**Osteoprotegerin Deficiency (Juvenile Paget's Disease): Responses to Oral and IV Bisphosphonates in 3 Children**

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Summary

Osteoprotegerin (OPG), the principal variant of juvenile Paget's disease, is a rare, autosomal recessive disorder in which markedly accelerated bone turnover occurs, leading to subperiosteal bone expansion and deformity. This case series documents responses to oral and intravenous bisphosphonates in 3 children with OPG, with improvements in bone metabolism and radiographic features, and lower markers of bone turnover.

**Introduction**

JP, previously called idiopathic or hereditary hyperphosphatasia, is an autosomal recessive disorder characterized by excessive bone turnover affecting the entire skeleton and presented with bone expansion and deformity along with variable clinical features. The principal variant is associated with OPG deficiency, characterized by increased bone turnover and radiographic features of subperiosteal bone expansion. The principal feature of Paget's disease is increased bone turnover affecting the entire skeleton and presenting with bone expansion and deformity along with variable clinical features. The principal feature of JPD is increased bone turnover affecting the entire skeleton and presenting with bone expansion and deformity along with variable clinical features. The principal feature of JPD is increased bone turnover affecting the entire skeleton and presenting with bone expansion and deformity along with variable clinical features. The principal feature of JPD is increased bone turnover affecting the entire skeleton and presenting with bone expansion and deformity along with variable clinical features.

**Discussion**

Osteoprotegerin (OPG) deficiency, the principal variant of juvenile Paget's disease, is a rare, autosomal recessive disorder in which markedly accelerated bone turnover occurs, leading to subperiosteal bone expansion and deformity. This case series documents responses to oral and intravenous bisphosphonates in 3 children with OPG, with improvements in bone metabolism and radiographic features, and lower markers of bone turnover.

**Case Descriptions**

**Patient 1**

A 17-year-old female presented with severe bone pain, torticollis, and decreased mobility. Radiographs showed subperiosteal bone expansion affecting the entire skeleton. Serum P and urinary calcium excretion remained elevated, but his ESR, ALP, and CRP were normal (Table). Radiographs showed a better defined bone cortices with improved mineralization of all bones.

**Patient 2**

A 15-year-old male presented with severe bone pain and large subperiosteal bone expansion affecting the entire skeleton. Serum P and urinary calcium excretion remained elevated, but his ESR, ALP, and CRP were normal (Table). Radiographs showed a better defined bone cortices with improved mineralization of all bones.

**Patient 3**

A 14-year-old male presented with severe bone pain and large subperiosteal bone expansion affecting the entire skeleton. Serum P and urinary calcium excretion remained elevated, but his ESR, ALP, and CRP were normal (Table). Radiographs showed a better defined bone cortices with improved mineralization of all bones.

**References**