AN UNUSUAL LIPOID RETICULOSIS OF BONE

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The patient described in this paper presented an affection of the skeleton which has not, so far as we are aware, been recorded previously.

CASE REPORT

A Muganda boy aged eight years was admitted to Mulago Hospital, Kampala, with a painful swelling of his left foot and leg. Ten months previously the distal phalanx of his left great toe had been amputated elsewhere for some septic condition. Two months later the left foot became painful and swollen and the swelling gradually spread up towards the knee. On examination the foot and the leg as far as the knee were diffusely swollen (Fig. 1). The overlying skin was normal though stretched: there was no oedema and no open or healed sinus. There was a slight rise in temperature over the affected area but no tenderness. Movement of the ankle joint was only slightly impaired. The affected leg was one inch longer than its fellow, measured from the knee to the ankle. There was no enlargement of the spleen. The patient was afebrile while under observation.

Investigations—White cell count 10,200 per cubic millimetre (neutrophils 56 per cent; lymphocytes 36 per cent; eosinophils 4 per cent; monocytes 4 per cent). Red cell count 4,340,000 per cubic millimetre. Haemoglobin 68 per cent (9.2 grammes per cent). Colour index 0.80. Examination of the urine showed no Bence-Jones proteose. The blood calcium was 9.8 milligrams per cent on two occasions at ten weeks interval. Blood phosphorus was 5 milligrams per cent and 4.5 milligrams per cent on two occasions at ten weeks interval.
Blood cholesterol was 196 milligrams per cent. The Kahn reaction was doubtfully positive. 

Radiographic examination (Dr J. Scott Brown)—The whole skeleton was examined. Lesions were found only in the left foot, the left tibia and fibula, and the right humerus. The affected

Figure 3—Left knee, showing early involvement of the lower femoral epiphysis. Figure 4—Antero-posterior radiograph of left foot. The right foot was normal.

Fig. 5
Lateral radiograph of left foot.

bones were radiographed again six months after the first examination and no significant change was detected. Left leg—The lesion involved the whole of the tibia and fibula (Fig. 2) and all the bones of the foot (Figs. 4 and 5). There was a small zone of abnormality in the left lower
Femoral epiphysis (Fig. 3). The bones were irregularly expanded, their cortices were thin in some places not detectable. The whole thickness of the bones was occupied by fairly widely spaced coarse trabeculae. In the upper third of the fibula there was a core of coarse trabeculae in the normal line of the shaft of the bone, and radiating perpendicularly from it spicules of trabeculation ending in a faint line of calcification marking the new boundary of the bone. The right humerus showed a cyst-like area in the middle of its shaft (Fig. 6). The overlying bone was expanded, and its cortex thinned. Proximal and distal to the zone of rarefaction the original films showed coarse and thickened medullary trabeculation. No lesion was seen in any of the other bones of the body, and there was no suggestion of "Erlenmeyer flask" deformity in the lower portions of the femoral shafts.

Histological examination (Dr Basil Elmes)—The left tibia and right humerus were explored.

and in both soft friable tissue was found under a thin cortex of bone. The specimen from the tibia was soft and did not require decalcification. The bone and marrow were replaced by a vascular cellular tissue composed of fibroblasts, large pale rounded or polyhedral cells with granular or finely vacuolated cytoplasm, and a sprinkling of plasma cells, lymphocytes and eosinophils (Fig. 7). The large pale cells were conspicuous (Fig. 8) and probably contained lipoid material; they were indistinguishable from Gaucher cells. Each contained one or two small darkly staining nuclei, sometimes eccentric; in a few there were several nuclei but no giant cells of the osteoclastoma type were seen. There were a few fragments of degenerate bone but there was no evidence of bone regeneration. The specimen from the humerus showed changes essentially the same as those in the tibia, though sheets of large pale "Gaucher" cells dominated the picture and the structure was more delicate and less fibroblastic. There were also small collections of spindle-shaped cholesterol clefts not seen in the other specimen.
Fig. 7
Photomicrograph of section from left tibia (×60).

Fig. 8
Photomicrograph of section from left tibia (×320).

THE JOURNAL OF BONE AND JOINT SURGERY
Subsequent history—The child's parents removed him from hospital. After repeated attempts to get him back, the medical officer of the district in which he lives persuaded them to allow him to drive the child 150 miles to Kampala for further radiographic examination. This was undertaken one and a half years after the first examination. Although there was considerable local extension of the lesions, most noticeable in the left tibia and fibula, the only evidence of involvement of bones previously unaffected was a lesion in the left lower femoral epiphysis, a suspicion of which had been detected in the previous films. The skull was still normal.

DISCUSSION

The profusely honeycombed appearance of the changes in the left leg suggested angiomatous infiltration of the bones, but the histological findings put this out of court. The involvement of every bone below the knee might be regarded as indicating a vascular error of some kind, but, apart from slightly increased warmth of the skin, there were no signs of changes in the soft tissues. The overgrowth of the tibia and fibula was probably explained by the abnormal vascularity of the bones.

The lesion in the humerus was compatible with a diagnosis of several different affections, including fibrous dysplasia and one of the lipoidoses: the histology seems to point towards the latter. Though the finding of foam cells in a section does not absolutely exclude fibrous dysplasia, the number present in this case, and their presence in both humerus and tibia, seemed to favour a diagnosis of one of the lipoidoses. According to Snapper (1949) mistakes are more likely to be made in the opposite direction, since secondary fibrotic changes in an old lesion in one of the lipoidoses, with disappearance of all lipid-containing cells, may readily lead to a wrong diagnosis. He advocated the selection of what appears to be a young lesion for biopsy in a doubtful case.

Somewhat similar honeycombing of several bones in a foot has been described, but this was in a case of angio-endothelioma (Fairbank 1951). Hauser and Constant (1948) reported a case with extensive honeycombing of the pelvis and upper femora, very suggestive of the changes seen in this case, but this also proved to be due to angio-endothelioma. A case of interest is that published by Parsons and Ebbs (1940) which, curiously enough, was regarded as Gaucher's disease until the child died from an attack of broncho-pneumonia, when complete post-mortem examination revealed that the widespread cystic changes in the skeleton were due to cavernous angiomata: similar changes were found in the spleen and other organs and also in the soft tissues.

We know of no other case, not even of the most widespread polyostotic fibrous dysplasia, which showed such complete and almost uniform changes in all the bones of the distal half of a limb as are seen here: the profuseness of the changes appears to be unique. We suggest that this case must be regarded as belonging to one of the lipoid reticuloses, possibly of the Gaucher type, but the absence of enlargement of the spleen is a strong point against this last suggestion.

Our thanks are due to Mr T. N. Salthouse for the photograph.

REFERENCES


