

welcome

Welcome to the **last TREAT-NMD newsletter of 2008!** This has been an exciting year, and the highlight of this edition is undoubtedly the "Christmas Message" from our coordinators, but we also have a conference report from Chile, the launch of the Finnish DMD and SMA patient registries, and news of a CMD therapeutic targets conference in 2009.

As always, 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. Back-issues of this newsletter can be found on our website at <http://www.treat-nmd.eu/patients/news/ezine-archive/>

All the best for 2009!

Katie, Volker, Steve, Emma, Rachel and Samantha: the Newcastle TREAT-NMD team

at a glance...

06-09 Jan 2009 [Workshop: The Multiple Faces of Lamins in Aging and Disease](#)

02-04 Feb 2009 [TREAT-NMD Governing Board meeting](#)

28 Feb 2009 - 01 Mar 2009 [First Asian conference on Duchenne muscular dystrophy \(DMD\)](#)

26-27 Mar 2009 [UK Neuromuscular Translational Research Conference](#)

12-15 May 2009 [The Nottingham Systematic Review Course 2009](#)

21-23 May 2009 [International conference in Ukraine: Recent standards in diagnosis, treatment and medical care for some rare neuromuscular diseases](#)

09-11 Jul 2009 ["Therapeutic Targets in CMD", Emory University, Atlanta, Georgia](#)



update

A Christmas message from the coordinators

This has been an exciting year for TREAT-NMD, one that has seen the network take tremendous steps forward in its aim of readying the neuromuscular field for clinical trials. We would like to take this opportunity to thank you all for your support and for the effort that so many of you have put into our work this year, and also to take a step back and have a look at a few of the successes that we have achieved together and our plans for 2009.



Patient registries

When TREAT-NMD was launched last year, only five national patient registries for DMD and SMA were planned. But thanks to the tremendous efforts, commitment and resources from colleagues and particularly patient organisations across the world, we now have 12 [national registries for DMD and 10 for SMA](#) already running, with a further 12 expected to come online in the near future. In addition to this we have held a [workshop to determine future strategies for other global registries for rare neuromuscular diseases](#), supported a registry for myotubular myopathy, and linked up with an initiative for a [registry for all CMDs](#). 2009 should see the launch of a number of additional registries for other neuromuscular conditions.

The importance of these registries for clinical trials cannot be underestimated. Having a single source of locatable patients with identified genetic mutations will save researchers and the pharmaceutical industry valuable time and resources in trial planning and when recruiting patients for clinical trials, and thus in the end make sure treatments are approved more quickly.

Network of clinical trial and patient care sites

As well as finding the patients for trials, TREAT-NMD has also set up a centralised source of information about [clinical sites with the expertise to host a clinical trial](#). When a researcher or pharmaceutical company is planning a trial, this registry of over 100 trial sites worldwide means they have all the data they need at their fingertips. In addition, it is a valuable tool for keeping all clinicians and patients informed about best practice in care standards.

Regulatory links

Regulatory approval is required before any clinical trial can take place, and it was encouraging to see the success of our harmonized [approach to the EMEA regarding outcome measures for SMA trials in October](#). A second meeting focusing on DMD trials is planned for 2009. Together with the [regulatory affairs database](#), this initiative helps to map the regulatory environment to ensure that all the regulatory requirements for upcoming trials can be more easily met.

Research tools

2009 also saw the publication of [standard operating procedures for work on animal models of disease](#), a major accomplishment involving close collaboration with the US Wellstone Institute. The [Registry of Outcome measures](#) is another key tool establishing the measures that are used to test treatment efficacy in neuromuscular conditions. A [publication on harmonisation of outcome measures for DMD and SMA](#) is another major achievement of this year.

Standards of care

The publication in 2007 of an international consensus on [SMA care standards](#) was well received by patients and clinicians as a guide to best practice. It will be followed early in 2009 by the publication of DMD standards, and TREAT-NMD is already working with patient groups on "patient-friendly" versions of these guidelines and their global dissemination.

Clinical trials platform

In addition to the very real chance of industry-led trials in the coming years, TREAT-NMD is also involved in other exciting developments regarding the setting up of a trials platform to fast-track

09-12 Sep 2009 IDMC-7
International Myotonic Dystrophy
Consortium

17-19 Nov 2009 TREAT-NMD / NIH
International Conference

efficacy studies for drugs that already have marketing authorisation for other indications, creating a rolling schedule of rapid small-scale utility trials to determine whether particular treatments warrant further development.

All of this is evidence that over the past year TREAT-NMD has developed into a major player in the neuromuscular field, one that will make a real difference to the way trials in neuromuscular diseases will be carried out in future. As coordinators we have been delighted to see the way so many supporters worldwide have already become so closely involved with our activities, and we look forward to developing further links in 2009.

It only remains for us to thank all our collaborators once again for their support, and to wish you a happy holiday season and all the best in 2009.

Katie and Volker

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Finnish patient registries launched

Another country in the [TREAT-NMD patient registries initiative](#) came online in November 2008 as the Finnish SMA and DMD Patient Registry was officially launched and patient registration began. The Registry is owned and maintained by the patient organization Lihastautiliitto ry (The Finnish Neuromuscular Disorders Association) in association with TREAT-NMD and is situated in the city of Turku in south-west Finland. All five university hospitals of Finland have participated in setting up this registry, the curators of which are Sinikka Loukamaa, nurse and diagnosis advisor, working at Lihastautiliitto, and Jaana Lähdetie, child neurologist, M.D., Ph.D., working at Turku University Central Hospital.



Patients have access to the informed consent and registration forms via the internet either through the TREAT-NMD website at <http://www.treat-nmd.eu/patients/patient-registries/fi-dmdsma> or directly from the Finnish patient organization website at <http://www.lihastautiliitto.fi/index.php?mid=252> or by telephone (int-358-2-2739730). The registry can provide information and forms to both Finnish and Swedish-speaking patients in Finland. The forms can be emailed and subsequently printed out by the patient or sent to his or her home address by mail. The signed forms should be mailed back to Lihastautiliitto where the data will be stored securely and filtered to be further transferred anonymously to the international TREAT-NMD Patient Registry.

Finnish doctors and nurses involved in the care of neuromuscular patients will be informed about the registry and registry flyers will be distributed to various hospitals in Finland in 2009. The voluntary basis of registration is stressed by collecting the registry data from patients themselves. If they give their permission, the curators will contact genetic laboratories to enquire details about genetic testing and/or muscle biopsies.

Patient registries are a key element of TREAT-NMD's work to accelerate clinical trials. By storing all the relevant information in one place, they make it easier for patients suitable for specific trials to be found and contacted quickly. Pharmaceutical companies have already shown interest in the patient registry, and are using it to obtain information regarding the feasibility and planning of multi-national clinical trials.

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EVELAM 2008 - a report from Latin America

The first Euro-Latinoamerican School of Myology (known as EVELAM for its acronym in Spanish) was held in Santiago, Chile, from December 11th to 13th. This initiative, designed as an "overseas" extension of the Summer School of Myology of Paris, was conceived by Dr. J. Andoni Urtizberea (Hendaye, France), Dr. Alberto L. Rosa (Cordoba, Argentina), Norma B. Romero (Paris, France) and Prof. Jorge A. Bevilacqua (University of Chile).



The meeting also attracted the presence of a select group of neuromuscular experts from Europe and Latin America sponsored by the Association Française contre les Myopathies (AFM); the Federación Española de Enfermedades Neuromusculares (ASEM); TREAT-NMD and Teletón (Chile). The experts lectured on the wide spectrum of neuromuscular disorders ranging from basic knowledge to diagnosis and therapy of inherited myopathies, neuropathies and motor neuron diseases, including presentation and discussion of selected clinical cases and histopathological workshops. About 120 participants from all over Latin America including Mexico, Brazil, Uruguay, Ecuador, Venezuela, Argentina and Chile joined in the enthusiastic and friendly atmosphere in Santiago. The meeting served not only as a means to provide an update on the advances in Inherited Neuromuscular Disorders but also to start creating a network in Spanish-speaking Latin American centres to link with other existing international networks working for consensus in diagnosis, standard of care and therapeutic strategies.

The seminar opened with an introduction to the study of neuromuscular patients (Dr. Ricardo Erazo, Santiago Chile) followed by a comprehensive lecture on normal muscle structure (Mariana Casas, PhD, Institute of Medical Biosciences, Chile). Prof. Ana Lia Taratuto (Buenos Aires, Argentina)

discussed the general approach to a muscle biopsy. Dr. Ricardo Fadic (Universidad Católica de Chile) and Luis Gracia, MD, from the Institut de Myologie, Paris, covered basic, clinical and therapeutic aspects of dystrophinopathies. In the afternoon, Dr. Isabel Ila (Barcelona, Spain) disserted on Inflammatory myopathies and Dr. Alberto Rosa (Cordoba, Argentina) made an update on FHSD.

On day two, Prof. Enrico Bertini (Rome, Italy) lectured on congenital muscular dystrophies and Isabel Ila and Alberto Rosa summarized autosomal recessive and dominant limb girdle muscular dystrophies, emphasizing the most prevalent LGMD in the region. The afternoon was dedicated to motor neuron disease. Prof. Robert Guilloff from Imperial College London provided an update on current knowledge of ALS including the ongoing therapeutic trials, and Dr. Claudia Castiglioni (Santiago, Chile) continued with spinal muscular atrophies (SMA), while Susana Quijano-Roy, (Paris, France) gave an update on Emery-Dreifuss dystrophy and laminopathies and finally, Prof. Mario Medici from Montevideo, Uruguay, reviewed oculopharyngeal muscular dystrophy.

Day three focused on myasthenic syndromes, with Dr. Gabriel Cea, University of Chile, and Dr. Cristina Vasquez, Montevideo Uruguay, presenting on hereditary neuropathies. The conference was closed by three lectures on therapeutics and multicentre clinical trials in SMA given by Prof. Enrico Bertini of TREAT-NMD; gene-based therapy lectured by Luis Garcia and physical therapy of neuromuscular patients discussed by Susana Quijano-Roy.

The organizers were eager to join the TREAT-NMD network, particularly in participating in the DMD and SMA patient registries initiative.

Based on the success of the imitative, EVELAM 2009 is already scheduled and is to be held in Montevideo, Uruguay. Don't miss it!

Report by Jorge A. Bevilacqua and Enrico Bertini

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Therapeutic Targets in CMD - conference in July 2009

Principal investigator: Dr. Carsten Bonnemann
Co-organizers: Dr. Anne Rutkowski and Patrick May, Cure CMD



This unique conference will identify and initiate opportunities for translational research in the congenital muscular dystrophies (CMDs). The primary goal of the conference is to address, evaluate and achieve consensus on therapeutic targets in the CMDs by bringing together experts ranging from basic science to clinical trial design in rare disorders. A successful outcome will lead to the construction of a roadmap and investment "portfolio" based on the identification of short term, mid term and long range scientific targets and their subsequent development into treatment strategies with strong translational potential.

Based on the current understanding of possible therapeutic targets and conceivable therapeutic strategies, topics will be grouped into the following core areas: apoptosis (mechanism and control), atrophy, endurance/exercise pathways, fibrosis and extracellular matrix control, inflammatory mediators, virally mediated gene delivery and mutation directed therapy, upregulation of glycosylation, and stem and satellite cell based therapy (replacement therapy and pharmacologic manipulation of regenerative and proliferative potential). Strengths and weaknesses of current clinical and basic science resources will be identified, including: available animal models, assays, patient registries, biorepositories and existing clinical trial networks. Expert clinicians with participation from an FDA representative will guide discussion germane to clinical trial design options in rare disease to develop and adapt strategies for efficient implementation of smaller clinical trials as well as open pilot trials. Related to this, an additional core area will focus on the development of outcome measures, biomarkers and the development of additional tools such as quality of life measurements.

Select pharmaceutical company participation will bridge current basic science research to industry based pharmaceutical development. MDA, NIH and TREAT-NMD representation will help broaden the perspective and synergize collaborative networks for future funding requests focused on CMD Translational Targets.

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