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Case Report

Melorheostosis of the Fibula: A Case Report

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Abstract

Melorheostosis is a relatively rare benign sclerosing bone dysplasia, which is also known as Leri-Joanny disease. Recent genetic research proposed the MAP2K1 oncogene mutation, which leads to this rare disease. The pathology usually manifests with severe pain, dysfunction, and deformity in a single limb. The diagnosis can be made through radiographic evaluation with positive dripping candle wax sign. Management of such condition is mainly symptomatic and not curative. Surgical debulking is indicated to regain functional status and alleviate severe pain.

Keywords: Leri-Joanny disease; Melorheostosis; Sclerotic bone lesion

Introduction

Melorheostosis is a relatively rare benign sclerosing bone dysplasia. The name comes from the Greek terms Melos “limbs”, rheos “flow”, and osteon “bone”. This disorder is typically segmental. [1] It commonly presents in adolescence or early adulthood, affects men and women equally, and has a prevalence of 0.9 per million. [2] A loss of function mutation in the LEMD3 gene “12q12-12q14.3”, a protein of the inner nuclear membrane involved in bone morphogenic protein and tumor growth factor signaling, is one possible cause of melorheostosis. On the other hand, another study found no mutation in three patients with sporadic melorheostosis, hence the precise causes are currently unknown. [1] The most common symptoms in most patients are pain, deformities, restricted range of motion, contractures, muscle atrophy, and limb swelling. [3] Majority of the time, the diagnosis can be made through radiographic evaluation with positive “dripping candle wax” sign. The diagnosis can be supported by analytical normal serum calcium, phosphorus, and alkaline phosphatase. The anatomopathological tests including histological findings are nonspecific, and frequently show a mixture of mature bone in a dense formation with increased trabecular bone. [2] Unsurprisingly, melorheostosis was noticed to be associated with various diseases, including linear scleroderma, rheumatoid arthritis, tuberous sclerosis, and neurofibromatosis. [4] Although the management of such condition is mainly symptomatic and not curative, non-surgical and surgical options are available. Hence, treatment is usually tailored for patient’s clinical presentation and complications. [5] In this case report, we discuss a 30-year- old, Saudi male who experienced sever progressive pain in the right leg over 6 years not relieved by physiotherapy or oral analgesics. Investigations and imaging studies pointed toward the diagnosis of Melorheostosis.

Case Presentation

A 30-year-old Saudi male with no history of medical illness or previous trauma presented to the orthopedic clinic with right leg pain, swelling and limitation of right knee movement for the past 8

years. The swelling and restriction of joint movement progressed gradually. There were no constitutional symptoms or relevant family history. On physical examination, the patient had a tender swelling over the right fibula and hyperesthesia of the lateral peroneal nerve in the right leg. There were no other abnormalities found in the left lower and upper limbs.

Laboratory findings including CBC, serum calcium, phosphorus, alkaline phosphatase, erythrocyte sedimentation rate, C-reactive protein, alpha fetoprotein, and carcinoembryonic antigen were all within normal ranges. The patient was referred to the radiology department for further evaluation. Initially, anteroposterior, and lateral radiographs of the right leg were obtained. Results showed proximal two-thirds cortical thickening and hyperostosis of the fibula consistent with the characteristic "dripping candle wax" appearance of melorheostosis. (Figure 1). Magnetic Resonance Imaging (MRI) and a biopsy were also ordered to confirm the diagnosis, exclude associated complication, and rule out malignancy and other pathologies. MRI demonstrated a lobulated periosteal thickening involving the posterolateral cortex of the right fibula, extending along the fibular shaft from the proximal metaphysis to a few centimeters proximal to the ankle joint. With encroachment on the medullary cavity, there were no masses of soft tissue or lymphadenopathy. There were no lytic bone lesions found. The peroneal nerve was likely being irritated by periosteal thickening (Figure 2). The patient initially was given trials of conservative management, but when the pain became intolerable and interfered with his daily activity, the patient requested an immediate relief of his symptoms. Surgical debulking of the protruding sclerotic bone from the fibula was performed to achieve a smooth margin. (Figure 3), After the surgical debulking the resected fibula was then sent for pathologic evaluation. The bone underwent decalcification process to facilitate sectioning and histologic evaluation. Microscopic examination revealed benign bone characterized by replacement of the normal cancellous bone by dense cortical bone with thickened trabeculae along with normal presence of the Haversian canals within the dense bone. The normal marrow in the inter trabecular spaces were replaced by fat. No nidus of osteoid tissue or cartilaginous component were seen. No cellular atypia or mitotic figures were identified (Figure 4). Finally, the patient was very satisfied with the outcomes as he experienced pain relief and full range of motion of his leg after 6 months follow up.

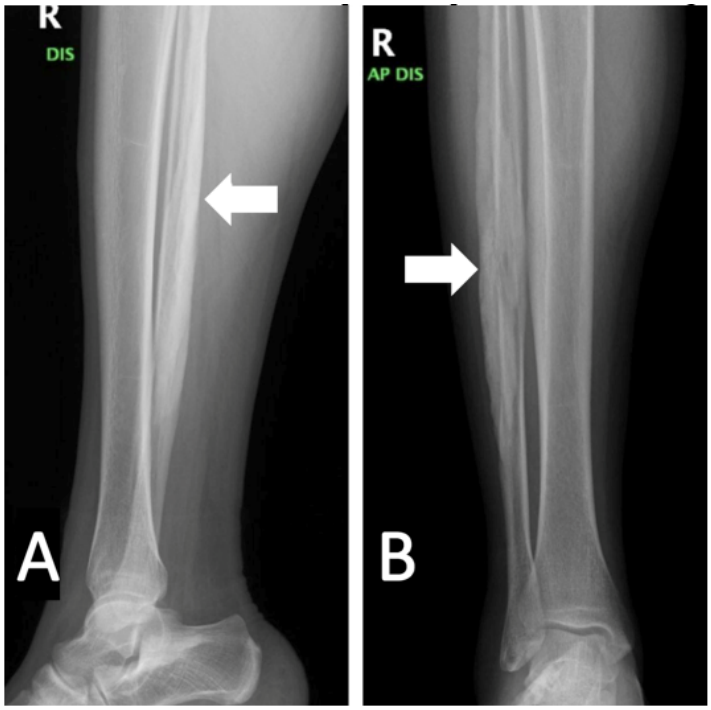

Figure 1: A,B: Lateral and anteroposterior radiographs of the right leg showing proximal two-thirds cortical thickening and hyperostosis of the fibula consistent with the characteristic **"dripping candle wax"** sign.

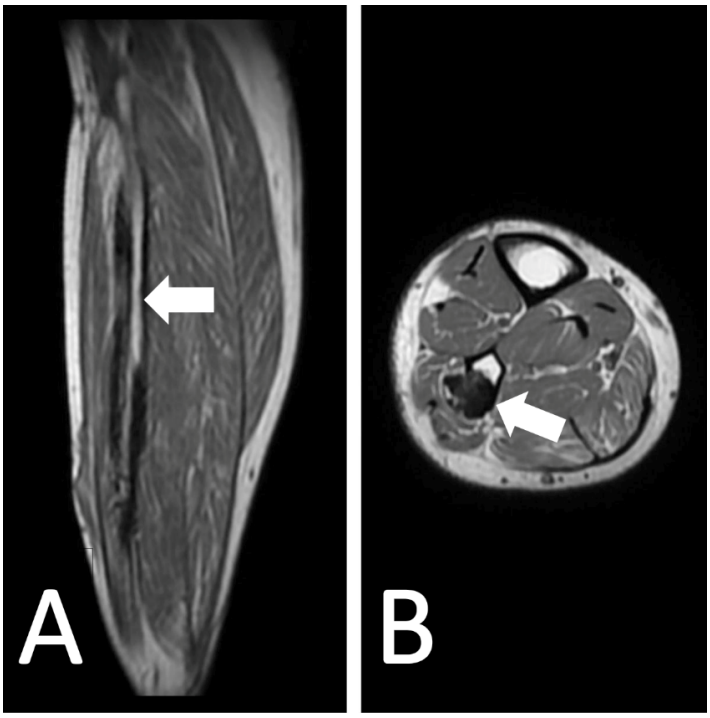

Figure 2: A: Sagittal T1- weighted, **B:** axial T1- weighted MRI showed lobulated periosteal thickening involving the posterolateral cortex of the right fibula, extending along the fibular shaft from the proximal metaphysis to a few centimeters proximal to the ankle joint, with encroachment on the medullary cavity, there are no masses of soft tissue or lymphadenopathy. There are no lytic bone lesions found.

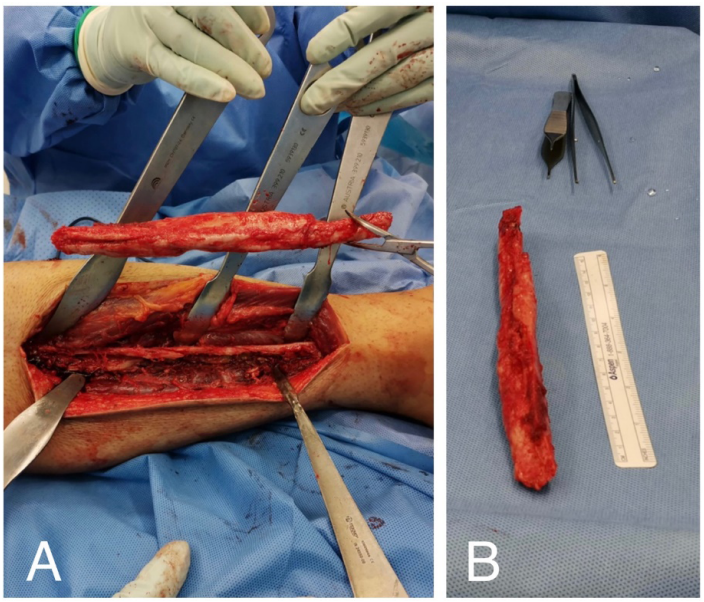

Figure 3: A: Intraoperative picture shows anterolateral approach of the leg aimed for pathology resection in the proximal fibula with 25 cm longitudinal incision. **B:** tumor was resected measuring 20 cm with enough margin and was 2 times more than the width of the healthy fibula which was entirely preserved.

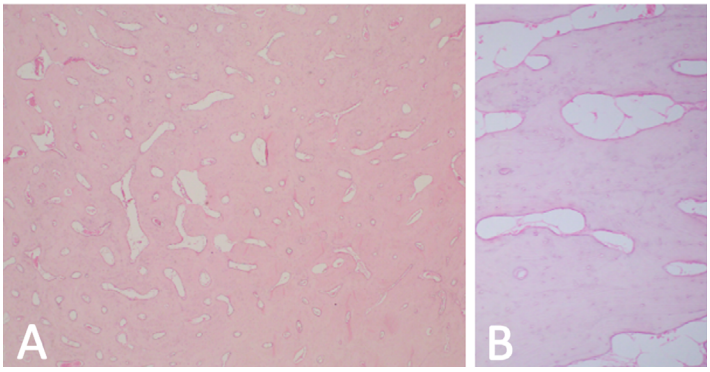

Figure 4: A,B: Low power views of histologic section of the resected specimen. The normal cancellous bone is replaced by dense cortical bone with thickened trabeculae and with presence of the Haversian canals. The normal marrow in the inter trabecular spaces is replaced by fat. No cellular atypia or mitotic figures seen.

Discussion

Melorheostosis is considered as a very rare benign sclerosing bone dysplasia which was described for the first time in 1922 by Leri and Joanny, which is also known as Leri-Joanny disease. [2,6,7] Although the theory of LEMD3 mutation was adopted initially as a key for the pathology emergence, recent genetic research proposed the MAP2K1 oncogene mutation, which contributes to over proliferation of osteoblast and subsequently leading to Melorheostosis disease. [8] As in our case, the pathology usually manifests with severe pain, dysfunction, and deformity in a single limb, as well as distributes in a pseudosclerotomal manner. [2,9] Pain was the main reason for our patient's intention to seek medical advice, this is similar to the result of the largest retrospective clinical analysis by Smith,GC. [9] To our knowledge, physiotherapy can decrease the symptoms as referred by Flowers KR. [10] However, our patients didn't get any improvement after full course of low load stress physiology, hence, surgical intervention was negotiated. The condition usually affects the cortex of bone, extending through the peristome layer and resembling dripping candle wax. [5] In our patient, dripping wax sign was appreciated in the radiograph image which is pathognomonic for melorheostosis. Other signs as such as myositis ossificans, osteopathic striatae and osteoma were reported in the literature.[11] However, these finding are less frequent and were not seen in our case.

Bisphosphonate therapy showed good results in controlling such condition, as it enhances osteoblasts and limit osteoclast activity. [12] However, bisphosphonate was not involved in our management plan due to weak level of evidence. Patient preference for surgical intervention is always attributed to the intention of immediate relief of the agonizing symptoms. Wordsworth P, et al.

suggested surgical resection among those patients, especially in case of impingement or nerve compression. [13] However, our patient was mainly suffering from severe pain, which limits his functional daily activity. The course of the condition is usually benign, surprisingly, Kotwal A et al, reported 2 cases with agressive transformation to malignant fibrous histiocytoma and osteosarcoma. [14] However, 2 year follow up after diagnosis of our patient was free from aggressive features which could exclude malignancy transformation. Last follow up of our patient was on 13 November 2022, showed no recurrence of symptoms or new radiological findings, however, the condition has a possibility of recurrence. [15] Interestingly, Gagliardi GG. Et al, stated that melorheostosis is associated with functional morbidity rather than mortality. [16] Fortunately, our patient regained his normal functional status after the surgical resection of the pathology.

Conclusion

Melorheostosis is a rare benign dysplastic condition mainly affects the cortex of long bones. Radiological evaluation can establish the diagnosis of melorheostosis; however, biopsy is essential to exclude malignancy. Methods of treatment varies depending on patient age, site of pathology, symptoms, and complication. In our case we chose surgical debulking due to long period of severe pain which led to limited functional status. six months follow up showed satisfactory outcome of the patient overall condition.

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