Phenotypic Presentation Of a Rare Case Of Kartagener Syndrome

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Abstract: Kartagener's syndrome is a rare, autosomal recessive genetic ciliary disorder comprising the triad of situs inversus, chronic sinusitis, and bronchiectasis. The main problem lies in the defective movement of cilia, leading to recurrent chest infections, upper respiratort tract infections, and infertility. We hereby report an unusual cases of this rare entity. The need for a high index of suspicion to make an early diagnosis cannot be overemphasized in such patients so that wherever possible, options for timely treatment of infertility may be offered and unnecessary evaluation of symptoms is avoided. [Adesh K Natl J Integr Res Med, 2019; 10(4):86-88]

Key Words: Bronchiectasis, Kartagener's Syndrome, Sinusitis, Situs Inversus

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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. Although Siewart 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 [1,2]. 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 [2] . In KS, the ultrastructural genetic defect leads to impaired ciliary motility which causes recurrent chest, ear , nose, throat, and sinus infections, and infertility.

Afzelius was the first to recognize the relationship between KS and male infertility when he observed lack of dynein arms in the sperms and cilia of four subjects, three with KS and a fourth one, brother of one of the three subjects [3]. 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. Also, although unproven, it seems likely that early diagnosis is important for the preservation of pulmonary function, quality of life, and life expectancy in this disease.

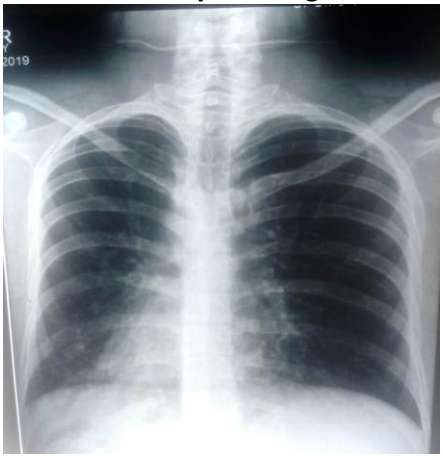
Case History: This was a 32-years-old non-smoker male. He presented to the outpatient with chief complaints of recurrent episodes of common cold, sneezing, and cough with expectoration and shortness of breath since childhood. He had been previously treated with antibiotics, antihistamines, bronchodilators, inhaled and oral corticosteroids, and but the response was only partial and temporary.

On examination, the vital parameters were within normal limits. Physical examination revealed grade 3 digital clubbing and apex beat on the right side in fifth intercostal space. His auscultation finding revealed bilateral wheeze and bilateral basal crackles, with heart sounds being best heard on the right side of the chest. Routine blood investigation (complete blood count ,differential blood count, liver function tests and kidney function tests, random blood sugar, absolute eosinophil count sent , they were found to be within normal range.

Sputum for acid fast bacilli examination and sputum culture for mycobacterium was negative, sputum also sent for gram staining and bacterial culture sensitivity. on gram staining gram negative bacilli was found and in culture pseudomnas was detected which was sensitive to piperacillin and tazatobactum and amikacin , sputum for fungal stain and culture was negative.

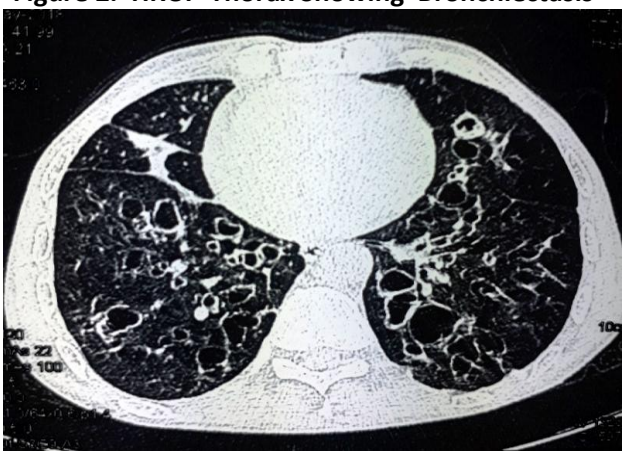
Chest X-ray postero-anterior (PA) view (Figure 1) revealed cardiac apex and aortic arch on the right side, suggesting dextrocardia.

Figure 1. Chest X-Ray Showing Dextrocardia



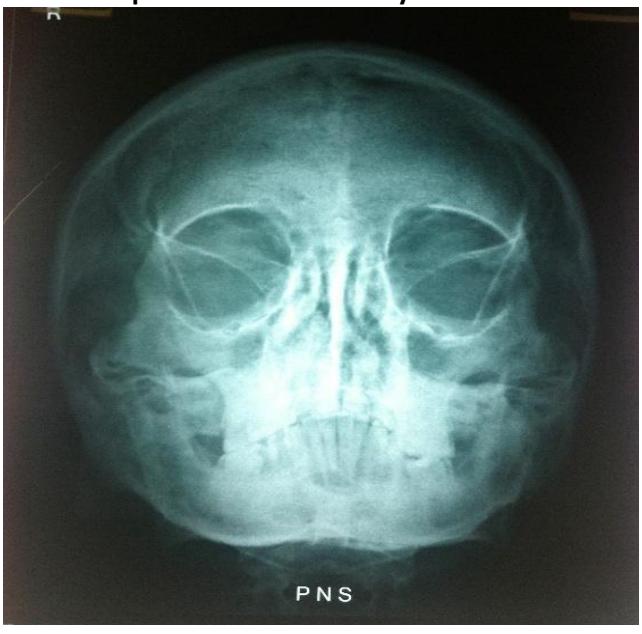
High Resolution Computed Tomography (HRCT) Chest (Figure 2) showed cystic bronchiectasis in both side predominantly in lower lobes.

Figure 2. HRCT Thorax Showing Bronchiectasis



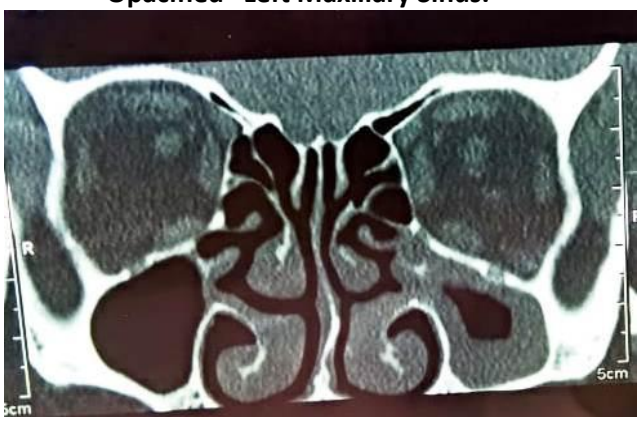
Electrocardiogram showed evidence of sinus tachycardia and Echocardiography revealed mirror image dextrocardia. Ultrasound of abdomen revealed liver on left side and spleen on right side suggesting situs inversus. X-ray Paranasal Sinuses (Figure 3) and CT scan Paranasal sinuses (saggital section, Figure 4) showed opacified left maxillary sinuses.

Figure 3. X-ray Paranasal Sinuses Showing Opacified left Maxillary Sinus



A semen analysis revealed azoospermia. Nasal brush biopsy on electron microscopy revealed intact inner and outer dynein arms with few admixed cilia show abnormalities of arrangements of the microtubules. In treatment antibiotics given as per sensitivity pattern for 2 weeks and inhaled bronchodilators and inhaled steroids given with chest physiotherapy. Patient continued on Azithromycin 500 mg thrice weekly and supportive and symptomatic treatment given in follow up.

Figure .4 CT Scan Paranasal sinuses Showing Opacified Left Maxillary Sinus.



Discussion: Primary ciliary dyskinesia (PCDs) is a congenital disorder of ciliary motility and about 50% of PCD patients have situs inversus. These cases of PCD with situs inversus are known as Kartagener's syndrome.

PCD is a phenotypically and genetically heterogeneous condition wherein the primary defect is in the ultrastructure or function of cilia.[4] Such defects are identified in approximately 90% of PCD patients and involve the outer dynein arms, inner dynein arms, or both. 8% Of the PCD patients carry mutations of the dynein genes *DNAI* and *DNAH5* [5,6].

The most common defect, first described by Afzelius, is a reduction in the number of dynein arms, which decreases the ciliary beat frequency[3]. The defective ciliary motility/immotility is the underlying defect which leads to accumulation of secretions and consequent recurrent sinusitis, bronchiectasis, infertility, and situs inversus. The age at which this condition diagnosed is often variable because severity of symptoms varies in different patients as some patients may have symptoms since birth and in others symptoms appear later.

Diagnostic criteria for this condition include clinical picture suggestive of recurrent chest infections, bronchitis, and rhinitis since childhood, along with one or more of the following: (a) situs inversus in the patient/sibling (b) alive but immotile spermatozoa; (c) reduced

or absent transbronchial mucociliary clearance and (d) cilia showing characteristic ultrastructural defect on electron microscopy [7].

In our cases, the diagnosis was essentially clinico-radiological, with azoospermia. Most infertile patients with KS have a normal spermatozoid count, but with a structural defect and a complete lack of motility[7].

Male patients with KS invariably present infertility, while women present reduced fertility. [7] Infertility in male KS patients is due to diminished sperm motility, while in females it is due to defective ovum transport because of dyskinetic motion of oviductal cilia, suggesting that the ciliated endosalpinx is essential for human reproduction [4].

The treatment of infertility in these patients should be individualized depending on sperm motility. In cases where there is no sperm motility, intracytoplasmic sperm injection (ICSI) may be the most appropriate treatment. However, if sperm motility is present, a trial of *in vitro* fertilization (IVF) should be considered[4]. One concern regarding the fertility treatment of men with PCD is the possibility that the resultant child has the risk of being affected by the same condition.

Conclusion: Kartagener's syndrome is a rare, autosomal recessive genetic ciliary disorder. There is need for a high index of suspicion to make an early diagnosis and timely intervention for the preservation of pulmonary function, quality of life, and life expectancy in this disease.
Introduction

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