MORQUIO DISEASE IN TWO SISTER. CASE REPORT

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BACKGROUND

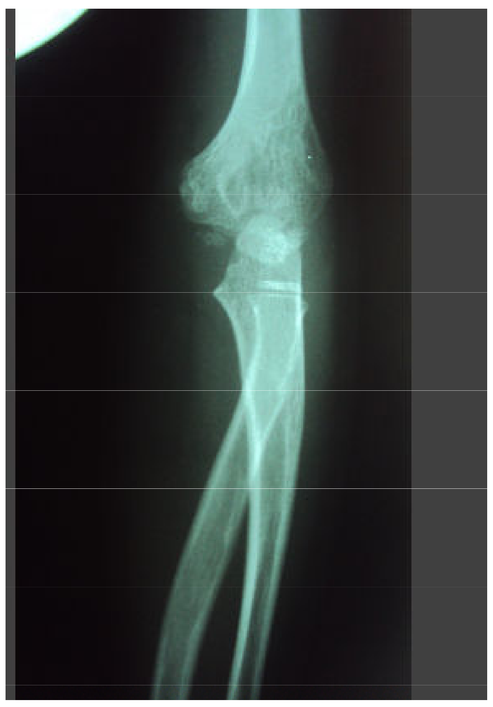
Morquio disease was described by the Uruguayan pediatrician Luis Morquio.
It is a congenital disease caused by a deficiency of the enzyme N ACETILGALACTOSAMINE 6 SULFATASE (MPS IV A) or B GALACTOSIDASE (MPS IV B) and it frequency is 1/100.000 live births. Accumulation of mucopolysacharides in tissues results in short stature, skeletal anomalies (vertebral column deformities), loss of hearing, visual anomalies (corneal opacities), cardiac, hepatic and respiratory problems life expectancy is 40 years

OBJECTIVE

To describe two patients with Morquio´s disease type IVA.

CLINICAL DESCRIPTION

joint thickening and genu valgus and genu valgus.

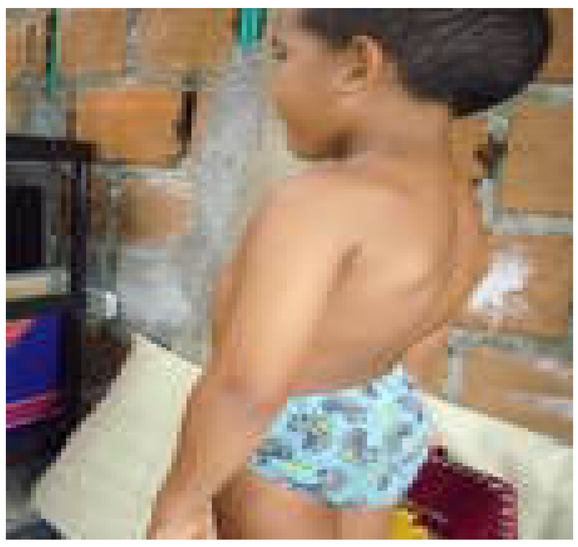


PRESENTING PROBLEMS

Description of physical findings, radiologic and laboratory finding in patients with short stature and Morquio disease.
Two sister with 18 month of age and 4 years of age, consulting for short stature (>3SD for age) deformities started age 18 months, dolichocephalia, serrated teeth, thoracic kyphosis, pectum carinatum, globular abdomen, joint thickening and genu valgus and no corneal opacities. Also had affection of thoracic spinal cord and atlanto-occipital subluxation and back fixing atlanto occipital. Enzymatic activity of GA lactose 6 sulfate was in leucocytes decreased (Morquio type IVA).

CLINICAL MANAGEMENT

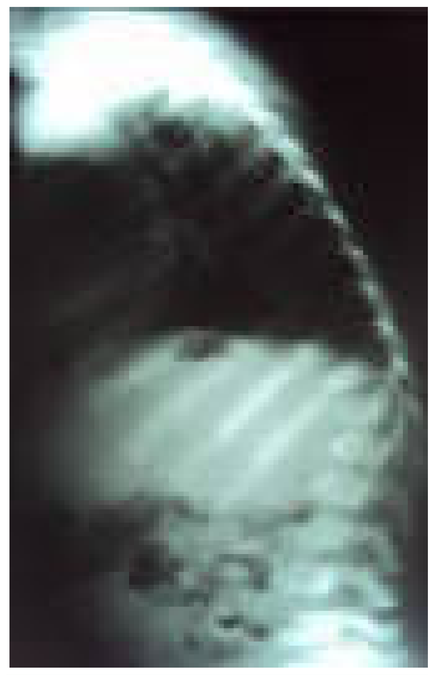
Morquio type IV A should be suspected in two sister patients with short stature and skeletal deformities are being studied. The patient must receive a multidisciplinary management: Cardiology Orthopaedic Ophthalmology Endocrinology Pediatrics, Genetics

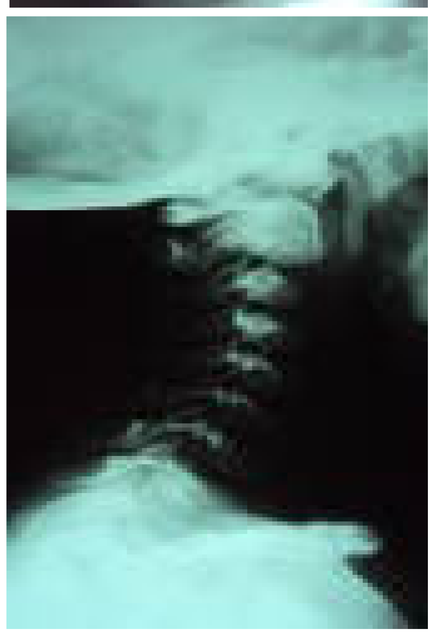


Kyphosis

RESULTS

With spinal cord compromise thoracic and subluxation atlanto occipital and fixing atlanto occipital.





DISCUSION

The management multidisciplinary in this patients is very important.

Enzyme replacement therapy (ERT) and hematopoyetic stem cell therapy (HSCT) have emerged as a treatment for mucopolysacharidoses disorders, including Morquio A disease.
There is promising results with new enzymatic replacement therapy.

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