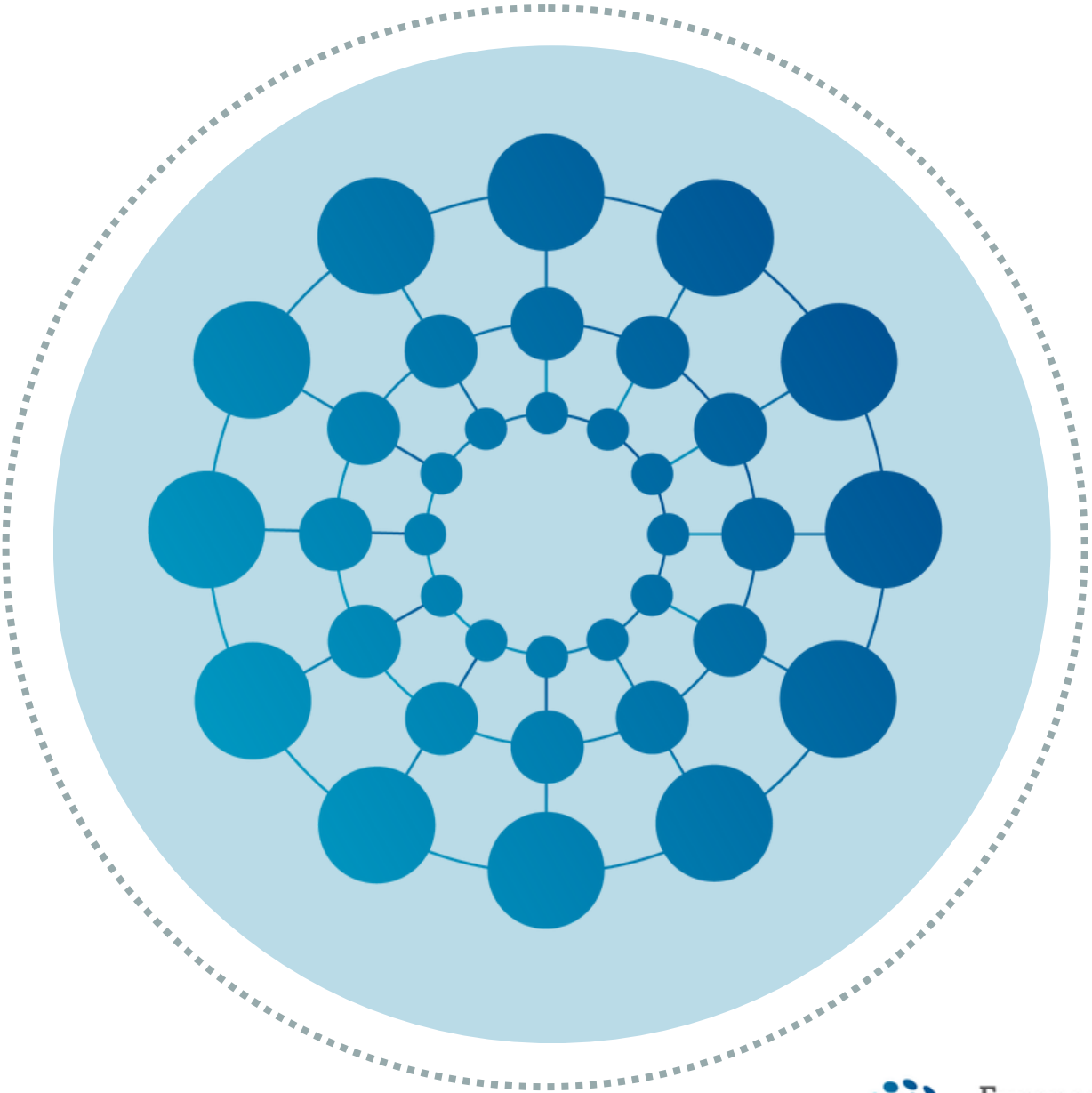
EURO-NMD Registry Hub

European Reference Network for neuromuscular diseases

INFORMATION LETTER



European Reference Network
for rare or low prevalence complex diseases

Network Neuromuscular Diseases (ERN EURO-NMD)

1 INTRODUCTION

The EURO-NMD registry is the first European Registry that encompasses all rare neuromuscular disorders (NMDs). It is a web-based, interoperable, multi-centre patient registry established by the EURO-NMD Registry Consortium and affiliated to the European Reference Network for patients with rare neuromuscular diseases (ERN EURO-NMD).

1.1 Aims

The ERN EURO-NMD involves 82 healthcare providers (HCPs) from 24 European Union Members countries and Norway, which are recognised as centres of expertise for rare NMDs and seeing more than 100,000 NMD patients annually. While the HCPs in the ERN are collecting valuable data in several registries, there is currently no unified NMD or NMD disease-specific registry in use by all of them.

The EURO-NMD registry will improve care delivery and address the fragmentation of NMD data, reducing differences in clinical practice across Europe. Simultaneously, it will provide sufficient data to initiate research and clinical trials and to inform policy and regulatory decisions. Finally, the registry is configured for federated data sharing with other registries, contributing to accruing the rare diseases data's critical mass.

1.2 Registry design

The EURO-NMD Registry was designed to collect longitudinal data on neuromuscular patients seen at EURO-NMD's 82 HCPs as part of routine care.

The Registry is built on REDCap, a secure online relational database system hosted in Freiburg and designed as a central database. Each participating centre has access only to data from its participants.

The Registry is part of the EURO-NMD Registry Hub, a federated network of FAIR-compliant connected data sources. This allows for joint analysis with data stored in multiple FAIR sources to answer research questions without sharing patient-level data via so-called "federated queries".

The data collected for the Registry will not entail additional clinical procedures or appointments by the HCPs beyond those currently provided.

1.3 Patient inclusion criteria

Adult and paediatric patients diagnosed with or suspected of having a neuromuscular disease who are being treated or followed up by HCPs who are full members and affiliate partners of the ERN EURO-NMD.

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1.4 Data to be collected

Mandatory data elements include the "Set of common data elements for Rare Disease Registration" (CDEs) developed by the European Commission (https://eu-rd-platform.jrc.ec.europa.eu/set-of-common-data-elements). The set consists of 16 data elements that are considered to be essential for research. All ERNs are obliged to collect these elements.

In addition to these core data elements, the Registry holds a set of common data elements for all neuromuscular diseases collectively agreed by the EURO-NMD experts, as well as disease-specific data elements for the different thematic disease groups covered by the ERN EURO-NMD: Neuropathies, Myopathies, Mitochondrial Diseases, Neuromuscular Junction Disorders, and Motor Neuron Diseases). Together, these data enable the calculation of disease-specific Key Performance Indicators (KPIs).

1.5 Data Collection Process

Patients or their representatives have to sign an informed consent before data is entered in the Registry. To this end, patient information and consent form in one of the EU languages is provided to participating sites. Participating sites are responsible for ensuring that the content of the informed consent meets national and local requirements.

Data is pseudonymised at the HCP level so that no identifying data is entered in the Registry.

1.5.1 Possible scenarios for the provision of data

A) Centralised approach: data submitted to EURO-NMD Registry

- Manual or bulk data entry directly into the REDCap-system hosted by University Medical Center Freiburg. Data can be extracted from local systems into csv files for import into the ERN Registry (no additional installations required at data providers).

B) Decentralised approach by connecting your local system to the EURO-NMD Registry Hub

- Data exchange with the EURO-NMD Registry by adopting the FAIR principles. This requires the FAIRification of existing registries including harmonisation of existing data, and the installation of additional software to host the FAIR data and metadata.

Please note: if you already have structured data collection at your HCP, either of these approaches enable the possibility of avoiding duplicate data entry. We are happy to work with you to find the most appropriate solution for your needs.

1.6 Security and Privacy

The Registry complies with EU data protection legislation (GDPR). It incorporates a range of technical mechanisms to ensure information security and data quality and to respect patient's rights. Patients participating in the registry consent to using their data in specific ways (care, research, commercial

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queries) and can withdraw their consent at any time, in which case the data is removed from the Registry. Personal data is pseudonymized and kept in a separated resource, in this case, the clinical file at his HCP. Therefore, the local investigator is able to identify the patient if necessary, for example if the patient wants to withdraw his data.

To ensure that our processes are well defined and do not leave any security risks, the Registry Coordinator has carried out a Data Processing Impact Assessment (DPIA) in accordance with the principles set out in Article 35 of the GDPR.

1.7 Ethics

APHP is the coordinator of the EURO-NMD Registry as well as a data provider. The Registry is hosted by the University Medical Centre Freiburg (UKLFR), which is also a data provider. Each data provider (HCPs participating in the Registry) is a joint controller concerning the processing of personal data. HCPs are required to sign the data-sharing agreement with the Registry Consortium, which defines the roles and responsibilities of the parties involved. HCPs are also responsible for obtaining and maintaining appropriate local ethical/governance approval *(e.g. from IRB or similar)* before accessing and enrolling patients into the EURO-NMD Registry.

1.8 Registry roll out

The onboarding of HCPs into the Registry will be done in stages, starting with a subset of pilot sites, who we will work closely with to ensure that the onboarding processes and tools are robust and well-documented. The roll-out will then be opened to the rest of the network.

A Registry Helpdesk will provide technical support for the data collection and the installation of data integration solutions (e.g. data transfer from an existing database or registry, etc.).

Each Full Member and Affiliated Partner HCP will be offered a financial compensation of €1,500 for the first 100 patients enrolled in the Registry.

1.8.1 How to start

Your site can start registration in the EURO-NMD Registry when the legally responsible person in your organisation hierarchy has signed the data sharing agreement allowing you to participate with data.

If a centre would like to be part of the pilot phase and help us to ensure the success of the Registry roll-out, it should get in touch with the registry helpdesk at registryhelpdesk.euronmd@outlook.com

1.9 Registry Governance

The Registry is under the oversight of a Steering Committee (SC) with representatives from the EURO-NMD Registry consortium partner organisations, including:

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> Assistance Publique–Hôpitaux de Paris (AP-HP)
> University Medical Centre Freiburg (UKLFR)
> Radboud University Medical Center (Radboudumc)
> World Duchenne Organization (WDO)
> Duchenne Data Foundation (DDF)
> Institute of Myology (AIM)
> French Muscular Dystrophy Association (AFM-Téléthon).

The SC is chaired by the EURO-NMD Coordinator, who is also the Registry Coordinator (Dr Teresinha Evangelista, AP-HP), and supported by a management team based at the ERN EURO-NMD Coordination Office in Paris and by the IT Operations Team based in Freiburg.

1.10 Registry funding

The EURO-NMD Registry setup was co-funded by the 3rd EU Health Programme under Grant Agreement No 947598. The Registry has also received funding from the EU4health program (grant agreement 101085084) and will continue to be funded for the 2023-2027 period under the call EU4H-2023-ERN-IBA.

2 WHAT IS THE IMPACT OF THE EURO-NMD REGISTRY?

The EURO-NMD Registry will:

- Enable research opportunities from translational to epidemiological studies to clinical trials, and will accelerate the generation of new, more accurate knowledge about rare NMDs.
- Provide insights into the natural history of the individual NMDs.
- Help describe care pathways for patients with NMDs.
- Help understand and evaluate standard-of-care practices at European, National, and HCP levels.
- Provide new evidence base to implement improvements in NMD care across countries and HCPs.
- Increase research accessibility for patients by identifying populations eligible to participate in clinical trials or other research opportunities through the registry.

3 BENEFITS OF PARTICIPATING

Through the EURO-NMD Registry, HCPs can:

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- Store patient data, produce reports summarising patient data, and download data for local analyses or to support presentations and publications.
- Access live summary dashboards of their own data at any time for regular monitoring and performance review, as well as written KPI reports that include benchmarked comparisons against aggregate data to identify strengths and areas for improvement.
- Access aggregated or pseudonymised data to address their research questions or recruit participants for studies.
- Publish data from their HCP at any time, including comparisons of local HCP data with Registry published data, and participate in Registry-wide publications where HCP data have been used.

4 PREREQUISITES FOR PARTICIPATION

To participate in the EURO-NMD Registry, HCPs must sign the data-sharing agreement with the EURO-NMD Registry Consortium and obtain any site-specific ethical and governance approvals for the provision of data to the EURO-NMD Registry.

To ensure the success of the Registry, HCPs should:

- inform eligible patients about the Registry.
- obtain informed patient consent before entering data into the Registry.
- actively participate in the registry by enrolling patients admitted to the HCP.
- complete follow-up questionnaires annually after the baseline visit.
- collect at a minimum the mandatory set of data variables that have been chosen in cooperation with the EU and are known as the Common Data Elements (CDEs).
- assist with data quality assurance processes.
- adhere to the EURO-NMD Registry Publication Policy
- ensure adequate resources, including named staff with specific responsibility for the EURO-NMD Registry at your site, and a contact person who will be responsible for all registry-related communications with the Registry support team.

Once your HCP has obtained the necessary approvals to participate in the Registry, HCP staff will receive training (online or in person) in the use of the tool, and will be provided with the necessary manuals, documentation and follow-up to support their learning.

If you have any queries, please contact the Registry team on
registryhelpdesk.euronmd@outlook.com

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