



# Reumatología Clínica

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Images in clinical rheumatology

## A woman with joint pain and enlarge dupper limbs

Mujer con artralgias y aumento de tamaño de miembros superiores

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### Clinical case

We present a 32-year-old woman with a history of hypothyroidism, on replacement therapy, menstruating and with no children. She had no family history of osteoarticular disease. She told us that she had been diagnosed with muscular dystrophy when she was four years old in her country of origin. She had no medical notes or history for this, stating that this diagnosis had been given because she had been late in starting to walk and that no muscular biopsy or specific study had been carried out. She was consulting due to mechanical arthralgia of lengthy evolution, with no joint swelling, no morning stiffness; the patient said her upper limbs had enlarged over the years and the diameter of her thighs and legs had also increased. Originally she had gone to our hospital's Neurology Department, where analysis gave normal CPK, an electromyogram showed no evidence of myopathy and an MRI of the pelvic girdle did not reveal myositis; consequently, she was sent to our department for study. The examination highlighted the absence of synovitis, generally limited passive and active joint distances, normal muscular strength and an increase in the upper limb length. Her walk also drew attention with a certain oscillation of the pelvis, exaggerated lumbar lordosis and latero-lateral sway. The tendon reflexes were normal and there was no deformity in the toes or fingers. The simple radiological study carried out showed cortical thickening and sclerosis of the shafts of tubular bone. There was irregular and heterogeneous osteosclerosis, mottled areas of radiolucency with greater endosteal than periosteal involvement and narrowing of the spinal canals (Figure). The epiphyses were respected radiologically. Densitometry of femoral neck had a Z-score of +6.2.

### Discussion

The radiological findings suggest bone dysplasia. The number of bone dysplasias is high and there is no general agreement on their nomenclature and classification. However, due to an increase in human genome knowledge, exact genotyping is now possible when the genetic locus is known. In this case, diaphyseal involvement with respect to the epiphysis made us think that it was a dysplasia of the craniotubular hyperostosis group—the most common of which known by the eponym Camurati-Engelmann disease. It is a dysplasia with progressive bilateral and symmetrical thickening of the long bone shafts, which affects the metaphysis but not epiphysis as it progresses.<sup>1</sup> The skull can be affected but this was not the case with our patient. The presentation normally occurs in childhood with joint pain, muscle weakness, waddling gait and fatigue. Its inheritance pattern is autosomal dominant and it is due to the mutation of the transforming growth factor  $\beta 1$  (TGF $\beta 1$ ) gene, which has 10 known variants.<sup>2</sup> The genetic study of our patient confirmed the suspected diagnosis for c.652C→T, p.Arg218Cys mutation, a previously-described mutation for this disease.<sup>2</sup>

The patient was treated with NSAIDs based on the joint pain, given that the degree of pain did not seem to require greater analgesia. The drugs most commonly used in literature are glucocorticoids, which given their anti-inflammatory effect, seem to relieve the bone pain in these patients. Bisphosphonates, mainly intravenous pamidronate, have been used to treat this disease but with contradictory results.<sup>3,4</sup>

### Diagnosis

Diaphyseal dysplasia, Camurati-Engelmann disease type.

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**Figure.** Images of upper and lower limb shafts that show cortical thickening and sclerosis of tubular bones, as well as irregular, heterogeneous osteosclerosis with mottled areas of radiolucency.

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