Enzyme Therapy Fights The Rare And Often-Deadly Pompe Disease

Erasmus University

MyozymeTM is the first medicine produced for Pompe disease and the first ever treatment for an inherited muscle disorder. Pompe disease is a rare genetic disorder, which can affect infants, children, and adults; only about 5,000 to 10,000 people in the world suffer from this condition. Progressive muscle weakness is the most common symptom of both the infantile onset and the late-onset forms of this disease. It limits mobility and respiratory function.

No therapy was available for this devastating condition until 2006. Fundamental and applied clinical research at Erasmus University Medical Center in Rotterdam, The Netherlands, finally resulted in the first pharmaceutical product that is effective in treating Pompe disease. A defective gene in the body causes this disease because the gene cannot produce acid alpha-glucosidase, an enzyme that breaks down glycogen, a form of sugar.

> *This inability leads to accumulation of glycogen in the muscle, which is followed by loss of*

muscle function.

A.J.J. Reuser, Ph.D. and A.T. van der Ploeg, M.D., at Erasmus University Medical Center, initiated the development and manufacturing of recombinant human alpha-glucosidase in cultured cells and milk of transgenic animals. Genzyme, a Boston-based biotechnology company introduced the product MyozymeTM to the health care market in 2006 after receiving FDA and EMEA approval. The Erasmus University Medical Center, the Prinses Beatrix Fonds, patient associations and industrial partners provided funding for this work.

Clinical trials have shown that MyozymeTM improves the ventilator-free survival rate in patients with infantileonset Pompe disease and the condition of affected children and adults.

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