Funded by the European Union

European Reference Network for rare or low prevalence complex diseases

Network Neuromuscular Diseases (ERN EURO-NMD)

8th ERN EURO-NMD Annual Meeting

ERDERA – Funding and Clinical Research Network contributions for ERNs development

5th – 7th March 2025

Gisèle BONNE

Centre de Recherche en Myologie

Sorbonne Université - Inserm - Institut de Myologie,

G.H. Pitié-Salpêtrière, Paris, France

CENTRE DE RECHERCHE EN MYOLOGIE

SORBONNE UNIVERSITÉ CRÉATEURS DE FUTURS DEPUIS 1257

RÉPUBLIQUE FRANÇAISE Liberté Égalité Fraternité

Inserm La science pour la santé From science to health

INSTITUTE OF MYOLOGY CURE THROUGH INNOVATION

The ERN EURO-NMD is funded by the European Commission under the EU4Health programme (EURO-NMD 23-27 — 101156434 — EU4H-2023-ERN2-IBA)

Connecting the dots in the rare diseases research ecosystem

Objectives

To improve the health and well-being of 30 million people living with a rare disease in Europe, by making Europe a world-leader in RD research and innovation.

- Diagnosis established or enrolment in systematic research in average within 6 months after coming to medical attention
- New effective therapies for rare diseases approved in Europe and beyond, the majority of which addressing diseases without approved options
- Better understanding of the impact of rare diseases on patients, families and society to improve quality of life

ERDERA
European Rare Diseases Research Alliance

181 Organisations

40 funders

81 research performing organisations

9 patients' organisations

3 research infrastructures

24 private for-profit partners (industry & SME)

24 other (univ, hospital, non-profit, public administration)

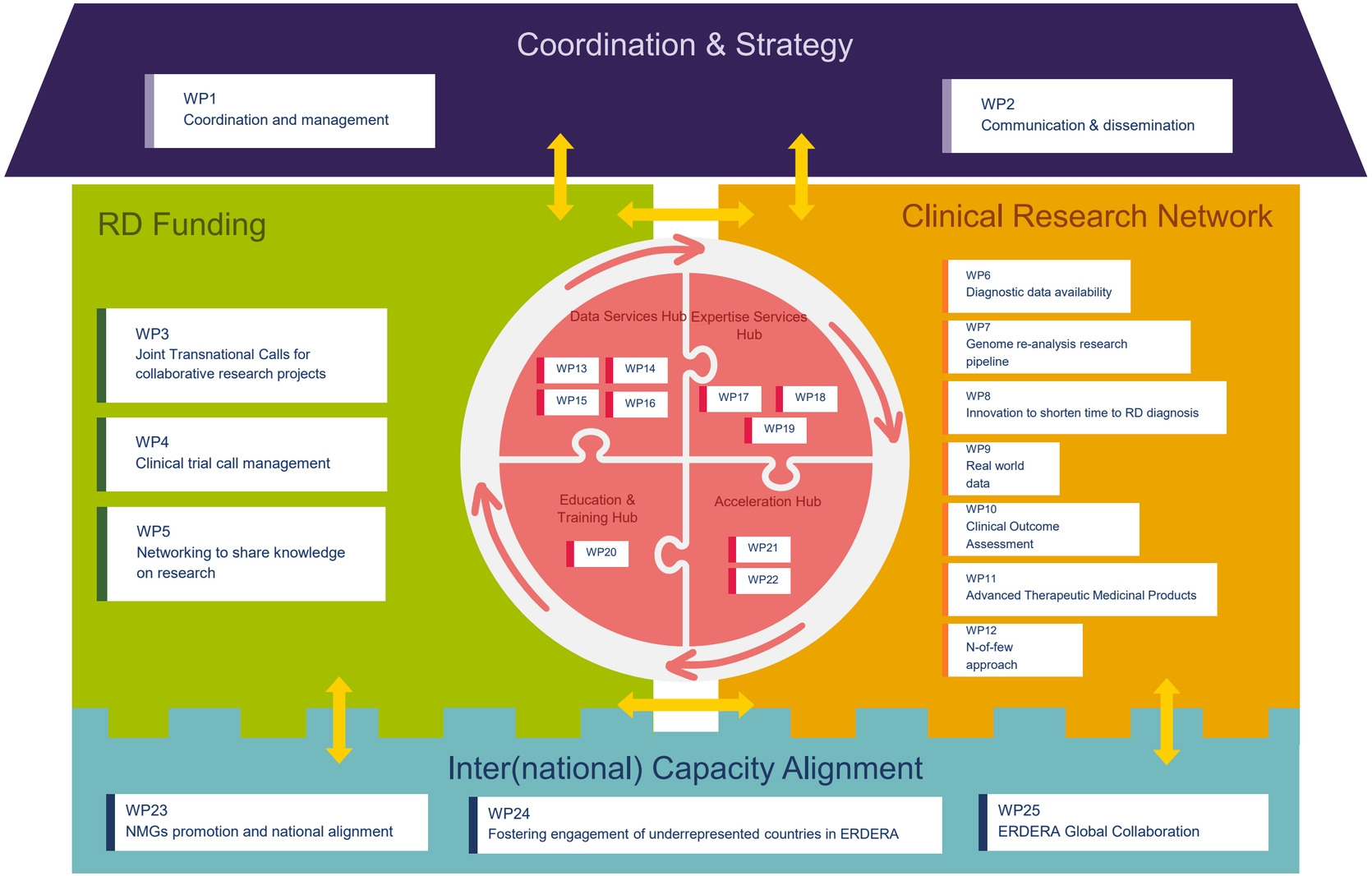
36 Countries

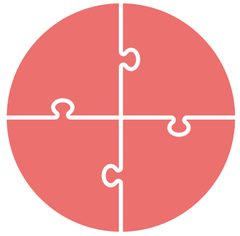
25 EU member states

8 associated countries

3 non-EU*

*at the time of proposal submission





WP13
Rare Diseases-Virtual Platform (RD-VP): Finding and accessing the data ecosystem

WP14
Data readiness services

WP15
Data sharing and analysis services

WP16
Knowledge bases and ontologies for RD research

WP17
Mentoring and consultancy

WP18
Regulatory support service

WP19
Methodological Support

WP20
Education and training in rare diseases research

WP21
Technology accelerator

WP22
Public-Private Collaboration Accelerator

Co-funded by the European Union

National Mirror Groups

ERDERA European Rare Diseases Research Alliance

The purpose of NMGs

- The concept of NMGs was developed to help bridge the gap between the European/International RD level, on the one hand, and national RD level on the other: effectively, this means
  - Channeling national needs, realities and good practices into ERDERA, to help shape its activities
  - Optimise the dissemination – and hopefully the implementation and use – of the outputs and lessons of ERDERA into the national landscape
- Furthermore, NMGs should enable more focused interactions and good practice sharing *between* countries
- NB – Can be an existing or new group/body: and can be mostly research-focused or look at broader RD issues.

Funding Opportunities

ERDERA European Rare Diseases Research Alliance

Joint Transnational Calls

MAIN GOAL: enable scientists in different countries to build an effective collaboration on a common interdisciplinary research project based on complementarities and sharing of expertise, with a clear future benefit for patients

Typical success rate: 1st stage vs final funding = 10-12%; 2nd stage vs final funding 35 -50%. Typical overall project budget 1.0 – 1.5 Million €

https://erdera.org/funding/#joint-translational-call

Launched every year in December with pre-announcement the latest in November

2-stage evaluation process. 3-years projects

A minimum of 3/4 eligible partners and a max. of 6 per project (can be extended to 8 according to specific conditions)
PAOs participation is financed

Networking Support Scheme

Financial support to applicants for fostering organization of workshops or conferences for new research networks or existing/expanding research networks to strengthen collaborations and to enable exchange of knowledge.

Share knowledge on research among relevant transnational European and transcontinental RD stakeholders including clinicians, basic researchers, patients' organisations/RD support groups, as well as PhD students, post-docs and early career researchers, and industry

Build new European and global research networks or expanding existing research networks on RDs and rare cancers to support patient-centred research, and to include stakeholders of widening countries and young researchers

30 000 €/event

Continuously open (eligibility check every 6 months)

Collaborative (min. 3 applicants from 3 ERDERA countries)

Clinical trials call

MAIN GOAL: Current treatments are usually supportive rather than disease-modifying, leaving most patients with rare diseases with considerable unmet medical needs. Tackling these issues will require **valid and reproducible clinical trials, and opportunities for those affected by rare diseases to influence and participate in these trials.**

Definition of call topic and rules in 2025. Opening of the call in 2026. Final decision by end of 2027 and funding of clinical trials between 2028-2031 with possible extension to 2034.

Two committees: Clinical Trials Scientific Committee (CTSC) responsible for triage of pre-proposals based on scientific merit and overall relevance; and Clinical Trails Evaluation Board (CTEB) responsible for the evaluation of full proposals.

30 million € budget from the European Commission + possible additional funds from national funding bodies

Participation of patients to be defined

Support services for RD research acceleration

ERDERA European Rare Diseases Research Alliance

Expert Mentoring & Innovation Management Toolbox for RD research projects

- In collaboration with eatris
  European infrastructure for translational medicine
- **What is this?** → Service for researchers planning a project that has **translational potential** for RDs
- **Why?** → to increase the impact of projects by providing tailored expertise along each step of the translational pathway
- **When?** → in the pre-proposal stage OR after having received funding
- **How?** → after preliminary discussion and (if needed) confidentiality agreement, EATRIS identifies the most suitable experts that advise and/or accompany the proposal/project
- **Which expertise?** → You can get support on the following topics: Translational feasibility/ Regulatory compliance/Product classification/Quality assurance and control/Intellectual property strategy/Suitability of analytical readouts/Manufacturing/Therapy development
- **How much does it cost?** → service is provided for free
- **Interested? More info** → contact us: coordination@erdera.org & **https://www.ejprarediseases.org/mentoring/**
- **IMT: https://imt.ejprarediseases.org → < 400 resources and specific use cases curated to maintain database updated**

Categories

- Translational Project Management
- Research and Drug Development
- Regulatory Science
  - Preclinical Studies
  - Early Acces Support
  - Biological
  - Clinical Development
  - Marketing Authorisation
  - Ethic and Legal Issues
  - HTA
- Funding Sources
- Intellectual Property

Subjects

All Subjects | Clinical Trials Toolkit | Research and Drug Development | Regulatory Affairs | Early Access Support | Clinical Development | Clinical Trial Design | Scientific Advice | Clinical Trials | Orphan Drug | Clinical Trial | Rare Disease | Guideline | General Data Protection Regulation | GPDR | Regulatory Advice | Advanced Therapy | Biomarker

Regulatory Science

The information proposed in this section provides key elements on the regulation of medicines for Rare Diseases in human and provides support in the:
Health Technology Assessment,
Drug repurposing,
Preclinical suties,
Early access support,
Clinical Development,
Marketing Authorization,
Ethic and legal issues,
Regulatory Agencies.

Showing **217** results for: Regulatory Science

All Subjects

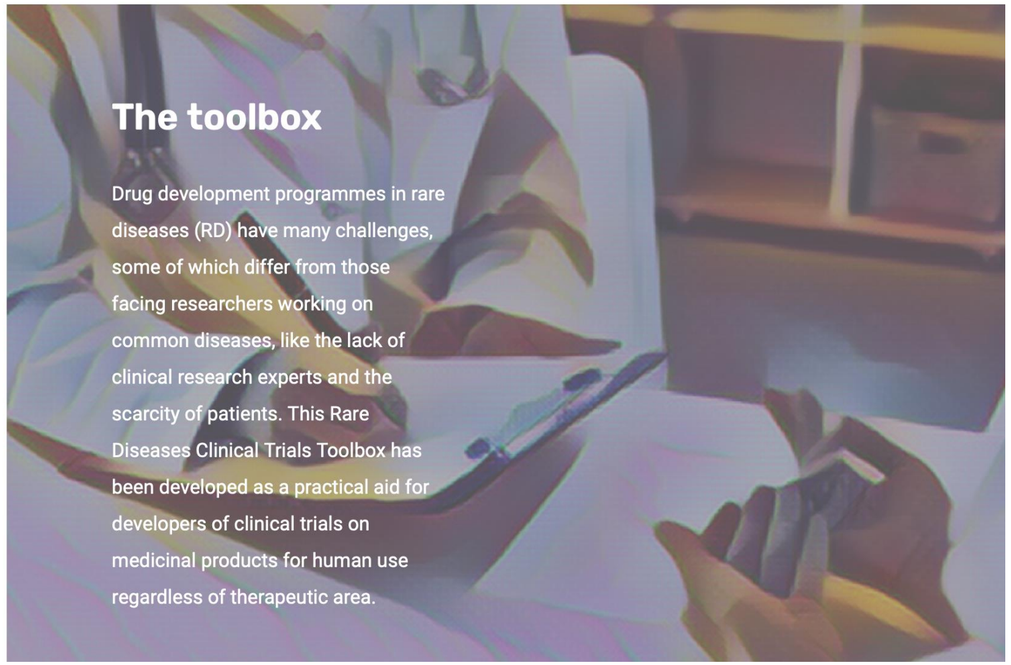
A neutral comparison of statistical methods for analyzing longitudinally measured ordinal outcomes in rare diseases

Published by Wiley

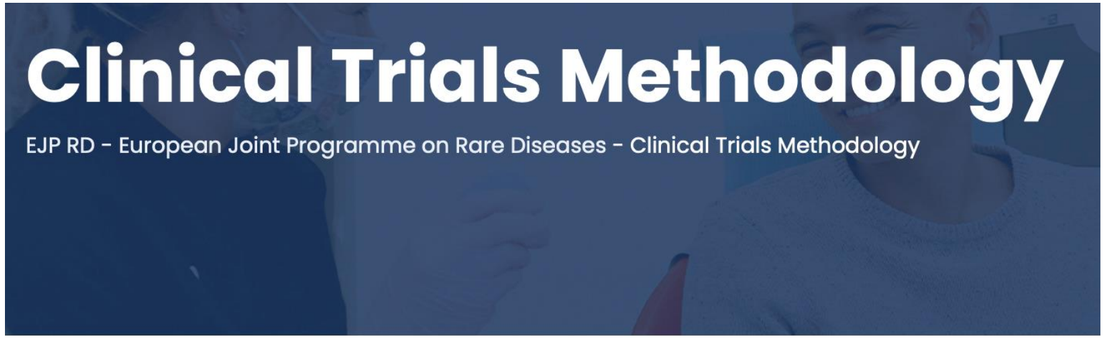
Research and Drug Development | Clinical Trial Design

How to Analyze Continuous and Discrete Repeated Measures in Small-Sample Cross-Over Trials?

Clinical Trials Toolbox & Methodology Support



https://www.ejprarediseases.org/rare-diseases-clinical-trials-toolbox/



https://www.ejprarediseases.org/clinical-trials-methodology/

Basic Courses | Intermediate Courses | Advanced Webinars | Videos

Cover:

- Recommended **courses for fundamentals in CTs** (not developed by EJP RD)
- **Intermediate courses** aim to provide a specialized statistical training to fill the knowledge gap in clinical trial methodologies
- **Advances webinars** addressed to people willing to conduct clinical research in rare diseases with the objective to train them in terminology, communication and understanding of RD clinical trial methodology.

Some topics covered: randomisation/composite endpoints/statistical evaluation of surrogate endpoints/challenges with master protocols, etc.

Improving the data ecosystem

ERDERA European Rare Diseases Research Alliance

ERDERA Data Hub & EJP RD Virtual Platform

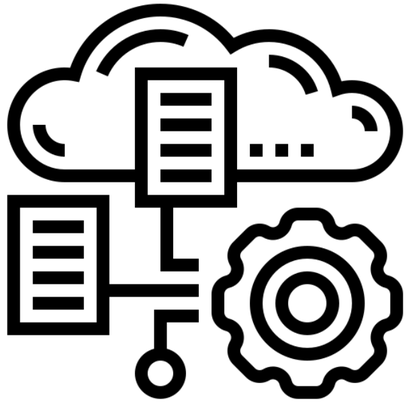
22 resources connected (11 registries, 3 catalogues, 2 gen-phen deposition infras, 5 knowledge bases, 1 project)

Over +**100 biological pathways** created

3 linkage/discoverability levels

https://vp.ejprarediseases.org

RD VIRTUAL PLATFORM



Data (re)analysis

FAIR stewardship

Computing infrastructure

RD DATA HUB

https://vp.ejprarediseases.org

ERDERA European Rare Diseases Research Alliance

EJP RD

Education & training opportunities

ERDERA European Rare Diseases Research Alliance

Massive Online Open Courses

Free & open to all

Foundation for Rare Diseases & European Joint Programme on Rare Diseases

Diagnosing Rare Diseases: from the Clinic to Research and back

4.9 (54 reviews)

5 weeks · 3 hrs per week

Included in Unlimited

Find out more

Foundation for Rare Diseases & European Joint Programme on Rare Diseases

Innovative Therapies and Personalised Medicine for Rare Diseases

4.6 (5 reviews)

5 weeks · 4 hrs per week

Included in Unlimited

Find out more

Foundation for Rare Diseases & European Joint Programme on Rare Diseases

Health Data Ethics & Regulatory Frameworks in Rare Disease Research

4 weeks · 3 hrs per week

Included in Unlimited

Find out more

Foundation for Rare Diseases & European Joint Programme on Rare Diseases

NMD

From Lab to Clinic: Translational Research for Rare Diseases

4.9 (15 reviews)

5 weeks · 4 hrs per week

Included in Unlimited

Find out more

https://www.futurelearn.com/partners/french-foundation-rare-diseases

Education & training

Patients' and young researchers' training: Blended and scalable training programme for patients & young researchers across the whole RD research pipeline

Training workshops for paediatric patients

RD research training for multistakeholder community

Trainings to empower RD diagnostic research

Trainings to empower RD clinical research

Trainings to empower knowledge on research methodologies

Trainings to empower acceleration of research and RD clinical trial methodologies and management

Trainings to empower RD data research

Clinical Research Network

DIAGNOSIS

- Diagnostic data availability
- Genome re-analysis research pipeline
- Innovation to shorten time to RD diagnosis

CLINICAL TRIAL READINESS

- Real world data
- Clinical Outcome Assessment

THERAPY DEVELOPMENT

- Advanced Therapeutic Medicinal Products
- N-of-few approaches

ERDERA in house research

ERDERA European Rare Diseases Research Alliance

CRN: Diagnostic Research workstream

ERDERA
European Rare Diseases Research Alliance

nature medicine

Article

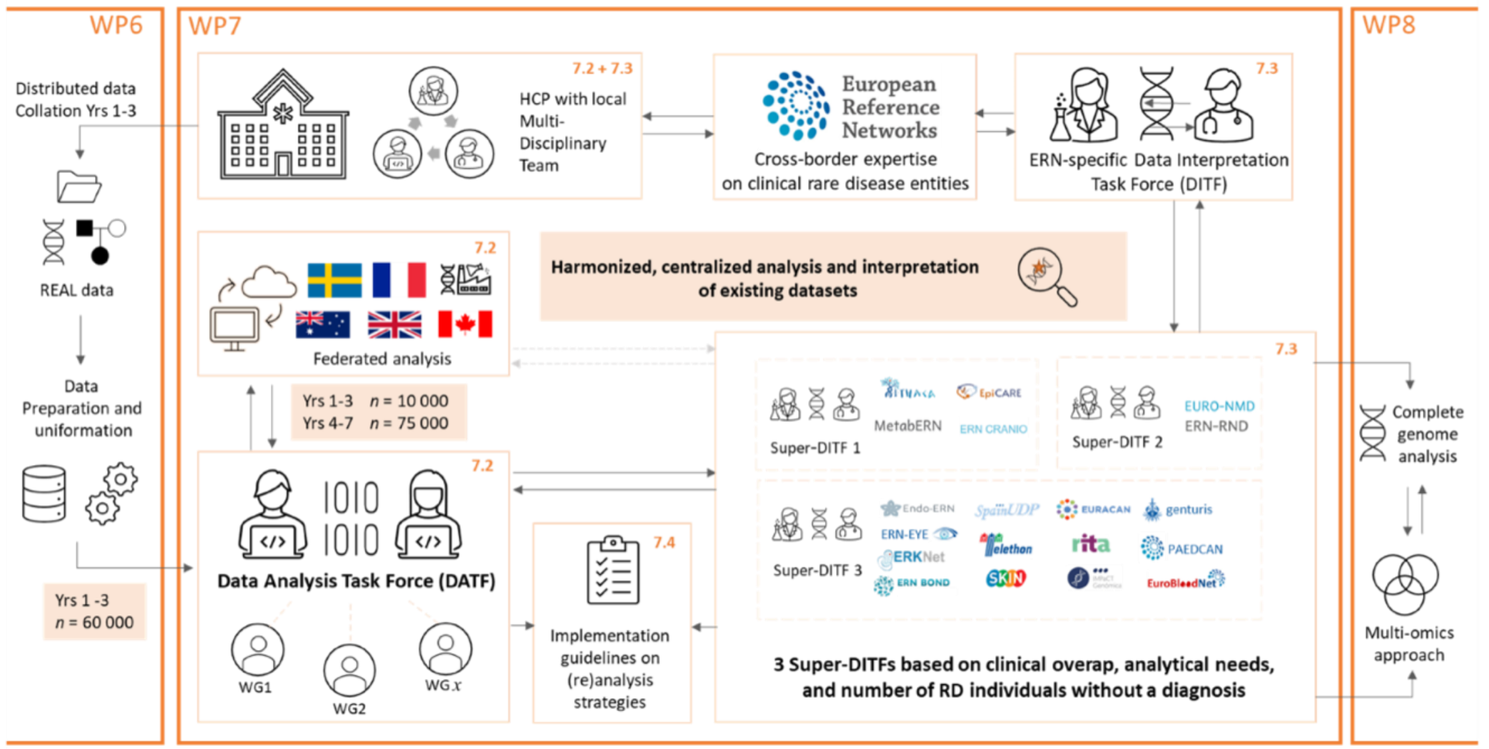
https://doi.org/10.1038/s41591-024-03420-w

Genomic reanalysis of a pan-European rare-disease resource yields new diagnoses

SolveRD
Solving the Unsolved Rare Diseases

8.4% new diagnoses
(6.3% for Euro NMD)

ERDERA Diagnostics workflow



CLINICAL RESEARCH NETWORK _ Outcome research

Real World Data

- **Task 9.1 –** Use of primary healthcare data (EHRs) for RD outcome research
- **Task 9.2 –** Use of population-based data for RD outcome research
- **Task 9.3 –** Integrating patient cohorts for natural history / standard-of-care reference studies
- **Task 9.4 –** Development of a blueprint and inventory of regulatory-grade natural history cohort data
- **Task 9.5 –** Disease progression modelling and prognostic biomarker research
- **Task 9.6 –** Development of a regulatory grade clinical trial simulation platform for rare diseases

Clinical Outcome Assessment

- **Task 10.1** – Platform for regulatory-grade patient-centred COA development and validation
- **Task 10.2** – Development and Implementation of Clinical Outcome Assessment Tools
- **Task 10.3** – Unveiling the Hidden Burden: Estimating the Socioeconomic Impact of Rare Diseases for Informed Decision Making and Resource Allocation

CRN: Outcome Research workstream _ Involvement of ERNs

	9.1	9.2	9.3	9.4	9.5	9.6	10.1	10.2
BOND			x					
CRANIO	x						x	
ENDO			x					
EpiCare	x	x					x*	x
ERKNet			x	x		x		x
ERNICA		x						
EURO-NMD / DDF	x				x	x	x*	x
RND				x	x		x*	x
EuroBloodNet	x			x	x		x	x
eUROGEN	x							
EYE				x			x*	
ITHACA			x				x	
MetabERN		x					x*	

* Mito-InterERN workgroup

CLINICAL RESEARCH NETWORK _ Innovative therapies

Advanced Therapeutic Medicinal Products

- **Task 11.1** – Identify and rank disease indications requiring ATMPs
- **Task 11.2** – Match technical development with prioritised needs
- **Task 11.3** – Elaborate PoC studies to test the development pipeline
- **Task 11.4** – Evaluate the selected platforms for clinical trials requirement and joint transnational call

N-of-few approach

- **12.1** – Academic Platform for Tailored Antisense Oligonucleotide Therapies
- **12.2** – Identification of patient relevant-outcomes (n-of-1/few) and run in natural history study
- **12.1** – Treatment/study design and analysis
- **12.1** – Implementation of first in human treatment infrastructure
- **12.1** – Case studies

Beyond ERDERA

ERDERA European Rare Diseases Research Alliance

FUNDING

ERA4Health Partnership

PerMed

European Directory of Registries (ERDRI.dor)

Central Metadata Repository (ERDRI.mdr)

ERDRI

Search Broker (ERDRI.sebro)

SPIDER

EOSC

The European Open Science Cloud

EOSC-Life

DATA SERVICE HUB

Global Alliance for Genomics & Health

Collaborate. Innovate. Accelerate.

DARWIN EU

ACCELERATION HUB

INNOVATIVE SME

innovative health initiative

EHDS

HealthData@EU Pilot

JARDIN

European Genomic Data Infrastructure

RARE DISEASE MOONSHOT

SCALING UP PUBLIC-PRIVATE PARTNERSHIPS TO ACCELERATE RESEARCH IN RARE DISEASES

eit Health

SCREEN 4CARE

THCS

TRANSFORMING HEALTH AND CARE SYSTEMS

EDUCATION & TRAINING

CLINICAL RESEARCH NETWORK

European Rare Diseases Research Alliance

Thank you!

Follow us

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