EJP RD
European Joint Programme on Rare Diseases

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

Grant agreement number 825575

Del 1.5
Third report from the face-to-face ExCom and Policy Board meeting

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

Due date of deliverable: month 31

Dissemination level:
Public

Table of Contents

List of Abbreviations

AOB	Any Other Business
AWP	Annual Work Plan
C-Path	Critical Path Institute
Coo	[EJP RD] Coordination [Team]
EC	European Commission
EHR	Electronic Health Record
EJP RD	European Joint Programme on Rare Diseases
EOSC	European Open Science Cloud
ERN(s)	European Reference Network(s)
ExCom	Executive Committee
GA	General Assembly
GB	Governing Board
IMI	Innovative Medicines Initiative
IRB	Institutional Review Boards
JLA	James Lind Alliance
JTCs	Joint Transnational Calls
KPI	Key Performance Indicator
KRI	Key Result Indicator
LTP	Linked Third Party
MOOC	Massive Open Online Course
MS	Member States
NCATS	National Center for Advancing Translational Sciences

NORD	National Organisation of Rare Disorders
NSS	Networking Support Scheme
NSS	Networking Support Scheme
OD	Orphan Drugs
P0	Pillar 0
P1	Pillar 1
P2	Pillar 2
P3	Pillar 3
P4	Pillar 4
PAO	Patient Advocacy Organisations
PB	Policy Board
PCO	Patient Centered Outcome
PM	Person Month
PPP	Public-Private Partnership
RD	Rare Disease(s)
RDCA-DAP	Rare Disease Cures Accelerator-Data and Analytics Platform
RDCRN	Rare Diseases Clinical Research Network
RWD	Real-World Data
RWE	Real-World Evidence
VP	Virtual Platform
WP	Work Package

EJP RD Executive Committee

6th of July 2021

9:30 – 13:00

Online

Attached document:
Slides presented during the meeting (ppt presentation) – **Annex1**

List of participants

Name Surname	Institution	Role	Presence
Daria Julkowska	INSERM	coordinator WP1 - WP5	Present
Ralph Schuster	DLR	P1 coleader WP6	Present
Sonja van Weely	ZonMw	P1 coleader WP7	Present
Ana Rath	INSERM (Orphanet)	P2 coleader WP10 -WP11	Present
Franz Schaefer	UKL-HD	P2 coleader WP13	Excused
Virginie Bros-Facer	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

Domenica Taruscio	ISS	WP2 coleader	Present
Viviana Giannuzzi	FGB	WP4	Present
Annalisa Landi	FGB	WP4	Present
Elena Beltrami	FTELE	WP4 - WP19	Present
Barbara Sanavio	FTELE	WP4 - WP19	Present
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
Roseline Favresse	FFRD	WP16	Present
Holm Graessner	EKUT	WP17	Present
Krystyna Chrzanowska	IPCZD	WP18	Present
Maurizio Scarpa	HSK	WP20	-
Ralf-Dieter Hilgers	UKA	WP20	Present
Tanja Bülow	UKA	WP20	Present
Alberto Pereira		ERN research group coo	-
Catherine Nguyen	INSERM	IT GGB director	-
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
Alexander Parry	INSERM	Coo team	Present
Tanguy Onakoy	INSERM	Coo team - WP5	Present

Agenda

9:30 – 10h25	**AWP Y4 budget** • Pillar 0 (10 min: presentation [Blandine] + discussion [All]) • Pillar 1 (10 min: presentation [Blandine] + discussion [All]) • Pillar 3 (10 min: presentation [Blandine] + discussion [All]) • Pillar 4 (10 min: presentation [Blandine] + discussion [All]) • Pillar 2 (10 min: presentation [Blandine] + discussion [All])
10h25 – 10h35	***Break***
10h35 – 11h30	**Discussion on the extension of the EJP RD** (Administrative constraints to take into account; risks of non-extension; how long; which activities) • Presentation [Daria / Blandine] (10min) • General discussion [All] (45min)
11h30 – 11h40	***Break***
11h40 – 12h15	**C-Path collaboration update and ways forward** • Presentation [Coo Team + partners involved in C-Path collaboration] (10min) • General discussion (25min)
12h15 – 12h30	**AOB** • Contribution to the European Open Science Cloud (EOSC) • Other?

Minutes

AWP Y4 budget

See slides 1-20 for complete information.

Additional points of discussion

- The majority of the budget transfer showed in the presentation are reallocation of budget in the same Pillar. The budget leftovers from each Pillar and additional need are showed in the slide 27.

Pillar 0

See slides 2-5 for complete information

Pillar 1

See slides 6-8 for complete information

Pillar 3

See slides 9-12 for complete information

Additional points of discussion

- The discussion for WP14 budget is still ongoing.
- If a partner declares more Persons Month (PM) than what was foreseen in the corresponding Annual Work Plan (AWP), Coo has to accept it (based on the timesheet and report of costs). However, some of the PM declared can be as in-kind, not reimbursed. In particular, in P3, activities are reimbursed at 80-90%, thus some in-kind is needed for all activities. However, we should not be in a situation where a partner spends in 2 years all of planned PM for 5 years and not able to continue the planned work because of this.
- Some numbers for budget transfer need to be confirmed by EURORDIS with financial officer.
- Virginie Bros-Facer informed the ExCom about a recent decision of the European Commission (EC) to cancel operating grant from which EURORDIS benefited for several years. From the end of this year, substantial funding will thus not exist for EURORDIS. This funding was used in particular to cover some EURORDIS core activities, such as the EURORDIS Summer School for which only a small percentage of the costs was assigned to EJP RD. To cover the costs of the Summer School 2022, could some EJP RD unused budget be used? The expected cost of such face-to-face event would be 40-50k€ in total, with 35-40k€ needed.
- An additional budget (25k€ max) to provide a series of motion design videos for the WP16 MOOC on diagnosis to create pedagogical videos to be included in the MOOC and available on other types of media should be added.
- In WP16, some discussion with ELIXIR Linked Third Party (LTP) is needed as they are not willing anymore to be involved in WP16 but would like to keep EJP RD associated funding for other activities. In case the LTP is not willing to do these WP16 activities, then the associated budget should be transferred to another EJP RD partners willing to do this activity.

Pillar 4
See slides 13-15 for complete information.

Additional points of discussion

- The institution in charge of the organisation of the "Mini-symposium for the Demonstration and Innovation projects" should be defined. Coo could keep the budget as central budget and transfer it to the organisation in charge once decided. In any case, the organiser of the symposium should be involved in all discussion around the organisation of this event.
- No budget transfer needed in WP19.

Pillar 2
See slides 16-19 for complete information.

Additional points of discussion

- Ana will send information for WP11 allocation to Blandine: 1PM from central budget for new set of resources foreseen
- A decision is needed on where the unspent budget to be centralised should be kept to make it easy to allocate it to events/activities. It is needed to be agile with this budget.
    - Budget could be centralised at the Coo level (Central budget line) and allocated to partners depending on who will do what. Only an estimation of the budget to be spent in following year is needed for the AWPs. At the end, if the amount planned was not exact, it can be adjusted. The centralised funding means that the budget is for the workshops regardless of the institution that ends up organising the workshop.
    - Also, budget could be kept at the level of Work Package leader budget, as initially foreseen and it will be up to the WP Leader to pay the bills related to the workshops. Attention: **no invoices between partners are allowed.**

  We want to avoid, as much as possible, redistribution of budget from one institution to another. Working in a system where budget lines have to be transferred from one partner to another for each organised workshop would make it very difficult for the financial reporting at the end of each year.

  A possibility could be to have the 120 000€ kept at the Coo level as Central budget to be allocated + 40k€ left at the level of WPL budget to organise the workshops.
- Question from EBI on budget needed on some activities will be discussed in a TC already planned with Carl.

ACTIONS

⇨ All WP Leaders and Pillar Leaders should confirm final numbers and send final needed information to Blandine Castrillo by July 15th the latest.

Discussion on the extension of the EJP RD

(Administrative constraints to take into account; risks of non-extension; how long; which activities)

See slides 21-27 for complete information

Additional points of discussion:

- It has to be kept in mind that in any case, there will be an overlap between the new Rare Diseases Partnership (to start in 2024) and a possible extension of the EJP RD.
- It should not be considered that for EC, asking for an extension means extending the whole project. Some next phase activity should be planned in the RD partnership rather than be extended in the EJP RD.

- Extension request would be sent to EC somewhere in Year 4. Justification/information to provide for the extension would be updated at the time the extension would be requested.
- Other activities, such as services developed by P2 could also be highlighted in the presentation. If we can demonstrate by 8-12 months that some services provided by P2 are directly connected to research projects currently funded within Pillar 1, those activities/connection can be included when the extension will be requested to the EC. It is important to have difference between services necessary for projects to be achieved and activities to be developed in the Partnership. If things are also implemented as service in the Partnership, nothing prevents the funded projects to use service in the RD Partnership.
- Some delay in P2/P4 collaboration due to COVID-19.
- There has been an impact of COVID also on Innovation projects (P4), only 2 projects have been funded. Some important topics identified by experts are not covered in those projects. WP20 would like to launch "speed projects", allowing to go directly to specific identified experts with a specific question/topic to develop a project answering that question. This would be a quicker process, as the calls are taking too much time and the drug development world is fast changing ➔ to be discussed. Would be very difficult at this time to organise, with no guarantee that projects will be finished on time, we should not launch new call for projects at this stage.
- Discussion on the content of the RD partnership is ongoing. In the next 8-12 months, we will have more information. In Q1 of 2022, we will have to deliver the first draft of the RD Partnership. RD Partnership discussion are not driven by EJP RD partners, other stakeholders are also involved. Two meetings have been organised so far by the EC with Member States (MS), which were mainly informative meetings. In the last meeting, EC asked about the composition of the writing group: MS will nominate people to participate in the writing groups.
- The EJP RD Coo propose to organise a F2F meeting of the ExCom in December 2021 to discuss the RD Partnership.
- It is important in the coming months to see which activities will be part of the RD Partnership, what should be finalised within EJP RD, what would request extension to be finalised in the EJP RD.
- It will be important to highlight activities that will be included in the RD Partnership (e.g., on data management, data quality, data discovery) are also important and needed, even if not in the extension.
- The P2 plan was to build a basic Virtual Platform (VP) in 5 years, so it will not be ready to use in 2.5 year. Also, COVID-19 slowed things done: the VP might not have been achieved as planned in 5 years, would need to extend for 6 months to be sure that will be finalised by the end of the EJP RD project.
- As the financing of the whole EJP RD is depending on the funding agencies and funding of P1 projects, there is a need for 1 year extension.
- Networking Support Scheme (NSS) activities (P1) have also been impacted by the COVID-19, it would be good to consider including it also in the extension.

- The justification of the extension to the EC should not be based on the non-spending of the budget by the end of the 5 years. It will be difficult to obtain a 1-year extension, but EC is willing to help us and will give us advice on what should be in the extension request and how.

ACTIONS

- ⇨ It is clear that a 1-year extension is needed for P1 funded projects. It should be discussed with EC to include also in the extension, 6-month extension for all other activities that have been also impacted by COVID-19 and need some extra months to finalise the activities planned in the EJP RD.
- ⇨ In the coming months, the content of the RD Partnership will become clearer. Coo will keep the ExCom informed on advancement of the discussion. A dedicated ExCom meeting will be organised F2F in December 2021 to discuss the RD Partnership.
- ⇨ The justification/content of the extension will be updated in 8-12 months, when the request will be drafted to be submitted to the EC.
- ⇨ The proposition of extension will be presented for validation at the General Assembly this year.

C-Path collaboration update and ways forward

EJP RD and C-Path are collaborating on multiple projects since the approval of the collaboration by the General Assembly in 2020. Since then, the Directorate of C-Path recently changed. A meeting with the new director took place at the end of May 2021. At this occasion, EJP RD partners learned that C-Path decided to expand largely their strategy in Europe. They moved to Amsterdam and hired people from Europe to reinforce their capacity to operate in Europe. For the last 2 years, they have created a centralised platform, centralising the data using specific ontologies; the data are available for research, also services to access data in a secured manner are available and it is possible to do research within a secured environment. They now target Europe and want to integrate EU resources, understand complexity of EU landscape and to initiate discussions with EU stakeholders.
Such a quick expansion in EU was not expected. The question to EJP RD is now: how do we do with this collaboration? What should we propose to them?

Discussion:

- Opportunity to develop a hybrid model: Federated model is the goal and motivation for our collaboration, Centralised model goes faster for certain purposes (e.g., clinical trials). We should strengthen the partnership to make them part of the ecosystem.

- There is a need to know more what they are offering, to be able to compare things that are comparable. Also, sustainability and cost efficiency should be looked at. If they truly have a better platform, we should, on behalf of the RD community, go with them. But there is likely to be significant differences. We need to understand more what they are offering.
- Who does the harmonisation? EJP RD strategy is that you should be the one making your data interoperable, not paying a company and becoming dependant of this company.
- There is a significant opportunity to collaborate and make the platforms interoperable, but we should not become dependent.

ACTIONS

- ⇨ There is a general agreement to explore the opportunities for collaboration within the General Assembly dedicated EJP RD / C-Path session.
- ⇨ Coo will send an email to ExCom to collect who would be interested to have a preparatory call with C-Path in advance of the General Assembly (GA) [around end of August]

AOB

Contribution to EOSC

One of the impacts identified for the EJP RD is the contribution to the European Open Science cloud.

Discussion:

- VP is EOSC compliant, ready to be integrated.

EJP RD Policy Board and Governing Board meeting

7th of July 2021
13:00 – 18:00
Online

Attached document:
Slides presented during the meeting (PowerPoint presentation) – **Annex 2**

List of participants

Name		Board	Country/ [organisation]
Alessandra	Renieri	PB	Italy
Alexander	Parry	Coo	France
Alysha	Croker	PB	Canada
Amanda	Borens	Speaker	USA
Ana	Rath	ExCom	France
Anabela	Isidro	GB	Portugal
Andrea	Corazza	Invited	Belgium
Aniket	Sharma	Coo	France
Annalisa	Landi	ExCom	Italy
Anthony	Brookes	GB / ExCom	Great Britain
Anton	Ussi	ExCom	The Netherlands
Armelle	Degeorges	PB	France
Avi	Israeli	PB	Israel
Bertrand	Schwartz	PB	France
Birute	Tumiene	ExCom	Lithuania
Blandine	Castrillo	Coo	France
Catherine	Nguyen	ExCom	France
Chris	Evelo	ExCom	Netherlands
Christina	Kyriakopoulou	EC	[EC]
Christine	Fetro	ExCom	France
Claudio	Carta	ExCom	Italy
Daria	Julkowska	Coo	France
Domenica	Taruscio	ExCom	Italy
Étienne	Richer	GB	Canada
Eva	Bermejo-Sanchez	ExCom	Spain
Florence	Guillot	Excom	France
Florence	Quist	GB	Belgium
Friederike	Ehrhart	ExCom	The Netherlands

Galliano	Zanello	Coo	France
Günter	Schreier	GB	Austria
Hélène	Le Borgne	PB	[EC]
Ingeborg	Barisic	PB	Hungary
Irit	Allon	ExCom	Israel
Jale	Sahin	PB / GB	Turkey
Jordi	Llinares Garcia	PB	[EMA]
Jose	Valverde	PB	[EC]
Judita	Klosaková	PB	Czech Republic
Juliane	Halftermeyer	Coo	France
Krystyna	Chrzanowska	ExCom	Poland
Leo	Schultze Kool	GB	The Netherlands
Liron	Even-Faitelson	ExCom	Israel
Lucia	Monaco	PB	[IRDiRC]
Maciej	Gajewski	Speaker	[Alexion]
Manuel	Posada	ExCom	Spain
Marco	Roos	ExCom	The Netherlands
Mari	Teesalu	PB	Estonia
Myriam	Cevallos	PB	Switzerland
Patrícia	Maciel	PB	Portugal
Pierre	Meulien	PB	[IMI]
Ralf-Dieter	Hilgers	ExCom	Germany
Ralph	Schuster	GB / ExCom	Germany
Rima	Nabbout	ExCom	France
Roseline	Favresse	ExCom	France
Sean	Sapcariu	GB	Luxembourg
Simon	Bennett	Speaker	Belgium
Sonja	van Weely	ExCom	The Netherlands
Tanguy	Onakoy	Coo	France
Theda	Wessel	PB	Germany
Tiina	Urv	Speaker	USA
Valentina	Bottarelli	PB	EURORDIS
Virginie	Bros-facer	GB / ExCom	EURORDIS
Vittoria	Carraro	Speaker	[EUCOPE]
Viviana	Giannuzzi	ExCom	Italy
Yanis	Mimouni	Coo	France
Živilė	Ruželė	GB	Lithuania

Agenda

13:00 – 13:25	**Welcome word and introduction to the EJP RD**	Daria Julkowska (INSERM) Coo
13:25 – 13:30	**Welcome of new members**	All
13:30 – 14:00	**Summary of EJP RD activities, achievements and impact in Year 1 to 3**	Daria Julkowska (INSERM) Coo
14:00 – 15:00	**Annual Work Plan Year 4 – Feedback from the Boards**	All
15:00 – 15:20	**Coffee break**	
How could collaborations with industry for Rare Diseases be implemented?		Presentations: 1h30 Discussion: 1h00
15:20 – 15:35	Introduction: How does industry collaborate with academia (translational and clinical aspects)?	Anton Ussi (EATRIS), Simon Bennett (Biogen)
15:35 – 15:50	Challenges and opportunities of the Rare Diseases Research Challenges of the EJP RD	Christine Fetro, FFRD
15:50 – 16:20	Proposals on how to improve the R&D ecosystem for basic research and company take-up of development by the European Expert Group on Orphan Drug Incentives	OD Experts Group members: Vittoria Carraro, EUCOPE & Maciej Gajewski, Alexion
16:20 – 16:35	Rare Diseases Clinical Research Networks – boosting RD therapy development and clinical trials in public-private setting	Tiina Urv, NCATS
16:35 – 16:50	Critical Path Institute RDCA-DAP experience with private stakeholders	Amanda Borens, CPATH
16:50 – 17:50	Round table and general discussion	All participants
17:50 – 18:00	**Summary and next steps**	Daria Julkowska (INSERM) Coo

Minutes

Welcome of new members

- **Alysha Croker – Canada**: Manager of the office for paediatric Patients involvement at "Health Canada"
- **Válter Fonseca – Portugal**: New representative from Portugal Ministry of Health; represented by Carla Pereira
- **Myriam Cevallos – Switzerland**: State Secretariat for Education Research & Innovation representative
- **Gyorgy Pfliegler – Hungary**: Ministry of Health representative
- **European Commission representatives:**
    - Changes in DG RTD: new representative will replace Catherine Berens
        - The RD team remains with **Helene Le Borgne** and **Christina Kyriakopoulou** (team leader)
    - Jose Valverde in DG Santé (unit B3) fully engaged with the European Reference Networks (ERNs)

Summary of EJP RD activities, achievements and impact in Year 1 to 3

See slides 2 to 28

Discussion

- EJP RD has many Key Performance Indicators (KPIs) being monitored in the work plan, do we have a higher level of measurements that are matched with the impacts presented?; and how are they linked with the impact?
- Regarding the recommendation of the EJP RD interim-review on the more visibility of impact indicators, is it a communication issue or missing item reflecting this high-level indicator?
    - The two questions are linked together, EJP RD has 93 Key Result Indicators (KRIs) and 40 KPIs that are set at the operational level for each Work Package. There is, to date, no direct connection with the overarching impacts.
    - Some indicators are too granular; this means that there is an opportunity to identify other (missing & overarching) indicators. The work of the EC Expert Group will be helpful.
    - The recommendation of the experts is also on the improvement of the dissemination and communication.

- This need for monitoring may be considered daunting because of the expected amount of work it requires, that might hinder planning and

operational flexibility. Also, the real value would come from the emphasis on the real-world impact.

- It seems that EJP RD is setting the question and report on its own progress. Is there a scope to increase the external monitoring (i.e., having external stakeholders perform the monitoring)?
    - The new (monitoring) framework is going towards less and better indicators. The indicators linked to key impact pathways are measured anyway (less reporting).
    - For the partnerships: many indicators are linked to the "external" additionality of initiatives.
    - Regarding the revision of the whole monitoring framework: EJP RD defined its own KPIs/KRIs. The monitoring is performed by a partner not involved in the work monitored. The updated monitoring framework should be endorsed by the stakeholders.

Annual Work Plan Year 4 – Feedback from the Boards

See slides 29 to 45

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

- **Taking into account your overall knowledge of EJP RD and AWP Y4: what is missing in AWP Y4?**
    - No feedback was provided.

- **How can we still better integrate EU-13 countries?**
    - A lot of areas within the EC instruments have issues with the integration/participation of EU-13 countries. This includes the Innovative Medicines Initiative (IMI) that identified the following aspects to consider:
        - **Pure awareness in these countries**: use every possible mechanism to have active and targeted outreach to those countries that should be participating because of their expertise and activity
        - **Is there a critical mass in a specific area in these countries**?
            - are their national/regional strategies that may align with RD?
            - are their National/regional funding that can be leveraged?
    - The EJP RD WP2 performed a survey to check, among others, the situation with the involvement/integration of EU-13 countries including the mutual alignment of EJP RD actions with their national plans (if any). An improvement was noticed when compared to the previous survey. These results will be presented

at a dedicated Strategic Meeting on July 8, with policy makers among other interested attendees.

- Many countries are without healthcare providers involved in ERNs. With the expansion of ERNs there will be new affiliated partners and new full partners, some of which are located in the EU-13 countries.
- For the Lithuanian Research Council:
  - The widening measures applied for the Joint Transnational Calls (JTCs) of EJP RD performed very well compared to other multinational calls managed in which LRC is participating. The involvement of EU-13 participants was successful, there is nothing to recommend for this Action.
  - Regarding the training activities, Lithuanian partners are aware of the programme and opportunities. There might be a need to improve communication, but for the moment Lithuanian partners are satisfied.
- → It might be interesting to show graphically the representation of different countries in the Networking Support Scheme and highlight the dynamic collaboration between them.
- The online format of meetings and events can leverage the involvement of EU-13 countries

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- **How do you present and get back to your national stakeholders with the key points of the EJP RD AWP Y4 ?**
  - No specific feedback was provided.

- **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.
  - EJP RD coordination is considering (depending on the resource availability) translating EJP RD website and specific dissemination campaigns to EU national languages.

- **Taking into account EJP RD developments in previous years and year 4, how would you take them to promote better data structuring and standardisation in your countries (apart from the connection to the VP)?**
  - No specific feedback was provided.

Questions from the EJP RD policy Board and Governing Board and answers provided

Theda Wessel (DE): Is there any activity allowing to figure out what are the most pressing questions for patients (taking into consideration the limited resources). For example, the James Lind Alliance (JLA)[1] brings together clinicians, patients and carers to discuss research priorities. Is there a structured way to finding the most relevant questions for patients?

- There is a nice chain of activities set up in EJP RD for this purpose:
  - the support to Networking Events that link *patients* and *researchers*; the former expressing their *needs* and the latter identifying *priorities*. The Networking Support Scheme addresses this first need of linking people together;
  - In the JTC patients' input is encouraged from the start to define the research questions. EJP RD developed a short guide for Patients Partnerships in rare diseases research projects with concrete examples to develop research projects together; funding of PAO is provided within EJP RD JTCs;
  - Training on scientific innovation and translational research is provided. A comprehensive presentation of RD research is performed with different examples on how to engage with researchers. Patients and researchers are also invited to share their experiences and illustrate success stories;
  - Even in the field of very sophisticated methodologies, Patient Advocacy Organisations (PAO) are in the task force leading this work, and patients are represented in the discussions on prioritisation of innovation call topics. This highlighted major topics: patient centred outcomes (PCO); patients registries and natural history studies;
  - These patients' participations are not only encouraged, but also financed. Patients' organisations are considered as full member in the research projects.
- There are a lot of commonalities in "patients' involvement" and "input" that need to be taken into account. They include "understanding patients' disease", "a better diagnosis", "drug development and repurposing" and "environmental factors ".

[1] The James Lind Alliance (JLA) is a non-profit making initiative established in 2004. It brings patients, carers and clinicians together in Priority Setting Partnerships (PSPs) to identify and prioritise the Top 10 unanswered questions or evidence uncertainties that they agree are the most important. The aim of this is to make sure that health research funders are aware of the issues that matter most to the people who need to use the research in their everyday lives.

Requests addressed to the EJP RD Policy Board and Governing Board

- **EJP RD needs to reach out to medical and PhD students of EU-13 countries for the Massive Open Online Course (MOOC) "Diagnosing Rare Diseases: from the Clinic to Research and back". There is a need for recommendation on some networks and medical associations.**
- **The Policy Board was asked to share, if possible, any opportunity that arouses from the fast changes linked to the COVID-19 pandemic.**

ACTIONS

- ⇨ **The EC Expert Group will help** EJP RD consortium to identify other (missing) monitoring indicators (matched with the EJP RD impacts).
- ⇨ **EJP RD is suggested to** show graphically the representation of different countries in the Networking Support Scheme and highlight the dynamic collaboration between them.
- ⇨ **EJP RD coordination** needs to finish the evaluation (depending on the resource availability) for translating EJP RD website and specific dissemination campaigns to EU national languages.
- ⇨ **The Policy Board help is requested** to reach out to medical and PhD students of EU 13 countries for the MOOC "Diagnosing Rare Diseases: from the Clinic to Research and back". There is a need for recommendation on some networks and medical associations.
- ⇨ **The Policy Board is asked** to share, if possible, any opportunity that arouses from the fast changes linked to the COVID-19 pandemic.

How could collaborations with industry for Rare Diseases be implemented?

Introduction: How does industry collaborate with academia (translational and clinical aspects)?

See slides 46 to 54

Challenges and opportunities of the Rare Diseases Research Challenges of the EJP RD

See slides 55 to 68

Discussion

Studies have shown that new products/innovations taken forward as spinouts only ever succeed after many rounds of refocusing and at least several dozen millions of (money) investment. There is a need for industry partners with considerable financial power; patience/tolerance for how long it [research] will take and experience to guide academia. Industry partners might rightfully be very picky over what ideas to run with.
The Public Private Partnerships seem far easier in the US, where investment and risk tolerance are much more available.

Proposals on how to improve the R&D ecosystem for basic research and company take-up of development by the European Expert Group on Orphan Drug Incentives

See slides 69 to 76

Discussion

Did the Working Expert Group address drug repurposing?

- The group tried to avoid cutting the problem into different product types. There are a lot of discussions on that. It was decided not to look at a specific development model.
- When looking at regulatory recommendation and specifically recommendation on orphan drugs, a need that is tangential to the repurposing topic was highlighted: "off label use".

Regarding the Public-Private Partnership (PPP) fund, why is there a need to establish a different funding mechanism when there is already the Innovative Medicines Initiative (IMI), for example, where all the players are involved?

- The IMI priorities are not specifically focused on RD. It is hard to compete with big population health risks and big areas that requires public attention. This is why establishing a specific mechanism focusing on RD can have more success for RD community, and is in fact a need.

Rare Diseases Clinical Research Networks – boosting RD therapy development and clinical trials in public-private setting

See slides 77 to 92

Critical Path Institute RDCA-DAP experience with private stakeholders

See slides 93 to 101

Round table and general discussion

The quality of registries is fundamental, would the next generation of registries be with quantitative data?

- It was deceiving for C-Path to find that the quality of data collected for years in registries often are not consistent and with a quality not allowing for therapy development.
- What helps is to have the regulatory agencies pushing for the application of data standards. C-Path is publishing a paper together with NORD (National Organisation of Rare Disorders) to guide on the tools to use to build patients' registries
  - → EJP RD will liaise with C-Path on this topic.

Regarding the Orphan Drugs (OD) expert group, every stakeholder is expected to participate to the OD regulation; which relationship do you see between the revision of the regulation and PPP?

- **Should researchers work with all the other stakeholders to define specific objectives for unmet needs?**
- **Which step of the R&D process would benefit more from PPP?:**
  - **sharing data?**
  - **looking for study sites?**
  - **comply with local rules or different medicines agencies?**

    - How a multistakeholder approach should be used for the definition of unmet need is a paramount, it should take into consideration different views (patients, researchers, regulators, health technology assessors, etc.)
    - To take into account the criterion "prioritisation", there is a need for a product that will be used, then the question on "how will there be a weighting" or "a common denominator for all the stakeholders" arise.
    - It was found that in the areas of ultra-RD, when PPP interact with regulation to try to impact unmet needs the issue faced is not about market exclusivity. It is where collaboration is needed the most. It is need everywhere; thus, it is not possible to prioritise.
    - The more people from different areas are gathered the more mutual understanding is reached.

Regarding the PPP commonalities in the presentations, there are some kind of centralisations to address PPP. The EU has a hub to accelerate addressing the need of therapy development. Tiina's presentation is a centralised option. What we are doing in EJP RD as a whole is also getting ready for faster therapy development.

→ We need to share experience with the Rare Diseases Clinical Research Network (RDCRN) & C-Path, mutualise ideas and also look at what is needed to be put in place to make a PPP come to a reality.

US colleagues are trying to use data from Electronic Health Records (EHR). How is it envisioned and how RD data from this resource is recognised? [Capturing data not coming from registries is a major topic especially for regulatory compliance on RD data].

Also, using different systems in C-Path and RDCRN; how both are collaborating to increase the critical mass of RD data?

- For regulatory grade data, the experience with FDA when it comes to unmet needs including RD, focuses on working in full provenance: every piece of data coming in raw format is accessible and all codes are checked in public repository: it is possible to reproduce the process from raw data.
- For EHR data: this is really an open question to the world. The question remains unsolved since various systems are used. Considerations on using Artificial Intelligence to solve this are ongoing.
    - Outside from RD world, in neonatal area, C-Path is trying to address the boundaries and Quality assessment tools for using EHR data.
- C-path worked with the National Center for Advancing Translational Sciences (NCATS) on multiple consortium efforts. Both data coordinating centres are working together to make sure that interoperability is there for any development.
- RDCA-DAP is not a centralised repository, it has APIs to connect to other platforms. A hub is located in Amsterdam, thus allowing to address GDPR required in EU.
    - Defining procedure and protocols to make data semantically interoperability is at the exploration phase with EJP RD.
    - C-Path developed APIs to collect data from any registry uploading it. It is moving towards a federated access to build an ecosystem and not a centralised repository.

For the use of Electronic Health Record to collect Real-World Evidence (RWE), how can we advance in this space?

- For RWE, it is important to have a specific policy framework, the translation of Real-World Data (RWD) into RWE is not a trivial task, and there is a learning by doing curve.
- There is a need for more regulatory guidance on how RWD can be used. C-Path is trying to provide FDA with suggestions; e.g., for the case of neonates where physicians are trying to guess for treatment (and dosing), EHR are used to look at lab values and detect adverse events. There is a need to interact with regulatory agencies.
    - The political perspective is now evolving, acceptance is increasing as RWE is seen as complementary (by regulators and HTA bodies) especially in areas of scarce data (such as RD). For example, this year, blinded case-studies are being performed and are considered by regulators.
- The RDCRN in US and ERNs in EU are among the best positioned stakeholders to lead the work on RWE.

All the software should be open, within a list of industrial/commercial collaborators there are software providers that do not share their code. How to make sure that the code used is correct?

- One experience from C-Path leads to building a software code used for quantitative medicine (NONlinear Mixed Effects Modelling) on R and shares this code.
- RDCRN is following the same model: having everything open source and available.

Rigor in setting up everything is needed for PPP to make data regulatory compliant. There should be a question if the process (study design, etc.) is designed for confirmatory research. The PPP enables academia to get advantage from industry experience involving rigor and specific tools.

GDPR hinders the developments of EHR use in this area. There is a need for a higher-level policy discussion involving patients' representatives.

In US, the Institutional Review Boards (IRB) are often viewed as paternalistic when it comes to the re-use of HER for research purposes.

It seems that the technical challenges of plugins entities/technologies together can be addressed. There are compatibilities between EJP RD & C-Path.

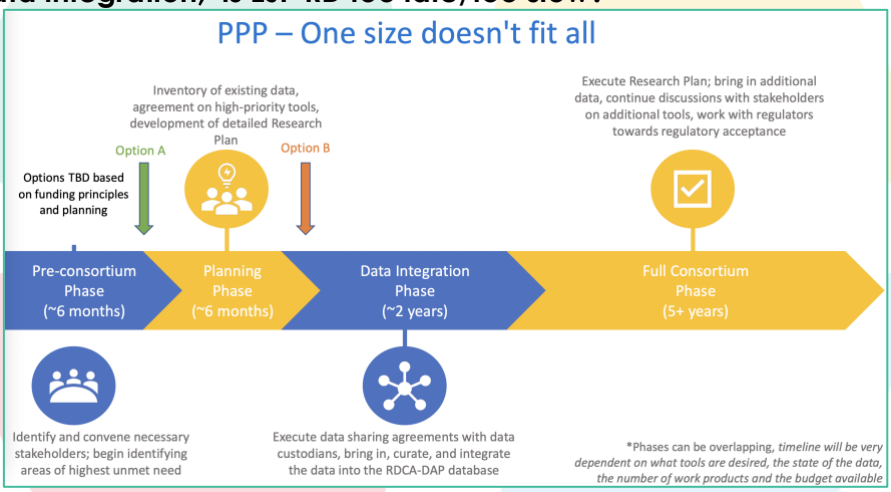
- **The challenge below, that is on the data side (regulatory grade data). Before getting hands on data there is a need to have an objective. How C-Path & RDCRN address the question of data suitability before getting hands on it?**
- **Above, the technology question there is the ELSI aspects. Where are you in addressing ELSI issues?**
    - The bigger challenge is the human challenge: "what happens to the data before you get it". At C-Path, there is a 2-years process to gather data with a diminishing return because of missingness (in data capture or in several of the studies selected).
    - The biggest challenge is to get custodian sharing "all" the data. Sometimes GDPR provisions allow custodians to remove some data that will lack for the research purpose.
        - To overcome this situation, conversations with medical ethicists and regulators are engaged (patients enter trials at great sacrifice so that others will benefit). Companies and Academics can be quite conservative in sharing data.
    - Not all the data gathered is valuable for research. Academic data is academic data (often lacking the needed quality). There is a need to meet standards as there is no replacement for rigorously collected data (garbage in = garbage out).
    - Regarding the prospective collection of data: C-Path is not running trials; it is partnering with stakeholders collecting it and prescribing best practices on what and how to collect data. Based on this process, C-Path observes better collected data.
    - Education is key, especially when working with PAO with less money and lot of passion spending huge efforts in collecting data.

- Related publication: "*Share and protect our health data: an evidence-based approach to rare disease patients' perspectives on data sharing and data protection - quantitative survey and recommendations*"

In the presentations there was a mention on the importance of having interactive and iterative process between industry and academia. How can we set up in a manageable scale this kind of collaboration (at global scale) in a regulated setting?

- There is a need to define long-term desirable achievements involving the research funders and policy makers to set up non-heavy programmes equally targeted. Creating structured framework for interactions with an end-to-end view with life-cycle management.
- Education is key not only for patients but also for academic researchers.
  - ERN interactions within EJP RD bring stakeholders close.

It takes to C-Path 6 months for pre-consortium discussion, 6 months for planning and 2 years for data integration; is EJP RD too late/too slow?



- The RD ecosystem is more complex than a single therapeutic area. The graphic (above) is eliciting a focused therapeutic area (e.g., Parkinson disease and Duchenne muscular dystrophy)
- We try to take the generalised framework and process and repeat them for other areas; this may be a lot longer that a 5-year plan.

ACTIONS

- ⇨ EJP RD will liaise with C-Path for the topics on the application and promotion of data standards use.
- ⇨ EJP RD needs to share experience with the Rare Diseases Clinical Research Network (RDCRN) and C-Path, mutualise ideas and also look at what is needed to be put in place to make a PPP come to a reality.

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

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