PRESS RELEASE

May 15, Evry (France)

ASGCT 18th Annual Meeting 2015:

Ana Buj Bello receives the Outstanding New Investigator Award

Dr. Ana Buj Bello, Inserm research scientist and head of the neuromuscular disorders team at Genethon, received the Outstanding New Investigator Award from the American Society of Gene & Cell Therapy (ASGCT) at the 18th Annual Meeting (May 13-16, 2015/New Orleans). This award recognizes four researchers every year for the quality and importance of their work in gene and cell therapy.

Ana Buj Bello is awarded for 10 years of pioneering work on gene therapy and currently leads a translational gene therapy program for myotubular myopathy, a very severe muscular disease t due to mutations in the gene MTM1, at Genethon, the laboratory of the AFM-Telethon.

In 2008, her work provided the proof of concept of the efficacy of an intramuscular injection of an adeno-associated viral vector (AAV) expressing the MTM1 gene in a mouse model of myotubular myopathy.

In 2009, Dr. Ana Buj Bello's team initiated at Genethon the first intravenous gene therapy assays in the mouse model of the disease to correct MTM1 gene dysfunction in all muscles of the body.

In 2014, she demonstrated, with her team, and in collaboration with two other teams in the United States (University of Washington and Harvard Medical School), the efficacy of this gene therapy in a dog model of myotubular myopathy. MTM1 gene transfer restored muscle function and prolonged the life of the treated animals (*Science Translational Medicine*, January 22, 2014). [Discover the images of treated dogs]

Dr. Ana Buj Bello: "*Receiving this award is an honour and a great satisfaction after many years of research dedicated to the gene therapy of myotubular myopathy. The next step is now to launch a clinical trial to treat children affected by this disease.* "

Frédéric Revah, CEO Genethon is also pleased: "*We are particularly proud of this recognition of the extraordinary work of Ana Buj Bello. This pride is shared by all teams at Genethon that contribute to the highly complex development and production of this product for the treatment of myotubular myopathy, for which further development is conducted in partnership with the biotechnology company Audentes Therapeutics, who will also be responsible for its commercialisation*".

Myopathy myotubular

Myotubular myopathy is a severe X-linked genetic disease that affects 1 in 50,000 newborn boys. It is due to mutations in the MTM1 gene encoding myotubularin, a protein required for the functioning of muscle fibres. The disease causes generalized hypotonia and muscle weakness leading to death during early childhood. There is currently no effective treatment for this rare disease.

About Genethon - www.genethon.fr

Created by the AFM-Telethon and funded by incomes from Telethon's activities, Genethon's mission is to provide innovative gene therapies for rare disorders. After a pioneering role in deciphering the human genome, Genethon is today, with over 200 researchers, physicians, engineers, regulatory affairs specialists, etc., a leading research center for preclinical and clinical development of gene therapy treatments for rare diseases. Genethon has the largest laboratory for GMP production of gene therapy products, Genethon Bioprod. In 2015, Genethon was one of 16 winners of the World Innovation Competition 2030 for its project on the development of an industrial process for production of gene therapy vectors.

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