Melorheostosis- melting the myths

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Purpose

Melorheostosis (Leri Disease) is a rare, benign, non-hereditary mesenchymal disorder that most commonly affects the long bones of the lower limbs. Classic melorheostosis manifests as a sclerosing bony dysplasia with irregular cortical and endosteal hyperostosis, giving a highly characteristic “dripping candle wax” appearance on plain radiography [1]. However, melorheostosis may have atypical appearances, involve multiple bones and result in extra-osseous abnormalities, such as intra-articular extension and fibrous or vascular soft tissue masses that may calcify or ossify over time [2].

Classic melorheostosis may be diagnosed on plain radiography alone. However, lack of familiarity with this rare disorder often results in additional but not necessarily contributory forms of imaging. Atypical manifestations of melorheostosis, and its initial detection on cross-sectional imaging, may cause diagnostic confusion with other more aggressive conditions.

This exhibit:

1. Demonstrates an overview of melorheostosis and its various manifestations, and dispels some myths about the disorder.
2. Illustrates the range of multi-modality imaging appearances of classic and less common forms of melorheostosis on plain radiography, CT, MRI and scintigraphy.

Methods and Materials

We have reviewed cases of melorheostosis managed in our unit over the last four years, and present a selection of cases with multimodality images illustrating the various classic and less typical appearances of melorheostosis on plain radiography, CT, MRI and bone scintigraphy.

Results
Clinical Features:

**Myth 1: “Melorheostosis is an asymptomatic disorder”**.

In fact, melorheostosis usually presents with symptoms in late childhood or young adult life. Clinical presentation is often with pain, stiffness, reduced range of movement, contractures, soft tissue masses, limb length discrepancy and deformity. Scleroderma-like skin changes may also occur over the areas of bony abnormality [3].

Some cases of melorheostosis may be asymptomatic for many years, and may be detected incidentally at any age on various forms of imaging.

Melorheostosis often has a chronic progressive clinical course and can lead to significant disability, and may require surgical management of contractures and limb deformities. Medical therapy with bisphosphonate infusion has also been reported to help manage symptoms in some cases [4].

Disease Patterns:

**Myth 2: “Melorheostosis is a disorder of long bones only”**.

Melorheostosis most frequently affects appendicular long bones, particularly in the lower limb. However, it may rarely involve the thoracic cage, skull and spine. Involvement of the axial skeleton usually requires cross-sectional imaging for evaluation and to help distinguish it from aggressive disorders. Melorheostosis may also affect multiple bones in the same region or side of the body, which may be in a sclerotome distribution [5].

**Myth 3: “Melorheostosis is only a disorder of cortical bone”**.

In fact, melorheostosis is a mesenchymal disorder, which can result in a sclerosing bony dysplasia as well as extra-osseous soft tissue masses. The hyperostosis, which typically affects the outer cortex, may also involve the endosteum and extend into the medullary cavity. Intra-articular extension may also be seen resulting in joint ankylosis and contractures [6-7]. Symptomatic forms of melorheostosis often relate to the extra-osseous manifestations of this disorder, particularly para-articular soft tissue masses that interfere with movement or cause compressive symptoms.
Imaging Appearances:

**Plain radiography:**

Plain radiographs remain invaluable in diagnosing the classic features of melorheostosis. Melorheostosis most commonly results in irregular cortical and endosteal thickening on one side of a single tubular bone, usually in a lower limb long bone, with a proximal to distal pattern of extension. This gives the characteristic "melting" or "dripping candle wax" appearance of the cortical changes seen in melorheostosis, which is most easily appreciated on plain radiography or on coronal and sagittal reconstructed CT images.

**Computed Tomography (CT):**

CT is useful for assessing the calcified soft tissue components, intra-articular extension, and for surgical planning. The variably mineralised extra-osseous manifestations may be a direct extension of the osseous abnormality or be separate from the bony changes. CT also helps demonstrate the degree of medullary cavity obliteration by endosteal extension. Axial imaging is particularly valuable for evaluating uncommon sites of involvement such as the spine, where there may be encroachment of neural structures by direct osseous extension or by adjacent soft tissue masses.

**Magnetic Resonance Imaging (MRI):**

MRI confirms the signal characteristics of the osseous components and helps exclude aggressive features such as bone marrow infiltration that may be seen with malignant disorders. The hyperostotic cortical and subcortical changes seen in melorheostosis demonstrate low signal intensity on T1- and T2-weighted sequences in keeping with sclerosis. MRI is particularly useful for evaluating the extra-osseous components and their anatomical relationships to adjacent structures. The soft tissue masses have variable signal intensity characteristics depending upon their composition and degree of mineralisation. Following administration of intravenous gadolinium contrast, no enhancement of the cortical hyperostosis is seen, but there is variable enhancement of the soft tissue masses [8].

\[^{99m}\text{Tc}}\text{-methylene diphosphonate (MDP) bone scintigraphy:}\]
Bone scintigraphy may demonstrate moderate to intense asymmetrical cortical uptake of tracer in the hyperostotic segments, which may cross joints to involve adjacent bones, whilst no abnormal uptake is seen in the adjacent uninvolved medullary cavity [9-10]. Bone scintigraphy is generally not necessary in the imaging assessment of melorheostosis.

Pathology:

Melorheostosis can be diagnosed by its characteristic appearances and the lack of aggressive features on imaging, and it does not generally require biopsy. If biopsied, histological evaluation of the bony abnormality shows thickened and enlarged cortical bone. The haversian canals are normal but may have an irregular arrangement, and the bone marrow space may exhibit increased cellularity. The soft tissue components may exhibit fibrous tissue with variable mineralisation [11].

Case Series:

We present a selection of cases of melorheostosis to illustrate the range of multimodality imaging features described above.

Case 1: Classic melorheostosis of the femur with cortical hyperostosis on plain radiography and MRI (Figures 1-3).

Case 2: Classic melorheostosis of tibia with marked cortical and endosteal hyperostosis on plain radiography and CT (Figures 4-7).

Case 3: Melorheostosis involving multiple right lower limb bones with soft tissue calcification on plain radiography (Figures 8-10).

Case 4: Melorheostosis involving multiple regional bones and soft tissue sites on the same side of the body, on plain radiography (Figures 11-15).

Case 5: Melorheostosis at an uncommon site- the thoracic cage, on plain radiography (Figure 16).

Case 6: Melorheostosis at a rare site- the lumbar spine, on plain radiography, CT and MRI (Figures 17-20).
Case 7: Melorheostosis incidental to other diseases- Rheumatoid arthritis, on plain radiography of the hands (Figure 21).

Case 8: Melorheostosis of the distal femur and proximal tibia with a partially ossified soft tissue mass on plain radiography and MRI (Figures 22-25).

Case 9: Osseous and extra-osseous manifestations of melorheostosis around the right hip on multi-modality imaging- plain radiograph, CT, MRI and bone scintigraphy (Figures 26-30).

Case 10: Melorheostosis around the knee with intra-articular ossified components, on plain radiography and MRI (Figures 31-32).

Images for this section:
Fig. 1: Case 1: Classic melorheostosis of the femur. Plain lateral radiograph demonstrates irregular cortical hyperostosis involving only one side of the femur, along the anterior distal diaphysis (arrow), giving a classic "dripping candle wax" appearance.
Fig. 2: Case 1: Classic melorheostosis of the femur. Plain anteroposterior radiograph shows the irregular hyperostosis of the anterior femoral cortex projected over the centre of the distal femur (arrow).

Fig. 3: Case 1: Classic melorheostosis of the femur. Sagittal proton density weighted MR sequence demonstrates very low signal intensity corresponding to the cortical hyperostosis in the anterior distal femur (arrow).
Fig. 4: Case 2: Classic melorheostosis of tibia. Plain anteroposterior radiograph demonstrates marked cortical (arrowhead) and endosteal (arrow) hyperostosis of the anterior tibial diaphysis.
**Fig. 5:** Case 2: Classic melorheostosis of tibia. Plain lateral radiograph demonstrates marked cortical (arrowhead) and endosteal (arrow) hyperostosis of the anterior tibial diaphysis.
**Fig. 6:** Case 2: Classic melorheostosis of tibia. Sagittal CT shows marked cortical (arrowhead) and endosteal (arrow) hyperostosis of the anterior tibial diaphysis. No soft tissue mass is evident.

**Fig. 7:** Case 2: Classic melorheostosis of tibia. Axial CT shows marked cortical (arrowhead) and endosteal (arrow) hyperostosis of the anterior tibial diaphysis. The endosteal thickening is extending into the medullary cavity.
Fig. 8: Case 3: Melorheostosis involving multiple right lower limb bones with soft tissue calcification. Plain anteroposterior radiograph of the fibula shows wavy cortical...
hyperostosis with a classic "dripping candle wax" appearance (arrow). Soft tissue calcification is projected over the ankle, better seen in Figure 9.

**Fig. 9:** Case 3: Melorheostosis involving multiple right lower limb bones with soft tissue calcification. Plain lateral radiograph of the foot and ankle shows irregular cortical hyperostosis of the distal fibula and a metatarsal (arrows), and soft tissue calcification in the posterior ankle joint recess and over the dorsum of the foot (arrowheads).
Fig. 10: Case 3: Melorheostosis involving multiple right lower limb bones with soft tissue calcification. Plain dorsal-plantar radiograph of the foot shows irregular cortical hyperostosis of the distal fibula, lateral mid foot, fourth metatarsal, and third proximal, middle and distal phalanges (arrows). There is soft tissue calcification projected over the lateral tarsal bones and the base of the lateral metatarsals (arrowheads).

Fig. 11: Case 4: Melorheostosis involving multiple regional bones and soft tissue sites. Plain anteroposterior radiograph of the right shoulder shows marked hyperostosis of the clavicle, scapula and humerus. Soft tissue calcification is projected over the infraclavicular region and shoulder. A walking crutch is also evident, hinting at lower limb problems.
Fig. 12: Case 4: Melorheostosis involving multiple regional bones and soft tissue sites. Plain lateral radiograph of the right elbow demonstrates cortical hyperostosis of the humerus, radius and ulna (arrows) and para-articular soft tissue calcification (arrowhead).
**Fig. 13:** Case 4: Melorheostosis involving multiple regional bones. Plain dorsal-palmar radiograph of the right hand shows cortical hyperostosis of the right thumb metacarpal and phalanges, trapezium, trapezoid, scaphoid and distal radius.
Fig. 14: Case 4: Melorheostosis involving multiple regional bones. Plain oblique radiograph of the right foot demonstrates florid hyperostosis of the first to third toe phalanges, metatarsals, medial tarsal bones and distal tibia. There is ankylosis across the first metatarso-phalangeal joint.
Fig. 15: Case 4: Melorheostosis involving multiple regional bones. Plain anteroposterior radiograph of the right hip illustrates extensive cortical thickening and sclerosis of the right hemipelvis and femur with secondary osteoarthritis of the hip joint, which was subsequently replaced by a total hip prosthesis.

Fig. 16: Case 5: Melorheostosis at an uncommon site- the thoracic cage. Plain postero-anterior radiograph demonstrates hyperostosis of the right second rib with adjacent soft tissue calcification.
**Fig. 17:** Case 6: Melorheostosis at a rare site- the lumbar spine. Plain coronal radiograph demonstrates dense lobular ossification projected over the right side of L4 and L5 vertebrae and adjacent paravertebral soft tissues.
**Fig. 18:** Case 6: Melorheostosis at a rare site- the lumbar spine. Right para-sagittal CT image demonstrates hyperostosis of the L4 and L5 vertebral bodies and posterior elements on the right side. There is also dense lobular ossification in the posterior paravertebral soft tissues.

![CT image of the lumbar spine](image1)

**Fig. 19:** Case 6: Melorheostosis at a rare site- the lumbar spine. Axial CT image on bony windows demonstrates hyperostosis of the right side of the L5 vertebral body and posterior elements, encroaching the neural canal. There is also dense lobular ossification in the right posterior paravertebral soft tissues.

![Axial CT image](image2)
Fig. 20: Case 6: Melorheostosis at a rare site- the lumbar spine. Axial T2-weighted MR image at the same level as the CT in Figure 19 demonstrates very low signal intensity corresponding to the hyperostosis on the right side of the L5 vertebral body and posterior elements and the ossified posterior paravertebral soft tissues. There is extension of the hyperostosis into the neural canal with mild displacement of the thecal sac.
Fig. 21: Case 7: Melorheostosis incidental to other diseases. Plain dorsal-palmar radiograph of both hands shows a symmetrical erosive arthropathy with features consistent with Rheumatoid arthritis. In addition, there is a longitudinal area of cortical hyperostosis in the left little finger middle phalanx (arrow) in keeping with melorheostosis.
Fig. 22: Case 8: Melorheostosis of the distal femur and proximal tibia with a partially ossified soft tissue mass. Anteroposterior plain radiograph of the knee demonstrates irregular thickening and sclerosis of the distal medial femoral and proximal tibial cortices (arrows). There are further longitudinal bands of endosteal sclerosis extending to the medial femoral condyle. The medial tibial plateau also demonstrates cortical hyperostosis. There is also soft tissue swelling around the distal medial thigh with multiple foci of ossification (arrowheads).
Fig. 23: Case 8: Melorheostosis of the distal femur and proximal tibia with a partially ossified soft tissue mass. Coronal proton density MR image of the knee demonstrates very low signal intensity corresponding to the areas of irregular thickening and sclerosis in the distal medial femoral cortex (arrow), medial femoral condyle and proximal tibia. There is a predominantly fatty signal intensity soft tissue mass in the distal medial thigh (arrowhead) containing multiple foci of very low signal intensity in keeping with ossification (curved arrow).
Fig. 24: Case 8: Melorheostosis of the distal femur and proximal tibia with a partially ossified soft tissue mass. Coronal proton density fat-suppressed MR image of the knee at the same level as Figure 23, demonstrates very low signal intensity corresponding to the areas of irregular thickening and sclerosis in the distal medial femoral cortex (arrow), medial femoral condyle and proximal tibia. The majority of the signal intensity of the soft tissue mass in the distal medial thigh (arrowhead) is suppressed on this sequence, confirming fatty content. The soft tissue mass contains multiple foci of very low signal intensity in keeping with ossification (curved arrow). Areas of fluid signal intensity seen within the soft tissue mass are compatible with oedema. No bone marrow oedema is seen in relation to the hyperostotic segments of melorheostosis.

Fig. 25: Case 8: Melorheostosis of the distal femur with a partially ossified soft tissue mass. Axial proton density fat-suppressed MR image of the knee demonstrates very
low signal intensity corresponding to the longitudinal bands of sclerosis in the medial femoral condyle (arrow). The soft tissue mass in the distal medial thigh extending to the medial patellofemoral joint is comprised of predominantly fatty signal intensity, which is suppressed on this sequence (arrowhead). The soft tissue mass also contains multiple foci of very low signal intensity in keeping with ossification (curved arrow). There is patchy fluid signal intensity in keeping with oedema evident within the mass and most marked around the ossified components. There is also cortical thinning and irregularity of the distal medial femoral condyle suggestive of mechanical erosion secondary to the adjacent partially ossified mass. No bone marrow oedema is evident.
**Fig. 26:** Case 9: Osseous and extra-osseous manifestations of melorheostosis around the right hip. Anteroposterior plain radiograph of the right hip demonstrates sclerosis and thickening of the medial cortex of the femoral head and proximal diaphysis (arrows). There is also an area of soft tissue calcification inferomedial to the hip joint (arrowhead).

**Fig. 27:** Case 9: Osseous and extra-osseous manifestations of melorheostosis around the right hip. Axial CT image of the right hip on bony windows demonstrates a heterogeneous soft tissue mass inferomedial to the hip joint and around the distal iliopsoas tendon, which contains multiple foci of calcification (arrowhead) and adjacent fluid density (arrow).
**Fig. 28:** Case 9: Osseous and extra-osseous manifestations of melorheostosis around the right hip. Coronal T1-weighted MR image of the right hip demonstrates very low signal intensity corresponding to the sclerosis and thickening seen in the medial cortex of the femoral head and proximal diaphysis (arrows). There is also a soft tissue mass of predominantly intermediate signal intensity inferomedial to the hip joint, which contains areas of low signal intensity in keeping with soft tissue calcification (arrowhead) as seen on plain radiography and CT.

![Coronal T1-weighted MR image of the right hip demonstrating osseous and extra-osseous manifestations of melorheostosis around the right hip.](image)

**Fig. 29:** Case 9: Osseous and extra-osseous manifestations of melorheostosis around the right hip. Axial proton density fat-suppressed MR image of the right hip demonstrates a heterogeneous soft tissue mass inferomedial to the hip joint and around the distal iliopsoas tendon, which contains areas of low signal intensity in keeping with soft tissue calcification (arrowhead). There is also surrounding fluid signal intensity (arrow).
Fig. 30: Case 9: Osseous and extra-osseous manifestations of melorheostosis around the right hip. 99mTc-methylene diphosphonate (MDP) whole body bone scintigraphy with a spot lateral view of the right femur demonstrates areas of moderate to intense tracer uptake in the cortex of the right medial femoral head and medial proximal femoral diaphysis, corresponding to the hyperostotic segments seen on plain radiography, CT and MRI. No abnormal uptake is seen in the adjacent uninvolved medullary cavity.
**Fig. 31:** Case 10: Melorheostosis around the knee with intra-articular ossified components. Lateral knee radiograph demonstrates irregular cortical and endosteal hyperostosis in the anterior distal femoral meta-diaphysis and proximal tibial diaphysis (arrows), and also in the patella. There is a calcified soft tissue mass projected over Hoffa's fat pad and the anterior tibiofemoral joint recess (arrowhead).
Fig. 32: Case 10: Melorheostosis around the knee with intra-articular ossified components. Sagittal gradient echo MR image demonstrates very low signal intensity corresponding to the areas of cortical and endosteal hyperostosis in the anterior distal femoral meta-diaphysis (arrow) and in the patella. There is a calcified soft tissue mass in the anterior tibiofemoral joint recess associated with an effusion and extending to Hoffa's fat pad (arrowhead). Whilst there is a differential diagnosis (such as synovial osteochondromatosis or pigmented villonodular synovitis) for the intra-articular MRI appearances alone, their co-existence with the characteristic cortical hyperostosis in the anterior femur, tibia and patella make the overall diagnosis in this case most in keeping with melorheostosis.
Conclusion

Melorheostosis is a rare and pathologically benign disorder, but can result in significant pain and functional disability. It may present with classic or less common patterns of cortical bone and soft tissue abnormality that may cause a diagnostic dilemma when detected incidentally on cross-sectional imaging. Familiarity with the range of manifestations and imaging appearances of melorheostosis may enable this rare condition to be diagnosed on imaging without the need for biopsy.

References


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