International Journal of Otorhinolaryngology and Head and Neck Surgery
Supreety et al. Int J Otorhinolaryngol Head Neck Surg. 2023 Jan;9(1):107-109
http://www.ijorl.com
pISSN 2454-5929 | eISSN 2454-5937

DOI: https://dx.doi.org/10.18203/issn.2454-5929.ijohns20223388

Case Report

Kartagener's syndrome: a rare case report

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Received: 09 October 2022
Accepted: 10 December 2022

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ABSTRACT

Kartagener's syndrome is an autosomal recessive primary ciliary dyskinesia. It is due to ciliary dysfunction consisting of triad of situs inversus, bronchiectasis, chronic sinusitis. Normal ciliary function is critical for respiratory host defence, motility of sperm and ensures proper visceral orientation during embryogenesis. In Kartagener syndrome, the gene mutation is at DNAI1 and DNAH5 leading to pathophysiological changes like ciliary immobility causing accumulation of secretions and consequent recurrent sinusitis. The severity of symptoms and the age at which the condition is diagnosed is quite variable, even though the symptoms are present from birth. We are reporting a case of 12-year-old male who came to ear, nose and throat outpatient department (ENT OPD) with complaints of bilateral nasal obstruction for the past 6 years associated with bilateral mucopurulent nasal discharge, having pale, greyish white polyps filling bilateral nasal cavities. Clinical examination and imaging findings revealed ethmoidal polyposis with chronic sinusitis, bronchiectasis, and situs inversus totalis. Patient underwent revision endoscopic intranasal ethmoidectomy with functinal endoscopic sinus surgery.

Keywords: Kartagener's syndrome, Primary ciliary dyskinesia, Situs inversus, Bronchiectasis, Ethmoidal polyposis

INTRODUCTION

Kartagener's syndrome is a rare hereditary disorder. It was first described by Siewart in 1904. A Swiss paediatrician Kartagener recognized the clinical syndrome in 1933. Eliasson et al first coined the term "immotile cilia syndrome" for Kartagener's syndrome and associated infertility with chronic sinopulmonary infections.[1,2]

Genetic basis for the variety of defects affecting ciliary structure and function in PCD is not clear till date. Mutations in more than 30 different genes have been mentioned.[3] The disease occurs as a direct result of congenital defects in motile cilia covering the respiratory epithelia leading to impairment of the mucociliary clearance causing recurrent sinopulmonary infections. Its incidence is approximately 1 in 30,000 live births.

The clinical phenotype is broad and overlaps with other chronic airways diseases causing chronic upper and lower respiratory tract infections. The incidence and severity differ from one patient to another, even among siblings. The estimated prevalence of PCD is about 1 in 16,000 live births, but this could be an underestimation due to missed diagnosis. Around 50% of the patients with primary ciliary dyskinesia have situs inversus.

CASE REPORT

Presenting a case of 12-year-old male who came to ENT OPD with complaints of bilateral nasal obstruction (L>R) for the past 6 years associated with bilateral mucopurulent nasal discharge. On examination patient was having pale, greyish white polyps filling bilateral nasal cavities.

Bilateral nasal obstruction was aggravated on exposure to cold air and on consuming cold items and partly relieved on taking medication. Bilateral mucopurulent nasal discharge is continuous in nature, non-foul smelling, non-bloodstained, aggravated on taking cold items, partly relieved on taking medication. There is history of mouth

breathing, snoring, dust allergy and anosmia. There is no history of bilateral ear pain, ear discharge, throat pain, difficulty in swallowing, epistaxis, fever and headache.

There is history of similar complaints 7 years ago for which the patient underwent functional endoscopic sinus surgery. Patient has normal appetite, mixed diet, adequate sleep, no addictions/allergies, bladder and bowel habits are regular. There is no relevant family history.

On general examination patient is thin built and his vitals are stable. There is no evidence of pallor, icterus, cyanosis, clubbing, lymphadenopathy and pedal edema. On anterior rhinoscopy smooth, pale, greyish mass seen in both nostrils along with mucopurulent discharge, the polyps are not sensitive to touch and not bleeding on touch. On posterior rhinoscopy greyish polypoidal masses filling both the choana is seen. Olfaction is absent. Cold spatula test and cotton wool test both are decreased on right side. No palpable lymph nodes on clinical examination.

Clinical examination and imaging findings revealed chronic sinusitis, bronchiectasis, dextrocardia, and situs inversus totalis. Patient and relatives were counselled about the nature of the disease and after written and verbal informed consent, patient underwent revision endoscopic intranasal ethmoidectomy and functional endoscopic sinus surgery involving removal of polyps and restoration of drainage and ventilation of sinuses. Postoperative period was uneventful. Histopathology report was suggestive of features of inflammatory polyp.

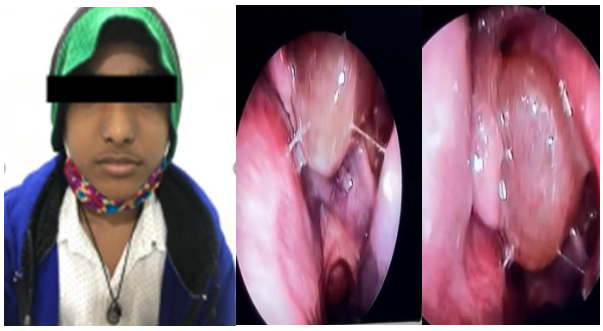

Figure 1: 12-year-old male having pale, smooth, glistening mass seen in bilateral nasal cavity.

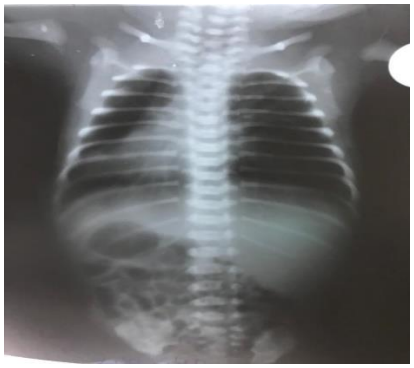

Figure 2: Dextrocardia.

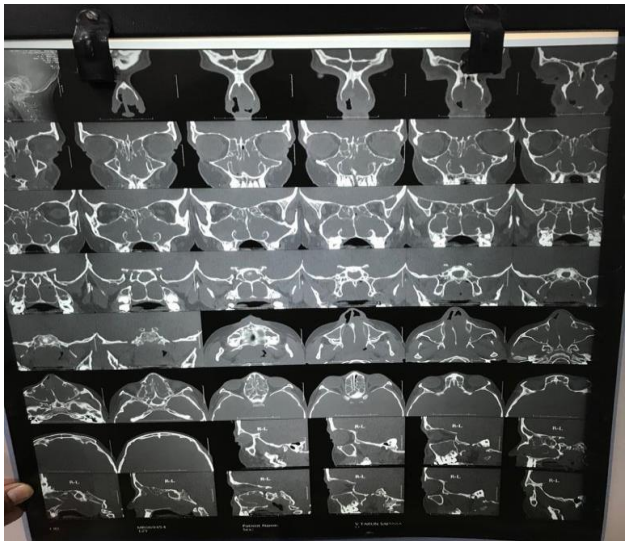

Figure 3: CT scan imaging involving all paranasal sinuses.

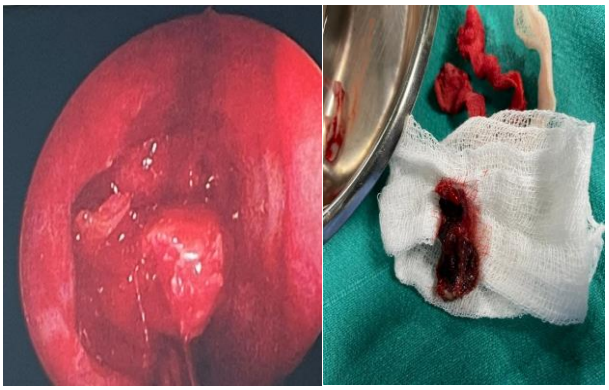

Figure 4: Intraop images of polyp.

DISCUSSION

Lack or dysfunction of dynein arms, radial spokes and microtubules of cilia are recognized structural and functional abnormalities of ciliary ultrastructures, encoded by the mutated genes DNAI1 and DNAH5. These faulty genes cause the cilia to be of the wrong size and shape or move in the wrong way, making ciliary motility defective.[4,5]

Pseudostratified ciliated columnar epithelium lines the nasopharynx, middle ear, paranasal sinuses, larynx, trachea and bronchi. Ciliary ultra-structures are coded by the gene DNAI1 and DNAH5. Mutation of these genes leads to disruption in the functioning of mucociliary clearance causing ineffective movement and leading to sinonasal, aural and pulmonary infections. Nasal involvement occurs usually in the form of chronic rhinitis, sinusitis and nasal polyposis where nasal obstruction and rhinorrhoea are the main complaints.[5]

Sinusitis in Kartagener syndrome is the least distinctive feature and can coexist with the absence or hypoplasia of

one or more sinuses, nasal polyposis or infection. Normal ciliary beating is also necessary for visceral rotation and orientation during embryonic development. Patients with KS may have either situs solitus i.e., dextrocardia only or situs inversus totalis where all the visceral structures are on opposite side.[6]

The diagnosis of Kartagener syndrome is often made incidentally on routine radiological examination which includes X-ray which is the first imaging modality that reveals dextrocardia and situs inversus as an incidental finding on routine check-up. High resolution computed tomography (HRCT) of chest shows bronchiectatic changes and ultrasonogram of abdomen will also reveal situs inversus totalis. CT scan of paranasal sinuses delineates pansinusitis, polyposis or hypoplasia of sinuses.

In our case clinical examination and imaging findings revealed chronic sinusitis with nasal polyposis, bronchiectasis, dextrocardia, and situs inversus totalis. Patient was managed medically for upper respiratory tract infection followed by endoscopic intranasal ethmoidectomy and functional endoscopic sinus surgery. Postoperatively patient was relieved of symptoms. He was asked to come for repeated follow ups.

CONCLUSION

Primary ciliary dyskinesia causes altered ciliary motility that causes multiple symptoms affecting the quality of life due to chronic upper and lower airway disease. Management includes systematic line of investigations and a multidisciplinary team for proper diagnosis and treatment. Initial line of treatment of nasal polyposis and chronic rhinosinusitis in these patients include ethmoidectomy with functional endoscopic sinus surgery and the importance of nasal irrigations and medical treatment to prevent recurrences. A wide inferior nasal antrostomy and the extensive middle meatal antrostomy allow proper nasal irrigations, thus promoting the mechanical drainage of the paranasal sinuses in these patients where there is improper mucociliary clearance. Genetic counselling should be addressed once Kartagener's syndrome is diagnosed.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

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Cite this article as: Supreety, Singh TD. Kartagener's syndrome – a rare case report. Int J Otorhinolaryngol Head Neck Surg 2023;9:107-9.