Late-onset Pompe patients in US begin receiving new therapy

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The first commercially available treatment in the United States for patients with late-onset Pompe disease was administered today (Wednesday, June 16) at the University of Florida.

Pompe disease is a rare form of muscular dystrophy and has been the focus of a research program at UF for more than 10 years. It is now part of expanded efforts in neuromuscular disease research.

People 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. Many patients need ventilators to breathe.

The therapy, developed by Genzyme Corp. and marketed under the name Lumizyme, involves intravenous infusions to replace the missing GAA enzyme in patients over 8 years of age.

"We are privileged to participate in the care of patients with Pompe disease and have a dedicated team in both clinical care and research for this form of muscular dystrophy," said Dr. Barry Byrne, the director of the Powell Gene Therapy Center and a member of the UF Genetics Institute. "The use of Lumizyme in the United States is the culmination of many years of work by basic science and clinical researchers around the world. Access to Lumizyme has been long-awaited by the patient community and this marks an important chapter as a specific therapy for this neuromuscular disease."

Although rare, late-onset Pompe disease can occur to patients even in their 60s, who begin showing signs of muscle weakness and respiratory problems, often undiagnosed at an earlier age.

Monique Griffin, 35, of Orlando, was the first patient at UF to receive commercially available Lumizyme -- technically known as alglucosidase alfa.

She was diagnosed with Pompe disease in January 2010, and has been receiving enzyme infusions on a study basis since March. She had formerly been employed as a communications specialist at a casino-resort in Las Vegas before being sidelined by the condition.

She can walk short distances, but largely relies on an electric scooter to move about.

"I noticed some improvement in mobility right after the first few treatments," Griffin said. "This has been a very long process. I had symptoms for 10 years before I finally got a Pompe diagnosis, and I was in constant pain for most of 2009, so I have already felt some benefits of this treatment. I still have flare-ups, but I am not as tired and have had some slight improvements in endurance and mobility. I still only walk very short distances, but I am more stable."

The infusion takes about four hours.

"This is an important day for the Pompe community, especially for those patients with late-onset Pompe disease in the United States who are awaiting treatment for this devastating disease," said John Butler, president of Personalized Genetic Health at Genzyme. "We appreciate the efforts of the University of Florida over the years to support the Pompe community, and their partnership with Genzyme to provide access to therapy for patients as quickly as possible."

During Lumizyme's preapproval period, Genzyme worked with patients and physicians to assure that the most severely affected late-onset patients in the United States could access therapy.

"There has been a long period of review and discussion," said Byrne, a pediatric cardiologist.

"Now that the treatment's commercially available, there will certainly be an impact for a larger proportion of patients. Future research of this type will be greatly facilitated by UF's Clinical and Translational Science Award from the National Institutes of Health."

Provided by University of Florida