An Unusual Presentation of Kartagener’s Syndrome
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Abstract
Kartagener’s syndrome is a rare disorder which is seen in nearly half of the cases of primary ciliary dyskinesia. We report an unusual case of Kartagener’s syndrome where the patient had associated ventricular septal defect, pectus excavatum and was fertile.

Introduction
Kartagener’s syndrome is an autosomal recessive condition occurring with a frequency of 1:30,000 – 1:40,000. It is characterized by the classic triad of situs inversus, bronchiectasis and sinusitis. The condition was first described by Siewert in 1904 but details of the condition were given by Manes Kartagener in 1933 and it is known by his name ever since.

Case History
A 26 years old male, married and father of 2 children, presented with history of productive cough, headache and fever off and on for the past 4-5 years. For the last 2 years the patient had also developed exertional breathlessness and for the past 2 months he had occasional episodes of hemoptysis.

On examination patient had clubbing and pectus excavatum (Fig. 1). On systemic examination a pansystolic murmur was present over the precordium. Bilateral crepitations and rhonchi were present.

A chest X-ray was done which showed dextrocardia (Fig. 2) and cardiomegaly. Subsequent investigations revealed situs inversus, bronchiectasis, ventricular septal defect. He was diagnosed as a case of Kartagener’s syndrome and treated with antibiotics, mucolytics and bronchodilators and he showed signs of improvement. The patient was discharged and is in follow up with us.

Discussion
Kartagener’s syndrome in seen in 50% cases of primary ciliary dyskinesia. Primary ciliary dyskinesia is an inherited autosomal recessive condition characterized by bronchiectasis (Fig. 3), sinusitis (Fig. 4) and otitis media. When situs inversus is associated with primary ciliary dyskinesia then it is referred to as Kartagener’s syndrome.

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. 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. Additionally since visceral rotation during development depends upon ciliary motion, situs inversus is seen in Kartagener’s syndrome (Fig. 5). 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. Diagnosis of this condition is usually clinical accompanied by imaging studies.

Our case was unusual for 3 reasons. Firstly our patient was fertile and had two children. He did not agree to a seminal fluid analysis. Males with Kartagener’s syndrome are generally infertile but there are few reports of fertility with Kartagener’s syndrome. Munro et al in their study found a third of...
their patients of primary ciliary dyskinesia to have normal spermatozoa and 2 patients were fertile.

Second the patient had ventricular septal defect. Congenital heart diseases have been reported with Kartagener’s syndrome but these are rare. Kennedy et al\(^2\) in their study of congenital heart disease in primary ciliary dyskinesia found 1 patient out of 21 to have VSD. Tkebuchava et al\(^3\) found 4 out of 9 patients to be having VSD.

Lastly our patient also had pectus excavatum. Kennedy et al\(^3\) found 10% of patients with primary ciliary dyskinesia to have pectus excavatum. It is unclear whether a genetic or pathophysiological link between pectus excavatum and Kartagener’s syndrome exists.

**Conclusion**

Kartagener’s syndrome is a rare condition that we come across. Association of fertility, congenital heart defects and pectus excavatum are very rare with this syndrome. We need documentation of more cases to find out these associations with Kartagener’s syndrome.

**References**