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CASE REPORT KARTAGENER'S SYNDROME

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ABSTRACT
Background: Kartagener's syndrome is a subset of primary ciliary dyskinesia, an autosomal recessive inherited disordercharacterized by the clinical triad of chronic sinusitis, bronchiectasis, and situs inversus. Abnormal ciliary structure or function leading to impaired ciliary motility is the main pathophysiologic problem in Kartagener's syndrome. **Case Presentation:** A 47 year old female presented with productive cough from one month and hemoptysis. She suffered fromsimilar complaints 1 year back. Laboratory workup showed leukocytosis (>12000 cells) Clinical and imaging findings revealed chronic sinusitis, bronchiectasis, dextrocardia, and situs inversus and she was diagnosed with kartagener syndrome. She was given medical treatment in the form of antibiotics, antipyretics, mucolytics, inhaled bronchodilators and nebulizer solution. **Conclusion:** Patients with Kartagener's syndrome present with chronic recurrent sinopulmonary infections. As the correct diagnosis is often delayed by years, it may cause chronic respiratory problems with reduced quality of life. Genetic counseling and fertility issues should be addressed once Kartagener's syndrome is diagnosed.

KEYWORDS: Kartagener's syndrome, Primary ciliary dyskinesia, Chronic sinusitis, Bronchiectasis, Situs inversus.

BACKGROUND
Kartagener syndrome is a subset of primary ciliary dyskinesia, an autosomal recessive condition characterized by abnormal ciliary structure or function, leading to impaired mucociliary clearance. The syndrome includes the clinical triad of chronic sinusitis, bronchiectasis, and situs inversus. Normal ciliary function is critical for respiratory host defense and motility of sperm, and ensures proper visceral orientation during embryogenesis. In KS, 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

CASE PRESENTATION
A 47 year old patient presented with productive cough and hemoptysis from one month . She had similar episodes one to three years back along with history of chronic sinusitis. On physical examination, she was nourished, conscious, and oriented. Her blood pressure (BP) was 120/70 mmHg, pulse rate (PR) 80 beats per minute, respiratory rate (RR) 20 breaths per minute, and temperature (T°) 37.5 °C. Her arterial oxygen saturation (SaO2) was 95% with room air. A respiratory system examination revealed coarse crackles and scattered rhonchi on both basal lung fields. On cardiovascular examination, apex beat was felt on right fifth intercostal space along midclavicular line. Sputum for acid-fast bacilli (AFB) staining was negative for Mycobacterium tuberculosis. Serum chemistries were normal. A chest X-ray revealed cardiac apex and aortic arch on right side. CT thorax showed inverted position of chest and scanned abdominal organs S/O Situs inversus totalis with e/o cylindrical bronchiectasis seen in lingular segment of RUL and in left middle lobe. Patches of consolidation were seen in B/L lung fields suggesting infective etiology. Thus, a diagnosisof KS was made on the basis of clinical presentation and imaging features.

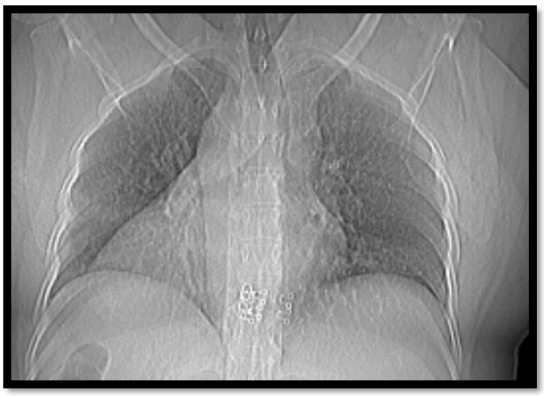

Fig. 1: Scanogram showing dextrocardia with right sided aortic arch.

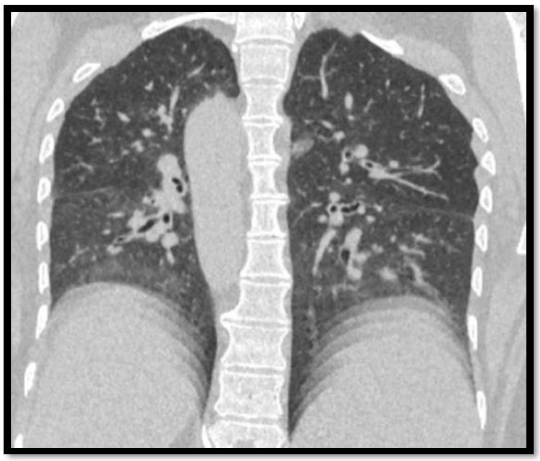

Fig. 2: Coronal HRCT image showing cylindrical bronchiectasis in B/L lung fields

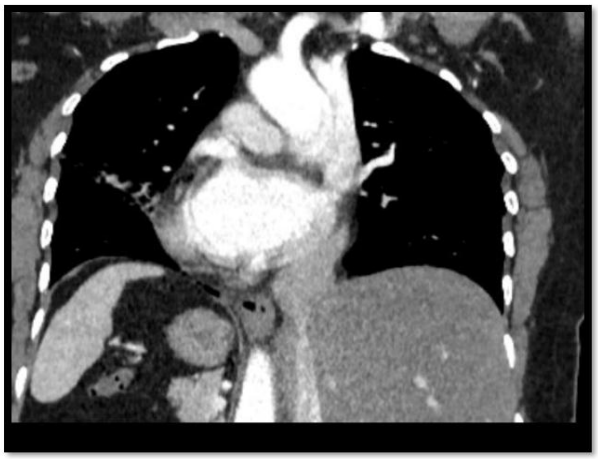

Fig. 3: Coronal CECT image showing dextrocardia and inverted position of scanned abdominal organs

DISCUSSION

KS is a rare, autosomal recessive ciliopathic disorder characterized by the clinical triad of chronic sinusitis, bronchiectasis, and situs inversus. Normal ciliary function is critical for respiratory tract host defense, sperm motility, and normal visceral orientation during embryogenesis. Lack or dysfunction of dynein arms, radial spokes, and microtubules of cilia are recognized structural and functional abnormalities of ciliary ultrastructures, encoded by the mutated genes DNAI1 and DNAH5. These faulty genes cause the cilia to be the wrong size or shape or move in the wrong way, making ciliary motility defective . 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 ora sibling, (b) alive but immotile spermatozoa, (c) absent or impaired tracheobronchial clearance, and (d) cilia showing characteristic ultrastructural defect on electron microscopy . Laboratory screening tests include exhaled nasal nitric oxide level determination and saccharin test for assessing nasal epithelial mucociliary function. High-speed video microscopy for assessing ciliary beat frequency and pattern, transmission electron microscopic for detecting ultrastructural ciliary defect, and genetic testing for DNAI1 and DNAH5 mutations are confirmatory laboratory tests.

CONCLUSION

As there is no easy, reliable non-invasive diagnostic test for KS and the correct diagnosis is often delayed by years, it may cause chronic respiratory problems with reduced quality of life. Genetic counseling and fertility issues should be addressed once KS is diagnosed.

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