Hypophosphatasia

Hypophosphatasia (HPP) is a rare inherited bone disease which affects approximately 1:100,000 people. It is defined by a missing enzyme known as tissue non-specific alkaline phosphatase (TNSALP) which helps calcium and phosphate to be integrated into bone. It is also found in many other tissues in the body. The missing enzyme leads to calcium and phosphate building up in the body, and in severe forms this causes high serum levels of calcium and phosphate as well as kidney calcification (nephrocalcinosis). Also, other metabolites that build up are strong inhibitors of bone formation which means that bone, and teeth, cannot be made properly. This causes fractures, bone pain as well as dental abnormalities.

There are five different forms of this condition; the Perinatal form, Infantile form, Childhood form, Adulthood form and Odontohypophosphatasia. Severity of HPP varies greatly. In general, the more severe forms are the ones that present earlier in life and develop over time.

Synonyms

Alternative names for this condition are:
- HPP

Further information about all forms of HPP is available from Climb.

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