EJP RD

European Joint Programme on Rare Diseases

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First report from the face-to-face ExCom and Policy Board meeting

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Partner 1 – INSERM

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PU - Public

EJP RD Executive Committee

3rd of July 2019

11:00 – 18:00

Covent Garden building, room COVE A2 0/129, Square Rogier, Brussels

Attached document:
Slides presented during the meeting (ppt presentation) – See Annex 1 (p23)

List of participants:

Name Surname	Institution	Role	Presence
Ana Rath	INSERM (Orphanet)	P2 coleader WP10 -11	Present
Anthony Brookes	ULEIC	WPL 10 - 12	Present
Anton Ussi	EATRIS	P4 coleader WPL 3 - 19	Present
Biruté Tumiene	VUHSK	P3 coleader 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
Laura Cellai	ISS	WP 14	Present (TC)
Daria Julkowska	INSERM	Coordinator WPL 1 - 5	Present
Domenica Taruscio	ISS	WPL 2	Present
Eva Bermejo-Sanchez	ISCIII	WPL 2	Present
Manuel Posada	ISCIII	WPL 2	Absent
Franz Schaefer	UKL-HD	P2 coleader WPL 13	Present
Holm Graessner	EKUT	WPL 17	Present
Irit Allon	CSO-MOH	WPL 9	Present
Krystyna Chrzanowska	IPCZD	WPL 18	Present
Luca Sangiorgi	IOR	Chair of ERN Research Group	Absent
Marco Roos	LUMC	WPL 12	Present
Maurizio Scarpa	HSK	WPL 20	Present
Ralf-Dieter Hilgers	UKA	WP20	Present
Tania Berger	UKA	WP20	Present
Ralph Schuster	DLR	P1 coleader WPL 6	Present
Rima Nabbout	AP-HP	P4 coleader	Present

		WPL 20	
Roseline Favresse	FFRD	WPL 16	Present
Emilie Bonnaud	FFRD	WP 16	Present
Serge Amselem	INSERM (RaDiCo)	WPL 3	Absent
Sonia Gueguen	INSERM (RaDiCo)	WPL 3	Absent
Daphne Jaoui	INSERM (RaDiCo)	WPL 3	Absent
Sergi Beltran	CNAG-CRG	WPL 11	Present
Sonja van Weely	ZonMw	P1 coleader WPL 7	Present
Stefano Benvenuti	FTELE	WPL 4 - 19	Excused
Elena Beltrami	FTELE	WPL 4 - 19	Present
Virginie Bros-Facer	EURORDIS	P3 coleader WPL 15 - 18	Present
Viviana Gianuzzi	FGB	WPL 4	Present (TC)
Annalisa Landi	FGB	WPL 4	Present
Bertrand Schwartz	MESRI	MESRI	Absent
Juliane Halftermeyer	IT	Coordination team	Present

Agenda:

09:00 – 11:00	*AREB meeting (for AREB and OG members only)*	
10:00 – 11:00	Photo session for available ExCom members	
11:00 – 11:15	Coffee break + photo session continues	
	Welcome from the coordination	
11:15 – 13:00	Key Performance Indicators for the EJP RD - presentation & discussion with ExCom	Giovanni Migliaccio (CVBF)
	Prioritization strategy process - in depth discussion with ExCom on the selection of elements to prioritize *(methodological aspects will be discussed with Policy Board)*	Eva Bermejo and Manuel Posada (ISCIII)
13:00 – 14:00	**LUNCH**	
14:00 – 14:30	Pillar 0 : Update on EJP RD communication strategy	Eleonora Passeri (Inserm) + All
14:30 – 16:10	WPs and Pillars operational issues • Pillar 0 • Pillar 1 • Pillar 2 • Pillar 3 • Pillar 4 Topics pre-identified in advance of the meeting: - EJP RD in IRDiRC (as official partner) – which strategy to adopt - EJP RD collaboration with non-EJP RD partners - Central Helpdesk - Financial questions/issues from WPs - Sustainability of EJP RD activities (training, Pillar 2 tools, etc.) - EJP RD in GA4GH – current status and next steps	All
16:10 – 16:40	**Coffee break**	

16:40 – 17:00	WPs and Pillars operational issues continue	All
17:00 – 18:00	ERNs & EJP RD: common agenda Including EJP RD and individual ERN training activities alignment	Franz Schaefer (UKL-HD) and Luca Sangiorgi (IOR)
18:00 – 18:15	AOB	

19:30	Dinner

Key Performance Indicators for the EJP RD - presentation & discussion with ExCom

See slides 3-13 for complete information

2 types of KPIs can be defined:

- KPIs for results: specific to WPs. KPIs of results can be the achievement of deliverable or milestone.
- KPIs for performance measure of the procedures: standardized metrics per Pillar/for the whole EJP RD

Objective: set up maximum 10 KPIs per Work Package ➔ KPIs should allow us to measure the efficiency of the process, not only the result of the process.
It is important that KPIs measure something that matters for the project and show that the project is doing well. The KPIs should also allow to identify emerging problems before the milestone/deliverable in the project.
We need to define a process to detect a problem in the project and have a mechanism to follow the management of the project: for example, it is important to check at the level of the WP the participation of partners, in case of no participation of a partner, a process of action should be defined (first involvement of the Pillar leader then of the Coordination or Operating Group if necessary)

Decisions:

- ⇨ It has been agreed that indicators such as email's answer rate or participation to meeting will not be used as KPIs in the EJP RD
- ⇨ WP leaders must define indicators of results in their WPs (Key Results Indicators (KRIs))
- ⇨ A process to measure the performance of the project as a whole and to identify potential problems and mitigation process for performance has to be defined and agreed on: common rules must be agreed at the Pillar level.

Prioritization strategy process - in depth discussion with ExCom on the selection of elements to prioritize

See slides 14-60 for complete information

- ⇨ The new version of the deliverables D2.1 – Final list of prioritization criteria and D2.2 – Prioritization scheme including decision-making process should be sent to the ExCom to be discussed and agreed. This document has not been shared prior the meeting, thus limiting the comments from the ExCom.

Recommendations from the ExCom:

- The EJP RD work plan for the 5 years is very detailed but we should keep the opportunity of flexibility given by the Annual Work Plan description.
- The Prioritization scheme (once validated by the ExCom) could be a tool to make the project better thanks to that flexibility. Thus, it should be transformed into "Guidelines for prioritization".
- Some criteria should be more present like innovation.
- It is also important to have input from the community and from end-users in this process. This input has to be balanced with the risk measurement.
- The Policy Board should be involved to bring specific national needs in the discussion and advise the consortium on issues of relevance.

Decisions:

- ⇨ The "Guidelines for prioritization" document have to be revised by the TL based on the comments from the ExCom and shared with the ExCom for final comments and validation. Notably, it should not be limited to only one method but also include some other methods that may be proposed by the ExCom members.
- ⇨ This scheme will not be used to compare tasks one to another but will be applied to a process of decision (at the level of the Task or WP), at the condition that alternatives that need to be prioritized and to choose between are present.
- ⇨ The prioritization scheme will also be used internally in each pillar in order to help decision process on particular tasks.

Pillar 0: Update on EJP RD communication strategy

See slides 61-76 for complete information

The communication strategy should focus first on European countries before going further international. However, it is also important to bring new ideas for communication that could go beyond EU.

There is a real need for research communication to and from ERN ➔ to be discussed with Eleonora to see what can be done (for example, we could have an ERN corner on the EJP RD website to feature specific content from ERN networks).

It will be important also to continue to highlight publications from E-Rare-3 and future EJP RD JTCs funded projects.

We could also think of releasing some content in the language of the networks: this would need the help of partners.

Decisions:

- ⇨ Pillar leaders, WP leaders and Task leaders have to share information and give input from the EJP RD activities to Eleonora in order to push forward some activities.
- ⇨ The newsletters should be renamed to get no confusion between the 'internal' and 'external' ones.

WPs and Pillars operational issues

EJP RD in IRDiRC (as official partner) – which strategy to adopt
See slides 77-78 for complete information

During the last IRDiRC meeting in Leiden, some connections between IRDiRC Task Forces and the work planned in the EJP RD have been established: important not to be redundant in our activities.
Spontaneous candidature to the Task Forces (by EJP RD members for example) can be done through the IRDiRC website ➔ EJP RD coordination can help to share these information to its network.

Should EJP RD be part of IRDiRC?
The EJP RD could be represented in the FCC (Funders Constituent Committee). For this we need to show that at least 10 million dollars of funding will be spent in 5 years by the project.
In any case it is important that people already in IRDiRC and participating in the EJP RD make the link between initiatives.
The Task Forces (TF) will now be at the center of the organization of IRDiRC, it will be important to have representatives of the EJP RD in all Task Forces. If we identify a synergy between a TF and the EJP RD, it should be mandatory to have an EJP RD representative in the TF.

Decision:

- ⇨ Based on the discussion, a strategy will be proposed to the EJP RD General Assembly for comments. Discussion will take place during the GA in September to take a common decision and agree on a procedure and a representative if we apply for a membership in IRDiRC.

Interaction with other important stakeholders
See slides 79-81 for complete information

GA4GH: Tony Brookes and Sergi Beltran were nominated as champions to represent the EJP RD as driver project in GA4GH. As a driver project, EJP RD have to indicate what are our needs, participate in the elaboration of new standards and test those standards, products ➔ there is a need to have more people involved. Partners will be contacted to propose them involvement is specific GA4GH activities.

1+MG: countries engaged in the initiative should engage to facilitate shareability of data. Daria is involved in the Working Group 8 – Use case RD.

C4C: the connection between projects is made through various levels as several EJP RD partners are involved in both projects.

Rare2030: some EJP RD partners involved; currently no formalized collaboration.

EIT Heath: could build some common actions based upon trainings developed by the EJP RD and activities of Pillar 4.

Other proposed programmes to engage with:

- All projects related to FAIRification (FAIRFAIR, FAIR4Helath, GoFAIR etc.) should be connected
- Important to have a connection with IMI: have been invited to be part of the Policy Board and also in some tasks of the project (i.e. WP20). *Post meeting information: an IMI representative has been nominated in the EJP RD Policy Board*
- SolveRD: no need for a formal link but we should be sure that existing tools are being used and not redone.

Action:

⇨ A session dedicated to presentation/discussion with the stakeholders/projects we want to collaborate with will be added in the agenda of ExCom meetings and the representatives of the respective initiatives will be invited to participate.

EJP RD collaboration with non-EJP RD partners
See slides 82-83 for complete information

When collaboration with non-EJP RD is needed, a CDA (Confidential Disclosure Agreement) should be signed between the so-called Consultant and the Pillar leader. Memorandum of Understanding (MoU) can also be a choice if needed.

In Flagship projects, a category of "supporters" has been defined. This could be adapted to the EJP RD.

⇨ Rules of engagement of associated partners have to be defined: associated partners DO NOT represent EJP RD except if they are authorized.

Central Helpdesk
See slides 84-85 for complete information

The needs for the Central Helpdesk are increasing from activities in different Pillars ➔ will need IT development to set up a ticketing tool allowing to follow all emails, timelines and store answers.

Decisions:

⇨ a simple contact form with sub-categories will be first put in place on the EJP RD website
⇨ a conference call will be set-up to discuss the needs and how to proceed for a more elaborated tool.

Financial questions
See slides 86-87 for complete information

Travel budget:
It is not possible to have additional money for travel than the budget planned but it could be possible to increase this budget if some money is put aside from some other activities ➔ possible solutions need to be discussed with financial officers of the institutions: shift of personnel to travel budget compensated by increase of in kind contribution for example.
Shift of budget from one partner to another:
In the budget, it is possible to shift:

- personnel costs from one year to another (has to be underlined every year in the AWP);
- budget from one partner to another partner but ATTENTION:
  - these changes needs to be reported to the coordination in advance
  - have to be sure that both partners are ok with the change
  - be careful, cost of person/month (PM) are different from one partner to another. What is shifted is the total cost in euros and not the number of PM (no increase of budget is possible even if the total costs of PMs is higher).

Actions:

- ⇨ distribution of PM and other costs for the AWP Year 2 have to be defined by the end of August
- ⇨ budget file per partner will be sent by Nadia Ibellaatti (administration@ejprarediseases.org)

Financial reporting:
The financial reporting will take place every end of year (Report at 12 months) as well as a scientific report➔ have to be delivered by the end of February 2020 for the first year.
A training (in the form of webinar or eventually a F2F meeting) for financial officers will be set up by Nadia: will probably take place in September. A guidebook will also be developed to help with the reporting.

Use of budget initially set aside for FAIRification activities:
A FAIRification envelope budget is in the Central budget of the coordination as its use have not been defined yet. It was proposed to use it in the context of the ERN call as small liaison budget to allow the visit of experts in the EJP RD ➔ need to have a description of how this budget will be used to be decided/validated in the GA in September.

Use of sponsors/additional funding for the EJP RD activities:
Any additional funding in the EJP RD is considered as income and means that the EC budget will be reduced of that amount. In case there is an under-estimation of the cost of a task this will have to be demonstrated and the impact of the private funding on the activity will have to be described. In any case the EJP RD should not make any profit, so the whole budget (EJP RD one + sponsored) must at the end be equal to the total costs of the activity (no extra income that could be used for other activity is allowed).

Sustainability of the EJP RD activities
See slides 88-89 for complete information

A whole session will be dedicated to sustainability during the GA meeting on 17-19 of September:

- ⇨ the models of sustainability should be shared in advance of the meeting by WP3
- ⇨ A handbook providing guidelines to help the partners to get in the process of sustainability will be prepared and also shared.

ERNs & EJP RD: common agenda
See slides 90-111 for complete information

A survey has been sent to the 950 ERN units from the EJP RD to have a clear idea of their needs. We received answers from 1/3.
In the ERN governance, a Working group on Research has been established: rotating chair (change every year) – the current chair is Alberto Perreira. Alberto Perreira will be invited to the EJP RD Operating Group. In addition, the chair of ERN coordinators - Franz Schaeffer already participates in the OG (as Pillar 2 co-leader) and provides additional liaison between ERNs and the EJP RD

- ⇨ it will be important to communicate and disseminate the existing tools and provide more information on the EU research infrastructures to ERNs as the survey showed that the awareness on them is very low when the need for what they provide is high.

Several interactions are already existing between EJP RD and ERNs:

- WP12: registry collaboration → interaction between the Work Focus Team 11 from Pillar 2 and Registry Taskforce of ERNs
- WP13: Cross-omics collaboration → interaction between WP13 and people who answered the survey and agreed to be re-contacted: a direct collaboration with the researchers can start.
- WP17: ERN Research Training Programme → WP dedicated to develop research training programs for ERNs. A clear interest exists from the ERNs to have trainings on scientific tools and methodologies and on concrete research skills with a format of training workshops/seminar and physical visit of junior researchers. Proposition to established 1/ Focused training seminars/workshops for ERN researchers (up to 45 workshops for a total budget o 1Million €) and 2/ short-term exchange visit program for ERN researchers (total budget of 0.6 Million €) → program start is anticipated for Q4 2019
- WP20: Clinical study support office and Demonstrator projects.

The WP14 provides trainings responding to the identified needs of the ERNs but for the first trainings organized there were not a high participation from ERNs: participation is free of charge but participants should fund their own travel fees.

Action:

- ⇨ A direct communication could be done to the ERNs units and not only ERN coordinators: this have to be done through the EC communication officer that has been delegated to the ERNs
- ⇨ Part of the budget of WP17 could be used to fulfil needs of ERNs for travel/accommodation without creating overlap between trainings.

EJP RD Policy Board meeting

4th of July 2019

9:00 – 17:30

Albert Borschette building, rue Froissart, Brussels, room AB-1.A

Attached document:
Slides presented during the meeting – see Annex 2 (p135)

List of participants:

Name	Country	Board	Presence
Ardigo Diego	Italy	Policy Board	Present
Allon Irit	Israel	Executive Committee	Present
Ayvazyan Naira	Armenia	Policy Board	Present
Azzopardi Miriam	Malta	Policy Board	Present
Barisic Ingeborg	Croatia	Policy Board	Present
Beltran Sergi	Spain	Executive Committee	Present
Berrens Catherine	EC DG RTD	Policy Board	Present
Bonnaud Emilie	France	Executive Committee	Present
Bottarelli Valentina	EURORDIS	Policy Board	Present
Beltrami Elena	Italy	Executive Committee	Present
Berger Tanja	Germany	Executive Committee	Present
Bermejo-Sanchez Eva	Spain	Executive Committee	Present
Brookes Anthony	UK	Executive Committee	Excused
Bros-Facer Virginie	France	Executive Committee	Present
Carta Claudio	Italy	Executive Committee	Present
Chrzanowska Krystyna	Poland	Executive Committee	Present
Denisovienė Dainė	Lithuania	Policy Board	Present
Dolezalova Pavla	Czech Republic	Policy Board	Present
Nabbout Rima	France	Executive Committee	Present
Evelo Chris	Netherlands	Executive Committee	Present

Favresse Roseline	France	Executive Committee	Present
Fetro Christine	France	Executive Committee	Present
Foltanova Tatiana	Slovak Republic	Policy Board	Present
Gamst Nora	Norway	Policy Board	Present
Giannuzzi Viviana (TC)	Italy	Executive Committee	Present
Graessner Holm	Germany	Executive Committee	Excused
Grimm Bernard	EUROPABIO	Policy Board	Present
Guglielmi Gaetano	Italy	Policy Board	Present
Halftermeyer Juliane	France	Executive Committee	Present
Hilgers Ralf-Dieter	Germany	Executive Committee	Present
Ikonen Tuija	Finland	Policy Board	Present
Israeli Abraham	Israel	Policy Board	Present
Julkowska Daria	France	Executive Committee	Present
Klosakova Judita	Czech Republic	Policy Board	Present
Khmaladze Ekaterine	Georgia	Policy Board	Present
Kolyva Sosanna	Greece	Policy Board	Present
Landi Annalisa	Italy	Executive Committee	Present
Le Borgne Hélène	EC DG Santé	Policy Board	Present
Martin Simona	EC JRC	Invited	Present
Monaco Lucia	IRDiRC	Policy Board	Present
Nguyen Catherine	France	Executive Committee	Present
Nicholl Ciaran	EC JRC	Invited	Excused
Paluste Heli	Estonia	Policy Board	Excused
Passeri Eleonora	France	Executive Committee	Present
Preuss Monika	UK	Policy Board	Present
Rath Ana	France	Executive Committee	Present
Reichel Judith	Germany	Policy Board	Present
Roos Marco	Netherlands	Executive Committee	Present
Rzepecki Ryszard	Poland	Policy Board	Excused
Sahin Jale (TC)	Turkey	Policy Board	Present
Sangiorgi Luca	Italy	Executive Committee	Excused
Scarpa Maurizio	Italy	Executive Committee	Present

Schuster Ralph	Germany	Executive Committee	Present
Schaefer Franz	Germany	Executive Committee	Present
Schwartz Bertrand	France	Policy Board	Present
Simelyte Egle	EC DG RTD	Project Officer	Present
Taruscio Domenica	Italy	Executive Committee	Present
Tumiene Birute	Lithuania	Executive Committee	Present
Ussi Anton	Netherlands	Executive Committee	Present
Valverde Jose A.	EC DG CNECT	Policy Board	Present
van Weely Sonja	Netherlands	Executive Committee	Present
Weinbach Jérôme	France	Policy Board	Present
Wessel Theda	Germany	Policy Board	Present
Wijnhoud Maaike	Netherlands	Policy Board	Present

Agenda:

9:00 – 9:30	Welcome word and introduction to the EJP RD	Daria Julkowska (Inserm) Coo
9:30 – 10:00	Round table	All
10:00 – 10:30	Pillar 0 – Coordination and transversal activities : *Presentation of achievements (M1 – M7) and of activities planned in the Annual Work Plan Year 2 with Policy Board [25min] + immediate questions [5min]*	Daria Julkowska (Inserm)
10:30 – 10:50	**Coffee break**	
10:50 – 11:20	Pillar 1 – Collaborative research funding : *Presentation of achievements (M1 – M7) and of activities planned in the Annual Work Plan Year 2 with Policy Board [25min] + immediate questions [5min]*	Ralph Schuster (DLR) and Sonja van Weely (ZonMw)
11:20 – 11:50	Pillar 2 – Innovative coordinated access to data and services for transformative rare diseases research : *Presentation of achievements (M1 – M7) and of activities planned in the Annual Work Plan Year 2 with Policy Board [25min] + immediate questions [5min]*	Ana Rath (Inserm) and Franz Schaefer (UKL-HD)
11:50 – 12:20	Pillar 3 – Capacity building and empowerment : *Presentation of achievements (M1 – M7) and of activities planned in the Annual Work Plan Year 2 with Policy Board [25min] + immediate questions [5min]*	Virginie Bros-Facer (EURORDIS) and Biruté Tumiene (VUHSK)
12:20 – 13:20	**LUNCH**	
13:20 – 13:50	Pillar 4 – Accelerating the translation of high potential projects and improving outcomes of clinical studies in small populations : *Presentation of achievements (M1 – M7) and of activities planned in the Annual Work Plan Year 2 with Policy Board [25min] + immediate questions [5min]*	Rima Nabbout (APHP) and Anton Ussi (EATRIS)
13:50 – 14:50	Open discussion of activities planned in the Annual Work Plan Year 2 with Policy Board	All
14:50 – 15:10	**Coffee break**	

15:10 – 16:10	Prioritization strategy process (indicators and methodology) – *Presentation and discussion with PB*	Eva Bermejo and Manuel Posada (ISCIII)
16:10 – 17:00	Stakeholders needs and challenges	Policy Board members round table
17:00 – 17:20	EJP RD under Horizon Europe	All
17:20 – 17:30	Summary & Next steps	Daria Julkowska (Inserm) Coo

Minutes:

Presentation of achievements (M1 – M7) and of activities planned in the Annual Work Plan Year 2:

- **Pillar 0 – Coordination and transversal activities, Daria Julkowska, EJP RD coordinator**
- **Pillar 1 – Collaborative research funding, Ralph Schuster & Sonja van Weely, Pillar 1 chairs**
- **Pillar 2 – Innovative coordinated access to data and services for transformative rare diseases research, Ana Rath & Franz Schaefer, Pillar 2 chairs**
- **Pillar 3 – Capacity building and empowerment, Virginie Bros-Facer & Biruté Tumiene, Pillar 3 chairs**
- **Pillar 4 – Accelerating the translation of high potential projects and improving outcomes of clinical studies in small populations, Rima Nabbout & Anton Ussi, Pillar 4 chairs.**

For complete information please see the slides (annex). There were no immediate comments or questions after the presentations.

Open discussion of activities planned in the Annual Work Plan Year 2 with Policy Board / Stakeholders needs and challenges

Comments from the Policy Board members (in order of speaking):

UK: there is a great interest for the Pillar 2 activities of the EJP RD and federation of data as it goes along with other important UK actions, for example their engagement in the 1+ Million Genomes project and support of genomics research. Since the registries are crucial for rare diseases there is a clear need for their connectivity and interoperability; thus efforts should be made in this direction but taking into account what exists and exploiting it.

Armenia: since Armenia is currently present in the EJP RD through its linked third party they would be interested to participate more but a discussion with national stakeholders is needed. It is also not yet clear how the EJP RD activities may overlap with some other EU projects and possible co-funding of the same activities of the involved national teams.

Georgia: the EJP RD is very complex and for the newcomers may be difficult to follow. It would be good to have some document/summary for the beginners, some indications on where to start, the main message that should be distributed to the national stakeholders.
Finland: they have just updated their national plan for RDs, they also have some ERN nodes but not sufficiently of research in the field so some strategic planning would be needed (especially for health sciences and genomic centers). The EJP RD can provide some new options for collaboration with other Nordic countries. Finland has a dedicated law for the secondary use of health and social data, which can be helpful in relation to the work done in Pillar 2. Moreover, the Finland developed regional and national registries to which FAIR criteria should apply.

Lithuania: is in line with the position of Georgia for the development of dedicated support documents (even in national language) for target groups to inform about the EJP RD, its goals and what is expected as outcome after 5 years of the programme so the next steps could be already planned.

Norway: has some limited participation in Pillar 2 but could engage more and surely would profit from the outputs. Currently the RD strategy is under development in Norway so the EJP RD (and participation in it) should be helpful. It would be good to receive some instructions on the National Mirror Groups (how to compose them, what is expected from them, etc.)

Slovakia: the data infrastructures are of great interest for the country. It is also important that the eHealth networks are connected and use the same standards. It would be expected that some innovative ways for ICT will be also applied/proposed.

Malta: is currently in process of upgrading its database and would like to receive some support so it is done according to standards. In addition they are in process of the development of RD strategy, as well as expansion through the ERNs. The connection with the EJP RD should help in better shaping of the strategy.

European Commission DG RTD: the Commission is pleased to see the advancement of this joint effort despite its complexity. However, the international efforts should not be forgotten and thus the connection to IRDiRC is very important.

European Commission JRC: the JRC is willing to help in the strategic orientation of the EJP RD and its alignment with national and EU efforts. Its participation in the Pillar 2 activities is part of that support.

European Commission DG Santé: the directorate is happy to help in the alignment with national strategies, synergies with Orphanet, ERNs and RD policy.

Eurordis: supported the project from the start and is please to see that patients are involved in different type of activities. The fact that Policy Board is multi-stakeholder is

of great importance as a body of such character and impact was missing since several years.

IRDiRC: the progress of the EJP RD is great and there is no other programme if equal size and depth worldwide. That is why it is also very challenging and the dissemination of clear message is key. IRDiRC also produced some resources, guidelines and tools that are not always used or known, which is a loss for community. The integration of the Scientific Secretariat of IRDiRC in the EJP RD coordination was a very good and strategic idea as it allows better alignment and already resulted in common planning of some joint activities. It is also important to underline that the new SciSec team that took over only in January 2019 was able to grasp and run the secretariat smoothly in only few months.

EuropaBIO: data and registries are key priority also for industry but there is a need to focus first on the "low hanging fruits" first like the quality of data/registries. The complexity and fragmentation of RD research in Europe remains as a problem. The activities proposed in the EJP RD Pillar 4 – mentoring of projects and support for translation – are very good and will be useful. Some examples from the US (Boston) informal mentoring communities could be used.
The best way to sustain the project is to demonstrate that all milestones are achieved on time and to showcase success stories with ERNs.

Czech Republic: it is key to not to duplicate the activities thus sharing of the information and disease specific activities are very important. In addition, the education of students, researchers and young doctors is crucial as it also contributes in better/improved healthcare.

Germany: supports RD research since many years and thus naturally joined EJP RD. German partners are involved in the programme at many levels and the ministries are interested to see what will be the outcomes of the programme after 1 year of existence.

European Commission DG CNECT: the 1+ Million Genomes initiative has many aspects in common with the EJP RD and both projects should bring valuable outcomes. Data is key but all the problems are far from being solved so we must continue.
The creation of National Mirror Groups is very interesting for the strategy at national level as it can bring research and healthcare together.

Israel: the healthcare systems are very different in all countries and thus should not be the main goal of the EJP RD. Similarly the ethical aspects and cultural differences may be a real challenge. The training and data issues are very important. The EJP RD should demonstrate through the achievement of respective milestones that it is on a good track and expected impact is there.

Greece: the NMGs are very important also for Greece. The activities of Pillar 4 are very innovative but we should not forget to have a good synergy with the EU and national

infrastructures; same for Pillar 2 in relation to data and connection to Elixir infrastructure. The Policy Board could be of great help for the sustainability issue but it is necessary to identify what would be the tasks in which PB can get involved.

France: the challenge we face is to bring stakeholders from different horizons but for the moment the efforts are going in good direction. The EJP RD is a future model for Partnerships under Horizon Europe but its sustainability is a key issue. Although specific Partnership on rare diseases is already mentioned in the preparatory documents for Horizon Europe some questions related to administrative and financial issues remain open.
RD research is very much driven by patients needs and also helped already a lot in understanding other more common phenomena. Thus, the pathways from RD data to other diseases or research areas (systems biology) are very important.
The interaction with 1+ MG is of added value and EJP RD will gain on visibility through it but the joint effort should help in better tools for annotated data from healthcare.
Now that the consortium and pillars / WPs' working groups are in place, the activities should demonstrate good interactions between the different pillars, which is not yet so visible, for example the projects funded under Pillar 1 should be assisted for translation of their results or their data integration in Pillar 2 Virtual Platform. The progress should be measured with clear impact indicators, baseline and target values that will be useful for convincing policy makers to support EJP RD sustainability.
As for the future of the EJP RD, it is needed to also take into account the health policy perspective, like disease prevention and health promotion. Thus, a win-win situation should be installed within the EJP RD – good balance between research and translation to healthcare. EJP RD should be a platform for policy makers and EMA regulators to support for instance the HTA evidence (e.g organizing joint real life drug impact studies between MS). Similarly, the issue of equal access to treatments for all patients in all Member States is of relevance as well as public-private collaborations.
Finally, a good sustainability strategy will also consist in designing and swarming new projects at regional, national and European levels connected to EJP RD. Therefore, it is crucial to connect the EJP RD and mutually inform about the opportunities at EU and national level to progressively build a network of connected/supporting projects.

Croatia: it is important to have a strong engagement of ERNs and good connection with the EMA. The NMGs are of relevance and can help in dissemination at national level. It is very important to connect well the local data resources, training opportunities and efforts that will be provided under 1+ MG project.

Italy: the EJP RD is a new model of collaboration and thus it is essential that it demonstrates first that such model can work. The dissemination should be based on set of simple messages targeting specific stakeholders.

Turkey: currently in Turkey a dedicated group of parliament members was established to work on the subject of RD research and regulation of access to treatments. However, Turkey does not have a dedicated RD national plan. EJP RD is a good platform to complement national efforts and increase the capacity of Turkish

researchers but also underline the potential of Turkey (especially the number of patients). The NMG will be of help and can be the driver for the creation of the plan for RDs.
The activities of Pillar 3 are a very important tool but it would be good to increase the capacity per training (allow higher number of participants) especially for the associated countries. If possible, it would be nice to have some courses delivered at national level. In fact, due to the complexity of the EJP RD the support from national stakeholders is crucial as they can help in expanding the capacity of the programme and its dissemination.

EUCOPE: the EJP RD is an interesting project brining together new stakeholders and possible boost for innovation. The activities of Pillar 3 & 4 should help in increasing of the capacity of drug development and delivery. Thus, the support from industry is key and should be used for both mentoring but also capacity building.

Summary of discussion and recommendations:

1. Policy board members highlighted the promising progresses accomplished by the EJP RD in the first 6 months given the high complexity of the project. This is a huge and complex project but it has no comparison worldwide in the rare diseases field. It puts together different topics and bring the results to the patients.

2. The Policy Board can be key players to pass the right messages to ensure that rare diseases (RD) are high on the European Commission (EC) agenda in particular connecting with the European Council. Involvement in the future programme in health could be also important to all go on the same direction.

3. The Policy Board members are a way for the EJP RD to get engage with new national funding agencies that could participate in the future EJP RD Joint Transnational Calls (JTCs). PB members should be ambassadors of the EJP RD in their countries.

4. PB members could be also more involved in concrete tasks such as sustainability: members could accompany the project on this question towards Horizon Europe, national and European strategies.

5. EJP RD could also be a central project helping the development of satellite projects involving EJP RD partners to develop its power of action.

6. Pillar 2 is incredibly complex but critically important. The connection of registries is highly important for the research community but also for policy makers as they are reassured of the added value of the already invested money.

7. The creation of National Mirror Groups (NMGs) will allow the link between heath and research sectors and will also allow countries to develop national strategies. ➔ EJP RD coordination will prepare and share a framework to create and organize NMGs in all EJP RD countries based on the example of the French NMG: it involves different types

of stakeholders (ministries of Health and Research, representative of EJP RD partners, representatives of patients, of research and health care community, ERNs and French equivalent of ERNs) and allows to have a feedback from the national community: important to make sure that what is happening at national level is taken into account in the EJP RD and the other way around. The NMGs should be a place of dialog between stakeholders. Especially in small countries, there is a high need to have a connection between different ecosystems. Orphanet local teams should also be involved in the NMGs.

8. Patient involvement is well embedded in the project. It is important to have an ecosystem of various stakeholders in the project and in the Policy Board.
Research in RD is pushed by patient needs and has always been a strong driver to understand cell biology, physiology, etc. this should continue and it is still important to sustain RD research. The way now taken through data and pathways is of major importance in this field.

Recommendation 1: To better demonstrate the added value of the project, some KPIs are needed as well as impact indicators: important to show what was the baseline and what has been achieved and to show it through measurable indicators.

Recommendation 2: It is important to highlight in the work plan of the EJP RD (achievements and planned work) the connection between the pillars.
Some practical examples are needed on the user side, in particular for Pillar 2: a work force has been set up on these questions. It is necessary to show that the project is answering the needs of patients and research community.

Recommendation 3: The EJP RD should continue to look for synergies with other existing programs not to develop things already developed somewhere else. For example, common things could be learned with the 1+Million Genome initiative: it is good that many common representatives are present in both projects.

Recommendation 4: The EJP RD should help countries and especially the countries that are new in the field to start to move forward; provide useful tips on where to start and provide PB members simple take home messages. PB members underlined that in order to organize the RD community in countries, it is very important to educate researchers and health care professionals in sub-specialties. To provide good quality data, people have to be qualified. ➔ EJP RD members reminded that the registration to EJP RD trainings is available on the EJP RD website and communication on the trainings will be done through the large networks of partners (that are part of the EJP RD like Orphanet, ERNs, Eurordis, etc.) to the community.

Recommendation 5: In general, a more simple communication on the program should be prepared to communicate more easily with persons not directly involved in the project activities with specific messages depending on the target groups (i.e. patients, charities, researchers, doctors, etc.) highlighting the goals of the project. A big effort should be done in the dissemination of the message.

Recommendation 6: Additional milestones should be added in Pillar 2 to measure the achievements and to be able to communicate to other stakeholders (ERNs, etc.) on those achievements. Some countries would also like support from the EJP RD side to upgrade their databases and help them to collect data in a standardized way.
At the same time Policy Board members are invited to encourage the national support for data collection especially through ERN registries as this is the basis of the success of Pillar 2 Virtual Platform.
Data and registries are also in the top priority for biotechnology industry. One important first step for this work is to measure the quality of registry/data.

Recommendation 7: The mentoring activity presented in Pillar 4 is a great idea that should be more developed in Europe, on the model of what already exists for example in Boston or in California. For example the set-up of unformal community of expertise could be expanded a bit further.
Even so the EJP RD project is oriented toward research, its activity will increase capacity, skills, knowledge and support research in the long journey of drug development. Some industries are interested to be part of this process and are ready to support researchers. A close interaction with EMA should be developed (planned in particular in Pilar 4).

Recommendation 8: The establishment of synergies between EJP RD and ERNs should be continued.

Recommendation 9: In general, it is important that all stakeholders spell out their needs so that the project can evolve fitting at best those expressed needs from the users.

Prioritization strategy process (indicators and methodology)
See attached slides for complete information

Since the EJP RD partners were exhaustive in the description of the planned work for the 5 years it was underlined that at this stage only a few elements would need prioritization and eventual choice between several options. However, the EJP RD Executive Committee would like to use the prioritization methodology/decision support tool to help the decision process in a task/sub-task of the project. Thus, only the points of major importance would be discussed with the Policy Board and submitted to their advice (rather than mathematical prioritization process).

Recommendations from the Policy Board:
Despite the fact that the prioritization process will take place at the consortium level and as a guiding process the four categories of indicators that have been proposed are not sufficient and there should be more room for measurement of the impact of an action and for the urgency of the proposed activity. Criteria should also address critical unmet needs in RD and impact of research on identified ecosystems' gaps.

Each criterion should be accompany by a range of descriptive parameters and descriptors coming with the score. This would allow a clear cut justification of the scores.
The proposed approach presented is very theoretical and should be more pragmatic: it would be important to take into account in the decision process also the policy makers needs.

- ⇨ For taking decision, a consensus should be reached, not only using a mathematical process. This process should be used as a starting point to sort options in an unbiased way but should be followed by a discussion to take decisions. As an internal tool, the decision process can be self-referential. In case external opinion is needed, a committee involving different stakeholders would have to be build.
- ⇨ The PB recommends to make some simulation on the use of those indicators before using them for taking decisions in the project: check in particular if using the same weight for all criteria is the good balance to use. For this the consortium could go back on what has been done in Year 1 and see what could be the outcome using the proposed model.

The EC underlined that the Annual Work Plans implemented in the EJP instrument are done on purpose to adapt priorities in the project. Thus, even if initially the prioritization process was intended to include the Policy Board and now is revised as a guiding process for the EJP RD consortium members, the Commission is prepared to discuss this modification and accept if the deviation is justified.

EJP RD under Horizon Europe

The Horizon Europe programme is still under negotiation. Currently, a strategic planning exercise is done involving all relevant stakeholders to prepare the first work programme (should be ready at the end of 2020).
A public consultation on general orientation of Horizon Europe has been launched ➔ you are all invited to participate in it:
https://ec.europa.eu/eusurvey/runner/HorizonEurope_Codesign_2021-2024
The results are expected early September.
On the 26th of September, during the EU Research & Innovation Days a conference is organized to discussion future orientations and a session dedicated to RD is planned.
In the orientation document prepared by the EC in collaboration with the Member States, some place have been kept for co-funded partnership in RD At present it is very broad, not to preempt what will happen in the next years and what will be the needs in a few years. It is also important to underline that Horizon Europe is prepared in collaboration with other (than DG RTD) DGs as RDs have relevant activities in other programmes.

Next steps:

- Finalise the constitution of Policy Board
- Finalise the creation of National Mirror Groups

- All PB members will receive the final version of the Annual Work Plan that should be approved by the EC in October
- All PB members will be regularly informed about the EJP RD achievements
- The coordination will liaise with the respective PB members and NMG to provide dedicated content or participate in meetings if requested
- Next regular Policy Board meeting: **8 July 2020**
- Possible additional PB meeting – Dec 2019/Jan 2020 to celebrate 1 year of the EJP RD

Additional information: On September 16th 2019, a satellite meeting to the Consortium General Assembly of the EJP RD will take place to discuss the needs and ways to engage EU13 countries in EJP RD activities. The meeting will take place at the Medical University of Gdansk, Gdansk, Poland. The PB members are welcome to participate and to disseminate the information at national level to relevant stakeholders.

Annex 1

Slides presented during the EJP RD Executive Committee

European Joint Programme on Rare Diseases

Executive Committee meeting

3rd of July

Brussels, Belgium

EUROPEAN JOINT PROGRAMME RARE DISEASES

Objectives of the meeting

- Pre-identify the performance and results KPIs and agree on the procedure to measure them
- Discuss & agree on the prioritization strategy process – what will need prioritization?
- Discuss the needs for communication and its implementation rules (coo + partners)
- Discuss the interaction with IRDiRC
- Discuss and agree on the strategy for interaction with other important stakeholders/project (1+MG, GA4GH, C4C, etc.)
- Agree on how to work with non-EJP RD partners (perimeter, process?)
- Present & agree on the implementation of the Helpdesk and association with different Pillars' services
- Discuss financial questions from WPs (budget & PMs allocation)
- Sustainability of the EJP RD activities – inputs from Pillars
- Discuss the EJP RD & ERNs common agenda

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Task 1.3.1: defining KPIs

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RARE DISEASES

Metrics are important

- To measure what we have done (Results)
  - Timing
  - Quality
  - Integrating many groups and teams
- To measure how we are working (Performance)
  - Effective working is needed for good results
  - Working with single teams, allow improvements in real time

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Key metrics (a selection of metrics)

- Reflect activities that are essential for the whole organization
- Key Results Indicators – what outputs are mission-critical?
- Key Performance Indicators – which behaviours does everybody need to show?
- KPIs need to be selective
  - < 10

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Goals of the process to set metrics

1. Define performance metrics
2. Define data collection methods
3. Start data collection
4. Review progress using metrics

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Proposed process 1

Goal	Task	Owner	Timeline	Other input needed
Define metrics	WP propose metrics to Pillar leads	WP leads	July 15th	
	Pillar leads validate WP metrics and identify pillar metrics and KPIs	Pillar leads	July 31st	
	Exec ratifies Pillar metrics and identifies any EJP KPIs	ExecCo	???	
Define data collection	Draft principles and likely procedures	CVBF	July 15th	
	Revise principles and likely procedures	CVBF	July 31st	
	Finalise procedures	CVBF	After ExecCo ratifies Pillar and WP metrics	

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Define performance metrics

1. Why?
2. What?
3. How?
4. Who?
5. When?
6. Consequences

What difference will this metric make?

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Proposed process 1 (discussion points in red)

Goal	Task	Owner	Timeline	Other input needed
Define metrics	WP propose metrics to Pillar leads	WP leads	July 15th	
	Pillar leads validate WP metrics and identify pillar metrics and KPIs	Pillar leads	July 31st	
	Exec ratifies Pillar metrics and identifies any EJP KPIs	ExecCo	???	
Define data collection	Draft principles and likely procedures	CVBF	July 15th	
	Revise principles and likely procedures	CVBF	July 31st	
	Finalise procedures	CVBF	After ExecCo ratifies Pillar and WP metrics	

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Define data collection

1. What? Nature of data
2. How? Source: Automated or Manual
3. Who? Name
4. When? Duties
5. Handling? Destination / Processing / Querying

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Proposed process 2 (discussion points in red)

Goal	Task	Owner	Timeline
Start data collection	Implement procedures	To be defined in final version of description of procedures	??
Review metrics	Review pillar metrics	Pillar leads	To be defined by Pillar
	Review EJP JP metrics and selected pillar metrics	ExecCo	December 2019 (or sooner if needed)

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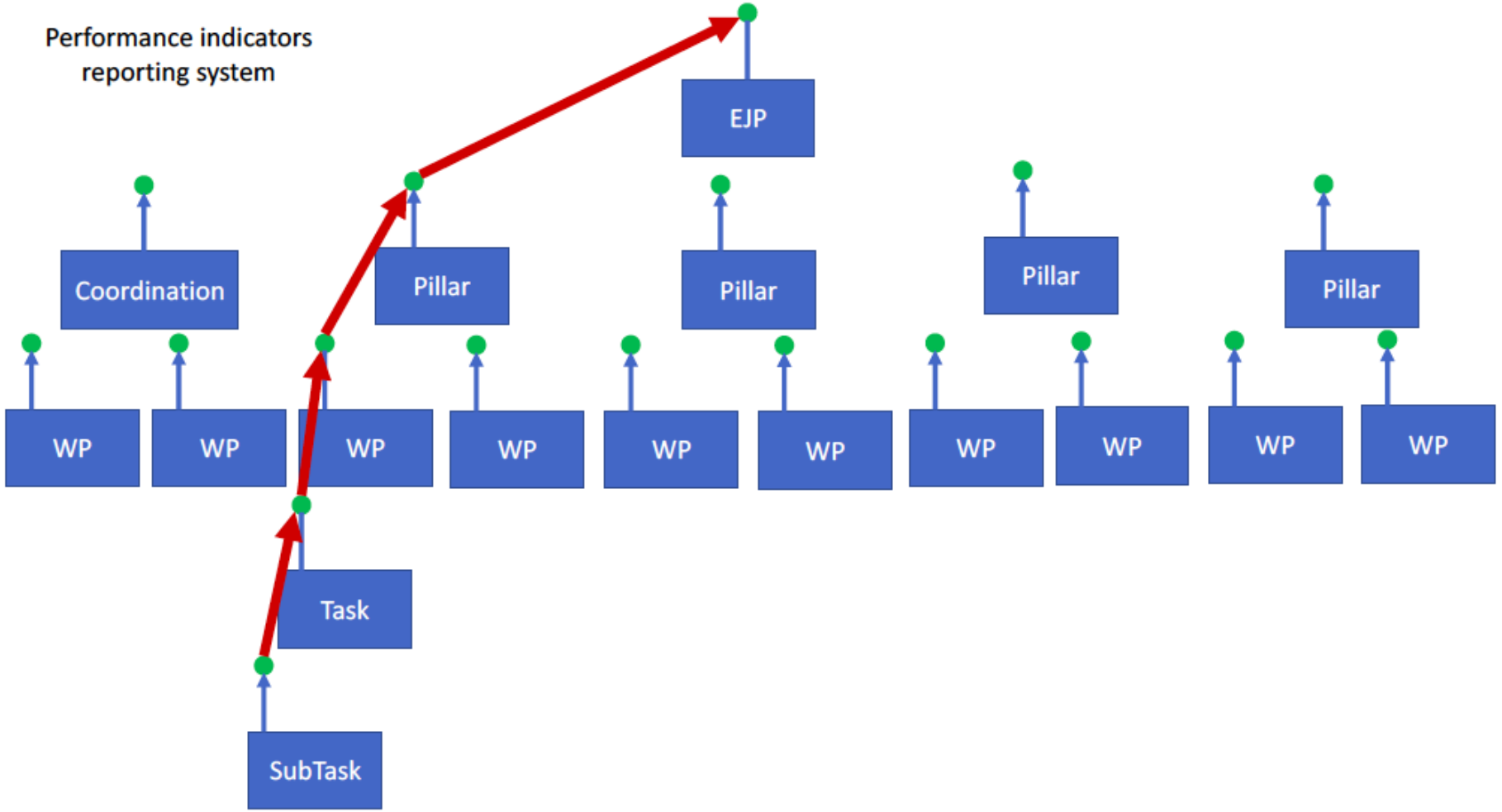
Discussion points

- When to escalate?



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Next steps

- Exec Committee to validate process
- WP / Pillars to select metrics
  - Including when to escalate
- ExecCo to ratify metrics
- Start data collection
- Start using metrics

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Prioritisation strategy process

Selection of elements to prioritize

Eva Bermejo-Sanchez & Manuel Posada (ISCIII)

Executive Committee Meeting

Brussels, July 3, 2019

EUROPEAN JOINT PROGRAMME RARE DISEASES

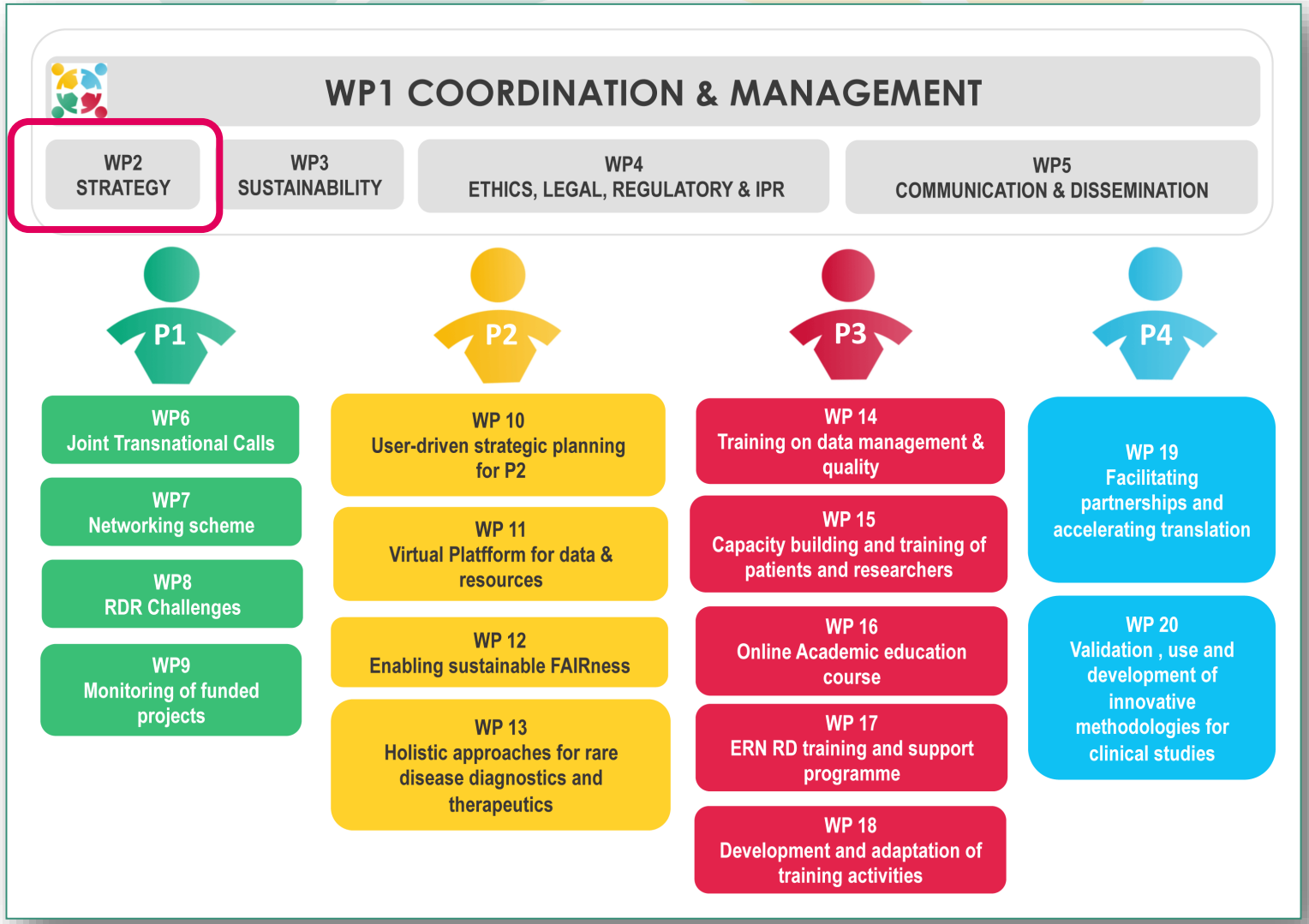
WP2
Integrative Research and Innovation Strategy

WPLs: ISCIII & ISS

Eva Bermejo-Sánchez and Domenica Taruscio

EUROPEAN JOINT PROGRAMME RARE DISEASES

Work package 2:
Integrative Research and Innovation Strategy



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OBJECTIVES

Work package 2 overall objectives

Goal: This WP focuses on the development of EJP RD research and innovation strategy in connection to all related stakeholders.

Specific objectives:

- To **map the needs** for RD research and innovation;
- To **define the prioritization model for the EJP RD actions** as part of the annual planning and in connection to WP3 (Sustainability) and WP4 (Ethical, Regulatory, Legal and IPR framework);
- To **prioritize topics for the joint transnational calls (JTCs)** to be implemented within Pillar 1;
- To **feed the medium and long-term RD research and innovation strategy** in collaboration with IRDiRC;
- To **prepare a Scoping Paper** to be promoted (and implemented) by the national mirror groups and within the current and forthcoming EC Framework Programme.

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Work package 2 year 1 objectives

- To **map the research and innovation needs**
- To **define the prioritization model for the EJP RD actions**;
- To **prioritize topics for the joint transnational calls (JTCs)** to be implemented within Pillar 1;
- To **build a list of R&I needs requiring a medium and long-term approach** and **related Task Forces**
- To **prepare a Scoping Paper** to be promoted (and implemented) by the national mirror groups and within the current and forthcoming EC Framework Programme.

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Prioritisation strategy process

Prepared by CVBF

Angelica Intini (AI) & Giovanni Migliaccio (GM)

EUROPEAN JOINT PROGRAMME RARE DISEASES

Prioritisation strategy process

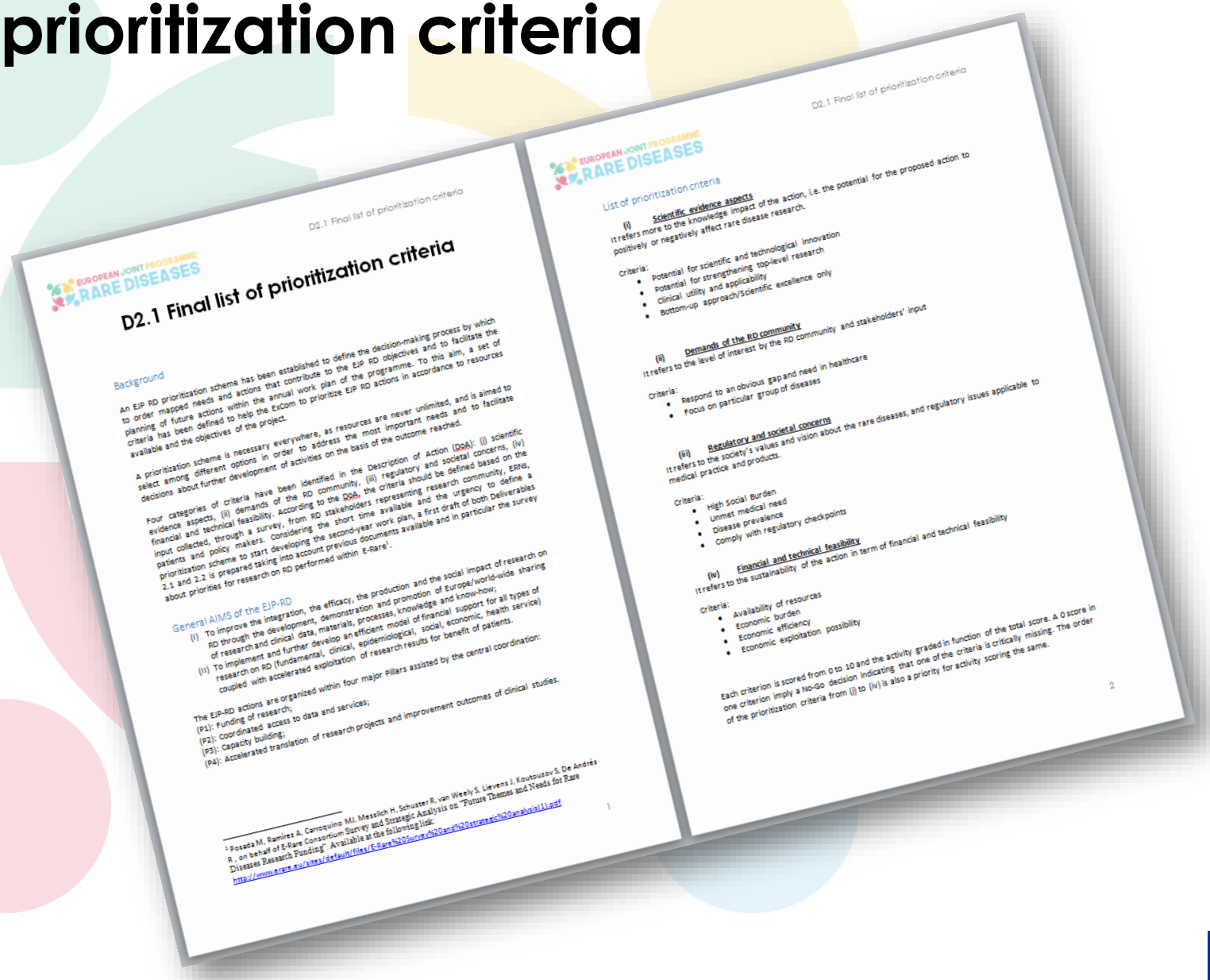
Angelica Intini (AI) & Giovanni Migliaccio (GM)(CVBF)

[presentation by Eva Bermejo-Sanchez &
Manuel Posada (ISCIII), on behalf of AI &GM]

Executive Committee Meeting – Conference Call
May 27, 2019

EUROPEAN JOINT PROGRAMME
RARE DISEASES

D2.1 – Final list of prioritization criteria



D2.1 Final list of prioritization criteria

EUROPEAN JOINT PROGRAMME RARE DISEASES

D2.1 Final list of prioritization criteria

Background

An EJP RD prioritization scheme has been established to define the decision-making process by which to order mapped needs and actions that contribute to the EJP RD objectives and to facilitate the planning of future actions within the annual work plan of the programme. To this aim, a set of criteria has been defined to help the ExCom to prioritize EJP RD actions in accordance to resources available and the objectives of the project.

A prioritization scheme is necessary everywhere, as resources are never unlimited, and is aimed to select among different options in order to address the most important needs and to facilitate decisions about further development of activities on the basis of the outcome reached.

Four categories of criteria have been identified in the Description of Action (DoA): (i) scientific evidence aspects, (ii) demands of the RD community, (iii) regulatory and societal concerns, (iv) financial and technical feasibility. According to the DoA, the criteria should be defined based on the input collected, through a survey, from RD stakeholders representing research community, ERNs, patients and policy makers. Considering the short time available and the urgency to define a prioritization scheme to start developing the second-year work plan, a first draft of both Deliverables 2.1 and 2.2 is prepared taking into account previous documents available and in particular the survey about priorities for research on RD performed within E-Rare[1].

General AIMS of the EJP-RD

(I) To improve the integration, the efficacy, the production and the social impact of research on RD through the development, demonstration and promotion of Europe/world-wide sharing of research and clinical data, materials, processes, knowledge and know-how;
(II) To implement and further develop an efficient model of financial support for all types of research on RD (fundamental, clinical, epidemiological, social, economic, health service) coupled with accelerated exploitation of research results for benefit of patients.

The EJP-RD actions are organized within four major pillars assisted by the central coordination:
(P1): Funding of research;
(P2): Coordinated access to data and services;
(P3): Capacity building;
(P4): Accelerated translation of research projects and improvement outcomes of clinical studies.

[1] Posada M, Ramírez A, Carroquino MJ, Messlich H, Schuster R, van Weely S, Lievens J, Koutsouov S, De Andrés R., on behalf of E-Rare Consortium Survey and Strategic Analysis on "Future Themes and Needs for Rare Diseases Research Funding". Available at the following link: http://www.erare.eu/sites/default/files/E-Rare%20Survey%20and%20strategic%20analysis41.pdf

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D2.1 Final list of prioritization criteria

EUROPEAN JOINT PROGRAMME RARE DISEASES

List of prioritization criteria

(i) **Scientific evidence aspects**
It refers more to the knowledge impact of the action, i.e. the potential for the proposed action to positively or negatively affect rare disease research.

Criteria:
- Potential for scientific and technological innovation
- Potential for strengthening top-level research
- Clinical utility and applicability
- Bottom-up approach/Scientific excellence only

(ii) **Demands of the RD community**
It refers to the level of interest by the RD community and stakeholders' input

Criteria:
- Respond to an obvious gap and need in healthcare
- Focus on particular group of diseases

(iii) **Regulatory and societal concerns**
It refers to the society's values and vision about the rare diseases, and regulatory issues applicable to medical practice and products.

Criteria:
- High Social Burden
- Unmet medical need
- Disease prevalence
- Comply with regulatory checkpoints

(iv) **Financial and technical feasibility**
It refers to the sustainability of the action in term of financial and technical feasibility

Criteria:
- Availability of resources
- Economic burden
- Economic efficiency
- Economic exploitation possibility

Each criterion is scored from 0 to 10 and the activity graded in function of the total score. A 0 score in one criterion imply a No-Go decision indicating that one of the criteria is critically missing. The order of the prioritization criteria from (i) to (iv) is also a priority for activity scoring the same.

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EJP RD

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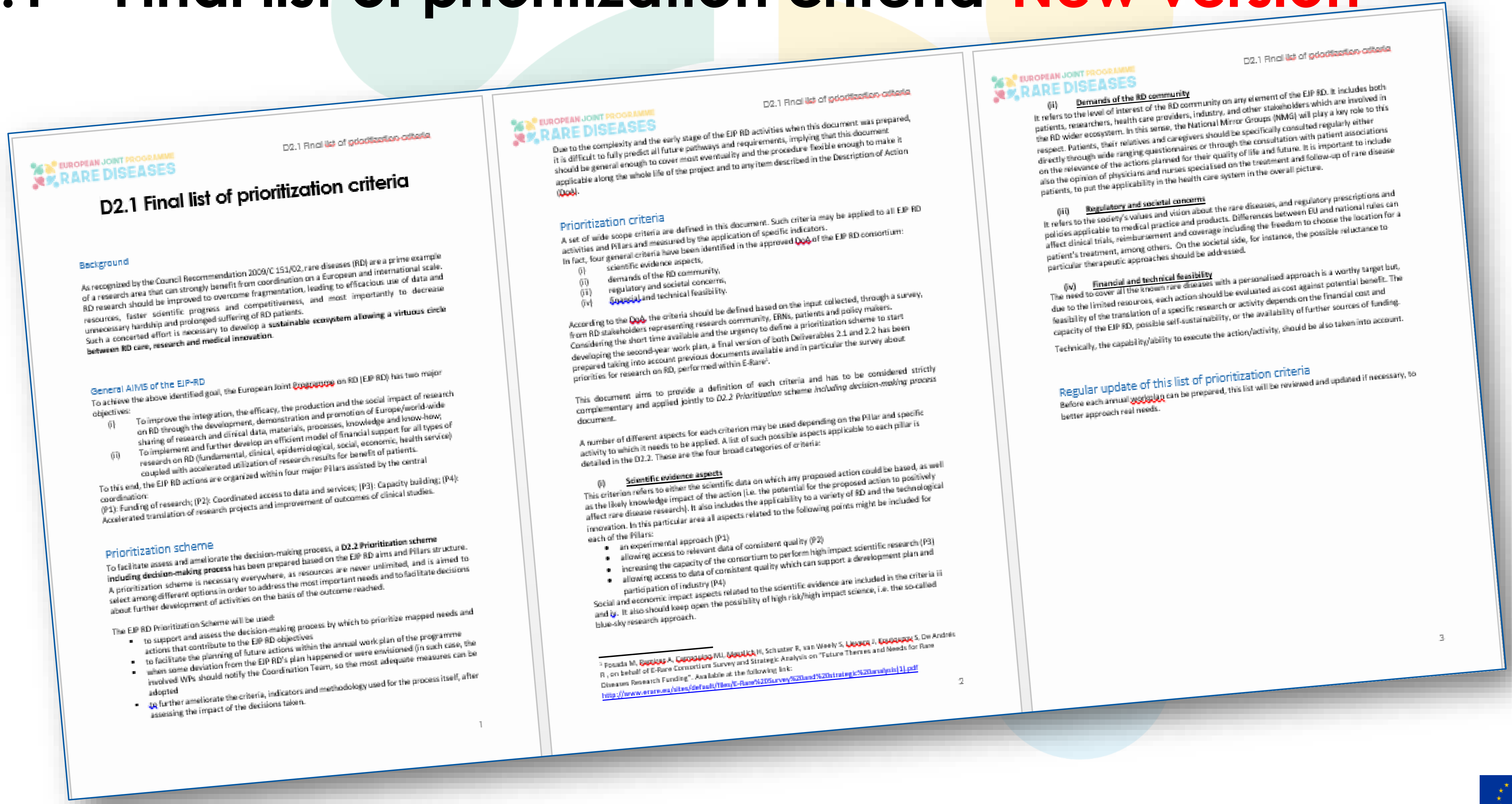
D2.1 – Final list of prioritization criteria-New version


D2.1 Final list of prioritization criteria

EUROPEAN JOINT PROGRAMME RARE DISEASES

D2.1 Final list of prioritization criteria

Background

As recognized by the Council Recommendation 2009/C 151/02, rare diseases (RD) are a prime example of a research area that can strongly benefit from coordination on a European and international scale. RD research should be improved to overcome fragmentation, leading to efficacious use of data and resources, faster scientific progress and competitiveness, and most importantly to decrease unnecessary hardship and prolonged suffering of RD patients. Such a concerted effort is necessary to develop a **sustainable ecosystem allowing a virtuous circle between RD care, research and medical innovation**.

General AIMS of the EJP-RD

To achieve the above identified goal, the European Joint Programme on RD (EJP RD) has two major objectives:

(i) To improve the integration, the efficacy, the production and the social impact of research on RD through the development, demonstration and promotion of Europe/world-wide sharing of research and clinical data, materials, processes, knowledge and know-how;

(ii) To implement and further develop an efficient model of financial support for all types of research on RD (fundamental, clinical, epidemiological, social, economic, health service) coupled with accelerated utilization of research results for benefit of patients.

To this end, the EJP RD actions are organized within four major Pillars assisted by the central coordination: (P1): Funding of research; (P2): Coordinated access to data and services; (P3): Capacity building; (P4): Accelerated translation of research projects and improvement of outcomes of clinical studies.

Prioritization scheme

To facilitate assess and ameliorate the decision-making process, a **D2.2 Prioritization scheme including decision-making process** has been prepared based on the EJP RD aims and Pillars structure. A prioritization scheme is necessary everywhere, as resources are never unlimited, and is aimed to select among different options in order to address the most important needs and to facilitate decisions about further development of activities on the basis of the outcome reached.

The EJP RD Prioritization Scheme will be used:

- to support and assess the decision-making process by which to prioritize mapped needs and actions that contribute to the EJP RD objectives
- to facilitate the planning of future actions within the annual work plan of the programme when some deviation from the EJP RD's plan happened or were envisioned (in such case, the involved WPs should notify the Coordination Team, so the most adequate measures can be adopted
- to further ameliorate the criteria, indicators and methodology used for the process itself, after assessing the impact of the decisions taken.

1

D2.1 Final list of prioritization criteria

EUROPEAN JOINT PROGRAMME RARE DISEASES

Due to the complexity and the early stage of the EJP RD activities when this document was prepared, it is difficult to fully predict all future pathways and requirements, implying that this document should be general enough to cover most eventuality and the procedure flexible enough to make it applicable along the whole life of the project and to any item described in the Description of Action (DoA).

Prioritization criteria

A set of wide scope criteria are defined in this document. Such criteria may be applied to all EJP RD activities and Pillars and measured by the application of specific indicators. In fact, four general criteria have been identified in the approved DoA of the EJP RD consortium:

(i) scientific evidence aspects,
(ii) demands of the RD community,
(iii) regulatory and societal concerns,
(iv) financial and technical feasibility.

According to the DoA, the criteria should be defined based on the input collected, through a survey, from RD stakeholders representing research community, ERNs, patients and policy makers. Considering the short time available and the urgency to define a prioritization scheme to start developing the second-year work plan, a final version of both Deliverables 2.1 and 2.2 has been prepared taking into account previous documents available and in particular the survey about priorities for research on RD, performed within E-Rare[1].

This document aims to provide a definition of each criteria and has to be considered strictly complementary and applied jointly to D2.2 *Prioritization scheme including decision-making process* document.

A number of different aspects for each criterion may be used depending on the Pillar and specific activity to which it needs to be applied. A list of such possible aspects applicable to each pillar is detailed in the D2.2. These are the four broad categories of criteria:

(i) Scientific evidence aspects

This criterion refers to either the scientific data on which any proposed action could be based, as well as the likely knowledge impact of the action (i.e. the potential for the proposed action to positively affect rare disease research). It also includes the applicability to a variety of RD and the technological innovation. In this particular area all aspects related to the following points might be included for each of the Pillars:

- an experimental approach (P1)
- allowing access to relevant data of consistent quality (P2)
- increasing the capacity of the consortium to perform high impact scientific research (P3)
- allowing access to data of consistent quality which can support a development plan and participation of industry (P4)

Social and economic impact aspects related to the scientific evidence are included in the criteria ii and iv. It also should keep open the possibility of high risk/high impact science, i.e. the so-called blue-sky research approach.

[1] Posada M, Baynam A, Campanelli MJ, Mauritz H, Schuster R, van Weely S, Lausen J, Kerzendorf S, De Andres R, on behalf of E-Rare Consortium Survey and Strategic Analysis on "Future Themes and Needs for Rare Diseases Research Funding". Available at the following link: http://www.erare.eu/sites/default/files/E-Rare%20Survey%20and%20strategic%20analysis(1).pdf

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D2.1 Final list of prioritization criteria

EUROPEAN JOINT PROGRAMME RARE DISEASES

(ii) Demands of the RD community

It refers to the level of interest of the RD community on any element of the EJP RD. It includes both patients, researchers, health care providers, industry, and other stakeholders which are involved in the RD wider ecosystem. In this sense, the National Mirror Groups (NMG) will play a key role to this respect. Patients, their relatives and caregivers should be specifically consulted regularly either directly through wide ranging questionnaires or through the consultation with patient associations on the relevance of the actions planned for their quality of life and future. It is important to include also the opinion of physicians and nurses specialised on the treatment and follow-up of rare disease patients, to put the applicability in the health care system in the overall picture.

(iii) Regulatory and societal concerns

It refers to the society's values and vision about the rare diseases, and regulatory prescriptions and policies applicable to medical practice and products. Differences between EU and national rules can affect clinical trials, reimbursement and coverage including the freedom to choose the location for a patient's treatment, among others. On the societal side, for instance, the possible reluctance to particular therapeutic approaches should be addressed.

(iv) Financial and technical feasibility

The need to cover all the known rare diseases with a personalised approach is a worthy target but, due to the limited resources, each action should be evaluated as cost against potential benefit. The feasibility of the translation of a specific research or activity depends on the financial cost and capacity of the EJP RD, possible self-sustainability, or the availability of further sources of funding.

Technically, the capability/ability to execute the action/activity, should be also taken into account.

Regular update of this list of prioritization criteria

Before each annual workplan can be prepared, this list will be reviewed and updated if necessary, to better approach real needs.

3

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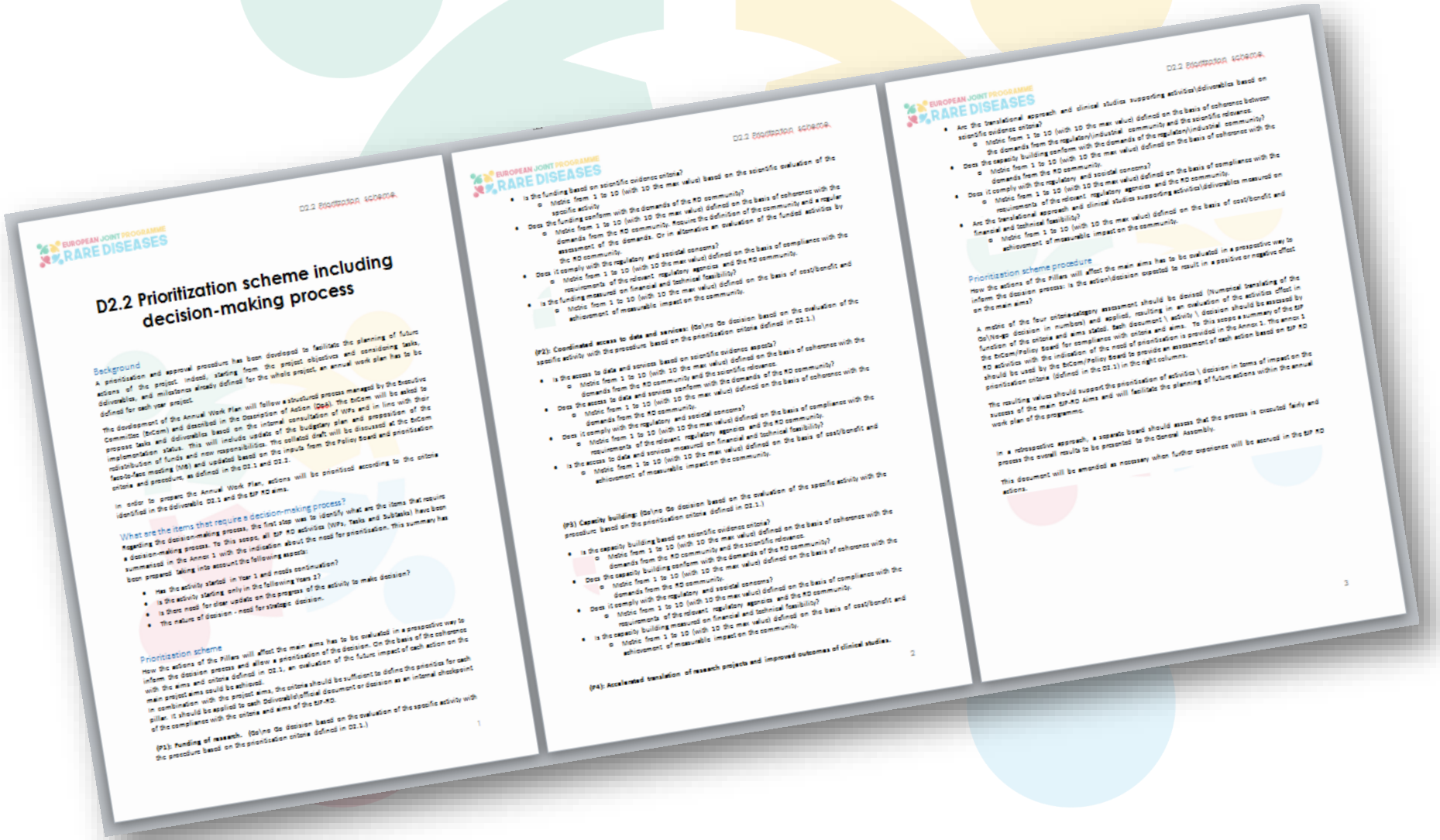
D2.2 – Prioritization scheme including decision-making process



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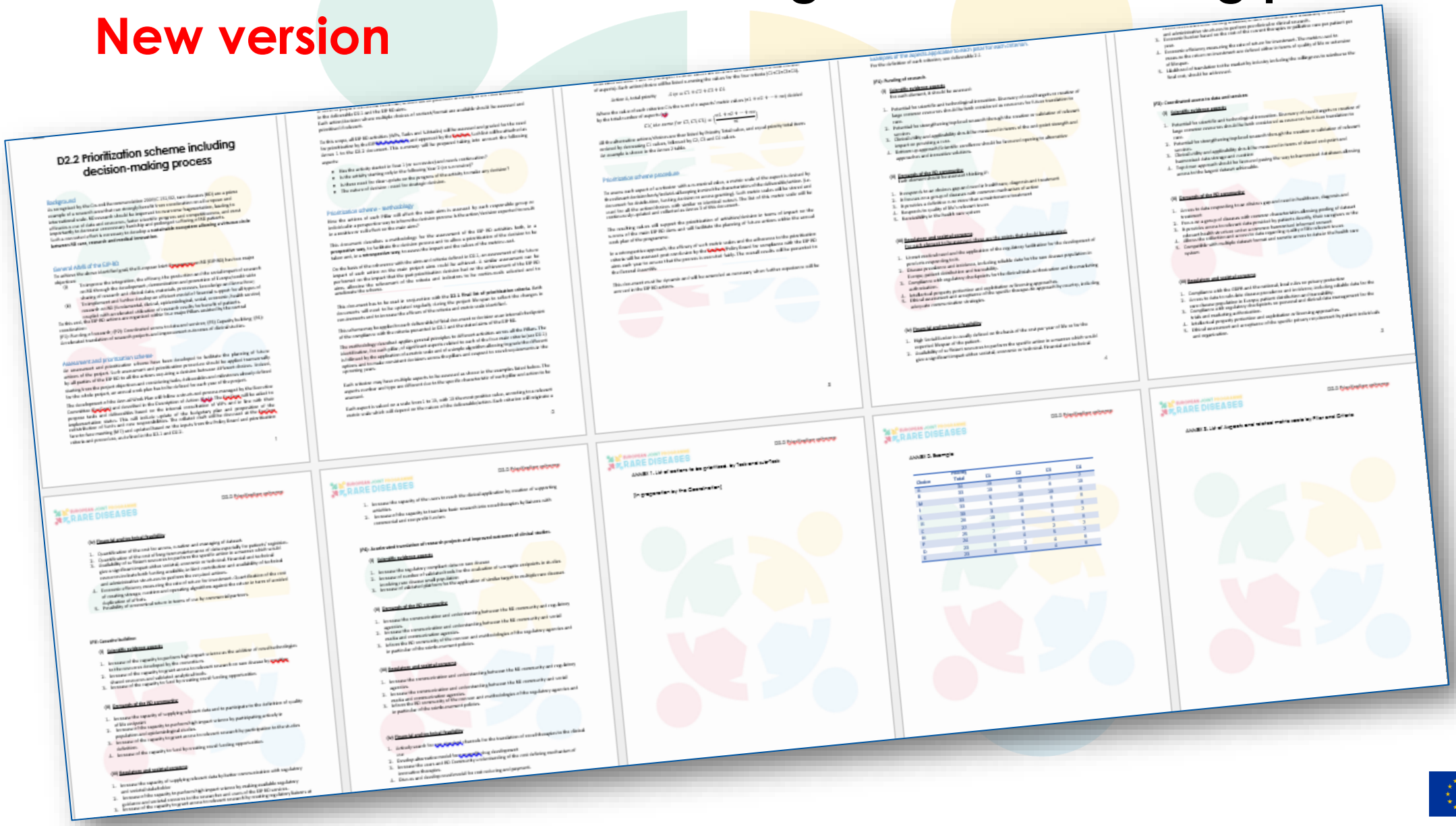
D2.2 – Prioritization scheme including decision-making process

New version



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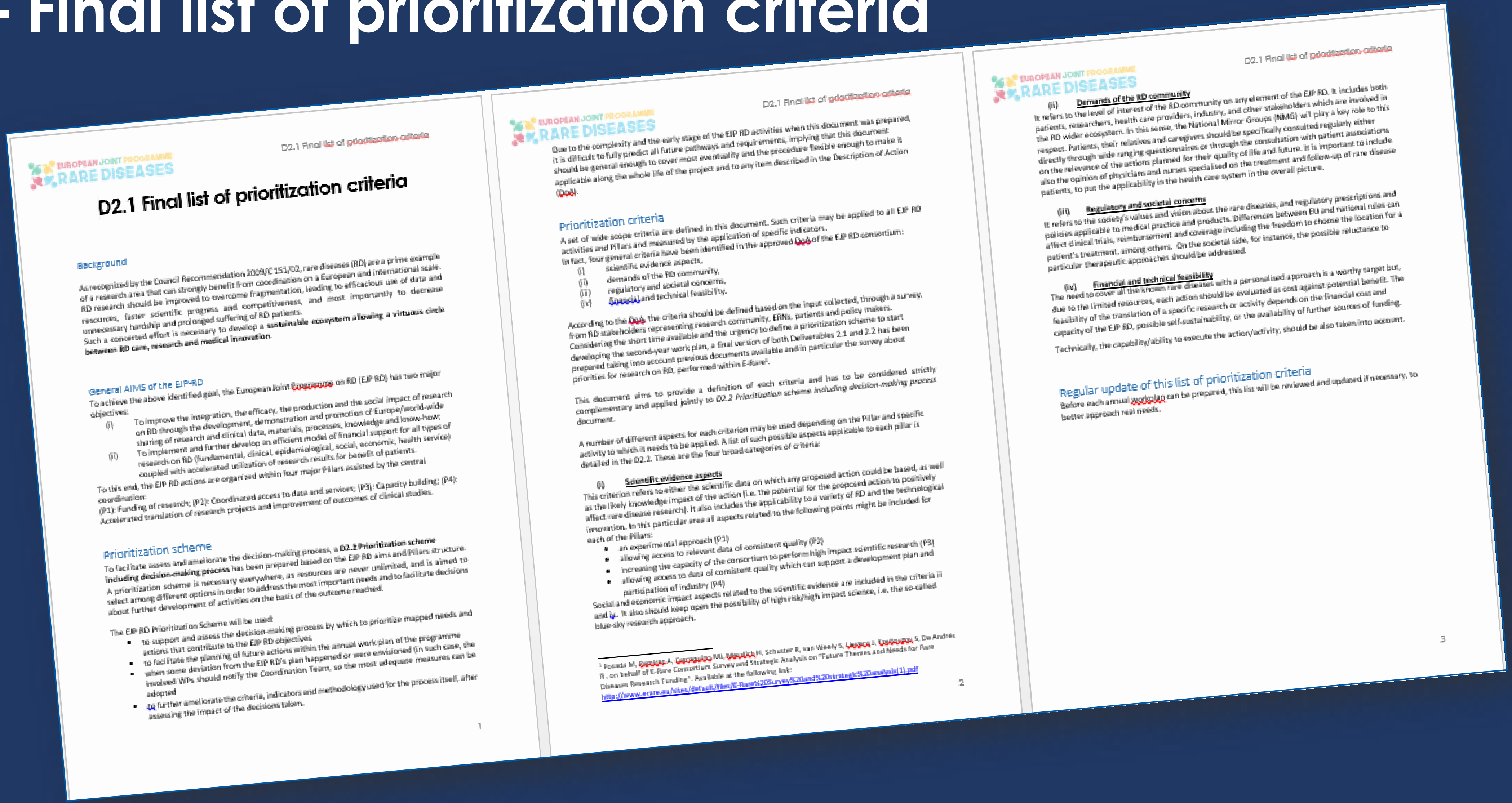
D2.1 – Final list of prioritization criteria



EUROPEAN JOINT PROGRAMME
RARE DISEASES

D2.1 – Final list of prioritization criteria

According to the Proposal, the criteria will be defined based on the input collected (through a survey) from RD stakeholders representing research community, ERNs, patients and policy makers.

For this first year the criteria and scheme were defined taking into account previous documents available, and this will be updated when the survey can be addressed to all the stakeholders

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D2.1 – Final list of prioritization criteria

- To facilitate assess and ameliorate the decision-making process, a **D2.2 Prioritization scheme including decision-making process** has been prepared based on the EJP RD aims and Pillars structure.
- A **prioritization scheme is necessary everywhere**, as **resources are never unlimited**, and is aimed to select among different options in order to address the most important needs and to facilitate decisions about further development of activities on the basis of the outcome reached.

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D2.2 – Prioritization scheme – It will be used:

- to support and assess the **decision-making process** by which to prioritize mapped needs and actions that contribute to the EJP RD objectives
- to facilitate the **planning of future actions** within the annual work plan of the programme
- when some **deviation from the EJP RD's plan** happened or were envisioned (in such case, the involved WPs should notify the Coordination Team, so the most adequate measures can be adopted
- to **further ameliorate the criteria, indicators and methodology** used for the process itself, after assessing the impact of the decisions taken.

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D2.2 – Prioritization scheme

Due to the **complexity and the early stage of the EJP RD** activities when this document was prepared:

- It is **difficult to fully predict** all future pathways and requirements
- This implies that **this document should be general enough** to cover most eventuality **and the procedure flexible enough** to make it applicable along the whole life of the project and to any item included in the Description of Action (DoA).

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D2.1 – Final list of prioritization criteria

Four wide scope categories of criteria:

- Scientific evidence aspects
- Demands of the RD community
- Regulatory and societal concerns
- Financial and technical feasibility

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D2.1 – Final list of prioritization criteria

Regular update of the list of prioritization criteria

Before each annual workplan can be prepared, this list will be reviewed and updated if necessary, to better approach real needs

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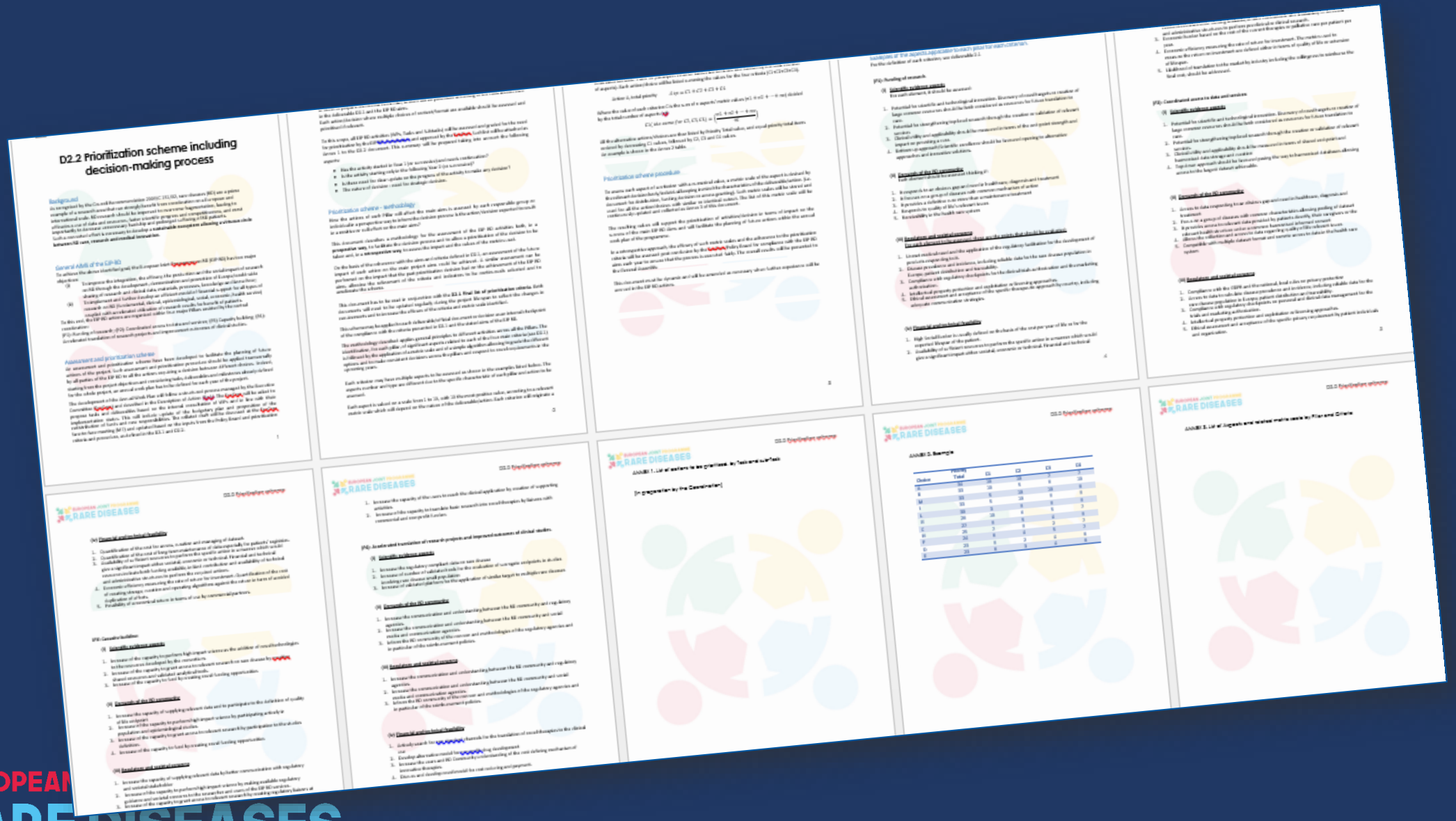
D2.2 – Prioritization scheme including decision-making process



D2.2 – Prioritization scheme including decision-making process

An assessment and prioritization scheme have been developed:

- To **facilitate the planning of future actions** of the project.
- **It should be applied transversally** by all parties of the EJP RD to all the actions requiring a decision between different choices.
- Indeed, starting from the project objectives and considering tasks, deliverables and milestones already defined for the whole project, an **annual work plan has to be defined for each year of the project**.

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D2.2 – Prioritization scheme – Transversally applicable

3.1a. List of workpackages

TITLE	LEAD BENEFICIARY
COORDINATION & TRANSVERSAL ACTIVITIES	INSERM (Coo)
WP1. Coordination and management	INSERM (Coo)
WP2. Integrative research and innovation strategy	ISCIII, ISS
WP3. Sustainability strategy and business plan	INSERM (RaDiCo), EATRIS, ISCIII
WP4. Ethical, regulatory, legal and IPR framework of the EJP RD	FGB
WP5. Communication & dissemination	INSERM (Coo)
PILLAR 1: RESEARCH COLLABORATIVE FUNDING	DLR, ZonMw
WP6. Joint Transnational Calls for collaborative research projects	DLR
WP7. Networking to share knowledge on rare diseases	ZonMw
WP8. Rare Disease Research Challenges	FFRD
WP9. Monitoring of funded projects	CSO/MOH
PILLAR 2: INNOVATIVE COORDINATED ACCESS TO DATA AND SERVICES FOR TRANSFORMATIVE RARE DISEASES RESEARCH	INSERM (Orphanet), UKL-HD
WP10. User-driven strategic planning and transversal activities for Pillar 2 data ecosystem	INSERM (Orphanet), ULEIC
WP11. Common virtual platform for discoverable data and resources for RD research	INSERM (Orphanet), CNAG
WP12. Enabling sustainable FAIRness and Federation at the record level for RD data, patients and samples	ULEIC, LUMC
WP13. Enabling multidisciplinary, holistic approaches for rare disease diagnostics and therapeutics	UM, UKL-HD
PILLAR 3: CAPACITY BUILDING AND EMPOWERMENT	EURORDIS, VUHSK
WP14. Training on data management & quality	ISS
WP15. Capacity building and training of patients and researchers in Rare Disease research and processes	EURORDIS
WP16. Online Academic education course	FFRD
WP17. ERN RD training and support programme	EKUT
WP18. Development and adaptation of training activities	VUHSK, CMHI, EURORDIS
PILLAR 4: ACCELERATING THE TRANSLATION OF HIGH POTENTIAL PROJECTS AND IMRPOVING OUTCOMES OF CLINICAL STUDIES IN SMALL POPULATIONS	APHP, EATRIS
WP19. Facilitating partnerships and accelerating translation for higher patient impact	EATRIS, FTELE
WP20. Accelerating the validation, use and development of innovative methodologies tailored for clinical trials in RDs	UKA, APHP, HSK (MetabERN)

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Prioritization scheme

- How the actions of the Pillars will affect the main aims has to be **evaluated in a prospective way** to inform the decision process and allow a prioritization of the decision.

- In combination with the project aims, **the criteria should be sufficient to define the priorities for each pillar.**

- It should be **applied to each Deliverable/official document or decision** as an internal checkpoint of the compliance with the criteria and aims of the EJP-RD.

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What are the items that require a decision-making process?

All EJP RD activities (WPs, Tasks and Subtasks) are been summarized with the indication about the need for prioritization.

This summary has been prepared taking into account the following aspects:

- Has the activity started in Year 1 and needs continuation?
- Is the activity starting only in the following Year 2?
- Is there need for clear update on the progress of the activity to make decision?
- The nature of decision - need for strategic decision.

This procedure is applicable for all the years of the project

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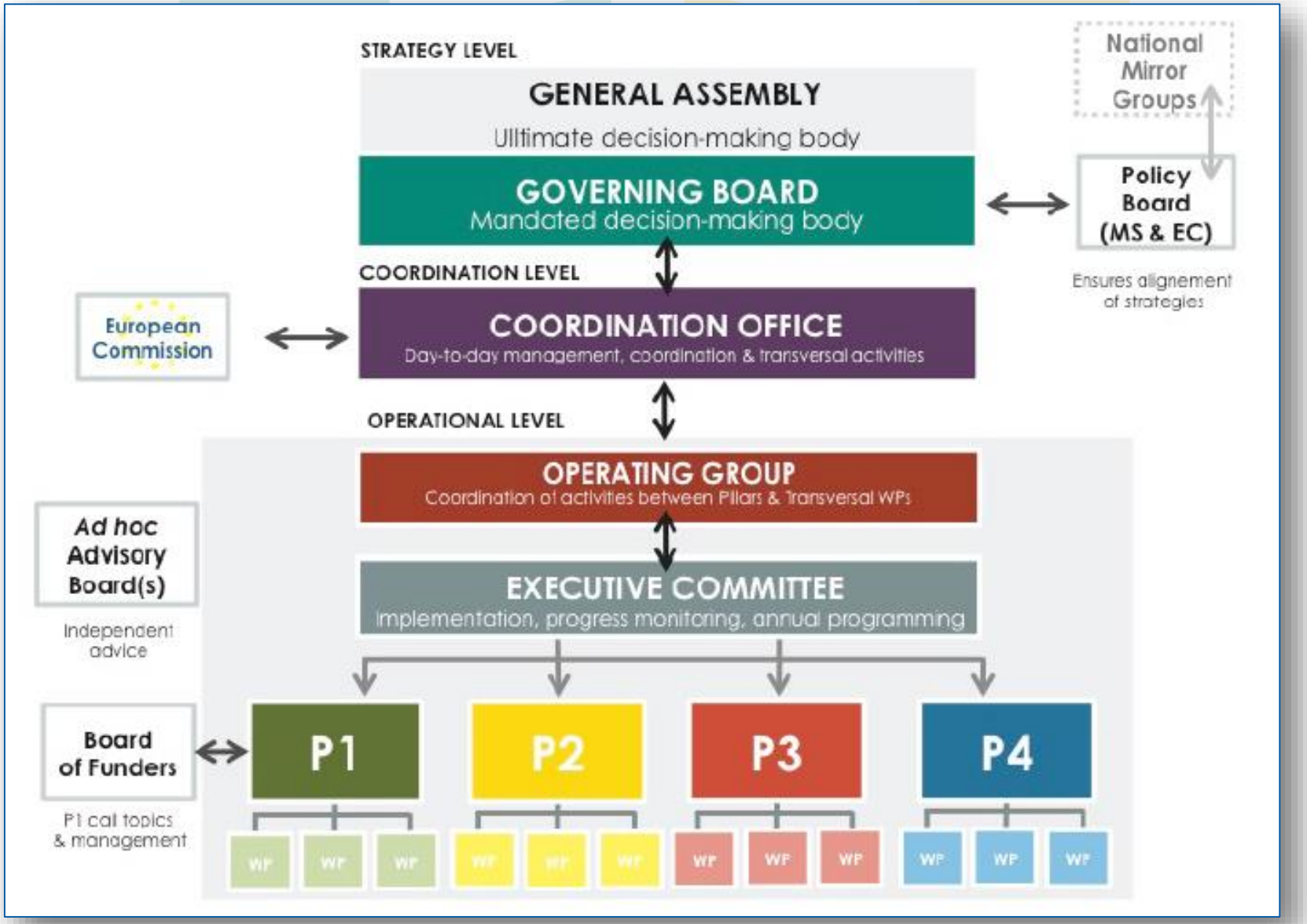
Decision-making process



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Decision-making process - Prioritisation

Coordination

- The development of the Annual Work Plan will follow a structured process initiated and managed from the Coordination and the Executive Committee (ExeCom)

Executive Committee

- The ExeCom will be asked to **propose tasks and deliverables** based on the internal consultation of WPs and in line with their implementation status. This will include **update of the budgetary plan** and proposition of the redistribution of funds and new responsibilities.

Policy Board

- The collated draft will be **discussed at the ExeCom face-to-face meeting (M7)** and updated based on the **inputs from the Policy Board** and prioritization criteria and procedure, as defined in the D2.1 and D2.2.

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Decision-making process - Prioritisation

Criteria

- Four categories

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D2.1 – Final list of prioritization criteria

Four wide scope categories of criteria:

- Scientific evidence aspects
- Demands of the RD community
- Regulatory and societal concerns
- Financial and technical feasibility

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Decision-making process - Prioritisation

Criteria

- Four categories

Aspects in each Pillar/WP/T/ST

- Variable number

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Prioritisation - Scoring

Criteria

Action A, total priority:

$$A\ tp = C1 + C2 + C3 + C4$$

Aspects in each Pillar/WP/T/ST

$$C1 = \left(\frac{n1 + n2 + \cdots + nx}{nt}\right)$$

$$(\ the\ same\ for\ C2, C3, C4)$$

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Prioritisation - Scoring

EUROPEAN JOINT PROGRAMME RARE DISEASES

D2.2 Prioritization scheme

ANNEX 2. Example

Choice	Priority Total	C1	C2	C3	C4
A	34	10	10	7	7
B	33	10	5	8	10
M	33	5	10	10	8
I	33	5	10	9	9
L	30	3	9	9	9
G	28	10	6	5	7
C	27	9	5	4	9
H	25	7	9	2	7
F	24	8	4	5	7
D	23	9	2	4	8
E	23	8	3	4	8

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Prioritisation - Scoring

Examples of the aspects applicable to each pillar for each criterion.

For the definition of each criterion, see deliverable 2.1

(P1): Funding of research.

(i) Scientific evidence aspects

For each element, it should be assessed:

1. Potential for scientific and technological innovation. Discovery of novel targets or creation of large common resources should be both considered as resources for future translation to care.
2. Potential for strengthening top-level research through the creation or validation of relevant services
3. Clinical utility and applicability should be measured in terms of the end-point strength and impact on providing a cure.
4. Bottom-up approach/Scientific excellence should be favoured opening to alternative approaches and innovative solutions.

(ii) Demands of the RD community

Each element should be assessed thinking if:

1. It responds to an obvious gap and need in healthcare, diagnosis and treatment
2. It focuses on a group of diseases with common mechanism of action
3. It provides a definitive cure more than a maintenance treatment
4. Responds to quality of life's relevant issues
5. Receivability in the health care system

(iii) Regulatory and societal concerns

For each element to be assessed, these are the points that should be evaluated:

1. Unmet medical need and the application of the regulatory facilitation for the development of products responding to it.
2. Disease prevalence and incidence, including reliable data for the rare disease population in Europe, patient distribution and traceability.
3. Compliance with regulatory checkpoints for the clinical trials authorization and the marketing authorization.
4. Intellectual property protection and exploitation or licensing approaches.
5. Ethical assessment and acceptance of the specific therapeutic approach by country, including adequate communication strategies.

(iv) Financial and technical feasibility

1. High Social Burden is usually defined on the basis of the cost per year of life or for the expected lifespan of the patient.
2. Availability of sufficient resources to perform the specific action in a manner which would give a significant impact either societal, economic or technical. Financial and technical

EUROPEAN JOINT PROGRAMME RARE DISEASES
D2.2 Prioritization scheme

resources indicate both funding available, in-kind contribution and availability of technical and administrative structures to perform pre-clinical or clinical research.

3. Economic burden based on the cost of the current therapies or palliative care per patient per year.
4. Economic efficiency measuring the rate of return for investment. The metrics used to measure the return on investment are defined either in terms of quality of life or extension of lifespan.
5. Likelihood of translation to the market by industry including the willingness to reimburse the final cost, should be addressed.

(P2): Coordinated access to data and services:

(i) Scientific evidence aspects

1. Potential for scientific and technological innovation. Discovery of novel targets or creation of large common resources should be both considered as resources for future translation to care.
2. Potential for strengthening top-level research through the creation or validation of relevant services.
3. Clinical utility and applicability should be measured in terms of shared end-point and harmonised data storage and curation
4. Top-down approach should be favoured paving the way to harmonised databases allowing access to the largest dataset achievable.

(ii) Demands of the RD community

1. Access to data responding to an obvious gap and need in healthcare, diagnosis and treatment
2. Focus on a group of diseases with common characteristics allowing pooling of dataset
3. It provides access to relevant data provided by patients directly, their caregivers or the relevant health structure under a common harmonised informed consent
4. Allows the collection and access to data regarding quality of life relevant issues
5. Compatible with multiple dataset format and remote access to data in the health care system

(iii) Regulatory and societal concerns

1. Compliance with the GDPR and the national, local rules on privacy protection
2. Access to data to calculate disease prevalence and incidence, including reliable data for the rare disease population in Europe, patient distribution and traceability.
3. Compliance with regulatory checkpoints on personal and clinical data management for the trials and marketing authorization.
4. Intellectual property protection and exploitation or licensing approaches.
5. Ethical assessment and acceptance of the specific privacy requirement by patient individuals and organization.

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Prioritisation - Scoring

(iv) Financial and technical feasibility

1. Quantification of the cost for access, curation and managing of dataset.
2. Quantification of the cost of long-term maintenance of data especially for patients' registries.
3. Availability of sufficient resources to perform the specific action in a manner which would give a significant impact either societal, economic or technical. Financial and technical resources indicate both funding available, in-kind contribution and availability of technical and administrative structures to perform the required actions.
4. Economic efficiency measuring the rate of return for investment. Quantification of the cost of creating storage, curation and operating algorithms against the return in term of avoided duplication of efforts.
5. Possibility of economical return in terms of use by commercial partners.

(P3) Capacity building:

(i) Scientific evidence aspects

1. Increase of the capacity to perform high impact science as the addition of novel technologies to the resources developed by the consortium.
2. Increase of the capacity to grant access to relevant research on rare disease by creating shared resources and validated analytical tools.
3. Increase of the capacity to fund by creating novel funding opportunities

(ii) Demands of the RD community

1. Increase the capacity of supplying relevant data and to participate to the definition of quality of life endpoint
2. Increase of the capacity to perform high impact science by participating actively in population and epidemiological studies.
3. Increase of the capacity to grant access to relevant research by participation to the studies definition.
4. Increase of the capacity to fund by creating novel funding opportunities

(iii) Regulatory and societal concerns

1. Increase the capacity of supplying relevant data by better communication with regulatory and societal stakeholder
2. Increase of the capacity to perform high impact science by making available regulatory guidance and societal concerns to the researcher and users of the EJP RD services.
3. Increase of the capacity to grant access to relevant research by creating regulatory liaisons at the early stage of research.

(iv) Financial and technical feasibility

EUROPEAN JOINT PROGRAMME RARE DISEASES D2.2 Prioritization scheme

1. Increase the capacity of the users to reach the clinical application by creation of supporting activities.
2. Increase of the capacity to translate basic research into novel therapies by liaisons with commercial and non-profit funders

(P4): Accelerated translation of research projects and improved outcomes of clinical studies.

(i) Scientific evidence aspects

1. Increase the regulatory compliant data on rare disease
2. Increase of number of validated tools for the evaluation of surrogate endpoints in studies involving rare disease small population
3. Increase of validated platform for the application of similar target to multiple rare diseases

(ii) Demands of the RD community

1. Increase the communication and understanding between the RD community and regulatory agencies.
2. Increase the communication and understanding between the RD community and social media and communication agencies.
3. Inform the RD community of the concern and methodologies of the regulatory agencies and in particular of the reimbursement policies.

(iii) Regulatory and societal concerns

1. Increase the communication and understanding between the RD community and regulatory agencies.
2. Increase the communication and understanding between the RD community and social media and communication agencies.
3. Inform the RD community of the concern and methodologies of the regulatory agencies and in particular of the reimbursement policies.

(iv) Financial and technical feasibility

1. Actively search for non standard channels for the translation of novel therapies to the clinical use
2. Develop alternative model for non profit drug development
3. Increase the users and RD Community understanding of the cost defining mechanism of innovative therapies.
4. Discuss and develop novel model for cost reducing and payment.

The above list of aspects for each criterion is only a partial example and will be revised regularly according to the indication of the ExeCom and the assessment of the prioritization scheme performed in a retrospective fashion on the actions and decisions made by the consortium.

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Prioritization scheme procedure

- **Prospective assessment** to inform the decision process
- To **assess each aspect of a criterion with a numerical value**, a metric scale of the aspect is devised by the relevant decision body/individual keeping in mind the characteristics of the deliverable/action. (i.e. document for distribution, funding decision or access granting). Such metric scales will be stored and used for all the action/choices with similar or identical output. The list of this metric scale will be continuously updated and collected as Annex 3 of this document.
- The **resulting values will support the prioritization of activities/decision** in terms of impact on the success of the main EJP RD Aims and will facilitate the planning of future actions within the annual work plan of the programme.
- In a **retrospective approach**, the efficacy of such metric scales and the adherence to the prioritization criteria will be assessed post conclusion by the ExeCom/Policy Board for compliance with the EJP RD aims each year to ensure that the process is executed fairly. The overall results could be presented to the General Assembly.
- This **document must be dynamic** and will be amended as necessary when further experience will be accrued in the EJP RD actions.

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What are the items that require a decision-making process?

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Decision-making process - Prioritisation

TASK
Task 1.1 Implementation and responsibilities of the coordination office (MS 1.1;1.2;1.3)
SubTask 1.1.1: Scientific Secretariat of IRDiRC
Task 1.2 Coordination & support within and across pillars
SubTask 1.2.1: Development of Annual Progress Report and Annual Work Plans
SubTask 1.2.2: Organization of EJP RD governance meetings
SubTask 1.2.3: EJP RD internal communication
SubTask 1.2.4: Initiating new partnerships
Task 1.3 Monitoring of the EJP RD activities and achievements
SubTask 1.3.1: Identification of key performance indicators
SubTask 1.3.2: Implementation of the monitoring process
Task 1.4 Data management plan

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Decision-making process - Prioritisation

TASK
Task 2.1 Prioritization scheme for EJP RD actions (MS 2.1;2.2)
Task 2.2 Mapping the research and innovation need (MS2.3)
Task 2.3 Scientific programming of joint transnational calls
Task 2.4 Management of the medium, longer-term research strategy questions and dedicated linkage with Task Forces of IRDiRC
Task 2.5 Translation/impact of prioritization on national and EU strategies
Task 3.1 Service roadmap alignment of the needs, expectations and engagement of the different RD research stakeholders in Europe with respect to EJP RD sustainability
Subtask 3.1.1 integration and maintainance of the information collected on needs, expectations and possible contributions of National Programme Owners (PO) and Programme Managers (PM)
Subtask 3.1.2 integration and maintainance of the information collected on needs, expectations and possible contributions of other stakeholders
Task 3.2 Preparation of the sustainable EJP-RD services catalogue, supporting the EJP-RD dissemination and communication activities
Task 3.3 Preparation of the EJP-RD sustainability plan with business plan (MS3.1)
Task 3.4 Sustainability roadmap (MS3.2)

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Decision-making process - Prioritisation

TASK
Task 4.2 Managing ethical and regulatory issues (MS4.2)
Task 4.3 Managing legal and IPR issues (MS4.3)
Task 5.1 EJP RD external communication & dissemination of results (MS5.1)
Subtask 5.1.1: Implementation of the EJP RD communication strategy
Subtask 5.1.2: Communication and dissemination tools
Subtask 5.1.3: Support of communication and dissemination activities of IRDiRC
Task 5.2 Integrating EJP-RD communication and dissemination strategies with the strategies of involved stakeholders (MS5.2;5.3)
Task 6.1 1st co-funded Joint Transnational Call (JTC 2019)
Subtask 6.1.1 Topic selection and eligibility criteria
Subtask 6.1.2 Preparation of the call and establishment of the joint call secretariat
Subtask 6.1.3 Evaluation of the joint call
Subtask 6.1.4: Funding decision
Subtask 6.1.5 Quality management of call procedures
Task 6.2 2nd co-funded Joint Transnational Call (JTC 2020)
Task 6.3 3rd Joint Transnational Call (JTC 2021) not co-funded by the EC
Task 6.4 4th Joint Transnational Call (JTC 2022) not co-funded by the EC

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Decision-making process - Prioritisation

TASK
Task 7.1 Development of the networking scheme (MS7.1)
Task 7.2 Application and evaluation of proposals for the Networking Scheme
Task 7.3 Early assessment of the Networking scheme
Task 8.1 Development of the scheme (MS8.1)
Task 8.2 Identification of challenges
Task 8.3 The call for projects, eligibility criteria and evaluation
Task 8.4 Management of the funding resources, establishment of contracts and distribution of funding to selected projects
Task 9.1 Monitoring of Joint Transnational Calls for collaborative research projects (MS9.1)
Subtask 9.1.1 Definition/Update of the indicators for ex-post evaluation of funded projects in each call
Subtask 9.1.2 Adaptation of the monitoring tool for follow-up and assessment of funded projects
Subtask 9.1.3 Follow-up, monitoring and assessment of funded projects in WP6, including organization of mid-term monitoring meetings for each Call
Subtask 9.1.4 Assessment of E-Rare 3 Joint Call Funding programme
Task 9.2 Monitoring of networking to share knowledge on rare diseases (MS9.2)
Subtask 9.2.1 Definition of indicators for ex-post evaluation of funded networking events
Subtask 9.2.2 Adaptation of monitoring tool for follow-up and assessment of funded networking events
Subtask 9.2.3 Assessment of the funded networking events
Task 9.3 Monitoring of Rare Disease Research Challenge (MS9.3)

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Decision-making process - Prioritisation

TASK
Task 10.1 Convene yearly retreats leading to Pillar 2 annual strategic plans (MS10.1)
Subtask 10.1.1 Whole-RD-community annual retreat
Subtask 10.1.2 Annual strategic plan for Pillar 2
Task 10.2 Coordinating Pillar 2 Work Packages outputs and aligning to RD community needs (MS10.2)
Subtask 10.2.1 Bi-monthly TCs and reports with WPs leaders
Subtask 10.2.2 Community surveys and structured interviews
Task 10.3 Coordination of technical GDPR implementation
Task 10.4 Quality oversight
Task 10.5 Sustainability and scaling up (MS10.3;10.4)
Task 11.1 Metadata and ontological model definition for cataloging resources for RD research (MS11.1)
Subtask 11.1.1: A set of metadata will be defined to describe each RD resource to be included in the virtual platform
Subtask 11.1.2 Application ontology to provide the metadata model for the future VP
Subtask 11.1.3 Prioritisation of resources to be included
Task 11.2 FAIR-compliant virtual platform for discovery of RD resources
Subtask 11.2.1 datasets from Orphanet (on biobanks and registries), BBMRI and ERDRI will be compared to detect overlaps and gaps on data described in each catalog
Subtask 11.2.2 Selected projects and trials will be pre-annotated with metadata from the Orphanet catalog
Subtask 11.2.3 ELIXIR's bio.tools catalogue will continue its adaptation for the RD community
Subtask 11.2.4 development of a API and a SPARQL access point

EJP RD

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Decision-making process - Prioritisation

Task 11.3 Data deposition and access to data infrastructures for RD research (MS11.2;11.3)
Subtask 11.3.1 Grow and develop deposition and access capabilities of existing RD-related databases
Subtask 11.3.2 Implement a unified authentication and authorisation mechanism, based on ELIXIR's AAI
Subtask 11.3.3 Adopt a Privacy-Preserving Record Linkage solution to enable data from the same individual to be connected across resources
Subtask 11.3.4 Provide data deposition guidelines and documentation
Task 11.4 Provision of RD analysis and data sharing capabilities through online resources (MS11.4)
Subtask 11.4.1 Provide RD phenotype-genotype analysis and data sharing capabilities through the RDConnect platform for molecular diagnostics and gene discovery
Subtask 11.4.2 Collaborative integrated analysis of multi-omics data from selected use cases
Subtask 11.4.3 Provide capabilities for custom analyses through cloud-based solutions
Subtask 11.4.4 User-driven implementation of new analysis functionalities and tools

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Decision-making process - Prioritisation

Task 12.1 Compare and align core interoperability standards of relevance (MS12.1)
Subtask 12.1.1 Metadata and data models
Subtask 12.1.2 Federation architectures and network registers
Subtask 12.1.3 Automated Accessibility and Reusability elements
Subtask 12.1.4 Query languages and data exchange formats
Subtask 12.1.5 Ontologies & semantics for record-level clinical and preclinical information
Task 12.2 Multi-team software development to enable the FAIR ecosystem (MS12.2)
Subtask 12.2.1 Semantic and syntactic harmonisation of data records
Subtask 12.2.2 Data anonymisation, extraction, obfuscation, and privacy control
Subtask 12.2.3 Metadata-level, data-level and sample-level discovery support
Subtask 12.2.4 Management of consent and data use conditions, plus automated access control
Task 12.3 Combining expertise for practical FAIRification support (MS12.3;12.4)
Subtask 12.3.1 Coordinating teams of experts
12.3.2 Brokered hosting service
Subtask 12.3.3 FAIR data stewardship service

EJP RD

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Decision-making process - Prioritisation

TASK
Task 13.1 Fill a number of pre identified gaps to allow integrative systems biology approaches for rare diseases
Subtask 13.1.1. Pathways created and expert curated
Subtask 13.1.2 Mapping to genes
Subtask 13.1.3 Mapping (combinations) of variants to protein function
Subtask 13.1.4 Link to external FAIR data
Subtask 13.1.5 Network repository:
Subtask 13.1.6 Environmental lifestyle
Subtask 13.1.7 Treatment drugs
Subtask 13.1.8 Environmental toxicology
Subtask 13.1.9 Workflow leading to understanding of disease mechanisms and diagnosis
Subtask 13.1.10 Link to Adverse Outcome Pathway approach
Task 13.2 Organise proof of principle studies to identify efficacy of the work done and identify remaining (including new) gaps (MS13.1;13.2)
WP14. Training on data management & quality (MS14.2)
Task 14.1 Training on the Orphanet nomenclature and RD ontologies for RD research (MS14.1)
Subtask 14.1.1 Assessment of needs and establishment of SMART learning objectives for the Orphanet nomenclature and RD ontologies for RD research training
Subtask 14.1.2 Design and development of training materials
Task 14.2 Standards and quality of genetics/genomics data in laboratory and clinical research practice
Task 14.3 Training on strategies to foster solutions of undiagnosed rare disease cases
Task 14.4 Training for biobanks and researchers/clinicians on sample data management
Subtask 14.4.1: Kicking off EJP RD biobank training program
Subtask 14.4.2 Biobank training Workshops
Task 14.5 Training on rare disease registries and FAIRification of data at the source
Task 14.6 Training on the European Rare Diseases Registry Infrastructure (ERDRI)

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Decision-making process - Prioritisation

Task 15.1 ExPRESS Expert Patients and Researchers EURORDIS Summer School
Task 15.2 Training for patient advocates on scientific innovation and translational research aspects in rare diseases
Task 15.3 Training for patient representatives and advocates on leadership and communication skills
Task 15.4 Educational materials and activities for paediatric patients
Task 16.1 Assess the needs, target audience, main topics of the academic education course
Task 16.2 Developing the online course (MS16.1,16.2,16.3,16.4;16.5)
Subtask 16.2.1: Format, technical issues, status, and accreditation
Subtask 16.2.2: Identify and work with relevant European universities, learned societies and other institutions to develop the content of the academic course
Task 16.3 Follow-up, assessment and monitoring of the e-learning course
Task 17.1 Mapping to collect information on preferences, needs and resources from the ERNs ecosystem (MS17.1)
Task 17.2 Development of training programs for the ERN training networks based on the results of survey (MS17.2)
Task 17.3 Deliver training programs through the ERN training networks
Task 17.4 Accreditation of training activities
Task 18.1 Evaluation of developing needs according to progress of work in Pillars 2 and 4 and relevant adaptation of training activities
Task 18.2 Evaluation of strategic priorities and development of existing training activities according to specific needs of EU-13 countries
Task 18.3 Evaluation of developing ERN training and support needs and adaptation of training activities in WP17 of Pillar 3

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Decision-making process - Prioritisation

TASK
Task 19.1 Accelerating translation (MS19.1)
Subtask 19.1.1: Innovation Management Toolbox
Subtask 19.1.2: Assessment of translational research projects
Subtask 19.1.3: Real-time mentoring and technical support for translational research projects
Task 19.2 Support in exploitation and follow-on funding
Subtask 19.2.1: Assess potential of projects
Subtask 19.2.2: Development and exploitation plan
Subtask 19.2.3: Follow-on funding
Subtask 19.2.4: Partnering support
Task 19.3 Evaluation of Innovation Management and exploitation support tasks (MS19.2)
Task 19.4 Roadmap for a European investment platform for RD (MS19.3)
Task 20.1 Task Force Group (MS20.1)
Subtask 20.1.1 Establishment the Task force group
Subtask 20.1.2 Create, disseminate and analyse surveys for the ERNs
Subtask 20.1.3: Mapping existing methodologies and prioritize the needs for demonstration or innovative methodologies for clinical studies in rare diseases
Task 20.2 Support in design and planning of RD clinical trials (MS20.4)
Subtask 20.2.1 Establishment of the clinical study support office
Subtask 20.2.2 Operation of the clinical study helpdesk
Task 20.3 Demonstration projects on existing statistical methodologies to improve RD clinical trials (MS20.2)
Subtask 20.3.1 Launch of the call for demonstration pilot projects
Task 20.4 Projects on innovative methodologies to improve RD clinical trials in limited populations (MS20.3)

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What are the items that require a decision-making process?

All EJP RD activities (WPs, Tasks and Subtasks) are been summarized with the indication about the need for prioritization.

This summary has been prepared taking into account the following aspects:

- Has the activity started in Year 1 and needs continuation?
- Is the activity starting only in the following Year 2?
- Is there need for clear update on the progress of the activity to make decision?
- The nature of decision - need for strategic decision.

This procedure is applicable for all the years of the project

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Thank you for your attention and comments

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EJP RD
Communication Strategy

Eleonora Passeri

EUROPEAN JOINT PROGRAMME
RARE DISEASES

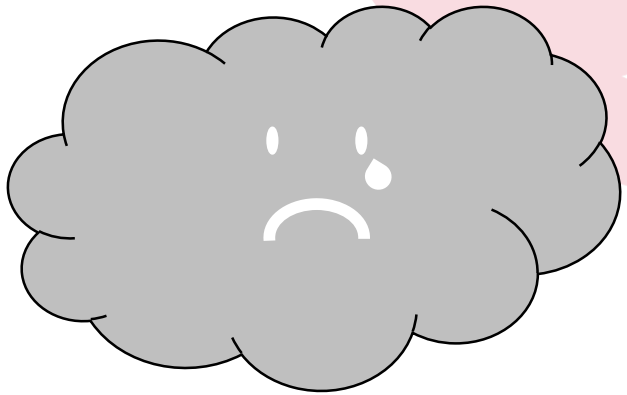
EJP RD Communication “History”

January

July

December

2019



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EJP RD Communication “Tools”

Website	http://www.ejprarediseases.org/
Newsletters	Internal and External –monthly-
Twitter account	@EJPRareDiseases
Communication Pack	Templates (presentation, poster, certificates), logos, EU funding statements for poster and publications, color codes

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Communication: Definition“S”

- A process by which **information is exchanged** between individuals through a common system of **symbols, signs, or behavior**
- Information communicated = **transmitted**
- A **verbal or written** message
- A **system** (phones, computers) for transmitting or exchanging information
- Personnel **engaged** in communicating
- A **technique for expressing** ideas effectively (as in speech)

Communicare: Lat., der. di *communis* «**comune**»

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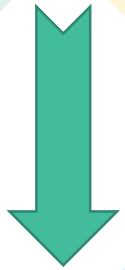
EJP RD Communication “key words”

- **Help** & **Collaboration**
- **ENGAGE** with INTERNAL & EXTERNAL STAKEHOLDERS



- **MAXIMIZE** the EJP RD **IMPACT**
- **Leadership position** for the EJP RD

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EJP RD Communication “outputs”: Website

Implementation on going:

- New NLs registration section by July
- Central Helpdesk by September
- Pillar descriptions (activities, members, etc.) by December

How?

- Collaborating with **you** to collect the info
- Engaging with the EJP RD community

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EJP RD Communication “outputs”: External NLs

Increase the registrations of 1,000 units by December

How?

- Establishing external networks with other partners (e.g. ERA-Learn, EU-Openscreen, Camridge Rare Diseases)
- Press releases
- Use EJP RD twitter account and my social media connections

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EJP RD Communication “outputs”: Twitter

Increase the Followers

- Doubled up the number of followers in less than a month
- Reach 1,000 followers by December 2019

How?

- Tweeting or Re-Tweeting 3-4 times per week
- Connecting with others (following them)
- Establishing professional connections with Comm. or Project managers in the RD, science, media, etc.

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Structuring the EJP RD Network

Internal

- Task force in Comm.

Teddy, c4c, Eurordis,

Orphanet, ERNs

- Establish Comm. References

per each Pillar (?)

Outside

Comm. Managers of

other stakeholders at EU

and international level

- Reaching out media

(interviews, podcast, etc.)

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Process of Communication

- Podcast recording with DNA Radio (US)
- Interview with Larry Luxner, journalist at Bionews Services
- Editorial or news –article- at Horizon Magazine

Plus

- Partnership with Cambridge Rare Disease for the #RareSummit2019, Researcher Radio, and SciLifeLab
- Rare Disease Film Festival partnership (finalize it by July)

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What we can provide

- Templates (power point presentation and poster) and our logistic support
- Webinars account to create EJP RD webinars in collaboration with you
- Share on our website webinars done by EJP RD partners or by externals but with contents related to rare diseases

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What we would like to have from you

- To be informed about conferences you will be going to represent EJP RD:
    a. Final « yes » to presentations/posters
    b. Keep your presentations/posters for our internal record
- Have pics and short descriptions of the EJP RD training courses, summer schools, etc. (TBS on our website and twitter)
- Twitter: Tag @EJPRareDiseases and use #ejprd

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EJP RD Communication “Brainstorm”

- #WomenInScience or the #menels (social media campaigns)
- Collaborate with Eurordis to engage with Patient (host them on our website?)
- EJP RD Rare Disease Day campaing (Feb. 28th 2020)
- EJP RD videos by lobbying with film schools
- EJP RD vignettes by collaborating with artists

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EUROPEAN JOINT PROGRAMME RARE DISEASES

IRDiRC

INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM

EJP RD

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Thank you
&
Happy Summer!

EUROPEAN JOINT PROGRAMME
RARE DISEASES

Questions or comments?

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Interaction with IRDiRC

Interaction with IRDiRC

Current status of interaction:

- EJPRD follows IRDiRC recommendations to contribute to its goals
  - No EJP RD independent Scientific Board
  - IRDiRC chair & vice-chair are part of the EJP RD Policy Board
  - EJP RD coordination office integrates the Scientific Secretariat of IRDiRC
  - Contribution to the implementation of IRDiRC Roadmap and Task Forces
    - Alignment of the Roadmap implementation agenda with the EJP RD Annual Work Plans
    - Proposition of EJP RD experts for the TF

Current status of official representation:

- EJP RD is not represented in IRDiRC as member of any of the constituent committees (Funders/Companies/Patient Organisations)
- At present the group of funders is represented by E-Rare-3 (based on the E-Rare-3 funding commitment, not exhaustive in terms of funders involved in the EJP RD; not representing EJP RD Pillar 1)
- Some countries have double representation in IRDiRC Funders Committee in the name of their national RD spending (DE, FR, IT, NL, ES) and as part of the E-Rare-3 group of funders (via their commitment to the transnational calls)
- INSERM applied to be partner of IRDiRC but as an independent institution

Next steps:

- How do we want to engage/represent EJP RD? → as a whole? Only Pillar 1?
- Any kind of "innovative" model of engagement we can propose
- Who should represent the EJP RD?
- ???

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Interaction with other important stakeholders

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RARE DISEASES

Interaction with other stakeholders/projects

Who are the stakeholders?

- GA4GH → EJP RD is GA4GH driver project
- 1+MG → EJP RD was invited to join 1+MG Working Group on RDs (WG8 – use cases RD) & as potential stakeholder for the CSA application that will accompany 1+MG project
- C4C → EJP RD is connected to C4C through the implication of ULiv (monitoring/KPI/communication activities)
- Rare 2030 → some EJP RD partners involved; currently no formalised collaboration
- EIT Health → contact established
- Other?

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Interaction with other stakeholders/projects

How do we want to work with them?

- Formal invitation to join the EJP RD ExCom meetings/TCs through a dedicated discussion session
- Engagement of EJP RD experts in the WG of GA4GH and 1+MG
- Formalised participation of the EJP RD members in the specific boards of other projects
- Extended communication & share of information
- ????

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How to work with non-EJP RD partners?

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How do we want to work with the non-EJP RD partners?

Floor open for comments from ExCom members

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Central Helpdesk

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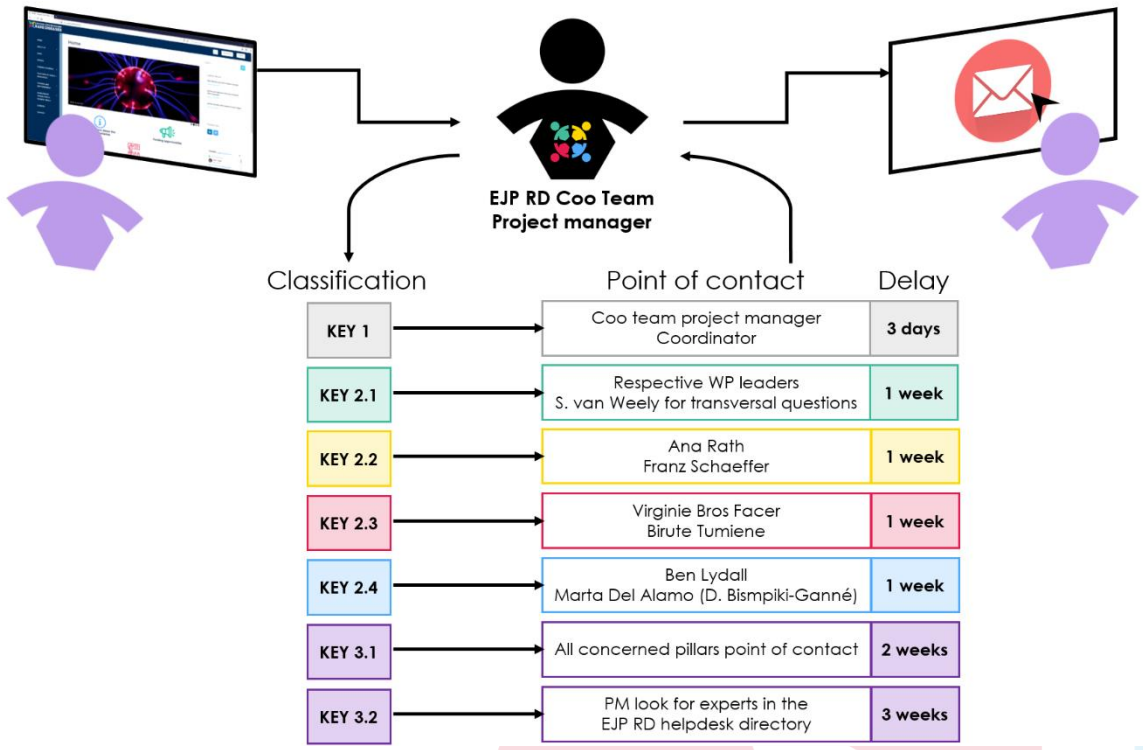
RARE DISEASES

EJP RD Helpdesk



In addition of the general incoming requests, the Helpdesk should also include:

- Pillar 2: tool allowing ERNs to ask questions and see previous question asked (+answers)
- Pillar 4: EJP RD Clinical Study Support Office

➔ A ticketing tool might be needed to follow all emails, timelines and store answers.

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Financial questions

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RARE DISEASES

Financial questions

- Travel budget
- Shifting of budget/PMs from 1 year to the other
- Shifting of budget from 1 partner to the other due to over take of activities
- Shifting of budget from 1 partner to the other due to new activity or replacement of activity by a new one

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Sustainability of the EJP RD activities

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Sustainability of the EJP RD activities

- Training
- Tool boxes
- Tools/services created by Pillars (especially P2 & P4)
- ???

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EJP RD & ERNs common agenda

Franz Schaefer

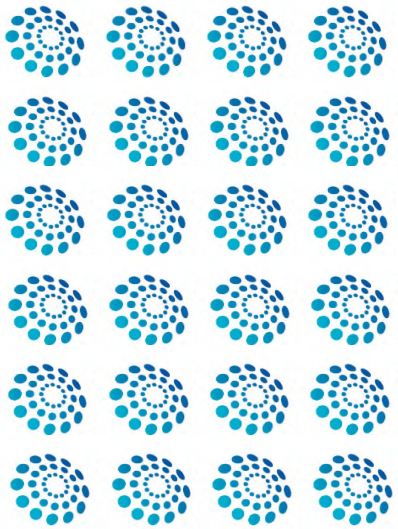
EUROPEAN JOINT PROGRAMME RARE DISEASES

European Reference Networks

EUROPEAN JOINT PROGRAMME RARE DISEASES

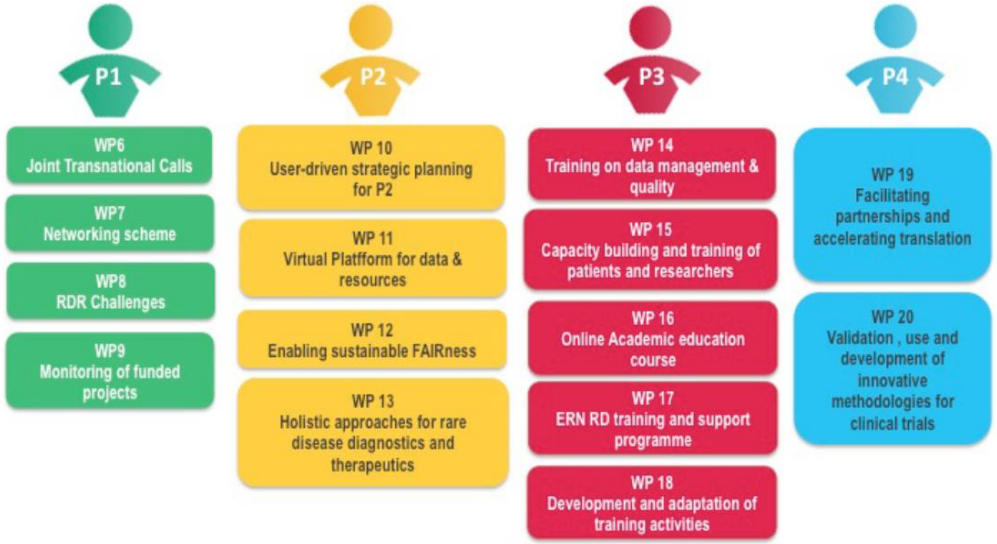
ERNs

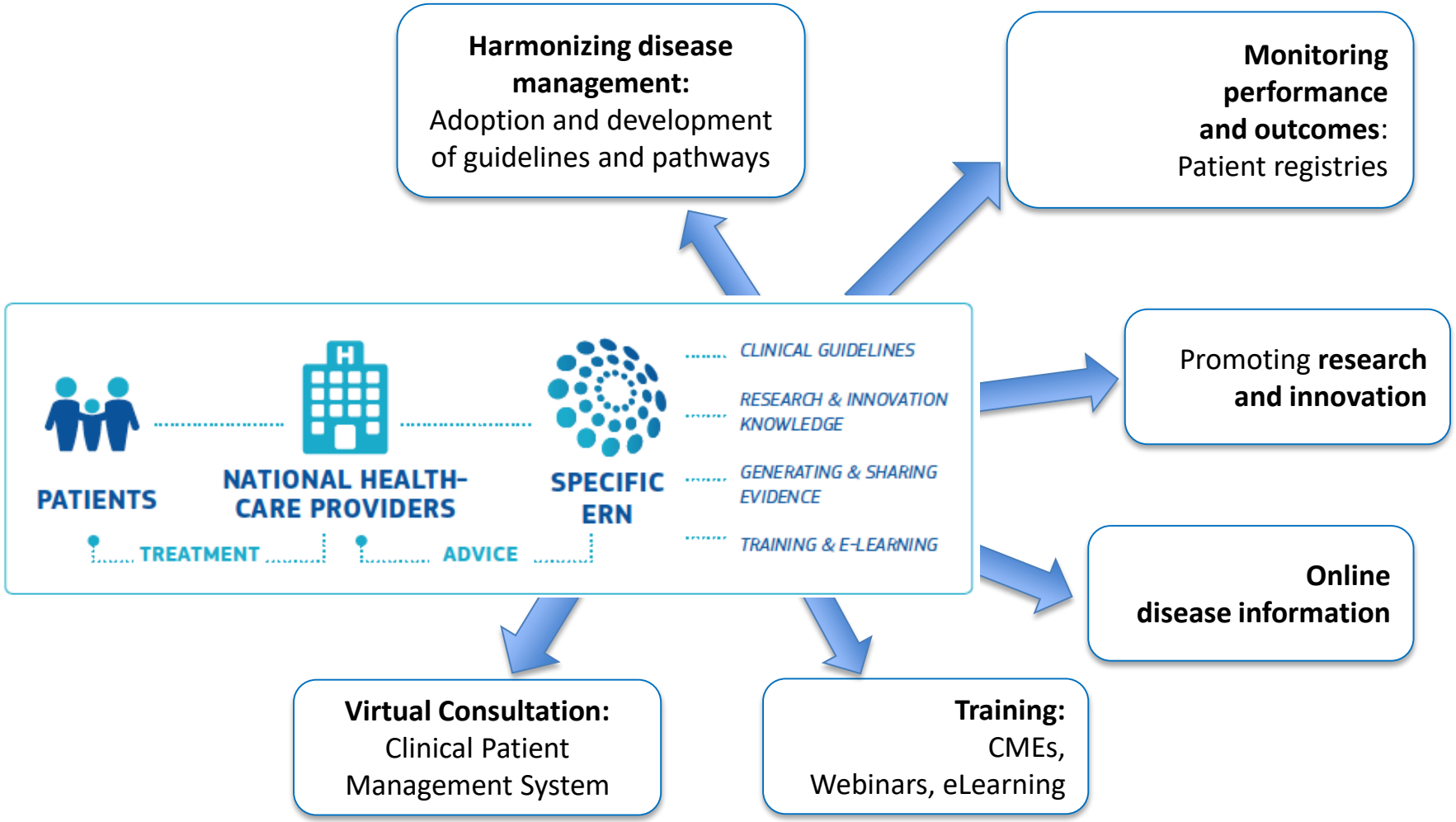
1 mio RD patients



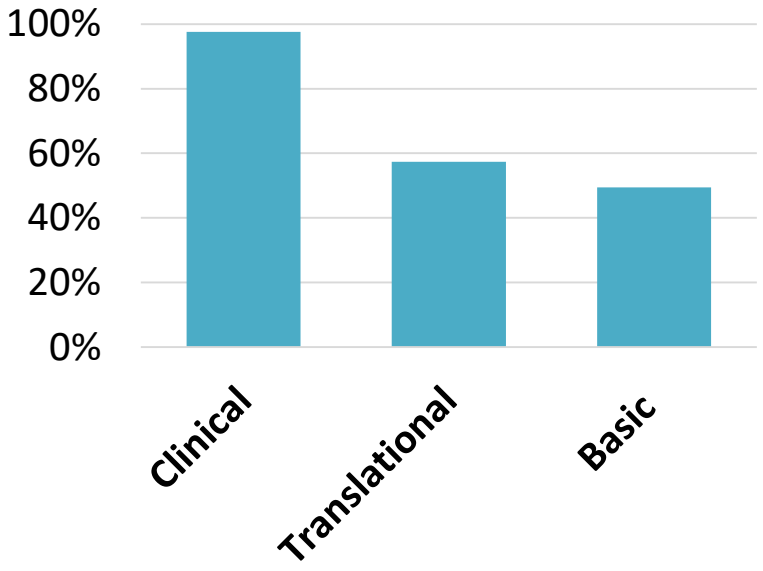
EJP-RD

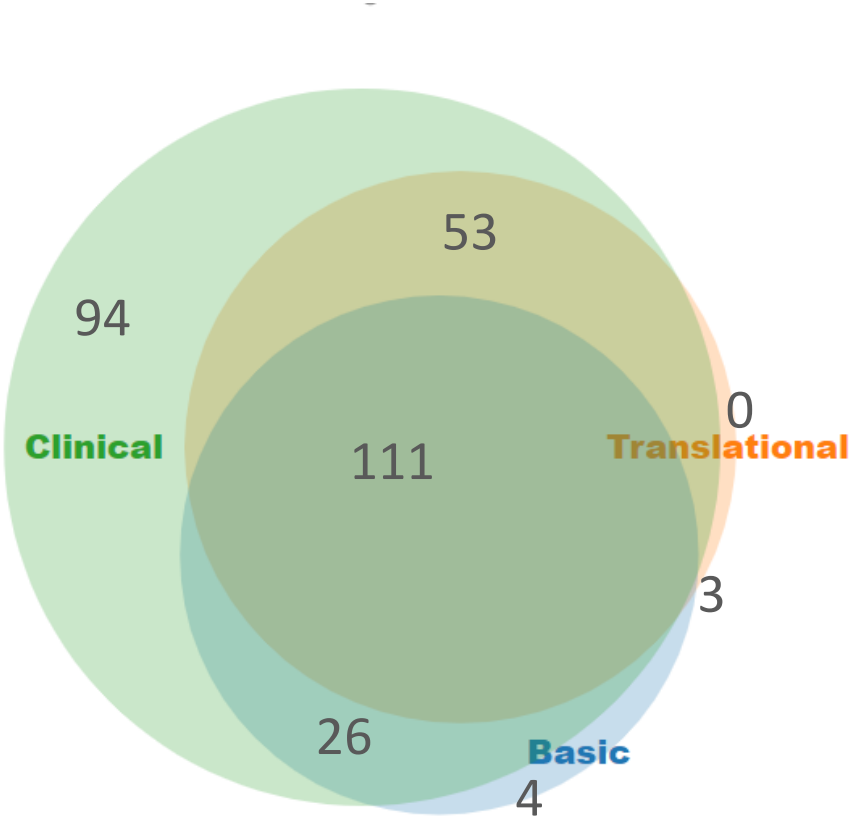
>100 mio € research funds, >100 research groups



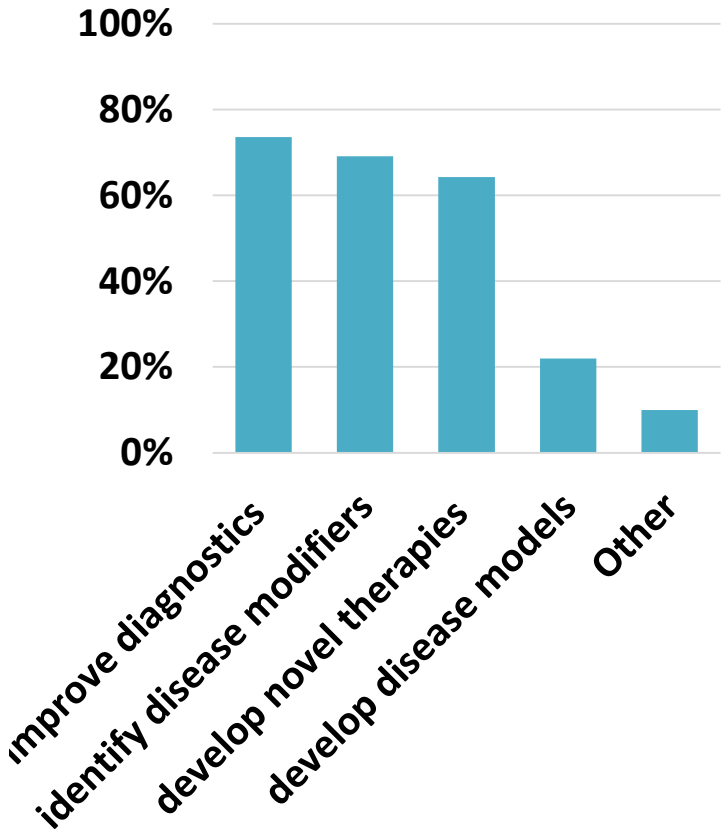

Harmonizing disease management: Adoption and development of guidelines and pathways
Monitoring performance and outcomes: Patient registries
Promoting research and innovation
Online disease information
Virtual Consultation: Clinical Patient Management System
Training: CMEs, Webinars, eLearning
PATIENTS
NATIONAL HEALTH-CARE PROVIDERS
SPECIFIC ERN
CLINICAL GUIDELINES
RESEARCH & INNOVATION KNOWLEDGE
GENERATING & SHARING EVIDENCE
TRAINING & E-LEARNING
TREATMENT
ADVICE

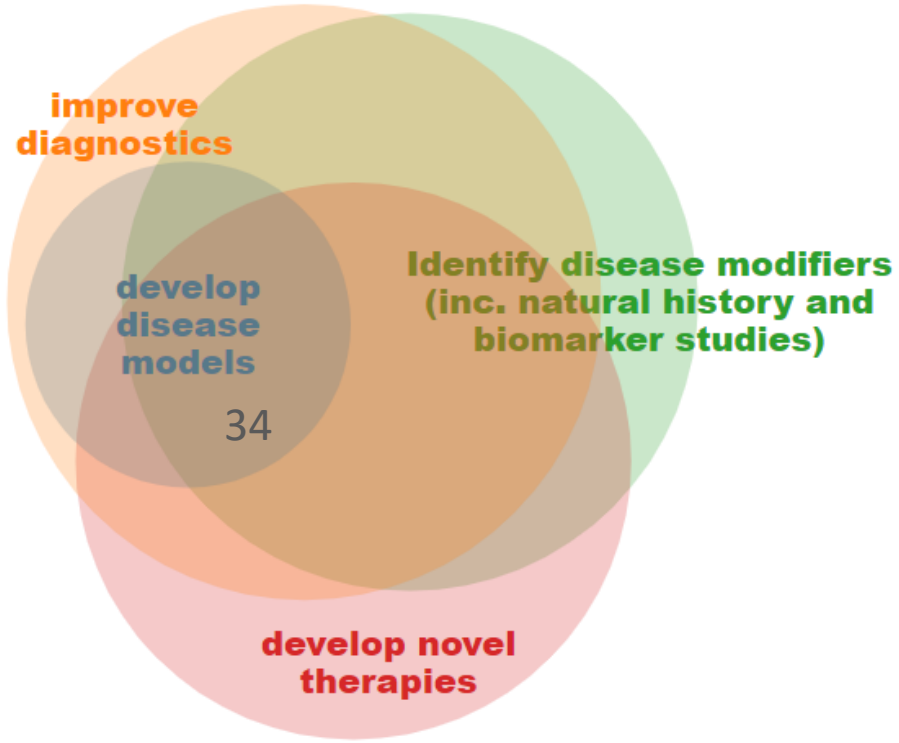
Types of research in ERNs





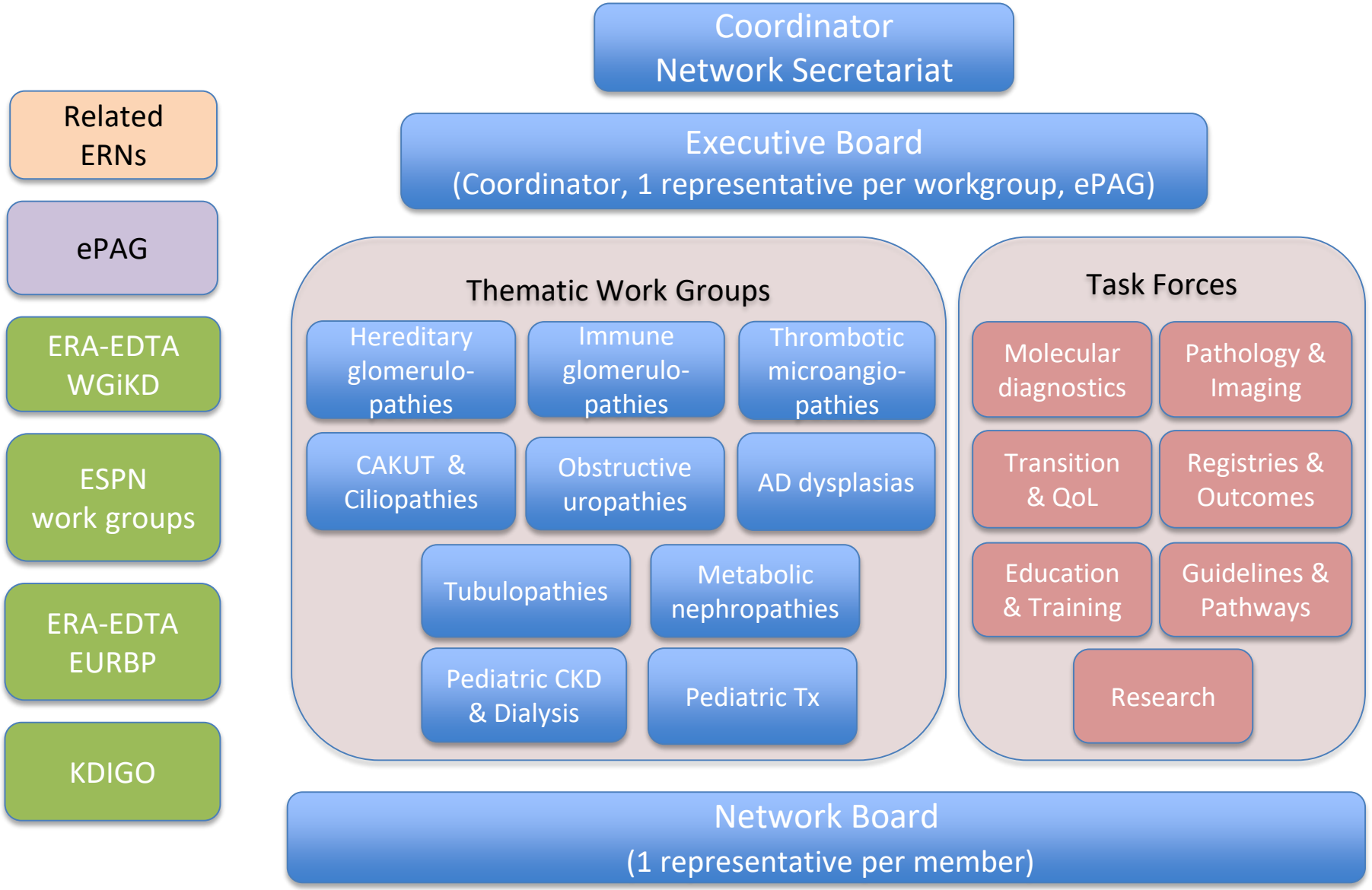
Main purposes of research in ERNs



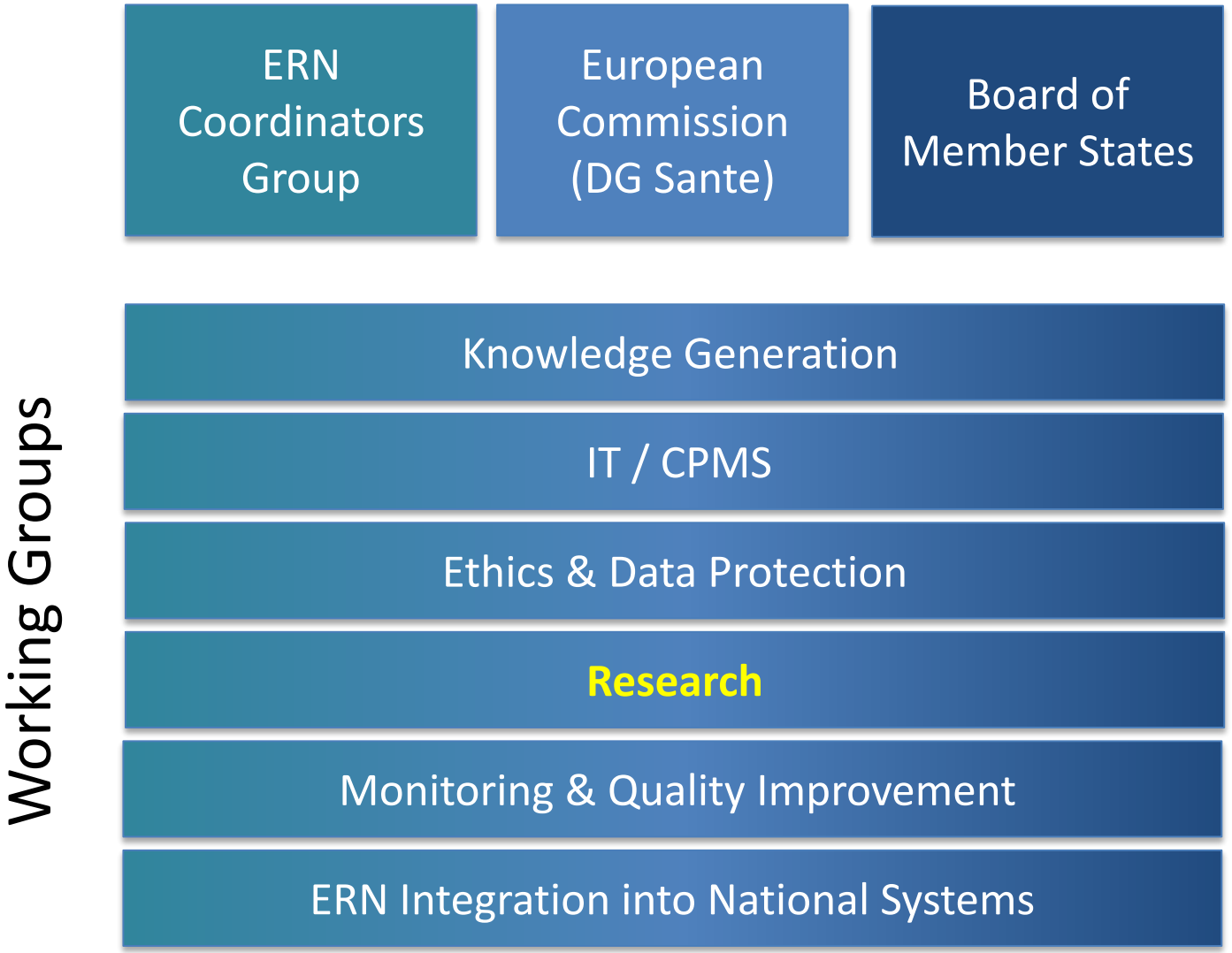


ERKNet European Rare Kidney Disease Reference Network

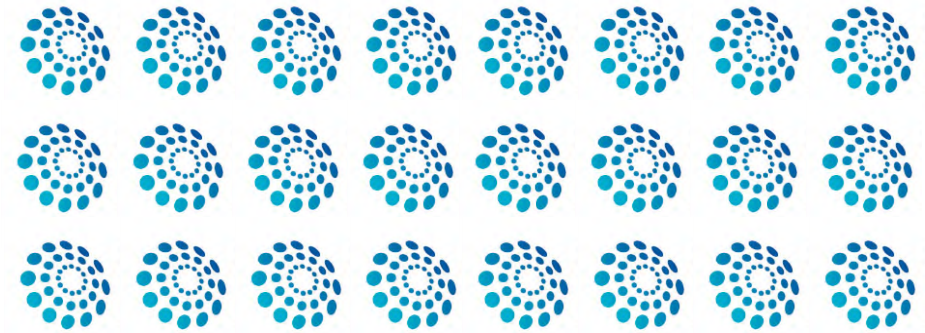
European Reference Networks



ERN Governance



European Joint Programme on Rare Diseases



WG Research

Functions/Role:

Advocacy

Coordination

Facilitation

Information flow

Activities:

Monthly TCs

EMA Clinical Trials Workshop 6/18

Research priorities meeting 1/19

via EJP membership

Relay research information to ERN members

ERN Research Working Group

Chairs:
Alberto Pereira (ERN Endo), Luca Sangiorgi (ERN BOND),
Eduardo Lopez Granados (TransplantChild), Franz Schaefer (ERKNet)

EC liaison: Helene Le Borgne

BoMS liaison: György Pflugler (Hungary)

EMA liaison: Luca Sangiorgi

EJP-RD liaison: Franz Schaefer

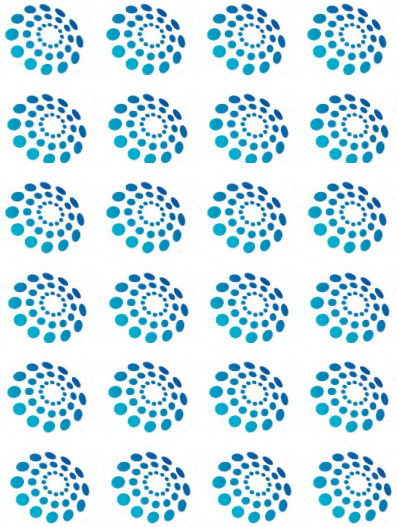
Registry Task force: Eduardo Lopez Granados

European Reference Networks

EUROPEAN JOINT PROGRAMME RARE DISEASES

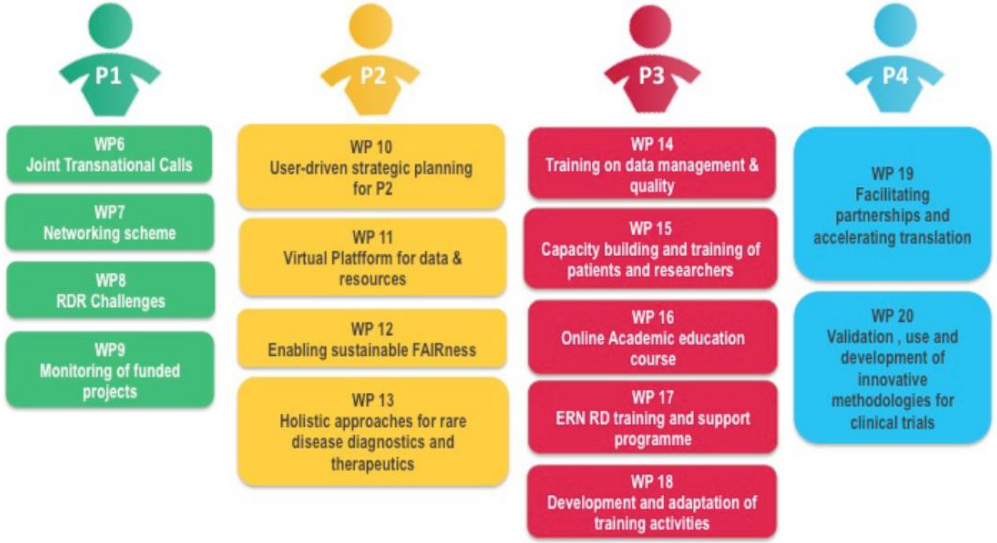
ERNs

1 mio. RD patients

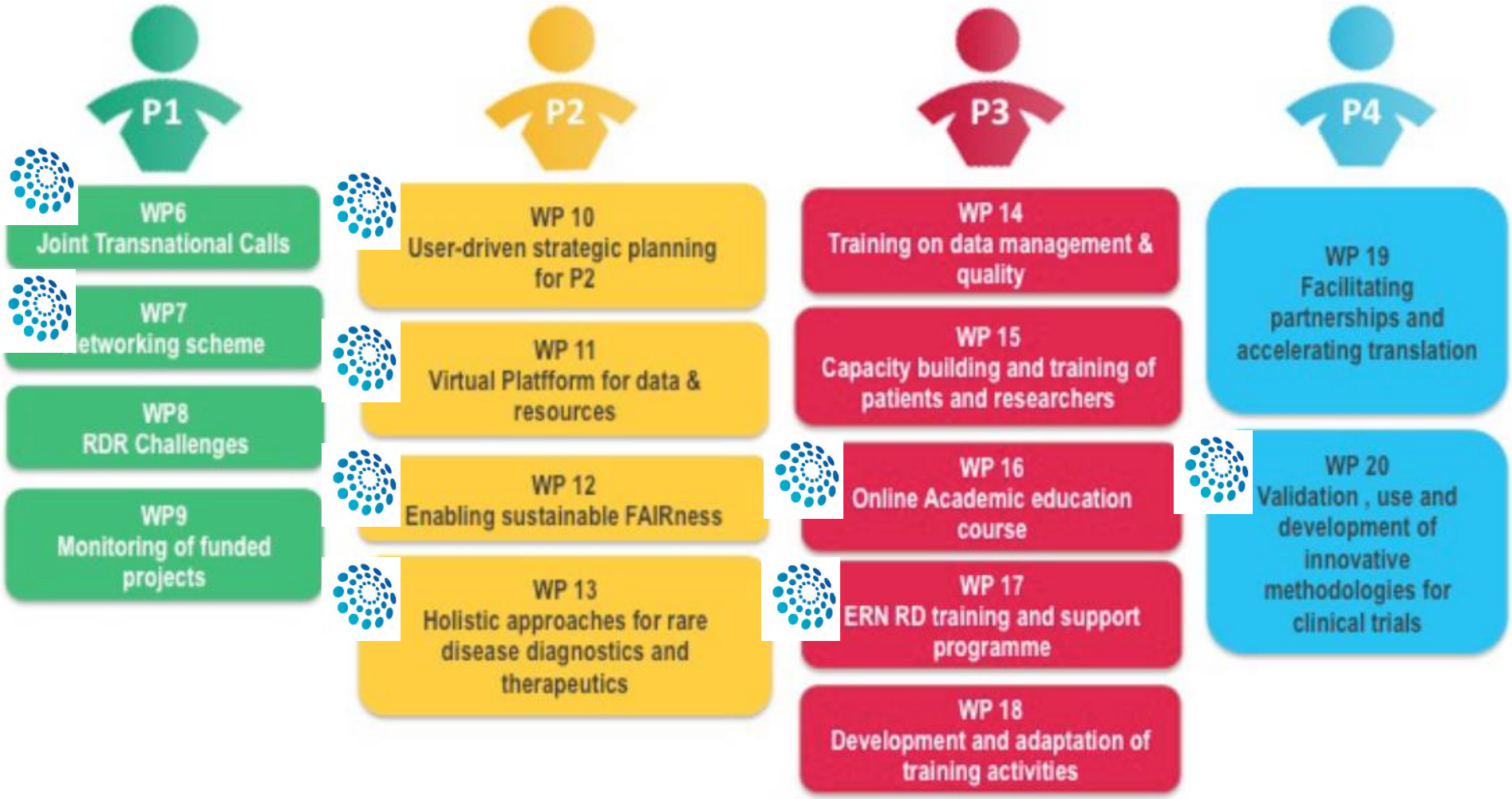


EJP-RD

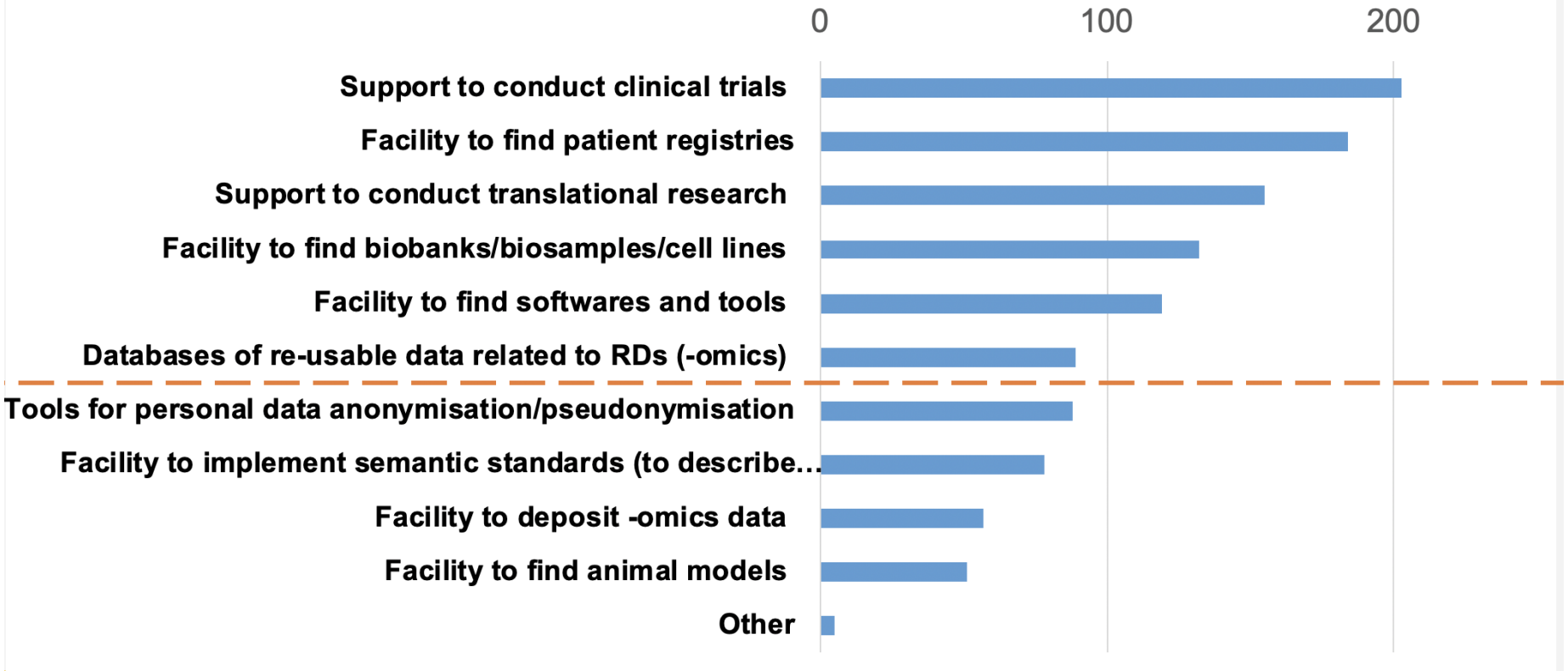
>100 mio €, >100 research groups



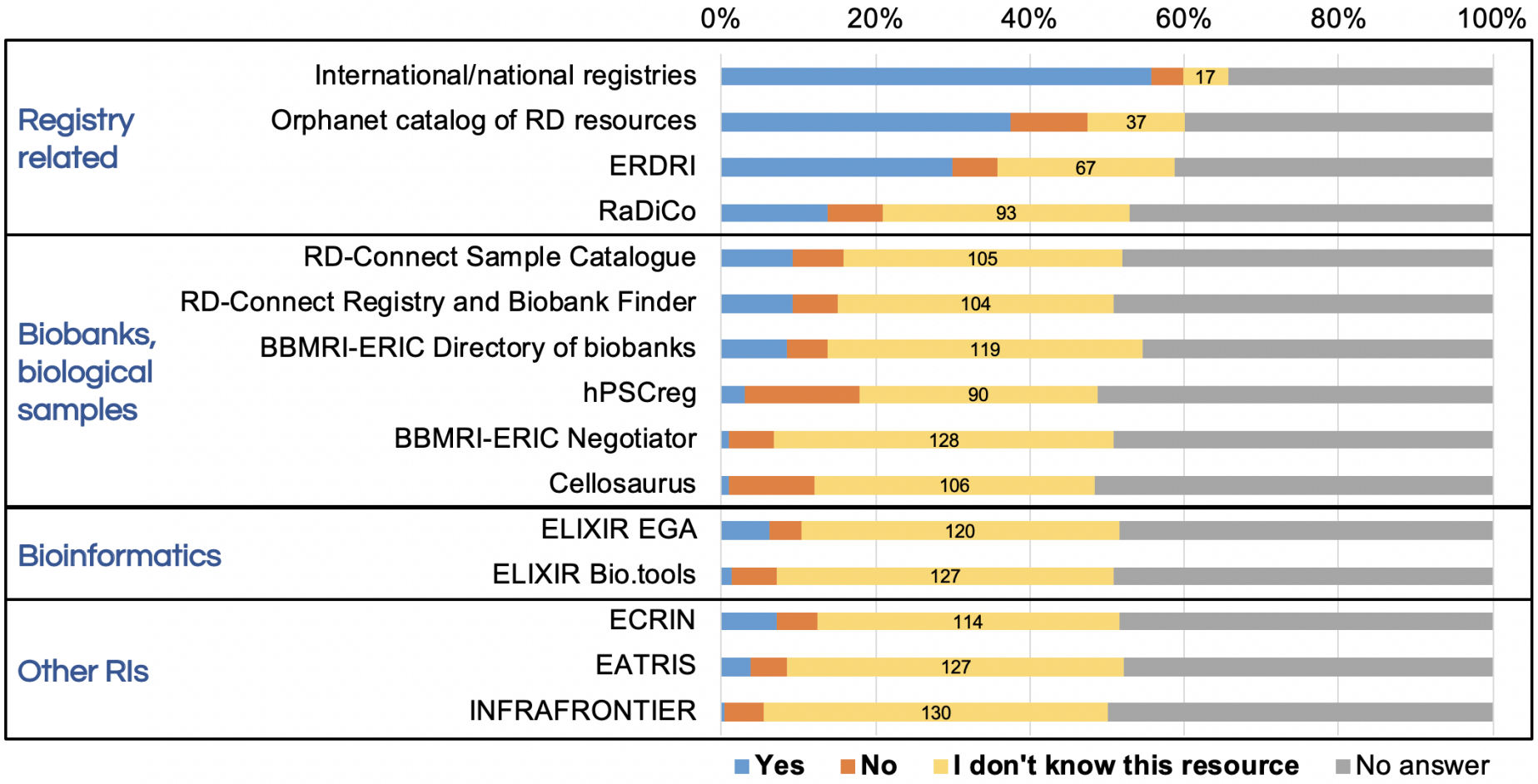
European Joint Programme on Rare Diseases



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



Which of these research infrastructure & resources are of utmost importance for your research?



Please describe your interest in making your research data FAIR?

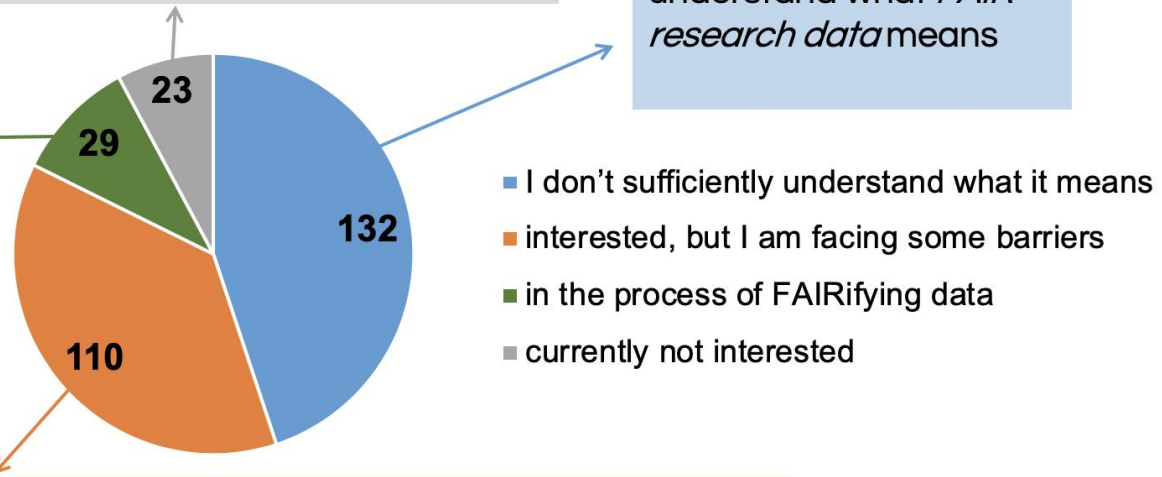
What could raise your interest?

50%	Seeing any added value
50%	Having the advantages outweigh the drawbacks
Other	NO TIME

45% of respondents feel they do not sufficiently understand what *FAIR research data* means

In what stage is your FAIRification process?

47% Initiation
24% Planning
26% Execution
1 case: Close (completed)



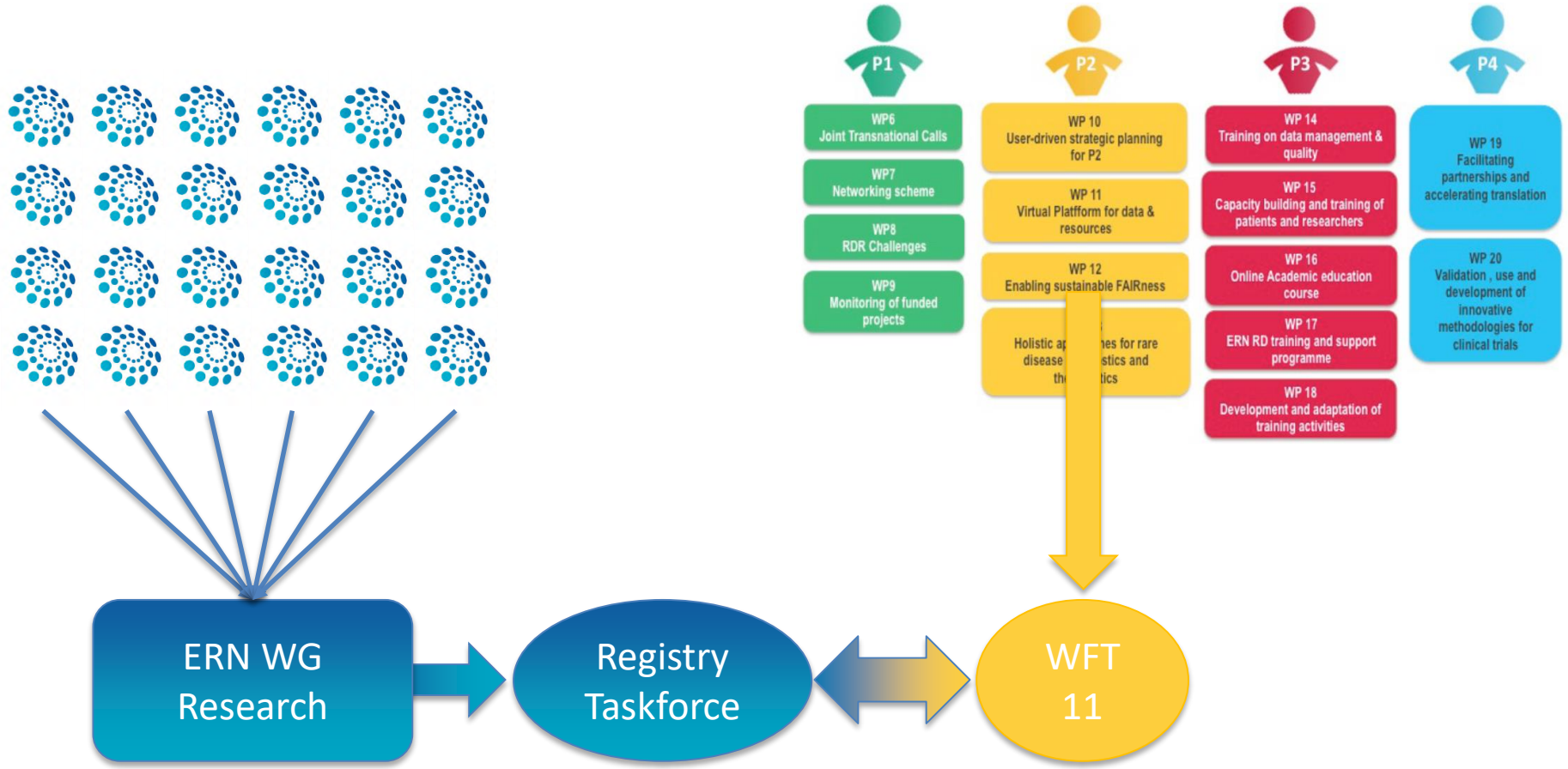
What are these barriers?

28%	No resources available for this effort
18%	No software or servers to make the data discoverable/sharable
17%	No consent/authority to make my data more widely accessible
7%	Don't know what standards to use
Other	NO TIME

WP12: Registry Collaboration

ERNs

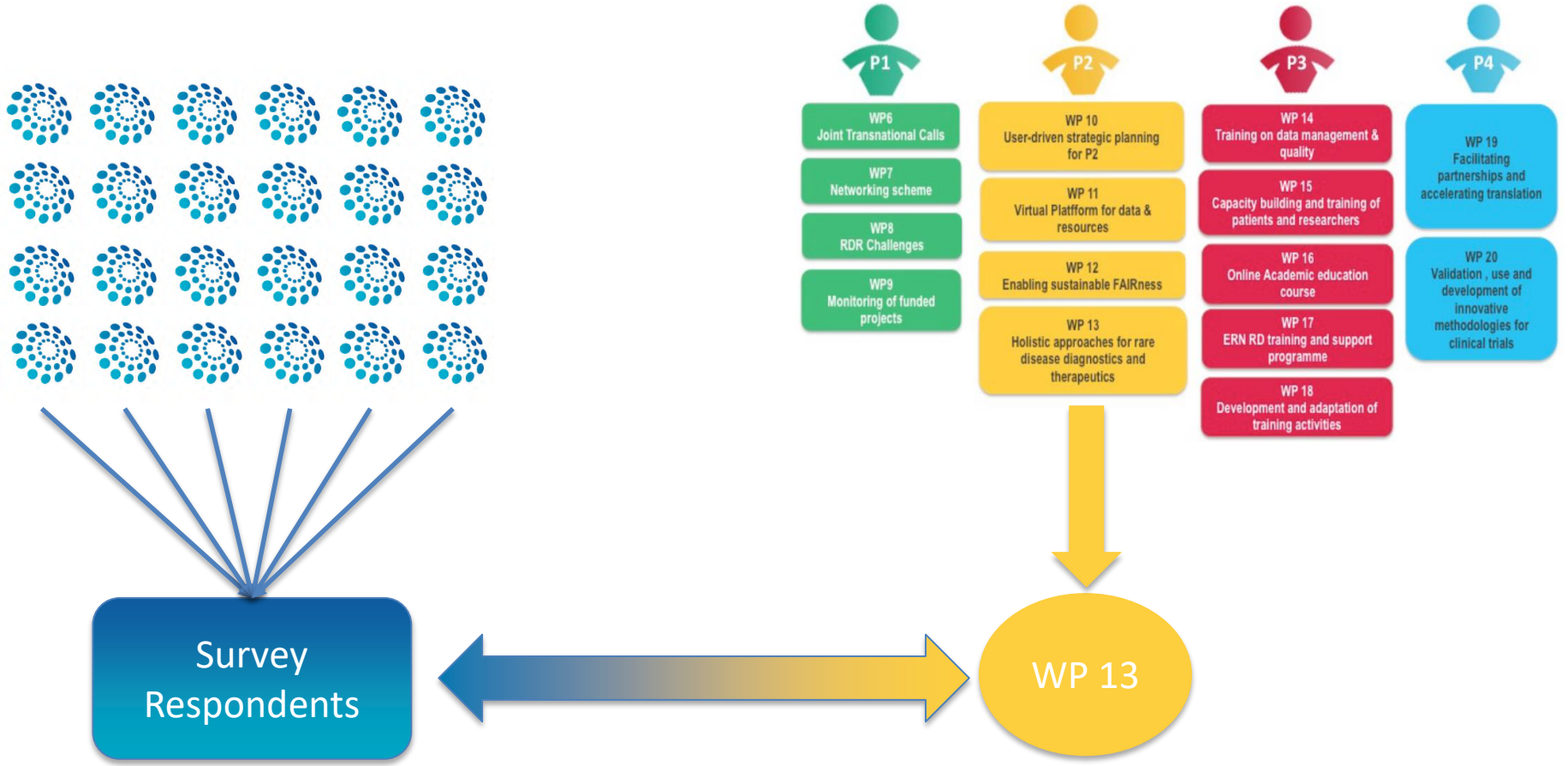
EJP-RD



WP13: Cross-Omics Collaboration

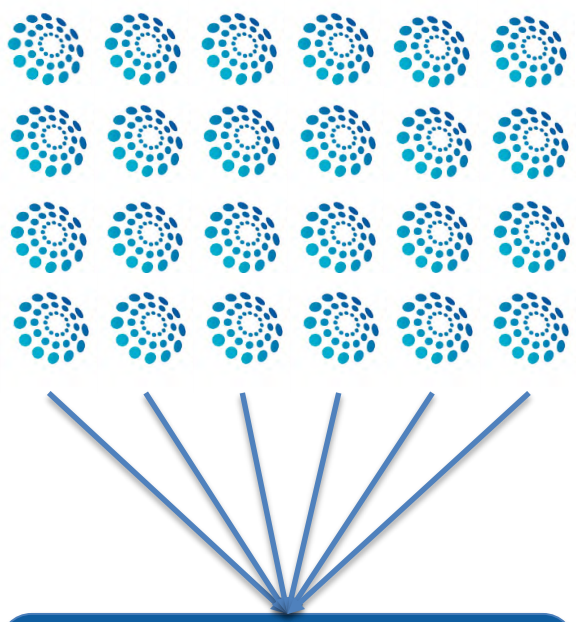
ERNs

EJP-RD



WP17: ERN Research Training Programme

ERNs

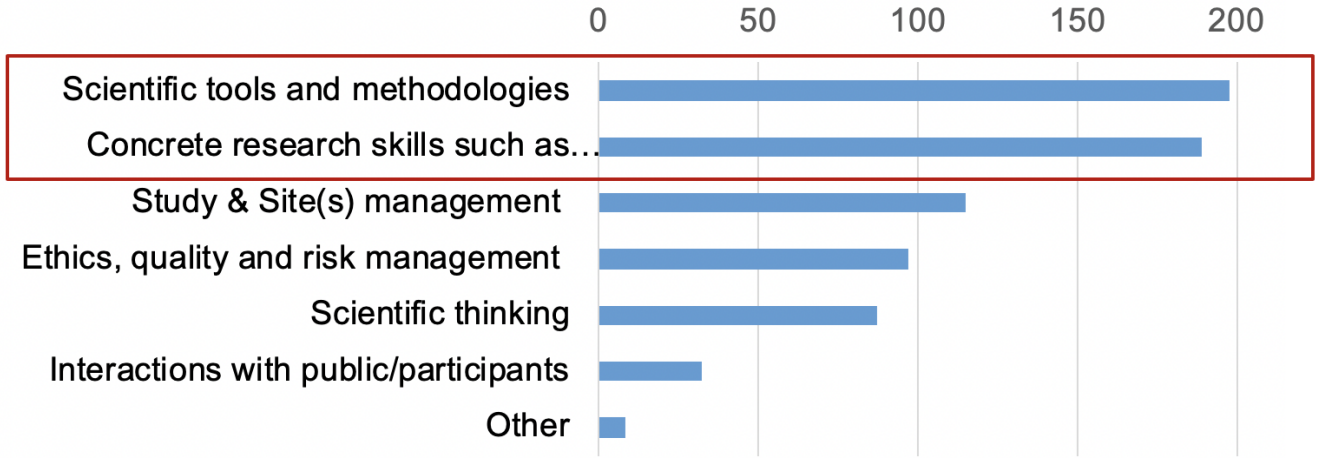


950 ERN units
150 Affiliated Partners

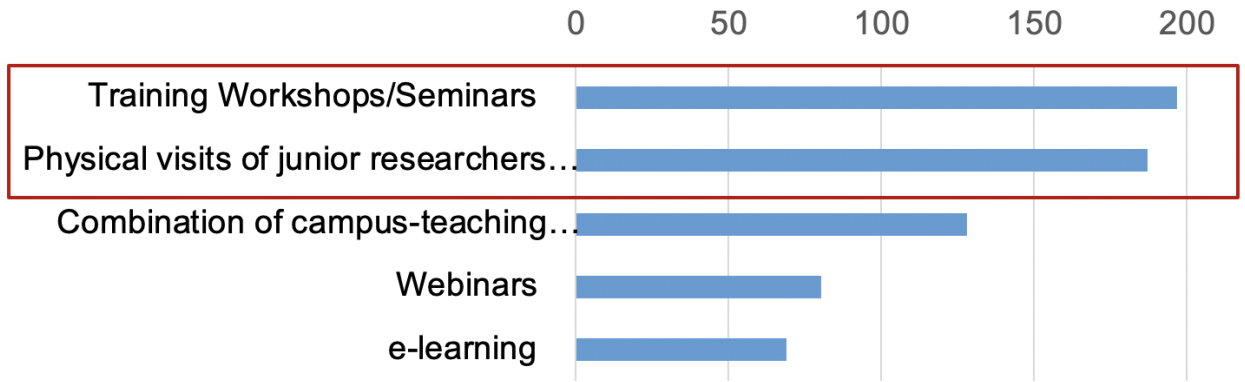
EJP-RD

WP 17

F3. What, in your view, are the most important research skills training domains that need to be addressed to help ERN HCPs raise the level of their research?



F4. Which of the following types of training measures would address these domains most efficiently for your group?



WP 17: ERN Research Teaching and Training Programme

Focus group meeting: June 26, Leiden

WP 17: ERN Research Teaching and Training Programme

Focused training seminars/workshops for ERN researchers

- Targeted budget: 1 mio. €
- Up to 45 workshops
- Interactive, 1.5 days
- 20-30 attendees by application
- Topic proposals by ERN and EJP investigators, 6-monthly deadlines
- Technical guidelines for local organizers tbd
- Pre/post-workshop evaluation

Short-term exchange visit programme for ERN researchers

- Targeted budget: 0.6 mio €
- 300 person months
- Duration: 2 weeks to 3 months
- Early-stage (≥2nd-yr) researchers
- Application: Research plan, added value to researcher and ERN
- Host and/or sending institution ERN-linked
- Priority: ITC residents, females, new collaborations
- Report within 1 month of completion

Anticipated programme start: Q4 2019

WP20: Clinical Study Support Office Demonstrator Projects

1) **Support Office for design and planning of clinical trial**

2) **Demonstration projects on existing statistical methodologies for clinical trials in RD:**

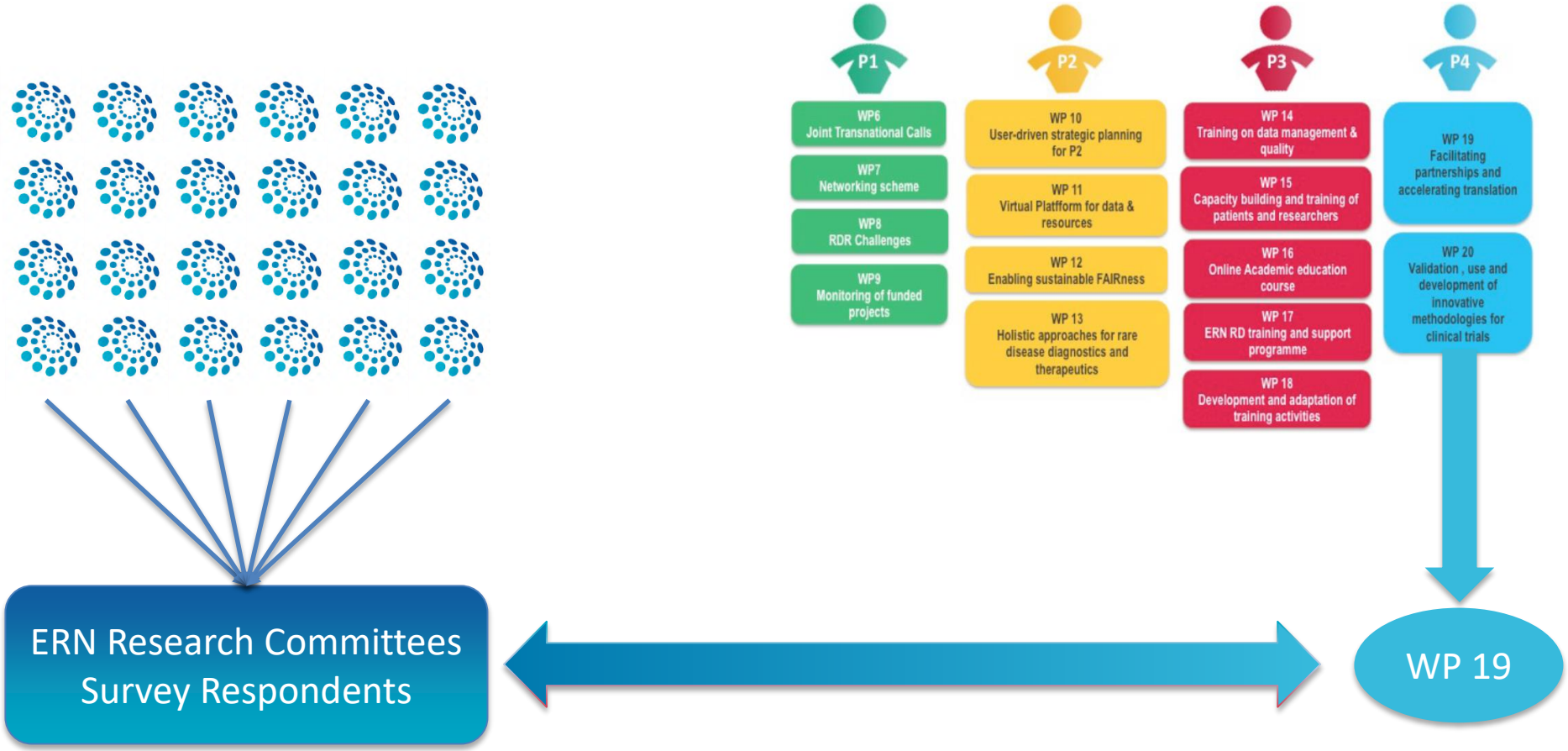
6 small scale demonstration projects

- to provide proof of efficacy of innovative trial methodologies

- based on real RD data or running co-funded pilot studies (with industry or private partners)

- preferred source: completed clinical trial data from ERNs

WP20: Clinical Study Support Office
Demonstrator Projects



Annex 2

Slides presented during the EJP RD Policy Board meeting

EUROPEAN JOINT PROGRAMME ON RARE DISEASES

Policy Board Meeting

4th of July
Brussels, Belgium

EUROPEAN JOINT PROGRAMME
RARE DISEASES

9:00 – 9:30	Welcome word and introduction to the EJP RD	Daria Julkowska (Inserm) Coo
9:30 – 10:00	Round table	All
10:00 – 10:30	Pillar 0 – Coordination and transversal activities : *Presentation of achievements (M1 – M7) and of activities planned in the Annual Work Plan Year 2 with Policy Board [25min] + immediate questions [5min]*	Daria Julkowska (Inserm)
10:30 – 10:50	**Coffee break**	
10:50 – 11:20	Pillar 1 – Collaborative research funding : *Presentation of achievements (M1 – M7) and of activities planned in the Annual Work Plan Year 2 with Policy Board [25min] + immediate questions [5min]*	Ralph Schuster (DLR) and Sonja van Weely (ZonMw)
11:20 – 11:50	Pillar 2 – Innovative coordinated access to data and services for transformative rare diseases research : *Presentation of achievements (M1 – M7) and of activities planned in the Annual Work Plan Year 2 with Policy Board [25min] + immediate questions [5min]*	Ana Rath (Inserm) and Franz Schaefer (UKL-HD)
11:50 – 12:20	Pillar 3 – Capacity building and empowerment : *Presentation of achievements (M1 – M7) and of activities planned in the Annual Work Plan Year 2 with Policy Board [25min] + immediate questions [5min]*	Virginie Bros-Facer (EURORDIS) and Biruté Tumiene (VUHSK)
12:20 – 13:20	**LUNCH**	
13:20 – 13:50	Pillar 4 – Accelerating the translation of high potential projects and improving outcomes of clinical studies in small populations : *Presentation of achievements (M1 – M7) and of activities planned in the Annual Work Plan Year 2 with Policy Board [25min] + immediate questions [5min]*	Rima Nabbout (APHP) and Anton Ussi (EATRIS)
13:50 – 14:50	Open discussion of activities planned in the Annual Work Plan Year 2 with Policy Board	All
14:50 – 15:10	**Coffee break**	

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15:10 – 16:10	Prioritization strategy process (indicators and methodology) – *Presentation and discussion with PB*	Eva Bermejo and Manuel Posada (ISCIII)
16:10 – 17:00	Stakeholders needs and challenges	Policy Board members round table
17:00 – 17:20	EJP RD under Horizon Europe	All
17:20 – 17:30	Summary & Next steps	Daria Julkowska (Inserm) Coo

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INTRODUCTION TO EJP RD

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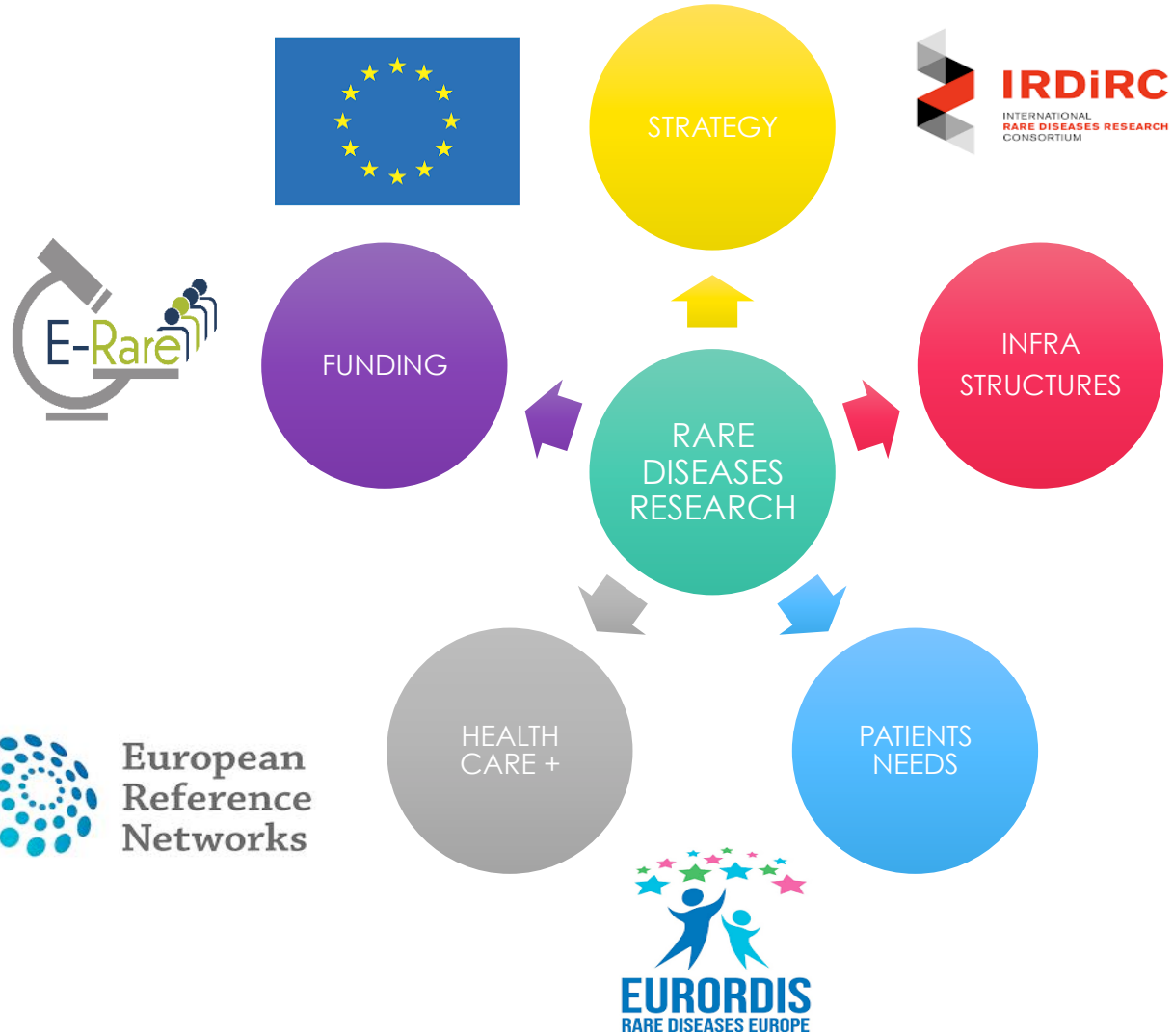
RARE DISEASES

Rare Diseases Landscape in Europe



IRDiRC
INTERNATIONAL RARE DISEASES RESEARCH CONSORTIUM

INFRAFRONTIER
mouse disease models

RD Connect

ePTRI
European Paediatric Translational Research Infrastructure

orphanet

BBMRI-ERIC
gateway for health

ECRIN
EUROPEAN CLINICAL RESEARCH INFRASTRUCTURE NETWORK

Joint Research Centre
JRC

EU-OPENSCREEN
Chemical keys for life's locks

elixir

conect 4children
COLLABORATIVE NETWORK FOR EUROPEAN CLINICAL TRIALS FOR CHILDREN

eatris

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Objectives

Main objective:

Create a research and innovation pipeline "from bench to bedside" ensuring rapid translation of research results into clinical applications and uptake in healthcare for the benefit of patients

Specific objective:

Improve integration, efficacy, production and social impact of research on rare diseases through the development, demonstration and promotion of sharing of research and clinical data, materials, processes, knowledge and know-how, and an efficient model of financial support for research on rare diseases

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IRDiRC Goals 2017–2027

Released 9 August 2017

VISION: *Enable all people living with a rare disease to receive an accurate diagnosis, care, and available therapy within one year of coming to medical attention*

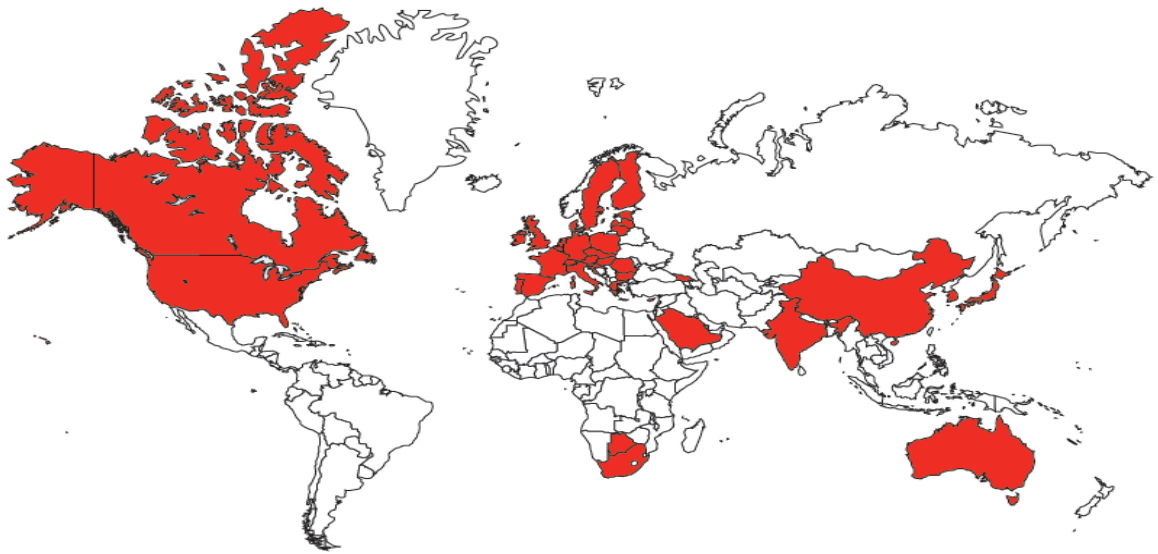
GOAL 1: All patients coming to medical attention with a suspected rare disease will be diagnosed within one year if their disorder is known in the medical literature; all currently undiagnosable individuals will enter a globally coordinated diagnostic and research pipeline.

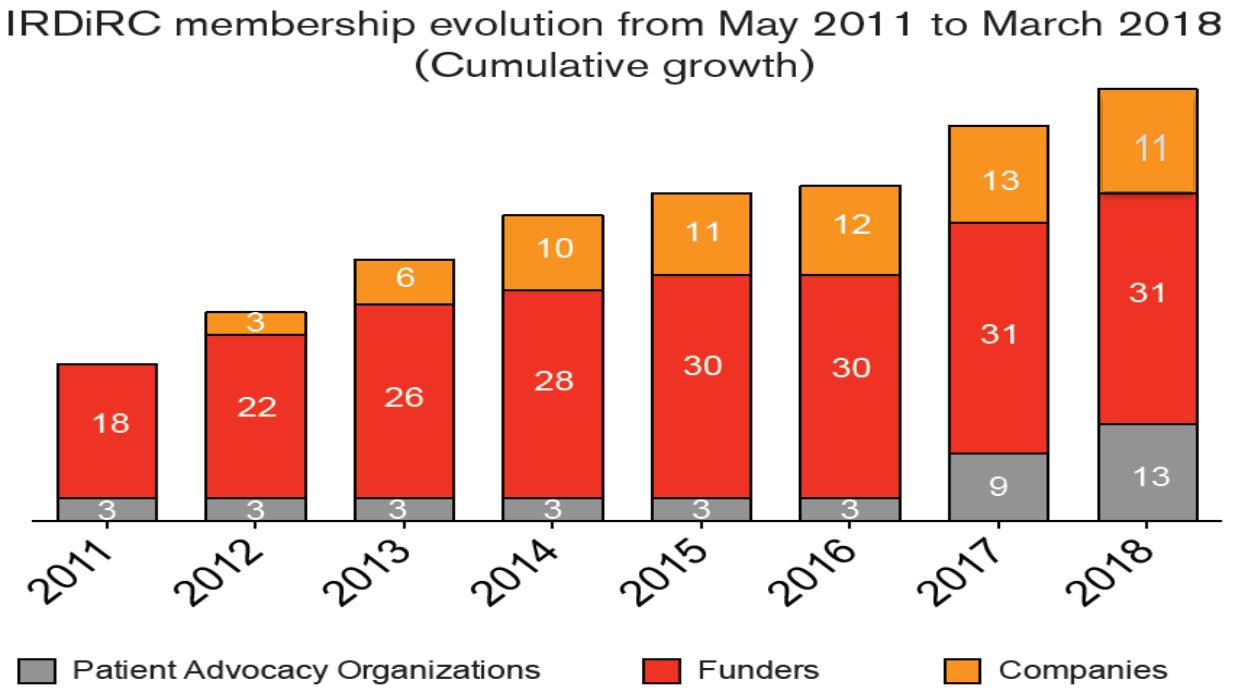
GOAL 2: 1000 new therapies for rare diseases will be approved, the majority of which will focus on diseases without approved options.

GOAL 3: Methodologies will be developed to assess the impact of diagnoses and therapies on rare disease patients.

IRDiRC
INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM

IRDiRC Consortium Assembly
Representation





- 31 funders
- 11 companies
- 13 patient advocates organizations

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CONSORTIUM

IRDiRC Committees
Mission

- Identify roadblocks/priorities
- Implement Task Forces and activities to address priorities/gaps
- Establish and promulgate best practices, operating procedures, quality standards, roadmap to address priorities
- Inform other Committees of scientific and programmatic states, needs, opportunities, emerging issues

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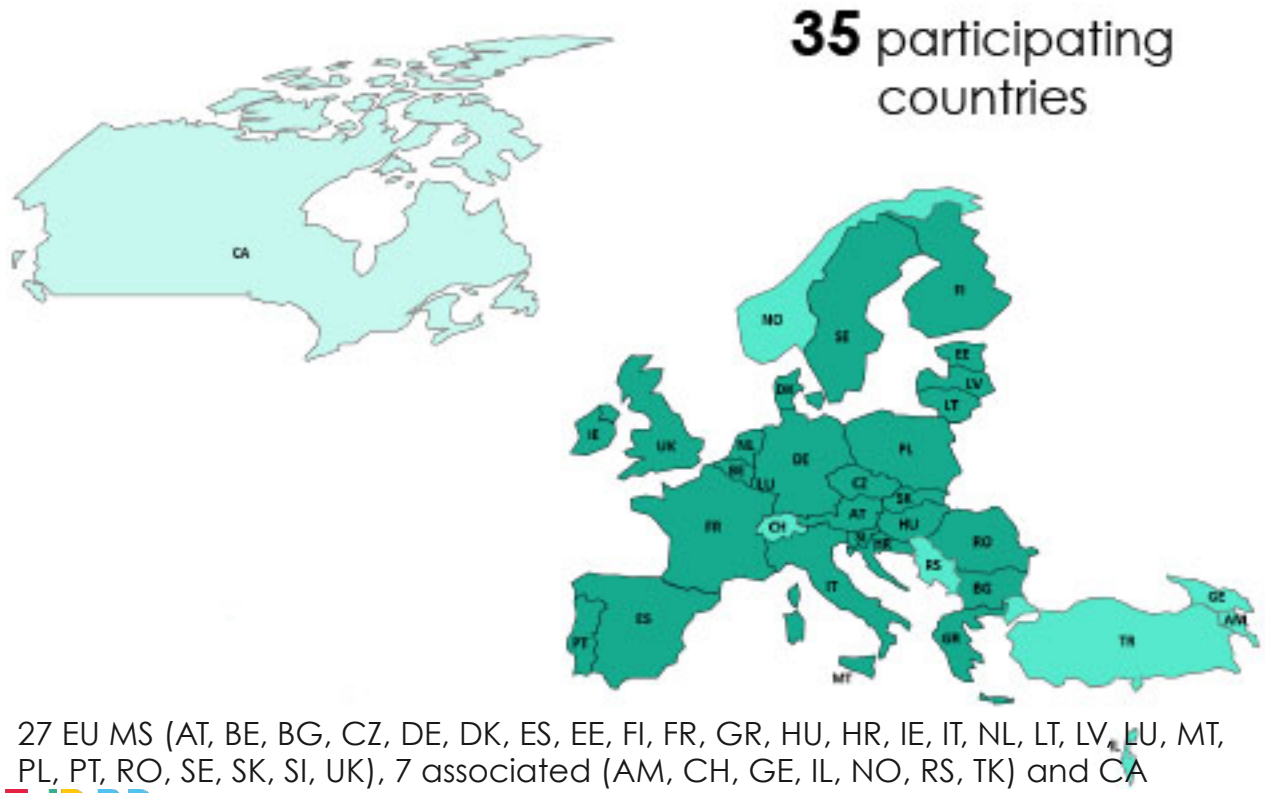
Main facts about the EJP RD

Jan 2019 → **Dec 2023**

Total budget (min. submitted): **101 M€** (→ expected > 110 M€)

Union contribution: ***55 M€*** (70% reimbursement rate)



27 EU MS (AT, BE, BG, CZ, DE, DK, ES, EE, FI, FR, GR, HU, HR, IE, IT, NL, LT, LV, LU, MT, PL, PT, RO, SE, SK, SI, UK), 7 associated (AM, CH, GE, IL, NO, RS, TK) and CA

88 beneficiaries

- 31 research funding bodies/ministries
- 12 research institutes
- 22 universities/hospital universities
- 11 hospitals
- 5 EU infrastructures (BBMRI, EATRIS, ECRIN, ELIXIR, INFRAFRONTIER) + EORTC
- EURORDIS & ePAGs
- 5 charities/foundations (FTELE, AFM, FFRD, FGB, BSF)

+ 50 Linked Third Parties

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EJP RD STRUCTURE

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From science to health

COORDINATION
& TRANSVERSAL ACTIVITIES

INTEGRATIVE RESEARCH STRATEGY

SUSTAINABILITY

ETHICAL & REGULATORY

COMMUNICATION

1 FUNDING

2 COORDINATED ACCESS TO DATA & SERVICES

3 CAPACITY BUIDLING & EMPOWERMENT

4 ACCELERATING TRANSLATION OF RESEARCH & THERAPY DEVELOPMENT

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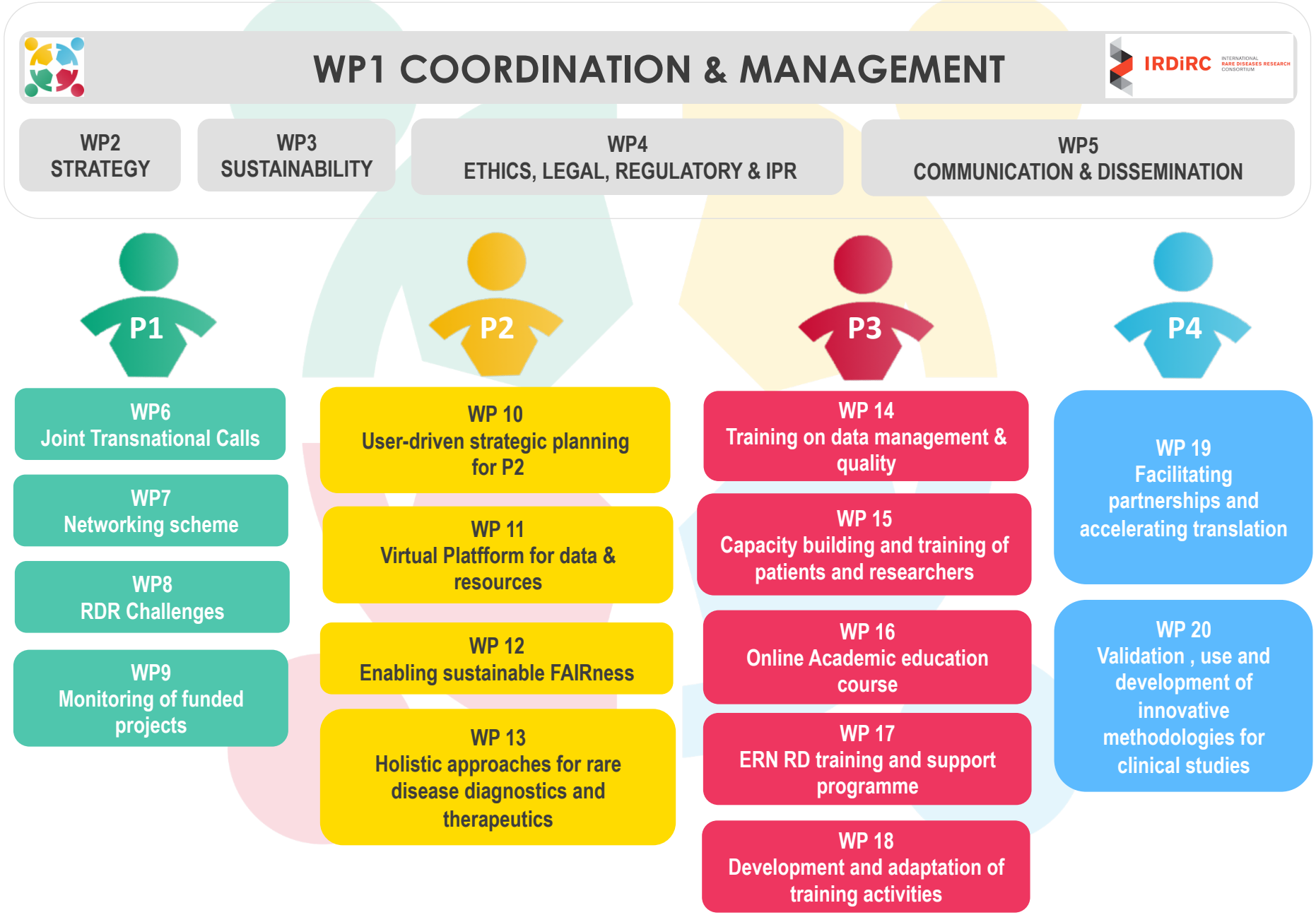
Why together is better than separately?



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Pillar 1: Collaborative research funding

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RARE DISEASES

Pillar 1: Activities

WP6: Joint Transnational Calls for collaborative research projects

Open to research teams from countries with funders involved – min of 4 teams from 4 countries. Patient Advocacy Organisation (PAO) can receive funding. Topics spanning from pre-clinical, translational to clinical research

WP7: Networking to share knowledge on rare diseases

Small support schemes for networking (workshops/events/share of knowledge) – 30 K€ max – open all year long – open to all countries involved in EJP RD

WP8: Rare disease research challenges

Public-private (small scale) partnerships – challenges set by industry (high levle TRL) and validated by EJP RD – short term (max. 18 months) projects - open to all countries involved in EJP RD

WP9: Monitoring of funded projects

Monitoring of all projects funded through EJP RD and previous E-Rare projects

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Pillar 2: Innovative coordinated access to data and services for transformative rare diseases research

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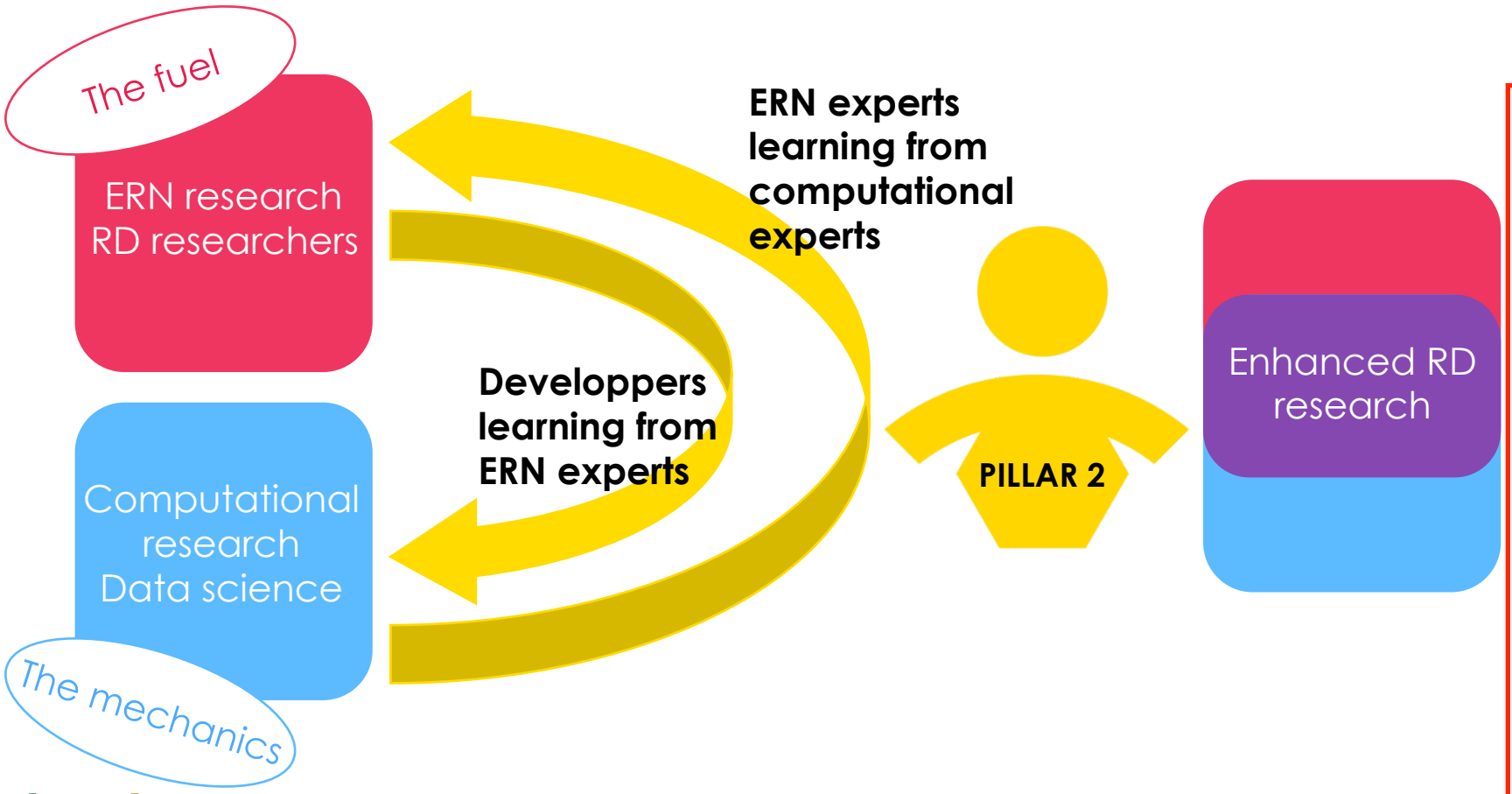
RARE DISEASES

Pillar 2 target: FAIR-based virtual platform



A powerful substrate for translational research:

- **Centralized services for collections (resource-level)**
  - Sample, biobanks, registries, infrastructures and tools catalogue
  - Analysis platform for omics data
  - Curated rare disease-centered information and data
- **Federated services for data elements (record-level)**
  - FAIR 'at source'
  - Data, patients, and samples - linked and discoverable
  - Consents and data use conditions also represented

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Pillar 2: Activities

- **WP10: User-driven strategic planning and transversal activities for Pillar 2 data ecosystem**

Annual strategic meetings with users (ERNs) & developers to define the priorities – coordination of outputs & needs – technical GDPR implementation – quality, sustainability and scaling up

- **WP11: Common virtual platform for discoverable data and resources for RD research**

Metadata & ontological models – FAIR compliance – data deposition & access to data infras – online tools

- **WP12: Enabling sustainable FAIRness and Federation at the record for RD data, patients and samples**

Alignement of core interoperability standards – software for FAIR ecosystem – FAIRification support

- **WP13: Enabling multidisciplinary, holistic approaches for rare diseases diagnostics and therapeutics**

System biology approaches for RD – biological pathways – variants to function – environmental toxicology – treatment drugs - proof of principle studies

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Pillar 3: Capacity building and empowerment

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RARE DISEASES

Pillar 3: Activities

- **WP14: Training on data management & quality**

Orphanet nomenclature – standards & quality of genetics/genomics data in clinical practice – strategies to foster undiagnosed diseases – biobanks sample data management – rare diseases registries & FAIRification at source – European Rare Diseases Registry Infrastructure

- **WP15: Capacity building & training of patients and researchers in rare diseases research and processes**

Expert Patients and Researchers EURORDIS Summer school – scientific innovation and translation research aspects in RDs for patient advocates – leadership & communication skills for patient advocates and representatives – education material and activities for paediatric patients

- **WP16: Online academic education course**

Based on assessed needs of the RD community – in collaboration with universities – 10 to 12 modules with accreditation – e-learning format open to all – Future Learn platform

- **WP17: ERN RD training & support programmes**

Based on four groups (Neuro, Neoplasm & malformation, Organs, Systemic) – preferences, needs and resources of ERNs – tailored for and performed by ERNs

- **WP18: Development and adaptation of training activities**

Evaluation of developing needs according to progress of Pillars 2 & 4 – specific needs of EU 13 countries – emerging needs of ERNs

Pillar 4: Accelerating the translation of high potential projects and improving outcomes of clinical studies in small populations

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RARE DISEASES

Pillar 4: Activities

WP19: Facilitating partnerships and accelerating translation for higher patient impact

Innovation management toolbox – assessment and real time mentoring of translational projects – support in exploitation and follow-on funding – partnering support – roadmap for European investment platform for RD

WP20: Accelerating the validation, use and development of innovative methodologies tailored for clinical trials in RDs

Key Task Force group - Support in design and planning of RD clinical studies with ECRIN – demonstration projects on existing statistical methodologies to improve RD clinical trials – innovative methodologies to improve RD clinical trials in limited populations (validation of outcomes from ASTERIX, IDeAI, InSPiRe)

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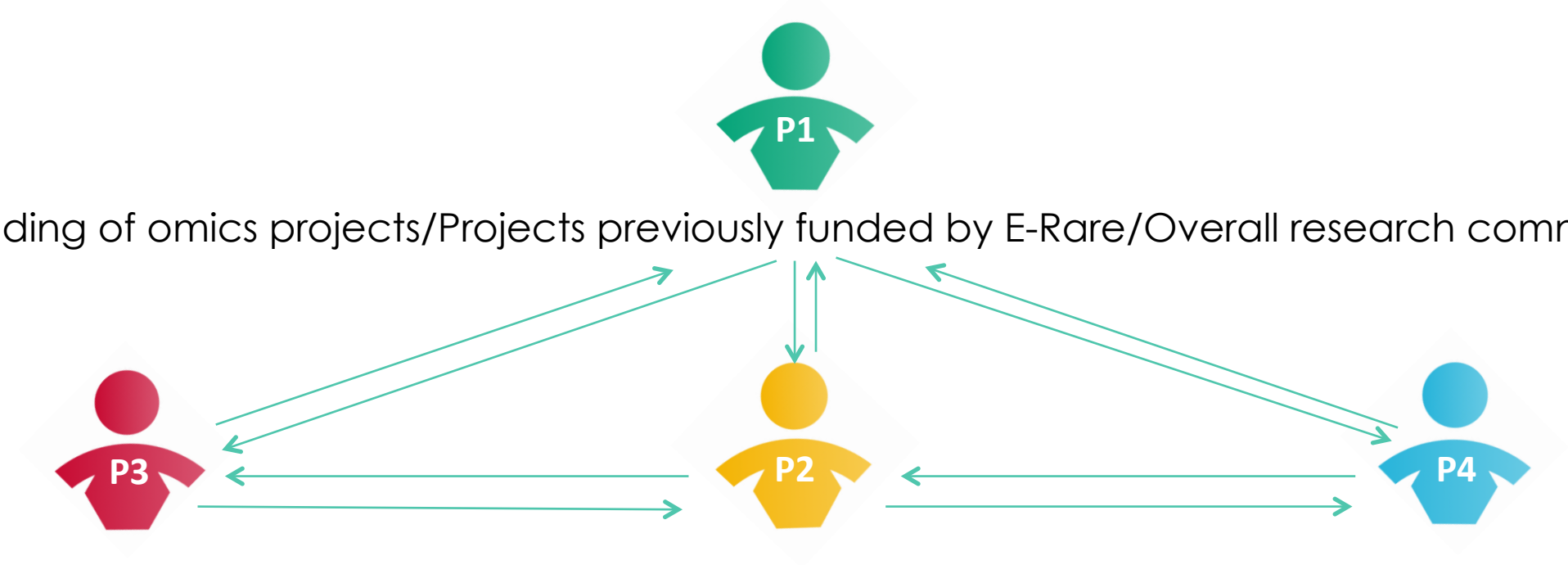

- Funding of omics projects/Projects previously funded by E-Rare/Overall research community

- Access to dedicated trainings
- Increased knowledge of new generations
- Development of new relevant trainings

- Access to and deposit of data
- Availability of additional resources & tools
- Contribution to the development of the virtual platform, interaction and input for ERNs

- Access to direct support by innovation managers & tools
- Direct expertise from ERNs
- Translation of gen(omic) results into accelerated diagnosis & treatment

INTERNATIONAL, EU, NATIONAL, REGIONAL STRATEGIES & FACILITIES

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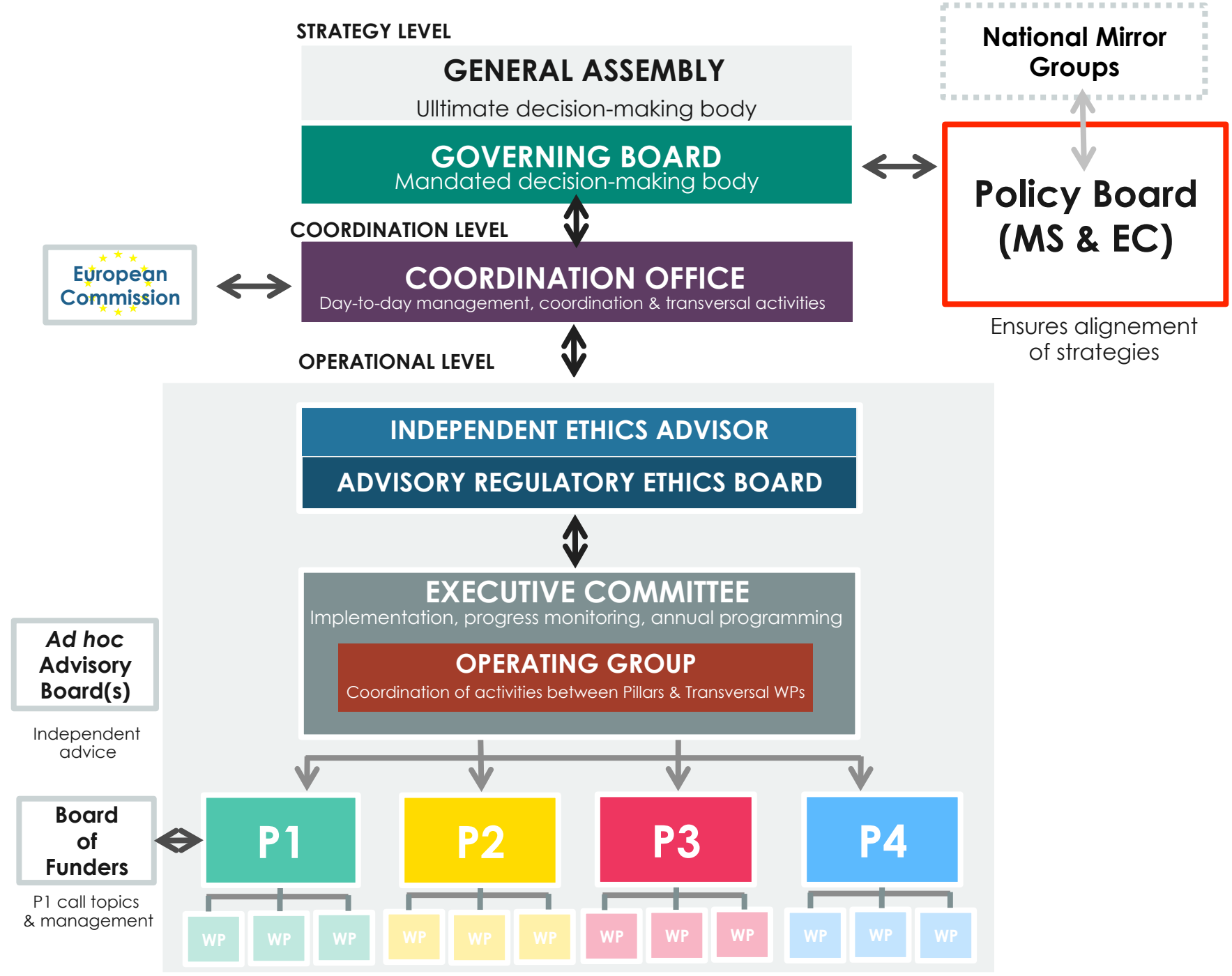
EJP RD GOVERNANCE

EUROPEAN JOINT PROGRAMME
RARE DISEASES



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POLICY BOARD & BOARD of FUNDERS

- The **POLICY BOARD** have a major role in ensuring the dialogue and translation through its participation is EJP RD strategy and sustainability development. It meets once a year.
- The Policy Board will be constituted from:
  - Representatives of national ministries of research and health;
  - Representatives of European Commission Directorates: DG RTD, DG Santé, DG Connect;
  - Representative of the pharmaceutical industry and public-private initiatives (e.g. EFPIA, IMI, EUCOPE, EuropaBio);
  - Representative of regulatory authorities (e.g. European Medicines Agency, EMA, esp. Committee for Orphan Medicinal Products, COMP, EuNetHTA);
  - Chair of the European Strategy Forum on Research Infrastructures (ESFRI);
  - Chair and vice-chair of the International Rare Diseases Research Consortium (IRDiRC)

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NATIONAL MIRROR GROUPS

It is up to each participating country to decide on how to establish a common voting position on each agenda item, so that the vote faithfully represents a consensual national position and not the one of any specific institution.

- **NATIONAL MIRROR GROUP:**
- Participating countries will be strongly advised to constitute NMG, bringing together the national representatives of the EJP RD and other relevant RD stakeholders. The creation and composition of a NMG is at the discretion of each participating country. Although not mandatory, it is expected that the establishment of National Mirror Groups will ensure that national activities, strategies and needs are taken into account when taking decisions at the EJP RD level and when designing the annual work plans.
- NMG ensures national coordination, contribute to the objectives of the EJP RD and benefit from it
- Is expected to include representatives of the National plan for RD, national nodes of the European Reference Networks, relevant national authorities and research institutions (whether participating to the EJP RD or not), as well as the relevant national partners of the EJP RD and **GB member** that will report NMG views and positions during GB meetings.

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ROUND TABLE

EUROPEAN JOINT PROGRAMME
RARE DISEASES

Presentation of EJP RD achievements, activities & Annual Work Plan for Year 2

EUROPEAN JOINT PROGRAMME RARE DISEASES

Coordination & transversal activities

EUROPEAN JOINT PROGRAMME RARE DISEASES

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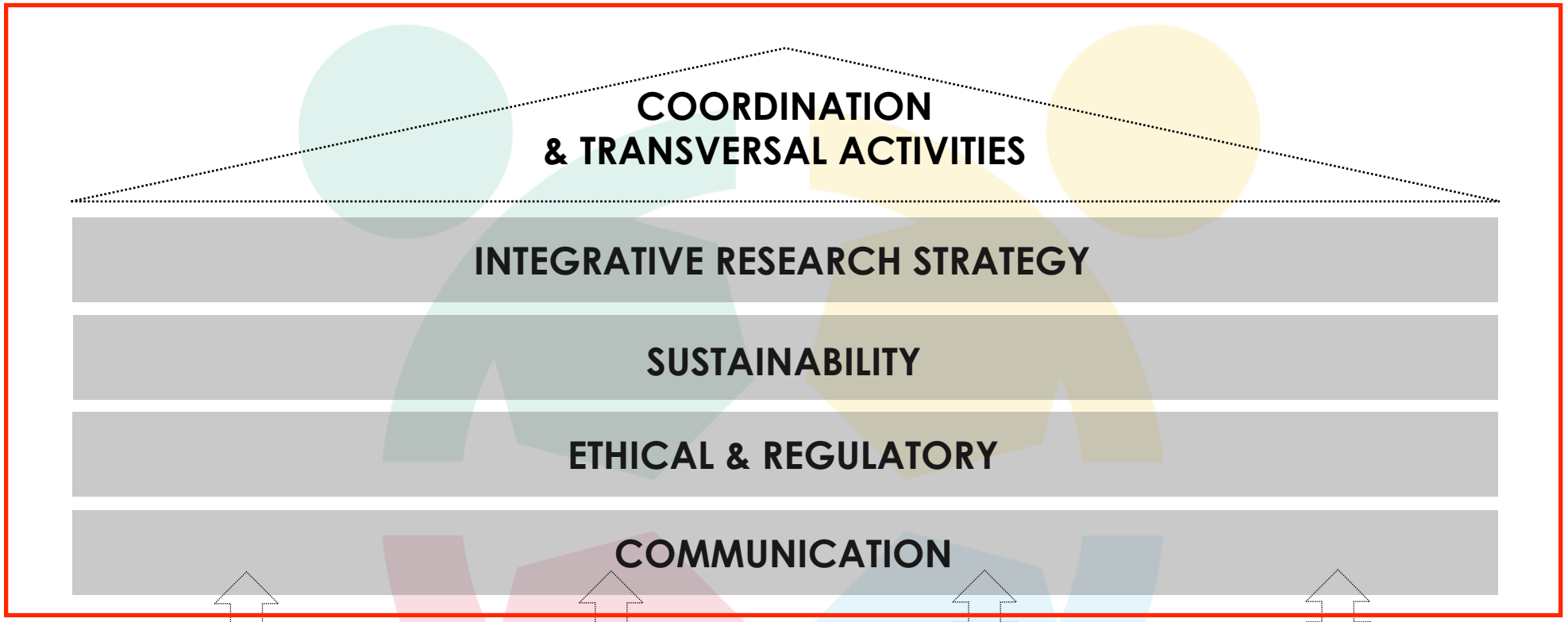
From science to health



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COORDINATOR

- Supervision of DAY-TO-DAY strategic, operational management, legal, financial & communication aspects
- Representation of EJP RD at EU & international level
- Primary contact for the European Commission

COMMUNICATION OFFICER

- Communication & dissemination activities of the EJP RD as a whole
- Communication activities of specific Pillars
- Support to SciSec of IRDiRC

PROJECT MANAGERS

- Day-to-day operational management
- Support to ExCom, Pillar leaders and WP leaders
- Scientific Secretariat of IRDiRC
- Central Helpdesk

FINANCIAL OFFICER

- Financial follow up and report of the EJP RD
- Advise and support to ALL EJP RD partners

ASSISTANT

- Assistance in the organisation of EJP RD & IRDiRC meetings
- Day-to-day support for the coordination team

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Coordination and transversal activities

- **WP1: Programme management & coordination**

Coordination office & governance – SciSec of IRDiRC – Central Helpdesk – Annual work plans – monitoring/KPIs – Data management plan

- **WP2: Integrative research & innovation strategy**

Prioritization strategy – mapping of research needs – scientific programming of calls – medium & long term strategy – alignement of national and EU strategies

- **WP3: Sustainability**

Roadmap of needs & expectations – sustainable service catalogue – EJP RD sustainability business plan

- **WP4: Ethics, legal, regulatory & IPR**

AREB – management of transversal legal, ethical & IPR issues

- **WP5: Communication & dissemination**

External communication & dissemination of EJP RD & IRDiRC results – expansion to & interactions with stakeholders

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M1 – M7 Achievements

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RARE DISEASES

P0: M1-M7 achievements

- <u>WP1: Coordination and management</u>
  - Coordination and governance structures set up (coordination office, OG, ExCom, GA, GB & PB, IRDiRC SciSec)→ regular meetings in place
  - Smooth uptake of IRDiRC activities: organisation of meetings, NL, follow up of existing Task Forces
  - The Central Helpdesk has been established and the internal database of EJP RD experts has been created
  - EJP RD internal communication: monthly internal NL, common agenda

  - Monitoring activities handbook submitted: proposition of a list of potential KPIs to be used for the monitoring of the EJP RD as a whole in terms of efficiency (performance of the consortium) and scientific performance (outcome) with the indication of the ways to measure them
  - Set up of the coordination office → some delays in the recruitment of the financials and administrative manager

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P0: M1-M7 achievements

- WP2: Integrative research and innovation strategy
  - A state-of-the-art of existing strategies/priorities/needs has been performed
  - A list of initial prioritisation criteria proposed

- WP3: Sustainability strategy and business plan
  - Development of a survey about sustainability of resources (initially planned to be sent to NMG, will be sent to WPL)
  - A handbook describing key sustainability considerations was created and will be disseminated together with the survey
  - An annotated contact database of stakeholders for the EJP RD has been set up
  - Activities run in coordination with Pillars to ensure that sustainability considerations are embedded into activities from the outset

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P0: M1-M7 achievements

- WP4: Ethical, regulatory, legal and IPR framework of the EJP RD
  - Advisory Regulatory Ethics Board (AREB) has been set up (1st meeting 03/07)
  - The coordination is in process of recruitment of an "Ethics Advisor" - ethics/regulatory experts - to overseen the whole EJP RD
  - The set of IP template agreements has been postponed to be aligned with the provisions of the Consortium Agreement not yet finalised

- WP5: Communication & Dissemination
  - EJP RD website, internal and external NL, twitter
  - A communication pack (templates, EJP RD standard presentation) was disseminated to partners
  - EJP RD has been presented in multiple conferences/workshop by the coordination/ partners participating in the programme
  - Internal network for communication was established
  - The joint scientific committee for the RE(ACT)-IRDiRC 2020 congress has been established

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M7 – M12 Planned Activities

EUROPEAN JOINT PROGRAMME
RARE DISEASES

P0: activities planned M7-M12

- **WP1**
  - First GA/consortium meeting to be held in Gdansk, 17-19 September 2019
  - Delivery of the final version of Annual Work Plan for Year 2
  - Preparation and delivery of the training workshop for EJP RD administrative officer
  - Follow up of IRDiRC activities (F2F meeting in November)
  - Finalize the Data Management Plan
  - Implement the acquisition, monitoring and control of KPIs
- **WP2**
  - List of prioritisation criteria to be validated by the Policy Board during the face-to-face meeting → finalisation of the prioritisation procedure
  - After discussion of the Policy Board, the outcomes of the mapping and prioritization will be reformulated and delivered in the form of Scoping Paper
  - Delivery of the First Analysis of national state of play and alignment process with EJP RD
- **WP3**
  - Sustainability strategies that exist mostly in the EU research fields collected and compiled to be analysed and collated into a report in Y2.
  - Follow up to survey with interviews and analysis and communication with all WP Leaders to collect information on respective needs, expectations and possible contributions.
  - Further development of Sustainability handbook as a resource for all WPs.
  - Annotated contacts database for all key stakeholder for the EJP-RD produced and shared with WPs 2 & 5.
- **WP4**
  - Contract between the Ethics Advisor and the Coordinator expected to be finalized in the next few months
  - First AREB meeting → first AREB report
  - Update on the ethical/regulatory/legal/IP issues for EJP RD partners (presented & produced in form of report)
- **WP5**
  - Continue communication & dissemination activities → update the Communication Plan based on inputs collected from ExCom members

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EJP RD
Annual Work Plan
Year 2

EUROPEAN JOINT PROGRAMME
RARE DISEASES

P0 : Annual Work Plan of Year 2

- WP1: Coordination & management
  - **Expand the EJP RD** consortium by involving more the under-represented countries, **include Cyprus, strengthen the collaboration with Policy Board & IRDiRC countries**
  - Start the **preparation of the next phase of the EJP RD** (under Horizon Europe) targeting innovation (EIT Health, IMI, etc.)
  - **Implement IRDiRC's Roadmap 2020** (Task Forces) in alignment with EJP RD activities:
    - Continuing activities: Orphan drug development guidebook; Database for funders; Data sharing policies; Patient engagement in research; Gene & drug counter; Indigenous populations; Natural history studies; Clinical research networks
    - New proposed activities: Chrysalis project; New technologies & integrated omics; Molecular aetiology of rare diseases; Drug repurposing guidebook; Access to drugs for all.
  - **Expand the Central Helpdesk** & demonstrate its efficiency for RD community

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P0 : Annual Work Plan of Year 2

- WP2: Research & innovation strategy
  - **Prioritize** items to be implemented in Year 3 (calls for projects; training activities; steps for development of Pillar 2 virtual platform)
  - Liaise and **develop medium & long-term strategy in collaboration with National Mirror Groups & Policy Board**
- WP3: Sustainability
  - Develop the **catalogue of sustainable services of the EJP RD**
  - Prepare the **report on sustainability models** and their application to EJP RD (per activity/type of service & globally)
- WP4: Ethics, regulatory, legal and IP issues
  - **Support partners and monitor ethical, regulatory & legal compliance** of EJP RD actions (calls, funded projects, activities implemented by different pillars)
  - Provide the **IP support on demand**
  - Keep EJP RD partners **up to date on any new ethical, regulatory or legal changes** that may influence RD research

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P0 : Annual Work Plan of Year 2

- WP5: Communication & dissemination
  - Follow up the **implementation of Communication & Dissemination plan**
  - **Increase the visibility of the EJP RD** by specific actions (Twitter; interviews; publications; dedicated actions)
  - **Organize RE(ACT) – IRDiRC Congress 2020** (11 – 14 of March 2020, Berlin)

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How to better involve all countries?

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RARE DISEASES

P0 : How to more involve all participating countries in the EJP RD?

- Coordination → ensure participation of all countries (currently Cyprus missing)
- Stronger connection between EJP RD & Policy Board → dedicated meetings/ support for national requests
- National Mirror Groups will be key to ensure the dialogue
- Ensure that the information on needs/bottlenecks but also about strengths/ capacities is provided for EJP RD requiring strategic decisions
- Enhance and exploit the capacity of national stakeholders for communication & dissemination (provide information in national languages when possible)

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Pillar 1: Collaborative research funding

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RARE DISEASES

P1 : Collaborative research funding: Global objectives

- Enhance the cooperation between scientists working on rare diseases across Europe and beyond
- Reduce fragmentation of research
- Increase knowledge in the fields of basic, translational, clinical, social and health systems research
- Accelerate development of diagnostic tools and treatments
- Enable translation of research results
- Contribute to the objectives of the International Rare Diseases Research Consortium (IRDiRC)

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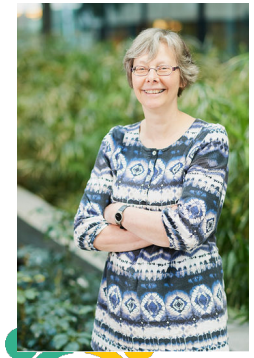
P1 : Collaborative research funding Global activities

WP6: Joint Transnational Calls for collaborative research projects

- WPleader: Ralph Schuster, DLR-PT
- 2 cofunded calls + 2 non-cofunded – expect to fund research for 60-80 Mio. €– open to research teams from countries with funders involved

WP7: Networking to share knowledge on rare diseases



- WPleader: Sonja van Weely, ZonMw
- Small support schemes for networking (workshops/events/share of knowledge) – 30 K€ max – 2 M€ budget – open all year long – open to all countries involved in EJP RD

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P1 : Collaborative research funding Global activities

WP8: Rare disease research challenges

- WPleader: Christine Fetro, FFRD; Virginie Bros-Facer, EURORDIS
- Public-private (small scale) partnerships – challenges set by industry and validated by EJP RD – short term projects – 1.5 M€ budget

WP9: Monitoring of funded projects

- WPleader: Irit Allon, CSO-MOH
- Monitoring of all projects funded through EJPRD P1 and previous projects through E-Rare

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P1 : M1-M7 achievements

- WP6: Joint transnational Calls (JTCs)
  - JTC 2019 topic "Transnational research projects to accelerate diagnosis and/or explore disease progression and mechanisms of rare diseases"
    - Participation of 31 funders from 23 countries
    - Total earmarked national budget of about 27 Mio. €
    - 220 pre-proposals submitted, of which 217 eligible according to formal criteria
    - 52 pre-proposals selected for submission of a full proposal after evaluation in April 2019 by the Scientific Evaluation Committee
    - It is expected that about 25-30 projects could be funded in this call after the second evaluation in July-September.

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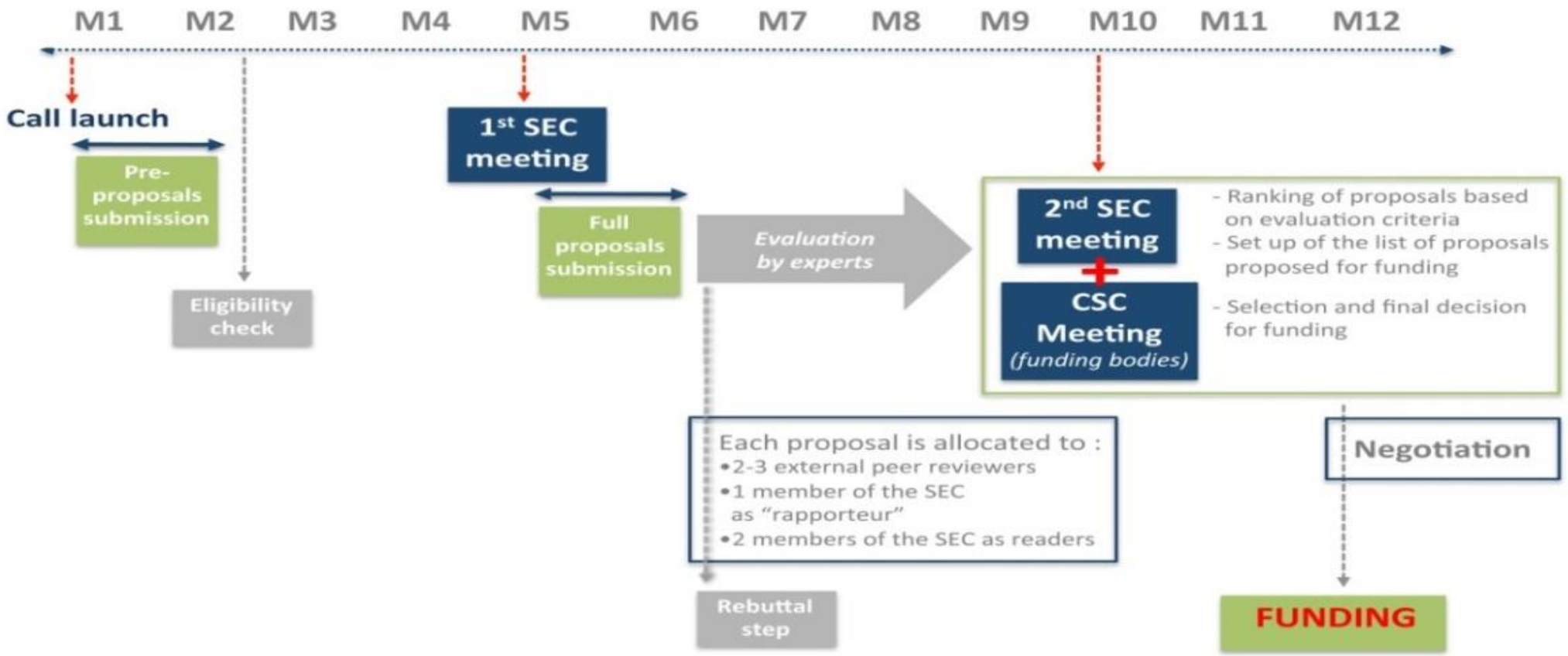
P1 : WP6 - Transnational calls – how it works



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P1 : M1-M7 achievements

- WP6: Preparation of second and third JTC
  - JTC 2020: first step to prepare potential topics which tentatively could focus on pre-clinical therapy development
  - JTC 2021: in the context of ERA-Net E-Rare-3 a Workshop on Social and Human Sciences (SHS) Research is being prepared for September 2019 that may be chosen as the topic for JTC 2021

- WP9: Monitoring of funded projects
  - Preparation of the foundation for the monitoring work package in general
  - The list of indicators for ex-post evaluation of the funded projects in JTCs has been updated and refined
  - The online monitoring tool is being adapted accordingly

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P1 : M1-M7 achievements

WP7 and WP8: Development of new funding initiatives

- WP7 Networking Support Scheme
  - Draft documents for the application and evaluation procedure are written and discussed with a working group consisting of funding agencies

- WP8 Rare Disease Research Challenges
  - Two Workshops have resulted in 3-4 challenges elaborated by 1-2 industrial partners per challenge
  - The 2-stage process has been discussed for the 3-4 pre-identified challenges with estimation of the budgets required including contribution of the industry, and expected outputs

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P1 : activities planned M7-M12

- **WP6: Joint Transnational Calls**
  - Evaluation, selection and granting of JTC 2019 full proposal projects that will be cofunded by the EC
  - Choice of final topic and launch of JTC 2020 Call
  - Workshop on SHS research to prepare potential JTC 2021 topic

- **WP7: Networking Support Scheme**
  - Implementation of the Networking Support Scheme that is foreseen in Q3

- **WP8: Rare Disease Research Challenges**
  - Preparation of Call documents in which the approved challenges will be described
  - Preparation of networking/partnering event for academia and industry to prepare proposals
  - Pre-announcement of the Call (M12)

- **WP9: Monitoring funded projects**
  - Preparation of list of indicators for ex-post evaluation of the funded networking events (WP7)
  - Adaptation of the online monitoring tool for the funded networking events (WP7)

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P1 : Annual Work Plan of Year 2

Objectives and activities

- WP6: Joint Transnational Calls
  - Implementation of JTC 2020: Evaluation and granting JTC 2020 projects that will be cofunded by the EC (M13-M24)
  - Choice of final topic and launch of JTC 2021 (M16-M24)

- WP7: Networking Support Scheme
  - Further implementation of the Networking Support Scheme (M13-M24)
  - Early assessment of the Networking Scheme and further improvement (M21-M24)

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P1 : Annual Work Plan of Year 2

Objectives and activities

- WP8: Rare Disease Research Challenges
  - Implementation of the Rare Disease Research Challenges funding scheme:
    - Networking/partnering event takes place (M13)
    - Announcement of the call (M14)
    - Submission, eligibility check and selection procedure of first phase, proof of concept projects, responding to the proposed challenges (M14-M21)
    - Funding decision of first phase projects and establishment of contracts (M21-M24)

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P1 : Annual Work Plan of Year 2

Objectives and activities

- WP9: Monitoring funded projects
  - Annual monitoring of funded E-Rare JTC 2017 and E-Rare JTC 2018 projects (M13-M24)
  - Monitoring, analysing and writing a report on the first set of networking events (WP7) (M13-M24)
  - Developing the indicators and adaptation of the monitoring tool for the Rare Disease Challenge scheme (WP8) (M17-M24)

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P1 : how to more involve all European countries in the EJP RD?

- Inform funding agencies in all European countries about joining the Joint Transnational Calls (WP6)
- Involve health care professionals, researchers and patient advocacy organizations from all European countries in networking events (WP7)
- Invite representatives of academia in all European countries for the networking/partnering meeting in WP8 to prepare projects for the Rare Disease Research Challenges

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Pillar 2: Innovative coordinated access to data and services for transformative rare diseases research

EUROPEAN JOINT PROGRAMME
RARE DISEASES

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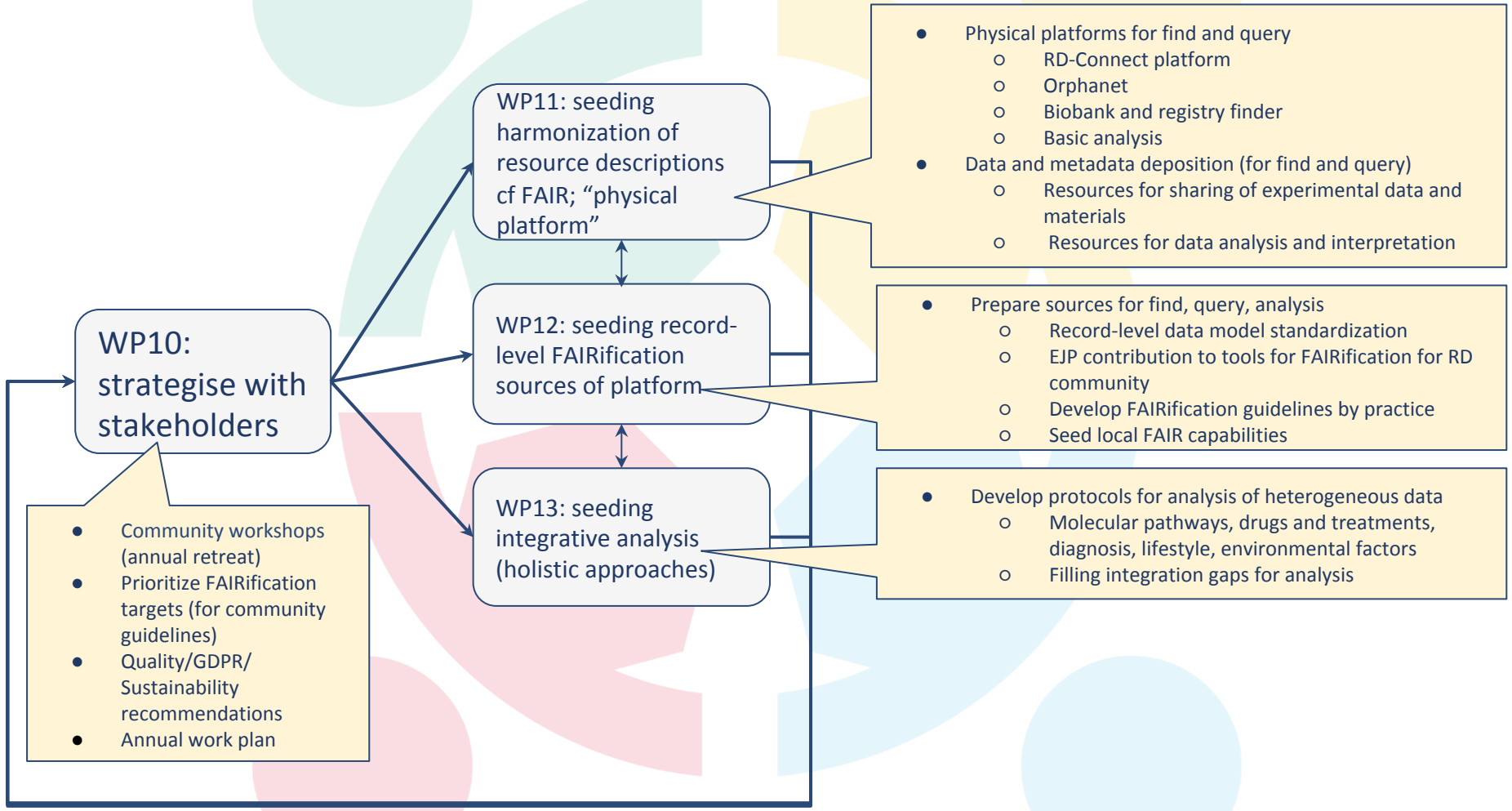
PILLAR 2 WORKFLOW



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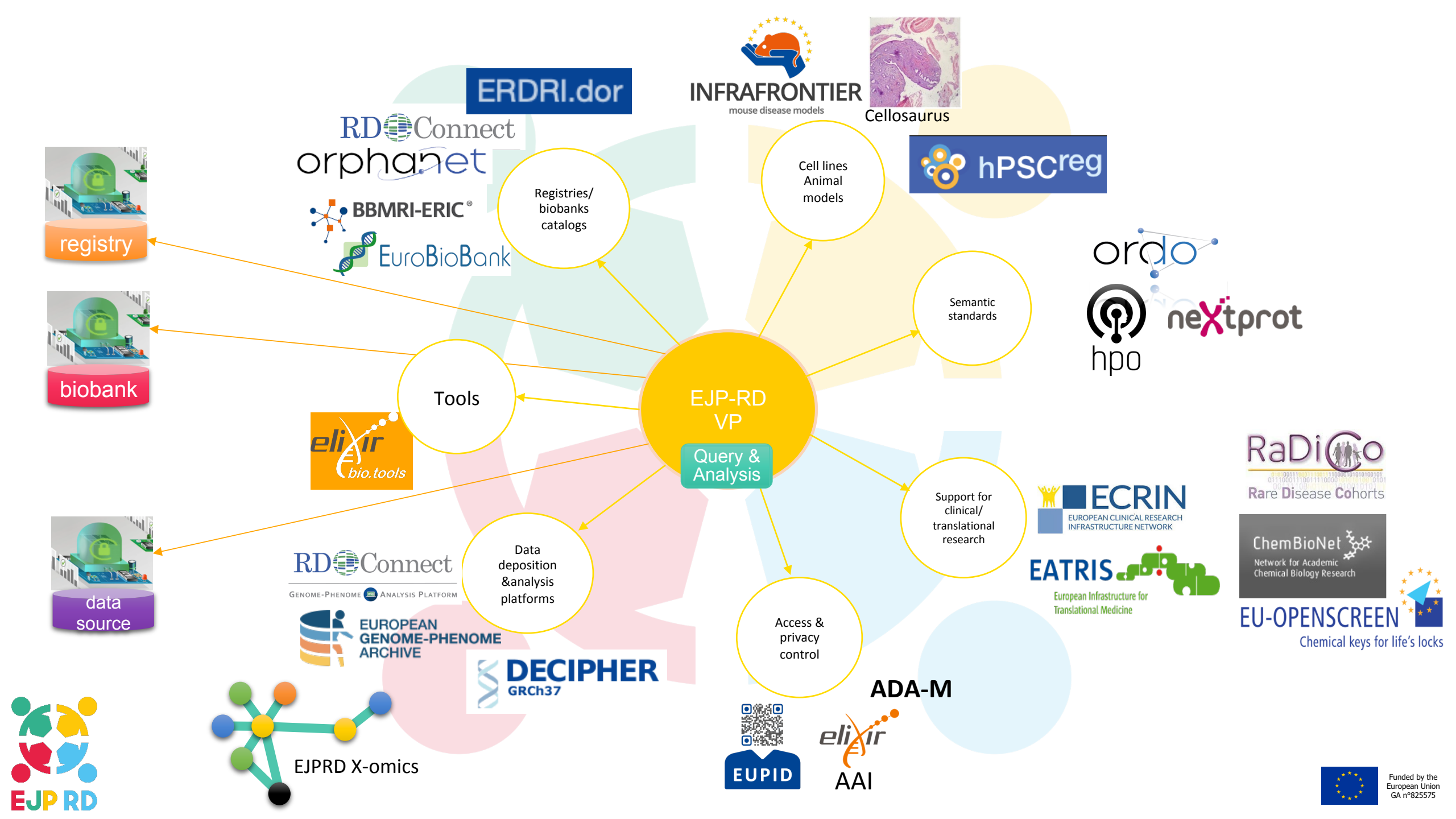
ERDRI.dor
INFRAFRONTIER
mouse disease models
Cellosaurus
RD Connect
orphanet
BBMRI-ERIC
EuroBioBank
hPSCreg
registry
biobank
data source
Registries/ biobanks catalogs
Cell lines Animal models
Semantic standards
Tools
EJP-RD VP
Query & Analysis
Support for clinical/ translational research
Data deposition &analysis platforms
Access & privacy control
elixir bio.tools
ordo
hpo
neXtprot
RaDiCo
Rare Disease Cohorts
ECRIN
EUROPEAN CLINICAL RESEARCH INFRASTRUCTURE NETWORK
EATRIS
European Infrastructure for Translational Medicine
ChemBioNet
Network for Academic Chemical Biology Research
EU-OPENSCREEN
Chemical keys for life's locks
RD Connect
GENOME-PHENOME ANALYSIS PLATFORM
EUROPEAN GENOME-PHENOME ARCHIVE
DECIPHER
GRCh37
ADA-M
EUPID
elixir
AAI
EJPRD X-omics
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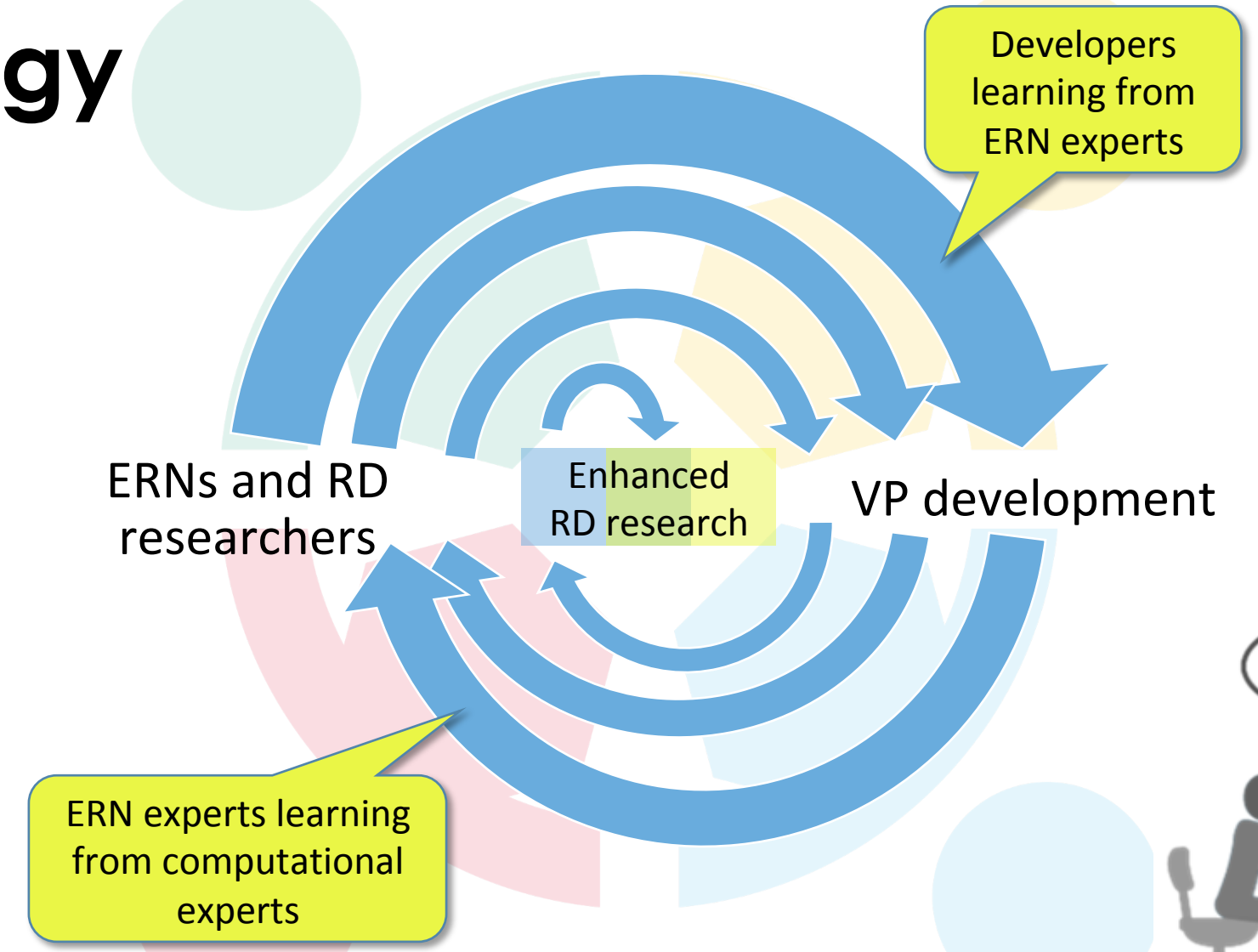
Strategy



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P2: M1-M12 achievements and planned activities

- WP10: User-driven strategic planning and transversal activities for Pillar 2 data ecosystem
  - A **survey** has been sent to ERNs to analyse their needs
  - **1st Annual Retreat**: decision on priorities for the next year → Annual strategic report to be prepared
  - A set of **quality criteria and quality level** have been **proposed** (based on ELIXIR's set)
  - Ongoing **work with WP3 on sustainability**: collection of information on what should be made sustainable in P2
  - EJP RD selected as **GA4GH Driver Project** → contribution to the establishment of global genome data sharing standards
  - Report on the State of the Art of existing resources to be prepared

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P2: M1-M12 achievements and planned activities

- WP11: Common virtual platform for discoverable data and resources for RD research
  - **WP11-WP12 Explorathon**: first draft of the Virtual Platform architecture proposed
  - **Startathon**: first metadata model proposed
  - **Submission interface, automated selection of research projects specific for RD** and curation interface have been developed and are being tested by Orphanet and IRDiRC Funders Constituent Committee (FCC) representatives. The analysis interface is being developed
  - **ELIXIR's bio.tools** catalogue continued its adaptation for the RD community
  - **Available analysis functionalities** were displayed in the Pillar 2 annual retreat and initial points of collaboration were initiated: for example, the MatchMaker Exchange connection between RD-Connect GPAP and DECIPHER has been launched
  - Ongoing activities that should be available at the end of Year1
    - Ontological model of resources metadata,
    - VP of RD resources annotated with EJP RD ontological model,
    - additional facilities integrated to resources regarding data deposition and access
    - a report on processed genome-phenome datasets and mutli-omics use cases analysed

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P2: M1-M12 achievements and planned activities

- WP12: Enabling sustainable FAIRness and Federation at the record level for RD data, patients and samples
  - WP11-WP12 **Explorathon: first draft of the Virtual Platform architecture proposed; VP module teams started working**
  - Use cases and scenarios for all tasks, including the metadata for records are being collected to be further prioritized (with WP11)
  - A Work Focus Team was formed to develop and evolve **interoperability considerations for ERN registries,** to help them be compatible with the EJPRD Virtual Platform and the underlying FAIR principles
  - **WFs setup for WP12** to go into building mode, some transitioned from pre-EJPRD initiatives

Explorathon Vienna April 3-4

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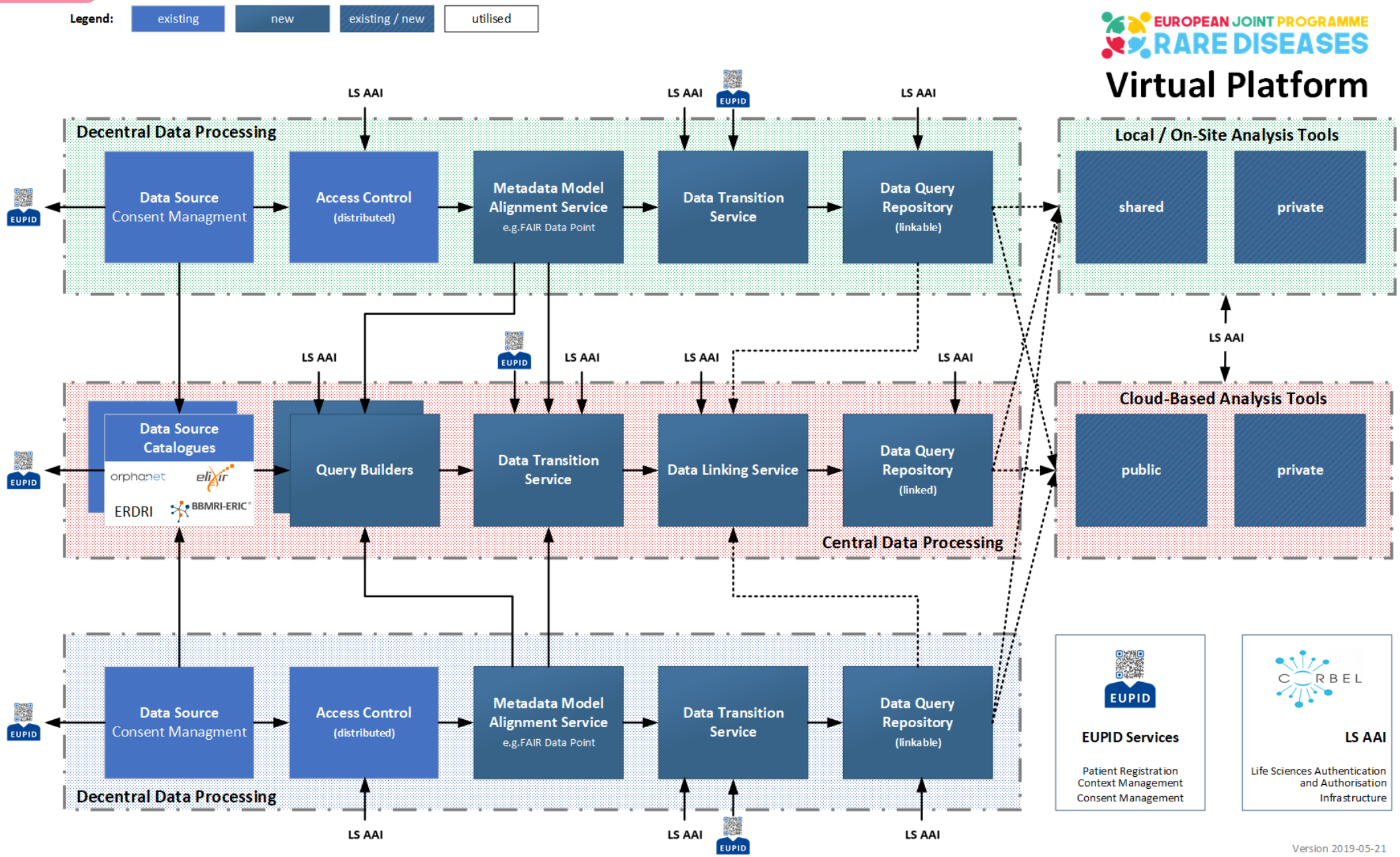
VP structure



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Version 2019-05-21

P2: M1-M12 achievements and planned activities

- WP13: Enabling multidisciplinary, holistic approaches for rare disease diagnostics and tehrapeutics
  - A rare disease pathway portal was established: **37 rare disease pathways** (on WikiPathways database) interesting genes and diseases were extracted from the ERN survey for future pathway creation work
  - **Pilot work** will be on histone deacetylase inhibition as molecular initiating event of neural tube defects (integration with EuToxRisk work on AOP), that will be presented at the European Teratology Society conference (Cologne, September 2019).
  - **Workshop** to teach pathway creation and to address and apply FAIR approaches to pathway development (**November 2019**)

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P2 : activities planned M7-M12:Work Foci

- **WF1: Use cases**: setting up research questions by ERNs partners that will drive the development of VP components based on real-world needs
- **WF2: Overall architecture**: global overviewing the VP components and connections between them
- **WF3: Data sources**: working up the integration of different sources of data, including ERN's registries as a priority
- **WF4: FAIRification:** allowing data sources to become progressively FAIR, pertaining to incorporating technical services from Pillar 2 and collaboration with local data stewards, focussing on ERN registries and selected OMICS data resources.
- **WF5: Distributed and federated consent control:** Defining where and how consent control is done based on the state-of-the-art and fitting it into the overall architecture of the VP. Defining other legal bases and definitions of roles (controller vs. processor in GDPR) for entities contributing or interfacing to the VP.
- **WF6: AAI:** Providing authentication and authorization infrastructure (AAI) to be used by other components of VP. Building on ELIXIR AAI, BBMRI-ERIC AAI and the upcoming LifeScience AAI.
- **WF7: Personal data linkage service:** identify datasets which belong to the same person (privacy-preserving record linkage)
- **WF8: Query builder**
- **WF9: Unicity/identifier resolution**: identifying and resolving duplicated data
- **WF10: Metadata model and alignment service:** computable ontology-based model of interoperable data descriptors using semantic standards.
- **WF11: Interoperability standards & GA4GH partnership**: mapping expertise and standards between GA4GH, ERNs, and EJP-RD Partners, with an initial focus on suggestions and guidelines for ERN registries
- **WF12: Resources for sharing experimental data and materials**
- **WF13: Resources for experimental data and analysis interpretation**
- **WF14: Biological networks analysis methods**
- **WF15: Adverse outcome pathways**

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P2 : Annual Work Plan of Year 2

- **WP10:** User-driven strategic planning and transversal activities for Pillar 2 data ecosystem
  - To **expand the first proof-of-principle** achieved in year 1 and **based on simple use cases**, so as to incorporate more complex ones, by **coordinating the development of the different modules of the Virtual Platform (VP) organised in Work Foci**,
    - WFs & agile as a general strategy for P2
    - moving from exploratory phase into building phase that will continue in Y2
    - focus on ERN registries
    - essential: involvement of stakeholders in agile process; working on models to establish this.

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P2 : Annual Work Plan of Year 2

- **WP11:** Common virtual platform for discoverable data and resources for RD research
  - **Achieve full interoperability between catalogs** addressed in year 1 and the VP, and expand it to **new prioritized resources** (RD-Connect GPAP, ECRIN and EATRIS) as well as to the toolbox developed in Pillar 4.
  - **Provide data on ongoing research projects and trials** for gap and opportunities analysis in order to serve the global research strategy at EJP and IRDiRC level.
  - **Adapt and integrate data deposition resources** according to RD community needs by working with representatives identified through the 2019 survey to the ERNs and new funded EJP-RD JTC projects.
  - Implement new user friendly and **cloud data analysis functionalities** for the RD community prioritised after testing. and evaluation of existing resources and tools by the task force established in Task 11.4 during 2019.

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P2 : Annual Work Plan of Year 2

- **WP12:** Enabling sustainable FAIRness and Federation at the record level for RD data, patients and samples
  - To **identify and expose core standards** for Findability, Accessibility, Interoperability and Reusability (FAIR) of assets (data, sample, subject), **concentrating on data level**, **guided by the needs of the EJP RD stakeholders** including ERNs
  - To **align and create joint working practices** between multi-site teams that will assess the availability and interoperability of their own and other open-source tools, defining and maintaining a solution architecture for the VP, and for community needs at the data level in general. This will be done in a manner **that ensures compliance with currently popular standards to enable selected RD data to be sufficiently FAIR**
  - the aim of establishing **a toolbox for data stewards**, and a toolbox for system engingeers to make resources FAIRer
  - First focus on CDEs, practical use cases requires going beyond CDEs which is part of agile process.

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P2 : Annual Work Plan of Year 2

- **WP13:** Enabling multidisciplinary, holistic approaches for rare disease diagnostics and therapeutics
  - To identify (from the pool of EJP RD partners and especially ERNs) the groups with relevant omics data, their needs for multi-omics analysis and **focus on delivering expert curated rare disease pathways** (on the online pathway repository WikiPathways) to **start including**
    - **genetic variant-to-disease**
    - **variant-to-protein function mapping**
    - **knowledge from drug and environmental toxicology**

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P2 : how to more involve all European countries in the EJP RD?

- **1+MG**: signatory MS; engage non-signatory EJPRD countries and vice-versa; Liaise with national genome programmes
  - Sharing best practices for data collection, deposition and sharing (1+MG as use-case for EJPRD) and EJPRD participating to the RD use case (WG8)
- Disseminate interoperability standards and FAIR principles to **national registries**, and to include them to the coordinated access VP
- ERNs to disseminate usability of the VP from every MS; **Follow the ERNs expansion**
- Involve **infrastructures nodes** from countries that are not in EJPRD: (Ex: ELIXIR:Luxembourg)

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Pillar 3: Capacity building and empowerment

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P3 : Capacity building and empowerment Global objectives

- **Increase the level of knowledge and know-how within the RD research and care community,** including through ERNs and RD patient representatives (adult & pediatric)

- **Enhance the innovation potential of the RD community**
  - Comprehensive, coherent & accessible EU RD research training programme
    - Rotating F2F courses, fellowships, online academic course
  - Leverage on existing training expertise & resources and create new experts
    - Development of existing & new courses with faculty members with a wide range of expertise (EJP partners)
  - Accelerate access to diagnostic tools and treatments for RDs
    - Facilitating access to training in latest advances in scientific innovation and clinical research relevant for RDs

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P3 : Capacity building and empowerment Specific objectives

- **WP14: Training on data management & quality- Claudio Carta, ISS - 6 training activities (5 courses in year 1 and 7 in year 2)**
  - Decrease RD data fragmentation and increase data quality which will raise the level of capacities and help data sharing and networking within the RD community (existing and new courses)

- **WP15: Capacity building and training of patients and researchers - Virginie Bros-Facer, EURORDIS – 4 training activities (2 in year 1 and 3 in year 2)**
  - Improve RD research & innovation and enhance uptake of research results by building the capacity of the patient community and other key stakeholders (existing and new courses)

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P3 : Capacity building and empowerment Specific objectives

- **WP16: Online academic education course - Roseline Favresse & Emilie Bonnaud , FFRD**
  - Provide a EU-wide streamlined education programme on RD research to all interested stakeholders via an e-learning (brand new)

- **WP17: ERN RD training and support programme - Holm Graessner, University of Tubingen**
  - Deliver research training programs for the European Reference Networks (ERNs) focusing on cross-cutting and over-arching research themes (brand new)
- **WP18: Development and adaptation of training activities -Birute Tumiene, VUHSK &Krystyna Chrzanowska, IPCZD (CHMI)**
  - Ensure that activities within Pillar 3 address the developing education and training needs in RD research of key stakeholders across different EU countries

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P3: M1-M7 achievements and planned activities M7-M12

- WP14: Training on data management & quality
  - Training on the Orphanet nomenclature and RD ontologies for RD research (14.1) – Sylvie Maiella & Marie Verrey (Orphanet)
    - Orphanet Training manager has been hired with a short delay
    - Currently conducting interviews with volunteers among all relevant stakeholders to define what needs to be included in the Orphanet nomenclature and ontologies training modules
  - Standards and quality of genetics/genomics data in laboratory and clinical research practice (14.2) – Gert Matthijs (KULeuven)
    - programme of the first course is finalized and registration is open until 31st July for the course scheduled in October in Leuven

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P3: M1-M7 achievements and planned activities M7-M12

- WP14: Training on data management & quality
  - Training for biobanks and researchers/clinicians on sample data management (14.4) – Mary Wang (FTELE)
    - 25 participants attended the training on 1-2 April in Milan and 2 travel fellowships were awarded (one to a patient representative and one to an EU13 participant);
    - Feedbacks on the training were very positives:
    - mixture of lectures + practical sessions in small multi-stakeholder groups using problem-based learning (PBL) methodology;
    - Vilnius course in preparation (29-30 October)

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P3: M1-M7 achievements and planned activities M7-M12

- WP14: Training on data management & quality

  - Training on rare disease registries and FAIRification of data at the source (14.5) – Claudio Carta (ISS)
    - programme is finalized and registration just closed for course in September in Rome;
    - Up to 3 patient fellowships and 3 fellowships for participants from EU13 countries will be granted

  - EU RD Platform ERDRI Training Workshop (14.6) – Andri Papadopoulou (JRC)
    - Ispra, 25th June 2019

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P3: M1-M7 achievements and planned activities

- WP15: Capacity building and training of patients and researchers in Rare Disease research and processes
  - ExPRESS Expert Patients and Researchers EURORDIS Summer School took place on 10-14 June 2019 (15.1):
    - 35 participants (28 patient representatives and 7 researchers) from 16 countries representing 25 rare diseases;
    - All would recommend training to others and 86% of the respondents strongly agreed that the EURORDIS Summer School effectively builds the capacity of patient advocates on Medicines Research & Development.

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P3: M1-M7 achievements and planned activities

- <u>WP15: Capacity building and training of patients and researchers in Rare Disease research and processes</u>
  - Training for patient representatives and advocates on leadership and communication skills will take place on 25-26 November 2019 (15.3):
    - 57 participants from 19 countries representing 21 ERNs are registered.
  - For both courses, preparatory webinars have been held and online training courses followed by participants ahead of the face to face trainings.

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P3: M1-M7 achievements and planned activities

- WP16: Online Academic education course
  - Orientations of the EJP RD online academic course are refined and prioritized based on interviews held with 30+ stakeholders, ERN survey and survey towards non-ERN targets (EJP-RD partners, academics, research organisations, PO, other stakeholders);
  - Targeted audience has been refined. 2 levels for each module will be implemented: level 1: one short video addressing basic definitions and an overview of the subject; level 2: an advanced session with 5 to 10 units of advanced content and exercises (certification only for level 2);
  - A benchmark of existing RD academic courses at EU level has been developed and a list of online courses on RD research and related topics has been compiled since January and contains 40 courses.

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P3: M1-M7 achievements and planned activities

- WP17: ERN RD training and support programme
  - Based on the ERN survey results, fellow exchanges and workshops/seminars will be the two main schemes of the ERN research training programme;
  - A focus group meeting was held on 26 June to develop the ERN research training program based on the EJP-RD ERN survey results: topic prioritisation, participants/fellow selection and assessment;
  - The ERN RD training and support programme will be finalised by July 2019 in year 1;
  - The implementation of the programme will start in the second half of year 1 as planned.

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P3 : Annual Work Plan of Year 2
Streamlining and improving processes across P3

- Develop & implement common standards for course registration, selection of participants & fellowship assessment (aligned with KPI developed in WP1);
- Define core and specific criteria for quality and impact assessment – in close collaboration with WP1;
- Develop sustainability strategy for the training course in close collaboration with WP3;
- Develop/adapt a dissemination & communication strategy specifically for the training courses in close collaboration with WP3 and WP5.

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P3 : Annual Work Plan of Year 2

- WP14: Training on data management & quality
  - **Increase the number of trainees (x2)** for existing trainings (omics quality in clinical practice , RD registries & data FAIRification, biobanking)
  - Implement new training courses on Orphanet ontology and Undiagnosed Diseases
- WP15: Capacity building & training of patients
  - **Enhance patients' training and empowerment** through the innovative modules proposed with the addition of the Winter school (scientific innovation and translational research)
- WP16: E-learning academic course
  - Launch **first modules of e-learning academic course** open to all
- WP17: ERN RD training & support programme
  - Implement **ERN** dedicated training in form of **exchange of fellows and workshops**
- WP18: Expand the **knowledge sharing and adapt to needs** with/for EU13 countries

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Timetable 2020 Pillar 3 Training courses

Date	Activity
March	**EURORDIS Winter School on scientific innovation & translational research (15.2) Paris, France**
April	**Training on strategies to foster solutions of undiagnosed rare disease cases (14.3) Rome, Italy**
June	**ExPRESS Expert Patients and Researchers EURORDIS Summer School (15.1) Barcelona, Spain**
July + other dates	**Orphanet Train the Trainers (14.1.3) Paris, France + national trainings (14.1.4) multiples locations**
September	**International Summer School on Rare Disease Registries and FAIRification of Data (14.5) Rome, Italy**
October	**Standards and quality of genetics/genomics data in laboratory and clinical research practice (14.2) Istanbul, Turkey**
November	**EURORDIS Leadership School on Healthcare & Research (15.3) Gdansk, Poland**
2 dates TBC	**Two training wokshops for biobanks and researchers/clinicians on sample data management (14.4) Graz, Austria + Madrid, Spain**
Multiple dates	**ERN training support programme (17) via fellow exchanges and Seminars multiples locations**

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P3 : how to more involve all European countries in the EJP RD?

- All residential training courses offer several fellowships for participants from EU13 countries;
- Rotation of some courses (Leadership, biobank, genetic standards, orphanet trainings) will increase active participation of partners (within the organisation of the course and also as faculty/trainers);
- Develop/adapt a dissemination & communication strategy specifically for the training courses to attract wider representation of participants/trainees;

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Pillar 4: Accelerating the translation of high potential projects and improving outcomes of clinical studies in small populations

EUROPEAN JOINT PROGRAMME
RARE DISEASES

P4 : Accelerating the translation of high potential projects and improving outcomes of clinical studies in small populations

Global Objectives

1) to actively support researchers in navigating the translational and clinical pathway by providing access to the best resources, facilities and expertise

2) To improve clinical development outcomes in rare diseases R&D by prioritizing and validating promising methodological concepts and outcome measures

3) To improve industrial uptake of academic research results and reduce barriers to public-private & public-public collaboration

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P4 : Accelerating the translation of high potential projects and improving outcomes of clinical studies in small populations

Work Packages

WP19 Facilitating partnerships and accelerating translation for higher patient impact

- The overall aim is to provide researchers the competences to support rigorous translational research, to secure follow-on funding and find partners for the development of new treatments and diagnostics for rare diseases.
- This WP will support the RD community to more effectively translate high quality research into high impact interventions for the RD patient community.
- The specific objectives are:
  - ● Empower researchers to conduct rigorous translational research by providing self-help resources and active project mentoring and technical support;
  - ● Improve the uptake of successful research projects towards clinical implementation with active exploitation and follow-on funding support;
  - ● Improve efficiency and outcomes of translational research, and develop novel funding concept to reduce the 'valley of death' funding gap.

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P4 : Accelerating the translation of high potential projects and improving outcomes of clinical studies in small populations

Work Packages

WP20 Accelerating the validation, use and development of innovative methodologies tailored for clinical trials in RDs

- The overall objective of this WP is to foster the development of innovative methodologies tailored for clinical studies in RDs.
- The specific objectives are to map the best methodologies for clinical studies in RDs and to validate innovative and promising design. This will be achieved by a 3D methodology: Disseminate, Demonstrate, Develop & Design.
- This WP will support ERNs to use the most adapted methodologies improving clinical trial studies in RDs.
- Once validated, agreed and accepted by all stakeholders, the innovative methodologies will foster the achievement of IRDiRC goals.

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M1 – M7 Achievements

EUROPEAN JOINT PROGRAMME
RARE DISEASES

P4 : M1-M7 achievements

- **Preparation of Innovation Management Toolbox**
  - Work has begun to design the online environment and on creating an Advisory Committee on Therapeutics handbook and a project management manual.
- **Assessment of Translational Research Projects**
  - Projects funded through E-Rare 3 have been contacted describe the support that the EJP consortium makes available in terms of innovation management and follow-on funding/ exploitation and assess their interest. Several projects have already responded to the call for support.
- **Real-time mentoring and technical support for translational research projects**
  - The Innovation Coordinators have identified a pool of mentors who will support the EJP RD translational research projects and have over 60 individuals in a database, especially including experts in those areas where academic research groups usually do not have expertise.
  - For building the expert panel they have attended several meetings with CROs, Start-ups and research groups which are working on translational rare disease projects.

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P4 : M1-M7 achievements

- **Support in exploitation and follow-on funding**
  - The set-up of evaluation panels is ongoing.
- **Mapping of existing POC funding opportunities**
  - Mapping of initiatives with a wide span being carried out.
- **Roadmap for a European investment platform for RD**
  - The initiative has been introduced to the European Investment Fund and the European Investment Bank in order to understand in particular EIF's strategy on technology transfer as well as its priorities. Involvement of VC in the panel of experts also represents an opportunity to brainstorm on possible models for the investment platform.

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P4 : M1-M7 achievements

- **Task 20.1 : Task force group on clinical studies**

Subtask 20.1.1: The Task Force Group (TFG) on clinical studies was established in a face to face meeting by M1(January 23rd 2019) as foreseen and had a meeting with the ERNs research group.

- TFG elected its chair, Dr. Rima Nabbout (WP20 leader) and met again in a F2F meeting at M4 (April 17th-18th 2019) and maintained contact via monthly TC meetings.

Subtask 20.1.2 : Create, disseminate and analyse surveys for ERNs

- TFG participated in the EJP-RD survey addressed for ERNs with specific questions investigating the current state of the art and most required needs in clinical research in rare diseases from ERNs. Final results were discussed during the F2F meeting in M4 (April 17th-18th 2019) and further TC meetings.
- This helped the TFG to identify the major needs of the ERNs in the field of clinical studies and to develop the topics of the calls. This survey was done in collaboration with the research group of the ERNs.

Subtask 20.1.3 : Mapping existing methodologies and prioritize the needs for demonstration or innovative methodologies for clinical studies in RD (action previewed M1-M12):

- The current methodological standard for clinical trials in rare diseases was reviewed, summarised, and the state of the art was presented to the TFG in the face to face meeting in M4(April 17th-18th 2019) . Possible areas for demonstration projects were identified (Deliverable 20.4,M6) and the organization of the calls initiated aiming to be ready for the calls launch on M9.

- Finally, a request to the EMA for them to appoint a permanent member at the TFG is undergoing in order to establish together the regulatory background of proposed methodologies.

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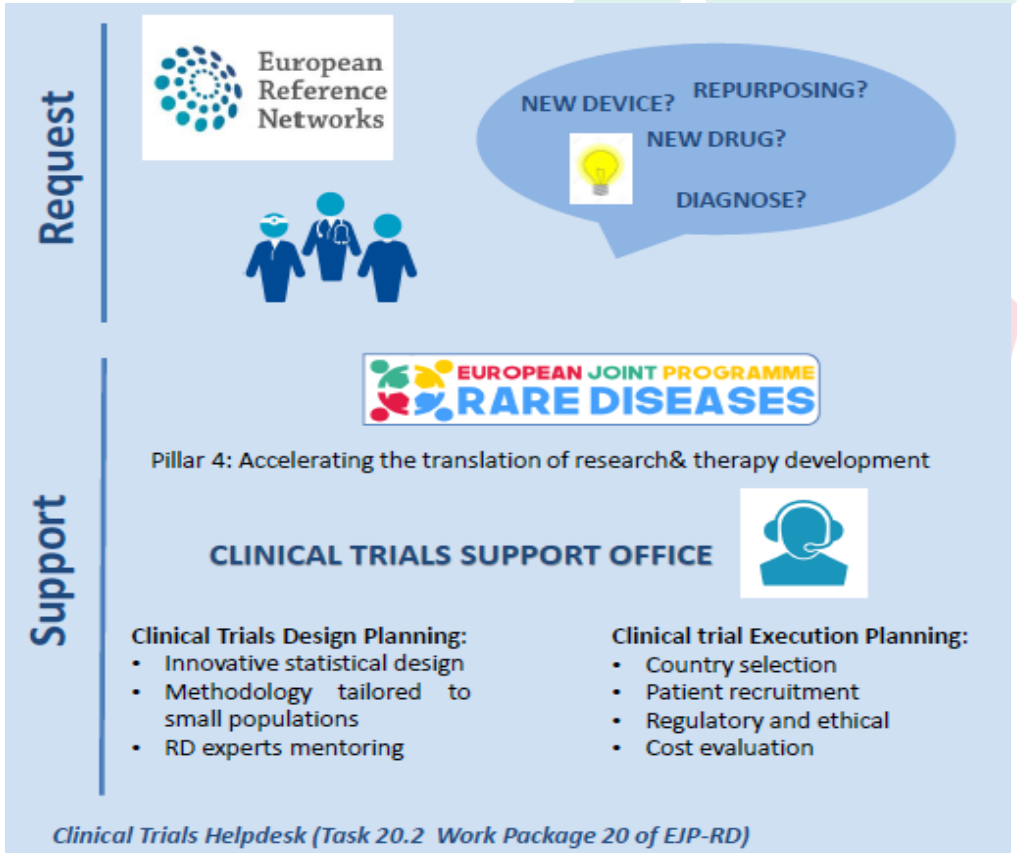
EJP-RD Clinical Study Support Office

Task 20.2 : ***Support in design and planning of RD clinical studies***
Subtask 20.2.1: Establishment of the clinical study support office (M1-M3) :
The support office was established before month 3 (March 18th 2019) (Milestone MS 20.4) of the year 1. A meeting (Teleconference) with all Task members was held by ECRIN with the goal of establishing the Terms of Reference of the Clinical Study support office for rare diseases investigators. Scope, specific tasks, eligibility criteria, governance, composition and responsibility and approach were discussed. The "Terms of Reference" (M6, Deliverable 20.1) document describes the mission, composition and activities of the support office, and the eligibility of the investigators requesting support (first restricted to members of ERNs, then expanded to the Pillar 1 calls or projects on clinical trials.



Aim of the office:
The EJP RD **Clinical Study Support Office** will offer support to ERN (European Reference Networks) investigators to plan clinical studies and new diagnosis procedures for the benefit of RD patients.
- Methodology/Trial design
- Operational: Multicenter/ multinational trials

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M7 – M12 Planned Activities

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RARE DISEASES

P4 : activities planned M7-M12

- **WP19 Facilitating partnerships and accelerating translation for higher patient impact**
  - Add resources to the management toolbox, including finalised ACT handbook and Project Management Manual, in the shared online environment
  - Further development of pool of mentors and matching of mentors with projects
  - Innovation Coordinators to work with projects selected
  - Progress work with CureDravet consortium and address requests coming from coordinators (additional indication of interest were received)
  - The evaluation process for 2019 will be established based on the quantity and kind of projects requesting support.
  - Call to industry experts at the multi-stakeholder workshop held for WP8, experts to be included in the panel
  - Finalise map of the PoC funding opportunities
  - Mapping of the need for a follow-on funding platform focused on RDs is ongoing in particular to establish necessary size of investment
  - Brainstorming with contacted stake holders to identify possible models
  - Further discussion with EIF

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P4 : activities planned M7-M12

- ***<u>WP20</u> Accelerating the validation, use and development of innovative methodologies tailored for clinical trials in RDs***
  - **Subtask 20.1.1: Continue activities and communication in the Task Force Group**

The communication will be continued via monthly TC and Face-to-Face meetings (September 16, 2019).

  - **Subtask 20.2.2 : Operation of the clinical study helpdesk (M3-M12)**

Terms of Reference of the Clinical Study Support Office for Rare Diseases will be finalised soon.

  - **Task 20.3 : Demonstration (Validation) projects task (M9-M12)**

  - **Subtask 20.3.1 : Launch of the call for demonstration pilot projects (M9-M12)**

Launch of the internal call for validation of novel methodologies in a 2-step procedure.

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EJP RD
Annual Work Plan
Year 2

EUROPEAN JOINT PROGRAMME
RARE DISEASES

P4 : Annual Work Plan of Year 2

- **Innovation Management Toolbox**
  - Continue to develop and add resources to the online virtual library that was developed and initiated online in Y1, with feedback from users sought and responded to.
- **Assessment of translational research projects**
  - Analysis of the ongoing projects from E-Rare-3 will be concluded, while projects from Pillar 1 will be analysed starting at M12.
- **Real-time mentoring and technical support for translational research projects**
  - Further expansion and development of the pool of mentors.
  - Recruitment of Innovation Coordinators into specific E-Rare 3 projects already selected.
  - Extension and development of mentoring activities.

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P4 : Annual Work Plan of Year 2

- **Support in exploitation and follow-on funding**
  - Implementation of the plans defined in year 1 for those projects funded through E-Rare calls approaching their termination and having reached at least a technology readiness level 4 (TRL4), provided interested PIs request such support and assessment additional E-Rare projects approaching their term, also based on indications from the Innovation Coordinators.
- **Assess potential of projects**
  - Optimisation of evaluation panels (throughout EJP): after a core panel of experts has been identified in year 1,this will constantly be enriched with additional expertise in order to match the pool of projects to be evaluated.
  - Assessment of additional projects (e.g. JTC 2016 and 2017).
- **Development and exploitation plan**
  - Definition of a development and exploitation plan for additional projects assessed, including research activities, Ip strategy unmet needs and sustainability planning.
- **Follow-on Funding**
  - Support in applying to POC funding opportunities and matching of selected projects with POC funding opportunities.
- **Partnering support**
  - Support researchers in the implementation of the development and exploitation plan defined in year 1.
- **Roadmap for a European investment platform for RD**
  - Based on the mapping effort made in year 1, partners will brainstorm with entrepreneurs, venture capital and pharma companies in the space in order to define the most suitable business model and funding scheme.

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P4 : Annual Work Plan of Year 2

- **Subtask 20.1.1: Continue activities and communication in the Task Force Group**
  - The TFG established in Y1 will continue the work on improved methodologies for clinical studies in rare diseases.
  - The work will continue during Y2 by critical revision of the efficiency and updating the process. The communication will be continued via monthly TC meetings and two F2F meetings are foreseen in Y2. The innovation project calls will be organized during Y2.
  - Paper adapted for clinicians on known and adapted methodologies.
- **Subtask 20.1.2 : Disseminate current progress of demonstration pilots on scientific congresses for rare diseases.**
  - Disseminate the activities and results of Y1 findings (EJP RD survey; literature review of state of the art methodology in rare diseases). A dissemination paper is previewed for the state of the art addressing the ready to use methodologies that were already validated. The stakeholders will be informed via presentations at the relevant congress of rare diseases (RE(ACT)-IRDiRC 2020, ECRD) ,and other congresses about the demonstration pilots. A proposition of a cycle of courses to empower the experts and the patients will be proposed for Pillar 3 (WP15 and WP17) aiming at a better dissemination of the new methodologies for providers and users.
- **Subtask 20.2.1: : Operation of the clinical study helpdesk**
  - Within Year 2 the main activity will be personalized contacts with each ERN to evaluate:
    - Their needs in terms of clinical studies
    - Their needs in terms of support / services to study planning and conduct
  - Information will be first gathered from surveys already circulated among ERNs in the frame of previous initiatives.
  - Additional information about the involvement of the ERNs in clinical trials will be evaluated in collaboration with Orphanet database on clinical trials.
  - This overview will be disseminated for all stakeholders.
  - Further needs will be uncovered from the requests and requesters of the general Central and Clinical Trial helpdesk offices.
- **Subtask 20.2.2 : Monitoring activities of the clinical study helpdesk**
  - Between M13 and M24 the support office will continuously work on ERNs' requests under the terms included in the Terms of Reference document. Monitoring of the helpdesk will be done in order to identify trends and gaps on the procedures and activities. Analysis of the performance will allow improvement of the provided support in terms of quality and timing.

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P4 : Annual Work Plan of Year 2

- **Task 20.3 : Demonstration (Validation) projects task**
  - The implementation of the selected demonstration pilots will be done by M15. To tune up the selected demonstration pilots and the potential methodologies to improve the clinical trial set ups a one day symposium will be conducted.
  - The progress of the demonstration pilots will be closely monitored on an every second month basis by the TFG to be able to identify the progress and next steps. As soon as results are available the TFG will be informed by pilot leaders.
  - The TFG will stimulate a layman description of the findings.
- **Subtask 20.4 : Projects on innovative methodologies to improve RD clinical trials in limited populations**
  - The launch of the innovation projects will be done at M15. The implementation of the selected innovation projects will achieved at M21. To tune up the selected innovation projects chosen to improve clinical trial methodology in rare diseases with respect to endpoints, natural history cause, level of evidence assessment, efficient designing of bridging studies and Pharmacometric modeling, a one day symposium will be conducted. The progress of the innovation projects will be closely monitored at least on an every second month basis Initiating at M21 by the TFG.

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How to better involve all countries?

EUROPEAN JOINT PROGRAMME
RARE DISEASES

P4 : how to more involve all European countries in the EJP RD?

- Every Beneficiary of EJP RD has a responsibility to be an **ambassador and champion of EJP**
- Create centrally coordinated **'marketing' campaign**
- Utilise each partner's network to **identify and approach the key stakeholders** in target countries
- Develop **communications materials** to support advocacy process, identify meetings (conferences etc.)
- Ambassadors work with coordination to **meet stakeholders to market EJP**
- **Diffusion** via EMA, C4C,scientific congresses, ERNs etc.

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OPEN DISCUSSION of AWP Year 2

-

Input from Policy Board

EUROPEAN JOINT PROGRAMME
RARE DISEASES

Prioritisation strategy process (Indicators & Methodology)

Eva Bermejo-Sanchez & Manuel Posada (ISCIII, Spain)

EJP RD Policy Board Meeting

Brussels, July 4, 2019

EUROPEAN JOINT PROGRAMME RARE DISEASES

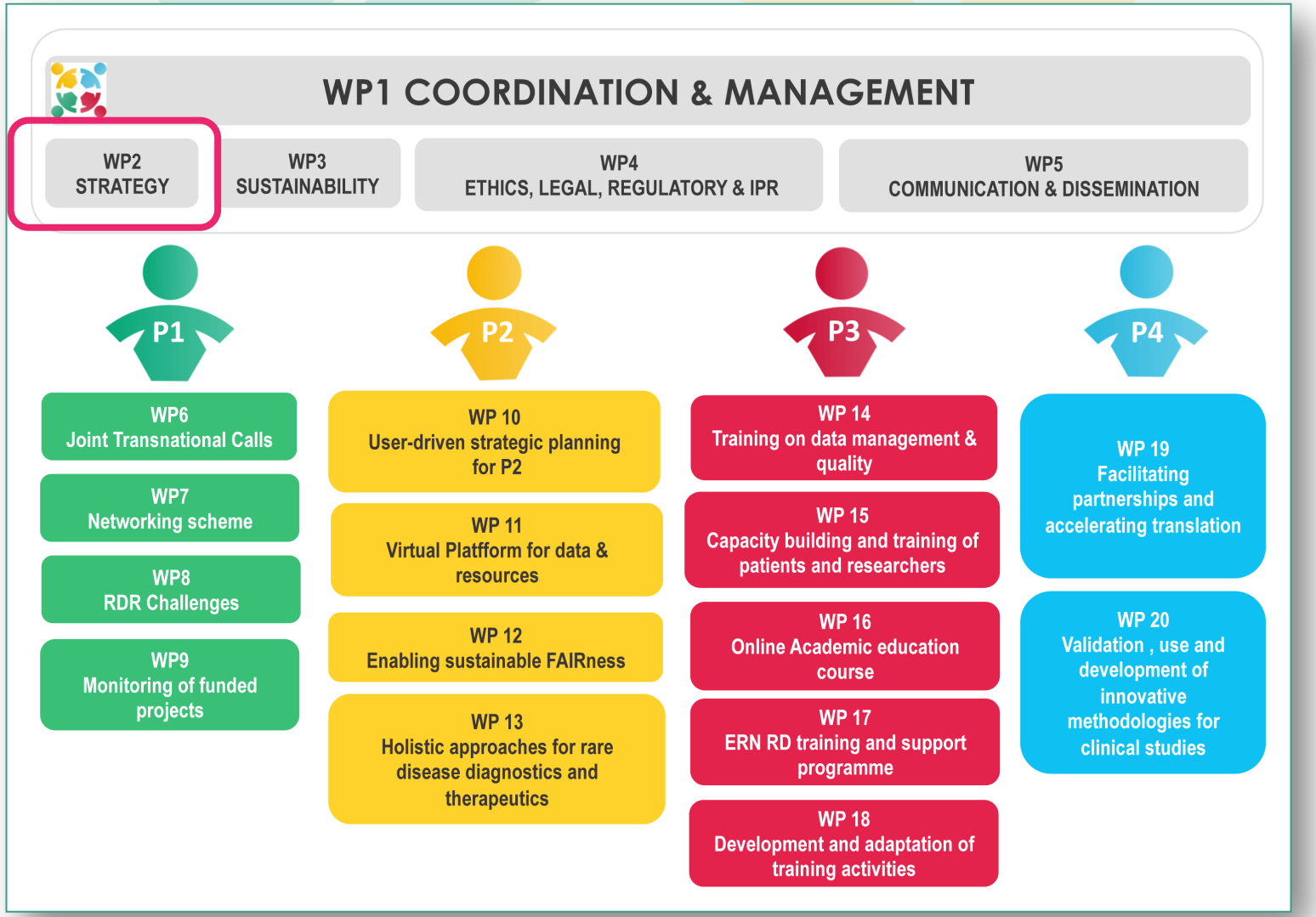
WP2
Integrative Research and Innovation Strategy

WPLs: ISCIII & ISS

Eva Bermejo-Sánchez and Domenica Taruscio

EUROPEAN JOINT PROGRAMME
RARE DISEASES

Work package 2:
Integrative Research and Innovation Strategy



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OBJECTIVES

Work package 2 overall objectives

Goal: This WP focuses on the development of EJP RD research and innovation strategy in connection to all related stakeholders.

Specific objectives:

- To **map the needs** for RD research and innovation;
- To **define the prioritization model for the EJP RD actions** as part of the annual planning and in connection to WP3 (Sustainability) and WP4 (Ethical, Regulatory, Legal and IPR framework);
- To **prioritize topics for the joint transnational calls (JTCs)** to be implemented within Pillar 1;
- To **feed the medium and long-term RD research and innovation strategy** in collaboration with IRDiRC;
- To **prepare a Scoping Paper** to be promoted (and implemented) by the national mirror groups and within the current and forthcoming EC Framework Programme.

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Work package 2 year 1 objectives

- To **map the research and innovation needs**
- To **define the prioritization model for the EJP RD actions**;
- To **prioritize topics for the joint transnational calls (JTCs)** to be implemented within Pillar 1;
- To **build a list of R&I needs requiring a medium and long-term approach** and **related Task Forces**
- To **prepare a Scoping Paper** to be promoted (and implemented) by the national mirror groups and within the current and forthcoming EC Framework Programme.

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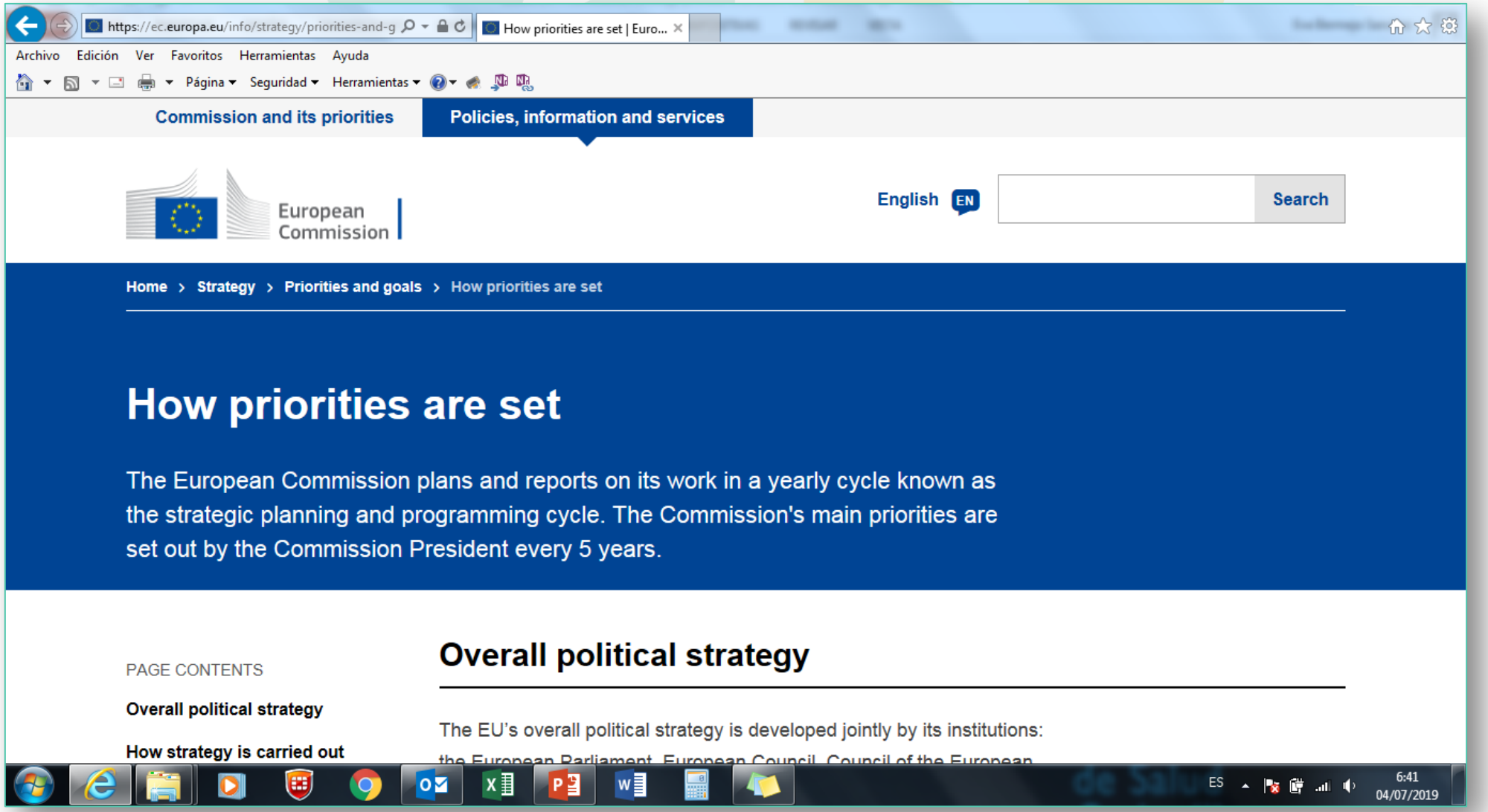
Prioritization is not anything new



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Need for prioritisation exercise

- A **prioritization scheme is necessary everywhere**, as **resources are never unlimited**, and is aimed to select among different options in order to address the most important needs and to facilitate decisions about further development of activities on the basis of the outcome reached.

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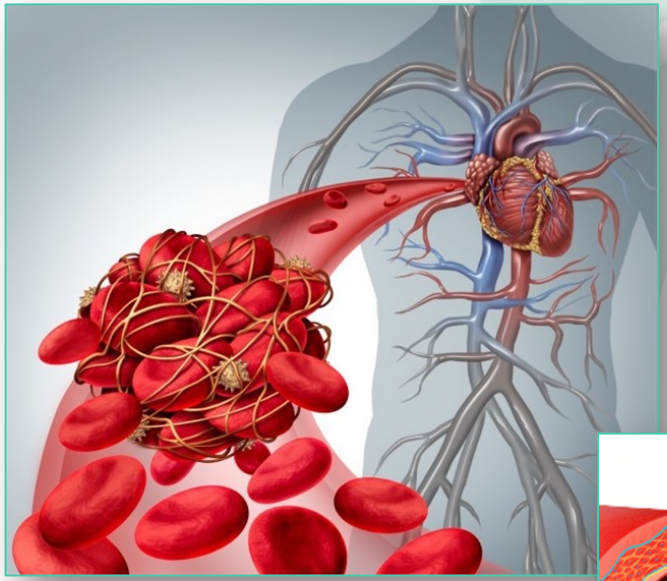
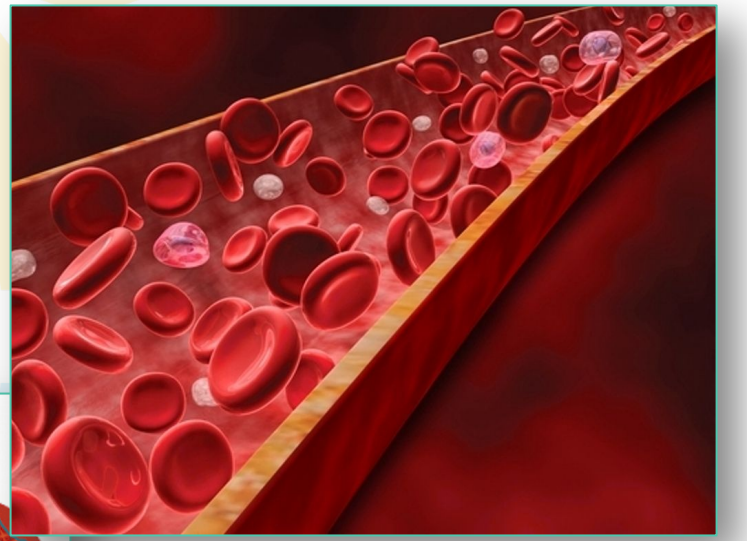
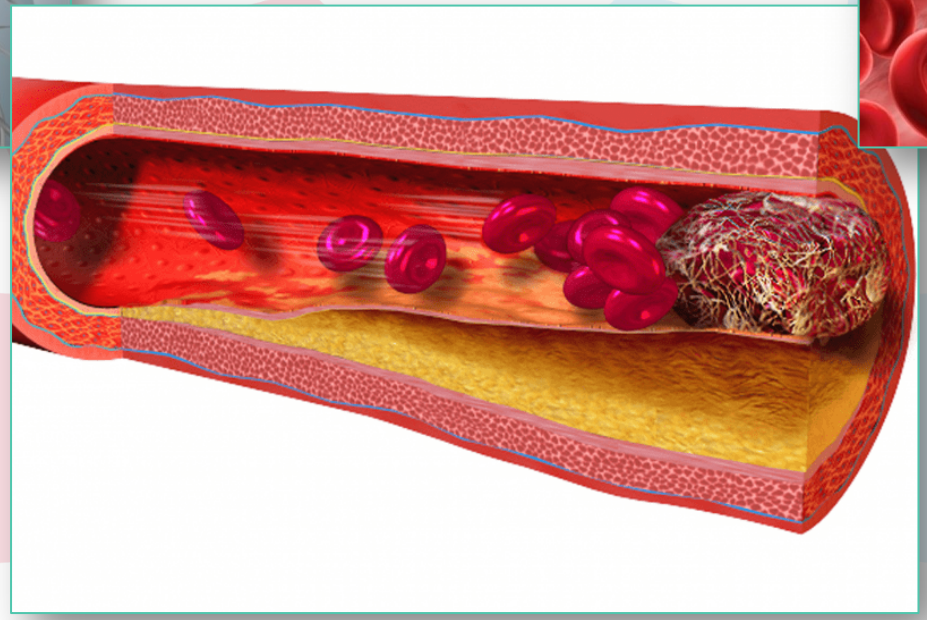
Need for prioritisation exercise







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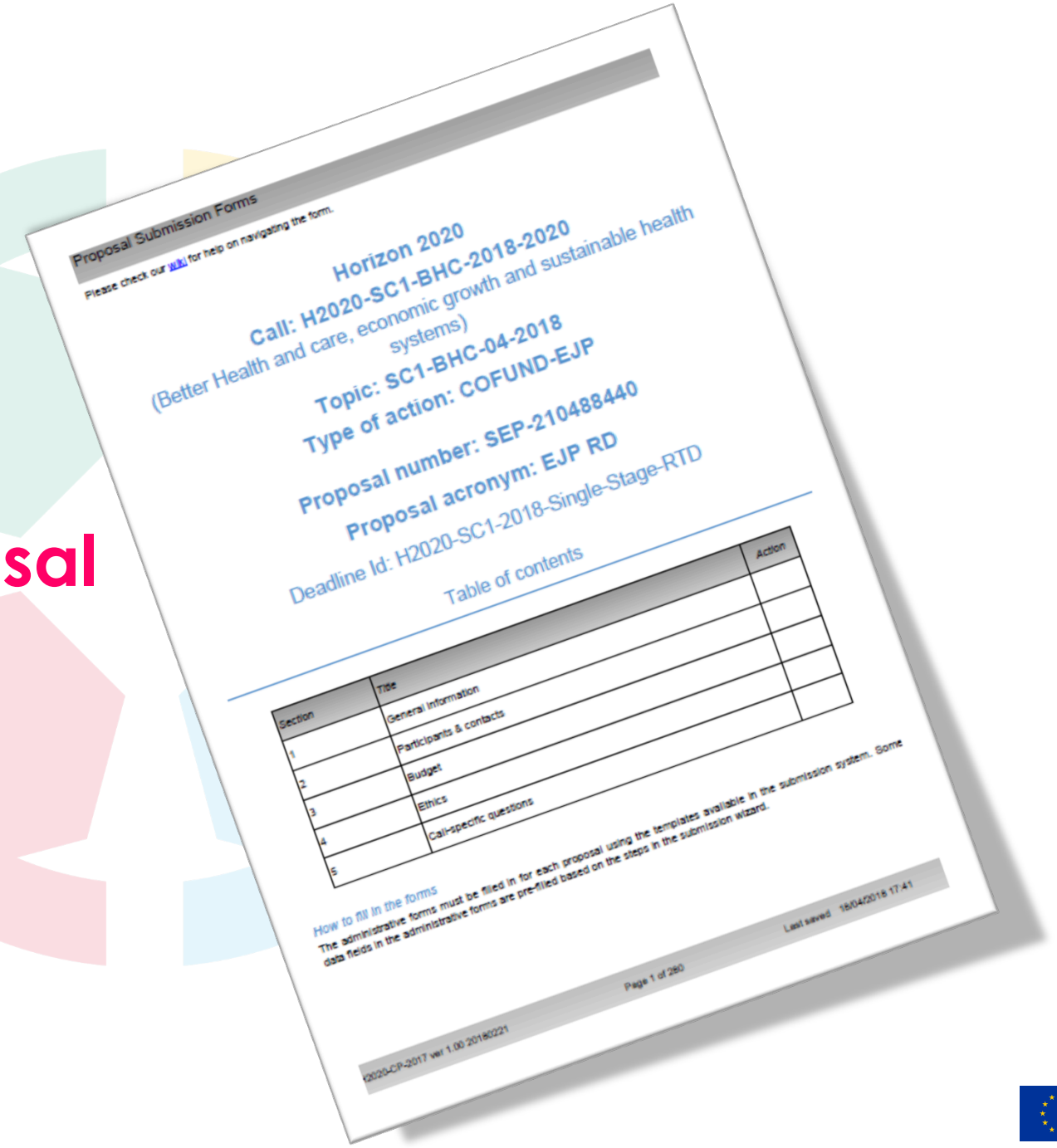
Prioritisation strategy process

EUROPEAN JOINT PROGRAMME
RARE DISEASES

EJP RD – Approved Proposal



Proposal Submission Forms

Please check our wiki for help on navigating the form.

Horizon 2020

Call: H2020-SC1-BHC-2018-2020

(Better Health and care, economic growth and sustainable health systems)

Topic: SC1-BHC-04-2018

Type of action: COFUND-EJP

Proposal number: SEP-210488440

Proposal acronym: EJP RD

Deadline Id: H2020-SC1-2018-Single-Stage-RTD

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How to fill in the forms

The administrative forms must be filled in for each proposal using the templates available in the submission system. Some data fields in the administrative forms are pre-filled based on the steps in the submission wizard.

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Deliverables in the Proposal

Deliverables:

D2.1: Final list of prioritization criteria
D2.2: Prioritization scheme including decision-making process
D2.3: Summary document on mapped research and innovation needs
D2.4: Scoping paper
D2.5: List of refined JTC topics
D2.6: List of research and innovation needs requiring medium- or long-term approach and related Task Forces

D2.7: Analysis of national state of play and alignment process with EJP RD
D2.8: Report from strategic workshop with national policy makers

EJP RD

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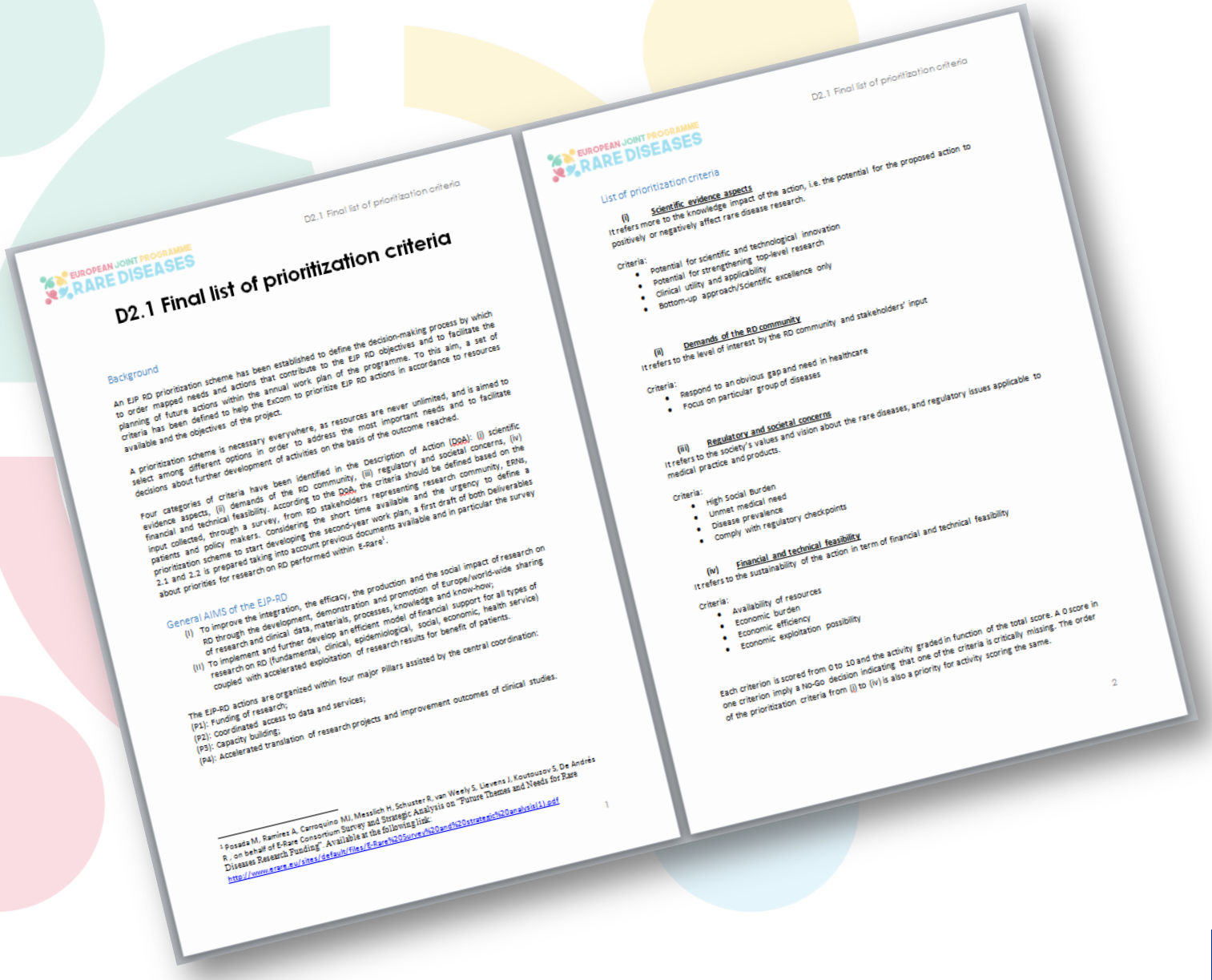
D2.1 – Final list of prioritization criteria



D2.1 Final list of prioritization criteria

EUROPEAN JOINT PROGRAMME RARE DISEASES

D2.1 Final list of prioritization criteria

Background

An EJP RD prioritization scheme has been established to define the decision-making process by which to order mapped needs and actions that contribute to the EJP RD objectives and to facilitate the planning of future actions within the annual work plan of the programme. To this aim, a set of criteria has been defined to help the ExCom to prioritize EJP RD actions in accordance to resources available and the objectives of the project.

A prioritization scheme is necessary everywhere, as resources are never unlimited, and is aimed to select among different options in order to address the most important needs and to facilitate decisions about further development of activities on the basis of the outcome reached.

Four categories of criteria have been identified in the Description of Action (DoA): (i) scientific evidence aspects, (ii) demands of the RD community, (iii) regulatory and societal concerns, (iv) financial and technical feasibility. According to the DoA, the criteria should be defined based on the input collected, through a survey, from RD stakeholders representing research community, ERNs, patients and policy makers. Considering the short time available and the urgency to define a prioritization scheme to start developing the second-year work plan, a first draft of both Deliverables 2.1 and 2.2 is prepared taking into account previous documents available and in particular the survey about priorities for research on RD performed within E-Rare[1].

General AIMS of the EJP-RD

(I) To improve the integration, the efficacy, the production and the social impact of research on RD through the development, demonstration and promotion of Europe/world-wide sharing of research and clinical data, materials, processes, knowledge and know-how;
(II) To implement and further develop an efficient model of financial support for all types of research on RD (fundamental, clinical, epidemiological, social, economic, health service) coupled with accelerated exploitation of research results for benefit of patients.

The EJP-RD actions are organized within four major Pillars assisted by the central coordination:
(P1): Funding of research;
(P2): Coordinated access to data and services;
(P3): Capacity building;
(P4): Accelerated translation of research projects and improvement outcomes of clinical studies.

[1] Posada M, Ramírez A, Carroquino MJ, Mesalich H, Schuster R, van Weely S, Lievens J, Koutsouav S, De Andrés R., on behalf of E-Rare Consortium Survey and Strategic Analysis on "Future Themes and Needs for Rare Diseases Research Funding". Available at the following link: http://www.erare.eu/sites/default/files/E-Rare%20Survey%20and%20strategic%20analysis11.pdf

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D2.1 Final list of prioritization criteria

EUROPEAN JOINT PROGRAMME RARE DISEASES

List of prioritization criteria

(i) **Scientific evidence aspects**
It refers more to the knowledge impact of the action, i.e. the potential for the proposed action to positively or negatively affect rare disease research.

Criteria:
- Potential for scientific and technological innovation
- Potential for strengthening top-level research
- Clinical utility and applicability
- Bottom-up approach/Scientific excellence only

(ii) **Demands of the RD community**
It refers to the level of interest by the RD community and stakeholders' input

Criteria:
- Respond to an obvious gap and need in healthcare
- Focus on particular group of diseases

(iii) **Regulatory and societal concerns**
It refers to the society's values and vision about the rare diseases, and regulatory issues applicable to medical practice and products.

Criteria:
- High Social Burden
- Unmet medical need
- Disease prevalence
- Comply with regulatory checkpoints

(iv) **Financial and technical feasibility**
It refers to the sustainability of the action in term of financial and technical feasibility

Criteria:
- Availability of resources
- Economic burden
- Economic efficiency
- Economic exploitation possibility

Each criterion is scored from 0 to 10 and the activity graded in function of the total score. A 0 score in one criterion imply a No-Go decision indicating that one of the criteria is critically missing. The order of the prioritization criteria from (i) to (iv) is also a priority for activity scoring the same.

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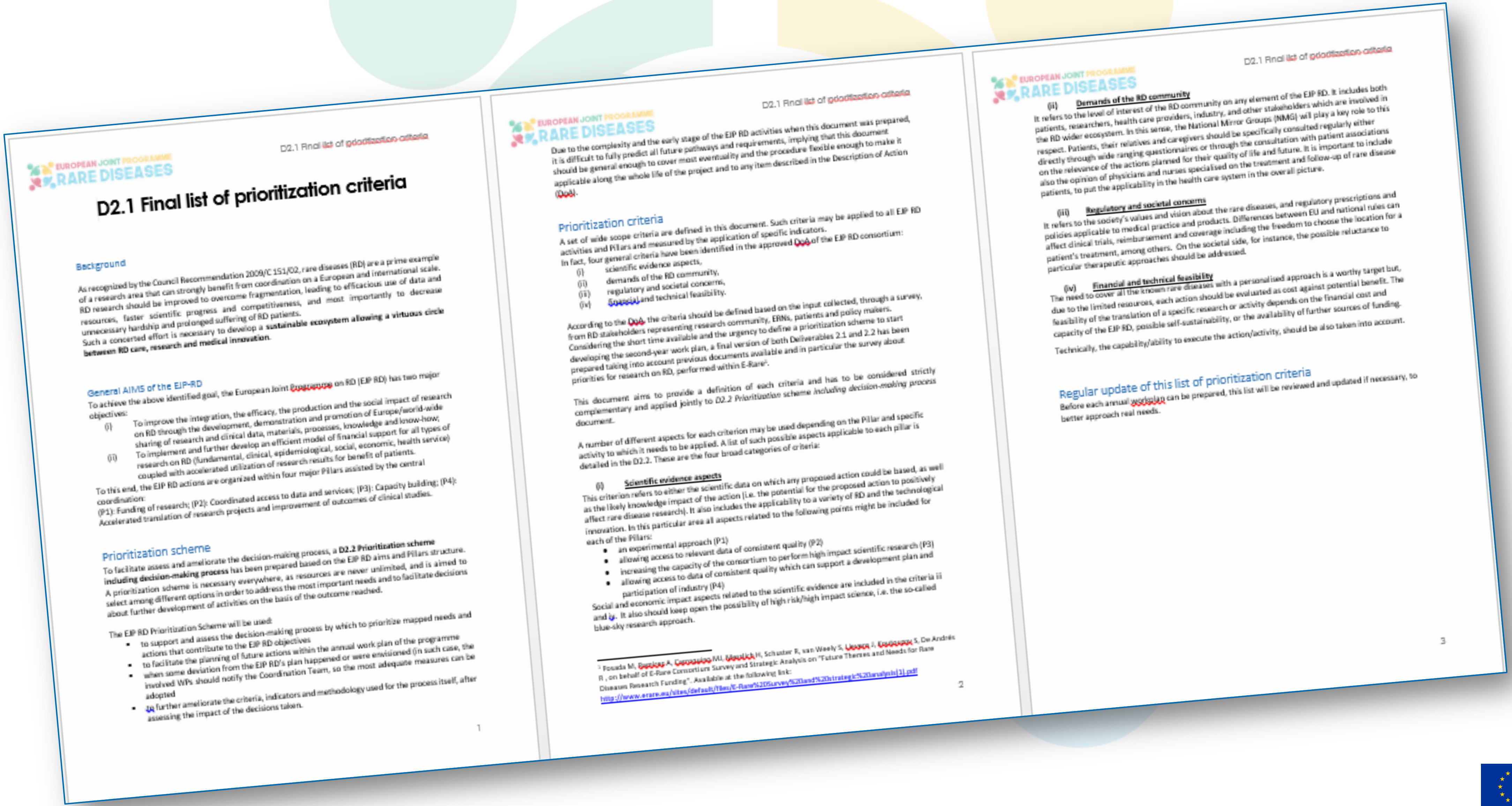
D2.1 – Final list of prioritization criteria-New version


D2.1 Final list of prioritization criteria

EUROPEAN JOINT PROGRAMME RARE DISEASES

D2.1 Final list of prioritization criteria

Background

As recognized by the Council Recommendation 2009/C 151/02, rare diseases (RD) are a prime example of a research area that can strongly benefit from coordination on a European and international scale. RD research should be improved to overcome fragmentation, leading to efficacious use of data and resources, faster scientific progress and competitiveness, and most importantly to decrease unnecessary hardship and prolonged suffering of RD patients. Such a concerted effort is necessary to develop a **sustainable ecosystem allowing a virtuous circle between RD care, research and medical innovation**.

General AIMS of the EJP-RD

To achieve the above identified goal, the European Joint Programme on RD (EJP RD) has two major objectives:

(i) To improve the integration, the efficacy, the production and the social impact of research on RD through the development, demonstration and promotion of Europe/world-wide sharing of research and clinical data, materials, processes, knowledge and know-how;

(ii) To implement and further develop an efficient model of financial support for all types of research on RD (fundamental, clinical, epidemiological, social, economic, health service) coupled with accelerated utilization of research results for benefit of patients.

To this end, the EJP RD actions are organized within four major Pillars assisted by the central coordination: (P1): Funding of research; (P2): Coordinated access to data and services; (P3): Capacity building; (P4): Accelerated translation of research projects and improvement of outcomes of clinical studies.

Prioritization scheme

To facilitate assess and ameliorate the decision-making process, a **D2.2 Prioritization scheme including decision-making process** has been prepared based on the EJP RD aims and Pillars structure. A prioritization scheme is necessary everywhere, as resources are never unlimited, and is aimed to select among different options in order to address the most important needs and to facilitate decisions about further development of activities on the basis of the outcome reached.

The EJP RD Prioritization Scheme will be used:

- to support and assess the decision-making process by which to prioritize mapped needs and actions that contribute to the EJP RD objectives
- to facilitate the planning of future actions within the annual work plan of the programme when some deviation from the EJP RD's plan happened or were envisioned (in such case, the involved WPs should notify the Coordination Team, so the most adequate measures can be adopted
- to further ameliorate the criteria, indicators and methodology used for the process itself, after assessing the impact of the decisions taken.

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D2.1 Final list of prioritization criteria

EUROPEAN JOINT PROGRAMME RARE DISEASES

Due to the complexity and the early stage of the EJP RD activities when this document was prepared, it is difficult to fully predict all future pathways and requirements, implying that this document should be general enough to cover most eventuality and the procedure flexible enough to make it applicable along the whole life of the project and to any item described in the Description of Action (DoA).

Prioritization criteria

A set of wide scope criteria are defined in this document. Such criteria may be applied to all EJP RD activities and Pillars and measured by the application of specific indicators. In fact, four general criteria have been identified in the approved DoA of the EJP RD consortium:

(i) scientific evidence aspects,
(ii) demands of the RD community,
(iii) regulatory and societal concerns,
(iv) financial and technical feasibility.

According to the DoA, the criteria should be defined based on the input collected, through a survey, from RD stakeholders representing research community, ERNs, patients and policy makers. Considering the short time available and the urgency to define a prioritization scheme to start developing the second-year work plan, a final version of both Deliverables 2.1 and 2.2 has been prepared taking into account previous documents available and in particular the survey about priorities for research on RD, performed within E-Rare[1].

This document aims to provide a definition of each criteria and has to be considered strictly complementary and applied jointly to *D2.2 Prioritization scheme including decision-making process* document.

A number of different aspects for each criterion may be used depending on the Pillar and specific activity to which it needs to be applied. A list of such possible aspects applicable to each pillar is detailed in the D2.2. These are the four broad categories of criteria:

(i) **Scientific evidence aspects**

This criterion refers to either the scientific data on which any proposed action could be based, as well as the likely knowledge impact of the action (i.e. the potential for the proposed action to positively affect rare disease research). It also includes the applicability to a variety of RD and the technological innovation. In this particular area all aspects related to the following points might be included for each of the Pillars:

- an experimental approach (P1)
- allowing access to relevant data of consistent quality (P2)
- increasing the capacity of the consortium to perform high impact scientific research (P3)
- allowing access to data of consistent quality which can support a development plan and participation of industry (P4)

Social and economic impact aspects related to the scientific evidence are included in the criteria ii and iv. It also should keep open the possibility of high risk/high impact science, i.e. the so-called blue-sky research approach.

[1] Posada M, ... on behalf of E-Rare Consortium Survey and Strategic Analysis on "Future Themes and Needs for Rare Diseases Research Funding". Available at the following link: http://www.erare.eu/sites/default/files/E-Rare%20Survey%20and%20strategic%20analysis(1).pdf

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D2.1 Final list of prioritization criteria

EUROPEAN JOINT PROGRAMME RARE DISEASES

(ii) **Demands of the RD community**

It refers to the level of interest of the RD community on any element of the EJP RD. It includes both patients, researchers, health care providers, industry, and other stakeholders which are involved in the RD wider ecosystem. In this sense, the National Mirror Groups (NMG) will play a key role to this respect. Patients, their relatives and caregivers should be specifically consulted regularly either directly through wide ranging questionnaires or through the consultation with patient associations on the relevance of the actions planned for their quality of life and future. It is important to include also the opinion of physicians and nurses specialised on the treatment and follow-up of rare disease patients, to put the applicability in the health care system in the overall picture.

(iii) **Regulatory and societal concerns**

It refers to the society's values and vision about the rare diseases, and regulatory prescriptions and policies applicable to medical practice and products. Differences between EU and national rules can affect clinical trials, reimbursement and coverage including the freedom to choose the location for a patient's treatment, among others. On the societal side, for instance, the possible reluctance to particular therapeutic approaches should be addressed.

(iv) **Financial and technical feasibility**

The need to cover all the known rare diseases with a personalised approach is a worthy target but, due to the limited resources, each action should be evaluated as cost against potential benefit. The feasibility of the translation of a specific research or activity depends on the financial cost and capacity of the EJP RD, possible self-sustainability, or the availability of further sources of funding. Technically, the capability/ability to execute the action/activity, should be also taken into account.

Regular update of this list of prioritization criteria

Before each annual workplan can be prepared, this list will be reviewed and updated if necessary, to better approach real needs.

3

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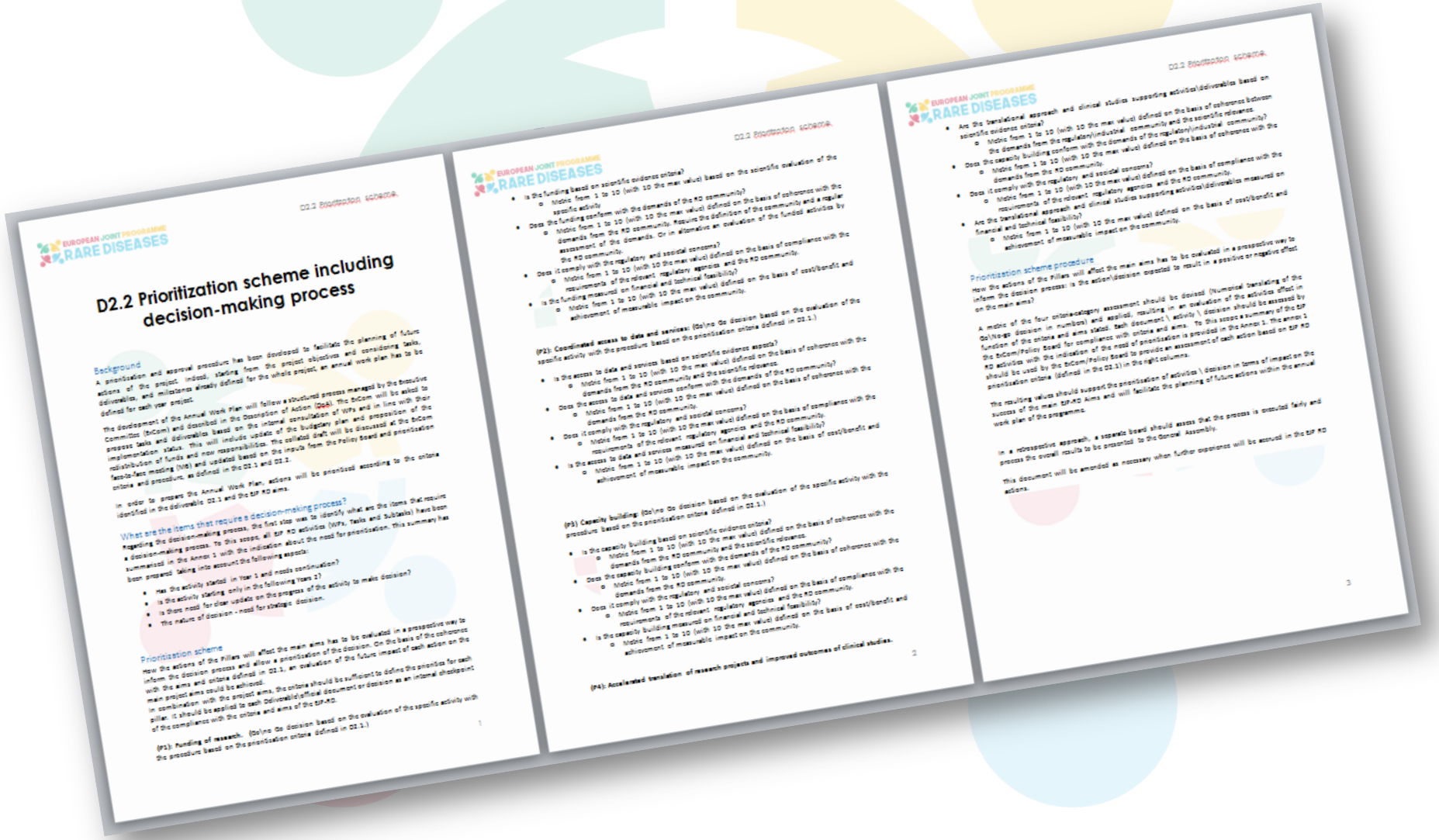
D2.2 – Prioritization scheme including decision-making process



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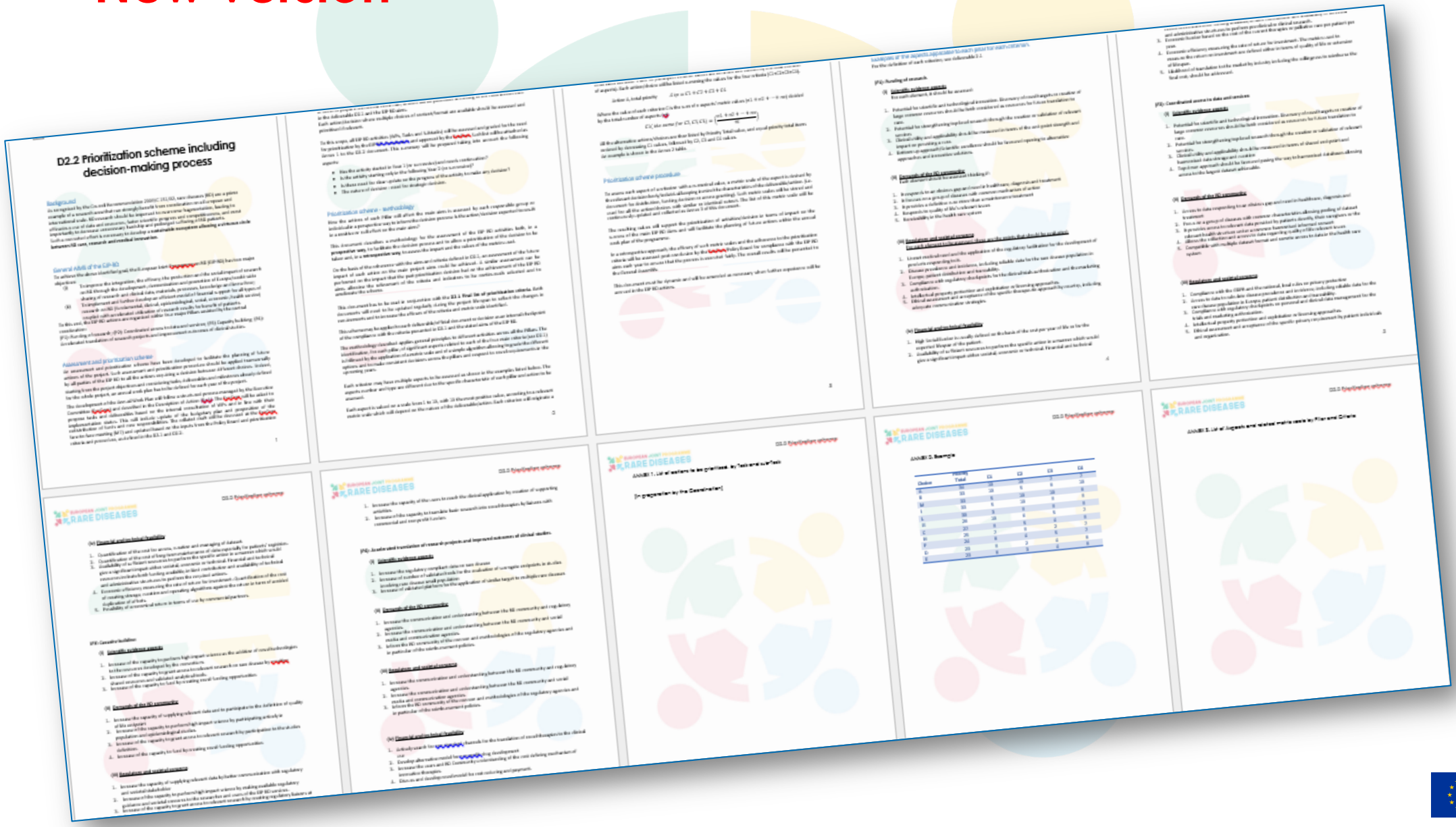
D2.2 – Prioritization scheme including decision-making process

New version



The Executive Committee agreed
to name it:

Guidelines for prioritization

EUROPEAN JOINT PROGRAMME
RARE DISEASES

Good News for the Policy Board

EJP RD Gantt Chart

TASK	Start	End	YEAR 1	YEAR 2	YEAR 3	YEAR 4	YEAR 5
WP1. Coordination and management							
Task 1.1 Implementation and responsibilities of the coordination office (MS 1.1;1.2;1.3)	1	60	M M				
Task 1.2 Coordination & support within and across pillars	1	60					
Task 1.3 Monitoring of the EJP RD activities and achievements	1	60					
Task 1.4 Data management plan	1	60					
WP2. Integrative research and innovation strategy							
Task 2.1 Prioritization scheme for EJP RD actions (MS 2.1;2.2)	1	36	M M				
Task 2.2 Mapping the research and innovation need (MS2.3)	1	48	M				
Task 2.3 Scientific programming of joint transnational calls	1	36					
Task 2.4 Management of the medium, longer-term research strategy questions and dedicated linkage with Task Forces of IRDiRC	1	60					
Task 2.5 Translation/impact of prioritization on national and EU strategies	6	60					
WP3. Sustainability strategy and business plan							
Task 3.1 Service roadmap alignment of the needs, expectations and engagement of the different RD research stakeholders in Europe with respect to EJP RD sustainability	1	36					
Task 3.2 Preparation of the sustainable EJP-RD services catalogue, supporting the EJP-RD dissemination and communication activities	12	36					
Task 3.3 Preparation of the EJP-RD sustainability plan with business plan (MS3.1)	1	42				M	
Task 3.4 Sustainability roadmap (MS3.2)	37	60					M
WP4. Ethical, regulatory, legal and IPR framework of the EJP RD							
Task 4.1 Setting up the 'Advisory Regulatory Ethics Board (MS4.1)	1	60	M				
Task 4.2 Managing ethical and regulatory issues (MS4.2)	1	60	M	M	M	M	
Task 4.3 Managing legal and IPR issues (MS4.3)	1	60	M				
WP5. Communication & dissemination							
Task 5.1 EJP RD external communication & dissemination of results (MS5.1)	1	60	M				
Task 5.2 Integrating EJP-RD communication and dissemination strategies with the strategies of involved stakeholders (MS5.2;5.3)	1	60	M M	M	M	M	
WP6. Joint Transnational Calls for collaborative research projects							
Task 6.1 1st co-funded Joint Transnational Call (JTC 2019)	1	12					
Task 6.2 2nd co-funded Joint Transnational Call (JTC 2020)	4	24					
Task 6.3 3rd Joint Transnational Call (JTC 2021) not co-funded by the EC	16	36					
Task 6.4 4th Joint Transnational Call (JTC 2022) not co-funded by the EC	28	48					
WP7. Networking to share knowledge on rare diseases							
Task 7.1 Development of the networking scheme (MS7.1)	1	7	M				
Task 7.2 Application and evaluation of proposals for the Networking Scheme	7	60					
Task 7.3 Early assessment of the Networking scheme	21	24					
WP8. Rare Disease Research Challenges							
Task 8.1 Development of the scheme (MS8.1)	1	8	M				
Task 8.2 Identification of challenges	9	12					
Task 8.3 The call for projects, eligibility criteria and evaluation	13	52					
Task 8.4 Management of the funding resources, establishment of contracts and distribution of funding to selected projects	17	50					
WP9. Monitoring of funded projects							
Task 9.1 Monitoring of Joint Transnational Calls for collaborative research projects (MS9.1)	1	60	M				
Task 9.2 Monitoring of networking to share knowledge on rare diseases (MS9.2)	1	60	M				
Task 9.3 Monitoring of Rare Disease Research Challenge (MS9.3)	17	56					M
WP10. User-driven strategic planning and transversal activities for Pillar 2 data ecosystem							
Task 10.1 Convene yearly retreats leading to Pillar 2 annual strategic plans (MS10.1)	1	60	M	M	M	M	M
Task 10.2 Coordinating Pillar 2 Work Packages outputs and aligning to RD community needs (MS10.2)	1	60	M	M	M	M	M
Task 10.3 Coordination of technical GDPR implementation	1	60					
Task 10.4 Quality oversight	1	60					
Task 10.5 Sustainability and scaling up (MS10.3;10.4)	1	60		M			

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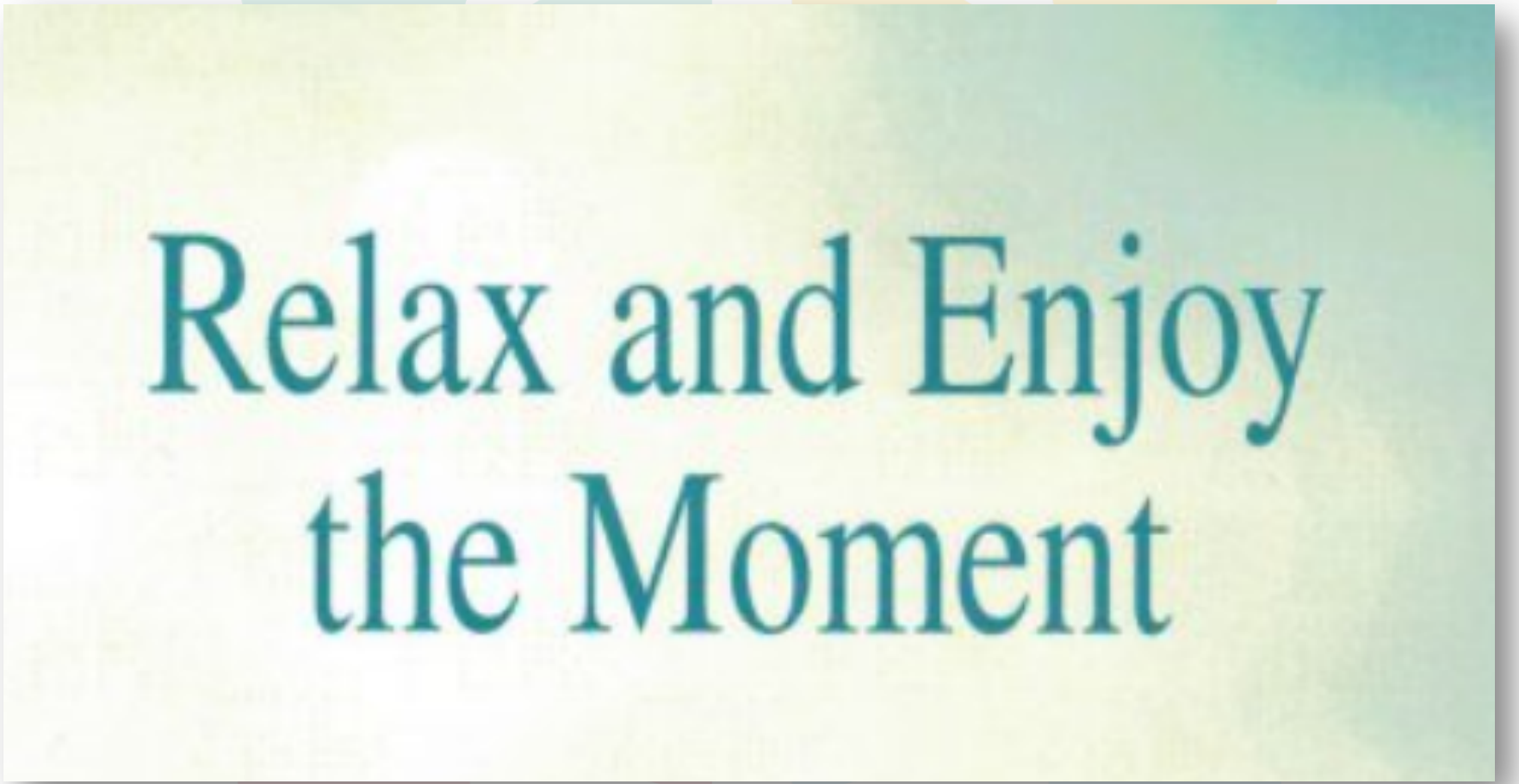

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Guidelines for prioritization

According to the Proposal, **some criteria will be defined** based on the input collected (through a survey) from RD stakeholders representing research community, ERNs, patients and policy makers.

For this first year, the criteria and scheme for prioritization were defined taking into account previous documents available, and this will be updated when the survey can be addressed to all the stakeholders

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Guidelines for prioritization – They will be used:

- to support and assess the **decision-making process** by which to prioritize mapped needs and actions that contribute to the EJP RD objectives
- to facilitate the **planning of future actions** within the annual work plan of the programme
- when some **deviation from the EJP RD's plan** happened or were envisioned (in such case, the involved WPs should notify the Coordination Team, so the most adequate measures can be adopted
- to **further ameliorate the criteria, indicators and methodology** used for the process itself, after assessing the impact of the decisions taken.

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Guidelines for prioritization

Due to the **complexity and the early stage of the EJP RD** activities when this document was prepared:

- It is **difficult to fully predict** all future pathways and requirements
- This implies that **this document should be general enough** to cover most eventuality **and the procedure flexible enough** to make it applicable along the whole life of the project and to any item included in the Description of Action (DoA).

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Guidelines for prioritization

Four wide scope categories of criteria:

- Scientific evidence and innovation
- Demands of the RD community
- Regulatory and societal concerns
- Financial and technical feasibility, including sustainability

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Guidelines for prioritization

Regular update of the list of prioritization criteria

Before each annual workplan can be prepared, this list will be reviewed (and updated if necessary), to **better approach real needs**

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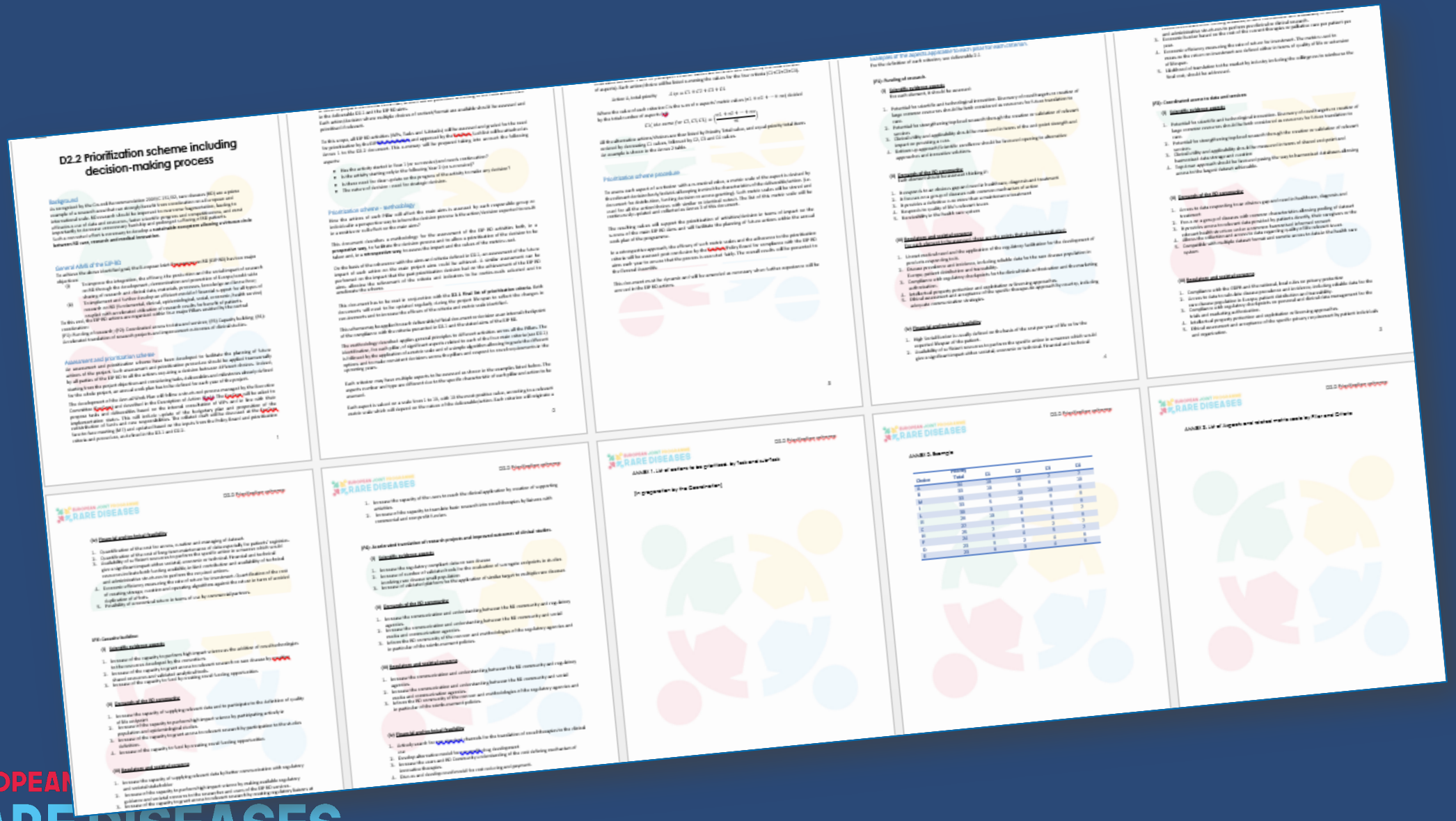
D2.2 – Prioritization scheme including decision-making process



EUROPEAN RARE DISEASES

Guidelines for prioritization

An assessment and prioritization scheme have been developed:

- To **facilitate the planning of future actions** of the project.
- **It should be applied transversally** by all parties of the EJP RD to all the actions requiring a decision between different choices.
- Indeed, starting from the project objectives and considering tasks, deliverables and milestones already defined for the whole project, an **annual work plan has to be defined for each year of the project**.

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Guidelines for prioritization – Transversally applicable

3.1a. List of workpackages

TITLE	LEAD BENEFICIARY
COORDINATION & TRANSVERSAL ACTIVITIES	INSERM (Coo)
WP1. Coordination and management	INSERM (Coo)
WP2. Integrative research and innovation strategy	ISCIII, ISS
WP3. Sustainability strategy and business plan	INSERM (RaDiCo), EATRIS, ISCIII
WP4. Ethical, regulatory, legal and IPR framework of the EJP RD	FGB
WP5. Communication & dissemination	INSERM (Coo)
PILLAR 1: RESEARCH COLLABORATIVE FUNDING	DLR, ZonMw
WP6. Joint Transnational Calls for collaborative research projects	DLR
WP7. Networking to share knowledge on rare diseases	ZonMw
WP8. Rare Disease Research Challenges	FFRD
WP9. Monitoring of funded projects	CSO/MOH
PILLAR 2: INNOVATIVE COORDINATED ACCESS TO DATA AND SERVICES FOR TRANSFORMATIVE RARE DISEASES RESEARCH	INSERM (Orphanet), UKL-HD
WP10. User-driven strategic planning and transversal activities for Pillar 2 data ecosystem	INSERM (Orphanet), ULEIC
WP11. Common virtual platform for discoverable data and resources for RD research	INSERM (Orphanet), CNAG
WP12. Enabling sustainable FAIRness and Federation at the record level for RD data, patients and samples	ULEIC, LUMC
WP13. Enabling multidisciplinary, holistic approaches for rare disease diagnostics and therapeutics	UM, UKL-HD
PILLAR 3: CAPACITY BUILDING AND EMPOWERMENT	EURORDIS, VUHSK
WP14. Training on data management & quality	ISS
WP15. Capacity building and training of patients and researchers in Rare Disease research and processes	EURORDIS
WP16. Online Academic education course	FFRD
WP17. ERN RD training and support programme	EKUT
WP18. Development and adaptation of training activities	VUHSK, CMHI, EURORDIS
PILLAR 4: ACCELERATING THE TRANSLATION OF HIGH POTENTIAL PROJECTS AND IMRPOVING OUTCOMES OF CLINICAL STUDIES IN SMALL POPULATIONS	APHP, EATRIS
WP19. Facilitating partnerships and accelerating translation for higher patient impact	EATRIS, FTELE
WP20. Accelerating the validation, use and development of innovative methodologies tailored for clinical trials in RDs	UKA, APHP, HSK (MetabERN)

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Prioritization scheme

- How the actions of the Pillars will affect the main aims has to be **evaluated in a prospective way** to inform the decision process and allow a prioritization of the decision.

- In combination with the project aims, **the criteria should be sufficient to define the priorities for each pillar.**

- It should be **applied to each Deliverable/official document or decision** as an internal checkpoint of the compliance with the criteria and aims of the EJP-RD.

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What are the items that require a decision-making process?

All EJP RD activities (WPs, Tasks and Subtasks) are being summarized with the indication about the need for prioritization.

This summary is being prepared taking into account the following aspects:

- Has the activity started in Year 1 and needs continuation?
- Is the activity starting only in the following Year 2?
- Is there need for clear update on the progress of the activity to make decision?
- The nature of decision - need for strategic decision.

This procedure is applicable for all the years of the project

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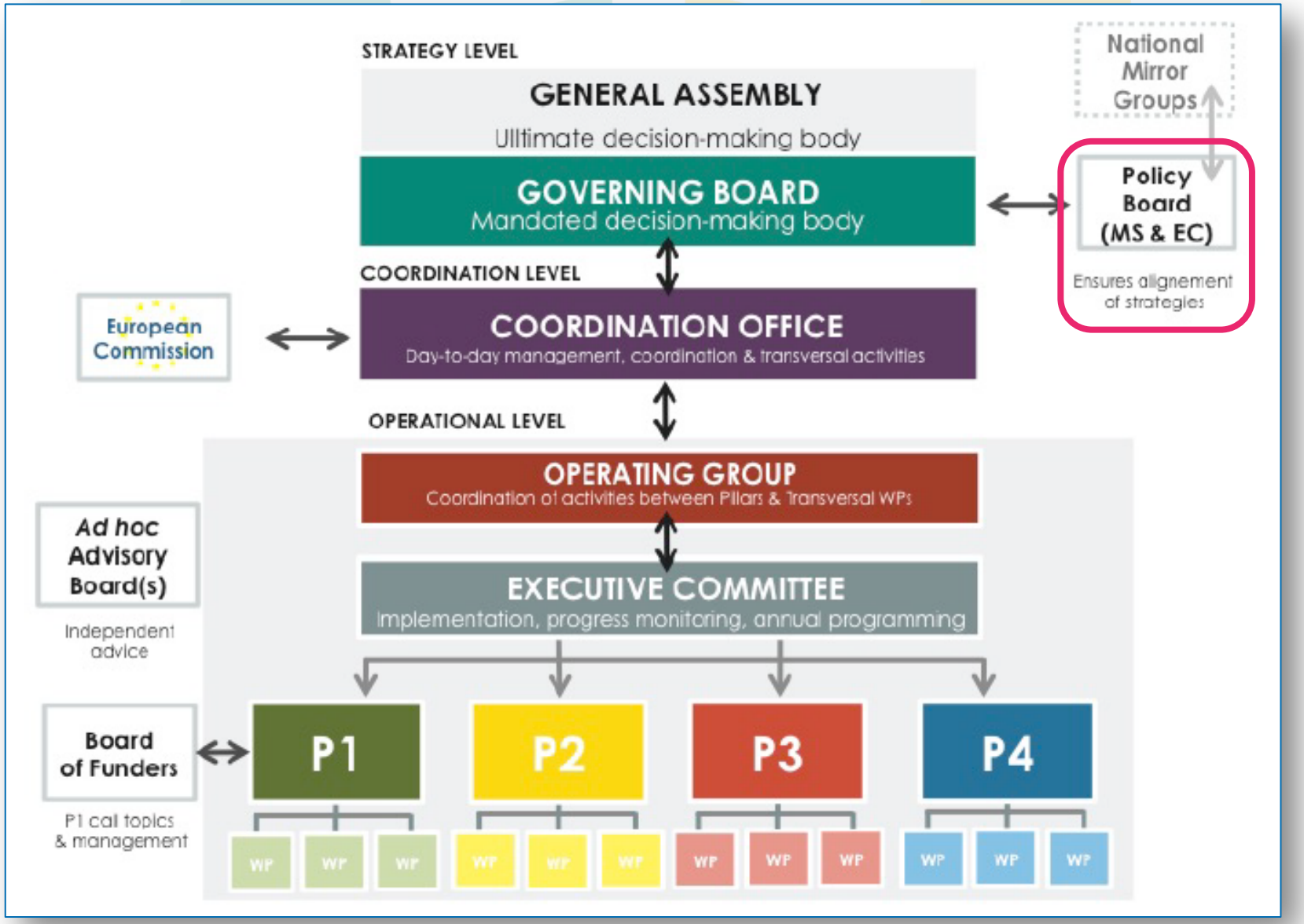
Decision-making process



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Decision-making process
Annual Work Plan – Prioritisation

Coordination

- The development of the **Annual Work Plan** will follow a structured process initiated and managed from the Coordination and the Executive Committee (ExeCom)

Executive Committee

- The ExeCom will be asked to **propose tasks and deliverables** based on the internal consultation of WPs and in line with their implementation status. This will include, if necessary, **update of the budgetary plan** and proposition of the redistribution of funds and new responsibilities.

Policy Board

- The collated draft will be **discussed at the ExeCom face-to-face meeting (M7)** and updated based on the **inputs from the Policy Board** and prioritization guidelines, as defined in the D2.1 and D2.2.

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Decision-making process - Prioritisation

Criteria

- Four categories

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Guidelines for prioritization

Four wide scope categories of criteria:

- Scientific evidence and innovation
- Demands of the RD community
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- Financial and technical feasibility, including sustainability

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Decision-making process - Prioritisation

Criteria

- Four categories

Aspects in each Pillar/ WP/T/ST

- Variable number

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Prioritisation - Scoring

Criteria

Action A, total priority:

$$A\,tp = C1 + C2 + C3 + C4$$

Aspects in each Pillar/ WP/T/ST

$$C1 = (n1 + n2 + \ldots + nx/nt)$$

$$(\textit{the same for } C2, C3, C4)$$

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Prioritisation - Scoring

EUROPEAN JOINT PROGRAMME RARE DISEASES

D2.2 Prioritization scheme

ANNEX 2. Example

Choice	Priority Total	C1	C2	C3	C4
A	34	10	10	7	7
B	33	10	5	8	10
M	33	5	10	10	8
I	33	5	10	9	9
L	30	3	9	9	9
G	28	10	6	5	7
C	27	9	5	4	9
H	25	7	9	2	7
F	24	8	4	5	7
D	23	9	2	4	8
E	23	8	3	4	8

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Prioritisation – Scoring – According to activities

Examples of the aspects applicable to each pillar for each criterion.

For the definition of each criterion, see deliverable 2.1

(P1): Funding of research.

(i) Scientific evidence aspects

For each element, it should be assessed:

1. Potential for scientific and technological innovation. Discovery of novel targets or creation of large common resources should be both considered as resources for future translation to care.
2. Potential for strengthening top-level research through the creation or validation of relevant services
3. Clinical utility and applicability should be measured in terms of the end-point strength and impact on providing a cure.
4. Bottom-up approach/Scientific excellence should be favoured opening to alternative approaches and innovative solutions.

(ii) Demands of the RD community

Each element should be assessed thinking if:

1. It responds to an obvious gap and need in healthcare, diagnosis and treatment
2. It focuses on a group of diseases with common mechanism of action
3. It provides a definitive cure more than a maintenance treatment
4. Responds to quality of life's relevant issues
5. Receivability in the health care system

(iii) Regulatory and societal concerns

For each element to be assessed, these are the points that should be evaluated:

1. Unmet medical need and the application of the regulatory facilitation for the development of products responding to it.
2. Disease prevalence and incidence, including reliable data for the rare disease population in Europe, patient distribution and traceability.
3. Compliance with regulatory checkpoints for the clinical trials authorization and the marketing authorization.
4. Intellectual property protection and exploitation or licensing approaches.
5. Ethical assessment and acceptance of the specific therapeutic approach by country, including adequate communication strategies.

(iv) Financial and technical feasibility

1. High Social Burden is usually defined on the basis of the cost per year of life or for the expected lifespan of the patient.
2. Availability of sufficient resources to perform the specific action in a manner which would give a significant impact either societal, economic or technical. Financial and technical

EUROPEAN JOINT PROGRAMME RARE DISEASES D2.2 Prioritization scheme

resources indicate both funding available, in-kind contribution and availability of technical and administrative structures to perform pre-clinical or clinical research.

3. Economic burden based on the cost of the current therapies or palliative care per patient per year.
4. Economic efficiency measuring the rate of return for investment. The metrics used to measure the return on investment are defined either in terms of quality of life or extension of lifespan.
5. Likelihood of translation to the market by industry including the willingness to reimburse the final cost, should be addressed.

(P2): Coordinated access to data and services:

(i) Scientific evidence aspects

1. Potential for scientific and technological innovation. Discovery of novel targets or creation of large common resources should be both considered as resources for future translation to care.
2. Potential for strengthening top-level research through the creation or validation of relevant services.
3. Clinical utility and applicability should be measured in terms of shared end-point and harmonised data storage and curation
4. Top-down approach should be favoured paving the way to harmonised databases allowing access to the largest dataset achievable.

(ii) Demands of the RD community

1. Access to data responding to an obvious gap and need in healthcare, diagnosis and treatment
2. Focus on a group of diseases with common characteristics allowing pooling of dataset
3. It provides access to relevant data provided by patients directly, their caregivers or the relevant health structure under a common harmonised informed consent
4. Allows the collection and access to data regarding quality of life relevant issues
5. Compatible with multiple dataset format and remote access to data in the health care system

(iii) Regulatory and societal concerns

1. Compliance with the GDPR and the national, local rules on privacy protection
2. Access to data to calculate disease prevalence and incidence, including reliable data for the rare disease population in Europe, patient distribution and traceability.
3. Compliance with regulatory checkpoints on personal and clinical data management for the trials and marketing authorization.
4. Intellectual property protection and exploitation or licensing approaches.
5. Ethical assessment and acceptance of the specific privacy requirement by patient individuals and organization.

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Guidelines for prioritization

- **Prospective assessment** to inform the decision process
- To **assess each aspect of a criterion with a numerical value**, a metric scale of the aspect is devised by the relevant decision body/individual keeping in mind the characteristics of the deliverable/action. (i.e. document for distribution, funding decision or access granting). Such metric scales will be stored and used for all the action/choices with similar or identical output.
- The **resulting values will support the prioritization of activities/decision** in terms of impact on the success of the main EJP RD Aims and will facilitate the planning of future actions within the annual work plan of the programme.
- In a **retrospective approach**, the efficacy of such metric scales and the adherence to the prioritization criteria will be assessed post conclusion by the ExeCom/Policy Board for compliance with the EJP RD aims each year to ensure that the process is executed fairly. The overall results could be presented to the General Assembly.
- This **document must be dynamic** and will be amended as necessary when further experience will be accrued in the EJP RD actions.

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What are the items that require a decision-making process?

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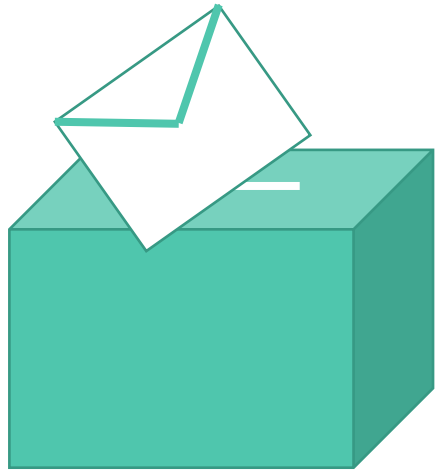
Guidelines for prioritization



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Next actions on this Summary on Research & Innovation Needs

- Once constituted, **the Policy Board** will receive this document in order to deliberate, prioritize (according to prioritization scheme developed under Task 2.1) and **issue recommendations to the EJP RD Executive Committee on the actions to implement within subsequent Annual Work Plan** and the ones that require medium and longer-term investigation.
- Also, the **National Mirror Groups**, once constituted, will be consulted on R&I needs so their input can be implemented to this Summary.
- In addition to **informing Annual Work Plan**, the outcomes of the mapping and prioritization will be reformulated in the form of **Scoping Paper** (prepared by the Task 2.2 leaders with help of the Coordination) to be transmitted to the leaders of Task 2.3, 2.4 and 2.5 for complementary actions.
- Taking into account the overall objective of the EJP RD is to establish a sustainable structure for support of RD research and innovation, **the mapping shall expand beyond mere scientific requisites and encompass other connected domains** comprising: (i) Diagnosis & healthcare; (ii) Regulatory & ethics (including the open data & science issues); (iii) EU competitiveness & innovation.
- **New inputs will be collected from all stakeholders on the first trimester of 2020 and every year until the end of the EJP RD.**

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Messages to bring home

- The EJP RD proposal was prepared on the asumption of prioritisation.
- All the activities are planned, distributed and prioritized since the beginning till the end **(Good news for the Policy Board)**
- Prioritisation is needed
- Prioritisation is part of our common strategy (WP2)
- Our Guidelines and prioritisation model are useful tools made available for all, but may not be the only ones
- Participation of the Policy Board will be necessary mainly on specific unusual circunstances.
- **Prioritisation is an opportunity for all to increase the impact of EJP RD**

EJP RD

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Thank you for your attention and comments

EUROPEAN JOINT PROGRAMME RARE DISEASES

Special thanks for accepting being part of the Policy Board and for your inputs for prioritisation when needed

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EJP RD under Horizon Europe

EUROPEAN JOINT PROGRAMME
RARE DISEASES

EJP RD under Horizon Europe

How can we prepare for Horizon Europe today?

- Establishment of key partnerships with projects/initiatives that are complementary to the EJP RD and where we can demonstrate the added value of the EJP RD
  - 1+ Million Genomes Initiative (21 signatory countries; 1+MG sequences & shared by 2022 for the benefit of health and research; possible joint NMGs)
  - Global Alliance for Genomics & Health (GA4GH) (EJP RD as driver project with strong contribution to the development of standards for responsible genomic data sharing)
  - Connect4Children (C4C) (pan-European pediatric clinical trials network)
- Stronger innovation
  - to establish long-term collaboration with industrial partners with help of EFPIA, EUCOPE, EuropaBio, IMI
  - European Institute for Innovation & Technology - Health (health innovation, training & research)
- Establishment of connections with EU Council & Parliament to strengthen the cause of RDs

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Next Steps

EUROPEAN JOINT PROGRAMME
RARE DISEASES

Next Steps

- Finalise the constitution of Policy Board
- Finalise the creation of National Mirror Groups
- All PB members will receive the final version of the Annual Work Plan that should be approved by the EC in October
- All PB members will be regularly informed about the EJP RD achievements
- The coordination will liaise with the respective PB members and NMG to provide dedicated content or participate in meetings if requested
- Next regular Policy Board meeting: **8 July 2020**
- Possible additional PB meeting – Dec 2019/Jan 2020 to celebrate 1 year of the EJP RD

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Next Steps – what do we expect from you?

Help us and use us!

- To create strong connection with your (national) RD community → by identifying and connecting to relevant stakeholders
- To support the RD stakeholders → by acting for creation of the National Mirror Group and vehicle the voice of your RD community
- To help us engage with other type of stakeholders to enhance the innovation and sustainability of the EJP RD by engaging with members of EFPIA, EUCOPE, EuropaBio, EIT Health
- Use us to help you connecting with other partners in your country, bother us with questions on how you can use the EJP RD, etc.
- Use us to help you put in place dedicated webinars, meetings, events
- Disseminate, communicate → **Be the EJP RD Ambassador**

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THANK YOU

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