TREAT-NMD Neuromuscular Network

27 February 2009

TREAT-NMD newsletter no. 49

welcome

Welcome to the latest TREAT-NMD newsletter. This week's edition features reports on a TREAT-NMD training course and the Czech patient registry for DMD, information about tomorrow's international Rare Disease Day, and news about an honour received by Guenter Scheuerbrandt, author of the renowned Duchenne Research Reports, for services to DMD.

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/

Best wishes,

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

at a glance...

26-27 Mar 2009 UK Neuromuscular Translational Research Conference

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

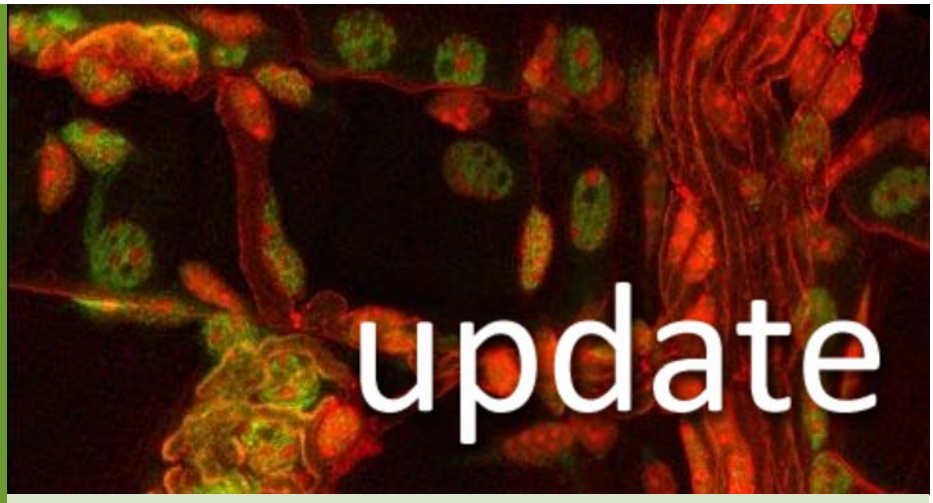
01-03 Jun 2009 Update in Neuromuscular Disorders course in London

04-06 Jun 2009 TREAT-NMD workshop: clinical trial design in neuromuscular diseases

09-11 Jul 2009 "Therapeutic Targets in CMD", Emory University, Atlanta, Georgia

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

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



Successful first TREAT-NMD Standards of Care training course

Training course on Standards of Care of Spinal Muscular Atrophy (SMA) and Duchenne Muscular Dystrophy (DMD)

February 4-5, 2009, Budapest, Hungary

TREAT-NMD, in cooperation with the National Institute of Environmental Health in Budapest, Hungary, organised a training course on Standards of Care for Spinal Muscular Atrophy (SMA) and Duchenne Muscular Dystrophy (DMD). This course took place from February 4-5, 2009 in Budapest, immediately following the TREAT-NMD Governing Board meeting. The aim of was to offer clinicians located in CEE countries the opportunity to attend a course given by specialists in SMA and / or DMD at a low cost.

The audience for this first training course consisted of 43 participants originating from 8 different CEE countries (Hungary, Romania, Serbia, Turkey, Macedonia, Croatia, Poland and Slovenia). Attendees included neurologists, paediatric neurologists, geneticists, parents and patient organisations.

The first training day focussed on “Networking for neuromuscular disorders for clinicians”, “Diagnosis and standards of care in DMD”, “Outcome measures and assessment tools in neuromuscular disorders", "Clinical trials on DMD” and “Genetic background, standards of care and clinical trials in SMA”. During the second day, the topics “Patient registries in DMD and SMA, global registries”, “Neuromuscular disorders and respiratory care in the Netherlands”, “Reference centre from concept to reality - A history of rare disease plans and initiation of reference centre model for gold standard implementation of care, registry to facilitate involvement in clinical trials”, “Social aspects, transfer from paediatrics to adulthood - Rehabilitation of patients” and “Oral and dental care in NMD patients” were covered.
All participants are to receive a thank you pack including the CD of all presentations given during the training. They also received a questionnaire, which was developed to get an insight into the value and appropriateness of this first training course on the one hand and the existing training needs among the audience on the other. This feedback will provide TREAT-NMD with a valuable tool to adjust and extend the training opportunities planned for the future.

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National seminar on Czech patient registry informs doctors about registries initiative

The Czech Parent Project for Duchenne muscular dystrophy together with the Slovak Muscular Dystrophy Association held a national meeting for doctors and patients in Prague on 19 February 09. The aim of this meeting was to inform the community about the Czech and Slovak patient registry for Duchenne and Becker muscular dystrophy and to enlist the support of doctors across the Czech Republic and Slovakia in registering patients in the system.

The Czech patient registry - which is now part of the TREAT-NMD registries initiative - was one of the first national DMD registries to come online, as early as 2005. It was developed by the Institute of Biostatistics and Analyses of Masaryk University, Brno, in collaboration with Parent Project Czech Republic and Brno University Hospital. It has an online interface, which is available in English. Meeting attendees received a live demonstration of the system, which is one of around thirty patient registries for various medical conditions run by the Institute and thus built to a very high technical standard with ISO quality certification.

In addition to presentations by the two parent associations on their activities and by Dr Taťána Maříková on genetic testing, meeting participants also received an overview by Dr Petr Vondráček of Brno University Hospital of current clinical trials in DMD, including those by AVI, Prosensa and PTC Therapeutics, and a presentation by TREAT-NMD on the way the Czech patient registry fits into the broader TREAT-NMD Global Database for DMD, which is currently receiving requests from several

companies interested in receiving information on the location of patients suitable for their upcoming trials as part of their feasibility planning for those trials. The opportunity to present the Czech registry in this international context provides a major incentive for doctors and patients to get involved with the registries initiative, as it is evidence that obtaining an accurate genetic diagnosis and registering in a registry does give patients a greater chance not only of being involved in a clinical trial for a promising new therapy, but at a later stage of being located so they can receive a suitable therapy once it has obtained marketing approval.

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Guenter Scheuerbrandt receives German Order of Merit for services to Duchenne

Guenter Scheuerbrandt, whose Duchenne Research Reports are known the world over for the accessible and comprehensive overview they provide of current approaches to research and treatment of DMD, was awarded the Order of Merit of the Federal Republic of Germany at a special ceremony on 5 February in Hinterzarten in the Black Forest. The Order of Merit was instituted in 1951 and is the highest tribute the Federal Republic of Germany can pay to individuals for services to the nation.

Guenter explains his background in Duchenne testing and his motivation for writing these reports in a summary below. His most recent reports are available here.

"On the suggestion of Hans Zellweger at the University of Iowa and Peter E. Becker at the University of Goettingen, I developed, between 1974 and 1977, an early detection program for Duchenne muscular dystrophy based on a kinetic bioluminescence screening test for creatine kinase in dry blood spots. Starting in 1977, this test ("CK-Suchtest") was offered in a voluntary program to all families with newborn boys in Germany performed in my private laboratory near Freiburg (Muscle & Nerve, 1986: 9, 11-23). The parents had to pay privately €15 (after 2002) for the test. Until February 2004, we tested 501,500 boys between 4 weeks and 1 year of age and found 150 boys with Duchenne (1:3,300) and 35 with Becker muscular dystrophy (1:14,300).

From the beginning of my work, it became clear that the families needed detailed information about the disease and the scientific research being done worldwide for finding a therapy for their sons. Therefore, I began soon after the start of our screening program to write research reports in an easy language which the patients, their families, and other people without a scientific background could easily understand. I continue writing these reports (and interviews), first in English, then in German, and Ricardo Rojas in Mexico, who has Becker dystrophy, translates them into Spanish (www.duchenne-information.eu). The last report "Research Approaches for a Therapy of Duchenne Muscular Dystrophy", published in May 2008 (21 pages), will be updated in the spring of 2009 with information from the 2008 Parent-Project meetings in Philadelphia and London and from new publications."

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Rare Disease Day 2009: February 28!

neuromuscular diseases are classified as "rare diseases". A disease or disorder is defined as rare in Europe when it affects less than 1 in 2000 citizens (Orphan Drug Regulation 141/2000). Rare diseases may affect 30 million European Union citizens. Awareness-raising events for rare diseases as a whole, initiated by bodies such as TREAT-NMD partner EURORDIS, the European Organisation for Rare Diseases, help to influence public policy towards healthcare for all rare diseases.

About Rare Disease Day 2009

Rare diseases are chronic, progressive, debilitating, disabling, severe and often life-threatening. Information is scarce and research is insufficient. People affected face challenges such as diagnosis delay, misdiagnosis, psychological burden and lack of practical support. Many rare disease patients are denied their right to the highest attainable standard of health and continue to advocate their need to overcome common obstacles.

The main objective of Rare Disease Day 2009 is to raise awareness with policy makers and the public of rare diseases and of their impact on patients' lives
Thus the aim of the day is to reinforce their importance as:

(i) A public health priority

(ii) The need for Centres of Expertise

Other main objectives of Rare Disease Day are to:

- Raise awareness on rare diseases
- Strengthen one voice of patients
- Give hope and information to patients
- Bring stakeholders closer together
- Coordinate policy actions in different countries
- Inspire continued growth of the awareness of rare diseases
- Get equity in access to care and treatment

For further information, visit http://www.rarediseaseday.org/

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www.treat-nmd.eu