EJP RD
European Joint Programme on Rare Diseases

H2020-SC1-2018-Single-Stage-RTD
SC1-BHC-04-2018
Rare Disease European Joint Programme Cofund

Grant agreement number 825575

Del 1.7
Fifth report from the face-to-face
ExCom and Policy Board meeting

Organisation name of lead beneficiary for this deliverable:
Partner 01 – INSERM

Due date of deliverable: month 55

Dissemination level:
Public
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<th>Abbreviation</th>
<th>Description</th>
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<tr>
<td>AOB</td>
<td>Any Other Business</td>
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<tr>
<td>AREB</td>
<td>Advisory Regulatory Ethics Board</td>
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<td>AWP</td>
<td>Annual Work Plan</td>
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<td>Consortium Agreement(s)</td>
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<td>CRN(s)</td>
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<td>European Credits Transfer System</td>
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<td>European Rare Disease Research Coordination and Support Action consortium</td>
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<td>ERN(s)</td>
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<td>European Patient Identity Management</td>
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<td>IHI</td>
<td>Innovative Health Initiative</td>
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<td>Innovation Management Toolbox</td>
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<td>National Mirror Group</td>
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<td>Orphan Drug Development Guidebook</td>
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<td>Person Month</td>
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<td>VP</td>
<td>Virtual Platform</td>
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<td>Work Package</td>
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EJP RD Executive Committee

3rd of July 2023
11:30 – 18:00 CEST
Hybrid meeting

Attached documents:
- Slides presented during the meeting: file “EJPRD_ExCom_20230703_all-slides”

List of participants

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<tr>
<td>Daria Julkowska</td>
<td>INSERM</td>
<td>coordinator WP1 - WP5</td>
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<tr>
<td>Ralph Schuster</td>
<td>DLR</td>
<td>P1 coleader WP6</td>
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<tr>
<td>Sonja van Weely</td>
<td>ZonMw</td>
<td>P1 coleader WP7</td>
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<tr>
<td>Ana Rath</td>
<td>INSERM (Orphanet)</td>
<td>P2 coleader WP10 - WP11</td>
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<td>Franz Schaefer</td>
<td>UKL-HD</td>
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<td>Roseline Favresse</td>
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<td>P3 coleader WP15 - WP18</td>
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<td>Biruté Tumiene</td>
<td>VUHSK</td>
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<td>Anton Ussi</td>
<td>EATRIS</td>
<td>P4 coleader WP3 - WP19</td>
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<td>Rima Nabbout</td>
<td>AP-HP</td>
<td>P4 coleader WP20</td>
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<td>Eva Bermejo-Sanchez</td>
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<td>WP2 and WP3 coleader</td>
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<td>Christine Fetro</td>
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<td>Irit Allon</td>
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<td>Sergi Beltran</td>
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<td>Pauline Adam</td>
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### Agenda

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<tr>
<td>11:30 – 11:45</td>
<td>Welcome from coordination</td>
<td>EJP RD Coordination</td>
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| 11:45 – 12:45 | AWP Y6: presentation of planned activities  
10 minutes per pillar | Pillar Leaders and All                                                  |
| 10:40 – 10:55 | Lunch Break                                                           |                                 |
| 13:45 – 15:45 | AWP Y6: Budget and Prioritisation                                      | All                             |
| 15:45 – 16:15 | Break                                                                 |                                 |
| 16:15 – 17:45 | Communication strategy for the last year of EJP RD                    | WP5 Leader & All                |
| 17:45 – 18:00 | AOB, Next steps                                                       | EJP RD Coordination             |
Minutes

AWP Y6: presentation of planned activities
See slides 2 to 40 “EJPRD_ExCom_20230703_all-slides”

Discussion:

- Pillar 0:
  - At the end of Year 6 (end August 2024): Periodic Report 6 (January to August 2024) and Final report (January 2019 to August 2024) to be submitted to EC.
  - P2 Consortium Agreement still ongoing: final reminder will be sent after the meeting.

- Pillar 1:
  - Ongoing discussion in funding activities on how to better capture in the future aspects related to data management, Data Management Plan, FAIRification, ethic, etc.; could be done through mandatory training.

- Pillar 2:
  - The Version v1 of the Virtual Platform (VP) was launched on Friday, June 30th: 22 resources are connected.
  - Closer collaboration with the funded project should be developed in the future: partners are trying to put in place better models in the Rare Diseases Partnership to have tighter connection (with all support activities). Possible proposed solution to better connect are: trainings (use of incentives?), wider dissemination, alignment with needs of researchers, online data marketplace/matchmaking, etc.
  - There is a need to better demonstrate that the VP is serving the RD community. In EJP RD, initial focus was done on the ERNs, but now need to be expanded: better dissemination (in lay language, not technical) is needed as well as training of users/future users now that the VP has been launched.

- Pillar 4:
  - Some exchanges ongoing between the Clinical Study Support (WP20) and WP4 of ERICA.
  - Training recorded material during the workshop should be further disseminated as open source webinars.

ACTIONS

→ Based on lessons learned from EJP RD, Consortium Agreement(s) (CA) of the Rare Diseases Partnership (RDP) (Framework CA and specific CA if needed for some activities) should be signed before the start of the partnership.

→ Lessons learned to be developed in Year 6 for P1 should include feedback from funders, patients, applicants.

→ Pillar 2 will launch a final survey to the RD community to evaluate results of the EJP RD (results to be compared to the initial survey); all other Pillars should indicate to P2 if they would like to include questions.
AWP Y6: Budget and Prioritisation

Discussion:

- **Pillar 0.** The pillar 0 additional budget for year 6 is identified around €600,000 mainly for coordination activities (WP1) and follow up on ethics activities (WP4). The estimations are based on the previous declared costs and estimation of year 5 and year 6 in the annual work plan (person-months). Participants agreed that only budget needed for year 6 should be allocated to avoid rewarding partners who overspent in the first years of the project.

  In WP4, the budget was overall under planned from the start. The planned budget did not consider all the activities that should have ethic reviews (JTCs projects, Networking Support Scheme project, fellowships, and workshops). WP4 leaders will review the allocation of PM in year 6 for some partners that are not active to reduce costs.

- **Pillar 1.** In pillar 1, there will not need major additional budget to cover year 6 based on the available central budget set aside. WP7 and WP9 personnel will need some budget and costs for organisation of the WP9 monitoring meeting in 2024. We removed the partners that overspent in the previous years from the calculation.

- **Pillar 3.** In pillar 3, there is no identified additional budget needed for 2024. The person-months in 2024 will be covered by internal unused budget (in personnel but mainly in other direct costs) especially in WP15 and WP16.

- **Pillar 4.** In WP19, with agreement of partners, reallocation of unused personnel budget will be transferred to the WP19 partners (EATRIS, FTELE and CVBF) to cover their needs for year 6. Additional other direct costs will be requested for the IMT and ODDG maintenance.

  The additional budget in WP20 were requested in the last annual work plan year 5, no further need was identified.

- **Pillar 2.** Pillar 2 needs internal prioritization of activities and budget for year 6 because there is not enough left to have all activities continued in year 6. Pillar 2 leaders will organise individual meetings with partners to identify budget reallocation. €275,402 have been identified as additional need after internal reallocation and use of central budgets.

**ACTIONS**

- Coordination will share the results of the discussion to the pillar leaders for validation.
- Pillar 2 will organise individual meetings with partners to identify budget reallocation.
- The budget transfers will be listed in the annual work plan year 6 and subject to validation by the General Assembly in September 2023.
Communication strategy for the last year of EJP RD
See slides 42 to 55 “EJPRD_ExCom_20230703_all-slides”

Discussion

- The meeting discussed the current status of stakeholder outreach in different Pillars. Some stakeholders are not fully reached. The group acknowledged that further engagement is necessary to fully achieve the objectives. Moreover, a revision of the initial targeted stakeholder needs to be done.
- Stakeholder objectives and involvement were discussed.
  - The suggestion to address stakeholder objectives with the Policy Board was made.
- Proactive national-level communication was emphasized.
- It was mentioned that the current strategy lacked focus on national-level activities, particularly involving health policy makers.
- Active and passive communication methods were highlighted.
- Proposals for webinars, tutorials, and incentives for researchers were put forward, inspired by Swiss health practices.
- The importance of informing the wider rare disease community about RD and projects was stressed.
- The idea of collecting input for the Rare Disease Day video was suggested.
  - Video with compiled testimonials for a general audience were proposed.
  - The RD Day video’s target audience was debated, with one opinion for RD patients and another for a broader audience.
  - A general theme showcasing progress and added benefits was suggested.
- Shared dissemination responsibility was highlighted.
- Collaborating with patient groups to expand outreach was recommended.
- The team agreed to enhance outreach beyond the current community.
- Inputs from the team on moving forward with the communication strategy were sought.

ACTIONS

→ Update the targeted audience and expand outreach
→ Collaborate with existing organizations for targeted outreach
→ Develop targeted tutorials and materials for different stakeholders
→ Dissemination of EJP RD outputs through different formats (journalistic reports, videos, events...)
→ Make the website more user-friendly

AOB
No other topics discussed.
EJP RD Policy Board and Governing Board meeting

4th of July 2023
9:30 – 18:00 CEST
Hybrid meeting

Attached document:
- Slides presented during the meeting: file “EIPRD_PB-GB-meeting_July2023-All-slides”

List of participants

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<th>Name</th>
<th>Board</th>
<th>Country</th>
<th>Presence</th>
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<tr>
<td>Alexandra Tataru</td>
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<td>France</td>
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<td>Ana Rath</td>
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<td>Anton Ussi</td>
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<td>Invited [Copenhagen Economist]</td>
<td>Denmark</td>
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<tr>
<td>Christina Kyriakopoulou</td>
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## Agenda

<table>
<thead>
<tr>
<th>Time</th>
<th>Session Description</th>
<th>Organizer</th>
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<tbody>
<tr>
<td>9:30 – 9:40</td>
<td>Welcome from coordination</td>
<td>EJP RD Coordination</td>
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<td>9:40 – 10:40</td>
<td>EJP RD – Sustainability</td>
<td>Copenhagen Economics &amp; All</td>
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<tr>
<td>10:40 – 10:55</td>
<td>Break</td>
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| 10:55 – 12:30 | EJP RD – Pillars’ contributions towards objectives of EJP RD: summary of achievements and final activities for Year 6  
• 15 minutes per pillar  
• Feedback from the board | Pillar Leaders & All                  |
| 12:30 – 13:30 | Lunch break                                                                          |                                    |
| 13:30 – 14:45 | Rare Diseases Partnership – Presentation of the Strategic Research & Innovation Agenda  
Feedback from the Boards | EJP RD coordination & All            |
| 14:45 – 16:00 | Rare Diseases Partnership – Presentation of the proposal  
Feedback from the Boards | EJP RD coordination & All            |
| 16:00 – 16:30 | Break                                                                                |                                    |
| 16:30 – 17:45 | The place of the Rare Diseases Partnership in the overall RD landscape                | EJP RD coordination & All          |
| 17:45 – 18:00 | AOB, Next steps                                                                      | EJP RD coordination                |
Minutes
EJP RD – Sustainability
See slides 3 to 14 “EJPRD_PB-GB-meeting_July2023-All-slides”

Discussion

• **Stakeholders consulted/interviewed**: The stakeholders consulted/interviewed for the sustainability strategy of EJP RD developed by Copenhagen Economics include pharma companies, small biotech firms, investor communities interested in funding start-ups, and patient representatives.

• **Public-Private Non-Profit Model**: There is a discussion about proposing a public-private non-profit model for the sustainability strategy. The focus is on commercial use and exploring different payment models, such as companies paying for specific services or providing access to data analysis through subscription fees.

• **Exit Strategy and Sustainability**: The strategy acknowledges the recommendation from Horizon 2020 projects and emphasizes the need for an exit strategy. It aims to provide services, expertise, and a data hub while advancing in a sustainable way beyond relying solely on public funds. Open-source provisions are considered, but sustainable funding beyond public grants is emphasized.

• **Role of SMEs**: There is recognition of a marketplace of small and medium-sized enterprises (SMEs) within the academic and data groups. Some SMEs can be customers, and it is believed that pharma companies can also be customers for specific expertise and treatment value solutions.

• **Access to Data Discovery and Standardized Methodologies**: The strategy emphasizes that access to data discovery should be available to everyone, but additional layers can be added. Developing standardized methodologies and ensuring regulatory compliance are important for working towards the same goal of delivering treatments to patients.

• **Connecting Sustainability with Private Companies**: The challenge is how to convince private companies to pay fees for services. One approach discussed is making patient cohorts accessible on the platform, distinguishing between discovery level and access level. Long-term targets include developing a business model for accessing data for commercial purposes.

• **Business Development Model**: There is a missing model in the business development part, which involves accompanying a customer to access research tools. This would be a service provided by experts who understand specific needs and have a strong network.

• **Building Attractiveness**: The strategy aims to build attractiveness not only for customers but also for resources joining the network. An attractive membership offer, along with a strong service offer, is deemed important. Consideration is given to the legal structure as well.

• **Ethical Compliance and Data Ethics**: The importance of ethical and regulatory compliant data is highlighted. The usefulness of data, especially for commercial partners, should be considered along with ethics compliance of patients.
**ACTIONS**

→ The recommendations from Copenhagen Economics will be considered in the next steps of the European Joint Program on Rare Diseases and the development and implementation of the Rare Diseases Partnership.

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**EJP RD – Pillars’ contributions towards objectives of EJP RD: summary of achievements and final activities for Year 6**

See slides 15 to 98 "EJPRD_PB-GB-meeting_July2023-All-slides"

**Discussion**

- **Pillar 3**
  - The possibility to calculate ECTs is being considered in the RD partnership through the building of university diplomas.
  - In order to replicate training in the countries, train-the-trainer model will be further developed in the RDP, with the contribution of the National Mirror Groups and in collaboration with the WP ‘Fostering engagement of underrepresented countries’ to adapt the training to national needs.

- **Pillar 2**
  - First version of the VP launched just before the meeting: it will be difficult to measure the impact on the resources access at the beginning.

---

**ACTIONS**

→ The Policy Board was offered the possibility to further comment on the draft AWP Y6 by email until July 17th: no comments received.

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**Rare Diseases Partnership – Presentation of the Strategic Research & Innovation Agenda**

See slides 100 - 131 "EJPRD_PB-GB-meeting_July2023-All-slides"

**Discussion**

- The SRIA should be the guiding document for the RD Partnership. It should not be too operational. More operational KPIs will be followed through the monitoring of the RDP.
- SRIA is a living document but is a high level roadmap. Revision of the SRIA in the lifetime of the RDP will be part of activities on Strategy (but will not be done annually, it is not an Annual Work Plan) == some factor inducing revision of the SRIA could be included in this Task (for example, revision of Orphan regulation).
- Monitoring of KPI should be computed automatically as much as possible.
- Monitoring should be done on activities/areas where the RDP can really have an impact (also to limit the list of KPIs).
Some reference should be set up for the monitoring of the KPIs, evolution of KPIs have to be comparable with something.

**ACTIONS**

→ **SRIA Task Force** will finalise the KPIs and the SRIA, taking into account results from the public consultation.

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**Rare Diseases Partnership – Presentation of the proposal**

See slides 132 to 162 “[EJPRD_PB-GB-meeting_July2023-All-slides]”

**Discussion**

No Discussion/comments with/from the audience on this topic.

**ACTIONS**

→ **Coordination team and WP/Tasks leaders** will follow the planned timeline for the development of the Rare Diseases Partnership.

---

**The place of the Rare Diseases Partnership in the overall RD landscape**

See slides 163 to 169 “[EJPRD_PB-GB-meeting_July2023-All-slides]”

**Discussion**

The RDP mission is to support the Research and Innovation services from across the Europe in a centralized mode so that every high-quality research project will benefit from cross-disciplinary expertise, goal-oriented study planning and efficient execution.

- Differences were highlighted between the Innovation Health Initiative (IHI) projects and RDP which is a co-funded project.
- The review of the portfolio-management criteria was performed, with a focus on how the RD partnership fits in the current landscape and what are the possibilities for collaboration with existing initiatives (36 programs were pre-identified), but attention should be given to the objectives of the other initiatives to avoid duplication.
- Concerns were raised if the Virtual Platform (VP) development will compete with the European Health Data Space, and how complementarity can be ensured; the reusage of electronic health records in the CRN workstream was also mentioned.
- Portfolio management: concerns were raised that the monitoring activity has the lowest priority, and that the agility and flexibility should be an economic priority, but enough flexibility should be given that priorities might change over time; in this sense, the mechanism in the partnership should be adapted by design to allow switching priorities; linkage with the financial aspects, including partners’ capacity (i.e., centralization of the diagnostic pipeline budget).
- Clear deliverables and milestones are crucial for the evaluation at Year 3, simultaneously a certain level of flexibility should be guaranteed not omitting certain implications that will be possibly associated such as amendments or administration constraints.
Suggestions were presented to adopt a more generic approach for drafting a 7-year planning, but having in place an agile methodology for implementation. The possible involvement of external reviewers might be considered, alongside with having a strategic evaluation process of the “go/no-go” decisions based on the Annual Work Plan, budget prioritization and alignment of activities’ outputs with the scope of the project.

Having an accommodating approach in terms of planning of the 7-year budget is essential to fit the long-term goal, including the budget restrictions that might apply at some time point.

The subcontracting prospects will be managed following the Horizon Europe rules, making certain that is time limited and addresses a certain pre-identified gap, and should not be confounded with the service provision.

**ACTIONS**

- **RDP** should focus on the collaboration with existing initiatives, adopting a synergistic mechanism, and avoiding the duplication of work and objectives
- Special attention should be given to the monitoring activity
- Have clear deliverables and milestones for Year 3 evaluation
- **RDP** should propose a flexible approach that accommodates the substitution of priorities over time, aligned with the strategic evaluation process for the 7-years and budget prioritization
- **RDP coordination** should ensure that subcontracting will be implemented just when it addresses a specific gap that cannot be fulfilled with an existing partner’s expertise and technical capabilities

**AOB**

No other topics discussed.
Annexes

Annex 1 – Slides presented during the EJP RD Executive Committee meeting
- See attached document: “Annex1_20230703_EJPRD_ExCom-Meeting_slides”

Annex 2 – Slides presented during the EJP RD Policy Board and Governing Board meeting
- See attached document: “Annex2_20230704_PB-GB-Meeting_Slides”
EUROPEAN JOINT PROGRAMME ON RARE DISEASES (EJP RD)
Annual Work Plan Year 6

Presentation of planned activities
COORDINATION, TRANSVERSAL ACTIVITIES & COMMUNICATION
WP 1 - Coordination and management

Objectives for Year 6

• day-to-day operational and contractual management
• Organize final Consortium meeting (May/June 2024)
• Deliver the Annual Progress Report for Y5
• Implement the risk management strategy (when necessary)
• Implement the monitoring of the EJP RD activities
• Completion of remaining ongoing IRDiRC activities, including activities developed under Roadmap 2023
• Ensure the continued development and the sustainability of the RD research ecosystem
WP 2 - Integrative research and innovation strategy

Objectives for Year 6

• To collaborate with National Mirror Groups (NMG) or national stakeholders in the RD field (in the absence of constituted NMG) to keep constant dialogue between EJP RD and national RD agendas, adapt the activities of the EJP RD and capture complementary actions enhancing the impact of the EJP RD.

• To develop new National Mirror Groups in EJP RD beneficiary countries
WP 3 - Sustainability strategy and business plan

Objectives for Year 6

• Provide support and feedback on sustainability considerations for all potentially sustainable outputs that might require attention to this respect in Y6, if any.
• To adapt the Sustainability and Business model Plan according to the maturity/evolution of assets/elements, if needed.
• To update and continue the identification of EJP RD sustainable activities
WP 4 - Ethical, regulatory, legal and IPR framework of the EJP RD

WP4 activities aim at providing all Pillars with the proper strategy to address ethical, regulatory, legal and Intellectual Property Right (IPR) issues and at ensuring that relevant rules are complied with within the course of the EJP RD.

Objectives for Year 6

• Giving to project’s partners, upon request, advice on the ethics provisions and regulatory requirements to perform ethically-sounded and regulatory-compliant research and data collection, and to protect patient rights
• Continuing the collaboration with all Pillars requiring ethics and regulatory expertise to perform their activities
• Continuing the collaboration with the Ethics Advisor team
• Performing a second round of the Ethics Follow-up of the EJP RD funded projects
• Continuing the update of ethics and regulatory provisions to all partners
• Continuing the IPR monitoring of Results
• Continuing the IPR support upon requests, including promotion of the establishment of interinstitutional agreements for the management and exploitation of co-owned results among relevant WPs.
WP 5 - Communication & dissemination

Objectives for Year 6

• Launch and promote a comprehensive campaign and platform to disseminate the output, achievements, and impacts of EJP RD, with the aim of reaching a wider audience. This includes leveraging conventional communication tools, establishing a dedicated platform, and utilizing social media platforms.

• Develop and execute a Rare Disease Day campaign to raise awareness, engage stakeholders, and promote understanding of rare diseases.

• With the goal of improving communication and dissemination efforts, IRDiRC has taken a proactive step of creating the IRDiRC Communication Strategy Sub-Committee. This sub-committee is tasked with developing a comprehensive and strategic framework to enhance the communication and dissemination efforts of IRDiRC’s activities. IRDiRC aims to maximize the visibility, utilization, and translation of the outputs generated by its Task Forces, Working Groups and other activities.
PILLAR 1
FUNDING OPPORTUNITIES
Plans for Year 6
WP6 Joint Transnational Calls for collaborative research projects

- Implementation of JTC 2023: Natural History Studies addressing unmet needs in Rare Diseases

  - 80 submitted projects
  - 27 selected projects
  - 21 funders
  - 16 countries

- Review by funders of the lessons learned for all WP6 Calls to prepare future Calls

- Review by Working group on patient engagement in research on lessons learned for all JTC calls to further adapt call documents and procedures for future calls.
WP6 Joint Transnational Calls for collaborative research projects

- Connect funded projects with activities and services Pillars 2-4
- Connect successful projects with WP5 Communication
WP7 Networking Support Scheme

- Finalise the administration and finances of the last networking events taking place at the end of Year 5 and in early Year 6
- Write lessons learned from the Networking Support Scheme for future scheme
- Connect successful events with WP5 Communication
WP8 Rare Diseases Research (RDR) Challenges

In Mid-End Year 5 it is decided after an evaluation process whether all three RDR Challenge projects will start the second phase of 12 months.

In Year 6 the second-phase projects will be followed and final reports will be analysed.

Final distribution of funding to the projects

<table>
<thead>
<tr>
<th>Challenge</th>
<th>Project title (Acronym)</th>
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<tr>
<td>#1 Development of a non-invasive tool for measuring rare disease patient mobility in daily living</td>
<td>Digital tools 4 Rare Diseases (DT4RD)</td>
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<tr>
<td>#2 Delivery system for intranasal administration of biological drugs to neonates</td>
<td>Intranasal device for neonates (INDENEO)</td>
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<tr>
<td>#4 Pre-clinical assay to detect instability of microsatellite repeat expansions</td>
<td>Development and validation of a novel pre-clinical assay to detect triplet repeat expansions (TRXAssay)</td>
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WP9 Monitoring of the results of the funded projects

WP6 – Joint Transnational Calls (E-Rare and EJP RD)

- Continue the monitoring of E-Rare-3 JTC2016, JTC2017 and JTC2018 through annual and final reports
- Update the monitoring tool for the EJP RD JTC2022
- Monitor EJP RD’s co-funded projects under JTC2019 and JTC2020 using annual and final reports
- Monitor EJP RD’s funded projects under the additional call JTC2021 and JTC2022
- Organise midterm monitoring meetings for the JTC2021
WP9 Monitoring of the results of the funded projects

WP7 Networking Support Scheme
Monitor and analyse the networking support scheme funded events

WP8 Rare Diseases Research Challenges
Monitor and follow-up on the 2nd phases of the RDR challenges funded projects
EJP RD Virtual Platform

Plans for Year 6

Ana Rath, Franz Schaefer, and Pillar 2 WP and WF Leaders
Pillar 2 evolution: long learning curve now paying off

2019
Startathons
« Deconfusion »
AR

2020
FAIRathons
Building-blocks
AR
AR

2021
Connectathons
Integration
AR
VP V0

2022
Progressive full-service delivery
VP V1
VP V2
VP V3
VP V4
VP V5
API refactoring

2023

2024
PR

https://vp.ejprarediseases.org
The Virtual Platform: a network of federated resources:

- Registries/biobanks catalogues
- Cell lines/Animal models
- Knowledge bases
- Tools catalogues
- Data deposition & analysis platforms
- Support for clinical/translational research

Decreasing fragmentation
Increasing interoperability
Increasing RD-readiness
through
Harmonisation
Standardisation
in a flexible way:
Common methodologies, Multiple technical solutions

V1: https://vp.ejprarediseases.org
Onboarding at diverse levels of connection to the Virtual Platform

**Level 1**
- Metadata discovery
  - basic description of resource
  - overview of content

**Level 2**
- Discovery of limited parts of datasets
  - output: yes/no, counts, etc..

**Level 3**
- Querying of datasets
  - record-level querying

**Level 4**
- Federated Analysis
  - Answer advanced research questions

---

Onboarding guidance document (ongoing)

Onboarding F2F and follow-up workshops
The EJP-RD Virtual Platform

The Virtual Platform (VP) is a growing network of Findable, Accessible, Interoperable and Reusable (FAIR) resources, ready to serve the rare disease RD research community.

It includes catalogues of resources, registries, biobanks, knowledge bases and tools compliant with agreed standards.

The VP Portal allows you to search the VP network resources at once in real time to find those of interest to your research.

Discover rare diseases resources and data:

Search for a disease name (e.g. ADPKD), gene (e.g. PKD1), or Orphacode (e.g. 730)

Tools and Resources

EXPLORE
Explore Rare Disease resources with the EJP Mind Map

GUIDE
Find out how to make your data more FAIR

CONNECT
Contact us for information or feedback

21 resources connected

11 registries

3 catalogues

2 genome-phenome deposition infrastructures

5 knowledge bases
Final actions to achieve Y5 strategic plan

- Continue increasing visibility of EJP RD available resources through the development of the VP discoverability portal
- Consolidate the technical components of the VP as an integrated architecture, and update the Virtual Platform Specifications (VIPS) accordingly, including specifications for ‘federated analysis on FAIR data’
- Document GDPR compliance, quality assessment and technical sustainability of VP components, and set up a governance for the VP
- Facilitate the onboarding of resources in the VP as a network for improved discoverability and queryability
- Enlarge the VP to other prioritised resources in a federated manner
- Continue expanding the EJP RD FAIRification stewardship programme
- Tackle the secure accessibility and reusability challenge
- Improve and further development of data deposition and analysis facilities
- Demonstrate that the VP specifications for interoperability prepare resources for federated analysis on FAIR data
- Enhance and expand RD pathways creation and analysis based on case studies, and making them findable through the Virtual Platform
Pillar 2 Y6 main goals

- Finalize onboarding of all EJP RD resources at L1 and L2 (for data sources)
  - Final versions of metadata and record-level data models
  - VP compliance assessed resources (including FAIRness)
  - Updated documentation

- Allow for L3 and L4 by
  - Final specifications for communicating with semantic data models for federated analysis on FAIR data
  - Fully implement LifeScience AAI
  - Fully implement data access and reuse conditions specifications
  - Deliver Proof of concept of PPRL interoperability
  - Deliver Proof of concept of federated analysis on FAIR data – demonstrate VP technology is ready

- Reflect on new functionalities in the VP Portal

- Deliver a fully documented VP
  - ViPS
  - Quality/technical sustainability assessed components
  - Documentation in GitHub
WP10- Reinforce the integration among teams to more efficient delivery

38 ST (WP11-12-13) → 14 WF → 9/10 WF
WP10

- Annual retreat to re-adjust plans and prioritization on February
- Finalize GDPR/quality/technical sustainability assessment
- Continue dissemination
  - Training sessions
- Final survey to compare impact vs initial 1019 survey
WP11-12

- Onboard 100% of EJP RD resources
  - In full ViPS compliance

- Update the metadata and record-level data models as needed

- Finalize new query functionalities

- Final version of onboarding guidance document and dissemination material

- Update the Resource MindMap as needed

- Prove VP specs are prepared for federated analysis on FAIR data
WP11 Deposition and analysis

- Finalize last improvements in data deposition resources or last updated in documentation not finished in Y5 (Infrafrontier, BBMRI sample catalogue, RaDiCo)
- Finalize the network GPAP/Infrafrontier/ hPSCreg
- Finalize AAI implementation
- Finalize PPRL POC
- Finalize federated analysis use-cases on multi-omics and on genome interoperability (with WP12-WP13) and provide documentation (lessons-learned, use of VRE…)
- Finalize the provision of capabilities for custom analyses through cloud-based solutions
WP11 - IRDiRC FCC

- Continue updating the analysis platform with FCC input
- Continue training sessions on project deposition and aggregated data analysis

https://rare-research.orphanet.org
WP12

- Finalize the consolidation of model alignment services (semantic mappings, semantic model alignments between extended DCDE/CDE and OMOP, FIHR, C-DISC)

- Implementation of CCE/DUC profiles and ODRL in view of
  - The implementation of a BBMRI-negotiator-like/DUOS-like for the VP
  - The implementation of a « triage agent » (access rights vs use intent)
  - Advising on machine understandable metadata to include in resource metadata to inform agents on the locally applicable responsible* record level analysis policy

- Finalize POC of federated analysis on FAIR-data

- Finalize POC on PPRL interoperability

- Consolidate FAIRification material, including FAIR maturity assessment

- Updating technical documentation including ViPS and technical sustainability plan

* ‘Responsible’ as in ethically and legally responsible use of data, for a definition see https://ec.europa.eu/programmes/horizon2020/en/h2020-section/responsible-research-innovation
WP13

- Finalise scientific papers for publication
- Refine and extend work on AOP and rare disease networks overlap
- Collaborate and support for bringing tools and workflows into the VP
- Collaboration with ELIXIR to create rare disease systems biology service bundle
Needs for dissemination: TBD

- Videos and tutorials for researchers, patients and patient representatives, funders, industry
- Assistance on delivery of impactful posts and news when a new functionality is launched / a new resource is onboarded
- Training sessions / webinars
- A publication in a scientific / bioinformatics journal
### Pillar 3
Trainig and capacity building: objectives

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| **Training on data management & quality**
- Decrease RD data fragmentation and increase data quality through training
- Provide training on data management & quality to increase the level of capacities and help data sharing and networking
- Training activities on standardization of RD data, standards & quality in genetic testing, strategies for undiagnosed RD cases, RD sample data management, RD registries. | **Capacity building and training of patients and researchers**
- Training on therapeutic development & regulatory processes for medicinal products in RDs for patients and researchers
- Providing the knowledge & skills required for patients to become legitimate collaborators in RD research
- Empowering patient representatives as equal, valued, and efficient partners in research
- Provide younger patients with specific knowledge, skills and educational tools on RD research | **Online Academic education course**
- Provide an EU multidisciplinary and transversal online research education course
- Identify needs, audience and topics
- Develop a series of 5 Massive Open Online Courses (MOOCs) in collaboration with KoL and experts
- Monitor the MOOCs and assess their impact. | **ERN RD training and support programme**
- Map and assess the existing landscape of the ERN research training programs
- Develop training programs consisting of crosscutting and overarching research training activities
- Establish an accreditation process | **Development of new trainings**
- Evaluate the state-of-the-art of RD research education and training across several axes
- Assess the impact of EJPRD's special provisions to increase accessibility
- Define the needs and gaps, challenges and opportunities for the further improvement. |
Pillar 3 - Year 6

Training on data management & quality (WP14)

• Organize the fifth edition of the “International course: Training on strategies to foster solutions of undiagnosed rare disease cases” in Rome at ISS originally planned in the AWP Y5 as an extra edition of the course - April 2024 - 4 travel and accommodation fellowships

Capacity building/training of patients (WP15)

• Organize the ExPRESS Expert Patients and Researchers EURORDIS Summer School (Medicine Research and Development) and the EURORDIS School on Scientific Innovation & Translational Research face to face in Spring 2024, preceded by a 4-6 months online intensive programme of pre-training and webinars. Open to 10 researchers. Both cohorts from the two schools will be on-site at the same time, with common days on Day 4 & 5 and the opportunity to exchange at breaks and during the lab visits. To be confirmed in Barcelona, ES.
Pillar 3 - Year 6

Online academic education course (WP16)

- In agreement with WP20’s partners, the MOOC#4 development (Clinical Trials Methodologies) was stopped due to the difficulty in adapting the content to the MOOC format, thus, finally, a total of 4 MOOCs will be developed by the end of the EJP RD programme.

- **EJP RD MOOC1 “Diagnosing Rare Diseases: from research to the clinic and back”** will be run continuously with 2 facilitation windows. New contents will be added (Q4 2023), on AI and Omics approaches.

- **EJP RD MOOC2 “Innovative Therapies and Personalized Medicine: new keys for the treatment of rare diseases”** will be launched in autumn/winter 2023. It will be run continuously, as per the “on demand” run model.

- **EJP RD MOOC3 “From Lab to clinic: translational research for rare diseases”** will be run continuously, as per the “on demand” run model and, as for the other MOOCs, two facilitation window per year will be guaranteed. If necessary, based on the results of the impact assessment of the previous run, further modifications will be made.

- **EJP RD MOOC5 “Rare disease data for research purposes: ethics and regulatory considerations”** will be launched in Q1 of 2024. Once launched, the MOOC will be run continuously.

- All EJP RD MOOCs will continue to be developed with the Creative Commons copyright CC BY-NC-SA.
- All EJP RD MOOCs include several testimonies from clinicians, researchers, patients and patients’ representatives. Whenever required, anonymization will be ensured in order to answer patients’ wishes to remain unidentified from work, family, or for other personal reasons.

- Follow up of the quantitative and qualitative analysis.
- Continuous monitoring & adaptation of the existing MOOCs.
Pillar 3 – Year 6

ERN RD training and support programme (WP17)

WORKSHOPS:

• Processing of the last reimbursements and final reports of the 6th call for workshops, being conducted in the second half of year 5, will be completed in Q2 of year 6.
• Finalisation and evaluation of all calls for ERN research workshops will be performed from M61 to M68.

FELLOWSHIPS:

• The last cohort of fellows will finalize their research stays.
  • Surveys: will serve to assess the effectiveness of the programme.
  • Reports: the fellows will also be requested to submit reports. Two types of reports are solicited: short-term reports, due within 1-3 months after the completion of the research stay, and long-term reports, expected approximately one year after the conclusion. These reports allow the fellows to share their research findings and provide valuable reflections on the impact of their research activities.
• During this final year, the necessary settlements will be made to ensure the smooth and efficient closure of the programme.
  • A comprehensive compilation of statistics will be conducted, covering all calls.
  • These statistics will provide a comprehensive overview and serve as a valuable resource for future assessment analysis.
Pillar 3 – Year 6

Development and adaptation of training activities (WP18)

- Development of the new training modules (M60-62) - Task leader: VUHSK; Participants: INSERM, UKL-HD, LUMC, EMBL-EBI, BBMRI, ULEIC, AMC.
  - The Organizational Committee will finalize the development of the two courses, including development of a training programme, contents and materials.
- Organization of the new training activities (M62-64) - Task leader: VUHSK; Participants: INSERM, UKL-HD, LUMC, EMBL-EBI, BBMRI, ULEIC, AMC.
  - In collaboration with EJP RD Coordination, the further activities will be accomplished: dissemination of information on the new courses targeted at the EJP RD Virtual Platform contributors and users; preparation and launch of the calls for the courses, selection and registration of course participants (M60-63).
- Task 18.8: Delivering the new training activities (M62 and M64) - Task leader: VUHSK, Participants: INSERM, UKL-HD, LUMC, EMBL-EBI, BBMRI, ULEIC, AMC.
  - The new training activities will be delivered to the training participants: the course on EJP RD Virtual Platform for the VP contributors (M62) and the course on EJP RD Virtual Platform for VP users (M64).
ACCELERATED TRANSLATION OF RESEARCH RESULTS & CLINICAL TRIALS
WP19: Activities foreseen for Year 6

**IMT - ODDG**
- Finalize Use cases in progress
- Maintenance of the IMT and ODDG will be undertaken
- Engagement with key stakeholders to ensure coherent long term innovation funding and investments
- KPI’s analysis and dissemination actions to increase awareness

**Mentoring services**
- WP19 partners will continue providing support for the projects funded by the JTC calls including the ongoing JTC2022;
- Support new projects for mentoring even they are outside of the EU funded programs.
- Comprehensive dissemination will be undertaken by identifying and directly contacting additional researchers and organizations working on rare diseases to offer the developed services.
Planned Pillar 4/WP 20 activities in AWP Y6

• Close monitoring of the progress and completion of the funded projects.

• Pursue disseminating the progress of the projects through scientific articles, meetings, and presentations at scientific conferences.

• Foster the clinical study support office work: evaluate and support RD projects focusing on methodological/study design requests and operational support advice enhancing the quality and number of RD clinical trials and projects/collaborations for clinical research on RD. (Collab ERN, ERICA)

• Refining the Clinical Trial toolbox, ensuring its implementation in the virtual platform and improving the performance of academic-sponsored clinical trials for rare diseases
Planned Pillar 4/WP 20 activities in AWP Y6

• Foster **external collaborations** (ERICA, EMA, OJRD).

• Provide additional **advanced courses** and develop **intermediate courses** on Clinical Trials Methodologies adapted to rare diseases clinical trials stakeholders.

• Assisting with transversal activities across the demonstration and innovation projects.

• Provide additional Intermediate courses (Webinar November 2023, April 2024)

• **Organize the final WP20 meeting** to present the results of the different demonstration and innovation projects and provide an intensive training course on CT methodologies. (June 2024)
Annual Work Plan Year 6

Budget and Prioritisation
Communication strategy for the last year of EJP RD
Communication Strategy

Last year of EJP RD
Objectives and strategy

Communication & Dissemination on the results & impact of EJP RD

Reach the missing targeted audience in addition to the current reached audience

Rare Disease Day campaign
Current status reached stakeholders

(Initially targeted)
Standard communication actions per campaigns

- Social Media Message
- Email to targeted audience
- Newsletter article
- Website news & slider (when appropriate)

**Visual**

**Addition in Year 6: Sponsored Targeted Posts!**
<table>
<thead>
<tr>
<th>WP</th>
<th>Action/Activity</th>
<th>Channel</th>
<th>Timeline</th>
<th>Target Audience</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Internal news dedicated to EJP RD Partners</td>
<td>Internal Newsletter</td>
<td>Every other month</td>
<td>EJP RD Partners</td>
</tr>
<tr>
<td>1</td>
<td>MsTeams Posts</td>
<td>MsTeams</td>
<td>Continuous</td>
<td>EJP RD Partners</td>
</tr>
<tr>
<td>1</td>
<td>Helpdesk</td>
<td>Website</td>
<td>Continuous</td>
<td>All Stakeholders</td>
</tr>
</tbody>
</table>
| 2  | Medium and long-term strategy - Task Forces: creation of IRDiRC Communication Subcommittee to expand the outreach of IRDiRC and make the website more user-friendly | Website     | January 2024 | • Research and health policy makers  
|    |                                                                                 |             |              | • Research funders  
|    |                                                                                 |             |              | • Industry  
|    |                                                                                 |             |              | • Patient(s)/Organisations  
|    |                                                                                 |             |              | • RD researchers  
|    |                                                                                 |             |              | • Healthcare providers            |
## Coordination & Management

<table>
<thead>
<tr>
<th>WP</th>
<th>Action/Activity</th>
<th>Channel</th>
<th>Timeline</th>
<th>Target Audience</th>
</tr>
</thead>
</table>
| 3  | Sustainability roadmap: Deliverable D3.5 (Second Proposal of structure, governance and financial model and global sustainability roadmap) | Deliverable (Website) | 30 June 2023  | • Policy makers  
• Ministries  
• Program owners and managers  
• European Commission  
• SMEs and industry  
• Foundations and learned societies  
• Patients’ organizations |
| 4  | Management of EJP RD ethics, regulatory & legal issues: Communication of news related to ethics, regulatory & legal issues | Internal Newsletter | Bi-Monthly     | • EJP RD partners  
• RD stakeholders at large  
• Regulatory agencies |
|    |                                                                                | Upload documents in the AREB Section on the Website | Continuous     |                                                               |
### Coordination & Management

<table>
<thead>
<tr>
<th>WP</th>
<th>Action/Activity</th>
<th>Channel</th>
<th>Timeline</th>
<th>Target Audience</th>
</tr>
</thead>
<tbody>
<tr>
<td>5</td>
<td>Creation of an interactive platform to showcase EJP RD outputs, impacts, and tools produced</td>
<td>Website</td>
<td>October/November 2023</td>
<td>RD community at large General public</td>
</tr>
<tr>
<td>5</td>
<td>Rare Disease Day Campaign</td>
<td>Video</td>
<td>28 February 2024</td>
<td></td>
</tr>
</tbody>
</table>
Fundings and Calls

<table>
<thead>
<tr>
<th>WP</th>
<th>Action/Activity</th>
<th>Channel</th>
<th>Timeline</th>
<th>Target Audience</th>
</tr>
</thead>
<tbody>
<tr>
<td>6 - 7 - 8 - 9</td>
<td>Journalistic report for selected projects written in lay language for general public</td>
<td>Website + Standard communication procedure</td>
<td>July 2023 - August 2024</td>
<td>All stakeholders</td>
</tr>
</tbody>
</table>
# Coordinated Access to Data and Services

<table>
<thead>
<tr>
<th>WP</th>
<th>Action/Activity</th>
<th>Channel</th>
<th>Timeline</th>
<th>Target Audience</th>
</tr>
</thead>
</table>
| 10 | Launch of the Virtual Platform: Communication Campaign | Standard communication procedure + Additional email personalized for target audience not reached (Patients, Funders, Industry, SMEs) | July 2023 then continuous communication | • RD researchers and clinicians (including P1 funded projects)  
• ERNs and national reference centers/hospitals  
• Patients  
• Policy makers/ministries  
• Funders  
• Industry (EFPIA)  
• SMEs (EUCOPE) |
| 11 | RD analysis tools and data sharing capabilities | Open Access Publications | Continuous | • RD researchers and clinicians (including P1 funded projects)  
• Systems Biologists and Data scientists ERNs and national reference centers/hospitals  
• Policy makers  
• Funders  
• Industry (including pharma) |
# PILLAR 2: Coordinated Access to Data and Services

<table>
<thead>
<tr>
<th>WP</th>
<th>Action/Activity</th>
<th>Channel</th>
<th>Timeline</th>
<th>Target Audience</th>
</tr>
</thead>
<tbody>
<tr>
<td>12</td>
<td>Organisation of data stewarding collaborations, advocacy, and self-help networks for practical FAIRification support</td>
<td>Open Access Publications</td>
<td>Continuous</td>
<td>RD researchers and clinicians, Research institutes and hospitals, Infrastructures, Funders, Policy makers, Industry</td>
</tr>
<tr>
<td>13</td>
<td>Innovative holistic approaches for rare diseases diagnosis and therapeutics</td>
<td>Event Participation</td>
<td>Continuous</td>
<td>RD researchers and clinicians, Research institutes and hospitals, Infrastructures, Funders, Policy makers, Industry, SMEs (EUCOPE)</td>
</tr>
<tr>
<td></td>
<td>P2 in general Videos and tutorial for researchers, patients, patient representatives, funders, industry</td>
<td>Video</td>
<td>Continuous</td>
<td>Researchers, Patients/Patients Representatives, Funders, Industry</td>
</tr>
</tbody>
</table>
## Training and Empowerment

<table>
<thead>
<tr>
<th>WP</th>
<th>Action/Activity</th>
<th>Channel</th>
<th>Timeline</th>
<th>Target Audience</th>
</tr>
</thead>
<tbody>
<tr>
<td>14 - 15 - 16 - 17 - 18</td>
<td>Communication campaign for trainings</td>
<td>Standard communication procedure</td>
<td>Continuous</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
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<tr>
<td></td>
<td>Joint campaign with EURORDIS dedicated to patients/patients advocacy groups</td>
<td>Continuous</td>
<td></td>
<td></td>
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<tr>
<td></td>
<td></td>
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<tr>
<td></td>
<td>Joint campaign with EURORDIS dedicated to patients/patients advocacy groups</td>
<td>Continuous</td>
<td></td>
<td>Young Public (12-18)</td>
</tr>
</tbody>
</table>
## Training and Empowerment

<table>
<thead>
<tr>
<th>WP</th>
<th>Action/Activity</th>
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<th>Timeline</th>
<th>Target Audience</th>
</tr>
</thead>
</table>
| 16 | Campaign on new MOOCs                                                           | Standard communication procedure | 2024     | • Academic clinicians and researchers  
|    |                                                                                  |                                |          | • Medical & biomedical students  
|    |                                                                                  |                                |          | • Industry researchers/scientists  
|    |                                                                                  |                                |          | • RD patient representatives and advocates  
|    |                                                                                  |                                |          | • Industry  
|    |                                                                                  |                                |          | • Lecturers  
|    |                                                                                  |                                |          | • University managers  
|    |                                                                                  |                                |          | • Professional organizations  
<p>|    |                                                                                  |                                |          | • Organizations of academic institutions |
| 17 | Publication of the fellows 2023 on EJP RD website with their presentation       | Website                        | Continuous | Academic clinicians and researchers involved in ERNs |</p>
<table>
<thead>
<tr>
<th>WP</th>
<th>Action/Activity</th>
<th>Channel</th>
<th>Timeline</th>
<th>Target Audience</th>
</tr>
</thead>
</table>
| 19 | Testimony Campaign for Mentoring                        | Website + Youtube | April 2024 | • Researchers with a potential product  
• E-Rare & Pillar 1 funded projects                                                  |
| 19 | Testimony Campaign for Follow-on Funding Support        | Website + Youtube | April 2024 | • Researchers with an evaluated product  
• Investors (industry, venture capitals, charities, etc.)                              |
| 20 | Publication of Innovation Management Toolbox (IMT) use cases on Youtube channel | Youtube    | Continuous | • RD research community  
• ERNs  
• RD research community  
• Institutional innovation managers                                                  |
| 20 | Testimony Campaign for Clinical Studies Support Office (Support Office & Clinical Trial Methodology) | Website + Youtube | April 2024 | • Researchers with a potential product  
• Clinicians with a potential project of a clinical study                             |
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 Nº825575
EUROPEAN JOINT PROGRAMME ON RARE DISEASES (EJP RD)

Policy Board and Governing Board meeting

PB-GB meeting
04/07/2023, Bruxelles - Belgium
Welcome from Coordination
EJP RD sustainability
STRATEGY AND BUSINESS PLAN
Ensuring the viability of EJP RD’s outputs in a future commercial setting

European Joint Programming on Rare Diseases (EJP RD)
June 2023
The EJP RD outputs may be lost post 2024 and a financially sustainable solution is needed

**Partners**

130 institutions (including all 24 ERNs) from 35 countries

**The governance structure**

Research project under Horizon2020

**Organization**

5 pillars and 20 Work Packages

**The budget**

€101 million
55% comes Horizon 2020

**Time horizon**

December 2018 - December 2023
Extended to December 2024
A sustainable solution – the Rare Disease Hub

Problems

- Scattered and disconnected biology and patient data resources.
- Uncertainty in the regulatory environment due to limited clinical evidence.
- Small populations in clinical trials.

Solution

- Data Hub
- Capacity Building Hub
- Expertise Hub
The RD Data Hub

Key features:
- Fully developed, secure virtual platform
- Clear overview of available resources, data and analytical tools
- Service level agreements facilitate data sharing and access
- Incentives in place promote data sharing
- ERNs use the data hub for sharing the patient data

Resources
- Rare disease catalogues
- Animal models and cell lines
- Care pathways
- other

Data
- Genomics/multi-omic data
- Patients’ clinical and biosamples data registers

Analytical tools
- Genomics/multi-omic analysis
- AI / Machine Learning (ML)

Source: Illustration by Copenhagen Economics
The RD Capacity Building Hub

Massive Open Online Courses - MOOC

Topics: Diagnosing rare diseases: from clinic to research and back. Introduction to Translational Research for Rare Diseases
Target audience: Research community, patients and patient representatives and doctors
Experts teaching the course: Research community and ERNs

Support for patient and their representatives

Topics: Training on scientific innovation and translational research aspects in rare diseases. Training for patient representatives and advocates on leadership and communication skills. Educational materials and activities for pediatric patients.
Target audience: Patient representatives and advocates
Experts teaching the course: EURORDIS

Clinical trials in small populations

Target audience: Organizations conducting clinical trials, EMA, the EU and national HTA bodies.
Experts teaching the course: Research community

Data sharing & use

Topics: Introduction to FAIR principles, data management and organization, data access and sharing, governance, privacy and security.
Target audience: Organizations collecting the data, e.g. ERN, biobanks, patient’s organizations
Experts teaching the course: RD Hub

Key features:
- Active and targeted outreach
- Regular updates of training content
- Business oriented interface & delivery

Source: Illustration by Copenhagen Economics
The RD Expertise Hub

**Clinical evidence for rare disease treatments**
Support to EMA, EU HTA, RD research consortia and clinical trial sponsors on the data collection, study design and methodology. It develops best practices for collecting and developing evidence for ultra rare diseases.

**Ethical support**
Experts support the design and execution of clinical trials e.g. advice on the study protocol, assess the potential risks and benefits to participants.

**Legal services**
Supports contracting between providers of omic-data and patient registers and the users of the data.

**FAIRification stewardship**
As part of FAIRification, the RD expertise hub supports the process of implementing and overseeing the adoption of FAIR principles in data processing.

Source: Illustration by Copenhagen Economics
Why is the RD Hub different from other existing solutions?

- **Trustful community**
  - MOOC
  - Ethics

- **Capacity Building Hub**
  - Patient Support
  - Resources

- **Quality driven by excellence**
  - Clinical trials/evidence
  - Data FAIRification

- **Expertise Hub**
  - Analytical tools
  - Data

- **Significant public investment**

Source: Illustration by Copenhagen Economics
Potential customers

Table: Types and ranking of organisations sponsoring clinical trials in Europe
Ranking by the number of ongoing and planned clinical trials

<table>
<thead>
<tr>
<th>Private companies, Top 10</th>
<th>Publicly listed companies, Top 10</th>
</tr>
</thead>
<tbody>
<tr>
<td>1. Celgene Group</td>
<td>1. F. Hoffmann-La Roche Ltd</td>
</tr>
<tr>
<td>2. PPD Inc.</td>
<td>2. Merck &amp; Co Inc</td>
</tr>
<tr>
<td>3. Parexel International Corp</td>
<td>3. AstraZeneca Plc</td>
</tr>
<tr>
<td>4. Labcorp Drug Development</td>
<td>4. Bristol-Myers Squibb Co</td>
</tr>
<tr>
<td>5. Boehringer Ingelheim International GmbH</td>
<td>5. Johnson &amp; Johnson</td>
</tr>
<tr>
<td>7. PRA Health Sciences Inc</td>
<td>7. Pfizer Inc</td>
</tr>
<tr>
<td>8. Alexion Pharmaceuticals Inc</td>
<td>8. Sanofi</td>
</tr>
<tr>
<td>10. Novotech Australia Pty Ltd</td>
<td>10. IQVIA Holdings Inc</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Government, Top 10</th>
<th>Institutions, Top 10</th>
</tr>
</thead>
<tbody>
<tr>
<td>1. Ministry of Health, France</td>
<td>1. Sarah Cannon Research Institute LLC, USA</td>
</tr>
<tr>
<td>2. Medical Research Council, UK</td>
<td>2. Assistance Publique – Hopitaux de Paris, France</td>
</tr>
<tr>
<td>3. German Research Foundation, Germany</td>
<td>3. Cancer Research UK, UK</td>
</tr>
<tr>
<td>4. Federal Ministry of Education and Research, Germany</td>
<td>4. Erasmus MC, Netherlands</td>
</tr>
<tr>
<td>5. European Commission</td>
<td>5. University College London, UK</td>
</tr>
<tr>
<td>7. The Swedish Research Council, Sweden</td>
<td>7. Rigshospitalet, Denmark</td>
</tr>
<tr>
<td>8. Region Skåne, Sweden</td>
<td>8. Aarhus University Hospital, Denmark</td>
</tr>
<tr>
<td>9. Directorate General of Health Care Provision, France</td>
<td>9. Oslo University Hospital, Norway</td>
</tr>
<tr>
<td>10. Vastra Gotalandsregionen, Sweden</td>
<td>10. KWF Kankerbestrijding, Netherlands</td>
</tr>
</tbody>
</table>

Source: Copenhagen Economics based on GlobalData.
Closing the gap between status quo and the vision

Completeness of EJP RD outputs

Status quo and the vision

The RD Hub

Status quo

The gap

The solutions

Foster a customer-oriented mindset

Establish a sustainable governance structure and legal entity

Accelerate the development of the Virtual Platform

Source: Copenhagen Economics
# Timeline for the path to sustainability in 2023 and 2024

<table>
<thead>
<tr>
<th>Activity</th>
<th>2023</th>
<th>2024</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Sep</td>
<td>Oct</td>
</tr>
<tr>
<td>Enhance data architecture and comprehensive data integration</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Develop incentives for data sharing and access</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Develop service level agreements (SLA) and contract models</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Decide on a legal entity and governance structure</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Design efficient decision-making processes</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Test prototypes for different levels of data access</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Make your presence known</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Fully fledged business plan including the operation costs</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Communication strategy to gain support</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Gain political support for the RD Hub</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

01-06-2024
STRATEGY AND BUSINESS PLAN
Ensuring the viability of EJP RD’s outputs in a commercial setting

AUTHORS
Christian Jervelund
Malwina Mejer, PhD
Lærke Kilsdal
Pillars’ contributions towards objectives of EJP RD

Summary of achievements and final activities for Year 6
COORDINATION, TRANSVERSAL ACTIVITIES & COMMUNICATION
## Pillar 0 – Expected impacts

### Overall impact: Improved alignment of national/regional activities and policies in RD

- efficiently transcribe EJP RD activities and outcomes at regional, national, EU and international levels
- each country will be urged to put in place National Mirror Group (NMG) bringing national partners participating directly in the EJP RD and additional actors of the RD field.

### Specific impact 2: Decrease fragmentation of rare diseases expertise and research resources

- Through the participation of 87 partners including policy makers, funders, research organizations, ERNs, infrastructures and patients from 32 countries, EJP RD programme is unique in achieving centralised critical mass of expertise and research resources from Europe and beyond.

### Specific impact 6: Follow the policies and contribute to the objectives of the International Rare Diseases Research Consortium (IRDiRC)

- The organization of the EJP RD consortium reflects well the organization of IRDiRC by bringing under single umbrella funders, research performing institutions, healthcare providers, patients and linking to industry. In addition, the majority of EJP RD partners are also members of IRDiRC. This will not only allow but also accelerate the follow up of IRDiRC policies and contribution to its objectives. Furthermore, constant connection to IRDiRC will be maintained by the integration of IRDiRC chair and vice chair in the EJP RD Policy Board, incorporation of IRDiRC Scientific Secretariat in the EJP RD Coordination Office, implementation of joint Task Forces and participation of rare diseases experts and other EJP RD members in IRDiRC scientific and constituent committees.

### Contribution to all other Specific Impacts

- The contribution of the EJP RD to the improvement of lives of RD patients by providing new and optimized treatment options and diagnostic tools will be achieved as a sum of efforts provided within different pillars and transversal activities strengthened by the central coordination and close linkage with relevant policy stakeholders to translate these efforts at regional, national and EU levels.
Pillar 0 – Achievements at M55

Overall impact: **Improved alignment of national/regional activities and policies in RD**

- efficiently transcribe EJP RD activities and outcomes at regional, national, EU and international levels
- each country will be urged to put in place National Mirror Group (NMG) bringing national partners participating directly in the EJP RD and additional actors of the RD field.

• **Increased awareness of the rare diseases research ecosystem** – EJP RD is featured on websites of national and regional funding bodies, research institutions, all ERNs and patients’ organisations (e.g. 19,600 results on google for "European joint programme on Rare Diseases")

• **Initiation and/or empowerment of National Mirror Groups** bringing all RD stakeholders (e.g., creation of NMG in the Netherlands, Poland, UK and Portugal, full alignment of actions between National Plan for Rare Diseases and EJP RD in France). The preparation of the Rare Diseases Partnership allowed Coo Team to identify new people to be part of the NMGs to be built. ➔ not all EJP RD countries have a NMGs yet: work will continue

• **Alignment with national strategies is now visible**: e.g., in France the EJP RD work, notably in relation to implementation of federated Virtual Platform, standards, ontologies and methods used, is indicated as mandatory for the alignment of national resources (newly created or to be updated rare diseases registries and/or databases), cohorts and health data hub that will host RD data.

• **Between 23 and 86.6% of national activities are aligned or complementary to EJP RD** actions (23% for P4 innovative methodologies in CTs and 86% for support of data repositories and tools)
Pillar 0 – Achievements at M55

Specific impact 2: Decrease fragmentation of rare diseases expertise and research resources

- Through the participation of 87 partners including policy makers, funders, research organizations, ERNs, infrastructures and patients from 32 countries, EJP RD programme is unique in achieving centralised critical mass of expertise and research resources from Europe and beyond.

EJP RD Helpdesk

- over 450 experts in the current database
- Expansion to other resources (paediatric, regulatory expertise from other networks)
### Pillar 0 – Achievements at M55

**Specific impact 6: Follow the policies and contribute to the objectives of the International Rare Diseases Research Consortium (IRDiRC)**

- The organization of the EJP RD consortium reflects well the organization of IRDiRC by bringing under single umbrella funders, research performing institutions, healthcare providers, patients and linking to industry. In addition, some of the EJP RD partners are also members of IRDiRC. This will not only allow but also accelerate the follow up of IRDiRC policies and contribution to its objectives. Furthermore, constant connection to IRDiRC is maintained by the integration of IRDiRC chair and vice chair in the EJP RD Policy Board, incorporation of IRDiRC Scientific Secretariat in the EJP RD Coordination Office, implementation of joint Task Forces and participation of rare diseases experts and other EJP RD members in IRDiRC scientific and constituent committees, organization of joint events.

### Consortium Assembly

| 11 FCC members | 1 PACC member |

### Scientific Committees

| 12 experts from EJP RD beneficiary institutions involved in IRDiRC Scientific Committees |

### Joint Action

- Machine Readable Consent and Use Conditions Task Force

### Task Forces

- 30 experts from EJP RD beneficiary institutions serving in IRDiRC Task Forces/Working Groups

### Topic Identification

- IRDiRC experts advising on possible topics in EJP RD calls
- IRDiRC Chairs as part of the EJP RD Policy Board
Pillar 0 – Achievements at M55

Contribution to all other Specific Impacts

• The contribution of the EJP RD to the improvement of lives of RD patients by providing new and optimized treatment options and diagnostic tools will be achieved as a sum of efforts provided within different pillars and transversal activities strengthened by the central coordination and close linkage with relevant policy stakeholders to translate these efforts at regional, national and EU levels.

Coordination across all Pillars, Work Packages and external stakeholders:

- 78 Operating Group meeting
- 24 Executive Committee meetings
- 5 Policy Board meetings
- 4 General Assembly and consortium meetings
- 2 Strategic meetings on National alignment
- 25 Advisory, Regulatory and Ethics Board meetings
- 5 (6) Annual Work Plans
- 4 Technical reports & 3 review meetings
- 8 established collaborations with EU and international stakeholders
Objectives for Year 6

- day-to-day operational and contractual management
- Organize final Consortium meeting (May/June 2024)
- Deliver the Annual Progress Report for Y5
- Implement the risk management strategy (when necessary)
- Implement the monitoring of the EJP RD activities
- Completion of remaining ongoing IRDiRC activities, including activities developed under Roadmap 2023
- Ensure the continued development and the sustainability of the RD research ecosystem
AWP Y6: WP 2 - Integrative research and innovation strategy

Objectives for Year 6

• To collaborate with National Mirror Groups (NMG) or national stakeholders in the RD field (in the absence of constituted NMG) to keep constant dialogue between EJP RD and national RD agendas, adapt the activities of the EJP RD and capture complementary actions enhancing the impact of the EJP RD.

• To develop new National Mirror Groups in EJP RD beneficiary countries
AWP Y6: WP 3 - Sustainability strategy and business plan

Objectives for Year 6

- Provide support and feedback on sustainability considerations for all potentially sustainable outputs that might require attention to this respect in Y6, if any.
- To adapt the Sustainability and Business model Plan according to the maturity/evolution of assets/elements, if needed.
- To update and continue the identification of EJP RD sustainable activities
WP4 activities aim at providing all Pillars with the proper strategy to address ethical, regulatory, legal and Intellectual Property Right (IPR) issues and at ensuring that relevant rules are complied with within the course of the EJP RD.

Objectives for Year 6

- Giving to project’s partners, upon request, advice on the ethics provisions and regulatory requirements to perform ethically-sounded and regulatory-compliant research and data collection, and to protect patient rights
- Continuing the collaboration with all Pillars requiring ethics and regulatory expertise to perform their activities
- Continuing the collaboration with the Ethics Advisor team
- Performing a second round of the Ethics Follow-up of the EJP RD funded projects
- Continuing the update of ethics and regulatory provisions to all partners
- Continuing the IPR monitoring of Results
- Continuing the IPR support upon requests, including promotion of the establishment of interinstitutional agreements for the management and exploitation of co-owned results among relevant WPs.
Objectives for Year 6

- Launch and promote a comprehensive campaign and platform to disseminate the output, achievements, and impacts of EJP RD, with the aim of reaching a wider audience. This includes leveraging conventional communication tools, establishing a dedicated platform, and utilizing social media platforms.

- Develop and execute a Rare Disease Day campaign to raise awareness, engage stakeholders, and promote understanding of rare diseases.

- With the goal of improving communication and dissemination efforts, IRDiRC has taken a proactive step of creating the IRDiRC Communication Strategy Sub-Committee. This sub-committee is tasked with developing a comprehensive and strategic framework to enhance the communication and dissemination efforts of IRDiRC’s activities. IRDiRC aims to maximize the visibility, utilization, and translation of the outputs generated by its Task Forces, Working Groups and other activities.
PILLAR 1
FUNDING OPPORTUNITIES
Impacts and Plans for Year 6
<table>
<thead>
<tr>
<th>Specific impact 1: Improve lives of rare disease patients by providing new and optimised treatment options and diagnostic tools for these diseases</th>
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Pillar 1 – Expected impacts

Specific impact 5: Reinforce the EU’s role as a global leader for rare diseases

• The joint effort will leverage national funding resources from 22 countries estimated at more than 60M € for 5 years.
• Variety of proposed funding schemes fostering transnational research, Networks and Rare Disease Challenges essential to enhance excellent research

Specific impact 6: Follow the policies and contribute to the objectives of the International Rare Diseases Research Consortium (IRDiRC)

• The support to IRDiRC policies and objectives is translated through introduction of IRDiRC recommendations in guidelines for researchers as well as implementation of joint transnational calls targeting topics identified by IRDiRC as of most importance.
Pillar 1 – Achievements at M55

Specific impact 1: Improve lives of rare disease patients by providing new and optimised treatment options and diagnostic tools for these diseases

- Financing of well-organized, coordinated science that includes basic, translational, clinical, social and health economic research that will develop new and optimised treatment options and diagnostic tools

**JTC 2019: Research to accelerate diagnosis and/or explore disease progression and mechanisms of rare diseases**
- 31 funders
- 220 submitted projects
- 52 selected projects
- 22 funded projects
- 30.5 million € spent in total
  - Including 6 Mio. € from European Commission

**JTC 2020: Pre-clinical research to develop effective therapies for rare diseases**
- 29 funders
- 173 submitted projects
- 30 selected projects
- 18 funded projects
- 24.5 million € spent in total
  - Including 2.7 Mio. € from European Commission

**JTC 2021: Social sciences and Humanities Research to improve health care implementation and everyday life of people living with a rare disease**
- 19 funders
- 37 submitted projects
- 22 selected projects
- 12 funded projects
- 11.5 million € spent in total
  - Including 0.4 Mio. € from European Commission

**JTC 2022: Development of new analytic tools and pathways to accelerate diagnosis and facilitate diagnostic monitoring of rare diseases**
- 27 funders
- 101 submitted projects
- 21 selected projects
- 12 funded projects
- 17 million € spent in total
Pillar 1 – Achievements at M55

Specific impact 2: Decrease fragmentation of rare diseases expertise and research resources

- Pooling of resources in funded research projects
- Integration of higher number of funding agencies in JTC funding

- Funding of 64 research projects with 432 research groups with 83.5 Mio. €
- Start with 31 funders from 23 countries, one additional funder from Australia (MRFF) and one from Italy (Telethon Italy) recruited
Pillar 1 – Achievements at M55

Specific impact 3: Increase the EU's capacity to innovate in the field of rare diseases

- All funding activities foster the increase of research and knowledge capacity for rare diseases in Europe
- Rare Disease Challenge funding scheme aims at removing general obstacles for innovation in the field of therapeutic research by bringing together public and private stakeholders

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<tr>
<th>Challenge</th>
<th>Project title (Acronym)</th>
<th>Lead applicant</th>
<th>Nº of partners involved</th>
<th>Nº of countries involved</th>
<th>Industry sponsors</th>
<th>Total funding</th>
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<td>SME: UK</td>
<td>5 (2 SME + 2 Academia + 1 PAO)</td>
<td>3 (Netherlands: France; UK)</td>
<td>Chiesi and CSL Behring</td>
<td>575 000 €</td>
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<td>#2 Delivery system for intranasal administration of biological drugs to neonates</td>
<td>Intranasal device for neonates (INDENEO)</td>
<td>SME: France</td>
<td>3 (1 SME + 2 Academia)</td>
<td>2 (France: Belgium)</td>
<td>Chiesi</td>
<td>487 500 €</td>
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<td>#4 Pre-clinical assay to detect instability of microsatellite repeat expansions</td>
<td>Development and validation of a novel pre-clinical assay to detect triplet repeat expansions (TRXAssay)</td>
<td>Academia: Ireland</td>
<td>3 (Academia)</td>
<td>2 (Ireland: UK)</td>
<td>LoQus23</td>
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Pillar 1 – Achievements at M55

Specific impact 4: Improve healthcare systems' capacity to take up research results

- The funding of socio-economic and health care/health services related projects will facilitate a better understanding of the real life problems and possible solutions for better uptake of research results in the general practice
- Exchange of knowledge facilitated through the Networking schemes will lead to an increased uptake of research results by providing the space to share diagnostic practices and guidelines and strengthen the collaborations by different stakeholders

- Funding of 12 research projects in social and human sciences with 11.5 Mio. €

- NSS results
  - Knowledge transfer
  - Formation of new collaborations between different stakeholders
  - Formation of new dedicated working groups
  - Setting up new goals and work plans

NSS ROUNDS 1-9

- Number of Applicants: 349
- Number of Countries: 33
- Underrepresented Countries: 15
- NSS Budget | Co-funding: ~1.24M | 0.8M
- Topic: Rare Diseases | Rare Cancers: 42 | 13
Pillar 1 – Achievements at M55

Specific impact 5: Reinforce the EU's role as a global leader for rare diseases

- The joint effort will leverage national funding resources from 22 countries estimated at more than 60M € for 5 years.
- Variety of proposed funding schemes fostering transnational research, Networks and Rare Disease Challenges essential to enhance excellent research

Specific impact 6: Follow the policies and contribute to the objectives of the International Rare Diseases Research Consortium (IRDiRC)

- The support to IRDiRC policies and objectives is translated through introduction of IRDiRC recommendations in guidelines for researchers as well as implementation of joint transnational calls targeting topics identified by IRDiRC as of most importance.

- All funded research activities have contributed to reinforcing EU leadership role and fostering IRDiRC objectives

- IRDiRC experts and resources were instrumental in shaping research topics for joint transnational calls
WP6 Joint Transnational Calls for collaborative research projects

- Implementation of JTC 2023: Natural History Studies addressing unmet needs in Rare Diseases
  - 80 Submitted projects
  - 27 Selected projects
  - tbd
  - 21 funders
  - 16 countries

- Review by funders of the lessons learned for all WP6 Calls to prepare future Calls

- Review by Working group on patient engagement in research on lessons learned for all JTC calls to further adapt call documents and procedures for future calls.
WP6 Joint Transnational Calls for collaborative research projects

- Connect funded projects with activities and services Pillars 2-4
- Connect successful projects with WP5 Communication
WP7 Networking Support Scheme

- Finalise the administration and finances of the last networking events taking place at the end of Year 5 and in early Year 6
- Write lessons learned from the Networking Support Scheme for future scheme
- Connect successful events with WP5 Communication
In Mid-End Year 5 it is decided after an evaluation process whether all three RDR Challenge projects will start the second phase of 12 months.

In Year 6 the second-phase projects will be followed and final reports will be analysed.

Final distribution of funding to the projects.

**WP8 Rare Diseases Research (RDR) Challenges**

**Public-private projects**

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WP9 Monitoring of the results of the funded projects

WP6 – Joint Transnational Calls (E-Rare and EJP RD)

- Continue the monitoring of E-Rare-3 JTC2016, JTC2017 and JTC2018 through annual and final reports
- Update the monitoring tool for the EJP RD JTC2022
- Monitor EJP RD’s co-funded projects under JTC2019 and JTC2020 using annual and final reports
- Monitor EJP RD’s funded projects under the additional call JTC2021 and JTC2022
- Organise midterm monitoring meetings for the JTC2021
WP9 Monitoring of the results of the funded projects

WP7 Networking Support Scheme
- Monitor and analyse the networking support scheme funded events

WP8 Rare Diseases Research Challenges
- Monitor and follow-up on the 2nd phases of the RDR challenges funded projects
VIRTUAL PLATFORM OF DATA, TOOLS & RESOURCES
Pillar 2 – Expected impacts

Specific impact 1: Improve lives of rare disease patients by providing new and optimised treatment options and diagnostic tools for these diseases

- Making data in registries, biobanks and knowledge bases FAIR and data sources and infrastructures standardized and interoperable for data use optimization
- Generating re-usable knowledge out of multi-omics data from research for biomarkers and targets identification and for re-use in diagnostic pipelines

Specific impact 2: Decrease fragmentation of rare diseases expertise and research resources

- Creating a network of RD-relevant resources and data sources that is harmonised, interoperable for humans and machines, and easy to find for researchers
- Creating the conditions for expanding the network

Specific impact 3: Increase the EU's capacity to innovate in the field of rare diseases

- EU resources are now better adapted for RD research
- EU resources are now in capacity to computationally interact with each other to increase their innovation potential

Specific impact 4: Improve healthcare systems' capacity to take up research results

- The outputs of Pillar 2 activities (models, standards, mapping services, model transformation tools) will be provided to member states to ease the integration of ERNs in national health systems, bridging healthcare and research
Pillar 2 – Expected impacts

Specific impact 5: **Reinforce the EU's role as a global leader for rare diseases**

- Creating a federated architecture of a diversity of resources that is unique in the world
- Setting up a model based on standards that is a blueprint for other networks (health data, genomics...)

Specific impact 6: **Follow the policies and contribute to the objectives of the International Rare Diseases Research Consortium (IRDiRC)**

- The support to IRDiRC policies and objectives is translated through the provision of a strategic instrument to understand and analyse the state of play of RD funded research.
Pillar 2 evolution: long learning curve now paying

- 2019: Startathons
  - « Deconfusion »
- 2020: FAIRathons
  - Building-blocks
- 2021: Connectathons
  - Integration
- 2022: Progressive full-service delivery
  - API refactoring
  - VP V1
  - VP V2
  - VP V3
  - VP V4
  - VP V5
- 2023
- 2024

https://vp.ejprarediseases.org
P2 achievements: From a heterogeneous, non-interoperable, scattered resources landscape ...
To the Virtual Platform: a network of federated resources

- Registries/biobanks catalogues
- Cell lines Animal models
- Knowledge bases
- Support for clinical/translational research
- Tools catalogues
- Data deposition & analysis platforms

Decreasing fragmentation
Increasing interoperability
Increasing RD-readiness through Harmonisation
Standardisation in a flexible way:
Common methodologies, Multiple technical solutions

V1: https://vp.ejprarediseases.org
Onboarding at diverse levels of connection to the Virtual Platform

Level 1
- Metadata discovery
- Basic description of resource
- Overview of content

Level 2
- Discovery of limited parts of datasets
  - output: yes/no, counts, etc..

Level 3
- Querying of datasets
  - record-level querying

Level 4
- Federated Analysis
  - Answer advanced research questions

What is it?
What it contains?
Answer my complex question

Y6: onboard 100% of EJP RD partner resources
Succeed L3 for ready resources
LifeScience AAI fully implemented
POC Federated analysis on FAIR data
The EJP-RD Virtual Platform

The Virtual Platform (VP) is a growing network of Findable, Accessible, Interoperable and Reusable (FAIR) resources, ready to serve the rare disease RD research community.

It includes catalogues of resources, registries, biobanks, knowledge bases and tools compliant with agreed standards.

The VP Portal allows you to search the VP network resources at once in real time to find those of interest to your research.

Discover rare diseases resources and data:

Search for a disease name (e.g. ADPKD), gene (e.g. PKD1), or Orphacode (e.g. 730)

Advanced Search to filter by specific criteria

Tools and Resources

Explore Rare Disease resources with the EJP Mind Map
Find out how to make your data more FAIR
Contact us for information or feedback

22 resources connected

- 11 registries
- 3 catalogues
- 2 genome-phenome deposition infrastructures
- 5 knowledge bases
- 1 project

https://vp.ejprarediseases.org
Behind the scenes

Figure 4: Illustration of the levels by which resources can contribute to and benefit from the Virtual Platform. Resources that apply the recommended extendible standards ‘for machines’ at source automatically increase the functionality of the Virtual Platform for the community.
The VP is a technical architecture minimized central components to federated enhanced resources Del AD49 Virtual Platform Specification (VIPS v2.0)
The VP architecture is well documented


https://github.com/epj-rd- vp
The VP is based on standards

- Meta-data standards (DCAT-based)
- Standards on data file formats, markup, and annotation
- Standard data element sets
- Standards on data models
- Standards on data ontology, terminology and vocabulary
- Standards on data discovery
- Standards on data exchange mechanisms
- Standards on security, authentication, and authorisation
- Tools

[Compile]
Updated a list of >90 standards and tools used in FAIR-based implementations relevant for Rare Diseases (Del 12.3)

[Guide]
The smart guidance WF developed a question-based knowledge map for the ELIXIR Data Stewardship Wizard to provide user-tailored guidance – the first version is focused on ERN registries

[Simplify]
A small set of standards and tools were selected for VP ‘onboarding’ at different levels
VP components are assessed for their technical sustainability

Following technical developments in Y4, the strategy to assess sustainability has been adapted to include quality and GDPR compliance review.

Evaluation meetings conducted after completion of the kit

1. Component dependencies
2. Licensing and ownership
3. Contingency plans
4. Documentation with respect to potential users
5. Credentials
6. Technical information
7. Interoperability
VP is adaptive

https://specs.fairdatapoint.org/

VP is based on FAIR data models at the source

- Rare disease resource (e.g. a registry)
- Compatible with EJP RD VP specs & FAIR principles
- Web User Interface
- Application Programming Interface(s)
- Machine understandable declaration of content

Semantic ‘measurement-process’ modelling pattern for observational data

- A reusable, scalable, and queryable data model ‘for machines’ using standard ontologies, applied at source
- Updated the core model and modules for the 16 common data elements for patient registries
- Mappings and bridging solutions to other semantic frameworks & formats (e.g. FHIR, OMOP, C-DISC, OBO Foundry, GA4GH)

Kaliyaperumal & Wilkinson et al., 2021
P2 provides tools and support for resources

**VP onboarding**

- *Specs & tools ready for resources creating the VP network* = ‘onboarding’
- FDP and FDP index specifications for humans and machines Findability and Queryability (https://specs.fairdatapoint.org/)
- Technical facilities to build up resources’ metadata provision: a common method, multiple technical solutions
  - Spreadsheet
  - FDP local deployment / FAIR-in-a-box
  - Software solutions that implemented specifications for metadata provisioning (e.g. MOLGENIS)
  - Onboarding guide document initiated (in finalization)
- An onboarding Hackathon (December 2022 + follow-up WS)
- Orphanet services for improving querying (for VPPortal and for resources)
  - Semantic mapping service API
  - RD Hierarchies service API
  - Genes/diseases mapper API

**FAIRification**

“A Model for Guiding Data Stewards of European Rare Disease Patient Registries”
van Damme et al., CODATA Data Science Journal, under review
https://www.ejprarediseases.org/the-smart-guidance-tool-for-the-fairification-of-rare-disease-registries/

Tackling the reusability challenge: ongoing work

Core effort
“Light” Semantics
For software engineers

End-User Tooling
Data Use Conditions (DUC) Template
DUC JSON Template
END-USER TOOLING
EJP DUC/CCE Creator
DUC in DCAT (not ongoing)
END-USER TOOLING
ODRL CCE Builder
ODRL CCE -> Narrative multilingual Translator
ChatGPT-based CCE builder (nascent)
Automated Negotiator (nascent)

Parallel effort
Semantics for machines

Survey of Consent and Data Usage Forms
Common Consent Elements (CCEs) Templates
CCE JSON Templates
Reference DUC/CCE Creator
CCE JSON Templates
Molgenis DUC/CCE Creator
CCE JSON Templates

Y6: finalize implementations initiated second half Y5
Tackling the privacy preserving record-linkage challenge: ongoing work

PPRL use cases
- GPAP ↔ 1 ERN-PaedCan Registry
- EJP-RD Portal ↔ EUPID Services

PPRL interoperability
- Technical implementation of selected approaches (MainSEL, EUPID)
  - Publish – results might also be used by Spider etc. in the future
- Further analyse potential solutions for PPRL Interoperability
  - Publication on status and concepts for interoperability

Y6: finalize PPRL interoperability POC
Finalize PPRL implementation use-case (EUPID)
Pillar 2 partners have improved data deposition and analysis resources for RD

- BBMRI-ERIC directory / RD-Connect Registry and Biobank Finder
- RaDiCo
- HPSCreg
- Cellosaurus
- INFRAFRONTIER
- MetaboLights
- CTSR
- EGA
- DECIPHER
- RD-Connect GPAP

Most resources made steps towards onboarding to the VP (level 1 or level 2)

RD-Connect GPAP: new GUI with many new functionalities. Implemented Phenopackets 2.0.
BBMRI-ERIC Directory: UI/UX improvements, mapping ICD-10 and ORDO, new RD search category
RD-Connect Registry and Biobank Finder: all entries migrated to Molgenis. Negotiator PoC.
RaDiCo: application of FAIR data principles to data and metadata, onboarding to VP.
Virtual Cluster Environment & workflows: new documentation, towards 2-factor-auth.
Knowledge generation from collaborative multi-omics data analysis

Y6: dissemination and publications
Finalising AOP pathways work

4 case studies – (multi) omics datasets from funded projects

- FAIR data analysis workflows
- Scientific publications, pathways
- Additional tools for comparability of multi-omics results

Investigating overlap with chemical compounds
- Drugs (Pathway-network extension workflow and RareGenoScope)
- Nutrition (publication)
- Adverse outcome pathways (to be explored)

Genetic variant prioritization and analysis
- New features for the VEP (Ensembl)

115 rare disease pathways available and reusable

Lessons learned: VP sustainability aspects

Aspects

Technical sustainability
- Of the components
- Of the ecosystem

Support evolutivity
- Quantitative growing as a network
- Qualitative diversify the network

Maintain and create value
- Develop services
- Become a major actor: attractivity

Needs

Evolutive technical maintenance

Strategic, tactical and operational capacity

Development and valorisation
VP sustainability & scaling-up aspects

Needs

• Evolutive technical maintenance

• Strategic, tactical and operational capacity

• Development and valorisation

Solutions

• Technical sustainability

• Rules & traceability

• CTO

• A well-defined Governance

• Business model
  • Membership?
  • Public funding?
  • Sales?
CAPACITY BUILDING & TRAINING

EJP RD

Policy Board and Governing Board

4 July 2023, Brussels

Birute Tumiene, VUHSK
Roseline Favresse, EURORDIS
Pillar 3
Training and capacity building: objectives

WP14
Training on data management & quality
- Decrease RD data fragmentation and increase data quality through training
- Provide training on data management & quality to increase the level of capacities and help data sharing and networking
- Training activities on standardization of RD data, standards & quality in genetic testing, strategies for undiagnosed RD cases, RD sample data management, RD registries.

WP15
Capacity building and training of patients and researchers
- Training on therapeutic development & regulatory processes for medicinal products in RDs for patients and researchers
- Providing the knowledge & skills required for patients to become legitimate collaborators in RD research
- Empowering patient representatives as equal, valued, and efficient partners in research
- Provide younger patients with specific knowledge, skills and educational tools on RD research

WP16
Online Academic education course
- Provide an EU multidisciplinary and transversal online research education course
- Identify needs, audience and topics
- Develop a series of 5 Massive Open Online Courses (MOOCs) in collaboration with KoL and experts
- Monitor the MOOCs and assess their impact.

WP17
ERN RD training and support programme
- Map and assess the existing landscape of the ERN research training programs
- Develop training programs consisting of crosscutting and overarching research training activities
- Establish an accreditation process

WP18
Development of new trainings
- Evaluate the state-of-the-art of RD research education and training across several axes
- Assess the impact of EJPRD’s special provisions to increase accessibility
- Define the needs and gaps, challenges and opportunities for the further improvement;
Pillar 3 - Achievements

Training on data management & quality (WP14)

- 9 national training sessions have been organised in 5 different countries (France, Italy, Norway, Spain, Turkey) on the Orphanet nomenclature and RD ontologies (309 trainees of which 29 in the TfT)

- 4 editions of the training course on Standards and quality of genetics/genomics data in laboratory and clinical research practice organized in 4 different countries (Belgium, Turkey, Italy, Germany) with a mix of in-person and online editions (107 trainees); next edition will be in Warsaw

- 4 editions of the international course Training on Strategies to foster solutions of undiagnosed rare disease cases with a mix of in-person and online editions (104 trainees)

- 9 training workshops on RD Biobanks in 8 different Countries (Italy, Lithuania, France, Switzerland, Germany, Netherlands, Poland, Spain) (268 trainees)

- 4 editions of the 5-day face-to-face training course International Summer School on RD registries and FAIRification of data with a mix of in-person and online editions (104 trainees)

892 trainees from 2019 to June 2023

Impact

Increased level of knowledge and capacity of the RD research and care community as a whole on standards for data, samples and genetic testing; on data management, on FAIRification of data.

Supporting efforts of the FAIR ecosystem as well as efforts to decrease RD data fragmentation and research duplication

Faster transfer to the RD community of the knowledge and tools produced in EJP RD
Pillar 3 - Achievements

Capacity building/training of patients (WP15)

Impact

2024
- 5 editions of the ExPRESS Expert Patients and Researchers, aka EURORDIS Summer School on Medicine Research and Development (157 trainees, incl 35 researchers) (3 online, 2 F2F)

2024
- 4 editions of the EURORDIS School on Scientific Innovation & Translational Research (107 trainees) (3 online, 1 F2F)
  - Training for patient representatives and advocates on leadership and communication skills organized (138 trainees) (2 F2F, 1 online)
  - 3 trainings on RD research for young patients organized (2 face to face: Lyon and Barcelona, one online in 2020-21) (12-18 y.o.) (33 trainees)

440 trainees from 2019 to 2023

Increased number of patient representatives involved in JTC and NSS calls

Supporting EMA protocol assistance
+150 patient rep involved in protocol assistance at the EMA

Supporting patient representation in EMA committees
4 patients selected as members of the EMA Paediatric Committee and in the Committee for Orphan Medicinal Products

New patients in IRDIRC task forces

Empowering expert patients to provide critical reviews (EJP RD JTC and NSS Calls)

Empowering young patients’ advocates by developing an assent form with and for young patients to process and share data for RD in ERNs
Pillar 3 - Achievements

Online academic education course (WP16)

- MOOC#1 - Diagnosing RD: from the Clinics to Research and back
  - Codeveloped by FFRD, ERN Ithaca, ERN Genturis, EURORDIS
  - Continuous opening with 2 annual facilitation windows in 2022
  - 1765 new participants joined the course in 2022, with a total of +5200 participants from 148 countries since April 2021
  - In 2022: new contents initiated: videos’ series on the ethical aspects, series of motion videos on omics approaches as well as a series of text steps on AI
  - 12 young researchers (mostly from ESHG-Y) helped in mentoring the facilitation windows.
Pillar 3 – Achievements

Online academic education course (WP16)

• MOOC#3 - From lab to clinic: translational research for rare diseases
  • Codeveloped by FFRD, EATRIS, LUMC, EURORDIS, Euro-NMD
  • Launched in October 2022
  • +1400 enrolled learners coming from 112 countries
  • 2 facilitation windows per year, during which experts in the field are online for 2 months in order to answer to participants’ questions
  • 7 young researchers, mainly contacted via the ESHG-Y helped in mentoring the facilitation windows.
Pillar 3 – Achievements

Online academic education course (WP16)

- 2 additional MOOCs continued to be developed in 2022
  - MOOC#2 - *Innovative therapies and personalized medicine approaches for RD*
    - Developed by FFRD, CVBF, ERN Transplant Child
    - Launch foreseen in Autumn 2023
  - MOOC#5 - *Rare Diseases data for research purposes: ethics and regulatory considerations*
    - Developed by FFRD, FGB, EURORDIS, ERN Epicare
    - Launch foreseen in early 2024
- 1 MOOC stopped: *Clinical trial methodologies for RDs*
  - Development planned in collaboration with EJP RD WP20
  - Cancelled due to the difficulty of adapting the topic to the MOOC format

2049 enrolled trainees in 2022
6655 enrolled trainees from 2021 (1st MOOC released in April 2021)

Impact

- 92% and 100% positive feedback for the two MOOCs available
- Very heterogenous audience (from patient to medical specialists through researchers from various disciplines and at very different career stages)
- Discussions ongoing to translate the Diagnostic MOOC in different languages, in order to reach a larger public with the financial support of external stakeholders, starting with French
- International Outreach: 40% of the learners of the MOOCs are from outside Europe
- Sustainability plans being discussed with ERNs (ITHACA and EURO-NMD)
Pillar 3 – Achievements

ERN RD training and support programme (WP17)

• ERN RD training and support programme (WP17)
  • 6 calls for workshops
  • 6 calls for fellowships
  • In 2022, 7 workshops have been successfully conducted, 4 of which in hybrid format and 3 in ‘in-person’ format. The workshops have been attended by different nr. of participants depending amongst other on the event format. (+180 in person/+170 online)
  • EACCME (European Accreditation Council for Continuing Medical Education) obtained for 3 workshops (7 to 11 CME points) → CME points are not driving the attendance at such workshops (based on evaluations performed)
  • All fellows (usually performing a 2-6 month-fellowship) were very to extremely satisfied and were able to achieve most of their goals during their fellowship.

89 fellows
31 workshops

Impact

Newly created networks and working groups of experts deriving from workshops & fellowships that are eager to work jointly on new projects and proposals.

Discussions leading to new publications (on outcome measures e.g.)

Exchange of perspectives between basic scientists, clinical scientists and patient representatives.

Increased research capacities and knowledge amongst the ERN centres and affiliated research and healthcare centres.
Objectives of the study (WP18)

- To evaluate the **state-of-the-art** of RD research education and training across several axes;
- To assess the **impact of EJPRD's** special provisions to increase accessibility;
- To define the **needs and gaps, challenges and opportunities** for the further improvement;
- To comprise the **conclusions and recommendations** and to disseminate the outcomes of the study to relevant stakeholders.

Limitations of the study

- Lack of published data or any other resources for the evaluation of the general RD research education and training.
- Focus on short-term (vs. long-term) impacts.
- Definition of “RD research education and training” vs. more general “RD education and training”.

*More than 90 references, 28 EJPRD internal documents and multiple meetings/discussions/calls.*
State-of-the-art of RD research education and training

- a general lack of knowledge and awareness about RD among the multistakeholder RD research community.
- a huge lack of knowledge and awareness about available RD research resources and data management aspects among the multistakeholder RD research community.

- objective evaluation of knowledge of multistakeholder community: correct answer rates for various questions from 2% to 91%.
- self-rated knowledge on RD: insufficient and poor – from 45% to 98%, especially among non-specialists;
- educational and informational sources: academic training not useful or insufficient for 7% - 17% of specialists and 80% of GPs) [Vandeborne 2019]; continuous medical education, scientific literature and conferences are considerably more important as a source of information on RD for practicing specialists; the Internet was mentioned as an important source of information about RD by a considerably higher number of practicing professionals as compared to students.
- awareness of where to find information about RD (e.g., Orphanet): from 0.9% to 85%.
- self-rated readiness to provide care to RD patients: not ready from 28% to 94%.
- RD patients are not recognized in practice: e.g., encountered just 4.2% during the last year [Miteva 2011] or just 52% overall [Vandeborne 2019].
- general willingness to broaden knowledge on RD: from 44% to 95%.
Challenges and opportunities for RD research education and training

• Although the need for RD education and training is evident from both public health and learners’ perspective, there are multiple challenges, including:
  ➢ concept of rarity;
  ➢ novelty, rapid development and expansion of RD field;
  ➢ heterogeneity of RD and multistakeholder community;
  ➢ lack of role of professional organizations;
  ➢ lack of awareness about existing educational resources;
  ➢ unequal competitive conditions as compared to more common diseases;
  ➢ a higher reliance on international networking and collaboration;
  ➢ increasing complexity of ELSI, data management and regulatory issues, and
  ➢ geographic inequities.

Other factors may present both challenges and opportunities for RD research education and training:
  • interconnections with innovative fields; the role of RD patients and PAOs; digital transformation of teaching and learning; professionalism, social accountability, cultural safety and responsiveness.
## Activity | Targeted stakeholders | Keywords
--- | --- | ---
P3 WP 14.1: Training on the Orphanet nomenclature and RD ontologies for RD research. Training for Trainers; National courses. | Orphanet National Teams | Ontologies, RD codification, data management

P3 WP 14.2: Standards and quality of genetics/genomics data in laboratory and clinical research practice F2F courses. | Laboratory scientists, clinical geneticists, medical specialists, policy makers and assessors for laboratory accreditation, patient representatives with a basic knowledge of biology or medicine. | Genetic diagnostics, genomic technologies, quality assurance and management of laboratories, data analysis and management

P3 WP 14.3 Training on strategies to foster solutions of undiagnosed rare disease cases F2F courses. | Clinicians, medical specialists, rare disease patient representatives, multistakeholder community | Undiagnosed diseases, multi-omics, functional analyses, diagnostic pathways, networking and matchmaking

P3 WP 14.4: Training for biobanks and researchers/clinicians on sample data management F2F courses. | Clinicians, data managers, biobanking specialists, rare disease patient representatives and registry curators, database managers, rare disease patients representatives, multistakeholder community | Biobanking, management of data and samples, quality assurance, ontologies, ethical, legal and social issues (ELSI), stem cells

P3 WP 14.5: Training on rare disease registries and FAIRification of data at the source F2F courses. | Clinicians, medical specialists, registry curators, database managers, rare disease patients representatives, multistakeholder community | RD registries, data FAIRification, ontologies, data management
<table>
<thead>
<tr>
<th>Activity</th>
<th>Targeted stakeholders</th>
<th>Keywords</th>
</tr>
</thead>
<tbody>
<tr>
<td>P3 WP15.1 - ExPRESS Expert Patients and Researchers EURORDIS Summer School Pre-training and e-learning courses; F2F courses</td>
<td>Patient advocates, researchers</td>
<td>Clinical trials, Orphan drugs, regulatory and ethicolegal issues, patient engagement, pharmacovigilance, European Medicines Agency, Health Technology Assessment</td>
</tr>
<tr>
<td>P3 WP15.2 – Training for patient advocates on scientific innovation and translational research - EURORDIS Winter School Pre-training and e-learning courses; F2F courses</td>
<td>Patient advocates</td>
<td>Translational research, genetics, bioinformatics, ERNs, RD diagnostics, undiagnosed diseases, gene/advanced therapies, genome editing, patient engagement</td>
</tr>
<tr>
<td>P3 WP15.3 – Training for patient advocates on leadership and communication skills Pre-training and e-learning courses; F2F courses</td>
<td>Patient advocates</td>
<td>Leadership, self-awareness, conflict resolution strategies, authority, negotiation, networking, communication</td>
</tr>
<tr>
<td>P3 WP 15.4: Educational materials and activities for paediatric patients e-learning; F2F courses</td>
<td>Pediatric patient advocates</td>
<td>Rare diseases, patient engagement, clinical research, ethicolegal issues, informed consent/assent form, patient wellbeing, Rare Disease Day</td>
</tr>
<tr>
<td>Activity</td>
<td>Targeted stakeholders</td>
<td>Keywords</td>
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</tr>
<tr>
<td>P3 WP16 MOOC#1 - “Diagnosing Rare Diseases: from the Clinic to Research and back” On-line academic course.</td>
<td>Students, multistakeholder community (researchers, clinicians, patients and patients' representatives)</td>
<td>General concepts about RD and genetic diagnostics, genomic technologies, care pathways, patient-centeredness, undiagnosed diseases, genetic consultation, genetic research</td>
</tr>
<tr>
<td>P3 WP16 MOOC#2 - Innovative personalized therapies On-line academic course.</td>
<td>Students, multistakeholder community (researchers, clinicians, patients and patients' representatives)</td>
<td>Rare disease treatment, innovative therapies, gene &amp; cell therapy, regenerative medicine, genome editing, personalized medicine</td>
</tr>
<tr>
<td>P3 WP16 MOOC#3 - Translational Research On-line academic course.</td>
<td>Students, multistakeholder community (researchers, clinicians, patients and patients' representatives)</td>
<td>Translational research, preclinical and clinical research, disease models, biomarkers, clinical trials, regulatory issues, ethicolegal aspects, postmarketing</td>
</tr>
<tr>
<td>P3 WP16 MOOC#4 Rare Disease Clinical Trials innovative methodologies On-line academic course.</td>
<td>Students, multistakeholder community (researchers, clinicians, patients and patients' representatives)</td>
<td>Small population clinical trials, clinical trial designs, statistical analysis</td>
</tr>
<tr>
<td>P3 WP17 Research training workshops</td>
<td>Clinicians, multistakeholder community</td>
<td>N/A</td>
</tr>
<tr>
<td>P3 WP17 Fellowships for research mobility secondments</td>
<td>Young clinicians, multistakeholder community</td>
<td>N/A</td>
</tr>
</tbody>
</table>
EJPRD education and capacity building programme: evaluation

- **R & I pipeline**: although the whole R & I pipeline is covered, current EJPRD educational programme is more responsive to the needs of researchers of pre-clinical and clinical studies and end-users (e.g., patient representatives) as compared to basic studies. However, basic research studies are intended to prepare scientists for the further specialized studies, including RD research studies, hence their smaller role in the RD research education and training.

- **Career stages**: EJPRD education and training programme encompass the whole axis of career stages from students (MOOCs in WP16) to junior investigators (the vast majority of education and training activities in the EJPRD), and to advanced researchers (horizon-scanning educational activities, as webinars on new trends and innovations). However, although targeted audiences for any given EJPRD educational activity were pre-identified (e.g., MOOCs in the WP16 were developed as academic courses mostly targeted at students), the courses and trainings have been attended by the vast range of participants.

- **Multistakeholder community**: EJPRD education and training programme is targeted at a vast range of multistakeholder RD research community including not only researchers, but also patient representatives, clinicians, multidisciplinary team members, bioinformaticians, laboratory technicians, biostatisticians, biobank and data managers, research nurses, etc.

- **Geographical coverage**: the geographical coverage was investigated from the perspective of “teachers” (composition of the teaching faculty) and from the perspective of learners. Although educators from certain EU-14 countries (e.g., DE, FR, NL, IT, ES) dominate in the teaching faculty, composition of the learners is much more diverse and includes a vast range of not only European countries, but also learners from all the continents (especially for the on-line courses). Therefore, education and training activities may help EJPRD to achieve the global impact on RD research.
**RD research topics:** The final list of RD research topic categories was identified from the keywords of existing educational and training activities and the brainstorming on any missing items:

- Pre-clinical studies (incl. disease pathomechanisms and models, biomarkers, natural history studies, etc.).
- Clinical trials (incl. small population trials, drug repurposing, medical devices, advanced therapies, etc.).
- RD registries and biobanks.
- Data science (incl. RD data management, resources, tools, FAIRification, application of AI technologies).
- RD diagnostics and undiagnosed diseases (incl. phenotyping, innovative methodologies for solving undiagnosed diseases, omics, functional analyses, etc.).
- Practical aspects of research (incl. ethicolegal issues, data management and sharing, patient engagement).
- Socioeconomical studies in RD (incl. RD burden investigations, innovative care organization, implementation science, health outcomes research, etc.).
- Social sciences and humanities (incl. equity and stigmatization, social determinants of health, psychological and social impact of RD, etc.).

- EJPRD education and training programme covers the vast majority of these topics to various extent. Somewhat less covered topics include education and training on socioeconomical studies, social sciences and humanities, preclinical studies and data science.
Impact of EJPRD: participation of widening countries

Empowerment and capacity building:
Participants from widening countries comprised:
- from **12 to 50%** of course participants in WP14 trainings;
- **20%** of ERN research mobility fellowships;
- **10%** of beneficiaries in ERN Research training workshops.
- Widening countries comprised from **18% to 33%** in WP15.

Ideally, participants with enhanced capacities in RD research will become active applicants for project proposals.

Rare disease research funding:
- **Networking scheme** (“COST-like” activities):
  In the first five rounds, funded applicants from widening countries comprised **21% (31 of 151)** of applicants.
- **Joint transnational calls (JTCs)**. **Widening principles were applied in E-RARE since 2015 and in EJPRD.**
Impact of EJPRD in the landscape of RD research education and training

- Participants from **widening countries** are highly active users of EJPRD education and training activities; special EJPRD provisions to increase participation from widening countries not only empower local communities with knowledge and skills in RD research, but also augment their experience to provide RD research education and training locally, and may be one of the factors to increase participation in research activities.

- Through education and training activities, directed at both patients and researchers, EJPRD provides a strong basis for **patient-centredness** in RD research. It may be one of the factors (together with improved regulations for PAO participation in JTCs) for a significant growth in PAO participation in EJPRD JTCs.

- RD education and training activities complement ERN educational and training programme, may empower ERN community with RD research knowledge and skills and foster its incorporation into the overall RD research ecosystem.

- Although data on **national RD research education and training activities are very limited**, some insights about insufficiency of these activities may be drawn from both literature data and surveys. The major responsibility of MS would be provision of basic RD and RD research education (provided mostly in universities and university hospitals), while major European and international efforts are required to provide highly-specialized RD research education and training.

- A promising option is a concept of “**training of trainers**” that is provided internationally and ensure standardization, high quality and up-to-dateness of trainings, that are further spread to national networks and adapted to local needs (e.g., EJPRD WP14.1).
Conclusions and recommendations

Actions at three levels are required:
(1) level of organisations, (2) national level, and (3) European and global level.

- Coherence of RD research education and training activities based on a common strategy across Europe and globally.
- Better alignment of national and transnational RD research education and training activities to fulfil the needs across RD research educational pyramid.
- Awareness-raising and education based on existing resources.
- Incorporation of RD research education and training into the overall RD research ecosystem to ensure up-to-date, empowering education and training and timely response to arising needs.
- Continuing efforts to diminish inequities and to foster inclusiveness in RD research through special provisions for underserved groups and underrepresented countries.
- Commitment for a long-term strategy for the RD research workforce development.
ACCELERATED TRANSLATION OF RESEARCH RESULTS & CLINICAL TRIALS
The overall aim of this work package is to provide researchers the mix of competences needed to support rigorous translational research, to secure follow-on funding and find partners for the development of new treatments and diagnostics for rare diseases. The main objectives are:

WP19.1: Translation acceleration
WP19.2: Support in exploitation and follow-on funding
WP19.3: Evaluation of Innovation Management and exploitation support tasks
WP19.4: Roadmap for a European investment platform for RD.

The aim is to foster the development of innovative methodologies tailored for clinical studies in RDs. The specific objectives are to map the best methodologies for clinical studies in RDs and to validate innovative and promising design and analysis methodologies for therapy evaluation in some specific rare diseases. This WP will support ERNs to use the most adapted methodologies improving clinical trial studies in RDs

WP20.1: Task Force Group
WP20.2: Support in design and planning of RD clinical studies
WP20.3: Demonstration projects on existing statistical methodologies to improve RD clinical trials
WP20.4: Projects on innovative methodologies to improve RD clinical trials in limited populations
Task 19.1 The Innovation Management Toolbox (IMT)

A reference library of resources in rare disease translational medicine. *(launched June 2022).*

- 450 resources
- 15 Use cases
- Integration of catalogues and toolkits:
  - ERICA catalogue of services
  - The Orphan drug development guide (ODDG)
  - The Clinical trial toolbox.
  - EJPRD online courses
- Resources Developed internally:
  - The ACT Tool kit. (Newcastle U)
  - Translational Research Management Manual (EATRIS)
  - Mentoring packages (WP19)

- A curation process was set up and implemented

- WP19 has worked closely with Pillar 2 to ensure discoverability of IMT elements in the virtual platform and avoid duplications.
Impact

Google Analytics
June 2022 – June 2023

Sessions: 3366
Visitants: 1956

IMT obtained the IRDiRC recognition
WP19.1 Mentoring and technical support for translational research projects

- Mentoring services was offered to shortlisted projects from JTC2020 (15 projects) and JTC2022 (13 projects) including a Mentoring Kick off webinar and one-on-one mentoring calls.

- **42 experts** in database to support the mentoring service and mentoring and legal documents were updated and published on the mentoring webpage

- **3 Mentoring packages have been created in the IMT** to support and guide the call applicants during the preparation and after receiving the funds.

- **Follow up mentoring services** is being offered to the funded JTC2020 projects (incl those that had not requested mentoring but were also funded) and to projects receiving other funding (E-Rare-2, H2020 Excellent Science, ... ) or projects without funding.
EJP RD Mentoring Overview

JTC2020 – 30 Applications (15x mentoring)
Pre-clinical Research to Develop Effective Therapies for Rare Diseases

- Diverse mentored topics.
- All projects received individual mentoring
- 13 experts involved.
- 400 hours of effort.
- Experimental Design: Drug screening, clinical trials, animal model, gene therapy, drug repurposing.
- 8 projects received funds.
- 11 projects requested follow-on mentoring.

JTC2022 - 21 Applications (13x mentoring)
Development of new analytic tools and pathways to accelerate diagnosis and facilitate diagnostic monitoring of rare diseases

- Kick-off webinar
- The range of topics was narrower due to the call topic and prior webinar.
- Fewer experts involved (4) and less hours per expert (135).
- DM planning, Innovation management, technology transfer, how to develop the project plan and «go»/«no go» decision/tollgates.
WP19.2 Support in exploitation and follow-on funding

- Updated the list containing public and private funding opportunities including grants, venture capitals, accelerators, incubators, business angels, and crowd fundings.
- Identified and expanded the list of experts with experiences in the field.
- Provided list of funding opportunities, patient advocate organizations, landscape of the companies developing for the therapy, advice on scientific and regulatory aspects to scientists requesting such support.
- A database of different funding resources from private entities and public agencies were integrated into the Innovation Management Toolbox (IMT).
- The database is regularly updated to make sure that the links of funding resources work well. Moreover, use cases on different categories have been being built for new functionalities of the IMT.
WP19.3 Evaluation of Innovation Management and exploitation support tasks

• WP19 has been actively engaging with the research funders (Pillar 1 and Health units at EC), WP3 Sustainability, and Projects such as ERICA (CSA project for the ERNs) and REMEDI4ALL with the intent to disseminate the mentoring service to these communities more broadly and hear back from the community on their satisfaction with the service.

• ERNLUNG Survey to evaluate the support provided to the rare disease research community.
WP19.3 ERNLUNG Survey (52 participants)

Have you ever used the Innovation Management Toolbox (IMT) offered by the EJP RD?

- Yes: 3
- No: 49

If you were conducting a translational research project, would this toolbox be beneficial for you?

- Yes: 38
- No: 11

If you already used the Innovation Management Toolbox, how would you evaluate:

- Very good
- Good
- Average
- Poor

- its search engine
- User-friendliness
- the usefulness of use cases
- the usefulness of Question and Answer section
- the usefulness of the available resources

100% 0%
WP19.3 ERNLUNG Survey (52 participants)

Have you ever heard about the mentoring for translational research service offered by the EJP RD?
- Yes: 21
- No: 31

Have you ever used the mentoring for translational research service offered by the EJP RD?
- Yes: 8
- No: 13

Was your application successful?
- Yes: 7
- No: 1
- The results are not yet available: 0
- Maybe: 3

Do you think that the mentoring contributed to the success of your application?
- Yes: 4
- No: 0

What kind of mentoring support would be useful for you?
- Quality and reproducibility of...: 9
- Design of experimental models: 8
- Regulatory advice: 22
- Drug development and scale-up: 10
- Innovation management and training: 10
- Pre-clinical models: 5
- Project management and milestone planning: 15
- Data management plan: 9
- Data FAIRification: 4
- Other: 1

If you had the opportunity to use this service when preparing your application and/or during the lifetime of your project, do you think that this kind of service would be useful for you?
- Yes: 33
- No: 11

If you had the opportunity to use this service when preparing your application and/or during the lifetime of your project, do you think that this kind of service would be useful for you?
- Yes: 33
- No: 11
WP19.4 Roadmap for a European investment platform for RD

- The report “Del 19.6 Roadmap for EU investment platform for RDs” was submitted to EC portal. It shows different financing models for rare diseases ranging from non-profit funding, to for-profit funding, public-private partnerships and new emerging models such as venture philanthropy or public benefit company. The committee has approved the report after a review meeting.
WP19: Activities foreseen for Year 6

**IMT - ODDG**
- Finalise Use cases in progress
- Maintenance of the IMT and ODDG will be undertaken
- Engagement with key stakeholders to ensure coherent long term innovation funding and investments
- KPI’s analysis and dissemination actions to increase awareness

**Mentoring services**
- WP19 partners will continue providing support for the projects funded by the JTC calls including the ongoing JTC2022;
- Support new projects for mentoring even they are outside of the EU funded programs.
- Comprehensive dissemination will be undertaken by identifying and directly contacting additional researchers and organizations working on rare diseases to offer the developed services.
WP20 Impact: Decrease fragmentation of rare diseases expertise and research resources

- Building of a community of methodologists involved in small samples trials with strong and trustful links with the clinical researchers
- Organization of five advanced courses (webinars) addressing methodologies in rare diseases clinical trial conduction, and training a wide range of stakeholders with an increasing participation and a successful communication plan
- Increasing dissemination (webinars, retreat) and with the series of papers with OJRD (2 papers accepted, 5 previewed)
Impact: Increase the EU's capacity to innovate in the field of rare diseases

Adaptation of RD statistical methodologies

- Funding of 3 demonstration and 2 innovation projects to validate new innovative methodologies for RD clinical trials
  - Positive effect on the interactions with ERNs and other stakeholders
  - Key used cases to help in establishing future methodologies for RDs

CT Toolbox kit implementation

- WP 20 developed and implemented the Clinical Trials Tool kit to support, guide and enhance clinical research, further need for communication and dissemination.
<table>
<thead>
<tr>
<th>Planned WP20 activities</th>
<th>Achieved by end of year 5</th>
<th>AWP Y6</th>
</tr>
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<tbody>
<tr>
<td><strong>Demonstration projects</strong></td>
<td>• EPISTOP IDEAL: December 2023&lt;br&gt;• EBStatmax: July 2023&lt;br&gt;• Improve PSP</td>
<td>• n/a&lt;br&gt;• n/a&lt;br&gt;• April 2024</td>
</tr>
<tr>
<td><strong>Innovation projects</strong></td>
<td>• iSTORE&lt;br&gt;• Evidence RND</td>
<td>• April 2024&lt;br&gt;• April 2024</td>
</tr>
<tr>
<td><strong>Advanced Webinars</strong></td>
<td>• Does Randomization matter in RD clinical trials?&lt;br&gt;• Composite endpoints including patient relevant endpoints (Quality of Life)&lt;br&gt;• The Statistical Evaluation of Surrogate Endpoints in Clinical Trials&lt;br&gt;• Statistical and operational challenges with master protocols&lt;br&gt;• Replicated N-of-1 RCTs for Rare Diseases&lt;br&gt;• Item response models for analysing assessments in rare diseases&lt;br&gt;• EFPIA Webinar: TBD, December 2023 (all available on EJPRD site with papers done or in prep)</td>
<td>• Modelling natural history in longitudinal data-Challenges and Solutions (2024)</td>
</tr>
<tr>
<td>Planned WP20 activities</td>
<td>Achieved by end of year 5</td>
<td>AWP Y6</td>
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<tr>
<td>Intermediate course Needs and request of the community</td>
<td>• One course to be provided</td>
<td>• Five courses to be provided</td>
</tr>
<tr>
<td>Clinical trial support office</td>
<td>• Support and guide ERNs/RD investigators focusing on the methodological/study design and providing support advice &lt;br&gt;• Raise awareness of the service &lt;br&gt;• Increase the quality and number of RD clinical trials and projects/collaborations for clinical research on RD</td>
<td>• Pursue its achieved work &lt;br&gt;• Uncover ERN roadmap for clinical research to evaluate their current and future needs in terms of support to set up clinical trials. &lt;br&gt;• Communicate and disseminate</td>
</tr>
<tr>
<td>Clinical Trial toolbox kit</td>
<td>• Development and implementation of the CT toolbox</td>
<td>• Update and refine and improve the CT toolbox performance &lt;br&gt;• Improve the performance of academic sponsored clinical trials &lt;br&gt;• Communicate and disseminate</td>
</tr>
<tr>
<td>PILLAR</td>
<td>ELEMENT/ASSET</td>
<td>TYPE</td>
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</table>
| 4      | Clinical Trials Support Office                    | Service            | • Leverage the expertise of partners with years of experience in their respective fields  
|        |                                                   |                    | • Support/advice for the planning and design of clinical studies for Rare Diseases (RD), mainly multinational research  
|        |                                                   |                    | • Enhance the quality and number of RD clinical trials and of projects/collaborations for clinical research on RD   |
| 4      | CT toolbox                                        | Service            | • Implementation of the CT toolbox in the virtual platform  
|        |                                                   |                    | • Improves the performance of academic-sponsored clinical trials for rare diseases: advice in designing and conducting publicly funded clinical trials in Europe     |
|        | Building a community of methodologists in data and CTs | More than a service | • Unique group dedicated and available for all researchers and clinicians to guide the CTs in RD  
|        |                                                   |                    | • Unique possibility to link with EMA/FDA with the help of the regulatory expertise of the EJPRD and further of the RD partnership |
European RARE DISEASES PARTNERSHIP

Policy Board & Governing Board Meeting

04/07/2023
SRIA update
What is SRIA?

The Strategic Research and Innovation Agenda is a partnership’s strategy document, which identifies the partnership’s targeted impact, foreseen portfolio of activities, measurable expected outcomes and resources within a defined timeframe.

A SRIA should be able to translate the vision of the partnership in a long-term systemic approach to define the logic, rationales and principles of its operations also involving dealing with emerging uncertainties.
RD Partnership & SRIA key steps:

- RDP concept paper published in Feb 2022
- SRIA Task Force constituted in May 2022
- Development of the SRIA from June 2022 to Jan 2023 (1st draft version)
- Development of KPIs
- Public consultation May – June 2023
SRIA development timeline

2023

This timeline is subject to change (last update 27-06-2023)
Main tasks of the SRIA Task Force

Develop & propose a SRIA draft – to be subject of consultation(s) by EC group & more widely of a public consultation

• Agree on the process for SRIA preparation (type of activities that will be part of this process, e.g., organisation of Working Groups, public consultation, process for inviting experts, etc.)

• Agree on the structure of the SRIA (level of granularity with which different topics will be addressed)

• Help in coordinating the work of the SRIA process and notably of the working groups if created. (But not everything needs to be agreed at the 1st SRIA meeting...)

+ SRIA Task Force members can be active actors in allowing their national community to reflect on the Concept Paper and thus help fine-tuning/improving the plans for the Partnership!
SRIA Task Force composition

Active experts involved in the Concept paper development representing (but not limited to):

• Various fields of activity
  • (preclinical, translational and clinical research; drug development and diagnostics innovation; biostatistics; data science; regulatory science; research funding);

• Different types of stakeholders
  • (research organisation/institutions; hospitals/university hospitals; EU research infrastructure; patients’ organisations; foundations; funding bodies; regulatory & health technology assessment bodies, Member States representatives, European Commission);

• Relevant programmes, initiatives and networks
  • (EJP RD; Solve-RD; ERNs; Innovative Health Initiative; European Health Data space; DARWIN EU; CSA STARS; C-PATH).
RDP concept paper: on the right track, but…

- Reduce amount of text
- Bring in logical pathways from resources and actions to outcomes and impacts
- Maybe lose one of the streams (proposal SDG 17) or integrate elsewhere (seems difficult to measure)
- Be more concrete on the level of actions and resources & link to pathways
- Select most meaningful / impactful elements from table 2 targets

E.g. How to measure? What is the exact objective?
Example of Partnership Specific Impact Pathway (PSIPs) from Innovative Health Initiative
SRIA TF meeting 23-09-2022 - output

- Use IRDiRC goals (adapted if needed) as new General Objectives
- Revise the objectives of the partnership and formulate new PSIPs
  - Coo team makes proposition
  - Small group of volunteer experts from SRIA TF group revises
  - Coo adapts and shares with the whole TF group
RD Partnership adaptation of the PSIPs based on feedback received

**Step 7 (adaptation of the PSIPs according to Task Force Feedback)**

**VISION:** To improve the health and well-being of 30 million persons living with a rare disease in Europe, by making Europe a world leader in RD research and innovation, and delivering concrete health benefits to rare disease patients, through better prevention, diagnosis and treatment.

**GO1**

**Diagnosis established or subject to systematic research within 6 months after coming to medical attention**

- **GSO1:** Generation of knowledge and its translation into medical and holistic intervention.

**GSO2:**

Healthcare and research data are accessible and used for scientific and regulatory evaluation and healthcare delivery.

**GSO3:**

1. Meaningful engagement of people living with a rare disease in research to healthcare cycle (OR)
2. All activities embody leadership, empowerment or engagement, as equal partners, of people living with rare disease (OR)
3. All activities empower as equal partners people living with rare disease.

**GSO4:**

Increased capacity and skills of RD stakeholders to optimise research and healthcare continuum.

**GSO5:**

Integrated multinational and multi-stakeholder Research & Innovation ecosystem for RD.

**GO2**

**1000 new therapies for rare diseases approved**

**GO3**

**Better understanding of the impact of RD on patients, families and society**

**GO4**

Investment in patient needs-led research and innovation

**GO5**

Support robust data, resources and expertise infrastructure

**GSO3:**

1. Coordination and alignment of European, national and regional research strategies and resources (OR)
2. Coordination and alignment of international and regional research strategies and resources.

**GSO4:**

Training and education of RD stakeholders.

**GSO5:**

Multi-stakeholder collaboration
Step 8 (THE PSIPs exercise) 🎉 🎉 🎉

001 → S05, S01, S03, S02
002 → S05, S01, S02
003 → S05 (and all other S0s)
004 → S04
005 → S04, S05, S01
S01 → GO2
S02 = S03, S00
Reminder _ SO specific development methodology

Analysis of the PSIP – example for the SO5

(Integrated multinational and multi-stakeholder Research & Innovation ecosystem for RD)
Final PSIPs

VISION: To improve the health and well-being of 30 million persons living with a rare disease in Europe, by making Europe a world leader in RD research and innovation, and delivering concrete health benefits to rare disease patients, through better prevention, diagnosis and treatment.
Reminder_ SRIA development Methodology

Considered inputs:

• **Identification of elements in the RDP Concept Paper** (Especially for scopes and challenges)

• **Analysis of the PSIPs** – Interactions between Specific Objectives (SOs) and:
  - Operational Objectives (OOs) as inputs
  - Others SOs interactions
  - General Objective(s) (GOs) as target(s)

• **Identification of key Points captured by SRIA TF:**
  - Ideas for SRIA developments captured from all the comments provided during the development of the PSIPS and the redefinition of the Feb
  - presented during the SRIA TF meeting on 23/09/2022

• **Using IHI SRIA for inspiration** (conciseness & structure)
Vision of the Rare Diseases Partnership

1.1. Missions
1.2. Building on Lessons learned
1.3. Intervention logic - Partnership Specific Impact Pathway (PSIP)
1.4. General Objectives
   1.4.1. Diagnosis established or subject to systematic research in average within 6 months after coming to medical attention
   1.4.2. 1000 new therapies for rare diseases approved
   1.4.3. Better understanding of the impact of RD on patients, families and society
1.5. Activities and resources (Operational Objectives)
1.6. Thematic focus?
1.7. Synergies with other initiatives

2. Specific Objectives of the Rare Diseases Partnership

2.1. Specific Objective X: XXX
   2.1.1. Challenge
   2.1.2. Scope (if feasible)
   2.1.3. Potential Outputs
   2.1.4. Specific Outcomes

3. Performance Indicators

4. Conclusions

5. Annexes

5.1. European Partnerships, EU Missions, EU Programmes, Projects and organisations of potential relevance

5.2. Glossary
Operational Objectives (OOs)

• OO1: Investment in patient need-led research and innovation

• OO2: Support robust data, resources and expertise infrastructure

• OO3: Coordination and alignment of European, (inter)national and regional research strategies and resources

• OO4: Training and education (of RD stakeholders)

• OO5: Multi-stakeholder collaboration
SO1: Generation of knowledge and its translation into medical and holistic intervention

SO2: Healthcare and research data are accessible, and used, for scientific and regulatory evaluation and healthcare delivery

SO3: All activities empower, as equal partners, people living with rare disease

SO4: Increased capacity and skills of RD stakeholders to optimise research to healthcare continuum

SO5: Integrated multinational and multi-stakeholder Research & Innovation ecosystem for RD
GO1: Diagnosis established on subject to systematic research in average within 6 months after coming to medical attention

GO2: 100 new therapies for RDs approved

GO3: Better understanding of RD impact on patients, families and society
• Number of undiagnosed patients who receive a confirmed diagnosis or enrolled in systematic research within 6 months after first medical examination at secondary care level, facilitated by the partnership.

• Number of countries having undiagnosed programmes/activities.

• Number of improvements on the time to diagnose patients seeking medical attention for an unknown condition.

• Best practices developed within diagnosis-translational pipelines disseminated or adopted or implemented, by diagnostic centres.

• Number of improvements (efficiency, quality) in all steps underlying diagnosis, from gains in fundamental research (e.g., biomarkers) to the clinical journey of a patient.
- Number of new therapies approved for rare diseases per year.
- Number of clinical trials conducted for new therapies for rare diseases
- Number of partnerships between industry, academia, and government to develop new therapies for rare diseases
- Time to approval for new therapies for rare diseases
Number of research studies conducted on the impact of rare diseases on patients, families, and society

Number of publications and presentations on the impact of rare diseases in scientific conferences, policy briefings, and media outlets

Number of policy changes or initiatives at local, national, and international levels aimed at addressing the impact of rare diseases on patients, families, and society

Number of collaborations between patient groups, academic researchers, industry, and government to address the impact of rare diseases

Increase in funding for research on the impact of rare diseases on patients, families, and society
SRIA _ KPIs

SO1 - Generation of knowledge and its translation into medical and holistic intervention

• Number of publications resulting from RD research projects supported by the Partnership

• Number of funded RD projects

• Number of transitions from one phase in the value chain to the next

• Number of collaborations between academic researchers, industry, and patient advocacy organizations to develop and implement medical and holistic interventions for RD

• Number of RD research projects supported by the Partnership (or a previous co-fund on Rare Diseases) resulting in drugs approved by EMA/FDA, patents and new companies
SRIA _ KPIs

SO2 - Healthcare and research data are accessible, and used, for scientific and regulatory evaluation and healthcare delivery

- Number of healthcare and research data sources that are made available for scientific and regulatory evaluation and healthcare delivery

- Number of validated Patient-Centred Outcome Measures (PCOMs) included in the comprehensive data infrastructure based on FAIR principles.

- Number of clinical trials that are initiated or have progressed due to improved trial readiness and therapeutic options through FAIR data use

- Number of researchers, patients, and clinicians who are re-using and sharing rare disease data to implement multinational research, as evidenced by published research papers, patents, and collaboration agreements.

- Number of cases where healthcare and research data use led to a clinically or biomedically relevant outcome (interventions, diagnosing an undiagnosed case, new biomarker, new candidate drug for repurposing)
SRIA _ KPIs

SO3 - All activities empower, as equal partners, people living with rare disease

• Number of patients empowered, within the Partnership, through capacity-building and training activities related to research.

• Number of funded research projects that involve patients/patient organisations as co-designers.

• Number of guidelines developed with patients or patient organisations (when they exist) to support equitable patient inclusion in research, and their adoption by relevant stakeholders.

• Percentage of research studies that have involved patient representatives in their governance and decision-making structures.

• Number/percentage of research questions/interventions that have been informed by patient needs and preferences
SRIA _ KPIs

SO4 - Increased capacity and skills of RD stakeholders to optimise research to healthcare continuum

- Number of RD stakeholders who participate in training programs to enhance their research skills and capacity
- Number of national/regional training and education programs aligned with?/triggered by?/supported by? the RDP
- Number of train-the-trainer programmes implemented for capacity building at national level (including under-represented countries) and (as sub-indicators) number of trainers trained and of countries involved in such programmes (per year and per country)
- Total number of researchers who have participated in education/training programmes per year and per country
- Number of transdisciplinary research training programs developed and implemented at the European level.
SRIA _ KPIs

SO5 - Integrated multinational and multi-stakeholder Research & Innovation ecosystem for RD

• Number of active National Mirror Groups per year.

• Number of countries with sustainable national RD research strategies aligned with EU and international collaborations supported by the Partnership.

• Number of clinical trials conducted in multiple countries per year.

• Increase in the number of funding programs and initiatives dedicated to RD research and innovation, at both national and European levels.

• Number of complementarities and synergies established with other relevant programmes and initiatives.
Collecting information necessary for KPIs measurement and follow-up

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<tr>
<th>European Partnership [title]</th>
<th>Monitoring and evaluation framework, draft 1, [date]</th>
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<td>Overall vision: [max 500 characters]</td>
<td>What is a measure of success? Please use quantitative (Key Performance) and qualitative indicators, and link them to a point in time</td>
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<table>
<thead>
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<tr>
<td>GO2</td>
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<td>GO3</td>
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<td>SO4</td>
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<td>OO4</td>
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*add more lines, as needed.
6. Type of the organisation

- Research Funding organisation: 12
- Ministry: 0
- Research Performing organisation: 22
- Patient Advocacy Organisation: 16
- University: 54
- Hospital: 52
- Charity: 5
- Pharmaceutical industry: 5
- SME: 4
- Other: 15

9. Do you complete this survey:

- in your own name: 102
- in the name of your organisation: 36

31. My organisation is involved in the initiative(s) listed in the SRIA Annex 1

- Yes: 70
- No: 64

Position

- Director of the center
- Scientific Director
- Assistant Professor
- Professor
- Vice President
- Medical Director
- Clinical Director
- Senior Consultant
- Head of Department
- Manager of research
- research leader
- Associate Professor

Expertise

- Biomedical researcher
- Clinical researcher
- Clinician researcher
- Researcher in rare diseases
- Clinician and researcher
- researcher - molecular field
- research clinician
- Pediatric and research
- Pediatric and Biomedical researcher
- Primary care research
- Ultrasound and research
- Export patient
- Genetics and researchers
- Genetists and clinicians
- Disease Research
- Genetics
- genetics and clinicians
- Clinician research
- rare disorders
- patient research
10. **GO1**: Diagnosis established or enrollment in systematic research in average within 6 months after coming to medical attention

Do you agree with the proposed General Objective?

- Yes: 182
- No: 5

12. **GO2**: 1000 new therapies for rare diseases approved

Do you agree with the proposed General Objective?

- Yes: 125
- No: 11

14. **GO3**: Better understanding of the impact of rare diseases on patients, families and societies

Do you agree with the proposed General Objective?

- Yes: 130
- No: 5
SRIA Public Consultation

**Rating Outputs according to importance to the RD Research & Innovation landscape**

**SO1**

- Generation of knowledge and its translation into medical and holistic intervention

**SO2**

- Healthcare and research data are accessible, and used, for scientific and regulatory evaluation and healthcare delivery

**SO3**

- All activities empower, as equal partners, people living with rare disease

**SO4**

- Increased capacity and skills of RD stakeholders to optimise research to healthcare continuum

**SO5**

- Integrated multinational and multi-stakeholder Research & Innovation ecosystem for RD
SRIA Public Consultation

Rating **Outcomes** according to their **relevance** to the Specific Objective

<table>
<thead>
<tr>
<th>SO1</th>
<th>SO2</th>
<th>SO3</th>
<th>SO4</th>
<th>SO5</th>
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<tr>
<td>Generation of knowledge and its translation into medical and holistic intervention</td>
<td>Healthcare and research data are accessible, and used, for scientific and regulatory evaluation and healthcare delivery</td>
<td>All activities empower, as equal partners, people living with rare disease</td>
<td>Increased capacity and skills of RD stakeholders to optimise research to healthcare continuum</td>
<td>Integrated multinational and multi-stakeholder Research &amp; Innovation ecosystem for RD</td>
</tr>
</tbody>
</table>

**Not relevant**

**Relevant**
Examples:

- RD policy and capacity across countries needs to be addressed to ensure appropriate for implementation of research, unmet needs and treatment approvals

- Use AI technologies to train the trainers, especially without any language barrier issues is a good solution to help exchange of expertise, and transmit knowledge, ensuring a connection with all existing RD projects and initiatives

- Discrepancies between Patient Advocacy Groups across countries / disease areas needs to be addressed, e.g. some are supported by Governments and national societies

- The SRIA should also reference the EMA/HMA Big Data initiative, which DARWIN EU is part of.

- Generation of knowledge for rare diseases should be done though incentives. The current research career focused on academic production (papers, citation, awards, etc) is against working with rare conditions with some exceptions. Hence, new and different types of incentives would track the interest of researchers in rare diseases.
Proposal update
European Partnerships – Why?

**Horizon Europe** will support the next-generation of European Partnerships to **deliver on global challenges** through concerted R&I effort with the Member States, private sector, foundations and other stakeholders.

**Horizon Europe** expects partnerships to take a “**systemic approach in the achievement of the objectives**”, including “to ensure coordination with other relevant R&I initiatives”.

**European Partnerships**

- provide mechanisms to link R&I closely to policy needs
- develop close synergies with national and regional programmes
- bring together a broad range of innovation actors to work towards a common goal
- turn research into socio-economic results

e.g. IHI is an Institutionalised Partnership and RD Partnership is a co-funded Partnership
Overview of 49 candidate European Partnerships according to Horizon Europe structure

Partnership portfolio resulting from the Strategic Planning: priorities were discussed together with the Member States
WHAT ARE THE REMAINING NEEDS?

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

INFRASTRUCTURES
Orphanet, RD Connect, ERDRI, Solve-RD, EATRIS, ECRIN, BBMRI, etc.

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

PATIENTS NEEDS
EURORDIS

HEALTHCARE +
European Reference Networks (ERN)

RESEARCH ECOSYSTEM
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 in numbers

+2300 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

94 beneficiaries
10 hospitals
13 research institutes
31 research funding bodies/ministries
29 universities/hospital universities
5 EU infrastructures
5 charities/foundations
EURORDIS

+47 linked third parties
+100% associated networks

Coordinated by ALL 24 ERNs
Remaining needs

- 95% of RDs are disregarded in terms of research and lack effective treatment options.
- 50% of RD patients still do not have confirmed molecular diagnosis.
- 4 years - is an average time to be diagnosed when RD is known.
- 52% of RD patients and carers, RD translates into severe impact on their daily life.
Rare Diseases Partnership

Vision

**SUPPORT**
- robust patient need-led research

**DEVELOP**
- new treatments and diagnostic pathways

**UTILIZE**
- the power of health and research data and spearhead the digital transformational change in RD research and innovation

**SUPPORT**
- the coordination and alignment of national and regional research strategies, including the establishment of strong public-private collaborations
Rare Diseases Partnership

Mission

Bring supporting R&I services from across Europe under one roof so that every high-quality RD research project will benefit from cross-disciplinary expertise, goal-oriented study planning and efficient execution.

Enable every consenting patient living with a rare disease to be findable and enrolled in a suitable clinical study, by boosting generation of regulatory-level and FAIR-compliant data from diversity of sources, with the ultimate goal to fasten advances in prevention, diagnosis, disease knowledge and treatment.

Make Europe a global leader on rare disease research through a significant increase in investment to spur innovation, leading to job creation and improving EU competitiveness in R&I.
Capacity building

Data integration and coordination services

Ethics & regulatory Support

Mentoring service

Annual Joint Transnational Calls

Networking Support Scheme

Meaningful and exploitable results

Competitive Research Funding and Transversal Support Services

Acceleration hub

Networking Support

Patient Engagement Facilitation

Data integration and coordination services

Patient Engagement Facilitation

Ethics & regulatory Support

Mentoring service
Clinical Research Network

Innovative diagnostics
Natural History Studies
RWE generation
Identification & validation of PCOMs
Biomarkers & surrogate endpoints

Accelerated diagnosis
Clinical trial readiness
New therapies

National alignment & capacities
Clinical expertise & capacities
Public-private collaboration
Technological expertise
Infrastructure

Diagnostic research support
Biostatistical Guidance
COA/PCOM
Data Capitalization Hub
Ad-Hoc support
CT support
RD Partnership timeline

- End 2019: Validation of RD Partnership as part of the HE
- Jan 2021: first strategic meeting to discuss the concept
- Apr 2021: first official meeting with Member States
- Oct 2021: organisation of experts group to develop concept paper (180 experts)
- Feb 2022: publication of RDP concept paper (validated by the EC)
- Feb-Apr 2022: 30 national meetings to mobilise national resources
- May 2022: organisation of SRIA Task Force (80 experts) → May 2023 SRIA opened for public consultation
- Apr 2023: request for LOI from national & international stakeholders
- 5-6 of April 2023 – 1st writing phase meeting
  - 250 people, open to all, organisation in session, sharing of ideas to start forming the WPs
- 2nd of May 2023 – 2nd writing meeting
  - 150 people, open to representatives from interested organisations, 6 parallel sessions covering pre-defined groups of activities, proposition of WPs
- May 2023: organising working groups for each WP and start of the writing process
- June 1st: online meeting to present the first draft of the WPs + session dedicated to the discussion on governance
- June, writing continues
- 21-23 of June, in person writing meeting to finalise the complete draft of the proposal
- Summer time: finalisation of the admin aspects, feedback from the EC
- 18 of Sep: submission
More than 290 persons assigned to different WG

164 LoIs received

39 "group" meetings already took place!

€ 176 405 000 in-cash commitment

€287 633 474 In-kind commitment

19 theme co-leads supporting 6 main work streams
Final Organisation with Work Packages
Coordination & communication

<table>
<thead>
<tr>
<th>Coordination &amp; management</th>
<th>Communication</th>
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<tbody>
<tr>
<td>• <strong>Task 1.1</strong> – Project/Programme coordination</td>
<td>• <strong>Task 2.1</strong> – Communication and dissemination (C&amp;D) strategy</td>
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<tr>
<td>• <strong>Task 1.2</strong> – Governance &amp; strategy</td>
<td>• <strong>Task 2.2</strong> – C&amp;D Tactical plans &amp; coordination with partnering organisations to maximise outreach</td>
</tr>
<tr>
<td>• <strong>Task 1.3</strong> – Monitoring of RDP activities</td>
<td>• <strong>Task 2.3</strong> – Communication tools and support to Work Packages</td>
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<tr>
<td>• <strong>Task 1.4</strong> – Data Management Plan</td>
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<td>• <strong>Task 1.5</strong> – Sustainability strategy</td>
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<td>• <strong>Task 1.6</strong> – Ethics compliance</td>
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<td>• <strong>Task 1.7</strong> – IPR management</td>
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</table>

Activities proposed in this figure are not exhaustive
Joint transnational calls for collaborative research projects

- **Tasks 3.1** – Topics selection and definition of eligibility criteria
- **Task 3.2** – Joint Transnational Call implementation
- **Task 3.3** – Engagement of patients

Clinical trial call management

- **Tasks 4.1** – Develop the call framework
- **Task 4.2** – Open the call and select trials for funding
- **Task 4.3** – Project implementation, project monitoring and financial management

Networking to share knowledge

- **Tasks 5.1** – Preparation and launching of the scheme
- **Task 5.2** – Evaluation of the selected proposals after each collection date and funding decision
- **Task 5.3** – Quality management

Activities proposed in this figure are not exhaustive
<table>
<thead>
<tr>
<th>Data Readiness</th>
<th>Genome re-analysis research pipeline</th>
<th>Genomic innovation to shorten time to diagnosis</th>
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</thead>
<tbody>
<tr>
<td><strong>Task 6.1</strong> – Coordinate Pan-European diagnostic research data readiness and collation effort</td>
<td><strong>Task 7.1</strong> – Exome and genome re-analysis pipeline coordination and monitoring</td>
<td><strong>Task 8.1</strong> – Enable access of complete genome sequencing for RD in underrepresented countries</td>
</tr>
<tr>
<td><strong>Task 6.2</strong> – Data standardisation, submission and harmonisation</td>
<td><strong>Task 7.2</strong> – Standardised exome and genome re-analysis beyond state-of-the-art diagnostics</td>
<td><strong>Task 8.2</strong> – Enable complete genome sequencing and analysis for RD to shorten time to diagnosis</td>
</tr>
<tr>
<td><strong>Task 6.3</strong> – Data archival and data access</td>
<td><strong>Task 7.3</strong> – Novel gene and mechanisms discovery through genome re-analysis</td>
<td><strong>Task 8.3</strong> – Complete genome mapping for RD to shorten time to diagnosis</td>
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<td><strong>Task 7.4</strong> – Develop and leverage knowledge for variant interpretation</td>
<td><strong>Task 8.4</strong> – New genomics/transcriptomics analysis capabilities to understand genetic variation in RD</td>
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<td><strong>Task 7.5</strong> – Translation to clinic</td>
<td><strong>Task 8.5</strong> – Multi-omics data integration to shorten time to diagnosis in RD</td>
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Activities proposed in this figure are not exhaustive
Real World Data

- **Task 9.1** – Use of primary healthcare data (EHRs) for RD outcome research
- **Task 9.2** – Use of population-based data for RD outcome research
- **Task 9.3** – Integration of patient cohorts for natural history/standard-of-care reference studies
- **Task 9.4** – Development of a blueprint and inventory of regulatory-grade natural history cohort data
- **Task 9.5** – Disease progression modelling and prognostic biomarker research
- **Task 9.6** – Development of a model-based clinical trial simulation platform for rare diseases

Clinical Outcome Assessment

- **Task 10.1** – Platform for patient-focused outcome development and validation
- **Task 10.2** – Development and Implementation of Clinical Outcome Assessment Tools
- **Task 10.3** – Unveiling the Hidden Burden: Estimating the Socioeconomic Impact of Rare Diseases for Informed Decision Making and Resource Allocation

Activities proposed in this figure are not exhaustive
Advanced Therapeutic Medicinal Products

• **Task 11.1** – Identify and rank disease indications requiring ATMPs
• **Task 11.2** – Match technical development with prioritised needs
• **Task 11.3** – Elaborate Proof of Concept studies to test the development pipeline

N-of-few approach

• **12.1** – Academic Platform for Tailored Antisense Oligonucleotide Therapies

Activities proposed in this figure are not exhaustive
## CLINICAL RESEARCH NETWORK _ Task Forces

<table>
<thead>
<tr>
<th>TF Objectives</th>
<th>Drug Trials</th>
<th>Non-pharmacological interventions</th>
</tr>
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</table>
| • Inform the RD Partnership to prepare for Clinical Studies  
  • Capitalize on the RD Partnership activities (e.g., from the CRN outcome research studies)  
  • Provide Guidance for investigator initiated trials.  
  • Identify the list of criteria for trials proposals (beyond Orphan Drug designation) | • Protocol design guidance  
• Modeling & simulation plan (study design optimisation)  
• Measures to optimise recruitment  
• Best practice & working procedures novel sampling approach  
• Standardisation of efficacy and safety data collection (including paediatric-specific variables/meta-data.)  
• Approaches to study preparedness | • Review and analyze the current landscape (current CT, developments and regulatory aspects)  
• Provide guidance for non-pharmacological therapy development (incl. Unmet technical and functional needs, patient involvement, academics/SME)  
• Stimulate further development of non-pharmacological therapy research & development (research proposal criteria e.g., SSH, HTA; mapping experimentation facilities; network of developers) |

Activities proposed in this figure are not exhaustive
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# Expertise Services Hub

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<tr>
<th>Mentoring and Consultancy</th>
<th>Regulatory Support</th>
<th>Methodological Support</th>
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<tbody>
<tr>
<td><strong>Task 17.1</strong> - Execution of the Mentoring Program</td>
<td><strong>Task 18.1</strong> - Regulatory support to preclinical research</td>
<td><strong>Task 19.1</strong> - Knowledge transfer towards the local clinical trial teams</td>
</tr>
<tr>
<td><strong>Task 17.2</strong> - Consultancy Services</td>
<td><strong>Task 18.2</strong> – Regulatory support to clinical research</td>
<td><strong>Task 19.2</strong> - Novel methodologies for the use of all available knowledge, including Real World Data</td>
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<td><strong>Task 19.3</strong> – Data analysis methodologies when data are multivariate, hierarchical, incomplete and of differing data types</td>
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<td><strong>Task 19.4</strong> - Non-paramedic and randomized-based methodology</td>
</tr>
</tbody>
</table>
Education & training in RD research

Education and Training in RD research

• Task 20.1 – Patients and young researchers’ trainings

• Task 20.2 - Identification and fulfilment of training needs

• Task 20.3 - RD research training for multistakeholder communities

• Task 20.4- European Curriculum on RD research

Activities proposed in this figure are not exhaustive
# Acceleration Hub

## Innovative therapies technology accelerator

- **Task 21.1** – rAAVs  
- **Task 21.2** – mRNA  
- **Task 21.3** – Synthetic nanoparticles or extracellular vesicles  
- **Task 21.4** – Gene Editing  
- **Task 21.5** – Therapy response and immunogenicity

## Public-private collaboration accelerator

- **Task 22.1** – Setting up the marketplace  
- **Task 22.2** – Acceleration readiness  
- **Task 22.3** – Asset profiling and development  
- **Task 22.4** – Matchmaking and marketing

*Activities proposed in this figure are not exhaustive*
(Inter)national capacity alignment

### National Mirror Groups promotion & national alignment
- Task 23.1 - Fostering creation of National Mirror Groups
- Task 23.2 - Deployment and operations of National Mirror Groups
- Task 23.3 - Animation of National Mirror Groups synergies

### Fostering engagement of underrepresented countries
- Task 24.1 - Promoting capacity development actions
- Task 24.2 - Undertaking advocacy and awareness efforts to UCs added value
- Task 24.3 - Support actions to improve UC participation in all RDP activities
- Task 24.4 - Mobility actions for UCs

### RDP Global collaboration
- Task 25.1 - Establish Strategic Alliances with European and International Programmes, Projects, Initiatives and Universities.
- Task 25.2 - Support to IRDIRC activities and dissemination of outcomes.
- Task 25.3 - Promote the International Dimension of the Clinical Research Network of RDP by building global networks among clinical research networks and patient organizations.

Activities proposed in this figure are not exhaustive
European Rare Diseases Partnership
Pathways as an RD Ecosystem
RDP MISSION
Bring supporting R&I services from across Europe under one roof so that every high-quality RD research project will benefit from cross-disciplinary expertise, goal-oriented study planning and efficient execution.

**Project results**
- Acceleration Hub (market place, follow on funding, industry incubators)
- Dissemination & communication

**Project execution**
- Education & training
  - Expertise (mentoring & consultancy)
  - Regulatory advice
- Data tools (e.g. analysis)

**Build networks**
- Networking to share knowledge

**Build proposal**
- Expertise Support Hub
- Data Support Hub
- Technology Accelerator

**Obtain funding**
- Joint transnational calls
- Clinical trials
RDP MISSION
Enable every consenting patient living with a rare disease to be findable and enrolled in a suitable clinical study, by boosting generation of regulatory-level and FAIR-compliant data from diversity of sources, with the ultimate goal to fasten advances in prevention, diagnosis, disease knowledge and treatment.

**Plan study**
- Expertise service hub (CT study design, regulatory, methodology)
- CRN CT simulation model(s)
- Innovative therapies pipeline (demonstrators & trial readiness)

**Implement CT**
- Drug trials, ATMPs and non-pharmacological
- Collaborative networks (C4C, industry...)

**Accelerate diagnosis**
- CRN diagnostic work stream

**Collect FAIR data**
- Data service hub
- JTC funded projects
- CRN (population & RD registries data)

**Analyse & (re)use FAIR data**
- Data service hub
- CRN (outcome research: RWE, PCOMs...)
RDP MISSION
Make Europe a global leader on rare disease research through a significant increase in investment to spur innovation, leading to job creation and improving EU competitiveness in R&I

New investments
- Expansion to other funding schemes
- Direct involvement of industry, charities, etc. as potential investors

Innovative results
- Profit from data “in one place”
- Profit from at hand regulatory expertise
- Profit from technology developments “validated” by industry

Strategic investment & national alignment
- JTCs & CRN activities
- Strategy of RDP

Training & education
- For new generations
- To increase the capacity & knowledge of more advanced stakeholders
- To increase the quality of delivered research

Assist projects
- Expertise of RDP
The place of Rare Diseases Partnership in the overall RD landscape
Expert Group on support of the Strategic Coordinating Process

Mandate 2022-24
Our mission:

The **Expert Group for support of the Strategic Coordination process for Partnerships** was entrusted with three tasks:

• Prepare evidence-based independent advice to the Commission on the development of the portfolio of European Partnerships, taking into account emerging R&I priorities, common challenges and EU political priorities that require orchestrated large-scale investments → currently being finalised

• Support the drafting of the 2024 edition of the BMR on partnerships, and engage with European Partnerships, Member States (MS) and Associated Countries (AC) in the preparation of the report, including data collection → preparatory process started

• Develop recommendations for next steps and required support for the strategic coordinating process → forthcoming
Integrated approach for assessing the landscape of European Partnerships

A. Forward-looking analysis of issues
1. Identification of sources
2. Characterisation of sources
3. Review of sources – identification of FLIs
4. Extraction of tech areas
5. Validation of FLIs and tech areas with EPs
6. Matching FLIs and tech areas with EP profiles
7. Prioritisation of matching results

B. Review of the legal basis criteria
1. Review of legal basis
2. Clarification/update of criteria
3. Application of criteria

C. Review of portfolio management (PM) criteria
1. Review of PM resources
2. Clarification/update of criteria
3. Application of criteria

D. Assessment of EPs’ appropriateness/potential to address the area(s)

Consultation with EPs and them. experts

C. Identification of areas less addressed or gaps

Consultation with EPs and them. experts

Assessment of EPs as instrument in relation to area(s)

WS with EC and EPs

Guidance and input on:
- The need for a new partnership
- The need to continue partnerships:
  - In their current form and resources (“Keep”)
  - Upscale ("Keep and increase investment")
  - Downscale ("Keep and decrease investment")
  - Merge or split decisions ("Keep and reconfigure")
- The need to disinvest / exit
C. Review of portfolio-management criteria

Global overview of the relevance of the proposed dimensions.
Source: Expert Team analysis (2023)

<table>
<thead>
<tr>
<th>Dimension / criterion</th>
<th>Priority</th>
</tr>
</thead>
<tbody>
<tr>
<td>Agility and flexibility</td>
<td>High</td>
</tr>
<tr>
<td>Synergies (internal)</td>
<td>High</td>
</tr>
<tr>
<td>Synergies (external)</td>
<td>High</td>
</tr>
<tr>
<td>Market Readiness</td>
<td>High</td>
</tr>
<tr>
<td>TRL evolution</td>
<td>High</td>
</tr>
<tr>
<td>IP generation</td>
<td>High</td>
</tr>
<tr>
<td>Sunk costs / irreversibility</td>
<td>Medium</td>
</tr>
<tr>
<td>Trust and stakeholder flexibility</td>
<td>Medium</td>
</tr>
<tr>
<td>Scale</td>
<td>Medium</td>
</tr>
<tr>
<td>Diversified portfolio management</td>
<td>Low</td>
</tr>
<tr>
<td>Monitoring and evaluation</td>
<td>Low</td>
</tr>
</tbody>
</table>

The set suggested in this report can be the basis which can be reviewed/refined/updated depending on the specific aims of the assessment task and the type of partnerships targeted.

When addressing an identified gap, where there are no partnerships already or which is less addressed by the existing ones, desk research will not suffice due to lack of information.

It is advisable to apply the methods suggested under each of the portfolio management criteria as described in Chapter 2.2 and more analytically in Annex 2 of the report.
36 Programmes / institutions / schemes identified with potential relevance for collaboration with the RD Partnership

→ Includes several other EU programmes such as:
  - Horizon Europe
  - EU4Health
  - EU Mission Cancer
  - European Innovation Council
  - Digital Europe Programme
  - European Regional Development Fund
  - European Social Fund Plus (ESF+)
  - Invest EU
FUNDING

ACCELERATOR HUB / Innovation

EDUCATION & TRAINING

DATA SERVICE HUB

CRN (including Healthcare pathway, diagnostic)
THANK YOU

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