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Modern approach to children with osteogenesis imperfecta

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Osteogenesis Imperfecta (OI) is characterized by bone fragility. At least seven discrete types have been described ranging from mild disease to a lethal form. In a large number of cases, mutations in one of the two genes encoding type I collagen have been found. In forms recently described (types V, VI, VII), such mutations have been excluded. In two other forms, (Bruck, and osteoporosis - pseudoglioma syndromes) defects in other proteins have been characterized. In OI, bone fragility stems from: decreased bone mass, disturbed organization of bone tissue, and altered bone geometry (size and shape). Histologic studies have shown that increased bone turnover is the rule in OI bone. This justifies using bisphosphonates in order to reduce osteoclast mediated bone resorption. Initial results are encouraging. Cyclical intravenous pamidronate administration reduces bone pain and fracture incidence, and increases bone density and level of ambulation, with minimal side effects. Effects on bone include increase in size of vertebral bodies and thickening of cortical bone. These results allow for more efficacious corrective surgery using intramedullary rodding of the long bones and paravertegral instrumentation. Specific occupational and physiotherapy programs are integral parts of the treatment protocol. This multidisciplinary approach will prevail until strategies aiming at the correction of the basic defect(s) will have come to fruition.

Keywords: osteogenesis imperfecta; bone fragility; collagen type I; osteoporosis; bisphosphonates; intramedullary rodding

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