



## TREAT-NMD Partner Newsletter No. 3

23 March 2007

Welcome to the 3<sup>rd</sup> weekly newsletter for all TREAT-NMD partners!

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### 1. About this newsletter and the mailing list

You are a "key contact" for your partner organisation and you have been subscribed to a mailing list called "treatnmd-partners", which is intended to be a simple way for partners to keep in touch with one another. If you ever want to write a message to reach the key contacts in every partner organisation, write to [treatnmd-partners@newcastle.ac.uk](mailto:treatnmd-partners@newcastle.ac.uk) and your message will automatically be distributed to everyone.

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### 2. Send us your news and views!

We encourage all partners to send their own news and updates, either directly to all partners by writing to this list at [treatnmd-partners@newcastle.ac.uk](mailto:treatnmd-partners@newcastle.ac.uk) or to [emma.heslop@newcastle.ac.uk](mailto:emma.heslop@newcastle.ac.uk) for inclusion in the next newsletter. In future we'd like to include other sections in the newsletter, such as one on newly published research that is of interest to the network. What else would you like us to include? Write to [emma.heslop@newcastle.ac.uk](mailto:emma.heslop@newcastle.ac.uk) with your feedback.

Please e-mail us with any information you have on upcoming education and training opportunities including workshops, conferences, funding, exchange programmes, clinical placements, visiting professorships and lectureships.

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### 3. Publicising TREAT-NMD at conferences

As part of efforts to raise the awareness of and publicise TREAT-NMD at conferences, workshops or other meetings we have produced a general double-sided colour flyer introducing TREAT-NMD and detailing the TREAT-NMD partner organisations. The flyer is now **available for download** from the TREAT-NMD web site at <http://www.treat-nmd.eu/assets/documents/TREAT-NMDFlyer.pdf>. We will shortly be adding an editable version in Microsoft Publisher format, which you can download and have printed yourself.

If you are planning to attend any workshops, conferences or meetings please let us know and please take our promotional material along to help promote TREAT-NMD. The TREAT-NMD logo is available in 3 sizes to download from the website. Please also ensure that you include it on any abstract, papers and posters you prepare in which you mention TREAT-NMD.

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#### **4. Resources**

The TREAT-NMD website will expand substantially over the coming months and will provide useful resources for partners – for reporting on activities, for creating email forums, uploading presentations and conference/workshop notifications. Please let us know what you would like to see on YOUR website!

A second, more general newsletter will be distributed to our wider audience on at least a quarterly basis beginning in May 2007. If you make any new contacts in the neuromuscular world over the coming months and years please promote TREAT-NMD and provide us with their e-mail address so that we can add them to our distribution list.

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#### **5. Progress update on active work packages**

##### WP05.1 – Design and Implement the Clinical Trials Coordination Centre (Rudolf Korinthenberg, WP Leader)

Key personnel have now been hired – the IT Manager will start in April and the Project Physician and Regulatory Affairs Manager in May. Questions regarding clinical studies and GCP can now be directed to the work package members. A workshop covering the methodologies in clinical trials is planned for the end of the year.

##### WP06.2 – Integrating tools and methods for SMA (Enrico Bertini, WP Leader)

Working with the TREAT-NMD group on outcome measures, we will obtain a consensus on the evaluation of SMA patients presenting with varying functional severity of disease. The validation of the scale for evaluating patients with SMA type I is underway, and evaluation protocols for SMA types II and III will be discussed at the ENMC meeting in May 2007.

Involved in preparing and activating the Parent/Caregiver Survey, which has been translated into several European languages, and a version for the 'Survey Monkey' web site has also been created.

##### WP09.1 – Assessment tools and outcome measures (Eugenio Mercuri, WP Leader)

A review has been undertaken into the principal outcome measures available that have been used in recent studies in the neuromuscular field. In validating functional scales we have focused mainly on spinal muscular atrophy (SMA) and Duchenne muscular dystrophy (DMD). Regarding the tools for SMA 1 we are finalizing the validation of the CHOP-INTEND functional scale and we are assessing its reliability in SMA 1 and in normal children and its responsiveness to clinical changes. Regarding the tools for SMA 2, we are comparing the responsiveness of the adapted Hammersmith Functional Motor Scale and of the original Hammersmith Functional Motor Scale (HFMS) (Main et al. 2003) in patients with different clinical severity. Regarding strong SMA 2 and SMA 3 patients, there is the need to develop a functional scale without the ceiling effect of the HFMS and to add other outcome measures (myometry, timed walking etc). This aspect is under debate and will be discussed at the ENMC meeting in May 2007. We have also worked on development and validation of "upper limbs module" to be added to other scales, such as the HFMS, to better assess the functional changes in upper limbs in children with neuromuscular disorders. At the moment we are using this module in SMA 2 and 3 and DMD patients. Regarding DMD, the Hammersmith functional scale by Scott et al. (1982) and the North Star scale have been evaluated and data of both scales in patients with different severity have been collected.

There is a lack of tools to assess paediatric neuromuscular patient' quality of life (QoL). The most promising tools seem to be the "PedsQL neuromuscular model" questionnaire, which is currently in the validation process in the US. To spread the use of this tool in Europe a cross-language validation is required, and the research fellow from this work package has recently attended a meeting in Boston with the US group to agree on the methodology to be used.

#### WP10.1 – Harmonise and Develop Patients Diagnosis and Care (Thomas Sejersen, WP Leader)

TREAT-NMD participation has been initiated in work by the Centre for Disease Control (CDC) to develop standards of diagnosis and standards of care for DMD. An agreement has been reached to aim for consensus documents to be common for CDC and TREAT-NMD. Expert panel meetings are planned for June 2007 in Atlanta, Georgia, with five TREAT-NMD representatives in attendance.

The International Standard of Care Committee for SMA (SCC) has initiated work on developing a consensus document on standards of care, with participation from several members of the TREAT-NMD network, and under the European Chairmanship of Enrico Bertini. This document will be published shortly. A TREAT-NMD steering group on standards of diagnosis and care has been established and will produce guidelines applicable to Europe, based on the SCC document, following publication. An email discussion group has been established and the SCC document will be discussed with SCC representative Ching Wang at a meeting (arranged by ENMC) on May 11-13.

#### WP07.2 – Select Appropriate Mammalian Models (Markus Ruegg, WP Leader)

To help with this WP Dr. Raffaella Willmann ([raffaella.willmann@unibas.ch](mailto:raffaella.willmann@unibas.ch)) has been recruited to interface with the partners to assess current animal models, compile a critical review of these models and reach consensus on which models will form the basis for further study. A drafting of a first report is now underway and to facilitate discussions among the partners an e-mail account has been created so all interested parties can send their suggestions and opinions. Please send your comments on this WP to [animalmodels-bioz@unibas.ch](mailto:animalmodels-bioz@unibas.ch)

In order to assist Raffaella with compiling this information it is important for us to assess the know-how around the network, and therefore I urge you to send your comments and suggestions on your experiences with animal models to the email address above. A first meeting with the partners of WP07.2 will take place on Thursday May 3, 2007 at the Biozentrum in Basel. Raffaella will forward further details on the meeting soon.

Can we please request that you send your work package updates to [stephen.lynn@newcastle.ac.uk](mailto:stephen.lynn@newcastle.ac.uk). Thank you to those of you who have already supplied updates. The 'highlights' of the results will be included in future newsletters.

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#### **6. Questions to be included in 'Fact finding Questionnaire' – deadline 29 March 2007**

Emma Heslop and Rachel Thompson contacted you recently to ask partners who need to gather information from other partners as part of their work-package responsibilities if they would like to do this via our general Fact Finding Questionnaire. An example of a question might be 'what NMD workshops and conferences are currently available in your institution / country'. Thank-you to those partners who have responded but we would like to encourage all partners to think about the information they are required to collect as part of their work-packages and formulate questions to e-mail to [emma.heslop@newcastle.ac.uk](mailto:emma.heslop@newcastle.ac.uk) or [r.h.thompson@newcastle.ac.uk](mailto:r.h.thompson@newcastle.ac.uk). The questions will then be included in the Fact Finding Questionnaire and distributed to all partners. Results will be made available to all partners upon completion. This will ultimately speed up and streamline the data collection, helping us all to achieve our deliverable deadlines. If you would like to have questions included in the questionnaire, please email them to Emma or Rachel by the 29 March.

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## 7. Latest News / Research

149th ENMC International Workshop and 1st TREAT-NMD Workshop on  
"Planning Phase I/II Clinical Trials Using Systemically delivered Antisense Oligonucleotides in Duchenne  
Muscular Dystrophy"

The workshop was organized with the support of ENMC, the TREAT-NMD EU Network of Excellence and  
PPMD on the 23rd – 25th February 2007, Naarden, The Netherlands.

Please click on the link below to view the Workshop Lay statement.

<http://www.treat-nmd.eu/assets/documents/lay-statement-AON-007.pdf>

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## 8. Conference Report

### THE TREAT-NMD INITIATIVE PRESENTED AT THE ITALIAN TELETHON CONVENTION

Report by Anna Ambrosini, Telethon

Last week, the TREAT-NMD initiative was presented by Prof. Bushby at the Italian Telethon Scientific  
Convention.

The meeting, which gathered all Telethon-funded scientists with an ongoing project on genetic disease and  
their collaborators – approximately 700 participants, was held in Salsomaggiore Terme (Parma, Italy), 12<sup>th</sup>-  
14<sup>th</sup> March. Prof. Bushby was invited to chair a workshop dedicated to the most innovative therapeutic  
strategies for muscular dystrophies. In her lecture, she highlighted advances and problems in translational  
research in this field, and presented the TREAT-NMD programme as a possible means of advancing preclinical  
research towards a clinical application.

This was an excellent opportunity to share once again the scopes of the TREAT-NMD project with the Italian  
neuromuscular research community, in order to strengthen the interest of those already involved as clinical  
network under the leadership of Telethon, and to increase the level of awareness in basic scientists working in  
this field.

Prof. Bushby's overview of the bottlenecks which arise while progressing along the pathway from lab to clinic  
was also of general interest to investigators working on other diseases which have genetic origins and rarity in  
common, since it clarified and discussed common problems faced by all of them.

Once more, it appeared clear how challenging the TREAT-NMD initiative will be and how relevant the impact  
of an international platform for clinical studies in NMD can be. This should encourage us all to work harder  
towards reaching the goals of this programme.

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## 9. Calls for Papers / Posters



**12th International WMS Congress, Sicily, 17-20 October 2007**

### Call for Abstracts – Deadline is 31 March 2007

Please remember to submit your abstracts for the upcoming WMS conference before 31 March 2007. If you  
submit an abstract for this conference, please let us know – we hope that TREAT-NMD will be well  
represented.

<http://www.worldmusclesociety.org/>

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## 10. Articles

TREAT NMD in Muscle news from around the world by Prof. Kate M.D. Bushby and Prof. Volker Straub  
Published in Neuromuscular Disorders 17 (2007) 269-271

This article, which serves as an introduction to TREAT-NMD, is now available online via the following DOI link:  
<http://dx.doi.org/10.1016/j.nmd.2007.02.002>

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## 11. Calls for Proposals

### DG SANCO launches a new Call for Proposals 2007

As part of its Programme of Community Action in the field of Public Health (2003-2008), DG SANCO has launched a new **Call for Proposals** (see [http://ec.europa.eu/phea/calls/call\\_for\\_proposals\\_en.html](http://ec.europa.eu/phea/calls/call_for_proposals_en.html)). Following the adoption of the Public Health Programme's 2007 **Work Plan** earlier the same month, the Public Health Executive Agency published a Call for Proposals on 16 February 2007. The programme is based on 3 main objectives: health information, health risks and health determinants.

Submissions deadline: 21 May 2007

Thereafter the Public Health Executive Agency (PHEA) will undertake the evaluation of the submitted proposals. The selection procedure is expected to last until autumn 2007. No information on the evaluation of projects can be provided during the selection procedure.

This year, the call will be focussed on certain key actions taken on previously and on several new fields not previously covered by submitted proposals made following earlier calls. The projects to be financed should be innovatory and should not take place over a period of more than 3 years.

With regard to rare diseases, projects should be concentrated on the following aspects:

- Development of strategies and mechanisms for exchange of information between patients with a rare disease and action to help improve epidemiological studies, codification, classification and definitions
- Support to European network centres of reference for rare diseases aiming at developing recommendations of best practice in terms of treatment, sharing of information on diseases and performance evaluation.

The total amount of funds given to this call for proposals is estimated at €33 800 000. Given the complementary and motivating effects of Community funding, at least 40% of project costs must be funded by other sources.

### First trans-national Call for Proposals for European research projects on rare diseases

The ERA-Net for research programmes on rare diseases (E-Rare) has been established to coordinate the research efforts of European countries in the field of rare diseases. Ten partners are taking part in this project which is coordinated by the French Institute for Research on Rare diseases/INSERM. The six E-Rare partners include the National Research Agency (ANR), France; the Federal Ministry of Education and Research (BMBF), Germany; the National Institute of Health (ISS), Italy; the Chief Scientist Office of the Ministry of Health (CSO/MOH), Israel; the Institute of Health Carlos III (ISCIII), Spain; the Scientific and Technological Research Council of Turkey (TÜBİTAK), Turkey. These partners have decided to open the first joint Call for funding multilateral trans-national research projects on rare diseases.

The aim of the Call is to enable scientists in different countries to build an effective collaboration on a common research project based on complementarities and the sharing of expertise. The projects shall involve a group of rare diseases or a single rare disease according to the European definition i.e. a disease affecting not more than 5:10,000 persons in the EU.

Further information can be obtained from the [E-Rare web site](#)

### **New Calls for Proposals at the Brocher Foundation**

The **Brocher Foundation** is calling for scientific projects to be undertaken during 1-6 month stays in Geneva where it is based. It is inviting submissions from researchers on the following topics:

- Ethical, legal and social issues relating to genetics
- Ethical, legal and social issues relating to biotechnologies, biomedical engineering or nanomedicine
- Health law, healthcare economy, philosophical approaches to healthcare or biotechnologies.

Deadline: 15 May 2007

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### **12. Upcoming Conferences**

#### **EURORDIS 10th Anniversary, Annual Membership Meeting & Workshop: gaining access to rare disease research resources**

Date: 4-5 May 2007

Venue: Institut Pasteur, Paris, France

[http://www.eurordis.org/IMG/pdf/EN\\_programme\\_AMM\\_Paris07.pdf](http://www.eurordis.org/IMG/pdf/EN_programme_AMM_Paris07.pdf)

#### **European Conference on Rare Disease Research**

Date: 13 September 2007

Venue: Charlemagne building- Brussels, Belgium

#### **European Human Genetics Conference 2007**

Date: 16-19 June 2007

Venue: Nice, France

<http://www.orpha.net/actor/EuropaNews/2007/doc/epposi.pdf>

#### **4th European Conference on Rare Diseases (ECRD 2007)**

Date: 27-28 November 2007

Venue: Lisbon, Portugal

[http://www.eurordis.org/article.php3?id\\_article=1351](http://www.eurordis.org/article.php3?id_article=1351)

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