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Kartagener's Syndrome: A Rare Case Report

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Abstract: ***Kartagener's syndrome is a rare autosomal recessive disorder. It is characterised by a Kartagener's triad of bronchiectasis, sinusitis and situs inversus. Its estimated incidence is approximately 1 in 30, 000 live births [1, 2]. It is a subgroup of primary ciliary dyskinesia. Here we present a case of 30 year old female who was brought to the emergency department with an alleged history of unknown compound consumption. On examination patient was having breathlessness, to investigate this the patient was advised HRCT Chest and ultrasonography of abdomen. On HRCT chest cystic bronchiectasis were observed and heart apex was noted on right side. On ultrasonography of abdomen situs inversus was observed. After performing CT head sinusitis was also observed and Kartagener's syndrome was diagnosed. On taking detailed history patient was married since 7 years and was unable conceive. Early diagnosis of this syndrome can help in early treatment of infertility and other associated conditionsin such patients.***

Keywords: Bronchiectasis; Sinusitis; Situs inversus; Kartagener's syndrome; Dextrocardia

1. Introduction

Multiple works of literature have been published to date about the Kartagener's syndrome. Here we are presenting a case of Kartagener's syndrome in a 30 year old female; which was diagnosed incidentally. Kartagener syndrome (KS) is a subset of a larger group of ciliary motility disorders called primary ciliary dyskinesias (PCDs) [3]. KS is an inherited autosomal recessive disorder that causes defects in the action of ciliary movement, comprises of triad sinusitis, situs inversus, and bronchiectasis [4]. Being an autosomal recessive disorder it is mainly observed in children born to consanguineous marriages. It was first described by Siewert in 1904; however, Manes Kartagener recognized the clinical syndrome in 1933. Males and females are affected equally [5]. The main pathophysiology behind this syndrome is abnormal motility of cilia; which leads to recurrent episodes of respiratory infections, sinusitis and male as well as female infertility. If this patients are diagnosed earlier, it may help in early treatment of the associated conditions like infertility. Hence, we are presenting this case to increase the index of suspicion of Kartagener's syndrome in general populations.

2. Case Report

We present a case of a 30 - year - old female from Maharashtra, India who presented to emergency department of Government Medical College and Hospital Miraj with alleged history of unknown compound consumption. On admission the patient was having breathlessness, after clinical examination the patient was found tachypneic. So, to further evaluate the patient, she was advised high resolution CT scan (HRCT) of Chest and ultrasonography of abdomen. HRCT of the patient was performed on a "SIEMENS SOMATOM Definition AS 128 slice" equipment; on chest topogram which is taken as a scout image for HRCT chest cardiac shadow was noted on right and liver shadow was visualised on left side (image 1) After performing HRCT chest cystic bronchiectasis was observed in bilateral lung fields and dextrocardia was noted (image 2, 3). Ultrasonography was performed on "MINDRAY DC - 60" ultrasonography equipment; on ultrasonographic examination, liver along with IVC was noted in left hypochondrium just superior to upper pole of left kidney and spleen was noted in right hypochondrium (image 4, 5). Pancreas was noted in normal position. On taking detailed history she was married since last 7 years and was unable to conceive since then which was suggestive of infertility which is a feature of Kartagener's syndrome. So, we advised CT head and PNS (paranasal sinuses) of the patient to look for any polyp or sinusitis. After performing CT head pansinusitis was observed (image 5). Looking at the findings of CT scan, ultrasonography and history given by the patient we diagnosed Kartagener's syndrome in this patient.

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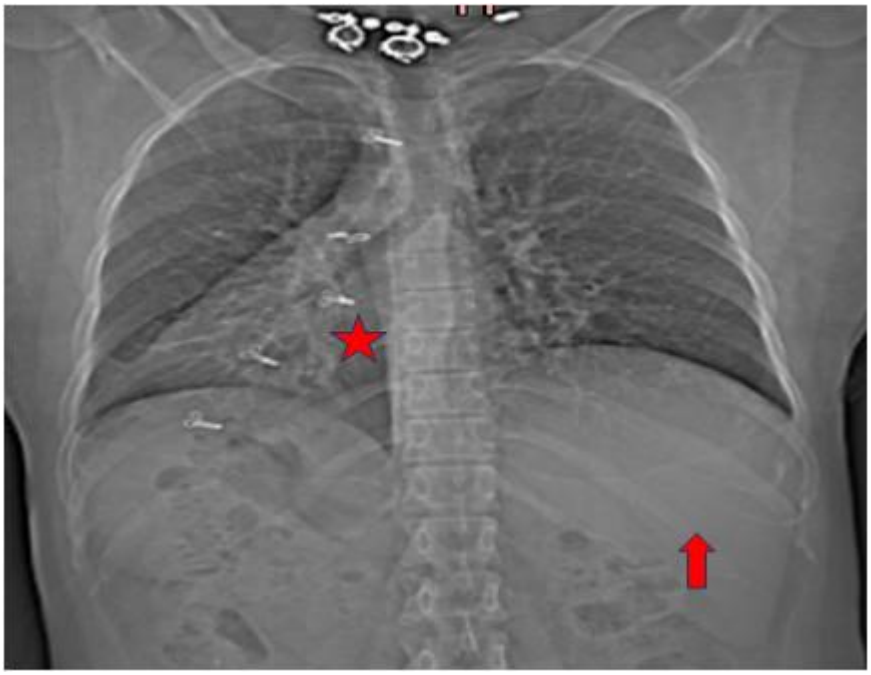

Image 1: Topogram (scout image) for HRCT Chest showing cardiac silohoutte on right side (asterisk) and liver shadow on left side (broad arrow)

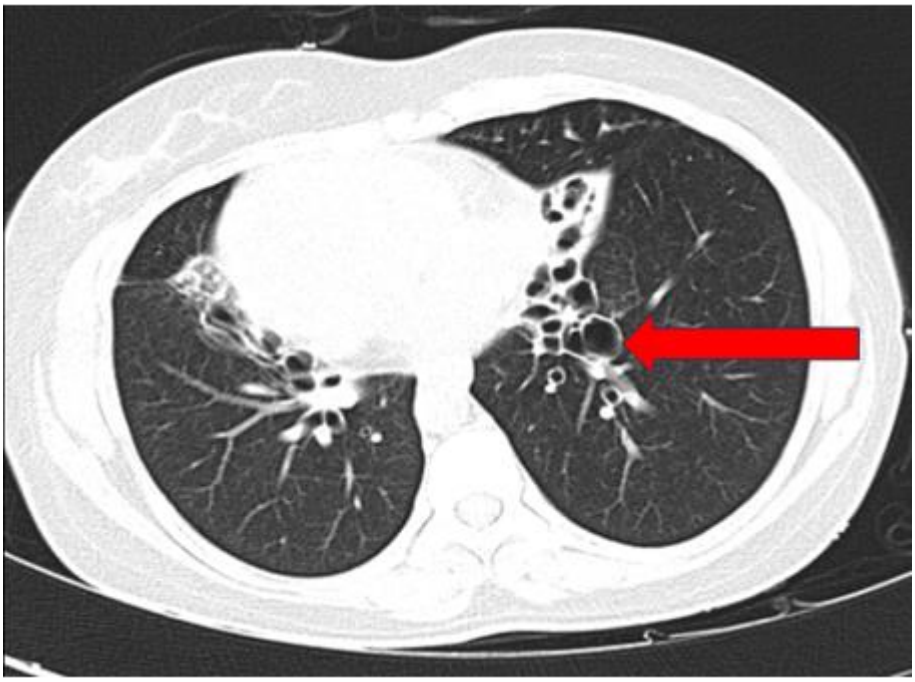

Image 2: Axial section CT scan of chest showing cystic bronchiectasis noted in bilateral lung fields (arrow).

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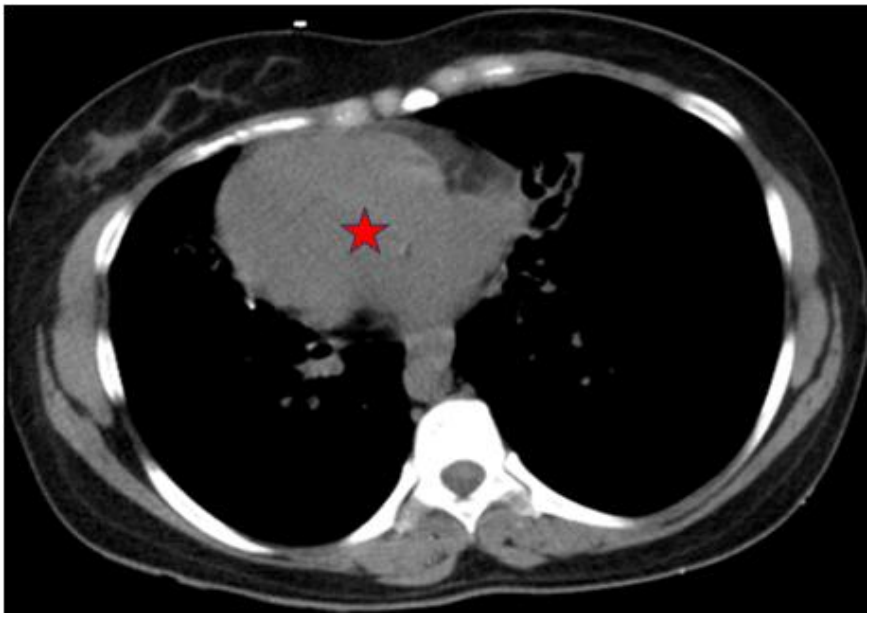

Image 3: Axial section CT scan of chest showing dextrocardia (asterisk)

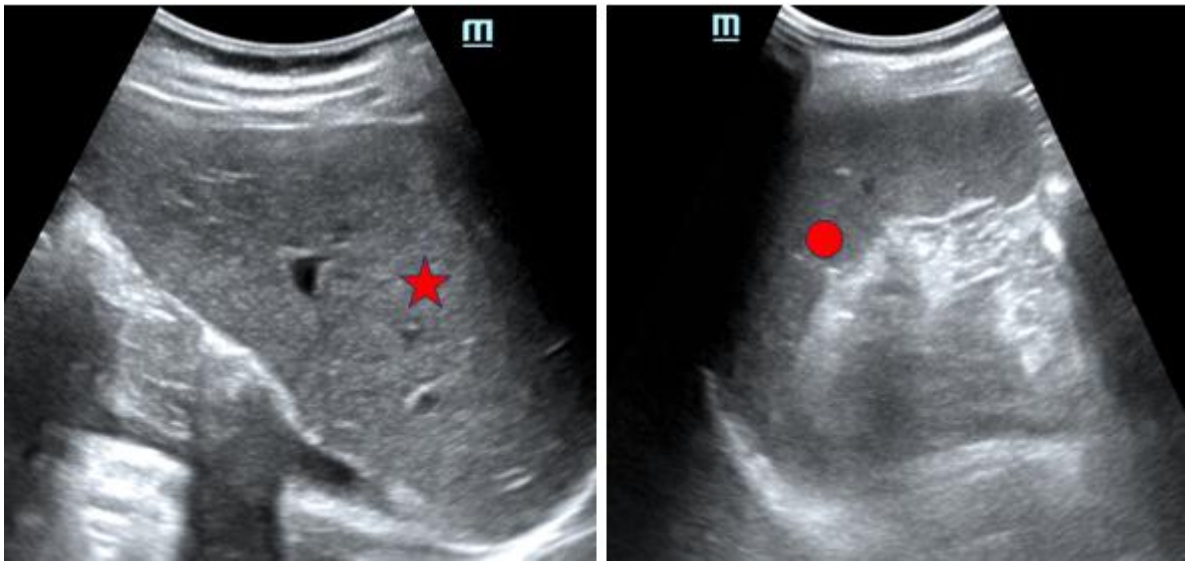

Image 4: Ultrasonographic images showing liver in left hypochondrium (asterisk) and spleen in right hypochondrium (circle)

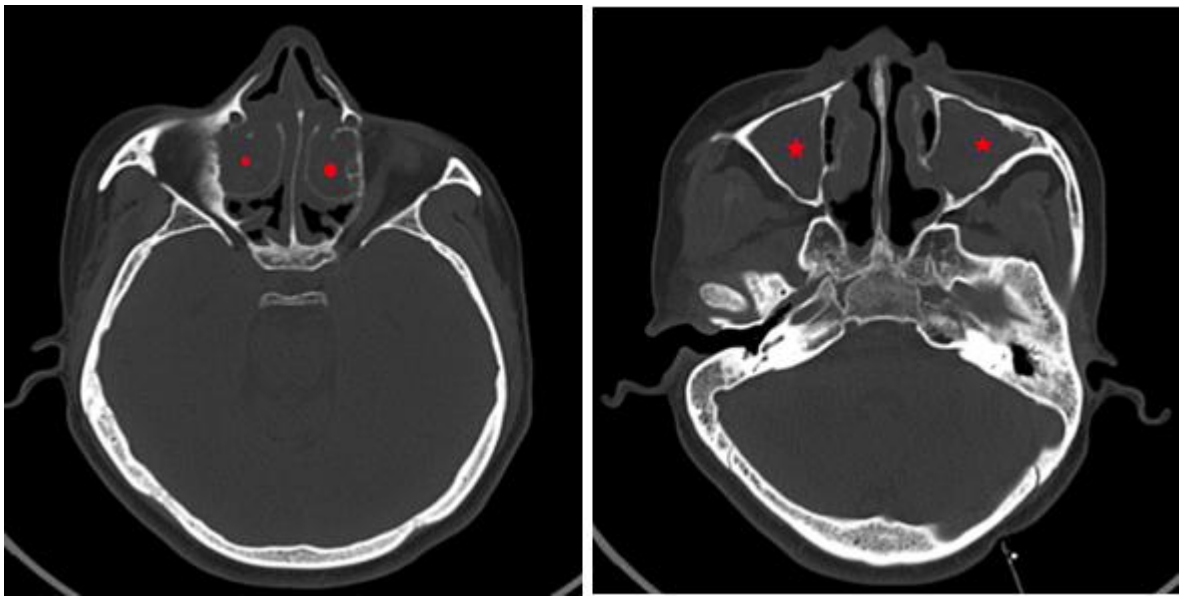

Image 5: AxialCT scan of head in bone window reconstruction showing mucosal thickening in bilateral ethmoid (red circle) and bilateral maxillary sinus (asterisk)

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3. Discussion

Kartagener's syndrome is a rare autosomal recessive disorder. Its estimated incidence is approximately 1 in 30, 000 live births [1, 2]. Kartagener syndrome is a subset of a larger group of ciliary motility disorders called primary ciliary dyskinesias [3]. KS is an inherited autosomal recessive disorder that causes defects in the action of ciliary movement, comprises of triad sinusitis, situs inversus, and bronchiectasis [4]. It was first described by Siewert in 1904; however, Manes Kartagener recognized the clinical syndrome in 1933. Males and females are affected equally [5]. Patients of Kartagener's syndrome may present with recurrent respiratorty tract infections, atypical asthma, chronic rhinosinusitis, male or female infertility. As the disease progresses clinical and radiographic evidence of bronchiectasis develops.

In Kartagener's syndrome, the gene mutation at DNAI1 and DNAH5 leads to impaired ciliary motility, which predisposes to recurrent sinopulmonary infections, infertility, and errors with left–right body orientation [6]. These faulty genes cause the cilia to be the wrong size or shape or move in the wrong way, making ciliary motility defective. Numerous defects have been described in components which comprise cilia, including lack of or dysfunction of inner and/or outer dynein arms, radial spokes, and microtubules.

Bronchiectasis is much more prevalent in people with situs inversus than in the general population. Sinusitis is the other component of the triad, which was markedly observed in our patient. Abnormal ciliary movements lead to an accumulation of secretions inside the paranasal sinuses. CT scan of the sinuses may show sinusitis, with mucosal thickening of the maxillary, ethmoid and frontal sinuses. In our patient mucosal thickening was noted in bilateral maxillary, ethmoid, frontal and sphenoid sinuses suggesting pansinusitis.

The diagnostic criteria recommended for this syndrome include history of chronic bronchial infection and rhinitis from early childhood, combined with one or more of following features: (a) situs inversus or dextrocardia in a patient or a sibling, (b) alive but immotile spermatozoa, (c) absent or impaired tracheobronchial clearance, and (d) cilia showing characteristic ultrastructural defect on electron microscopy [7, 8].

Radiological findings revealed bronchiectasis, dextrocardia, and situs inversus, which met the diagnostic criteria for Kartagener's syndrome. Laboratory screening and confirmatory tests, which required a better clinical setup, were not done in this case.

Treatment of Kartagener's syndrome is mainly based upon the treating the symptoms. Main role of treatment is to reduce symptoms and slow disease progression. Acute episodes of respiratory tract infections can be treated with antibiotics. But, to improve the quality of life in such patients prophylactic measures such as appropriate immunizations, mainly influenza vaccine and pneumococcal vaccine are recommended. Patients who develop recurrent pneumonia or hemoptysis and do not respond to antibiotics may benefit from segmental lung resection or lobectomy [5]. The long - term prognosis of patients with KS is good, with many patients living to an advanced age. Decreased quality of life is caused by chronic respiratory symptoms [9].

4. Conclusion

Kartagener's syndrome is a rare disease entity, but early diagnosis of this can lead to early treatment of conditions associated with this disease. If diagnosed in appropriate child birth age it can help in establishing the cause of infertility and can avoid additional investigations and will be beneficial to patient in terms of cost. Early diagnosis can help to give prophylactic vaccinations and to advise patients the proper prophylactic practices which will improve patients quality of life in Kartagener's syndrome. So, Kartagener's syndrome must be suspected in such patients. It can be an incidental finding on imaging investigations, so there should be high suspicion index for this because it will help to treat the patients and improve the quality of life of patients living with Kartagener's syndrome.

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