Available online at www.sciencedirect.com

SciVerse ScienceDirect

Journal of the Chinese Medical Association 74 (2011) 469–472

www.jcma-online.com

Case Report

Melorheostosis of the ulna

Shalimar Abdullah, Gerry M.H. Pang, Nor Hazla Mohamed-Haflah*, Jamari Sapuan

Department of Orthopaedics, Faculty of Medicine, Universiti Kebangsaan Malaysia, Kuala Lumpur, Malaysia

Received September 1, 2010; accepted December 20, 2010

Abstract

Melorheostosis is a rare osteosclerotic bone dysplasia. It is usually characterized by dull and aching pain, reduced joint motion and contractures. Classic radiograph findings are of undulating cortical hyperostosis along the length of the bone, simulating a "dripping candlewax appearance". We report two cases of melorheostosis of the ulna bone, diagnosed 6 years apart in two different females in their early 20s. Both the patients presented with the characteristic features of dull and aching pain in the forearm and were treated conservatively. However, we misdiagnosed the first case as bone malignancy and subjected the patient to a biopsy. For the second case, with hindsight we made the correct diagnosis based only on the classic clinical history and radiographs. We believe that the discussion of a misdiagnosed case of melorheostosis with salient findings may be important for clinicians and orthopedicians in day-to-day clinical practice.

Keywords: hyperostosis; melorheostosis; pain

1. Introduction

Melorheostosis is a rare osteosclerotic bone dysplasia of unknown etiology, characterized by slowly progressing hyperostosis.[1] The first reported case, described by Leri and Joanny in 1922, was a patient who had involvement of an upper extremity. Melorheostosis usually affects the long bones of the upper and lower limbs but may also involve the short bones of the hand and foot and, rarely, the axial skeleton.[2] It is pertinent to mention that the nonmalignant fibrosing tumors in the pediatric hand or juvenile fibromatoses are clinically challenging because of their rare occurrence.[3]

The name of the disease derives from combining two Greek words, *melos* (member or limb) and *rhein* (flow), producing a "flowing limb" describing the radiographic appearance of hyperostosis resembling molten wax running down the cortex of the bone.

Although the cause is unknown, a sclerotomal pattern is usually evident. A sclerotome is a zone of skeleton supplied by an individual spinal sensory nerve. This may be due to a monomelic tendency and linear track pattern of melorheostosis, as was initially reported by Putti, hence suggesting the name "osteosis eburnisans monomelica".[4]

An earlier researcher had proposed mosaicism as a cause because the occurrence is sporadic, and there is an asymmetric "segmental" pattern with variable extent of involvement.[5] Although melorheostosis is a nonhereditary disorder, it has been suggested that melorheostosis may originate as a type of segmental manifestation of osteopoikilosis, an autosomal dominant disorder.[6]

2. Case report

2.1. Case 1

A 22-year-old female presented in 2002 with swelling and dull pain over her right forearm. She had started to notice the swelling 4 years before, and the pain started approximately 1 year prior to consultation. The pain was vaguely described by the patient as an uneasy feeling over the forearm and

* Corresponding author. Dr. Nor Hazla Mohamed-Haflah, Department of Orthopaedics and Traumatology, Faculty of Medicine, Universiti Kebangsaan Malaysia, Jalan Yaakob Latiff, Bandar Tun Razak, Cheras 56000, Kuala Lumpur, Malaysia.

E-mail address: hazla1971@yahoo.com (N.H. Mohamed-Haflah).

1726-4901/$ - see front matter
doi:10.1016/j.jcma.2011.08.019

accentuated by resting her forearm against hard surfaces. However she was still able to carry on with her daily activities. She gave no constitutional symptoms and no significant family history. There was no other location of involvement clinically.

On examination, there was a diffuse swelling of the right forearm on the ulnar side with no thickening or puckering on the overlying skin (Fig. 1). There was no limitation of motion or rigidity of the elbow. On palpation, there was a knobby feeling on the ulna bone, which was nontender. Serum calcium, phosphorus and alkaline phospatase levels were also found to be normal.

Anteroposterior and lateral radiographs of the right forearm (Fig. 2) showed dense sclerosis involving the posterior cortex and adjacent medullary cavity of the proximal ulna with wavy margin. It was running from the distal third of the ulna down to involve the posterior two-thirds of the bone and distal end of the humerus. However, the elbow joint was not involved. No bony expansion was detected. The radius bone appeared to be normal. We did not request a radiologist's report, nor did we subject the patient to a computed tomography (CT) or magnetic resonance imaging (MRI) scan. We felt that neither investigation would add more information. Because the bony protuberance was easily palpable, there was no requirement for a guided biopsy.

The provisional diagnosis of bony malignancy was made, and the patient underwent a trephine biopsy of her bone. However, the trephine biopsy failed because the bone was too hard, and we took a biopsy with a bone nibbler. Because our oncologists routinely require a histopathological report, we did not consult them prior to biopsy.

Histopathological examination showed fragments of bony trabeculae with attached fibrous tissue. Skeletal muscle bundle fibers, myxoid areas and a few smaller fragments of necrotic bony tissue were seen (Fig. 3). The patient was diagnosed with melorheostosis.

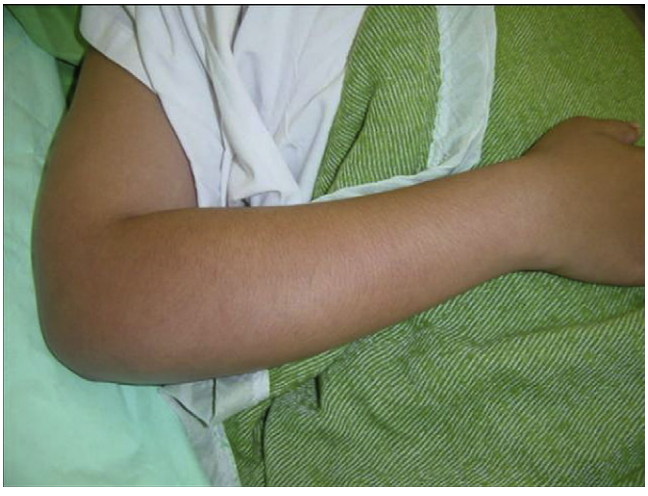

Fig. 1. Clinical view of the first patient's right forearm with a diffuse swelling on the ulnar side with no thickening or puckering on the overlying skin.

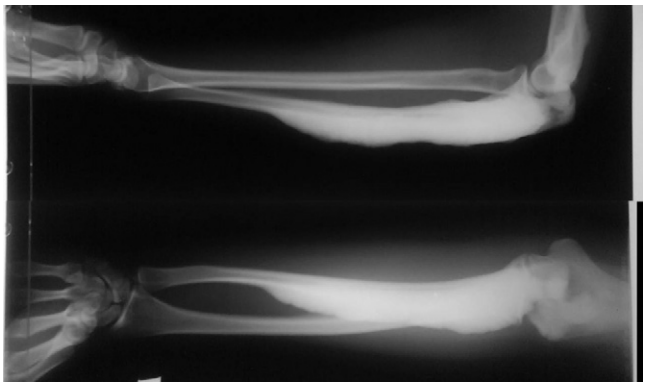

Fig. 2. Radiographs of the first patient. Dense sclerosis involving the posterior cortex and adjacent medullary cavity of the proximal ulna with wavy margin. The elbow joint is not involved. The radius appeared normal.

At her 8-year follow-up, this patient complained of pain only on lifting weights more than 5 kg. She had also noticed a gradual limitation of her elbow range of motion at 65–120°. Her pronation was 0–45° and supination 0–75°. This is not surprising because the melorheostosis had progressed into the elbow joint (Fig. 4).

2.2. Case 2

A 26-year-old female presented in June 2008 with dull pain over her left forearm for the past 2 years. The pain was also vaguely described, and she was still able to carry on with her daily duties as a registered nurse. She reported no constitutional symptoms, and no significant family history was obtained. Examination showed no swelling of the left forearm, normal overlying skin and no limitation of motion or rigidity of the elbow joint. Clinically, the patient had no other locations which were involved. Blood investigations were found to be normal.

Anteroposterior and lateral radiographs of the left forearm (Fig. 5) showed dense sclerosis of the ulnar cortex and

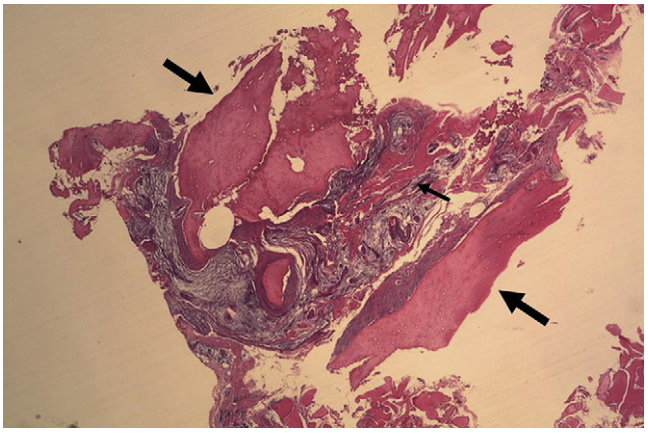

Fig. 3. Section shows necrotic bones (thick arrow) and fibrous tissue (thin arrow) with myxoid area (H&E: 20×).

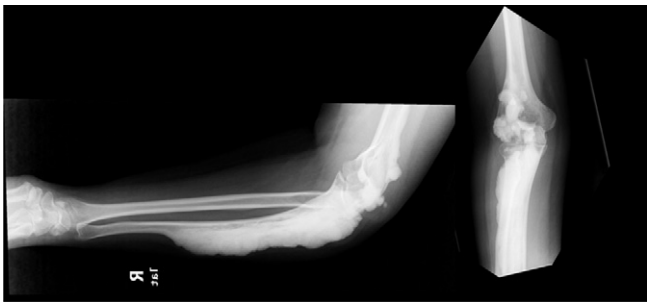

Fig. 4. Radiograph of the first patient at her 8-year follow-up. Marked thickening and sclerosis of the cortical diaphyses and metaphyses of the right ulnar bone had crossed into the elbow joint and distal humerus.

adjacent medullary cavity similar to Case 1. The wavy hyperostosis was running down to the proximal end of ulna without involving the elbow joint. From these characteristic clinical and radiographic features, and with our previous experience, we focused on a definitive diagnosis of melorheostosis. The patient received analgesics for the pain.

At her 2-year follow-up, she continued to complain of pain. We have discussed various options of management, and she agrees to analgesia and observation.

3. Discussion

We misdiagnosed the first case as bone malignancy and subjected the patient to a biopsy. However, for the second case, with hindsight, we made the correct diagnosis based only on the classical clinical history and radiographs.

The two cases were challenging, especially when the clinical findings were nonspecific and radiographic findings virtually established the diagnosis. Physical examination contributed very little to making a diagnosis. Characteristic features were a dull and aching pain, reduced joint motion and contractures. However, the most consistent feature was pain, and it was pain that brought the patients to the physician.[7]

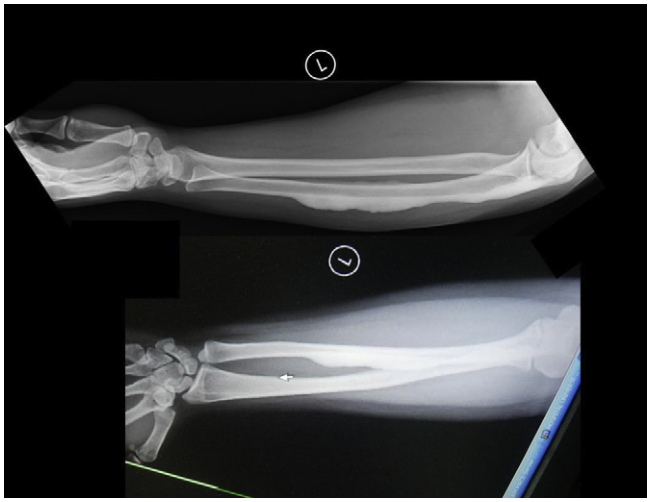

Fig. 5. Radiographs of the second patient. Similar dense sclerosis of the ulnar bone and adjacent medullary cavity. The wavy hyperostosis runs down to the proximal end of ulna without involving the elbow joint.

Localized bone enlargement can cause pain and loss of movement in a nearby joint. Involvement of the soft tissues can lead to contractures. There are reports of successful release of an elbow contracture secondary to melorheostosis.[8] In children, there is a possibility of presentation with limb length discrepancy, deformity or joint contractures even before any radiographic evidence of bony changes are seen.[9] A recent article reported a 10-year-old boy with a severely deformed foot with metatarsus adductus undergoing osteotomies and bone grafting with a successful outcome.[10]

The classic radiographic finding of undulating cortical hyperostosis along the length of the bone simulating molten wax flowing down the side of a lit candle is pathognomonic for melorheostosis.[1,5,10,11] In an earlier study on 23 cases, it was concluded that the classical finding should be revised because there were more osteoma-like and osteopathia striata-like patterns seen rather than the classic candle wax appearance.[5]

The new bone formation mostly involves one side of the cortex of a tubular bone with periosteal and endosteal involvement, and another distinguishing feature is the sclerotomal involvement. Both our cases involved the T1 sclerotome.

Other clinicians have diagnosed melorheostosis based only on clinical and plain radiographic assessments.[10] Radiographic investigations such as CT scan and MRI can further characterize the lesion but rarely contribute to the diagnosis.[2] A clear demarcation between normal and abnormal bone can be revealed with a CT scan, and it is also helpful to confirm or exclude continuity of osseous and soft tissue abnormalities. MRI features of melorheostosis include hyperostosis, which appears as uniform hypointensity on all imaging sequences. It is useful for detecting the presence and extent of associated mineralized and nonmineralized soft tissue masses.

Histopathological examinations of materials from other reports including ours have revealed no characteristic appearance and are inconclusive. There are reports of increased vascular pattern, obliteration of vessel lumina in places, perivascular ossification, atrophic bone marrow rich in osteoblasts, and compactness of the lamellae.[2,12] Cytokines such as transforming growth factor-β and FGF are actively involved in melorheostosis.[12]

It has to be borne in mind that the differential diagnoses include myositis ossificans, osteoma, parosteal osteosarcoma, osteomyelitis, reflex sympathetic dystrophy, neurofibromatosis, soft-tissue sarcoma and metastatic lesions.

To the best of our knowledge, only a few cases of melorheostosis have been reported in the South East Asian region, and we would like to add our cases to the International database.[13]

In conclusion, simple clinical evaluation and radiographs are often adequate to diagnose melorheostosis. We intend to create awareness amongst orthopedic surgeons and general practitioners regarding the presentation of this rare condition in order to prevent unnecessary biopsies or radical procedures. We believe our humble opinion may be beneficial for day-to-day clinical practice.

References

1. Campbell CJ, Papademetriou T, Bonfiglio M. Melorheostosis: a report of the clinical, roentgenographic, and pathological findings in fourteen cases. *J Bone Joint Surg Am* 1968;**50**:1281−304.
2. Greenspan A, Azouz EM. Bone dysplasia series. Melorheostosis: review and update. *Can Assoc Radiol J* 1999;**50**:324−30.
3. Netscher DT, Baumholtz MA, Popek E, Schneider AM. Non-malignant fibrosing tumors in the pediatric hand: a clinicopathologic case review. *Hand* 2009;**4**:2−11.
4. Dillehunt RB, Chuinard EG. Melorheostosis leri: a case report. *J Bone Joint Surg Am* 1936;**18**:991−6.
5. Freyschmidt J. Melorheostosis: a review of 23 cases. *Eur Radiol* 2001;**11**: 474−9.
6. Happle R. Melorheostosis may originate as a type 2 segmental manifestation of osteopoikilosis. *Am J Med Genet A* 2004;**125A**:221−3.
7. Wallace EH, Dana MS. Melorheostosis: relief of pain by sympathectomy. *J Bone Joint Surg Am* 1950;**32**:422−7.
8. Gong HS, Lee KH, Oh JH, Chung JH, Baek GH, Chung MS. Successful elbow contracture release secondary to melorheostosis: a case report. *J Bone Joint Surg Am* 2008;**90**:1106−11.
9. Adeyomoye AA, Awosanya GO, Arogundade RA. Melorheotosis of leri: report of a case in a young African. *Niger Postgrad Med J* 2004;**11**:234−6.
10. Gagliardi GG, Mahan KT. Melorheostosis: a literature review and case report with surgical considerations. *J Foot Ankle Surg* 2010;**49**:80−5.
11. Rozencwig R, Wilson MR, McFarland GB. Melorheostosis. *Am J Orthop* 1997;**26**:83−9.
12. Hoshi K, Amizuka N, Kurokawa T, Nakamura K, Shiro R, Ozawa H. Histopathological characterization of melorheostosis. *Orthopaedics* 2001; **24**:273−7.
13. World Map of Melorheostosis. http://www.melorheostosis.com/default_files/Page1403.htm.