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Case Report

A Case Report of Kartagener's Syndrome: Clinical Presentation, Diagnosis, and Management Challenges

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Abstract

Background: Kartagener's Syndrome (KS) is a rare autosomal recessive genetic disorder characterized by defects in the structure and function of cilia. It is a subset of primary ciliary dyskinesia (PCD) and is defined by a triad of chronic sinusitis, bronchiectasis, and situs inversus totalis. The condition often leads to infertility due to impaired sperm motility. Given its rarity and overlapping symptoms with other respiratory diseases, KS is frequently misdiagnosed. *Case Presentation:* Our client was a 40-year-old male who presented to the Rivers State University Teaching Hospital, Port Harcourt, Rivers State, South-south Nigeria with recurrent productive cough, rhinorrhea, exertional breathlessness, and wheezing—symptoms have been present since childhood. He had been managed for bronchial asthma at a peripheral center and also reported a history of primary infertility for six years. Clinical examination revealed respiratory distress, oxygen saturation of 87% on room air, and auscultatory findings of coarse crepitations and inspiratory rhonchi. Notably, cardiac auscultation localized heart sounds to the right side of the chest, raising suspicion of situs inversus. A chest computed tomography (CT) scan confirmed cystic bronchiectasis and situs inversus totalis, leading to a diagnosis of Kartagener's Syndrome. Spirometry demonstrated an obstuctive ventilatory pattern with significantly reduced Forced Expiratory Volume in 1 second (FEV1), Forced Vital Capacity (FVC) and FEV1/FVC ratios. The patient was managed with antibiotics, bronchodilators, steroids, antihistamines, and chest physiotherapy. Educating and counseling the patient on disease condition, referred to psychotherapists/ Social support group. He remains under follow-up in the respiratory clinic for long-term care. *Conclusion* Kartagener's Syndrome, though rare, has been reported globally (1–5). Due to symptom overlap with asthma and other chronic respiratory conditions, it is frequently underdiagnosed. Early recognition using clinical evaluation and imaging studies is crucial for appropriate management and improved patient outcomes.

Keywords

Primary Ciliary Dysfunction, Kartagener's Syndrome, Clinical Presentation, Diagnosis, Management, Nigeria, Challenges

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1. Introduction

Kartagener's Syndrome (KS) is a rare genetic disorder first described in 1933 by Manes Kartagener [1-5]. It is inherited in an autosomal recessive manner and results from mutations in genes such as Dynein, Axonemal, Intermediate Chain 1(DNAI1), Dynein, Axonemal, Heavy Chain 5 (DNAH5), and Coiled-Coil Domain-Containing protein 39 (CCDC39), which are responsible for the normal function of cilia [6-8]. Ciliary dysfunction impairs mucociliary clearance in the respiratory tract, leading to chronic infections, sinusitis, and bronchiectasis [9]. Additionally, ciliary defects disrupt embryonic organ positioning, resulting in situs inversus totalis—a complete mirroring of thoracic and abdominal organs [3]. Patients with KS frequently experience: • Recurrent respiratory infections • Chronic sinusitis • Bronchiectasis • Infertility (due to sperm immotility in males) • Situs inversus totalis [5, 10]. Diagnosis is confirmed using a combination of:

1. Clinical history (chronic respiratory symptoms and infertility)
2. Imaging (Chest X-ray or CT scan revealing bronchiectasis and situs inversus)
3. Specialized tests: • Nasal nitric oxide measurement (low levels suggest Primary Ciliary Dysfunction, [PCD]) • Transmission electron microscopy (identifying ciliary defects) • Genetic testing (to identify specific mutations) [11-12]. There is no definitive cure for KS. Management focuses on symptom control and prevention of complications using: • Airway clearance therapy (chest physiotherapy, postural drainage) • Antibiotics (for recurrent infections) • Bronchodilators and mucolytics (to improve airway function) • Assisted reproductive techniques (such as in vitro fertilization for infertility) [5, 13-15]

2. Case Presentation

A 40-year-old male presented to the Rivers State University Teaching Hospital, Port Harcourt, Rivers State, South-south Nigeria with a longstanding history of recurrent productive cough and rhinorrhea dating back to childhood. He also reported exertional breathlessness, poor effort tolerance, and wheezing. Prior to this visit, he had been managed at a peripheral health facility for bronchial asthma, without significant improvement. He also had a six-year history of infertility.

2.1. Clinical Examination Respiratory Examination Findings

The patient was in respiratory distress • Oxygen saturation was 87% on room air, this however improved with oxygen supplementation • Coarse crepitations were heard in the mid and lower lung zones bilaterally • Inspiratory rhonchi were noted in the right lung zones • Heart sounds were predominantly heard on the right side of the chest, suggestive of situs inversus • No other significant abnormalities were detected.

2.2. Investigations

• Chest Computed Tomography (CT) Scan • Cystic bronchiectasis • Situs inversus totalis • Spirometry: • Forced Expiratory Volume in 1 second (FEV1), Forced Vital Capacity (FVC) and FEV1/FVC were reduced, indicating obstructive ventilatory impairment. Semen analysis was significantly deranged.

2.3. Diagnosis

The diagnosis of Kartagener's Syndrome was made based on the clinical presentation and imaging findings

2.4. Management and Outcome

The patient was started on• Antibiotics (to treat recurrent infections) • Bronchodilators (to relieve airway obstruction) • Steroids (for inflammation control) • Antihistamines (to manage allergic symptoms) • Chest physiotherapy (for airway clearance) He showed gradual improvement and was scheduled for long-term follow-up in the respiratory clinic to monitor disease progression and prevent complications.

3. Results

Attached are results of some of the patient investigations; Chest Computed tomography.

Table 1. *Spirometric indices.*

Indices	Predicted	Measured pre-bronchodilator	%Predicted	Measured post bronchodilator	%Predicted	◇(Change)
FEV1(L)	3.2	1.60	50	2.0	63	+0.4L (25%)
FVC (L)	4.0	3.2	80	3.5	88	+0.3L (9%)
FEV1/FVC (%)	80.0	50.0	-	0.57	-	+7%
PEFR (L/s)	8.0	4.5	56	5.8	72	+1.3L/s (29%)

Indices	Predicted	Measured pre-bronchodilator	%Predicted	Measured post bronchodilator	%Predicted	◇(Change)
FEF25-75%(L/s)	3.5	1.1	31	1.7	49	+0.6L/s (55%)

Table 2. *Semen analysis report.*

Parameter	Result	Reference Range
Volume	2.5	1.5-5ml
Appearance	Opalescent	Whitish, opaque
PH	7.4	7.2-8.0
Concentration	8million/ml	15-200million/ml
Total sperm count	20million	Million
Motility (Progression count)	10%	Greater than 40%
Motility (Non-progressive)	15%	Less than 10%
Non-motile	75%	Less than 40%
Morphology	2%	Greater than 4% (WHO standard)
Vitality (live sperm)	50%	Greater than 58%
Round cells	2million /ml	Less than 5million /ml
Leucocytes	0.5million /ml	Less than 1million/ml

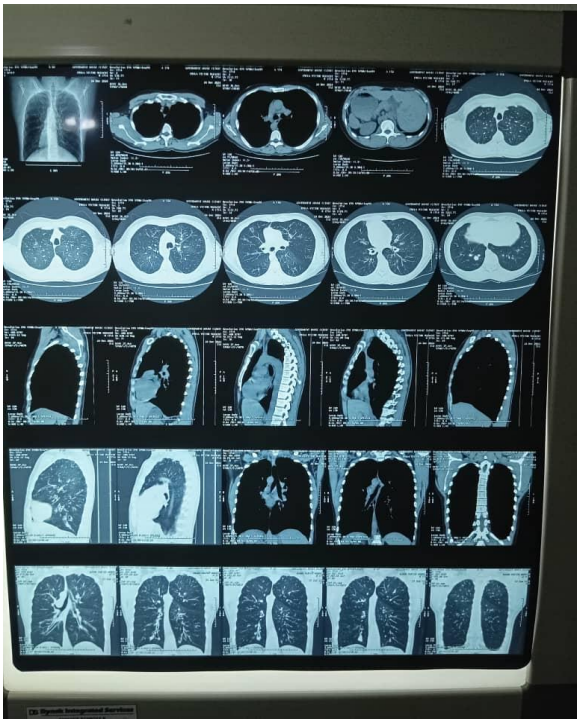

Figure 1. *Computed tomography scan.*

4. Discussion

Kartagener's Syndrome is a rare but significant cause of chronic respiratory disease and infertility. The triad of bronchiectasis, sinusitis, and situs inversus total is should prompt consideration of KS, particularly in patients with unexplained recurrent respiratory infections [6, 7].

4.1. Challenges in Diagnosis

The overlapping symptoms with asthma and chronic bronchitis often lead to misdiagnosis. Genetic testing is not readily available in resource-limited settings, making diagnosis reliant on clinical suspicion and imaging.

4.2. Long-term Management

Pulmonary rehabilitation is essential to maintain airway function. Lifelong monitoring is necessary to prevent progressive lung damage. Assisted reproductive techniques may be required for affected males.

4.3. Prognosis

With early diagnosis and appropriate management, patients with KS can lead relatively normal lives. However, untreated

bronchiectasis can lead to progressive lung damage, increasing the risk of respiratory failure.

5. Conclusion

Kartagener syndrome is not as scarcely seen as was previously assumed in sub-saharan Africa. This case highlights the importance of recognizing Kartagener's Syndrome early to ensure timely and appropriate management. Given its non-specific symptoms, KS is often mistaken for asthma or other chronic respiratory diseases, leading to delayed diagnosis. Clinicians should have a high index of suspicion, especially in patients with recurrent respiratory symptoms, situs inversus, and infertility. Availability of genetic test will also aid early diagnosis.

Abbreviations

PCD	Primary Ciliary Dysfunction
DNAI1	Dynein, Axonemal, Intermediate Chain 1;
DNAH5	Dynein, AXonemal, Heavy Chain 5
KS	Kartagener's Syndrome
CCDC39	Coiled-Coil Domain-Containing Protein 39
CT	Computed Tomography
FEV1	Forced Expiratory Volume in 1 Second
FVC	Forced Vital Capacity

Conflicts of Interest

No Conflicts of Interest exists among the authors.

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