Melorheostosis: A Retrospective Clinical Analysis of 24 Patients at an Academic Medical Center

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Abstract

Background: Current understanding of the clinical features of persons with melorheostosis is restricted primarily to individual case reports and small case series.

Objective: To assess the clinical features of patients with melorheostosis treated at our institution from 1972 through 2010.

Design: Chart review.

Setting: Tertiary academic medical center.

Participants: Twenty-three patients with “definite” and one patient with “probable” melorheostosis based on radiographic criteria.

Methods: The eligible study cohort was identified through an index database. Further diagnostic confirmation of patients with melorheostosis was performed by radiographic review.

Main Outcome Measurements: We evaluated age at first visit to our institution, gender, affected body area, number of bones affected, presenting symptoms, surgical evaluation, and therapies provided.

Results: The average age at first evaluation at our clinic was 36.5 years (median 41.5 years, range 3-68 years). The female-to-male ratio was 4:1. The lower extremity was most commonly affected (66.6%), followed by upper extremity (33.3%), spine (16.6%), and head (8.3%). One-third of patients had involvement of a single bone; two-thirds had multiple bone involvement. Pain was the most common presenting concern (83.3%), followed by deformity (54.1%), limitation of movement (45.8%), numbness (37.5%), and weakness (25.0%). Most patients had a physician evaluation (87.5%); patients also underwent orthopedic surgery (45.8%), physical therapy (33.3%), and occupational therapy (12.5%).

Conclusions: Melorheostosis is a rare sclerotic bone disease resulting in pain, deformity, and dysfunction. An interdisciplinary approach to care should include nonoperative and operative evaluation, as well as appropriate therapies. A prospective approach to evaluation, including imaging and physical examinations, would provide valuable longitudinal data.

Introduction

Melorheostosis is a rare disorder of unknown cause characterized by dense sclerotic bone that occurs in a sclerotomal distribution. Leri [1] first described melorheostosis in 1922 as “a deformation of the fingers characterized by a slight spacing of the extremities of the index and medius of the left hand” in a 39-year-old woman [1-5].

The term melorheostosis is derived from the Greek roots melos (limb), rhein (flowing), and ostosis (bone formation). Classically, the disease is characterized by thickening of cortical bone resembling dripping candle wax [6]. The characteristic distribution is sclerotomal, which means that the abnormality follows the osseous innervation pattern. Melorheostosis can present at any age but typically appears in late childhood or adolescence [4] and historically affects both genders equally. Deformity, contracture, pain, stiffness, and limited range of motion are signs that can herald the presence of affected bone. Overlying skin and soft tissue are often involved, showing thickening, shininess, fibrosis, erythema, linear scleroderma, hyperpigmentation, edema, fibrosis, and a scleroderma-like appearance [7].
Melorheostosis typically affects long bones (Figure 1). The lower extremities are affected more often than upper extremities, although small bones of the hand and the foot also may be affected. Rarely, the skull and axial skeleton are affected. Melorheostosis often has a diagnostic appearance on radiographs, with flowing cortical sclerosis in a sclerotomal distribution; however, it can mimic other conditions radiographically. Myositis ossificans, osteoma, and parosteal osteosarcoma can have an appearance to that of melorheostosis. Melorheostosis may be associated with other sclerotic bone dysplasias, including osteopoikilosis and osteopathia striata.

Associations also have been made with scleroderma and Buschke-Ollendorff syndrome, an asymptomatic condition characterized by skin growths (connective tissue nevi) and bone abnormalities primarily in the epiphysis of trabecular bone [4,7].

The cause of melorheostosis is unknown. Initially, investigators postulated that a defect in the \textit{LEMD3} gene caused the disease. Research thus far has not determined a conclusive link to this gene, however. Hellemans et al [8] found that only 16% of patients with sporadic melorheostosis had an identifiable \textit{LEMD3} loss of function, suggesting that a unifying cause continues

Figure 1. Examples of areas affected by melorheostosis.
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