



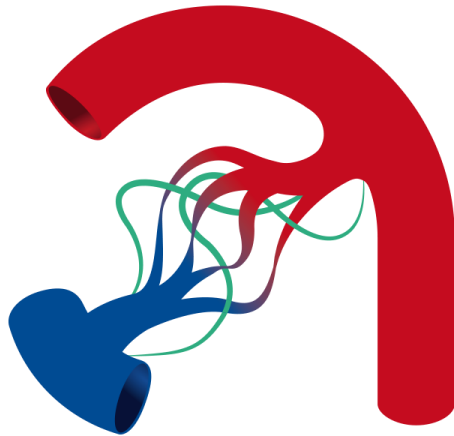
**European  
Reference  
Network**

for rare or low prevalence  
complex diseases



**Network**

Vascular Diseases  
(VASCERN)



 [#VASCERNdays2020](https://twitter.com/VASCERNdays2020)



**08.30-9.00 Research:  
European Joint  
Programme on Rare  
Diseases (EJP RD): calls and  
opportunities & Q&A (Daria  
Julkowska and Yanis**

**Mimouni)**

# EUROPEAN JOINT PROGRAMME ON RARE DISEASES (EJP RD)

VASCERN Days  
22–24 of October 2020



# 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, 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

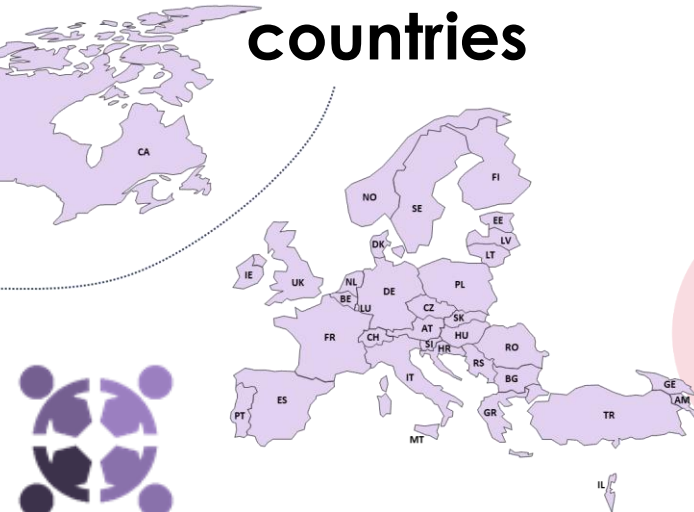
**85%** of  
**European RD community**  
(directly or indirectly)  
involved in **EJP RD**

**750** people:

**650** Scientifics  
**100** Admin

**35**

participating  
**countries**



**88** 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**

**And 100% of the associated networks**

**EURORDIS:**  
884 RD patient  
organisations  
72 countries

**EATRIS**  
13 main national nodes

**INFRAFRONTIER**  
23 partners  
15 countries

**24 ERNs:**  
300 institutions  
>950 healthcare units  
26 countries

**ECRIN**  
12 main national nodes

**ELIXIR**  
220 research  
organisation  
23 partners

**BBMRI**  
1 international partner  
21 main national nodes  
20 countries



# EJP RD STRUCTURE

Coordinated by



# EJP RD, organised in four Pillars with central coordination, is a supreme instrument to grant high-level strategic organisation empowering all stakeholders and performance of RD research activities

## Pillar 1: Collaborative RD research funding

- Funds RD collaborative research
- Supports networking & sharing of knowledge between clinicians, scientists and patients
- Fosters RD challenges through private-public partnerships

- Builds the capacity of RD stakeholders by providing top-level trainings available to all
- Supports ERNs in delivery of cross-cutting education programmes fostering new generations of clinicians
- Addresses training needs & empowers EU 13 countries

## Pillar 3: Capacity building & empowerment

## Pillar 2: Coordinated access to data, tools and services

- Builds FAIR-based, user driven Virtual Platform of data, resources & services open to all
- Accelerates RD research by ensuring access to powerful substrates & tools
- Provides innovative approaches to RD diagnosis through application of systems biology

## Pillar 4: Accelerating the translation of research results and clinical studies

- Facilitates partnerships & accelerates translation of research results by providing mentoring & innovation management assistance
- Ensures tailored support for design & planning of RD clinical studies
- Advances validation & use of innovative methodologies for clinical studies

## Coordination & Transversal activities

- Ensures the overall research & innovation strategy
- Engages with policy makers and guarantees expansion to relevant stakeholders
- Measures the EJP RD results, performance and ensures compliance
- Translates each action in dissemination & communication item
- Brings future sustainability/ business solutions

**EJP RD**

-

**what is there for me?**

# FUNDING, TRAINING & SUPPORT OFFICES



***Project idea: going beyond clinical outcome measures  
– how to improve patient healthcare outcomes?***

Clinicians + patients + social sciences experts

# Joint Transnational Call 2021

**Objective: support transnational, innovative, and interdisciplinary Social Sciences and Humanities research projects to improve healthcare implementation and everyday life of people living with a rare disease**

## List of topics:

- **Health & social care services** research to improve patient and familial/household health outcomes
- **Economic Impact of Rare diseases**
- **Psychological and Social Impact of Rare diseases**
- Studies addressing **the impact/burden of the delay in diagnosis and of the lack of therapeutic intervention.**
- **e-Health in rare diseases:** Use of innovative technology systems for care practices in health and social services
- Development and enhancement of **health outcomes research methods** in rare diseases
- Effects of **pandemic crisis** and the **global outbreak alert** and response on the rare disease field, and the emergence of innovative care pathways in this regard.

**Other research topics are possible as long as they focus on SSH research and are not in the excluded topics list.**



**The call is scheduled to open in December 2020  
with a pre-preposal submission deadline in February 2021**

# Joint Transnational Call 2021

## ✿ The following approaches and topics are excluded from the scope of the call:

- Interventional clinical trials to prove efficacy of drugs, treatments, surgical procedures, medical technology procedures. This also includes studies comparing efficacy, e.g. B. two surgical techniques or therapies. Clinical phase IV pharmacovigilance studies cannot be funded either.
- Studies on the exclusive testing of the safety of medical devices.
- Health technology assessment reports (HTA) for a specific product
- Projects focussing on meta-analyses and systematic reviews
- Creation of new registers or establishment of new long-term cohorts and / or promotion of existing registers or long-term cohorts.
- Development of new digital or technological tools.
- Projects to accelerate diagnosis and/or explore disease progression and mechanisms of rare diseases as covered in EJP RD JTC 2019.
- Development of new therapies as covered in EJP RD JTC 2020.
- Projects focussing only on rare neurodegenerative diseases which are within the main focus of the Joint Programming Initiative on Neurodegenerative Disease Research (JPND; <http://www.neurodegenerationresearch.eu/>). These are: Alzheimer's disease and other dementias; Parkinson's disease (PD) and PD-related disorders; Prion disease; Motor Neuron Diseases; Huntington's disease; Spinal Muscular Atrophy and dominant forms of Spinocerebellar Ataxia. Interested researchers should refer to the relevant JPND calls.
- Rare infectious diseases, rare cancers and rare adverse drug events in treatments of common diseases.

**MORE INFORMATION:** <https://www.ejprarediseases.org/index.php/early-announcement-jtc2021/>

# EJP RD internal call for projects on innovative statistical methodologies to improve RD clinical trials in limited populations

- Aims to develop innovative statistical methodologies to address unmet needs associated the development and the analysis of clinical trials in limited populations
- Submission of projects is limited to partners from institutions beneficiaries of the EJP RD. This **includes all 24 ERNs** and Linked Third Parties or, parties bound by the Network Agreement with the beneficiary institution (and thus being able to integrate EJP RD project as Linked Third Party at later stage)
- **Opening of the call is foreseen in late November 2020**
- A networking/matchmaking event will be organised in January 2021

*Are you looking for (e.g):*

*Gathering of experts & patients to discuss and share knowledge?*

*Expanding your network to include new stakeholders?*

*Finding ways to gather and support future consortium that plans to apply to EC calls?*

# Networking Support Scheme

- 🌟 **Objective:** encourage sharing of knowledge on rare diseases and rare cancers
  - ✂ to support health care professionals, researchers and patient advocacy organizations with a networking grant to re-organize themselves into transnational (clinical) research networks
  - ✂ that focus on a (group of) rare disease(s), a (group of) rare cancer(s) or on cohorts of undiagnosed patients that are suspected of suffering from a rare disease
- 🌟 **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**
- 🌟 30K€ max per event
- 🌟 **Applications:**
  - ✂ open on a continuous basis. The applications will be collected every three months and the eligibility will be checked. First collection of application: **December 1, 2020 at 14:00 (CET)**
  - ✂ open to all countries involved in EJP RD (Applicants from Canada are not eligible for funding)
  - ✂ the consortium submitting an application must involve a **minimum of three eligible applicants** from **at least three different countries** participating in the EJP RD at the time of the application. A **maximum of 10 partners** per application is eligible

**MORE INFORMATION:** <http://www.ejprardiseases.org/index.php/networking-support/>

*Are you looking to:*

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

# Research Mobility Fellowship

🌟 **Aim:** financially support **PhD students** and **medical doctors** working in **ERN-member institutions** to undertake **short scientific visits** (secondments) up to 3 months fostering specialist research training outside their countries of residence and within one of the ERN host institutions. Through this training measure the fellows should acquire at their host (secondment) institution new competences and knowledge related to their research on rare diseases and with benefit to their ERN.

🌟 **Applicants/Application profile:**

- ✘ PhD students with a minimum of one year of research experience OR physicians having finished their first year of specialist training
- ✘ Be affiliated to an **ERN Full Member or to an ERN-Affiliated Partner Institution** from one of the 24 [ERNs](#) at the time when the application is submitted, as well as during the proposed period of the training stay
- ✘ The host (secondment) institutions must be Full or Affiliated Members of an ERN at the time when the application is submitted, as well as during the proposed period of the training stay
- ✘ Added value to ERN of the mobility stay



The call opened on October 1st 2020

**Deadline for submission: 13th November 2020**

**MORE INFORMATION:** <https://www.ejprarediseases.org/index.php/training-and-empowerment/ern-trainings/>



*Are you looking to:*

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

# Research Training Workshop

🌟 **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. Training themes may include innovative research methodologies, diagnostic research methodologies, interdisciplinary treatment approaches, such as gene therapy and transplantation, etc. Moreover, the workshops will be aiming to provide a cross-ERN added value.

The workshops will be implemented as two day events. Topics can be proposed by the ERNs or by investigators belonging to EJP RD beneficiary institutions.

🌟 **25k€ max/event**

🌟 **Applicants/Application profile:**

- ✖️ A affiliated to any EJP RD beneficiary institution
- ✖️ A affiliated to an ERN Full Member
- ✖️ A affiliated to an ERN Partner institution at the time when the application is submitted, as well as during the period of the execution of the workshop



2 calls per year - the call closed on October 12 2020  
**Next call will open in Q1 2021**

**MORE INFORMATION:** <https://www.ejprarediseases.org/index.php/training-and-empowerment/ern-trainings/>

*Are you looking to:*

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

# Other EJP RD trainings

- 🌟 **Data management & quality:** the training activities cover a variety of topics including but not limited to *variant interpretation, data management, registries, FAIRification process, Orphacodes, biobanking, and undiagnosed cases.*
- 🌟 **Patients & Researchers training:** training modules dedicated to building the capacity of the patient community and other key stakeholders, including training “expert” paediatric patients (new in 2021) on rare diseases, paediatric medicines development and clinical research.
- 🌟 **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 implementation of the first modules are expected by the end of 2020.

**MORE INFORMATION:** <https://www.ejprarediseases.org/index.php/training-and-empowerment/>

*You have project and/or preliminary results for e.g. new therapy/biomarker/device*

*&*

*You are looking for (e.g):*

- further regulatory support*
- advice on how to get interest of potential sponsor or derisk your project*
- further financial support*

# Support for innovation & research translation

## Facilitating partnerships and accelerating translation for higher patient impact

### Innovation management toolbox

- Integration of various resources supporting research translation (including the IRDiRC Orphan Drug Dev. Guide)
- Connection with Pillar 2 to make it interoperable with the Virtual Platform

### Support for translation & innovation mentoring

- Pool of mentors to support the translation of research projects
- Assessment of the translation needs of E-Rare, EJP RD funded projects and ERN projects

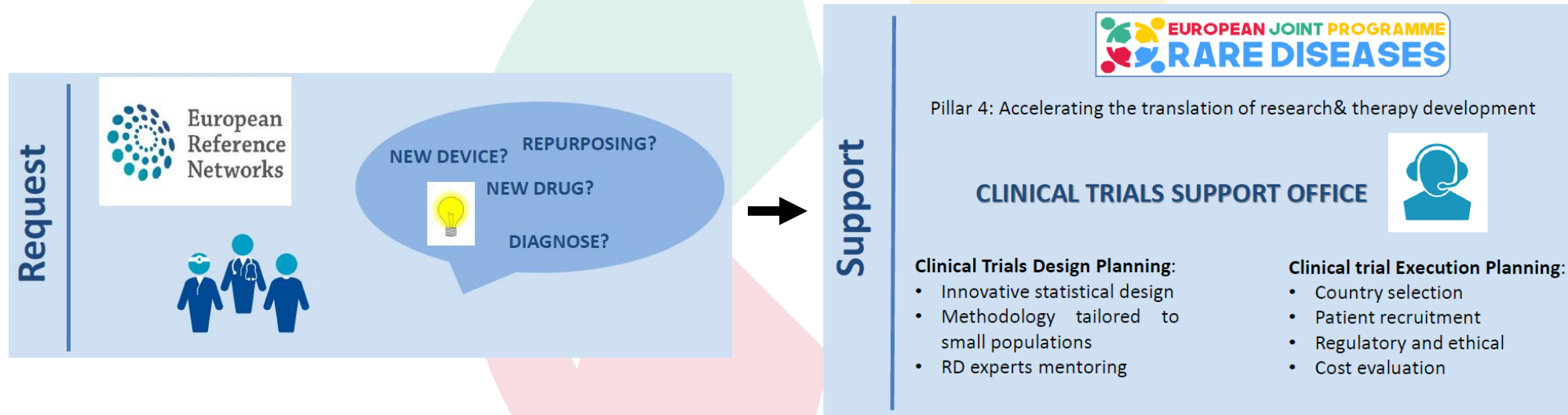
*You have idea about possible multinational clinical study but for which e.g. there is no interest from industry?*

*&*

*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*

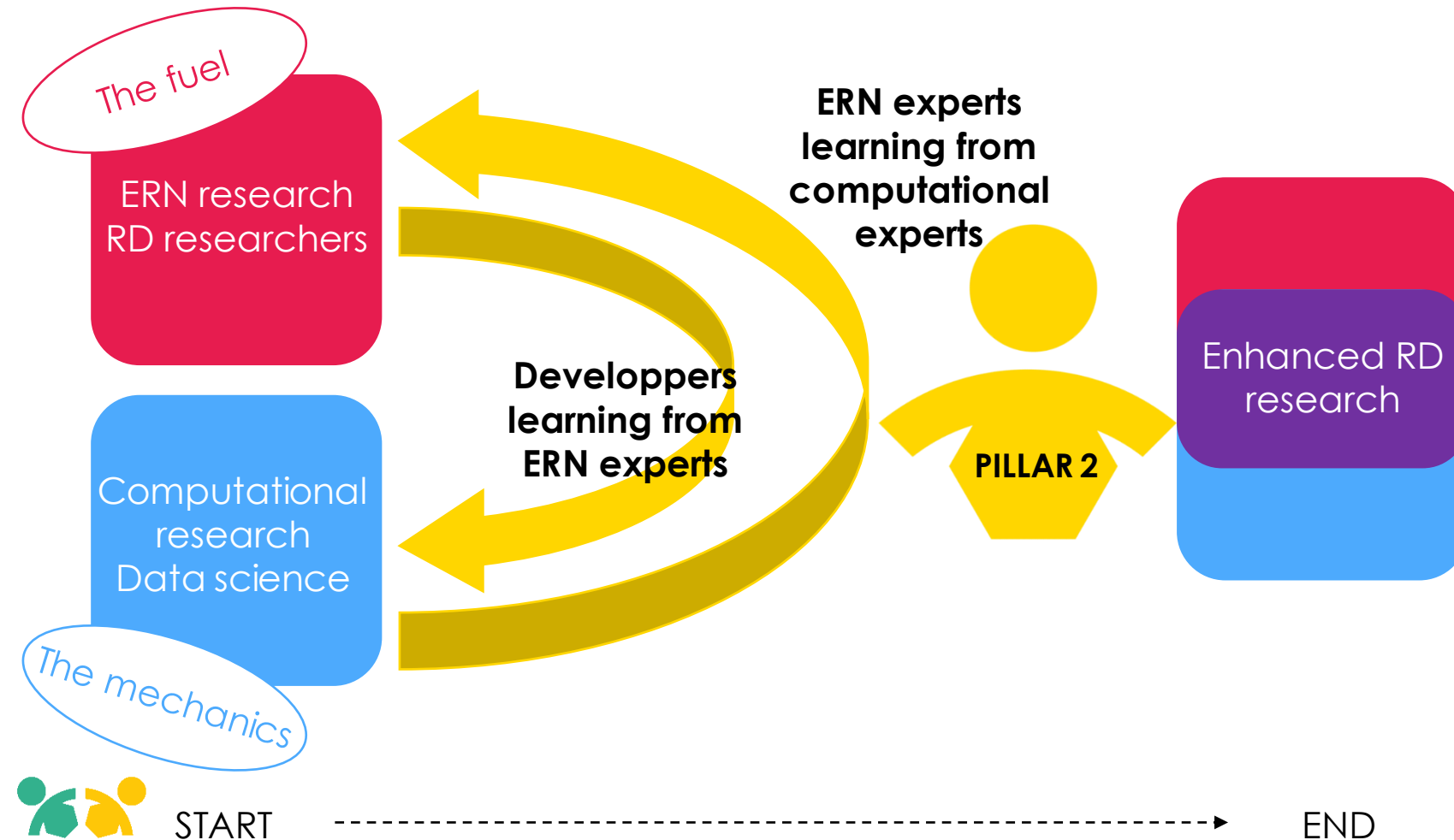
# Multinational CTs support office





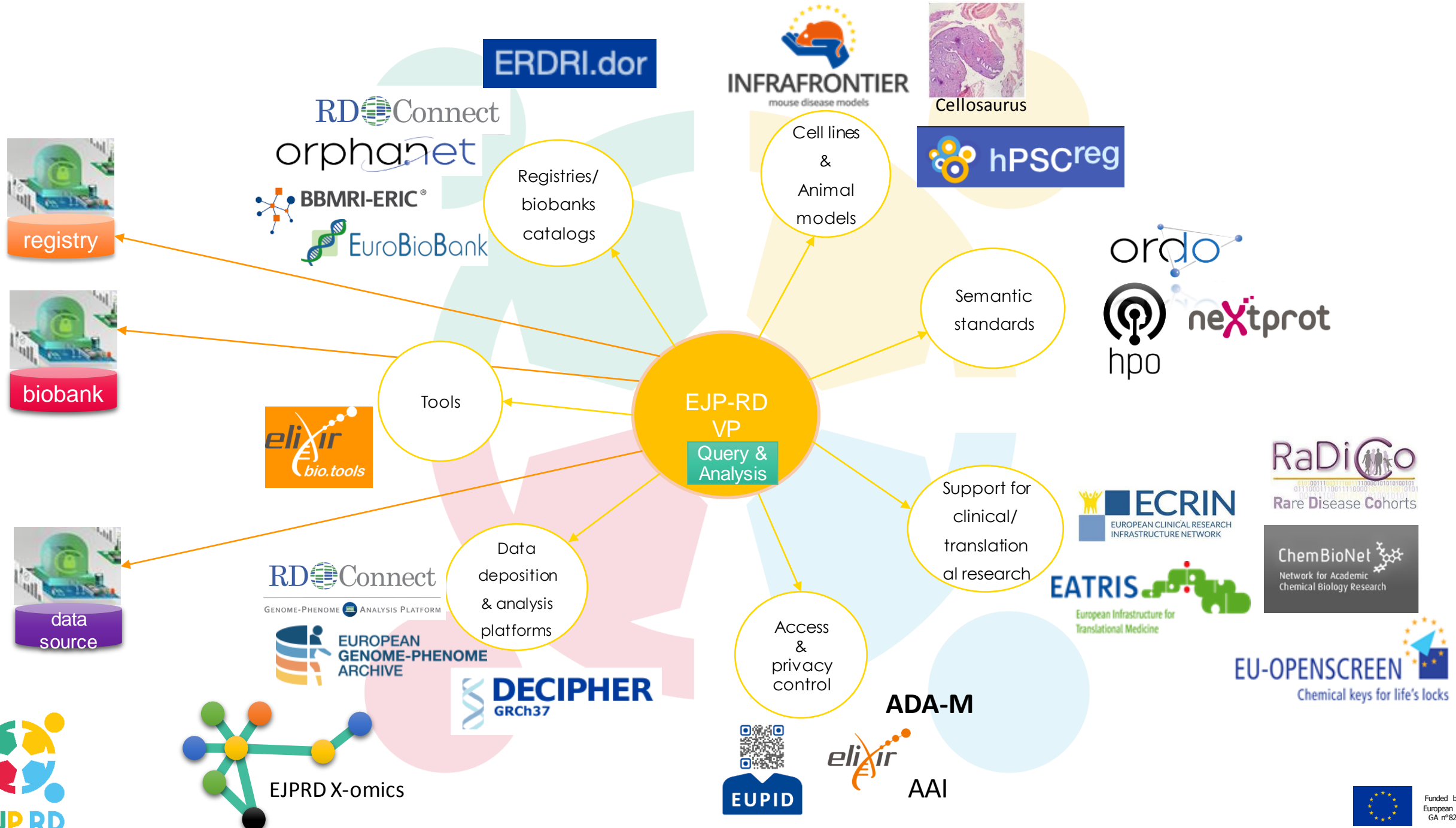
# VIRTUAL PLATFORM & RELATED SERVICES

# Pillar 2 target: FAIR-based virtual platform

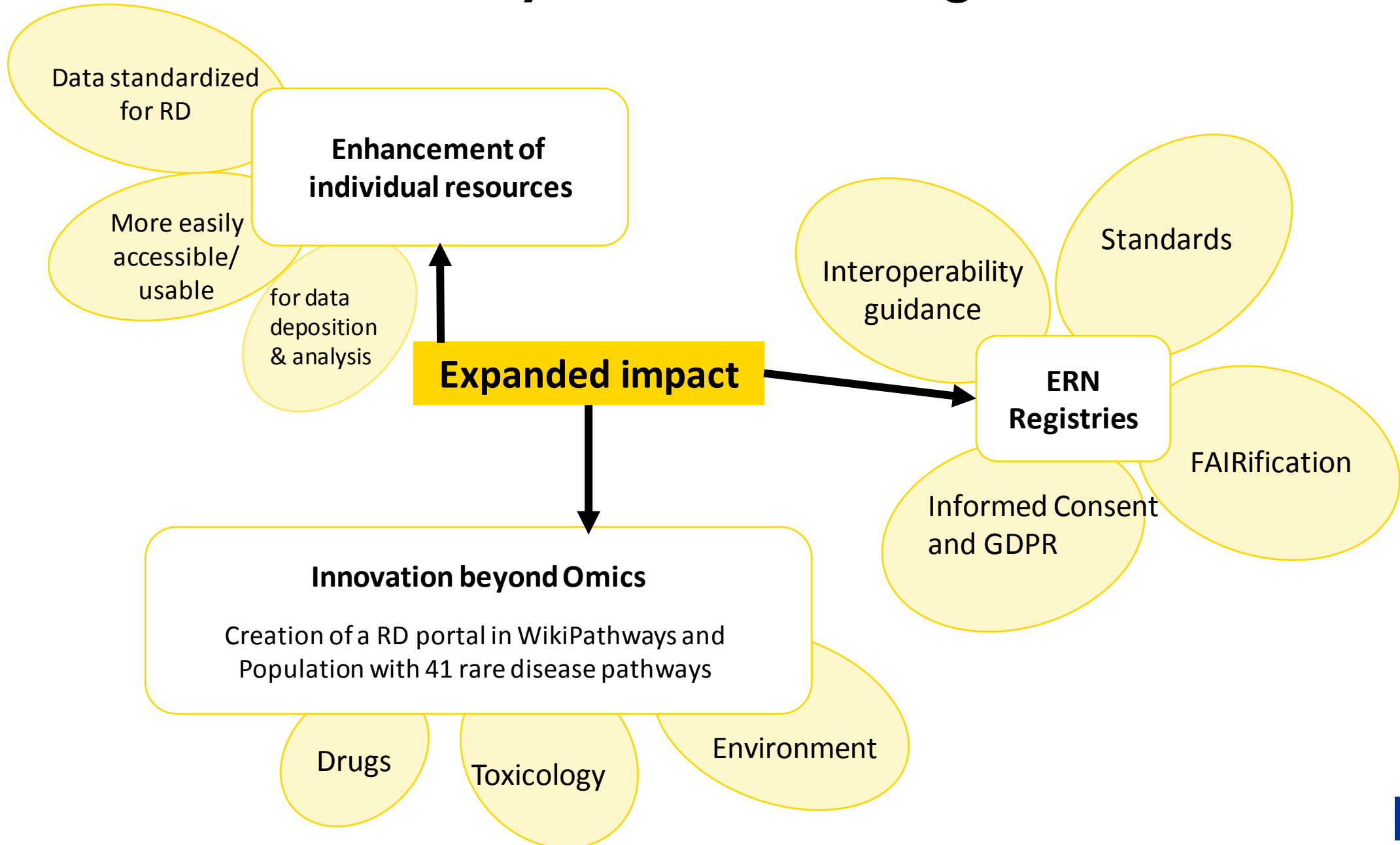


A powerful substrate for translational research:

- 🌟 **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



# Beyond VP building



# Pillar0 (WP4) – Registries Support

## Registry Ethics Submission

- ✿ The **Working group for Informed Consent Process Facilitation (including the AREB)** has worked to provide solution to ERN submitting their registries for Ethics Committees approval (using the ERKNet 1+ year plus experience that submitted to 35 **Ethics Committees**).
- ✿ **Finalising a harmonised framework (15/11/2020):**
  - ✿ provide an **addendum to the generic EU ICF to ERN registries** to be **fully GDPR compliant and compatible with secondary research use** including EJP RD research data focus
    - ✿ the web link to the addendum could be printed on the CPMS/Registry ICF to allow patients to have the additional missing information);
  - ✿ To provide **information for minors**;
  - ✿ To provide the **current experience from ERKnet** with the summary table and the information on the addendum to the other ERN requests

# Pillar2 – Registries Support

## FAIRification

🌟 Meetings between the FAIRification Stewards and the teams of the 5 VASCERN Registries

- 🔗 To **complete the inventory of implementations** (for FAIRer Registries)

- 🔗 **Plan the extensive support** required for some registries (Vascular EDS [RaDiCo])

🌟 EJP RD FAIRification **Hackathon material shared:**

- 🔗 Link: [EJPRD Workshop 2020-06 Hackathon Implementation CDE semantic model for ERNs](#)

- 🔗 Useful for Redcap platform development to have full FAIR semantic model, FDP (redcap to RDF conversion possible in 2 different methods)

🌟 ERNs FAIRification Experience Sharing is being planned through MsTeams

# Pillar2 – Involvement in Use Cases

Activation

Team Channel  
creation

Identify  
Champions

Identify users

TC use case  
description

Minimum  
Viable Product

Overall  
architecture

## Counting

Counting persons with specific rare conditions (Proof of Concept of the VP)

**May need stakeholders (registries) feedback**

## Consent

A structured manner of encoding consent conditions in a human and machine readable format

**Will need more stakeholders (registries) involvement**

## Catalogue

How stakeholders (*researchers, patient organisations, experts, etc.*) currently (and want to) use RD catalogue of catalogue to answer particular questions

**Needs more stakeholders involvement**

## Registry CDE Linking

to query CDE values across rare disease resources to get answers to questions

**Future call for query tool extension in Q1/Q2 2021**

## Multi-omics analysis

Added value of multi-omics data for diagnosis, disease modifiers (including environmental exposure), identification of drug targets and druggable pathways.

**Next phase for use-cases using registries with mutiomics data to be expected**

# Pillar2 – Involvement in Other Activities

## Domain Specific CDE

🌟 Plans for the creation with JRC a channel dedicated to Domain Specific CDE development (to be confirmed)

🌟 **PPL registry team contribution would be valuable**



We all work for the benefit of rare diseases patients  
& EJP RD is here to support YOU!

# CONTACT US

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# THANK YOU

VASCERN Days  
22–24 of October 2020

