

welcome

Welcome to the latest TREAT-NMD newsletter.

This week's edition includes...

- information on the recently published best practice guidelines on molecular diagnostics in DMD/BMD
- an update on the first six months of the BIO-NMD Project
- information from Families of SMA ahead of their upcoming conference
- a call for future workshop applications from ENMC

We would like to thank those who have contributed to this week's edition. This newsletter relies on input from our readers. If you have anything you wish to be included in the next newsletter please contact us at info@treat-nmd.eu

at a glance...

[19-23 Jun 2010 20th Meeting of the European Neurological Society - Berlin, Germany](#)

[24-27 Jun 2010 Parent Project Muscular Dystrophy Annual Conference - Denver, Colorado, USA](#)

[24-27 Jun 2010 Families of SMA Annual Conference - Santa Clara, California, USA](#)

[15-16 Jul 2010 MDR2010 - Padova, Italy](#)

[19-23 Jul 2010 12th International Congress on Neuromuscular Diseases - Naples, Italy](#)

[30 Jul - 1 Aug The Biennial FSHD International Patient and Researcher Network Meeting - Las Vegas, Nevada, USA](#)

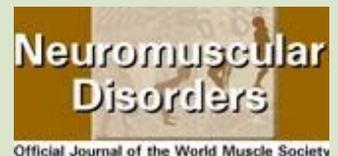
[29 Aug - 1 Sep European Meeting on Next Generation Sequencing - Leiden, Netherlands](#)

[9-10 Sept 2010 MD2010 - 4th conference for the Muscular](#)



Best practice guidelines for molecular testing published in Neuromuscular Disorders

In view of recent developments in both molecular testing techniques and therapeutical approaches for DMD, the need to update the guidelines for molecular testing of dystrophinopathies, which dated back to the 1990's, was identified. To this end, a Best practice meeting on Duchenne /Becker muscular dystrophies was held in Naarden, the Netherlands on November 14-16, 2008. The meeting report has recently been published in Neuromuscular Disorders and is available to download free of charge.



The meeting was held under the auspices of Eurogentest, TREAT-NMD, EMQN, and was organized by ENMC. It convened 30 scientists and clinicians from 21 countries from Europe, the USA, India, and Australia. 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.

During the meeting discussions the content of the guidelines was agreed, which includes the basic characteristics of the gene, the approaches to diagnose affected patients, to carrier detection, and prenatal diagnosis. It also includes recommendations for result interpretations, mutation nomenclature, and report writing.

To download the article from Neuromuscular disorders please click [here](#).

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BIO-NMD: 6 month project update

BIO-NMD is a collaboration of 12 European partners working together with the common aim of trying to identify possible biomarkers in people with neuromuscular disease. The ultimate goal is to improve the development of treatments and diagnosis of people affected by neuromuscular disease, specifically Duchenne Muscular Dystrophy and Collagen VI myopathies.



The project is currently in its 6th month and significant progress has been made. A brief and edited summary of the work done by the 12 partners so far is detailed below. A full copy of the update can be found on the [BIO-NMD website](#).

University of Ferrara - project co-ordinator - has obtained approval from Local Ethics Committees together with licences for animal experiments. A task force has been set-up for selecting the patients to be studied and a list of genes for analysis has been built-up.

Leiden University Medical Centre has screened the serum of Duchenne patients for possible protein biomarkers. They have so far found two proteins with elevated levels in DMD patients compared to healthy age-matched controls.

Newcastle University has created a patient-friendly part of the project's website in collaboration with patients' groups (www.bio-nmd.eu). They have also been working as part of the clinical task force in selecting patient sub-groups and establishing a standard protocol for the collection of samples.

University of Padova has studied mice with a genetic mutation which means they do not produce

[Dystrophy Association of Western Australia - Perth, Western Australia](#)

[18 Sept 2010 Muscular Dystrophy Campaign Conference - Birmingham, UK](#)

[20-22 Sept 2010 Muscle Study Group Annual Meeting - Buffalo, NY, USA](#)

[12-16 Oct 2010 World Muscle Society International Congress - Kumamoto, Japan](#)

[12-13 Nov 2010 Action Duchenne 8th Annual Conference - London, UK](#)

Please note: This is only a selection of upcoming meetings. To see all our listed meetings click [here](#).

collagen VI in order to gain new insight into the molecular mechanisms of collagen VI disease.

Institute of Child Health, University College London has been actively involved in the selection of patient characteristics for the study, patient recruitment and RNA sequencing.

University of Rome Tor Vergata are keeping up to date with the European Regulatory Agency concerning new rules and guidelines about genomic biomarkers in order to guide future work of the other partners in BIO-NMD.

Institut National de la Santé et de la Recherche Médicale has begun to identify key tools used by partners and experts within BIO-NMD with the help of a questionnaire. They have also started development of the software that will be able to predict the effects of certain genetic mutations. University of Milan has focused attention on studies of mice deficient in collagen VI in order to provide new information about the effect that different protein expression has on the animal model. Preliminary results have indicated that different muscles are affected in a different way by collagen VI mutations. Conclusion of the analysis is planned for the first week of July 2010.

Royal Institute of Technology Stockholm has installed a new LUMINEX instrument which enables analysis of 384 samples in one single run. They have also been in contact with BIO-NMD partners to discuss which type of patient samples (blood, serum or plasma) should be analyzed, the amount required for the analysis and the number of samples needed.

Ariadne Genomics Inc has developed a detailed plan for building the first snapshot of an NMD knowledge base. They have also started outlining the molecular mechanisms of various muscular dystrophies.

AppliedBiosystems has been working on a method for the targeted re-sequencing of 80 genes related to NMD. The plan is to get samples and a list of genes from the University of Ferrara and start designing the exact technique to be used for analysis.

Projets et Réseaux de Recherche organised the Kick-Off Meeting for the project along with the project coordinator (UNIFE). The team has also monitored the achievement of the first 6 month's targets and related reports, and supported the BIO-NMD partners in their work on a daily basis.

More information about BIO-NMD is available from the website at www.bio-nmd.eu or by contacting Cathy Turner, Communication and Dissemination Officer - catherine.turner@ncl.ac.uk Tel: +44 (0) 191 241 8659

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Families of SMA Conference approaches

The Annual Families of SMA Conference hosts the USA's largest gathering of those affected by SMA and leading researchers from around the world, to share ideas and hope for the future.

The conference running from 24- 27th June in Santa Clara, California allows families to learn about the latest SMA research, increase their knowledge of the disease, share the latest in disease management techniques and network with other families and leading medical and scientific professionals.

FSMA's approach has the crucial component that both researchers and families work closely together as a team. They are dedicated to educating families and healthcare professionals and to bringing the entire community together for growth and support.

By fund raising over the last 25 years, the organisation has funded \$50 million(US) in SMA research and has funded more than 140 Basic Research Grants at 70 institutions around the world.

FSMA is dedicated to creating a treatment and cure by funding and advancing a comprehensive research program which is split into three distinct parts.

- Basic Research Grants (to unravel the biology of SMA)
- Drug Discovery Projects (to make new SMA drugs)
- Clinical Trial Initiatives (to test drugs in humans)

Along with funding and directing the leading SMA research, FSMA provides core resources and assistance that help families navigate through life with SMA, providing a stable, unbiased platform for SMA families to live active, engaged, hopeful lives. Funds raised are distributed via an advisory board consisting of medical and scientific experts, to fund SMA research projects and also improve patient care for those with SMA.

Further details of FSMA can be downloaded [here](#) and details of their conference can be found [here](#).

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ENMC is accepting applications for workshops in 2011

The European Neuromuscular Centre (ENMC) aims to encourage and facilitate collaborative research into neuromuscular diseases (NMD). In order to foster these goals, the ENMC sponsors grants for international workshops on NMD.



For ENMC workshops to be conducted in the first half of 2011, the deadline for submitting the applications is September 15, 2010. Further information about the procedure and the forms to be completed for a workshop application can be downloaded from our website www.enmc.org.

ENMC workshop proposals should be based on well focused topics within the neuromuscular field. The chairperson of an ENMC Workshop is responsible for the scientific aspects of the programme and inviting participants. Scientists who participate in ENMC workshops will normally have published relevant papers in international peer reviewed journals. All ENMC workshops are held in Naarden, The Netherlands and take place during the weekend to allow full participation and reduced flight and accommodation costs.

If you have any questions regarding this, please do not hesitate to contact the ENMC office enmc@enmc.org

Deadline for submission, September 15, 2010

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