

**TREAT-NMD Partner Newsletter No. 13
and Club of Interest Newsletter No. 7**

1st June 2007

Welcome to the seventh newsletter for the TREAT-NMD Club of Interest. This week's edition features a report from the recent University of Basel TREAT-NMD workshop on 'selecting appropriate mammalian animal models' and an interesting contribution from the Czech Republic regarding Czech interest in TREAT-NMD and their project to create a "Virtual Neuromuscular Centre" (see section 3).

We hope you enjoy the newsletter and look forward to hearing your comments - write to info@treat-nmd.eu with anything you'd like to say. Feel free to forward this message to anybody you think might find it of interest, or invite them to sign up to receive the newsletter by visiting our website.

Best wishes,

Katie, Volker, Stephen, Emma, Arron and Rachel – the TREAT-NMD coordination team

Newsletter contents

- 1. About this newsletter**
- 2. Working with us**
- 3. TREAT-NMD news and reports**
- 4. Calls for proposals**
- 5. Upcoming conferences, meetings and workshops**
- 6. Partner-specific items**
- 7. Send us your news and views!**



1. About this newsletter

This is a weekly newsletter sent to all members of TREAT-NMD's "Club of Interest" worldwide. We are receiving new subscriptions all the time, so if you've missed the earlier editions of the newsletter and would like to catch up, please visit our newsletter archive online at <http://www.treat-nmd.eu/news/newsletter/> where you will find all back-issues. If you have received this letter from a friend or colleague and would like to subscribe directly, please visit our website at <http://www.treat-nmd.eu/> where you will find a subscription form at the bottom of the homepage. You can also use the same form if you no longer wish to receive this newsletter – just select the unsubscribe button.

2. Working with us

TREAT-NMD aims to be an inclusive rather than an exclusive network, and you do not have to be based in Europe or be a partner to be involved. International collaboration with experts from all over the world is already taking place, and new links are being developed.

If you are involved in any of TREAT-NMD's areas of interest and have something you'd like to say or a suggestion of where we could work together, we encourage you to get in touch by writing to us at info@treat-nmd.eu

3. TREAT-NMD news and reports

A Czech "Virtual Neuromuscular Centre"

Last week the TREAT-NMD Coordination Team in Newcastle were visited by a team of Czech delegates interested in the future integration of the Czech Republic into the TREAT-NMD network.

As regular readers of this newsletter are aware, one of TREAT-NMD's primary aims is to reduce the fragmentation that is currently hindering research and treatment of neuromuscular diseases in Europe and worldwide. This fragmentation often exists not only internationally but also within countries – patients in one part of a country might receive different treatment and differing levels of care to patients in another part of the same country, and the fact that one particular clinician or treatment centre is involved in TREAT-NMD does not necessarily mean that others in that country are even aware of the network. This is a key challenge we have to overcome, and for this reason we are always keen to hear of cases where a number of groups and institutions in the same country are able to work together to create the in-country networks necessary to ensure that best practice and latest developments can easily be spread throughout the country. Precisely such an initiative – the creation of a "virtual neuromuscular centre" – is already underway in the Czech Republic, and we were very interested to meet with some of the originators of this significant project.



Report by Dr Petr Vondráček (pvondracek@fnbrno.cz)

The Czech delegation invited to visit the TREAT-NMD Coordination Centre consisted of four participants: neuromuscular consultant Dr Petr Vondráček and geneticist/molecular biologist Dr Jana Sedláčková (both of Masaryk University in Brno), plus two representatives of Czech Parent Project – Pavlína Petrásková and Miroslav Stuchlík. Our discussions focused on several main areas:

- The Czech team provided an overview of the current situation in the Czech Republic, where we have a network of five university hospitals collaborating in the field of diagnostics and treatment of



neuromuscular diseases. These centres consist of multidisciplinary teams of clinicians, geneticists, molecular biologists and pathologists who have access to advanced diagnostic and IT equipment.

- We expressed our desire to integrate our national network into the TREAT-NMD project, within the framework of which we would be interested in actively participating in multicentric clinical trials of new drugs and therapeutic strategies.
- We presented our online database of Czech and Slovak DMD/BMD patients and female carriers, which is a joint project between Czech Parent Project and Masaryk University and already contains the full phenotypic and genotypic profiles of 126 patients. This database can be accessed online at <http://dystrophy.registry.cz/>. Plans are already underway to integrate our database into the European-wide TREAT-NMD database and we are in close contact with the leaders of this particular TREAT-NMD activity.
- We detailed our plans for our project for a national “Virtual Neuromuscular Centre”, which would ensure the comprehensive multidisciplinary management of patients with DMD/BMD, SMA and other less common neuromuscular diseases in the Czech and Slovak republics. Within this project we are prepared to adopt and implement TREAT-NMD’s recommended European standards. Czech Parent Project intend to submit a grant request to the Czech Ministry of Health in order to secure the funding for this project. Included in the proposal will be funding for the position of a “liaison officer” responsible for the coordination of the activities of the Czech virtual network in relation to the activities of TREAT-NMD.
- We would value the opportunity for our clinicians, molecular biologists, physiotherapists etc. to take part in the training programmes that TREAT-NMD intends to organise in order that we can implement European standards in our clinical and diagnostic practice as effectively as possible.



About Czech Parent Project – summary by Pavlína Petrásková (parentproject@parentproject.cz)

From its launch in 2001, Parent Project in the Czech Republic was aware that it was necessary to work closely with specialists to ensure its projects were professionally sound. We also knew that establishing a database and specialist clinics in the Czech Republic would be a necessity. It was clear that we would receive hardly any state funding for our 500 patients, even though we drafted a number of projects and requests. Nevertheless, with the help of parents we eventually managed to finance our first major project, the database. All parents in our team work on a voluntary basis, but we naturally have to pay for the work of doctors and other specialists.

It was Parent Project who came up with the idea for a Neuromuscular Centre in the Czech Republic: we are beginning the process of acquiring funds for this project and for the potential integration of the Czech Republic into TREAT-NMD and into joint research projects. Patient care is paid for by the state through the public health insurance scheme, but there remains the question of what care is required, and whether potential future treatments for NMDs will be funded. We therefore intend to continue to support our main priorities:

- joint research projects within TREAT-NMD,
- improving specialist care for patients with NMDs and thereby improving their quality of life.

We are able to raise funds from our own resources, from various European calls, from the Ministry of Health and from private foundations and donors. This year we decided that we will also set up a “Parent Project Foundation”, which will be devoted exclusively to supporting research projects.

We are very glad that TREAT-NMD has come into being – we had ourselves been constantly wondering how to unite individual countries with the aim of accelerating drug development and slowing the progress of the disease. What your team is doing is simply fantastic and we do not want to be merely passive members but would wish to actively help reach our shared goals. That is our mission.



Report on the first workshop of WP 7.2 – Select appropriate mammalian animal models By Dr. Raffaella Willmann, University of Basel

A first workshop was held on May 21 at the Biozentrum of the University of Basel (Basel, Switzerland) discussing WP 7.2: “Select appropriate mammalian animal models”. The partners participated either by attending the workshop or by contributing to the discussion by means of a telephone conference. From the Club of Interest, Drs. Reginald Bittner (DMD mouse model; Vienna), Judith Melki (SMA mouse model; Evry), Stephane Blot (DMD dog model; Alfort), Annamaria de Luca (exercised mdx-model; Bari) were present or participated on the phone.

The main objective of the workshop was to discuss the currently available mammalian animal models and to reach a consensus as to which models are useful for the acceleration of preclinical development. The discussion regarding updating the list of currently used animal models was very lively. It also became clear that some of the animal models discussed, although of high potential benefit, do not yet fulfill critical criteria such as general accessibility for the entire community, low cost, and availability of reliable reference data. On this basis, the WP 7.2 team will now proceed to the final assessment that will allow selecting those mammalian models that are best suited for preclinical studies and that will serve as reference for emerging new animal models.

Active discussion lists

- registries and biobanks,
- standardised assessment of animal models,
- outcome measures for clinical trials (especially patient’s perspective),
- standards of diagnosis and care in DMD and SMA.

If you would like to be involved in any of these discussions or if you know someone who would like to contribute, please let the TREAT-NMD office know (by writing to rachel.thompson@treat-nmd.eu).

Call for contacts (AFM)

VLCAD deficiency is a rare hereditary disease responsible for a myopathy with exercise intolerance, muscle weakness and myalgia. Affected patients are subject to rhabdomyolysis in response to exercise or other stress situations, and this manifestation of the disease is well recognized by the appearance of brown urine, due to myoglobinuria. There is to date no treatment for this myopathy. The disease is due to mutations of a gene encoding the Very Long Chain AcylCoA Dehydrogenase, an enzyme which belongs to the mitochondrial Fatty Acid β -Oxidation (often referred as FAO or β -oxidation) pathway. The disease is well characterized at the molecular level, and is part of the disorders that are systematically screened in newborns in Germany, in Australia and the US, through neonatal screening programs.

Recent research conducted in France led to the suggestion that some existing drugs might have beneficial effects in this pathology, as well as in other myopathies due to FAO disorders.

Before considering the organization of any tests in patients, we need to estimate the potential cohort of patients who present with *myopathy due to VLCAD deficiency*, with *myopathy due to defect in another FAO defect*, or with *myopathy of unknown origin* (showing similarities with the VLCAD or FAO deficiencies on the basis of clinical and biological investigations).

This call for contacts aims to collect information on the number of patients who fulfill these criteria and their geographical location – these are essential factors to evaluate the feasibility of an eventual clinical trial.

This call for contacts is coordinated by the Association Française contre les Myopathies (AFM). If you are able to contribute any information, please contact Hervé Laouenan at hlaouenan@afm.genethon.fr



4. Calls for proposals

We regularly publish calls for proposals that might be of relevance to TREAT-NMD in this section of the newsletter. The network has ambitious plans in areas such as training and education, and since these are not covered by our initial funding from the EU, we have to seek funds from elsewhere. We strongly encourage anyone outside the network who is interested in developing links with us to look through these calls and see if there is anything of relevance to you. We also encourage partners to consider possibilities for academic exchanges via these proposals.

Attracting additional funding for network activities is vital if we are to ensure the future sustainability of the network. There are a number of research-based calls for proposals under FP7 that would be suitable for the network, and we would like to ensure that the network has every opportunity to obtain funding through FP7. Therefore, it is important that the Coordination Team at Newcastle (stephen.lynn@treat-nmd.eu) are kept up-to-date on any of your research proposals that are associated with the network activities, as we can offer help and support as well as a united front in which we work together to win European funding either as a single partner or in collaboration with other partners and non-network colleagues.

The call below is specifically related to exchanges involving the UK, but there are likely to be similar initiatives organised by other countries. If you are aware of any that might be of interest to readers of this newsletter, please send details to stephen.lynn@treat-nmd.eu so that we can publicise them in future newsletters. This particular call came to our attention rather late, but we understand it is likely to be repeated next year, so we encourage anyone considering an exchange programme next year to bear this in mind.

The British Council

The British Council is announcing a call for proposals under its initiative aimed at supporting new links between early stage researchers in the UK and in other countries.

The Researcher Exchange Programme (RXP) is a £250,000 initiative that provides individual researchers with awards covering travel and subsistence costs, and some consumables costs, needed to develop new scientific collaborations and contacts through exchange visits of between one week and three months' duration.

The research link can be in any area of science, engineering and technology, including social sciences and humanities.

The term 'early stage researcher' refers to researchers at the beginning of their research careers.

The applicant must have:

- either obtained a doctoral qualification or be within twelve months of obtaining a doctoral qualification.
- no more than two years full tenure in a university or research institute.
- no more than six years overall active researcher experience.

The maximum award that can be applied for is £5,000.

Please note that more than one researcher from the same sending or receiving research group can apply for an award, but each must submit a separate application.

The closing date for applications is 2 June 2007.

For full details and an online application form go to www.britishcouncil.org/science-rxp



5. Upcoming conferences, meetings and workshops



European Society for Muscle Research

- XXXVIth EMC meeting of the European Society for Muscle Research
- Stockholm, Sweden September 8–12, 2007
- <http://www.esmr.org/EMCSt1.htm>

TREAT-NMD meetings will be advertised in both the weekly newsletters and the TREAT-NMD website – please see the “news and events” section for details.

If there is an event you would like us to publicise in future newsletters and on our website, please send details to rachel.thompson@treat-nmd.eu

6. Partner-specific items

TREAT-NMD 6 month activity report

The 6 month activity report is due on the 15th June 2007.

1. The Activity Leader should ask each **Work Package Leader** from her/his Activity to complete the information on her/his WP (i.e. Work Package objectives, Progress towards objectives, Ethics, Deviations from project work program, deliverable and milestones and dissemination of knowledge). WP leaders can do this by updating their 3 month report and sending it to their Activity Leader.
2. The Activity Leader **consolidates** information from the Work Packages of her/his Activity, and provides an overview of actions undertaken.
3. The Activity Leader **sends** the consolidated report to ACIES (eu-new@acies.fr) no later than the **Friday 15th June**

Cost-Effort forms

The first overview on costs and resources spent during these 6 first months of TREAT-NMD is required before the Governing Board meeting at the end of June. Therefore, could you please send ACIES your cost and efforts follow-up sheets before the **15th June**? These sheets, designed for each partner, are **downloadable** from the private part of the website (<http://www.treat-nmd.eu/private/> under “Cost forms”).

Presentation of questionnaire results

Partners who asked for questions to be included in the fact-finding questionnaire received their results last week. If you are one of the partners who has been asked to present a poster at the Governing Board Meeting, we would ask you to devote one part of the poster to providing a brief summary of any important findings from the questionnaire and any action required. If you have any questions regarding this please contact emma.heslop@treat-nmd.eu.

Posters and presentations for Governing Board meeting

Partners have been asked by Stephen Lynn to prepare and present posters and/or presentations for the Governing Board meeting. Templates are available to download from the private section of the website <http://www.treat-nmd.eu/private/>.

As a general guide, can we please request that all posters should be printed A1 size (594x841 mm or 23.4x33.1 inches). The posters will be on display from the evening of Sunday 1st July until the close of the meeting on Tuesday 3rd July. The UNEW team are very happy to help you in preparing the content for these posters, so please feel free to contact us.

You will also find on this web page a template for producing PowerPoint presentations related to the network. Please use this template when designing presentations that you intend to give on your activities within the network. We hope you find these templates useful.



Reimbursement guidelines

As the number of TREAT-NMD activities are increasing across the network reimbursement guidelines have been drawn up to help simplify your planning and implementation of TREAT-NMD related meetings and workshops. These guidelines have been posted on the private section of the web site and will be incorporated into the next version of the TREAT-NMD Project Management Manual. Please print-out and read these guidelines – they will help you to correctly identify the source for reimbursement when attending workshops and meetings.

TREAT-NMD flyers

We have had a number of glossy flyers printed to distribute within the neuromuscular community and would be happy to send some to any partner wanting to help publicise TREAT-NMD, whether this be at international conferences or internal meetings. If you would like to receive a pack please e-mail Rachel Thompson at rachel.thompson@treat-nmd.eu

Discussion forums / lists

Would you like us to set up a discussion forum for you on the TREAT-NMD website?
If so, please e-mail rachel.thompson@treat-nmd.eu.

Calls for proposals / funding opportunities

Please forward to us at the Coordination Office any calls for proposals and funding opportunities you receive within your institution. We will then advertise these in the newsletter and on the website.

7. Send us your news and views!

We encourage all partners and supporters to send their own news and updates and we will be happy to include them in future editions of the newsletter. Please send your contributions to emma.heslop@treat-nmd.eu

