ACTA RADIOLÓGICA PORTUGUESA
September-December 2024 | Vol 36 nº 3 | 33-34 https://doi.org/10.25748/arp.33668

Images of Interest / Imagens de Interesse

Osteoporosis-Pseudoglioma Syndrome, a Rare Entity

Síndrome Osteoporose-Pseudoglioma, uma Entidade Rara

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Received: 18/11/2023
Accepted: 29/01/2024
Published: 31/12/2024

Abstract

Osteoporosis-pseudoglioma syndrome is a rare autosomal recessive disorder of severe juvenile osteoporosis and congenital blindness. This disease is characterized by intellectual disability, osteoporosis of bones and eye abnormalities. It is usually diagnosed in early childhood, with affected children displaying early onset blindness, severe osteoporosis, short stature and fractures. On plain radiographs, patients usually manifest severe thinning of bones, bowing of extremities, and spinal deformities.

Keywords

Osteoporosis-pseudoglioma syndrome; Blindness; Juvenile osteoporosis.

Resumo

A síndrome de osteoporose-pseudoglioma é uma doença rara autossómica recessiva que se caracteriza por osteoporose grave na infância e cegueira congénita. Esta patologia é também caracterizada por deficiência intelectual, fragilidade óssea e anomalias oculares. Geralmente, é diagnosticada na infância, com as crianças afetadas a apresentarem cegueira precoce, osteoporose grave, estatura reduzida e fraturas. Nas radiografias simples, os doentes apresentam-se com afilamento acentuado dos ossos, curvatura dos membros e deformidades na coluna vertebral.

Palavras-chave

Síndrome osteoporose-pseudoglioma; Cegueira; Osteoporose juvenil.

Case Presentation

An 18-year-old male, with intellectual disability, presented with congenital blindness and a history of repeated fractures since early childhood, including a fracture of the right femur at 14 years of age. The patient is a refugee from a sub-developed country and no further medical history was available.
On clinical examination, there is marked scoliosis as well as bilateral microphthalmia and anophthalmia.
A typical skeletal survey was performed using conventional x-ray with bilateral projections of hands, forearms, humerus, feet, leg, femur, pelvis, spine, and skull.
Frontal and lateral skull radiographs revealed generalized cortical thinning as well as a reduction of the orbital spaces (Figure 1).
AP x-ray of the spine shows dorsal and lumbar dextroscoliosis (Figure 2).
AP x-ray of the forearms shows bowing of the radius and ulna, as well as thinning of the diaphysis and widening of the metaphysis (Figure 3).
Also showing the bowing of the forearm bones.
Chilaiditi syndrome was observed.
Bone mineral density (BMD) was measured by dual energy X-ray absorptiometry (DXA), confirming osteoporosis.

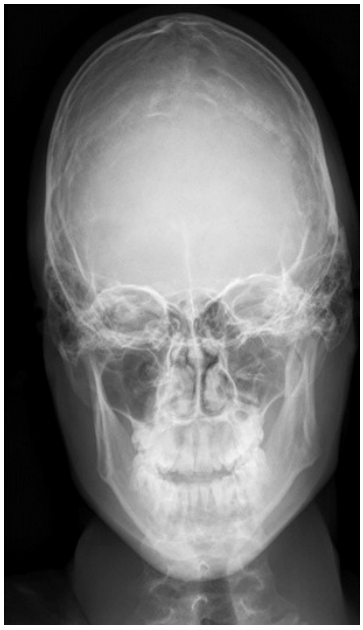

Figure 1 – AP x-ray of the skull shows generalized cortical thinning as well as reduction of the orbital spaces.

Discussion

Osteoporosis-pseudoglioma syndrome (OPPG) is a rare autosomal recessive disorder of severe juvenile osteoporosis and congenital blindness.[1] It is caused by inactivating mutations in the gene encoding low-density lipoprotein receptor-related protein 5 (LRP5), responsible for the Wnt/β-catenin signaling pathway. To date, around 80 cases have been reported worldwide and the estimated prevalence is 1/2.000.000.[1] A large proportion of cases emerges in populations with a high rate of consanguinity.
OPPG is characterized by intellectual disability, osteoporosis of bones and eye abnormalities. It is usually diagnosed in early childhood, with affected children displaying early onset blindness, severe osteoporosis, short stature and fractures.

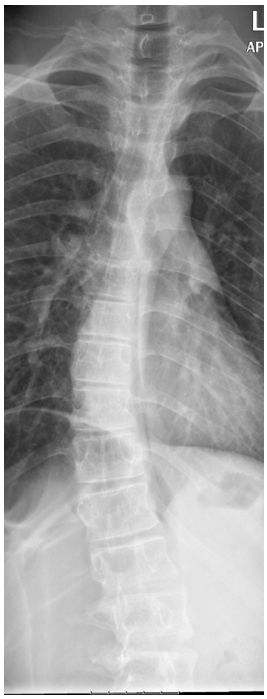

Figure 2 – AP x-ray of the spine with dorsal and lumbar dextroscoliosis.

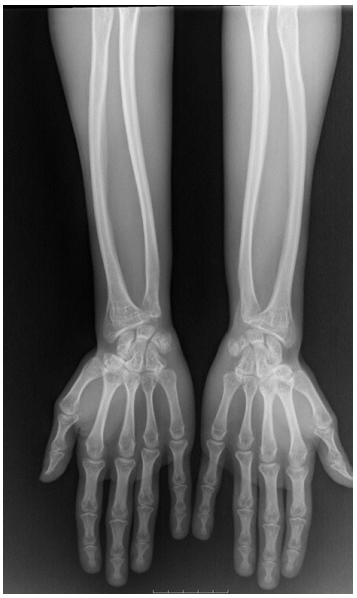

Figure 3 – AP x-ray of the forearms shows thinning of the diaphysis and widening of the metaphysis of the ulna and radius with decreased density of these bones.

Because most patients are blind before the age of 25, a bone examination should be performed on infants who present with eye abnormalities, such as, but not limited to, microphthalmia, microcornea, corneal clouding or vitreoretinal detachment to rule out this syndrome. Similarly, infants who present with severe bone abnormalities should undergo eye examination.[2]

On plain radiographs, patients usually manifest severe thinning of bones, bowing of extremities, and spinal deformities, such as fish mouth vertebra. One should be aware that radiological characteristics of the patients may present marked differences in the severity of the clinical phenotype and the degree of bone deformity.[3]

The final diagnosis is made via genetic testing.

Treatment options are regular bisphosphonates.

The main differential diagnosis for this disease was Osteogenesis imperfecta which was ruled out, as this patient had no hearing loss, blue sclera, joint laxity, contractures or teeth problems.

Radiologists should be aware of OPPG as a diagnostic possibility when facing osteoporosis in infants with eye abnormalities.

Ethical Disclosures / Divulgações Éticas

Conflicts of interest: The authors have no conflicts of interest to declare.

Conflitos de interesse: Os autores declaram não possuir conflitos de interesse.

Financing Support: This work has not received any contribution, grant or scholarship.

Suporte financeiro: O presente trabalho não foi suportado por nenhum subsídio ou bolsa.

Confidentiality of data: The authors declare that they have followed the protocols of their work center on the publication of data from patients.

Confidencialidade dos dados: Os autores declaram ter seguido os protocolos do seu centro de trabalho acerca da publicação dos dados de doentes.

Protection of human and animal subjects: The authors declare that the procedures followed were in accordance with the regulations of the relevant clinical research ethics committee and with those of the Code of Ethics of the World Medical Association (Declaration of Helsinki).

Protecção de pessoas e animais: Os autores declaram que os procedimentos seguidos estavam de acordo com os regulamentos estabelecidos pelos responsáveis da Comissão de Investigação Clínica e Ética e de acordo com a Declaração de Helsínquia da Associação Médica Mundial.

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