

Melorheostosis in proximal phalanx and metacarpal bone: a short-mid-term follow up

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KEYWORDS

Melorheostosis;
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Sclerosing
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Candle bone disease

ABSTRACT

Melorheostosis is a rare sclerosing hyperostosis that affects the appendicular skeleton more than the axial skeleton. This disorder also affects the cutaneous and soft tissues and leads to the formation of fibrosis and contracture. Due to the unique features of melorheostosis (dripping candle wax appearance), plain radiographs are often sufficient for diagnosis. Herein we report a case of melorheostosis in proximal phalanx and metacarpal bone. The case is a 21-year-old girl with intense pain in her left hand. She underwent conservative treatment and followed for five years. Outcome measures indicate pain reduction despite no significant changes in radiographs.

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Abbreviations

CT, Computed tomography; MRI, Magnetic resonance imaging; VAS, Visual analogue scale; ROM, Range of motion; MCP, Metacarpophalangeal; PIP, Proximal interphalangeal; NSAIDs, Non-steroidal anti-inflammatory drugs;

Introduction

Melorheostosis, candle bone disease, or Leri disease is a rare bone dysplasia with an incidence of less than one in a million (1). It is an uncommon sclerosing hyperostosis with unknown etiology and equal frequency of occurrence between both sexes (2). This benign bone disorder was first discovered by Leri and Joanny in 1922 (3). Although melorheostosis has been reported in the various regions of the appendicular skeleton, the upper limb bones have been rarely affected (4). Moreover, there is often dermal involvement and soft tissue contracture along with bone involvement (5).

The occurrence of this infrequent disease is usually accompanied by pain. Swelling, deformities, tenderness and limited range of motion can be considered as the other symptoms. Nevertheless, melorheostosis can sometimes be completely asymptomatic (4, 6). X-ray imaging, bone scans, computed tomography (CT) scan and magnetic resonance imaging (MRI) assessment have been used to diagnose melorheostosis (7). However, most of these diagnosis methods are often done to rule out differential diagnosis such as infection, osteoma, asphyxia myositis, malignancies, or in suspected cases with multiple causes.

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In this article, we report a rare case of melorheostosis arising on the left 4th proximal phalanx and metacarpal bone after obtaining a signed informed consent from the patient.

Case Report

The case was a 21-year-old female with relatively moderate pain (6/10 in severity on visual analogue scale (VAS)) of the left upper limb (forearm, ulnar side of the wrist and 4th finger) of around 3 years' duration who was referred to our outpatient hand clinic in referral orthopedic hospital. The pain had occurred without any trauma and was intensified with activity and sometimes in the nighttime. In general clinical evaluation, no records of fever, night sweating or significant weight loss were evident, and her full neurovascular assessment was normal. Moreover, Hematological and biochemical tests were clear without any noticeable abnormal condition. The Patient's medical history was clear and there were neither family histories of cancer nor her, though.

Physical examination revealed tenderness upon palpation on the dorsal side of the left 4th proximal phalanx which was prominent. Any movement restriction was not seen so that the range of motion (ROM) was normal (The active ROM of the metacarpophalangeal (MCP) joint was 10 degrees of extension to 80 degrees of flexion and the active ROM of the proximal interphalangeal (PIP) joint was 0 degrees of extension to 90 degrees of flexion).

Plain radiography of the left hand illustrates sclerosis in the left 4th proximal phalanx and metacarpal bone and the hook of the hamate bone (Figure 1). Dripping candle wax pattern was consistent with a diagnosis of melorheostosis. A bone scan revealed increased radiotracer uptake in the left 4th proximal phalanx and left 4th metacarpal bone, accompanied by regional reactive hyperemia which implied benign bony lesions such as melorheostosis or fibrous dysplasia (Figure 2).

Nevertheless, the lack of any increased radiotracer uptake on the flow and blood pool images endorsed the non-vascular lesions.

The patient underwent symptomatic therapy (non-steroidal anti-inflammatory drugs (NSAIDs), occupational and eventually intermittent splinting therapy) which led to a decrease in pain (1/10 severity on VAS) during a five-year follow-up. However, radiographic findings were persistent and indicated neither improvement nor progression of the disease (Figure 3).



Figure 1. Baseline plain radiography of the left hand. a: anteroposterior (AP) view; b: lateral view; c: oblique view.

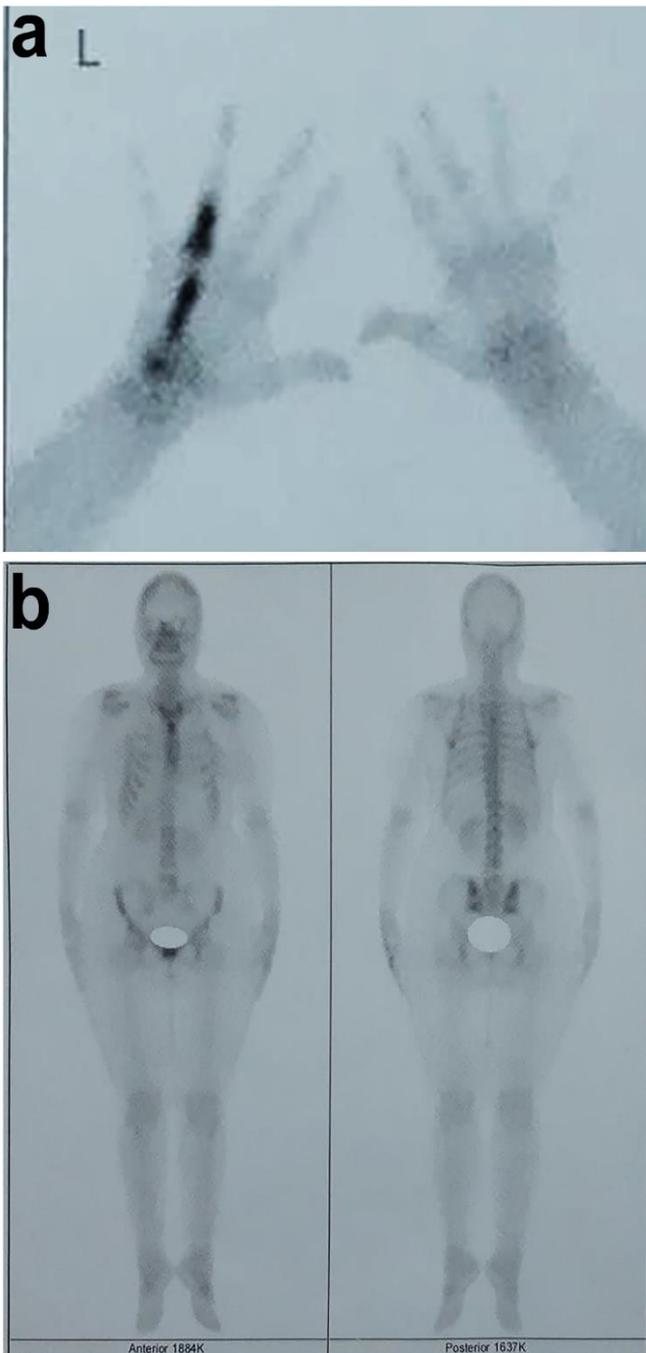


Figure 2. Bone scan. a: the dorsal view of a bone scan of the hands. The left fourth proximal phalanx and metacarpal bone illustrated intense tracer uptake in the bone phase. b: whole body bone scan.

Discussion

Melorheostosis, as an uncommon dysplasia, is characterized by immoderate thickening of the cortical bones, dermal involvement and soft tissue contracture. Approximately 50% of cases are diagnosed at the age of 20 (4). Among the various diagnostic methods such as MRI, Bone scan and CT scan which have been used for the prevention of differential diagnosis in peculiar cases, conventional radiography is the most appropriate method.

A typical pathognomonic presence of melorheostosis, dripping candle wax, could be revealed by X-ray imaging (8). Although the etiology of this disorder has remained uncertain, somatic mosaic MAP2K1 mutations have been reported in some patients with melorheostosis (9).

There is no standard and definite cure for melorheostosis until the date and existing options are individualized based on the patients' conditions such as age, pain severity, bone deformities and lifestyle (8). Physical therapy, occupational therapy, medications including NSAIDs and surgery are suggested treatment methods which have been reported to minimize the symptoms of melorheostosis in different studies (10-12).

In the case reported in this study, melorheostosis in the left 4th proximal phalanx and metacarpal bone was diagnosed by the assessment of bone scan and radiographic findings. In order to symptomatic management, NSAIDs, occupational and intermittent splinting therapy were prescribed for her, and she was followed up for five years with regular radiography. In a similar study, Abed et al. reported a case of melorheostosis in the hand of a 33-year-old Saudi female who suffered from mild to moderate hand pain. They only referred her to physiotherapy and followed her up for four years. During this period, no change in her symptoms and radiographs were reported (13).

Conclusion

We report melorheostosis in the left 4th proximal phalanx and metacarpal bone with short-mid-term follow-up. After five years, symptomatic therapy led to a decrease in pain while no radiographic changes were seen. We recommend that symptomatic therapy and analgesic medications such as NSAIDs or other anti-inflammatory drugs be used for pain control in melorheostosis patients.

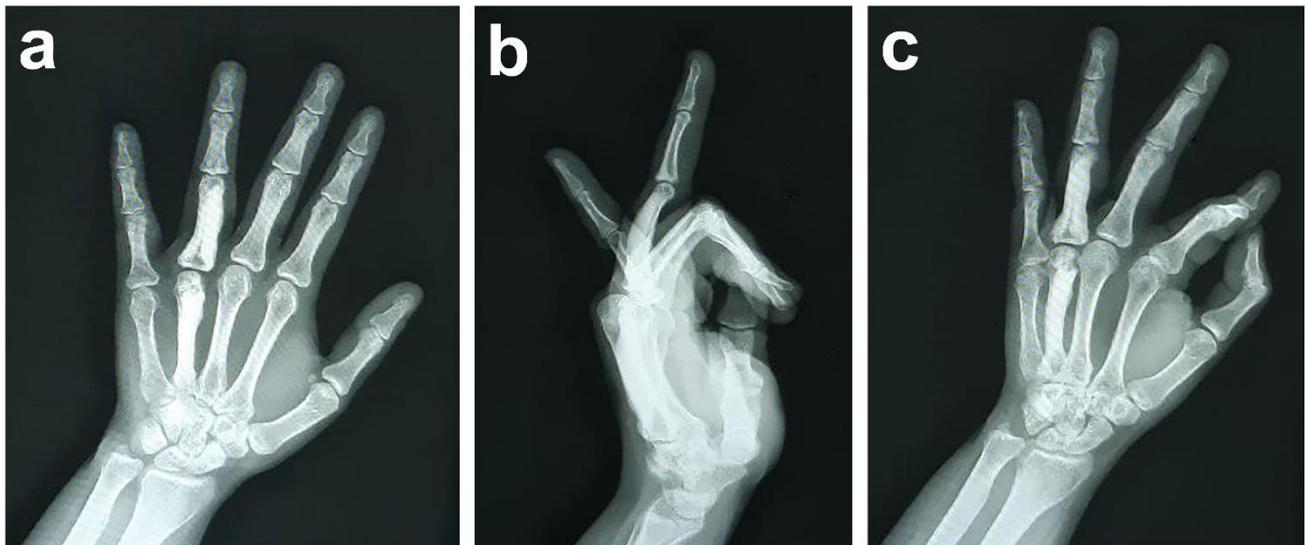


Figure 3. Plain radiography of the left hand after 5 years follow up. a: anteroposterior (AP) view; b: lateral view; c: oblique view.

Declaration

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Conflicts of interest/Competing interests

The authors declare no conflict of interest.

Authors' contributions

FNM and MH conceived the design of the study. MH prepared the manuscript. MM and MR reviewed literature and performed necessary interventions. SR, MM, and HR participated in data collection. All authors contributed toward drafting and critically revising the report and agree to be accountable for all aspects of the work.

Ethics approval

Not applicable.

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