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Case Reports

Mucopolysaccharidosis Type 1(Hurler Syndrome)- About A Case

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Abstract

Introduction: Hurler syndrome is an autosomal recessive inborn error of metabolism, resulting from deficiency of a-liduronidase, a catabolic mucopolysaccharidase, which leads to excessive systemic storage of the glycosaminoglycans (GAGs) heparan sulphate and dermatan sulphate [1]. Affected children may appear normal at birth. However, progression of the disease results in characteristic facial changes, hepatosplenomegaly cardiomyopathy, major skeletal abnormalities and CNS damage, resulting in severe disability, intellectual regression and death, usually by the age of 10 years. We report the case of a 7-year-old girl who was consulting for a visual impairment with photophobia. The ophthalmologic examination showed corneal opacities associated with bilateral papilled edema with an eye tone of 18mmHG for the eye right and 19 mmHG for the left eye. The abdominal examination finds a protruding abdomen with a hepato splenomegaly. Cardiovascular examination with doppler echo objective thickening mitro-aortic and tricuspid valves. The diagnosis was evoked before facial dysmorphism and then confirmed biologically (MPStype I-H). A bilateral trabeculectomy was performed while waiting for a marrow allograft. The early diagnosis of MPS, before the formation of neurological deficits, has become essential, since the treatment can stop the evolution. Thus, a better knowledge of the clinical picture by ophthalmologists could improve the prognosis.

Keywords: Mucopolysaccharidosis type1, eyes, diagnosis.

INTRODUCTION

Hurler syndrome is an autosomal recessive inborn error of metabolism, resulting from deficiency of a-liduronidase, a catabolic mucopolysaccharidase, which leads to excessive systemic storage of the glycosaminoglycans (GAGs) heparan sulphate and dermatan sulphate [1].

Affected children may appear normal at birth. However, progression of the disease results in characteristic facial changes, hepatosplenomegaly cardiomyopathy, major skeletal abnormalities and CNS damage, resulting in severe disability, intellectual regression and death, usually by the age of 10 years.

Three significant and progressive ocular manifestations occur (corneal clouding, retinal pigmentary degeneration and optic atrophy) with varying severity in this syndrome [1, 3].

In some patients, trabecular involvement will result in the development of secondary open-angle glaucoma.

CASE REPORT

We report the case of a 7-year-old girl who was consulting for a visual impairment with photophobia. The ophthalmologic examination showed corneal opacities associated with bilateral papilled edema with an eye tone of 18mmHG for the eye right and 19 mmHG for the left eye.

The abdominal examination finds a protruding abdomen with a hepato splenomegaly.

Cardiovascular examination with doppler echo objective thickening mitro-aortic and tricuspid valves.

The diagnosis was evoked before facial dysmorphism and then confirmed biologically (MPStype I-H). A bilateral trabeculectomy was performed while waiting for a marrow allograft.

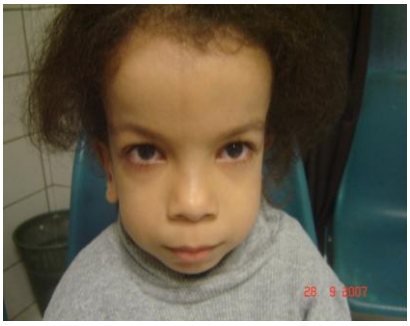

Fig-1: Facial dysmorphism

Fig-2: Corneal opacities

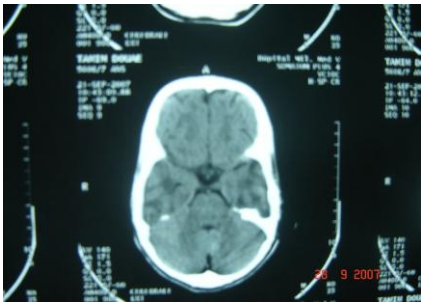

Fig-3: MRI image in sagittal section seq T1 Iso-intense, discretely heterogeneous process repressing the frontal lobe G at the top deformation of the cranial box

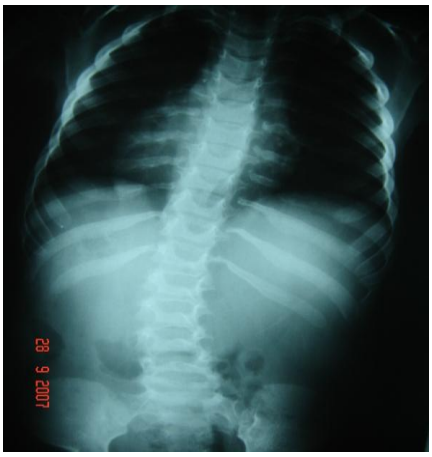

Fig-4: Bone deformities

DISCUSSION

The accumulation of glycosaminoglycans, present in various forms in ocular tissues, may result in stromal opacities, secondary glaucoma, pigmentary retinopathy, and / or papillary edema. While ophthalmological involvement is often secondary, it can lead the ophthalmologist to the diagnosis of MPS.

The Ocular Features of the Mucopolysaccharidoses
Cornea and anterior segment

Glycosaminoglycan deposition in the corneal stroma has been suggested by some authors to cause a progressive increase in corneal thickness.

However, in an animal model of MPS VI, the cornea was found to be thinned. Deposition in the anterior chamber structures and iris results in narrowing of the angle and poor pupillary dilation, leading to increased risk of pupil block glaucoma. Secondary alterations in corneal collagen structure and organization also occur.

Peripheral vascularization of the cornea may occur as a result of chronic corneal edema due to raised intraocular pressure or secondary to corneal exposure associated with exophthalmos [3].

Multiple iridociliary cysts have been reported in two patients with MPS IS and MPS VI.

B. Glaucoma

Glaucoma is thought to occur in MPS due to accumulation of GAG within anterior segment structures, resulting in narrowing of the angle, and due to GAG accumulation within trabecular cells leading to obstruction of trabecular outflow.

C. Lens

Cataracts may occur but are usually clinically insignificant. Subcortical lens opacities have been reported in three siblings with MPS IV (Morquio) syndrome, and bilateral peripheral cataracts in MPS IIID (Sanfilippo) [3].

D. Sclera

The sclera of individuals with MPS is thickened due to deposition of GAG. Bilateral uveal effusions have been reported in a patient with Hunter syndrome. This was presumed to be secondary to scleral thickening and reduced number of vortex veins. Thickening of sclera may also contribute to optic nerve swelling.

E. Optic nerve

Optic nerve head involvement is common in MPS IH and MPS IH/S: a review of 108 MPS patients by Collins found that optic nerve swelling occurred in 57% of patients with MPS IH (Hurler) and 43% of patients with MPS IH/S (Hurler/Sheie).

F. Retina

Pigmentary retinopathy has an insidious onset and is often masked by corneal problems. Patients may complain of night blindness and problems with peripheral vision. Clinical signs of retinopathy include arteriolar narrowing, indistinct foveal reflex and pigmentation, retinal pigment epithelium (RPE) atrophy [3], and later bone-spicules, optic disk pallor, and associated visual field constriction.

G. Ocular motility

Motility disorders are common in MPS patients, especially exotropia. Esotropia may reflect raised intracranial pressure. Acquired Brown syndrome, a mechanical limitation of movement of the superior oblique tendon resulting in limitation of elevation in adduction.

There are not many avaliable treatment options for late Hurler syndrome. But during recent technological advancement, there are some options.

The treatment technique depends on the condition of the individual and the affected organs. Marrow transplantation can improve the symptoms and was successfully performed first time in the year 1981. The bone marrow transplant should be performed to begin very much to prevent mental retardation. Marrow transplantation is performed after whole body radiotherapy, and is highly effective treatment of Hurler syndrome, except for bone and eye disease [2].

A new technique, umbilical cord is in use for transplants. In this technique, stem cells provided by the donor are transplanted and are as effective as bone marrow transplantation and further this technique does not require total radiation before grafting [4]. The third enzyme replacement technique is also in place. Children with severe Hurler syndrome have progressive mental retardation and a short duration is expected.

CONCLUSION

The early diagnosis of MPS, before the formation of neurological deficits, has become essential, since the treatment can stop the evolution. Thus, a better knowledge of the clinical picture by ophthalmologists could improve the prognosis.

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