



Hierarchical clustering of rare diseases based on functional impacts to support the identification of potential Patient-Centered Outcomes Measures (PCOMs) for Rare Diseases research

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RD in cluster #22

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## Grant from:

## MAIN OUTCOMES:

- 551 RDs were clustered into 47 clusters with a specific functional impact profile
- Next step: Identification of RD-PCOMs within each cluster and function-specific PCOMs matching cluster functional impact profiles
- Impact: This work is a very promising step to identify PCOMs for RD lacking disease-specific and therefore turn RD patient experience into meaningful and comparable data

**RESULTS** 

- 1. First clustering work: 57 RD clusters, including 35 with less than 5 RDs and a large cluster of 65 RDs
- 2. Refinement: forcing cluster size to 10 RDs and merging subtypes under the related disorder
- 3. Second clustering work after refinement: 442 RD were gathered into 47 clusters, ranging from 10 to 12 RDs
- 4. Clusters validation by RD experts is on-going

NTDT-PRO© | Nontransfusion-Beta-thalassemia Dependent Thalassemia – PRO **Budd-Chiari syndrome** 22 Patient-Reported Outcome (PRO) **Cystic fibrosis** PedsQL™ 3.0 Rheumatology Module **Dermatomyositis** MAP - USA | Myositis Activity Profile Immune-mediated necrotizing myopathy PedsQL™ 3.0 Rheumatology Module Juvenile dermatomyositis Juvenile polymyositis MAP - USA | Myositis Activity Profile **Polymyositis** Refsum disease Sickle cell anemia **25 PRO TEMPI** syndrome Von Hippel-Lindau disease Cluster example with existing RD-specific PCOMs

RD-specific PCOMs

Cluster #22 functional impact profile Engaging in sexual relations / Engaging in intimate relationships Performing vigorous activities Moving outside their home / Moving within their home

- Drinking / Eating
- Dressing/Undressing
- Managing their health / Washing themselves
- Falling asleep Participating in cultural activities
- /Practicing sports Playing with others (as a child)
- Socializing / Taking part in community life Travelling
- Handling emotions and mood

1. RD 2. Qualitative 3. Identification of existing PCOMs qualitative data data clustering **Cluster A with functional** impact profile A Self-Care Motor Self-Care Self-Care RD<sup>A</sup> **RD**<sup>A</sup> **RD**<sup>A</sup> PCOMs match with \_\_\_\_ Self-Care Interviewees: OR Cluster A impact clinicians and Self-Care **RD**<sup>B</sup> profile patient RD<sup>B</sup> **RD**<sup>B</sup> 조조조 ||| ||| ||| **№** ■ **№** representatives Motor  $\subseteq =$ **METHODS** Hierarchical Daily activity **Tool**: Orphanet clustering: Multiple **Cluster B with functional** Disability **RD**<sup>c</sup> Daily activity impact profile B **RD**<sup>c</sup> Factorial Analysis + Questionnaire (113 Refinement items) derived and **RD**<sup>c</sup> Communication adapted from the Daily activity Communication WHO **RD**<sup>D</sup> Daily activity RD<sup>C</sup> – specific PCOM Classification of We also used Natural Language Processing RD<sup>D</sup> to identify PCOMs for RD Functioning (ICF) could be suitable for **RD**<sup>D</sup> Visit the poster **PCR85** "Facilitating the Communication Identification of Appropriate Patient-Centered RD<sup>D</sup> since they share RD<sup>C</sup> – specific Outcome Measures (PCOMS) in Rare functional similarities Communication Disease (RD) Clinical Research Using **PCOM** Functional Impacts Matching".by scanning the QR Code at the top right-hand Functional Daily activity Communication impacts **442 Rare** 6.000 **551 Rare PCOMs** diseases diseases PR@@OLID orphadata We connected these two databases!

RESSOURCES

Mapi ResearchTrust database with data on 6.000 PCOMs

https://eprovide.mapi-trust.org/

Visit the poster PCR28 "Facilitating and Accelerating Patient-Centered Outcomes Measures Awareness and Use in Clinical Research in Rare Diseases" by scanning

the QR Code at the top right-hand

Orphanet database of RD functional consequences annotation http://www.orphadata.org

**OBJECTIVE** 

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CHALLENGES

Identify existing PCOMs suitable for RDs based on functional impacts similarities

The lack of specific validated PCOMs in RDs is a strong limitation in patient centric clinical research and development of disease specific PCOMs for more than 7,000 RDs is not a realistic objective