EUROPEAN JOINT PROGRAMME ON RARE DISEASES (EJP RD)

Rima Nabbout, MD, PhD
Necker Enfants Malades Hospital, APHP,
Institut Imagine, Université de Paris, Paris, France

EUROPEAN JOINT PROGRAMME
RARE DISEASES

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

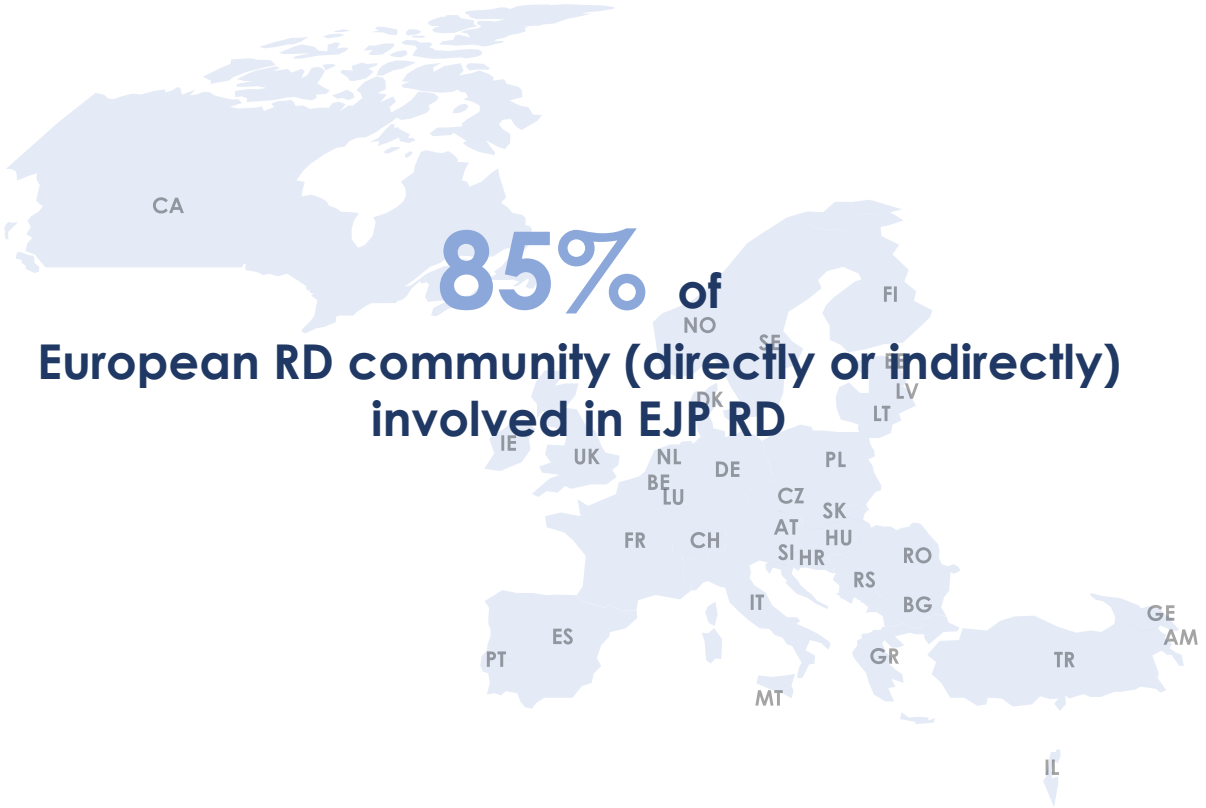
Coordinated by Inserm

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)



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

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

EURORDIS RARE DISEASES EUROPE
eatris
elixir
INFRAFRONTIER mouse disease models
European Reference Networks
ECRIN EUROPEAN CLINICAL RESEARCH INFRASTRUCTURE NETWORK
BBMRI-ERIC gateway for health

EJP RD STRUCTURE

Coordinated by Inserm
La science pour la santé
From science to health

COORDINATION
& TRANSVERSAL ACTIVITIES

INTEGRATIVE RESEARCH STRATEGY

SUSTAINABILITY

ETHICAL & REGULATORY

COMMUNICATION

1 FUNDING

2 COORDINATED ACCESS TO DATA & SERVICES

3 CAPACITY BUIDLING & EMPOWERMENT

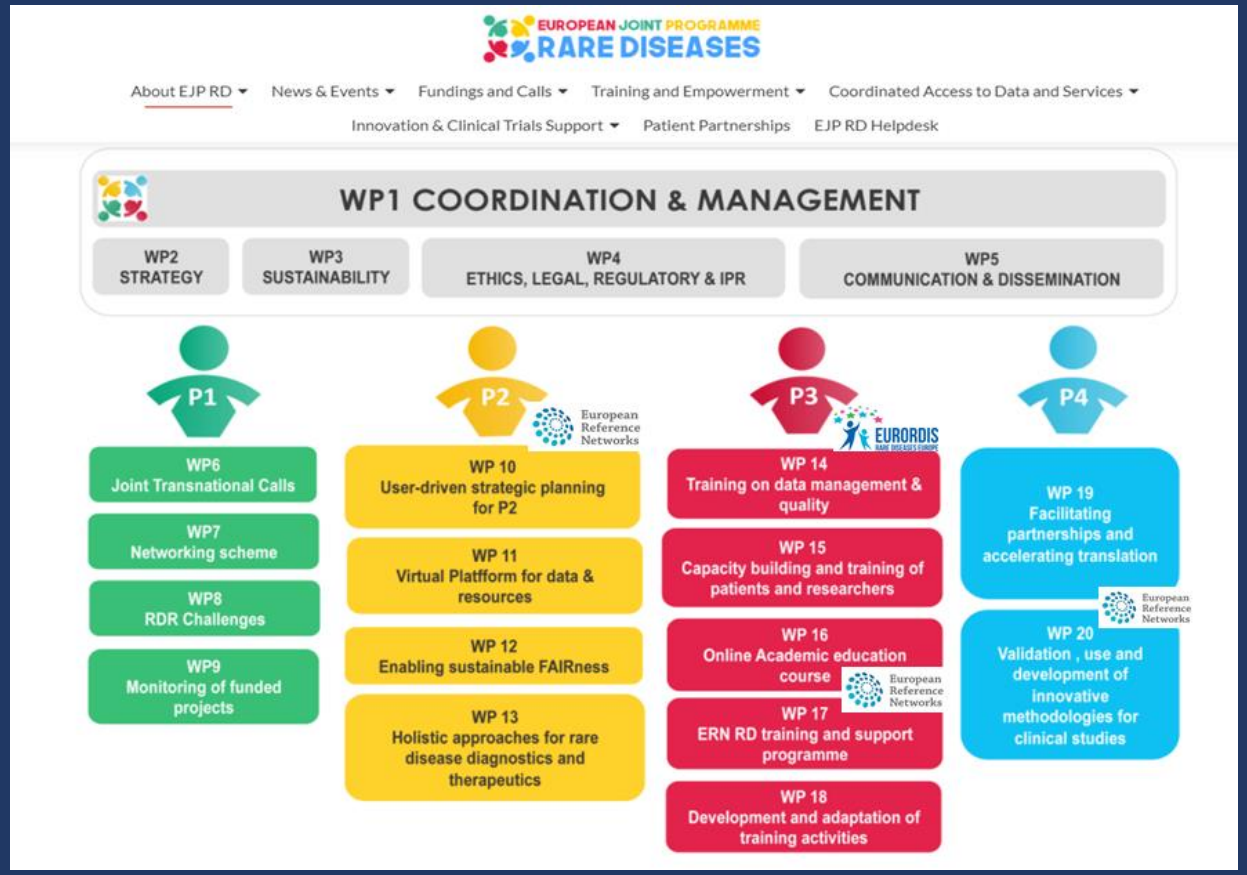
4 ACCELERATING TRANSLATION OF RESEARCH & THERAPY DEVELOPMENT

EJP RD

Funded by the European Union
GA n°825575

EJP RD
what is there for RD stakeholders?

FUNDING, TRAINING & SUPPORT OFFICES



EUROPEAN JOINT PROGRAMME
RARE DISEASES

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/

EUROPEAN JOINT PROGRAMME
RARE DISEASES

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

EUROPEAN JOINT PROGRAMME RARE DISEASES

European Reference 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.

EUROPEAN JOINT PROGRAMME RARE DISEASES

European Reference Networks

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.

EUROPEAN JOINT PROGRAMME RARE DISEASES

European Reference Networks

Are you looking to:

Train your members or gain knowledge in rare diseases research related aspect?

EUROPEAN JOINT PROGRAMME
RARE DISEASES

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

P3

EURORDIS

WP 14
Training on data management & quality

WP 15
Capacity building and training of patients and researchers

WP 16
Online Academic education course

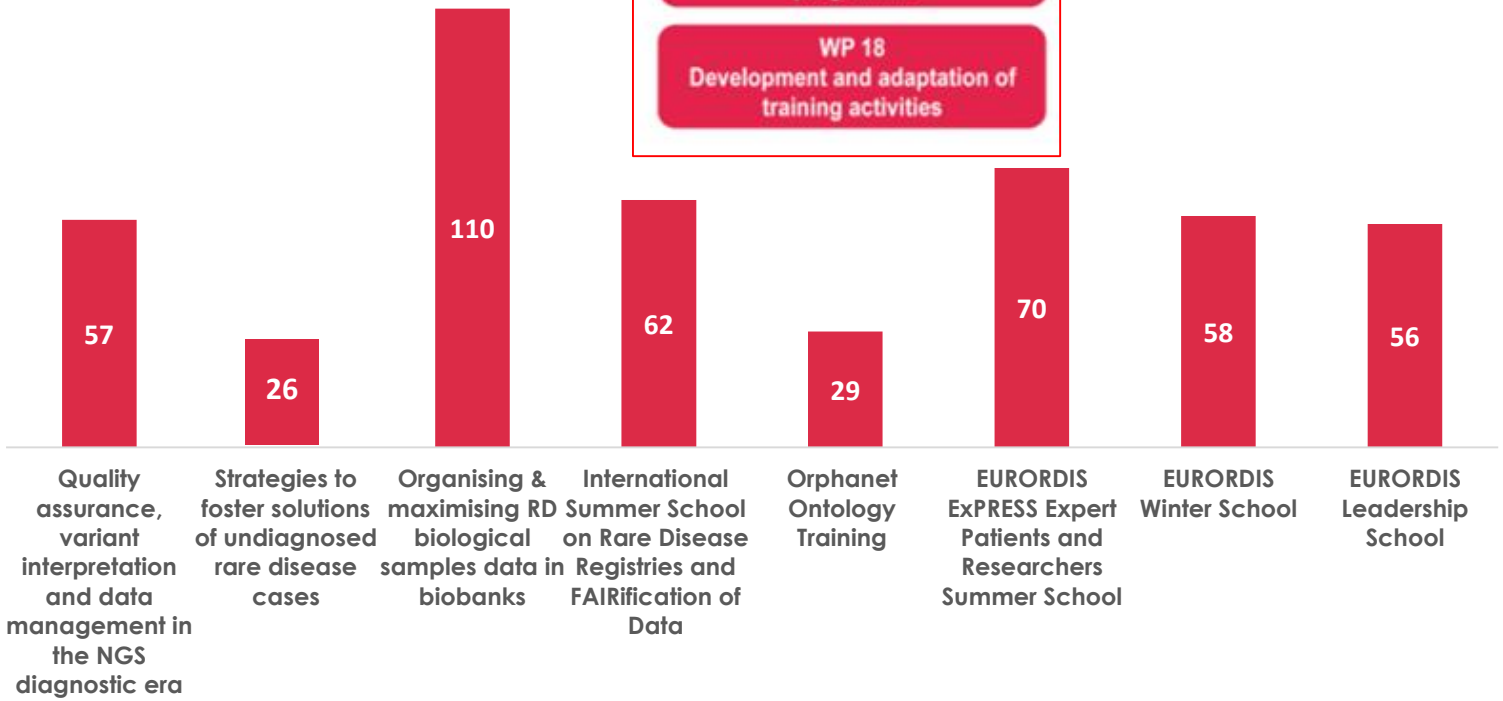
European Reference Networks

WP 17
ERN RD training and support programme

WP 18
Development and adaptation of training activities

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 define d in year 3



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

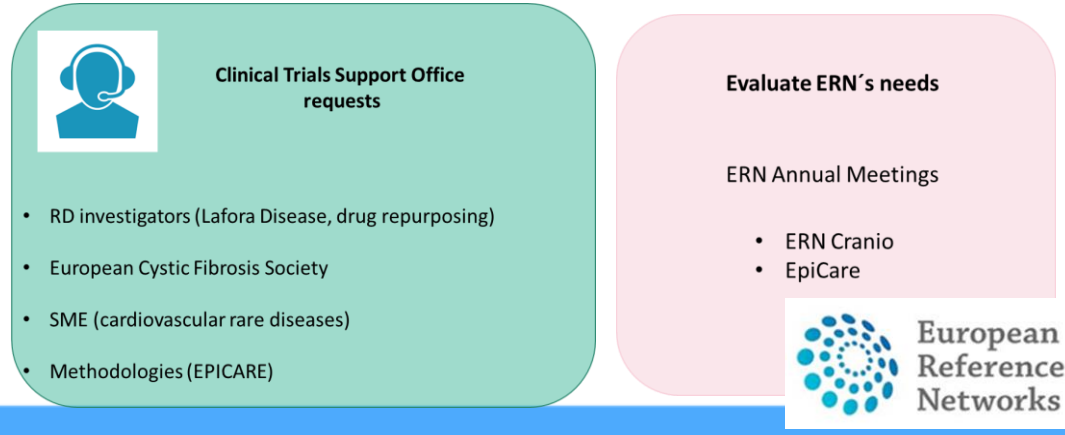
You have idea about possible multinational clinical study but for which e.g. there is no interest from industry? &
You are looking for (e.g):
- advice on how to advance with such project
- advice on how to put in place clinical study with public sponsors
- advice on overall management & regulatory issues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

EUROPEAN JOINT PROGRAMME
RARE DISEASES

EJP RD support offices

Clinical studies support office

- Specialised infrastructures: ECRIN & experts in statistical methodologies (WP20), partners with expertise in multinational CT/Studies
- CT execution planning: country selections, patient recruitment, cost evaluation, regulatory & ethics
- Facilitated access to national support & additional expertise (e.g. paediatric CTs, regulatory, ethics)



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

EJP RD Virtual Platform

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

INFRASTRUCTURES & CATALOGUES
Induced improvement for explotation 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
- Funding

PATIENTS

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

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

THANK YOU

www.ejprarediseases.org

coordination@ejprarediseases.org

helpdesk@ejprarediseases.org

Follow us on social media

@EJPRarediseases

The EJP RD initiative has received funding from the European Union's Horizon 2020 research and innovation programme under grant agreement N°825575

EUROPEAN JOINT PROGRAMME RARE DISEASES