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Inherited degenerative chondropathy – an autosomal dominant new clinical entity: report two cases and follow-up of four cases

MARY KURIEN*, M. S. SESHADRI,†, ANAND ZACHARIAH†

Abstract

Four cases of the rare disorder, inherited degenerative chondropathy have been previously reported (Kurien *et al.*, 1989). A five-year follow-up of these patients and two additional cases are presented in this report. The progress of this disease appears to be arrested after regular dapsone therapy and there was no other organ involvement noted during the follow-up period.

Key words: Cartilage, abnormalities; Nose; Larynx; Spine; Hearing loss, sensorineural; autosomal dominant

Introduction

Inherited degenerative chondropathy has been described previously in four of our patients (Kurien *et al.*, 1989). Here we report on two additional patients, who are also siblings. They presented with a saddle-nose deformity, present since birth, and delayed onset of laryngeal stenosis with premature calcification of the laryngeal cartilages but absence of the other features of relapsing polychondritis (McAdam *et al.*, 1976; Moloney, 1978; Bauby *et al.*, 1991). However these two patients have, in addition, sensorineural hearing loss and involvement of tracheal cartilages with loss of lumbar lordosis and thoracic kyphosis causing straight back syndrome. This type of degenerative disease of the cartilages has not been described in any of the known connective tissue disorders, inherited or acquired (McKusick, 1966).

Report of cases

The clinical features and the results of the laboratory investigations are given in Table I. These patients were siblings and their family tree is presented in Figure 1. Both were noted to have an identical nasal deformity present since birth (Figure 2). Figure 3 (a and b) illustrates the calcification of the laryngeal and tracheal cartilages.

Both these patients came to our hospital primarily for augmentation rhinoplasty. As the first patient had clinical and radiological evidence of moderate glottic and subglottic stenosis, rhinoplasty was deferred due to the possibility of intubation difficulty and of a tracheostomy. However in the second patient, although there was evidence of only mild subglottic stenosis, endotracheal intubation for augmentation rhinoplasty was unsuccessful and emergency tracheostomy had to be done. Both these patients have been started on dapsone (Gouet *et al.*, 1984).

Discussion

Inherited degenerative chondropathy involving laryngeal and nasal cartilages has been described previously (Kurien *et al.*, 1989). These two patients also have inherited degenerative chondropathy. It differs from relapsing polychondritis in that these patients are younger, there is a positive family history, clinical signs of episodic inflammation of the involved cartilages are absent but X-ray evidence of premature calcification of the laryngeal and tracheal cartilages is present. Relapsing polychondritis, in contrast, is most prevalent in the fourth decade and is characterized by recurrent episodes of inflammatory seronegative polyarthritis with auricular, nasal and laryngeal chondritis. Occasional ocular, cardiovascular and renal involvement are reported. Laryngeal and tracheal manifestations in relapsing polychondritis are most often end-stage complications (McAdam *et al.*, 1976; Cyril and Joseph, 1993).

In addition to the saddle-nose deformity and laryngeal stenosis with cartilage calcification, these two patients were found to be of short stature, had associated tracheal stenosis, loss of thoracic kyphosis and lumbar lordosis

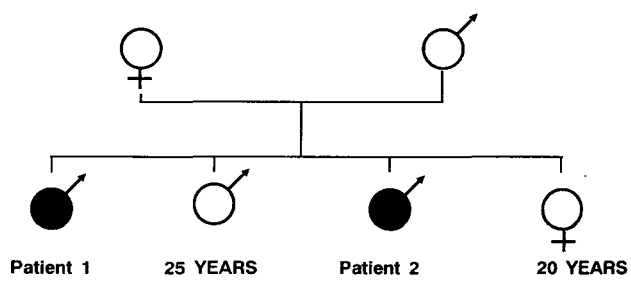

FIG. 1

Family tree for the patients showing two of the four siblings were affected. No first degree relative of either parent had a similar illness.

From the Departments of Otolaryngology*, and General Medicine†, Christian Medical College Hospital, Vellore, South India.
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TABLE I

	Patient 1	Patient 2
Age of presentation	27 years	21 years
Clinical presentation:		
Saddle-nose deformity	Since birth	Since birth
Hearing loss	For 15 years	For 18 years
Change in voice	For 10 years	Not present
Breathing difficulty	For 5 years	Not present
Larynx		
Epiglottis: infantile	Present	Present
Vestibular folds: oedematous	Present	Not present
True folds: mild reduction in the anteroposterior diameter and reduction in abduction	Present	Not present
Immediate subglottic stenosis	Moderate	Mild
Ear: pinna, canal and tympanic membrane	Normal	Normal
Systemic examination (cardiovascular, renal, ocular, articular)	Normal	Normal
Complete blood counts/ESR/blood sugar/serum creatinine/ calcium and phosphorus	Normal	Normal
VDRL	Nonreactive	Nonreactive
24 hour urine:		
Protein	Normal	Normal
Mucopolysaccharide	Absent	Absent
2D Echo cardiogram	Normal	Normal
X-rays:		
Chest (PA)	Normal	Normal
Spine (lateral):		
Loss of lumbar lordosis and thoracic kyphosis	Present	Present
Neck (lateral):	(Figure 3a)	(Figure 3b)
Calcification of the epiglottis, thyroid cricoid and upper tracheal rings	Present	Present
Audiogram:		
Hearing loss (sensorineural)	Moderate	Severe
Indication for tracheostomy	Not done	Failed endotracheal intubation
History of fever, arthralgia, painful swelling of nasal/ear/laryngeal cartilages	Absent	Absent

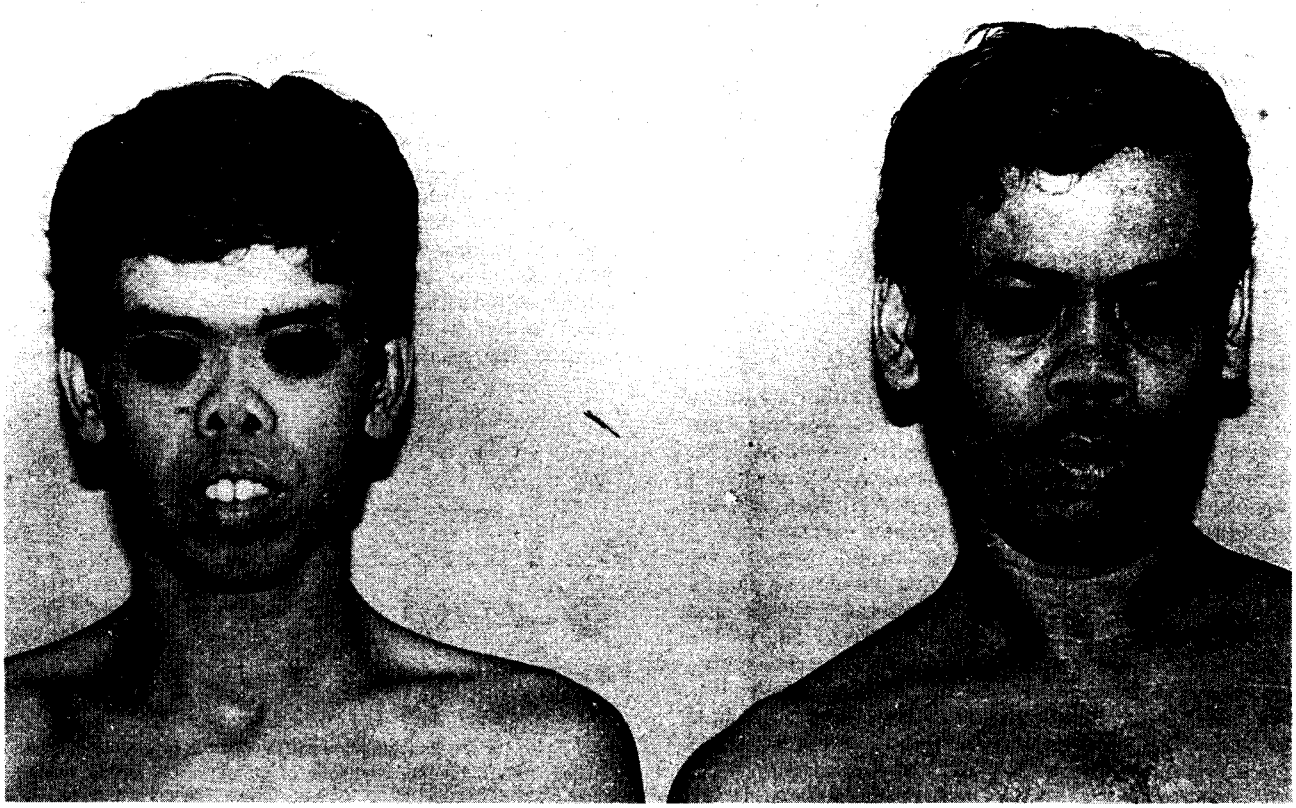

FIG. 2

Photograph of the two patients: patient 1 (right) and patient 2 (left).

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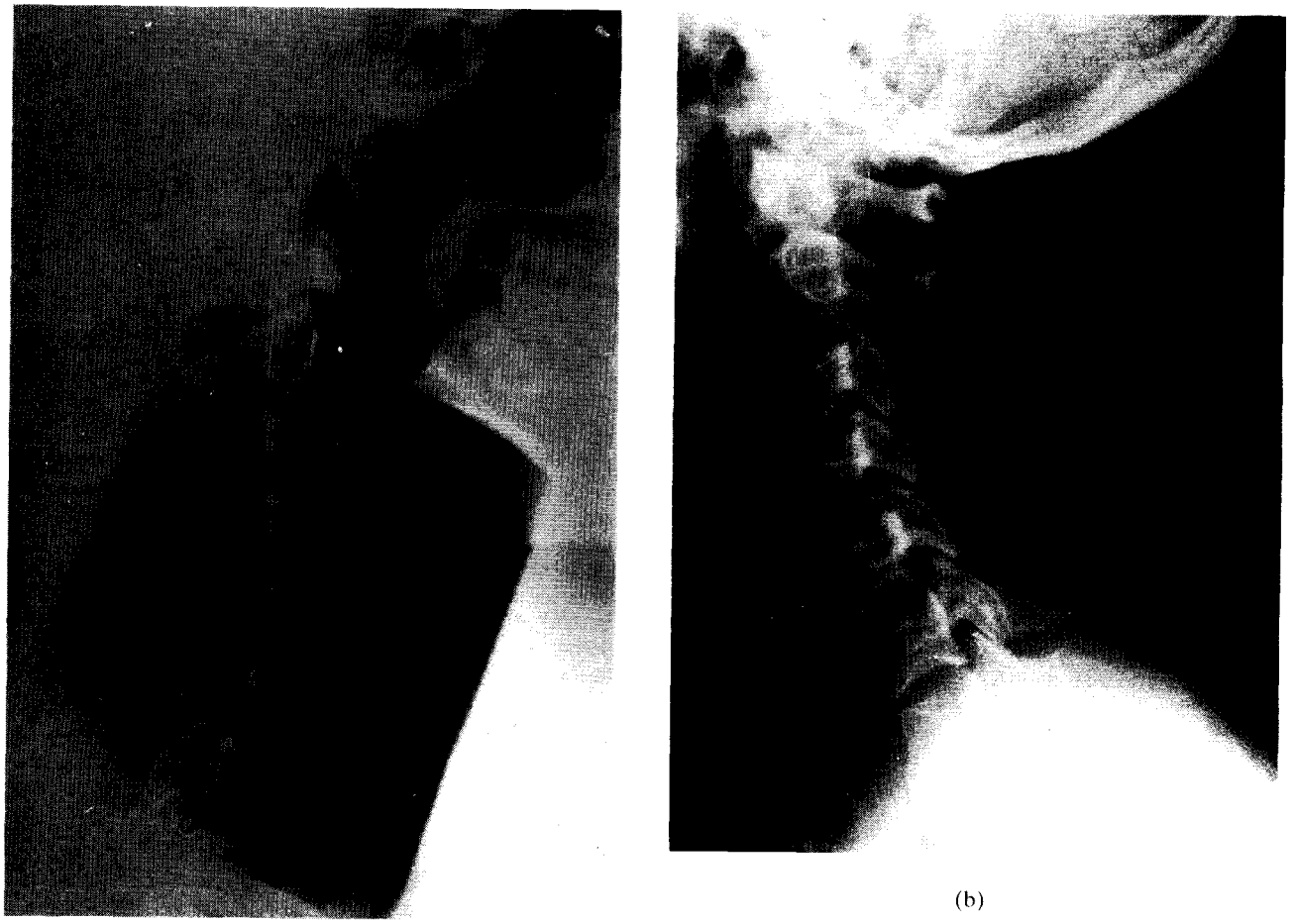

(a) (b)

FIG. 3

X-ray of the lateral neck: (a) patient 1; and (b) patient 2. Note the extensive calcification of the epiglottis, thyroid, cricoid and tracheal cartilages.

causing straight back syndrome and a sensorineural hearing loss. In our earlier report (Kurien *et al.*, 1989) the patients had only the first two features suggesting the involvement of the hyaline cartilages. This may be because all of them presented at less than 16 years old and involvement of elastic cartilages may be a delayed feature. The mode of inheritance in this report appears to be autosomal dominant as the affected brothers have two other siblings who are normal (Figure 1). This was also the pattern observed in our previous report (Kurien *et al.*, 1989).

A five-year follow-up of the four patients in our previous report who are on dapsone therapy (Gouet *et al.*, 1984) shows no evidence of progression of the disease and no other additional features have been detected. A follow-up of both the brothers in this report after two years of dapsone therapy (Kurien *et al.*, 1989) has shown no worsening of stridor. Tracheostomy is being avoided in the elder brother. Both cases have not developed any other new symptoms.

All the six patients are being continued on dapsone therapy.

Conclusion

Inherited degenerative chondropathy is an autosomal dominant inherited disorder of cartilage (hyaline cartilage is affected earlier than the elastic cartilage) with secondary dystrophic calcification. Classical presentation is of an adolescent or young adult with a saddle-nose deformity present since birth and delayed onset of laryngeal and tracheal stenosis due to premature calcification of those cartilages. There is no history of episodic fever with athralgia and painful swelling of the involved cartilages. The laryngeal stenosis may necessitate tracheostomy eventually or earlier if elective endotracheal intubation is attempted. There can be associated somatic abnormalities like short stature, loss of thoracic kyphosis and lumbar lordosis, and sensorineural hearing loss. Dapsone appears to prevent the progression of this disorder.

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Address for correspondence:
Dr Mary Kurien, M.B.B.S., D.L.O., M.S.,
Associate Professor,
Department of ENT,
Christian Medical College Hospital, Vellore,
Tamil Nadu 632004, South India.

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