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TEST YOURSELF: ANSWER

Hypophosphatasia

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Discussion

Hypophosphatasia is a rare inborn error of metabolism that occurs in approximately one per 100,000 live births [1]. Hypophosphatasia is characterized by defective mineralization of bone and/or teeth in the context of low activity of serum and bone alkaline phosphatase(ALP). Reduced serum ALP activity, increasing phosphoethanolamine, inorganic pyrophosphates and pydoxal-5'-phosphate in serum and urine determine the diagnosis of hypophosphatasia [2]. Hypophosphatasia is due to mutations in the alkaline phosphatase gene encoding the tissue nonspecific alkaline phosphatase. The deficiency of ALP limits the availability of phosphate giving rise to serum phosphate decreasing and serum calcium increasing without phosphate to interact with, and the failure to form the Ca-PO4 matrix leads to fragile bones and early loss of teeth. Clinical presentation widely varies, from death in utero to cases in which pathologic fractures first present only in adulthood [3]. Adults may be troubled by recurring fractures in their feet and painful, partial fractures in their long bones. Some patients can be self-limited or self-cure. At present, there is no curative treatment for hypophosphatasia.

Five clinical forms of hypophosphatasia have been reported [4]. The forms of perinatal (lethal), infantile, childhood and adult are determined by the age when their skeletal lesions are discovered. The other form, odontohypophosphatasia, refers to children and adults who have no skeletal problems, but only biochemical and dental manifestations; this usually involves premature loss of teeth.

For adult manifestations of childhood hypophosphatasia and the adult form of hypophosphatasia, an increased incidence of poorly healing stress fractures, especially of the metatarsals, may occur. Pseudo-fractures are one of the hallmarks, often occurring in the lateral aspect of long bones. There is also a predilection for chondrocalcinosis and marked osteoarthropathy later in life [5]. The disease can be distinguished from osteoporosis by specialized biochemical testing.

In the presented case, the adult patient with hypophosphatasia revealed decreased blood ALP (18 U/L, normal value:32-92U/L), increased blood phosphoethanolamine (28.9 μmol/L, normal value: 2.1-3.3 μmol/L) and raised urine phosphoethanolamine (1820 μmol/L, normal value: 170-990 μmol/L) and elevated blood calcium (3.5 mmol/L, normal value:2.3-2.6 mmol/L). The X-ray of the patient manifested widened ends of femurs and tibias, multiple fractures and bowing of bilateral tibias. Besides, stenosis of vertebral canal and osteosclerosis on end plates of lumbar vertebrae were revealed, which was not found to be described in the literature.

The case presentation can be found at doi:10.1007/s00256-012-1456-8.

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