FAIRification of Neuromuscular Disease Data

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FACTS & FIGURES

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LinkedIn:	PaLaDIn

CHALLENGE

- Neuromuscular diseases (NMD) are rare**,** hence patient data is inherently scarce and fragmented
- Combining these fragmented sources is essential to generate meaningful evidence
- Within PaLaDIn we are developing a data collection platform (**Interactium**) that integrates different sources of neuromuscular disease data (e.g., wearables, PROMs, patient preferences, registry data).

Findable **A**ccessible **I**nteroperable **R**eusable

My PhD aims to make this data **F**indable **A**ccessible **I**nteroperable and **R**eusable (FAIR) [1] .

APPROACH & METHODOLOGY

Problem → **Identifying** the current data collection efforts in NMDs by conducting a scoping review

Feasibility → **Mapping** the TREAT-NMD core datasets to CARE-SM[2], a semantic model for clinical and registry data

Scalability → **Accelerating** the FAIRification of NMD data by automating the mappings with AI

Value → **Evaluating** the pipeline and its benefits

RESULTS

NMD registries are semantically **fragmented and heterogenous** and there are little standards for reuse

Mapping TREAT-NMD core datasets to CARE-SM **enhances the FAIRness** of the data, because CARE-SM's ontology-based structure makes data machine-readable and interoperable

Impact on patients

Shortens path from patient data to insight

Interoperable data strengthens the evidence base for rare disease research

Supports faster trial recruitment

Manual mapping doesn't scale: we are planning to use AI to automate FAIRification

VALUE OF IHI COLLABORATION

Direct access to TREAT-NMD's Global Registry Network

Working with patient organizations, clinicians and industry partners

Opened pathways for multiple collaborations with researchers around the world

CAREER IMPACT

- Presented our work at multiple international conferences/workshops/events and co-organized workshops
- Learned to communicate with both patient and industry stakeholders
- Gained a lot of valuable contacts in the rare disease community

References:

[1] Wilkinson, Mark D., et al. "The FAIR Guiding Principles for scientific data management and stewardship." *Scientific data* 3.1 (2016): 1-9.

[2] Alarcón-Moreno, Pablo, and Mark Denis Wilkinson. "Take care of your patient data: clinical and registry entries (care) semantic Model." *Proceedings of the 15th International Conference on Semantic Web Applications and Tools for Health Care and Life Sciences (SWAT4HCLS 2024)*. Vol. 3890. 2024.

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