“HEART OFF-SITE MAKING DIAGNOSIS RIGHT”: A CASE REPORT ON KARTAGENER SYNDROME

(GENERAL MEDICINE)

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BACKGROUND

Kartagener syndrome is a subset of primary ciliary dyskinesia, an autosomal recessive inherited disorder characterized 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 syndrome.

CASE PRESENTATION

A 15-year-old boy came with recurrent episodes of nasal congestion with itching and paranasal discomfort, and productive cough since 2 years, aggravated since a week.

❖On examination vital parameters were within normal limits and grade 2 clubbing was present.

❖On inspection –nasal mucosa was congested ,nasal cavity was filled with mucus.
❖On palpation –B/L maxillary sinus tender. Apex beat felt in right 5 intercostal space 1cm medial to midclavicular line.
❖On Auscultation- B/L, Crepitations all over the lung fields present. Heart sounds best heard on right side of chest.
❖Clinical and imaging findings revelation: Ecg – dextrocardia, chest x ray – cardiac apex and aortic arch on right. X-ray of paranasal sinus – mucosal thickening and haziness of maxillary sinus.

❖USG abdomen –normal liver, gallbladder on left, spleen on right side.HRCT chest shows tractional bronchiectatic changes, hematological parameters shows rise in total counts.
❖2D ECHO – was consistent with dextrocardia, all 4 chambers were normal with EF -65%.
❖Semen analysis done at time of discharge shows Azoospermia. Audiogram shows B/L mild conductive hearing loss.
❖Doppler study confirmed situs inversus of aorta and inferior venacava.
❖Electron microscopic confirmation of the ultrastructural ciliary defect showing primary ciliary dyskinesia.
❖Sputum for gram stain, AFB ,CBNAAT negative for TUBERCULOSIS ,SPUTUM CULTURES showed klebsiella.

Patient was treated with antibiotics, mucolytics, bronchodilators, chest physiotherapy and other supportive treatment.

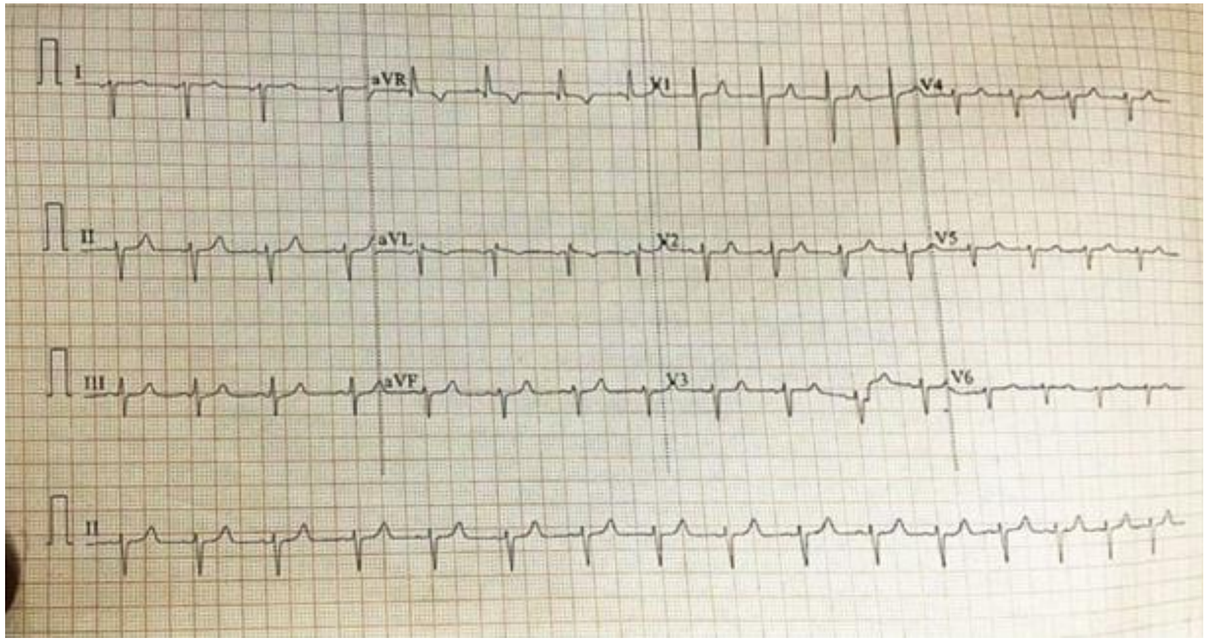

Fig 1: ECG showing dextrocardia

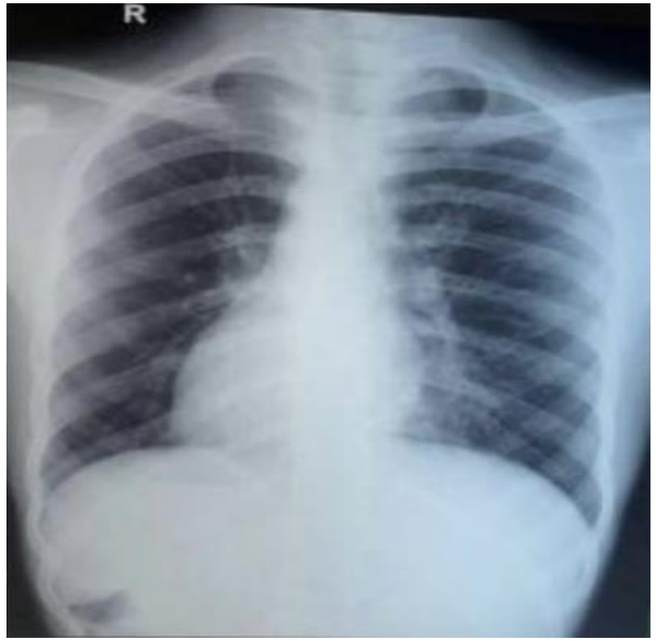

Fig 2: Chest X-ray – cardiac apex and aortic arch on right side

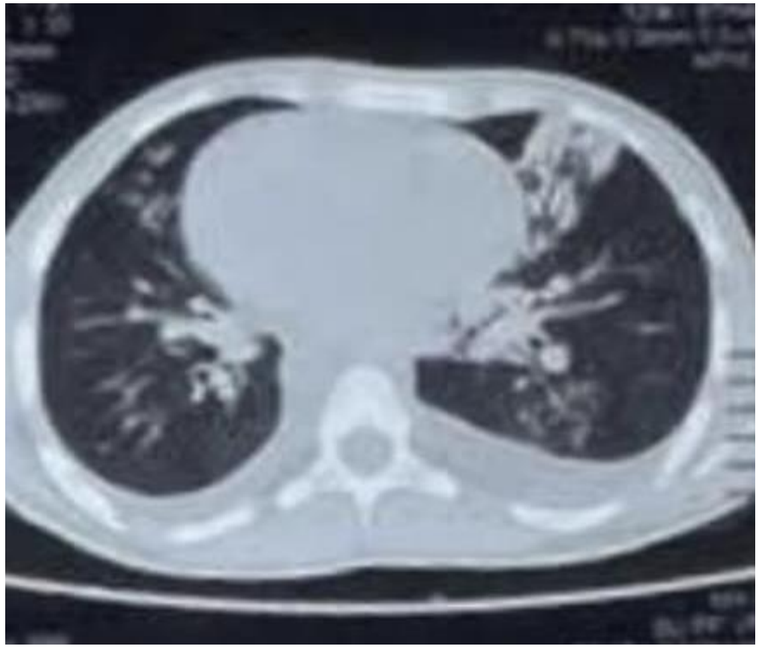

Fig 3: HRCT scan chest shows tractional bronchiectatic changes

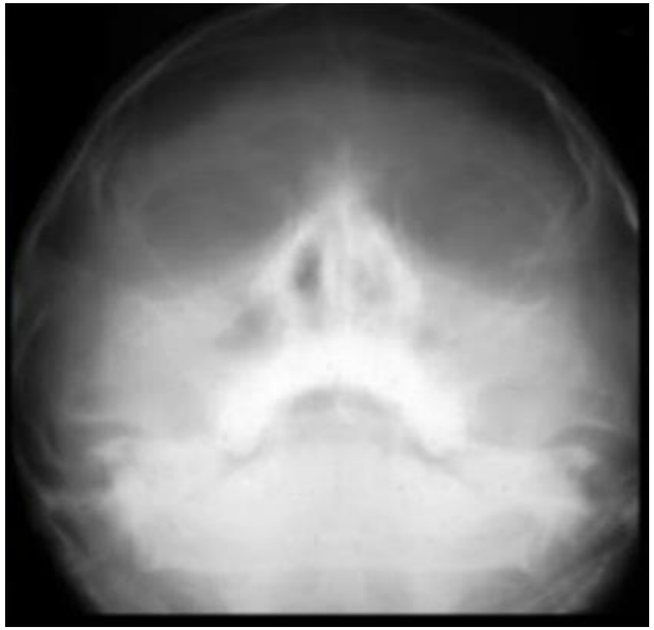

Fig 4: X-ray of Paranasal sinus viewing mucosal thickening and haziness of maxillary sinus

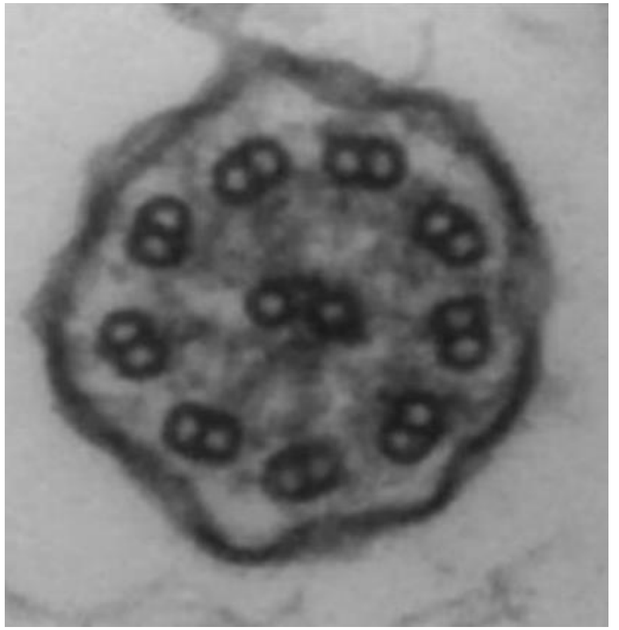

Fig 5: Electron microscopy showing PCD

DISCUSSION

Kartagener syndrome results due to the genetic defect in the ciliary structure leads to impaired mucociliary clearance and the resulting symptoms. Disorders of ciliary motility may be congenital or acquired. Congenital disorders are labeled as Primary Ciliary Dyskinesias. Nearly 50% of PCD patients have situs inversus. Such cases of PCD with situs inversus are known as Kartagener's syndrome. 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. The severity of symptoms and the age at which the condition is diagnosed is quite variable, even though the symptoms are present from birth. KS may be associated with reversible airflow obstruction.

Apart from the triad of KS- “chronic sinusitis, bronchiectasis, and situs inversus.”, two types of tests done for diagnosis of PCD – screening tests (exhaled nasal nitric oxide measurement which is usually low in PCD, and saccharin test to assess mucociliary function of nasal epithelium) and diagnostic tests (ciliary beat pattern and frequency analysis using video recording, and electron microscopic confirmation of the ultrastructural ciliary defect). The samples for these tests for examining motility and ultrastructure of cilia may be obtained by biopsy of nasal mucosa and laparoscopic biopsies of tubal mucosa in females.

Male patients with KS invariably present infertility, while women present reduced fertility. 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. In cases where there is no sperm motility, ICSI may be the most appropriate treatment, if sperm motility is present, a trial of IVF should be considered.

Key strategies for treatment include **airway clearance** techniques like chest physiotherapy and mucolytics, targeted antibiotics for infections, and bronchodilators for respiratory support. Nasal irrigation and surgery may be needed for chronic sinusitis. Regular monitoring of lung function, vaccinations and multidisciplinary care are crucial.

CONCLUSION

As the patient has sinusitis, bronchiectasis, situs inversus - the triad of Kartageners syndrome. A diagnosis was made based on clinical presentation, imaging features and confirmatory test by nasal biopsy and electron microscopy showing absence of dynein arms in the ultrastructure of cilia. Prophylactic antibiotics, hand hygiene practices, adequate ventilation, reassurance, psychiatric counselling constitutes the main stay to these patients. Genetic counselling and infertility issues were also addressed.

REFERENCES

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❖ Harrison, T. R., Jameson, J. L., Kasper, D. L., Longo, D. L., Fauci, A. S., Hauser, S. L., & Loscalzo, J. (Eds.). (2023). Harrison's principles of internal medicine (21st ed.). McGraw-Hill Education.