ERDERA

European Rare Diseases
Research Alliance

ERDERA: Connecting Rare Disease Research, Data and Biobanking Across Europe

Connecting the dots in the rare diseases research ecosystem

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Rare diseases and biobanks in Europe: key facts

Individually rare, collectively common: reusing samples and data speeds up diagnosis

1 in 2,000
EU definition: no more than 5 people in 10,000

6,000+
rare diseases (Orphanet); 72% genetic, 70% paediatric onset

~30 M
people in Europe live with a rare disease (263–446 M worldwide)

~500/~3,700
biobanks / collections listed by BBMRI-ERIC

506
families diagnosed by reanalysing existing data (Solve-RD)

10,000
genomic datasets from undiagnosed cases, ERDERA year 1

Rare disease projects

Connect4children
Pediatric trial network
IMI2 · 2018–24

Screen4Care
Newborn screening + AI
IMI2 · 2021 · €25M

PaLaDIn
Rare neuromuscular data
IHI · 2024 · €21M

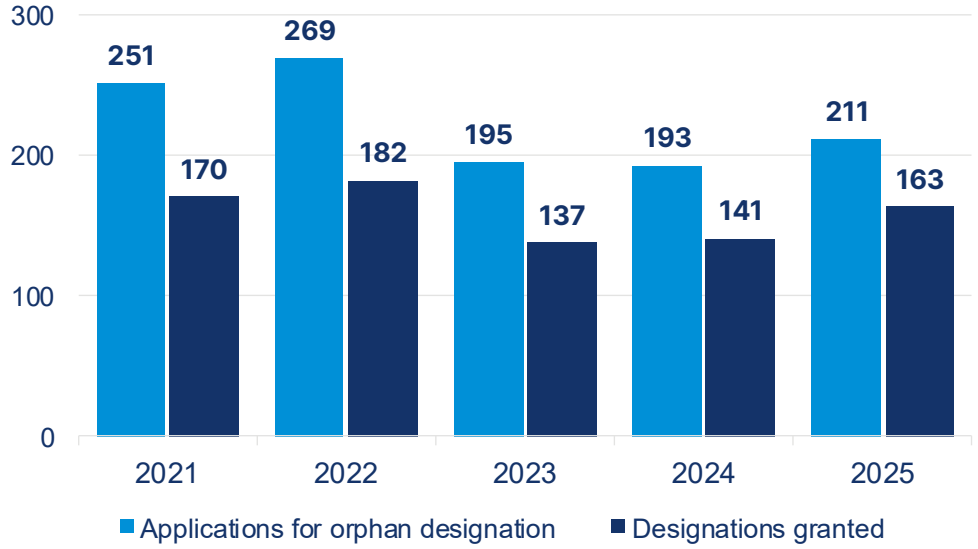
RealiseD
Ultra-rare disease trials
IHI · 2025 · €17M

Sources: EU Orphan Regulation (EC) 141/2000; Nguengang Wakap et al., Eur J Hum Genet 2020 (Orphanet); ERDERA; BBMRI-ERIC / ESFRI RI Portfolio 2024; Laurie et al., Nat Med 2025; IMI and IHI project websites and launch releases.

ERDERA European Rare Diseases Research Alliance

EU orphan medicines: a rebound in 2025

Applications dipped in 2023–24 and recovered in 2025; about 18 authorisations a year



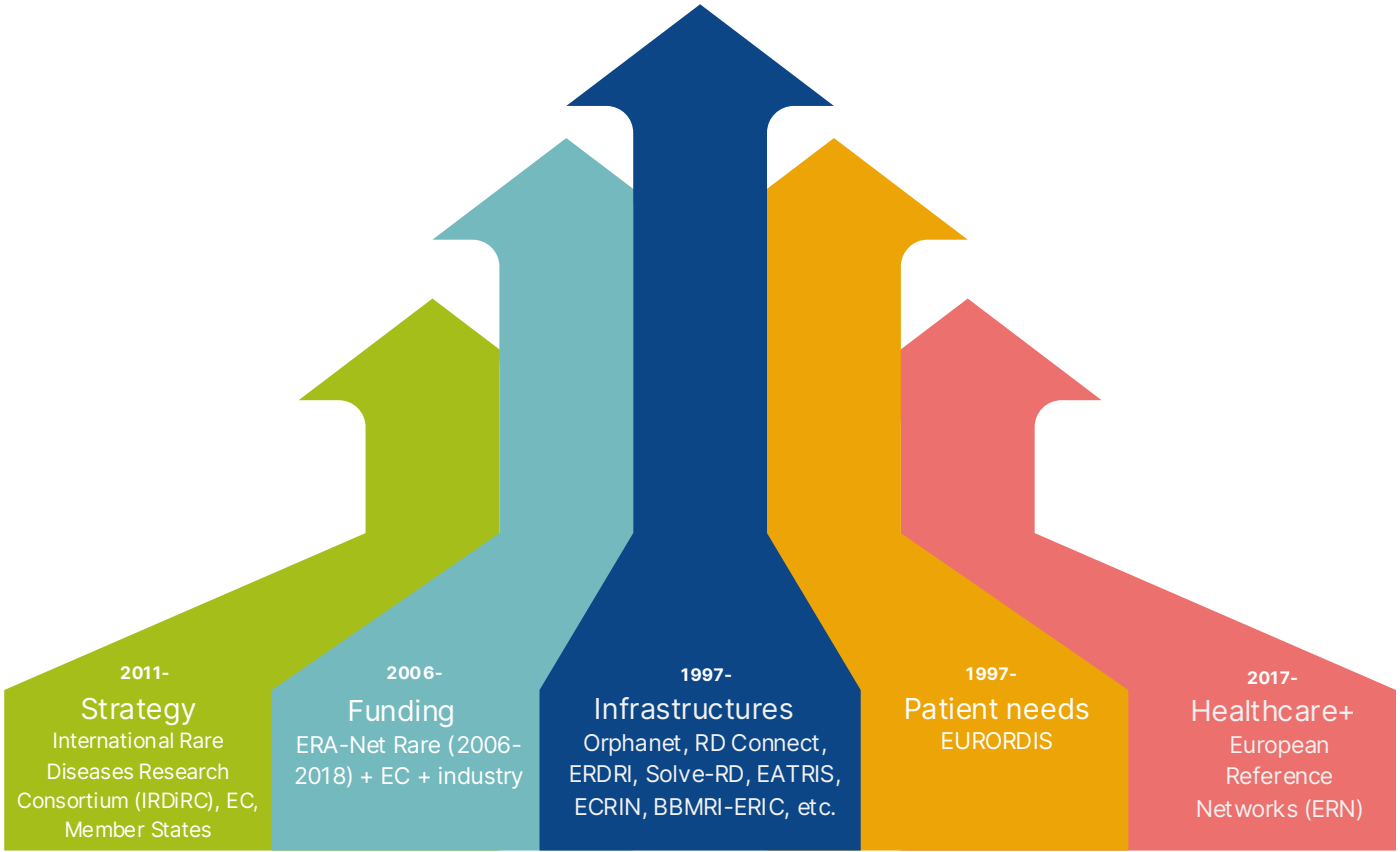
211
applications in 2025 (+9% vs 2024); 15% advanced therapies

163
designations granted in 2025 (+16% vs 2024)

17
orphan medicines newly authorised in 2025; 278 since 2000

Source: EMA, Annual report on the use of the special contribution for orphan medicinal products, Year 2025 (EMA/375104/2025, January 2026): Table 2 and Figure 2. Percentage changes calculated from the table.

Towards a comprehensive RD Research Ecosystem



Co-funded by the European Union

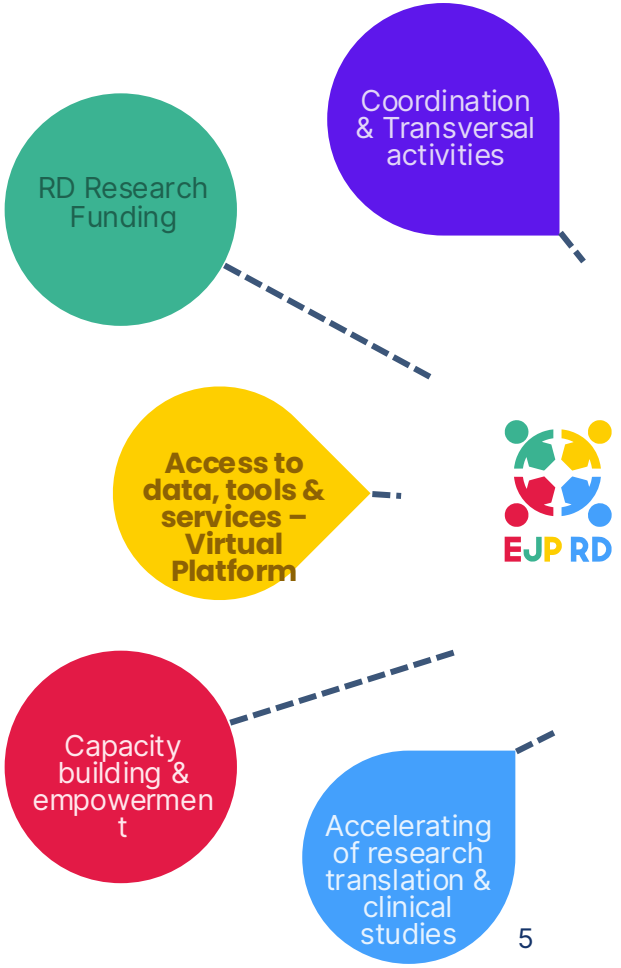
The EJP RD initiative has received funding from the European Union's Horizon 2020 research & innovation programme under grant agreement N°825575

Main objective

Create a research and innovation pipeline "from bench to bedside" ensuring rapid translation of research results into clinical applications and uptake in healthcare for the benefit of patients

Mode of action:

Large programme that integrates existing infrastructures, trainings, funding programmes and tools, expands them and develops new essential ones to offer harmonized (and centralized) RD research ecosystem that is easy to use for scientists and produces benefits for patients in the most efficient way



What is EJP RD



+2300 People

35 Participating countries

101 M€ Budget

130 Institutions

All 24 ERNs

EJP RD activities and achievements

Innovation Management Toolbox (511 resources)

Collaborative Research Funding

RD Virtual Platform

Training and empowerment

Accelerated Translation of Research Results and Clinical Studies

207 projects funded through the JTC

69 supported events through NSS

3 projects funded through RDR

More than 10 000 stakeholders trained

RD **data management & quality** (968)

Patient trainings (425)

28 **inter-ERN workshop**

91 **ERN fellowships** attributed (77 completed)

3 MOOCs (diagnosis, translational research, data & ethics)

7 769 trainees from 150 countries)

RD Virtual Platform

(https://vp.ejprarediseases.org)

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

Over +**100 biological pathways** created

104 M € invested in collaborative research projects (2019-2023)

Expanding beyond EJP RD

Increased scope

EUROPEAN JOINT PROGRAMME RARE DISEASES --> ERDERA European Rare Diseases Research Alliance

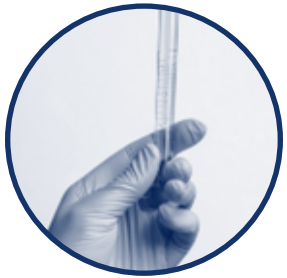
Pathway to impact

- Accelerated diagnosis
- New effective therapies approved
- Better understanding of rare disease impact

Co-funded by the European Union

Connecting the dots

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

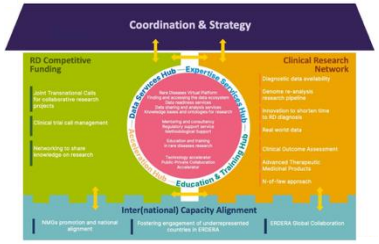
ERDERA spans **25 European Union countries** (except Croatia and Malta) and **numerous associated and non-EU countries**, fostering international cooperation (Australia, Canada, Iceland, Israel, Georgia, Morocco, New Zealand, Norway, Serbia, Switzerland, Türkiye, United Kingdom)

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

Competitive and In-house Research & Innovation

ERDERA Research Funding

Mentoring & Education

National capacity & growth

Tools & Services to Enhance Outputs

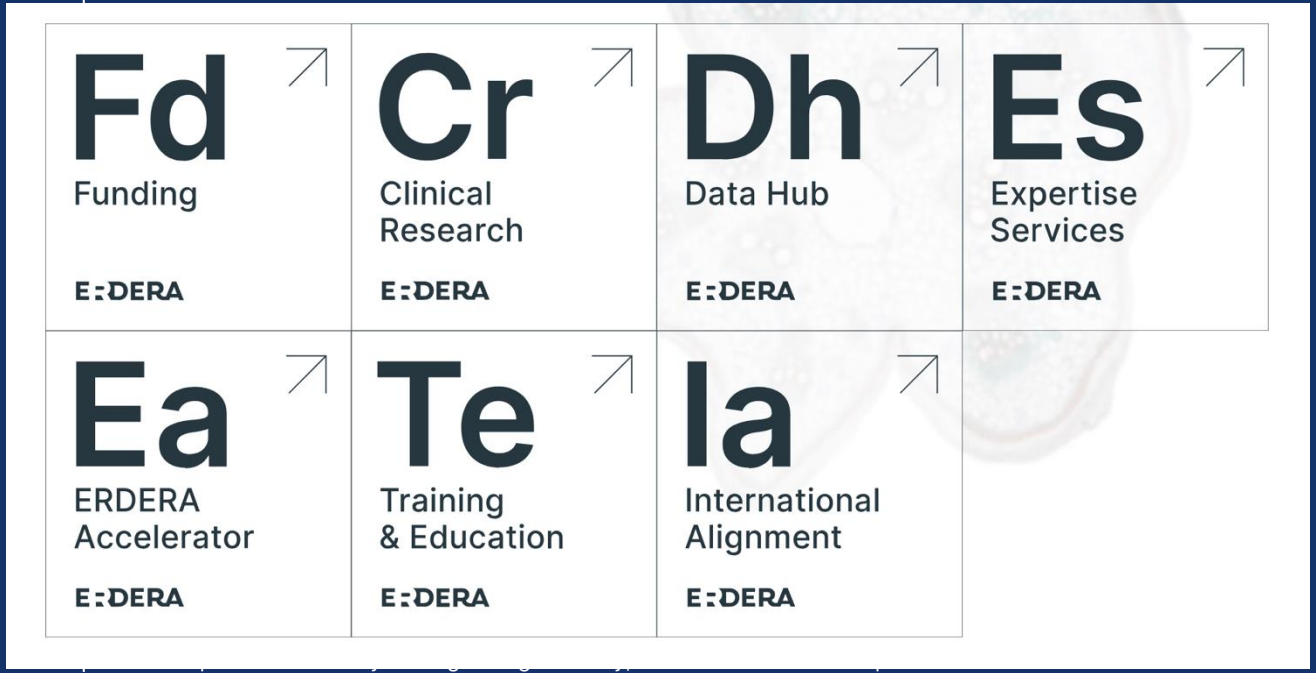
Exploratory Due Dilligence

Public-private Accelerator

ERDERA in action

European Rare Diseases Research Alliance

Research Services



Clinical Research Network

Diagnostics, Outcome Research, Therapies

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



Data Service Hub

Virtual Platform (VP)

FAIR by design

Find patients with particular condition in multiple sites

Machine-Readable Consent and Access conditions

Re-usable Data for clinical trials optimisation (e.g., external control arms, natural history studies)



https://www.ejprarediseases.org/wp-content/uploads/2021/10/EJPRD_P2_AD36_PU_Virtual-Platform-Specification.pdf

ERDERA Data Hub & EJP RD Virtual Platform

1143 resources connected (registries, catalogues, gen-phen deposition infras, knowledge bases, projects) (186 biobanks)

Over +**100 biological pathways** created

3 linkage/discoverability levels

https://vp.erdera.org/

RD VIRTUAL PLATFORM



Data (re)analysis

FAIR stewardship

Computing infrastructure

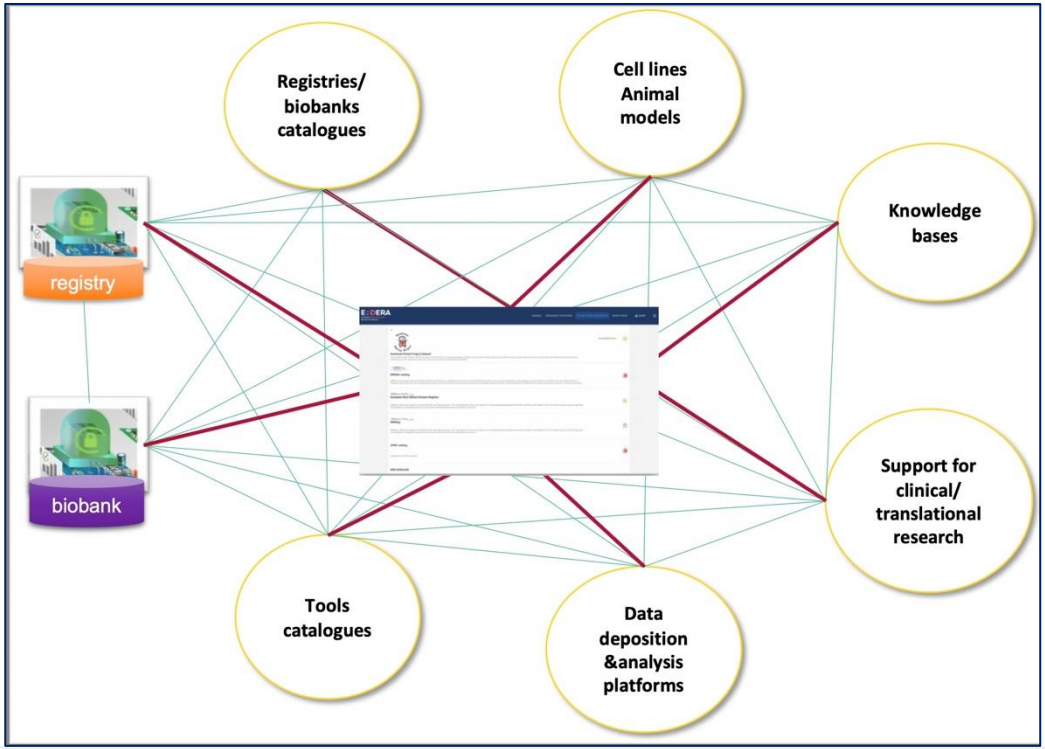
RD DATA HUB SERVICE (DSH)

DSH= A **service-oriented** ecosystem of solutions of which ERDERA partners are the **co-creators**

ERDERA European Rare Diseases Research Alliance

EJP RD

The RD-Virtual Platform curated ecosystem



https://vp.erdera.org/

ERDERA European Rare Diseases Research Alliance

Decreasing fragmentation

Increasing interoperability

Increasing RD-readiness

through

Harmonisation

Standardisation

in a flexible way:

Common methodologies, Multiple technical solutions

BBMRI IS A DISTRIBUTED ERIC

Operating in health and life sciences

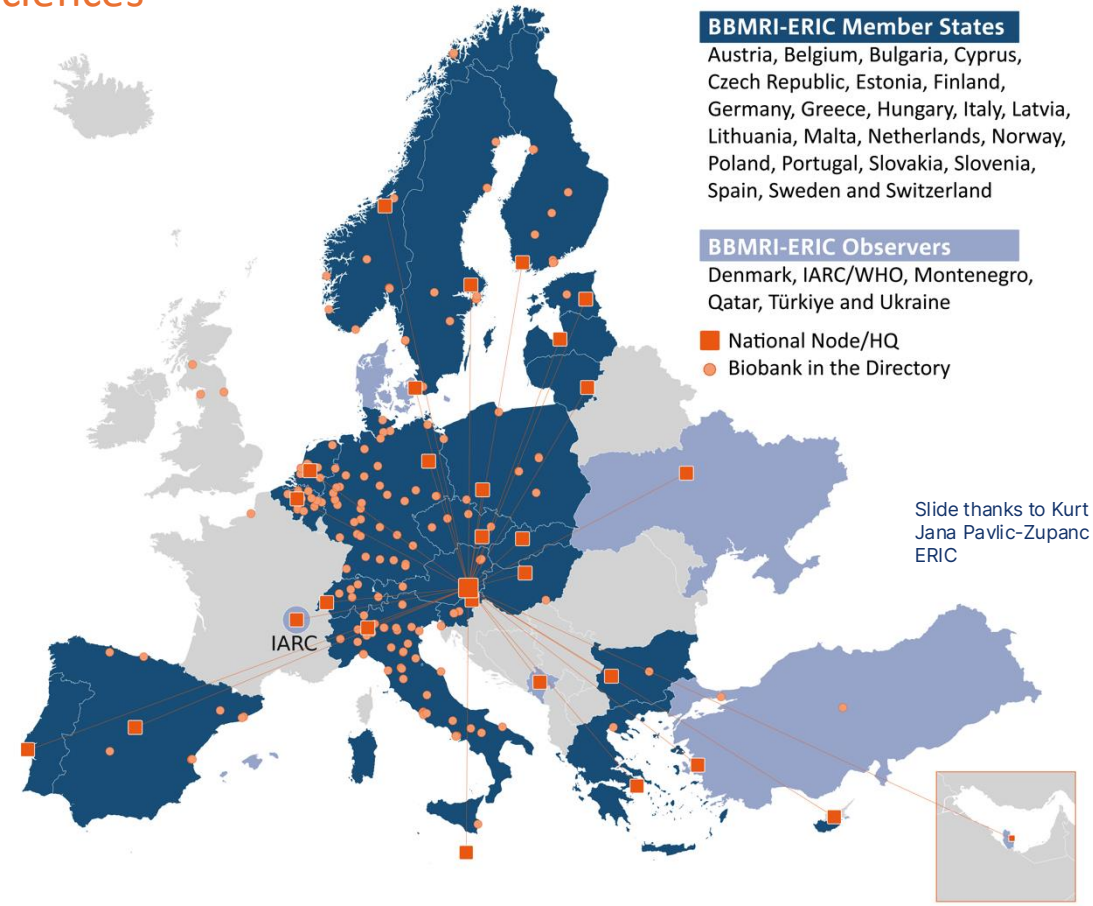
BBMRI-ERIC®
The European research infrastructure for biobanking and biomolecular resources in health and life sciences

THE BBMRI COMMUNITY

- **28** National Nodes + IARC
- **~ 500** biobanks across 33 countries
- **32** ELSI experts
- **130** Information Technology experts
- **313** Quality Management experts
- **3** Biotechnology Expert Centres
- **1** Headquarter

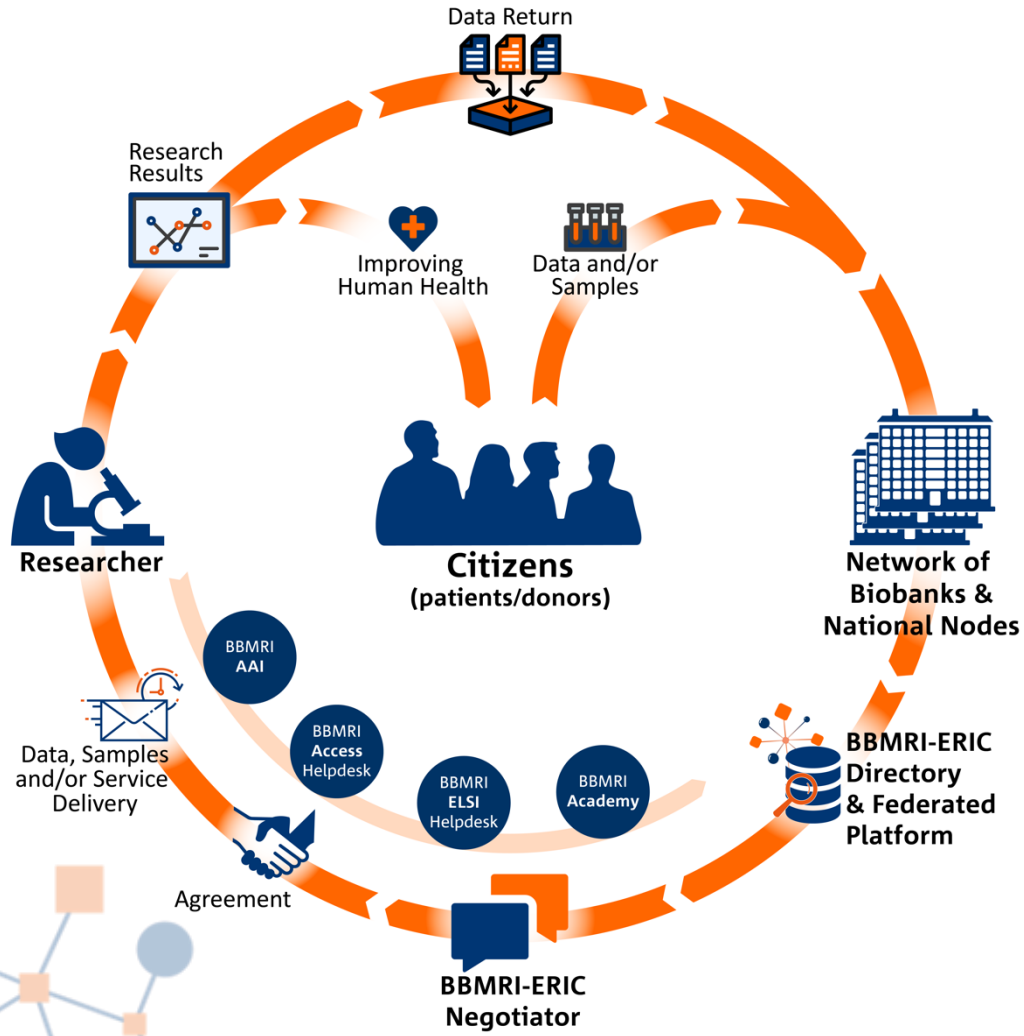
SERVING SOCIETY, ACADEMIA AND INDUSTRY ALSO AS DATA INFRASTRUCTURE

Orange square= where biobanks are located.



Slide thanks to Kurt Majcen and Jana Pavlic-Zupanc @BBMRI-ERIC

BBMRI-ERIC®
biobanking for a healthier world



BBMRI-ERIC's MIABIS* FAIR Data include:

- >8k harmonised OMOP variables of clinical phenotype data with biochemical measurements,
- >10PB whole slide or radiology image data,
- OMICs data (> 155K genomes, proteomics, metabolomics etc.),
- National Registries connections with e.g. survival data and
- other data types incl. surveys, genealogy, patient-provided data.

*MIABIS: Minimum Information ABoutBIobanking data Sharing

Training & Education in:

BBMRI-ERIC® *Academy*

- ELSI
- IT
- QM
- Biobank Development
- Health & Life Science Research/Development
- Outreach/Engagement

BBMRI-ERIC Directory is largest biobank catalogue worldwide:

- ~500 biobanks
- 3,768 collections
- 33 countries.

BBMRI-ERIC Federated Platform features:

- 460,000 donors with
- 155,000 genomes across
- 10 EU countries today.

ERDERA Virtual Platform – BBMRI-ERIC Connection

Discoverable BBMRI-ERIC Directory collections connected to the Virtual Platform via L1/L2 connectors.

1

BBMRI offers FAIR data points. This allows querying BBMRI biobanks in the VP.

2

Normally, biobanks are included in BBMRI when they are a member state, except for RD. Also, OPRHA codes are represented (when available).

3

Accessible
Federated, GDPR-compliant query with machine-readable consent conditions.

Expertise Services

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

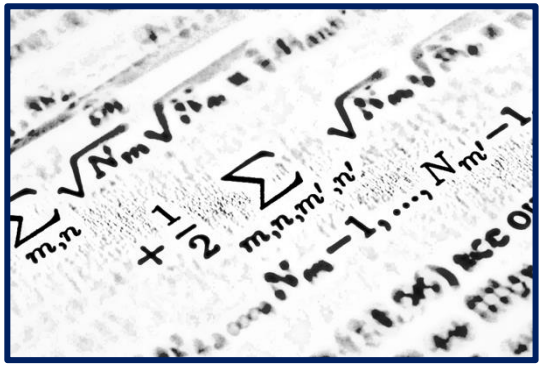


REGULATORY

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

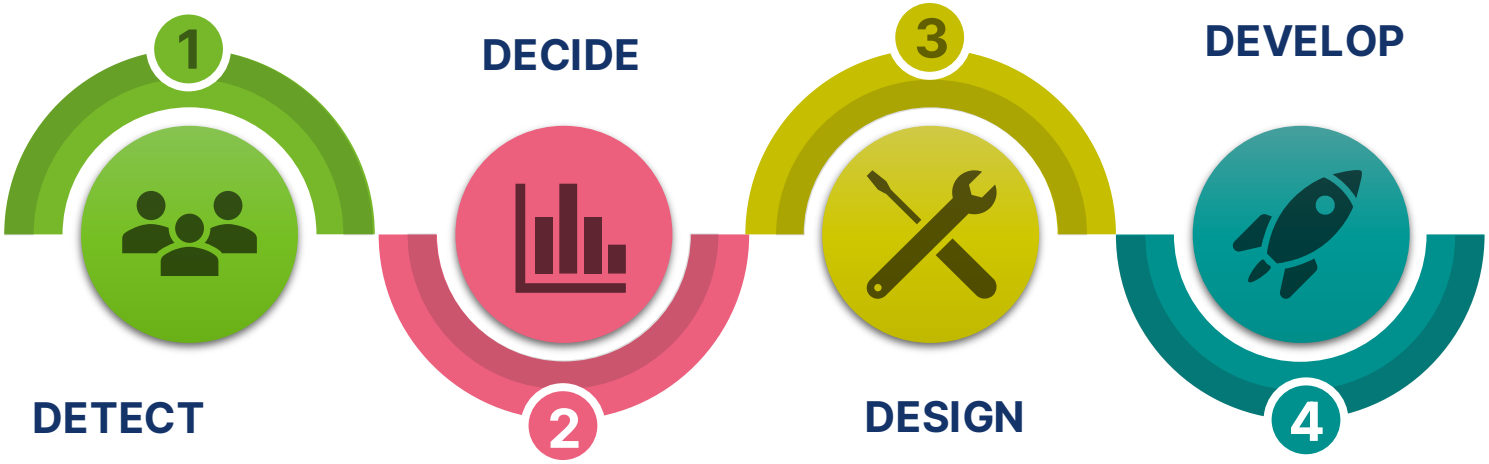
RD trial methodology with initiation of surrogate endpoints evaluation

Handling missing data

ERDERA Accelerator

Public-Private Partnerships Accelerator

Acceleration Hub is both a **translational** and **public-private collaboration** accelerator Hub in **rare diseases**



Projects **sourcing**

ERDERA and EJP RD JTC External sources

Strong translational potential

Investor mapping

Projects **selection** & eligibility

- Solid scientific rationale
- Clear unmet medical need
- Project feasibility

Investor-oriented

Support Services Offered

- Translational mentorship
- IP & Regulatory support
- Data management

Investor-ready

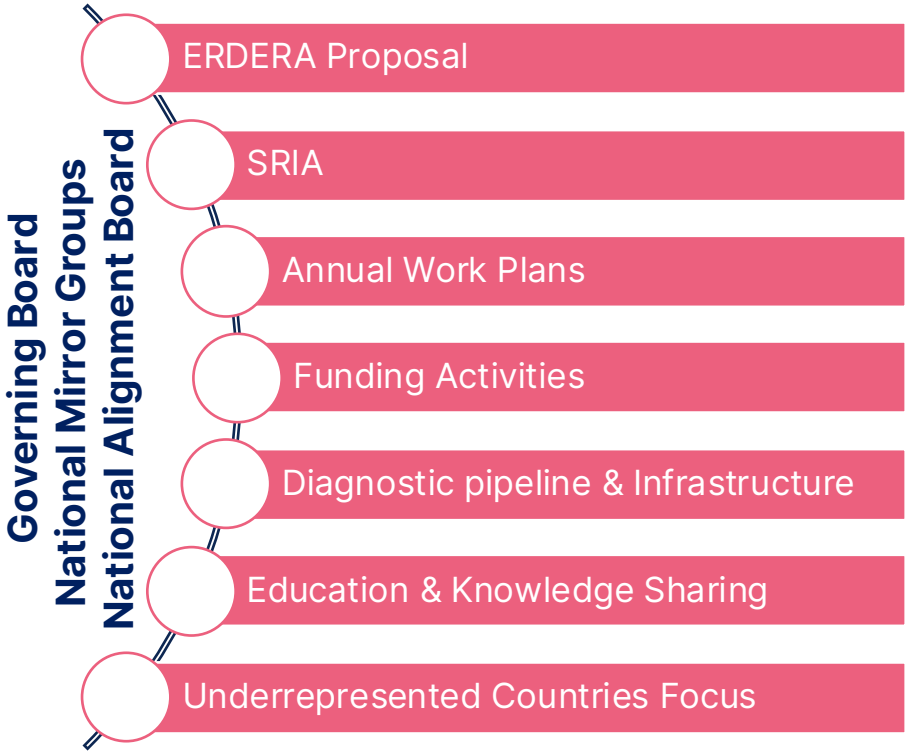
Advancing projects towards IND-enabling studies

- Investor search
- Investor roadshow

Match with investors and industry interests

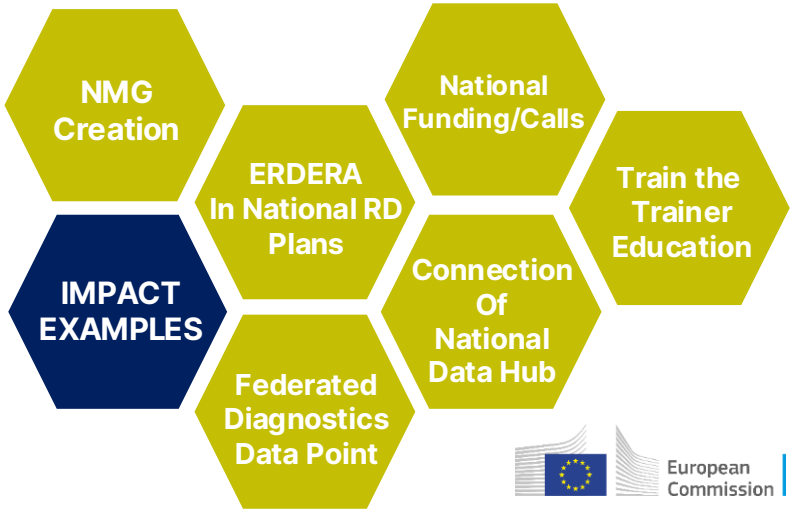
Inter(national) Alignment

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

JARDIN

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

SCREEN 4CARE

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

Competitive funding & support services

What are the opportunities of collaboration that ERDERA can offer?



Yearly Joint Transnational Calls

These calls support **multinational research projects** that are of broad scope. Topics include improving diagnosis, developing new therapies, and understanding the social impact of rare diseases.



Networking Support Scheme

This scheme offers **small grants** (30 000 €) **to support events or workshops** aimed at fostering knowledge sharing and community building.



Multinational Clinical Trials Call

A call for **multinational clinical trials** is expected in 2026.

Training& Education

TRAINING & EDUCATION

2400 stakeholders trained. Mainly MOOCs

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, 58 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

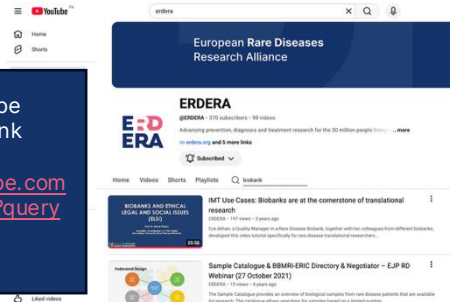
Submitting genomic and clinical data for re-analysis – **133 participants, 26 countries**

Clinical annotation using HPOs – **114 participants**

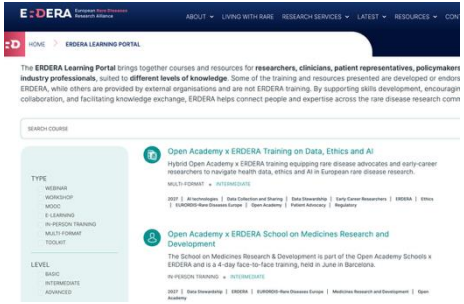
Advancing diagnostics in underrepresented countries – **150 participants, 30 countries**

ERDERA Education Resources

ERDERA Youtube Channel/ biobank resources: https://www.youtube.com/@ERDERA/search?query=biobank



ERDERA YouTube channel - biobank training videos



ERDERA Learning Portal - courses & training resources



EJP RD guidance: organising rare disease biosample data



EJP RD workshop: genetic biobanks for rare disease research

Big thank you to the ERDERA Coordination team

Inserm
La science pour la santé
From science to health



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Coordinator



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RD fundings–Expertise Services Hub



Pauline Adam
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ERDERA

European Rare Diseases Research Alliance

Thank you!

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