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Authors

Silver, Matthew
Thomas, Cyril
Montgomery, Erica

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One in a Million: A Case of Arm and Leg Pain and Deformity

Matthew Silver, MD*
Cyril Thomas, MS PA-C*
Erica Montgomery, PA-S†

*Kaiser Permanente, Department of Emergency Medicine, San Diego, California
† University of Southern California, Keck School of Medicine, Los Angeles, California

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A 38-year-old Hispanic woman with no known past medical or family history presented to the emergency department with severe, intractable left upper and lower extremity pain and inability to walk for 2 days. The woman reported a history of chronic, progressive left hand, arm, and leg deformity over the previous 2 years with episodic flares of severe pain. The woman had not reported any trauma or systemic symptoms. Physical exam revealed deformity and hypertrophy of the second and third digits of the left hand (Figure 1), the left elbow, left thigh, and lower aspect of her left leg without significant joint swelling, warmth or redness. She exhibited significantly limited and painful range of motion at the joints in the left upper and lower extremity. Plain radiographs were obtained (Figure 2).

DIAGNOSIS

Melorheostosis was first described by Leri and Joanny in 1922.¹ It is a rare osteosclerotic bone dysplasia caused by a non-



Figure 1: Photograph of the patient's left hand demonstrating marked deformity and apparent swelling of the 2nd and 3rd digits and metacarpal bones.



Figure 2: (A) Radiograph of the left hand showing dense sclerosis, lengthening, and expansion of the second and third phalanges and metacarpals with increased density of the lunate, capitate, and trapezoid and, (B) radiograph of the left proximal femur showing diffuse dense cortical thickening with characteristic "flowing candle wax" appearance.

inheritable, developmental error in the LEMD3 gene.² Its incidence is about 0.9 per million and affects males and females equally.³ The pattern of the dysplasia generally follows a sclerotomal distribution, representing the zones of the skeleton supplied by individual spinal sensory nerves, and is typically asymmetric involving the extremities and rarely the axial skeleton. The name melorheostosis is derived from its characteristic "melting wax flowing down a candlestick" appearance on radiologic exam.³

Common presenting symptoms include pain, edema, stiffness, and deformity. The treatment of melorheostosis includes symptomatic treatment for pain, surgery to correct deformities, and amputation if necessary.

Our patient was admitted to the hospital for pain management. Two days later she was discharged to a skilled nursing facility for continued physical therapy, occupational therapy, and medical management.

Address for Correspondence: Matthew A. Silver, MD, Kaiser Permanente, San Diego Medical Center, San Diego, CA. Email: Matthew.a.silver@kp.org.

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