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Case Report

Immotile Cilia Syndrome-A Rare Case

Manudev Mehta, Mukesh Goyal*, Ramakant Dixit, Pallavi Pillai

Department of Respiratory Medicine, J.L.N Medical College, Ajmer, Rajasthan, India.

Abstract

Immotile cilia syndrome is a hereditary condition resulting in defective ciliary movement or complete ciliary immobility within the airways and other bodily regions. Kartagener syndrome is a specific subset of immotile cilia syndrome, distinguished by situs inversus viscerum. The characteristic triad of Kartagener syndrome includes bronchiectasis, situs inversus, and chronic sinusitis. This report presents an atypical case of immotile cilia syndrome in a female aged 38 years with infertility. The diagnosis was established upon a recurrent respiratory tract infection history and radiographic evidence of maxillary sinusitis, bronchiectasis, dextrocardia, and situs inversus. She had been unable to conceive despite 18 years of marriage, indicating possible infertility due to this. She was treated symptomatically with adequate medical management.

Keywords: Immotile cilia syndrome, Bronchiectasis, Infertility.

Introduction

Immotile cilia syndrome (ICS) is an uncommon genetic disorder distinguished by all cilia in the body being either immotile or exhibiting an abnormal, inefficient beating pattern with prevalence of about 1 in 30,000 to 1 in 60,000 people.[1] It follows an autosomal recessive inheritance pattern. The majority of identified mutations affect the dynein axonemal heavy chain 5 (DNAH5) or dynein axonemal intermediate chain 1 (DNAI1) genes, which are essential components of the outer dynein arms in cilia.[2]

Microscopic (electron) analysis of bronchial and nasal mucosal biopsies from patients has identified distinct ultrastructural abnormalities in respiratory cilia. These include deficiencies in outer or inner dynein arms, or both, as well as radial spoke defects and microtubular transposition anomalies.

Fertility in women with Kartagener's syndrome (primary ciliary dyskinesia) varies, likely due to the impaired motility of the oviductal cilia. Proper ciliary function in the endosalpinx is crucial for successful human reproduction. [3]

This report presents a case of ICS with infertility, representing an atypical manifestation of the condition. Given its rarity and the absence of recent literature on similar cases, this case adds valuable insight into the clinical spectrum of ICS.

Case Report

A 38-year-old female presented with complaints of shortness of breath, serous nasal discharge (mostly seasonal), cough with expectoration, and mild fever for the last month. She had a history of recurrent episodes of similar illness since early childhood. Over the past five years, she was hospitalized 11 times, with the highest frequency of admissions occurring in the last year.

Although no infertility workup was conducted, she had never conceived despite 18 years of marriage without using any measures, suggesting primary infertility.

On physical examination, she was in respiratory distress with a respiratory rate of 28/min with oxygen saturation of 84% on room air with a pulse rate of 116 beats per minute. She had a low BMI of 15 kg/m^2, and was thin-built with adequate nutrition. During respiratory system examination,

Address for correspondence: Mukesh Goyal
Department of Respiratory Medicine, J.L.N Medical College, Ajmer, Rajasthan, India.
E-mail: libra.mukesh@rajasthan.in

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accessory muscles of respiration were working, apex beat was palpated on the right side, at the 5th intercostal space along the midclavicular line. Percussion revealed bilateral resonant percussion note but cardiac dullness toward the right side up to the midclavicular line.

Auscultation revealed bilateral pan-respiratory coarse crepitations at the infrascapular area and heart sounds over the right side.

Chest skiagram showed bilateral bronchiectasis changes at mid and lower zones with cardiac apex on the right side, suggesting dextrocardia and mild cardiomegaly (Figure 1). Skiagram of paranasal sinus in water's view radiographed haziness at both maxillary sinuses, suggestive of maxillary sinusitis (Figure 2). A high-resolution computed tomographic (HRCT) scan of the chest was performed, which revealed central bronchiectasis along with dextrocardia (Figure 3).

Echocardiography confirmed the dextrocardia with enlargement of right heart chambers and raised right ventricular systolic pressure suggestive of mild pulmonary hypertension.

Ultrasonography of the abdomen was done with the liver and spleen positioned on the left side, indicative of situs inversus.

Sputum smear microscopy revealed no acid-fast bacilli and subsequent examination for CBNAAT revealed no mycobacterial tuberculosis detected. The bacterial culture of sputum revealed the growth of *E. coli* with a sensitive pattern of antibiotics. Sputum KOH revealed budding yeast cells. Her blood investigations did not reveal any significant deviation from the normal range. However, the nasal mucociliary clearance test using saccharin, bronchoscopy, and electron microscopic analysis of the nasal mucosa could not be conducted.

She got multiple consultations in the past for her symptoms and was treated with a diagnosis of allergic bronchitis, bronchiectasis, and pneumonia but the diagnosis could not be confirmed. On the basis of her clinical and radiological presentation diagnosis of immotile cilia syndrome was made.

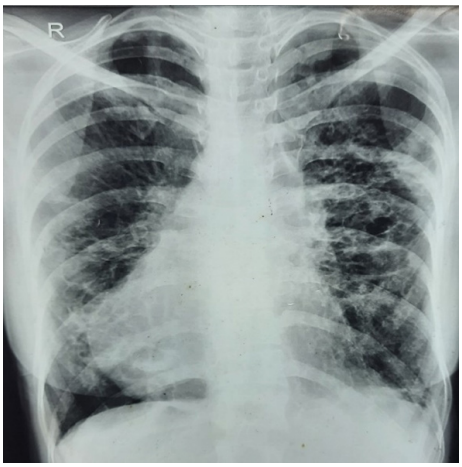

Figure 1: Chest x-ray PA view Showing bilateral bronchiectasis changes along with dextrocardia

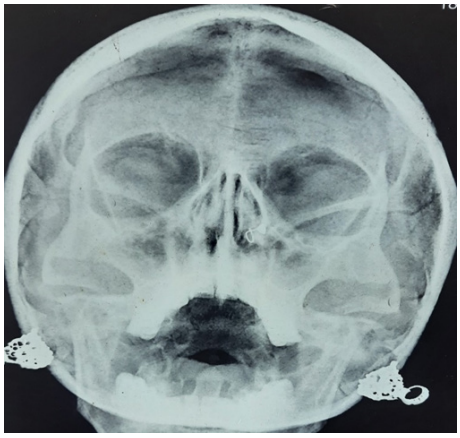

Figure 2: Xray PNS (Water's view) showing bilateral Maxillary sinusitis

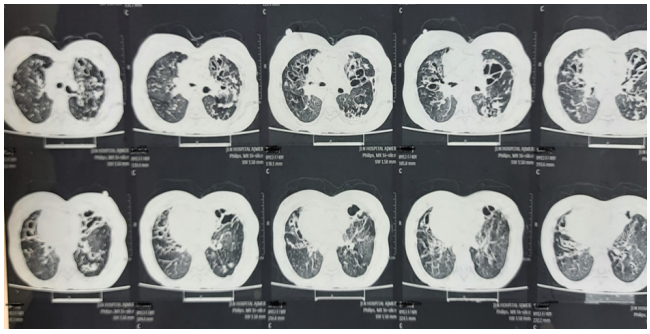

Figure 3: HRCT chest sections showing cystic dilatation of bronchus with thickened bronchial wall suggestive of bronchiectasis at both lungs

She was managed conservatively with adequate medical management with oxygen therapy, broad-spectrum antibiotics as per antibiotic sensitivity and supportive treatment. She was also referred to an infertility specialist.

Discussion

Immotile cilia syndrome is a genetic disorder in which a molecular defect leads to immotile or otherwise dysfunctional cilia. Ciliary motility disorders can be either acquired or inherited, with congenital forms classified as primary ciliary dyskinesias (PCDs). Approximately 50% of these patients exhibit situs inversus. When PCD is accompanied by situs inversus, it is termed Kartagener's syndrome. The characteristic triad of situs inversus, bronchiectasis, and sinusitis have been linked to Kartagener's name after he reported four cases in 1933.[4] Siewart first narrated this syndrome in 1904, still, Kartagener is credited for revealing the etiological relationship between the components of the triad, leading to the syndrome being named after him.[5]

The clinical manifestations of primary ciliary dyskinesia (PCD) can vary widely. Some patients experience neonatal respiratory distress, which may later develop into a persistent

productive cough due to bronchiectasis, treatment-resistant atypical asthma, chronic rhinosinusitis, recurrent otitis media, subfertility, and ectopic pregnancy in females or infertility in males. As the disease progresses, bronchiectasis and obstructive lung impairment become more apparent, with radiographic and clinical signs sometimes detectable as early as preschool age.

The term "primary ciliary dyskinesia" has been largely replaced by "immotile cilia syndrome" (ICS). There have been reports of ICS being linked to various other conditions, including hepatic parenchymal steatosis[6], retinitis pigmentosa caused by defective cilia in the retinal pigment epithelium[7], polysplenia, and extrahepatic biliary atresia[8] suggesting a common dysmorphogenetic process.

In our case, the diagnosis got established due to characteristic clinical presentation and radiological findings. The patient remained asymptomatic during the first decade of life and was later diagnosed with the Kartagener triad, consisting of situs inversus, sinusitis and bronchiectasis. She also had primary infertility, which, combined with the triad, constitutes immotile cilia syndrome. Although male infertility is commonly described in literature[9], infertility among females is less commonly described making this case unique.

Kartagener syndrome and cystic fibrosis are usually diagnosed in neonates or early childhood due to recurrent pulmonary infections. Laboratory screening tests include measuring exhaled levels of nasal nitric oxide and performing the saccharin test to evaluate nasal mucociliary function. The definitive diagnosis is confirmed through electron microscopy, which identifies ultrastructural abnormalities in the ciliary apparatus, which is largely unavailable at most centers.

Early diagnosis and routine follow-ups are essential to prevent complications, as no definitive cure exists for this condition. Delayed detection of bronchiectasis can lead to a poorer prognosis, even with optimal treatment. Complications may severely affect the patient's quality of life. Although no specific treatment can restore normal ciliary function, treatment for ICS focuses on improving lung function and slowing disease progression by airway clearance, chest physical therapy, antibiotics, bronchodilators and inhaled corticosteroids, oxygen therapy, etc and lung transplantation as a last resort.

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