

Rare Case of Vitamin D Deficiency without Elevation of Alkaline Phosphatase

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1. Introduction

Elevated serum Alkaline phosphatase acts as screening marker of vitamin D deficiency. Failure of its elevation might result in misdiagnosis. Estimation of both radiological findings and serum Alkaline Phosphatase helps in early recognition of metabolic bone diseases. One of the rare differentials that can be considered with normal alkaline phosphatase and with low vitamin D levels being hypophosphatasia, which is a rare genetic disorder characterized by abnormal development of bone and teeth.

2. Case Report

- Hereby presenting a case of a 6-year-old male child who came with complaints of bilateral wrist and knee swelling since 10 days, with bowing of legs, decreased appetite since 8 days. Swelling was gradual and non-progressive in nature.
- On examination swelling bilateral wrist and knee present (non tender),pigeon shaped chest with beading present.
- Severe wasting with stunting was present.
- Double medial malleolus with Bowed legs observed.
- Baseline investigations were done where CBC was within normal limit, ESR 20, serum electrolytes WNL, serum Calcium was low (6.97),serum Phosphorus was low(2.6),serum Magnesium WNL, Alkaline Phosphatase was 122.4,serum PTH was normal (49.5),serum 1,25 OH Vitamin D being low (10.6), VBG done s/o Ph 7.33,HCO₃ 20.5,PCO₂ 39.8.serum creatinine WNL(0.2)
- Xray bilateral wrist and bilateral knee s/o radiological changes like cupping, fraying and metaphyseal widening.

3. Discussion

Alkaline Phosphatase is present in many human tissues such as bone, intestine, kidney, liver and WBC.

Damage of these tissues results in release of alkaline phosphatase in the blood stream.

Major function of alkaline phosphatase is transportation across cell membrane. Normal range of alkaline phosphatase being 44-147 U/L.

Vitamin D deficiency primarily affects the bone as there is lack of mineralization of bones, thus causing elevation of alkaline phosphatase in almost all categories of rickets. Making it serve as an important marker of Vitamin D deficiency.

As per management protocol the patient was started on calcium supplementation and was given 6,00,000IU Vitamin D.

Pt was told to follow up on OPD basis after 4 weeks with repeat Xray so as to see improvement in mentioned radiological changes.

Xray bilateral wrist done after 6 weeks of giving vitamin D showed improvement. Patient is being followed up on OPD basis.