Mucopolysaccharidosis Type 1

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Mucopolysaccharidosis(MPS) are a group of lysosomal storage disorders caused by deficiency of alfa-L-idorinidase. MPS is a progressive multisystem tissue and organ dysfunction, including cardiac, respiratory, musculoskeletal and visceral manifestations.

MPS 1 divided into three clinical phenotype: Hurler, Hurler–Scheie, Scheie syndromes.

Infants with severe disease (Hurler syndrome) are usually diagnosed in the first year of life. Upper airway obstruction and frequent ear, nose and throat infections dominate the clininical picture at early stage and the full clinical picture of short stature, hepatosplenomegaly, coarse face, cardiac involvement and learning difficulty and manifestation evolves over the 2nd, 3rd years of life.

Skeletal involvement (dysostosis multiplex) include enlarged sella turcica, broad ribs, tapering of metacarpals and hooke shape vertebrae.

Diagnostic tests

The diagnosis of MPS1 is based on clinical suspicion supported by appropriate clinical and radiological examinations followed by urinary examination for glycosaminoglican excretion and then specific enzyme assay.

Treatment

Treatment includes haematopoietic stem cell transplantation (HSCT) and enzyme replacement therapy (ERT).

ERT (aldurazyme) using recombinant enzymes are approved for patients with MPS I. It reduces the number of episodes of sleep apnea, ameliorates rate of growth and joint mobility, and reduces organomegaly and urinary GAG excretion. ERT is a safer treatment for these disorders, but limited by an inability of the enzyme to cross the blood – brain barrier when given intravenous

Bone marrow transplantation from related or unrelated donors or cord blood transplantation results in significant clinical improvement of somatic disease in MPS I. Clinical effects include increased life expectancy, resolution or improvement of growth failure, hepatosplenomegaly, joint stiffness, facial appearance, obstructive sleep apnea, heart disease, communicating hydrocephalus, and hearing loss. Enzyme activity in serum and urinary GAG excretion normalize.

Keywords: Mucopolysaccharidosis 1; Child; Diagnosis; Therapy

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