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Otological manifestations of a new familial polyostotic bone disorder

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Abstract
Fifty members of a family with a unique autosomal dominant bone disease were investigated. Nineteen of the family members were either known to have, or were strongly suspected of having the disease. All but one of these had a hearing loss which was conductive in the younger age group and mixed in the older members. The common finding in those who had middle ear surgery was replacement of the long process of incus by a fibrous band. The histological features were similar to those found in Paget's disease. The age of onset, distribution of lesions and radiographic findings, however, were not typical of this disorder.

Introduction

The first member of this family presented to the Orthopaedic Department in 1939 with a swelling of the forearm and a pathological fracture of the left radius. Initial investigations suggested that this was an atypical form of either osteitis fibrosa cystica (hyperparathyroidism) or fibrous dysplasia of bone. When other similarly affected members of the family presented in the early 1950s it became obvious that this was a familial disease with an autosomal dominant pattern (Fig. 1).

Radiographic features

The bony abnormality was seen most commonly in the limbs and only occasionally in the girdles and axial skeleton. This was unlike the distribution of bony lesions in Paget's disease. The characteristic findings in this family were areas of bone expansion progressing to translucent lesions. The rate of progression of these was much faster than that found in Paget's disease. Unfortunately, we have been unable to get radiographic information about the temporal bones in these patients. Radioistope scans

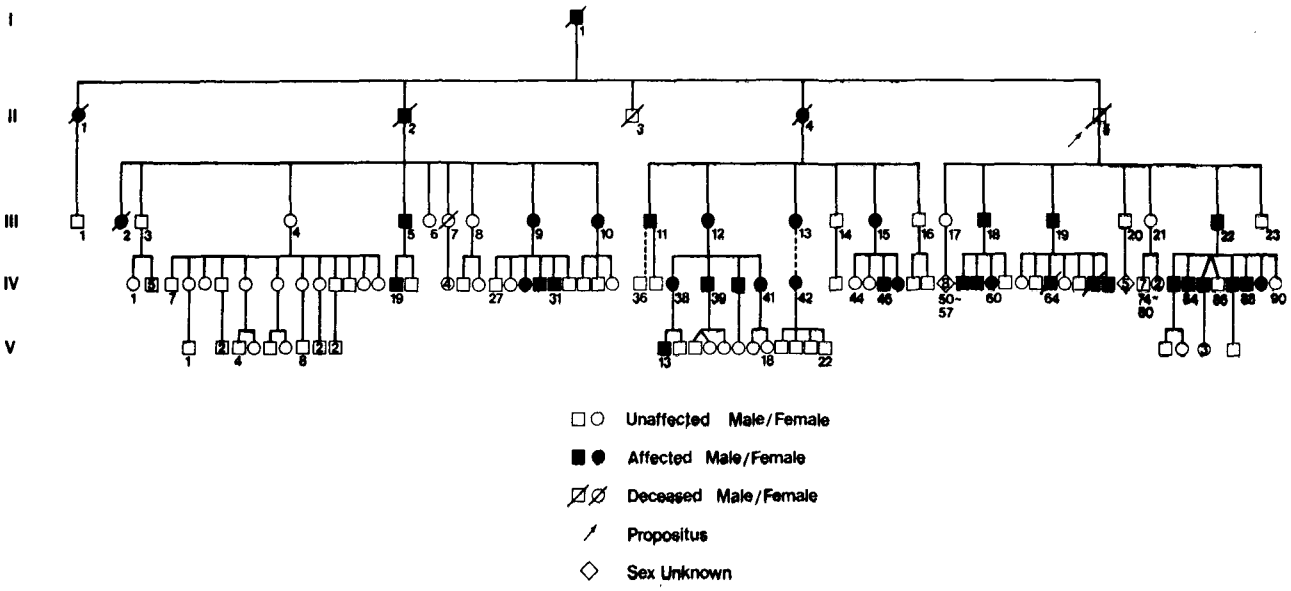

FIG. 1
Family Tree.

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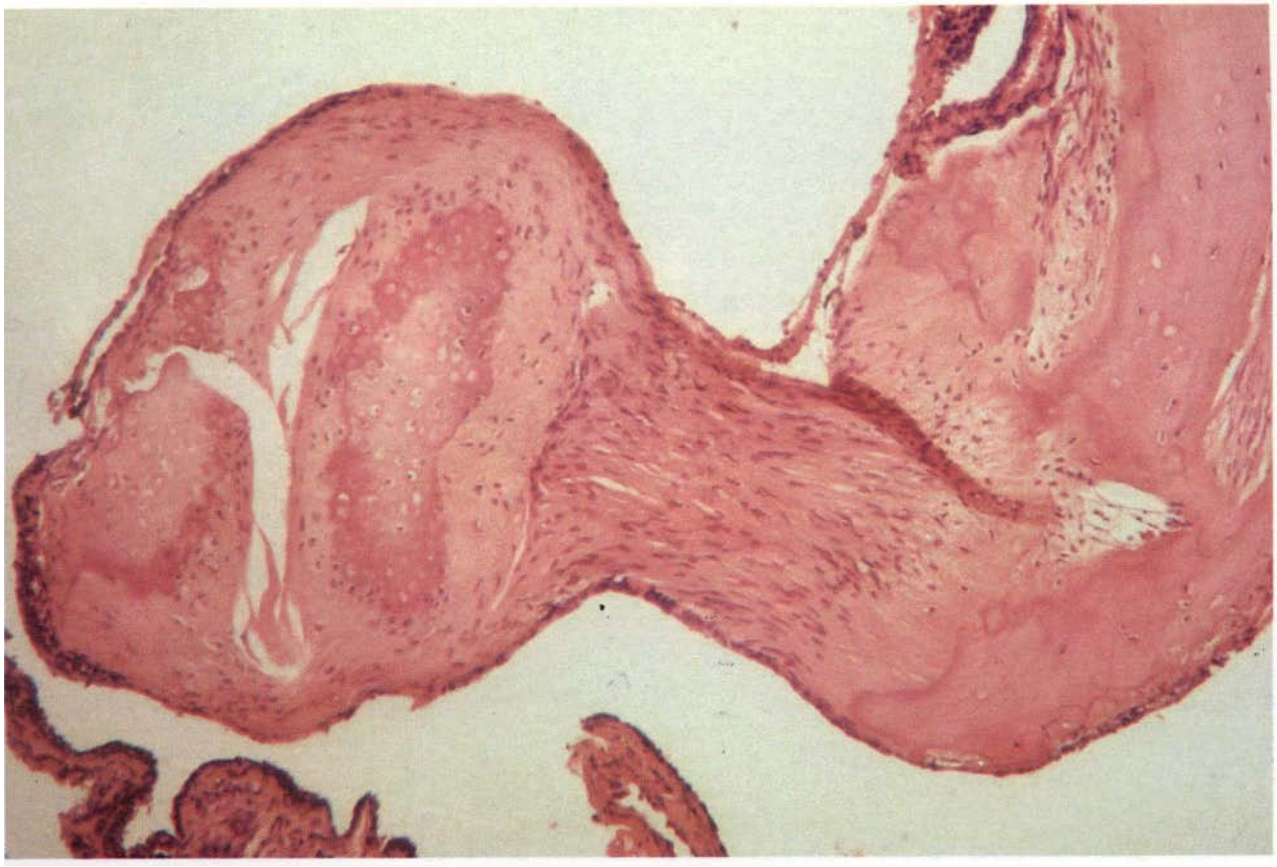

FIG. 2
Stapes removed at surgery.

demonstrated proportionately greater uptake of isotope in the tibia compared to the femur. This feature would appear to be unique to this family and is not present in any other previously described bone disorder.

Biochemical features

Serum alkaline phosphatase and urinary hydroxyproline levels were raised, reflecting increased osteoblastic and osteoclastic activity respectively. Serum calcium and phosphate levels were normal and there was no increase in parathyroid hormone levels. The erythrocyte sedimentation rate was normal in all members of the family.

Histopathological features

Light microscopy of long bone biopsies from these patients showed a wide range of features of increasing severity with duration of disease. Early in the disease, increased members of osteoblasts lined the bony trabeculae along with focal collections of osteoclasts. Mosaic cement lines were prominent. With progression the bony matrix decreased, osteoblasts remained prominent and the amount of fibrous tissue increased and in the final stages affected areas of bone were largely replaced by fatty tissue.

In cases where the stapes was examined the changes were less marked than in the long bones, but examination under polarised light showed increased amounts of woven bone. Figure 2 shows the stapes crura removed at surgery from one of the family. It demonstrates features often found in Paget's disease with marbelling and increased cellularity. When examined under polarised light there was loss of lamellar pattern and abnormal woven bone was present (Fig. 3).

Otological features

Fifty members of the family, aged between 3 years 6 months and 59 years attended the Otolaryngology Department of the Royal Victoria Hospital for assessment. A routine history and examination were followed by pure-tone audiometry and tympanometry.

Nine of the 50 subjects were known not to have the disease; all nine had normal hearing and type A tympanograms.

Of the other 41, nineteen were either known to have, or were strongly suspected of having the disease. All but one of these had a hearing loss. The remaining 22 family members had not been investigated for the bone disease since they were under 20 years of age and biochemical tests were difficult to interpret. None of these 22 had a conductive hearing loss not due to secretory otitis media.

In the youngest members of the family (ages 5–24) the hearing loss was purely conductive with the air-bone gap averaging 20–30 dB. The tympanic membranes looked normal and moved with pneumatic otoscopy. There was a characteristic tympanogram (Fig. 4). The middle ear pressure was within the normal range but there was a very high compliance typical of that found with ossicular chain discontinuity.

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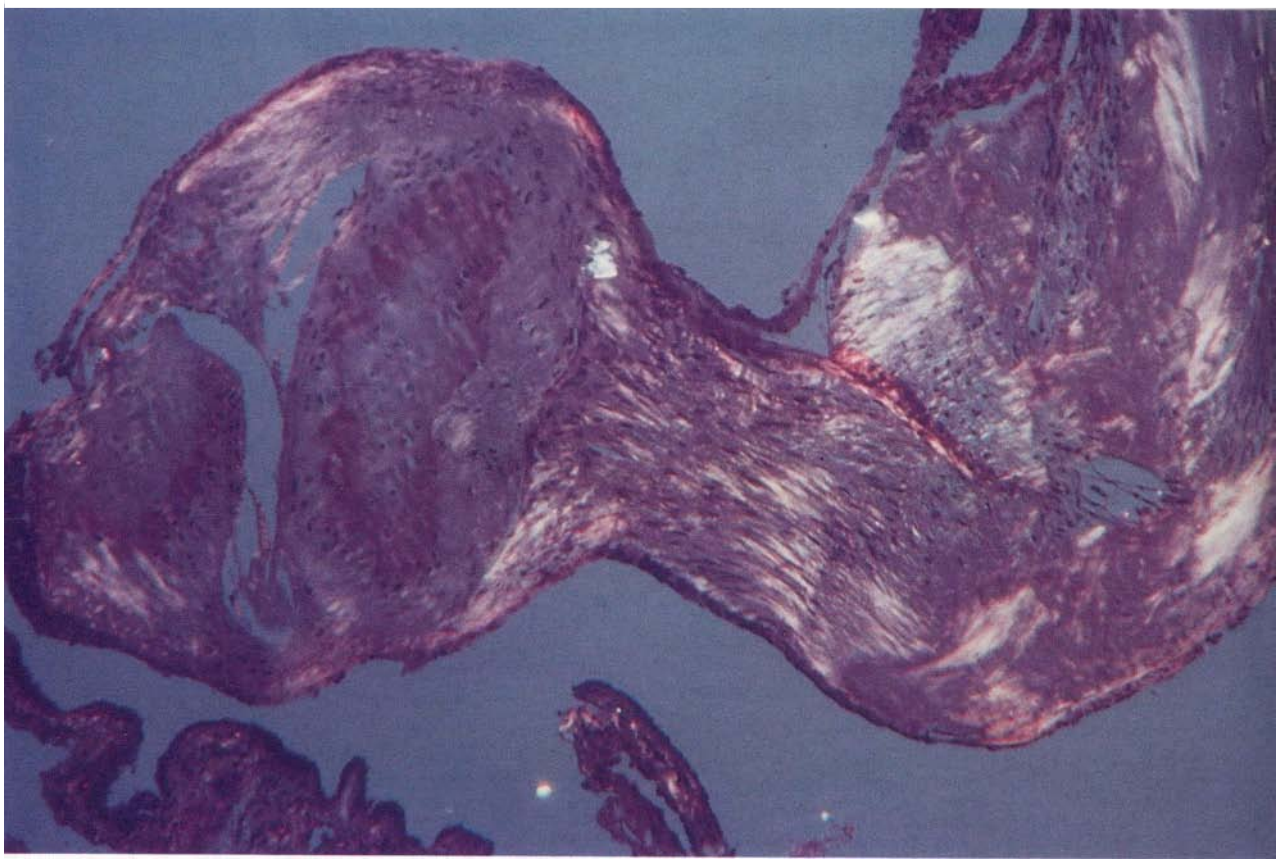

FIG. 3
Stapes under polarised light.

In the older affected members of the family the hearing loss was mixed, with an air-bone gap of 40–45 dB. There was often a well marked notch at 2000 Hz (Fig. 5). The high frequency hearing loss was typically worse than that for the low frequencies. Speech discrimination remained at 80 per cent or better in all but five ears. Tympanometry in this group showed the same abnormally high compliance.

Surgical findings

Ten members of the family have had middle ear surgery by one of two experienced otologists. Two of the patients have had surgery to both ears making a total of twelve ears.

In eleven ears the common findings was that the long process had been replaced by what looked like fibrous tissue. In two of these ears the body of incus was also affected (patients 4 and 7). In one ear (patient 9) the incus was absent. The stapes crura were noted to be abnormal in pateints 1, 2, 3, 4, 5, 9 and 10. In most cases they were replaced by what looked like fibrous tissue, though in one case the crura were thought to be thicker than normal (patient 3).

Five of the patients had a stapes footplate abnormality. In two patients (3, 8) the footplate was thick but

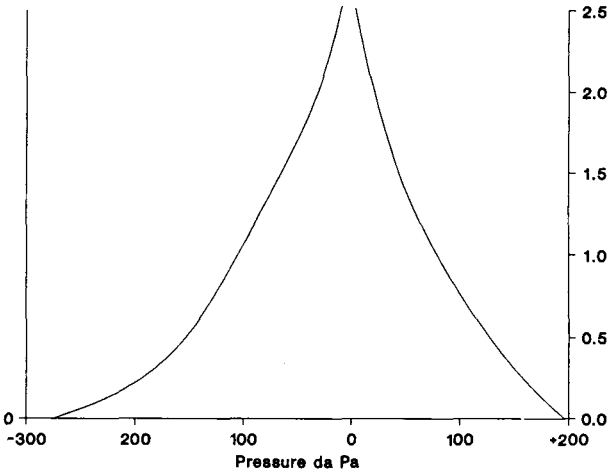

FIG. 4
Tympanogram from affected member of family.

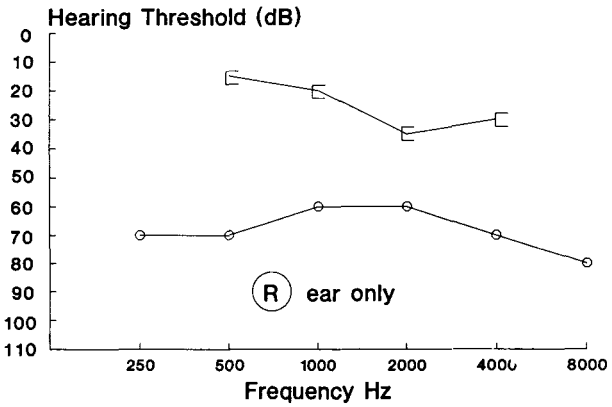

FIG. 5
Typical audiogram from affected adult.

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TABLE I

Patient		Middle Ear Surgery	AB gap (dB) Before surgery	AB gap (dB) After surgery
1	1970	Homograft incus, stapes to malleus	42	42
	1972	Fibrous stapes arch. Boomerang strut stapes F.P. to tympanic membrance		38
2	1980	Adhesions divided. Long process incus thin	18	25
	1983	Stapedectomy (teflon prosthesis)		3
3	1981	Right ear. Homologous incus as malleus stapes assembly	42	28
	1981	Right ear. Stapes abnormally mobile. Proplast prosthesis, oval window to malleus		13
	1982	Right ear. Prosthesis slipped. Homologous incus (oval window to malleus)		12
	1987	Left ear. Homologous incus as malleus stapes assembly	48	42
4	1983	Right ear. Homologous incus footplate to malleus	43	12
	1986	Left ear. Homologous incus footplate to malleus	40	7
5	1984	Autologous incus and homograft incus (footplate to malleus)	38	37
	1985	Incus repositioned		37
6	1984	Torp, footplate to malleus	35	45
7	1987	Homologous incus, footplate to malleus	55	35
8	1987	Homologous incus as malleus stapes assembly	36	30
9	1987	Homologous incus oval window to malleus	32	5
10	1987	Cervital prosthesis incus to stapes footplate	30	45
	1988	Cervital prosthesis moved medially, hole in footplate. Fat graft, prosthesis repositioned		13

mobile. Patient 2 had an immobile footplate and patient 7 had no footplate, the oval window being closed by a 'dark membrane'.

None of the patients had any obvious abnormality of the malleus or reduced movement of the malleus head in the attic.

The results of surgery have been disappointing. Six of the 12 ears have a satisfactory closure of the air-bone gap to within 15 dB. In the other ears there was no improvement in hearing after surgery, though none of these ears developed a sensorineural hearing loss as a result of surgery.

Discussion

The precise nature of the bone disease in this family is unknown. The differential diagnosis includes polyostotic fibrous dysplasia, osteitis fibrosa cystica (hyperparathyroidism) and Paget's disease.

The histological features of both long bone biopsies and ossicles removed at surgery were similar to those found in Paget's disease. Again this diagnosis was supported by the biochemical findings. On the other hand, the distribution of bony lesions, radioisotope scans and early onset of symptoms were atypical.

One notable feature in this family was the early onset of conductive hearing loss. Juvenile Paget's disease is very rare (Smith, 1977). Patients who have hearing loss due to Paget's disease usually have a mixed loss (Davies, 1968; Harner *et al.*, 1978). Davies found that the air-bone gap averaged 30 dB for females and 20 dB for males. In the Belfast family the average air-bone gap in established disease was 40–45 dB, typical of that found with ossicular discontinuity. There was further evidence of this in the abnormally high compliances found by tympanometry.

The surgical findings in this family included disease of the long process of incus, stapes crura and footplate. Paget's disease only rarely involves the stapes footplate (Davies, 1970; Schuknecht, 1974). In addition, in Paget's disease epitympanic spurs and changes in the ossicles may result in decreased mobility of the malleus and incus. None of the patients in this study had disease affecting the malleus and in all the malleus mobility was normal.

Fibrous dysplasia of the temporal bone was reviewed by Nager, *et al.*, (1982). They noted the early onset of clinical symptoms, usually in the 10–20 age group. Most patients presented with progressive hearing loss, which was usually conductive in the early stages. Familial polyostotic fibrous dysplasia, however, is extremely rare. The only reported example involved in the craniofacial skeleton alone. None of the Belfast family had obvious craniofacial abnormalities. Nager, *et al.*, described the surgical findings in three cases. In one there was distortion of the middle ear with the 'ossicles involved'. In another the malleus was noted to be thicker than normal and in the third the long process of incus had disappeared. In the Belfast family there was no histological evidence of fibrous dysplasia in either long bone biopsies or ossicles removed at surgery, apart from fibrous replacement of the long process of incus.

The other differential diagnosis, osteitis fibrosa cystica, can be excluded by the normal serum calcium, phosphate and parathormone levels in this family.

Conclusions

This family presented with an autosomal dominant dysplasia of bone. The histological features might suggest that this is an atypical form of Paget's disease. The age of onset, distribution of lesions, radiographic and middle ears findings are against this diagnosis. There is no evidence of a polyostotic fibrous dysplasia. This family would appear to have a previously undescribed bone disorder, the cause of which is as yet unknown.

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