

A Case Report of Kartagener's Syndrome

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Abstract: Kartagener syndrome is a triad of situs inversus, bronchiectasis, and either nasal polyps or recurrent sinusitis. It is a rare condition and a delay in its diagnosis or wrong diagnosis can lead to serious respiratory complications. This case report is about an 18-year-old patient who was having recurrent upper and lower respiratory tract infection since childhood, treated as same. We noticed prominence of heart sound on right side instead of normally on left side, so we evaluate patient with interest and found a rare condition as its cause i.e. Kartagener's syndrome. Also recognition of situs inversus is important for preventing surgical mishaps that result from the failure to recognize reversed anatomy or an atypical history.

Key words: Kartagener syndrome, situs inversus, bronchiectasis, nasal polyps, recurrent sinusitis.

I. INTRODUCTION

Primary ciliary dyskinesia (PCD), also called immotile ciliary syndrome or Kartagener syndrome, is a rare, ciliopathic, autosomal recessive genetic disorder that causes defects in the action of cilia lining the respiratory tract (lower and upper, sinuses, Eustachian tube, middle ear), fallopian tube, and flagella of sperm cells.¹⁻³ The phrase "immotile ciliary syndrome" is no longer favored as the cilia do have movement, but are merely inefficient or unsynchronized.⁴ Respiratory epithelial motile cilia, which resemble microscopic "hairs" (although structurally and biologically unrelated to hair), are complex organelles that beat synchronously in the respiratory tract, moving mucus toward the throat.⁵ Normally, cilia beat 7 to 22 times per second, and any impairment can result in poor mucociliary clearance, with subsequent upper and lower respiratory infection. Cilia also are involved in other biological processes (such as nitric oxide production), which are currently the subject of dozens of research efforts.⁶ As the functions of cilia become better understood, the understanding of PCD should be expected to advance. When accompanied by the combination of situs inversus (reversal of the internal organs), chronic sinusitis, and bronchiectasis, it is known as Kartagener syndrome (only 50% of primary ciliary dyskinesia cases include situs inversus). It is a genetically heterogeneous disorder affecting motile cilia which are made up of approximately 250 proteins.⁷ Around 90% of individuals with PCD have ultrastructural defects affecting protein(s) in the outer and/or inner dynein arms which give cilia their motility, with roughly 38% of these defects caused by mutations on two genes, DNAI1 and DNAH5, both of which code for proteins found in the ciliary outer dynein arm.⁸⁻¹⁰

II. CASE REPORT

18-year-old male patient presented in OPD with recurrent history of common cold and headache since childhood with episodic fever, cough with expectoration and worsening of symptoms. For that he consulted many physicians and investigated repeatedly for blood investigations, chest x-ray and sputum examination. His record revealed that he received several courses of antibiotics, antihistamines, and even anti-tuberculous drugs but response was only partial and temporary. In examination we noticed that heart sound are more prominent on right side, instead of normally on left side, even though trachea is central. Also cardiac impulse was located in 5th inter costal space, medial to mid-clavicular line on right side, tidal percussion for liver was evident in 5th inter costal space on left side and Traube's area was situated on right side. For confirmation of finding we repeat chest x-ray with proper labeling of side, advised ultrasound of abdomen and electrocardiogram. In new x-ray (figure-1) findings from previous x-rays were reconfirmed, ultrasound of abdomen showed spleen on right side of abdomen while liver on left side suggestive of situs inversus. ECG showed QS waves and inverted T waves in lead-1 along with other features of dextrocardia (figure-2). So diagnosis of Dextrocardia with Situs inversus was made. As the patient was of dextrocardia with situs inversus and have repeated history of upper and lower respiratory tract infection, we evaluated patient for Kartagener's syndrome. We advised him sinus radiograph and HRCT of chest. Semen analysis for sperm motility was not done as the patient declined for it. Sinus radiograph showed mucosal thickening, opacified cavities and other feature of chronic sinusitis. HRCT of chest showed bronchiectasis in bilateral lower lobes, more on left side (figure-3). Considering the clinical picture of sinusitis, bronchiectasis, and situs inversus the clinical diagnosis of Kartagener's syndrome was made. The

condition was explained to the patient and he was treated with antibiotics, antipyretics, mucolytics and regular chest physio-therapy.

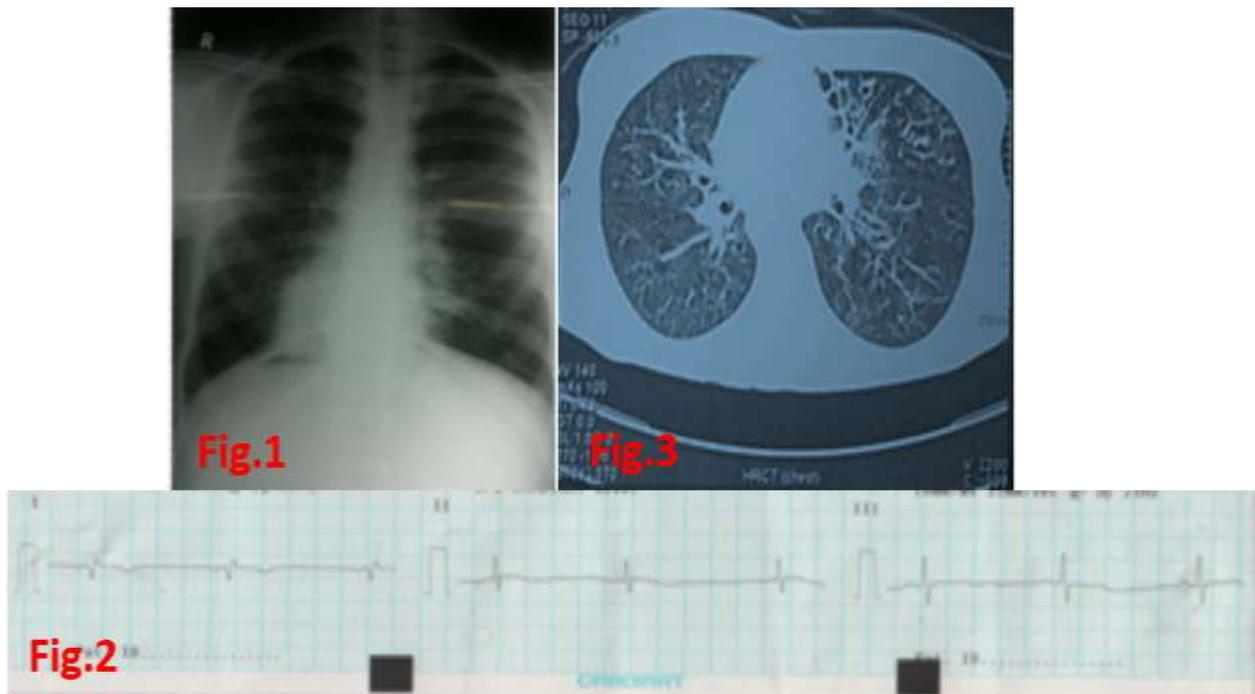


Figure 1: Cxr showing dextrocardia and fundic gas under right dome of diaphragm.

Figure 2: QS with inverted T wave in Lead 1.

Figure 3: HRCT of chest showing bilateral bronchiectasis more on left.

III. DISCUSSION

Numerous defects have been described in both the conditions including abnormalities in dynein arms, radial spokes and microtubules. As a result of these abnormalities cilia become dyskinetic and their propulsive action is diminished.¹¹⁻¹³ This leads to impaired bacterial clearance resulting in recurrent lower and upper respiratory tract infections. Also because sperm motility depends on proper ciliary movement, males with this condition are generally infertile.¹⁴ Additionally since visceral rotation during development depends upon ciliary motion, situs inversus is seen in Kartagener's syndrome.¹⁵ Demonstration of abnormal ciliary movement needs electron microscopic studies of biopsies obtained from nasal mucosa or trachea. However these procedures are invasive and available at specialized centres only.¹⁶ Diagnosis of this condition is usually clinical accompanied by imaging studies. A 1998 review noted that life expectancy is usually normal, but that there have occasionally been reported neonatal deaths due to PCD. A longitudinal study followed 151 adults with PCD for a median of 7 years. Within that span, 7 persons died with a median age of 65.^{17,18} The classic symptom combination associated with PCD was first described in 1904, while Kartagener published his first report on the subject in 1933.¹⁹⁻²¹ There are no standardized effective treatment strategies for the condition. Severe fatal respiratory failure can develop; long-term treatment with macrolides such as clarithromycin, erythromycin and azithromycin has been empirically applied for the treatment of primary ciliary dyskinesia in Japan, though controversial due to the effects of the medications.²²⁻²⁵

IV. RATIONALE OF TREATMENT

Treatment of PCD is aimed at relieving symptoms and preventing complications.²⁶ Early recognition of the disease and prompt antibiotic treatment are the keys to minimise the irreversible lung damage.²⁷⁻²⁸ 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.²⁹⁻³¹ 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.³²⁻³⁴ 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.³⁵

V. CONCLUSION

Kartagener Syndrome is a rare condition which in many a times remains wrongly diagnosed or undiagnosed. Many a times patient undergoes a battery of investigations and unnecessary drugs including Anti Tubercular Drugs as in this case. In a Country like India where tuberculosis has a high burden such irrational use of anti-tubercular drugs only on the basis of cough with expectoration and fever can result in Prospective Multi Drug Resistant Tuberculosis cases. So prompt and proper Diagnosis of Kartagener syndrome is of utmost importance.

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