# EUROPEAN JOINT PROGRAMME on RARE DISEASES (EJP RD)

# VASCERN Days 2021

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European Reference Network
for rare or low prevalence complex diseases
Network Vascular Diseases (VASCERN)

EUROPEAN JOINT PROGRAMME RARE DISEASES

08/10/ 2021

# 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

EJP RD

EJP RD

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

# EJP RD in numbers

**+1800** people

**35** participating countries

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

**ALL 24 ERNs**

**101 M€** Budget

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


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

**91 beneficiaries**

**10** hospitals
**12** research institutes
**31** research funding bodies/ ministries
**27** universities/hospital universities
**5** EU infrastructures
**5** charities/foundations
EURORDIS

**+ 52 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 – A glimpse on 30 months work

## Coordination & transversal activities

Qualified coordination team & support

Agile governance & strategy

Sustainability planning from 1st day

Extensive ethics & regulatory support

Perfomant communication & dissemination

## Accelerating of research translation & clinical studies

Innovation Management Toolbox created

19 projects mentored

DataBase of funding opportunities

3 demonstration projects + 2 Innovation projects

Collaboration with EMA established

## Capacity building & empowerment

7 F2F + 9 online courses

500 participants trained

15 ERN workshop financed

33 ERN fellowships attributed

1st Online education MOOC launched


## RD research funding

2 JTCs – 55 M€ - 40 projects

18 Networking events – 487 K€

3 RD Research public-private challenges

78% funded projects involve patient organisations

## Access to data, tools & services

VP building blocks developed & upgraded (incld. Metadata model)

First set of resources linked

Pilot tools to query resources & data discovery in test phase

70 biological pathways created

# EJP RD
# -
# What is there for me?

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# FUNDING, TRAINING & SUPPORT OFFICES

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# Joint Transnational Calls

- **MAIN GOAL:** enable scientists 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

- **MAIN CHARACTERISTICS (some minor modifications may apply every year):**
  - Launched every year in December, pre-announcement in November
  - 2-stage evaluation process (short pre-proposals + invitation to submit full proposal after 1st round of scientific evaluation)
  - Pre-proposal submission stage open for 60 days
  - A minimum of 3 eligible research teams and a max. of 6 per project (can be extended to 8 according to specific conditions)
  - **Involvement of under-represented countries is encouraged**
  - **Involvement of Patient Advocacy Organisations is encouraged** (and can be financed)
    - Guide on Patient Partnership in RD Research (link)
  - Projects are multinational but funding is national (contract is signed by national funding bodies)
  - Typical success rate: 1st stage vs final funding = 10-12%; 2nd stage vs final funding 35 - 50%

**Next call (JTC 2022) will Pre-announced in November 2021**

https://www.ejprarediseases.org/index.php/fundings-and-calls/

*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?*

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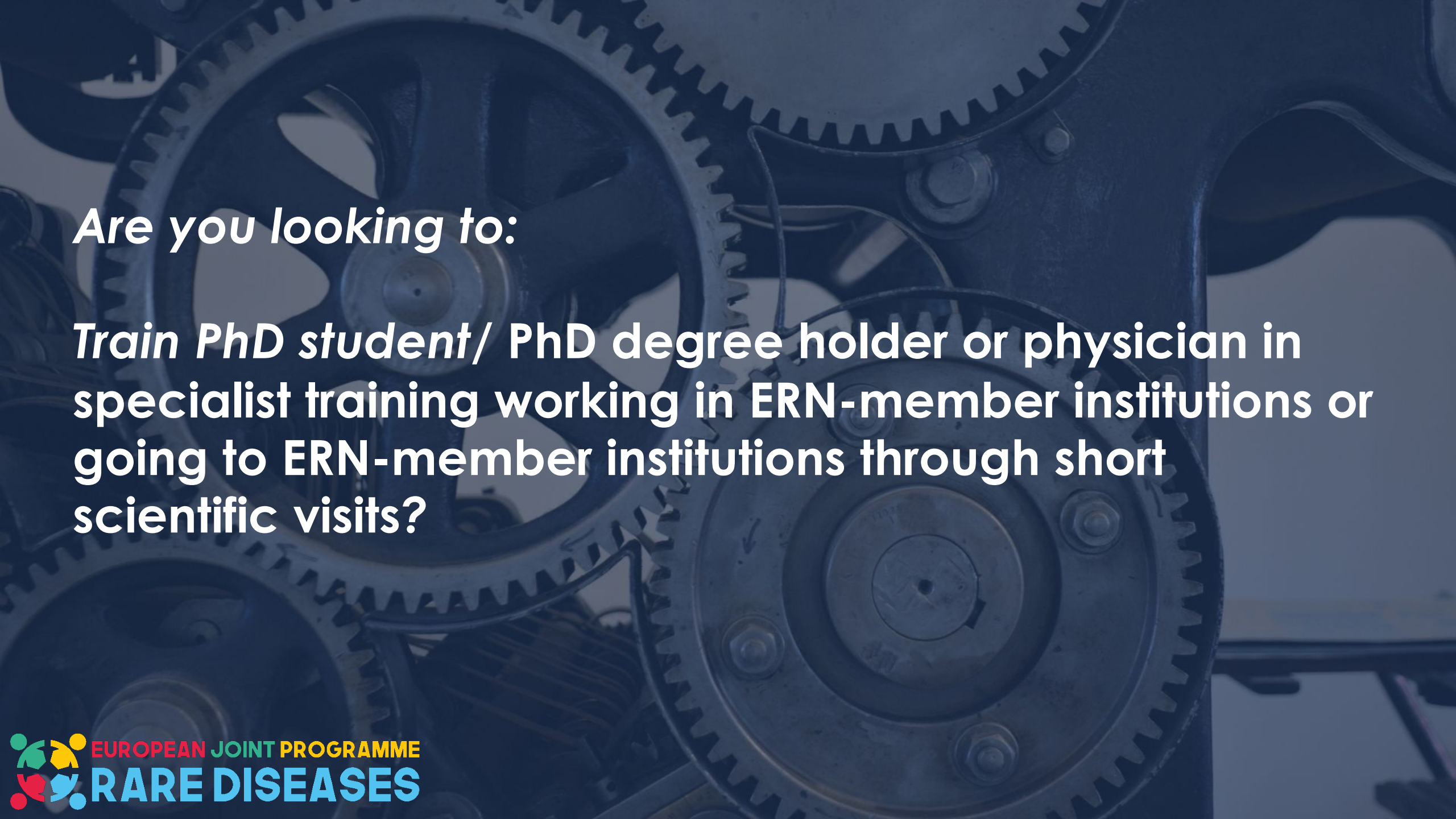
# Networking Support Scheme

- **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.
    - Next collection of application:

| | |
|---|---|
| December 2, 2021 at 14:00 (CET) | March 1, 2022 at 14.00 (CET) |
| September 1, 2022 at 14.00 (CEST) | December 1, 2022 at 14.00 (CET). |

  - **Selected networking events are published on the EJP RD website**. These may be used as **inspiration for building the applying consortium or defining topics for an application**
  - **FAQ document here**
  - 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

https://www.ejprarediseases.org/our-actions-and-services/funding-opportunities/calls/networking-support/



*Are you looking to:*

*Train PhD student/* PhD degree holder or physician in specialist training working in ERN-member institutions or going to ERN-member institutions through short scientific visits?

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# Research Mobility Fellowship

- **Aim:** financially support **PhD students, medical doctors & post-docs working in ERN-member institutions or going to ERN-member institutions** to undertake **short scientific visits** (secondments) up to **6 months** fostering specialist research training outside their countries of residence. Through this training measure the fellows should acquire or share new competences and knowledge related to their research on rare diseases and with benefit to the (home or host) ERN.
- **Applicants/Application profile**:
  - **PhD student** with at least one year of research experience, **PhD degree holder** within five years after doctorate (PhD) or **physician in specialist training** having completed their first year of training and not longer than within five years of completion of specialization
  - Residency in one of the countries beneficiaries of EJP RD
  - Both host and home institution of fellow exchange must be located in one of the countries beneficiaries of EJP RD
  - **Either the home or 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

**Pre-Anouncement for Fellowship: 18th October 2021**
**Deadline for submission: 28th November 2021**

- **Fellows** who were **unable to go to their host institution as well as future fellows will have 18 months from receiving a positive response** from WP17 **to conduct their training** (i.e. Call 2 at the latest by 30.06.2022).

https://www.ejprarediseases.org/our-actions-and-services/funding-opportunities/calls/

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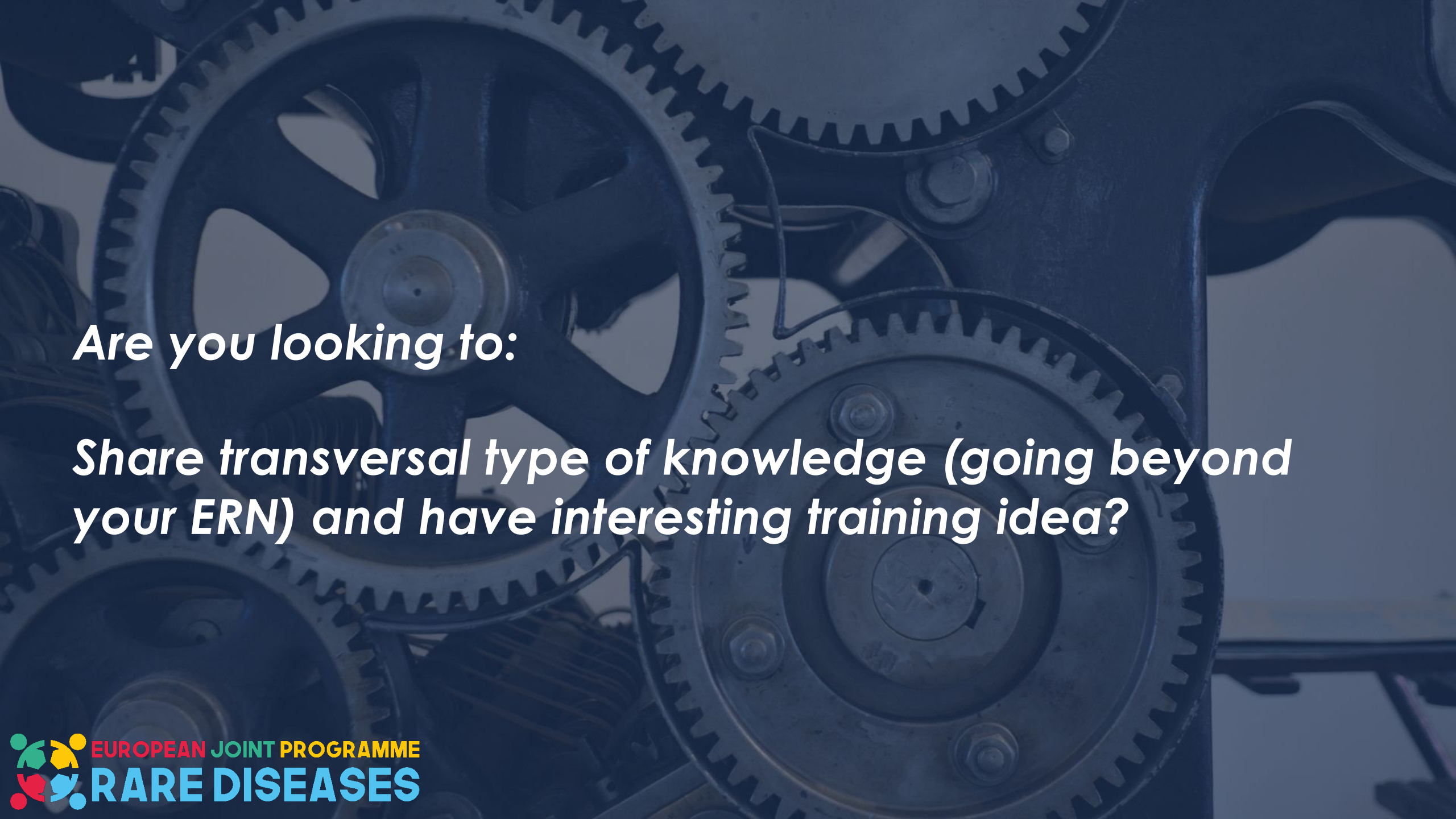

*Are you looking to:*

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

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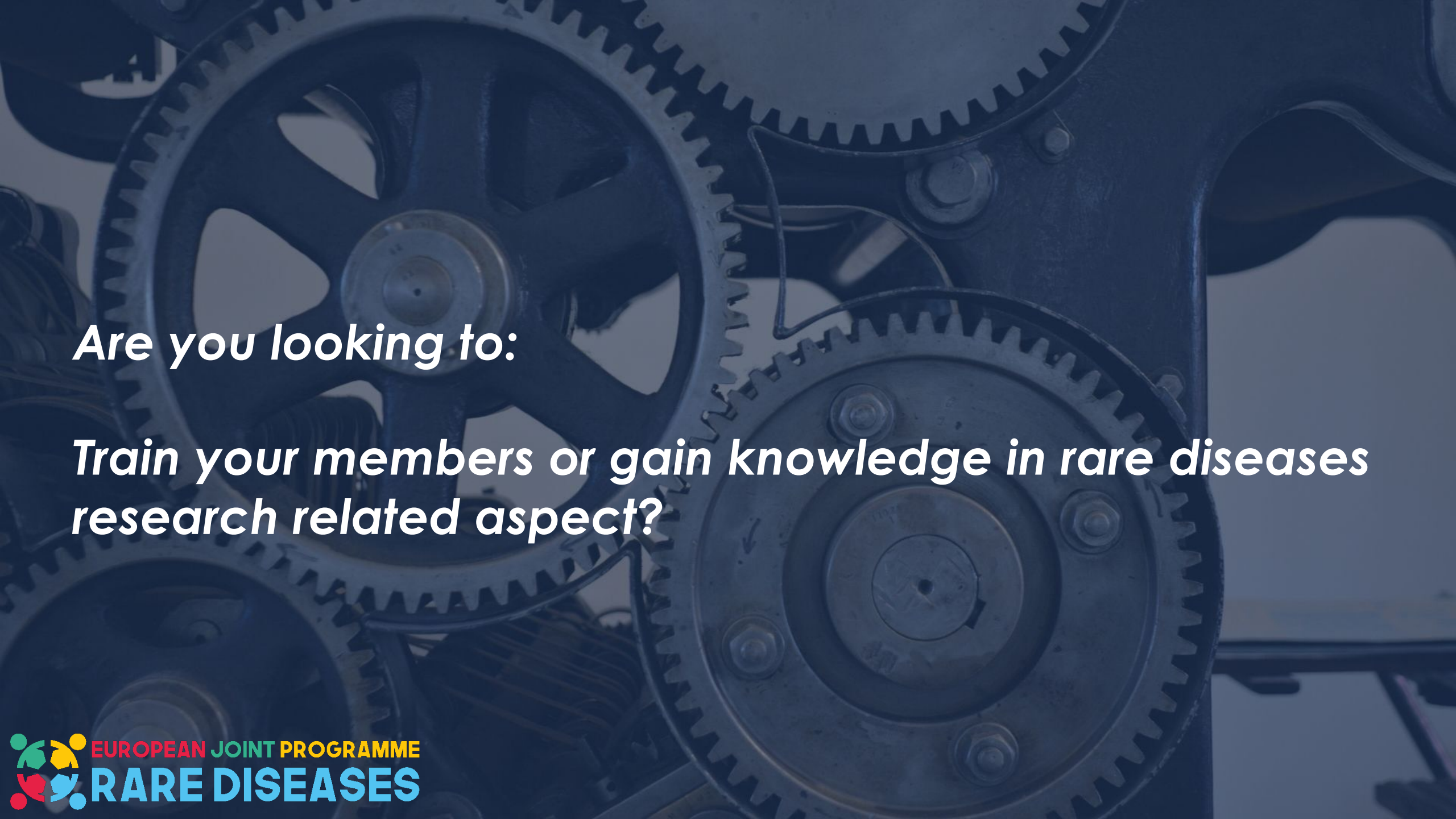
# Research Training Workshop

- **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.
- Topics can be proposed by the ERNs or by investigators belonging to EJP RD beneficiary institutions.
- **25k€ max/event**
- **Applicants/Application profile**:
  - Affiliated to any EJP RD beneficiary institution
  - Affiliated to an ERN Full Member
  - 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
**Next call will open in Q1 2022**

- The selected workshops in Call 2 have to be conducted by latest: 31/05.2022
- And the ones selected in Call 3 have to be conducted by latest 31/12/2022

https://www.ejprarediseases.org/our-actions-and-services/training-and-education/ern-workshops/



*Are you looking to:*

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

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# 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*.
  - **Next available event: RD Clinical Trials Randomisation / Online (registration open very soon)**
  - **Further upcoming events:**
    - **course on Undiagnosed Cases _ 11-13 April 2022**
    - **Summer School on RD Registries and FAIRification of data _ 26-30 September 2022**
    - **NGS _ 19-21 October 2022**

https://www.ejprarediseases.org/our-actions-and-services/training-and-education/rd-research-trainings/

- **Patients & Researchers training:** training modules dedicated to **building the capacity of the patient community and other key stakeholders**, including
  - training "expert" paediatric patients on rare diseases – Q1 2022,
  - medicines development,
  - Scientific Innovation and Translational Research,
  - Leadership and Communication Skills

https://www.ejprarediseases.org/our-actions-and-services/training-and-education/patients-training/

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# Other EJP RD trainings

- **Online Academic Education course:** on transversal and multidisciplinary aspects of rare diseases research are made available for all stakeholders in a fully online format. The course foresees the development of Different modules.
  - **MOOC #1Diagnosing Rare Diseases: from the Clinic to Research and back.**
    - **Run2 Registration still open: https://www.futurelearn.com/courses/rare-genetic-disease.**
    - Run3  Spring 2022 - Run 4 Autumn 2022
  - MOOC#2 **- Innovative personalized medicine - Sept. 2022**
  - MOOC#3 - **Translational Research - March/April 2022**
  - MOOC#4 - **Statistical methodologies for clinical trials – end 2022/early 2023**
  - MOOC#5 - **Data & Ethics - end 2022/early 2023**

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*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*

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# 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: https://orphandrugguide.org)
- Connection with Pillar 2 to make it interoperable with the Virtual Platform

**Beta-Test of the Innovation Management Toolbox Mid-November 2021**

**To get involved as a tester (prime access), contact** coordination@ejprarediseases.org

## 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**

**Access to the service:**

**https://eatris.eu/services/expert-mentoring-service-rare-disease-researchers/**

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*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*

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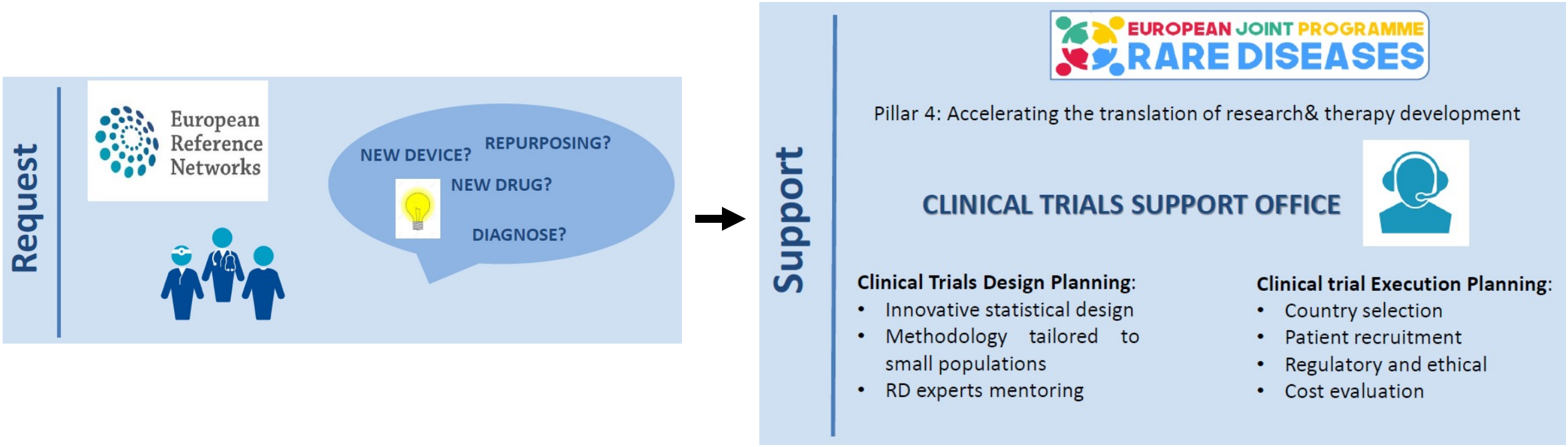
# Multinational CTs support office




**Access to through EJP RD Helpdesk:**
**https://www.ejprarediseases.org/our-actions-and-services/central-helpdesk/**

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# VIRTUAL PLATFORM & RELATED SERVICES

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# Virtual Platform (VP)

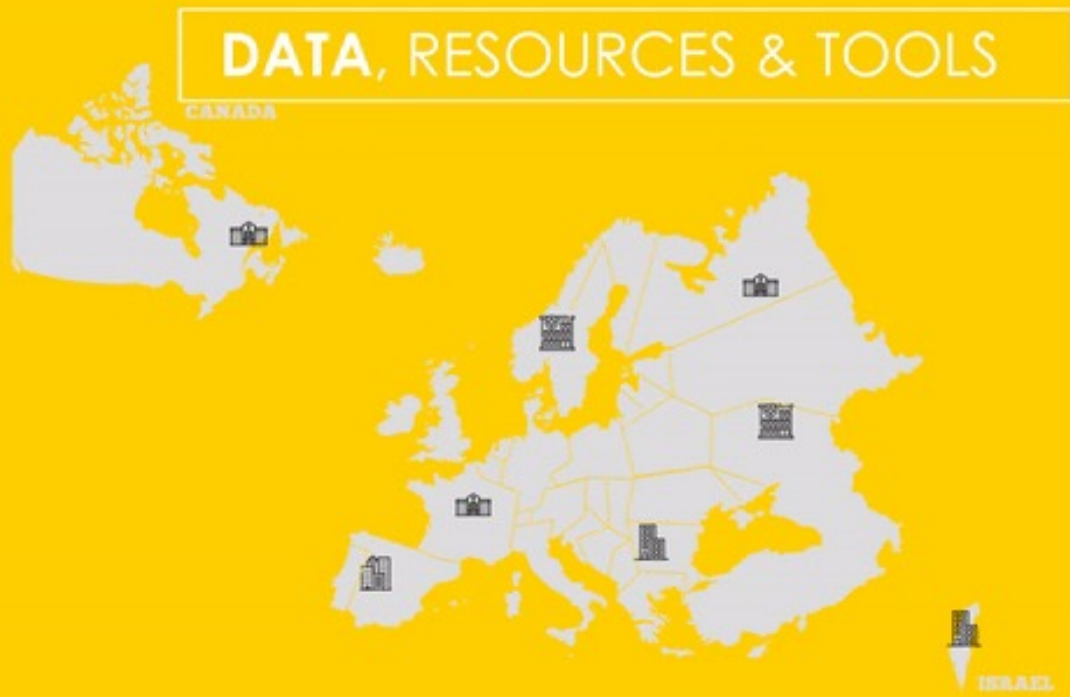
Federated

Standardized

GDPR-compliant

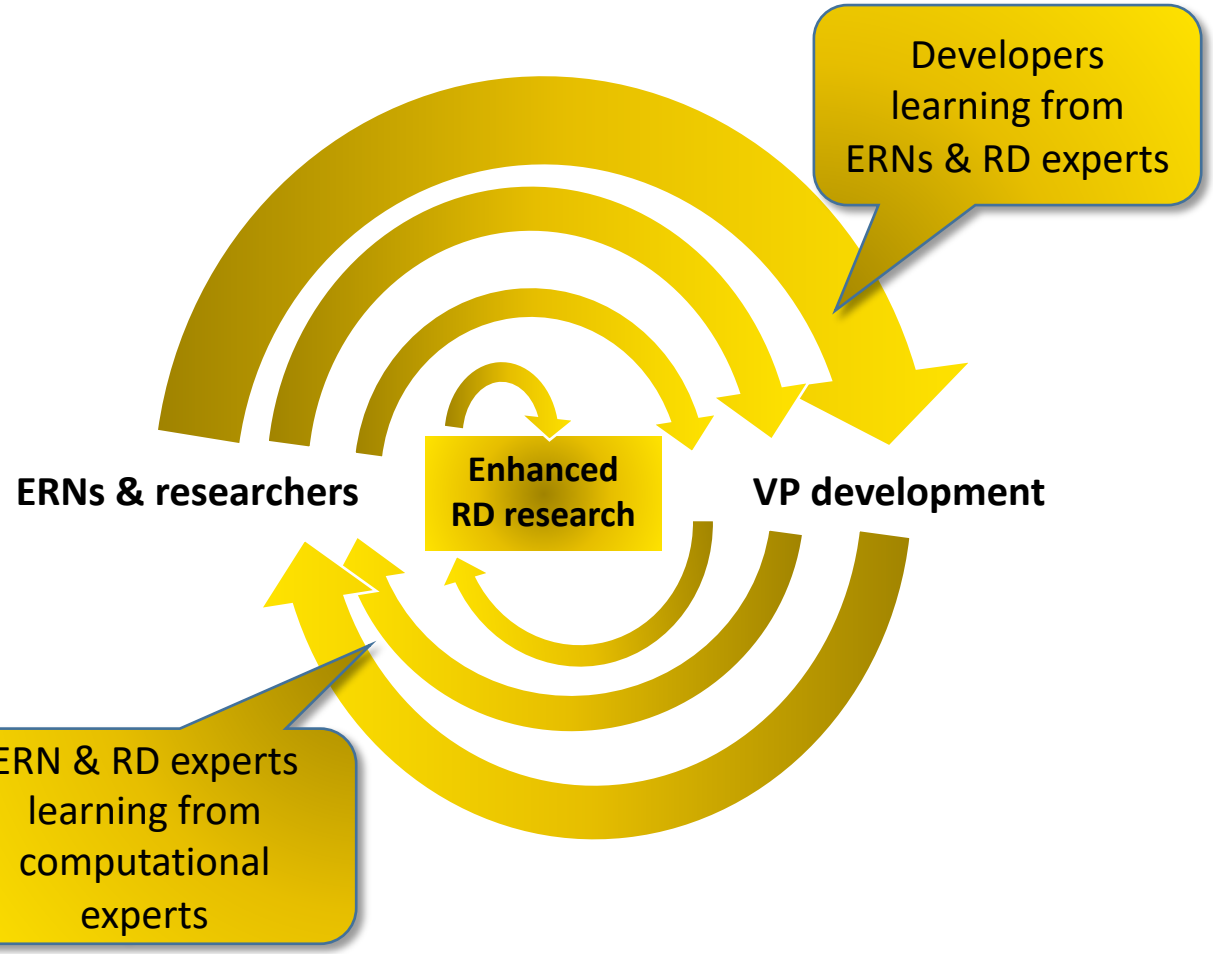
Sustainable

Quality assessed




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

# Strategy




**Use cases as a starting point**

Activation → Identify Champions → Identify users → Use case description → Minimum Viable Product → VP component

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# 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**

# Finding resources via EJP RD

New tool to search for current available resources related to Rare Diseases. A simple interface to easily discover IRDiRC recognized and EJP RD funded resources & tools.

PATIENT REGISTRIES AND COHORTS

BIOBANKS

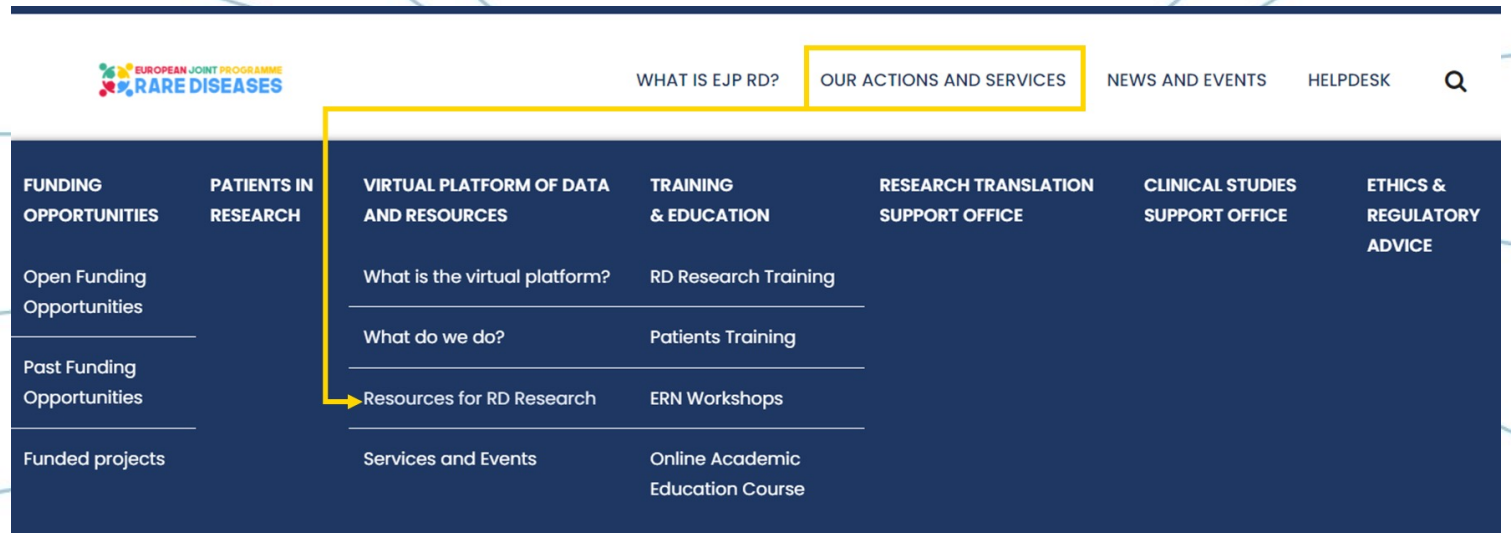
ANIMAL MODELS AND CELL LINES

ARCHIVE/SHARE EXPERIMENTAL AND PHENOTYPIC HUMAN DATA

ANALYSE EXPERIMENTAL AND PHENOTYPIC HUMAN DATA

RD CLINICAL EXPERTS

https://resourcemap.ejprarediseases.org/




STANDARDS

IRDIRC RECOGNIZED GUIDELINES

TOOLS FOR RESEARCH

KNOWLEDGE BASES

TOOLS & SERVICES FOR TRANSLATIONAL AND CLINICAL

# Workshops & Webinars Material

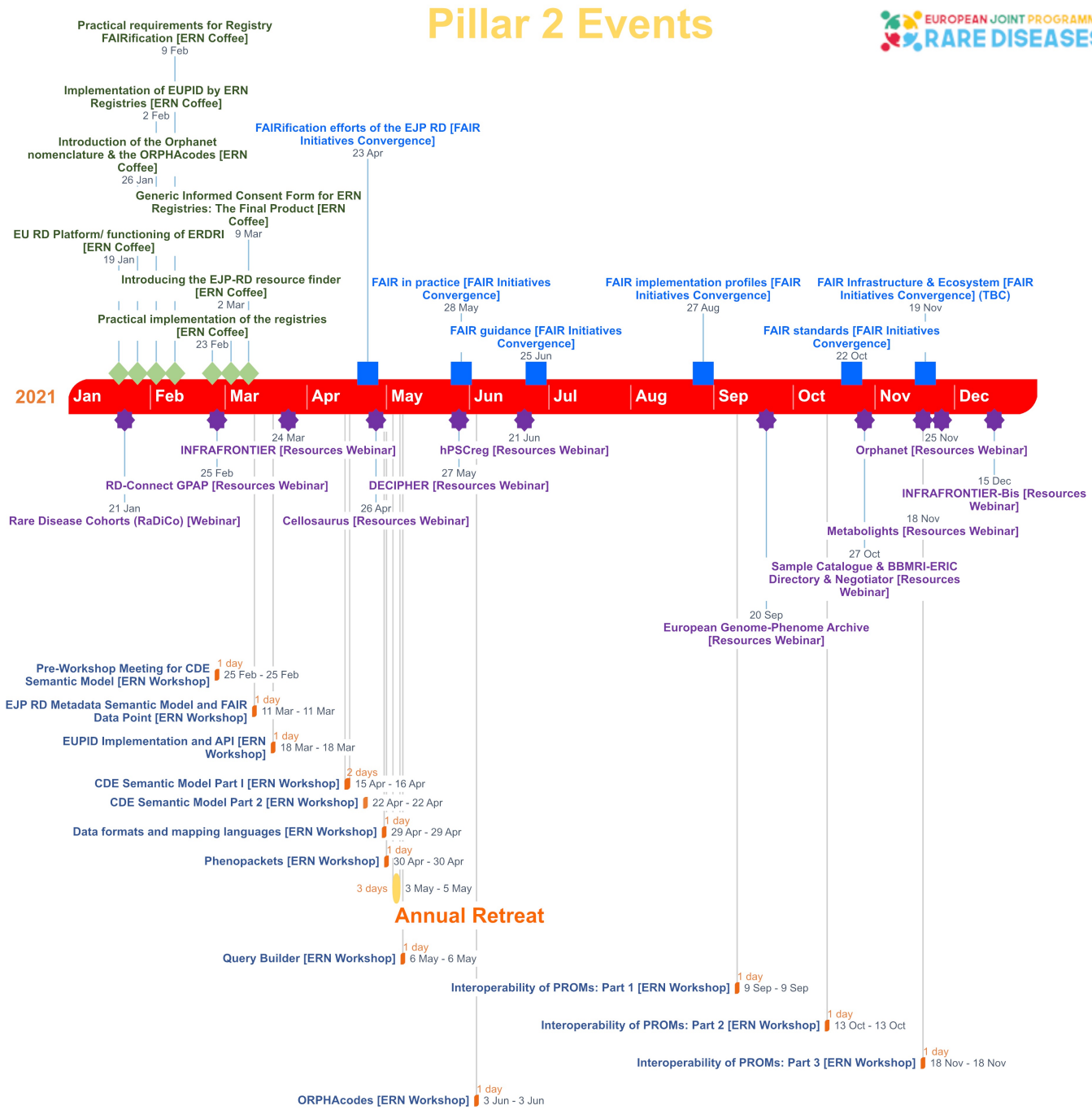
Video recording and presentation slides of past webinars

+

description & connection links to upcoming ones

available on dedicated **SharePoint sites**

[Timeline link (subject to updates)]



# EJP RD Resources Webinars

- Monthly webinars to demo the Virtual Platform related resources with aims to introduce:
  - What is this resource?
  - How can this resource be helpful for clinicians / researchers involved in rare disease studies?
  - How can this resource be used by the audience?

**SharePoint site:**

**https://ejprd.sharepoint.com/sites/EJPRD-Resources-Webinar**

- If issue for access, email b42f2456.ejprd-project.eu@fr.teams.ms to request access to this SharePoint site and to the ERNs team (please indicate the name of your ERN as well)

**Next webinars** (always 3-4pm):

27/10/2021 Sample Catalogue & BBMRI-ERIC Directory & Negotiator

18/11/2021 Metabolights

25/11/2021 Orphanet

15/12/2021 Infrafrontier - bis

# EJP RD ERN Events

- Webinars and workshops to address ERN needs
  - Non-technical Webinars
  - ERN technical hands-on Workshops (Primary target audience is ERN Registry Developers/ Programmers or data stewards for the practical aspects of the FAIRification tasks)

## SharePoint site:

**https://ejprd.sharepoint.com/sites/EJPRD-ERN-EVENTS**

- If issue for access, email b42f2456.ejprd-project.eu@fr.teams.ms to request access to this SharePoint site and to the ERNs team (please indicate the name of your ERN as well)

## Next workshops:

13/10/2021_10:00-12:00 CET

- **Interoperability of PROMs in the context of ERNs:** Join the conversation! How can we make the PROMs for ERNs FAIR?

18/11/2021 _10:00-11:00 CET

- Interoperability of PROMs in the context of ERNs: **Proposed solution and implementation plan**

# FAIR Initiatives Convergence

- A series of workshops organised with external experts to identify convergence in FAIRification procedures and guidance developed in other projects.

**SharePoint site:**

**https://ejprd.sharepoint.com/sites/FAIR-Initiatives-Convergence**

- If issue for access, email b42f2456.ejprd-project.eu@fr.teams.ms to request access to this SharePoint site and to the ERNs team (please indicate the name of your ERN as well)

**Next webinars** (always on Friday 4-5pm):

22/10/2021 FAIR standards

TBC FAIR Infrastructure & Ecosystem

# Harmonising patient consent

**Common informed consent forms**
(ICF) for RD registries

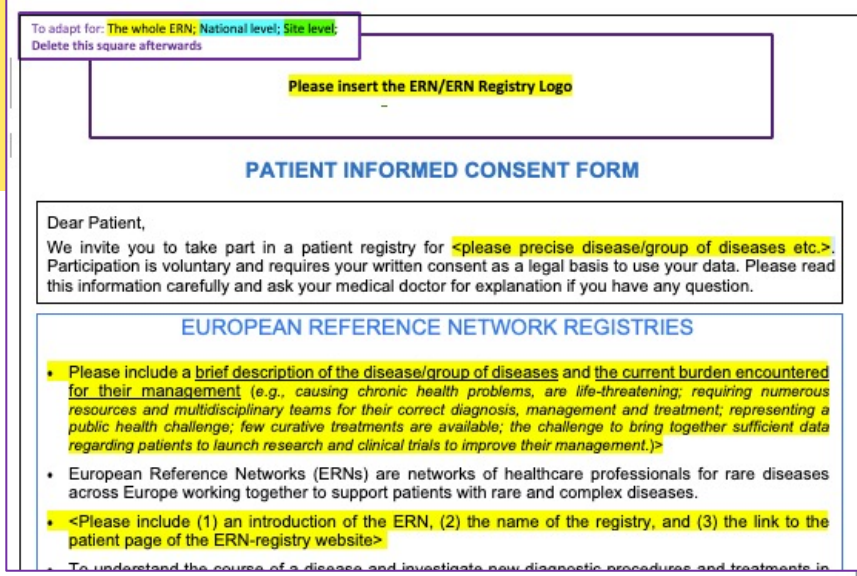
**Adaptation at: ERN level; National level; Site level**

Using **standards & ontologies**
to make it **machine-readable**

- Adult version link
- Parents version link
- Folder link for translated versions*

*[Accessible through EJP RD SharePoint for EJP RD Partners]*

*into Dutch, Greek, Hebrew, Italian, Lithuanian, Norwegian, Polish, Slovak, Spanish, Portuguese, Estonian, Latvian, Danish, Finnish, French, Swedish, Turkish, Arabic, Romanian, Bulgarian, German, Slovenian, Hungarian, Czech and Croatian


To adapt for: The whole ERN; National level; Site level;
Delete this square afterwards

Please insert the ERN/ERN Registry Logo

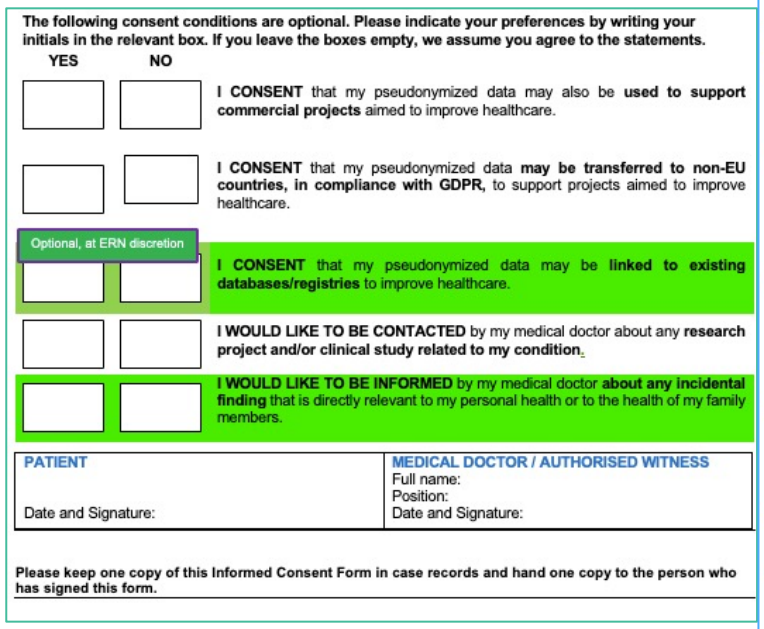
PATIENT INFORMED CONSENT FORM

Dear Patient,

We invite you to take part in a patient registry for <please precise disease/group of diseases etc.>. Participation is voluntary and requires your written consent as a legal basis to use your data. Please read this information carefully and ask your medical doctor for explanation if you have any question.

EUROPEAN REFERENCE NETWORK REGISTRIES

- Please include a brief description of the disease/group of diseases and the current burden encountered for their management (*e.g., causing chronic health problems, are life-threatening; requiring numerous resources and multidisciplinary teams for their correct diagnosis, management and treatment; representing a public health challenge; few curative treatments are available; the challenge to bring together sufficient data regarding patients to launch research and clinical trials to improve their management.*)>
- European Reference Networks (ERNs) are networks of healthcare professionals for rare diseases across Europe working together to support patients with rare and complex diseases.
- <Please include (1) an introduction of the ERN, (2) the name of the registry, and (3) the link to the patient page of the ERN-registry website>


**The following consent conditions are optional. Please indicate your preferences by writing your initials in the relevant box. If you leave the boxes empty, we assume you agree to the statements.**

YES NO

**I CONSENT** that my pseudonymized data may also be **used to support commercial projects** aimed to improve healthcare.

**I CONSENT** that my pseudonymized data **may be transferred to non-EU countries, in compliance with GDPR,** to support projects aimed to improve healthcare.

Optional, at ERN discretion

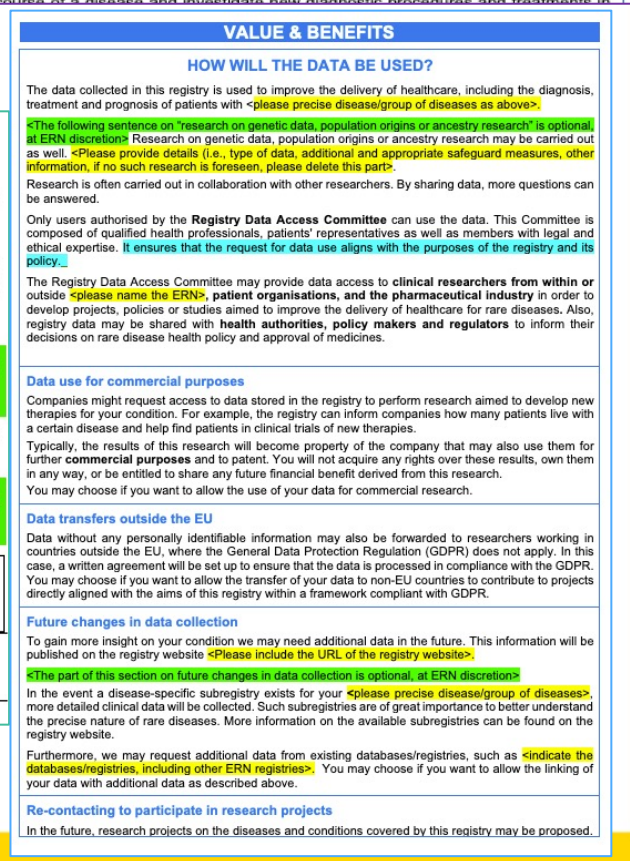
**I CONSENT** that my pseudonymized data may be **linked to existing databases/registries** to improve healthcare.

**I WOULD LIKE TO BE CONTACTED** by my medical doctor about any **research project and/or clinical study related to my condition.**

**I WOULD LIKE TO BE INFORMED** by my medical doctor **about any incidental finding** that is directly relevant to my personal health or to the health of my family members.

| PATIENT | MEDICAL DOCTOR / AUTHORISED WITNESS |
|---|---|
| Date and Signature: | Full name:<br>Position:<br>Date and Signature: |

**Please keep one copy of this Informed Consent Form in case records and hand one copy to the person who has signed this form.**


**VALUE & BENEFITS**

**HOW WILL THE DATA BE USED?**

The data collected in this registry is used to improve the delivery of healthcare, including the diagnosis, treatment and prognosis of patients with <please precise disease/group of diseases as above>.

<The following sentence on "research on genetic data, population origins or ancestry research" is optional, at ERN discretion> Research on genetic data, population origins or ancestry research may be carried out as well. <Please provide details (i.e., type of data, additional and appropriate safeguard measures, other information, if no such research is foreseen, please delete this part>.

Research is often carried out in collaboration with other researchers. By sharing data, more questions can be answered.

Only users authorised by the **Registry Data Access Committee** can use the data. This Committee is composed of qualified health professionals, patients' representatives as well as members with legal and ethical expertise. It ensures that the request for data use aligns with the purposes of the registry and its policy.

The Registry Data Access Committee may provide data access to **clinical researchers from within or** outside <please name the ERN>, **patient organisations, and the pharmaceutical industry** in order to develop projects, policies or studies aimed to improve the delivery of healthcare for rare diseases. Also, registry data may be shared with **health authorities, policy makers and regulators** to inform their decisions on rare disease health policy and approval of medicines.

**Data use for commercial purposes**

Companies might request access to data stored in the registry to perform research aimed to develop new therapies for your condition. For example, the registry can inform companies how many patients live with a certain disease and help find patients in clinical trials of new therapies.

Typically, the results of this research will become property of the company that may also use them for further **commercial purposes** and to patent. You will not acquire any rights over these results, own them in any way, or be entitled to share any future financial benefit derived from this research.

You may choose if you want to allow the use of your data for commercial research.

**Data transfers outside the EU**

Data without any personally identifiable information may also be forwarded to researchers working in countries outside the EU, where the General Data Protection Regulation (GDPR) does not apply. In this case, a written agreement will be set up to ensure that the data is processed in compliance with the GDPR. You may choose if you want to allow the transfer of your data to non-EU countries to contribute to projects directly aligned with the aims of this registry within a framework compliant with GDPR.

**Future changes in data collection**

To gain more insight on your condition we may need additional data in the future. This information will be published on the registry website <Please include the URL of the registry website>.

<The part of this section on future changes in data collection is optional, at ERN discretion>

In the event a disease-specific subregistry exists for your <please precise disease/group of diseases>, more detailed clinical data will be collected. Such subregistries are of great importance to better understand the precise nature of rare diseases. More information on the available subregistries can be found on the registry website.

Furthermore, we may request additional data from existing databases/registries, such as <indicate the databases/registries, including other ERN registries>. You may choose if you want to allow the linking of your data with additional data as described above.

**Re-contacting to participate in research projects**

In the future, research projects on the diseases and conditions covered by this registry may be proposed.

# Common Conditions of use Elements (CCEs)

| Category | Common Condition Elements |
|---|---|
| **Who** | Use by a commercial entity |
| **Where** | Geographical area<br>Jurisdiction |
| **When** | Time period of use |
| **Why** | Research use<br>Clinical use<br>Disease specific use<br>Use as control<br>Profit motivated use |
| **How** | Collaboration<br>Fees<br>Return of non-incidental results<br>Return of incidental findings<br>Re-identification of individuals |

## CCEs are

- **Generic** - Cover common issues
- **Atomic in nature** - Handle only **one** concept
- **Non-Directional** - No indication of permission

## CCEs do NOT

- Replace Ethics/Governance processes
- Encode complete consent forms
- Provide detailed encodings for automated decisions on access

**First use case being explored is Resource Discovery**

**Test out the creation of 'profiles' of consent and use conditions that apply to some asset or resource at:**

- Leicester (https://duc.le.ac.uk)

or

- Groningen (for Molgenis Platform) (https://irdirc.molgeniscloud.org/menu/main/home)

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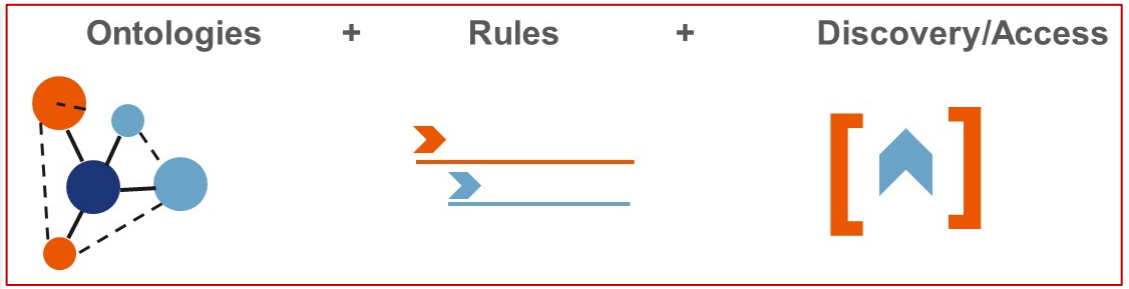
# Standardising models for access conditions

**Improved ontologies**, plus core set of terms relevant to the VP ('CCE')

**Validated data structure** for digital representation of consent and use conditions in the VP

**Framework semantic models**
Ontological representation of access conditions (consent, license) in FAIR metadata to support automatic assessment of dynamic data access decisions




**"Digital Use Conditions" (DUC) data structure**

**Integration as metadata model extension for resources discoverability (WIP)**

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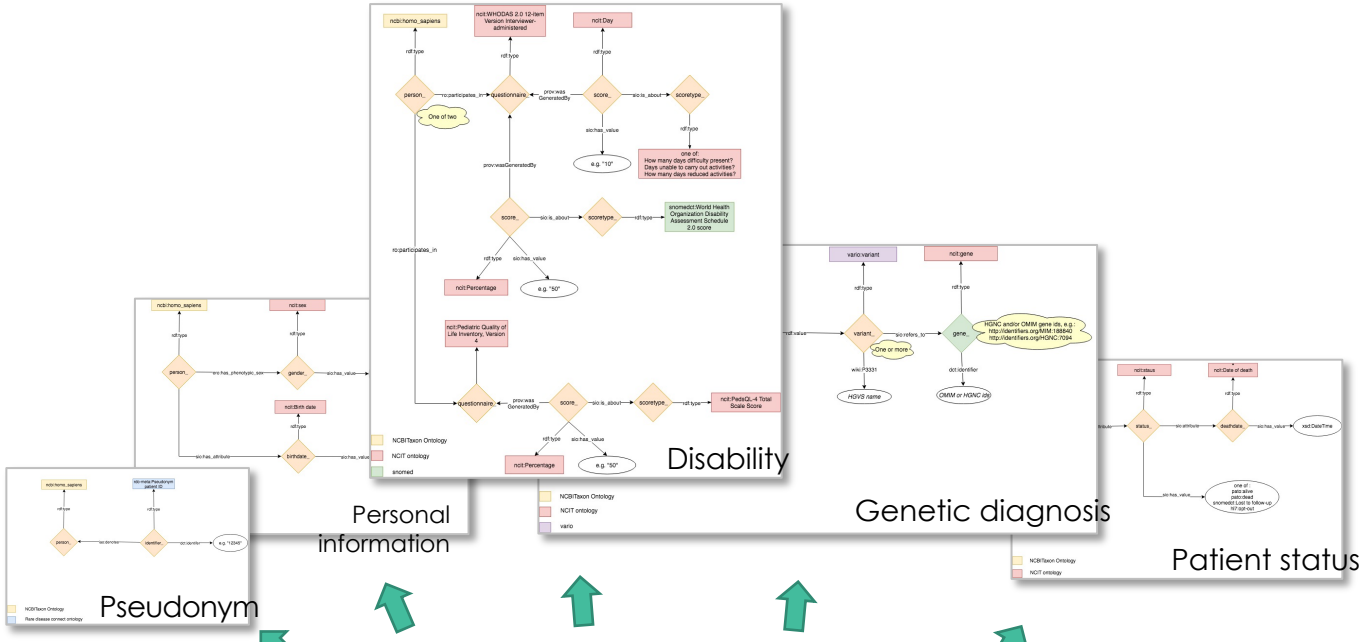
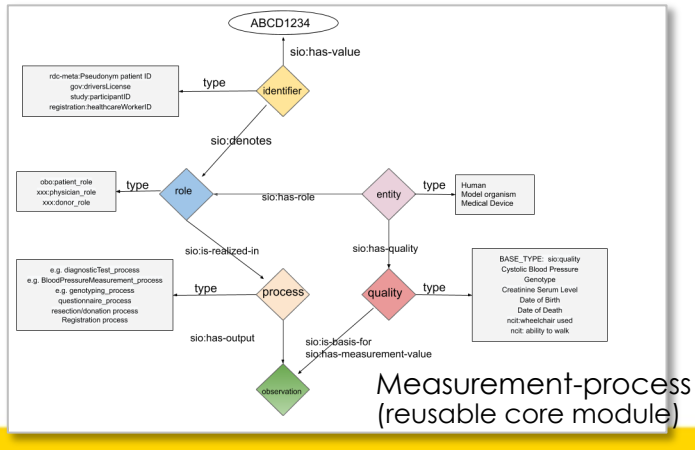
# Ontological model to expose shared understanding of what data elements mean in a rare disease resource




<new data element>

- **For resources to declare their content for machines**
  → VP adapts function to its sources
- Reusable core module (design pattern), extendable, queryable
  → **Asset for other health data projects**
- Based on global standard ontologies
  → globally interoperable, AI ready
- Mappings and bridging solutions
  (SSSOM & grlc for GA4GH schemas, FHIR, OMOP, C-DISC, OBO Foundry)
- Compatible with DCAT2-based EJP RD metadata model and FAIR data points
- Interacts with services that host reference models and mappings (e.g. FAIRsharing.org)

https://github.com/ejp-rd-vp/CDE-semantic-model
https://fairsharing.org/FAIRsharing.xVAQX9
**« CDE-in-a-box »** https://github.com/ejp-rd-vp/cde-in-box

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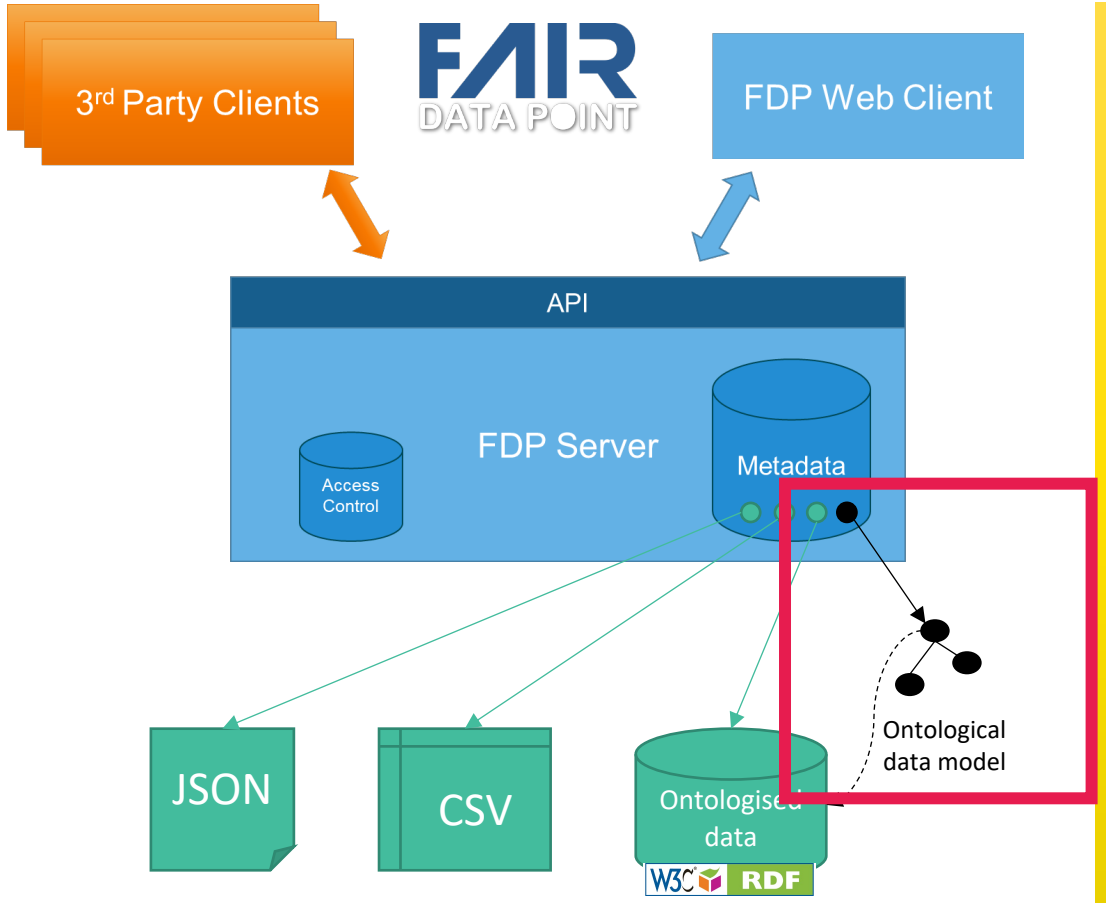
# Access to metadata: FAIR data point

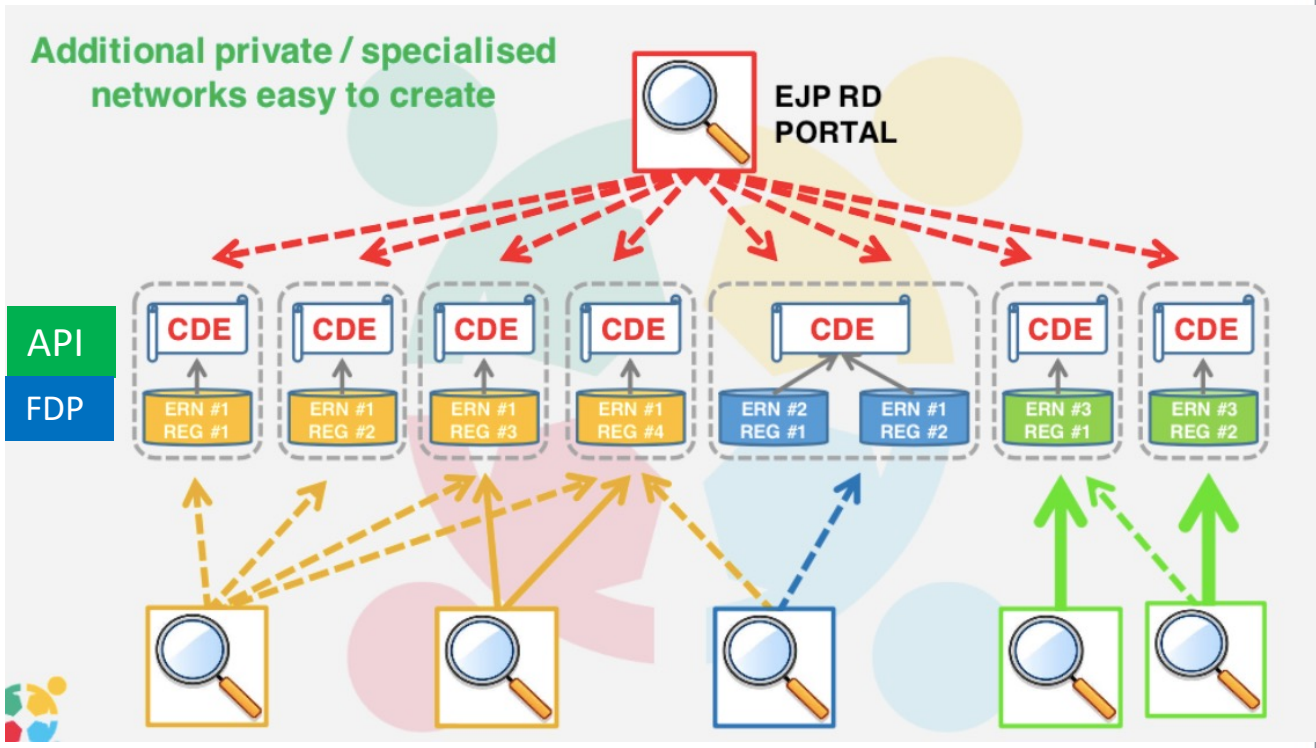



- **A metadata repository**
  - **Machine actionable information** about a resource:
    - The repository itself;
    - The type of resource such as catalogue, registry, dataset, software, ontologies, etc.;
    - Pointers to data sets in one or more formats (distributions)
    - Access conditions
  - Machine actionable by EJP RD metadata model (=extended DCAT2)
  - Access by REST API (follows DCAT2)
- Indexed in one or more **metadata indexes** (also FAIR data points)
- **Complements Query APIs** by seamless bridging between GA4GH API & SPARQL

**Specification:** https://github.com/FAIRDataTeam/FAIRDataPoint-Spec
**Reference code:** https://github.com/FAIRDataTeam/FAIRDataPoint
**Deployment documentation:** https://fairdatapoint.readthedocs.io/
**Licence:** MIT

# Creating technical interoperability (FDPs & APIs)

- **Application Programming Interface (API)** based on GA4GH Beacon-2 plus, leveraging EJP-RD Common Data Elements, and Common Consent Element models
- **Network management service** that holds and distributes the addresses of the participating resources
- **Discovery services** (including central query portal(s)) for safe record-level querying of a wide array of registries and biobanks
- **Complements metadata repository** (FAIR data point) **& ontologically linked data** by seamless bridging between GA4GH API & SPARQL




**QB implementation API**
https://github.com/ejp-rd-vp/query_builder_api

**FAIR data point specs**
https://github.com/FAIRDataTeam/FAIRDataPoint-Spec

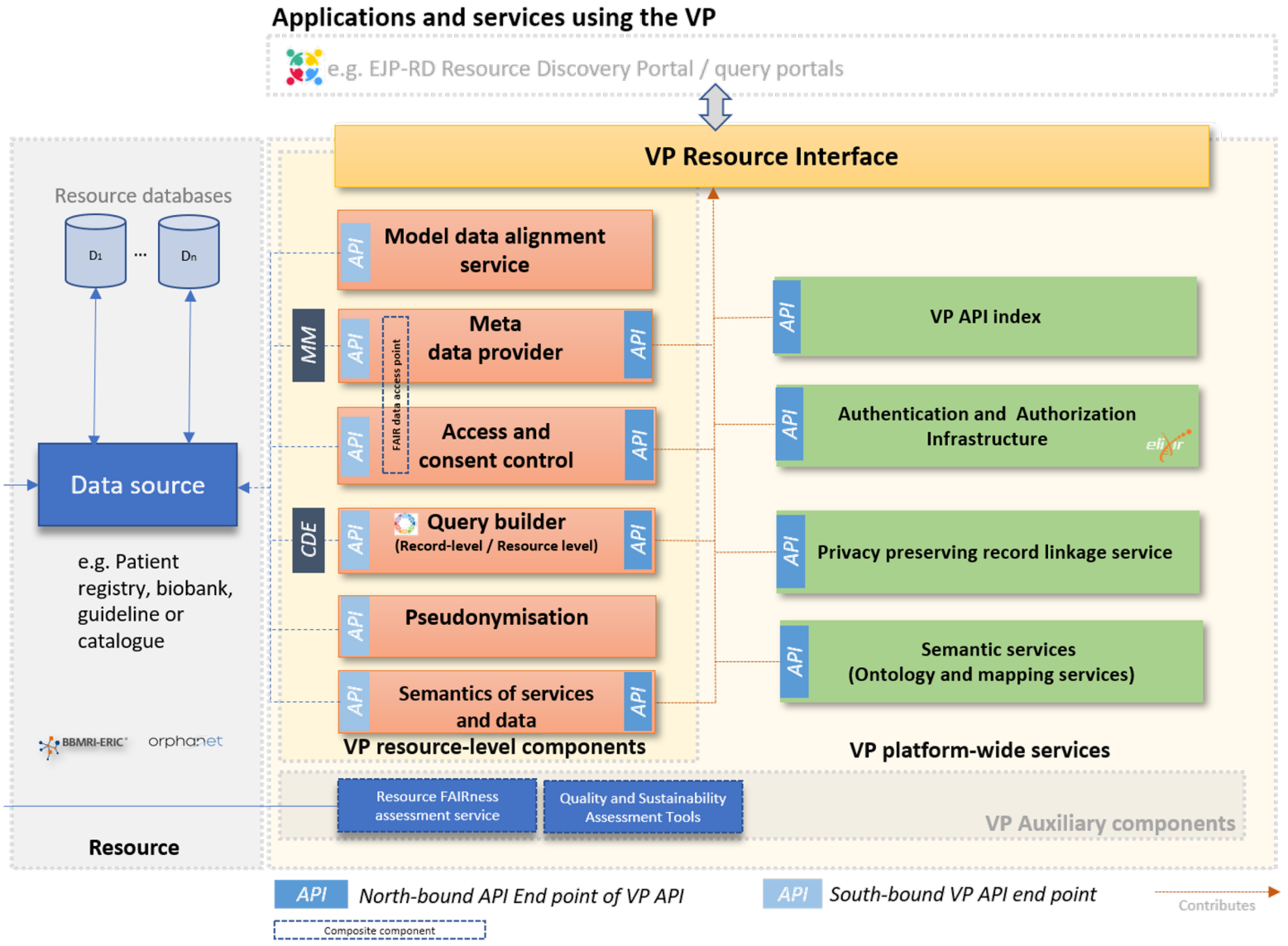
# Virtual Platform specifications

**First version to be disseminated today !**

**describes the first version of the virtual platform, laying out architectural principles, identifying stakeholders, core components and standards.**

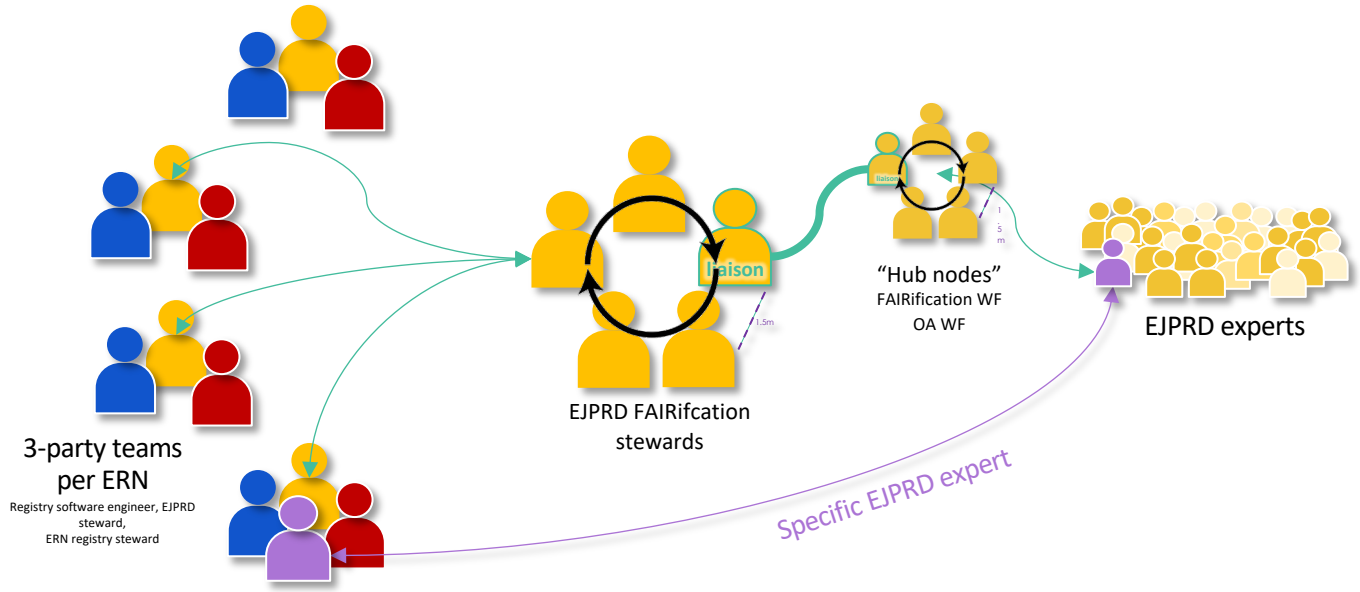
This document is an alpha version, meaning that components may change, new components may be added or removed in upcoming versions that extend or modify the core functionalities of the virtual platform.

**Will also be available in the [EJP RD Deliverables Folder]**



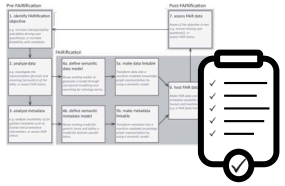
# Steward support for adopting VP specifications & FAIR principles

1. Documented FAIRification **procedures and experiences**
   First implementations within ERN registry software published; workshop on FAIRifying pathways by uploading to WikiPathways (WP13)
2. FAIRification **guidance** aligned with external projects
3. **Toolkits** for data stewards (compilation of guides, software tools, data models)
4. FAIRification **steward service** / project management support
5. FAIR+VP-enabled **registries**
6. **Demonstrate** benefits of FAIR-based analysis

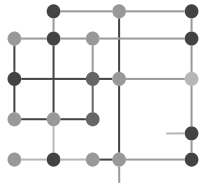




Assess standards & tools


Guidelines & metrics


Organise expertise

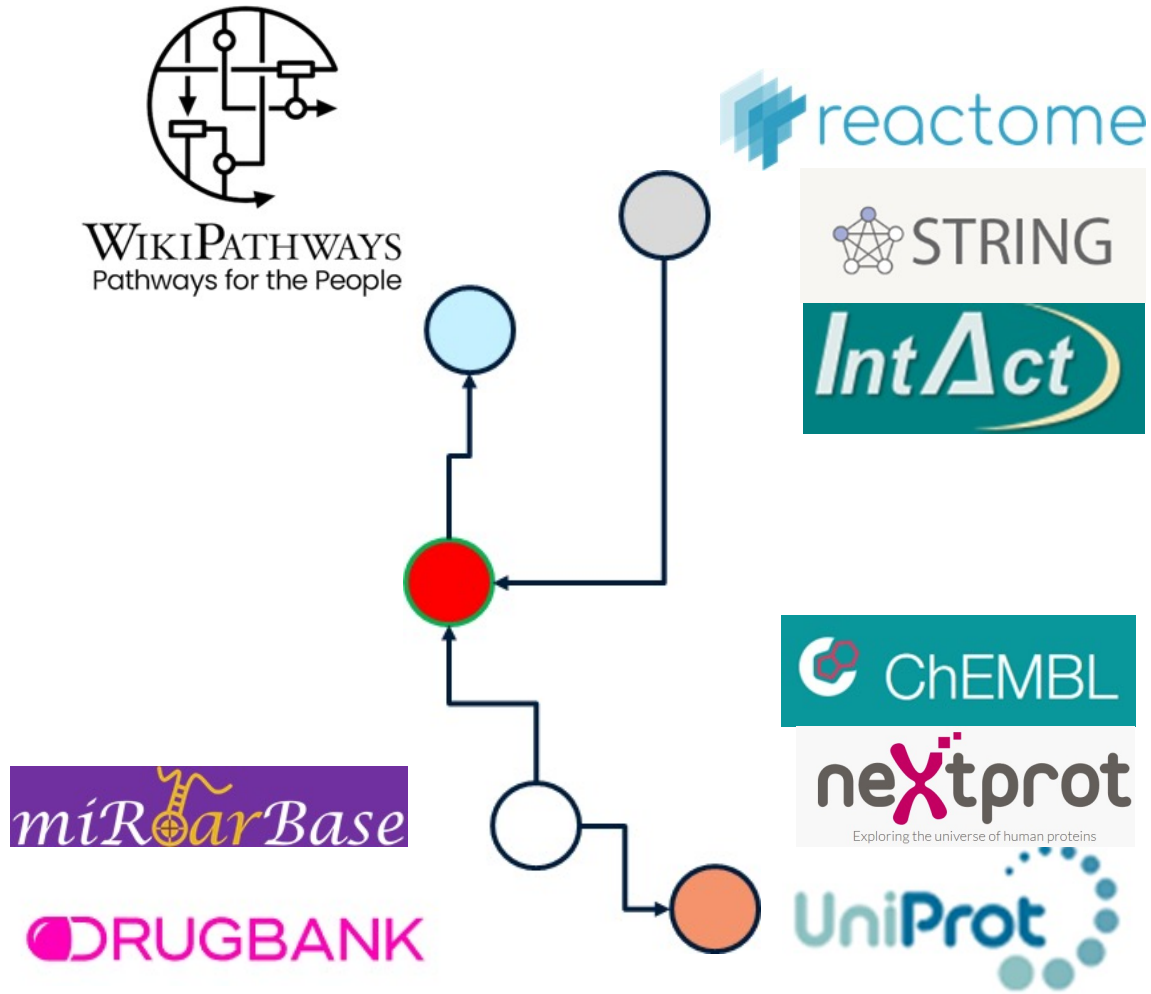

Align with infrastructure


Support training (P3)

https://www.ejprarediseases.org/services-and-events/fairification-stewardship-programme-for-ern-registries/

# Building cross-omics pathways and disease networks

- Systems biology integration of
  - **Knowledge** = pathway models, known interactions
  - **Experimental data** = single omic & multiomics data
  - **Additional information** = e.g. Environmental interplay (Nutrition, drugs), phenotypical information
- to build **disease networks** for understanding and identification of potential drug targets




[http://raredisease.wikipathways.org](http://raredisease.wikipathways.org)

- Holding 70 RD pathways
- Inborn Errors of Metabolism

(Blau et al., 2014, Springer)

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[WikiPathways Academy (link)](): Organized training examples for new WikiPathway Authors

# MultiOmics data analysis – Call for collaboration

- 3 Use cases done with ERNs – **we are looking for 2 more to go!**

- **What we can offer**
  - Thorough (re)analysis of your omics data, eventually drawing new conclusions
  - Help with a specific research question that require the integration of a special resource

- **Intake criteria**
  - Availability of a concrete research question in disease mechanisms, progression and modifiers
  - Availability of data from at least **two different -omics technology** from the same samples.
  - A group size of at least 20 cases and controls (if applicable)
  - Availability of rich phenotype descriptions, descriptions of sample collection and preservation
  - A standardized way of collecting and measuring samples
  - Permission to share the data among the EJP RD WP13 members
  - Commitment to jointly publish the results
  - Commitment to join in monthly meetings to discuss results and give feedback on the analysis process and to join at least one face-to-face workshop

Contact: Peter-Bram.tHoen@radboudumc.nl

EJP RD

# 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

European Reference Network for rare or low prevalence complex diseases

Network Vascular Diseases (VASCERN)