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:
TCNs 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 getting 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
  - deadline expected in February 2018

  - TRANSVERSAL TASK, OTHER ERNs will be invited to participate and be involved
SPECIFIC ACTIONS

- Extensive CNVs search in **KNOWN GENES** 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