

**TREAT-NMD Partner Newsletter No. 21
and Club of Interest Newsletter No. 15**

27th July 2007

Welcome to the fifteenth newsletter for the TREAT-NMD Club of Interest. This week's edition will be the last one for 5 weeks. We are having a break from the newsletter during August and we will be back on the 7th September to re-launch the newsletter.

This week's newsletter features a report from the Governing Board meeting with pictures (see if you can spot anyone you know) as well as information on a number of upcoming meetings and summer schools.

We hope you have a relaxing and enjoyable summer and look forward to the re-launch of the TREAT-NMD newsletter. In the meantime if you have any suggestions or comments please feel free to e-mail the coordination team.

Best wishes,

Katie, Volker, Stephen, Emma, Arron and Rachel – the TREAT-NMD coordination team

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1. About this newsletter

This is a weekly newsletter sent to all members of TREAT-NMD's "Club of Interest" worldwide. We are receiving new subscriptions all the time, so if you've missed the earlier editions of the newsletter and would like to catch up, please visit our newsletter archive online at <http://www.treat-nmd.eu/news/newsletter/> where you will find all the back-issues. If you have received this from a friend or colleague and would like to subscribe directly, please visit our website at <http://www.treat-nmd.eu/> where you will find a subscription form at the bottom of the homepage. You can also use the same form if you no longer wish to receive this newsletter – just select the unsubscribe button.

2. Working with us

TREAT-NMD aims to be an inclusive rather than an exclusive network, and you do not have to be based in Europe or be a partner to be involved. International collaboration with experts from all over the world is already taking place, and new links are being developed.

If you are involved in any of TREAT-NMD's areas of interest and have something you'd like to say or a suggestion of where we could work together, we encourage you to get in touch by writing to us at info@treat-nmd.eu. The coordination team in Newcastle will be happy to put you in touch with the person most relevant to your particular interest.

3. TREAT-NMD news and reports

Report from the TREAT-NMD Governing Board Meeting, 2-3 July 2007, Naarden, The Netherlands

By Stephen Lynn

This was the first meeting of the Governing Board since the Kick-Off Meeting back in January 2007. The one and a half-day meeting covered a wide and varied agenda that highlighted the impressive progress made by the network in the first six months. A welcome reception was held on Sunday evening to give the partners an opportunity to view posters produced by partners that highlighted their respective activity/workpackage achievements.



The poster-walk session proved to be very popular with the Board. This was an opportunity to discuss the progress made by the partners and to present their achievements in an easily digestible form. An extended poster session will be incorporated into future Governing Board meetings to allow partners to network and discuss their activities and achievements with each other.

Professor Kate Bushby provided some opening comments to the meeting in which she stated that the network partners were committed and working extremely hard to achieve the goals of the network. She also described the immense interest the network is receiving from other groups and countries around the world and how we are already thinking globally, and not just Europe!

The meeting began with a review of the network's Joint Programmes of Activities – covering Integration, Research, and the Spreading of Excellence. The review of the Joint Programme on Integration demonstrated how the TREAT-NMD web site has developed over the first six months, but more importantly, how the web site will be instrumental in the integration and communication of the network's activities both with partners and with the wider community. This will be an important tool that we will continue to develop and improve – and we need your input to help us achieve this. Professor Hanns Lochmüller gave an overview of the TREAT-NMD registry and the how this will integrate national registries into a European-wide database – which is



currently lacking for the industrial and research communities. A TREAT-NMD registry charter will be drafted to assist in the management and operation of the European registry.

The review of the Joint Programme of Research highlighted the need for a large-scale production facility for viral vectors. A 4,800 m² plant will be built and delivered by 2011. This €26 million facility will be funded by AFM, Région Ile de France, and Département Essonne.



The review of the Joint Programme of Spreading Excellence gave an overview of the training and education opportunities that could be incorporated into the network, as well as the potential development of an International Master's Course in Neuromyology. Plans are also in hand for TREAT-NMD to be represented at patient-focussed scientific meetings later this year to discuss ethical and regulatory issues and how they relate to patients with neuromuscular disease. This Joint Programme is also working hard to extend the network to Eastern Europe with discussions and visits with various countries already taken place.

During the Governing Board meeting we were honoured to have two members of our Scientific and Technological Advisory Committee present, Dr John Porter, Programme Director at the NINDS (NIH) and Dr Ségolène Aymé, Director of Research at Orphanet and leader of the EC Rare Diseases Task Force. Dr Aymé gave a brief overview of a communication on rare diseases that is currently in the drafting phase. This communication will address the support required to deal with rare diseases as well as what action the Community will take regarding improving health services for patients by increasing cooperation between agencies such as the EMEA, DG Research and DG SANCO. It is hoped that a final draft will be ready by the end of the year for consultation and will be adopted in 2008.

The Governing Board made a number of recommendations – one being the establishment of an Education and Training Task Force. Under the leadership of the ENMC, this task force will identify training and education opportunities, as well as schemes to allow the mobility of personnel between institutions within Europe and outside the European Community. The task force will begin to identify the gap that exists between supply and demand and define an action plan to address this issue.



The network has already attracted a great deal of interest from the international community, with people from over 68 countries having visited the web site to date, and a number of countries expressing interest in participating in the network. There is a need to enable interested parties to participate in the network and various ways to recognise such membership were discussed and will be implemented. Also to address this interest it was decided to implement a task force to help organise a large international meeting/workshop to be held in late 2009. It was proposed to hold an International Congress on Translational Research in Muscular Dystrophies and SMA. Delegates from academia, clinical, industry and patient representatives would be included. Dr John Porter indicated that the NIH would be willing to help with funding for such a large international meeting.



Some of the partners felt that the frequency of the TREAT-NMD newsletter was too often. However, others have reported strong support for keeping the status quo. Therefore, in order not to clog email inboxes, but still maintain a strong communication link with our readers, we have decided to publish the newsletter fortnightly. This newsletter will be the last weekly edition before we take a summer break, and we will be back with our fortnightly newsletter beginning Friday 7th September 2007. In the meantime, the Coordination Office in Newcastle will send out any important information for partners and supporters via brief e-mail notifications.



Thank you for all your support and input over the last 21 weekly newsletters – we appreciate all your comments and suggestions regarding this newsletter. Finally, from all the team here in Newcastle we want to wish you all a relaxing summer.

4. Funding calls



FP7 Call for Proposals (Capacities Work Programme: Part I – Infrastructures) Call identifier: FP7-INFRASTRUCTURES-2007-2

20 September 2007 at 17.00 Brussels local time
Indicative budget 64 million EUR

Line of action/Activity	Topics called	Funding scheme(s)	€ (million) indicative
1.1 Support to existing research infrastructures			
1.1.2 ICT based e-Infrastructures	INFRA-2007-1.2.3: e-Science Grid infrastructures	Combination Collaborative projects & Coordination and support actions	50
1.3 Support to policy development and programme implementation			
INFRA-2007-3.3: Studies, conferences and coordination actions supporting policy development, including international cooperation, for e-Infrastructures		Coordination and support actions	14

All the documents for making an application are at:

http://cordis.europa.eu/fp7/dc/index.cfm?fuseaction=UserSite.CapacitiesDetailsCallPage&call_id=66

5. Upcoming Conferences, Workshops, Meetings and Summer Schools



MOLEDA Summer School
Non-viral gene transfer into muscle and skin
16-22 September, 2007
Evry – Maison Alfort – Paris (France)

The new deadline for registration and abstract submission is August 27th, 2007
For Further information please visit: www.moleda.org

:: Institut de Myologie



11th Summer School of Myology
19-27 June, 2008
Institut de Myologie, Paris, France
<http://www.institut-myologie.org/>

3rd International Myology congress
26-30 May, 2008
Marseille, France
Organized by the AFM
Thomas Voit will chair the congress and Ketty Schwartz will be the honorary president.
Details to follow

5th International Congress of Rehabilitation
30 May- 1 June, 2008
Marseille, France
Registration details will follow
For further information or if you would like to present a poster, please contact
Jean Claude Riou, jcriou@afm.genethon.fr before October 15th



57th Annual Meeting of the American Society of Human Genetics
23-27 October, 2007
San Diego, California
<http://www.ashg.org/genetics/ashg/menu-anmeet.shtml>



6th International Myotonic Dystrophy Consortium Meeting
12-15 September 12-15, 2007,
University of Milan, Milan, Italy
<http://www.idmc-6.com/default.html>

6. Pick of the publications

Antisense-induced exon skipping for duplications in Duchenne muscular dystrophy Annemieke Aartsma-Rus¹, Anneke AM Janson², Gert-Jan B van Ommen¹ and Judith CT van Deutekom²



In this report exon skipping for patients with duplications is described for the first time. Interestingly, for patients with single exon duplications, exon skipping has the potential to generate wild type transcripts and dystrophins.

Abstract

Background

Antisense-mediated exon skipping is currently one of the most promising therapeutic approaches for Duchenne muscular dystrophy (DMD). Using antisense oligonucleotides (AONs) targeting specific exons the DMD reading frame is restored and partially functional dystrophins are produced. Following proof of concept in cultured muscle cells from patients with various deletions and point mutations, we now focus on single and multiple exon duplications. These mutations are in principle ideal targets for this approach since the specific skipping of duplicated exons would generate original, full-length transcripts.

Methods

Cultured muscle cells from DMD patients carrying duplications were transfected with AONs targeting the duplicated exons, and the dystrophin RNA and protein were analyzed.

Results



For two brothers with an exon 44 duplication, skipping was, even at suboptimal transfection conditions, so efficient that both exons 44 were skipped, thus generating, once more, an out-of-frame transcript. In such cases, one may resort to multi-exon skipping to restore the reading frame, as is shown here by inducing skipping of exon 43 and both exons 44. By contrast, in cells from a patient with an exon 45 duplication we were able to induce single exon 45 skipping, which allowed restoration of wild type dystrophin. The correction of a larger duplication (involving exons 52 to 62), by combinations of AONs targeting the outer exons, appeared problematic due to inefficient skipping and mistargeting of original instead of duplicated exons.

Conclusion

The correction of DMD duplications by exon skipping depends on the specific exons targeted. Its options vary from the ideal one, restoring for the first time the true, wild type dystrophin, to requiring more 'classical' skipping strategies, while the correction of multi-exon deletions may need the design of tailored approaches.

To View the full article please visit; <http://www.biomedcentral.com/1471-2350/8/43>

7. Job opportunities

Physical therapist position, Laboratory of physiology and neuromuscular evaluation, Myology Institute

The Myology Institute (Pitié-Salpêtrière hospital) is seeking a part-time or full-time physical therapist (renewable temporary contract that could lead to a permanent contract) to perform quantified muscle force measurements within the framework of clinical research protocols for certain neuromuscular pathologies. Training given.

Contact: jy.hogrel@institut-myologie.org

The Institute Charles Delaunay (Troyes University of technology) and the Laboratory of Neuromuscular Physiology (Myology Institute, Paris) is seeking a PhD student

Subject: Motor Unit Number Estimation (MUNE) and EMG decomposition: Application of Blind Source Separation (BSS) and comparison with other methods

Laboratory : Institute Charles Delaunay - Troyes University of technology
Laboratory of Neuromuscular Physiology-Myology Institute, Paris

PhD supervisors: Jacques Duchêne (Troyes) and Jean-Yves Hogrel (Paris)

Application deadline: July 31, 2007

Contact Myology Institute: jy.hogrel@institut-myologie.org

Contact UTT: jacques.duchene@utt.fr

The TREAT-NMD website now features a job opportunities page. Please visit <http://www.treat-nmd.eu/jobs.htm> to view all current job opportunities.

If you would like to advertise a relevant job opportunity on our site and in our newsletter, please write to rachel.thompson@treat-nmd.eu

8. Partner-specific items

Discussion forums / lists

Would you like us to set up a discussion forum for you on the TREAT-NMD website?

If so, please e-mail rachel.thompson@treat-nmd.eu.

Calls for proposals / funding opportunities

Please forward to us at the Coordination Office any calls for proposals and funding opportunities you receive within your institution. We will then advertise these in the newsletter and on the website.

Deliverable reports



We would like to thank the partners who contributed to and submitted their 6 month deliverable reports. We will be forwarding these to the EU in due course.

9. Send us your news and views!

We strongly encourage all partners and supporters to send their own news and updates and we will be happy to include them in future editions of the newsletter. Please send your contributions to emma.heslop@treat-nmd.eu

