CASE REPORT

Kartagener's syndrome: A case report

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Abstract

The primary defect in Kartagener's syndrome is at the ciliary level and so the normal ciliary activity is compromised causing impaired respiratory tract clearance, infertility and defective organ(s) orientation and left-right laterality predisposition during organogenesis. The authors came across the case of a 20-year-old female that was later diagnosed with Kartagener's syndrome. This case may be of particular interest to general physicians and pulmonologists.

Keywords: Kartagener's Syndrome, Bronchiectasis, Situs Inversus, Chronic Sinusitis

Introduction

Individuals with Kartagener's Syndrome (KS) often present with complaints of difficulty in breathing, shortness of breath or in some cases, a history of chronic sinusitis [1]. The condition though, is characterised by a classical triad of chronic or recurrent sinusitis, bronchiectasis, and situs inversus. Situs inversus may involve all the abdominal and thoracic organs (totalis) or may occur as only dextrocardia (solitus). In the case described below, the patient was displaying the classical triad of KS with situs inversus totalis. KS is more common in populations practicing consanguineous marriages. The reported incidence as of 2020, was 1 in 32,000 to 40,000 births [2]. There is no definitive treatment for KS but lifestyle modifications coupled with supportive therapy form the mainstay of the management of this condition.

Case Report

We present a case of a 20-year-old girl who was brought to the hospital with complaints of fever for the last 10 days, associated with cough. The fever was intermittent in nature and was associated with chills. There was no reported evening rise in temperature or history of night sweats. There were, however, generalized body pains and fatigue. The fever was associated with cough which was productive in nature with white sputum. The patient also complained of having shortness of breath on exertion which started from the day of onset of the fever (Grade II). There was no evidence of hypertension or diabetes mellitus in the patient. The patient's mother also complained of weight loss in the patient in the last one month and there was a significant past history of pneumonia four months ago. Upon asking for further past evidence of similar infections, it was reported that the patient regularly suffered from bouts of coryzas, rhinosinusitis, bronchitis, and lung infections. There was an associated headache whenever she suffered from upper respiratory tract infections which was aggravated on bending forward but relieved by steam inhalation and medication. There was also discharge from the nose at times of coryza, associated with the nasal blockade. Symptoms were aggravated by exposure to dust and the patient had a well-documented dust allergy. The mother

had no issues during pregnancy and no one else in the family exhibited similar complaints. The patient was a full-term, vaginally delivered, healthy baby who weighed approximately 3.0 kg at birth. She attained the required milestones at the appropriate ages. She attained menarche at the age of 13 and had been having irregular menstrual cycles since then. Based on general appearance, the patient looked weak and was moderately built. She was well-oriented to time, place, and person. Her blood pressure was recorded at 120/80 mm of Hg (recorded with the patient in a sitting position; left arm) while her pulse rate was 96 beats/minute. Her SpO_2 at room temperature was 94% with the respiratory rate found to be 28/minute. Physical examination revealed that the patient's body temperature was slightly elevated (100º F) and diaphoretic. Upon auscultation of the cardiovascular system, the cardiac apex was heard on the right side (5^{th} intercostal space) with the left side giving no sounds at all. There were no noticeable murmurs or thrills. Auscultation of the respiratory system brought to light bilateral coarse crepitation in the lower lobes of both lungs (right side more than the left). It was best heard in the infrascapular regions of both sides. The rest of the systemic examination revealed no abnormalities. The general and systemic findings pointed us towards subjecting the patient to radiological studies to confirm these findings while also helping in establishing a definitive diagnosis.

Investigations

Taking into account the relevant history and the physical examination, a provisional diagnosis of lower respiratory tract infection with dextrocardia was made.

Complete blood picture

The complete blood picture revealed an elevated White Blood Cell (WBC) count and an accompanying slight degree of anaemia. The patient's haemoglobin was just below the 12% with cut-off at 11%. The elevated WBC count pointed towards an ongoing infection.

Electrocardiogram (ECG)

The ECG revealed changes suggestive of dextrocardia. There was poor R wave progression from V1 to V6 and positive aVR.

2-Dimensional Echo study

The 2-Dimensional Echo study was done and revealed no regional wall motion abnormality and with no murmurs. 2D echo was done keeping in mind the abnormal ECG.

Sputum Culture

The patient's sputum was subjected to culture and microbiological studies. Pseudomonal growths were observed and the choice of antibiotic was made.

Radiological studies:

Chest X-Ray

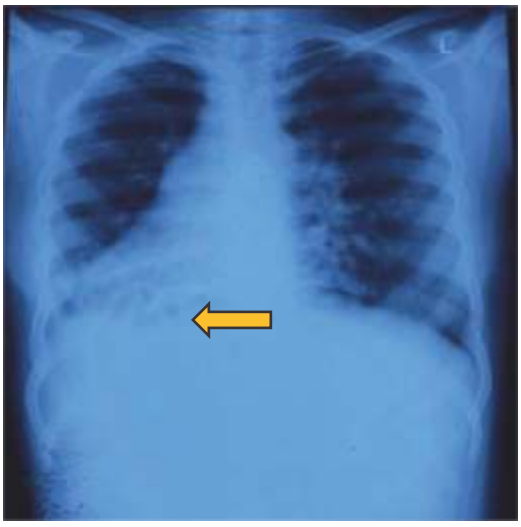

Figure 1: Right Cardiac apex

Chest X-ray (Postero-anterior (PA) view) revealed the cardiac apex pointing to the right. There was also a right-sided aortic arch and the right hemi-diaphragm presenting at a lower level than the left.

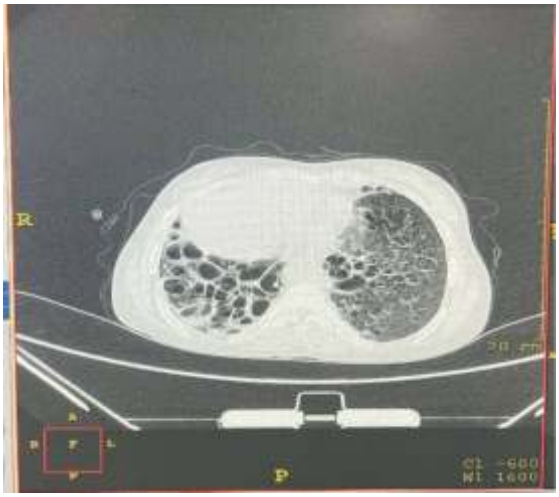

Figure 2: HRCT Chest

High-Resolution Computerised Tomography of the Thorax (HRCT-Chest**)** revealed the following:

1. Right-sided aortic arch with the prominent left ventricle, right ventricle and right atrium. There was also an appreciation of a prominent ascending aorta and MPA.
2. Well-defined cystic bronchiectasis changes in both lower lobes and lingular segment with some of the cysts showing air-fluid levels suggestive of infective aetiology.
3. Features of situs inverusus totalis.
4. Patchy consolidation in the posterior basal segment of the right lower lobe.
5. Streak of bilateral pleural effusion seen

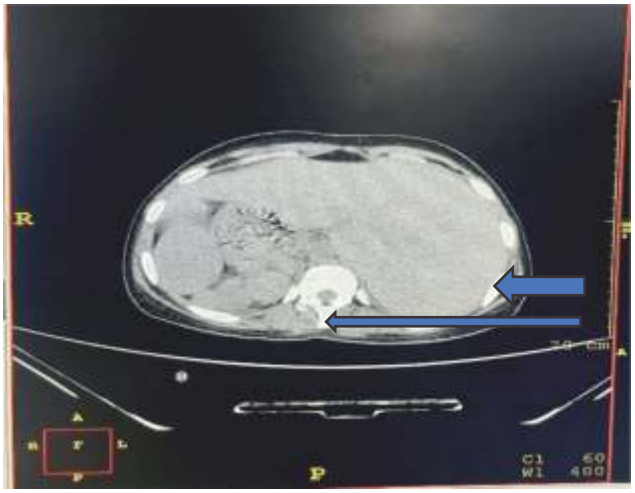

Figure 3: CT Abdomen

CT-Abdomen showed the following features:

1. Liver in the left hypochondrium.
2. Spleen in the right hypochondrium
3. Features suggestive of situs inversus totalis

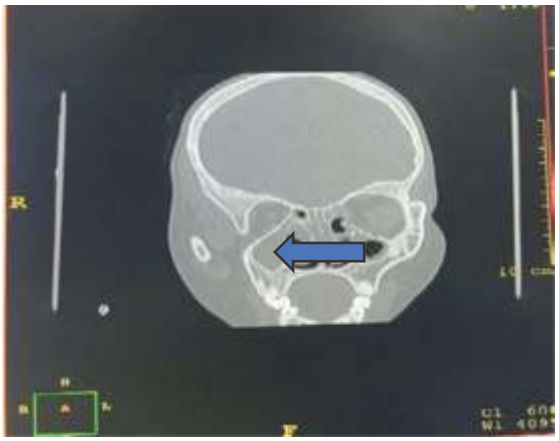

Figure 4: CT Paranasal Sinuses (PNS)

CT PNS revealed features suggestive of Maxillary, ethmoidal and frontal sinusitis.

Discussion

Keeping in mind the patient's clinical presentation and the findings obtained from the general and systemic examination coupled with the laboratory and radiological studies, a diagnosis of primary ciliary dyskinesia or KS was made. The patient in this case had displayed the classical findings of dextrocardia, situs inversus totalis and history of recurrent respiratory tract infections with an ongoing sinusitis infection (triad). These findings are classical of KS and thus, the diagnosis was made.

Normal ciliary function is essential for respiratory tract defence, normal organ orientation during embryogenesis and transport of the ova through the fallopian tubes/sperm motility. Mutations in DNAI1 and DNAH5 may result in structural abnormalities in cilia ranging from microtubular defects, lack or malfunctional radial spokes and dynein arms [4-5].

Diagnostic criteria of KS requires the presence of three key clinical features: (1) Abnormal or absent tracheobronchial clearance; (2) dextrocardia in the patient or in sibling; (3) alive sperm cells with impaired or absent motility/amenorrhea and/or infertility or subfertility in females; (4) ultrastructural defects in cilia as observed in electron microscopy; (5) laboratory screening for PCD helps in confirming the diagnosis but availability of the resources and the cost involved limits their usage. Estimation of exhaled nasal nitric oxide levels (less than 10% of normal), and saccharin test (prolonged ciliary clearance > 1 hour) are two of the routinely employed laboratory screening tests. High speed video microscopy to determine ciliary beat frequency (<11 Hz/s) and pattern and transmission electron microscopy to detect ultrastructural ciliary defects may also be employed. Genetic testing for DNAI1 and DNAH5 mutations are also carried out. In this case, lab screening tests and confirmatory tests were not carried out due to the lack of required resources.

Differential diagnoses for KS include malignancies, foreign body obstruction of the respiratory pathways and immunodeficient states such as IgG or IgA deficiency, leukocyte dysfunction [6]. Conditions such as Young's Syndrome (characterised by a triad of chronic rhinosinusitis with or without nasal polyp, azoospermia and bronchiectasis) and cystic fibrosis may resemble KS. The former though has been strongly associated with mercury exposure and usually presents in middle-aged men while the latter is due to mutations in the gene CF Transmembrane Conductance Regulator (CFTR) with bronchiectasis commonly affecting the upper lobes of the lung in the affected patients [7]. Genetic counselling [8] should be done when the diagnosis is made and the patient should be made aware of the details of their condition and the potential adjustments to their lifestyles that are to be made.

In most settings, diagnosis is often made on the basis of clinical signs and radiological evidence as there may not be adequate resources to run screening tests or confirm the syndrome. Moreover, patient affordability also becomes an important factor. While most patients lead near-normal lives, some may require bilateral lung transplantations in severe cases. DNase and mucolytic agents such as acetylcysteine and hypertonic saline may be tried in patients with respiratory symptoms or those with recurrent infections [9]. Surgical intervention may be rarely warranted but can be an option if the disease is localised [10].

Conclusion

It is imperative that such a diagnosis should be made as early as possible as supportive therapy can greatly enhance the quality of life in such patients. That being said, lifestyle modifications and education of the condition also plays a major role in the long-term survival of patients with KS. In this case, the patient and their family were educated on the necessary adjustments that they would have to make in terms of quality of life in order to produce the best prognosis. That being said, available treatment modalities were also explained while highlighting that supportive therapy would form the mainstay of the management of the disease. Our study therefore, highlights the classical clinical presentation of Kartagener's Syndrome in a young female and the challenges associated with it for the patient as well as the doctor.

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