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Mucopolysaccharidosis type I Hurler-Scheie syndrome affecting two sisters

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Mucopolysaccharidosis I (MPS I) is a rare inherited disorder characterized by physical deformities and developmental anomalies. Part of a group of clinically progressive disorders, it is caused by the deficiency of the lysosomal enzyme, α-L -iduronidase, which results in intralysosomal accumulation of dermatan sulfate and heparan sulfate and in turn causes cell dysfunction. Two sisters, one 11 years old and the other 7, both MPS type I H/S, came to our diagnostic center. Hand-wrist radiographs revealed bullet-shaped phalanges with proximal pointing of the second to fifth metacarpals. Ultrasonographic examination showed splenomegaly in the younger child. Radiography of the pelvis showed a narrow pelvis with flared iliac wings. A skull skiagram showed J-shaped sella.

Introduction

Mucopolysaccharidosis type I (MPS I) is a progressive multisystem disorder with features ranging over a continuum from mild to severe. Clinical features usually noted within the first two years are hepatosplenomegaly, skeletal deformity, coarse facial features, corneal clouding, large tongue, prominent forehead, joint stiffness, and short stature. Progressive skeletal dysplasia (dysostosis multiplex) involving all bones occurs in all individuals with severe MPS I.

Case report

Two sisters, one 11 years old and the other 7, both MPS type I H/S, came to our diagnostic center for a skeletal survey and abdominal sonography. On clinical examination, both children were short-statured (stunted growth) and presented coarse facial features. The elder's height was 105 cm and weight 23 kg, and the younger's height was 78 and weight 18 kg. This was less than the 3rd percentile on the NCHS standard. Both sisters had large heads, short necks, depressed nasal bridges, large thick tongues, and joint contractures (Figs. 1A and 1B). Medical history revealed that both suffered frequent respiratory infections (chronic sinus infections), stiffness of joints, and hepatomegaly (the last more severe in the elder sister). Their family history was noncontributory. On radiological evaluation, hand-wrist radiographs revealed bullet-shaped phalanges with proximal pointing of the second to fifth metacarpals (Figs. 2A and 2B, in the older and younger one, respectively). Radiography of the spine and ribs showed anterior notching in the thoraco-lumbar vertebral bodies, with mild inferior beaking in the L2 vertebra and oar-shaped ribs (Fig. 3). Radiography of the pelvis revealed a narrow pelvis with flared iliac wings (Fig. 4).

Ultrasonographic examination showed splenomegaly in the younger child (Fig. 5). A skull skiagram showed J-shaped sella (Fig. 6). Echocardiography showed mitral-valve thickening and mild mitral regurgitation as well (Fig. 7).

Later blood and urine examination of both patients showed deficient enzyme activity of MPS-1 (α-L-iduronidase). The values were 1.8 nmol/hr/ng in the elder, and 0.5 nmol/hr/mg in the younger child. Urinalysis revealed increased traces of iduronite sulphate and dermatan sulphate. The younger sister has an IQ of 50 (moderate mental retardation), and the elder has IQ of 70 (mild mental retardation). Other hematological investigations were within normal limits.

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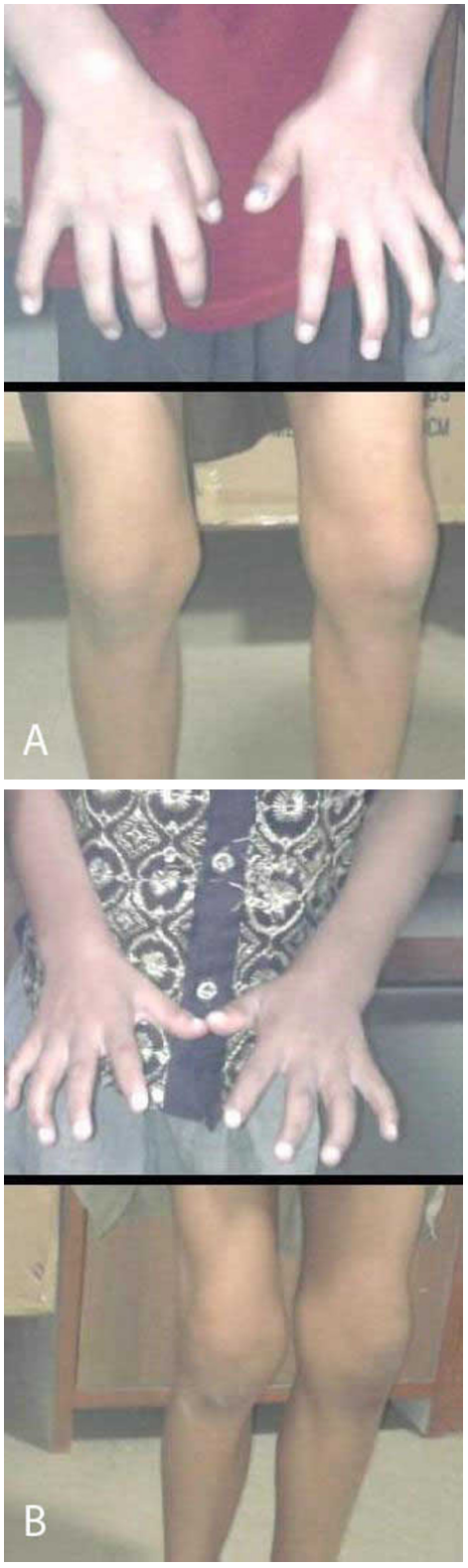

Figure 1. 7- and 11-year-old sisters with mucopolysaccharidosis I, Hurler-Scheie and Scheie syndrome. Picture of hands and knees of both children, showing contracture of joints.

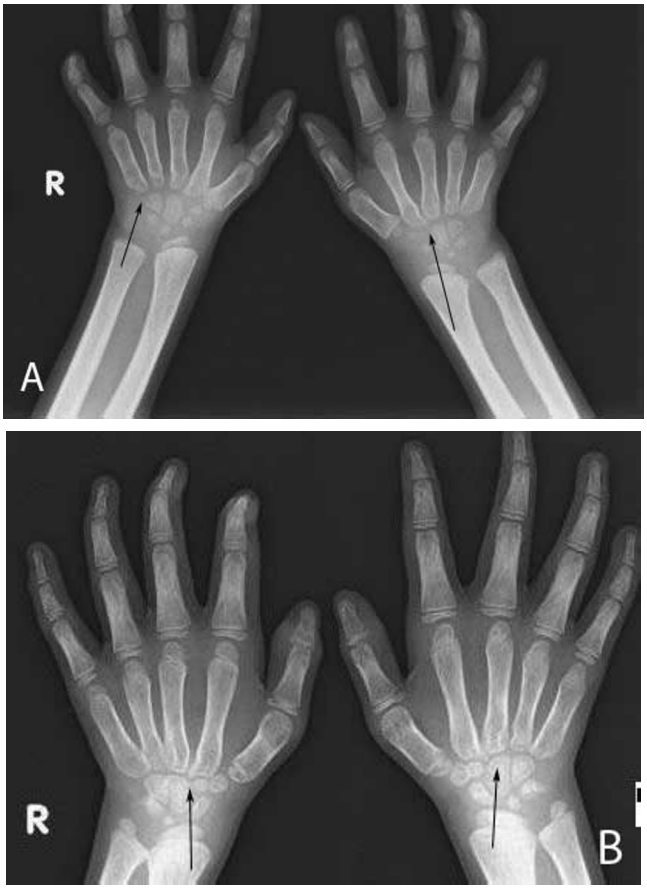

Figure 2. 7- and 11-year-old sisters with mucopolysaccharidosis I, Hurler-Scheie and Scheie syndrome. Hand-wrist skiagram. Arrows show bullet-shaped phalanges with proximal pointing of the second to fifth metacarpals. A. Older sister. B. Younger sister.

Discussion

Mucopolysaccharidosis-I (MPS I) is a lysosomal storage disorder inherited as an autosomal-recessive condition and is caused by a deficiency of the lysosomal enzyme α1 -iduronidase. This results in the progressive accumulation of glycosaminoglycans (GAG) within the lysosomes, leading to multiorgan dysfunction and damage (1). Patients affected with MPS I are unable to degrade the GAG, dermatan sulfate, and heparan sulfate, which provide structural support to the extracellular matrix and cartilaginous structures such as joints and heart valves (2).

MPS I has an estimated incidence of 1 case per 100,000 live births, and the attenuated type represents about 20% of the total MPS I population. MPS I includes separate diseases on the basis of clinical presentation: Hurler Syndrome (severe), Hurler-Scheie syndrome (intermediate), and Scheie syndrome (mild) (3). However, since MPS I has been recognized as a disease continuum due to variation in age of onset and rate of disease progression, an international panel composed of 12 experts on MPS I revised and