Kartagener’s Syndrome: A Triad of Bronchiectasis, Situs Inversus, and Chronic Sinusitis


Abstract

Primary ciliary dyskinesia (PCD) is a syndrome characterised by productive cough with bronchiectasis and sinusitis in early life, and reduced fertility later in life. PCD is a rare syndrome with an estimated incidence of 1: 20,000 to 30,000. Primary ciliary dyskinesia is a genetic disorder with manifestations present from early life and distinguishes it from acquired mucociliary disorders. Here we present one such case of Kartagener’s syndrome that presented with bronchiectasis, recurrent sinusitis with primary infertility, and situs inversus, which fits in the triad of Kartagener’s syndrome.

Key words: Primary ciliary dyskinesia, Primary infertility, Acquired mucociliary disorders.

Case report

A 45-years-old female presented with complaints of productive cough with profuse yellowish expectoration with postural variation for 15 days, alongwith low grade fever since 1 month. She had previous episodes of recurrent rhinosinusitis and profuse expectoration for many years. The patient had been married for 25 years and she was infertile with regular menstrual periods suggesting primary infertility.

On examination, the patient had temperature of 100° F, normal pulse and blood pressure with respiratory rate of 20/min. Apex beat was localised to right fifth intercostal space on mid-clavicular line; and on percussion, liver dullness was on left side – all this suggestive of situs inversus. On auscultating the respiratory system, there were bilateral, lower zone, coarse leathery crepitations present on inspiration as well as expiration.

On investigation, the patient had haemoglobin of 12 gm/dl and total leucocyte count of 12,000/cmm of blood, and ESR of 60 mm in 1st hour. Routine biochemical tests did not reveal any significant abnormality. Sputum culture was negative for acid-fast bacilli, but showed Gram-positive cocci. Chest radiography confirmed dextrocardia with the aortic arch lying on right side of the trachea with cystic bronchiectatic changes seen in both lower zones and mid-zones (Figure 1). HRCT (High resolution computed tomography) of chest showed bronchiectatic changes in left upper lobe, lingula, and right middle lobe, with few small areas of consolidation in both lower lobes and apical segments (Figure 2). USG abdomen showed liver and inferior vena cava on left side; and spleen, stomach, and aorta on right side. X-ray PNS showed changes of chronic maxillary sinusitis bilaterally.

The patient was investigated for infertility in the form of:
1. USG abdomen and pelvis
2. Laparoscopic examination of pelvis

Fig. 1: Chest radiography showing dextrocardia with aortic arch lying on right side of trachea with cystic bronchiectatic changes.
3. Hormonal assay for FSH, LH, and prolactin levels.

All reports were normal.

Pulmonary function tests (PFT) showed FEV₁/FVC to be 62% of predicted values, suggestive of obstructive pattern with minimal improvement in PFT after bronchodilators. The 'nasal saccharin transit time' test gave mucociliary clearance time of 60 minutes (Normal < 30 minutes)¹. However, due to lack of the facility of electron microscopy of the nasal cilia, we were not able to study the ultrastructural defect.

Thus, Kartagener's syndrome was diagnosed clinically. She was treated with intravenous antibiotics and mucolytic agents with chest physiotherapy. The majority of symptoms rapidly improved and she was discharged on low dose antibiotics in rotation.

Discussion

The triad of bronchiectasis, sinusitis, and situs inversus was first described by Siewert in 1903, although its usual eponym is Kartagener's syndrome – after the Swiss paediatrician who described four cases with similar features in 1933². By 1960, 300 cases had been reported and concept of a disease with congenital and generalised non-functioning of the cilia evolved. About 50% of the people affected with primary ciliary dyskinesia have situs inversus, so fit in the criteria of Kartagener's syndrome.

The clinical features of PCD have been described in primary ultrastructural defects in cilia. Ultrastructurally, cilia and spermatozoa tail are similar. The axoneme is the key component of the cytoskeleton and has a characteristic nine plus two arrays of microtubules (Figure 3). The nexin links and spokes seem to provide structural rigidity to the axoneme. Dynein arms extend from one side of a doublet in a clockwise direction, when viewed from the tip of the cilium. They contain most of the ATPase activity of the axoneme, and are important in releasing energy for sliding and bending of microtubules and ciliary motion³.

The tracheobronchial tree is ciliated to the level of the respiratory bronchioles, each ciliated cell having about 200 cilia. Mucociliary transport in the respiratory tract is important for normal respiratory function and resistance to respiratory infection⁴.

The typical clinical picture of PCD is a chronic productive cough which can usually be traced back to early childhood or infancy, chronic rhinitis often with nasal polyposis, chronic or recurrent maxillary sinusitis, and frequent ear infections in childhood. Bronchiectasis is not present at birth, but may develop early, sometimes even in childhood. The most common respiratory pathogens are Haemophilus Influenzae and Streptococcus pneumoniae. Most males are sterile, but many females have a lowered fertility. About 50% of patients have situs inversus viscerum⁵.

Fig. 2: HRCT of chest showing bronchiectatic changes in left upper lobe and lingula, and right middle lobe, with few small areas of consolidation in both lower lobes and apical segment.

Fig. 3: Cilia axoneme diagram.
Patients with suspected primary ciliary dyskinesia should have their mucociliary clearance measured, and the cilia should be examined by microscopy. Nasal mucociliary clearance can be measured by the ‘nasal saccharin transit time’ test in which a saccharin pellet is placed on the anterior end of the inferior turbinate and the time taken for the subject to notice the taste is recorded. This test requires the patient's co-operation and is not reliable in children under 10 years of age. Nasal cilia are easily accessible, and can be obtained from the inferior turbinate without anaesthesia by a non-invasive brush technique. Ciliary beat frequency can then be assessed by light microscopy and photometric techniques, and cilia fixed on electron microscopy. In males, the motility of sperm can be examined, and electron microscopy may show the characteristic ultrastructural defects.

Rationale of treatment

Treatment of PCD is aimed at relieving symptoms and preventing complications. Early recognition of the disease and prompt antibiotic treatment are the keys to minimise the irreversible lung damage. Physiotherapy with postural drainage and cessation of smoking are also important. Coughing should not be suppressed since it acts as a substitute for mucociliary clearance. Huffing from mid to low lung volume with forced expiratory manoeuvre helps to improve clearance.

Despite this being a chronic respiratory disease, life span seems to be normal. Infertile patients benefit from advanced micromanipulation techniques that allow non-motile or poorly motile sperm to penetrate, or by in vitro fertilisation techniques.

Thus, we should remember that any patient with a history of recurrent cough and cold, and bronchiectasis with infertility should be examined for Kartagener’s syndrome which is a part of PCD.

References