Kartagener’s Syndrome: Presenting in Fourth Decade

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ABSTRACT

Kartagener’s Syndrome (KS) is a rare ciliopathic autosomal recessive genetic disorder characterized by a triad of situs inversus, bronchiectasis, and chronic sinusitis. Although Siewart first described this condition in 1904, it was Kartagener - a Swiss pediatrician, who recognized the etiological correlation between elements of the triad and reported four cases in 1933 1. It is a type of Primary Ciliary Dyskinesia (PCD). The frequency of KS is 1:32,000 to 1:50,000 live births.

A 40-year-old male patient presented to the emergency department with complaints of fever, productive cough, dyspnea, and headache. He had a history or a recurrent cough for ten years with multiple hospitalizations. The patient had been married for 15 years and still had no offsprings for which he had been investigated previously which was suggestive of azoospermia. He was febrile with normal other vitals accept his respiratory rate which was 28 per minute with 88% saturation on room air. Apex beat could be localized on 5th ICS right side on mid clavicular line. On percussion heart, borders were confirmed on the right side with liver dullness on left side.

On auscultation, S1S2 were heard on the right side with coarse, leafery crepitations on inspiration and rhonchi on expiration. On investigating further his ECG showed features of dextrocardia and chest roentgenography was suggestive of dextrocardia with aortic arch on right side of the trachea with gastro-hepatic transposition suggestive of situs inversus with bronchietatic changes. Further High resolution computed tomography was done to confirm those findings along with ultrasound of abdomen which showed liver in left upper quadrant and spleen in right upper quadrant. A skull roentgenogram was also done which was suggestive of sinusitis. The ‘nasal saccharin transit time’ test gave mucociliary clearance time of 60 minutes (Normal < 30 minutes). Thus patient was diagnosed with Kartagener’s syndrome clinically and also on the basis of investigations. The patient was treated with oral steroids, bronchodilators, mucolytics, and antibiotics and over the course of 10 days patient gradually recovered and was discharged.

Keywords: Kartagener’s Syndrome, Primary Ciliary Dyskinesia, Situs inversus

1. INTRODUCTION

Kartagener’s Syndrome (KS) is a rare ciliopathic autosomal recessive genetic disorder characterized by a triad of situs inversus, bronchiectasis, and chronic sinusitis. Although Siewart first described this condition in 1904, it was Kartagener - a Swiss pediatrician, who recognized the etiological correlation between elements of the triad and reported four cases in 1933 1. It is a type of Primary Ciliary Dyskinesia (PCD). The frequency of KS is 1:32,000 to 1:50,000 live births.
2. CASE REPORT

A 40-year-old male patient presented in the emergency room with complaints of fever of moderate grade with chills and rigors since one month, with productive cough of white colored expectorant since 20 days, dyspnea on exertion since five days and headache with running nose since three days.

The patient gave a history of recurrent respiratory tract infections (RTI) since last ten years with frequency of almost 6 to 7 episodes per year, for which he used to get medications from a local doctor and was admitted for same reason multiple times since then he was aware of his dextrocardia. He was married for 15 years and yet had no offsprings. His previous medical records revealed that he had azoospermia with low sperm motility.

On examination, the patient was febrile with 100°F, with normal pulse and blood pressure while his respiratory rate was 28 per minute with oxygen saturation of 88% on room air and respiratory muscles of respiration were prominent. The patient was conscious but anxious. Apex beat was localized to the right 5th intercostal space on mid clavicular line, and cardiac borders were confirmed on percussion on the right side while liver dullness was present in the left hypochondriac region. On auscultation, S1 and S2 were heard best on the right side with no murmur. On respiratory auscultation, there were bilateral infrascapular coarse, leathery crepitations on inspiration and rhonchi on expiration. The patient was managed with antibiotics and bronchodilators and shifted to medical intensive care unit.

On investigating further his ECG (figure 1) showed right axis deviation with positive QRS complexes with upright P and T waves in aVR, with global negativity in lead I and poor progression of R wave in precordial leads suggestive of dextrocardia and chest roentgenography (figure 2) was suggestive of dextrocardia as well with aortic arch on right side of trachea with gastro-hepatic transposition suggestive of situs inversus with bronchiectatic changes.

Fig.1: ECG suggestive of dextrocardia
Fig.2: Chest radiography suggestive of situs inversus

Fig.3: Bronchietatic changes on HRCT thorax

Further high resolution computed tomography (Figure 3) was done to confirm those findings along with ultrasound of abdomen which showed liver in left upper quadrant and spleen in right upper quadrant. Echocardiography showed dextrocardia although there was no structural or functional abnormality. His hemogram showed mild leucopenia with ESR of 60mm in the first hour. His sputum culture was positive for gram positive cocci suggestive of RTI. A paranasal roentgenogram (figure 4) showed sinusitis. Pulmonologists were also consulted, and bronchoscopy was done which revealed mucosal inflammation and mucopurulent secretions and reversal of bronchial anatomy. The ‘nasal saccharin transit time’ test gave mucociliary clearance time of 60 minutes (Normal < 30 minutes).

Thus patient was diagnosed with Kartagener’s syndrome clinically and confirmed by investigations. The patient was treated with oral steroids, bronchodilators, mucolytics, and antibiotics and over the course of 10 days patient gradually recovered and was discharged with proper counseling about the nature of the disease.

3. DISCUSSION

Disorders of ciliary motility may be congenital or acquired. Congenital disorders are labeled as PCDs. PCD is a phenotypically and genetically heterogeneous condition where polypeptide species within the axoneme of cilia, sperm flagella, ciliary membrane or matrix are defective(2,3). Pathophysiologically, the underlying defect leads to accumulation of secretions and consequent recurrent sinusitis, bronchiectasis & infertility. Currently, three genes (DNAI1, DNAH5, and DNAH11) that encode for dynein proteins (axonemal and cytoplasmic) have been linked to PCD. Because the embryonic and nodal cilia are defective, body asymmetry occurs randomly. Hence approximately 50 percent of the patients have situs inversus. Such cases of PCD causing
bronchiectasis and sinusitis with situs inversus are known as Kartagener's syndrome (KS). Respiratory epithelium has approximately 200 cilia per cell that beat in a coordinated fashion to move respiratory secretions. As situs inversus has no serious adverse health consequences per se, so the condition often goes undetected until a chest roentgenography is obtained. Most patients with KS present in childhood (median age of diagnosis 5 to 5.5 years), but some also present in adulthood with the mean age of is 22 years\(^4\).

Patients usually present with chronic URTI and/or LRTI due to the ineffective mucociliary mechanism. Some male patients present later in life with sterility due to immotile spermatozoa while cases of semisterility in females have also been reported\(^2\). Occasionally, KS may be associated with the reversible airflow obstruction hence patient may present with respiratory distress\(^5\).

Diagnostic criteria for this condition include clinical picture of recurrent chest infections and rhinitis since childhood, along with one or more of the following: (1) situs inversus ; (2) alive but immotile spermatozoa; (3) reduced or absent transbronchial mucociliary clearance and (4) cilia showing characteristic ultrastructural defect on electron microscopy\(^6\).

Examination usually reveals pale and swollen nasal mucosa, mucopurulent secretions, with or without the presence of nasal polyps and impaired sense of smell. The recurrent chronic sinusitis typically produces sinus pressure headache in the frontal, maxillary and periorbital region. Obstructive lung disease may be another component of KS. It probably results from elevated levels of local inflammatory mediators in a chronically irritated airway.

Apart from fulfilling the criteria mentioned above, two types of tests are done for diagnosis of PCD – screening tests: exhaled nasal nitric oxide measurement which is usually low and saccharin test to assess mucociliary function and diagnostic tests: ciliary beat pattern and frequency analysis using video recording and electron microscopic confirmation of the ultrastructural ciliary defect. The samples are obtained by biopsy taken during bronchoscopy or laparoscopically from tubal mucosa in females; semen analysis in a male patient.

Treatment of PCD is aimed at relieving symptoms and preventing complications. Early recognition of the disease and prompt antibiotic treatment are the keys to minimize the irreversible lung damage. Physiotherapy with postural drainage and cessation of smoking are also important. Coughing should not be suppressed since it acts as a substitute for mucociliary clearance. Huffing from mid to low lung volume with forced expiratory maneuver helps to improve clearance. The prognosis is generally considered favorable, and life expectancy is usually normal. It is necessary to monitor the progression of the lung disease on every clinical visit at regular intervals.

Thus, we should remember that any patient with a history of a recurrent cough and headache with infertility should be examined for Kartagener’s syndrome which is a part of PCD. As it is progressive, it has a greater impact on health in the second half of life, producing significant morbidity and restriction of daily activity. Early diagnosis and prompt management may improve symptoms and quality of life and complications can be delayed.

REFERENCES