



POSNA

The Core Curriculum

Hypophosphatasia

Objectives

1. Describe the mode of transmission and enzymatic defect noted in patients with hypophosphatasia
2. Describe physical findings noted in patients with hypophosphatasia
3. Discuss laboratory findings in patients with hypophosphatasia
4. Describe radiographic features of hypophosphatasia

Discussion

Hypophosphatasia is a rare autosomal recessive trait, characterized by a defect in synthesis of alkaline phosphatase. Considerable phenotypic variation occurs, ranging from a lethal perinatal form to mild forms manifested by short stature with a tendency toward fracture, pseudofracture, and skeletal deformity. With the defect in alkaline phosphatase activity, the radiographic findings are compatible with osteopenia, and growth plate changes similar to rickets, such as widening of the growth plate. In hypophosphatasia, there are central cup shaped defects in the metaphysis adjacent to the physis that is typical of hypophosphatasia. The diagnosis is established by decreased serum levels of alkaline phosphatase, variations in the severity of the affected alleles appear to be related to the phenotypic heterogeneity. There is no effective treatment at present for the metabolic defect. Carriers have been noted to have decreased bone density and decreased levels of serum alkaline phosphatase. A recent paper describes a salubrious effect of nonsteroidal anti-inflammatory medications for symptomatic children with hypophosphatasia. Successful treatment of bone deformity with intramedullary nailing has been reported but delayed union is not unexpected.

References

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