05 June 2009
TREAT-NMD newsletter no. 56
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welcome

Welcome to our 56th Newsletter which begins with a report covering the international conference that was held recently in Kharkiv in the Ukraine. The three day conference focusing on the recent standards in diagnosis, treatment and medical care for some rare neuromuscular diseases attracted participants from a wide range of backgrounds. Ukrainian speakers talked about diagnosis, care and also treatment, whilst participants from various European countries covered topics such as international standards of care, physiotherapy, diagnostics and patient organisations.

The National Center for Canine Models for DMD (NCDMD) has a call for proposals, the goal of the organisation is to develop and sustain dog models of DMD to be used in preclinical therapy development studies. Funded jointly by both NINDS and NIAMS proposals need to be submitted by the 15th July for the steering committee's consideration.

Experts in Congenital Muscular Dystrophy are wanted to join a workshop to create a consensus document focusing on the standards of care and diagnosis. This is with a view to moving CMDs forwards toward a clinical trials readiness phase. The drive to develop this came from a meeting of TREAT-NMD staff, international CMD experts and Cure CMD.

We also include a report on Czech and Slovak registries for both DMD and BMD. This is available to read via a download link from the website. The document covers both the development of the registry in the two countries; this registry also provides a detailed phenotypic and genotypic description of patients, who have access to their own data.

We have a report from the American Academy of Neurology, their annual meeting was held in Seattle and TREAT-NMD was represented with two poster presentations one of which can be downloaded from our website.

International conference in Ukraine improves international collaboration

From 21 - 23 May 2009 the Ukrainian patient charity Children with Spinal Muscular Atrophy, together with the Institute of Neurology, Psychiatry and Narcology of the Academy of Medical Sciences in Kharkiv, Ukraine, hosted an international conference on "recent standards in diagnosis, treatment and medical care for some rare neuromuscular diseases". Participants included representatives from various institutions in Ukraine - health authorities, rehabilitation, diagnostics and everyday care - plus patients and families, and international participants. Speakers from across Ukraine gave presentations on various aspects of neuromuscular diagnosis, care and treatment, while participants from the UK, Sweden and France spoke on international care standards, physiotherapy, MRI techniques, LGMD diagnostics, social and ethical considerations, patient organisations, and TREAT-NMD.

One of the key outcomes of the meeting is the recognition that neuromuscular diseases require support at the national level within Ukraine. After the conference, consultation with Vladimir Martynyuk, Chief Child Neurologist of the Ministry of Health of Ukraine, resulted in an agreement on support for a national patient registry for SMA and DMD. The opportunity for creation of a special National Program for SMA and DMD at a national level was also discussed and seems to be a real possibility within a short time.

As well as these important national steps, closer international ties were established in a meeting between TREAT-NMD and representatives of the Ministry of Health and several other institutions (the Institute of Neurology, Psychiatry and Narcology of the Academy of Medical Sciences, IMBG - Department of Human Genomics, Institute of Molecular Biology and Genetics of the National Academy of Sciences of Ukraine, IPAG - Institute of Pediatrics, Obstetrics and Gynecology of the Academy of Medical Sciences of Ukraine), who are keen to work together with TREAT-NMD. A Memorandum of Understanding between the Ukrainian institutions and TREAT-NMD, AFM and the Institut de Myologie is also being planned, and this will hopefully result in new opportunities for international training and exchange visits between clinicians and scientists.

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National Center for Canine Models of DMD - Call for Proposals

The National Center for Canine Models of Duchenne Muscular Dystrophy (NCDMD) was established at the University of North Carolina at Chapel Hill in May 2008 through a U24 grant jointly funded by NINDS and NIAMS. Information on the Center can be found at http://www.ncdmd.org/.

The overarching goal of the NCDMD is to develop and sustain dog models of DMD to be used in preclinical therapy development studies ultimately leading to human clinical trials. In addition to producing dogs, the NCDMD provides high quality facilities and services to conduct independent, standardized testing of candidate therapeutics in compliance with GLP standards. Importantly, because the breeding colonies and key personnel are supported through the U24 grant, fees charged to investigators are reduced.

Proposals are being solicited to access the services and subsidy provided by the NCDMD. The application process, including the required form and support materials, is detailed on the NCDMD website. Decisions on funding of these applications will be made by the NCDMD Steering Committee (see membership under "About Us" on the website). Key criteria to be evaluated include results from

Günter Scheuerbrandt's exon skipping report that was featured in one of last month's newsletters has now been translated into Japanese and Spanish and are available to download from the website.

We also have included a reminder that the deadline for the TREAT-NMD DMD UK Coordinator post is fast approaching; the closing date for applications is Wednesday 17th June.

Finally the application process for ENMC workshops has changed, details of the new procedure and submission dates are mentioned.

We hope you find these articles informative, and please feel free to distribute them to colleagues who might find them of interest, or better still ask them to subscribe to our newsletter directly.

Have an enjoyable weekend and best wishes,

Katie, Volker, Hanns, Steve, Emma, Rachel, Samantha and Michael: the Newcastle TREAT-NMD team.

at a glance...

25-28 June 2009 PPMD Annual Connect Conference in Atlanta, Georgia.

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

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

09-12 Sept 2009 14th International Congress of the World Muscle Society, Geneva, Switzerland.

05-06 Oct 2009 6th UK SMA Research Conference, Edinburgh, UK

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

the mdx mouse, availability of preliminary data from normal dogs (pharmacokinetics of the proposed compound, etc), and the likelihood that the compound can be moved expeditiously to human trials. In response to NCDMD's initial RFA earlier this year, seven applications were received and two were funded.

Projects receiving a subsidy must complete sequential milestones to progress through the NCDMD. These milestones will vary with individual projects but will generally involve demonstrating initial success at the molecular or histological level before moving to physiological testing and imaging.

Investigators should contact NCDMD Director Joe Kornegay directly to discuss potential projects. Proposals must be submitted by July 15, 2009 and will be reviewed by the NCDMD Steering Committee in early August. Investigators will be notified of the Committee's funding decision by August 15.

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Experts in Congenital Muscular Dystrophy Required

Wanted: Experts in Congenital Muscular Dystrophy to Work on Consensus Document for CMD Standards of Diagnosis and Care.

The Congenital Muscular Dystrophy Standards of Care Workshop is a focused international effort to compose a consensus document that standardizes the care of patients with the congenital muscular dystrophies (CMDs) across several medical specialties. An initiative to drive the development of CMD consensus care standards arose from a recent TREAT-NMD sponsored workshop bringing together TREAT-NMD staff, international CMD experts and Cure CMD, a non-profit patient advocacy group. This workshop complements current efforts to move the CMDs into a clinical trial readiness phase and as such, builds upon parallel efforts to launch an international CMD patient registry (CMDIR) and identify therapeutic targets.

The steering committee consists of: Dr. Thomas Sejersen, Dr. Ching Wang and Dr. Anne Rutkowski. We have set a goal to recruit a minimum of 8-10 internationally recognized specialists with CMD experience from each of the following medical specialties:

1. Orthopedics / Rehabilitation
2. Pulmonary / Intensive care
3. Cardiac
4. Gastrointestinal / Nutrition
5. Neurology (congenital brain anomalies, developmental delay and epilepsy)
6. Diagnostic and Genetics
7. Palliative care (pain, end of life issues and psychosocial supports)
8. Dental / Oral care (will only require 3 specialists and one leader)

Each medical specialty forms a working group and together with the steering committee they form the SMC Standard of Care Committee (SOC). To facilitate the CMD Standards of Care Workshop, the steering committee has developed a timeline with set goals to drive a successful pre-workshop work effort allowing for the successful draft of the CMD Standards of Care document culminating in a proposed TREAT-NMD November workshop in Brussels, November 20-22nd to follow the TREAT-NMD/NIH conference "Bringing down the Barriers: Translational Medicine in Neuromuscular Disease".

If you are interested in participating in the CMD Standards of Care workshop, please send the following to anne.rutkowski@curecmd.com

1. Your updated curriculum vitae
2. Your clinical experience in neuromuscular disorders, including CMD
3. Your research interests
4. The subspecialty working group you would like to join

If you would like to recommend an individual who has particular expertise in any of the subspecialty areas working with CMD patients, please give the person's contact information to anne.rutkowski@curecmd.com

In order to ensure the high efficiency of the working groups, we ask that all committee members commit the time to participating in periodic conference calls prior and following the Workshop and make plans to attend the CMD SOC Workshop in Brussels in the fall.

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Czech and Slovak DMD Patient Registry Features in Neuromuscular Disorders

The Czech and Slovak DMD patient registry, one of the national registries feeding into the TREAT-NMD Global Registry, is described in Neuromuscular Disorders volume 19 (April 2009).

Effective planning of clinical trials requires an appropriate number of patients who fulfil given inclusion criteria. In the case of so called “orphan” diseases, such as Duchenne and Becker muscular dystrophy (DMD/BMD), the number of suitable patients within one country is usually limited. The Neuromuscular Disorders article describes the Czech experience developing a detailed registry of Czech and Slovak DMD/BMD patients. The online registry provides a detailed phenotypic and genotypic description of patients, who have access to their own data.

The main purpose of such a registry is the time-effective recruitment of eligible patients for a clinical trial or therapy and may allow the anticipation of possible future effects of appropriate therapy on individual patients. The importance of the DMD/BMD patient registries has recently also been rising with new clinical trials focused on mutation-specific approaches. Other outputs include assessment of epidemiology, phenotype and genotype relationships, or standards of care.

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American Academy of Neurology's Annual Meeting

At this year’s annual meeting of the American Academy of Neurology, 12.000 neuroscience professionals convened in Seattle, USA, from April 25 to May 2.

The AAN meeting is one of the largest neurology gatherings and platform for the latest developments in scientific and clinical research.

TREAT-NMD was represented by two poster presentations.

Joanne Auld presented the Registry of Outcome Measures and its applicability for clinical trials in various neuromuscular diseases.

The value of the TREAT-NMD patient registries for clinical trials and natural history studies was exemplified by analysis of the data derived from 500 SMA patients currently registered in the German and UK SMA patient registries; this poster was presented by Sarah Baumeister. The audience agreed that TREAT-NMD offers some important infrastructure for clinical trials and that patient registries can also be used for research into care and phenotype comparison between countries.

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Günter Scheuerbrandt's Exon Skipping Report now available in Japanese and Spanish

Günter Scheuerbrandt's exon skipping report that was featured in last month's newsletter has now been translated into both Japanese and Spanish. The translations were completed by Yumiko Yamauchi and Ricardo Rojas respectively.

These reports are now available to download from Günter's dedicated page on the website.

Günter who is currently working on a report about the Dutch Clinical Trials uses his indepth knowledge of DMD to explain exon skipping to a wider non-scientific audience.

The report explains the genetics behind the disease and also exon skipping, the most advanced genetic technique for an effective therapy of Duchenne muscular dystrophy. The report finishes with Günter talking to Dr. Kate Bushby about ongoing trials, which is also included in both translations.

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Vacancy: TREAT-NMD DMD UK Coordinator

The closing date for this post based in the TREAT-NMD Co-ordination office in Newcaslte is Wednesday 17th June 2009.

The position is funded in the first instance for 12 months by Action Duchenne

The role of this post will be to manage, develop and coordinate the dissemination and communication of TREAT-NMD activities to the UK Duchenne Muscular Dystrophy community, reporting directly to the TREAT-NMD Project Manager, as well as close links with the TREAT-NMD Project Coordinators and Action Duchenne.

It will be your responsibility to ensure that the objectives of the Network are communicated across the UK to the wider publics in a clear, concise and informative manner and so increase the public profile of the Network and its partners. You will be responsible for the facilitation of the involvement of UK clinical trial centres in upcoming DMD clinical trials by assisting the Principal investigators, identifying funding opportunities and carrying out regulatory document filing and reporting.

The UK coordinator will also be responsible for enhancing the excellence available in UK trials sites / centres by disseminating the TREAT-NMD standards of care and helping centres implement them, organising training within the framework of TREAT-NMD, where necessary. This is a diverse and challenging position for a person with excellent organisational and communication skills and an understanding of issues relating to translational medicine and clinical trials.

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Changes to ENMC Workshop Application Process

In order to establish a more efficient, standardised and transparent process for assessing workshop applications, ENMC will implement a new procedure for this, starting for workshops to be performed as of 2010.

This new procedure includes two grant review rounds per year.

For workshops to be conducted in the first half of 2010, the deadline for submitting the applications is September 15, 2009. The forms to be completed for a workshop application can be downloaded from our website www.enmc.org .

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

We do look forward to receiving your applications!

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