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A Rare Presentation of Kartagener Syndrome: Case Report

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Abstract:- 66/M who presented to us with recurrent H/o lower respiratory tract infections and headache since childhood.The patient also have a history of taking over the counter medications rather than consulting a physician and never been to hospital.When we heard the apex beat on Right 5 th intercostal space , it arouse suspicion of dextrocardia and situs inversus which were then confirmed by imaging investigations. Patient was then subjected to genetic study which showed mutation of DNAH5. The rarity of this case is of primary ciliary dyskinesia with preserved fertility,as in this case the patient had 5 children

I. INTRODUCTION

KS – an autosomal-rescessive disease with triad of chronic sinusitis+bronchiectasis+situs inversus.This disease has Incidence of 1in 30,000 live births.[1]The instance of 66/M presenting with KS is described here due to rarity of its presentation.

II. REPORT OF THE CASE

A 66/M presented with c/o recurrent headache × 14 days, aggravated x past 2 days. cough with expectoration x 10 days ,c/o recurrent headache .

The patient had similar complaints recurrently occurring since childhood for which he used only over the counter medications for symptomatic relief ,No H/O Smoking ,No comorbidites.

III. EXAMINATION

A. General Examination

- Concious,oriented,
- Pan digital clubbing+ grade 3
- Vitals Stable

B. Systemic Examination

- **R /S** – crests present bilaterally,diffusely all over the chest
- **CVS**-S1, S2+ apical impusle in **right 5 th ICS.**
- **CNS**-NFND
- **P/A-** No organomegaly,bowel sounds +

C. Examination of Paranasal Sinuses

Tenderness present in maxillary sinus and frontal sinus.

D. Investigation:

- **CHEST X-RAY** showed dextrocardia .
- **HRCT CHEST-** dextrocardia and features of bronchiectasis
- **CT PNS** – maxillary and frontal sinusitis
- **BRONCHOSCOPY**-Normal
- **USG& CT ABDOMEN - situs inversus.**As it met the triad of KS, probable diagnosis was made.
- **SPERM ANALYSIS** - normal **Although infertility is common (50-70%) in kartagener syndrome ,this patient has 5 children which is rare.**
- **GENETIC STUDY showed mutation of DNAH5**

➢ *X RAY CHEST*

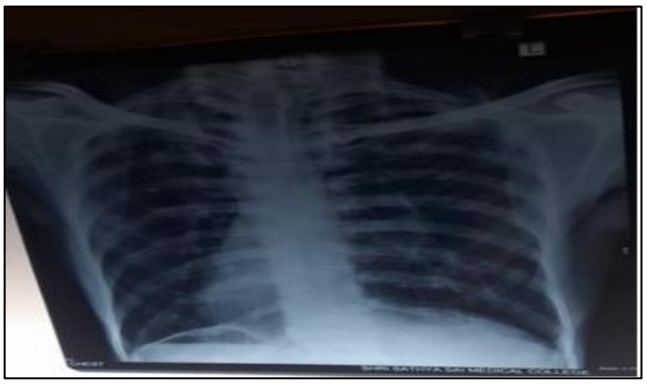

Fig 1: X-Ray Chest

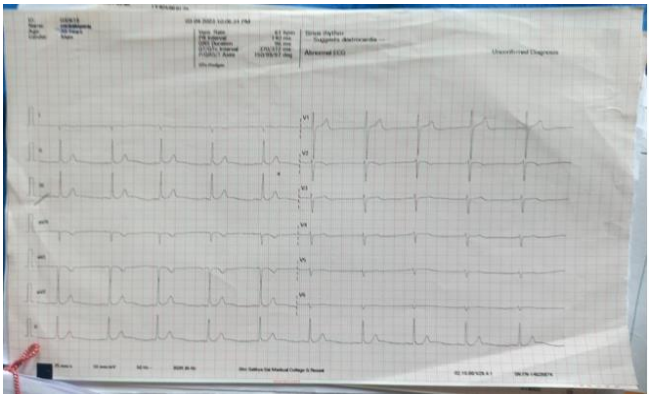

Fig 2: Patient

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Fig 3: ECG

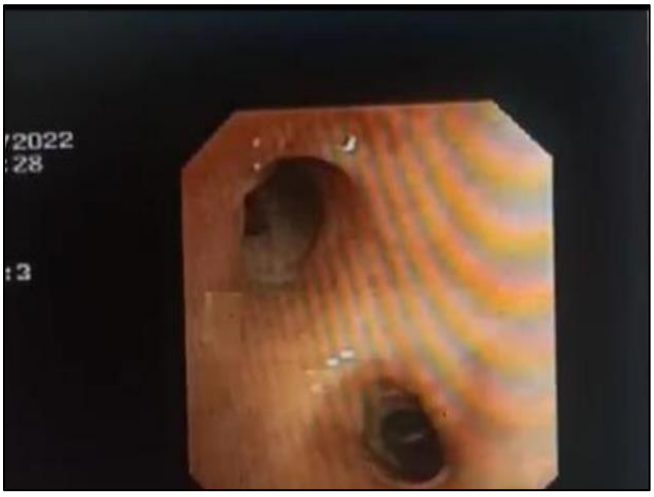

Fig 4: Bronchoscopy

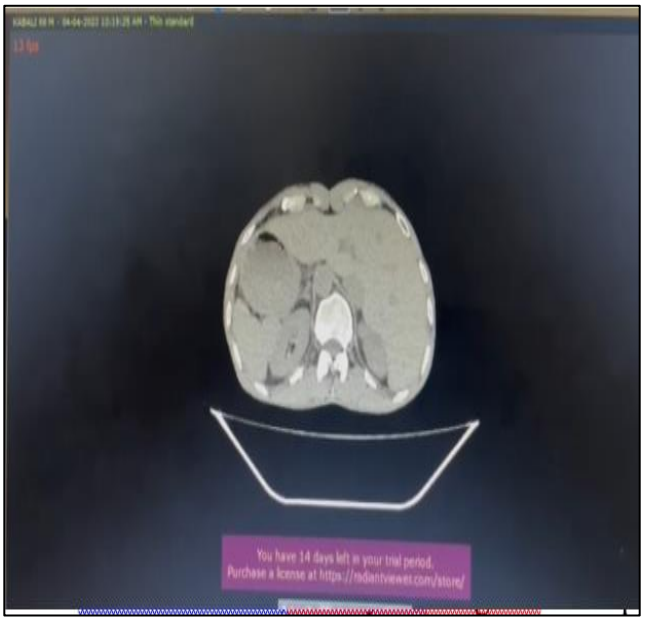

Fig 5: X-Ray PNS

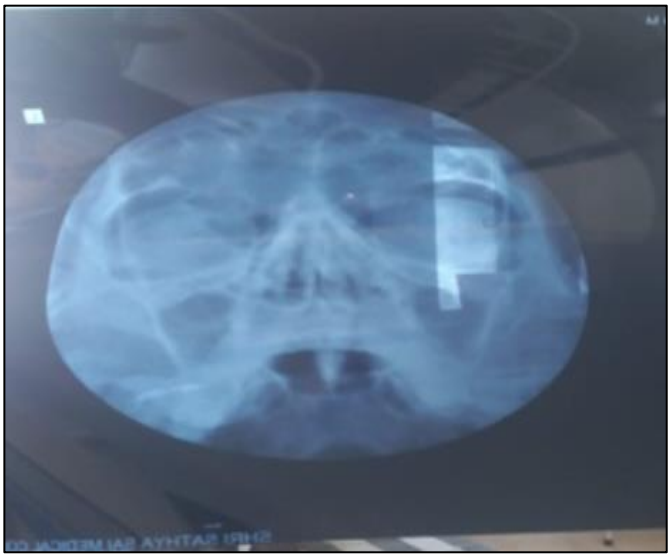
Fig 6: CT Abdomen

➢ *Treatment*

The patient was started with AMOXICLAV ,mucolytics&regular chest physiotherapy was given. Patient was adviced for flu vaccines.

IV. DISCUSSION

KS is a autosomal-recessive disorder with triad of chronic sinusitis+bronchiectasis+situs inversus. This disease has an incidence of 1in 30,000 live births. Chromosomal abnormalities reveal absent or defective function of dynein arms,microtubules . Mutation in DNAI1 and DNAH5 genes leading to defective ciliary motility. [2] This abnormality in ciliary motility during embryonal developmental stage predisposes to left– right lateralisation abnormalities—isolated dextrocardia or situs inversus totalis - transpositions of thoracic+abdominal organs,present in addition .The diagnostic criteria for kartagener syndrome is h/o recurrent chronic bronchial infection and rhinitis along with 1 or more of the below-findings (1) situs-inversus or dextrocardia in a patient/sibling (2) alive still spermatozoa immotility (3) absent/impaired tracheobronchial clearance (4) cilia showing characteristic ultra-structural defect on E/M .

V. ROUTINE INVESTIGATIONS TO BE SENT

1.usg abdomen showing situs inversus 2.x ray pns 3 chest roentogenogram PA view shows dextrocardia with right-sided aortic arch. 4.EM - ultrastructural ciliary defect, 5. genetic testing

➢ *Treatment*

Includes chest physiotherapy, mucolytics & antibiotics. Influenza and pneumococcal vaccines are adviced.[3].

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VI. CONCLUSION

Infertility may not always be present in kartagener syndrome as the patient in my case has five children(Rarity of this case).Any case with recurrent LRTI and sinusitis, genetic causes should be ruled out.Early diagnosis and appropriate treatment(low dose Long term antibiotics) improves quality of life of such patients

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