Press Release
Evry, France - 11th January 2018

Myotubular Myopathy

Genethon welcomes interim data of a phase I / II clinical trial with a gene therapy product developed in its laboratories

On September 21, Audentes Therapeutics, a biotechnology company, launched the first gene therapy clinical trial in children with myotubular myopathy disease using a gene therapy product designed at Genethon (Read the press release). At the 12-week timepoint, the first 3 children treated show early signs of efficacy.

Myotubular myopathy is an X-linked genetic disease affecting 1 in every 50,000 newborn boys. It is caused by mutations in the MTM1 gene encoding myotubularin, a protein involved in the development and function of muscle cells. It is characterized by extreme muscle weakness and respiratory failure, with an estimated 50% mortality rate by 18 months of age.

From Genethon-designed product to the early signs of muscle strength in treated children

With the support of the AFM-Téléthon and the Myotubular Trust, Dr Ana Buj-Bello led the Genethon team in designing an adeno-associated virus (AAV) capable of delivering the MTM1 gene to muscle cells. Preclinical studies demonstrated spectacular efficacy in a mouse model and in dogs naturally affected by the disease. On the basis of this early work, Audentes Therapeutics initiated an international phase I / II clinical trial, aimed at evaluating the safety and efficacy of the product in approximately 12 patients less than 5 years of age.

Twelve weeks after administration in the first child, Audentes Therapeutics reported positive interim data from the first cohort of patients, including:

- Significant improvements in neuromuscular function
- Significant improvements in respiratory function
- Improvement of the general condition of treated children

To discover in detail the results for each of the 3 children treated: http://prn.to/2lXCrsE

« These first promising results demonstrate Genethon's expertise in developing gene therapy products. It took 8 years of research and development and several million euros to reach this trial. With these first results, which will have to be confirmed by long-term analyses to establish the therapeutic benefit of the treatment, we are deeply proud that our expertise based on nearly twenty years of pioneering research conducted in gene therapy development now

benefits the first patients affected by this particularly serious disease, » said Frédéric Revah, CEO of Genethon.

Dr. Ana Buj-Bello, researcher at Genethon, author of preclinical development said: *« We have worked on the development of the vector and are delighted for the boys and their families about these promising preliminary results of the clinical trial, which give hope also for other genetic disorders of the muscle".*

About Genethon - www.genethon.fr

Created and financed by the AFM-Téléthon, Genethon's mission is to provide patients with innovative gene therapy treatments. After having played a pioneering role in the decoding of the human genome, Genethon is today, with more than 150 researchers, doctors, engineers, regulatory affairs specialists..., one of the main international centers of preclinical and clinical research and development of gene therapy for rare diseases.
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