Melorheostosis (MEL) is a rare benign congenital mesodermal disease. The disease can occur at any age and sex. The most common affected sites are the lower and upper limbs. It is characterized as hyperostosis of cortex. Its most prominent clinical features are articular pain, contracture, ossification of the soft tissue and articular limitation of motion. The diagnosis of MEL is usually based on clinical evaluation and the finding of the characteristic radiographic abnormalities. Bone scintigraphy can help to distinguish a focus of MEL from other lesions (osteopoikilosis, osteopatika striata, osteogenic sarcoma, and malignant fibrous histiocytoma) and monostotic, polystotic, or monomelic disease. Here, we present two cases with MEL which had also been evaluated by bone scintigraphy and its contribution to the definition of extension of the disease as monostotic, polystotic, or monomelic.

**Key Words:** Melorheostosis; radionuclide imaging; radiography


**Anahit Kelimeyer:** Meloreostoz; radyonuklit görüntüleme; radyografi


Bone Scintigraphy in Melorheostosis: Case Report

Melorheostosis'te Kemik Sintigrafisi

**ABSTRACT** Melorheostosis (MEL) is a rare benign congenital mesodermal disease. The disease can occur at any age and sex. The most common affected sites are the lower and upper limbs. It is characterized as hyperostosis of cortex. Its most prominent clinical features are articular pain, contracture, ossification of the soft tissue and articular limitation of motion. The diagnosis of MEL is usually based on clinical evaluation and the finding of the characteristic radiographic abnormalities. Bone scintigraphy can help to distinguish a focus of MEL from other lesions (osteopoikilosis, osteopatika striata, osteogenic sarcoma, and malignant fibrous histiocytoma) and monostotic, polystotic, or monomelic disease. Here, we present two cases with MEL which had also been evaluated by bone scintigraphy and its contribution to the definition of extension of the disease as monostotic, polystotic, or monomelic.

**Key Words:** Melorheostosis; radionuclide imaging; radiography


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Melorheostosis (MEL) is a rare benign congenital mesodermal disease. It is included in the “sclerotic bone dysplasias” along with other sclerosing disorders such as cranio-methaphyseal and diaphyseal dysplasias as well as the osteoscleroses: osteopetrosis and its “variants” pyknodysostosis, osteopoikilosis, osteopathia striata.1

It was first reported by Leri and Joanny in 1922.2 MEL is characterized by a “flowing” hyperostosis of cortex, which looks like wax dripping down one side of the candle3 and this appearance also gave the name of this anomaly, which is taken from the Greek words for member (melos) and flow (rhein).4
The disorder can occur in both sexes and at any age. The most affected sites are the lower and upper limbs. Its most prominent clinical features are articular pain, contracture, ossification of the soft tissue and articular limitation of motion. The diagnosis of MEL is usually based on clinical evaluation and finding of the characteristic radiographic abnormalities. The typical radiographic image presents resembling molten wax flowing down one side of a candle. Bone scintigraphy can help to distinguish a focus of MEL from other lesions and monostotic, polyostotic, or monomelic disease. Accumulation of bone-seeking radiopharmaceuticals with medullary cavity preservation may help differentiate MEL from osteopoikilosis and osteopathica striata, which do not show increased activity. This accumulation also helps differentiate among aggressive malignancies, which invade the cortex and cancellous bone.

Here, we present two cases with MEL which had also been evaluated by bone scintigraphy.

**CASE 1**

A 47-year-old man presented with a 3-year history of left upper limb swelling and pain. He described the pain as ensuing after writing and using his hand for an extended time but not during routine daily activities or at rest. Physical examination was normal except for swelling and pain on the dorsal side of the first metacarpal bone during resistive dorsal flexion of the thumb.

The plain radiograph showed massive sclerotic changes and the typical candle-wax-like thickening of the left distal humerus, the left radius, the left first and second metacarpals and phalangeal bones (Figure 1). The left radius and hand in particular, as well as the radiograph of the humerus led to a diagnosis of suspected melorheostosis.

The laboratory findings (blood calcium, phosphate levels and alkaline phosphatase) were in normal range.

Magnetic Resonance Imaging (MRI) study revealed osteosis at the left radius, left first metacarpal and phalangeal bones with hypointensity on all pulse sequences, findings suggesting MEL (Figure 2).

Three phase bone scintigraphy revealed high vascularity in dynamic and blood pool phases and increased activity at the left distal humerus, left elbow, left radius, carpal bones on the radial side and first and second metacarpals and phalangeal bones in static images (Figure 3).
A 34-year-old woman presented with a 10-year history of right-forearm pain. She described the pain during all routine daily activities or rest. Physical examination showed swelling and pain on the dorsal side of the fifth metacarpal bone.

The plain radiograph showed massive sclerotic changes and typical candle-wax-like thickening of the right distal ulna (Figure 4). Diagnosis of MEL was established, and although a three phase bone scintigraphy showed normal vascularity in dynamic and blood pool phases, it supported the diagnosis by increased uptake in the areas of radiographic hyperostosis in delayed images (Figure 5).

The laboratory findings (blood calcium, phosphate levels and alkaline phosphatase) were in normal range.
DISCUSSION

MEL is a rare hyperostotic, non-genetic, benign sclerosing bone displasia of unknown etiology. Several models for the pathogenesis of MEL have been proposed. Murray and McCredie suggest that the disease is a consequence of segmental sensory nerve damage. Other theories propose a vascular disorder, inflammation, a degenerative lesion of the connective tissue and embryonic damage.

Apart from pain, typical symptoms include tendon and muscle contracture, limitation of joint motion and ossification of the soft tissue. In the present cases, the main symptom was pain. Although MEL can affect any bone, the lower extremities are more frequently involved. Most reported cases involving the upper extremity are focused on the hand. The localization of the dis-

FIGURE 4: Plain of the right forearm of Case 2. The right distal ulna shows massive sclerotic changes.

FIGURE 5: Bone scintigraphy of Case 2, normal dynamic-blood pool phases (a) and extensive uptake of 99mTc-MDP in right distal ulna in static image (b) and whole body image (c) (monostotic type of MEL).
ease was also in the upper extremity in our both cases. MEL of the hand has been associated with bony spur formation and is complicated by an inflamed bursa. Other symptoms such as stiffness, swelling, numbness, tingling, carpal tunnel syndrome, and a slowly growing desmoid tumor of the soft tissues of the hand have also been reported. Involvement can encompass one bone (monostotic), more than one bone (polystotic), or one extremity (monomelic). In the present cases, case 1 was polystotic (left humerus, radius, carpals) and case 2 was monostotic (right ulna).

The classic radiographic feature of MEL is asymmetrical bands of sclerosis in an irregular, linear pattern often described as molten wax dripping down from one side of a candle. Radiological studies reveal cortical and endosteal hyperostosis extending to one side of the long tubular bones. In our both cases, the plain radiograph showed massive sclerotic changes and typically candle-wax-like thickening in the affected bones.

Radionuclide bone scanning is a useful method to distinguish a focus of MEL from other lesions. Accumulation of bone-seeking radiopharmaceuticals with medullary cavity preservation may help differentiate melorheostosis from osteopoikilosis and osteopathica striata, which do not show increased activity. This accumulation also helps differentiate among aggressive malignancies, which invade the cortex and cancellous bone. In MEL, focal increased radiopharmaceutical accumulation appeared in each radiographically abnormal area on bone scintigraphy. The factors responsible for uptake may include increased mass of the cortex, osteoblastic activity, local hyperemia, presence of immature collagen, and changes in capillary permeability. While our first case showed increased activity on all three phases, second case did not show markedly increased vascularity at dynamic and blood pool images. This finding could be due to the duration of the disease. The duration of the disease in the second case is longer than the first case (10 years vs 3 years). In addition, the intensity of inflammation might be different in the cases.

MRI is very useful in understanding soft tissue patho-anatomy and for preoperative planning. On MRI, there is decreased signal intensity localized to affected bone on all pulse sequences. Although MRI appearance of soft tissue masses associated with MEL is variable, advanced imaging allows visualization of mineralized and nonmineralized soft tissue. MRI of case 1 also showed hypointensity in all pulse sequences at the affected bones.

There is no specific treatment for MEL. Therapy is mainly symptomatic. Patient symptoms vary considerably in MEL, and consequently treatment should be individualized depending on the age and location. Surgery is possible to correct bone deformities and asymmetric bone growth. Surgery has been planned for our cases since the symptoms still continue and non specific medical therapy was ineffective.

In conclusion, MEL is a rare benign congenital mesodermal disease which is included in the “sclerotic bone dysplasias”. Although the diagnosis is usually based on clinical evaluation and the finding of the characteristic radiographic abnormalities, bone scintigraphy is useful to define the extent of the disease as monostotic, polystotic, or monomelic.

REFERENCES


20. By Vijay Kumar Jain, MS; Rajendra Kumar Arya, MS; Minakshi Bharadwaj, MD; Satish Kumar, MS. Melorheostosis: Clinicopathological Features, Diagnosis, and Management. Orthopedics 2009; 32:512.


