KARTAGENER’S SYNDROME: A CASE REPORT

V. N. Raju1, Banavath Durgaprasad Naik2, Tippagudisa Anil3, Vulli Venkatesh4, Netala S. Vara Prasad5

1Professor, Department of Pulmonary Medicine, Andhra Medical College.
2Post Graduate, Department of Pulmonary Medicine, Andhra Medical College.
3Post Graduate, Department of Pulmonary Medicine, Andhra Medical College.
4Post Graduate, Department of Pulmonary Medicine, Andhra Medical College.
5Post Graduate, Department of Pulmonary Medicine, Andhra Medical College.

ABSTRACT

BACKGROUND
Kartagener’s syndrome is a rare, autosomal recessive genetic ciliary disorder comprising the triad of situs inversus, chronic sinusitis, and bronchiectasis. Patient usually presents with chronic recurrent chest, ear, nose, throat infections and infertility. Situs inversus can be seen in about 50% of cases. Here we present one such case that presented with bronchiectasis, recurrent sinusitis, and situs inversus totalis, which fits in the triad of Kartagener’s syndrome. The same was diagnosed based on clinical presentation and radiological findings.

KEYWORDS
Situs Inversus, Chronic Sinusitis, and Bronchiectasis.


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. Comprising a triad of situs inversus, bronchiectasis and sinusitis. 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. An early diagnosis and timely treatment options may be offered for infertility in these young patients and also for the preservation of pulmonary function, quality of life, and life expectancy in this disease.

CASE REPORT
A 19-year-old unmarried female patient presented to outpatient department with chief complaints of common cold, sneezing, and cough with expectoration since one week. She has history of recurrent respiratory infection since childhood. On examination there is clubbing. On auscultation, bilateral wheeze and bilateral basal crackles were heard. On cardiovascular examination, apex beat and heart sounds were heard on right side, indicating dextrocardia. Electrocardiogram showed evidence of dextrocardia, with right axis and poor R wave in left side leads. Chest X-ray postero-anterior (PA) view (Fig 1) revealed cardiac apex and aortic arch on the right side, suggesting dextrocardia and left lower zone cystic changes suggesting bronchiectasis, fundic gas shadow on right side and elevated dome on left side. An ultrasound of the abdomen revealed a normal liver and gall bladder on the left side and a normal spleen on the right side.

High resolution CT (Fig 2) revealed dextrocardia, with situs inversus totalis, central bronchiectatic changes involving right middle lobe and lingular lobe with thickening of bronchi, multiple tiny nodular infiltrates in tree in bud pattern involving the right middle lobe, lingual, superior, anterior and lateral basal segments of both lower lobes. Liver on left side and stomach and spleen on right side in mediastinal window (Fig 3). X ray PNS (Water’s view) there is maxillary sinusitis with absence of frontal sinus (Fig 4). CT PNS showed gross deviation of nasal septum on left side, bilateral maxillary and ethmoidal sinusitis (Fig 5) and absent of frontal and sphenoidal sinus (Fig 6).

Fig. 1: Chest X-ray postero-anterior (PA) view

(Fig 1) revealed cardiac apex and aortic arch on the right side, suggesting dextrocardia and left lower lobe cystic changes suggesting bronchiectasis, fundic gas shadow on right side and elevated dome on left side.
(Fig 2) revealed dextrocardia, with situs inversus totalis, central bronchiectatic changes involving right middle lobe and lingular lobe with thickening of bronchi, multiple tiny nodular infiltrates in tree in bud pattern involving the right middle lobe, lingual, superior, anterior and lateral basal segments of both lower lobes.
DISCUSSION

Ciliary abnormalities are due to congenital defects of ciliary structure "Primary ciliary dyskinesia" and acquired nonspecific anomalies of the ciliary apparatus.\(^1\) Chronic sinusitis, bronchiectasis, and situs inversus are known as the clinical triad of Kartagener’s syndrome (KS).\(^2\) KS is now recognized as a clinical variant of primary ciliary dyskinesia (PCD). PCD is an autosomal recessive disorder characterized by inefficient or absent mucociliary clearance. The coexistence of PCD and situs inversus is called KS and occurs in 50% of PCD 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. Mutations have been identified in eight genes in PCD. Most of the disease-causing mutations identified to date involve two genes, these are genes coding for the dynein axonemal intermediate chain 1 (DNAI1) and the dynein axonemal heavy chain 5 (DNAH5) in ciliary outer dynein arms. Mutations in two genes have been associated with KS. Mutations in the coding region of DNAH11 account for situs inversus totalis and a minority of cases of PCD. Most patients with PCD have a history of neonatal respiratory distress. Rhinosinusitis and otitis media are cardinal features of the disease in PCD, and are responsible for much of the morbidity associated with PCD in early childhood. Most patients will present as chronic, productive cough due to the lack of effective mucociliary clearance leads to recurrent episodes of pneumonia or bronchitis and bronchiectasis.

Genitourinary system manifests as Male infertility due to the defects in sperm tail axonemes.\(^3,4\) Central nervous system manifests as hydrocephalus with PCD, hypothetically due to impaired cerebrospinal fluid flow secondary to dysfunctional motile cilia that line the ventricular ependymal cells. In Eye as retinitis pigmentosa.\(^5\) In Kidney presents as autosomal-dominant polycystic kidney disease.\(^6\) The diagnostic approach to PCD are mainly, ct scan, ct cerebellum, us abdomen, mucosal biopsy, semen analysis, saccharine test, audiological test and pulmonary function test, Genetic testing, nasal NO measurement, immunofluorescent analysis, and high-speed video-microscopy for the early detection and diagnosis of PCD. In our case, however, we could not perform these tests and the diagnosis was essentially clinico-radiological. There is no curative treatment only symptomatic treatment with antibiotics, mucolytics and vaccination and surgical lung transplantation.

CONCLUSION

Thus any patient with a history of recurrent cough and cold, and bronchiectasis with dextrocardia should be examined for Kartagener’s syndrome which is a part of PCD. KS is itself a rare entity, which we thought worth for sharing among medical personnel.

High index of suspicion will lead to prompt diagnosis. Earlier diagnosis and appropriated management can prevent fatal complications like respiratory failure and cor pulmonale.

REFERENCES