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Kartagener's Syndrome with Seizures: A Rare Case Report

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ABSTRACT

Primary ciliary dyskinesia (PCD) is a genetic disorder with an autosomal recessive mode of inheritance. It is caused by a defect in the structure of cilia, due to which ciliary movement and consequently, its function, are impaired. Sinusitis, nasal polyposis and otitis media with effusion are commonly seen among patients. Seizures are rare in such patients. We are presenting a rare case report of a patient with Kartagener's syndrome who presented with seizure.

Keywords: Kartagener's syndrome, seizure, sinus thrombosis

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,[1,2] comprising a triad of situs inversus, bronchiectasis and sinusitis.[1,2] Although Siewert 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.[2] The estimated prevalence of PCD is about 1 in 30,000,[3] though it may range from 1 in 12,500 to 1 in 50,000.[1]

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. Seizures are very rare in such patients. We present a case of a 38-year-old female patient of KS with seizures.

CASE REPORT

A 38-year-old nonsmoker female presented to us with chief complaints of throbbing type of diffuse headache since 5 days and 3 episodes of generalized tonic-clonic seizures since 2 days. There was no history of fever, nausea, vomiting or any symptoms suggestive of other focal neurological deficit. There was history of recurrent chest infections since her childhood and progressively increasing breathlessness for last 30 years. She had been married for last 20 years but had no children. She received anti-tubercular treatment for chronic cough for 8 months, 10-year back with no relief. Her family history revealed no paternal consanguinity.

On examination, patient was conscious but drowsy. Her pulse rate was 122/min and regular, respiratory rate was 27/min, blood pressure was 110/74 mmHg, SpO_2 81% on room air. Bilateral pedal edema and Grade III clubbing was present. On auscultation, diffuse rhonchi and crackles were heard. Rest of systemic examinations was within normal limits. Routine investigations were normal except high total leukocyte count (17,000/mm^3) with neutrophilic predominance. Electrocardiogram (ECG) was suggestive of dextrocardia. Electroencephalography (EEG) of the patient was normal. Chest X-ray film posteroanterior view showed bronchiectatic changes with dextrocardia (Fig. 1). CT abdomen was suggestive of situs inversus (Fig. 2). CECT thorax revealed bronchiectasis and situs inversus (Fig. 3). CT brain showed left middle cerebral artery territory hemorrhagic infarct (Fig. 4). CT venography brain of the patient was suggestive of left transverse sinus and superior sagittal sinus thrombosis (Fig. 5). Patient was managed with antibiotics, antiepileptics, low-molecular-weight heparin and other supportive treatment. Patient was doing well on follow-up.

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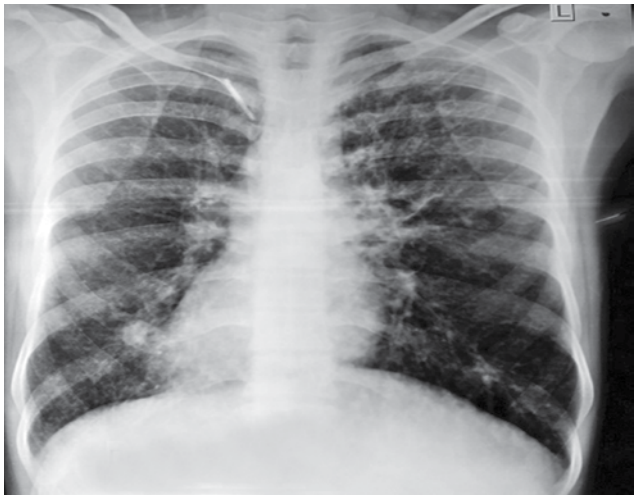

Figure 1. Chest X-ray film PA view showing dextrocardia.

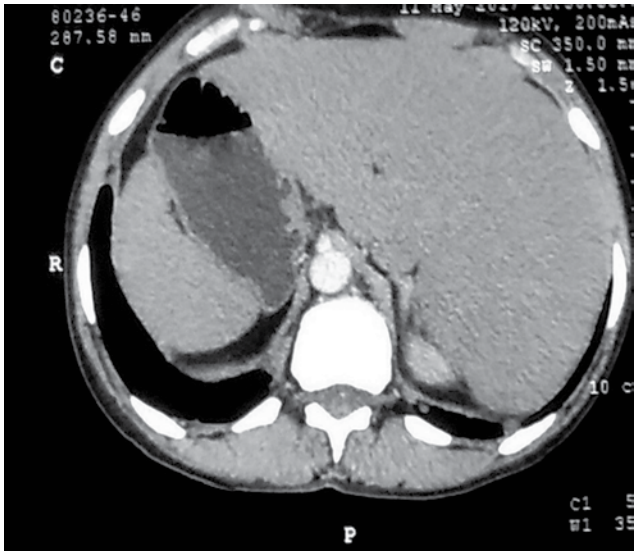

Figure 2. CT abdomen showing situs inversus.

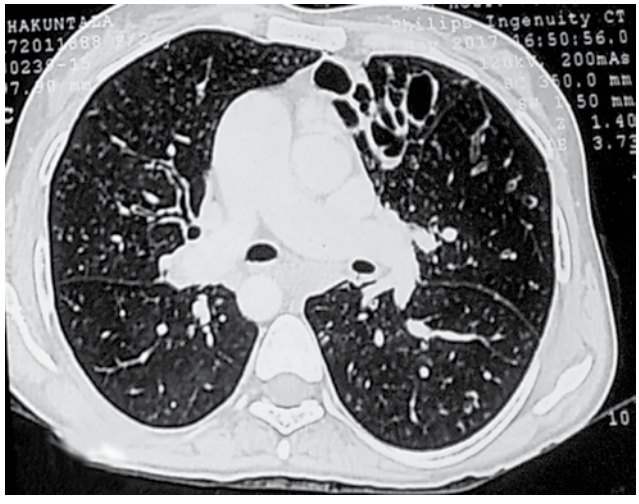

Figure 3. CECT thorax showing bronchicetatic changes in upper and lower lobe of left lung.

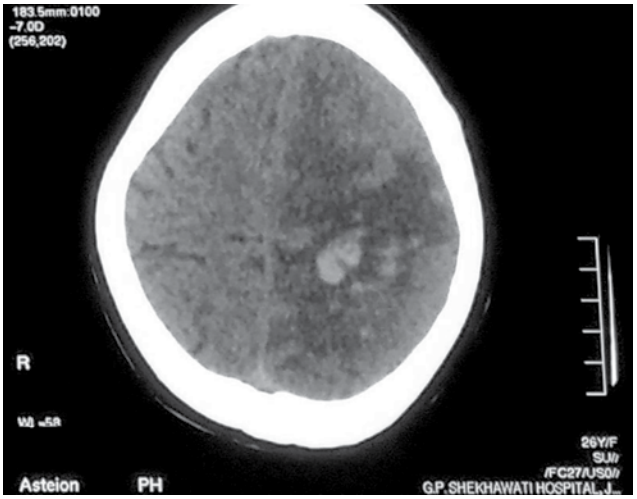

Figure 4. CT brain showing hemorrhagic infarct in left middle cerebral artery territory.

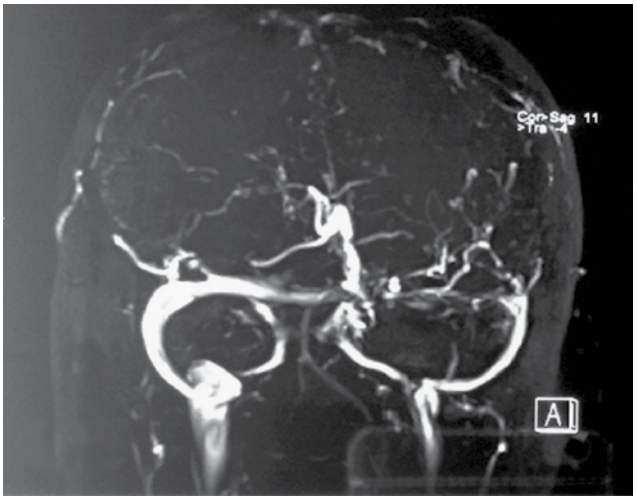

Figure 5. CT venogram brain showing left transverse and superior sagittal sinus thrombosis.

DISCUSSION

PCD is a rare genetic disorder with an autosomal recessive mode of inheritance.[4] It is caused by a defect in the dynein arm structure of cilia, due to which ciliary movement and consequently its function, are impaired. Ciliary movements are responsible for the rotation and orientation of internal organs in the 10th to 15th days of gestation. In PCD, the underlying ciliary dysfunction causes incomplete rotation or malrotation of one or many internal organs, most commonly the heart.[5] Isolated malrotation of the heart (situs solitus) is associated with severe anomalies of the vessels connecting the heart and is thus relatively rare as such cases have a very low survival. More commonly, a right-sided heart (dextrocardia) exists along with malrotation of the other internal organs,

namely: lungs, liver, spleen, kidneys and intestines (situs inversus).

About 50% of PCD patients develop situs inversus and KS, which has been classically described as a triad of dextrocardia, sinusitis and bronchiectasis, and male infertility; the incidence of KS is estimated to be around 1:15,000 with variable penetrance, and phenotypic differences have been observed because the underlying genetic mutation has a pleiotropic effect.

Radiology, in the form of a chest X-ray, quickly corroborates the clinical suspicion of dextrocardia, but may also reveal dextrocardia and situs inversus as an incidental finding on routine preoperative work-up. CT thorax may further delineate malrotation, and bronchiectasis if any, and other changes found in PCD. In the event of the CT scan being inconclusive, a Gallium-67 scan can establish the bronchiectatic changes.[6]

Seizures are not reported in any case in the literature as a manifestation of KS previously. In our case, patient developed venous sinus thrombosis as a result of chronic infection due to bronchiectasis, which may be the cause for seizures.

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