

*Case Report*

Kartagener's Syndrome: A Case Report

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ABSTRACT

Kartagener's syndrome is part of the larger group of disorders referred to as primary ciliary dyskinesias. Approximately one half of patients with primary ciliary dyskinesia have situs inversus and, thus, are classified as having Kartagener syndrome. Kartagener syndrome is characterized by the clinical triad of chronic sinusitis, bronchiectasis, and situs inversus. Here we report a case of 18 years old girl with all typical features of Kartagener's syndrome

Key-words: Kartagener's syndrome, chronic sinusitis, bronchiectasis, situs inversus

INTRODUCTION

The frequency of Kartagener syndrome is 1 case per 32,000 live births. It is a triad of Chronic Sinusitis, Situs Inversus and Bronchiectasis. It is a syndrome due to primary ciliary dyskinesia, a rare autosomal recessive disorder because of impaired ciliary movement. Recurrent upper and lower respiratory infection like otitis media, sinusitis, pneumonia leading to bronchiectasis occur due to impaired bacterial clearance. Second important feature of this syndrome is Situs Inversus – as visceral rotation during development depends upon proper ciliary motion, the positioning of normally lateralized organs become random.

CASE HISTORY

An 18 years old thin built girl came to emergency room with severe shortness of breath with history of recurrent upper & lower respiratory tract infection since childhood. On examination she had Pulse-80 / min, BP-110/70 mm Hg, RR-34/min. Systemic examination revealed bilateral basal crepitations. His apex beat was not palpable on usual site but at right 5th ICS, clubbing was present. On investigation her ECG showed dextrocardia(Figure.1), Chest X-RAY revealed bilateral basal bronchiectatic changes with situs inversus (Figure.2). CT scans Chest revealed extensive cystic bronchiectatic changes and complete situs inversus(Figure.3). X-ray PNS showed chronic sinusitis. The

combination of her symptoms & investigation made us to think for kartagener's syndrome. She was treated with intravenous antibiotics, bronchodilators and

mucolytic agents. She responded to treatment and discharged in stable condition after 7 days. She was advised regular follow up for monitoring.

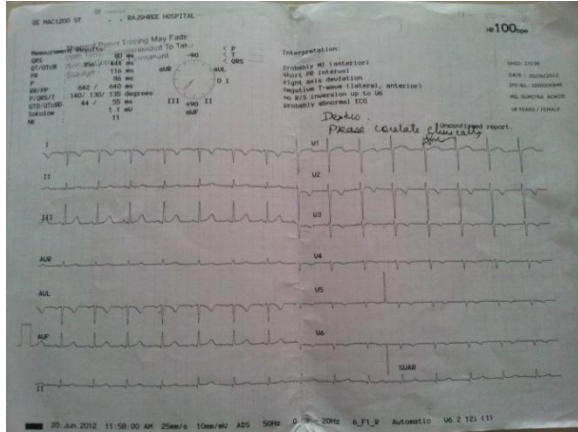


Figure.1: ECG suggestive of Dextrocardia.



Figure.2: Chest X-ray suggestive of bronchiectasis.

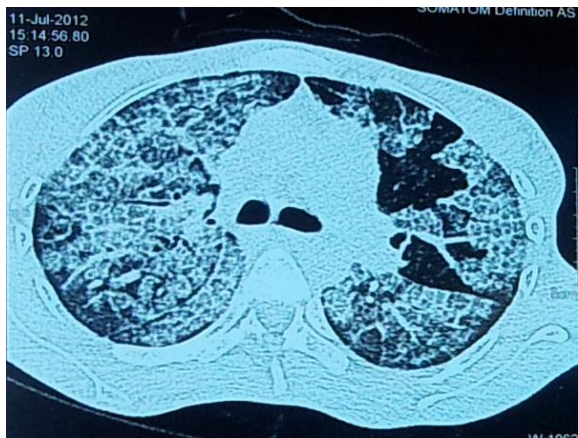


Figure.3: CT Scan chest suggestive of bilateral bronchiectasis.

DISCUSSION

Siewert first described the combination of situs inversus, chronic sinusitis, and bronchiectasis in 1904. However, Manes Kartagener^[1] first recognized this clinical triad as a distinct congenital syndrome in 1933. Because Kartagener described this syndrome in detail, it bears his name. Kartagener syndrome is inherited via an autosomal recessive pattern. Symptoms result from defective cilia motility. Camner and co-workers^[2] first suggested ciliary dyskinesia as the cause of Kartagener syndrome in

1975. They described 2 patients with Kartagener syndrome who had immotile cilia and immotile spermatozoa. These patients had poor mucociliary clearance because the cilia that lined their upper airways were not functioning.

In 1981, Rossman and co-workers^[3] coined the term primary ciliary dyskinesia (PCD) because some patients with Kartagener syndrome had cilia that were not immobile but exhibited an uncoordinated and inefficient movement pattern. Current nomenclature classifies all congenital ciliary disorders as primary ciliary dyskinesias in order to differentiate them from acquired types. Kartagener syndrome is part of the larger group of disorders referred to as primary ciliary dyskinesias. Approximately one half of patients with primary ciliary dyskinesia have situs inversus and, thus, are classified as having Kartagener syndrome. Afzelius proposed that normal ciliary beating is necessary for visceral rotation during embryonic development. In patients with primary ciliary dyskinesia, organ rotation occurs as a random event; therefore, half the

patients have situs inversus and the other half have normal situs.

Patients with primary ciliary dyskinesia exhibit a wide range of defects in ciliary ultrastructure and motility, which ultimately impairs ciliary beating and mucociliary clearance. The most common defect, first described by Afzelius, is a reduction in the number of dynein arms, which decreases the ciliary beat frequency.^[4]

Sturgess et al^[5] described how the radial spoke, which serves to translate outer microtubular sliding into cilial bending, was absent in some patients with primary ciliary dyskinesia. Cilia in other patients lacked central tubules; however, instead of the central tubules, an outer microtubular doublet transposed to the cell of the axoneme was present that displayed an abnormal 8+1 doublet-to-tubule pattern. Both the radial spoke and the transposed doublet defects impaired mucociliary clearance. Clinical manifestations include chronic upper and lower respiratory tract disease resulting from ineffective mucociliary clearance. Males demonstrate infertility secondary to immotile spermatozoa. Other features include digital clubbing and diminished female fertility. Primary ciliary dyskinesia has been associated with esophageal problems and congenital cardiac abnormalities. The majority of patients are seen by a physician more than 50 times before the diagnosis is made at an average age of 10-14 years.^[6]

The cause of primary ciliary dyskinesia is genetic, with an autosomal recessive inheritance pattern. Genome analysis has found primary ciliary dyskinesia to be genetically heterogenous. Genes *DNAH5* and *DNAI1* on bands 5p15.1 and 9p13, 3 respectively, are known to cause primary ciliary dyskinesias.^[7] Management is prevention of recurrent respiratory infection and treatment of infection with proper antibacterial therapy. If unilateral

involvement of lung is present sometime surgery is indicated.

CONCLUSION

Kartagener's syndrome can be a devastating illness for the thousands of patients who develop this condition each year. Clinicians should consider workup for Kartagener's syndrome in patients with recurrent respiratory tract infections since childhood. Early diagnosis and appropriate monitoring of Kartagener's syndrome may decrease complications.

REFERENCES

1. Kartagener M. Zur pathogenese der bronchiectasien. I Mitteilung: bronchiectasien bei situs viscerum inversus. *Betr Klin Tuberk.* 1933; 83:498-501.
2. Camner P, Mossberg B, Afzelius BA. Evidence of congenitally nonfunctioning cilia in the tracheobronchial tract in two subjects. *Am Rev Respir Dis.* 1975; 112:807-9.
3. Rossman CM, Forrest JB, Lee RM, Newhouse MT. The dyskinetic cilia syndrome. Ciliary motility in immotile cilia syndrome. *Chest.* 1980; 78:580-2.
4. Afzelius BA. A human syndrome caused by immotile cilia. *Science.* 1976; 193:317-9.
5. Sturgess JM, Chao J, Wong J, Aspin N, Turner JA. Cilia with defective radial spokes: a cause of human respiratory disease. *N Engl J Med.* 1979; 300:53-6.
6. Sommer JU, Schäfer K, Omran H, Olbrich H, Wallmeier J, Blum A, et al. ENT manifestations in patients with primary ciliary dyskinesia: prevalence and significance of otorhinolaryngologic

comorbidities. *Eur Arch
Otorhinolaryng.* 2011; 268:383-388
7. Bush A, Chodhari R, Collins N,
Copeland F, Hall P, Harcourt J.

Primary ciliary dyskinesia: current
state of the art. *Arch Dis Child.*
2007; 92:1136-40.

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