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The Erlenmeyer Flask Deformity on Computed Tomography

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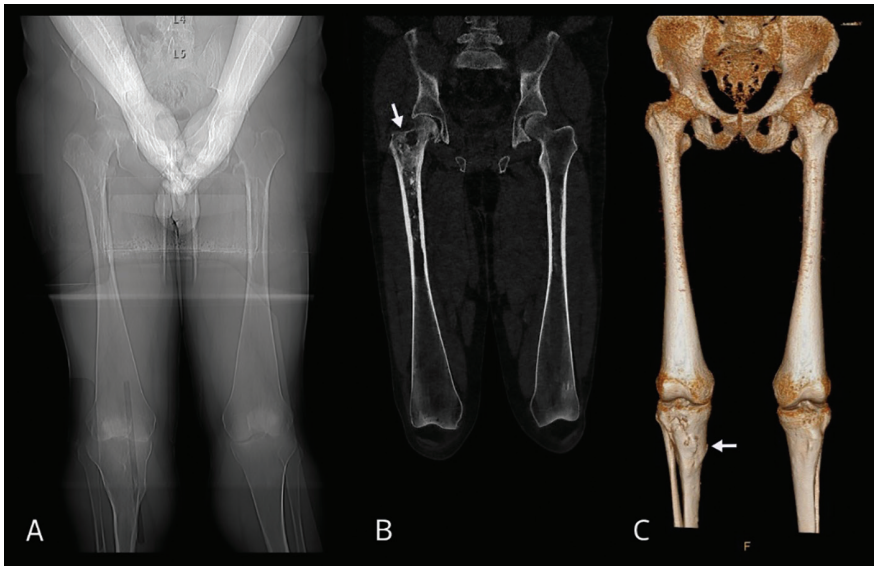

A 52-year-old male was referred to the Radiology Department for a scheduled skeletal survey using a whole-body low dose computed tomography (WBLD-CT) protocol (Panel 1 A & B). He was diagnosed with Gaucher disease (GD) type 1 in early childhood and was being treated by the Hematology Department with intravenous imiglucerase. He complained of chronic right hip pain, and mentioned a right tibial plateau fracture a few weeks prior as a result of a work accident, which was treated conservatively (Panel 1 C). WBLD-CT revealed a bone infarct involving the right femoral head and trochanters (assumed to be the cause of the chronic right hip pain) (Panel 1 B). Additionally, a widened distal diaphysis and diametaphyseal region on both femora was noted, with a thin cortex and straight borders, while the proximal diaphyses were narrow, resembling a flask, also known as metaphyseal flaring, or the Erlenmeyer flask deformity (EFD) (Panel 1 C). GD is a rare autosomal recessive disease responsible for accumulation of the glycolipid glucocerebroside within the lysosomes of the macrophages, principally in the bone marrow, spleen and liver, caused by the deficiency of glucocerebrosidase activity (1). EFD is the most common bone abnormality of GD and appears in 50-80% of adult cases (2, 3). Development occurs during puberty as a result of undertubulation. It may involve all tubular bones, predominantly the femur, and is typically asymptomatic with no proven predisposition to fragility fractures (1, 2). Besides GD, EFD manifests in numerous different diseases with improper osteoclast function, such as osteopetrosis, metaphyseal dysplasia (Pyle disease), Niemann-Pick disease, achondroplasia and thalassemia (1).

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Conflict of Interest: The authors declare that they have no conflict of interest.

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