Welcome

Welcome to our 73rd newsletter which includes:

- An introduction to the BIO-NMD project
- Details of the clinical trials workshop to be held in Freiburg
- Information on a hands-on workshop on non-invasive outcome measures at the Institut de Myologie
- SMA Europe's grant awards
- A new Cochrane report on ankle range of motion
- Information about the joint neuromuscular meeting in Vaals, Netherlands

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

BIO-NMD biomarkers project kicks off with a meeting in Ferrara

The BIO-NMD EU project (FP7, HEALTH-2009-2.4.4-1: Rare neurological diseases) “Identifying and validating pre-clinical biomarkers for diagnostics and therapeutics of Neuromuscular Disorders” was officially launched on the 28th-30th of January 2010, at a kick-off meeting in Ferrara.

The consortium is led by the University of Ferrara’s Prof. Alessandra Ferlini and involves 12 partners from across Europe and the US. BIO-NMD is funded until December 2012 and will focus current funding on identifying biomarkers in the collagen VI myopathies and Duchenne muscular dystrophy.

45 participants from Italy, the United Kingdom, the Netherlands, France, Germany, Sweden, and United States attended the kick-off meeting. Participants included BIO-NMD partners and members of the BIO-NMD committees including the scientific advisory board (SAB), patient association committee (PAC) and project ethics board (PEB). These participants included a number of patient associations and granting agencies, namely Telethon Italy, Association Francaise contre les Myopathies, Duchenne Parent Project, Muscular Dystrophy Campaign as well as representatives of the TREAT-NMD EU Network of Excellence and the EU NMD-CHIP project.

What is the aim of BIO-NMD?
BIO-NMD is a translational project devoted to biomarker discovery and validation in muscular dystrophies with the aim of improving disease and therapy monitoring and focused on Duchenne muscular dystrophy and the collagen VI myopathies. During the project the partners will first study muscle tissue or myogenic cells (available through the EuroBioBank), and then move to validate the identified biomarkers in blood and urine. Preliminary biomarkers identified during the first 18 months will subsequently be validated in prospective patient cohorts as part of the study.

BIO-NMD will adopt OMIC approaches (extensive genomic and proteomic high throughput analysis) for biomarkers discovery, in easily accessible patient specimens such as blood and urine, in order to maximize benefit and minimize pain and invasiveness for patients involved in diagnostic or therapeutic procedures.

Why are biomarkers important?
Several clinical trials for collagen VI myopathies and Duchenne muscular dystrophy are in the planning phases. Therefore, the development of disease specific biomarkers via the BIO-NMD project is very important. Biomarkers are critical tools used to monitor clinical trials as they allow a clinical trial expert to: determine if a drug is having a positive effect on improving disease; identify early side effects; and personalize the patient’s clinical treatment (see Personalized Health Care or PHC).

How will a disease-specific biomarker provide useful information?
Both Duchenne and collagen VI myopathies are variably progressive diseases, even within one particular genetic subtype. Innovative clinical trials are expensive and typically run for less than 18 months, making it difficult to see a “positive” drug effect that produces subtle but important changes on an affected child’s functional abilities. A biomarker can level the playing field and allow a clinical trial expert to determine how a drug is affecting each affected individual based upon changes in that individual’s biomarker levels over the course of the trial. Given the limited number of children and adults with a particular neuromuscular disease who can participate in future clinical trials, it is critically important to identify objective changes and increase the sensitivity and power of a study.

Clinical Trials Coordination Centre trial design workshop

at a glance...

18-20 Mar 2010 International Conference on Rare Diseases and Orphan Drugs - Buenos Aires, Argentina
25-26 Mar 2010 Asian Oceanian Myology Center Annual Meeting - Seoul, South Korea
10-17 Apr 2010 American Academy of Neurology, Annual Meeting - Toronto, Canada
3-7 May 2010 International Child Neurology Congress 2010 - Cairo, Egypt
10 May 2010 The George Karpati Symposium on Neuromuscular Disease: Innovation and Application, Montreal, Canada.
**Background**

One of the most common reasons for failed trials is poor protocol design. As neuromuscular disorders are very rare, clinical trials have to be multi-centre or even multinational to include enough patients. As a result, the study design for these trials is usually complex. Also, academic trials have come to face a changed regulatory environment following the implementation of the EU Clinical Trials Directive 2001/20/EC.

Therefore, the aim of this workshop is to improve the efficiency of clinical trials in neuromuscular disorders and other rare diseases by

- introducing future investigators to the fundamentals of effective clinical trial design by means of lectures
- developing study protocol drafts in small group sessions in conformance with ICH-GCP
- fostering contacts between investigators to facilitate later multinational cooperation through informal discussion sessions

**Target Audience**

This workshop is aimed at physicians specialising in neuromuscular disorders and other rare diseases who are interested in clinical trial work. One focus will be on study design in small numbers. Participants are asked to bring a draft study plan to provide the basis for writing a synopsis during the workshop.

**About the TREAT-NMD CTCC**

The Clinical Trials Coordination Centre was established in May 2007 as one activity of TREAT-NMD, a pan-European ‘network of excellence’ aimed at improving treatments and finding cures for patients with neuromuscular disorders. It was built up in collaboration with one of Germany’s leading Departments of Neuropaediatrics and Muscle Disorders and the Clinical Trials Center (ZKS) of the University Medical Center in Freiburg, Germany. Both have longstanding experience in the field of neuromuscular disorders and in the conducting of clinical trials.

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Institut de Myologie/TREAT-NMD hands-on workshop on non-invasive outcome measures for NMDs

30 Jun 2010 - 01 Jul 2010

A workshop led by outcome measures specialists at the Institut de Myologie (Pierre Carlier, director of the NMR laboratory and Jean-Yves Hogrel, director of the neuromuscular physiology and evaluation laboratory) will be held in Paris immediately following the Paris Summer School of Myology at the end of June 2010.

A maximum of four participants, preferably having taken part at one of the Myology Schools, will receive intensive hands-on training in the latest non-invasive outcome measures techniques, including force measurements and quantitative NMRI.

There is no cost for attendance. Applicants will be selected based on CV, short motivation letter and phone interview.

Brief program

Day 1: force measurements, activity monitoring

Day 2: NMRI: whole body-imaging, quantitative analysis of muscle composition and of inflammation, new perspectives in spectroscopy

For further details or to express your interest, please contact Pierre Carlier.

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New Cochrane review on ankle range of motion

New Cochrane Neuromuscular Disease Group review: Interventions for increasing ankle range of motion in patients with neuromuscular disease.

This new review looks at interventions to improve ankle flexibility in patients with neuromuscular disease and reports the results of randomised controlled trials in Duchenne muscular dystrophy and Charcot-Marie-Tooth disease.

“Loss of ankle flexibility is a common problem for people with neuromuscular disease. It can cause foot deformity, pain and problems walking. The purpose of this review was to assess the evidence regarding the effectiveness of interventions for improving ankle flexibility in people with neuromuscular disease. Four studies were included in the review involving a total of 148 participants. Two studies showed that wearing a night splint was no more effective than not wearing a night splint for increasing ankle flexibility in 26 people who had Charcot-Marie-Tooth disease type 1A. One study showed corticosteroids (prednisone) did not significantly improve ankle flexibility in 103 boys with Duchenne muscular dystrophy and the other study showed that while orthopaedic surgery initially increased ankle flexibility in 20 young boys with Duchenne muscular dystrophy this was not sustained in the long term. This review shows that, currently, there is limited evidence supporting any intervention for improving ankle flexibility in patients with Charcot-Marie-Tooth disease type 1A and Duchenne muscular dystrophy. More research is needed.”


Topics seeking authors

If you would be interested in becoming an author of a systematic review, please contact the Cochrane Neuromuscular Disease group at: cochraneemd@ion.ucl.ac.uk. The group provides support for authors and can assist with training via the UK Cochrane centre or overseas centres. Priority topics seeking authors include:

- Management of respiratory failure in acute neuromuscular and chest wall disorders
- Volume-cycled versus pressure cycled ventilation
- Weaning from mechanical ventilation in neuromuscular patients
- Diaphragm pacemaker
- Feeding: endoscopic gastrostomy
- Cough augmentation for SMA

You may know of other topics relevant to TREAT-NMD that have not yet been covered. If so, please let
us know - especially if you would like to volunteer as an author.

To learn more about the work of the group please visit our [website](#).

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**SMA Europe announces grant recipients**

SMA Europe is pleased to announce the award of about €650,000 for research into Spinal Muscular Atrophy – SMA, in response to its recent Call for Research Projects 2009.

SMA is a rare but devastating disease. Approximately one in every 6,000 children is born with it; it is the largest genetic killer of babies and knows no national or racial boundaries. Muscle strength is gradually reduced until even breathing unassisted is not possible for those worst affected.

Significant research progress has been made in recent years in understanding SMA, and much of this research has commonalities with that of other neuromuscular conditions.

The seven institutions which attracted support are:

- Columbia University, New York, USA: Professor Umrao Monani
- Molecular Genetics Institute of Montpellier, France: Professor Rémy Bordonne
- The University of Sheffield, UK: Professors Ke Ning and Mimoun Azzouz
- University of Western Australia, Australia: Professor Steve Wilton
- University of Bern, Switzerland: post-doctoral fellow Dr Rachel Nlend Nlend
- INSERM, Université de Nantes, France: post-doctoral fellow Dr Beatrice Joussemet
- Emory University, Atlanta, USA: post-doctoral fellow Dr Claudia Fallini

For further details of this year's awards and how to apply for next year please click [here](#).

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**Belgian, Dutch and German Joint Neuromuscular Meeting in Vaals 23-24 April**

As a logical continuation of the first successful meeting in 2007 of the Belgium-Dutch neuromuscular Study Club and the German Reference Center for Neuromuscular Diseases of the DGNN (Deutsche Gesellschaft für Neuropathologie und Neuroanatomie) the next meeting is being organized at the beautiful Kasteel Bloemendal in Vaals, April 23-24, 2010.

Vaals is located at the Drielandenpunt; the place where the borders of Belgium, Germany and the Netherlands meet.

The meeting includes state of the art lectures on motor neuron diseases, polyneuropathies, diseases of the neuromuscular junction and myopathies. Both clinical and basic aspects will be addressed.

The deadline for abstracts to submitted is now 26th March 2010. Please send abstracts no longer than 250 words including title, authors, affiliations to info@euronschool.eu. Further details can be found in the [PDF download](#) and online registration is available on the [Euron site](#).

Further details can be found in the meetings and events section of our website.