

Three Cases of Melorheostosis With Foot and Ankle Involvement

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Abstract

Melorheostosis is a rare and poorly understood condition of bone and soft tissue with a wide range of clinical presentations. This condition is typically characterized by cortical hyperostosis and pain in the involved extremity, but can also be associated with soft-tissue masses and limb deformities that may be additional sources of disability for those affected by this disease.

Characteristic radiographic findings can aid in establishing an accurate diagnosis and the condition should not be mistaken for more aggressive neoplasms. This chronic condition is typically managed nonoperatively, but more invasive measures may be necessary when nonoperative measures fail. In cases of surgical intervention, physicians and patients should be aware that this disease has a high recurrence rate.

Although there are only a few reports of melorheostosis in the foot and ankle, it is important to be aware of the difficulties the condition may cause in this anatomical location. Melorheostosis can be a source of significant morbidity when the foot and ankle are involved, especially when complicated by symptomatic soft-tissue masses.

In this article, we report 3 cases of melorheostosis in the foot and ankle with distinct presentations and variations in outcomes.

Melorheostosis is a poorly understood condition of bone and soft tissue that is typically characterized by pain and dysfunction in the involved extremity. The condition is uncommon, with an incidence of 0.9 case per million, making it a diagnostic dilemma.¹ Melorheostosis usually involves just one extremity, with the lower extremity affected twice as often as the upper extremity.²⁻⁶ History taking and physical examination are important aspects of evaluation, but

it is the typical radiographic finding of “flowing candle wax” in the involved bone that leads to the correct diagnosis.^{2,4,7,8} Many etiologies have been proposed for the condition, but given its classic dermatomal distribution, many believe the dysplasia is congenital.^{3,4,8,9} It typically has a benign course, but malignant transformation has been reported in rare instances.¹⁰

Clinical presentation can also differ by degree of osseous involvement and patient age, and can vary from incidental radiographic findings to debilitating pain and stiffness.^{2,3,8,9,11,12} Children may not have the typical findings of pain and cortical hyperostosis but instead may manifest disease in the form of limb deformity with more subtle radiographic findings.¹¹⁻¹³ When the lesion is metaphyseal, it may cross nearby joints and become a source of significant stiffness and decreased range of motion (ROM).² This condition has also been associated with soft-tissue findings and vascular malformations, which may be additional sources of pain and add to the difficulty in establishing an accurate diagnosis.^{2,7-9,13-15}

There are few accounts of manifestations of this disease in the foot and ankle.^{2,5,16} In this article, we report 3 cases of melorheostosis involving the foot and ankle with distinct outcomes, illustrating the wide variations in presentation. The living patients described here provided written informed consent for print and electronic publication of their case reports.

CASE REPORTS

Case 1

A 53-year-old woman presented with a 3-month history of atraumatic right leg pain that was aggravated by dancing and walking and relieved with rest and use of non-steroidal anti-inflammatory drugs (NSAIDs). The patient denied weight loss or any other constitutional symptoms.

Physical examination revealed no gross limb deformity, but there was a tender mass in the lateral aspect of the right

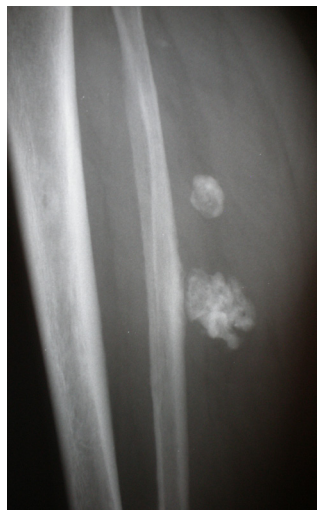


Figure 1. Initial radiographs of a 53 year-old female with atraumatic leg pain show a mineralized lesion with peripheral maturation.

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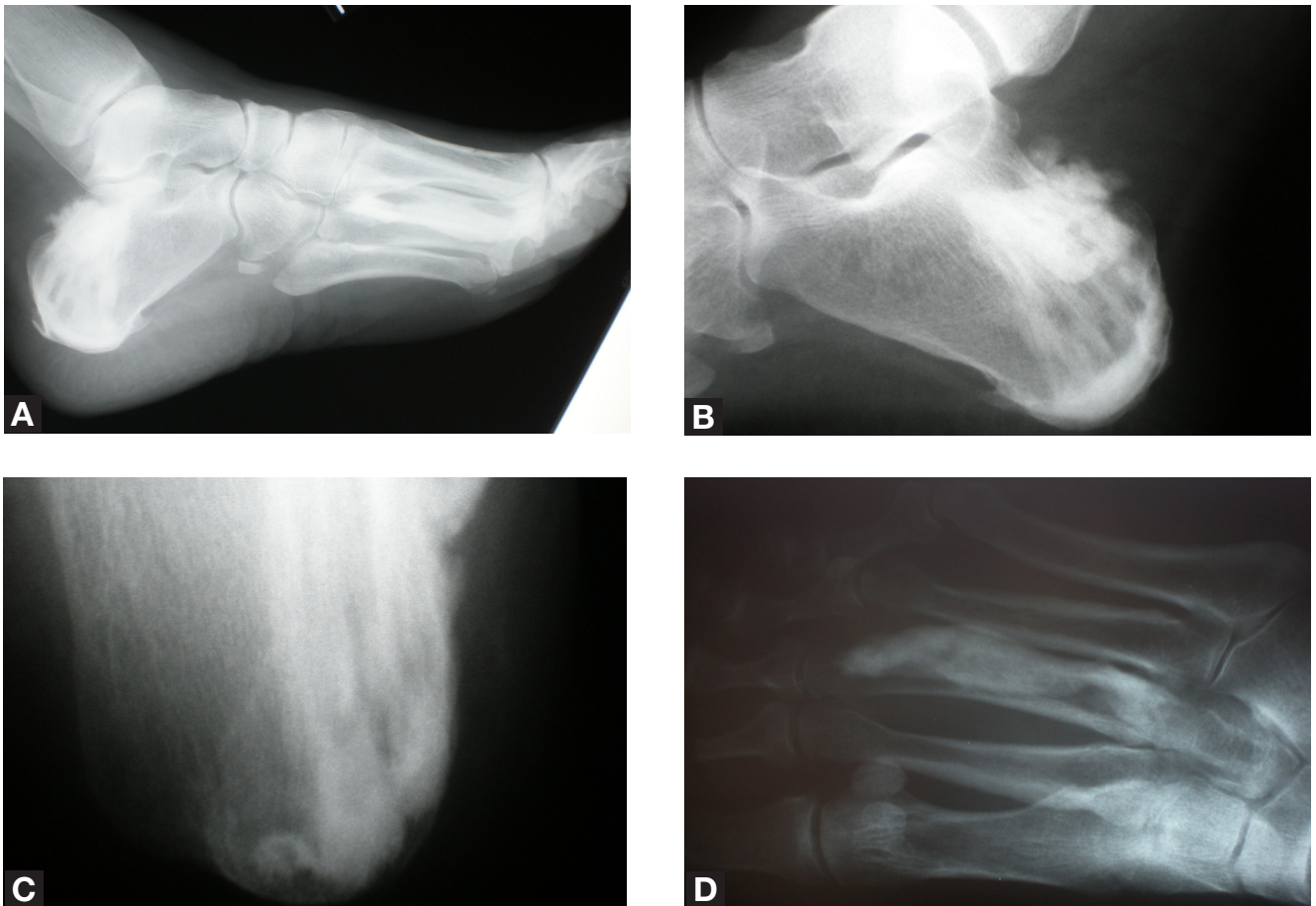


Figure 2. (A) Further imaging identified a hyperdense lesion, confluent with the cortex of the calcaneus and third metatarsal. (B) The lesion was reminiscent of flowing candle wax. (C) Despite the periarticular location of the lesion, the patient had full passive and active range of motion of the foot and ankle. (D) The patient had a good outcome with conservative measures, which included activity modifications and oral anti-inflammatory medications.

leg. This mass was freely mobile and not adherent to underlying bone. There were no overlying skin changes. The patient had full passive and active ROM of the knee and ankle.

The diagnosis of melorheostosis was based on typical radiographic findings. Radiography showed a mineralized mass with peripheral maturation in the midlateral leg (Figure 1) and a hyperdense lesion in the calcaneus and third metatarsal that appeared confluent and hyperostotic, reminiscent of flowing candle wax (Figures 2A-D). Computed tomography showed a nondestructive cortical density in the calcaneus. Bone scan showed intense and homogeneous radiotracer uptake in the right ankle and mid leg. On both T1 and T2 pulse-weighted sequences, magnetic resonance imaging (MRI) showed a 2.4×1.5-cm hypointense lesion abutting the periosteum of the posterior fibula.

Conservative measures with close follow-up were recommended. Annual visits showed no progression of disease. The patient continued to do well with activity modifications and use of oral anti-inflammatory medications and was content with her outcome over 8-year follow-up.

Case 2

A 34-year-old woman presented with a known diagnosis of melorheostosis dating back to age 15. She reported right ankle stiffness associated with a painful mass that interfered with shoe wear. She first noticed the mass 7 months earlier and stated that she previously had a similar mass in the knee that was surgically removed at another institution when the condition was first diagnosed. The patient denied constitutional symptoms or any history of trauma to the involved extremity.

Physical examination revealed a well-healed incision in the posterior aspect of the right knee, mild erythema, and swelling in the anteromedial aspect of the ankle, with a firm and fixed mass that was tender to palpation. There was a noticeable brownish discoloration of the skin extending from the foot up to the lateral aspect of the thigh (Figure 3). Radiography of the extremity showed a hyperostotic, cortically based lesion involving the distal femur, the distal tibia, and the forefoot (Figures 4A-B). On T1 and T2 pulse-weighted sequences, MRI showed uniform low signal changes along the anterior cortex of the tibia, the medial malleolus, the tibial plafond, the navicular, the base of the first metatar-



Figure 3. Clinical photo of a 34-year-old female with hyperpigmentation of the skin overlying the ankle. She presented with ankle stiffness associated with a painful mass.

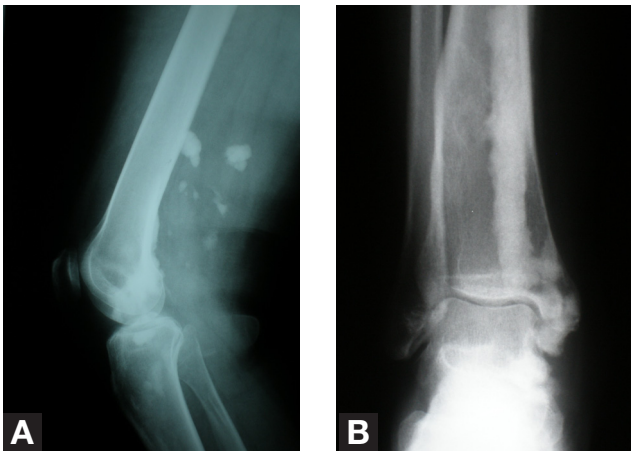


Figure 4. (A) Initial radiographs showed a hyperostotic, cortically based lesion along the distal femur associated with mineralized soft tissue masses. (B) The lesion also involved the distal tibia and forefoot of the same extremity.

sal, and the medial cuneiform. In addition, there was a heterogeneous, contrast-enhancing soft-tissue mass over the medial aspect of the talus. The lesions appeared to be mineralized, with predominantly low-signal intensity. The decision was made to treat the patient symptomatically with NSAIDs and regular radiographic follow-up.

Over the next 16 months, the patient came in for regular follow-up examinations and reported various concerns, including increasing knee pain and stiffness, as well as progressive and poorly controlled ankle symptoms. She continued conservative treatment with anti-inflammatory medications until September 2004, when she requested operative excision of the ankle mass. The soft-tissue mass was removed from the anterior medial ankle, adjacent to the talonavicular joint, without complication. Histologic evaluation of the mass revealed synovium with areas of chronic inflammation and myxoid changes, as well as areas of lamellar and woven bone with increased osteoclastic and osteoblastic activity.

The patient was followed closely. Outcomes were sat-

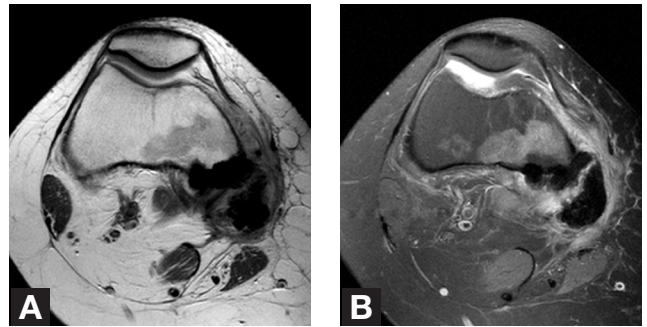


Figure 5. (A) Due to increasing pain about the knee, an MRI was obtained demonstrating a highly mineralized soft tissue mass with surrounding edema, as well as a low signal, cortically based lesion in the distal medial thigh. (B) The patient's symptoms were poorly controlled with medications, so the decision was made to proceed with excision of the ankle and knee soft tissue masses. She has done well throughout her 54 months of follow-up.

isfactory until February 2008, when she began to report knee pain that was poorly controlled with medication. Repeat MRI showed an exophytic low-signal cortical lesion with surrounding edema in the medial distal posterior femur with a large mineralized soft-tissue mass of low signal intensity in the distal medial thigh (Figures 5A-B). Given the patient's poorly controlled pain, bisphosphonates were started, and surgical intervention was discussed.

Five months later, after nonoperative therapy failed to control her symptoms, the patient requested resection of the knee mass. The mass was excised without complication, and histologic evaluation of the resected lesions revealed heterotopic bone with fibromyxomatous changes. Follow-up examinations performed during the 52 months after excision of the masses revealed significant improvement in pain and ROM of both knee and ankle.

Case 3

A 28-year-old woman presented with reports of dyspareunia, which she had for many months. Radiographs showed cortical hyperostosis in the iliac crest, the medial femur, the tibia, and along the medial hindfoot, extending to the first ray. In addition, a mineralized soft-tissue mass was identified in the right groin (Figures 6A-C). Bone scan showed hyperintense scintigraphic uptake, and MRI showed a hypointense cortical lesion on all pulse-weighted sequences in the involved bones.

The patient denied having symptoms in the foot and ankle, and agreed to nonoperative treatment for dyspareunia. As her anti-inflammatory medications provided poor relief of symptoms, and her concerns continued during NSAID-only management, the decision was made to add bisphosphonates to the regimen. The added medication did not help, and the patient continued to have pain with sexual intercourse. Seven years later, she died of unrelated causes, never obtaining full relief from her original symptoms.



Figure 6. (A) Radiographs of a 28-year-old female with the report of dysparinuria demonstrated a cortical hyperostosis involving the iliac crest, femur, tibia, and medial hindfoot, extending down the first ray. (B) Imaging also identified a mineralized soft tissue mass in the right groin. The patient continued to have symptoms despite oral anti-inflammatory medications and bisphosphonates. (C) The patient denied any pain or limited range of motion of the foot and ankle, despite the periarticular location of the lesion. The cortically based lesion demonstrated the typical flowing candle wax appearance associated with melorheostosis.

DISCUSSION

As these cases have demonstrated, melorheostosis has many clinical manifestations, ranging from a lack of overt symptoms to debilitating pain. These variations in presentation have been well described in the literature, particularly in relation to differences based on patient age. Previous authors have noted that children typically present with painless extremity deformities, whereas adults usually have painful lesions.^{2,3,8,9,11,12} The nature of the disease is poorly understood, and little is known about its evolution, but a proposed congenital etiology could explain the dermatomal distribution of the condition.^{3,4,8,9} There is no sex predilection, and most patients are diagnosed in childhood or early adolescence with progression into adulthood.^{2,6,8,9}

The condition is thought to be caused by a defect in intramembranous and endochondral bone formation.⁸ It typically involves the long bones of the lower extremity, but involvement of the small bones of the hands and feet has also been described.⁸ The disease can involve a single bone (monostotic), multiple bones in one extrem-

ity (monomelic), or multiple extremities (polyostotic).⁸ Cutaneous manifestations are not uncommon, with typical findings of tense, firm skin overlying osseous lesions.^{4,9} Hyperpigmentation, as was found in our second case, is a rare presentation of the condition but has also been described in the literature. The hyperpigmented lesion has been biopsied and histologically described as hyperkeratotic with dense proliferation of deep dermal collagen.¹⁷

Limb-length discrepancies have also been described. When the lesion is periarticular, contractures and joint stiffness may occur, especially when the lesion crosses the joint.² Intra-articular extension of this entity has been found in 35% of patients.¹⁴ Campbell and colleagues⁹ described 14 patients' clinical findings. All patients had decreased ROM in the involved extremity, and foot deformities were common. Most patients reported pain.

Radiography is the cornerstone of the diagnostic workup. Freyschmidt⁶ described 5 radiographic variants: osteoma-like, classic flowing candle wax, myositis ossificans-like, osteopathia striata-like, and mixed. The classic type was found in the minority of patients in that series. Bone scan typically shows intense radiotracer uptake, and uptake intensity and distribution may help distinguish this entity from other, similar ones.^{2,3,8} The lesion in our first case was initially thought to be myositis ossificans because of the peripheral maturation seen in the soft-tissue radiographic manifestations. This case may have been better classified as a myositis ossificans-like variant, as previously described. Increased uptake on bone scan, along with the radiographic finding in other bones of the extremity, assisted in making the correct diagnosis.

MRI usually shows a cortical lesion with decreased intensity on all pulse sequences, as was the case in our patients.^{8,14} Judkiewicz and colleagues¹⁴ reviewed the advanced imaging findings of 17 patients with melorheostosis; 76% of these patients had an associated soft-tissue mass. Depending on the extent of mineralization, these masses were generally infiltrating, heterogeneous, and contrast-enhancing, mimicking more aggressive neoplasms.¹⁴ All the patients in our series had melorheostosis with an associated soft-tissue mass, and most of their concerns stemmed from these masses. Even in the patients in whom most of the lower extremity was involved, the presenting symptoms, as well as the symptoms that were most difficult to control, involved the presence of a soft-tissue mass.

Woolridge and colleagues² described the typical histologic appearance of melorheostosis. They found that the cortical lesion is typically composed of an abnormal proliferation of compact bone, which can be woven, lamellar, or both, depending on maturation stage. They also described a lack of prominent osteoclastic activity.² The histologic makeup of the soft-tissue mass has been described as fibrovascular, osteocartilaginous, with

areas of adipose tissue and varying degrees of mineralization.¹⁴ Occasional eosinophilic degeneration of this predominantly fibrovascular tissue has been described; it is thought to result from tissue ischemia caused by the underlying cortical changes associated with the disease.¹⁵ The findings in our patients are somewhat different, as the degree of mineralization to the soft-tissue component was larger. This may be attributed to the long-standing history in these patients.

Management is typically nonoperative, except in limited clinical circumstances and after failed nonoperative management. Surgical management of limb deformities usually is unsuccessful, and recurrence is common.^{8,12} Success in the management of limb deformities has involved use of Ilizarov fixators for gradual correction of contractures.¹⁶ Pain control can be achieved with use of anti-inflammatory medications, as was found in our first patient, and bisphosphonate use has been espoused by some as an effective management method. The success of bisphosphonates in controlling pain associated with this condition has been attributed to the anti-inflammatory and antiarthritic properties of these medications, yet they have not been shown to alter the course of the bone or soft-tissue lesions.^{18,19} Successful pain control with use of such medications also varies in the literature and is illustrated in our 3 patients.²⁰ When nonoperative management fails, surgical intervention may be the only suitable option, but the patient should be counseled on the high incidence of recurrence associated with invasive measures.

This series illustrates the wide variations in outcomes found with the different management options and reinforces the fact that patients should be well informed about the unpredictable nature of this disease. Foot and ankle involvement is not uncommon in patients with this condition, and the spectrum of clinical presentations and outcomes should be understood by foot and ankle specialists. All 3 patients in this series had some pain and a soft-tissue mass associated with their affected extremity. A conservative measure, NSAID use, was successful in the first patient, had only moderate results in the second patient, and was unsuccessful in the third patient. Use of bisphosphonates was also only partially successful in the second and third patients, but good results were obtained with limited surgical intervention when it was used. This intervention was directed solely at the resection of symptomatic soft-tissue masses resistant to noninvasive measures and was not intended as a curative measure.

CONCLUSION

Melorheostosis is a poorly understood condition that is commonly associated with pain and deformity in the involved extremity. Foot and ankle involvement is not uncommon and may mitigate normal shoe wear and interfere with daily activities. Radiographic findings are typical

and traditionally have been described as flowing candle wax, but variations are possible, as described. Advanced imaging may be useful in patients with atypical radiographic findings to ascertain the proximity to normal adjacent structures and the pattern of mineralization or zonation. The goal of management is to provide symptomatic relief, which is usually achieved by nonoperative means. In this small series, surgery was successful in relieving pain and restoring normal function, but should be used with caution. In other situations, the patient may be afflicted with a painful, chronic condition that is very difficult to manage. These case reports illustrate the wide range in presentation and outcomes of melorheostosis of the lower limb, specifically the foot and ankle, and both patients and physicians should be well informed regarding the variation in clinical symptoms and treatment outcomes.

AUTHORS' DISCLOSURE STATEMENT

The authors report no actual or potential conflict of interest in relation to this article.

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