

**PAB POSITION LETTER**  
**CALL FOR PROPOSAL ON REGISTRIES DEDICATED TO ERN**

Patients' data registries in Europe in the neuromuscular field followed the spectacular progress of medicine over the past decades.

Due to the high number and heterogeneity of diseases, the variety of collected data types and the various national policies in the domain we observe today a multiplicity of initiatives drawing a highly uneven picture throughout the continent and among diseases.

The NMD Patient Community is one of the oldest, strongest and well structured communities in Rare Diseases. Patient owned registries are representing at the moment the biggest part of the NMD registries followed by Academic and HCP registries. All POs, HCPs and Academic registries at the moment operate in silos and none of them is FAIR and interconnected, crippling the research and development in NMDs.

While the last years new FAIR patient driven platforms are under development to tackle this issue on the main NMD diseases areas, a FAIR EURO NMD Registry Hub that promotes interoperability with a strong and joined patient and HCP governance could be a unique opportunity both for patients but also for HCP and Industry to promote patient centered care and research.

In the context of more precise diagnostic and the arrival of the first natural history modifying drugs the necessity of quantitative and qualitative data has never been as crucial as today. In these circumstances the call for proposal on registries dedicated to the ERN initiated by the European Commission got a positive feedback from the PAB.

The present letter intends to summarize patients' remarks, prerequisites, and visions on how the EURO-NMD network should positively respond to the call with the ambition for the EURO NMD Registry Hub to be a model for other RDs.

We think the EURO-NMD network should take a leading role on creating a registry for all diseases covered by the network.

This Registry Hub should be patient centered, able to communicate with the already existing disease specific registries and databases and should be designed in such a way that professionals and patients can add data in a way that requires a minimal effort. As agreed all the above will be supported with a joined and equal PO and HCPs access to data, and governance model.

Due to the cost in patient lives, no type of another - in silo- non patient centered, and poor governed registry would be supported by the NMD patient community, their Patient Organization and the PAB that would actively advocate against it.

### **Registry design**

The PAB agreed on several issues of importance regarding the design of the present registry.

As a minimum requirement, the registry must comply with European current practice:

- It must comply with EJP RD standards
- It must follow the FAIR principles.
- It must implement at least the JRC minimum core dataset

On the one hand, the registry must be able to interoperate with other registries and take a position of European Hub for neuromuscular condition registries. This will allow the registry to link with existing numerous - in silo - initiatives in the domain, promote their "FAIRifications", allow batch data entries and serve as the connection point of these registries, avoiding multiple data entry, duplications, fragmentation and actively promote data sharing.

On the other hand, the registry must provide with a conventional, manual, way of inputting the data. This is crucial so that no patient is excluded from the registry and in particular:

- Non diagnosed patients (patients that are suspected to have a neuromuscular disease, but do not have a specific diagnosis yet)
- Diagnosed patients with a condition for which there is no registry
- Patients that are not followed in reference centers
- Patients living in countries where there is no national registry

As a conclusion patients must be able to input themselves their own data which can be HCP validated if this is deemed necessary.

Individual data must be fully accessible and extractable for the patient in a machine readable format, according to GDPR, - respecting also the "Right to Forget" in case the patient desires to do so- and also accessible in an anonymized or pseudo anonymised format to all the stakeholders including patients and their organization, researchers, regulators and HCPs accompanied by a dynamic informed consent.

## **Software**

We encourage the EURO-NMD registry to be based on an already existing solution. This will drastically reduce the cost and the implementation time necessary.

The technical core platform could also be co-developed with other ERNs or reuse already existing ERN registry.

As we think that the project should be piloted by the current coordinating center of the ERN we find it equally important that a team of IT expert is in charge of the technical implementation of the registry. For efficacy reason this team could be made of professional that are familiar with medical system development and/or European Commission IT systems. The Patient Organizations and their Foundations can support with the technical experts in order to reduce the cost. The technical proposal should follow Open Access and Open Source solutions.

After the development of the registry specs and for transparency reasons, we should request at least three official quotes that will include the development and the 3 year maintenance cost.

## **Governance**

When comes the matter of governance it is important that patients' and clinicians' community are equally involved in the governance of the registry. According to EU Law the patient is the owner of his/her data and should be consulted when it comes to its use or reuse by other stakeholders.

In practice this means:

- That patients' and clinicians' weight is equal in the decisions made regarding the registry.
- Patients, Patients Organizations, Research Institutes, HCPs, Medical Organizations and Regulatory Authorities are considered equal partners when it comes to data access.
- A governance board should be installed consisting equally of representatives of both patients and clinicians.
- Industry is not allowed in the governance structure. Requests for anonymized data or reports from patients can be filed at the Registry Board.
- Patients will be approached by the Registry Board in a manner that respects their privacy (for instance they may be approached by the registry and respond if they like to participate in a specific research).
- The EURO NMD Registry Hub should develop with the support of the PAB a dynamic IC that will promote research while it protects patient preferences and privacy.
- The PAB and POs involved will initiate and support a Scientific Advice at the EMA for the EURO NMD Registry Hub. Following the discussion of the NMD patient community with the EMA and the Regulatory challenges in the development of medicines in the NMDs, the advice of the European Regulators in the design and the data collected is considered critical in order for the EURO NMD Registry Hub to be useful in the regulatory work and promote research and access to innovative therapies.

The EURreca registry (European Registries for Rare Endocrine Conditions) from Endo- ERN such balanced governance and could be considered as a model for the EURO-NMD registry.

## **Funding**

Funding of the registry must already be thought of in two aspects:

- The possibility to reach the ambition described in this document
- The sustainability of the registry on the long term

As a consequence complementary funding must be searched since the present call for proposal will not be enough.

In order for the EURO-NMD Registry Hub to be successful the EURO-NMD should have a strategic partnership with the Patient Organizations.

PAB and the Patient Organizations, will support such an endeavor by promoting awareness and recruitment, by supporting the interconnection of the existing patient registries that are already developed by the POs, but also by providing the essential technical and financial support that is needed.

With the Patient Organizations on board, the development of a second stage strategy could be developed where the involvement of other stakeholders like pharmaceutical companies would be designed and implemented to promote research and medicines development.

This strategy would be developed, potentially through a consortium, in which the governance would remain fully in the ERN hands.

## **Conclusions**

EUR-NMD PAB, the NMD Patient Community and the participating Patient Organizations are committed to support the development of a FAIR, Patient Centered EURO NMD Registry HUB that will be supported with a joined and equal PO and HCPs access to data, and governance model.

On behalf of PAB