

# Kartagener's Syndrome - A Case Report

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**Abstract:** *Kartagener's syndrome is a rare, autosomal recessive genetic ciliary disorder comprising the triad of situs inversus, chronic sinusitis, and bronchiectasis. The basic problem lies in the defective movement of cilia, leading to recurrent chest infections, ear/nose/throat symptoms, and infertility. We present a case of a 24-year-old female, who presented with recurrent episodes of cough and breathlessness. She was diagnosed with Kartagener's syndrome based on her clinical presentation and radiological findings.*

## “1. Introduction”

Kartagener's syndrome (KS) is a subset of a larger group of ciliary motility disorders called primary ciliary dyskinesias (PCDs). It is a genetic condition with an autosomal recessive inheritance,<sup>1,2</sup> comprising a triad of situs inversus, bronchiectasis and sinusitis. During the embryonic stage, organ position is determined by uniform ciliary beating but in Kartagener's Syndrome, due to ciliary dysmotility heart along with the other organs fail to move on to the left side, resulting in dextrocardia and situs inversus. Although Siewart first described this condition in 1904, it was Kartagener who recognized the etiological correlation between the elements of the triad and reported four cases in 1933.<sup>2</sup> The estimated prevalence of PCD is about 1 in 30,000<sup>3</sup>, though it may range from 1 in 12,500 to 1 in 50,000<sup>1</sup>. In KS, the ultrastructural genetic defect leads to impaired ciliary motility which causes recurrent chest, ear/nose/throat, sinus infections, and infertility. A high index of suspicion is needed to make an early diagnosis so that timely treatment options may be offered for infertility in these young patients, wherever feasible. Also, although unproven, it seems likely that early diagnosis is important for the preservation of pulmonary function, quality of life, and life expectancy in this disease<sup>4,5</sup>.

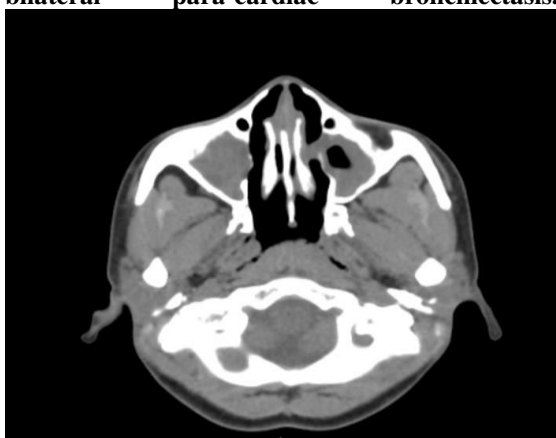
## “2. Case Report”

A 24 year old female came to medicine OPD with complaints of recurrent cough with copious

expectoration and breathlessness for 1 month. Cough and breathlessness were not associated with any postural or diurnal variation. She used to suffer from frequent cough and cold since childhood. No family history of asthma or atopy and recurrent respiratory infection were present. She is unmarried and is having normal menstrual history. Clinical examination revealed normal physical development with mild pallor. Local examination of the paranasal sinus shows tenderness over the bilateral frontal and maxillary sinuses. Systemic<sup>th</sup> examination shows apical impulse heard at right 5<sup>th</sup> inter costal space at midclavicular line instead on left side. On chest percussion cardiac dullness was elicited in right precordium. Hepatic dullness was elicited on the left side with a tympanic note on the right side. On auscultation, fine coarse crepitation heard over bilateral infra mammary, infra axillary and infra scapular area with heart sounds being best heard on the right hemi thorax. Her blood work up was unremarkable except for mild leukocytosis and sputum microscopy for AFB was negative. Chest X-ray postero-anterior (PA) view [Figure 1] revealed cardiac apex and aortic arch on the right side, suggesting dextrocardia and bilateral para cardiac cystic bronchiectatic changes and the hepatic shadow was seen on the left side and gastric fundus on the right side suggestive of situs inversus totalis. An ultrasound of the abdomen revealed a normal liver and gall bladder on the left side and a normal spleen on the right side. CT Paranasal sinuses [Figure 2] shows diffuse mucosal thickening of the all the paranasal sinuses suggestive of pan-sinusitis. CT-scan thorax plain [Figure 3] was done which showed features of dextrocardia with right sided aortic arch and bilateral cystic and tubular bronchiectasis. Upper abdomen cuts section [Figure 4] shows complete situs inversus with hepato-biliary system on the left side and spleen, gastric fundus and splenic flexure on right side.



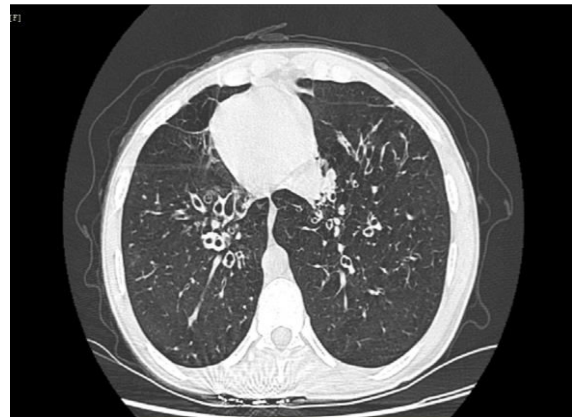
**Fig 1. Chest X-Ray Shows dextrocardia with bilateral para-cardiac bronchiectasis.**



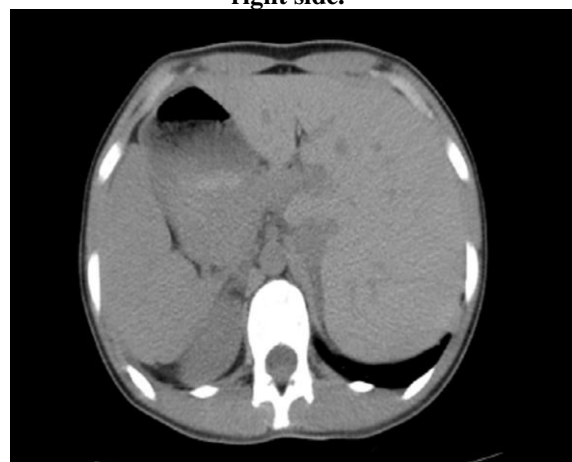
**Fig 2. CT Paranasal sinus axial section shows diffuse soft tissue thickening of bilateral maxillary sinuses.**

### “3. Discussion”

Disorders of ciliary motility may be congenital or acquired. Congenital disorders are labeled as Primary ciliary dyskinesia (PCDs). Nearly 50% of PCD patients have situs inversus. Such cases of PCD with situs inversus are known as Kartagener's syndrome<sup>6</sup>. PCD is phenotypically and genetically heterogeneous condition wherein the primary defect is in the ultrastructure or function of cilia<sup>7,8</sup>. Such defects are identified in approximately 90%<sup>9</sup> of PCD patients and involve the outer dynein arms, inner dynein arms, or both. 38%<sup>9</sup> of the PCD patients carry mutations of the dynein genes *DNAI10* and *DNAH5*<sup>6</sup>. Pathophysiologically, the underlying defect which leads to accumulation of secretions and consequent recurrent sinusitis, bronchiectasis, infertility, and situs inversus is the defective ciliary motility/immotility. The severity of symptoms and the age at which the condition is diagnosed is quite variable, even though the symptoms are present from birth.<sup>11,12</sup>



**Fig 3. HRCT Thorax shows bronchiectasis in both lower lobes with cardiac shadow seen on right side.**



**Fig 4. CT upper abdominal section shows liver on left side, spleen and stomach on right side.**

Patients with Kartagener's syndrome may have either situs solitus i.e. dextrocardia only or situs inversus totalis where all the viscera are on the opposite side, including left sided appendix (Jonsson *et al.*, 1982).

A recent study showed that heterotaxy (situs ambiguous or laterality defect other than situs inversus totalis) occurs with a prevalence of at least 6% and can be associated with complex congenital heart disease in PCD. Kartagener's syndrome can have variable presentations and severity due to its multisystem involvement and reversal of viscera orientation. Having situs inversus totalis, will present with left sided appendicitis if they develop this problem at some stage in their lives. Females can present with subfertility due to sluggish movement of ova in the fallopian tubes, while males demonstrate infertility secondary to immotile spermatozoa.

Diagnostic criteria for this condition include<sup>13</sup> clinical picture suggestive of recurrent chest infections, bronchitis, and rhinitis since childhood, along with one or more of the following: (1) situs

inversus in the patient/sibling; (2) alive but immotile spermatozoa; (3) reduced or absent transbronchial mucociliary clearance; and (4) cilia showing characteristic ultrastructural defect on electron microscopy. Apart from fulfilling the criteria mentioned above, two types of tests are done for diagnosis of PCD – screening tests (exhaled nasal nitric oxide measurement which is usually low in PCD, and saccharin test to assess mucociliary function of nasal epithelium) and diagnostic tests (ciliary beat pattern and frequency analysis using video recording, and electron microscopic confirmation of the ultrastructural ciliary defect). The samples for these tests for examining motility and ultrastructure of cilia may be obtained by biopsy of nasal mucosa and laparoscopic biopsies of tubal mucosa in females, as was done by Halbert *et al.*<sup>14</sup>

There are currently no reliable noninvasive diagnostic methods for this disease and the correct diagnosis is often delayed by years, causing irreversible pulmonary damage with subsequent morbidity. There was a 98% reduction of level of nasal nitric oxide concentration in patients with Kartagener syndrome compared to age-matched controls suggesting that nitric oxide measurements could be of help in the early diagnosis and hence management of the disease process. Early diagnosis and treatment is important to prevent long term sequelae and morbidity associated with it. Genetic counselling, social, psychological and fertility issues should be addressed once it is diagnosed and help these patients to live with. Kartagener's syndrome without morbidity and in a dignified way. Differential diagnosis is from cystic fibrosis (an inherited disorder) and, Young's syndrome, in addition to features of Kartagener's syndrome, has azoospermia. Treatment of this rare congenital disorder includes antibiotics, intravenous or oral, intermittent or continuous, and are used to treat upper and lower airway infections. Hemophilus influenzae and *Staphylococcus aureus* are the most common organisms (Bent and Olearczyk, 2007). Long-term low-dose prophylactic antibiotics may be necessary in children. Obstructive lung disease/bronchiectasis should be treated with inhaled bronchodilators, mucolytics, and chest physiotherapy. Influenza and pneumococcal vaccination should be encouraged. Lung transplantation and heart-lung transplantation have occasionally been tried in severe cases with some success (Otgün *et al.*, 2004; and Alvarez *et al.*, 2005).

To conclude, 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.

#### 4. References

1. Barthwal MS. Kartagener's syndrome in a fertile male - An uncommon variant. *Lung India.* 2006;23:123–5.
2. Dixit R, Dixit K, Jindal S, Shah KV. An unusual presentation of immotile-cilia syndrome with azoospermia: Case report and literature review. *Lung India.* 2009;26:142–5.
3. Seaton D. Bronchiectasis. In: Seaton A, & D, Leitch AG, editors. *Crofton & Douglas's respiratory diseases.* 5ed. Oxford: Blackwell Science ; 2004. pp794–828.
4. Barbato A, Frischer T, Kuehni CE, Snijders D, Azevedo I, Baktai G, et al. Primary ciliary dyskinesia: A consensus statement on diagnostic and treatment approaches in children. *Eur Respir J.* 2009;34:1264–76.
5. Marthin JK, Petersen N, Skovgaard LT, Nielsen KG. Lung function in patients with primary ciliary dyskinesia: A cross-sectional and 3-decade longitudinal study. *Am J Respir Crit Care Med.* 2010;181:1262–8.
6. Olbrich H, Häffner K, Kispert A, Völkel A, Volz A, Sasmaz G, et al. Mutations in DNAH5 cause primary ciliary dyskinesia and randomization of left-right asymmetry. *Nat Genet.* 2002;30:143–4.
7. Noone PG, Bali D, Carson JL, Sannuti A, Gipson CL, Ostrowski LE, et al. Discordant organ laterality in monozygotic twins with primary ciliary dyskinesia. *Am J Med Genet.* 1999;82:155–60.
8. Chodhari R, Mitchison HM, Meeks M. Cilia, primary ciliary dyskinesia and molecular genetics. *Paediatric Respir Rev.* 2004;5:69–76.
9. Zariwala MA, Knowles MR, Omran H. Genetic defects in ciliary structure and function. *Annu Rev Physiol.* 2007;69:423–50.
10. Loges NT, Olbrich H, Fenske L, Mussaffi H, Horvath J, Fliegauf M, et al. DNAI2 mutations cause primary ciliary dyskinesia with defects in the outer dynein arm. *Am J Hum Genet.* 2008;83:547–58.
11. Kordus RJ, Price RL, Davis JM, Whitman-Elia GF. Successful twin birth following blastocyst culture of embryos derived from the immotile ejaculated spermatozoa from a patient with primary ciliary dyskinesia: A case report. *J Assist Reprod Genet.* 2008;25:437–43.
12. Coren ME, Meeks M, Morrison I, Buchdahl RM, Bush A. Primary ciliary dyskinesia: Age at diagnosis and symptom history. *Acta Paediatr.* 2002;91:667–9.
13. Afzelius BA, Mossberg B. Immotile cilia. *Thorax.* 1980;35:401–4.
14. Halbert SA, Patton DL, Zarutskie PW, Soules MR. Function and structure of cilia in the fallopian tube of an infertile woman with Kartagener's syndrome. *Hum Reprod.* 1997;12:55–8.