

## Hereditary Hypophosphatasia In A 30 Year Old Female With Chronic Bone Pain

Hereditary hypophosphatasia (HPP) is a rare autosomal recessive disorder, characterized by disrupted mineralization of bones and teeth. It is often caused by loss-of-function mutations in the ALPL gene that encodes the tissue-nonspecific isoenzyme of alkaline phosphatase (ALP). Symptoms include defective mineralization of bone, premature loss of teeth, and decreased serum ALP activity. Severe cases may also include fractures, rickets, and respiratory insufficiency. Chronic bone pain is a common symptom, but less specific, and often preceded by gross deformities. Adult onset HPP can be missed, given its uncommon and complex nature. There have been few reports of HPP presenting in adulthood, which were mistaken for osteoporosis. In the current case, the patient was a 30 year old female with a long history of chronic, progressive bone pain since childhood who presented to her family medicine physician. Her ALP level at the time of the visit was 31 U/L (Ref: 35-147 U/L). She previously had low ALP levels intermixed with low-normal. The persistent, distressing bone pain and decreased ALP levels prompted genetic testing, which revealed an ALPL gene .571G>A (p.Glu191Lys) mutation, indicating HPP. Since diagnosis, she has normal DEXA scans, tibial x-rays, and renal ultrasound. Her course of treatment has mostly consisted of pain management with tapentadol, gabapentin, buprenorphine and her geneticist have discussed treatment with hormone replacement and teriparatide--modified parathyroid hormone that promotes bone growth. HPP is important to consider in patients with chronic bone pain, regardless of the consistency of ALP levels. It is also imperative to distinguish bone pain compared to pain of muscular origin, as seen with fibromyalgia. Diagnosis of HPP may provide more treatment options with teriparatide and ALP replacement.