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A Case Report of Kartagener Syndrome

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Abstract: Kartagener syndrome/Primary ciliary dyskinesia (PCD) is a rare ciliopathic autosomal recessive genetic disorder that causes defects in the action of cilia lining the respiratory tract (both lower and upper, sinuses, eustachian tube, middle ear) and fallopian tube as well as in the flagella of sperm cells. In 1933, Kartagener described a unique syndrome characterized by the triad of situs inversus, chronic sinusitis, and bronchiactesis, which was named Kartagener syndrome. Our case presented with recurrent respiratory tract infections with situs inversus, nasal polyps and early pulmonary changes of bronchiactesis.
Keywords: Ciliopathy, situs inversus, bronchiactesis, nasal polyp.

INTRODUCTION

Primary ciliary dyskinesia (PCD)/ Kartagener syndrome is an autosomal recessive disease with extensive genetic heterogeneity characterized by abnormal ciliary motion and impaired mucociliary clearance. In 1933, Kartagener described a unique syndrome characterized by the triad of situs inversus, chronic sinusitis, and bronchiactesis, which was named Kartagener syndrome [1]. Ultra structural and functional defects of cilia result in the lack of effective ciliary motility, causing abnormal mucociliary clearance. This leads to recurrent or persistent respiratory infections, sinusitis, otitis media, and male infertility. In 50% of the patients, ICS is associated with situs inversus [2].

The main consequence of impaired ciliary function is reduced or absent mucus clearance from the lungs, and susceptibility to chronic recurrent respiratory infections, including sinusitis, bronchitis, pneumonia, and otitis media. Progressive damage to the respiratory system is common, including progressive bronchiactesis beginning in early childhood, and sinus disease (sometimes becoming severe in adults) [3].

PCD/ Kartagener syndrome is a genetically heterogeneous disorder affecting motile cilia which are made up of approximately 250 proteins [4]. Around 90% of individuals with PCD/ Kartagener syndrome have ultra-structural defects affecting protein(s) in the outer and/or inner dynein arms which give cilia their motility, with roughly 38% of these defects caused by mutations on two genes DNAI1 and DNAH5, both of which code for proteins found in the ciliary outer dynein arm [5]. Absence of nodal ciliary function leads to defects such as situs inversus or heterotaxy. Splenic abnormalities such as polysplenia, asplenia and complex congenital heart defects are more common in individuals with situs ambiguous and PCD [6].

Studies have confirmed that ciliary beat pattern is associated with specific ultra structural defects in PCD/ Kartagener syndrome. New high-resolution digital high-speed video (DHSV) imaging has allowed the precise beat pattern of cilia to be viewed in 3 different planes in slow motion or frame-by-frame [7]. Gold standard for diagnosis of PCD is transmission electron microscopy.

CASE REPORT

A 7 year old girl presented to us with a history of repeated nasal blockage, thick nasal secretions and multiple hospital visits for recurrent respiratory tract infections since 2 years of age. Acute presentations were fever and cough for 6 days.

On examination her apex beat was felt on the right side and liver was palpable on the left side whose span was normal. On auscultation she had localized crackles on the left infra-mammary area and left sub-scapular area. Patient also had right sided nasal polyp between middle and inferior turbinate. Chest X-ray demonstrated patchy consolidation left lower zone and dextrocardia(Fig-2). Ultra sonography abdomen showed liver on the left side and a single spleen on the right side(Fig-3). HRCT chest reported diffuse ground glass haziness with lobar consolidation and dextrocardia with right sided aortic arch.

Based on the history and physical findings a diagnosis of Kartagener syndrome was made. Chest physiotherapy and appropriate antimicrobial therapy was advised, child is on follow-up.

Fig-1: Physical appearance of the patient

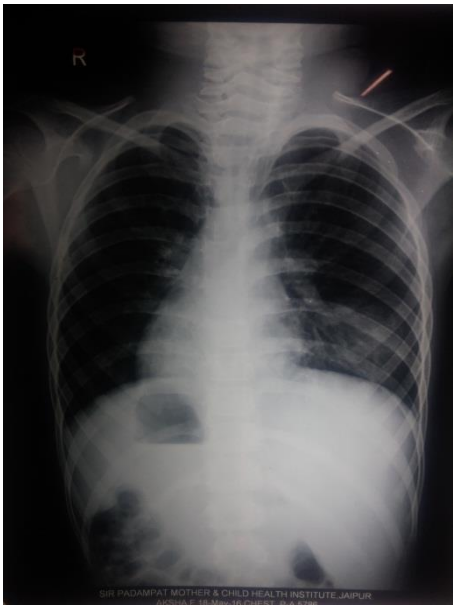

Fig-2: Chest X-ray

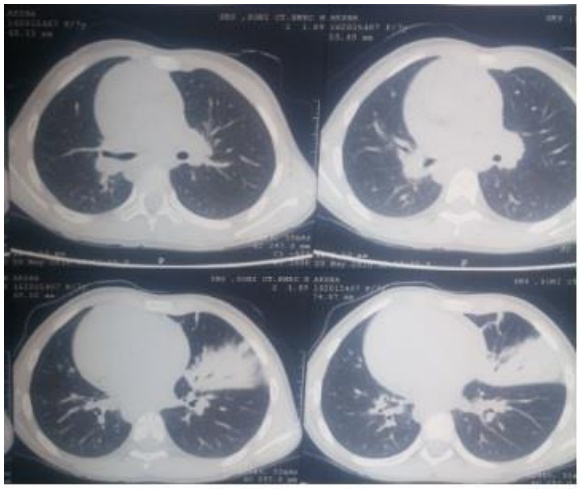

Fig-3: HRCT of Chest

DISCUSSION

Primary ciliary dyskinesia/Kartagener syndrome is genetically inherited disorder. Structures that make up the cilia including inner and/or outer dynein arms, central apparatus, radial spokes, etc. are missing or dysfunctional and thus the axoneme structure lacks the ability to move [8]. Dysfunction of the axonemal structure has been linked to the emerging class of disorders collectively known as ciliopathies, which includes PCD/Kartagener syndrome, Bardet-Biedl syndrome, hydrocephalus, polycystic kidney disease, polycystic liver disease, nephrolithiasis, Meckel-Gruber syndrome, and Joubert syndrome [9].

Defects in the ciliary component cause abnormal ciliary movements, resulting in impaired mucociliary clearance and manifesting as recurrent and or persistent sinopulmonary infections. In some patients with typical clinical manifestations of PCD and low levels of nasal nitric oxide, the ciliary ultra structure may appear normal suggesting functional abnormalities because of other defects in ciliary components [10].

Laterality defects are found in 50 % of children with PCD. Without nodal cilia in the embryonic period thoracoabdominal orientation are random, these patients have katanager triad, defined as situs inversus totalis, chronic sinusitis and bronchiactesis. Other laterality defects such as heterotaxy may coexist with congenital heart defects, asplenia or polysplenia.

Eleven genes with disease-causing mutations have been identified. These include 5 genes coding for outer dynein arm proteins (*DNAI1*, *DNAI2*, *DNAH5*, *DNAH11*, *TXNDC3*), 2 genes for radial spoke proteins (*RSPH4A*, *RSPH9*), and 4 genes for cytoplasmic proteins involved in dynein arm assembly (*c14orf104/KTU*, *LRRC50*) and in RPGR and OFD1 [11]. Mutations in *DNAI1* and *DNAH5* have been detected in 38% of patients with primary ciliary dyskinesia [11].

PCD is diagnosed with physical findings, clinical presentation and Specific ciliary ultra-structural defects identified by transmission electron microscopy or 'TEM.' This "gold standard" diagnostic test for primary ciliary dyskinesia requires a biopsy of ciliated airway tissue usually taken by brush sample or scraping of either the nose or the trachea [12]. Positive Clinical genetic testing can be done for confirmation of PCD. In addition, measurement of nasal nitric oxide (NO), while not diagnostic, can be a very useful screening tool in PCD, which is characterized by unusually low levels of nasal NO.

CONCLUSIONS

One should always keep in mind the possibilities of Kartagener syndrome in those patients presenting with recurrent upper and lower respiratory tract infections, sinusitis or bronchiactesis. Failure to recognize the condition may subject the patient to unnecessary and repeated hospital admissions, investigations and inappropriate treatment.

The progression of lung disease varies and is affected by the time of diagnosis, the ability of medical treatment to control symptoms, and the prevention of complications that affect the quality of life. Regular surveillance should include lung function testing, microbiological studies, and review of airway clearing techniques. Currently, management is mainly focused on chest physiotherapy and trail of human recombinant DNase.

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