

# TREAT-NMD

Serving the neuromuscular community



## INTRODUCTION

- 04 An introduction from the coordinators
- 05 Creating partnerships to build essential resources

## DEVELOPING RESEARCH RESOURCES

- 07 TREAT-NMD Advisory Committee for Therapeutics (TACT)
- 08 Biobanks for neuromuscular diseases
- 08 Preclinical research on animal models: creating consensus and developing SOPs
- 08 Facilitating research networking and collaboration

## SUPPORTING THERAPY DEVELOPMENT

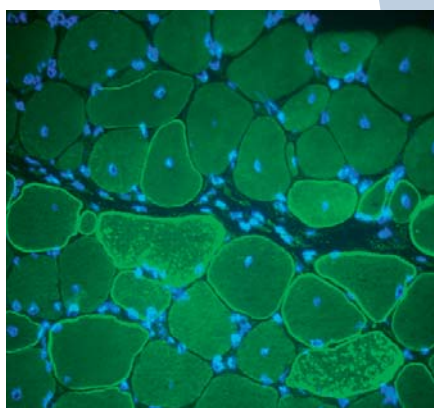
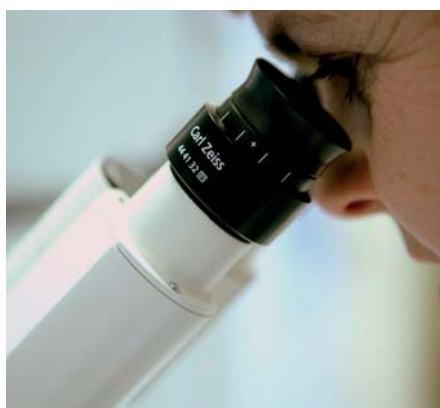
- 09 Patient registries
- 10 Outcome measures research and validation and the Registry of Outcome Measures (ROM)
- 11 Engaging with regulatory authorities
- 12 The Care and Trial Site Registry
- 13 Regulatory affairs database
- 13 Advisory support for industry

## IMPLEMENTING BEST PRACTICE

- 15 Standards of care guidelines
- 15 Training and education

## CATALYZING NEW DEVELOPMENTS

- 18 Catalyzing new developments
- 21 TREAT-NMD: the future
- 22 Acknowledgements
- 22 TREAT-NMD partners



# Introduction

## AN INTRODUCTION FROM THE COORDINATORS



“Recent years have seen rapid developments in the neuromuscular field and a corresponding surge in interest from the pharmaceutical industry. Promising preclinical results raise the potential for new therapies in the near future.

Yet translational research towards therapy development for neuromuscular diseases has faced a number of barriers.

For patients, promising research results have still not been translated into the treatments they hope for, while lack of standardized care guidelines prevents many from receiving optimal care.

For the biomedical industry, identifying the investigators and sites with the relevant expertise and accessing the appropriate patient cohorts for clinical trials has been a significant challenge.

For clinicians and researchers, lack of support tools such as validated clinical outcome measures or standard operating procedures for research protocols has held back therapeutic development.

TREAT-NMD addresses all these issues, uniting the stakeholders in the community and providing an infrastructure that is accelerating research and therapy development, increasing collaboration, improving patient care and helping to support ‘clinical trial readiness’ on an international scale.

TREAT-NMD was established as a EU-funded ‘network of excellence’ with the remit of ‘reshaping the research environment’ in the neuromuscular field. The network has developed from its European roots to become a global organization bringing together leading specialists, patient groups and industry representatives to ensure preparedness for the trials and therapies of the future while promoting best practice today.”

### Professors Kate Bushby and Volker Straub

TREAT-NMD coordinators  
Newcastle University, UK

“By providing carefully designed infrastructure to support all stages of therapy development, and promoting necessary collaborations among patients, advocacy groups, academic institutions, industry, and governmental agencies, internationally, TREAT-NMD is becoming *the* essential, go-to resource to advance novel treatments for devastating neuromuscular diseases.”

John D. Porter, Ph.D.  
Program Director,  
Neuromuscular Disease NINDS/NIH Office of Translational Research



## CREATING PARTNERSHIPS TO BUILD ESSENTIAL RESOURCES

At the heart of TREAT-NMD is a commitment to bringing real patient benefit through partnerships between clinical and scientific leaders in the field, advocacy groups and industry to develop the resources for therapy development and delivery.

### Partnering with patient advocacy groups

Close relationships with the major advocacy groups in the field have been key to the network's success. The initial European Union funding for the network was advocated for by patient groups, in particular the Association Française contre les Myopathies (AFM, the French Muscular Dystrophy Association), with the support of the European Organisation for Rare Diseases (EURORDIS). The network's original supporters have been joined by the major advocacy organizations across the globe, both disease-specific groups and those with a broader remit. To each of them TREAT-NMD can offer advice, expertise and key resources. In return, researchers and clinicians within TREAT-NMD can benefit from the knowledge and experience patient groups can provide.

“As a parent organization dedicated to the battle against neuromuscular diseases, AFM has been developing over the years an ambitious program aimed at building knowledge and setting up new treatments.

Overcoming bottlenecks has become a priority as therapeutic development progresses. In this view, TREAT-NMD is a key network that successfully addresses major issues. Important tools and strong expertise are being provided to the scientific and medical community, which will undoubtedly help accelerate cutting-edge therapies for otherwise unmet medical needs.”

Serge Braun, PharmD, PhD  
Scientific Director, AFM



# Introduction

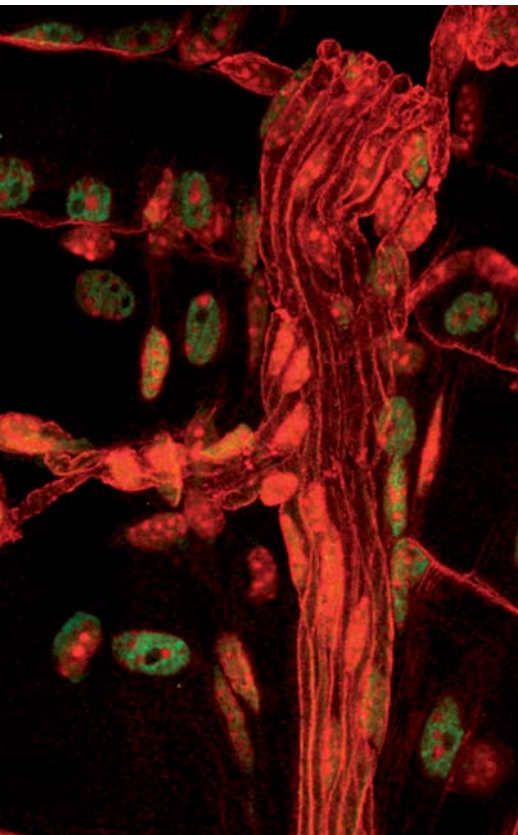
## Addressing all inherited neuromuscular diseases

The platform developed by TREAT-NMD, which initially focused on Duchenne muscular dystrophy (DMD) and spinal muscular atrophy (SMA), is now recognized to be of benefit to all inherited neuromuscular conditions.

Strong collaborations have been built across the neuromuscular community by engaging numerous international researchers and organizations worldwide, who are able to tap into the resources developed by TREAT-NMD. These resources span the research and clinical arenas and form a suite of tools that are essential for rapid progress towards new therapies.

Tools include:

- Advisory committee for therapeutics
- DNA, cell and tissue biobanks
- Consensus and SOP generation for animal models
- Patient registries
- Network of care and trial sites
- Outcome measures for NMD
- Regulatory links
- Support for further grants
- International standards for diagnosis and care
- Focused training and education



## Case study:

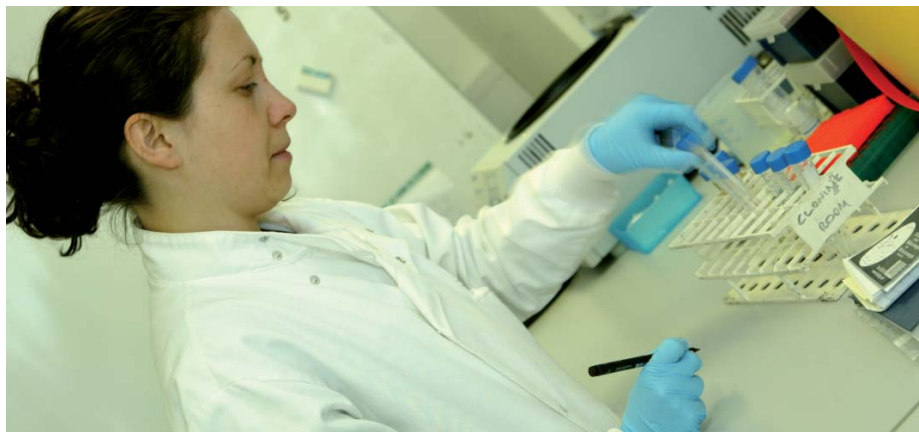
### Congenital muscular dystrophies

Partnership with Cure CMD, a US-based patient advocacy group for congenital muscular dystrophies

- Cure CMD registry for all CMD subtypes (CMDIR) established, feeding into TREAT-NMD gene-specific registries
- International conference addressed issues of trial-readiness in CMD, including outcome measures, animal models, and therapy prioritization.
- Consensus process led by Professor Thomas Sejersen (Stockholm) and Dr Ching Wang (Stanford University) involving workshops with leading CMD experts led to the development of care standards for CMDs
- Researchers with promising preclinical results on CMD targets are encouraged to submit applications to TACT for review

# Developing Research Resources

## TREAT-NMD ADVISORY COMMITTEE FOR THERAPEUTICS (TACT)



Of the many promising research results presented at conferences, published in journals and hailed as the basis for possible future treatments and cures, few progress into clinical trial. Evaluating the therapeutic potential of drugs seemingly ready for this step is a challenge not only for the patients who build hope on preclinical results and for the potential funders and industry sponsors of the research, but also for the researchers themselves.

TACT, the TREAT-NMD Advisory Committee for Therapeutics, is an expert multidisciplinary body that provides the neuromuscular community (clinicians, researchers, patient advocacy groups and industry) with independent and objective guidance on advancing new therapies (whether novel or repurposed) for neuromuscular diseases.

Its goal is to position the potential therapy along a realistic pathway to eventual clinical trial and registration by evaluating preclinical data as well as drug development considerations that are crucial for the conduct of studies that generate meaningful data. In close collaboration with the TREAT-NMD clinical trials coordination centre (Freiburg, Germany), TACT is also dedicated to providing information on optimal trial design and the resources available to investigators and industry for clinical trial planning and conduct.

“The strength of TACT is its broad scientific and development expertise coupled with the rigour and independence of its review process, performed in a truly global context. This committee can provide objective and constructive guidance from world experts in the field that will both help researchers focus on areas for development and ensure the wider community is better informed about the readiness of new therapies for the next step.”

Cristina Csimma, TACT chair

## Core Committee Members

- **Cristina Csimma PharmD MHP**  
Vice President, Drug Development, Virdante Pharmaceuticals Inc, Cambridge, MA, USA.
- **Didier Caizergues PharmD DESS**  
Head of the Regulatory Affairs Department at Généthon, France.
- **Petra Kaufmann MD MSc**  
Director of the Office of Clinical Research, NINDS, USA.
- **Rudolf Korinthenberg MD**  
Head of the Department of Neuropediatrics & Muscular Disorders, Children's Hospital, University Hospital Freiburg, Germany.
- **John McCall PhD**  
Senior Consultant, PharmaMacLLC, USA.
- **D. Elizabeth McNeil MD**  
Medical Officer, Food and Drug Administration, Rockville Maryland, USA.
- **Jerry Mendell MD**  
Director, Center for Gene Therapy, Columbus Children's Research Institute, Professor of Pediatrics and Neurology, Ohio State, University, USA.
- **Kanneboyina Nagaraju PhD MVSc**  
Associate Professor of Pediatrics, Children's National Medical Center, Washington DC, USA.
- **Dominic Wells PhD MA VetMB**  
Professor in Translational Medicine, Department of Veterinary Basic Sciences, Royal Veterinary College, University of London, UK.

Further details, together with the complete list of over 40 experts making up the full committee, are available online at [www.treat-nmd.eu/TACT](http://www.treat-nmd.eu/TACT)



# Developing Research Resources

## BIOBANKS FOR NEUROMUSCULAR DISEASES

The TREAT-NMD-integrated resource EuroBioBank is the only biobank network dedicated to rare disease research in Europe, and its samples are distributed to researchers across the globe. Led by the patient network EURORDIS (Rare Diseases Europe) with the guidance of EuroBioBank scientific director Professor Hanns Lochmüller (Newcastle), EuroBioBank provides human DNA, cell and tissue samples as a service to the global scientific community conducting research

on rare diseases. Since its inception as a separate EU-funded project under the Fifth Framework Programme (contract no. QLRI-CT-2002-02769), EuroBioBank has been referenced in over 100 research papers, and a total of approximately 400,000 samples are available to researchers worldwide via the online catalogue.

[www.eurobiobank.org](http://www.eurobiobank.org)

## PRECLINICAL RESEARCH ON ANIMAL MODELS: CREATING CONSENSUS AND DEVELOPING SOPS

Comparability of results between different research groups is a major issue in the preclinical field. As the result of international collaboration between animal model specialists worldwide led by TREAT-NMD (Professor Markus Rüegg, Basel and Santhera Pharmaceuticals, Liestal) and the Washington Wellstone Center (Professor Eric Hoffman and Dr Kanneboyina Nagaraju), a review of animal models for DMD has been published, and a comprehensive set of standard operating procedures or SOPs for various experimental protocols on DMD models has been drafted and made available online for the use of researchers across the world. The SOPs are attracting

numerous downloads, and this approach to harmonization is now also being applied to models of other diseases.

[www.treat-nmd.eu/animalmodels](http://www.treat-nmd.eu/animalmodels)



## FACILITATING RESEARCH NETWORKING AND COLLABORATION

TREAT-NMD is bringing scientists together in many different areas – from facilitating exchange on high throughput screening methodologies to optimizing production and safety of therapeutics. In one example, researchers working on different antisense oligonucleotide chemistries in different labs (the MDEX consortium led by Professor Francesco Muntoni in London and the Leiden lab led by Dr Annemieke Aartsma-Rus) have been able to set up a regular dialogue to discuss progress and results in a constructive and non-competitive way. This model for collaboration helps groups learn from one another and progress more quickly, without damaging their competitive advantage.

A major milestone for TREAT-NMD was the network's international conference with NIH in November 2009. With over 350 participants including researchers, clinicians, patient groups, industry and regulatory representatives and a focus on facilitating cross-talk to address the barriers that are still hindering neuromuscular translational research, it provided a unique opportunity for taking stock and setting future priorities.

[www.treat-nmd.eu/2009conference](http://www.treat-nmd.eu/2009conference)



# Supporting Therapy Development

## PATIENT REGISTRIES

In collaboration with clinicians and patient advocacy groups across the world, TREAT-NMD has created patient registries with future trials and therapies in mind. Led by Dr Christophe Bérout (INSERM, Montpellier), the global registries for DMD and SMA are recognized as a leading resource for trial planning and recruitment in these diseases at an international level and are being used by industry for this purpose. They are also open to enquiries from academic researchers. Registries for a range of other conditions are also underway or in preparation.

For patients, the registries are a valuable way of receiving information and feedback related to their condition and provide an important connection to the research community. The registries are governed by a charter and by an oversight committee including patient representatives.

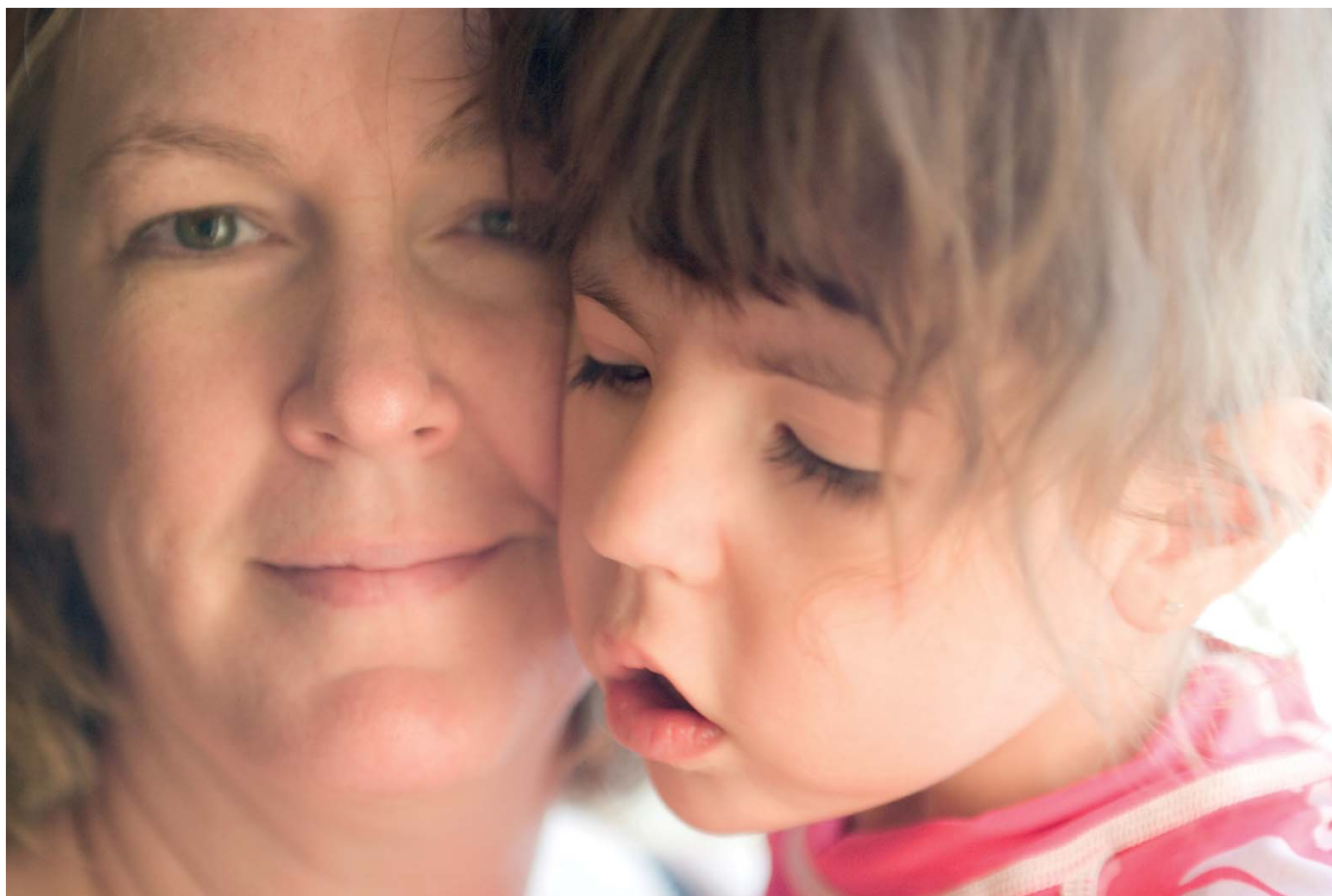
Key benefits of the TREAT-NMD registries:

- Single entry point for access to patient data worldwide
- Registries contain a core set of information including accurate, verified genetic diagnosis together with key clinical data items, all updated at least annually
- Powerful clinical trial feasibility tool: can filter patients by precise mutation, age, ambulation status, medication type and location
- Powerful recruitment tool: patients have consented to being contacted about trials for which they may be eligible
- Inbuilt patient data protection: patients are only contacted through their national registry; their data is kept confidential

[www.treat-nmd.eu/patientregistries](http://www.treat-nmd.eu/patientregistries)



# Supporting Therapy Development



## OUTCOME MEASURES RESEARCH AND VALIDATION AND THE REGISTRY OF OUTCOME MEASURES (ROM)

The correct choice of outcome measures for a clinical trial is critical to its ability to generate meaningful data that enables the therapy being trialled to move towards regulatory approval.

In an activity led by Professor Eugenio Mercuri (Rome), TREAT-NMD is involved in research to define and validate appropriate outcome measures (OMs) for different conditions and is engaging in open dialogue with the regulatory authorities over measures for use in pivotal trials.

In related initiatives, promising techniques requiring further development and standardization such as quantitative MRI are being moved forward and standard operating procedures (SOPs) generated.

The online TREAT-NMD Registry of Outcome Measures (ROM) and its associated tools are coordinated by Dr Michael Rose (London). These

resources are designed to give guidance, information and assistance to all professionals seeking details of outcome measures, including international collaborative teams of reviewers undertaking the crucial task of choosing the right outcome measures for neuromuscular disease trials, and to avoid duplication of this effort. The searchable registry contains information about outcome measures, including a description, details of validation, availability, contact details for providers, and references to related documents including manuals and training videos. Review teams can record OMs by category as being considered for a specific study or trial and benefit from seeing the OM choices being considered by other review teams. A manual gives advice on how to assess and select OMs.

[www.treat-nmd.eu/outcomemeasures](http://www.treat-nmd.eu/outcomemeasures)

## ENGAGING WITH REGULATORY AUTHORITIES

TREAT-NMD has proactively engaged with the European and US regulatory agencies, the EMA (formerly EMEA) and FDA, over key issues surrounding trials in neuromuscular disorders. The lack of previous trials in these conditions means that basic groundwork relating to drug registration trials still needs to be worked out with the regulators, with issues ranging from which outcome measures might be considered appropriate for granting of marketing approval in particular conditions to the

challenges of personalized medicine for highly mutation-specific therapies such as exon skipping.

By convening broad, strategic meetings with the regulatory authorities over these issues TREAT-NMD has been able to develop close links with EMA that will guide future development for new therapies. The approach has been welcomed as providing a unique forum for discussion that benefits both sides.

### Case study: EMA workshop on DMD

In September 2009 a broad group of 98 DMD experts assembled by TREAT-NMD – including clinicians involved in current clinical trials, patient/parent groups and pharmaceutical companies working on Duchenne therapies – met at the London offices of EMA, the European Medicines Agency, to discuss the regulatory issues surrounding the unprecedented level of personalization inherent in antisense oligonucleotide (AO) therapies for genetic conditions like DMD. Input from Duchenne patient groups and experts worldwide ensured that there was global representation at the meeting, which also included a representative of the US Food and Drug Administration (FDA).

Although more than 80% of Duchenne boys could potentially benefit from the “exon skipping” approach, each exon targeted requires a unique AO, and thus treating all the different mutations known to cause DMD requires the development of large numbers of AOs, each treating only a small subset of the patient population. This represents a personalized approach to therapy that is currently without precedent for a genetic disease, and concerns were raised from many different sectors that the regulatory hurdles might hinder the approval of drugs to treat the rarer mutations. The meeting with EMA therefore sought to take steps to identify a pathway that will allow the safe and efficient progress of these drugs through the approval process. The groups present were praised for bringing the field together in a constructive way.

More information:  
[www.treat-nmd.eu/EMEA-antisense](http://www.treat-nmd.eu/EMEA-antisense)

# Supporting Therapy Development

## THE CARE AND TRIAL SITE REGISTRY

Defined patient cohorts and experienced trial sites and investigators are essential requirements for any trial. The TREAT-NMD Care and Trial Site Registry (CTSR) is a database of information on clinical sites established by the Clinical Trials Coordination Centre under the leadership of the German MD-NET and Professor Rudolf Korinthenberg (Freiburg) to facilitate the selection of centres with the expertise to take part in clinical trials. Registered sites have completed detailed feasibility information regarding

their experience, facilities, equipment and personnel, equivalent to a company or CRO's early feasibility enquiry. Companies have already made use of the CTSR to assist in their site selection for upcoming trials.

The CTSR contains data on over 200 registered sites in over 40 countries worldwide.

[www.treat-nmd.eu/CTSR](http://www.treat-nmd.eu/CTSR)



## Case study: EMA workshop on SMA

In 2008 a workshop set up in collaboration with the International Coordinating Committee (ICC) for SMA helped set the collaborative agenda for future trials in SMA. In order for trials to move through the approval process without delays, consensus between trial planners and regulators on endpoints and novel methodologies is essential. Topics addressed at the meeting included outcome measures and trial design to best assess efficacy in phase I/II studies in SMA, ensuring the relevancy of efficacy outcome measures used in pilot studies to later studies, and facilitating progression from small phase I/II studies to larger studies in a way which would satisfy regulatory authorities.

The meeting established broader common ground between the regulatory authorities and those interested in running clinical trials in SMA, and outcomes included consensus on certain types of outcome measure and a definition of areas where further development is required. It was acknowledged that it is important to educate the regulators about disease mechanism and disease phenotypes, and meetings such as this one were therefore recognised as highly valuable to both sides.

More information:  
[www.treat-nmd.eu/EMEA\\_SMA](http://www.treat-nmd.eu/EMEA_SMA)



## REGULATORY AFFAIRS DATABASE

The regulatory affairs database is a source of specific regulatory information for investigators involved in clinical trial planning. It contains details of national legislation from Europe and the US.

European regulations and guidelines (e.g. from ICH and EMA) are also available.

[www.treat-nmd.eu/regulatoryaffairs](http://www.treat-nmd.eu/regulatoryaffairs)

## ADVISORY SUPPORT FOR INDUSTRY

TREAT-NMD brings together leading specialists in the neuromuscular field and is a natural partner for biotech and pharmaceutical companies developing new therapeutics. The unique tools, services and expertise available within the network can support, simplify and accelerate clinical studies.

the expertise to conduct specific trials and support with patient recruitment.

The network offers consultancy in all aspects of neuromuscular trial planning, from complete set-up of a full scientific advisory board for protocol development to advice on the selection of individual neuromuscular experts with preclinical, clinical, clinical evaluator, biostatistical or regulatory expertise.

Through TREAT-NMD companies can not only receive advisory support from leading neuromuscular experts, but also assistance in locating the centres with

[www.treat-nmd.eu/industry](http://www.treat-nmd.eu/industry)

## Case study: Industry enquiries

A number of companies have requested feasibility data from the TREAT-NMD patient registries and care and trial sites registry. Examples of requests include:

- numbers of patients with specific mutations (amenable to particular therapies moving into trial)
- patients stratified by age range and ambulation status
- details of steroid use and cardiac involvement (DMD)
- total numbers of patients per country meeting specific inclusion criteria (up to 30 countries worldwide)
- frequency of particular mutations in DMD
- feasibility data on trial sites including equipment and staffing, muscle biopsy experience, trial experience
- details of site diagnostic capabilities – availability of MLPA analysis, point mutation detection, deletion/duplication analysis

This information and more is readily available through the TREAT-NMD resources and this can dramatically speed up trial planning and thus lower the barriers to getting a trial established.

At the recruitment stage, eligible patients can subsequently be contacted through their national registries in parallel with site-based recruitment efforts.





# Implementing Best Practice

## STANDARDS OF CARE GUIDELINES

Variations in care standards between and even within countries not only impact on quality of life but also make comparison of trial results from different centres a challenge.

TREAT-NMD has worked with international specialist groups and patient advocacy organizations on international consensus documents setting out best practice in diagnosis and patient care and under the leadership of Professor Thomas Sejersen (Stockholm) has made these available in multiple languages in family-friendly form to patients worldwide. Standards of care for

spinal muscular atrophy and Duchenne muscular dystrophy are available through the TREAT-NMD website and as handy printed brochures. The network is expanding its collaborations on standards of care to encompass additional conditions and is committed to supporting their development and dissemination across the field.

SMA care standards:

[www.treat-nmd.eu/sma-care](http://www.treat-nmd.eu/sma-care)

DMD care standards:

[www.treat-nmd.eu/dmd-care](http://www.treat-nmd.eu/dmd-care)

## TRAINING AND EDUCATION

Under the leadership of the European Neuromuscular Centre (ENMC), TREAT-NMD has capitalized on the expertise available within the network and the availability of best-practice standards to develop training and education initiatives with the goals of raising care standards worldwide, training the next

generation of neuromuscular specialists, providing trial-focused training and disseminating expertise. Activities include focused workshops on care standards in neuromuscular disorders, patient registry curator training and support for major conferences worldwide.



# Implementing Best Practice



## Trial design training

Owing to the relative lack of neuromuscular trials until now, investigator-led trials have been a challenge because many potential investigators do not have sufficient experience in trial design. The TREAT-NMD Clinical Trials Coordination Centre in Freiburg runs workshops introducing future investigators to the fundamentals of effective clinical trial design and training them in developing study protocol drafts. These training workshops focus on the specific requirements of trials in neuromuscular diseases and explain the complexities of trial design in rare disorders and powering trials for small patient cohorts.

## Clinical evaluator / trial physiotherapist training

For a multicentre study, it is essential that procedures and evaluations are performed consistently. TREAT-NMD arranges this type of training for the pharmaceutical companies active in the field, making use of the expert staff available through the network, including leading trial physiotherapists Dr Michelle Eagle (Newcastle), Dr Julaine Florence (Washington) and Dr Elena Mazzone (Rome).

## Case study:

### DMD care standards generation

In January 2010 a major international consensus document setting out best practice in care for Duchenne muscular dystrophy (DMD) was published in the *Lancet Neurology* journal. The drafting of these guidelines was the result of a three-year-long project guided by the US Centers for Disease Control and Prevention (CDC) using a rigorous review process that required 84 international experts to consider more than 70 thousand different care scenarios. TREAT-NMD was integrally involved in the creation of this document through the managing editorship of network coordinator Kate Bushby. To ensure patients had access to information as early as possible, TREAT-NMD produced a set of "interim recommendations" over a year before the publication of the full document, and once the full published article became available a close collaboration between TREAT-NMD, advocacy groups and healthcare professionals resulted in the publication of a comprehensive "family guide" that summarizes the main recommendations in patient-friendly form. The full published *Lancet* document is now being used in countries across Europe by TREAT-NMD and the CARE-NMD project as the basis for lobbying and discussions with health authorities to work towards the implementation of best-practice care for all patients, while the family guide can be used by patients and families to assist them in discussions of their care with their doctors. The recommendations are also being supported by pharmaceutical companies active in the Duchenne field, who recognise not only that a standardized baseline level of care is of value in multicentre clinical trials but also that the recommendation of full genetic diagnosis for all patients is of great importance.

More information:

[www.treat-nmd.eu/dmd-care](http://www.treat-nmd.eu/dmd-care)



## Case study:

### SMA care standards generation

A recognition that recent progress in the understanding of the molecular pathogenesis of SMA and improvements in medical technology have not been matched by similar developments in the care of SMA patients led to a consensus publication in the *Journal of Child Neurology* in 2007. 12 core members of the International Standard of Care Committee (SCC) for SMA worked with over 60 SMA experts using a Delphi process and virtual and in-person meetings to achieve consensus on 5 care areas: diagnostic/new interventions, pulmonary, gastrointestinal/nutrition, orthopaedics/ rehabilitation, and palliative care. Consensus was achieved in several areas relating to common medical problems in SMA, the diagnostic strategies, recommendations for assessment and monitoring, and for therapeutic interventions in each care area. The published consensus statement addressed the 5 care areas according to the patient's functional level.

TREAT-NMD experts were not only involved in the drafting of the academic article but also worked to produce a comprehensive précis aimed at families and general practitioners. This has now been made available in multiple languages in booklet and online form, and feedback has indicated that it is seen as a tremendously valuable resource that allows patients and doctors to work together to achieve optimal care.

More information:

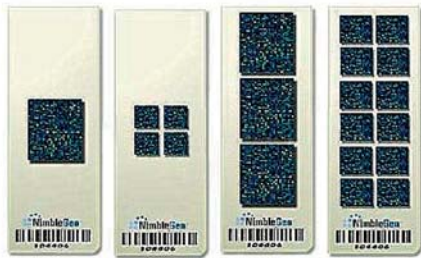
[www.treat-nmd.eu/sma-care](http://www.treat-nmd.eu/sma-care)



# Catalyzing New Developments

## CATALYZING NEW DEVELOPMENTS

In uniting such a wide range of neuromuscular specialists and other key stakeholders, TREAT-NMD has become a significant catalyst for new research proposals and spin-off projects, all of which can take advantage of the translational research platform established by the network. Numerous funding applications have already been successful on national, European and international levels and others are constantly in the pipeline. The following three examples of EU funded projects utilizing the TREAT-NMD infrastructure address advanced diagnostics, biomarker development and advancing care in NMD.



### NMD-chip

The NMD-chip project (contract no. 223026), led by Professor Nicolas Lévy (Marseille), was funded by the European Union's Seventh Framework Programme under a call for "high throughput molecular diagnostics.

The aim of NMD-chip is to design, develop and validate new sensitive high-throughput DNA arrays to efficiently diagnose patients affected by NMDs, specifically Duchenne / Becker muscular dystrophies (DMD/BMD), limb girdle muscular dystrophies (LGMD), congenital muscular dystrophies (CMD), and hereditary motor-sensory neuropathies or Charcot-Marie-Tooth neuropathies (CMT).

The new sensitive and reliable tools originating from this project will allow the assessment of all known genes implicated in a group of diseases at one time, and the efficient analysis of chip data through optimized read-out bioinformatic tools will provide results within 72 hours to one week and thus be cheaper than any "gene by gene" approach. It is anticipated that developing these NMD-chips could allow the cost of molecular diagnostics to be decreased by a factor of 10.

[www.nmd-chip.eu](http://www.nmd-chip.eu)

### BIO-NMD

BIO-NMD (contract no. 241665), led by Professor Alessandra Ferlini (Ferrara), is a research project on biomarker research and discovery that was funded by the European Union's Seventh Framework Programme in January 2010. Biomarkers are measurable bio-parameters that can be used to monitor disease progression, prognosis and drug response, thus optimizing the choice of appropriate and often personalized therapies. Early-stage clinical trials are typically so short that they provide limited opportunity to evaluate whether there is a meaningful clinical improvement. The identification of surrogate endpoints to help detect the activity of a new therapy in the early stages of development is essential to rapid progress on the development path. BIO-NMD aims to discover and validate new genomic and proteomic biomarkers, which will be validated both in animal models and in human samples before entering into a qualification process at the EMA.

[www.bio-nmd.eu](http://www.bio-nmd.eu)

## CARE-NMD

CARE-NMD, led by Dr Janbernd Kirschner (Freiburg), is a project funded by the EU's Executive Agency for Health and Consumers to implement care standards for DMD in 19 European countries.

Although expert centres for the care of patients with neuromuscular diseases do exist in most European countries, many patients still do not receive treatment in line with current guidelines. The CARE-NMD project is establishing an inclusive reference network of care centres specializing in the treatment of DMD to analyze current treatment practices for patients with DMD, identify inequalities between and within participating European countries, and host information and training workshops to ensure dissemination and implementation of treatment recommendations at national and European level. Barriers to implementation will be identified and collaborative solutions sought in order to help stakeholders and decision-makers in the area of healthcare to take further action to improve the care of patients with DMD and other neuromuscular disorders.

[www.care-nmd.eu](http://www.care-nmd.eu)









# Catalyzing New Developments

## TREAT-NMD: THE FUTURE

The platform TREAT-NMD has established for accelerating translational research and catalyzing new developments across the neuromuscular field is one that has already proved its value and that its coordinators are committed to sustaining in the long term. The future will see the network further develop this essential infrastructure, playing an increasing role in areas such as long-term surveillance of therapies coming into the clinic, continued regulatory and lobbying activities to support availability of drugs with proven efficacy, updating and implementation of care standards across the neuromuscular spectrum,

benchmarking of service delivery against care standards, supporting idea generation from the leaders in the field, and providing training and education to support the development of new care and trial sites with the expertise to treat neuromuscular patients.

All of this will ensure that the neuromuscular community can move forward together to develop the treatments that its patients and its practitioners have been awaiting for so long.

“Solutions for patients affected by severe and rare diseases can only emerge if all the actors work together. It is not an option but a prerequisite. This is exactly what TREAT-NMD does: it creates the opportunities to make everyone involved move forward together.”

Dr Ségolène Aymé  
Director of Orphanet



## ACKNOWLEDGEMENTS

From 2007 to 2011 inclusive, TREAT-NMD is a “network of excellence” supported through Priority 1 (Life Sciences, Genomics and Biotechnology for Health) of the European Union’s Sixth Framework Programme under contract number LSHM-CT-2006-036825.



### Image credits

We'd like to thank everyone who allowed us to use their photos in this brochure, in particular the following individuals:

**Front cover and p21:** Vitaliy and Yuliya Matyushenko ([www.csma.org.ua](http://www.csma.org.ua))

**p5:** Peter Streng ([www.enmc.org](http://www.enmc.org)) and Boris Šuštaršič ([www.eamda.org](http://www.eamda.org), [www.drustvo-distrofikov.si](http://www.drustvo-distrofikov.si))

**p10:** Lisa and Kayla Vittek ([www.myotonic.org](http://www.myotonic.org); [www.cureforkayla.com](http://www.cureforkayla.com)) and the staff and patients of the Newcastle Muscle Centre.

## TREAT-NMD PARTNERS

The following partners receive funding from the European Union through the TREAT-NMD network. Close external partnerships have also been developed with numerous other individuals and organizations across the world, and the network now has members in over 40 countries.



**TREAT-NMD Coordination Office**

Institute of Human Genetics  
Newcastle University  
International Centre for Life  
Newcastle upon Tyne  
NE1 3BZ  
United Kingdom

T: +44 (0)191 241 8605

F: +44 (0)191 241 8770

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

[www.treat-nmd.eu](http://www.treat-nmd.eu)