Hypophosphatasia

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Hypophosphatasia. ORPHA:436

Hypophosphatasia (HPP) is a rare heritable metabolic disorder characterized by defective mineralization of bone and/or teeth in the presence of reduced activity of unfractionated serum alkaline phosphatase (ALP). The clinical spectrum is extremely wide, from stillbirth at one end to fractures of the lower extremities in adulthood, at the other, or even no bone manifestations (odontohypophosphatasia).