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A rare case of monomelic melorheostosis

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Abstract

Melorheostosis is a rare entity belonging to the group of sclerotic bone dysplasias. Described for the first time in 1922 by Leri, it remains imperfectly known as clinical presentations are highly variable, and the etiological diagnosis is not fully elucidated. Here we present a rare case of melorheostosis of left upper limb involving left ulna and humerus extending to metacarpals and phalanges. In our case the disease has involved 2 long bones in same extremity diffusely.

Keywords: monomelic melorheostosis

1. Introduction

Melorheostosis is a rare benign non hereditary condition. It is known as a mesoderm sclerotic bone dysplasia, characterized by cortical hyperostosis with or without retraction of soft tissue. It was described for the first time in 1922 by Leri and Joanny as a dripping candle wax hyperostosis [9]. Since then, 300 cases have been reported in the literature. Its incidence is estimated 0.9 per million population. It affects both men and women at any age. The locations are highly variable, and mono ostotic forms were more often described. Polyostotic forms localized to the lower limbs are rare. The disease can remain silent and be discovered incidentally. If not, symptoms are made of distortion, variable pain or limitation of joint mobility. It is a disease whose etiology is imperfectly understood, the genetic factors and metabolic predisposition or malformation of the vessels are proposed, but the exact cause is to be determined [5, 6]. In our case the disease has involved 2 long bones of same extremity

Case Report

A 45-year-old man presented with a history of left upper extremity pain and swelling for 38 years and limitation of articular motion for 30 years. His limb pain initially appeared at the age of 6 years, most prominently in his left elbow. After middle school, the swelling and restriction of joint motion gradually progressed. During the previous 10 years, he often felt anesthesia in the ulnar areas of the left forearm and hand. There was no relevant family history or trauma, and his son had no similar problems.

On physical examination, the patient had hypertrophy of left upper limb. The circumferences of the left upper arm and forearm were 15 and 9 cm larger than those of the right side (Fig. 1A) with focal hypertrophy 5 cm proximal to the wrist near the ulnar border. The swelling was bony hard and nontender. High skin temperature and hyperpigmentation also were present. The joints below the left shoulder were completely or near-completely stiff. There was about 20° flexion of the shoulder, 80° abduction terminal restriction of internal and external rotation in neutral and abduction. There was fixed flexion deformity of the elbow of about 80°, 20° flexion deformity of the wrist with restricted dorsiflexion. He had decreased sensation in the ulnar half of the forearm and hand. Tinel's sign was positive around the territory of the sulcus for the ulnar nerve. No abnormality was detected in the right upper limb.

Plain radiographs (fig. 2A & 2B) showed extensive, dense, undulating or irregular cortical hyperostosis, resembling candle wax, extending along the length of both sides of left ulna and humerus including the carpal bones, central metacarpal and phalanges of middle finger, resulting in deformity of the bones and narrowing or disappearance of the medullary cavities. There was no distinct demarcation between the affected and normal bones. Plain radiographs of bones on the right side were normal.

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Laboratory findings for serum calcium, phosphorus, alkaline phosphatase, C-reactive protein, erythrocyte sedimentation rate were within normal limits. The pathology report described nonspecific, dense cortical bone. The hyperostotic bone consisted of mature and immature bone elements.



Fig. 1A



Fig. 2A



Fig. 2B

Discussion

Melorheostosis is a rare chronic bone disorder first described in 1922 by Leri and Joanny [9]. Men and women are equally affected, and no hereditary features have been discovered. The onset is insidious, and the first symptom of neuralgia or arthralgia usually occurs in childhood or early adolescence. Range-of-motion limitation and joint deformities develop gradually [1]. Our patient had initial onset of pain in the left upper limb at the age of about 6 to 8 years.

Melorheostosis may present in a monostotic, polyostotic, or monomelic form. The monomelic variant is the most common presentation of the disease [16]. In our patient, the entire left upper extremity was involved and the right extremities were completely normal.

Several hypotheses have been suggested concerning its etiology, such as an embryonic mesodermal disorder that affects osseous and soft tissue, a loss-of-function mutation in LEMD3 gene (sometimes referred to as MAN1), a vascular disturbance, and an inflammatory process [5, 15]. In 1979, Murray and McCredie [13] suggested an infection associated with nerve roots may be responsible for melorheostosis, because they detected a good correlation between hyperostotic lesions and the sclerotomes. This may in part explain the monomelic and linear track involvement and distribution of melorheostosis. Another possible explanation is involvement of the corticospinal tract or spinothalamic tract, but then the symptoms in the axial skeleton or nervous system also should be apparent. The etiology of melorheostosis remains obscure. Radiographic findings have been the cornerstone of the diagnosis, consisting of candle wax-like hyperostotic changes of the cortex, generally on one side of the bone. There is usually a distinct demarcation between the affected and normal bone. Dense linear areas are seen mainly in the cortex but also extending into the cancellous bone. Melorheostosis affects mainly the long bones of the upper and lower limbs, and also the short bones of the hand and foot, but rarely the axial skeleton [4]. In our patient, plain radiographs showed the classic radiographic features of melorheostosis, but the severity was unusual, including severe thickening and deformity of the bones. Almost all the bones and joints in the left upper limb was affected, every bone was extensively affected, and there was no distinct demarcation between the affected and normal bone. The radius and ulna and small bones in the hand were almost completely fused.

Routine laboratory findings usually are normal. Histologic findings are usually nonspecific and often show dense bone formation, a mixture of mature and immature bone elements, and thickened and enlarged bony trabeculae constituting largely primary Haversian systems [3]. Osteoclastic activity is not a prominent feature; however, osteoblastic activity along the margins of osteons is common [2].

The differential diagnosis for melorheostosis, especially for the forme fruste, includes osteopoikilosis, osteopathia striata, myositis ossificans, parosteal osteosarcoma, and osteoma [7, 14, 16]. In our patient, according to the clinical presentation, characteristic appearance of plain radiographs, and histologic features, the diagnosis was easily made.

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