

CASE REPORT

KARTAGENER'S SYNDROME: A CASE SERIES STUDY

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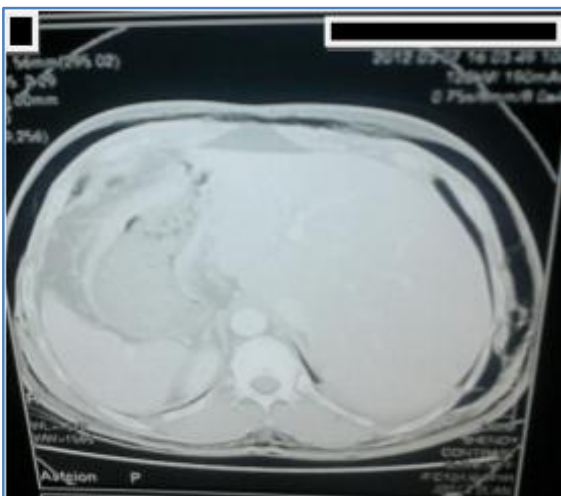
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ABSTRACT: Kartagener's Syndrome is a rare Autosomal recessive disorder presents as a triad of Sinusitis, Bronchiectasis and Situs inversus. Chronic Rhino sinusitis is a common condition seen is ENT OPD routinely. Of the several causes of Sinusitis, Kartagener's is rare entity and can remain undetected. This case series is to emphasize the diagnosis of the disease and alert the patient about it.

KEYWORDS: Kartagener's Syndrome, Bronchiectasis, Situs inversus, Dextrocardia, Primary ciliary dysfunction.

CASE 1: A 30 year old male patient presented to ENT OPD with complains of Nasal obstruction and nasal discharge on and off since 3 months. Nasal obstruction it was bilateral insidious in onset and gradually progressive, aggravated during winter months and subsided on taking medications. Nasal discharge was copious, thick and persistent, no h/o bleeding from the nose. Patient had hyposmia which was bilateral, associated with post nasal drip. Patient had history of headache which was bilateral and of mild degree which was more in morning. Patient is married and is having two children. His brothers are unmarried. No other significant past history was present On Anterior rhinoscopy there was septal deviation to right side with compensatory hypertrophy of the inferior turbinates was seen. Bilateral pale pink, soft, glistening, multiple, painless mass in both the nasal cavities was seen. Mass was insensitive to touch and doesn't bleed on probing. Probe can be passed all over except laterally. Diagnostic Nasal endoscopy confirms Anterior Rhinoscopy findings. Systemic examination revealed grade III clubbing further examination by general physician Dextro cardia, situs inversus totalis with spleen on the right side and liver on the left side were seen.

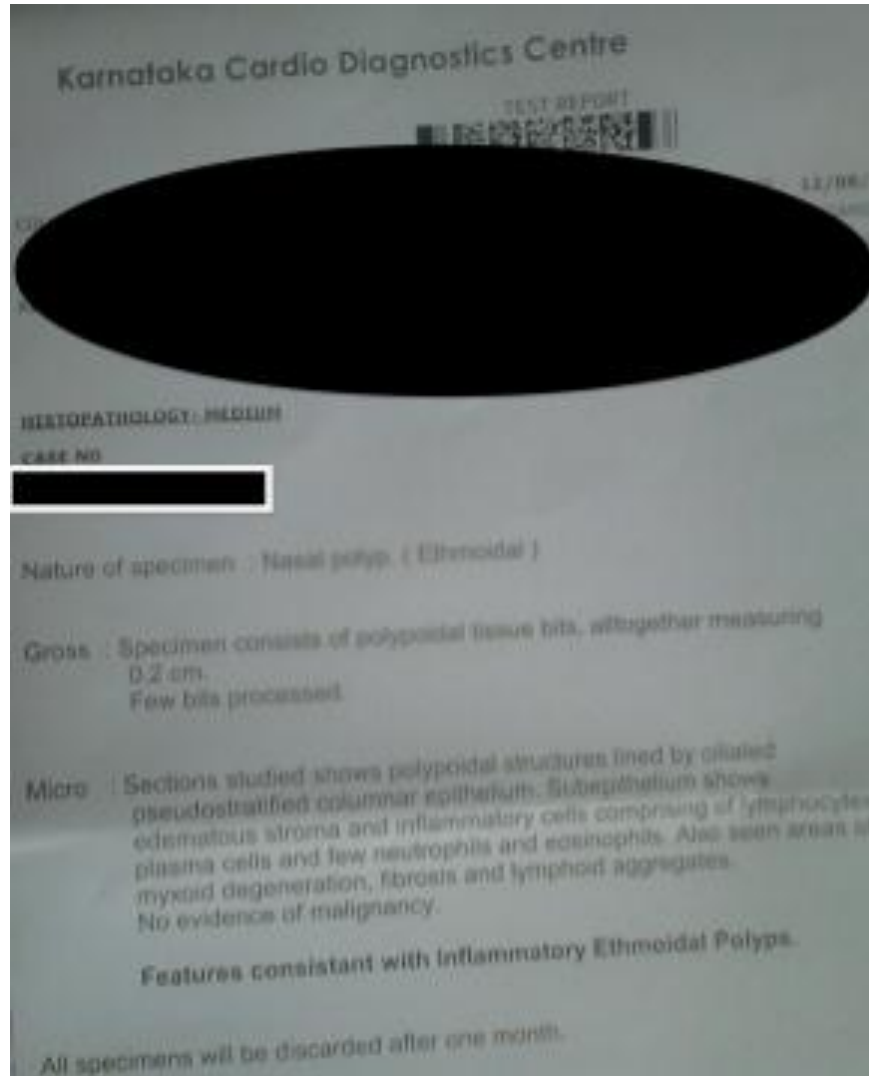


Situs inversus



Dextrocardia

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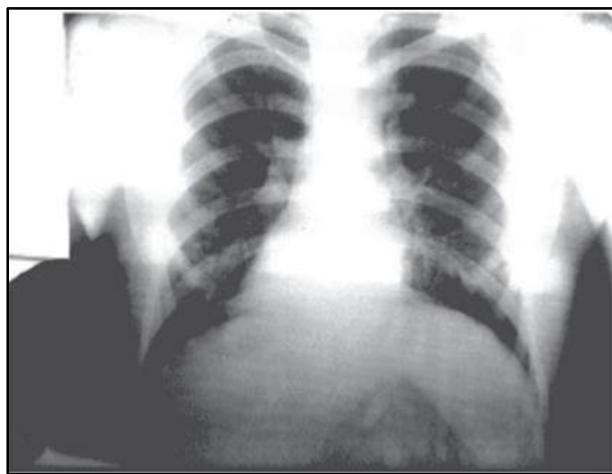
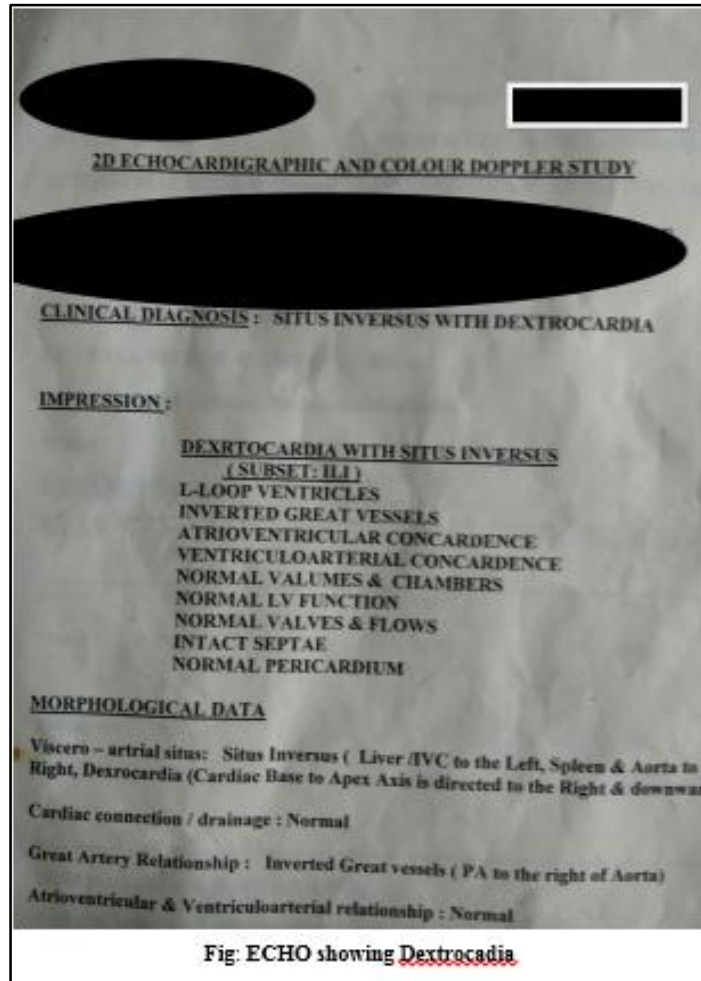


Routine blood investigation was done. X-ray of the nose and PNS showed opacification of the maxillary, frontal and ethmoid sinuses. CT of nose and PNS confirmed the X-ray findings, no bony irregularities and erosions were seen. X-ray of chest shows dextrocardia and bronchiectatic changes in both main bronchi. Ultrasound abdomen shows situs inversus totalis. HRCT thorax showed Dextrocardia with abdominal situs inversus involving left sided liver and right sided stomach and spleen. Treatment includes medical line of treatment and surgical line. Antibiotics, analgesics and decongestants were given. Functional endoscopic sinus surgery was done. Patient was referred to thoracic surgeon, paediatrician, and pulmonologist for further management.

CASE 2: A Case of 8yrs old male patient with chief complaints of common cold, sneezing and cough with expectoration and throat pain for past 2yrs. On clinical examination by Paediatrician dextrocardia was suspected. On examination, bilateral wheeze was present. X-ray chest revealed dextrocardia and chronic bronchitis. X-ray paranasal sinuses showed pansinusitis and x-ray of the neck showed enlarged adenoid. USG and CT of abdomen showed situs inversus where liver was on

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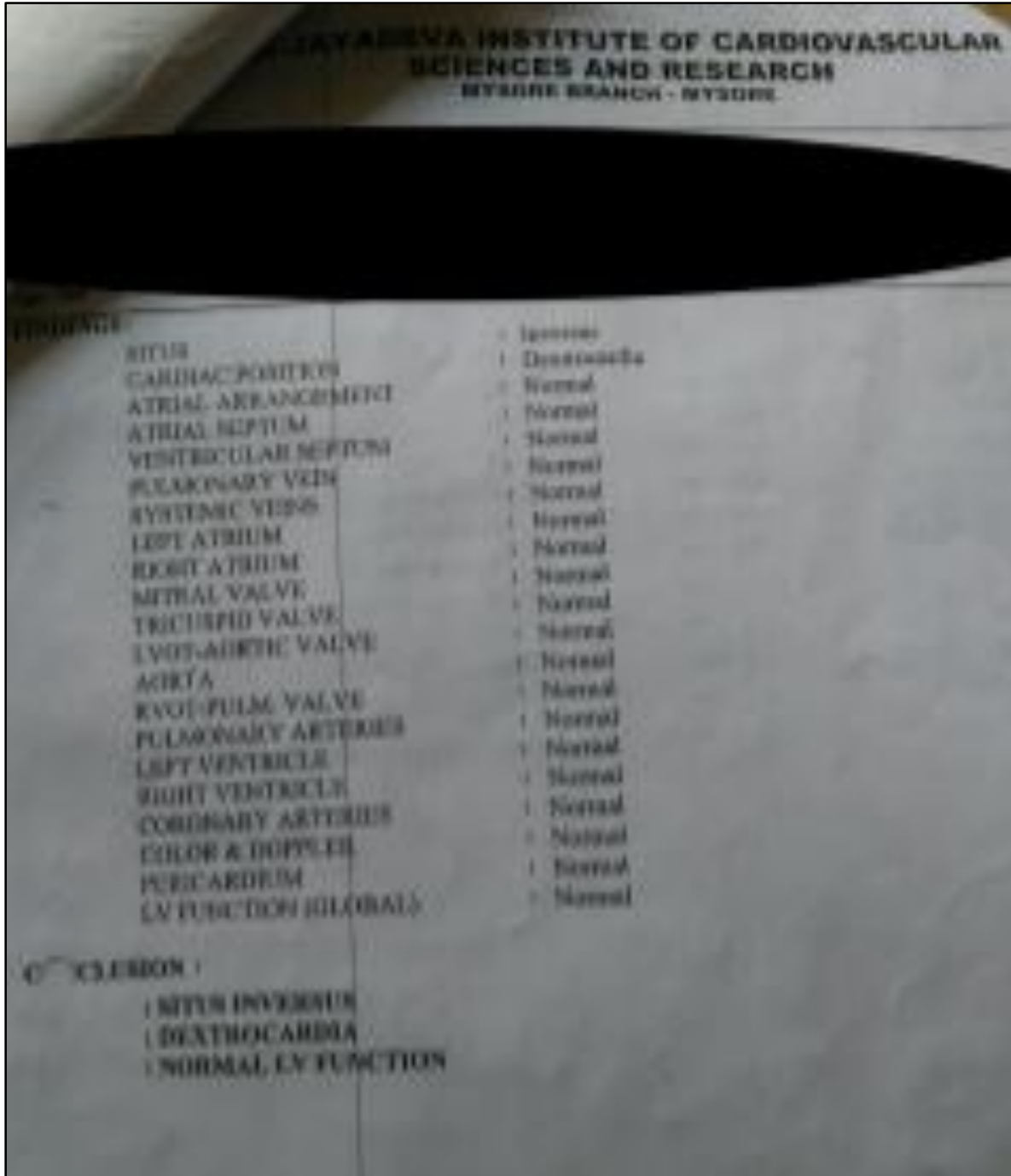
left and spleen on right. Treatment included Antibiotics, analgesics, steroids. Surgical treatment of adenotonsillectomy was done under general anaesthesia. Patient's parents were explained about the diseases and reassurance was given. Follow up is being done.



Chest x-ray (PA view)

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CASE 3: Case of 10yrs old female child with history of episodes of rhinorrhea, cough with expectoration, and throat pain and nasal obstruction. Clinical examination shows apex beat in right side suggested dextrocardia. X-ray chest shows dextrocardia and USG abdomen shows situs inversus. The diagnosis of kartagener's syndrome was made on the basis of Bronchiectasis, chronic sinusitis, inversus totalis. Management consists of medical line of treatment and physiotherapy. Surgical treatment was adenotonsillectomy under general anaesthesia. Patient is being followed up.



ECHO showing Dextrocardia

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X-ray showing Dextrocardia and bronchiectasis

ETIO PATHOGENEIS AND DISCUSSION: Kartagener's syndrome is a collection of bronchiectasis, situs inversus and dextrocardia.¹ Disorders of ciliary motility may be congenitally acquired. Congenital disorders are labeled as PCDs. Nearly 50% of PCD patients has situs inversus. Such cases of PCD with situs inversus are known as Kartagener's syndrome. PCD is a phenotypically and genetically heterogeneous condition where in the primary defect is in the ultrastructure or approximately 90% of PCD patients and involve the outer dynein arms, inner dynein arms, or both. 38% of the PCD patients carry mutations of the dynein genes DNAI and DNAH5. Pathophysiologically, the underlying defect which leads to accumulation of secretions and consequent recurrent sinusitis, bronchiectasis, infertility, and situs inversus is the defective ciliary motility/immobility. The severity of symptoms and the age at which the condition is diagnosed is quite variable, even though the symptoms are present from birth. Occasionally, Kartagener's syndrome may be associated with reversible airflow obstruction.²

Clinical progression of the disease is variable with lung transplantation required in severe cases. Diagnostic criteria for this condition include clinical picture suggestive of recurrent chest infections, bronchitis, and rhinitis since childhood, along with one or more of the following: (1) situs inversus in the patient/sibling; (2) alive but immotile spermatozoa; (3) reduced or absent transbronchial mucociliary clearance; and (4) cilia showing characteristic ultrastructural defect on electron microscopy.

Apart from fulfilling the criteria mentioned above, two types of tests are done for diagnosis of PCD—screening tests (Exhaled nasal nitric oxide measurement which is usually low in PCD, and saccharin test to assess mucociliary function of nasal epithelium) and diagnostic tests (Ciliary beat pattern and frequency analysis using video recording, and electron microscopic confirmation of the ultrastructural ciliary defect).² The samples for these tests for examining motility and ultrastructure of cilia may be obtained by biopsy of nasal mucosa and laparoscopic biopsies of tubal mucosa in females.¹ In our cases, however, we could not perform these tests and the diagnosis was essentially clinico-radiological.²

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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) keratosis obturans may be 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.³

Kartagener's Syndrome is a rare Autosomal recessive disorder presents as a triad of Sinusitis, Bronchiectasis and Situs inversus.⁴ Although the immotile cilia syndrome is a heterogeneous condition with regard to ultrastructure, and hence, the clinical profile seems to be fairly or even remarkably uniform perhaps not surprising when one considers the absence of mucociliary clearance as a common denominator. Characteristically, the respiratory tract disease can be traced back to early childhood or even infancy-often to the very day of birth. Neonatal respiratory distress is not uncommon. Chronic cough and expectoration of mucoid, mucopurulent, or at times purulent sputum is generally present and often tends to increase during the day rather than being most prominent in the morning as in smoker's chronic bronchitis. Atelectasis and pneumonia are fairly common.⁴

It has already been remarked that nearly all men with the immotile cilia syndrome are sterile ('Subfertile' Bartoloni et al. Axonemal beta heavy chain dynein DNAH9: cDNA sequence, genomic structure, and investigation of its role in primary ciliary dyskinesia. *Genomics*. 2001 Feb 15; 72(1): 21-33) because of immotility or poor motility of their spermatozoa but that some men have motile spermatozoa and are fertile. In typical cases, the volume of the ejaculate and the values for sperm number are within the normal range, and sperm in Males and ovum in females are equally affected. Most males with the syndrome have immotile spermatozoa and are sterile. Male sterility also can be observed in most, but not all, published Pedigrees. In rare cases, a Kartagener patient may have normally motile spermatozoa and be capable of fathering a healthy child. In a Norwegian population, major percent of all children with characteristic morphological features, are evaluated for therapy.

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 I 1 (Geremek et al., 2004). 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

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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), Electron microscope 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 Cystic Fibrosis, where there is progressive deterioration, and the majority may enjoy a normal or near-normal life (Bush et al., 2007). Organisms which may present in Kartagener's Syndrome are Haemophilus influenza, Staphylococcus aureus and pseudomonas. 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 stabilized 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. The role of physiotherapy in PCD has not been fully established. Physical exercise and breathing techniques are recommended. Positive expiratory pressure devices are helpful and regular follow-up is strongly advised.⁵ 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). It is imperative to employ diagnostic testing wherever possible in order to establish the diagnosis of PCD for optimal management, record-keeping and Genetic testing is useful for counselling.⁶ It has also been recognized in User's syndrome type-1, congenital heart diseases, congenital including folliculitis, nummular eczema, and pyoderma gangrenosum.⁷

Treatment is symptomatic and directed against complications in the upper and lower respiratory tract. There is no method available to restore ciliary and spermatozoal motility in situ. Antibiotics or chemotherapeutic agents may be given when there are signs of bacterial infection, such as increased purulence of sputum or bouts of sinusitis or otitis. In selected patients, intravenous administration of antibiotics may be necessary as well as extended courses of 1 to 3 months of oral antibiotics. The value of mucolytics is uncertain, but they may be tried in selected patients with tenacious secretions. Bronchodilators (Beta-adrenergics, methylxanthines, or anticholinergics) may be valuable in patients in whom there is airway obstruction with a bronchospasm.⁴

Physiotherapy is often important and, if started early in life, may prevent or delay the evolution of bronchiectasis and atelectasis. Abandonment of smoking is a most important preventive measure, since smoking probably accelerates deterioration of lung function. Surgical interventions against maxillary sinusitis, nasal polyposis, and middle ear disease are often performed repeatedly in these patients.⁵ (e. g., endonasal trepanation, Caldwell-Luc operation, polypectomy, tympanostomy). Such operations doubtless may be necessary in certain patients, but a certain amount of conservatism has been advocated in this context, since there often is a spontaneous recession in adulthood.

Thoracic surgical intervention against bronchieclasis is sometimes indicated. Although the choice may be difficult in individual patients. The symptoms of chronic bronchitis are not be expected

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to be cured by resenotional surgrey. A heart-lung transplantation has been performed in two patient suffering from the complete Kartagener Syndrome.⁴

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.

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