European Reference Network
for rare or low prevalence complex diseases

Network Neuromuscular Diseases (ERN EURO-NMD)

How can ERNs add value to clinical research in RD and highly specialised domains?

EURO-NMD Case study

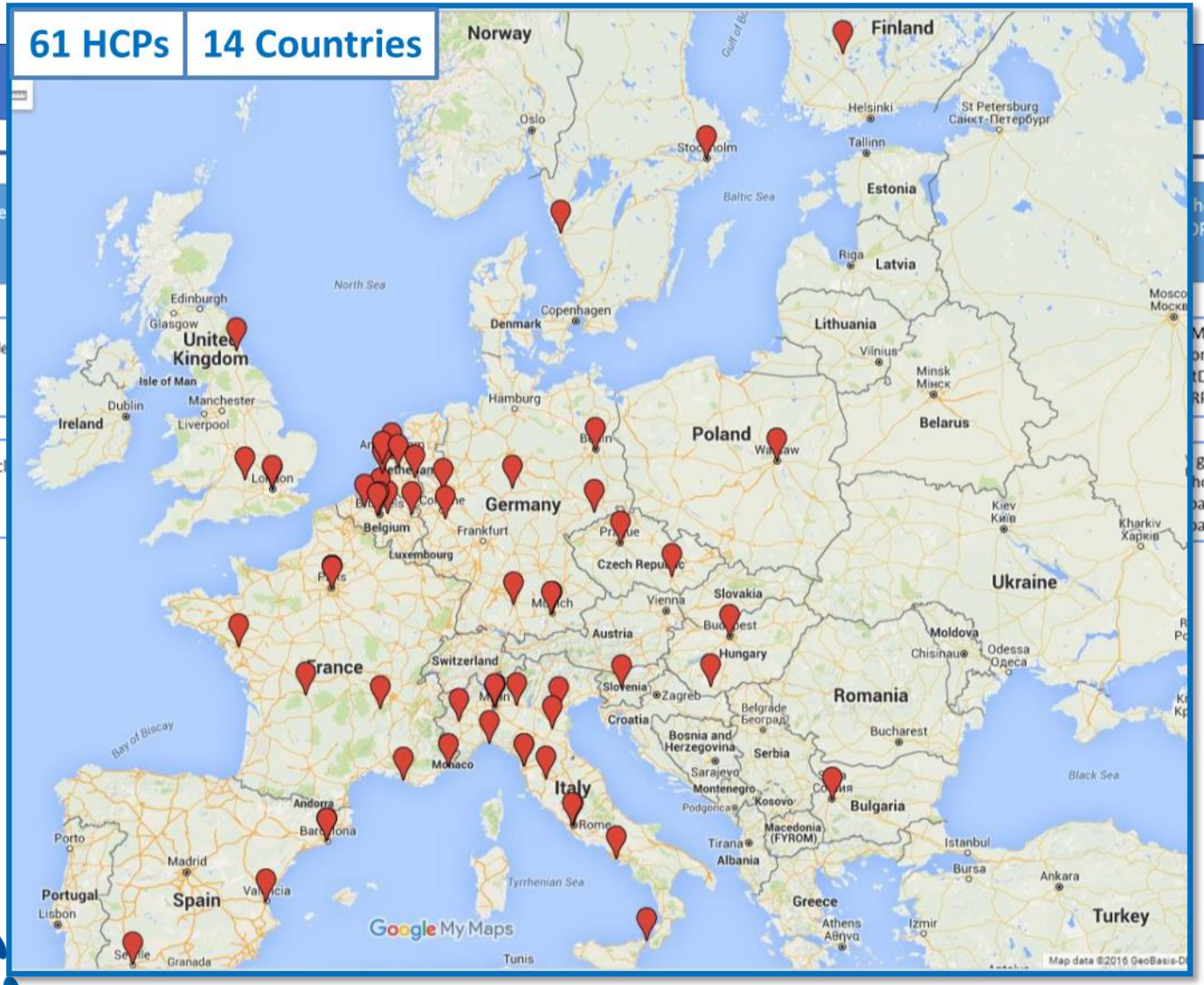
Teresinha Evangelista

RD-ACTION, European Medicines Agency, and DG SANTE Workshop

Rare muscle disease
ORPHA98472
1 in 3500

Aquired skeletal muscle diseases
ORPHA 206638

Genetic skeletal muscl disease
ORPHA 206634



hondrial Diseases
RPHA68380
1in 5000

Mitochondrial
omyopathies caused by
tDNA mutations
RPHA 68380

gene defects causing
hondrial encephalo-
pathy, myopathy or
pathy ORPHA 68380

EURO-NMD Strategic Research Plan

Main goals:

- Promote research activities within the network
- Ensure a harmonized baseline that will enable standardization and reuse of network data and samples for research
- Develop better Research Services

Key deliverables for the first year:

- Mapping of research infrastructures and studies for NMD patients

- Agreed consent elements
- Agreed data sharing standards and mechanisms

- Database of shared samples through deposition at biorepositories and –omics data with associated phenotypic data via submission to databases and RD-Connect

TREAT-NMD
Neuromuscular Network

Home | Membership | Newsletter | Intranet | Events | Cookies | Contact

About TREAT-NMD — All about the network
Resources — Tools and infrastructure
Research — Scientific and clinical
Care — Global best practice
Industry — Service overview
Disease Information

Resources
- Resources overview
- Care overview
- Neuromuscular care and trial centres
- Post marketing surveillance
- Patient Registries
- TACT
- Outcome measures
- BioBanks
- Social & ethical framework
- Training & education
- Regulatory affairs database
- Resources for researchers

Resources available through the network

"TREAT-NMD provides tools and infrastructure to help the neuromuscular field collaborate better internationally, addressing areas that often get missed in individual research projects. From patient registries to international consensus publications, the resources below are available to clinicians, researchers, industry and patients across the world."

Patient registries
Find out about different types of national and international registries for over ten neuromuscular diseases

Care and trial site registry
A database of clinical sites and medical centres caring for patients with NMDs and participating in clinical trials

Outcome measures
"Outcome measures" are the tests that investigators perform to decide whether a treatment being tested in a clinical trial is having any effect

TACT
An expert multidisciplinary body providing independent and objective guidance on advancing new therapies for NMDs

Biobanks
EuroBioBank: a network of biobanks distributing DNA, cell and tissue samples to scientists conducting research on NMDs

Regulatory affairs
A valuable source of advice to people who are involved in the planning of mono- or multi-centre clinical trials

Training and education
Information on specialist training courses covering neuromuscular disorders

Social and ethical framework
TREAT-NMD is undertaking research to explore, identify and examine ethical and social issues in clinical research of neuromuscular disorders

Registries

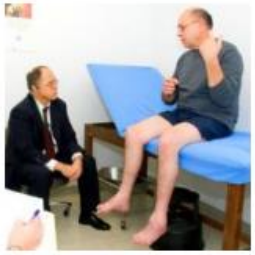
The registries were developed to:

- Help **researchers** to answer questions such as:
  how common the individual diseases are
- Support activities to improve patient care, such as the assessment of care standards in different countries.
- Help **Pharmaceutical** companies interested in locating patients for a clinical trial.
- Facilitate contact with **Patients.** Patients will be informed through their own national registry of upcoming trials

CMD
Congenital Muscular Dystrophies

Congenital Myasthenic Syndromes

CMS
Congenital Myasthenic Syndromes



CMT
Charcot Marie Tooth Disease

DM
Myotonic Dystrophy

DMD & BMD
Duchenne & Becker Muscular Dystrophy

Facioscapulohumeral Muscular Dystrophy

FSHD
Facioscapulohumeral Muscular Dystrophy

GNE / HIBM
Hereditary Inclusion Body Myopathy

LGMD
Limb Girdle Muscular Dystrophies

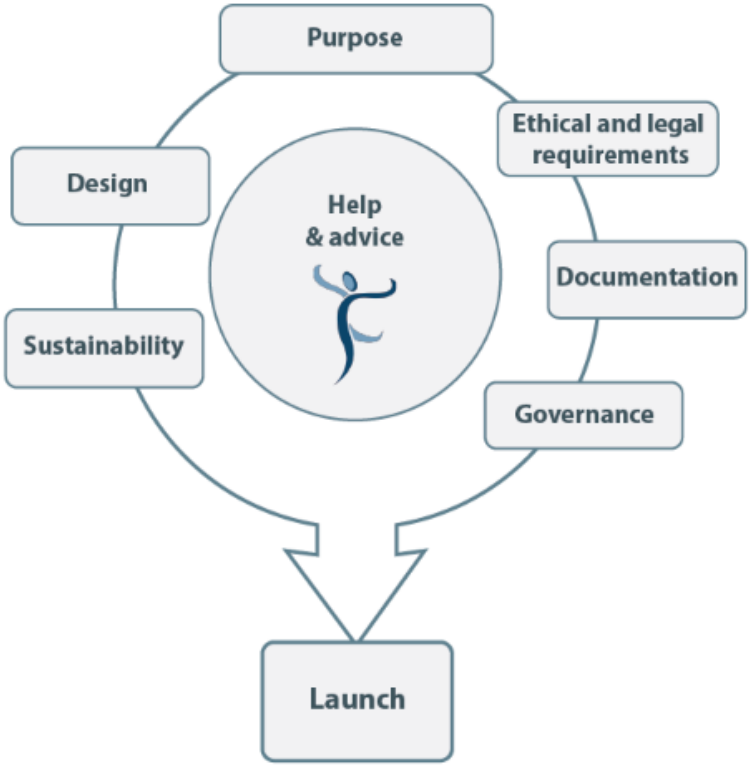
MTM / CNM
Myotubular & Centronuclear Myopathy

SMA
Spinal Muscular Atrophy

Registries tool kit

Things to consider when setting up a registry

TREAT-NMD, along with many patient organizations, are 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 to an elaborate database using bespoke software. That said many common factors still exist and we have tried to outline some of the fundamental things that should be considered when setting up any type of rare disease registry. Click on the boxes below for more information.



Care and Trial Site Registry – CTSR

- One of the major hurdles to overcome before initiating a clinical trial is to **identifying trial sites** able to

  recruit enough patients

  offer a specific standard of care

  offer experience in clinical trials

- The concept behind the CTSR was to **collect information** on **personnel, facilities and patient population** to help industry and clinical investigators select trial sites, and to help to **identify potential partners** for upcoming research projects.

Care and Trial Site Registry – CTSR

Established in 2007

It is a flexible database with the ability of being expanded with pertinent questions
From 2013 includes data on NMD and Neurodegenerative diseases

Registration at the CTSR was one of the specific criteria for the EURO-NMD HCPs

Advantages:

Real knowledge of the infrastructure of the centres

Allows identification of gaps in patient care

Information gathered in the CTSR

- **General Site Information**
- **Patient Cohorts**
  - Number of patients and available diagnostic tools
- **Clinical Trial Infrastructure**
  - Personnel and experience, GCP training, equipment
- **Care Settings**
  - Members of interdisciplinary team
  - Arrangements for transition from paediatric to adult care
  - Pulmonary and cardiologic care
- **Research and Education**
  - Participation in clinical trials
  - Peer-reviewed publications
  - Participation in networks
  - Training activities

TREAT-NMD Advisory Committee for Therapeutics (TACT)

- Established in 2009, TACT is a unique structure constituted by a multi-disciplinary international group of well recognized **academic** and **industry** drug development **experts** as well as representatives of **patient** foundations and **institutional and governmental scientific research centres**

- Review and provide guidance on the translation and development path of therapeutics programs in rare neuromuscular diseases.
    - Review therapeutics with the long-term goal of an intended clinical trial and potential registration.
    - Address issues of drug formulation, bioavailability and toxicology
    - Address possible regulatory requirements and marketing considerations.
    - Provide applicants with a comprehensive written review

Future (EURO-NMD/TREAT-NMD)

- EURO-NMD will make use of these available tools as a baseline for its clinical research activities (agreements have been established)
- The remit of the disease groups of EURO-NMD is broader than in TREAT-NMD; we will work together to expand and share activities to other disease areas (funding?)
- Concern: how to replicate the success of TREAT-NMD in engaging with Industry – It is a **safe, ethically-sound** and **mutually beneficial model** developed over many years… how to continue?

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Neuromuscul Disord. 2018 Apr 20. pii: S0960-8966(18)30196-2. doi: 10.1016/j.nmd.2018.04.009. [Epub ahe

Meeting report of the "Regulatory Exchange Matters" sessio
Conference:: Lessons in communication: How an early dialo
academics can further therapy development for neuromuscu
29 November 2017.

Aartsma-Rus A[1], Mercuri E[2], Vroom E[3], Balabanov P[4].

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Orphanet J Rare Dis. 2015 Apr 23;10:49. doi: 10.1186/s13023-015-0258-1.

The TREAT-NMD advisory committee for therapeutics (TACT): an innovative de-risking model to

nberg R[8], Flanigan KM[9], Kaufmann P[10], McNeil E[11], Mendell

) in cell and animal models only a handful resch clinical

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Orphanet J Rare Dis. 2013 Oct 23;8:171. doi: 10.1186/1750-1172-8-171.

The TREAT-NMD care and trial site registry: an online registry to facilitate clinical research for neuromuscular diseases.

Rodger S, Lochmüller H, Tassoni A, Gramsch K, König K, Bushby K, Straub V, Korinthenberg R, Kirschner J[1].

Author information

Author information

Abstract

Facilitating orphan drug development: Proceedings of the TREAT-NMD International Conference, December 2015, Washington, DC, USA.

Collaborators (44)

Author information