

ERDERA

European **Rare Diseases**
Research Alliance

European Rare Diseases Research Alliance

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INSERM, France

**2025 annual meeting of the European network of paediatric research
at EMA (Enpr-EMA)
19-20 November 2025, Amsterdam**

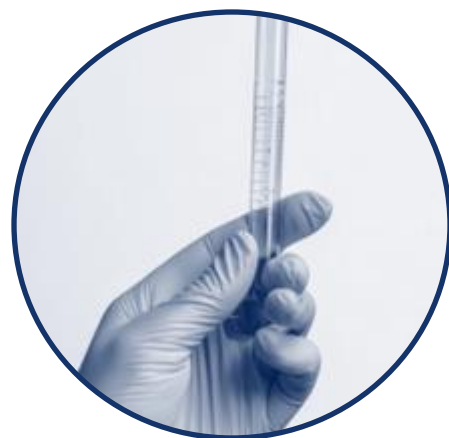


Co-funded by
the European Union

ERDERA has received funding from the European Union's Horizon Europe research and innovation programme under grant agreement N°101156595.

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**Improving the health and well-being
of 30 million people living with a rare disease
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 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: Our Global Scale & Investment



Diverse Partnership Network

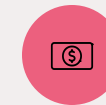
Our mission is powered by a broad network of nearly **180 organizations from 37 countries**, including:

- Funders
- Research Institutions
- Hospitals
- Research Infrastructures
- Patient Organizations
- Industry Partners



Extensive Global Reach

ERDERA spans **25 European Union countries** (except Croatia and Malta) and **numerous associated and non-EU countries**, fostering international cooperation (Australia, Canada, Iceland, Israel, Georgia, Morocco, New Zealand, Norway, Serbia, Switzerland, Türkiye, United Kingdom)

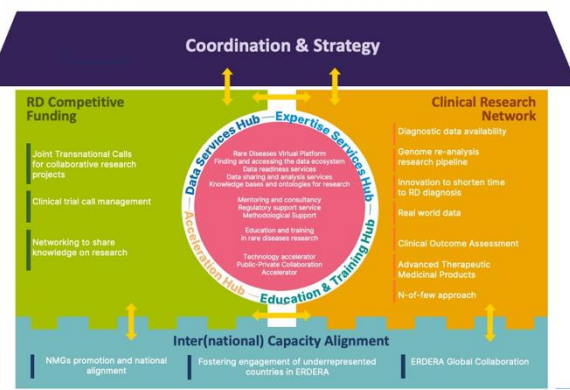


Substantial Financial Commitment

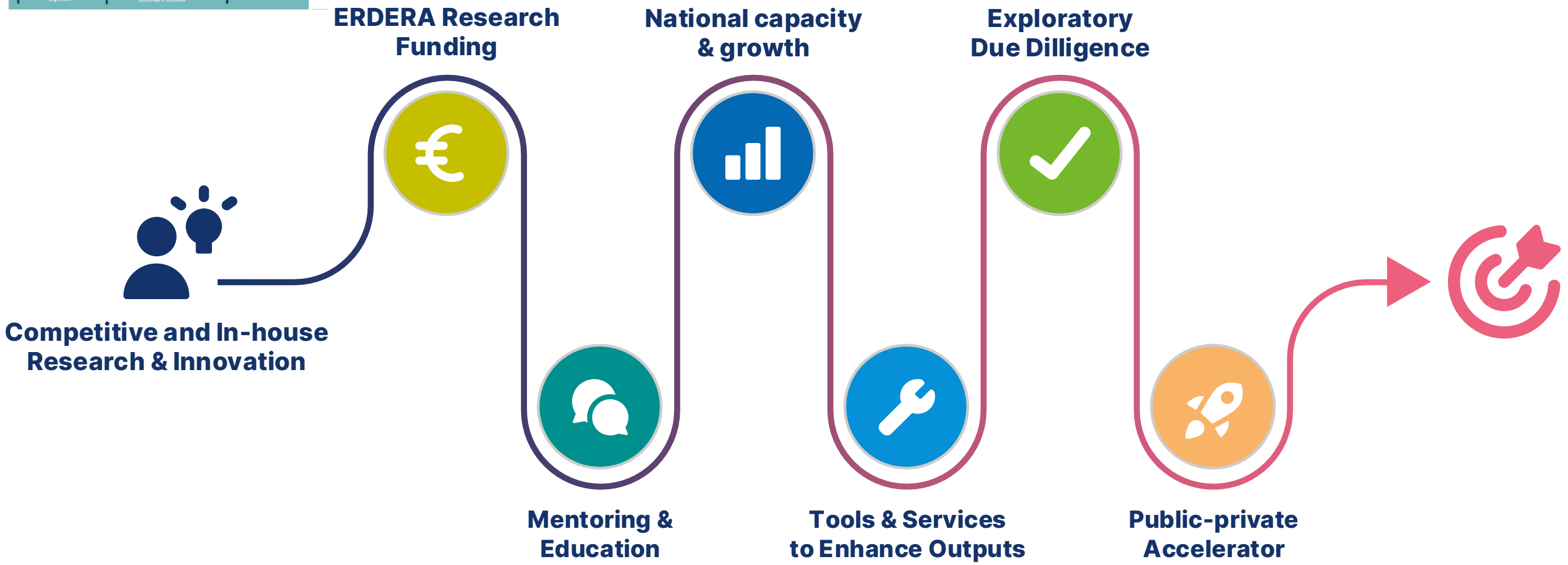
A significant financial commitment underpins our ambitious goals, with a total budget of approximately **€380 million**:

- **€150 million** investment from the European Commission
- Over €230 million contributed by participating countries and institutions
- **Every partner contributes** to our collective actions

ERDERA brings together diverse expertise and significant resources to advance its mission, demonstrating a strong commitment to global collaboration.



ERDERA's research & innovation support cycle



ERDERA in action

Competitive funding & support services

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



Call for Proposals 2026

Resolving unsolved cases in rare genetic and non-genetic diseases through variant validation and new technological approaches

→ Launch 10 December

✍ Pre-announcement now available

📅 Webinar:
16 December,
15:00–17:00 CET

Networking Support Scheme

A competitive funding instrument for networking events to share knowledge on rare diseases and rare cancers



Proposal round number 2

The current submission round ends on 7 April 2026 at 14:00 (CET)

161
Days

01
Hours

Funding: 30 000 € per event

**Application: Continuously open
(eligibility check every 6 months)**

**Eligibility: Collaborative
(minimum 3 applicants from 3 ERDERA countries)**

Program Goals

- Provides financial support for organizing workshops or conferences, fostering new or expanding research networks.
- Aims to strengthen collaborations and enable the exchange of knowledge.
- Build new or expand existing European and global research networks focusing on Rare Diseases (RDs) and rare cancers.

Clinical Trials Call

Clinical Trials Call funds controlled clinical research studies undertaken in humans to establish or confirm the safety and effectiveness of therapeutic interventions. These will benefit from ERDERA support for regulatory and methodological aspects.

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

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.

Driven by patients' needs from inception



Clinical Trials Call

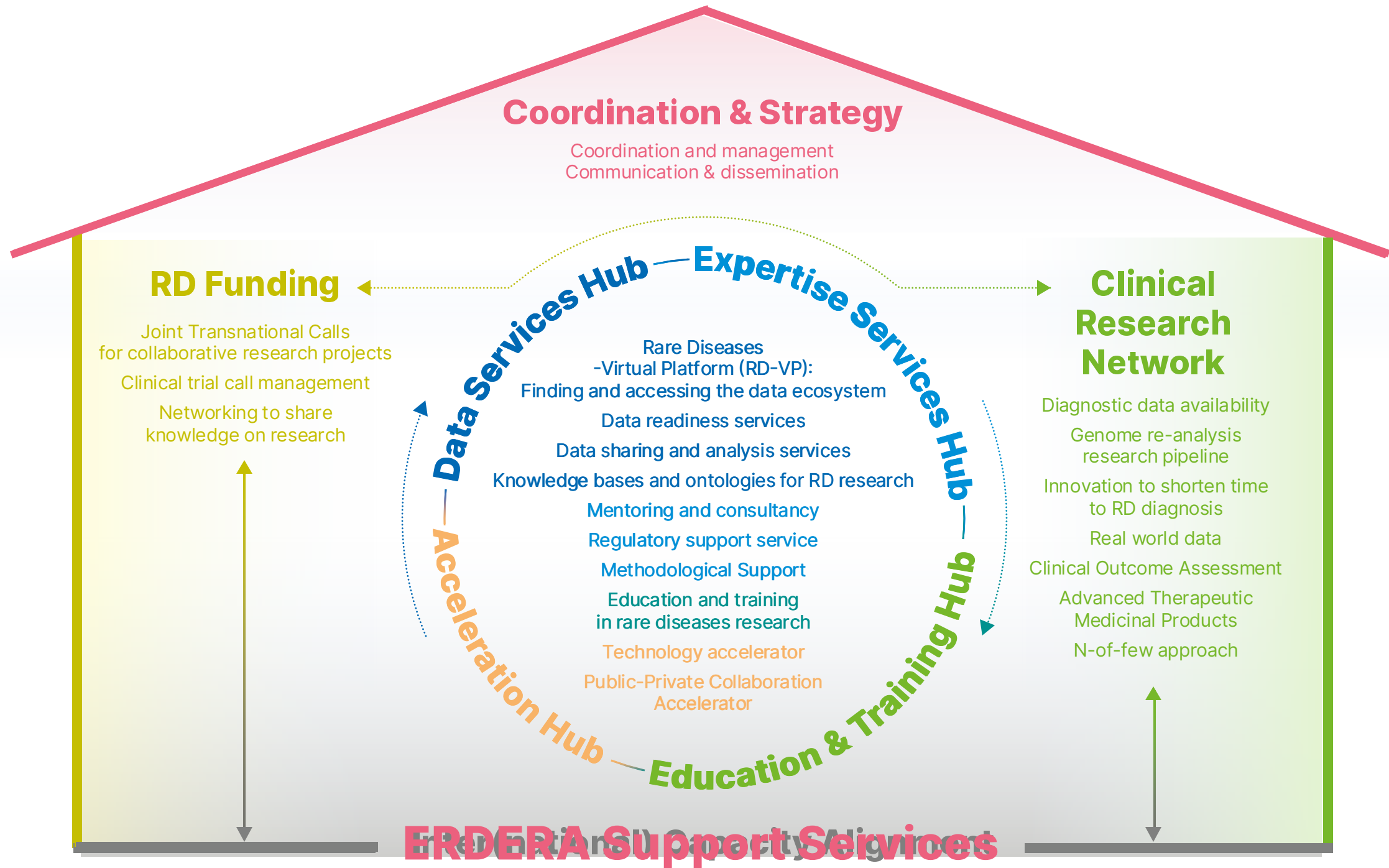
**SHORT GUIDE
ON PATIENT PARTNERSHIPS
IN RARE DISEASE
RESEARCH PROJECTS**

BASIC
PRE-CLINICAL
TRANSLATIONAL & SOCIAL

Written by the members
of the working group PEIRREP*
Guide first
published in July 2020
on www.ejprarediseases.org

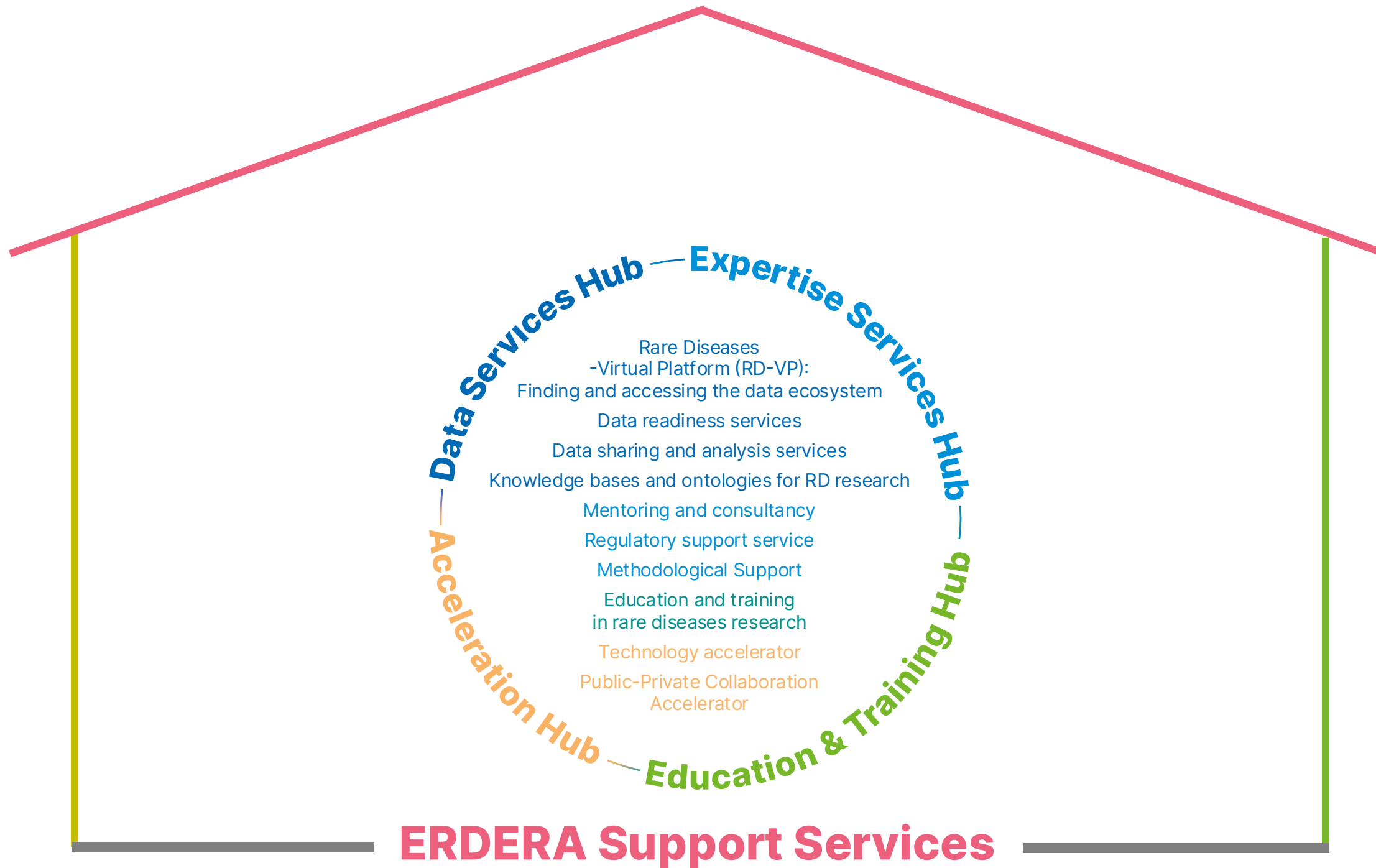
* Patient Engagement in
Biomedical Research Projects.





ERDERA Support Services

NMGs promotion and national alignment
Fostering engagement of underrepresented countries in ERDERA
ERDERA Global Collaboration



The Virtual Platform (VP) is a growing **network of Findable, Accessible, Interoperable and Reusable (FAIR) resources, ready to serve the rare disease (RD) research community**. It includes catalogues of over 45 resources, registries, biobanks, knowledge bases and tools compliant with agreed standards. The VP Portal allows you to search the VP network resources at once in real time to find those of interest to your research.

Discover rare diseases resources and data:

Search for a disease name (e.g. ADPKD), gene (e.g. PKD1), or Orph...



Patient Reported Outcome Measures (PROMs)

1 PROMs repository has been updated within **PROQOLID**, and a **user-friendly QALY interface** is under development. Currently, **1,200 rare disease-specific PROMs** are included.

Early Detection and Prenatal Screening

2 An **ontology of prenatal echography signs** is being developed to improve the **early detection of rare diseases** and harmonize data interpretation across clinical settings.

Functional Impact and Disease Burden

3 An **ontology describing the functional impact of rare diseases** has been developed and is being prepared for **public release**, supporting research, clinical assessment, and policy design.

Expertise Service Hub



MENTORING & CONSULTANCY

Mentoring Programme:

32 projects accompanied with over 30 mentors, handbook & tracking platform

Consultancy service: encompassing overall expertise available in ERDERA tailored to users needs

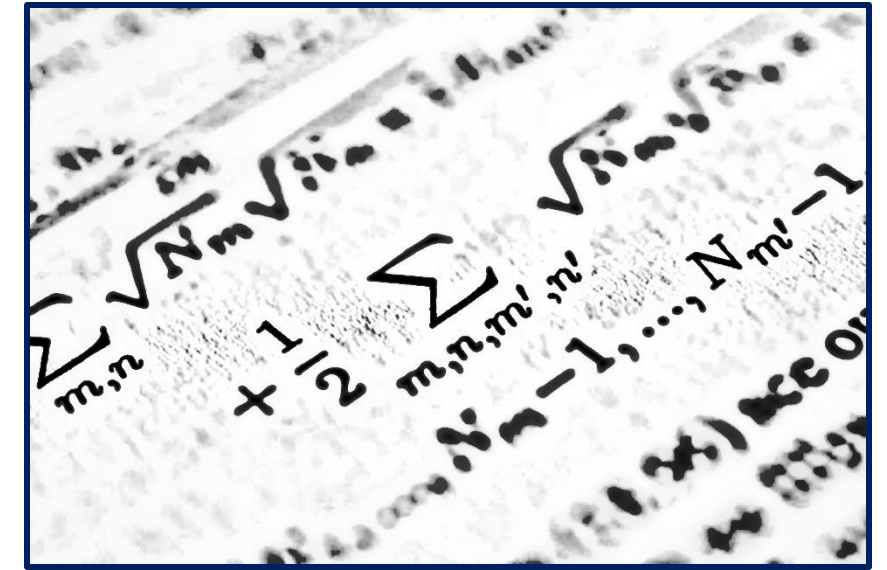


REGULATORY

Regulatory Support Group:

20 members, representing regulators, academia, industry, patients, non-for profit

Guidance on: regulatory requirement for pre-clinical & clinical research, data use, privacy, samples, etc.



METHODOLOGY

Clinical studies support:

Methodologies for multivariate, hierarchical, incomplete data and federated data analysis

RD trial methodology with initiation of surrogate endpoints evaluation

Handling missing data

Public-Private Collaboration Accelerator (PPCA)



The **ERDERA Public-Private Collaboration Accelerator (PPCA)** empowers researchers to turn promising translational ideas into investor-ready projects able to improve the lives of people living with rare diseases. By combining tailored scientific guidance with access to a unique European ecosystem of public and private partners, we help you de-risk and advance your work at the discovery and pre-clinical stages.

**Official Opening
for submissions
on January 2026**

TRAINING & EDUCATION



Diagnosing & Treating Rare Diseases

Diagnosing Rare Diseases: from the Clinic to Research and back – **1,021 participants**

Innovative Therapies and Personalised Medicine for Rare Diseases – **289 participants**

From Lab to Clinic: Translational Research for Rare Diseases – **190 participants**

Health Data, Ethics & Regulatory Frameworks – **111 participants**



FAIR Data Training

Understanding & value of FAIR

FAIR Guiding principles

Advanced concepts & Tools

300 participants, XX countries



Paediatric Training

27 young participants (ages 12–18)

From **7 Young Persons' Advisory Groups (YPAGs)**: Portugal, UK, Albania, Italy, Greece, Ireland



EURORDIS Open Academy

4-day international training with **two tracks**: *Medicines R&D* & *Scientific Innovation & Translational Research*

70 participants (60 patient advocates, 10 researchers)

31 countries, >52 rare diseases

15 expert faculty members delivering interactive sessions



Diagnostic Research

3 focused training sessions – **~400 participants** total

Submitting genomic and clinical data for re-analysis – **133 participants, 26 countries**

Clinical annotation using HPOs – **114 participants**

Advancing diagnostics in underrepresented countries – **150 participants, 30 countries**

2400 stakeholders trained

Clinical Research Network

Diagnostics, Outcome Research, Therapies



Accelerating Rare Disease Diagnosis & Therapies Across Europe



EU diagnostic pipeline

Robust Governance Framework to ensure high data quality and **coordinated access** (aligned with EHDS)

10 000 genomic data sets from patients across Europe with undiagnosed rare diseases

Leveraged **Long-Read Sequencing and RNA-seq** to **empower underrepresented countries** with advanced diagnostic capabilities



Electronic Health Records & Regulatory-grade Modelling

Practical **EHR→research workflow** defined and piloted (governance, queries, validation path).

First **regulatory-grade modelling result**: in genetic ataxias, adding digital motor measures to clinician scales yielded **~49–61% trial sample-size reduction** in simulations.



RD Therapies Pipeline

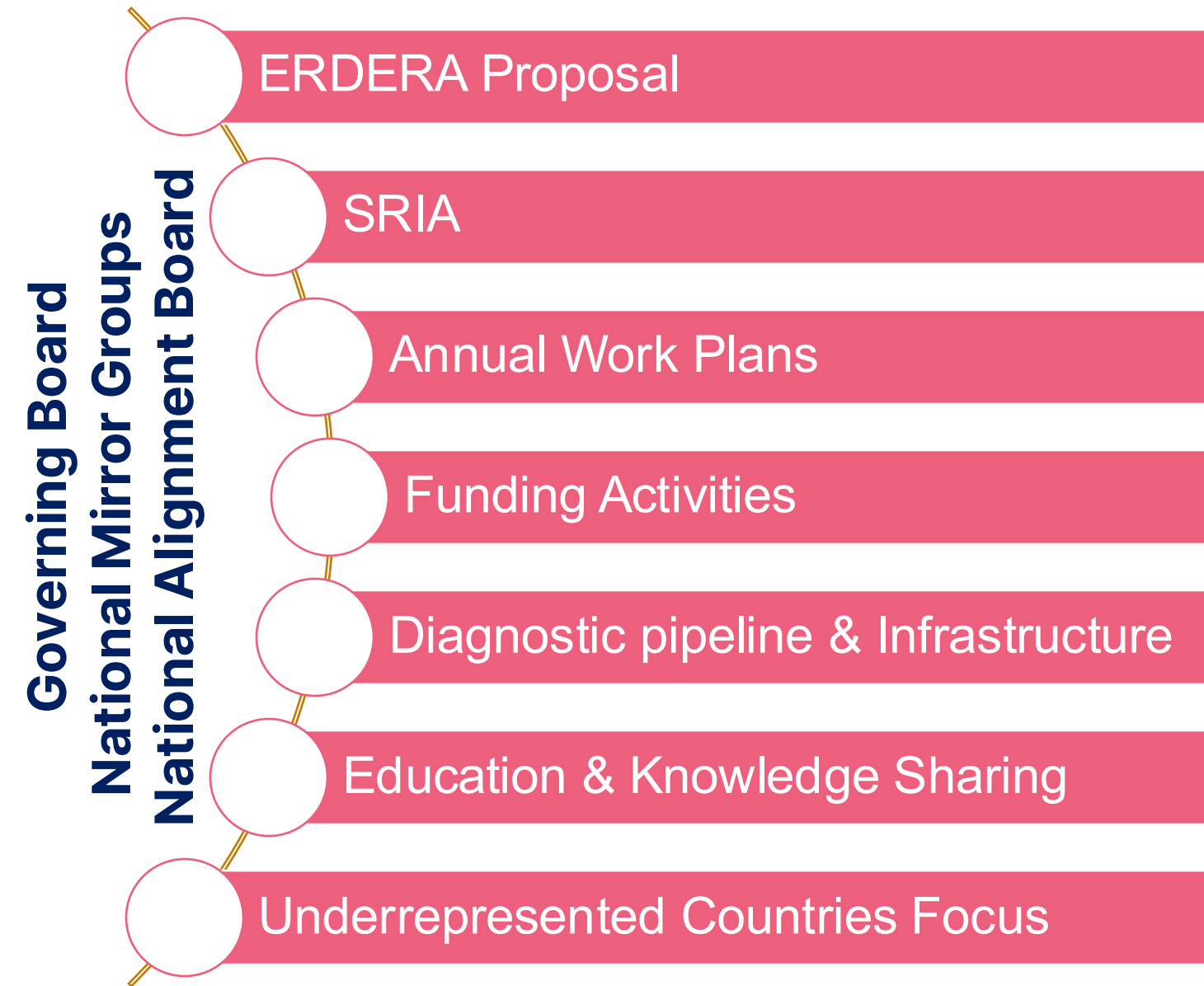
Developed a **transparent, evidence-based prioritisation framework** to guide ATMPs therapy development

Creation of a **matching tool** linking **rare disease categories with ATMP technology platforms**



Inter(national) Alignment

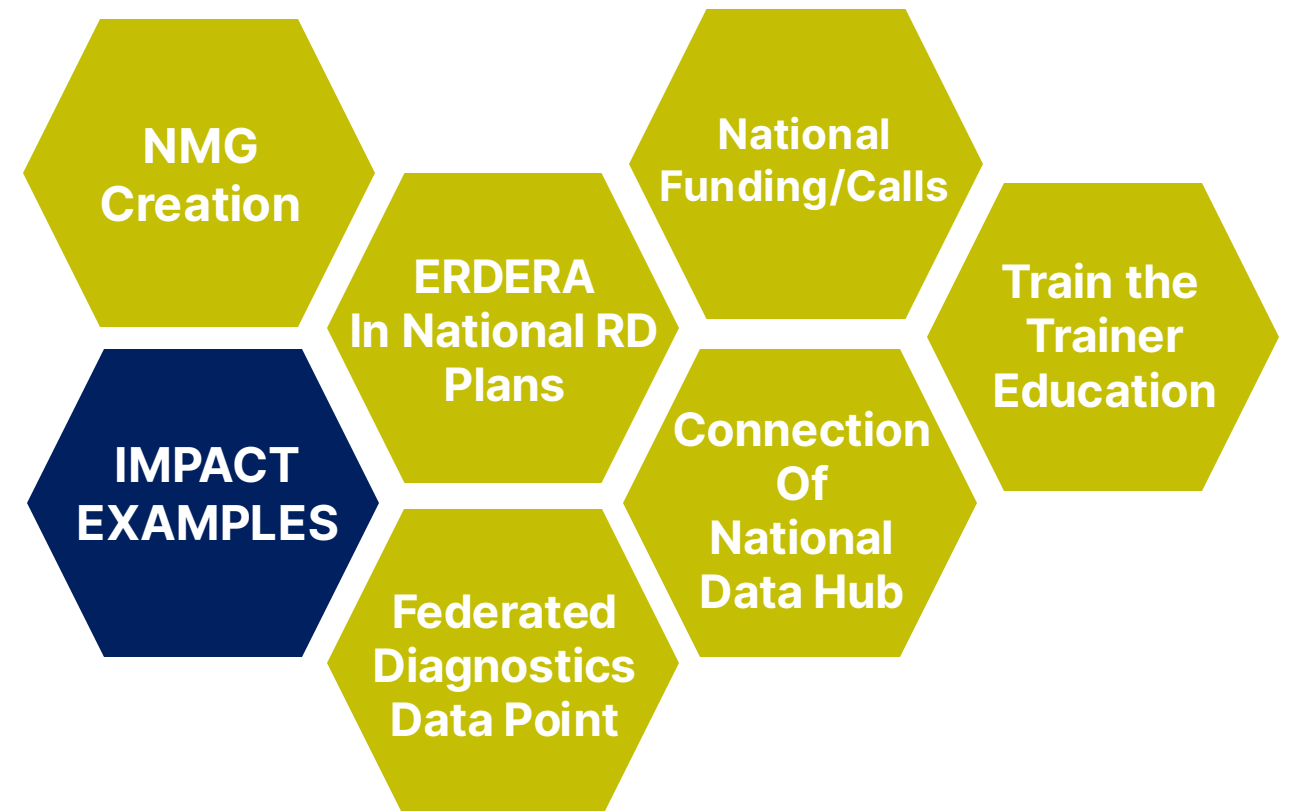
National Alignment



**18 NMGs established – 13 ongoing
37 as target for end of year 2**

NMGs COMPOSITION:

ERDERA GB Representative, National Competent Authority, National Funding Body, Academia, Hospitals, National RD Patients Alliance, Industry, HTA/Payers, ERNs



Strategic Alliances



**Joint Action on
Integration of ERNs into
National Healthcare
Systems**



**European
Genomic Data
Infrastructure**

**Enabling access to
genomic and related
phenotypic and clinical
data across Europe**



**Global Alliance
for Genomics & Health**
Collaborate. Innovate. Accelerate.

**Technical standards,
policy frameworks and
tools that expand
responsible, voluntary,
and secure use of
genomic and other
related health data**



**Shortening the path to
rare disease diagnosis
by using newborn
genetic screening and
digital technologies**

To the Brilliant Minds and Big Hearts Behind ERDERA Coo



Daria Julkowska
Coordinator



Anissa Bounabi
Assistant
Coordinator



Baptiste Eluard
Project Manager
RD fundings-
Expertise
Services Hub



Pauline Adam
Project Manager
Global
collaborations



**Clément
Moreau**
Project
Manager
National
Alignment



**Başak
Uysal**
Project Manager
Data Hub



**Raisa
Simoes**
Policy Officer



**Gustavo
Insaurralde**
Financial &
Administrative
Manager



Odile Heitz
Office Manager



Maica Llaveró
Co-head of EU-
Funded Projects



Anna Llobet
Project
Manager



Marina Derch
Project
Manager



Gisela Pairo
Communication
Lead



Freya Sentimarti
Junior Communication
Officer



Jordi Vaqué
Communication
Officer

Join ERDERA's Open Session

Join the free and online public session on 30th of October

[VIEW MORE](#)

ERDERA Funding

The ERDERA Networking Support Scheme
(NSS) Call **is open!**

[CLICK HERE](#)

www.erdera.org

ERD ERA

European Rare Diseases
Research Alliance



Thank you!

Follow us



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