakthrough UTI. He had normal blood pressure, urine output and serum creatinine 0.25 mg/dL at 9 months of age. Uroprophylaxis was stopped at 9 months of age. He was immunised as per the National Immunisation Schedule including pneumococcal vaccine, *Haemophilus influenzae* type b (Hib) vaccine and nasal influenza vaccine. Genetic counselling for future pregnancy was carried out for parents. At 1 year of age, he had a weight of 8.5 kg, length of 72 cm and head circumference of 46 cm, achieved all developmental milestones as per age and was thriving well on complimentary feeds. This case highlights the importance of a multidisciplinary approach using the expertise of neonatologist, respiratory therapists, occupational therapist, physiotherapist, paediatrician, cardiologist and ongoing support in managing such complex conditions.

## DISCUSSION

Pulmonary hypoplasia is defined as an absolute decrease in the lung volume and weight for gestational age. Pulmonary

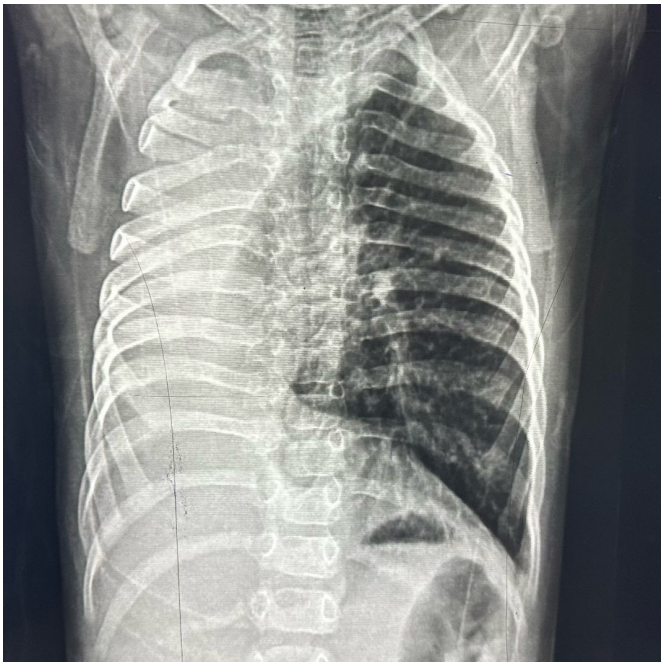

**Figure 4** Chest X-ray at 6 months of age showing homogeneous opacity in the right hemithorax with loss of right cardiac and diaphragmatic silhouette. Heart shadow is not visualised, and it is pulled towards the right side. The left lung is hyperinflated and herniated towards the right side. Fundal gas is seen on the left side. There are 13 ribs on the right side including an extra floating rib and cervical rib on the left side with no obvious fractures.

hypoplasia results from intrathoracic or extrathoracic compression of the lungs, amniotic fluid volume disturbance, phrenic nerve abnormalities leading to abnormal fetal breathing, decreased pulmonary blood flow, primary mesodermal defects or idiopathic causes. Common associated anomalies in pulmonary hypoplasia are renal causes (renal agenesis, dysplastic kidneys, urinary outlet obstruction), oligohydramnios, other anomalies like congenital diaphragmatic hernia (CDH) or congenital anomalies of the neuromuscular system and chromosomal abnormalities, including trisomy 13, 18 and 21.

The onset of pulmonary hypoplasia due to any antecedent cause will lead to more severe hypoplasia than late-onset hypoplasia. The prognosis of pulmonary hypoplasia depends on the underlying causes and severity. Neonatal mortality is reported to be 70%, and some instances of improvement are observed post amnioinfusion. The prognosis also depends on the associated severity of pulmonary hypertension.[8]

Lung hypoplasia can be classified as primary (idiopathic) or secondary (when it occurs in association with environmental factors or other congenital anomalies that may be implicated in its pathogenesis). According to estimates, there are one or two cases of primary hypoplasia for every 12 000 births. In newborn autopsy, the incidence of primary hypoplasia to varied degrees has been found to range from 7 to 26%.[5] Secondary pulmonary hypoplasia, which accounts for more than 85% of all cases, occurs in association with other abnormalities in the developing fetus. Most frequent causes are diaphragmatic hernia and renal defects.[8] Although the prevalence of the secondary form is unknown, it is probably more prevalent than is widely acknowledged due to its combination with a number of other anomalies and the complexity of pathological identification in several patients. Correctable causes have a better outcome. Most frequent correctable causes are diaphragmatic hernia and renal defects. Thoracic space-occupying lesions include CPAM (congenital pulmonary airway malformation), bronchopulmonary sequestration, mediastinal masses and lymphatic malformations, which are other correctable causes. When unilateral, the left lung is more frequently affected than the right.[9] In the present case report, we report a rare case of unilateral right lung hypoplasia.

In general terms, hypoplastic lungs are smaller and weigh less than one would anticipate given their age. Measurement of fetal lung length and comparison of the thoracic circumference with abdominal circumference, biparietal diameter or femur length is used for the diagnosis of pulmonary hypoplasia. However, these measurements can be erroneous in growth-restricted fetuses. Three-dimensional ultrasound-derived nomograms more reliably predict pulmonary volumes. Doppler ultrasound velocimetry can also be used to detect pulmonary vascular changes. This may correlate with the degree of pulmonary hypoplasia; however, reports of its usage are minimal until in recent years, MRI of the fetal lung is used as a better modality for diagnosing pulmonary hypoplasia. However, there is no universally available standardised values for its confirmation. Hence, overall antenatal diagnosis of pulmonary hypoplasia is at best modest, and it is best diagnosed postnatally. The presence of oligohydramnios and growth restriction raises the suspicion of pulmonary hypoplasia. The commonly used postnatal tests for confirming pulmonary hypoplasia are lung weight, lung weight/body weight ratio, radial alveolar count, alveolar count per unit volume and lung volume measurement and correlation with crown-rump length.[10] A decrease in the number of airway generation, ranging from around 50 to 75% of normal, is the most constant observation, despite variations in the degree and kind of changes between individual cases. Furthermore, there is often a drop in alveolar count, which one group of scientists assessed to be around one-third normal. This is frequently linked to a reduction in alveolar mass. While some researchers have observed an immature appearance, others have shown typical alveolar and airway development for gestational age. The pulmonary artery system has also been shown to exhibit abnormalities, including less elastic tissue in the bigger arteries, increased muscle in the arteries that should typically be muscular and muscle extension into the arteries that should ordinarily not be muscular.[1] Our baby also had reduced chest expansion on the right side and decreased breath sounds on the right along with PPHN.

Pulmonary hypoplasia frequently coexists with other congenital abnormalities and have significant rate of occurrences. There have been reports of anencephaly, diaphragmatic hernia, heart lesions, hepatic anomalies, thumb abnormalities, thoracic spine deformities, urinary tract abnormalities, renal anomalies and pleural effusions. These anomalies may be related to one another.[11] Our case had right-sided VUR along with right pulmonary hypoplasia, absent right main pulmonary artery along with subaortic VSD.

It is also known that primary pulmonary hypoplasia is rare and occurs due to alterations in the transcription factor and/or growth factor signalling. *NKX2-1* is a critical gene which encodes the protein thyroid transcription factor-1 and is necessary for lung development. Mutation in this gene can lead to pulmonary hypoplasia and is also associated with thyroid and ventral forebrain malformations. Pulmonary hypoplasia can be seen in association with chromosomal abnormalities, including trisomy 13, 18 and 21, and in multicomponent disorders, like Scimitar, Eagle-Barret, Pena-Ahokeir and Smith-Lemli-Opitz

syndrome.[8] Genetic workup could not be done in our case due to financial constraints.

A unilateral absence of the pulmonary artery is an unknown complication. The unilateral absence of the pulmonary artery (UAPA) is a rare congenital malformation. It is often associated with other cardiac anomalies. There are instances where it has been reported as an isolated lesion as well. Isolated absence of the right pulmonary artery is twice more frequent than that of the left pulmonary artery. Isolated UAPA is usually asymptomatic at birth and develops symptoms like exercise intolerance, dyspnoea, chest pain, haemoptysis and recurrent pulmonary infections during childhood or adulthood. As patients are predominantly asymptomatic, hence, the diagnosis of isolated UAPA is difficult in infancy. Due to the rarity of neonatal presentation, there is no consensus regarding the treatment of this malformation.[12] In our case, probably, the absent right pulmonary artery led to right pulmonary hypoplasia. The baby initially presented with PPHN which normalised on conservative management. To the best of our knowledge, this is the third case of neonatal presentation of UAPA reported in the literature to date.[12] Our patient did not need any long-term treatment and follow-up till 1 year was normal. The need of long-term cardiovascular follow-up was emphasised.

Medical and surgical therapy are used to treat hypoplasia, both before and after delivery. Pulmonary hypoplasia can present with preterm labour. In such a scenario, tocolytics, antenatal steroids and antibiotics form a part of immediate management of preterm labour. Antenatal intervention in the form of fetoscopic endoluminal tracheal occlusion is available in CDH to improve the pulmonary development. Postnatal management of pulmonary hypoplasia is by gentle ventilation strategy. Underlying conditions like obstructive uropathy, CDH are managed by surgical correction, while the outcome is grim when severe anomalies like renal agenesis, dysplastic kidneys are the aetiology of pulmonary hypoplasia. In certain instances, extracorporeal membrane oxygenation may be required. Dialysis could be necessary to maintain renal function.[1] Regarding UAPA treatment, there is no universally accepted approach. The patient's symptoms, the structure of the pulmonary arteries and the related aortopulmonary collaterals should all be taken into consideration while developing a treatment plan. These patients may benefit from primary versus staged pulmonary artery anastomosis, closure of specific collateral arteries that are not exclusively in charge of pulmonary blood flow or partial or complete pneumonectomy.[13] In our case, the baby was provided with invasive mechanical ventilation for 6 days after the birth, and thereafter, oxygen support was given through a heated humidified high-flow nasal cannula. He had UTI due to VUR which was managed conservatively; there was no associated renal failure.

Based on a survey of cases reported between 1977 and 2003, the death rate was high, 3 of the over 30 newborns with congenital primary pulmonary hypoplasia survived out of the NICU, while neonatal reports of outcome in UAPA are scarce. The reported neonatal median age of death in pulmonary hypoplasia after the delivery was 9 hours.[4] According to a retrospective study done in Spain, 75% of fatalities occur on the first day of birth, and 47% occur within the first 60 days of life.[14] A review of literature by Odd *et al* had concluded that majority of the affected infants of pulmonary hypoplasia succumb to ventilation failure.[15] Some of the babies who survive can present with recurrent infections or wheezing and may require pulmonectomy of non-functional hypoplastic lung.[16] In the present case, the baby is currently doing well at 1 year of age.

## Learning points

- Unilateral absent pulmonary artery with unilateral pulmonary hypoplasia is a rare congenital anomaly and survival beyond the neonatal period can be achieved with multidisciplinary care and long-term follow-up.
- Complete immunisation and vigilant follow-up are crucial to prevent and manage recurrent respiratory illnesses.
- High-quality antenatal and postnatal counselling for parents is essential for managing expectations and providing emotional support.

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