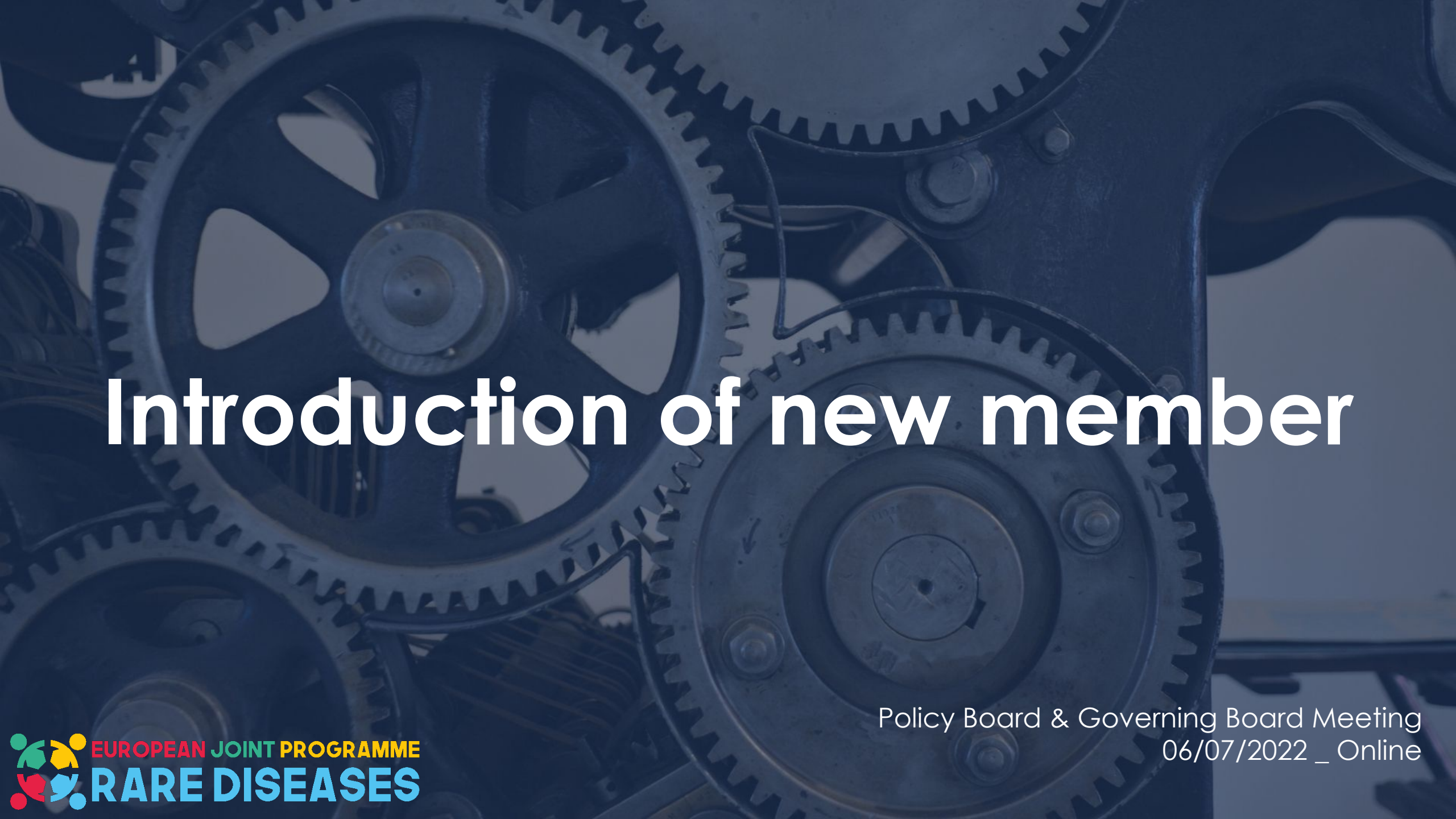
European Joint Programme on Rare Diseases

Policy Board & Governing Board Meeting 2022

Agenda

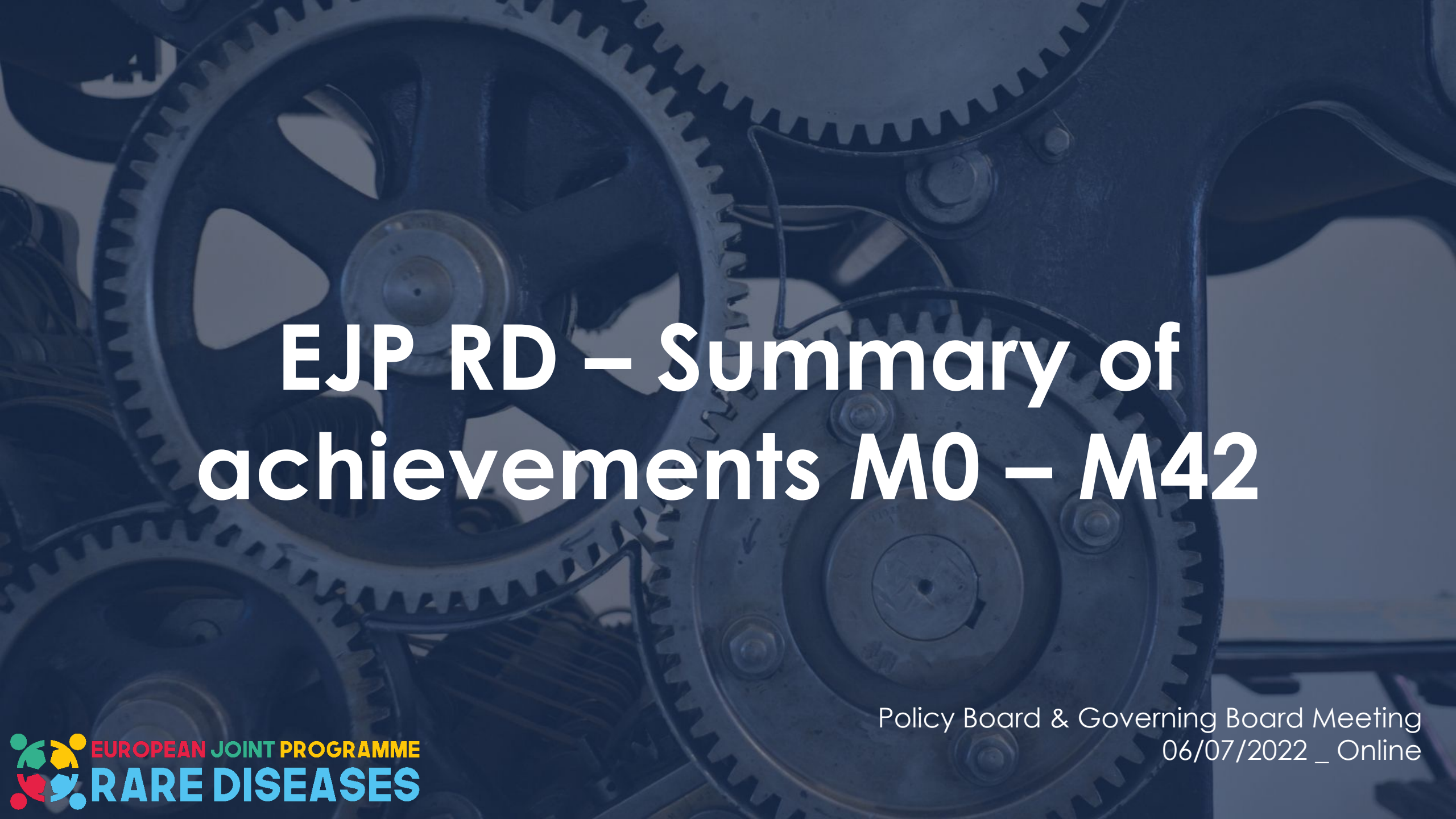
13:30 – 13:40	**Welcome from coordination and introduction of new members**	EJP RD Coordination
13:40 – 14:10	**EJP RD – Summary of achievements M0 – M42**	EJP RD Coordination
14:10 – 14:25	**EJP RD Pillar 3: Training gaps identified, and solutions proposed**	Birute Tumiene
14:25 – 15:40	**Annual Work Plan Year 5** Feedback from the Boards	Pillar Leaders & All
15:40 – 15:55	*Coffee break*	
15:55 – 17:10	**Rare Diseases Partnership** Update on the Concept Paper and timeline Feedback from the Boards	EJP RD coordination & All
17:10 – 17:55	**EJP RD sustainability**	EJP RD WP3 partners & All
17:55 – 18:00	**AOB, Next steps**	EJP RD Coordination



Introduction of new member

Policy Board & Governing Board Meeting
06/07/2022 _ Online

EUROPEAN JOINT PROGRAMME
RARE DISEASES



EJP RD – Summary of achievements M0 – M42

Policy Board & Governing Board Meeting
06/07/2022 _ Online

EUROPEAN JOINT PROGRAMME
RARE DISEASES

EJP RD

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

EJP RD in numbers

+2300 people

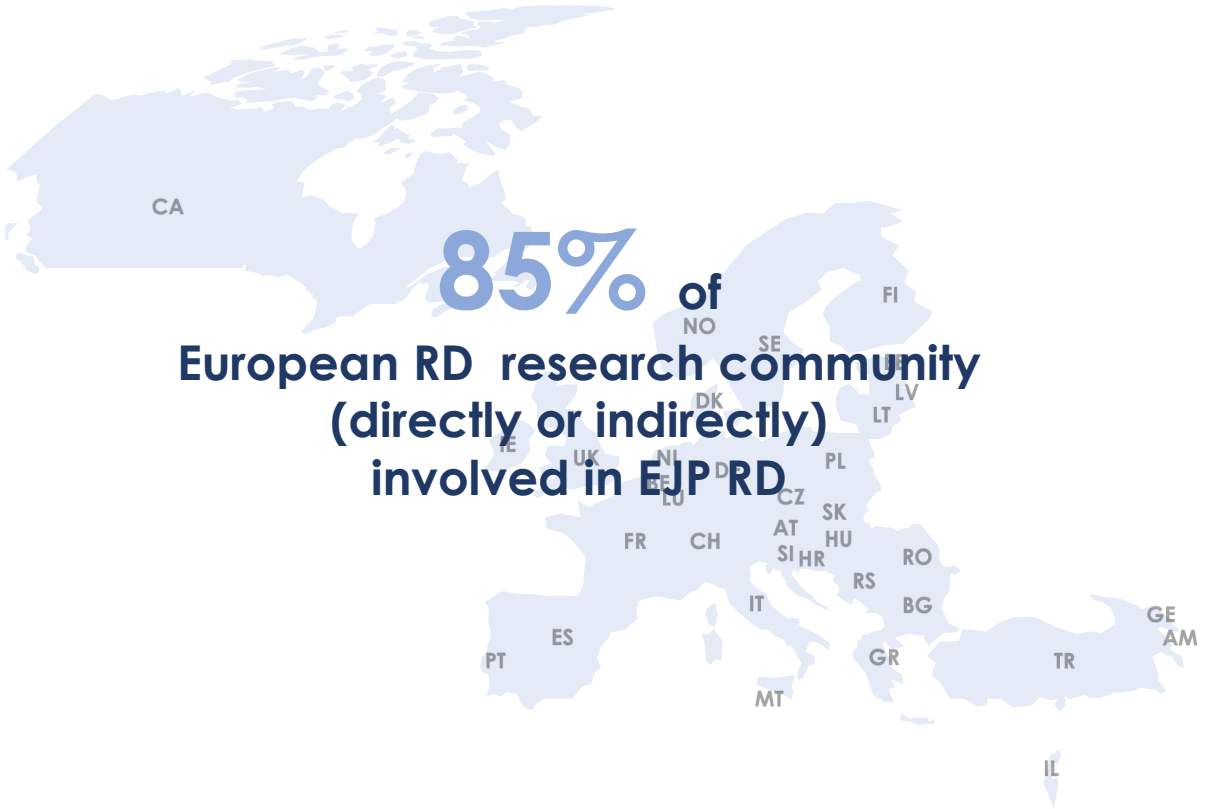
35 participating countries

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

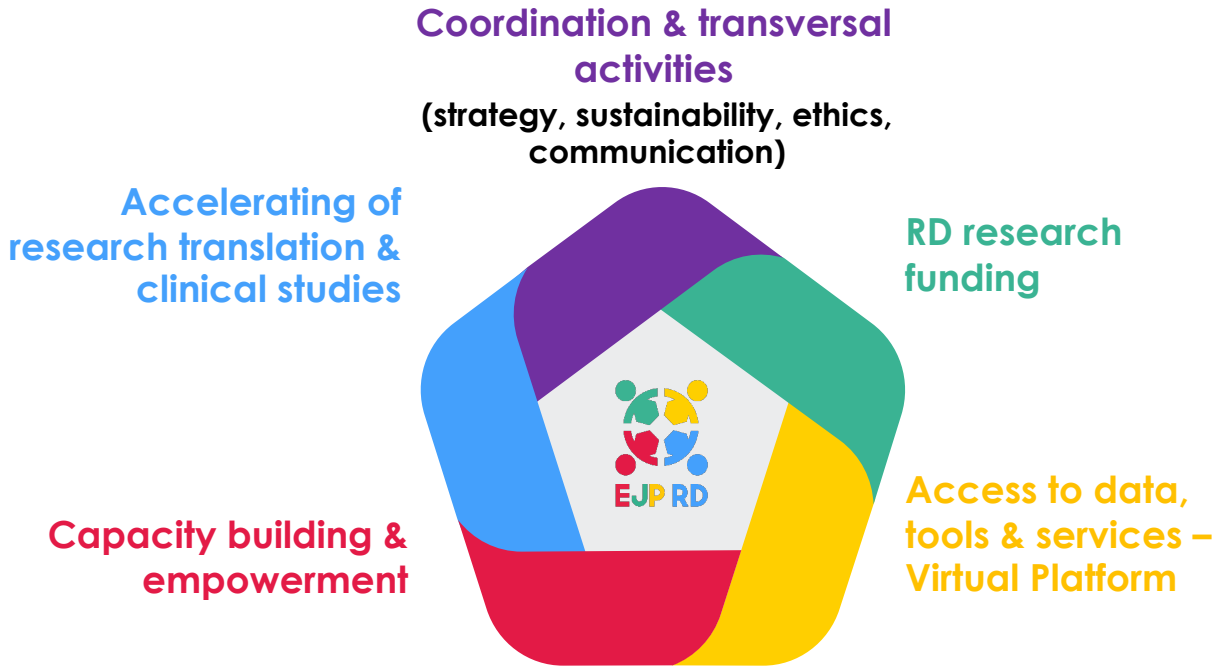
European Joint Programme on Rare Diseases – objectives & structure

- **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 Achievements 2019– June 2022

EUROPEAN JOINT PROGRAMME RARE DISEASES

Policy Board & Governing Board Meeting
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Overall impact: Improved alignment of national/regional activities and policies in RD

- **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. 14 200 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.

- **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.

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

- **The EJP RD standardization work is featured in the calls for projects** of the European Commission/Innovative Medicines Initiative and national calls as reference/recommendation that needs to be taken into account by applicants.

- **The work between ERNs and EJP RD on the registries and related Informed Consent Form** resulted in adaptation of the original ICF template (provided by the European Commission) to include national specificities and facilitate the validation of ERN registries by national ethics committees

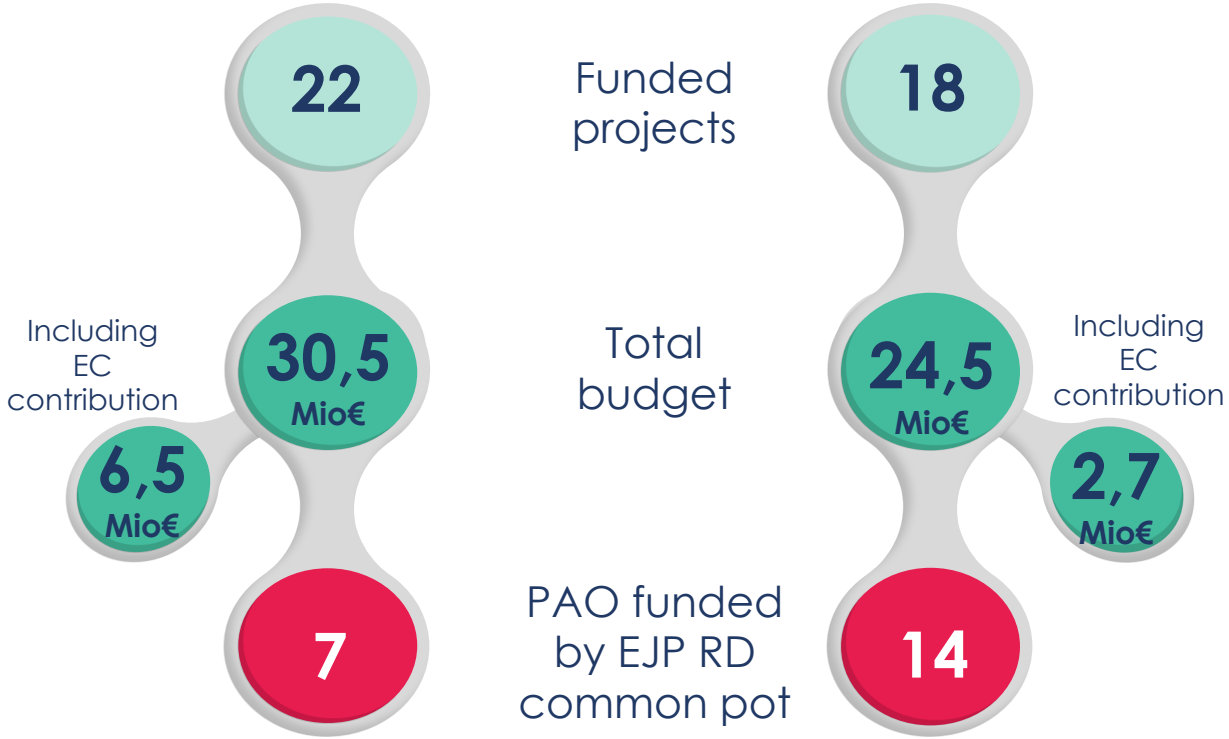
- **Between 23 and 86,6% of national activities are aligned or complementary to EJP RD** actions (23% for P4 innovative methodologies in CTs and 86% for support of data repositories and tools)

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

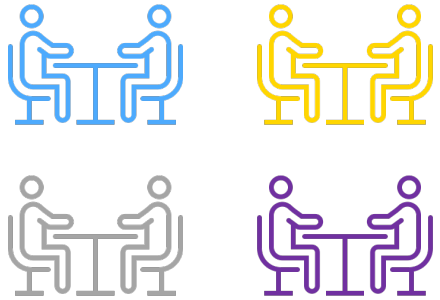
INVESTMENT IN RESEARCH

JTC 2019: Research to accelerate diagnosis and/or explore disease progression and mechanisms of rare diseases

JTC 2020: Pre-clinical research to develop effective therapies for rare diseases



ACCELERATE RESEARCH BY MENTORING



70+ experts recruited to provide mentoring for the research planning, funding and execution process

Since 2019, **44** requests were received.
30 projects were mentored.

2022 **Currently 16 total mentoring requests from JTC 2022 applicants, Follow-on mentoring for JTC 202.**

JTC 2022 : Development of new analytic tools and pathways to accelerate diagnosis and facilitate diagnostic monitoring of RD

EJP RD

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

Involvement and Capacity Building of Patients

- 275 Patient and patient advocates trained
  - 130 RD patients trained in medicine research and development
  - 140 RD patient advocates trained in translational research and scientific innovation
  - 14 Paediatric patients trained
- Participation of the patients associations to the JTC projects : from 45% in JTC2019 to **100 %**in JTC2021

BEYOND-OMICS APPROACHES

- Rare disease portal on WikiPathways: **100 RD pathways created** to date http://raredisease.wikipathways.org
- **Network approaches** (improving & accelerating rare disease diagnostics by combining several pathways and using multiomic data) **available & being expanded** to include **genetic variants** information and **nutritional data**.
  - The relating multi-omics analysis workflows on Congenital Anomalies of the Kidney and Urinary Tract (**CAKUT**) has been **made ready to be provided through the VP for reuse and reproducibility**.
- Variant interpretation tools and interfaces have been enhanced enabling accurate variant interpretation (for rare diseases diagnosis).

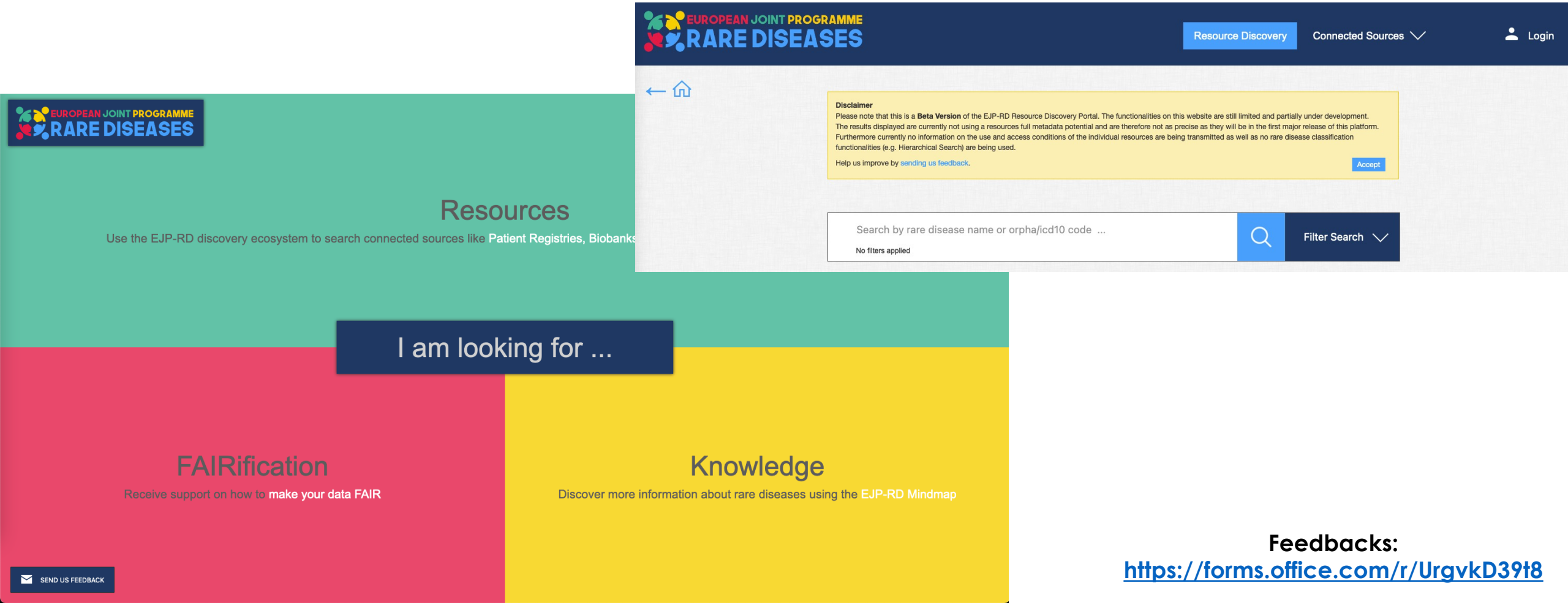
Launching of the Virtual Platform

EJP RD

Virtual Platform Version 0

Under development – guided by users feedbacks

https://vp.ejprarediseases.org/



Feedbacks:
https://forms.office.com/r/UrgvkD39t8

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

EJP RD Helpdesk

- over **450 experts** in the current database
- Expansion to other resources (paediatric, regulatory expertise from other networks)

Participation of under-represented countries

- JTC 2020 : 14 new partners included in full proposals for involvement of under-represented countries
- 22% of all training courses were provided in EU13 countries

Among different types of stakeholders:

- 260 patient advocates and 650 researchers trained in 2019-2022
- 24 research-focused trainings delivered, 650 participants in total (around 25-30 participants per training)
- 1965 persons enrolled in the MOOC training on RD diagnosis so far

*under-represented countries: Bulgaria, Croatia, Cyprus, Czech Republic, Estonia, Hungary, Latvia, Lithuania, Malta, Poland, Romania, Slovakia and Slovenia

EJP RD

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

Resources for Research enhanced

Working locally to make the whole ecosystem sustainable

- Improved user experience and usefulness (unique login, R2R connections)
- Improvements for RD data archive, discovery and access: **adapted for RD**
- Increased number of data collected for RD researchers
- Increased awareness (resource webinars)

--> 19 new series of webinars, new trainings and documentations have improved the access and usability of 11 resources addressing RD research specific needs

EJP RD

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

Rare Diseases Research Challenges

Three Rare Diseases Research challenges public-private projects were supported to advance innovation in RD field and shall lead to

- (i) development of a non-invasive tools for measuring RD patient mobility in daily living;
- (ii) delivery system for intranasal administration of biological drugs to neonates and
- (iii) development of pre-clinical assay to detect instability of microsatellite repeat expansions.

Adaptation of the methodologies

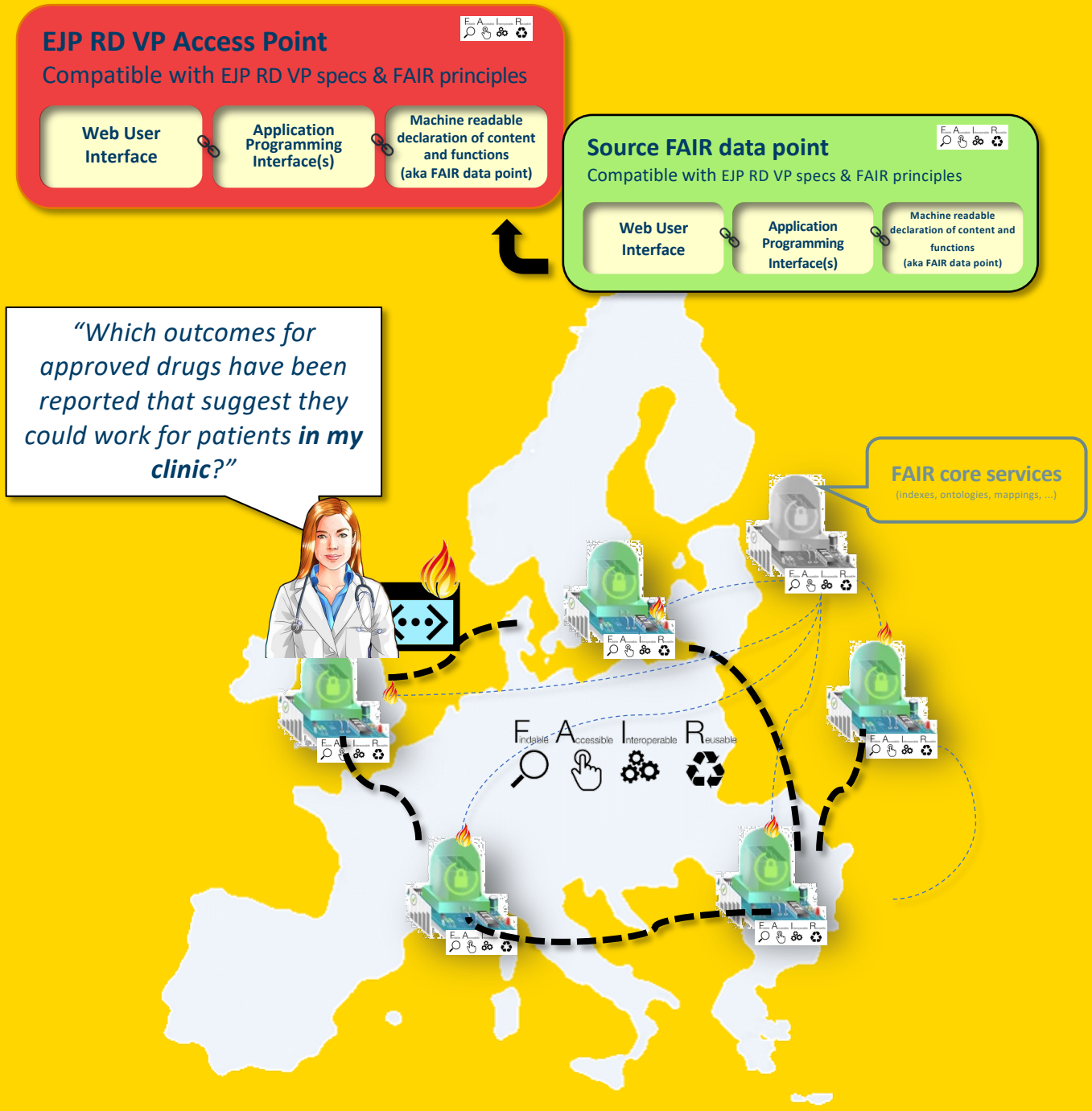
- Funding of **3** demonstration and **2** innovation **projects** to validate new innovative methodologies for RD clinical trials

Toolbox development

- EJP RD developed the **Innovation Management Toolbox** and **Clinical Trials Tool kit** supporting both pre-clinical translational and clinical research.

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

- **Mechanisms for continuous integration** of new data elements, standards and tools → provide **European research with a unique ecosystem** that will efficiently translate research into better care and medical innovation
- **Link healthcare and research** opening the road to diagnostics innovation thanks to the collaboration with the ERNs
- Develop **novel tools and pathways** for **comprehensive interpretation of data** and setting of standards



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

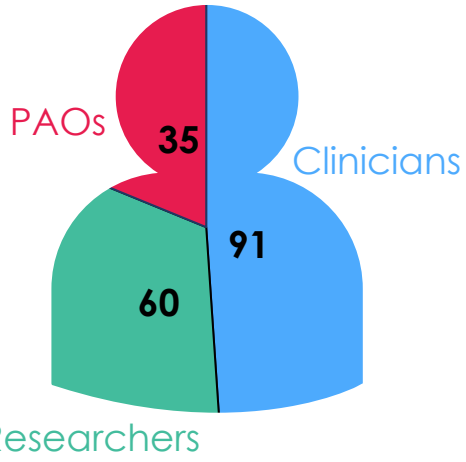
The **joint transnational call 2021** focused on funding of research related to **socio-economic, health care/health services, e-health and studies addressing the impact/burden of the delay in diagnosis and of the lack of therapeutic interventions**. This is the first time such type of multinational research is financed, with the aim of up taking the results to provide direct guidance/recommendations and impact healthcare systems and practices.

11.5 Mio€

12 funded projects

Accelerated share of knowledge & increased uptake of research results

Partners in the 23 NSS selected projects (Round 1 to 6)



ERNs dedicated trainings and fellowships

Funding of EJP RD ERN dedicated fellowships allows on exchange that contribute to bridging of healthcare and research by sharing of knowledge, joint research activities and clear benefit for each involved ERN :

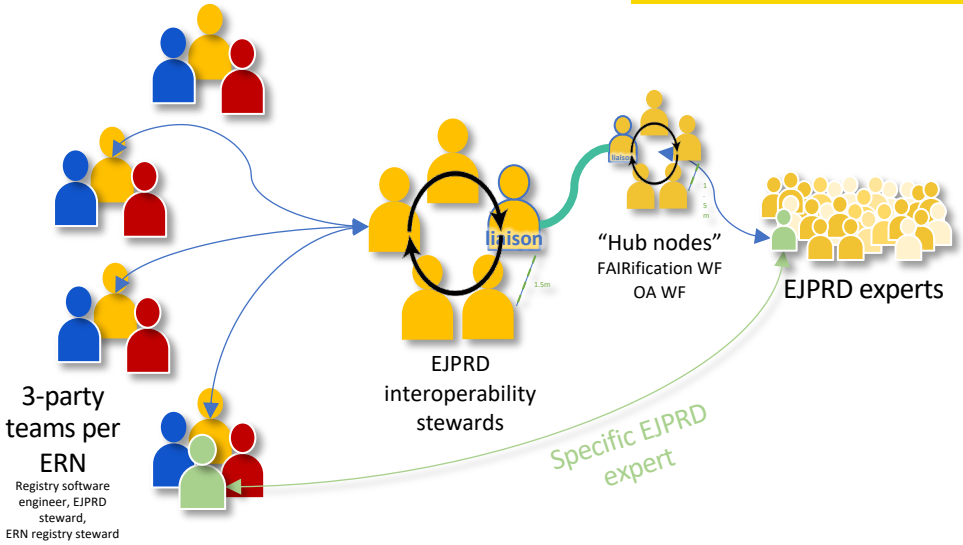
--> **33 Funded ERNs fellowships so far**

EJP RD

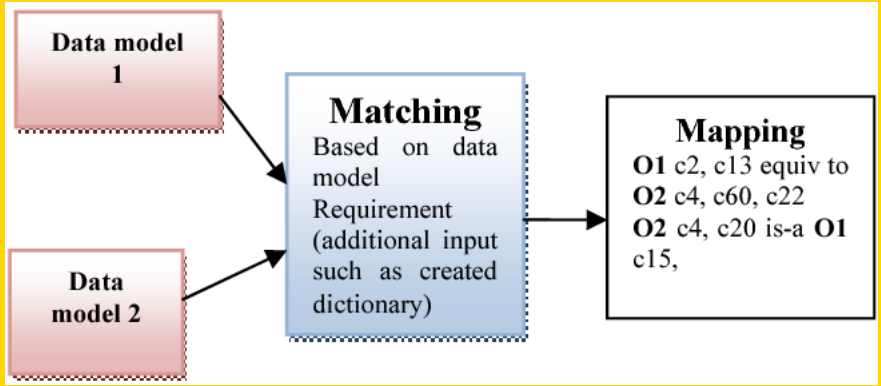
Specific impact 4: Improve healthcare systems' capacity to take up research results

Making ERN registries FAIR at the source to improve data sharing and re-use aiming to increase health data secondary use for research purposes

The FAIRification Stewardship Programme



77 FAIR standards and Tools mapped with 30 ERN registries



Data Model mapping

Data Models mapping efforts **including medical information standards as well as the data consent and access condition** representation and modelling is **paving the way to further integration of healthcare and research ecosystems** for a mutual take-up of data and findings.

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

- **EJP RD is recognized as major player** in the field of RDs by EU and international stakeholders

Synergy between EJP RD and ERICA **continues** for the collaborative and/or aligned Action. A joint workshop on "Finding rare disease registry data with the VP" was held in February 2022. **Frequent Alignment Actions** between EJP RD and ERICA has started. Timely mutual dissemination of Results.

EJPRD is contributing & providing PoC elements federated model, standards, ontologies building blocks for genome-phenome data federation for clinical research & healthcare

EJPRD collaborates with C4C to mutualise expertise for paediatric clinical trials, share guidelines & knowledge (e.g. training, clinical trials SOPs)

Interactions with TEHDaS were **through Orphanet** is involved; mainly in TEHDaS Work Package 6 that focuses on **data quality and standardisation within the context of the future European Health Data Space**. **Orphanet** was also one of the initiatives that were **interviewed as a producer and promoter of health data standards.** Additionally, Orphanet was able to **present a brief on the EJP RD VP** during the Gaia-x event

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

- **EJP RD is recognized as major player** in the field of RDs by EU and international stakeholders

EJPRD already engaged in the interaction with stakeholders involved in building the EHDS to contribute with its developments (VP) & support RD community

EUROPEAN MEDICINES AGENCY
SCIENCE MEDICINES HEALTH

Meetings with EMA representatives **to establish an interaction framework** and discuss key advances that could lead to **innovation in the field of statistical methodologies in small population clinical trials**.

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

EJPRD is actively contributing in the development and expansion of global standards for genomic data sharing

CRITICAL PATH INSTITUTE

The expertise of EJP RD in data modelling and standardization led to a **successful pilot to query transatlantic data** (through EJP RD metadata models, ontologies and standards) paving the way to interoperability between EJP RD and RDCA-DAP resources.

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

- **EJP RD is recognized as major player** in the field of RDs by EU and international stakeholders

- Medical Research Future Fund from Australia decided to join EJP RD **joint transnational call 2022** allowing on further expansion of RD knowledge and collaboration beyond the frontiers of Europe.

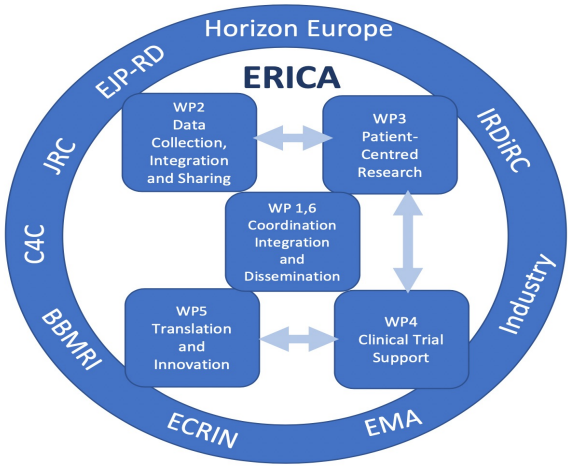
EJP RD Trainings are opened to international trainees :

- ERN workshops & ERN Fellowships

- E-Learning Academic Education
40% of participants/trainees of the EJP RD online education courses (MOOCs) are from outside of the EU demonstrating the added value of the EJP RD activities going beyond the EU borders.

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

EJP RD **Pillar 2 Partners (Virtual Platform)** are already leading global endeavours (Orphanet, HPO, FAIR data principles and metrics) or are deeply involved in global initiatives (IRDiRC, MME, Beacon, GA4GH)



>RD-CODE

European Reference Networks

ERN RWG Registries Taskforce

X-eHealth
Exchanging Electronic Health Records in a common framework

trainings

TEHDAS

EUROPEAN JOINT PROGRAMME RARE DISEASES

IRDiRC
INTERNATIONAL RARE DISEASES RESEARCH CONSORTIUM

PPRL
Consent

Rare Diseases

GO FAIR

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

EOSC-Life

B1MG Beyond one Million Genomes

CRITICAL PATH INSTITUTE

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

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 and Computable Consent

Clinical Research Network (CRN) Conference

IRDiRC- EJP RD-RE(ACT) Congress

Task Forces

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

Resource Integration

ODDG into WP19 Innovation Management Toolbox

Topic Identification

ELSI and WG3 feeding the JTC call on SHS

IRDiRC experts advising on possible topics in all EJP RD calls

IRDiRC Chairs as part of the EJP RD Policy Board

IRDiRC
INTERNATIONAL RARE DISEASES RESEARCH CONSORTIUM

THANK YOU

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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

EJPRD Pillar 3: training gaps identified and solutions proposed

Birutė Tumienė[1] & Roseline Favresse[2]

[1] Vilnius University Hospital Santaros Klinikos; [2]EURORDIS

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EUROPEAN JOINT PROGRAMME RARE DISEASES

EJP RD: Capacity building and empowerment framework in the RD ecosystem

RD research ecosystem:

- Universities;
- University hospitals;
- ERNs;
- Other research institutions;
- Research infrastructures;
- Patient organizations;
- Professional organizations…

The role of EJPRD:

- Providers;
- Contributors and collaborators;
- Ecosystem building;
- Advisors/ policy makers.

EJPRD, Pillar 3, Years 1-3:

- An **extensive and coherent programme** of education and trainings;
- Special provisions to increase **accessibility** for some **underserved groups and underrepresented countries**;
- **Patient-centredeness;**
- Progression of activities in Pillar 1, 2 and 4 → further **identification of educational needs.**
- The means **to share best practices** and collect extensive information for **continuous improvement.**

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

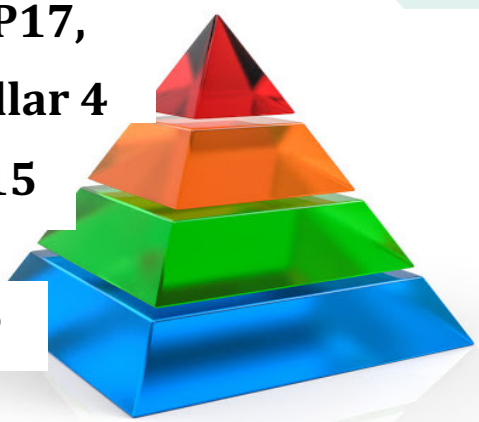
- Lack of published data or any other resources for the evaluation of the general RD research education and training.
- Focus on short-term (vs. long-term) impacts.
- Definition of "RD research education and training" vs. more general "RD education and training".

**More than 90 references, 28 EJPRD internal documents and multiple meetings/discussions/calls.*

Funded by the European Union
GA n°825575

EJP RD, Pillar 3, Years 1- 3

Principle of pyramid



- Experienced PI, leaders (highly – specialized and transversal knowledge and skills, know-how… ecosystem building)
- PhDs, postdocs;
- Multidisciplinary, multistakeholder community: clinicians, bioinformaticians, lab technicians, biobank and data managers, etc.
- Patients;
- Students (medical and other; homogenous, rigorous base of knowledge, direct teaching)

- RD (research) education and training **ecosystem is highly complex**: it involves many „learners“, „teachers“ and providers, and multiple forms of teaching and learning, including formal, non-formal and informal education.
- The **needs** of learners **are highly diverse** across the education and training continuum and follows a principle of a pyramid.
- **Many RD education and training resources** have been recently developed by the main RD organizations, ERNs, professional organizations and other stakeholders.

RD education and training resources

Type of resources	Knowledge, education and training resources
Knowledge bases, the main organizations for rare diseases	Orphanet; https://www.orpha.net/consor/cgi-bin/index.php
	OMIM*; https://www.omim.org/
	EURORDIS; https://www.eurordis.org/
	NORD; https://rarediseases.org/
	European Medicines Agency. Orphan designation: overview; https://www.ema.europa.eu/en/human-regulatory/overview/orphan-designation-overview
	IRDiRC; https://irdirc.org/
	GeneReviews; https://www.ncbi.nlm.nih.gov/books/NBK1116/
European Reference Networks	Information and links: https://www.orpha.net/consor/cgi-bin/Clinics_ERN.php?lng=EN; https://ec.europa.eu/health/ern_en Specific ERN websites, DG SANTE ERN Newsletter.
Professional organizations**	European Union of Medical Specialties (UEMS): Multidisciplinary Committee on Rare and Undiagnosed Diseases (MJC RUD); https://uems-genetics.org/links.html
	Society for the Study of Inborn Errors of Metabolism (SSIEM); https://www.ssiem.org/training
	Rare Cancers Europe; https://www.rarecancerseurope.org/events
	European Society of Human Genetics (ESHG)*; https://www.eshg.org/index.php?id=education
	International Society of Pediatric Oncology (SIOP); https://casehippo.com/spa/courses/catalog/siop/home
	European Society for Pediatric Nephrology (ESPN)*; https://www.espn-online.org/espn-ipna-erknet-educational-best-clinical-practice-webinars/#
	European Society of Endocrinology* https://www.ese-hormones.org/about-us/committees/rare-disease-committee/ and European Society of Pediatric Endocrinology* https://www.eurospe.org/education/
	International League Against Epilepsies*; https://www.ilae.org/education
	European Academy of Neurology*; https://www.ean.org/learn/joint-webinars
	European Respiratory Society*; https://www.ers-education.org/collections/educational-material-on-rare-diseases/
	European Society of Medical Oncology* (ESMO)
	European Hematology association; https://ehaweb.org/education/
	European Association for the Study of the Liver*; https://easlcampus.eu/ern-on-demand

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RD education and training resources

Education and training resources**	ESHG Genetic Educational Materials and Sources*; https://www.eurogems.org/index.html
	European School of Oncology*; https://www.eso.net/
	EURORDIS Open Academy; https://openacademy.eurordis.org/
	BBMRI.QM Academy*; https://www.bbmri-eric.eu/services/e-learning/
	Elixir Training Platform*; https://elixir-europe.org/platforms/training
	EATRIS Transmed Academy - course on translational medicine*; https://eatris.eu/services/education/*
	European Patients' Academy Webinars*; https://www.eupati.eu/category/webinar/
	European Patients' Academy Expert Training Course*; https://www.eupati.eu/eupati-training-course/
	Integrated DEsign and AnaLysis of clinical trials in small population group (IDeAl) resources; https://www.ideal.rwth-aachen.de/?page_id=1732
	Research Data Management online courses*; https://vidensportal.deic.dk/en/RDMELearn
	Patient-Centered Outcomes Research Institute (PCORI) Training: A Program for Rare Disease Patient Advocates; https://www.pcori.org/research-results/2015/pcor-training-program-rare-disease-patient-advocates
	Findacure's e-learning resources on rare diseases; https://portal.findacure.org.uk/
	FutureLearn courses on genomics*: • The Genomics Era: the Future of Genetics in Medicine https://www.futurelearn.com/courses/the-genomics-era; • Whole Genome Sequencing: Decoding the Language of Life and Health; https://www.futurelearn.com/courses/whole-genome-sequencing; • Genomic Technologies in Clinical Diagnostics: Next Generation Sequencing; https://www.futurelearn.com/courses/next-generation-sequencing; • Genomic Technologies in Clinical Diagnostics: Molecular Techniques; https://www.futurelearn.com/courses/molecular-techniques.
	Genetics education for primary care resources from the Gen-Equip project* https://www.primarycaregenetics.org/?page_id=109&lang=en.
	Medics4RareDiseases (M4RD) video library; https://www.m4rd.org/video-library/.
	Program on rare diseases "Excellence In pediatrics"; https://www.ineip.org/p2p_education_program_on_rare_diseases_excellence_in_pediatrics.
	Recordati rare diseases; https://www.rrd-foundation.org/en/courses
	Aarhus University, Rare Diseases in Translational and Personalized Medicine; https://kursuskatalog.au.dk/en/course/105020/Rare-Diseases-in-Translational-and-Personalized-Medicine.
	Wellcome Advanced Courses and Scientific Conferences – Genomics of Rare Disease; https://genetics.org.uk/events/wellcome-advanced-courses-and-scientific-conferences-genomics-of-rare-disease/.
	Queen's University Belfast; https://www.qub.ac.uk/sites/RareDisease/Events/.

*Resources that are not specific for rare diseases but include important aspects of rare diseases.
**Some of these resources may have limited duration or accessibility.

Funded by the European Union GA n°825575

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

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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

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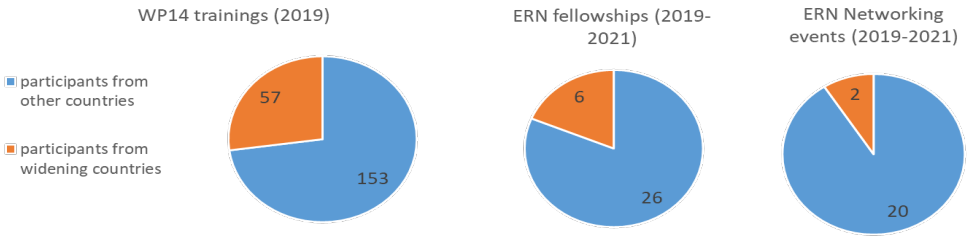
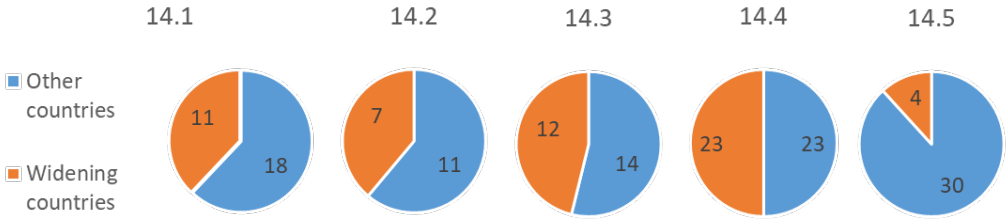
EJPRD: participation of widening countries

Empowerment and capacity building:

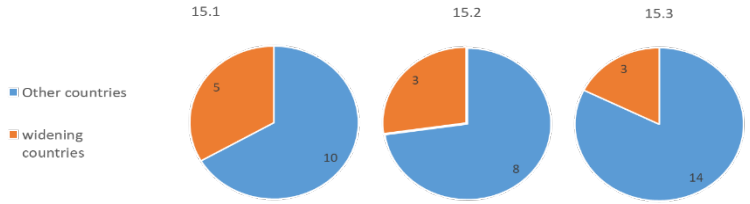
Participants from widening countries comprised:

- 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.

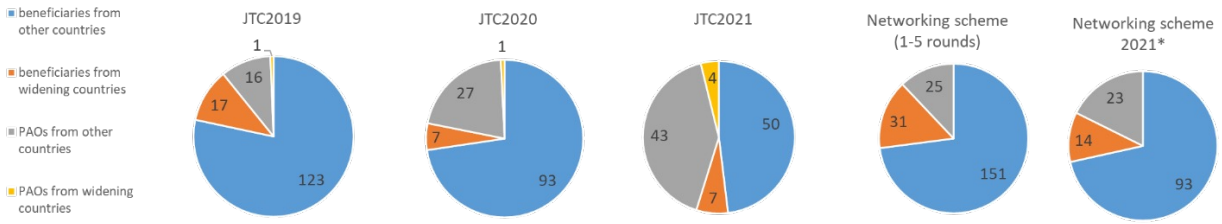


Rare disease research funding:

- **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.**



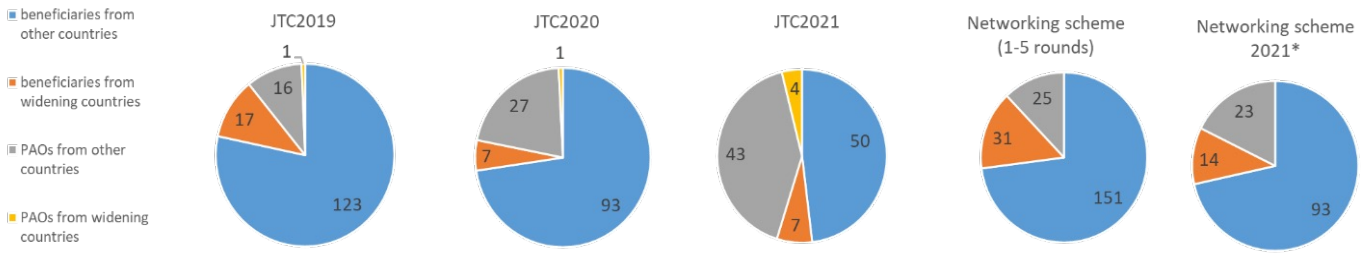
EJPRD: measures to increase participation of widening countries

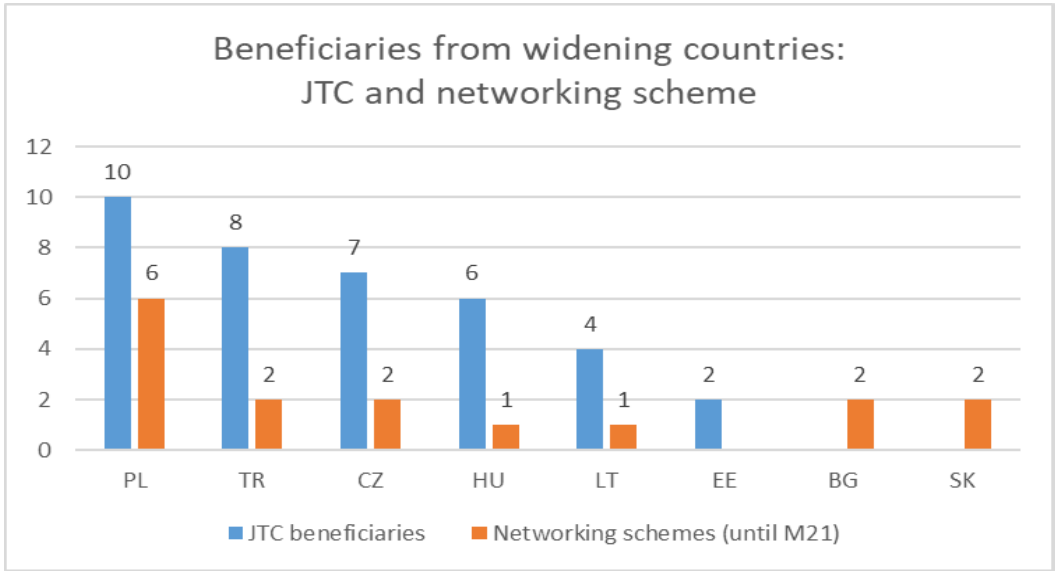
Number of JTC **funding organizations** from widening countries increased *from **1** in E-RARE-1 to **7** in EJPRD*.

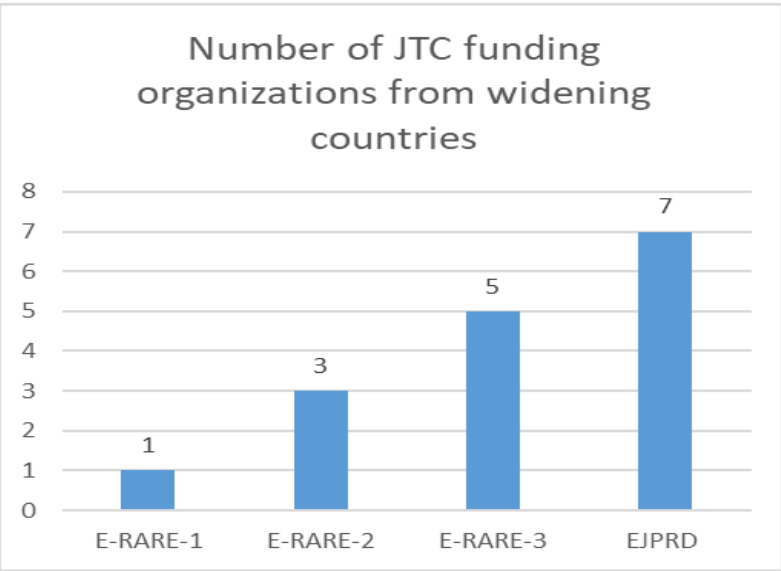
- **Beneficiaries** from widening countries comprised *from **7 to 12%** in EJPRD JTCs 2019 to 2021*.

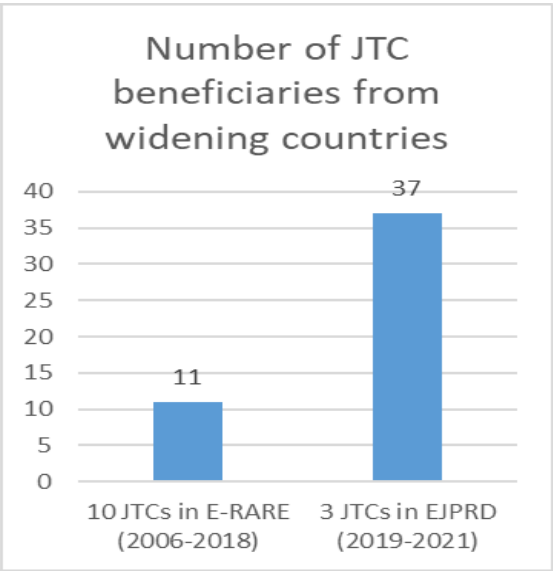
On average, 47% of EJPRD JTC consortia involved at least one partner from widening countries.

- Overall, beneficiaries from widening countries received ***4,1% to 8,7%** of funding* in JTC2019 and JTC2020.
- The average financial contributions per beneficiary were ***30% to 41%** lower* for beneficiaries from widening countries.
- However, widening principles may result in a win-win situation for all consortium participants: the average sum of the total requested funding was ***8% to 18% higher** in projects that involve participants from widening countries*.









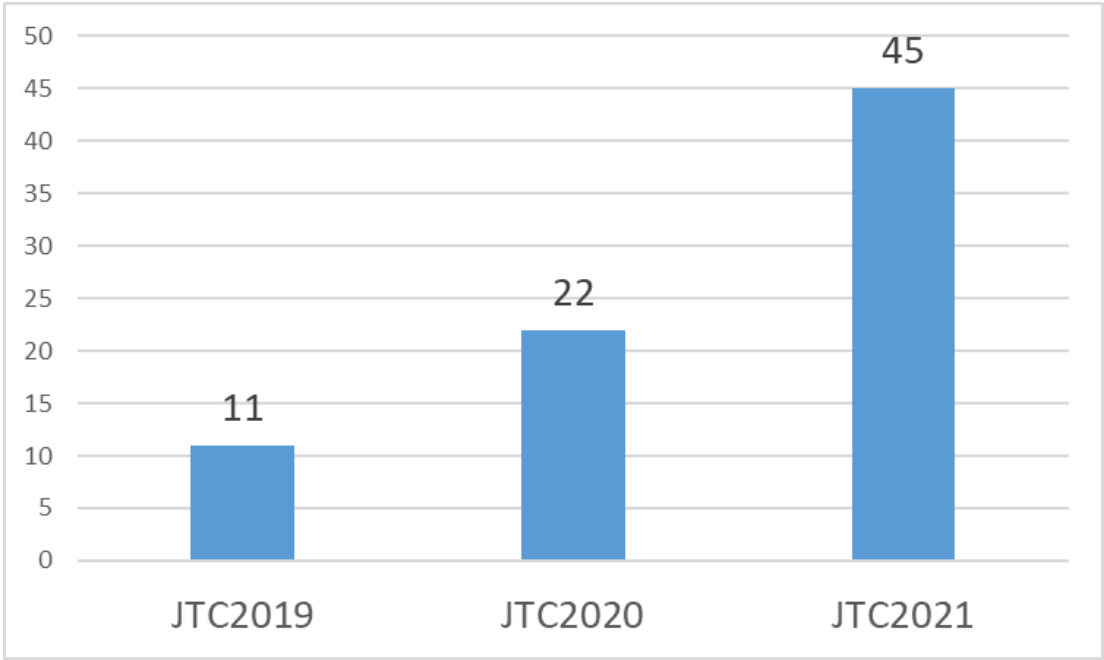
EJPRD: measures for patient-centeredness in RD research

Measures:

- **Trainings for patient representatives and advocates**.
- Trainings for **mixed audiences** of patients and researchers.
- **Short Guide on Patient Partnerships in Rare Disease Research Projects**.

Outcomes:

- A considerable growth of participation of PAOs in the JTCs: from 17 of 157 (**11%**) beneficiaries in JTC2019 to 28 of 128 (**22%**) in JTC2020 and to 47 of 104 (**45%**) in JTC2021; 92 patient organizations have been included into EJPRD JTCs over the three years.
- Financial contributions allocated to PAO were also growing from **0.9%** of all financial contributions allocated to JTC beneficiaries in JTC2019 to **2%** of all financial contributions allocated in JTC2020.



National alignment of RD education and training

- 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.
- a survey of ERN members [EJPRD D17.1]: only 51 of 291 (17.5%) respondents indicated that there is a specific research skills and support training available in their countries.
- the vast majority of identified educational activities are based in the EU-15 countries.
- a survey of national authorities [EJPRD D2.23]: 78% of interrogated national authorities (from AM, BG, HR, CZ, FR, GE, DE, IE, IT, LT, LU, PL, PT, RO, RS, ES, TK, UK) answered “yes” to the question “Does the NP/NS for RD promote and/or support training activities?” However, after analyzing available RD National Plans and Strategies (eighteen RD RD NP/NS, available in either English, Polish or Lithuanian, BG, CZ, HU, LT, PL, RO, SI, SK, AT, BE, DE, ES, FI, FR, IE, IT, NL, UK), it seems that support is mostly non-financial, through the endorsement of universities and university / teaching hospitals.

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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.

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Impact of EJPRD in the landscape of RD research education and training

- 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).

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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

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EJP RD update on the Annual Work Plan for year 5

EUROPEAN JOINT PROGRAMME RARE DISEASES

Policy Board & Governing Board Meeting
06/07/2022 _ Online

COORDINATION, TRANSVERSAL ACTIVITIES & COMMUNICATION

EUROPEAN JOINT PROGRAMME
RARE DISEASES

What's new in Pillar 0 in AWP Y5

- **WP1: Coordination & Management**
  - Continue Coordination activities (organisation of all governing Boards meeting, delivery of the Annual Progress Report for Year 4, monitoring, etc.)
  - Preparation of RD Partnership (in close connection with activities of WP2, national bodies, in particular through the support development of National Mirror Groups)
  - Implementation of IRDiRC Roadmap 2023

- **WP2: Strategy**
  - Overall support for EJP RD strategy:
    - To align with national strategies
    - To facilitate prioritisation
  - Organisation of the second strategic workshop with national policy makers

- **WP3: Sustainability**
  - Update of the catalogue of EJP RD "services" and Sustainability Handbook
  - Finalisation of the business plan for each of the EJP RD outputs & for EJP RD as a whole (in close collaboration with WP1, WP2 & WP4)

EJP RD

What's new in Pillar 0 in AWP Y5

- **WP4: Ethical, regulatory, legal and IPR support**
  - Continue to support all EJP RD partners (ethics monitoring or evaluation of funded projects, support on demand from WPs/pillars, continuous information on ethics/regulatory/legal updates)
  - Continue the work in connection with WP3 on identified IP needs

- **WP5: Communication & Dissemination**
  - Develop and apply the communication strategy for the RE(ACT) Congress and IRDiRC Conference 2023
  - Enhance the IRDiRC and EJP RD communication strategy to better disseminate the output of the work done
  - Showcase through communication actions the impact of EJP RD through the years

FUNDING OPPORTUNITIES

EUROPEAN JOINT PROGRAMME
RARE DISEASES

What's new in Pillar 1 in AWP Y5

- Implementing the fifth joint transnational call (JTC 2023) for projects on rare diseases and connecting funded projects with activities and services in Pillars 2-4 (WP6);

- Further reviewing of guidelines for patient partnerships by the Working group for patient engagement in research (PENREP) and increasing visibility of its short guide towards future applicants of JTC 2023 as well as more generally in the RD community (WP6);

- Further implementing the Networking Support Scheme for sharing of knowledge on rare diseases and rare cancers between relevant stakeholders through fostering the organization of workshops and/or conferences (WP7);

What's new in Pillar 1 in AWP Y5

- Monitoring the implementation of first phase projects of the Rare Disease Research Challenges until their mid-term evaluation; after assessment of the milestones/deliverables distribute funding to successful projects for second phase (WP8);

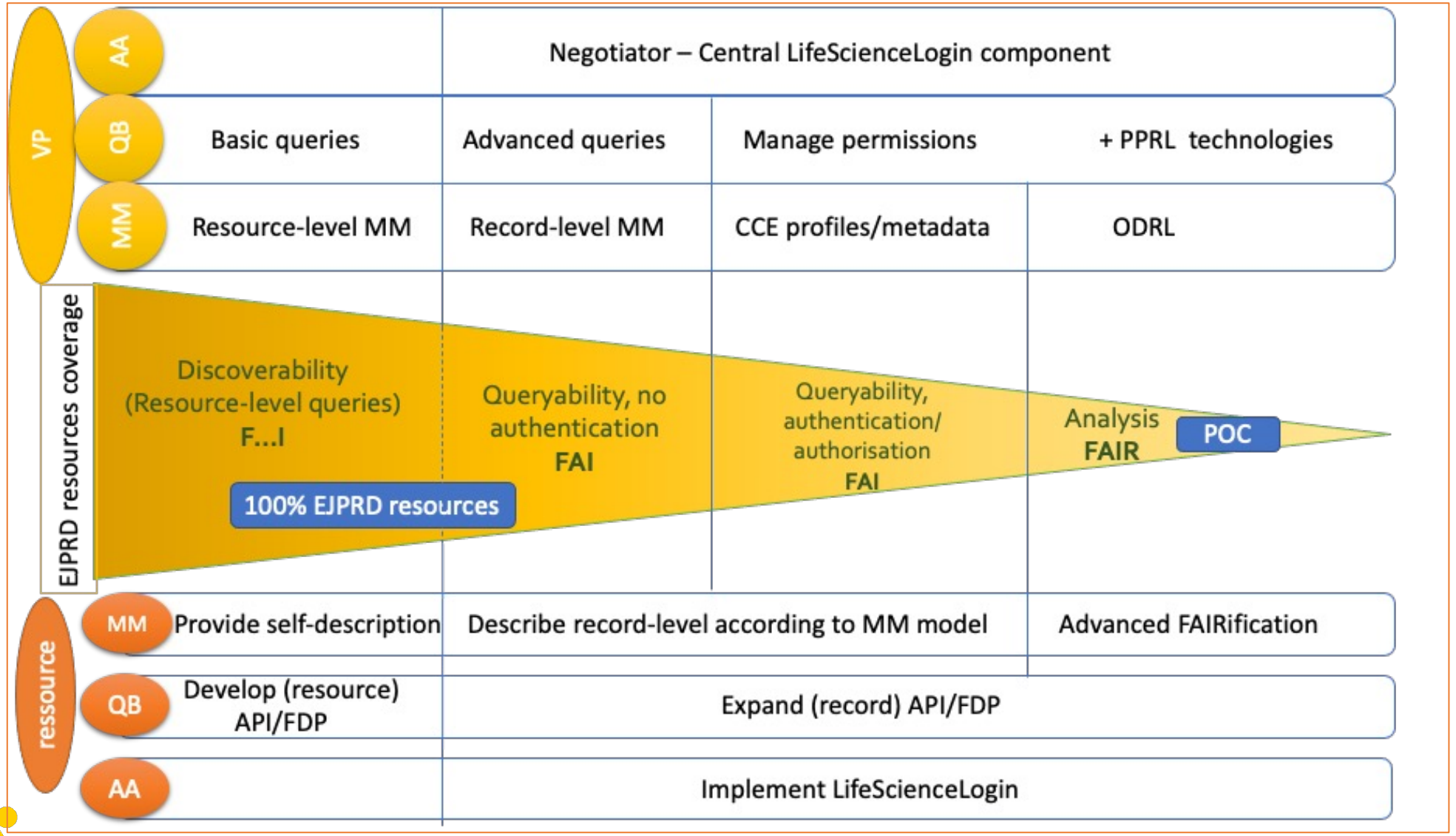
- Monitoring all selected projects in the different funding schemes in Pillar 1 (WP6-WP8) and in ERA-Net E-Rare to measure the success of these funding schemes and support applicants in further developing their projects for translation and dissemination (WP9).

EJP RD

VIRTUAL PLATFORM OF DATA, TOOLS & RESOURCES

EUROPEAN JOINT PROGRAMME
RARE DISEASES

What's new in Pillar 2 in AWP Y5



AA: Authentication & Authorisation;

API; Application Programming Interface;

CCE: Common Consent Elements;

FAIR: Findable, Accessible, Interoperable, Reusable (for machines & humans)

FDP: FAIR Data Point

MM: Metadata Model;

ODRL: Open Digital Rights Language;

POC: Proof of Concept;

PPRL: Privacy Preserving Record Linkage;

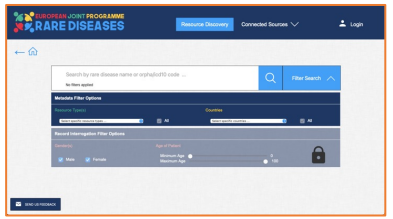
QB: Query Builder

VP: Virtual Platform

EJP RD

What's new in Pillar 2 in AWP Y5

- **VP as a network** supported by centralized minimum set of facilities
  - Methodology document onboarding resources in the VP
  - Sustainability, quality, GDPR metadata assessed and modelled.
- **VP as a central portal (EJP RD) https://vp.ejprarediseases.org/**
  - User interface design and development
  - Increase query facilities
  - Every 6-mo version releases from December 2022



- **VP Authentication and Access Control**
  - Common digital representations of use conditions towards machine readability
  - Implement LifeScience-AAI
- **Federated analysis POC (new Work Focus)**
  - Using system biology FAIR datasets and other heterogeneous data sources
- **Engaging further by dissemination and training**
  - 2 trainings planned
  - "EJPRD VP connected resource" label
  - Publish visually enhanced users' "Flash cards"
- **Research and ecosystem integration readiness**
  - Cloud facility for data analysis
  - Metadata model transformation for interoperability with health data and data spaces

EJP RD

TRAINING & EDUCATION

EUROPEAN JOINT PROGRAMME
RARE DISEASES

What's new in Pillar 3 in AWP Y5

- **Training on data management & quality (WP14)**
  - 2 new national trainings on the *Orphanet nomenclature and RD ontologies*
  - 5th edition of the training course on '*Standards and quality of genetics/genomics data in laboratory and clinical research practice*' in Warsaw, PL in Oct. 2023
  - 5th edition of the international course *'Training on Strategies to foster solutions of undiagnosed rare disease cases'* in Roma, IT in April 2023
  - 2 training workshops on *RD Biobanks* in Spring 2023 and Autumn 2023
  - 5th edition of the 5-day face-to-face training course *'International Summer School on RD registries and FAIRification of data'* in Sept. 2023 in Roma, IT

- **Capacity building and training of patients and researchers in RD Research (WP15)**
  - *EURORDIS Winter and Summer Schools* to be hosted the same week in Barcelona, SP to enhance interactions between faculty members and learners and maximize the learning experience
  - One face-to-face *Leadership School* to be held in Gdansk (PL) with a double capacity of learners
  - One new face-to-face training on RD research for *15 young patients* will be held (12-18 y.o.)

EJP RD

What's new in Pillar 3 in AWP Y5

- **Online academic education course (WP16)**
  - **2 new MOOCs delivery** planned in 2023:
    - *MOOC#4 - Statistical methodologies for clinical trials*
    - *MOOC#5 - Data and Rare diseases: ethical and regulatory considerations*
  - **Continuous opening of the 3 available MOOCs**
    - *MOOC#1 - Diagnosing RD: from the Clinics to Research and back*
    - *MOOC#2 - Innovative therapies and personalized medicine: new keys for the treatment of rare diseases - launch Q4 2022*
    - *MOOC#3 - Rare Diseases: translating research into health improvements» - launch Q4 2022)*

- **ERN RD training and support programme (WP17)**
  - Delivery of all remaining ERN training programmes already selected and funded
  - Accreditation process follow up for training activities

- **Continuous improvement and adaptation of training activities (WP18)**
  - Finalization and provision of two new courses on EJP RD Virtual Platform, targeted at the VP contributors and users (Q1 2023 and Q2-3 2023)
  - Final analysis of the Pillar 3 education and training activities and recommendations for the further actions.

EJP RD


ACCELERATED TRANSLATION OF
RESEARCH RESULTS & CLINICAL TRIALS
EUROPEAN JOINT PROGRAMME
RARE DISEASES

What's new in Pillar 4 in AWP Y5

WP19:

Final year push to **consolidate, strengthen and embed innovation support services in the community**:

- **Continue mentoring programme**, onboard successful JTC 2022s, **recruit new projects and mentors.** Sustainability model to be socialised with gatekeepers;
- **Explore with national funders/research communities expansion of mentoring programme** into national contexts;
- Big push to **ensure wide use of Innovation Management Toolbox**; **maintain and expand content and curation**;
- Work with innovation funding roadmap output to engage relevant stakeholders, including EFPIA, Moonshot signatories, national funders, charities, philanthropy for **long term value chain support for RD innovation**

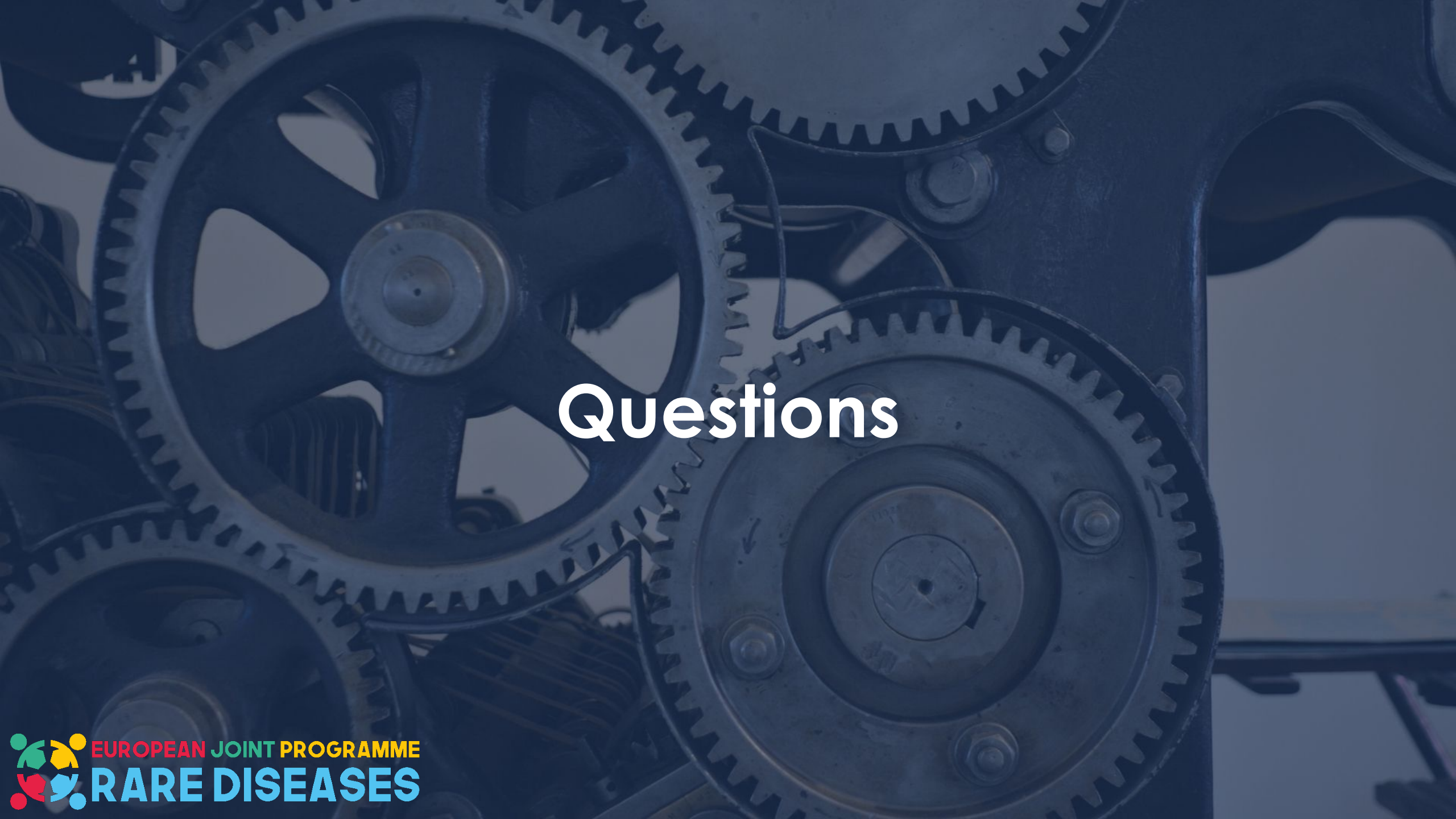
EJP RD

What's new in Pillar 4 in AWP Y5

WP20:

- Provide additional advanced courses and develop intermediate courses **on Clinical Trials Methodologies** adapted to rare diseases clinical trials stakeholders
- **Show first results** on demonstration and innovation projects, supervise their progression and **boost the collaboration between expert methodologists**
- Intensify the **Collaboration with EMA** with regard to transversal activities
- Disseminate the work of WP20 in **meetings, conferences, and scientific publications**
- Foster the **Clinical Studies Support Office** work by **targeted dissemination activities**, and appropriately **refining its assistance** to ensure the best quality assistance to Rare Diseases Clinical Trials applicants by addressing their different queries on unmet needs in this field
- **Further validate and provide tools required by clinicians for RDs Clinical Trials** including best practice documents, recommendations, and guidelines (**Clinical Trials toolkit**)

EJP RD



Questions

EUROPEAN JOINT PROGRAMME
RARE DISEASES

Feedback on the AWP Y5

- Taking into account your overall knowledge of EJP RD and AWP Y5: what is missing in AWP Y5?
- How can we still better integrate EU-13 countries ?
- How do you present and get back to your national stakeholders with the key points of the EJP RD AWP Y5 ?
- How to make the research resources and data sources more visible for researchers in your country ?

Rare Diseases Partnership

EUROPEAN JOINT PROGRAMME
RARE DISEASES

Policy Board & Governing Board Meeting
06/07/2022 _ Online

Vision

To leave no one behind, deliver a RD ecosystem that builds on the successes of previous programmes by:

- supporting robust patient need-led research,
- developing new therapies, diagnostic methods and pathways,
- spearheading the digital transformational change connecting the dots between care, patient data and research while
  - ensuring individual control over the use of personal data
  - and strong alignment of strategies in RD research across countries and regions.
- Finally structuring goal-oriented public-private collaborations targeted at interventions all along the R&D value chain

ensure that **the journey from knowledge to patient impact is expedited**, thereby optimizing EU innovation potential in RD.

EJP RD

Mission (to be accomplished by 2031)

- **Bring** to bear the high value **supporting services from across Europe under one roof** so that every high-quality RD research project will benefit from
  - cross-disciplinary expertise,
  - goal-oriented study planning
  - and efficient execution.
- Enable **every consenting patient living with a rare disease to be findable and enrolled in a suitable clinical study** that is necessary for generating advances in diagnosis, understanding of diseases, having regulatory-compliant data sets and developing treatments.
- Make **Europe a global leader in rare disease research** through
  - providing a suitable infrastructural and regulatory environment
  - as well as significant increase in investment

to spur innovation, leading to job creation and optimizing EU competitiveness in R&D and healthcare innovation.

EJP RD

VISION

Improve the health and well-being of people living with a rare disease by making Europe a world leader in innovation to address the unmet needs of 30 million persons living with a rare disease in Europe, thus supporting the EU commitment to UN 2030 Agenda's Sustainable Development Goals

UN 2030 AGENDA'S SUSTAINABLE DEVELOMENT GOALS

- SDG3 GOOD HEALTH AND WELL BEING
- SDG9 INDUSTY, INNOVATION AND INFRASTRUCTURE
- SDG10 REDUCED INEQUALITIES
- SDG17 PARTNERSHIPS FOR THE GOALS

GENERAL OBJECTIVES (Impact)

SPECIFIC OBJECTIVES (outcomes)

OPERATIONAL OBJECTIVES (Resources & actions)

GO1 Tackling diseases and reducing diseases burden

- SO1 Patient-need led relevant research enabled by outcome-oriented investments strategically deployed along the R&D value chain
- SO2 Better understanding of RD burden and impact assessment of interventions

GO2 Unlocking the full potential of new tools, technologies and digital solutions

- SO3 Interoperable, federated, evolving and scalable RD infrastructure of data, samples, resources and tools with necessary critical mass for meaningful RD research & innovation
- SO4 Active utilization in all Member States & Associated countries by all stakeholders of high-value, ethically and regulatory compliant data tools and services tailored to needs of RD research community

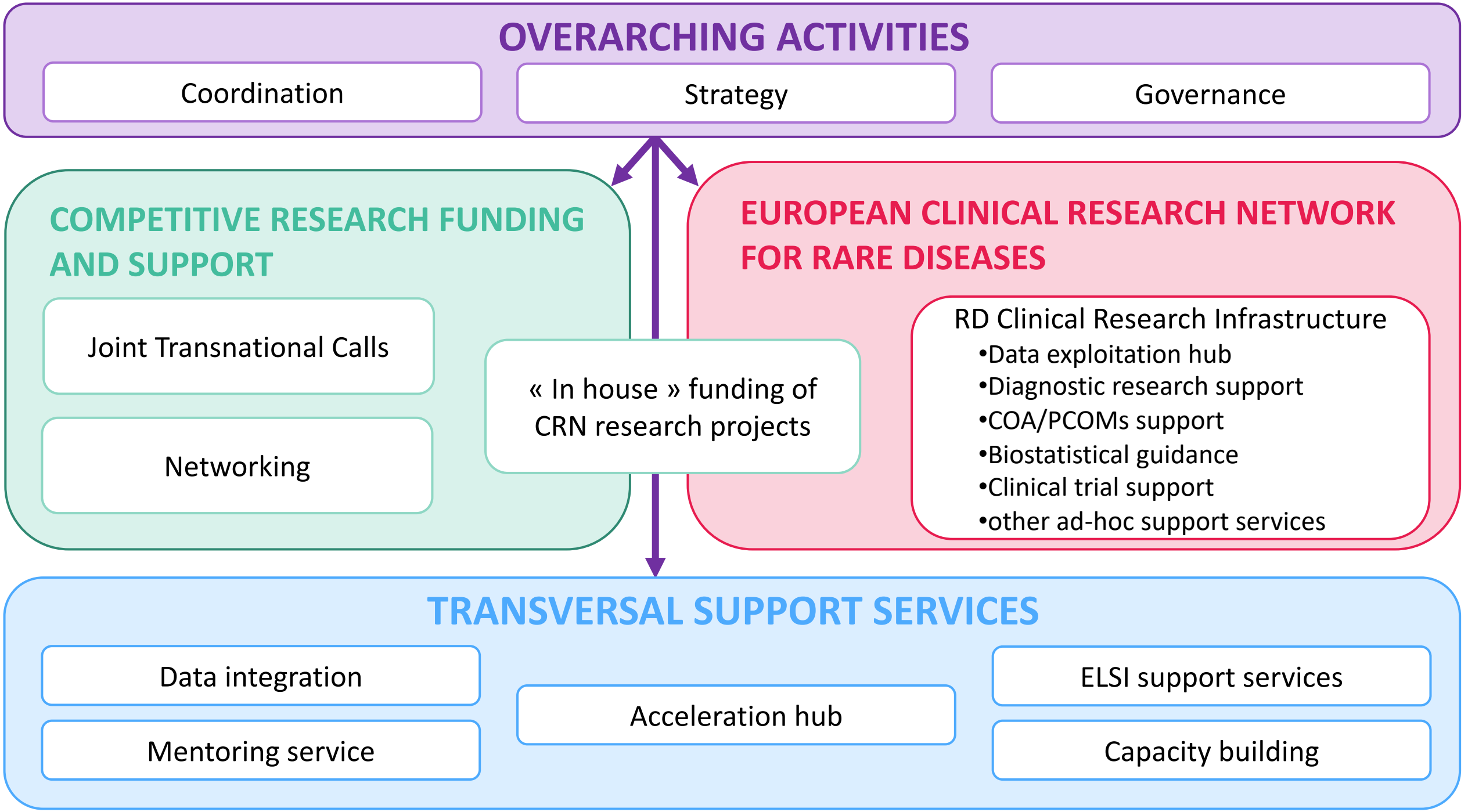
GO3 Timely, equitable access to innovative, sustainable and high-quality healthcare by virtue of a highly integrated research and healthcare system

- SO5 Decreased number of undiagnosed rare diseases and unsolvable cases as well as reduction in the duration of the diagnostic odyssey
- SO6 Increased capacity of RD stakeholders across Europe through quality training and skills development
- SO7 Reduced failure rates of therapeutic development

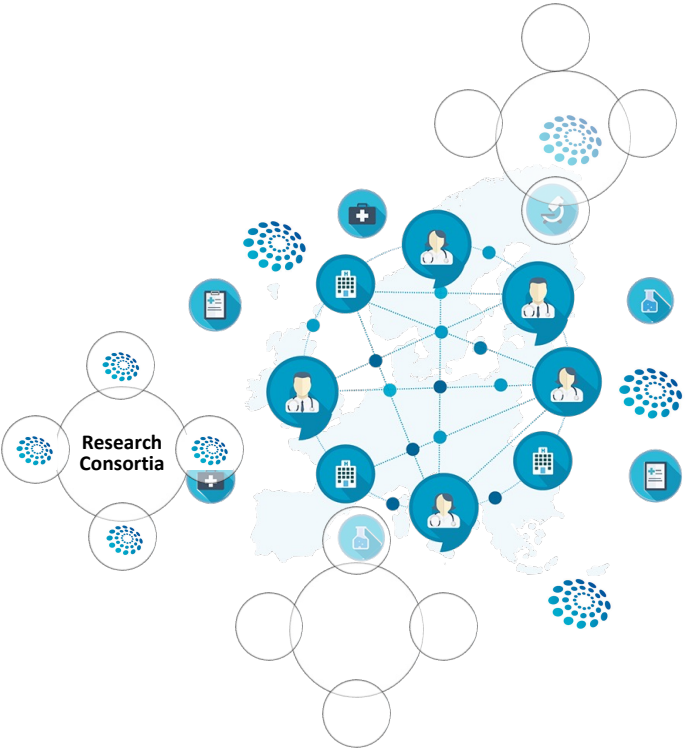
GO4 More effective outcomes from public-public, civil society and public-private collaborations

- SO8 Meaningful empowerment, engagement, and leadership - as equal partners - of people living with a rare disease
- SO9 Effective alignment of national RD strategies with EU objectives, leveraging shared resources and maximizing MS and Associated countries contributions
- SO10 Effective transcontinental collaboration

- OO1 **Funding and support to high quality, interdisciplinary, collaborative research**
- OO2 **Multistakeholder and cross sectoral collaborations**
- OO3 **Robust data, resources and expertise infrastructure**
- OO4 **Capacity building and empowerment**
- OO5 **Strategy alignment and coordination**

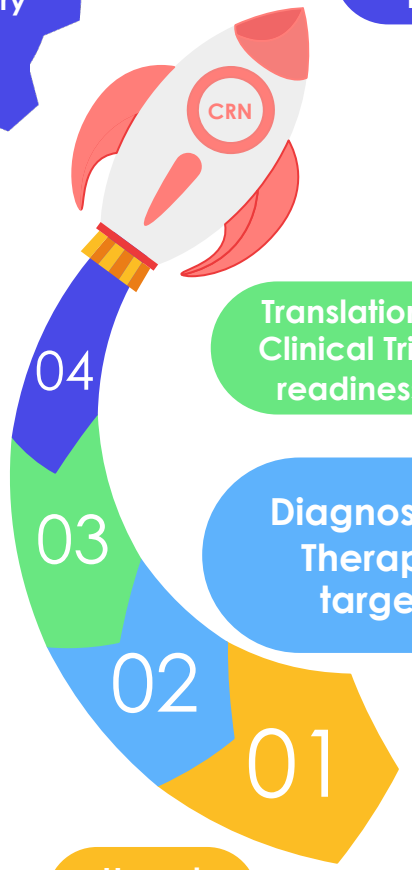

OVERARCHING ACTIVITIES
Coordination
Strategy
Governance
COMPETITIVE RESEARCH FUNDING AND SUPPORT
Joint Transnational Calls
Networking
« In house » funding of CRN research projects
EUROPEAN CLINICAL RESEARCH NETWORK FOR RARE DISEASES
RD Clinical Research Infrastructure
•Data exploitation hub
•Diagnostic research support
•COA/PCOMs support
•Biostatistical guidance
•Clinical trial support
•other ad-hoc support services
TRANSVERSAL SUPPORT SERVICES
Data integration
Mentoring service
Acceleration hub
ELSI support services
Capacity building

Clinical Research Network



synergy
COA/
PCOM
synergy
SYNERGY
with
complementary
structures/
process
Diagnostic
research
support
Biostatistical
Guidance
synergy
INFRASTRUCTURE
synergy
Data
Exploitation
Hub
Clinical
Trial
support
synergy
Ad-Hoc
support
synergy

Impactful results



Translation/
Clinical Trial
readiness

Diagnostic/
Therapy
target

Unmet
need

In House research

- RWE generation for RD outcomes
- Identification & validation of RD-specific PCOMs
- Discovery & validation of biomarkers and surrogate endpoints
- Innovative diagnostic research approaches
- Clinical Trial evidence generation

Project proposals

Projects selected

National Meetings Fostering Commitment

EUROPEAN JOINT PROGRAMME
RARE DISEASES

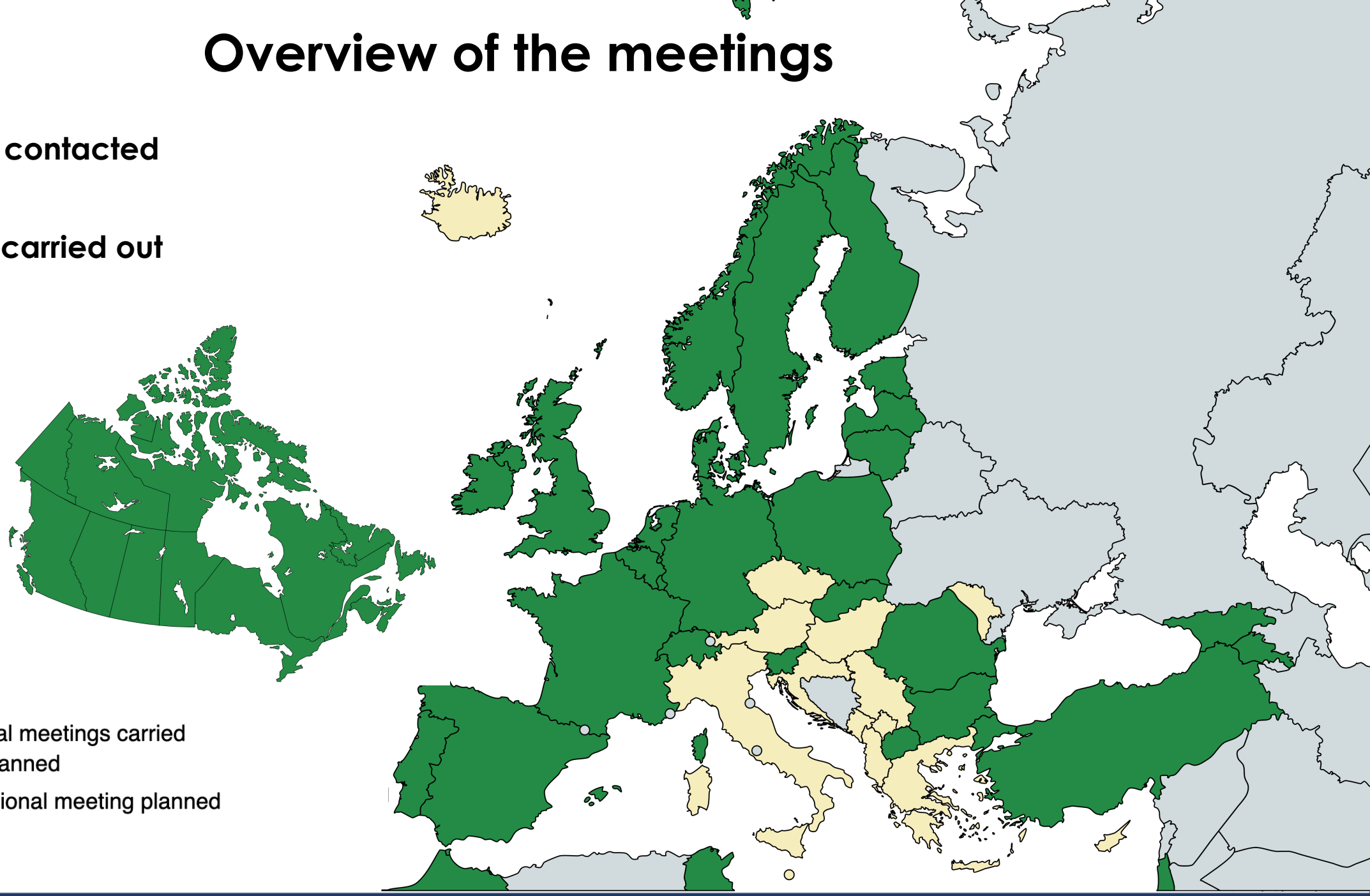
Agenda of the national meetings – Commitment for the RDP

- Rare Disease Partnership – Short Presentation
- In-Kind / In-Cash contributions - information
- Letter of commitment – Presentation of what is expected
- Identification of strengths – [Country] State of Art
- Exchange on the possible contribution, needs, institutions to be involved etc.

EJP RD

Overview of the meetings

- 44 countries contacted
- 30 Meetings carried out



Strategic Research & Innovation Agenda (SRIA) development

EUROPEAN JOINT PROGRAMME
RARE DISEASES

Objectives _ SRIA TF

1. Agree on the **structure of the SRIA**
   - (**level of granularity** with which different topics will be addressed);
2. 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.)
3. **Help in coordinating the work of the whole process**,
   - and notably of the working groups if created.

Members representativeness _ SRIA TF

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

- **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).

Composition of the SRIA Task Force

14 'Funders', including 6 representative of **funding agencies** + **8 experts nominated by Ministries**

2 IRDiRC representatives

2 representatives of **Patients organisations**

3 Experts for **ERNs** (+2 ERNs coordinators in the experts below)

11 Experts representing **relevant EU-funded projects or initiatives**

3 Experts on **Data**

2 Experts for **relevant Research infrastructures**

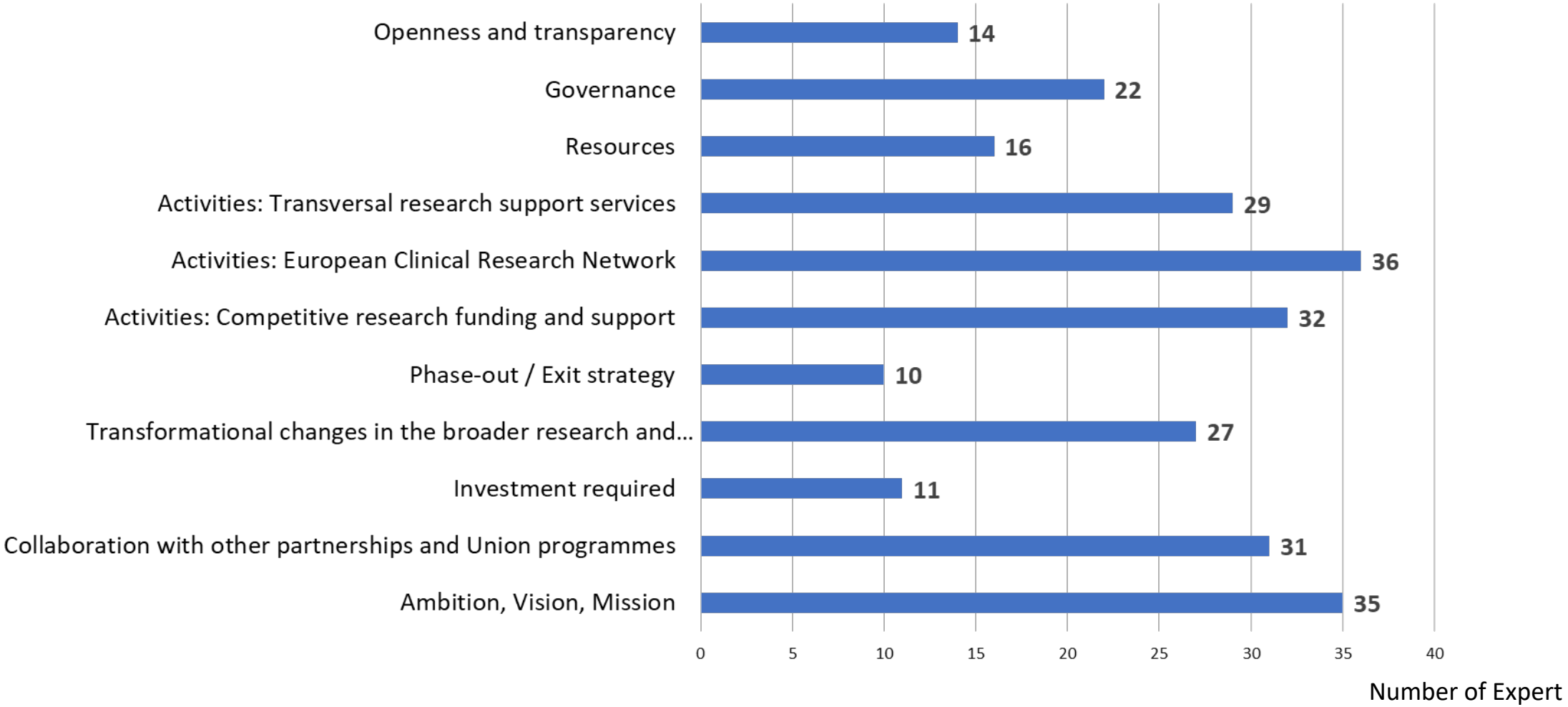
4 representatives of **Foundations**

2 representatives from **Other organisations** (Industry/Private)

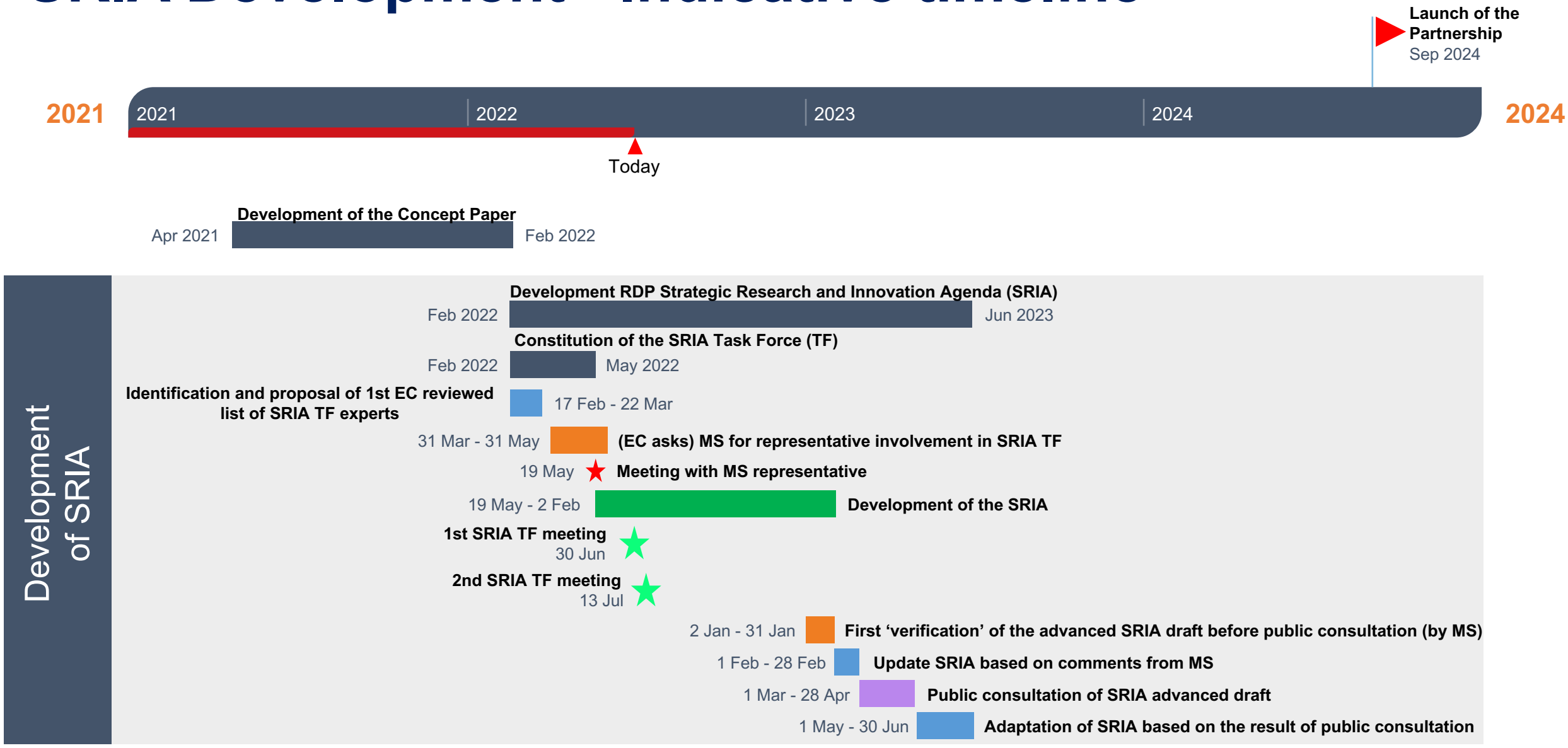
10 representatives of **European Commission and agencies (EMA, EUnetHTA, etc.)**

EJP RD

Distribution of the expertise in TF SRIA RDP



SRIA Development - Indicative timeline



This timeline is subject to change

Proposal for SRIA development process

(according to the Concept paper)

Preparatory/Initial phase

- Set-up a dedicated expert team to guide and oversee the development and implementation of the SIRA
- Identify the key stakeholders to include in the SRIA framework development process

Writing & Consultation process

- Initial reflection and analysis of gaps and opportunities
- Early ideas about scope, impact, outputs should be consulted by the core partners with several test audience
- Preliminary draft should be open for public consultation
- Above-mentioned results should be used for the SRIA revision
- Draft will continue to evolve as more information about the scope of other EU programmes become available

Validation phase

- Final SRIA will be submitted for approval of the General Assembly and EC (before signature of the Grant Agreement)
- SRIA will be reviewed at the occasion of the Partnership interim evaluation

EJP RD

Rare Diseases Partnership Feedback

Question Asked

- Indicate any feedback that would serve for the **development** of the **next phases of the Rare Diseases Partnership** (e.g., the development of the Strategic Research and Innovation Agenda)

- Indicate any **national activity or development** that would be **related to the Rare Diseases Partnership**

MIRO

https://miro.com/app/board/uXjVOnjU8yE=/?share_link_id=617003062087

Sustainability
Roadmap for EJP RD

WP3

Policy Board & Governing Board Meeting
06/07/2022 _ Online

EUROPEAN JOINT PROGRAMME
RARE DISEASES

Processe summary

Identification

- First survey on sustainability (Work package and Task leaders)
- Categorisation of element by relevance and type (resource, activity, output,service)

Development

- Analysis of first survey
- Follow-up interviews
- Feedback
- Second survey on sustainability (stakeholders & business model focused)

Refinement

- Business Model Canvas: Exercise & Analysis
- Validaton of value propositions

EJP RD catalogue

PILLAR	ELEMENT/ASSET	TYPE	VALUE-CHARACTERISATION (SUMMARISED)
0	**Coordination services EJP RD & IRDiRC SciSec**	Service, Resource	Proactive, adaptable and efficient planning, execution, monitoring and issue solving to optimise quality results, fostering links with other initiatives for efficiency and synergistic actions.
0	**Central Helpdesk**	Service, Resource	Provision of centralised easy access to RD expertise, with request processes and tailored services for users, outreaching through dissemination strategies to increase visibility of EJP RD services.
0	**Sustainability Handbook**	Output	The general guidance on sustainability key factors helps the RD Community and other areas to plan and operate in a sustainable manner.
0	**Advisory Regulatory Ethics Board (AREB ELSI Services)**	Service, Resource	Reference single contact point, easily identified, for ELSI questions, with the provision of quality and time-efficient ethics review of RD research proposals, accelerating their implementation.
0	**Re(ACT) Congress and IRDiRC Conference**	Activity	Promotion of scientific cooperation and research on rare and orphan diseases in a unique Face-to-Face congress that fosters RD Research and Policy independently on medical domain.

EJP RD

EJP RD catalogue

PILLAR	ELEMENT/ASSET	TYPE	VALUE-CHARACTERISATION (SUMMARISED)
1	**Monitoring**	Service	Tool that permits the centralisation and analysis of monitored funded projects, reinforced by feedback from experts. Annual collection of information from projects and analysis of their progress.
1	**Networking Support Scheme**	Activity	Organisation of networking events that involve RD research's stakeholders, key for RD research to advance. Encouragement of underrepresented stakeholders and countries' involvement.
1	**Research Challenges Scheme**	Activity	Innovative funding scheme involving consortia of applicants (Academia, SME, and PAOs) Advocacy Organizations) and industry sponsors. Public-private partnerships facilitator.

EJP RD

EJP RD catalogue

PILLAR	ELEMENT/ASSET	TYPE	VALUE-CHARACTERISATION (SUMMARISED)
3	**Orphanet training material & module**	Activity	Empowerment of Orphanet Network members at national level, to deliver local training of the nomenclature and ontology. Training for trainers' sessions and trainer's toolkit.
3	**Training quality assurance Next Generation Sequencing (NGS)**	Activity	Large scope on NGS diagnostics, with yearly updates, helping in the translation of research tools to diagnostic applications. Up to date trainings with at international level with expert trainers.
3	**Registries and undiagnosed courses**	Activity, Resource	Opportunity to meet and create networking among professionals involved in RD registries, undiagnosed rare conditions and FAIRification, promoting further interaction & collaborations
3	**Trainings on biobanks and samples**	Activity, Output	Favouring communication and knowledge exchange about RD research biobanking and related operating procedures to deliver training, increasing visibility and sharing of preparatory material.
3	**Program delivery for the 3 schools (EURORDIS Academy)**	Activity	Empowerment of rare disease patient advocates with knowledge and skills to take part in patient engagement roles side-by-side with researchers, regulators, and industry and policy makers.
3	**Workshops/training contents for paediatric patients**	Activity, Output	Facilitator of the involvement of paediatric patients in ethically sound rare diseases research. The workshops aim to provide young rare disease patients with engagement skills in RD research.
3	**Online academic course (MOOC)**	Activity, Output	Cover the lack of transversal training on RD, by delivering free, easily accessed, and ergonomic MOOCs on transversal rare diseases research topics, with scalable content, to a wide audience.
3	**ERN Workshops (training multiplicators)**	Activity, Output	Sharing of knowledge and methodologies applicable for several different RD fields, highly relevant for ERNs, at the European level, with expectative of accreditation.
3	**ERN Workshops material**	Output	ERNs Workshops (Training multiplicators) material. This training material comprehend research concepts and topics that are cutting edge and of cross-ERN benefit.
3	**Secondments (Mobility fellowships)**	Activity, Output	The fellows have the possibility to obtain a highly specific and tailored training, acquiring new research experiences and learning new methodologies, through exchanges between institutions.

EJP RD

EJP RD catalogue

PILLAR	ELEMENT/ASSET	TYPE	VALUE-CHARACTERISATION (SUMMARISED)
4	**Innovation Management Toolbox**	Service, Resource	Centralised repository of resources that provides searchability and answers to specific questions. It may contain outputs as the Sustainability Handbook.
4	**Mentoring Service**	Service	Easy access to full gamut of therapy development expertise that increases the success rates in translational research projects of individual researchers by stimulating interdisciplinarity and building capacities.
4	**Clinical Trials Support Office**	Service	Support/advice for the planning and design of clinical studies for Rare Diseases (RD), with special focus in multinational research. Built up on the expertise of partners with years of experience in their respective fields.

EJP RD

EJP RD catalogue*

**Under construction for P2*

PILLAR	ELEMENT/ASSET	TYPE	VALUE-CHARACTERISATION (SUMMARISED)
2	**Virtual Platform assets**	Service, Resource Activity, Output	Federated platform of resources for RD research that facilitates the discovery, access and analysis of data and samples scattered across several resources in Europe. It represents the central value of the research ecosystem.

**Under construction for P2*

VIRTUAL PLATFORM

STEPS

01

MATURITY
CONFIRM ASSETS x
MINIMAL VIABLE PRODUCT

↓

CATALOG OF PRODUCTS
WITH VALUE PROPOSITIONS

02

SET OF QUESTIONS FOR THE ANNUAL RETREAT (besides step 1)

- ❑ Operating model vs excelled model
- ❑ Components that could:
  - Be standalone vs Interdep*
  - Reusable/ Reusable for other purposes
  - Interchangeable with other platforms/ projects (distributed efforts)
  - Stakeholders (bundles)

03

ROUNDS OF
BUSINESS MODEL CANVAS

ALREADY STARTED

EXPECTED BEFORE
SUMMER

04

INFORMATION-SERVICE PROVIDER

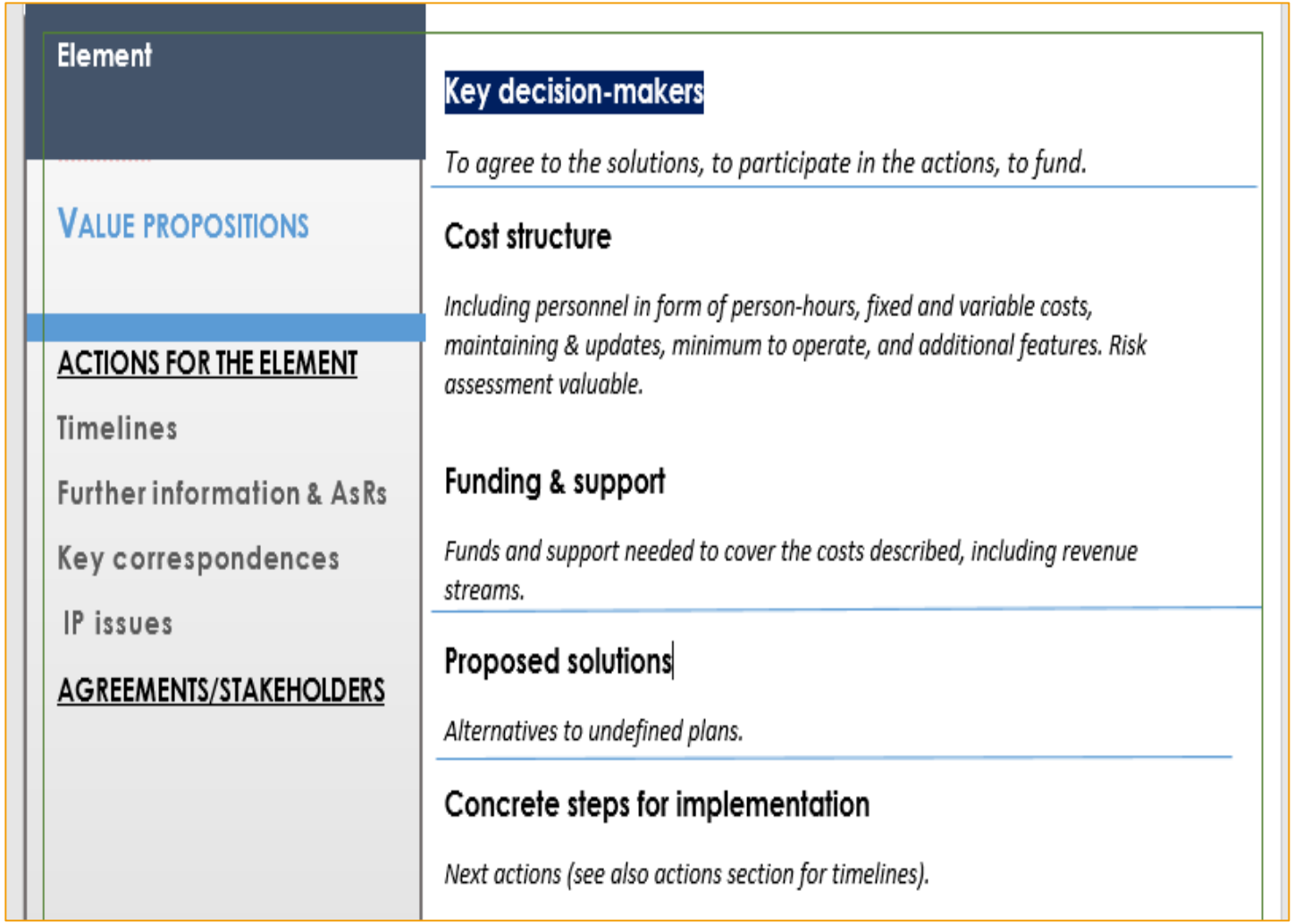
SUSTAINABILITY PLANS PROPOSALS/ALTERNATIVES

STAKEHOLDERS APPROACH

Individualised feedback

➢Will start a round of individualised feedback*

- Revisit value propositions if needed
- Costs structures when needed
- Funding alternatives and stakeholders approaches



*This feedback may be done in collaboration with the External Service Provider

Services Roadmap

Elements that:

- have considerations as services (Virtual Platform assets excluded for now);
- may share channels, communication, ítems and resources

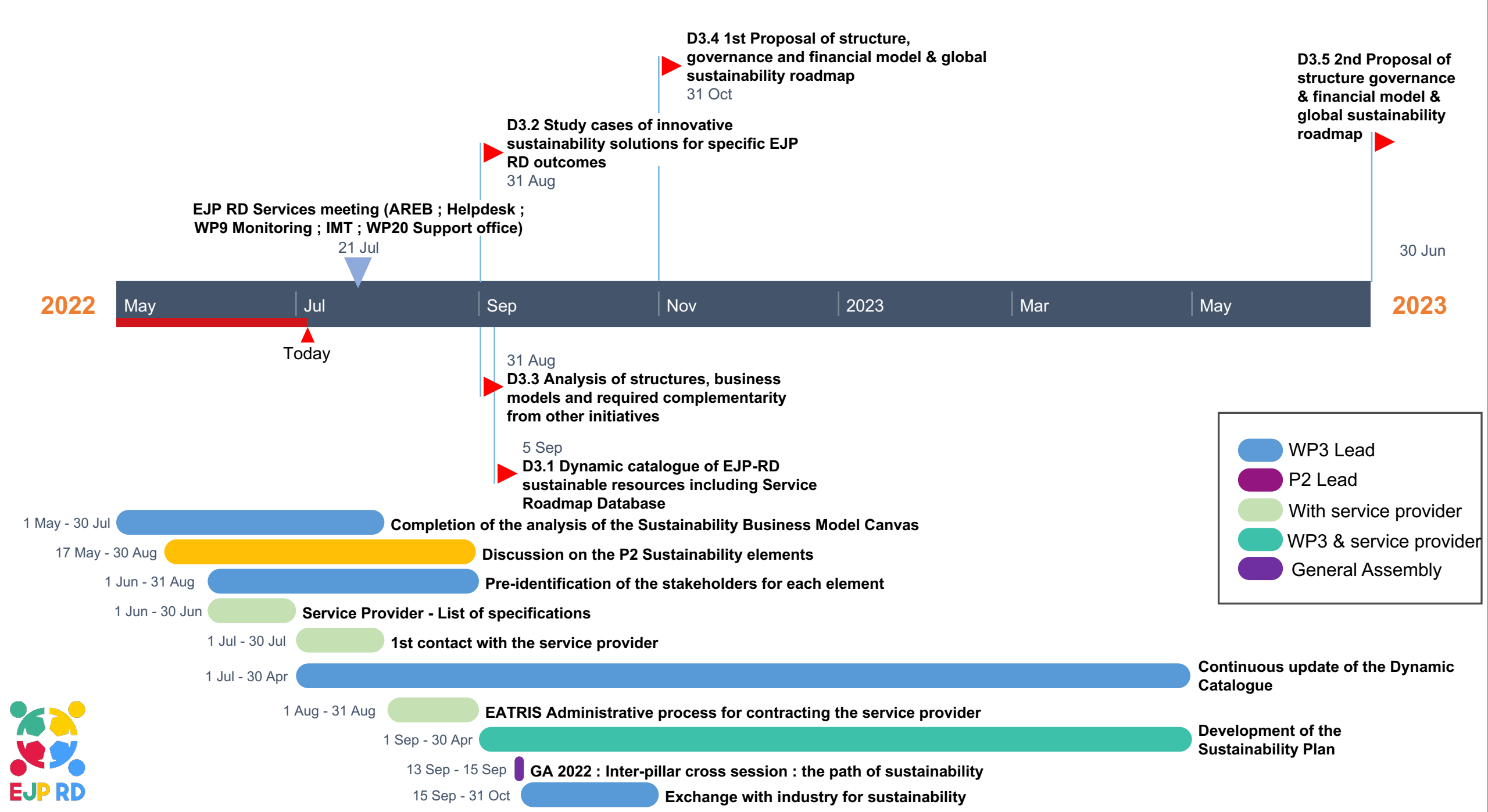
This roadmap includes:

- **Coordination services EJP RD & IRDiRC SciSec.**
- **Central Helpdesk.**
- **Advisory Regulatory Ethics Board (AREB).**
- **Monitoring.**
- **Innovation Management Toolbox.**
- **Mentoring services.**
- **Clinical Trials Support Office.**

An internal meeting is planned

Next actions

- Complete P2 processes (steps presented at P2 retreat)
- Individualised feedback
- Services Roadmap internal meeting
- Stakeholders approach, including industry connections and Policy Board suggestions and questions (last slide).


D3.4 1st Proposal of structure, governance and financial model & global sustainability roadmap
31 Oct
D3.5 2nd Proposal of structure governance & financial model & global sustainability roadmap
D3.2 Study cases of innovative sustainability solutions for specific EJP RD outcomes
31 Aug
EJP RD Services meeting (AREB ; Helpdesk ; WP9 Monitoring ; IMT ; WP20 Support office)
21 Jul
30 Jun
2022
May
Jul
Sep
Nov
2023
Mar
May
2023
Today
31 Aug
D3.3 Analysis of structures, business models and required complementarity from other initiatives
5 Sep
D3.1 Dynamic catalogue of EJP-RD sustainable resources including Service Roadmap Database
WP3 Lead
P2 Lead
With service provider
WP3 & service provider
General Assembly
1 May - 30 Jul
Completion of the analysis of the Sustainability Business Model Canvas
17 May - 30 Aug
Discussion on the P2 Sustainability elements
1 Jun - 31 Aug
Pre-identification of the stakeholders for each element
1 Jun - 30 Jun
Service Provider - List of specifications
1 Jul - 30 Jul
1st contact with the service provider
1 Jul - 30 Apr
Continuous update of the Dynamic Catalogue
1 Aug - 31 Aug
EATRIS Administrative process for contracting the service provider
1 Sep - 30 Apr
Development of the Sustainability Plan
13 Sep - 15 Sep
GA 2022 : Inter-pillar cross session : the path of sustainability
15 Sep - 31 Oct
Exchange with industry for sustainability

Policy Board meeting questions

- What **specific stakeholder(s) in your country** can contribute to the sustainability of the EJP RD elements?
- Does your country have any investment roadmap or support service for RD that might be aligned with the sustainability plan of EJP RD elements (apart from project calls?)
- Do you have any national resource that would connect to the Virtual Platform?
  - In case there is any, how is this national resource supported/sustained at the national level? (This information would be valuable also for the forthcoming Rare Disease Partnership (RDP))

EUROPEAN JOINT PROGRAMME
RARE DISEASES

THANK YOU!

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@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