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Kartagener Syndrome in a Young Zambian Male: A Case Report

Moses Chibamba[1*], Chipo Mushinda[1], Butemwe Kawanda[2], Leticia Mwiinga[1]

[1]Kabwe Women, Newborn and Children's Hospital, Kabwe, Zambia
[2]Key Laboratory of the Ministry of Health, University Teaching Hospital, Lusaka, Zambia
Email: *chibambamosesnyense@gmail.com, chipomushinda@gmail.com, butemwekawanda13@gmail.com, leticiamusante@gmail.com

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Abstract

Introduction: Kartagener Syndrome (KS), a subset of Primary Ciliary Dyskinesia (PCD), is a rare autosomal recessive disorder caused by defective motile cilia. The classical triad of chronic sinusitis, bronchiectasis, and situs inversus defines the syndrome. In many low-resource settings, KS is frequently underrecognized and often misdiagnosed as pulmonary tuberculosis due to overlapping respiratory symptoms. **Case Presentation:** This case involved a 13-year-old Black Zambian boy who presented with a long-standing history of intermittent productive cough and repeated lower tract infection treatment associated with chronic sinusitis with frequent exacerbation, since infancy. He was initially diagnosed with pulmonary tuberculosis based on clinical and radiological findings, despite the negative GeneXpert treatment continued with little improvement. His treatment comprised a three-month duration of rifampicin, isoniazid, ethambutol and pyrazinamide. His imaging investigations revealed dextrocardia on chest radiograph and complete situs inversus on abdominal ultrasonography. Echocardiography confirmed situs inversus totalis with preserved ventricular function. A high-resolution chest computed tomography scan was suggestive of complete situs inversus providing a higher suspicion of Kartagener Syndrome. Routine laboratory investigations were within normal range. His supportive management, included airway clearance therapy, mucolytics, and prophylactic antibiotics, with significant clinical improvement and no apparent exacerbation in the last 3 months. **Conclusion:** The diagnosis of Kartagener syndrome is typically challenging and often delayed because the clinical symptoms often mimic the common infectious disease. Since there is no ultimate definite treatment for Kartagener syndrome, early diagnosis and supportive management are critical to prevent irreversible chronic lifelong sequelae complications such as lung damage. This calls for strengthening diagnostic capacity, high index of suspicion in order to make

early diagnosis, appropriate timely management, and prevention of irreversible pulmonary damage.

Keywords

Primary Ciliary Dyskinesia, Kartagener Syndrome, Dextrocardia, Situs Inversus, Bronchiectasis, Case Report

1. Introduction

Kartagener Syndrome (KS) is known by several synonyms, including:

1) **Immotile Cilia Syndrome:** This term is rarely used now, as it is understood that the cilia are not always completely immotile but may show abnormal motility.

2) **Primary Ciliary Dyskinesia (PCD):** synonymously used for **Kartagener Type**.

3) **Siewert (Zivet) Syndrome**

4) **Chronic Sinobronchial Disease with Dextrocardia:** This term refers to the cardinal critical features of Kartagener syndrome: dextrocardia, bronchiectasis, and chronic sinusitis.

Primary Ciliary Dyskinesia (PCD) is a very rare autosomal recessive disorder, characterised by impaired mucociliary clearance, leading to chronic oto-sino-pulmonary disease, and in approximately 50% of cases shows a defect (e.g., situs inversus) [1].

Kartagener syndrome (KS) is a subset of broader spectrum of ciliary motility disorders known as primary ciliary dyskinesias (PCDs) [2]. KS is a rare inherited disorder inherited in an autosomal recessive manner. Although it was first mentioned by Siewert in 1904, it was Manes Kartagener who clinically described the syndrome in 1933 [3]. The condition is characterized by a unique set of symptoms, including but not limited to: chronic sinusitis, bronchiectasis, and situs inversus.

At a fundamental hereditary tier, the *dynein axonemal intermediate chain* 1 (DNAI1) and *dynein axonemal heavy chain* 5 (DNAH5) gene mutation in KS causes reduced ciliary motility, which escalates the risk of infertility, left-right body alignment problems, and recurrent sinopulmonary infections. Normal ciliary function is crucial for sperm movement, respiratory system defense, and the proper positioning of internal organs during development [3]. The disordered cilia also lead to poor clearance of mucus from the lungs, making these patients more prone to repeated respiratory infections [4].

Marking a diagnosis of primary Kartagener is complex, as there is no gold standard test and requires a combination of tests to typically make a diagnose the condition. The European Respiratory Society recommends using several diagnostic methods, including measuring nasal nitric oxide (nNO) levels, high-speed video microscopy, transmission electron microscopy (TEM), and genetic testing. When TEM is inaccessible, immunofluorescence staining of ciliary proteins can provide a likely diagnosis. Overall ultrastructural ciliary defects are common in

PCD, around 26% of patients diagnosed through genetics and other tests may not show visible defects in their cilia [5].

The incidence of PCD is estimated to be about 1 in 32,000 live births, and the incidence of KS overall ranges from 1 in 20,000 to 1 in 40,000 births [6] [7]. In Zambia, the prevalence of Kartagener syndrome remains largely unknown, with no documented case reports available, highlighting the scarcity of the condition. Factors leading to underreporting is partly attributed to limited access to testing in low-middle-income countries.

In this report, we present a case involving a 13-year-old male diagnosed with clinically suspected Kartagener syndrome. This case report aims to emphatically raise awareness about the importance of early diagnosis and intervention in managing the disease, which can substantially enhance the quality of life for affected individuals.

2. Case Presentation

A 13-year-old black male, came to our hospital with a long-standing history of recurrent chronic cough that produces thick, brownish sputum since infancy. His clinical manifestations were further complicated by intermittent episodes of respiratory distress with associated nasal congestion and discharge leading to multiple hospital admissions over the years, which led to further investigations and the initiation of anti-tuberculosis treatment based on clinical suspicion.

The cough was typically productive, yielding whitish sputum, and occasionally associated with chest pain. Notably, he did not experience hemoptysis, fever, weight loss, or night sweats. Review of other systems was unremarkable. His mother reported that he was delivered at the hospital after a full-term pregnancy via spontaneous vaginal delivery. Has no significant history of developmental delays. He is the last-born child and has four older siblings with no similar issues reported among all other family members. He has no history of smoking or alcohol intake.

On physical examination, SC was alert and active. Was Stable with normal vital signs. No visible nasal polyps. Lung auscultation revealed bibasal course crackles. There was no finger clubbing or central and peripheral cyanosis. Examination of the heart revealed that the cardiac apex was palpated on the right side, heart sounds were detected more on the right side with normal heart sounds and no added heart sounds. Abdominal examination and other systems were unremarkable.

Patient was admitted to the pediatric ward for further investigations which were revealing as shown in **Figures 1-5**.

The results of baseline laboratory tests performed (**Table 1**) to rule out concomitant infectious processes.

Upon discharge, he was recommended to undergo thoracic physiotherapy, employing techniques such as postural drainage, percussion, and vibrations to aid in mucus clearance from the airways. Patient's body weight was 50 kg, he was prescribed Azithromycin 500 mg, once a day three times a week amounting to 1500

mg a week for 6 months as guided by The BESTCILIA trial, and Guaifenesin syrup 200 mg three times daily. Patient was monitored for potential adverse effects with the use ECG and liver enzymes which showed no changes. Long term antimicrobial therapy was considered in the context of the potential for antimicrobial resistance. Discontinuation of Fixed Drug Combination for pulmonary tuberculosis management. Other potential causes of chronic suppurative respiratory disease such as cystic fibrosis, allergic bronchopulmonary aspergillosis were considered however, comprehensive investigations for these conditions were not available in our setting. Regular follow-ups for a period of 6 months after treatment initiation was done and showed continued progress with better symptom control and fewer episodes of respiratory distress. During the follow ups, pseudomonas aeruginosa was isolated from sputum. Antimicrobial susceptibility testing was performed and results informed the choice of antimicrobial therapy as susceptible to azithromycin.

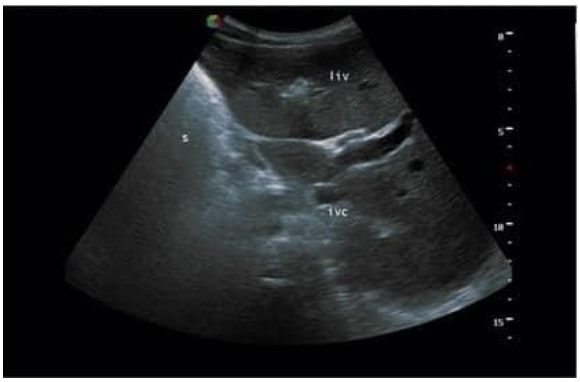

Figure 1. An ultrasound scan; demostrates normal organ architecture but revealed situs inversus (liver and inferior vena cava on the left side, and spleen on the right side).

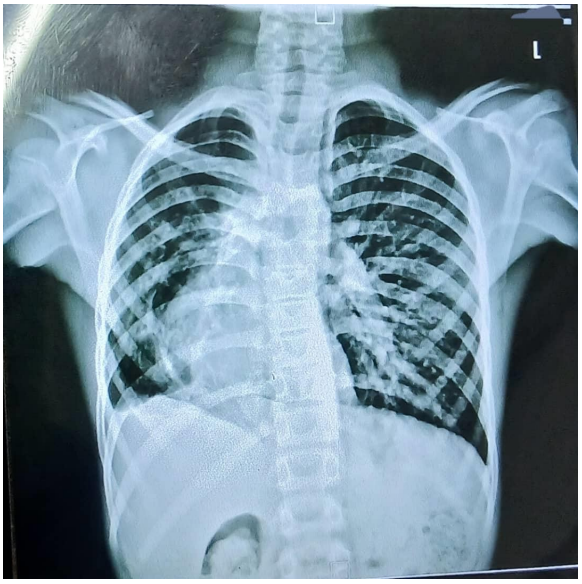

Figure 2. Chest X-ray on the posterior-anterior view showed dextrocardia with bilateral infiltrates (brochiectatic changes).

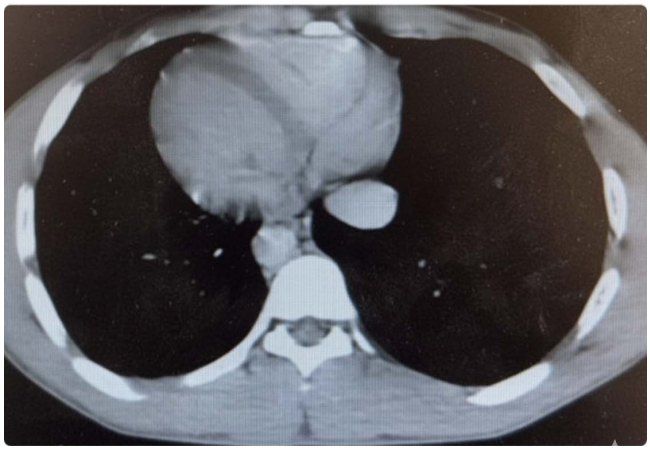

Figure 3. High-resolution chest computed tomography scans showing complete reversal of heart and great vessels.

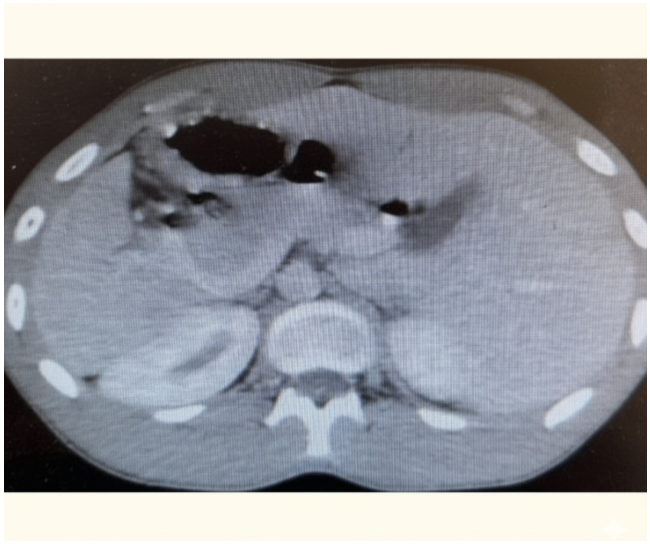

Figure 4. High-resolution chest computed tomography scans showing abdominal organs with stomach and spleen on the right side and liver on left side, suggestive of complete situs inversus.

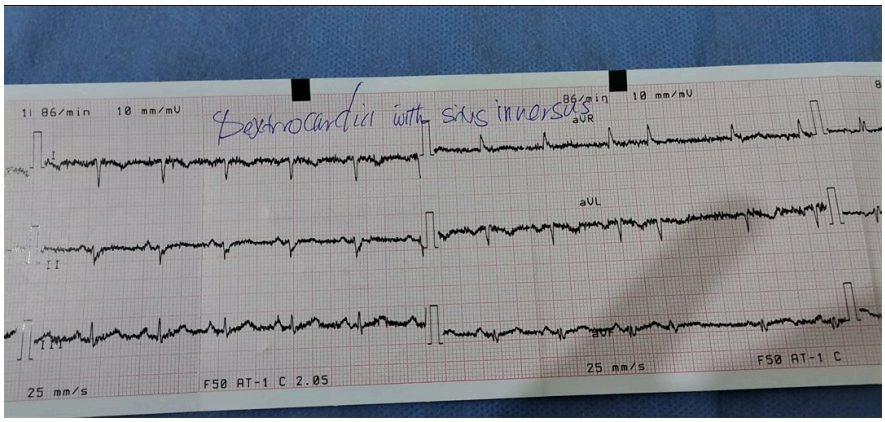

Figure 5. Electrocardiography (ECG) revealed a right axis deviation, an iverted p wave in lead I and avL.

Table 1. Laboratory values from follow up visits with reference ranges.

Lab results	At 2 months review	After 2 months	After 6 months	Reference
Hgb (g/dl)	12.8	13.1		13.2 - 18.3
Urine LAWBC (103/Ul)	6.7	7.4		2.8 - 8.1
Neutrophils (%)	52	56		40 - 60
Lymphocytes (%)	28.7	29.3		26.3 - 59.3
RBS (mg/dL)	N/A	N/A		N/A
Creatinine (μmol/dL)	41.00	Not done		26.52 - 44.2
Urea (mmol/L)	5.2	4.5		2.8 - 7.2
ESR	12	Not done		0 - 20
Gene-Xpert MTB/RIF Assay from sputum	Negative	Negative	Negative	
Urine LAM	Negative	Negative	Negative	
Sputum MCS	Collected	-	Pseudomonas Aeruginosa	
HIV status	Negative		Negative	
ALT (U/L)	10.5	Not done		<45
AST (U/L)	9.9	Not done		<35

3. Discussion

Disorders of ciliary motility may be congenital or acquired. The congenital form, Primary Ciliary Dyskinesia (PCD), is a genetically heterogeneous disorder caused by abnormalities in motile cilia [1]. Approximately half of affected individuals have situs inversus, and when this occurs alongside PCD, the condition is defined as Kartagener syndrome (KS) [1]. Although the exact prevalence is not known PCD is estimated to be 1 in 20,000 - 40,000 live births [3]. Since Siewert's description of a condition in 1904, several additional cases have been documented worldwide, including African countries [7]-[10].

Our patient, in this case report presented with situs inversus totalis and abiding chronic respiratory symptoms, raising a strong clinical suspicion of Kartagener syndrome. Although genetic testing could not be performed in our patient, no other family members clinically demonstrated respiratory symptoms or abnormal imaging.

PCD manifestations vary substantially. Some infants show respiratory distress at birth, while others develop persistent cough, recurrent infections, bronchiectasis, or chronic rhinosinusitis later in childhood. Otitis media, atypical asthma features, and in some cases hearing impairment may occur. Females may experience subfertility or ectopic pregnancy, and males often demonstrate infertility [11]. Because the disorder is congenital, symptoms often appear early, and clinicians should consider PCD in young children with recurrent upper and lower respira-

tory symptoms [12]. As disease progresses, radiologic bronchiectasis and obstructive impairment become evident even in preschool-aged patients [13]. Consistent with these features, our patient had a long history of productive cough, recurrent respiratory tract infection and chronic sinus disease.

Preventive care such as including influenza and pneumococcal vaccination and avoidance of environmental pollutants are recommended to limit infections [3] [14]. In this case, chest X-ray revealed bronchiectasis and the clinical history suggested recurrent infections.

Diagnosis of Kartagener syndrome relies on characteristic clinical features supported by imaging, ciliary motility studies, and genetic assessment when available [15]. Diagnostic criteria include chronic bronchial infections and rhinitis from early childhood, plus evidence of situs inversus/dextrocardia, immotile sperm, impaired mucociliary clearance, or ultrastructural ciliary defects [3]. Semen analysis may reveal immotile sperm, and mucociliary clearance can be assessed through the saccharin test. Nasal nitric oxide (NO) levels are also helpful, with PCD patients generally showing only 10% - 20% of normal values [1]. Due to resource limitations, advanced tests were not obtained; semen analysis could not be done as the patient did not consent to it. However, the combination of situs inversus and imaging-confirmed bronchiectasis supported a diagnosis of Kartagener syndrome.

Differential diagnoses such as cystic fibrosis, asthma, allergic bronchopulmonary aspergillosis, chronic aspiration, alpha-1 antitrypsin deficiency, and other bronchial disorders should be excluded [16]. In our patient, the medical history and physical examination showed features of situs inversus and chronic rhinosinusitis with recurrent exacerbations of respiratory tract symptoms supporting KS as the most plausible differential diagnosis.

There are no restorative and curative options yet, PCD management aims at controlling symptoms and preventing complications. Current treatment guidelines are partly adapted from cystic fibrosis and non-CF bronchiectasis guidelines, though PCD has dissimilar pathophysiology. As a result of meagerness of evidence-based treatment options, therapeutic approaches vary from country to country. An undisputable attest was established on Azithromycin as the only evidence-based chronic therapy with proven benefit in PCD, helping reduce symptoms and exacerbations [17]. Our patient was treated with low-dose azithromycin three times weekly, mucolytics and chest physiotherapy. This showed a significant noticeable improvement and regular clinical reviews.

A 2009 to 2021 retrospective pediatric study carried out in China established that patients taking azithromycin had fewer flare-ups and better exercise endurance compared to those not receiving the drug [18]. The paramount leverage of Azithromycin's is its anti-inflammatory effects which help lessening airway inflammation and mucus production, both of which characterize PCD. A Long-term low-dose therapy may also help preventing recurrent infections and its associated symptoms [3] [18].

Supplemental treatments with symptomatic relief for selected advanced cases of KS include but not limited to the following hypertonic saline, acetylcysteine, and recombinant human DNase (rhDNase), despite their properties, evidence shows that the efficacy still remains limited [1] [19]. In some severe cases, such as end-stage lung destruction surgical interventions may be considered, a case of successful lobectomy and bilateral lung transplantation for advanced KS have been reported [20] [21]. The prognosis for Kartagener syndrome is generally favorable when patients receive regular respiratory care and holistic monitoring and most patients have normal life expectancy [12]. Our patient will continue follow-up visits every 3 - 6 months for monitoring of symptoms, end organ function, and progression of the overall disease.

4. Conclusion

The diagnosis of Kartagener syndrome (KS) is often belated because its clinical presentation can be mistaken for common respiratory infections. Although there is no magic bullet for KS, early identification, diagnosis and management are vital to prevent irreversible lung damage and chronic lifelong complications. Therefore, high index of suspicion, early diagnosis can markedly ameliorate the long-term outlook for the impacted patients by allowing for timely interventions. Multidisciplinary collaboration and a holistic approach are key to facilitating an early diagnosis and ensuring appropriate treatment strategies.

Declaration of Figure Authenticity

The figures are created by the authors, who confirmed that the images are original with no duplication and have not been previously published in whole or in part.

Consent

Written informed consent was obtained from the patient's parents for publication of this case report and any accompanying images.

Author Contributions

Moses Chibamba and Butemwe Kawanda prepared the manuscript. Chipo Mushinda and Leticia Musante conceptualized and prepared the images. All authors reviewed and approved the manuscript.

Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

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