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Images of Interest / Imagens de Interesse

Radiographic Skeletal Features of Mucopolysaccharidosis Type VI

Manifestações Esqueléticas Radiográficas da Mucopolissacaridose Tipo VI

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Abstract

We present a case of a patient in his twenties, diagnosed with mucopolysaccharidosis type VI at 7 years old, and since then under enzyme replacement therapy. He has a clinical history of short stature, hypogonadism, moderate mitral and aortic valve insufficiency, bilateral hip dysplasia, bilateral carpal tunnel syndrome and bilateral corneal clouding. A skeletal radiographic evaluation showed typical radiological features of this disease, generally referred to as dysostosis multiplex.
Maroteaux-Lamy syndrome is a rare genetic disease, associated with several clinical manifestations, affecting virtually all organic systems. The most distinctive features of this syndrome are skeletal, with characteristic imaging findings on skeletal radiography, which we demonstrate in this article.

Keywords

Radiography; Bone; Mucopolysaccharidosis; Maroteaux-Lamy syndrome.

Resumo

Apresentamos um caso de um jovem adulto, diagnosticado com Mucopolissacaridose tipo VI aos 7 anos de idade, tendo iniciado terapêutica enzimática de substituição no momento do diagnóstico. Antecedentes pessoais de baixa estatura, hipogonadismo, insuficiência aórtica e mitral moderadas, displasia da anca bilateral, síndrome do túnel cárpico bilateral e opacificação da córnea bilateral. A avaliação radiográfica do esqueleto mostrou alterações típicas desta patologia, comumente designadas como disostose multiplex.
O Síndrome de Maroteaux-Lamy é uma doença genética rara, que se associa a diversas manifestações clínicas, com atingimento de praticamente todos os sistemas orgânicos. As alterações mais distintivas desta patologia são esqueléticas, com achados imagiológicos característicos na radiografia do esqueleto, que ilustramos neste artigo.

Palavras-chave

Radiografia; Osso; Mucopolissacaridose; Síndrome Maroteaux-Lamy.

Images in Case

We present a male patient, in his twenties, with established diagnosis of Mucopolysaccharidosis type VI by molecular genetic analysis with detection of a homozygotic mutation in arylsulfatase B (ARSB) gene, elevated levels of urinary glycosaminoglycan and demonstrated reduced arylsulfatase B (ASB) enzyme activity in isolated leukocytes and fibroblasts. He has been on enzyme replacement therapy with galsulfase ever since he was diagnosed at the age of 7. As main clinical features he has short stature and hypogonadism, treated with testosterone replacement therapy. He also suffers from moderate mitral and aortic valve insufficiency, controlled with a combination of furosemide, spironolactone and lisinopril. No other symptoms are reported. Regarding his surgical history, he was submitted to bilateral pelvic and femoral osteotomy with 18 months due to hip dysplasia, bilateral carpal tunnel surgical release to treat bilateral carpal tunnel syndrome, as well as bilateral corneal transplantation surgery as he had severe corneal clouding.
A skeletal radiographic evaluation was requested to monitor disease progression, in which typical radiological features were found (displayed on Figures 1, 2 and 3).

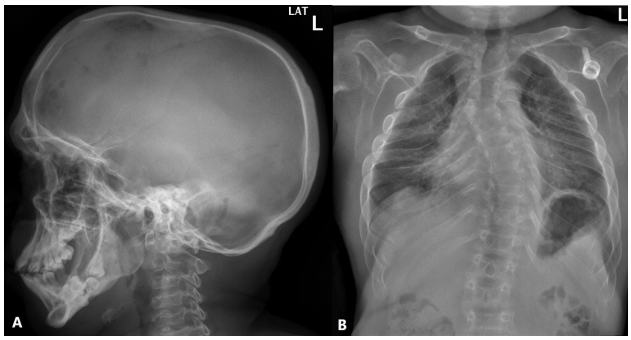

Figure 1 – Lateral skull radiograph (A) with macrocephaly, with widening of both transversal and antero-posterior diameters, "J-shaped" sella turcica, discrete thickening of cortical bone, prognathism and widely spaced teeth; Frontal chest radiograph (B) showing short and thickened clavicles, "paddle-shaped" ribs with anterior enlargement and posterior funnelling.

Discussion

Mucopolysaccharidosis type VI, also known as Maroteaux-Lamy syndrome, is a multisystem disease, caused by a mutation in the ARSB gene, leading to an autosomal recessive lysosomal storage disorder due to deficiency of ASB and associated accumulation of glycosaminoglycans, specifically dermatan-sulphate. Incidence rate per total live births in Portugal is estimated in 1 in 238,095, which illustrates the slight

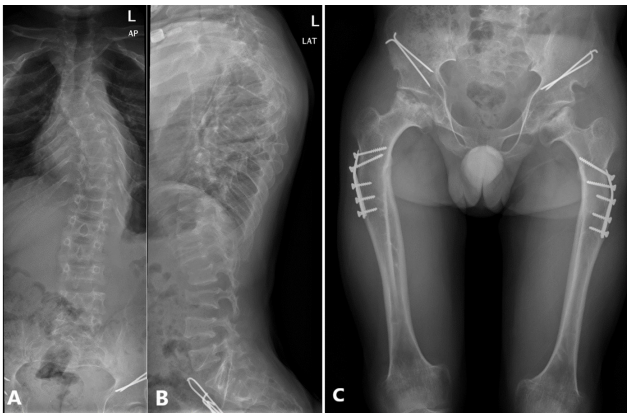

Figure 2 – Anteroposterior (A) and lateral (B) radiograph of the dorso-lumbar spine exhibiting "wedge-shaped" vertebral bodies, elongated vertebral pedicles, severe dextro-convex dorso-lumbar scoliosis producing dorsal gibbus deformity, straightening of the physiological lumbar lordosis; Anteroposterior hip and femoral radiograph (C) displaying rounded hypoplastic iliac wings, flat acetabular roof, with signs of surgical material related to hip dysplasia corrective surgery as well as long and narrow femoral neck, dysplastic femoral head, broad proximal femoral diaphysis.

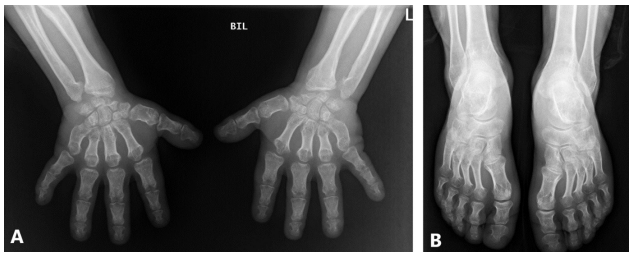

Figure 3 – Hands radiograph (A) with "V-shaped" hypoplastic distal ulna, shortening of carpal bones, as well as "bullet-shaped" phalanges; Feet radiograph (B) showing shortening of tarsal bones and phalanges.

relative increase in Mucopolysaccharidosis type VI frequency comparing to other types of mucopolysaccharidosis in Portuguese (16%) and Brazilian (18,5%) population. This can be partially explained by an uprisen prevalence of specific mutations, such as 1533del23, present in 23% of alleles amidst Brazilian Mucopolysaccharidosis type VI patients, which is also found in Portuguese patients, with unknown frequency. However, no specific ethnic group has been associated with a heightened risk of this syndrome.[1]

Maroteaux-Lamy syndrome can have numerous different clinical findings, with highly variable genetic findings, as well as phenotypic expression.[1]

Being a multisystem disease it has many clinical manifestations such as organomegaly, with hypertrophic cardiomyopathy and hepatosplenomegaly, cardiac valve dysfunction, respiratory symptoms, as these patients may suffer from reduced pulmonary function and sleep apnoea, ophthalmologic impairment, with corneal clouding and photophobia, as well as hearing disorders, with hearing loss due to repeated otitis media, sinusitis and also other symptoms such as inguinal or umbilical hernia and carpal tunnel syndrome.[1,2]

Maroteaux-Lamy syndrome exhibits characteristic skeletal chondrodystrophic abnormalities generally referred to as dysostosis multiplex, which are not exclusive to this disease, and can be found in other types of mucopolysaccharidosis, mucolipidoses and other storage diseases.[1,2] In this case our patient showed classic radiological features of Mucopolysaccharidosis type VI on skeletal radiographic evaluation.

Ethical Disclosures / Divulgações Éticas

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

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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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