Press Release
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Genethon is delighted about the launch of a gene therapy clinical trial for late-onset Pompe disease, led by the company Spark Therapeutics, using technologies developed at Genethon.

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.

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," explains Frédéric Revah, CEO of Genethon, "and which again illustrates the quality of our R&D to serve patients suffering from rare diseases."

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

Find out more about the Spark Therapeutics clinical trial

- RESOLUTE Study of late-onset Pompe disease (LOPD) for HCP
- Clinical Trials website

About Genethon

Genethon, a pioneer in gene therapy for rare diseases, was created in 1990 by AFM-Telethon, thanks to donations from the first French Telethons. The challenge at the time was huge: deciphering the human genome, tracking the genes responsible for genetic diseases and using this knowledge to create innovative medicines. Today, a product incorporating patents and the results of research work conducted at Genethon is available on the market for spinal muscular atrophy, a neuromuscular disease, and a second product is at the marketing authorisation application stage, for a rare disease affecting vision. A total of 9 other products resulting from R&D at Genethon, developed alone or in collaboration, are at the clinical trial stage around the world for rare diseases of the muscle, vision, liver, immune system and blood, and a further 8 products are at the preclinical stage.
Genethon.fr/en
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About Spark Therapeutics

At Spark Therapeutics, a fully integrated, commercial company committed to discovering, developing and delivering gene therapies, we challenge the inevitability of genetic diseases, including blindness, hemophilia, lysosomal storage disorders and neurodegenerative diseases. We currently have four programs in clinical trials. At Spark, a member of the Roche Group, we see the path to a world where no life is limited by genetic disease. For more information, visit www.sparktx.com, and follow us on Twitter and LinkedIn.

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