ERDERA – One Year of achievements

and forthcoming years

Xavier Mérit

ERDERA, Coordination, Strategy, Project Management

Funded by
the European Union

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30M

European touched

Representing aroung aroung 6% of european population population

7K+

Identified diseases

With regular new discoveries.

80%

Genetic origins

Majority of rare diseases are genetic - based

Diagnostic

Patients usually have to wait 5 years to get a precise diagnostic.

Inequalities of access

Important disparities between european countriesfor healthcare and treatments

Therapeutical developpement

High costs .or small populations. Only 5% have a treatment.

A disease is considered rare when it affects less than 5 persons out of 10 000

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ERDERA: Our Objectives

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

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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.

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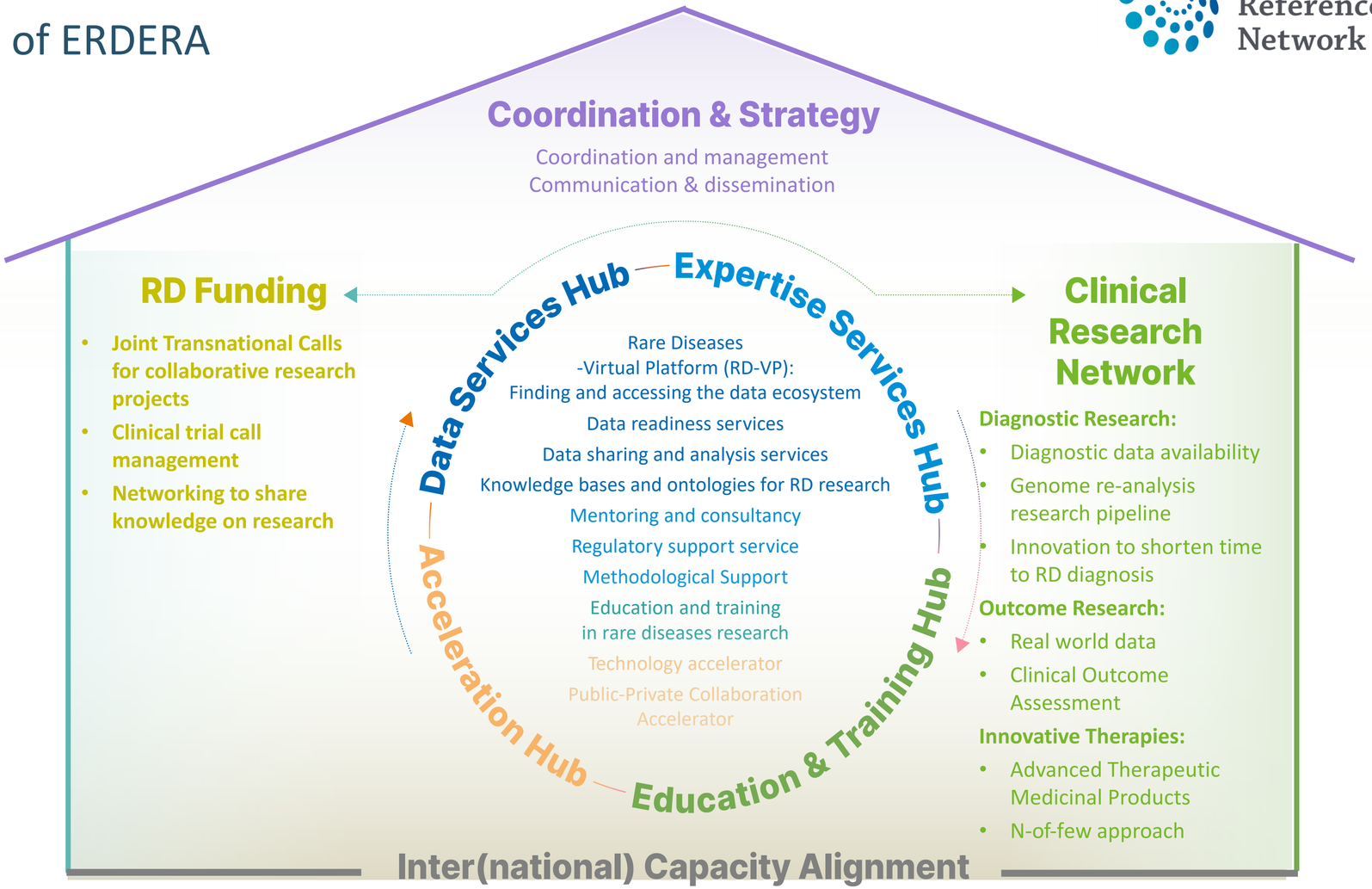
The house of ERDERA



NMGs promotion and national alignment

Fostering engagement of underrepresented countries in ERDERA

ERDERA Global Collaboration

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ERDERA in Action

- Joint Transnational Calls
- Networking Support Scheme
- Clinical Trials Call
- Data Hub
- Expertise Service Hub
- Public-Private Collaboration Accelerator (PPCA)
- TRAINING & EDUCATION
- Clinical Research Network
  - Diagnostics,
  - Outcome Research,
  - Therapies
- Inter(national) Alignment

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Joint Transnational Calls

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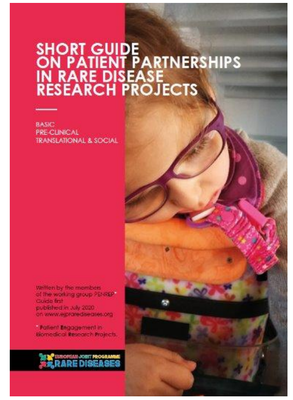
MAIN GOAL: enable scientists in different countries to build an effective collaboration on a common interdisciplinary research project based on complementarities and sharing of expertise, with a clear future benefit for patients

https://erdera.org/funding/#joint-transnational-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

- 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.

Funding: 30 000 € per event

Application: Continuously open (eligibility check every 6 months)

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

Clinical Trials Call

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.

Clinical Trials Call

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

Data Hub

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ERDERA | RESOURCE DISCOVERY | VP NETWORK RESOURCES | WHATS NEW! | LOGIN

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.

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Expertise Service Hub

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MENTORING & CONSULTANCY

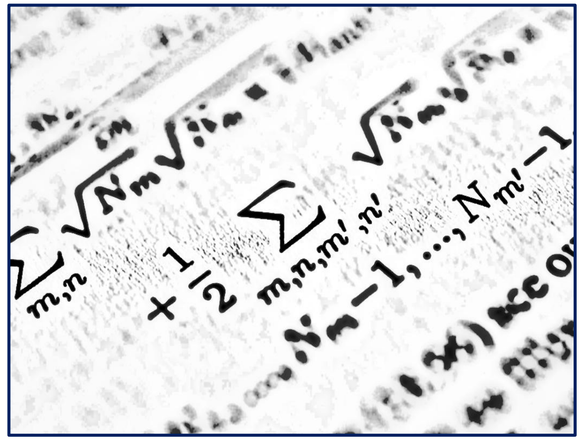
Mentoring Programme:
32 projects accompanied with over 30 mentors, handbook & tracking platform

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.



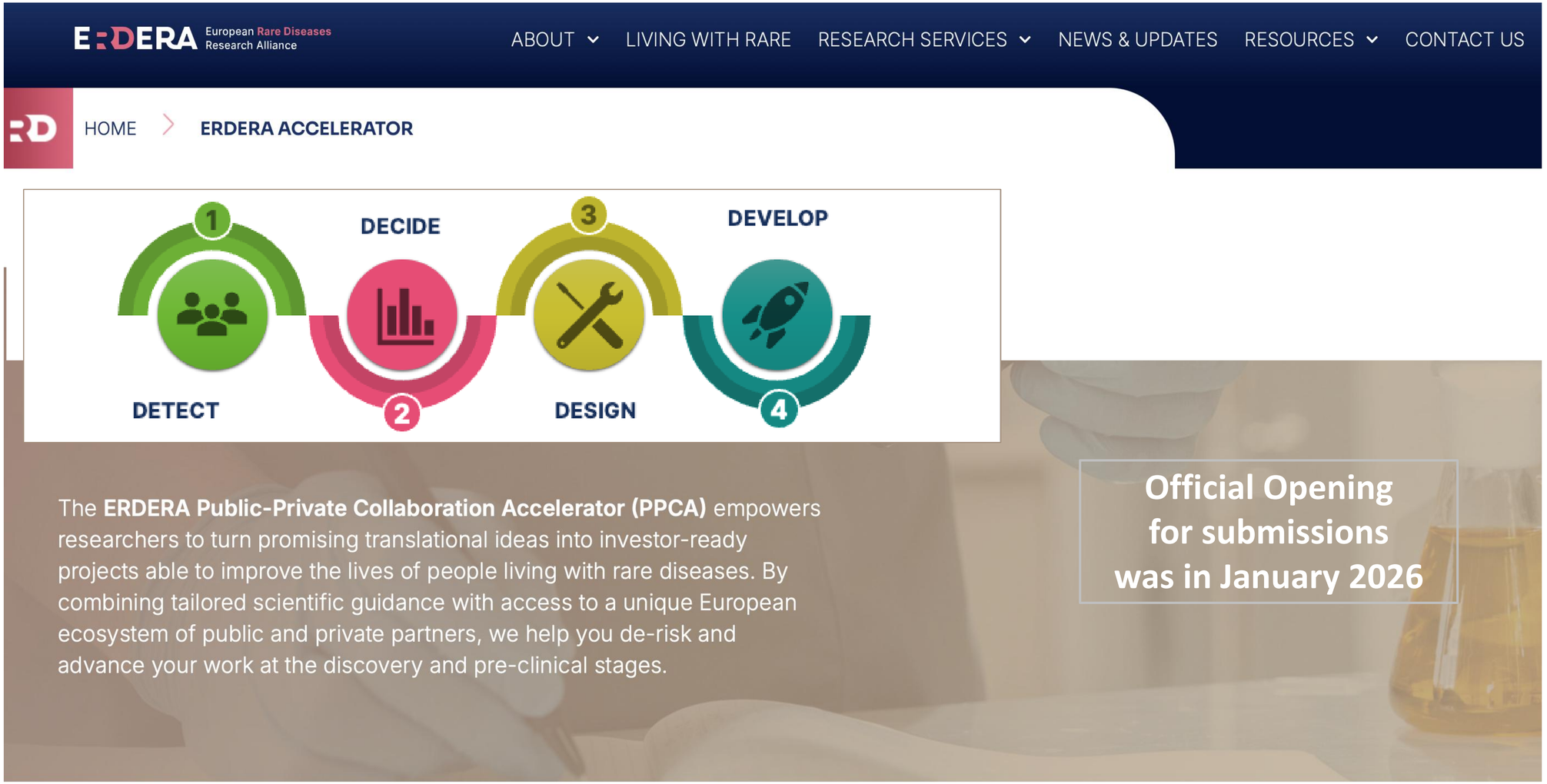
METHODOLOGY

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)



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Training & Education

2400 stakeholders trained

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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**

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Accelerating Rare Disease Diagnosis & Therapies Across Europe

EU diagnostic pipeline

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

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

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

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Inter(national) Alignment

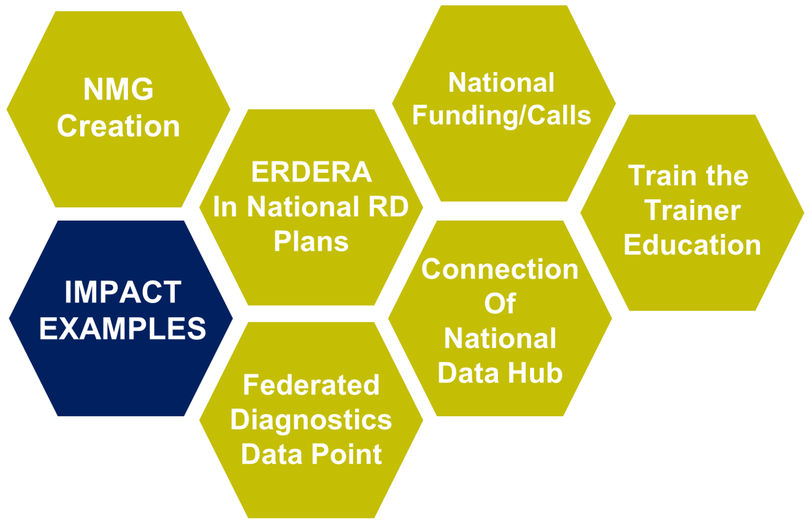
National Alignment

Governing Board
National Mirror Groups
National Alignment Board

- ERDERA Proposal
- SRIA
- Annual Work Plans
- Funding Activities
- Diagnostic pipeline & Infrastructure
- Education & Knowledge Sharing
- Underrepresented Countries Focus

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



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Strategic Alliances

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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

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To the Brilliant Minds and Big Hearts Behind ERDERA Coo

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Inserm
La science pour la santé
From science to health

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https://erdera.org/


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