Extensive melorheostosis of the upper extremity: Evaluation of disease activity with ^{99m}Tc-MDP bone

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Abstract
Melorheostosis is a rare, sporadic sclerosing bone dysplasia characterized by a "dripping candle wax" appearance on imaging. We report a case of a 29-year-old male with a 20-year progressive history of bone overgrowth and joint contracture in the left upper extremity. Technetium-99m-methylenediphosphonate (^{99m}Tc-MDP) bone scintigraphy demonstrated intense tracer uptake, indicating high metabolic activity despite the long-term disease course. Three-dimensional computed tomography (CT) reconstruction visualized extensive hyperostosis crossing multiple joints, while magnetic resonance imaging (MRI) revealed associated soft tissue involvement. The diagnosis was confirmed by biopsy. This case highlights the value of multimodality imaging in assessing the extent and activity of this rare condition to guide surgical intervention.

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Introduction

Melorheostosis, first described by Léri and Joanny in 1922, is an exceedingly rare osteosclerotic bone disease with an estimated incidence of 0.9 per million [1, 2]. It typically follows a sclerotomal distribution and is characterized by mesenchymal product overgrowth. Although benign, it can lead to severe pain, deformity, and functional impairment. While the diagnosis is primarily based on characteristic radiographic features, nuclear medicine imaging plays a crucial role in evaluating the metabolic activity of the lesions, which is vital for clinical management and surgical planning.

Case Presentation

A 29-year-old male presented with a 20-year history of progressive deformity and restricted movement in his left upper limb. The symptoms began insidiously in the left fifth finger during childhood and gradually extended to the forearm and wrist over two decades. There was no associated pain or prior trauma. Physical examination revealed multiple hard, bony prominences along the ulnar aspect of the left forearm and hand. Joint mobility was severely compromised: elbow extension was limited to 30°-40°, wrist dorsiflexion was 0°, and the proximal interphalangeal joints of the fourth and fifth fingers were ankylosed. Muscle strength remained Grade V, and no neurological deficits or skin pigmentations were noted.

Laboratory investigations, including parathyroid hormone (PTH), rheumatoid factor (RF), antinuclear antibodies (ANA), and alkaline phosphatase (ALP) were within normal limits. An upper extremity arteriovenous ultrasound was normal, ruling out associated vascular malformations.

Plain radiography (Figure 1A) and 3D computed tomography (CT) reconstruction (Figure 1B and 1C) demonstrated extensive, undulating cortical hyperostosis extending from the medial humeral epicondyle to the 5th digit phalanges. This classic "dripping candle wax" appearance involved the ulna and ulnar-sided carpal and metacarpal bones, crossing joint spaces and causing mechanical fusion. Coronal T1-weighted magnetic resonance imaging (MRI) (Figure 1D) demonstrated the flowing hyperostosis as signal voids along the ulna and carpus. Axial short tau inversion recovery (STIR) fast spin echo (FSE) MRI (Figure 1E) revealed thickened cortices and significant high-signal swelling

in the surrounding soft tissues, elucidating the severe functional impairment.

A technetium-99m-methylenediphosphonate (^{99m}Tc-MDP) whole-body bone scintigraphy (Figure 2) showed intense, asymmetric radiotracer accumulation along the entire length of the involved skeletal segments in the left upper limb. The intense uptake confirmed highly active osteoblastic metabolism within the sclerotic lesions.

Due to the severe functional impairment, the patient underwent an open biopsy and subsequent surgical intervention, including partial excision of the bony lesions and joint release of the elbow and wrist. Histopathological examination revealed marked hyperplasia and hypertrophy of the compact bone, narrowing of the medullary cavity with internal fibrous tissue proliferation, and surrounding cartilage hyperplasia, confirming the diagnosis of melorheostosis.

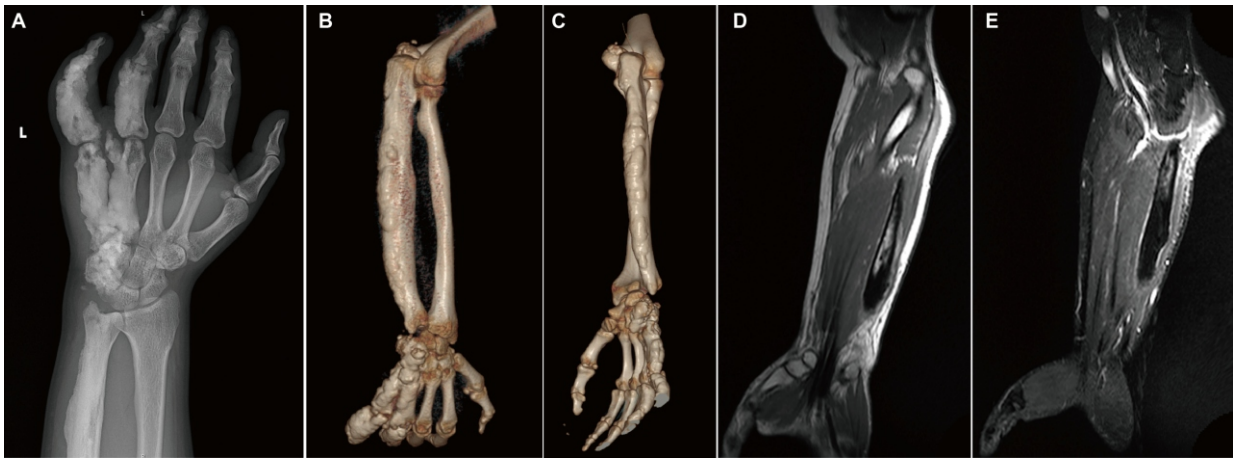

Figure 1. Plain radiograph, 3D CT reconstructions, and MRI of the left upper extremity. (A) Plain radiograph showing extensive undulating cortical hyperostosis with a classic "dripping candle wax" appearance along the ulnar aspect. (B, C) Three-dimensional CT reconstructions demonstrating hyperostosis extending from the medial humeral epicondyle to the 5th digit phalanges, crossing multiple joint spaces. (D) Coronal T1-weighted MRI showing the flowing hyperostosis as signal voids along the ulna and carpus. (E) Axial STIR FSE MRI revealing thickened cortices and high-signal soft tissue swelling surrounding the involved segments.

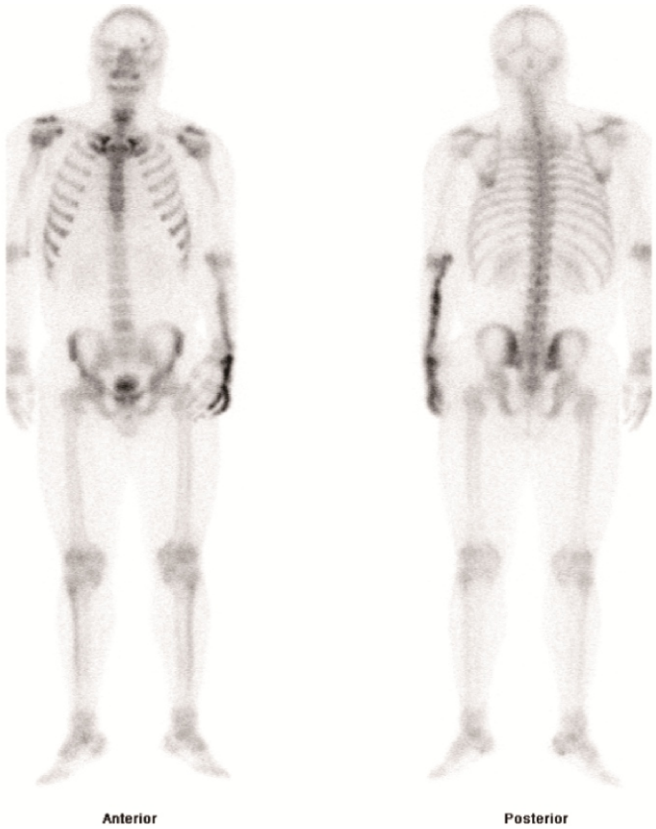

Figure 2. Whole-body bone scintigraphy (anterior and posterior views) following intravenous administration of ^{99m}Tc-MDP. Intense, asymmetric radiotracer accumulation is demonstrated along the entire length of the left upper limb, extending from the shoulder to the hand, confirming highly active osteoblastic metabolism within the sclerotic lesions.

Discussion

Melorheostosis is a rare clinical entity often presenting with a classic "dripping wax" sign on plain radiography or CT. Its pathogenesis has recently been linked to somatic mutations in the MAP2K1 gene [3], which leads to increased transforming growth factor-β (TGF-β) signaling and bone overgrowth [4, 5]. This case is remarkable for its 20-year progression and the extent of involvement, crossing multiple joints from the humerus to the hand. The normal ALP level, despite intense bone tracer uptake, underscores the focal rather than systemic nature of this metabolic activity.

Additionally, the increased T2 signal in the surrounding soft tissues on MRI elucidates the severe periarticular symptoms and joint contractures, demonstrating that the disease affects both bone and adjacent soft tissues. The normal vascular ultrasound also assisted in differentiating this condition from Klippel-Trenaunay syndrome [6].

In such chronic cases, bone scintigraphy is particularly valuable. While CT provides exquisite anatomical detail of the "wax-like" flow, ^{99m}Tc-MDP uptake reflects the current rate of bone turnover. The intense uptake observed in this patient indicates that the disease remained metabolically active even after two decades, providing a biological rationale for the continued clinical progression and justifying the decision for surgical salvage and joint release. The primary differential diagnoses include osteopoikilosis and osteopathia striata; however, the asymmetric, flowing cortical distribution and intense tracer uptake are highly specific for melorheostosis. For patients with significant joint restriction, surgical intervention aims to debulk the overgrowth and restore mobility, although the risk of recurrence remains [7].

In conclusion, the integration of 3D CT, MRI, and bone scintigraphy is essential for the comprehensive evaluation of melorheostosis. Bone scintigraphy, by demonstrating disease activity, complements anatomical imaging and assists in the clinical decision-making process for these rare and challenging skeletal dysplasias.

The authors declare that they have no conflicts of interest.

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