

**TREAT-NMD Partner Newsletter No. 17  
and Club of Interest Newsletter No. 11**

**29<sup>th</sup> June 2007**

Welcome to the eleventh newsletter for the TREAT-NMD Club of Interest. This week's edition features a report on the MD-NET annual meeting and an interesting overview of one of MD-NET's service structures, the centre for microsatellite analysis in Dresden (see section 3).

As readers are aware, we have been busy organising the first TREAT-NMD Governing Board Meeting, which will run from 1–3 July. This will be a great opportunity for all partners to get together to share information on the progress of their activities during our first six months and to make decisions about the future work of the network. A full report of the meeting will appear in a later edition of the newsletter. We look forward to seeing all the partners in Naarden this weekend!

We hope you enjoy the newsletter and look forward to hearing your comments – write to [info@treat-nmd.eu](mailto:info@treat-nmd.eu) with anything you'd like to say. Feel free to forward this message to anybody you think might find it of interest, or invite them to sign up to receive the newsletter by visiting our website.

Best wishes,

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

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- A satellite-style map of Europe and surrounding regions, showing landmasses in green and brown, and oceans in blue. The map is slightly faded and serves as a background for the newsletter contents list.

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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 back-issues. If you have received this letter 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.

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## 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](mailto:info@treat-nmd.eu)

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## 3. TREAT-NMD news and reports

### Microsatellite analyses as an economic tool for diagnosis in neuromuscular disorders

Angela Huebner & Maja von der Hagen, Dresden, Germany

Neuromuscular disorders (NMDs) represent a genetically and phenotypically heterogeneous group of inherited disorders comprising muscular dystrophies (MD), myopathies, congenital myasthenic syndromes (CMS), and myotonias. To date, more than 50 different gene loci have been linked to muscular dystrophies and myopathies which represent the main group of NMDs. The growing number of candidate genes for phenotypically similar diseases has raised the demand for economic diagnostic strategies. As one of the main core facilities of the German MD-NET we have developed DNA marker sets for 75 gene loci which can be applied for haplotype analyses in families with unclassified NMDs. In the past, this marker set has proved highly effective for the further narrowing of the number of differential diagnoses and for the characterization of the molecular defect in informative families with extensively prediagnosed but yet unclassified NMDs. So far we investigated 236 informative families and were able to reduce the number of candidate genes to one or two gene loci in 40 % of the families, and in about 30 % of all families the disease-causing mutation was subsequently identified. Informative families in whom all known candidate genes are excluded can be transferred to a genome wide linkage scan. This offers the possibility to detect and characterize novel genes involved in the regulation of muscle function.

We would like to present this facility to all TREAT-NMD partners and also to the wider Club of Interest as we think that this diagnostic service structure could be of value for some of the workpackages within TREAT-NMD. We are interested in contributing to a European Network of high throughput, reliable and cost effective diagnostic tests and would be prepared to participate in the FP7 diagnostic call.

We constantly update the screening sets with novel candidate gene loci and also offer the establishment of this diagnostic tool on special requests.

Additional information about our service centre can be found in our recent publication in Neuromuscular Disorders 2006 (see [http://www.treat-nmd.eu/assets/documents/Linkage\\_analysis\\_NMD.pdf](http://www.treat-nmd.eu/assets/documents/Linkage_analysis_NMD.pdf)) and in the list of gene loci currently integrated in our diagnostic tool. The list is available on the TREAT-NMD website here: [http://www.treat-nmd.eu/assets/documents/screening\\_sets.pdf](http://www.treat-nmd.eu/assets/documents/screening_sets.pdf)

We would be very happy to provide further information on our facilities to any interested parties. Please email [angela.huebner@uniklinikum-dresden.de](mailto:angela.huebner@uniklinikum-dresden.de) with any questions.



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## MD-NET Annual Meeting

Report by Hanns Lochmüller and Sarah Baumeister

On 8–10 June 2007, the annual meeting of the German Muscular Dystrophy Network (MD-NET; [www.md-net.org](http://www.md-net.org)) took place in Wuerzburg. MD-NET is partner 4 within TREAT-NMD and is involved in several workpackages, taking major responsibility as activity leader for activities A04 (biobanks and patient databases) and A05 (clinical trials coordination centre).

MD-NET was established in 2003 as a network of medical professionals and scientists from different areas to collaborate on muscular dystrophy research. The network's objectives are to deepen understanding of the pathogenesis of muscular dystrophies, optimize diagnostic procedures and develop effective treatment regimes. In order to achieve these goals, national service structures have been formed and expanded and MD-NET has successfully set up two controlled clinical trials with 15 national and 2 international clinical centres participating. MD-NET has established five service structures, which form the backbone of the network. Research in muscular dystrophies is coordinated and facilitated by the Muscle Tissue Culture Collection in Munich, the centre for microsatellite analysis in Dresden, the central facilities for the sequencing of genes involved in muscular dystrophies in Wuerzburg and Bochum, and the Coordination Centre for Clinical Trials in Freiburg. MD-NET service structures are well known beyond national borders and the Muscle Tissue Culture Collection has been part of a European network (Eurobiobank) since 2003.

One of the key themes at this year's annual meeting was TREAT-NMD and its implementation in Germany via MD-NET. Volker Straub, TREAT-NMD coordinator and, together with Hanns Lochmüller, coordinator of MD-NET, presented the structure and objectives of TREAT-NMD. Activity Leaders Rudolf Korinthenberg (A05) and Hanns Lochmüller (A04) reported on their Activities and showed the results that have already been achieved. MD-NET members were generally very interested in these efforts and several offered their expertise and active participation for future endeavours. Representatives from the German Duchenne Parent Project "Benni & Co" were excited about the progress made and offered additional support, especially for the upcoming TREAT-NMD DMD patients' registry. However, some scientists raised concerns that the main focus of TREAT-NMD funding is on building and maintaining administrative structures rather than "bench-side" projects. The coordinators responded that these structures will greatly aid clinicians and scientists to obtain additional funding for their research projects.

The two controlled clinical trials run by MD-NET, R17, a clinical trial on immunosuppressive treatment in Duchenne muscular dystrophy, and R19, a clinical trial on Deflazacort on dysferlinopathies, are still ongoing. R17 reached the recruitment goal of 150 DMD patients in February 2007 and will be completed by summer 2008. Several MD-NET research projects were presented, highlighting scientific excellence and the benefit of collaboration within MD-NET. As an invited speaker Christelle Etard (Karlsruhe) talked about "Roles of chaperone proteins in muscle development: insights from Zebrafish".

The future of MD-NET was another major topic at the meeting, since funding from the German Ministry for Science and Education will come to an end in 2008. It was stressed that MD-NET, now a registered association, will continue to thrive beyond this date. However, efforts are needed to obtain new funding. Future plans of MD-NET scientists include the establishment of a research group working on myofibrillar myopathies. The 2008 MD-NET meeting will be organized by the Department of Neuropathology in Aachen and chaired by J. Weis.

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## 4. Upcoming conferences, meetings and workshops



UPPMD Global Registry Meeting  
Amsterdam, The Netherlands  
29<sup>th</sup>-30<sup>th</sup> June 2007





**TREAT-NMD Governing Board meeting**  
Naarden, The Netherlands  
1–3 July 2007



**Outcome measures for experimental studies in Duchenne muscular dystrophy**  
Naarden, The Netherlands.  
30 June–1 July 2007



**Conference on Clinical research for Rare Diseases**  
Bethesda, Maryland, USA  
5 September 2007



**European Society for Muscle Research**  
XXXVIth EMC meeting of the European Society for Muscle Research  
Stockholm, Sweden September 8–12, 2007  
<http://www.esmr.org/EMCSth1.htm>  
Including session together with the European Muscle Development Network  
(MyoRes, <http://myores.org/site/intranet/events/show/7>)

**European Workshop on Centres of Reference for Rare Diseases**  
Prague, Czech Republic.  
13–14 July 2007

**Rare Diseases Research: Building on Success - a European Conference**  
Charlemagne building- Brussels, Belgium  
13 September 2007

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## 5. Pick of the articles

The TREAT-NMD newsletter occasionally highlights news and research that may have implications for patients with neuromuscular diseases.

### Two companies collaborate to develop rare disease medicines

AVI BioPharma and Ercole Biotech have announced a cross-licensing and collaboration agreement to develop medicines that may prove effective in treating genetic diseases Duchenne muscular dystrophy (DMD) and beta thalassemia. Under the terms of the agreement, AVI and Ercole will work together in the development of products, with AVI leading the DMD program and Ercole leading the thalassemia program. Each company has the option of co-funding the program led by the other party and sharing equally in the financial returns from resulting products. The technologies developed by Ercole and AVI allow manipulation of the RNA splicing process and the production by cells of clinically desirable variants of relevant proteins.

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## 6. Partner-specific items

### Governing Board meeting

The TREAT-NMD Governing Board meeting starts this coming Sunday (1 July). A report will appear in a future edition of this newsletter. We look forward to seeing you all in Naarden!

### 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](mailto: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.

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### **7. 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](mailto:emma.heslop@treat-nmd.eu)

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