ERICA ERN Research Conference, 11-13 December 2024

ERN Research:

the expected ERICA legacy

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A. Introduction: ERICA, a “coordination & support action” for ERN research – why? Where do we come from?

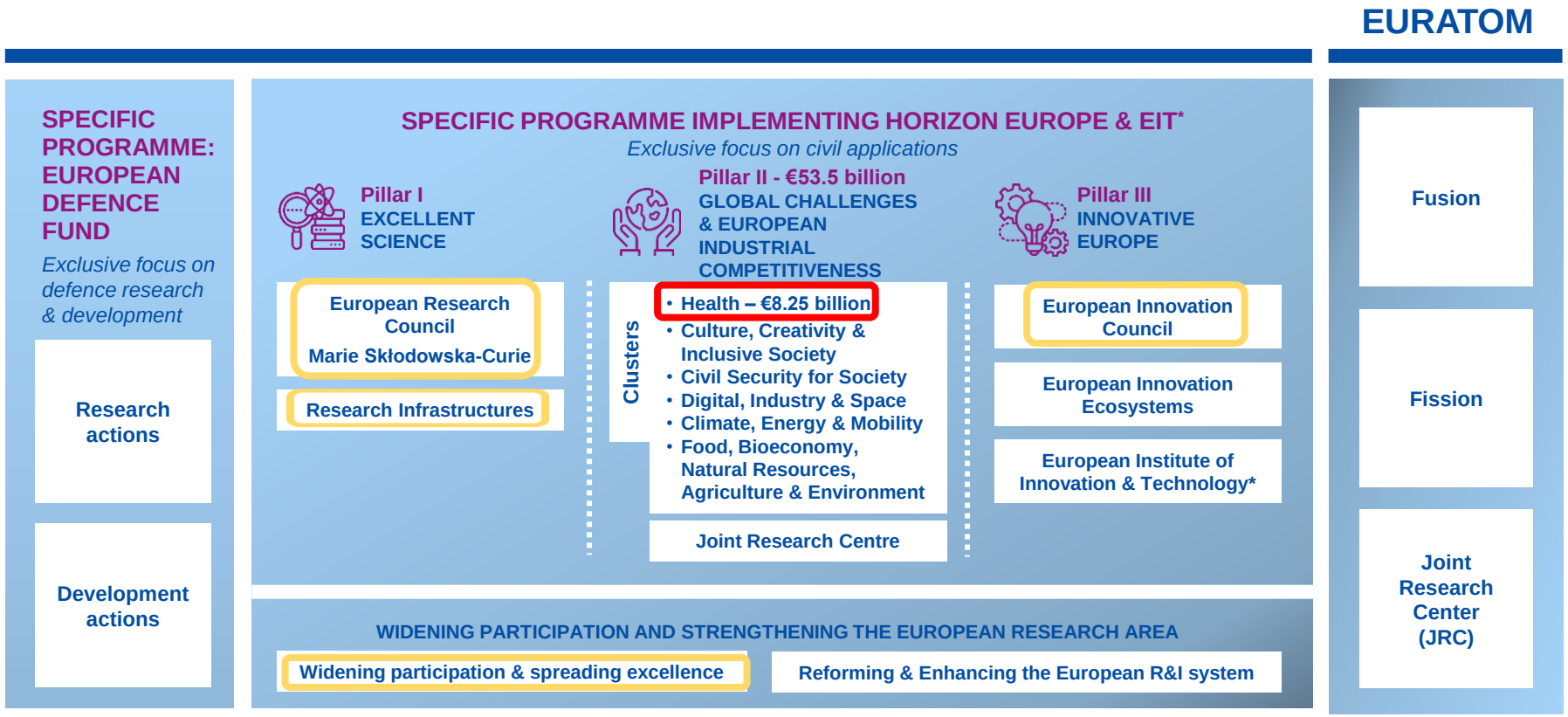
B. (No exhaustive) Horizon mapping of ERN research via EU-funded research projects – happening currently

C. ERNs in the European Partnership on Rare Diseases ERDERA – and outside of ERDERA – looking into the future

D. Conclusion

European Commission

HORIZON EUROPE (2021 – 2027) €95.5 billion



* The European Institute of Innovation & Technology (EIT) is not part of the Specific Programme

European Commission

A. Introduction: ERICA, a “coordination & support action” for ERN research

a “coordination & support action” Why a ‘CSA’ / what is a CSA?

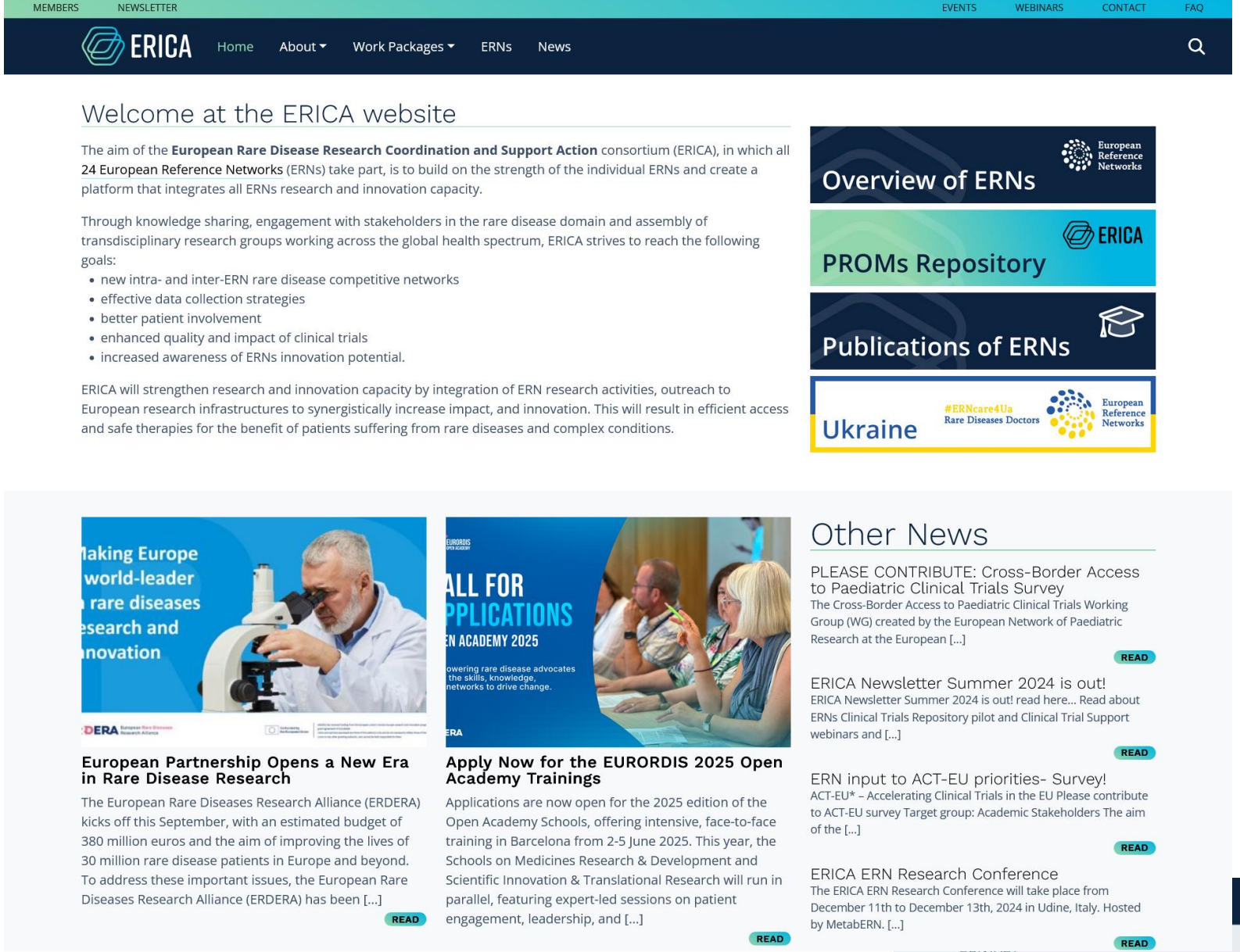
Programme(s)
•H2020-EU.3.1. - SOCIETAL CHALLENGES - Health, demographic change and well-being / H2020-EU.3.1.6. - Health care provision and integrated care

Topic(s)
•SC1-HCO-20-2020 - Coordination of clinical research activities of the European Reference Networks

- A ‘CSA’ is a project type in the Framework Programme covering service projects to coordinate or support research activities and policies (networking, exchanges, trans-national access to research infrastructures, studies, conferences, etc.)

- Meant for the 24 ERNs (supporting them equally + cross-fertilisation)

This activity will aim at enhancing research and innovation capacity of the ERNs in view of achieving the goals of the International Rare Diseases Research Consortium (IRDiRC) for bringing new diagnostic tools and therapies more efficiently to the patients and for developing methodologies to assess the impact of diagnoses and therapies on rare disease patients, taking into account sex and gender differences where relevant. . Support will be given to identify research priorities and potential synergies among ERNs and coordinate research and innovation activities to be tackled by ERNs. The project should address fostering collaboration in the field of clinical research among ERNs, ERN-independent clinical research collaborations and other stakeholders, such as research infrastructures, industry and patient organisations, as well as international collaboration with other clinical research networks. Close collaboration with the European Joint Programme on Rare Diseases will be necessary to ensure complementarity, to achieve relevant synergies and avoid overlaps. To ensure broad geographical representation and participation across ERNs the proposals shall involve participants from several countries and aim at engaging all approved ERNs and other relevant research networks in Europe.



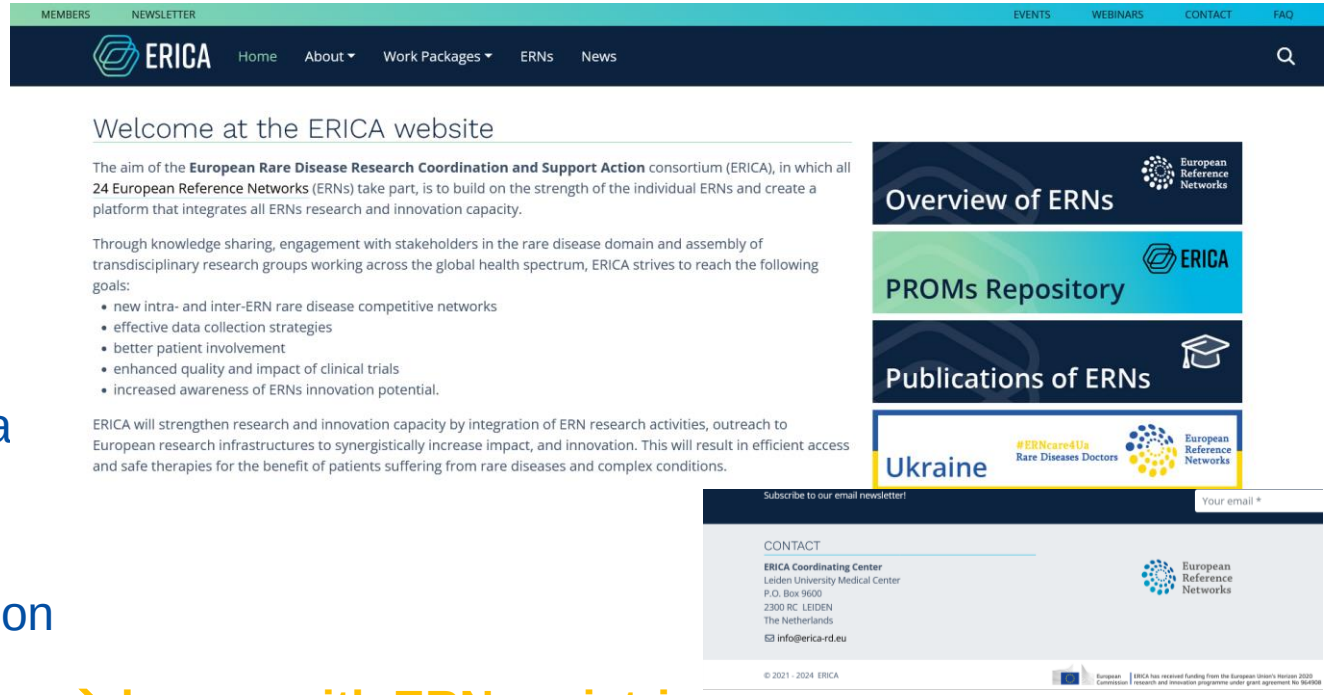
ERICA, the successful proposal, managed to gather all ERNs and other key partners and propose work streams relevant to all ERNs

ERICA, timeframe, workstreams…

- ERICA took over from the

« ERN Research Working Group »

- Started in March 2021 (covid times…) for 4 yea



- WP1: Consortium Management and Coordination
- **WP2: Data collection, Integration and Sharing → legacy with ERN registries**
- **WP3: Patient-Centred Research → legacy with better involvement of patients in RD research, better understanding of patients needs by clinicians and researchers etc.**
- **WP4: Clinical Trial Support → legacy with more/better clinical trials where ERN teams are involved**
- **WP5: Translation and Innovation → legacy with… more translation & innovation !**
- **WP6: Integration, Outreach & Dissemination → legacy with better awareness of the research potential of ERNs (in- and outside of the ERN community)**
- WP7: Ethics Requirements

European Commission

ERNs & their registries: a huge potential for ERN research, enhanced through ERICA efforts

ERN	ERN registry project
Endo-ERN	EuRRECa
ERKNet	ERK-Reg
ERN-LUNG	REGISTRY WAREHOUSE
MetabERN	U-IMD
ERN PaedCan	PARTNER
ERN GUARD-HEART	GUARD-Heart Registry
ERN ReCONNET	TogethERN ReCONNET
ERN ITHACA	ILIAD
ERN BOND	EuRR-Bone
ERN CRANIO	ERN CRANIO registry
ERN EURACAN	STARTER
ERN RND	ERN-RND Registry

ERN	ERN registry project
ERNICA	ERNICA registry
ERN EYE	REDgistry
ERN EpiCARE	EPICARE-GRANT
VASCERN	VASCERN Registries
ERN-Skin	ERN-Skin REGISTRY
ERN EURO-NMD	EURO-NMD Registry
ERN GENTURIS	GENTURIS registry
ERN RARE-LIVER	R-LIVER
ERN RITA	MERITA
ERN TRANSPLANT-CHILD	PETER
ERN EuroBloodNet	ENROL
ERN eUROGEN	ERN-eUROGEN registry

Joint Research Centre
JRC

European Platform on Rare Disease Registration (EU RD Platform)

Searchable, findable rare disease registry data

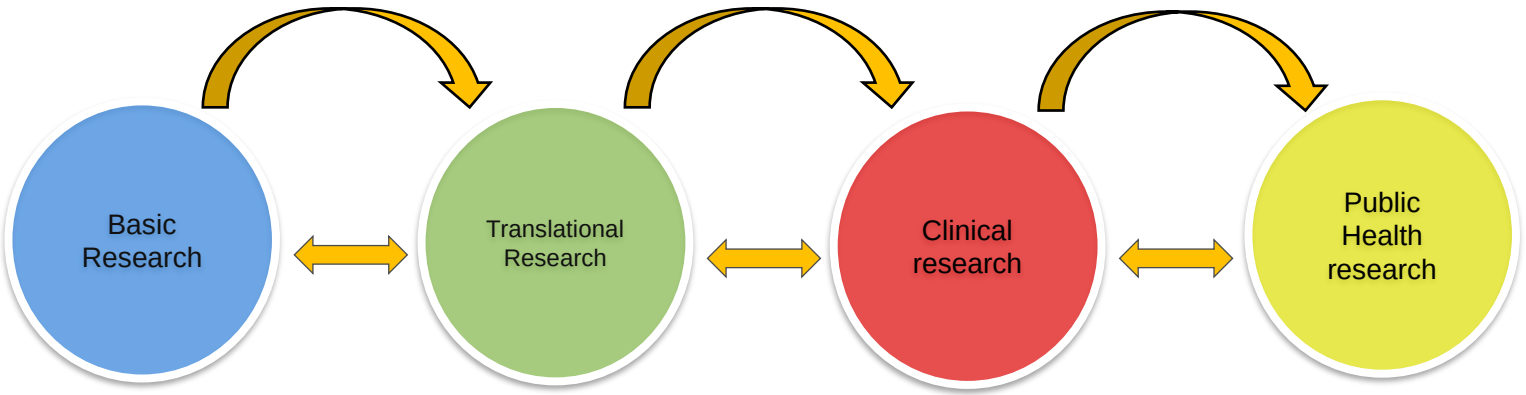
https://eu-rd-platform.jrc.ec.europa.eu/erdri_en

B. (No exhaustive) Horizon mapping of ERN research via EU-funded research projects

"ERN research" might mean many different types of research…

EU investment on research into rare diseases

FP7 (2007-2013) €3.8 billion
H2020 (2014-2020) €3.2 billion > 550 multi-partner projects



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B. Horizon mapping of ERN research via EU-funded research projects

ERICA legacy will depend on the state of play of research for respective ERNs and even disease areas.

And of course this was, and is still, very heterogeneous…

Let’s try some mapping – non exhaustive & for EU-funded research (= does not capture national funding, funding by charities, or even funding by EJP RD co-fund under Horizon 2020)

Collaborative research projects

HORIZON-HLTH-2022-DISEASE-06-04-two-stage: Development of new effective therapies for rare diseases

Relevant for:

ERN Euro-NMD

ERNs ITHACA and EpiCARE

ERN GUARD-Heart

Endo-ERN & more with

ERN Euro-NMD

ERN EuroBloodNet

ERN-EYE

MetabERN, ERN-RND & ERN EURO-NMD

ERKNet

- Drug REpurposing with Artificial intelligence for Muscular disorderS (for 5 rare neuromuscular disorders)
- EUropean network for neurodevelopmental RASopathies (Cardio-facio-cutaneous syndrome, Costello syndrome and SYNGAP1-related)
- Gene Therapy for treatment of rare inherited Arrhythmogenic Cardiomyopathy
- LightCure - Light for double specificity and efficacy without burden (congenital hyperinsulinism (CHI), cause of hypoglycemic brain injury with intellectual disability, epilepsy and cerebral palsy)
- Next-generation models and genetic therapies for rare neuromuscular diseases
- Nanomedicine Approach to Normalize Erythrocyte Maturation in Congenital Anemia by messenger RNA
- Novel advanced and repurposed therapeutics for vision restoration in a group of severe rare ocular surface diseases: from validation to first clinical investigations
- Accelerating drug repurposing for rare neurological, neurometabolic and neuromuscular disorders by exploiting SIMilarities in clinical and molecular PATHology www.simpathic.eu
- Therapies for Renal Ciliopathies

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All descriptions available on Cordis database: https://cordis.europa.eu/

Collaborative research projects

HORIZON-HLTH-2023-IND-06-04 - Modelling and simulation to address regulatory needs in the development of orphan and paediatric medicines

- INVENTS - **Innovative designs, extrapolation, simulation methods and evidence-tools for rare diseases addressing regulatory needs (2024-2028)**:

6,2 M€ budget
(99% EU contribution)

Relevant for all ERNs (and in particular for cooperation between ERNs & industry)

Innovative designs and tools for rare disease medicines evaluation: Assessing the efficacy and safety of new medicines for rare diseases, particularly paediatric conditions, poses several challenges. These include small patient sample sizes, patient and disease heterogeneity, and limited disease knowledge, which hinder effective treatment access and limit available options. INVENTS seeks to refine longitudinal model-based disease trajectories, improve extrapolation models, and implement in silico trials, considering patient's needs, to enhance regulatory decision-making. The project's outcomes are expected to enable researchers, regulators and the European pharmaceutical industry to leverage innovative clinical trial designs, in silico trials, and real-world data analysis approaches. Patients with rare diseases stand to benefit from increased and expedited access to effective and safe treatments.
https://cordis.europa.eu/project/id/101136365 ; https://invents-he.eu/

Relevant for all ERNs , in particular: ERNs ERN-RND, Euro-BloodNet and EURO-NMD

- ERAMET - **Ecosystem for rapid adoption of modelling and simulation METhods to address regulatory needs in the development of orphan and paediatric medicines (2024-2027)**: Limited patient populations and ethical constraints delay access to life-saving treatments for paediatric and orphan diseases. Traditional methods struggle to provide reliable assessments of drug efficacy and safety. In this context, the EU-funded ERAMET project integrates modelling and simulation methods to establish credibility within regulatory procedures. Its transparent ecosystem connects questions, data, and methods, facilitating adoption of advanced techniques like AI and hybrid approaches. Three pillars underpin ERAMET: a versatile repository, high-quality standards for data and methods, and an AI-driven platform for automated analysis and credibility assessment. 5 use-cases in 4 groups of RDs (ataxia, transfusion dependent haemoglobinopathies, bronchopulmonary dysplasia, degenerative neuromuscular disorders) demonstrate ERAMET's potential, aiming for regulatory approval of validated tools. Training initiatives will ensure stakeholders embrace this transformative paradigm.https://cordis.europa.eu/project/id/101137141

3,86 M€ budget
(100% EU contribution)

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Collaborative research projects

More EU-funded research projects (not funded under 'RD-specific' topics)

Relevant for:

- ERN EuroBloodNet
- ERN EURACAN
- ERN Euro-NMD
- MetabERN
- For all ERNs !
- ERNs PaedCan & EURACAN
- ERN TRANSPLANT-CHILD
- ERN GENTURIS

- **GenoMed4All:** Genomics and Personalized Medicine for all though Artificial Intelligence in **Haematological** Diseases *(funded under Topic:* DT-TDS-04-2020 - AI for Genomics and Personalised Medicine)
- **IDEA4RC**: Intelligent Ecosystem to improve the governance, the sharing and the re-use of health Data for Rare Cancers *(funded under Topic: HORIZON-HLTH-2021-TOOL-06-03 - Innovative tools for use and re-use of health data (in particular of electronic health records and/or patient registries)*
- **PaLaDin**: Patient Lifestyle and Disease Data Interactium - Innovative approach for neuromuscular disease patient data integration *(Topic: HORIZON-JU-IHI-2022-03-02 – Patient-generated evidence to improve outcomes, support decision making, and accelerate innovation)*

  Public-private project ! (IHI)
- **Recon4IMD**: Reconstruction and Computational Modelling for Inherited Metabolic Diseases *(funded under: Programme: HORIZON.2.1.5 - Tools, Technologies and Digital Solutions for Health and Care, including personalised medicine ; Topic: HORIZON-HLTH-2022-TOOL-12-01-two-stage - Computational models for new patient stratification strategies*
- **Remedi4All**: Building a sustainable European Innovation Platform to enhance the repurposing of medicines for all
- **REPO4EU**: Precision drug REPurpOsing For EUrope and the world
- **MONALISA**: A SIOPEN pragmatic clinical trial to MOnitor NeuroblastomA relapse with LIquid biopsy Sensitive Analysis (High-risk neuroblastoma) *(funded under the Cancer Mission)*
- **PROTECT-CHILD**: A PRivacy-prOTecting Environment for Child Transplants health-related and genomic data integration in the European Reference Network
- **PREVENTABLE**: Cancer Prevention VS Cancer Treatment: the Rare Tumour Risk Syndromes Battle

More EU-funded research projects (not funded under 'RD-specific topics)

Relevant for:

Marie Skłodowska-Curie Actions (MSCAs)

- ERKNet
- **DRUGTrain**: Drug repurposing and discovery multidisciplinary training network (Autosomal Dominant Polycystic Kidney Disease)
- ERN-LUNG
- **ORGESTRA**: Organoid technologies for disease modeling, drug discovery and development for rare diseases (Cystic fibrosis (CF) and cystinosis)
- **CF.Nbs.PA-SA**: Development of Nanobodies against Pseudomona aeruginosa and Staphylococcus aureus and evaluation as immunotherapeutics (Cystic fibrosis)
- All ERNs
- **GetRadi**: Gene Therapy of Rare Diseases

European Research Council (ERC) grants

- ERN CRANIO
- **CAD4FACE**: Computational modelling for personalised treatment of congenital craniofacial abnormalities (ERC 'Starting Grant')
- ERN RARE-LIVER and potentially all ERNs
- **IMPACT**: IMPlementation of Affordable gene Correction Therapies - Methylmalonic Acidemia (MMA) (ERC 'Proof of Concept' Grant)

And many more examples in various disease areas !

Be curious and search for your disease area in Cordis !

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More EU-funded research projects

Relevant for:

MetabERN

- **Nano4Rare**: Preclinical development of a nanomedicine candidate for Fabry rare disease treatment to enter clinical phase

EIC (Transition)

Nano4Rare team has successfully developed a new patent protected medicinal product candidate, named nanoGLA, for the treatment of Fabry disease, which is one of the most devasting LSD rare diseases. NanoGLA has been designated, in January 2021, as Orphan Drug by the European Medicine Agency (EMA). With the EU H2020 Smart4Fabry, nanoGLA was brought to an advanced stage of preclinical development (first GLP-toxicology in rats included). Under Phoenix EU Project (#953110)(2021-2025), nano-GLA production will be scaled-up and brought to GMP conditions. As a first objective, Nano4Rare project will use nanoGLA engineered batches produced in the frame of Phoenix project to complete the preclinical phase and generate sufficient quality data on safety, efficacy, and quality in order to get approval by regulators to proceed with clinical phase. As a second objective, a new spin-off company will be created for the advancement of the nanoGLA towards the market and the commercialisation of the patent protected nanoencapsulation platform to generate new product candidates for rare disease treatments.

relevant for ERN CRANIO, ERN-Skin and all ERNs involving surgery, skin expansion etc.

- **BIOMET4D**: Smart 4D biodegradable metallic shape-shifting implants for dynamic tissue restoration (101047008); Topic: HORIZON-EIC-2021-PATHFINDEROPEN-01-01 - EIC Pathfinder Open 2021

EIC (Pathfinder)

Relevant for several ERNs (GUARD-HEART, GENTURIS, EURACAN etc.)

- **TraffikGene-Tx**: Targeted Peptide Carriers for RNA Delivery

EIC (Transition)

ERN EuroBloodNet

HemaFAIR

- Fostering F.A.I.R. Data and Standards in Rare Hematological Diseases (from “Widening programme”, CY institution coordinating)

And more !!!

European Commission

Collaborative research projects

Other EU-funded research projects ('generic' but with some RDs as use cases)

HTx - Next Generation Health Technology Assessment to support patient-centred, societally oriented, real-time decision-making on access and reimbursement for health technologies throughout Europe (2019-2024)

HORIZON 2020 PROJECT

9,64 M€ budget
(100% EU contribution)

With Eurordis

Objectives HTx aimed to create a framework for next generation Health Technology Assessment (HTA) that supports patient-centred, societally oriented, and real-time decision-making for integrated healthcare throughout Europe. HTx will focus on therapeutic areas with high unmet need for which HTA information has to be provided on complex and personalised combinations of health technologies.
https://cordis.europa.eu/project/id/825162

More-EUROPA - More Effectively Using Registries to suppOrt PAtient-centered Regulatory and HTA decision-making (2023-2027)

HORIZON EUROPE PROJECT

7 M€ budget
(100% EU contribution)

With patient organisations: Eurordis and the European Multiple Sclerosis Platform AISBL

Setting standards for real-world evidence in the medicinal decision-making process: While randomized controlled trials (RCTs) remain the mainstay in drug development, approval, and reimbursement, the potential of real world data (RWD) to contribute to the understanding of drug effects is increasingly realized. Aim of the project is to develop, implement and establish evidentiary standards and methods to address the data and evidentiary needs of regulatory authorities and HTA bodies towards a more efficient use of RWD for the development, registration and assessment of medicinal products in Europe.
https://cordis.europa.eu/project/id/101095479

European Commission

Public-private project !

C4C: conect4children - Collaborative network for European clinical trials for children (2018-2025)

ONGOING → will be further expanded

Objectives

- Create a sustainable, integrated pan-European collaborative paediatric network that will speed up of high-quality clinical trials in children, ensuring that the voices of young patients and their families are heard.
- Build the capacity for conducting multinational paediatric clinical trials for all disease areas and all phases of the clinical drug development process.

Outcomes

- C4C established an “Always on” paediatric clinical research network of > 400 experts, > 220 sites organized in 18 National Hubs
- Set up and deployed a service to integrate input from children, young people, and families into Expert Advice about on paediatric clinical research
- Training Health Care Professionals how to work with children and young people
- Established the European Young People’s Advisory Group Network (eYPAGnet)

154.4 M€ budget
67 M€ EU contribution
87.4 M€ EFPIA contribution

10 industry partners
34 universities, research organisations, non-profit private
1 SME
1 patient organisation (Eurordis)
10 third parties

20 countries
18 EU Member States
2 non-EU

https://cordis.europa.eu/project/id/777389
https://conect4children.org/

European Commission

Public-private project !

Screen4Care (S4C) - Shortening the path to rare disease diagnosis by using newborn genetic screening and digital technologies (2021-2026)

Objectives **ONGOING !!**

- S4C will leverage the genomic and digital advent to develop and pilot genetic NBS and AI-guided symptom recognition algorithms, while accounting for all relevant legal, regulatory and ethical considerations. S4C aims to harmonize the results of existing efforts in a horizon scan, by looking at the totality of the available data resources, diagnostic algorithms, and other initiatives with similar ultimate goals.
- The genetic NBS will interrogate 1) currently treatable RDs (TREAT-map gene panel), 2) actionable RDs (ACT-map gene panel) in 18.000 new-borns in 3 EU countries (D, It, and Cz). Further, S4C will offer whole genome sequencing (WGS) to early symptomatic babies, tested negatively during panel-based NBS to identify known NBS-escaped RDs and novel genes/phenotypes.
- S4C will also provide two digital diagnosis support systems for RD on the basis of features and symptom complexes: 1) federated ML- and literature-evidence-based algorithm for continuous and automated screening of EHR and 2) meta symptom checker with virtual clinics for patients and HCP offering the possibility of increased accuracy of diagnosis and ongoing supports. Our ambitious goal is to evaluate the validity of our multi-pronged approach to shorten the time to diagnosis for all patients affect by RDs, improve value-based healthcare resource utilization, and hopefully reduce the suffering of millions of European citizens.

26,2 M€ budget
11,9 M€ EU contribution
14,3 M€ EFPIA contribution
-
industry partners
universities,
research organisations,
non-profit private, SMEs
1 patient organisation (Eurordis)
-

20 countries
18 EU Member States
2 non-EU

https://screen4care.eu/ ; https://cordis.europa.eu/project/id/101034427

European Commission

Public-private project !

FACILITATE - FrAmework for ClInicaL trIal participants daTA reutilization for a fully Transparent and Ethical ecosystem (2022-2025)

Objectives **ONGOING !**

Finding ways to access, use and reuse patient data

Has the EU General Data Protection Regulation (GDPR) limited Europe's capabilities in innovative drug development? The EU-funded FACILITATE project will explore the issue. Its aim is to find an ethical and GDPR-compliant framework for returning clinical trial data to study participants. Specifically, it will look for ways to allow patients' data to be accessed, used and reused. The project will also show how a patient-centred data-driven approach can improve the drug development and approval process. The findings will shed new light on future strategies and opportunities for medicines development and regulation.

https://cordis.europa.eu/project/id/101034366

7,56 M€ budget
3,26 M€ EU contribution
4,3 M€ EFPIA contribution

with patient organisations:
Eurordis, Stichting EUPATI Foundation +
Accademia del Paziente Esperto EUPATI APS

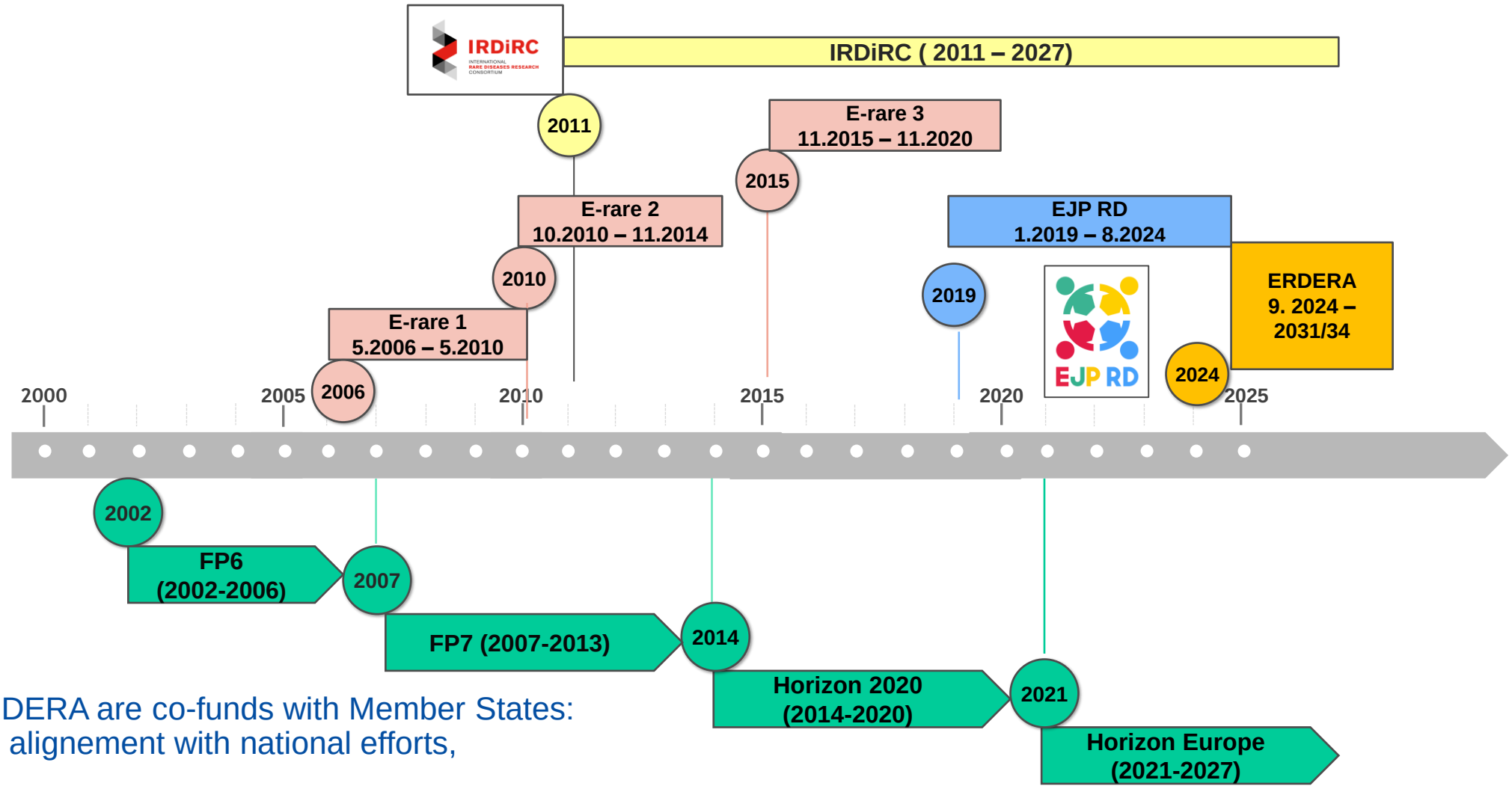
European Commission

C. ERNs in the European Partnership on Rare Diseases ERDERA – and outside of ERDERA – looking into the future

- ERNs in EJP RD (state of play in 2028-2019) versus ERNs in ERDERA (state of play in 2024)
- ERICA legacy in ERDERA + what is in for ERNs
- ERICA legacy ‘outside of ERDERA’…

European Commission

EU research & innovation on rare diseases (RD) - Coordination of national & international research funding



EJP RD & ERDERA are co-funds with Member States: contribution & alignement with national efforts,

And also with other Partnerships under Horizon Europe (ERA4Health, Transforming Health & care Systems (THCS), Personalised Medicine, Innovative Health Initiative etc.) or other actions such as JARDIN under EU4Health

European Commission

ERDERA
European Rare Diseases Research Alliance

178 Organisations

40 funders

81 research performing organisations

9 patients' organisations

3 research infrastructures

22 private for-profit partners (industry & SME)

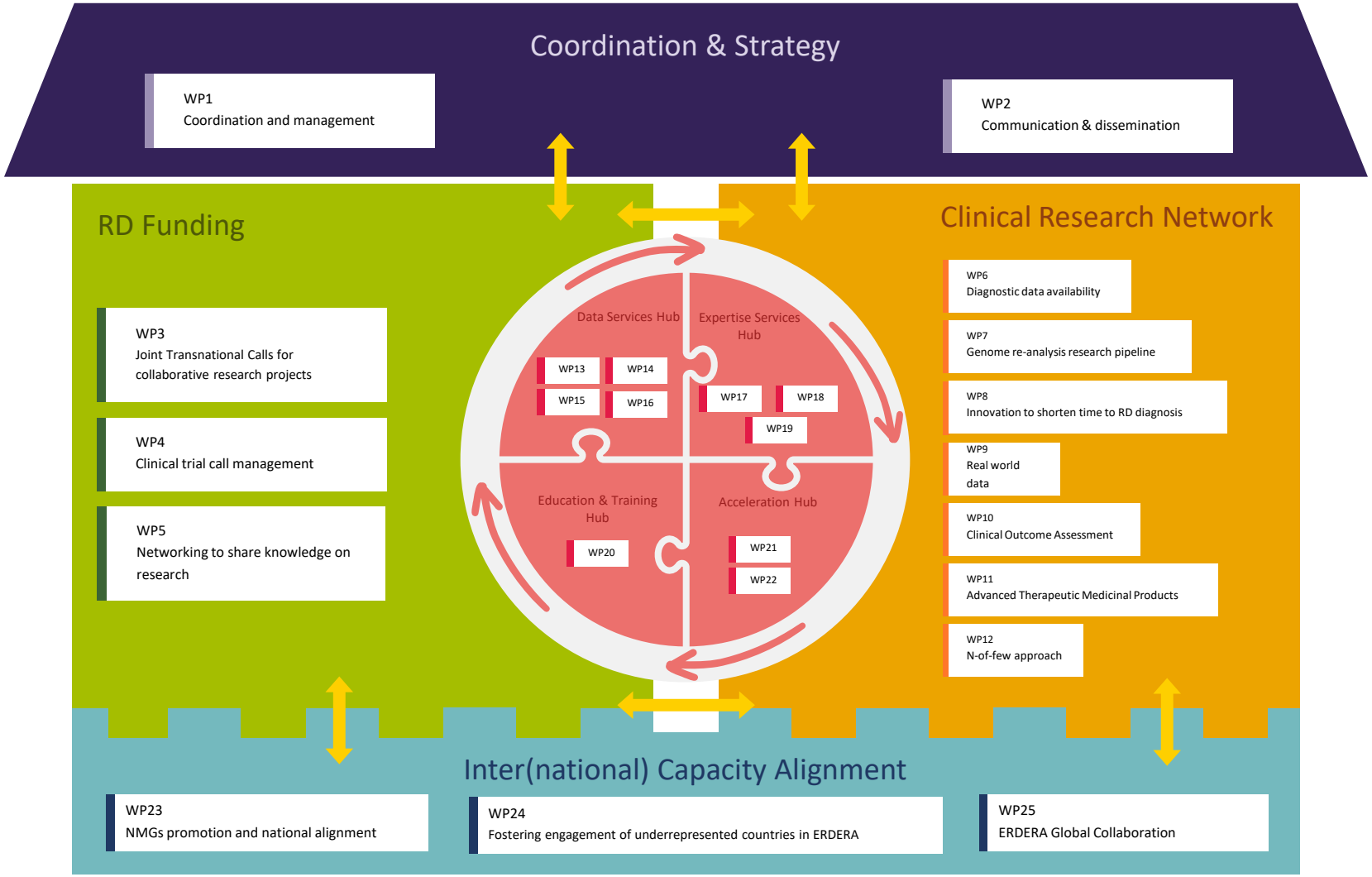
23 other (univ, hospital, non-profit, public administration)

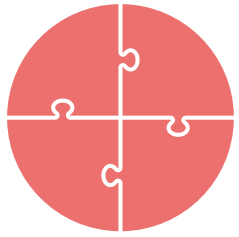
37 Countries

26 EU member states

8 associated countries

3 non-EU





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

ERDERA (European Rare Diseases Research Alliance)

ERDERA Work Packages (WPs):

COORDINATION AND STRATEGY
WP1: Coordination and management
WP2: Communication and dissemination

FUNDING *(! ERN Research !)*
WP3: Joint Transnational Calls (JTCs) for collaborative research projects
(ERICA legacy if helped preparing ERN teams to apply !)
WP4: Clinical trial call management (ERICA WP3 legacy)
WP5: Networking to share knowledge (ERICA WP4 legacy)

CLINICAL RESEARCH NETWORK (! ERN Research !)
WP6: Diagnostic data availability (Solve-RD legacy)
WP7: Genome re-analysis research pipeline (Solve-RD legacy)
WP8: Innovation to shorten time to RD diagnosis (Solve-RD legacy)
WP9: Real World Data (ERICA WP2 legacy)
WP10: Clinical Outcome Assessment (! ERN Research !)
WP11: ATMPs
WP12: N-of-few Approach

DATA SERVICE HUB
WP13: RD 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

EXPERT SERVICES HUB
WP17: Mentoring and consultancy
WP18: Regulatory support service
WP19: Methodological support

EDUCATION & TRAINING HUB
WP20: Education and Training on RD Research

ACCELERATION HUB
WP21: Technology accelerator
WP22: Public-Private Collaboration accelerator (IMI-IHI legacy)

INTER(NATIONAL) CAPACITY ALIGNMENT
WP23: National Mirror Groups' (NMGs) promotion and national alignment
WP24: Fostering engagement of underrepresented countries in ERDERA
WP25: ERDERA Global Collaborations (ERICA legacy)

WP26: Ethics requirements

ERDERA (European Rare Diseases Research Alliance)

Example of how to best include patients in research projects funded under ERDERA 'Joint Transnational Calls' (JTCs):

Nota Bene: patients' inclusion funded on EU funding since often not foreseen/possible via national/regional funding

• "The project team is expected to include **at least one patient partner (patient/caregiver/family member) or a patient advocacy organisation (PAO)** in the role of Principal Knowledge User (PKU), Knowledge User (KU) or Co-Applicant as appropriate. Details must be provided regarding the patient partner involvement plan and the consideration given to patient compensation must be explicit in the proposed budget."

 Categories of Partners:

Partners belonging to one of the following categories may request funding under a joint research proposal (according to country/regional regulations):

• Academia (research teams working in universities, other higher education institutions or research institutes),

• Clinical/public health sector (research teams working in hospitals/public health and/or other health care settings and health organisations),

• Enterprises (all sizes of private companies). Participation of small and medium-sized enterprises (SMEs) is encouraged when allowed by national/regional regulations,

• Patient advocacy organisations (PAOs).

ERDERA (European Rare Diseases Research Alliance)

Example of how to best include patients in research projects funded under ERDERA 'Joint Transnational Calls' (JTCs):

(2)

 Patient Advocacy Organisations and Patient Involvement/Partnership:

Consortia are expected to include and actively engage patient partners (patients/caregivers/family members) and/or patient advocacy organisations (PAOs) from the start when preparing their proposals.
For information on where to find patient partners and PAOs willing to be involved in research, please see:

- Orphanet portal for rare diseases and drugs patient organisation directory
- Rare Diseases Europe (EURORDIS)
- European Reference Networks (ERNs)
- European Patient's Academy on Therapeutic Innovation (EUPATI)
- European Patients' Forum https://www.eu-patient.eu/
- Research Patient Partnership resources (CIHR-IG)

The consortia should **clearly describe the role and responsibilities of the patient partners and PAOs**, how they will operate, at what levels and stages of the research, and provide justifications for allocated resources in a patient involvement plan. It is highly encouraged that patient partners and PAOs are involved in all levels of the proposed work, including in project design, by advising on prioritization, sitting on advisory groups, and/or being a member of the consortium steering group or the governance group. Patient partners and PAOs may be part of institutional scientific boards to discuss the proposal and subsequent study on issues such as:

- the research idea, for relevance to patient concerns;
- possible outcomes;
- informed consent;
- plain language summaries, and;
- patient input on appropriate outcome measures;
- possible patient intervention in the project;
- review of the data collected;
- dissemination of research findings;
- review of data use conditions and access procedures within and outside of the project consortium, within and beyond the runtime of the project.

ERDERA (European Rare Diseases Research Alliance)

ERDERA 'Joint Transnational Call 2025'

Announcement on 10 December 2024:

Join the 2025 Joint Transnational Call Webinar on 17 December

ERDERA @ERDERA_org · 8. Nov.

🌍 📢 Reminder: Our 2025 Joint Transnational Call for Proposals draft announcement is available for consultation!

🔗More information here: loom.ly/-YEsh3Y
👉Stay tuned for our webinar on 17 Dec to learn more!

2 4 272

The European Rare Diseases Research Alliance (ERDERA) is delighted to invite researchers, patient advocates, and early career professionals to a key **webinar** on **17 December 2024,** following the official launch of the 2025 Joint Transnational Call for Proposals for **"Pre-clinical therapy studies for rare diseases using small molecules and biologicals – development and validation"** on 10 December.

Click **here to register for the webinar.**

Agenda (CET)

- 14.00-14.10 Welcome and Introduction to ERDERA (Daria Julkowska)
- 14.10-14.30 JTC 2025 Introduction (Ralph Schuster)
- 14.30-14.40 Pre-clinical therapy development – Evaluators perspective (Christine Kinnon)
- 14.40-15.00 Q&A – Round 1
- 15.00-15.10 Patient engagement in research (Roseline Favresse, Avril Kennan)
- 15.10-15.20 ERDERA expertise support services (Viviana Giannuzzi, Toni Andreu, Rima Nabbout)
- 15.20-15.30 Data Hub and FAIR data (Marco Roos)
- 15.30-15.50 Q&A – Round 2
- 15.50-16.00 Closing remarks

Registration is mandatory

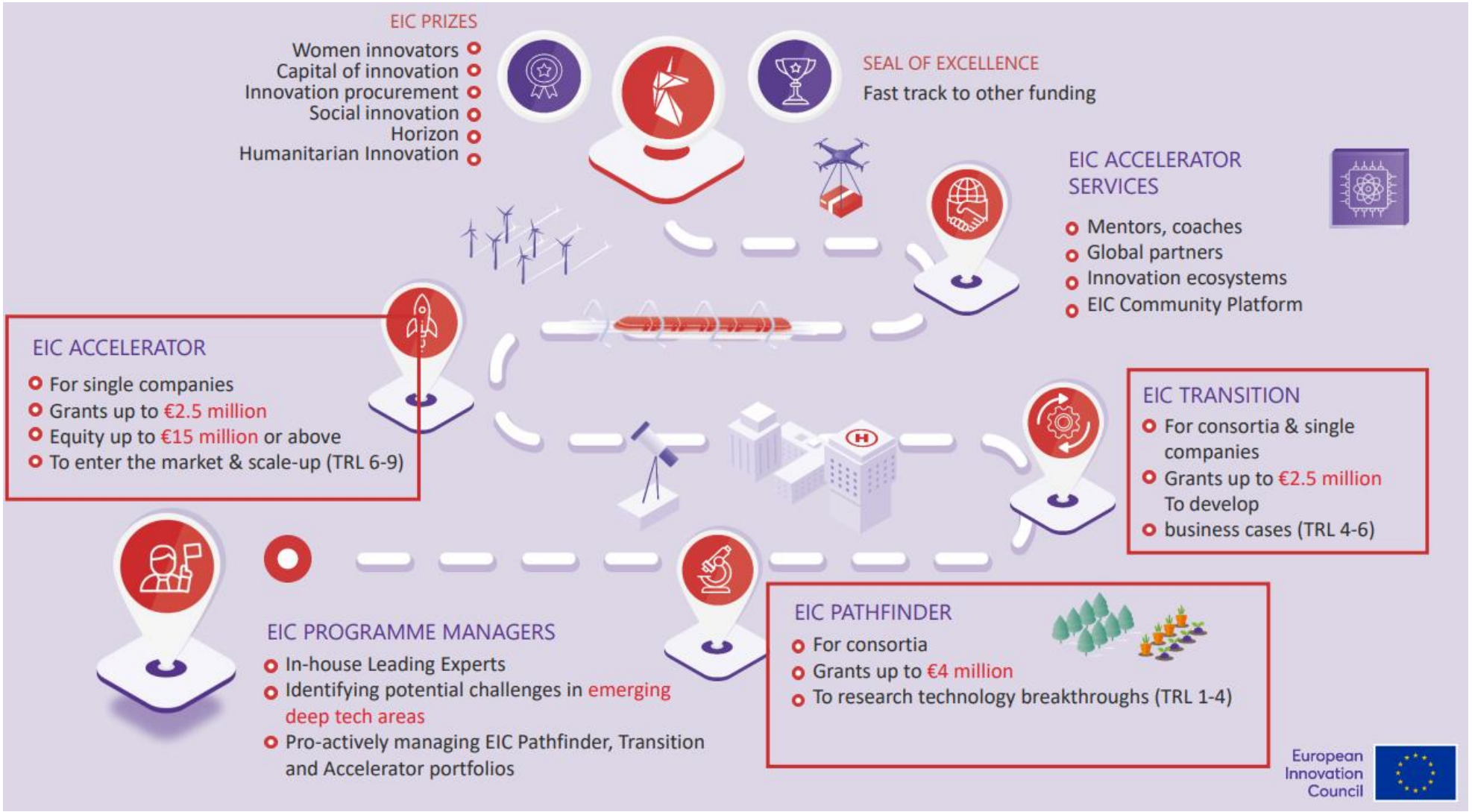
- The **registration deadline** is **17 December 2024.**
- Upon submitting your registration, you will receive connection links and calendar invitations to ensure seamless participation.

Note: The webinar will be recorded, and the video recording will be made available **exclusively** to registered participants.

Don't miss this chance to get ahead in the application process and ensure that your project stands out!

Find out more details about ERDERA's Joint Transnational Call 2025 – Preliminary announcement here.

For further thoughts & discussions…



Courtesy: Uschi (Orsolya) Symmons Programme Manager for Health and Biotechnology

EIC main instruments and characteristics

European Innovation Council

Pathfinder

- **Early stage research** on breakthrough technologies
- Grants up to €3/4 million
- Successor of FET (Open & Proactive)

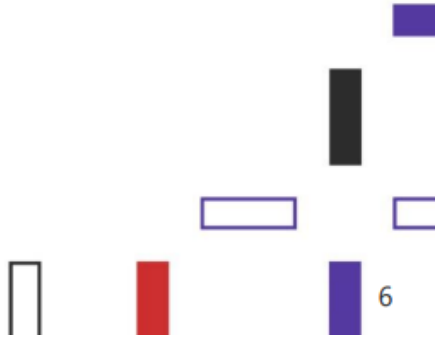
Transition

- **Technology maturation** from proof of concept to validation
- **Business & market readiness**
- Grants up to €2.5 million

Accelerator

- **Development & scale up** of deep-tech/ disruptive innovations by startups/ SMEs
- Blended finance (grants up to €2.5 million; equity investment up to €10 million)
- Successor of SME instrument

- **Business Acceleration Services** (coaches/ mentors, corporates, investors, ecosystem)
- **Pro-active management** (roadmaps, reviews, re-orientations, etc) with EIC Programme Managers
- **Connection with other EU / national funding programs** (ERC, EIT, collaborative) & national programmes



Courtesy: Uschi (Orsolya) Symmons Programme Manager for Health and Biotechnology

ESFRI RIs linked to health research

Imaging facilities

- **EUROBIOIMAGING – Imaging facilities**

Biological Resource Centres

- **BBMRI - Biobanks and Biomolecular Resources**
- **EMBRC - Marine biology resources**
- **EU-OPENSCREEN - Chemical libraries**
- **INFRAFRONTIER - Mouse archives and clinics**
- **MIRRI – Microbial resources**

Molecuclar Biology facilities

- **INSTRUCT - Structural biology facilities**

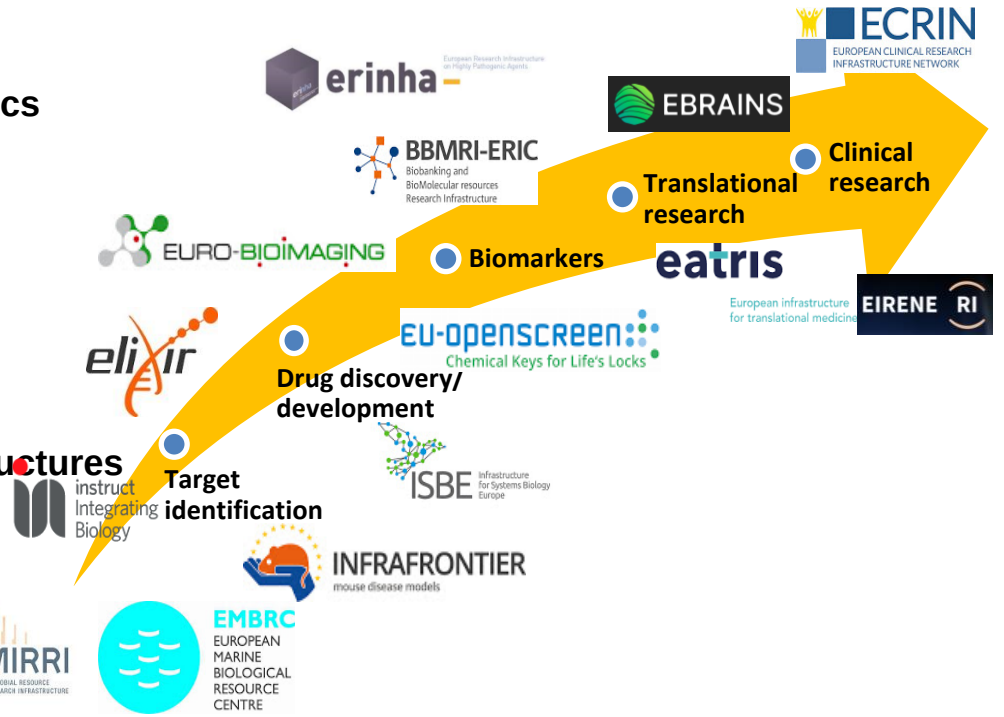
Bioinformatics resources

- **ELIXIR – Data repositories**
- **eBrains – European Brain Research Infrastructures**

Medical research facilities

- **EATRIS – Translational medicine research**
- **ECRIN - Clinical trial platform**
- **ERINHA - High-security labs**
- **EIRENE – Environmental Exposure Assessment**

Wide spectrum of research, discovery and development on health challenges



ERNs & their registries: a huge potential !

ERN	ERN registry project
Endo-ERN	EuRRECa
ERKNet	ERK-Reg
ERN-LUNG	REGISTRY WAREHOUSE
MetabERN	U-IMD
ERN PaedCan	PARTNER
ERN GUARD-HEART	GUARD-Heart Registry
ERN ReCONNET	TogethERN ReCONNET
ERN ITHACA	ILIAD
ERN BOND	EuRR-Bone
ERN CRANIO	ERN CRANIO registry
ERN EURACAN	STARTER
ERN RND	ERN-RND Registry

ERN	ERN registry project
ERNICA	ERNICA registry
ERN EYE	REDgistry
ERN EpiCARE	EPICARE-GRANT
VASCERN	VASCERN Registries
ERN-Skin	ERN-Skin REGISTRY
ERN EURO-NMD	EURO-NMD Registry
ERN GENTURIS	GENTURIS registry
ERN RARE-LIVER	R-LIVER
ERN RITA	MERITA
ERN TRANSPLANT-CHILD	PETER
ERN EuroBloodNet	ENROL
ERN eUROGEN	ERN-eUROGEN registry

Joint Research Centre
JRC

European Platform on Rare Disease Registration (EU RD Platform)

Searchable, findable rare disease registry data

https://eu-rd-platform.jrc.ec.europa.eu/erdri_en

JRC - EU RD Platform
SPIDER ISO 27001 certification

- Scope of Information Security Management System
- Information security policy
- Information Security risk assessment process
- Statement of Applicability
- Information Security risk treatment process
- Information Security objectives
- Evidence of competence of persons
- Operations and Planning
- Results of Information Security risk assess
- Results of monitoring and measuring
- Audit programme and audit results
- Management Review
- Nonconformities and their management
- Documents stated as necessary for the organization

ISO/IEC 27001 CERTIFICATION for SPIDER - TIMELINE

07 2022 - Definition of the Scope of Security

09 2022 ongoing
Preparation of policies/manual/trainings/statement of applicability/information security controls/management review etc. a complex process involving a number of internal and external services

- Internal audit - part 1 October 2024
- Internal audit - part 2 - November 2024

External Audit - Phase 1 - 2nd trimestre 2025

External Audit - Phase 2 - 4th trimestre 2025

Successful - Non conformities detected

- Lengthy process with limited additional resources while guaranteeing full functioning of the EU RD Platform, its security, further development and service to the registries.
- Interactions with many Commission's/JRC's departments - LISO (Informatics Security Office), IPO (ICT Programmes Office) / IT Security Coordinator's team, LSO (Local Security Office) etc.

This is the first system in the JRC to obtain the ISO/IEC 27001 Certification

European Commission

D. Conclusion

We can't yet predict the 'full legacy' of ERICA since it will also depend on how different ERN teams take ownership of and roll out ERICA work.

But we know already:

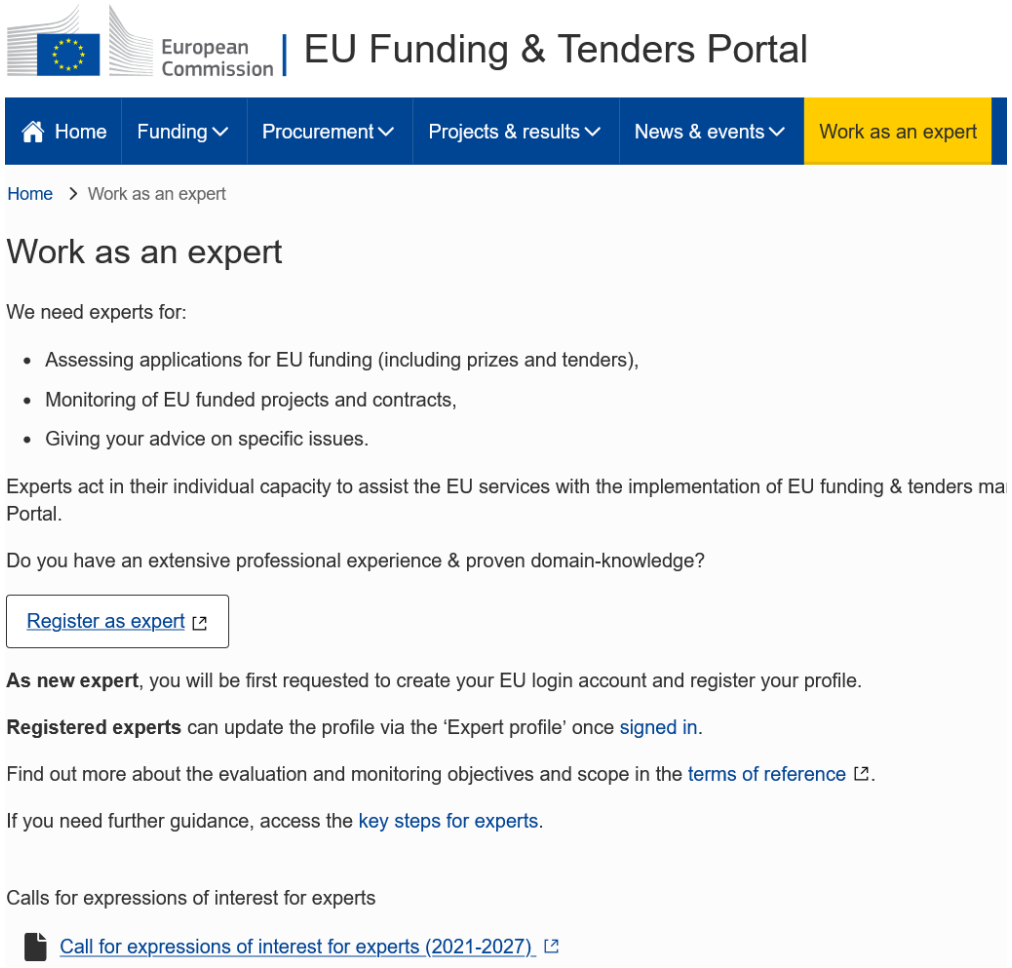
- that ERICA has allowed you to start acting collectively on ERN research across ERNs and involving all ERNs ! (be proud)

- that the following projects and initiative will benefit from ERICA efforts: (go on…)

    - ERDERA, various work streams (think also about the 2nd 'instalment' of ERDERA…)
    - RealiseD (IHI project – thus public-private, and with ERNs engaged !)
    - ERN registries
    - Together4RD – relations with industry
    - many more research projects on (groups of) rare diseases… (it can continue to grow !)

In addition…

Don’t hesitate to register to our Experts’ database to evaluate proposals submitted under various EU funding programmes, such as EU Research & Innovation Programme Horizon Europe



https://ec.europa.eu/info/funding-tenders/opportunities/portal/screen/work-as-an-expert

We are constanly looking for new experts to be involved in the evaluation and assessment of EU-funded projects and contracts…

and this would certainly also help you to better understand criteria and processes for EU funding.

European Commission

Thank you – and let's keep in touch

ec.europa.eu/

hadea.ec.europa.eu

europeancommission

europa.eu/ https://research-and-innovation.ec.europa.eu/research-area/health/rare-diseases

https://health.ec.europa.eu/rare-diseases-and-european-reference-networks/european-reference-networks
https://health.ec.europa.eu/medicinal-products/orphan-medicinal-products
https://eu-rd-platform.jrc.ec.europa.eu

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