

Researchers continue 'extraordinary measures' to tackle Pompe disease

January 26 2010

As scientists work to find new treatments for Pompe disease -- the devastating genetic "villain" that drives the efforts of the main characters in the new film "Extraordinary Measures" -- University of Florida researchers are hopeful that gene therapy will help patients in the late stages of the disease breathe on their own.

Clinical trials of a [gene therapy](#) for Pompe-related breathing problems in six infants are expected to begin at UF this summer.

Writing today in *Molecular Therapy*, scientists with UF's Powell Gene Therapy Center describe how they used a gel to deliver a corrective gene directly to the diaphragms of a [mouse model](#) of the disease. In people, Pompe disease is a form of muscular dystrophy that causes extreme [muscle weakness](#) and leads to severe breathing difficulty.

"In disease treatment we always think prevention is better and easier than reversal, but we don't always have the opportunity to prevent some diseases," said Dr. Barry Byrne, a UF pediatric cardiologist and the director of the Powell Gene Therapy Center. "When we find out that reversal of what could be considered permanent damage is possible, that is extremely encouraging."

Children with Pompe disease cannot produce the enzyme acid alpha-glucosidase, or GAA. Without the enzyme, sugars and starches that are stored in the body as glycogen accumulate and destroy [muscle cells](#), particularly those of the heart and respiratory muscles.

In the coming gene therapy trial, scientists will incorporate the correct gene to produce GAA into an adeno-associated virus, which already exists in most people, and inject it into each patient's diaphragm. The intent is to "infect" cells of Pompe patients with the genetic machinery they have been missing since birth.

In the latest experiments, UF scientists targeted the diaphragm — an essential muscle for healthy breathing — in mice at different stages of the disease.

"We were able to provide some level of correction in animals of different ages, but it may be more important that we are able to provide benefit to older ones," said Dr. Cathryn Mah, a UF assistant professor of pediatrics. "Many Pompe patients are older children and adults. The longer people have the disease, the weaker their muscles get. Respiratory complications are their No. 1 problem. If we can get some improvement for them, this is worthwhile to keep pursuing."

Byrne believes gene therapy is a way to augment the current treatment for Pompe patients, which involves intravenous infusions to replace the missing GAA enzyme.

Efforts to find a treatment for Pompe disease, some of them based on the work of UF's gene therapy and pediatric doctors, are depicted in Pulitzer Prize-winning reporter Geeta Anand's book "The Cure."

The book became the basis of the film "Extraordinary Measures" starring Harrison Ford and Brendan Fraser, which opened last weekend. Byrne provided technical advice to the producers during filming and has a brief moment as an extra in a scene with Fraser.

The film depicts the true story of John and Aileen Crowley, whose two youngest children were diagnosed with Pompe disease. Faced with no

options, John Crowley quit his job as a marketing executive and started a biotechnology company, Novazyme Pharmaceuticals Inc., which was eventually purchased by Genzyme Corp.

A cure has yet to be found, but Byrne is hopeful that advances can be made to improve the quality of life for patients and their families.

"We think the gene therapy aspect will work alongside the traditional treatment," said Byrne, who is also a member of the UF Genetics Institute. "In chemotherapy, a combination of treatments is used to benefit the patient, and we hope this will work in the same way."

Provided by University of Florida

Citation: Researchers continue 'extraordinary measures' to tackle Pompe disease (2010, January 26) retrieved 20 April 2024 from <https://medicalxpress.com/news/2010-01-extraordinary-tackle-pompe-disease.html>

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