

A registries hub against data siloing (ERN EURO-NMD registry)

H Pascal¹, A Tassoni², PAC 't Hoen³, H Lochmuller², J Kirschner²,
 G Paliouras⁴, E Vroom⁵, D Athanasiou⁴, SA Bakker⁴, F Lamy⁶,
 A Herman¹, G Bassez⁷, T Evangelista^{1,7}

ASSOCIATION INSTITUT DE MYOLOGIE, France¹; UNIVERSITÄTSKLINIKUM FREIBURG, Germany²; RADBOUDUMC, Netherlands³; STICHTING DUCHENNE DATA FOUNDATION, Netherlands⁴; WORLD DUCHENNE ORGANIZATION, Netherlands⁵; ASSOCIATION FRANCAISE CONTRE LES MYOPATHIES (AFM-Telethon)⁶, France; ASSISTANCE PUBLIQUE - HOPITAUX DE PARIS, France⁷

INTRODUCTION

Patient registries are key instruments for EURO-NMD to be able to deliver its objectives of care, research and education. EURO-NMD health care providers and patient organizations are currently active in more than 120, mostly disease specific, registries. None of them is used by all EURO-NMD centers and there is no unified NMD Registry in the EU. This dispersion of legacy data is known to hinder research.

AIMS

The development of the EURO-NMD registry was recently approved by HP-PJ-01-2019. Its general objective is to **build a hub for all neuromuscular diseases, including undiagnosed patients.**

- Harmonization of data collection amongst existing and future registries
- Development of an online system that will help with the evaluation of the ERN outcomes and
- Empowerment of patients and families to take an active role in the ERNs and healthcare
- Development of a registry hub that will allow linking and extraction of data from different sources
- Increase the amount and quality of NMD data available and ready to be used for care, healthcare planning; epidemiological and public health studies and research

PARTNERS

- Assistance publique – Hôpitaux de Paris (AP-HP) FR
- Medical Center – University of Freiburg (Freiburg) DE
- Stichting Katholieke Universiteit handelend onder de naam Radboudumc - Nijmegen (RUMC) NL
- Stichting United Parent Projects Muscular Dystrophy (UPPMD) NL
- Stichting Duchenne Data Foundation (DDF) NL
- Association Institut de Myologie (AIM) FR
- AFM- Telethon (AFM) FR

METHODS

- **Patient representative's involvement** from day 1
- Development by an **academic partner**
- Open architecture software - **REDCap**
- Follow **JRC and EJP-RD** specifications on rare diseases registries
- Generation of a **Privacy Preservation Record Link (PPLR) - EUPID** system
- Use of approved **Ontologies and Coding systems**
- Registration in the JRC's meta-registry platform - **ERDRI**
- Complete adherence to FAIR principles:

Interoperability layers to be **tested** by connecting the EURO-NMD registry with 4 other registries:

Dutch Neuromuscular Database CRAMP: as an example of a national, clinical registry that automatically imports data from the hospitals' clinical information systems

Duchenne Data Platform: as an example of patient organization-driven registry where patients or their relatives can register themselves and enter information about their health status and medical data

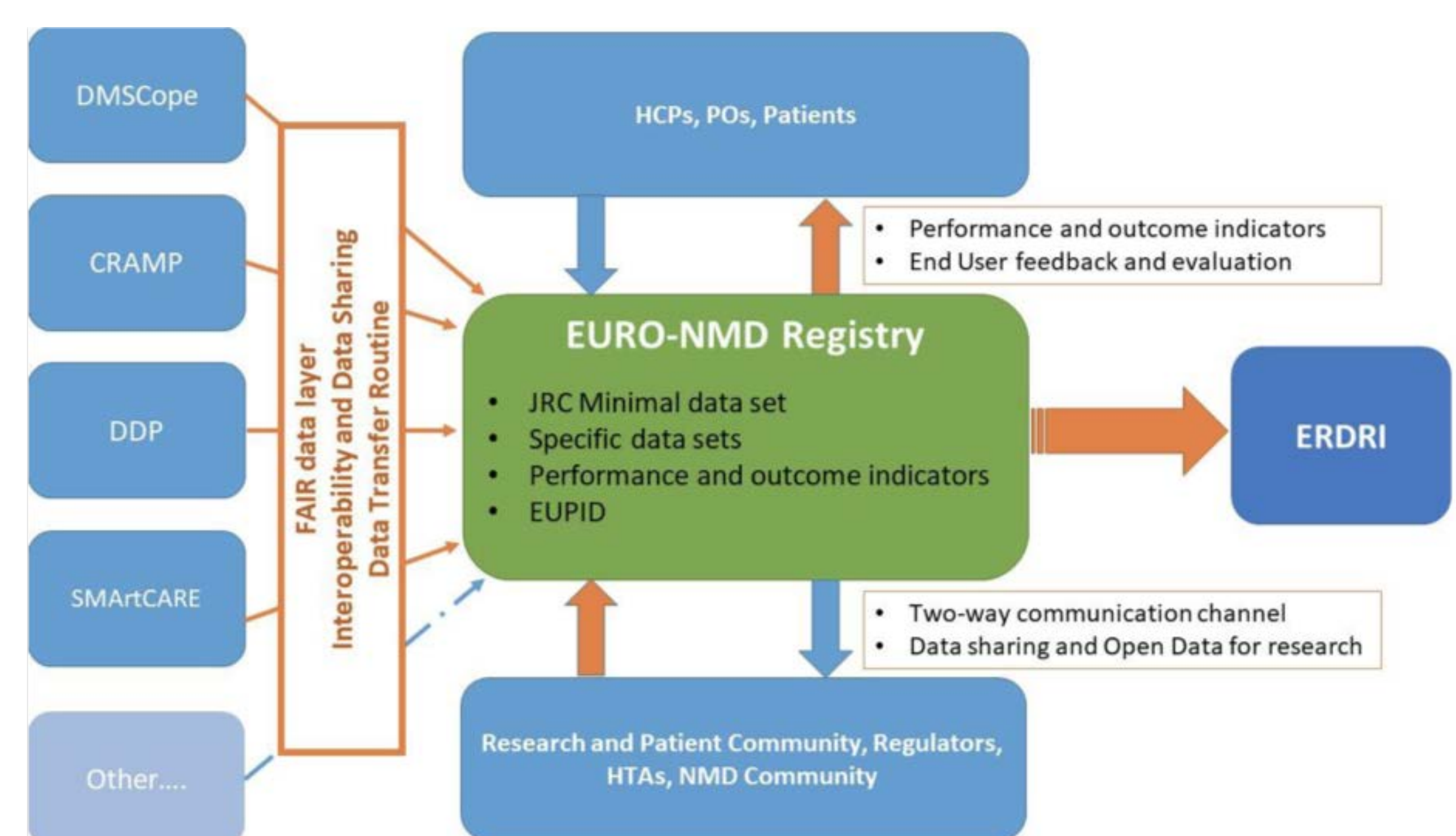
DM-Scope: FRENCH OBSERVATORY OF MYOTONIC DYSTROPHIES as an example of a national patient organization supported registry for Myotonic Dystrophy that allows both patient and clinician entered data

SMARtCARE: A platform to collect real-life outcome data of patients with spinal muscular atrophy as an example of registry focusing on patient recorded outcome measures (PROMs).

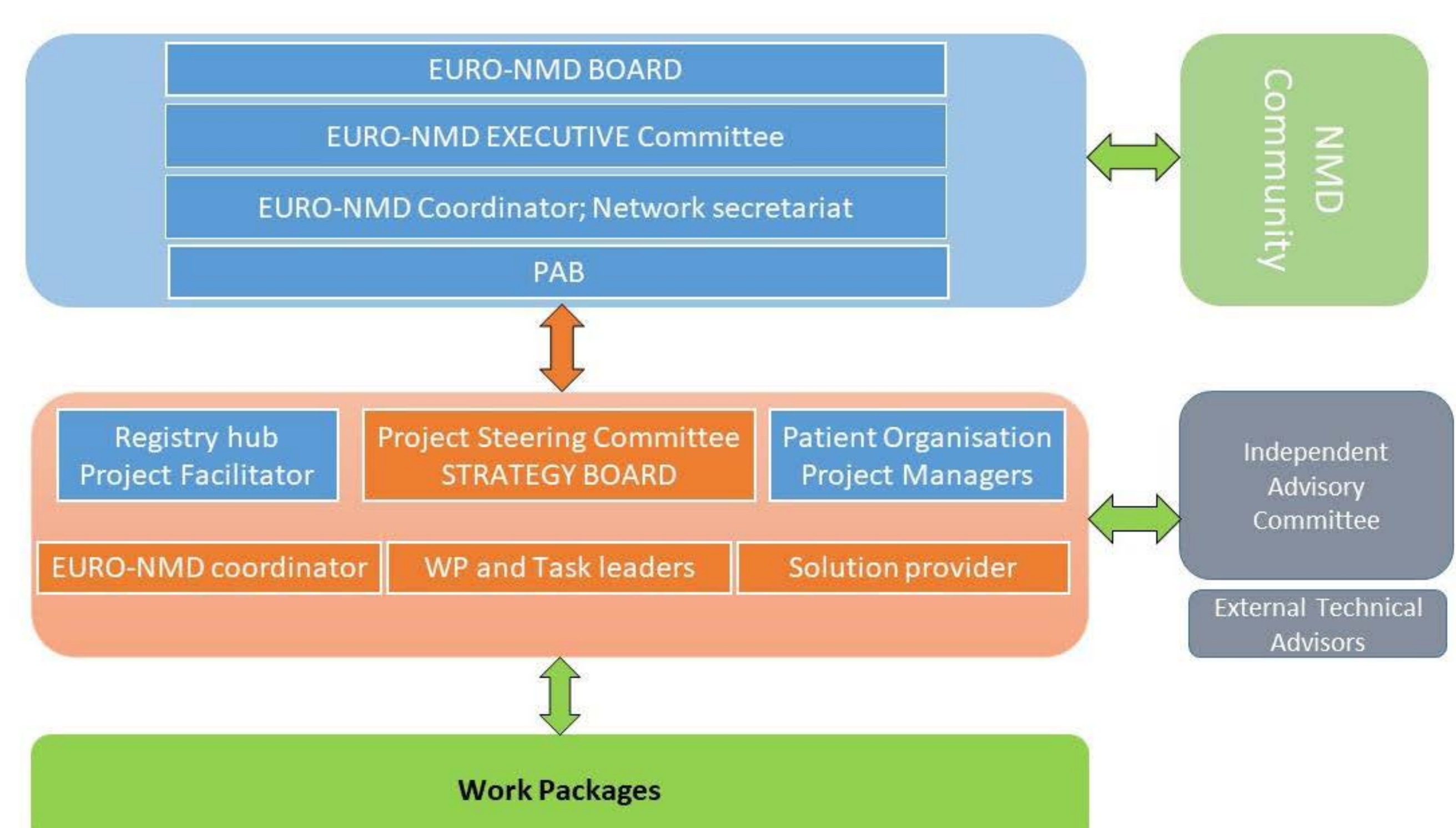
EURO-NMD, ERN for Rare Neuromuscular Diseases, spans 14 European countries, 61 reference centers, more than 100,000 patients.



Registry Hub Concept



Organisation of the EURO-NMD Registry Hub



CONCLUSION

A recognised challenge for rare diseases is the heterogeneity of legacy data sets and the multiplicity of existing registries. EURO-NMD HCPs and patient organizations are currently active in more than 120, mostly disease specific and patient run registries.

With the proposed approach and a successful implementation of the EURO-NMD Registry Hub, we will be able to achieve an efficient exchange of data with the major existing national and international neuromuscular registries and increase the "Research readiness" of our ERN.