Rare Diseases Landscape in Europe

Funded by the European Union GA n° 825575

Rare Diseases Research

Strategy

Infrastructures

Funding

Patients Needs

Healthcare +

European Reference Networks

EJP RD

Eurordis

 Rare Diseases Europe
Objectives

**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

**Specific objective:**
Improve integration, efficacy, production and social impact of research on rare diseases through the development, demonstration and promotion of sharing of research and clinical data, materials, processes, knowledge and know-how, and an efficient model of financial support for research on rare diseases
Main facts about the EJP RD

Jan 2019 Dec 2023

Total budget (min. submitted): 101 M€ (→ expected > 110 M€)

EU contribution: 55 M€

35 participating countries

88 beneficiaries
- 31 research funding bodies/ministries
- 12 research institutes
- 22 universities/hospital universities
- 11 hospitals
- 5 EU infrastructures (BBMRI, EATRIS, ECRIN, ELIXIR, INFRAFRONTIER) + EORTC
- EURORDIS & ePAGs
- 5 charities/foundations (FTELE, AFM, FFRD, FGB, BSF)
+ 50 Linked Third Parties

27 EU MS
7 associated
1 third country

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WP1 COORDINATION & MANAGEMENT

WP2 STRATEGY
WP3 SUSTAINABILITY
WP4 ETHICS, LEGAL, REGULATORY & IPR
WP5 COMMUNICATION & DISSEMINATION

P1
- WP6 Joint Transnational Calls
- WP7 Networking scheme
- WP8 RDR Challenges
- WP9 Monitoring of funded projects

P2
- WP10 User-driven strategic planning for P2
- WP11 Virtual Platform for data & resources
- WP12 Enabling sustainable FAIRness
- WP13 Holistic approaches for rare disease diagnostics and therapeutics

P3
- WP14 Training on data management & quality
- WP15 Capacity building and training of patients and researchers
- WP16 Online Academic education course
- WP17 ERN RD training and support programme
- WP18 Development and adaptation of training activities

P4
- WP19 Facilitating partnerships and accelerating translation
- WP20 Validation, use and development of innovative methodologies for clinical studies
Pillar 1: Collaborative research funding
Pillar 1

- Joint Transnational Calls for collaborative research projects
- Networking to share knowledge on rare diseases
- Rare disease research challenges
- Monitoring of funded projects

- Achievement of critical mass of knowledge & resources
- Accelerated diagnosis and treatment development
- New and expanded networks – inclusion of stakeholders, knowledge sharing
- New diseases targeted
- Public-Private Partnerships
- Proof of Concept and optimisation studies
### Pillar 1

**WP6: Joint Transnational Calls for collaborative research projects**

- 2 co-funded calls + 2 non-co-funded – 27 M€ budget for call 1 – open to research teams from countries with funders involved – min of 4 teams from 4 countries

**WP7: Networking to share knowledge on rare diseases**

- Small support schemes for networking (workshops/events/knowledge sharing) – 30 K€ max – open all year long – open to all countries involved in EJP RD

**WP8: Rare disease research challenges**

- Public-private (small scale) partnerships – challenges set by industry and validated by EJP RD – short term (max. 30 months) projects

**WP9: Monitoring of funded projects**

- Monitoring of all projects funded through EJP RD and previous E-Rare projects
WP6: Joint Transnational Calls

General information:

- 1 call / year
- Topics are decided based on the input from scientific community (through the prioritisation process) and other RD stakeholders
- Final decision on the call (final topic description, national/regional eligibility criteria) belongs to the participating funding (national/regional) agencies
- Funding agencies commit and earmark budget every year but there is no obligation to participate every year (may be dependent on the national budgetary-political configuration)
- The contribution of the EC serves to increase the overall capacity of funding (number of funded projects) and/or can be dedicated to a specific goal (e.g. funding of Patient Advocacy Organisations in JTC 2019). The decision on the use of EC contribution belongs to the funding agencies
- **Call schedule (similar every year):**
  - Pre-announcement: November
  - Opening: December
  - Pre-proposal submission: February
  - Decision selected pre-proposals: April/May
  - Submission of full proposals: June
  - Final decision: Sep/Oct
WP6: Joint Transnational Calls

1st Joint Transnational Call (2019)
- Total budget 27 M€ from participating Member States + min 5 M€ of EC contribution
- 31 funders from 23 countries
- Topic: Research projects to accelerate diagnosis and/or explore disease progression and mechanisms of rare diseases

Eligible partners (check the national rules!):
- Academia (research teams working in universities, other higher education institutions/research institutes)
- Clinical/public health sector (research teams working in hospitals/public health and/or other health care settings and health organisations)
- Enterprises. Participation of small and medium-size enterprises (SMEs) is encouraged when allowed by national/regional regulations
- Patient advocacy organisations

Other eligibility rules:
- Min. 4 eligible partners from 4 different countries
- Max. 6 partners per project (can be extended to 8 if underrepresented countries (CZ, EE, HU, LT, PL, SK, TK) participate)
Associated partners from countries NOT participating in the call may be part of the project if they participate with their own funding, their number is limited to 2
WP7: Networking to share knowledge on rare diseases

- **Objective:** Small support schemes for networking (workshops/events/share of knowledge/creation of new or enlargement of existing networks)
- **Max. funding amount:** 30 K€
- **Official opening of the first funding opportunity:** second semester 2019
- Open all year but 4 evaluation sessions per year (except 2019 – 2 sessions)
- Open to **all countries involved in the EJP RD**
- Funding should be spent within 1 year from the allocation (TBC)
- Reporting on the activity will be required after the completion of the networking activity
- Eligibility rules following Horizon 2020 criteria
- Activity under responsibility (WP leader) of ZonMw (NL)
WP8: Rare disease research challenges

- **Objective:** Public-private (small scale) partnerships
- The NC3R Crack it Challenge scheme as starting point – challenges set by industry (and validated by EJP RD)
- **2 workshops to define the challenges and finalise the call**
- Applications expected from academia and/or SMEs
- Short term (max. 30 months – 18m + 12m) projects with co-funding/in kind contribution from industry
- **Max. budget per project: 250 K€** (150 K€ in a first phase with possible extension of 100 K€ in the second phase)
- Eligibility & evaluation rules following Horizon 2020 criteria
- Activity under joint responsibility (WP leaders) of FFRD (FR) and EURORDIS
Pillar 2: Innovative coordinated access to data and services for transformative rare diseases research
Pillar 2 target: FAIR-based virtual platform

A powerful substrate for translational research:

- Centralized services for collections (resource-level)
  - Sample, biobanks, registries, infrastructures and tools catalogue
  - Analysis platform for omics data
  - Curated rare disease-centered information and data

- Federated services for data elements (record-level)
  - FAIR ‘at source’
  - Data, patients, and samples linked and discoverable
  - Consents and data use conditions also represented

The fuel

ERN research
RD researchers

ERN experts learning from computational experts

Developers learning from ERN experts

PILLAR 2

Enhanced RD research

The mechanics

Computational research
Data science
Pillar 2: Impact and outputs

- Decrease fragmentation of resources relevant for RD research: data, infrastructures and tools will be accessible in an integrated way
- Recommended standards, tools and services for FAIRification at record-level
- An ecosystem of federated rare disease data, patient and sample stations that enable discovery and analysis across countries and institutes
- X-omics: rare diseases pathways based on multi-omics, environmental and toxicology data mining

- Virtual Platform and services built according to end-users needs
- **Resources findable and exploitable** through the EJP RD virtual platform
- **User-friendly analysis interface of RD research projects and trials for funders**
- **Simplified access to resources** by adopting a common Authentication and Authorization Infrastructure (AAI)
- A system to **link anonymised data** from the same patient in different projects via Privacy Preserving Record Linkage (PPRL)
Pillar 3: Capacity building and empowerment
Pillar 3: Activities

WP14: Training on data management & quality
Orphanet nomenclature – standards & quality of genetics/genomics data in clinical practice – strategies to foster undiagnosed diseases – biobanks sample data management – rare diseases registries & FAIRification at source – European Rare Diseases Registry Infrastructure

WP15: Capacity building/training of patients and researchers in RD research and processes
Expert Patients and Researchers EURORDIS Summer school – scientific innovation and translation research aspects in RDs for patient advocates – leadership & communication skills for patient advocates and representatives – education material and activities for paediatric patients

WP16: Online academic education course
Based on assessed needs of the RD community – in collaboration with universities – 10 to 12 modules with accreditation – e-learning format open to all

WP17: ERN RD training & support programmes
Based on 4 groups (Neuro, Neoplasm & malformation, Organs, Systemic) – preferences, needs and resources of ERNs – tailored for and performed by ERNs
Pillar 3: Impact and outputs

- Comprehensive, coherent & accessible EU RD research training programme: Rotating F2F courses, fellowships, online academic course
- Leverage on existing training expertise & resources and create new experts
- Open access RD education (e-learning)

- Increased level of knowledge and know-how within the RD research and care community, including through ERNs and RD patient representatives
- Cultural and scientific changes to systematically develop patient-centred RD research integrating the patients' voice
- ERNs training program for further medical education/integration of ERN trainings in academic schemes (increased number of trained RD medical doctors/researchers)
Pillar 4: Accelerating the translation of high potential projects and improving outcomes of clinical studies in small populations
Pillar 4: Activities

**WP19: Facilitating partnerships and accelerating translation for higher patient impact**

Innovation management toolbox – assessment and real time mentoring of translational projects – support in exploitation and follow-on funding – partnering support – roadmap for European investment platform for RD

**WP20: Accelerating the validation, use and development of innovative methodologies tailored for clinical trials in RDs**

Key Task Force group - Support in design and planning of RD clinical studies with ECRIN – demonstration projects on existing statistical methodologies to improve RD clinical trials – innovative methodologies to improve RD clinical trials in limited populations
Pillar 4: Impact and outputs

- More effective translation of knowledge into clinical benefit
- Easy access for researchers and funders to expertise and resources in the innovation process
- Faster transition to clinic for high potential projects
- Improved communication between actors involved in RD clinical studies
- Improved clinical study methodologies for small populations

- Openly accessible resources for the R&D community to perform effective translation – from bench to bedside
- Active, in-project support for translation, follow-on funding and exploitation
- Roadmap & design for EU investment platform for early innovation funding in RD
- Innovative tailored clinical study methodologies necessary for RD research
THANK YOU

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