

Mucopolysaccharidosis Type VI: Five Years Treatment with Galsulfase

Jorge Sales Marques*

Pediatric department, Centro Hospitalar Vila Nova de Gaia, Portugal

***Corresponding Author:** Jorge Sales Marques, Pediatric department, Centro Hospitalar Vila Nova de Gaia, Portugal.

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Introduction

Mucopolysaccharidosis type VI, (MPS VI), also known as Maroteaux-Lamy syndrome, is an autosomal recessive inherited lysosomal disorder. The deficient enzyme is Arylsulfatase B. This result in multisystem accumulation of dermatan sulfate, one of the glycoaminoglycans (GAGS). The incidence is estimated to 1/340 000 live births. The child presented with coarse face, macrocephaly, corneal clouding, glaucoma, impaired hearing, enlarged tongue, sleep apnea, obstructive and restrictive airway disease, cardiomyopathy, cardiac arrhythmia, pulmonary hypertension, hepatosplenomegaly, umbilical and inguinal hernias, hip dysplasia, joint stiffness, spinal cord compression and carpal tunnel syndrome. The intelligence is normal. Galsulfase is the first replacement therapy for MPS VI. The dose is 1 mg/kg/day in four-hour infusion associated with 250 ml of 0,9% sodium chloride, every week. The treatment with Galsulfase, improved more endurance when the patient walks for 12 minutes, stairs climb for 3 minutes and also reduced urinary GAGS in clinical studies.

Purpose

Evaluate a thirteen years old boy with MPS VI after five-year treatment with galsulfase, diagnosed at 15 month of age with hypotonia, umbilical hernia, hip dysplasia. The Arylsulfatase B showed: 0 nmol/h/mg protein in the leucocytes (N: 109-522), 0 nm/h/mg in the fibroblasts (N: 122-664) and urine Gags 43 mg/nmol creatinine (N: 4-11).

He started treatment with galsulfase at eight years of age. The items that we choose for the comparative study were: weigh, height, growth velocity, body mass index, heart (ECG and ultrasound), lungs (respiratory function), vision and eye fundi, endurance test (12 minutes walk and 3 minutes' stairs climb) and Gags in urine.

Results

We didn't found any improvement in height, weight, growth velocity, vision, eyes fundi, ECG, psychomotor development after 5 years of treatment. The heart ultrasound, recovered from thick mitral and aortic valve, mitral insufficiency, dilated left auricle, systolic pressure of pulmonary artery – 30 mm, to small mitral and aortic insufficiency, normal structure of the heart and systolic pressure of pulmonary artery – 40 mm. The respiratory function improved from FEV₁/FEV- 85% to FEV₁/FVC- 97%. The 12-walk test, stairs climb and Gags in urine showed important improvement after therapy with galsulfase (Figures 1,2,3,4).

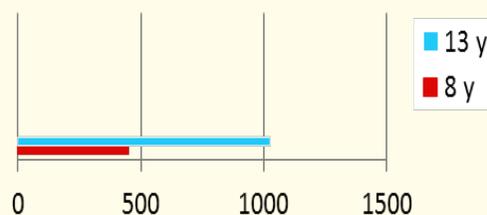


Figure 1: 12 m walk – meters.

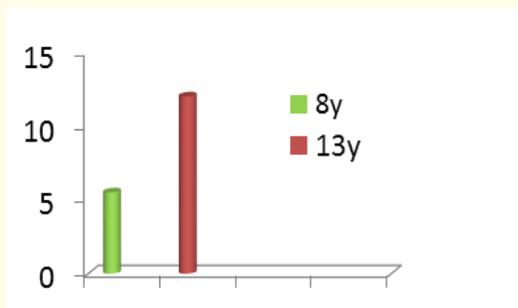


Figure 2: 12 minutes walk test.

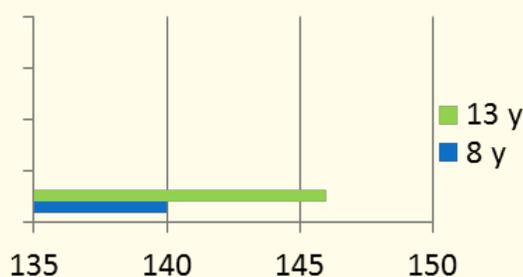


Figure 3: Stairs Climb.

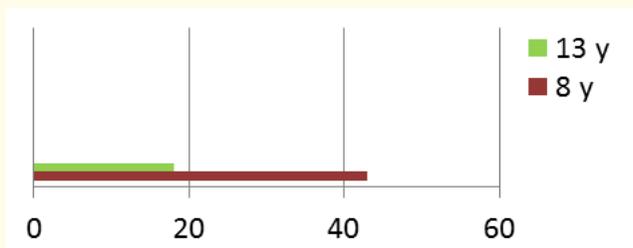


Figure 4: Gags (N: 4-11 nmol /creatinine).

Discussion

Our case confirmed that galsulfase improved endurance test and reduced urine Gags. The 12 minutes walk test showed an excellent result. The patient double the distance and completed the test. The 3 minutes' stairs climb has a much lower improvement, with increased of only six stairs after five years. Gags reduced significantly after the replace therapy with almost reaching normal range. Our study showed that standard deviation of growth, weight and growth velocity did not improved during this period. Heart and lung function improved well, particularly the FVC₁/FVC ratio that change to 97%, normal range for the age and sex. No evolution was found in the eyes that at the examination showed the same results after five year. The patient so far has no glaucoma and optic nerve affected. Galsulfase replacement therapy improved the patient endurance, respiratory function, heart structure and avoid so far more severe changes in the eyes.

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