

welcome

Welcome to the latest TREAT-NMD newsletter. This edition features a report on a workshop on DMD molecular diagnostics organised jointly by three major networks working in the field, news of the launch of the new NMD-chip project, an update on the CMD registry and information on the upcoming French Telethon.

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, 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 and Science and Technology Advisory Council Meetings](#)



International diagnostics meeting updates best-practice guidelines for DMD molecular diagnostics

A best practice meeting on Duchenne/Becker muscular dystrophies was held in Naarden, the Netherlands on November 14-16, 2008. The meeting was held under the auspices of TREAT-NMD, Eurogentest and EMQN, and was organized by ENMC. It convened 30 scientists and clinicians from Europe, the USA, India and Australia. In view of recent developments in both molecular testing techniques and therapeutic approaches, the aim of the meeting was to update

the guidelines for molecular testing of dystrophinopathies, which dated back to the 1990s. The presentations included updates on current approaches to testing, novel technologies, and a review on the current concepts for clinical trials, many of which rely on mutation-specific approaches. In parallel to this, the TREAT-NMD patient registries initiative aims to set up patient registries in all countries to improve trial readiness. These activities call for the harmonisation of diagnostic testing procedures, result interpretation, and reporting formats. During the discussions the content of the guideline updates was agreed, which includes the basic characteristics of the gene, the approaches to patient diagnosis, carrier detection and prenatal diagnosis. It also includes recommendations for result interpretations, mutation nomenclature and report writing.



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New NMD-chip project officially launched at Paris kick-off meeting

An exciting new project that recently won funding from the European Commission officially got underway at a kick-off meeting in Paris this week. The NMD-chip project aims to develop a "DNA chip" that will enable the quick, cost-effective and reliable sequencing of multiple neuromuscular genes.



Many cutting-edge therapies currently in development for neuromuscular diseases require a precise genetic diagnosis that identifies the precise mutation causing the disease, but since current methods of genetic testing are expensive and time-consuming, testing is not always available to patients. New technologies involving DNA-chip arrays designed to examine all possible mutations in the relevant genes in a single-step process have the potential to extend the availability of genetic testing to more patients by making the procedure dramatically cheaper and quicker.

Working closely with TREAT-NMD and including a number of TREAT-NMD partners, the NMD-chip project brings together participants across Europe and collaborators in the US to develop this technology for neuromuscular diseases. Project updates will be available via the TREAT-NMD newsletter on a regular basis.

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International registry for the CMDs moves a step closer

On November 18-19 a core working group of US congenital muscular dystrophy (CMD) experts (Dr. Carsten Bonnemann, Dr. Anne Connolly, Dr. Susan Iannaccone, Dr. Kathy Mathews,



Dr. Steve Moore, Dr. Tom Winder, Dr Susan Sparks) met with Indiana University registry experts (Dr. Jackie Jackson and Dr. Tatiana Foroud) and Cure CMD staff (Dr. Anne Rutkowski and Richard Cloud) in a workshop aimed at setting up a patient registry for the CMDs. A representative of TREAT-NMD (Dr. Anna Sarkozy) participated in the workshop to harmonize efforts in the US and Europe. The mission of the recently launched Cure CMD (www.curecmd.org) is to bring forward research, treatments and in the future, a cure for CMDs, working globally together with dedicated parent, government and research advocates. The workshop targeted creating a registry for all types of CMD and defining how the registry would function within the current infrastructure both in the US and abroad.



Unlike some other neuromuscular diseases such as DMD and SMA, the term CMD actually covers a large number of different inherited conditions caused by mutations in different genes. Since many potential future therapies are likely to be gene-specific, it is important that registries such as this one collect information on patients' precise genetic mutation wherever possible. This strategy is in line with the [TREAT-NMD registries initiative](#) for DMD and SMA, which aims to collect precise genetic information on patients worldwide in a secure manner so that whenever a potential treatment reaches the clinical trial stage, and later on when it is approved for general use, individuals who might benefit can be contacted as quickly as possible.

In addition to this aim of "trial readiness", the CMD registry aims to assist in the diagnosis, classification and prevalence assessment of CMD for natural disease progression studies and for preclinical and clinical therapeutic trials, and to drive pharmaceutical industry interest in the CMDs with investment in therapeutic targets.

The CMD registry is in the development stages with a goal to pilot the online version within 6 months to 1 year. During the meeting, attention was focused on connecting the CMD registry with existing national and international infrastructures and with TREAT-NMD. In particular, a subset of data elements will be used to direct patients with specific conditions classified under the general "CMD" banner into mutation-based global registries focused on clinical trials, such as the FKRP global registry currently under development by TREAT-NMD.

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French Telethon is major fundraising event

TREAT-NMD partner the AFM (the French Muscular Dystrophy Association), has set itself two missions: curing neuromuscular diseases and reducing the disabilities they cause. To achieve these goals, AFM has organised an annual "Telethon" on the first weekend in December every year since 1987. This major fund-raising operation, combining a 30-hour TV show and tens of thousands of local events across France, provides the majority of donations to the Association and also aims to raise public awareness of neuromuscular diseases.



With the investment of 845 million euros for its Cure mission between 1987 to the end of 2007, the AFM has become an indispensable actor in the field of research. In 2007, 66 million euros have been invested in almost 400 research programs.

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