EJP RD
European Joint Programme on Rare Diseases

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

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Del 1.6
Fourth report from the annual ExCom and Policy Board meeting

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

Due date of deliverable: month 43

Dissemination level:
Public

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List of Abbreviations

AAI	Authentication Authorisation Infrastructure
AOB	Any Other Business
AREB	Advisory Regulatory Ethics Board
AWP	Annual Work Plan
Coo	[EJP RD] Coordination [Team]
EC	European Commission
EJP RD	European Joint Programme on Rare Diseases
EMA	European Medicines Agency
ERN(s)	European Reference Network(s)
EUPID	European Patient Identity Management
ExCom	Executive Committee
GA	General Assembly
GB	Governing Board
GDPR	General Data Protection Regulation
IHI	Innovative Health Initiative
IMI	Innovative Medicines Initiative
IT	Information Technology
JTCs	Joint Transnational Calls
JRC	Joint Research Center (of the European Commission)
KPI	Key Performance Indicator
KRI	Key Result Indicator
MOOC	Massive Open Online Course
MS	Member States
NMG	National Mirror Group
NSS	Networking Support Scheme
OD	Orphan Drugs
P0	Pillar 0
P1	Pillar 1
P2	Pillar 2
P3	Pillar 3
P4	Pillar 4
PB	Policy Board
PM	Person Month
RD	Rare Disease
SRIA	Strategic Research and Innovative Agenda
VP	Virtual Platform
WP	Work Package

EJP RD Executive Committee

5th of July 2022
14:00 – 18:00
Online

Attached documents:

- *Slides presented during the meeting: file "Annex1_20220705_EJPRD_ExCom-Meeting_slides"*
- MIRO extract _ Addressing delays in tasks:
  - file "*Annex3_20220705_EJPRD_ExCom-Meeting_MIRO_Delays in Tasks*"
  - view link: https://miro.com/app/board/uXjVOonZYr4=/?share_link_id=172865242533
- MIRO extract _ Sustainability Roadmap for EJP RD
  - file "*Annex4_20220705_EJPRD_ExCom-Meeting_MIRO_Sustainability*"
  - view link: https://miro.com/app/board/uXjVOo9QeMI=/?share_link_id=582244008581

List of participants

Name	Institution	Role	Presence
Daria Julkowska	INSERM	coordinator WP1 - WP5	Excused
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	Present
Roseline Favresse	EURORDIS	P3 coleader WP15 - WP18	Present
Biruté Tumiene	VUHSK	P3 coleader WP18	Present
Anton Ussi	EATRIS	P4 coleader WP3 - WP19	Present
Rima Nabbout	AP-HP	P4 coleader WP20	Present
Eva Bermejo-Sanchez	ISCIII	WP2 and WP3 coleader	Present
Manuel Posada	ISCIII	WP2 and WP3 coleader	Present
Annalisa Landi	FGB	WP4	Present
Barbara Sanavio	FTELE	WP4 - WP19	Present

Name	Institution	Role	Presence
Christine Fetro	FFRD	WP8	Present
Irit Allon	CSO-MOH	WP9	Present
Anthony Brookes	ULEIC	WP10 - WP12	Present
Sergi Beltran	CNAG-CRG	WP11	Present
Marco Roos	LUMC	WP12	Present
Chris Evelo	UM	WP13	Present
Friederike Ehrhart	UM	WP13	Present
Claudio Carta	ISS	WP14	Present
Magda Granata	FFRD	WP16	Present
Krystyna Chrzanowska	IPCZD	WP18	Present
Tanja Bülow	UKA	WP20	Excused
Liron Even-Faitelson	CSO-MOH	WP9	Present
Maria Del Carmen Sanchez Gonzalez	ISCIII	WP2 - WP3	Present
Florence Guillot	ANR	WP6	Present
Ben Lydall	EATRIS	WP3	Present
Catherine Nguyen	INSERM	IT GGB director	Present
Juliane Halftermeyer	INSERM-Transfert	Coo team	Present
Aniket Sharma	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
Victor Ombredane	INSERM	Coo team	Present
Hiba Abou Daya	INSERM	Coo team	Present
Clement Moreau	INSERM	Coo team	Present
Loranne Charrier	INSERM	Coo team	Present

Agenda

○ **Addressing delays in tasks:** How to improve task execution and management until the end of EJP RD ○ In order to prepare the discussion, a survey was completed with information on current delays in Tasks in each WP as well as mitigation measures taken/to be implemented were indicated
○ **Sustainability of EJP RD results**
Break
○ **Update on the Rare Diseases Partnership**
○ **General Assembly and Consortium meeting**
AOB ○ Reminder about the AWP Y5 ○ Other

Minutes

Addressing delays in tasks

See MIRO extract _ Addressing delays in tasks:

- file "*Annex3_20220705_EJPRD_ExCom-Meeting_MIRO_Delays in Tasks*"
- view link: https://miro.com/app/board/uXjVOonZYr4=/?share_link_id=172865242533

1st delay: delay in the submission of deliverables on the EC portal

Discussion:

- More strict deadlines need to be implemented by the coordination. It was recommended to put the timeline that was established for each type of deliverable, and that considers the Pillar/Work Package leaders' review period, within a calendar to be made available through the Teams channel of the EJP RD "Leaders".
    - The current process is as follows: pillar leaders need to review the deliverables of their respective pillars. The Pillar 0 (P0) deliverables needs to be reviewed by the EJP RD Executive Committee (ExCom).
        - there is a defined timeline: deliverable will have to be submitted for ExCom 21 days before the submission deadline. The ExCom has 10 days to review it; the deliverable responsible (within its leading beneficiary) has 7 days to implement the suggested modifications, 3 days will be left for finalising the deliverable and
        - submit it to the EC.

- Reward system to be adopted for the people who send their deliverables on time.
    - Currently there is a system of virtual rewards on Microsoft Teams where acknowledgement of achievers can be highlighted. A designated EJP RD beneficiary is leading each deliverable, its responsible can use such system of rewarding.

ACTIONS

→ **Coordination will send more frequent reminders to announce the up-coming deliverables & declare them during Pillars and WP conference calls**

→ **Reward system to be utilized by the deliverable leader when submitting deliverables on time**

2nd delay: Rare Diseases Research challenges: 3 funded projects started later and will end after EJP RD

Discussion:

- Only one of the three projects would be concerned during the extension
- Mitigations proposed as the projects are co-funded by the EU and the industry: Have the third instalment paid by the EC only and the last instalment paid by the involved industry.

ACTIONS

→ **Coordination proposes the above mitigation measure to the EC to inquire about its feasibility.**

3rd delay: FAIRification of resource types other than ERN registries, (potential delay)

Discussion:

- Explanation from Pillar 2 partner: initially 10 resources were planned for FAIRIfication, that number increased as the work involved more than the 24 ERN registries (an overfit for the ERNs compared with what was planned); more support and efforts were and are being provided for these resources (than initially planned).
    - Nevertheless, the FAIRification of some resources is easier (requiring less efforts from the FAIRification stewards) as these latter have already performed substantial FAIRification work already such as the Work Package 13 resources that are being addressed the last two years of EJP RD (following the FAIRification (support) undertaking of resources other than ERNs registries)
- Additional FAIRification efforts (including those performed by the FAIRification stewards) are being considered within the budget allocations being performed for the Annual Work Plan Year 5 (and extension) of EJP RD. This also involves the reinforced coordinated work between the partners working on onboarding resources inside the Virtual Platform ecosystem (at the metadata level and the record level) and the FAIRification involved partners.
- Regarding the point raised by coordination, it should be brought to the Pillar 2 general calls, because it's kind of a strategic decisions on moving on, allocating resources for priorities for people that are already funded to do some things and that they are just shifting their focus.

ACTIONS

→ **Reinforce the ongoing coordinated work between the FAIRification involved partners and other working groups liaising with resources for onboarding into the Virtual Platform**

→ **Discuss the identified additional need of FAIRification resources (Person-Months) during the next Pillar 2 general call.**

4th delay: Pillar 3 Training for patient representatives and advocates on leadership & communication skills

Discussion:

- The mitigation measure adopted was doubling the capacity of the training for the last year as agreed with the EJP RD coordination (it will be organised in Poland and will host 60 participants instead of 30).
- No other training in pillar 3 will need such mitigation.

ACTIONS

→ Mitigation measures have been put in place to solve any delay - No further specific actions needed.

5th delay: Pillar 4 first mentoring report

Discussion:

- The original intention to delay the first report until after JTC 2022 was to add to the data set of the mentoring report and improve its quality. The first draft is currently in-preparation.

ACTIONS

→ Mitigation measures have been put in place to solve the delay - No further specific actions needed Mitigation measures have been put in place to solve the delay - No further specific actions needed.

6th delay: submission delay of the periodic report on the EC

Discussion:

- Delays are regularly linked with the amendment process of the European Commission
- Do we get feedbacks on the deliverables that we submit?
    - We received some feedback from the mid-term review in 2021, the experts that were involved in the review went into details for each deliverable that were submitted so far. All the deliverables are also reviewed by our Project Officer at the European Commission and can be rejected. If needed, the Project Officer can also request a review from external expert for some of the deliverables. These feedbacks do not report on the length of the deliverables (i.e., if they are too short or too long).
    - For the Year3 (2021) technical periodic report, we have been asked to provide a summary of the whole report to facilitate the review and validation. The full technical periodic report remains the reference for reporting the work progress. EJP RD is a large research programme with 21 Work Packages (and is not a single research project); it is difficult to report on the progress of its different parts through a short report.

ACTIONS

→ **Coordination will discuss the amendment process with the EC to explore the possibilities to have more flexibility, especially in consideration of the coming Rare Diseases Partnership.**

7th delay: JTC 2019-2020 projects end-date delayed due to COVID-19

Discussion:

- One possibility is to have some financial reports, like interim reports, by the end of EJP RD as a justification on how the funding went for these projects.

ACTIONS

→ **EJP RD coordination continues the follow-up with the European Commission to have the answer on the information needed to justify the full funding of (co-funded) JTC projects.**

→ **Prepare financial and interim reports at the end of the EJP RD as justifications of this funding.**

8th delay: (Virtual Platform) Architecture that demonstrably supports federated analytics, automatic adaptation and their demonstrations (potential delay)

Discussion:

- Federated analytics (on FAIR Data) task force has been set to move forward with some Virtual Platform aspects as a mitigation measure for this delay.
- what exactly did we commit to provide at the end of the project? What do we have to achieve?
  - Specific detailed information on federated analytics has not been provided at the start of EJP RD, but the aim is to have a Virtual Platform that is FAIR based to the record level (demonstrating machine-readability of record level data). It is also about the adaptation of the VP to what the resources provide, which is a demonstration of the FAIR approach.
  - The Virtual Platform was defined as having the capabilities to federating discovery query and analyse different heterogeneous sources of data; WP13 activity includes providing this verified piece of data that could be utilized for analysis purposes and demonstrate this analysis feasibility. Then, through the convergence of the different pieces of work in different Work Packages, go up to the federated analysis capabilities enabled by the resulting architecture and framework.

ACTIONS

→ No specific actions identified

9th delay: EUPID demonstration application delayed

Discussion:

- Pillar 2 partners expressed concerns regarding the delay in EUPID (European Patient Identity Management) implementation, that is an implementation of a Privacy-Preserving-Record-Linkage system that allows to link data of the same patient across resources (avoiding for example counting a patient twice and having the patient's data scattered). There were some perceived concerns about the security EUPID. Thus, another Privacy-Preserving-Record-Linkage

system, called SPIDER, was developed by the Joint Research Center (JRC) of the European Commission because of concerns that EUPID is not cyber- safe and can be hacked. Partners found themselves in front of two complementary or competing systems which complicated the implementation. EUPID security concerns did not result in any data breach/leak, the EUPID services were upgraded to address any security concern.

- One of the outputs of the meeting between the Pillar 2 involved partner, the EJP RD coordination and the JRC, was to provide the timeline for the work on Privacy-Preserving-Record-Linkage that is being updated and that shows the past and coming implementations of such systems. This timeline will be reviewed and discussed during the next pillar 2 general call with an emphasis on the pilots that were planned with EUPID, the next steps, and outputs foreseen until the end of EJP RD. JRC will look on how to integrate SPIDER into that timeline. Interoperability between the different Privacy-Preserving-Record-Linkage systems is needed (as different systems are already integrated in various resources.

ACTIONS

→ **Pillar 2 will review & update the timeline, Pilots and outputs of Privacy-Preserving-Record-Linkage implementation considering several systems**

10th delay: implementation of LifeScience AAI (if necessary)

Discussion:

- Pillar 2 partner expressed uncertainty regarding the activities or connections to which LifeScience AAI (Authentication Authorisation Infrastructure) implementation is needed. So far, we were working on the discovery of resources and metadata, so AAI was not needed for much (and not all) of the work performed until now. AAI is needed as we start digging more and more into the resources with data access authorisation arising needs. The point is to decide which resources needs AAI and what use-cases will be created for its implementation.
    - The implementation of LifeScience AAI is in progress now that this service is available since the past few months; the use cases about interacting with data will be developed in the next months. A first version of LifeScience AAI implementation will be released in the coming months.

ACTIONS

→ **Pillar 2 partners will discuss and develop implementation use-case(s) with LifeScience AAI**

11th delay: full onboarding all the planned resources on the VP (potential delay)

Discussion:

- The delay was discussed during the annual retreat of Pillar 2, and the partners designed a strategy to address it. Pillar 2 will be able to advance on the work and provide updates by the end of this year (2022).
- The level of onboarding is yet to be defined as there are different levels. Guidance to the resource on how to get to each level will need to be provided.
    - It was suggested to start onboarding the maximum feasible number of resources through the basic level and then build on that, guiding some resources to the most sophisticated level of integration to the Virtual Platform.
- The onboarding process is planned to be optimised and documented; resources will continue being onboarded while a general onboarding manual will be prepared to be used by the resources that will connect later to the Virtual Platform.
    - This onboarding process is linked to the Deliverables 10.x (the next one is deliverable 10.4). There are also other related deliverables from WP11 where we provide updates on how the data has been populated in each type of resources and what are the next steps towards improvement in the rare disease field. These deliverables could have a subsection on connection readiness or connection status to the VP ecosystem.

ACTIONS

→ **Define the different levels of resource onboarding into the Virtual Platform and guidance for each level**

→ **Provide update on the onboarded resources at the end of the year (2022)**

Sustainability of EJP RD results

See slides 2-14 from the attached presentation "Annex1_20220705_EJPRD_ExCom-Meeting_Slides"

Processes summary: see slide 3 for complete information

EJP RD catalogue of elements/assets: see slides 4-8 for complete information

Discussion:

- WP3 team should think about adding in the catalogue, resources that are being sustained (for now) and should define some criteria about the fact that the sustainability is being assured ; in order to avoid missing some elements.
    - This is planned considering the period after the EJP RD end. Indeed, certain elements can be sustained already. WP3 Coordination team should classify these elements in the catalogue and could think about making a separate report on what is already coming in as sustainable.
- Certain services are eventually set up outside of the EJP RD. Some definition of the elements considered would help.
    - WP3 Team should add some clear definitions on how it understands these terms, on the Sustainability Handbook.

- There is a need to foresee strictly what are the targets for the sustainability:
    - European level or other partners' national level
    - Adaptability
    - Scalability
  - For example, about the trainings in Genomics, maybe they are not needed anymore at European level, but it would be absolutely required on a national scale. Therefore there is a need to target our questions to distinct levels and to consider the adaptation of the scale.
  - some elements are not going to be sustained as they stand, in this case the aim would be therefore not to look at each activity and ask whether it will be maintained as it is, but what can be maintained from that activity and how it can be scaled up or how some of it can be used usefully even if the activity itself discontinues.

Sustainability of Pillar 2 elements: see slide 8 for complete information

Individualized feedback: see slide 9 for complete information

Sustainability roadmap of EJP RD services: see slide 10 for complete information

Discussion:

- Consideration about the fact that the roadmap of services did not change much since it was initiated. How much was it used during the EJP RD lifetime and which of these is really more worthy to be sustainable ? What was the effective activity or usefulness of these services ?
  - On one side, some of these elements are still being developed. On the other side, one of the tasks of the WP3 is to provide the roadmap by the end of the EJP RD. Besides, most of the elements presented were captured during the surveys on sustainability that were sent to the task leaders asking them to consider which elements were worth to sustain. So, the worthiness of the elements is assumed. In addition to that, some deliverables are showing the activity and the use of these services.
  - There is a need to think about "how to add the value of using these elements in order to promote their sustainability and to give the desire for the different stakeholders to use them"; "what can we "sell" for the future of the Partnership, or sell to other partners and investors?"

- EJP RD community is trying to sustain all these central services through the Rare Disease Partnership for the next 7 or 10 years. So why develop this very detailed and complex roadmap when we are actually in the process of defining the Strategic Research and Innovative Agenda (SRIA) of the Partnership where we try to take up as many of the useful elements that we have.
  - This is an important discussion that we are having about the SRIA, the role of the Rare Disease Partnership and how to link the EJP RD elements to this latter.

Sustainability workplan timeline: see slides 11-12 for complete information

Policy Board meeting questions: see slide 13 for complete information

Questions to be addressed to the Policy Board meeting:

- 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)

Discussion :

- The third question that will be addressed to the Policy Board meeting "how is this national resource supported/sustained at the national level ?" should be kept rather broad because in some countries they could have private support in addition to national support.
- Policy Board members have received information and documents in advance of the meeting to get prepared and the MIRO board will stay open until the 19th of July. Policy Board members will have the possibility to consult experts on their own country.

Miro Session

See MIRO extract _ Sustainability Roadmap for EJP RD

- file "*Annex4_20220705_EJPRD_ExCom-Meeting_MIRO_Sustainability*"
- view link: https://miro.com/app/board/uXjVOo9QeMI=/?share_link_id=582244008581

- **Question: *Which updates do you have relating stakeholders (sustainers) interested in funding the element?***
- **Exercise description:** each element / asset described in the previous presentation is included in the MIRO board. Each ExCom member added updates / inputs of potentials stakeholders that could ensure the sustainability of each element.
- **Additional comments:**
  - Regarding the platform for results, using Horizon Europe mandatory platform could help https://ec.europa.eu/info/funding-tenders/opportunities/portal/screen/opportunities/horizon-results-platform
  - People can offer services on top of the material (element/asset) and that extra expertise and counselling using the material can be charged as part of sustainability model.
  - The Innovation Management Toolbox and the mentoring service have a link between them: the mentoring service would probably refer people to innovation management toolbox, but they may be potentially standalone: One does not rely on the other.

- Is it feasible that some parties inside or outside the EJP RD would start offering a mentoring service not for free using the materials from EJP RD as a commercial activity?
  - ◊ A lot of the materials are based on these parties, it is possible to adopted a not-for-profit service for covering costs. This should not be very commercially viable.
  - ◊ there should be a business model for providing trainings: EJP RD developed the resources and the tutorial material. Potential 'clients' could pay mentors, for example for one day of lectures showing how our resources work.

- Regarding any potential "competition" between university/research institution technology transfer offices: unless they have regulatory - drug development expertise on rare disease, competition is limited for near clinic project (i.e. pre-competitive area).
- The work with transatlantic partners is particularly important; currently we may be duplicating a lot of work on both sides of the Ocean. Collaborations is needed, especially about sharing data from patients across continents.
- ISS (Italy) is obviously interested maintaining some of the training activities.

ACTIONS

→ **The WP3 Coordination Team must focus on the targets of activities to be sustained.**

→ **There is a need to consider the adaptation of the scale : a focus on what can be maintained from each activity and how it can be scaled up.**

Update on the Rare Diseases Partnership

See slides 16-28 from presentation "Annex1_20220705_EJPRD_ExCom-Meeting_Slides"

Discussion

- **About the proposed structure for the SRIA:**
  - From a technology direction, there are still opportunities that EJP RD is not covering entirely particularly for the analysis parts.
    - The General objective of the Rare Diseases Partnership (RDP) is addressing these concerns: it comprises the analytical part.
  - The proposed structure of the RDP is going straight from vision to objectives. With the objectives we might get too specific and not strategic.
    - In Horizon Europe, "General Objectives" mean "Impacts" and the "Specific objectives" are the outcomes of the whole Partnership. And then there are the Operational objectives as the concrete actions (as shown in the proposed intervention logic).

- The SRIA should not focus on "What are we going to do?" but "How are we going to do better?" There is a need to use the experience from EJP RD and try to make the ecosystem a better performing and coherent tool
  - The Chapter 1 needs to explain how we can improve the ecosystems performance by not only looking at the research, but on the actual system itself, what are the processes, how can we generate patient centrism and multidisciplinary. And then on each specific objective part: How we are going to implement? Who we are going to implement with, and these are the people or the communities in the national context.
- The need to gather the performance indicators is imposed by the EC. There are different types of performance indicators. For the moment there is a need to focus on the overarching one, the ones that are linking the different partnerships and the one of the RDP as a whole. Afterwards there will be more granularity in depth of the performance indicator linked to the operational ones that will be addressed at later stage. In this new model of monitoring for the partnership there are Performance indicator at several levels and that are going to be connected (one feeding the others). So, there will be the ones, that are most commonly known, relating to the operational objectives as the one we are having in EJP RD. But there will be others who are linked to the outcomes and others who are linked on the impact and others who are linked to the integration with other partnerships in the framework of Horizon Europe. On the RDP development Timeline, the Key Performance Indicators (KPIs) development will be developed from September 2022 to December 2022.

- **About the Specific Objective 1 : Patient-need led relevant research enabled by outcome-oriented Investments strategically deployed along the R&D value chain**
  - There will be a strong clinical focus of the RDP. That would mean that there is a need to have KPI which are quantitative, and which could demonstrate the functionality of the structures around the Clinical Research Network (CRN); considering that no clinical trials will currently be funded how can we demonstrate such functionality?
    - The synergies with other initiatives will be key and strong here. They should foster also launching the clinical trials, for example through a synergy with the EU4health, for EU investigator-initiated trials, or with collaboration with industry through IHI pr through direct industry collaboration.
    - There is a strong focus on clinic, but a strong focus is also put on the fundamental & pre-clinical side. The section thematic focus would need to be discussed by the SRIA Task Force to set such focus, considering that we are more on the research side (although it is intertwined with healthcare in the context of Rare Diseases).
      - ◊ In IHI, for example, the SRIA section 'thematic focus" describes criteria for projects prioritisation including the burden of diseases (as they are addressing the whole set

of diseases) and for themes that are not related to specific disease(s), they consider the transformational impact or innovation process of the different projects in order to be funded and that could also impact the work of IHI.

- It is important to be careful about leaving the treatment out of the care pathway. There is a need not to forget that treatment and therapies and clinical trials are also research and are highly needed.
  - There is a need to prioritize: the Concept Paper resource estimation is more than 500 million euros and for the moment we are at 200 million euros (considering the current EC contribution of 100 million euros).
  - The JTC experience on Investigator initiated clinical trials, was not conclusive. However, if the EU commits with enough money that is centralized and that is going to serve for funding the investigator Led trials within the RDP, this is a possibility.

- **About the SRIA development Timeline**
  - Changes to take into accounts after discussions:
    - The review of general objectives will be extended until the 30th of September 2022
    - The Final consolidation will start at the 1st of December 2022

ACTIONS

→ **The Coordination team and the SRIA Task Force will consider the comments received related to the structure and the content of the SRIA.**

→ **The Coordination team will update the SRIA timeline according to the discussion decisions**

2022 General Assembly and Consortium meeting updates

Updates from the coordination:

- Preliminary agenda has been shared with the EJP RD members as well as the venue information that was confirmed in Porto, Portugal.
- One interpillar session on September 12 from 9h00-10h30 is still empty. Coordination asked if the ExCom would like to use it for a new specific topic.
    - Suggestion about having instead separate parallel sessions for all pillars.
        - Pillar leaders are asked to inform the EJP RD coordination if such session is needed for their pillar.
- Hotel recommendations
    - EJP RD Coordination will check with the Portuguese colleagues on any hotel recommendations and/or event discounts.

ACTIONS

EJP RD Coordination has to:

→ **Send hotel recommendations to partners**

→ **Updated information on COVID-19 restrictions and travel conditions will be sent by coordination before the GA (ideally one week before)**

→ **Coordination reminded about registration deadline. The initial deadline (8th of July 2022) was extended until July 12th. A reminder will be sent on July 6th, 2022.**

ExCom members have to:

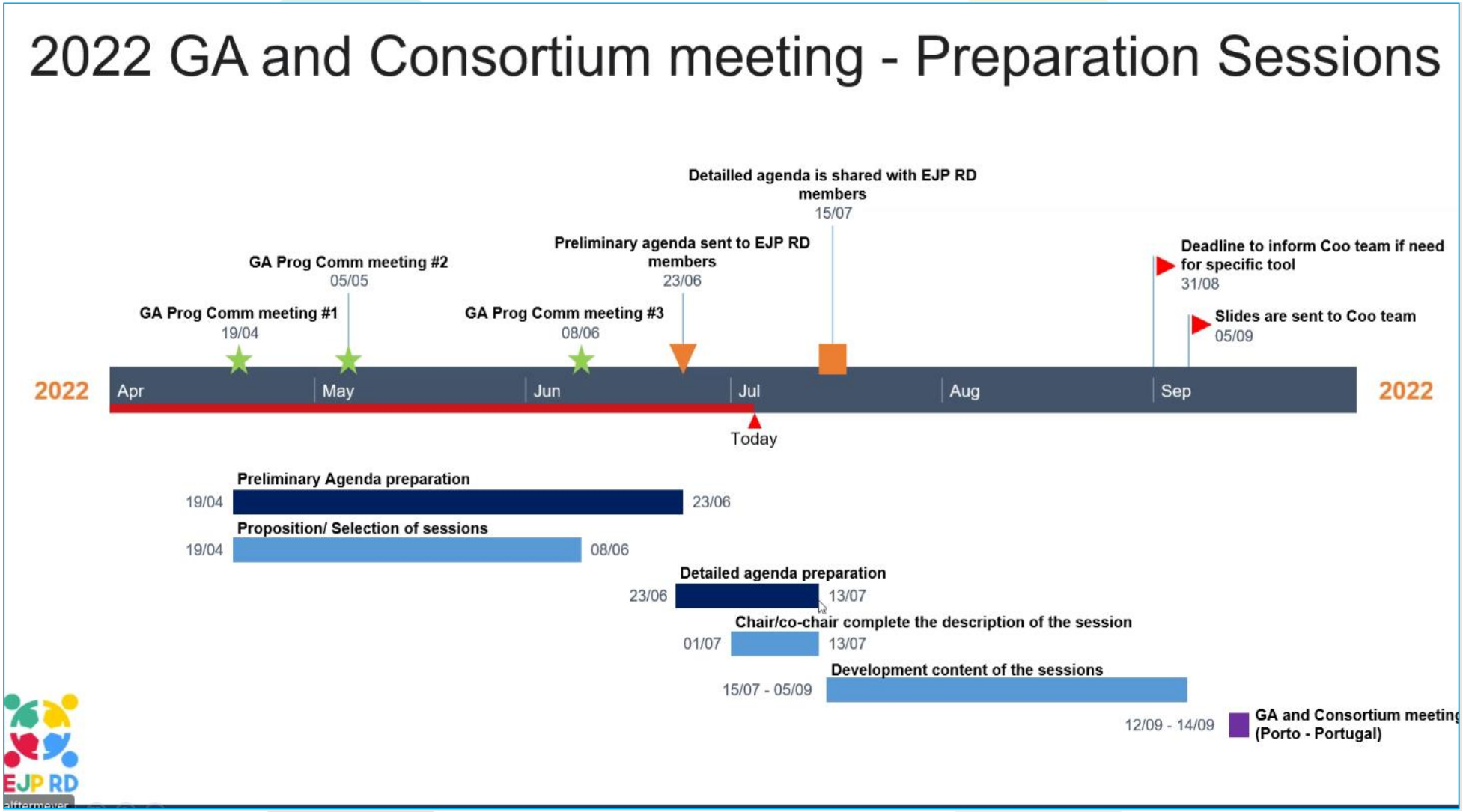
→ **Send their need for specific pillar session or idea of session for the remaining free slot to coordination by email until July 13th, 2022**. If nothing is received, the session will be cancelled and the sustainability session originally scheduled on day 3 will be moved to day 1 allowing for the meeting to finish earlier.

→ **Provide inputs about how to organise the two interactive use-case sessions.**

→ **Communicate the possible need of additional tools or materials for each session before the end of August 2022.**

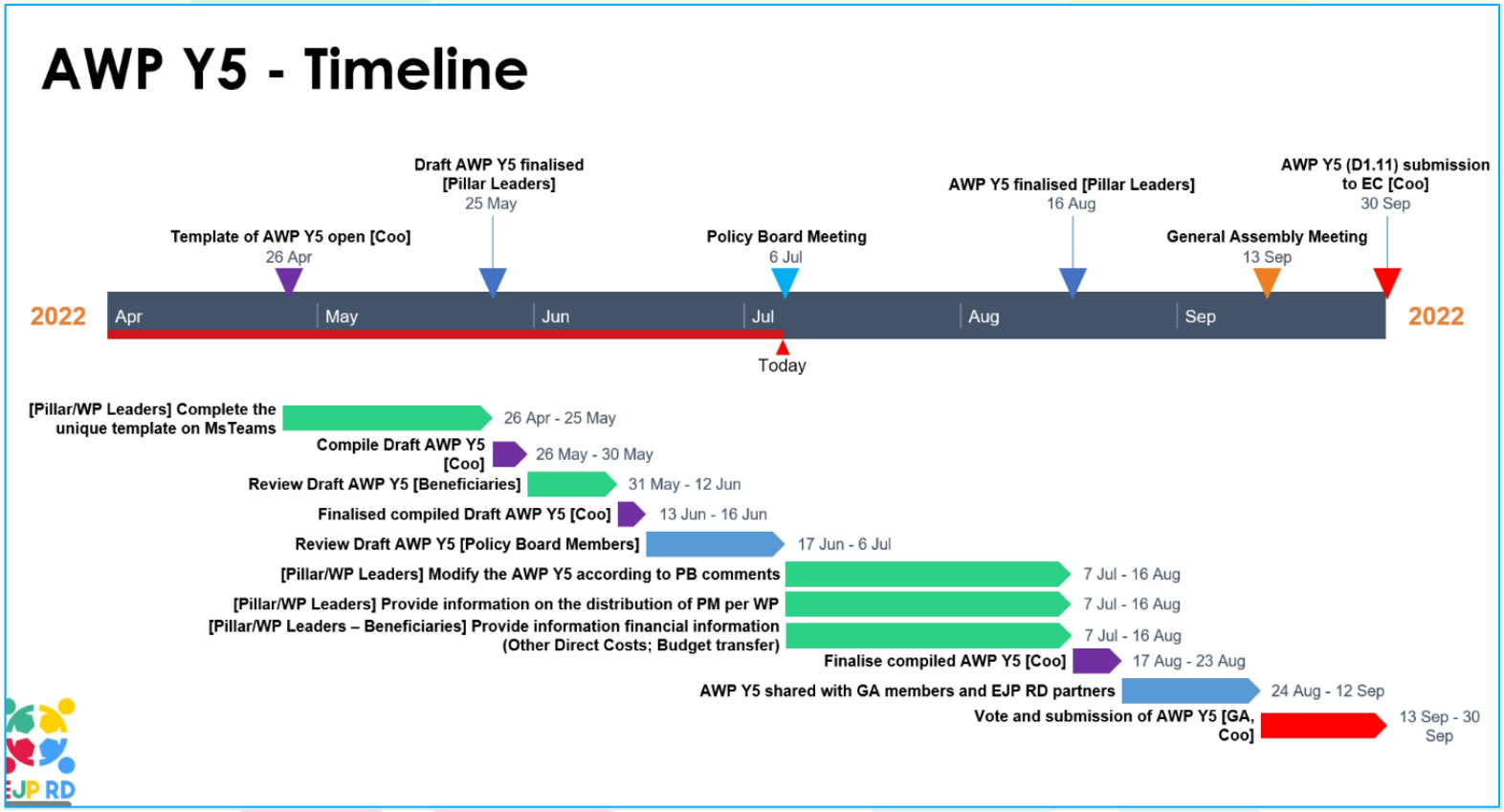
→ **Share the slides from speakers with the coordination by September 5th, 2022** (a dedicated folder in MsTeams will be provided end of August 2022 by the EJP RD coordination).

Timeline of GA meeting preparations:



AOB

Reminder on the Annual Work Plan Y5 timeline



- Policy Board members might give some comments on the AWP
- 3 Activities to be performed in the summer period before August 16th, 2022:
  - update the content of activities according to the Policy Board comments;
  - provide information about the distribution of Person-Months per Work Package;
  - provide the financial information on budget transfers that are needed.

EJP RD Policy Board and Governing Board meeting

6th of July 2022

13:30 – 18:00

Online

Attached document:

- *Slides presented during the meeting: file "Annex2_20220706_ PB-GB-Meeting_Slides"*
- MIRO extract _ Rare Diseases Partnership feedback:
  - file "*Annex5_20220706_EJPRD_PB-GB-Meeting_MIRO_RDP Feedback*"
  - view link: https://miro.com/app/board/uXjVOnjU8yE=/?share_link_id=618102436199
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  - view link: https://miro.com/app/board/uXjVOoUOpFQ=/?share_link_id=24315272785

List of participants

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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

Minutes

Welcome of new members

- **Tonia Bieber – Germany**: Federal Ministry of Education and Research representative.
- **Adrien Samson – Belgium**: Healthcare Biotech Manager - EuropaBio
- **Carmen Laplaza Santos:** European Commission - DG RTD
- **Szymon Bielecki:** European Commission - DG CNECT

Summary of EJP RD activities, achievements and impact (from Year 1 to 4)

See slides 4 to 24 "Annex2_20220706_EJPRD_PB-GB-Meeting_Slides"

Discussion

- **Regarding health system mobilisation and its readiness to take-up some of the new research results from EJP to RD patients? And how is that measured?**
    - Healthcare system is multi-dimensional, it includes several processes, many of which involve Data. When we are FAIRifying and building inter-operability between the Health information system elements such as registries and biobanks, leveraging for example on (meta)data models, standards and ontologies, we are opening the door from the Information Technology (IT) perspective between the healthcare system and the research ecosystem, so that one benefit the other through the mutual uptake of results (including data).
    - Additionally, through the funded projects, EJP RD is trying to accompany researchers in order to make their results FAIR from the start so that the results are discoverable and reusable.
    - Also, ERN representatives supported by EJP RD often have two hats: clinicians and researchers. In this regard there are some evaluations from the end-users about the benefits of EJP RD activities for ERNs (example, the joint EJP RD & ERICA workshop https://erica-rd.eu/wp-content/uploads/2022/06/ERICA-EJPRD-DataAccess-workshop-preliminary_agenda_v4.pdf). This is a kind of 'indirect/partial' effect on healthcare systems (uptake).
    - There are also the forthcoming results of the EJP RD JTC 2021 focused on Social Sciences & Humanities, including projects that more specifically target the development of new methodologies for the evaluation of RD patients including Quality of Life and other topics whose results shall be uptaken in healthcare systems.
    - The measure of the performance and impact of such work requires a long-term monitoring in which the Innovative Medicines Initiative and the Innovative Health Initiative could provide valuable guidance and return on experience.

- **Is there a significant difference regarding under-represented countries (uptake of research results) and the impact on patients?**
    - Expansion to the under-represented countries is a long process, that would take longer than the 5-years of EJP RD considering the starting situation with the resources that are currently involved; this why such expansion is planned for the RD partnership now that we are maturing the processes and tools/standards for such work. The RD ecosystem, including the virtual platform and all the IT services will be extended at the national level. This is one of the aims as described in the concept paper for the RD Partnership.
    - There are trainings planned in the Annual Work Plan focusing on the integration and use of the Virtual Platform ecosystem which would be adapted afterwards for national specificities, ensuring the exploitation/uptake of research results.
    - Indeed, one of the first preconditions for uptake of the results is the empowerment and capacity building. Indeed, in Pillar 3, some short-term measures are already present such as participants from under-represented countries are very active users of EJP RD services for education and training and are even more active than in other countries.
    - Every year, under-represented countries are getting more engaged in project proposals, which is also a short-term measure, but they are already providing a clear indication about the benefit.

ACTIONS

→ **The Policy Board** experience regarding indicators of healthcare impact and uptake of research results will be solicited

- **What about the regulatory acceptance and early engagement with the European Medicines Agency?**
- **What about the sharing of best practices between countries and regions and how are the less advanced countries benefiting from the more advanced ones especially at the level of diagnosis of RDs?**
    - For the aspects relating to the Virtual Platform, this has been planned since the beginning and a specific task for GDPR (General Data Protection Regulation) was put in place to be compliant with the regulation.
    - Regarding the research funding activities, this is one of the call text criteria where there is a need to obtain approvals. There is also the possibility to be accompanied by EJP RD afterwards regarding the intellectual property rights, legal and regulatory issues.
    - WP4, worked on an informed consent form securing the patient rights and fostering appropriate (data and patient) access conditions that are

regulatory compliant. This document was made available in 27 national languages (as an effort towards best practice sharing).

- Also, the Advisor Regulatory Ethics Board (AREB) of the EJP RD provides the applicants of EJP RD funded projects with 'regulatory suggestions' in order to undertake voluntary regulatory procedures, e.g., qualification procedure.
- Moreover, there is an online training course on regulatory aspects and data, (MOOC 5), which is currently in preparation to be launched next year.
- With respect to EJP RD interaction with EMA, it is currently concerning WP20 (calls) part focusing on clinical trials innovative methodologies that are been tested and others (innovative methodologies) being developed and tested. Discussion with EMA are ongoing to define a framework to qualify these methodologies that would then be promoted for use by the whole clinical trials community.
- Moreover, one of the WP19 activities is the mentoring activity that is accompanying the research projects, where key elements relating to the regulatory science are addressed by mentors and experts.
- Regulatory science is one of the key topics for the RD partnership as a continuation building on the current RD ecosystem to make it regulatory-compliant. This topic is expected to be addressed in most of the current specific objectives of the RD partnership.
- EJP RD is connected to IRDiRC, this latter has decided this year to create the regulatory science committee, (in addition to having the diagnostic, Therapeutic and inter-disciplinary committees), that will feed the EJP RD strategy as well.
- The ongoing EJP RD work on constituting National Mirror Groups (NMGs) will contribute to the sharing of best practices between countries and regions, including the under-represented countries, benefiting from the experience of the more advanced ones. These groups are also planned to meet to brainstorm and provide input for the Strategic Research and Innovative Agenda increasing thus the probability of benefit from the RD Partnership outputs.

- **The improved alignments of national and regional activities with EJP RD were presented as ranging between 23% and 86%, how does EJP RD follow this activity as it is critical for the KPIs of the future RD partnership where the alignment between national and European levels is a main goal?**
  - This is a long-standing effort from WP2 that focus on this alignment. EJP RD launches annual surveys inquiring on RD national plans and strategies with questions oriented towards the activities of the 4 pillars. This generates the provided overall estimation. Further development of the surveys is needed where additional questions can be added as elicited by the Policy Board & Governing Board meeting. Also, the need for a better explanation of the questions was noticed from the past national meetings that were organised to foster commitment to the RD Partnership. EJP RD currently have the figure of

23 to 86% alignment, depending on the activity, with a trend in improved alignment.

Presenting the training gaps identified and solutions proposed

See slides 25-39 "Annex2_20220706_EJPRD_PB-GB-Meeting_Slides"

- **Is there new approach to address the lack of knowledge and awareness about RD among the multistakeholder research community? And does the Annual Work Plan describe a new tactic to tackle this issue?**
    - Tactic is multi-pronged and there can't be a single solution. RD ecosystem is a better step to rectify the lack of knowledge, because the same participant which are educated through EJP RD later become participants of JTCs and also get to know about the resources and the virtual platform, etc.
    - The second major channel which is also developed is the national dissemination. For example, the train the trainers process that is planned to be scaled up for the RD Partnership. This is done through the development of training program for trainers that is standardized and of high quality and later it may be adapted to the local needs. It may be translated to local languages so that the trainings that are currently developed would be sustained. It was first applied in WP14.1 training on Orphanet nomenclature through national teams. So, there are several ways, on how to sustain, disseminate and scale up the trainings involving many levels and many measures.
    - What is noticed, for translational research, is that there is a little bit of a disconnection between what the research funder is requesting from the researcher and all these educational opportunities and tools available from the community. And since EJP RD brings the funder, the researcher and the infrastructures services together, it constitutes an opportunity also for the funder to be more prescriptive on encouraging people, especially if they are applying for grants to make use of the EJP RD materials, tools, infrastructures & services.
    - With regards to the funded projects, EJP RD is currently making kick-off meetings where all the programme services and tools are presented, so that participants become aware of their possibilities. It will be possible to measure the impact of this process as EJP RD is currently addressing questions to the funded researchers on how they use the services provided in the EJP RD. This will help in monitoring the use of the different services by the funded projects.
- A scalable system of amplification is required because this can't be done centrally. The national groups can be of direct benefit if they can be mobilized and if they have the resources to address the lack of knowledge and awareness.
- EC can disseminate the trainings to individual partners in different countries, which are a not much linked to the EJP RD network.

ACTIONS

→ **EJPRD will publish** the presented "training gaps identified and solutions proposed".

→ **The EC will help** EJP RD consortium to disseminate the trainings to individual partners in different countries, which are not much linked to the EJP RD network.

→ **The Policy Board is requested** to help with the national dissemination of the training activities

Annual Work Plan Year 5 – Feedback from the Boards

See slides 40 to 57 "Annex2_20220706_EJPRD_PB-GB-Meeting_Slides"

Questions submitted to the EJP RD policy Board and Governing Board, and feedbacks provided

- **Considering your overall knowledge of EJP RD and AWP Y5: what is missing in AWP Y5?**
  - No specific feedback was provided.

- **How does PB/GB members present and get back to their national stakeholders with the key points of the EJP RD about the AWP Y5?**
  - EJP RD Work Package leaders expressed difficulties in reaching the different countries with different national background and facilities to see what is suitable for each one of them. This will be facilitated by EJP RD during the coming NMGs meetings with the different countries.
  - In Portugal, the directorate general of health is building national guideline.
    - EJP RD coordination suggested to provide comments on this guideline according to the EJP RD activities and Results.
  - The possibility to arrange meetings between Work Package leaders and the national research communities was proposed to disseminate EJP RD services and activities. Such meetings would help in understanding the needs of the research communities.
  - In Germany, there is a group that looks at the national plan and all the RD issues. The outputs of EJP RD are regularly presented to the group and other funding initiatives in the field of Rare Diseases with the effort to present the most relevant outcomes to make the best use of them.
  - The Netherlands mirror group is also advocating the EJP RD approach and results promoting their take up .
  - Funders should be encouraged to promote the use of the standards of EJP RD ecosystem by the funded researchers. For example, funders would encourage research projects focused on data or building databases to consider connecting to the VP. Such strategy can be applied also to the trainings and any other activity.

- During the 2022 EJP RD General Assembly, a use-case session is planned to map the different type of activities and find out the different paths that would allow funded projects to know what EJP RD is doing and how to make use of it.

- The maturity model of the Beyond One Million Genome was presented to EJP RD by Sergi Beltran. This maturity model is meant to increase the capacities at the national and regional level as well.

ACTIONS

→ **EJP RD is suggested to** propose interventions from funders, research councils, funding infrastructure, facilities and academic institutes to prescribe to their research community the tools and services provided by EJP RD.

→ **EJP RD coordination proposed** to provide feedback on the national guideline prepared by the directorate general of health of Portugal.

→ **EJP RD coordination** needs to exploit national needs from the different countries during the NMGs meetings and the possibility to arrange meeting between Work Package/Activity leaders and national research communities.

- **Are there additional training needs that need to be set and how to ensure better translation of training needs?**
  - No specific feedback was provided.

- **How to make the research resources and data sources more visible for researchers in your country?**
  - No specific feedback was provided.

Preparation of the Rare Diseases Partnership

See slides 58-74 "Annex2_20220706_EJPRD_PB-GB-Meeting_Slides"

See MIRO extract _ Rare Diseases Partnership feedback:

- file "*Annex5_20220706_EJPRD_PB-GB-Meeting_MIRO_RDP Feedback*"
- view link: https://miro.com/app/board/uXjVOnjU8yE=/?share_link_id=618102436199

1st exercise: 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)

Comments:

- A central hub capability would need to be added to the Virtual Platform (e.g., for storage and analysis
- Next to an analysis hub, it is necessary to think of 'orchestration'. Analysis running over multiple sources needs orchestration. The Pillar 2 EJP RD did not do so much

address this aspect, but other projects did (e.g., Personal Health Train in NL and Germany).

- It would be interesting to have a continuous computational algorithm getting access to the data sources and learning from them including health data sources to automatically adapt to new resources in order to accelerate diagnosis, find new therapeutic option, accelerate the development of treatment, etc.
- The upscaling of the training should include national efforts.
- It is essential that the clinical research network is open and inclusive of the excellence in clinical research we have in Europe, besides the ERNs.

2nd exercise: Indicate any national activity or development that would be related to the Rare Diseases Partnership

Comments:

- In Canada, there is a rare diseases strategy that is being developed with a big research component that could be linked to a clinical trial network initiative.
- Netherlands: Development of the National Health research infrastructure (should be on translational research)
- ERICA-PROMs repository and state of the art: the 1st version of the repository is about to be launched after the summer as a webpage in ERICA website linking to the true repository that is a MAPI Research Trust pre-existing repository on clinical outcome assessment instruments including PCOMs and PROMs. After ERICA, for sustainability, this repository will be hosted by MAPI Research Trust.
- RD Moonshot Initiative : led by industry in order to see the different instruments for rare disease to foster public-private collaborations: good momentum to start collaborating in a broad sense.
    - EC plans to have in October 2022 the next meeting of the EC Group of national delegates from the different Member States and associated countries that are in interested in the RD partnership preparation → It is planned to invite colleagues from industry representatives to present this RD Moonshot Initiative.

Sustainability of EJP RD results

See slides from 75-88 "Annex2_20220706_EJPRD_PB-GB-Meeting_Slides"

See MIRO extract _ Sustainability Roadmap for EJP RD

- file "*Annex6_20220706_EJPRD_PB-GB-Meeting_MIRO_Sustainability*"
- view link: https://miro.com/app/board/uXjVOoUOpFQ=/?share_link_id=24315272785

1st exercise: Sustainability of EJP RD Results

Question 1: ***what specific stakeholder(s) in your country can contribute to the sustainability of the EJP RD elements? (if needed please indicate the country while answering in the posts-its)***

Additional points discussed:

- About adding new funders to IRDiRC : Membership in IRDiRC requires to respect some rules on funding Rare Diseases research :USD 10 Million over 5 years; an organism/institution should prove that it is spending USD 10 Million in the Rare Diseases program.
- About sustainability of IRDiRC : should IRDIRC funders have a share of their investment dedicated to the sustainability of the IRDiRC scientific secretariat as a mean to sustain it?
- (EJP) RD Central Helpdesk: one of the potential possibilities to sustain the Helpdesk would be having it as an EC Central service (as for the European IP Helpdesk)
    - This should be discussed at the EC level.
- Central services (such as the helpdesk) may consider having fees applied to process some requests that would need to remain confidential.
- The sustainability plan must consider a specific element: the different capacities of different countries: in terms of innovation, in terms of translation, in terms of trainings, etc. because the aim of EJP RD is to address the fragmentation of the national European level.

Question 2: ***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?)***

Additional points discussed:

- The Dutch Research Board is considering the service proposed by the FAIRification stewards as very valuable. So when the EJP RD ends, some Dutch FAIRification stewards should be sustained and used for other organisations : a scaling model that is currently being adopted.
    - The 'Train the trainers' approach could be linked with the FAIRification stewards for sustainability: as the Netherlands has highlighted as valuable service the work of the FAIRification stewards, we could amplify it with the 'train the trainers model' to increase the number of RDs specific stewardships in different countries.

2nd Exercise: VP National resources

Question: ***do you have any national resource that would connect to the Virtual Platform?***

- ***How is this national resource supported/sustained at the national/regional level?***

No further additional points. Were discussed apart from the MIRO inputs

Annexes

Annex 1 – Slides presented during the EJP RD Executive Committee meeting

- *See attached document: "Annex1_20220705_EJPRD_ExCom-Meeting_slides"*

Annex 2 – Slides presented during the EJP RD Policy Board and Governing Board meeting

- *See attached document: "Annex2_20220706_ PB-GB-Meeting_Slides"*

Annex 3 – ExCom meeting MIRO _ Addressing delays in tasks

- View link: https://miro.com/app/board/uXjVOonZYr4=/?share_link_id=172865242533
- *See attached document as well: "Annex3_20220705_EJPRD_ExCom-Meeting_MIRO_Delays in Tasks"*

Annex 4 – ExCom meeting MIRO _ Sustainability Roadmap for EJP RD

- View link: https://miro.com/app/board/uXjVOo9QeMI=/?share_link_id=582244008581
- *See attached document as well: "Annex4_20220705_EJPRD_ExCom-Meeting_MIRO_Sustainability"*

Annex 5 – Policy Board/Governing Board meeting MIRO _ Rare Diseases Partnership feedback

- View link: https://miro.com/app/board/uXjVOnjU8yE=/?share_link_id=618102436199
- *See attached document as well: "Annex5_20220706_EJPRD_PB-GB-Meeting_MIRO_RDP Feedback"*

Annex 6 – Policy Board/Governing Board meeting MIRO _ Sustainability Roadmap for EJP RD

- View link: https://miro.com/app/board/uXjVOoUOpFQ=/?share_link_id=24315272785
- *See attached document as well: "Annex6_20220706_EJPRD_PB-GB-Meeting_MIRO_Sustainability"*