

welcome

Welcome to the latest TREAT-NMD newsletter.

This edition includes:

- a call for abstracts for the 9th EPNS Congress
- a call for projects from the Myotubular Trust
- an article about the effects of technological advances in genetic sequencing
- programme information about the 8th Action Duchenne conference

We would like to thank those who have contributed to this 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...

11-12 Nov 2010 International Workshop for Glycosylation Defects in Muscular Dystrophies - Charlotte, North Carolina USA

12-13 Nov 2010 Action Duchenne 8th Annual Conference - London, UK

19-20 Nov 2010 Steps Forward in Pompe Disease - London, UK

9-11 Dec 2010 The 3rd Latin American Summer School of Myology EVELAM - Córdoba, Argentina

15 Dec 2010 Systemic delivery of AAV for neuro-muscular gene therapy - Evry, France

21-22 Feb 2011 7th Annual Update Symposium on Clinical Neurology and Neurophysiology - Tel Aviv, Israel

24-26 Mar 2011 International Congress of the European Society of Magnetic Resonance in Neuropediatrics - Amsterdam, Netherlands

29-30 Mar 2011 The Fourth UK



Call for Abstracts for the 9th EPNS Congress

The 9th European Pediatric Neurology Society Congress will be held in Cavtat / Dubrovnik in Croatia from **11th - 14th May 2011**.

Authors are invited to submit abstracts for both oral and poster presentations. The deadline for submission is **15th November 2010**. Topics include...

- Symptomatic epilepsy management and epilepsy surgery
- Neuromuscular disorders update
- Neurodegenerative and neurometabolic disorders genetics and treatment
- Neuroimmunology
- Pediatric neurocritical care
- CNS infections: immunology, complications and treatment
- Advances in neuroimaging

The Congress is a valuable opportunity for a comprehensive and wide-ranging update on the latest topics in paediatric neurology. The programme includes a specialist workshop on "advances in neuromuscular disorders: diagnosis and treatment - in cooperation with TREAT-NMD", to be held on Tuesday May 10th.

Further details of the congress and the call for abstracts can be found on the [congress website](#).



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Myotubular Trust call for projects

The **Myotubular Trust** is holding a third call for projects.

Applications need to be submitted by

mid January 2011

and awards will be made in

May / June 2011.

It is anticipated that funding will be made available for further projects that will help find a cure and / or a treatment for any of the three types of myotubular myopathy:

- congenital X-linked recessive
- congenital autosomal recessive
- autosomal dominant

Projects should focus on research that would not generally be funded by public or industrial funding sources. The call will be open internationally and awards are made to a named individuals.

The following types of application are sought:



Neuromuscular Translational
Research Conference - London, UK

7-8 April 2011 Chinese conference
on translational research in DMD -
Guangzhou, China

9-16 April 2011 63rd American
Academy of Neurology Annual
Meeting - Honolulu, Hawaii

9-13 May 2011 Myology Congress -
Lille, France

18-22 Oct 2011 WMS Conference
2011 - Algarve, Portugal

8-11 Nov 2011 TREAT-NMD
International Conference - Geneva,
Switzerland

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

1. A project grant applied for by a Principal Investigator to fund a project for 2-3 years duration to be carried out by a postdoctoral researcher, or PhD student

2. A Myotubular Trust fellowship – basic science (3-4 years duration), where the scientist has identified a group that he or she wants to work with.

In particular encouragement is given to the application of new technologies to research into myotubular myopathy, which may involve collaboration between different medical disciplines and / or different research institutions. Applications which involve joint funding with other organisations will be considered.

Past awards include:

- Gene therapy for x-linked myotubular myopathy and pathophysiology – Dr Anna Buj Bello, Genethon, Evry

- Membrane trafficking and T tubule structure and function in a canine model of centronuclear myopathies – Dr Richard Piercy, Royal Veterinary College, London

- Deciphering the molecular pathway involving centronuclear myopathy genes – Manuela D'Alessandro, IGBMC, Illkirch

Further information

If you wish to learn more about the Myotubular Trust, please visit www.myotubulartrust.org or email research@myotubulartrust.org

Further guidance and an application form can be found on the website.

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New technology can drive understanding of disease mechanisms

Technological advances are crucial to the understanding of the mechanisms behind neuromuscular disease and the quest for therapies. Raimo Tanzi, an industry partner ([Applied Biosystems/Life Technologies](#)) involved in the [BIO-NMD](#) project, discusses the advances in next-generation sequencing that are driving genetic research, enabling genetic diagnosis to be made available to increasing numbers of patients and underpinning the work to develop high-throughput "gene chips" in projects such as [NMD-chip](#).



"New discoveries that mark the progress of science may stem from the brilliant ideas of bright scientists, but more often than not are made possible by the progress of technologies for investigation. In this respect biology is no different from other sciences and, in the same way as more powerful telescopes enable astronomers to find new stars and planets, in the analysis of the biological reasons for diseases, more powerful DNA analysers can give a strong push to discoveries.

After about ten years of technological stability where the first generation of automated DNA sequencers - those which contributed to the publication of the first sequence of the human genome in 2001 - didn't undergo any major development, suddenly in 2008 a new generation of DNA analyzers was developed and commercialised. These new instruments increased productivity by more than 1 million fold, at the same time decreasing costs by 10,000 fold and bringing the cost of sequencing one billion bases down from 2 million Euros to less than 200. This dramatic leap forward opened the way to completely new methods of investigation which now may involve even the sequencing of the entire genome (3 billion bases) in an individual patient.

The time was thus ripe to prove the utility and value of the new technology in some challenging areas of research and here the BIO-NMD project came into play. The scope of this aspect of the project is to identify the reasons why some patients affected by neuromuscular diseases develop different levels of severity and/or respond differently to drug treatments even when the defective gene causing the disease is the same. This must involve differences in other areas of the genome of these individuals which, up to now, have not been possible to identify.

As participants in the BIO-NMD project we aim to develop novel investigational methods to allow the consortium to identify those genes involved in determining different levels of severity of the disease, which would in turn open up the possibility of identifying more effective drugs."

"Progress in science depends on new techniques, new discoveries, and new ideas, probably in that order." Sydney Brenner, 2002 Nobel Prize Winner

By: Raimo Tanzi
Partner in the BIO-NMD project
Director Business Development Next Generation Sequencing
[Life Technologies](#)

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Action Duchenne's 8th Annual Conference - London, UK

The Action Duchenne Conference this year will take place on Friday November 12th and Saturday November 13th at the Hilton Hotel Canary Wharf in London.



Keynote speakers include..

- Dr John Bourke
- Professor Kate Bushby
- Professor Mehul Dattani
- Professor Dame Kay Davies
- Dr Mike Gait
- Professor Veronica Hinton
- Dr Steve Shrewsbury
- Professor Steve Wilton

Sessions titles include...

- Human genome at 10: Patient trials for Duchenne
- Improving clinical trial design and outcomes
- Exon skipping - improving delivery and efficiency
- New methods for detailed genetic analysis
- Duchenne, ADHD, Dyslexia and Autism
- Endocrine issues and long term steroid use

In parallel to these sessions, various workshops and breakout sessions have been organised. Full details can be found in the [full conference programme](#) which is now available.

Registration can take place via the [Action Duchenne Shop](#) and a gala dinner has also been organised. For further details please visit the [Action Duchenne website](#) or contact Action Duchenne by [email](#).

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