PYCNODYSOSTOSIS

Z. EMAMI-AHARI, M. ZARABI and B. JAVID, SHIRAZ, IRAN

From the Nemazee Hospital, Shiraz

Pycnodysostosis was first described and its present term coined by Maroteaux and Lamy (1962, 1965, 1966) under the heading of diastrophic dwarfism. They distinguished the condition from cleidocranial dysostosis and osteopetrosis and claimed that Toulouse-Lautrec had suffered from it. The disease is rare, having been reported in only thirty-three patients (Elmore 1967). The purpose of this communication is to discuss the condition further and to present some unusual findings in an additional patient.

The disease is characterised by proportionate dwarfism, well developed secondary sex characteristics, peculiar facies, prominent forehead, beaked nose, receding jaw, abnormal dentition with a usually normal palate—although sometimes it is cleft—and certain skeletal changes with multiple spontaneous fractures; finally there is usually consanguinity in the parents (Maroteaux and Lamy 1966; Dusenberry and Kane 1967). The similarity in appearance of the patients (Figs. 1 to 3), regardless of sex and race, is striking (Elmore 1967).

CASE REPORT

A thirty-seven-year-old woman from a southern province in Iran was referred to us with a draining fistula from the right side of the mandible which had been present for two years since tooth extraction and fracture of the mandible. She gave a history of multiple fractures of the upper and lower limbs, so numerous that she could not remember how many. There was a pseudarthrosis in the midshaft of each tibia, obliging her to wear long leg calipers. She had had several operations on the mandible and on the legs, including plating and bone grafts. She gave no family history of multiple fractures or short stature in her parents, who were first cousins. Her mother was examined by one of us and found to be normal. Her
father, allegedly a normal person, had died from an unknown disease. Two siblings, both older than our patient, had died in early childhood of uncertain diseases at the ages of three and four years.

On admission the patient was in good general condition. Her height was 127 centimetres, with an upper to lower limb ratio of fifty-eight to seventy-three, a sitting height of 70 centimetres and weight of 34 kilograms. The upper limbs were deformed because of old malunited fractures. There were pseudarthroses of both tibiae with scars of previous operations on the shins. The hands and feet had short digits with overlying cutaneous wrinkles and were tapered off with large overriding nails (Figs. 4 and 5). Joint movements were all within normal limits. There was no defect in cranial or spinal nerves. She had wide cranial sutures with all the facial characteristics of pycnodysostosis. The teeth appeared to be normal although rather small, with normal implantation and no double rowing. The palate was narrow and shallow. Laboratory findings were all within normal limits, including blood haemoglobin, differential cell counts, serum calcium, phosphorus, alkaline and acid phosphatase.

![Fig. 4](image1)

Figs. 4 and 5
Figure 4 shows the short spoon-shaped fingers and overriding nails and wrinkles. Figure 5 shows the shins with the pseudarthroses.

Radiological findings—There was generalised increased density of the skeleton and thickening of the cortices of the long bones, but without obliteration of the medullary canals. The skull showed protruding parietal bones, open cranial sutures and obliteration of the frontal sinuses with increased density of the base of the skull (Fig. 6). Dental radiographs showed the roots of the teeth to be thickened and over-cemented but with no impacted or supernumerary teeth. The mandible was hypoplastic and there was a virtual loss of mandibular angles with a fracture at the right angle and a small sequestrum at the fracture site. Radiographs of the thoraco-lumbar spine showed compression fractures of several vertebral bodies with notching of the anterior surfaces (Fig. 7). The pelvis showed evidence of previous surgery, most probably for removal of a graft, increased acetabular angles, bilateral coxa plana and increase in neck-shaft angle (Fig. 8). The extremities revealed multiple malunited fractures in both forearms (Fig. 14) and femora and non-union in both tibiae (Figs. 9 and 10). The chest showed multiple old rib fractures (Fig. 13); the clavicles, heart and lungs were normal. Hands and feet demonstrated aplasia of the tufts of some phalanges and shortening of others (Figs. 11 and 12).
Figure 6—The skull shows open sutures and fontanelles with non-aerated paranasal sinus. The facial bones are small, the mandibular angle is decreased and on the right fractured, and a sequestrum is present. Figure 7 shows the spine. Figure 8 shows shallow acetabula with coxa plana but the pelvis otherwise normal. Note that in these and the following radiographs there is generalised bone density and wide intramedullary canals.
During her stay in hospital the patient sustained another pathological fracture from minor trauma, this time in the midshaft of the right humerus.

**Management**—The patient was first treated for osteomyelitis of the mandible by sequestrectomy and curettage. The tibial fractures were treated by insertion of intramedullary nails and plaster fixation (Fig. 15). The right leg showed clinical and radiological evidence of sound union four months after the operation and she is now bearing weight on it. At the time of review the left leg was still in plaster two months after nailing. The fracture of the humerus was treated by a plaster U-slab and there was sufficient callus formation after ten weeks to allow removal of the plaster.
Figure 13 shows the thorax: Figure 14 the left forearm with a fracture of the ulna; and Figure 15 the fractures of the tibiae healing after Kuntscher nailing.

### TABLE I

**The Salient Features of Osteopetrosis, Cleido-cranial Dysostosis and Pycnodysostosis**

<table>
<thead>
<tr>
<th>Features</th>
<th>Osteopetrosis</th>
<th>Cleido-cranial dysostosis</th>
<th>Pycnodysostosis</th>
</tr>
</thead>
<tbody>
<tr>
<td>Base of skull</td>
<td>Dense</td>
<td>Normal or rarely dense</td>
<td>Dense</td>
</tr>
<tr>
<td>Cranial sutures</td>
<td>Normal</td>
<td>Normal</td>
<td>Open</td>
</tr>
<tr>
<td>Paranasal sinuses</td>
<td>Unaerated</td>
<td>Normal</td>
<td>Unaerated or closed</td>
</tr>
<tr>
<td>Mandible</td>
<td>Normal</td>
<td>Normal</td>
<td>Loss of angle</td>
</tr>
<tr>
<td>Clavicle</td>
<td>Present and normal</td>
<td>Absent or dysplastic</td>
<td>Present, sometimes dysplastic</td>
</tr>
<tr>
<td>Hands and feet</td>
<td>Normal</td>
<td>Normal</td>
<td>Aplastic tufts, short phalanges, overriding nails</td>
</tr>
<tr>
<td>Pelvis</td>
<td>Coxa vara</td>
<td>Normal</td>
<td>Coxa plana</td>
</tr>
<tr>
<td>Spontaneous fractures</td>
<td>Present</td>
<td>Absent</td>
<td>Present</td>
</tr>
<tr>
<td>Bone texture</td>
<td>Dense with obliteration of intramedullary canals</td>
<td>Normal</td>
<td>Dense without obliteration of intramedullary canals</td>
</tr>
<tr>
<td>Blood findings</td>
<td>Aplastic anaemia</td>
<td>Normal</td>
<td>Normal</td>
</tr>
<tr>
<td>Genetic</td>
<td>Dominant; anaemic type, recessive</td>
<td>Dominant</td>
<td>Recessive</td>
</tr>
<tr>
<td>Stature</td>
<td>Usually normal</td>
<td>Usually normal</td>
<td>Short</td>
</tr>
</tbody>
</table>

**VOL. 51 B, NO. 2, MAY 1969**
DISCUSSION

Thirty-three cases of pycnodysostosis have been described since 1923 and this report adds one more. The condition has been reported in all races and in both sexes between the ages of nine months and forty-three years. The patients usually present with multiple fractures or abnormal stature and are found to have characteristic features of the disease. The differential diagnosis is between cleidocranial dysostosis and osteopetrosis, the three conditions sharing certain combined features (Table I). Cleidocranial dysostosis (Forland 1962) is an inheritable disorder transmitted as an autosomal dominant trait and is manifested by a usually normal stature, total or partial absence of one or both clavicles as a constant finding, and normal texture of bones except for increased density of the base of the skull in some cases. Osteopetrosis (Kneal and Sante 1951) is characterised by a generalised increased density of bone which is a dominant trait. The malignant forms are recessive, in which there is severe aplastic anaemia caused by the obliteration of the medullary canals, hepatosplenomegaly and early death. Pycnodysostosis is an autosomal recessive and the patient has characteristic facies, dwarfism, aplasia of tufts or of the digits, generalised increase in density of bones even though not sufficient to obliterate medullary canals or cranial orifices, abnormal dentition, and bone healing of normal rate with normal blood findings.

The following differences were observed between our patient and those previously reported.
1) There was no history of double rows of teeth, or of supernumerary teeth. 2) There was no delay in eruption of deciduous teeth or permanent teeth in the patient or her mother.
3) The palate appeared narrow and shallow. 4) There was no evidence of cleft palate in the patient or her mother.

SUMMARY

1. A patient is described, the thirty-fourth reported, who shows all the skeletal changes of pycnodysostosis with the exception of the oral findings.
2. It is suggested that the pseudarthroses of the tibiae in our patient cannot be taken as evidence against normal union because of the possibility of inadequate management of the fractures.
3. We consider that the use of strong intramedullary nails in the treatment of long bone fractures in these cases is advisable.

REFERENCES