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Melorheostosis with Osteopoikilosis - A Rare Case Report

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Abstract:
Melorheostosis with osteopoikilosis is a rare sclerosing bone dysplasia, combining the clinical and radiological features of melorheostosis and osteopoikilosis ,that has been reported in some families with osteopoikilosis and that is characterized by a variable presentation of limb pain and deformities.
*Melorheostosis is a rare sclerosing bone dysplasia that affects both cortical bone and adjacent soft tissue structures in a sclerotomal distribution. Melorheostosis may present as mixed or atypical osseous involvement in addition to the classically described "**dripping candle wax"** appearance of hyperostosis.*
Osteopoikilosis (OPK) is a benign, rare, asymptomatic osteosclerotic bone dysplasia which is inherited as an autosomal dominant trait. It may develop during childhood and persists throughout life. Diagnosis is usually made incidentally according to radiographs. It may be confused with other conditions, such as osteoblastic metastases.
*As **osteopoikilosis** is a benign, usually asymptomatic condition, the clinical signs are primarily those of melorheostosis that can manifest with joint contractures, pain, stiffness, limited joint functions, limb-length discrepancy and deformity.*
We report a 11-year-old male who has an incidental diagnosis of melorheostosis with osteopoikilosis .

Keywords- *bone metastases, Dripping candle wax, hyperostosis, melorheostosis, Leri disease, osteopoikilosis, sclerosing bone dysplasia,Sclerotomal distribution*

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I. Introduction

The U.S. National Organization for Rare Diseases defines rare diseases as conditions affecting fewer than 200,000 individuals.

Sclerosing bone dysplasias are a group of rare skeletal abnormalities that are caused by various disruptions of the bone developmental pathway. Diagnosis and classification of sclerosing bone dysplasias is based on a combination of clinical, morphological, and radiological criteria. The radiological approach suggested by Greenspan classifies these disorders as a disturbance in endochondral or intramembranous bone formation or modelling, or both.

Melorheostosis (eponym: Léri's disease) is a very rare lesion affecting the both sexes; no familiar trend has been reported. Though the condition has not been observed in a child under the age of 3 years, there is strong presumptive evidence that is presented at birth.
The term "**melorheostosis**" is derived from the Greek "me- los," meaning limb, "rhein," meaning flowing, and "ostosis," meaning bone formation. Leri and Joanny have described this disease since 1922 for the first time and it is not currently known clearly. . This rare condition has an estimated prevalence of approximately 1 in 1,000,000 reported between 1950 and 1979.
Patient may complain of pain and of restricted movements of joints but the condition is often asymptomatic. Some cases are associated with skin lesion, such as scleroderma , and with the vascular anomalies; joint contractures may be found in some patients.
The condition is characterised by the presence ofdense irregular bone running down the cortex of the long bone. Both the internal and external aspect of the cortex may be affected. Dense areas tend to be over grown and bowing may result. Murray and McCredi (1979) have pointed out that the distribution of the new bone corresponds to be a sclerotome, the segmental root nerve innervations of a bone.

The new bone has been likened to molten wax running down the side of burning candle. The lesions tend to be segmental and unilateral , through both limbs may be affected.
Occasionally the condition is bilateral but never symmetrical. Some lesion are progressive. The lower limbs are most commonly affected. Premature epiphyseal fusion may result, so that anaffected limb may be larger or smaller than normal.
The skull, spine and ribs are rarely affected. Ectopic bone may be found in the soft tissue around the joints between the affected bones.

Osteopoikilosis (OPK) is a rare inherited condition of the bones, transmitted as an autosomal dominant trait characterized by numerous hyperostotic areas that tend to localize in periarticular osseous regions. It is characterized by symmetrically distributed numerous, small, well-defined, homogenous circular or ovoid radiodensities clustered in epiphysis and metaphysis of long bones in periarticular region, and in some cases diffusely present throughout axial and appendicular skeleton. There is no age and sex predilection. It is usually asymptomatic but rarely in 15-20% patients slight juxta-articular pain and joint effusions can be seen. These are incidental radiological findings in most of the cases, also sometimes confused as bony metastasis. There are no specific clinical features; histological features are similar to bony island and it may be associated with connective tissue disorders, synovial osteochondromatosis, and a rare bone condition melorheostosis.
Indeed, the precise boundary between melorheostosis and other sclerosing bone conditions (osteopoikilosis, osteopathia striata and various other causes of hyperostosis and osteosclerosis) is somewhat blurred . Many aspects of the condition are still unexplained or contentious.

II. Case Report

A 11-year-old male presented with trauma due to fall (2 years back).Patient had insidious onset of pain and swelling in the left knee since one and half years, with associated history of trauma 2 years back.
Initially, the patient was managed conservatively, but after 6 months the pain increased. No history of weight loss, anorexia or night sweats was present. He had no history of joint disease, and there was no significant family history.

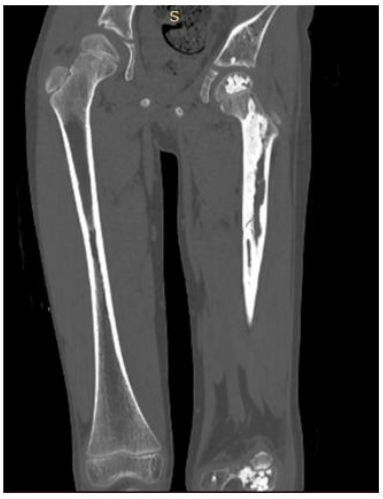
On general examination-
patient was conscious, co-operative, oriented to time, place and person; afebrile to touch , pulse rate of 80 beats/minute.

On local examination –
patient presented with swelling, tenderness and evidence of limb deformity (genu Valgum).

Laboratory evaluation - were performed
complete blood count, erythrocyte sedimentation rate, phosphate test, vitamin D, calcium levels, phosphorus, and C-reactive protein, which were all normal.

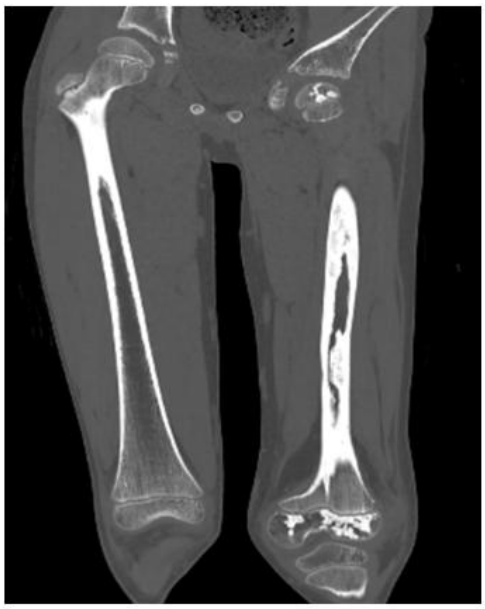
The patient then had a lower limb CT scan to assess.
Ct scan shows finding of Coronal images

FIG -1



Diffuse asymmetrical cortical thickening noted involving the left femur and the postero-inferior portion of left acetabulum with sub-periosteal and endosteal newbone formation with narrowing of the medullary cavity suggestive of Osteopoikilosis.

FIG -2

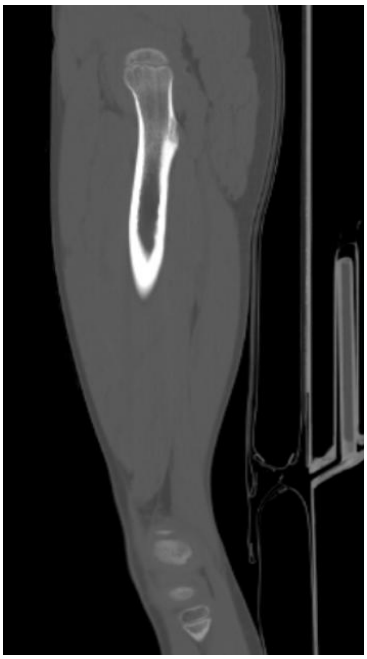


Multiple tiny subcentimeter sized bony island were noted involving the subarticularportions of the hip and head of femur - suggestive of Melorheostosis.

(FIG-1 and FIG-2)

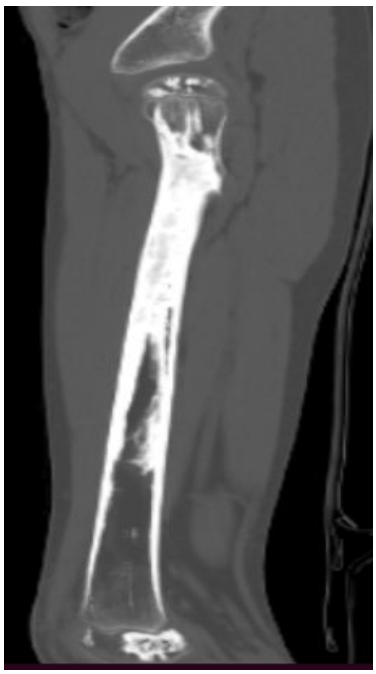
saggital images -

FIG -3



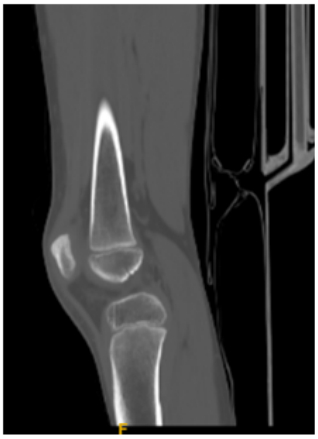
Right femur

FIG -4



left femur

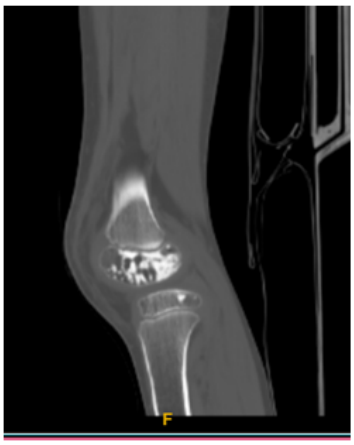
FIG -5



Right knee joint

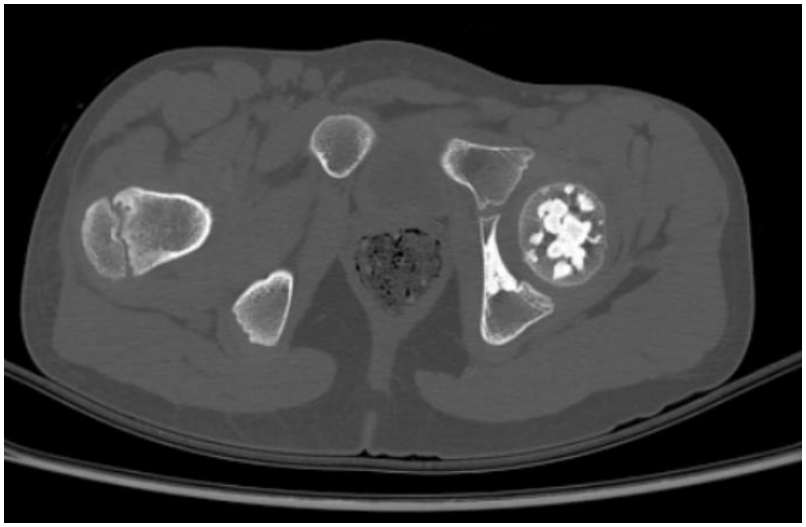
Multiple tiny subcentimeter sized bony island were noted involving the subarticular portions of the left knee joint (FIG – 6)

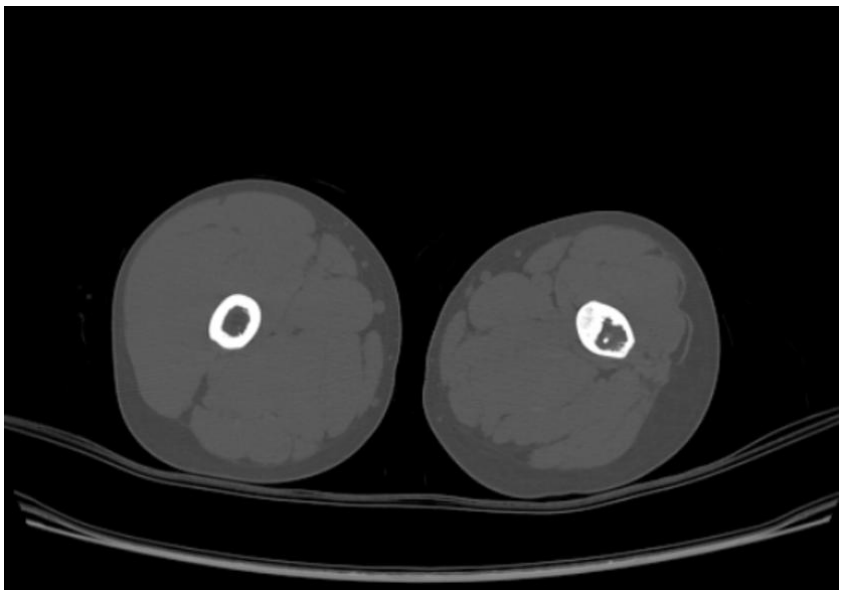
FIG -6



left knee joint

Axial images





Other findings:
Mild left hip and knee joint effusion was noted.
There was mild atrophy of the visualized muscles of the left thigh and hip region.

III. Conclusion:

Normally, osteopoikilosis and melorhosteosis occur separtarely, but this is a rare case in which both of them are seen simultaneously.

Ostepoikilosis is itself a rare inherited condition, transmitted by autosomal dominant trait and is associated with mixed sclerosing bone dysplasia, scleroderma and gunal-seber- basaran syndrome

Melorhostosis is a uncommon mesenchymal dysplasia manifestating in the regions of sclerosing bones. It is associated with scleroderma, vascular tumours and other tumours like osteosarcoma and malignant fibrous histiocytoma.

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