

Classic Presentation of a Rare Disease: Melorheostosis

PETROS EFTHIMIOU, MD, Assistant Professor of Clinical Medicine, Weill Cornell Medical College, New York, New York; OURANIA NASIS, MD, New Jersey Medical School, University Hospital, Newark, New Jersey; LINDA A. RUSSELL, MD, Assistant Professor of Clinical Medicine, Weill Cornell Medical College, Department of Medicine, Hospital for Special Surgery, 535 East 70th Street, New York, NY 10021, USA. Address reprint requests to Dr. P. Efthimiou. E-mail: Petros.Efthimiou@nychhc.org

A 49-year-old Caucasian woman with a history of melorheostosis since childhood presented with decreased range of motion and chronic pain in her left leg and knee after a fall that injured her left knee. Examination was notable for evidence of skin tightening over the knee and lower leg with no frank sclerodermatous changes. There was also evidence of bony indurations as well as venous varicosities over the left lower leg. Left knee examination revealed flexion contracture of about 15 degrees. Plain radiographs of the knee were taken that revealed “flowing” hyperostosis of the cortex, resembling wax dripping down a candle (arrows in Figure 1A, 1B). This finding is classic for melorheostosis, which is a rare mesodermal disease of poorly understood etiology that affects the skeleton and adjacent soft tissues. The term melorheostosis is derived from the Greek *melos*, meaning limb, and *rheos*, flow¹. Melorheostosis, also known as Leri’s disease, is one of a group

of sclerosing bone dysplasias that affects mostly the long bones of the upper and lower extremities. Patients can present at any age with a variety of symptoms, including pain, limb swelling, and limitations in motion of affected limbs. Patients may develop flexion contracture of hips or knees, and skin overlying an area of melorheostosis in the bone may appear tense, shiny, or erythematous. There is also an association with vascular lesions such as varices, hemangiomas, or aneurysms². This case is a classical presentation of melorheostosis.

REFERENCES

1. Freyschmidt J. Melorheostosis: a review of 23 cases. *Eur Radiol* 2001;11:474-9.
2. Greenspan A, Azouz EM. Bone dysplasia series. Melorheostosis: review and update. *Can Assoc Radiol J* 1999;50:324-30.



A

Figure 1. Radiographs of the knee reveal flowing hyperostosis of the cortex (arrows in A and B).



B

Figure 1B. Arrows indicate flowing hyperostosis of the cortex.