Case Report

Immotile Cilia Syndrome: A case Report

Immotile Cilia Syndrome

Ghulam Mustafa

ABSTRACT

Immotile cilia syndrome is a genetic disorder characterized by defects in the structure and function of cilia, which are hair-like structures that protrude from the surface of cells and play a role in various physiological processes, including the movement of mucus in the respiratory tract. PCD is often autosomal recessive to manifest the disease. The impaired movement of cilia in the respiratory tract can result in the ineffective clearance of mucus and debris. This makes individuals with PCD more susceptible to respiratory infections such as sinusitis, bronchitis, and pneumonia.

Key Words: Bronchiectasis, Immotile cilia syndrome, Kartagener's syndrome, Primary ciliary dyskinesia, Sinusitis, Situs inversus.

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INTRODUCTION

Kartagener's syndrome, also known as primary ciliary dyskinesia (PCD) or immotile cilia syndrome, which is a rare genetic disorder that is characterized by a triad of symptoms: situs inversus, bronchiectasis and chronic sinusitis[1]. The primary cause of Kartagener's syndrome is a defect in the structure or function of cilia. In individuals with Kartagener's syndrome, the cilia do not function properly, leading to the accumulation of mucus and recurrent respiratory infections[2]. Regarding the bronchiectasis in Kartagener's syndrome, there has been debate over whether it is truly congenital (present from birth) or acquired after damaging infections in early life[3]. Some studies suggest that the bronchiectasis may develop over time due to recurrent respiratory infections, while others propose that there may be congenital factors contributing to its development[4].

Eliasson et al[5] have established a solid pathophysiological foundation for this disorder. Electron microscopic examination demonstrates disarray of ciliary microtubules and the absence or partial absence of dynein arms, essential for both ciliary and sperm tail movements.

Situs inversus is indeed caused by defective embryonic organ movement around 10 to 15 days after implantation, it suggests that an error in the normal developmental process leads to the mirror-image arrangement of organs. Bronchiectasis and sinusitis can indeed result from mucus retention and infection.

We describe a case of female child with repeated respiratory distress and chest infection. The development of respiratory distress and chest crepitations in this child may suggest pulmonary complications. The accumulation of pulmonary secretions requiring physiotherapy for removal indicates potential issues with mucus clearance or respiratory function.

METHODS

A female child with history dates back to the age of 30 days of life, when she developed respiratory distress. After that, she often had same complaints for which she took treatment from local doctors till the age of 9 yrs.

1st Admission: During her initial admission to hospital, she presented with symptoms including productive cough, respiratory distress, cyanosis from 1 day, and exhibited signs of tachypnea, clubbing, pectus carinatum, and chest crepitations. Diagnostic workup was done on next day, including a chest X-ray, revealed bronchiectatic changes, and HRCT Chest showed mild interstitial thickening. On the sixth day of admission, a sweat chloride test was conducted, revealing elevated levels of 36 mmol/L; this result prompted a follow-up test performed after more than three months to mitigate the possibility of false-positive outcomes, yet the repeated test still indicated elevated levels, measuring 38 mmol/L. Echocardiography was performed 12th day of admission that revealed levocardia, structurally and functionally normal heart.

2nd Admission: At the time of this admission echocardiography was repeated showing moderate pulmonary hypertension. Subsequent HRCT Chest demonstrated hyperinflated lungs with perihilar bronchiectasis. She was treated with I.V antibiotics

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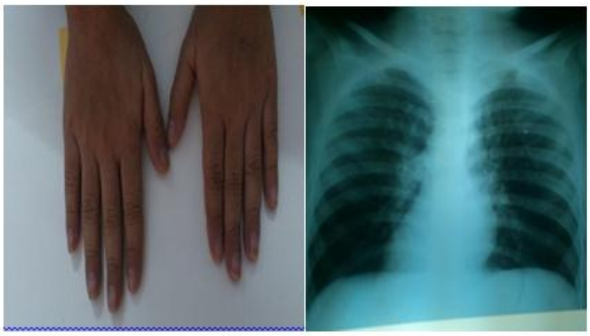
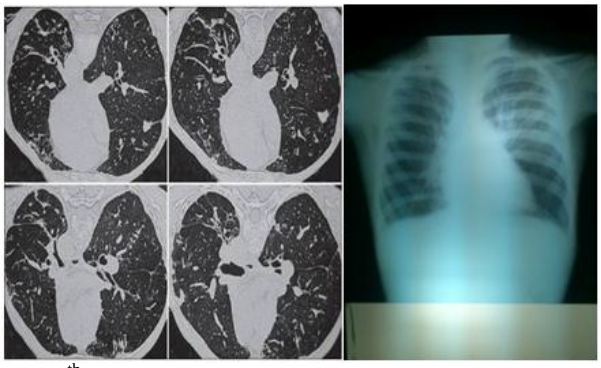
(Ceftriaxone, Cefuroxime), I.V hydrocortisone, nebulization, iron and multivitamin supplementation with chest physiotherapy. She was discharged on oral Cefuroxime, Multivitamin and was advised Chest physiotherapy. For next 16 months, she remained at home with Nebulization and Oxygen inhalation without significant work-up.



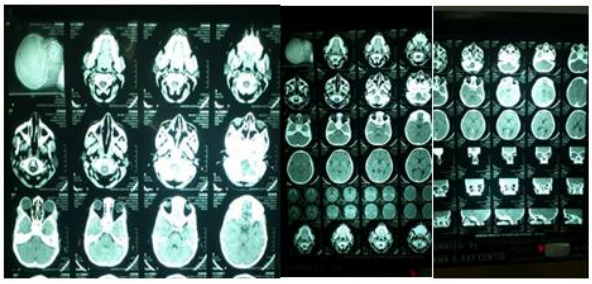
3rd Admission: 3rd time admitted at Hospital, with same complaints of productive cough and respiratory distress. Chest x ray showed hyperinflated lungs with perihilar opacities. HRCT Chest revealed bilateral hyperinflated lungs having bronchiectasis with peribronchial cuffing, areas of air-trapping, patch of fibrosis. On 20th day of admission 99m MAA Lung Perfusion Scan was performed showed multiple large segmental and sub-segmental perfusion defects in both lung field more on right lower lobe. Repeat echocardiography showed mild pulmonary hypertension. Immunoglobulin (IgG) levels were 13.5 g/L (5.4-18.2 g/L), Echocardiography showed borderline RV function, severe pulmonary hypertension, pulmonary artery systolic pressure 70 mmHg. During this admission she was given intravenous (IV) Ceftazidime and Amoxi-clav, Oral Co-Trimoxazole, Nebulization with Tobramycin, Normal saline, Hypertonic saline, Ipratropium and Beclomethasone, chest physiotherapy and on 27th day she was Oxygen free and afebrile.

After one-month Saccharine test (A bedside test to diagnose ciliary dyskinesia) was performed. A drop of saccharine was put in nose just in front of inferior meatus and the patient was asked to tell when she felt the taste of saccharine. The time for patient was 5 hours, not felt the taste for 1st test and on repeat test respectively while for control it was 5 min in each case. She was discharged on Tab. Co- Trimoxazole, Tab. Prednisolone, Nebulization with Ipratropium and Beclomethasone along with chest physiotherapy.

4th Admission: She was admitted at Hospital again. Rhino scintigraphy Scan was performed that didn't show prompt transit of tracer into nasopharynx with nasal mucociliary transport rate (NMTR) of 1.9 mm/min in left nostril and 2.18 mm/min in right nostrils (Normal Range of ~7mm/min). There is scintigraphic evidence of delayed mucociliary transport in both nostrils. This finding confirmed our diagnosis of Immotile Cilia Syndrome.



On 10th day of admission CT Scan PNS was performed that showed bilateral maxillary and ethmoidal sinusitis with non-pneumatization of bilateral frontal sinuses. After two days of CT PNS Delta F508 mutations showed both the alleles negative for CFTR gene mutations and patient was discharged on oral Amoxi-clav, Prednisolone, and Salbutamol.



DISCUSSION

Cilia line the epithelium in various anatomical structures, including the trachea, bronchi, nasopharynx, Eustachian tubes, fallopian tubes, and cerebral ventricles; the immotile cilia syndrome, characterized by immotile sperm due to dynein arm defects, is associated with impaired sperm tail movement, while defective embryonic cilia are believed to contribute to the random lateralization of viscera, resulting in a distribution of individuals with either levocardia or dextrocardia[6].Our ultrastructural findings closely resemble those described by an author in the context of immotile cilia syndrome. A study reported a case with the onset of cyanosis and respiratory distress early within few days of life. Another described a case with symptoms appearing at 2 days of life[6-8].

These cases highlight the variability in the presentation of Kartagener's syndrome in the neonatal period, with symptoms ranging from respiratory distress to nasal discharge and otitis media. The common thread in these cases is the presence of situs inversus and respiratory difficulties, which are key features of the syndrome. In this case we used saccharin test for final diagnosis of ciliary dyskinesia and mucus clearance. Riechmann et al[9] also described this test and concluded that the saccharin test, a cost-effective and straightforward procedure, is a clinically valuable method for assessing mucociliary clearance in older children and adults; a

normal test, when properly conducted, effectively rules out the diagnosis and eliminates the necessity for more intricate testing.

Early diagnosis is important to implement aggressive airway clearance and antibiotic measures, preventing the onset of bronchiectasis, and ensuring avoidance of inappropriate ear, nose, and throat procedures. The diagnostic process typically commences with functional studies, commonly involving the direct measurement of ciliary beat frequency on nasal epithelial cells. Subsequently, ciliary ultrastructure is examined through electron microscopy to further corroborate the diagnosis.

CONCLUSION

This case highlights a challenging diagnostic journey with recurrent respiratory distress ultimately diagnosed as Immotile Cilia Syndrome, emphasizing the importance of a comprehensive diagnostic workup in complex respiratory cases.

Author's Contribution:

Concept & Design of Study:	Ghulam Mustafa
Drafting:	Ghulam Mustafa
Data Analysis:	Ghulam Mustafa
Revisiting Critically:	Ghulam Mustafa
Final Approval of version:	Ghulam Mustafa

Conflict of Interest: The study has no conflict of interest to declare by any author.

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