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Melorheostosis of small bones of hand – rare presentation of a still rare disorder – A case report.

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Abstract

Introduction: Melorheostosis is a rare benign mixed sclerosing disorder chara cterized by meso dermal dys plasia of bone that has a sclerotomal distribution. We report a case of a 18 year old female with Melorheostosis of second and third metacarpals and capitate bone.

Case report: A 18-year female is presented in this case report with a history of deformity over the dorsum of right hand for 13 months. Physical examination revealed a bony hard, non-tender swelling on the dorsum of her hand. Overlying skin and adjacent joints were unaffected. Plain radiographs and CT scan showed dense, extensive, irregular cortical hyper ostosis, like candle wax, extend ing along the length of second metacarpal and the capitate bone. All other routine investigations were within normal limits. Oral alendronate along with sym ptomatic management with NSAIDS was provided to the patient.

Conclusion: Melorheostosis should be considered in the differential diagnosis when bony tumors of the hand are encountered.

Keywords: bone dysplasia, dripping candle wax, hyper ostosis, Melorheostosis

Introduction

Melorheostosis was first described in 1992 by Leri and Joanny [1]. It is a rare benign mixed sclerosing disorder characterized by mesodermal dysplasia of bone that has a sclerotomal distribution. It predominantly affects intra membranous ossification [2]. The word Melorheostosis derives its origin from the Greek 'Melos' meaning limb, 'Rhein' meaning to flow and 'ostasis' meaning bone formation. It is due to its classical radio logical appea rance of dense, irregular and eccentric hyper ostosis, which looks like the dripping of wax from the side of a candle, although other various patterns also exist. No hereditary patterns have been noted. It is seen to equally affect either gender. The disease commonly presents in childhood and early adolescence but may be diagnosed at any age [3]. It is insidious in onset and gradually prog ressive. A waxing and waning course may also be seen with quiescent and active periods [4].

The diaphysis of long bones are usually affected, especially of the lower limbs. The short bones of the hand and foot are relatively less affected [5]. Involve ment of the axial skeleton is not commonly seen. Bone pain is more prominent if there is active sub periosteal bone formation. Other presentations may include joint stiffness, limb length discrepancy, ossification of adja cent soft tissue structure and progressive deformity [3].

Case report

Here we present a case of a 18 year old female who presented with the history of deformity over the dorsum of her right hand. The deformity was present since last 13 months. Deformity were insidious in onset and gradually progressive in nature. It was not associated with any pain. No relevant positive family history was present. No history of trauma to the affected part was reported.

Informed consent was obtained from the patient regard ing the publication of the case. On physical exam ination, the patient had a swelling/mass on the dorsum of her right hand (Fig. 1 and 2). It was bony hard and non-tender. Overlying skin was of normal temperature. No ab normal pigmentation, hair growth or skin texture was noted. Adjacent joints were normal and no form of contra ctures were seen. No other anomaly was detected in the remainder of the right upper limb, contralateral upper limb, and bilateral lower limbs. Plain radiographs of the hand showed dense, extensive, irregular cortical hyper ostosis, like candle wax, extending along the length of second metacarpal (Fig. 3). This resulted in the medul lary cavity narrowing and deformity of the bone. Plain radio graphs of the contralateral hand, chest, pelvis, skull, spine were normal. Non contrast CT scan of the right hand showed the involvement of the second, third meta carpal and capitate bone by diffuse cortical thickening of the bone with dripping wax appearance suggestive of Melorheostosis (Fig. 4). Laboratory investigations were ordered, like CBC, S. phosphorus, S. Calcium, ESR, CRP, S. ALP, S. LDH which were within normal limits.

The patient was symptomatically managed with oral NSAIDs, Calcium and vitamin D supplements. Tab alend ronate 70mg weekly was prescribed to her. Active physio therapy was administered.

The patient was followed up monthly up to duration of 12 months during which the deformity did not progress in size nor any signs of transformation were seen.

Discussion

Melorheostosis is a rare, chronic, benign sclerosing dysplasia of bone, which affects the younger population. Its incidence is reported to be 0.9 per million, with no sex predilection [6]. The disorder classically is non heredi tary, with unclear etiology. Various authors have proposed numerous somatic gene mutations as the possible cause [7, 8, 9, 10]. Appendicular skeleton is com monly affected with two types of presentation i.e. mono or polyostotic, with most of the cases limited to only one limb (mono melic). In polyostotic forms involving one limb, the affected bones are characteristically contiguous with sclerotomal distribution, crossing joint lines frequently. Long bone diaphysis, especially lower limbs are commonly affected. Short bones of foot and hand are com paratively less affected [5], making this case a rare presentation of a still rare disorder. Axial skeleton involvement is seen rarely. Pain is the most common presentation in the disease. A wide range of types of pain are present, including nociceptive, neuropathic or skeletal pain.

Plain radiographs have the characteristic ‘dripping candle wax’ appearance. CT and MRI suggest a sclerotic pattern with medullary space reduction and involvement of the soft tissue (if any). Bone scintigraphy shows increased uptake, differentiating it from other sclerosing disorders, like osteo pathia striata, and osteo poikilosis, which have

normal scinti graphic study. Histopathological study are not very specific and show increment in the formation of osteoid with trabeculae thickening and irregular lamellae. Many reports of tumors associated with Melorheostosis have been published including osteogenic tumors, malignant fibrous histiocytoma, and dermoid tumors [11, 12]. Management is mainly symptomatic with analgesics like NSAIDs for pain management, and physiotherapy. More recently, bis phos phonates containing nitrogen were found to be effective, although their long-term efficacy is still questionable [13]. Various types of surgeries like excision of the hyperostotic bone, tendon lengthening, sym pathe ctomy, and amputation, are indicated, which are highly personalized according to the part of the skeleton involved and patient profile [14]. But most of the surgeries require revision, indicative of the progressive nature of the disorder [5].

The prognosis of patients Is variable, depending on various factors like age, anatomic location of the lesion, soft tissue involvement, etc. Life span remains normal for these patients, but considerable morbidity maybe be present.

Conclusion

Melorheostosis is a very rare, sporadic, sclerotic bone dysplasia of unknown cause, diagnosed by chara cteristic radio logical findings.

Patients must be evaluated and treated based on a comprehensive, individual strategy, which involves non-surgical and surgical plan, since no definite standard of care is established.

Clinical message: This article has a clinical significance in identifying different patterns in which Melorheostosis can present to a clinician, and hence helps in prompt diagnosis and treatment of such cases.

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Legend Figures

Figure 1: Lateral view of hand showing the swelling on the dorsum of hand.

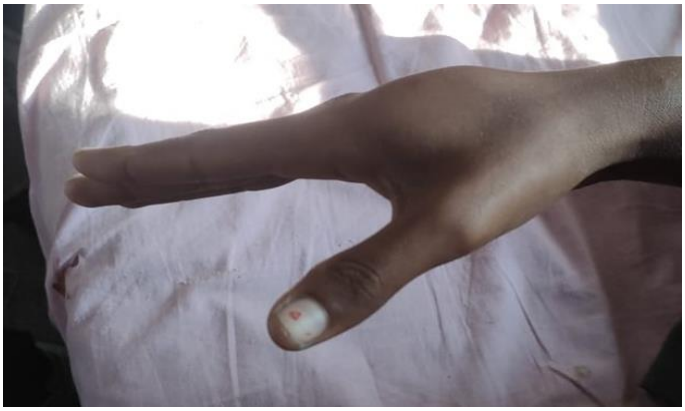

Figure 2: Clinical photo of the swelling on dorsum of hand.

Figure 3: Radio graph showing antero-posterior and oblique views of the affected hand. This shows dense, extensive, irregular cortical hyperostosis, like candle wax, extending along the length of second metacarpal.

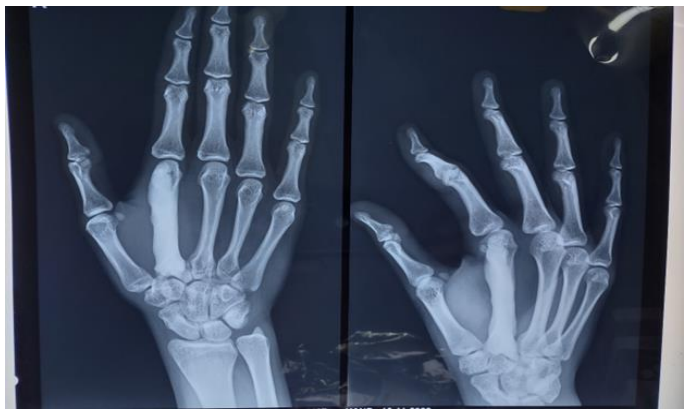

Figure 4: NCCT of the wrist and Hand showing cross and axial section with 3D reconstruction showed the involvement of the second, third metacarpal and capitate bone by diffuse cortical thickening of the bone with dripping wax appearance suggestive of Melorheostosis