EUROPEAN JOINT PROGRAMME ON RARE DISEASES (EJP RD)

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Objectives of the EJP RD

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

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

750 people
- 650 Scientifics
- 100 Admin

35 participating countries
- 26 EU MS, 7 associated (AM, CH, GE, IL, NO, RS, TK), UK and CA

101 M€ Budget
- Union contribution: 55 M€ (70% reimbursement rate)

87 beneficiaries
- 9 hospitals
- 12 research institutes
- 31 research funding bodies/ministries
- 24 universities/hospital universities
- 5 EU infrastructures
- 5 charities/foundations
- EURORDIS

+ 50 linked third parties
+ 100% associated networks

85% of European RD community (directly or indirectly) involved in EJP RD

Coordinated by Inserm
EJP RD STRUCTURE

COORDINATION & TRANSVERSAL ACTIVITIES

INTEGRATIVE RESEARCH STRATEGY

SUSTAINABILITY

ETHICAL & REGULATORY

COMMUNICATION

1. FUNDING

2. COORDINATED ACCESS TO DATA & SERVICES

3. CAPACITY BUILDING & EMPOWERMENT

4. ACCELERATING TRANSLATION OF RESEARCH & THERAPY DEVELOPMENT

Coordinated by Inserm

Funded by the European Union GA 825575
EJP RD

what is there for RD stakeholders?

FUNDING, TRAINING & SUPPORT OFFICES
JOIN TRANSNATIONAL CALLS

MAIN GOAL: enable scientists and researchers 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

MORE INFORMATION: https://www.ejprarediseases.org/index.php/fundings-and-calls/
Are you looking for:

Gathering of experts & patients to share knowledge?  
Expanding your network to include new stakeholders?  
Support future consortium to apply to EC calls?

Networking support scheme (continuous call)  
Objective: encourage sharing of knowledge on rare diseases to support health care professionals, researchers and patient advocacy organizations with a networking grant to re-organize themselves into transnational (clinical) research networks.
Are you looking to:

Train your PhD student/young MD within your ERN network or within other ERN network?

Research Mobility Fellowship (2 calls/year):
Aim: financially support PhD students, medical doctors & post-docs working in ERN-member institutions to undertake short scientific visits (secondments) up to 6 months fostering specialist research training outside their countries of residence and within one of the ERN host institutions.
Are you looking to:

*Share transversal type of knowledge (going beyond your ERN) and have interesting training idea?*

**Research training network (2 calls per year)**

Aim: identify the most suitable proposals for the organization of research training workshops of 2 days targeted to the ERNs needs. Selected research training workshops will have to train ERN researchers and clinicians in ERN relevant innovative training themes. Moreover, the workshops will be aiming to provide a cross-ERN added value.
Are you looking to:

*Train your members or gain knowledge in rare diseases research related aspect?*
Training & empowerment

- **7 face-to-face courses** in 5 countries (220 participants, 18 fellowships)
- **9 online courses** (270 participants)
- **500 stakeholders trained** so far increasing research potential of the multi-stakeholder EU RD research community

E-learning academic course on RD

- **Module 1**: RD Diagnosis
- **Module 2**: RD Innovative personalized therapies
- **Module 3**: RD translational research
- **Module 4 & 5**: to be defined in year 3

### Course Modules

- **WP 14**: Training on data management & quality
- **WP 15**: Capacity building and training of patients and researchers
- **WP 16**: Online Academic education course
- **WP 17**: ERN RD training and support programme
- **WP 18**: Development and adaptation of training activities

### Course Topics

- Quality assurance, variant interpretation and data management in the NGS diagnostic era
- Strategies to foster solutions of undiagnosed rare disease cases
- Organising & maximising RD samples biobanks
- International Summer School on Rare Disease Registries and FAIRification of Data
- Orphanet Ontology Training
- EURORDIS EXPRESS Expert Patients and Researchers Summer School
- EURORDIS Winter School
- EURORDIS Leadership School
You have project and/or preliminary results for e.g. new therapy/biomarker/device & You are looking for:
- Evaluation of translational potential of your project
- Further regulatory support
- Advice on how to get interest of potential sponsor or derisk your project

You have idea about possible multinational clinical study but for which e.g. there is no interest from industry? & You are looking for (e.g.):
- Advice on how to advance with such project
- Advice on how to put in place clinical study with public sponsors
- Advice on overall management & regulatory issues of multinational clinical studies
Support for Accelerated translation of research results

- Accompany research projects
  - Mentoring from their conception and throughout their lifetime & facilitate next steps
- Provide tools (based on use-cases) free of charge → innovation management toolbox
- Open to EJP RD beneficiary institutions, E-Rare & EJP RD funded projects, ERN projects
- Innovation Management Toolbox (to be open in coming weeks)

helpdesk@ejprarediseases.org
Methodological & regulatory

- Launch of **3 demonstration projects** validating novel methodologies in small population CTs (proof required by regulators) with

- Launch of **innovative methodologies** for clinical studies in limited populations (below 1 in 50 000)

- Direct collaboration with regulators: EMA and EU Innovation Network (national competent authorities)

Ethics/Informed Consent

**ERN Registry Informed Consent Form (ICF)**

- High level expertise: EJP RD Advisory regulatory & Ethics Board

- Close collaboration with all ERNs + proof-of-concept developed based on initial EC ICF & real feedback from national/institutional ethics committees

- Improved ERN registry informed consent including “customised choices” to meet National and Site specific requirements as well as GDPR compliance

- Keep easy to read (time & language) by patients & carers (including minors)
Virtual Platform (VP)

Federated
Standardized
GDPR-compliant
Sustainable
Quality assessed

Findable
Accessible
Interoperable
Reusable
EJP RD Virtual Platform

**Counting Patients with specific conditions**

**Explore & use (RD) Catalogues to answer questions**

**Make Consent machine readable for Automatic data Access**

**Use of multi-omics data for diagnosis & identification of drug targets**

**HEALTHCARE + European Reference Networks (ERN)**
Support & connection of ERN registries (patient health data)

**INFRASTRUCTURES & CATALOGUES**
Induced improvement for exploitation by RD community

**NATIONAL ALIGNMENT**
Referenced in PNMR3, FR funding opportunities for standard alignment with Health Data Hub, cohorts, FR RD DBs

**EU ALIGNMENT**
Referenced in EC/IMI calls for projects

**INTERNATIONAL**
Collaboration with international stakeholders (CPATH) on joint standards & methodologies
EJP RD – single entry point & solutions for all in rare diseases

**RESEARCHERS**
- Funding
- Research support services
- Training at every stage
- Access to resources & tools
- Access to extensive network & expertise

**CLINICIANS**
- Clinical studies support services
- Support for registries
- Access to resources & tools to accelerate diagnosis
- Access to extensive network & expertise

**PATIENTS**
- Access to RD specific expertise
- Networking
- Training at every stage
- Access to resources & tools
- Access to extensive network & expertise

**POLICY MAKERS & FUNDERS**
- Joint funding & strategy
  - Optimisation of investment in research
  - Access to support for national RD community
  - Access to extensive network & expertise
  - Holistic impact evaluation

**INTERNATIONAL PARTNERS**
- Access to extensive RD network & expertise
- Multiple collaboration opportunities
- Possibility of alignment
- Access to resources & tools
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