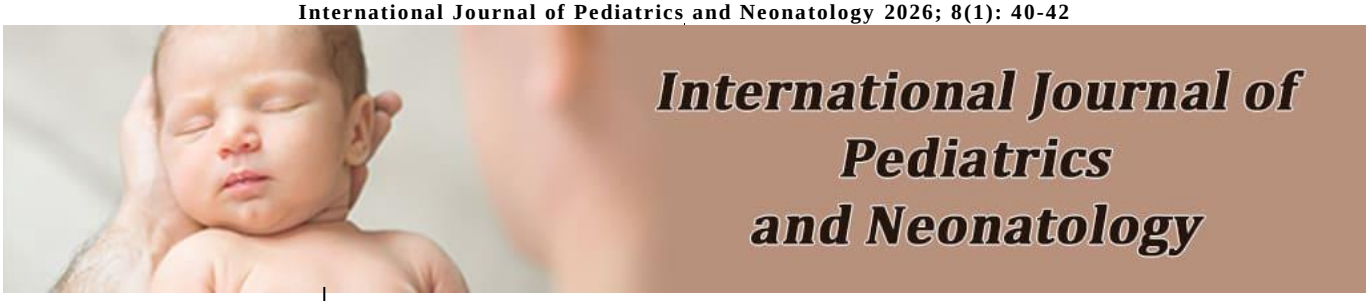

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A rare case of pulmonary alveolar proteinosis in a term neonate

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Abstract
Primary Alveolar Proteinosis (PAP) is a rare lung disorder caused by decreased clearance and accumulation of surfactant. It is classified into primary, secondary, and congenital forms. Congenital PAP, linked to mutations (e.g., in SP-B, SP-C, ABCA3), is a rare, potentially fatal condition in newborns, often presenting as respiratory failure. Diagnosis relies on lung biopsy and bronchoalveolar lavage (BAL). We report a case of male neonate, born at 38+3 weeks was admitted to the NICU for respiratory distress. CT imaging suggested PAP and bronchoalveolar lavage (BAL) confirmed the diagnosis by demonstrating lipid-laden macrophages. The infant showed clinical improvement following BAL, supportive care and was discharged off respiratory support with adequate feeding and weight gain. This case highlights that, early diagnosis and management can lead to favourable outcomes in congenital PAP, potentially avoiding the need for whole lung lavage or transplantation.

Keywords: Primary alveolar proteinosis, congenital pulmonary alveolar proteinosis, bronchoalveolar lavage, neonate, respiratory distress

Introduction
Primary Alveolar Proteinosis (PAP) is a rare intra-alveolar lung filling disorder due to surfactant accumulation. It is primarily due to decreased clearance of surfactant proteins within the alveoli [1]. The incidence of this rare disease is 0.2 cases per million and the estimated prevalence of 3.7-40 cases per million [2]. Since the initial description of this rare lung disease by Rosen *et al*. in 1958, a significant improvement was seen in its understanding. PAP is divided into three categories, with congenital PAP being the rarest (1%) and primary or autoimmune PAP being the most common form (90%), while secondary PAP accounts for 4% [3]. Congenital PAP is caused by gene mutation in surfactant proteins (SP): SP-B, SP-C and the ABCA3 transporter or by the absence of granulocyte/macrophage colony-stimulating factor (GM-CSF) receptor [4].
Neonatal PAP, a rare and potentially fatal condition affecting newborns, typically presents with clinical features ranging from asymptomatic to progressive respiratory failure. Lung biopsy and bronchoalveolar lavage confirm its diagnosis [5]. We present a rare case of a male neonate diagnosed with congenital PAP, initially manifesting as respiratory distress. Following bronchoalveolar lavage for PAP, the infant was successfully weaned off respiratory support, was tolerating enteral feeds, and demonstrated adequate weight gain upon discharge.

Case Report
A male neonate born at 38+3weeks gestation to a pregnancy-induced hypertension (PIH) mother via lower-segment cesarean section (LSCS) due to failed induction of labour. An antenatal anomaly scan revealed the presence of a single umbilical artery. The neonate cried immediately after birth, with APGAR scores of 6/10, 7/10. Due to respiratory distress (grunting, subcostal and intercostal retractions), the neonate was shifted to the NICU.
The neonate was started on high-flow nasal cannula (HFNC) at 6l/min. Chest radiography on day 1 of life showed bilateral diffuse haziness (Figure 1). The baby was weaned to room air on day 5 of life. 2D echo on day 2 of life showed mild-moderate patent ductus arteriosus (PDA), moderate pulmonary arterial hypertension (PAH).

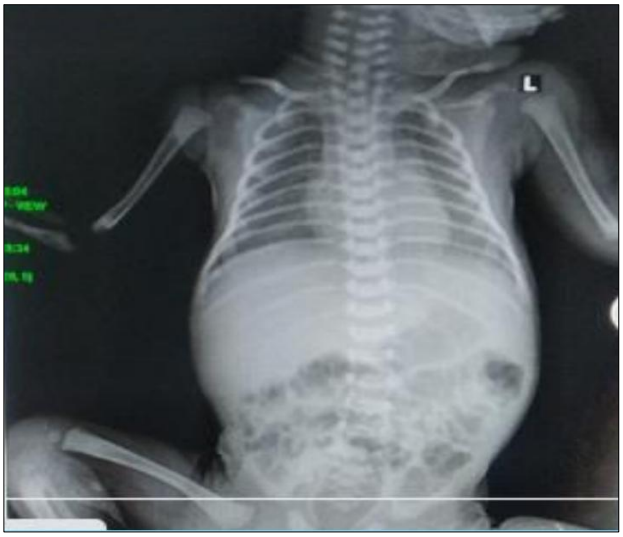

Fig 1: Chest X-Ray showing bilateral diffuse haziness

The neonate remained hemodynamically stable and did not require inotropic support. Feeds were started on day 2 of life, and slowly graded up as per tolerance. Initial sepsis work-up was negative, and antibiotics were stopped after 3 days. However, on day 8 of life, the baby had worsening tachypnoea and was restarted on HFNC support, sepsis work-up sent and started on iv antibiotics. Newborn screening (NBS) was normal. In view of persistent distress, microlaryngoscopy was performed on day 12 of life and was normal. Pulmonologist opinion was sought and CT chest (Figure 2: A, B, C, D) was done which showed diffuse ground glass opacities in bilateral lung parenchyma with subpleural sparing, suggestive of PAP.

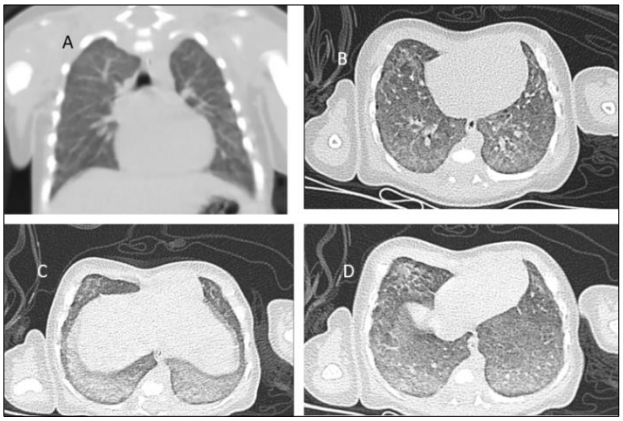

Fig 2: CT chest revealing diffuse ground glass opacities in bilateral lung parenchyma

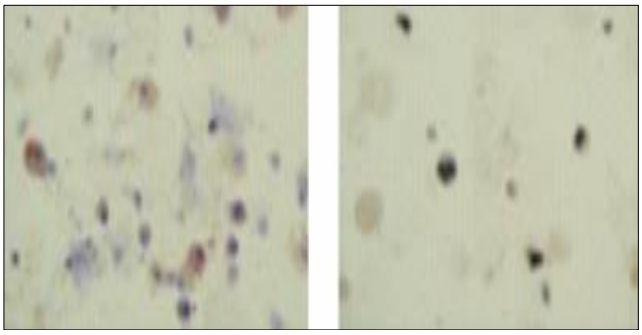

Fig 3: Histological examination of BAL fluid showing lipid laden macrophages

Bronchoalveolar lavage (BAL) was performed in view of PAP which revealed the presence of lipid laden macrophages. BAL smears studied are cellular composed of benign endobronchial, squamous epithelial cells along with many alveolar macrophages and occasional inflammatory cells. Numerous lipid laden macrophages are observed which is indicative of PAP (Figure 3). Geneticist opinion was sought, and genetic testing and immunoglobulin subset analysis was advised. The baby was symptomatically better post procedure and was weaned to room air by day 23 of life. The neonate was subsequently discharged off respiratory support, tolerating feeds well, with adequate weight gain.

Discussion

Pulmonary Alveolar Proteinosis (PAP) is a rare respiratory condition that is caused by the malfunction of the alveolar macrophages, resulting in the intra-alveolar accumulation of lipoproteinaceous material [6]. Although it lacks standardization and is highly invasive, performed under general anesthesia, Whole lung lavage is the current standard of care for PAP [7]. Use of corticosteroids (prednisolone), azithromycin, hydroxychloroquine may have some role in disease progression. Mutations in ABCA3 and SFTPB are associated with severe neonatal respiratory failure. Mechanical ventilation is frequently necessitated in congenital PAP [8]. In our case, the neonate improved following bronchoalveolar lavage and supportive respiratory care, without the need for mechanical ventilation.

The 1-year and 5-year post-transplant survival rates were approximately 81-83% and 56%, respectively. Infants transplanted before 1 year of age were reported to be more likely to have motor and speech delays compared with older children. In addition, chronic lung allograft dysfunction, most commonly as bronchiolitis obliterans, was a leading cause of late post-transplant mortality [9]. Emerging therapies include gene addition approaches targeting genetic surfactant dysfunction disorders. Adeno-associated viral vectors have been explored for SFTPB deficiency, while lentiviral vectors are under investigation for ABCA3-related disease as they can accommodate the larger ABCA3 gene [10].

High-resolution CT contributes significantly to the diagnosis of PAP by demonstrating widespread ground-glass opacification and septal involvement, findings that correlate with surfactant accumulation within the alveoli [11]. In our case, CT chest demonstrated diffuse ground-glass opacities with subpleural sparing, prompting further evaluation and ultimately supporting the diagnosis.

Primary and congenital forms of PAP are usually associated with chronic interstitial lung disease and may follow a slowly progressive course, with some patients ultimately requiring lung transplantation. Whole lung lavage remains the mainstay of treatment for primary PAP and is typically performed using normal saline warmed to body temperature to facilitate removal of accumulated surfactant material from the alveoli. The prognosis of idiopathic forms of PAP is variable, with cases of spontaneous remission as well as long-term symptom resolution after a single lavage procedure being reported [12]. In our case, the infant improved with bronchoalveolar lavage and supportive care, without requiring whole lung lavage or lung transplantation, suggesting a comparatively milder disease course.

Conclusions: Congenital pulmonary alveolar proteinosis (PAP) is a rare lung disorder that may present with early respiratory distress in neonates and requires a high index of suspicion for diagnosis. Our case highlights that timely evaluation, supportive interventions such as bronchoalveolar lavage can result in clinical improvement without the need for whole lung lavage or lung transplantation. Early recognition and timely intervention are essential for optimizing outcomes in affected infants.

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