GFB

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THE FIRST PHASE OF THE AMERICAN PROJECT ON THE LGMD2E HAS BEEN SUCCESSFULLY COMPLETED

In 2013 it has already been started, at the Research Institute by Nationwide Children's Hospital - Columbus Ohio, the beta-sarcoglycan gene transfer project for treatment of Limb Girdle Muscular Dystrophy Type 2E.

Dr. J.R. Mendell and Dr. L. Rodino Klapac had started their work, the scientific way to be followed is based on their experiences, gained by completing a similar trial for the treatment, throughout a gene transfer therapy, of the Limb Girdle Muscular Dystrophy Type 2D. Dr. Mendell explained in details aims and phases of the pathway to be followed:

Aim 1. Determination of pre-clinical efficacy of the transfer of human b-sarcoglycan gene, using recombinant adeno-associated virus to act as delivery vehicle, in b-sarcoglycan deficient mice. Time required: one year.

Aim 2. Regulatory preparation for a "recombinant adeno-associated virus human b-sarcoglycan" gene transfer intramuscular clinical trial, including formal toxicology/biodistribution study and clinical vector production. Time required: about one year.

Aim 3. Perform an intramuscular clinical gene therapy trial with recombinant adeno-associated virus human b-sarcoglycan transfer (into the "extensor digitorum brevis" muscle) in LGMD2E patients. Time required: about one year starting since the closing of the previous aim.

Today the first phase of the project has been successfully completed, in 2014 the second phase will be developed, so to reach the first clinical trial for beta-sarcoglycan in 2015.

Moreover, Dr. Mendell underlined his clear intention to take this project to the following important level (Aim 4) which target is the vascular delivery of the missed gene to the lower limbs. He is in fact confident this can be achieved with an outcome that will mean widespread gene expression into the treated muscles and functions improvement. At this purpose they are already testing and discussing, with U.S. FDA, a procedure which will be developed and carried out during the next eighteen months.

We wish to underline this trial-study, as we know up to now, is today **the most important worldwide scientific project entirely and solely committed to the research of a treatment for LGMD2E**. It is therefore extremely important for us and represents a substantial step in order to get more attention to the disease that affects our children. We can say it is a reward for all the efforts we have made till now, independently on the results and the applications it will produce.

THE GFB ONLUS HAS BEEN ENROLLED AT THE NATIONAL CENTRE FOR RARE DISEASES

The GFB ONLUS has been enrolled in the associations of patients' groups, at the National Institute of Health - National Centre for Rare Diseases.

The list can be found at the link:

http://www.iss.it/site/cnmr/dina/asso/as01.asp

A REGISTRY FOR PATIENTS LGMD2C

The American Foundation "Kurt + Peter Foundation" collects in its patient registry the data on patients with LGMD2C (gamma-sarcoglycan deficiency). This registration is done online at the link provided below. It is in 3 parts and lasts about 15 minutes. The answers will be very useful for the analysis of variable progression of this disease and several other features specific to the LGMD2C. We recommend you to complete the form in its entirety, although some of the questions are optional.

http://www.kurtpeterfoundation.org/patient_registry

A POSTER OF GFB ONLUS AT THE EUROPEAN CONGRESS OF RARE DISEASES ECRD2014 AND AT THE INTERNATIONAL CONGRESS OF NEUROMUSCULAR DISEASES ICNMD2014.

The GFB NGO will present a poster in electronic format at the European Congress of Rare Diseases ECRD2014, which will take place in Berlin, in the days 8-9-10 May, where the latest updates regarding the sarcoglycanopathies will be presented. From July 7 to 10, the researcher Paola Bonetti, a member of the Committee of the GFB medical and scientific non-profit organization, will present the poster of the association in Nice, at the International Congress of Neuromuscular Diseases ICNMD2014.

http://www.rare-diseases.eu/ http://icnmd2014.org/en/
