An Unusual Presentation of Primary Ciliary Dyskinesia: A Case Report on Kartagener Syndrome

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ABSTRACT

Primary ciliary dyskinesia (PCD) is an autosomal recessive condition characterised by abnormal ciliary motion, leading to impaired mucus clearance resulting in recurrent sinopulmonary infections, otitis media, and infertility. Kartagener syndrome (KS), a subset of PCD, presents with situs inversus, bronchiectasis, and chronic paranasal sinusitis. We describe a case of a young female exhibiting clinical and radiological features indicative of KS, including high-grade fever, cough with sputum, and shortness of breath. The patient was promptly managed for suspected and later culture-proven pseudomonas pneumonia. This case underscores the significance of early diagnosis, given that previous general practitioners had treated the patient without clear clinical or radiological evidence of the condition's nature and aetiology.

Key Words: *Primary ciliary dyskinesia, Kartagener syndrome, Pneumonia.*

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INTRODUCTION

Kartagener syndrome (KS), first described as a clinical syndrome by Manes Kartagener in 1933, falls under the category of primary ciliary dyskinesia (PCD), hallmarked by the presence of a clinical triad of chronic paranasal sinusitis, bronchiectasis, and situs inversus. This genetically heterogeneous disorder is inherited as an autosomal recessive disorder and is characterised by an altered motion of the cilia in the ciliated epithelium of the respiratory tract, paranasal sinuses, and middle ear.[1] Visceral rotation of the organs during embryonic development depends on normal ciliary beating which otherwise causes situs inversus.[2] Poor mucociliary clearance manifests in early childhood as recurrent sinusitis, pneumonia, and otitis media. Prevention of respiratory dysfunction is the prime goal of the management governed by antibiotics and symptomatic care.

CASE REPORT

A young female, in her first decade of life, presented in the outpatient department of a teaching hospital with high-grade fever, cough with greenish sputum, and difficulty in breathing for the past three days.

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She had a history of recurrent episodes of fever with facial pain. She also gave a history of episodic cough with sputum and shortness of breath beginning from early childhood for which she was treated by the local practitioners. On presentation, her blood pressure was 100/60 mmHg, pulse rate was 110 bpm, respiratory rate was 27 breaths/min and temperature was 101˚F. Physical examination was unremarkable except for mild conjunctival pallor and finger clubbing. On auscultation, wheezes were audible in both lung fields, and the apex beat was best heard at the right fifth intercostal space in the mid-clavicular line.

Routine blood tests were suggestive of neutrophilic leucocytosis with reactive thrombocytosis. Microcytic hypochromic anaemia was present. Liver and renal function tests were within limits. Arterial blood gas analysis revealed uncompensated respiratory alkalosis. Chest X-ray was advised which showed dextrocardia (Figure 1). High-resolution computed tomography (HRCT) of the chest was suggestive of bronchiectatic changes in both lungs (Figure 2). Ultrasound abdomen revealed liver on the left and spleen on the right side. Sputum for acid-fast bacilli (AFB) was negative. The patient was treated empirically with antibiotics targeted against *pseudomonas aeruginosa* as suggested by the history which was later confirmed by the culture of sputum. The patient was labelled as a case of KS due to strong evidence of history, and clinical and radiological workup which was later confirmed by nasal mucosal biopsy in which electron microscopic analysis of the cells from nasal scrapings showed decreased dynein arms in the respiratory epithelial cilia.

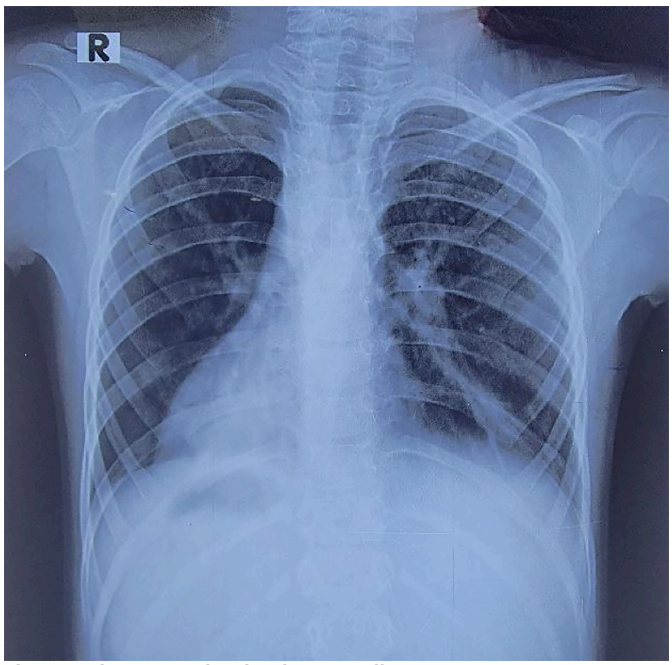

Figure 1: Chest X-ray showing dextrocardia.

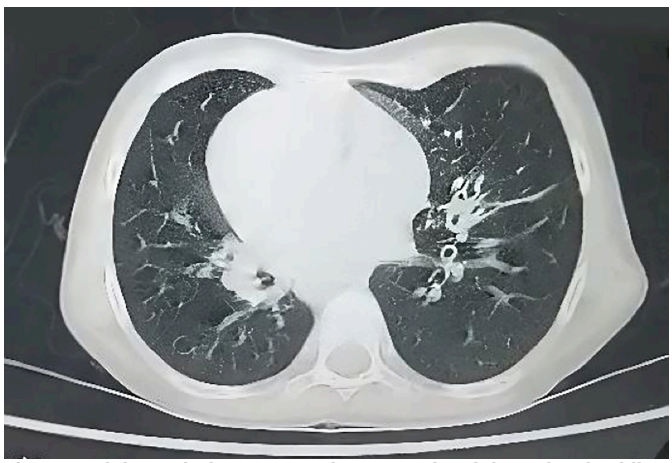

Figure 2: High-resolution computed tomography of chest showing bilateral bronchiectatic changes.

DISCUSSION

PCD is an autosomal recessive condition caused by the mutations in the genes, DNAH5 and DNAI1, located on chromosomes 5p15.2 and 9p21-p13, respectively.[3] This results in altered ciliary structure and function, impairing the beating of cilia and mucociliary clearing. The most common defect is a decrease in the number of dynein arms which adversely impacts the frequency of ciliary beating.[4] This results in chronic childhood infections which vary in the severity of clinical symptoms.

PCD when associated with chronic sinusitis, bronchiectasis, and situs inversus is termed KS. Patients often complain of long-standing, viscous, and mucoid nasal discharge from early childhood. Examination usually reveals edematous nasal mucosa that is pale. There is also an altered sense of smell. The recurrent sinusitis presents with fever and facial pain mostly at maxillary and periorbital regions. Recurrent middle ear infection is also a common presentation. Retracted tympanic membrane with poor motility and effusion can be found on the middle ear examination.

Chronic bronchitis, recurrent pneumonia, and bronchiectasis in KS are often caused by pseudomonal infection. Patients can present with high-grade fever and cough with sputum in copious amounts. Examination may reveal increased tactile fremitus, crackles, rhonchi, and occasionally wheezing. Obstructive lung disease may occur due to increased levels of inflammatory mediators in a long-standing affected airway. Cardiothoracic and abdominal examination and imaging reveal the reverse orientation of thoracic and abdominal organs called situs inversus.

A diagnostic workup should be soon initiated when history and clinical examinations give clues to the diagnosis.[5] Initial workup includes sinus and chest radiographs which typically demonstrate thickened, opacified sinus cavities and dextrocardia with lower lobe bronchiectatic changes. However, HRCT of chest is more sensitive in diagnosing the abnormalities in the airway and pulmonary parenchyma. Other less commonly done tests include the saccharine test which is done by placing saccharine in the nostril, and then measuring the velocity of transport into the nasopharynx, and estimating exhaled nitric oxide which is typically decreased in the presence of abnormal ciliary function.

Antibiotics are the mainstay of treatment but prophylactic antibiotics should be used with caution due to emerging antibiotic resistance in the paediatric population. Antibiotics should always be given in the light of bacterial culture and sensitivity, although some studies suggest long-term low-dose preventive antibiotics. Symptomatic relief can be given by inhaled bronchodilators and mucolytics if required.[6] Patients should be advised to have the pneumococcal vaccine and the influenza vaccine every year, in addition to the conventional childhood immunisations. Caregivers should be emphasised for regular hospital visits which should include spirometry, sputum culture, and imaging studies when needed. Well-managed patients usually become less problematic in their second decade of life and most of them live a healthy adult life. A bilateral lung transplant is only considered when medical management fails to improve the quality of life.[7]

In conclusion, this case underscores the need for early diagnosis and prompt treatment to improve patient outcomes.

PATIENT'S CONSENT:
Informed consent was obtained from the patient's parents.

COMPETING INTEREST:
The authors declared no competing interest.

AUTHORS' CONTRIBUTION:
NIB, AU, SK, ZT, OA: All the authors contributed to the design, drafting and critical revision of the manuscript, and approved the final version of the manuscript to be published.

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