EURO-NMD Registry Hub
European Reference Network for neuromuscular diseases

EURO-NMD Registry Hub

947598 - EURO-NMD Registry

D2.4 / MD.2 Mandatory Deliverable

Layman version of the final report

Lead contributor	UPPMD
Other contributors	AP-HP, UKLFR, RUMC, DDF, AIM, AFM, RUMC

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Abstract: MD.2 Mandatory Deliverable - Layman version of the final report. Deliverable D2.4 of the EURO-NMD Registry Hub project involves creating a Layman Version of the Final Report. This is done to ensure accessibility and understanding for patients and families living with rare neuromuscular conditions. Led by UPPMD and drawing on the comprehensive D1.04 Final report Y3: Contractual progress report, this task aims to present a concise summary of the project's outcomes and progress, without using too much technical jargon.

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History

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V0.1	13-06-2023	Draft
V0.2	25-06-2023	Final draft for Steering Committee
V0.2	28-06-2023	Consolidating comments, finalising document
V1	29-06-2023	Ready for submission in EU portal

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1. Introduction

1.1 About EURO-NMD Registry Hub

The EURO-NMD Registry Hub is a project to create a centralised and standardised database containing clinical and genetic data for patients living with rare neuromuscular diseases in Europe, as well as patient reported outcomes. It involves healthcare providers, patient organisations, and experts to share and collect important data. The main goal is to improve patient care and research by providing a centralised entry point for large amounts of information. The registry allows better collaboration and helps identify common patterns for improved diagnoses and treatments.

The project uses advanced technology to ensure data is accessible, secure, and follows international standards. Patients and their families are actively involved to address their needs and concerns. It's a groundbreaking effort to use data and teamwork to advance research and care for rare neuromuscular conditions in Europe.

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1.2 About this deliverable

Deliverable D2.4 of the EURO-NMD Registry Hub project creates a Layman Version of the Final Report. This report ensures that patients and families with rare neuromuscular conditions can understand the project's achievements and progress. Led by UPPMD and using the D1.04 Final report as a foundation, this task aims to present a clear summary without using complex language.

The Layman Version is easy to understand and accessible to a wider audience. It highlights the project's impact, challenges, and successes, allowing all stakeholders to grasp the significant work accomplished within the EURO-NMD Registry Hub.

The report keeps the rare neuromuscular community informed about progress and contributions made to improve the lives of those affected by these conditions. It demonstrates the project's commitment to research, care, and awareness for rare neuromuscular disorders, fostering engagement from all involved stakeholders.

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2. Summary per Work Package

2.1 Coordination and Management

As part of Work Package 1, APHP effectively led the coordination and management of the EURO-NMD Registry Hub project. The project implementation went according to plan and the main tasks due to the reporting period were completed. The EURO-NMD project manager, along with the Project Coordinator and the Steering Committee, have ensured smooth governance of the consortium. Key policy documents to facilitate data sharing, including data sharing agreements and consent forms, have been created and validated, and are expected to be finalised soon.

Most deliverables and milestones scheduled during the period have been successfully achieved or submitted to the European Commission (EC) as planned, with minor delays documented. The final report submission was delayed by one month. Additionally, complications in preparing and executing contractual agreements for the transfer of data to the registry prevented the enrollment of the first patient in a timely manner.

Task 1.2 focused on project operational management and monitoring, with APHP's diligent efforts to oversee the project's progress. Regular communication and support to partners were facilitated through emails, conference calls, and teleconferences with the Steering Committee. Notably, the in-person Steering Committee meeting in Porto allowed for fruitful discussions on various project aspects, followed by a training workshop for ERN HCPs.

APHP also took the lead in Task 1.3, focusing on Intellectual Property (IP) & Knowledge Management. A working group actively discussed guidelines for collaboration with other registries and ways to protect intellectual property and confidential information exchange.

Regarding sustainability (Task 1.4), the EURO-NMD Registry's funding was secured through current and upcoming grants, ensuring its operations beyond the project's end. In-kind support from consortium partners and the establishment of the registry's responsibility under the EURO-NMD Registry Consortium further contribute to its long-term viability.

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2.2 Dissemination of the project

Work Package 2 of the EURO-NMD Registry Hub project focused on disseminating project information to various stakeholders. Task 2.1 ensured effective communication with consortium partners through teleconferences, emails, and periodic meetings, keeping them informed about project achievements and progress. Task 2.2 involved communication with the IT developer, UKLFR, coordinating the implementation of new features and change requests for the EURO-NMD register. UKLFR also engaged with other organisations to share knowledge and contribute to the development of the registry.

In Task 2.3, the project engaged with Healthcare Providers (HCPs) mainly through the EURO-NMD website, social media channels, newsletters, and regular meetings of the Executive Committee and Board of the Network. Hands-on training workshops were also conducted during the EURO-NMD Annual Meeting to familiarise representatives from participating HCPs with the registry system and gather feedback for improvements.

Task 2.4, led by UPPMD and supported by all partners, focused on engaging with Patient Organizations (POs) and other stakeholders. Dissemination efforts targeted patient communities, families, and relevant organisations, using websites, social media, newsletters, and presentations during meetings and events. Engagement with pharmaceutical companies demonstrated the project's credibility and potential for impactful collaborations.

Overall, Work Package 2 successfully disseminated project updates, fostered collaborations, and raised awareness about the EURO-NMD Registry Hub's potential to benefit the neuromuscular and rare disease community. The engagement with various stakeholders reaffirms the project's commitment to making a positive impact in the field of neuromuscular disorders.

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2.3 Evaluation of the project's output and impact

In Work Package 3, the EURO-NMD Registry Hub project focused on evaluating its output and impact, with the establishment of the Independent Advisory Committee (IAC) taking centre stage. Led by UPPMD, the IAC comprised various experts in data management, registries, ethics, regulatory affairs, and policy.

Over the project's duration, the IAC held three meetings to offer valuable input and expertise. The meetings covered various aspects, including the scope of work packages, updates on progress, demonstrations of the registry setup, and discussions on Key Performance Metrics (KPMs) for quality and functionality. The IAC played a crucial role in evaluating the registry concept, monitoring its quality and functionality, and assessing the project's impact on clinical and research activities.

The IAC’s recommendations contributed to health policy and legislative advancements, supporting the registry's sustainable development and implementation within the Rare Neuromuscular European Reference Network and beyond. The IAC's involvement ensured that the project benefited from external insights, contributing to the overall success and effectiveness of the EURO-NMD Registry Hub.

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2.4 Design and development of the registry

In Work Package 4, the focus was on designing and developing the EURO-NMD Registry to ensure user-friendly access and functionality. Task 4.4 involved creating manuals, FAQs, and additional materials to support users. A quick-start guide was developed, providing instructions on accessing the registry, important navigation items, and how to add and edit patient records. Data import templates were also provided for bulk-upload of essential data for multiple patients simultaneously. Plans were made to expand training materials with videos and offer them in multiple languages. Additionally, a training environment was established for users to try out all available functions without affecting real data, making it possible to replicate the registry at separate locations.

Task 4.5 focused on testing and refining the software and IT systems. A first version of the registry was created and evaluated by real data-entry users from various healthcare providers (HCPs). Feedback was collected through structured forms and discussions, resulting in improvements to usability and database content. A hands-on training session during the EURO-NMD Annual Meeting gathered further feedback and feature requests, which were thoughtfully addressed and implemented. An anonymous user-experience feedback survey garnered positive responses, with improvements made based on the collected feedback. These efforts ensured the registry's usability and effectiveness, making it a valuable resource for healthcare professionals and researchers involved in the EURO-NMD project. Direct patient input in the registry is still under development but foreseen in the near future.

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2.5 Interoperability and FAIRification

In Work Package 5, the focus was on achieving interoperability and FAIRification of the EURO-NMD Registry, enabling seamless data exchange and accessibility.

An important milestone was achieved under this work package where a proof-of-concept demonstrated how five different rare disease registries for neuromuscular disorders can be jointly queried because they are interoperable. This milestone was a joint effort from a number of partners building the registry hub for EURO-NMD. Because the registry hub connects different FAIR registries, it is now possible to conduct the same queries related to neuromuscular diseases in multiple, independent registries simultaneously without exposing sensitive patient details. The registries that participated in this proof-of-concept with mock data are: CRAMP (Computer Registry of All Myopathies and Polyneuropathies, Netherlands), DDP (Duchenne Data Platform, patient-led registry for Duchenne Muscular Dystrophy, Netherlands), DM-Scope (National registry for Myotonic Dystrophies, France), SMArtCARE (Clinical registry for Spinal Muscular Atrophy, Germany), and the central European Reference Network (EURO-NMD) registry. In order to support others in their FAIRification efforts, this demonstration prototype was made publicly accessible.

FAIR stands for Findable, Accessible, Interoperable, and Reusable. It is the acronym used to describe a global initiative to make data more valuable by increasing the ability of computers to find, interpret, integrate, and analyse those data autonomously. In this prototype, the computer calls a special Web address where specified and pre-approved queries (e.g. patient-count or phenotypic observations) are executed inside the FAIR registry's secure space, and anonymized data are given back to the computer as output. The prototype depends on a publicly available database of queries that were manually curated and filtered by experts in FAIR and neuromuscular diseases. FAIR makes it possible for the same query to be executed over independent resources, and thus sharing those queries leads to convergence between registries.

RUMC, Freiburg and other partners worked closely in developing for EURO-NMD this privacy-preserving federation over multiple, independent and FAIR registries.

The successful demonstration prototype and proof-of-concepts served as best practices and are now implemented in other European Reference Networks (ERNs) and the European Joint Program on Rare Diseases' virtual platform.

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2.6 Development of Quality and Outcome indicators

In Work Package 6, the focus was on developing quality and outcome indicators to assess care quality performance of rare neuromuscular disease centres within the EURO-NMD Registry.

Task 6.2, led by UKLFR in collaboration with RUMC, UPPMD, DDF, AIM, and AFM, involved the integration of key performance and outcome indicators (KPIs) into the registry.

A total of 83 KPIs were carefully selected by EURO-NMD experts through a consensus seeking process. These KPIs serve as essential metrics to measure and monitor the performance of the centres involved. While all data variables for measuring centre performance were incorporated into the data dictionary, only a limited set of KPIs have been implemented in the live dashboard-views within the REDCap database at this stage.

To provide more detailed and customised reports, an additional pipeline is being developed. This pipeline will export the required data via the REDCap API, prepare summary statistics and graphs in statistical software, and then upload the results back into the REDCap File Repository in a restricted folder for each participating centre. This will enable local investigators at the participating centres to view continuously updated KPI statistics for their patients and compare them with the overall patient cohort in the registry (KPI benchmarking).

The integration of these KPIs and the continuous monitoring of their statistics will help assess the quality of care and outcomes for patients with rare neuromuscular diseases, ensuring that the registry functions effectively as a valuable tool for improving patient care and research within the EURO-NMD project.

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