

Kartagener's Syndrome - A Case Report

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Abstract: *Kartagener's syndrome is a rare autosomal recessive syndrome characterized by bronchiectasis, situs inversus and chronic sinusitis. The triad is usually seen in a young age group and is punctuated by recurrent upper respiratory tract infections and pneumonia. The basic problem lies in ciliary motility, leading to recurrent chest infections and infertility. We hereby present a case of 34 year male who presented to us with recurrent cough, shortness of breath and fever.*

Keywords: Kartagener's syndrome, Bronchiectasis, situs inversus.

1. Background

Kartagener's syndrome is a subset of a larger group 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.¹ 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. The estimated prevalence of PCD is about 1 in 30,000² though it may range from 1 in 12,500 to 1 in 50,000. In KS, the ultrastructural genetic defect leads to impaired ciliary motility which causes recurrent chest, ear/nose/ throat (ENT), and sinus infections, and infertility.

2. Case Report

We reported a case of a 34 year married male presented in emergency department with chief complaints of recurrent episode of cough with copious amount of expectoration since childhood with worsening of cough, fever and shortness of breath and not having children despite being married for 7 years. He has received anti-tubercular treatment for these complaints 5 years back with no relief. No family history of asthma and atopy was present. On examination, the B.P was

124/76 mm of Hg, pulse rate 93bpm, and oxygen saturation was 75% on room air. Clinical examination revealed grade 3 clubbing and apex beat on right side in fifth intercostal space. On auscultation, bilateral biphasic crepts were present with wheeze. Heart sounds were best heard on right side of chest. Chest X-ray (figure 1) revealed cardiac apex and aortic arch on right side suggesting dextrocardia. Sputum examination for acid fast bacilli and cbnaat was negative and culture was suggestive of pseudomonas aeruginosa sensitive to ciprofloxacin and cefepime. A high resolution computerized tomography (HRCT) scan (figure 2,3) of thorax suggestive of extensive cystic bronchiectatic changes in bilateral lung fields with bronchiectatic cyst in upper lobe. On ultrasound of abdomen Liver present on left side and measure 12.6 cm, spleen present on right side of body, pancreas was normal with head on left side and tail on right side. Suggestive of situs inversus totalis. Observing the presence of bronchiectasis, dextrocardia, and history of infertility his semen analysis was done and it revealed complete aspermia with normal seminal fluid content. Echocardiography showed dextrocardia with normal atrioventricular concordance without any structural abnormality. Xray of paranasal sinus revealed maxillary sinusitis. He was started on cefepime 2 gm iv bd for 10 days along with nebulization with salbutamol and budesonide.

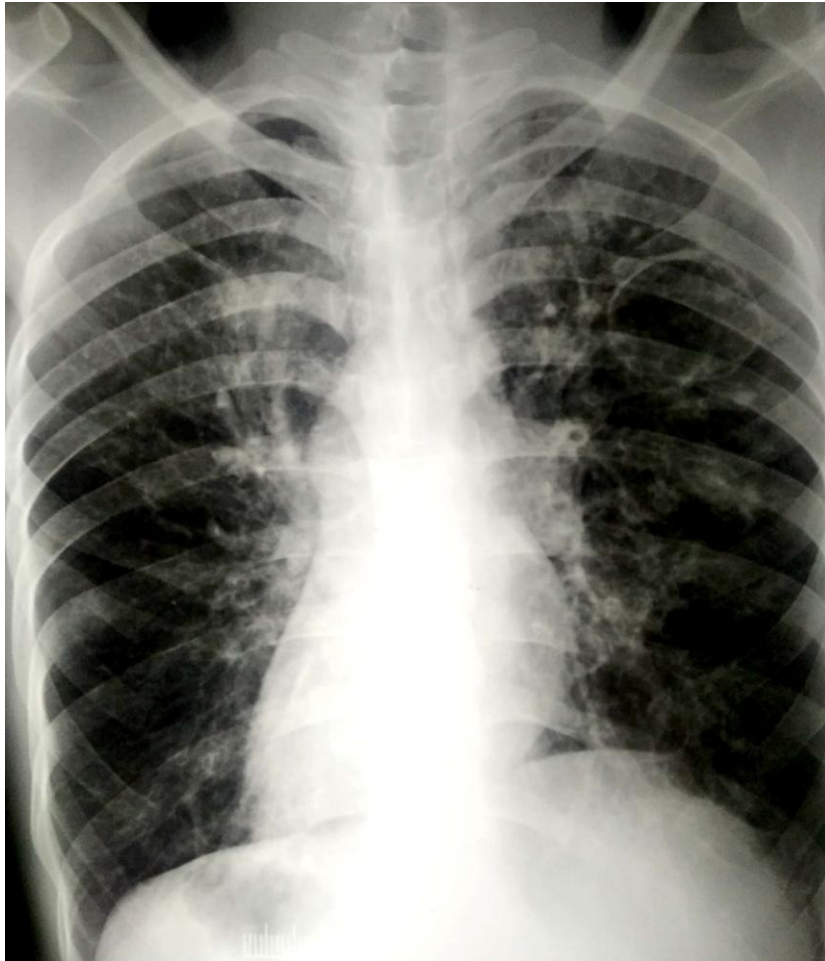


Figure 1: Chest xray showing dextrocardia with cavity in upper lobe

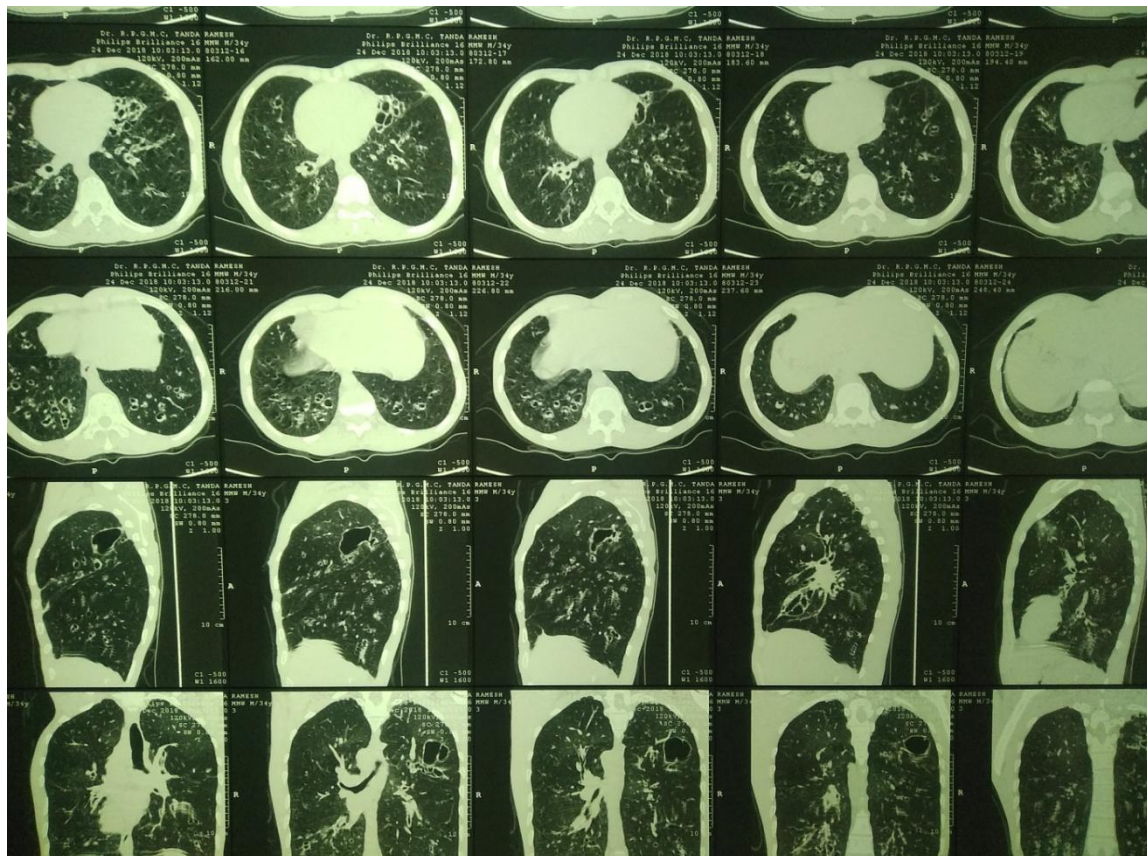


Figure 2: HRCT thorax showing extensive bronchiectasis.

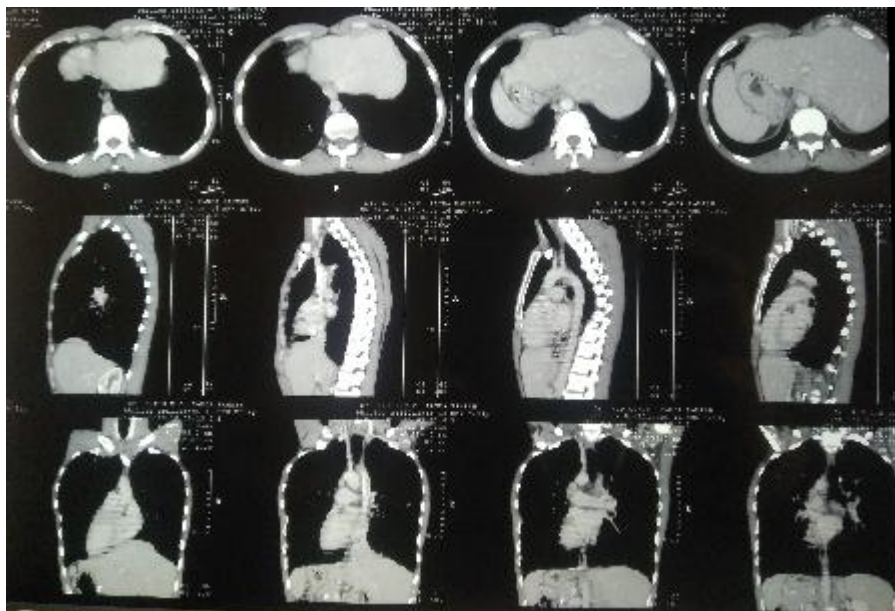


Figure 3: HRCT showing dextrocardia

3. Discussion

Disorders of ciliary motility may be congenital or acquired. Congenital disorders are labeled as PCDs. Nearly 50% of PCD patients has situs inversus. Such cases of PCD with situs inversus are known as Kartagener's syndrome.³

PCD is a phenotypically and genetically heterogeneous condition wherein the primary defect is in the ultrastructure or function of cilia.^{4,5} Such defects are identified in approximately 90% of PCD patients and involve the outer dynein arms, inner dynein arms, or both. The defect is in polypeptide species within axoneme of cilia, sperm flagella or in protein of ciliary membrane and matrix or protein for assembly of cilia.

Pathophysiologically, the underlying defect which leads to accumulation of secretions and consequent recurrent sinusitis, bronchiectasis, infertility, and situs inversus is the defective ciliary motility/immotility.⁶ The severity of symptoms and the age at which the condition is diagnosed is quite variable. Clinical progression of the disease is variable with lung transplantation required in severe cases.

Diagnostic criteria for this condition include⁷ 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.

There is no gold standard test available for diagnosis of the condition yet test like nasal nitric oxide test and ciliary beat pattern and frequency analysis using video recording and electron microscopy of cilia structure is done. In our case, however, we could not perform these tests and the diagnosis was essentially clinico-radiological.

4. Conclusion

Kartagener's syndrome is a rare disorder with constellation of bronchiectasis, infertility, situs inversus and sinusitis. The aim of this article was to present classic findings of Kartagener's syndrome, so that patients with recurrent sinusitis and cough with sputum can be evaluated for primary ciliary dyskinesia.

5. Declaration of patient consent

The authors certify that they have obtained all appropriate consent forms, in which patient has given their consent for images and clinical information to be reported in journal. The patient understand the name and initials will not be published and due efforts will be made to conceal identity, but anonymity cannot be guaranteed.

Conflict of interest: Nil

Source of support and sponsorship: Nil

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