RESEARCH PAPER | Medical Science | Volume : 5 | Issue : 3 | March 2015 | ISSN - 2249-555X

Melorheostosis With Unusual Paraarticular Ossification and Long-Term Follow Up – A Case Report

KEYWORDS: melorheostosis Léri

Anastasika Poposka
Clinic for Orthopedic Surgery University Clinical Center "Mother Teresa", Skopje, Macedonia

Milan Samardziski
Clinic for Orthopedic Surgery University Clinical Center "Mother Teresa", Skopje, Macedonia

Roza Dzoleva-Tolevska
Clinic for Orthopedic Surgery University Clinical Center "Mother Teresa", Skopje, Macedonia

Nenad Atanasov
Clinic for Orthopedic Surgery University Clinical Center "Mother Teresa", Skopje, Macedonia

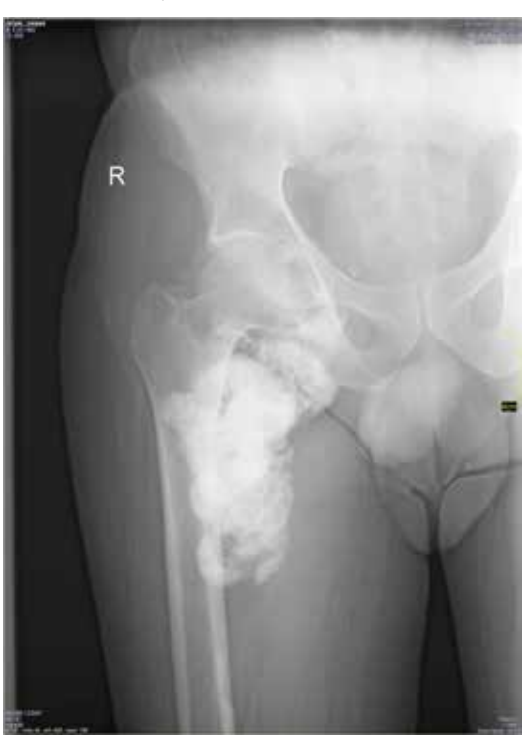
ABSTRACT *Melorheostosis is a rare bone disease characterized by a "flowing" hyperostosis of the cortex, first described in 1922 by Léri and Joanny. It frequently involves a single limb characterized by hyperostotic linear bone densities. The radiographic appearance has been linked to "dripping wax down one side of a candle". It has been suspected that melorheostosis is due to a loss-of-function mutation in LEMD3 gene (also called MAN1), which encodes an inner nuclear membrane protein. However, the etiology of melorheostosis remains unknown.*

We report a case of a 72-year-old male patient, with unusual for the desease paraarticular ossification, whose left leg was affected by melorheostosis more than 37 years ago.

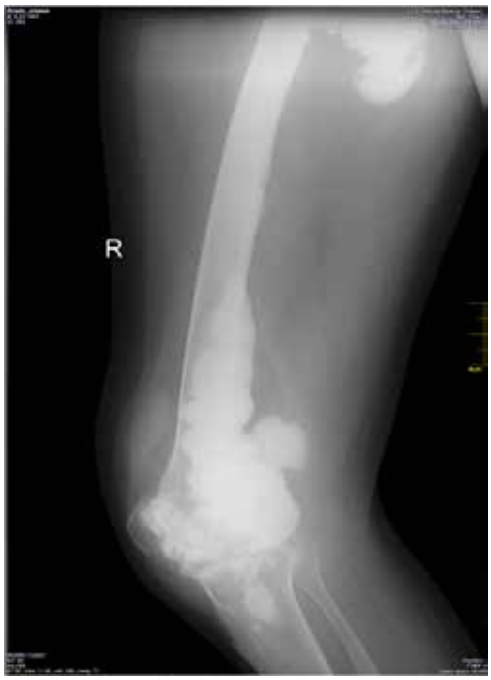
Introduction: Melorheostosis is a rare bone disease characterized by a "flowing" hyperostosis of the cortex, first described in 1922 by Léri and Joanny. It frequently involves a single limb characterized by hyperostotic linear bone densities. The radiographic appearance has been linked to "dripping wax down one side of a candle". It has been suspected that melorheostosis is due to a loss-of-function mutation in LEMD3 gene (also called MAN1), which encodes an inner nuclear membrane protein. However, the etiology of melorheostosis up to date remains unknown.

Case Report: We present a case of a 72-year-old male patient, diagnosed with single extremity (left leg) localized melorheostosis and long term follow-up. He presented with chronic, blunt left leg pain, restricted hip and knee motion and left foot deformity with skin callosity. The extent and severity of symptoms on the left leg varied depending on his professional or daily activities. Left groin and left inguinal pain was often present. He also complained on a very slow growing solid tumor in his medial femoral region and tumor on the first left metatarsal, resembling atypical "hallux valgus" deformity. The tumor in the proximal part of the femur caused lack of flexion and pain in the left hip. Sometimes he had pain in his left knee and he couldn't afford to kneel for longer time. Deformity of his foot was accompanied with reactive skin callosity of the sole and pain increased after prolonged walking or standing. In the history of the illness, the patient mentioned that more than 37 years ago he was diagnosed with "some rare chronic and proliferative benign disease". Having in mind that he had a mild pain (he had occasionally used pain killers or non-steroid antirheumatic medications), he has not taken into consideration the suggestions to be surgically treated.

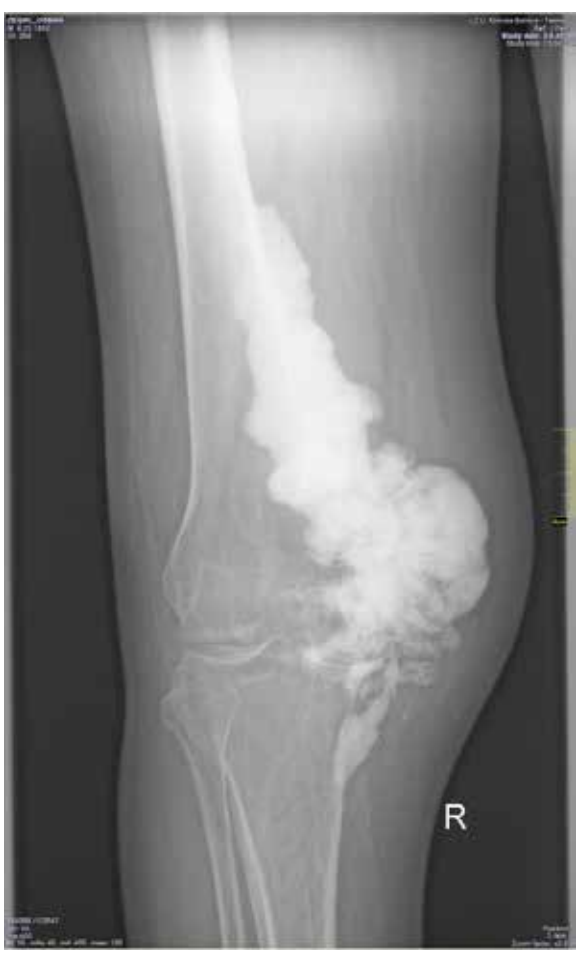
The x-ray images showed a large, abundant, para-articular ossification of the left hip. More typical hyperostosis with para-articular ossification is present on distal part of the medial cortex of the femur (Figure 1a, 1b). Medial cortex of the tibia and medial side ray of the foot bones are hyperostotic. Abundant para-articular ossification medial to the first metatarsophalangeal joint is formed. The hyperostosis resembles atypical "halux valgus" deformity. The characteristic radiographic features consist of irregular hyperostotic changes of the cortex, along the medial side of the bone, resembling melted wax flowing down one side of a candle. The paraarticular ossifications are unusual clinical and radiographic features for the desease.



Volume : 5 | Issue : 3 | March 2015 | ISSN - 2249-555X



a b



c d

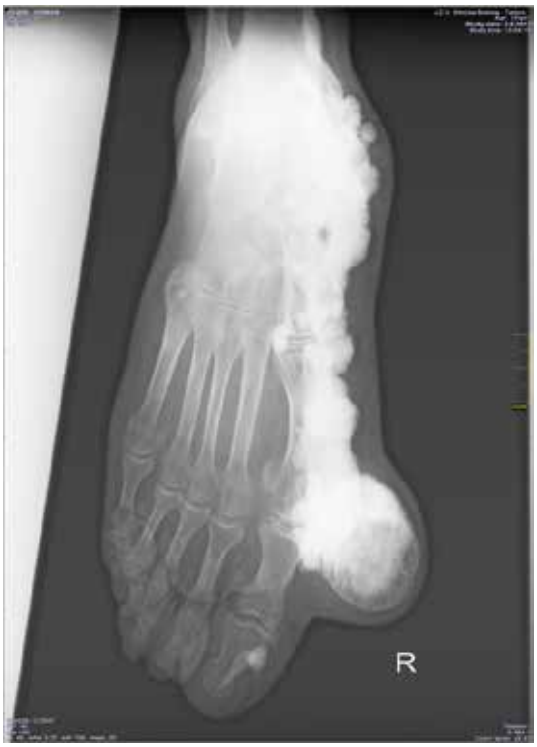


Figure 1. a) X-ray of proximal femur, with abundant parosteal ossification resembling solid tumor, unusual for melerhostosis, b) Lateral view of the femur presenting massive para-articular ossification in the proximal and distal meta-diaphyseal parts, **c)** X-ray of the distal tibia with "dripping candle wax" medial cortex of the bone, **e)** Frontal x-ray of the foot with ossification on the whole medial side (with large para-articular ossification of the first metatarsal bone) resembling atypical "hallux valgus" deformity.

The patient was offered to have para-articular ossifications of the left hip and the first metatarsophalangeal joint of the left foot to be surgically treated, but he declined such procedures. The well-fitted custom-made orthopedic shoes helped him to feel comfortable in everyday activities.

Discussion: Melorheostosis is rare dysplasia characterized by a "flowing" hyperostosis of the cortex, first described in 1922 by Léri and Joanny. [(1)] Patients present at of any age, and both sexes are affected equally. Onset is usually insidious, with deformity of the extremity, pain, limb stiffness and limitation of motion in the joints first manifesting in late childhood or early adolescence and progressing into adult life. [(2)] The characteristic radiographic appearance consists of irregular hyperostotic changes of the cortex, generally on one side of the bone, resembling melted wax drippingdown one side of a candle. This appearance gave the anomaly its name, which is taken from the Greek words for limb (melos) and flow (rhein).[(3)] There is usually a distinct demarcation between the affected and normal bone. Dense, sclerotic linear areas are seen mainly in the cortex but also extending into the cancellous bone. Melorheostosis affects mainly the long bones of the upper and lower limbs, but also the short bones of the hand and foot and, rarely, the axial skeleton. Sometimes it can produce *para-articular* ossifications, protruding into the vicinity of the bone, and associated with pain. [(4)] Melorheostosis

Volume : 5 | Issue : 3 | March 2015 | ISSN - 2249-555X

is a rare condition affecting the axial skeleton; it should be a definite consideration in the differential diagnosis of unilateral or segmental lesions of cortical hyperostosis in the spine because accurate detection can prevent an unwarranted biopsy. Although radiographic and bone scintigraphic appearance of melorheostosis has been well described (ie, undulating cortical thickening and marked increased uptake of radionuclide), CT and MR imaging helps confirm and accurately localize the zones of hyperostosis in the spine and provide assessment of the degrees of narrowing of the spinal canal and foramina. [(5, 6)] Further, even though the MR imaging appearance of soft tissue masses associated with melorheostosis is variable, mineralized and nonmineralized soft tissue abnormalities should be recognized as another manifestation of this disease. MR imaging aids in confirmation of the diagnosis and in the accurate detection and determination of the extent of soft tissue involvement. Melorheostosis can be complicated by various soft tissue changes. There have been cases with this disorder in the radius, clinically presented with symptoms of carpal tunnel syndrome. Treatment is case-dependent and usually is conservative. In rare cases biopsy must be done, due to osteogenic osteosarcoma; atypical form of infection or other unusual condition must be excluded. Sometimes, when joints are seriously affected or there is compression type neuralgia surgery must be considered. [(7)]

The etiology of melorheostosis is still unknown. It was not considered to be genetically induced condition. But in 2004 it has been reported that melorheostosis is due to a loss-of-function mutation in LEMD3 gene (also called MAN1), which encodes an inner nuclear membrane protein. It was reported that heterozygous, loss-of-function, germline mutations in the LEMD3 gene (LEMD3 or MAN1) might cause both osteopoikilosis and Buschke-Ollendorff syndrome. (8) In 2007 Mumm et al confirmed that osteopoikilosis and Buschke-Ollendorff individuals, including those with melorheostosis-like lesions, had heterozygous, deactivating, germline LEMD3 mutations. However, the etiology of melorheostosis still remains unknown. (9)

REFERENCE

1. Leri A, Joanny J. Une affection non décrite des os hyperostose "en coulée" sur toute la longeur d'un member ou "melorhéostose." Bull Memoires Soc Med Hopitaux Paris 1922;46:1141–5. | 2. Freyschmidt J. Melorheostosis: a review of 23 cases. Eur Radiol. 2001; 11(3): 474-9. | 3. Greenspan A, Azouz EM. Bone dysplasia series. Melorheostosis: review and update. Can Assoc Radiol J. 1999; 50(5):324-30. | 4. Dissing I, Zafirovski G. Para-articular ossification associated with melorheostosis Leri. Acta Orthp Scand; 1979, 50:717-9. | 5. Michiels I, Schaub T, Scheinzabach M. [Article in German] Melorheostosis, osteopoikilosis and ostiopathia striata. Their clinical significance and the value of scintigraphy in the differential diagnosis. Beitr Orthop Traumatol. 1990; 37(6):317-30. | 6. Motimayaa A.M, Meyersb S.P. Melorheostosis Involving the Cervical and Upper Thoracic Spine: Radiographic, CT, and MR Imaging Findings. AJNR, 2006; 27: 1198-1200. | 7. De Vos J, Mulliez A, De Loore G. Mélorhéostose de l'humérus : une affection rare dans le diagnostic différentiel du syndrome du canal carpien, Chirurgie de la Main 2010; 29(2)125–7. | 8. Hellemans J, Preobrazhenska O, et all. Loss-of-function mutations in LEMD3 result in osteopoikilosis, Buschke-Ollendorff syndrome and melorheostosis. Nature Genetics. 2004; 36: 1213-8. | 9. Mumm S, Wenkert D, Zhang X, McAlister WH, Mier RJ, Whyte MP. Deactivating germline mutations in LEMD3 cause osteopoikilosis and Buschke-Ollendorff syndrome, but not sporadic melorheostosis. J Bone Miner Res. 2007; 22(2):243-50. |