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Skeletal Radiology

Dyschondrosteosis (Léri-Weill Syndrome) Observed in a VIth Century Skeleton

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Abstract. Study of a case of dyschondrosteosis (Léri-Weill syndrome) was detected in a sixth-century adult skeleton.

In addition to the typical characteristics of dyschondrosteosis (shortness of the forearms and lower legs accompanied by Madelung's deformity of the radius), examination of the skeleton revealed bilateral acetabular dysplasia.

No degenerative change (osteoarthrosis) was observed in the joints directly associated with these malformations.

Key words: Dyschondrosteosis — Léri-Weill syndrome — Congenital dysplasias — Paleopathology.

Although many physicians tend to consider paleopathology simply as an academic exercise, useful only to anthropologists, paleontologists, and archeologists, its essential concern with the skeleton renders its findings important also to radiologists, rheumatologists, and orthopaedic surgeons, since it may provide material usually unavailable in the normal practice of pathology. Moreover, if the diagnosis of specimens from the past can be expressed within modern radiological terms, valuable data are supplied for medical research. In addition, such cultural information may be of psychological value in discussions between the doctor and his patients, particularly in the case of genetic counselling.

This paper describes an interdisciplinary study, anthropological and anatomicoradiological, of a case of dyschondrosteosis (Léri-Weill syndrome) detected in a skeleton dating from the VIth Century, exhumed in Geneva, Switzerland. A complete anthropological account of this skeleton will be published in the report of the "XIIIe Colloque des anthropologistes de langue française, Caen 1977" [3].

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Case Report

The skeleton examined was that of an adult, probably a female about sixty years of age (No 1974-64, catalogue of the Department of Anthropology; No T. 10448/77 of the Department of Pathology). The stature of the living person is estimated to have been slightly more than 150 cm.

The most striking feature of the skeleton was bilateral and symmetrical shortness of the limbs, which was essentially mesomelic, affecting mainly the forearms, and accompanied by symmetrical Madelung's deformities of the distal extremity of each radius (Figs. 1 and 2). The anthropological study, however, also revealed slight bilateral shortening of each humerus and femur. The long bones of the limbs were thick and had pronounced insertion tuberosities, particularly along the linea aspera. Bilateral acetabular dysplasia was present also, accompanied by mild flattening of the femoral heads, more marked on the right side. The femoral necks were short (Fig. 3).

No signs of osteoarthrosis of the wrists or hips were observed, but some indications of osteoarthrosis of the left knee were evident (i.e. osteophytic rims around the femoral and tibial condyles and eburnation of both sides of the posterior part of both lateral femoral and tibial surfaces). Similar changes had affected a trapeziometacarpal joint and a proximal inter-phalangeal joint of a foot. Observation of the vertebrae revealed osteophytes of lumbar spondylosis without any ankylosing hyperostotic bridges; vertebral arthrotic remodelling, either anterior (between vertebral bodies) or posterior (between articular processes) was absent.

Radiological examination of the skeleton revealed no abnormalities other than those described above. The degenerative lesions which were present, being unrelated to the joints adjacent to dysplastic skeletal involvement, were considered to be attributable simply to a coincidental ageing process.

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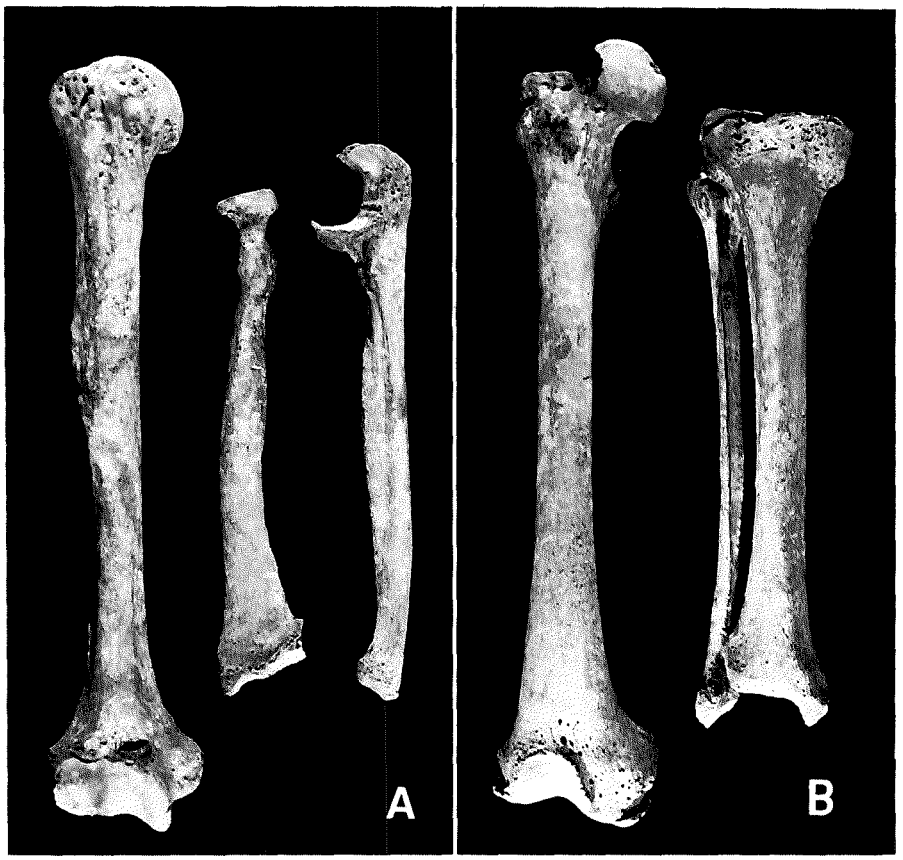

Fig. 1A and B. Bones of right arm and leg.
A Upper limb (×0.4). Shortness of the ulna and the radius with Madelung's deformity. Angularity of the radius and obliquity of its distal articular surface. Congruence of proximal and distal radio-ulnar articular surfaces
B Lower limb (×0.3). Congruence of proximal and distal articular surfaces of the tibia and fibula

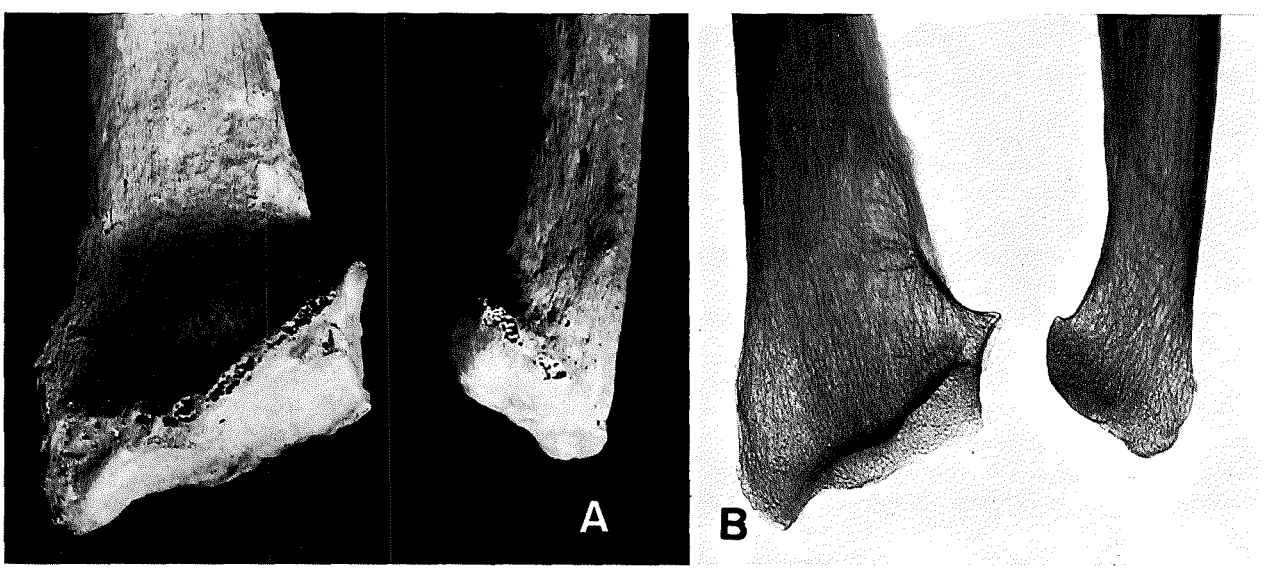

Fig. 2A and B. Distal extremities of right ulna and radius (×1). **A** Macroscopic anterior view. **B** Radiograph

Discussion

The symmetrical mesomelic shortness of the limbs, accompanied by Madelung's deformities observed in the skeleton under study indicates clearly a typical case of dyschondrosteosis, an osteochondrodysplastic condition described in 1929 by Léri and Weill [5]. More frequently observed in women than in men [4], the condition has been reported in Europe, United States, and Japan [4, 6]. It is considered to be hereditary

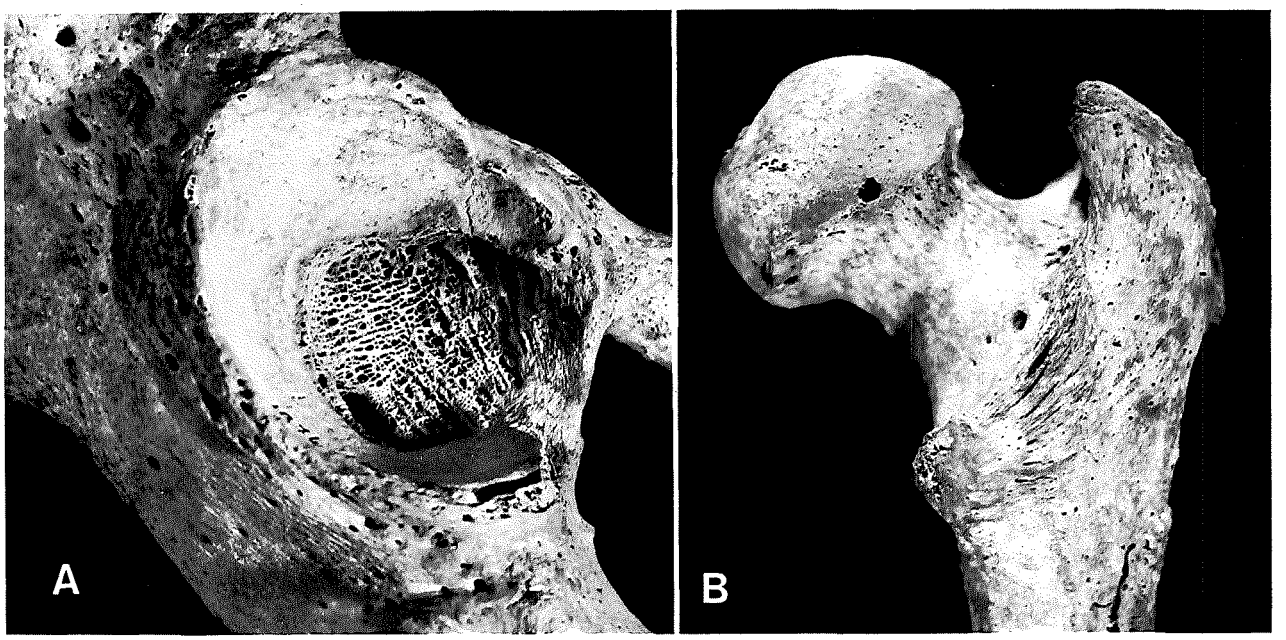

Fig. 3A and B. Dysplastic right hip joint. **A** Posterior view of the acetabulum (×0.7): obliquity of the roof. **B** Posterior view of proximal end of the femur (×0.9): mild flattening of the head and shortening of the neck

and transmitted by an autosomal dominant mode [4, 6], but with a diversity of expression. Madelung's deformity may occur as a *forme fruste* of dyschondrosteosis, this entity being manifested only by an abnormality of the wrist, occasionally unilateral, and not necessarily being accompanied by shortness of stature [4, 6].

Although the case under study seems relatively mild, compared with other cases reported in the literature [1, 4], it included certain elements previously described in the literature, but not regarded as essential criteria for the diagnosis of the condition. Thus dyschondrosteosis can be accompanied by some other malformations [4] and by periosteal bony overgrowth [2, 4, 5]. Indeed, in the present case, we observed bilateral acetabular dysplasia and periosteal thickening on tendon insertions. The presence of such bony overgrowths in a living person with dyschondrosteosis should therefore suggest to the physician the clinical necessity of assessing mechanical and muscular function.

It can be concluded that the malformations described above, at least in the relatively mild form encountered in the present study, do not, *per se*, cause osteoarthrosis. Although signs of osteoarthrosis, probably related to ageing, were present elsewhere in the skeleton, no arthrotic remodelling directly related to the dysplasia was observed.

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