



# TREAT-NMD

## Neuromuscular Network



16<sup>th</sup> November 2007 · Newsletter No. 21

Welcome to the latest newsletter from the TREAT-NMD network. This edition features a meeting report from last week's patient registry curators' meeting in Montpellier, plus a number of calls for participants in our activities – see page 3.

Please forward any items that you would like to be included in future editions of the newsletter to [info@treat-nmd.eu](mailto:info@treat-nmd.eu).

Best wishes,

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

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### About this newsletter

This is a fortnightly newsletter sent to all members of TREAT-NMD's "Club of Interest" worldwide. Earlier editions of the newsletter can be found online at [www.treat-nmd.eu/news/newsletter/index.htm](http://www.treat-nmd.eu/news/newsletter/index.htm). If you would like to subscribe directly, please visit our website at [www.treat-nmd.eu/](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.

### 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](mailto:info@treat-nmd.eu). The coordination team in Newcastle will be happy to put you in touch with the person most relevant to your particular interest.

## Report from the TREAT-NMD Registry Curators Training Course

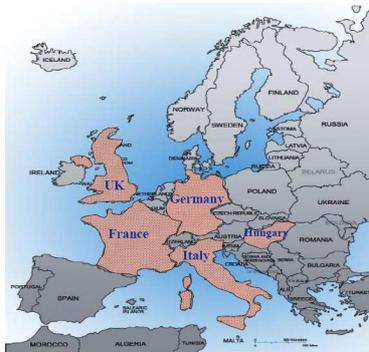
7-9 November 2007, Montpellier, France

The TREAT-NMD patient registries project has been receiving an enormous level of interest and support from around the world. It is clearly recognised that creating a reliable source of information on patients with neuromuscular disorders will speed up the process of locating patients suitable for clinical trials as well as providing other useful information about the prevalence of disorders and allowing patients to be kept informed about the latest research of relevance to their condition. Registries for DMD and SMA are already well underway, while those for other disorders will follow in the coming months and years.



Last week over 50 delegates from 16 countries travelled to Montpellier in the south of France to receive in-depth information on registries for SMA and DMD and gain hands-on training with the software that will be used to run the TREAT-NMD Global Patient Registry. Since TREAT-NMD started in January 2007, the will to move forward with a harmonised registry has been extraordinary and plans are in place for international collaboration involving up to 19 different existing or planned registries. TREAT-NMD activity leader Hanns Lochmüller gave an overview of the registry and its objectives, as well as discussing the Registry Charter that describes both the legal and ethical conduct of users, access rules to the database as well as third party involvement. This was followed by an excellent presentation from Sylvie Tuffery-Giraud on the genetic items in the characterisation of the various mutations. A handbook describing these items will be drafted and made available through the TREAT-NMD website and a forum will be opened for direct discussion of the details of patient registration to support the individual curators.

Planned TREAT-NMD registries on DMD and SMA (2006)



Véronique Humbertclaude then described the mandatory and highly encouraged clinical items that would be included in the global registry. These items have been generated by a process of consensus and this in itself should make the registry a valuable resource for future clinical trials. Christophe Bérout then gave a practical demonstration of the ease with which mutation information can be entered using the UMD software into the registry, as demonstrated by the success of both genetic and non-genetic specialists who entered test data. This was a very interactive session and the support of the Montpellier team was much appreciated.

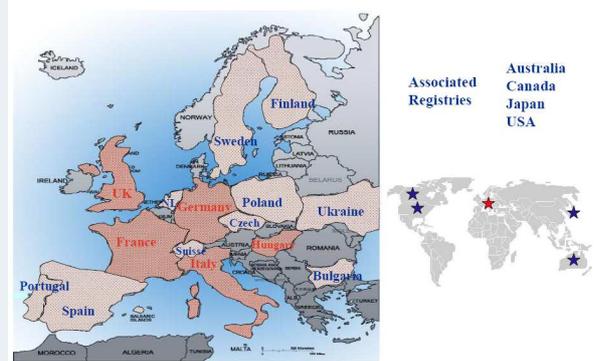
The second day gave participants an opportunity to hear from the various national registries, not only in Europe, but from around the world (see maps), and to discuss how each of these registries can interface with the TREAT-NMD Global Registry.

This was a very successful and well-planned meeting and we must congratulate our colleagues at INSERM for their efforts. The TREAT-NMD Global Registry will be launched at the beginning of 2008, and future training courses will be arranged to ensure that we work with all national registry efforts to deliver a single point of contact for industry as they plan and develop future clinical trials in neuromuscular diseases.

All the presentations from this training meeting are now available on the TREAT-NMD web site along with other relevant information via the link below.

<http://www.treat-nmd.eu/registries.htm>

TREAT-NMD registries on SMA and DMD (Nov 2007)



### Call for participants in DMD care standards discussion

The process of drawing up a comprehensive set of recommendations for standards of care in DMD on the basis of true international consensus is ongoing, under the auspices of the CDC Care Considerations project, and is likely to be complete late in 2008. In the intervening period, TREAT-NMD is working to get as much of the existing information as possible summarised and out into the public domain. A group of experts, including co-authors of existing DMD management guidelines, is being invited to collaborate with the TREAT-NMD activity leaders with the aim of collecting and comparing existing management recommendations and creating brief and practical consensus statements that will be useful in their own right and also able to feed into the ongoing CDC discussion.



If you are interested in contributing to this discussion, please write to TREAT-NMD activity leader Thomas Sejersen at [thomas.sejersen@ki.se](mailto:thomas.sejersen@ki.se), stating which of the following discussions you would like to be involved in:

- Diagnosis
- Cardiovascular care
- Orthopaedic/Surgical care
- Psychosocial care
- GI/Nutritional care
- Neurological care
- Respiratory care
- Rehabilitation management
- Oral care

### Upper limb assessments for DMD – request for input from occupational therapists

Clinical trials of new therapies naturally require validated methods of assessing treatment outcomes. There is some concern that non-ambulatory boys will be excluded from participating in upcoming DMD trials if reliable upper limb assessments are not agreed on.

Developing appropriate scales for upper limb assessment requires input from occupational therapists, and TREAT-NMD is very interested in hearing from OTs and others with an interest in this area.

If you would like to participate in this discussion, please contact the TREAT-NMD coordination office at [info@treat-nmd.eu](mailto:info@treat-nmd.eu)

### Request for information about care costs for DMD

Some of TREAT-NMD's industrial partners are interested in establishing costs for the burden of care for DMD patients. This is something that needs to be identified both from a government lobbying point of view and also to convince commercial organisations that there is benefit to the health care system to generate medicines for DMD and thus reduce these hidden costs. The usual figures needed for this kind of cost evaluation are average total healthcare costs per patient (diagnosis, follow on visits to consultants, prescriptions, physio, wheelchair costs, intensive care time, spinal surgery, ankle surgery, respiratory intervention, pneumonia treatment) and the hidden costs such as loss of parental income, loss of income to society due to premature death, cost of carers, preparation for education. If anyone has all or some these figures for respective countries could they forward them on to the TREAT-NMD office ([info@treat-nmd.eu](mailto:info@treat-nmd.eu)) for dissemination and future discussion.

### AFM looking for volunteers to appear in the Telethon



The AFM has decided to organise an event for this year's Telethon to highlight the pan-European fight to deliver treatments for NMD. This will take place in Metz on 7th and 8th December and will consist of workshops, meetings and a round table with a public audience in a "European Gene Café" in the afternoon of 8th December.

It would be fantastic to have TREAT-NMD partners and other people interested in the project taking part in this event especially those from Germany, Luxembourg, Belgium, the Netherlands, Switzerland and the UK.

The objective is to involve a minimum of 30 members from European partners participating in this event which could be the opportunity to communicate through different media (press, radio, TV) first in France, but in other countries involved too.

If you are interested please contact Hervé Laouenan who is coordinating the event.

[hlaouenan@afm.genethon.fr](mailto:hlaouenan@afm.genethon.fr)

## Meetings

### Open Forum on the Future of European Networks of Excellence

Brussels, Belgium

20th November, 2007

A group of 56 EU-funded Networks of Excellence, including TREAT-NMD, has drafted an opinion paper calling for action from the European Commission to secure the long-term future of the most successful integrated networks. The signatories believe that Networks of Excellence face an uncertain future, and urge the Commission to take four actions to ensure their long-term sustainability. The proposed actions address the Commission's commitment to Networks of Excellence, funding, a review of the Networks and further exploitation of the networks.

One of the main causes for concern relate to a reduction of funding for Networks of Excellence and difficulties in setting up a permanent legal structure. The Commission has responded by saying that it is looking to re-examine how Networks of Excellence can be used 'as an instrument to take a real step forward in the European Research Area'. An independent review of the current Networks of Excellence is due to take place soon.

TREAT-NMD is already planning to ensure the future sustainability of the network, and will work closely with partners and stakeholders to establish a lasting and durable entity.

### European Conference on Rare Diseases

Lisbon, Portugal

27-28th November, 2007

The TREAT-NMD Neuromuscular Network will be presented at this meeting, which is an opportunity for patients and health care professionals to review policies, strategies and examples of successful action among the rare disease community in Europe. The conference is sponsored by the European Commission through the DG Public Health and organised by Eurordis, who are also a partner in TREAT-NMD.

### Launch of DuchenneConnect—a resource for the Duchenne/Becker community

Parent Project Muscular Dystrophy (PPMD) has launched a new resource website and registry, DuchenneConnect, that aims to provide resources to help with early, appropriate, and least invasive diagnosis, understand benefits and limitations of genetic testing and access resources and services to include care and treatment.

Representatives from DuchenneConnect attended the TREAT-NMD Registry Curator Training Course in Montpellier where they described their registry and discussed ways in which they can collaborate with the TREAT-NMD Global Patient Registry. TREAT-NMD are already working closely with PPMD, and other patient organisations, and we are committed to harmonising our efforts with national registries to ensure that all patients are represented in the TREAT-NMD Global Patient Registry.

Further information on DuchenneConnect can be found on their web site.

<http://www.duchenneconnect.org>

### Send us your news and views!

We strongly 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](mailto:emma.heslop@treat-nmd.eu)



## The TREAT-NMD Neuromuscular Network