PACHYDERMOPERIOSTOSIS WITH ACRO-OSTEOLYSIS

A REPORT OF FIVE CASES

P. B. GUYER, F. J. BRUNTON, M. W. G. WREN

From the Departments of Radiology of the Southampton General and Royal Portsmouth Hospitals

Five patients with pachydermoperiostosis are described. Three of them showed the incomplete form, and two the forme fruste. Four of the five patients showed acro-osteolysis, a finding which is not well recognised in this syndrome. The natural history of the disease is reviewed, and the differential diagnosis is discussed, with particular reference to hypertrophic pulmonary osteo-arthritis.

Pachydermoperiostosis is an uncommon clinical condition, and as far as we can ascertain fewer than 100 cases have so far been described. Angel (1957) claimed to describe the first three cases in England. We have been unable to find any record of a description of pachydermoperiostosis with acro-osteolysis, although this association is mentioned in textbooks by Kreekl (1971) and Taybi (1975). Weens and Brown (1945) describe a patient with periosteal thickening and acro-osteolysis without actually mentioning the diagnosis of pachydermoperiostosis. We therefore now wish to record five further patients with this curious condition, four of them showing varying degrees of acro-osteolysis.

CASE HISTORIES

Case 1. A man aged twenty-nine, of Chinese-Malayan extraction, presented with a complaint of poor vision. Examination revealed ptosis due to thickening of the skin of the eyelids, which was part of a generalised thickening of the skin of the face (Fig. 1). There was enlargement of the hands and feet, with clubbing (Fig. 2). He was diagnosed as having acromegaly, but radiographs of the skull were normal, and films of the hands and feet showed marked periosteal new bone formation (Figs. 3 and 4), which was also evident in the long bones (Fig. 5). Examination of some of his relatives revealed no evidence of the condition in them.

Case 2. A man aged forty-eight, from a small remote rural community in Hampshire, was referred to the orthopaedic department with pain in the knees. On examination he was found to have enlargement of the wrists and knees with large hands and grossly clubbed fingers and toes (Fig. 6); he was diabetic and also suffered from valvular and ischaemic

Fig. 1
Case 1. Figure 1—Thickening of the skin of the face. Figure 2—Clubbing of the fingers. Figure 3—New bone formation in the hands.
Case 1. Figures 4 and 5—New bone formation in feet and long bones.

Fig. 4

Fig. 5

Fig. 6

Fig. 7

Fig. 8

Fig. 9

Case 2. Figure 6—Gross clubbing of the fingers. Figure 7—Loss of normal tubulation in the long bones; calcification of the tendo calcaneus. Figure 8—Absorption of the tufts of the terminal phalanges. Figure 9—Absorption more marked with periostitis on the calcaneus.
heart disease and prostatic enlargement. On further questioning he said that the changes in his hands had been present since adolescence but had never given rise to any symptoms. He admitted to hyperhidrosis of the hands. His face showed marked coarsening of the features due to thickening of the skin. Radiological examination revealed widespread gross enlargement of bones with periosteal thickening and irregularity; there was ossification extending into the tendo calcaneus, and there was loss of the normal tubulation of the long bones (Fig. 7). Radiographs of the hands revealed partial absorption of the tufts of the terminal phalanges (Fig. 8), and in the feet this absorption was marked and there was also periostitis affecting the calcaneus (Fig. 9). In the twenty-seven years he was under observation there was no change in radiological appearances, but the formation of periosteal new bone at the elbows had necessitated ulnar nerve decompression and he had had recurrent haemarthroses in the knees and elbows, which were severely affected by osteoarthritis. He recently died of congestive heart failure aged seventy-six.

**Case 3.** The sister of Case 2 was seen shortly after the diagnosis was made in her brother. She was aged fifty-eight and had never suffered on account of her condition. Physical examination revealed coarsening of the facial features due to thickening of the skin, and pronounced clubbing of the fingers and toes. Radiographs showed typical periosteal thickening and irregularity of the shafts of the long bones, and partial absorption of the tufts of the terminal phalanges (Figs. 10 and 11).

**Case 4.** A woman aged forty-two of Anglo-Indian extraction presented with symptoms of diabetes mellitus. Physical examination revealed pronounced clubbing of the fingers, and there was some
coarsening of the facial features. Radiographs of the hands revealed gross acro-osteolysis, and similar changes were present in the feet (Figs. 12 and 13). Examination of her long bones revealed a loss of the normal tubulation, particularly in the tibiae, but there was also fine periosteal new bone (Fig. 14). There was no known family history.

**Case 5.** A man aged fifty-six presented with varicose ulceration of the leg. He had excessive swelling of the legs, enlargement of the hands and feet, and marked clubbing of the fingers and toes. Radiographs of the legs showed extensive periostitis, which was initially attributed to varicose ulceration or pulmonary osteo-arthritis, but his chest radiograph was normal.

Six years later he was seen again. Radiographs of hands now showed absorption of the tufts of the terminal phalanges (Fig. 15), and those of the long bones showed a marked periosteal new bone formation (Fig. 16). A diagnosis of pachydermoperiostosis was made by Dr R. Thursfield. No mention was made in the notes of any family history, nor of the age of onset of his symptoms. The dermatologist who saw him made no mention of any alteration in his facial features which are therefore presumed to have been normal.

**DISCUSSION**

This clinical condition was first described in two brothers by Friedreich (1868) and further patients were noted by Marie (1890). Both these authors were commenting on the clinical condition affecting the hands and feet, supported by post-mortem studies in one of Friedreich's two cases. Unna (1907) described the condition of cutis verticis gyrata, and Grönberg (1927) associated the latter condition with that described by Friedreich (1868). The combination of findings is sometimes referred to as the Touraine–Solente–Golé syndrome, following their description of cases and review of the literature in 1935 (Susmano et al. 1968). Uehlinger (1941) also described this syndrome. Details of the first of our patients were presented to the Royal Society of Medicine by Langston (1950). Vogl and Goldfischer (1962) described two further patients, one a Negro and the other a Puerto Rican, and commented that the condition was uncommon in women, only ten such cases having been reported by that date. Other descriptions have come from Egypt (Shawarby and Ibrahim 1962), Africa (Findlay and Oosthuizen 1951), Japan (Schinz et al. 1951; Katoh 1969), the United States (Rimoin 1965; Herman et al. 1965; Harbison and Nice 1971), Scandinavia (Ursing 1970), the West Indies (Curth, Firschein and Alpert 1961) and France (Vague 1948).

Most authors describe the onset in adolescence, but both an earlier and a later onset of symptoms have been described (Currario et al. 1961; Vogl and Goldfischer 1962; Ursing 1970). The abnormalities consist of enlargement of the hands and feet with clubbing of the fingers and toes and distal thickening of the soft tissues. These changes were present in varying degrees in all our patients. In addition, the clinical features include coarsening of the skin of the face and scalp; the former was present in all our patients, but none of them had scalp changes. The extent of the periosteal changes may vary from fine (Case 4, Figs. 12 and 13) to gross (Case 2, Fig. 6). Although it is stated to be rare for the tarsus and carpus to be involved, a tarsal involvement was present in our second patient. Only Uehlinger (1941) has described involvement of the pelvis. The skull may be affected (Schönemberg 1954) but the lumbar spine is said to be always normal, as it was in all our patients.

With the exceptions mentioned earlier, acroosteolysis has not been related to pachydermoperiostosis in case histories recorded in the medical literature. In one instance a careful radiological survey of the skeleton was needed to detect the other bone changes in a patient with gross acro-osteolysis, but in the others the degree of osteolysis was less marked, even when other bone changes were gross. The association of osteolysis with finger clubbing helps to differentiate this condition from others of which osteolysis is a feature with the exception of Cheney's syndrome, in which there are also other abnormalities including osteoporotic wedging of vertebrae, edentulism, and wide skull sutures. Familial osteolysis starts in childhood, as do hereditary multicentric osteolysis, hereditary osteolysis, and osteolysis with nephropathy; they also have other bone abnormalities and distinctive clinical features. Pyknodysostosis is accompanied by bone sclerosis and dwarfism; vinyl chloride poisoning is distinguished by exposure to the chemical, and the absence of clubbing. Scleroderma is accompanied by cutaneous vascular changes, hyperparathyroidism by biochemical abnormalities, and psoriasis by erosive arthritis and skin lesions.

Pachydermoperiostosis generally progresses for approximately ten years before arresting spontaneously, and rarely gives rise to symptoms, although hyperhidrosis and arthralgia may occur and give cause for complaint. The condition is described as occurring in
complete or incomplete forms, or as a forme fruste. The complete syndrome consists of pachydermia with periostitis and cutis verticis gyrata; the incomplete form spares the scalp, and the forme fruste has minimal periostitis (Ursing 1970). Based on this classification our first three patients showed the incomplete form, and the last two the forme fruste. Between 40 and 50 per cent of patients gave a family history of the condition (Brugsch 1941; Vague 1948; de Seze and Jurmand 1950; Vogl and Goldfischer 1962), through a recessive or incompletely autosomal dominant mode of inheritance. Curth et al. (1961) consider familial clubbing to be a minimal expression of the syndrome.

The main differential diagnosis is from hypertrophic pulmonary osteo-arthritis. Nearly all the clinical abnormalities described with pachydermoperiostosis have been described in both conditions, and it appears that the lesion can only be satisfactorily differentiated radiologically by the firm exclusion of a pulmonary lesion (which is usually, but not always, neoplastic). However, there are clinical differences, particularly the age of onset, the presence of a family history in pachydermoperiostosis, and of pain in the secondary form in which the periosteal changes are generally less marked. Although the condition has been confused with acromegaly, the absence of endocrine disturbances and the presence of the marked bone changes make the differentiation possible (Camp and Scanlon 1948). Acromegaly enhances tufting of the terminal phalanges, and does not cause osteolysis. Thyroid acropachy also must be considered in the differential diagnosis, but this is usually associated with exophthalmos and pretibial myxoedema, and there may be other evidence of disturbed thyroid function. Paget’s disease of bone should not present any problems since the radiological features are quite distinctive.

We wish to thank our colleagues for permission to publish details of the case histories of their patients.

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