ERDERA

European Rare Diseases
Research Alliance

Making Europe a world-leader in rare diseases research and innovation

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INSERM, France

ERDERA JTC 2025 Information Webinar
17 of December 2024
Online

Co-funded by
the European Union

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Why and what is ERDERA?

Why a partnership for rare diseases

- There are more than **7000** different rare diseases, affecting **30 million** people **million** people in Europe.

- Each of us knows someone in their close environment, family or work, living work, living with a rare disease, sometimes we don't even know they have have such a disease.

- For those people, in Europe, on average, **it still takes 4 years to get diagnosed**. **diagnosed**. During that time, they may be misdiagnosed and shuffled between shuffled between doctors without clear answers.

- When the diagnosis finally comes, they find that **no treatment exist for over for over 90% of rare diseases**—or if they do, they might not be available in available in their home country.

ERDERA's Mission

ERDERA aims to change this by **bringing together the strengths of academia, academia, industry, and healthcare to make diagnosis faster, treatments more treatments more accessible, and quality of life better for people living with rare with rare diseases**. With ERDERA, we harness the power of research and technology, and we actively engage people living with these diseases, so their so their needs drive our mission.

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)

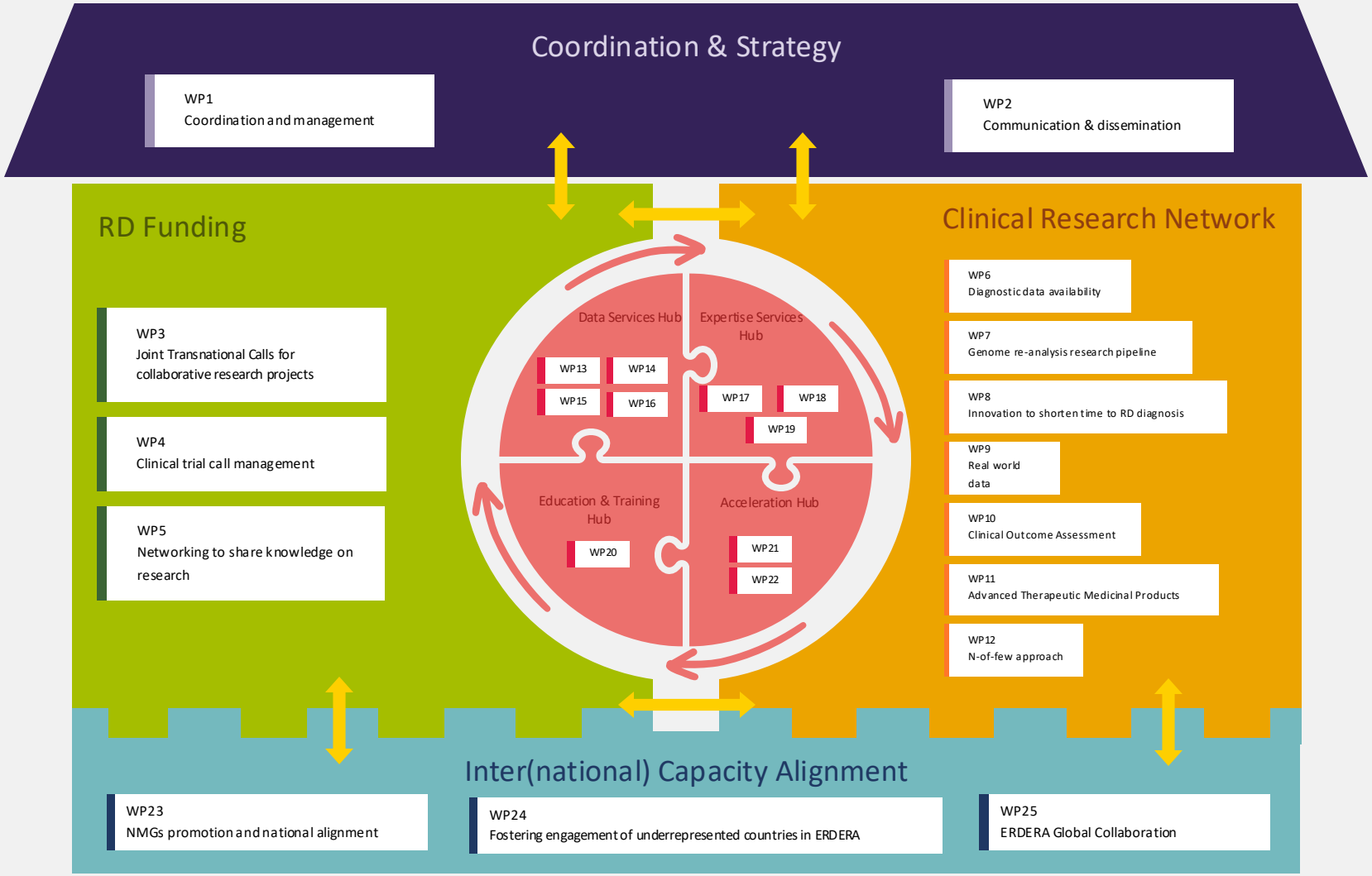
37 Countries

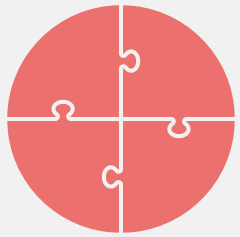
26 EU member states

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

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Information Webinar

ERDERA Joint Transnational Call 2025

Pre-clinical therapy studies for rare diseases using small molecules and biologicals – development and validation

Ralph Schuster, DLR Projektträger
17 December 2024

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Agenda

14.00-14.10	**Welcome and Introduction to ERDERA**	**(Daria Julkowska)**
14.10-14.30	**JTC 2025 Introduction**	**(Ralph Schuster)**
14.30-14.40	**Patient engagement in research**	**(Roseline Favresse, Avril Kennan, Laura Benkemoun)**
14.40-15.00	**Q&A Round 1 (*time to submit questions!*)**	
15.00-15.20	**ERDERA expertise support services**	**(Rima Nabbout, Viviana Giannuzzi, Toni Andreu)**
15.20-15.30	**Data Hub and FAIR data**	**(Marco Roos)**
15.30-15.50	**Q&A Round 2 (*time to submit questions!*)**	
15.50-16.00	**Closing remarks**	**(Ralph Schuster)**

Aim of the call - **Pre-clinical therapy studies**

- The aim of the call is to enable scientists in different countries to build an effective collaboration on a common interdisciplinary research project based on complementarities and sharing of expertise, with the expected impact being future use of the results to benefit patients.

- Projects should focus on **a group of rare diseases or on a single rare disease if there is no valid rationale/evidence for the benefit of grouping diseases.** If focusing on a single rare disease, applicants must clearly specify why working on a group of rare diseases is not appropriate. The classification of rare diseases follows the European definition, i.e. a disease affecting not more than **five in 10.000 persons** in the European Community, EC associated states, and Canada.

- Translatability into humans should be the key focus of the projects, and applicants should demonstrate access to relevant scientific or regulatory expertise (e.g., through innovation task forces or competent national authorities).

Call topic: **Pre-clinical therapy studies for rare diseases using small molecules and biologicals – development and validation**

Research studies on therapies using small molecules, small non-coding chemically synthesized nucleic acid-based therapies, repurposed drugs or biologicals (e.g., antibodies or proteins such as enzymes, immune modulators or growth factors etc.). Proposals must cover **at least two** of the following areas**:**

1. development of novel therapies in a pre-clinical setting through cell, organoid and animal disease model studies, and/or use of *in silico* or artificial intelligence models to accelerate the success rate of the pre-clinical stage
2. development of predictive and pharmacodynamics biomarkers correlated to the efficiency of the therapy in a preclinical setting that could serve as surrogate endpoints
3. replication of pre-clinical studies in an independent lab to increase validity of exploratory findings
4. pre-clinical proof of concept studies for evidence of pharmacological activity *in vitro* and *in vivo*, pharmaco-kinetics and pharmaco-dynamics of the investigational drug (i.e., small molecule(s) and/or biologic) and first toxicology and safety data as well as studies to support readiness for initiating clinical trial authorization conforming to regulatory requirements

Excluded Approaches and Topics

- ATMP therapies (gene therapy medicinal product (including mRNA-based therapies), somatic cell therapy medicinal product, tissue engineered product, according to EMA definition).
- Development of new cell/organoid/animal models, which should already be established.
- Set-up or extension of natural history studies / patient registries.
- Interventional clinical trials to prove efficacy of drugs/treatments/surgical procedures/medical procedures. This includes studies comparing efficacy, e.g., two surgical techniques or therapies, and projects whose main objective is the implementation of a clinical phase IV pharmacovigilance study.
- Projects focusing only on rare neurodegenerative diseases that are within the focus of the Joint Programming Initiative on Neurodegenerative Disease Research (JPND). These are: Alzheimer's disease and other dementias; Parkinson's disease (PD) and PD-related disorders; Prion diseases; Motor Neuron Diseases; Huntington's disease; Spinal Muscular Atrophy and dominant forms of Spinocerebellar Ataxia. Interested researchers should refer to the relevant JPND calls. **However, childhood dementias/neurodegenerative diseases are eligible.**
- Rare infectious diseases, rare cancers and rare adverse drug events in treatments of common diseases. **Rare diseases with a predisposition to cancer are eligible.**

General considerations 1

- Projects should focus on rare diseases or disease groups with high unmet medical need, high disease burden, and no currently approved therapeutic options in Europe (European marketing authorisation). Preferably, they should address group(s) of rare diseases with commonalities such as, but not exclusively, shared molecular etiologies and/or clinical symptoms, such that the same drug and/or drug combinations could be used for clinical trials of multiple diseases
- Existing knowledge from multiple sources (natural history studies/registries, real-world data/evidence, multi-omics, medical imaging, etc.) should be used to underpin the therapeutic hypothesis and therapeutics development.
- Consortia **performing preclinical development of therapeutics** are strongly advised to engage or consult experts in the various stages of product development to ensure that the data generated is suitable for future regulatory filings such as for application of receiving orphan designation and/or clinical trial preparedness for regulatory advice and authorisation → establish one or more: **Target validation, Suitable formulation and route-of-administration, Right Tissue, Right safety profile, Right patient, Readiness for clinical trial application (CTA)-directed studies**

General considerations 2

- **For the development of novel therapies or pre-clinical proof-of-principle studies** → Orphan medicine designation (OD) planning, EMA Scientific Advice Working Party (SAWP) and/or Innovation Task Force (ITF) early engagement, target validation in relevant preclinical disease and/or models
- **Validation or development of predictive and pharmacodynamics biomarkers** → Robust analytical procedures, analytical validation using high quality samples from an independent collection, should follow a risk-based approach
- **Describe and justify the use of disease models** → how the model replicates the pathology or human condition as well as aspects of the therapy target, justification on use of animals, availability of the model, statistics for robust and well controlled pre-clinical efficiency studies, primary endpoints
- The **design of the study** (sample collection, statistical power, interpretation, relevant models for hypothesis validation) must be well justified and should be part of the proposal.
- Study design, preclinical models and reagents should be selected **to facilitate approval in human trials** and future clinical grade manufacturing.

General considerations 3

- Appropriate bioinformatics and statistical methods, whenever included and justified, should constitute, an integral part of the proposal, and the relevant personnel should be clearly specified. These personnel should either be an eligible partner of the consortium, part of the research group of an eligible partner or involved as direct contractors of an eligible partner. They cannot be external collaborators that participate with their own funding. **Their responsibilities must be clearly described and align with the requested resources and a CV must be provided.**
- Data generated or newly collected for the project must be made ready for reuse according to **FAIR principles**. This should be achieved by contributing to the creation of the ERDERA Data Hub, a collaborative responsibility of the ERDERA partnership. Effort and budget must be earmarked for FAIR data stewardship and a milestone should be included to mark the contribution.
- Risk management should be considered including the identification of possible bottlenecks and go/no go contingencies.
- The analysis of IP status, freedom to operate and access to therapeutic molecules for development should be clearly described.

Eligible countries/regions and budget

- **35 funding agencies from 27 EU, EU associated countries & Canada, co-funded by European commission**
- **Participating countries:** Austria, Belgium, Bulgaria, Canada, Cyprus, Czech Republic, Denmark, Estonia, France, Germany, Hungary, Iceland, Ireland, Israel, Italy, Latvia, Lithuania, Luxembourg, Norway, Poland, Portugal, Slovakia, Spain, Sweden, Switzerland, The Netherlands, Türkiye
- **Each funder funds only their respective national/regional teams → check guidelines!! → contact your national/regional contact point!!**
- **Partners from other countries (e.g. UK, USA, China) can only participate as collaborators with own funding**
- **3 years projects**
- **Overall budget: 32,6 Mio €**
- **Expected number of funded projects: ~25**
- **Usual success rate: from pre-proposal stage→funding 10-15%, pre-proposal→ full proposal 30%, full proposal→ funding 40-50%**

Consortium partners and composition 1

- **Categories of Partners** (depending on national/regional regulations)
  - Academia (research teams working in universities, other higher education institutions or research institutes),
  - Clinical/public health sector (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).

- **Consortium Makeup**
  - **four to six** eligible principal investigator **partners** from at least **four different participating countries**
  - **One of these four to six partners must be an Early Career Researcher** (ECR)
  - **The number of partners can be increased to 8 in two cases:**
    - The inclusion of partners from participating countries usually underrepresented in projects (UR: Czech Republic, Estonia, Latvia, Lithuania, Slovakia, Türkiye), OR
    - The inclusion of an additional ECR as full partner (see section 5.6).
  - **Patient advocacy organizations** do not count toward the total number of partners in the consortium
  - No more than **2 eligible partners from the same country** in a consortium (Further national/regional limits may apply)

Consortium partners and composition 2

- **What is a partner? a collaborator? a sub-contractor?**

  - **Partner:** a group must contribute substantially to at least one of the project's work packages. If the only role of a group is to provide patient access, data or samples for the study, they will not be considered as partners of the consortium, but can be included otherwise, via cooperation agreements (as collaborators) or subcontracting.

  - **Collaborator:** research groups that secure their own funding, can come from a non-participating country. Collaborators cannot be work package leaders, and their contribution to the consortium must be described. As they do not receive funding as part of this call, they do not count toward the limit of 8 partners requesting research funding (nor is there a limit of collaborators per country, as long as their participation is justified). Collaborators must supply a letter of intent and a CV as well as be entered in the online submission system.

  - **Sub-contractor**: may cover only a limited part of the research activity, and their contribution to the consortium must be described. They do not count toward the limit of 8 partners requesting research funding (nor is there a limit of subcontractors per country, as long as their participation is justified and if subcontracting is possible according to national/regional funding rules).

Early Career researchers (ECR)

- At least one **Early Career Researcher** must join a consortium as a full research partner and is therefore subject to the same eligibility criteria as other partners.
- **In general, ECRs can either be PhD holders or medical doctors.**
- **PhD holders**: Scientists who have received their PhD no more than seven years prior to the application deadline.
- **Medical doctors**: Physicians who have completed specialist medical training no more than seven years prior to the application deadline. For physicians with a PhD, the date of the completed specialist medical training remains the relevant date.
- **Extensions** to this period are allowed in case of reasonably justified career breaks: absence for parental leave, family care leave, long-term sickness leave, and compulsory military service.
- **National/regional eligibility criteria**, definitions and time limits might differ → refer to national guidelines and contact your national/regional funder
- **Non-eligibility of the ECR due to not conforming with the eligibility date requirements will lead to exclusion of the whole consortium from the application process.**

Patient Advocacy Organisations

- 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.

- 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.

- Depending on their activities within the consortium and on the specific guidelines from each funding agencies, PAOs from participating countries can participate as partners, collaborators or sub-contractors

- PAOs representative of rare disease patients within a Member State or throughout the EU/EEA can be funded through **central funding mechanism**, max 25.000 € per project – please check guidelines!

Application Timeline

Call opens

December 10, 2024

Informational webinar

December 17, 2024

Add. Webinar Jan 14, 2025

Pre-proposal submission

February 13, 2025

Eligibility check

Early March 2025

Invitation full proposal

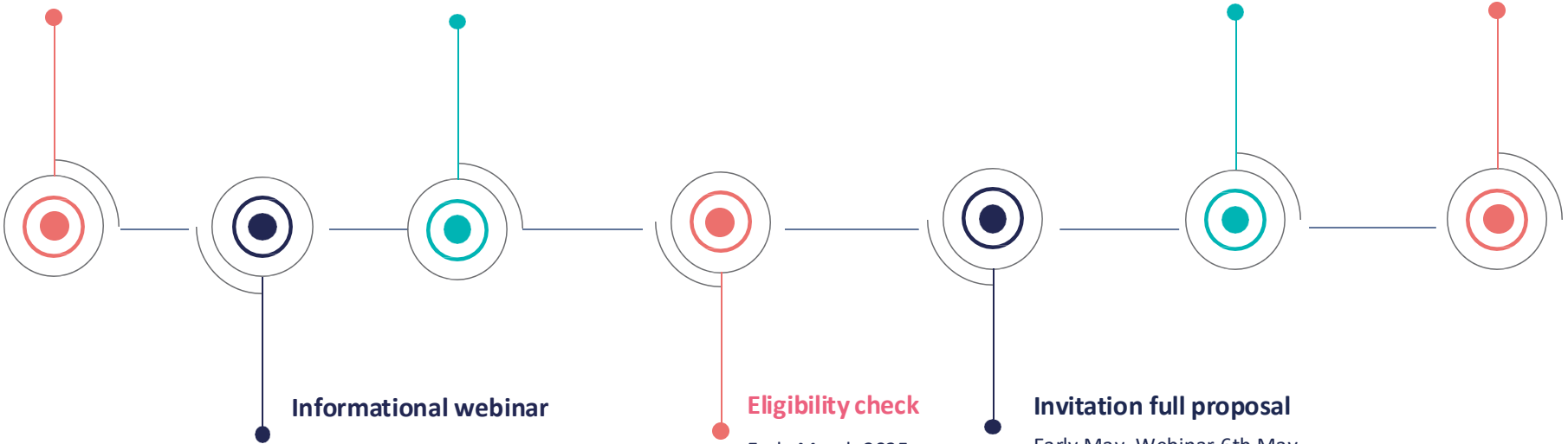
Early May, Webinar 6th May

Full proposal submission

July 9, 2025

Decision

Dec 2025



Registration and submission

- One joint pre-proposal (in English) to be submitted by coordinator to JCS via the electronic submission system: https://funding.erdera.org **no later than February 13th, 2025 at 2:00 p.m. (CET)**
- **Platform to open latest by January 13, 2025**
- **Call text, guidelines, PAO declaration of honour and online pre-proposal submission form preview available for download at https://erdera.org/funding/**
- **PRE-PROPOSAL SUBMISSION FORM PREVIEW ONLY PROVIDES INFORMATION ABOUT THE SUBMISSION PLATFORM. IT IS NOT INTENDED TO BE FILLED OUT AND UPLOADED. ONLY PROPOSALS THAT HAVE BEEN ENTERED ELECTRONICALLY AT FUNDING.ERDERA.ORG WILL BE EVALUATED!**
- **CHECK PREVIEW, PREPARE NECESSARY DATA, ONLY PLAIN TEXT CAN BE ENTERED FOR DESCRIPTIONS. CHARACTER RESTRICTIONS MUST BE RESPECTED!**
- **Documents to upload in the platform**
  - Diagram of the work plan (Timeline, workflow and interconnections of work packages (Gantt chart, Pert or similar, max. 1 page))
  - Diagrams, figures, tables etc. to support the work plan description (max. 2 pages)
  - List of references
  - Date and signature page of all project partners asking for funding
  - Letter of intent for collaborators
  - Declaration of honor for PAOs
- ***Eligibility pre-check / parallel submission necessary for some funding agencies***

Description of the project

Introduction and background (max. 4.500 characters)

Project description (max.13.500 characters)

- **Objectives and hypothesis**
- **Soundness and pertinence**
- **Workplan and Methodology**

Impact (max. 1.500 characters)

Added value of the consortium (max. 1.500 characters)

Patient Advocacy Organisations (PAOs) engagement/involvement (max. 2.000 characters)

Results of previous EJP RD or E-Rare funded project, only if applicable (max. 4.500 characters)

Participant information

Narrative CVs

Evaluation

- **Scientific Evaluation Committee (SEC)** consisting of internationally recognized, independent, scientific experts from the fields of biomedical science (e.g. basic research, clinical research, translational research, drug development, regulatory, animal/cell model expertise) plus experts in patient engagement (full proposal only)
- **Evaluation criteria** according to Horizon Europe for **pre-proposals:**
  - **Excellence**
    - Clarity and pertinence of the objectives;
    - Credibility of the proposed approach and methodology;
    - Soundness of the concept;
    - Innovative potential:
    - Competence of participating research partners
  - **Impact**
    - Potential and readiness of the expected results to be translatable for future clinical applications
    - Benefit to patients, their families, and carers;
    - Added value of transnational collaboration:
  - **Implementation**
    - Feasibility of the project
    - Complementarity of the participants within the consortium, including the integration of PAOs or patient partners;
- **Scoring** – see call text
- **Full proposal**: additional criteria, independent ethical evaluation

Frequently asked questions 1

- Can research groups from countries not participating in the call (e.g. UK, USA, China etc.) receive funding?
  - No, they can only particpate as collaborators with their own funding.
- Can Patient Advocacy Organisations from USA receive funding?
  - No, only PAOs representative of rare disease patients within a Member State or throughout the EU/EEA can be funded through central funding mechanism. Other groups only with own funding or subcontractors depending on national/regional regulations.
- Is there a partnering platform for this call?
  - Unfortunately, there is no partnering platform available for this call. It may be offered for future calls.
- Is there a restriction of how many proposals a research group can be part of?
  - There is no general restriction, however some national/regional restrictions may exist → please consult guidelines.
- Is disease xyz considered a rare disease?
  - Refer to definition „disease affecting not more than **five in 10.000 persons** in the European Community, EC associated states, and Canada", consult Orphanet or scientific literature for prevalence data
- Is research on rare cancers eligible for funding?
  - No, rare cancers are excluded from the call. Please check TRANSCAN and EC calls (e.g Cancer Mission etc.) for funding opportunities.
- Are gene or cell therapy approaches eligible for funding?
  - No, therapy approaches that fall in the ATMP category are not eligible. These might be subject of a future call.
- Are therapy approaches like ASOs, siRNA, suppressor tRNAs eligible?
  - Yes, if the fall under the definition of „small non-coding chemically synthesized nucleic acid-based therapies".

Frequently asked questions 2

- Do you fund validation or other forms of research involving intervention in human subjects?
  - No, only pre-clinical research is funded.

- *"Projects should focus on rare diseases or disease groups with high unmet medical need, high disease burden, and no currently approved therapeutic options in Europe (European marketing authorisation)"* – can we also apply for therapies in diseases that do have approved therapeutic options?
  - Yes, that is possible, but the high unmet medical need in comparision to already existing therapy options needs to be demonstrated.

- Do projects need to address a group of diseases or can the also focus on a single disease?
  - Applicants are strongly encouraged to work on groups of rare diseases with commonalities and assemble criteria of meaningful grouping of the rare diseases under study, based on state-of-the-art scientific discoveries and clinical practice, to achieve higher global impact, but single rare diseases are possbile as well with specification why working on a group of rare diseases is not appropriate.

- What are the budget/eligibility criteria for the different national/regional funders/ PAO funding?
  - Please check guidelines and/or contact your local funder.

- How many months can ECR candidates add to the cut off date of seven years for maternity/paternity leave?
  - As many as they actually took for the leave.

- What is the cut-off date for the ECR seven years qualification?
  - Submission deadline of pre-proposal, 13th February 2025.

- Where can I find information on previously funded projects in similar calls?
  - Please visit the EJP RD Website: https://www.ejprarediseases.org/our-actions-and-services/funding-opportunities/funded-projects/ and https://funded-projects.ejprarediseases.org/

European Rare Diseases Research Alliance

Contact Joint Call Secretariat

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Patient partnerships in preclinical studies

Why and how involving patients in research from the start?

Roseline Favresse, EURORDIS-Rare Diseases Europe
Laura Benkemoun, Foundation for Rare Diseases
Avril Kennan, Health Research Charities Ireland

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JTCs partners

- Partners belonging to one of the following categories may request funding:
  - Academia
  - Clinical/public health sector
  - Enterprises
  - **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.
- Funding for PAOs is limited to a total of **25,000 € over 3 years**.
- PAOs can also be involved through national/regional funding or subcontracting depending on the proposed tasks and national/regional funding rules.

What is a Patient partner / Patient Advocacy Organization?

Who can apply for funding?

- Patient Advocacy Organisations (PAO) only.

Definition of rare disease patient advocacy organisations:

- Patient advocacy organisations are defined as not-for-profit organisations, which are patient focused, and where patients and/or carers and/or family members of patients represent a majority of members in governing bodies. These are:
  - Umbrella organisations (e.g. representing either European organisations and/or national umbrella organisations for rare diseases).
  - European rare disease specific organisations (i.e. representing national organisations or individual patients on rare diseases) and
  - National rare disease specific organisations.

How do I find a Patient partner / Patient advocacy organisation (PAO)?

Existing databases and directories

- EURORDIS – Rare Diseases Europe
- ORPHANET – Directory of patients' organisations
- Rare Diseases National Alliances
- European Patient Advocacy Groups (ePAGs)

What if there are several patient organisations in place for the same disease?

→ Check if a European federation/association exists and is active for the disease

→ Try to identify the patient organisation that is more involved in research activities

What if there is no patient organisation for the disease we target?

→ Check with the relevant umbrella organisations (there may be an association in a different country)

→ There is still an opportunity to involve an umbrella organisation that may be active in the group of diseases you target

→ Foresee the involvement of individual patients in activities at some point of time if there is no structured organisation in place to advise/co-design

PPIE, what it is?

Patient and Public Involvement and Engagement: working **with** patients and the public to shape research and engage with it.

Patient and public involvement and engagement (PPIE) describes the **different ways in which members of the public can inform and shape research.**

"PPIE is different to research participation where members of the public can directly take part in a study, for example by being given a new treatment as part of a clinical trial." – University of Birmingham

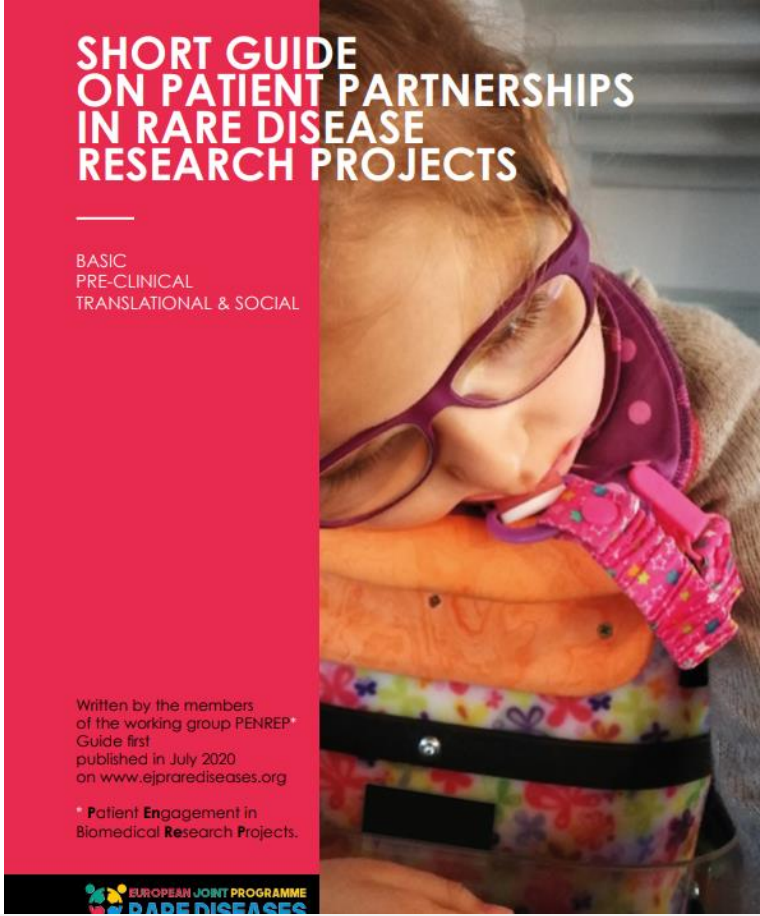
*"Patient and public involvement (PPI) entails **research being carried out 'with' or 'by'** members of the public, rather than 'to', 'about' or 'for' them"*- National Institute of Health Research

PPIE is for **everyone.**

What is patient partnership? The PENREP Short Guide

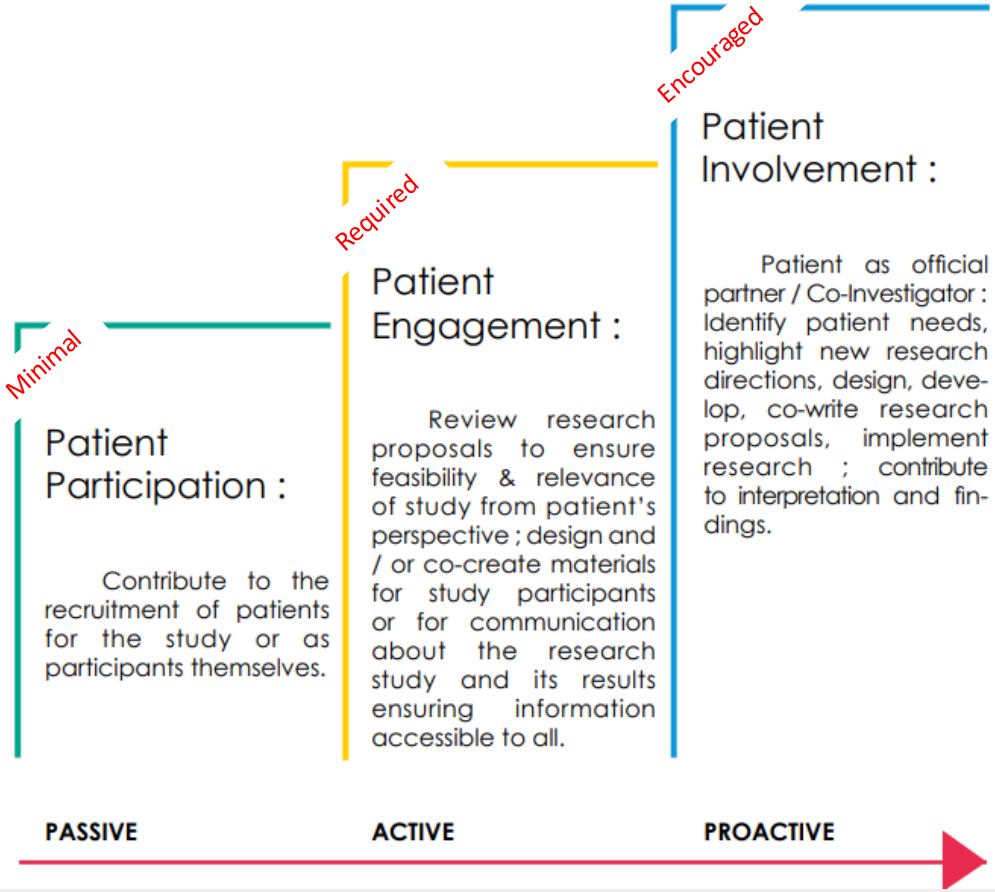
- Defines what patient partnerships are
- Defines the benefits of involving patients
- Provides examples of concrete patient partnerships
- Highlights the common pitfalls that could be avoided
- Provides an indicative checklist for self-evaluation
- Embeds testimonies from successful applicants

https://www.ejprarediseases.org/wp-content/uploads/2021/03/SHORT-GUIDE-ON-PATIENT-PARTNERSHIPS-IN-RARE-DISEASE-RESEARCH-PROJECTS.pdf



A continuum of active and meaningful partnership

Depending on the project and its state of development, all these options may happen concomitantly or iteratively/progressively



Patient partnerships' benefits

STRONGER FUNDING APPLICATIONS

EXPANDED OUTREACH & IMPROVED COMMUNICATION

Patients can assist in the creation of communications, translating information into accessible language to reach a wider community more efficiently.

GREATER RELEVANCE

Involving patients ensures that researchers demonstrate accountability of public money investment as research results translate into concrete benefits and address patients needs.

NEW IDEAS

MOTIVATION & FOCUS

Hearing directly from people living with a specific RD can provide you and your project with meaning and context.

GREATER IMPACT

Patient partners are excellent advocates to generate public interest and impact, raise awareness of the research and facilitate further funding through collaborations with charities.

Succeeding in partnerships between patient organisations & researchers

- What should a good partnership look like?
- Understanding the charity perspective
- Understanding the researcher perspective
  - Recommendations for charities & other civil society organisations
  - Recommendations for researchers
  - Recommendations for universities and other research institutions
  - Recommendations for research funders

https://hrci.ie/hrci-ppi-ignite-network-charities-researchers-partnering-guide/

Patient engagement in early discovery? The PFMD Guide

- Prepare the partnership- Ensure that long term partnerships with patients are created and nurtured
- Understand the condition profile - Define unmet needs
- Develop research methodology - Identify optimal approaches to address research objectives
- TPP and Target Patient Value Profile - Create a target value profile that represents the patients' perspective on the product

https://pemsuite.org/How-to-Guides/Early-Discovery.pdf

Avoiding common pitfalls (examples)

" Patient organisations will recruit patients as donors for the biobank . "

Not enough explanation is given as to how this will be achieved. Who ? How ? When ? Was the patient organisation involved in developing the recruitment strategy ?
If involvement / engagement activities are not planned, please provide an explanation as to why it was not possible in this project.

"Patient and public involvement (PPI) entails research being carried out 'with' or 'by' members of the public, rather than 'to', 'about' or 'for' them"- National Institute of Health Research

" The applicants are in contact with patients and patient organisations so patients will be engaged / involved throughout the research project "

" Patient organisations will be responsible for disseminating the research results to their communities "

Any specific roles and responsibilities need to be discussed and agreed between the researchers and the patient organisations (or patients) before submitting the proposal and need to be detailed in the proposal.

Generic statements are not useful to evaluators and need to be expanded to include the descriptions of the responsibilities of the different partners.

« There is actualy no cure so our project answers to an unmeet medical need »

Gain insights on the current therapies patients are using and identify how this drug might fit in or impact the patient. Understand the comorbid conditions. Ask about the effect the medicine should have on the condition and what might be the preferred frequency the medicine could be taken. A side effect might be tolerable for 3 weeks and another for 3 years. Discuss with patients to understand what is important to them.

Checklist for self-evaluation of applicants (extract)

- ☐ *Have discussions between researchers and patient representatives taken place before identifying the research questions and writing the proposal ?*
- ☐ *Have you described how the patients/patient representatives were identified and selected ?*
- ☐ *Has the input of patients/patient representatives been integrated in the development of the proposed research project ? Have you described what changed / improved as a result of this input ?*
- ☐ *Have clear roles and responsibilities been assigned to the patients / patient representatives in the project ?*
- ☐ *Have the Patient Partnership activities been clearly explained (who, what and when) ?*
- ☐ *Have the available resources of respective partners been maximised to the benefit of the research project (e.g. registries, know-how, networks, communication channels) ?*
- ☐ *Have the approaches through which the patients / patient representatives will be engaged / involved / participate in the project been described (e.g. focus groups, interviews, surveys etc.) ?*
- ☐ *Has a process been included to ensure two-way communication between the partners throughout the life of the project ?*

European Rare Diseases Research Alliance

Thank you!

Laura Benkemoun - laura.benkemoun@fondation-maladiesrares.com

Avril Kennan - Avril@hrci.ie

Roseline Favresse - roseline.favresse@eurordis.org

On behalf of all ERDERA patient partners

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ERDERA
European Rare Diseases
Research Alliance

Making Europe a world-leader in rare diseases research and innovation

ERDERA expertise support services

Viviana Giannuzzi, Toni Andreu, Rima Nabbout

JTC 2025 – 17 December 2024

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Expertise Workstream

- Data Hub
- Acceleration Hub
- Education & Training Workstream
- Ethics Advisory Group

Mentoring/Consultancy service
WP17

Regulatory Support Service
WP18

Methodological Support
WP19

EXPERTISE SERVICES HUB

Know-how and **support** on specific aspect of translational research and clinical studies

Efficient execution and transability of results and innovation of research activities

Patient needs targeted and less costly development process for clinical implementation of products for RD

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Mentoring service

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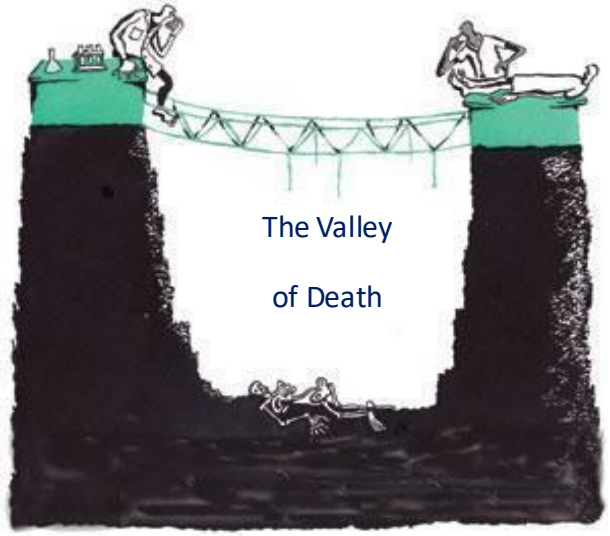
Mentoring service

Improve the feasibility of translational and clinical research projects



Nature, 2008

Challenges of translational projects:

- Poor predictive pre-clinical models
- Mode of action not fully validated
- Sub-optimal clinical trial design
- IP not secured – no freedom to operate
- Limited regulatory experience
- Limited data management knowledge

Execution of the Mentoring service

- **To Whom**: Shortlisted JTC's proposals and also projects from other funding schemes.
- **Format**: Webinar and 1-on1 meetings with expert mentors who provide advise on specific areas
- **Cost**: Free of charge, full confidentiality (signed Letter of engagement/CDA)
- **When**: During 2nd stage to prepare the final proposal , through full project lifetime

Mentoring and application Timeline

Call opens

December 10, 2024

Informational webinars

December 17, 2024

January 14, 2025

Pre-proposal submission

February 13, 2025

Shortlisting

April, 2025

Kick off Webinar

May 6, 2025

1-on-1 meetings

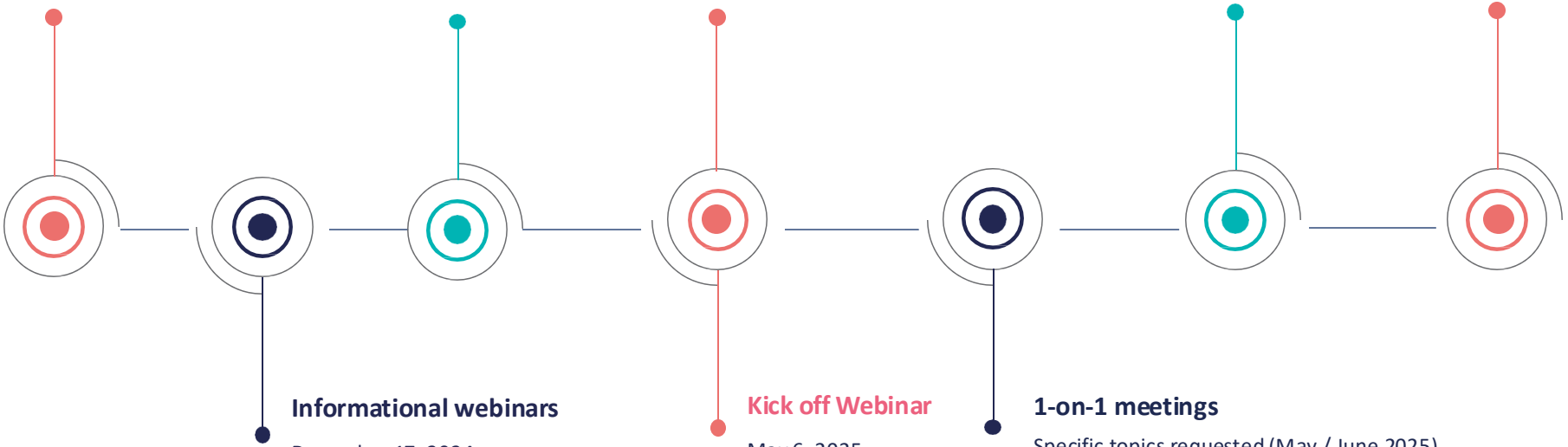
Specific topics requested (May / June 2025)

Full proposal submission

July 9, 2025

Decision

Dec 2025



Testimonial

Davide Gabellini
Group Leader Division of Genetics and Cell Biology
San Raffaele Scientific Institute

EJPRD JTC 2020 participant

During a period of about one month, I interacted with various professionals collaborating with EATRIS to discuss issues related to ***preclinical models; medical statistics; technology transfer, industrialisation and intellectual property; regulatory affairs***.

The support has been ***professional, timely, creative, flexible and accurate.*** *Always ready to accommodate any request for the benefit of the project. All of this while maintaining a friendly and positive attitude.*
Thanks also to the mentoring support, ***my application was funded.***
In summary, the ***mentoring professionals are well trained, honest, patient and meticulous***. *I believe they are an ideal choice for mentoring service provider."*

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Regulatory Support Service

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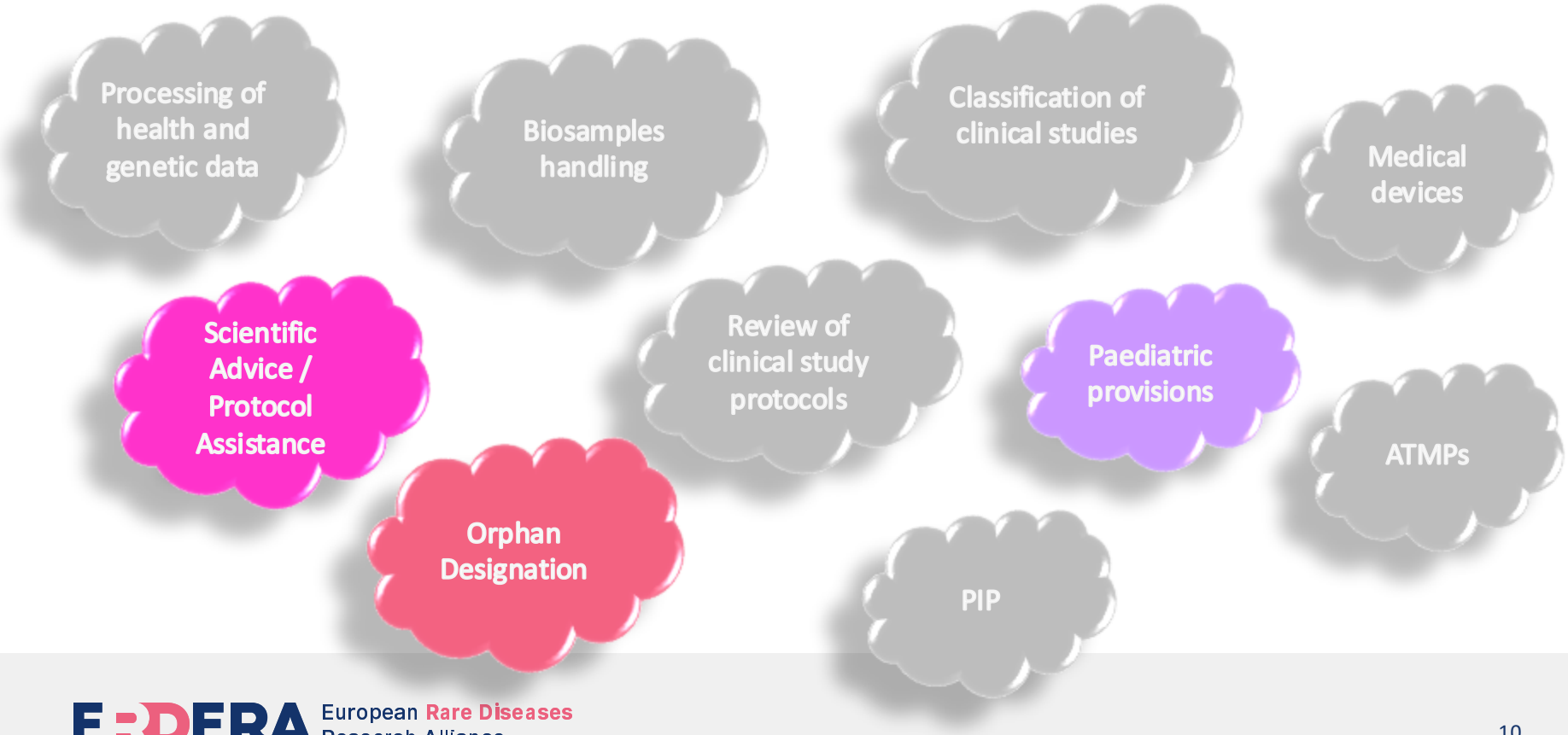
Regulatory support service

Facilitate the engagement with regulatory agencies

Preparatory activities to help identify the most suitable regulatory procedures

Regulatory issues relevant for RD community



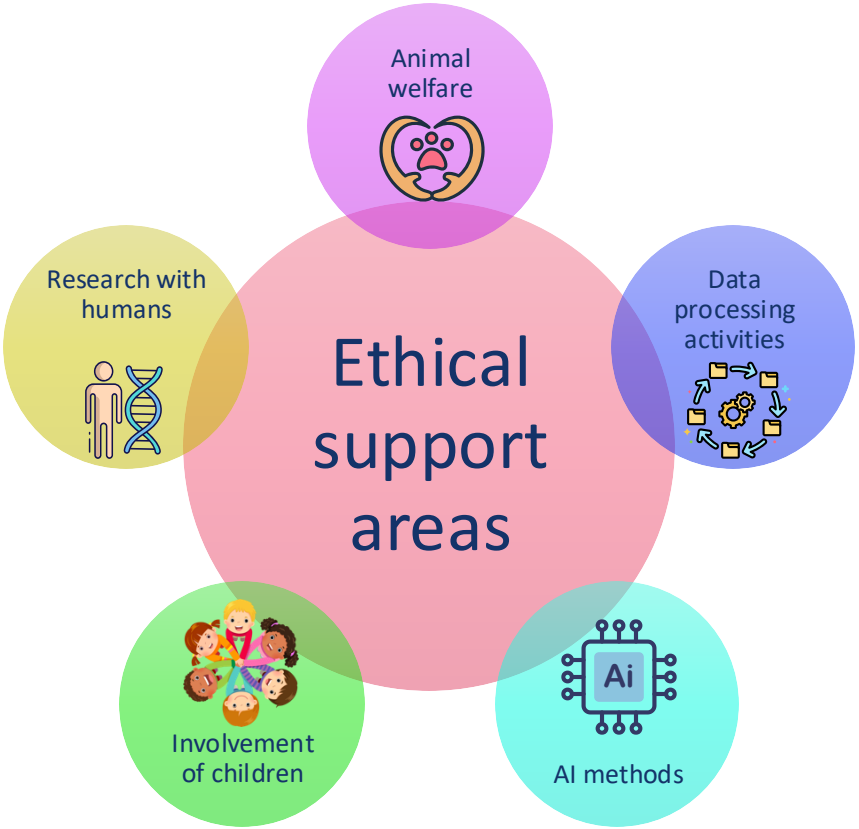
Regulatory support for preclinical studies

- EMA Orphan Designation
- Scientific Advice / Protocol Assistance & Qualification of novel methodologies for medicine development (pharmacometrics, innovative trial designs, RWD and registries, biomarkers/endpoints)
- EMA Innovation Task Force (ITF) Briefing Meeting with the EU-Innovation Network for innovative products
- Paediatric provisions, Paediatric Investigation Plan (PIP)

Ethics Advisory Group

To **guarantee and support ethical compliance in all project activities** during the implementation and throughout their research phases

Through the ethics follow up



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Methodological support

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Challenges in Preclinical research

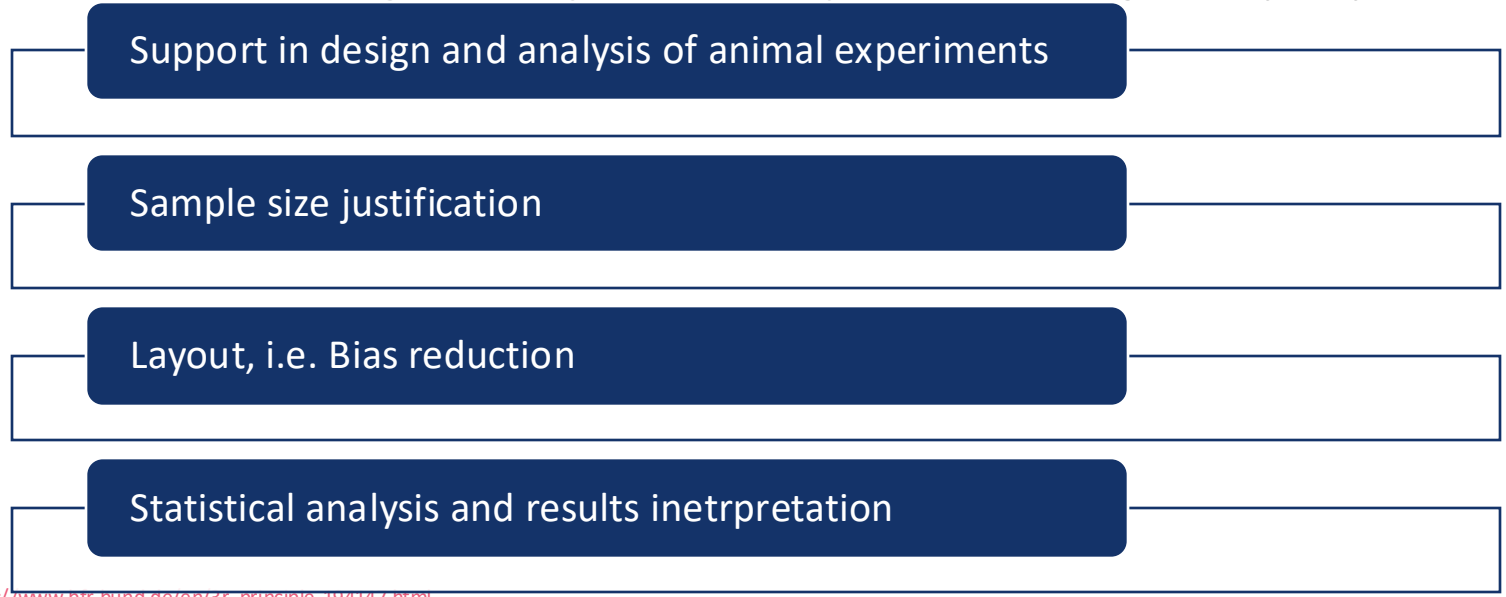
The valley of death….

- **Translation from models to human**: Animal studies do not reliably predict Human Outcomes
- **Translation of drug efficacy:** 9/10 Drugs that appear promising in animal studies go to fail in Human Clinical Trials
- **Data integration and analysis:** a major gap in data intergration and analysis of in vivo and in silico models
- **Design:** Animal studies are flawed by design

https://aavs.org/animals-science/problems-animal-research/

Support in ERDERA from WP19

- **Mentoring in Cooperation with EATRIS WP17 with respect to design and analysis of preclinical trials**
- **Consultation in Design and Analysis of Animal Experiments reflecting the 3R principle with respect to:**



https://www.bfr.bund.de/en/3r_principle-194147.html

Begley, C., Ellis, L. Raise standards for preclinical cancer research. *Nature* 483, 531–533 (2012).

van der Worp HB, Howells DW, Sena ES, Porritt MJ, Rewell S, O'Collins V, et al. (2010) Can Animal Models of Disease Reliably Inform Human Studies? PLoS Med 7(3): e1000245.

Support and expertise Workstream welcomes you!

Mentoring/Consultancy service
WP17

EATRIS
Toni Andreu
toniandreu@eatris.eu

Regulatory Support Service
WP18

Benzi Foundation
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Methodological Support
WP19

APHP
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Making Europe a world-leader in rare diseases research and innovation

ERDERA Data Services Hub (DSH)

Marco Roos presenter
Ana Rath & Ronald Cornet leading

JTC 2025 webinar 17th December 2024

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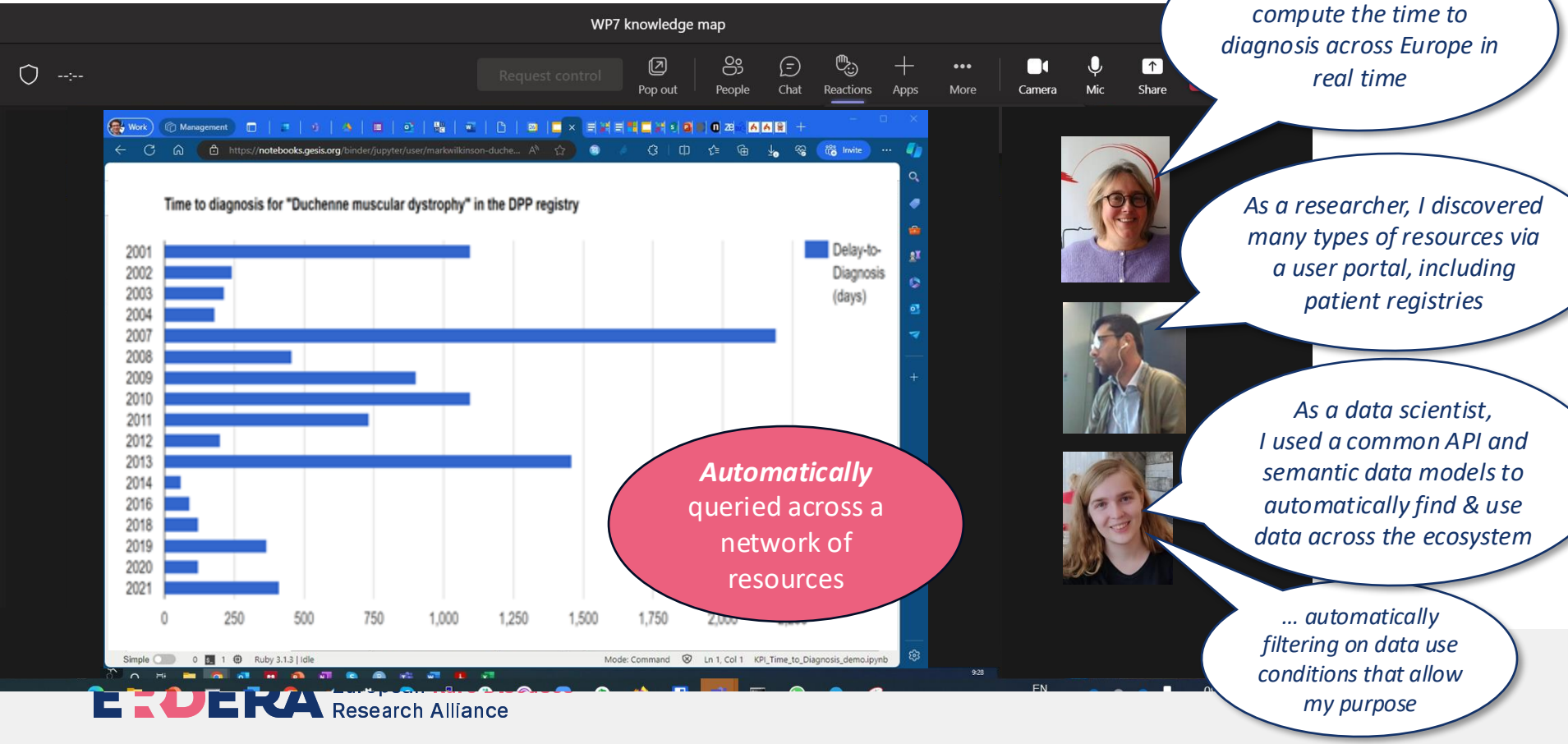
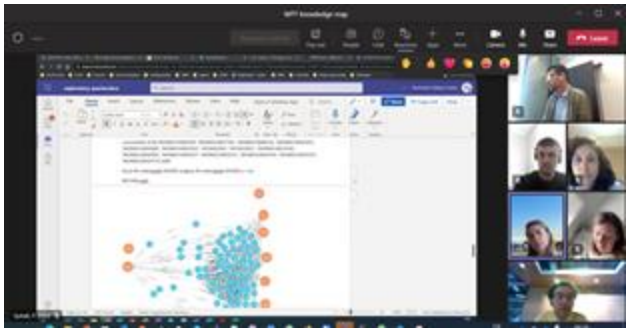
Presentation outline

- An example of the use of resources that contribute to a standards-based network of interconnected data resources
- Introduction to the ERDERA RD Data Services ecosystem
- Recommendations for pre-proposals

Reuse example of RD registries collecting ‘Common Data Elements’ FAIRly, including diagnosis & first hospital visit



Other examples of potential automated use of the RD ecosystem (*non-exhaustive list*)



- **Think of *your* scenario!**
- Real time hypothesis generation/evidence finding
- Continuous machine learning across the ecosystem
- Continuously update incidence statistics
- Suggest possible diagnoses, genes, pathways, treatments, repurposable drugs, with evidence
- Find clinical trials for patients & patients for trials
- Find and use authoritative mappings between codes
- Collaborative scenarios with industry

Opportunity to contribute to an RD ecosystem

It is vital that data collected for one goal are reused for other goals…

…using multiple sources of data as if all are in one database (but distributed in reality)

…by multiple stakeholders (patients, clinicians, data scientists, regulatory experts, policy makers)

… computationally Findable, Accessible, Interoperable, Reusable to boost research

Contribute as ERDERA partner to collaborative automated data exploration and analysis *by and on behalf of people living with a rare disease* (PLWRD)

ERDERA Data Services Hub RD ecosystem

Developed to facilitate data capture, integration, analysis and sharing across the RD community

FAIR (Findable, Accessible, Interoperable, Reusable) data sources

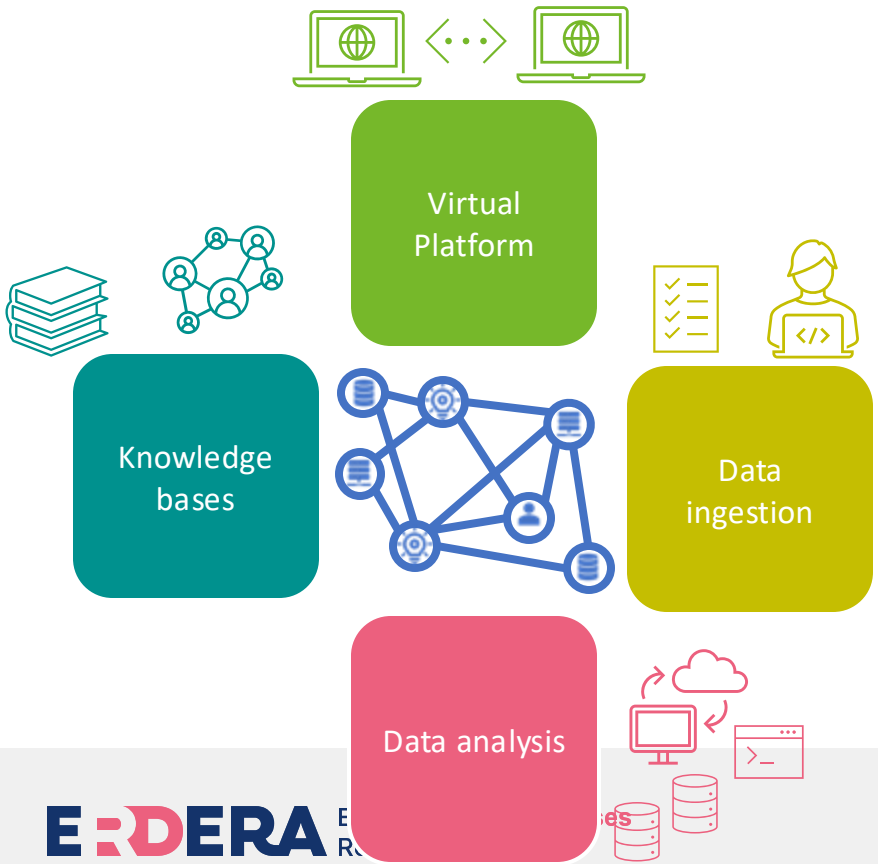
Data analysis pipelines

Knowledge bases

Federated data infrastructure - Virtual Platform

Towards enhancing the RD data and knowledge bases globally to benefit People Living With a Rare Disease (PLWRD)

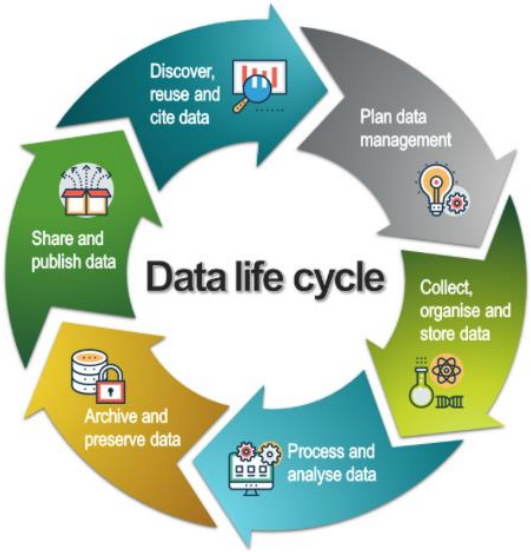
Data Services Hub RD ecosystem



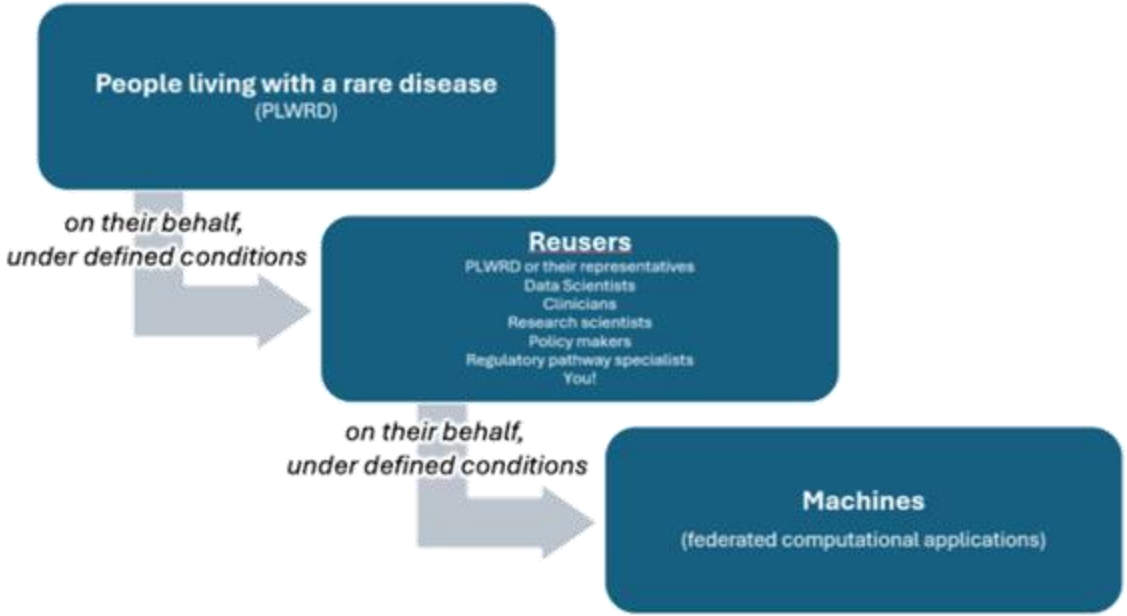
- **RD Virtual Platform (RD-VP)** federated ecosystem enabling to access and find RD data
- **Data readiness** adhering to standards and nomenclature ensuring data entering the RD-VP ecosystem are F+A+I+R for automated use
- **Data sharing and analysis** of genome-phenome data integrated into a federated infrastructure within the RD-VP
- **RD knowledge bases and ontologies** expanding and curating repositories, disease maps, semantic models and Patient-Centred Outcome Measures (PCOMs) across the RD ecosystem

Data Service Hub & RD-VP objectives

Facilitate data collection/generation by ERDERA partners that is sustainably standardised to enable automated finding, accessing, interoperating, and reusing for the benefit of PLWRD



Taken from www.slu.se/dms



Benefiting from the DSH RD ecosystem

- Minimal set of guidelines to facilitate FAIRification across the ERDERA RD ecosystem include:
  - Standardised approach to data generation/collection
  - Specified common nomenclature, ontologies, and ontology-based data models ‘for machines’
  - Tools to deliver accessible and comprehensive metadata (machine actionable descriptions of what you share)
  - Enabling computational and automated applications to scale
  - **Benefit to PLWRD and RD ecosystem sustainability**
- Enable multiple stakeholders to access a rich set of data services from the RD ecosystem to expand the utility of your service for PLWRD
- Access to a broad set of interconnected resources supporting your research in RD
  - Registries, genome-phenome databases, curated knowledge bases, molecular pathways, disease maps

Resources for implementation & training

Towards enhancing the RD data and knowledge bases globally to benefit PLWRD

Tools and guidelines for "Do It Yourself" contributions to the FAIR-based ecosystem
Recommended: always plan to engage with the ERDERA Expertise Hub to ensure a functional contribution

Online awareness training to advanced "Bring Your Own Data" workshops
Entry level webinars for FAIR project management; technical hackathon + training to learn, implement, exploit FAIR with experts

A platform for collaboratively adapting *YOUR TOOLS* and adopting *YOUR DOMAIN STANDARDs* to implement FAIR principles and evolve the ecosystem

Recommendations for proposals

JTC 2025: *"Pre-clinical therapy studies for rare diseases using small molecules and biologicals – development and validation"*

For data that your project generates or collects…

Standardising your data generation/collection approach to adhere to FAIR principles and to contribute to the RD ecosystem
Include the role of Data Steward

Engage with the ERDERA Data Services Hub to implement guidelines and specifications for data ingested into the RD ecosystem and to exploit the results

Access training and resources available in the ERDERA RD ecosystem to drive forward research questions to fruition and complement research outcomes

towards improving the lives of People Living With a Rare Disease

European Rare Diseases Research Alliance

Thank you!

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