

WP3 - Patient-Centered Research in Rare Diseases

- 4th ERICA General Assembly -

Udine, December 11th, 2024

Agenda

1. PCOMs Repository

- A. Update on the current content
- B. Ever dynamic & evolving contribution to the repository

2. Pilot studies

- A. Presentation of the 3 studies
 - i. Application in Clinical Trials: Satisfy Trial (ERN-EuroBloodNet) **Andreas Glenthøj**
 - ii. Application in Registry: Peter Registry (ERN Transplantchild) **Miguel Clemente Bernal**
 - iii. Application in Policy-making process: (Rare Diseases Ireland) **Vicky McGrath**

3. Continuity in ERDERA & future PCOMs/PROMs development

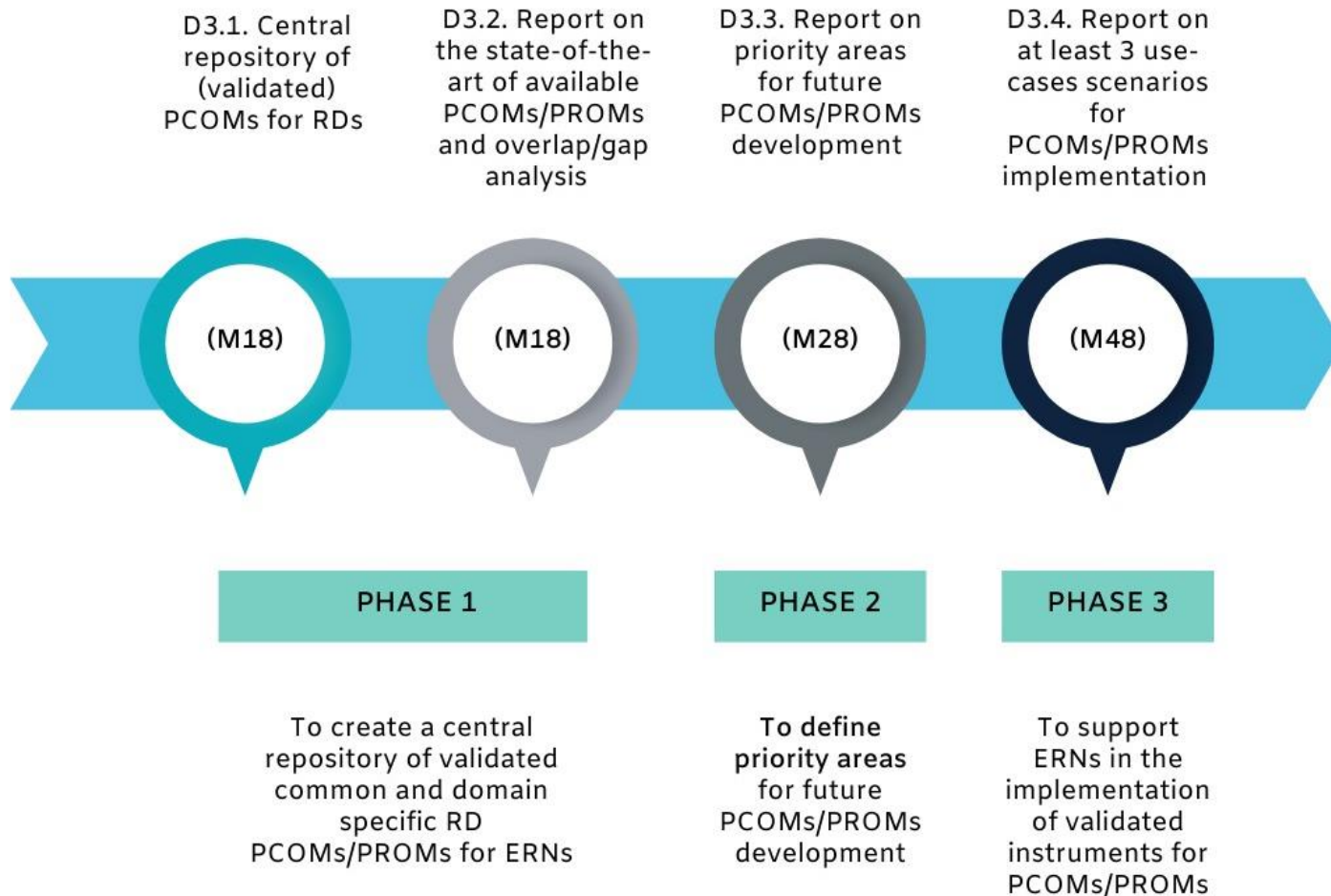
4. Questions & Answers

WP3 Objectives

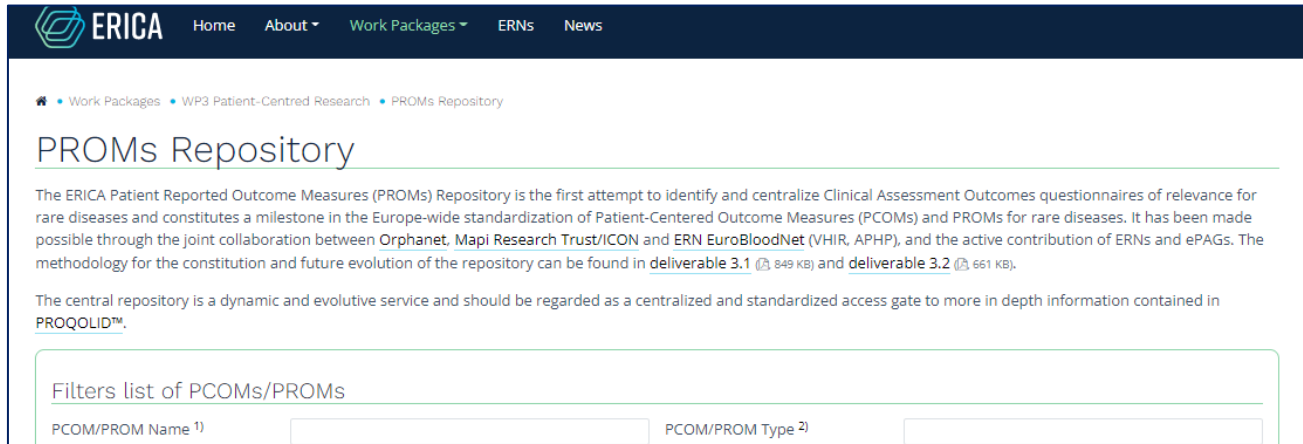
- 1. To create a central repository** of PCOMs/PROMs for ERNs
- 2. To define priority areas** for future PCOMs/PROMs development
- 3. To support ERNs** in the implementation of validated instruments for PCOMs/PROMs in 3 Pilot studies

➔ Review of each objective achievements and continuity in ERDERA

Timeline



1. PCOMs Repository



The screenshot shows the ERICA website's navigation bar with links for Home, About, Work Packages, ERNs, and News. Below the navigation bar, there is a breadcrumb trail: Work Packages > WP3 Patient-Centred Research > PROMs Repository. The main heading is "PROMs Repository". The text below the heading describes the repository as the first attempt to identify and centralize Clinical Assessment Outcomes questionnaires for rare diseases, mentioning its collaboration with Orphanet, Mapi Research Trust/ICON, and ERN EuroBloodNet. It also refers to deliverables 3.1 and 3.2. A section titled "Filters list of PCOMs/PROMs" contains two input fields: "PCOM/PROM Name 1)" and "PCOM/PROM Type 2)".

→ Repository historical

- **V1.0 released in August 2022**
- **Feedback from ERNs: to include ObsROMs + recently developed PROMs/ObsROMs**
- **V2.0 in November 2023**

ERICA PROMs repository

<https://erica-rd.eu/work-packages/patient-centred-research/proms-repository/>

Repository v2.0 content:

- Observer-Reported Outcome Measures (ObsROMs) (242)
- ERN Survey and direct feedback (4)
- PROMs developed in RD: MRT Literature review and Orphanet watch (73)
- Quality of life PROMs and ObsROMs (88)

→ Repository current content (dec 2024): (1081)

- Rare Diseases specific PROMs/ObsROMs (444)
- Function specific PROMs/ObsROMs (383)
- Quality of life PROMs/ObsROMs (145)
- Clinical practice PROMs/ObsROMs (109)

Ever dynamic & evolving contribution to the repository

PROMs Repository

The ERICA Patient Reported Outcome Measures (PROMs) Repository is the first attempt to identify and centralize Clinical Assessment Outcomes questionnaires of relevance for rare diseases and constitutes a milestone in the Europe-wide standardization of Patient-Centered Outcome Measures (PCOMs) and PROMs for rare diseases. It has been made possible through the joint collaboration between [Orphanet](#), [Mapi Research Trust/ICON](#) and [ERN EuroBloodNet](#) (VHIR, APHP), and the active contribution of ERNs and ePAGs. The methodology for the constitution and future evolution of the repository can be found in [deliverable 3.1](#) (📎 849 KB) and [deliverable 3.2](#) (📎 661 KB).

The central repository is a dynamic and evolutive service and should be regarded as a centralized and standardized access gate to more in depth information contained in [PROQOLID™](#).

NEWS! (May 2023): Content of the repository has been enriched with Observer-Reported Outcome Measures (ObsROMs), with PROMs & ObsROMs measuring quality of life as well as PROMs & ObsROMs recently developed for rare diseases.

Your contribution is crucial to identify new measures and keep alive and up-to date the repository – If you know about PROMs, ObsROMs or other Patient Centered-Clinical Outcomes Assessment (PC-COA) measures (Clinician-Reported Outcome Measure (ClinROM) and Performance Outcome (PerfO)) developed or validated for any rare diseases not included yet in the repository.

Please Contribute →

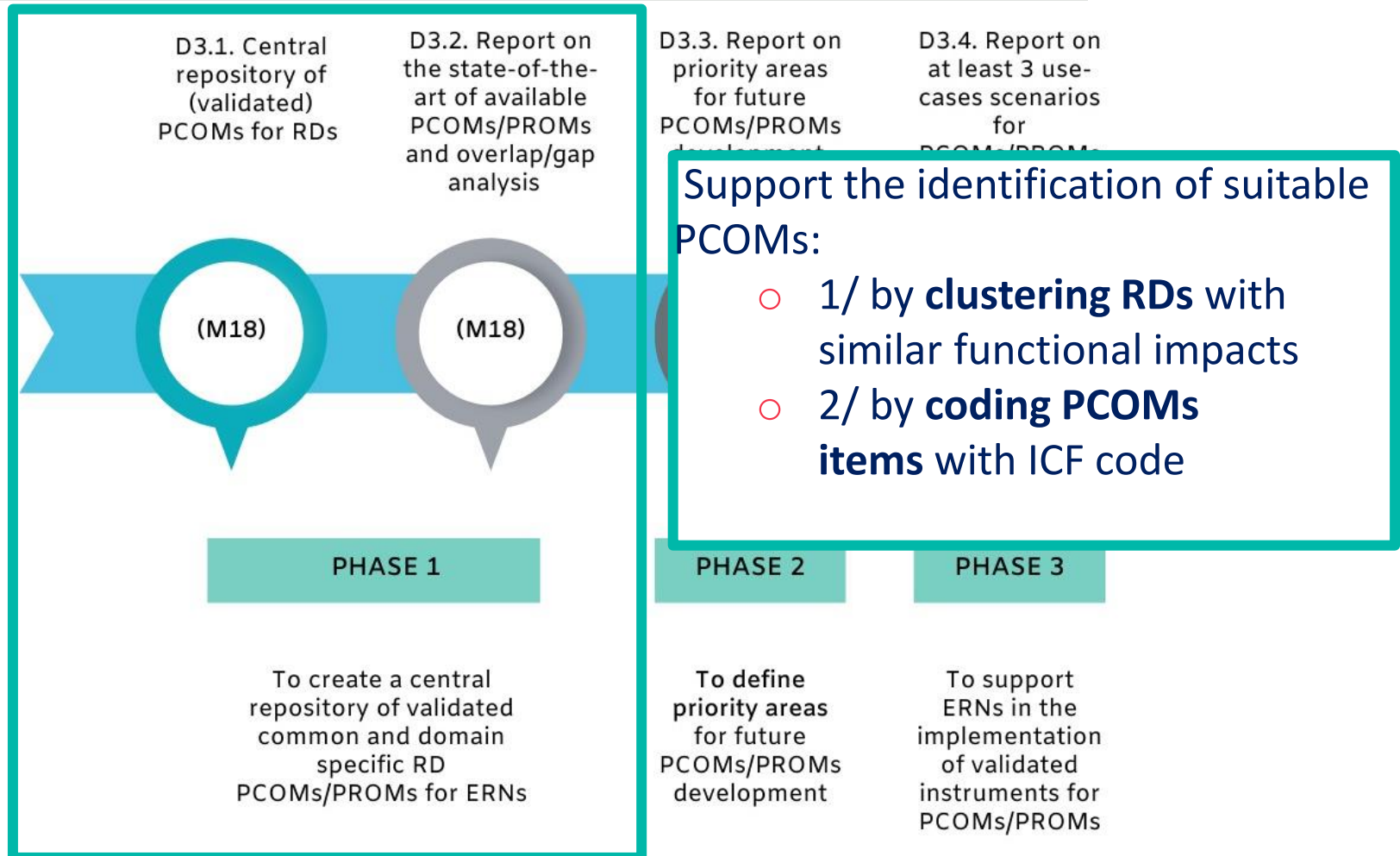


To contribute to the repository:
the ERN Contribution Form

Timeline



erica-rd.eu/work-packages/patient-centred-research/proms-repository/



2. Pilot studies

Timeline

Application in **Clinical Trials**

Satisfy Trial (ERN-EuroBloodNet) *Andreas Glenthoj*: PROMs Selection Process.
Associating PROMs in PKD in Clinical Trials in Danish Red Blood Cell centers.

Application in **Registry**

Peter Registry (ERN Transplantchild) *Miguel Clemente Bernal*: Registry points for clinical trials gathering (Web)+ PROMs questionnaire (mobile app) for monitoring transplant phases for pediatric population

Application in **Policy-making process**,

Vicky McGrath, CEO Rare Diseases Ireland.
PROMs as tools to support patient submissions for reimbursement decision making for orphan medicines (OMPS) at the NCPE/HSE.

D3.4. Report on at least 3 use-cases scenarios for PCOMs/PROMs implementation

(M48)

PHASE 3

To support ERNs in the implementation of validated instruments for PCOMs/PROMs

To be submitted by M48

3 pilots identified

Application in Clinical Trials:

Satisfy Trial (ERN-EuroBloodNet) – **Thomas Doeven, Andreas Glenthoj**

Application Registry:

Peter Registry (ERN T) **Guilherme Clemente Bernal**


REC

Application in policy-making process:

Rare Diseases Ireland - **Vicky McGrath**



PAEDIATRIC
TRANSPLANTATION
EUROPEAN
REGISTRY



peter

PETER:

PaEdiatric T ransplantation E uropean R egistry

Miguel Clemente Bernal
e-Health Support Manager, ERN-TransplantChild

3. Continuity in ERDERA & future PCOMs/PROMs development

Continuity in ERDERA – PCOMs repository

→ Development of a new interface to identify suitable COA for Rare Disease trials (WP16.1)

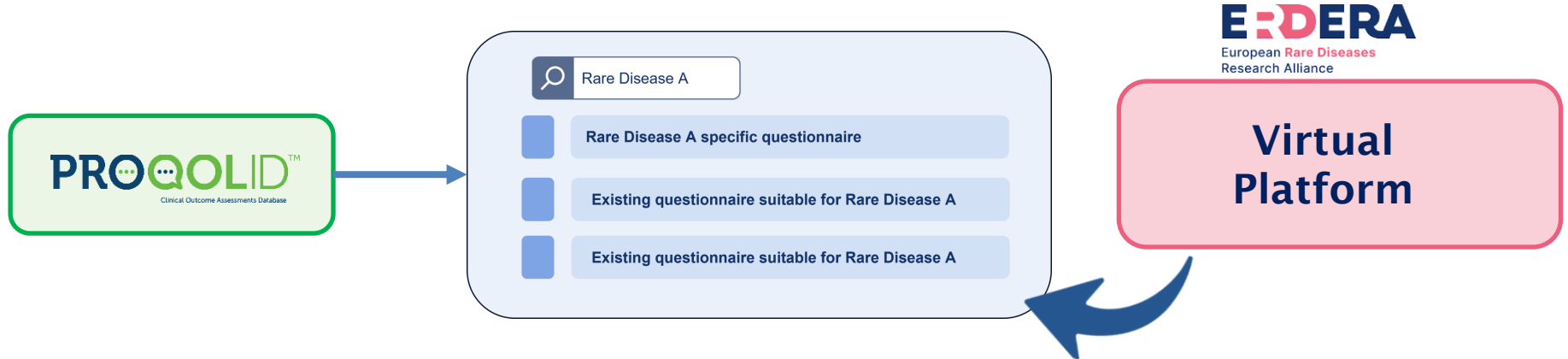
New interface



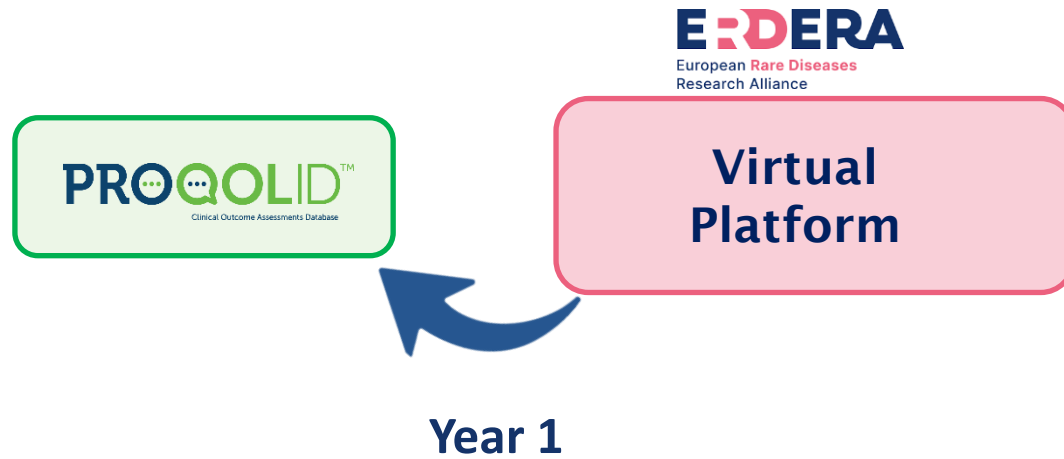
Search bar:

- Rare Disease A specific questionnaire
- Existing questionnaire suitable for Rare Disease A
- Existing questionnaire suitable for Rare Disease A

Continuity in ERDERA – PCOMs repository



Continuity in ERDERA – PCOMs repository



Continuity in ERDERA – PCOMs repository

- **Year 1:** Connect the PROQOLID™ to the Virtual Platform (according to Orphacodes and Rare Disease name)
 - ➔ ERICA repository transfer to the ERDERA
 - ➔ Access to all RD-specific PCOMs
 - ➔ Updated & dynamic content
 - ➔ PerfO & ClinRO also included
 - ➔ PROQOLID free access continuity for ERNs
- **Year 2-3:** Development of the PCOMs suggestion platform based on:
 - RD functional impact analysis
 - ERN PCOMs endorsement

Pathways to Optimized RD-RWD Collection and Analysis

9.1

Utilizing Electronic Health Records Across Interoperability and Language Barriers

9.2

Utilizing Population Based RD Data for Outcomes Research

9.3

Integrating patient cohorts for natural history / standard of care reference studies

9.4

Developing blueprints for regulatory-grade RD-RWD cohort data

9.5

Developing tools for modeling and predicting RD progression

9.6

Developing a regulatory-grade platform to simulate clinical trial interventions

Continuity in ERDERA – Pilot studies (Development and Implementation of PCOMs) Lead by María del Mar Mañú Pereira

Year 1: Development of methodologies to collect and implement data from PCOMs related to patient registries, considering legal, technological, and regulatory requirements

- **Study Protocol Development:** Collect clinical and technical requirements from end-users and standardize data (**Collaboration with Data Services Hub**)
- **Identification of Mobile Apps and Devices** and assessment of their research readiness (**Collaboration with WP14: Data readiness services**)
- **Clarification of Regulatory Aspects**, including privacy and security protections in mobile health (**Collaboration with WP18: Regulatory support service**)

Year 2-3:

- Development/Adaptation of Mobile Health Solutions for use with patient registries (ERN Registries)
- Execution of Clinical Studies (**Collaboration with WP15: Data sharing and analysis services**)
- Validation of Patient-Generated Data
- Development of a Data Visualization Framework (**Collaboration with WP15: Data sharing and analysis services**)

Case Studies

T10.2: Development and Implementation of COA Tools

1

Rare neuromuscular
diseases
ERN-NMD
PI: Evangelista
AP-HP

2

Rare neurological
diseases
ERN-RND
PI: Synofzik
EKUT

3

Rare Kidney
Disorders
ERKNet
PI: Schaefer
UKHD

4

Rare Hematological
Disorders
ERN-EuroBloodNet
PI: Mañú-Pereira
VHIR

Questions & Answers





THANK YOU!