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

Table of Contents

List of Abbreviations

AOB	Any Other Business
AREB	Advisory Regulatory Ethics Board
AWP	Annual Work Plan
CA	Consortium Agreement(s)
Coo	[EJP RD] Coordination [Team]
CRN(s)	Clinical Research Network(s)
EC	European Commission
ECTS	European Credits Transfer System
EJP RD	European Joint Programme on Rare Diseases
EMA	European Medicines Agency
ERICA	European Rare Disease Research Coordination and Support Action consortium
ERN(s)	European Reference Network(s)
EUPID	European Patient Identity Management
ExCom	Executive Committee
GB	Governing Board
IHI	Innovative Health Initiative
IMI	Innovative Medicines Initiative
IMT	Innovation Management Toolbox
JTCs	Joint Transnational Calls
KPI	Key Performance Indicator
NMG	National Mirror Group
NSS	Networking Support Scheme
ODDG	Orphan Drug Development Guidebook
P1	Pillar 1
P2	Pillar 2
P3	Pillar 3
P4	Pillar 4
PB	Policy Board
PM	Person Month
RD	Rare Disease
SMEs	Small and Medium-sized Enterprises
SRIA	Strategic Research and Innovative Agenda
VP	Virtual Platform
WP	Work Package

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

Name	Institution	Role	Presence
Daria Julkowska	INSERM	coordinator WP1 - WP5	Present
Ralph Schuster	DLR	P1 coleader WP6	Present
Sonja van Weely	ZonMw	P1 coleader WP7	Present
Ana Rath	INSERM (Orphanet)	P2 coleader WP10 -WP11	Present
Franz Schaefer	UKL-HD	P2 coleader WP13	Online
Roseline Favresse	EURORDIS	P3 coleader WP15 - WP18	Present
Biruté Tumiene	VUHSK	P3 coleader WP18	Online
Anton Ussi	EATRIS	P4 coleader WP3 - WP19	Online
Rima Nabbout	AP-HP	P4 coleader WP20	Online
Eva Bermejo-Sanchez	ISCIII	WP2 and WP3 coleader	Present
Annalisa Landi	FGB	WP4	Present
Viviana Giannuzzi	FGB	WP4	Present
Barbara Sanavio	FTELE	WP4 - WP19	Present
Christine Fetro	FFRD	WP8	Present
Irit Allon	CSO-MOH	WP9	Online
Anthony Brookes	ULEIC	WP10 - WP12	Present
Sergi Beltran	CNAG-CRG	WP11	Online
Luca Zalatnai	CNAG-CRG	WP11	Online
Marco Roos	LUMC	WP12	Online
Chris Evelo	UM	WP13	Present
Friederike Ehrhart	UM	WP13	Present
Claudio Carta	ISS	WP14	Online
Magda Granata	FFRD	WP16	Present

Name	Institution	Role	Presence
Krystyna Chrzanowska	IPCZD	WP18	Present
Ralf-Dieter Hilgers	UKA	WP20	Online
Liron Even-Faitelson	CSO-MOH	WP9	Online
Maria Del Carmen Sanchez Gonzalez	ISCIII	WP2 - WP3	Present
Florence Guillot	ANR	WP6	Online
Ben Lydall	EATRIS	WP3	Excused
Catherine Nguyen	INSERM	IT GGB director	Present
Juliane Halftermeyer	INSERM-Transfert	Coo team	Present
Ngangta Mbaidoum Abanga	INSERM	Coo team	Present
Blandine Castrillo	INSERM	Coo team	Present
Yanis Mimouni	INSERM	Coo team	Present
Galliano Zanello	INSERM	Coo team	Present
Alexandra Tataru	INSERM	Coo team	Present
Marie-Catherine Letinturier	INSERM	Coo team	Present
Tanguy Onakoy	INSERM	Coo team - WP5	Present
Pauline Adam	INSERM	Coo team	Present
Hiba Abou Daya	INSERM	Coo team	Online
Clement Moreau	INSERM	Coo team	Present
Madeline Chatel	INSERM	Coo team	Present

Agenda

11:30 – 11:45	**Welcome from coordination**	EJP RD Coordination
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.**

→ **P4 last annual retreat should be organised back to back with the final EJP RD consortium meeting (date not yet defined) to optimise the costs.**

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 "EJPRD_PB-GB-meeting_July2023-All-slides"*

List of participants

Name	Board	Country	Presence
Alexandra Tataru	Coo	France	Present
Ana Rath	ExCom	France	Present
Annalisa Landi	ExCom	Italy	Present
Anthony Brookes	GB / ExCom	United Kingdom	Present
Anton Ussi	ExCom	The Netherlands	Online
Avi Israeli	PB	Israel	Online
Barbara Sanavio	ExCom	Italy	Present
Biruté Tumiene	ExCom	Lithuania	Present
Blandine Castrillo	Coo	France	Present
Chris Evelo	ExCom	The Netherlands	Present
Christian Jervelund	Invited [Copenhagen Economist]	Denmark	Online
Christina Kyriakopoulou	EC	[EC]	Present
Christine Fetro	ExCom	France	Present
Claudio Carta	ExCom	Italy	Online
Clement Moreau	Coo	France	Present
Daria Julkowska	Coo	France	Present
Etienne Richer	GB	Canada	Present
Eva Bermejo-Sanchez	ExCom	Spain	Present
Florence Guillot	ExCom	France	Online

Franz Schaefer	ExCom	Germany	Present
Friederike Ehrhart	ExCom	The Netherlands	Present
Galliano Zanello	Coo	France	Present
Guenter Schreier	GB	Austria	Online
Helene Le Borgne	PB	[EC]	Present
Hiba Abou Daya	Coo	France	Online
Irit Allon	ExCom	Israel	Online
Juliane Halftermeyer	Coo	France	Present
Kateryna Kratzer	GB	Canada	Online
Kristien Peeters	GB	Belgium	Present
Krystyna Chrzanowska	ExCom	Poland	Present
Lærke Kilsdal	Invited [Copenhagen Economist]	Denmark	Online
Liron Even-Faitelson	ExCom	Israel	Online
Madeline Chatel	Coo	France	Present
Magda Granata	ExCom	France	Present
Malwina Mejer	Invited [Copenhagen Economist]	Denmark	Present
Marco Roos	ExCom	The Netherlands	Online
Maria Del Carmen Sanchez Gonzalez	ExCom	Spain	Present
Marie-Catherine Letinturier	Coo	France	Present
Ngangta Mbaidoum Abanga	Coo	France	Present
Onur Burak Dursun	PB	Turkey	Online
Patricia Maciel	PB	Portugal	Present
Pauline Adam	Coo	France	Present
Ralph Schuster	GB / ExCom	Germany	Present
Ralf-Dieter Hilgers	ExCom	Germany	Online
Rima Nabbout	ExCom	France	Online
Rita Cavaleiro	GB	Portugal	Online
Roseline Favresse	ExCom	France	Present
Sean Sapcariu	GB	Luxembourg	Online
Sergi Beltran	ExCom	SPain	Online
Simona Bifolchi	PB	Italy	Online
Sonja van Weely	ExCom	The Netherlands	Present
Tanguy Onakoy	Coo	France	Present
Tonia Bieber	PB	Germany	Online

Viviana Giannuzzi	ExCom	Italy	Present
Yanis Mimouni	Coo	France	Present
Živilė Ruželė	GB	Lithuania	Online

Agenda

9:30 – 9:40	**Welcome from coordination**	EJP RD Coordination
9:40 – 10:40	**EJP RD – Sustainability**	Copenhagen Economics & All
10:40 – 10:55	*Break*	
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 • Feeback 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.

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.

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.

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)

EUROPEAN JOINT PROGRAMME
RARE DISEASES

ExCom meeting
03/07/2023, Bruxelles - Belgium

Annual Work Plan Year 6

Presentation of planned activities

EUROPEAN JOINT PROGRAMME RARE DISEASES

COORDINATION, TRANSVERSAL ACTIVITIES & COMMUNICATION

EUROPEAN JOINT PROGRAMME
RARE DISEASES

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.

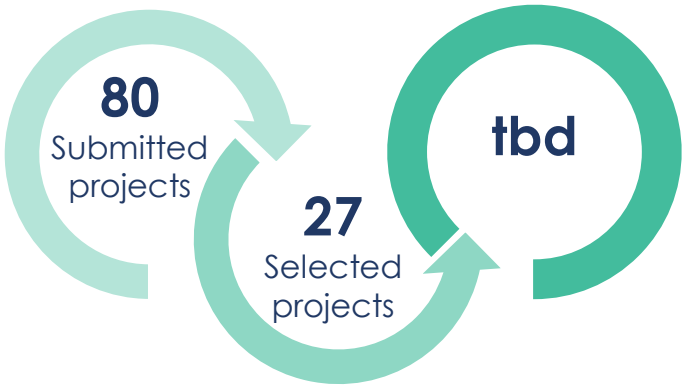
PILLAR 1
FUNDING OPPORTUNITIES
Plans for Year 6

EUROPEAN JOINT PROGRAMME
RARE DISEASES

WP6 Joint Transnational Calls for collaborative research projects

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

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 lessions learned for all JTC calls to further adapt call documents and procedures for future calls.

EJP RD

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

Public-private projects

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

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

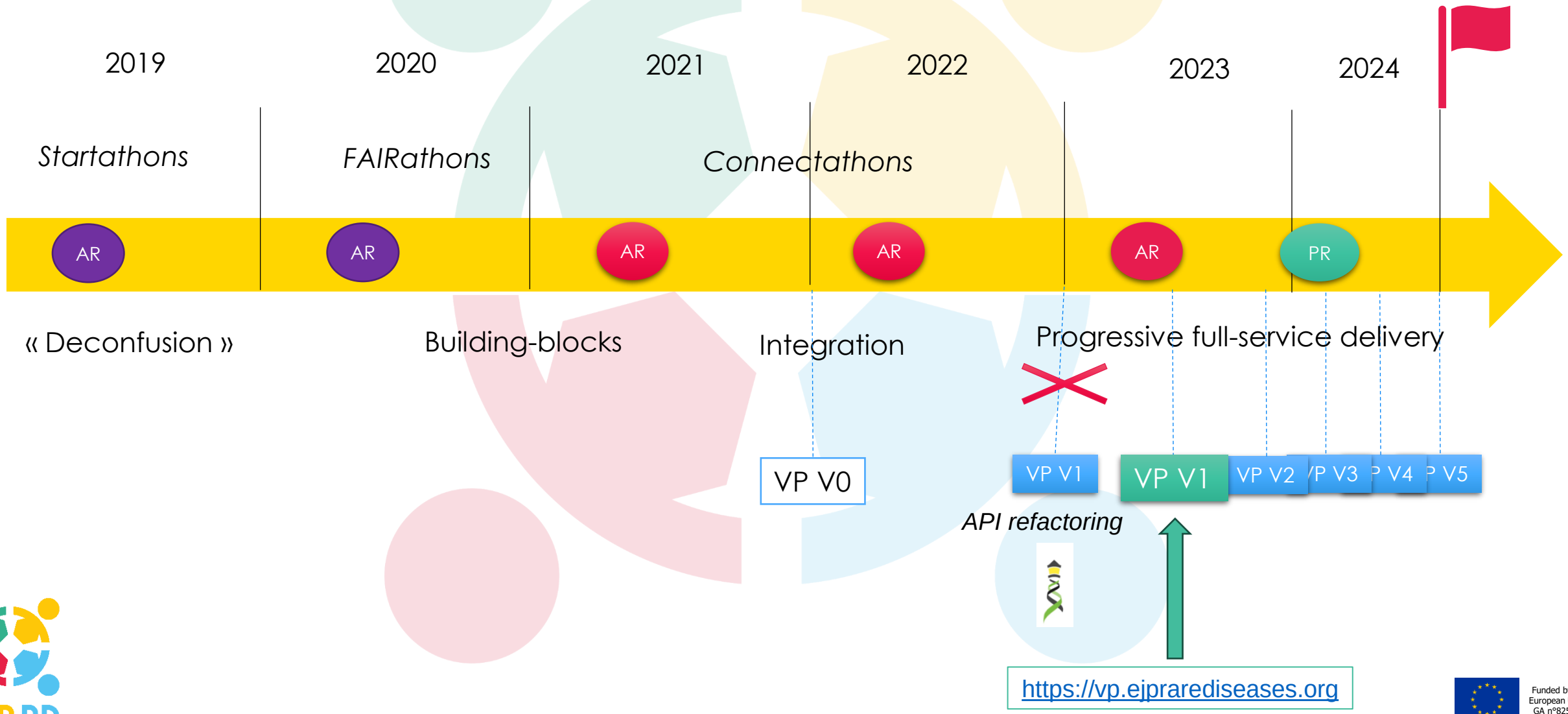
EJP RD Virtual Platform

Plans for Year 6

Ana Rath, Franz Schaefer, and Pillar 2 WP and WF Leaders

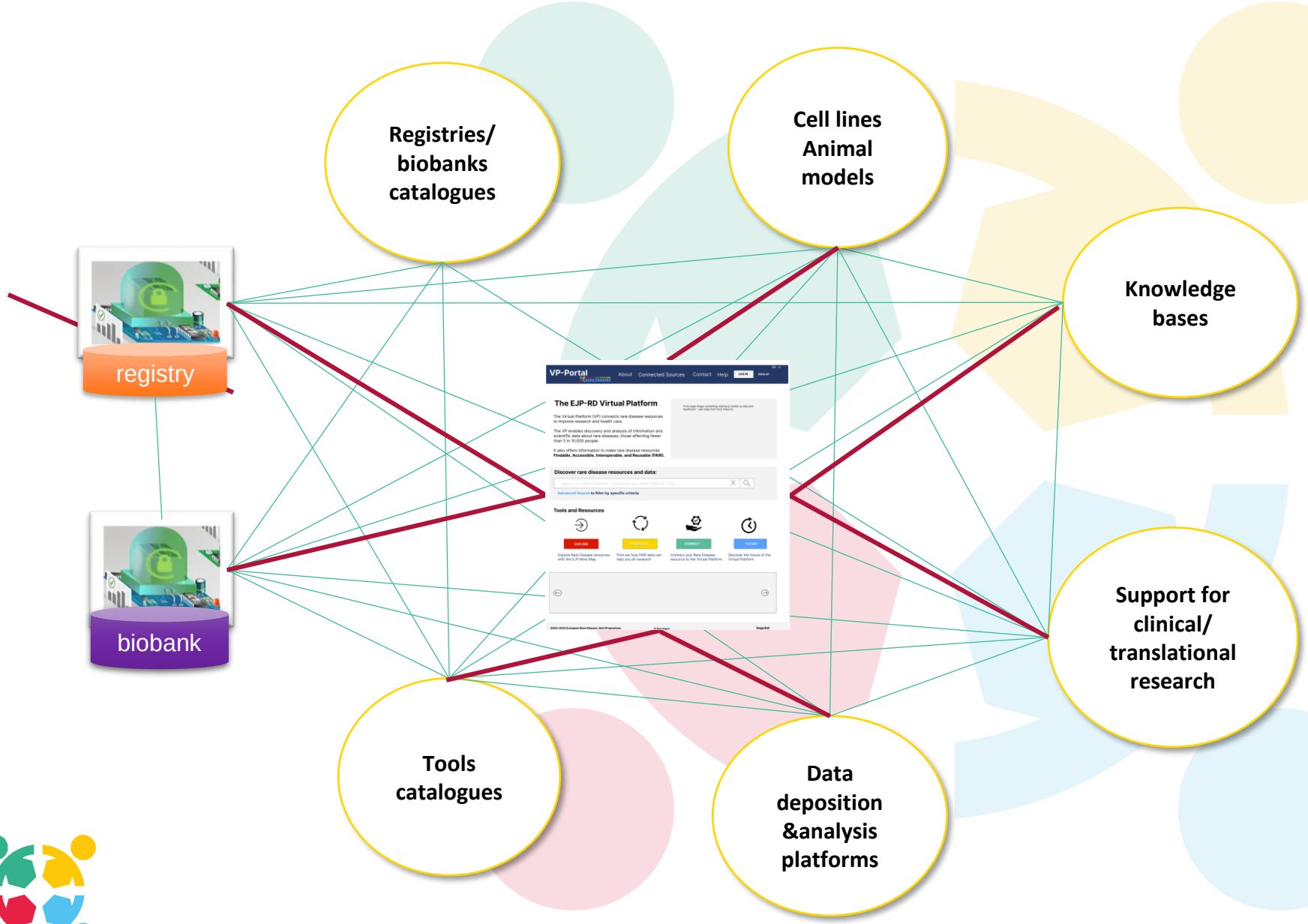
EUROPEAN JOINT PROGRAMME
RARE DISEASES

Pillar 2 evolution: long learning curve now paying



Funded by the European Union
GA n°825575

The Virtual Platform: a network of federated resources:



Decreasing fragmentation

Increasing interoperability

Increasing RD-readiness

through

Harmonisation

Standardisation

in a flexible way:

Common methodologies, Multipe technical solutions

V1: https://vp.ejprarediseases.org

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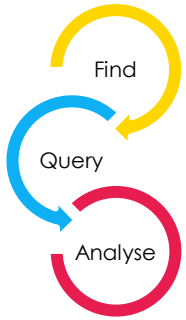
Onboarding at diverse levels of connection to the Virtual Platform



Onboarding guidance document (ongoing)

Onboarding F2F and follow-up workshops

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

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EUROPEAN JOINT PROGRAMME RARE DISEASES VP-PORTAL

RESOURCE DISCOVERY VP NETWORK RESOURCES LOGIN

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
Explore Rare Disease resources with the EJP Mind Map

GUIDANCE
Find out how to make your data more FAIR

CONNECT
Contact us for information or feedback

2020-2022 European Rare Diseases Joint Programme

EUROPEAN JOINT PROGRAMME RARE DISEASES

This work is supported by the funding from the European Union's Horizon 2020 research and innovation programme under the EJP RD COFUND-EJP N° 825575

Legal Notice

V1

- 21 resources connected
  - 11 registries
  - 3 catalogues
  - 2 genome-phenome deposition infrastructures
  - 5 knowledge bases

EJP RD

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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 expande RD pathways creation and analysis based on case studies, and making them findable through the Virtual Platform

EJP RD

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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 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

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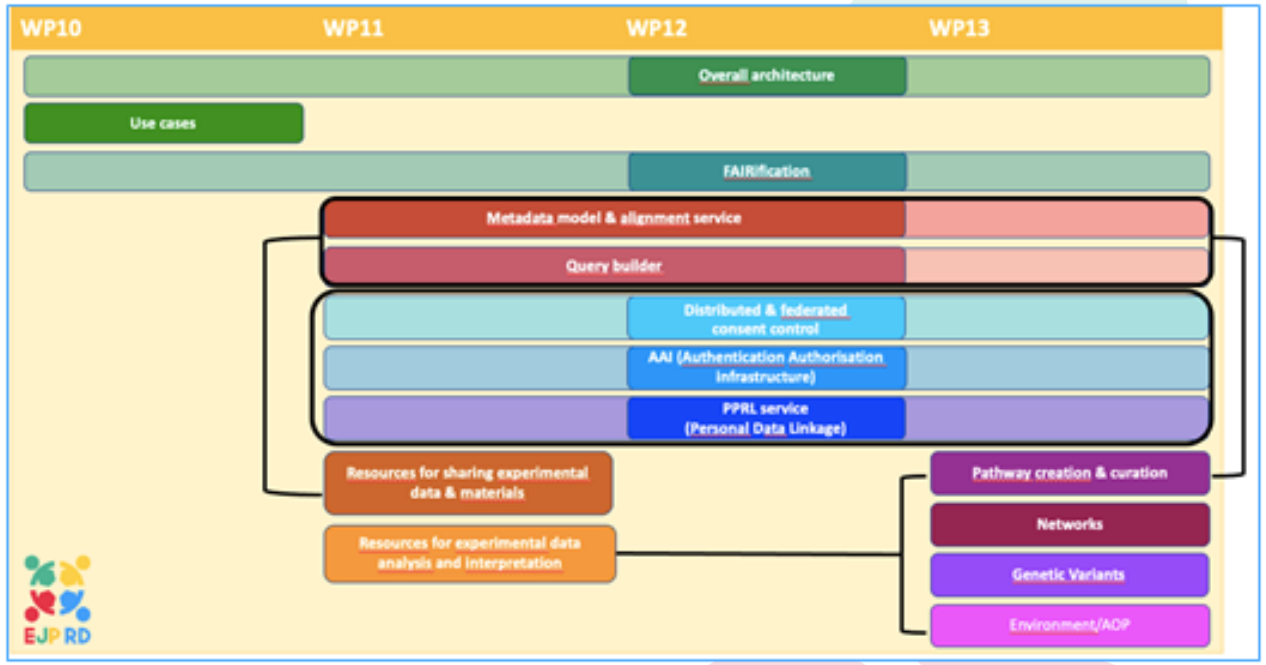
WP10- Reinforce the integration among teams to more efficient delivery



Overall architecture & technical oversight

Metadata & data modelling

FAIRification & onboarding

Access, Use, and Consent Conditions Control

Privacy Preserving Record Linkage

L1&L2 VP query
Reference Implementation

L3&L4 VP analysing
Demonstration of readiness

Resources for Experimental Data Analysis and Interpretation

Joint Collaborative Analysis

Sustainability, Quality, and GDPR Assessment

38 ST (WP11-12-13) → 14 WF → 9/10 WF

EJP RD

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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

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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

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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

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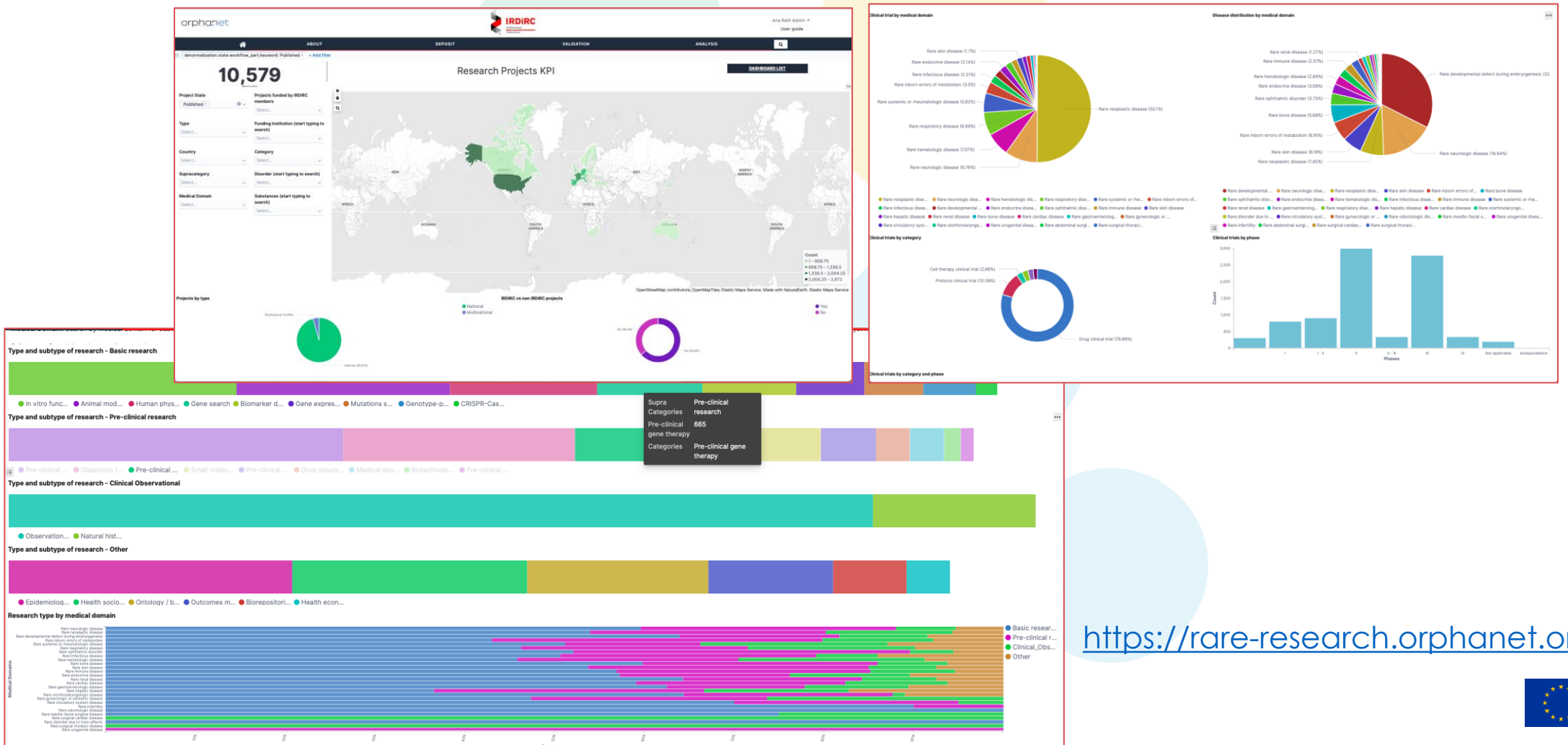
WP11- IRDiRC FCC

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



https://rare-research.orphanet.org

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WP12

- Finalize the consolidation of model alignment services (semantic mappings, sematic model aligments 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

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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

EJP RD

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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

EJP RD

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TRAINING & EDUCATION

EUROPEAN JOINT PROGRAMME
RARE DISEASES

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;

EJP RD

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.

EJP RD

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 disease***" 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.

EJP RD

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.

EJP RD

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).

EJP RD

ACCELERATED TRANSLATION OF RESEARCH RESULTS & CLINICAL TRIALS

EUROPEAN JOINT PROGRAMME
RARE DISEASES

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.

EJP RD

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

EJP RD

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)

EJP RD

Annual Work Plan Year 6

Budget and Prioritisation

EUROPEAN JOINT PROGRAMME RARE DISEASES

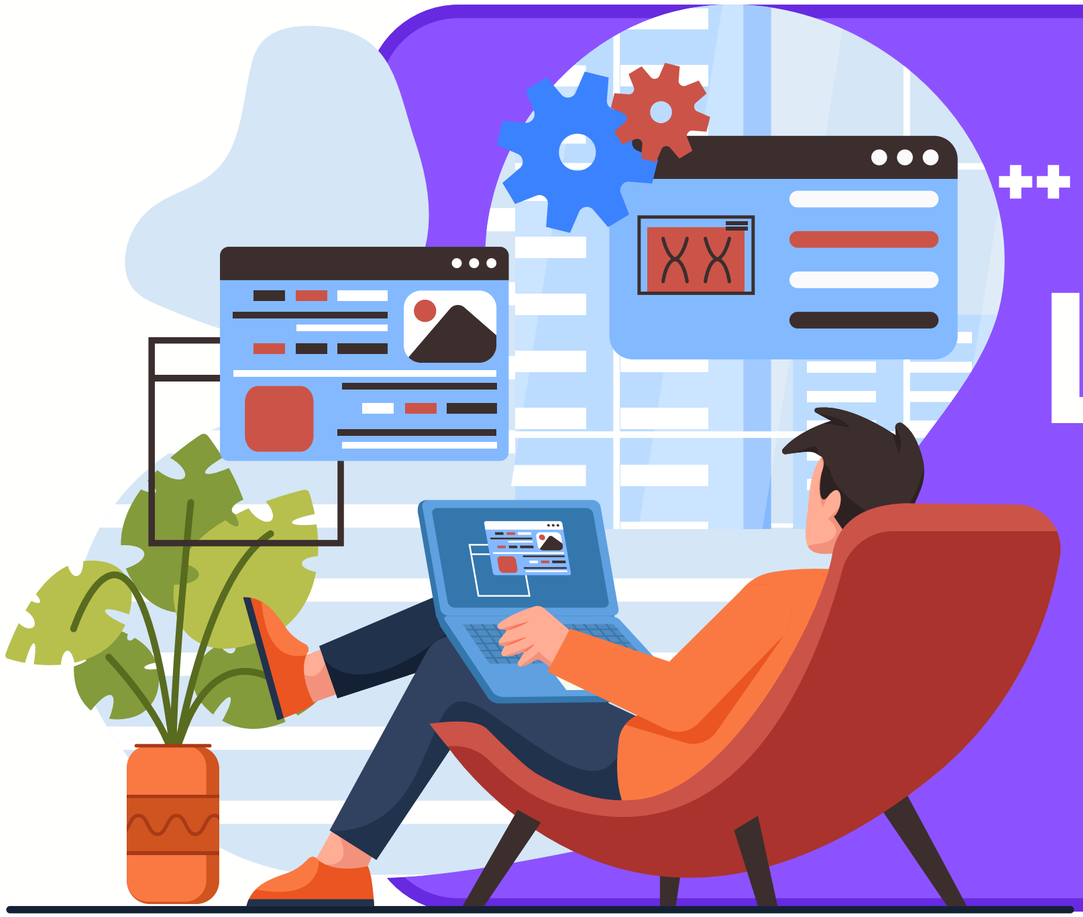
Communication strategy for the last year of EJP RD

EUROPEAN JOINT PROGRAMME RARE DISEASES

Tanguy Onakoy
WP5

ExCom Meeting
3 July 2023
Brussels, Belgium

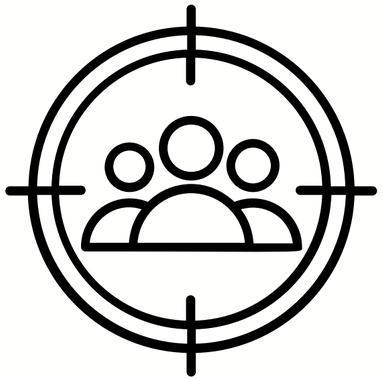
Communication Strategy



Last year of EJP RD

Objectives and strategy

Communication & Dissemination on the results & impact of EJP RD

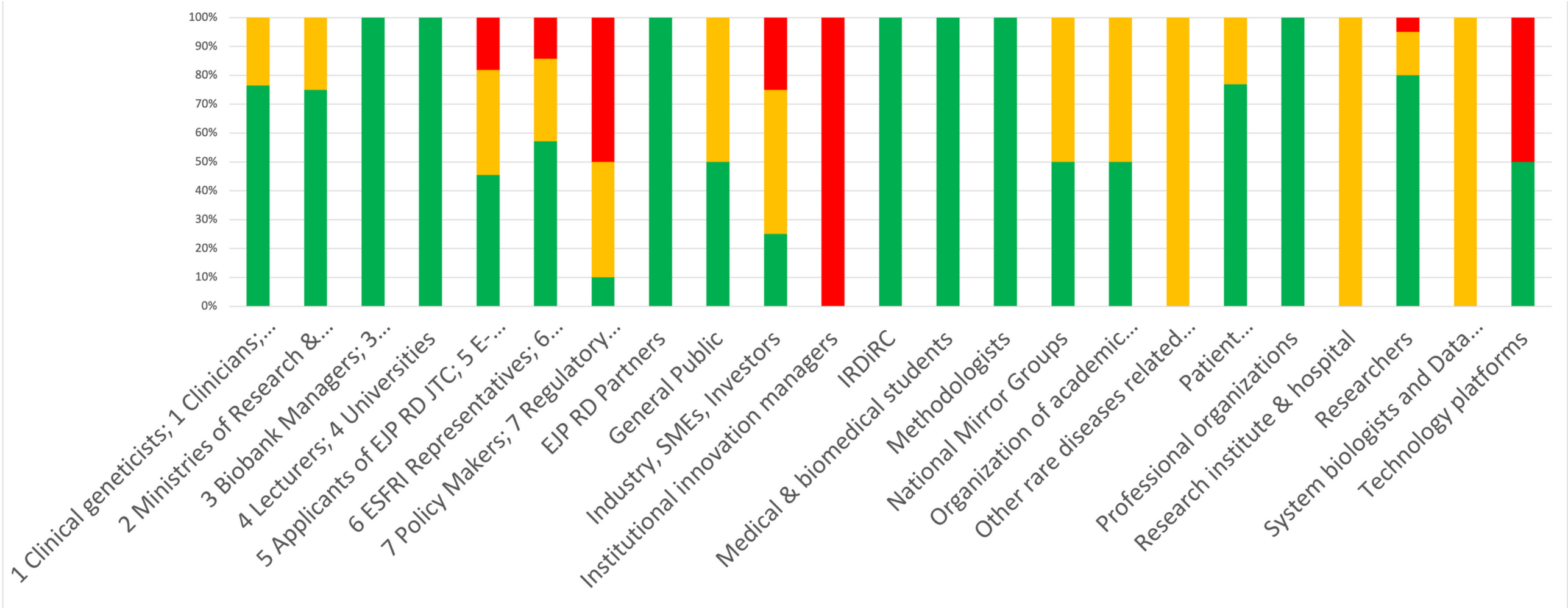


Reach the missing targeted audience in addition to the current reached audience

Rare Disease Day campaign

Current status reached stakeholders

(initially targeted)



Reached

Partially Reached

Not Reached

Standard communication actions per campaigns

Social Media Message

Visual

Newsletter article

Website news & slider (when appropriate)

Email to targeted audience

Addition in Year 6: Sponsored Targeted Posts!

PILLAR 0

Coordination & Management

WP	Action/Activity	Channel	Timeline	Target Audience
1	Internal news dedicated to EJP RD Partners	Internal Newsletter	Every other month	EJP RD Partners
1	MsTeams Posts	MsTeams	Continuous	EJP RD Partners
1	Helpdesk	Website	Continuous	All Stakeholders
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

PILLAR 0

Coordination & Management

WP	Action/Activity	Channel	Timeline	Target Audience
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	

PILLAR 0

Coordination & Management

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

Fundings and Calls

WP	Action/Activity	Channel	Timeline	Target Audience
6 - 7 - 8 - 9	Journalistic report for selected projects written in lay language for general public	Website + Standard communication procedure	July 2023 - August 2024	All stakeholders

PILLAR 2

Coordinated Access to Data and Services

WP	Action/Activity	Channel	Timeline	Target Audience
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 Event (Workshop/Webinar)	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

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

PILLAR 3

Training and Empowerment

WP	Action/Activity	Channel	Timeline	Target Audience
14 - 15 - 16 - 17 - 18	Communication campaign for trainings	Standard communication procedure	Continuous	• Academic researchers • Medical & biomedical students • Industry researchers/scientists • Industry • Lecturers • University managers • Professional organizations • Organizations of academic institutions • Biobank managers • Laboratory scientists & technicians • Clinical geneticists
		Joint campaign with EURORDIS dedicated to patients/patients advocacy groups	Continuous	• Rare disease patients and representatives • ePAGs (RD patient rep involved in • ERNs)
		Joint campaign with EURORDIS dedicated to patients/patients advocacy groups	Continuous	Young Public (12-18)

PILLAR 3

Training and Empowerment

WP	Action/Activity	Channel	Timeline	Target Audience
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 • 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

PILLAR 4

Innovation and Clinical Trials Support

WP	Action/Activity	Channel	Timeline	Target Audience
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 aclinical 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 N°825575

EUROPEAN JOINT PROGRAMME RARE DISEASES

EUROPEAN JOINT PROGRAMME ON RARE DISEASES (EJP RD)

Policy Board and Governing Board meeting

EUROPEAN JOINT PROGRAMME RARE DISEASES

PB-GB meeting
04/07/2023, Bruxelles - Belgium

Welcome from Coordination

EUROPEAN JOINT PROGRAMME RARE DISEASES

EJP RD sustainability

EUROPEAN JOINT PROGRAMME
RARE DISEASES

Copenhagen Economics

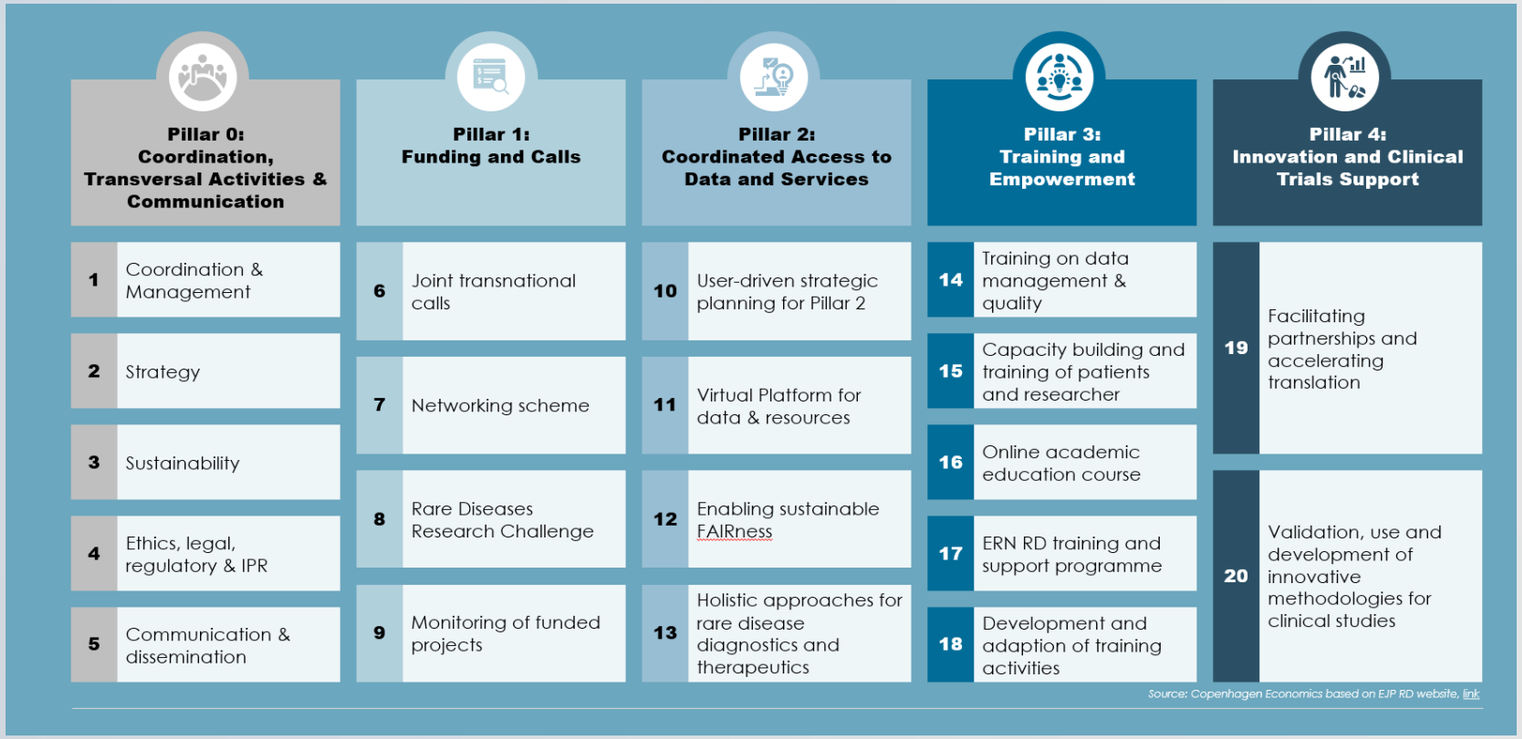
STRATEGY AND BUSINESS PLAN

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

EUROPEAN JOINT PROGRAMME RARE DISEASES



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

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

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

Topics: Innovative statistical methodologies for small population clinical trials. Statistical and operational challenges with master protocols. Evidence types, best practices on evidence collection and presentation.
Target audience: Organizations conducting clinical trials, EMA, the EU and national HTA bodies.
Experts teaching the course: Research community

Key features:

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

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

Source: Illustration by Copenhagen Economics

The RD Expertise Hub

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.

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.

FAIRification stewardship

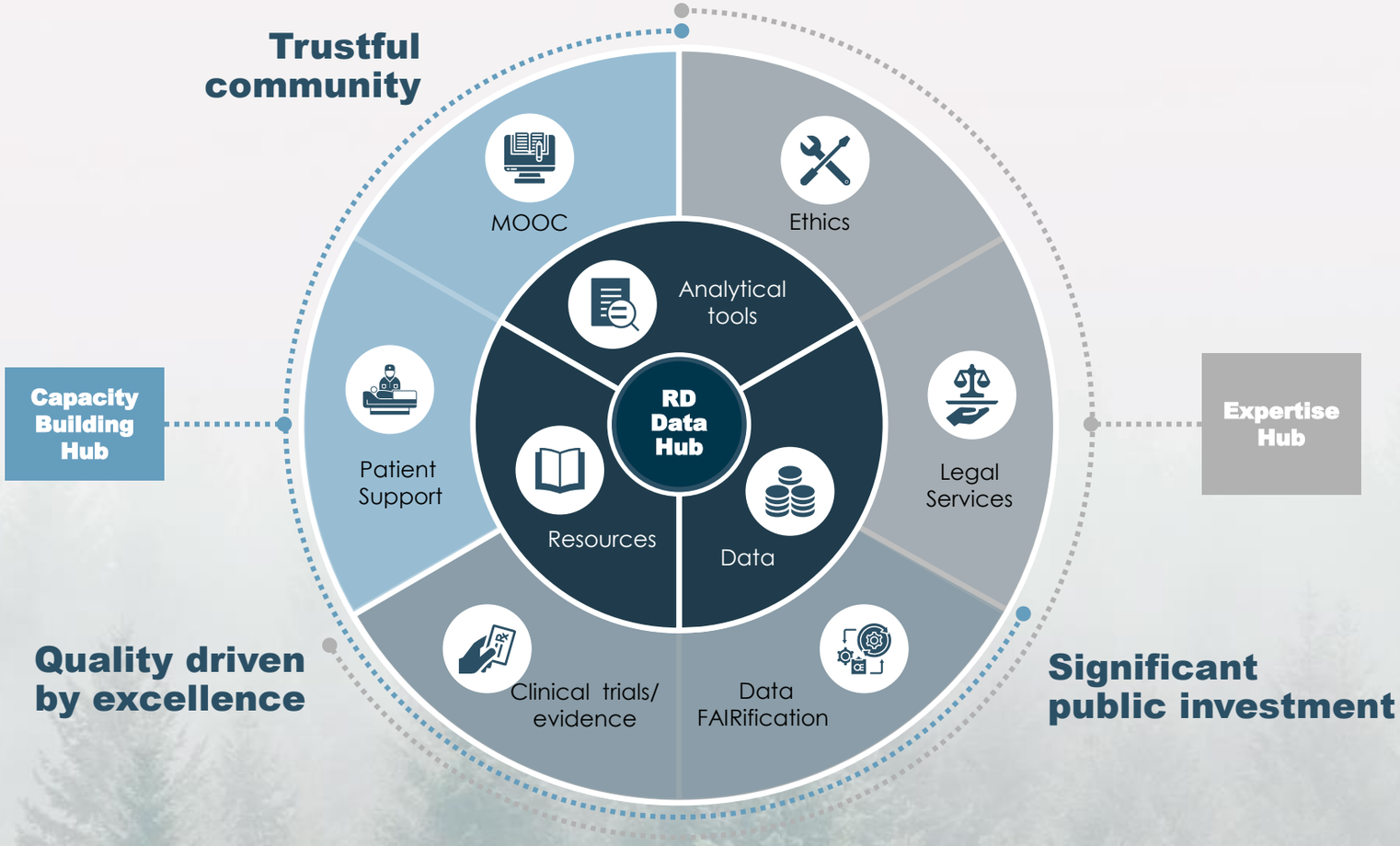
As part of FAIRification, the RD expertise hub supports the process of implementing and overseeing the adoption of FAIR principles in data processing.

Source: Illustration by Copenhagen Economics

Source: Illustration by Copenhagen Economics

6

Why is the RD Hub different from other existing solutions?



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

	Private companies, Top 10
1	Celgene Group
2	PPD Inc.
3	Parexel International Corp
4	Labcorp Drug Development
5	Bohringer Ingelheim International GmbH
6	Genentech USA Inc
7	PRA Health Sciences Inc
8	Alexion Pharmaceuticals Inc
9	Pharmasyntez
10	Novotech Australia Pty Ltd

	Publicly listed companies, Top 10
1	F. Hoffmann-La Roche Ltd
2	Merck & Co Inc
3	AstraZeneca Plc
4	Bristol-Myers Squibb Co
5	Johnson & Johnson
6	Novartis AG
7	Pfizer Inc
8	Sanofi
9	AbbVie Inc
10	IQVIA Holdings Inc

	Government, Top 10
1	Ministry of Health, France
2	Medical Research Council, UK
3	German Research Foundation, Germany
4	Federal Ministry of Education and Research, Germany
5	European Commission
6	Ministry of Health, Italy
7	The Swedish Research Council, Sweden
8	Region Skåne, Sweden
9	Directorate General of Health Care Provision, France
10	Vastra Gotalandsregionen, Sweden

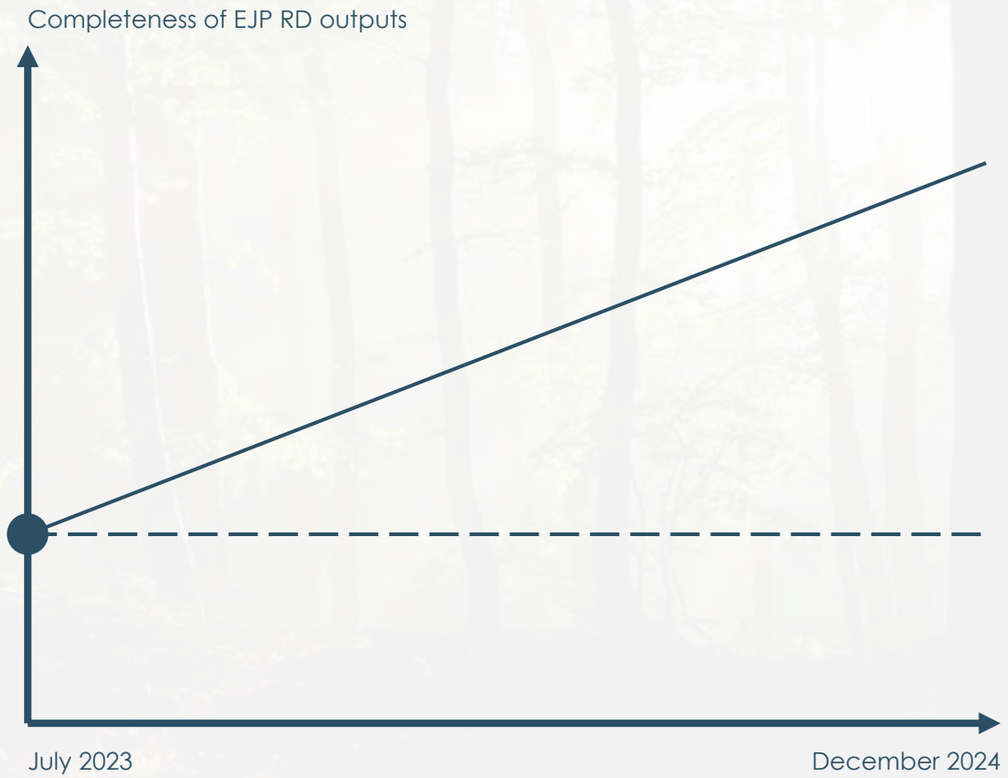
	Institutions, Top 10
1	Sarah Cannon Research Institute LLC, USA
2	Assistance Publique – Hopitaux de Paria, France
3	Cancer Research UK, UK
4	Erasmus MC, Netherlands
5	University College London, UK
6	University of Birmingham, UK
7	Rigshospitalet, Denmark
8	Aarhus University Hospital, Denmark
9	Oslo University Hospital, Norway
10	KWF Kankerbestrijding, Netherlands

Source: Copenhagen Economics based on GlobalData.

1) Copenhagen Economics based on GlobalData

Closing the gap between status quo and the vision

Status quo and the vision



The gap

The solutions

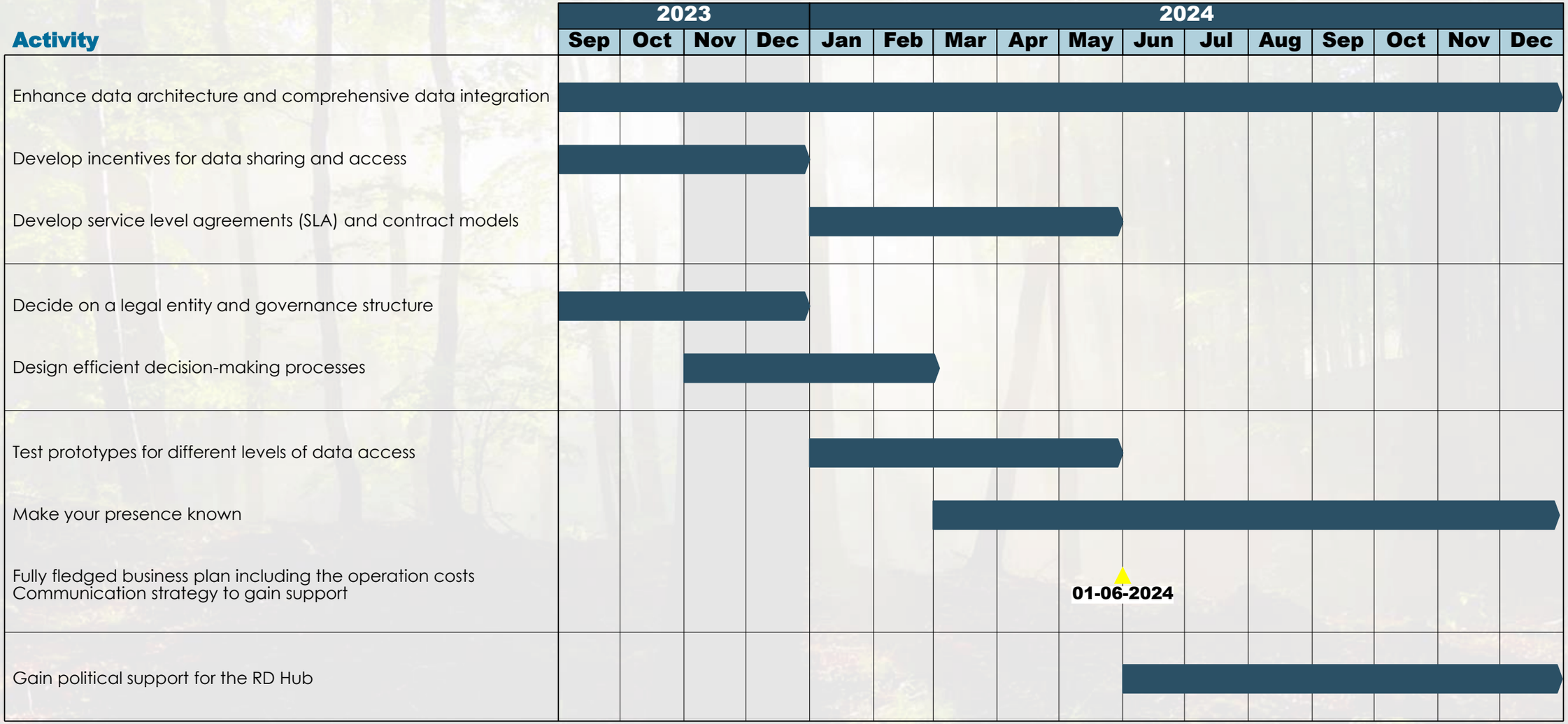
Foster a customer-oriented mindset

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



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

EUROPEAN JOINT PROGRAMME RARE DISEASES

COORDINATION, TRANSVERSAL ACTIVITIES & COMMUNICATION

EUROPEAN JOINT PROGRAMME
RARE DISEASES

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.

EJP RD

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)

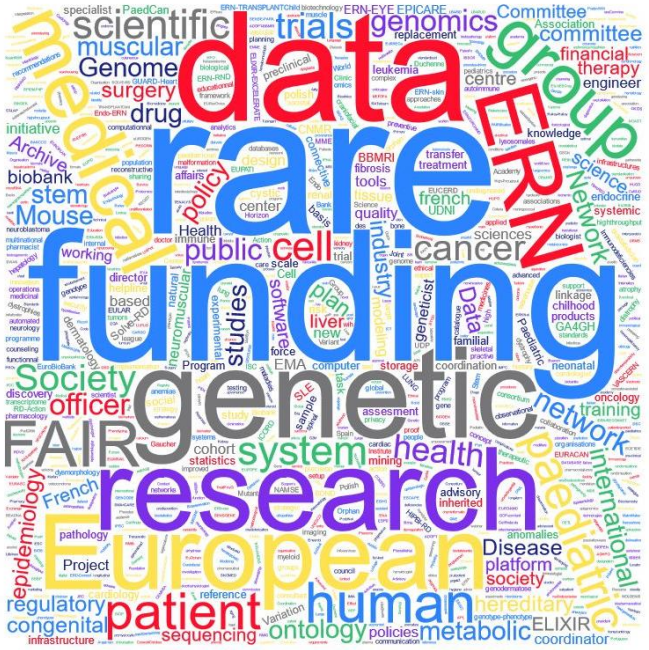
Pillar 0 – Achievements at M55

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

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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

IRDiRC
INTERNATIONAL RARE DISEASES RESEARCH CONSORTIUM

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

AWP Y6: 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

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

AWP Y6: 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.

AWP Y6: 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
Impacts and Plans for Year 6

EUROPEAN JOINT PROGRAMME
RARE DISEASES

Pillar 1 – Expected impacts

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

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

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

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.

EJP RD

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.

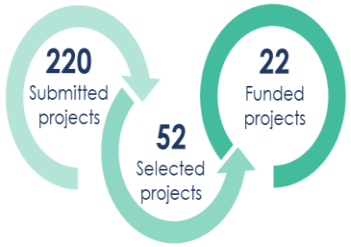
Pillar 1 – Achievements at M55

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
23 countries



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
22 countries

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
16 countries

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
24 countries

17 million € spent in total

EJP RD

EVENT DATE

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

Challenge	**Project title (Acronym)**	**Lead applicant**	N° of partners involved	N° of countries	**Industry sponsors**	**Total funding**
#1Develop--ment of a non-invasive tool for measuring rare disease patient mobility in daily living	**Digital tools 4 Rare Diseases (DT4RD)**	**SME; UK**	5 (2 SME + 2 Academia + 1 PAO)	3 (Netherlands; France; UK)	**Chiesi and CSL Behring**	**575 000 €**
#2 Delivery system for intranasal administration of biological drugs to neonates	**Intranasal device for neonates (INDENEO)**	**SME ; France**	3 (1 SME + 2 Academia)	2 (France; Belgium)	**Chiesi**	**487 500 €**
#4 Pre-clinical assay to detect instability of microsatellite repeat expansions	**Development and validation of a novel pre-clinical assay to detect triplet repeat expansions (TRXAssay)**	**Academia; Ireland**	3 (Academia)	2 (Ireland; UK)	**LoQus23**	**487 500 €**

EJP RD

EVENT DATE

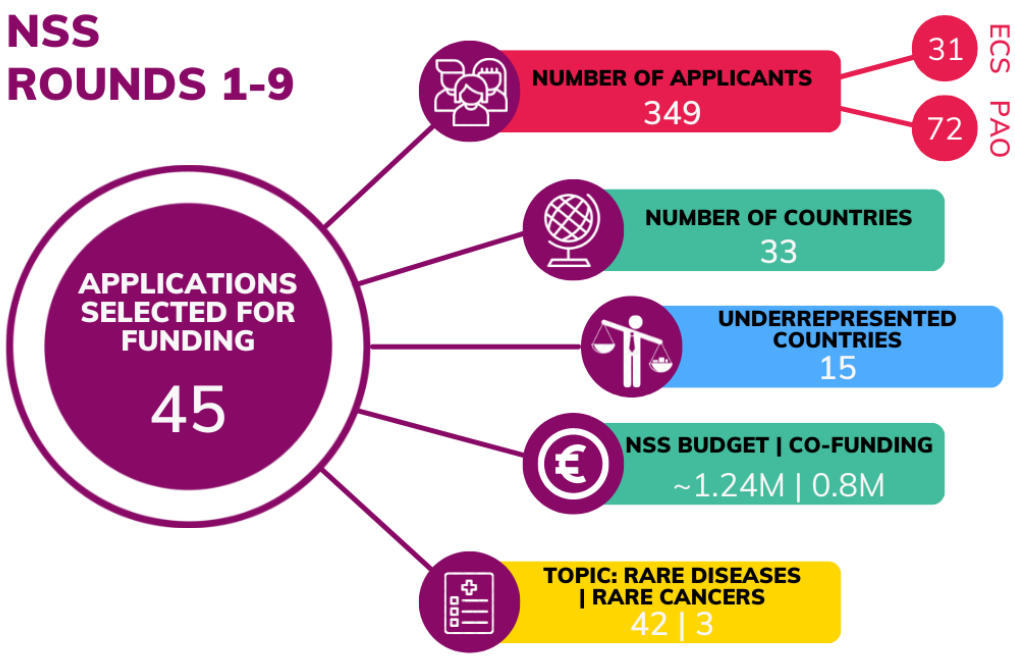
Pillar 1 – Achievements at M55

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



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.

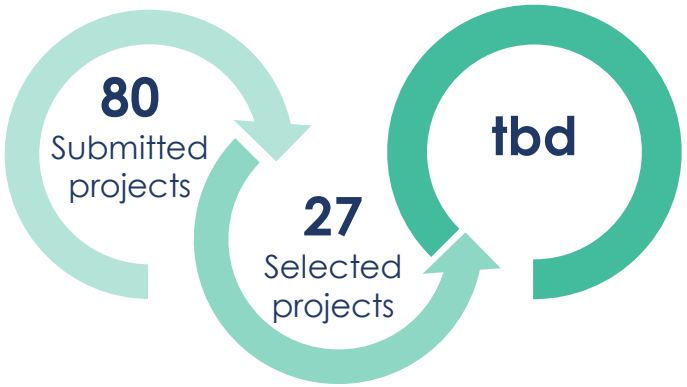
- All funded research activities have contributed to reinforcing EU leadership role and fostering IRDIRC objectives
- IRDIRC experts and resources were instrumental in shaping research topics for joint transnational calls

EJP RD

WP6 Joint Transnational Calls for collaborative research projects

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

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 lessions learned for all JTC calls to further adapt call documents and procedures for future calls.

EJP RD

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

Public-private projects

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

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 OF DATA, TOOLS & RESOURCES

EUROPEAN JOINT PROGRAMME
RARE DISEASES

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 computationnally 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

EJP RD

Pillar 2 – Expected impacts

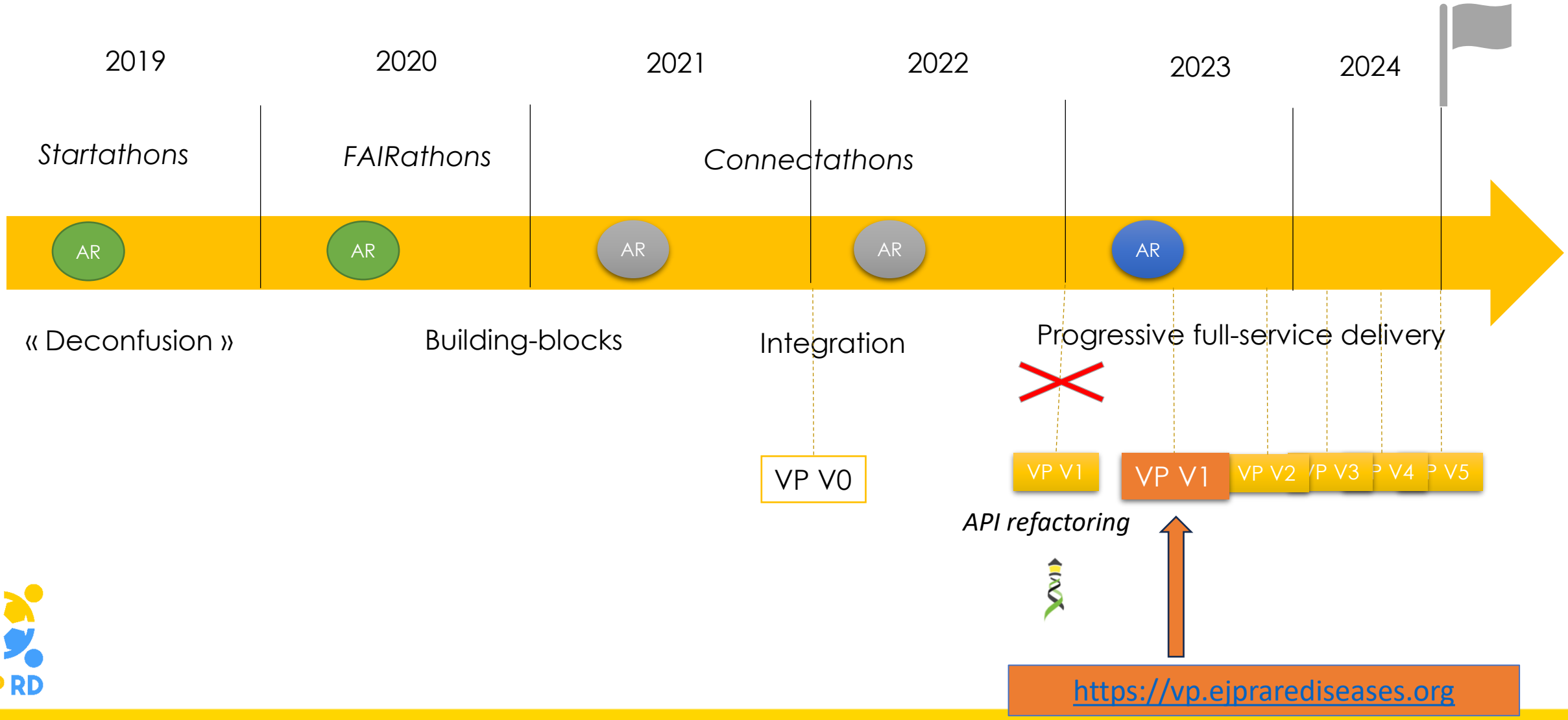
Specific impact 5: **Reinforce the EU's role as a global leader for rare diseases**

- 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

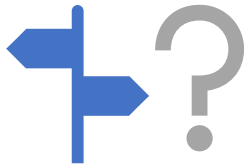


P2 achievements: From a heterogeneous, non-interoperable, scattered resources landscape …

Registries/ biobanks catalogues

Cell lines Animal models

Knowledge bases



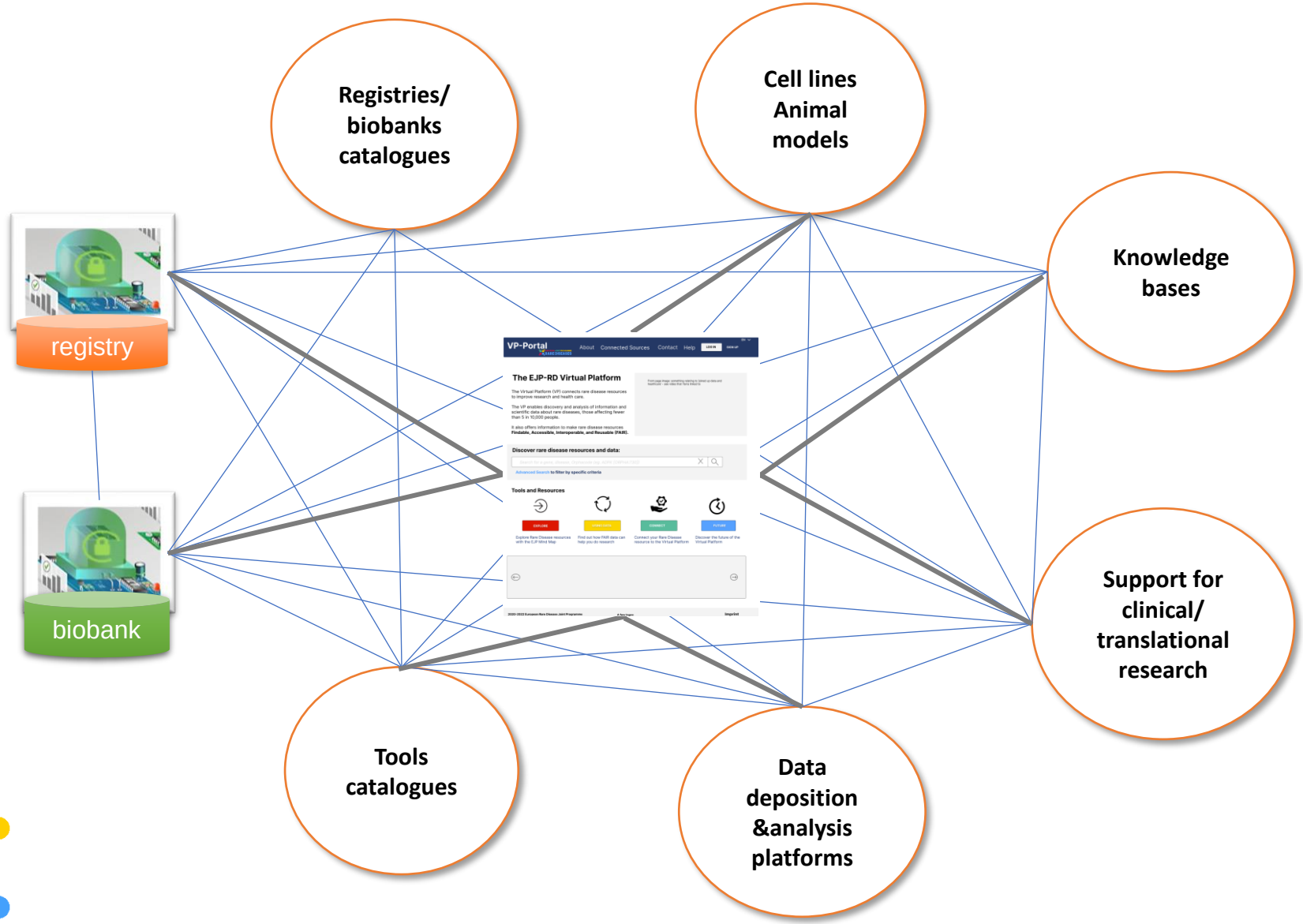
Support for clinical/ translational research

Tools catalogues

Data deposition & analysis platforms

EJP RD

To the Virtual Platform: a network of federated resources



Decreasing fragmentation

Increasing interoperability

Increasing RD-readiness

through

Harmonisation

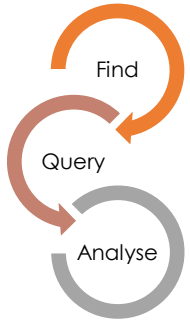
Standardisation

in a flexible way:

Common methodologies, Multipe technical solutions

V1: https://vp.ejprarediseases.org

Onboarding at diverse levels of connection to the Virtual Platform



LIFE SCIENCE RI

Y6: onboard 100% of EJP RD partner resources

Succeed L3 for ready resources
LifeScience AAI fully implemented

POC Federated analysis on FAIR data

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

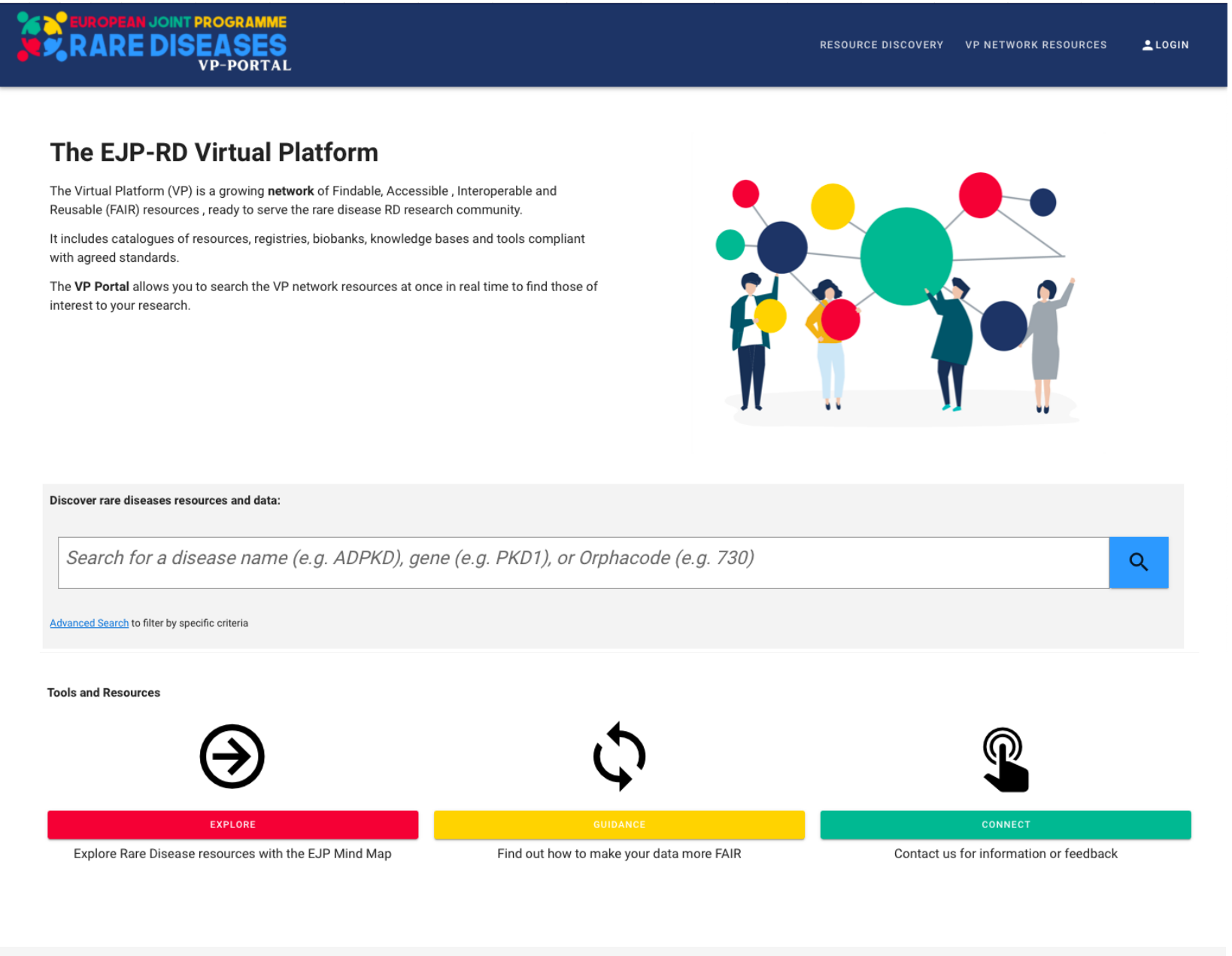
- Federated Analysis
  - Answer advanced research questions

What is it?

What it contains?

Answer my complex question

EJP RD



V1

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

https://vp.ejpraretdiseases.org

Behind the scenes

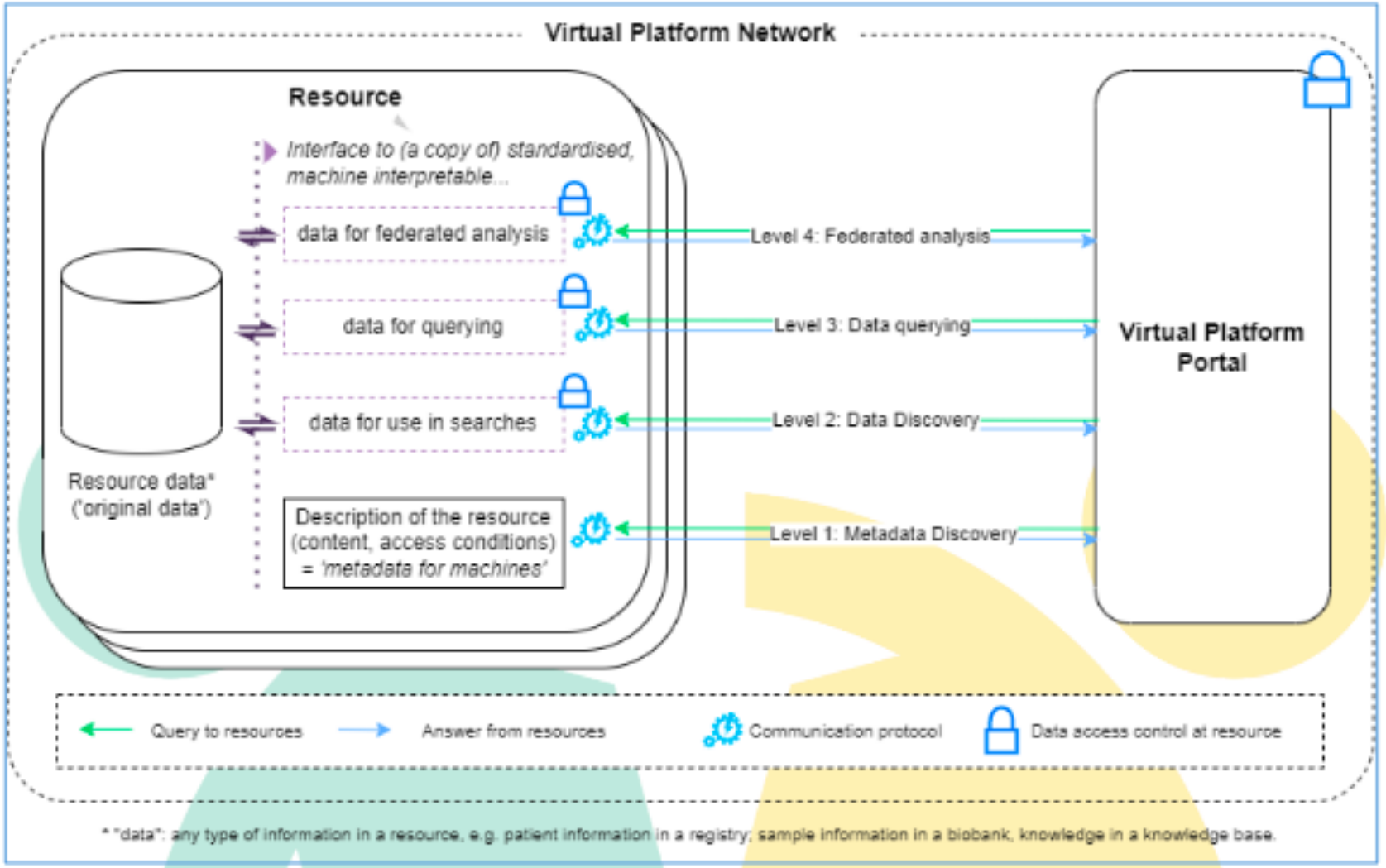
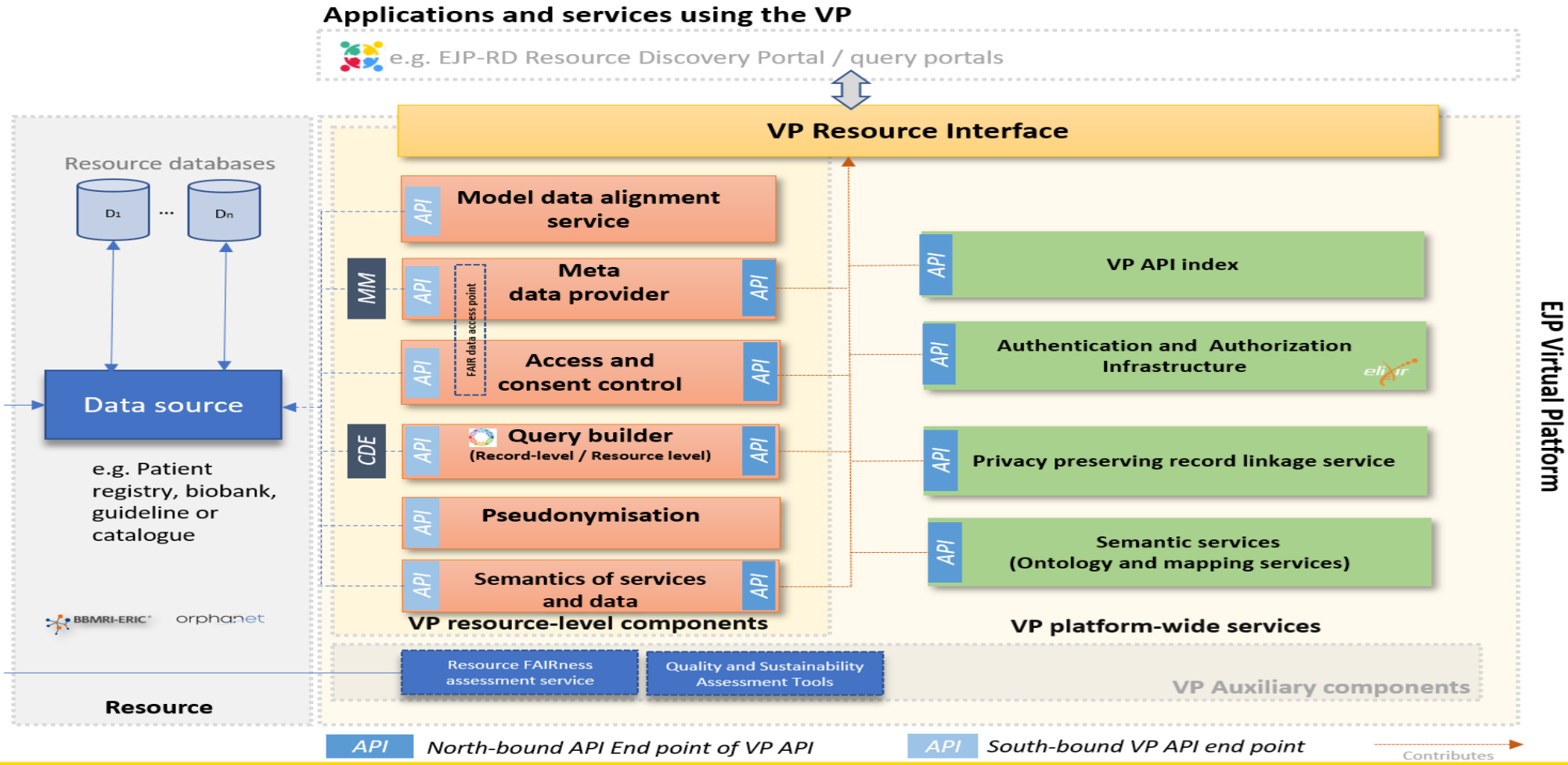


Figure 4: Illustration of the levels by which resources can contribute to and benefit from the Virtual Platform. Resources that apply the recommended extendible standards 'for machines' at source automatically increase the functionality of the Virtual Platform for the community

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

EUROPEAN JOINT PROGRAMME RARE DISEASES

AD49
Virtual Platform Specification (VIPS) v2.0

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

AD49

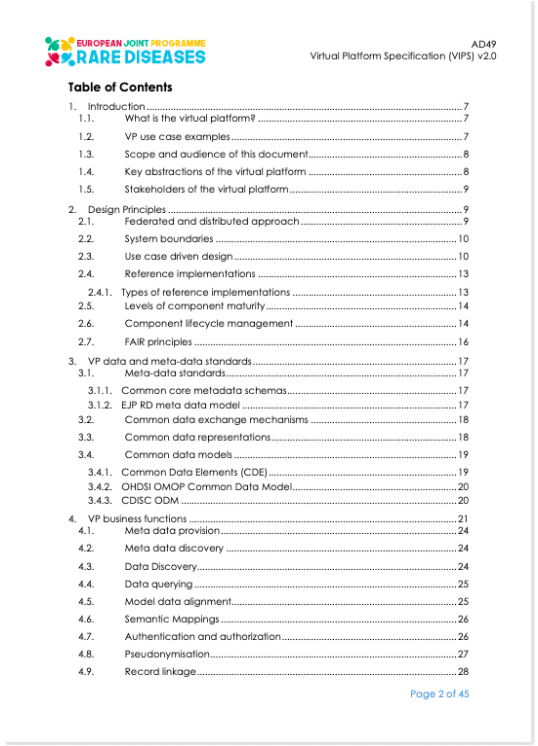
Virtual Platform Specification (VIPS) V2.0

Organisation name of lead beneficiary for this deliverable:
Partner 03 - AIT

Due date of deliverable: month 45

Dissemination level:
Public

Page 1 of 45

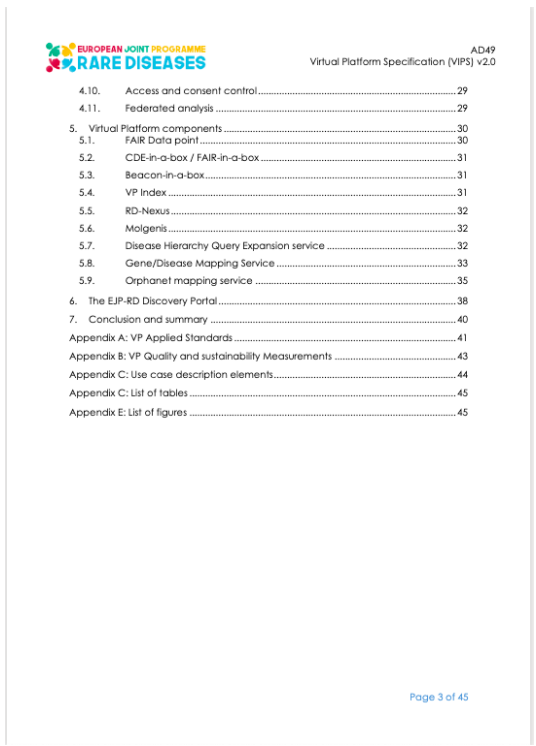


EUROPEAN JOINT PROGRAMME RARE DISEASES

AD49
Virtual Platform Specification (VIPS) v2.0

Table of Contents

Page 2 of 45

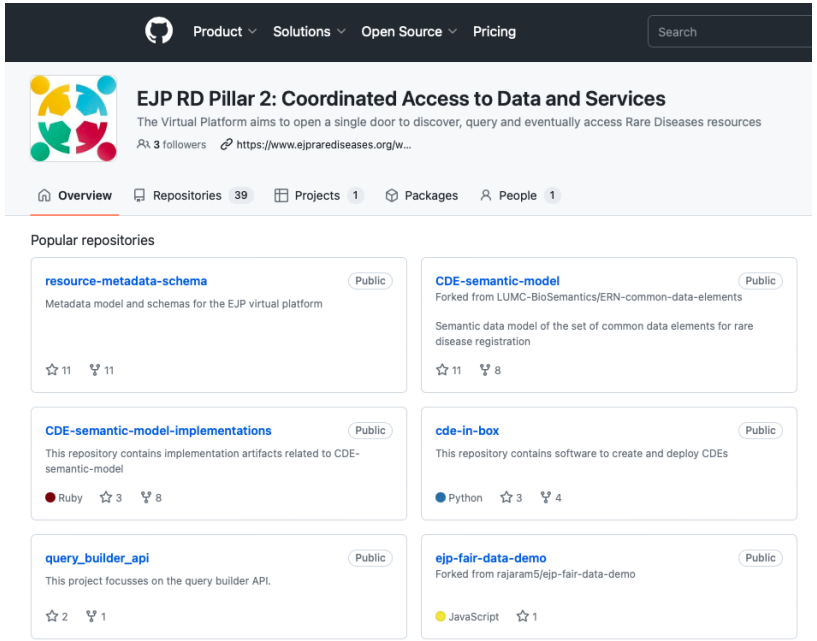


EUROPEAN JOINT PROGRAMME RARE DISEASES

AD49
Virtual Platform Specification (VIPS) v2.0

Page 3 of 45

Y6: Final documentation



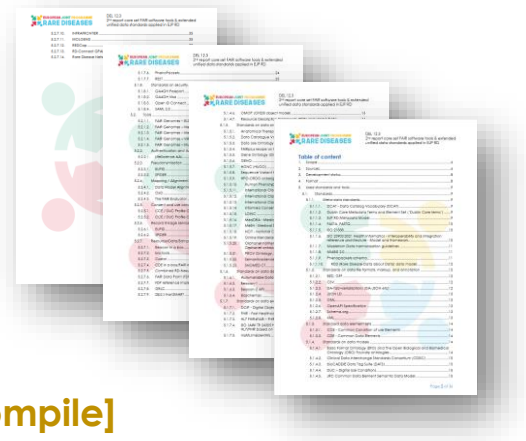
https://www.ejprarediseases.org/wp-content/uploads/2023/05/EJPRD_P2_AD49_PU_Virtual-Platform-Specification-V2.0.pdf

https://github.com/ejp-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

Y6: Final version



[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

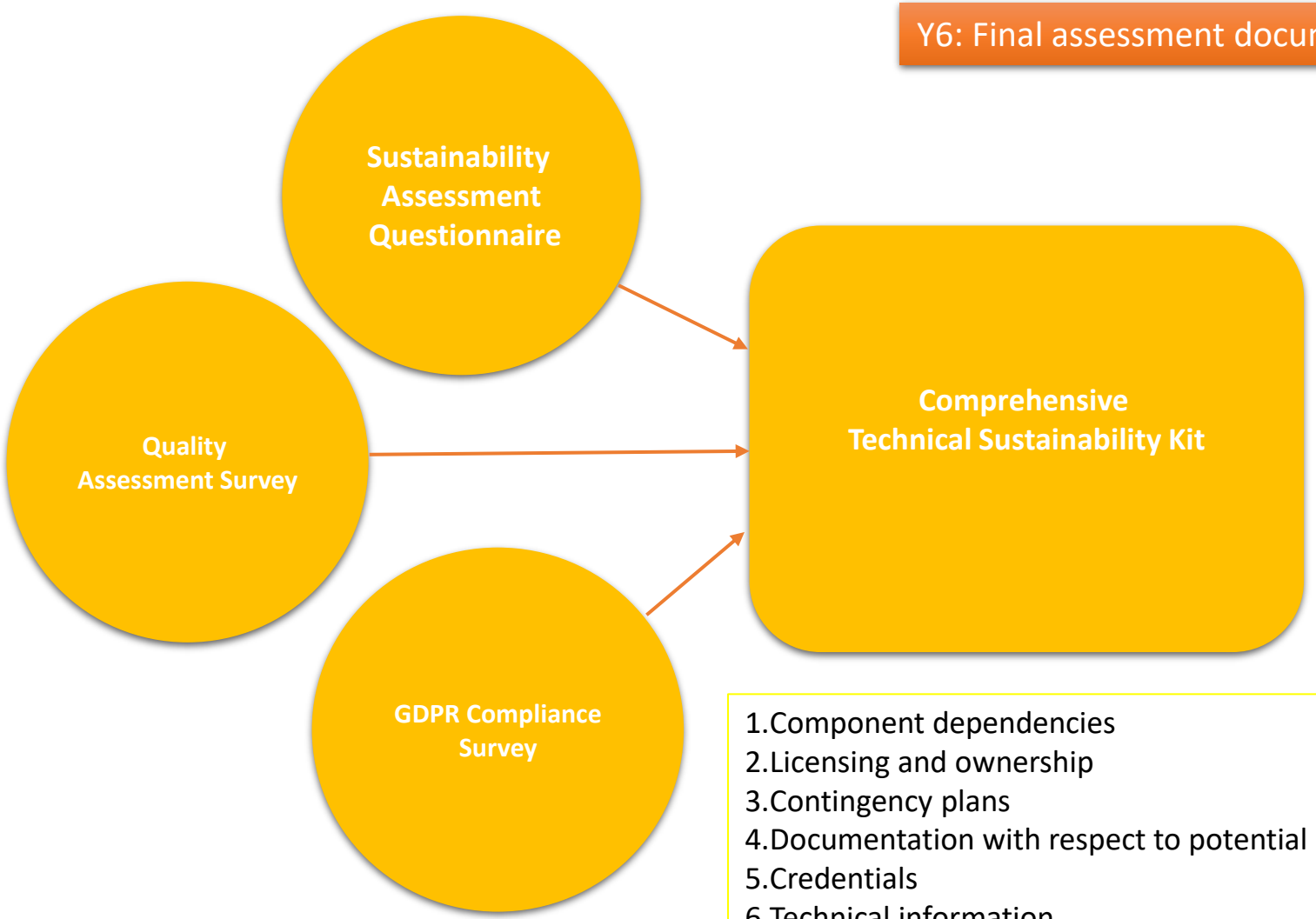
https://www.ejprarediseases.org/wp-content/uploads/2023/02/EJPRD_P2_D12.03_PU_2nd-Report-Core-Set-FAIR-Software-Tools-Extended-Set-Unified-FAIR-Data-Standards-Applied-in-EJP-RD_VF.pdf

VP components are assessed for their technical sustainability

Y6: Final assessment documented

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

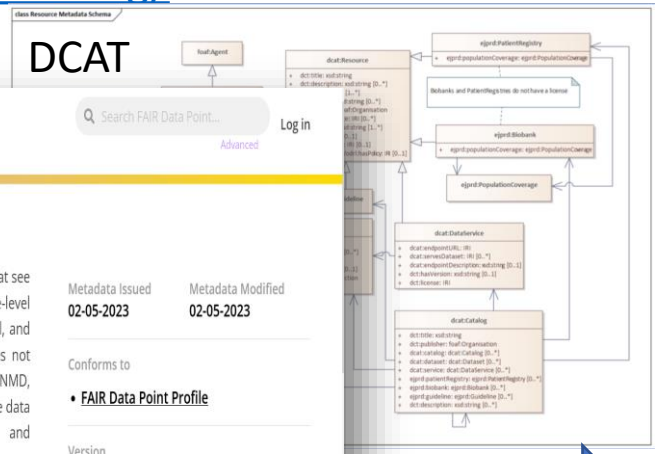
Evaluation meetings conducted after completion of the kit

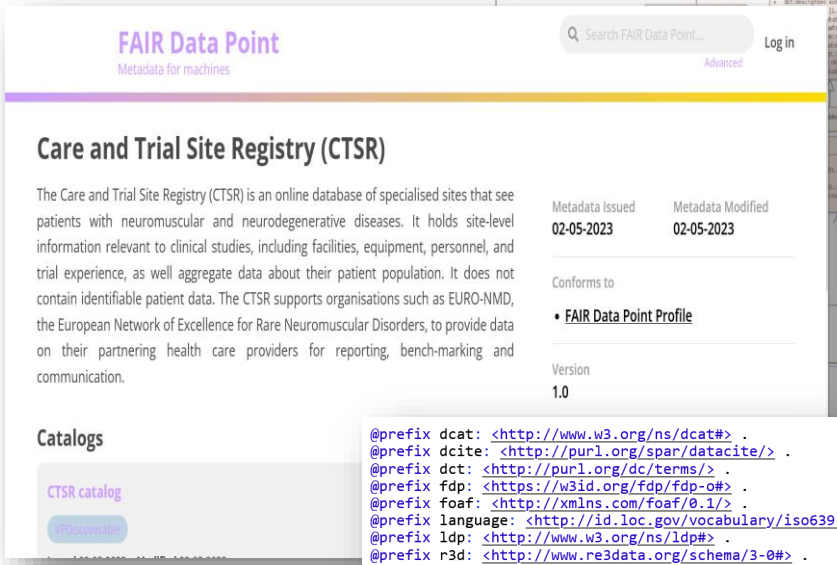


1. Component dependencies
2. Licensing and ownership
3. Contingency plans
4. Documentation with respect to potential users
5. Credentials
6. Technical information
7. Interoperability

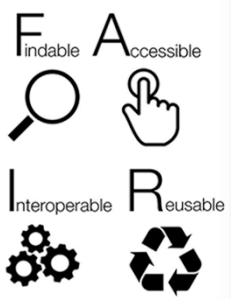
VP is adaptive

https://specs.fairdatapoint.org/ Bonino da Silva Santos *et al.* FAIR Data Point: A FAIR-Oriented Approach for Metadata Publication. *Data Intelligence* 2023; 5 (1): 163–183. https://doi.org/10.1162/dint_a_00160





…for humans
generated from →

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@prefix dcite: <http://purl.org/spar/datacite/> .
@prefix dct: <http://purl.org/dc/terms/> .
@prefix fdp: <https://w3id.org/fdp/fdp-o#> .
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@prefix language: <http://id.loc.gov/vocabulary/iso639-1/> .
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@prefix xsd: <http://www.w3.org/2001/XMLSchema#> .

<https://w3id.org/ctsr-fdp> dct:accessRights <https://w3id.org/ctsr
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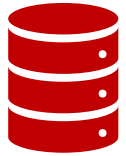
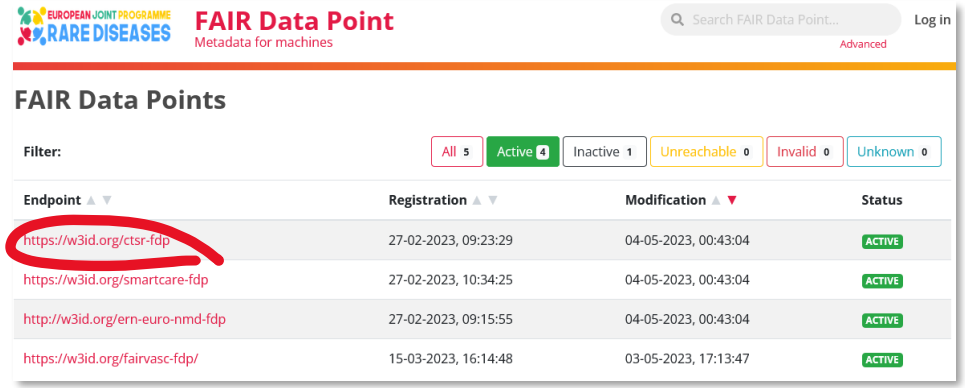
FAIR for machines
(extendible, fully ontology-qualified description)

VP FDP index





Index user interface
https://index.vp.ejprarediseases.org/
(also a FAIR Data Point)

```
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@prefix dcterms: <http://purl.org/dc/terms/> .
@prefix foaf: <http://xmlns.com/foaf/0.1/> .
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@prefix xsd: <http://www.w3.org/2001/XMLSchema#> .

<https://index.vp.ejprarediseases.org> dcterms:accessRights <https://index.vp.ejprarediseases.org#accessRights> ;
dcterms:conformsTo <https://index.vp.ejprarediseases.org/profile/77aaad6a-0136-4c6e-88b9-07ffccd0ee4c> ;
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dcterms:publisher <https://index.vp.ejprarediseases.org#publisher> ;
dcterms:title "My FAIR Data Point" ;
<http://www.re3data.org/schema/3-0#repositoryIdentifier> <https://index.vp.ejprarediseases.org#identifier> ;
<http://www.w3.org/1999/02/22-rdf-syntax-ns#type> dcat:DataService ,
dcat:Resource ,
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<https://w3id.org/fdp/fdp-o#MetadataService> ;
<http://www.w3.org/2000/01/rdf-schema#label> "My FAIR Data Point" ;
```

Index FAIR Data Point *for machines*
https://index.vp.ejprarediseases.org/?format=ttl

EJP RD

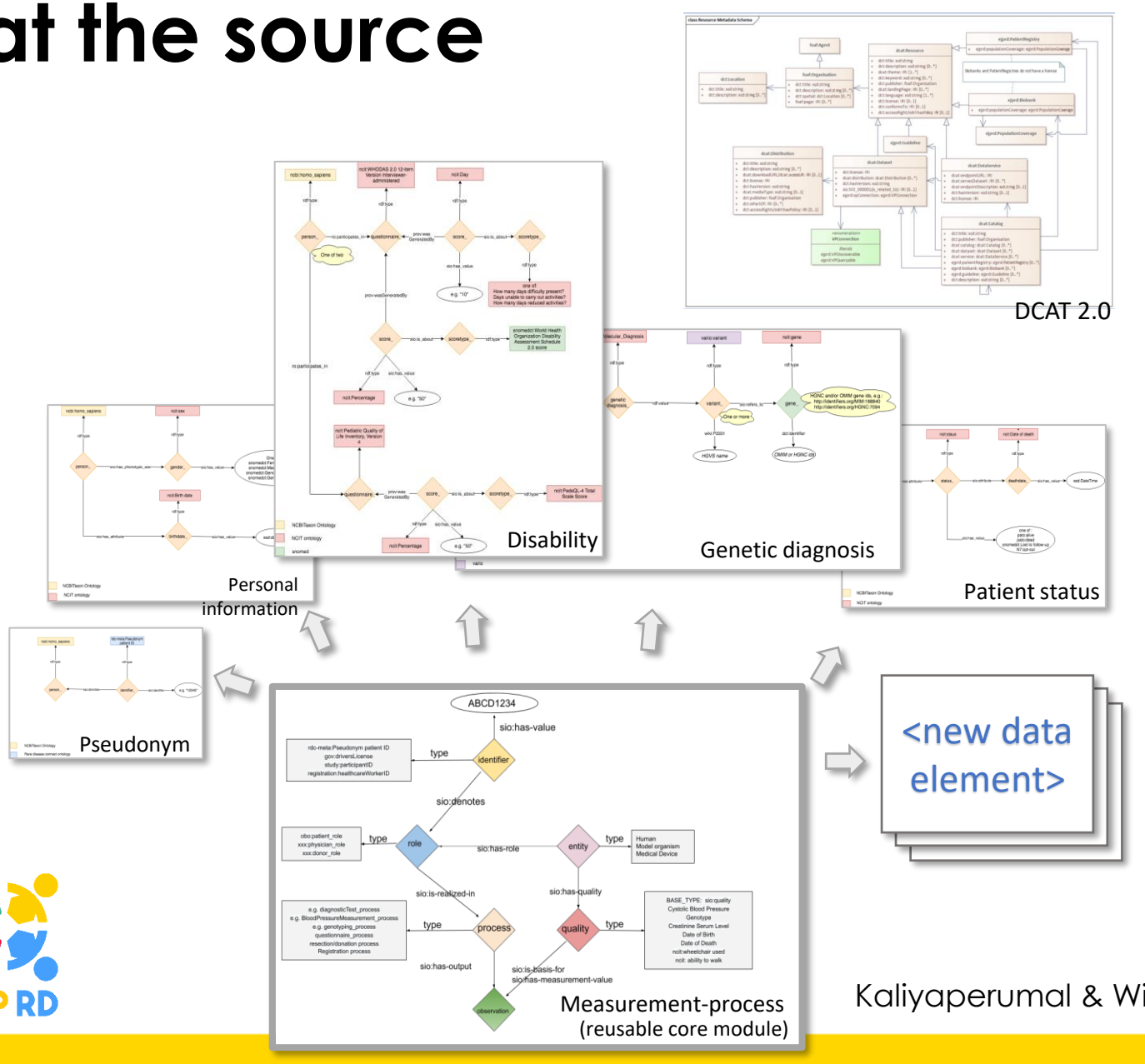
Virtual Platform Network Virtual Platform Portal

VP is based on FAIR data models at the source

Y6: Final adaptations of models and Mapping / transformation services

Rare disease resource (e.g. a registry)
Compatible with EJP RD VP specs & FAIR

- 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

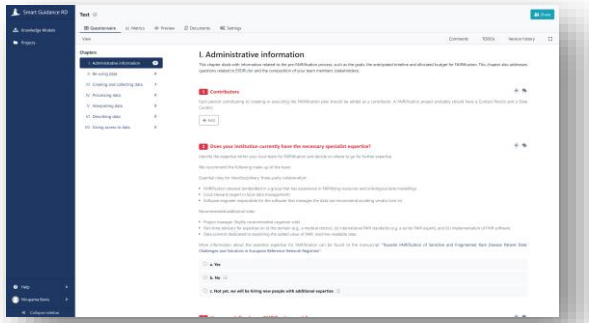
Y6: Final material Dissemination & training

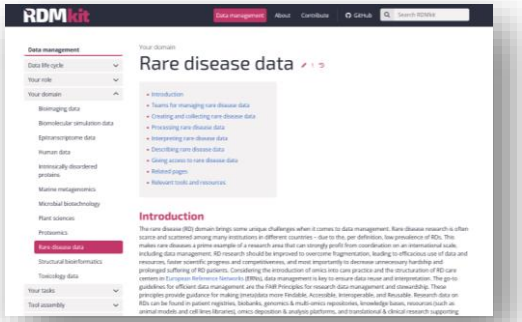
VP onboarding

- *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

https://www.ejprarediseases.org/wp-content/uploads/2023/02/EJPRD_P2_D11.09_PU_Third-update-Virtual-platform-of-RD-resources-annotated-with-EJP-ontological-model_VF.pdf

FAIRification



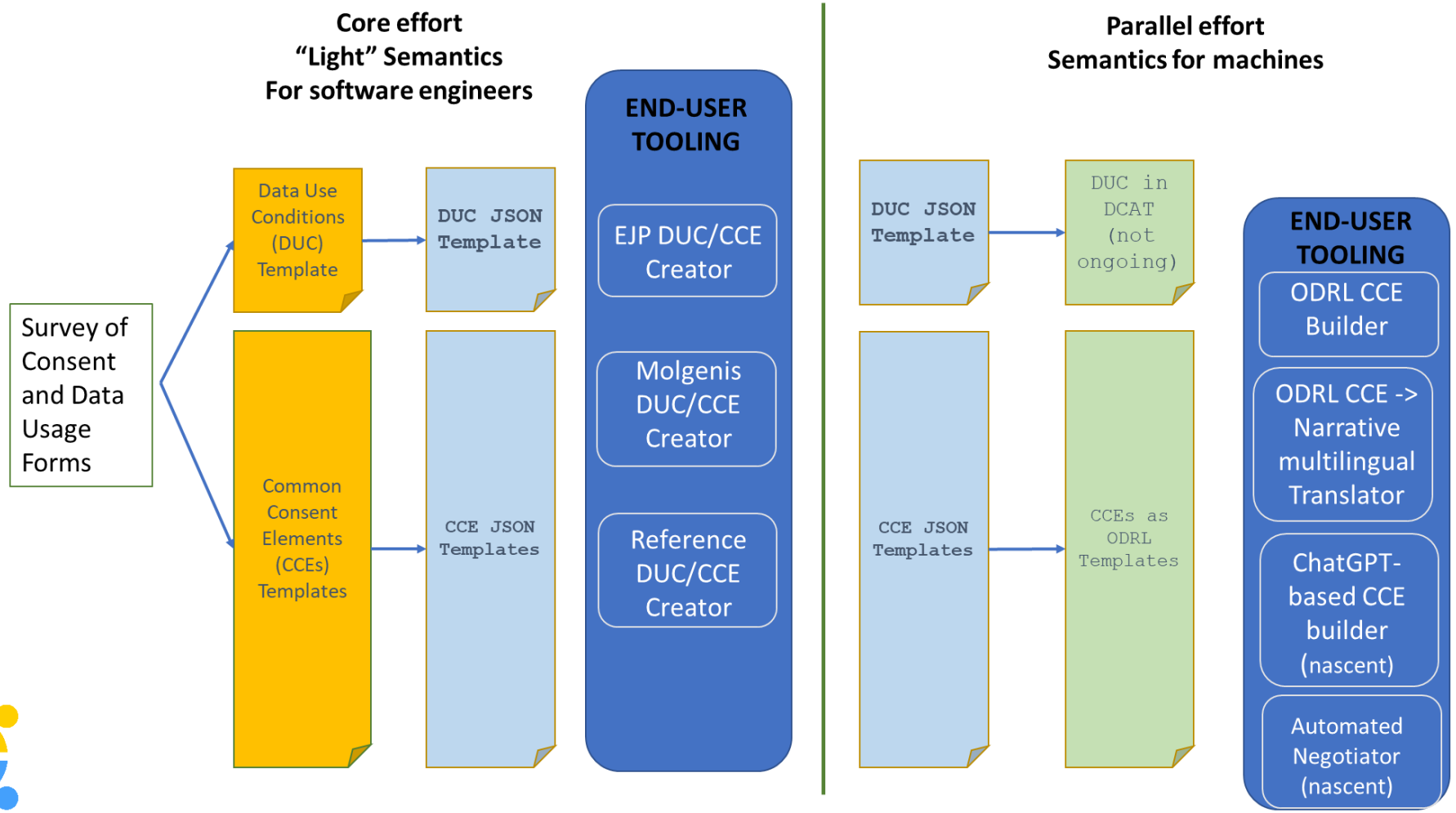


"A Model for Guiding Data Stewards of European Rare Disease Patient Registries"
van Damme *et al.*, CODATA Data Science Journal, *under review*
https://www.ejprarediseases.org/the-smart-guidance-tool-for-the-fairification-of-rare-disease-registries/

https://www.ejprarediseases.org/fairification/

Tackling the reusability challenge: ongoing work

Y6: finalize implementations initiated second half Y5



Tackling the privacy preserving record-linkage challenge: ongoing work

Y6: finalize PPRL interoperability POC

Finalize PPRL implementation use-case (EUPID)

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

Pillar 2 partners have improved data deposition and analysis resources for RD

Y6: last final improvements and documentation

- **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

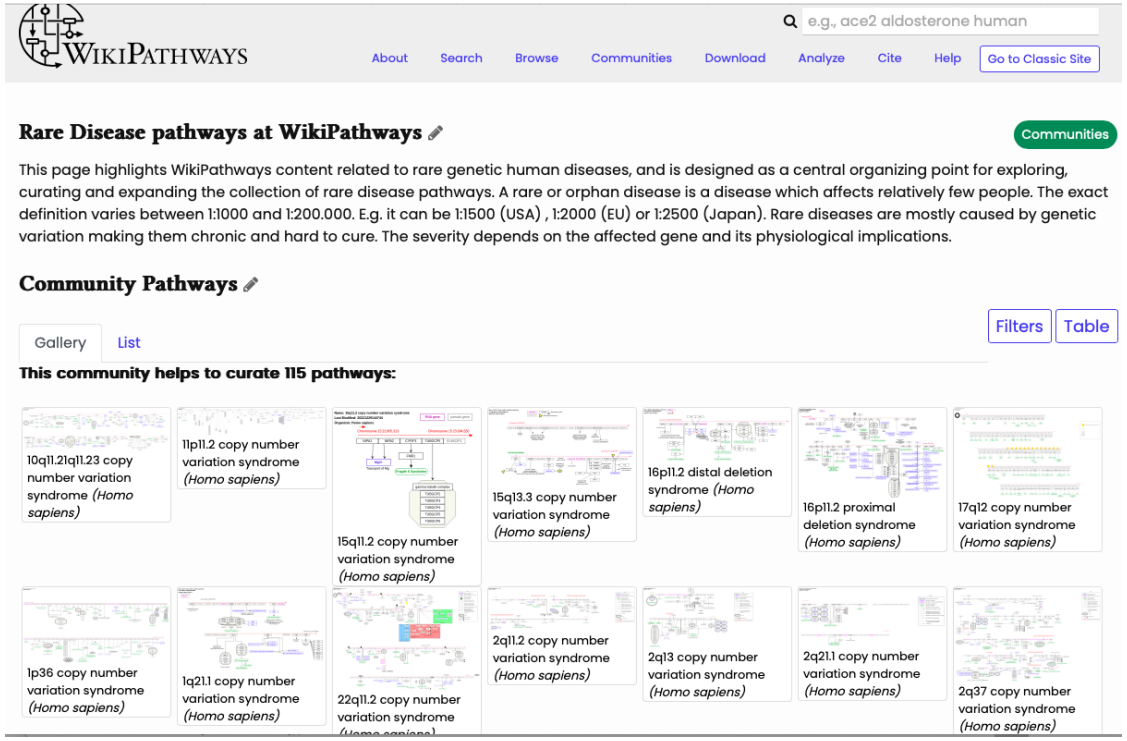
RD-Connect Registry and Biobank Finder: all entries migrated to Molgenis. Negotiator PoC.

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



115 rare disease pathways available and reusable

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

FAIR data cube

- 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)

https://www.ejpraregiseases.org/wp-content/uploads/2022/01/EJPRD_P2_D13.01_PU_Collection-of-curated-pathways-sub-Portal-WikiPathways_VF.pdf

Lessons learned: VP sustainability aspects

Aspects	Needs
Technical sustainability Of the components Of the ecosystem	Evolutive technical maintenance
Support evolutivity Quantitative growing as a network Qualitative diversify the network	Strategic, tactical and operational capacity
Maintain and create value Develop services Become a major actor: attractivity	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

EUROPEAN JOINT PROGRAMME RARE DISEASES

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;

EJP RD

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)*

2024

- 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

EJP RD

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)

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

Impact

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)

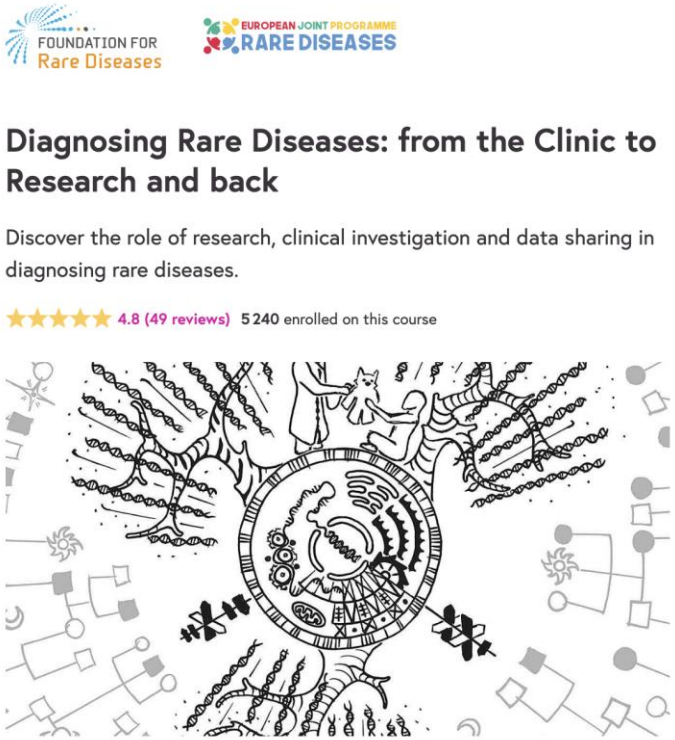
Empowering young patients' advocates by developing an assent form with and for young patients to process and share data for RD in ERNs

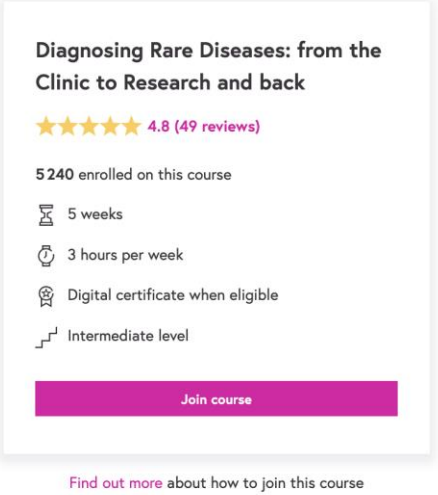
EJP RD

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.

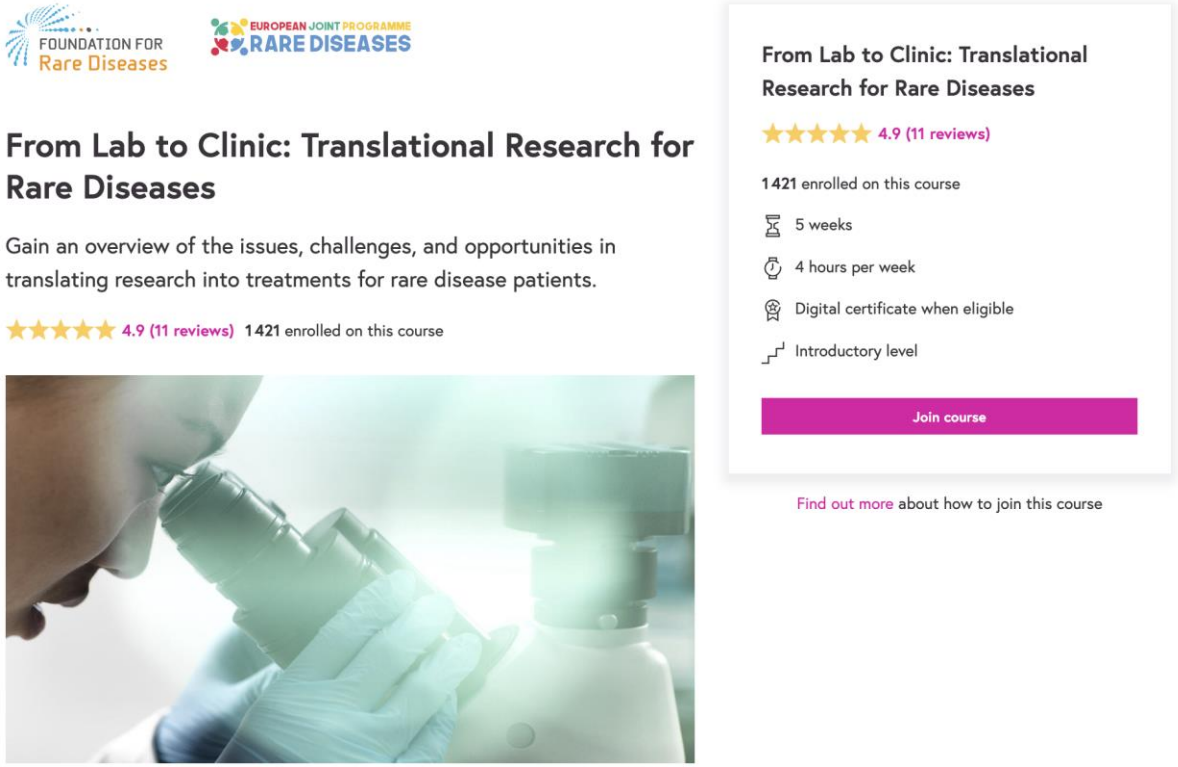




Pillar 3 – Achievements

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**
  - 2024 MOOC#2 - ***Innovative therapies and personalized medicine approaches for RD***
    - Developed by **FFRD, CVBF, ERN Transplant Child**
    - Launch foreseen in Autumn 2023
  - 2024 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)

EJP RD

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.

EJP RD

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".

EJP RD

**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 mutistakeholder 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%.

EJP RD

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.

EJP RD

EJPRD education and capacity building programme: overview

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	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

EJP RD

EJPRD education and capacity building programme: overview

Activity	Targeted stakeholders	Keywords
P3 WP15.1 - ExPRESS Expert Patients and Researchers EURORDIS Summer School Pre-training and e-learning courses; F2F courses	Patient advocates, researchers	Clinical trials, Orphan drugs, regulatory and ethicolegal issues, patient engagement, pharmacovigilance, European Medicines Agency, Health Technology Assessment
P3 WP15.2 – Training for patient advocates on scientific innovation and translational research - EURORDIS Winter School Pre-training and e-learning courses; F2F courses	Patient advocates	Translational research, genetics, bioinformatics, ERNs, RD diagnostics, undiagnosed diseases, gene/advanced therapies, genome editing, patient engagement
P3 WP15.3 – Training for patient advocates on leadership and communication skills Pre-training and e-learning courses; F2F courses	Patient advocates	Leadership, self-awareness, conflict resolution strategies, authority, negotiation, networking, communication
P3 WP 15.4: Educational materials and activities for paediatric patients e-learning; F2F courses	Pediatric patient advocates	Rare diseases, patient engagement, clinical research, ethicolegal issues, informed consent/assent form, patient wellbeing, Rare Disease Day

EJP RD

EJPRD education and capacity building programme: overview

Activity	Targeted stakeholders	Keywords
P3 WP16 MOOC#1 - "Diagnosing Rare Diseases: from the Clinic to Research and back" On-line academic course.	Students, multistakeholder community (researchers, clinicians, patients and patients' representatives)	General concepts about RD and genetic diagnostics, genomic technologies, care pathways, patient-centeredness, undiagnosed diseases, genetic consultation, genetic research
P3 WP16 MOOC#2 - Innovative personalized therapies On-line academic course.	Students, multistakeholder community (researchers, clinicians, patients and patients' representatives)	Rare disease treatment, innovative therapies, gene & cell therapy, regenerative medicine, genome editing, personalized medicine
P3 WP16 MOOC#3 - Translational Research On-line academic course.	Students, multistakeholder community (researchers, clinicians, patients and patients' representatives)	Translational research, preclinical and clinical research, disease models, biomarkers, clinical trials, regulatory issues, ethicolegal aspects, postmarketing
P3 WP16 MOOC#4 Rare Disease Clinical Trials innovative methodologies On-line academic course.	Students, multistakeholder community (researchers, clinicians, patients and patients' representatives)	Small population clinical trials, clinical trial designs, statistical analysis
P3 WP17 Research training workshops	Clinicians, multistakeholder community	N/A
P3 WP17 Fellowships for research mobility secondments	Young clinicians, multistakeholder community	N/A

EJP RD

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.

EJP RD

EJPRD education and capacity building programme: evaluation

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.).

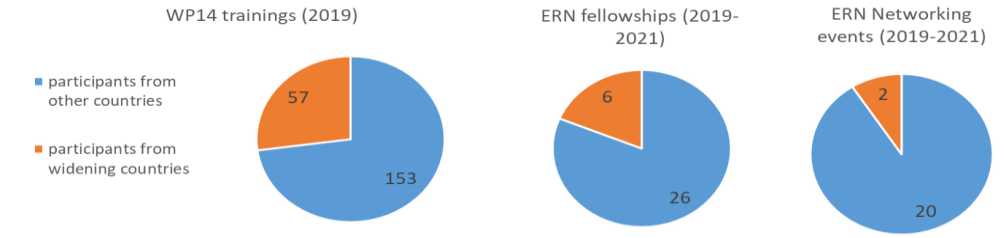
- EJPRD education and training programme covers the vast majority of these topics to various extent. Somewhat less covered topics include education and training on socioeconomical studies, social sciences and humanities, preclinical studies and data science.

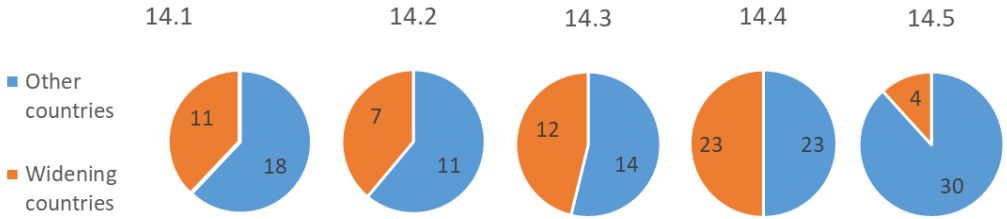
Impact of EJPRD: participation of widening countries

Empowerment and capacity building:

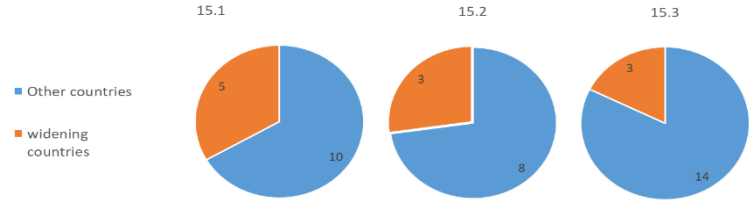
Participants from widening countries comprised:

- 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.

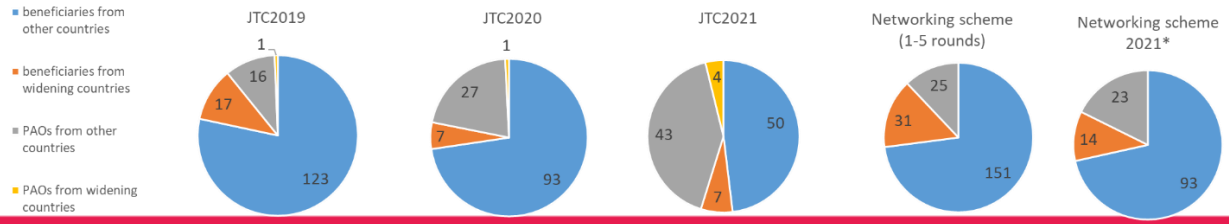


Rare disease research funding:

- **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-centredeness** 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).

EJP RD

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.

EJP RD

ACCELERATED TRANSLATION OF RESEARCH RESULTS & CLINICAL TRIALS

EUROPEAN JOINT PROGRAMME
RARE DISEASES

PILLAR 4

WP19

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.

WP20

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

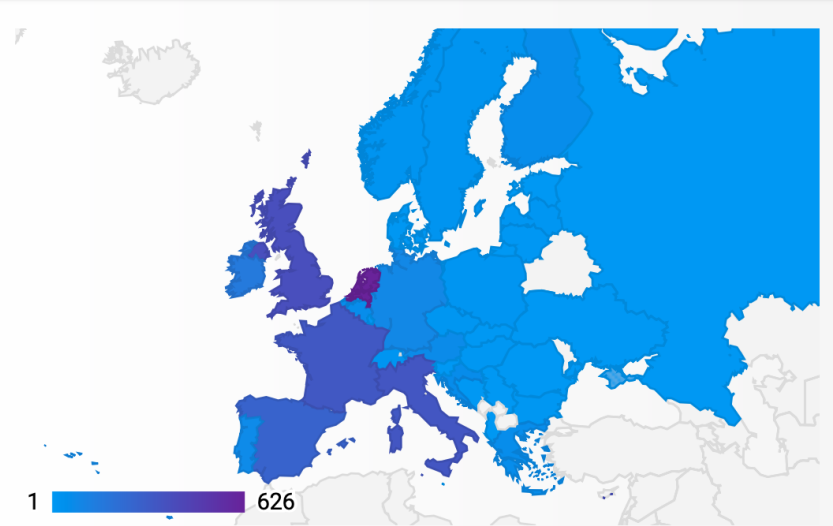
- WP19 has worked closely with Pillar 2 to ensure discoverability of IMT elements in the virtual platform and avoid duplications.

Impact

Google Analytics
June 2022 – June 2023

Sessions: ***3366***
Visitants: ***1956***

	Category	Sessies
1.	research and drug development	85
2.	regulatory science	67
3.	translational project management	66
4.	funding sources	65
5.	research and drug development -...	47
6.	regulatory science -> preclinical s...	30
7.	research and drug development -...	30
8.	regulatory science -> early acces ...	29
9.	intellectual property	28
10.	research and drug development -...	28



	Land	Sessies	Weergaven
1.	Netherlands	626	
2.	United Kingdom	383	
3.	Italy	344	
4.	United States	338	
5.	France	335	

IMT obtained the IRDiRC recognition

IRDiRC
RECOGNIZED RESOURCES

EJP RD

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.

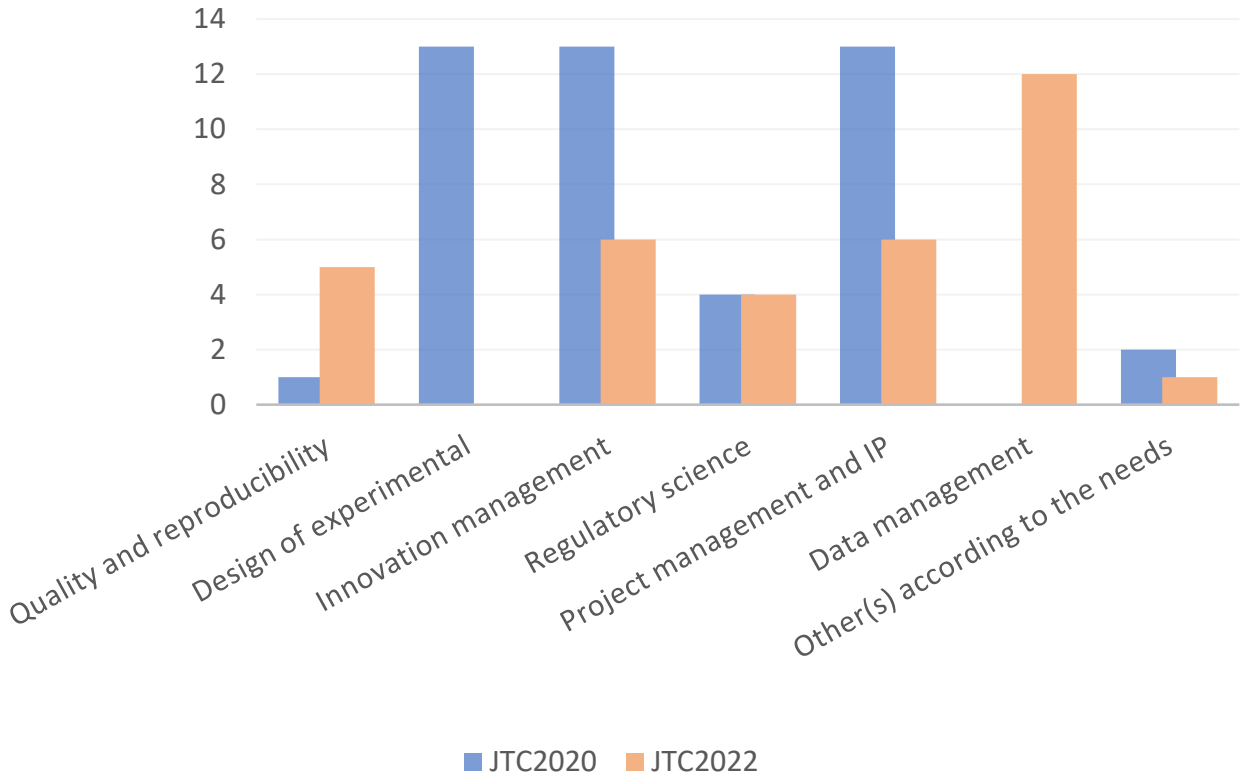
- **42 experts** in database to support the mentoring service and mentoring and legal documents were updated and published on the mentoring webpage

- **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.

Impact

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.**
- **11projects 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**.

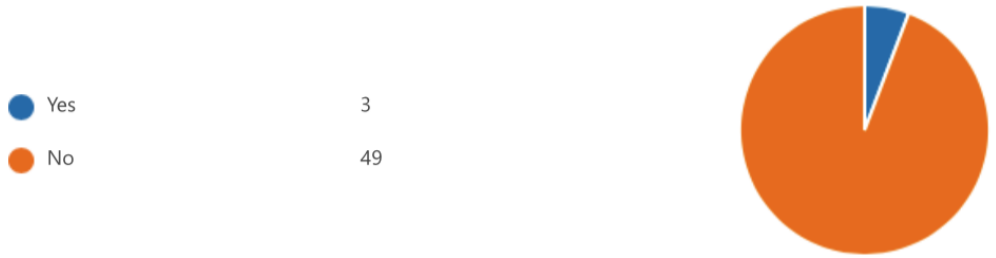
EJP RD

WP19.3 Evaluation of Innovation Management and exploitation support tasks

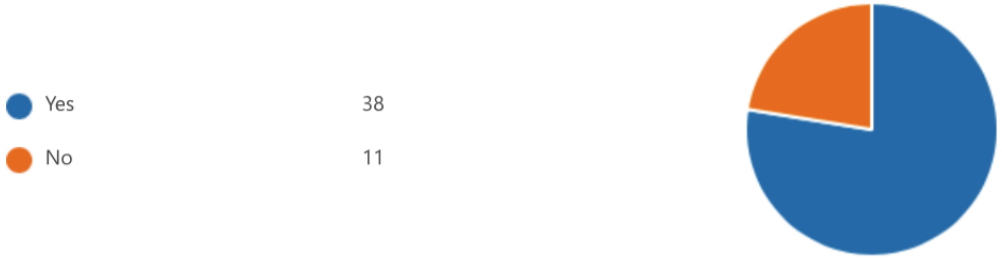
- WP19 has been actively engaging with the research funders (Pillar 1 and Health units at EC), WP3 Sustainability, and Projects such as ERICA (CSA project for the ERNs) and REMEDI4ALL with the intent to disseminate the mentoring service to these communities more broadly and hear back from the community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

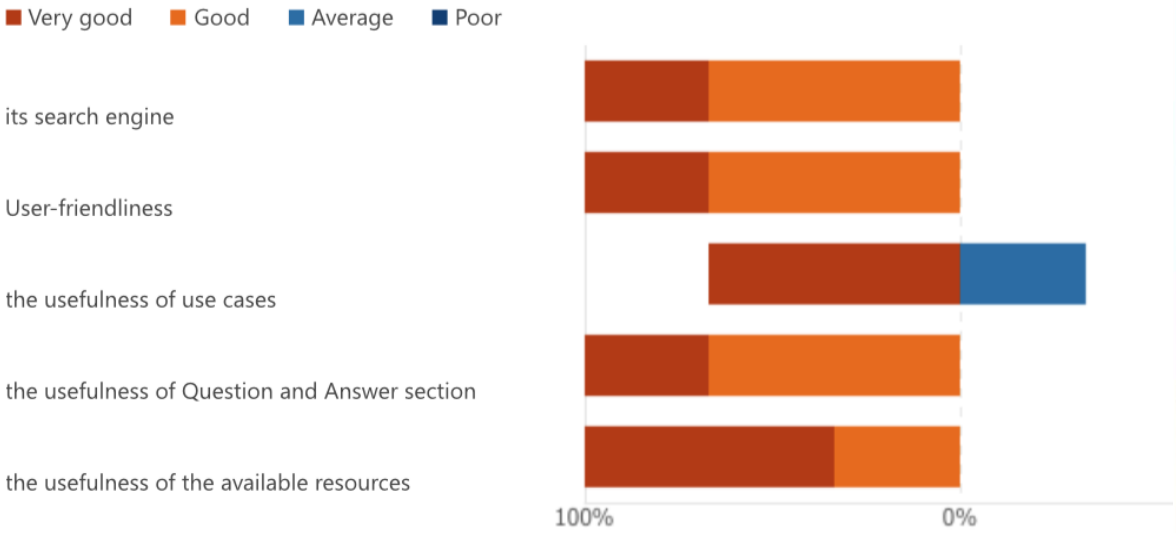
Have you ever used the Innovation Management Toolbox (IMT) offered by the EJP RD?



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



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



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

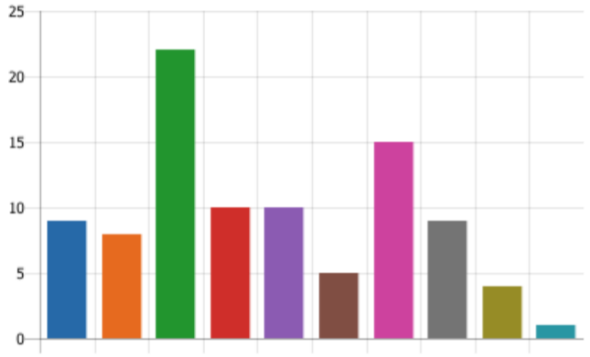
Was your application successful?

Yes	7
No	1
The results are not yet available	0

Do you think that the mentoring contributed to the success of your application?

Yes	4
No	0
Maybe	3

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-...	10
Innovation management and t...	10
Pre-clinical models	5
Project management and mile...	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

EJP RD

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.

EJP RD

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 awarness

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.

EJP RD

WP20 Impact: Decrease fragmentation of rare diseases expertise and research resources

- Building of a community of methodologists involved in smmall 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.

Planned WP20 activities	Achieved by end of year 5	AWP Y6
Demonstration projects	• EPISTOP IDEAL: December 2023 • EBStatmax: July 2023 • Improve PSP	• n/a • n/a • April 2024
Innovation projects	• iSTORE • Evidence RND	• April 2024 • April 2024
Advanced Webinars	• Does Randomization matter in RD clinical trials? • Composite endpoints including patient relevant endpoints (Quality of Life) • The Statistical Evaluation of Surrogate Endpoints in Clinical Trials • Statistical and operational challenges with master protocols • Replicated N-of-1 RCTs for Rare Diseases • Item response models for analysing assessments in rare diseases • EFPIA Webinar: TBD, December 2023 (all available on EJPRD site with papers done or in prep)	• Modelling natural history in longitudinal data-Challenges and Solutions (2024)

Planned WP20 activities	Achieved by end of year 5	AWP Y6
Intermediate course Needs and request of the community	• One course to be provided	• Five courses to be provided
Clinical trial support office	• Support and guide ERNs/RD investigators focusing on the methodological/study design and providing support advice • Raise awareness of the service • Increase the quality and number of RD clinical trials and projects/collaborations for clinical research on RD	• Pursue its achieved work • Uncover ERN roadmap for clinical research to evaluate their current and future needs in terms of support to set up clinical trials. • Communicate and disseminate
Clinical Trial toolbox kit	• Development and implementation of the CT toolbox	• Update and refine and improve the CT toolbox performance • Improve the performance of academic sponsored clinical trials • Communicate and disseminate

Sustainability

PILLAR	ELEMENT/ASSET	TYPE	VALUE-CHARACTERISATION (SUMMARISED)
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 tan 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

EUROPEAN
RARE DISEASES
PARTNERSHIP

SRIA update

EC group of national delegates - 11th meeting
29/06/2023 _ online

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

SRIA Development

- 26 Apr - 4 May — **1st round of selection/reformulation KPI**
- 5 May — **KPI SRIA TF meeting**
- 22 May - 29 May — **2nd round of selection/reformulation KPI**
- 10 May - 14 Jun — **Public consultation of SRIA advanced v1**
- 15 Jun - 20 Jun — **First analysis of the public consultation by Coo**
- 27 Jun - 10 Jul — **KPIs finalisation including methodology for measurement & public consultation results**
- 27 Jun - 11 Jul — **Update SRIA version**
- 12 Jul — **SRIA version 2.0**
- 12 Jul - 20 Aug — **SRIA finalisation**
- 21 Aug — **SRIA final version before submission**

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)

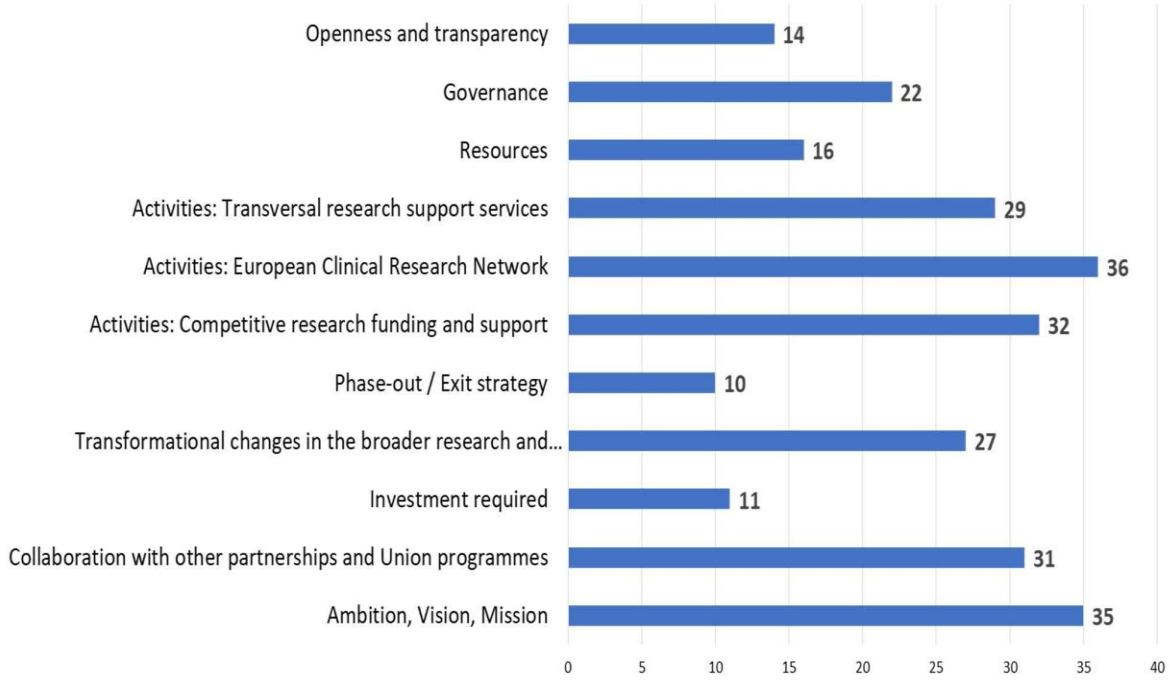
- Help in coordinating the work of the SRIA process and notably of the working groups if created. (But not everything needs to be agreed at the 1st SRIA meeting…)

+ 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!

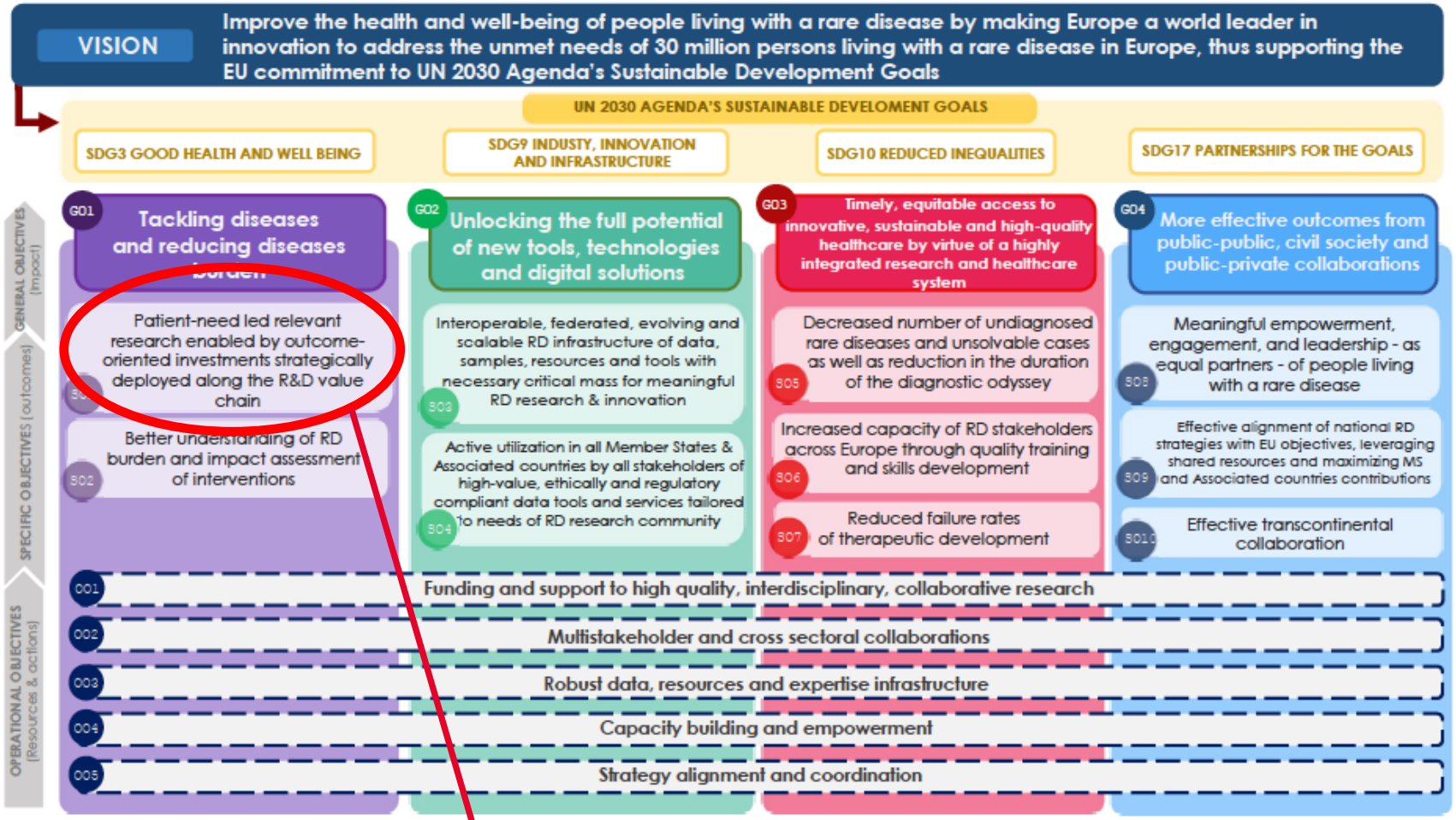
SRIA Task Force composition

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).



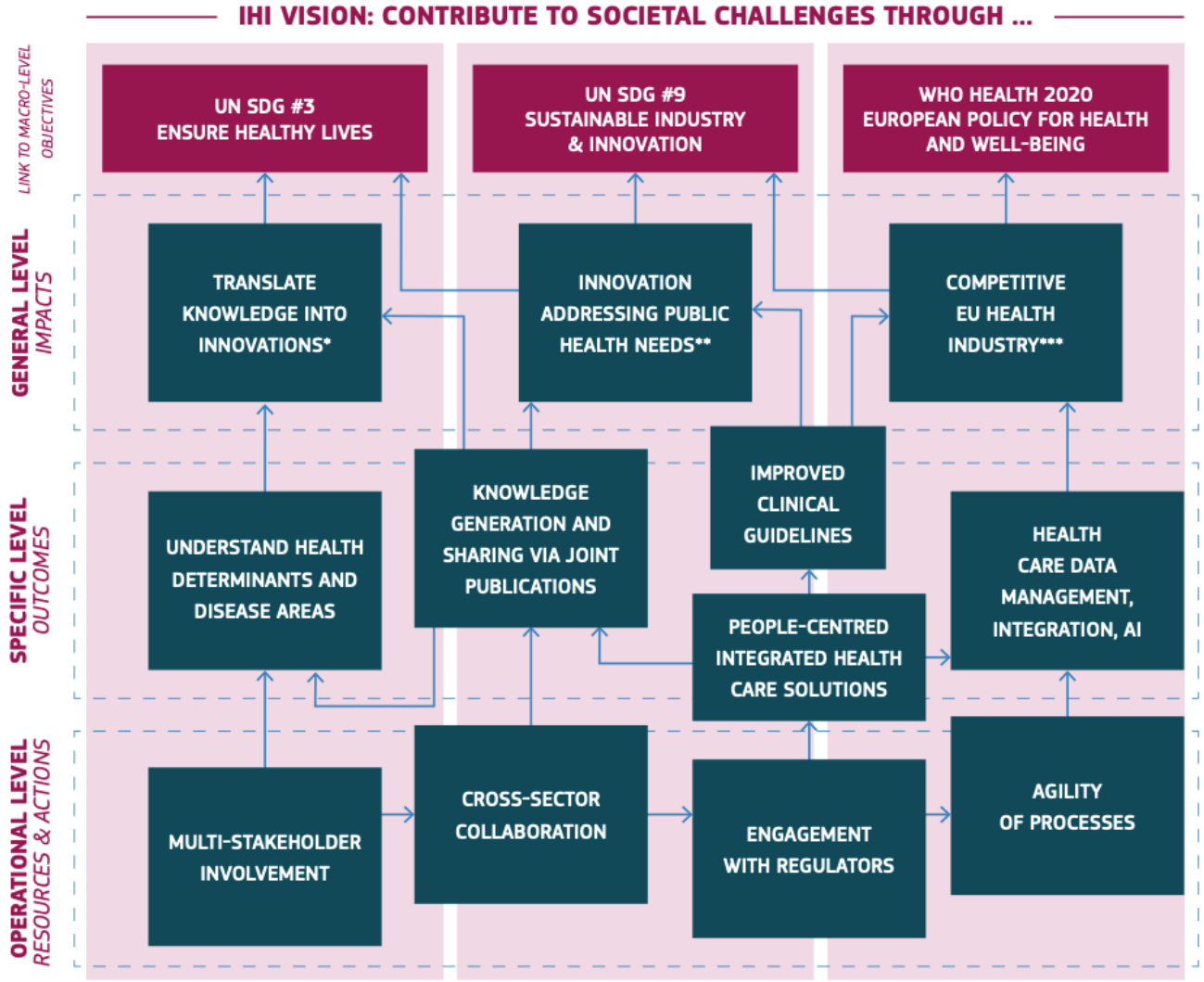
RDP concept paper: on the right track, but…



- 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

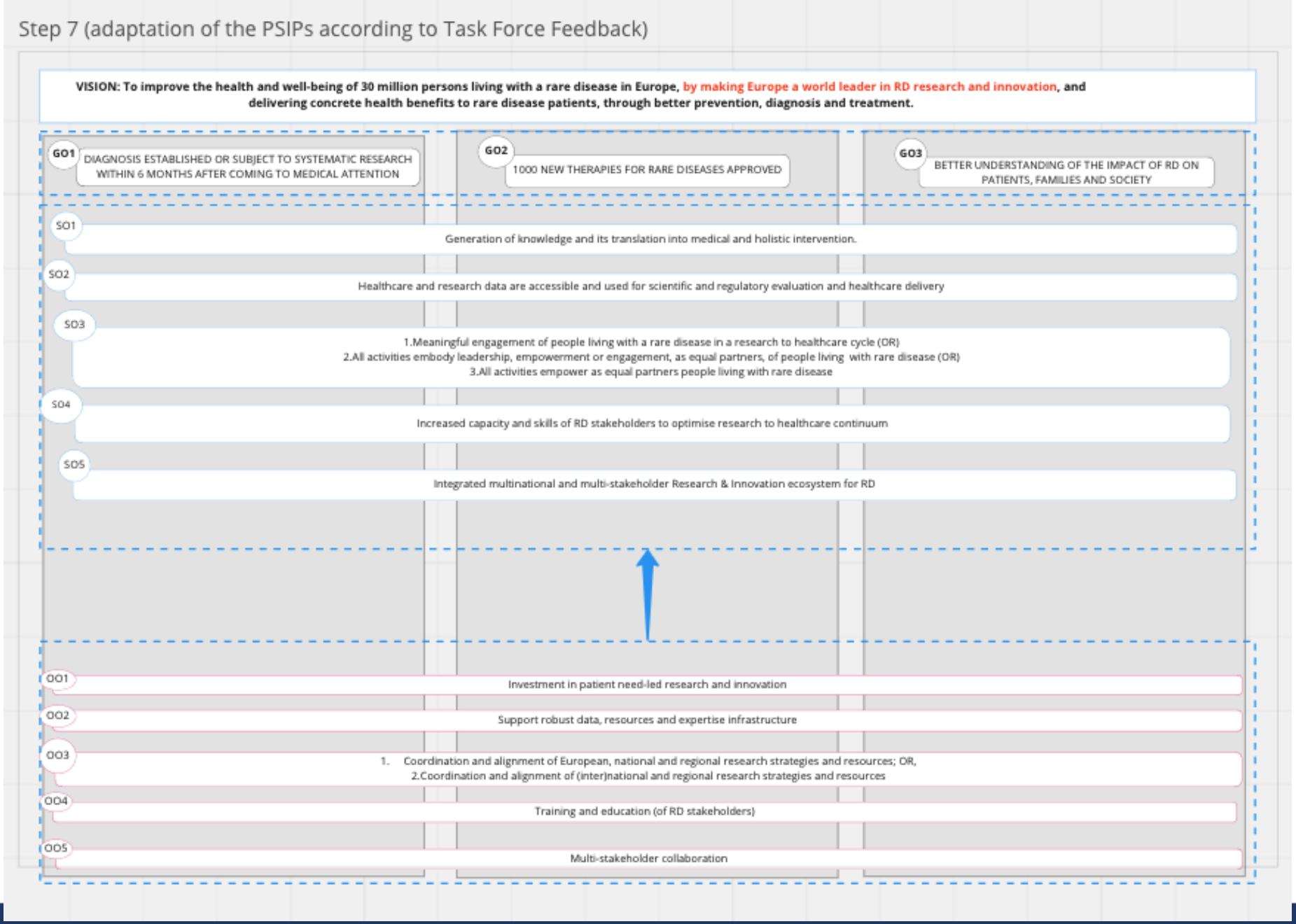


ERA LEARN

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 volounteer experts from SRIA TF group revises
  - Coo adapts and shares with the whole TF group

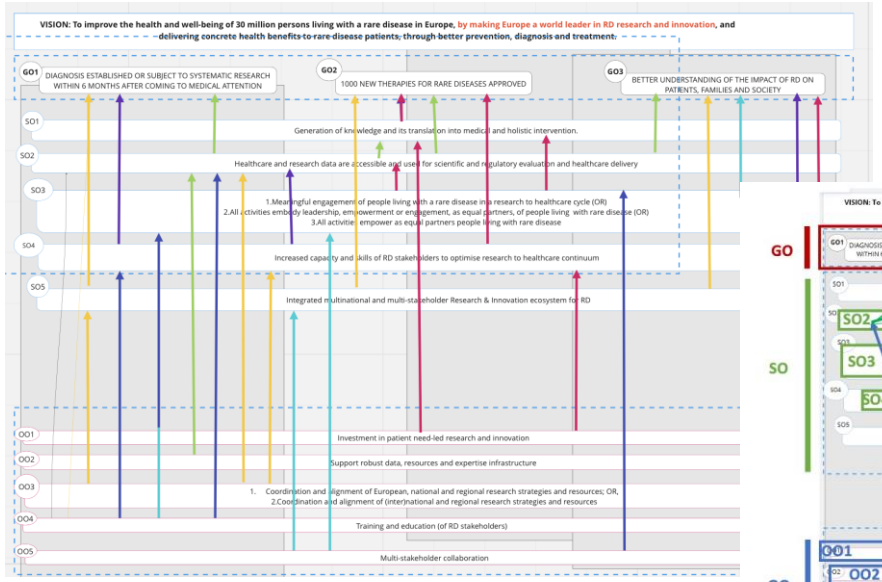
RD Partnership adaptation of the PSIPs based on feedback received

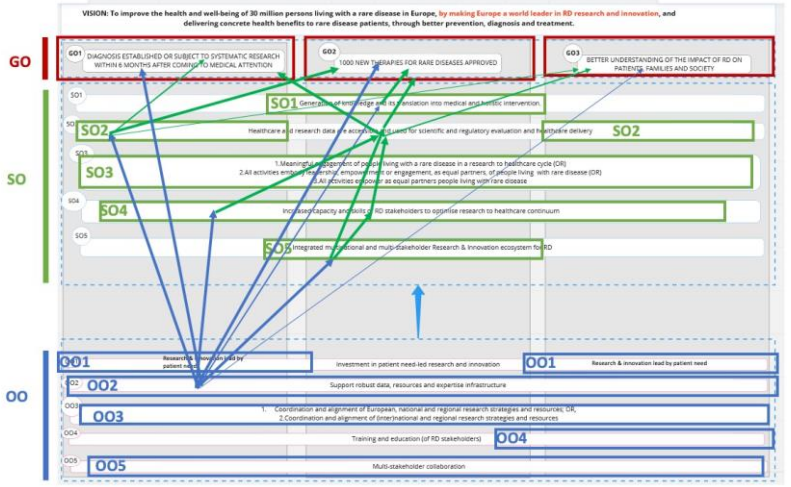


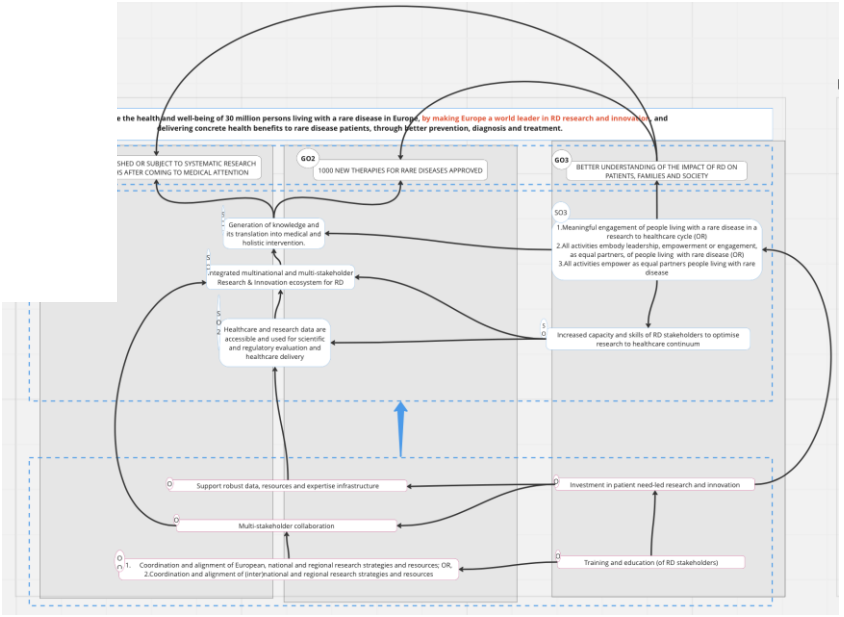
EJP RD

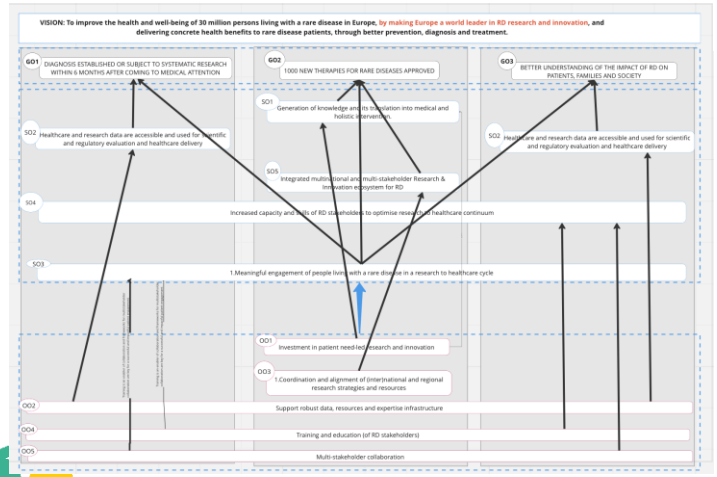
Step 8 (THE PSIPs exercise) 😱 😱 😱

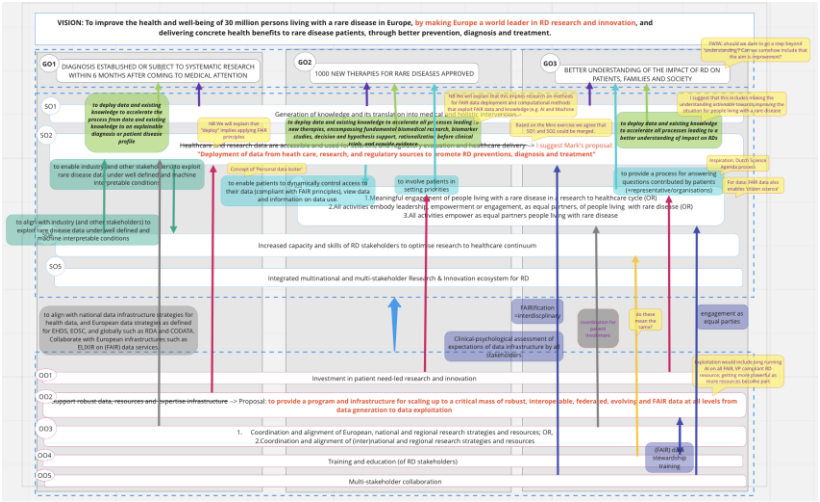
OO1 -> SO5, SO1, SO3, SO2
OO2 -> SO5, SO1, SO2
OO3 -> SO5 (and all other SOs)
OO4 -> SO4
OO5 -> SO4, SO5, SO1
SO1 -> GO2
SO2 -> GO2, GO3

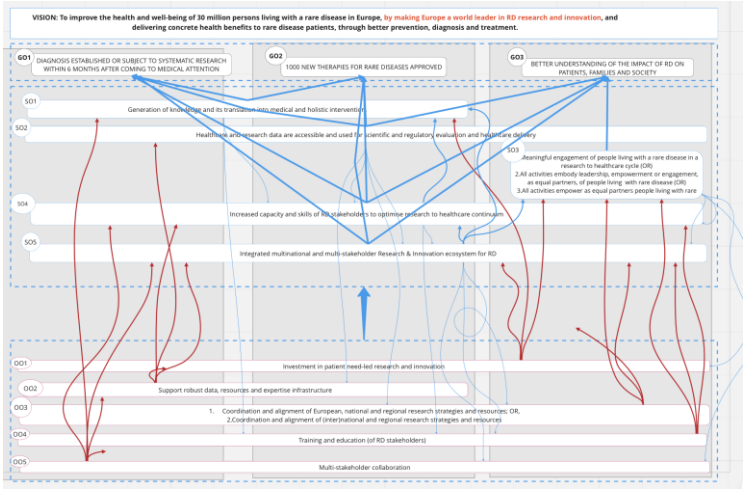










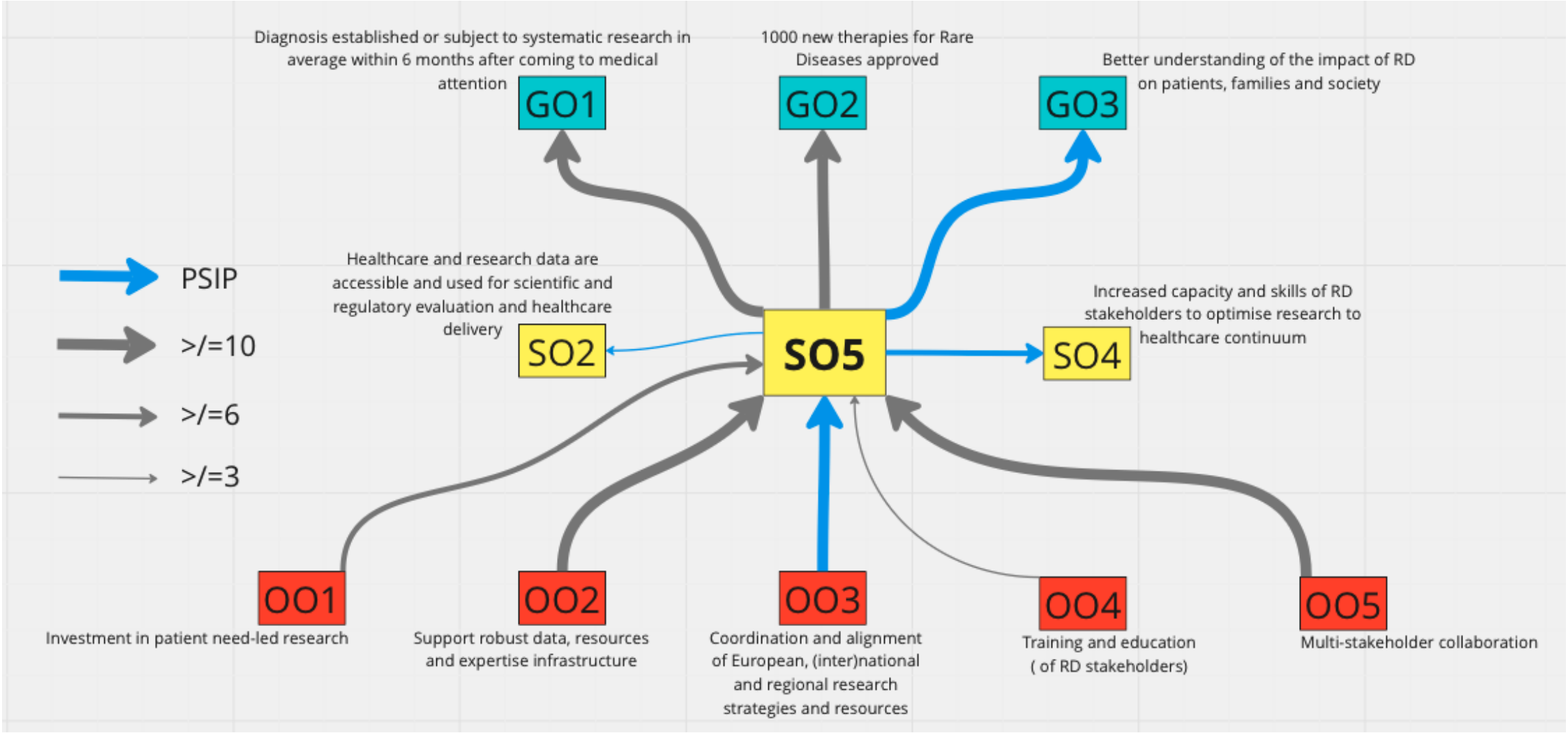


EJP RD

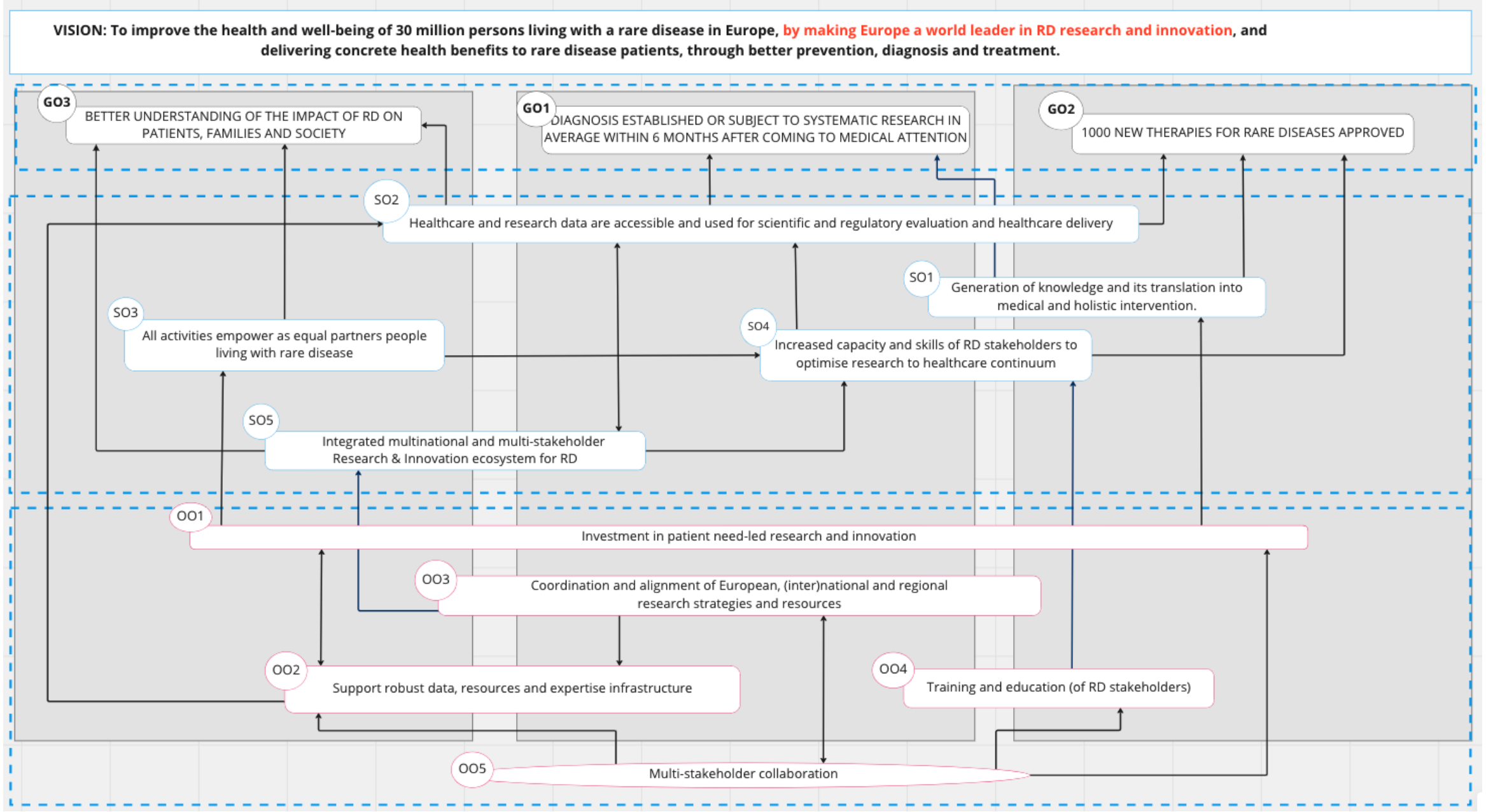
Reminder _ SO specific development methodology

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.
GO3
BETTER UNDERSTANDING OF THE IMPACT OF RD ON PATIENTS, FAMILIES AND SOCIETY
GO1
DIAGNOSIS ESTABLISHED OR SUBJECT TO SYSTEMATIC RESEARCH IN AVERAGE WITHIN 6 MONTHS AFTER COMING TO MEDICAL ATTENTION
GO2
1000 NEW THERAPIES FOR RARE DISEASES APPROVED
SO2
Healthcare and research data are accessible and used for scientific and regulatory evaluation and healthcare delivery
SO1
Generation of knowledge and its translation into medical and holistic intervention.
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
OO1
Investment in patient need-led research and innovation
OO3
Coordination and alignment of European, (inter)national and regional research strategies and resources
OO2
Support robust data, resources and expertise infrastructure
OO4
Training and education (of RD stakeholders)
OO5
Multi-stakeholder collaboration

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)

  See next slide
- **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)

SRIA Layout

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

SRIA _ KPIs

GO1 - DIAGNOSIS ESTABLISHED OR ENROLLMENT IN SYSTEMATIC RESEARCH IN AVERAGE WITHIN 6 MONTHS AFTER COMING TO MEDICAL ATTENTION

- 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.

SRIA _ KPIs

GO2 - 1000 NEW THERAPIES FOR RARE DISEASES APPROVED

- 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

SRIA _ KPIs

GO3 - BETTER UNDERSTANDING OF THE IMPACT OF RD ON PATIENTS, FAMILIES AND SOCIETY

- 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.

SRIA _ KPIs

Current step

Collecting information necessary for KPIs measurement and follow-up

European Partnership [title]		**Monitoring and evaluation framework**, draft 1, [date]			
Overall vision: [max 500 characters]					
Objectives		**What is a measure of success?** Please use quantitative (Key Performance) and qualitative indicators, and link them to a point in time	**Which is the data source and methodology used** [project data, study, ….]	**Who is responsible for monitoring and providing the data / information** **When will it be collected?**	**Baseline and target**
General objectives (linked to impact indicators)	GO1				
	GO2				
	GO3				
Specific objectives* (linked to outcome/result indicators)	SO1				
	SO2				
	SO3				
	SO4				
Operational objectives* (linked to output indicators)	OO1				
	OO2				
	OO3				
	OO4				

*add more lines, as needed.

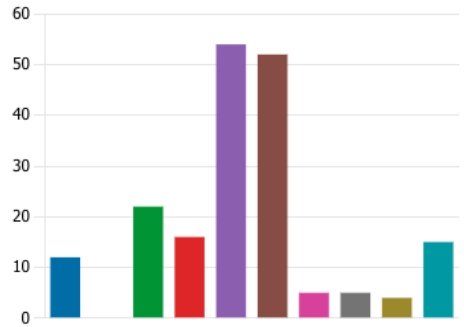
SRIA Public Consultation

Respondent Typology

6. Type of the organisation

More Details

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



138

Responses

9. Do you complete this survey:

More Details Insights

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

More Details Insights

Yes	70
No	64

South United Belgium West Spain / Barcelona Norway / Mid
UK UK/Scotland FRANCE **Spain** **Netherlands**
Norway ITALY Poland **Germany** Germany / Bavaria
Portugal PARIS / FRANCE Spain/Catalonia
South AFrica

Position

Senior Consultant
Patient Vice President Pediatric Head of Department Pediatric Urology
Medical
Clinical **Professor** Research Policy Officer
Director of the center Senior Group leader
Scientific Director Manager researcher
Assistant Professor head of research Associate Professor

Expertise

research clinician researcher - molecular researcher in laminopathies
Clinician research
Pediatrician and researcher Clinician and researcher field geneticists and clinicians
Urology and researcher rare disorders
Expert patient **Clinical** **research** **patient**
diagnostics **rare diseases**
basic researcher
Biomedical researcher clinician scientist Disease Research

SRIA Public Consultation

General Objectives agreement

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 ?

More Details Insights

Yes	132
No	5

12. GO2 : 1000 new therapies for rare diseases approved

Do you agree with the proposed General Objective ?

More Details Insights

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 ?

More Details Insights

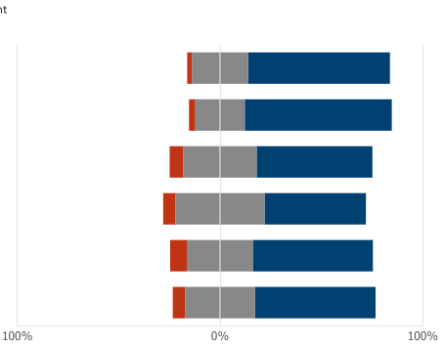
Yes	130
No	5

SRIA Public Consultation

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

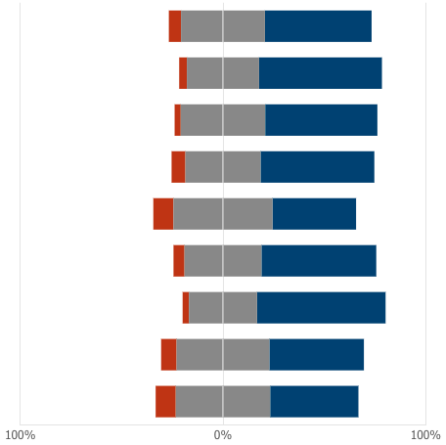
Not so important **Important** **Very important**

SO1



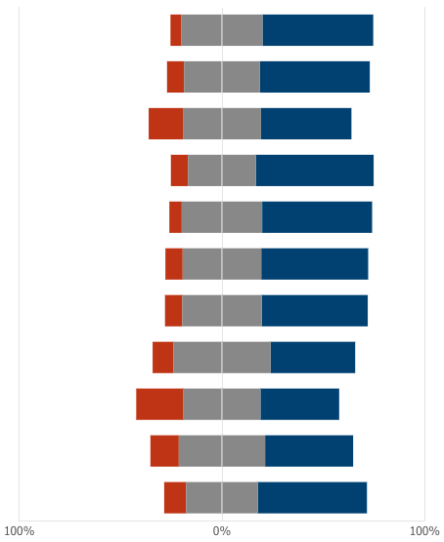
Generation of knowledge and its translation into medical and holistic intervention

SO2



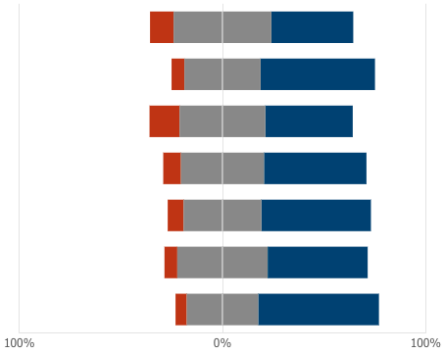
Healthcare and research data are accessible, and used, for scientific and regulatory evaluation and healthcare delivery

SO3



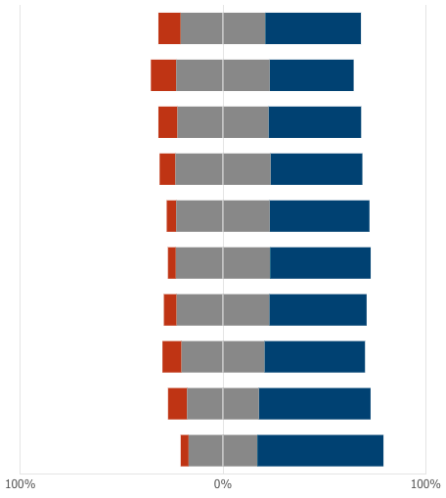
All activities empower, as equal partners, people living with rare disease

SO4



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

SO5



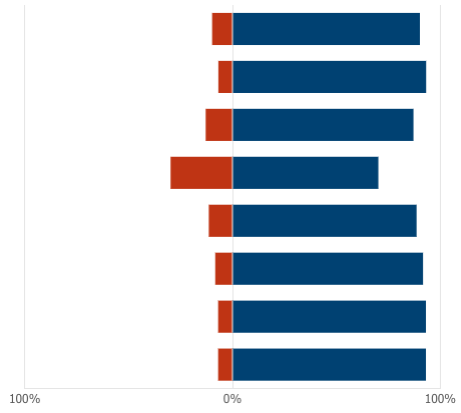
Integrated multinational and multi-stakeholder Research & Innovation ecosystem for RD

SRIA Public Consultation

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

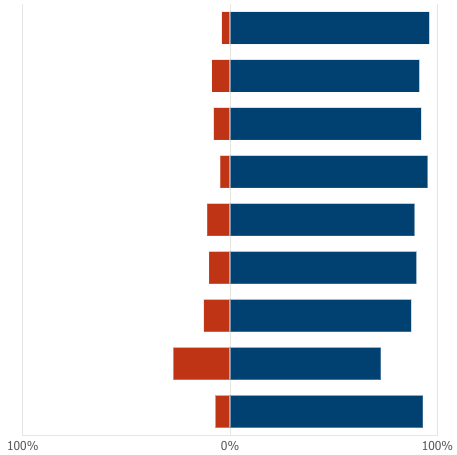
Not relevant **Relevant**

SO1



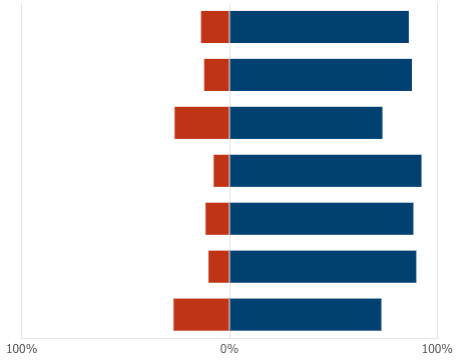
Generation of knowledge and its translation into medical and holistic intervention

SO2



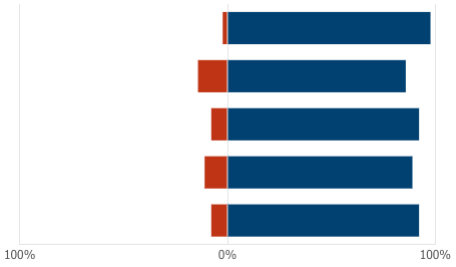
Healthcare and research data are accessible, and used, for scientific and regulatory evaluation and healthcare delivery

SO3



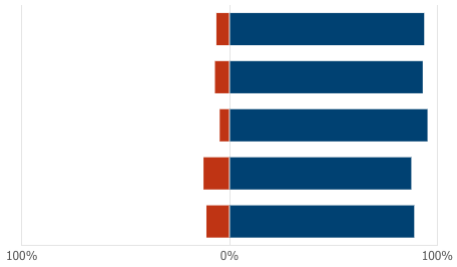
All activities empower, as equal partners, people living with rare disease

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
Comments provided

304
- Comments provided

111
- comments identified by Coo for SRIA-TF & WP Leaders consideration

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.

EUROPEAN
RARE DISEASES
PARTNERSHIP

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".

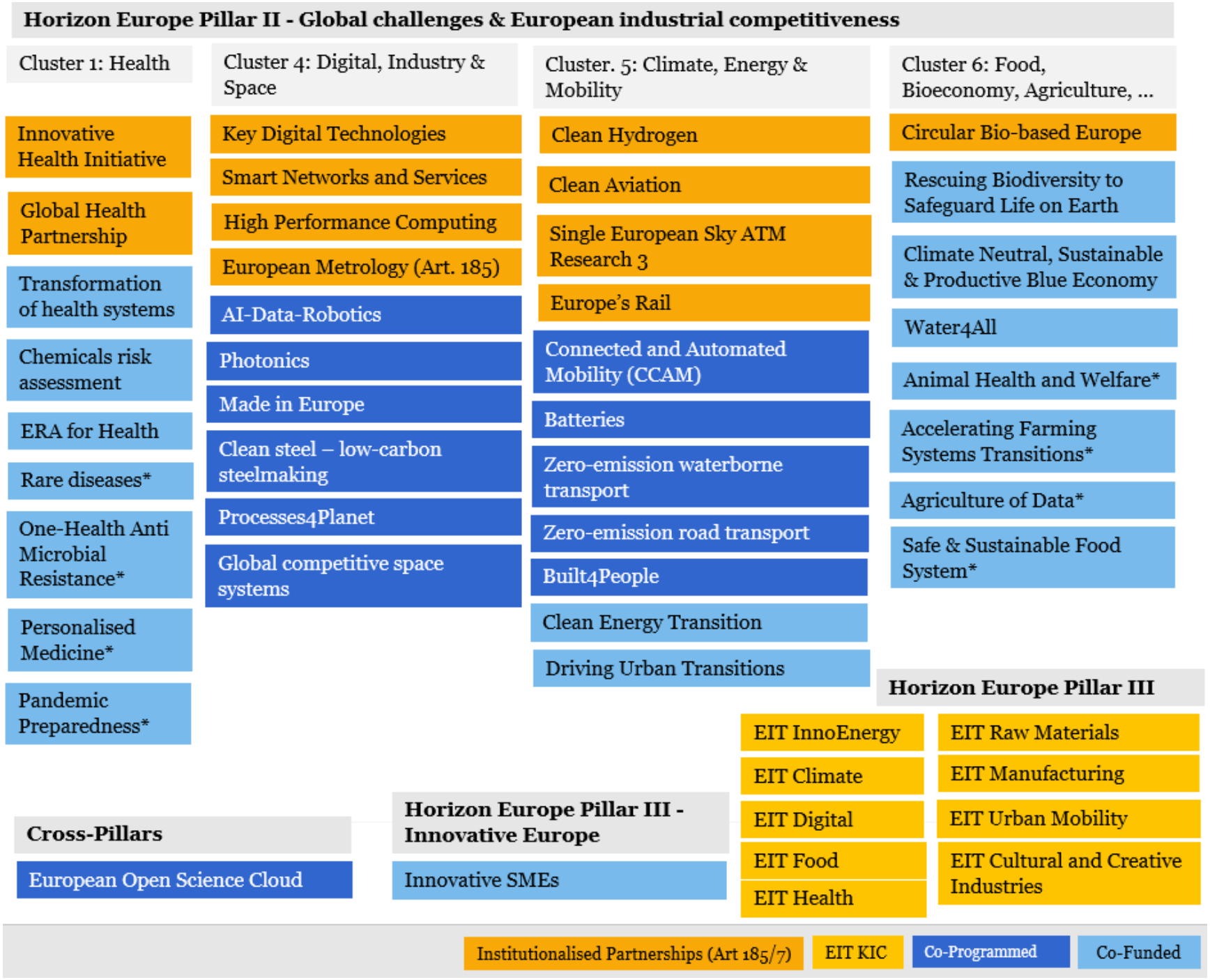
European Partnerships

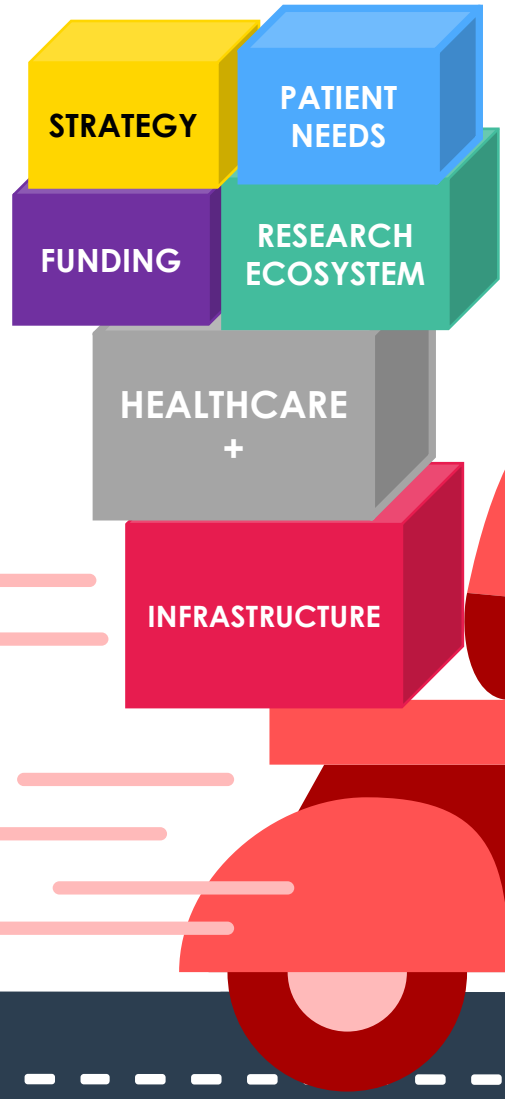
- provide mechanisms to link R&I closely to policy needs
- develop close synergies with national and regional programmes
- bring together a broad range of innovation actors to work towards a common goal
- turn research into socio-economic results

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





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

EJP RD

WHAT ARE THE REMAINING NEEDS?

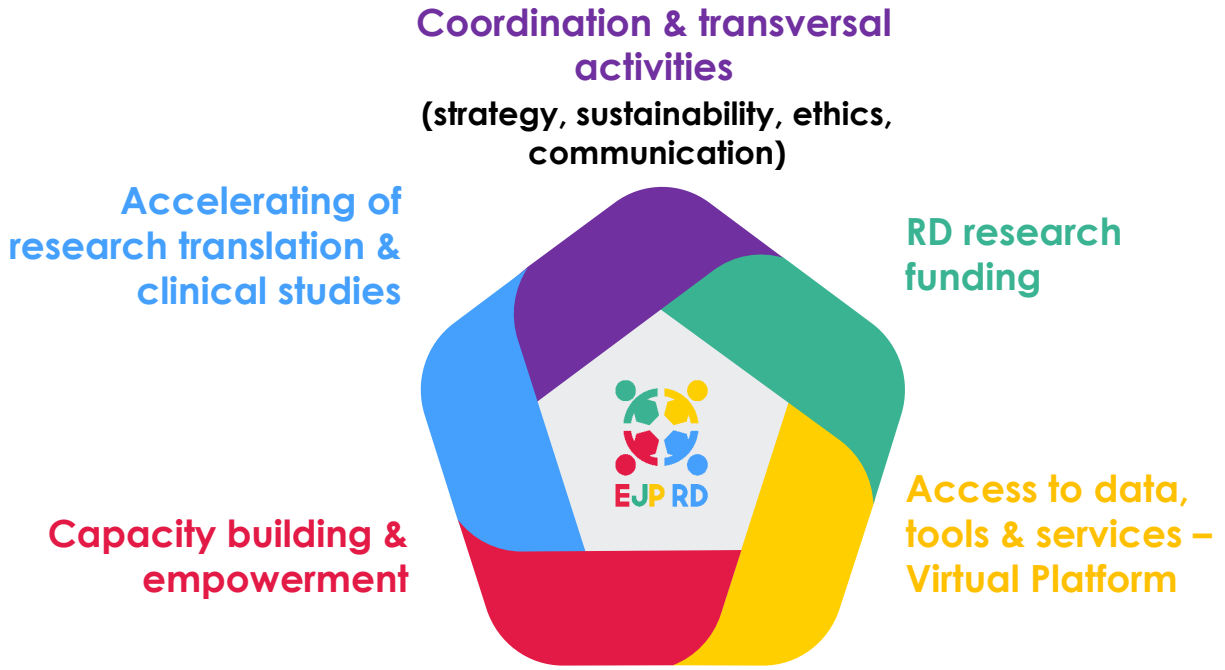
European Joint Programme on Rare Diseases – objectives & structure

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

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

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

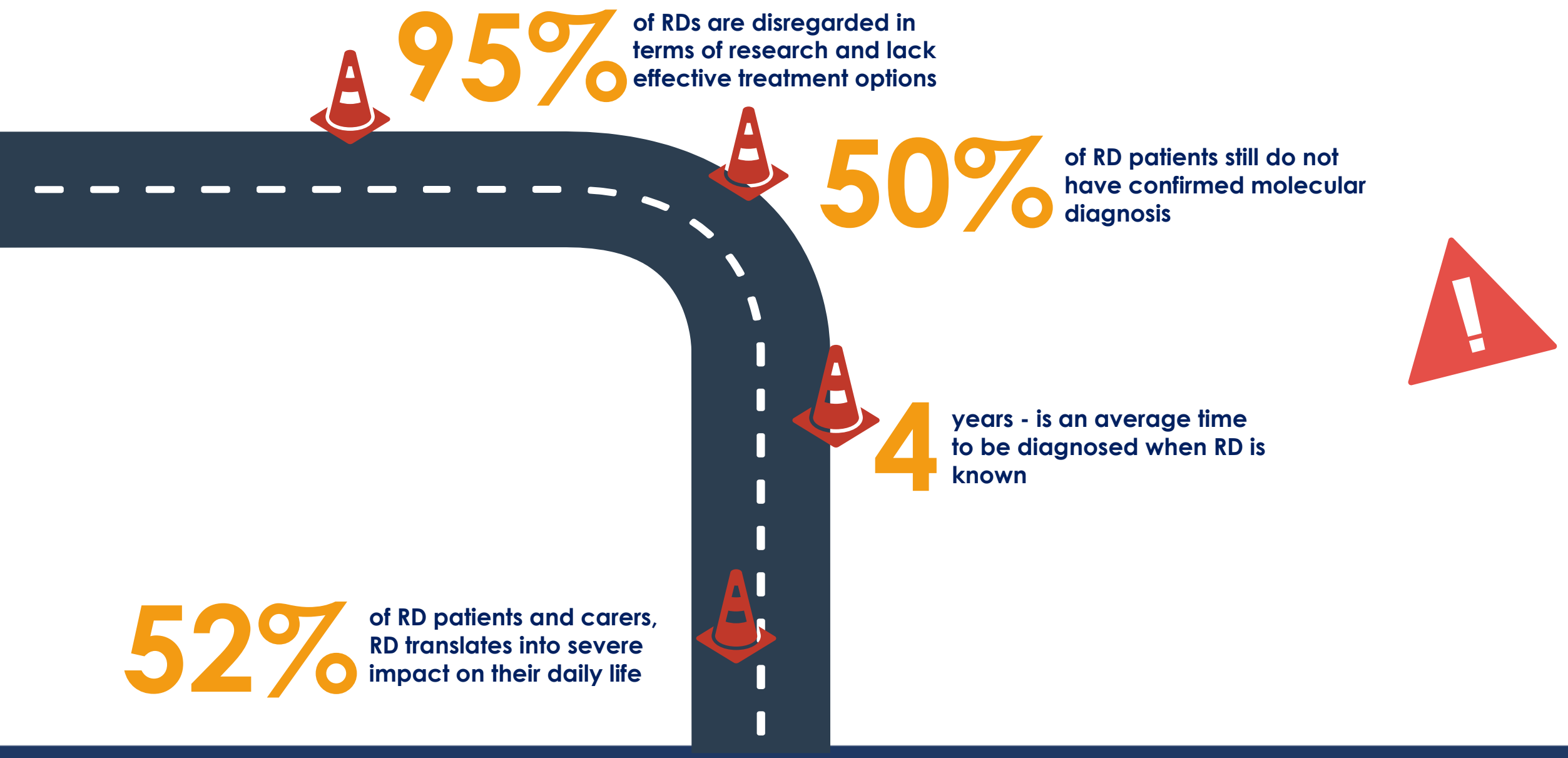
10 hospitals
13 research institutes
31 research funding bodies/ministries
29 universities/hospital universities
5 EU infrastructures
5 charities/foundations
EURORDIS

+ 47 linked third parties

+100% associated networks

EURORDIS RARE DISEASES EUROPE
eatris
elixir
INFRAFRONTIER mouse disease models
European Reference Networks
ECRIN EUROPEAN CLINICAL RESEARCH INFRASTRUCTURE NETWORK
BBMRI-ERIC® gateway for health

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

SUPPORT

robust patient need-led research

UTILIZE

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

Rare Diseases Partnership Vision

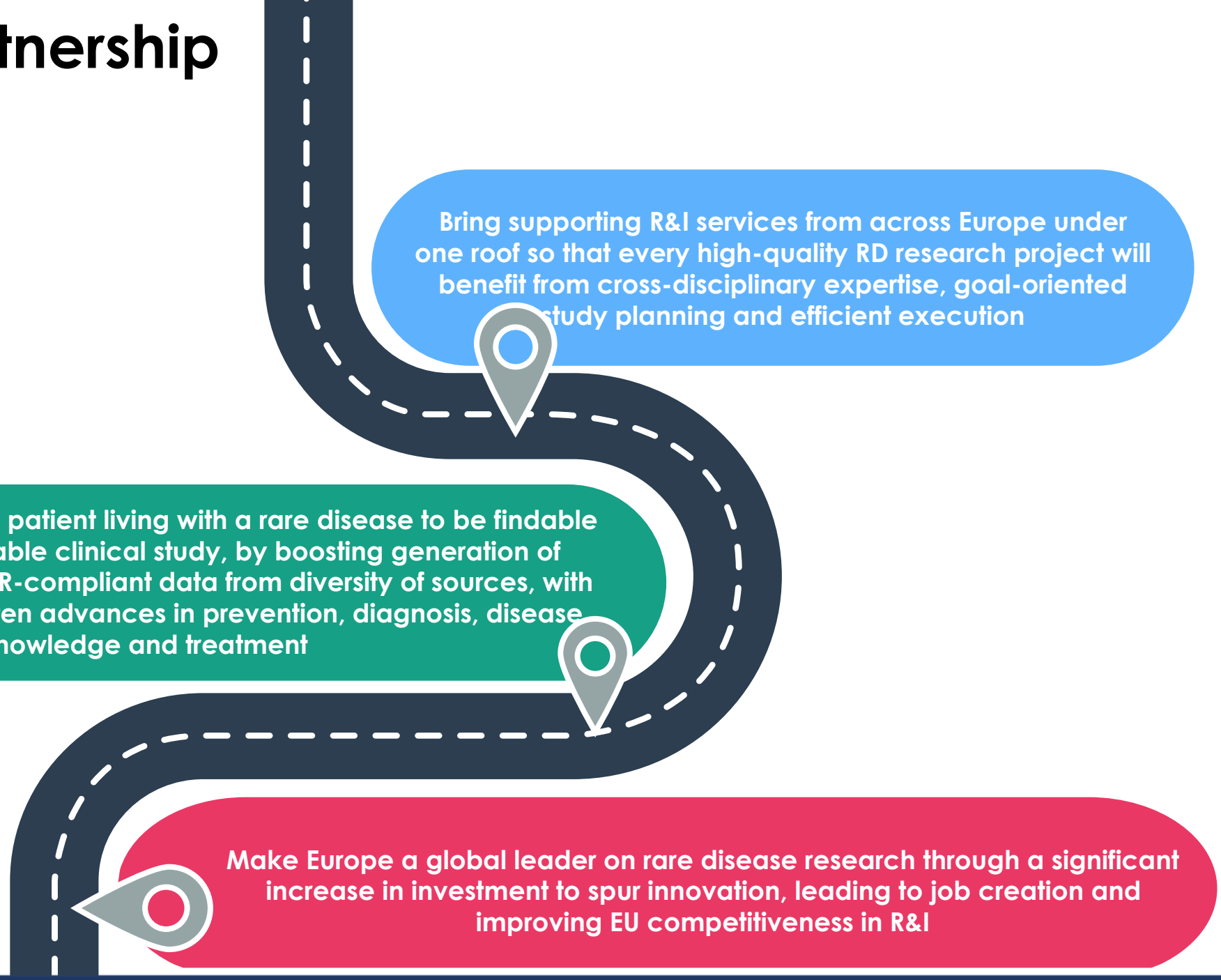
DEVELOP

new treatments and diagnostic pathways

SUPPORT

the coordination and alignment of national and regional research strategies, including the establishment of strong public-private collaborations

Rare Diseases Partnership Mission



Competitive Research Funding and Transversal Support Services

Patient Engagement Facilitation

Networking Support Scheme

Capacity building

Annual Joint Transnational Calls

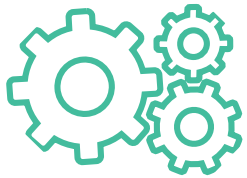
Mentoring service

Acceleration hub

Meaningful and exploitable results

Ethics & regulatory Support

Data integration and coordination services



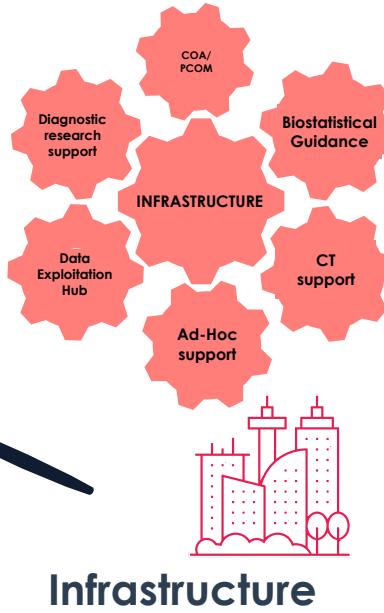
Clinical Research Network

Clinical expertise & capacities

National alignment & capacities

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

Technological expertise



Infrastructure

Public-private collaboration

Accelerated diagnosis
Clinical trial readiness
New therapies

EUROPEAN CLINICAL RESEARCH NETWORK FOR RARE DISEASES

RD Clinical Research Infrastructure
Data exploitation hub
Diagnostic research support
COA/PCOMs support
Biostatistical guidance
Clinical trials support

COMPETITIVE RESEARCH FUNDING AND SUPPORT

Joint Transnational Calls, Networking

Rare Diseases Partnership

TRANSVERSAL SUPPORT SERVICE

Mentoring service
Data integration
Capacity building
Ethics & regulatory support
Acceleration hub

COORDINATION, STRATEGY, GOVERNANCE

Public-private collaboration
Maximisation of national alignment & contributions
Joint multi-stakeholder strategy
Patients as drivers

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

SOME FACTS

More than 290 persons assigned to different WG

19 theme co-leads supporting 6 main work streams

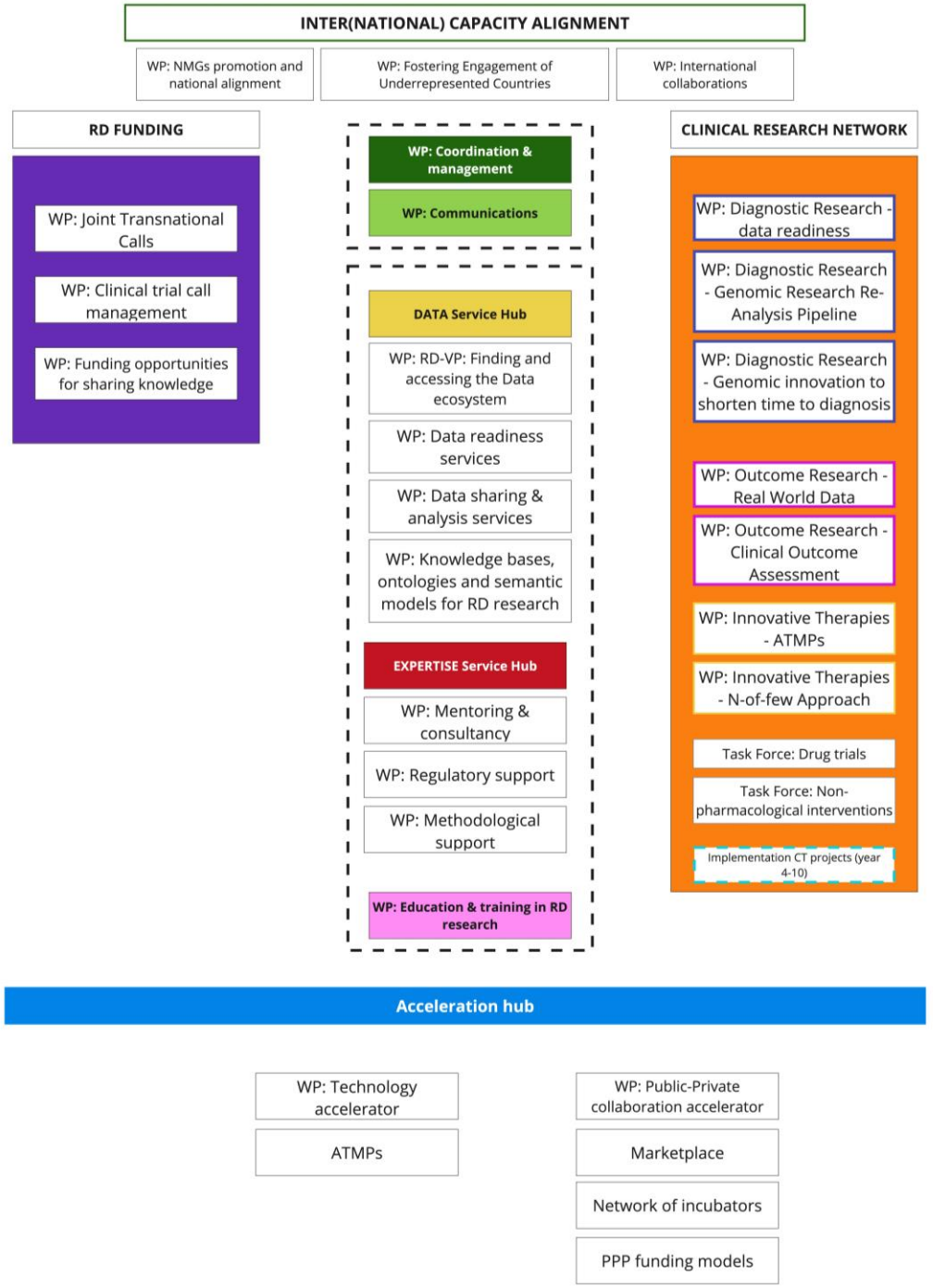
164 LoIs received

39 "group" meetings already took place!

€ 176 405 000
in-cash commitment

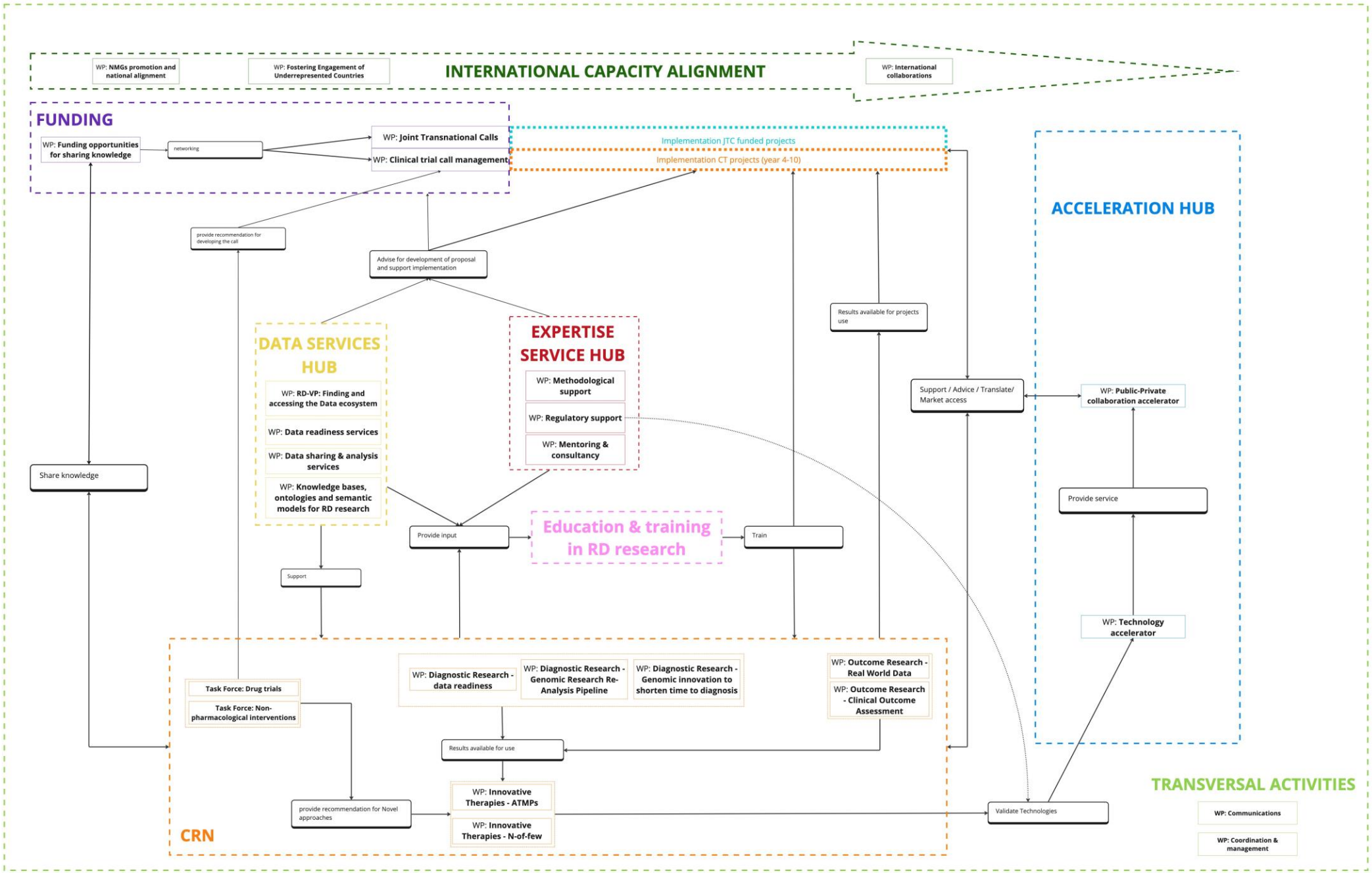
€287 633 474
In-kind commitment

Final Organisation with Work Packages



European Rare Diseases Partnership

Work Package Pathways as an RD Ecosystem



Coordination & communication

Coordination & management

- **Task 1.1** – Project/Programme coordination
- **Task 1.2** – Governance & strategy
- **Task 1.3** – Monitoring of RDP activities
- **Task 1.4** – Data Management Plan
- **Task 1.5** – Sustainability strategy
- **Task 1.6** – Ethics compliance
- **Task 1.7** – IPR management

Communication

- **Task 2.1** – Communication and dissemination (C&D) strategy
- **Task 2.2** – C&D Tactical plans & coordination with partnering organisations to maximise outreach
- **Task 2.3** – Communication tools and support to Work Packages

Activities proposed in this figure are not exhaustive

RD funding

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

CLINICAL RESEARCH NETWORK _ Diagnostic research

Data Readiness

- **Task 6.1 –** Coordinate Pan-European diagnostic research data readiness and collation effort
- **Task 6.2 –** Data standardisation, submission and harmonisation
- **Task 6.3 –** Data archival and data access

Genome re-analysis research pipeline

- **Task 7.1 –** Exome and genome re-analysis pipeline coordination and monitoring
- **Task 7.2 –** Standardised exome and genome re-analysis beyond state-of-the-art diagnostics
- **Task 7.3 –** Novel gene and mechanisms discovery through genome re-analysis
- **Task 7.4 –** Develop and leverage knowledge for variant interpretation
- **Task 7.5 –** Translation to clinic

Genomic innovation to shorten time to diagnosis

- **Task 8.1 –** Enable access of complete genome sequencing for RD in underrepresented countries
- **Task 8.2 –** Enable complete genome sequencing and analysis for RD to shorten time to diagnosis
- **Task 8.3 –** Complete genome mapping for RD to shorten time to diagnosis
- **Task 8.4 –** New genomics/transcriptomics analysis capabilities to understand genetic variation in RD
- **Task 8.5 –** Multi-omics data integration to shorten time to diagnosis in RD

Activities proposed in this figure are not exhaustive

CLINICAL RESEARCH NETWORK _ Outcome research

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

CLINICAL RESEARCH NETWORK _ Innovative therapies

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

TF Objectives

- **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)

Drug Trials

- **Protocol design guidance**
- **Modeling & simulation plan** (study design optimisation)
- Measures to **optimise recruit**ment
- Best practice & working procedures **novel sampling approach**
- **Standardisation of efficacy and safety data collection** (including paediatric-specific variables/meta-data.)
- **Approaches to study preparedness**

Non-pharmacological interventions

- **Review and analyze the current landscape** (current CT, developments and regulatory aspects)
- **Provide guidance for non-pharmacological therapy development** (includ. 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 developpers)

Activities proposed in this figure are not exhaustive

DATA SERVICE HUB

RD-VP: finding & accessing data ecosystem

- **13.1 –** VP/data service hub steering
- **13.2 –** RD-VP evolution and scaling up
- **13.3 –** VP onboarding services

Data readiness service

- **14.1 –** Data ingestion services
- **14.2 –** Services for making data findable, accessible, interoperable, reusable for automated applications
- **14.3 –** Services to prepare data for the regulatory pathway

Data sharing and analysis services

- **15.1 –** Data archiving and sharing services
- **15.2 –** Data analysis infrastructure as a service
- **15.3 –** Global genomics analysis network

Knowledge bases, ontologies & semantic models for RD research

- **16.1** – Creation of a repository of FAIR Patient-centered Outcome Measures (PCOMs)/Patient Reported Outcomes Measures (PROMs)
- **16.2** - Antenatal echographic and pathologic RD phenotypes knowledge base and ontology
- **16.3** – Treatabolome
- **16.4** – Rare disease maps
- **16.5** – Improving and expanding RD Ontologies and semantic models: ORDO,HOOM,HPO, functional impact of RD

Activities proposed in this figure are not exhaustive

Expertise Services Hub

Mentoring and Consultancy

- **Task 17.1 -** Execution of the Mentoring Program
- **Task 17.2 -** Consultancy Services

Regulatory Support

- **Task 18.1 -** Regulatory support to preclinical research
- **Task 18.2 –** Regulatory support to clinical research

Methodological Support

- **Task 19.1 -** Knowledge transfer towards the local clinical trial teams
- **Task 19.2 -** Novel methodologies for the use of all available knowledge, including Real World Data
- **Task 19.3 –** Data analysis methodologies when data are multivariate, hierarchical, incomplete and of differering data types
- **Task 19.4 -** Non-paramedic and randomized-based methodology

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	Fostering engagement of underrepresented countries	RDP Global collaboration
• 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	• 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	• 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

Implement CT

Drug trials, ATMPs and non-pharmacological

Collaborative networks (C4C, industry…)

Plan study

Expertise service hub (CT study design, regulatory, methodology)

CRN CT simulation model(s)

Innovative therapies pipeline (demonstrators & trial readiness)

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:

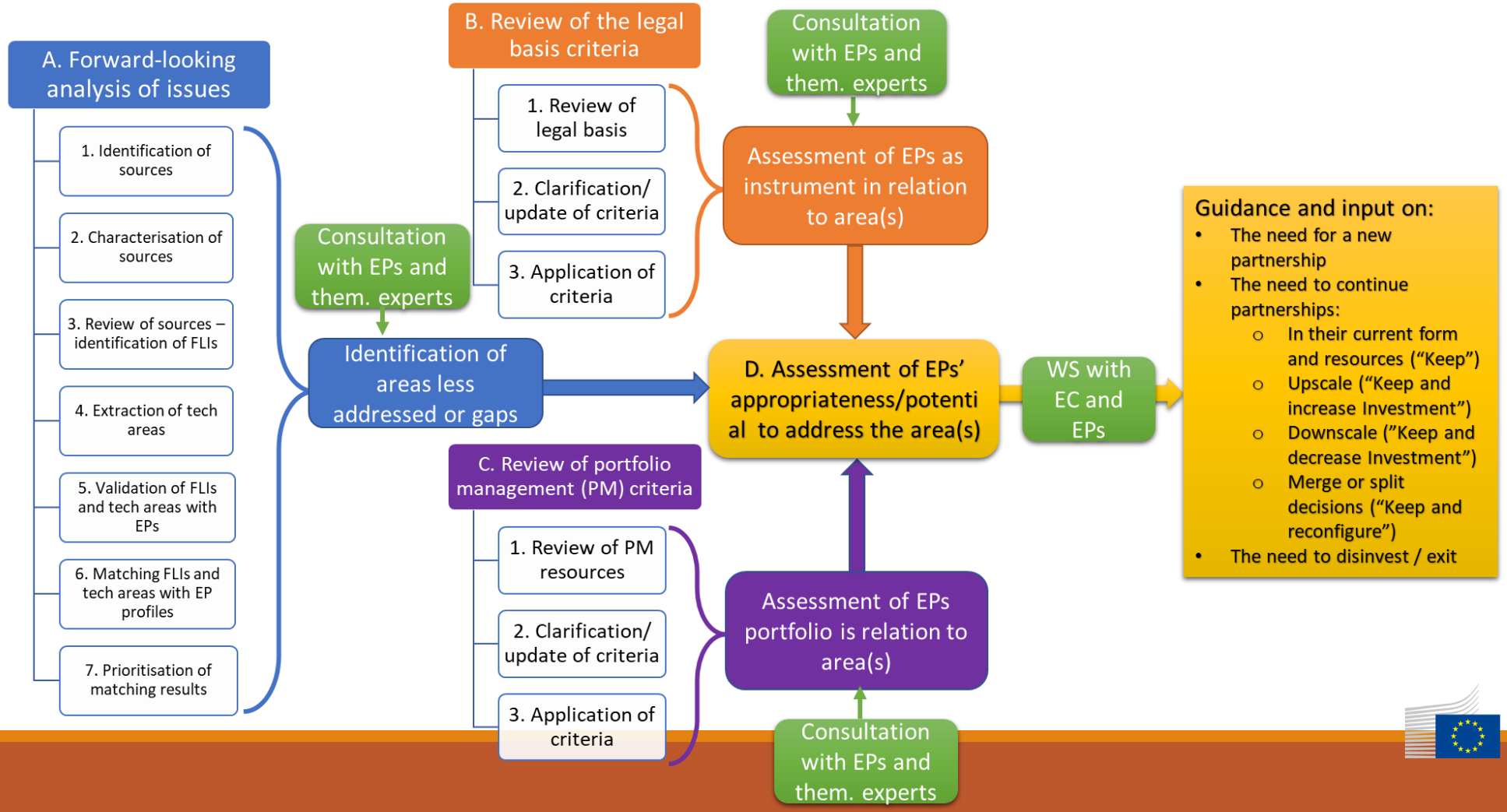
The **Expert Group for support of the Strategic Coordination process for Partnerships**[4] was entrusted with three tasks:

- 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



C. Review of portfolio-management criteria

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

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

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

SRIA

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 Develoment Fund
- European Social Fund Plus (ESF+)
- Invest EU

FUNDING

ERA4Health Partnership

PerMed

ERDRI

SPIDER

EOSC The European Open Science Cloud

EOSC-Life

DATA SERVICE HUB

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

DARWIN EU

ACCELERATOR HUB / Innovation

INNOVATIVE SME

innovative health initiative

EHDS
HealthData@EU Pilot

JARDIN

European Genomic Data Infrastructure

RARE DISEASE MOONSHOT
SCALING UP PUBLIC-PRIVATE PARTNERSHIPS TO ACCELERATE RESEARCH IN RARE DISEASES

eit Health

SCREEN 4CARE

THCS
TRANSFORMING HEALTH AND CARE SYSTEMS

EDUCATION & TRAINING

CRN (including Healthcare pathway, diagnostic)

THANK YOU

www.ejprarediseases.org

coordination@ejprarediseases.org

helpdesk@ejprarediseases.org

Follow us on social media

@EJPRarediseases

The EJP RD initiative has received funding from the European Union's Horizon 2020 research and innovation programme under grant agreement N°825575

EUROPEAN JOINT PROGRAMME RARE DISEASES