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Hunter Disease: When We Suspected?

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Abstract

Hunter disease, also known as mucopolysaccharidosis type 2 (MPS 2), is a lysosomal storage disorders (LSD) caused by a deficiency of iduronate-2-sulfatase. It is a rare x-linked disorder.

The patient will appear with Hurler-like phenotype with coarse face, hepatosplenomegaly, cardiovascular disorders, dysostosis multiplex, dwarfism, deafness, but no corneal clouding like in Hurler disorder.

These patients can be treated with enzyme replace therapy (ERT), Elaprase, 0.5 mg/kg IV once a week.

Genetic counselling and prenatal diagnosis should be offered for the couple because the risk of recurrence is 50% in the next pregnancy.

Keywords: *Hunter; Diagnosis; Enzyme Replace Therapy*

Hunter disease, also known as mucopolysaccharidosis type 2 (MPS 2), is a lysosomal storage disorders (LSD) caused by a deficiency of iduronate-2-sulfatase. The glycosaminoglycans (GAGS) in urine showed heparan and dermatan sulfate.

It is a rare x-linked disorder, with incidence around 1 out of 100000 males. Females are carriers.

The disorder involves several organs, causing aggressive behavior and developmental delay.

The physical findings are similar to MPS 1. There are two forms of presentation: a severe and a mild type. In this last form, the intelligence can be normal or only presented with mild delay. The patient will appear with Hurler-like phenotype with coarse face, hepatosplenomegaly, cardiovascular disorders, dysostosis multiplex, dwarfism, deafness, but no corneal clouding like in Hurler disorder.

The age of presentation is 1 - 3 years in the severe form and 1 - 5 years in the mild one.

The radiologic changes in this type 2 disorder are dysostosis multiplex, with short and thick long bones. The clavicle has widened ends.

We can also find flattened acetabulum, ovoid vertebrae, dorsal kyphosis and ribs with “oar shape”.

The skull is large and deformed due to craniosynostosis, with a thickened calvarium and an abnormal "j" or boot-shaped sella turcica.

The basilar skull may cause cervical vertebrae fusion causing narrowed spinal channel.

The diagnose of Hunter disorder is based in urine test for GAGS, blood tests for enzyme activity and gene test for ILS.

These patients can be treated with enzyme replace therapy (ERT), Elaprase, 0.5 mg/kg IV once a week with infusion of 1 to 3 hours each time. The most common adverse effects are pyrexia and headache.

Even with the ERT, when the patient present with severe form, many complications can appear, particularly breathing problems, heart disease, joint abnormalities, carpal tunnel syndrome and behavioral changes.

Physical therapy is important to improve the quality of life of the affected child.

Genetic counselling and prenatal diagnosis should be offered for the couple because the risk of recurrence is 50% in the next pregnancy.

Conclusion

Hunter disorder is underdiagnosed in the first year of life but after this period we can diagnose the disorder based on the clinical and radiological findings. The earlier the diagnosis the better the prognosis after starting the treatment with enzyme replace therapy, although in the severe form, even with the treatment can cause several bones and heart problems [1-3].

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