Features of Nine Adult Cases of Osteogenesis Imperfecta

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Introduction: Osteogenesis imperfecta (OI) is a congenital bone disorder characterized by common osteoporosis, recurrent fractures and deformities. Most cases are caused by mutations in the *COL1A1* and *COL1A2* genes responsible for type-1 collagen production. Treatment of the patients with OI is aimed at increasing bone strength and personal functionality in order to prevent fracture, maintain mobility and decrease pain. We present the features of 9 adult cases of OI.

Cases: Nine cases of OI (Female: 7, Male: 2) were retrospectively investigated. In this study, following parameters were evaluated: the age, gender, family medical history, duration of disease, frequency of fractures, bone mineral density scores, duration and types of medical therapy, bone turnover markers, the level of 25-hydroxy vitamin D. The mean of age of our patients was 24. The age at the time of diagnosis was between 1.5 and 15 years old. Oral alendronate, pamidronat and zoledronic acid were given and no fracture was observed with this bisphosphonates treatment.

Results: Thanks to bisphosphonates treatment, life quality of the patients with OI has increased recently. Although not curative, bisphosphonates treatment has proven to decrease the number of fractures. Lately, no new fractures have been observed in our patients to whom oral alendronate, pamidronat and zoledronic acid were given. Bisphosphonates treatment seems to be the most efficient one until novel treatment modalities are found.

Key words: Osteogenesis imperfecta, osteoporosis, fractures, deformities, bisphosphonates treatment