JALALABAD MEDICAL JOURNAL

Case Report

A Case Report on Kartegener's Syndrome

Habiba Jamila Khan[1], Naznin Akther[2], Ashith Chandra Das[3], Rabiul Hasan[4], Tahmina Jahan Chowdhury[5], Jannatul Ferdush Chowdhury[6]

[1,7]Registrar, Department of Paediatrics, Jalalabad Ragib-Rabeya Medical College Hospital, Sylhet.
[2,5,6]Assistant Professor, Department of Paediatrics, Jalalabad Ragib-Rabeya Medical College, Sylhet.
[3,4]Associate Professor, Department of Paediatrics, Jalalabad Ragib- Rabeya Medical College, Sylhet.

ABSTRACT

Kartagener's syndrome is a rare, autosomal recessive genetic ciliary disorder that comprises the triad of situs inversus, chronic sinusitis, and bronchiectasis. The basic problem lies in the defective movement of cilia, leading to recurrent chest infections, ear, nose and throat symptoms, and infertility. We are hereby going to report a case of a 9-year-old female child with Kartegener's syndrome from our hospital, diagnosed clinically and by relevant radiological investigations.

Keywords: *Bronchiectasis, Kartagener's syndrome, Sinusitis, Situs inversus.*

[Jalalabad Med J 2021; 18 (2): 59-62]

INTRODUCTION

Kartagener's syndrome (KS) is a subgroup of ciliary motility disorders called primary ciliary dyskinesias (PCDs). Although an X-linked inheritance has been reported, it is an autosomal recessive hereditary disorder with a triad of situs inversus, bronchiectasis, and sinusitis[1,2]. Seiwert first described this condition in 1904, but it was Manes Kartagener, who established the etiological association between the members of the triad and documented four cases in 1933[3]. The estimated prevalence of PCD is about 1 in 30,000, though it may range from 1 in 12,500 to 1 in 50,000, but its prevalence in children with repeated respiratory infections has been estimated to be as high as 5%[2,3]. The extreme structural genetic abnormality in KS produces ciliary motility impairment, which leads to recurrent chest, ear, nose, and throat infections, as well as infertility. A high index of suspicion is required in order to make an early diagnosis and offer prompt treatment choices for infertility in these young patients. Also, although unproven, it seems likely that early diagnosis is important for the preservation of pulmonary function, quality of life, and life expectancy in this disease[4,5].

Address of Correspondence:

Dr. Habiba Jamila Khan, Registrar, Department of Paediatrics, Jalalabad Ragib-Rabeya Medical College Hospital, Sylhet. Mobile: +880171689315. Email: habibasorin@gmail.com

CASE REPORT

A 9-year-old female child weighing 22 kg, the youngest one of three siblings of non-consanguineous parents, was admitted with a history of productive cough and respiratory difficulties for the last 15 days and a fever for the last 10 days. The cough was productive in nature and associated with mucopurulent sputum, copious in amount, especially in the morning. The cough was worsening day by day, but didn't interfere with her daily activities. She had a history of recurrent headaches and similar episodes of illness since her infancy and was treated with multiple oral antibiotics each time. On examination, she was malnourished (BMI of 13, which was less than the 5th centile), conscious, and oriented. Her pulse rate was 90 beats per minute, respiratory rate was 30 breaths/minutes, the temperature was 102°F, and

clubbing was present. On auscultation, there were coarse crepitations over both lung fields. According to the CVS examination, the apex beat was on the right 5th intercostal space. The heart sound was better heard on the right side, with no murmur. The upper border of liver dullness was not found in the right 5th intercostal space, and no organomegaly was found on abdominal examination. Laboratory studies revealed an Hb% of 12.7 gm/dl, a WBC count of 15.3 K/microL, a platelet count of 310 K/microL, an ESR was 44 mm in the 1st hour, febrile antigen test was negative, urine R/E was normal, and C/S showed no bacterial growth. The MT test was negative. CXR P/A view revealed dextrocardia, bronchiectatic changes in the left lower lung zone, and a sub diaphragmatic fundic gas shadow on the right side (Figure-1). The paranasal air sinuses were consistent with bilateral maxillary sinusitis (Figure-2). A CT scan of the chest revealed a left middle lobe collapse consolidation with bronchiectatic changes, as well as a right-sided pulmonary inflammatory lesion, dextrocardia, and situs inversus (Figures 3a and 3b). A diagnosis of Kartagener's syndrome was made on the basis of features of clinical presentation and imaging features. The patient was treated with parenteral antibiotics, antipyrectics, mucolytics, and physiotherapy. After 13 days of hospitalization, she became symptom free and was discharged with the advice of frequent follow-up, but unfortunately, she missed the follow-up.

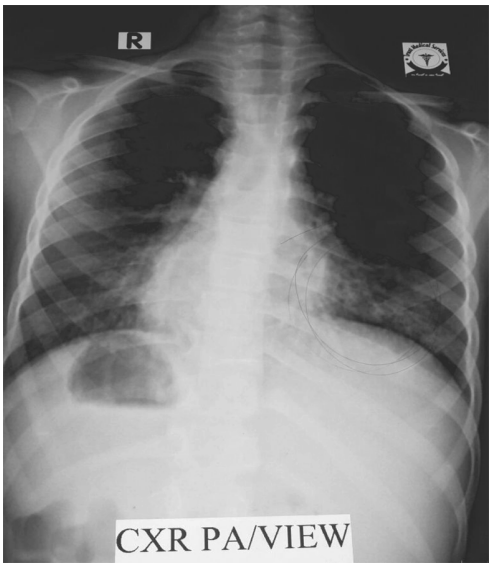


Figure-1: Dextrocardia, Bronchiectatic change with situs inversus.

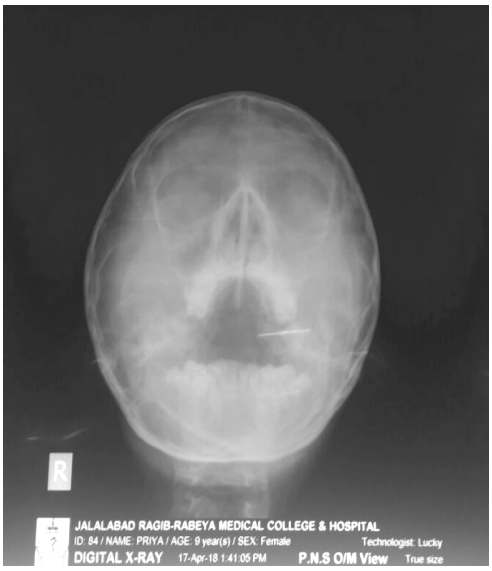
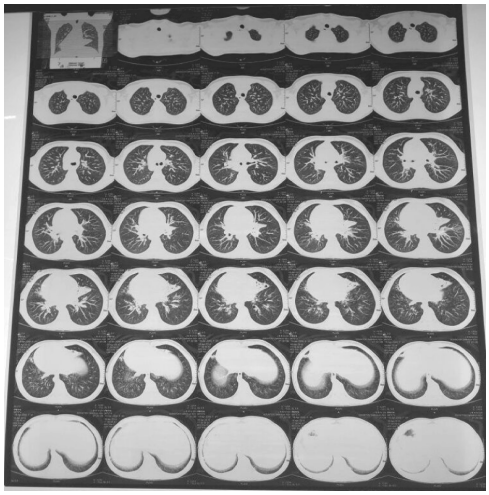
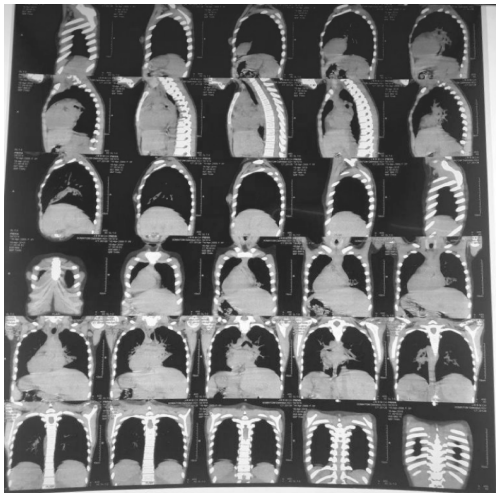


Figure-2: Maxillary sinusitis.



(a)



(b)

Figure-3a & 3b: CT scan of chest revealed middle lobe collapse consolidation with bronchiectatic changes and pulmonary inflammatory lesion (Rt), dextrocardia and situs inversus.

DISCUSSION

Disarrays of ciliary motility might be congenital or acquired. Congenital disarrays are categorized as PCDs. Almost half of PCD patients have situs inversus. Such cases of PCD with situs inversus are established as Kartagener's syndrome. PCD is a phenotypically and genetically heterogeneous condition with the primary defect being in the ultra structure or funnction of cilia[5,6]. Such defects are noted in roughly 90% of PCD patients and entail the outer dynein arms, inner dynein arms, or both. About 38% of PCD patients carry mutations of the dynein genes DNAI and DNAH5[7-9]. It has been suggested that typical ciliary beating is essential for visceral revolving at the time of embryonic development. In patients with PCD, half of the patients will have situs inversus, i.e., they will be the cases of KS and the other half will be the typical situs because of random rotation. Pathophysiologically, the primary defect that causes the accumulation of secretions and subsequent recurring sinusitis, bronchiectasis, infertility, and situs inversus is defective ciliary motility or immotility. The rigorousness of symptoms and the era at which the condition is diagnosed are reasonably erratic, even though the symptoms have been present since birth. Clinical progression of the disease is inconsistent with lung transplantation, which is required in severe cases. Diagnostic criteria for this condition comprise a clinical picture indicative of recurring chest infections, bronchitis, and rhinitis since childhood, together with one or more of the following: (1) a patient/sibling with situs inversus; (2) alive but immotile spermatozoa; (3) reduced or absent transbronchial mucociliary clearance; and (4) cilia with ultrastructural defects on electron microscopy. Our patient was a 9-year-old female child who had presented with recurrent chest infections since infancy, along with sinusitis and rhinitis, in addition to situs inversus and dextrocardia. Apart from fulfilling the criteria mentioned above, two types of tests are done for the diagnosis of PCD– screening tests (Exhaled nasal nitric oxide measurement, which is usually low in PCD, and saccharin test to evaluate mucociliary function of the nasal epithelium) and diagnostic tests (Ciliary beat pattern and frequency analysis using video recording, and electron microscopic confirmation of the ultrastructural ciliary defect). The samples for those tests for examining the motility and ultrastructure of cilia can be obtained by taking biopsies from both the nasal and tubal mucosa in females[9]. Abnormal laboratory findings in KS include low nasal nitric oxide level (~10% of normal), prolonged saccharin clearance time (>1 hour), low ciliary beat frequency (<11 Hz/second), absent ciliary ultrastructure (Dynein arms), and mutated DNAI1 and DNAH5 gene[10,11]. However, in our case, we could not perform these tests, and so the diagnosis was based upon clinico-radiological findings. There is no definitive curative treatment for the complete recovery of KS. The purpose of the treatment is to clear up respiratory system secretions and get control over infections. Therefore, antibiotic treatment is prescribed for acute bacterial exacerbations along with bronchodilator therapy and chest physiotherapy, which are recommended for the excretion of secretions. In addition, non-surgical methods such as the destruction of the bronchial wall and irreversible dilatation are defined for the treatment of bronchiectasis as first-choice methods. The intention of surgical treatment in bronchiectasis is to resect all bronchiectatic areas to make the patient more asymptomatic. Patients with KS having severe problems because of bronchiectasis may undergo bronchiectasis surgery. A variety of organ pathologies might be reported with KS, including congenital heart disease, esophageal disease, biliary atresia, hydrocephalus, Marfan’s syndrome, renal and pancreatic dysplasia, Hirschsprung’s disease, varicocele, and small intestine malrotations, polysplenia, and kyphoscoliosis[12]. Prakash S.B and Leela Tejaswini mentioned a case of an 8-year-old boy with a common cold, productive cough, and throat pain for 2 years. His X-ray chest revealed dextrocardia, chronic bronchitis, and pansinusitis in addition to an enlarged adenoid and situs inversus, which was confirmed by USG and CT abdomen. Another case reported by Prakash S.B and Leela Tejaswini stated that a 10-year-old female child with a history of episodic rhinorrhea, productive cough, throat pain, and nasal obstruction. She had dextrocardia, chronic sinusitis, and situs inversus revealed by USG[13]. In addition, Manjunath S. Pandit et al. also reported a case of a 7-year-old girl who presented with a history of repeated nasal blockage, thick nasal secretion, and multiple hospital visits with RTI since 2 years of age, along with an acute onset of fever and cough. Additionally, she had dextrocardia with situs inversus[14]. Furthermore, some other findings were also found in Kartegener's syndrome, such as nasal polyps, chronic nasal obstruction with nasal discharge, hyposmia (Bilateral), enlarged adenoid, pectus carinatum, and fertility[12,13]. Both the reported cases of Prakash S.B. and Leela Tejaswini had

adenotonsillectomy under general anesthesia in addition to medical treatment. It is worth mentioning that all of the cases, including our reported case, were managed by proper counseling, antimicrobial therapy, and chest physiotherapy.

CONCLUSION

Kartegener's syndrome is a rare genetic disorder that often results in significant morbidity and mortality. It may prove challenging if a high index of suspicion is not made. It is believed that the progression and extent of lung disease can be slowed with early diagnosis and therapy. Thus, routine surveillance studies recommended for the care of children with PCD include regular spirometry, chest imaging, and sputum or oropharyngeal cultures to assess respiratory flora.

REFERENCES

1. Barthwal MS. Kartagener's syndrome in a fertile male- An uncommon variant. Lung India 2006; 23 (3): 123-5.
2. Ferkol T. Primary Ciliary Dyskinesia (Immotile Cilia Syndrome). In: Nelson Text Book of Pediatrics. Stanton BF, Schor NF, St Geme JW, Behrman RE, editors. 21st ed. Philadelphia: Saunders, Elsevier; 2019. p 2113-16.
3. Seaton D. Bronchiectasis. In: Seaton A, Seaton D, Leitch AG, editors. Crofton and Douglas's respiratory diseases. 5th ed. Oxford: Blackwell Science; 2004. p 794-828.
4. Barbato A, Frischer T, Kuehni CE, Snijders D, Azevedo I, Baktai G, et al. Primary ciliary dyskinesia: A consensus statement on diagnostic and treatment approaches in children. Eur Respir J 2009; 34 (6): 1264–76.
5. Marthin JK, Petersen N, Skovgaard LT, Nielsen KG. Lung function in patients with primary ciliary dyskinesia: A cross-sectional and 3-decade longitudinal study. Am J Respir Crit Care Med 2010; 181 (11): 1262-8.
6. Mahsud I, Shahab-ud-Din. Kartegener's syndrome: a case report. Gomal J Med Sci 2006; 4 (2): 79-81.
7. Chodhari R, Mitchison HM, Meeks M. Cilia, primary ciliary dyskinesia and molecular genetics. Paediatr Respir Rev 2004; 5 (1): 69-76.
8. Chilvers MA, Rutman A, O'Callaghan C. Ciliary beat pattern is associated with specific ultrastructural defects in primary ciliary dyskinesia. J Allergy Clin Immunol 2003; 112 (3): 518-24.
9. Zariwala MA, Knowles MR, Omran H. Genetic defects in ciliary structure and function. Annu Rev Physiol 2007; 69 (1): 423-50.
10. Lobo LJ, Zariwala MA, Noone PG. Ciliary dyskinesia: primary ciliary dyskinesia in adults. Eur Respir Mon 2011; 52: 130-49.
11. Rafi MK. Katagener's syndrome –A rare case series in female patients. Indian J Med Case Rep 2016; 5 (4): 33-40.
12. Tokur M, Tokur N, Yenibertiz D, Korkmaz C. A case of the kartegener syndrome accompanied by pectus carinatum deformity. J Clin Anal Med 2014; 5 (6): 519-20.
13. Prakash SB, Leela Tejaswini RM. Kartegener's syndrome: a case series study. J Evol Med Den Sci 2015; 4 (65): 11404-11.
14. Pandit MS, Gupta A, Mathur P, Sharma M, Garg M, Kumar R. A case report of Kartagener syndrome. Sch J Med Case Rep 2016; 4 (6): 409-11.