

# PROGRAMME CONJOINT EUROPÉEN SUR LES MALADIES RARES

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# EUROPEAN JOINT PROGRAMME ON RARE DISEASES

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# PAYSAGE MALADIES RARES EN EUROPE

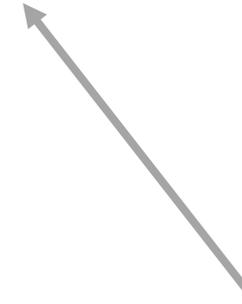
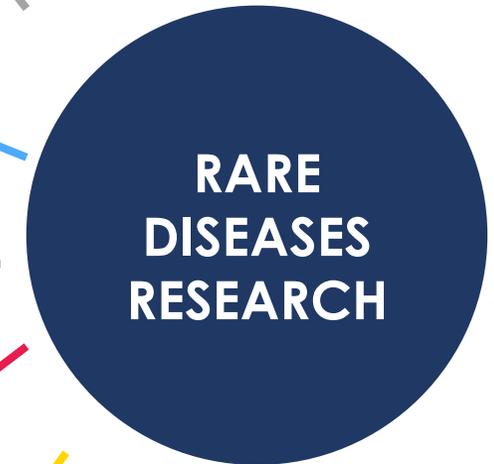
**HEALTHCARE +**  
**European Reference Networks (ERN)**  
More than 900 units from 300 hospitals covering 26 countries

**PATIENT NEEDS**  
**EURORDIS**  
962 patient organisations from 73 countries

**FUNDING**  
ERA-Net Rare (2006-2018) + EC

**INFRASTRUCTURES**  
RD-specific: Orphanet, RD Connect, ERDRI, Solve-RD  
ECRIN, EATRIS, BBMRI-ERIC, ELIXIR, EU-OPENSURE, EPTRI, INFRAFRONTIER,  
Conect4Children

**STRATEGY**  
International Rare Diseases Research Consortium (IRDiRC), EC, Member States





**International,  
European &  
national research  
strategy &  
funding**



**Consortium International de la Recherche sur les  
Maladies Rares**

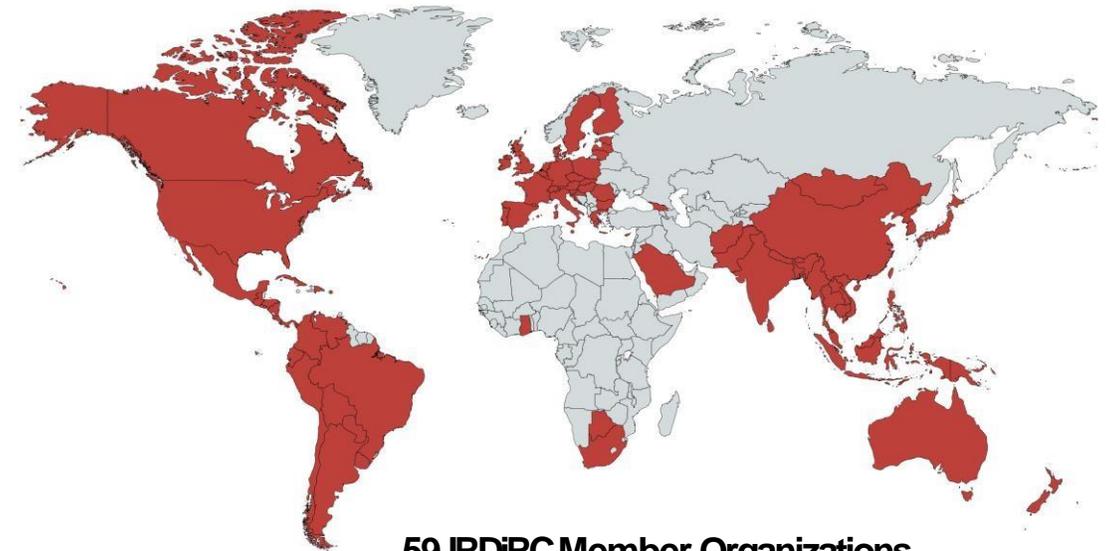
**International Rare Diseases Research Consortium**

# International Rare Diseases Research Consortium (IRDiRC)

**IRDiRC** is a **collaborative network of different type of stakeholders** (funders, research organizations, patients' advocacy organization, and industry) of which **main goal is to provide solutions/recommendations and subsequently implement them** at national/international level to achieve shared objectives.

**IRDiRC does not finance (as an organization) directly research or other type of support;** its members participate in the network for free (their dedicated time).

- ▶ Launched in 2011
- ▶ **3 Constituent Committees** formed by
  - 32 Governmental and non-profit **Funding Bodies**
  - 11 **Companies**
  - 16 **Patient Advocacy Organizations**
- ▶ **3 Scientific Committees**
  - **Diagnostics**
  - **Therapies**
  - **Interdisciplinary**



**59 IRDiRC Member Organizations  
from 22 Countries**

- Europe 21
- North America 20
- South America 1
- Asia 11
- Oceania 3
- Africa 3

# IRDiRC Vision et Objectifs à l'horizon 2027

## Vision

Enable all people living with a rare disease to receive an accurate diagnosis, care, and available therapy within one year of coming to medical attention

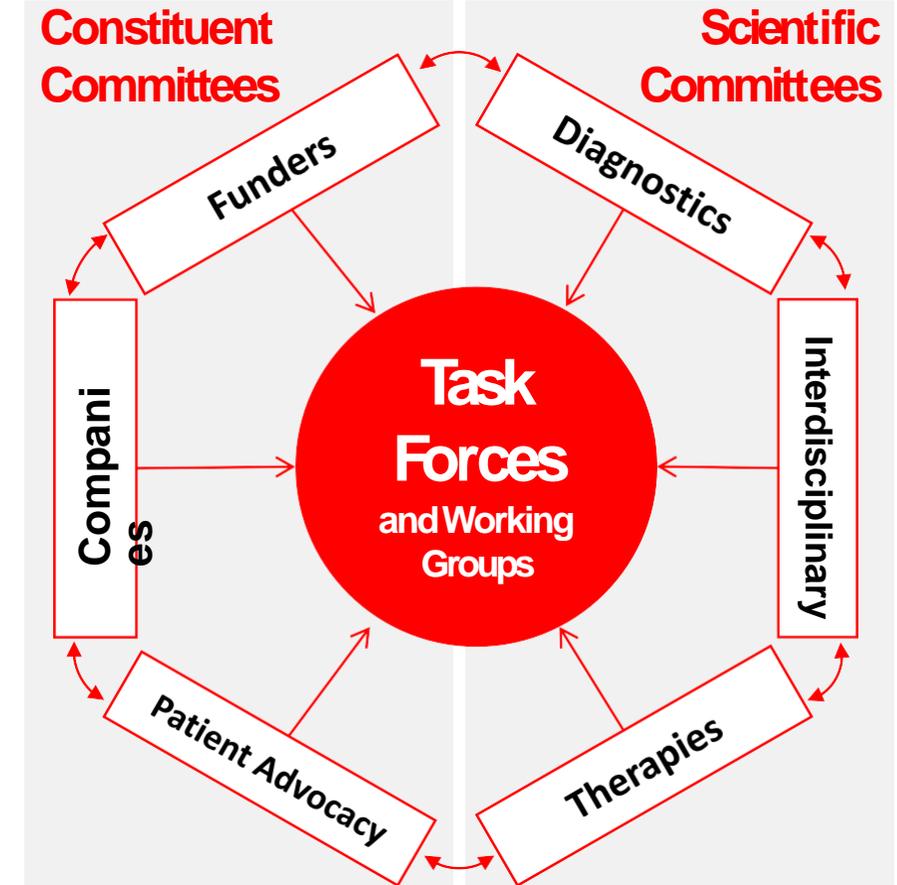
## Goals

1. All patients coming to medical attention with a suspected rare disease will be **diagnosed within one year** if their disorder is known in the medical literature; all currently undiagnosable individuals will enter a globally coordinated diagnostic and research pipeline
2. **1000 new therapies** for rare diseases will be approved, the majority of which will focus on diseases without approved options
3. Methodologies will be developed to **assess the impact of diagnoses and therapies** on rare diseases patients



# IRDIRC – mode opératoire

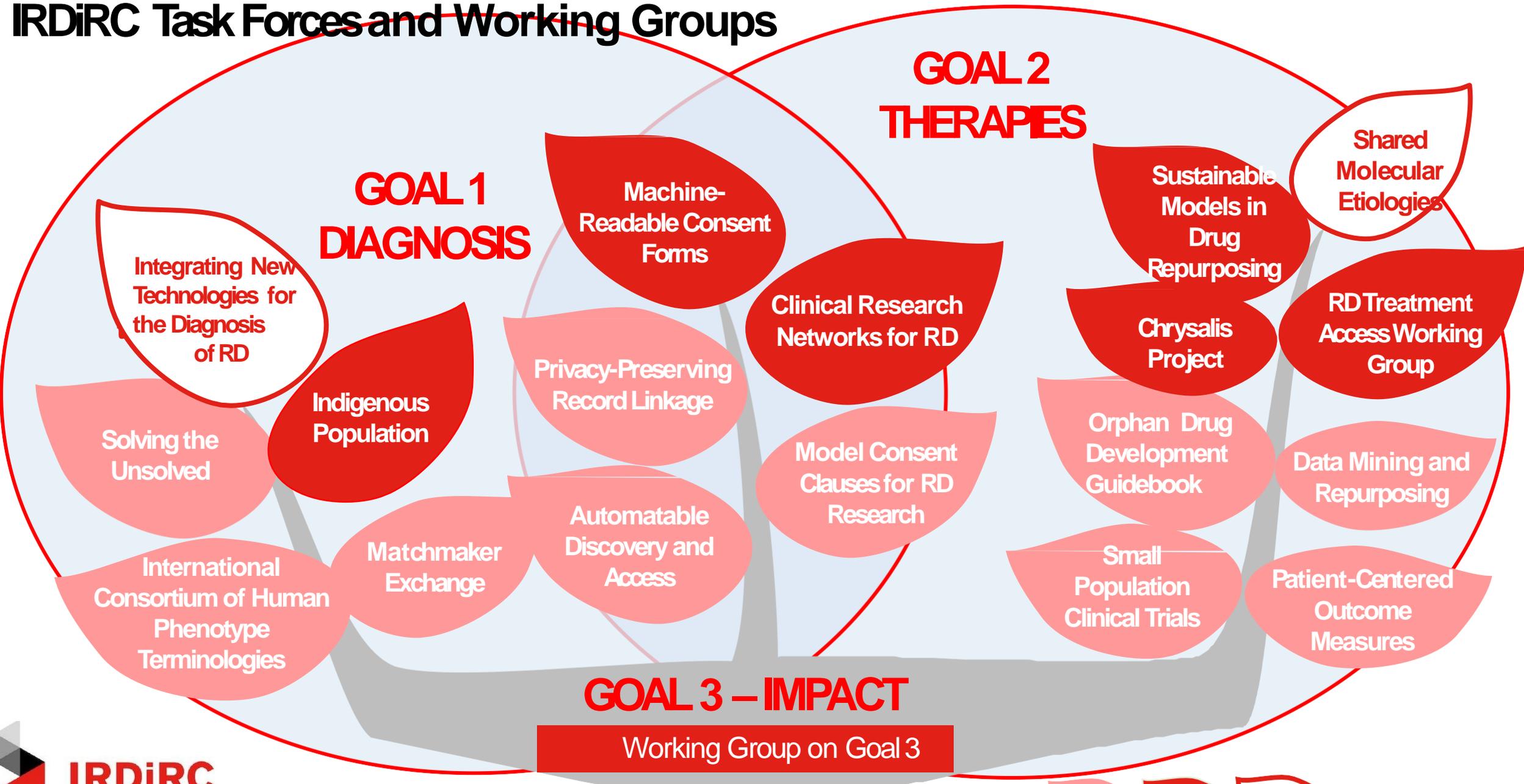
- ▶ The Consortium Assembly agrees upon **priority actions** tackling the three IRDiRC goals proposed by IRDiRC Committees
  - Dedicated **Task Forces** address the selected topics and develop solutions through policy recommendations and guidelines and/or technical applications
  - Working Groups address exploratory projects that may develop into Task Forces or collaborations with other stakeholders



Nomination calls for **Task Forces/Working Groups** and **Scientific Committees** published on [www.irdirc.org](http://www.irdirc.org)

**!!! STAY TUNED !!!**

# IRDiRC Task Forces and Working Groups



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# Objectifs d'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, 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

**1339**  
people

# EJP RD en chiffres

**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

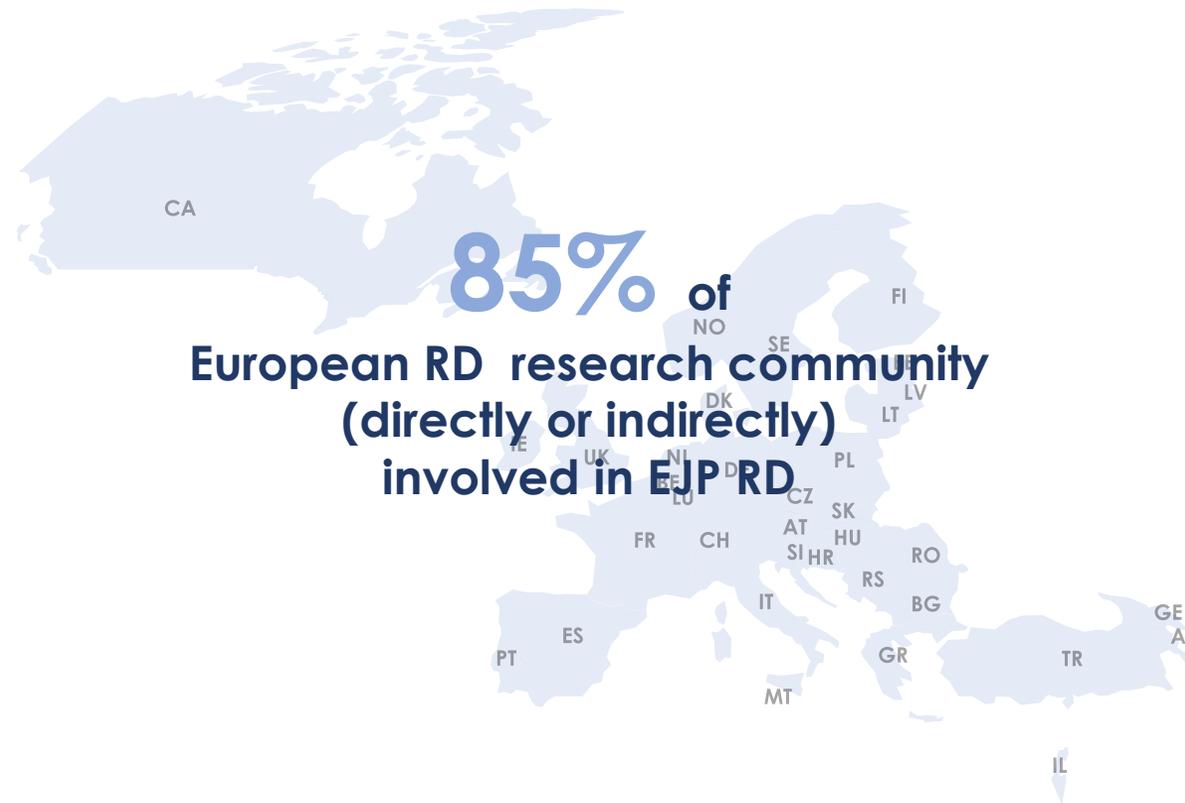


**35** participating countries

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)





# Participation de la Suisse

- 🌈 SCHWEIZERISCHER NATIONALFONDS ZUR FORDERUNG DER WISSENSCHAFTLICHEN FORSCHUNG (SNSF)
- 🌈 LES HOPITAUX UNIVERSITAIRES DE GENEVE (Orphanet Switzerland)
- 🌈 SIB INSTITUT SUISSE DE BIOINFORMATIQUE (Elixir-CH node)
- 🌈 BLACKSWAN FOUNDATION



# EJP RD STRUCTURE

Coordinated by



## Coordination & transversal activities

(strategy, sustainability, ethics, communication)

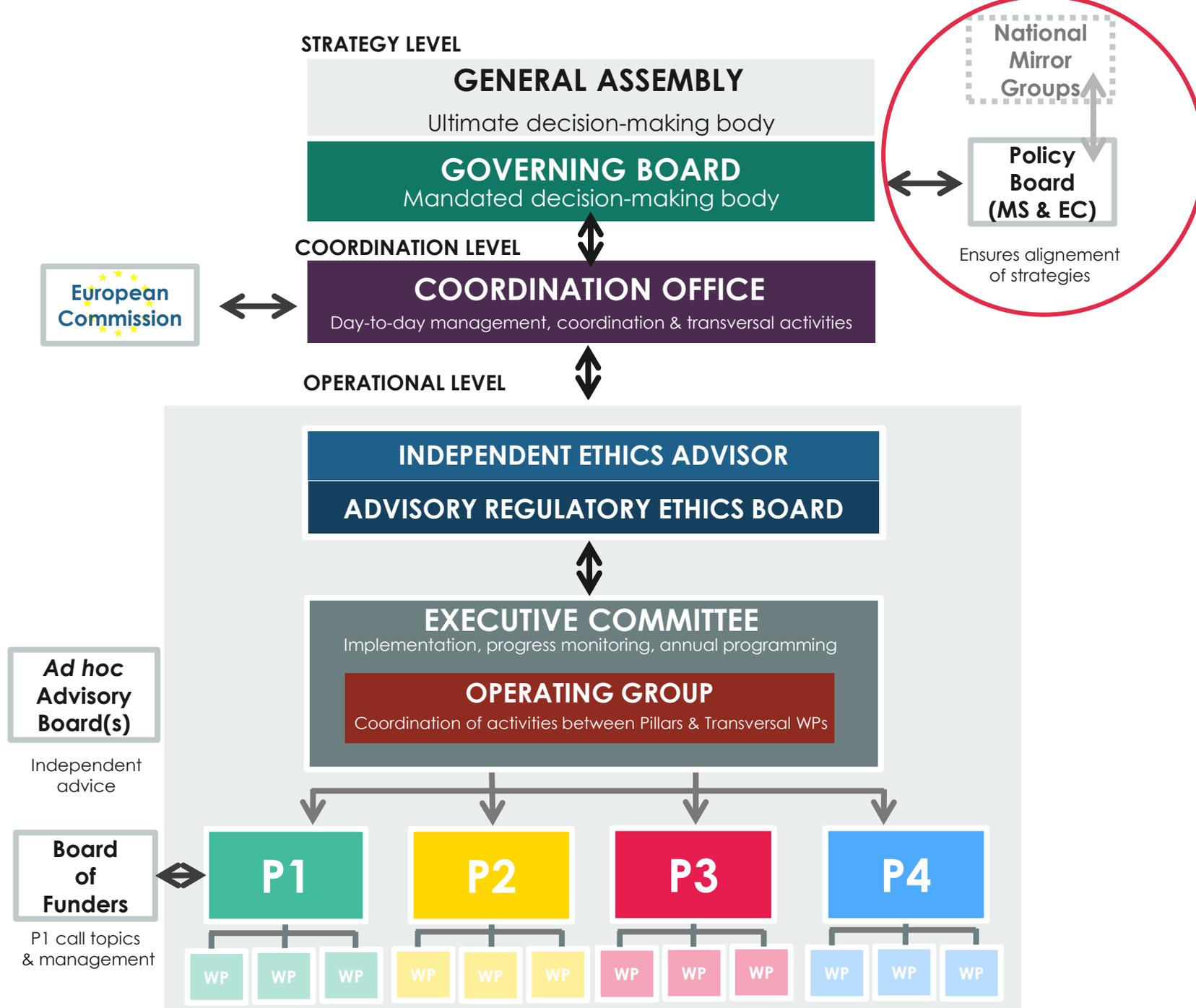
Accelerating of research  
translation & clinical studies

RD research funding

Capacity building &  
empowerment

Access to data, tools &  
services – Virtual Platform







# FINANCEMENT DE LA RECHERCHE COLLABORATIVE

**JTC 2019: Research projects to accelerate diagnosis and/or explore disease progression and mechanisms of rare diseases**

**31 funders**  
**23 countries**



**30.5 million €**  
spent in total  
Including 6 million€ from the  
European Commission

**JTC 2020: Pre-clinical research to develop effective therapies for rare diseases** (launched in December 2019)

**29 funders**  
**22 countries**



**24.5 million €**  
spent in total  
Including 2.7 million€ from  
the European Commission

**39 diseases/groups**

Including at least 6  
with prevalence  $1 < 1000000$

**Successful**  
**« widening »:** 14 new  
partners included in full  
proposals



# Support beyond funding

## Improve Involvement of Patients in Research

- At all stages: topics definition, evaluation, patient-driven research projects
- In the Joint Transnational Call 2019, **36%** of funded projects involved PAOs

In **2020**, it increased to **78%** of the funded projects



## Networking Support Scheme

- 28 applications (3 rounds)
- 11 recommended for funding (2 rounds)
- 302 419 € invested

## Rare Disease Research Challenges

- 4 challenges set up with industry partners
- 10 applications received
- 3 recommended for funding

## ERN Support Scheme

- WORKSHOPS
- 15 applications (2 rounds)
- 7 recommended for funding (2 rounds)
- FELLOWSHIPS
- 27 applications (2 rounds)
- 6 fellows funded (1st round)

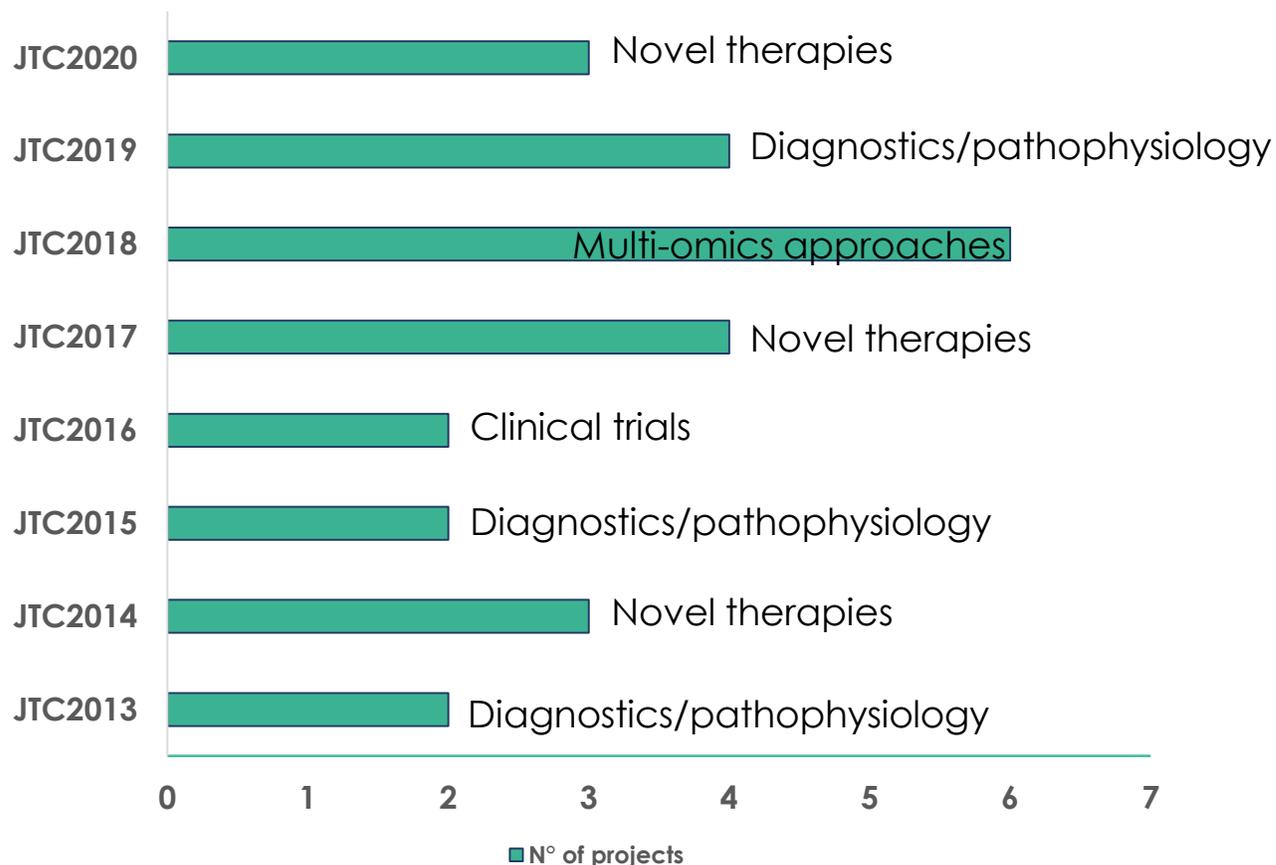


# Joint Transnational Calls – participation de la Suisse

Involved in funding activities since 2013 (via SNSF) → 2013-2018 E-Rare ERA-Net + 2019-now EJP RD

Budget → 6 012 929,72 CH invested

N° of funded projects per call



## Institutions involved in 27 funded projects:

- University of Zürich (8 projects)
- École polytechnique fédérale de Zurich (3)
- University of Lausanne (3)
- Ecole Polytechnique Federale de Lausanne (2)
- University of Geneva (2)
- University of Bern (1)
- University of Basel (5)
- Kantonsspital St. Gallen (1)
- Institute for Research in Biomedicine (1)
- Institute of Molecular and Clinical Ophthalmology, Basel (IOB) (1)

# PLATEFORME VIRTUELLE DES DONNES, RESSOURCES ET OUTILS



# Virtual Platform (VP)

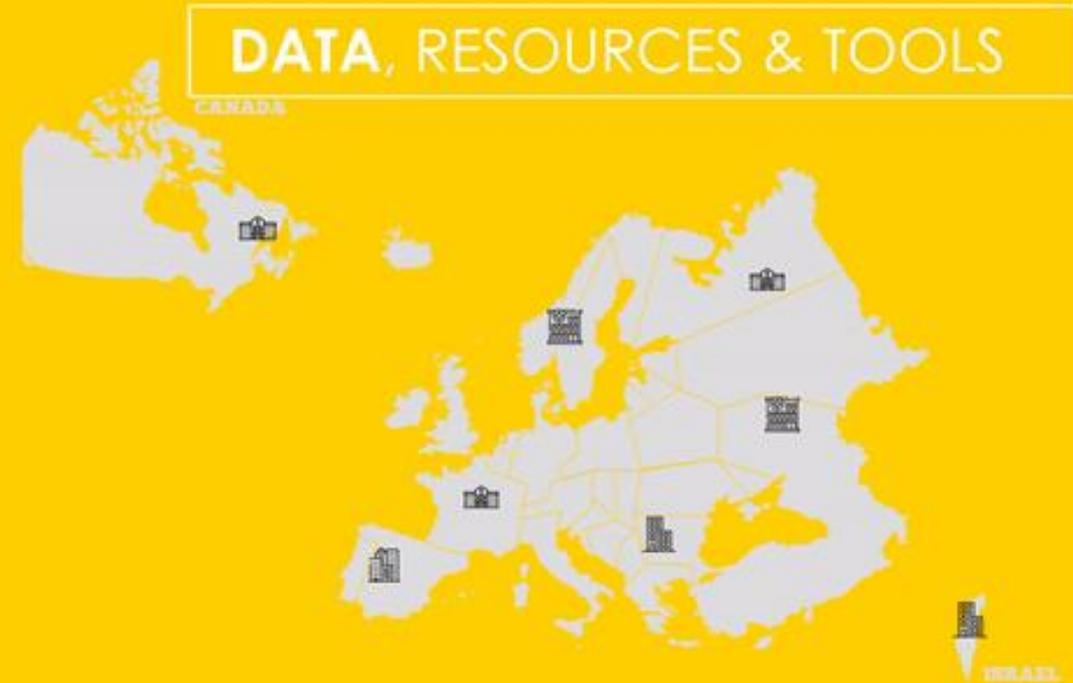
**Federated**

**Standardized**

**GDPR-compliant**

**Sustainable**

**Quality assessed**



**F**indable  
**A**ccessible  
**I**nteroperable  
**R**eusable

# What is the issue?

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



# What is needed to provide solutions?

## Enhancement of existing resources

- ✚ Making resources more RD friendly
- ✚ Improving inter-connectivity

## (Meta)Data Models

- ✚ Common models to link & exchange data across multiple IT systems

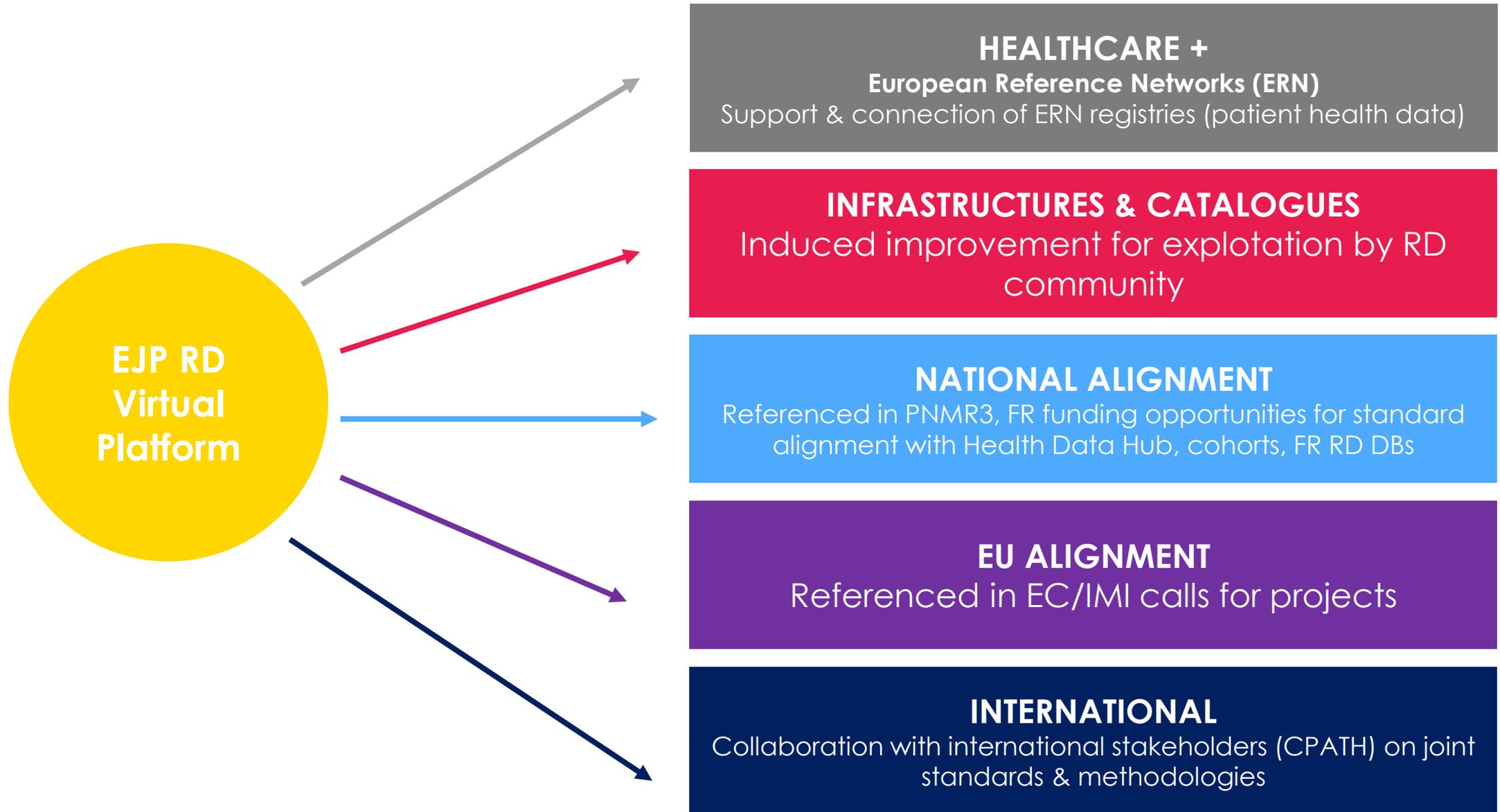
## EJPRD asset for other health data projects

Develop **tools for discovery of existing resources** and available data

Develop **tools** and apply standards to allow **Access to data for reuse**

RD portal for exploitable rare disease pathways enabling multi-omics analysis

# EJP RD Virtual Platform – international reference



# FORMATION & EDUCATION

## 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
- **7 ERN workshops** financed
- **15 ERN fellowships** attributed
- **1st Online education MOOC** created

## Free e-learning academic course on RD

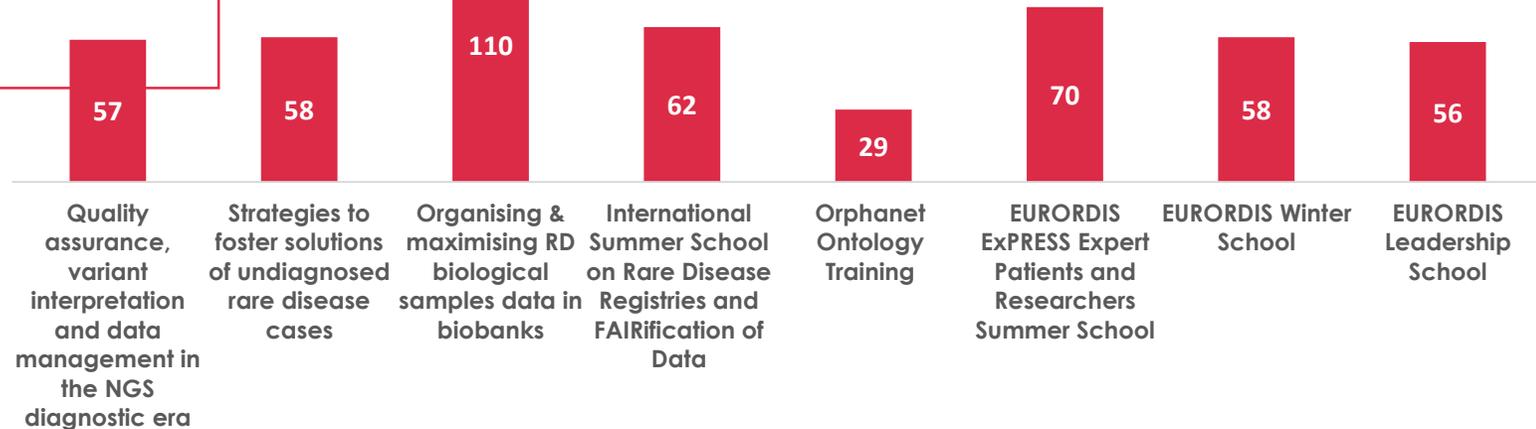
Module 1: RD  
Diagnosis

Module 2: RD  
Innovative  
personalized  
therapies

Module 3: RD  
translational  
research

Module 4 & 5:  
to be defined  
in year 3

50 – 60 hours of training



**Diversity & transversality**

**Large target span**

**No 1-shots but regular revision**

**Leverage national capacity**

# EJP RD trainings

- 🌐 **Educational academic course:** on transversal and multidisciplinary aspects of rare diseases research will be made available for all stakeholders in a fully online format. The course foresees the development of 5 modules. **The first module on Diagnosis was just launched!**

A promotional graphic for an online course. The background is a photograph of a person's hands writing in a notebook on a desk with a laptop and a coffee cup. The text is overlaid on the image.

**ONLINE COURSE**

**Diagnosing Rare Diseases:  
from the Clinic to Research and back**

This course offers an exploration of key questions around rare disease diagnostic research and issues today, as well as insights into patient experiences.  
Join today!

Duration: 5 weeks      Weekly study: 3 hours

# ACCELERER TRASLATION DE LA RECHERCHE & ET LES ETUDES CLINIQUES



## Regulatory

- Launch of 3 demonstration projects validating novel methodologies in small population CTs (proof required by regulators)
- Direct collaboration with regulators: EMA and EU Innovation Network (national competent authorities)
- International innovation: collaboration with Critical Path Institute (co-financed by FDA & industry) to support regulatory research for the benefit of RD patients



## EJP RD removes barriers to innovation

### Accelerated translation of research results

- Accompany research projects
  - Expert mentoring from their conception and throughout their lifetime & facilitate next steps
  - Provide tools (based on use-cases) free of charge → innovation management toolbox
  - Follow-on funding planning & support

### Clinical studies support office

- Specialised infrastructures: ECRIN & experts in statistical methodologies, 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)

# EJP RD – UN RESEAU D'EXPERTISE

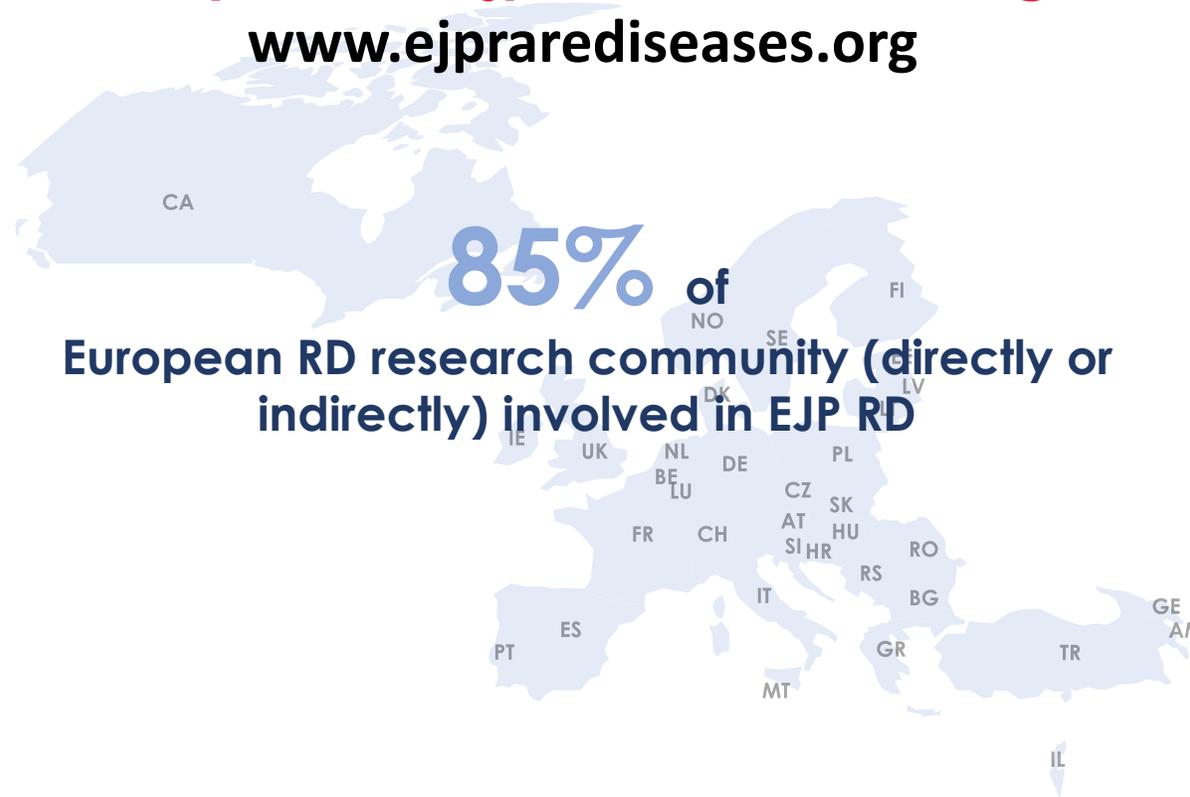
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[helpdesk@ejprarediseases.org](mailto:helpdesk@ejprarediseases.org)  
[www.ejprarediseases.org](http://www.ejprarediseases.org)



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# EJP RD – single entry point & solutions for all

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

# MERCI

[www.ejprarediseases.org](http://www.ejprarediseases.org)

[coordination@ejprarediseases.org](mailto:coordination@ejprarediseases.org)

[helpdesk@ejprarediseases.org](mailto:helpdesk@ejprarediseases.org)

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