European Joint Programme on Rare Diseases (EJP RD)

Yanis Mimouni - INSERM
EJP RD coordination team
Rare Diseases Landscape in Europe

1.4 billion € for RD research in FP7 & Horizon 2020

More than 900 units from 300 hospitals covering 26 countries

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

869 patient organisations from 76 countries

Funded by the European Union

Ga n° 825575

RARE DISEASES RESEARCH STRATEGY

INFRA STRUCTURES

RARE DISEASES RESEARCH

FUNDING

PATIENTS NEEDS

HEALTH CARE +
EUROPEAN JOINT PROGRAMME ON RARE DISEASES
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: 884 RD patient organisations 72 countries
+ ECRIN: 300 institutions >950 healthcare units 26 countries
+ EATRIS: 13 main national nodes
+ INFRAFRONTIER: 23 partners 15 countries
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+ ELIXIR: 220 research organisation 23 partners
+ BBMRI: 1 international partner 21 main national nodes 20 countries

50 Linked Third Parties

Jan 2019 Dec 2023

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50 Linked Third Parties
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 STRUCTURE

COORDINATION & TRANSVERSAL ACTIVITIES

INTEGRATIVE RESEARCH STRATEGY

SUSTAINABILITY

ETHICAL & REGULATORY

COMMUNICATION

1. FUNDING

2. COORDINATED ACCESS TO DATA & SERVICES

3. CAPACITY BUILDING & EMPOWERMENT

4. ACCELERATING TRANSLATION OF RESEARCH & THERAPY DEVELOPMENT
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

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

- Ideas generation & Sharing
- Discussion for potential of publication & Setting-up Publication Working Group
- Follow-up of undergoing articles
  - Journal Selection
    - OPEN ACCESS
  - Publication plan & Assignment of article parts (intro/review, methods, results, etc.)
  - Articles Writing & Adaptation
  - Article Submission & Addressing reviewers comments

Submission to EJPRD OG

ExCom Meeting

Publication Working Group

EJPRD Coo support

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

<table>
<thead>
<tr>
<th>Describe Background</th>
<th>Specific limitations and/or conditions for implementation (Article 25.2 Grant Agreement)</th>
<th>Specific limitations and/or conditions for Exploitation (Article 25.3 Grant Agreement)</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

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
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.
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)
Data Management Plan Compliance to:

FINDABLE
F1 Metadata are assigned a globally unique and persistent identifier.
F2 Data are described with rich metadata.
F3 Metadata clearly and explicitly include the identifier of the data they describe.
F4 Metadata are registered or indexed in a searchable resource.

ACCESSIBLE
A1 Metadata are retrievable by their identifier using a standardized communication protocol; the protocol is open, free and universally implementable; the protocol allows for an authentication and authorization procedure where necessary.
A2 Metadata are accessible, even when the data are no longer available.

INTEROPERABLE
I1 Metadata use a formal, accessible, shared and broadly applicable language for knowledge representation.
I2 Metadata use vocabularies that follow FAIR principles.
I3 Metadata include qualified references to other metadata.

REUSABLE
R1 Metadata are richly described with a plurality of accurate and relevant attributes.
R2 Metadata are released with a clear and accessible data usage license.
R3 Metadata are associated with detailed provenance.
R4 Metadata meet domain-relevant community standards.

DESCRIBE
Describe provenance, usage and organization of data with standardized metadata (DataCite, RDA standards, DublinCore). Make metadata available even if data are not.

OPEN
Open your data using standardized licenses (ex. Creative Commons). Limitations may apply to the openness (ex. embargo). Disclose files in open formats, even alongside proprietary formats.

LINK
Use persistent identifiers for datasets (ex. DOI, HANDLE, URN) and tag all the metadata with the same identifiers. Cross-link datasets with linked-data standards (RDF).

PUBLISH
Deposit datasets in data repositories, favoring services with user-friendly interfaces.

“Data should be as open as possible, as closed as necessary.”
Carlos Moedas
EU Commissioner

How FAIR are your data? Take the FAIR self-assessment test²
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.

### File formats extensions for reusability/preservation:

<table>
<thead>
<tr>
<th>Type of data</th>
<th>APPROPRIATE</th>
<th>ACCEPTABLE</th>
<th>NOT SUITABLE</th>
</tr>
</thead>
<tbody>
<tr>
<td>Tabular data with minimal metadata</td>
<td>.csv - .tab - .ods - .SQL</td>
<td>.xml if appropriate DTD - .xlsx</td>
<td>.xls - .xlsb</td>
</tr>
<tr>
<td>Code</td>
<td>.m - .R - .py - .jypnb - .rstudio - .rmd - NetCDF</td>
<td>.sdd</td>
<td>.mat - .rdata</td>
</tr>
<tr>
<td>Digital audio data</td>
<td>.flac - .wav - .ogg</td>
<td>.mp3 - .mp4 - .aiff</td>
<td></td>
</tr>
<tr>
<td>Digital video data</td>
<td>.mp4 - .mj2 - .avi - .mkv</td>
<td>.ogm - .webm</td>
<td>.wmv - .mov</td>
</tr>
<tr>
<td>CAD/vector and raster data</td>
<td>.x3d - .x3dv - .x3db - PDF3D .pdf</td>
<td>.dwg - .dxf</td>
<td></td>
</tr>
<tr>
<td>Generic data</td>
<td>.xml - .json - .rdf</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
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 backed up**
- 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**.

**DESCRIPTING**

**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 preformatted 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**.
## Data Management Plan Recommendations

### Data Masking

#### ADVANTAGES
**WHY IT’S WORTH**
- Complies with law
- Makes data shareable
- 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

<table>
<thead>
<tr>
<th>Pseudonymization (Reversible)</th>
<th>Anonymization (Irreversible)</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>REPLACING</strong></td>
<td><strong>GENERALIZING</strong></td>
</tr>
<tr>
<td>Replace data by identifiers. The key is stored separately and securely.</td>
<td>Diminish granularity by generalizing the variables. Appropriate for data too specific or unique records.</td>
</tr>
<tr>
<td><strong>ENCRYPTING</strong></td>
<td><strong>SHUFFLING</strong></td>
</tr>
<tr>
<td>Encrypt the data and store the key securely. Appropriate for long-term preservation, not for data publishing.</td>
<td>Shuffle data over one / several columns without compromising their utility.</td>
</tr>
<tr>
<td><strong>FAKING</strong></td>
<td><strong>REMOVING</strong></td>
</tr>
<tr>
<td>Prevent the identification of specific records, adding fake data while preserving correlations.</td>
<td>Suppress data or part of the outlier records. Appropriate for processing identifiers.</td>
</tr>
</tbody>
</table>

### SOME TOOLS

<table>
<thead>
<tr>
<th>TO MASK IDENTITY OR ASSESS IDENTIFICATION RISKS</th>
</tr>
</thead>
<tbody>
<tr>
<td>- ARX Data Anonymization Tool <a href="1">Java</a></td>
</tr>
<tr>
<td>- Annesia (Oracle) [2]</td>
</tr>
<tr>
<td>- ARGUS (Java) [1]</td>
</tr>
<tr>
<td>- sqlMicro [3]</td>
</tr>
<tr>
<td>- Differential privacy queries <a href="9">SQL</a></td>
</tr>
<tr>
<td>- Faker (Python) [10]</td>
</tr>
</tbody>
</table>

#### SUPPORT AND LAWS

- GDPR

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**3rd PARTY DATA**

Using commercial datasets or collaborating 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.
## Data Management Plan recommendations

### Storage, publication and preservation

<table>
<thead>
<tr>
<th>RESEARCH DATA</th>
</tr>
</thead>
<tbody>
<tr>
<td>• Raw Data</td>
</tr>
<tr>
<td>• Processed Data</td>
</tr>
<tr>
<td>• Metadata</td>
</tr>
<tr>
<td>• Codes / Algorithms</td>
</tr>
<tr>
<td>• Virtual machines</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>STORAGE</th>
</tr>
</thead>
<tbody>
<tr>
<td>• NAS</td>
</tr>
<tr>
<td>• Cloud solutions</td>
</tr>
<tr>
<td>• Local servers</td>
</tr>
<tr>
<td>• Shared databases</td>
</tr>
<tr>
<td>• ELN / LIMS</td>
</tr>
<tr>
<td>• Data management system</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>PUBLISHING</th>
</tr>
</thead>
<tbody>
<tr>
<td>• Data papers</td>
</tr>
<tr>
<td>• Journals servers</td>
</tr>
<tr>
<td>• Data repositories</td>
</tr>
<tr>
<td>• Preprints</td>
</tr>
<tr>
<td>• Data citation mechanisms</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>PRESERVATION</th>
</tr>
</thead>
<tbody>
<tr>
<td>• Data repositories</td>
</tr>
<tr>
<td>• Cold data</td>
</tr>
<tr>
<td>• Post-processed, curated data</td>
</tr>
<tr>
<td>• Archive-ready format converted files</td>
</tr>
<tr>
<td>• Certified, standardized Archival Management System</td>
</tr>
</tbody>
</table>

### STAKEHOLDERS
- Teams
- Institutions
- Funders
- Research partners
- Private partners
- Research and scientific IT services providers

### Publishing and deposit conditions
- Data ownership
- Stakeholders consent
- Compliance with protection laws
- Ensuring data integrity
- Providing appropriate metadata
- Clarifying reuse licensing
- Setting up embargoes and sampling rules, if needed

### Preserving criteria
- Historical and scientific data value
- Data quality and uniqueness
- Reliability of sources
- Data preparation cost
- Repository and maintenance cost
- Deposit responsibility

### How long to preserve?
- At least 10 years for the SNSF
- Evaluate preserving criteria
- Mind the retention and disposal schedules
- Stick to administrative and legal stakeholders requirements
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
### Examples of Public Deliverables

*available after EC validation*

<table>
<thead>
<tr>
<th>Number</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>D2.1</td>
<td>Final list of prioritization criteria*</td>
</tr>
<tr>
<td>D2.2</td>
<td>Prioritization scheme including decision-making process*</td>
</tr>
<tr>
<td>D5.1</td>
<td>EJP RD website</td>
</tr>
<tr>
<td>D5.2</td>
<td>EJP RD Newsletter</td>
</tr>
<tr>
<td>D10.5</td>
<td>Report on the State of the Art of existing resources*</td>
</tr>
<tr>
<td>D11.1</td>
<td>First Ontological model of resources metadata*</td>
</tr>
<tr>
<td>D11.6</td>
<td>Virtual platform of RD resources annotated with EJP ontological model*</td>
</tr>
<tr>
<td>D11.11</td>
<td>First version of Additional facilities integrated to resources regarding data deposition and access, including user guidelines and documentation*</td>
</tr>
<tr>
<td>D11.16</td>
<td>First Report on processed genome-phenome datasets and multi-omics use cases analysed, including description of new cloud and online analysis functionalities and tools*</td>
</tr>
<tr>
<td>D14.3</td>
<td>First Report on course on interpretation of genetic variants and quality standards*</td>
</tr>
<tr>
<td>D14.7</td>
<td>First Report on sample data management training workshops*</td>
</tr>
<tr>
<td>D14.9</td>
<td>First Report on International course on Rare Disease Registries and FAIRification of data at source*</td>
</tr>
<tr>
<td>D15.1</td>
<td>First Report on Expert Patients and Researchers EURORDIS Summer School*</td>
</tr>
<tr>
<td>D15.5</td>
<td>First Report on EURORDIS’ Leadership Programme*</td>
</tr>
<tr>
<td>D17.1</td>
<td>Results of survey on preferences, needs and resources from the ERNs ecosystem*</td>
</tr>
<tr>
<td>D17.2</td>
<td>EJP RD ERN training programmes*</td>
</tr>
</tbody>
</table>
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
THANK YOU
Pillar 1: Collaborative research funding
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.
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 RDR challenges 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
Pillar 2: Innovative coordinated access to data and services for transformative rare diseases research
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
PILLAR 2 WORKFLOW

WP10: strategise with stakeholders
- Community workshops (annual retreat)
- Prioritize FAIRification targets (for community guidelines)
- Quality/GDPR/Sustainability recommendations
- Annual work plan

WP11: seeding harmonization at the resource-level of FAIR: "physical platform"

WP12: seeding record-level FAIRification sources of platform

WP13: seeding integrative X-omics analysis (holistic approaches)

- Physical platforms for find and query
  o RD-Connect platform
  o Orphanet
  o Biobank and registry finder
- Data and metadata deposition (for find and query)
  o Resources for sharing of experimental data and materials
  o Resources for data analysis and interpretation

- Prepare sources for find, query, analysis
  o Record-level data model standardization
  o EJP contribution to tools for FAIRification for RD community
  o Develop FAIRification guidelines by practice
  o Seed local FAIR capabilities

- Develop protocols for analysis of heterogeneous data
  o Molecular pathways, drugs and treatments, diagnosis, lifestyle, environmental factors
  o Filling integration gaps for analysis
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.
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)
Pillar 3: Capacity building and empowerment
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)
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
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 at assessing the needs, target audience and main topics of the academic education course has been done, results are available on the EJP RD website.
Pillar 4: Accelerating the translation of high potential projects and improving outcomes of clinical studies in small populations
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, IDeAI, InSPiRe).
EJP RD GOVERNANCE
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).
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