Title

Improvements in young adult male receiving ERT for Pompe disease

Journal

Molecular Genetics and Metabolism, 102(3)

ISSN

1096-7192

Authors

Simon, Julie

Kimonis, Virginia

Publication Date

2011

DOI

10.1016/j.ymgme.2010.11.160

94) Improvements in young adult male receiving ERT for Pompe disease

Julie Simon, Virginia Kimonis

Department of Pediatrics, Section of Human Genetics, University of California Irvine, Irvine, CA, USA

Background: Pompe disease, first described in 1932, is a glycogen storage disorder as well as a lysosomal storage disorder that results in accumulation of glycogen in major organs, particularly the skeletal, cardiac, and smooth muscles, and is represented by a wide spectrum of symptoms with varying age of onset, severity, and rate of progression. The late-onset form, with symptoms presenting in childhood through adulthood, is characterized by proximal muscle weakness, respiratory insufficiency, and no cardiac involvement, which is the cardinal sign of classic or infantile-onset Pompe disease. It is caused by the deficiency of enzyme acid alpha-glucosidase (GAA) and inherited in an autosomal recessive manner.

Here we describe a 15-year-old male diagnosed with Pompe disease at 11 years of age due to elevated urine hexose tetrasaccharide and reduced GAA activity on dried blood spot analysis confirmed via fibroblast analysis in February 2007. He initially presented with fatigue and difficulty breathing secondary to weak diaphragm, scoliosis, and progressive muscle disease of unknown etiology two years prior to diagnosis. He initiated enzyme replacement therapy (ERT) of alglucosidase alfa in May 2007. The infusions are well tolerated for the past three years. One minor setback was experienced following posterior spinal fusion level T4 to sacrum in June 2009. His 6-minute walk test and pulmonary function tests both decreased dramatically, but since have continued an upward trend following recovery.

Results: Continued use of ERT results in observable improvements in 15-year-old male with Pompe disease. His data following ERT demonstrates an increase in pulmonary function (FVC measured pre-ERT 0.72 L to 1.66 L currently), an increase in distance covered in the 6-minute walk test (pre-ERT 1414 ft/6 min to 1830 ft/6 min currently), decrease in his BiPAP IPAP and EPAP settings from 28 to 15 cm H_2O and 18 to 6 cm H_2O respectively for pre-ERT and currently, an increase in strength by dynamometry. The increase in strength was noted particularly in our patient's grip strength increasing from 28 lbs pre-ERT to 65 lbs currently. Presently, his improvements following ERT will be paired with a concurrent submaximal aerobic exercise regime and increase in protein ingestion to further slow the progression of the disease.