SHORT COMMUNICATION

BIPHOSPHONATES IN HYPOPHOSPHATASIA: NOT THE EVIL?

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Use of biphosphonates is not recommended in hypophosphatasia, because of their potential deleterious effects on bone formation and mineralization. On another hand, managing the hypercalcemia phasis of infantile hypophosphatasia with sustained excess of calcium urine excretion may be a serious concern. We report here the case of a young girl with a severe form of infantile hypophosphatasia. Femur bowing was detected during pregnancy and the diagnosis hypophosphatasia was done during the first month of life with low alkaline phosphatase level and an homozygous mutation of the TNSAP gene (c.896T > C, p.L299P). When 3-months old she developed severe hypercalcemia with high urine calcium level. Usual therapeutics including calcium restriction was inefficient. Decision was taken to administer two intravenous cures of pamidronate (0.5 mg/kg each) in a week, with no further injection. Calcemia was appropriately controlled within the 4 days following injections and, with further significant reduction of the calcium intake, remained stable. In addition, lowering calcium level allowed to free PTH secretion. PTH level increased, and, strikingly, alkaline phosphatase level increased by two folds. Improvement of the growth and weight curves was observed in months following pamidronate administration. She further developed craniosynostosis when 12-years old, but no fractures. Therefore, while not being recommended as chronic treatment biphosphonates can be used without deleterious effect for treating acute phasis of hypercalcemia and hypercalciuria in hypophosphatasia,