

The European charity Myotubular Trust is supporting Généthon in developing gene therapy for myotubular myopathy

Evry (France), June 24, 2013. The European charity Myotubular Trust announces its support to the development of a pre-clinical gene therapy trial for myotubular myopathy, a project co-ordinated by Dr. Anna Buj-Bello at Généthon (Evry, France).

Congenital myotubular myopathy is a neuromuscular disease of children caused by mutations in the MTM1 gene encoding myotubularin, a protein involved in the functioning of muscle cells. Since 2009, Dr Buj-Bello has been working at Généthon on the development of a genetic therapy for this disease, which affects 1 out of 50,000 newborn males.

The European charity Myotubular Trust, based in the UK, the largest association of patients and parents in Europe for this disease, decided to support Dr Buj-Bello's team with a grant of € 120,000 over 2 years.

Anne Lennox, General Director of the Myotubular Trust stated: *"We are pleased to support the work of Dr Anna Buj-Bello, who was the first researcher to demonstrate that gene transfer extends the life of mice suffering from myotubular myopathy. Thanks to many years of research and international co-operation, the work of Généthon, a world-renowned research organization with unique expertise in the field of gene therapy, allows us to hope that we will soon have a treatment for this disease".*

Fulvio Mavilio, Scientific director of Généthon, said: *"The support given by the Myotubular Trust will enable Dr Buj-Bello's team to make quicker progress in developing a gene therapy clinical trial for myotubular myopathy. For such a rare disease, co-operation with an international patient association is a major advantage and a key success factor".*

In the context of the project funded by the Myotubular Trust, Généthon will deploy its experience and know-how in developing gene therapies for rare diseases. Généthon has unique expertise in this field: it is the promoter of two ongoing international clinical trials for primary immunodeficiencies, participates in a trial addressing a rare eye disease that will start before the end of 2013, and has several trials in preparation addressing rare diseases of the muscles, blood, liver and central nervous system.

Myotubular myopathy

Myotubular myopathy is a rare genetic disease presenting at birth and transmitted by an X-linked recessive mode. It therefore mainly affects boys. It is a severe disease leading to general hypotonia and death, generally during the first years of life. The disease is caused by a mutation in the MTM1 gene, encoding a protein – myotubularin – involved in lipid metabolism in the muscle.

The symptoms at birth are mainly hypotonia and respiratory distress. Problems with swallowing, paralysis of the facial muscles responsible for eye movements, dropping eyelids and deformations of the chest and feet are frequently associated with this disease.

About Généthon

Généthon, located in Evry, France, is a non-profit organisation dedicated to the development of biotherapies for orphan genetic diseases. Généthon has unique experience in the international research community in gene therapy and muscle disorders, and has several clinical trials ongoing or in preparation for neuromuscular, blood, liver and eye diseases. Généthon has one of the largest viral vector production facilities in the world, and has unique expertise in assessing the quality and efficacy of viral vectors for clinical application.

About Myotubular Trust

Anne Lennox and Wendy Hughes, two parents of children affected by myotubular myopathy, set up the Myotubular Trust in February 2006. It was very clear that as a rare condition, research into myotubular myopathy could lag substantially behind the scientific developments in other fields of muscle disease, due to lack of dedicated funds, and the difficulty of "competing" with other more common diseases. The Founding Patron is the renowned Professor Victor Dubowitz, Emeritus Professor of Paediatrics at University of London and President of the World Muscle Society. Professor Francesco Muntoni, Professor of Pediatric Neurology and Head of The Dubowitz Neuromuscular Centre at the Institute of Child Health/Great Ormond Street Hospital for Children, London, serves as scientific advisor and Chair of the Scientific Advisory Board, which supervises research grant awards. In 2010 the Trust became incorporated as a company limited by guarantee (07260229) registered in England and Wales.

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