

## **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 1.4**

# **Second 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 19

**Dissemination level:**

Public

# EJP RD Executive Committee

**7<sup>th</sup> of July 2020**

**9:50 – 16:00**

Online

**Attached document:**

Slides presented during the meeting (ppt presentation) – Annex1

**List of participants:**

<b>Name Surname</b>	<b>Institution</b>	<b>Role</b>	<b>Presence</b>
Ana Rath	INSERM (Orphanet)	P2 co-leader WP10 -11	Present
Anthony Brookes	ULEIC	WPL 10 - 12	Present
Anton Ussi	EATRIS	P4 co-leader WPL 3 - 19	Present
Birutė Tumiene	VUHSK	P3 co-leader WPL 18	Present
Catherine Nguyen	INSERM	IT GGB director	Present
Chris Evelo	UM	WPL 13	Present
Christine Fetro	FFRD	WPL 8	Present
Claudio Carta	ISS	WPL 14	Present
Daria Julkowska	INSERM	Coordinator WPL 1 - 5	Present
Domenica Taruscio	ISS	WPL 2	Present
Eva Bermejo-Sanchez	ISCIII	WPL 2 - 3	Present
Manuel Posada	ISCIII	WPL 2 - 3	Present
Franz Schaefer	UKL-HD	P2 co-leader WPL 13	Present
Holm Graessner	EKUT	WPL 17	Present
Irit Allon	CSO-MOH	WPL 9	Present
Krystyna Chrzanowska	IPCZD	WPL 18	Absent
Alberto Pereira	LUMC	Chair of ERN Research Group	Absent
Marco Roos	LUMC	WPL 12	Present
Maurizio Scarpa	HSK	WPL 20	Absent
Ralf-Dieter Hilgers	UKA	WPL 20	Present
Tanja Bülow	UKA	WPL 20	Present
Ralph Schuster	DLR	P1 co-leader WPL 6	Present
Rima Nabbout	AP-HP	P4 co-leader WPL 20	Present

Roseline Favresse	FFRD	WPL 16	Present
Daphné Jaoui	INSERM (RaDiCo)	WPL 3	Present
Sergi Beltran	CNAG-CRG	WPL 11	Present
Sonja van Weely	ZonMw	P1 co-leader WPL 7	Excused
Elena Beltrami	FTELE	WPL 4 - 19	Present
Virginie Bros-Facer	EURORDIS	P3 co-leader WPL 15 - 18	Present
Viviana Gianuzzi	FGB	WPL 4	Present
Annalisa Landi	FGB	WPL 4	Present
Bertrand Schwartz	MESRI	MESRI	Absent
Yanis Mimouni	INSERM	Coordination team	Present
Katerina Tzima	INSERM	Coordination team	Present
Blandine Castrillo	INSERM	Coordination team	Present
Galliano Zanello	INSERM	Coordination team	Present
Carla D'Angelo	INSERM	Coordination team	Present
Eleonora Passeri	INSERM	WPL 5	Present
Juliane Halftermeyer	IT	Coordination team	Present
Ben Lydall	EATRIS	WPL 3 - 19	Present
Joana Namorado	RHIZOME	Ethics Advisor	Present
Anne Demoisy	RHIZOME	Ethics Advisor	Present
Mary Wang	FTELE	[Speaker]	Present
Barbara Sanavio	FTELE		Present

**Agenda:**

<b>9:50 – 10:00</b>	Welcome from the coordination	
<b>10:00 – 11:00</b>	<b>Optimisation of the EJP RD work plan (AWP Y3 and following years):</b> <ul style="list-style-type: none"> <li>New activities foreseen</li> <li>Potential synergies (e.g. training activities)</li> <li>Actions related to COVID-19 mitigation measures (e.g. re-organisation of actions into online events, support required, etc.)</li> <li>How do we deal with planned activities that are not working as expected? (what are the mitigation measures implemented by different pillars/WPs, when do you decide that the GO/NO GO decision is necessary)</li> </ul>	All attendants
<b>11:00 – 11:15</b>	<b>Break</b>	
<b>11:15 – 12:15</b>	<b>Introduction to financial optimisation:</b> <ul style="list-style-type: none"> <li>Presentation of the update of the current status of the budget including the possible "savings" and changes to be implemented in year 3 (according to what was reported in AWPY3)</li> <li>Propositions on budget to consider by ExCom members in the finalization of the AWPY3</li> </ul>	All attendants
<b>12:15-13:15</b>	Continuation of discussion if necessary	
<b>13:15 – 14:00</b>	<b>Lunch</b>	

14:00 – 14:45	<b>Pillar 2 ERN &amp; RD Researchers surveys:</b> • "deep dive" and exploitation	Mary Wang (FTELE)
14:45 – 15:00	<b>Break</b>	
15:00 – 15:30	<b>Sustainability survey:</b> • results, • what was expected? • how to complete the following surveys?	Ben Lydall (EATRIS)
15:30 – 16:00	<b>EJP RD Ethics Advisor:</b> • presentation of our expert and interactions to come	Anne Demoisy

**Minutes:**

**Optimisation of the EJP RD work plan (AWP Y3 and following years):**

See slides 2-13 for complete information

[New activities foreseen](#)

See slides 2-7 for complete information

Any new activity (not planned in Annex 1 or activity starting now - not started in Y1 or Y2) will have impact on the budget → the budget has to be validated by the General Assembly (GA) through the validation of the AWPY3.

**Additional points of discussion**

- In the Pillar 2, no new activity but more a refinement of foreseen activities. There is one new activity, but is already under development in Year 2, which is the webpage displaying a MindMap to allow finding RD resources already integrated in the VP.
- There is a need to enable people to discover/look for experts as well (not part of the initial plan). This action would need coordination outside P2 as it is quite transversal. A pool of experts is already available through the Helpdesk and can be expanded beyond. This major need was also identified in Pillar 4 with a huge missing link with people in need for knowledge and methodologies. **A new activity, in link with the Central Helpdesk, should be developed.** Some person/month should be saved for this activity to help developing this facility for EJP RD. We should start from detailed mapping of "internal" EJP RD expertise and apply to it the tools that will allow finding experts. However, this can be further expanded since in many EJP RD activities, partners are having more and more connection to external experts whose roles should be well identified. Having such network of experts and having people coming to EJP RD to look for EJP RD expertise but also for our collaborators would add value for the sustainability of EJP RD as a network.
- Another important element (in relation to in house expertise) is the provision of dedicated training to spread knowledge (especially, expertise developed in P0, P2 and P4). These could be supported (possibly) through the WP18.

For novel RD training needs in Pillar 3 WP18 (activity to start in Y3), there is a huge lack of data on what is existing, the synergies with other pillars and their scientific content. A dialogue with other pillars will be established for detecting experts to implement training activities.

- Some additional deliverables have to be added in the WP18.

- Following the COVID-19 crisis, some trainings in P3 were successfully implemented online. In the future, there will be a need to identify trainings that could be done online and the ones that need a face-to-face. There could be a discussion with WP16 as some trainings could thus be developed as module on the e-learning platform.

**Actions:**

- ⇒ The new activity on the pool of experts in link with the Central Helpdesk should be discussed at an upcoming ExCom meeting
- ⇒ The new activity on the pool of experts in link with the Central Helpdesk has to be included in the Annual Work Plan Year 3: description of the activity + budget
- ⇒ Work between Clinical Trial Helpdesk and the Toolbox (WP19 and P2) on how to organise existing expertise in a toolbox for people to go and find answers but without exposing the whole problem

Potential synergies

See slides 8-9 for complete information

**Additional points of discussion:**

- A training on good scientific practices, reproducibility, etc. should be proposed to EJP RD funded researchers (interaction P1/P3/P4). Whether this training should be mandatory or not has to be discussed. It could be dedicated to young researchers involved in the projects (PhD, Post-Docs) to generate a new generation of well-trained people. Education on statistics would also be very useful for funded researchers (synergy of WP19 and WP20 already started on that area). It is important to make sure that data produced by funded projects are usable and available within EJP RD and the RD community.
- It is important to have a connection between the project we fund and the tools we develop. Today, we strongly encourage the funded projects to deposit their data in databases that are part of VP. It could be enforced in the future, but as the contracts are established by the funding agencies at national level, it is important to demonstrate its usefulness to have the willingness of funding organisations.
- Trainings on CT methodologies need to be regulator compliant and thus should be first validated by the regulators. EMA has working groups on methodologies involving pharmacometrics. A connection through Mats Karlson (Uppsala University), leading the EMA working group on that topic can be done via Ralf-Dieter Hilgers
- For any question regarding IRDiRC Task Forces of your interest, do not hesitate to contact Carla D'Angelo ([Carla.dangelo@ejprarediseases.org](mailto:Carla.dangelo@ejprarediseases.org)) or Galliano Zanello ([galliano.zanello@ejprarediseases.org](mailto:galliano.zanello@ejprarediseases.org))

**Action:**

- ⇒ The WPs for which synergies were detected should establish (when needed) dedicated working group to advance on the implementation of the proposed synergistic actions.

Actions related to COVID-19 mitigation measures

See slides 10-11 for complete information

**Additional points of discussion:**

- The possibility to propose a centralized support for online events funded through EJP RD activities (WP7 and WP17) was discussed. It was underlined that the support should not necessarily include the “call in/webinar” platform (like MS Teams) since this is already working well but rather focus on specific tools to use during such events (e.g. e-learning tools, mind mapping, calls notebook) as well as technical support during the event.

**Actions:**

- ⇒ Collect the needs of tools and the description of how the workshop will be implemented and performed.
- ⇒ Guidelines to organize more complex online events will be provided, based on the lessons learned, and procedures established during the preparation of the P2 annual retreat

[How do we deal with planned activities that are not working as expected? - feedback by Pillar](#)

See slides 12-13 for complete information

**Pillar 1:**

Activities running in line with work planned.

For the WP7, mitigation measures have been put in place after the Covid-19 crisis. We will need to check how this is accepted by the community (as sometimes organisation of online events is not considered the best option) and evaluate the situation within next 6 months.

**Pillar 2:**

Workflow starting with use cases may rise prioritization issues that need to be better streamlined. For this purpose, it was decided that Pillar 2 leaders will have open slot time every two weeks dedicated to task leaders and any other P2 partners to discuss specific issues and make some guidance. P2 leaders could also ask a person to come for clarification and specific reporting.

**Pillar 3:**

The transfer of planned trainings as online courses during the Covid-19 crisis required additional human resources (WP14 and WP15) that is balanced with the money not spent in the physical meeting.

Only the Leadership school planed in Gdansk end of November will be rescheduled in Year 5 as it is important to keep it as a face-to-face meeting (networking). The go/no go decision will be taken based on lack of applicants and satisfaction surveys (post-training).

**Action:**

- ⇒ The development of the 2<sup>nd</sup> module for the e-learning platform is a bit behind the schedule because its development was planned with 2 ERNs that are not available at the moment. That is why WP16 is inviting P2 and P4 partners to discuss and (if agreed) establish new programme to deliver new module

#### **Pillar 4:**

P4 activities were not very much impacted by the Covid-19 crisis.

It was noticed in the WP20 Task Force Group that the participation of some TFG members was very low (meaning not present for over 70% of the time). In order to solve this issue, first an email was sent to those people to ask about their willingness for involvement and dedicated time for this task. Each member had the possibility to confirm their contribution or resign. Currently, the WP Leaders are reviewing all tasks and budget for each partner in order to evaluate as much as possible partners' involvement for the 6 last months of year 2 and for future involvement in year 3. The new proposition of budget distribution will be presented to Pillar 4 partners and (upon agreement) new distribution will be proposed for the GA validation in AWP3.

Some changes will be implemented in WP20 due to the start of new activities like education programme or implementation of the call for innovation projects. It was also agreed that the publication materials supporting the dissemination of the information on novel methodologies will be postponed for later in order to include the inputs from the webinars and funded demonstration projects.

WP19 did not face any go/no go decision. The portfolio of projects mentored is growing. It was first difficult to get some projects on board, so it was enlarged to ERNs projects. In addition, WP19 is accompanying currently JTC2020 applicants with mentoring service; this would be continued with JTC2020 projects that will be recommended for funding, and the capacity of the service should be ok for the future. The Innovation management toolbox development is ongoing, and it should be expanded by ODDG (Galaxy Guide) of IRDiRC but it may require some prioritisation of workload.

#### **Coordination:**

At the coordination level, the workload is expected to be growing. It is planned to increase capacity of the team by recruiting an additional person whose activity would be related to MsTeams (e.g. online event support) and who, on a daily basis, would share the global workload to ensure support for each pillar.

#### **Transversal activities**

Some planned deliverables are overlapping. It was decided to produce smaller version of the Del. or to propose merging of deliverables into one to be sure to deliver meaningful documents. This will be reflected through the changes applied in the AWP Y3.

For the WP4, an unexpected workload came especially from Pillar 1 funded project Ethics monitoring and the involvement of AREB in project activities will be higher than initially foreseen.

As for the WP2 and actions related to the strategy establishment, the constitution of National mirror group being very asymmetrical in the different EJP RD countries their input cannot be taken into account properly yet while it would have been and will be very useful. In the meantime, a thorough yearly analysis of research needs is being provided to feed the strategy set up for each AWP.

#### **Actions:**

- ⇒ The Policy Board and Governing Board will be solicited for help and regularly informed during the joint meeting, so their contribution helps in the set-up of such national bodies.
- ⇒ A survey to ask interest to be part of NMG directly sent from EJP RD coordination (preferably to pre-identified target organizations, for more successful contacts) would help to constitute those groups (for example in Spain)

## Introduction to financial optimisation

See slides 14-23 for complete information

### [Presentation of the update of the current status of the budget](#)

### [Propositions on budget to consider by ExCom members in the finalization of the AWPY3](#)

- Attention for travels budget: some partners already used a big part of their budget and should take into account that the repartition should be equilibrated for 5 years and their presence is expected at yearly meetings.
- Whenever other goods and services budget was planned for a specific activity and assigned to a partner responsible for this activity, it should not be considered that such budget (if not spent) belongs to this beneficiary. Therefore, it cannot be transferred or used for example to cover the travel costs of this specific partner. The remaining budget from other goods and service should go back at pillar level as reserve budget for other activities.
- The redistribution of the budget should be discussed at Pillar level. No new budget can be generated.
- P1 meetings are also identified as travel budget: they are 100% covered and have to be declared. **(information added in the presentation)**
- The information from Pillar 2 on remaining budget in 2020 due to Covid-19 crisis is missing (except for Annual Retreat) → partners are **requested to share information** with Blandine to complete the table.
- We have to be careful to spend the budget during the whole project and not keep too much money to spend in year 5. The EC reimburses based on the actual spending so if partners do not spend enough, the EC will reimburse only what has been declared, which may lead to the need to advance money by partners in coming years (due to the very low rate of the EC reimbursement).
- It seems that there is a lot of small-scale needs in each pillar that were abandoned because it was not possible to cover them with initial budget → pillars should consider covering them with remaining (unspent) budget (only if really needed).
- All partners, including the ones with 'smaller' activity shall report their actions. It is the responsibility of WPL to monitor TL works and vice-versa (through calls, minutes, reports). It has to be done in transparent manner in order to avoid that some partners work without consulting or reporting on what they are doing to the others (that are impacted by their actions). All pieces of work in the EJP RD are connected.
- In case some partners are not involved anymore in tasks, it is possible to re-allocate their responsibilities (and thus budget) to another partner, but such change requires justification.



**Actions:**

- ⇒ The final AWPY3 budget has to be finalized by August 18<sup>th</sup>, 2020.
- ⇒ In case partners underspend their budget because they are not being involved: WPL/PL should take the lead to discuss the issue with the partners and come to the coordination once the final decision is taken.

A huge thank you to all EJP RD partners for their amazing commitment during the Covid-19 crisis to keep the timeline. We can proudly say that all activities are running with almost the same pace despite the crisis.

**Pillar 2 ERN & RD Researchers surveys**

*See slides 24-52 for complete information*

The surveys allowed Pillar 2 to build its strategy for the development of the Virtual Platform. The RD research community was targeted through the survey: ERNs in 2019 and RD researchers in 2020.

Based on the results of the survey, a publication will be prepared by P2 (with support of the coordination).

**Additional points of discussion:**

- We have to make an effort to make sure that the tools and services developed and provided by EJP RD are known and easy to use by the RD community.
  - Pillar 2 should engage as soon as possible with P1 funded projects
  - We could use the Policy Board to help in encouraging the national RD community to use our resources. We should also think to target national funding agencies to make it mandatory to deposit the data produced by the funded projects.
  - We have to be careful with overload of information, which is a barrier to have people aware of existing standards and tools.
  - We need to adapt the existing to RD research challenges and to show how the tools/infrastructures/etc. fit the needs of the researchers/stakeholders
  - Creation of a dedicated webpage, mind map and additional proactive actions are needed (e.g. similarly to WP 19 Innovation Management)
- It would be important to repeat the survey at the end of the project to see the results (and hopefully improvements).

**Actions:**

- ⇒ Pro-active actions (for example webinars, specific recommendations or requirements from funding agencies, etc.) need to be taken in order to be sure that the tools and services developed and provided by EJP RD are known and easy to use by the RD community
- ⇒ Development of a webpage and MindMap with the available resources

### Sustainability survey

See slides 53-72 for complete information

#### Additional points of discussion:

- The engagement of people for the sustainability of activities is key and should be done as soon as possible.
- The WP3 should be aware of the discussions that are taking place regarding sustainability of EJP RD activities with external partners in order to be able to provide help. The negotiation takes time. WP3 can monitor the process and define EJP RD position only if they are aware of the discussion at the earliest.
- A catalogue of the resources, including business model for sustainability from external partners will be developed through WP3.
- The survey was quite complex to answer so partners are not 100% sure that the answer provided are accurate in some parts of the survey: this will be discussed through direct exchange between WP3 and WP/Task leaders.
- For some elements reported through the survey, a pattern could be found, and common sustainability plan could be established for different activities.
- With so many elements reported, prioritization will be needed and will depend on various scenarios (e.g. EJP RD continues to be funded or not)
- Some critical elements developed under EJP RD are very much dependent on the sustainability of the others. Therefore, it is necessary that partners keep in mind the sustainability and IP issues at all time and are accompanied by WP3.

#### Actions:

- ⇒ Over the next few months, direct communication between WP3 and WP/Task leaders will be established.
- ⇒ The follow-up of the first survey will be launched at the end of the year through a simpler structure and together with a guide to explain the terms used and what is expected → to be replaced by interviews and related minutes/reports whenever possible (since another survey may not be efficient to achieve WP3 goals)
- ⇒ A specific Team/Channel will be created to raise any issue and give more support on the sustainability → to be discussed with Yanis and WP3
- ⇒ Set up a working group to discuss how to deal with external partners on sustainability questions.

### EJP RD Ethics Advisor

See slides 73-95 for complete information

#### Additional points of discussion

- The central contact point in relation to ethics will remain the WP4 Leader (FGB) and the AREB. The procedure in place to contact the AREB with questions on EJP RD activities will remain.
- Within deliverables, a collection of good examples of consent forms will be done with help of partners.

- Among the deliverables, the report of External Ethics Advisor is the responsibility of the Ethics Advisor, while other deliverables remain under the responsibility of INSERM.



# EJP RD Policy Board and Governing Board meeting

8<sup>th</sup> of July 2020

13:00 – 18:00

Online

**Attached document:**

Slides presented during the meeting – Annex 2

**List of participants:**

Name		Board	Country	
Janis	Ancans	Policy Board / Governing Board	Latvia	Present
Pilar	Aparicio Azcárraga	Policy Board	Spain	Replaced by Yolanda Agra
Juan	Arenas	1+MG		Present
Gonzalo	Arévalo	Governing Board	Spain	Present
Anželika	Balčiūnienė	Policy Board	Lithuania	Present
Wolfgang	Ballensiefen	ICPerMed		Present
Ingeborg	Barisic	Policy Board	Croatia	Present
Catherine	Berens	Policy Board	-	Present
Eva	Bermejo-Sanchez	ExCom		Present
Valentina	Bottarelli	Policy Board	-	Present
Anthony	Brookes	Governing Board / ExCom	UK	Present
Virginie	Bros-Facer	Governing Board / ExCom	EURORDIS	Present
Tanja	Bullow	ExCom		Present
Onur	Burak Dursun	Policy Board	Turkey	Present
Claudio	Carta	ExCom		Present
Blandine	Castrillo	ExCom		Present
Nikki	Coutts			Present
Carla	D'Angelo	ExCom		Present

Anne	Demoisy	EJPRD Ethics Advisor		Present
Sofia	Dimitropoulou	Governing Board	Greece	Present
Pavla	Dolezalova	Policy Board	Czech Republic	Present
Roseline	Favresse	ExCom		Present
Christine	Fetro	ExCom		Present
Bernard	Grimm	Policy Board	-	Present
Juliane	Halftermeyer	ExCom		Present
Ralf-Dieter	Hilgers	ExCom		Present
Tuija	Ikonen	Policy Board	Finland	Present
Richard	Imrich	Policy Board	Slovakia	Present
Avi	Israeli	Policy Board	Israel	Present
Daria	Julkowska	ExCom		Present
Judita	Klosakova	Policy Board / Governing Board	Czech Republic	Present
christina	Kyriakopoulou	European Commission		Present
Annalisa	Landi	ExCom		Present
Hélène	Le Borgne	Policy Board	-	Present
David K	Lee	Policy Board	Canada	Present
Jordi	Llinares	Policy Board	-	Present
Anabela	Lopes Isidro	Governing Board	Portugal	Replaced by Sandra Alves
Patrícia	Maciel	Policy Board	Portugal	Present
Pierre	Meulien	Policy Board	-	Present
Yanis	Mimouni	ExCom		Present
Lucia	Monaco	Policy Board	-	Present
Rima	Nabbout	ExCom		Present
Joana	Namorado	EJPRD Ethics Advisor		Present
Catherine	Nguyen	ExCom		Present
Eleonora	Passeri	ExCom		Present
David	Pearce	Policy Board	-	Present
Florence	Quist	Governing Board	Belgium - Wallon	Present
Ana	Rath	ExCom		Present

Alessandra Renieri	Policy Board	Italy	Present
Etienne Richer	Governing Board	Canada	Present
Zivile Ruzele	Governing Board	Lithuania	Present
Ryszard Rzepecki	Policy Board	Poland	Present
Jale Sahin	Policy Board / Governing Board	Turkey	Present
Gunnar Sandberg	Governing Board	Sweden	Present
Franz Schaefer	ExCom		Present
Guenter Schreier	Governing Board	Austria	Present
Leo J Schultze Kool	Governing Board	Netherlands	Present
Ralph Schuster	Governing Board / ExCom	Germany	Present
Bertrand Schwartz	Policy Board / ExCom	France	Present
Serena Scollen	1+MG		Present
Supriya Sharma	Policy Board	Canada	Present
Egle Simelyte	European Commission		Present
Pilar Soler			Present
Domenica Taruscio	ExCom		Present
Biruté Tumiene	ExCom		Present
Katerina Tzima	ExCom		Present
Anton Ussi	ExCom		Present
Sonja van Weely	ExCom		Excused
Jérôme Weinbach	Policy Board	France	Present
Theda Wessel	Policy Board	Germany	Present
Galliano Zanello	ExCom		Present

## Agenda

13:00 – 13:15	<b>Welcome word and introduction to the EJP RD</b>	Daria Julkowska (INSERM) Coo
13:15 – 13:30	<a href="#"><u>Presentation of new members</u></a>	All
13:30 – 14:00	<a href="#"><u>Summary of EJP RD activities and achievements in Year 1</u></a>	Daria Julkowska (INSERM) Coo
14:00 – 15:30	<a href="#"><u>Annual Work Plan Year 3 – Discussion on strategic points</u></a>	All
15:30 – 15:50	<b>Coffee break</b>	
15:50 – 16:50	<a href="#"><u>Erreur ! Résultat incorrect pour une table.</u></a> <ul style="list-style-type: none"> <li>• What are the advancements in building NMGs in different countries?</li> <li>• What are the best practices?</li> <li>• How best we can engage the community making sure that RDs remain an important point on the policy agendas?</li> </ul>	All
16:50 – 17:50	<a href="#"><u>Alignment with other strategic initiatives</u></a> (1+MG, Personalized Medicine and other to be reported by PB members)	All
17:50 – 18:00	<a href="#"><u>Erreur ! Source du renvoi introuvable.</u></a>	Daria Julkowska (INSERM) Coo

## Table of Contents

<b><i>Presentation of new members</i></b> .....	<b>16</b>
<b><i>Summary of EJP RD activities and achievements in Year 1</i></b> .....	<b>16</b>
<b>ACTIONS</b> .....	<b>18</b>
<b><i>Annual Work Plan Year 3 – Discussion on strategic points</i></b> .....	<b>18</b>
<b>Pillar 0</b> .....	<b>18</b>
<b>Pillar 1</b> .....	<b>20</b>
<b>Pillar 2</b> .....	<b>21</b>
<b>Pillar 3</b> .....	<b>22</b>
<b>Pillar 4</b> .....	<b>24</b>
<b><i>Rare Diseases National Mirror Groups</i></b> .....	<b>26</b>
<b><i>Alignment with other strategic initiatives</i></b> .....	<b>27</b>
<b><i>Summary and Next steps</i></b> .....	<b>Erreur ! Signet non défini.</b>

## Minutes:

---

### Presentation of new members

- Pierre Meulien: executive director of Innovative Medicines Initiative (IMI), European public-private partnership
- Janis Ancans: Latvia, Ministry of Education and Science, Horizon 2020 contact point at the Programme Management agency
- Richard Imrich: Slovakia, director national institute Rheumatic diseases, Nominated by the Ministry of Education, Science, Research and Sport
- Onur Burak Dursun: Turkey, Associate professor of Child and adolescent Psychiatry, established new unit for RD and autism, a body for Policy making, represents the ministry of health since 2020.
- Anželika Balčiūnienė: Lithuania, responsible for children care and RD in the Ministry of Health Lithuania
- Alessandra Renieri: Italy, Sienna, full professor in Medical Genetics at the University of Sienna, represents the Ministry of Education, University and Research
- Pilar Aparicio Azcárraga: Spain, Ministry of Health, Social Services and Equality, General Director of Public Health, represented by Yolanda Agra, Deputy Director of Quality Assurance and innovation for the RD national strategy
- Patricia Masia: Portugal, Associate professor of Molecular Genetics and biochemistry/biotechnology, representing the Ministry for Higher Education, science and technology
- Ryszard Rzepecki: Poland, Medical Biotechnology Professor working in RD, assigned by the national center of RD in Poland.
- Supriya Sarma: Canada, Chief medical advisor at Health Canada,
- David K Lee: Canada, Chief Regulatory Officer at Health Canada

### Summary of EJP RD activities and achievements in Year 1

[See slides 4 to 17](#)

#### Discussion

A better strategy to involve end-user was suggested including open questions, direct consultations and close teleconferences with the two survey respondents (end users).

- This is already addressed. The two surveys captured the end-user's needs, brought new ones, allowed to redirect the strategy and plan dedicated support. The work in EJP RD, and particularly in Pillar 2 is based on collaboration with them, the different elements are developed through direct consultation with end-users. This is the same for the EJP RD training activities.
- Moreover, the survey respondents, who expressed their will to be re-contacted, will be solicited for structured interviews on specific topics

The EC was pleased with the promising progress made so far given the complexity and size of the EJP RD. there is no other comparable project. It has high expectations to



showcase how to catalyse an RD ecosystem that provides innovative solutions benefiting RD patients.

- The Pillar 1 achievements are important, they are similar for the calls launched at the EC level.
- The Pillar 2 complex endeavour of putting data together through linking resources is at the centre of the EU data space that the EC is trying to develop; EJP RD is performing significant steps. Taking into account the users' needs (through surveys) is important to ensure that the developments answer these needs.
- Guidance on the development of ERN registries should take into account what is already existing; the collaboration with the Joint Research Centre (JRC) and ERNs is satisfying. collaboration to build on what is existing
- The Clinical Trial Support Office (helpdesk) is a nice outcome of Pillar 4 to address the field where difficulties lie according to EC view. It will be advertised.
- The EC wants to propose some selected projects to benefit from the EJP RD mentoring activities
- Dissemination activities are important. The EC also contribute to disseminate results of EJP RD and congratulates this latter for the concise YouTube video that presents the different activities: <https://youtu.be/ip1cTodXC3k>
  - tailored communication messages targeting different groups (patients; doctors, researchers, etc.) is recommended to foster the buy-in of all these stakeholders and increase the probability of success.

The EJP RD Data related work is also strategic for industry. There is a Big interest in liaising much more with industry who is also an end-user of RD data.

The heterogeneity of how data is collected stored and made accessible is the first the main encountered barrier. Th EJP RD opted for a federated architecture. This means that the data stays within the data-source and the data exposure and access is harmonised. The RD data resources are brought one by one to be equipped for being queried.

There are sometimes competing technologies; it is extremely important to have robust set of criteria to ensure that the semantic and quality standards, the GDPR and the sustainability requirements are carefully selected.

This is the main lesson learned so far for the EJP RD Data related work

The close collab EJP RD with ERNs is important to achieve the goals, it is currently taking place and will increase.

Collaboration on data sharing and consent issues are tackled by groups. The gained experience and guidance on how obstacles can be passed will be shared.

The EJP RD helpdesk on the EJP RD: to promote collaboration and to submit end-user cases

The EJP RD helpdesk is receiving different demands addressed collaboratively by EJP RD experts. It can be used to submit end-user cases/needs and promote collaboration: <https://www.ejprarediseases.org/index.php/ejp-rd-helpdesk/>

The improvement of this helpdesk is planned.

## ACTIONS

- ⇒ EJP RD (WP19 & coordination) will liaise with the EC to get the selected projects that need WP19 mentoring services
- ⇒ EJP RD need to disseminate tailored communication messages targeting different groups (patients; doctors, researchers, etc.)
- ⇒ Share experience on the management of data sharing and consent issues

## Annual Work Plan Year 3 – Discussion on strategic points

See slides 18 to 24

Only specific strategic issues to discuss with boards

### Pillar 0

- How to best prepare the next phase – Rare Diseases Partnership under Horizon Europe ?
- What is the most efficient approach to ensure EJP RD (elements/services) sustainability and their support at the national, EU and international level ?

The preparation for the Rare Diseases Partnership under Horizon Europe should start before the strategic meeting in 2021 organised as a back-to-back meeting with the IRDiRC/ReACT congress taking place on 12-13 January 2021. The strategic meeting will focus on how the RD future is seen beyond EJP RD and how to tackle Horizon Europe

There is a need to elicit:

- how all the pillars were successful in connecting the dots of the current fragmentation to build the RD ecosystem.
- What in 18 months is the most valued part of this Ecosystem.

It is important to define where the most valuable KPI would lie for the construct we have today. Then, ask others where they see the remaining gap.

Although challenging, the digital infrastructure gap is being addressed. However, the need to undertake it over a long time, a decade, is needed. It is important for IMI, as a funder, to make sure that connections between the various initiatives are made to avoid creating other Silos. The linkage of EJP RD with big (infra)structures and other projects represent the strength on what we can build.

Each element contributing to the EJP RD overall objective has its own Key Performance Indicator (KPI) or Key Result Indicator (KRI). The dedicated task for monitoring is in place; Work Packages were asked to provide KPIs and KRIs. This was a long-term process; the indicators were progressively developed and need to be revised every six months. Some external input and evaluation would be valuable. Considering them all together shows that reaching stakeholders to link or strengthen current link with the RD community and other programmes is progressive.

In many countries, there are no funding programmes for RD research. EJP RD instrument is valuable for them, providing the RD research support.

The creation of the Virtual Platform (VP) is step wise process; it is impressive to see how much there is yet to solve. Collaboration with end users including ERN is of utmost important.

There are some bottlenecks, such as regulatory and ethics issues at national level, that are yet hard to solve. This also concerns the training, translation and clinical trials activities.

**The most added value until now: the level of connection (RD Ecosystem) that was not seen before.** We can directly observe how stakeholders make connections, establish common cultures, share the **highest standards and become a common community.** **The Policy Board plays an important role for the connection to the Member States and to a broader Ecosystem.**

The example of the Czech Republic was mentioned. A website for all RD activities was developed and a national RD registry was put in place. The country is now Completing the RD strategies for the next 10 years. The main problems are communication, information sharing and harmonisation of activities. Finding information easily needs to be improved.

The EC will not start embarking on partnership where there is no added value. This was also requested by Member States; the mushrooming of partnerships leads to focus on meaningful ones.

Good and strong KPI are not only needed to demonstrate at the operational level but also at the policy level. The demonstration of a better care and better delivery of innovation to patients fostering personalised medicine is needed. The focus of Policy delivery KPI is important.

Horizon Europe (HE) still being prepared. The work is carried out in various Directorates and public consultation are performed to identify orientation for HE. The document for orientation and strategic planning is open for consultation ([https://ec.europa.eu/info/horizon-europe-next-research-and-innovation-framework-programme\\_en](https://ec.europa.eu/info/horizon-europe-next-research-and-innovation-framework-programme_en)). It describes priorities and expected impact over the first 4 years of HE. The Directorates focus on the partnerships to be launched in the first 2 years (2021 & 2022); the RD partnership discussion is expected to start at the end of 2020 or the beginning of 2021 gathering the important RD stakeholders. This discussion should be transparent and will take into account the EJP RD achievements, especially where the added value would lie. The call will be prepared in 2023

**The planned midterm review of the EJP RD with external experts will help in providing feedbacks on what went well, what still remains to be done possibly through other partnerships.**

From the industry perspective, there is a need:

- to showcase progress in two topics:
  - The Digital space through the building of the registries and the Virtual Platform
  - Clinical Trials to show how progress was made to accelerate development
- to evaluate the quality of the networks through

- the integration of ERNs and as many countries as possible
- the evaluation of the strength of the network

## ACTIONS

- ⇒ Be connected to other programs, e.g. IMI (show this connection in respective actions)
- ⇒ Define a KPI to reflect on the level of connection with other related programs (Evaluate the quality and impact of the EJP RD network)
- ⇒ Develop a Policy delivery KPI to demonstrate a better care and better delivery of innovation to patients fostering personalised medicine
- ⇒ Prepare & Monitor the EJP RD achievements with added value that will be taken into account during the Horizon Europe RD partnership discussion.
- ⇒ Define a KPI to monitor the level of integration and the strength of the EJP RD network
- ⇒ Involve the Policy Board More to liaise and align with national RD/Health/Research Activities

## Pillar 1

- Topic of the Joint Transnational Call (JTC) 2022 needs to be defined : the previously envisaged topic is now obsolete due to the existing difficulty to fund clinical trials within EJP RD funding schemes

A "second" round of already closed calls of earlier years (JTC 2019, 2020, 2021) was proposed. Keeping calls on basic research in RD should be considered as the EU needs in this field are not covered yet. Research groups of small countries where there is no dedicated national programs for RD are in need of the basic research funding.

The EMA free scientific advice for academia was reminded: <https://www.ema.europa.eu/en/news/academia-developing-medicines-rare-diseases-receive-free-ema-scientific-advice>

The EJP RD should serve as a lasting general hub for all the activities that are complementary and ancillary to research projects to successfully bring concrete results to the patient.

The EJP RD Pillar 1 is a sounding board testing the capacity of other pillars to support RD research and serve the RD community.

Excellent science should be promoted by Pillar 1 and be complemented by EC calls but on the other hand EC should refer to EJP RD in its calls (we need to make sure that we are ready to support such referencing by suitable actions and services)

The patients' organisations are included in the call topics development pathway (e.g. through EURORDIS involvement). This is continued for next call topics definition.

The Horizon Europe activities should refer to the EJP RD as a reference point to all those support activities.

Regarding the orientations towards the first Strategic Plan for Horizon Europe ([https://ec.europa.eu/info/files/orientations-towards-first-strategic-plan-horizon-europe\\_en](https://ec.europa.eu/info/files/orientations-towards-first-strategic-plan-horizon-europe_en)), this first work programme shared was developed in co-creation with stakeholders following a successful session for RD during the [European Research and Innovation Days](#). In the document RD is present different places notably for the intervention area, and most importantly the mention of future partnerships starting on 2024.

For the Development of the Work programme, confidential discussion with Member States representatives are ongoing.

The strategic discussion on the next steps and EJP RD future will address the possibility to include networks other than ERN. All ERN need to apply to the cross-border healthcare directive. The question about the inclusion of Turkey is about EU memberships. ERN work closely with professional associations; Turkish participation could be enhanced through this professional networks. Turkey is now part of Orphanet network (part of EJP RD) and in that framework it will allow to make Turkish RD research activities visible.

Of note, the IRDiRC Task Force on Clinical Research Networks aims to create a map with the existing clinical research networks all over the world and analyse their characteristics to provide recommendation in order to enhance and expand the collaboration (to different countries).

The EJP RD need the help of both the Policy Board and the Governing Board for the dissemination of the next JTC (2021) Call on Humanities and Social Sciences (SHS). The headlined description is included in the slide No 21.

## ACTIONS

- ⇒ The Policy Board and the Governing Board will be solicited by EJP RD to help with the dissemination of the next JTC (2021) Call on Humanities and Social Sciences (SHS).
- ⇒ The JTC 2022 Topics will be defined taking into account the topics from previous calls and the possible extension of funded projects
- ⇒ Elicit how EJP RD is serving as hub to activities that are COMPLEMENTARY to research projects that are being financed
- ⇒ Excellent science should be promoted by Pillar 1 and be complemented by EC calls but on the other hand EC should refer to EJP RD in its calls (**we need to make sure that we are ready to support such referencing by suitable actions and services**)
- ⇒

## Pillar 2

- The heterogeneous interpretation of GDPR at member state and institutional level

Inform consent procedure is not facilitated by GDPR in Europe; there is a heterogeneous interpretation by member states and institutions (one of the problem is that independent local Ethics Boards can add requirements on top of GDPR such as clinical trials like requirements including the request for patients' insurance for registries).

The current funding to develop and support the development of the ERN registries provides unique opportunity to build registries **FAIR** (Findable, Accessible, Interoperable, Re-usable) from the start. We aim to find ways to tackle this GDPR interpretation issue in order to complete this development process by the end of 2021.

EJP RD is setting-up an Informed Consent template for ERN Registries in order to increase the probability of Ethics approval across European involved institutions.

The work on machine readable consent is performed in parallel (in line with the objective of IRDiRC and a continuation of a previous activity); it will take more time.

What procedure could be applied to help to come for more uniform interpretation of GDPR in clinical trials?

**This important issue of GDPR interpretation impact on the informed consent process, data sharing and re-use for research purposes is not restricted to RD** (e.g. the attractiveness of the EU for all clinical trials is concerned). There is a need to have a joint learning on this. Many activities dealing with this issue are ongoing in IMI, for e.g., the multinational Harmony project on haematological malignancies have already dealt with enormous amount of GDPR issues. Cross project learnings on how the problem was solved is in place. **IMI would like to share experience and Knowledge with EJP RD on this topic.** Even though data sharing framework and data protection may be different in Canada (related issues across Canadian provinces were encountered), **the Canadian colleagues would like to collaborate.**

The EC is aware that this issue is recurrent since 2018. The EC should be informed on any issue encountered to allow for linking information, identifying signals triggering urgent action (and prioritisation) and try to find common solving tool. **EJP RD should take into account the recommendations published in relation to COVID-19** ([https://edpb.europa.eu/sites/edpb/files/files/file1/edpb\\_guidelines\\_202003\\_healthdatascientificresearchcovid19\\_en.pdf](https://edpb.europa.eu/sites/edpb/files/files/file1/edpb_guidelines_202003_healthdatascientificresearchcovid19_en.pdf)) **and the forthcoming guidelines for processing of health data for research.**

### ACTIONS

- ⇒ EJP RD will liaise with IMI and Canadian Colleagues to exchange current experience and knowledge on GDPR compliance for informed consent process and data sharing
- ⇒ The EC should be informed on the GDPRD issues impacting informed consent process, data sharing and reuse and (if possible) engaged in the related actions to better follow up and support when needed

## Pillar 3

---

- Identification of new education & training needs/gaps

The education and training on RD research are insufficient, fragmented and geographically unequal across Europe. The EJP RD has already started to fill this gap. The capacity building is highly dynamic, activities and standards are constantly developed and established. There is a need to have a deep evaluation of existing resources and identify synergies and gaps to feed the Pillar 3 Programme.

The survey for ERNs and RD researchers will be used to embed the EJP RD outputs in the practice of stakeholders, through trainings, to achieve high level RD research. **The recommendations from “key” National stakeholders to develop RD research are also needed.**

The list of trainings will be presented to the RD community, including the Policy and Governing Boards and submitted through the usual Annual Work Plan to be established in Year 4.

The face-to-face trainings have limited capacity, Online Academic courses are being developed.

**At the national level the challenge is “the scalability of the training programme”; the language might be a hurdle.**

Courses dedicated to train the “trainers” will allow for the provision of the pillar3 trainings in national languages.

The role of the Member States to do this training pull from the central design is a key factor for success. Member states can adapt these training in their own national languages.

**How to measure the impact of these trainings on healthcare or measure any other outcome eliciting the implementation of their teachings ?**

Because of the fact that the training courses are free, and the number of registrations exceeds the attendance capacity, questionnaires are sent to registered participants in order to select the attendants whose background, position and career planning are fitting the training objectives the most. When this is combined with the matching of the selected training participants' profile/position (using medical schools directories for e.g.) with the existence and implementation needs of a national rare disease plan/strategy, and with post-training satisfaction questionnaires, the probability of impacting RD research and healthcare can be enhanced (e.g. through the adoption of significant clinical outcome for therapy development and care management).

Trainings were proposed for the JTC applicants to improve the quality and the use of standards in RD research. Getting accreditations from the trainings provided would increase their impact (and value).

Currently, it is not possible to really assess this long-term impact on RD research and healthcare. Follow-up surveys of the training's attendants need to be planned. Mid-Term indicators should be identified including the level of involvement in further collaborations and networks.

More generally, the national uptake of what EJP RD is producing could be considered as a main indicator. The role of National Mirror Groups is important in conveying the results and promoting their uptake.

## ACTIONS

- ⇒ Reach key national stakeholder (particularly those involved in existing or planned/related National RD Plan) to adapt training courses and provide guidance
  - ⇒ Prioritize the gaps that need to be filled through EJP RD action
- ⇒ Assess the (long-term) impact of the training outcomes (on healthcare and professional development) through follow-up survey of attendants
  - ⇒ Identify mid-term indicators for this impact assessment (including the level of involvement in further collaborations and networks.)
  - ⇒ Assess the national uptake of EJP RD outcomes

## Pillar 4

The demonstration projects aiming at testing previously published innovative statistical methodologies (based on outcomes of Asterix, IDeAI, InSPiRe) are ongoing; complementary related training activities are being planned.

Bringing together existing services to develop new regulatory validated analytical methodologies (through the (re)use of real Clinical Trial cases and the involvement of different stakeholders working on genetics, pharmacometrics, surrogate endpoints and/or registry data [ERNs and Patients]) is hindered by the lack of visibility.

There is a need to engage with Regulators at the national and international level (EMA, FDA) as well as with existing clinical trial platforms gathering patients, industry and regulators. Most of these latter are disease focused, a platform dedicated to RD or a group of RD can be considered.

The European Clinical Research Infrastructure (ECRIN) as well as other Research Infrastructures are not yet very well known by the RD community. ECRIN is an EJP RD full beneficiary working with the other Pillar 4 beneficiaries (experts in Clinical Trial methodologies) to bring together the aforementioned scattered stakeholders.

The EMA (Jodi Llinares) would be interested to engage in the EJP RD action on the development of new regulatory validated analytical methodologies through the liaison with its biostatistics Working party. Other points where collaboration with regulatory agencies was mentioned in the Annual Work Plan was also noted by EMA.

A workshop at EMA could be planned; EMA representatives will be invited for the January 2021 Policy Meeting.

The ICMRA coalition (<http://icmra.info/drupal/en/strategicinitiatives>) gathers several regulatory agencies (and where EMA is chairing the Board of Directors) have an innovation pillar looking at novel therapy development pathways that have been successful in facilitating the review of products brought to the market with innovative methodologies. Their work on innovation is more on technologies (e.g. bioprinting). Discussion to support EJP RD action (on innovative methodologies) can be considered.

Moving beyond the clinical trial domain to address Market Access and pharmacoeconomic topics is not considered by EJP RD although there is a need for



a “holistic view” that includes leading researchers to understand the steps undertaken beyond Clinical trials.

EJP RD will tackle the prioritised identified gaps relating to the above mentioned methodologies. Market Access and pharmacoeconomic related gaps would be addressed by the next phase of the programme. Moreover, drug pricing has a socio-political component that is less technical and often country-specific; the sponsors are the key stakeholders for discussing these topics (drug repurposing could be an exception to this).

## ACTIONS

- ⇒ Consider setting up an RD platform for Clinical trial modelling (or linking to an existing one)
- ⇒ Follow-up with EMA (Jordi Llinares) for:
  - ⇒ the action on the development of new regulatory validated analytical methodologies (liaison with EMA biostatistics Working party)
  - ⇒ other points needing collaboration with regulatory agencies as mentioned in the AWP
- ⇒ Follow up with Jordi Llinares and Supriya Sarma on the possible interaction with The ICMRA for innovative methodologies.
- ⇒ Plan a Pillar4 discussion with EMA and invite EMA representative to the January 2021 Policy Meeting

## Rare Diseases National Mirror Groups

One of the recommendations received from Policy Board during the last 2019 meeting was to prepare a guideline on how to set National Mirror Groups (NMG) when these latter do not yet exist.

During the 2019 General Assembly (GA) meeting, a back to back meeting with the RD Polish Community was held for setting up a NMG. Another back to back meeting with the Portuguese RD community was foreseen during the planned 2020 GA meeting. Despite the COVID-19 context the Portuguese stakeholders started the work to set up a NMG. The obstacle faced is the difficulty to identify the right person at the right position to be committed to this NMG.

National Alliances for Rare Diseases (that federate patient organisations) should be used as ambassadors, to support the establishment of NMG. Patients at national level are used to discuss with different stakeholders EURORDIS is available to help and coordinate with their involvement.

NMG establishment in underrepresented countries requires support from EJP RD through central coordination and experience sharing (French experience and Polish Experience [Ryszard Rzepecki is willing to help for this regard]). This is even more important considering that often in those countries, such as Lithuania, there is no national research programme for rare diseases, hence, European Programmes are almost the single opportunity for their RD researchers.

There are many opportunities provided to these underrepresented countries through Joint Transnational Calls and Training activities, their exploitation for the set-up of NMG should be considered.

Orphanet nodes in each countries together with clinicians need to be involved in the set-up of NMG.

An issue might remain for the participation of the funding agencies that have no specific RD calls and of the National Authorities (such as Ministries) that might be more concerned by the clinical part relating to RD and less by research part.

The National Cancer Plan can be considered for Rare Diseases.

The Dutch NMG is being established, gathering large group with all the different categories listed in the "[EJP RD National Mirror Groups Terms of Reference](#)".

An Italian working group is building the second RD National Plan. It includes members from the Ministry of Health, Research, Planning DG, National Alliance and Charities, ERN representatives, the "Istituto Superiore di Sanità" (ISS), Orphanet contact point (Prof. Dallapiccola; involved in the network of Italian genomes), the Medicine Agency, and an Agency for assessing healthcare providers. This working group can be considered as the Italian NMG whose mandate will include providing feedback and commenting on EJP RD activities.

The work provided for setting-up the EJP RD NMG is appreciated by the EC. The EJP RD should not operate by itself; it is important to link on what is planned and what is not planned at the national level in order to optimize resource utilization through collaborative and complementary efforts.

## ACTIONS

- ⇒ Follow-up with EURORDIS to coordinate the involvement of National Alliances in setting up NMG.
- ⇒ EJP RD central Coordination will propose support for the establishment of NMG in underrepresented countries through NMG set up experience sharing and the exploitation of these countries' involvement in JTCs and training activities
- ⇒ Liaise with Orphanet National Nodes
- ⇒ Propose to the Italian National RD Plan Working group to be the EJP RD NMG
- ⇒ Set-up NMG in coordination with national RD plans.

---

## Alignment with other strategic initiatives

It is important to have a clear view and understand the work of the different initiatives (with a focus on "1+MG" and "IC PerMed") to make alignment and synergy possible.

Regarding NMG, EJP RD identified that at the national level there is a missing connection between the stakeholders and the people representing different projects.

1+MG acknowledged that the state of NMG constitution in each country is very different, this initiative wants to develop guidelines on how to establish a NMG and share it with the persons identified as driving their implementation at the national level.

There are no NMG foreseen in IC PerMed but within this latter initiative, meetings with Research and Health Ministries and other partners are held in France, Italy and Germany to allow the implementation of Personalised Medicine. In this field, it is important that research remains connected with healthcare. This is important as well for the RD field.

Some of the recommendations on connecting EJP RD with 1+MG and IC PerMed and the involvement in the upcoming programmes include:

- Ensuring that the different initiatives are connected as much as possible and mutually feed each other's reflection and planning (next steps)
  - while warranting that the right data, the right medicine systems, AI, diagnostic development, treatment development are made available
    - as an example, EJP RD and 1+MG could feed a more aligned and integrated vision on how to implement personalised medicine using genomics
    - deep phenotyping and next generation phenotyping should go hand in hand with the deep genotyping
- the need of a large unified project where the visions of EJP RD, 1+MG and IC PerMed are operationalised to impact the Health System. This can be done through IMI
  - ELIXIR is a key sustainability platform for a lot of IMI projects

- EJP RD will strive to connect to initiatives that will stem from the IMI call on Shortening the path to rare disease diagnosis by using newborn genetic screening and digital technologies
- The mutual involvement in Horizon Europe that takes into consideration this important close link between research and healthcare aspects. Also, the new [EU4Health](#) will boost preparedness for cross-border health threats and strengthen health systems so that they can face epidemics as well as long-term challenges by stimulating (among other fields) access to health care for vulnerable groups
- Ensuring that EJP RD feeds the reflection on the European Health Data space
- Demonstrating that what is being developed at the level of Rare Diseases can have an impact on the policies.
  - EJP RD needs to impact the Orphan Drug regulation

The EC needs to set up expert liaison groups gathering different stakeholders from the various RD related or linked initiatives in order to avoid redundancies, facilitate alignment and synergy, allow for better prioritisation of the RD community needs and foster the translation of results that impact the patients' care and prevention. This leads to the idea of setting up a networking of the networks under the EC impulse and monitoring.

Of note, the Steering Group on Promotion and Prevention set up by Commission's DG SANTE with Member States' representatives, although not usually gathering persons involved RD at the forefront, continues to discuss rare disease policies while looking for synergies.

## ACTIONS

- ⇒ Set up alignment calls between EJP RD 1+MG and IC PerMed
- ⇒ Set up with IMI a process/strategy to make EJP RD results impact the Health System
- ⇒ Engage the initiatives mutual involvement in the preparation of Horizon Europe, EU4Health and the European Health Data space
- ⇒ Liaise with the EC to set up expert liaison groups gathering different stakeholders from the various RD related or linked initiatives



# ExCom Annual Meeting

# Identification of new actions foreseen by Pillars

# New actions foreseen – Pillar 0

## WP1 – Coordination and management

- 🌟 Strategic meeting with Policy Board in January 2021 + support for the development of Rare Diseases Partnership under Horizon Europe
- 🌟 National strategic meetings supporting creation and/or organisation of National Mirror Groups
- 🌟 New Task Forces of IRDiRC and potential joint EJP RD – IRDiRC actions (according to the Roadmap 2021)

## WP5 – Communication & dissemination

- 🌟 RE(ACT) – IRDiRC Congress 2021 in January 2021 (since postponed from 2020)
- 🌟 IRDiRC 10th anniversary



# New actions foreseen – Pillar 1

## WP6, Task 6.5 - Working group for patient engagement in research

- Continuation (stage 2) of work with the members of the **working group on improving patient engagement in research projects** by organising several teleconferences in year 3 to:
  - 1) assess and review usability of the guide
  - 2) discuss the organisation of a webinar for prospective applicants wishing to learn more about patient engagement and patient partnerships in research projects
- Organisation of a webinar** that will be recorded and widely disseminated using EJP RD communication channels as well as that of other EJP RD partners
- Organisation of a **follow up workshop** in year 4 with the members of the working group **to consolidate the first version of the guide** adding learnings from the JTCs of 2019-2021 and the webinar(s).

## WP6 - Joint Transnational Calls for collaborative research projects

- The possibility to pay PAO experts with the remaining budget from evaluation meetings not organised due to Covid-19 crisis

# New actions foreseen – Pillar 2

- ✿ Moving from a Centralised VP to Federated Query & Access VP with:
  - ✿ catalogues for RD
  - ✿ genomics and multi-omics data deposition and analysis resources,
- ✿ Extend the VP to:
  - ✿ infrastructures providing support to clinical and translational research
  - ✿ additional genomics and multi-omics data deposition and analysis resources
  - ✿ resources providing animal models and cell lines
- ✿ FAIRification: Interoperability Data stewards Team set-up and service deployment
- ✿ **Virtual Platform Specification (VIPS) document** that describes and specifies the Overall Architecture (OA) of the EJP RD Virtual Platform (*extendable as the understanding and agreements of the VP design and implementation principles evolve*)
- ✿ Setting up all components/services for testing and validation: **Connectathon(s)**

# New actions foreseen – Pillar 3

## WP15, Task 15.4 – Educational materials and activities for paediatric patients

- 🌟 New educational programme for paediatric patients to empower them to take an active role in decision-making process related to research and to play an advisory role in the drug development process
- 🌟 Paediatric patient experts training course will be organized every year and addressed to 12-18 years' patients with chronic rare diseases
- 🌟 Training plan tailored for young people will be developed by M30

## WP18 – New needs in training

- 🌟 Development of new training courses to fill the gaps in alignment with progress of work and increase access to underserved groups
- 🌟 Identification of needs & gaps in capacity building arising from the activities of Pillar 2 and 4 and ERNs
- 🌟 Opportunity to gather training needs from all pillars will be discussed at the GA meeting

# New actions foreseen – Pillar 4\_WP19

## **Task 19.1: Accelerating translation**

- ✿ Integration of the ODDG into the Innovation Management Toolbox and Pillar 2 Virtual Platform
- ✿ Mentoring of JTC 2020 funded projects

## **Task 19.2: Support in exploitation and follow-on funding**

- ✿ Development of an exploitation plan for all the assessed projects

## **Task 19.4: Roadmap for a European investment platform for RD**

- ✿ Development of the roadmap for EU investment platform for RDs

## **Task 20.4: Projects on innovative methodologies to improve RD clinical trials in limited populations**

- ✿ Organisation of networking events for the creation of a consortia for the innovation projects
- ✿ Implementation of the selected innovation projects

## **Task 20.5: Educational program to disseminate advanced statistical trial methodology**

- ✿ Disseminate knowledge on basic statistical clinical trials methodology
- ✿ Create an education program on advanced statistical trial methodologies suited for rare disease clinical trials (workshops, webinars).

# Optimisation of work – synergies between WPs

# Potential synergies between WPs

- 🌟 Patient engagement in research: collaboration of **WP6** and **P3** for development of a training?
- 🌟 **WP19** mentoring of Pillar 1 funded projects (**WP6**)
- 🌟 Target Pillar 1 funded projects (**WP6**) to specific trainings from **P3** based on the call topic with the help of **WP5**
- 🌟 **Pillar 2** Training on Virtual Cluster Environments (**WP12**) and how to use available tools and pipelines to come to a variant interpretation → **WP17 & WP18/WP14**
- 🌟 Work on FAIRification and data deposition (**WP12**) with Pillar 1 funded projects (**WP6**) (particularly those generating multi-omics and liaison with **WP13**)
- 🌟 Innovative Clinical Trial (CT) Methodologies (**WP20**) and registry data access through VP (e.g. adaptive [Bayesian] design, missing data, control arms, etc.) (**WP12**)
- 🌟 **WP20** innovative CT methodologies and **WP13** multi-omics networks (drug targets <-> Basket and Umbrella Trials)
- 🌟 Continuation of "Machine Readable and Computable Consent Models" Task Force (**EJP RD & IRDiRC joint action**) through **Pillar 2** Use Case Work Focus Team in liaison with GA4GH Regulatory and Ethics Work Stream
- 🌟 **New IRDiRC Task Forces** "Shared Molecular Etiologies (SaME)", "Integrating New Technologies for the Diagnosis of Rare Disease" and **WP13**
- 🌟 **WP20** Education proposal on innovative methodologies → **WP17 & WP18**
- 🌟 **WP20** training support/workshop for Clinical Trials → **WP7 & WP17**

# Optimisation of work – COVID-19 mitigation measures

# Actions (taken or planned) related to COVID-19 mitigation measures

- 🌟 **For the majority of the WPs:** re-organisation of Face to Face meetings into online events
- 🌟 **WP7 - Networking to share knowledge on rare diseases:** Due to the Covid-19 crisis, the format of the events funded through this scheme are enlarged to online events
- 🌟 **Pillar 2 (Annual Retreat):** development of methodology on the set-up of online Events through Ms Teams
- 🌟 **Possibility of Centralised Support:**
  - 🌟 Workshop/events **centralised tool** available for EJP RD partners and funded workshops (e.g. WP7, WP17)



# Optimisation of work – GO/NO GO for activities failing to deliver

# How do we deal with planned activities that are not working as expected?

## Discussion on:

- What are the mitigation measures implemented by different Pillars / Work Packages?
- When do you decide that the Go / No Go decision is necessary?

# Introduction to financial optimisation

# Current status of the person-months used in Y1

01/01/2019-31/12/2019	Pillar 0	Pillar 1	Pillar 2	Pillar 3	Pillar 4	Total
<b>Actual Person-months declared</b>	<b>121,94</b>	<b>47,79</b>	<b>345,80</b>	<b>75,89</b>	<b>86,63</b>	<b>678,05</b>
<b>Planned Person-months for year 1</b>	159,70	57,60	593,00	95,80	124,00	1 030,10
<b>Actual compared to planned year 1</b>	76%	83%	58%	79%	70%	66%
<b>Planned Person-months for 5 years</b>	721,66	209,38	2 923,73	473,50	607,00	4 935,27

 During the 1<sup>st</sup> year, the EJP RD consortium spent:
 

-  66% of the person-months planned for year 1;
-  14% of the global person-months planned (5 years).

# Current status of the costs incurred in Y1

01/01/2019-31/12/2019	ACTUAL COSTS DECLARED (IN EUROS)	PLANNED COSTS YEAR 1 (IN EUROS)	ACTUAL COMPARED TO PLANNED YEAR 1	COSTS PLANNED FOR 5 YEARS (IN EUROS)
Personnel costs	3 745 495	6 094 794	61%	30 473 970
Travels	368 731	437 094	84%	2 185 470
Other Goods and services	185 221	1 860 342	10%	9 301 710
Subcontracting	15 600	116 000	13%	580 000
Direct costs of providing financial support to third parties (e)	849 623	8 303 000	10%	41 515 000
Indirect costs	1 074 862	2 093 557	51%	10 467 787
<b>Total costs</b>	<b>6 239 531</b>	<b>18 904 787</b>	<b>33%</b>	<b>94 523 937</b>

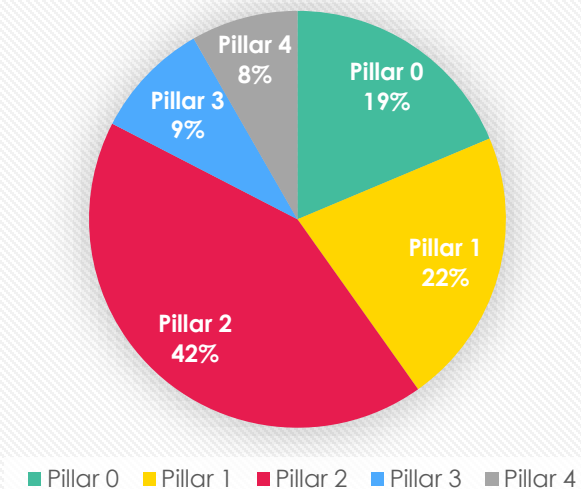
✚ During the 1<sup>st</sup> year, the EJP RD Consortium spent:

- ✚ 33% of the budget planned for year 1: with achievement of **84% of planned travels and 61% of planned personnel costs.**
- ✚ 7% of the total budget planned (<20%): with 17% of travels and 12% in personnel costs.

# Current status of costs incurred per pillar in Y1

Costs category	Pillar 0	Pillar 1	Pillar 2	Pillar 3	Pillar 4	Total Actual costs
Personnel costs	617 287	289 817	2 038 645	397 583	402 162	3 745 495
Travels	183 044	77 475	53 921	42 669	11 623	368 731
Other Goods and services	131 666	26 483	11 730	14 917	425	185 221
Subcontracting	0	0	15 600	0	0	15 600
Direct costs of providing financial support to third parties (e)	0	849 623	0	0	0	849 623
Indirect costs	232 999	98 444	526 074	113 792	103 553	1 074 862
<b>Total costs</b>	<b>1 164 996</b>	<b>1 341 842</b>	<b>2 645 969</b>	<b>568 961</b>	<b>517 763</b>	<b>6 239 531</b>

Costs per pillar Y1



# Reminder on use of travel costs

## Travel budgets identified:

- ✘ All partners received a budget of 5 900 € to travel to GA meetings (5 meetings in total; 100% reimbursement rate)
- ✘ ExCom members received budget to travel to ExCom meetings (5 meetings + kick off; 100% reimbursement rate)
- ✘ Partners involved in P2 received budget to travel to the Annual Retreats (5 meetings; 70% reimbursement rate)

**There is no additional budget for travels to any extra meetings foreseen**

**The coordination is verifying the eligibility of travel costs for each annual reporting**

**If you are planning to attend the meeting and finance your travel costs from EJP RD budget always check with the coordination in advance**

## Possible choices to cover additional travel costs

- ✘ From external resources (other project (if eligible); institutional budget)
- ✘ By increasing in kind contribution & using part of budget dedicated to personnel

ATTENTION: we want to avoid any situations where the majority of the budget of a partner is used for travels!

# Conclusions on year 1

Under use of person-months and personnel costs to be addressed in the annual work plan(s) and reorganisation of costs:

☘ Pillar 0: WP2

☘ Pillar 1: WP7 & WP9

☘ Pillar 2: WP12 & WP13

☘ Pillar 3: WP16 & WP17

☘ Pillar 4: WP20

01/01/2019-31/12/2019	WP2	WP7	WP9	WP12	WP13	WP16	WP17	WP20
Planned PM Y1	19,70	6,90	3,00	251,00	54,70	24,00	17,00	62,00
Actual PM Y1	6,23	2,21	1,48	129,20	31,24	9,27	3,50	33,19
Difference Planned-Actual	13,47	4,69	1,52	121,80	23,46	14,73	13,50	28,81
Achievement %	32%	32%	49%	51%	57%	39%	21%	54%

☘ /!\ Individual Travel costs

☘ Other Goods and services underspending: planning and reorganisation necessity

☘ Impact on payments : EC payments are based on actual costs reported, we can expect that the third payment will be reduced and partners will have to advance payments for Y4



# Identified remaining 2020 budget from AWP2 due to COVID-19

Other Goods and services planned budget 2020	Pillar0	Pillar1	Pillar2	Pillar3	Pillar4	TOTAL
INSERM (Coord)	135 000	70 000			35 000	305 000
FFRD	20 000			564		20 564
FTELE				3 154		3 154
ISS			12 480	22 230		34 710
UMCG			10 000			10 000
UM			11 750			11 750
MUG				7 000		7 000
ISCI				4 500		4 500
CNAG-CRG			5 500			5 500
ULEIC			5 800			5 800
GUF			1 315			1 315
LUMC			14 750			14 750
RUMC			17 500			17 500
AMC			4 300			4 300
BBMRI				5 200		5 200
EATRIS					800	800
ELIXIR			6 414			6 414
EURORDIS				16 045		16 045
BSF	115 000					115 000
ACU/ACURARE				3 500		3 500
<b>TOTAL costs planned</b>	<b>270 000</b>	<b>70 000</b>	<b>89 809</b>	<b>62 193</b>	<b>35 800</b>	<b>527 802</b>
Costs incurred before COVID-19	131 000	7 000		36 000		174 000
<b>Remaining Other Goods &amp; Services Budget 2020</b>	<b>139 000</b>	<b>63 000</b>	<b>89 809</b>	<b>27 193</b>	<b>35 800</b>	<b>354 802</b>

# What can be done with the remaining budget?

🌟 Pillar 1, WP6 SEC meetings : remaining budget **can be used for the payment of PAO experts in further calls.**

- 🌟 Need internal pillar approval
- 🌟 Transfer explanation note/table to COORD
- 🌟 Application in the internal budget

**: remaining budget can be kept at pillar 1 level as a “reserve” envelope** but not directly attached to a participant and could be used to implement a 5th call if possible. The remaining budget can be distributed within pillar 1 after declaration of actual Costs upon approval of EC and PL Leaders.

🌟 Pillar 2, Workshops activities & Pillar 2 annual retreat : **remaining budget can be kept at pillar 2 level as a “reserve” envelope until redistribution of budget internally.**

🌟 Pillar 3, Trainings activities: **remaining budget can be kept at pillar 3 level as a “reserve” envelope** but not directly attached to a participants. The remaining budget can be distributed within pillar 3 after declaration of actual costs upon approval of EC and PL Leaders.

🌟 Pillar 4, WP20 SEC meetings: **remaining budget can be kept at pillar 4 level as a “reserve” envelope** but not directly attached to the initial beneficiary. The remaining budget can be distributed within pillar 4 after declaration of actual costs upon approval of EC and PL Leaders.

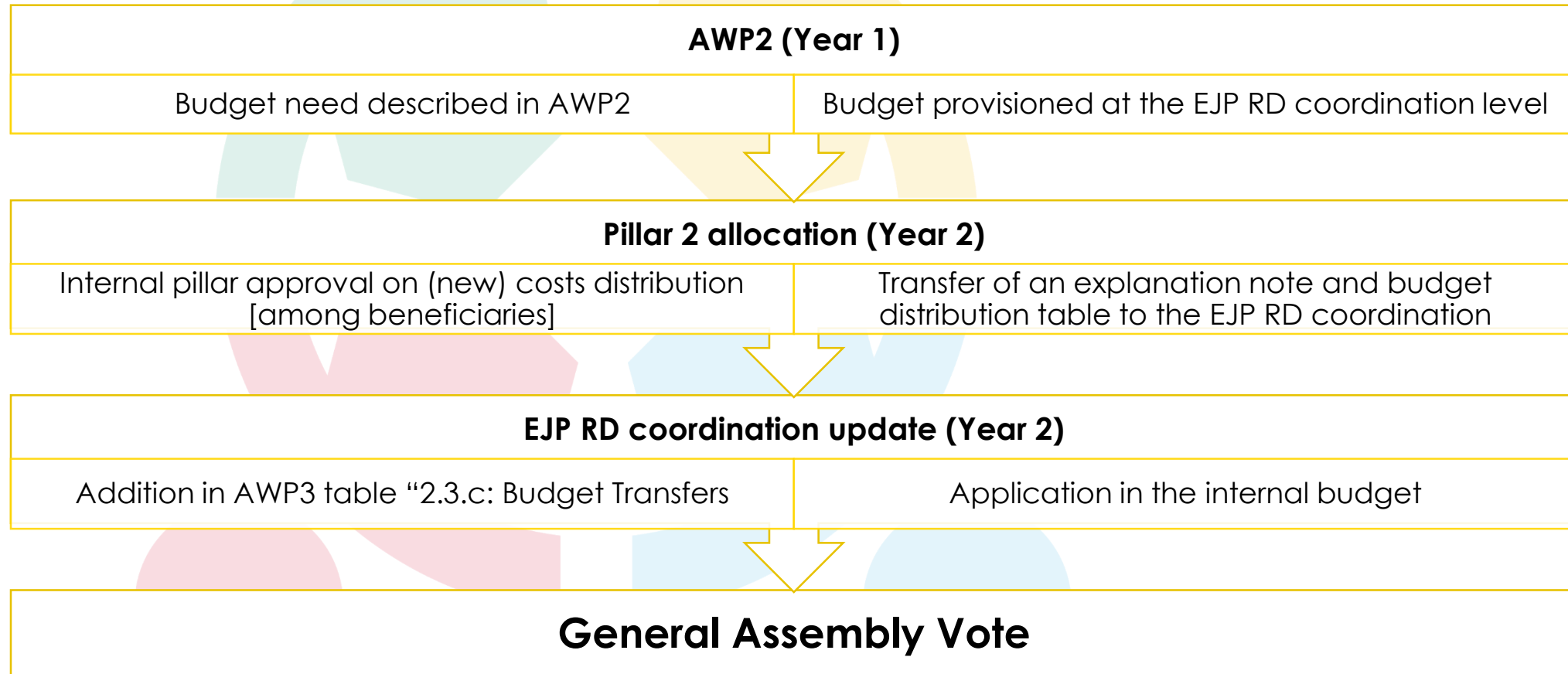
🌟 **Changes need to be integrated in the annual work plan and internal budget and approved by the General Assembly**

# What to do when you want to re-allocate the budget from one/multiple partner(s) to another?

- 🌟 Transfers of amounts between beneficiaries or between budget categories (or both) do NOT require an amendment, provided that the **action is implemented in line with Annex 1 (no changes to the action, it is not new activity)**
- 🌟 **However, they need to be implemented in the internal budget, in the annual work plan(s) and voted by the General Assembly**
- 🌟 Actions:
  - 🌟 Partner(s) discuss issue with WP Leaders and PL Leaders
  - 🌟 When approved by Pillar Leaders, write an explanation note to coordination unit with pillar leaders in cc.
  - 🌟 Introduction in the annual work plan and internal budget **to be voted in the General Assembly**

# What to do when the budget validated in the previous AWP for specific activities needs to be re-allocated during the year with the start of these activities?

Example for the Subtask “12.3.3 FAIR data stewardship service”



# Pillar 2 ERN & RD Researchers surveys

- *"deep dive" and exploitation* -

Mary Wang (FTELE)

7-July-2020

# Rare disease research community – who?



**European Reference Networks (ERNs)** are virtual networks involving healthcare providers across Europe with the aim to tackle complex or rare diseases. There are 24 thematic networks: over 900 specialised healthcare units located in 313 hospitals in 25 Member States (plus Norway).



Source: [https://ec.europa.eu/health/ern\\_en](https://ec.europa.eu/health/ern_en)



**Rare Disease researchers** are academic scientists in universities and research centres working on any research projects related to rare diseases. They can be working to understand disease mechanisms using disease models as well as translational research. Some EU-level research projects are funded by E-Rare and EJP RD Joint Transnational Calls.

**Stakeholders** being involved for Pillar 2 Use Cases Work Focus:

- ERNs
- Researchers
- Patient representatives
- Funding agencies

# Topic of the surveys

1. About the researcher, type of research and needs;
2. Data generation and storage;
3. Data annotation and FAIRification;
4. Use of existing resources and infrastructures for research data

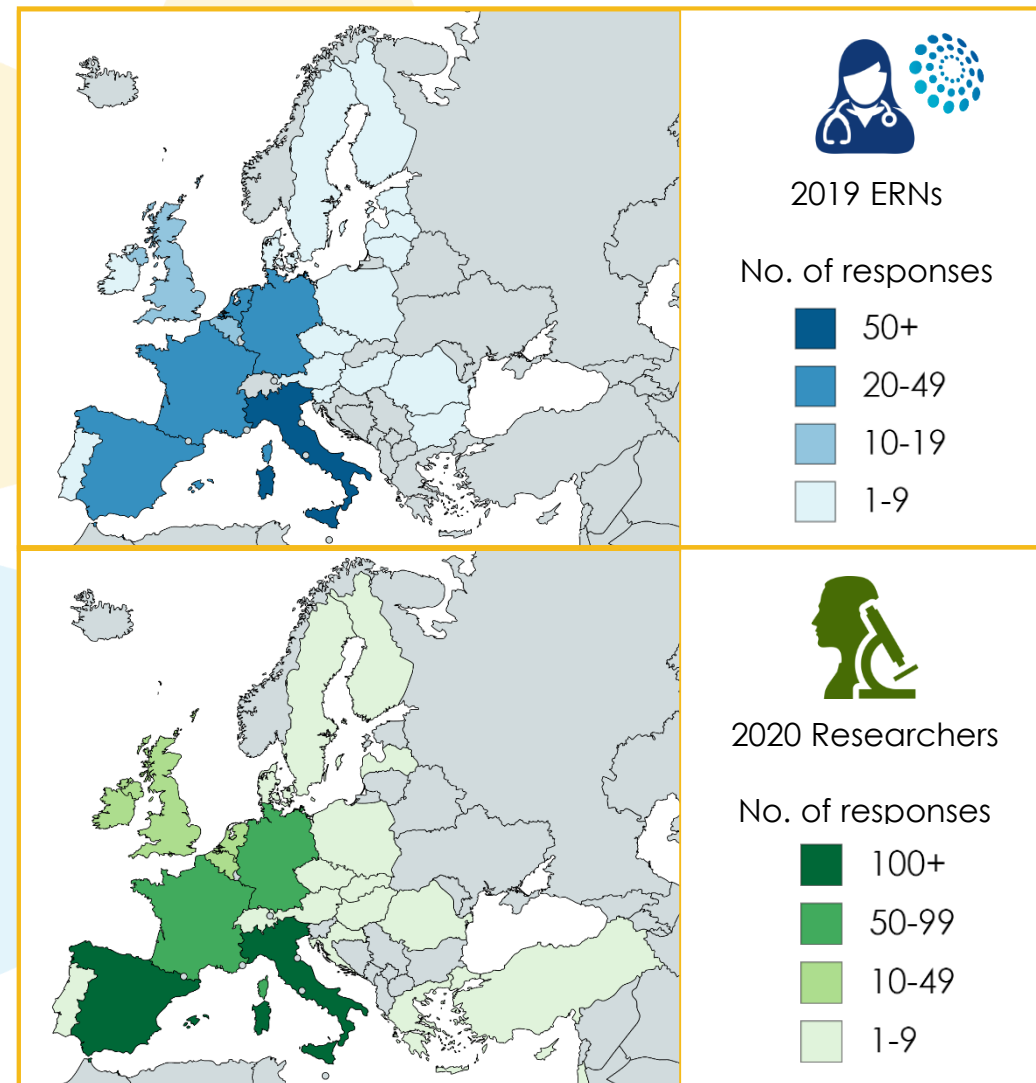
# Research Community Surveys

## 2019 ERN

- 🌈 Surveyed 25/3 – 11/4 2019 (17 days)
- 🌈 73 questions (P2, P3, P4)
- 🌈 **291 responses** received; 30.1% response rate
- 🌈 All 24 ERNs responded to the survey

## 2020 Researcher

- 🌈 Surveyed 27/3 – 21/4 2020 (25 days)
- 🌈 47 questions (P2 only)
- 🌈 **451 responses** received from 31 countries
- 🌈 83% of respondents are Principle Investigators

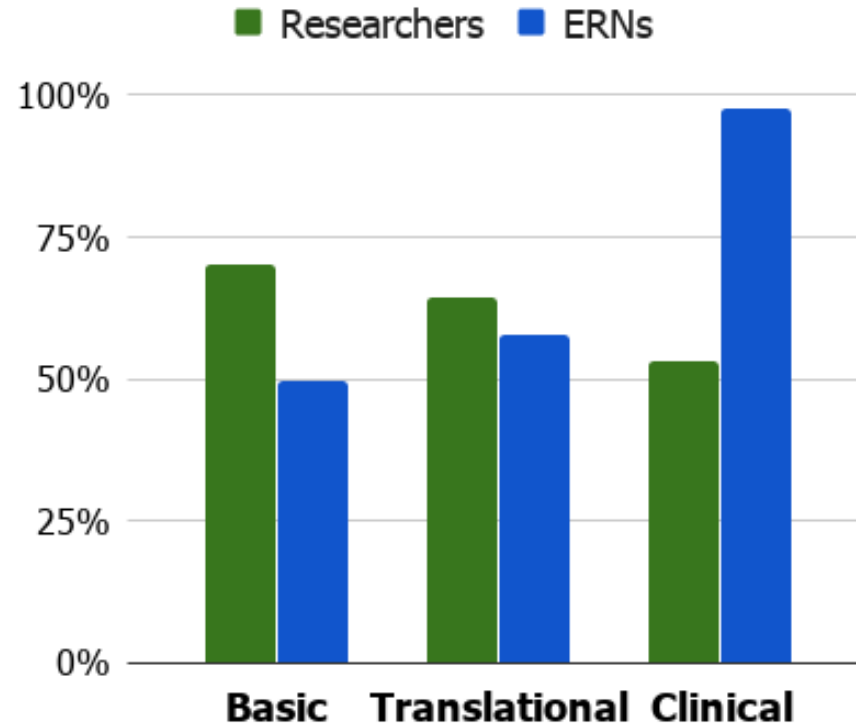
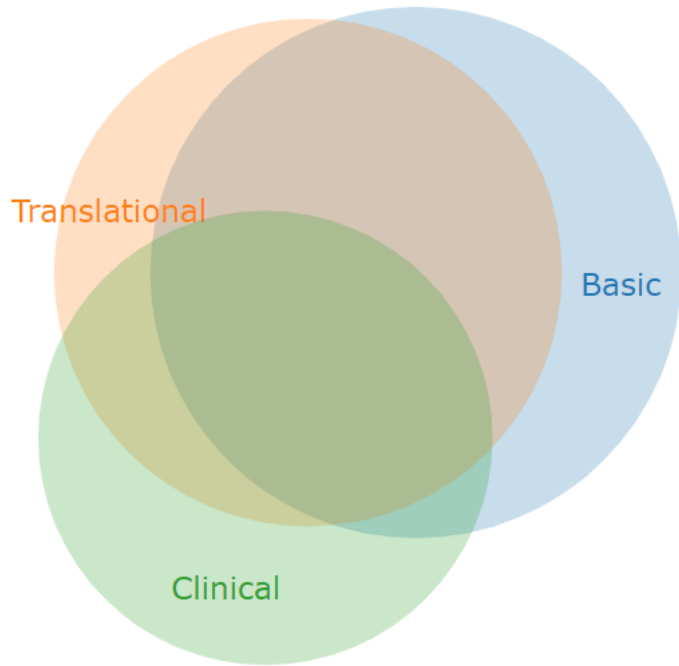


Special mentions: Yanis Mimouni, Juliane Halftermeyer, Eleonora Passeri, Daria Julkowska, Franz Schaefer, Ana Rath, Florence Guillot, H el ene Le Borgne

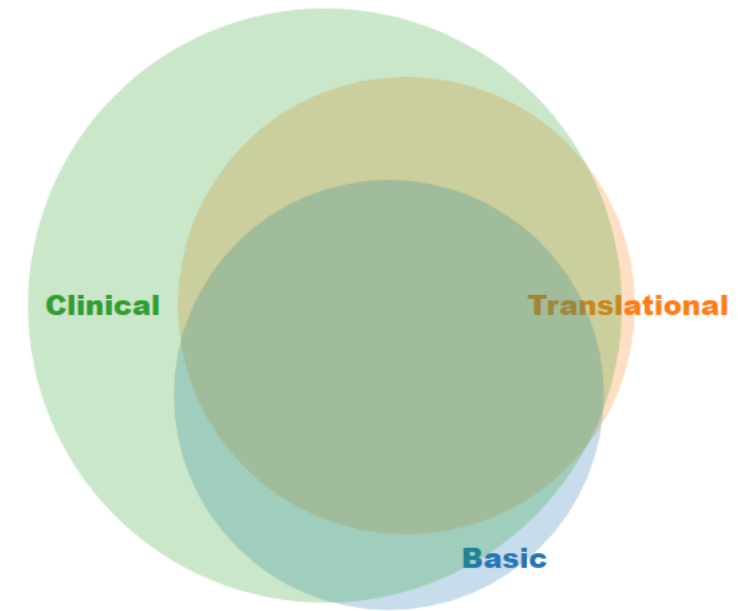


# Types of research performed by ERNs and researchers

2020 Researchers

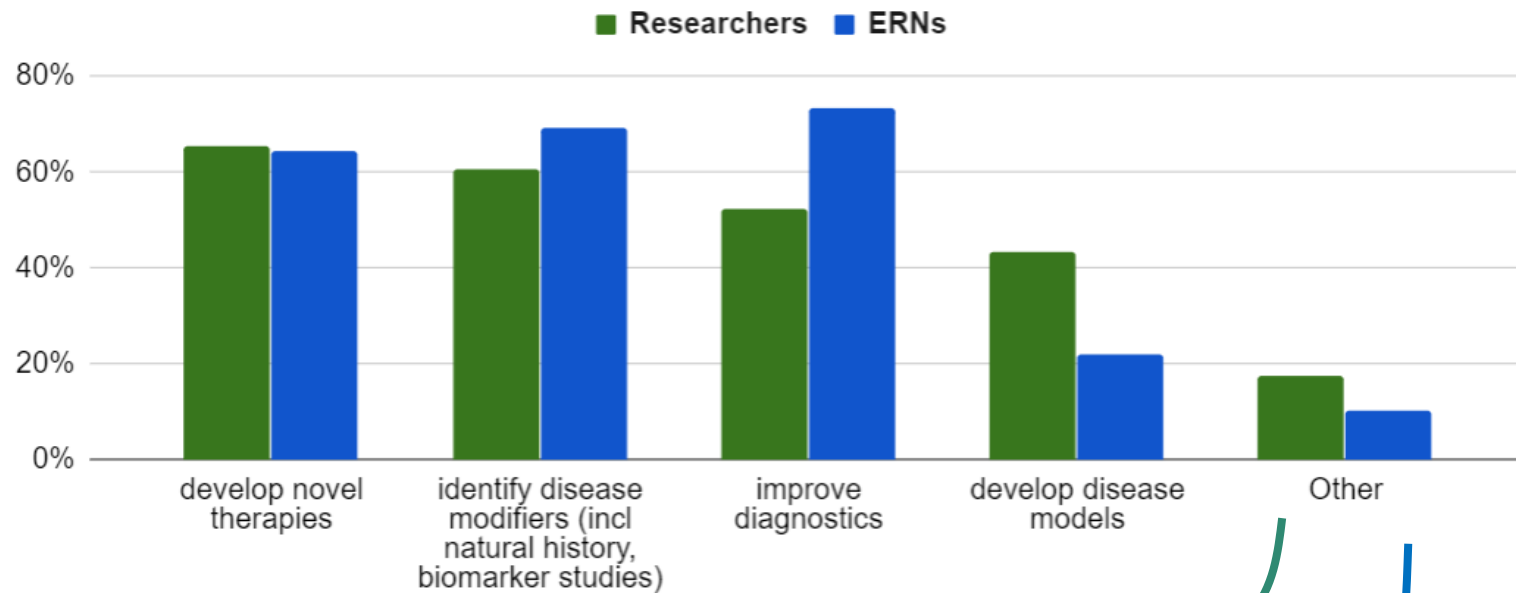


2019 ERNs



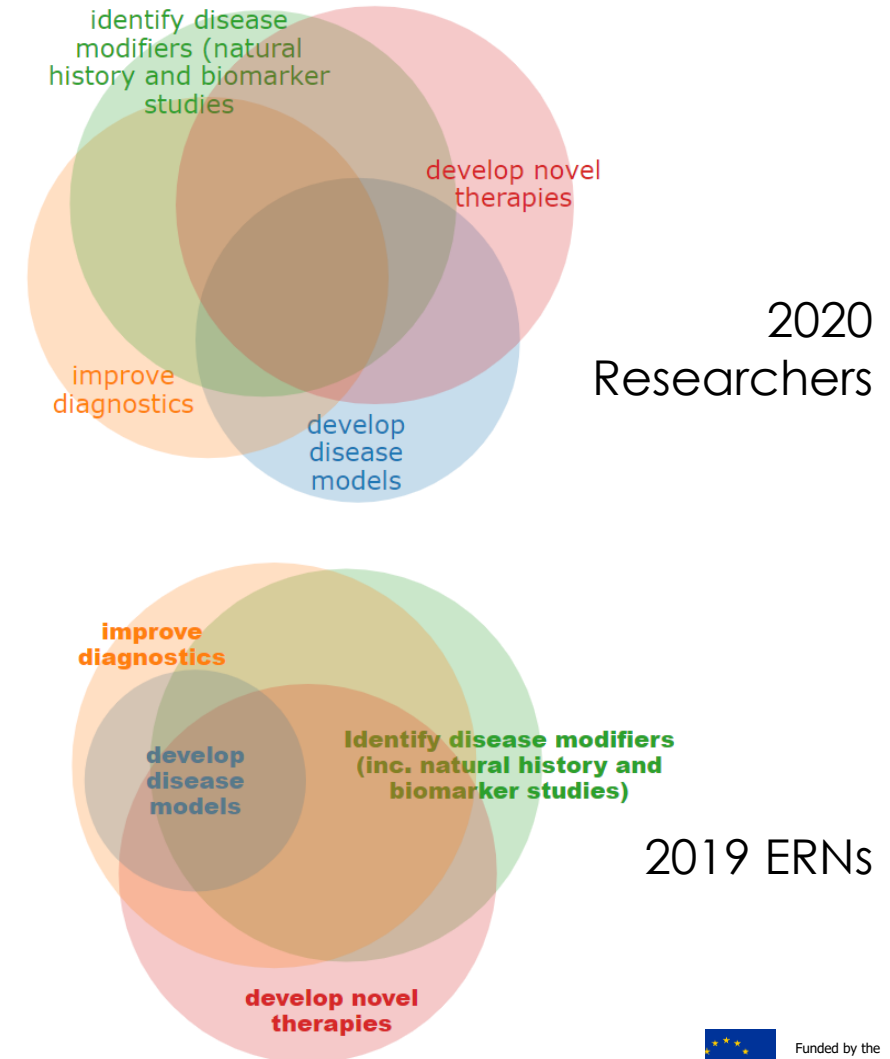
Researchers are a lot more heterogeneous in terms of research performed. ERNs appear more homogeneous.

# The main purpose of the current research



Understanding disease mechanism

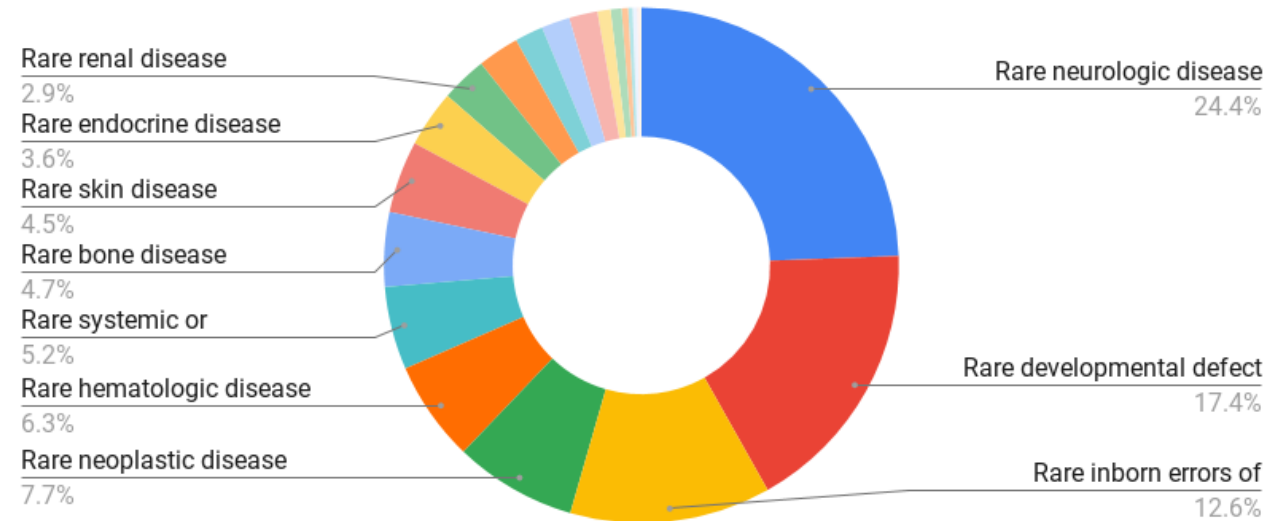
Innovative/improve care



# Which rare diseases do researchers work on?

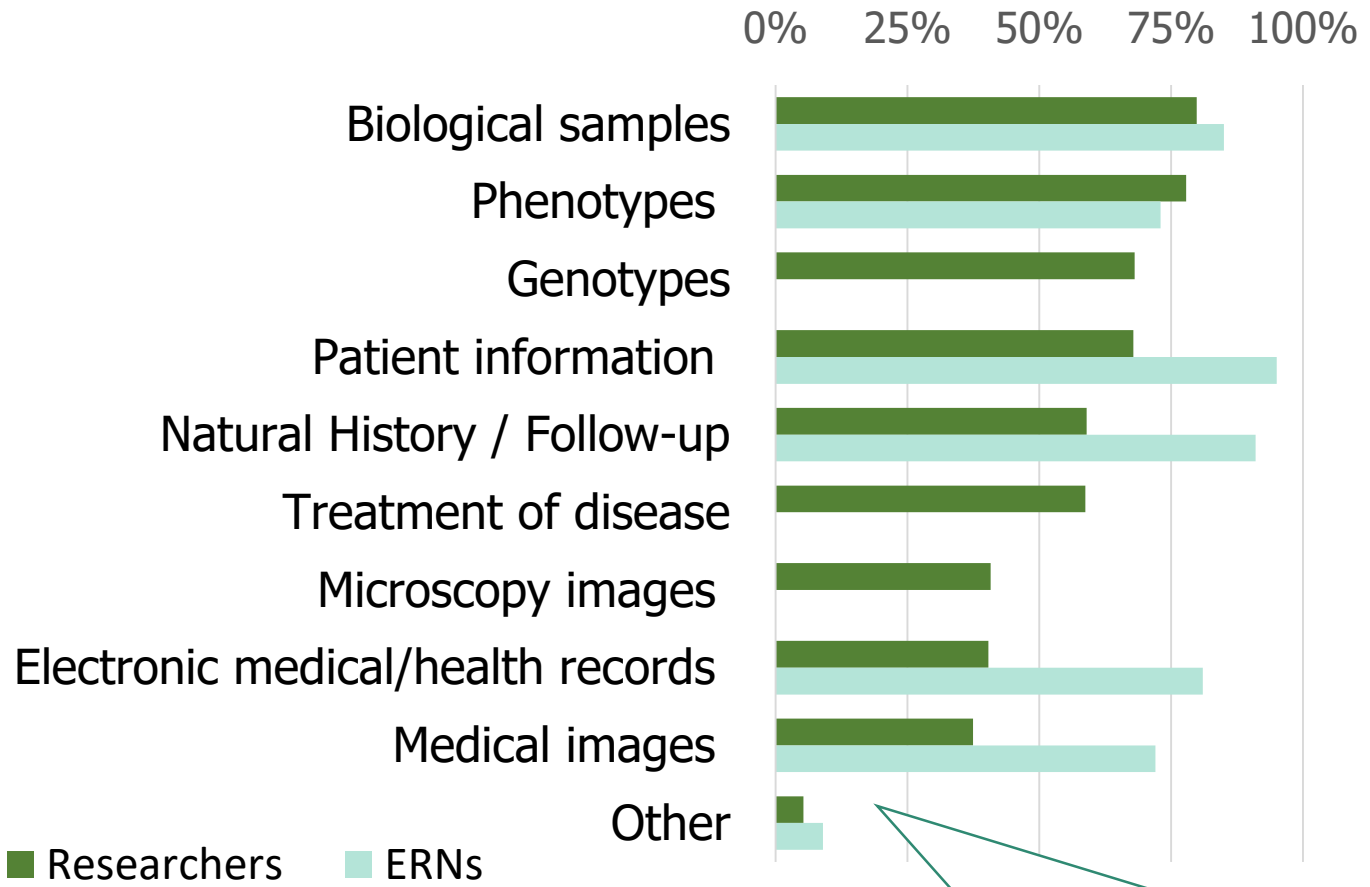
- Total of **629 disorders** and **109 disease groups** mentioned
- These diseases and groups map to **21 disease classifications** out of the 35 Orphanet Disease Classification
- 54.5% of diseases mentioned fall in 3 major disease classifications:
  1. **Rare neurologic disease**
  2. **Rare developmental defect during embryogenesis**
  3. **Rare inborn errors of metabolism**

The rare diseases researchers work on



# Data generation

# Which of the following data types do you collect or use?



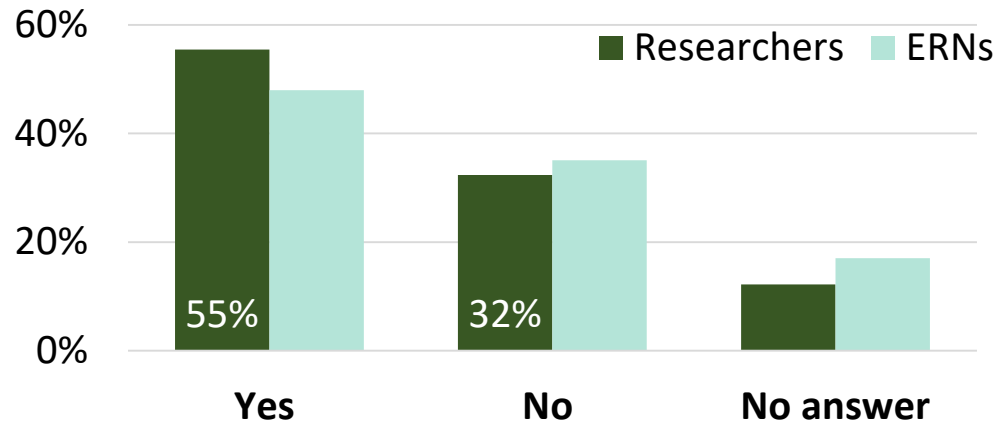
Researchers use fewer data types related to patient health.

Researchers use biological samples, phenotype and genotype data.

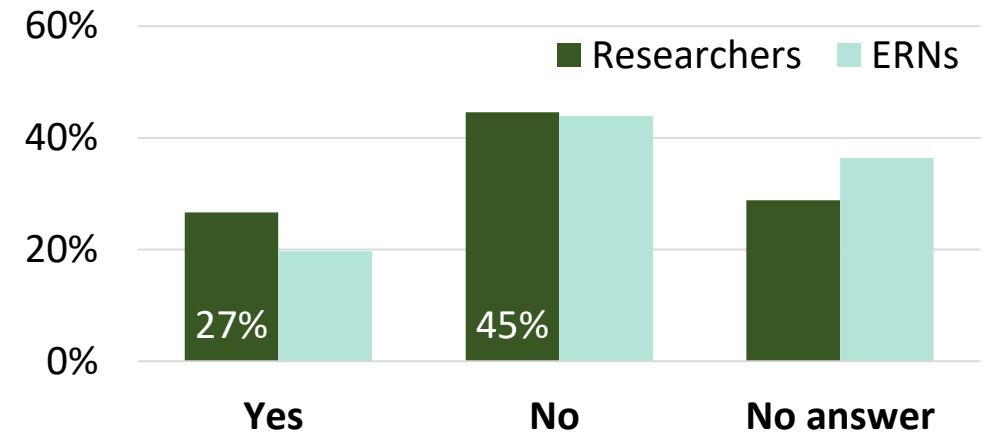
Behavioural data, molecular structures, Non-E HR, mouse data



## Do you generate or use –omics data?



## Do you have different types of –omics data that originate from the same sample, from multi-omic studies?

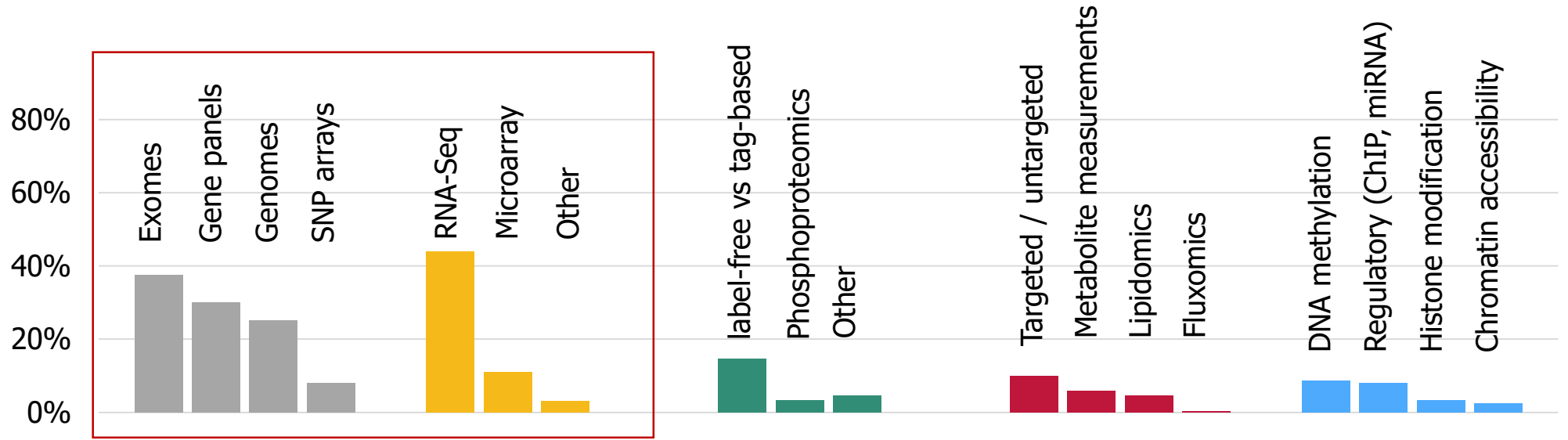


Over half of the researcher respondents generate or use –omics data. Under a third have data from multi-omic studies.

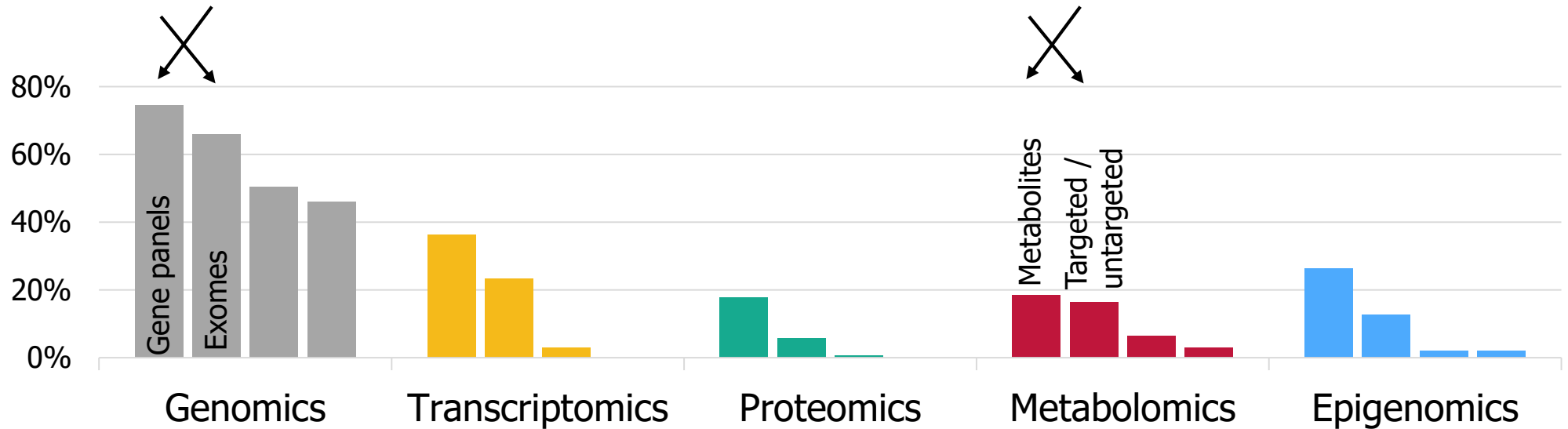
Similar responses from ERNs and researchers.

# If YES, which -omics data?

2020  
Researchers



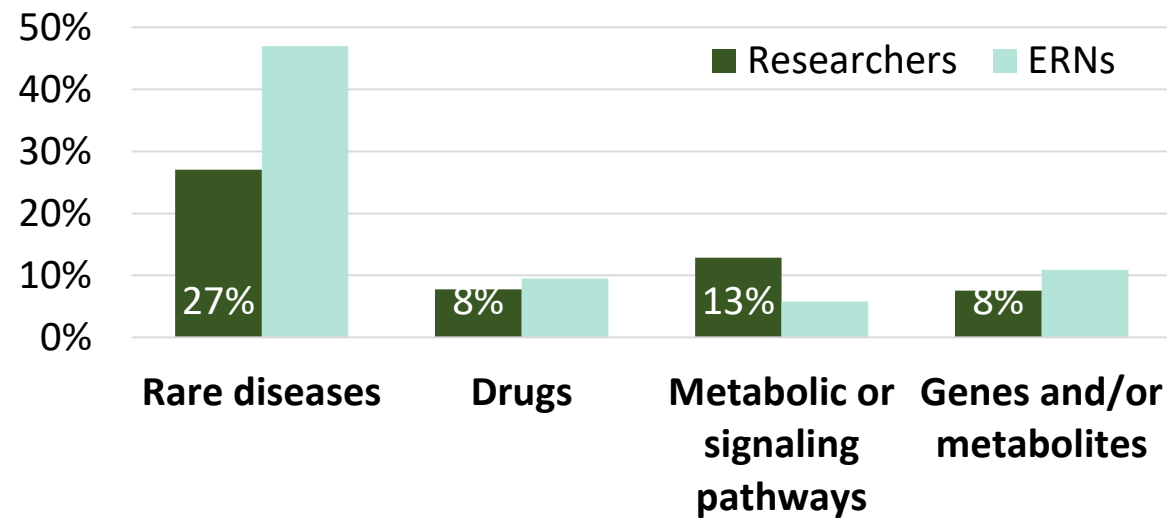
2019  
ERNs



The most common -omics generate/used are *genomics* and *transcriptomics*



# Do you have any data on diseases / risk factors / drugs / metabolic / signaling pathways?



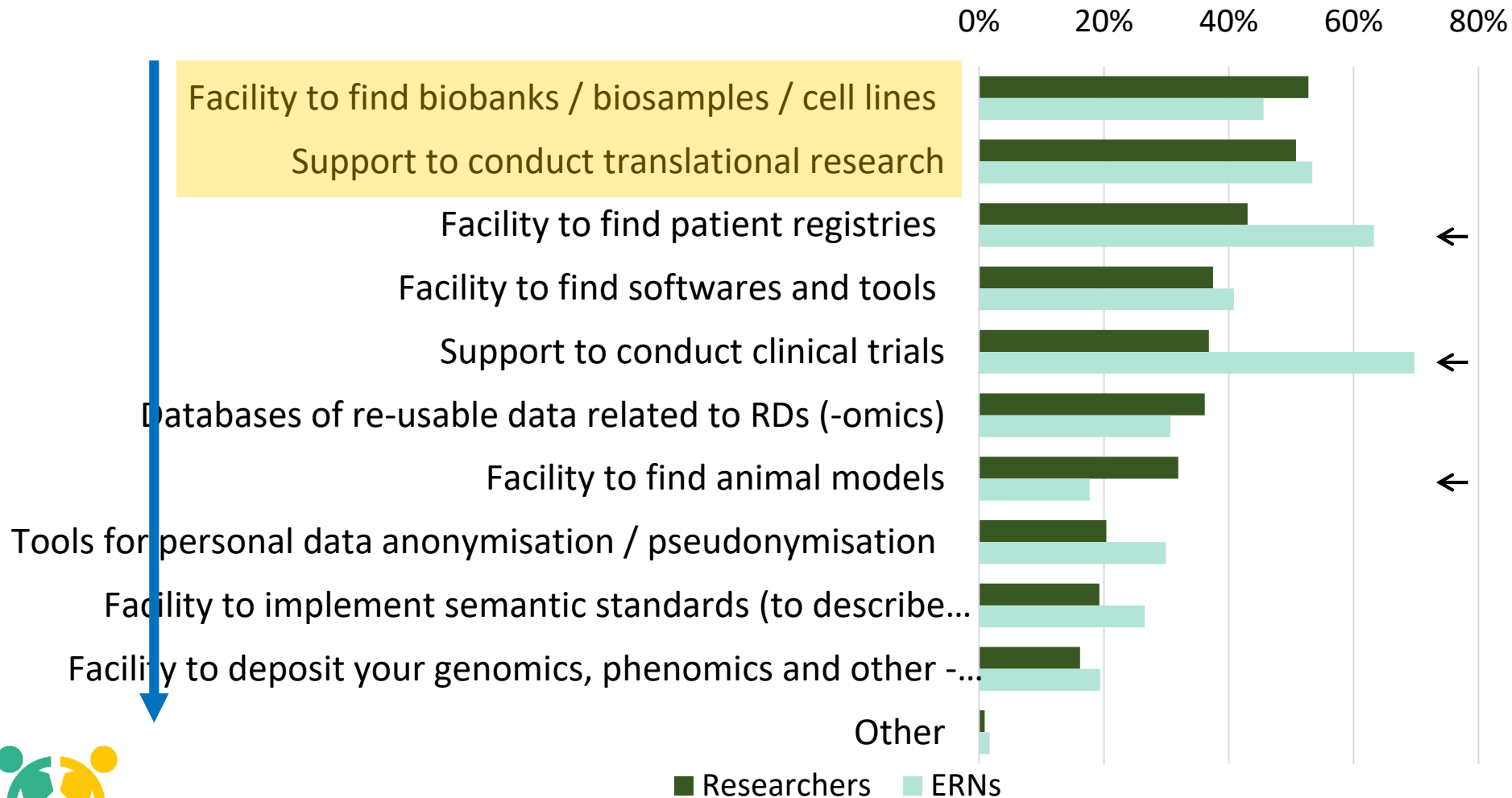
-> Could these data be added into an existing pathway or structured into a new one?

Yes 26%  
No 4%  
**31%**

I cannot judge if my data could fit an existing pathway or structured into a new one



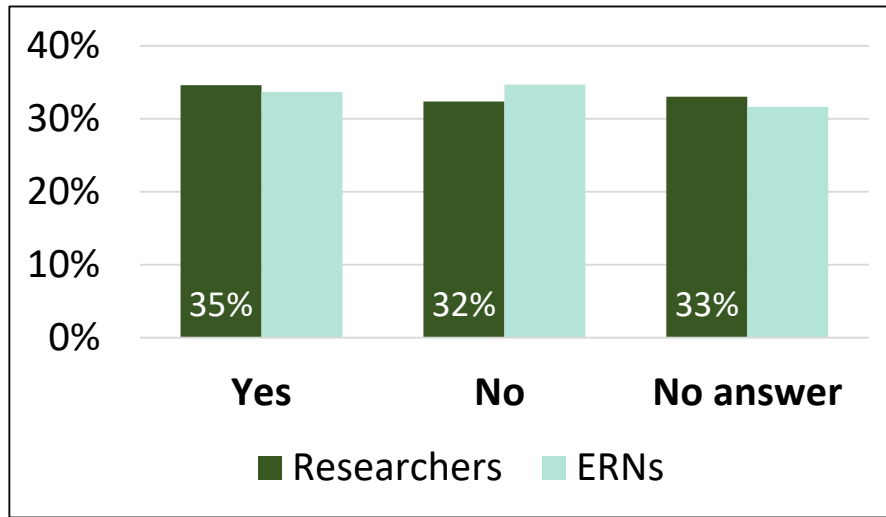
# Which of these services are of utmost importance for your research?



Researchers and ERNs require different services, this is most likely due to the differences in the type of research they perform.

# Data annotation and FAIRification

# Do you use ontologies or standards to annotate your data?



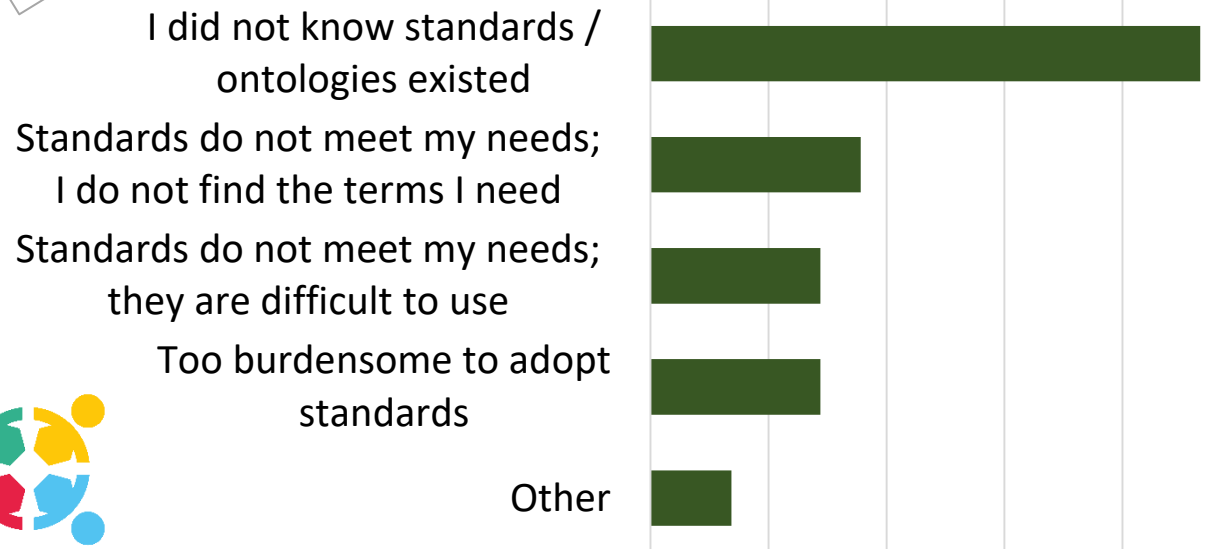
Yes

The top standards used by both researchers and ERNs are: OMIM, ICD, HPO and ORDO

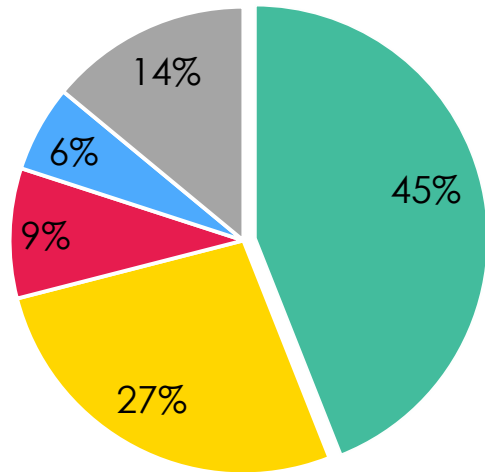
Need to improve awareness!

No

0% 10% 20% 30% 40% 50%



# Please describe your interest in making your research data FAIR?



I don't sufficiently understand what "FAIR research data" means.

I am interested, but I am facing some barriers to get involved.

I am in the process of FAIRifying my data.

I am currently not interested in this kind of activity.

Not applicable

## **What are these barriers?**

70% No resources for this effort  
 43% No software/servers to make the data discoverable  
 32% No consent/authority to make data more accessible  
 32% Don't know what standards to use

## **Stage of FAIRification?**

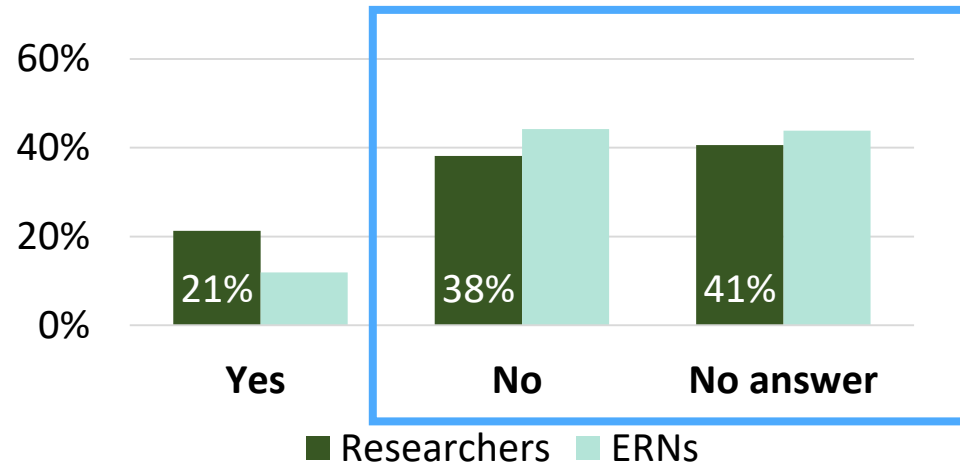
52% Initiation  
 19% Planning  
 33% Execution  
 2% Closed

## **What might raise interest?**

63% Seeing added value  
 24% If advantages outweigh drawbacks

# Use of existing resources and infrastructures for research data

## Do you usually deposit and/or share your -omics data in open or controlled access resources?

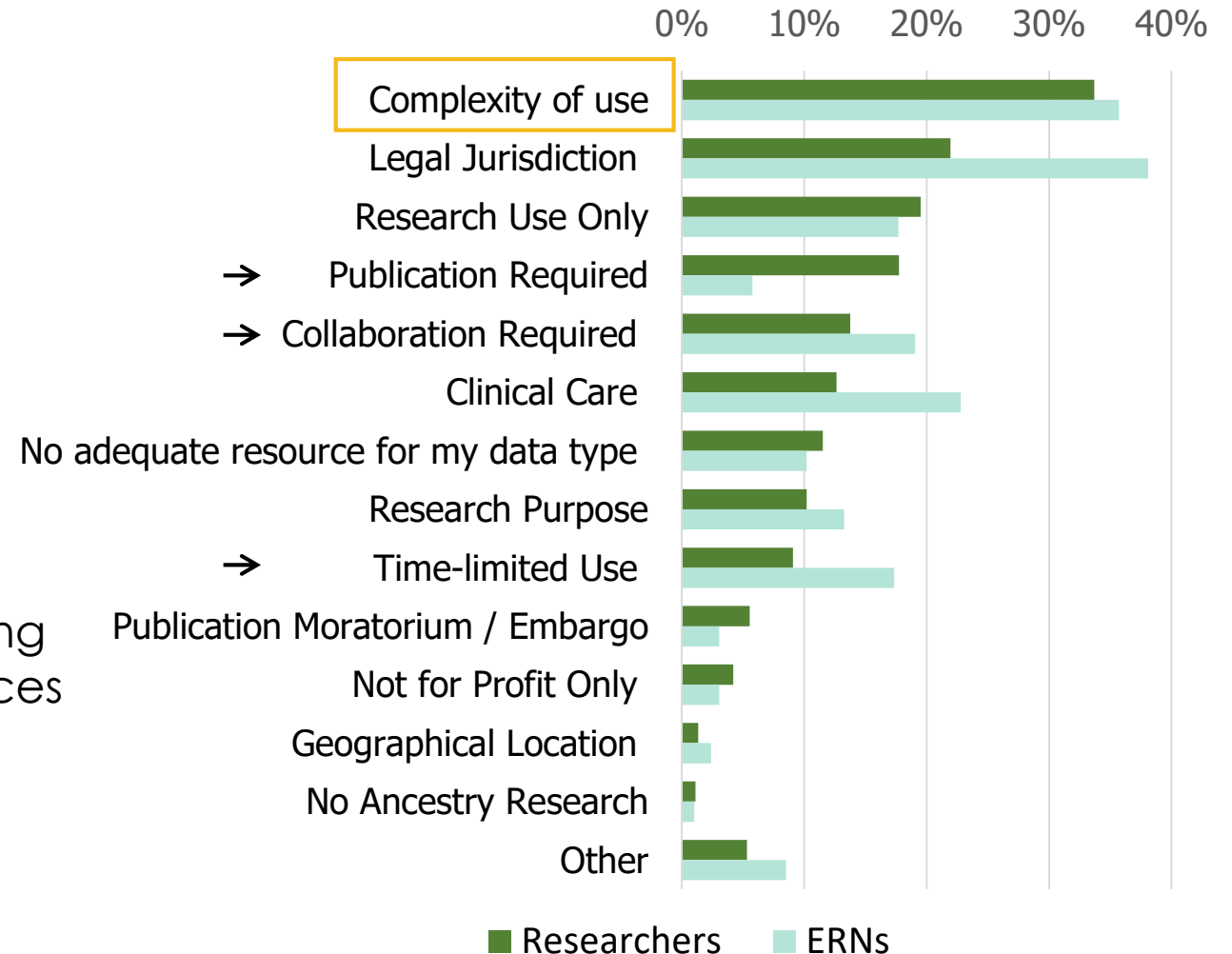


*Complexity of use* remains the most limiting factor to deposit or share data in resources

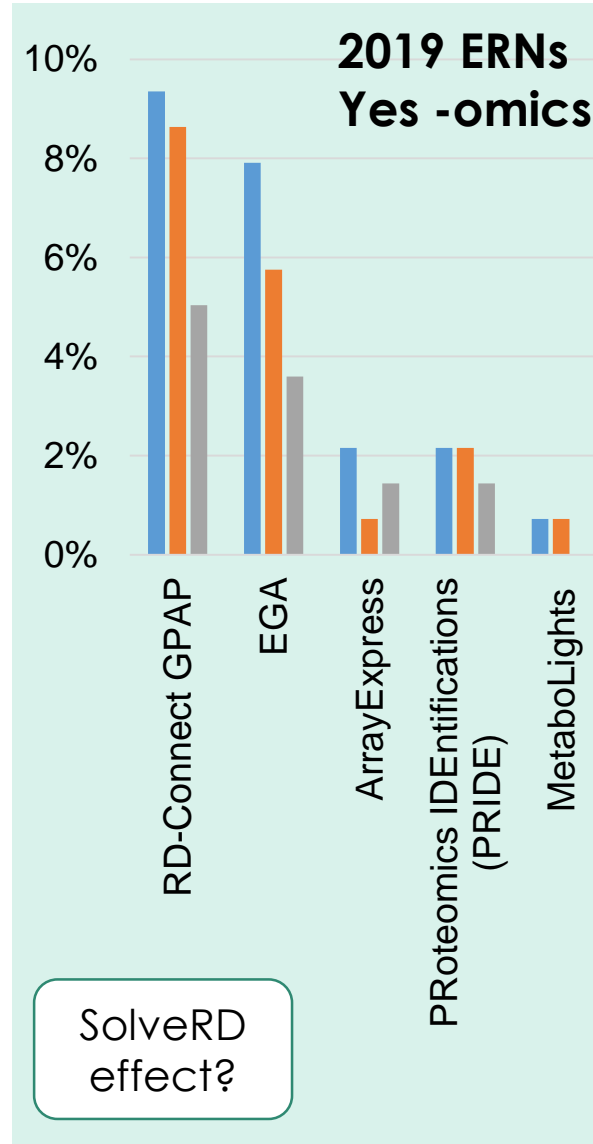
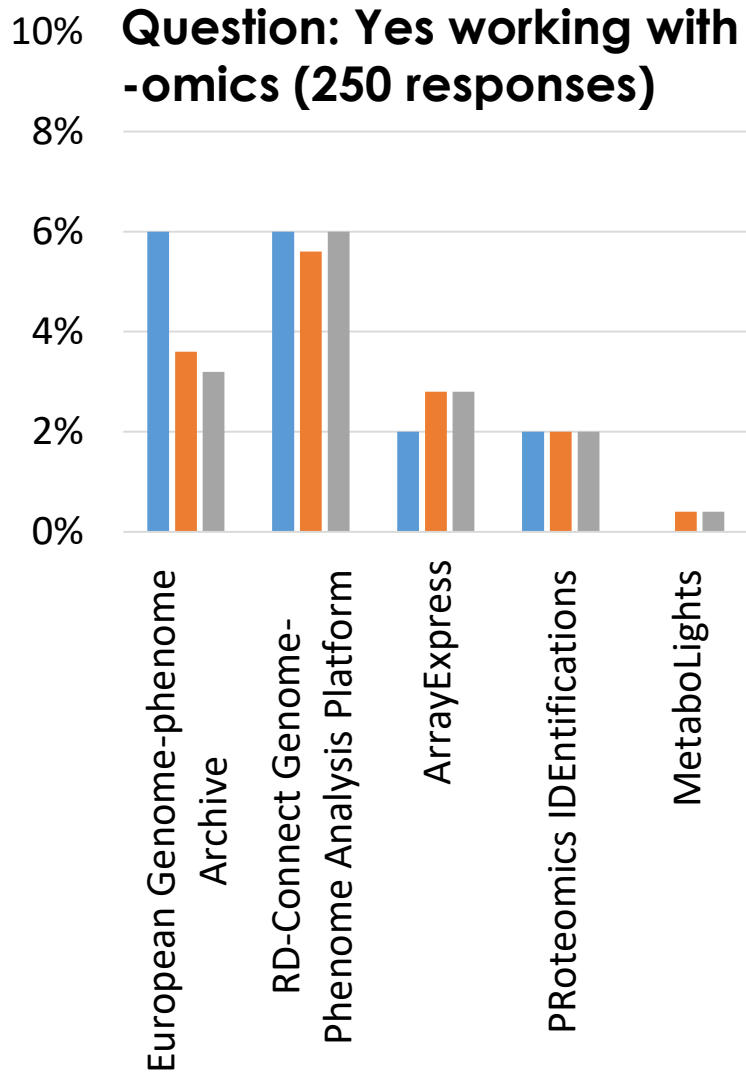
Make it simple!

How can we encourage sharing?

## What are the issues that are more limiting for you to deposit / share your data in such open or controlled access resources?



# Do you use any of the following resources as your primary data deposition / data sharing / data analysis mechanism?

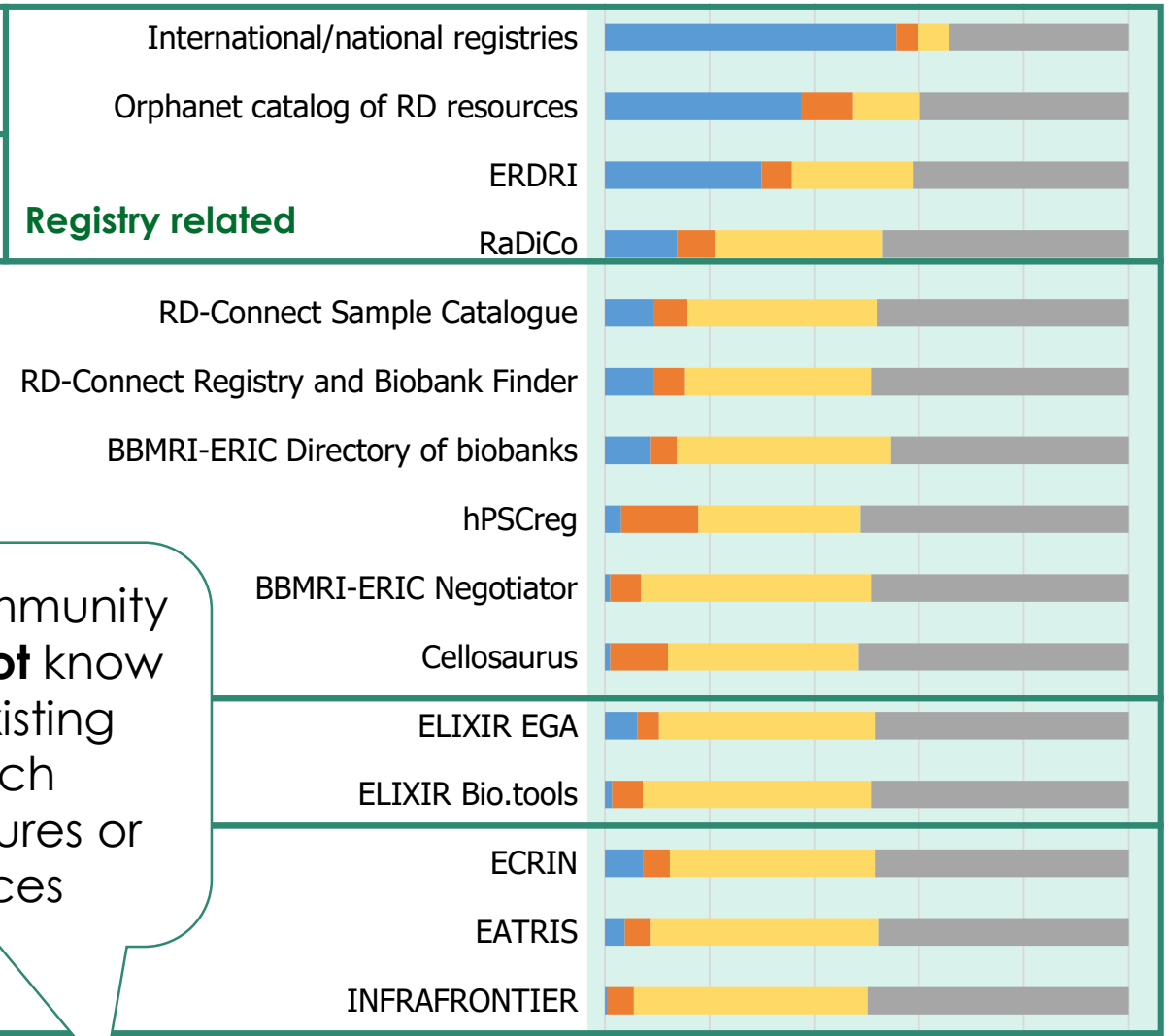
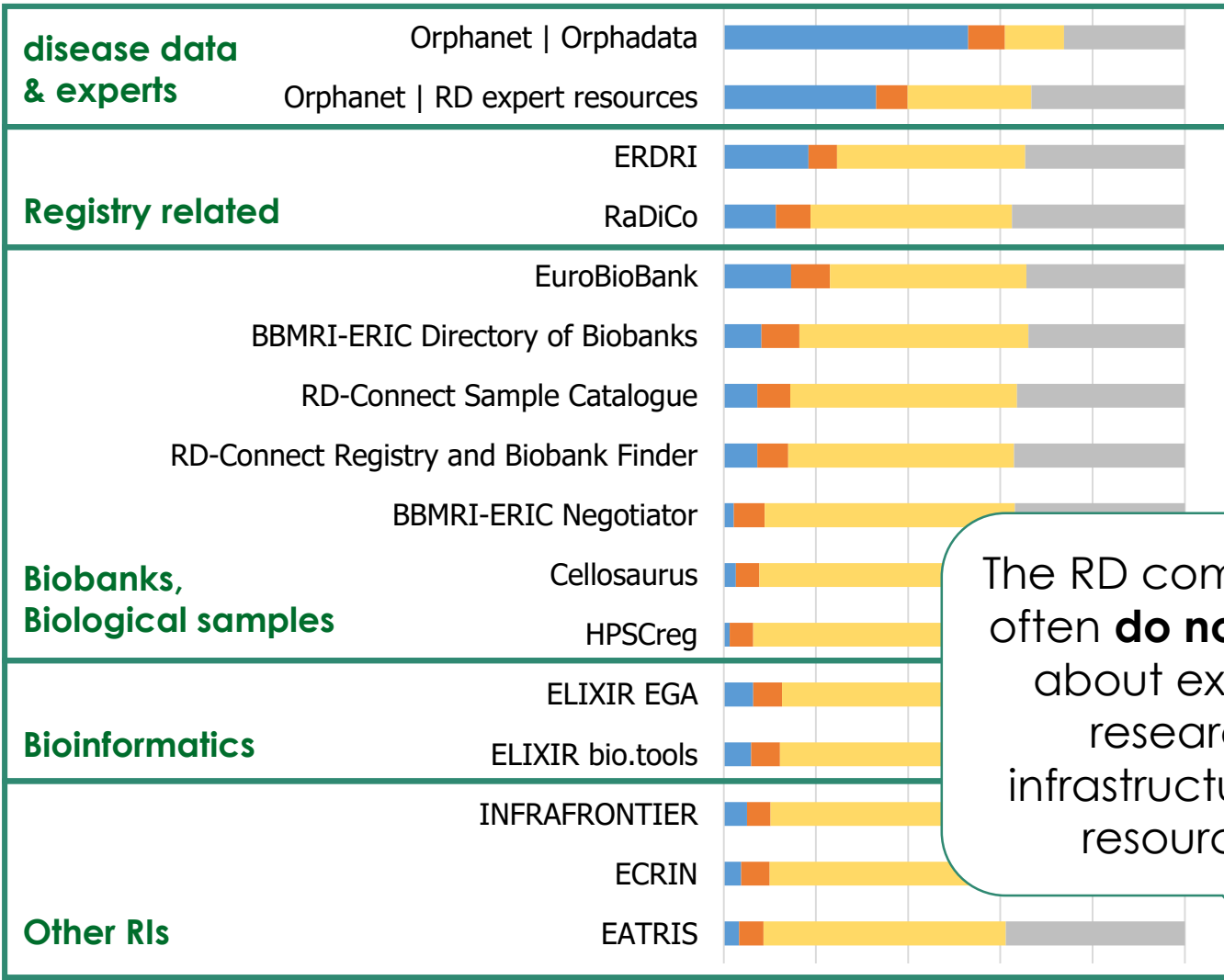


A very small fraction of researchers use the EJPRD related resources for their data deposition, sharing or analysis.

# Are these *infrastructures or databases* of utmost importance for your research?

0% 20% 40% 60% 80% 100%

0% 20% 40% 60% 80% 100%



The RD community often **do not** know about existing infrastructures or resources

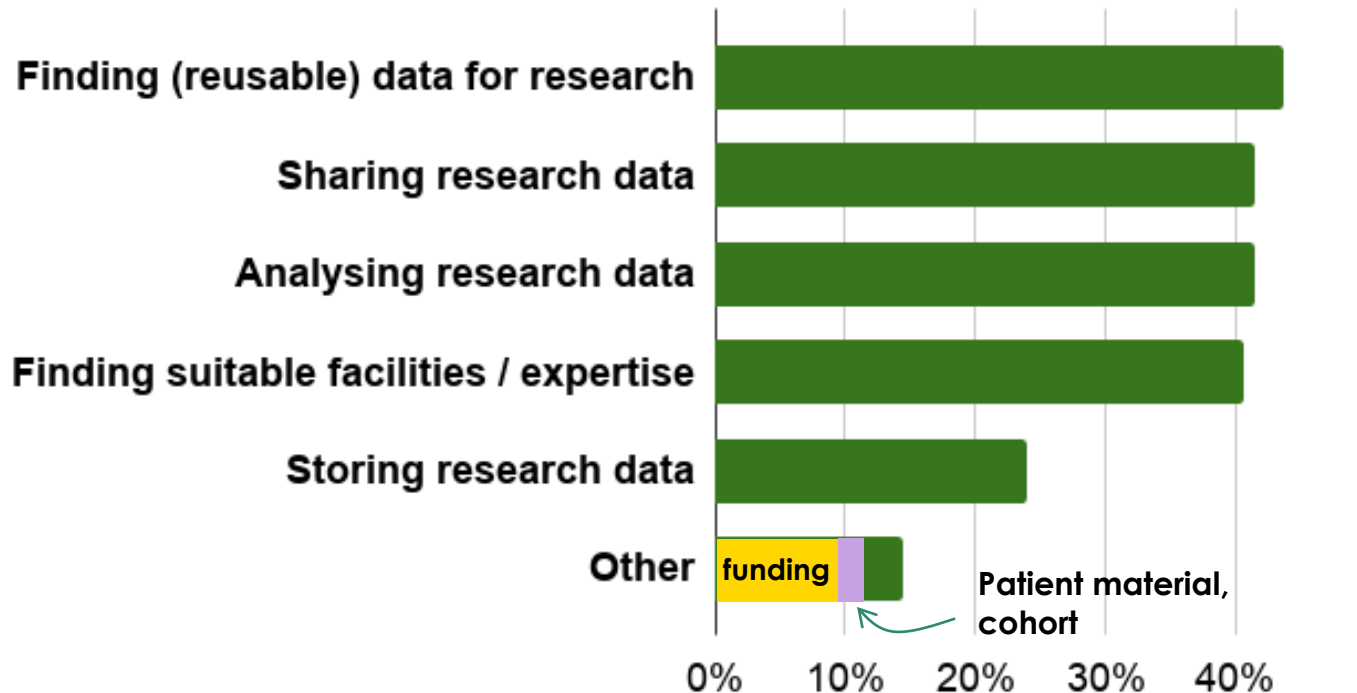
■ Yes ■ No ■ I don't know this resource ■ No answer

2019 ERNs



# General research needs

# To perform your research on rare diseases, do you face challenges in any of the following?



**What are your research needs other than financial / funding considerations?**

314 answers >>

Researchers face general challenges in finding, sharing and analysing data. Similarly there are challenges finding suitable facilities or expertises for their research.

# Needs other than funding: Wish list (1/2)

Type of need	count
Expertise & resources: Bioinformatics, Omics, biosamples, resources, models, Experts, databases, collaboration for resources	138
General collaboration, Training	88
Clinical development, Translational	13
GDPR, ELSI	6

## Access to patient materials

- Collaborations with clinicians to get access to patient samples and to know the clinical aspect of the diseases
- Availability of patient samples for basic researchers. Usually, patient samples are "owned" by the clinical researchs who took the samples or they "belong" to the research networks which collected the samples. For basic reseachers without contact to those networks it is VERY difficult to access patient samples. I suggest establishing a kind of "ADDGENE for patient samples".

## Multi-omics pipeline and expertise

- excellent easy-to-access and easy-to-manage multi-omics pipelines which allow easy data upload and easy data analysis of own omics data (transcriptomics, proteomics, etc)

▪ Access to data such as flow sequencing data/single cell datas and facilitating collaborations with teams of bioinformaticians capable of interrogating these datas in relation to our biological questions and shaping them for us would be great to move forward.

# Needs other than funding: Wish list (2/2)

## Rare disease database

- Specific information bases on rare diseases that contain clinical, genetic and affective information (there are some but are the exception). Numbers are power and at present most information is scattered and unavailable.
- A European proteomic database to record the differential expression levels of proteins studied in Rare Diseases.

## Translation to clinic

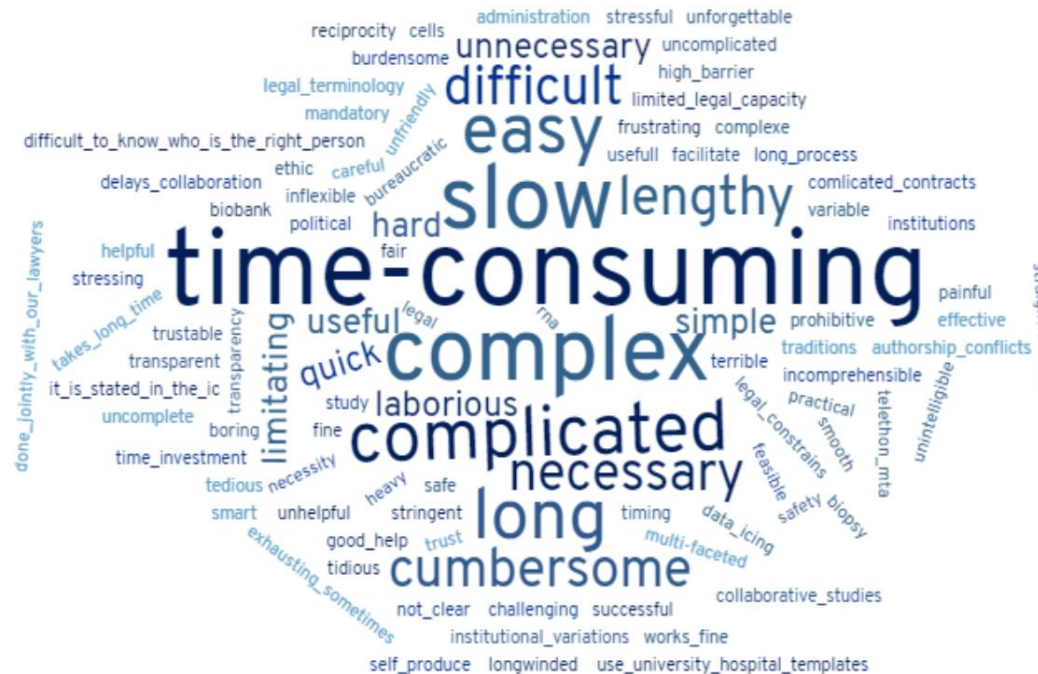
Collaboration with clinicians experts on the disease, to evaluate if potential treatments in animal models can be extrapolated to humans. Experts on extrapolating preclinical results into clinical trials, regulatory paperwork, etc

## Collaboration

- As a basic researcher working with mice, I wish I have more access to clinical records, including background and epileptic EEG recordings from patients
- My research needs of a close collaboration with clinicians.
- A research network that would include pharmaceutical companies.
- Make access to international research more accessible for small research groups.

**Data management skills, Human resources, Training, Data privacy, Bureaucracy**

# Have you used a DTA and/or a MTA that are compliant with GDPR for your research activities?

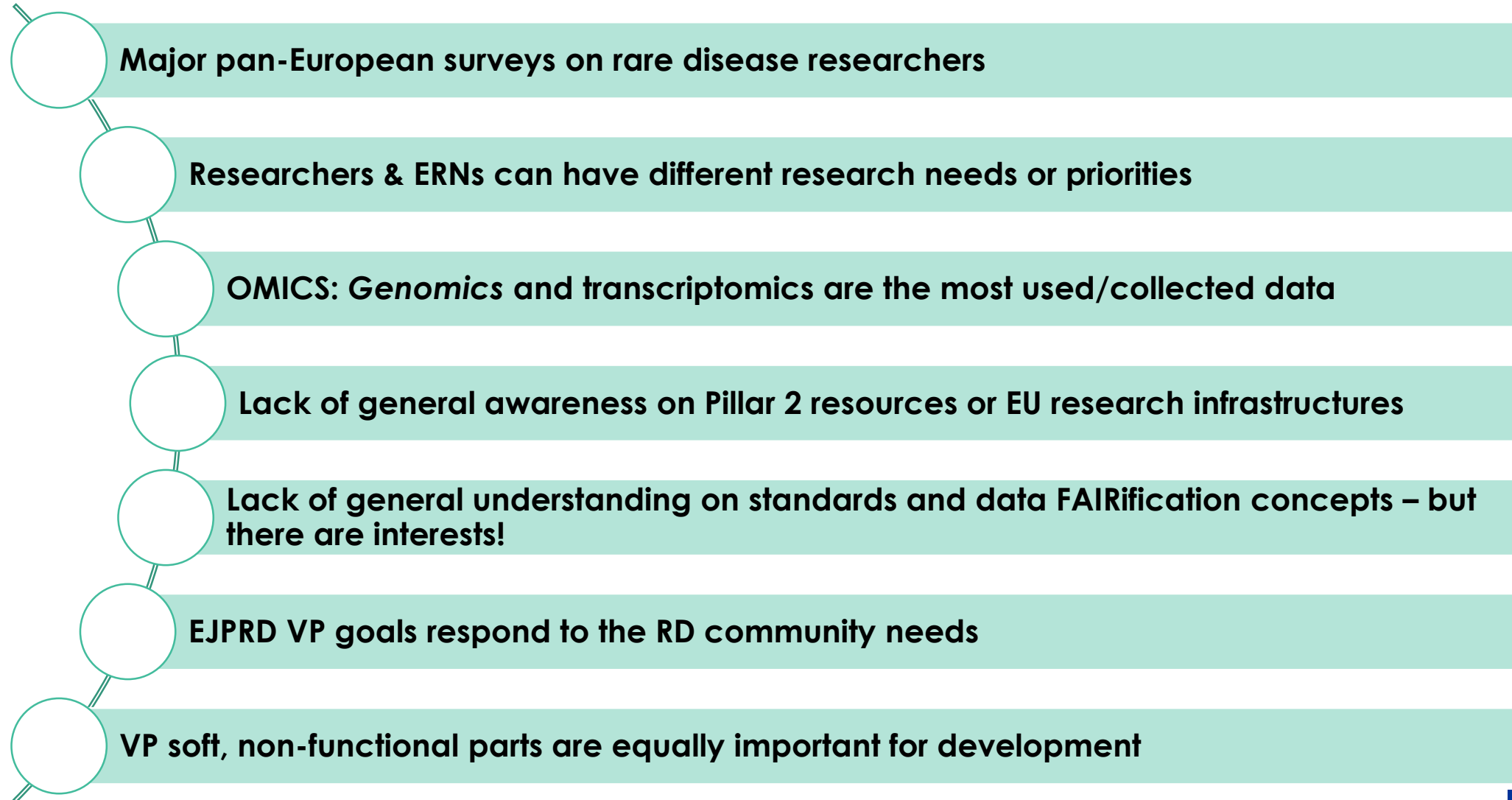


## Top 5 mentioned words (out of 187 words submitted)

time-consuming	17
complex	16
slow	12
complicated	11
easy	9

# Key messages & discussion

# Key messages



# Key questions

- 🌍 How can we leverage on different parts of the EJR RD to
  - 🌍 Drive best practices in RD research?
  - 🌍 Ensure output of EJPRD are known?
  - 🌍 **Encourage** adoption and use of standards and resources?
  - 🌍 Encourage collaboration among researchers and clinicians?



Mary Wang, PhD  
mwang@telethon.it



# WP3-Sustainability – Survey and Next Steps

Executive Committee Annual Meeting

July 6th, 2020

# Survey and Results

# Survey on Sustainability

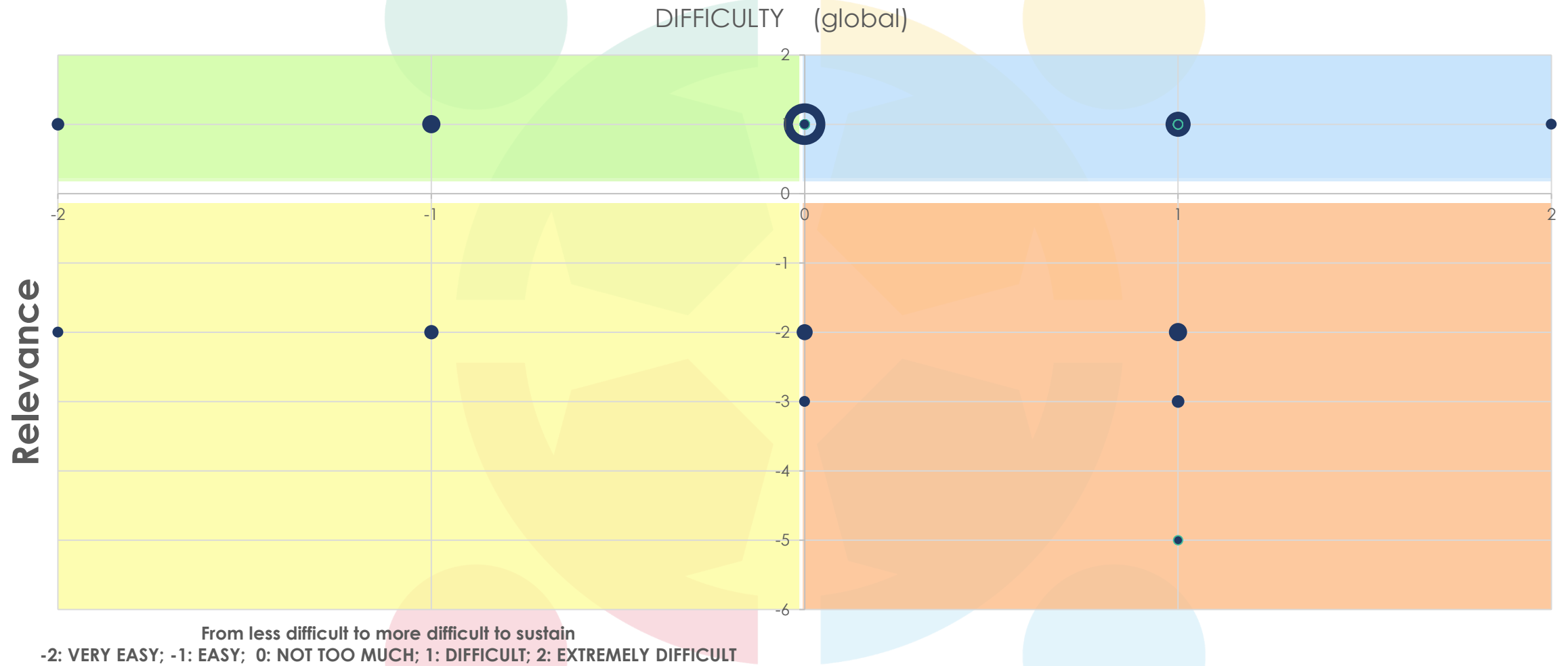
- 🌱 Identification of EJP RD elements that would be valuable to sustain beyond the life of the EJP
- 🌱 Launched for WP Leaders: February 2020
- 🌱 Launched for Task Leaders: May 2020

# Survey Results

Responses	Survey to Work Package Leaders	Survey to Task Leaders	Totals
<b>TOTAL responses</b>	<b>13</b>	<b>18</b>	<b>31</b>
<b>Number of elements identified</b>	<b>21 (13 MR)</b>	<b>43 (18 MR)</b>	<b>64 (31 MR)</b>

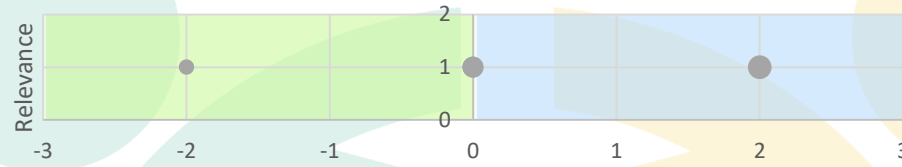
MR: Element selected as Most Relevant

# Elements by difficulty to sustain (global)



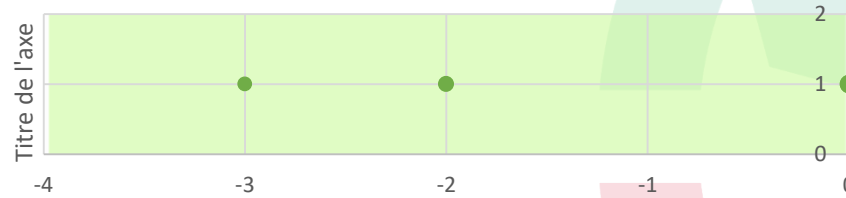
# Elements by difficulty to sustain (by Pillar)

DIFFICULTY PILLAR 0



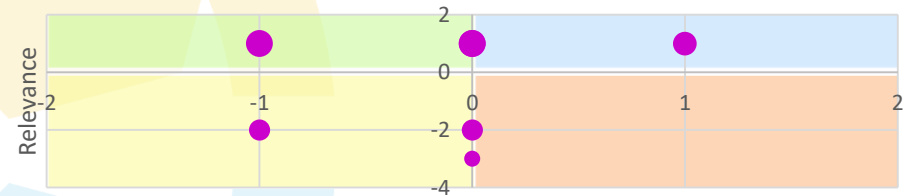
From less difficult to more difficult to sustain  
 -2: VERY EASY; -1: EASY; 0: NOT TOO MUCH; 1: DIFFICULT; 2: EXTREMELY...

DIFFICULTY PILLAR 1



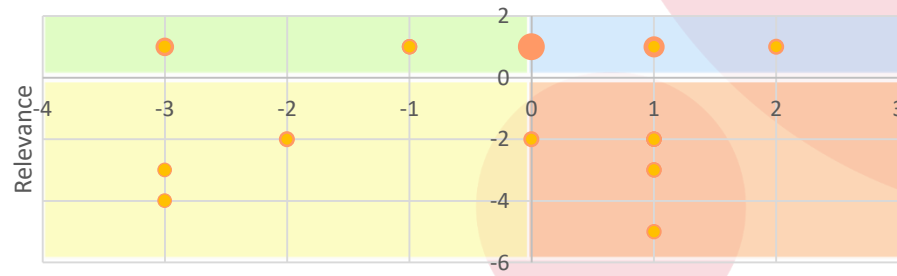
From less difficult to more difficult to sustain  
 -2: VERY EASY; -1: EASY; 0: NOT TOO MUCH; -3: MISSING

DIFFICULTY PILLAR 3



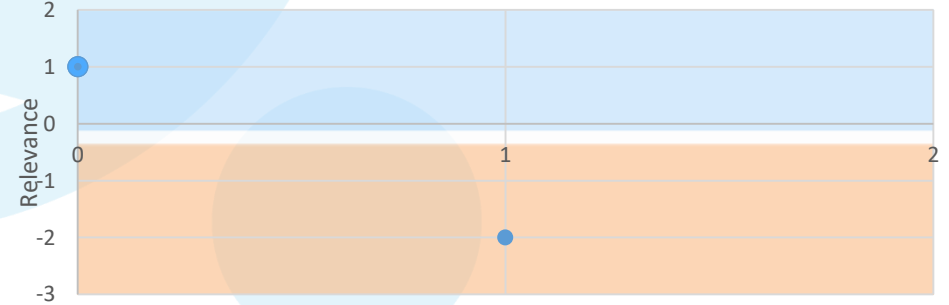
From less difficult to more difficult to sustain  
 -2: VERY EASY; -1: EASY; 0: NOT TOO MUCH; 1: DIFFICULT; 2:..

DIFFICULTY PILLAR 2



From less difficult to more difficult to sustain  
 -2: VERY EASY; -1: EASY; 0: NOT TOO MUCH; 1: DIFFICULT; 2:..

DIFFICULTY PILLAR 4



From less difficult to more difficult to sustain  
 -2: VERY EASY; -1: EASY; 0: NOT TOO MUCH; 1: DIFFICULT; 2:..

## PILLAR 0

SHORT TITLE	RELEVANCE	DIFFICULTY
<b>Call topics of joint transnational calls</b>	MR	VERY EASY
<b>AREB</b>	MR	NOT TOO MUCH
<b>Sustainability Handbook (including such elements of roadmaps, business planning and services catalogue as are not specific to the EJP RD activities) (Sustainability Handbook)</b>	MR	NOT TOO MUCH
<b>Scientific Advisory Board appointed to advise for the scientific programming of joint transnational calls.</b>	MR	NOT TOO MUCH
<b>EJP RD Central Helpdesk (Helpdesk)</b>	MR	DIFFICULT
<b>Re (ACT) congress - IRDiRC conference</b>	MR	DIFFICULT
<b>Mapping of research and innovation needs</b>	MR	DIFFICULT
<b>Service Roadmap Database for sustainability purposes</b>	MR	DIFFICULT



## PILLAR 1

SHORT TITLE	RELEVANCE	DIFFICULTY
<b>Joint transnational call toolbox</b>	MR	VERY EASY
<b>Monitoring tool</b>	MR	NOT TOO MUCH
<b>Public-Private Partnerships (PPP) and RDR Challenges innovative funding scheme as a tool for sustainable development (Public Private Partnerships)</b>	MR	NOT TOO MUCH
<b>The organisation of a European funding scheme for Networks (Organisation of application and evaluation of European networks taking into account the Horizon 2020 rules)</b>	MR	MISSING*

\* Most of info obtained, but no data on difficulty

# PILLAR 2

## Elements by pillar

SHORT TITLE	RELEVANCE	DIFFICULTY
API and data model standards for federated querying at the record level (Generic Query API (Beacon) and digital consent (ADA-M) standards)	2°	VERY EASY
Cafe Variome derived discovery installations (Cafe Variome (customised as PaR-RaDiGM and RD-NEXUS software))	MR	EASY
Domain-specific Common Data Elements (DCDEs) (Domain-specific Common Data Elements (DCDEs))	MR	EASY
DECIPHER	MR	EASY
Federated Query Broker	MR	NOT TOO MUCH
EUPID	MR	NOT TOO MUCH
BBMRI-ERIC Directory	MR	NOT TOO MUCH
Digital Research Environment for Analysis and Integration of multi-omics data	MR	NOT TOO MUCH
RD-Connect Biobank & Registry Finder Catalogue of Biobanks and Registries for Rare Diseases (Catalogue of Rare Disease Biobanks and Registries at the aggregated level)	2°	NOT TOO MUCH
FAIRification guidelines & stewardship wizard	MR	NOT TOO MUCH

# PILLAR 2

## Elements by pillar

SHORT TITLE	RELEVANCE	DIFFICULTY
<b>Cellosaurus</b>	MR	DIFFICULT
<b>Curation of mouse strain data, especially if submitted from individual researchers from the scientific community. This both includes assigning of official nomenclature and identifiers for mouse strains, genes and alleles, and linking to rare disease related information.</b>	MR	DIFFICULT
<b>Keep import of rare disease related information up-to-date as structure of both the INFRAFRONTIER/EMMA database and rare disease resources evolve over the years. Also make sure that the data provided from INFRAFRONTIER/EMMA to the EJP RD via an API stays compatible with the virtual platform. (Keep automated processes for data integration up-to-date).</b>	2°	DIFFICULT
<b>Research projects and clinical trial analysis platform (Metadata exposure service for resources)</b>	2°	DIFFICULT
<b>Data validation service/tool (SHex/SHACL)</b>	5°	DIFFICULT
<b>FAIR steward support service &amp; FAIR training (BYOD)</b>	2°	DIFFICULT
<b>Toolbox for FAIR stewards &amp; engineers</b>	3°	DIFFICULT
<b>RD-Connect Genome-Phenome Analysis Platform (GPAP)</b>	MR	DIFFICULT
<b>Providing training for researchers in submission of EJPRD related data to the appropriate resources</b>	2°	DIFFICULT
<b>Providing training materials and best practices guidance to be used by other databases and researchers for training and integration</b>	3°	DIFFICULT
<b>Defining specifications for capturing, curating and integrating EJPRD related data in as appropriate in national and global resources eg MetaboLights (Data specification, curation and integration)</b>	MR	EXTREMELY DIFFICULT

## PILLAR 2

SHORT TITLE	RELEVANCE	DIFFICULTY
Metadata exposure service for resources (Research projects and clinical trial analysis platform)	MR	MISSING*
Metadata alignment service/tool	3°	MISSING*
Query builder service	4°	MISSING*
RD-Connect Sample Catalogue Catalogue of Individual Samples from RD Biobanks. (Record Level catalogue of Samples)	MR	MISSING*

\* Most of info obtained, but no data on difficulty

# PILLAR 3

## Elements by pillar

SHORT TITLE	RELEVANCE	DIFFICULTY
Training contents for paediatric patients	MR	EASY
Training module (Task 14.1)	2°	EASY
Training material (Task 14.1)	MR	EASY
Secondments - acquiring NEW research experiences / methodologies	2°	EASY
Preparatory material and training materials	2°	EASY
the actual training workshops, related to biobanks and biological materials (biobank & biological samples training)	MR	NOT TOO MUCH
Preparatory materials from the workshops: 1) Problem cases for "problem based learning", 2) Preparatory guide for facilitators 3) design of workshop sessions 4) list of appropriate speakers	2°	NOT TOO MUCH
Workshops for paediatric expert patients	2°	NOT TOO MUCH
Annual Training Courses on "Quality assurance. variant interpretation and data management in the NGS diagnostics era"	MR	NOT TOO MUCH
ERN Research Workshops	MR	NOT TOO MUCH
Web Services (for TC, survey....) (Web System)* (same for Task 14.3&14.5)	2°	NOT TOO MUCH
Training material *(same for Task 14.3&14.5))	3°	NOT TOO MUCH
Workshops to share knowledge and create training multipliers	MR	NOT TOO MUCH
Biobank, biosample training workshops	MR	NOT TOO MUCH
Programme delivery for the 3 schools	MR	DIFFICULT
Online academic course	MR	DIFFICULT
Course* (same for Task 14.3&14.5)	MR	DIFFICULT

## PILLAR 4

SHORT TITLE	RELEVANCE	DIFFICULTY
<b>Innovation Management Toolbox</b>	MR	NOT TOO MUCH
<b>Clinical Trials Support Office</b>	MR	NOT TOO MUCH
<b>Mentoring Service</b>	2°	DIFFICULT

# Identified elements without information

SHORT TITLE	PILLAR	WP/TASK	RELEVANCE
Exchange of best practice and knowledge	0	2.5	MR
PRIDE	2	11.3	MR
Demonstration projects call	4	23.3	MR
HSL	1	8	MR
Lists of researchers/ projects	1	9	2°
Life Science AAI	2	12.2	2°
Programme developed of the 3 training courses (Summer, Winter and Leadership schools)	3	15	2°
Faculty for the 3 schools	3	15	2°
ERN Research fellowship exchanges	3	17.3	2°
Report of funded Demonstration Projects	4	23.3	2°
Training materials: presentations from speakers, mock data for data harmonisation exercise, tool demo/documentation, reference lists	3	14,4	3°
Report of funded methodology Projects	4	23.3	3°
ADA-M / DUO	2	12.2	3°
FAIR maturity test tool	2	12.3	4°
Semantic data models & FAIR data point specifications	2	12.3	5°
FAIR resource catalogues/indexes	2	12.3	6°

# Lessons for Follow-Up Survey



## Complexity of Survey and IT issues

- ⌘ Simpler Structure for respondents
- ⌘ Accompanying Guide to explain terms used, give examples, and describe Lymesurvey process and issues
- ⌘ Teams channel for technical video and presentation, discussion and queries
- ⌘ Improved response rate and detail by focusing on Task level rather than WP level



# Next Steps

# Analysis of Results

- Follow up interviews with all respondents to enable full understanding and assessment of all elements
- For all elements assess and report to ExCom on:
  - Particular critical sustainability issues requiring immediate action (IP protection, systems design, etc.)
  - General issues with sustainability and how best to work with WP and Task Leaders to address these (Advice and information to be provided in brochure and Toolbox)

# Follow-Up Survey

- 🌈 Follow up survey to be launched end 2020
  - 🌐 Accompanying information and guidelines
  - 🌐 Tailored to identify critical factors to be addressed

# Action Plan

- 🌍 Analyse and report on all elements assessing critical risks and opportunities and recommend actions
- 🌍 Monitoring process – Excom review of Report and planned actions
- 🌍 Individual engagement and resources to be provided to ensure sustainability considerations embedded in all WPs
- 🌍 Development of Sustainability Roadmap



 **Thanks from WP3**

**ISCIII, EATRIS, INSERM RaDiCo  
and DTL-P ELIXIR NL**

# EJP RD Executive Committee Meeting 7 July 2020

European Commission Ethics Requirements  
And role of the Ethics Advisors

By Anne Demoisy

And Joana Namorado ([www.rhizome.be](http://www.rhizome.be))

# Overview

1. The external Ethics Advisors' roles and team
2. Ethics in Horizon 2020 and in EJP-RD
3. The ethics requirements and deliverables
4. KEY Ethics concepts - CONSENT and DATA PROTECTION
5. The Work-package21 Deliverables
6. Timeline and Partners' contribution
7. Ethics added value ?
8. Time for questions ?

# 1. The external Ethics Advisors' team

**RHIZOME s.a. Ethics and Technology**

**www.rhizome.be**

• Mrs Anne  
DEMOISY

• Rhizome  
Director

• Philosopher and  
Computer Scientist

• Senior Consultant in Ethics and  
Technology within H2020 projects,  
Advisor and Auditor to the  
European Commission

• [anne.demoisy@ejprd-project.eu](mailto:anne.demoisy@ejprd-project.eu)



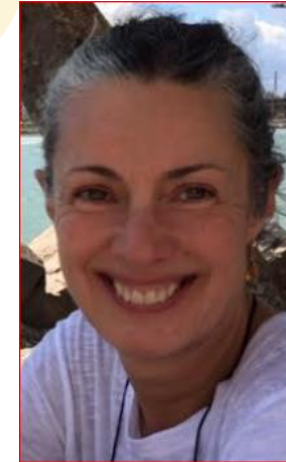
• Dr Joana  
NAMORADO

• Rhizome Partner  
from Citolab Ltd

• Physician

• Senior consultant in medical ethics,  
previously working at the Council of  
EU Ministers and at the European  
Commission RTD - Health

• [joana.namorado@ejprd-project.eu](mailto:joana.namorado@ejprd-project.eu)





# The external Ethics Advisors' roles

- Provide guidance to the Coordination Team on the structure and content of the WP21 ethics deliverables
- Review the ethics deliverables before their submission to the European Commission
- Cooperate closely with the WP4 in charge of the EJP-RD ethics and legal tasks
- Support the WP4 in writing the annual reports on ethics
- Participate to the AREB (Advisory Regulatory and Ethics Board) and to relevant meetings
- Provide ad-hoc advice during project life.

## 2. Ethics in Horizon2020



- Respect of the HORIZON 2020 Ethics principles
- Compliance to the National legal & ethics requirements and codes of practice of the Member States where the research is performed. (i.e. for human involvement in field tests, legal requirements for research in Non EU countries)
- The General Data Protection Regulation (GDPR 2016/679 - 25May 2018) protecting the citizens' privacy and increasing the responsibility of partners collecting and processing data within European Union.

# Ethics in EJP-RD



- An Initial Ethics Review was performed at program start by a panel of Ethics and Data protection experts mandated by the European Commission.
- The Reviewers identified 15 ethics and data protection requirements regrouped within Work-package 21.
- These WP21 deliverables will be reviewed by the European Commission experts during the ethics audit/checks.

# 3. Ethics Requirements and Deliverables



## EU legislation in the spotlight

- Clinical Trials **Regulation**
- Data Protection **Regulation**
- ATMP **Regulation**
- Stem Cell Products
- Blood and Tissues **Revision**
- Food legislation (Health claims)REFIT
- Medicinal Products
- Protection of Animals for experimental purposes

# 4. KEY ETHICS CONCEPTS : CONSENT is the focus of research with Humans, clinical trials, samples, data processing and third countries involvement

🌈 Able to Consent

🌈 Cannot consent to illegal

🌈 IC is a PROCESS

🌈 IC for what, where & when

🌈 Registration of CT

🌈 Benefit sharing

🌈 Ownership of Clinical data

🌈 Not able to Consent

🌈 Justification

🌈 Participation as a right

🌈 Mitigation

🌈 Benefit to individual

🌈 Re-consent

# The issue of DATA – a dual nature

## 🌈 Data as content

- 🌈 Background
- 🌈 Foreground
- 🌈 Capital to Share
- 🌈 Patentable or not
- 🌈 Exploitation part of Consortium Agreement



## 🌈 Personal Data

- 🌈 What data Support
- 🌈 Portability
- 🌈 How long and destruction
- 🌈 What for and Meta-data
- 🌈 Information within information
- 🌈 Privacy is civil liberty
- 🌈 GDPR



# 5. THE WP21 DELIVERABLES

## D21.3 Creation of the AREB -Advisory and Regulatory Ethics Board

- Creation of a Board to monitor the Ethics and Data Protection issues raised by the program
- Writing an annual report to the European Commission on how issues are managed within EJP-RD





# D21.14 and D21.15 Appointment of external expertise in Ethics

- Is expected to deliver an annual independent report
- Is expected to join the AREB -Advisory and Regulatory Ethics Board
- The Board should make sure the H2020 framework for ethics is applied for the planned subsidiary projects

## Collaboration on the Ethics



# D21.4 to D21.9

## Human Participants- Informed Consent procedures

- Criteria for participants recruitment
- Justification for involvement of children and adults unable to consent
- Informed Consent/Assent procedures and templates
- Copies of ethics approvals for the research

An Involved Patient = a  
Consenting Patient



# D21.4 to D21.9 Human Participants- Informed Consent procedures

## What **MUST** be in the Consent Form

## What must be Re- enforced in the **CONSENT** process

- This is research project
- Aim+duration+benefit
- Foreseen risks
- Alternatives
- Confidentiality
- Treatment/compensation +information
- Contact for rights/claims
- Contact for injury
- Voluntary participation
- No penalty on stopping
- Consent for WHAT



*"Paying my fee will also help as evidence for our insanity defense."*

# D21.1 ANIMALS - Directive 2010/63/EU



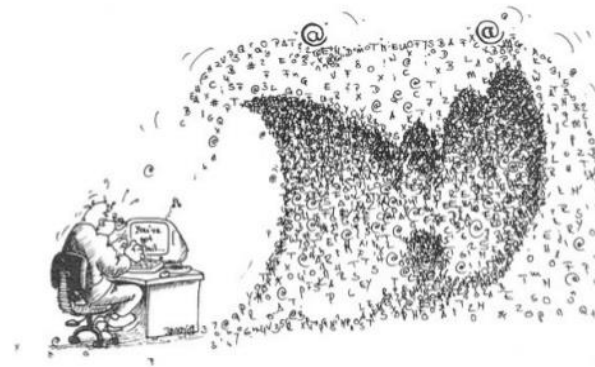
- Procedures to ensure animals welfare
- Respect of 3Rs
- Training requirements for researchers
- Justification when inclusion of non human primates



# D21.12 & D21.13 - Personal Data Processing procedures and authorisations

- Procedures for data collection storage, protection, retention and destruction
- Confirmation they comply to GDPR
- Relevant authorisations to use previously collected data
- Meta-data and Merged DB

Getting lost in your data



# D21.10 to D21.11 - Personal Data Processing and DPOs

- Check of need of declaration of compliance at national level
- Identification of the Data Protection Officers (DPOs)
- Statement of compliance by the DPOs



# D21.2 NON EU Countries

- Procedures for Transfer of personal data /material between EU and Non EU
- Details on transfer and copies of authorisation (when needed)
- Confirmation of compliance to H2020 standards in Non EU countries



# 6. TIMELINE





# 6. Timeline and Partners' contribution

- 🌟 Meeting with the WP4 on synchronisation of ethic activities –17 July
- 🌟 Onboarding and preparation of the external Ethics Advisor initial assessment – 15 September
- 🌟 Support the Coordination in the preparation of the ethics deliverables
- 🌟 Prioritisation of the deliverables and discussion with the European Commission
- 🌟 From September your contribution will be requested for providing Informed Consent procedures and forms, data protection policies and all needed information in order to fulfill the requirements.

# 7. ETHICS ADDED VALUE ?

# Ethics: Added Value?

Risk\* Assessment

Risk\* Management

Basis for validation and Regulatory files

And provides a trace

IP /ownership of reseach – it is a chronological written (often Notarized)Timeline for an idea – that cannot be published

\* Ethics risk is Reputational BUT can become Financial and legal

Attorneys for Plaintiffs

UNITED STATES DISTRICT COURT  
SOUTHERN DISTRICT OF NEW YORK

THE AUTHORS GUILD, INC., THE AUSTRALIAN SOCIETY OF AUTHORS LIMITED, UNION DES ÉCRIVAINES ET DES ÉCRIVAINS QUÉBÉCOIS, PAT CUMMINGS, ANGELO LOURAKIS, ROXANA ROBINSON, ANDRÉ BOY, JAMES SHAPIRO, DANIELE SIMPSON, T.J. STILES and FAY WELDON,

Plaintiffs,

-against-

HATHITRUST, THE REGENTS OF THE UNIVERSITY OF MICHIGAN, THE REGENTS OF THE UNIVERSITY OF CALIFORNIA, THE BOARD OF REGENTS OF THE UNIVERSITY OF WISCONSIN SYSTEM, THE TRUSTEES OF INDIANA UNIVERSITY and CORNELL UNIVERSITY,

Defendants.

Colorado Casualty Insurance Company, a Colorado corporation,

Plaintiff,

vs.

PERPETUAL STORAGE, INC., a California corporation; UNIVERSITY OF UTAH, a body politic and corporate of the state of Utah, on behalf of UNIVERSITY OF UTAH HOSPITALS AND CLINICS; UNIVERSITY OF UTAH HEALTH SCIENCES CENTER,

Defendants.

"I am stunned offences on this scale were possible in the VW Group."

Mr Winterkorn at the time news of the scandal broke

BMJ

Updated: Anil Potti, Duke Cancer Researcher Accused of Misconduct, Resigns

By Zachary Treisman, Taylor Delaney | November 23, 2016

Updated 8:30 p.m. with comments from Dr. Anil Potti, IISG Director Huntington Willard and Dr. Michael Coyle, IISG vice president for medical affairs.

The Duke cancer researcher who has been under investigation for research misconduct since this summer has resigned.

"Full responsibility in data handling that have come months."

According to Utah incur the security

Ethics in the News

EIN Report on Challenges for Journalism in the Post-truth Era



# 8. TIME FOR QUESTIONS ?



# EJP RD Policy Board & Governing Board Meeting

# Welcome word

# Presentation of new members

Name	Institution	Country
Pierre Meulien	Innovative Medicines Initiative (IMI)	EU
Janis Ancans	Ministry of Education and Science	Latvia
Richard Imrich	Ministry of Education, Science, Research and Sport	Slovak Republic
Onur Burak Dursun	Ministry of Health	Turkey
Anželika Balčiūnienė	Ministry of Health	Lithuania
Alessandra Renieri	Ministry of of Education, University and Research	Italy
Pilar Aparicio Azcárraga	Ministry of Health, Social Services and Equality	Spain
Patricia Masia:	Ministry for Higher Education, science and technology	Portugal
Ryszard Rzepecki	National center of RD	Poland
Supriya Sarma	Health Canada	Canada
David K Lee	Health Canada	Canada



# 18 months of EJP RD activities and achievements

**85%** of  
**European RD community**  
(directly or indirectly)  
involved in **EJP RD**

**750** people:

**650** Scientifics  
**100** Admin

**35**

participating  
**countries**



**87** Beneficiaries:

**9** hospitals  
**12** research institutes  
**31** research funding bodies/ministries  
**24** universities/hospital universities  
**5** EU infrastructures  
**5** charities/foundations  
EURORDIS

+

**50**

**Linked Third Parties**

**And 100% of the associated networks**

**EURORDIS:**

884 RD patient  
organisations  
72 countries

**EATRIS**

13 main national nodes

**INFRAFRONTIER**

23 partners  
15 countries

**24 ERNs:**

300 institutions  
>950 healthcare units  
26 countries

**ECRIN**

12 main national nodes

**ELIXIR**

220 research  
organisation  
23 partners

**BBMRI**

1 international partner  
21 main national nodes  
20 countries



# EJP RD STRUCTURE

Coordinated by



# M1-M18 Coordination and transversal (Pillar 0) activities & achievements

- ✿ Established (and powerful) governance and organisation → Policy Board, Governing Board, General Assembly
- ✿ Professional, knowledgeable and efficient coordination team
- ✿ Seamless take over of IRDiRC Scientific Secretariat
- ✿ Smooth research strategy establishment process
- ✿ Very efficient ethics, regulatory and legal advice support ensured by AREB
- ✿ Communication & dissemination achieving steady growth (1000 web visits/day, 100 new NL subscription/month, >1300 twitter followers)
- ✿ EJP RD is recognized as major European player in the field of rare diseases → opening the doors to new collaborations (GA4GH, C-PATH, 1+MG, etc.)

# M1-M18 Pillar 1 activities & achievements

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

31 funders  
23 countries

220  
Submitted  
projects

52  
Selected  
projects

22  
Funded  
projects

30,5 million €

spent in total

Including 6 million€ from the  
European Commission

✿ JTC 2020: *Pre-clinical research to develop effective therapies for rare diseases* (launched in December 2019)

29 funders  
22 countries

173  
Submitted  
projects

30  
Selected  
projects

??  
Funded  
projects

- **Successful « widening »:** 14 new partners included in Full proposals
- Around **50%** of Full proposals used the **mentoring service** (WP19)
- **PAOs are included in all consortia**

✿ JTC 2021: *expected to focus on Social Sciences and Humanities Research to improve health care implementation and everyday life of people living with a rare disease*

Ongoing preparation of potential topics for JTC2021

# M1-M18 Pillar 1 activities & achievements

- ✿ EJP RD supports **engagement of patients in research** → preparation of a **guide on how to engage/include patients in research projects**, especially in the pre-clinical and social sciences and humanities research
- ✿ EJP RD encourages **sharing of knowledge on rare diseases and rare cancers** of health care professionals, researchers and patients via **Networking Support Scheme (NSS)**:
  - ✿ New funding scheme developed during Year 1 → The first call opened on M12 (First round completed)

**9 applications received** **Evaluation** **6 funded applications**

- ✿ EJP RD allows a **long term monitoring** of both EJP RD and E-Rare (ERA-Net) funded projects
- ✿ EJP RD volunteered to be one of the **pilot projects to align monitoring with the European Commission** as requested by Member States

# M1-M18 Pillar 1 activities & achievements

## Rare Diseases Research Challenge Call

- First RD targeted **public/private collaboration** for small scale projects with rapid uptake
- **Co-funded** by EJP RD and industry partners

### Challenge 1

*Chiesi and CSL  
Behring*

Development of a non-invasive tool for measuring rare disease patient mobility in daily living

### Challenge 2

*Ipsen*

Continuous remote assessment tools to characterize mobility in rare bone disorders in real world setting

### Challenge 3

*Chiesi*

Delivery system for intranasal administration of biological drugs to neonates

### Challenge 4

*Cydan and Pfizer*

Cellular assay to detect instability of microsatellite repeat expansions

- Call **launched on April 2<sup>nd</sup> 2020 (M16), closed on June 30, 2020 (M18)**

# M1-M18 Pillar 2 activities & achievements

- ERNs and RD research community needs captured respectively through 2019 (291 responses ) and 2020 (451 responses) surveys
- Strategic plan, working organisation and development workflow set up involving Pillar 2 “developers” and end-users ”ERNs representatives, RD research community”
- Pillar 2 **Virtual Platform (VP) resources mapped and described**

**Established and consolidated engagement with international expert** networks and organisations such as the GA4GH and GO FAIR



To set up the basis of the creation of an **operational Virtual Platform**



To influence and adopt international standards for a **FAIR Virtual Platform**



# M1-M18 Pillar 2 activities & achievements



- Based on use-cases
- **Federated architecture** drafted



- **Authentication Authorisation Infrastructure**, consent control
- **privacy-preserving record-linkage** (EUPID)
- **Automated GDPR compliant access protocol** (Automatable Discovery and Access Matrix [ADA-M]; Data Use Ontology [DUO])



- **Quality** criteria
- **Sustainability** criteria
- **FAIRness**
- **Standardisation**
- Pillar 2 **GDPR guidelines**

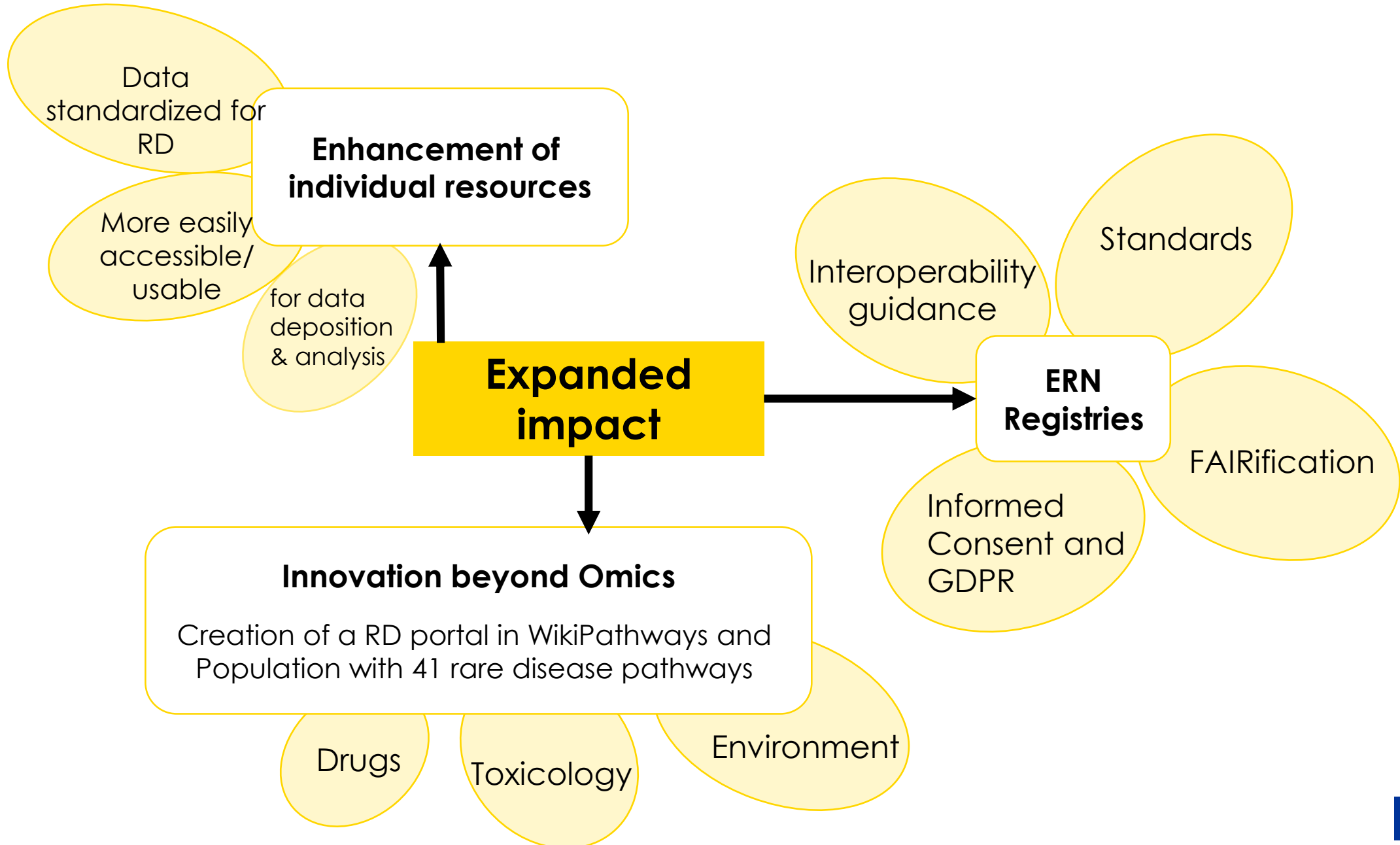
→ **Level 1:** 1st central access version ([Linked-data platform](#)) released for catalogues (e.g EDRDI, Orphanet, RD-Connect Sample Catalogue & Biobank-Registry Finder)

- **extendible** through individual registries/resources
- and **allowing for interpreting data coming from multiple heterogeneous sources.**

→ **Level 2 (deeper):** 1st semantic metadata model to represent the record-level of data (incl. the [Common Data Element](#) of JRC\*)

\*Pseudonym, Personal information, Patient Status, Care Pathway, Disease history, Diagnosis, Research, Disability)

# M1-M18 Pillar 2 activities & achievements



# M1-M18 Pillar 3 activities & achievements

## Overall achievements

## WP14 & WP15:

- 7 face-to-face courses in 5 countries by end of 2019 (220 participants, 18 fellowships)
- 4 online courses by M18 (130 participants)
- 350 stakeholders trained so far increasing research potential of the multi-stakeholder EU RD research community

Number of participants by trainings (M1 to M18)

Quality assurance, variant interpretation and data management in the NGS...  
Strategies to foster solutions of undiagnosed rare disease cases (Task 14.3)  
Sample data management training workshops (Task 14.4)  
Rare Disease Registries (RD Registries) and FAIRification of data (Task 14.5)  
EURORDIS Summer School (Task 15.1)  
EURORDIS Winter School (Task 15.2)  
EURORDIS Leadership School (Task 15.3)

58

# M1-M18 Pillar 3 activities & achievements

## WP16

- 🌍 Outreach to an international scale with the EJP RD e-learning academic course is within reach:
  - 🔗 Choice of the e-learning platform was finalized by the end of 2019 and the contract signed for the development of five modules amounting to around 50 to 60 hours of online learning
  - 🔗 The design of the module 1 has been finalized and the first session will run by the end of 2020. It will include 5 different units delivered over 5 weeks, 2 hours/week

Module 1: RD  
Diagnosis

Module 2: RD  
Innovative  
personalized  
therapies

Module 3: RD  
translational  
research

Module 4 & 5:  
to be define d  
in year 3

## WP17

- 🌍 A new level o collaboration between ERNs is being introduced with the implementation of the training programs for the ERN training networks tailored according to their needs
  - 🔗 The first round of calls for ERN-driven cross-cutting workshop and research mobility fellowship proposals calls has been implemented and the awardees selected.



Research  
workshop calls

5 applications  
3 workshop  
topics  
selected



Research  
mobility  
fellowship calls

16  
applications  
6 projects  
selected for  
funding

# M1-M18 Pillar 4 activities & achievements

## Facilitating partnerships and accelerating translation for higher patient impact

### Innovation management toolbox

- Integration of various resources supporting research translation (including the IRDiRC Orphan Drug Dev. Guide)
- Connection with Pillar 2 to make it interoperable with the Virtual Platform

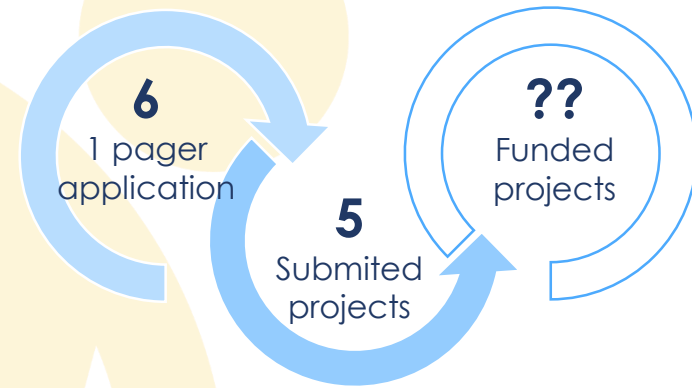
### Support for translation & innovation mentoring

- Pool of mentors to support the translation of research projects
- Assessment of the translation needs of E-Rare, EJP RD funded projects and ERN projects
- First confidentiality agreement signed to work with E-Rare funded project
- Mentoring of 15 projects within EJP RD JTC2020

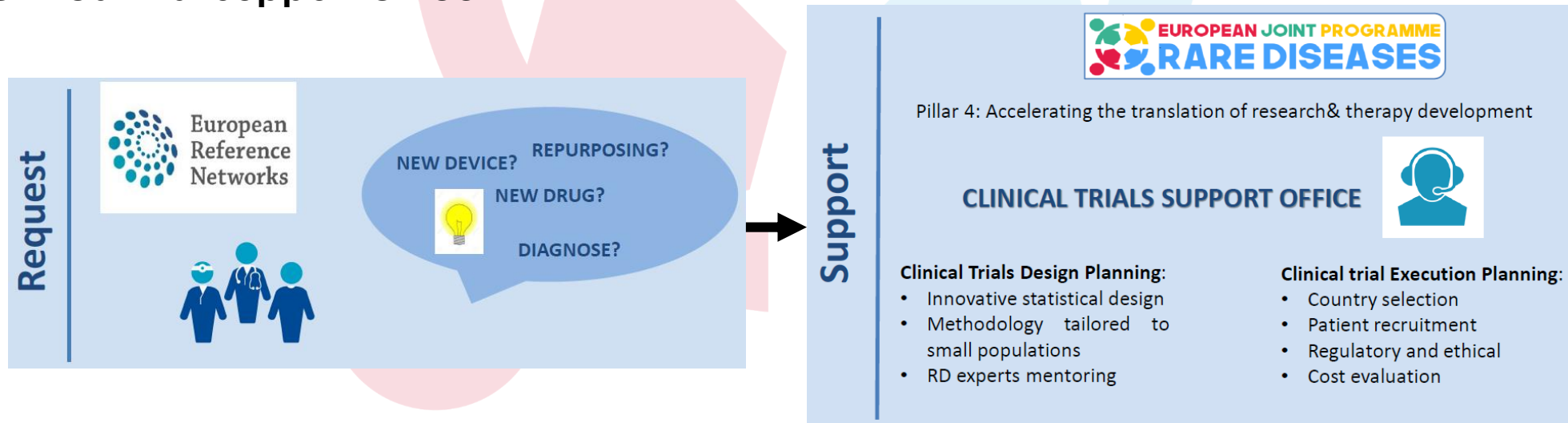
# M1-M18 Pillar 4 activities & achievements

## 🌟 Clinical Trials Methodology Demonstration Projects

- 9 innovative statistical methodologies (based on outcomes of Asterix, IDeAI, InSPiRe)
- Clinical experts matchmaking with methodology experts
- Validation of selected methodologies as proof-of-concept innovation in small population clinical studies



## 🌟 Clinical trial Support Office



# Annual Work Plan Year 3

## Discussion on strategic points

# Strategic points – Pillar 0

## **Requiring PB-GB input:**

-  How to best prepare the next phase – Rare Diseases Partnership under Horizon Europe
-  The most efficient approach to ensure EJP RD (elements/services) sustainability → follow up at national/EU/international level

## **For PB-GB information and/or support:**

-  Policy Board and stakeholders strategy meeting 12-13 of January 2021, Hotel Melia, Berlin, Germany
-  Support for National Mirror Groups





# Strategic points – Pillar 1

## **Requiring PB-GB input:**

-  Topic of JTC2022 needs to be defined : the previously envisaged topic is now obsolete due to the existing difficulty to fund clinical trials within EJP RD funding schemes

## **For PB-GB information and/or support:**

-  Need support from PB/GB for the dissemination of the JTC2021 to the researchers in the area of Social Sciences and Humanities
-  Networking Support Scheme: effect of Covid-19 on this type of scheme → information on how the EJP RD is adapting to this new situation

# Potential JTC 2021 Topics

- 🌟 **Health care & social services** research to improve patient health outcomes
- 🌟 **Health Economics Impact**
- 🌟 **Social Impact of Rare diseases**
- 🌟 Studies addressing the **impact/burden of the delay in diagnosis and of the lack of therapeutic intervention.**
- 🌟 **e-Health in rare diseases:** Use of innovative technology systems for care practices in health and social services
- 🌟 Development and enhancement of **health outcomes research methods** in rare diseases
- 🌟 **Effects of the global outbreak alert and response** on the rare disease field, and the emergence of innovative care pathways in this regard

# Strategic points – Pillar 2

## **Requiring PB-GB input:**

- ⌘ The heterogeneous interpretation of GDPR at member state and institutional level

## **For PB-GB information and/or support:**

- ⌘ Extension of the VP by connecting resources that are more researchers-oriented (in addition to those that are clinical-researchers oriented)
- ⌘ Adoption of a full-federated architecture roadmap, however starting from some centralized developments
- ⌘ Development of cloud analysis facilities
- ⌘ Concrete plans for ERNs registries FAIRification
- ⌘ Progress in X-omics pathways and networks developments

# Strategic points – Pillar 3

## Requiring PB-GB input:

Identification of new education & training needs/gaps

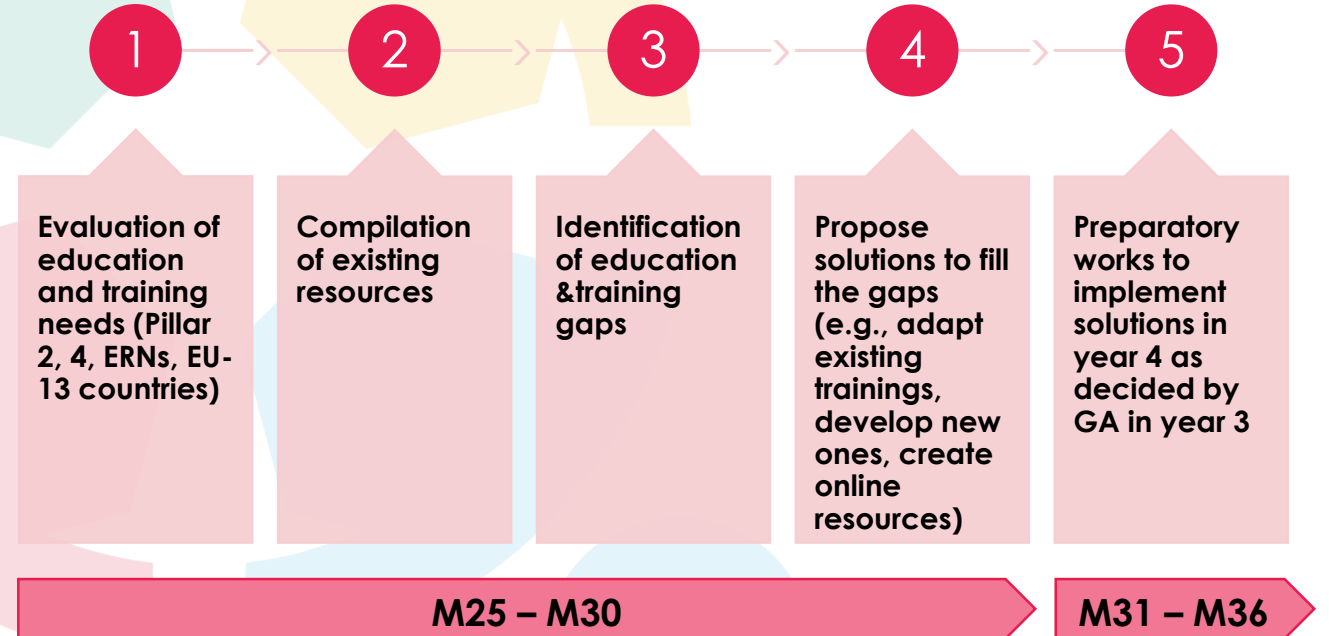
### RATIONALE

It was identified that education and training on RD research is insufficient, fragmented and geographically unequal across Europe

The EJP RD has already started to fill this gap (350 stakeholders trained so far)

**WP18 goal is to ensure that capacity building activities within Pillar 3 address the developing education and training needs in RD research of key stakeholders across different EU countries**

**Year 4: delivery of novel education and training activities**



**Budget total: 155 970  
(Travels & catering = 60 250)**

# Strategic points – Pillar 4

## **Requiring PB-GB input:**

- ✿ Follow-on funding service of WP19: how to ensure that PB are aware and notify WP19 leadership of Suitable funds (pre-seed, seed and Proof-of-concept funds) in their regions
- ✿ How to best engage regulators (EMA, FDA) to validate the adoption of innovative clinical trial methodologies

## **For PB-GB information and/or support:**

- ✿ Support in raising awareness and access to information regarding the Pillar 4 support services (research translation support services and clinical studies support office)
- ✿ Support to engage with different stakeholders and sponsors (EU-funded initiatives, international initiatives, industry) to ensure that WP20 (demonstration & innovation) projects translate into clinical trials for the benefit of patients

# Rare Diseases National Mirror Groups

# NMG: Objective and Role

## National Mirror Groups

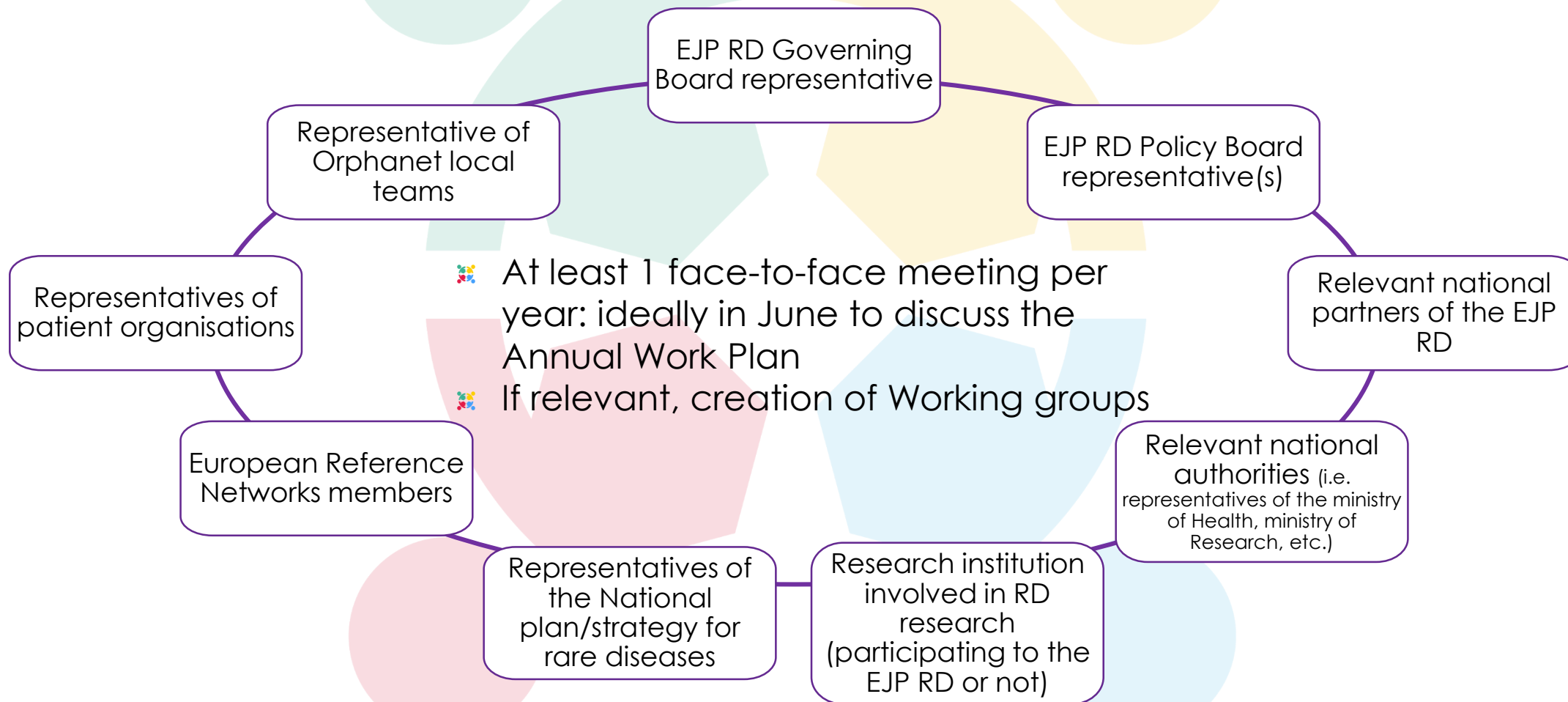
**Coordinate** the participation of **national actors** in the field of **RD** into the EJP RD activities

Define the **national position and priorities** to be reported to the EJP RD and for its Annual Work Plan

Ensure the **alignment** between **EJP RD** and **national strategy** in the field RD by promoting EJP RD actions and outcomes at national level

→ to ensure **national coordination** of and with **all rare diseases stakeholders** to facilitate the **alignment** between national and EJP RD activities, to **contribute to the objectives** of the EJP RD and **benefit from it**

# NMG: Composition and Organisation





# Rare Diseases National Mirror Groups

## FRANCE

# French National Mirror Group

🌈 Set up in May 2019 in the joint context of **EJP RD** and the **French National Plan for Rare Diseases 3** (PNMR3)

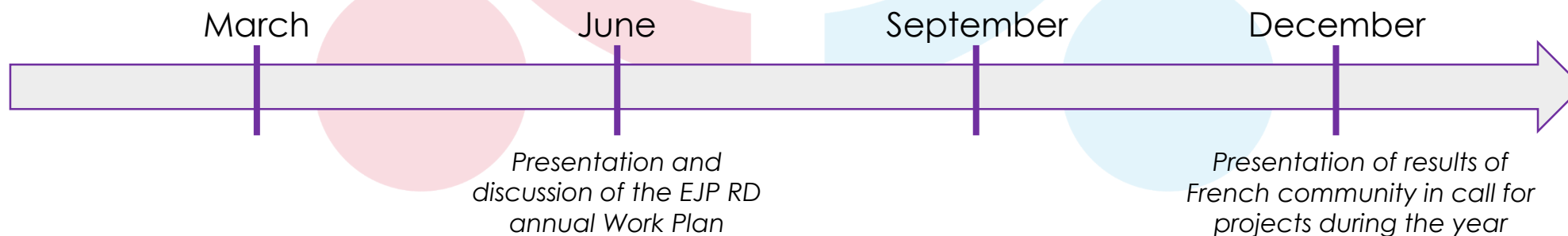
## Missions of the FR NMG:

- Coordinate the participation of rare diseases French community to EJP RD activities
- Sustain the setup of consortia, through the 'Filières', for the submission of collaborative projects to international and european call for projects
- Be pro-active to ensure the fluidity of clinical research until basic research to return to the patient

# French National Mirror Group

## Composition:

- ✘ Representatives of Research institutions involved in RD research: Inserm (Aviesan), APHP
  - ✘ Representatives of national authorities: 'Direction Générale de la Recherche et Innovation (DGRI)' and 'Direction Générale de l'Offre de Soins' (DGOS)
  - ✘ National partners of the EJP RD: Orphanet, IMAGINE, FFRD, RaDiCo, EURORDIS, AFM, ANR, ERNs
  - ✘ Alliance Maladies Rares (national alliance of RD patient organisations)
  - ✘ Representatives of Filières Santé Maladies Rares (French RD centers of reference)
- ✘ Animated by EJP RD coordinator and DGRI and chaired by Pr Elisabeth Tournier-Lasserre, research vice-chair of the PNMR3
- ✘ 4 meetings per year



# French National Mirror Group

## Benefits & challenges

### BENEFITS:

- Close collaboration with national stakeholders spanning beyond EJP RD partners
- Possibility to consult FR community in advance and establish FR position as well as collect needs
- Connection between EJP RD and national activities (RD National Plan, other initiatives e.g. Plan France Genomics 2025) allowing true impact (e.g. integration of EJP RD developed standards in FR databases; building FR scientific community in response to specific calls, etc.)
- Expanded support for national community, e.g. request to adapt EJP RD Helpdesk to accept demands in national languages or produce communication materials in national language

# French National Mirror Group

## Benefits & challenges

### CHALLENGES:

- Keeping multiple (busy) stakeholders on board → can be solved by good organisation and scoping of meetings & actions of NMG
- Good balance between national position/recommendations and final decision at EU (EJP RD) level → requires clarity and transparency on the decision making process and role of the NMG
- Alignment of agendas (national and EJP RD planning) and short deadlines for feedback (e.g. on Annual Work Plan)

# Rare Diseases National Mirror Groups

## PORTUGAL

# National Mirror Group - Portugal

- ✿ In Portugal, the set up of the NMG started during 2019
- ✿ First, the two EJP-RD Portuguese Members, INSA (National Health Institute of Health Dr. Ricardo Jorge) and FCT (Foundation for Science and Technology) agreed that a major partner which should be involved in the coordination of NMG was DGS (Directorate General for Health) that is also represented in the EJP-RD Policy Board and contacted DGS in that regard.
- ✿ - Two face-to-face meetings have occurred (one in the end of 2019 and the other in the beginning of 2020) between representatives of these 3 institutions.

# National Mirror Group - Portugal

- It was agreed that the Portuguese NMG will be coordinated by INSA in closer collaboration with FCT and DGS.
- These three institutions also decided that other national relevant stakeholders in the rare diseases field will be contacted to participate in the NMG. In a first stage it is planned to contact:
  - Portuguese Commission for the Reference Centers
  - Portuguese rare disease patient organisation
  - Orphanet Portugal
- Due to the Covid-19 pandemic situation, those contacts were not yet established but as soon as possible those stakeholders will be contacted and a first meeting will be scheduled



# Alignment with other strategic initiatives



# International Consortium for Personalised Medicine

## Personalised Medicine – A definition

*PM [ ] “refers to a medical model using characterisation of individuals’ phenotypes and genotypes (e.g. **molecular profiling, medical imaging, lifestyle data**) for tailoring the right **therapeutic** strategy for the right person at the right time, and/or to determine the **predisposition** to disease and/or to deliver timely and targeted **prevention**”.*

*According to:*

*Horizon 2020 and the European Council Conclusions on personalised medicine for patients (2015/C 421/03)*

# The International Consortium for Personalised Medicine - **ICPerMed**

Wolfgang Ballensiefen, DLR PT, Germany, ICPerMed vice-chair

July 8<sup>th</sup> 2020

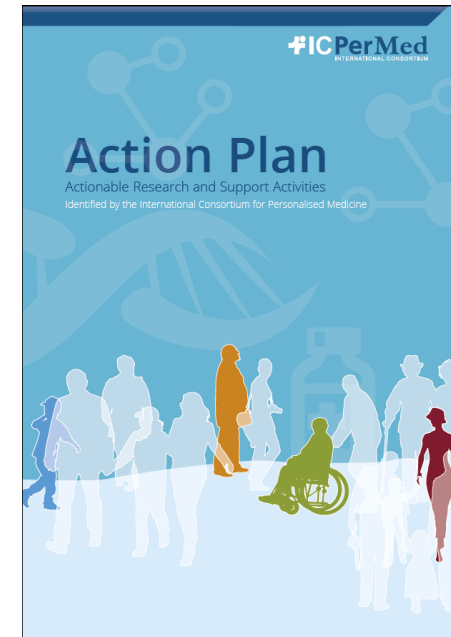


## ICPerMed – An International Consortium

- **41** European and international members and **5** observers, representing federal and regional **ministries**, **funders** and the European Commission (**EC**) in order to:
- support the personalised medicine science base through a coordinated approach of research activities,
- foster research to investigate the benefits of personalised medicine to citizens and healthcare systems and
- pave the way for personalised medicine approaches for the citizens

# ICPerMed Action Plan, March 2017

- (A) **Actionable research** and
  - (B) **support activities** for personalised medicine approaches
- Basis for the Joint Transnational Calls (JTCs) of ERA PerMed
  - Input to regional, national and European strategic discussion and actions of research funders and policy



# ICPerMed Vision Paper, December 2019

- Future vision of ICPerMed on personalised medicine research and implementation by 2030:

**“How can personalised approaches pave the way to next-generation medicine?”**

- Based on the consultation of European and international experts, covering the entire range of relevant sectors and professional backgrounds
- Available for download on the ICPerMed website:  
<https://www.icpermed.eu/en/activities-vision-paper.php>
- Including two accompanying publications

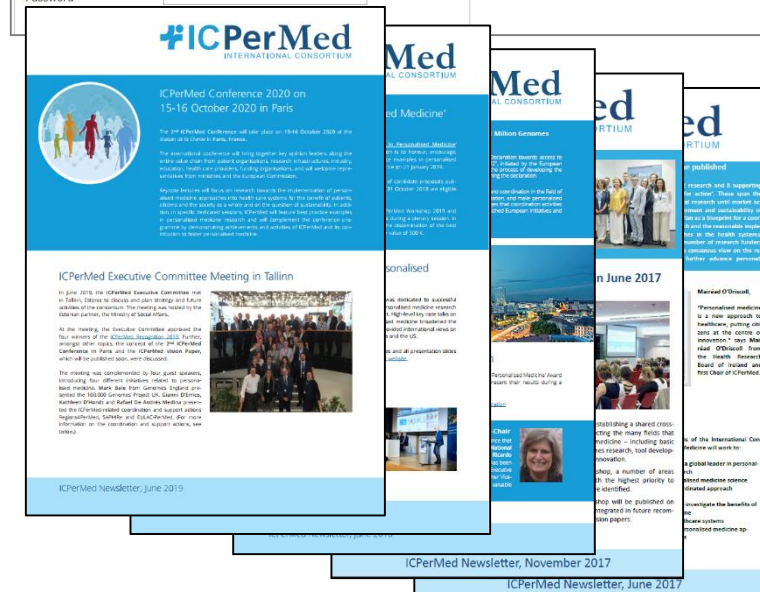
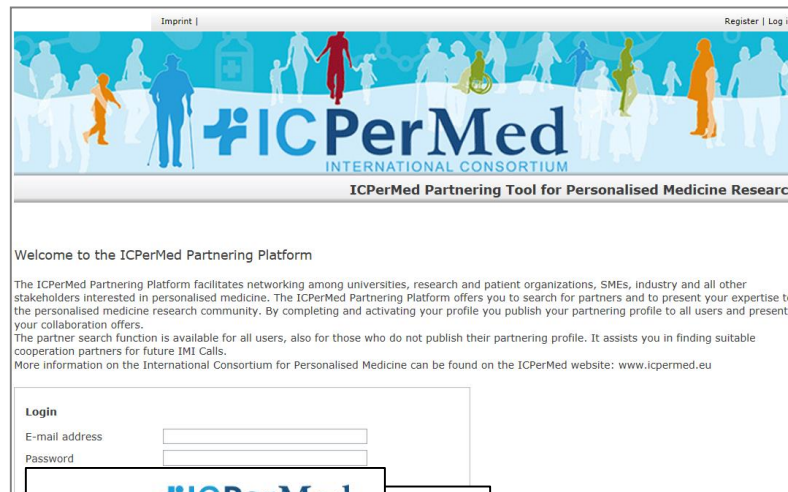


# ICPerMed Activities, Tools and Documents

- Conferences
  - **Berlin, November 2018:** “Personalised Medicine in Action”
  - **Paris, February 2021:** “Personalised Medicine - From Vision to Practice”
- Workshops
  - **Milano, June 2017:** “Innovative Concepts on Data Generation and use for Personalised Medicine Research”
  - **Madrid, November 2019:** “Personalised Medicine for All Citizens and Patients within Sustainable Implementation”
- ICPerMed Recognition:
  - 2018, 2019 and 2020
- Workshop and „State of the Art” reports on the webpage
- 3 Best Practise examples on the ICPerMed webpage



# Webpage, Partnering Tool & Newsletter etc.



- Platform for research organisations to find potential cooperation **partners**
- Over 660 entries so far
- was used e.g. by ERA PerMed and Flagship Canada/EU calls
- **Newsletter** and Stakeholder Group for staying connected with ICPeMed
- registration on ICPeMed website

# ICPerMed Funder's Survey and Database




**Welcome to the ICPerMed mapping database - funding for research in Personalised Medicine**

The ICPerMed database gives an overview on the actions in the field of Personalised Medicine (PM) performed in the ICPerMed member states and may assist you in finding suitable funding opportunities and information about potential funders in your respective country and/or region.

Via the ICPerMed database, ICPerMed members (e.g. funding agencies and ministries) provide available information about their national/regional funding activities in PM. The different national/regional funding schemes and general funding information are described. The database allows the user to search for programmes susceptible to finance research projects in the field of PM. This includes not only programmes that are ongoing but also those that are already completed as well as others that will start in the near future.

The ICPerMed database is open to the public after simple registration.

The ICPerMed database reflects the progress of the ICPerMed consortium towards the objectives and implementation of the ICPerMed Action Plan. More information on the International Consortium for Personalised Medicine (ICPerMed) can be found on the ICPerMed website: [www.icpermed.eu](http://www.icpermed.eu).

E-Mail

Password

Remember me

[Forgot your password? Register](#)

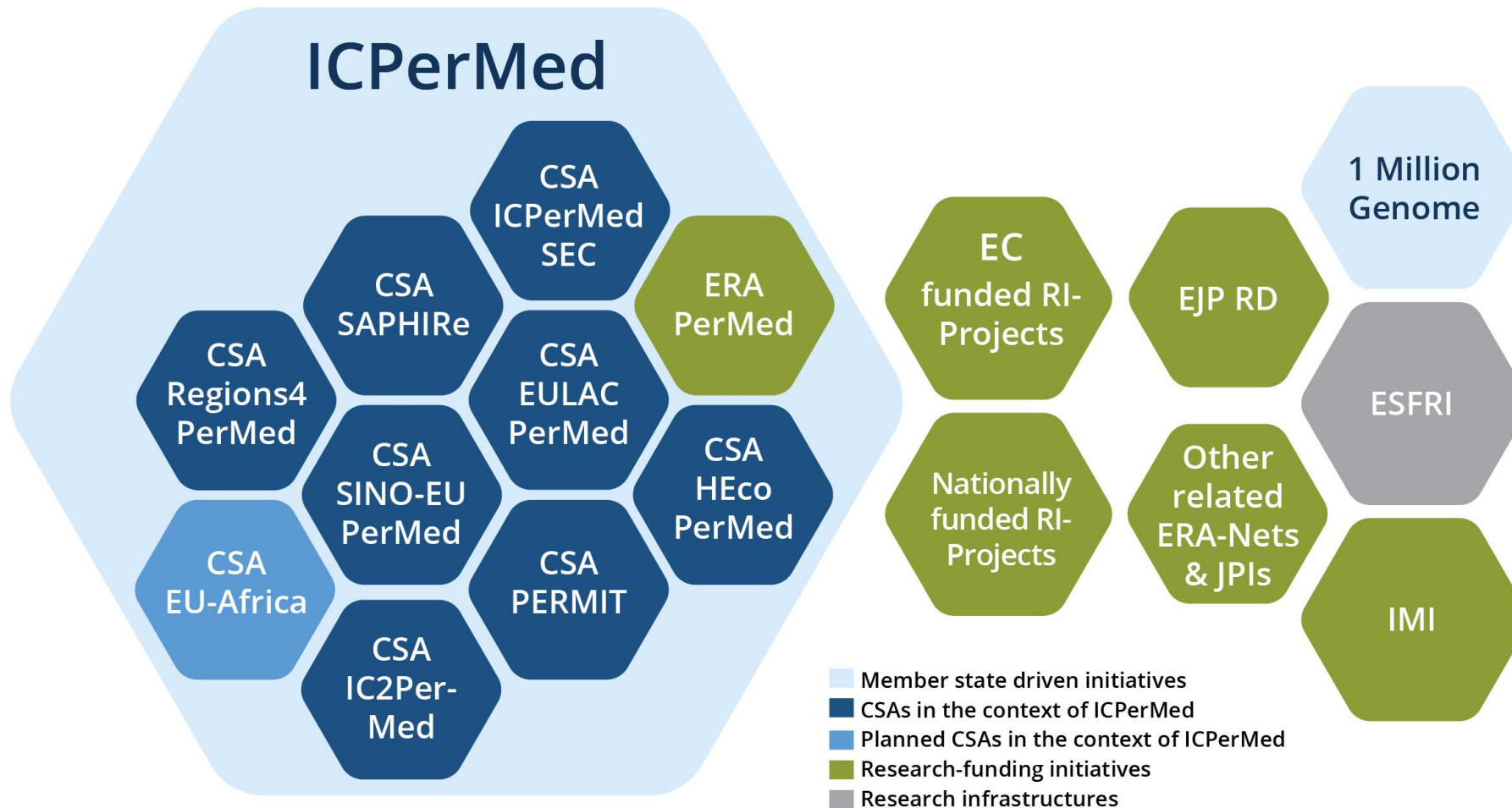
- **Mapping Activity:**  
Ongoing funding programmes in Personalised Medicine
- Survey sent to all ICPerMed members
- Results are available in the ICPerMed mapping database
- open to the public after simple registration

## ICPerMed Statements

ICPerMed published joined statements on important topics or developments related to Personalised Medicine.

- 1<sup>st</sup> statement: May 2018 about the One Million Genomes Declaration: <https://www.icpermed.eu/en/statement-concerning-the-1-million-genomes-declaration.php>
- 2<sup>nd</sup> statement: November 2019 (together with ERA PerMed) about a planned European Partnership for Personalised Medicine (EP PerMed): <https://www.icpermed.eu/en/joint-icpermed-and-era-permed-statement.php>

# ICPerMed „Family“ and related initiatives



More information about ICPerMed are  
available

on the Webpage: [www.icpermed.eu](http://www.icpermed.eu)

or via mail request to: [ICPerMed@dlr.de](mailto:ICPerMed@dlr.de)

The background features four stylized human figures in teal, yellow, pink, and blue, arranged in a circle. Each figure is composed of a circular head and a larger, abstract body shape. The teal figure is at the top left, yellow at the top right, pink at the bottom left, and blue at the bottom right.

# 1+ Million Genomes Initiative



# 1+MG Initiative & B1MG Project

---

EJP RD Policy Board and Governing Board meeting  
8th July 2020

Serena Scollen (ELIXIR) B1MG Project Coordinator

On behalf of 1+MG in Ruben Kok and Bruno Dallapiccola's absence

# 1+MG Declaration of cooperation - April 2018



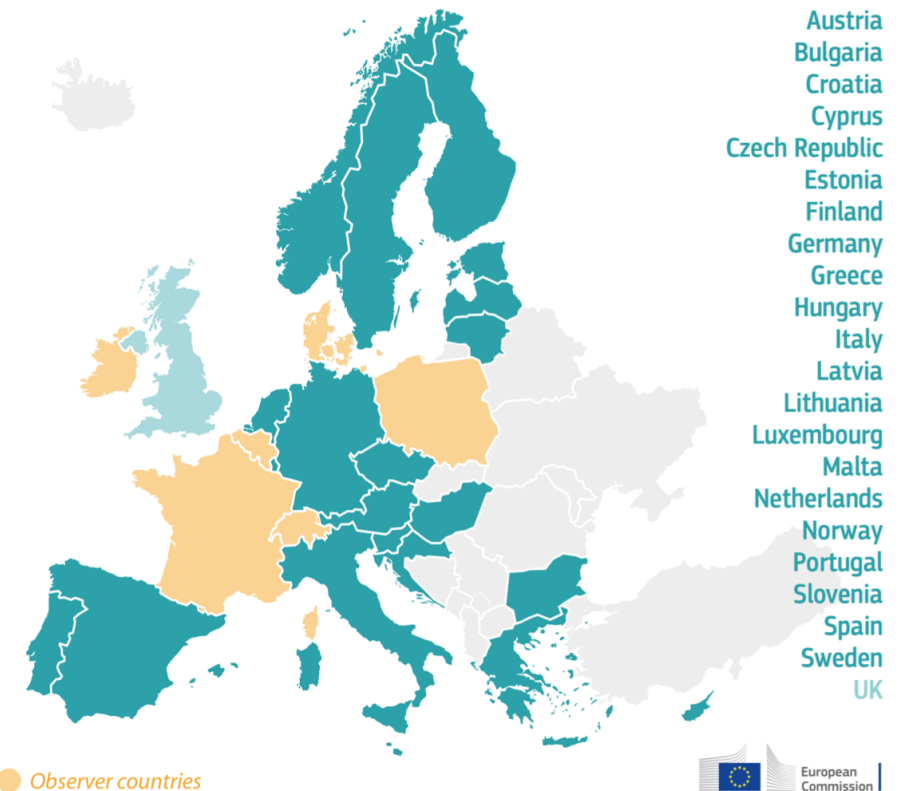
DECLARATION OF COOPERATION

*Towards access to at least 1 million sequenced genomes in the European Union by 2022*

<https://ec.europa.eu/digital-single-market/en/european-1-million-genomes-initiative>



## EU countries agreed to cooperate in linking genomic data across borders



**22 countries have now signed; 6 are observers**





# Working groups addressing key issues

## Coordination: 1+MG coordination team & NMGs coordinators

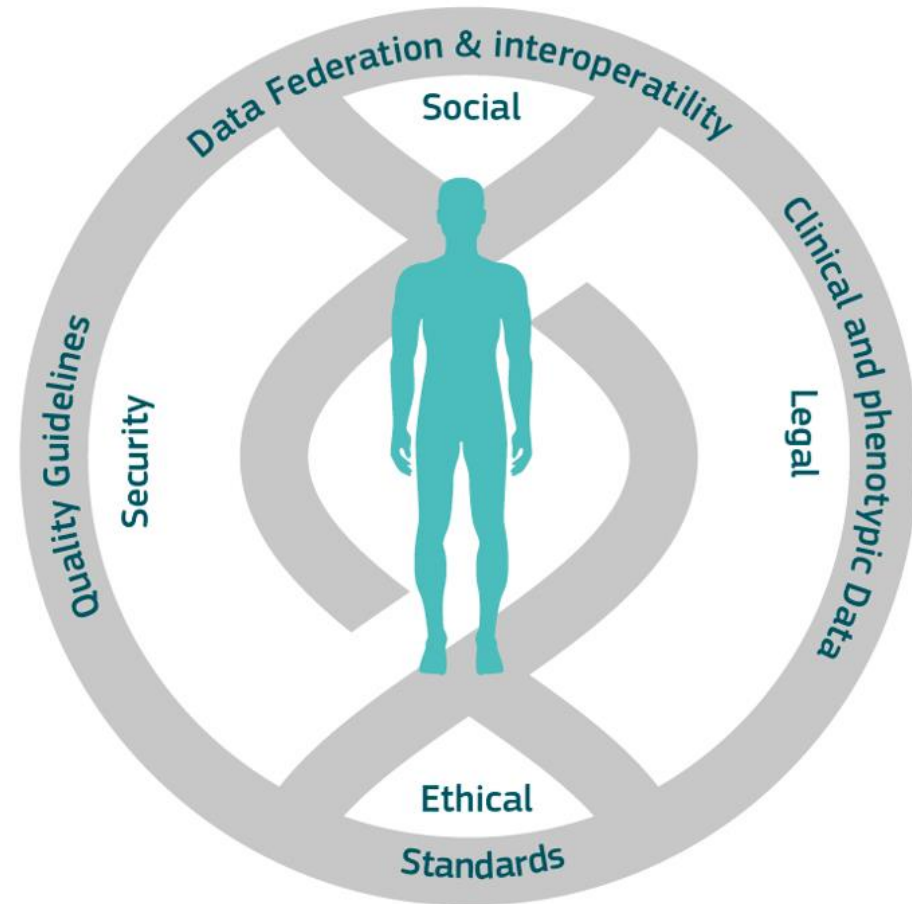
### 1. Organisation and governance

## Technical WGs: NMGs experts

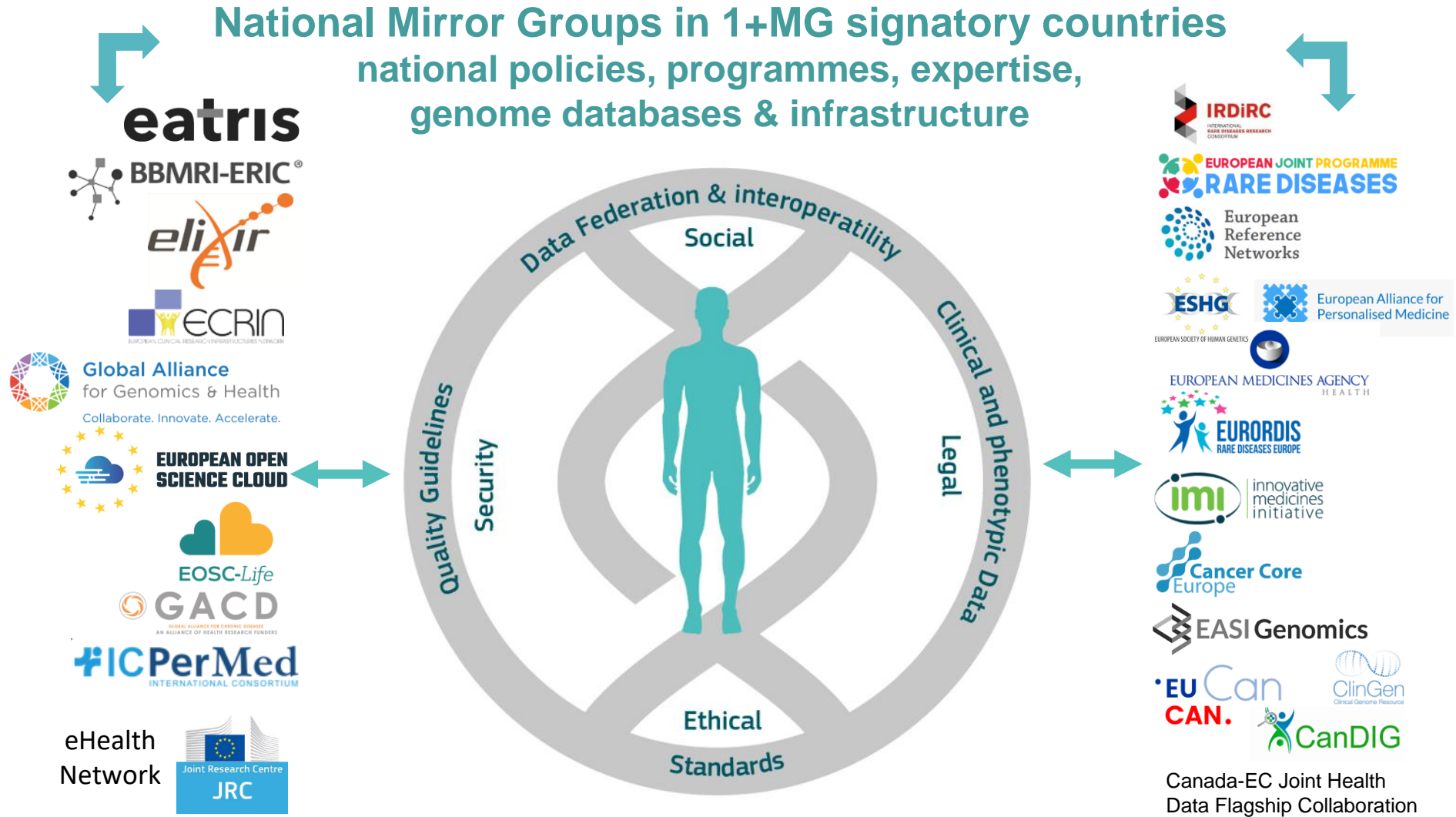
1. ELSI aspects
2. Clinical data
3. Quality
4. Infrastructure
5. Healthcare implementation

## Use cases WGs: NMGs experts

1. Industry involvement
2. Rare diseases
3. Cancer
4. Common Complex diseases
5. Infectious diseases -> COVID-19



# Establishing a European learning framework



# 1+MG Roadmap adopted Feb 4 2020

## Objectives

1. Engage local, regional, national and European stakeholders to define the requirements for cross-border access to genomics and personalised medicine data
1. Translate requirements for data quality, standards, technical infrastructure, and ethical, legal and social issues (ELSI) into technical specifications and implementation guidelines that captures European best practice
1. Drive adoption and support long-term operation by organisations at local, regional, national and European level by providing guidance for phased development via a maturity model and a methodology for economic evaluation



# 1+MG Roadmap – key aspects

- **Key use cases: Rare diseases, Cancer, Common & Complex diseases**
  - Highly different in approach, similar in need for genome information
  - Direct impact via national healthcare systems
  - Many ongoing major European initiatives to build on
- **Federated infrastructure offering access to local data across borders**
  - HQ WGS is focus, WES supportive; perspective of PRS towards panels
  - Minimal set of associated phenotypic data
  - Distributed analysis; requires harmonisation of data (FAIR, clinical standards)
- **Collective ethical and legal framework, national flexibility**
  - ELSI toolkit for sharing very privacy sensitive genomics and health data



# 1+MG Roadmap - Timeline



**1+ million  
accessible genomes**



2020

## “Engage”

### **Governance, cooperation, and collaboration**

Governance model, national “mirror groups”  
Terms and conditions for distributed access  
Guidelines on good genomic practice  
Funding, communication

2021

## “Translate”

### **Infrastructure, guidelines, and pilots**

Distributed, authorised and secure access  
GDPR, legal guidelines  
Technical specifications, interoperability  
Secure infrastructure and tools

2022

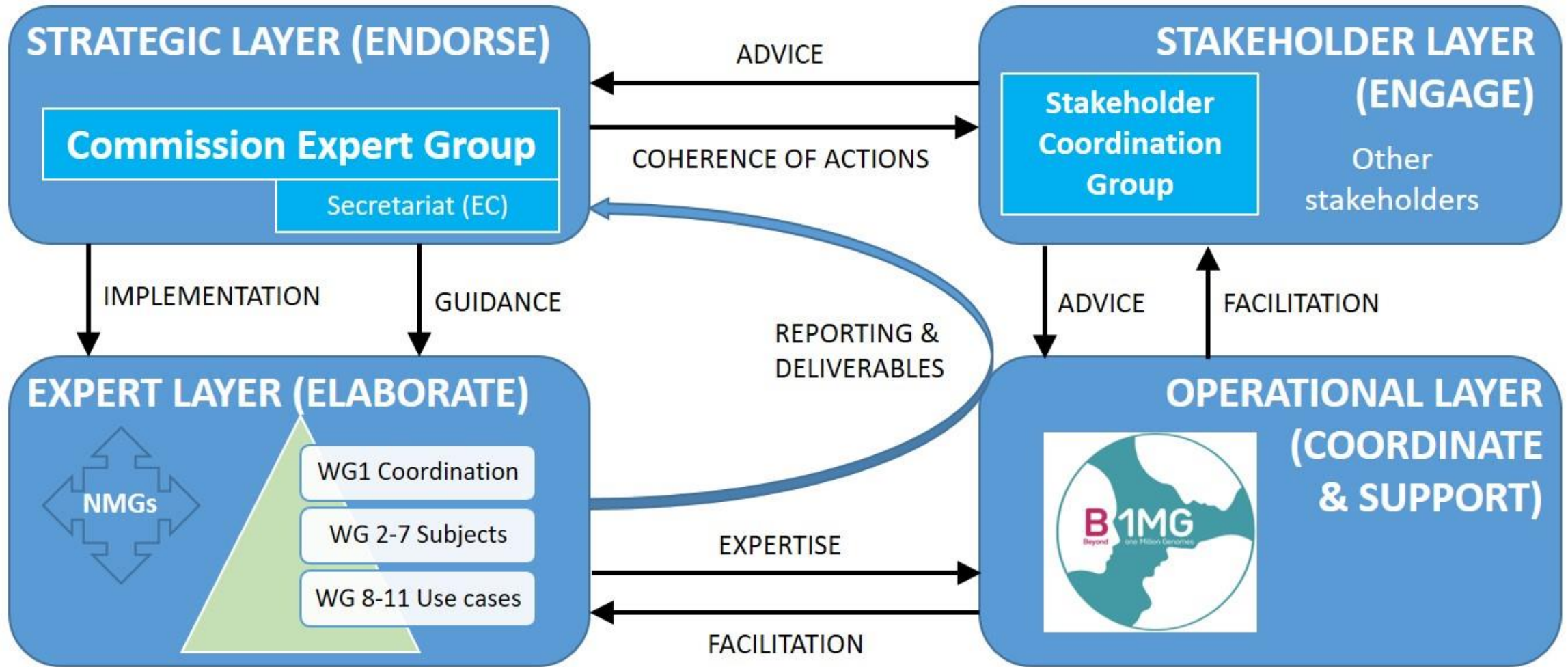
## “Drive”

### **Sharing, scaling, and sustaining**

Coordinated data governance  
Plan for scale-up of the infrastructure  
Economic evaluation  
Roadmap for longer-term sustainability

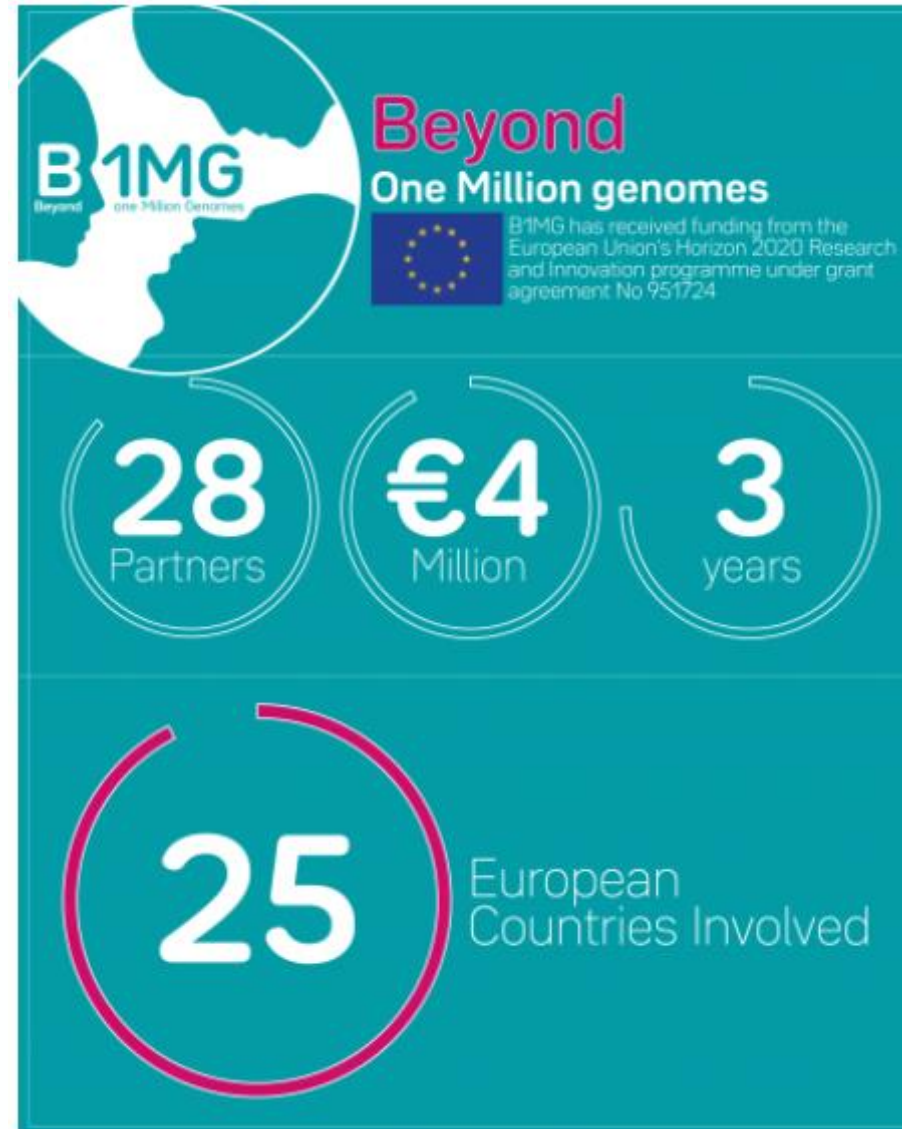


# Organisational structure



# B1MG Project

- Coordination and Support Action
  - No (or limited) research driven activity
  - 1+MG signatories countries + France, Belgium & Switzerland
- Started 1st June 2020 to May 2023
  - Held the project kick off meeting June 4th



# Consortia: 28 partners



Legal Pathways Life Sciences Law



European Alliance for Personalised Medicine



- Many many stakeholders, as well as the 25 countries involved



# B1MG project will (aligned to 1+MG roadmap)...

Drive the development of **European infrastructure for federated and secure cross-border access to personalised medicine data** by:

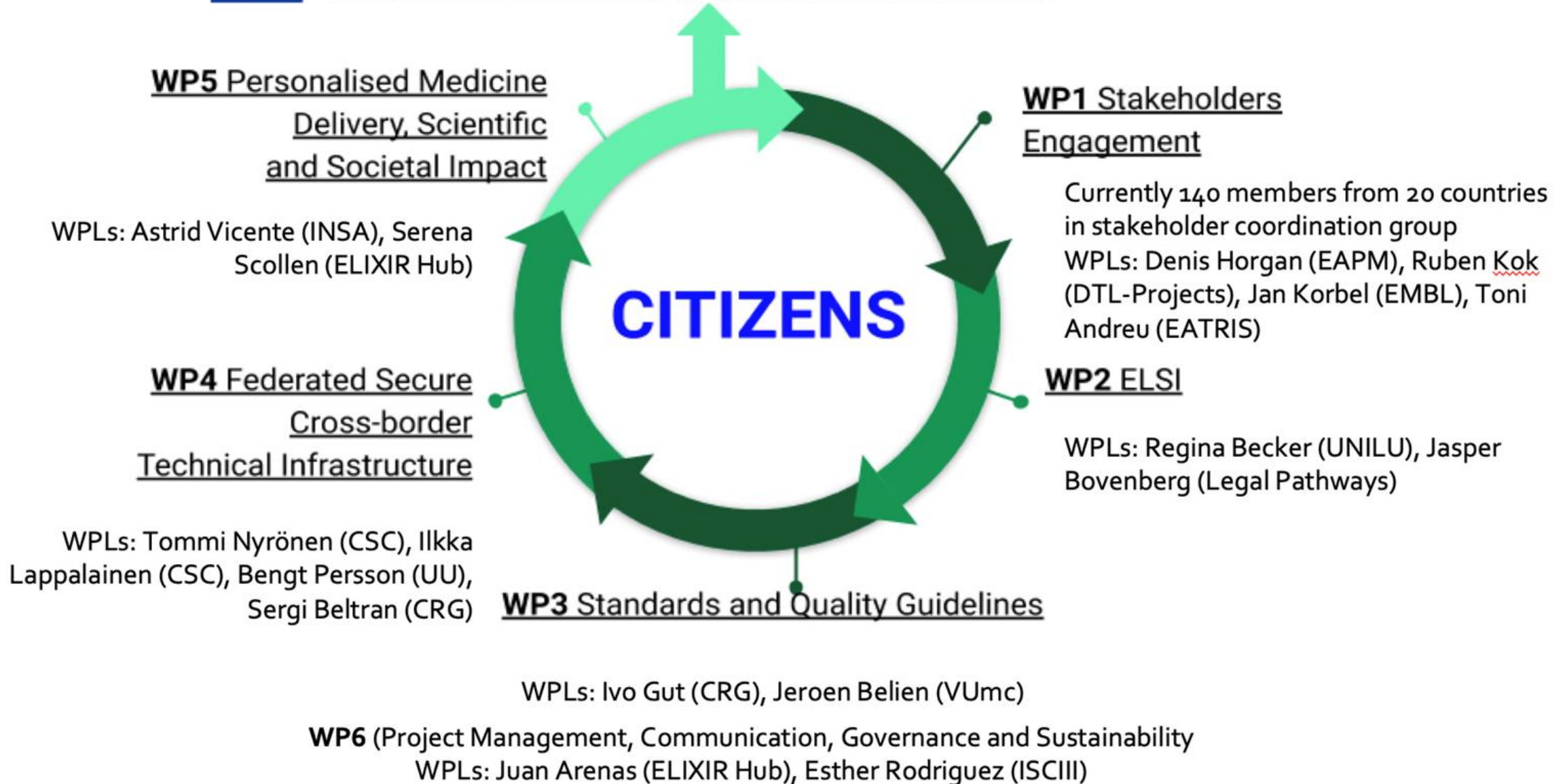
Engaging local, regional, national and European stakeholders to **define the requirements** for cross-border access to genomics and personalised medicine data

Translating requirements for data quality, standards, technical infrastructure, and ELSI into **technical specifications and implementation guidelines** that captures European best practice

Driving **adoption and support long-term operation** by organisations at a local, regional, national and European level by providing guidance on phased development (via the B1MG maturity level model), and a methodology for economic evaluation



## FEDERATED SECURE CROSS-BORDER ACCESS TO PERSONALISED MEDICINE DATA BOOSTS EU RESEARCH AND INNOVATION



### Implementation

- Maturity level model: guidance enabling self-assessment and step-by-step integration with infrastructure standards and recommendations
- Country visits for knowledge exchange
- Economic model for evaluating PM approaches

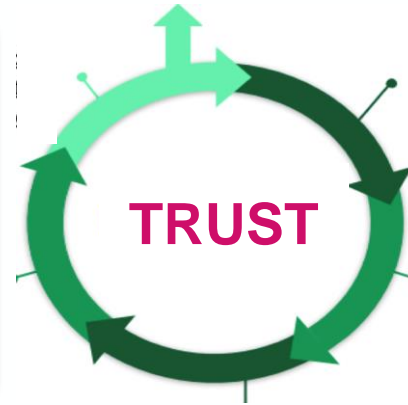
# PM data can be accessed across Europe successfully

### Stakeholder Coordination Group

- to contribute and shape outcomes
- national and EC policy makers, academics, industry, patient and citizen organisations, genetics and health care professionals, healthcare care system representatives, planners, funders...

### Infrastructure requirements to enable cross-border services

- security requirements, interoperability standards
- technical components
- coordinate catalogue of existing 'synthetic' data
- safely test cross-border interoperability



### ELSI toolkit

- consolidate national requirements, policies and recommendations
- build upon good practice developed in EU projects and national genomic initiatives

### Standards and quality guidelines

- minimal requirements for the provenance of the samples, the generation of the whole genome sequencing data including, the bioinformatics analysis, and the description of patient-specific phenotype and clinical data considering national particularities, such as clinical systems and languages

### Sustainability and guidance for establishment of National Mirror Groups

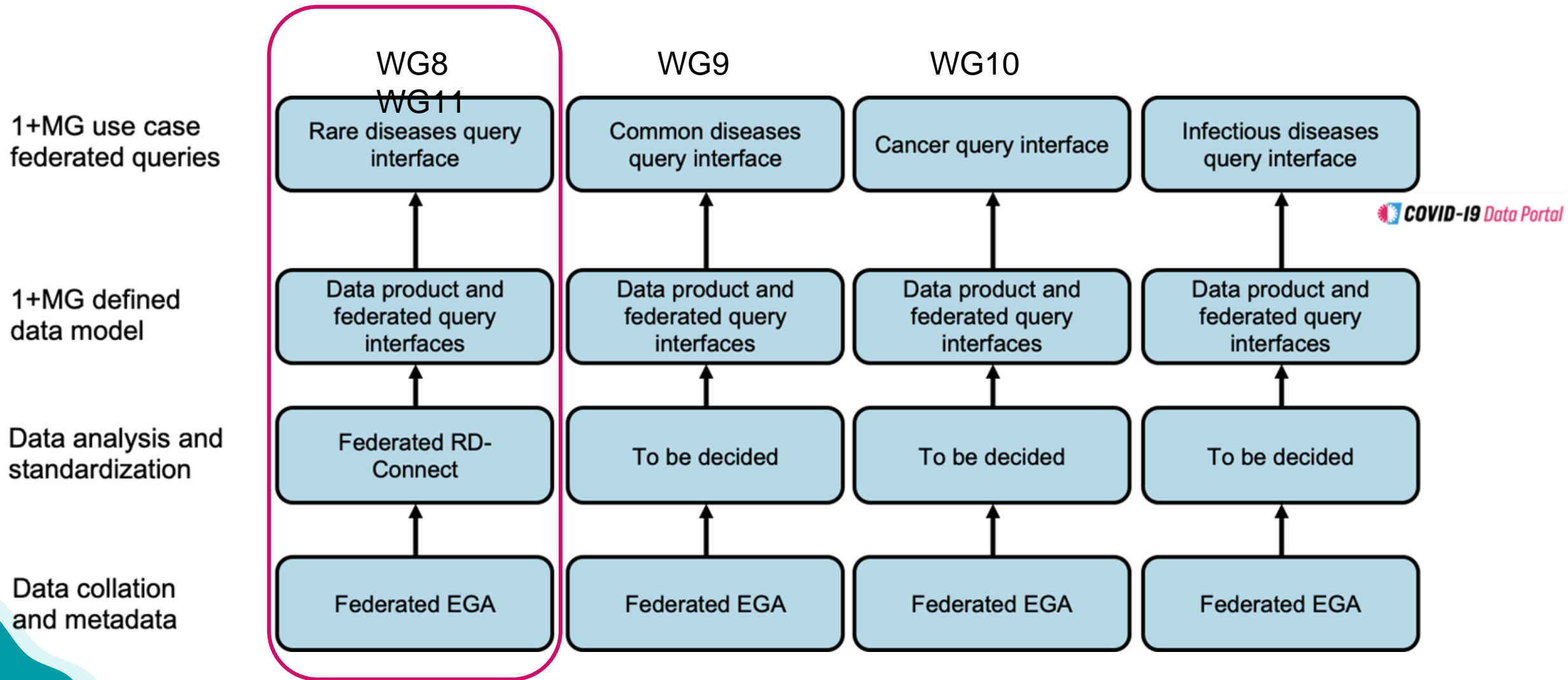
# Maturity level model

B1MG Maturity Level Model Key Areas (Illustrative Example)					
#	Standards	Security	Federate Cross-border Infrastructure	Personalised Medicine Delivery	Citizens
5	Optimized	Transnational access to data according to applicable regulations and best practices. Machine ready.	Federation to machine ready infrastructure based on common global standards	Personalised Medicine delivered at global scale, reduction of cost for the Health Care Systems	Global access to data allow to tackle global issues and accelerate translation into Health Care Systems
4	European	Machine ready transnational access to data according to applicable regulations across EU	<ul style="list-style-type: none"> <li>Project outcomes integrated into the model</li> <li>Practical guidance on the steps required for an organisation to engage with the federated secure cross-border infrastructure to access personalised medicine data at local, regional, national and european level.</li> <li>For situations where no integration exists between genomics and health data, all the way to a level where data can be accessed transnationally and fulfilling all applicable regulations</li> </ul>		diagnostic and therapeutic data at EU level. Improvements on patient quality of life.
3	National	Alignment of regulations across national level			patients benefits from PM data at national level
2	Regional	Alignment of regulations across regional level			patients benefits from PM data at regional level
1	Initial	Lack of alignment with regulation prevents sharing PM data			No national infrastructure available

# 1+MG WG participation in B1MG WPs

1+MG WG vs B1MG WP		B1MG (EC H2020)					
		WP1 Stakeholders	WP2 ELSI	WP3 Standards & Quality Guidelines	WP4 Federated secure cross-border Technical infrastructure	WP5 Delivering Personalised Medicine cross-borders	WP6 Coordination Office
1+M Genome	Scope, stakeholders and governance (WG1)	Stakeholders					Governance, Sustainability
	Ethical, Legal, and Societal Issues (WG2)		Ethical, Legal, and Societal Issues				
	common standards for capturing clinical and phenotypic data requirements (WG3)			Common Standards + Phenotypic data requirements			
	good sequencing practice / development of standards for clinical interpretation (WG4)			Sequencing practices + standards for clinical interpretation			
	interoperability, transfer between countries, local/federated system incl. systems development and deployment and data access governance (WG5)				interoperability, transfer between countries, local/federated system incl. systems development and deployment and data access governance (Technical Aspects)		
	health economics and outcome research (WG6)					health economics and outcome research	
	involvement of the private sector (incentives, IP, contribution and access) (WG7)	involvement of the private sector					
	rare diseases (WG8)	rare diseases	rare diseases	rare diseases		rare diseases	
	cancer (WG9)	cancer	cancer	cancer		cancer	
	common, complex diseases (WG10)	common, complex diseases	common, complex diseases	common, complex diseases		common, complex diseases	
	National Mirror Groups (NMG)	Mirror Groups					Mirror Groups

# Rare diseases as a B1MG use case



# Additional points of alignment

- Serena Scollen (ELIXIR Hub and B1MG Coordinator) to join EJP RD Executive Committee meetings to provide regular updates when requested
- Juan Arenas (B1MG WP6 lead) attending EJP RD Coordination meetings and EJP RD GA
- 1+MG WG members and B1MG participants are involved in numerous tasks across all 4 pillars (e.g. Ivo Gut, Sergi Beltran,..)
- Overlap in stakeholders e.g. GA4GH

# Call for action

- Get in contact with your National Mirror Group coordinators on the 1+MG to be involved in the 1+MG WGs that are relevant for you
  - Participation welcome, but particularly in WG6, WG7 and WG11
  - Additional information about National Mirror Groups [b1mg-nmg-coordinators@elixir-europe.org](mailto:b1mg-nmg-coordinators@elixir-europe.org).
- [Register to B1MG Newsletter](#) to be aware of project progress
- Further questions: [b1mg-coordination@elixir-europe.org](mailto:b1mg-coordination@elixir-europe.org)



# Discussion / Questions



# Priorities/planning: timeline 2020 (2)

	I-2020	II-2020
<b>WG5</b>	<ul style="list-style-type: none"> <li>- Suitable synthetic or public data sets identified for demonstrators</li> <li>- National contact details collated for appropriate technological solutions</li> </ul>	<ul style="list-style-type: none"> <li>- Selection of first ring of infrastructure test-locations across signatory countries</li> <li>- Proof of Concept of federated data storage infrastructure: demonstrator supporting secure and interoperable data management processes, e.g. based on Federated EGA technology but integrated to the national context. At this point demonstrator supports connection of specified data resources.</li> </ul>
<b>WG6+1</b>	<ul style="list-style-type: none"> <li>- Inventory of HTA methodology initiatives</li> </ul>	<ul style="list-style-type: none"> <li>- Inventory of best practices on:               <ol style="list-style-type: none"> <li>a) education of professionals,</li> <li>b) information for citizens/patients</li> <li>c) effective communication material</li> </ol> </li> <li>- Engagement with relevant mirror groups and other experts from national healthcare systems for co-creation of Maturity Level Model</li> </ul>
<b>WG8-10</b>	<ul style="list-style-type: none"> <li>- Open and effective communication channels with relevant national mirror groups</li> <li>- Inventory of use cases genomes available and situation regarding:               <ol style="list-style-type: none"> <li>a) consents for sharing;</li> <li>b) phenotypic data available;</li> <li>c) sequencing standards</li> </ol> </li> <li>- Inventory of stakeholders and engagement with relevant initiatives for each use case</li> <li>- Inventory of available national and European funding and funding gaps, for each use case</li> <li>- Inventory of funding opportunities for each use case</li> </ul>	<ul style="list-style-type: none"> <li>- Alignment of use cases activities with relevant initiatives roadmaps</li> <li>- Interoperability with WG 2, 3 and 4 regarding challenges and needs specific to each use case</li> <li>- Funding applications for use cases</li> </ul>



# Summary and Next Steps

Thank you