

ORIGINAL ARTICLE

KARTAGENER SYNDROME: A NOT SO RARE PHENOMENON

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Background: Kartagener Syndrome is characterized by ciliary dyskinesia and is inherited in autosomal recessive manner. It occurs in 1:20,000–30,000 live births in general population. Its prevalence varies from region to region. **Methods:** This case series describes four patients of Kartagener Syndrome who were diagnosed in the departments of medicine and pulmonology between year 2009 and 2013. **Results:** The age of youngest patient was 15 years while the oldest patient was 19 years old. All of them were males. Mean age was 17.5 years. They had presented with history of shortness of breath and productive cough. One of them had presented with rhinorrhea and nasal blockage as the main symptom. **Conclusion:** Although Kartagener Syndrome is often thought of as a rare disorder, yet it might not be as rare as it is thought of. Considering this and the fact that half of the patients with Kartagener Syndrome do not have situs inversus, large scale studies with family trees are required to trace patients with Kartagener Syndrome.

Keywords: Kartagener Syndrome, Autosomal Recessive, Dextrocardia, Bronchiectasis

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INTRODUCTION

Kartagener Syndrome, also known as the primary ciliary dyskinesia or immotile cilia syndrome, is a triad of chronic sinus disease, bronchiectasis and situs inversus. This syndrome was first described in 1933 by Kartagener.^{1,2} It is believed that situs inversus which is present in only half of patients with Kartagener Syndrome², is the first event and results from a failed or faulty organ movement within 2 weeks of implantation. The other two features, bronchiectasis and chronic sinusitis, are a result of infective processes and retention of mucus after the birth.³

MATERIAL AND METHODS

This case series describes four patients of Kartagener Syndrome who presented to departments of Medicine and Pulmonology, Ayub Teaching Hospital, Abbottabad between 2009 and 2013. These patients had presented at different time with symptoms. All these patients were young adults and the most common presenting symptom was productive cough accompanying shortness of breath as the main complaint. Only one of these patients had presented with foul-smelling nasal discharge. Although he did report presence of productive cough in winter months. Detailed medical history was obtained from these patients and a thorough physical examination was done. Baseline investigations were done for each patient and HRCT for confirmation of diagnosis of bronchiectasis as well as situs inversus was also done when required. Only one patient consented to semen analysis. The detail of these cases is as follows in no particular order:

CASE-1

A 19 year old male presented to the OPD with a history of chronic productive cough, shortness of breath and

nasal obstruction. These symptoms have been present for past many years. Nasal discharge and intermittent nasal obstruction had been present since childhood. He had been having repeated chest infections especially in winter for the past couple of years and the amount of produced sputum was another cause of concern for the patient. On examination, the patient was not in respiratory distress. Grade-2 clubbing was noted. He had a deviated nasal septum with partial blockage of right nostril along with swollen inferior turbinates bilaterally. On chest examination, the apex beat was felt in right 5th intercostal space. On auscultation bilateral coarse crepitations were heard in the lower part of chest on either side. His TLC was 9.6×10^3 /uL. There were 71.2% granulocytes, 19% were lymphocytes and 9.8% were monocytes. He was anaemic with an Hb level of 9.6 gm/dl. Haematocrit was 33, MCV was borderline normal 79.3%, MCH was low, i.e., 23.1%, MCHC was normal (29.1%) and RDW was also normal (16.1%). Platelets were 82,000. Serum biochemistry was normal except for a slightly raised serum alkaline phosphatase levels. The ESR was 64 mm/1st hour. X-ray chest (Figure-1) was requested which showed heart on right side of the chest. In order to confirm the presence of bronchiectasis, a CT scan of chest was advised to patient which he refused because he could not afford the costs. X-ray PNS was ordered which showed haziness of the maxillary sinuses, more on right side and a deviated nasal septum. Semen analysis showed 35% immotile sperms in addition to the 20% sperms with sluggish motility. 40% of the sperms were morphologically abnormal. Situs inversus was noted on ultrasound abdomen. ECG showed a deviation of the axis to right side, Positive QRS complexes in lead avR, inversion of all waves in lead-I, and absence of R wave progression in chest leads. A diagnosis of Kartagener Syndrome was made.

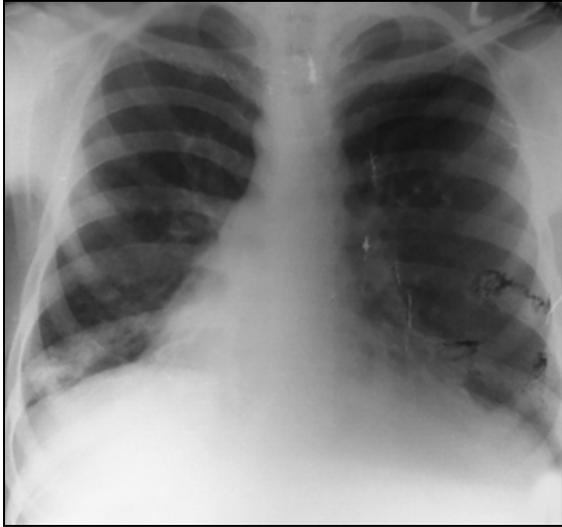


Figure-1: Chest X-ray of case-1.



Figure-2: X-Ray PNS of case-1

CASE-2

A 17 year old male was admitted in the pulmonology unit with a history of worsening exertional dyspnea, frequent exacerbations of “chest infection”, and productive cough. The patient was having productive cough and progressive dyspnea for the previous 8 years. He was diagnosed as a case of pulmonary TB 8 years ago and had completed the anti-tuberculosis treatment. His “chest infection” started after this and it kept progressing. Over the previous two years, the mobility of the patient had reduced to significant extent. He reported that he was fine when admitted to a hospital and would not last a few days at home. His elder brother informed the attending physician that their grandfather had “asthma” and had died young

because of it. On examination, patient appeared distressed and short of breath while sitting. Grade-3 clubbing was present. He was cyanosed. Chest examination revealed apex beat in right 5th intercostal space and bilateral coarse crepitations in the lower part of chest. X-ray showed heart in right side of the chest. An ECG confirmed the presence of dextrocardia and a subsequent echocardiography moderate RV systolic dysfunction and severe pulmonary hypertension were noted. CT scan chest showed extensive bilateral bronchiectasis and situs inversus. Ultrasound Abdomen done and it also showed situs inversus.



Figure-3: Chest X-ray of case-2

CASE-3

A 19 years old male presented to the pulmonology OPD with a history of chronic productive cough, frontal headaches and nasal discharge since childhood. He also gave history of repeated episodes of fever and bad chest for the last 7–8 years. According to the available record, he had consulted many physicians for his symptoms but no relief was provided despite having used many medications. There was no other medical history. He had not had any surgery in the past except for repeated admissions for the same problems over past many years. On physical examination, the patient was having fever of 39.4 C. He did not have clubbing. His blood pressure was 120/80 mmHg. He had a slightly deviated nasal septum, with hypertrophied inferior turbinates. On chest examination he had bilateral coarse crackles as well as scattered rhonchi audible on auscultation with the stethoscope. Apex beat was palpable in the right 5th inter-costal space where heart sounds were best audible. The X-ray chest showed

dextrocardia and the ECG confirmed it. A CT scan of chest was ordered which showed extensive bilateral bronchiectasis and situs inversus. He was admitted to the pulmonology unit for further management.

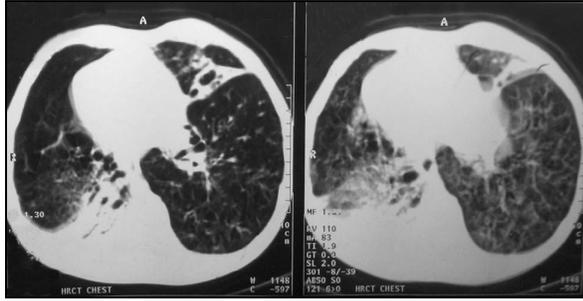


Figure-4: CT Chest of case-3.

CASE-4

A 15 year-old male presented to the OPD with a history of repeated throat infections and productive cough which, initially was subsequent to the throat infections but later became permanent, for the last 5 years. His father was worried about the patient's falling health and stated that the boy was not gaining weight and had lost 9–10 KG weight during this period. The patient had recently begun producing large quantities of sputum and this was the main reason for presenting to the OPD. On examination the patient was visibly short of breath and was coughing. His temperature was 39 C. Blood pressure was 115/75 mmHg. He was mentally alert and well oriented in time, place and person. He had grade 2 clubbing but no pallor or jaundice was present. Palpation of chest revealed the apex beat to be in the right part of chest where the heart sounds were clearly audible when compared with the left side of the heart. On auscultation of the chest diffuse bilateral coarse crackles over both infra scapular regions as well as rhonchi were heard. Rest of the physical and systemic examination was normal. A provisional diagnosis of dextrocardia was made which was confirmed with an X-ray and an ECG. A CT scan chest showed diffuse bronchiectasis in lower lobes bilaterally. The diagnosis was changed to Kartagener Syndrome and patient was admitted to the medical unit for further management.

DISCUSSION

There are many causes of infections of the upper and lower respiratory tract. Most common infections in our region include bacterial infections, mycobacterial infections and viral infections. A study of a multitude of factors that might influence the prevalence of upper as well as lower respiratory tract infections in young population in Pakistan concluded that climate had a major role in prevalence of upper and lower respiratory tract infections.⁴ This situation demands

that when a patient presents with symptoms and signs of respiratory infection, the commonest causes need to be considered. Only when the patient doesn't improve with conventional treatments, alternative diagnoses are sought. Kartagener Syndrome is one such example. It should be considered with an early-onset bronchiectasis. It is a rare autosomal recessive disorder characterized by abnormal function of cilia in most cases.⁵ Most of the clinical manifestations of Kartagener Syndrome e.g., otitis media, bronchiectasis, recurring episodes of rhinosinusitis, infertility in men, recurrent pneumonias etc are a result of the abnormal function of the cilia. Since Kartagener first described the classical triad of this syndrome, several hundred research papers have been published covering various aspects of this syndrome.⁵ The prevalence of Kartagener syndrome is variable: from 1 in 8000 in Scandinavia to 1 in 11,000 in the US.⁵ There are no exact prevalence data of Kartagener Syndrome in Pakistan. However sporadic cases of Kartagener Syndrome or situs inversus have been reported.^{6–10} Although situs inversus is present only in half of the patients with Kartagener Syndrome, its presence in our patients made it easier for us to diagnose the syndrome. Having once been described by Aristotle as the “punishment by gods”⁶, it is now known to be part of a group of diseases called primary ciliary dyskinesias⁸, in which the main defect is in the dynein arms of cilia in up-to 96% of cases.¹¹

This defect in the cilia is responsible for many manifestations of the syndrome including infertility in men. Women with KS can become pregnant but men are usually infertile.⁸ The presence of classical triad is usually sufficient to alert the physicians to the diagnosis, however confirmation of diagnosis requires certain specialized tests including biopsy, genetic studies, semen analysis including sperm count and sperm motility evaluation and measurement of exhaled nitric oxide which probably has a good role in ruling out the syndrome than confirming the diagnosis.¹²

This condition is best managed conservatively with chest physiotherapy, treatment and prevention of respiratory infections and supportive care including vaccination and LTOT is usually required. One of our patients presented to us with severe pulmonary hypertension.

In addition to pseudomonas infections, the commonest microbial organisms found in the patients with KS are *Staphylococcus aureus* and *Hemophilus influenzae*.¹³ Patients usually require long-term prophylactic oral or intravenous antibiotics. Inhaled bronchodilators, steroids and mucolytics can be prescribed for the management of obstructive lung disease.¹⁴ Surgical options for Kartagener Syndrome

include tympanostomy tubes, endoscopic sinus surgery, in case of pulmonary disease lobectomy, lingulectomy, segmentectomy and lung transplantation & heart-lung transplantation.¹⁴

CONCLUSION

We feel that Kartagener Syndrome is not as rare as it is thought to be. The chest physicians should keep a low threshold for these cases especially when 50% of Kartagener syndrome can present with no situs inversus. Proper and timely management of these cases can delay many of the sequelae of this syndrome if not eliminate them all.

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