Melorheostosis in a 12-year-old child

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Abstract

Melorheostosis (of Leri and Joanny) is a rare, non-genetic sclerotic dysplatic bone disorder presenting at any age, usually from late childhood to adulthood. Its aetiology is unknown. It affects mainly the long bones of the upper and lower limbs, but also the short bones of the hand and foot and, rarely, the axial skeleton. Onset is usually insidious, with pain, deformity of the extremity, limb stiffness and limitation of joint motion, as well as thickening and fibrosis of the overlying skin and muscle atrophy being the common clinical features found on presentation. In adults the characteristic radiographic appearance consists of irregular hyperostotic changes of the cortex, generally on one side of the bone, resembling melted wax dripping down one side of a candle, from which melorheostosis derives its name. This, however, is not a feature of the radiographic appearance in children, where the external cortical margin retains its regular outline. Children more often present with a discrepancy in limb size rather than with pain (as is seen in adults). We present our case of melorheostosis in a 12-year-old child to highlight the main clinical and radiological differences found between children and adults.

Key words
Sclerosis, bone dysplasia, cortical hyperostosis, medullary encroachment, limb asymmetry, growth disturbances

Case report

A 12-year-old male presented with a small left hand. On examination there was clinodactyly of all the fingers of the left hand. There was stiffness of the wrist and all the metacarpophalangeal joints, with restricted range of movement both in flexion and extension. The overlying skin was indurated and the first web space was tight. The patient was tender to palpation over the wrist joint and the third carpometacarpal joint.

Plain films of both hands and the left forearm were done. The right hand was normal. All the left carpal bones were smaller and diffusely sclerotic compared with the right side. The left hand was globally smaller than the right (Figure 1). There was cortical sclerosis with thickening extending into the medullary cavities of all five metacarpals as well as of the proximal phalanx of the thumb. The phalanges of the left middle and ring

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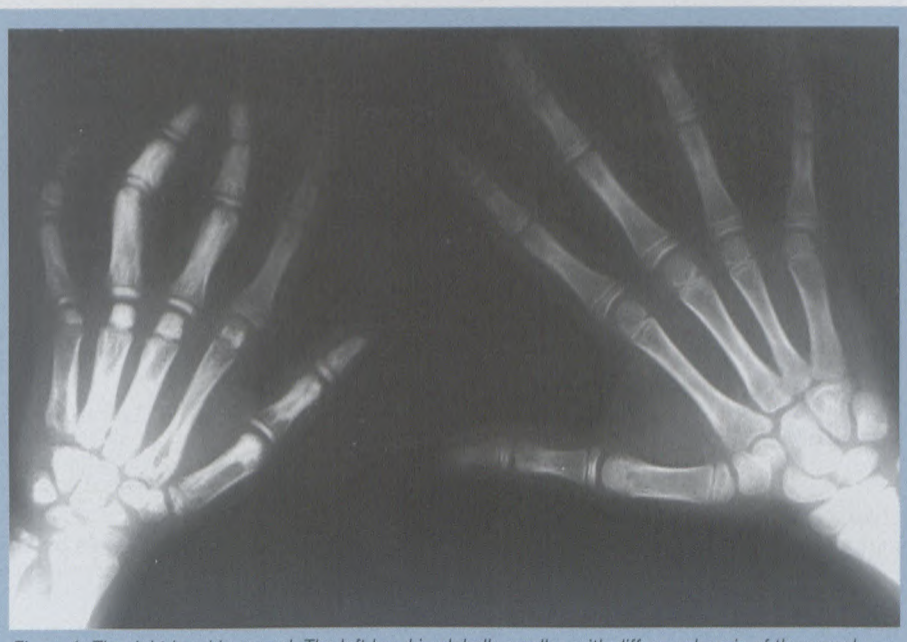

Figure 1: The right hand is normal. The left hand is globally smaller, with diffuse sclerosis of the carpal bones. The first web space on the left is smaller than on the right.

fingers showed diffuse sclerosis. There was also expansion and loss of tubulation in two of the metacarpals involved and in all components of the ring finger. The cortical margin, however, retained a regular outline throughout (Figure 2). Both the left radius and ulna showed cortical thickening in the diaphyseal region. The ulna was affected to a greater degree, with both anterior and posterior cortices showing thickening. The radial involvement was restricted to the medial cortical aspect. There was encroachment on the medullary cavity in both bones, but the external cortical margin was not disturbed (Figure 3).

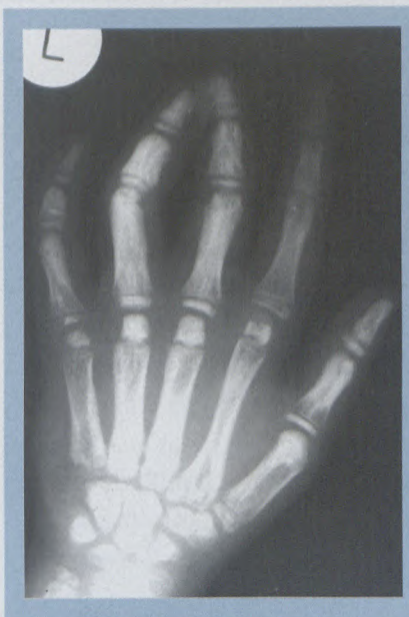

Figure 2: There is cortical sclerosis with thickening extending into the medullary cavities of all five metacarpals as well as in the proximal phalanx of the thumb. The phalanges of the left middle and ring fingers showed diffuse sclerosis. There is expansion and loss of tubulation in two of the metacarpals involved and in the components of the ring finger. The cortical margins retain their regular outline.

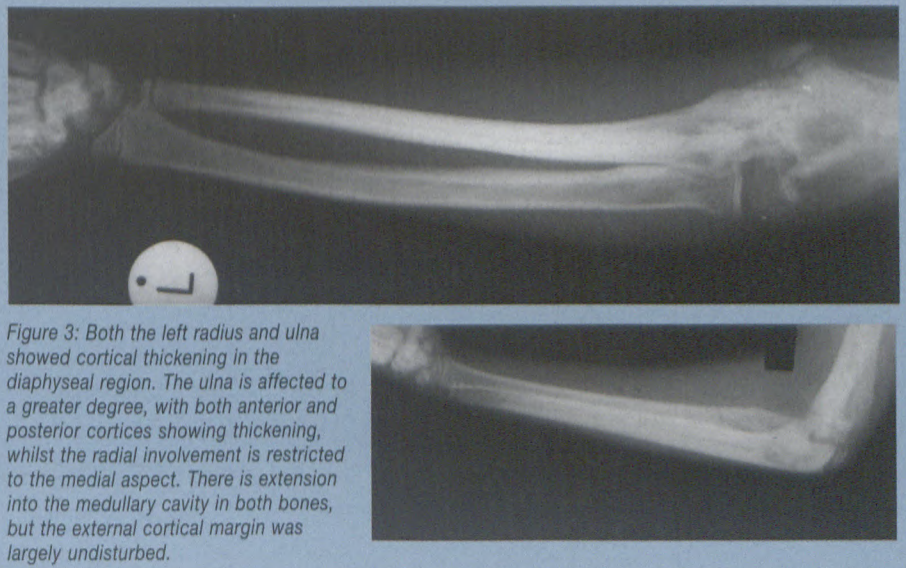

Figure 3: Both the left radius and ulna showed cortical thickening in the diaphyseal region. The ulna is affected to a greater degree, with both anterior and posterior cortices showing thickening, whilst the radial involvement is restricted to the medial aspect. There is extension into the medullary cavity in both bones, but the external cortical margin was largely undisturbed.

The diagnosis of melorheostosis was made radiologically on the basis of a multifocal unilateral sclerotic bone disease with retarded growth associated with the positive clinical features of a small limb, tenderness, joint stiffness with limited range of movement and overlying skin changes.

Discussion

Melorheostosis of Leri and Joanny, first described in 1922, is a rare non-genetic slowly progressive sclerosing skeletal dysplasia that usually presents late in childhood and affects the sexes equally. Common clinical manifestations include bone pain (encountered more in adults), joint stiffness, asymmetry of limb size and skin and soft tissue lesions. The asymmetry of limb size was the presenting feature in our patient. The bone pain is in contrast to other bone dysplasias. It may coexist with osteopoikolosis and osteopathia striata as well as with tumours or malformations of blood vessels or lymphatics. Up to 1994, 320 cases had been reported.

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Radiological features include mono- or polyostotic limb involvement (including hands, feet, shoulder girdle and pelvis). The tubular bones of the lower limbs are the sites of predilection. Less commonly the axial skeleton is involved. The abnormality is usually limited to a single bone or limb and sometimes may skip a segment. Even when the disorder is seen bilaterally, the abnormalities are never symmetrical. Linear cortical hyperostosis following the long axis of the bones is seen with extension into the medullary cavity. This is a predominant feature in our patient. Usually only one side of the bone is affected, as is seen in the left radius of our patient. Sclerosis may be complete in the carpal and tarsal bones, as was the case in the carpal bones of our patient. The epiphyseal growth plate may close prematurely, resulting in growth discrepancies. This is clearly demonstrated in our case, where all the bones of the left wrist and hand were smaller than those on the right.

It is important to note that the "flowing wax" appearance seen in adults, when the linear density extends beyond the cortex as multiple periosteal outgrowths, is not a feature in children. In childhood the hyperostosis does not extend beyond the boundaries of the cortex and the external contours of the bone are undisturbed.

Conclusion

The above features are so distinctive that the unilateral endosteal sclerosis (even when the "flowing wax" appearance is not present in children) is virtually diagnostic of melorheostosis.

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Computed tomography appearances of focal and diffuse hepatic disease in children

and radiological features. Dynamic multi-phase CT scanning has revolutionised lesion characterisation and is currently the modality of choice for imaging hepatic masses. Consideration of imaging characteristics in conjunction with clinical and laboratory data allows the formulation of a focused differential diagnosis.

Acknowledgements

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A practical approach to the chest x-ray of a child with known or suspected congenital heart disease

Evidence of infection

Patients with CHD are more susceptible to lower respiratory tract infections. It is vitally important that any report on a chest X-ray of a child with CHD mentions the presence or absence of features of infection. Often a lower respiratory tract infection leads to decompensation and admission to hospital. Infection may be acute or chronic and in the South African context features of tuberculosis must be excluded.

Conclusion

With the above approach, the radiologist will hopefully be able to offer useful reports to the clinicians managing patients and thus play an integral role in the multidisciplinary management of patients with congenital heart disease.

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