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Case Report

Kartagener's syndrome - A case report

K.N. Sricharan,[1] Sayd Mohammed Rajeeb,[1] Praveen Kumar John,[2] S. Rajesh[1]

[1]Department of General Medicine, A.J. Institute of Medical Sciences, Mangalore, India
[2]Department of Radiology, A.J. Institute of Medical Sciences, Mangalore, India

Abstract

Kartagener's syndrome is a rare, ciliopathic, autosomal recessive genetic disorder that causes a defect in the action of the cilia lining the respiratory tract. Patients usually present with chronic recurrent rhinosinusitis, otitis media, pneumonia, and bronchiectasis caused by pseudomonal infection. Situs inversus can be seen in about 50% of cases. Diagnosis can be made by tests to prove impaired cilia function, biopsy, and genetic studies. Treatment is supportive. In severe cases, the prognosis can be fatal. We present a case of a 24-year-old man, who presented with acute respiratory symptoms. He was diagnosed with Kartagener's syndrome based on his clinical presentation, laboratory and radiological findings.

Keywords: Dextrocardia; Genetic disorder; Kartagener's syndrome

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1. Introduction

Kartagener's syndrome is a rare, ciliopathic, autosomal recessive genetic disorder that is characterized by dextrocardia, bronchiectasis, otitis media and rhionsinusitis.[1,2,3] Basic problem is defective movement of the cilia. Males are generally infertile because of immotile sperms. Diagnosis can be made by tests to prove impaired cilia function, biopsy, and genetic studies. However, cases of semi-sterility in females have been reported.[4] Prompt detection and treatment of Kartagener's syndrome can prevent its various complications.[5] We present a case of 24 year old male patient with Kartagener's syndrome.

2. Case report

A 24 year old male patient presented with complaints of chest pain, breathing difficulty, fever and recurrent episodes of sinusitis. There was no history of allergy, atopy, bronchial asthma or lower respiratory infections. On general examination his vitals were: heart rate 120 beats/minute; respiratory rate-30 breaths/min; oxygen saturation 91% on 5 L of oxygen; blood pressure 150/90 mmHg; and temperature of 102.2 F. Physical examination revealed patient in moderate respiratory distress with diffuse ronchi and diminished air movement in both lungs. Other physical examination findings were unremarkable. His blood work up was unremarkable except for mild leucocytosis. Patient was admitted in medical intensive care unit (MICU). Chest X-ray revealed features suggestive of situs inversus and haziness with bilateral basal bronchiectatic changes. Ultrasonography of the abdomen showed situs inverus totalis. High resolution CT of the thorax

(Fig.1) revealed bilateral basal bronchiectasis with secondary infection. He was diagnosed with Kartagener's syndrome based on his clinical presentation, laboratory evaluation and radiological features. Saccharin clearance test was done and consistent with Kartagener's syndrome.[6] CT of paranasal sinuses (Fig.2) revealed thickening of paranasal mucosa which suggested chronic sinusitis. Semen analysis revealed non motile spermatozoa and oligospermia. He was started on IV antibiotics and supportive medications. Patient's condition improved and was shifted out of ICU by the fifth day of presentation and discharged from hospital after one week.

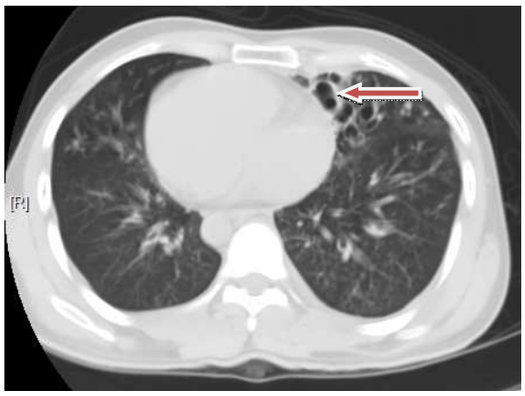

Fig.1: High resolution CT Thorax-Bronchiectatic changes

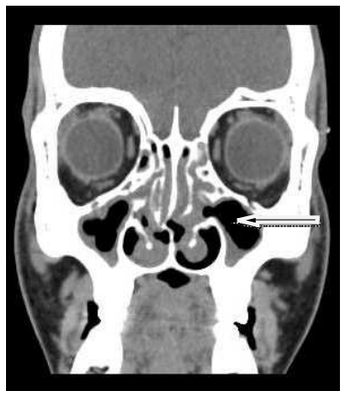

Fig.2: CT Paranasal sinuses-Mucosal thickening

3. Discussion

Kartagener in 1933 described a unique syndrome characterized by the triad of situs inversus, chronic sinusitis and bronchiectasis now dubbed as Kartagener's syndrome.[4] Later patients with this condition were noted to have defects in the ultra structure of cilia. Based on this feature Afzelius coined the term immotile cilia.[10] Later studies showed that disorganized motion, rather than immotile cilia, resulted in the uncordinated and ineffective ciliary beat, hence coined the term ciliary dyskinesia syndrome.[10] Primary ciliary dyskinesia is an autosomal recessive disease with extensive genetic heterogenecity characterized by abnormal ciliary motion and impaired mucociliary clearance. Ultrastructural and functional defects of cilia result in the lack of affective ciliary motility causing abnormal mucociliary clearance. This leads to recurrent and or persistent respiratory infections, sinusitis, otitis media, and male infertility. In 46% of the patients it is associated with situs inversus.[4]

Primary ciliary dyskiesia is an autosomal recessive heterogenous group of conditions with variable clinical findings. The disease phenotype is caused by defects of respiratory cilia, spermatids and cilia of embryonic mode.

Currently three genes (DNAI1, DNAH5 and DNAH11) that encode for dynein proteins (axonemal and cytoplasmic) have been linked to recessive primary ciliary dyskinesia.[8] It is hypothezised that given the small number of well characterized affected families, the large size of the dynein genes, the different dynein proteins present in the axomere and the large number of regulatory structural proteins necessary for ciliaray function, dynein mutation may not be the only cause of primary ciliary dyskinesia. Since its initial description in 1933, Kartagener's

syndrome (total situs inversus, bronchiectasis and either nasal polyp or recurrent sinusitis) and the description by Afzelius in 1975 of the defects in the dynein arm underline this condition, incomplete forms of this syndrome have increasingly been recognized. Thus clinical findings may be varied in patients with primary ciliary dykinesia, including respiratory distress in neonates, recurrent respiratory tract infection, bronchiectasis, situs inversus, infertility and hydrocephalus, singly or in various combinations. The study by Noone et al. looking at 94 patients from 68 families showed that cough was present in 100% of patients, bronchiectasis (98%), sinusitis (47%), otitis media (92%) and situs inversus (46%).[7] Treatment of Kartagener's syndrome includes daily chest physiotherapy, antibiotics with good anti pseudomonal coverage,and supportive pulmonary care. Surgical intervention for bronchiectasis is rarely recommended, but can be beneficial when the disease is localized.[9]

Differential diagnoses for immotile-cilia syndrome include malignancy, interstitial lung diseases including idiopathic pulmonary fibrosis and idiopathic interstitial pneumonia and other conditions associated with bronchiectasis which include acquired (foreign body aspiration, tumor, lymphadenopathy, chronic obstructive pulmonary disease,and mucoid impaction) and congenital bronchial obstruction (bronchomalacia, pulmonary sequestration, and yellow nail syndrome), recurrent infection (immunodeficiencies), abnormal secretion disorder (cystic fibrosis), and other miscellaneous conditions (alpha-1 antitrypsin deficiency and connective tissue disease).[10] Primary ciliary dyskinesia (Kartagener's syndrome) is in itself a rare clinical entity, which we thought worth of sharing among medical personnel. High index of suspicion will lead to prompt diagnosis. Earlier goal directed treatment can prevent fatal complications.

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Corresponding author

Dr. Sayd Mohamed Rajeeb
Postgraduate cum Tutor
Department of General Medicine
A.J.Institute of Medical Sciences
Mangalore, India
Mobile: +91 7760063897
E-mail: rajeeb80@yahoo.com