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Case Report

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An atypical case of Kartagener's syndrome

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ABSTRACT

Kartagener's syndrome is an autosomal recessive disease characterized by the tetrad of situs inversus, bronchiectasis, sinusitis and infertility. It is a subset of a larger group of disease known as primary ciliary dyskinesia. A 23 year old male patient presented to our hospital with complaints of Right sided headache for the past 3 weeks. General Physical examination was normal except that the heart sounds were heard over the right side. Dextrocardia was suspected. Routine ENT and Diagnostic nasal endoscopy examination revealed deviated septum towards the left along with pale greyish polyp like mass in the right nasal cavity. CT PNS was ordered. It showed bilateral frontal, ethmoidal and maxillary sinusitis with Right nasal polyp. Routine Blood investigations were normal but Chest X-ray showed dextrocardia with fundal shadow over the right side and normal ECG was obtained only when the chest leads were reversed. USG abdomen confirmed the suspicion of situs inversus totalis. To rule out Kartageners syndrome HRCT Chest was done which revealed normal lungs with no findings. Sperm analysis was also done which was normal except for the count (<15 million/ml). Saccharin test was performed and it was delayed (>20 min). Anesthetic fitness was obtained and he was taken up for surgery - Septoplasty with fess. The diagnosis of kartageners syndrome was done based on clinical and radiological findings, with variation in form of no bronchiectasis and oligospermia. This type of atypical presentation of Kartagener's syndrome might be a rare sub type of the disease which is infrequently reported.

Keywords: Primary ciliary dyskinesia, Oligospermia, Saccharin test, Nasal polyp, Situs inversus

INTRODUCTION

Kartagener's syndrome is an autosomal recessive disease characterized by the tetrad of situs inversus, bronchiectasis, sinusitis and infertility. This condition is a subset of a larger group of disease known as primary ciliary dyskinesia previously known as immotile ciliary syndrome. The incidence of Kartagener's syndrome (KS) ranges from 1 in 20,000 to 1 in 60,000.[1]

Only half the patients present with all the features a condition designated as complete KS, compared with incomplete KS, typically defined as cases in which situs inverus does not occur.[2]

The underlying basic defect lies in the ciliary ultrastructure which lines the respiratory tract, due to which ciliary motility and consequently its function are impaired. This causes recurrent chest, ear/nose/throat (ENT), and sinus infections, and infertility. Infertility is due to defective functioning of the tail of the sperm.

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.[3,4]

CASE REPORT

A 23 year old male patient presented to our outpatient at Shri Sathya Sai Medical College and Research Institute

with complaints of right sided headache for the past 3 weeks. The pain suddenly developed in the right temporal and frontal regions 3 weeks back which had a pricking quality and was continuous in nature. The patient had no other significant complaints. There was no history of any co morbid condition or previous surgery. The family history was also insignificant.

General physical examination was normal except that the heart sounds were heard over the right side with the apex heart beat in the right fifth intercostals space. Dextrocardia was suspected and chest X-ray along with ECG was ordered. Routine blood investigations were also done. Routine ENT examination revealed deviated septum towards the left with right inferior turbinate hypertrophy along with pale greyish polyp like mass in the right nasal cavity. There were secretions present in the left nasal cavity. Throat and ear were normal.

Diagnostic nasal endoscopy revealed multiple pale grey polyps in the right nasal cavity obstructing the right middle meatus with a boggy middle turbinate. There was a high septal deviation towards the left side with spur. The left middle turbinate also showed polypoidal changes. Hence a provisional diagnosis of right sinonasal polyposis with left DNS was made. CT PNS was ordered to confirm the diagnosis and determine the extent of the polyp

CT scan showed bilateral frontal, ethmoidal and maxillary sinusitis with right nasal polyp. There was bilateral osteo-meatal complex block. So the patient was diagnosed as chronic sinusitis with right sinonasal polyp with left DNS and was prepared for FESS with septoplasty.

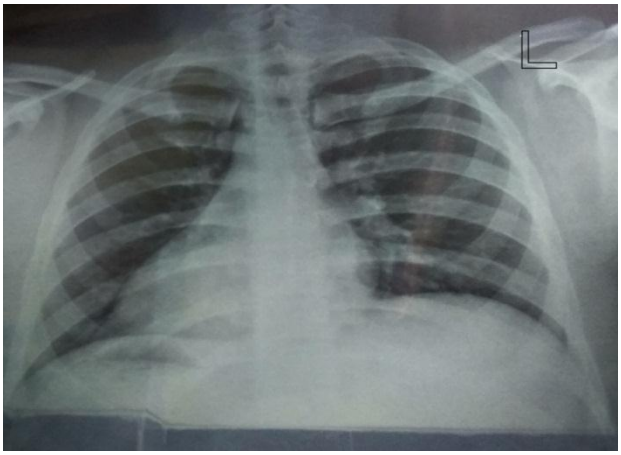

Figure 1: Chest X-ray showing heart in the right side with fundal shadow below it.

Routine blood investigations were normal but chest X-ray showed dextrocardia with fundal shadow over the right side (Figure 1). ECG showed changes related to dextrocarida (Figure 2) and normal ECG was obtained only when the chest leads were reversed. USG abdomen was ordered to rule out situs inversus totalis. USG confirmed the suspicion of situs inversus totalis. Now we suspected that the patient might have KS.

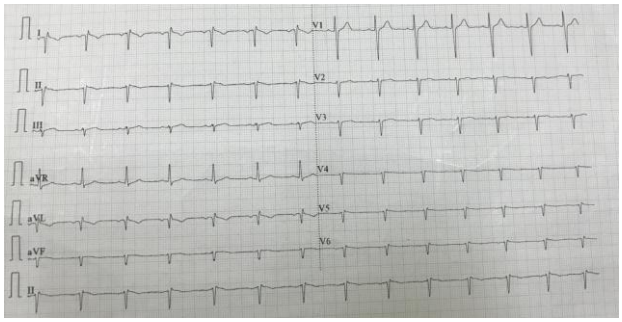

Figure 2: ECG with unreversed leads showing positive wave in aVR and tall R wave in V1 and absent R wave in V6.

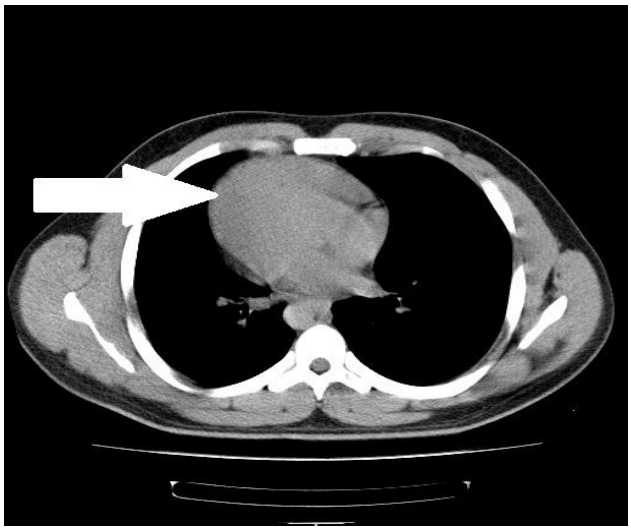

Figure 3: Arrow marks showing the heart in the right side.

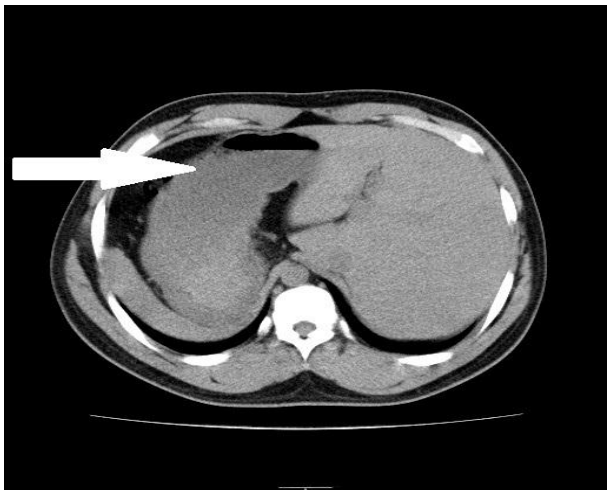

Figure 4: Arrow mark showing the stomach on the right side with air.

Chest medicine and venerology opinions were obtained. Chest Medicine had asked for HRCT chest to rule out any bronchiectasis. HRCT chest revealed normal lungs with no findings. While CT abdomen screening showed evidence of situs inversus totalis (Figure 3 and 4). Venerology had asked for sperm analysis as there was no other significant history.

Sperm analysis was normal except for the count which was less than 15 million/ml. Saccharin test was performed to confirm the ciliary dysfuction and as suspected it was delayed (>20 min) thereby confirming our diagnosis of KS. Anesthetic fitness was obtained and after explaining the condition to the patient, he was taken up for surgery. Septoplasty was done with bilateral uncinectomy, middle meatal antrostomy with anterior ethmoidectomy with removal of polpy, which was sent for biopsy. Partial turbinoplasty was done for the right middle and inferior turbinates. Patient recovered well and came for follow up without any problems. Histopathology of the sample came as hyperplastic sinonasal polyp.

DISCUSSION

Primary ciliary dyskinesia is a phenotypically and genetically heterogeneous condition wherein the primary defect is in the ultrastructure or function of cilia.[5,6] The main cause maybe defect in genes of the outer or inner dynein arms or both.

Pathophysiologically, the underlying defect is the defective ciliary motility/immotility. The severity of symptoms and the age at which the condition is diagnosed is quite variable.[7,8] Progression of the disease in KS is variable.

Diagnostic criteria for this condition include[9] clinical picture suggestive of recurrent chest infections, bronchitis, and rhinitis since childhood, along with one or more of the following: (1) situs inversus in the patient/sibling; (2) alive but immotile spermatozoa; (3) reduced or absent transbronchial mucociliary clearance; and (4) cilia showing characteristic ultrastructural defect on electron microscopy.

Nowadays two types tests are done for PCD apart from fulfilling the above criteria –

1. Screening tests (exhaled nasal nitric oxide measurement which is usually low in PCD, and saccharin test to assess mucociliary function of nasal epithelium) and
2. Diagnostic tests (ciliary beat pattern and frequency analysis using video recording, and electron microscopic confirmation of the ultrastructural ciliary defect).

In this particular case, only saccharin test was done which was delayed, thereby confirming the diagnosis. The diagnosis of this case was done based on clinical and radiological findings, with variation in form of no bronchiectasis in chest and oligospermia. These situations have been infrequently reported previously, and it could be possible that they are a variantion of KS.[10-12] Most infertile patients with KS have a normal spermatozoid count, but with a structural defect and a complete lack of motility.[13]

CONCLUSION

Atypical nature of this case is highlighted by 1. KS (primary ciliary dyskinesia) is usually reported with immotile sperms, but oligospermia has been rarely reported, 2. This patient does not classify as a complete KS case as he did not have the classic triad of the Syndrome (no evidence of bronchiectasis), nor is he a typical incomplete KS (because of presence of situs inversus). 3: This type of atypical presentation of KS might be a rare sub type of the disease which is infrequently reported. Hence we are reporting this case.

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