

MELORHEOSTOSIS

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Abstract

This case study is a report on the rare mesodermal disorder typically characterized by abnormalities of the skeleton and soft tissues. We present a 56 years old woman who was diagnosed with melorheostosis affecting the left arm. Chronic pain odema and cosmetic deformities were her presenting problems. Melorheostosis is a bony dysplasia with characteristic X-ray appearance resembling wax dripping down one side of the candle. Soft tissue calcification and even ossification may rarely be seen. In some rare and complicated cases corrective surgery or amputation may be done in very painful and ischemic limbs (1). Until very recently the aetiology of melorheostosis was unknown but now it has been established that melorheostosis is due to a loss-of-function mutation in LEMD 3 gene (also called MAN 1), which encodes an inner nuclear membrane protein (2). This is the first reported case in this region. The purpose of this case report is to describe the presentation and course of the disease. A comprehensive review of literature describing etiology, clinical aspects, diagnosis and treatment is included. Patients symptoms vary considerable in melorheostosis and consequently their treatment should be individualized.

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