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A Genetically Confirmed Case Of Activated PI3Kδ Syndrome Presenting With Recurrent Pneumonia And Chronic Lung Disease

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Abstract
Background:
Activated phosphoinositide 3-kinase delta syndrome (APDS) is a rare autosomal dominant inborn error of immunity caused by gain-of-function variants in PIK3CD or PIK3R1. It is characterised by recurrent infections, immune dysregulation, lymphoproliferation, and progressive end-organ damage, particularly affecting the lungs. Due to its heterogeneous presentation, diagnosis is frequently delayed.
Case Presentation:
We report a genetically confirmed case of APDS in a 16 years old female child who presented with recurrent lower respiratory tract infections since early childhood, progressing to chronic lung disease. The clinical course was marked by multiple hospital admissions for pneumonia, persistent respiratory symptoms, and failure to thrive. Examination and imaging revealed generalised and mesenteric lymphadenopathy with intermittent hepatosplenomegaly, raising initial concern for lymphoproliferative or malignant disorders.
Investigations:
Extensive infectious work-up, including tuberculosis and HIV screening, was negative. Serial haematological evaluations showed fluctuating cytopenia and inflammatory markers. Imaging studies demonstrated abdominal and pelvic lymphadenopathy. Histopathological evaluation of the mesenteric lymph node biopsy revealed reactive lymphoid hyperplasia with preserved architecture and no evidence of lymphoma. Genetic testing using next-generation sequencing identified a heterozygous pathogenic variant in the PIK3CD gene, confirming the diagnosis of APDS.
Management and Outcome:
The patient was managed with immunoglobulin replacement therapy, antimicrobial prophylaxis, and immunomodulation with sirolimus. Therapeutic drug monitoring was performed to maintain optimal sirolimus levels. Following initiation of targeted therapy, there was a reduction in the frequency of respiratory infections, stabilisation of lymphadenopathy, and clinical improvement.
Conclusion:
This case highlights the importance of considering APDS in children with recurrent pneumonia, chronic lung disease, and lymphoproliferation. Early genetic diagnosis allows targeted therapy, prevents unnecessary invasive investigations, and may limit progressive organ damage. Increased awareness of APDS is essential for timely diagnosis and improved outcomes in affected patients.
Keywords: *Activated PI3K delta syndrome, APDS, PIK3CD, recurrent pneumonia, primary immunodeficiency, lymphadenopathy, chronic lung disease*

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