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Third Analysis of national state of play and alignment process with EJP RD

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Summary

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List of abbreviation

ERN(s)	European Reference Network(s)
EJP RD	European Joint Programme on Rare Diseases
EU	European Union
FAIR	Findable, Accessible, Interoperable, Reusable
IPR	Intellectual Property Rights
M	Month
MS	Member States
NMG(s)	National Mirror Group(s)
NP	National Plan(s)
NS	National Strategy(ies)
PB	Policy Board

R&I	Research and Innovation
RD	Rare Disease(s)
WP	Work Package

Executive summary

In Work Package 2 (WP2) "Integrative research and innovation strategy" of the European Joint Programme on Rare Diseases (EJP RD), special attention is specifically given to the development of the EJP RD research and innovation strategy in connection to all related stakeholders, counting among its particular objectives, to focus on the translation/impact of the prioritization on national and European Union (EU) strategies.

A key instrument required to fulfil the objectives of WP2 is a periodical analysis, carried out through a dedicated survey, involving the EU Member States (MS), and other countries involved in the EJP RD, and investigating their national alignments with the EJP RD relevant/complementary actions. A specific focus of this analysis is set on EU-13 Countries[1] and relates to their specific needs, obstacles and advancements. At present, data have been collected through specific surveys performed in autumn 2020 and spring 2021, targeting National Mirror Groups (NMGs) if constituted, or in their absence, persons involved in the National Plans or Strategies (NP/NS) for RD. The surveys address the topics covered by the 4 major non-transversal Pillars of the EJP RD and aim at assessing whether the NP/NS for RD or other national relevant RD undertakings align with the actions promoted by the EJP RD. The attention given to NP/NS for RD is motivated by their key role as instruments to reach common RD strategies at European/international level. As not all countries are provided with NP/NS for RD and as not all the national initiatives fall under the umbrella of the NP/NS for RD, also other relevant national RD initiatives have been investigated through the survey.

The outcomes of the present study furnish a description of the status quo of the 27 countries that participated to the surveys.

The results contribute to the evaluation of the achievements reached in the RD field and of the gaps that need specific interventions towards advisable advancements. Some gaps were already known on national level, and in fact the EJP RD is in place. Through this effort, such gaps and some other have been assessed simultaneously in a transversal way. Based on this analysis of RD activities in the different countries the EJP RD may define (additional or revised) activities to take up to fill gaps on European level in its last years.

Highlights and next actions

- The present report gives a picture, based on the responses of 27 countries (20 EU and 7 Associated/Other countries) that participated to the surveys in 2020 and/or in 2021. These results contribute to the delineation of the alignment status of the national policies for RD with the actions promoted by the EJP RD and provide the baseline for further assessments.
- Through the identification of the achievements and critical issues, specific areas of intervention at national and international level are signalled, in order to advance towards the desirable alignment between the actions promoted by the efforts of the EJP RD and the national/international RD undertakings.

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[1] Bulgaria, Croatia, Cyprus, Czech Republic, Estonia, Hungary, Latvia, Lithuania, Malta, Poland, Romania, Slovakia, Slovenia

- From the analysis performed up to now, the national RD policies for which there seem to be globally a greater alignment with the EJP RD actions are those related primarily to the field of "Capacity building and Empowerment" and then of "Resources and Services to foster research on rare diseases" (that fall respectively under Pillar 3 and Pillar 2 of the EJP RD).

- The national RD actions that evidence a general lower alignment with the actions promoted by the EJP RD are those within the fields of "National and International Investments on research on rare diseases", and "Accelerated translation of research projects and improvement of outcomes of clinical studies" (respectively falling in Pillar 1 and Pillar 4 of the EJP RD). In particular, the actions related to Pillar 4 "Accelerated translation of research projects and improvement of outcomes of clinical studies" appear as the most critical area with a low rate of implementation in the participating countries. This allows that some follow-up actions can be proposed at national level for better alignment.

- The results obtained for the EU-13 Countries regarding the activities of the 4 EJP RD Pillars demonstrate the need for specific interventions (different types, depending on the aforesaid activity and local conditionings) at national level, and the main achievements and criticalities appear to fall in the same areas as for the other EU MS countries.

- The questions dedicated specifically to EU-13 Countries evidence that their main perceived obstacles are funding, investment and limited links to potential RD research Partners. Although each country's conditionings are better known locally, probably sharing each national experience in those EU-13 countries and designing a common approach would be beneficial and would help overcome those obstacles,

- A WP2 Strategic Meeting held in July 2021 has facilitated the dialogue between relevant policy stakeholders on the status of the national alignments with the EJP RD actions. The meeting has focused on: (1) the presentation of the current state of advancements and achievements reached through the work of the EJP RD; (2) the focus on some country's experiences in terms of adopted best practices and main challenges to be faced; (3) the way to move forward, towards the enhancement and coordination of national and international efforts in the RD field. The meeting allowed to draw conclusions on some of the main emerged challenges, and funding has been identified as the main obstacle for the full implementation of RD research potential in many countries. The constitution of NMGs as tools to ensure national coordination of activities, strategies and for addressing the national needs has moreover arisen as a strongly advisable objective, according to several successful experiences.

- For the Fourth Analysis of national state of play and alignment process with EJP RD, a dedicated survey will be distributed in order to update the outcomes that are the objective of the present document, to obtain information from the countries that are not yet represented in this Third Analysis and to compare the results. This will help to assess the state of implementation of the proposals enacted by the EJP RD activities.

Introduction and Objective

The analysis of the national state of play and alignment process with EJP RD constitutes one of the activities to be performed periodically within WP 2. WP 2 concentrates on the progress of the EJP RD research and innovation strategy in connection to all related stakeholders. Within this framework, Task 2.5 ("Translation/impact of prioritization on National and EU strategies") leaders are expected to collect information from EU MS on the EJP RD relevant/complementary actions performed at national level, via a specific survey targeting, ideally NMGs and/or key persons deeply involved in the NP/NS for RD of their country. In fact, Task 2.5 concentrates both on exploring the impact of the European dimension of the EJP RD on the undertakings at national and EU level, and on the involvement of the Policy Board on the activities of the EJP RD itself.

The reports of Task 2.5 analysing the national alignments with the EJP RD and based on the results of the above mentioned survey are delivered at months (M) 9, 21, 32 and 55 of the EJP RD. The present deliverable is the third analysis of this series (due date of the deliverable M 32, year 3 of the EJP RD) and describes the results of the surveys launched in year 2020 and 2021. Specific focus of these analysis is made on EU-13 Countries, in respect to their specific needs, obstacles and advancements. In addition, the presentations held in the context of the Strategic Meeting of July 2021allowed to complement and illustrate the information received in the survey through the example of some country experience.

The results of survey investigating the national alignments with the actions promoted by the EJP RD contribute furthermore to the outcomes of Task 2.2 "Mapping the research and innovation strategy" and in the formulation of the Scoping Papers that are meant to be promoted by the NMGs and within the current and forthcoming EC Framework Programme. Task 2.2 is directed at identifying and regularly update the Research and Innovation (R&I) needs that feed the EJP RD Annual Plans and contribute to the development of the long-term strategy of the EJP RD. The mapping exercise includes the inputs from different types of stakeholders (also beyond the EJP RD beneficiaries and their Linked Third Parties) and is yearly reformulated in the form of a public Scoping Paper to be transmitted to the leaders of tasks 2.3[2], 2,4 and 2.5 for complementary actions.

In this way the mapping of R&I needs and the analysis of the national state of play and alignment process with the EJP RD represent useful and highly connected tools for the benefit of the integrative research and innovation strategy of the EJP RD, and for the RD community at a broader level.

The main objective of the present deliverable is to progress in the analysis of the state of play of relevant national RD activities and of their alignment with EJP RD. The report has been preceded by the "First-" and "Second Analysis of national state of play and alignment process with EJP RD".. The results of these analysis encompass: (i) the initia-

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[2] Task 2.3 "Scientific programming of joint transnational calls". Task 2.4 "Management of the medium, loner-term research strategy questions and dedicated linkage with Task Forces of IRDiRC"

tives and efforts regarding RD policies in EU MS and in Associated/Other countries participating in the EJP RD, and (ii) the alignment status of those policies with the main actions promoted by EJP RD. This two-fold focusing should offer an insight on the advisable mutual influence that should occur between the efforts and activities of the EJP RD and the existing RD policies.
D2.23 falls under the objectives of Pillar 0 the "Transversal and Communication" Pillar of the EJP RD.

The results discussed in this deliverable will contribute to: (i) deepen the understanding of the alignment status of the national policies for RD with the EJP RD actions, (ii) identify the achievements and critical issues faced by the countries in the RD field, (iii) suggest specific actions to be implemented at national and international level for a better alignment with the intentions set by the EJP RD, (iv) focus on the specific challenges encountered by the EU-13 Countries for more tailored actions to be promoted, (v) feed the discussion at the occasion of the Strategic Workshops with relevant stakeholders, and (vi) contribute to the mapping of the R&I needs of Task 2.2. of the EJP RD.

In July 2021 a Strategy Meeting with relevant stakeholders took place online and highlights on the results shown in the present deliverable D2.23 have been presented.

Background

The background and premises standing behind the formulation of the present document and encompassing the "soft-law documents" and initiatives that in the past years were dedicated to stimulate the countries to adopt NP/NS for RD as key instrument for the progressing in the diagnosis, treatment and care for people with RD, have been already described in the "First-"and "Second Analysis of national state of play and alignment process with EJP RD", and will not be further presented.

Methodology

The first survey titled "National Plans and Strategies for Rare Diseases" launched between September and November 2020[3], and addressed at the 35 EJP RD countries has been slightly adapted and enhanced in 2021. The 2021 survey has been structured so as to: (i) collect information from the countries that did not participate in the 2020 edition (ii) ask the countries that participated in the 2020 edition if they wanted to give some updates[4]. The survey (for updates and new participants) has been launched between May and June 2021.

The surveys collected in 2020 and 2021 are similar in terms of contents but the 2021 edition has been enhanced, as expected form the results of D2.22, regarding some aspects like the formulation of separated questions on the NP/NS for RD and on other relevant national initiatives. A relevant aspect of improvement of the 2021 survey has

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[3] The results of the first Survey on National Plans and Strategies for Rare have been sumbitted in D2.22.

[4] Countries that gave their agreement to be recontacted for further collaborative initiatives in the 2020 edition of the survey

been also that the countries not provided with/developing a NP/NS for RD had the opportunity to answer to the questions on the NP/NS for RD (referring to their developing NP/NS for RD) and to the questions regarding other relevant national initiatives.

For the content of the survey see Annex 1[5].

The survey, composed of multiple-choice and open questions, is divided into different sections:

- GDPR section
- Section on general information on the person/officer completing the survey (for further follow-up if necessary and for updates in the forthcoming editions of the survey)
- Section on information linked to the areas of interest of the 4 non-transversal Pillars (Pillar 1-4) of the EJP RD, specifically dedicated to analysing the alignment of the NP/NS for RD with the actions promoted by EJP RD. The titles and areas of interest of the 4 Pillars are as follows: “Pillar 1: National and International Investments on research in the field of rare diseases”, “Pillar 2: Resources and Services to foster research on rare diseases”, “Pillar 3: Capacity building and Empowerment”, and “Pillar 4: Accelerated translation of research projects and improvement of outcomes of clinical studies”
- Section on information linked to the areas of interest of the 4 non-transversal Pillars (Pillar 1-4) of the EJP RD, specifically dedicated to analysing the alignment of other national initiatives than the NP/NS in the RD field with the actions promoted by EJP RD. The titles of the areas of interest of the 4 Pillars are as above
- Section dedicated to the EU-13 Countries, and investigating their main perceived obstacles and barriers for the development, improvement and translation of RD research results, as well as for their participation in EU/International projects
- A final open question inviting the respondents to give free comments on aspects regarding the RD field not considered in the survey.

Privacy issues and delivery platform

The survey “National Plans and Strategies for Rare Diseases” is GDPR compliant (EU Regulation 2016/679 of the European Parliament and of the Council of 27 April 2016 on the protection of natural persons with regard to the processing of personal data and of the free movement of such data) and has been distributed (and the data gathered) through the Microsoft Forms Platform of the EJP RD.

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[5] The survey has been elaborated with branching possibilities, so that the respondents that already participated in autumn 2020 could choose on which topic they wanted to give eventual updates and skip the topics that required no updates. A pdf with the answers given by each respondent of the 2020 edition of the survey has been sent by email to the participants that in 2020 gave their agreement to be recontacted.

Target and timing of the survey

The main planned target of the survey are the NMGs as key actors for identifying, discussing and bringing the national needs to the upper level. To date, the NMGs have not been constituted in most countries (in part due to the difficult interactions during the COVID-19 pandemic). For this reason, a refined list of contacts has been created by the Coordination Team (based also on previous similar experiences in the RD field) and the 2.5 Task Leaders, for the distribution of the survey among persons directly/indirectly involved in the NP/NS for RD in the EJP RD countries (and broadly among contacts in the EU MS[6]). It is important to note that the survey itself invites the respondents that might not be directly involved in the NP/NS for RD to indicate contact persons directly involved/ more involved in the development or implementation of the NP/NS, as a deep knowledge of the situation in the country is needed to respond.

The survey has been distributed via email at the beginning of May 2021 to EUROPLAN contacts, Orphanet-INSERM contacts and EJP RD Partners. The deadline for completing the survey was 20 June 2021. Between the launching and the deadline period the contacted persons have been periodically asked if assistance in completing the survey was needed.

Results

A detailed summary of the results of the survey is provided below, question by question.

It is important to underline that, given the short laps of time passed between the 2020 and 2021 editions of the survey (6 months), the outcomes of the present document summarize the results obtained in 2020 and 2021. The answers of the countries who furnished updates in 2021 have been considered in these results, whereas for the countries that did not give updates, the answers given in 2020 were considered still valid. To these results those of the new participants were added.

General information

A total of 27 countries (Figure 1) contributed to the results of the survey. Thus, 77% of the 35 countries participating in the EJP RD are represented by the outcomes of this deliverable.

Of these:

- 20 are MS (of which 10 are EU-13 Countries)
- 7 are Associated/Other countries.

The countries who answered the survey are:

Armenia, Belgium, Bulgaria, Canada, Croatia, Czech Republic, Estonia, France, Georgia, Germany, Hungary, Ireland, Israel, Italy, Latvia, Lithuania, Luxembourg,

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[6] Cyprus is not participating in EJP RD but has been contacted as EU country

Poland, Portugal, Romania, Serbia, Slovakia, Spain, Sweden, The Netherlands, Turkey, UK[7].

Missing responses from EU MS and Other/Associated countries:
Austria, Cyprus, Denmark, Finland, Greece, Malta, Norway, Slovenia, Switzerland.

The updates (total number of updating countries=15) have been given by countries that are developing their first NP/NS for RD (Canada, Israel, Turkey, Georgia, Armenia), or are in the process of updating it (e.g., Czech Republic, Italy, Romania) or that wanted to provide more detailed information compared to that submitted in 2020. For the first, the answers refer to the forthcoming NP/NS for RD.

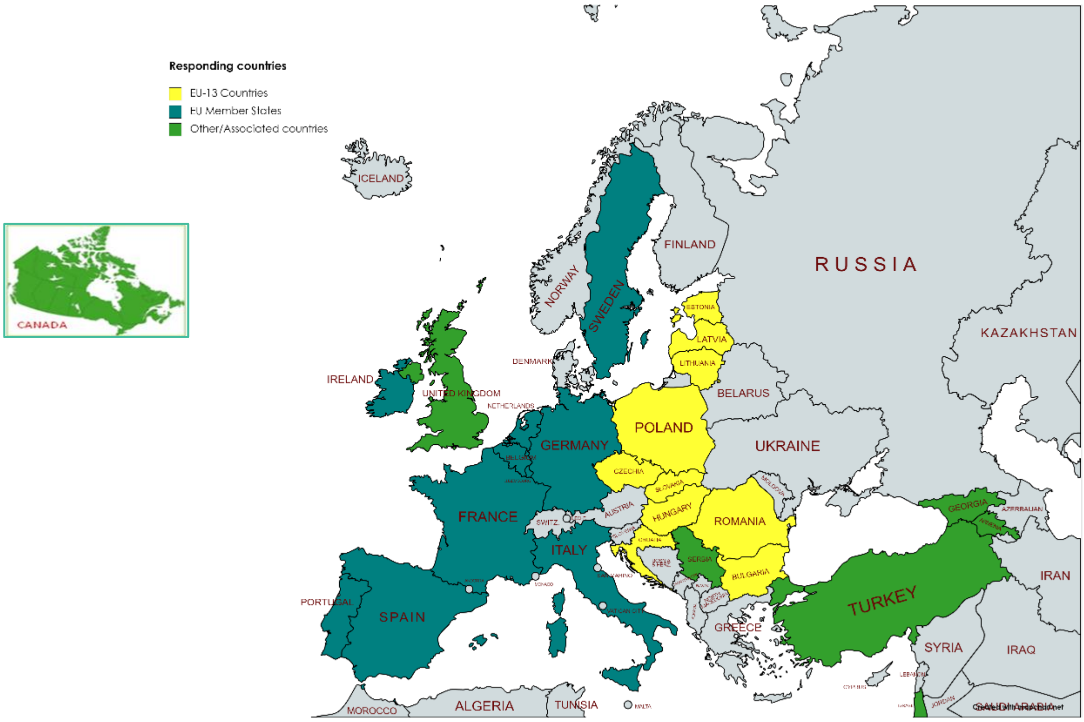

Figure 1. The 27 countries who answered to the survey (Blue: EU Member States, Yellow: EU-13 Countries, Green: Other/Associated Countries)

78% of the surveys have been completed by persons directly involved in the NP/NS for RD of their country, or in the development and construction of a NP/NS for RD. The remaining 22% of the surveys have been completed by persons working in Universities,

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[7] In the results of the present deliverable Hungary and Sweden figure among "no data", because the only available data on these countries is the absence of an active NP/NS for RD (Hungary) and the complete absence for a NP/NS for RD (Sweden). Hungary has been contacted in 2021 to complete the new edition of the survey but did not answer and Sweden did not agree in 2020 to be recontacted for further collaborative initiatives. Both countries are therefore not considered in the percentages.
Similarly, countries that did not give information regarding some topics in the 2021 edition of the survey have not been considered for the calculation of the relative percentages

National Research Funds, Research Councils, National Institutes of Health, or National Academies of Science.

Is there an approved NP/NS for RD in your country?

Yes (70%): Belgium, Bulgaria, Croatia, Czech Republic, Estonia, France, Germany, Ireland, Italy, Latvia, Lithuania, Luxembourg, Portugal, Romania, Serbia, Slovakia, Spain, The Netherlands, UK
No (4%): Sweden
No but it is under development in my country (26%): Armenia, Canada, Georgia, Hungary, Israel, Poland, Turkey
I don't know: -

Figure 2 illustrates the status quo of the countries that declare to have adopted a NP/NS for RD at some stage, the countries that declare that the NP/NS for RD is under development and the countries that affirm not to have adopted a NP/NS for RD yet.

18 Countries transmitted a link to the NP/NS for RD of their country (see Annex 2).

As of the information received to date, in 12 of the countries declaring to have a NP/NS for RD, the NP/NS is expired (Bulgaria, Croatia, Czech Republic, Estonia, Hungary, Ireland, Italy, Latvia, Lithuania, Portugal, Romania, The Netherlands) and currently there is no active NP/NS for RD. Some countries adopted "open-ended" NP/NS for RD (e.g., Belgium, Germany).

Moreover, 58% of the countries affirms to perform a periodical evaluation of the NP/NS for RD.

Complete information on the NP/NS of the responding countries is compiled in Table 1.

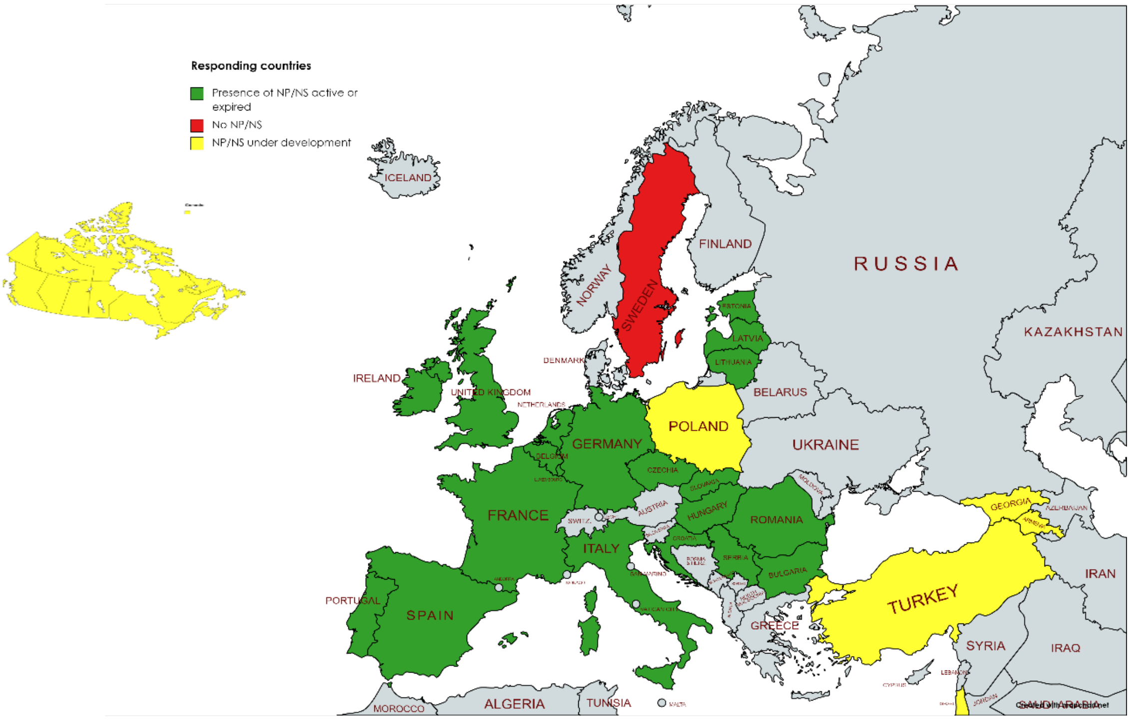

Figure 2: Status quo regarding NP/NS for RD (Red: countries who declare the complete absence of a NP/NS for RD; Yellow: countries that are developing a NP/NS for RD; Green: countries with a NP/NS for RD, active or expired)

Table 1. Details of the NP/NS for RD in the surveyed countries

Country	Year of approval	Year of expiry	Periodical evaluation	Under development
ARMENIA				✓
BELGIUM	2014			
BULGARIA	2008	2013		
CROATIA	2015	2020		
CANADA				✓
CZECH REPUBLIC	2018 (last edition)	2020	✓	
ESTONIA	2014	2017		
FRANCE	2018 (last edition)		✓	

Country	Year of approval	Year of expiry	Periodical evaluation	Under development
GEORGIA				✓
GERMANY	2013		✓	
HUNGARY	2013	2020		
IRELAND	2014	2018	✓	
ISRAEL				✓
ITALY	2013	2016		
LATVIA	2013	2020		
LITHUANIA	2013	2020	✓	
LUXEMBOURG	2018		✓	
POLAND				✓
PORTUGAL	2015 (last edition)	2020	✓	
ROMANIA	2014	2020	✓	
SERBIA	2019			
SLOVAKIA	2012			
SPAIN	2009 (last edition)		✓	
SWEDEN	No NP/NS for RD	No NP/NS for RD	No NP/NS for RD	No NP/NS for RD
THE NETHERLANDS	2013	2018	✓	
TURKEY				✓
UK	2013		✓	

Alignment process with EJP RD

The following outcomes refer to the countries who declare having a NP/NS for RD, active or expired, and to the countries that are developing a NP/NS, and concern the alignment process of the NP/NS for RD, and of other relevant national initiatives in the RD field, with the relevant/complementary actions promoted by the EJP RD.

Pillar 1: “National and International Investments in the field of RD”

Does the NP/NS for RD in your country promote national calls for research projects on rare diseases?

Yes (59%): Armenia, Croatia, France, Georgia, Germany, Ireland, Latvia, Luxembourg, Portugal, Romania, Spain, The Netherlands, Turkey, UK

No (33%): Belgium, Bulgaria, Czech Republic, Estonia, Italy, Lithuania, Poland, Serbia

I don’t know (8%): Canada, Slovakia

No data: Hungary, Israel, Sweden

Does the NP/NS for RD in your country promote transnational calls for research projects on RD?

Yes (50%): Armenia, Croatia, Czech Republic, France, Germany, Ireland, Luxembourg, Portugal, Romania, Spain, The Netherlands, UK

No (38%): Belgium, Bulgaria, Estonia, Georgia, Italy, Lithuania, Poland, Serbia, Turkey

I don’t know (12%): Canada, Latvia, Slovakia

No data: Hungary, Israel, Sweden

In comparison to the promotion of trans*national* calls (50%), a higher percentage of NP/NS for RD promotes *national* calls for research projects on RD (59%).

The NP/NS for RD of eleven countries (46%) promotes both national and transnational calls for research projects on RD (Armenia, Croatia, France, Germany, Ireland, Luxembourg, Portugal, Romania, Spain, The Netherlands, UK), while for two countries (8%) the NP/NS for RD promotes only transnational calls (Czech Republic, Portugal). Two countries (8%) declare that their NP/NS promotes only national calls for research projects (Georgia, Turkey).

For seven Countries (30%) the NP/NS for RD does not support neither national nor transnational calls for research projects on RD (Belgium, Bulgaria, Estonia, Italy, Lithuania, Poland, Serbia). Two countries (8%) affirm not to know whether the NP/NS for RD supports national and/or transnational calls for research projects (Canada, Slovakia).

Considering the free text comments in respect to the promotion of national and transnational calls for research projects endorsed by the NP/NS for RD, most of the promoted national and transnational calls for research projects fall in the field of translational research, first, and then of basic research. Furthermore, some NP/NS for RD also recommend RD research networking to facilitate international collaboration with relevant registries, organisations and consortia, including IRDiRC.

Does the NP/NS for RD in your country foresee investments for networking to share knowledge on RD?

Yes (46%): Armenia, Bulgaria, Croatia, Georgia, Ireland, Lithuania, Luxembourg, Spain, The Netherlands, Turkey, UK

I don't know (29%): Belgium, Canada, Czech Republic, France, Latvia, Romania, Slovakia

No (25%): Estonia, Germany, Italy, Poland, Portugal, Serbia

No data: Hungary, Israel, Sweden

In the free text comments, the most frequently cited areas are the development/participation in European Reference Networks (ERNs) and in Centres of Expertise, and the support to national/international events for RD. A dedicated axis of the NP/NS for RD for networking activities is also reported by some country. Apart from the NP/NS for RD, the establishment of national genetics and genomics medicine networks has been referred as a governments' priority and National Registries have also been cited as investments to share knowledge on RD. Universities, professional societies, public health institutions, Ministries of Health, National Science Funds, University medical centres figure among the involved parties in this area. Participation in Orphanet and the establishment and maintenance of RD dedicated websites have been mentioned, too.

Are there other public funding initiatives (than the NP/NS) in your country for research and/or networking in the field of RD?

Yes (68%): Belgium, Canada, Czech Republic, Estonia, France, Germany, Ireland, Israel, Italy, Luxembourg, Poland, Portugal, Romania, Slovakia, Spain, Turkey, UK

No (20%): Bulgaria, Georgia, Lithuania, Serbia, The Netherlands

I don't know (12%): Armenia, Croatia, Latvia

No data: Hungary, Sweden

When asked to describe the public funding initiatives for research and/or networking in the field of RD, the participation in national/international funded projects (specific and non-specific for RD) have been reported. These funded projects are promoted by national research agencies, Ministries of Health, national authorities, Universities or Academia, covering mostly the field of translational and basic research.

Public or private funding initiatives for national and transnational calls cover the topics of gene identification, understanding of biological mechanisms, socio-economic aspects of RD, patient engagement, network activities and therapy development. Joint funding schemes of some countries are not RD specific but include the RD field. In some cases, also the pharmaceutical industry, charities and RD research foundations support research on RD.

Are there private funding initiatives in your country for research and/or networking in the field of RD?

Yes (48%): Belgium, Canada, France, Germany, Ireland, Israel, Italy, Lithuania, Portugal, Spain, The Netherlands, UK
I don't know (28%): Armenia, Bulgaria, Croatia, Latvia, Romania, Slovakia, Turkey
No (24%): Czech Republic, Estonia, Georgia, Luxembourg, Poland, Serbia **No data:** Hungary, Sweden

Eleven countries (44%) claim the existence of both public and private funding initiatives, other than those promoted by the NP/NS for RD, for research and/or networking in the field of RD (Belgium, Canada, France, Germany, Ireland, Israel, Italy, Lithuania, Portugal, Spain, UK), seven countries[8] (28%) of public funding initiatives only (Czech Republic, Estonia, Luxembourg, Poland, Romania, Slovakia, Turkey). One country (4%) answers to have private funding initiatives only (Lithuania).

Two countries (8%) declare that there are no other public or private funding initiatives for research and/or networking in the field of RD (Georgia, Serbia). In one country the NP/NS for RD does not foresee investments for networking to share knowledge on RD either (Serbia).

The free text comments on the private funding initiatives for research/networking reveal that these are mostly supported by charities, patients' associations and pharmaceutical industries.

Pillar 2: "Resources and services to foster research on RD"

Is there an advisory body for national experts for EU Research and Innovation policy in your country?

Yes, an advisory body exists, but not specific for RD (52%): Bulgaria, Canada, Czech Republic, Estonia, France, Ireland, Israel, Italy, Luxembourg, Poland, Serbia, The Netherlands, Turkey

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[8] Romania, Slovakia and Turkey answer "Yes" for public funding initiatives but "I don't know" for private funding initiatives.

I don't know (32%): Armenia, Belgium, Croatia, Latvia, Lithuania, Slovakia, UK
Yes, an advisory body exists specific for RD (8%): Portugal, Romania
No (8%): Georgia, Germany **No data**: Hungary, Sweden

The presence of an advisory body of national experts for EU Research and Innovation (R&I) is reported in 56% (n=15) of the countries (Bulgaria, Canada, Czech Republic, Estonia, France, Ireland, Israel, Italy, Luxembourg, Portugal, Romania, Serbia, Spain, The Netherlands, Turkey) and in two of these countries (Romania and Portugal) the advisory body is specific for RD.

How does the NP/NS for RD of your country foresee the support of data repositories and tools in RD research? (Already existing examples: RD-Connect platform, European Rare Disease Registry Infrastructure (ERDRI), Cellosaurus, Infrafrontier, ORDO, HPO, DECIPHER, EGA)

Promoting both their implementation and development (52%): Belgium, Bulgaria, France, Germany, Ireland, Italy, Lithuania, Luxembourg, Serbia, Spain, Turkey UK
Promoting the implementation of such data repositories and tools (13%): Czech Republic, Georgia, Romania
The NP/NS for RD does not foresee the support of data repositories and tools in rare disease research (13%): Estonia, Poland, Portugal
Promoting the development of new data repositories and tools (13%): Armenia, Croatia, The Netherlands
I don't know (9%): Latvia, Slovakia **No data**: Canada, Hungary, Israel, Sweden

Globally, in 78% (n=18) of the countries the NP/NS for RD foresees the support of data repositories and tools dedicated to RD research (Armenia, Belgium, Bulgaria, Croatia, Czech Republic, France, Georgia, Germany, Ireland, Italy, Lithuania, Luxembourg, Romania, Serbia, Spain, The Netherlands, Turkey, UK), enacted through their implementation, their development, or both.

The topics covered by data repositories and tools in RD research, endorsed by the NP/NS for RD, and by other public and private initiatives have been investigated through specific questions, as described in the tables that follow (Table 2, Table 3, Table 4). In Table 2, the topics covered by the data repositories and tools for RD research that are supported by the NP/NS for RD of the participating countries are summarized. They have been ordered from the most to the less covered topic.

Table 2. Topics covered by the data repositories and tools for RD research that are supported by the NP/NS for RD, and countries covering them.

Topics of data repositories and tools for RD research	**Support by the NP/NS of:**
“Registries catalogues”	Armenia, Belgium, Bulgaria, Croatia, Czech Republic, France, Georgia, Germany, Ireland, Italy, Lithuania, Luxembourg, Romania, Serbia, Turkey
“Ontologies and codification”	Belgium, Croatia, Czech Republic, France, Georgia, Germany, Ireland, Italy, Lithuania, Luxembourg, Spain, UK
“Biobanks catalogues”	Armenia, Belgium, Czech Republic, France, Georgia, Ireland, Italy, Lithuania, The Netherlands, Turkey
“Support for clinical/translational research”	Armenia, Czech Republic, France, Georgia, Ireland, Italy, Portugal, The Netherlands, UK
“Data deposition & analysis”	Belgium, Bulgaria, France, Ireland, Italy, Lithuania, Portugal, Turkey, UK
“Tools”	Armenia, Bulgaria, France, Lithuania, Turkey, UK
“Access & privacy control”	Bulgaria, France, Georgia, Lithuania
“OMICS services”	France
“Cell lines”	Italy
“Animal models”	Italy
“Semantic standards”	France
“Other”	Belgium, Luxembourg

Are there other public funding initiatives in your country than NP/NS for RD for the development of data repositories and tools to support research on RD?

Yes (44%): Belgium, Bulgaria, Canada, Czech Republic, France, Ireland, Italy, Serbia, Spain, The Netherlands, Turkey
No (36%): Estonia, Georgia, Germany, Israel, Lithuania, Luxembourg, Poland, Portugal, UK
I don't know (20%): Armenia, Croatia, Latvia, Romania, Slovakia **No data**: Hungary, Sweden

The table below (Table 3) shows the covered topics in the eleven countries that report the presence of public initiatives other than the NP/NS for RD for the development of data repositories and tools to support research, presented from the most to the less covered topic.

Table 3. Topics covered by public funding initiatives other than the NP/NS for RD

Topics covered by other public funding initiatives than the NP/NS for RD for the development of data repositories and tools to support research in RD	Countries that cover the topic
"Registries catalogue"	Bulgaria, Canada, Czech Republic, France, Ireland, Italy, Serbia, Spain, The Netherlands
"Data deposition and analysis"	Bulgaria, Canada, France, Ireland, The Netherlands
"Biobanks catalogue"	Canada, Ireland, Serbia, The Netherlands
"Tools"	Bulgaria, Canada, The Netherlands, Turkey
"Support for clinical/translational research"	Canada, Ireland, The Netherlands
"Animal models"	Canada, France, The Netherlands
"Cell lines"	Canada, Serbia, The Netherlands
"Access & privacy control"	Bulgaria, Canada, The Netherlands
"Semantic standards"	The Netherlands

Are there other private funding initiatives in your country than the NP/NS for RD for the development of data repositories and tools to support research on RD?

I don't know (48%): Armenia, Belgium, Canada, Croatia, France, Germany, Israel, Latvia, Romania, Slovakia, Spain, Turkey
Yes (24%): Bulgaria, Ireland, Italy, Portugal, The Netherlands, UK
No (28%): Czech Republic, Estonia, Georgia, Lithuania, Luxembourg, Poland, Serbia **No data**: Hungary, Sweden

The table below (Table 4) shows the covered topics in the six countries who affirm the presence of private funding initiatives, apart from the NP/NS for RD, for the development of data repositories and tools to support research, listed from the most to the less covered topic.

Table 4. Topics covered by private funding initiatives, apart from the NP/NS for RD

Topics covered by other private funding initiatives than the NP/NS for RD for the development of data repositories and tools to support research in RD	Countries that cover the topic
"Registries catalogue"	Bulgaria, Ireland, Italy, Portugal, The Netherlands
"Biobanks catalogue"	Ireland, Italy, Portugal, The Netherlands
"Data deposition and analysis"	Bulgaria, Ireland, Italy, The Netherlands
"Support for clinical/translational research"	Bulgaria, Ireland, The Netherlands
"Tools"	Bulgaria, Italy, The Netherlands
"Cell lines"	Portugal The Netherlands
"Animal models"	Portugal, The Netherlands
"Access & privacy control"	Bulgaria, The Netherlands
"Semantic standards"	-
"Other"	UK-Patient Registries

Analysing the results of these last three items of the survey, it appears that, in addition to the support given to data repositories and tools in RD research by the NP/NS for RD, in four countries (16%) the support is given also by both other public and private funding initiatives (Bulgaria, Ireland, Italy and The Netherlands), in two countries (8%) also by private funding initiatives (Portugal and UK), and in six countries (24%) also by other public funding initiatives (Belgium, Canada, Czech Republic, France, Serbia, Turkey[9]).

Furthermore, in four countries (16%) the support is given by the NP/NS for RD only (Georgia, Germany, Lithuania, Luxembourg[10]). Three other countries (12%) whose NP/NS for RD supports the development/implementation of data repositories and tools for RD research answer "I don't know" regarding the existence of other public or private funding initiatives in this field (Armenia, Croatia, Romania).

In one country data repositories and tools for RD research are supported only by other private funding initiatives (Portugal).

In two countries (8%) there is lack of support to data repositories and tools for research in RD, that is not endorsed by the NP/NS for RD nor by other public or private funding initiatives (Estonia, Poland).

Finally, two countries (8%) answered not knowing if data repositories and tools are supported by the NP/NS for RD and/or by other public or private funding initiatives (Latvia, Slovakia).

In general, the support provided by the NP/NS for RD to data repositories and tools in RD research is higher than the support issued by other public and/or private funding initiatives.

Does the NP/NS for RD support FAIR data in your country? (*FAIR: Findable, Accessible, Interoperable, Reusable)*

No (50%): Belgium, Bulgaria, Czech Republic, Georgia, Lithuania, Luxembourg, Poland, Portugal, Romania, Serbia, The Netherlands, Turkey
I don't know (29%): Armenia, Croatia, Estonia, Israel, Latvia, Slovakia, Spain
Yes (21%): France, Germany, Ireland, Italy, UK
No data: Canada, Hungary, Sweden

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[9] Among these, Canada, France, Spain and Turkey answer "I don't know" to the question on other private funding initiatives for the development of data repositories and tools in RD research

[10] Germany answers that there are no other public funding initiatives, but "I don't know" regarding private initiatives

Do other initiatives than the NP/NS for RD support FAIR data in your country? (*FAIR: Findable, Accessible, Interoperable, Reusable)*

I don't know (44%): Armenia, Bulgaria, Croatia, Estonia, Israel, Latvia, Poland, Portugal, Slovakia, Spain, Turkey
Yes (36%): Belgium, Canada, Czech Republic, France, Germany, Ireland, Italy, Luxembourg, The Netherlands
No (20%): Georgia, Lithuania, Romania, Serbia, UK
No data: Hungary, Sweden

Considering the outcomes shown in the tables above, globally, in 56% (n=14[11]) of the countries FAIR data are supported by the NP/NS for RD and/or by other initiatives.

In one country (4%) the support to FAIR data is given only by the NP/NS for RD (UK), in five countries (20%) only by other initiatives (Belgium, Canada, Czech Republic, Luxembourg, The Netherlands) and in four countries (16%) by both (France, Germany, Ireland, Italy).

In four countries (16%) FAIR data are not supported at all (Georgia, Lithuania, Romania, Serbia). Seven countries (28%) answer not to know if the NP/NS for RD or other national initiatives support FAIR data (Armenia, Croatia, Estonia, Israel, Latvia, Slovakia, Spain). Finally, four countries (16%) whose NP/NS for RD does not support FAIR data do not know if other initiatives do so.

The free text comments reflect what has been already outlined with respect to the support of FAIR data, namely that some countries declare to adopt the FAIR principles through the endorsement of the NP/NS for RD or through other initiatives, while some other countries affirm to adopt the FAIR principles, even if not explicitly outlined in the NP/NS or by other national policies.

Is the adoption of multidisciplinary holistic approaches for RD diagnostics and therapeutics promoted by the NP/NS for RD of your country?

Yes (81%): Armenia, Belgium, Bulgaria, Croatia, France, Georgia, Germany, Ireland, Italy, Latvia, Lithuania, Luxembourg, Poland, Portugal, Romania, Spain, The Netherlands, Turkey, UK
No (9%): Czech Republic, Estonia
I don't know (4%): Slovakia **No data**: Canada, Israel, Hungary, Sweden

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[11] This percentage that comprises both the support of FAIR data endorsed by the NP/NS for RD and/or by other initiatives is calculated on 15 countries, even though for Canada there are no data regarding the support endorsed by the NP/NS for RD

Is the adoption of multidisciplinary holistic approaches for RD diagnostics and therapeutics promoted by other initiatives than the NP/NS for RD of your country?

Yes (44%): Belgium, Bulgaria, Canada, France, Germany, Ireland, Italy, Latvia, Lithuania, Portugal, Romania
No (28%): Czech Republic, Estonia, Georgia, Luxembourg, Poland, Serbia, UK
I don't know (28%): Armenia, Croatia, Israel, Spain, Slovakia, The Netherlands, Turkey **No data**: Sweden, Hungary

The adoption of multidisciplinary holistic approaches for diagnostics and therapeutics is promoted globally by the NP/NS for RD and/or by other initiatives in 83%[12] (n=20) of the countries, in ten countries (40%) both by the NP/NS for RD and by other initiatives (Belgium, Bulgaria, France, Germany, Ireland, Italy, Latvia, Lithuania, Portugal, Romania) and in five countries (20%) only by the NP/NS for RD (Georgia, Luxembourg, Poland, Serbia, UK). Five countries (20%) that support the adoption of multidisciplinary holistic approaches for diagnostics and therapeutics through the NP/NS do not know if other national initiatives do so (Armenia, Croatia, Spain, The Netherlands, Turkey) and for one country (4%) other initiatives give this kind of support, but there are no data regarding the support endorsed by the NP/NS for RD (Canada)

Furthermore, in two countries (8%) the adoption of multidisciplinary holistic approaches for diagnostics and therapeutics is not promoted neither by the NP/NS for RD nor by other initiatives (Czech Republic, Estonia), whereas one country (4%) answers not to know if the promotion is adopted by the NP/NS for RD or by other national initiatives (Slovakia). One further country (4%) answers not to know with respect to other national initiatives, while there are no data on this topic regarding the NP/NS for RD (Israel).

From the free comments it emerges that the adoption of multidisciplinary holistic approaches is promoted by the NP/NS or by other initiatives for RD, primarily through the development of national networks, national centres, national programmes, dedicated committees, Centres of Reference, Centres of Expertise and participation in European Reference Networks (ERNs). Some NP/NS for RD explicitly mention the need for inter-ministerial, intersectoral and interinstitutional cooperation for a complementary use of the medical, social, scientific and technological resources.

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[12] This percentage is calculated on the twenty countries that answered that their NP/NS for RD and/or other national initiatives promote the adoption of multidisciplinary holistic approaches, although it has to be considered that for two of them (Canada and Israel) there are no data regarding the promotion by the NP/NS for RD

Pillar 3 "Capacity building and empowerment"

Does the NP/NS for RD promote and/or support training activities?

Yes (78%): Armenia, Bulgaria, Croatia Czech Republic, France, Georgia, Germany, Ireland, Italy, Lithuania, Luxembourg, Poland, Portugal, Romania, Serbia, Spain, Turkey, UK
No (18%): Belgium, Estonia, Latvia, The Netherlands
I don't know (6%): Slovakia **No data**: Canada, Israel, Hungary, Sweden

The topics of the training activities supported/promoted by the NP/NS for RD are summarized, in descending frequency order, in the following table (Table 5).

Table 5. Topics covered by the training activities supported by the NP/NS for RD

Topics covered by the training activities supported by the NP/NS for RD	Countries that cover the topic
"Empowerment of the patients"	Bulgaria, Croatia, Czech Republic, France, Georgia, Ireland, Lithuania, Luxembourg, Poland, Portugal, Romania, Serbia, Spain
"Registries"	Bulgaria, Croatia, Czech Republic, France, Georgia, Ireland, Italy, Lithuania, Luxembourg, Poland, Romania
"Data management"	Armenia, Bulgaria, Czech Republic, France, Georgia, Ireland, Lithuania, Turkey
"Online education courses"	Czech Republic, Ireland, Georgia, Lithuania, Poland, Portugal, Romania
"Standard and quality of genetics/genomics data in clinical practice and laboratories"	Armenia, Bulgaria, Czech Republic, Lithuania, Poland, Romania, Turkey
"Biobanks"	Armenia, Ireland, Lithuania, Poland, Turkey
"Data quality"	Armenia, Bulgaria, Georgia, Lithuania
"Other"	Lithuania, UK, Germany
"FAIR data"	Italy, Turkey

Apart from the provisions made in the NP/NS for RD, are there any other training, mentoring and coaching activities in the field of RD provided in your country?

Yes (56%): Belgium, Bulgaria, Canada, Czech Republic, Germany, Ireland, Italy, Lithuania, Portugal, Serbia, Slovakia, Spain, The Netherlands, UK
I don't know (28%): Armenia, Croatia, Estonia, France, Latvia, Romania, Turkey
No (16%): Georgia, Israel, Luxembourg, Poland **No data:** Hungary, Sweden

Ten countries (40%) promote/support training activities both through the NP/NS for RD and through other national initiatives (Bulgaria, Czech Republic, Germany, Ireland, Italy, Lithuania, Portugal, Serbia, Spain, UK).

Three countries (12%) support training activities only through the NP/NS for RD (Georgia, Luxembourg, Poland) and two countries (8%) only through initiatives other than NP/NS for RD (Belgium and The Netherlands).

Six countries (24%) whose NP/NS for RD supports training activities, do not know if the support is endorsed also through other initiatives (Armenia, Croatia, France, Latvia, Romania, Turkey)

One country (4%) affirms that other national initiatives than the NP/NS for RD support training activities but gives no data regarding the NP/NS (Canada), and one country (4%) answers not to know with respect to the NP/NS for RD but that other national initiatives support training activities (Slovakia). Other two countries (8%), affirm that the NP/NS for RD does not support training activities, and not to know about other national initiatives (Estonia, Latvia).

The free text comments regarding the specification on the training activities supported/promoted by the NP/NS for RD, show that the addressed topics cover mainly: the empowerment of patients (most cited training activity), the training of practitioners (general or specialists in RD), trainings on data and registries. Some countries mention the introduction of mandatory trainings on RD for all medical students.

The other described training activities that are promoted by initiatives different from the NP/NS for RD are trainings provided mainly through the patients' associations, the National Centres for RD, and the Universities.

Pillar 4 “Accelerated translation of research projects and improvement of outcomes of clinical studies”

Does the NP/NS for RD of your country promote a rapid translation of the research results in clinical studies and healthcare?

Yes (48%): Croatia, Czech Republic, France, Georgia, Ireland, Italy, Romania, Spain, The Netherlands, Turkey, UK
No (39%): Armenia, Belgium, Bulgaria, Estonia, Germany, Lithuania, Poland, Portugal, Serbia
I don’t know (13%): Latvia, Luxembourg, Slovakia **No data**: Canada, Israel, Sweden, Hungary

Are there other initiatives that promote a rapid translation of research results in clinical studies and healthcare in the field of RD, other than the NP/NS for RD in your country?

No (36%): Czech Republic, Estonia, Georgia, Germany, Israel, Lithuania, Luxembourg, Poland, Serbia
I don’t know (36%): Armenia, Belgium, Bulgaria, Croatia, France, Latvia, Romania, Slovakia, Spain
Yes (28%): Canada, Ireland, Italy, Portugal, The Netherlands, Turkey, UK **No data**: Hungary, Sweden

Globally, twelve countries (53%) reveal to promote a rapid translation of research results in clinical studies and healthcare through the NP/NS for RD and/or through other initiatives (Canada, Croatia, Czech Republic, France, Georgia, Ireland, Italy, Portugal, Romania, Spain, The Netherlands, Turkey, UK).

Five countries (21%) assert that the rapid translation of research results in clinical studies and healthcare is promoted both by the NP/NS for RD and by other initiatives (Ireland, Italy, The Netherlands, Turkey, UK). Of the other six countries whose NP/NS for RD promotes the rapid translation of research results in clinical studies and healthcare, four countries (17%) affirm not knowing whether other initiatives do so (Croatia, France, Romania, Spain), and two countries (8%) that other initiatives do not endorse this promotion (Czech Republic, Georgia).

For one country (4%) presenting other initiatives for the rapid translation of research results in clinical studies and healthcare there are no data regarding the NP/NS for RD on this topic (Canada), and in another country (4%) other initiatives enforce the promotion, but not the NP/NS for RD (Portugal). One further country (4%) gives no information regarding the NP/NS for RD and answers not to know with respect to other initiatives (Israel).

In five countries (21%) the rapid translation of research results in clinical studies and healthcare is not promoted by the NP/NS for RD, nor by other initiatives (Estonia, Germany, Lithuania, Poland, Serbia). Three countries (13%) answer that there are only other initiatives than the NP/NS for RD for the rapid translation of research results in clinical studies and healthcare, and not to know about other initiatives (Belgium, Bulgaria, Luxemburg). Two countries (8%) do not know if the NP/NS for RD and or other initiatives promote the rapid translation of research results in clinical studies and healthcare (Latvia, Slovakia).

The free text comments indicate that the rapid translation of research results in clinical studies and healthcare is implemented through the creation of dedicated groups for innovation and RD Task Forces, or through the collaboration of different stakeholders for the production of Clinical Practice Guidelines for RD, based on the translation of RD research in clinical studies and healthcare; other described means for the rapid translation are the constitution of health research institutes with the participation of different research centres for the promotion of translational research with a better transfer of the scientific advances or with dedicated research programmes for RD. New clinical procedures, drugs and medical devices have also been reported as examples of the rapid translation of research results in clinical studies and healthcare, endorsed by the NP/NS for RD.

The specifications on the other initiatives for a rapid translation of research results in clinical studies and healthcare, different from the NP/NS for RD, make reference to ad hoc initiatives undertaken by medical societies, universities, pharmaceutical industries and medical treatment facilities. National clinical programmes (specific and non-specific for RD) for designing models of care, clinical pathways and guidelines, supporting and guiding implementation and developing innovative, efficient and evidence-based solutions that can be applied on a national basis in the health system are also mentioned. Specific national programmes for translational research on RD are named as well. Some countries mention a Rare Disease Taskforce.

Does the NP/NS for RD of your country promote the development of innovative methodologies tailored for clinical trials in RD?

No (61%): Armenia, Belgium, Bulgaria, Czech Republic, Estonia, Georgia, Germany, Luxembourg, Poland, Portugal, Romania, Serbia, The Netherlands, Turkey
I don't know (22%): Croatia, France, Latvia, Slovakia, Spain
Yes (17%): Ireland, Italy, Lithuania, UK **No data:** Canada, Israel, Sweden, Hungary

Regarding the support of innovative methodologies tailored for clinical trials, patient's registries as well as registries on ongoing clinical trials, and the large involvement of the patients and their families have been mentioned as open response to the question. Access to new and innovative medicines, innovative solutions and technologies in different areas of RD research and healthcare, improvement of the connection between research and care and the envisaging of the development of "clinical trials gateways" to provide information to the patients about research trials, have been referred too.

Focus on EU-13 Countries with regard to specific needs, obstacles and advancements

Ten of the EU-13 Countries answered to the survey (Bulgaria, Croatia, Czech Republic, Estonia, Hungary, Latvia, Lithuania, Poland, Romania, Slovakia[13]). The NP/NS for RD of Poland is under development, whereas in all the other responding EU-13 countries the NP/NS for RD appears to be expired and not yet replaced or updated.

As to the question[14] on the main perceived obstacles and barriers for the development, improvement and translation of RD research results, "Funding" is indicated by 89% of the participating EU-13 Countries; "Difficulties in accessing to national resources for funding of research and development of RD projects" is mentioned by 56% of the countries; "Lack of options for exploitation of research results at national level" by 40%; whereas "Language" and "Other" are pointed out by 11% of the responding countries.

The answers are summarized in the table below (Table 6), in descending order of perceived difficulty.

Table 6. Main perceived obstacles and barriers for the development, improvement and translation of RD research results

Main perceived obstacles and barriers for the development, improvement and translation of RD research results (ordered in decreasing order of perceived difficulty)	**Perceived as an obstacle/barrier by:**
"Funding"	Bulgaria, Croatia, Czech Republic, Estonia, Latvia, Lithuania, Poland, Romania
"Difficulties in accessing to national resources for funding of research and development of RD projects"	Bulgaria, Croatia, Latvia, Lithuania, Romania
"Lack of options for exploitation of research results at national level "	Estonia, Lithuania
"Language"	Slovakia
"Other"	Slovakia
No data	Hungary

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[13] In this deliverable all collaborative efforts have been valued and taken into account, although it has to be considered that for Slovakia the survey has been filled out by a person not involved in the NP/NS for RD, that answered "I don't know" to a great part of the items. Despite this, the answers have been included in the present analysis as they contain also relevant contents.

[14] Multiple answers were possible for the two questions dedicated to the EU-13 Countries

In the free text comments, the lack of funding is referred as the most frequent obstacle and barrier by all the respondents, often related to the lack of interest for RD at national level, with a consequent dearth of expertise and of dedicated national efforts.

Regarding the participation in EU/International projects in the RD field, 78% of the countries indicates "Limited links to potential partners" as the most important estimated obstacle and barrier. The other critical aspects are indicated in the following order of frequency: "Lack of information on funding opportunities" (56 %) "Bureaucratic application on reporting procedures" (50%), "Limited skills on drafting proposals" (44%), "Irrelevance of programme topics and goals to own research agenda" (2%), "Quality of support provided by national contact points" (22%).

The answers are summarized in the following table (Table 7).

Table 7. Most important estimated obstacles and barriers for the participation in EU/International projects in the RD field

Most important estimated obstacles and barriers for the participation in EU/International projects in the RD field	**Perceived as an obstacle/barrier by:**
"Limited links to potential partners"	Bulgaria, Croatia, Czech Republic, Estonia, Lithuania, Romania, Slovakia
"Lack of information on funding opportunities"	Estonia, Latvia, Lithuania, Poland, Romania
"Bureaucratic application on reporting procedures"	Bulgaria, Croatia, Czech Republic, Estonia, Romania
"Limited skills on drafting proposals"	Bulgaria, Czech Republic, Lithuania, Poland
"Quality of support provided by national contact points"	Czech Republic, Slovakia
"Irrelevance of programme topics and goals to own research agenda"	Lithuania, Poland
No data	Hungary

Furthermore, the free text comments on the obstacles and barriers to the participation in EU/International projects in the RD field describe the scarcity of experience and the need to partner more experienced institutions, that in turn is pointed out as the overall critical aspect in the multiple-choice questions (" Limited links to potential partners"). Poor support services to the interpretation of core elements of EU funding process, like

Ethics, Intellectual Property Rights (IPR), consortium agreements are as well stressed out.

Focus on EU-13 Countries in respect the alignment status with the 4 non-transversal Pillars of the EJP RD

Below a summary is presented with the results regarding the alignment status of the 9[15] EU-13 Countries who participated in the survey (Bulgaria, Croatia, Czech Republic, Estonia, Latvia, Lithuania, Poland, Romania, Slovakia) with the 4 EJP RD Pillars, highlighted in points:

Pillar 1: "National and International Investments in the field of RD"

- In three countries (33%) the NP/NS for RD promotes national calls for research projects on RD (Croatia, Latvia, Romania), in five countries (56%, Bulgaria, Czech Republic, Estonia, Lithuania, Poland) the NP/NS for RD does not promote national calls for research projects on RD, and one country (11%) answers "I don't know" (Slovakia).
- In three countries (33%) the NP/NS for RD promotes transnational calls for research projects (Croatia, Czech Republic, Romania). In four countries (45%) it does not promote transnational calls for research projects (Bulgaria, Estonia, Lithuania, Poland), and two countries (22%) answer "I don't know" (Latvia, Slovakia).
- In three countries (33%) the NP/NS for RD foresees investments to share knowledge on RD (Bulgaria, Croatia, Lithuania). In two countries (22%) it does not foresee investments to share knowledge on RD (Estonia, Poland), and four countries (45%) answer "I don't know" (Czech Republic, Latvia, Romania, Slovakia).

Other public/private initiatives for research and/or networking in the field of RD:

- Five countries (56%) have other public funding initiatives for research and/or networking in the field of RD (Czech Republic, Estonia, Poland, Romania, Slovakia), two countries (22%) do not (Bulgaria, Lithuania) and two countries (22%) answer "I don't know" (Croatia, Latvia).
- One country (11%) has private funding initiatives for research and/or networking in the field of RD (Lithuania), three countries (33%) do not have such private funding initiatives (Czech Republic, Estonia, Poland), and five (56%) countries answer "I don't know" (Bulgaria, Croatia, Latvia, Romania, Slovakia).

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[15] Hungary is not included in the present results, as it declared in the 2020 survey not to have a NP/NS for RD, (and has not responded neither to the items investigating the alignment with the 4 EJP RD Pillars, nor the section dedicated to EU 13 Countries) and did not participate to the 2021 edition.

<u>Pillar 2</u> "Resources and services to foster research on RD"

- In five countries (56%) there is an advisory body of national experts for EU Research and Innovation policy (Bulgaria, Czech Republic, Estonia, Poland, Romania), and in one of these countries (Romania), the advisory body is specific for RD; four countries (44%) answer "I don't know" (Croatia, Latvia, Lithuania, Slovakia).
- In five countries (56%) the NP/NS for RD foresees the support of data repositories and tools in RD research (Bulgaria, Croatia, Czech Republic, Lithuania, Romania) in two countries (22%) the support is not foreseen (Estonia, Poland), and two countries (22%) answer "I don't know" (Latvia, Slovakia).
- The topics supported by the NP/NS for RD of the five above cited countries (Bulgaria, Croatia, Czech Republic, Lithuania, Romania) with regard to data repositories and tools for RD research are named in this order: "Registries catalogues", "Ontologies and codification", "Biobanks catalogues", "Data deposition and analysis", "Tools", "Access & privacy control" and "Support to clinical/translational research".
- With regard to other public funding initiatives for the development of data repositories and tools, these are reported in two countries (22%, Bulgaria and Czech Republic), and not supported in three countries (33%, Estonia, Lithuania, Poland), while four (44%) countries answer "I don't know" (Croatia, Latvia, Romania, Slovakia).
- The topics covered by the countries with other public funding initiatives for the development of data repositories and tools (Bulgaria and Czech Republic) are "Registries catalogues", "Data deposition and analysis", "Tools" and "Access & privacy control".
- As for the private funding initiatives for the development of data repositories and tools, these are reported in one country (12%, Bulgaria), and not supported in four countries (44%, Czech Republic, Estonia, Lithuania, Poland), while four countries (44%) answer "I don't know" (Croatia, Latvia, Slovakia, Romania).
- The topics covered by the country with private funding initiatives for the development of data repositories and tools (Bulgaria) are "Registries catalogues", "Support to clinical/translational research", "Data deposition and analysis", "Tools" and "Access & privacy control".
- FAIR data are supported only by initiatives other than the NP/NS for RD in one country (11%, Czech Republic). The remaining countries (89%) declare that neither the NP/NS for RD, nor other initiatives support FAIR data, or not be informed on the topic (Bulgaria, Croatia, Czech Republic, Estonia, Latvia, Poland, Romania, Slovakia).
- The adoption of multidisciplinary holistic approaches for RD diagnostics and therapeutics is promoted by the NP/NS for RD in six countries (67%, Bulgaria, Croatia, Latvia, Lithuania, Poland, Romania), not promoted in two countries (22%, Czech Republic, Estonia), and one country (11%) answers "I don't know"

(Slovakia). When considering other national initiatives than the NP/NS for RD on this topic, these are mentioned in four countries (45% Bulgaria, Latvia, Lithuania, Romania), not present in three countries (33% Czech Republic, Estonia, Poland) while two countries (22%) answer "I don't know" (Croatia, Slovakia).

<u>Pillar 3</u> "Capacity building and empowerment"

- In six countries (67%, Bulgaria, Croatia, Czech Republic, Lithuania, Poland, Romania), the NP/NS for RD promotes and/or supports training activities; in two countries (22%, Estonia, Latvia) it does not endorse this support, and one country (11%) answers "I don't know" (Slovakia).

- The training activities supported/promoted by the NP/NS for RD of the six above listed countries cover all the topics investigated through the dedicated question of the survey except for trainings on FAIR data, even if not all the six countries cover the same topics.

- As to the endorsement of training activities by initiatives other than the NP/NS for RD, four countries (44%) declare the presence of other initiatives (Bulgaria, Czech Republic, Lithuania, Slovakia), four countries (44%) answer "I don't know" (Croatia, Estonia, Latvia, Romania) and one country (12%) answers that there are no such initiatives (Poland).

<u>Pillar 4</u> "Accelerated translation of research projects and improvement of outcomes of clinical studies"

- The NP/NS for RD promotes the rapid translation of research results into clinical studies and healthcare in three countries (33% Croatia, Czech Republic, Romania), while in 4 countries (44%, Bulgaria, Estonia, Lithuania, Poland), the NP/NS for RD does not back up this promotion, and two countries answer "I don't know" (Latvia, Slovakia).

- Initiatives (other than the NP/NS for RD) that promote the rapid translation of research results into clinical studies and healthcare are not reported by any of the country.

- The development of innovative methodologies tailored for clinical trials is reported in the NP/NS for RD of one country (11%, Lithuania), but is not present in the NP/NS of five countries (56%, Bulgaria, Czech Republic, Estonia, Poland, Romania), while the other three countries (33%) answer "I don't know" (Croatia, Latvia, Slovakia).

<u>Final open question (to all the countries, including EU-13 Countries)</u>

The final open question on other possible aspects that were not considered elsewhere in the survey has been addressed to all the survey respondents (including EU-13 Countries) declaring the presence of a NP/NS for RD in their country and has been filled out by 38% (n=6) of them (Bulgaria, Estonia, Ireland, Luxembourg, Romania, The Netherlands). The comments that could be of interest for the conclusions to be drawn from these results are: (i) a certain slowdown due the COVID-19 pandemic during 2020; (ii) the challenges set by the absence of a critical mass of RD patients in small countries; (iii) the role of patients as partners for the advancement in research and care.

Discussion

The results of this deliverable represent the analysis of the national state of play and alignment process with EJP RD of relevant national RD actions as portrayed through the outcomes obtained for the period November 2020-June 2021. The short time span (six months) elapsed between the spread of the first and the second survey allows to consider the outcomes as a single result and permits to widen the number of countries participating to the analysis, giving this way a more precise picture. In order to have all participating countries on the same baseline, the 2020 responding countries had the possibility to give eventual updates or to add information in 2021.

This third analysis can therefore be considered as an image of the current situation and will serve as reference for further assessments and actions. Even if not comprising 8 of the EJP RD participating countries (23%), the data obtained through this enquiry can nevertheless give important insights of the general state of relevant national RD policies and of their alignment with the EJP RD actions. The series of deliverables analysing the national state of play can be considered as a relevant tool for detecting key aspects that should be addressed by the future actions of the EJP RD. Key points for each Pillar and specific key points for EU13 countries are listed and detailed below. Furthermore, an assessment of the impacts of the EJP RD activities on the national policies for RD should be an objective of the fourth and last analysis of this series.

The gathered information has at least three important aspects. First it can serve as a baseline for further assessments for the responding countries; furthermore, the achievements shown by some countries can constitute a positive reference for other countries; thirdly, the non-responding countries might constitute a specific target with particular and even common reasons (to be analysed) for the lack of participation.

General information

Globally, a general good presence of NP/NS for RD in the responding countries can be observed, with 78% of the countries having adopted a NP/NS for RD at some stage, and 26% being in the process of developing a NP/NS for RD. It has nonetheless to be observed that many of the time-bound NP/NS for RD expired at that to date there has been no replacement (93% of the countries that appear having a time-bound NP/NS for RD). The survey that will be launched for the Fourth and last Analysis of this series should address the issue of the possible reasons for the non-renewal of the NP/NS for RD in order to detect possible shared and/or country specific obstacles, and plan tailored actions.

Furthermore, obtaining data also from the missing countries and encouraging the development or update of the NP/NS for RD of the countries that currently do not have an approved/active NP/NS for RD are important objectives to be envisaged.

With respect to the alignment status with the four non -transversal Pillars of the EJP RD, the following considerations can be stressed out:

Alignment with Pillar 1 “National and International investments on research in the field of RD”:

Key point: support to investments to share knowledge on RD research activities needs to be fostered to achieve a better alignment with the activities of Pillar 1. Transnational and national calls are also in need of attention, especially in EU-13 Countries.

The survey results show that the aspect of RD research needing to be strengthened most is the support towards investments to share knowledge on RD, promoted only by 46% of the NP/NS for RD. It should be noted, however, that the EJP RD is already provided with a Networking Support Scheme (WP 7), that addresses the support of RD knowledge-sharing between the countries. This resource is available to all EJP RD participating countries and should be better disseminated through the EJP RD Partners to encounter the needs of stronger knowledge-sharing on RD.

There is also a need to direct the efforts for an enhancement of national and transnational calls for research projects, as these are promoted respectively in 59% and 50% of the countries. Although this can appear as a relatively positive outcome, research on RD is of pivotal importance and should be furthermore enhanced.

Besides of the relatively positive result on the support of the NP/NS for RD for national and transnational calls for research projects (50% of the NP/NS for RD supports both), it is of crucial importance to consider that in 29% of the NP/NS for RD neither national nor transnational calls for research projects are promoted. From the survey results, this appears to affect mainly the EU-13 Countries (57% of their NP/NS for RD does not support national nor transnational calls), for which it might be advisable to draw dedicated strategies. The eventual absence of the endorsement through the NP/NS for RD of transnational calls should not be taken as an overall result, as some countries (e.g., Italy or Lithuania) do participate in transnational calls, even if this participation is not directly promoted by their NP/NS for RD. In general, the strategies to enhance the promotion of national and transnational calls for research projects should be planned and enacted both at national level and European level; to this latter purpose, the EJP RD always tries to increase the dialogue with the national and regional stakeholders and elaborate interventions to counterbalance eventual inequalities between the countries.

The latter statement might be read as consistent with what emerges regarding the public funding initiatives, other than the NP/NS for RD, for research and/or networking in the RD field. Public funding initiatives appear to be present in 68% of the countries, with the participation to national/international funded projects. This somewhat positive result, compared to what emerged regarding the support to RD research could be explained by the fact that in many countries research on RD is probably addressed by general health research policies, while it should benefit from dedicated efforts and policies, and should receive a broader and specific attention within the NP/NS for RD. This proposition is of particular relevance for those countries (globally 12% of the countries) that declare to have only private funding initiatives for research and/or networking in the RD field or not having any support for research and/or networking at all (neither through the NP/NS nor through other public/private funding initiatives).

Alignment with Pillar 2 "Resources and services to foster solutions in the field of RD":

Key point: The constitution of dedicated RD advisory bodies for Research and Innovation and the attention to FAIR data to be reinforced within the NP/NS for RD appear to be the areas that require interventions for a better alignment with the actions of Pillar 2.

In many countries, there is an advisory body for EU Research and Innovation policy, but only in two countries this body is specific for rare diseases. The outcomes described for Pillar 1 related to the globally low support to research on RD could also be in connection with the absence of a RD dedicated advisory body, that could direct and optimize the efforts in RD research.

A dedicated advisory body could speed up the efforts in the RD field of the different countries, establishing a reference point committed to the advancement in multiple areas in the field.

A good alignment with the topics afforded by Pillar 2 of the EJP RD has been observed with respect to the support of data repositories and tools in RD research, endorsed globally by the NP/NS for RD and/or by other public or private funding initiatives in 78% of the countries.

Apart from the support given by the NP/NS for RD, data repositories and tools in RD research receive a greater support from other public than from private funding initiatives (44% vs 24%).

The data repositories and tools for RD research supported by the NP/NS for RD cover the topics mostly of "Registries catalogues", "Ontologies and codification", "Biobanks catalogues", "Support for clinical/translational research" and "Data deposition and analysis ", while the other areas, namely, "Tools", "Access and privacy control", "OMICS services", "Cell lines", "Animal models" or "Semantic standards" receive a lower attention. A similar distribution can be observed for the other public and private funding initiatives, with "Tools" receiving a better support by other public funding initiatives, when compared to the NP/NS for RD or to private funding initiatives.

A moderately good support to FAIR data is observed globally if considering the NP/NS for RD and/or other national RD initiatives (56% of the countries), with 21% of the countries enacting this support through the NP/NS for RD (with or without the addition of other initiatives). Alongside this partly encouraging general outcome, it must be pointed out that in 20% of the responding countries the support is enforced only through initiatives other than the NP/NS, and in other 16% of the countries FAIR data are not supported at all (neither through the NP/NS for RD, nor through other initiatives). The limited attention given to FAIR data in the NP/NS for RD could also rely on the fact, among other, that various NP/NS for RD have been approved at a date when the FAIR data principles were not yet widely disseminated.

Multidisciplinary holistic approaches for RD diagnostics and therapeutics receive globally a good endorsement through the NP/NS for RD. Only 19% of the NP/NS for RD do not to promote such approach. A worse outcome emerges with respect to initiatives other than the NP/NS for RD, as only 44% declares to do so. Overall, this appears as a favourable result, showing the presence of an attention to the RD patients' multidisciplinary take in charge, especially through the NP/NS for RD.

From the survey results it comes up that there is no evident disparity between EU-13 Countries and the other responding countries in respect to the topics investigated in this section of the survey, as 56% of the EU-13 Countries declare to have an advisory body of national experts for EU R&I (even if in only one EU-13 Country this is specific for RD), and that the area of support to data repositories and tools is rather well endorsed by their NP/NS (56%). A less encouraging result for the EU-13 Countries regards the support to data repositories and tools by other public initiatives (22%) as well by other private initiatives (12%). The data on the covered topics do not differ when compared to the global results of the survey.

Only one EU-13 Country supports FAIR data; the support is endorsed through initiatives other than the NP/NS for RD, and this emerges to be the only effort in this area in the EU-13 Countries. As for the adoption of multidisciplinary holistic approaches, these appear as rather present (67% of the EU-13 Countries), whereas other initiatives than the NP/NS for RD on this area require a dedicated effort (45% of the EU-13 Countries have such other initiatives).

Alignment with Pillar 3 "Capacity building and empowerment"

Key points: Trainings on FAIR data demand for a special dedicated and overall attention for an alignment with the Pillar 3 activities.

The attention dedicated to capacity building and empowerment reveals being widespread, with 78% of the responding countries declaring to endorse training activities in their NP/NS for RD, and 56% through other initiatives. When looking at the topics covered by the training activities backed up by the NP/NS for RD, "Empowerment of the patients", "Registries", "Data management", "Online education courses", "Standard and quality of genetics/genomics data in clinical practice and laboratories" are all rather well covered. Less attention is given to trainings on "Biobanks" and "Data quality", and, most important for the purposes of the present deliverable, it emerges that trainings on FAIR data are endorsed only in the NP/Ns of two countries. This is consistent with the outcome obtained for Pillar 2 regarding the endorsement of FAIR data that arises to be lacking in a high percentage of NP/NS for RD. The main effort for an alignment with the Pillar 3 activities relates heavily in the promotion, in the NP/NS for RD, of trainings dedicated to FAIR data.

Currently, it is not possible to assess, from the collected data, which topics are covered by training activities different from those promoted by the NP/NS for RD, as this information is lacking for several countries and will be collected in the forthcoming analysis of this series.

When focusing on EU-13 Countries, it can be observed that in these countries training activities are promoted at a certain degree (67%), suggesting that these countries are engaging in RD-dedicated education efforts.

Alignment with Pillar 4 "Accelerated translation of research projects and improvement of outcomes of clinical studies"

Key points: the activities that fall under the action of Pillar 4, and namely the rapid translation of research results in clinical studies and healthcare and the development of innovative methodologies tailored for clinical trials need intervention to achieve better results, particularly in EU-13 Countries.

Even if globally there appears to be a relative positive alignment towards the promotion of a rapid translation of clinical studies and healthcare (53% of the responding countries, if considering NP/NS for RD and/or other initiatives), it can be stated that the attention dedicated to this issue through the NP/NS for RD is not widespread and represents a topic that needs to be sustained to improve the alignment with the actions of Pillar 4.

More in detail, the survey results show a need to reinforce the rapid translation of the research results in clinical studies and healthcare, that is tackled only by 48% of the NP/NS for RD and by 28 % of other initiatives.

Similarly, a high percentage of countries (61%) declares that the NP/NS for RD does not promote the development of innovative methodologies tailored for clinical trials in RD, or not to have information on this topic (22%).

The above-described results could reflect the need to update the NP/NS for RD on these issues and indicate areas that need intervention to raise the alignment status with the EJP RD activities.

EU-13 Countries' specific needs, obstacles and advancements

In respect to the adoption of a NP/NS for RD, it can be observed that 90% of the 10 responding EU-13 Countries (counting also Hungary) adopted a Plan or Strategy at some stage. On the other hand, it arises that these NP/NS expired and have not been replaced to date (for some countries there is the information that in the period 2020-2021 the NP/NS was under update (Czech Republic, Romania), while for the other countries this information is not available from the survey results.

Considering the responses of the participating EU-13 Countries, the most relevant fields that have been pointed out by the survey participants, and that need to be faced to overcome the main perceived obstacles and barriers for the progress, improvement and translation of RD research results, are (named in order by decreasing frequency): "Funding", "Difficulties in accessing to national resources for funding of research and development of RD projects", "Lack of options for exploitation of research results at national level".

Other relevant obstacles and barriers indicated by the participating EU-13 Countries that require to be addressed with respect to the participation in EU/international projects in the RD field are, in order (by decreasing frequency): "Limited links to potential partners", "Lack of information on funding opportunities", "Bureaucratic application of funding procedures".

Reading these obstacles and barriers in connection with the results obtained in the sections addressing the alignment status with the four EJP RD Pillars, some considerations might be drawn. Namely, it emerged that the EU-13 Countries show criticalities in:

- The participation in national and transnational calls for research projects.
- The investments to share knowledge on RD.
- The development of data repositories and tools besides the efforts endorsed through the NP/NS for RD.
- The adoption of FAIR data.
- The promotion of the rapid translation of research results into clinical studies and healthcare.

Overall, considering the results from all the participating countries, it can be stated that the highest alignment with the activities of the EJP RD occurs currently with the actions of Pillar 3 "Capacity building and empowerment", followed first by the actions promoted by the activities of Pillar 2 "Resources and services to foster research on RD", and then of Pillar 1 "National and International Investments in the field of RD", whereas the lowest alignment seems to concern the activities promoted by Pillar 4 "Accelerated translation of research projects and improvement of outcomes of clinical studies".

In addition and integration to the conclusions above, some considerations on positive aspects regarding national RD activities can be reported based on the presentations held during the Strategy Meeting of July 2021. The countries that advised on their country experience indicated a growing involvement of the patients and patients' association in all levels of the RD dedicated strategies, a great participation of the ERNs, the introduction of national RD helplines, the improvement of the new-born screenings, the adoption of the Orpha nomenclature and the translation of the Orphacodes in their country-language.

Finally, it can be stated that, as expected, there have not been relevant differences between the responses given by countries that answered to the first and second edition on the survey. This can be motivated by the short space of time elapsed between the surveys, the outspread of the Covid-19 pandemic that channelled the resources ad efforts of the countries to face the emergency situation, and the fact that many NP/NS were expired or expiring by 2020.

Conclusions and next actions

The results collected through this survey allow to have a more precise overview in respect to what has been observed via the survey launched in autumn 2020 and will serve as starting point for the planning of specific and targeted EJP RD actions. They will serve furthermore as a baseline for a comparative analysis at month 55 of the EJP RD, when the Fourth Analysis of this series is foreseen.

The present results seem to confirm the main critical points already highlighted in D2.22 as well as the gaps to be filled to fulfil a good alignment with the proposals of the EJP RD.

More in detail, the presented outcomes point out several issues to be addressed to progress in the implementation process of the EJP RD proposals at national and global level. They can be considered as important tools to identify the challenges to be tackled by the future EJP RD activities. The identified issues are presented in the tables below, for the 4 Pillars, with some options and broad suggestions for dedicated actions, proposed also in the light of the discussions held at the occasion of the Strategy Meeting of July 2021. These suggestions have to be considered as general hints and points of reflection to be forwarded to the Pillar Leaders, who in turn can develop and work out specific and more informed interventions and actions in their Work Plans.

Table 8 Challenges and suggestions for actions for Pillar 1

Challenge	Suggested action
• promotion of RD national and transnational calls for research projects	• foster the dialogue with national and regional stakeholders to identify common and local needs • organise workshops (online and face to face), illustrating the EJP RD Funding Schemes possibilities and inviting especially national policy makers, in order to stimulate the awareness on the need of the national/local support to employ the EU opportunities • explore the bureaucratic "bottlenecks" of funding procedures to enhance dedicated strategies
• the support towards investments to share knowledge	• disseminate the information of the added value of the Networking Support Scheme (WP 7 of the EJP RD) between EJP RD Partners

Table 9 Challenges and suggestions for actions for Pillar 2

Challenge	Suggested action
• lack of dedicated RD advisory bodies for Research and Innovation	• promote the importance and stimulate the constitution of

	NMGs as RD multi-stakeholder bodies • stimulate all EJP RD countries to have representatives of Ministries (Research and/or Health) inside the Policy Board of the EJP RD to help the connection with national policy makers
• the adoption of the FAIR principles	• disseminate the importance of the adoption of the FAIR principles and encourage their inclusion in the NP/NS for RD
• the promotion of private and public funding initiatives, beyond the NP/NS for RD to support data repositories and tools for RD research	• encourage and maintain the connection between EJP RD and other RD projects and spread the information to raise awareness on their specific actions and potential • production of some generic material on FAIRification, Orpha nomenclature, data management standardisation and harmonisation to leverage the potential of the work of the EJP RD for the national RD registries • diffusion of the EJP RD dedicated Scheme for the RD Challenges to boost the public/private collaboration
• the fostering of multidisciplinary holistic approaches also beyond the NP/NS for RD	• encourage the participation of the NMGs as multistakeholder bodies

Table 10 Challenges and suggestions for actions for Pillar 3

Challenge	Suggested action
• lack of trainings on FAIR data, and other relevant topics	• trainings on FAIR data, to be addressed especially by the NP/NS for RD as well as trainings on other topics that emerged

	to be less covered, mainly on Biobanks and Data Quality. Strengthen the visibility of the existing training courses promoted by the EJP RD • spread the information on the fellowships for PhD students for the mobility inside and between the ERNs that are promoted by the EJP RD

Table 11 Challenges and suggestions for actions for Pillar 4

Challenge	Suggested action
• the reinforcement of the promotion of both the rapid translation of research results in clinical studies and healthcare	• Promote closer, regular contact between researchers, research centres and academia, and the patients
• the development of innovative methodologies tailored for clinical trials.	• Disseminate the Innovative methodologic aspects for clinical trials developed in the EJP RD.

Regarding the EU-13 Countries, the reported difficulties and barriers for the development, improvement and translation of RD research results should be addressed by increasing funding dedicated to RD, facilitating the access to funding for research and development of RD projects, and enlarging the possibilities to exploit research results at national level. These efforts should be complemented by initiatives to enhance the participation in EU/International projects in the RD field, with a special attention to facilitating the links to potential partners, easing the retrieval of information on funding opportunities, assisting on bureaucratic application on funding procedures and improving the skills in drafting proposals.

The results obtained through the survey launched in 2020 have been presented at the occasion of the Policy Board Meeting in January 2021and the data, integrated with the outcomes obtained by the 2021 survey have been shown as highlights during the Strategic Meeting with relevant RD policy stakeholders held in July 2021. The presentation of the outcomes achieved through the Strategic Meeting will be the objective of a dedicated deliverable (D2.25 "First Report from strategic workshop with national policy makers"), to be submitted in M 34.

Furthermore, the outcomes of the present analysis will help in identifying the R&I needs, that are yearly mapped in Task 2.2.

In summary, it seems highly desirable that further actions of the EJP RD include specific strategies to be implemented both at national and international level to counterbalance the most critical emerged issues in the areas of research, use of data and resources, empowerment of all stakeholders, and fast scientific progress.

The **next actions** will be:

1. The results of the present document will be shared internally with each Pillar to stimulate the reflection on how to face the specific arisen issues and plan dedicated interventions.
2. For the Fourth Analysis of national state of play and alignment process with EJP RD, a dedicated survey will be launched, and the results will be reported in month 55 of the EJP RD. The survey will ideally reach the countries that did not participate to the analysis to date and will allow a comparison with the present results with the aim to draw an assessment of the impacts of the EJP RD actions on relevant national RD policies and initiatives.
3. The results of the Fourth Analysis will be presented at the Strategic Workshop with relevant policy stakeholders, foreseen for month 57.
4. The results of the present deliverable will be transmitted to Task 2.2 Leaders to feed the mapping of the R&I needs for targeted actions that will encounter the emerged criticalities and increase the effectiveness of the EJP RD efforts.
5. The submission at M34 of D2.25 “First Report from strategic workshop with national policy makers” that will furnish further important indications on the way forwards, as identified and discussed by relevant RD stakeholders during the meeting.

Annex 1 Survey "National Plans and Strategies for Rare Diseases"

National Plans and Strategies for Rare Diseases Edition 2021

This survey aims at collecting information from EU Member States on the state of the art regarding the development and implementation of National Plans and Strategies for rare diseases and on the alignment process with the European Joint Programme on Rare Diseases, EJP RD, (GA 825575) relevant/complementary actions performed at national level, with a specific focus on EU 13-Countries in respect to their specific needs, obstacles and advancements. Please fill in the survey by referring to the National Plan or Strategy for rare diseases in your Country. Be aware that some questions do not relate directly to the National Plan or Strategy for rare diseases but concern the rare disease field in a broader context.

GDPR

This survey form is specifically dedicated to collect information for the purpose of the European Joint Programme on Rare Diseases, EJP RD, WP 2 "Integrative Research and Innovation Strategy", "Task 2.5 "Translation/impact of prioritization on national and EU strategies". We collect Personal Data freely provided by the user including (but not limited to): name, email address, and any other details specifically asked in registration forms. EJP RD does not share personally identifiable information with unrelated Third Parties. However, we may disclose, transfer or share your Personal Data- anonymized or in its original format- with certain third parties without further notice to you, only for the purpose of the organization and follow up of this event. Information collected on this form will be held in compliance with the General Data Protection Regulation (EU Regulation 2016/679) (GDPR) of the European Parliament and of the Council of 27 April 2016 on the protection of natural persons with regard to the processing of personal data and on the free movement of such data. Data will be processed and stored for a maximum of 15 years. If you want to have more information on data processing, for example know how your personal data is being processed, or if you want to exercise your rights according to articles 15-22 of the GDPR, or if you notice a personal data breach according to Articles 33-34, please contact the data controller who determines the purposes and means of the processing of personal data. Contact details: coordination@ejprarediseases.org (mailto:coordination@ejprarediseases.org)

I have read the above mentioned information and

(Check any that apply)

1. I authorise the processing of personal data, in compliance with the European General Data Protection Regulation, Reg (EU) 2016/679 for the specific purpose they are collected (any communication of personal data to private or public subject will be allowed only for the specific purpose they are collected)

2. I authorise to be contacted for involvement in future collaborative initiatives, which might fall within the scope of my research activity I authorise to be contacted for dissemination and communication activities (e.g., newsletters, invitations to meetings)

General information

First Name

Last Name

Email address

Institution

Country

Is there an approved National Plan/Strategy for rare diseases (NP/NS for RD) in your Country?

- *Yes*
- *No*
- *Yes, but the NP/NS for RD of my country expired*
- *No, but it is under development in my country*
- *I don't know*

Please provide the link to the NP/NS for RD of your country (active or expired)

When was the NP/NS for RD approved in your country?

Is there a periodical evaluation of the NP/NS for RD in your country?

- *Yes*
- *No*
- *I don't know*

How many editions of the NP/NS for RD has your country adopted by now?

If the NP/NS for RD expired, please specify when it expired

Are you directly involved in the implementation or development of the NP/NS for RD of your country?

- *Yes*
- *No*

If not, could you kindly provide a contact of a person who is directly involved in the NP/NS for RD of your country?

- *Yes*
- *No*

Please provide the name of the person who is directly involved in the NP/NS for RD of your country

Please provide the email address of the person who is directly involved in the NP/NS for RD of your country

National and International Investments in the field of RD

Does the NP/NS for RD of your country promote national calls for research projects?

- *Yes*
- *No*
- *I don't know*

If yes, please specify on which topic

Does the NP/NS for RD of your country promote transnational calls for research projects?

- *Yes*
- *No*
- *I don't know*

If yes, please specify on which topic

Does the NP/NS for RD of your country foresee investments to share knowledge?

- *Yes*
- *No*
- *I don't know*

If yes, please specify on which topic

Resources and Services to foster research on RD

Does the NP/NS for RD of your country support data repositories and tools for research on RD?

- *No*
- *I don't know*
- *Yes, the NP/NS for RD promotes the development of data repositories and tools for RD research*
- *Yes, the NP/NS for RD promotes the implementation of data repositories and tools for RD research*
- *Yes, the NP/NS for RD promotes both the development and implementation of data repositories and tools for RD research*

If the NP/NS for RD of your country supports data repositories and tools for research on RD, please specify the topic (possible multiple choice)

- *Registries catalogue*
- *Biobanks catalogue*
- *Ontologies and codification*
- *OMIC services*
- *Cell lines*
- *Animal models*
- *Semantic Standards*
- *Support for clinical/translational research*
- *Access & privacy control*
- *Data deposition & analysis*
- *Tools*
- *Other*

Does the NP/NS for RD of your country support FAIR* data? (Findable, Accessible, Interoperable, Reusable)

- *Yes*
- *No*

- *I don't know*

If yes, please specify how the NP/NS for RD of your country supports FAIR data

Does the NP/NS for RD promote the adoption of multidisciplinary/holistic approaches for RD?

- *Yes*
- *No*
- *I don't know*

If yes, please specify on which topic

Capacity building and empowerment

Does the NP/NS for RD of your country promote training activities for RD?

- *Yes*
- *No*
- *I don't know*

If yes, on which topics? (Possible multiple choice)

- *Data management*
- *Data quality*
- *FAIR data*
- *Standards and quality of genetics/genomics data in clinical practice and laboratories*
- *Registries*
- *Biobanks*
- *Empowerment of the patients*
- *Online education courses*

Accelerated translation of research projects and improvement of clinical studies and healthcare

Does the NP/NS for RD of your country promote a rapid translation of research results in clinical studies and healthcare?

- *Yes*
- *No*
- *I don't know*

If yes, please specify on which topic

Does the NP/NS for RD of your Country promote the development of innovative methodologies tailored for clinical trials?

- *Yes*
- *No*
- *I don't know*

If yes, please specify on which topic

Are there other relevant initiatives for RD in your country, than the NP/NS?

- *Yes*
- *No*
- *I don’t know*

Are you directly involved in other initiatives for RD in your country, other than the NP/NS?

- *Yes*
- *No*

If yes, please specify on which topic

Are there other public funding initiatives that promote national calls for research/networking in the field of RD in your country, apart from the NP/NS for RD?

- *Yes*
- *No*
- *I don't know*

If yes, please specify on which topic

Are there private funding initiatives for national calls for research/networking in the field of RD in your country?

- *Yes*
- *No*

- *I don't know*

If yes, please specify on which topic

Are there other public funding initiatives that promote transnational calls for research/networking in the field of RD in your country, apart form the NP/NS for RD?

- *Yes*
- *No*
- *I don't know*

If yes, please specify on which topic

Are there private funding initiatives for transnational calls for research/networking in the field of RD in your country?

- *Yes*
- *No*
- *I don't know*

If yes, please specify on which topic

Are investments to share knowledge foreseen by other public national initiatives than the NP/NS for RDs?

- *Yes*
- *No*
- *I don't know*

If yes, please specify on which topic

Are investments to share knowledge foreseen by private national initiatives for RD?

- *Yes*
- *No*
- *I don't know*

If yes, please specify on which topic

Is there an advisory body of national experts for Research and Innovation in your country?

(Possible multiple choice)

- *Yes, and advisory exists, but not specific for RD*
- *Yes, an advisory body exists, specific for RD*
- *No*
- *I don't know*

Do other public initiatives than the NP/NS for RD of your country support data repositories and tools for research on RD?

- *Yes, other public initiatives promote the development of data repositories and tools for RD*
- *research*
- *Yes, other public initiatives promote the implementation of data repositories and tools for RD*
- *research*
- *Yes, other public initiatives support both the development and implementation of data*
- *repositories and tools for RD research*
- *No*
- *I don't know*

If yes, please specify on which topic (possible multiple choice)

- *Registries catalogue*
- *Biobanks catalogue*
- *Ontologies and codification*
- *OMIC services*
- *Cell lines*
- *Animal models*
- *Semantic standards*
- *Support for clinical/translational research*
- *Access & privacy control*
- *Data deposition and analysis*
- *Tools*

Do other private initiatives of your country support data repositories and tools for

research on RD?

- *Yes, other private initiatives promote the development of data repositories and tools for RD*
- *research*
- *Yes, other private initiatives promote the implementation of data repositories and tools for RD*
- *research*
- *Yes, other private initiatives support both the development and implementation of data*
- *repositories and tools for RD research*
- *No*
- *I don't know*

If yes, please specify on which topic (possible multiple choice)

- *Registries catalogue*
- *Biobanks catalogue*
- *Ontologies and codification*
- *OMIC service*
- *Cell lines*
- *Animal models*
- *Semantic standards*
- *Support for clinical/translational research*
- *Access & privacy control*
- *Data deposition and analysis*
- *Tools*

Do other national initiatives for RD than the NP/NS support FAIR* data? (*Findable, Accessible, Interoperable, Reusable)

- *Yes*
- *No*
- *I don't know*

If yes, please specify how FAIR data are supported by other initiatives than the NP/NS for RD

Are there other national initiatives than the NP/NS for RD that promote the adoption of multidisciplinary approaches for RD?

- *Yes*
- *No*
- *I don't know*

If yes, please specify on which topic

Do other initiatives than the NP/NS for RD support training activities for RD?

- *Yes*
- *No*
- *I don't know*

If yes, please specify on which topic (possible multiple choice)

- *Data management*
- *Data quality*
- *FAIR data*
- *Standards and quality of genetics/genomics data in clinical practice and laboratories*
- *Registries*
- *Biobanks*
- *Empowerment of the patients*
- *Online education courses*
- *Other*

Do other initiatives than the NP/NS for RD promote a rapid translation of research results in clinical studies and healthcare?

- *Yes*
- *No*
- *I don't know*

If yes, please specify on which topic

Do other initiatives than the NP/NS for RD promote the development of innovative methodologies tailored for clinical trials?

- *Yes*
- *No*
- *I don't know*

If yes, please specify on which topic

Do you belong to an EU-13 Country?

- *Yes*
- *No*

If yes, to which EU-13 Country do you belong?

- *Bulgaria*
- *Croatia*
- *Cyprus*
- *Czech Republic*
- *Estonia*
- *Hungary*
- *Latvia*
- *Lithuania*
- *Malta*
- *Poland*
- *Romania*
- *Slovakia*
- *Slovenia*

Based on your experience, what are the main obstacles and barriers in your country for the development, improvement and translation of RD research results? (Possible multiple choice)

- *Language*
- *Funding*
- *Difficulties in accessing to national resources for funding research and development of RD*
- *projects*
- *Lack of options*

Regarding your participation in EU/International projects in the RD field, what do you estimate to be the most important/obstacles and barriers? (Possible multiple choice)

- *Limited links on drafting proposals*
- *Lack of information on funding opportunities*
- *Limited links to potential partners*
- *Bureaucratic application on responding procedures*
- *Irrelevance of programme topics and goals to own research agenda*
- *Quality of support provided by national contact points*

Do you want to highlight any other aspect regarding the national policies and initiatives for RD of your country that were not included in the present survey?

- *Yes*
- *No*

If yes, please specify

Annex 2 Links to the NP/NS for RD

Belgium:

https://www.health.belgium.be/sites/default/files/uploads/fields/fpshealth_theme_file/belgisch_plan_voor_zeldzame_ziekten.pdf

Bulgaria:

https://www.mh.government.bg/media/filer_public/2015/04/17/programa-redki-bolesti-2009-2013.pdf

The Bulgarian National Programme for Rare Diseases (2009 - 2013) officially ended on December 31, 2013:

- Since then, there have been no official talks to adopt a second edition of the Programme;

Croatia:

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