Case Study

Kartagener's Syndrome – a triad or tetrad?

Akshay Mehta[1], Lakshay Mehta[1], Jyoti Gupta[2], Bhawna Malik[3]

Received on:
01-May-2019

Accepted for Publication:
30-Jun-2019

Correspondence to:
Akshay Mehta, Swansea, United Kingdom
akshaymehta88@yahoo.in

Author's Affiliation:
1- Department of Neonates, Singleton Hospital, Swansea, Wales, United Kingdom
2- Fortis Memorial and Research Institute, Gurugram, Haryana, India
3- KJIVF Centre, New Delhi, India

Abstract:

Kartagener's Syndrome is an autosomal recessive disease characterized by triad of situs inversus, bronchiectasis and sinusitis. This anarchy affects the activity of ciliary cytoskeletal proteins important for the movement of cilia, thus causing multisystemic manifestation of varying severity.

Apart from highlighting clinical case of infrequent incidence, including a brief literature review emphasizing the need of evaluating the patients presenting with recurrent cough and cold in detail for primary ciliary dyskinesia associated with Kartagener's Syndrome and stressing the importance of early diagnosis and management; this paper also intends to share an observation of presence of malrotation in two unrelated cases of Kartagener's syndrome each one of which had malrotation with volvulus as an associated feature and thus discussing the possibility that Kartagener's syndrome might be tetrad rather than a triad as we consider it.

Keywords: Kartagener's syndrome, Bronchiectasis, Sinusitis, Situs inversus, Malrotation

Introduction

Kartagener's Syndrome was described for the first time by Siewert in 1904 but the minutiae of the syndromic associations were specified by Manes Kartagener.1 Basic problem is in ciliary motility and this entity is also known as primary ciliary dyskinesia. When associated with situs inversus, the patients with Primary Ciliary Dyskinesia (PCD) are labelled as Kartagener's Syndrome.2 We present two case reports whose participation involved informed consent.

Case 1

A 10 year old female presented in OPD with productive cough with postural variation and breathlessness on exertion of 15 days duration with history of recurrent attacks of respiratory tract infection since early childhood with no seasonal variation. Fever and headache accompanied most of these episodes. Patient had received several treatments including anti tubercular drugs in the past but with not much response. Patient also had a past history of bilious vomiting on Day 2 of life for which patient was investigated and diagnosed with malrotation with midgut volvulus and was operated for the same on Day 8 of life (confirmed with previous medical records). Parents were informed then that the patient has situs invertus. No investigations were however done on patient thereafter till her presentation to our hospital. On examination, she was febrile but with no signs of respiratory distress. Tenderness was present over the maxillary sinuses. On chest examination, wheeze and crepitations were present bilaterally but more on the right lower lobe region. Cardiac pulsations were seen and felt predominantly on the right side. Trachea was, however, centrally positioned ruling out mediastinal shift. Physical examination of the abdomen revealed surgical scar in paraumbilical region and liver dullness on the left side. On investigations, Mucosal thickening and opacified sinus cavities suggestive of chronic sinusitis were seen on sinus radiography. Chest X-ray suggested a right-sided cardiac shadow with a wedge-shaped opacity in the right retrocardiac area suggestive of bronchiectasis. ECG showed waveform patterns suggestive of dextrocardia. Echocardiogram confirmed the diagnosis of dextrocardia. Ultrasonography abdomen revealed spleen on the right side of the abdomen while liver on the left. Barium swallow depicted presence of fundal gas shadow on right side. CT Chest suggested presence of bronchiectasis of the medial segment of right lower lobe and also the inverted pattern of major bronchi. Sputum microscopy for AFB and sputum culture and sensitivity were negative. Considering the clinical condition of the patient and corroborating it with investigations, diagnosis of Kartagener's Syndrome was made.

Case 2

A 12 year-old boy presented to our institute with recurrent history of cough with yellowish expectoration intermittently for past five years; often associated with fever and wheezing. Patient had been managed with antibiotics with occasional steroids prior to visiting us. He underwent Ladd's procedure in early post natal period in view of malrotation with midgut volvulus which was diagnosed based on upper GI series (confirmed with past medical records available with the patient). On examination he was dyspenic, febrile and grade 2 clubbing was present. Auscultation of the chest revealed diffuse bilateral rhonchi with scattered crepitations; with heart sounds appreciable on the right. Apex beat was palpable over the fifth intercostal space on the right side of chest. Surgical scar was noticed in supra umbilical region. Rest of the examination was normal. Chest X-ray, ECG and Echocardiography confirmed dextrocardia. Chest X-ray was also suggestive of increased bronchovesicular markings and suspicious bronchiectatic features. Ultrasonography of abdomen revealed situsinversus. HRCT chest confirmed bronchiectatic changes. X-ray and CT para nasal sinus revealed pansinusitis. Based on these findings diagnosis of Kartagener's syndrome was confirmed. Condition was explained to the parents and the patients were treated with antibiotics, antipyretics, mucolytics and inhaled bronchodilators in both the cases and are being observed on regular follow-up basis.

Discussion

Kartagener's Syndrome has a prevalence between 1/15000 to 1/32000 in general population and is characterized by a triad of situs inversus, bronchiectasis and sinusitis.1 Embryonic cilia located in the embryonic node are presumed to propel morphogens and helps in development of embryonic sidedness. Abnormal nodal cilia results in Situs Inversus which is the earliest and most striking component of the triad and is characterized by location of the internal organs on the opposite side of the body from the normal position.3 Involvement of airway cilia results in defective mucociliary clearance of airway secretions predisposing for recurring respiratory tract infections ultimately resulting in bronchiectasis.4 Pansinusitis results from accumulation of secretions inside the paranasal sinuses secondary to the ciliary dysfunction.5 Other manifestations include infertility due to defective sperm motility or poor ovum transfer in fallopian tube. Very rarely ciliary dyskinesia may result in hydrocephalus because of involvement of ventricular cilia which are located on the ependymal lining of the brain.2 Intestinal malrotation is defined as congenital abnormal position of the bowel within the peritoneal cavity and usually involves both small and the large bowel. It is accompanied by abnormal bowel fixation by mesenteric bands or absence of fixation of portions of

the bowel, leading to increased risks of bowel obstruction, acute or chronic volvulus, and bowel necrosis.6 The gut develops from the yolk sac; grows rapidly and by 12 weeks, after a series of complex steps involving a total rotation of 270°, it undergoes fixation in the normal position in the abdomen. Various degrees of malrotation of the small or large bowel may occur, depending on the developmental stage at which normal embryologic rotation failed.7 Malrotation of the bowel is associated with a number of syndromes.8 We could not find any reported association between intestinal malrotation and Kartagener's syndrome. One possible explanation that the authors propose: Ciliary dyskinesia which is primary pathology in Kartagener's syndrome might affect rotation and fixation of gut as well and thus, result in malrotation. We also request others to revisit past history of patients with Kartagener's syndrome in their follow up so as to arrive at some conclusion whether Kartagener's syndrome is a quadriad or its mere observational coincidence. Apart from this we also propose that every patient who presents with history of recurrent cold and cough responding minimally to appropriate and prolonged treatment should be examined in detail, for it can timely reveal a rare but potentially dangerous clinical entity like Kartagener's Syndrome. Apart from improving the quality of life by a guided treatment, it can also forecast few important issues like infertility in adulthood and also surgical intervention, if needed later in life can be planned accordingly. We also condemn the practice of giving antitubercular treatment without proper examination and investigation like done in our patient (Case 1) prior to visit to us.

References

1. Seaton D. Bronchiectasis. In: Seaton A, Seaton D, Leitch AG(ed). Crofton and Douglas's respiratory diseases (5th ed). Oxford: Blackwell Science, 2004: 794–828.

2. 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: 1264–1276.

3. Afzelius BA. A human syndrome caused by immotile cilia Science 1976; 193: 317-319.

4. Coren ME, Meeks M, Morrison I, Buchdahl RM, Bush A. Primary ciliary dyskinesia: age at diagnosis and symptom history Acta Paediatr 2002; 91667–91669.

5. Greenstone M, Stanley P, Cole P, Mackay I. Upper airway manifestations of primary ciliary dyskinesia J Laryngol Otol. 1985; 99(10): 985–991.

6. Strouse PJ. Disorders of intestinal rotation and fixation ("malrotation") Pediatr Radiol 2004; 34: 837–51.

7. Applegate KE, Anderson JM, Klatte E. Intestinal malrotation in children: a problemsolving approach to the upper gastrointestinal series Radiographics. 2006; 26: 1485-1500.

8. Taybi H, Lachman R. Radiology of syndromes, metabolic disorders, and skeletal dysplasias (3rd ed). Chicago, III: Yearbook Medical Publishers. 1990; 825–826.