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The dripping candle wax sign of melorheostosis

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Abstract

Melorheostosis is a rare benign bone disease including dysostosis and sclerosis. Dripping candle wax presence is a common and typical sign of melorheostosis. This sign appears as irregular hyperostosis of the cortical bone which is likened to melted wax flowing down one side of a candle. It can sometimes cause pain, stiffness joint, or limitation of motion in the affected areas implicitly but mostly has no symptoms. It is usually observed on plain radiography; its appearance is generally hyperplasia on one side of the bone. We report a 33-year-old male who has an incidental diagnosis of melorheostosis post-trauma.

Keywords

Dripping candle wax, hyperostosis, melorheostosis, Leri disease, sclerosing bone dysplasia

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Introduction

Melorheostosis or Leri disease is an exceptionally rare benign sclerosing bone dysplasia, typically affects both cortical bone and its surrounding soft tissue structures in a sclerotomy distribution. The origin of its name comes from the Greek ("melos" = "limb", "rhein" = "to flow", "ostos" = "bone"). The density distribution of melorheostosis likewise the segmental distribution can correspond with the anatomical distribution of nerve roots or blood vessels. Any bone in the skeletal system can be affected by this disease, but the long bones of the lower extremities are the most common position.[1,2] Leri and Joanny[2] have described this disease since 1922 for the first time and it is not currently known clearly. Clinical manifestations are variable and the etiological diagnosis is not absolutely clear. Melorheostosis is a developmental abnormality that belongs to one of the groups of sclerosing dysplasias of bone.[3] All ages can be affected (popular age range: 2–64 years). Its demonstration is often discreet until adolescence and occurs equally in both sexes.[4,5] We report a case of an adult man who presented to an emergency with the cause of chest wall pain post-trauma. His disease is detected by plain radiographs and computed tomography (CT). The aim of this article provides several points and additions to literature about the clinical manifestations, imaging features, treatment, and prognosis of melorheostosis.

Case report

A 33-year-old male presented with trauma and left thoracic ache. Legs and hands of him were slightly rubbed due to an injury. There were no signs of danger, and no muscle weakness or respiratory problems. There was no swelling, no deformity, and no color change in the joints of the limbs. He had no history of joint disease, and there was no related family history. Laboratory evaluation included complete blood count, erythrocyte sedimentation rate, phosphate test, vitamin D, calcium levels, phosphorus, and C-reactive protein, which were all normal. Rheumatoid factor, anti-cyclic

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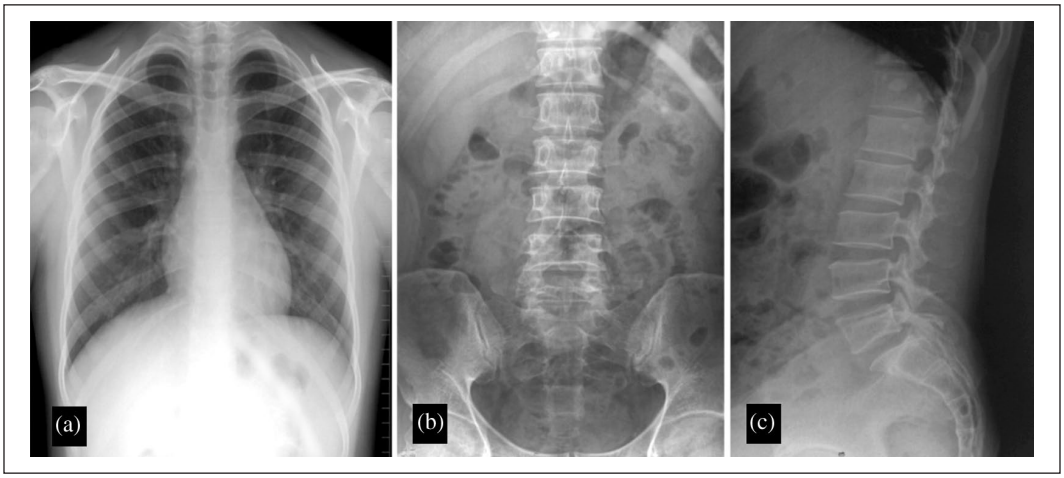

Figure 1. Radiographs of the chest (a), anteroposterior (b) and lateral (c) views of a lumbar spine showing hyperostosis involving 11th rib on the left side, and involving thoracic and lumbar vertebrae. This is a characteristic sign of melorheostosis and the appearance of cortical is irregular hyperostosis. It typically occurs on one side of the involved bone and has been likened to melted wax flowing down one side of a candle.

citrullinated peptide (CCP), and antinuclear antibodies were also tested to rule out joint diseases, and all were negative.

The patient underwent radiographs of the chest, the lumbar spine, and the limbs which showed hyperostosis involving ribs bone, the bones of the limbs, both anterior and posterior thoracic-lumbar spine segments. Radiographs of the limbs showed cortical hyperostosis in multiple bones including the femur, tibia, fibula, hands, and feet bones. They revealed the multiple areas of dense linear hyperostosis and resembled melted wax dripping down one side of burning candles. Based on these characteristic radiographic features, this patient is diagnosed with melorheostosis (Figures 1 and 2).

The patient then had a chest CT scan to assess more the chest wall, lungs, and pleura. CT images also showed signs of melorheostosis with hypertrophy, dysplasia, and sclerosis (Figure 3). It demonstrated sclerosis of the spine and hyperostosis of several ribs bone. The patient was asked to have magnetic resonance imaging (MRI) for advanced imaging and he agreed to do it. MRI showed low signal intensity areas in the corresponding bone. Some areas of the ribs were slightly enlarged and the signal was reduced on all pulse sequences. The vertebrae were similarly affected by CT (Figure 4).

The patient was prescribed painkillers and wound care for 5 days. He was re-examined after a month; the pain from his trauma had disappeared completely. We followed up his melorheostosis for a year, patient status then was normal.

Discussion

Melorheostosis is characterized by bone hypertrophy, dysplasia, and sclerosis. The dripping candle wax sign of melorheostosis stems from the sclerosis of one side of the cortex and appears along the long axis of the bone.[4] The most common position effected by melorheostosis is the long bones of the limbs and the auxiliary skeleton. It can sometimes be found in the bones of the hands and feet but rarely found in the axial skeleton.[6,7] It may affect single bone or multiple; however, bilateral involvement cases being reported are rare situations.[8,9]

The origination of melorheostosis has not been clarified yet, and many theories have been proposed.[10] Murray and McCredie[11] hypothesize that one of the causes of melorheostosis with sclerotomes is a segmental sensory lesion of neural crest happening during embryogenesis. Fryns[12] has suggested mosaicism to explain the sporadic occurrence of dysplasia. Lately, the phenomenon of sclerosing bone dysplasias is usually associated with genetic work in several families with melorheostosis, osteopoikilosis, and Buschke-Ollendorff syndrome.[13] Several research groups indicate that this disease is related to gene mutations. It has been proved by Hellemans et al.[14] that the melorheostosis is the result of a loss-of-function mutation in LEMD3 on chromosome 12q. Other study groups have demonstrated several genes codes for a nuclear protein that normally inhibits both transforming growth factor and bone morphogenic protein leading to melorheostosis.[15]

Recent studies of melorheostosis lesional tissue prove that most of the cases arise from somatic MAP2K1 mutations, although a small number of them may arise from other genes in related pathways, such as KRAS. Cases related to the MAP2K1 mutation can cause the appearance of "candle wax dripping sign" on radiographs.[16] In the study of Kang et al.,[17] they report that mutation of MAP2K1 can inhibit differentiation and mineralization of BMP2-mediated osteoblast. Mosaicism pattern is also found in the skin adjacent bone lesions in four of five patients. This shows that the MAP2K1 oncogene is significant in bone formation of human and opens a perspective of treatment for melorheostosis by gene therapy in the future. Genetic research has not been elucidated in our case, but it may also be related to

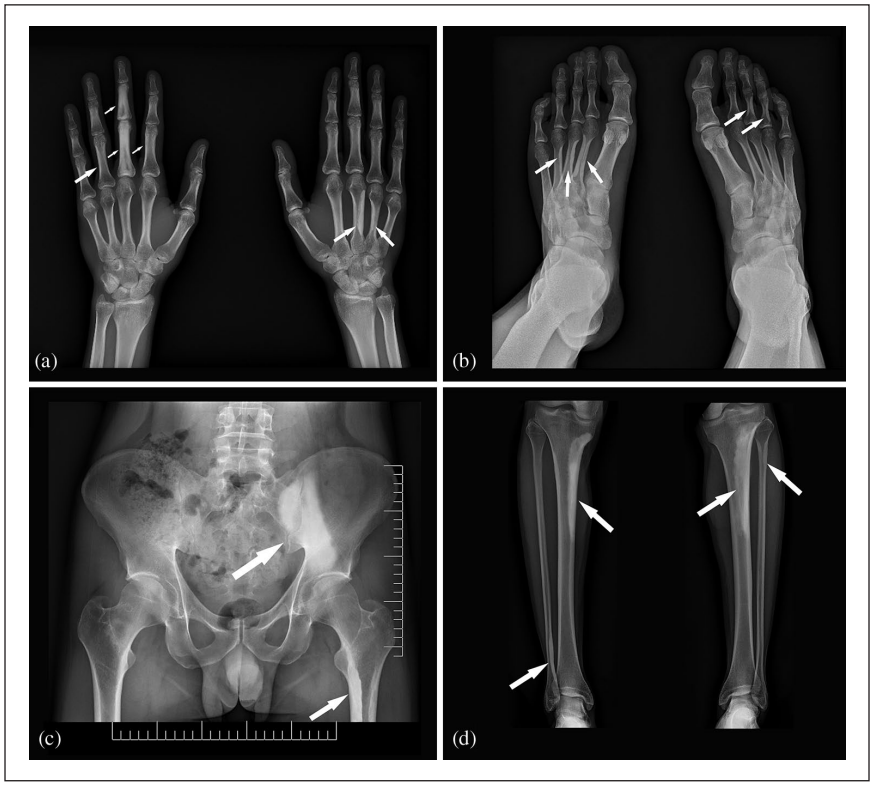

Figure 2. Radiographs of the metacarpal, metatarsal, pelvis, and legs. (a) Radiographs of two hands demonstrating a hyperdense lesion in several bones with obliteration of the medulla in the middle finger of the left hand. (b) Radiographs of two feet with localized cortical hyperostosis of several metatarsal bones. (c) Radiographs of the pelvis showing cortical hyperostosis extending from left iliac bone to left femur. (d) Radiographs of two legs showing regional sclerosis of cortically based endosteal in the tibia and fibula with characteristic melted candle wax appearance (*arrows*).

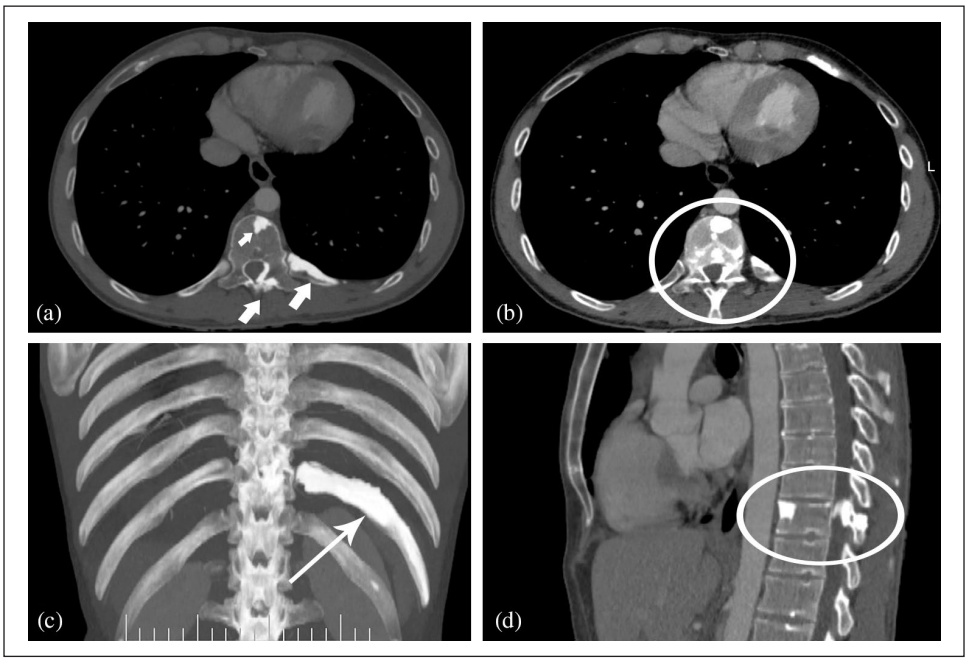

Figure 3. (a and b) Axial CT scan images showing hyperostosis involving 11th rib left, thoracic, and lumbar vertebrae. (c) CT reconstruction image of the coronal plane showing the left 11th entire rib with hypertrophy and sclerotic. (d) Sagittal reformatted CT image showing hyperostosis involving the anterior and posterior segment of thoracic vertebrae. Note the spinal canal is not narrow (*arrows and circles*).

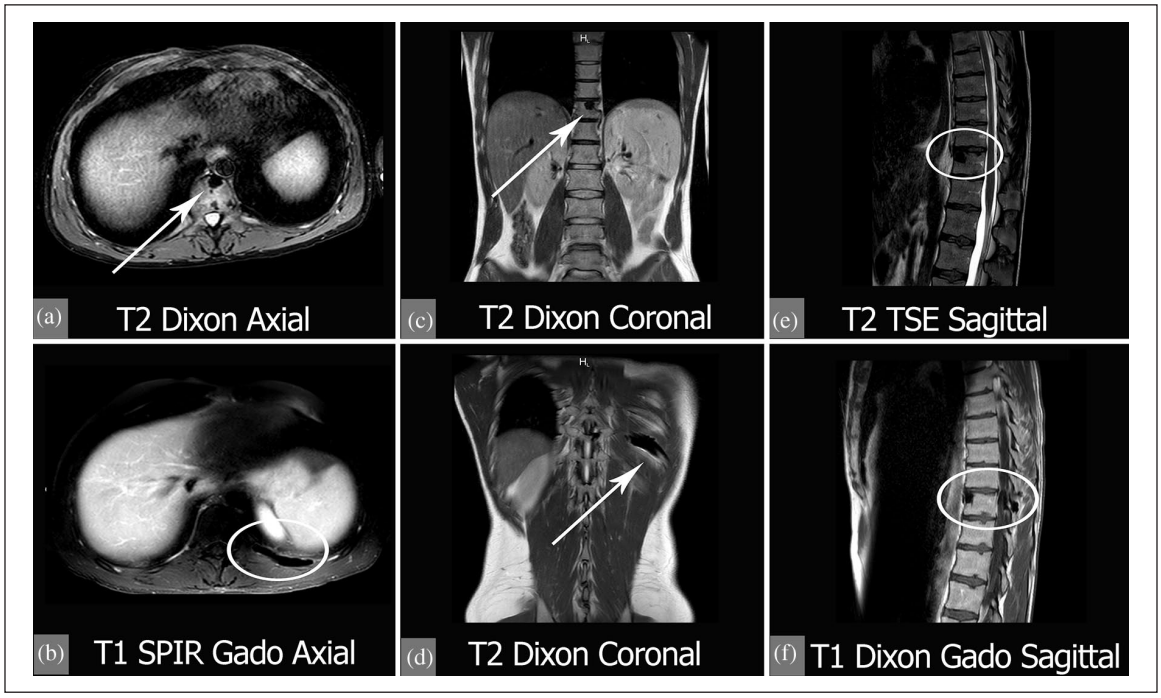

Figure 4. MRI of melorheostosis on T2-Weighted sequences (a, c, d, and e) and T1-Gado sequences (b and f). Images showing some areas of the ribs are enlarged and hypointensity of a signal on all pulse sequences. Hyperostosis of the vertebrae as a hypointense signal is also observed (*arrows and circles*).

the MAP2K1 mutation. In this case, because the appearance of radiographs is similar to the studies of MAP2K1, however, clinical manifestations are not suitable yet.

Melorheostosis is often clinically asymptomatic. But sometimes the most common presentation is bony swelling, pain, localized growth abnormalities, stiffness, or joint deformities. It may lead to amputation if a serious defect is present. When the surrounding soft tissues are also involved, there could occur a scleroderma pattern of the skin that involves edema, hypertrichosis, subcutaneous fibrosis, fibromas, or fibrolipomas.[18] Associations with vascular anomalies have also been reported.[5,19] Laboratory parameters for phosphorus, serum calcium, and alkaline phosphatase levels are normal.[20] Various nerve compression syndromes have also been described, including carpal tunnel syndrome and spinal nerve root compressions.[16] Smith et al.[21] have reported in clinical review with 24 patients, pain and limb deformity are the commonest presenting symptoms, followed by limitation of movement, weakness, and numbness. Histopathologic examination of pathological bone specimens demonstrates unclear hyperostotic periosteal bone formation pattern, with fibrotic transforms and thickening of bone rafts in the bone marrow spaces.[22]

Melorheostosis inclines to be involved in segmental and unilateral in an arrangement. They can affect one or more sclerotomes positions which are a region of a bone innervated from a spinal sensory nerve individually.[11,18] The distribution of melorheostosis on radiographs in our patients shows non-compliance with sclerotome pattern; however, this is not yet clear. According to Jha et al.,[23] only 17% of the 30 patients in their research match with sclerotome. Melorheostosis does not always have typical imaged characteristics that even may overlap and vary radiology manifests. Its characteristics appear in many patterns such as myositis ossificans-like, osteoma-like, osteopathic striata-like, or a mixed pattern which make diagnosis very difficult and complicated.[4] In a study case series involving 23 patients, only five patients have classic radiographic features.[19] CT and MRI are usually not needed for diagnosis except for complex cases. If CT is performed, undulating cortical hyperplasia areas will appear as areas of high density on CT which are similar to radiographs.[24] There are not many articles on MRI pulse sequences in the affected bone area. Encroachment into the marrow may be observed well on MRI, relating to the endosteal.[24,25] Bone scintigraphy usually presents in this disease, showing moderately increased uptake of radiopharmaceutical tracer, principally concentrated on the cortex.[26] Our patient has not assigned to perform nuclear scintigraphy.

Although a typical appearance on the radiology of melorheostosis is characteristic of diagnosis, it may be readily distinguished from other lesions. Image findings may have a range of different manifestations and need to be distinguished from osteosarcoma, osteoid osteoma, parosteal osteosarcoma, osteochondroma, osteopetrosis, osteomyelitis, osteopoikilosis, pycnodysostosis, osteopathic, fibrous dysplasia, and myositis ossificans.[16,27]

A variety of surgical and conservative treatments have been used for melorheostosis to try to relieve the pain and related defects. The conservative treatments which are used include drugs such as Adalat, bisphosphonates, and non-steroidal anti-inflammatory drugs.[28] In our case, the patient did not show symptoms related to melorheostosis; so no prescription. Other conservative treatments with no surgery requirement include manipulations, physical therapy, brace, serial casting, sympathectomies, and nerve block.[29] In many cases, melorheostosis needs surgical treatment such as soft tissue procedures including tendon lengthening, capsulotomy, fasciotomy, fibrous, and osseous tissue excision. Other therapies include hyperostotic bone resection, bone orthopedic, and even amputation consideration.[30] Conservative treatments are often ineffective in treating severe limb deformities associated with melorheostosis, and surgical treatment recurrences are common.[25–30]

Conclusion

Melorheostosis is a rare benign disease. The appearance of it on radiographic is likened to the appearance of melting wax flowing down of a candle. Dripping candle wax appearance is a classic characteristic sign in radiographs and CT. If the diagnose confirmed by the radiology, the management should avoid unnecessary investigation or biopsy. This disease can be treated with conservation methods or surgery.

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Ethical approval

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Informed consent

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References

1. Franca P, Ferrreira C, Figueiredo R, et al. Melorheostosis. *Radiol Bras* 2015; 48(1): 60–61.
2. Leri A and Joanny J. Une affection non décrite des os hyperostose “en coulée” sur toute la longeur d’un member ou “melorhéostose.” *Bull Mem Soc Med Hop Paris* 1922; 46: 1141–1145.
3. Boulet C, Madani H, Lenchik L, et al. Sclerosing bone dysplasias: genetic, clinical and radiology update of hereditary and non-hereditary disorders. *Br J Radiol* 2016; 89(1062): 20150349.
4. Kotwal A and Clarke B. Melorheostosis: a rare sclerosing bone dysplasia. *Curr Osteoporos Rep* 2017; 15(4): 335–342.
5. Kumar R, Sankhala S and Bijarnia I. Melorheostosis: case report of rare disease. *J Orthop Case Rep* 2014; 4(2): 25–27.
6. Fick C, Fratzl-Zelman N, Roschger P, et al. Melorheostosis: a clinical, pathologic, and radiologic case series. *Am J Surg Pathol* 2019; 43(11): 1554–1559.
7. McDermott M, Branstetter B and Seethala R. Craniofacial melorheostosis. *J Comput Assist Tomogr* 2008; 32(5): 825–827.
8. Biaou O, Avimadje M, Guira O, et al. Melorheostosis with bilateral involvement in a black African patient. *Joint Bone Spine* 2004; 71(1): 70–72.
9. Kherfani A and Mahjoub H. Melorheostosis: a rare entity: a case report. *Pan Afr Med J* 2014; 18: 251.
10. John B, Sharma A and Pandey R. Managing recurrence in intraarticular melorheostosis involving the knee joint: a case report. *J Orthop Case Rep* 2017; 7(5): 29–33.
11. Murray R and McCredie J. Melorheostosis and the sclerotomes: a radiological correlation. *Skeletal Radiol* 1979; 4(2): 57–71.
12. Fryns J. Melorheostosis and somatic mosaicism. *Am J Med Genet* 1995; 58(2): 199.
13. Pope V, Dupuis L, Kannu P, et al. Buschke-Ollendorff syndrome: a novel case series and systematic review. *Br J Dermatol* 2016; 174(4): 723–729.
14. Hellemans J, Preobrazhenska O, Willaert A, et al. Loss-of-function mutations in LEMD3 result in osteopoikilosis, Buschke-Ollendorff syndrome and melorheostosis. *Nat Genet* 2004; 36(11): 1213–1218.
15. Mumm S, Wenkert D, Zhang X, et al. Deactivating germline mutations in LEMD3 cause osteopoikilosis and Buschke-Ollendorff syndrome, but not sporadic melorheostosis. *J Bone Miner Res* 2007; 22: 243–250.
16. Wordsworth P and Chan M. Melorheostosis and osteopoikilosis: a review of clinical features and pathogenesis. *Calcif Tissue Int* 2019; 104(5): 530–543.
17. Kang H, Jha S, Deng Z, et al. Somatic activating mutations in MAP2K1 cause melorheostosis. *Nat Commun* 2018; 9(1): 1390.
18. Birtane M, Eryavuz M, Unalan H, et al. Melorheostosis: report of a new case with linear scleroderma. *Clin Rheumatol* 1998; 17(6): 543–545.
19. Freyschmidt J. Melorheostosis: a review of 23 cases. *Eur Radiol* 2001; 11(3): 474–479.
20. Sureka B, Mittal M, Udhaya K, et al. Melorheostosis: two atypical cases. *Indian J Radiol Imaging* 2014; 24(2): 192–195.
21. Smith G, Pingree M, Freeman L, et al. Melorheostosis: a retrospective clinical analysis of 24 patients at the Mayo Clinic. *PM R* 2017; 9(3): 283–288.
22. Motimaya A and Meyers S. Melorheostosis involving the cervical and upper thoracic spine: radiographic, CT, and MR imaging findings. *Am J Neuroradiol* 2006; 27(6): 1198–1200.
23. Jha S, Laucis N, Kim L, et al. CT analysis of anatomical distribution of melorheostosis challenges the sclerotome hypothesis. *Bone* 2018; 117: 31–36.

24. Ashish G, Shashikant J, Ajay P, et al. Melorheostosis of the foot: a case report of a rare entity with a review of multimodality imaging emphasizing the importance of conventional radiography in diagnosis. *J Orthop Case Rep* 2016; 6(1): 79–81.
25. Alothman M, Alkhamees L and Al Subaie AM. Melorheostosis of the leg: a case report. *J Radiol Case Rep* 2018; 12(11): 12–17.
26. Hassan A, Khalid M and Khawar S. Detection of melorheostosis in a young lady with upper limb pain on three phase bone scintigram/SPECT-CT. *Clin Cases Miner Bone Metab* 2016; 13(1): 48–50.
27. Yildirim C, Ozyurek S, Cicek E, et al. Melorheostosis in the upper extremity. *Orthopedics* 2009; 32(4): 38064.
28. Jain V, Arya R, Bharadwaj M, et al. Melorheostosis: clinicopathological features, diagnosis, and management. *Orthopedics* 2009; 32(7): 512.
29. Zhang C, Dai W, Yang Y, et al. Melorheostosis and a review of the literature in China. *Intractable Rare Dis Res* 2013; 2(2): 51–54.
30. Ethunandan M, Khosla N, Tilley E, et al. Melorheostosis involving the craniofacial skeleton. *J Craniofac Surg* 2004; 15(6): 1062–1065.