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A 25 Years Old Female with Meloheostosis: A Case Report

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ABSTRACT

Background: Melorheostosis is a rare sclerosing bone dysplasia characterized by its classic radiographic feature of flowing/dripping candle wax. It usually affects one limb, more often the lower extremity, and rarely the axial skeleton. The etiology remains unknown and various theories have been proposed to explain the pathogenesis of this disease. It often has an insidious onset in early adult life. Symptoms include pain, oedema, and limitation of joint movement.
Case presentation: A patient aged 25 year old male complaint of pain on the left lower extremity for the last 3 years. There is muscle atrophy at left extremity, bowing of the thigh, awelling at the foot. There is palpable mass at the left bottom, solid consistency and fixed. There are also limitation ROM of hip, knee and ankle. From the radiography examination showed inhomogen thickening of the periosteal cortical and endosteal, dripping candle wax appearance, and sclerotic lesion at soft tissue.
Conculsion: X-ray is a sufficient method for diagnosis of melorheostosis.Other imaging techniques are essential for decision about therapeutic intervention (CT or MRI). Laboratory findings are usually in physiological range. Symptomatic therapy proved to be sufficient in subjective symptoms management.

Keywords: Melorheostosis, sclerosing bone dysplasia, dripping candle wax

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BACKGROUND

Melorheostosis is a rare bone disease. Only about 300 cases have been reported world-wide. The etiology is still unknown. It is a developmental anomaly of bone formation with the evidence of inheritance. The small part of patients have mutations in LEMD3 group, but this mutations was not present in most of the cases (Vaclav et al, 2015).

Melorheostosis is a rare, non-familial, benign, sclerosing dysplasia affecting the skeleton and adjacent soft tissues. L'eri and Joanny gave the initial description of this disease in 1922. The etymology of this bizarre idiopathic disorder is derived from its pathognomonic roentgenographic appearance resembling "flow-ing candle wax" [melos = limb; rhein = flow; osteon = bone]. Patients usually present in late adolescence or early adulthood. The onset is insidious with a chronic progressive course. There is no sex or familial predilection. The usual presenting complaints are pain, swelling, deformity, and joint stiffness. The exact incidence ofthis unique, yet, uncommon disease involving the foot and ankle is not known (Dhillon, 2017).

We report one case of melorheostosis.

Case presentation

A 25 year old male complaint of pain on the left lower extremity for the last 3 years. Patien feel swelling on his foot since he was in the kindegarten, then when he was in primary school he feel his knee become stiffed and when college he feel a lump on his left hip and feel pain when he was sitting. This complain was getting worse until last 3 years. There is no history of trauma, no night pain, there is loss of body weight for the last 3 years, and loss of appetite. Patient has consulted to some hospital, and get analgetic treatment.

In the physical examination of the left lower extremity, there is muscle atrophy, bowing of the thigh, swelling at the foot. There is a palpable mass at left bottom region sized 8 x 5 cm, fixed, solid consistency and tenderness (+). ROM of hip, knee and ankle limited due to pain.

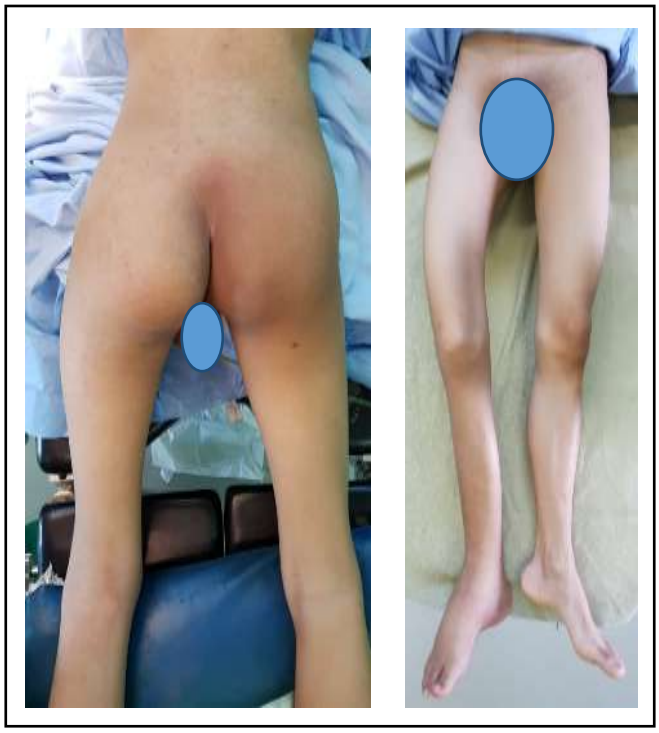

Figure 1. Physical finding of the patient

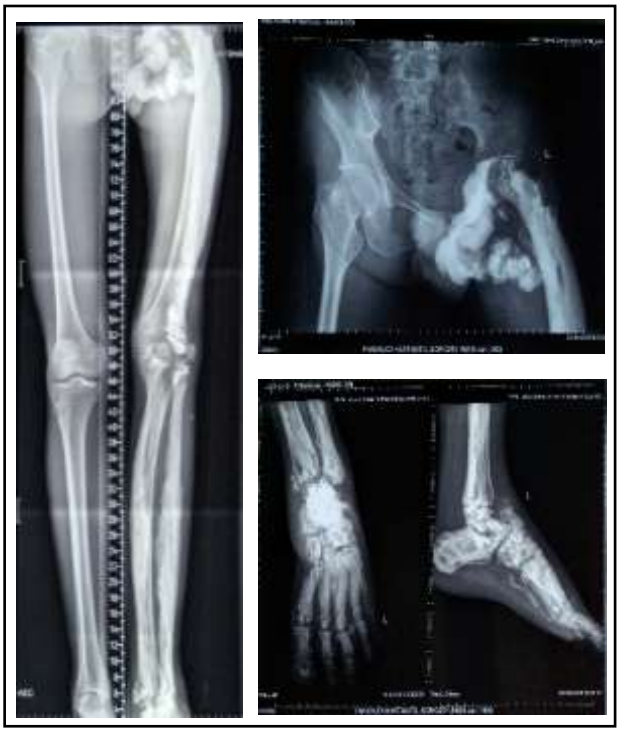

Figure 2. Radiological examination

From the radiography examination of the left lower extremity showed in homogenous thickening of the periosteal cortical and endosteal, dripping candle wax appearance and sclerotic lesion at soft tissue (Figures 2 and 3). Patient we diagnosed with melorheostisis of the left lower extremity then we perform excision of the mass at the hip that causes pain.

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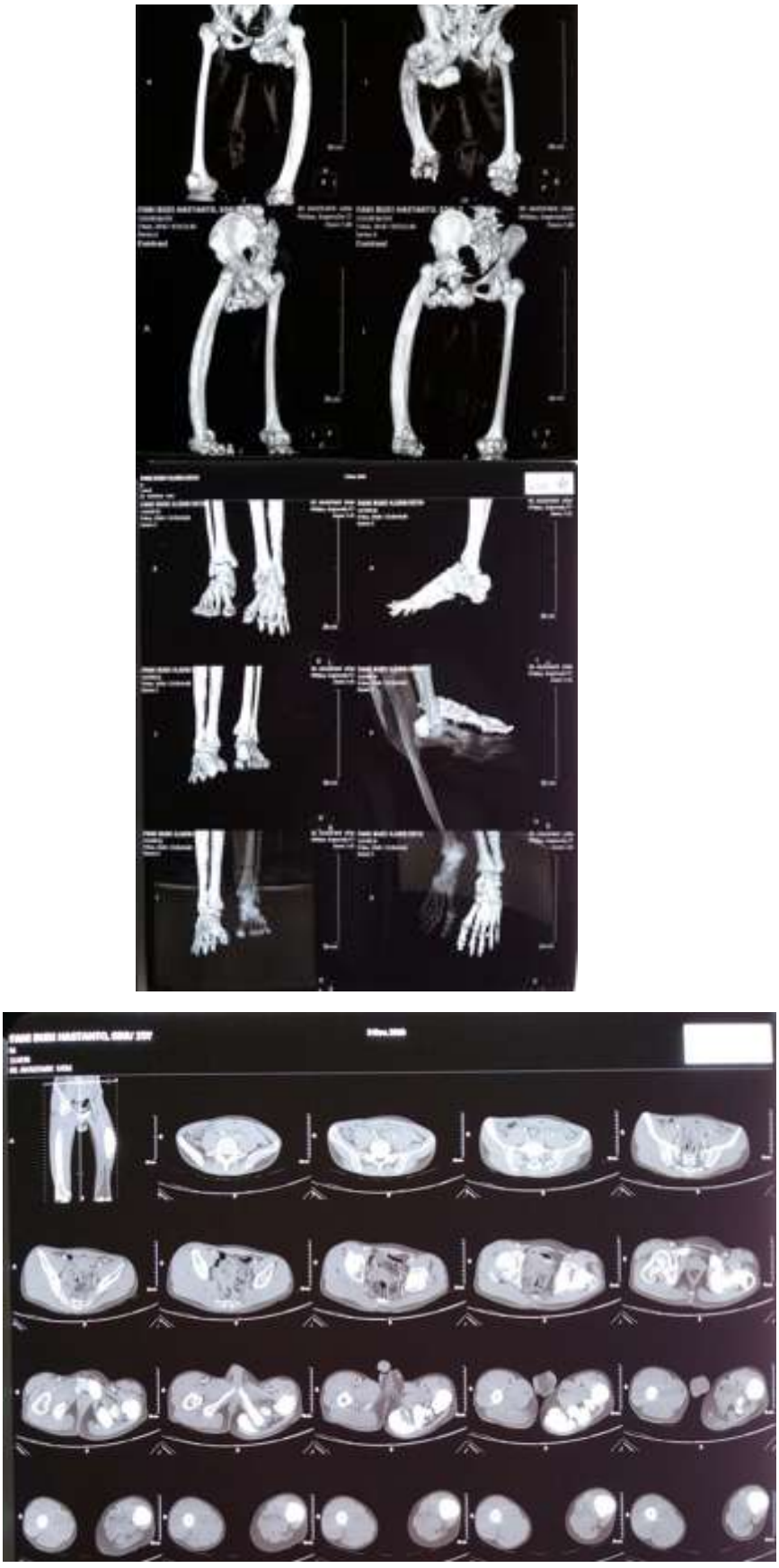

Figure 3. CT scan of lower extremity

After we diagnosed patient with melorheostosis of the leftlower extremity, because patient complaint of pain at ischium area, we perform excision at ischium region with prosedure:

1. Patien in prone position with general anesthesia
2. Incision posterior approach to ischium
3. Identify sciatic nerve, set aside
4. Identify tumor mass and excision the prominence tumor with oscillating saw

After that we examine tumour sample for patological finding, and we get the normal osteosit without any abnormal cell.

Figure 4. Mass at ischium region during operation

Figure 5. Mass after excision at ischium region

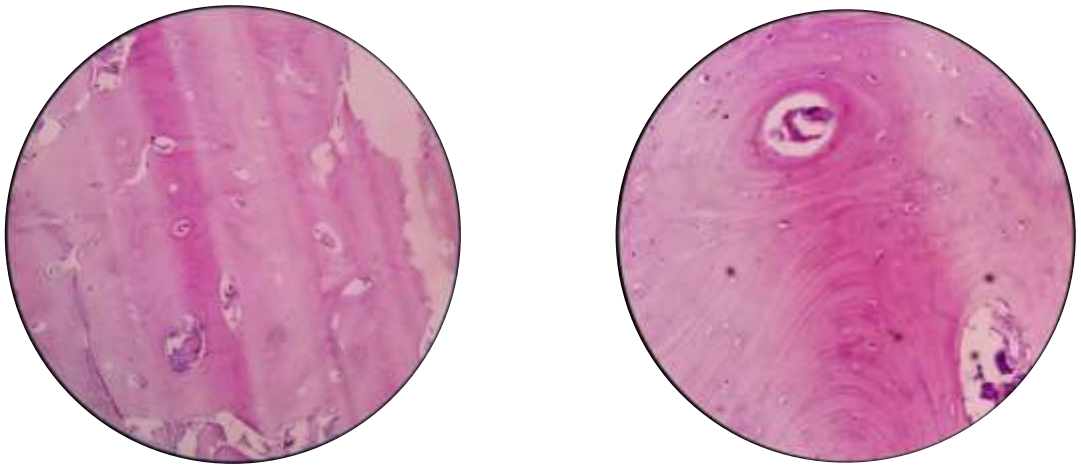

Figure 6. Histopatology finding of sample show nonspecific, only reveal of normal cell without any malignancy sign

DISCUSSION

Melorheostosis, also known as Leri disease, belongs to the class of sclerosing bone dysplasia with an incidence of less than one case per million. The condition is thought to be caused by a defect in intra membranous and endochondrial bone formation leading toirregular thickening of cortical bone (cortical hyperostosis) (Agarwal et al. 2017).

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It may affect a single bone (monostotic), a single limb (monomelic), or multiple bones (polyostotic). The monomelic variant is more common, with the lower extremity affected twice as often as the upper extremity (Dhillon, 2017).

In our patient it was monomelic presentation from hip to the foot. Gabriel et al. (2016) also revealed in his study that monomielic is typical in their population of study

Pain is by far the most common reason for physician referral among patients with melorheostosis. Our patient come to our wih chief complaint of pain. Sith et al. study also show that pain is the most reason for physician with melorheostosis (Agarwal et al. 2017).

Skin and subcutaneous tissue involvement can result in fibrosis and joint contractures leading to deformity and limb-length discrepancy (Agarwal et al. 2017). In our patient, he was complain in joint stiffed from hip joint, knee joint and ankle joint.

According to Freyschmidt (2001), The diagnosis of melorheostosis was made on the basis of clinical and radiologic pattern.

Diagnostic criteria for establishing the diagnosis "melorheostosis" were one of the following:

1. Typical radiologic pattern with the following or dripping candle wax phenolmenon on the inner and/or outer surface of the involved bone(s)
2. In the case of an osteoma-like hyperostosis with orientation in the long axis of the involved bone following subset of criteria:
   a. The lesion diameter would have to be more than 5 cm or more
   b. More than one bone would have to be involved
   c. Only eccentrically located lesions at or in the bone were eligible
   d. If only one bone was involved, other signs, such as circum scribed sclera derma or subcutaneous fibrosis above the involved skeleton, were mandatory to establish the diagnosis
3. Radiographic pattern with long and dense hyperostotic striations near the inner side of the cortex in two or more bones, but unilateral in contrast to the typical pattern of genuine osteopathi astriata
4. In the case of myosit ossification neuro pathic like pattern following subset of criteria:
   a. Ossifications adjacent to a joint in two or more unilateral regions with or without intra osseous hyperostosis
   b. In contrast to classic myositis ossification, the ossifications would have to be nodularly arranged and not appear as structured lamellar bone
   c. Exclusion criteria: history of direct trauma to the region of interest and/or neurologic deficit (Freysshmidt. 2001).

The etiology of melorheostosis remains unknown. However, one possible etiology of melorheostosis is a loss of function mutation in the LEMD3 gene [12q12–12q14.3], a protein of the inner nuclear membrane involved in bone morphogenic protein and tumor growth factor-β signalling. There have been various theories proposed to explain the pathogenesis of this disease such as a developmental disorder theory, ischemic theory, telangiectatic theory, and infective theory. Currently, there are two major hypotheses in existence. In 1979, Murray and McCredie correlated melorheostosis with sclerotomes, hypothesizing that melorheostosis might be the result of a segmental sensory lesion due to a specific infection, insult, or injury to segments of the neural crest during embryogenesis, which partially explains the peculiar mono-

melic involvement of melorheostosis. In 1995, Fryns proposed mosaicism to explain the sporadic occurrence of dysplasia which suggests that the asymmetric involvement of skeletal structures and concomitant vascular and hamartomatous changes in the overlying soft tissues result from an early post-zygotic mutation of the mesenchyme which explains why the extent of involvement is so variable and why the incidence ratio in both genders is equal (Agarwal et al., 2017).

Histopathology reveals nonspecific periosteal bone formation with thickened trabeculae and fibrotic changes in the marrow spaces. Our result for histopathology also normal, didn't show any malignancy (Agarwal et al., 2017).

Laboratory findings for serum calcium, phosphorus, erythrocyte sedimentation rate, and alkaline phosphatase levels have been reported to be within normal limits. Our results for alkaline phosphatase levels was normal, but higher level for erythrocyte sedimentation rate.

Flowing cortical hyperostosis along one side of the shaft of the long bone resembling "melting wax flowing down the side of a candle" is the characteristic radiographic appearance of melorheostosis. The lesions are typically eccentrically placed with no evidence of bony destruction. There is usually a distinct demarcation between the affected and normal bone. Computed tomography [CT] scans and three-dimensional reconstruction show candle wax-like or massive, rough hyperostosis around cortical and cancellous bones, accompanied by deformity of the bones, narrowing of the medullary cavity (Agarwal et al., 2017). Our patient had radiologicaly typical for this.

Various conservative or surgical methods have been practiced in treating the pain and deformities associated with melorheostosis. Conservative therapies include oral medications such as bisphosphonates, NSAIDs, and nifedipine. Surgical procedures consist of soft-tissue procedures such as tendon lengthening, excision of fibrous and osseous tissue, fasciotomy, capsuletomy, osteotomies and excision of hyperostoses, arthrodesis, and amputation. We perform osteotomy for the tumor at ischial that cause pain complain from patient, after osteotomy patient feel better (Agarwal et al., 2017)

This patient had localized melorheostosis at left lower extremity which was treated with simple analgesics in several hospitals without relief. We treated this patient with osteotomy of the tumor mass at left ischium that caused persistent pain for patient especially whule he was sitting. After the surgery patient feel better and feel more enjoy while sit.

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