CASE REPORT

Melorheostosis at Left Lower Limb of a 22 Years Old Female: A Rare Case

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ABSTRACT

Sclerosing bone dysplasias are skeletal abnormalities of varying severity with a wide range of radiologic, clinical, and genetic features. It is divided into Hereditary and Non-hereditary. Types of hereditary sclerosing bone dysplasia include osteopetrosis, osteopoikilosis, osteopathia striata, progressive diaphyseal dysplasia, hereditary multiple diaphyseal sclerosis, hyperostosis corticalis generalisata. Nonhereditary dysplasias include intramedullary osteosclerosis, melorheostosis, and overlap syndromes.[1] Our case report below describe about Melorheostosis of the lower limb.

Key words: Melorheostosis, Sclerosing bone dysplasias, skeletal abnormalities

CASE HISTORY

We report a 22-years-old female from makassar, south sulawesi, Indonesia, presented with pain at her left lower limb, and limping. Pain presented since 10 years ago, not related to her activity, never seek medical advice before. She also felt limping when walking that gradually progressed until she was 20 years old. There was no relevant family history, there is no history of trauma, and no history of fever. Patient has graduated from senior high school, has not married and has not had any job. Patient still able to do daily activity living.

On physical examination, the patient had hard swelling of her left thigh and leg. The swelling was bony hard, non tender, non hyperpigmented, skin temperature was normal and leg length discrepancy was 6 cm.

Range of Motion (ROM) of hip and ankle joint are within normal limit. ROM of left knee active and passively was 0^0 - 90^0 of flexion. No other abnormality was detected in the right lower limb or her upper limbs.

Plain radiographs showed extensive, dense, undulating or irregular cortical hyperostosis, resembling candle wax, extending along the length of bone, resulting in deformity of the bone and narrowing of the medullary cavities.

Laboratory findings like serum calcium, phosphate, alkaline phosphatase concentration, markers of inflammation and other findngs were within normal limit.[4] Biopsy was not done since the clinical findings and X-rays were definitely diagnostic of melorheostosis.[4]

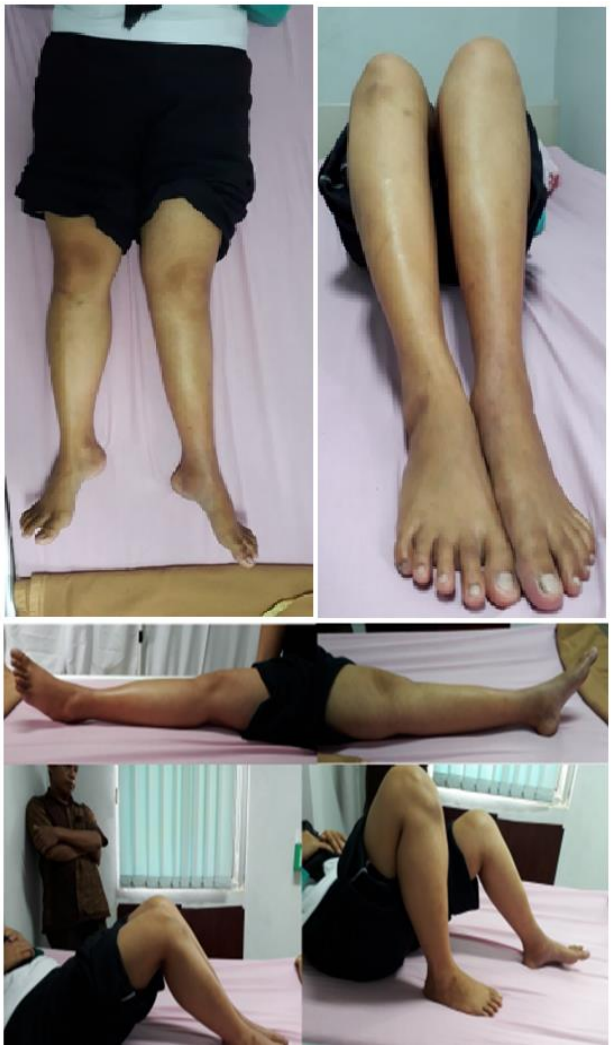

Fig. 1. Clinical findings (reproduced with the patient's permission)

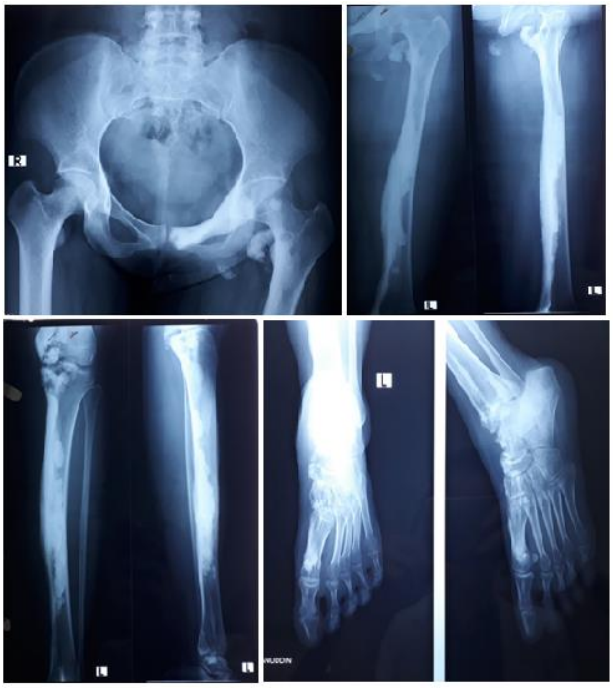

Fig. 2. Plain Radiographs of left lower limb (reproduced with the patient's permission)

Treatment is symptomatic based on the complaint of the patient. Our patient has been treated with nonsteroidal anti-inflammatory drug and physiotherapy with good symptomatic response. Patient still able to perfom her daily activity living and control routinely to our outpatient departement.

DISCUSSION

Melorhesotosis (synonyms Candle Bone Disease, Melting Wax Syndrome, Leri Disease), first described by Leri and Joanny in 1922 as "Hyperostose en Coutee", is a rare benign disorder of Sclerosing bony dysplasia, affecting more commonly appendicular skeleton than axial skeleton and also its adjacent soft tissue. Melorheostosis may present in a monostotic (affect one bone), polyostotic (affect many bones) or monomelic form (affect one limb). In appedicular skeleton cases, lower limb is more common than upper limb. Melorheostosis is derives from the Greek word, melos = limb, rhein = flow, osteon = bone; referring to the radiographic appearance that resemble wax flowing down one side of candle. Men and women are equally affected. This disorder begin in childhood with varies clinical manifestation from 3 years to 61 years.[2]

It may also present with pain, joint stiffness, contractures, limb-length discrepancy and hyperpigmentation of overlying skin and scleroderma. The exact cause of diseaseis remain unclear. There are several hypothesis are given. Murray and McCredie in 1978, hypothesis was embryonic infection of nerve rrot causes neural scarring and segmental bone sclerosis responsible for melorheostosis. The other hypothesis, one possible etiology of melorheostosis is loss of function mutation in the LEMD3 gene (12q12-12q14.3), a protein involved in bone morphogenic protein and tumor growth factor-β signaling.[3]

Melorheostosis mainly affects, the long bones of the upper and lower limbs, and also the short bones of the hand and foot, but rarely the axial skeleton. In this case, the disease affected the pelvis, long bone of femur and tibia, and also her left foot. And the patient still able to do daily activity living. [4]

Routine laboratory findings usually are normal. Histologic findings are usually nonspecific and

often show dense bone formation, a mixture of mature and immature bone elements. [4] Most patient receives nonoperative treatment. Surgical treatment is reserved for contractures and deformities.[5]

CONCLUSION

This is the first reported case in our region. The purpose of this case report is to describe the presentation and course of the disease. A comprehensive review of literature describing etiology, clinical aspects, diagnosis and treatment is included. Patients symptoms vary considerable in melorheostosis and consequently their treatment should be individualize

The diagnosis can be attained only by clinical and radiographic finding (dripping candle wax sign). All routine laboratory findings usually normal and the histological findings are nonspecific. Most patient receives nonoperative treatment. Surgical treatment is reserved for contractures and deformities. Our patient had been treated with oral analgetic, physiotherapy and proper education, which is given a good response. Patient still able to perfom her daily activity.

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