Case Report:

Kartagener's syndrome: Case Report

*Dr. Amit A. Palange, Dr. Mohd. Shahid, Dr. Manjit S. Sisode, Dr. Bhumika Vaishnav

Department of Medicine, P.Dr.D.Y.Ptln Medical College, Pimpri, Pune, India.

*Corresponding author*: Email: amitapalange@yahoo.com

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 hereby reported a case of this rare entity – an infertile female with bronchiectasis and chronic sinusitis. The need for a high index of suspicion to make an early diagnosis cannot be overemphasized in such patients so that wherever possible, options for timely treatment of infertility may be offered and unnecessary evaluation of symptoms is avoided.

**KEY WORDS:** Bronchiectasis, Kartagener's syndrome, sinusitis, situs inversus.

**BACKGROUND:**

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. Comprising a triad of situs inversus, bronchiectasis and sinusitis. 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. The estimated prevalence of PCD is about 1 in 30,000, though it may range from 1 in 12,500 to 1 in 50,000. In KS, the ultrastructural genetic defect leads to impaired ciliary motility which causes recurrent chest, ear/nose/throat (ENT), and 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. However, this has not been confirmed and further large prospective studies are needed.

**CASE REPORT**

We reported a case of a 22 year old married female patient presented in outpatient department with chief complaints of recurrent episodes of common cold, sneezing, and cough with expectoration for past 4-5 years. The patient also revealed that she frequently developed cough, cold, rhinorrhea, nasal blockade, and ear discharge during childhood. She also gives history of not having children despite being married for last 4 years. On cardiovascular examination, dextrocardia was suspected. On auscultation, bilateral wheeze and right basal crackles were audible, with heart sounds being best heard on the right side of the chest. Electrocardiogram showed evidence of dextrocardia. Chest X-ray postero-anterior (PA) view revealed cardiac apex and aortic arch on the right side, suggesting dextrocardia. An ultrasound of the abdomen revealed a normal liver and gall bladder on the left side and a normal spleen on the right side. Contrast-enhanced computed tomography (CECT) chest revealed dextrocardia, nodular opacities in bilateral lung fields suggestive of bronchiectasis.
DISCUSSION

Disorders of ciliary motility may be congenital or acquired. Congenital disorders are labeled as PCDs. Nearly 50% of PCD patients have situs inversus. Such cases of PCD with situs inversus are known as Kartagener's syndrome. PCD is a phenotypically and genetically heterogeneous condition wherein the primary defect is in the ultrastructure or function of cilia. Such defects are identified in approximately 90% of PCD patients and involve the outer dynein arms, inner dynein arms, or both. 38% of the PCD patients carry mutations of the dynein genes DNAI and DNAH5. 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. Occasionally, Kartagener's syndrome may be associated with reversible airflow obstruction. Clinical progression of the disease is variable with lung transplantation required in severe cases. Diagnostic criteria for this condition include 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. In our cases, however, we could not perform these tests and the diagnosis was essentially clinico-radiological.
The issue of fertility was not addressed in the initial published reports of patients with KS until Arge reported three male patients with this syndrome having immotile spermatozoa and sterility. Male patients with KS invariably present infertility, while women present reduced fertility. Infertility in male KS patients is due to diminished sperm motility, while in females it is due to defective ovum transport because of dyskinetic motion of oviductal cilia, suggesting that the ciliated endosalpinx is essential for human reproduction. The development of assisted reproductive techniques has allowed rational treatment for these patients, and to date, there have been reported pregnancies using subzonal insemination (SUZI) and intracytoplasmic sperm injection (ICSI). Until more is known with regard to the genetic control of PCD, it is suggested that treatment should be individualized depending on sperm motility. In cases where there is no sperm motility, ICSI may be the most appropriate treatment. However, if sperm motility is present, a trial of in vitro fertilization (IVF) should be considered. One concern regarding the fertility treatment of men with PCD is the possibility that the resultant child has the risk of being affected by the same condition. To conclude, KS patients are frequently troubled by repeated infection episodes for which they have to seek medical attention and this is largely the reason for their morbidity. But infertility is also one important aspect that needs to be adequately addressed in their evaluation so that they may be offered a suitable option that could help them have children.

REFERENCES