European Joint Programme on Rare Diseases (EJP RD)

Yanis Mimouni - INSERM

EJP RD coordination team

EUROPEAN JOINT PROGRAMME RARE DISEASES

CHIST-ERA Workshop on Open Science in Transnational Research

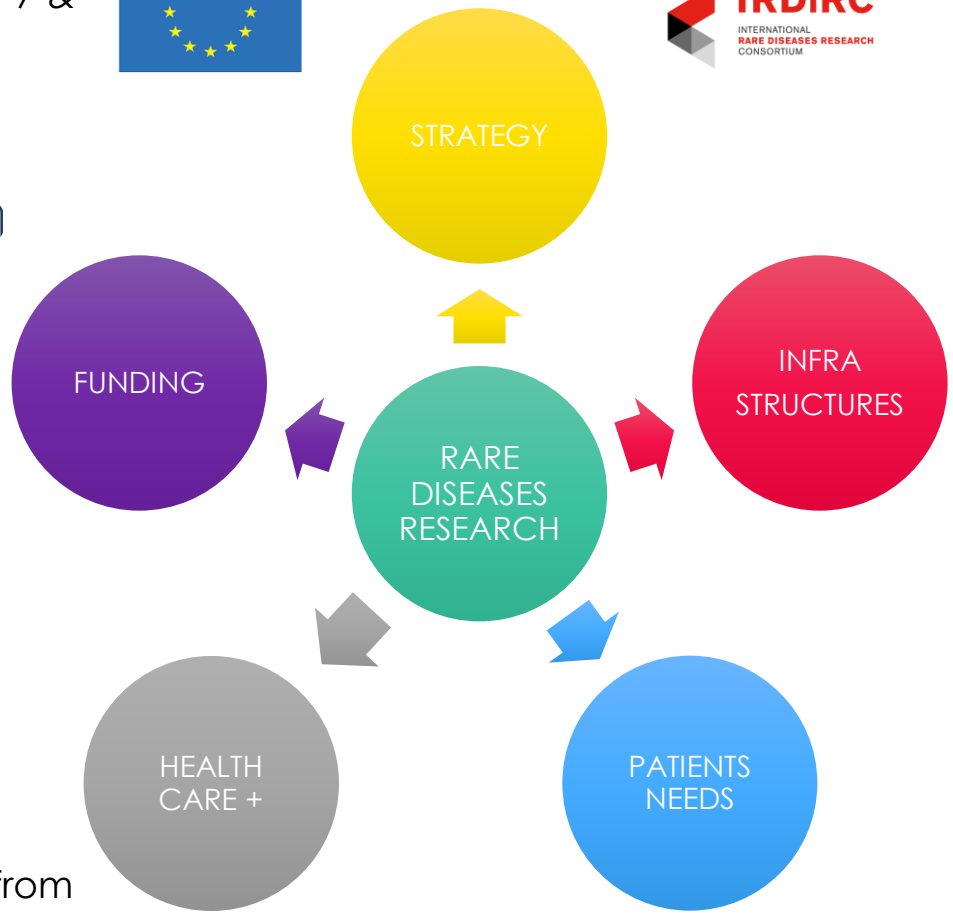
Bern, Switzerland

06/03/2020

Rare Diseases Landscape in Europe

1.4 billion € for RD research in FP7 & Horizon 2020

25 funders from 17 countries
Over 120 M€ invested in RD research



More than 900 units from 300 hospitals covering 26 countries

869 patient organisations from 76 countries

EUROPEAN JOINT PROGRAMME ON RARE DISEASES

EUROPEAN JOINT PROGRAMME
RARE DISEASES

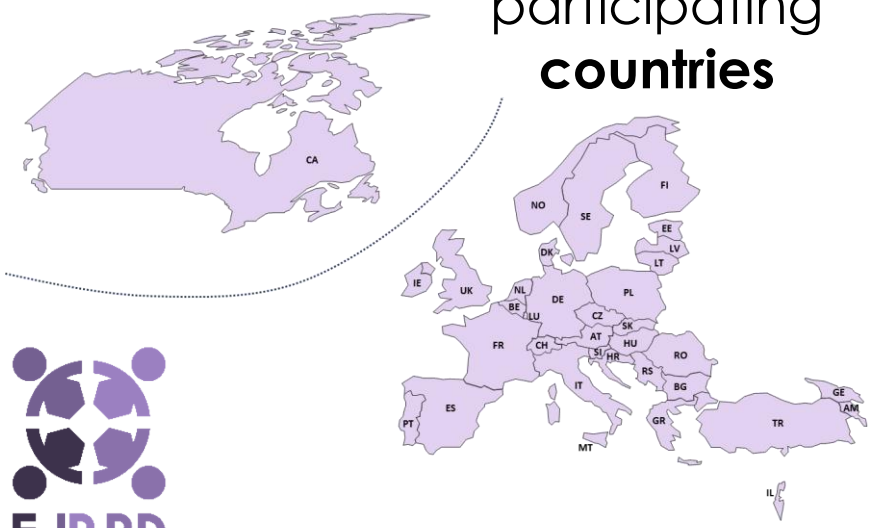
Jan 2019 → **Dec 2023**

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

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

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

35
participating
countries



88 Beneficiaries:

10 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 the associated networks

24 ERNs:
300 institutions
>950 healthcare units
26 countries

EURORDIS:
884 RD patient organisations
72 countries

ECRIN
12 main national nodes

EATRIS
13 main national nodes

ELIXIR
220 research organisation
23 partners

INFRAFRONTIER
23 partners
15 countries

BBMRI
1 international partner
21 main national nodes
20 countries

EJP RD

Funded by the European Union
GA n°825575

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

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

OPEN SCIENCE "ORIENTED"

EJP RD

Funded by the European Union
GA n°825575

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, 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

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

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

Pillar 3: Capacity building & empowerment

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 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

EFFORTS TOWARD OPEN SCIENCE POLICY

EUROPEAN JOINT PROGRAMME
RARE DISEASES

Under H2020

- **Open Access to all peer-reviewed scientific publications relating to EJP RD results: an obligation**
  - **Public Deliverables** (available on EJP RD website after EC validation)

- **Open Access to research data** - the right to access and reuse digital research data **under the terms and conditions set out in the Grant Agreement**.

EJP RD

Funded by the European Union
GA n°825575

EJP RD Grant Agreement _ open science considerations

DISSEMINATION OF RESULTS — OPEN ACCESS — VISIBILITY OF EU FUNDING

- Each beneficiary must - **as soon as possible** - '**disseminate**' its **results** by **disclosing them to the public** by appropriate means (other than those resulting from protecting or exploiting the results), including in scientific publications (in any medium). Unless it goes against their legitimate interests
  - **Open access to scientific publications**
  - **Open access to research data**

Funded by the European Union
GA n°825575

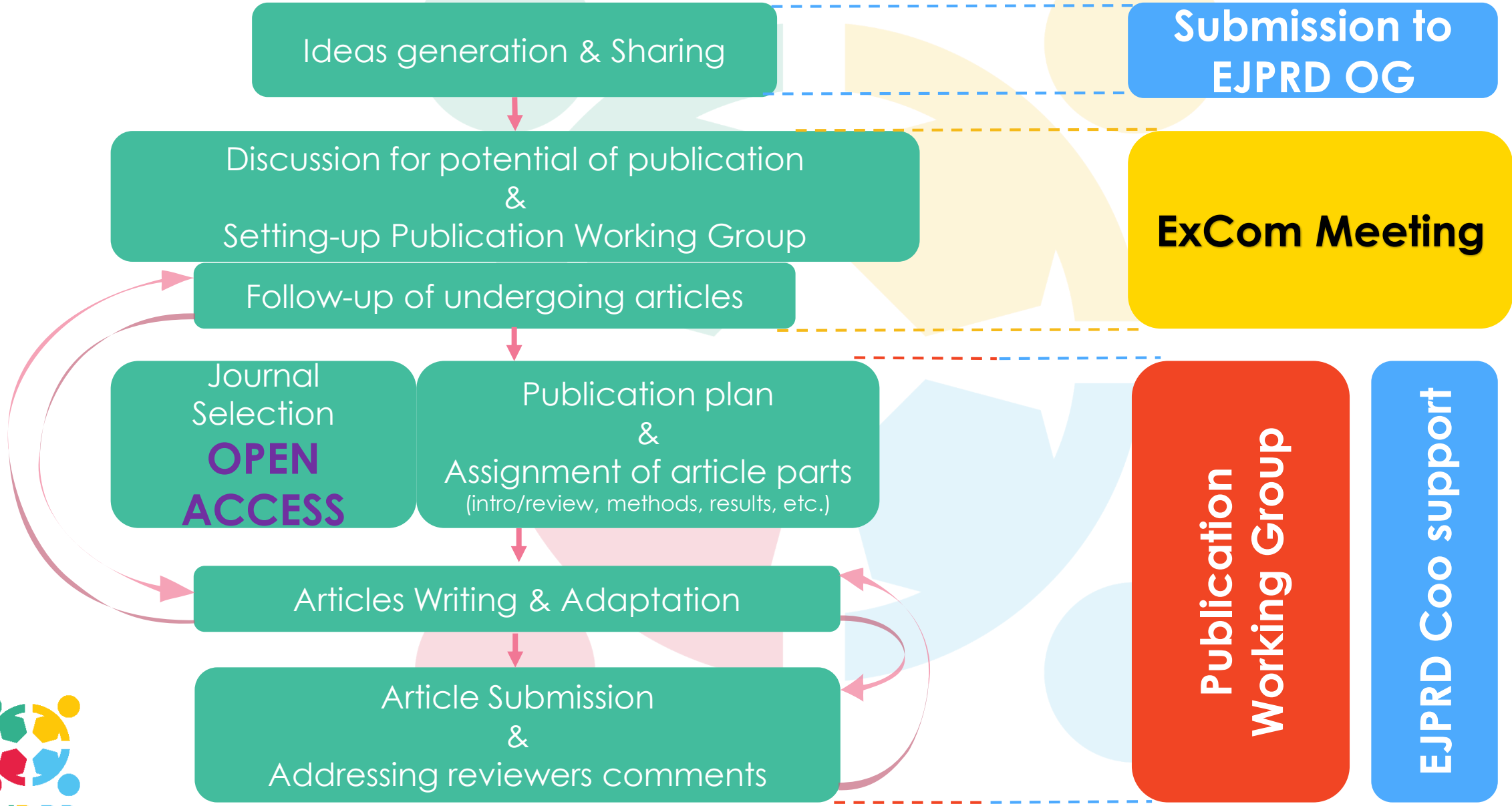
Open access to Scientific publications

- Results obtained :
  - **after the start of the EJP RD** (January 2019)

    AND
  - **Multi-partner publication** (at least two partners from the EJP RD in the authors' list)
- **Inform** the EJP RD Coordination
  - **when the paper is under preparation**
  - of **the costs "OPEN ACCESS" of the publication**:
    - EJP RD Coordination will include those costs in the Communication budget
- **Acknowledgment :**
  - *"X.X (person initials) work is supported by the funding from the European Union's Horizon 2020 research and innovation programme under the EJP RD COFUND-EJP N° 825575"*

Funded by the European Union GA n°825575

Overarching publications (Policy, white papers, scientific with large scope, etc.)



Funded by the European Union GA n°825575

EJP RD Grant Agreement _ open science considerations

Open access to research data

The beneficiaries must:

a) **deposit in a research data repository** and take measures to make it possible for third parties **to access, mine, exploit, reproduce and disseminate - free of charge for any user -** the following:

   (i) **data**, including **associated metadata**, **needed to validate the results presented in scientific publications**, as soon as possible;

   (ii) **other data**, including associated metadata, **as specified** and within the deadlines laid down **in the 'data management plan'**;

b) **provide information** — via the repository — **about tools and instruments at the disposal of the beneficiaries and necessary for validating the results** (and - where possible - provide the tools and instruments themselves)

Funded by the European Union
GA n°825575

Funded Projects (Pillar 1 and Pillar 4)

- Joint Transnational Calls for collaborative research projects
- Networking Support Scheme Call to share knowledge on rare diseases
- Rare Diseases Research (RDR) challenges Call
- Demonstration projects internal call on existing statistical methodologies to improve RD clinical trials

Comply to

Open Access to Research Data

Required in the call text

EJP RD

Funded by the European Union
GA n°825575

EJP RD Pillar 2 Agreement

Purpose:

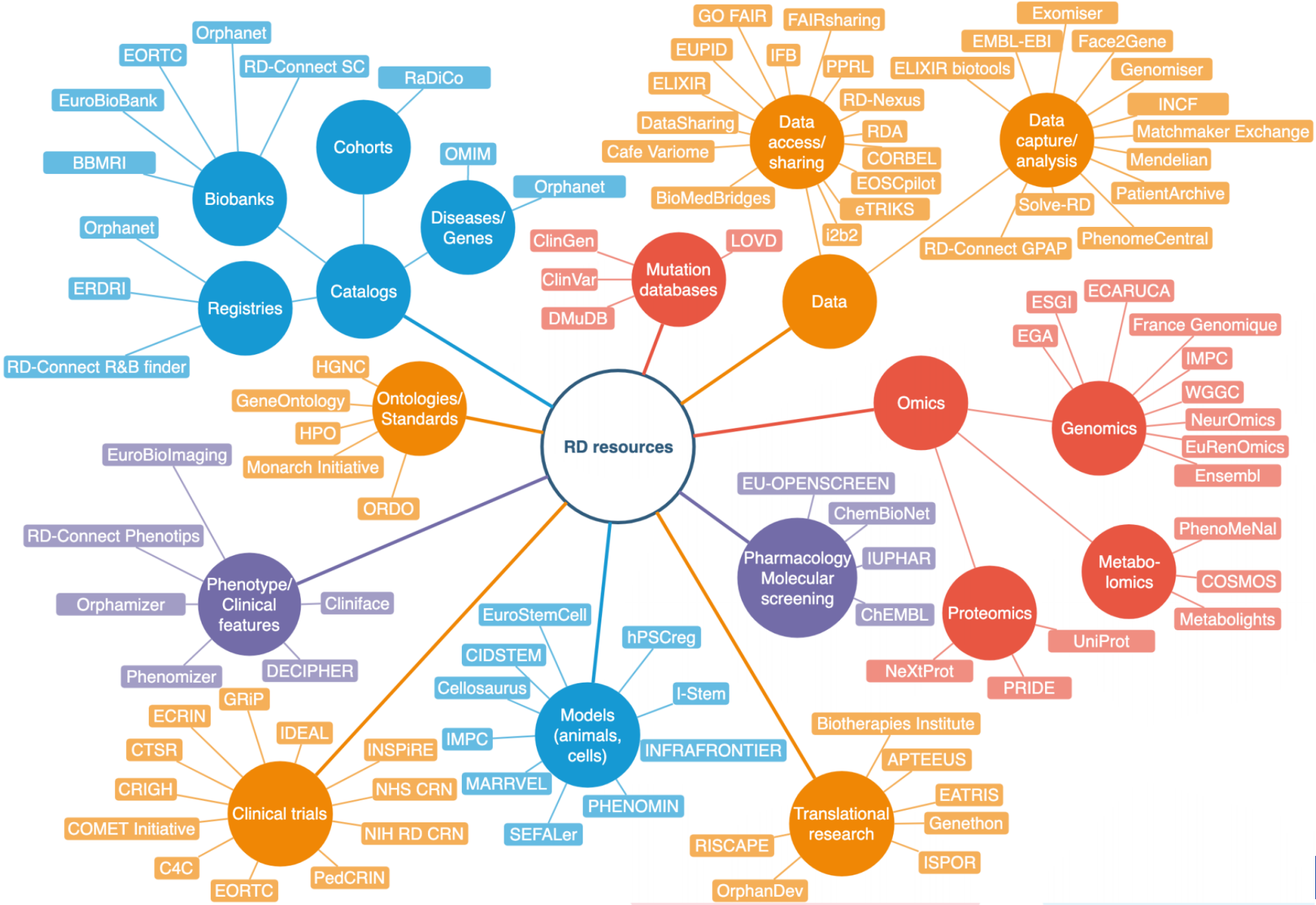
- **To specify**, with respect to the Pillar 2 Work Programme within the EJP RD Action, **the relationship among the *Pillar 2 Parties***, in particular regarding the organisation of the work between the *Pillar 2 Parties*:
  - Service Supplier;
  - and/or Data Supplier;
  - and/or Tools Supplier;
  - and/or Developers/ Innovators;
  - and/or End-Users providing user Expertise/ knowledge

EJP RD

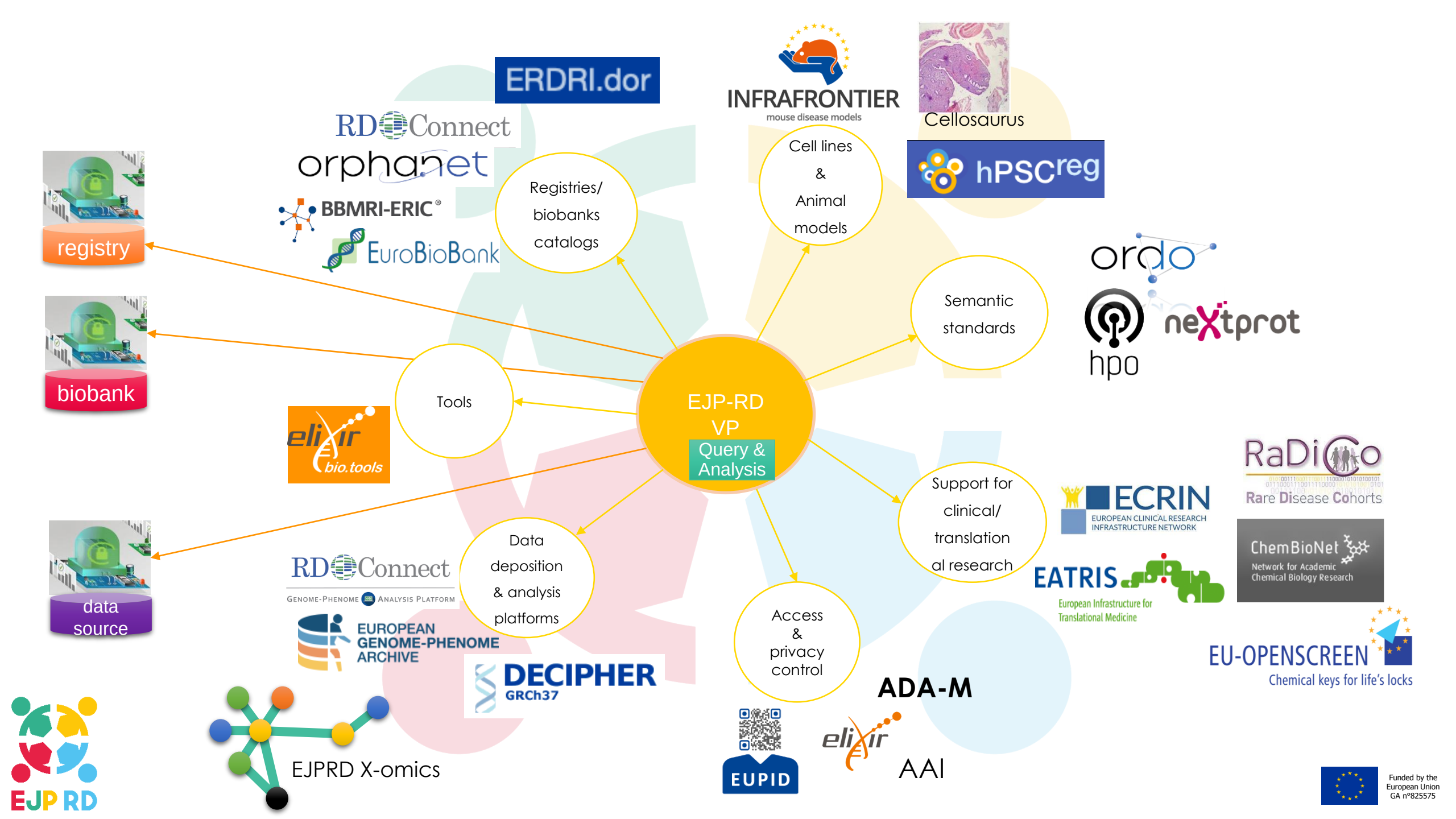
Funded by the European Union
GA n°825575

Deliverable 10.5 "Report on the State of the Art of existing resources in Europe"

Fig2. **RD research resources landscape**



Funded by the European Union
GA n°825575


ERDRI.dor
RD Connect
orphanet
BBMRI-ERIC
EuroBioBank
INFRAFRONTIER
mouse disease models
Cellosaurus
hPSCreg
registry
biobank
data source
Registries/ biobanks catalogs
Cell lines & Animal models
Semantic standards
Tools
EJP-RD VP
Query & Analysis
Data deposition & analysis platforms
Access & privacy control
Support for clinical/ translational research
ordo
neXtprot
hpo
elixir bio.tools
RD Connect
Genome-Phenome Analysis Platform
EUROPEAN GENOME-PHENOME ARCHIVE
DECIPHER
GRCh37
ECRIN
EUROPEAN CLINICAL RESEARCH INFRASTRUCTURE NETWORK
RaDiCo
Rare Disease Cohorts
ChemBioNet
Network for Academic Chemical Biology Research
EATRIS
European Infrastructure for Translational Medicine
EU-OPENSCREEN
Chemical keys for life's locks
EJP RD
EJPRD X-omics
EUPID
ADA-M
elixir
AAI
Funded by the European Union
GA n°825575

Pillar 2 Agreement _ Open science considerations

Responsibilities of Beneficiaries

- **Any Developing Party** undertakes to carry out its developments **taking into account the compatibility issues between Software Licences to prevent contamination** between Software.
- If a Developing Party wishes to **use an Open Source Licence**, in the Pillar 2, it must **notify all the others Developing Parties**.
  - A contaminating Software or contaminating Licence means **Open Source Licence whose Licence is required for Software derived from it**. For example
    - the GPL and CeCILL version A Licences are called contaminating Licences.
    - The LGPL, and CeCILL version B and version C Licences are not contaminating.

EJP RD

Funded by the European Union
GA n°825575

Pillar 2 Agreement _ Open science considerations

Specific provisions for Open Source Software

- In order to **enable the Parties to determine the effects of using an Open Source Software** relating to the Use or Exploitation of Results and to express any disagreement regarding the use of Open Source Software, **the Party who wishes to use it**, within the framework of the Project, must **provide to** the **Data and Software Committee**, on express request from another Party, **all necessary information** relating to the Open Source Software which is applicable to it.
- The Data and Software Committee, shall be **responsible for verifying that this request does not affect the objectives of the Project and/or the interests of a Party**

EJP RD

Funded by the European Union
GA n°825575

Pillar 2 Agreement _ Open science considerations _ Background

Background (Grant Agreement definition)

- Data, know-how or information - whatever its form or nature (tangible or intangible), including any rights such as intellectual property rights - that:
  a) is held by the beneficiaries before they acceded to the Agreement, and
  b) is needed to implement the action or exploit the results

- Because the background is needed, Access Rights have to be granted in principle, but Parties must identify and agree amongst them on the Background for the project →

EJP RD

Funded by the European Union
GA n°825575

Pillar 2 Agreement _ Open science considerations _ Background

- Option 1: The following background is hereby identified and agreed upon for the Project. Specific limitations and/or conditions, shall be as mentioned hereunder:

Describe Background	**Specific limitations and/or conditions for implementation** (Article 25.2 Grant Agreement)	**Specific limitations and/or conditions for Exploitation** (Article 25.3 Grant Agreement)

- Option 2: No data, know-how or information of **"beneficiary"** shall be Needed by another Party for implementation of the Project or Exploitation of that other Party's Results

Access Conditions Defined

Funded by the European Union
GA n°825575

Pillar 2 Work Foci _ Promoting data access through VP

- **FAIRification WF**: allowing data sources to become progressively FAIR, pertaining to incorporating technical services from Pillar 2 and collaboration with local data stewards, focusing on ERN registries and selected OMICS data resources
- **Distributed and federated consent control WF**: defining where and how consent control is done based on the state-of-the-art and fitting it into the overall architecture of the VP. Defining other legal bases and definitions of roles (controller vs. processor in GDPR) for entities contributing or interfacing to the VP
- **Authentication Authorisation Infrastructure WF**: providing Authentication and Authorization Infrastructure (AAI) to be used by other components of VP
- **Personal data linkage service WF**: identify datasets which belong to the same person (Privacy-Preserving Record Linkage)
- **Resources for sharing experimental data and materials WF:** Improving, adapting, scaling-up and documenting resources for data and material deposition, access and sharing
- **Query builder WF:** Developing a federated discoverability and query facility for the VP

EJP RD

Funded by the European Union
GA n°825575

Defined Cross-Pillar Collaborations _

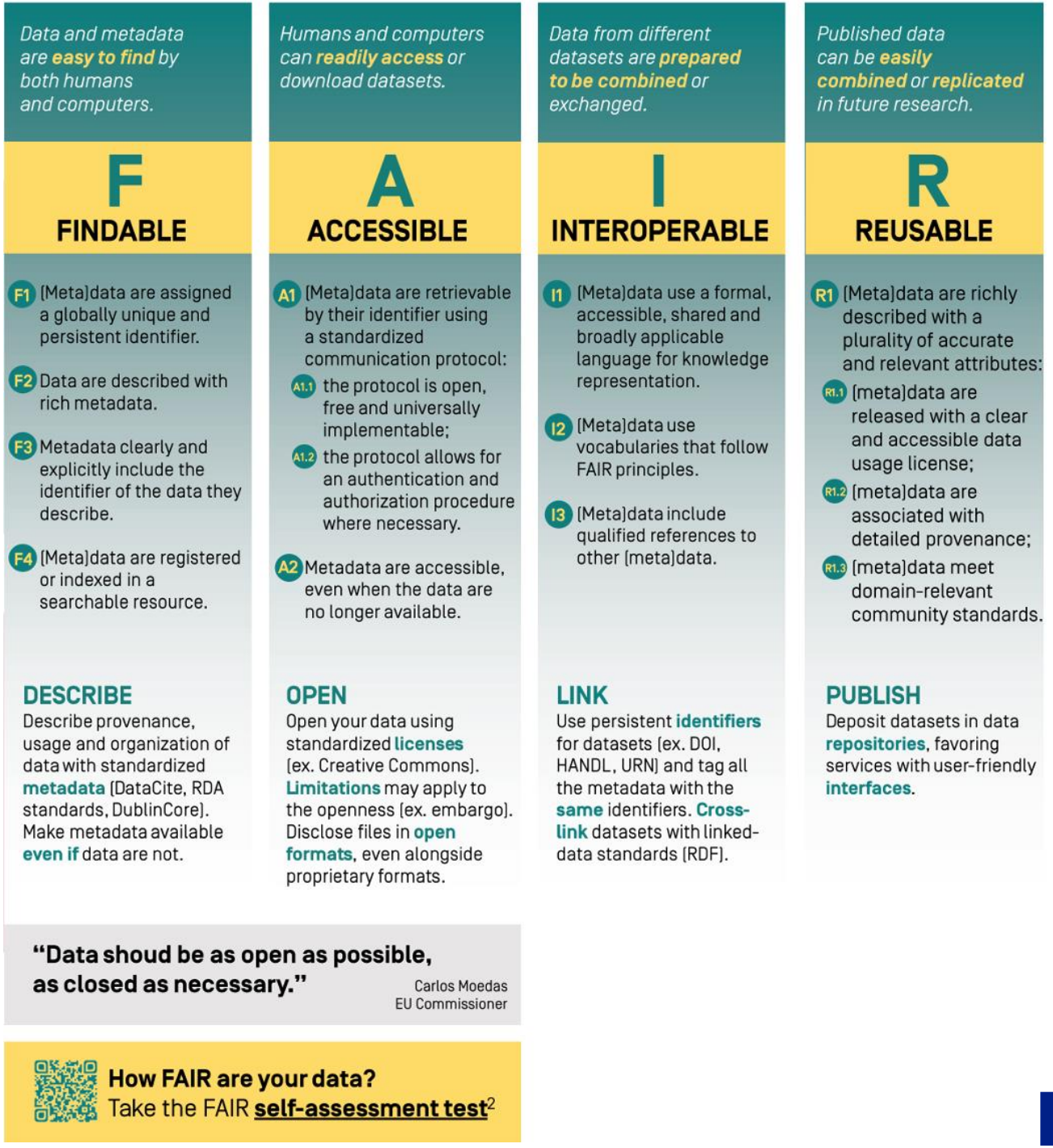
Promoting Open Science / data access

- *Innovation Management Toolbox integration with Central Helpdesk & VP*
- *ERN Registry Task Force& Pillar 2 Interoperability Work Focus*
- *JTC2019 funded team's preparation by Pillar 2 (Data deposition, FAIRification, VP requirements, training)*

EJP RD

Funded by the European Union
GA n°825575

Data Management Plan _ Compliance to:



EJP RD

Funded by the European Union
GA n° 825575

Data Management Plan _ recommendations

File Format

When listing out the data formats you will be using, make sure to include:

- The necessary software to view the data (e.g. SPSS v.3; Microsoft Excel 97-2003).
- Information about version control.
- If data are stored in one format during collection and analysis and then transferred to another format for preservation: list out features that may be lost in data conversion such as system specific labels.

When selecting file formats for archiving, the formats should ideally be:

- Non-proprietary, unencrypted, uncompressed, commonly used by the research community.
- Compliant to an open, documented standard: interoperable among diverse platforms and applications, fully published and available royalty-free, fully and independently implementable by multiple software providers on multiple platforms without any intellectual property[2].

File formats extensions for reusability/preservation:

Type of data	APPROPRIATE	ACCEPTABLE	NOT SUITABLE
Tabular data with extensive metadata	.csv - .hdf5	.txt - .html - .tex - .por	
Tabular data with minimal metadata	.csv - .tab - .ods - SQL	.xml if appropriate DTD - .xlsx	.xls - .xlsb
Textual data	.pdf - .txt - .odt - .odm - .tex - .md -.htm - .xml	.pptx - .pdf with embedded forms - .rtf	.doc - .ppt
Code	.m - .R - .py - .iypnb - .rstudio - .rmd – NetCDF	.sdd	.mat - .rdata
Digital image data	.tif - .png - .svg - .jpeg	jpg - .jp2 - .tif - .tiff - .pdf - .gif - .bmp	.indd - .ait - .psd
Digital audio data	.flac - .wav - .ogg	.mp3 - .mp4 - .aif	
Digital video data	.mp4 - .mj2 - .avi - .mkv	.ogm - .webm	.wmv - .mov
Geospatial data	NetCDF, tabular GIS attribute data, .shp - .shx - .dbf - .prj - .sbx - .sbn - PostGIS - .tif - .tfw - GeoJSON	.mdb - .mif	
CAD/vector and raster data	.x3d - .x3dv - .x3db - PDF3D .pdf	.dwg - .dxf	
Generic data	.xml - .json - .rdf		

EJP RD

Funded by the European Union GA n° 825575

Data Management Plan _ recommendations

A better efficiency in Code Management

VERSIONING

Versioning systems are powerful tools for code management. The most used is **Git**, it's free and open :

- It allows to **track changes** and to undo changes if needed. You can manage easily different versions of your code
- Connected to a repository your code and its modifications are **automatically backuped**
- You can also **work in team** easily on the same code

SHARING

In order to **share your code and make it visible**, repositories provide various services like version management system, wikis, task management and issues tracking, one of the most known is **Github**.

DESCRIBING

README documentation is a really important part of coding. It allows you to **explain your code**, for you and others. You should add rich metadata and documentation (README, LICENSE, comments on code...) on any publication of the code.

Some tools like sphinx-doc.org and doxygen.nl can help you by going through your code and generating a preformated documentation.

LICENSING

It is important to explain **how your code can be used** by others (and related restrictions).

You have at least three options :

- Open source licenses (permissive as MIT or GPL)
- Academic licenses (restrict commercial usage)
- Commercial licenses (reserve commercial usage)

PUBLISHING

Don't forget to **generate a DOI** to uniquely identify a version of your software and to easily cite it.

Most code repository (like Zenodo or c4science) generate a DOI for your deposit.

TIP : Github provides an integration with Zenodo.

PRESERVING

Preservation is important for keeping your work secure and also for scientific validation.

C4science is a solution to **preserve your code** for the long term. If you are using another code repository, you can **always make a copy on c4science for preservation.**

EJP RD

Funded by the European Union GA n° 825575

Data Management Plan _ recommendations

Data Masking

ADVANTAGES
WHY IT'S WORTH

- Complies with law
- Makes data sharable
- Prevents data misuse
- Makes data publishable

APPLICABILITY
TESTS ON HUMANS / SENSITIVE DATA

- Name, identification number, location data, online identifier, etc.
- Factors specific to the physical, physiological, genetic, mental, economic, cultural or social identity

TECHNIQUES

PSEUDONYMIZATION
REVERSIBLE
(FOR WORKING DATA)

REPLACING
Replace data by identifiers. The key is stored separately and securely.

ENCRYPTING
Encrypt the data and store the key securely. Appropriate for long-term preservation, not for data publishing.

ANONYMIZATION
IRREVERSIBLE
(FOR PUBLISHED DATA)

GENERALIZING
Diminish granularity by generalizing the variables. Appropriate for data too specific or unique records.

SHUFFLING
Shuffle data over one / several columns without compromising their utility.

FAKING
Prevent the identification of specific records, adding fake data while preserving correlations.

REMOVING
Suppress data or part of the outlier records. Appropriate for processing identifiers.

3RD PARTY DATA
Using commercial datasets or collaborationg in a joint research? Then, define a **contract** for data sharing or publication, and distinguish between research **authors** and data **owners**.

HINT
Mitigate the identification risk, but preserve the data utility for research.

SOME TOOLS
TO MASK IDENTITY OR ASSESS IDENTIFICATION RISKS

- ARX Data Anonymization Tool [Java][1]
- Amnesia [online][2]
- ARGUS [Java][3]
- sdcMicro [R][4]
- Differential privacy queries [SQL][5]
- Faker [Python][6]

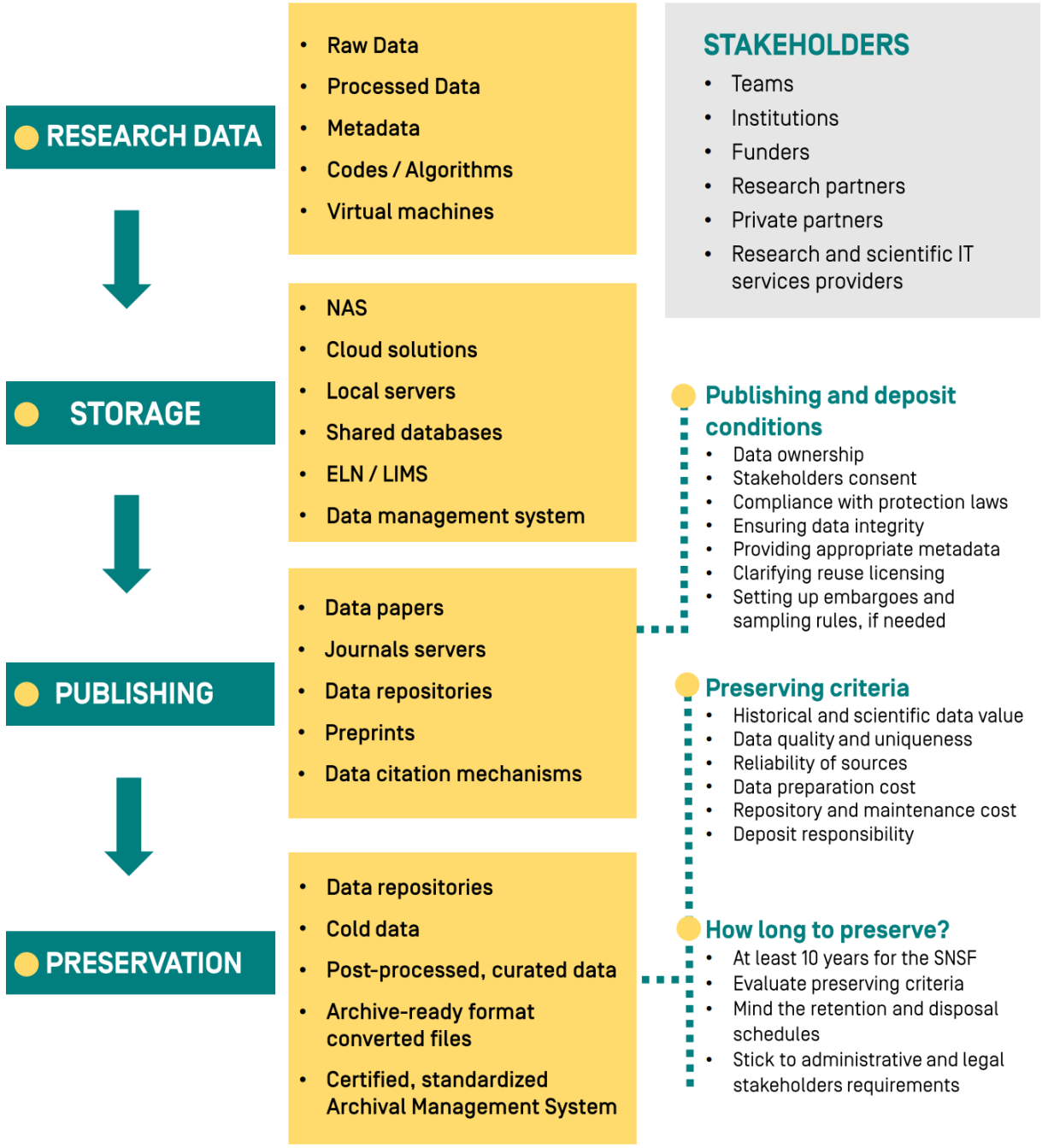
SUPPORT AND LAWS

GDPR

Funded by the European Union
GA n° 825575

Data Management Plan _ recommendations

Storage, publication and preservation



EJP RD

Funded by the European Union GA n° 825575

European Open Science Cloud (EOSC)

- EJP RD **described impact**: actively contributing to the EOSC
  - (i) **supporting** the EOSC to **apply FAIR Principles** (in league with the GO FAIR network),
  - (ii) **aligning** with EOSC on the respective **data plus software quality assurance approaches and the data governance principles**
  - (iii) **developing** the EJP RD data **Virtual Platform** to be a multi-tenant **federated** architecture able **to support a plurality of service providers and service consumers**, as needed for EOSC
  - (iv) **exploring** cross-fertilising **sustainability models for the life science ecosystem** with **EJP RD** being the **leader on rare diseases** aspects of this ecosystem

EJP RD

Funded by the European Union
GA n°825575

EJP RD 2019 _ Open science "results" (non-exhaustive)

Examples of Public Deliverables

*available after EC validation

D2.1	Final list of prioritization criteria*
D2.2	Prioritization scheme including decision-making process*
D5.1	EJP RD website
D5.2	EJP RD Newsletter

D10.5	Report on the State of the Art of existing resources*
D11.1	First Ontological model of resources metadata*
D11.6	Virtual platform of RD resources annotated with EJP ontological model*
D11.11	First version of Additional facilities integrated to resources regarding data deposition and access, including user guidelines and documentation*
D11.16	First Report on processed genome-phenome datasets and multi-omics use cases analysed, including description of new cloud and online analysis functionalities and tools*

D14.3	First Report on course on interpretation of genetic variants and quality standards*
D14.7	First Report on sample data management training workshops*
D14.9	First Report on International course on Rare Disease Registries and FAIRification of data at source*
D15.1	First Report on Expert Patients and Researchers EURORDIS Summer School*
D15.5	First Report on EURORDIS' Leadership Programme*
D17.1	Results of survey on preferences, needs and resources from the ERNs ecosystem*
D17.2	EJP RD ERN training programmes*

EJP RD

Funded by the European Union GA n°825575

EJP RD 2019 _ Open science "results" (non-exhaustive)

- **Publications**
  - 5 Open Access
- **GitHub**
  - Main https://github.com/ejp-rd-vp 7 repositories, 188 commits
  - https://github.com/EBISPOT/EJP-Ontology 138 commits
- **WikiPathways** https://www.wikipathways.org/index.php/Portal:RareDisease
  - 41 Pathways
- **Ndex network database** http://www.ndexbio.org/#/networkset/d4048ad7-1281-11ea-bb65-0ac135e8bacf
  - 2 networks
- Set up cloud-analysis virtual machine and **evaluated workflows reproducibility in collaboration with EOSC Life** to adopt:
  - **Common Workflow Language**
  - **Research Objects-Crate** (establishing a lightweight approach to packaging research data with their metadata) as a mechanism to work towards reproducibility and repeatability;
  - **Galaxy*** as a friendly front-end to implement workflows

*open-source application to develop and maintain a system that enables researchers without informatics expertise to perform computational analyses through the web

Funded by the European Union GA n°825575

THANK YOU

EUROPEAN JOINT PROGRAMME
RARE DISEASES

BACK-UP

EUROPEAN JOINT PROGRAMME
RARE DISEASES

Pillar 1: Collaborative research funding

EUROPEAN JOINT PROGRAMME
RARE DISEASES

Pillar 1: Activities

WP6: Joint Transnational Calls (JTC) for collaborative research projects

Open to research teams from countries with funders involved – min of 4 teams from 4 countries. Topics spanning from pre-clinical, translational to clinical research.

WP7: Networking Support Scheme (NSS) Call to share knowledge on rare diseases

Encourages research networks to share knowledge on rare diseases and rare cancers. Small support schemes for networking (workshops/events/share of knowledge). Max 10 participants from 3 countries. 30 K€ max. Open on a continuous basis.

EJP RD

Funded by the European Union
GA n°825575

Pillar 1: Activities

WP8: Rare Diseases Research (RDR) challenges Call & Networking Event

Facilitates and funds collaboration between industry, academia, SMEs, and patient organizations to solve specific research challenges in rare diseases.

#4 RDRchallenges are:

- Delivery system for intranasal administration of biological drugs to neonates
- Characterize Rare Bone Disorders (RBD) Mobility Challenges in Real World Setting
- Development of a non-invasive tool for measuring rare disease patient mobility in daily living
- Pre-clinical assay to detect instability of microsatellite repeat expansions) set by industry and validated by EJP RD.

1.5 Mio€ total budget (375 000€ per project). Short term (max. 18 months). Registration for the Networking Event is closed. The call will be launched in March.

WP9: Monitoring of funded projects

Monitoring of all projects funded through EJP RD and previous E-Rare projects

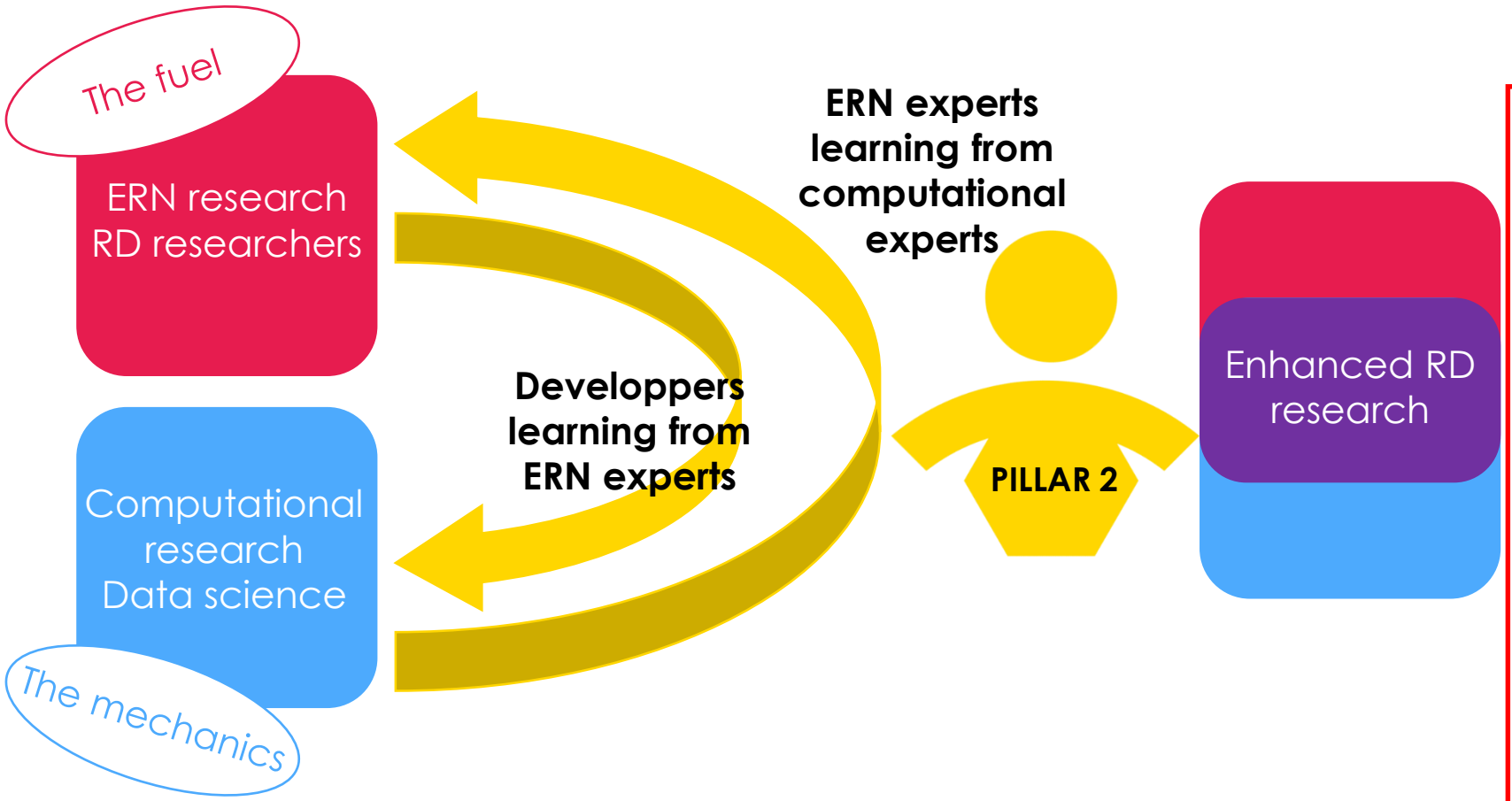
EJP RD

Funded by the European Union
GA n°825575

Pillar 2: Innovative coordinated access to data and services for transformative rare diseases research

EUROPEAN JOINT PROGRAMME RARE DISEASES

Pillar 2 target: FAIR-based virtual platform



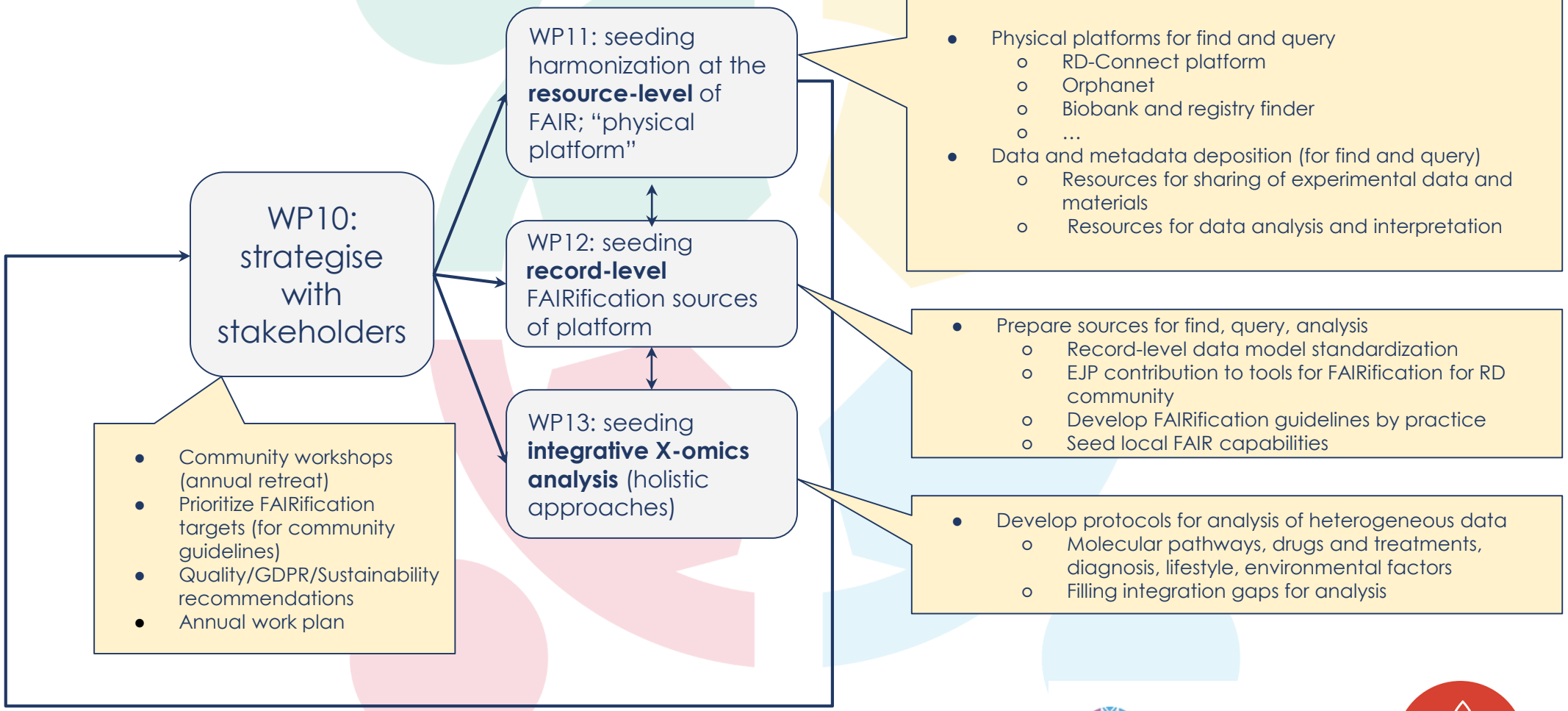
A powerful substrate for translational research:

- **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

EJP RD

Funded by the European Union
GA n°825575

PILLAR 2 WORKFLOW



EJP RD

Global Alliance for Genomics & Health
Collaborate. Innovate. Accelerate.

Funded by the European Union
GA n°825575

Pillar 2: Activities

- **WP10: User-driven strategic planning and transversal activities for Pillar 2 data ecosystem**

Annual strategic meetings with users (ERNs) & developers to define the priorities – coordination of outputs & needs – technical GDPR implementation – quality, sustainability and scaling up

- **WP11: Common virtual platform for discoverable data and resources for RD research**

Metadata & ontological models – FAIR compliance – data deposition & access to data infras – online tools. A first EJP RD « Linked Data Platform » –virtual platform- has been set up and it is currently available on the EJP RD website.

- **WP12: Enabling sustainable FAIRness and Federation at the record for RD data, patients and samples**

Alignement of core interoperability standards – software for FAIR ecosystem – FAIRification support. Pillar2 proposal for ERN registries: a document to help ERNs consider registry interoperability topics and how to work with EJP-RD.

- **WP13: Enabling multidisciplinary, holistic approaches for rare diseases diagnostics and therapeutics**

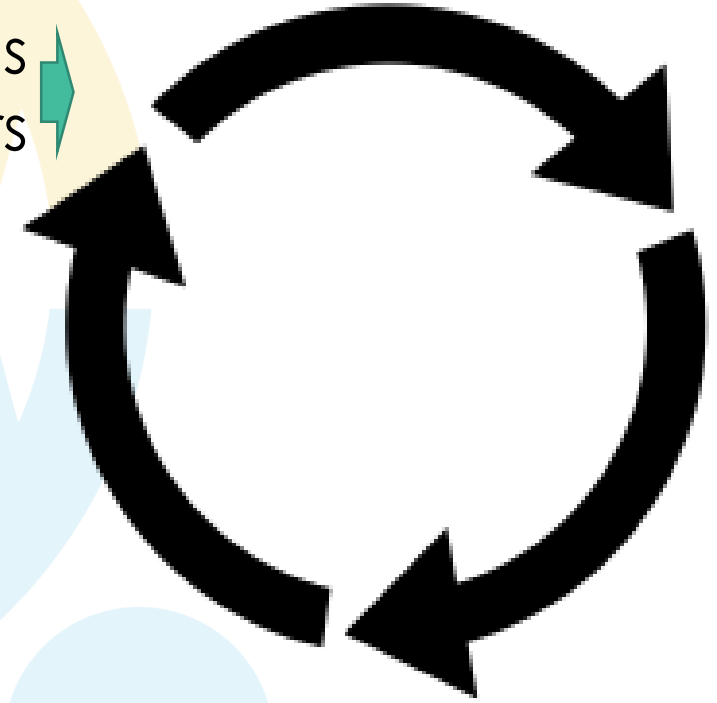
System biology approaches for RD – biological pathways – variants to function – environmental toxicology – treatment drugs - proof of principle studies. The video "Yakup's Journey to Hope" documenting one Rare Disease patient's diagnostic journey, has been recently released, it was prepared together by EJP RD members and GA4GH.

Funded by the European Union GA n°825575

Role of use cases

Use case descriptions
Volunteer stakeholders

Requests for use cases
Results for feedback



Stakeholder representatives
+ P2 mediators
(Use case work focus)

Pillar 2 agile development cycles
(all work foci)

EJP RD

Funded by the European Union
GA n°825575

Pillar 3: Capacity building and empowerment

EUROPEAN JOINT PROGRAMME
RARE DISEASES

Pillar 3: Activities

- **WP14: Trainings on data management & quality**
- **Objectives**: Decrease RD data fragmentation and increase data quality which will raise the level of capacities and help data sharing and networking within the RD community (existing and new courses)
  - **Courses**:
    - 1) Orphanet nomenclature
    - 2) standards & quality of genetics/genomics data in clinical practice
    - 3) strategies to foster undiagnosed diseases
    - 4) biobanks sample data management
    - 5) rare diseases registries & FAIRification at source – European Rare Diseases Registry Infrastructure

- **WP15: Capacity building & training of patients and researchers in rare diseases research and processes**
- **Objectives**: Improve RD research & innovation and enhance uptake of research results by building the capacity of the patient community and other key stakeholders (existing and new courses)
  - **Courses:**
    - 1) EURORDIS Summer school on Medicines Research & Development
    - 2) EURORDIS Winter School on scientific innovation and translation research
    - 3) EURORDIS Leadership School on Healthcare and Research
    - 4) Education material and activities for paediatric patients (2021)

EJP RD

Funded by the European Union
GA n°825575

Pillar 3: Activities

WP16: Online academic education course

- **Objectives:** Provide an EU-wide streamlined education programme on RD research to all interested stakeholders via an e-learning (brand new);
- Based on assessed needs of the RD community – in collaboration with universities – 10 to 12 modules with accreditation – e-learning format open to all – Future Learn platform

WP17: ERN RD training & support programmes

- **Objectives:** Deliver research training programs for the European Reference Networks (ERNs) focusing on cross-cutting and overarching research themes (brand new).

Based on four groups (Neuro, Neoplasm & malformation, Organs, Systemic) – preferences, needs and resources of ERNs – tailored for and performed by ERNs.

The aim is to **deliver training programs as research training workshops** (2 days workshop) **and/or research fellow exchange** through the ERN training groups (across national borders with a duration of 2 weeks up to 3 months).

WP18: Development and adaptation of training activities

- **Objectives**: Ensure that activities within Pillar 3 address the developing education and training needs in RD research of key stakeholders across different EU countries (specific needs of EU 13 countries), according to progress of Pillars 2 & 4 and emerging needs of ERNs

EJP RD

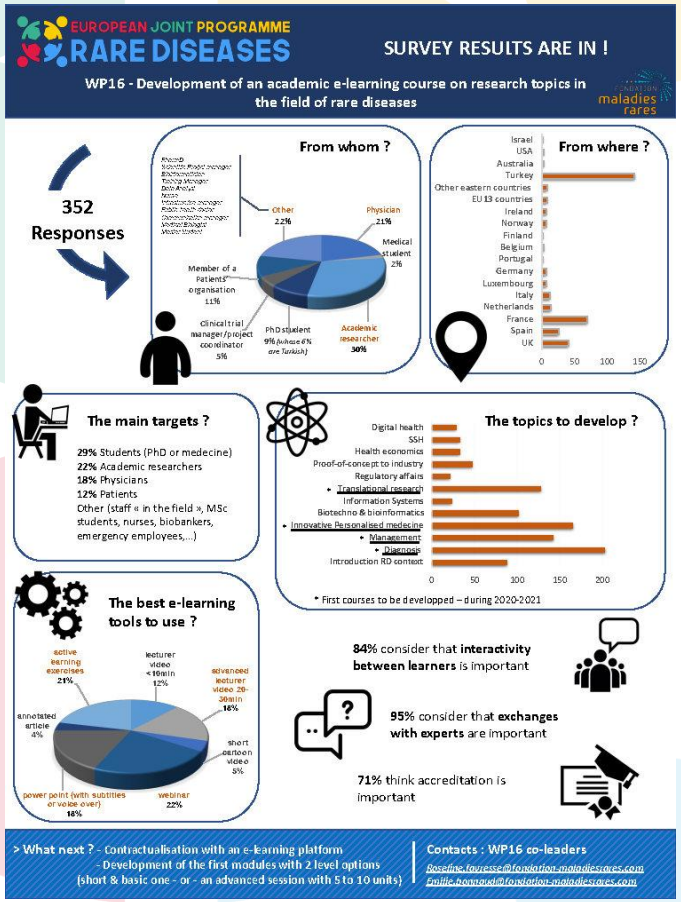
Funded by the European Union
GA n°825575

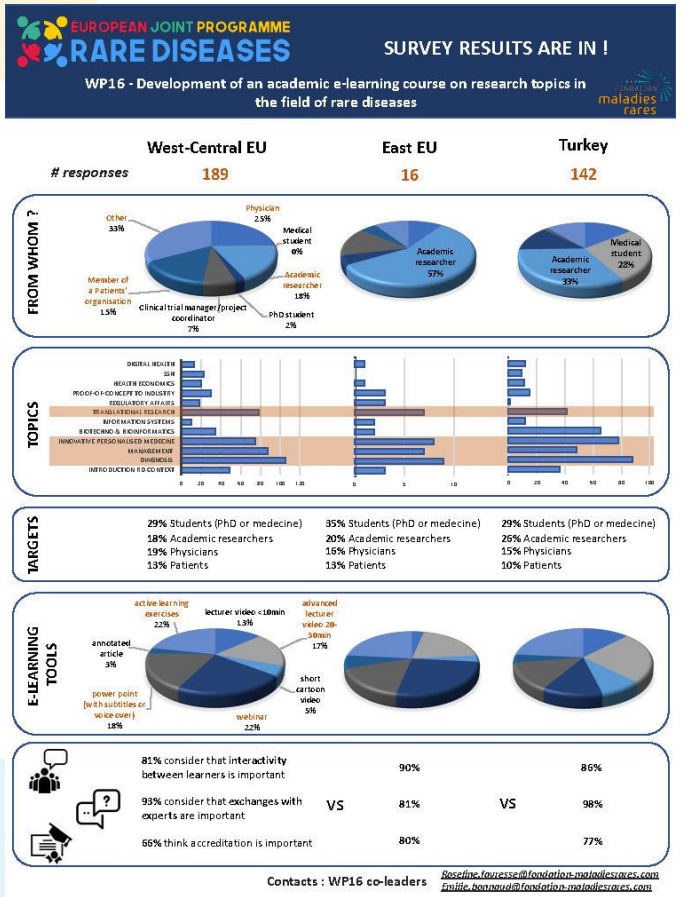
Pillar 3: Activities

- **WP16: Online academic education course**

Provide a EU-wide streamlined education programme on RD research to all interested stakeholders via an e-learning (brand new):

- Based on assessed needs of the RD community
- Done in collaboration with universities
- 10 to 12 modules with accreditation
- e-learning format open to all – Future Learn platform.
- A survey aimed to assessing the needs, target audience and main topics of the academic education course has been done, results are available on the EJP RD website.





Funded by the European Union GA n°825575

Pillar 4: Accelerating the translation of high potential projects and improving outcomes of clinical studies in small populations

EUROPEAN JOINT PROGRAMME
RARE DISEASES

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 (TFG) - 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 (validation of outcomes from ASTERIX, IDeAl, InSPiRe).

EJP RD

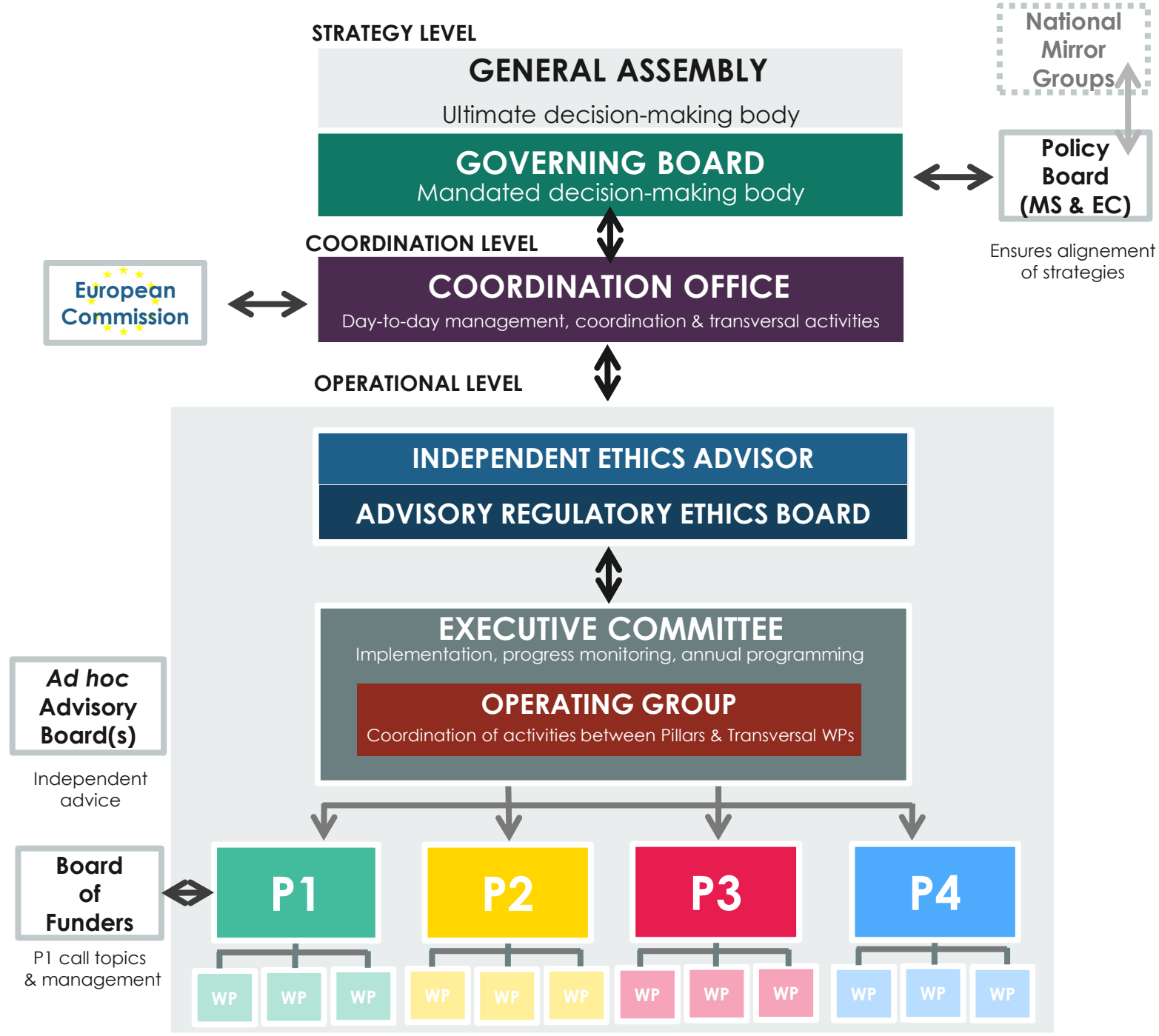
Funded by the European Union
GA n°825575

EJP RD

Funded by the
European Union
GA n°825575

EJP RD GOVERNANCE

EUROPEAN JOINT PROGRAMME
RARE DISEASES



Funded by the European Union
GA n°825575

POLICY BOARD

- The **POLICY BOARD** will have a major role in ensuring this dialogue and translation through its participation is EJP RD strategy and sustainability development. It will meet once a year.
- The Policy Board will be constituted from:

- Representatives of national ministries of research and health;
- Representatives of European Commission Directorates: DG RTD, DG Santé, DG Connect;
- Representative of patients (EURORDIS);
- Representative of the pharmaceutical industry and public-private initiatives (e.g. EFPIA, IMI, EUCOPE, EuropaBio);
- Representative of regulatory authorities (e.g. European Medicines Agency, EMA, esp. Committee for Orphan Medicinal Products, COMP, EuNetHTA);
- Chair of the European Strategy Forum on Research Infrastructures (ESFRI);
- Chair and vice-chair of the International Rare Diseases Research Consortium (IRDiRC).

EJP RD

Funded by the European Union
GA n°825575

NATIONAL MIRROR GROUPS (NMGs)

Objective of the NMGs

The role of the NMGs will be to ensure national coordination of and with all rare diseases stakeholders to facilitate the alignment between national and EJP RD activities, to contribute to the objectives of the EJP RD and benefit from it.

Composition of the National Mirror Groups:

Although the creation and composition of aN NMG is at the discretion of each participating country, it is recommended to involve the following stakeholders:

- EJP RD Governing Board representative
- EJP RD Policy Board representative(s)
- Relevant national partners of the EJP RD
- Relevant national authorities (i.e. representatives of the ministry of Health, ministry of Research, etc.)
- Representatives of the National plan/strategy for rare diseases
- European Reference Networks members
- Research institution involved in RD research (participating to the EJP RD or not)
- Representatives of patient organisations
- Representative of Orphanet local teams

Funded by the European Union GA n°825575