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Case Report

Pyle disease (metaphyseal dysplasia) presenting in two adult sisters

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ABSTRACT

Pyle's disease is an extremely rare skeletal disorder characterized by a benign course and an autosomal recessive genetic pattern of inheritance. Its causal mutation is still unknown. In the medical literature, fewer than 30 cases have been described to date. We report the case of two female siblings, daughters of consanguineous parents, referred to the radiology department complaining of genu valgum. Laboratory tests showed no other relevant findings. Conventional radiography plain films revealed Erlenmeyer flask deformity in bilateral femorotibial metaphyses, metaphyseal flaring of long bones, and mild sclerosis of the skull base. The clinicoradiological dissociation, along with the characteristic imaging findings, was consistent with the diagnosis of Pyle's disease. Intervention is not required in most cases, but orthopedic treatment may be required for genu valgum or fractures. Therefore, these cases emphasize the pivotal role conventional radiography plays in the correct diagnosis of this rare entity, allowing for appropriate genetic counseling.

Introduction

Pyle's metaphyseal dysplasia is a rare genetic skeletal disorder of benign course, inherited in an autosomal recessive pattern, whose causal genetic mutation is still unknown [1,2]. Edwin Pyle, an American orthopedic surgeon, first reported the disease in 1931, describing bone deformities involving the skull and limbs of a 5-year-old child [3,4].

There is striking clinicoradiological dissociation, with mild clinical manifestations. Genu valgum is the most common

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feature on physical examination and is frequently the only consistent finding [1]. Other findings that may be observed on physical examination include bilateral and symmetrical enlargement of the knee, proximal two-thirds of the humerus, distal two-thirds of the radius and ulna, as well as the proximal phalanges and distal metacarpal bones [3,5,6]. Other clinical manifestations include caries, poor dental implantation, malocclusion, mandibular prognathism, limited extension of the elbow, muscle weakness, and arthralgia [5,7]. Some cases may present spinal deformities such as scoliosis [1,7].

Considering both the rarity of this disease and its typical radiographic presentation, we report the cases of two female siblings, daughters of consanguineous parents, with an emphasis on imaging findings. We also discuss the differential diagnosis along with a brief review of the literature.

Case report

Case 1

A 53-year-old woman was referred to the radiology department complaining of genu valgum and joint pain. Her family had noted widening of the knees at the age of 8 years, which progressed over time. There was no functional disability. The patient sought medical care several times, receiving the diagnosis of osteoarthritis with no further investigation. Over the last year, she had developed a more pronounced arthralgia that was not relieved by common analgesics. There was no visual, auditory, cognitive, or psychomotor disturbance.

The most evident finding on physical examination was the marked genu valgum (Fig. 1).

Mild facial dysmorphism, prognathism, and prominent ears and frontal bossing were also noted. The patient showed partial edentulism with caries and poor hygienic condition of the remaining teeth. There were no signs of anemia, jaundice, organomegaly, fractures, or motor disturbances.

Family history revealed parental consanguinity and similar phenotypic characteristics in 2 of her 8 siblings. The parents had no musculoskeletal deformities (Fig. 2).

Laboratory tests did not show any significant alterations.

Conventional radiography plain films of the patient's knees revealed Erlenmeyer flask deformity, characterized by marked femorotibial metaphyseal flaring, and associated cortical bone thinning (Fig. 3).

The additional radiographic survey, which included other long bones, skull, and spine, exposed the symmetry and the systemic nature of the skeletal disorder. Most of the long bones showed undertubulation and loss of their usual morphology. The proximal portions of the clavicles and ribs were expanded (Fig. 4A), as well as the proximal two-thirds of the humerus, which presented an arcuate shape (Fig. 4B). Other tubular bones shared similar characteristics, such as the distal portions of the radius and ulna, proximal and distal portions of the fibula, head of the metacarpals, and base of the phalanges (Fig. 4C). The ischiopubic rami were also diffusely enlarged, thereby narrowing the obturator foramina (Fig. 4D). Despite striking changes in tubular bones, skull radiographs showed only mild basal sclerosis, mandibular prominence, and poor pneumatization of paranasal sinuses and mastoid air cells (Figs. 4E and F).

Preoperative three-dimensional tomographic reconstruction of the lower limbs for surgical planning provided detailed analysis of the Erlenmeyer flask deformity and highlighted the S-shaped aspect of the tibia (Fig. 5A).

Additional T1-weighted magnetic resonance imaging (MRI) of knees revealed normal bone marrow signal intensity (Fig. 5B).

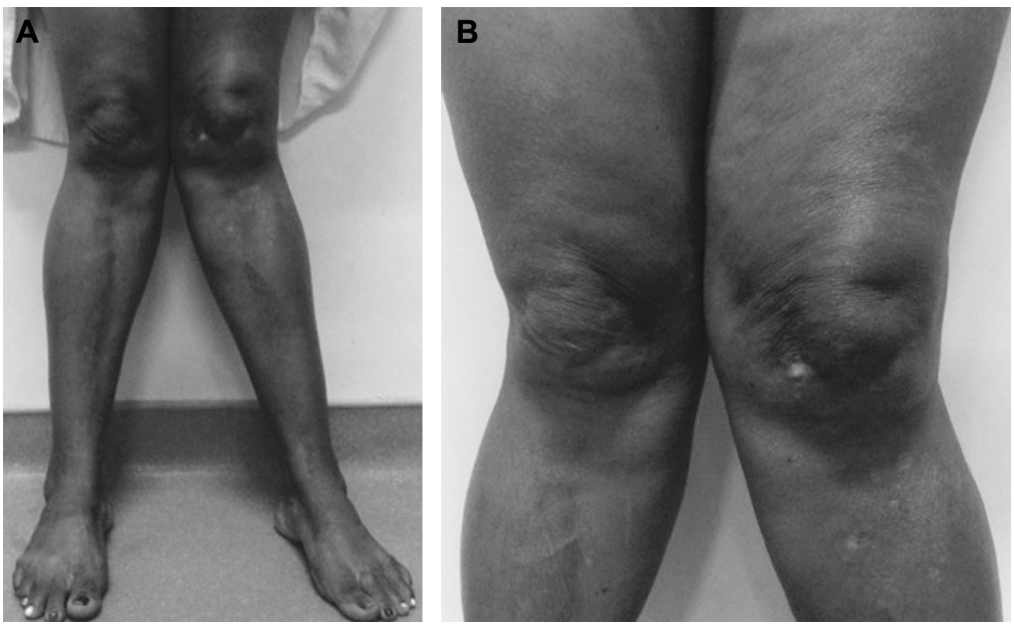

Fig. 1 – Photographs of the lower limbs of patient 1. (A) Genu valgum deformity. (B) Zoomed-in view. Greater detail of the bilateral widening of distal thighs and knees, accompanied by genu valgum.