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Melorheostosis: presentation of a clinical case

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Summary
Melorheostosis is a form of hyperostosis which affects both bone and the adjacent soft tissues. Its incidence is variable, although it is higher in the second and third decades of life due to the slowly progressive nature of the disease. It generally presents with pain which may cause significant functional limitation. We may be assisted in its diagnosis by its characteristic radiological image which resembles “wax melting down the side of a candle”. A case of melorheostosis is presented with clinical findings and radiological characteristics. The patient had previously been diagnosed with Paget’s disease of bone, so we proposed a differential diagnosis of this pathology.

Key words: melorheostosis, Paget’s disease of bone, differential diagnosis.
Introduction
The term melorheostosis is derived from the Greek suffixes melos (limbs), rhein (fluid/flow) and osteon (bone). This disease is also known as hyperostotic osteopathy or Leri and Joanny disease (the first to describe the disease in 1922). It is a rare form of hyperostosis which affects both the bone tissue and the adjacent soft tissues. It incidence is 0.9 cases per million population. In 50% of cases it is diagnosed before the age of 20, without predilection regarding the sex of the person. It consists of a benign process without associated mortality, but which generates functional limitations. Its etiology and etiopathogenesis are unknown. Genetic alterations associated with the disease have recently been described. Any bone may be compromised, with the lower limbs being the most affected. The diagnosis is usually carried out through diagnostic imaging techniques, notable among which is simple radiography. With this we may observe an image which resembles “wax dripping down the side of a candle”, a sign which gives us a secure diagnosis in most cases. The treatment is mainly symptomatic, and only occasionally requires recourse to surgery.

Clinical Case
A male patient 36 years of age referred in 2009 for a check up to his bone metabolism unit (BMU) and diagnosed in a private bone disease clinic with monostotic Paget’s disease of bone in the left radius. The patient brought with him a biopsy and risedronate carried out in 2007. He was treated with risedronate at a dose used for osteoporosis. On confirmation of a good level of P1NP (amino-terminal propeptide of procollagen type 1), the same year, 2009, the risedronate was withdrawn. From then, and up to November 2014 he was periodically checked for the blood level of markers for bone remodelling, with a new cycle of risedronate at a low dose indicated for him for a few months until the levels increased.

Also in this period a gammagraphy was performed without there being seen significant alterations from the earlier measurements. On administering the treatment the patient’s local pain improved, but in November 2014 he spontaneously attended the clinic with his arm in a sling saying that two days before he had suffered an accidental fall and was experiencing intense pain in the left carpal region. In a physical examination a haematoma was found in the back of the hand and the distal third of the forearm, oedema, effacement of the tendinous sulci of the back of the hand, functional weakness in the wrist and a local increase in temperature. With the clinical suspicion of fracture, he was referred from the bone metabolism unit to the accident and emergency department where X-rays were carried out. He was discharged with the diagnosis of contusion there being no evidence of fracture, but a large hyperostotic lesion was observed (Figure 1) in the simple X-rays, for which reason the patient again attended the BMU. In spite of a fracture not being seen in the X-rays, but there being clinical evidence, a computerised axial tomography (CAT) was requested urgently, and a nuclear magnetic resonance (NMR) scan for a deferred study of the hyperostotic lesion. In the CAT scan a fracture was seen in the hamate and trapezoid bones. The limb was immobilised with a posterior ferrule. Once the traumatological emergency was resolved, in the following days the radiological lesion of the radius was evaluated with the so called sign of “dripping melted wax” being identified. Given the possibility of melorheostosis, this option was suggested to the radiology and nuclear medicine service for their consideration. In the end, it was accepted as an alternative diagnosis to Paget’s disease of bone, their gammagraphy being indistinguishable. Once the new diagnosis of melorheostosis was confirmed and agreed we resumed the anamnesis, which, notably, recounted a fall at the age of 14 when practicing sport. The patient said that that he had suffered intense pain in the radius, but that he did not attend any health centre and hid it from his parents, having had since then some deformity. The pain subsided some weeks after the fall. We deduced that the patient had fractured his radius, and by neither immobilising nor reducing the fracture he was left with this deformity which can be seen in Figure 1, but which bears no direct relation with hyperostosis. Besides the striking central image, there are other areas of hyperostosis in the interior of the distal end of the radius, and in the proximal third.

Discussion
The exposition and development of the case presented invites a number of points of reflection. Firstly: should all diagnoses be called into question even if they are properly documented? In the case we are dealing with “everything had already been done”: the X-rays, although we never saw them, only the reports; also the pathological anatomy report following a biopsy, and the report of a gammagraphy, which could be viewed, having been stored in the hospital in the patient’s clinical record.

The second point of reflection is related to the trauma and raises the issue of the validity of clinical data as against complementary examination. Although the carpal fractures were not observed in the simple X-ray, the clinical evidence and our insistence led us to request an urgent CAT scan.

With regard to melorheostosis, we present this case because of the infrequency of the disease itself and the doubts which may be raised when trying to make a correct differential diagnosis with other pathologies.

Generally, when it affects a long bone, we can in practice obtain a diagnosis using a simple X-ray with the characteristic image of “dripping melted wax flowing down the side of a candle”. However, on many occasions we need a bone biopsy or bone gammagraphy to discount pathologies which affect bone metabolism. The initial diagnosis was not totally uncertain given that in Paget’s disease,
in response to bone resorption, there is an increase in bone formation, resulting in an increase in the thickness of some trabeculae and an irregular hypertrophy of the trabecular bone. As a consequence, the bone marrow is infiltrated with an excess of fibrous connective tissue and of blood vessels which lead to hypervascularisation, findings which are compatible with melorheostosis where the cortical hyperostosis causes thickening and trabecular bone and vascular increase. It should be borne in mind that in melorheostosis the microscopic appearance is not always the same, since it depends on the point in time at which the sample is obtained, similarly to Paget’s disease of bone. However, in order to carry out a differential diagnosis with the latter condition it is noteworthy that in melorheostosis, as well as presenting intramembranous ossification with the increase in the activity of the osteoblasts, there are certain consistent alterations such as an irregular diameter of the Haversian canals and an irregular lamellar pattern in the trabecular area, anatomopathological data which facilitate a definitive diagnosis. In relation to the gammagraphy, in Paget’s disease there is an increased radiopharmacological capture which may give us images similar to melorheostosis. This leads one to suppose that in melorheostosis there is an increase in bone metabolism which translates into an increased trace due to the presence of immature collagen and changes in vascular permeability. In addition, the gammagraphy also allows the differential diagnosis of melorheostosis with other diseases which develop with hyperostotic lesions such as osteopoikilosis and striated osteopathy, two conditions in which there is no gammagraphy capture. Lastly, the lightest forms of melorheostosis present more difficulties in differential diagnosis with periosteal osteosarcoma or myositis ossificans.

In the case we present, a credible documented prior diagnosis, a conservative attitude on our part in preventing exposure to ionising radiation and the superimposition of the data of two different diseases, meant that for several years the patient had an erroneous diagnosis, until a fortuitous trauma and a new X-ray invited a re-evaluation of the case.

**Bibliography**