7th ERN EURO-NMD

ANNUAL MEETING

EMQN Scheme

21 February 2024

EURO-NMD activities

Carla D'ANGELO

European Reference Network
for rare or low prevalence complex diseases

Network Neuromuscular Diseases (ERN EURO-NMD)

Funded by the European Union

The ERN EURO-NMD is funded by the European Commission under the EU4Health programme (EURO-NMD 23-27 — 101156434 — EU4H-2023-ERN2-IBA)

EMQN Pilot EQA scheme NGS-based NMD diagnostics

- External Quality Assessment scheme of NGS-based testing for NMDs
  - By comparing testing procedures and consistency of results, we aim to identify areas needing improvement & harmonise practices across the ERN
- WHY THIS IS IMPORTANT?
  - Clinical and genetic heterogeneity: >700 NMD disease genes *(PMID: 34930546)*
  - Genetically heterogeneous conditions: 39 genes identified to cause LGMD
  - EQA is currently offered for some NMD genes as single gene test (DMD, SMA, CMT, and few others), but no dedicated schemes are currently offered for panel testing of NMD genes

EMQN: European Molecular Genetics Quality Network
EQA: External Quality Assessment

EMQN Pilot EQA scheme NGS-based NMD diagnostics

- [2020 survey] Genetic testing offer for inherited neuromuscular diseases within the EURO-NMD reference network: A European survey study (PMID: 32946487)
  - NGS is used by 94% of centres (55 HCPs)
  - 60% offer NGS for all patients that fulfil criteria for NMD of genetic origin

Oct. 2022
Negotiations with EMQN started

Nov. 2022
Letter of intent & collaboration agreement

Apr. 2023
New scheme proposal sent

- Gene selection has been made based on 3 main criteria:
  - genes which pathogenic variations are causing similar phenotypes
  - genes which pathogenic variations cause frequent diseases (more than 1/10.000)
  - genes which pathogenic variations appear most frequently as SNVs
  - genes which pathogenic variation cause hereditary muscle disorders

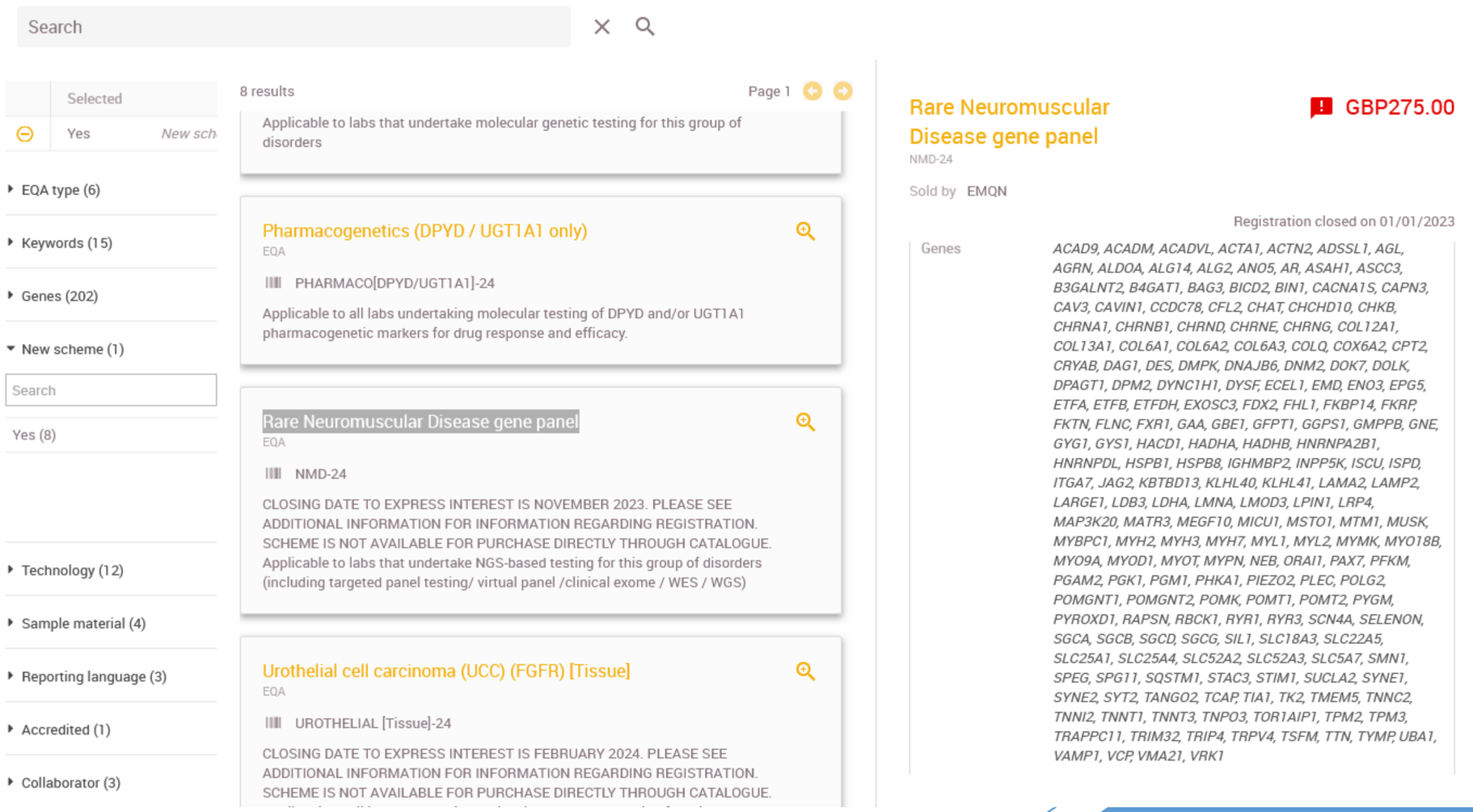
EMQN Pilot EQA scheme NGS-based NMD diagnostics

June 2023
formal approval by EMQN

July-Sept 2023
EOI from genetic labs in the EURO-NMD HCPs

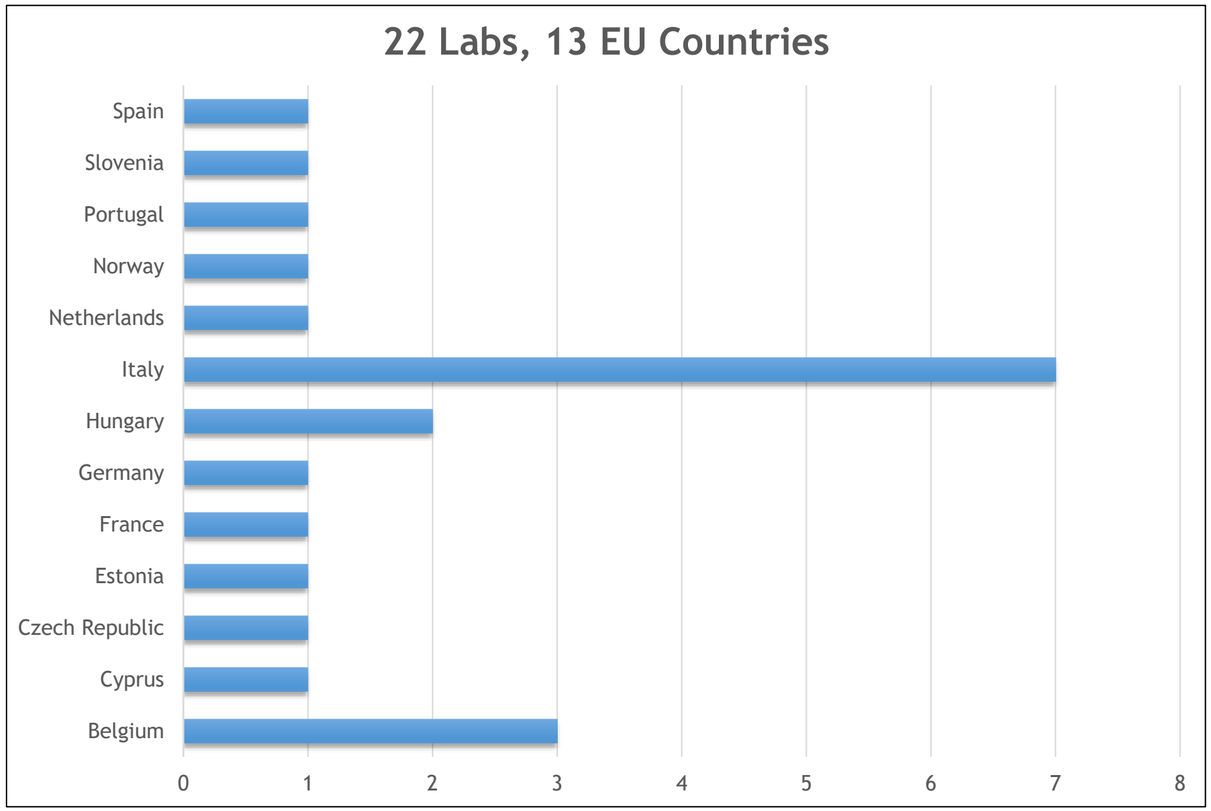
- Scheme was opened to 30 EURO-NMD HCPs:
  - performing NGS-based tests for NMD diagnosis (including targeted panel testing/virtual panel/clinical exome/WES/WGS)
  - using any appropriate gene panel for NMDs
  - performing both wet and dry NGS-based tests (no tertiary labs accepted)

EQA scheme catalogue 2024

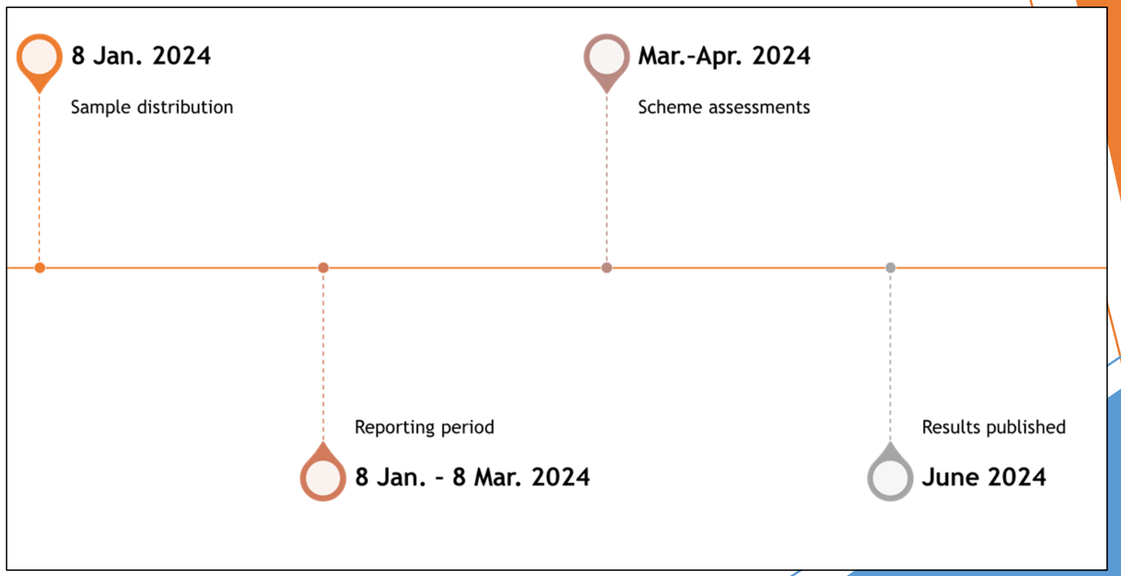


EMQN Pilot EQA scheme NGS-based NMD diagnostics

Participating Labs



Scheme timelines



Thank you !

Scheme organizers

ERN EURO-NMD

- Alessandra Ferlini (Ferrara)
- Vincenzo Nigro (Naples University),
- Rosario Santos (University Hospital of Porto)

EMQN

- Simon Patton
- Nicola Wolstenholme
- Rachel Taylor

- If you are interested, please contact: c.dangelo@ern-euro-nmd.eu

EURO-NMD

NEWSLETTER

Scan to subscribe !

The ERN EURO-NMD is funded by the European Commission under the EU4Health programme (EURO-NMD 23-27 — 101156434 — EU4H-2023-ERN2-IBA)

Funded by the European Union