# TREAT-NMD Partner Newsletter No. 4 30 March 2007

Welcome to the 4th weekly newsletter for all TREAT-NMD partners!

**Newsletter contents**



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**1. About this newsletter and the mailing list**

You are a "key contact" for your partner organisation and you have been subscribed to a mailing list called "treatnmd-partners", which is intended to be a simple way for partners to keep in touch with one another. If you ever want to write a message to reach the key contacts in every partner organisation, write to treatnmd-partners@newcastle.ac.uk and your message will automatically be distributed to everyone.

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**2. Send us your news and views!**

We encourage all partners to send their own news and updates, either directly to all partners by writing to this list at treatnmd-partners@newcastle.ac.uk or to emma.heslop@newcastle.ac.uk for inclusion in the next newsletter. What else would you like us to include in the newsletter? Write to emma.heslop@newcastle.ac.uk with your feedback.
Please e-mail us with any information you have on upcoming education and training opportunities including workshops, conferences, funding, exchange programmes, clinical placements, visiting professorships and lectureships.

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**3. Publicising TREAT-NMD**

As part of efforts to raise the awareness of and publicise TREAT-NMD at conferences, workshops and other meetings we have produced a general double-sided colour flyer introducing TREAT-NMD and detailing the TREAT-NMD partner organisations. The flyer is now available for download from the TREAT-NMD web site at http://www.treat-nmd.eu/assets/documents/TREAT-NMD_Flyer.pdf. We have also added an editable version

in Microsoft Publisher format (higher resolution, better for those intending to print it), which you can download and have printed yourself at http://www.treat-nmd.eu/public_html/private/docs/TREAT-NMD_Flyer.pub

If you are planning to attend any workshops, conferences or meetings please let us know and please take our promotional material along to help promote TREAT-NMD. The TREAT-NMD logo is available to download from the partners area of the website at http://www.treat-nmd.eu/private/ (please contact r.h.thompson@ncl.ac.uk if you need to be provided with login details). Please also ensure that you include it on any abstract, papers and posters you prepare in which you mention TREAT-NMD.

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**4. Resources**

The TREAT-NMD website is growing all the time to provide a useful resource for partners – for reporting on activities, for creating email forums, uploading presentations and conference/workshop notifications.

Over the coming months the partners area of the website will undergo a complete transformation to provide a fully fledged "intranet" for partners. Further details of this will be published in next week's newsletter.

Now that our IT support officer is in place, we will be revamping the "public" face of the website too. You will shortly be receiving forms to fill in so that we can improve the sections giving details of what each partner is involved in. In the meantime, if you have an opinion on what else should appear on the site (either as a "public" page so all site visitors are informed about TREAT-NMD in the way you'd like them to be, or as a private facility within the partners area), please do let us know.

The following documents have been uploaded to the TREAT-NMD website and are now available to download.

Final version of the lay statement on the AON workshop:
http://www.treat-nmd.eu/assets/documents/Lay_statement_AON_2007.pdf

Back issues of the newsletter:
http://www.treat-nmd.eu/assets/documents/TREAT-NMD_partner_newsletter_No1.pdf
http://www.treat-nmd.eu/assets/documents/TREAT-NMD_partner_newsletter_No2.pdf
http://www.treat-nmd.eu/assets/documents/TREAT-NMD_partner_newsletter_No3.pdf

Minutes from the kick-off meeting:
http://www.treat-nmd.eu/private/pdf/Minutes_KOM_18-19.01.2007_v7.pdf

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**5. TREAT-NMD link from your website**

In an effort to increase the profile of TREAT-NMD we are asking partners to add a link to the TREAT-NMD website from their existing website. Many of the partners have already done this – thank you! For those of you who have not, we would be very grateful if you could arrange for a link to be created.

If you would like us to provide you with a snippet of html code for the link that can simply be copied and pasted into your website, please contact r.h.thompson@ncl.ac.uk. Alternatively the TREAT-NMD logo is available on the website (http://www.treat-nmd.eu/private/) for you to create your own hyperlink.

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**6. 3-Monthly Report – deadline 9th April**

Bénédicte Charrin from ACIES recently emailed all Activity Leaders asking them to complete and submit a 3 monthly report. We would like to remind you that it is the Activity Leaders' responsibility to ensure that the

Work Package Leaders (for the activity that they are responsible for) submit their reports to the AL and that the AL completes the form and returns it to Bénédicte Charrin (eu-new@acies.fr) before 9th April 2007.

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**7. 'Fact finding Questionnaire'**

The deadline for questions to be included into the 'fact finding questionnaire' has now passed. We would like to thank the people who contributed and assure you we will be issuing the questionnaire to all partners within the next few weeks. We look forward to your responses.

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**8. Training opportunity**

The EuroBioBank network (www.eurobiobank.org) coordinated by Eurordis offers specialised training working with neuromuscular human biomaterial. Prof. Dr. Hanns Lochmüller, MTCC, University of Munich, Germany, and A04 leader, can organise training with primary human muscle cells.

Myoblast training courses are available in Munich for TREAT-NMD members during the following weeks:
06.08.2007 - 10.08.2007
15.10.2007 - 19.10.2007

Prof. Lochmüller is also willing to coordinate a limited number of courses "on demand" to accommodate the calendars of the trainees. If any TREAT-NMD partners are interested can they please email Prof. Lochmüller at hanns.lochmueller@med.uni-muenchen.de

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**9. Introducing our TREAT-NMD UK Coordinator**

Elaine Scott, research physiotherapist and project coordinator from the Muscular Dystrophy Campaign, will work alongside the TREAT-NMD team based at the University of Newcastle, the new MRC Centre for Translational Research at London and Newcastle and other involved partners.

The coordinator role in the UK will facilitate the implementation of TREAT-NMD network priorities such as cohesion of registries and databases, clinical trial networks, development and implementation of standards of care and outcome measures, and UK contribution to Biobanks. This post will ensure the UK's full involvement in these exciting activities, with harmonisation of workload and lack of duplication of effort. This will allow the better integration of UK clinicians and other groups into the TREAT-NMD effort.

Activities will include:

- Development of networks within the UK for delivery of clinical trials in neuromuscular diseases.
- Contribution to the development of patient databases within the TREAT-NMD network activity.
- Contribution to the activities within TREAT-NMD on the generation of outcome measures, standards of care and diagnosis.
- Dissemination of these standards within the UK clinical and patient communities.
- Dissemination of information on Biobank activities, co-ordination of UK contributions.

Clearly, many of the issues Elaine will be dealing with will be of relevance in other countries too, and any partners who are facing similar coordination issues in their own countries are welcome to contact Elaine at e_scott@btopenworld.com for discussion.

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**10. The Scientific and Technological Advisory Council (STAC)**

**List of Members:**

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| John Porter | NIH/NINDS (USA) |
| Kenneth Fischbeck | NIH (USA) |
| Maria Grazia Roncarolo | HSR-TIGET (Italy) |
| Martin McGlynn | StemCells Inc (USA) |
| Roland Foisner | Max F. Perutz Laboratories (Austria) |
| Serge Herson | Université Pierre & Marie Curie (France) |
| Krzysztof Jagla / Christophe Marcelle | Myores (France) |
| Boris Sustarsic | EAMDA (Slovenia) |
| Pat Furlong | PPMD, MDA (USA) |
| Marianne De Visser | University of Amsterdam (Netherlands) |
| Ségolène Aymé | Orphanet (France) |

**John Porter, Program Director, Extramural Research Program, NIH/NINDS**

Dr. John Porter is Program Director for Neuromuscular Disease at the National Institute for Neurological Disorders and Stroke (NINDS). He directs an extramural research grant program for the NINDS that funds and manages research grants and cooperative agreements in the areas of basic and translational research in neuromuscular disease. His program focuses on inherited and acquired peripheral neuropathies, normal biology of and disorders affecting the neuromuscular junction, and neuromuscular disorders affecting skeletal muscle proper, including myotonic dystrophy and Duchenne/Becker, facioscapulohumeral, congenital, limb girdle, oculopharyngeal, and Emery-Dreifus muscular dystrophies. He also serves as executive secretary for the interagency Muscular Dystrophy Coordinating Committee. Prior to joining the NIH, Dr. Porter was Professor of Neurology at Case Western Reserve University. Dr. Porter received his undergraduate degree in Biology from the College of William and Mary and his Ph.D. in Anatomy from Medical College of Virginia and completed postdoctoral training in systems neurophysiology at the University of Alabama at Birmingham. His research focused upon extraocular muscle biology in health and disease, including the mechanisms responsible for its novel responses to a variety of neuromuscular disorders. Dr. Porter continues to consult on gene expression study design and bioinformatics for projects in the areas of muscle development and disease.

**Kenneth Fischbeck, Adjunct Investigator, National Human Genome Research Institute, Senior Investigator Neurogenetics Branch, NIH/NINDS**

As part of the National Institute of Neurological Disorders and Stroke (NINDS), Dr. Fischbeck's laboratory is studying the mechanisms of hereditary neurological and neuromuscular disorders, particularly the polyglutamine expansion neurodegenerative diseases. His laboratory, in the Neurogenetics Branch of NINDS, investigates the causes of hereditary neurological diseases, with the goal of developing effective treatments for these disorders. Particular areas of research interest in the Fischbeck lab include the polyglutamine expansion diseases (Huntington's disease, Kennedy's disease and spinocerebellar ataxia), spinal muscular atrophy, Charcot-Marie-Tooth disease, muscular dystrophy, hereditary motor neuron disease, and Friedreich's ataxia. The disease mechanisms are studied in cell culture and other model systems.

**Maria Grazia Roncarolo, Director, The San Raffaele Telethon Institute for Gene Therapy (HSR-TIGET)**

The overall goal of the Pediatric Clinical Research Unit is the application of basic research discoveries to the treatment of genetic diseases, focusing on primary immunodeficiencies and metabolic diseases. This Pediatric Clinical Research Unit (PCRU) was established through a dedicated grant of the Fondazione Telethon to the San Raffaele Telethon Institute for Gene Therapy (HSR-TIGET). The Unit is led by Prof Maria Grazia Roncarolo and operates at HSR-TIGET, and at the Divisions of Hematology, Pediatrics and Neurology, through dedicated medical personnel and trained nursing and technical staff. It provides a suitable environment to translate the results from basic and clinical research into clinical practice and viceversa. Patients with primary immunodeficiencies are seen first at our "Pediatric Immunology" clinic of the San Raffaele Hospital. Children affected by primary immunodeficiencies are offered advanced diagnostic tests, immunological follow-up and adequate therapies.

Recent Scientific career

- 11/94-10/01 Associate Professor in Pediatrics, Department of Pediatrics, School of Medicine, University of Turin, Turin, Italy.

- 11/01-present Associate Professor in Pediatrics, School of Medicine and Surgery, University Vita-Salute San Raffaele, Milan, Italy.
- 2/98-12/98 Director of Cellular Therapy Laboratory, Telethon Institute for Gene Therapy, Istituto Scientifico San Raffaele Hospital (HSR-TIGET), Milan, Italy.
- 12/98-05/00 Co-Director of Telethon Institute for Gene Therapy (HSR-TIGET), Scientific Institute H.S. Raffaele - Milan, Italy.
- 06/00-present Director of San Raffaele Telethon Institute for Gene Therapy (HSR-TIGET) – Milan (Italy)

### Martin McGlynn, President and Chief Executive Officer, StemCells, Inc

Martin McGlynn joined StemCells, Inc. in January of 2001 as President and Chief Executive Officer. He was elected to the Company's Board of Directors on February 6, 2001.

A seasoned leader, Mr. McGlynn joined the Biotech industry in 1994 having held several executive positions in the life sciences and healthcare sector. As the President and Chief Executive Officer of Pharmadigm, Inc., a privately held, venture capital backed start-up company in Salt Lake City, Mr. McGlynn led the company's efforts surrounding the research and development of a new class of anti-inflammatory agents. This successful endeavor utilized Mr. McGlynn's prior experience in FDA and USDA regulated industries in Europe, Canada and the United States.

In 1990, Mr. McGlynn joined the BOC Group as the President of Anaquest, Inc., a global leader in anesthesia and acute care pharmaceuticals headquartered in New Jersey. Prior to Anaquest, he held several positions with Abbott International Ltd. During his 13 year tenure with Abbott, he rose through the ranks and was appointed President and General Manager, Abbott Canada Ltd., in 1982 and a Vice President of Abbott International Ltd. in 1986. This succession built on Mr. McGlynn's early career experiences with Becton Dickinson, Ireland, Ltd. and The International Meat Company, a USDA-approved beef processing company, located in Dublin, Ireland.

Mr. McGlynn is a native of Dublin, Ireland and a graduate of University College, Dublin. He is a former member of the Board of Directors of the Confederation of Irish Industries (CII) and served two terms as a Board Member of the Pharmaceutical Manufacturers Association of Canada (PMAC). Mr. McGlynn has been an invited speaker on stem cell technology at numerous life science and healthcare conferences including in 2006, the 7th Harvard Business School Healthcare Conference, the Wharton Healthcare Business Conference, and the California Institute for Regenerative Medicine Conference on Industry and Stem Cells in California.

### Roland Foisner, Max F. Perutz Laboratories

Chair of Division of Molecular Cell Biology, Department of Medical Biochemistry, Medical University Vienna.
Coordinator of the International PhD programme at the Vienna Biocenter
Coordinator of the EU-FP6 project "Euro-Laminopathies"
Principal Investigator of numerous national and international research projects (majority of which is funded by the Austrian Science Research Fund, FWF)
The majority of studies deals with nuclear lamina proteins: their potential functions in chromatin organization, in nuclear architecture, and in the control of gene expression, cell proliferation and differentiation, their dynamic behaviour during mitosis and differentiation, and their involvement in human diseases, termed laminopathies.
These diseases are linked to mutations in genes encoding lamina proteins and include muscular dystrophy, cardiomyopathy, lipodystrophy, peripheral neuropathy, and premature ageing, but the molecular disease mechanisms are poorly understood. In a second research focus in the lab, we study molecular mechanisms involved in late stage tumor progression, tumor invasion and metastasis, with a particular emphasis on the role of cell adhesion proteins. As experimental systems we mainly use transgenic mouse models and primary and immortalized cell culture differentiation models.

### Serge Herson, La Pitié-Salpêtrière hospital, Paris

Serge Herson is Professor and Head of internal medicine at the La Pitié-Salpêtrière hospital (Paris).
He is a specialist in neuromuscular diseases, especially inflammatory myopathies. He is principal investigator of several cutting edge therapy clinical trials, including the first gene transfer trial for Duchenne muscular dystrophy carried out between 2001 and 2004.
His bibliography includes about 200 peer reviewed papers and books.

### Krzysztof Jagla / Christophe Marcelle, MYORES (France)

MYORES is the first European Network of Excellence dedicated to studying normal and aberrant muscle development function and repair. It brings together 37 European research groups belonging to 23 research organisations in seven countries. They are working on six different model systems, both vertebrate and invertebrate, the idea being to combine the strengths of these different model systems.

Krzysztof Jagla, research director in Inserm's U384 laboratory in Clermont-Ferrand, France, acts as MYORES coordinator, and Christophe Marcelle of the Université Aix-Marseille II as the network's sub-coordinator.

### Boris Šuštaršič

Boris Šuštaršič of the Slovenian Muscular Dystrophy Association has been president of the European Alliance of Muscular Dystrophy Associations (EAMDA) since the 2003 AGM in Malta. EAMDA's history dates back to 1970 when an exploratory conference was held in The Netherlands at which European muscular dystrophy associations met to discuss common ground and interests. 1971 saw the inaugural meeting of the European Alliance of Muscular Dystrophy Associations. The number of members has increased from 9 in 1971 to 23 in 2003. Milestones in EAMDA's history have included facilitating the emergence of organisations such as the EAMDA Youth Organisation (EYO) in 1989, the European Neuromuscular Centre (ENMC) also in 1989, the World Alliance of Muscular Dystrophy Associations (WAMDA) in 1990, and the European Consortium on Chronic Respiratory Insufficiency (ECCRI) in 1993. 1997 has seen the launching of the European Platform for Patients' Organisation. EAMDA is also a member of the European Disability Forum (EDF).

### Pat Furlong, Founding President & CEO Parent Project Muscular Dystrophy (PPMD)

Pat Furlong graduated from Mt. St. Joseph College in Cincinnati, Ohio with a BS in Nursing. She attended Graduate School at Ohio State University. While attending Ohio State, Pat spent most of her time in the Medical Intensive Care Unit. After marrying Dr. Tom Furlong, Pat ran the Renal Dialysis Unit and Patient Education Centre at Akron General Hospital.

Pat and Tom had 4 children, 2 girls -Jenny and Michelle and 2 boys - Christopher and Patrick. Pat continued her career on a part-time basis, teaching patient education classes at Middletown Regional Hospital.

In June 1984, Chris and Patrick were diagnosed with DMD. Pat immersed herself in DMD, working to understand the pathology of the disorder, research investment and mechanisms for optimal care.

Pat became frustrated because of the overall lack of significant investment in research and little understanding about issues related to Care.

In 1994, Pat Furlong and Donna Saccomanno initiated Parent Project Muscular Dystrophy specifically focused on Duchenne and Becker MD.

### Marianne de Visser, Professor of Neuromuscular Disease, University of Amsterdam

Marianne de Visser graduated in 1975 from the University of Amsterdam, Medical Facility. She carried out training in Neurology (Academic Hospital of the University of Amsterdam) from 1976-1980 and completed her PhD Thesis in 1981: Becker muscular dystrophy: a neurological, cardiological and computed tomographic study.

Since 1993 Marianne has held the position of Professor of Neuromuscular Diseases, Academic Medical Centre (AMC), University of Amsterdam, The Netherlands. Other appointments include; Chair of the board of the Educational Institute of Medicine of the Academic Medical Centre-University of Amsterdam 1997 – 2002; Vice-president of the Health Council of The Netherlands (since 1 September 2002); Chair of the 'Neurozintuigen' Division of the AMC (since 1 March 2004)

Marianne has over 140 publications in peer-reviewed international medical journals, mainly in the field of neuromuscular diseases, 15 publications in Dutch medical journals, and about 20 (contributions to) medical books. Her research focus on is on Motor neuron disease, Hereditary neuropathies and Inflammatory myopathies.

### Ségolène Aymé, Director of Research, Orphanet

Ségolène Aymé is a medical geneticist, director of research at the French Medical Research Council (INSERM). She is the executive manager of the Inserm department dedicated to information on rare diseases in Paris. This service established in 1997 the European database of rare diseases and orphan drugs named Orphanet (www.orpha.net), currently funded by the French ministry of health, the Inserm and the European commission (DG Public Health and DG Research). She was the founder and the first president (1997 to 1999) of the International Federation of Human Genetics Societies. She is the current chairperson of the Public and

Professional Policy Committee of the European Society of Human Genetics which is releasing recommendations and guidelines (http://www.eshg.org) and the leader of the Rare Diseases Task Force established by the European Commission, DG Public Health and Consumers' protection. She is also the chairperson of the French Society of Human Genetics and the editor-in-chief of the Orphanet Journal of Rare Diseases (www.ojrd.com).

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**11. Latest News / Research**

- **Industry pleased with orphan medical guidelines**

EU pharmaceutical companies have welcomed draft Commission guidelines that they say provide legal certainty on market exclusivity for orphan medicines that are developed for diseases or conditions that affect fewer than 5 in every 10,000 people. In the EU, 10-year market exclusivity is given as an incentive to encourage developers to invest in such diseases. The guidelines now enter a consultation period.

Press release:
http://www.orpha.net/actor/EuropaNews/2007/doc/ebe-europa.pdf
Draft guidelines:
http://ec.europa.eu/enterprise/pharmaceuticals/pharmacos/docs/doc2007/2007_03/draft_guideline_art8-2_200702.pdf

- **FP7 Electronic Submission System (EPSS)**

An Electronic Submission System (EPSS) is now available for some FP7 calls, including Health and European Research Council (ERC). The online system can be accessed from the relevant FP7 call page on Cordis at http://cordis.europa.eu/fp7/dc/index.cfm.

- **The Basel Biozentrum Taps Phenyx as Protein Identification Platform**

Biozentrum of the University of Basel will integrate GeneBio's Phenyx software platform for MS data analysis into its multi-tiered proteomics cluster.
http://www.genomicsproteomics.com

- **Santhera Announces Appointment of Cesare Mondadori as Head of Research**

LIESTAL, Switzerland, March 28, 2007--Santhera Pharmaceuticals (SWX:SANN), a Swiss specialty pharmaceutical company with a focus on neuromuscular diseases, announced today that it has appointed Dr Cesare Mondadori to the position of Senior Vice President Research. As Head of Research, he will oversee all research activities and have responsibility for the Company's preclinical programs reporting directly to Dr Thomas Meier, Chief Scientific Officer of Santhera.

"Cesare is a distinguished pharmaceutical research manager with a track record in basic research, drug discovery and preclinical drug development, particularly in the CNS area. He brings to Santhera almost 30 years of experience both in big pharmaceutical and biotech companies. His background, ranging from target discovery and assay development to in vivo pharmacology, preclinical development as well as preparation of clinical studies, will be of great value in further enhancing Santhera's preclinical organization," said Dr Thomas Meier, Chief Scientific Officer of Santhera. "It is with great pleasure that we welcome Cesare to Santhera and I am confident that he will make a significant contribution to advancing our preclinical programs in neuromuscular diseases."

Dr Mondadori joins Santhera from Neuro3d, Mulhouse, France, where he was Chief Scientific Officer with responsibility for all aspects of drug discovery and development of this CNS company. Prior to this position, Dr Mondadori was Head of CNS research at Aventis (1999 to 2000) and before at Hoechst Marion Russell and Marion Merrell Dow respectively (1996 to 1999) with focus on target discovery, assay development, target validation and screening in several CNS indications. In this role he was a member of the drug innovation and

approval teams as well as the scientific review committees. Dr Mondadori began his scientific career at Ciba-Geigy, Basel, Switzerland in 1978 as a research scientist in psychopharmacology. From 1981 to 1982 he was associate professor at the Department of Psychology of the University of Toronto, Canada. Dr Mondadori holds a PhD in neurosciences from the Swiss Federal Institute of Technology (ETH) Zurich, Switzerland.

http://www.medadnews.com/News/Index.cfm?articleid=427557

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**12. Progress update on active work packages**

Thank you to all the work package leaders who provided a progress update on their active work packages, the highlights of which were published in last week's newsletter (3). We feel that this was a valuable section and would like to continue to provide updates to partners regarding the progress of active work packages. We would therefore encourage you to e-mail stephen.lynn@newcastle.ac.uk with further updates as and when they occur.

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**13. Calls for Papers / Posters**

The SMA Foundation would like to remind you of the opportunity to submit abstracts to the

**18th International Symposium on ALS/MND**
December 1 – December 3, 2007
Toronto, Canada

**The organizers have informed us that there will be a session dedicated to SMA in the program. They will provide a grant to support up to 20 free registrations for SMA researchers with accepted abstracts.**

- **The deadline for abstract submission is May 11, 2007**
- Abstracts are processed online, please click here to submit
- Please see attachment for additional details
- For more information on the meeting please click here

We hope you will be able to take advantage of this unique opportunity to mingle and network with our ALS colleagues.

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**14. Articles**

**Inflammatory pathway leads to Duchenne muscular dystrophy**

Duchenne muscular dystrophy (DMD) is characterized by progressive muscle wasting and usually results in death by the age of 30. Although mutations in the dystrophin gene cause DMD, they trigger persistent inflammation, which is probably what intensifies disease progression.

In a study that appears online on March 22 in advance of publication in the April print issue of the Journal of Clinical Investigation, Denis Guttridge and colleagues from Ohio State University, Columbus, show that a signaling pathway associated with chronic inflammation (the IKK/NF-kappa-B pathway) is upregulated in both muscle cells and immune cells from individuals with DMD and from mice lacking expression of dystrophin. Impaired IKK/NF-kappa-B signaling in mice lacking expression of dystrophin (achieved by either knocking out one of the genes encoding the p65 NF-kappa-B subunit or treating the mice with an IKK inhibitor) improved their muscle function and muscle regeneration. The authors therefore suggest that the IKK/NF-kappa-B signaling pathway might provide a viable therapeutic target for the treatment of DMD.-Journal of Clinical Investigation

http://www.huliq.com/16168/inflammatory-pathway-leads-to-duchenne-muscular-dystrophy

**15. Upcoming Conferences**

The 5th EuroBioBank Annual Network Meeting will be held on 10-11 May 2007 in Paris and will have a focus on neuromuscular diseases.

Location: Hôpital Broussais, Plateforme Maladies Rares, 96 rue Didot, 75014 Paris, France.