Pycnodysostosis

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SUMMARY The first Scottish family with pycnodysostosis is reported. The clinical and radiological findings in the two affected men are recorded.

This rare sclerosing bone dystrophy was first described by Maroteaux and Lamy in 1962¹ as a variant of osteopetrosis (Albers-Schonberg disease). They suggested the eponymous title of 'maladie de Toulouse-Lautrec' who is thought to have been

affected by this disorder.² There have been a number of published reviews and reports of other cases since then, with over 100 cases having been listed.³ In this first Scottish family we report two brothers who illustrate the chief features of this proximal short limbed dwarfism and the problems in managing ununited fractures. They are the oldest subjects so far reported.

Case reports

Case 1 (II.2, fig 1) was born in 1932 after a normal pregnancy; his father was 33 and his mother 24 years old. His early life was normal, with his first fracture occurring at the age of eight years. X rays at that time showed general osteosclerosis of the skeleton. Thereafter he suffered a series of fractures of limb bones (fig 2) and his growth became stunted. Nevertheless he married and worked as a music teacher. He had a minor episode of osteomyelitis of

FIG 1 Family pedigree.

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FIG 2 AP radiograph of the pelvis of case II.2 aged 52 years showing the ununited fracture of the right femoral neck and the incomplete internal fixation.

his mandible at the age of 40 after a tooth extraction. This quickly responded to treatment with antibiotics. Since 1978 his two most recent fractures in the left femoral shaft and right femoral neck have failed to unite despite internal fixation, prolonged splintage in bed, and electrical induction therapy.

On examination, case 1 is 160 cm tall with marked shortening of his upper arms and thighs. His nose and chin are pointed. The forehead is prominent with some bossing, but the occiput is flattened. The angle of the mandible appears normal. His head looks large with a circumference of 59.8 cm. The palate is grooved and high arched with irregular dentition. He also has middle ear deafness. His hands are a striking feature with shortening of all the digits, especially the distal phalanges which are ulnar deviated, except for the normal right thumb. Here the nail is also normal, whereas on the fingers the nail is short, sometimes lens shaped (that is, greater width than length), and overhanging the pulp. He has a mild kyphoscoliosis. The big toes are short with fixed plantar flexion. The only joint stiffness is in the knees where it is of recent origin consequent upon his ununited femoral fractures.

Case 2 (II.4, fig 1), the younger brother, was born in 1944 after a normal pregnancy. He was noted to have shorter limbs than average and at the age of eight suffered the first of several fractures of both femora and humeri which have failed to unite. He has worked for only short periods and is now almost

housebound, moving with difficulty on crutches with both legs splinted. Electrical induction therapy applied diligently over a period of 18 months has had no effect on one femoral fracture.

He is approximately 144.5 cm in height with marked shortening of the humeri and femora. His features are sharp without hypoplasia but with a prominent forehead. The teeth are irregular and the palate ridged. The skull is dolichocephalic (fig 3). The fingers are short and deformed with lens shaped nails (fig 4). The shoulders and clavicles appear normal although shoulder movement is limited by ununited fractures of the necks of the humeri. The spine is normal. The right thigh is oedematous with an ununited fracture. The big toes are shorter than the second toes.

Family history

The pedigree is shown in fig 1. The father's family can be traced in the district back to 1620, the mother's for a much shorter period, though one line is also of local origin. We have not been able to show any consanguinity, but this cannot be conclusively excluded. Their mother when aged 70 had a hip replacement arthroplasty, when her bone was found to be macroscopically and radiologically normal. She has had no biochemical or haematological abnormality. Their older brother, II.1, and his two children have been examined. No external locomotor abnormality was found and all three were of normal stature. The fourth sib, a sister, died of an unknown cause aged one month. The wife of case 1

FIG 3 Skull x ray of case II.4.

FIG 4 A view of the hands of case II.4 at the age of 40 to show lenticulate nails and irregular resorption of the phalanges.

is unrelated, though from the same area, and is under 150 cm in height. Their two children are aged 23 and 24 years. Although also of short stature, only one has sustained a fracture. They do not show any locomotor abnormality and radiographs of their hands are normal.

RADIOGRAPHS

A very large number of *x* rays has been taken of both patients over the past 20 years mainly featuring the multiple limb fractures. From the first, osteosclerosis is the most notable feature in all parts of the skeleton. In the skull there is some thickening of the calvarium but no lack of fusion of the sutures and no Wormian bones. The mandibles show angles of 120° to 130°. The frontal sinuses are very poorly developed.

The long bones show osteosclerosis throughout their length, but this is more obvious proximally. There is obliteration of the corticomedullary junctions. The clavicles are of normal shape and size.

The hands and fingers show increased density and resorption of the terminal phalanges and there is some resorption of some middle phalanges also (fig 5).

Discussion

As a form of proximal short limbed dwarfism, pycnodysostosis is distinguished by the disproportionately large calvarium, small face with prominent nose, obtuse mandibular angles, and recurrent fractures. The terminal phalanges are shortened by dysplasia and perhaps resorption. That

the lenticulate shape of the nails is related to the loss of bulk of the phalanx is strongly suggested by the normal appearance of the nail on the normal right thumb in case 1.

It is the appearance of the fingers which is characteristic as to suggest the correct diagnosis in subject with recurrent fractures and radiological osteosclerosis. Other dysmorphic features, such as a

FIG 5 AP radiograph of the left fingers of case II.4 aged 40 years. The irregular absorption of the terminal and middle phalanges is clearly seen.

TABLE Clinical and radiological features of pycnodysostosis.

Previous reports	Case 11.2	Case 11.4
Short stature	+	+
Facial hypoplasia	—	—
Hooked nose	pointed	pointed
Frontal bossing	+	+
Receding chin	prominent	prominent
Narrow, ridged palate	+	+
Irregular dentition	+	+
Short terminal phalanges	+	+
Lenticulate nails	+	+
Bone fragility	+	+
Osteomyelitis	+	—
Osteosclerosis	+	+
Ununited fractures	+	+
Persistent fontanelles and sutures	—	—
Hypoplasia of acromial end of clavicle	—	—
Mandibular angle	130°	120°
Lack of segmentation of atlas/axis	—	—

ridged yet narrow palate, are present (table) and distinguish it from osteopetrosis, as does the absence of anaemia. Osteosclerosis is not found in cleidocranial dysostosis or osteogenesis imperfecta. Pycnodysostosis is inherited as an autosomal recessive disorder. Although X linkage could not be excluded in this family, consanguinity is possible and indeed quite likely in their small, stable community, though it could not be formally proved. A figure of

30% has been recorded in reported series of pycnodysostosis.⁴ No chromosomal abnormalities were present in the two patients reported.

Several authors have commented on the difficulty in securing union of the long bone fractures and the problems of internal fixation.⁵ We are not aware of any other attempts to produce bone union in this condition by electrical coils or implanted electrodes.

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