Amputation: A Rare Surgical Indication in Melorheostosis

Khalid Masood1

1Hand and Upper Limb Surgery Center, Lahore, Pakistan

ABSTRACT

Melorheostosis is a rare and progressive mesenchymal dysplasia characterized by thickening (hyperostosis) of the cortical bone. It is a developmental error that has been described to occur due to a mutation in LEMD3 gene. It affects bone and soft tissue development resulting in severe functional limitation; chronic pain, malformed or immobilized muscles, tendons or ligaments limb deformities and leg length discrepancies. X-ray imaging is the preferred diagnostic tool for melorheostosis, which often reveals a pattern of thickened bone resembling dripping candle wax.

Keywords: Melorheostosis; Mesenchymal Dysplasia; LEMD3; Dripping candle wax

INTRODUCTION

Melorheostosis (also known as Leri disease) is an uncommon mesenchymal dysplasia manifesting as regions of sclerosing bone with a characteristic dripping wax appearance [1-3]. It is a rare and progressive disorder characterized by hyperostosis of the cortical bone, affecting both bone and soft tissue growth and development. It results in severe functional limitation, extensive pain, soft tissue contractures and limb and hand deformities [4]. This disease was first described by Leri and Joanny as dripping candle wax hyperostosis and about 300 cases have been reported since then [4]. The incidence is estimated to be 0.9 per million population and equally affects men and women, at any age [1]. The location of occurrence is highly variable with monostotic forms being more common than polyostotic forms [5]. It is a developmental error occurring due to loss of function mutation in LEMD3 gene [6]. This case report describes this rare condition occurring in a polyostotic form that has never been reported in Pakistan.

CASE REPORT

A 68-year-old female complained of gross swelling of her right middle finger with a great degree of restriction in middle finger movements and mild restriction and pain in movements of the index and ring fingers for past 4 years. The swelling had increased in size over the years and there was no relief in pain on taking standard therapies. There was no positive past medical or surgical history. Her personal and family history was noncontributory. There was no known history of trauma during childhood or in recent years.

On clinical examination, there was no active or passive movement at the proximal interphalangeal, distal interphalangeal and metacarpophalangeal joint of the right middle finger. The range of motion decreased by 10 degrees in the metacarpophalangeal joints of index and ring finger. There was a severe functional disability of right hand which interfered in daily routine work.

The patient had been taking NSAIADS and anti-inflammatory drugs without improvement. The patient underwent a series of radiological tests, including X-ray of the right hand which suspected fibrodysplasia of right middle finger and carpal bones including capitate and lunate.
There was dripping candle wax hyperostosis extending over the metacarpals and phalanx of a right middle finger on X-rays and other scans. The CT-Scan and a 3-D scan revealed the typical flowing wax melorheostosis, with significant multifocal sclerotic lesions involving phalanx and metacarpal and carpal bones. A wide spectrum of differential diagnosis related to hyperostosis on radiographs was considered, which included: melorheostosis, myositis ossific ANS, osteoma, focal scleroderma, parosteal osteosarcoma, Caffey’s disease and sclerotic metastasis (e.g. breast and prostate).

Due to the severe functional disability of the right hand and no improvement seen on conservative treatment, a more invasive approach of amputation of the right middle finger along with the affected metacarpal was considered. Amputated finger was sent as a biopsy sample, which confirmed the preliminary diagnosis of melorheostosis. Physiotherapy including active and passive flexion/extension at metacarpophalangeal and interphalangeal joints of remaining fingers was started. Full hand function was successfully achieved within one month.

**DISCUSSION**

Melorheostosis is a rare non-genetic developmental anomaly first described in 1922 by Léri and Joanny as flowing hyperostosis resembling dripping candle wax [1, 3]. The name is derived from the Greek words for limb (Melos) and flow (rhein), due to characteristic flowing hyperostosis [3].

It is one of the developmental diseases of the bone density and is included in other sclerosing disorders such as cranial-metaphyseal and diaphyseal dysplasia as well as the osteoscleroses, osteopetrosis and its variants pyknodysostosis, osteopoikilosis and osteopathic

**Figure 1:** Pre-op, showing swelling of the Middle finger of right hand

**Figure 2:** X-Ray and 3-D showing dripping candle wax appearance

**Figure 3:** Zero Post operative day along with the amputated finger

**Figure 4:** Follow-up visit
Melorheostosis predominantly affects the appendicular skeleton and is most common in the long bones of the upper and lower extremities, although it can be seen in the hands and feet as well. Melorheostosis has rarely been reported in the axial skeleton [2]. There are reports of several associated clinical entities, including scleroderma and von Recklinghausen disease [3].

Histological findings include variable degrees of cortical thickening consisting of chondroid islands surrounded by mature lamellar and woven bone, as well as adjacent zones of fibrocartilage with irregular surface fibrillation. Soft tissue abnormalities consisting of osseous, chondroid, vascular, and fibrocartilaginous tissue have been reported in ≤76% of cases of melorheostosis [10]. This disease may present at any age and may be seen in children as well as in adults. There is no sex predilection. The condition is often asymptomatic, but sometimes pain, stiffness, and limb deformity may be present along with the limitation of movement [11]. There may be a severe functional limitation, extensive pain, soft tissue contractures, limb and hand deformities, leg length discrepancies and rarely a malformation of blood or lymph vessels [4].

On plain radiography, melorheostosis is classically described as periosteal hyperostosis; the most frequent appearance is an osteoma-like hyperostosis on the endosteal surface of the bone. The lesions are typically eccentrically placed with no evidence of bony destruction. Isotope bone scanning reveals increased uptake in the same distribution seen on plain radiography reflecting an increase in bone metabolism. CT scan reveals the lesion and a clear demarcation between the normal and abnormal bone. MRI shows soft tissue and bone lesions as areas of low signals on all sequences [11]. In the vast majority of cases, CT and MRI are not needed for diagnosis.

Most cases are benign and may be treated conservatively with bisphosphonates which inhibit osteoclast-mediated bone reporting by direct and indirect actions on osteoblasts and macrophages [11]. And most cases are treated with NSAIDS or Zoledronic acid for symptomatic relief. Sometimes, treatment may include surgery for tendon lengthening and correction of deformities. In severe and very rare cases, amputation of the part of the affected limb is indicated because of contractures causing limited functionality and ischemia. The recent literature mentions that conservative treatment with bisphosphonates and COX-2 inhibitors in combination with naftidrofuryl can delay surgery solution [13].

Melorheostosis is rare and benign with a chronic course, often interspersed with remissions. The diagnosis is usually made by plain radiography, CT scan or MRI and if any doubt biopsy may be necessary. The treatment is essentially symptomatic. But, in cases of severe functional disability and no improvements on symptomatic treatment, a more radical approach to surgery is considered. The presented case report is one of the very few cases from Pakistan, requiring the radical approach of amputation in order to restore full hand function.

REFERENCES

