International Journal of Research in Orthopaedics
Agarwal S et al. Int J Res Orthop. 2017 May;3(3):635-638
http://www.ijoro.org

Case Report

DOI: http://dx.doi.org/10.18203/issn.2455-4510.IntJResOrthop20171915

Localised melorheostosis

Sunny Agarwal[1], Vishesh Khanna[2], Mathew Varghese[1], Suresh B.[3]*

[1]Department of Orthopaedics, 1[St] Stephen's Hospital, Tis Hazari, New Delhi, India
[2]Fellow in Arthoplasty, Sunshine Hospital, Secunderbad, Telangana, India
[3]Subbaiah Institute of Medical Sciences, Purle, Shivamogga, Karnataka, India

Received: 15 January 2017
Revised: 09 March 2017
Accepted: 16 March 2017

***Correspondence:**
Dr. Suresh B.,
E-mail: sureshb008@gmail.com

Copyright: © the author(s), publisher and licensee Medip Academy. This is an open-access article distributed under the terms of the Creative Commons Attribution Non-Commercial License, which permits unrestricted non-commercial use, distribution, and reproduction in any medium, provided the original work is properly cited.

ABSTRACT

Melorheostosis is a sclerosing bone dysplasia of unknown aetiology. Diagnosis is mainly based on a combination of clinical and imaging studies. The classical dripping candle wax picture on plain radiograph is diagnostic. We report a case of localized melorheostosis of a 14 year old boy on daily analgesics for two years for pain relief. Radiographs showed endosteal bone formation like dripping candle wax. Biopsy and decompression of the hyperostosis was done. However biopsy did not relieve his symptoms. Based on literature survey he was given a single infusion of zoledronic acid. This gave dramatic relief in pain.

Keywords: Localised melorheostosis, Flowing/dripping candle wax, Bisphosphonates, Zoledronic acid

INTRODUCTION

Melorheostosis is a rare sclerosing bone dysplasia characterized by its classic radiographic feature of flowing/dripping candle wax. It usually affects one limb, more often the lower extremity, and rarely the axial skeleton. The etiology remains unknown and various theories have been proposed to explain the pathogenesis of this disease. It often has an insidious onset in early adult life. Symptoms include pain, oedema and limitation of joint movement. Pain is often more common in adults because of subperiostial bone formation. Treatment in most instances has been symptomatic. Conservative therapies include oral medications such as bis-phosphonates, NSAIDs, and nifedipine. Surgical procedures consist of soft-tissue procedures such as excision of fibrous and osseous tissue, fasciotomy, capsulotomy, osteotomies and excision of hyperostoses, arthrodesis, and amputation.[1-7]

CASE REPORT

A 14 year old male child presented with a history of pain and swelling in the left leg for last 2 years. Pain was insidious in onset, dull aching and gradually progressive in nature. Initially he could manage with analgesics being taken once in 2-3 days but for last 1 year he was taking it twice or thrice in a day. Pain used to aggravate on exertion and relieve mildly on taking rest.

On examination there was a bony prominence with tenderness over the proximal 1/3[rd] of leg without redness and local rise of temperature (Figure 1). Range of motion of knee and ankle joints were within normal limits.

Plain radiographs showed flowing/dripping wax pattern involvement of the proximal half of the left tibia (Figure 2). It was involving the endosteal surface of the anterior cortex with clear demarcation between the affected and normal bone. CT scan showing flowing bone pattern on the endosteal surface of anterior cortex (Figure 3).

Laboratory results of serum calcium, phosphorus, ESR, serum alkaline phosphatase were within normal limits. Skeletal survey did not reveal involvement of any other site.

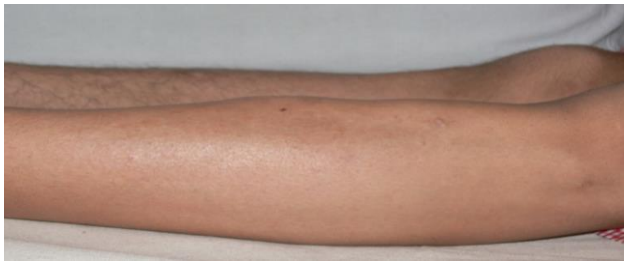

Figure 1: Bony prominence over the proximal 1/3rd of leg.

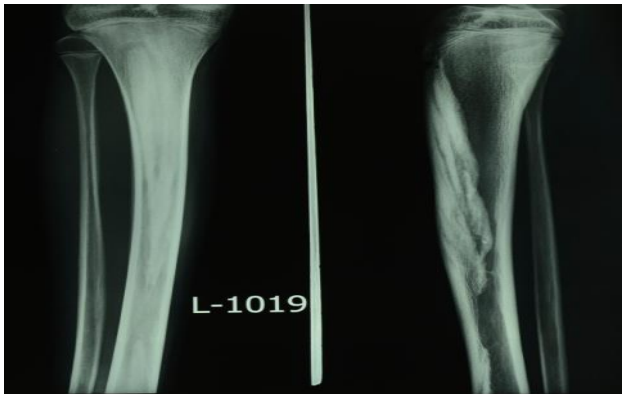

Figure 2: Flowing/dripping wax pattern involvement of the proximal half of the left tibia.

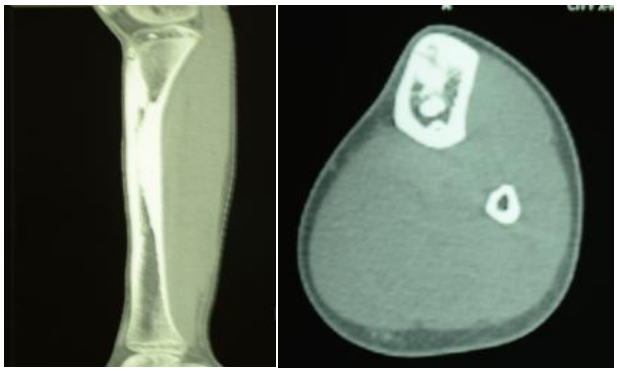

Figure 3: Flowing bone pattern on the endosteal surface of anterior cortex.

Decompression of the hyperostosis was done through a bone window (Figure 4) and biopsy of the excised mass revealed cortical bone with crushed marrow.

Patient was discharged with a slab and was kept non weight bearing to prevent a pathological fracture. But patient had no relief in pain and he had to continue analgesics twice or thrice in a day. Patient was then given a single infusion of 3 mg zoledronic acid [bisphosphonates] given over 30 minutes which resulted in a dramatic improvement in pain. Patient has been walking pain free ever since and has not required any further treatment.

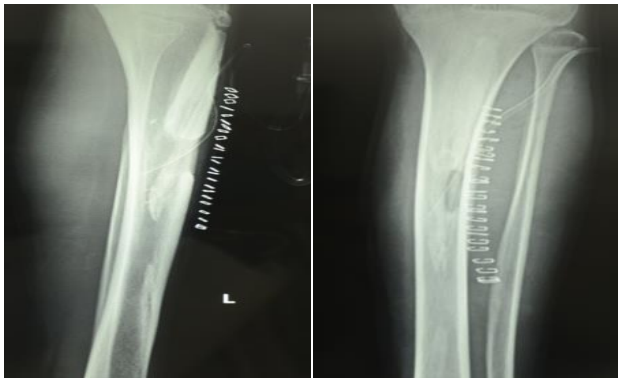

Figure 4: Decompression of the hyperostosis was done through a bone window.

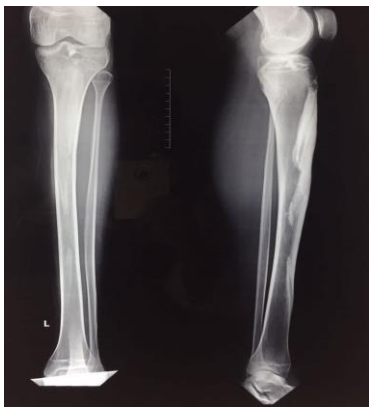

Figure 5: At the latest follow up period of 1 ½ years.

DISCUSSION

Melorheostosis is a rare, non-hereditary, benign, sclerosing mesodermal dysplasia which affects the skeleton and adjacent soft tissues.[1-3] It got this name because of the characteristic periosteal hyperostosis along the cortex of long bones which looks similar to the flowing or dripping of candle wax [originated from Greek, melos = limb, rhein = to flow, ostos = bone].[4-6] Melorheostosis is also known by other synonyms such as candle disease of the bone, and osteosis eburnisans monomelica.[8,9]

Melorheostosis was first described by Leri and Joanny in 1922, and was also called Leri's disease or syndrome thereafter. This condition may affect only one bone [monostotic form, representing a forme fruste of the disorder], one limb [monomelic form], or multiple bones [polyostotic form]. It was monostotic in our patient. Skin and subcutaneous tissue involvement can result in fibrosis and joint contractures leading to deformity and limb-length discrepancy.[3,10,11]

The etiology of melorheostosis remains unknown. However, One possible etiology of melorheostosis is 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-β signalling.[12] There have been various theories proposed to explain the pathogenesis of this disease such as a developmental disorder theory, ischemic theory, telangiectatic theory, and infective theory.[13] Currently, there are two major hypotheses in existence. In 1979, Murray and McCredie correlated melorheostosis with sclerotomes, hypothesizing that melorheostosis might be the result of a segmental sensory lesion due to a specific infection, insult, or injury to segments of the neural crest during embryogenesis, which partially explains the peculiar monomelic involvement of melorheostosis.[14] In 1995, Fryns proposed mosaicism to explain the sporadic occurrence of dysplasia which suggests that the asymmetric involvement of skeletal structures and concomitant vascular and hamartomatous changes in the overlying soft tissues result from an early post-zygotic mutation of the mesenchyme which explains why the extent of involvement is so variable and why the incidence ratio in both genders is equal.[15]

Histopathology reveals nonspecific periosteal bone formation with thickened trabeculae and fibrotic changes in the marrow spaces.[1-3] Our results were comparable.

Symptoms include pain, limb stiffness, limitation of motion in the joints, and deformity of the involved extremity, usually do not manifest until late childhood or early adolescence and tend to progress into adult life.[1-4] The disease usually exhibits a chronic course with periods of exacerbation and arrest. Our patient had pain and swelling for 2 years with periods of exacerbation and rest initially. Potential causes of bone pain in melorheostosis include increased osteoclastic bone resorption and activation of pain receptors, raised intraosseous pressure and increased vascularity secondary to hyperosteosis and soft tissue involvement around joints.

Laboratory findings for serum calcium, phosphorus, erythrocyte sedimentation rate, and alkaline phosphatase levels have been reported to be within normal limits.[1-3,6] Our results were comparable.

Flowing cortical hyperostosis along one side of the shaft of the long bone resembling "melting wax flowing down the side of a candle" is the characteristic radiographic appearance of melorheostosis.[2-4,16] The lesions are typically eccentrically placed with no evidence of bony destruction. There is usually a distinct demarcation between the affected and normal bone. Our patient had radiographic finding that were typical of melorheostosis. 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.[3] Isotope bone scanning reveals increased uptake in the same distribution seen on plain radiography reflecting an increase in bone metabolism. Computed tomography [CT] scans and three-dimensional reconstruction show candle wax-like or massive, rough hyperostosis around cortical and cancellous bones, accompanied by deformity of the bones, narrowing of the medullary cavity.

The differential diagnosis for melorheostosis includes osteopoikilosis, osteopathic striata, myositis ossificans, parosteal osteosarcoma, and osteoma.[16-18]

Various conservative or surgical methods have been practiced in treating the pain and deformities associated with melorheostosis. Conservative therapies include oral medications such as bisphosphonates, NSAIDs, and nifedipine. Surgical procedures consist of soft-tissue procedures such as tendon lengthening, excision of fibrous and osseous tissue, fasciotomy, capsulotomy, osteotomies and excision of hyperostoses, arthrodesis, and amputation.[1-5,7,8,14,19] When treating limb deformities associated with surgical treatments in melorheostosis patients, recurrences are common. We treated our patient initially with decompression of the hyperostosis which did not improve his symptoms. Then we treated him with single infusion of zoledronic acid which took him off from analgesics.

Bisphosphonates are used for symptomatic control in both malignant and non-malignant diseases associated with increased bone turnover. They have been shown to decrease bone pain, slow progression of bone lesions and decrease the risk of pathological fracture.[20,21] Nitrogen containing bisphosphonates inhibit osteoclast-mediated bone resportion by direct and indirect actions on osteoblasts and macrophages. Bisphosphonates also decrease bone vascularity and zoledronic acid has been shown *in vitro* to be a potent inhibitor of angiogenesis.[22] Thus, bisphosphonate treatment via a number of mechanisms would be expected to reduce inflammatory bone pain and symptoms in melorheostosis. Donath *et al* described a case of melorheostosis with extensive bilateral disease and elevated alkaline phosphatase where the patient was treated with 30 mg of pamidronate daily for 6 days which resulted in a rapid improvement in symptoms.[23]

It is recognised that osteonecrosis of jaw is common, although still very rare, with bisphosphonate treatment. Therefore it is important to ensure good dental hygiene and adequate 25-hydroxy vitamin D levels prior to administering intravenous bisphosphonates due to the increased risk of osteonecrosis of the jaw.[24]

CONCLUSION

This patient had localized melorheostosis which was treated with simple analgesics in several hospitals without relief. When we saw him, we also could not explain the severity of pain and therefore kept the possibility of sclerosing osteomyelitis. However, the

decision to take a biopsy was based on his clinical symptom of severe pain. At surgery, unlike sclerosing osteomyelitis, the bone was soft and could be peeled away in layers. We have not seen this reported in the literature. His temporary relief after surgery with recurrence soon after is an indicator that mere decompression or biopsy will not relieve the pain in patients of melorheostosis. While it is reported in literature, we did not expect the dramatic pain relief that the patient had from zoledronic acid. We present this case to highlight the importance of suspecting such lesions as a localized melorheostosis and to report on the dramatic pain relief from zoledronic acid. It is not clear from literature the duration of treatment with zoledronic acid.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

REFERENCES

1. Gagliardi GG, Mahan KT. Melorheostosis: a literature review and case report with surgical considerations. J Foot Ankle Surg. 2010;49(1):80-5.
2. Jain VK, Arya RK, Bharadwaj M, Kumar S. Melorheostosis: clinicopathological features, diagnosis, and management. Orthopedics. 2009;32(7):512.
3. Greenspan A, Azouz EM. Bone dysplasia series. Melorheostosis: review and update. Can Assoc Radiol J. 1999;50(5):324-30.
4. Campbell CJ, Papademetriou T, Bonfiglio M. Melorheostosis. A report of the clinical, roentgenographic, and pathological findings in fourteen cases. J Bone Joint Surg Am. 1968;50(7):1281–304.
5. Taylor PM. Melorheostosis. J Foot Surg. 1976;15(2):55–8.
6. Freyschmidt J. Melorheostosis: a review of 23 cases. Eur Radiol. 2001;11(3):474–9.
7. Gong HS, Lee KH, Oh JH, Chung J-H, Baek GH, Chung MS. Successful elbow contracture release secondary to melorheostosis. A case report. J Bone Joint Surg Am. 2008;90(5):1106–11.
8. Ihde LL, Forrester DM, Gottsegen CJ, Masih S, Patel DB, Vachon LA, et al. Sclerosing bone dysplasias: review and differentiation from other causes of osteosclerosis. Radiogr Rev Publ Radiol Soc N Am Inc. 2011;31(7):1865–82.
9. Rozencwaig R, Wilson MR, McFarland GB. Melorheostosis. Am J Orthop Belle Mead NJ. 1997;26(2):83–9.
10. Siegel A, Williams H. Linear scleroderma and melorheostosis. Br J Radiol. 1992;65(771):266–8.
11. Judkiewicz AM, Murphey MD, Resnik CS, Newberg AH, Temple HT, Smith WS. Advanced imaging of melorheostosis with emphasis on MRI. Skeletal Radiol. 2001;30(8):447–53.
12. Hellemans J, Preobrazhenska O, Willaert A, Debeer P, Verdonk PCM, Costa T, et al. Loss-of-function mutations in LEMD3 result in osteopoikilosis, Buschke-Ollendorff syndrome and melorheostosis. Nat Genet. 2004;36(11):1213–8.
13. Lasserre C. A case of melorheostosis. Rev Rhum Mal Osteoartic. 1960;27:347–50.
14. Murray RO, McCredie J. Melorheostosis and the sclerotomes: a radiological correlation. Skeletal Radiol. 1979;4(2):57–71.
15. Fryns JP. Melorheostosis and somatic mosaicism. Am J Med Genet. 1995;58(2):199.
16. Spieth ME, Greenspan A, Forrester DM, Ansari AN, Kimura RL, Siegel ME. Radionuclide imaging in forme fruste of melorheostosis. Clin Nucl Med. 1994;19(6):512–5.
17. Kawabata H, Tsuyuguchi Y, Kawai H, Yasui N. Melorheostosis of the upper limb: a report of two cases. J Hand Surg. 1984;9(6):871–6.
18. Zeiller SC, Vaccaro AR, Wimberley DW, Albert TJ, Harrop JS, Hilibrand AS. Severe myelopathy resulting from melorheostosis of the cervicothoracic spine. A case report. J Bone Joint Surg Am. 2005;87(12):2759–62.
19. Zhang C, Dai W, Yang Y, Tang Q, Yao Z. Melorheostosis and a review of the literature in China. Intractable Rare Dis Res. 2013;2(2):51–4.
20. Saad F, Lipton A. Clinical benefits and considerations of bisphosphonate treatment in metastatic bone disease. Semin Oncol. 2007;34(4):17-23.
21. Theriault RL. Zoledronic acid (Zometa) use in bone disease. Expert Rev Anticancer Ther. 2003;3(2):157–66.
22. Wood J, Bonjean K, Ruetz S, Bellahcène A, Devy L, Foidart JM, et al. Novel antiangiogenic effects of the bisphosphonate compound zoledronic acid. J Pharmacol Exp Ther. 2002;302(3):1055–61.
23. Donáth J, Poór G, Kiss C, Fornet B, Genant H. A typical form of active melorheostosis and its treatment with bisphosphonate. Skeletal Radiol. 2002;31(12):709–13.
24. Khosla S, Burr D, Cauley J, Dempster DW, Ebeling PR, Felsenberg D, et al. Bisphosphonate-associated osteonecrosis of the jaw: report of a task force of the American Society for Bone and Mineral Research. J Bone Miner Res. 2007;22(10):1479–91.

Cite this article as: Agarwal S, Khanna V, Varghese M, Suresh B. Localised melorheostosis. Int J Res Orthop 2017;3:635-8.