

welcome

Welcome to the latest TREAT-NMD newsletter. This edition includes details of a DMD animal models report published in *Neuromuscular Disorders*, news of the Action Duchenne 2009 conference, and a call for experts on care standards for Nemaline and other Congenital Myopathies to work on a consensus document. Although there has been rapid progress in the molecular genetic understanding of how these diseases develop, medical care in this area has not enjoyed such progression. The collaboration between TREAT-NMD, the Nemaline Consortium and US experts aims to address this issue.

Details of the CMD Standards of Care meeting in Brussels are also featured. With this meeting scheduled to take place immediately before our own International Conference, Brussels is definitely the place to be in November 2009! Don't forget to submit your abstract before 17 July.

Best wishes from,
Katie, Volker, Hanns, Steve, Emma, Rachel, Samantha and Michael,
the Newcastle TREAT-NMD team

at a glance...

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

21-23 Sept 2009 Muscle Study Group Annual Meeting, Buffalo, New York

25-26 Sept 2009 SMA 'at the Eve of the Cure' conference, Warsaw, Poland

05-06 Oct 2009 6th UK SMA



Seeking Experts in Standard of Care for Nemaline and other Congenital Myopathies

Experts are required to join an inter-continental collaboration between TREAT-NMD, the Nemaline Consortium and US experts to form a Committee for the Standard of Care for Nemaline and other Congenital Myopathies, specifically Central-Core, Multi-Minicore and Myotubular Myopathy. There has been rapid progress in the understanding of the molecular genetics and pathogenesis of Nemaline and other congenital myopathies in recent years. However, the level of the medical care for this group of patients has not matched the progress of the current advances in medical technology. The aim is to form a committee to look into this issue and to achieve a consensus for the standard of care for this group of patients.



The SOC Committee for Nemaline and other Congenital Myopathies is an international group of experts in this area. The effort is currently supported by "A Foundation Building Strength" - <http://www.buildingstrength.org/> - and it will include experts in pediatric neurology, genetics and diagnostics, pulmonary, orthopedics/physical therapy and rehabilitation, and gastroenterology/nutrition and speech pathology. Each of these disciplines will form a working group to work on the recommendation for that specific care area.

Delphi survey techniques to solicit experts' opinion on the care issues will be used. There are plans to hold a workshop at Stanford University by the Spring of 2010 to discuss the results of this survey and to achieve consensus for the recommendation for each care area. Periodic conference calls will be held to discuss the progress in this project. It is anticipated that this project will take about two years and that the final product will be a published Consensus Statement for the Standard of Care for Nemaline and other Congenital Myopathies.

Currently there is a particular need for experts in gastroenterology/nutrition, and speech pathologists with expertise in these congenital myopathies to join the committee.

If you are interested in participating please contact the Chair of the Committee, Dr. Ching H. Wang at his e-mail address: wangch@stanford.edu.

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CMD Standard of Care Conference

Immediately prior to the TREAT-NMD international conference this November in Brussels, a conference on CMD care standards will take place. The CMD Standard of Care (SOC) initiative was launched at a recent TREAT-NMD sponsored meeting in April 2009, bringing together international CMD experts, Cure CMD (a patient advocacy group) and TREAT-NMD staff with participation by Sharon Hesterlee (MDA). The meeting recognized the critical need to address SOC as part of the larger effort to promote CMD clinical trial readiness, decreasing variability in CMD medical practice patterns, evaluating CMD diagnostics in a variety of international settings and delineating the CMD medical care issues with an emphasis on a multidisciplinary approach.



In order to define the CMDs, the meeting participants defined the CMDs as a group based on their clinical presentation as neuromuscular disorders with congenital and early childhood onset presenting

Research Conference, Edinburgh, UK

23-24 Oct 2009 Action Duchenne International Conference Holiday Inn Bloomsbury London

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

with hypotonia, contractures, myopathic or dystrophic muscle biopsy findings and a variable CK. This definition recognizes that the line between the CMDs, congenital myopathies and LGMDs is blurred with a gene classification approach demonstrating a gene specific spectrum of disease with clinical heterogeneity. The current genes included in the CMD definition include: the collagen VI myopathies, Ullrich CMD and Bethlem myopathy (col6a1, col6a2, col6a3), merosin deficient CMD (LAMA2), the dystroglycanopathies (FKRP, fukutin, LARGE, POMT1, POMT2, POMGnT1), Rigid Spine muscular dystrophy (SEPN1), alpha 9 integrin and lamin A/C mutations with CMD presentation.

The CMD SOC effort is being led by Dr. Thomas Sejersen, Dr. Ching Wang and Dr. Anne Rutkowski. Drs. Sejersen and Wang have already collaborated to produce the spinal muscular atrophy (SMA) SOC guidelines. Dr. Wang established a credible and rigorous approach to developing the SMA SOC guidelines using a modified Delphi methodology. Dr. Anne Rutkowski is Vice Chair of Cure CMD, a nonprofit CMD patient advocacy group. Cure CMD will launch an international CMD patient registry (CMDIR) in July 2009.

The CMD SOC initiative will draw upon international experts in the following subspecialty areas: pulmonary/ICU, GI/nutrition, cardiac, orthopedics, neurology, dental care, palliative care and diagnostics. Each subspecialty group will have a CMD expert neurologist to provide guidance by focusing the group upon CMD specific care issues while underscoring CMD heterogeneity. Nomination to the subspecialty areas occurred through a survey of recognized CMD neurologist leaders and an advertisement placed in the TREAT-NMD newsletter.

At the upcoming July 2009 conference, Therapeutic Targets in the CMDs, a small group of committed CMD neurologists will convene to review the methodology, SOC design and delineate the prototype worksheet for the first SOC survey. The survey will be sent through email discussion groups to address integral CMD care issues, barriers and unknowns. A follow up survey will summarize key care issues identified and solicit current practice patterns, incorporating varied global medical practice settings and resources.

A November conference (November 14th-16th) in Brussels will build upon the first survey with in depth reviews of available literature, patient databases and practice patterns. A total of 45 participants numbering 5-6 from each subspecialty area with MDA representation will convene to review preliminary surveys regarding specific care issues and current practice patterns in light of disease heterogeneity. Conference participants will be chosen on the basis of level of expertise and involvement in subspecialty email discussion groups. The conference will place a premium on structured discussion both within each subspecialty and as a multidisciplinary team focused on improving quality of life and prognosis for those with CMD.

The deliverables for the November conference include:

- Poster presentation of conference outcomes by Dr. Susan Iannaccone at the subsequent NIH-TREAT-NMD conference: Bringing Down the Barriers, Nov 17-19th, Brussels
- Draft of a preliminary CMD Standard of Care document to be finalized and published (April 2010)
- Draft of a preliminary lay friendly CMD SOC document to be posted on advocacy websites (April 2010)
- Draft of a document in parallel to review current CMD diagnostic strategies and approaches, including the diagnostic algorithm from the University of Iowa CMD conference, summer 2008.

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Pre-clinical testing for DMD - workshop report now available online

Kanneboyina Nagaraju and Raffaella Willmann on behalf of the TREAT-NMD Network and the Wellstone Muscular Dystrophy Cooperative Research Network have produced a report from the Pre-Clinical Testing for Duchenne Dystrophy Workshops entitled "Developing standard procedures for murine and canine efficacy studies of DMD therapeutics". This report has now been accepted by Neuromuscular Disorders and is available online.



Held in Washington DC and Zurich, the two workshops organised jointly by TREAT-NMD and the Wellstone Muscular Dystrophy Center at the Children's National Medical Center in Washington, DC focused on the standardization of protocols (SOPs) for recommended models i.e. mice and dogs to evaluate Duchenne Muscular Dystrophy therapeutics.

Topics covered in the report include:

- Animal models, methods and variability in pre-clinical evaluations
- Muscle function and histology readouts in mdx mice, toxicology evaluations
- Prioritization of endpoints and assignments of draft protocols
- Cardiac and respiratory function, histology, biochemistry, electrophysiology readouts in mdx mice

The report is available online [here](#) and as a PDF [here](#).

Finalized SOPs are available on the TREAT-NMD website via a link that allows download of protocols and contact to the author for questions and suggestions - <http://www.treat-nmd.eu/research/preclinical/SOPs/>

All researchers involved in use of Duchenne models are encouraged to make use of these protocols and to assist with their future refinement.

In the future it is intended that they will also be posted on the NIH Wellstone Portal. The SOPs will be updated annually by the author and reviewer, taking into consideration feedback and suggestions that they may have received from users.

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Action Duchenne International Conference

The annual Action Duchenne International Conference is scheduled for 23 - 24 October 2009. This two day conference will be held in the Holiday Inn, Bloomsbury London.

This is the 7th annual conference from Action Duchenne and will include top researchers and experts in Duchenne. Here you will have the chance to meet with other families and learn of all the up to date research and medical care as well as practical workshops on physiotherapy, learning and behaviour and much more. A special TREAT-NMD session will include speakers and workshops on the latest international initiatives in Duchenne. More details will be available shortly from www.actionduchenne.org



Action Duchenne International Conference
Holiday Inn,
Bloomsbury,
London
23 October 2009 - 24 October 2009

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