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A Case Report on Kartagener Syndrome or Primary Ciliary Dyskinesia – A Rare Genetic Disorder of Ciliary Motility

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A B S T R A C T

Kartagener syndrome (KS) is a rare genetic disorder of autosomal recessive inheritance which is manifested by the classical triad of bronchiectasis, chronic sinusitis, and situs inversus along with infertility in males and reduced fertility in females due to impaired or defective ciliary movement and comes under the types of primary ciliary dyskinesia (PCD). This study aims to contribute to the incidence of Kartagener syndrome by reporting this rare genetic disorder. We enlighten a case of a 65-year-old female, a resident of Aligarh, U.P., India, who presented with recurrent episodes of exacerbation of chest infection and sinusitis. After evaluating the patient through history, general physical, systemic examination, and investigation we found the classical triad of bronchiectasis, situs inversus, and chronic sinusitis, and diagnosed the case as Kartagener syndrome. Reporting such cases in a given geographical area helps in the diagnosis of such rare diseases, as one can keep them in the differentials of recurrent chest infections. Early diagnosis of such disorders prevents patients as well as the health care system from the unnecessary burden of repeated investigation and helps in better outcomes and may result in improved quality of life.

Introduction

Kartagener syndrome is a rare genetic disorder characterized by a classic triad of symptoms: bronchiectasis, situs inversus, and chronic sinusitis. It is a form of primary ciliary dyskinesia (PCD), which involves the dysfunction of cilia, the hair-like structures that line various organs, including the respiratory tract. Siewert first described the combination of situs inversus, chronic sinusitis, bronchiectasis and ciliary dyskinesia as a Kartagener's syndrome (KS) in 1904 (Seiwert, 1904). It is a genetic condition with an autosomal recessive inheritance (Seiwert, 1904; Kartagener, 1933). Camner *et al.,* (1975) first suggested ciliary dyskinesia as the cause of KS in 1975.

In 1977, Eliasson *et al.,* (1977) first coined the term "immotile cilia syndrome" for KS to categorize infertility with chronic sinopulmonary infections. The estimated prevalence of primary ciliary dyskinesia (PCD) is about 1 in 30,000, (Barthwal, 2006) though it may range from 1 in 12,500 to 1 in 50,000 (Dixit *et al.,* 2009). In KS, the ultrastructural genetic defect leads to impaired ciliary motility which causes recurrent chest, ear, nose, throat,

and sinus infections, and infertility (Mayank Mishra *et al.,* 2012). 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 (Samuel, 1987). 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 (Mayank Mishra *et al.,* 2012; Samuel, 1987).

Case Presentation

A 65-year-old female non-smoker from Islamabad Bhatta, Aligarh, U.P., presented to the outpatient department of the Moalejat Department at Ajmal Khan Tibbiya College and Hospital. She was admitted to the inpatient department due to complaints of recurrent nasal congestion accompanied by itching, paranasal discomfort, and a cough with expectoration lasting over 30 years, along with progressively worsening shortness of breath. Her sputum was yellow, foul-smelling, and purulent, often brought up by postural drainage in the early morning after waking up.

Over the years, she had multiple hospital admissions and was diagnosed with chronic sinusitis, chronic bronchitis, and recurrent pneumonia. In the last 20 years, she experienced frequent exacerbations of her cough, producing copious purulent sputum. Notably, she had never received anti-tubercular treatment. Three years prior, an ENT specialist diagnosed her with chronic sinusitis and treated her with antibiotics and intranasal steroids.

She was married for 38 years and a mother of four children, her medical history included numerous visits to a paediatrician in her childhood for recurrent chest infections. Family history showed no parental consanguinity and no other relatives with similar issues.

During the physical examination, she appeared lean and thin, exhibiting grade 1 clubbing with slight nail bed angle obliteration, MRC grade 2 dyspnoea, and bilateral hypertrophy of the inferior turbinate. There was no palpable lymphadenopathy, pallor, icterus, or cyanosis. Her vital signs were: blood pressure 110/72 mmHg, pulse rate 92 beats per minute, respiratory rate 20 breaths per minute, and temperature 37.5 °C. Arterial oxygen saturation was 93% on room air. Cardiovascular examination revealed the heart apex beat in the fifth intercostal space at the right mid-clavicular line, with heart sounds best heard on the right side of the thorax. Respiratory examination showed coarse crackles and scattered rhonchi in both lung fields. Other systemic examinations were normal.

Laboratory tests showed haemoglobin at 11.1 gm/dl, a total leukocyte count of 11,680/µl with 74.7% granulocytes and 17.9% lymphocytes, and a platelet count of 316,000/µl. Sputum tests for acid-fast bacilli were negative for Mycobacterium tuberculosis. Serum chemistry, including fasting and postprandial blood sugar, blood urea, serum creatinine, liver function tests, and lipid profile, were all normal. X-ray Chest PA view showing right sided aortic arch and dextrocardia, along with fibrotic bands and right paracardiac bronchiectasis. Ultrasound examination of abdomen showed spleen on the right side, and liver and inferior vena cava on the left side which is suggestive of situs inversus. CT thorax image morphology is s/o complete situs inversus with liver and trilobed lung on left side and spleen, stomach and bilobed lung on right side. It shows dextrocardia with apex of heart towards right side. Consolidation patch with air bronchograms noted involving right lower lobe with multiple centrilobular ground glass nodules with many of them giving tree in bud appearance involving B/L lungs (infective), B/L apical pleural thickening, Shows multiple enlarged and sub-centimetric mediastinal lymph nodes in prevascular, paraaortic, B/L upper and lower paratracheal, B/L hilar and subcarinal region with many of them showing calcification, however no evidence of conglomeration noted. (Size of largest~ 14 X 22 mm in subcarinal region). It also shows multiple small subpleural fibrotic streaks and strands involving most of the B/L lung fields and few intraparenchymal fibrotic patches in left middle lobe with adjacent tractional bronchiectatic changes (old infective sequalae). Few cuts of abdomen taken shows no significant diagnostic abnormality. Bony window: Show degenerative changes in the spine.

Based on her clinical history, presentation, physical examination, and radiographic imaging findings, she was diagnosed with Kartagener Syndrome. During her hospital stay, she received a short course of a third-generation cephalosporin with clavulanate for pulmonary infection and was prescribed broad-spectrum antibiotics for prophylaxis. Additionally, inhaled corticosteroids, mucolytics, bronchodilators, and immunizations for influenza and pneumococcus were recommended, along with chest physiotherapy.

To enhance her treatment, she was advised to take a decoction of herbs including *Irsa* (*Iris insata*), *Katan* (*Linum usitatissimum*), *Asl as soos* (*Glycyrrhiza glabra*), *Arhusa* (*Adhatoda vasica*), and *Zoofa* (*Hyssopus Officinalis*), each 4 grams, along with one tablespoon (5 grams) of *Laooq khyar shamber*, twice daily for one month. She showed satisfactory improvement in symptoms and general well-being following this treatment regimen.

Results and 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 (Olbrich *et al.,* 2002). Defects in the ciliary component cause abnormal ciliary movements, resulting in impaired mucociliary clearance and manifesting as recurrent and / or persistent sinopulmonary infections (Marthin *et al.,* 2010).

Key Features

1. **Bronchiectasis**: This condition involves abnormal widening of the bronchi, causing chronic respiratory symptoms such as a persistent cough, thick mucus production, and frequent lung infections, ultimately leading to significant lung damage over time.

2. **Situs Inversus**: This is a congenital anomaly where the internal organs are arranged in a mirror image of their typical positions, such as the heart being located on the right side of the chest (dextrocardia).

3. **Chronic Sinusitis**: Patients often experience chronic inflammation of the sinuses, resulting in symptoms such as nasal congestion, facial pain, and recurrent sinus infections (Kartagener, 1933; Camner *et al.,* 1975; Eliasson *et al.,* 1977).

4. **Infertility**: Most infertile patients with KS have a normal spermatozoid count, but with a structural defect and a complete lack of motility (Afzelius and Eliasson, 1983). Infertility in male with KS is due to diminished sperm motility, while in females it is due to defective ovum transport because of the dyskinetic motion of oviductal cilia, suggesting that the ciliated endo-salpinx is essential for human reproduction (McComb *et al.,* 1986).

5. **Pathophysiology:** Normal ciliary function is critical for respiratory host defence and motility of sperm, and ensures proper visceral orientation during embryogenesis. The underlying cause of Kartagener syndrome is linked to genetic mutations affecting ciliary function. The most commonly involved gene is *DNAH5*, which encodes a protein that is essential for the movement of cilia. These defective cilia fail to effectively clear mucus and pathogens from the respiratory tract, leading to the accumulation of secretions and resulting infections, which predispose to recurrent sinopulmonary infections, infertility, and errors with left-right body orientation (Gupta *et al.,* 2012; Shyama and Prudence, 2017).

Diagnosis

Diagnosis is primarily based on clinical presentation and can be confirmed through various approaches:

- **Imaging**: Chest X-rays or CT scans can identify bronchiectasis and assess lung health.
- **Genetic Testing**: Identifying mutations in genes related to ciliary function can confirm the diagnosis.
- **Ciliary Motility Tests**: These tests assess the movement of cilia and can help determine their functionality (Gupta *et al.,* 2012).

Management

The management of Kartagener syndrome aims to relieve symptoms and prevent complications:

- **Airway Clearance Techniques**: Techniques like chest physiotherapy and devices to help clear mucus.
- **Antibiotic Therapy**: Antibiotics are prescribed to treat and prevent respiratory infections.
- **Sinus Management**: In cases of chronic sinusitis, medications or surgical interventions may be necessary (Shyama and Prudence, 2017).

Prognosis

Although there is variability in the prognosis of individuals with Kartagener syndrome, many patients can lead active lives, if appropriate treatment given. Maintaining respiratory health and general wellbeing requires routine monitoring and therapy modifications.

Figure.1 X-ray Paranasal sinus Water's view showing bilateral opacified maxillary sinuses. Note: the correct side marker on the right upper corner

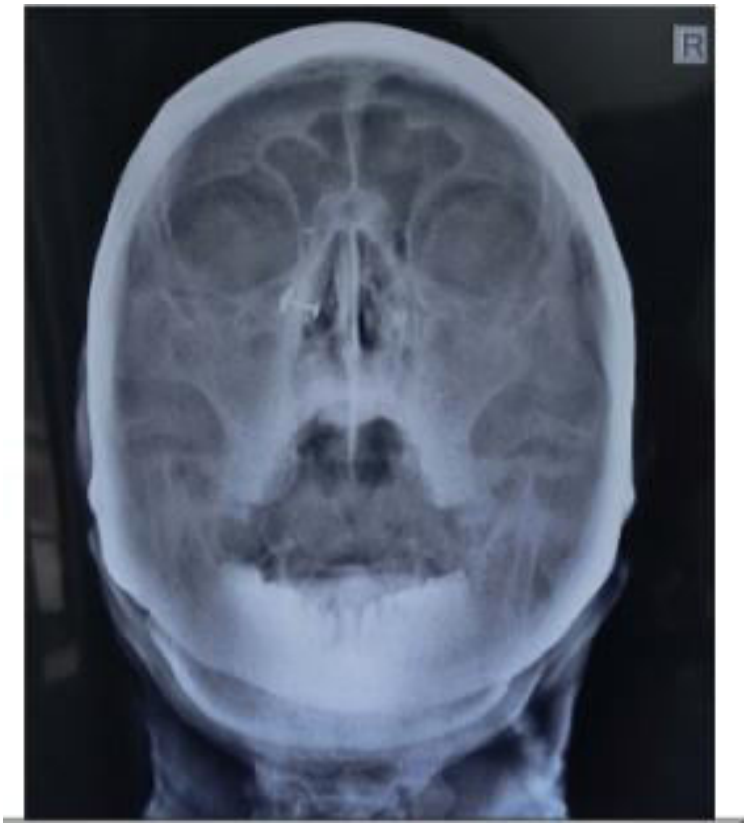


Figure.2 Axial section CT scan of the chest showing cystic bronchiectasis noted in bilateral lung fields

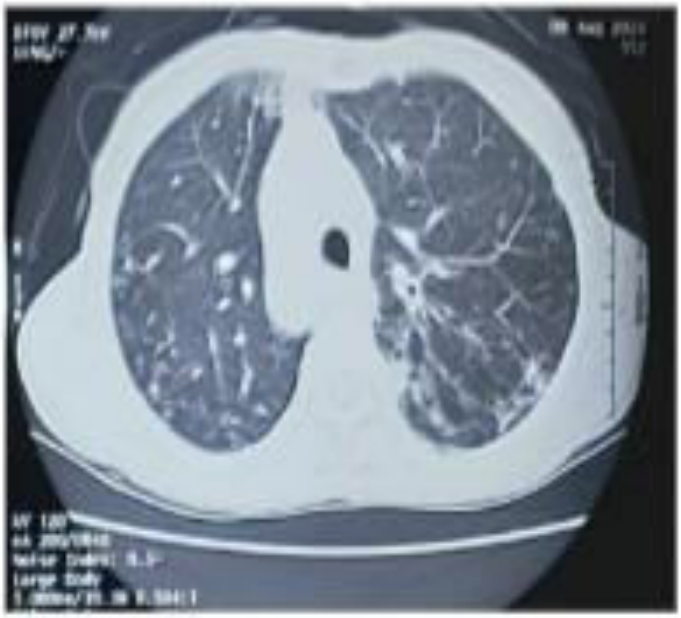
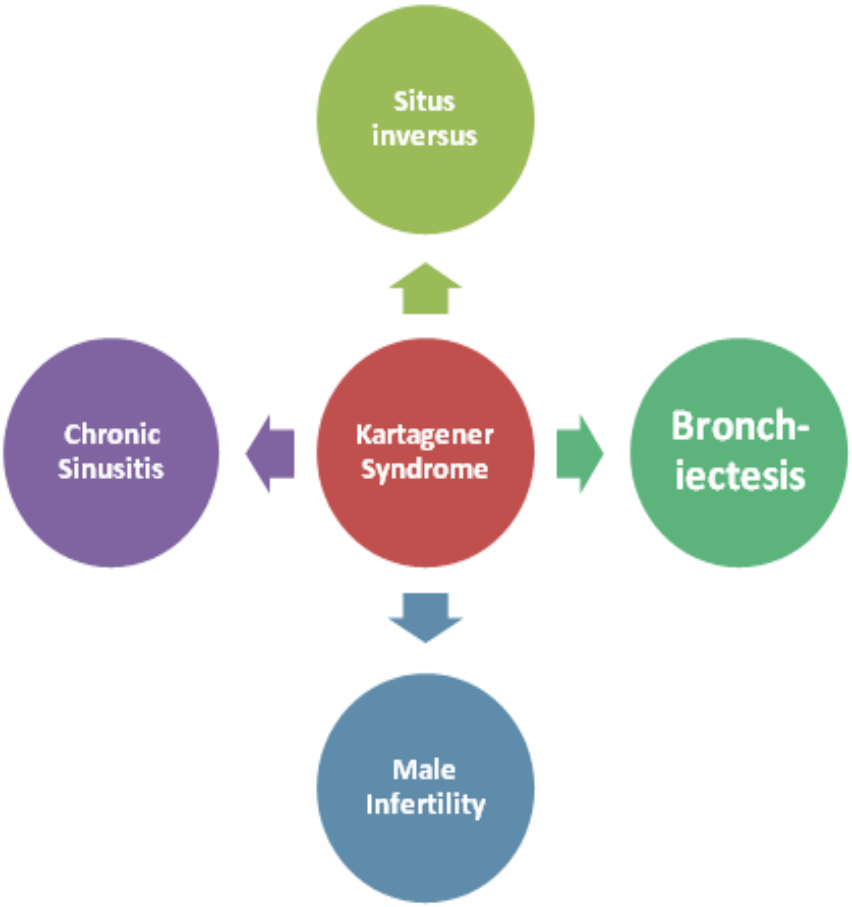


Figure.4



In this study patient presented with recurrent episodes of sinopulmonary infections. Imaging findings revealed bronchiectasis, dextrocardia, and situs inversus, which met the diagnostic criteria for Kartagener Syndrome. Laboratory screening and confirmatory tests, which required a better clinical setup, were not done.

Standard treatment for sinopulmonary problems in people with KS had been given to the patient which includes chest physiotherapy, mucolytics, and antibiotics along with adjuvant therapy with Unani medicine which includes drugs like *Irsa, Katan, Asl as soos, Arhusa, zoofa. Khyarshambar*as these drugs possess properties like *Mohallil* (resolvent), *Munaffis Balgham* (expectorant), *Mufattehurooq Khashna* (bronchodilator), *Musakkin-e-Sual* (Antitussive) which are responsible for better outcome in our patient (Ali Syed Safiuddin and Unani Advia Mufrada, 1993; Anonymous, 1956; Kirtikar and Basu, 1981; Khan Mohd Azam and Muheet e Azam, 2013I; Khan Mohd Azam and Muheet e Azam, 2013II; Khan Mohd Azam and Muheet e Azam, 2013IV; Al Razi Abu Bakar Mohammad Bin Zakariya, 1999).

In conclusion, while Kartagener Syndrome is a rare genetic condition, the case presented here demonstrates its occurrence in Aligarh, Uttar Pradesh. It should be considered in the differential diagnosis for patients with sinusitis, recurrent chest infections, situs inversus, and specific radiological features. The absence of a simple, reliable, and non-invasive diagnostic test for KS often leads to delays in diagnosis, which can result in chronic respiratory issues and a reduced quality of life. Timely and accurate identification of KS can prevent unnecessary investigations, multiple exposures in radiodiagnosis, and inappropriate antibiotic use, ultimately enhancing patients' quality of life. Additionally, genetic counselling and fertility concerns should be addressed upon diagnosis.

Author Contributions

Tooba Hayat: Investigation, formal analysis, writing—original draft. Mohd Mushfiq: Validation, methodology, writing—reviewing. Jamal Azmat:—Formal analysis, writing—review and editing.

Data Availability

The datasets generated during and/or analyzed during the current study are available from the corresponding author on reasonable request.

Declarations

Ethical Approval Not applicable.

Consent to Participate Not applicable.

Consent to Publish Not applicable.

Conflict of Interest The authors declare no competing interests.

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