European Reference Network
for rare or low prevalence complex diseases

Network
Neuromuscular Diseases (ERN EURO-NMD)

Coordinator
The Newcastle upon Tyne Hospitals NHS Foundation Trust — United Kingdom

DIAGNOSTIC TASK GENETIC TESTING

BOARD

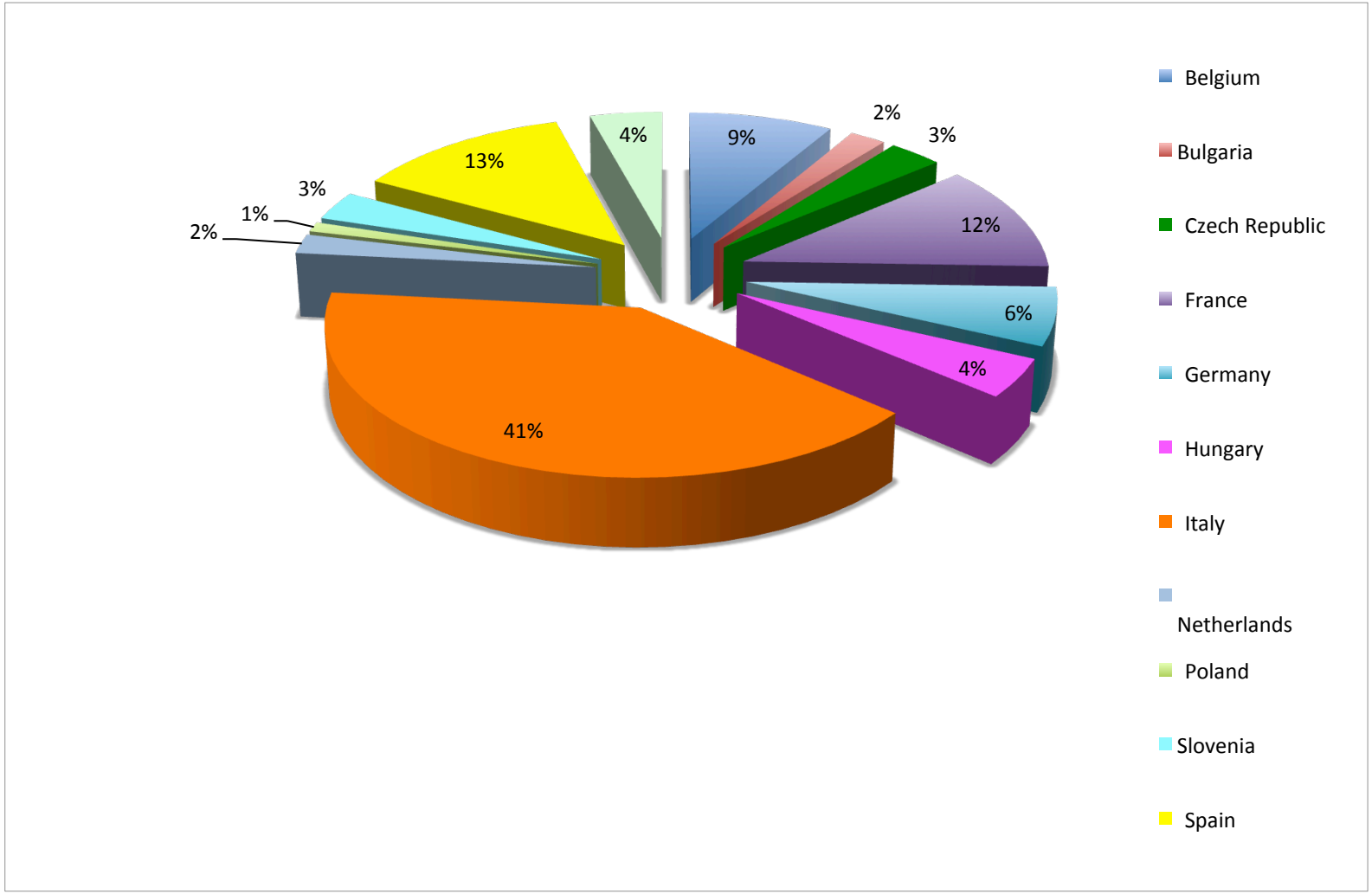
Alessandra Ferlini (Ferrara, Italy) - chair
Serenella Servidei – (Rome, Policlinico Gemelli, Rome, Italy)
Silvere van Der Maarel - (Leiden, LUMC, The Netherlands)
Borut Peterlin - (Ljubljana, Slovenia)

Patient Advisory Board (PAB)

Francoise Rouault, AFM
Martin Bobrow, MDUK

EU CENTERS/HCPs who manifested interest

A TOTAL OF 97 MEMBERS FROM:



TCONs and MEETINGS

- TC 3° of July
- TC 16° of August
- Meeting at WMS, St Malo Oct. 2017 (Ferlini, Maarel, Servidei)
- Meeting at SIGU, Neaple Nov. 2017 (Ferlini, Peterlin)
- Meeting in Freiburg (Ferlini, Servidei, Peterlin)

MAIN ISSUES

- Diagnostic genetic testing needs within the ERN
- Revising genetic diagnostic guidelines, educational aspects, phenomics
- Stakeholders interaction: RD-Connect, ESHG, Eurogentest, EMQN, EAN and other EU ERN networks
- NGS methodologies and applications into clinical practice: SURVEY
- SOLVE-RD EU PROJECT “solve the unsolved” collaboration and interaction

GOALS

- Survey and Questionnaire to list diseases currently genetically diagnosed within the EURO-NMD HCPs
- Strong interaction with **RD-Connect** for data sharing and NGS data databases
- Reviewing Genetic Guidelines
- Drafting non existing genetic guidelines
- Facing the bottleneck of CNVs search in KNOWN GENES in NMDs (negative at panels, WES or with incomplete genotype definition)
- Funds scouting
- Telegenetics

ONGOING ACTIONS

- **Questionnaire**

Survey within the ERN about the NMD diagnostic tools
-gene panels
-WES
-WGS

- NMDs covered
- Number of genes covered
- Techniques
- Platforms

GOALS
MAPPING THE NOT DIAGNOSED NMDs

- The questionnaire −first draft

ONGOING ACTIONS

- **GUIDELINES**

- CONTACTS WITH:
- EMQN, EQA, PT (Simon Patton)
- SOLVE-RD, other ERNs (Holm Graessner)
- ESHG (Olaf Riess)

  - Review and eventual update of the existing genetic Guidelines

  - IN PROGRESS

ONGOING ACTIONS

- **Funding opportunity for networking**
- Importance of get funding to have meetings also expanding the attendees across EU and also beyond

- **ENMC workshop application** (deadline March 2018, meeting at the end of 2018), with the main focus on NMD genetic diagnosis
- Provisional title: **EU Roadmap of diagnostic needs for neuromuscular patients: what we have and what we don't. Briefly:** what is available in EU and what is not (genetic orphans); orphan genes in NMD in terms of lacking genetic testing.

ONGOING ACTIONS

- **Funding opportunity for networking**

- Importance of get funding to have meetings also expanding the attendees across EU and also beyond

- **EU COST ACTION** on NMDs GENETIC TESTING
  - dedline expected in February 2018
  - TRANSVERSAL TASK, OTHER ERNs will be invited to participate and be involved

SPECIFIC ACTIONS

- **Extensive CNVs search in <u>KNOWN GENES</u> for NMDs**

- **INDUSTRIES COLLABORATION**
  - **Evaluate co-development of easy-to-use diagnostic kits to identify CNVs in the routine lab activity**

- Preliminary discussion already started with Multiplicom and MRC Holland
- Collaboration to develop new “AD-HOC-NMD” diagnostic tools
- Other Industries will be contacted.

- **ENMC** WS excellent opportunity also to have Industries invited and prime collaborations

SPECIFIC ACTIONS

- **Telegenetics**

Transversal task

Country-based Initiatives already ongoing in Italy and in the Netherland

- Just start talking

Thanks

- PLEASE HAVE YOUR QUESTIONS