



TREAT-NMD

Neuromuscular Network

2nd May 2008 · Newsletter No. 32



Welcome to the latest newsletter. This edition features information on six TREAT-NMD fellowships to the WMS 2008 teaching course in Newcastle; a focus on our partners in Hungary, the National Institute of Environmental Health; details of Santhera's upcoming Phase II clinical trial in Ullrich CMD; and information on this autumn's European Research Conference in Paediatric Neurology.

Please forward any items that you would like to be included in future editions to info@treat-nmd.eu.

Best wishes,

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

*****IMPORTANT MESSAGE TO ALL OUR READERS*****

We'd like to remind readers that receiving this newsletter does not automatically make you a member of TREAT-NMD. If you are interested in developing closer links with us, we encourage you to read our Members' Charter and complete the membership application form. These documents can be found on our web site at the address below.

Become a Member of TREAT-NMD

If you are interested in becoming a member of the TREAT-NMD Network please visit our web site to download our membership charter. An application form is also available for download. The web link to our Members' section is: http://www.treat-nmd.eu/news/item/?members_charter We look forward to welcoming new members!

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. If you would like to subscribe directly, please visit our website at 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. The coordination team in Newcastle will be happy to put you in touch with the person most relevant to your particular interest.

Newsletter contents

About this newsletter	1
Working with us.....	1
TREAT-NMD news and reports.....	2
Partner focus	3
Meetings and workshops.....	5
Other news and reports.....	6

CTCC Workshop

Clinical Trials in Neuromuscular Diseases

19th-21st June 2008

The TREAT-NMD CTCC team cordially invites you to come to Freiburg in June 2008 to participate in the first TREAT-NMD *Clinical Trials in Neuromuscular Diseases* Workshop

In this workshop, focused on clinical trial development in the field of neuromuscular diseases, participants will learn how to develop a trial protocol from scratch. Lectures on study design, biostatistics, ICH-GCP and European regulatory requirements will provide fundamental information.

Ideally, participants will attend the workshop with a solid idea of a planned study. During hands-on sessions there will be time to develop a protocol synopsis and members of the TREAT-NMD CTCC and other experts will assist participants in all aspects of protocol development.

The workshop offers an outstanding opportunity to meet other researchers and discuss trial projects and perspectives.



For more information:

CTCC Workshop Programme: http://www.treat-nmd.eu/assets/documents/Workshop-schedule_o80303.pdf

Flyer and registration form: http://www.treat-nmd.eu/assets/documents/Flyer_o80229.pdf

For further information or to express an interest please contact: annette.pohl@uniklinik-freiburg.de

WMS 2008 Teaching Course Fellowships supported by TREAT- NMD

TREAT-NMD is offering 6 fellowships of €500 each to Central and Eastern European (CEE) delegates to attend the 6th WMS Satellite Teaching Course from 28-29th September 2008. The course is intended to improve the diagnostic competence of professionals dealing with patients with neuromuscular diseases and will be held immediately prior to the opening of the 13th International WMS Congress in Newcastle Gateshead, UK (29th September—2nd October).

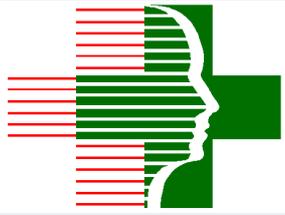
The course is intended to improve the diagnostic competence of professionals dealing with patients with neuromuscular diseases and will be held immediately prior to the opening of the 13th International WMS Congress in Newcastle Gateshead, UK (29th September—2nd October).

<http://www.wms2008.com/training.htm>

WMS
World Muscle Society

13th International WMS Congress
29th September - 2nd October 2008
Newcastle Gateshead, UK

A multidisciplinary scientific society dedicated to advancement and dissemination of knowledge in the neuromuscular field for the benefit of patients



Focus on the National Institute of Environmental Health (NIEH)

P.O. Box 1437 Budapest 839, 2-6. Gyali Street, 1097 Budapest, Hungary

The Department of Molecular Genetics and Diagnostics at the National Institute of Environmental Health in Hungary specialises in the DNA diagnosis of neuromuscular disorders. The laboratory was established in 1993 and performs diagnostic activities as the only centre in Hungary for spinal muscular atrophy, Charcot-Marie-Tooth type 1A, facioscapulohumeral dystrophy, myotonic dystrophy type 1, congenital myasthenic syndrome and fragile X(A) syndrome. Although Duchenne/Becker muscular dystrophy is also diagnosed in other centres in Hungary, this laboratory is the only one which provides deletion/duplication screening for the entire dystrophin gene, therefore most of the families are sent here for diagnosis, including prenatal cases. SMA families are exclusively sent to NIEH for diagnosis, carrier analysis and prenatal diagnosis. DNA and muscle samples of patients with LGMD and CMD are also collected and protein and DNA analyses of the patients are performed by international collaboration.

The department maintains a DNA bank and a database of clinical symptoms and identified/unidentified mutations of 2729 patients/family members with neuromuscular diseases which has been in operation since 1993. It participates in the EUROBIOBANK project funded by FP5. It has a nationwide collaboration with paediatric neurologists, neurologists, genetic counsellors of clinics and hospitals in Hungary, as well as a long-term international scientific collaboration with LMU Munich Dept. of Neurology (Germany) and with departments of Human Genetics of the Universities of Turku (Finland) and Leiden (The Netherlands). From time to time, diagnostic services are also provided mainly in SMA, DMD/BMD and FSHD patients from other Eastern European countries, like Rumania, Bosnia, Croatia, Bulgaria.

NIEH is involved in Activity 04 (Databases and Biobanks). The main objective of NIEH involvement in WP 04.2 is to create and maintain a TREAT-NMD Patient Registry for Spinal Muscular Atrophy and Duchenne/Becker Muscular Dystrophy in the Eastern European countries. The existence of national patient registries is crucial for future therapeutic trials.

People involved in WP4.2

Dr. Veronika Karcagi, PhD is Head of the Molecular Genetics and Diagnostics Department at the National Institute of Environmental Health in Budapest. She graduated as a biologist MSc. at the Eötvös Lorand University, Budapest in 1975. In 1996 she received her PhD degree in neurobiology and in 1998 her certification in human genetics. She acts as the national coordinator of EMQN for Hungary. She is member of the Hungarian National Standardization Committee of Clinical Geneticists since 2004. She is also a member of the executive committee of the Hungarian Society of Clinical Neurogenetics since 1997 and is the general secretary of the Hungarian Society of Human Genetics since 2004. In relation to this project, she works as a Database Curator for TREAT-NMD for Eastern European Countries.



karcagi.veronika@oki.antsz.hu

Marianna Nagymihaly studied biology and finished her master's degree in 2007 at the Eötvös Lorand University, Budapest. She performs the carrier testing and estimation of SMN2 copy number for all Hungarian families affected by spinal muscular atrophy with real-time PCR. In addition to her activities in the scientific work, she is employed as a TREAT-NMD database manager for Eastern-European Countries.



nagymihaly.marianna@oki.antsz.hu



Henriett Piko has worked as a staff member since 2003 and is currently conducting a PhD. She graduated as a molecular biologist, Msc. in 2000 at the Kossuth Lajos University, Debrecen. She is an expert on DMD/BMD and LGMD diagnosis. Her PhD thesis, about molecular diagnosis of Hungarian DMD/BMD patients, will be completed in 2008.

piko.henriett@oki.antsz.hu



Viktor Vancso started to work in the department in September 2007. He is currently a PhD student as well and graduated as a molecular biologist, Msc. in 2003 at the Eötvös Lorand University, Budapest. His special field is myotonic dystrophy type 1 and facioscapulohumeral muscular dystrophy. He also actively contributes to the diagnostic work of DMD/BMD by MLPA analysis.

vancso.viktor@oki.antsz.hu

NIEH Personnel involved in other TREAT-NMD Activities



Dr. Agnes Herczegfalvi MD, PhD is a paediatric neurologist and member of the National Standardization Committee of Neurologists. She is the head of the Department of Neurology of Bethesda Children's Hospital in Budapest. She is the key person in clinical diagnosis of Hungarian DMD and SMA patients and therefore contributes to WPo4.2. In addition she is a member of the TREAT-NMD Project Ethics Council (PEC).

herczegfalvi@bethesda.hu

Other Associated Personnel involved in TREAT-NMD via NIEH



Dr. Laszlo Timar, MD is a genetic counsellor at National Institute of Children's Health in Budapest. There is a long-term collaboration with him on SMA and other NMDs. He acts as an expert on ethical issues, genetic counselling and prenatal diagnosis.

timar.l@ogyei.hu

Mrs Gabriella Oberrecht is president of the Hungarian Patient Association for Duchenne Muscular Dystrophy and the National Institute has developed close links with the Parent Association.

oberrecht.gabriella@tvnetwork.hu or icgabi@hu.inter.net

For further information on NIEH please contact **Dr. Veronika Karcagi**
karcagi.veronika@oki.antsz.hu

Santhera Phase II clinical trial with SNT-317

TREAT-NMD partner Santhera Pharmaceuticals of Switzerland is planning a phase II clinical trial with SNT-317 (INN: omigapil) in children with collagen VI disorder (Ullrich Congenital Muscular Dystrophy). The investigational drug was in-licensed from Novartis last year. Together with appointed Principal Investigator Thomas Voit (Institut de Myologie, Paris), Santhera recently held a meeting of 16 clinical experts from Europe and the USA to discuss the study design and reach a broad consensus on endpoints and patient eligibility criteria. Attending the meeting in Basel on April 22nd were representatives from TREAT-NMD Enrico Bertini (Rome), Janbernd Kirschner (Freiburg), Rudolf Korinthenberg (Freiburg), Francesco Muntoni (London), Volker Straub (Newcastle) and Thomas Voit (Paris) as well as Carole Bérard (Lyon), Carsten Bönnemann (Philadelphia), Brigitte Chabrol (Marseille), Nicolao Deconinck (Brussels), Brigitte Estournet (Paris), Kevin Flanigan (Salt Lake City), Pierre-Yves Jeannot (Lausanne), Andrea Klein (Zürich), Amelie Nadeau (London), Ulrike Schara (Essen) along with members of Santhera's scientific and clinical teams.

Study sites in Europe and the US will be selected in the coming months considering geographic location as well as patient availability and other practical issues.

Further information is available from the Santhera web site: <http://www.santhera.com>



Meetings and workshops

European Research Conference in Paediatric Neurology

October 3 - 4 2008, Tübingen, Germany



The European Research Conference in Paediatric Neurology will take place on October 3 - 4, 2008 in Tübingen, Germany. The Research Conference is supported by the European Paediatric Neurology Society (EPNS) and welcomes researchers from all over Europe.

Its aims are:

- to support especially young researchers to meet their colleagues in the field for an exchange in order to facilitate collaboration,
- to foster the building of research networks.

Most of the meeting is therefore reserved for exchange in working groups.

The working group in Neuromuscular Diseases is organized by Markus Schülke (markus.schuelke@charite.de), Thomas Sejersen (thomas.sejersen@ki.se), Volker Straub (volker.straub@newcastle.ac.uk) and Nathalie Goemans (nathalie.goemans@uz.kuleuven.ac.be)

Registrants must sign up for the appropriate working group and prepare a short abstract of the research topic which should be presented in brief within the working group session (see abstract form).

Visit <http://www.treat-nmd.eu/news/item/?european-research-conference-in-paediatric-neurology> to download the conference programme and application forms.

Please note that the meeting is not sponsored. All participants have to cover their costs; a fee of EUR 70 will be requested to cover meals (coffees, lunches and dinner).

The registration form and abstract are available via the TREAT-NMD website at . Completed forms should be sent to the Tübingen conference secretariat at andrea.rohrer@med.uni-tuebingen.de.

Abstracts should include affiliation, research topic and methodology, and if possible specific interest for exchange and should not exceed 2000 characters. Abstracts will then be forwarded to the working group leaders.

You will find a list of Tübingen hotels on the registration form, and a number of rooms have been pre-reserved. As hotel rooms in Tübingen are limited it is advisable to make reservations as soon as possible.

We hope you are interested in this meeting and look forward to welcoming you to Tübingen.

Warm regards from the neuromuscular working group leaders: Markus, Thomas, Volker and Nathalie

International Symposium On Psychosocial Aspects Of Neuromuscular Disorders

June 20th-21st, 2008

Venue: "Pedro Icaza" La Comercial Auditorium and "ESIDE" Auditorium

Organized by: Department of Psychology, FICE, University of Deusto, Bilbao

For further information and registration:

www.siapen2008.deusto.es

Other news and reports

Family Guide to the Consensus Statement for Standard of Care in SMA

The Patient Advisory Group of the International Coordinating Committee for SMA Clinical Trials (the ICC) has produced a family guide to the consensus statement for standard of care in spinal muscular atrophy. The original document was published last year in the Journal of Child Neurology and contains comprehensive expert recommendations for the care of people with SMA. A number of TREAT-NMD partners were involved in drafting these recommendations and TREAT-NMD has also produced a user-friendly précis of the document, available on the TREAT-NMD website.



This new and useful document from the PAG explains the expert recommendations in family-friendly language. The family guide is available from the following link: <http://tinyurl.com/5h8gek>

TREAT-NMD recommends that you use this family guide in conjunction with the précis of the full standards document available on the TREAT-NMD website at <http://www.treat-nmd.eu/soc/eng/sma/> to help you understand what experts recommend for the treatment of the different types of SMA.



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