

Kartagener Syndrome: Congenital Variety of Primary Ciliary Dyskinesia with Infertility

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Abstract

We report a rare case of Kartagener's Syndrome, congenital variety of Primary Ciliary Dyskinesia (PCD) with infertility. The patient exhibited the classical triad of which was elucidated by Manes Kartagener in 1933. The frequency of KS in the United States is 1 case per 32,000 live births. Situs inversus occurs randomly in half the patients with PCD; therefore, for every patient with KS, another patient has PCD but not situs inversus. Current nomenclature classifies all congenital ciliary disorders as PCDs in order to differentiate them from acquired types. KS is part of the larger group of disorders referred to as PCDs. Approximately one half of patients with PCD have situs inversus and, thus, are classified as having KS.

Keywords: Immotile cilia syndrome; Primary ciliary dyskinesia (PCD); Situs inversus; Chronic sinusitis; Bronchiectasis.

1. Introduction

The condition now known as Primary Ciliary Dyskinesia (PCD), a case of which was reported by Siewert was first properly recognised by Kartagener. Subsequent work by Afzelius¹ demonstrated that patients with Kartagener's syndrome had a motility defect in the cilia of respiratory mucosa, in the lungs and sinuses, and that in addition in males there can also be a defect of sperm motility², which results in reduced fertility³. Defective ciliary movement due to primary ciliary dyskinesia can result in impairment of pulmonary defence mechanism⁴⁻⁵ with immunological deficiency, poor bacterial clearance recurrent infection of broncho-pulmonary tree, bronchiectasis and/or cystic fibrosis. There could be association of motility dysfunction in form of poor spermatozoal movement and male sterility.⁶ Siewert first described the combination of situs inversus, chronic sinusitis and bronchiectasis in 1904. However, Manes Kartagener first recognized this clinical triad as a distinct congenital syndrome in 1933.⁷ The syndrome can be diagnosed clinically and has significant prognostic value in management of bronchiectasis associated with infertility due to ciliary dyskinesia and proper counseling assisted reproductive technique can change the prognosis of such cases.

2. Case History

Our patient was 32 year old male farmer, referred from Department of Chest and Tuberculosis, with c/o recurrent episodes of cough with expectoration low grade fever and breathlessness on moderate exertion. He had two episodes of hemoptysis which subsided with treatment of antibiotics from local practitioner. He gave history of ear discharge similar episodic attack during his early childhood. His clinical symptomatology was aggravated with episodes of hemoptysis since 2 months. He was married, living with his wife but has no issue for last 17 years. His present admission revealed a young patient of moderate build, early clubbing, mild pallor no cyanosis or lymphadenopathy. His vitals were normal with normal pulsations. He is normotensive with normal appearance of chest wall. Examination revealed absent apex beat on left side. Auscultation revealed coarse rales bilateral with loud pulmonary sound second (P2). Liver was palpable on left hypochondrium. No lump on right side. His laboratory reports were normal but for mild leukocytosis and raised ESR sputum examination did not reveal AFB and culture for other microbial were negative. His liver function tests including serum amylase and lipase were normal. His semen analysis revealed normal counts but defective sperm motility.

3. His radiological work up included

Sinus radiographs typically demonstrate mucosal thickening, opacified sinus cavities, and hypoplastic frontal sinuses. Chest radiographs (Fig. 1) illustrated bronchial wall thickening as manifestation of chronic infection, bronchiectasis and situs inversus (in 50% of patients with PCD). The presence of situs inversus strongly suggests KS. Bronchiectasis was seen in the lower lobes in patients with KS and immunoglobulin deficiency, while bronchiectasis predominantly occurs in the upper lobes of patients with cystic fibrosis.

High-resolution CT scan of the chest extended to upper abdomen was performed and showed typical lower lobe bronchiectasis with secondary infection and bronchial wall thickening, extended scan showed situs inversus totalis. (Fig. 2). An additional Barium meal was done (not required but for academic interest) showed the stomach on the right side.

4. Discussion

Kartagener Syndrome is an autosomal recessive disorder primarily manifesting as ciliary movement disorder. The frequency is 1 case per 32000 live births and Situs inversus totalis occurs randomly in half patients with primary ciliary disorder.⁸ However primary ciliary dyskinesia is responsible for 5 to 10% of cases of bronchiectasis.⁹

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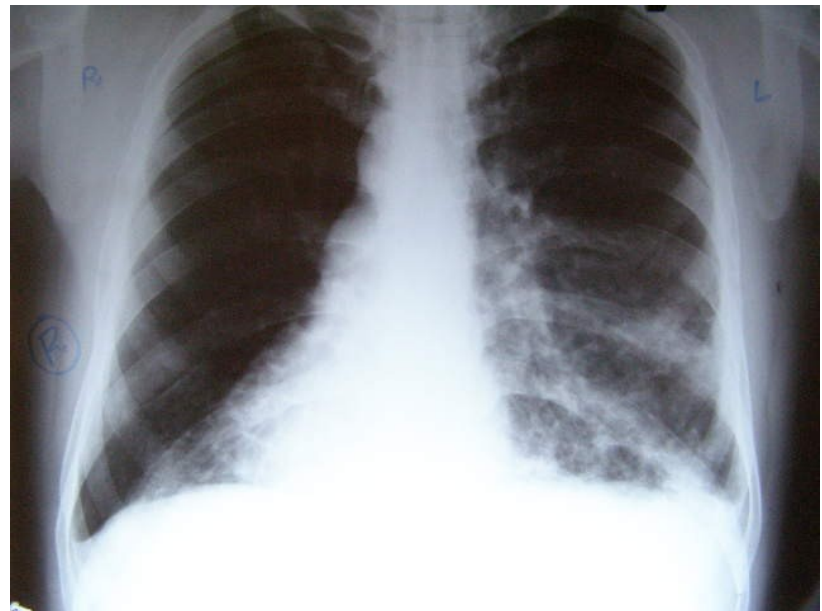


Figure 1: Chest radiograph illustrated bronchial wall thickening as manifestation of chronic infection and bronchiectasis



Figure 2: High-resolution CT scan of the chest showed typical lower lobe bronchiectasis with secondary infection and bronchial wall thickening

Numerous defects are encompassed under this syndrome including abnormalities of dyenin arms,¹⁰⁻¹¹ radial spokes and microtubules.⁶⁻¹² The cilia becomes dyskinetic their co-ordinated propulsive action is diminished and natural defence mechanism for microbial is impaired.¹³ This leads to recurrent upper and lower respiratory tract infection¹⁴ such as sinusitis, otitis media¹⁵ and bronchiectasis. Since sperm motility depends upon proper functioning of ciliary movements there is defective sperm motility and thus leading to infertility.¹⁶ The early recognition of clinical syndrome in a patient of upper respiratory tract with bronchiectasis should raise a suspicion especially in the background of infertility. The condition can be diagnosed clinically and corroborated and confirmed radiologically and hence treated accordingly.¹⁷ Attempts have been made to treat infertile couples through ART and especially Intracytoplasmic injection (ICSI) of processed spermatozoa.¹⁸

Imaging Studies which are required to corroborate the classical triad of KS are sinus radiographs, chest radiographs, high-resolution CT scan of the chest is the most sensitive modality for documenting early and subtle abnormalities within airways and pulmonary parenchyma when compared to routine chest radiographs. Consideration should be given to this imaging technique early in the presentation of PCD syndromes, when a chest radiograph may not be sensitive enough to identify disease processes or when another differential is being considered.

5. References

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