

Infantile Hypophosphatasia: An unusual Presentation and Novel Gene Mutation

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Rec date: 30 Oct, 2014, Acc date: 31 Oct, 2014, Pub date: 2 Nov, 2014

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Clinical Image



Radiographic findings suggesting severe osteopenia, rachitic changes and low alkaline phosphatase levels.

Patient was born at term and was admitted on day 1 of life for seizure like activity. Severe osteopenia changes were discovered incidentally on chest x-ray obtained for NG tube placement at 3 weeks of life. Lab evaluation showed very low alkaline phosphatase levels and hypercalcemia. Skeletal survey showed severe rachitic changes in long bone with metaphyseal fraying widening and cupping.

Analysis of the ALPL gene demonstrated two mutations thus confirming the diagnosis of hypophosphatasia. One of the two mutations was not reported previously.