Kartagener syndrome - a case report

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Abstract:
Kartagener syndrome, rare disorder, a type of primary ciliary dyskinesia, is an autosomal recessive inherited disorder of special cells called cilia. Kartagener syndrome is estimated to occur in 1 per 32,000 live births, and affects both males and females. 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. We report a case of Kartagener's syndrome where the patient associated with cor pulmonale.

Keyword: kartagener syndrome, dextrocardia, primary ciliary dyskinesia

INTRODUCTION
Ciliary abnormalities are classified into two categories; specific congenital defects of ciliary structure incident to the "primary ciliary dyskinesia" and acquired nonspecific anomalies of the ciliary apparatus. Chronic sinusitis, bronchiectasis, and situs inversus are known as the clinical triad of Kartagener's syndrome (KS). Kartagener's syndrome is now recognized as a clinical variant of primary ciliary dyskinesia (PCD). Primary ciliary dyskinesia is an autosomal recessive disorder characterized by inefficient or absent mucociliary clearance. The coexistence of primary ciliary dyskinesia and situs inversus is called Kartagener's syndrome and occurs in 50% of primary ciliary dyskinesia patients. Situs inversus can be defined as the random distribution of internal organs during embryogenesis, probably due to the absence of the ciliary activity that is responsible for normal organ distribution.

CASE REPORT:

Fig. 1 PATIENT
Mr. G. (Fig.1) 24years old unmarried male, admitted with the history of recurrent respiratory tract infections for 10 years, cough with copious mucopurulent sputum production 50ml per day and dyspnoea for 6 months, more for 2months, class
IV with orthopnoea and paroxysmal nocturnal dyspnoea. His younger brother died due to a chronic respiratory illness 5 years ago. Patient is not a smoker. Routine basic investigations were normal; sputum for acid fast bacilli was negative. Chest x-ray (Fig. 2) showed cardio thoracic ratio 0.6, heart shadow in right side, aorta in right side, stomach gas bubble visualized in right side and cystic changes present in bilateral lower lobes. ECG showed sinus tachycardia with right atrial and right ventricle enlargement with features of dextrocardia. Echocardiogram showed dextrocardia, liver in left side, RA, RV – dilated, severe pulmonary hypertension with moderate tricuspid regurgitation. Ultrasoundogram showed dextrocardia with situs inversus, congested liver.

Fig. 2 CHEST X RAY
CT thorax (Fig. 3) showed dilated pulmonary artery, aortic arch in right side, cardiomegaly, RA, RV grossly enlarged, descending aorta on right side. Nonhomogeneous opacity with multiple cavity involving both lower lobes, liver in left side and spleen in right side.

Fig. 3 CT THORAX
X-ray paranasal sinuses (Fig. 4) showed agenesis of bilateral frontal sinuses, bilateral maxillary sinusitis, deviated nasal septum to right side, left inferior turbinate hypertrophy. He is not willing for semen analysis.

Fig. 4 X-RAY PNS
Treated with antibiotics, bronchodilators, mucolytics, anti-failure measures and chest physiotherapy. Patient improved well with above measures, discharged and he is on regular follow up now.

DISCUSSION
Primary ciliary dyskinesia (PCD), also known as immotile ciliary syndrome or Kartagener Syndrome (KS), is a rare, ciliopathic, autosomal recessive genetic disorder that causes a defect in the action of the cilia lining the respiratory tract and fallopian
tube. It occurs with a frequency of 1:30,000 – 1:40,000. It is characterized by the classic triad of situs inversus, bronchiectasis and sinusitis. Genetically heterogeneous disorder affecting motile cilia which are made up of approximately 250 proteins. Around 90% of individuals have ultra structural 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.

The main consequence of impaired ciliary function is reduced or absent mucus clearance from the lungs, and susceptibility to chronic recurrent respiratory infections, including sinusitis, bronchitis, pneumonia, and otitis media. Progressive damage to the respiratory system is common, including progressive bronchiectasis beginning in early childhood, and sinus disease (sometimes becoming severe in adults). However, diagnosis is often missed early in life despite the characteristic signs and symptoms. Kartagener syndrome is recognized by the three main symptoms of chronic sinusitis, bronchiectasis, and situs inversus. Chest x-ray or computed tomography (CT) scan detect lung changes characteristic of the syndrome. Taking biopsy of the lining of the trachea, lung, or sinuses can allow microscopic examination of the cells that line the respiratory tract, which can identify defective cilia.

Medical care for a person with Kartagener syndrome focuses on prevention of respiratory infections, and prompt treatment of any that may occur. Antibiotics can relieve sinusitis, and inhaled medications and respiratory therapy can help if chronic lung disease develops. Small tubes may be placed through the eardrums to allow infections and fluid to drain out of the middle ear. Adults, especially men, may have difficulty with fertility, and may benefit from consulting a fertility specialist. It usually becomes less problematic near the end of the patient's second decade and many patients have near normal adult lives. Our patient had the typical features of recurrent respiratory infections, bronchiectasis, absent frontal sinuses, Dextrocardia with situs inversus totalis associated with cor pulmonale.

**CONCLUSION**

Kartagener's Syndrome should always be considered in patients with recurrent respiratory infections and situs inversus. Symptoms result from impaired mucociliary clearance. We had the patient with bronchiectasis, absent frontal sinuses, Dextrocardia with situs inversus totalis associated with cor pulmonale.

**REFERENCES**


