Mucopolysaccharidosis type I Hurler-Scheie syndrome: A rare case report

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Abstract

Mucopolysaccharidosis I (MPS I) is a rare inherited disorder that belongs to a group of clinically progressive disorders and is caused by the deficiency of the lysosomal enzyme, \( \alpha_1 \)-iduronidase. MPS I has been recently classified into a severe (Hurler syndrome) and an attenuated type (Hurler-Scheie and Scheie syndromes). The purpose of this article was to describe a rare case of MPS type I, attenuated type (Hurler-Scheie) affecting a 15-year-old Indian child.

Keywords: Autosomal recessive, iduronidase, mucopolysaccharidosis

Introduction

Mucopolysaccharidosis-I (MPS I) is a lysosomal storage disorder inherited as an autosomal-recessive condition and is caused due to deficiency of the lysosomal enzyme \( \alpha_1 \)-iduronidase, which results in the progressive accumulation of glycosaminoglycans (GAG) within the lysosomes, subsequently leading to multiorgan dysfunction and damage. 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.[1]

MPS I has an estimated incidence of 1 case per 100,000 live births,[2,3] and the attenuated type represented about 20% of the total MPS I population.[4] MPS I includes separate diseases on the basis of clinical presentation, Hurler Syndrome (severe), Hurler-Scheie syndrome (intermediate) and Scheie syndrome (mild). However, with MPS I being recognized as a disease continuum due to variation in age of onset and rate of disease progression, an international panel comprising of 12 (International) experts on MPS I revised and updated the initial guidelines in the year 2008 on the basis of additional clinical data and therapeutic advances. Based on their recommendations, MPS I has been classified into two broader groups, severe MPS I (Hurler Syndrome) and attenuated MPS I (Hurler-Scheie and Scheie syndromes).[1]

The purpose of this article is to describe a rare case of a 15-year-old female child affected with MPS I, Hurler-Scheie (H/S).

Case Report

A 15-year-old Indian female with a known diagnosis of MPS type I H/S was referred to our dental clinic by a pediatrician for dental care. On physical examination, she appeared short statured (stunted growth) with lumbar kyphosis and presented coarse facial features [Figure 1]. Medical history revealed that she suffered frequent respiratory infections (chronic sinus infections), stiffness of joints and hepatomegaly. Gradual hearing loss and vision impairment has been reported with evidence of corneal clouding [Figure 2]. Her family history was non-contributory.

Intraoral examination revealed macroglossia, high-arched palate, anterior open bite and decayed 11, 12, 21, 16, 26, 36 and 46 (FDI notation). Oral hygiene of the patient was good with no evidence of significant deposits of calculus [Figure 3].

Cardiological evaluation including echocardiogram revealed mild mitral regurgitation with good left ventricular function. Anteroposterior view of the chest showed widened metaphyses and oar-shaped ribs, which were suggestive of dysostosis multiplex [Figure 4]. Hand–wrist radiographs revealed bullet-shaped phalanges with proximal pointing of the second to fifth metacarpals [Figure 5]. Urine analysis revealed increased traces of iduronite sulfate and dermatan sulfate.

Dental management needed multiple visits and included restoration of 16, 26, 36 and 46 with silver amalgam and 11, 12 and 21 with composite restoration. Further, importance of home oral care and follow-up visits was emphasized to the

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parents. The patient is currently under enzyme replacement therapy and regular medical evaluation involving the multidisciplinary team.

Discussion

MPS I, caused by deficiency of the lysosomal enzyme, alfa-L-iduronidase, has been mapped to the chromosome band 4p16.3. Children with severe MPS I usually die within the first decade of life as a result of cardiorespiratory failure and progressive neurological disease. On the contrary, most patients with attenuated MPS I survive into adulthood. However, cases of attenuated MPS I show wide variation with respect to age of presentation, symptoms and disease course.

The clinical presentation may be limited to growth failure, cloudy corneas, mild coarsening of facial features, hepatomegaly and micrognathism in MPS I H/S. Skeletal changes of hands and spine are minimal. The typical features appear at about the age of 12 years, including macrocephaly and coarse facies, and restriction of joint mobility with deformities becomes apparent at the age of 12–15 years. The combination of metaphyseal deformities and thickened joint capsules secondary to GAG deposition and fibrosis has been related to joint function abnormalities. Frequent upper and lower respiratory tract infections are common and occur secondary to enlargement of tonsils and adenoids and enlarged tongue.

Patients with MPS I H/S may show dental abnormalities...
including enamel defects, carious teeth, dentigerous cysts and abscesses. However, our patient showed caries involving the permanent first molars in all the four quadrants and maxillary incisors, high-arched palate and open bite due to macroglossia and mandibular prognathism. Regular dental and radiological evaluation should be performed every 6 months and parents or caregivers should be educated about dental home care.[1] Our patient presented good oral hygiene, which can be attributed to the supervision and involvement of parents in oral care and normal cognitive development of the patient.

Cardiac abnormalities are common among patients with MPS I, and worsen with age. It has been recommended to undergo cardiac evaluation every 1 or 2 years after an initial diagnosis.[1] Monitoring of patients with MPS I H/S by a multidisciplinary team should include regular assessments, supportive care and treatment of a variety of systemic complications. Delayed diagnosis in patients with attenuated MPS I may be attributed to lack of disease awareness among parents and physicians.[8,9]

With the advent of hematopoietic stem cell transplantation and, more recently, enzyme replacement therapy, there exists a need for early diagnosis, better disease recognition and management.[3] Early detection of the disease and appropriate management through a multidisciplinary approach is recommended to improve the quality of life.

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