

Kartagener's Syndrome: A Rare Case Report

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Abstract: *Kartagener's syndrome, a rare genetic disorder, is a clinical challenge to be diagnosed on initial presentation. Multitude of problems associated with syndrome may hamper the quality of life of the affected person and family as a whole. To improve overall prognosis it necessitates to be diagnosed as early as possible. High suspicion should be kept in children presenting with recurrent chest and ENT symptoms so that early diagnosis and treatment is initiated. Genetic and fertility counselling should be offered to persons and family as and when necessary. Research to be taken to make easy, reliable and affordable testing and treatment for all.*

Keywords: Kartagener's syndrome, situs inversus, chronic sinusitis, primary ciliary dyskinesia

1. Introduction

Kartagener's syndrome is a subset of primary ciliary dyskinesia which are genetically inherited as an autosomal recessive inheritance. Typically manifest as a classical triad of chronic sinusitis, bronchiectasis and situs inversus [1,2]. It is a genetic mutation in DNAI1 and DNAH5 genes which lead to recurrent sinopulmonary infections, infertility and error in left-right body orientation [3].

2. Case Report

A 12 years old female child from lower socioeconomic strata presented to ENT OPD with history of recurrent nasal obstruction, chronic cough and chronic nasal discharge and parents gives history of being inattentive in school and some hearing problems. Pulse 90/min regular high volume, BP 100/60mmHg, clubbing grade 1 seen, no cyanosis. Child was thin built 15 kg in weight, pale, on examination bilateral vesicular breathing heard with occasional crepitations lower zones and apex beat heard on the right side on auscultation. On ENT examination, the bilateral nasal cavity full of thick yellowish discharge and bilateral tympanic membrane were intact but dull looking. Right sided otoscopy was suggestive of secretory otitis media. Pure tone audiometry was suggestive of conductive hearing loss 40 dB on right side and 33 dB and tympanometry suggestive of type B tympanogram right ear and type C in left ear. X Ray PNS and X Ray chest were advised.

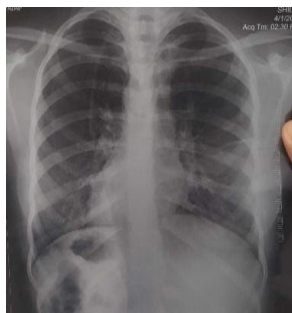


Figure 1: Chest radiograph showing Dextrocardia and fundal gas shadow on right side



Figure 2: X ray PNS Showing Bilateral maxillary sinusitis

It was found to have bilateral maxillary sinusitis on X Ray PNS and dextrocardia on chest X Ray along with fibrotic bands on both side lower lung fields, with fundal gas shadow on right side.

Diagnosis of Kartagener's syndrome was made on the basis of clinical presentation and radiographic evidence and clinical examination suggestive of ciliary dysfunction. Child was prescribed oral and nasal decongestants, a 14 days course of oral antibiotics, and mucolytic agents in syrup preparation. Routine blood investigations were prescribed and the child was advised paediatric consultation and pulmonary consultation. Grommet insertion was planned after 14 days of follow up but the child was lost during the follow up.

3. Discussion

Kartagener's syndrome is a subset of primary ciliary dyskinesia characterised by the classical triad of chronic sinusitis, bronchiectasis and situs inversus. Overall incidence of the disorder worldwide is approximately 1 in 30,000 live births [1, 2].

Impaired ciliary function due to defect in ultra-structure of ciliary elements encoded by mutation in genes DNAI1 and DNAH5, leads to defects right from embryogenesis, varying from situs solitus to situs inversus totalis [3, 4]

In countries like ours these disorders are underdiagnosed and lately diagnosed as many do not seek medical attention. Recurrent sinopulmonary infections, failure to thrive are noticed commonly in such patients. Infertility due to immotile cilia is seen associated with these disorders. As no

easy, reliable and non-invasive method of diagnosis is available it leads to further delay in making proper diagnosis and many patients are also lost during follow up as in our case. So one should focus on clinical diagnosis and supportive investigations and be vigilant with recurrent Sinopulmonary problem, recurrent otitis media in paediatric population.

Supportive treatment in the form of chest physiotherapy, mucolytic agents and long term low dose antibiotics may help those with frequent exacerbation of bronchiectasis. Immunization of children with influenza and pneumococcal vaccine should be done to minimize risk of recurrent respiratory tract infections.

Few of them may require interventions like tympanostomy tube insertion and functional endoscopic surgery in ENT setup. Collective efforts through a multidisciplinary approach may help in improving quality of life in such patients.

4. Conclusion

As there is no easy, reliable and non-invasive method to diagnose Kartagener's syndrome, it may remain undiagnosed for years after initial presentation [5]. High degree of suspicion should be kept in paediatric patients presenting with recurrent respiratory, recurrent otitis media, chronic sinusitis to ENT and recurrent pneumonias, failure to thrive, growth retardation to paediatric OPD. Early diagnosis will be beneficial for the patient to improve quality of life and improve life expectancy [5]. These patients require a multidisciplinary approach, regular follow up with a chest physician, medical specialist and ENT specialist. Genetic counselling should be offered to such patients and fertility issues should be addressed appropriately [6].

5. Conflict of interest

None declared

6. Financial Disclosure

The facilities for the study including laboratory are available in the institute. No financial charges will accrue to the study subject. The study is not funded by any agency.

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