



Gene Therapy for Pompe Disease

Glycogen storage disorder type II or Pompe disease is a rare, incurable disease caused by a genetic defect resulting in deficiencies of the enzyme acid alpha-glucosidase (GAA). This deficiency of GAA activity results in excessive storage of glycogen in lysosomes, eventually disrupting the architecture and function of affected cells. Patients suffer from cardiomyopathy, muscle weakness, respiratory distress, and significantly shortened life-spans. There is no cure for Pompe disease, and treatment options are not very effective. University of Florida researchers have developed a novel plasmid to package high-titer rAAV1 vectors for use as a gene-therapy treatment for Pompe disease. This therapy has been shown to be effective in maintaining glycogen clearance in an animal model of Pompe disease.

Applications

Novel AAV plasmid useful for:

- ◆ Gene-replacement therapy for Pompe disease
- ◆ Creation of other AAV vectors for other monogenic disorders

Advantages

- ◆ Compared to AAV2 vectors, lower doses of this vector are needed, resulting in an overall decline in manufacturing and treatment costs
- ◆ Plasmid can be used to package large quantities of high-titer rAAV1 vectors, making this technology applicable to treat a wide variety of monogenic diseases
- ◆ Provides long lasting therapy for Pompe disease, for which there are currently no effective treatments

The Technology

This invention provides the potential for long-term gene replacement therapy for Pompe disease. University of Florida inventors created a novel plasmid used to package large quantities of high-titer rAAV1 with cDNAs encoding GAA cloned in. Experiments were completed in (*Gaa*^{-/-}) knockout mice treated with the AAV1 vector encoding the mouse GAA gene. Our researchers observed functional enzymatic replacement and glycogen clearance in these animals after gene-replacement therapy.

contact

Elizabeth Garami
University of Florida
Office of Technology Licensing
352/392-8929 • email: egarami@ufl.edu
Reference UF #10859 ~ Patent pending



UNIVERSITY OF
FLORIDA

Office of Technology Licensing

*Facilitating Technology Transfer
To Serve Faculty and Community*
www.otl.ufl.edu