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A case report: Kartagener's Syndrome

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Abstract
A rare autosomal recessive disorder Kartagener's syndrome is a classic triad of sinusitis, situs inversus and bronchiectasis which is a type of primary ciliary dyskinesia (PCD). We present a case of a 34 years old male with sinusitis, situs inversus and bronchiectasis and diagnosis of this rare congenital disorder is important in the overall prognosis of the syndrome.

Keywords: kartagener's syndrome, bronchiectasis, sinusitis, situs inversus, primary ciliary dyskinesia

Introduction
Siewert first described the combination of situs inversus, chronic sinusitis, bronchiectasis and ciliary dyskinesia as a Kartagener's syndrome (KS) in 1904 [1]. It is a genetic condition with an autosomal recessive inheritance [2, 3]. The estimated prevalence of primary ciliary dyskinesia (PCD) is about 1 in 30,000, [3] though it may range from 1 in 12,500 to 1 in 50,000.[2] In KS, the ultrastructural genetic defect leads to impaired ciliary motility which causes recurrent chest, ear, nose, throat, and sinus infections, and infertility [4]. Male patients with this syndrome are almost invariably infertile because of non-motile spermatozoa. The non-motility is due to variety of ultrastructural defects in respiratory cilia and sperm tail [5]. Also, although unproven, it seems likely that early diagnosis is important for the preservation of pulmonary function, quality of life and life expectancy in this disease [6, 7].

Case report
A 34 years old male presented to pulmonary medicine OPD at C. U. Shah medical college with history of recurrent cough with expectoration whitish-yellowish in colour and muco-purulent in nature, nasal discharge and obstruction, difficulty in breathing on heavy exertion with episodes of fever, weight loss of about two to three kg over past six month, long term use of antibiotics for past one to two years. He also taken anti tuberculosis treatment for pulmonary koch's 20 years back. He was smoker and tobacco chewer.
On examination, he was a moderately built and nourished, having coughing and short of breath. He was conscious, co-operative and well oriented for time, place & person. He has no pallor, icterus, cyanosis, lymphadenopathy, JVP raised and oedema feet with clubbing grade II.
On auscultation of the chest revealed diffuse bilateral ronchi with scattered crepitations over both infrascapular regions. Chest expansion was reduced.
We found normal cardiac sounds on right side of chest. Also, Apex beat was palpable over the fifth intercostal space on the right side of chest and rest of the physical and systemic examination findings were normal.
On investigation, chest X-ray PA view (Figure - 1) had cardiac shadow on the right side so that it would suggested that dextrocardia.

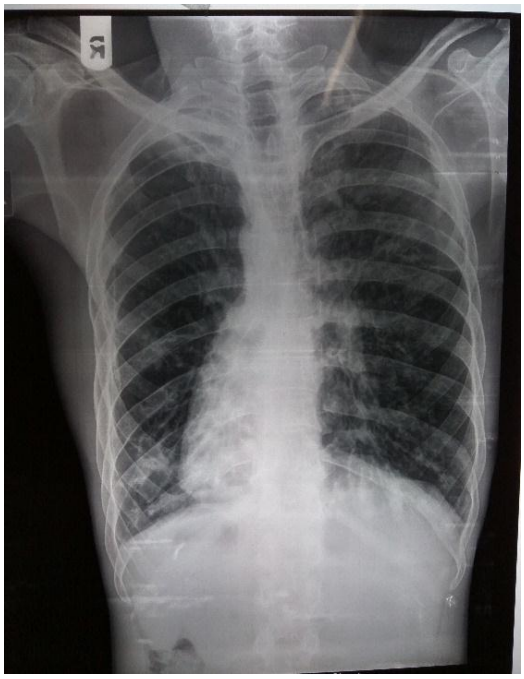

Fig 1: X-ray chest PA view

Electrocardiogram (ECG) was normal on right sided chest leads (Figure - 2) and both right and left sided chest leads, which revealed inverted *"*P*"* waves in L1 and AVL.

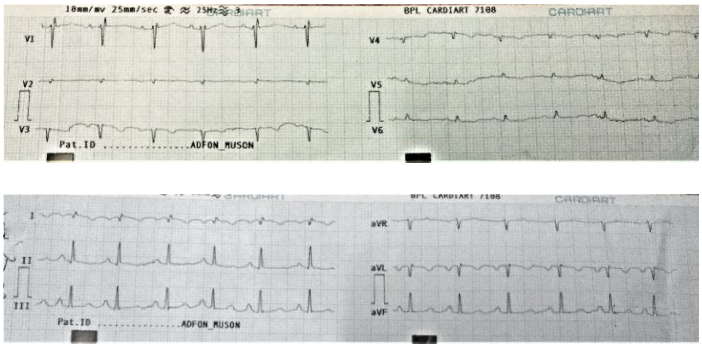

Fig 2: Electrocardiogram

X-ray of paranasal sinuses showed finding of sinusitis.

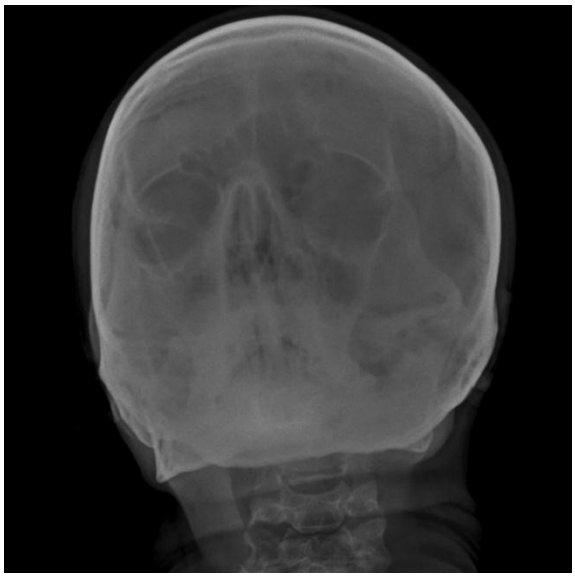

Fig 3: X-ray of paranasal sinuses

HRCT of thorax showed finding of bronchiectasis. (Fig-4)

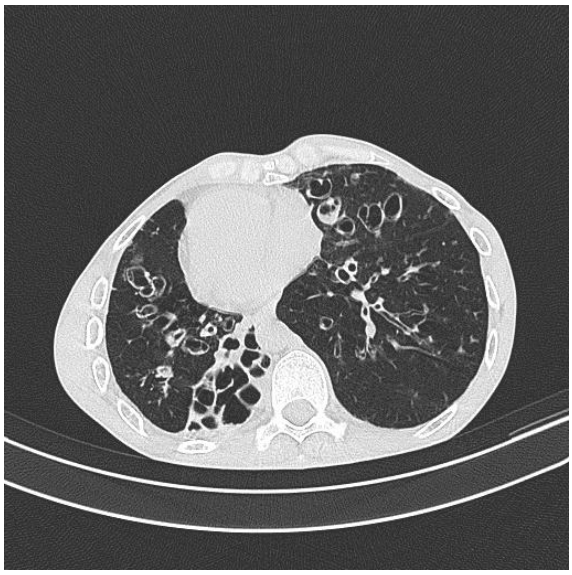

Fig 4: HRCT of thorax

Ultrasound of abdomen showed spleen on right side and liver on left side. (Fig - 5)

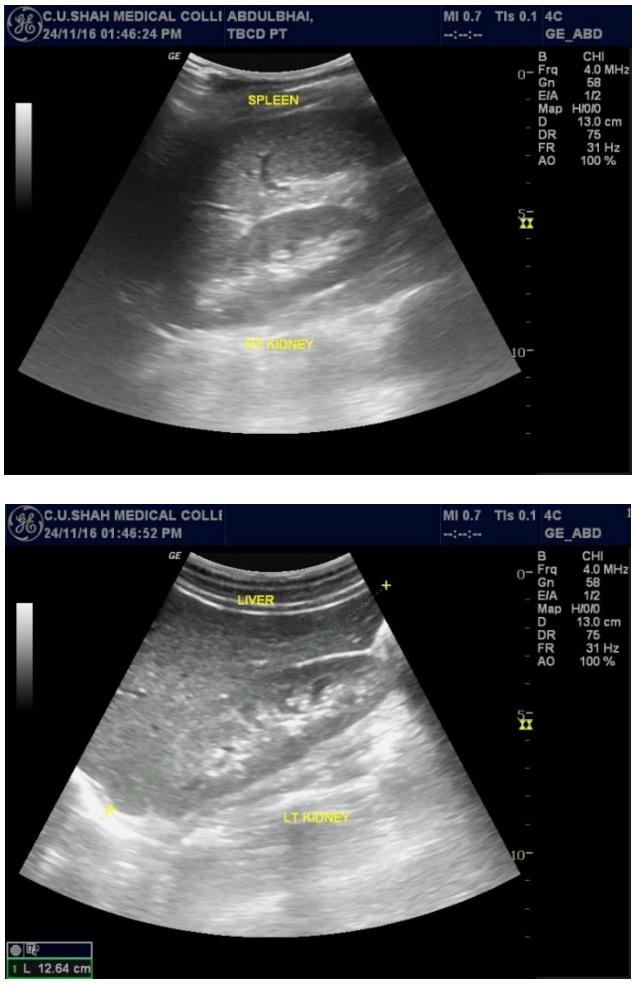

Fig 5: Ultrasound of abdomen.

Sputum culture had grown Klebsiella pneumoniae on MacConkey culture media.

Treatment was intended to relieve symptoms and prevent irreversible complications. Third generation cephalosporin with clavulanate antibiotics were given for acute Klebsiella pneumoniae infection and broad spectrum antibiotic was given for prophylaxis. Inhaled corticosteroids, mucolytics, bronchodilators, influenza and pneumococcal immunisation and chest physiotherapy manoeuvers were benefited to patient.

Discussion

Kartagener's syndrome is a very rare congenital malformation. Disorders of ciliary non-motility may be congenital or acquired. Congenital disorders are labelled as PCDs. Nearly 50% of PCD patients have situs inversus. Such cases of PCD with situs inversus are known as Kartagener's syndrome [8].

Defects in the ciliary component cause abnormal ciliary movements, resulting in impaired mucociliary clearance and manifesting as recurrent and / or persistent sinopulmonary infections.

Dynein arm defects manifest as a total or a partial absence of either both inner or both outer dynein arms or involve just the inner or outer arms. Sometimes, shortened dynein arms are the only defect. Recent studies show differential functions of both inner and outer dynein arms and correlate ciliary beat

frequency directly with the number of outer dynein arms. The ciliary beat frequency is not correlated with the number of inner dynein arms.

Microtubular transposition defects occur in the form of absence of the central pair of tubules with transposition of the outer doublet to the centre. Other defects, such as ciliary aplasia, ciliary disorientation [8], mal-aligned central pair of microtubules in adjacent cilia and basal body abnormalities may occur after viral infections, making it unclear if they are primary or secondary defects [9].

Moreover, in some patients with typical clinical manifestations of PCD and low levels of nasal nitric oxide, the ciliary ultrastructure may appear normal, suggesting functional abnormalities because of other defects in ciliary components. This includes few patients with biallelic mutations *in* DNAH11 [10].

Studies have confirmed that ciliary beat pattern is associated with specific ultrastructural defects in PCD [11].

Generally, a child must inherit faulty genes from both parents to have PCD. These genes affect how cilia grow and function. Cilia are tiny, hair-like structures that line the airways. The airways include your nose and linked air passages, mouth, larynx, trachea, bronchi, and their branches. Cilia move mucus (a slimy substance) through airways toward mouth to be coughed or sneezed out of body. The mucus contains inhaled dust, bacteria and other small particles. Faulty genes may cause the cilia to be the wrong size or shape or move in the wrong way. Sometimes the cilia are missing altogether. If the cilia don't work well, bacteria stay in airways. This can cause breathing problems, infections and other disorders.

With PCD, this process is very complex. Researchers are still learning how the disease is inherited and which genes are involved.

The disease also affects people from all racial and ethnic groups. Some people with PCD have breathing problems from the moment of birth. However, other people can go through all or most of their lives without knowing that they have the disease.

Common signs, symptoms and complications with PCD include the following:

- Sinuses
- Chronic nasal congestion
- Runny nose with mucus and pus discharge
- Chronic sinus infections
- Ears:
- Chronic middle ear infections
- Hearing loss
- Lungs:
- Respiratory distress in new born
- Chronic cough
- Recurrent pneumonia
- Collapse of part or all of a lung

The diagnostic criteria recommended for this syndrome are 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 or a sibling, (b) living but non-motile spermatozoa, (c) absent or impaired tracheobronchial clearance [14].

The symptoms and severity of PCD varies from person to person and it also shows seasonal variation. Diagnosing PCD in children can be hard. This is because some PCD symptoms such as, ear infections, chronic cough and runny nose are common in children, even if they don't have PCD. Also, the disease may be confused with another condition, such as cystic fibrosis.

Most infertile patients with KS have a normal spermatozoid count, but with a structural defect and a complete lack of motility [15]. Infertility in male with KS is due to diminished sperm motility, while in females it is due to defective ovum transport because of dyskinetic motion of oviductal cilia, suggesting that the ciliated endosalpinx is essential for human reproduction [16].

In treatment part of this syndrome, standard treatments for breathing and lung problems in people who have PCD are chest physical therapy (CPT), exercise, and antibiotics. Continuous or intermittent, oral or intravenous antibiotics treat upper and lower airway infections. Long-term low-dose prophylactic antibiotics may be necessary in children [17].

Hemophilus influenzae and Staphylococcus aureus are the common organisms.[18] Obstructive lung disease / bronchiectasis should be treated with inhaled bronchodilators, mucolytics and chest physiotherapy. Influenza and pneumococcal vaccination should be encouraged.[17]

Surgical care involves tympanostomy tubes that will reduce recurrent infections and conductive hearing loss. Endoscopic sinus surgery and the formation of a nasal antral window underneath the inferior turbinate may afford a transient improvement in upper and lower respiratory tract symptoms [17].

People who have PCD may develop a serious lung condition called bronchiectasis. If bronchiectasis severely affects part of lung, surgery may be used to remove that area of lung. In very rare cases, if other treatments haven't worked, lung transplant may be an option for severe lung disease. Taiana and associates reported successful pulmonary operations in a 25 years old man, consisting of left lower lobectomy, lingulectomy and anterior segmentectomy of the left upper lobe [19].

Conclusion

Kartagener`s syndrome is a rare condition, occasionally picked up by the physician. It may prove challenging if high index of suspicion is not made.

Thorough clinical evaluation, adequate investigations, proper timely management, etc. go a long way in alleviating the patient`s symptoms, reducing the morbidity and improving the quality of life.

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