



Grant from:



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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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

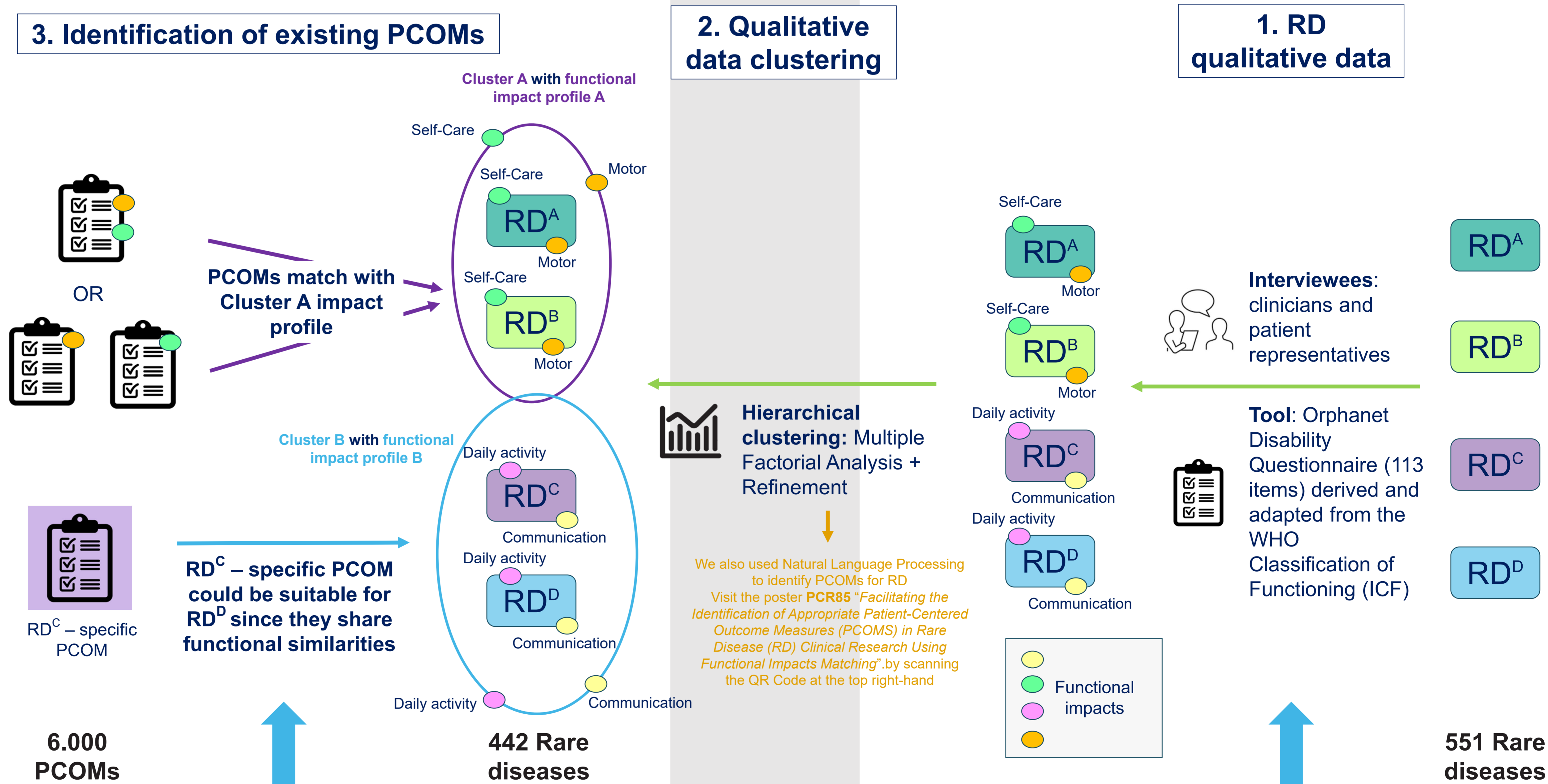
RESULTS

- 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

Cluster example with existing RD-specific PCOMs

RD in cluster #22	RD-specific PCOMs	Cluster #22 functional impact profile
Beta-thalassemia	NTDT-PRO® Nontransfusion-Dependent Thalassemia – PRO	<ul style="list-style-type: none"> • Engaging in sexual relations / Engaging in intimate relationships • Performing vigorous activities • Moving outside their home / Moving within their home • Dressing/Undressing • Drinking / Eating • 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
Budd-Chiari syndrome	-	
Cystic fibrosis	22 Patient-Reported Outcome (PRO)	
Dermatomyositis	PedsQL™ 3.0 Rheumatology Module MAP - USA Myositis Activity Profile	
Immune-mediated necrotizing myopathy	-	
Juvenile dermatomyositis	PedsQL™ 3.0 Rheumatology Module	
Juvenile polymyositis	-	
Polymyositis	MAP - USA Myositis Activity Profile	
Refsum disease	-	
Sickle cell anemia	25 PRO	
TEMPI syndrome	-	
Von Hippel-Lindau disease	-	

METHODS



RESSOURCES

PROQOLID™
Clinical Outcome Assessments Database
Mapi ResearchTrust database with data on 6.000 PCOMs
<https://eprovide.mapi-trust.org/>

We connected these two databases! 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

orphadata
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