ERDERA

European Rare Diseases
Research Alliance

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EURORDIS Open Academy SITR School
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Co-funded by
the European Union

ERDERA has received funding from the European Union's Horizon Europe research and innovation programme under grant agreement N°101156595.
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Towards a comprehensive RD Research Ecosystem

EUROPEAN JOINT PROGRAMME
RARE DISEASES
2019 - 2024


COLLABORATION

What is EJP RD

+2300 People

35 Participating countries

101 M€ Budget

130 Institutions

All 24 ERNs

EJP RD activities and achievements

Innovation Management Toolbox (511 resources)

Collaborative Research Funding

RD Virtual Platform

Training and empowerment

Accelerated Translation of Research Results and Clinical Studies

207 projects funded through the JTC

69 supported events through NSS

3 projects funded through RDR

More than 10 000 stakeholders trained

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

Patient trainings (425)

28 **inter-ERN workshop**

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

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

7 769 trainees from 150 countries)

RD Virtual Platform

(https://vp.ejprarediseases.org)

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

Over +**100 biological pathways** created

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

Expanding beyond EJP RD

Increased scope

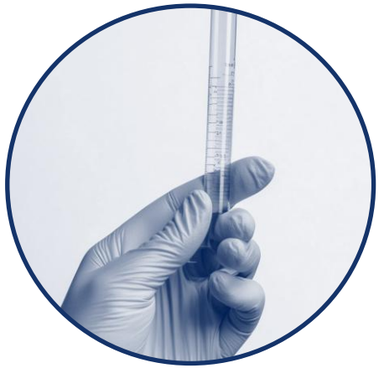
EUROPEAN JOINT PROGRAMME RARE DISEASES --> ERDERA

European Rare Diseases Research Alliance

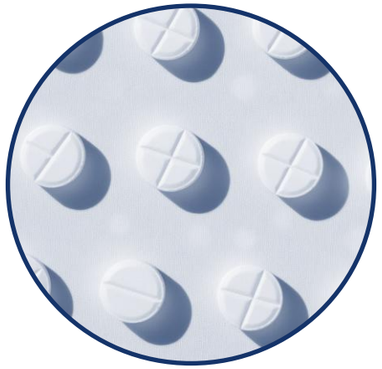
Pathway to impact

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

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

Our alliance includes

+170

Organisations

Funders

Research performing organisations

Patients' organisations

Research infrastructures

Private for-profit partners (industry & SME)

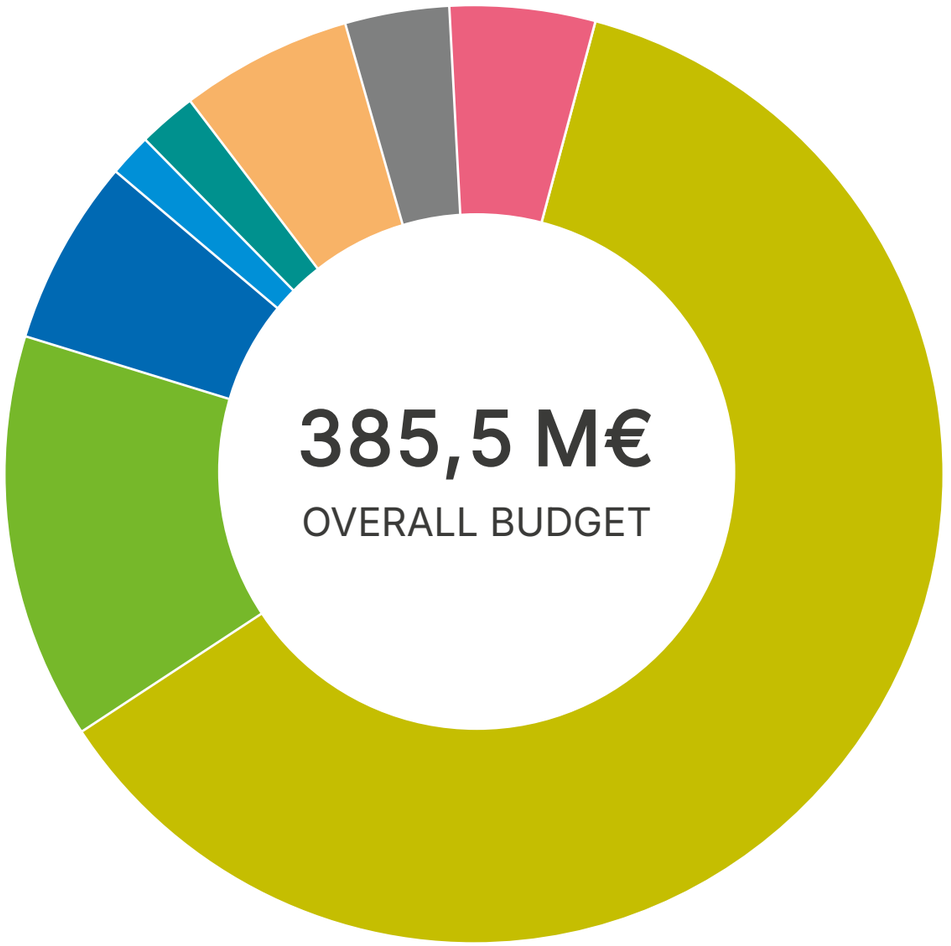
Other entities (universities, hospitals, non-profits, public administrations)

37

Countries

In Europe and beyond

Total Budget* per Work Stream



244,886.552 €

18,609.020 €

13,886,916 €

23,408.401 €

8,643,325 €

5,335.450 €

20,125.025 €

50,734.575 €

*estimated for 7 years, includes: associated partners, in-kind and in-cash contributions, and 150 M€ of EC contribution

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WP1
Coordination and management

Coordination & Strategy

RD Competitive Funding

- Joint Transnational Calls for collaborative research projects
- Clinical trial call management
- Networking to share knowledge on research

Data Services Hub — Expertise Services Hub — Education & Training Hub — Acceleration Hub

Rare Diseases Virtual Platform
Finding and accessing the data ecosystem
Data readiness services
Data sharing and analysis services
Knowledge bases and ontologies for research

Mentoring and consultancy
Regulatory support service
Methodological Support

Education and training
in rare diseases research

Technology accelerator
Public-Private Collaboration
Accelerator

Clinical Research Network

- Diagnostic data availability
- Genome re-analysis research pipeline
- Innovation to shorten time to RD diagnosis
- Real world data
- Clinical Outcome Assessment
- Advanced Therapeutic Medicinal Products
- N-of-few approach

Inter(national) Capacity Alignment

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

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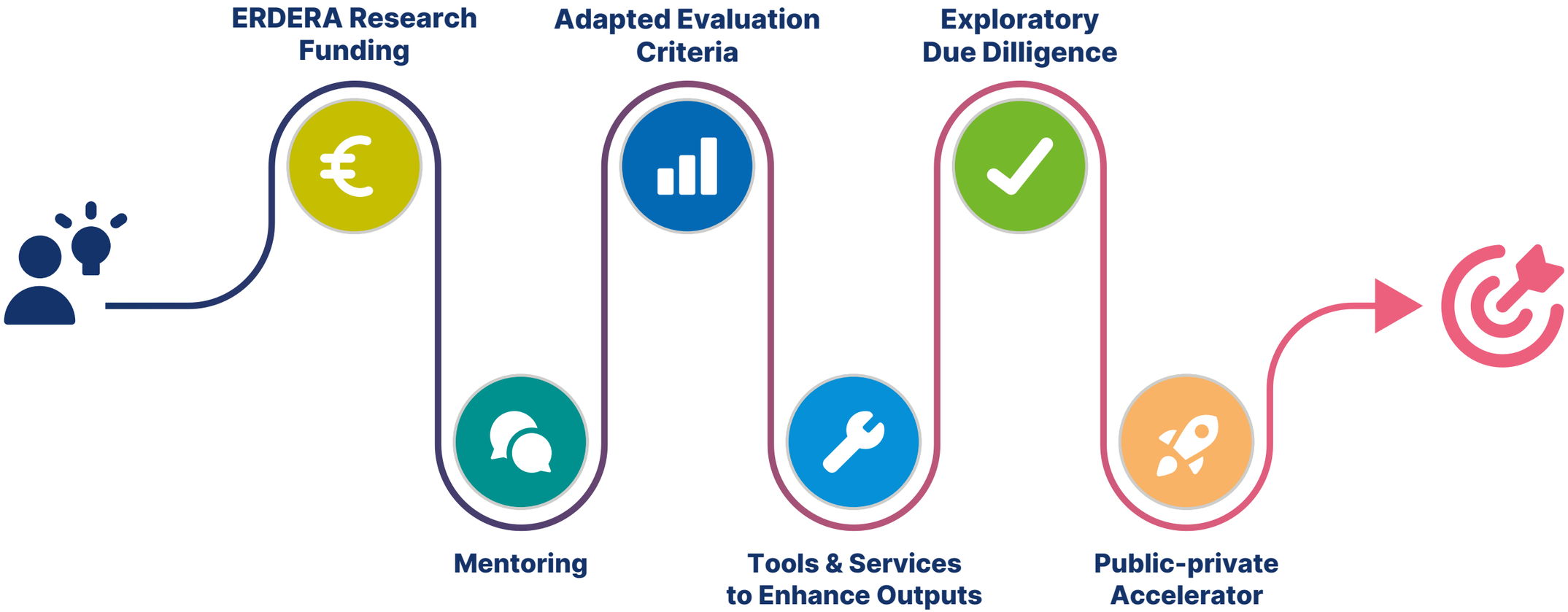
ERDERA innovation support cycle to boost clinical research & expedite innovation



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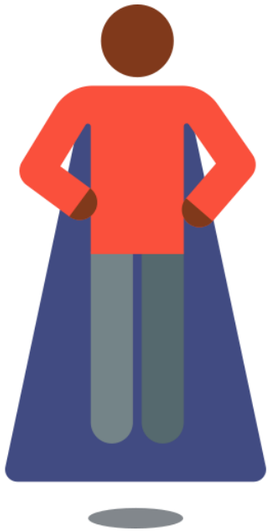

Your voice/opinion/contribution via:

- Governing, executive and advisory bodies
- Patient & Public Engagement and Involvement (PPIE)
- Direct involvement as patient expert
- Open consultation/survey

Your benefit via:

- Participation/coordination of a Networking project/event
- Participation/partnership in multinational research project
- Participation in offered education & training
- Participation/coordination of a National Mirror Group
- Accelerated access to expertise/advice

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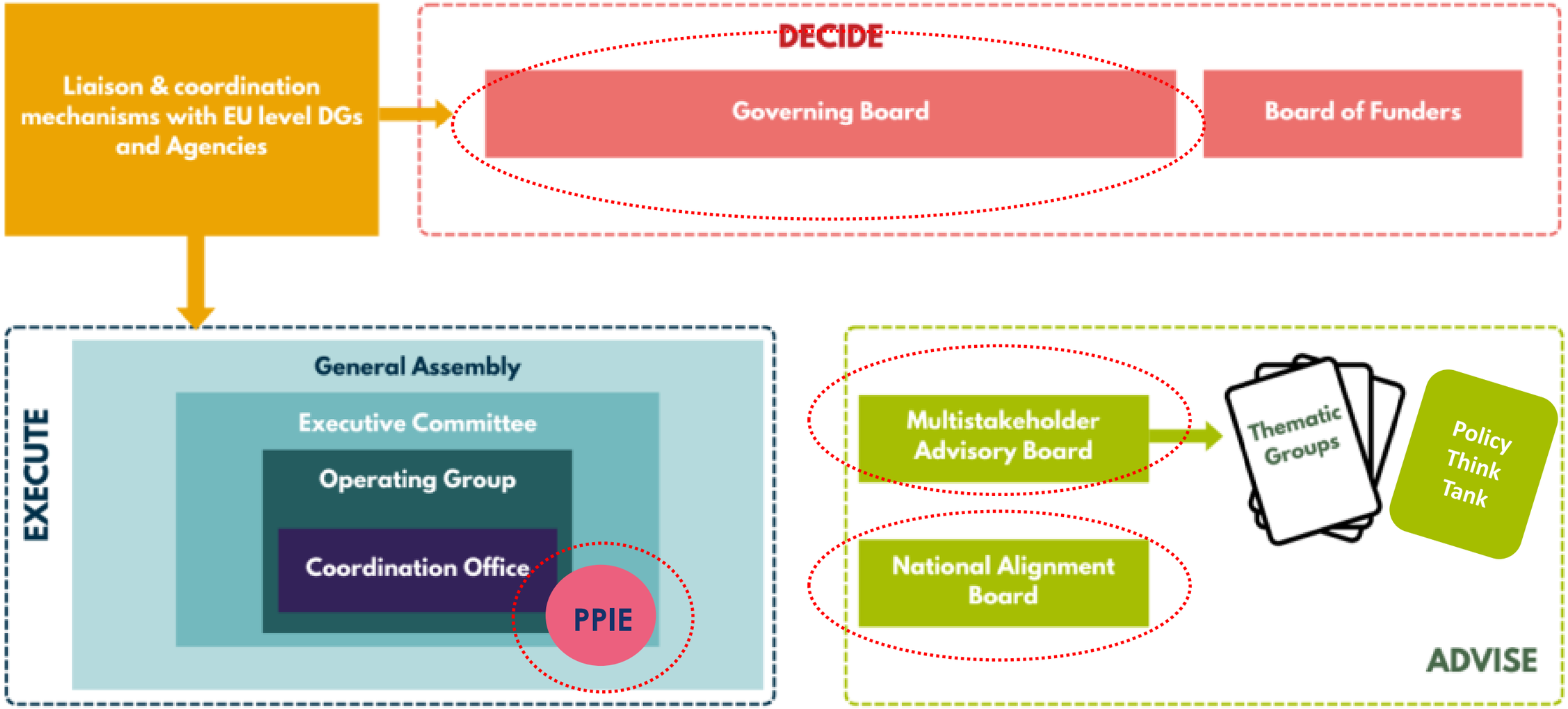

Policy Think Tank

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What

- Embedded in WP Coordination
- Encompass all representatives of PAOs in ERDERA
- Supported by operational team of PAOs representatives

Why

- Ensure that PPIE is achieved throughout ERDERA
- Act as the advocacy arm of ERDERA
- Develop framework for PPIE in all parts of the partnership

How

- Hub to answer inquiries from patients
- Collecting needs & expectations
- Indentifying patient to part in tasks
- Links with PAOs groups and networks
- Developing resources
- Supporting coo

NMGs should:

- Seek to unite the RD research community in their national territory (and either integrate the 'care' community too, or else forge robust links to suitable groups/structures)
- Bridge the gap between national/regional RD activities and expertise, on the one hand, and the international level on the other (embodied by, as a minimum ERDERA, but considering also IRDiRC and any other relevant pan-RD structures or initiatives)
- Ensure that all key stakeholders in their national territory are represented in the NMG – including the following, as far as possible, plus representatives of any other relevant national structures:
  - researchers, clinicians, people with lived experience (ideally national alliance for RD organisations, where existing), ERN representatives, national competent authorities, key funders, regulatory experts, industry
- Contribute, directly or indirectly, to the support of an active, specific and meaningful national RD plan/strategy in the country, with multistakeholder engagement

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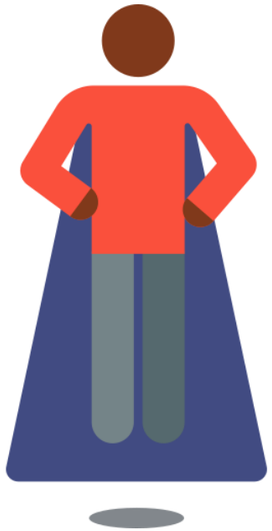
The involvement of national alliances within each NMG is very important



Each NMG must have patient organisations representatives involved

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What are the opportunities of collaboration that ERDERA can offer?

Yearly Joint Transnational Calls

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

Networking Support Scheme

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

Multinational Clinical Trials Call

A call for multinational clinical trials is expected in 2026.

Direct Collaboration Opportunities

Collaborate directly with ERDERA's research teams involved in "in-house" research.

IMPROVE INVOLVEMENT OF PATIENTS IN RESEARCH

- At all stages: topics definition, evaluation, patient-driven research projects

- In the Joint Transnational Call 2019, 36% and In 2021 we reached 100% of funded projects involved PAOs

- ERDERA finances PAO participation in projects

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Financial support to applicants for fostering organization of workshops or conferences for new research networks or existing/expanding research networks to strengthen collaborations and to enable exchange of knowledge.

Share knowledge on research among relevant transnational European and transcontinental RD stakeholders including clinicians, basic researchers, patients' organisations/RD support groups, as well as PhD students, post-docs and early career researchers, and industry

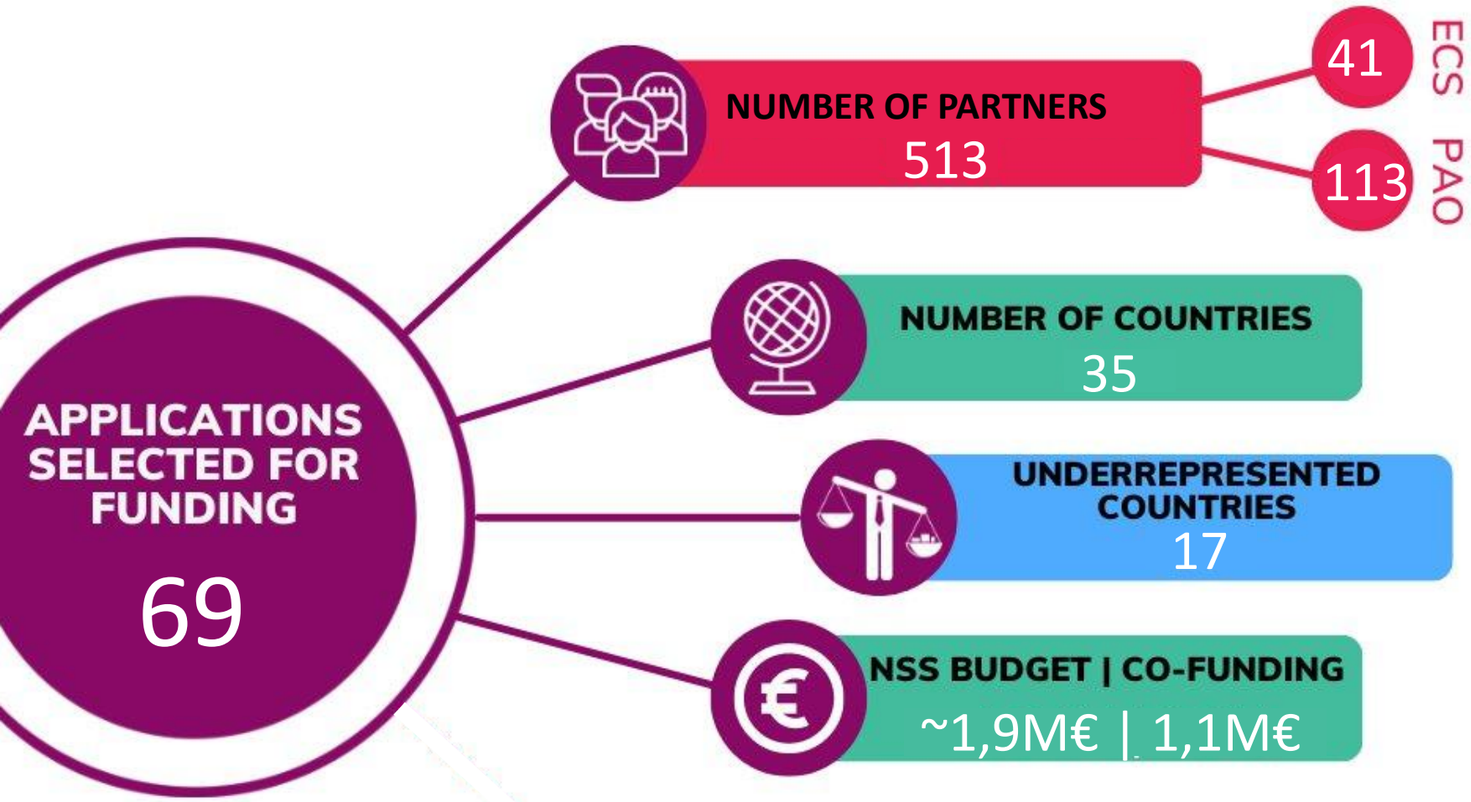
Build new European and global research networks or expanding existing research networks on RDs and rare cancers to support patient-centred research, and to include stakeholders of widening countries and young researchers

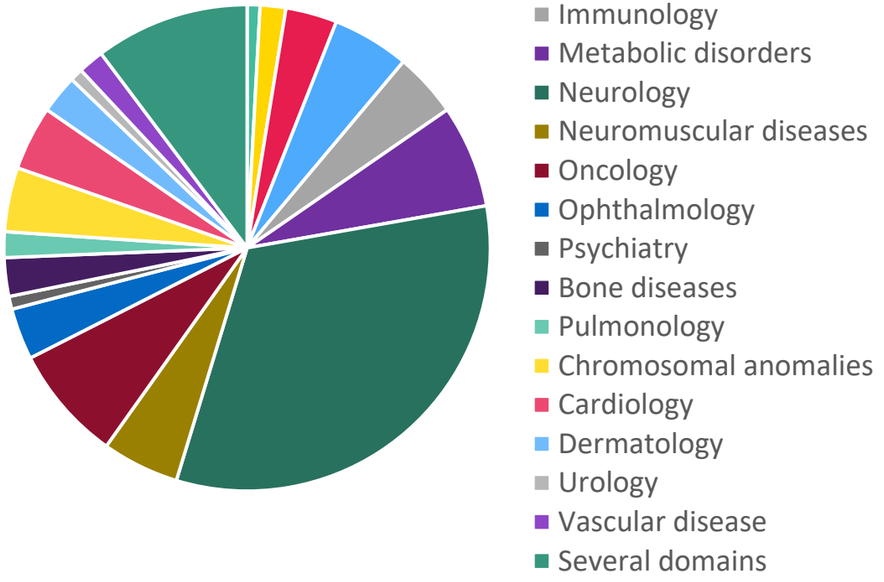
30 000 €/event

Continuously open (eligibility check every 6 months)

Collaborative (min. 3 applicants from 3 ERDERA countries)

NSS in EJP RD





Medical domain distribution of received NSS applications

*ECS: Early Career Scientists
*PAO: Patient Advocacy Organisations

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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

Typical success rate: 1st stage vs final funding = 10-12%; 2nd stage vs final funding 35 -50%. Typical overall project budget 1.0 – 1.5 Million €

https://erdera.org/funding/#joint-translational-call

Launched every year in December with pre-announcement the latest in November

2-stage evaluation process. 3-years projects

A minimum of 3/4 eligible partners and a max. of 6 per project
(can be extended to 8 according to specific conditions)
PAOs participation is financed

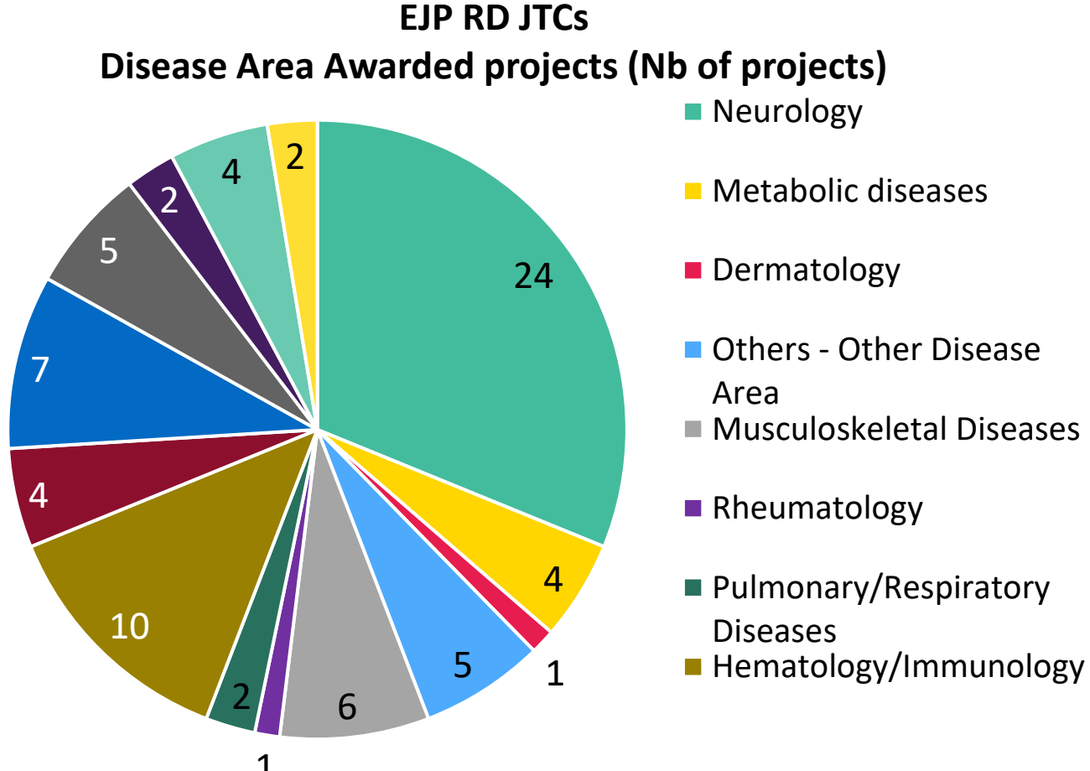
Joint Transnational Calls in EJP RD

- **5 calls launched:**
  - **JTC 2019 –** Research projects to accelerate diagnosis and/or explore disease progression and mechanisms of rare diseases
  - **JTC2020 –** Pre-clinical research to develop effective therapies for rare diseases
  - **JTC2021 –** Social Sciences and Humanities Research to Improve Health Care Implementation and Everyday Life of People Living with a Rare Disease
  - **JTC2022 –** Development of new analytic tools and pathways to accelerate diagnosis and facilitate diagnostic monitoring of rare diseases
  - **JTC2023 –** Natural History Studies addressing unmet needs in Rare Diseases

77 funded projects

More than **104million euros**

➔For more information on past calls (call documents, etc.):
https://www.ejprarediseases.org/past-funding-opportunities/



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MAIN GOAL: Current treatments are usually supportive rather than disease-modifying, leaving most patients with rare diseases with considerable unmet medical needs. Tackling these issues will require **valid and reproducible clinical trials, and opportunities for those affected by rare diseases to influence and participate in these trials.**

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.

Two committees: Clinical Trials Scientific Committee (CTSC) responsible for triage of pre-proposals based on scientific merit and overall relevance; and Clinical Trails Evaluation Board (CTEB) responsible for the evaluation of full proposals.

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

Participation of patients to be defined

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Free & open to all

Foundation for Rare Diseases & European Joint Programme on Rare Diseases

Diagnosing Rare Diseases: from the Clinic to Research and back

4.9 (54 reviews)

5 weeks · 3 hrs per week

Included in Unlimited

Find out more

Foundation for Rare Diseases & European Joint Programme on Rare Diseases

Innovative Therapies and Personalised Medicine for Rare Diseases

4.6 (5 reviews)

5 weeks · 4 hrs per week

Included in Unlimited

Find out more

Foundation for Rare Diseases & European Joint Programme on Rare Diseases

Health Data Ethics & Regulatory Frameworks in Rare Disease Research

4 weeks · 3 hrs per week

Included in Unlimited

Find out more

Foundation for Rare Diseases & European Joint Programme on Rare Diseases

From Lab to Clinic: Translational Research for Rare Diseases

4.9 (15 reviews)

5 weeks · 4 hrs per week

Included in Unlimited

Find out more

https://www.futurelearn.com/partners/french-foundation-rare-diseases

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Patients' and young researchers' training: Blended and scalable training programme for patients & young researchers across the whole RD research pipeline

Training workshops for paediatric patients

RD research training for multistakeholder community

- **Trainings to empower RD diagnostic research**
- **Trainings to empower RD clinical research**
- **Trainings to empower knowledge on research methodologies**
- **Trainings to empower acceleration of research and RD clinical trial methodologies and management**
- **Trainings to empower RD data research**

Agenda - ERDERA Young Patients Training

Day 1 - Friday 2nd May, 2025

08:00 – 09:30 | Opening Session
- Welcome & introduction to the ERDERA training (15 mins)
- Welcome & introduction from the KIDS groups (30 mins)
- The role of advocacy and patient organizations (30 mins)
- Q&A and discussion (15 mins)

09:30 – 10:00 | Break

10:00 – 11:30 | Rare and Genetic Diseases Specificities
- Overview of rare and genetic diseases in pediatric patients (30 mins)
- Challenges in diagnosis, research and care (30 mins)
- Case studies and interactive discussion (30 mins)

11:30 – 12:00 | Break

12:00 – 13:30 | Patients' and Children's Rights
- Legal and ethical considerations in pediatric research (30 mins)
- The role of young patients in matters related to their health (30 mins)
- Group discussion and real-world examples (30 mins)

13:30 – 14:30 | Lunch Break

14:30 – 16:00 | Translational Research Challenges
- Innovative therapies; ethical and legal issues in pediatric research (45 mins)
- Unmet needs and prioritization in health and research decision-making (45 mins)
- Interactive session with case-based scenarios (30 mins)

16:00 – 16:30 | Break

16:30 – 18:00 | Activities

ERDERA European Rare Diseases Research Alliance

TEDDY NETWORK European Network of Excellence for Paediatric Research

In collaboration with: RARE DISEASES GREECE

With the support of:

Day 2 - Saturday 3rd May, 2025

08:00 – 09:30 | Patient Engagement in Clinical Trials
- Involvement in CTs and innovative studies (PROMs, PREMs, etc.) (30 mins)
- Consent/assent, clinical study protocol, and lay summaries (30 mins)
- Q&A and participant reflections (30 mins)

09:30 – 10:00 | Break

10:00 – 11:30 | Orphan Medicinal Products & Pharmacovigilance
- Availability and access to orphan drugs in the EU (30 mins)
- Pharmacovigilance and off-label use in pediatric settings (30 mins)
- Group discussion and case analysis (30 mins)

11:30 – 12:00 | Break

12:00 – 13:30 | Closing Session
- Key takeaways from the training (40 mins)
- Participant feedback and evaluation (40 mins)
- Closing remarks (10 mins)

13:30 – 14:30 | Lunch Break

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In collaboration with: RARE DISEASES GREECE

With the support of:

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Virtual Platform (VP)
FAIR by design

Find patients with particular condition in multiple sites

Machine-Readable Consent and Access conditions

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

- ✓ Federated
- ✓ Standardized
- ✓ GDPR-compliant
- ✓ Sustainable
- ✓ Quality assessed

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

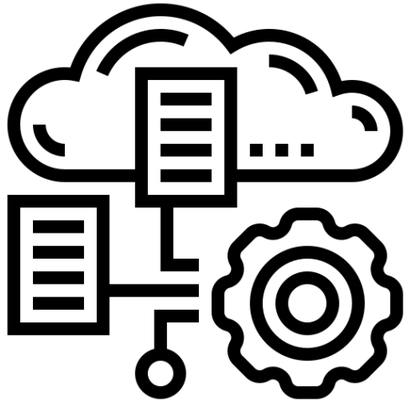
45 resources connected (registries, catalogues, gen-phen deposition infras, knowledge bases, projects)

Over +**100 biological pathways** created

3 linkage/discoverability levels

https://vp.ejprarediseases.org

RD VIRTUAL PLATFORM



Data (re)analysis

FAIR stewardship

Computing infrastructure

RD DATA HUB

https://vp.ejprarediseases.org

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ERDERA in house research

DIAGNOSIS

- Diagnostic data availability
- Genome re-analysis research pipeline
- Innovation to shorten time to RD diagnosis

CLINICAL TRIAL READINESS

- Real world data
- Clinical Outcome Assessment

THERAPY DEVELOPMENT

- Advanced Therapeutic Medicinal Products
- N-of-few approaches

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