GFB
BETA-SARCOGLYCANOPATHY FAMILY GROUP

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THE GFB ONLUS AND PROF. ANGELINI CORRADO HAS ORGANIZED THE CONGRESS "LGMD DAYS"

On days 15, 16, 17 October will be held the Conference "LGMD DAYS" in Lido di Venezia (VE) at the Institute IRCCS "San Camillo," Via Alberoni no. 70.

The Conference, in Italian language, will be entirely devoted to the clinical aspects of Limb Girdle Muscular Dystrophy, with a special focus on Sarcoglycanopathies.

On the Association's website, at the following link, have been published

- The provisional program of the conference,
- the registration form,
- any accommodation in Hotel,
- information on ferries to reach the location of the congress.

http://www.beta-sarcoglicanopatie.it/index.php?option=com_content&view=article&id=150

THE MYO-SEQ PROJECT: THE CTSR MADE IT POSSIBLE

There are a number of specific challenges at every stage of rare disease (RD) therapy development: incomplete knowledge and understanding of disease prevalence and disease course, genotype-phenotype correlation, modifying factors, response to treatment, potential side-effects, as well as difficulties in accessing biomaterials and identifying patients. In the neuromuscular field, that last specific challenge has become less cumbersome thanks to the existence of the Care and Trial Site Registry (CTSR).

About MYO-SEQ: Next Generation Sequencing (NGS) and other -omics technologies, in combination with powerful IT infrastructure and sophisticated bioinformatics tools, allows, in principle, the entire genetic profile (exome, genome) of any RD patient to be deciphered. It can also enable the integration of this information with clinical data (e.g. from deep phenotyping, ontologies, electronic health records) as well as data from clinical studies (such as response to treatment, side effects, pharmacogenomics) and biomarker studies (proteomics, metabolomics).

In this context, the neuromuscular team at Newcastle University has conceived the MYO-SEQ project that will focus on the application of NGS, in particular Whole Exome Sequencing (WES), in a large cohort of patients with unexplained limb-girdle weakness (LGW). Focusing on undiagnosed patients with a clearly defined clinical phenotype will enable increased diagnostic rates for disease causing mutations in known genes in this cohort, while the use of WES provides scope both for new gene discovery and for additional research into disease modifiers and genotype-phenotype correlation with substantial cost effectiveness.

The initial inquiry to test the feasibility of the study was carried out with a short questionnaire publicized via the CTSR. The excellent response from the CTSR affiliated clinical centres has directly resulted in the engagement of private and non-for-profit funders to support the execution of the project that is expected to start in August 2014. In 55 European centres more than 1000 patients will be sequenced in the next year and a half.

If you have questions about MYO-SEQ or would like to participate, please contact Monica Ensini (monica.ensini@ncl.ac.uk)

The CTSR has proven here quite clearly its capability to galvanize interest and collaboration efforts that can then be translated into action in a fast and effective way.

THE GFB ONLUS WAS RECORDED IN THE REGIONAL REGISTER OF FAMILY SOLIDARITY ASSOCIATIONS

After a year of its establishment as a Volunteer Organization, the GFB ONLUS was also recorded in the regional register of Lombardy of Family Solidarity Associations .

ON THE SITE OF THE GFB ONLUS THE COUNT OF PATIENTS BELONGING TO THE GROUP

The GFB ONLUS has published on its website the count of patients so far registered in the group since 2010, with particular reference to the country of origin

http://www.beta-sarcoglicanopathy.org/index.php?option=com_content&view=article&id=47&Itemid=53