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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: Karatgner syndrome, seizure, sinus thrombosis

1. Introduction

Kartagener's syndrome (KS) is a subset of a larger group of ciliary motility disorders called primary ciliarydyskinesias (PCDs). It is a genetic condition with an autosomal recessive inheritance, [1, 2] comprising a triad of situsinversus, bronchiectasis and sinusitis. [1, 2] Although Siewartfirst described this condition in 1904, it was Kartagenerwho 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 42 year old female patient of KS with seizures.

2. Case Report

A 42 year old non smoker female presented to us with chief complaints of throbbing type of diffuse headache since 5 days and 4 episodes of generalised 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 24 years but had no children. She received anti - tubercular treatment for chronic cough for 9 months ten year back with no relief. Her family history revealed no paternal consanguinity. On examination, patient was conscious but drowsy. Her pulse rate was 114 per minute and regular, respiratory rate was 31/min, blood pressure was 118/78mmhg, spo2 80% on room air. Bilateral pedal oedema and grade III clubbing was present. On auscultation, diffuse ronchi and crackles were heard. Rest of systemic examinations was within normal limits. Routine investigations were normal except high total leucocyte count (19700) with neutrophilic predominance. ECGand 2D Echo was suggestive of dextrocardia without any significant abnormality. EEG of the patient was normal. Chest X ray film PA view shows bronchectiatic changes with dextrocardia. USG abdomen suggestive of situsinversus. HRCT thorax revealed bronchectiasis and situsin versus. CT brain shows MCA territory hemorrhagic infarct. CT venography brain of the patient was suggestive of left transverse sinus and superior sagittal sinus thrombosis. Patient was managed with antibiotics, antiepileptics, low molecular weight heparin and other supportive treatment. Patient was doing well on follow up.

3. Discussion

PCD is a rare genetic disorder with an autosomal recessive mode of inheritance (Bush et al., 1998) (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 (Afzelius, 1976) (5). Isolated malrotation of the heart (situssolitus) is associated with severe a 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 (situsinversus).

About 50% of PCD patients develop situsinversus 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: 15000 (Bush et al., 1998) with variable penetrance, and phenotypic differences have been observed because the underlying genetic mutation has a pleiotropic effect (Holmes, 1964).

Radiology, in the form of a chest X - ray, quickly corroborates the clinical suspicion of dextrocardia, but may also reveal dextrocardia and situsinversus as an incidental finding on routine pre - operative workup. CT thorax may further delineate malrotation, and bronchiectasis if any, and other changes found in PCD (Barker, 2002). In the event of

the CT scan being inconclusive, a Gallium - 67 can establish the bronchiectatic changes (Becker, 2000) (6).

Seizures are reported very rarely 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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