IOSR Journal of Dental and Medical Sciences (IOSR-JDMS)
e-ISSN: 2279-0853, p-ISSN: 2279-0861.Volume 21, Issue 8 Ser.1 (August. 2022), PP 01-05
www.iosrjournals.org

A Case of Primary Ciliary Dyskinesia with Kartageners Syndrome

Dr.Sindhuja P.G Dr.Gopichand M.D, Prof and HOD;
Dr.Aruna Asst Prof; Dr.Suresh kumar Asst Prof; Dr.Bhanu Naik Asst Prof

Date of Submission: 21-07-2022 Date of Acceptance: 05-08-2022

I. Introduction

- Primary ciliary dyskinesia is a rare autosomal recessive disorder characterised by impaired ciliary function leading to chronic sinopulmonary disease, persistent middle ear infection, situs inversus and infertility.

II. Case Report

- A 45 year old male patient, presented to department of Pulmonary Medicine, GGH, Vijayawada with complaints of cough with expectoration, breathlessness and fever since 7 days.
- History of recurrent episodes of similar complaints and rhinitis are present since childhood which are on and off.
- No comorbidities and never smoker.
- Second degree consanguineous marriage and has no children.

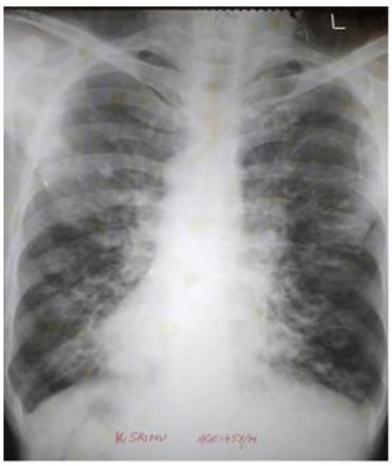
ON EXAMINATION

- Clubbing is present.
- Saturation is 82% with room air and rest of the vitals are normal.
- Deviation of nasal septum towards left side
- Tenderness over bilateral maxillary sinus region
- Apex beat-on right side 5th ICS 1cm medial to midclavicular line.
- On auscultation-coarse biphasic crepitations heard in bilateral mammary ,infra axillary, infrascapular and interscapular regions.

CHEST X RAY

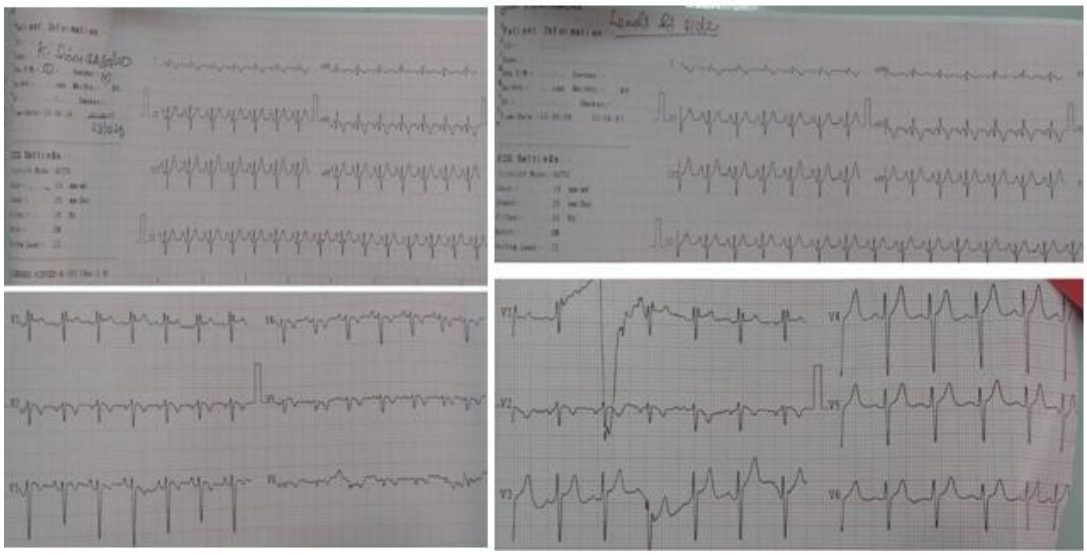
Showed cystic lesions in bilateral lower zones with dextrocardia.

CHEST X RAY



ECG
ECG revealed features of dextrocardia with right axis deviation and poor R wave progression and findings were normal when chest leads are placed on right side.

ECG

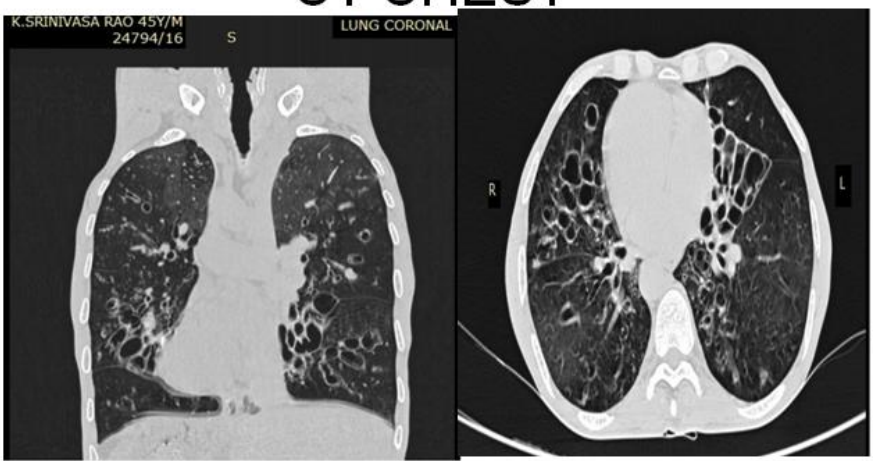


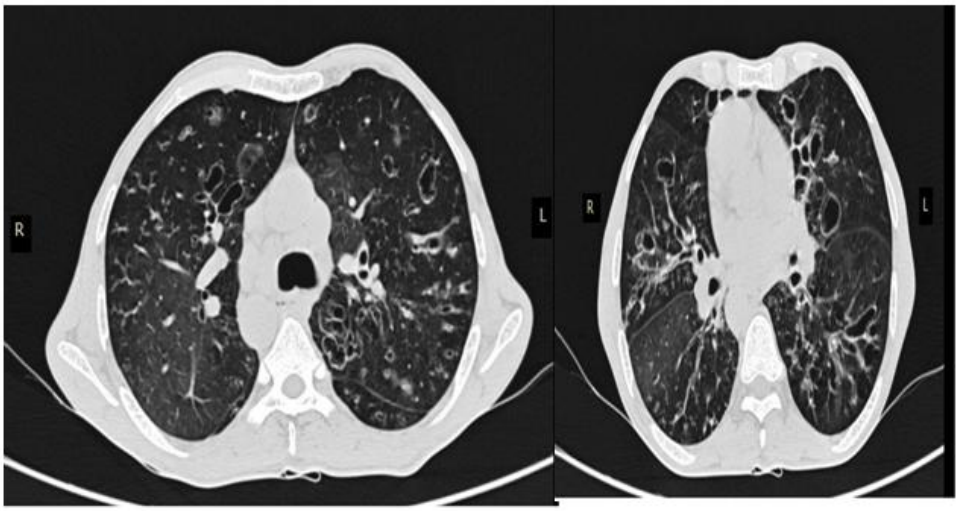
USG ABDOMEN
Suggestive of Situs Inversus

PFT
Showed moderate obstructive and mild restrictive pattern.

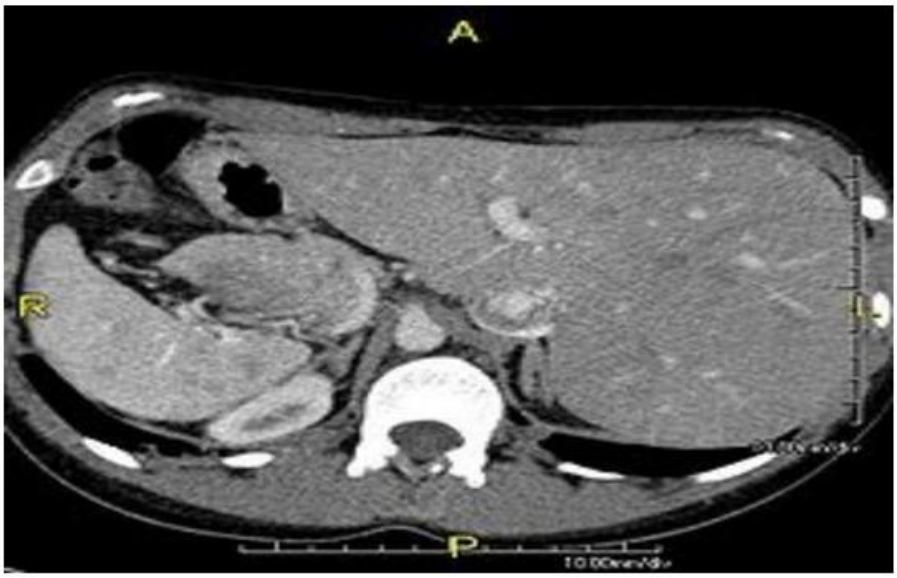
CT CHEST
Showed cystic and tubular bronchiectasis predominantly in bilateral lower lobes, right middle and lingular lobes and situs inversus totalis with right sided aortic arch, dextrocardia and total transpostion of abdominal viscera.

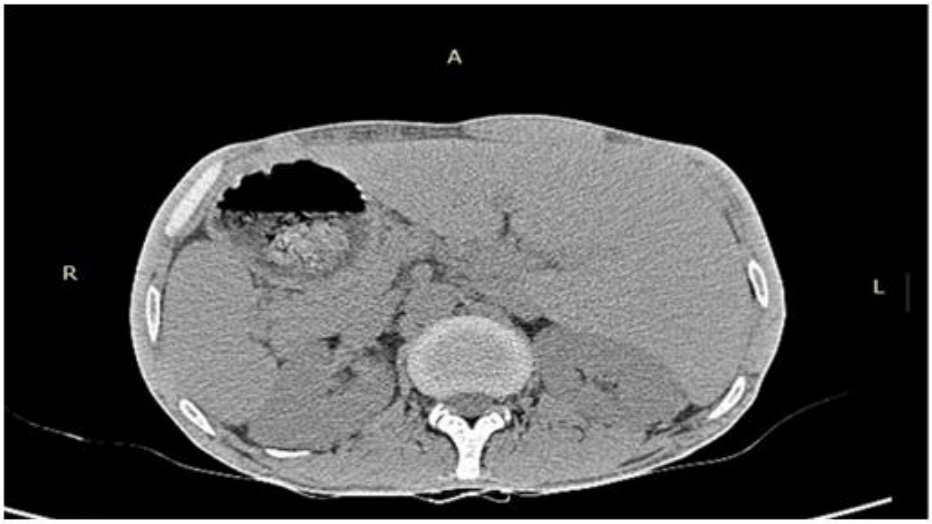
CT CHEST





CT ABDOMEN

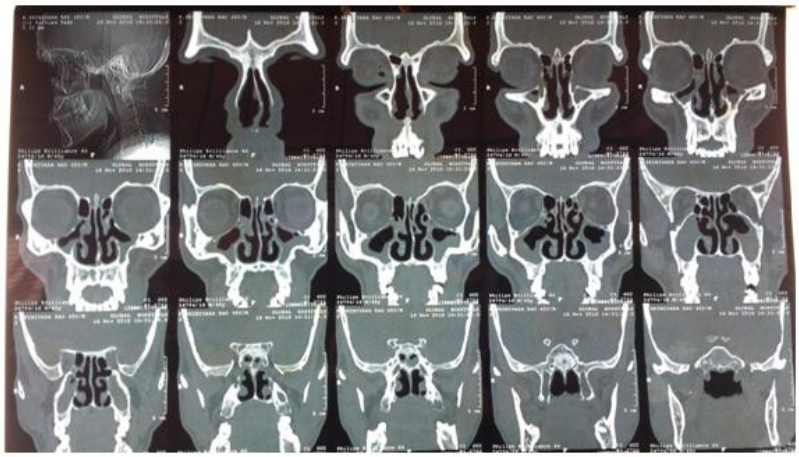


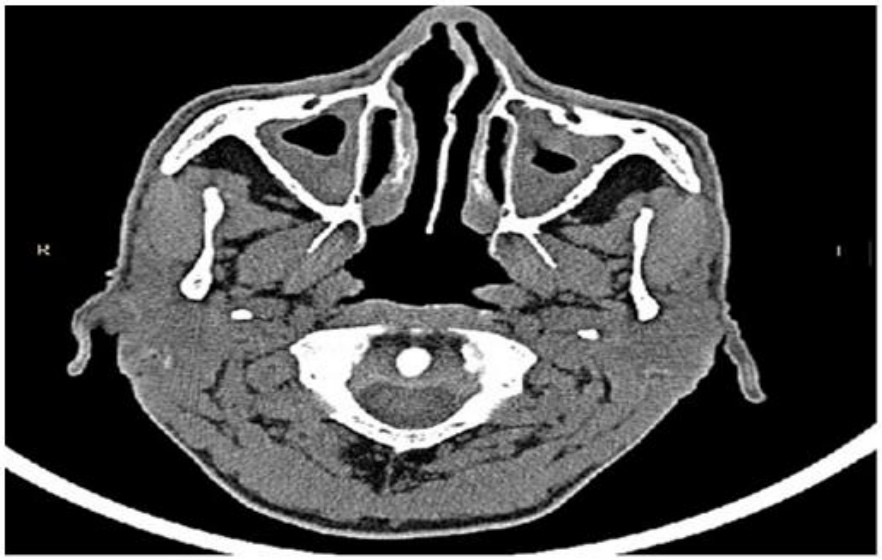


CT PNS

- CT PNS showed mucosal thickening suggestive of chronic sinusitis involving bilateral maxillary, sphenoid and ethmoid sinuses. Frontal sinuses are under pneumatised.

CT PNS





- Saccharin test revealed transit time of 1hour 15 minutes.
- Seminal fluid analysis showed normal volume and count with only 15% of sperms actively motile, 55% non motile and 30% sluggishly motile sperms.

III. Discussion

- All congenital ciliary disorders are labelled as Primary ciliary dyskinesia(PCD).Rare, ciliopathic, Autosomal Recessive genetic disorder that causes defects in the action of cilia lining the respiratory tract and fallopian tube, as well as in the flagella of sperm cells.
- The phrase "IMMOTILE CILIARY SYNDROME" is no longer favored as the cilia do have movement, but may be inefficient or unsynchronized. When accompanied by the combination of situs inversus (reversal of the internal organs), chronic sinusitis, and bronchiectasis, it is known as KARTAGENER SYNDROME (only 50% of primary ciliary dyskinesia cases include situs inversus).
- The reported frequency is 1 per 26,000- 40,000 live births.No racial and sexual predeliction reported.
- In abnormalities of cilia, structural abnormalities are the most common , although abnormalities of radial spokes and microtubules can occur.

- Specific investigations are functional assessment by mucociliary clearance test(saccharin test), Ciliary beat frequency(CBF) and ciliary beat pattern(CBP), Nasal Nitric oxide measurement, Cell culture system.
- Structural assessment by Electron microscopy and genetic analysis are
- No specific treatment available as the cause is genetic. Symptomatic management include antibiotics, chest physiotherapy, mucolytics and bronchodilators.
- In case of persistent symptoms, surgeries like Tympanostomy,FESS,lobectomy,lung transplantation may be helpful.

IV. Conclusion

- The progression of lung disease varies and is affected by the time of diagnosis, the ability of medical treatment to control symptoms, and the prevention of complications that affect the quality of life.
- Regular spirometry, chest imaging and sputum cultures to be done to slow down progression and extent of lung disease by early diagnosis

References

[1]. Lucas JS, Leigh MW. Diagnosis of primary ciliary dyskinesia: searching for a gold standard. Eur Respir J 2014; 44: 1418–1422.
[2]. Boon M, Smits A, Cuppens H, et al. Primary ciliary dyskinesia: critical evaluation of clinical symptoms and diagnosis in patients with normal and abnormal ultrastructure. Orphanet J Rare Dis 2014; 9: 11.
[3]. Werner C, Lablans M, Ataian M, et al. An international registry for primary ciliary dyskinesia. Eur Respir J 2016; 47: 849–859
[4]. Afzelius BA. A human syndrome caused by immotile cilia. Science 1976; 193: 317–319

Dr.Sindhuja, et. al. "'A Case of Primary Ciliary Dyskinesia with Kartageners Syndrome." *IOSR Journal of Dental and Medical Sciences (IOSR-JDMS)*, 21(08), 2022, pp. 01-05.