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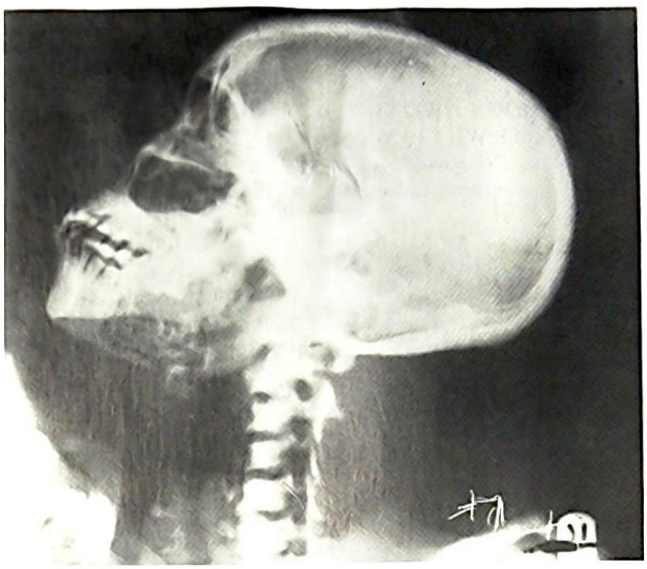

Fig. 1. X-ray of patient's skull — showing chronic osteomyelitis of the left mandible. A sequestrium is present Note the sclerotic base of skull and the vertebral bodies.

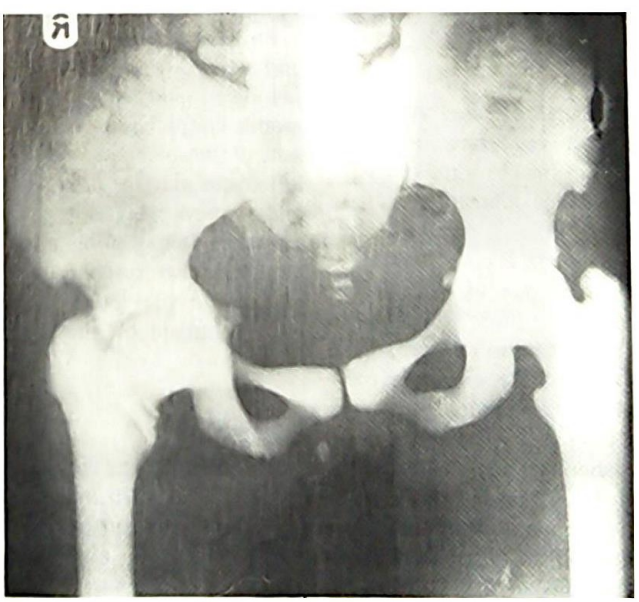

Fig. 2. X-ray of patient's pelvis — showing generalized bone sclerosis with obliteration of the medulla in the femur.

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Osteopetrosis in a Nigerian woman — a case report

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Summary

A case of osteopetrosis complicated by chronic osteomyelitis of the mandible, pathological fractures, pancytopaenia and splenomegaly is presented. Family studies revealed a dominantly inherited pattern.

Résumé

Nous avons étudié un cas d'ostéoporose avec incidence d'ostéomyélite de la mandibule, fractures pathologiques, pancytopénie et splénomégalie. L'étude des antécédents a révélé une forte tendance héréditaire.

Introduction

Osteopetrosis (marble-bone disease, Albers-Schonberg disease) is a very rare osseous disorder, first described in 1904 by a radiologist Albers-Schonberg (cited by Johnston *et al.*, 1968).

The aetiology of this disease is poorly understood. It is thought to be due to osteoclast dysfunction (Johnston *et al.*, 1968; Rimoin & Horton, 1978). Two distinct forms of osteopetrosis are recognized; the infantile, malignant or congenital form, which is inherited in an autosomal recessive fashion, and the benign adult form, which is inherited in an autosomal dominant fashion. The latter is usually compatible with normal survival, although with some complications, while the former is aggressive and invariably fatal, usually in the early life (Johnston *et al.*, 1968, Loria-Cortes, Qvesada-Calvo & Cordero-Chaverri, 1977).

A series of cases of osteopetrosis presenting at the University College Hospital (UCH), Ibadan, Nigeria, was described by Kolawole in 1971. The present report further extends his observation and underlines the dominant pattern of inheritance of the adult variant of the disease.

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Case report

A 43-year-old female Yoruba farmer A.S. (UCH No. QC. 24709) presented in 1985 with a history of painful swelling of the left lower jaw of one year's duration, purulent discharge from the left lower jaw, pain in the left thigh, tiredness and weight loss, all of about 6–8 months' duration.

Physical examination revealed an ill-looking and wasted woman. She was febrile, mildly jaundiced and very pale. There were small mobile lymph nodes in the left axilla. The lower pole of the spleen was palpated 15 cm below the left costal margin; it was not tender. The liver and kidneys were not palpable. There was a diffuse tender swelling of the left mandible with an extra-oral discharging sinus. There was trismus on the left side of the mouth. The patient limped on the left side.

Investigations

Skull X-ray showed chronic osteomyelitis of the left mandible with a sequestrum within it. A pathological fracture of the left mandible was also present. The entire skull was very dense but much denser at the base (Fig. 1). Skeletal survey revealed generalized and uniformly increased density of the whole skeleton. The medullary cavity was almost completely obliterated. There was a pathological fracture of the left thigh (Figs 1, 2 and 3).

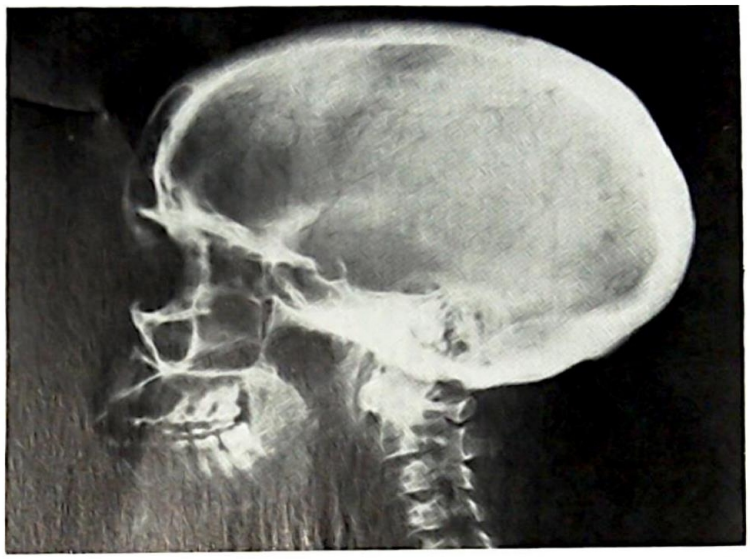

Fig. 4. X-ray of patient's sister's skull — showing increased sclerosis of the calvanium, especially at the base of skull.

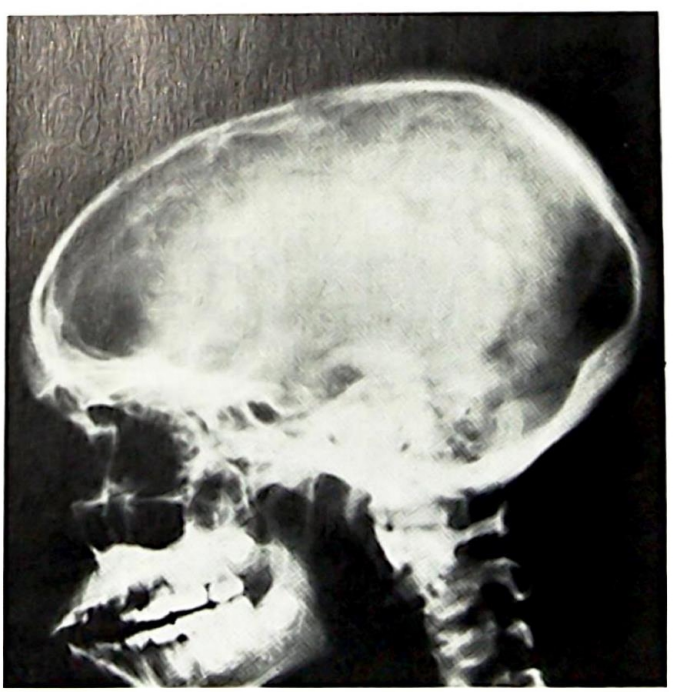

Fig. 5. X-ray of patient's daughter's skull — showing sclerosis at the base.

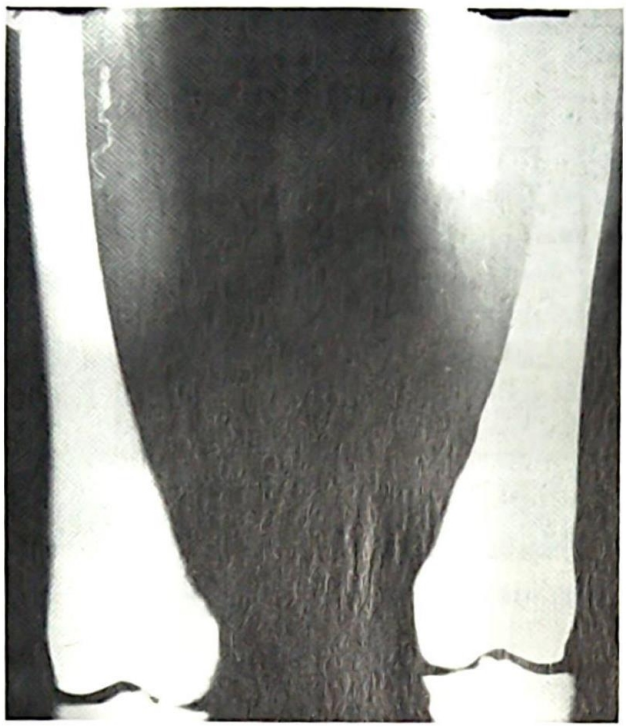

Fig. 3. X-ray of patient's femora — showing generalized sclerosis and a pathological fracture of the proximal shaft on the left side.

Haematological investigations showed pancytopaenia. Haematocrit ranged from 10–20%, white cell count ranged fom 2.1×10^3–$4.2 \times 10^3/mm^3$. Platelets were on the lower limits of normal. The peripheral blood film showed anisocytosis and tear-drop poikylocytisis. Direct anti-human globulin test and Ham's test were negative. Bone marrow aspirations at both the iliac crests and the sternum, as well as biopsy, were unsuccessful owing to stony hardness of the bone. Biochemical examinations were unremarkable: serum calcium and phosphate were normal (9.3 mg/dl and 3.0 mg/dl, respectively): alkaline phosphatase was normal (8.0 K-A units/dl) and acid phosphatase was also normal (3.7 K-A units/dl). Liver function tests were normal. Histological report of the axillary lymph node biopsy revealed extramedullary haematopoiesis.

In order to establish the heritable nature of this patient's illness, two healthy relations, a 48-year-old sister and a 23-year-old daughter were examined, radiologically (Figs 4 and 5). The bone changes observed in them were similar to those of the patient, thus, suggesting a dominant inheritance form of her disease, which showed clinical features of osteopetrosis.

Discussion

Osteopetrosis is a rare osseous disorder. Its aetiology is poorly understood. It is thought to result from deficient osteoclast bone resorption resulting in excessive bone mineralization and the sclerotic changes that are characteristic of this desease (Jonhston *et al.*, 1968; Rimoin & Horton, 1978; Hawke, John & Bailey, 1981). The bone is dense but brittle and is prone to pathological fractures (Johnston *et al.*, 1968; Kuo & Davis, 1981).

Family studies and clinical features in many of the affected patients indicated that osteopetrosis is probably a genetic disorder (Johnston *et al.*, 1968; Loria-Cortes *et al.*, 1977; Horton, Schimke & Iyama, 1980) and two distinct forms of inheritance are recognized; a

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