Birth Defects, Dysmorphology, Skeletal Dysplasia, Craniofacial Anomalies

AB030. Study of bone turnover markers and treatment monitoring in osteogenesis imperfecta

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Background: Osteogenesis imperfecta (OI), the most prevalent heritable disorders of bone, results from mutations involving several genes responsible for the synthesis or intracellular processing of type I collagen. Bisphosphonates are potent antiresorptive agents, decreasing the incidence of fractures and pain, while increasing energy levels and bone mineral density.

Methods: The study included 26 OI patients and 30 controls. Samples were collected from patients at baseline, 6 months and 1 year from bisphosphonate treatment onset. Samples were subjected to biochemical measurements of serum calcium, inorganic phosphorus, parathyroid hormone, 25(OH)vitamin D, 1,25(OH)2vitamin D, bone formation markers; osteocalcin and procollagen type I N-telopeptide (P1NP) and type I collagen degradation markers; urinary type I collagen helical peptide α1(I), N-telopeptide, non-isomerized C-telopeptide, β-isomerized C-telopeptide, pyridinoline and deoxypyridinoline.

Results: Patients were classified according to their clinical severity into three groups; mild, moderate and severe. No significant difference was observed for the biochemical measurements at different time points in all groups of patients and controls, except parathyroid hormone. All bone formation markers and type I collagen degradation markers showed significant differences at different time points in different patient groups and controls. No correlation was detected between the mode of inheritance and any parameter at any time point. Molecular data were available in 9 patients. Variations in some markers were observed in patients with *SERPINF1* and *CRTAP* gene mutations.

Conclusions: The clinical and genetic heterogeneity of OI were reconfirmed as patients with the same mutated gene showed different clinical severity. Bone formation markers and type I collagen degradation markers are valuable markers for monitoring the effect of bisphosphonate treatment in different clinical groups of patients. However, P1NP is a better bone formation marker. For evaluation of bone metabolism in patients, comparing bone turnover markers according to clinical severity and molecular findings is more informative than mode of inheritance.

Keywords: Osteogenesis imperfecta (OI); bisphosphonate; bone formation markers; type I collagen

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