International Journal of Advances in Medicine
Giripunje P et al. Int J Adv Med. 2025 Sep;12(5):502-504
http://www.ijmedicine.com
pISSN 2349-3925 | eISSN 2349-3933
DOI: https://dx.doi.org/10.18203/2349-3933.ijam20252538

Case Report

Diagnostic uncertainty between cystic fibrosis and allergic bronchopulmonary aspergillosis in a young adult male: a case report

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Received: 12 June 2025
Revised: 10 July 2025
Accepted: 14 July 2025

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ABSTRACT

A 23-year-old male presented with persistent respiratory issues and HRCT findings indicating bronchiectasis. Both cystic fibrosis (CF) and allergic bronchopulmonary aspergillosis (ABPA) were considered due to overlapping clinical and radiological features. Notably, elevated Aspergillus-specific IgE levels supported ABPA, while systemic and reproductive features pointed towards CF. This case exemplifies the diagnostic complexity when two conditions with converging presentations must be carefully distinguished.

Keywords: Cystic fibrosis, Allergic bronchopulmonary aspergillosis, Bronchiectasis, Chronic rhinosinusitis

INTRODUCTION

Bronchiectasis in younger adults often warrants an in-depth etiological investigation. Cystic fibrosis (CF), an autosomal recessive disorder involving CFTR gene mutations, is classically diagnosed in childhood but can occasionally manifest later with milder or atypical symptoms. ABPA, a hypersensitivity reaction to *Aspergillus fumigatus*, frequently develops in individuals with asthma or pre-existing CF. Distinguishing between the two is essential, as misdiagnosis may delay appropriate therapeutic intervention and lead to disease progression.[1,2]

CASE REPORT

A 23-year-old male presented with a long-standing productive cough, chronic nasal obstruction, and general fatigue, accompanied by a history of recurrent lower respiratory tract infections since early childhood, with no prior diagnosis of asthma or tuberculosis. Physical examination revealed persistent nasal discharge, turbinate oedema, suboptimal growth, and borderline underweight status. Signs of hypogonadism were noted, including mild bilateral gynecomastia, sparse facial hair, small testicular volume, and delayed pubertal milestones, with a semen analysis confirming azoospermia. These findings suggested features consistent with hypogonadism and possible congenital bilateral absence of the vas deferens (CBAVD), a known manifestation of CF-related male infertility. Diagnostic workups included an HRCT thorax which showed cystic and cylindrical bronchiectasis, along with centrilobular nodules in a "tree-in-bud" pattern across multiple lobes (Figure 1). Serological markers were significant for elevated Aspergillus-specific IgE (109.00 kUA/l) and C-reactive protein (24 mg/l), mild eosinophilia, and a deficient Vitamin D level (20.6 ng/ml). An ENT endoscopy confirmed chronic rhinosinusitis with bilateral mucosal inflammation. All other routine tests, including liver, kidney, thyroid functions, viral markers, abdominal ultrasound, and echocardiogram, were normal.

Differential considerations

Cystic fibrosis

The presence of multi-systemic involvement-including chronic pulmonary disease, upper airway pathology,

vitamin D deficiency, azoospermia, and underdeveloped secondary sexual characteristics-strongly suggests CF. While classic features such as meconium ileus or pancreatic insufficiency were absent, atypical or mild CF phenotypes can present primarily with respiratory and reproductive manifestations in adulthood.[3,4] Diagnostic confirmation through sweat chloride analysis and CFTR mutation testing is recommended.[5]

Allergic bronchopulmonary aspergillosis

The elevated aspergillus-specific IgE, alongside central bronchiectasis and sinus involvement, favours a diagnosis of ABPA. Although asthma was not documented, some ABPA cases have been reported in non-asthmatic individuals, particularly when CF coexists or airway colonization is longstanding.[6]

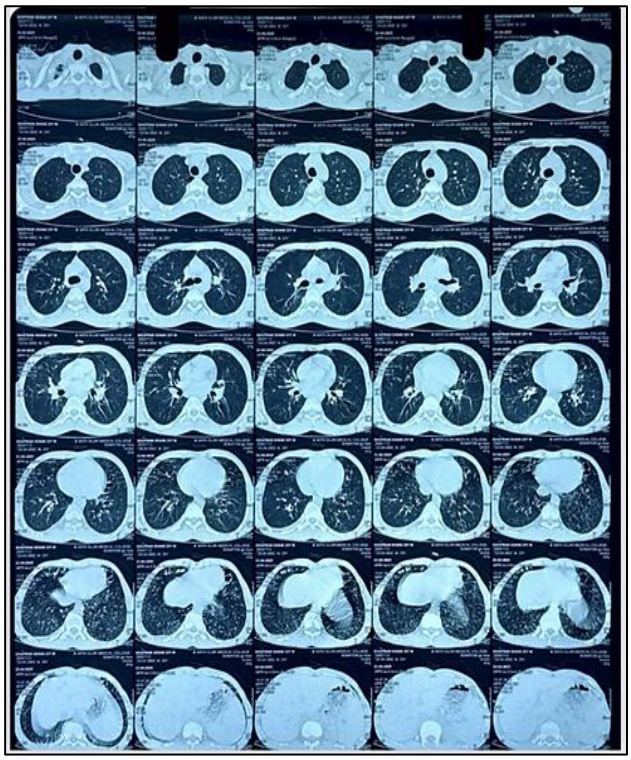

Figure 1: CT chest showing "tree-in-bud" appearance with cystic bronchiectasis.

DISCUSSION

This case represents a diagnostic overlap where two respiratory conditions with similar radiological and clinical features coexist. The markedly raised Aspergillus-specific IgE meets a key diagnostic criterion for ABPA, though the systemic involvement-particularly delayed pubertal development and infertility-supports the diagnosis of CF.

It is important to note that up to 97% of males with CF exhibit azoospermia due to CBAVD, and that hypogonadism is not uncommon in long-standing disease.[7,8] Late diagnoses of CF have been increasingly recognized, particularly in adults with non-classical presentations or monoallelic CFTR mutations.[9] Tools such as nasal potential difference testing or extended CFTR gene panels can help in these ambiguous cases. ABPA and CF may not be mutually exclusive; in fact, ABPA frequently complicates CF and worsens respiratory outcomes. In such cases, management must balance immunosuppressive treatment for ABPA and aggressive airway clearance strategies for CF.

CONCLUSION

This report illustrates the diagnostic complexity in a young adult with respiratory and reproductive abnormalities suggestive of both CF and ABPA. While serological findings support ABPA, clinical evidence leans strongly toward an atypical CF phenotype. A comprehensive diagnostic strategy including sweat testing, CFTR gene analysis, and immunological profiling is essential for timely and accurate diagnosis. Early intervention can significantly impact prognosis and quality of life.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

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Cite this article as: Giripunje P, Singh H, Roy S, Paul UK. Diagnostic uncertainty between cystic fibrosis and allergic bronchopulmonary aspergillosis in a young adult male: a case report. Int J Adv Med 2025;12:502-4.