Spark Therapeutics launches gene therapy clinical trial for late-onset Pompe disease

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Reviewed by Emily Henderson, B.Sc. Feb 3 2021

Pompe disease is a hereditary genetic disorder caused by a deficiency of acid alpha-glucosidase leading to build-up of glycogen in the lyosomes, which then causes cell damage in various tissues, in particular the heart, the muscles, the liver and the nervous system.

In patients living with late-onset Pompe disease the respiratory system and mobility are most frequently the most affected by the disease, often requiring the use of a wheelchair and respiratory assistance, and life expectancy can be reduced.

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The company Spark Therapeutics has just announced that a first study participant has been dosed in the USA as part of an international gene therapy clinical trial. The product tested - SPK-3006 - is the result of collaboration with Genethon. It utilizes technologies developed at Genethon, and which, in mouse models, have already helped correct the build-up of glycogen in the muscles and nervous system.

The purpose of this study is to evaluate the safety, tolerability, and efficacy of a single intravenous infusion of SPK-3006 in adults with clinically moderate, late-onset Pompe disease receiving enzyme replacement therapy (ERT). This study is taking place first in centers in the USA, and then should be continuing in Europe.

> *"We are delighted about the start-up of this clinical trial for late-onset Pompe disease led by Spark Therapeutics, with a transgene that integrates technologies designed at and licensed from Genethon, where the in-vivo proof of concept in pre-clinical models was demonstrated. It is an important step in the treatment of this serious disorder, for which there are currently few therapeutic options, and which again illustrates the quality of our R&D to serve patients suffering from rare diseases."*
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> *Frédéric Revah, CEO, Genethon*

The study plans to recruit and treat around twenty patients, and is set to end in October 2023.