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RESPONSE TO PAMIDRONATE THERAPY AND PHARMACOGENETICS IN PATIENTS WITH OSTEOGENESIS IMPERFECTA

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INTRODUCTION/BACKGOUND
Osteogenesis imperfecta (OI), is a genetically heterogeneous connective tissue disorder associated with skeletal fragility, deformity and growth deficiency. Intravenous bisphosphonate therapy is the mainstay of medical treatment of this condition. Due to the scarcity of regional data, this study aimed to investigate the genetic profile and clinical response to bisphosphonate therapy in a cohort of Malaysian patients.

METHODOLOGY
Genetic analysis was performed on 14 children (6 females, 8 males) with OI at Hospital Putrajaya. Three children with Type I and eleven with Type III OI. All patients received bisphosphonate therapy. Clinical, biochemical and radiological data were gathered prior to initiation of treatment and at subsequent intervals during treatment. Targeted gene sequencing using the Ion AmpliSeq platform on the Ion Torrent™ system identified genetic mutations which were validated using Sanger sequencing. In silico analysis evaluated their potential impact at the protein level.

RESULT
All patients had involvement of long bone fractures, with the addition of thoracic or lumbar vertebrae involvement in 50% of patients. A positive family history was noted in 28% of patients. Bisphosphonate therapy was started at a median age of 4 [3.1 - 10.7] years with 42.8% of patients starting treatment before the age of 2. The median duration of treatment was 6.4 (2.8-14.9) years. All patients showed a significant reduction in fracture rate while on intravenous bisphosphonate therapy, with annual fracture rates decreasing from 2.4 to 0.8 fractures per year on average. Additionally, all patients reported an improvement in bone pain. There was no significant difference in fracture rate between COL1A1 and COL1A2 positive patients.

CONCLUSION
This study highlights the genetic heterogeneity of OI patients with COL1A1 and COL1A2 in the Malaysian population and supports the efficacy of pamidronate therapy in improving skeletal outcomes.