



How can EURO-NMD help develop and implement care guidelines for NMDs?

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# How can EURO-NMD help develop and implement care guidelines for NMDs?

- What are ERNs
- EURO-NMD in a glance
- Proposed plans and methodologies for guideline development and implementation
- Stumbling blocks in the production of guidelines for RD
- Specific ERN related problems
- How to try to move forward



# What are ERNs?

- **Virtual networks** connecting expert centres across Europe
- **Aim:** Tackle complex or rare diseases and conditions that require highly specialised treatment and/or concentration of knowledge and resources



# How can ERNs change the way we look into RD?

- Major focus on :

## e-HEALTH

\*allow access to the best expertise reducing the geographic barriers

\*allow timely exchange of knowledge

\*avoid unnecessary travel

## Training and education

- Can **facilitate large clinical studies** and **help with research** by **gathering large pool** of patient data and samples



# Some ERNs related numbers

- Approved in December 2016 and launched in March 2017
- > than 900 highly specialised healthcare units
- 313 hospitals in 25 Member States (plus Norway)
- 24 ERNs in different areas of expertise

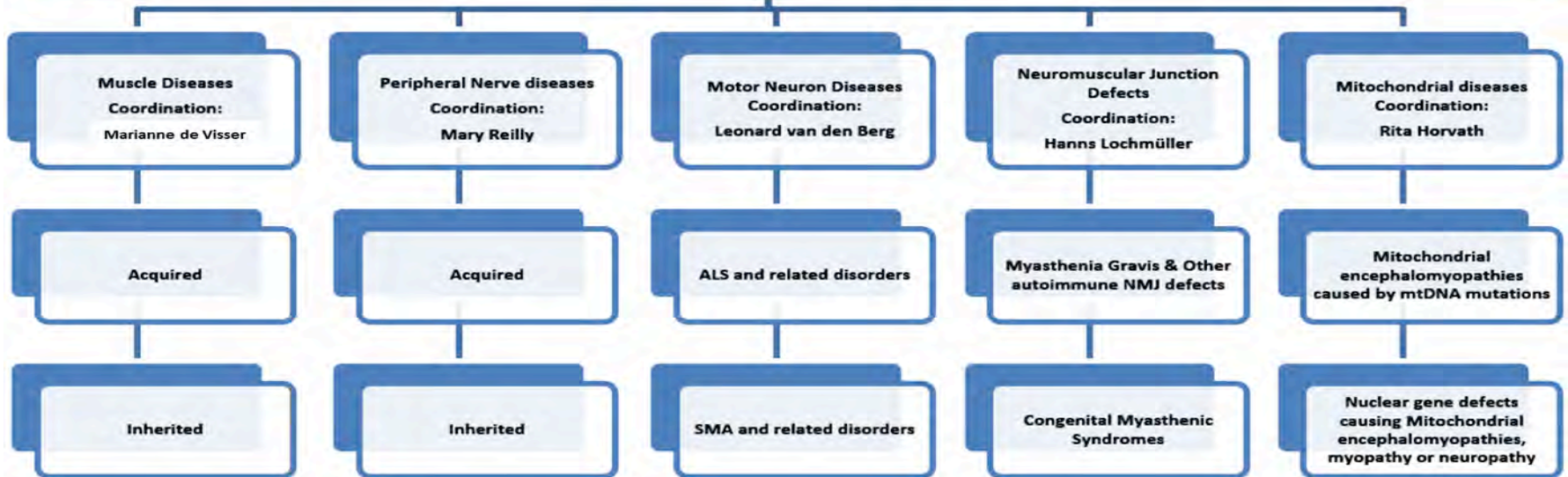


# ERN Directory

Endo-ERN	European Reference Network on endocrine conditions
ERKNet	European Reference Network on kidney diseases
ERN BOND	European Reference Network on bone disorders
ERN CRANIO	European Reference Network on craniofacial anomalies and ENT disorders
ERN EpiCARE	European Reference Network on epilepsies
ERN EURACAN	European Reference Network on adult cancers (solid tumours)
ERN EuroBloodNet	European Reference Network on haematological diseases
ERN eUROGEN	European Reference Network on urogenital diseases and conditions
ERN EURO-NMD	European Reference Network on neuromuscular diseases
ERN EYE	European Reference Network on eye diseases
ERN GENTURIS	European Reference Network on genetic tumour risk syndromes
ERN GUARD-HEART	European Reference Network on diseases of the heart
ERNICA	European Reference Network on inherited and congenital anomalies
ERN ITHACA	European Reference Network on congenital malformations and rare intellectual disability
ERN LUNG	European Reference Network on respiratory diseases
ERN PaedCan	European Reference Network on paediatric cancer (haemato-oncology)
ERN RARE-LIVER	European Reference Network on hepatological diseases
ERN ReCONNET	European Reference Network on connective tissue and musculoskeletal diseases
ERN RITA	European Reference Network on immunodeficiency, autoinflammatory and autoimmune diseases
ERN-RND	European Reference Network on neurological diseases
ERN Skin	European Reference Network on skin disorders
ERN TRANSPLANT-CHILD	European Reference Network on transplantation in children
MetabERN	European Reference Network on hereditary metabolic disorders
VASCERN	European Reference Network on multisystemic vascular diseases



## Rare Neuromuscular Diseases (NMDs) (ORPHA68381)











- 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



## Care

- E-Health (teleconsultation, multidisciplinary board for queries patient/tests related)
- **Guidelines, Outcome measures**

## Education

- Training and continuous education programmes
- Target Healthcare professionals, Patients, Public

## Research

- Clinical trials
- Registries
- Biobanks
- Networking; harmonization and standardisation of procedures



# Healthcare Guidelines

- Guidelines should be based on the best available evidence
- Should involve all relevant groups
- Are an ideal tool to support decision makers
- Help to reduce unnecessary variation in practice
- Should provide support rather than dictate care

**How can EURO-NMD help with the implementation and development of Guidelines?**



# EURO-NMD plan for the development of guidelines

- Gap analysis in different disease areas/diagnostic tools
- Evaluate and endorse existing guidelines
- Develop new guidelines where they do not exist or the provision is inadequate – Prioritise the most relevant ones
- Make those available through the web-site and monitor implementation



# EURO-NMD plan for the development of guidelines

Development of guidelines and outcome measures, is the responsibility of:

- **Specialist Disease Groups**
- **Diagnostic Tools Groups**

Each group will:

- Be supported by the **PAB**
- Will engage with **external advisors** when needed



# EURO-NMD plan for the development of guidelines

- ‘Gap Analysis Reports’; guideline prioritisation
- **To develop** guidelines: GRADE system, RARE-bestpractices work
- **Audit** existing guidelines: AGREE II system
- **Translations:** 24 EU official languages

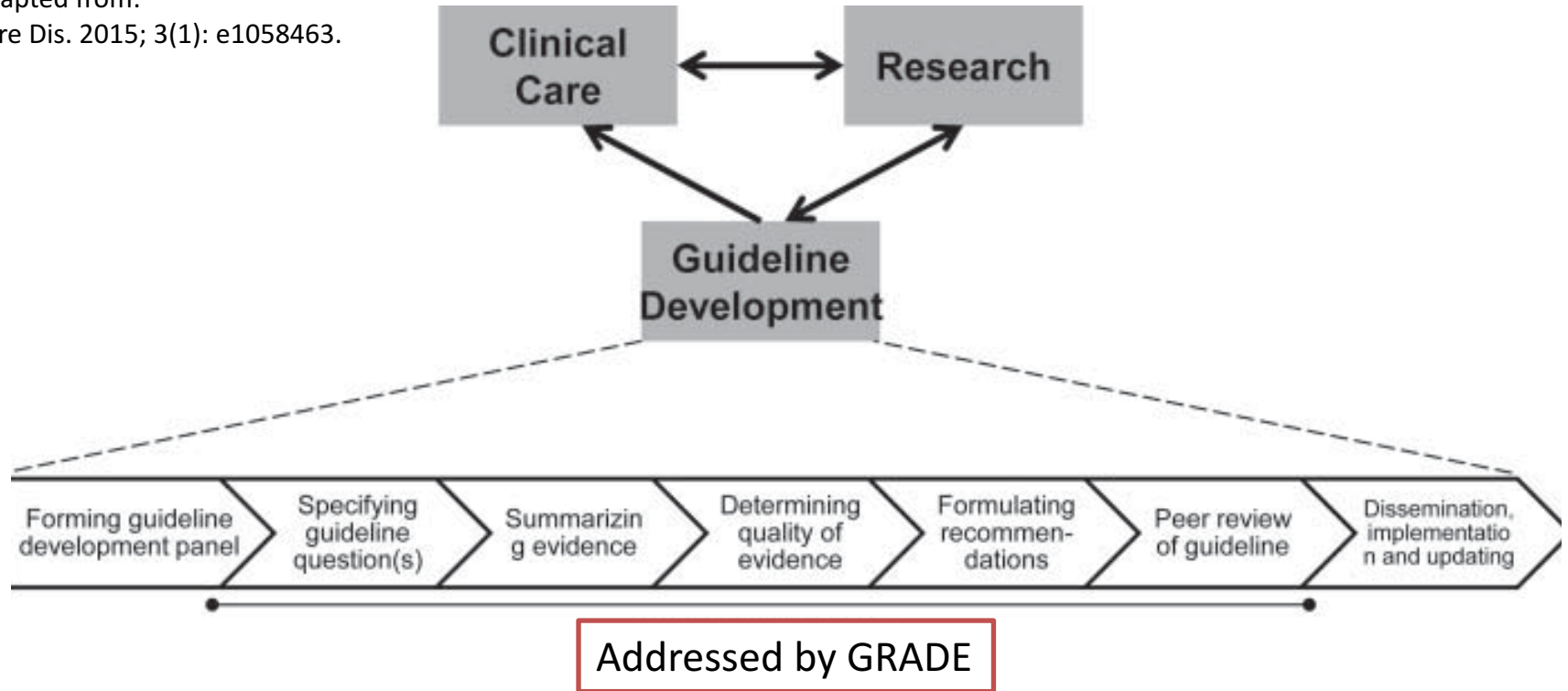
(HCPs + patient organizations)



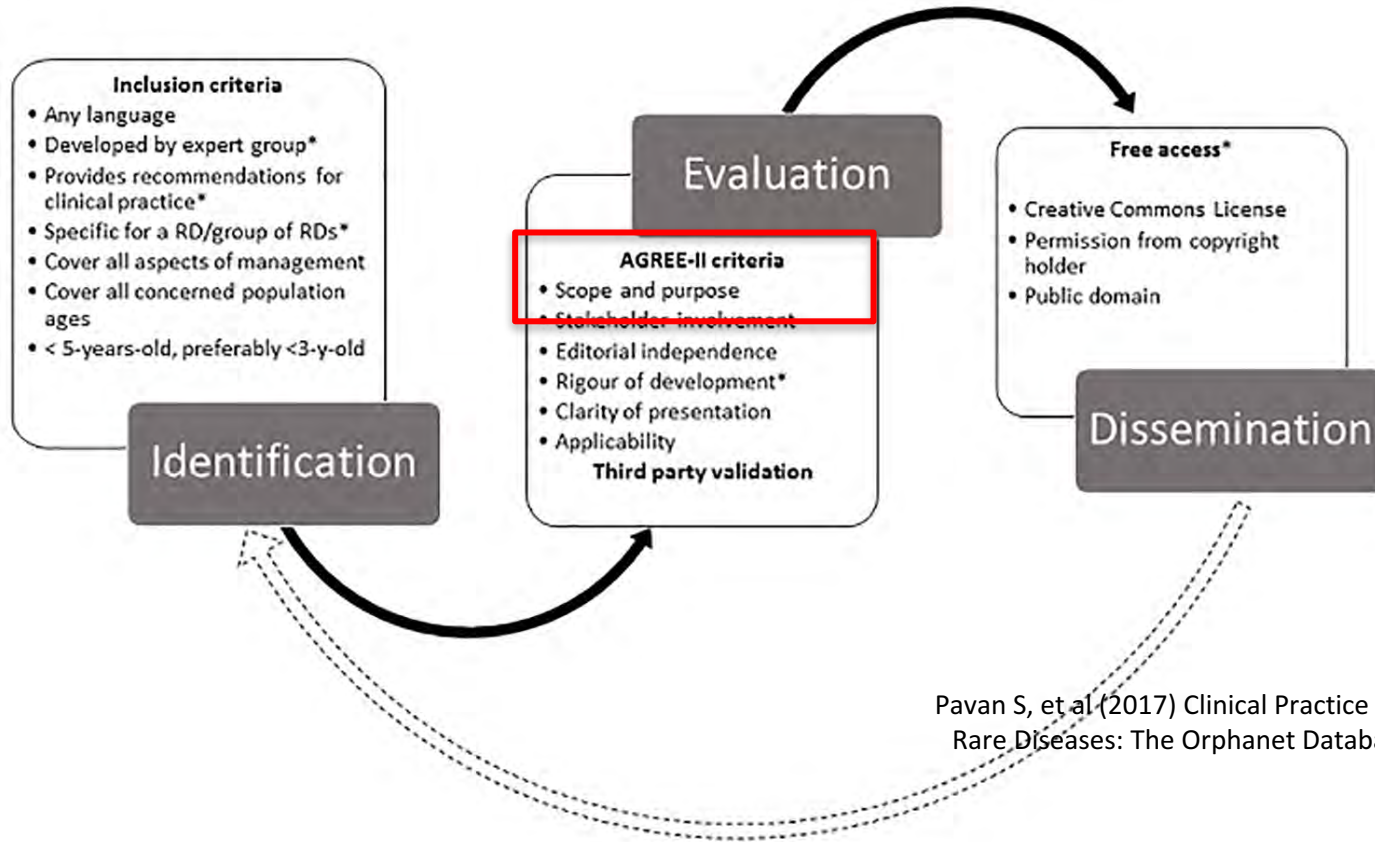


# RARE - bestpractices

Adapted from:  
Rare Dis. 2015; 3(1): e1058463.



# The ORPHANET database



Pavan S, et al (2017) Clinical Practice Guidelines for Rare Diseases: The Orphanet Database. PLoS ONE



# Constrains of producing guidelines for RD (NMD)

- How to prioritize guidelines and determine the level of relevance
- Scarcity of published data on RDs (low quality)
  - Rarity of randomised controlled trials
  - Limited therapeutic options
  - No published evidence for outcomes, or for patient values and preferences
- High costs of development of guidelines
- Lack of clinical expertise



# How the scene is changing

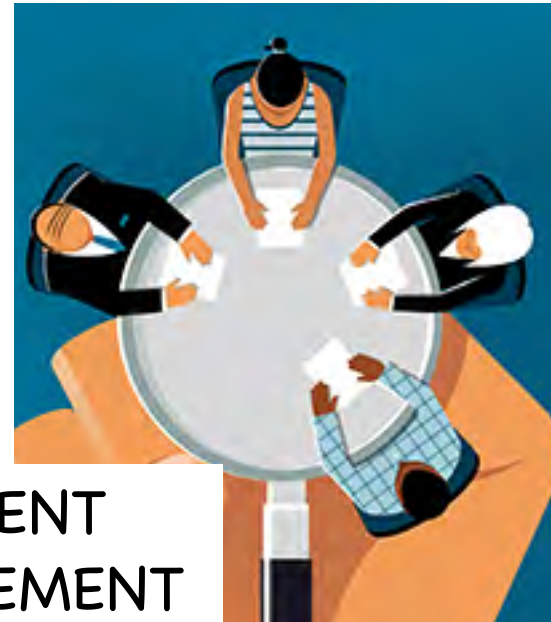
- RD have been a privileged target of health policies in Europe:  
dedicated funding,  
CBHCD,  
awareness campaigns,  
Centres of Expertise, National Plans for RD, ERN...
- The development and dissemination of guidelines for RDs has gathered increasing attention from Member States:  
shorten the time to diagnosis  
Improve the quality of care  
Reduce costs and help with resources allocation  
Promote equity in care



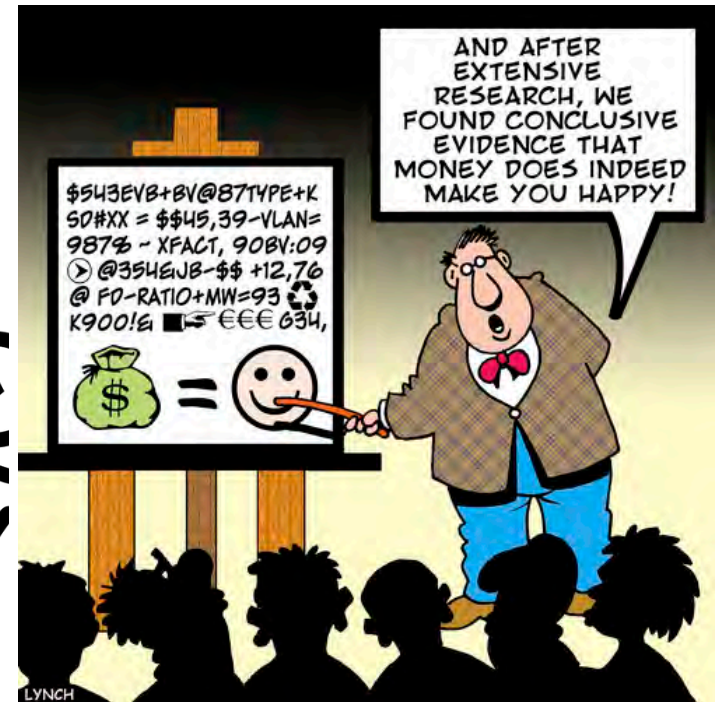
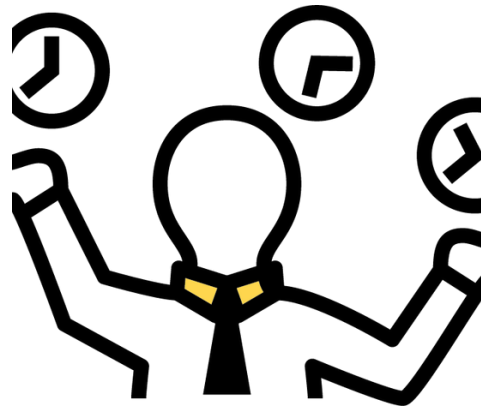
# Some problems are solved with EURO-NMD



PATIENT  
ENGAGEMENT



# Some problems persist





# How to move forward?

- Work in collaboration with learned societies
- Collaborate with other ERNs for the use of common tools
- Collaborate with Rare-Bestpractices, Orphanet
- Look for sponsorship for expert meetings (ENMC; EJP)



# Dissemination and implementation of guidelines

- Evidence about methodologies for guideline dissemination and implementation is **scarce and not evidence based**



## EURO-NMD dissemination strategies

- **Educational materials** (Scientific publications, Leaflets with lay and specialised information, Orphanet database, patient organisations, EURO-NMD web-site)
- **Educational meetings** (participation in conferences, lectures, workshops)
- **Audit the implementation** and feedback to the ERN members
- **Patient-mediated information**



## Summer School for NMDs 2-6 July 2018 Newcastle-upon-Tyne, UK

This course will address the following aspects as it travels along this established pathway.



## First EURO-NMD Meeting 29-30 November 2017 Freiburg, Germany

The network's healthcare providers will get the chance to meet face-to-face promote their work, and more.

 @euro\_nmd

[ern-euro-nmd.eu](http://ern-euro-nmd.eu)





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Building bridges and breaking barriers in rare neuromuscular diseases