Chapter 9

Metabolic bone disorders

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Rickets

Rickets is a clinical syndrome that represents a spectrum of metabolic disorders with similar radiological and histopathological abnormalities resulting from inadequate or delayed mineralization of newly synthesized organic matrix (osteoid) in the immature skeleton before physeal fusion. When seen in adults, the same radiological, biochemical and clinical changes are termed as "osteomalacia" as these refer to the mature skeleton. By definition, rickets is found only in children prior to the closure of the growth plates, while osteomalacia occurs in adults.

Pathophysiology

Vitamin D is a steroid hormone that plays a major role in calcium and phosphorus homeostasis and hence bone mineralization. Vitamin D prohormone undergoes two sequential hydroxylations in the body to become biologically active, the first one being in the liver and then the kidney. Vitamin D in its active form is biochemically 1,25-dihydroxyvitamin D, 1,25(OH_2) D. The second hydroxylation is closely regulated by parathyroid hormone (PTH), calcium, phosphorus and vitamin D. The main targets of vitamin D action at the organ level are the intestine and bone. Vitamin D has two actions that have diametrically opposite outcomes. In conjunction with PTH, it acts on bone, to promote osteolysis by osteoclasts, which results in release and mobilization of calcium from bone to blood. It also mediates the mineralization of organic matrix by calcium and phosphorus deposition. Understanding this basic mechanism is integral in approaching metabolic bone diseases involving abnormalities of vitamin D or PTH.

The etiological factors for rickets may be congenital or acquired. Any factor that interferes with vitamin D metabolism involving intake, its hydroxylation in the liver or kidney, or end-organ resistance to the action of the hormone can lead to rickets. Malnutrition, decreased sun exposure, malabsorption states involving the pancreas, small intestine and liver, and abnormal hydroxylation states are the usual suspects.

As rickets results from a disturbance in metabolism, the underlying disease should be diagnosed. The causes of rickets can be classified into 11 main categories, as shown in Table 9.1.

Imaging findings

The clinical and radiographic features of rickets depend on the age of the patient at which it occurs, the relative maturation of the affected bones and the severity of vitamin D deficiency. There is delayed or abnormal ossification of bone leading to skeletal retardation and osteopenia. The earliest radiographic finding of rickets consists of widening at the growth plate along the longitudinal axis of the bone followed by a decrease in the density of the bone along the metaphyseal side of the growth plate (Figure 9.1). With progressive disease, the widening of the growth plate increases and the zone of provisional calcification becomes irregular. Fraying and disorganization of spongy bone in the metaphyses follows (Figures 9.2, 9.3).

The radiographic changes are maximally seen in zones of active and rapid growth, including the costochondral junctions of the middle ribs, distal femur, proximal humerus, both ends of the tibia and distal ends of the radius and ulna.

In the skull, incessant accumulation of unossified osteoid in the frontal and parietal regions results in the prominence of the frontal bones, called frontal bossing. Other manifestations include Wormian bones, flattening of the posterior skull, basilar invagination and squaring of the skull (craniotabes).

Other key findings including sequelae of bone weakening include deformities of the long bones, both of the shaft and its junction with the cartilage. Genu varum (bow legs) or genu valgum (knock knees) can be seen in toddlers. Anterior bowing of the tibia (saber shin) is also seen. In the pelvis and hips, one may see a triradiate pelvis due to intrusion of the spine into the soft pelvis, appearing as a triflanged-shaped pelvis, and slipped capital femoral epiphysis may be seen. With increasing age, other deformities like scoliosis and bending of long bones may lead to reduced height. Greenstick fractures in the weakened long bones, delayed eruption of teeth, hypoplasia

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