



## What is the Diagnosis?

<sup>1</sup>Mathew D Crawford MD, <sup>2</sup>William C Eward DVM MD

### ABSTRACT

Melorheostosis is a rare, benign connective tissue disorder affecting both bone and soft tissues. Its insidious and variable presentation makes it a challenging clinical diagnosis. We describe the case of a patient presenting with monomelic involvement including significant contractures of the left upper extremity. This case illustrates the functional morbidity associated with melorheostosis as well as its characteristic (i.e. radiographs with flowing hyperostosis) and more rare (i.e. local gigantism) findings.

**Keywords:** Melorheostosis, Hyperostosis, Skeletal dysplasia.

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### CASE REPORT

DO is a 58-year-old right-hand dominant man who sustained a left comminuted distal third clavicle fracture 5 weeks ago. He presents to an orthopaedic oncology clinic for evaluation of multiple calcific bodies around his left humeral head and sclerotic changes of the scapula and humerus. The patient has been treated nonoperatively in a sling. He has pain at the site of his fracture but does not report any shoulder pain.

On examination of his left upper extremity, the patient has limited shoulder range of motion secondary to pain at the site of the clavicle fracture. He also reports mild baseline limitation of movement in the shoulder. The patient has a 30° flexion contracture of the elbow with only 30° of active flexion. The patient has 20° of volar wrist flexion, no wrist extension, and no supination/pronation. He has significant hypertrophy and bony overgrowth of the index and long fingers with no functional movement of the IP or MP joints of these fingers.

The patient states that he has had limited range of motion of his elbow, wrist and hand since he was a child and that the bony overgrowth and functional

limitation of his fingers has progressively worsened over the years. The patient works in sales and although he has had cosmetic concerns regarding his fingers, he has not pursued surgical intervention as the fingers have not caused him significant pain. The patient had a prior fracture of his left clavicle but otherwise has no relevant medical history. His sister has scoliosis but his family history is also unremarkable. Radiographs of his entire left upper extremity were obtained at this visit.

He says that he was once told he had osteopetrosis. Does that seem correct? What is your diagnosis?

### DISCUSSION

The patient's radiographs show irregular hyperostotic cortical changes of the humerus, radius and ulna. There are periarticular nodular calcifications in the shoulder and elbow as well as patchy, mottled ossifications of the proximal forearm, distal radioulnar joint, carpus and second/third digits (Figs 1, 2, 4 and 5). These findings in combination with his clinical presentation of asymmetric joint contractures and increased extremity circumference with overlying skin changes (Fig. 3) are hallmarks of melorheostosis.

Melorheostosis is a rare, benign connective tissue disorder affecting both bone and soft tissues. Best regarded as a skeletal dysplasia, it has an estimated prevalence of 0.9 cases per million affecting men and women equally. It was first described in 1922 by Léri and Joanny as a linear pattern of hyperostosis along the major axis of long bones.<sup>1-4</sup> It has no associated mortality but the functional morbidity can be considerable as it is associated with progressive joint and soft-tissue contracture with chronic pain common in later stages of the disease.

The clinical presentation of melorheostosis is insidious and the symptoms are variable. These commonly include dull pain, decreased joint motion, limb swelling, and paresthesias over the involved tissues. Deformities consist of soft-tissue contractures and increased extremity circumference (with decreased bone length more common than increased length). The overlying skin may be tense, shiny, fibrosed or covered with varices. The disorder can involve one bone (monostotic), more than one bone (polyostotic), one extremity (monomelic) or present in a more generalized fashion. Although it can occur in any location, lower extremities are more commonly involved than upper extremities. Symptoms first

<sup>1,2</sup>Department of Orthopaedic Surgery, Duke University Medical Center, Durham, NC, USA

**Corresponding Author:** Mathew D Crawford, Department of Orthopaedic Surgery, Duke University Medical Center, Box 3000 Durham, NC 27710, USA, e-mail: mathew.crawford@duke.edu





**Fig. 1:** Anteroposterior view of the left shoulder demonstrating a distal 1/3 clavicle fracture with multiple calcific bodies around the shoulder joint



**Fig. 2:** Full length view of the left humerus which shows calcific bodies around the left elbow in addition to those seen at the shoulder



**Fig. 3:** Clinical photograph demonstrating significant hypertrophy of the patient's left index and long fingers



**Fig. 4:** Posteroanterior view of the left forearm demonstrating calcification around the elbow, proximal forearm, wrist and hand

manifest in late childhood or early adolescence and progress into adult life.<sup>3,4</sup> However, it can also be discovered as an asymptomatic, incidental finding.

The characteristic radiographic appearance consists of irregular, flowing hyperostosis in long bones, often described as melted wax dripping down a candle. This appearance gives the anomaly its name, which is taken from the Greek nomenclature—melos (limb), rhein (flow), osteon (bone). Periarticular soft-tissue ossifications are also common.<sup>5</sup> Freyschmidt<sup>6</sup> defined an array of radiographic patterns to better characterize the scope of presentation. These include the classic pattern of flowing candle wax on the inner and/or outer surface of bone; an osteoma-like hyperostosis that is eccentric, >5 cm, and involving more than 1 bone; an osteopathia striata-like pattern with unilateral, long, and dense hyperostotic striations near the inner side of the cortex in multiple bones and a pattern similar to periarticular myositis



**Fig. 5:** Posteroanterior view of the left hand demonstrating significant calcification in the distribution of the hypertrophy of the index and long fingers

ossificans but with ossifications in a nodular arrangement that do not appear like structured lamellar bone and with no history of trauma. In this patient's case, the lesions

present in the shoulder joint may represent myositis ossificans or they may represent a second problem—synovial chondromatosis.

The etiology of melorheostosis is unknown. A few theories include a vascular disturbance, inflammatory process or endocrinopathy. Murray and McCredie<sup>7</sup> suggested that the peculiar monomelic and linear hyperostotic involvement of melorheostosis may result from an embryonic insult to a spinal sensory nerve, producing bone scarring in a skeletal distribution (scleroterm). Freyschmidt<sup>6</sup> proposed that the sporadic occurrence, variable extent of disease, and equal gender ratio is better explained by a lethal gene that survives only in a mosaic state. Recent studies<sup>8</sup> have reported that loss of function mutations in the LEMD3 gene, which encodes an inner nuclear membrane protein, may be involved in the pathogenesis of melorheostosis and osteopoikilosis.

Melorheostosis is difficult to treat and has led many to advocate training patients to use the unaffected extremity in a dominant fashion. Treatment is symptomatic and the primary goals are pain relief and restoration of full joint range of motion. Conservative management includes analgesia, manipulation, braces, serial casting, PT, nerve blocks and sympathectomy. Surgical management consists of soft tissue release, capsulotomy, fasciotomy, tendon lengthening, corrective osteotomy, excision of hyperostotic bone, arthroplasty, ilizarov lengthening, contralateral epiphysiodesis, arthrodesis and amputation.<sup>3,4</sup> While conservative measures are often ineffective, recurrences are very common after surgical management.<sup>2,9</sup> Because melorheostosis is a progressive condition, surgical intervention in children should be delayed until after skeletal maturity to avoid additional procedures.<sup>9,10</sup> Melorheostosis exhibits a slow and chronic course and its prognosis is variable and depends on location as well as soft-tissue extension. Although most cases of melorheostosis are not associated with local gigantism, in some cases

melorheostosis causes local gigantism, and this is almost certainly the cause of DO's macrodactyly.

DO's monomelic presentation is late in the course of the disease as he has significant soft tissue involvement with elbow and wrist joint contractures and overgrowth of the carpus and second and third digits. His radiographs show both the classic appearance of flowing candle wax and periarticular nodular ossifications similar to myositis ossificans. As DO has lived with this disorder for most of his life and has no associated pain, he is not interested in pursuing any treatment.

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