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Endoped Abstract

Osteogenesis Imperfecta-Related Osteoporosis: Between Low Bone Mineral Density and Multiple Fractures

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Abstract: Osteoporosis is reflected by low bone mineral density (BMD) at central Dual-Energy X-ray Absorptiometry (DXA) as well as clinical complications like low-trauma or spontaneous fractures. While typical primary osteoporosis is menopause-related, among the secondary causes of osteoporosis, osteogenesis imperfecta (OI) is listed for children, teenagers and adults. Underlining more than 17 mutations, and a heterogeneous clinical presentation, decreased BMD is associated with multiple fractures and impaired peak bone mass with lifelong effects.

Keywords: bone; fracture; osteoporosis; osteogenesis imperfect; skeleton

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Background

Osteoporosis is reflected by low bone mineral density (BMD) at central Dual-Energy X-ray Absorptiometry (DXA) as well as clinical complications like low-trauma or spontaneous fractures [1,2]. While typical primary osteoporosis is menopause-related, among the secondary causes of osteoporosis, osteogenesis imperfecta (OI) is listed for children, teenagers and adults [3,4]. Underlining more than 17 mutations, and a heterogeneous clinical presentation, decreased BMD is associated with multiple fractures and impaired peak bone mass with lifelong effects [5].

Aims

Our purpose is to introduce two index cases coming from two different families with or without specific therapy for severe osteoporosis during transition to adulthood.

Material and Methods

This is a case series.

Results

Case 1

This is a 24-year old female known with osteogenesis imperfecta (OI) and secondary osteoporosis since the age of 16 and multiple fragility fractures since early childhood. Her mother is OI +ve and she also has one brother and one sister that are OI-negative. At age of 16, lumbar BMD was 0.8 g/sqcm, Z-score of −3 SD and intravenous (IV) ibandronat was offered to her for 5 years with a BMD increase to 0.933 g/sqcm, Z-score of −1.3 SD, but a new asymptomatic fracture at lumbar L1 vertebra was detected at X-Ray so she was switched to yearly zolendronic acid 5 mg. After 2 years, L BMD was increased to 0.951 g/sqcm, Z-score of −1.2 SD, thus, for the moment, drug holiday was initiated; only cholecalciferol 1000 UI/day was continued. No side effect to IV bisphosphonates was registered.

Case 2

This is a 25-year old male (one brother OI-negative) with a history of >15 fractures since the age of 9 months, without prior medication, who has an L BMD 0.689 g/sqcm, Z-score −4 SD and moderate T 7,8,9 vertebral fractures at computed tomography (CT) scan. His 49-year father, also OI +ve, without previous medication, and multiple fractures, has an L BMD 0.776 g/sqcm, Z-score −2.9 SD and decreased T9-L5 vertebras height based on CT analyzes. For both patients, alkaline phosphatase was mildly increased to 160 U/L, respectively 134 U/L (normal: 38–129 U/L). Weekly oral alendronate 5600 U in addition to cholecalciferol 1000 UI/day was initiated.

Conclusions

Bisphosphonates are useful for OI-related OP. The decision of therapy is related to improving the peak bone mass potential in teenagers and young adults.

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Conflicts of Interest: The authors declare no conflict of interest.

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