Chapter 17

Melorheostosis

Historical and Nosological Considerations

Léri and Joanni (1922) described a 39-year-old woman who had ‘hyperostosis flowing along the whole length of the limb.’ Subsequently Léri and Lièvre (1928) termed the condition ‘melorheostosis’ from the Greek melos, a limb, and rheos, a stream.

Numerous reports followed and it became apparent that the disorder had important clinical consequences. Green et al. (1962) discussed the distinction between melorheostosis and osteopoikilosis, and Campbell et al. (1968) reviewed the clinical, radiographic and pathological findings in 14 cases.

Wagers et al. (1972) considered the condition from the dermatological standpoint and Soffa et al. (1975) mentioned that about 5% of patients have linear skin lesions overlying the affected bone. In a review of the literature, Beauvais et al. (1977) commented that about 200 cases had been described. Murray (1978) discussed the features of 30 patients, whose ages ranged from 2 to 76 years, and pointed out that the distribution of the skeletal changes often corresponds to the anatomical arrangement of the sclerotomes. On this basis, he suggested that melorheostosis might be the end result of a sensory nerve lesion.

Clinical Features

Melorheostosis is a slowly progressive disorder which usually presents in late childhood. The manifestations are variable, and the diagnosis may be made by chance following radiographic investigation of obscure skeletal discomfort, limb deformities, or dermal lesions.

In contrast to the majority of sclerosing bone dysplasias, melorheostosis may cause considerable bone pain. This symptom is most common in adulthood and is precipitated by activity or fatigue. Disturbed bone growth in childhood can lead to limb-length discrepancy and secondary problems, including genu valgum and scoliosis.

The skin over the affected bones may be sclerodermatous or indurated and erythematous. The subcutaneous tissues and muscles may be involved and
soft-tissue contractures may compound the patient’s disability. In some individuals with melorheostosis these changes are severe; in others they are entirely absent.

The simultaneous occurrence of arteriovenous aneurysms and melorheostosis was observed by Murray (1951) and Patrick (1969). Isolated reports of concomitant neurofibromatous and tuberous sclerosis are of doubtful significance and there are no other systemic ramifications. Neither pathological fractures nor malignant degeneration have been reported.

Radiographic Manifestations

The classical radiographic feature of melorheostosis in adulthood is heaped-up sclerotic bone which gives the appearance of wax flowing down the side of a candle. In childhood the hyperostosis does not extend beyond the boundaries of the cortex, and external contours of the bone are not disturbed.

Skull, Spine, Chest, and Pelvis

Involvement of the skull, spine and thoracic cage is rare, but when the flat bones are affected, the sclerosis is irregular and patchy. In the pelvis sclerosis is maximal around the acetabula and it may extend across the ilia in a streaky, fan-shaped configuration.

Limbs

The tubular bones are the site of predilection and the lower limbs are more frequently involved than the arms. The abnormalities are usually limited to a single bone or limb and sometimes skip a segment. They may be bilateral, but involvement is never symmetrical.

The longitudinal density encroaches upon the medullary cavity, and usually only one side of the bone is affected. In adults the linear density extends beyond the cortex in multiple periosteal outgrowths, which give the 'flowing wax' appearance. Involvement of an epiphysis may be partial but sclerosis may be complete in the small bones of the carpus and tarsus. There may be premature closure of the epiphyseal plate, with unequal growth of an affected limb. Ectopic bone may form in adjacent soft tissues, and bony outgrowths within the joints may limit movement.

Comment

The aetiology of melorheostosis is unknown. Males and females are affected with equal frequency; there is no familial tendency and no evidence to indicate a genetic basis.