Review

Kartagener’s syndrome- A case report

Jayita Poduval* and Murali Poduval

1Department of Otolaryngology, Faculty of Medicine, AIMST University Semeling, Bedong-08100, Malaysia.
2Faculty of Medicine, AIMST University, Malaysia.

Accepted 10 February, 2011

Primary Ciliary Dyskinesia (PCD) is a genetic disorder with an autosomal recessive mode of inheritance. It is caused by a defect in the structure of cilia, due to which ciliary movement, and consequently, its function, are impaired. Sinusitis, nasal polyposis, and otitis media with effusion are commonly seen among patients presenting to the otolaryngologist, and PCD should be considered as an aetiological factor in these cases, especially in the refractory ones. A case presenting to an otolaryngologist is described with a review of salient literature.

Key words: Nasal polyp, primary ciliary dyskinesia, Kartagener’s syndrome.

INTRODUCTION

Kartagener’s syndrome (KS) is a primary ciliary dyskinesia (PCD) in which there is malrotation of internal organs (situs inversus) bronchiectasis, sinusitis and male infertility (Afzelius BA, 1976). All these abnormalities are due to an inherent defect in ciliary structure and function.

Sinusitis, nasal polyposis, and otitis media with effusion are common problems among patients presenting to the otolaryngologist, and it is important to keep PCD in mind as an aetiological factor in these cases, especially the refractory ones. Prompt detection and treatment of PCD can prevent its various complications (Bush et al., 2007). This report is a review of the diagnosis and management of KS from an otolaryngological perspective.

CASE REPORT

A 10-year old boy presented with the history of nasal obstruction and rhinorrhoea for 6 months. There was no history of allergy, atopy, bronchial asthma or upper and lower respiratory infections. There were no other ear, nose or throat complaints. On examination, he was found to have bilateral nasal polyposis and viscid mucopurulent nasal discharge. Diagnostic nasal endoscopy (DNE) revealed polyps in both the middle meati and polypoid changes of the middle turbinate mucosa on both sides.

There was no evidence of otitis media with effusion (OME) as confirmed by otoscopy and impedance audiometry. General examination was normal save for dextrocardia.

He was advised surgery under general anaesthesia, for which the routine blood and urine tests were done and a chest X-ray obtained, which revealed dextrocardia and situs inversus.

A screening CT of the paranasal sinuses (Figure 1) prior to surgery showed limited polyposis in the region of the infundibulum on both sides, without gross involvement of the sinuses. A screening CT of the thorax (Figure 2) did not show any changes of bronchiectasis (Barker, 2002).

Referral to the paediatrician, pulmonologist and cardiologist did not reveal any active lesion, and the patient was taken up for surgery. Functional endoscopic sinus surgery (FESS) was done and the polyps removed and sent for histopathology, which showed features of non-allergic nasal polyps with oedema and scant cellularity. No other diagnostic testing was done as there were no facilities for the same.

Post-operatively the patient was put on oral antibiotics, nasal douches and intranasal steroid spray, and follows up regularly for suction clearance of the nose. 3 months post-op the polyps have not recurred, but the patient continues to have troublesome viscid rhinorrhoea, and has just started to develop mild respiratory symptoms. Physiotherapy and chest consultation have been instituted, and all the treatment has been provided free of
cost so far. Efforts are on to dispense antibiotics as and when necessary, and immunization as well, at a nominal or no cost to the patient, as he hails from a poor background.

**DISCUSSION**

PCD is a rare genetic disorder with an autosomal recessive mode of inheritance (Bush et al., 1998). It is caused by a defect in the dynein arm structure of cilia, due to which ciliary movement, and consequently its function, are impaired. Ciliary movements are responsible for the rotation and orientation of internal organs in the 10th to 15th days of gestation. In PCD the underlying ciliary dysfunction causes incomplete rotation or malrotation of one or many internal organs, most commonly the heart (Afzelius, 1976). Isolated malrotation of the heart (situs solitus) is associated with severe anomalies of the vessels connecting the heart and is thus
relatively rare as such cases have a very low survival. More commonly, a right-sided heart (dextrocardia) exists along with malrotation of the other internal organs, namely: lungs, liver, spleen, kidneys and intestines (situs inversus).

About 50% of PCD patients develop situs inversus and KS, which has been classically described as a triad of dextrocardia, sinusitis and bronchiectasis, and male infertility; the incidence of KS is estimated to be around 1:15000 (Bush et al., 1998) with variable penetrance, and phenotypic differences have been observed because the underlying genetic mutation has a pleiotropic effect (Holmes, 1964). Researchers have found possible explanations for the inflammatory changes in PCD, from Ig A deficiencies (Holmes, 1964) to low nasal nitric oxide (NO) levels and abnormal mucus properties though the exact mechanism remains poorly understood (Bush et al., 2006).

Clinically, patients present to the otolaryngologist with nasal obstruction, rhinorrhoea and deafness if symptoms pertaining to the other systems have not yet manifested. Nasal polyps, by and large, are a result of allergy (Marsden, 1978), and quite common and severe in cystic fibrosis (CF) but are not often seen in PCD; tonsillar hypertrophy and obstructive sleep apnoea are also rare in these patients; OME (Otitis media with effusion) is the most common otolaryngological problem in PCD but may stabilise by adolescence (Bush et al., 2007). In our patient, however, nasal polyposis was present and OME was absent. Mainly the finding of dextrocardia on general examination and X-ray pointed to a possible diagnosis of KS.

Radiology, in the form of a chest X-ray, quickly corroborates the clinical suspicion of dextrocardia, but may also reveal dextrocardia and situs inversus as an incidental finding on routine pre-operative workup. CT thorax may further delineate malrotation, and bronchiectasis if any, and other changes found in PCD (Barker, 2002). In the event of the CT scan being inconclusive, a Gallium-67 can establish the bronchiectatic changes (Becker, 2000).

Proper diagnostic tools are required for the definitive diagnosis of PCD. Screening tests like saccharin clearance test are simple and cheap (Canciani et al., 1988), but may be cumbersome in children, time-consuming, and most importantly, do not differentiate between primary and secondary ciliary dysfunction.

Nasal nitric oxide measurement is a rapid and reliable method for diagnosing PCD (Wodehouse et al., 2003) and is superior to measuring the exhaled NO (Narang et al., 2002) but is not widely available in all centres and all countries. Spirometric assessments are usually added to NO measurements for corroborative or research purposes (Bush et al., 2007).

Recent advances in genetic research have confirmed mutations of DNA H 5 and DNA 11 (Geremek et al., 2004). Genetic testing is useful for counselling and has the potential for developing gene therapy for the management of PCD in the future. Primary epithelial cell culture can be done both for gene testing and electron microscopy (EM) (Jorissen et al., 2000) in cases where the tissue and brush biopsy specimens are inadequate.

Transmission electron microscopy remains the most definitive method of establishing the diagnosis of PCD as the exact structural changes can be visualised. Ciliary beat frequency (Afzelius, 2004), ciliary beat pattern analysis by high-speed video photography (Stannard et al., 2004), EM of ciliary ultrastructure (Teknos et al., 1997) and measurement of ciliary disorientation (Stannard et al., 2004) are recommended wherever facilities exist.

At present, gene testing and EM are out of reach for many patients of PCD. Treatment may have to be instituted at the earliest based on clinical evaluation alone as the prognosis remains good in contrast to CF, where there is progressive deterioration, and the majority may enjoy a normal or near-normal life (Bush et al., 2007). At least in the case of Kartagener’s syndrome, a clinical diagnosis may be all that is needed if diagnostic testing is not available or feasible.

Medical management, usually multidisciplinary, consists of effective control of infections with antibiotics. Immunization is also advised, as is antibiotic prophylaxis. PCD patients are adequately stabilised with medical therapy alone (Bush et al., 2007).

Surgical treatment in the form of FESS has proved to be beneficial in patients with polyposis and sinusitis (Parsons et al., 1993) even though the number of patients studied was small and the follow-up period was less than 3 years. In fact, the treatment of PCD is not strictly evidence-based.

Special consideration is required at the time of administering anaesthesia; care must be taken to avoid nasal tubes and airways, and to observe strict aseptic precautions (Anthony, 1992).

The role of physiotherapy in PCD has not been fully established (Bush et al., 2007). Physical exercise and breathing techniques are recommended. Positive expiratory pressure devices (Volsko et al., 2003) are helpful and regular follow-up is strongly advised (Mcmanus et al., 2003).

Bronchiectasis, pneumonia and other pulmonary complications like collapse and fibrosis are preventable to a large extent; surgical interventions like lobectomy and pneumonectomy may not be required at all (Bush et al., 2007).

This case study was greatly limited by the inability to perform diagnostic testing. However, it focuses on the issue from the otolaryngologist’s perspective. It is a single case study and would have benefited if more cases were added to study the spectrum of presentation and behaviour of otolaryngological problems in this population. It is imperative to employ diagnostic testing wherever possible in order to establish the diagnosis of PCD for optimal management, record-keeping and
research purposes.

CONCLUSION

In the routine practice of a primary care physician or general otolaryngologist, the management of rare conditions like PCD, especially KS, may prove challenging if a high index of suspicion is not maintained. In the absence or scarcity of diagnostic facilities, thorough clinical evaluation and a multidisciplinary approach go a long way in reducing morbidity and enhancing the quality of life of these patients.

ACKNOWLEDGEMENTS

This case was managed at the Pondicherry Institute of Medical Sciences, India, where the author was Assistant Professor and Consultant in Otolaryngology. The author acknowledges the guidance of the then Head of Department Dr Thambi A Cherian.

REFERENCES