Effect of Bisphosphonate Therapy in Pediatric Skeletal Dysplasia: A Cross-Sectional Study

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ABSTRACT

Skeletal dysplasia is a group of rare, often progressive genetic disorders that impact bone development and joint function. These conditions commonly present with joint pain, stiffness, and limited mobility, resulting in reduced quality of life in affected children. Bisphosphonates have been explored as a potential symptomatic therapy in some patients with skeletal dysplasia, though evidence remains limited. To assess the clinical characteristics of pediatric patients with skeletal dysplasia in a tertiary center and evaluate the symptomatic and functional outcomes associated with bisphosphonate therapy. This cross-sectional study included 11 pediatric patients diagnosed with various forms of skeletal dysplasia at King Abdulaziz University Hospital. Diagnoses were confirmed clinically and genetically. Musculoskeletal symptoms (joint pain, stiffness, swelling, and range of motion) and functional status were evaluated using structured interviews and validated tools, including the Visual Analog Scale (VAS) for pain and the Short Form-12 (SF-12) for physical (PCS) and mental (MCS) component scores. Seven patients received bisphosphonate treatment (zoledronic acid), allowing for between-group and within-patient comparisons. Data was analyzed using non-parametric statistical tests. The mean age at disease onset was 5.6 ± 3.6 years, with diagnosis delayed until 9.7 ± 5.0 years. The most common diagnosis was Progressive Pseudorheumatoid Dysplasia (PPD). High parental consanguinity (91%) and family history (55%) were observed. At baseline, 82% reported joint pain, 73% had a limited range of motion, and 64% experienced stiffness and swelling. Bisphosphonate-treated patients had lower symptom prevalence than non-users, with significantly higher PCS scores (48.8 ± 14.6 vs. 43.1 ± 9.3; p = 0.03). Within-patient analysis revealed a significant improvement in PCS following treatment (from 30.2 ± 13.4 to 48.8 ± 14.6; p = 0.01) and a trend toward reduction in joint pain (from 100% to 29%; p = 0.06). Bisphosphonate therapy, particularly with zoledronic acid, may offer symptomatic relief and improve physical function in pediatric patients with skeletal dysplasia. Although limited by a small sample size and a short follow-up period, these findings suggest a potential role for bisphosphonates as part of a multimodal management strategy in selected patients. Further longitudinal studies with larger cohorts are warranted to confirm efficacy and long-term safety.

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