EURO-NMD: the European Reference Network for rare neuromuscular diseases

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What is an ERN?

• A network of health care providers/centres providing highly specialised healthcare, with the purpose of improving access to diagnosis, treatment and care for patients across Europe”
Building bridges and breaking barriers in rare neuromuscular diseases

61 HCPs | 14 Countries

15 Italy
8 France
8 Germany
5 Spain
5 Belgium
5 The Netherlands
4 UK
2 Czech Republic
2 Hungary
2 Sweden
1 Poland
1 Bulgaria
1 Finland
1 Slovenia
Rare Neuromuscular Diseases (NMDs) (ORPHA68381)

- Muscle Diseases
  Coordination: Marianne de Visser
  - Acquired
    - Inherited
  - Inherited

- Peripheral Nerve diseases
  Coordination: Mary Reilly
  - Acquired
    - Inherited

- Motor Neuron Diseases
  Coordination: Leonard van den Berg
  - ALS and related disorders
  - SMA and related disorders
  - Congenital Myasthenic Syndromes

- Neuromuscular Junction Defects
  Coordination: Hanns Lochmüller
  - Myasthenia Gravis & Other autoimmune NMJ defects
  - Mitochondrial encephalomyopathies caused by mtDNA mutations
  - Nuclear gene defects causing Mitochondrial encephalomyopathies, myopathy or neuropathy

- Mitochondrial diseases
  Coordination: Rita Horvath

Building bridges and breaking barriers in rare neuromuscular diseases
Diagram 1 – Care Pathways
1st year Goals

i. To establish a functional Europe-wide network
ii. To collate and review existing information on NMDs
iii. To provide a map of expertise across Europe and define pathways for clinicians and patients to access the centres of excellence;

These primary goals were for most achieved
EURO NMD

Interaction with other ERNs and Learned Societies
Interaction with the HCPs and PAB
Interaction with the EC

Working Groups
- Organisation
- Mapping the expertise
- Defining goals
- Guidelines
- Other

What about the Future

Building bridges and breaking barriers in rare neuromuscular diseases
Interaction with the European Commission

➢ **IT advisory Board**

EURO-NMD is part of the IT advisory board
The IT advisory group gives advice to the EC on the development of the ERN IT Platform: ERN Collaborative Platform and the CPMS
Currently working on a specific data-set for NMD

➢ **Cross-border healthcare and business continuity working group**

EURO-NMD co-chairs this WG with the coordinator of the ERN-RND
EURO-NMD – Year One
achievements

Website Launched in June 2017

First Conference Nov 17

...all this and the secretariat did not reach full capacity until October 2017

59 of 61 HCP fully engaged

CPMS Piloted and ready to refer
EURO-NMD current work

**Prioritising PROMS:**
- ALS
- SMA
- Myasthenia Gravis
- Peripheral Neuropathies and
- Myotonic Dystrophy

**Working on new guidelines:**
- Guillain-Barre Syndrome
- Myositis
- GNE Myopathy
- Paediatric Myasthenia Gravis

**Starting Work on S4R and SOLVE RD**

**Planning the Summer School and 2018 training programme**

**Receiving and managing the diagnosis and treatment of the first patients through CPMS**

Building bridges and breaking barriers in rare neuromuscular diseases
EURO-NMD involvement in ENMC workshops proposals

• 238th ENMC International Workshop on “Updating management recommendations of cardiac dystrophinopathy (CARDIAC)”. This workshop will be held from 30 November – 2 December 2018

• 236th ENMC International Workshop on “GNE myopathy (also known as HIBM, Nonaka disease, and quadriceps sparing myopathy) (GNEM). This workshop will be held from 14 – 16 September 2018

• ENMC workshop proposal - Designing a roadmap of genetic diagnosis needs for Neuromuscular patients : meeting ERN EURO-NMD mission
PAB
Chair: Francois Lamy Co-Chairs: Florence Duguet

• **Primary focus for year 1:** establishing work processes
  
  ✓ recruitment and appointment procedure for new members
  ✓ terms of reference
  ✓ PAB booklet

• There is a need to **enhance cooperation between HCPs and patient representatives.**
Education Advisory Board
Chair: Andoni URTIZBEREA Co-Chair: Jean-Philippe PLANCON

- > 80 people core group of 10 members

- **Priority:** questionnaire to identify existing resources and the unmet needs

- Euro-NMD and TREAT-NMD endorsed Neuromuscular **Translational Summer School**

- Define **criteria for endorsement of educational events**

- Organisation of a teaching course to be held at the next EPNS meeting in Athens 2019 with the EUROPEAN PAEDIATRIC NEUROLOGIC
Education Plan - Initiatives

For the Website/Collaborative platform:

• Planned **2 Webinars** on thematic areas
  eg: neurophysiology (MND diagnosis; CMT diagnostic...)
  eg: Inflammatory myopathies diagnostic or workout of a suspicion of a mitochondrial disease

• Planned **educational videos** on basic aspects such as:
  MMT
  CMS/MG assessment
  Processing of a muscle biopsy/nerve biopsy
NEUROPHYSIOLOGY
Chair: Peter Van den Bergh and Camiel Verhamme  Co-Chair: Lucio Santoro

• Neurophysiology expertise and representative questionnaires

• Future projects
  Collecting data on neurophysiological practice in the HCPs
  Creating awareness about differences in practices
  Endorsing and supporting guidelines and consensus statements; making them available for reference to the HCPs
  Inclusion of experts in specific neurophysiology fields
  Supporting initiatives to standardise digital export format of ENMG machines
Euro-NMD imaging group
Chair: Pierre Carlier

• Main focus:
  tests of the image handling in the CPMS and readiness for HCPs requests

  collaboration with the MYO-MRI scan-bank with the objectives of
  1/ giving access to the HCPs to the NMD teaching atlas of scan-bank,
  2/ enrichment of scan bank with imaging cases solved by Euro-NMD

Organise imaging teaching courses
  2-day workshop - Institute of Myology in Paris, June 25th and 26th.

# Existing SOPs for muscle imaging have been selected

# New SOPs are due in 2018:

  NMR imaging update and an MRN (magnetic resonance neurography)
Pathology
Chair: Bjarne Udd (Finland)  Co-Chair: Anders Oldfors (Sweden)

• Working on recommendations for harmonizing neuromuscular pathology methodological practices
  recommendations on reporting pathology findings in order to create systemic platforms

• Currently working on drafts for these recommendations

• Planned: a face-to-face meeting towards the end on 2018 to finalize the recommendations.
Genetics
Chair: Alexandra Ferlini Co-Chairs: Serenella Servidei, Borut Peterlin, Silvere van der Marel

AIMS
• Revising genetic diagnostic guidelines, educational aspects, data sharing.
• NGS methodologies and applications into clinical practice

GOALS
• Questionnaire to list diseases genetically diagnosed by the ER-NMD HCPs
• Strong interaction with RD-Connect for data sharing
• Questionnaire shared with other ERNs (rare neurological, Epicare, etc.)

• **ENMC workshop proposal** - Designing a roadmap of genetic diagnosis needs for Neuromuscular patients: meeting ERN EURO-NMD mission.

• **Transversal Genetic TASK** – preparation for a Cost action
Progress on setting up the ERNs

• Challenges

✓ **Motivate** 61 HCP to be active and engage with the work
✓ Delineate the **European map** of existing resources/expertise and practices
✓ **Transparency** towards partners regarding the closed calls for grants
✓ **Co-funding** mechanisms for grant application
✓ **CPMS** and the CEF grant delay
✓ **Indemnity** regarding the written opinions (e-Consultation)
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“EURO-NMD is one of the 24 European Reference Networks (ERNs) approved by the ERN Board of Member States. The ERNs are co-funded by the European Union (Health Programme and CEF). For more information about the ERNs and the EU health strategy, please visit ec.europa.eu/health/ern”