Osteogenesis Imperfecta (OI) is known to affect about 1/15,000–20,000 individuals in the world [1]. OI is a rare congenital disorder characterised by bone fragility. Mutation in collagen-related genes results in deficiency of collagen, which forms the bone matrix. This can result in fractures, deformities of the spine, and effects on other organs. A child's perception of pain can be influenced by a variety of circumstances, including repetitive fractures, bone deformities, muscle atrophy, mobility issues, and psychological concerns. Based on clinical characteristics and the course of the disease, OI subtypes are categorised; types I, II, III, and IV. OI Type I is considered the mildest form of the condition, with normal growth patterns and a low incidence of fractures during the perinatal period. Type II OI is more severe and causes death during the prenatal period. In contrast, OI Type III is a more serious presentation of the disorder. People with type III OI have significant bone deformities, and have fractures often even after birth. The most clinically heterogeneous type of OI is Type IV, with members displaying mild to severe symptoms. OI is diagnosed by using ultrasonography, radiography, bone densitometry, and serum electrolytes (calcium and phosphorous). Bone and skin biopsy also helps in diagnosis by studying collagen tissue. Individual differences in treatment response and outcome are due to genetic complexity in each kind of OI. Medicinal treatment (bisphosphonates) is the most common approach used to manage pain but these treatments' primary drawbacks include their comparatively low efficacy, their lack of benefits, the emergence of treatment resistance in certain individuals, or their cytotoxic adverse effects. The multidisciplinary approach to manage OI involves occupational therapy, physical therapy, orthotics and by other health care professionals including orthopaedic surgeon, dieticians, and speech therapists. Ongoing studies have provide new avenues for the management of OI, including medical, rehabilitation, and surgical treatments. Physical therapy maximise mobility and independence while lowering the risk of fracture and bone loss. The physiotherapists play a crucial role by providing counselling to the parents of OI infants based on disease severity and help the children with OI reach adult independence and maintain optimal musculoskeletal health. Parents are advised to handle and position the baby safely, with the ultimate goal of enabling the child to gain mobility on their own, whether that means utilising a wheelchair in the future or walking aids. Observational studies have shown evidence that supervised exercise regimes can enhance physical capacity and muscle strength.

REFERENCES