The Journal of Foot & Ankle Surgery 49 (2010) 80–85

Contents lists available at ScienceDirect

The Journal of Foot & Ankle Surgery

journal homepage: www.jfas.org

Melorheostosis: A Literature Review and Case Report with Surgical Considerations

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ARTICLE INFO

Level of Clinical Evidence: 4
Keywords:
bone graft
contracture
dehiscence
fibrosis
foot
metatarsus adductus
sclerosis

ABSTRACT

Melorheostosis is a rare disorder marked by increased bony sclerosis on radiographs. In addition to bone changes, the skin and soft tissues overlying affected bone often demonstrate increased fibrosis, which can create joint contracture. These can all affect surgical planning for a patient with melorheostosis. This article reviews the literature and describes the surgical intervention and 4-year follow-up of a 10-year-old boy with melorheostosis.

In 1922, Léri and Joanny (1) described a pattern of hyperostosis distinguished by a linear pattern of distribution along the major axis of long bones, and they termed this condition melorheostosis. Melorheostosis is not associated with increased mortality; however, there is an associated functional morbidity. The long bones of the lower extremity are most commonly affected (2). This disease is associated with progressive contracture of involved joints and soft tissues. Women and men are affected equally by the disorder, which usually presents after early childhood (3).

Although some cases of melorheostosis are discovered by coincidence before the development of signs or symptoms, it is often pain, stiffness, and limitation of joint motion or deformity that initially prompts patients to seek medical attention (2). The onset is insidious and symptoms vary considerably, from none to severe deformity owing to contracture (3, 4). Although pain, which ranges from dull to sharp and penetrating, is present in almost all cases of melorheostosis, it is rarely constant or severe, and it is often aggravated by activity (2, 3, 5). Soft tissue fibrosis and contractures are often present and result in restriction of joint motion. In Campbell et al's (2) study of 14 patients, the most common location of soft tissue contracture was the plantar fascia. At times, the condition may be associated with premature closure of the epiphyseal plate, and the resultant disturbance in longitudinal growth can cause shortening and angulation of the affected bone (2). The number of limbs that are affected varies from one to all of the extremities, and lower extremity involvement is more common than upper extremity melorheostosis (6). Often the affected limb is shorter, and limb length discrepancies between 1.2 cm and 10 cm have been noted (2, 6). Patients with melorheostosis are also noted to have muscle and bone changes adjacent to the affected bone, and skeletal muscle atrophy has been documented (7). Younge et al (6) noted a woody, firm thickening of the skin in patients with the disorder. The skin may be tense, shiny, or erythematous, and varices are sometimes seen (2).

The pathognomonic radiographic sign of melorheostosis entails irregular, flowing hyperostosis in long bones, and is commonly described as wax flowing down a candle (2–4, 8). The longitudinal arrangement of the hyperostosis is the most important radiographic diagnostic feature of the disease. Children, moreover, exhibit streakiness in long bones, and spotting in the pelvis and small bones of the feet and hands (6). The rate of hyperostosis is more rapid in children, and gradually the rate slows in adulthood (4). Murray and McCreadie (9) explained the linear distribution of skeletal hyperostosis in terms of sclerotomes, which are patterns of skeletal sensory innervation that are analogous to myotomes and dermatomes.

In addition to the typical radiographic presentation of flowing candle wax, Freyschmidt (8) described 3 additional patterns that may be included when diagnosing melorheostosis radiologically. The first is an osteomalike hyperostosis with orientation in the long axis of the involved bone. The lesion must also be 5 cm or larger, involve more than 1 bone, and be eccentrically located. If only 1 bone is involved, other signs such as circumscribed scleroderma or subcutaneous fibrosis above the involved skeletal lesion must be noted. The second pattern demonstrates unilateral, long, and dense hyperostotic striations near the inner side of the cortex in 2 or more bones. The third pattern is similar to periarticular myositis ossificans neuropathica in 2

Financial Disclosure: None reported.
Conflict of Interest: None reported.
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doi:10.1053/j.jfas.2009.08.004

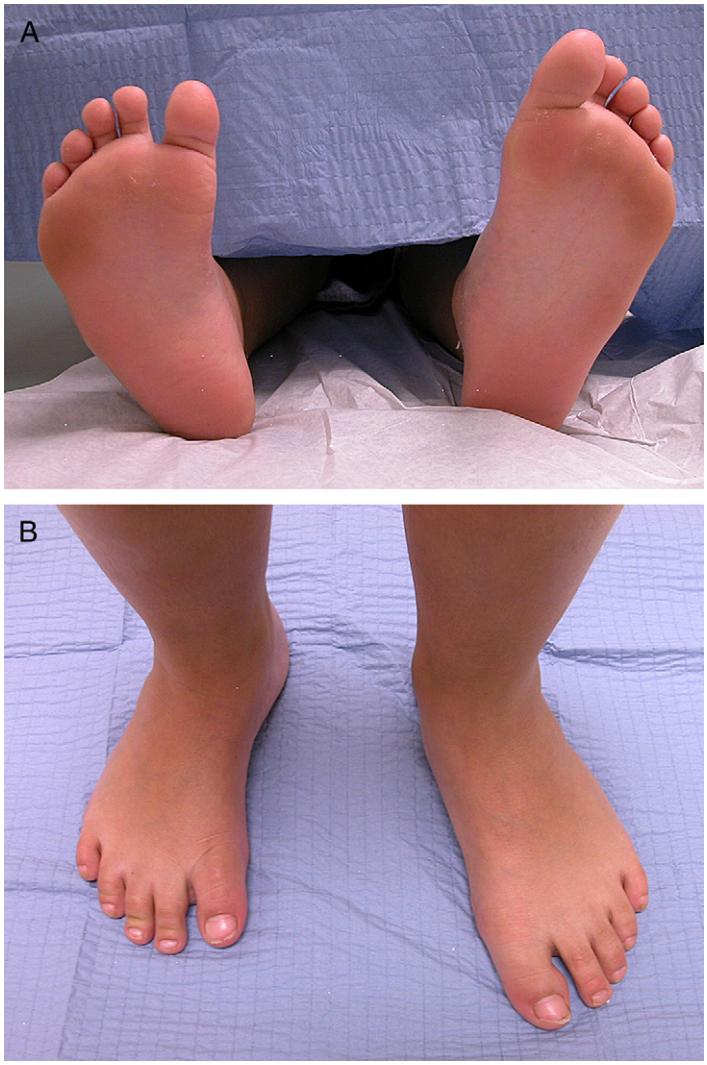

Fig. 1. Preoperative plantar view (*A*) and weight-bearing dorsoplantar view (*B*) of the 10-year-old patient's feet, depicting a C-shaped right foot with metatarsus adductus and melorheostosis.

or more unilateral regions with or without intraosseous hyperostosis. In contrast to classic myositis ossificans, however, the ossifications must be nodularly arranged and should not appear as structured lamellar bone. In addition, there must be no history of direct trauma and/or neurological deficit localized to the region of interest (8).

Grossly, melorheostosis is associated with fibrosis of the skin and subcutaneous tissues adjacent to the affected areas of bone. The muscles of affected limbs are typically edematous and poorly developed. Macroscopic analysis of melorheostosis reveals bone with thickened cortices and trabeculae (2). The histological findings of melorheostosis are considered to be nonspecific (6, 10), and similar to those associated with other hyperostotic bone conditions such as osteopoikilosis (2). Haversian canals are of unequal diameter (3), and the dense trabeculae almost obliterate the Haversian systems (2, 3). If the physis is affected, the epiphyseal plate is replaced with dense bone adjacent to the hyperostotic bone of the epiphysis and metaphysis (2). The medullar bone displays a fibro-fatty marrow (2, 7). Microscopically, the soft tissues about the involved bone display nonspecific fibrosis including perivascular fibrosis with obliteration of blood vessels (6, 7).

There is no definitive diagnostic lab test for melorheostosis, and the results of complete blood count, as well as serum phosphorus and alkaline phosphatase tests are typically normal (3). There has been a report of elevated alkaline phosphatase in a patient with melorheostosis; and this was thought to be associated with an increase in osteoblastic activity (11).

There have been several proposed causes of melorheostosis and the exact mechanism is still unknown. Although a hereditary tendency has been ruled out, the disorder appears to be congenital in nature (3). The linear distribution suggests an embryonic lesion of the somatopleure causing maldevelopment of the limb bud. In addition to the limbs, melorheostosis occasionally affects the spinal column, skull, and ribs; and this pattern suggests maldevelopment of the mesodermal cells of the somites (2). Because of their proximity, some cells of the somites may migrate to the somatopleure before limb bud formation between the fourth and seventh postovulatory week; thus, Campbell et al (2) proposed that the potential for the disease rests in the mesodermal cells before, or at the time of, limb bud formation. Murray and McCredie (9) suggested that melorheostosis may be the late consequence of a segmental sensory nerve lesion that accounts for the sclerotomal distribution of the disorder's hyperostosis. They further proposed that the nerve lesion may be the result of a postnatal peripheral neuropathy in a manner similar to herpes zoster, and the hyperostosis represents nerve root irritation. They concluded that the associated ectopic bone is attributable to the involvement of the corresponding myotome (9). Furthermore, affected skin fibroblasts have been shown to have several cell adhesion proteins with altered expression, and thus, may play a role in the development of the hyperostosis and associated soft tissue abnormalities of melorheostosis (12).

It is interesting to note that bone callus distraction used to correct a limb length discrepancy in a patient with melorheostosis has also given some insight into the possible etiology and mechanism of this disease. The regenerated bone had the appearance of the melorheostotic bone in which the corticotomy had been performed. Marshall and Bradish (13) concluded that because callus formation in callotasis is largely intramembranous, the underlying pathology of melorheostosis is likely related to intramembranous ossification.

Many different conservative and surgical treatments have been used in trying to treat the pain and deformities associated with melorheostosis. Conservative therapies used include oral medications such as bisphosphonates, nonsteroidal anti-inflammatory medications, and nifedipine. Other nonsurgical treatment modalities include physical therapy, manipulations, braces, serial casting, nerve block, and sympathectomies. Surgical procedures used in treating patients with melorheostosis include tendon lengthening, limb lengthening, excision of fibrous tissue, fasciotomies, capsulotomies, osteotomies, excision of hyperostoses, arthrodesis, contralateral epiphysiodesis, and amputation (3). Conservative modalities are typically ineffective when treating limb deformities associated with melorheostosis and surgical treatments often result in recurrence of deformities (3).

Conservatively, various medications have been used to help relieve pain associated with melorheostosis. In one report (11), a patient known to have melorheostosis presented with increased pain in his back and limbs; laboratory tests revealed that the patient had an elevated serum alkaline phosphatase level, indicating an increase in osteoblastic activity. For this reason, the authors treated the patient with an infusion of bisphosphonate for 6 days. Within 1 week there was a decrease in swelling and pain of the right foot, and after 3 weeks all bone and articular pain had decreased and the serum alkaline phosphatase level also decreased. The use of nifedipine, a vasodilator, in a 42-year-old female with an aching, burning pain localized deep in her upper left thigh that limited her activity has also been reported (14). In that report, the patient was successfully treated with nifedipine