

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



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 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

✕ 🔍

	Selected	
⊖	Yes	<i>New sch</i>

8 results

Page 1 ⏪ ⏩

- ▶ EQA type (6)
- ▶ Keywords (15)
- ▶ Genes (202)
- ▼ New scheme (1)

- Yes (8)

- ▶ Technology (12)
- ▶ Sample material (4)
- ▶ Reporting language (3)
- ▶ Accredited (1)
- ▶ Collaborator (3)

Applicable to labs that undertake molecular genetic testing for this group of disorders

Pharmacogenetics (DPYD / UGT1A1 only) 🔍

EQA

▮▮▮ PHARMACO[DPYD/UGT1A1]-24

Applicable to all labs undertaking molecular testing of DPYD and/or UGT1A1 pharmacogenetic markers for drug response and efficacy.

Rare Neuromuscular Disease gene panel 🔍

EQA

▮▮▮ NMD-24

CLOSING DATE TO EXPRESS INTEREST IS NOVEMBER 2023. PLEASE SEE ADDITIONAL INFORMATION FOR INFORMATION REGARDING REGISTRATION. SCHEME IS NOT AVAILABLE FOR PURCHASE DIRECTLY THROUGH CATALOGUE.

Applicable to labs that undertake NGS-based testing for this group of disorders (including targeted panel testing/ virtual panel /clinical exome / WES / WGS)

Urothelial cell carcinoma (UCC) (FGFR) [Tissue] 🔍

EQA

▮▮▮ UROTHELIAL [Tissue]-24

CLOSING DATE TO EXPRESS INTEREST IS FEBRUARY 2024. PLEASE SEE ADDITIONAL INFORMATION FOR INFORMATION REGARDING REGISTRATION. SCHEME IS NOT AVAILABLE FOR PURCHASE DIRECTLY THROUGH CATALOGUE.

Rare Neuromuscular Disease gene panel

! **GBP275.00**

NMD-24

Sold by EMQN

Registration closed on 01/01/2023

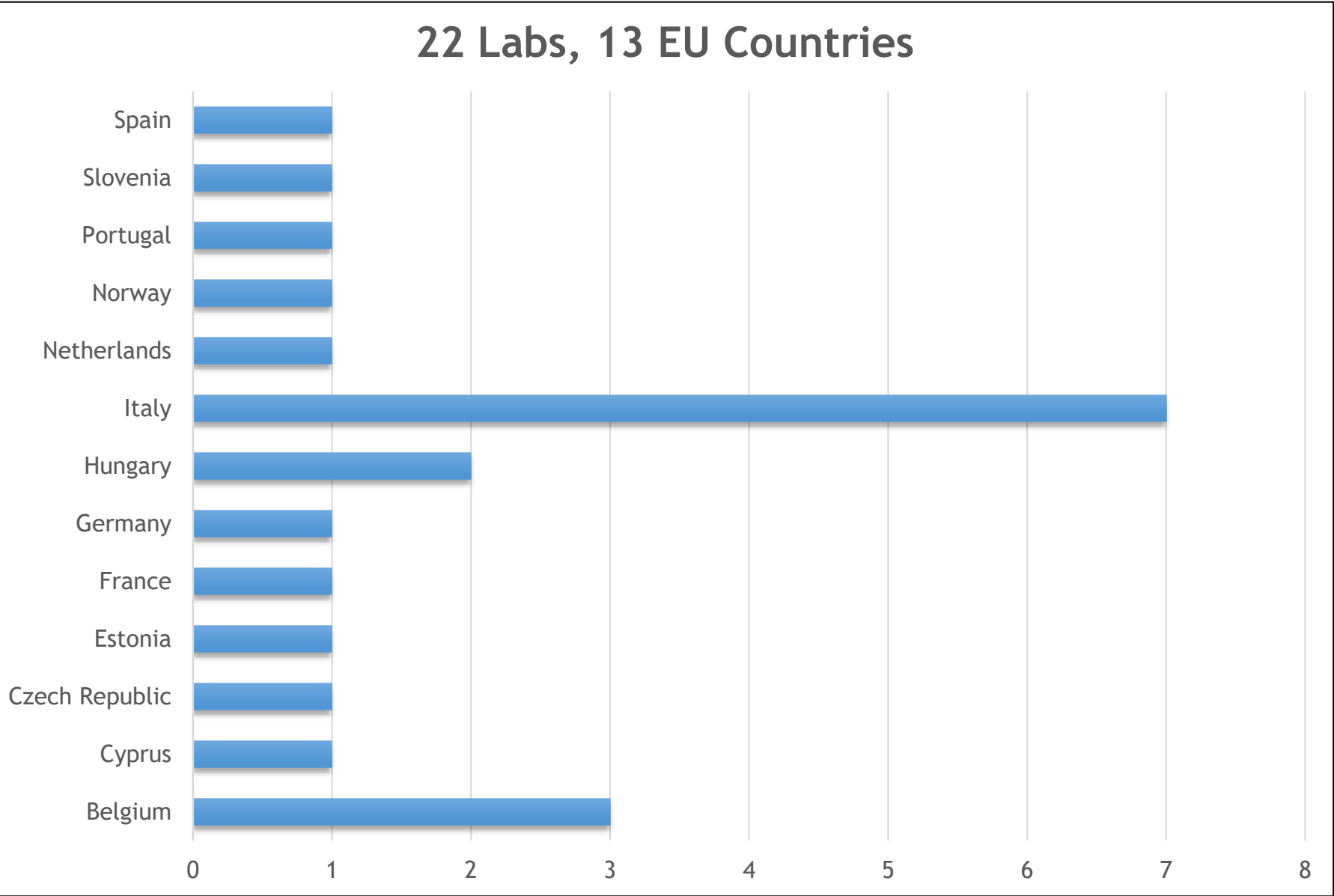
Genes

ACAD9, ACADM, ACADVL, ACTA1, ACTN2, ADSSL1, AGL, AGRN, ALDOA, ALG14, ALG2, ANO5, AR, ASAH1, ASCC3, B3GALNT2, B4GAT1, BAG3, BICD2, BIN1, CACNA1S, CAPN3, CAV3, CAVIN1, CCDC78, CFL2, CHAT, CHCHD10, CHKB, CHRNA1, CHRN1, CHRN2, CHRNE, CHRNG, COL12A1, COL13A1, COL6A1, COL6A2, COL6A3, COLQ, COX6A2, CPT2, CRYAB, DAG1, DES, DMPK, DNAJB6, DNM2, DOK7, DOLK, DPAGT1, DPM2, DYNC1H1, DYSF, ECEL1, EMD, ENO3, EPG5, ETFA, ETFB, ETFDH, EXOSC3, FDX2, FHL1, FKBP14, FKBP, FKTN, FLNC, FXR1, GAA, GBE1, GFPT1, GGPS1, GMPPB, GNE, GYG1, GYS1, HACD1, HADHA, HADHB, HNRNPA2B1, HNRNPDL, HSPB1, HSPB8, IGHMBP2, INPP5K, ISCU, ISPD, ITGA7, JAG2, KBTBD13, KLHL40, KLHL41, LAMA2, LAMP2, LARGE1, LDB3, LDHA, LMNA, LMOD3, LPIN1, LRP4, MAP3K20, MATR3, MEGF10, MICU1, MSTO1, MTM1, MUSK, MYBPC1, MYH2, MYH3, MYH7, MYL1, MYL2, MYMK, MYO18B, MYO9A, MYO10, MYO11, MYO12, MYO13, MYO14, MYO15, MYO16, MYO17, MYO18, MYO19, MYO20, MYO21, MYO22, MYO23, MYO24, MYO25, MYO26, MYO27, MYO28, MYO29, MYO30, MYO31, MYO32, MYO33, MYO34, MYO35, 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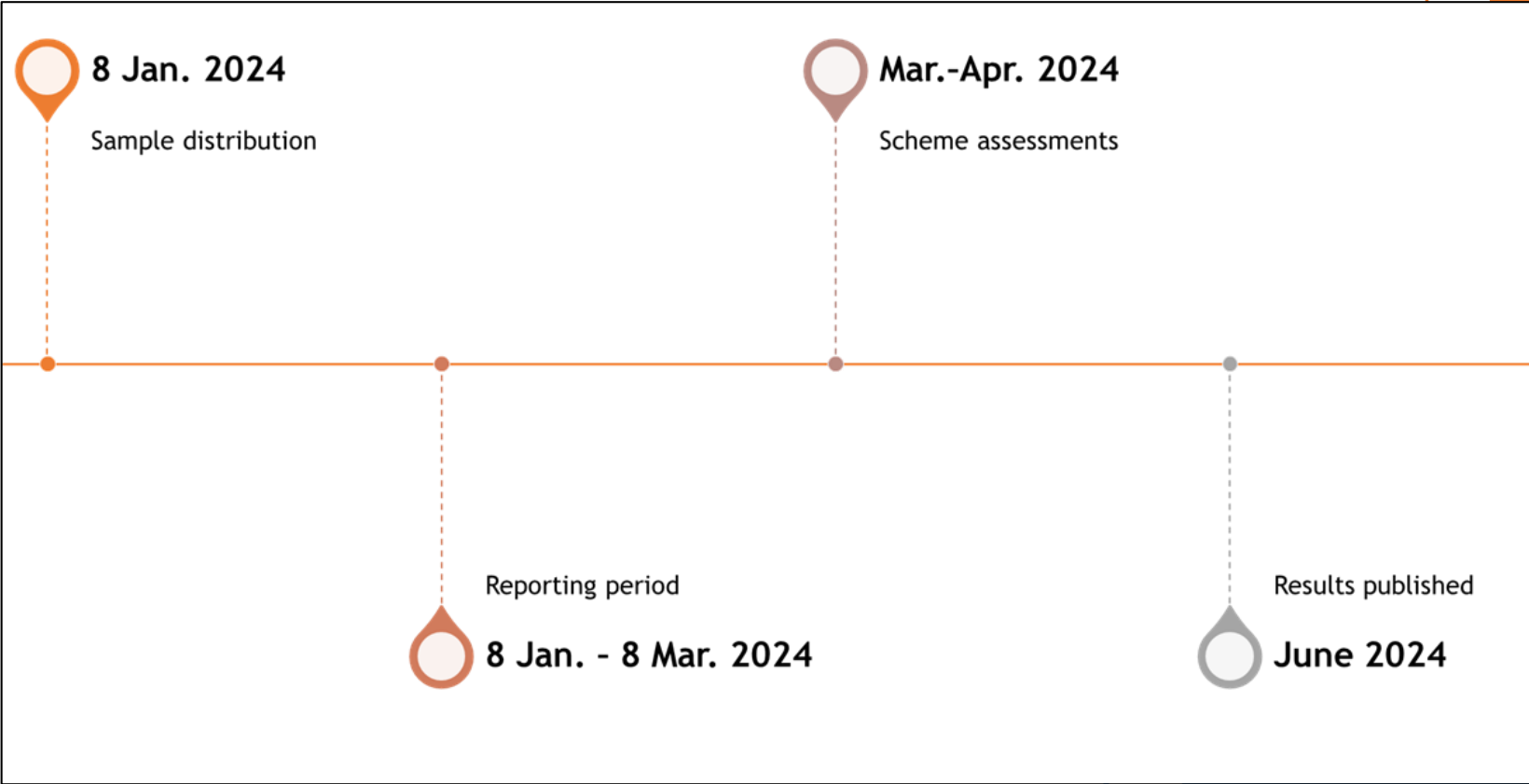
EMQN Pilot EQA scheme NGS-based NMD diagnostics

Participating Labs

22 Labs, 13 EU Countries



Scheme timelines



Thank you !

EURO-NMD NEWSLETTER

Scheme organizers

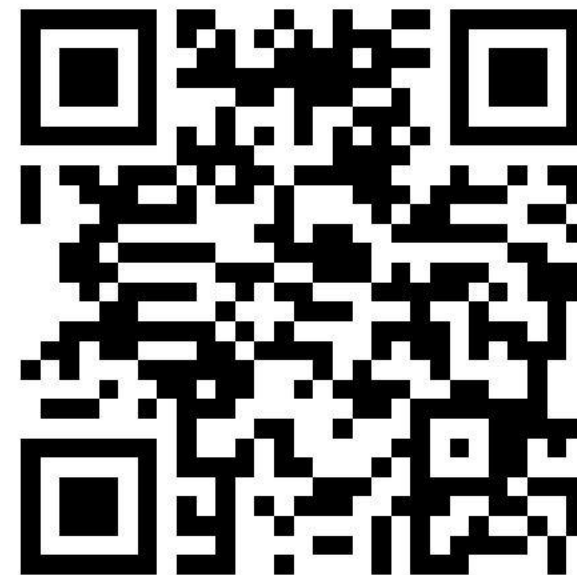
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