VOLUME - 15, ISSUE - 04, APRIL - 2026 • PRINT ISSN No. 2277 - 8160 • DOI : 10.36106/gjra

Original Research Paper

Paediatrics

SEVERE ABCA3 MUTATION-ASSOCIATED PULMONARY ALVEOLAR PROTEINOSIS IN A TODDLER WITH UNEXPECTED CLINICAL RECOVERY: A CASE REPORT

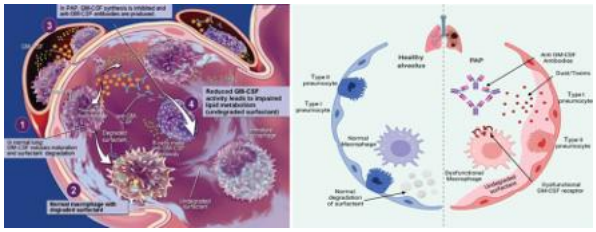
Dr. Shruti Naresh Dhale	MD, Professor and Head of Department, Paediatrics, Government Medical College, Alibag, Maharashtra, India
Dr Sudhakar Ashok Bantewad	MD, Associate Professor, Paediatrics, Government Medical College, Alibag, Maharashtra, India
Dr Abhishek Satwarao Pate*	MD, Senior Resident, Government Medical College, Alibag, Maharashtra, India *Corresponding Author
Dr Pratiksha Vilas Shinde	NBEMS Diploma, 1st Year PG Resident, Paediatrics, Government Medical College, Alibag, Maharashtra, India
Dr Sanket Hansraj Vaidya	MD, Assistant Professor, Paediatrics, Government Medical College, Alibag, Maharashtra, India
Dr Sourabh Singh	NBEMS Diploma, 1st Year PG Resident, Government Medical College, Alibag, Maharashtra, India

ABSTRACT Pulmonary alveolar proteinosis (PAP) is a rare disorder characterized by intra-alveolar accumulation of surfactant leading to impaired gas exchange and progressive respiratory insufficiency. Genetic causes, particularly ABCA3 mutations, are associated with severe early-onset disease and often poor prognosis. We report a 2-year-6-month-old male who presented with fever, cough, and progressive respiratory distress. High-resolution computed tomography (HRCT) revealed a "crazy paving" pattern suggestive of PAP. Bronchoalveolar lavage findings supported the diagnosis, and whole exome sequencing confirmed an ABCA3 mutation. The child required prolonged oxygen therapy, non-invasive ventilation, and underwent two sessions of total lung lavage. Additional therapies included hydroxychloroquine, corticosteroids, intravenous immunoglobulin (IVIG), and antibiotics. Despite an initially severe clinical course, gradual improvement was observed. The child was successfully weaned off oxygen and discharged. On follow-up, oxygen saturation remains within normal limits with stable respiratory status. This case highlights the heterogeneous clinical course of ABCA3-related PAP and demonstrates that recovery is possible with sustained supportive and interventional management.

KEYWORDS : Pulmonary Alveolar Proteinosis, ABCA3 Mutation, Total Lung Lavage, Pediatric Interstitial Lung Disease, IVIG

INTRODUCTION

Pulmonary alveolar proteinosis is a rare lung disorder characterized by accumulation of surfactant within alveoli, leading to impaired gas exchange. Genetic causes such as ABCA3 mutations are associated with severe disease in early childhood and variable prognosis.



Case Presentation

A 2-year-6-month-old male, born full-term by LSCS with normal birth weight, presented in August 2024 with fever for 4 days, cough, and progressive respiratory distress. He was initially admitted at Civil Hospital, Alibag, where symptomatic treatment and oxygen therapy were started.

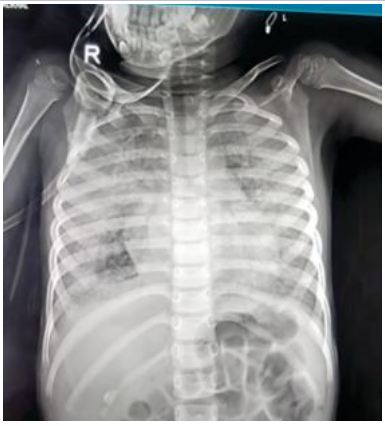
Due to persistent hypoxia, he was referred to MGM Hospital, Panvel, where he remained admitted for one month. During this period, echocardiography was normal, and hydroxychloroquine therapy was initiated.

Owing to continued oxygen dependency, he was referred to KEM Hospital, Mumbai. HRCT chest revealed diffuse ground-glass opacities with interlobular septal thickening forming a "crazy paving" pattern suggestive of pulmonary alveolar proteinosis. Repeat echocardiography showed mild tricuspid regurgitation

The child was further referred to Wadia Hospital for bronchoscopy. Bronchoalveolar lavage yielded milky fluid, and whole exome sequencing confirmed an ABCA3 mutation. Despite undergoing total lung lavage, no immediate improvement was noted.

The child required escalation of respiratory support, including non-invasive ventilation and high-flow oxygen. Repeat bronchoscopy and second lung lavage were performed. BAL culture grew Pseudomonas aeruginosa, treated with appropriate antibiotics. Corticosteroids and 3 doses of IVIG (1g/kg/dose) were also administered.

Gradual improvement was observed, and the child was successfully weaned off oxygen. At discharge, he was stable and maintaining normal oxygen saturation on room air.



1st Chest X-ray on Admission

Chest X-ray on Discharge

Repeat admission after 1 year was done for respiratory complains. Symptomatic management & 2 doses of IVIG(1g/kg/dose) were given. Spontaneous recovery was seen after IVIG administration

CONCLUSION

This case highlights that recovery is possible in ABCA3-related pulmonary alveolar proteinosis despite severe initial presentation. Persistent supportive care, repeated therapeutic interventions, and immunomodulatory therapies like IVIG(1g/kg/dose) may contribute to spontaneous recovery & clinical improvement. Long-term follow-up is essential due to the variable clinical course.

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