PCR243

Facilitating And Accelerating Patient-Reported Outcomes (PROs) Selection To Use For Clinical Research And Rare Diseases (RD)

- A European Rare Disease Research Coordination and Support Action (ERICA) project -

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- Objectives -

The lack of specific validated PROs in RD is a strong limitation in patient centric clinical research. However, the development of disease-specific PROs for more than 7,000 RD is not a realistic objective. As part of the ERICA project, we worked at maximizing existing knowledge on RD and optimizing use of existing PROs by developing a PROs Repository to support the identification of PROs eligible for use in RD clinical research.

- Methods - 1/ PROs functional coding and PROs Repository

Orphanet conducted a systematic documentation of the functional consequences of 551 RD using the Orphanet Disability Questionnaire (ODQ), derived from the International Classification of Functioning (ICF).

To identify PROs measuring RD functional impacts, MRT applies the same ICF coding rule to a selection of PROs identified in MRT PROQOLID™ database and from a survey conducted among 24 European Reference Networks (ERN).

PROs eligible for use in Rare Diseases

- The ERICA PROs Repository -

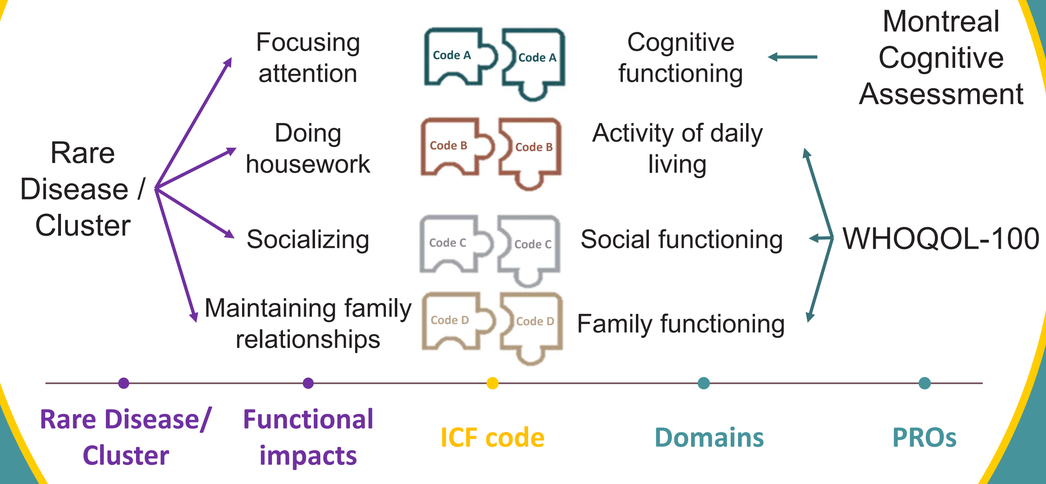

Fig 1: ERICA PROs Repository – Content

Available at: PROMs Repository | ERICA (erica-rd.eu)

2/ RD functional clustering:

To extend the identification of eligible PROs suitable for RD, a Multiple Factorial Analysis (MFA) has been performed to group together RD sharing similar functional impacts in clusters.

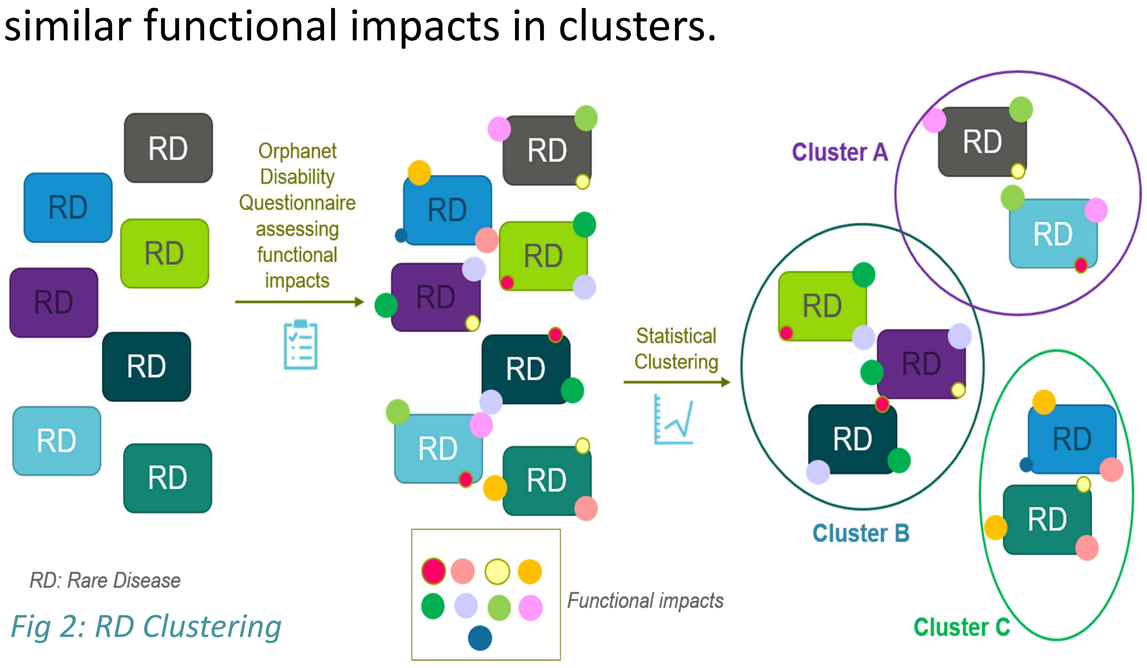

Fig 2: RD Clustering

- Results - 1/ PROs functional coding and PROs Repository

259 PROs developed in RD, 151 extra non-RD PROs measuring functional impacts (*e.g.* Motor Skills, Communication, Understanding and learning) and 160 measuring concepts of Interest for ERN *(e.g.* Burden, Self-efficacy, Adherence, Independence) were selected from PROQOLID™. The ERN survey identified 211 additional PROs. The **781 identified PROs** were domains-coded. They are now described in the **ERICA PROs Repository**, in a user- friendly interface allowing for quick and refined searches for relevant PROs:

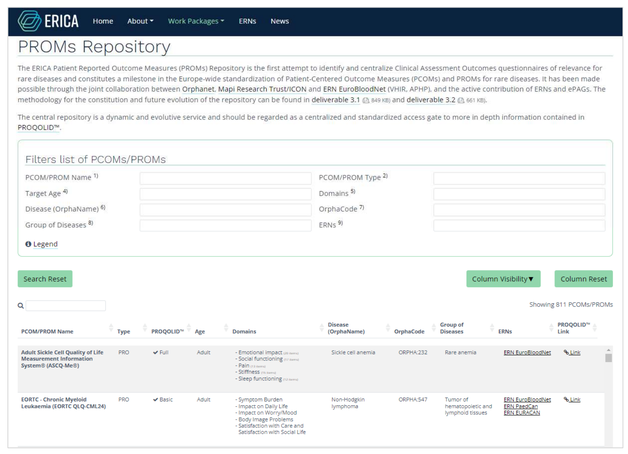

Fig 3: ERICA PROs Repository - Platform

Within PROQOLID™, there is additional PROs information, which can help ERNs decide if the selected PRO is well suited to their needs.

- Results - 2/ RD functional clustering

The 551 RD were grouped into 57 clusters sharing similar functional impacts, of which 21 include 6 to 58 RD.

SOURCES:
PROQOLID™: https://eprovide.mapi-trust.org/
Orphanet: https://www.orpha.net/consor/cgi-bin/index.php
Eurobloodnet: https://eurobloodnet.eu/
ERICA (WP3): https://erica-rd.eu/work-packages/patient-centred-research/

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- Conclusion -

The ERICA PROs Repository is the first attempt to identify and centralize PROs of relevance for RD. The clustering work will be implemented into the repository to extend the search results to RD with similar functional impacts thus to identify more PROs eligible for use in RD.