Case Report

A rare cause of pneumothorax in a neonate: A case report

Anjum Hussain[1], Poorva Gohiya[2], Nitesh Upadhyay[3], Mohammed Shahid[3]

From, [1]Post graduate resident, [2]Associate Professor, [3]Assistant Professor, Department of Pediatrics, Gandhi medical college, Bhopal, MP, India

ABSTRACT

Congenital pulmonary airway malformation (CPAM) is a rare developmental lung malformation, typically presenting with respiratory distress in the neonatal period. It is typically diagnosed by fetal ultrasound. We describe a baby boy (22 days old) presenting with respiratory distress. He was admitted at birth for perinatal asphyxia and respiratory distress and was discharged on the 10th day of life. Examination showed severe subcostal retraction with tracheal shift towards the left and reduced breath sounds on the right side of the chest. X-rays of the chest revealed a hyperlucent right lung with a significant mediastinal shift towards the left and compression of the left lung. High-resolution computed tomography (HRCT) of the chest demonstrated a large multicystic lesion almost replacing the right upper and middle lobes, suggestive of CPAM. A lobectomy was planned, but the baby succumbed before surgical intervention. A pathological diagnosis could not be made. CPAM is a rare but significant cause of neonatal respiratory distress, often mimicking conditions like pneumothorax, which can lead to diagnostic challenges. This case underscores the importance of considering CPAM in neonates with persistent respiratory distress and highlights the role of imaging in early diagnosis. Timely recognition and surgical intervention remain crucial to improving outcomes and preventing fatal complications.

Key words: Congenital Pulmonary Airway Malformation, Pneumothorax, Respiratory Distress Syndrome, Lung Malformations

Pediatric lung disorders encompass a wide range of congenital and acquired conditions, including bronchopulmonary dysplasia, congenital pulmonary airway malformation (CPAM), pulmonary sequestration, congenital lobar emphysema, pneumonia, asthma, cystic fibrosis, bronchiolitis, and tuberculosis, each presenting with varying degrees of respiratory compromise and requiring tailored management approaches. Congenital pulmonary airway malformation (CPAM), previously referred to as congenital cystic adenomatoid malformation (CCAM), results from the cessation of lung development during the sixth and seventh week of foetal development. Some authors believe they are hamartomatous lesions of the bronchial tree. It presents in 0.004% of all pregnancies and constitutes almost 95% of all congenital cystic lung diseases. The incidence of CPAM is reported to be 1 in 10000 to 1 in 35000 births. They contribute to 42% of paediatric lung resections, 86% identified by age 5 [1]. They occur sporadically with no genetic predisposition (except type 4) and no association with maternal factors [2].

In 2002, Stocker proposed the term congenital pulmonary airway malformation (CPAM) as all the lesions are neither cystic nor adenomatoid. In this CPAM classification, five types have been described based on morphology and histology. Type 0 arises from the trachea and is typically fatal due to respiratory insufficiency. The most common is Type 1 (~70%), which consists of large cysts and has a good prognosis after surgical resection. Type 2 features smaller cysts and is often associated with other congenital anomalies. Type 3 presents as microcystic lesions and can lead to severe pulmonary hypoplasia and fetal hydrops. Type 4 originates from distal acinar structures and has a risk of malignant transformation, including pleuropulmonary blastoma [3, 4].

CPAM exhibits a spectrum of clinical presentations, ranging from asymptomatic cases detected on prenatal ultrasonography to severe neonatal respiratory distress. The severity of symptoms largely depends on the size and location of the lesion. In severe cases, large cystic lesions can cause pulmonary hypoplasia, mediastinal shift, and hydrops fetalis, leading to fetal demise. In the developed world, the majority of cases are diagnosed prenatally in foetal ultrasound. Computed tomography of the chest (CT Chest) is often diagnostic, but histological examination is definitive. Management depends on the severity and size of the lesion. Prenatally, large lesions with hydrops may require interventions like steroids or fetal surgery. Postnatally, asymptomatic cases are monitored, but surgery (lobectomy/segmentectomy) is often advised to prevent

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Correspondence to: Dr. Anjum Hussain K T, Department of Pediatrics, Gandhi medical college, Bhopal, Madhya Pradesh, India.

Email: dranjumhussainkt@gmail.com

complications, leading to good long-term respiratory outcomes.

CASE REPORT

A 22-day-old male baby weighing 3.2 kg presented with a history of breathing difficulty for the past 3 days. He was earlier admitted at birth for 10 days for perinatal asphyxia and respiratory distress and was discharged. On admission to the special newborn care unit (SNCU), he was afebrile, with a heart rate of 122 beats per minute, a respiratory rate of 64 breaths per minute, and a SpO_2 of 80%. Respiratory system examination revealed severe subcostal retraction, hyperresonance on the right side with mediastinal shift towards the left, and reduced breath sounds on the right side. Based on the above clinical findings, a provisional diagnosis of pneumothorax was considered. On admission, the baby was kept under oxygen via nasal prongs at 0.5L/min. Laboratory investigations showed haemoglobin 16.7gm%, total leucocyte count 4200/mm^3, and the platelet count 1.5 lakh/mm^3. On the chest x-ray, the right lung looks collapsed with significant opacity. [Figure 1] The primary diagnosis was pneumothorax, and differential diagnoses of Congenital diaphragmatic hernia (CDH), Pneumonia, and CPAM were considered.

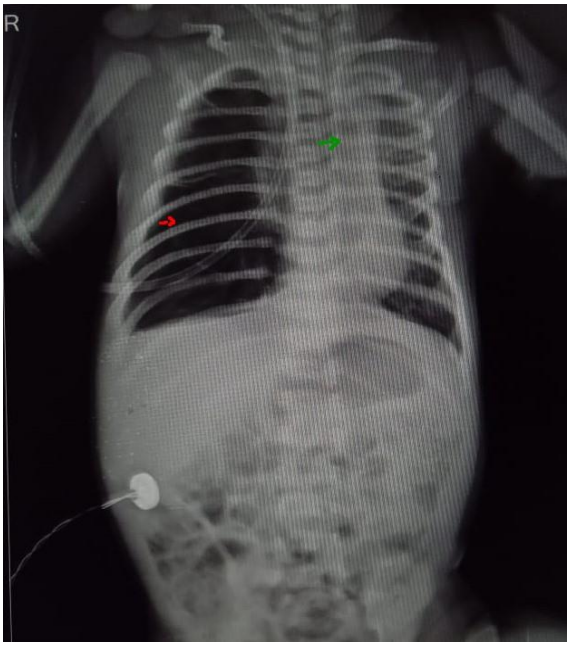

Figure 1 - X-ray chest showing hyperlucent right lung (red arrow) with significant mediastinal shift towards left (green arrow) and compression of left lung

An intercostal drainage was inserted in the right chest. However, the respiratory distress did not settle. High-resolution computerised tomography (HRCT) of the chest was done, which revealed a large multicystic lesion measuring approximately 2.9*6.2*3.1cm3, almost replacing the right upper and middle lobes, with a contralateral shift of mediastinum compressing the left lower lobe. The left lung was aerated normally. [Figure 2] The possibility of congenital pulmonary airway malformation of the right lung was kept. A surgical review was done, and surgical resection of the affected lobes was planned. However, respiratory distress worsened, and the baby was intubated. He succumbed before surgical intervention could be done.

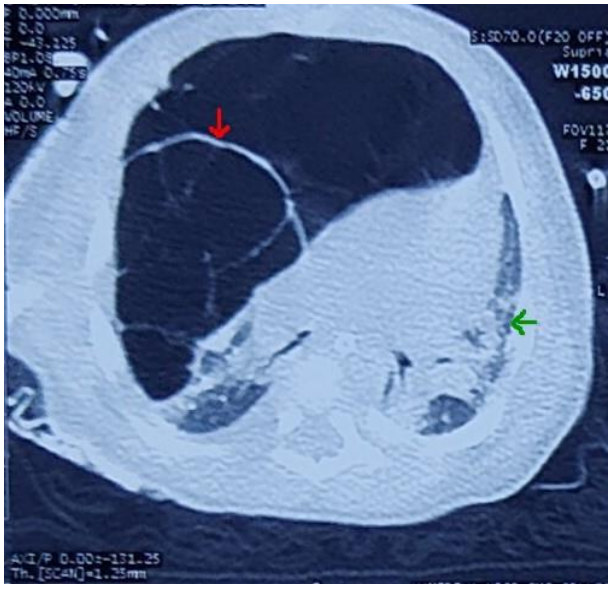

Figure 2 - HRCT Chest showing a large multicystic lesion almost replacing the right upper and middle lobe (red arrow) with contralateral shift of mediastinum compressing the left lower lobe (green arrow).

DISCUSSION

CCAMs were first reported in 1897 by Stoerk et al. [5]. Although rare overall, CPAM accounts for 95% of congenital cystic lung diseases [1]. They are the most common congenital parenchymal lung malformations. In 2002, Stocker proposed a classification system dividing CPAM into five distinct types based on histopathological and morphological features, reflecting the progression of developmental abnormalities through the tracheobronchial tree. Type 0 arises from the trachea and is usually lethal due to complete pulmonary insufficiency. Type 1 is the most common (~70% of cases), characterised by large cysts (>2 cm) lined by pseudostratified columnar epithelium. It has an excellent prognosis after surgical resection. They rarely undergo malignant transformation into broncheo-alveolar carcinoma. Type 2, the second most common type, is associated with bronchiolar structures and smaller cysts. It is frequently related with other congenital anomalies (e.g., renal agenesis, cardiac defects, diaphragmatic hernia). Type 3, also arising from the bronchiolar region, presents with microcystic lesions and can cause fetal hydrops due to severe pulmonary hypoplasia.

Type 4 originates from distal acinar structures and has a high association with pleuropulmonary blastomas, necessitating close monitoring for malignancy [3, 4]. They can often present as pneumothorax. CPAM may present at birth with respiratory distress or may go through infancy into childhood and may present with repeated pulmonary infections,

hemoptysis, pneumothorax, chest pain, malignancy, or as an incidental finding in radiological images [6, 7]. Severe cases may present in utero as pulmonary hypoplasia, mediastinal shift, and hydrops fetalis, leading to fetal demise [8].

CPAM is typically diagnosed through a combination of prenatal imaging and postnatal assessments. CPAM is diagnosed during prenatal ultrasound examination, where it may present as an echogenic mass in the fetal thorax, although it is identified in the second trimester in the majority of cases. Further evaluation using fetal MRI can provide detailed information about the lesion's size, location, and potential impact on surrounding structures. After birth, asymptomatic cases may be identified incidentally during imaging for other reasons. However, symptomatic infants might undergo chest X-rays and computed tomography (CT) scans to confirm the diagnosis and assess the extent of the malformation [9]. However, tissue diagnosis is essential for a definitive diagnosis. Histologically, CPAM lesions exhibit disorganized lung architecture, cystic spaces, and abnormal epithelial differentiation.

Neonatal pneumothorax management depends on size and cardiovascular stability. Small and medium pneumothoraces are managed with close observation. Large pneumothoraces in stable infants are managed with chest drain insertion. Tension pneumothoraces with cardiovascular instability are managed with emergency needle thoracocentesis followed by chest drain placement. Ventilatory support should be optimised by minimising pressure and ensuring adequate expiratory time. Regular chest X-rays help monitor progress [10]. The management of CPAM varies, with different approaches depending on clinical severity and affected lung volume. Large CPAM lesions causing hydrops may require in utero interventions, including steroid therapy or fetal surgery. Postnatally, asymptomatic infants are often monitored with serial imaging.

However, many experts advocate for elective surgical resection to prevent recurrent infections, pneumothorax, or potential malignant transformation. Lobectomy or segmentectomy, depending on the lesion size and location [11, 12], is the definitive treatment. Early intervention has been shown to result in good long-term pulmonary function. With advancements in fetal imaging, surgical techniques, and neonatal care, the prognosis of CPAM has improved significantly. Timely surgical resection generally results in excellent long-term respiratory outcomes in infants, depending on remaining normal lung tissue, highlighting the need for early detection and multidisciplinary management.

Although CDH also causes pulmonary hypoplasia and mediastinal shift, it involves the displacement of abdominal contents into the thoracic cavity and presents with scaphoid abdomen and cyanosis. X-ray findings include bowel loops in the thoracic cavity, mediastinal shift and absence of diaphragmatic contour. Pneumonia may also present with pneumothorax but is associated with systemic signs like fever and sepsis and improves on antibiotic therapy. X-ray findings include diffuse or focal consolidation, air bronchograms and pleural effusion.

CONCLUSION

CPAM is a rare but significant congenital lung anomaly that can present with varied clinical manifestations, ranging from asymptomatic cases to severe neonatal respiratory distress. This case highlights the diagnostic challenge of differentiating CPAM from other neonatal respiratory conditions, such as pneumothorax, congenital diaphragmatic hernia, and pneumonia. The failure to improve after chest drain placement and the subsequent HRCT findings led to the definitive diagnosis of CPAM. Despite advancements in prenatal imaging and neonatal management, severe cases may have high morbidity and mortality, as seen in our patient. Early recognition and timely surgical intervention remain crucial in improving outcomes. This case underscores the importance of a multidisciplinary approach in managing neonatal respiratory distress and the need for heightened clinical suspicion when pneumothorax fails to resolve as expected.

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