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TREAT-NMD Alliance Brochure

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**Keyword List:**

**Abstract:**  
The TREAT-NMD brochure is one of our key dissemination tools. The brochure was first produced in 2010 and has been updated in 2013 to reflect the development of the TREAT-NMD Alliance.

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1. **Introduction**

The TREAT-NMD brochure is one of our key dissemination tools. The brochure was first produced in 2010 and has been updated in 2013 to reflect the development of the TREAT-NMD Alliance.

The contents of the brochure cover the following TREAT-NMD activities, tools and resources:

**ABOUT THE TREAT-NMD ALLIANCE**
- An introduction
- Governance
- Membership
- Communication platform

**RESEARCH RESOURCES**
- TREAT-NMD Advisory Committee for Therapeutics (TACT)
- Biobanks for neuromuscular diseases
- Preclinical research on animal models and Standard Operating Procedures
- Facilitating research networking and collaboration

**SUPPORTING THERAPY DEVELOPMENT**
- Patient registries
- The Care and Trial Site Registry
- Outcome measures research and validation and the Registry of Outcome Measures (ROM)
- Regulatory interactions
- Regulatory affairs database
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**IMPLEMENTING BEST PRACTICE**
- Standards of care guidelines
- Clinical evaluator/trial physiotherapist training
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- Accelerating translational research and catalyzing new developments
- 3GB Test
- BIOIMAGE-NMD
- COST
- EUCERD Joint Action
Neuromics
Next generation sequencing
OPTIMISTIC
RARE-Bestpractices
RD-Connect
SCOPE-NMD
SKIP-NMD

2. **Presentation of results**

The current draft version of the updated brochure is presented below.

**About the TREAT-NMD Alliance**

**Introduction**

The TREAT-NMD Alliance is a global research network for the neuromuscular field. It provides an infrastructure to support and foster collaboration between clinicians, scientists, and patients, accelerate therapy development for rare inherited neuromuscular disorders, improve patient care through publication and dissemination of best-practice consensus guidelines, and provide reliable information for patients and professionals.

TREAT-NMD was established in 2007 as an 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 clinical trials and therapies of the future while promoting best practice today. The activities of the TREAT-NMD Alliance address the bottlenecks delaying the development of treatments, from animal model assessment, patient registries and outcome measures for clinical trials to standards of patient care.

**Addressing all inherited neuromuscular diseases**

The platform developed by TREAT-NMD, which initially focused on Duchenne Muscular Dystrophy (DMD) and Spinal Muscular Atrophy (SMA), has now been expanded to include many other rare inherited neuromuscular conditions.

Strong collaborations have been built across the entire neuromuscular community by engaging numerous international researchers and organizations worldwide, who are able to benefit from 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 among others:

- Advisory committee for therapeutics
- DNA, cell and tissue biobanks
- Standard Operating Procedures for animal models
- Patient registries
- Network of care and trial sites
- Outcome measures for NMD
TREAT-NMD was initially an EU ‘Network of Excellence’ funded under the 6th Framework Programme. Following the end of this funding TREAT-NMD developed a new charter and governance structure, enabling the network to continue to grow and develop scientifically and geographically. In 2012, the TREAT-NMD Executive Committee, made up of both academic and patient representatives took over governance of the network. The members of the Executive Committee were elected from the TREAT-NMD Task Force and from member patient organisations.

The Executive Committee is responsible for providing overall policy and strategic direction to the TREAT-NMD Alliance, oversees activities and progress, and delegates responsibility for day-to-day operations to the representatives of the various activity groups.

The TREAT-NMD Task Force was set up to oversee the transition from the network's EU funding status to the Alliance. The Task Force, consisting of key clinical and scientific experts from around the world, plays an important role in current and future activities of the TREAT-NMD Alliance whilst also guiding the development of the Alliance’s future strategy.

Membership

TREAT-NMD recognises the valuable work in the neuromuscular field that many organizations, institutions, companies and individuals are undertaking with an invitation to become a member of the TREAT-NMD Alliance. Members benefit from closer ties with the network and with the TREAT-NMD Alliance work together with them to help implement the goals, from improved patient care to specialist scientific training, worldwide.

All TREAT-NMD Alliance members agree to adhere to the charter as part of a requirement of membership. If you or an organization wishes to be considered for membership of the TREAT-NMD Alliance, please visit the TREAT-NMD website.

Communication platform

The network benefits from an established communication platform that continues to develop and adapt to satisfy the growing needs of the Alliance. At the heart of the platform is the main TREAT-NMD website which annually attracts in excess of 275,000 page hits from over 50,000 people around the world whilst our newsletter keeps 3,500 stakeholders up to date with the issues relating to the neuromuscular field on a monthly basis.

In response to the growth in popularity of social media, the network now regularly keeps its increasing number of followers ‘instantly’ informed via its Twitter account @TREAT_NMD.

A number of related projects have since benefited from our experience and have integrated key aspects of the network’s communication platform into their infrastructures.

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 actually progress into clinical trials. 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 rare neuromuscular diseases.

The goal of each review is to position the potential therapy along a realistic and well informed pathway to clinical trials, and eventual registration, by evaluating the supporting preclinical data and all other critical drug development considerations that are necessary for objective decision-making and for the design and conduct of studies that generate meaningful data and have the potential to be funded longer term.

Further details about TACT, together with the complete list of over 50 experts making up the full committee, are available online at www.treat-nmd.eu/TACT

Biobanks for neuromuscular diseases

The TREAT-NMD-integrated resource, EuroBioBank, is a unique network of biobanks that stores and distributes quality DNA, cell and tissue samples for scientists conducting research on rare diseases, including neuromuscular disorders. Biobanks and biomaterial collections across the world may join the EuroBioBank. All samples remain in the possession of the member biobank, with EuroBioBank acting as a clearing house or "virtual" bank with an online catalogue and search engine for locating samples. Researchers from anywhere in the world who locate a sample of interest through the catalogue then liaise directly with the bank holding the sample, with sample exchange being facilitated by conditions set out in the EuroBioBank charter and standardised material transfer agreements. EuroBioBank has been referenced in over 100 research papers, and over 400,000 samples are available to researchers worldwide via the online catalogue. As of January 2012, the Fondazione Telethon provides the administrative support for coordinating the EuroBioBank network and hosting the website.

Preclinical research on animal models and Standard Operating Procedures (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, a review of animal models for DMD, SMA and CMD has been published, and a comprehensive set of standard operating procedures (SOPs) for various experimental protocols on DMD and SMA 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. Standard protocols are followed up and regularly updated about every two years. This task is currently funded by the Swiss Foundation for Research on Muscle Disease (FSRMM), as a member of the TREAT-NMD Alliance.

Facilitating research networking and collaboration

TREAT-NMD is bringing together scientists in many different areas which have resulted in spin off EU funded research networks such as BIO-NMD (focusing on biomarker research) and NEUROMICS. Furthermore it fosters networking activities such as the one initiated on exon skipping by Dr Annemieke Aartsma-Rus and Prof Francesco Muntoni, involving 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 and currently receives funding from the cooperation of science and technology (COST).

Since 2009, TREAT-NMD has been organising regular conferences which bring together patients, academics, clinicians, patient registry curators and industry representatives. These events are an excellent opportunity for networking, looking at the new areas of research and encouraging collaboration with the aim of addressing barriers that are still hindering neuromuscular translational research.
Supporting Therapy Development

Patient registries

In collaboration with clinicians and patient advocacy groups across the world, TREAT-NMD has created patient registries with future clinical trials and therapies in mind. The global registries for DMD and SMA are recognized as a leading resource for clinical 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 they provide an important connection to the research community. The registries are governed by a charter and by an oversight committee which includes 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 clinical trials for which they may be eligible
• Inbuilt patient data protection: patients are only contacted through their national registry; their data is kept confidential

Outcome measures research and validation

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 clinical 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.

Regulatory interactions

TREAT-NMD has proactively engaged with the European and US regulatory agencies, the EMA and FDA, over key issues surrounding clinical trials in neuromuscular disorders. The lack of previous clinical 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 all.

Case Study:
In 2011, the EMA published a concept paper on the need for a guideline on the treatment of Duchenne and Becker muscular dystrophy, which it released for public consultation. TREAT-NMD submitted its response to this concept paper in September 2011 and at the time invited EMA to meet with TREAT-NMD representatives to discuss our comments in more detail. In March 2013, the EMA published its draft guideline on the clinical investigation of medicinal products for the treatment of Duchenne and Becker muscular dystrophy, which was released for public consultation. TREAT-NMD worked with its stakeholders to prepare a submission to the consultation, which also included a workshop held on the 21st June 2013 in London to discuss the issues raised in the draft guideline. The workshop brought together academics, patient representatives, industry representatives and individual experts who provide advice to EMA committees. TREAT-NMD submitted its response to the draft guideline in August 2013.

The Care and Trial Site Registry

The Care and Trial Site Registry (CTSR) has provided information on personnel, facilities and patient populations across clinical centres with expertise in the neuromuscular field since 2007. In 2013, it was expanded to also cover the field of rare neurodegenerative diseases (NDD). It has functioned as a database of care and clinical trial sites, providing information to industry, health professionals and patients seeking specialized centres worldwide.

The CTSR has developed a facility (called PhenoSearch) which offers the possibility to collaborate with other researchers treating patients with a similar phenotype. PhenoSearch will make it easier for researchers and clinicians across the world to share information about undiagnosed patients and will hopefully facilitate collaboration between groups through ‘match-making’ those with similar cases.

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 clinical trials in SMA. In order for clinical trials to move through the approval process without delays, consensus between 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 through 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.

Regulatory affairs database

The TREAT-NMD Regulatory Affairs Database is a valuable source of advice to people who are involved in the planning of mono- or multi-centre clinical trials within different European countries. It contains contact addresses of national authorities as well as national legislation and other useful documents.

The database has been developed by the Clinical Trials Unit of the University Medical Center, Freiburg within the TREAT-NMD Alliance and in co-operation with the European Clinical Research Infrastructures Network (ECRIN), information provided in this database is updated on a regular basis (at least once a year).

Advisory and service 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.
Through TREAT-NMD companies can not only receive advisory support from leading neuromuscular experts, but also assistance in locating the centres with the expertise to conduct clinical 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.

**Case study: Industry enquiries**

A number of companies have requested feasibility data and support with patient recruitment from the TREAT-NMD patient registries and Care and Trial Site Registry. Examples of these 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
- assistance with the recruitment of patients for phase III study

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 clinical 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 clinical 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 has made these available in multiple languages in a family-friendly form to patients worldwide. Standards of care for spinal muscular atrophy and Duchenne muscular dystrophy and congenital muscular dystrophies are available through the TREAT-NMD website and as 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.

**Registries Toolkit**

TREAT-NMD, along with many patient organizations, is experienced in the creation and implementation of registries for neuromuscular conditions. All registries are tailored to not only the disease they cater for but also the location in which they operate, making the creation of each registry a unique process. A registry can vary by purpose and design and can be a very simple collection of data through to an elaborate database using bespoke
software. However, many common factors still exist and TREAT-NMD has developed a toolkit which outlines some of the fundamental issues that should be considered when setting up any type of rare disease registry.

**Bringing real patient benefit**

At the heart of the TREAT-NMD is a commitment to bring 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. Close relationships with the advocacy groups in the field have been important to TREAT-NMD’s success. TREAT-NMD can offer to the patient organisations, both disease-specific groups and those with a wider remit, advice, expertise and key resources. In return, researchers and clinicians within the TREAT-NMD Alliance as well as industry and regulatory agencies, can benefit from the knowledge and experience that patients provide.

Also, TREAT-NMD is undertaking research to explore, identify and examine ethical and social issues in clinical research of neuromuscular disorders. The Project Ethics Council identifies and examines ethical and social aspects of research taking place in the context of TREAT-NMD.

**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

**Case study: DMD care standards generation**

The publication in January 2010 in The Lancet Neurology of international consensus standards of care for Duchenne muscular dystrophy (DMD) marked the culmination of a three-year process led by the US Centers for Disease Control and Prevention (CDC). Under the managing editorship of the then-TREAT-NMD Network Coordinator Kate Bushby, 84 international experts in all areas of DMD care rigorously reviewed more than 70,000 clinical scenarios to create the consensus guidelines. TREAT-NMD produced interim recommendations in advance of the full publication to ensure that patients had access to best-practice care recommendations as soon as possible. Subsequently, a “Family Guide” was created through close collaboration between TREAT-NMD, advocacy groups and healthcare professionals, which sets out the care recommendations in lay terms. As of 2014, this is available in 30 languages, with additional translations in progress. Both the Lancet recommendations and the Family Guide have been used around the world for lobbying and negotiations with healthcare authorities for better care provision, including as part of the CARE-NMD project in Europe. The recommendations are also supported by pharmaceutical companies, who recognise that standardised baseline care and full genetic diagnosis of all patients is critical to successful multicentre clinical trials. In 2014, the CDC began a review process to ensure that the care recommendations reflected the latest clinical knowledge of DMD, and to include additional areas of care such as emergency and adult care. TREAT-NMD is closely involved in this process in close collaboration with healthcare professionals and patient advocacy organisations.

**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 document written in a more accessible style 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.

Case Study: Myotonic Dystrophy: Preparing for Clinical Trials

In 2009 TREAT-NMD, in collaboration with the Canadian based Marigold Foundation, organised a workshop to assess clinical trial-readiness in myotonic dystrophy. This workshop was attended by 26 participants representing eight countries and included scientists, clinicians, patient representatives and industry. The workshop built upon previous tools developed as part of the TREAT-NMD network for patient registries and outcome measures.

One of the main outcomes of the workshop was an international consensus on a dataset for myotonic dystrophy patient registries. This includes a limited number of mandatory data items (e.g. genetic confirmation, date of birth, name, and address) and additional highly encouraged items (e.g. cardiac, pulmonary information). This dataset has been implemented in an ever growing number of national patient registries around the world allowing a virtual global registry to be created.

The TREAT-NMD Alliance continues developing and sharing best practice in myotonic dystrophy and is part of the European Commission funded OPTIMISTIC study. A collaboration between European partners working to improve quality of life and clinical practice for these patients.

Collaborative initiatives

In uniting such a wide range of neuromuscular specialists and other key stakeholders, TREAT-NMD has been 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. Below you can find examples of some of the TREAT-NMD related projects:

3GB Test

New technologies that allow efficient sequencing of a whole human genome in a diagnostic setting will have an enormous impact on diagnostic centres replacing many existing molecular and cytogenetic tests. Patients deserve to benefit from our vastly growing knowledge on functional genomics. 3Gb-testing is the ideal method to bring these benefits to the public.

This project aims to prepare Europe for innovations in molecular testing. Quality assessment schemes, HTA and guidelines have to be in place. Healthcare professionals must be aware of the impending change and potential impact on practice. The Consortium will inform the healthcare community and make recommendations to the European Commission, the European Society of Human Genetics, and national organizations relevant to this field. A key output will be a validated roadmap for the implementation of diagnostic genome sequencing in Europe.

BIOIMAGE-NMD

The overarching objective of the BIOIMAGE-NMD project is to deliver combined structural and molecular imaging biomarkers with proven utility for the detection of therapeutic effects in patients with rare neuromuscular diseases (NMD). The project will have three specific objectives:
To develop a new generation of muscle diffusion Magnetic Resonance Imaging (MRI). This diffusion imaging technology will be used to augment a state of the art simultaneous MRI / Magnetic Resonance Spectroscopic Imaging (MRSI) protocol for quantitative muscle imaging.

To provide a proof of principle in Duchenne Muscular Dystrophy (DMD) that simultaneous MRI/MRSI can be used as a biomarker to monitor therapeutic efficacy in clinical trials in neuromuscular diseases.

To develop a novel simultaneous Positron Emission Tomography (PET)/MRI technology to advance innovative drug development programmes for personalised medicines based on Antisense Oligonucleotide technology.

**COST Actions**

**MYO-MRI** - Applications of MR imaging and spectroscopy techniques in neuromuscular disease: collaboration on outcome measures and pattern recognition for diagnostics and therapy development”, is the COST Action BM1304. The project aims to improve diagnosis and understanding of muscle pathology: online atlas, develop multicentric imaging outcome measures: SOPs, explore new contrasts, targets and imaging techniques for NMD: clinical testing and look into strategies for muscle imaging texture analysis: validated algorithms.

**The Exon Skipping COST Action** BM1207 aims to advance the development of antisense-mediated exon skipping for rare diseases, focusing on Duchenne muscular dystrophy for which this approach is currently assessed in phase 3 clinical trials. The project involves all key stakeholders (scientists, clinicians, regulators, industry and patients) this COST Action aims to overcome challenges through networking to allow clinical implementation of antisense-mediated exon skipping for as many rare disease patients as possible.

**EUCERD Joint Action: working for rare diseases**

The EUCERD Joint Action: Working for Rare Diseases, was originally established to support the activities of the European Union Committee of Experts on Rare Diseases (EUCERD). The project supports the European Commission in formulating and implementing the European Union's activities in the rare disease field, by facilitating the exchange of policies and practices between all stakeholders and Member States. The project is active across many diverse areas of the field, and is expected to deliver numerous outcomes such as:

- accelerated support for the development and implementation of national plans and strategies for rare diseases:
- the comprehensive coding and classification of rare diseases, to ensure visibility of these conditions in healthcare systems and to produce a greater quantity of interoperable data:
- a demonstration of the importance of developing specialised social services for people living with rare diseases (beyond the purely medical sphere);
- means of improving access to higher quality healthcare for rare disease patients, regardless of where they live in the EU;
- the development of a model for the integration and sustainability of rare disease initiatives and infrastructures.

The 'mini-projects' within the overall Joint Action will produce shared tools and resources for the benefit of the whole rare disease community, and will also define and publicize the state of the art in terms of rare disease activities in Europe.

**Neuromics**

The NEUROMICS is an integrated European project on omics research in rare neuromuscular and neurodegenerative diseases. Its work plan integrates a group of expert centres with five aims:

- to unravel a large portion of still unknown genetic causes of ten categories of NDD and NMD using next-generation genomics approaches;
- to provide a diagnostic strategy to detect most genetic causes in clinical practice;
- to develop biomarkers which better describe phenotype and predict progression using transcriptomic, proteomic, metabolomic, and lipidomic technologies;
• to use Omics approaches for disease pathway elucidation in animal models, as well as on patient derived iPSC and other cell types;
• to utilize Omics-based results for stratified therapy development for NMD/NDD.

OPTIMISTIC
An observational prolonged trial in myotonic dystrophy type 1 to improve QoL standards, target identification collaboration.

The aim of OPTIMISTIC is to improve clinical practice in the management of patients with Myotonic dystrophy for which no dedicated treatment is currently available. To this end OPTIMISTIC compares two intervention regimes (exercise training and cognitive behavioural therapy (CBT)) and evaluates their effectiveness and adverse events, with particular attention given to the definition of appropriate outcome measures and new clinical guidelines for DM1 management. Relevant stakeholders such as national and European patient organisations are engaged and rapid uptake of developed guidelines is ensured by dissemination plans involving these stakeholders as well as the international TREAT-NMD network and other DM1 specific networks.

RARE-Bestpractices
This is a platform for sharing best practices for management of rare diseases. The aim of Rare-Bestpractices is to improve clinical management of RD patients and narrow the existing gap in quality of health care among EU member states as well as in other countries, by collecting, evaluating and disseminating best practice recommendations and sharing knowledge globally.

Rare-Bestpractices will develop a sustainable global networking platform, supporting the collection of standardized and validated data and the exchange of knowledge and reliable information among 14 countries. This project fosters synergistic collaboration among experts, patient representatives, agencies, institutions, networks and organizations experienced in best practice guidelines development, at a regional, national and international level.

RD-Connect
An integrated platform connecting registries, biobanks and clinical bioinformatics for rare disease research.

RD-Connect is a unique global infrastructure project that links up databases, registries, biobanks and clinical bioinformatics data used in rare disease research into a central resource for researchers worldwide.

In a six-year project funded by the European Union but uniting researchers across the world, it will develop an integrated research platform in which complete clinical profiles are combined with -omics data and sample availability for rare disease research, in particular research funded under the International Rare Diseases Research Consortium (IRDiRC).

SCOPE-NMD
The main goal of the FP7 funded SCOPE-DMD project is to further advance and accelerate the development of PRO045, an exon-skipping compound for DMD. PRO045 is currently in a Phase IIb dose-escalating clinical trial to assess its safety and efficacy. The chemically-modified AON induces exon 45 skipping in the dystrophin gene and could be suitable for approximately 8% of all DMD patients.

SKIP-NMD
The idea of the SKIP-NMD project is to perform a safety assessment of a lead PMO compound to skip exon 53, and perform a randomised study looking at safety, biochemical efficacy and exploratory clinical efficacy of this novel antisense in ambulant boys affected by DMD. Exploratory measures will be analysed including muscle MRI and serum biomarkers to assess non-invasively response to therapy. Three UK charities (AD; MDC; DMS support group), one Italian (DMP Parent Project Italy, and one French (AFM) are involved in this project.
There are a number of companies involved in the project, from those involved in PMO production, to companies which manufacture some of the new outcome measure equipment, to companies involved in preclinical toxicology and MRI data acquisition and analysis.