TREAT-NMD Neuromuscular Network

30th May 2008 · Newsletter No. 34

Welcome to the latest newsletter. This edition features a report from the EuroBioBank Annual Meeting and a call for Eastern European applicants for 6 TREAT-NMD Fellowships to attend the WMS Teaching Course.

The TREAT-NMD web site is currently migrating to a new format and the web pages will be updated on an ongoing basis over the coming days. Please bear with us during this transitional period. Thank you.

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

Best wishes,

The TREAT-NMD coordination team

Newsletter contents

*****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 have not completed our membership application form, you are not a member! 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/research/get-involved/. 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 our web site. 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.

CALL FOR APPLICATIONS: 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).

Course details can be found at: http://www.wms2008.com/training.htm

The application form is available from the WMS web site at:

http://www.wms2008.com/pdf/TREAT-NMD_WMS_%20fellowship_application_form.pdf

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Important step for Sweden to enter the TREAT-NMD Global Patient Registry initiative

On April 25-26th the Swedish Muscular Dystrophy Association (SMDF, www.smdf.se) arranged a symposium in Jönköping entitled "Duchenne and Becker muscular dystrophy, Update and hope for the future". Similar to previous SMDF symposia, this was over-booked and well represented with approximately 150 participants each day. The programme was the same both days, with professionals attending the first day, and boys/men with DMD/BMD and their families attending the second day. Hanns Lochmuller gave an overview of TREAT-NMD and specific information on the TREAT-NMD work on patient registries for DMD and SMA. Thomas Sejersen discussed DMD patient registries from a Swedish point of view, and also the TREAT-NMD work on Standards of Care for DMD. During the meeting consensus was reached on details of the DMD registry, and its initial financing was secured by SMDF.

Other topics covered by invited speakers during the symposium were "Animal models for DMD" by Dominic Wells, of the Gene Targeting Group, Imperial Colege, London, and "Translational Duchenne muscular dystrophy research" by Kanneboyina Nagaraju, Director of the Pre-Clinical Drug Testing facility at Children´s National Medical Centre, Washington DC. Anna-Karin Krogsmark and Mar Tulinius, of Göteborg, finally summarized planned or ongoing DMD studies in which Swedish centers are involved, namely CINRG, PTC124 phase 2b study, and the Prosensa phase 1 study on exon skipping.

During the 2nd day Christina and Elwyn Mandley, resigning founders of the Swedish Muscular Dystrophy Association, were thanked for all their years of hard work and important contributions to the SMDF. The art piece "Myogenesis" by Victor Dubowitz was presented to them as a token of appreciation.

EuroBioBank/TREAT-NMD Meeting Report 2008

The joint 6th annual EuroBioBank Network/ WP04.1 TREAT-NMD meeting was held on 5-6th May 2008 at the Eurordis headquarters in Paris. The meeting was well-attended by 20 participants, including representatives from all participating member biobanks, of patient organisations such as Eurordis and AFM, and also one of the leaders of the new European Biobanking & Biomolecular Resources Research Infrastructure (BBMRI), a major European scientific strategic plan for the next 10-20 years.

EuroBioBank (http://www.eurobiobank.org) is a European network of DNA, Cell and Tissue Banks founded in 2001 and coordinated by the European Organisation for Rare Diseases (Eurordis). The network is currently composed of 12 biobanks from 7 EU countries, most of them focused on the field of neuromuscular research.

The objective of EuroBioBank is to further improve and extend a network of European biobanks where scientists are encouraged to deposit biomaterials and are also welcome to obtain specific material they need for their research. Within TREAT-NMD, the special focus is on neuromuscular diseases and translational research.

Work package WP04.1 is headed by Eurordis with the goal of improving and adapting the existing EuroBioBank structures to the objectives and needs of TREAT-NMD, in particular to facilitate the exchange of neuromuscular biomaterials.

During the meeting, the EuroBioBank members were interested to hear about the Year 1 achievements of TREAT-NMD and see how they fit into the larger picture and how their work contributes to the success of this project. They were challenged to achieve higher quality in their biobanking activities, in particular quality control of samples. This can be obtained by assessing the samples and implementing a customer satisfaction survey.

Emphasis was placed again on recruiting new members and a proposal for an online membership form was made. This year, we had a new member applying for membership and were pleased to vote in as permanent member of EuroBioBank, Dr Barbara Garavaglia, representing a bank from Fondazione I.R.C.C.S Istituto Neurologico "C.Besta", Milan, Italy, specialised in neurometabolic and paediatric movement disorders.

Emphasis was also placed on the mutual benefits to be gained through collaborating with the BBMRI. As an associated organisation in the BBMRI, the EuroBioBank Network invited Dr Georges Dagher, Inserm, France, to give a talk on this new infrastructure and explore the areas of collaboration, in particular how EuroBioBank could contribute its experience of harmonisation of biobanking practices and its work on legal/ ethical issues on biobanks, to avoid duplication of efforts.

Professor Hanns Lochmüller was re-elected Scientific Coordinator of the EuroBioBank Network.

TREAT-NMD Governing Board Meeting

25-27th June 2008, Newcastle, UK

The TREAT-NMD Governing Board will convene in Newcastle in June to discuss the progress of the Network as well as planning for the future aims and objectives.

The Board will hear from Activity Leaders on the progress with the development of the Network tools, such as the Global Patient Registries, Standards of Care and Diagnosis, the Clinical Trials Coordination Centre (CTCC) and the Joint Programme of Research. The meeting will be very interactive and will aim to encourage further collaboration across the various activities and help develop a strategy for sustaining TREAT-NMD following the initial support of the European Commission which ends in 2011.

We look forward to welcoming our Governing Board representatives to Newcastle for this meeting.

Santhera Presents Efficacy Data of SNT-317 in Congenital Muscular Dystrophy at the International Congress of Myology

Santhera Pharmaceuticals, a Swiss specialty pharmaceutical company and partner in TREAT-NMD, announced this week positive preclinical data with SNT-317 (INN: omigapil) in Congenital Muscular Dystrophy (CMD) and was presented at the 3rd International Congress of Myology in Marseille, France. Tests in disease-relevant models have shown that SNT-317 prevents apoptosis and ameliorates the pathology of laminin-alpha2 deficient muscular dystrophy. Recently, omigapil was granted orphan drug designation in the European Union for the two most common sub-types of Congenital Muscular Dystrophy. Omigapil is an anti-apoptotic compound structurally derived from R(-)deprenyl (selegiline) but unlike selegiline exhibits virtually no monoamine oxidase Type B inhibiting activity and is not metabolized to amphetamines.

The efficacy of the compound was tested in a well established animal model for laminin alpha2-deficient muscular dystrophy. SNT-317 was applied orally and the results show that SNT-317 ameliorated key pathology hallmarks of the animal model. Specifically, oral administration of the compound reduced apoptosis in muscle and preserved muscle histology, reduced body weight loss, mitigated skeletal deformation and improved locomotion. Moreover, SNT-317 increased the 50% survival time from ~35 days to ~85 days and ~105 days in mice treated with 0.1 mg/kg and 1 mg/kg, respectively. In addition, it was shown that co-administration of mini-agrin had additive beneficial effects.

Thomas Meier, Chief Scientific Officer of Santhera, commented: "Our preclinical research has shown that SNT-317 reduces apoptosis and preserves muscle histology resulting in increased body weight, mitigated skeletal deformation, improved locomotion and increased life span. Based on these data, we believe omigapil is a potential therapeutic option for certain forms of CMD. At present there are no drugs approved or in advanced clinical development for the treatment of this very devastating disease."

In June 2007, Santhera in-licensed omigapil from Novartis for development in CMD and other neuromuscular diseases. Recently, the European Medicines Agency (EMEA) granted orphan drug designation for the compound in CMD with merosin (laminin-alpha2) deficiency (MDC1A) and with collagen VI deficiency (Ullrich Congenital Muscular Dystrophy and Bethlem Myopathy), the two most common subtypes of CMD.

Adult life with Spinal Muscular Atrophy (SMA)

Follow the web presentation of data and stories about the Danish SMA II population

A comprehensive survey of adult people with SMA II has been carried out by the Rehabiliterings Center for Muskelsvind (RCfM), the national Danish centre of excellence in neuromuscular rehabilitation. The results are now being published successively on the Internet, for the benefit of study participants, families, patient organizations, and the professional SMA communities.

An estimated 80% of the adult Danish population with the diagnosis has participated in an interview study ranging from specific questions about family relations, housing, education, communication skills, personal care, etc., to views on broader issues such as social relations, love, the future and quality of life.

In addition to the presentation of numerical data, people with SMA II are portrayed through a number of journalistic features giving readers a different perspective on their everyday life.

There is a global lack of knowledge about adult life with SMA II. The information gathered in this study aims to improve the information and counselling offered to those living with the diagnosis and to develop the knowledge of professionals working in the field.

To access the results go to http://www.rcfm.dk/forskning-udvikling/adult-life-with-sma-ii/

Besides survey results and personal portraits, you will also find further information on the study background, subjects and methods used.

Comments and questions are welcomed and should be sent to joje@rcfm.dk (Jørgen Jeppesen).

Large-scale community annotation – WikiProfessional

A new collaborative website has been launched initially focusing on Life Sciences and more in particular on proteins and their role in biology and medicine. The WikiProfessional technology underlying the site has been developed based upon the collaborative Wikipedia approach. Described in BioMed Central's open access journal *Genome Biology*, WikiProfessional provides a method for community annotation on a huge scale.

The article is written by Barend Mons of the Erasmus Medical Center in Rotterdam, and the Leiden University Medical Center, The Netherlands, and his co-authors from Brazil, The Netherlands, Switzerland, the UK and the USA. They include Amos Bairoch of UniProt, Michael Ashburner of GO and Jimmy Wales, the co-founder of Wikipedia. The article can be found at http://genomebiology.com/.

The source material for WikiProfessional Life Sciences comes from a mixture of existing authoritative databases (such as the Unified Medical Language System, UniProtKB/Swiss-Prot, IntAct and GO), supplemented by concepts mined from scientific papers published in public literature databases. The automated data mining identifies 'facts' in these available resources, such as protein functions or protein-disease relationships. This process created over one million biomedical concept clouds – called 'Knowlets' – around each individual concept. The developers of the site now hope that many researchers will follow their call to annotate, via WikiProfessional, the Knowlets for which they are leading experts. The method enables researchers to add data even from sources that are not openly available, such as from journals only accessible via publishers' databases, immensely enhancing the potential for comprehensive coverage. Each page of text called up via the system is automatically indexed and concepts are connected to the WikiSpace, so that their definition comes up and the information can be edited directly from the page.

The resulting data in the Wiki is fully and freely accessible to the public, and entries can be annotated by any registered user. Mons said: "We here call on a million minds to annotate a million concepts and collect new facts from full-text literature with the immediate reward of collaborative knowledge discovery and recognition of Wiki-contributions to the scientific community."

Launched in 2001, Wikipedia is a freely available, collaboratively created online encyclopedia. WikiProfessional requires user registration and maps to Wikipedia. It has been created as part of the WikiProfessional initiative and plans to add new workspaces such as WikiPeople (an intellectual networking environment), and WikiChemicals for other communities, are well underway.

A preview of the WikiProfessional technology is available on the WikiProfessional website and can be fully experienced at the WikiProfessional portal (http://www.wikiprofessional.org/).

Meetings and workshops

Clinical Trials Coordination Centre Workshop — Clinical Trials in Neuromuscular Diseases

19-21st June 2008, Freiburg, Germany

Participants are invited to this first TREAT-NMD *Clinical Trials in Neuromuscular Diseases* Workshop. The focus of the workshop is clinical trial development for neuromuscular diseases and participants will learn how to develop a trial protocol from concept to execution. Sessions on study design, biostatistics, ICH-GCP and European regulatory requirements will provide fundamental information for participants. There will be a number of hands-on sessions designed to help participants develop a protocol synopsis and experts will be available to help in all aspects of protocol development.

Further information can be found on the new TREAT-NMD web site at:
http://www.treat-nmd.eu/research/events/events/26/

7th Annual Meeting of the Muscular Dystrophy Coordinating Committee (MDCC)

23rd June 2008, Washington DC

TREAT-NMD will present an overview of its activities and explore further partnering and collaborative opportunities at the meeting organised by the NINDS/NIH.

TREAT-NMD Governing Board Meeting

25-27th June 2008, Newcastle, UK

This is the next regular meeting of the TREAT-NMD Governing Board to discuss the work of the Network. This meeting will be immediately preceded by the Project Ethics Council (PEC) meeting on the afternoon of the 25th.

The next Governing Board meeting will be in Budapest, Hungary from 2-4th February 2009.

Partner section

TREAT-NMD Governing Board Meeting in Newcastle

The agenda and supporting documentation for the Governing Board meeting can be found on the intranet site at
https://intranet-treatnmd.ncl.ac.uk/portal

Please contact Stephen Lynn (stephen.lynn@treat-nmd.eu) with questions regarding this meeting.

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