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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
MS	Member States
NMG(s)	National Mirror Group(s)
NP	National Plan(s)
NS	National Strategy(ies)
PB	Policy Board
RD	Rare Disease(s)
WP	Work Package

Executive summary

The European Joint Programme on Rare Diseases (EJP RD) has among its major objectives to improve the integration, efficacy, production and social impact of research on rare diseases (RD) through the development, demonstration and promotion of Europe/world-wide sharing of research and clinical data, materials, processes, knowledge and know-how.

In EJP RD 's Work Package 2 (WP2) "Integrative research and innovation strategy" 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 prioritization on national and European Union (EU) strategies.

An analysis performed periodically, involving the EU Member States on the EJP RD relevant/complementary actions performed at national level, with a specific focus on EU-13 Countries¹, is among the key instruments required to fulfil the objectives of WP2. For this analysis, data were collected through a specific survey performed in 2020, targeting National Mirror Groups if constituted, or in their absence, to relevant (deeply involved) persons in the national plans of strategies, addressing the topics of the 4 major Pillars of the EJP RD. Attention is given to National Plans (NP) and National Strategies (NS) for RD, and to other relevant national RD initiatives, as tools to reach a common strategy at European level with the aim to: (i) facilitate the consistency of national initiatives with the main issues and existing good practices in the RD field, (ii) prepare the ground for possible synergies and cooperative approaches for the benefit of RD patients, as stated in the Council Recommendation of 8th June 2009 on an action in the field of rare diseases.

The outcomes of the present study furnish a first description of the alignment status of national RD initiatives/undertakings with the EJP RD relevant complementary actions in the countries who participated to the survey, contributing therefore to the process of evaluation of the achievements and gaps that need specific interventions for the advisable advancements in the RD field.

Highlights and next actions

- The present report gives a partial picture, based on the responses of 21 countries (16 EU and 5 Associated/Other countries) that participated to the survey in 2020. These results can contribute to a first understanding of the alignment of the national policies for RD with the action promoted by the EJP RD and provides the base for further developments.
- Through the identification of the achievements and critical issues, specific actions to be implemented at national and international level are suggested, 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.
- Globally, the national RD policies for which there seem to be 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

¹ Bulgaria, Croatia, Cyprus, Czechia, Estonia, Hungary, Latvia, Lithuania, Malta, Poland, Romania, Slovakia, Slovenia

research on rare diseases" (that fall respectively under Pillar 3 and Pillar 2 of the EJP RD). In EU-13 countries, some differences (e.g., the absence of a dedicated RD advisory body in 5 of the 6 EU-13 Countries) and similarities (e.g., the limited links to potential partners has been indicated as the most important obstacle and barrier to the participation in EU/International research projects in the RD field in all 6 EU-13 Countries) have been observed.

- The national RD actions that evidence a 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. In EU-13 countries, funding and investment are the main problems, according to the survey results.
- The WP2 Strategic Meeting of July 2021 will facilitate the dialogue between relevant policy stakeholders on the alignment status with EJP RD, emerging from the first survey results. The meeting will focus more in detail on three main points: (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.
- For the Third Analysis (within the EJP RD) of national state of play and alignment process of EJP RD, an enhanced edition of the survey will be distributed in order to update the outcomes that are the objective of the present document and to obtain information from the countries that are not yet represented in this Second Analysis.

Introduction and Objective

One of the main actions of Work Package 2 (WP2) is to focus on the progress of the European Joint Programme on Rare Diseases (EJP RD) research and innovation strategy in connection to all related stakeholders. Within this framework, Task 2.5 leaders are expected to collect information from EU Member States (MS) on the EJP RD relevant/complementary actions performed at national level, via a specific survey targeting National Mirror Groups (NMGs). In the absence of NMGs in many countries, key persons (deeply involved in the national plan/strategy) received the survey, and they were asked to forward the survey to the relevant person if those are currently the ones more involved in the National Plans (NP)/National Strategies (NS).

The present deliverable D2.22 “Second Analysis of national state of play and alignment process with the European Joint Programme on Rare Diseases” is a public report that aims at exploring the state of the art of the existing undertakings and relevant actions at national, European and international level in the field of rare diseases (RD). The analysis concentrates on the 35 countries participating to the EJP RD. The results of this analysis encompass: (i) the initiatives and efforts regarding RD policies in EU Member States (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. A specific consideration is given to EU-13 Member States with respect to their specific needs, obstacles and advancements, as it was expressed in the Description of Action of the EJP RD.

The main objective of the present deliverable is therefore to analyse the existence of national policies/actions for RD and to assess their alignment with the actions promoted by the EJP RD, in EU countries and in Associated/Other countries participating in the EJP RD, with a special attention to EU-13 Countries. The focus on EU-13 Countries is motivated by the fact that these countries currently still lag behind in RD research so their wider participation is limited by challenges such as lack of RD research and training, or funding and economic constraints for attending international conferences/workshops. Therefore, their needs differ from those of other EU MS. The participation of EU-13 Countries in EJP RD was tailored to progressively increase their capacity and go beyond the initially available budgetary, administrative and scientific resources. It is therefore envisaged that the EU-13 Countries’ needs will be interpreted through the prioritization strategy.

D2.22 falls under the objectives of Pillar 0, WP2, more precisely of task 2.5 “Translation/impact of prioritization on national and EU strategies” and is the second document of a series of reports, regarding the evolution of national alignments with EJP RD.

The “First Analysis of national state of play and alignment process with EJP RD” was already submitted as Deliverable 2.21.

The analyses included here are intended to verify that the actions promoted by the EJP RD have a significant impact on the RD policies at national, European and

international level and serve as a basis for the identification of the topics that need to be developed, strengthened or enhanced for the general benefit of the RD community. The premise is that synergies would benefit the progress in this field.

The results submitted in this deliverable have also been presented at the occasion of the EJP RD Policy Meeting that took place in January 2021 with the presence of relevant policy makers and stakeholders.

The submission of the present deliverable has been postponed with respect to the planned due date, given the COVID-19 pandemic, which slowed down the data collection process. It was important to have as many respondents as possible, since currently gathered data through the survey will serve as reference for further developments.

Background

Since many years, several improvements have been achieved to continue progressing in the diagnosis, treatment and care for people with RD.

The Council Recommendation of June 8, 2009, on an action in the field of RD (2009/C 151/02)², a key “soft law” policy document, emphasized the need of EU MS to adopt Plans and Strategies for RD as essential tool, among others, to move forward in the RD field. It therefore encouraged MS to elaborate and adopt a Plan or Strategy for RD as soon as possible and preferably by the end of 2013, at the latest. In 2014, the European Commission published an Implementation Report³ addressed to the European Parliament, the Council, the European Economic and Social Committee and the Committee of the Regions, highlighting various actions to continue supporting MS, and stressing the need to continue to enhance the development of high-quality National RD Plans and Strategies in the EU.

Numerous initiatives of European Expert Groups/Committees for RD moved also in this direction, such as the Commission Expert Group on RD (CEGRD, 2014-2016), the European Union Committee of Experts on RD (EUCERD, 2012-2015), the European Project for RD National Plans Development (EUROPLAN, 2008-2011, 2012-2015)⁴, RD-ACTION, with its Overview Reports of 2016⁵ and 2018⁶.

Other relevant initiatives are summarized in Annex 1 of EJP RD's Deliverable 2.21 “First analysis of national state of play and alignment process with EJP RD”.

Methodology

In order to collect information on the state of the art regarding the national undertakings for RD, and especially of the development, implementation and update of the NP/NS for RD in EU countries (and in other countries participating to the EJP RD) a specific survey was elaborated in 2019 and 2020, after a deep assessment among WP2 leaders and the Coordination Team of the EJP RD. The survey, titled “National

² https://ec.europa.eu/health/ph_threats/non_com/docs/rare_com_en.pdf.

³ https://ec.europa.eu/health/sites/health/files/rare_diseases/docs/2014_rarediseases_implementationreport_en.pdf

⁴ [EUROPLAN, 2008-2011, 2012-2015](#)

⁵ <http://www.rd-action.eu/wp-content/uploads/2015/10/Overview-Report-State-of-the-Art-2016-Final-for-Portal.pdf>

⁶ <http://www.rd-action.eu/wp-content/uploads/2018/09/Final-Overview-Report-State-of-the-Art-2018-version.pdf>

Plans and Strategies for Rare Diseases", launched in 2020, is composed of multiple choice and open questions, as reported in Annex 1. The survey was divided into different sections:

- Sections 1-3 collect general information on the person/officer completing the survey (for further follow-up if necessary) and investigate the existence and some details regarding the NP/NS for RD of the country.
- Sections 4-7 of the survey reflect information linked to the areas of interest of the 4 non-transversal Pillars (Pillar 1-4) of the EJP RD, and are specifically dedicated to analysing the alignment of the NP/NS in the RD field 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".
- A special section of the survey (Section 8) is dedicated to the EU-13 Countries, and investigates 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.
While for the sections 4-7 the main topics of the four non-transversal Pillars of the EJP RD have been taken as reference for the elaboration of the questions, section 8 has been elaborated taking into account the results of the document "*Overcoming innovation gaps in the EU-13 Member States*", a report of the European Parliamentary Research Service (EPRS), of March 2018⁷. Such study aimed at exploring, identifying and shedding light on the reasons for the low and uneven participation rate of EU-13 Countries in research projects, in order to improve their performance in Horizon 2020 and FP9. The report stated that, after almost 20 years of access to the opportunities of the Framework Programmes (FPs)- the EU's primary instruments for the creation of the European Research Area-, the EU-13 Countries are still behind the results of other MS. It also highlights that the knowledge that is produced in the European context needs to be applied at national level, what can show some local difficulties in some countries. That uneven participation is an issue that impacts on the achievements of the higher objectives of the EU FPs as such.
- A final open question (Section 9) invites the respondent to give free comments on aspects regarding the NP/NS for RD, that are not included in the survey.

The estimated time for completing the survey is 45 minutes.

The percentage results of the answers to the survey provided by the countries which participated in the present study are assessed, and the results summarized in Annex 2, whereas a separate analysis is provided for the non-responding countries, based on the results collected by EUROPLAN and RD-ACTION ("*Overview Report on the State of the Art of Rare Disease Activities in Europe, 2018 Version*") and the formal texts of the National Plans/Strategies that are available through the internet (referring to their official and non-official English versions) (Annex 3).

⁷ [https://www.europarl.europa.eu/RegData/etudes/STUD/2018/614537/EPRS_STU\(2018\)614537_EN.pdf](https://www.europarl.europa.eu/RegData/etudes/STUD/2018/614537/EPRS_STU(2018)614537_EN.pdf)

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 LisyLime Survey Platform of INSERM.

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 MS⁸). It is important to note that the survey itself invites the respondent to forward it to another person who might be 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 content of the survey was finalized at the end of June 2020 and was distributed via email at the beginning of September 2020 to EUROPLAN contacts, Orphanet-INSERM contacts and EJP RD Partners.

The deadline for completing the survey was extended from 16th October to 2nd November 2020, following the requests of some of the respondents, and such extension was motivated by the great variety of topics investigated by the survey and by the burden of the emergency situation given by the COVID-19 pandemic.

Results

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

This summary of the results, apart from providing some introductory general information, has been structured to give detailed data on the responses gathered, question by question, following the same order that was established in the survey.

General information

A total of 21 countries (Figure 1) contributed to the results of the survey (Figure1). Thus, 60% of the 35 countries participating in the EJP RD are represented by the outcomes of this deliverable.

⁸ Cyprus is not participating in EJP RD but has been contacted as EU country

Of these:

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

The countries who answered the survey are:

Bulgaria, Canada, Czech Republic, Estonia, France, Germany, Hungary, Ireland, Israel, Italy, Lithuania, Luxembourg, Portugal, Romania, Serbia, Slovakia, Spain, Sweden, The Netherlands, Turkey, UK.

Missing responses from EU MS and Other/Associated countries:

Austria, Armenia, Belgium, Croatia, Cyprus, Denmark, Finland, Georgia, Greece, Latvia, Malta, Norway, Poland, Slovenia, Switzerland.

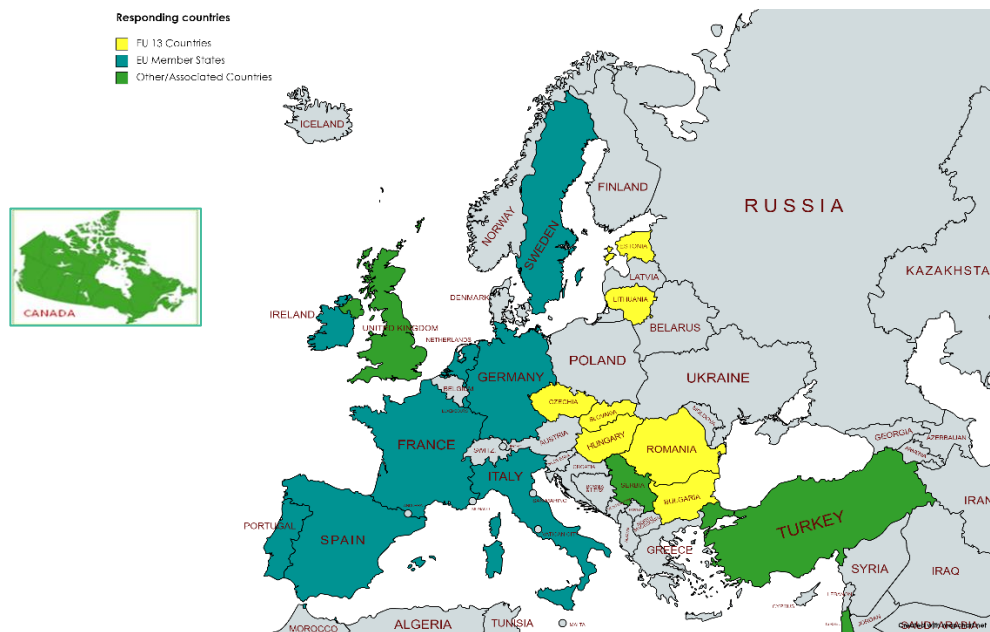


Figure 1. The 21 countries who answered to the survey (Blue: EU Member States, Yellow: EU 13 Countries, Green: Other Associated Countries)

Four additional surveys (with identified persons filling them) were totally or almost entirely completed but not submitted. With the consent of the persons who filled them out and who gave their agreement to be contacted for the purposes of the survey, those surveys have also been considered and included in the analysis. Two of these surveys came from countries that already had submitted completed surveys (France, UK), but contained additional information, and two other surveys were from countries not present in the fully submitted surveys (Canada, Slovakia).

76% of the surveys (n=16) 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 24% of the surveys have been completed by persons working in Universities, 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 (76%): Bulgaria, Czech Republic, Estonia, France, Germany, Ireland, Italy, Lithuania, Luxembourg, Portugal, Romania, Serbia, Slovakia, Spain, The Netherlands, UK
No (5%): Sweden
No but it is under development in my country (19%): Canada, Hungary, Israel, Turkey
I don't know: -

Figure 2 illustrate 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.

15 Countries transmitted a link to the NP/NS for RD of their country (see Annex 3).

In 3 of the countries declaring to have a NP/NS for RD, the NP/NS is expired (Bulgaria, Italy and Estonia) and currently there is no active NP/NS for RD.

Hungary answered that the country does not have a NP/NS for RD and that the NP/NS is under development, but from the results of RD-Action Report of 2018 it appears that a NP/NS for the period 2013-2020 did exist. Hungary should be included among the countries with an expired NP/NS for RD.

Ireland stated that the National Rare Disease Plan 2014-2018 was extended by a Ministerial order in 2019 and was part of the Irish Government's Programme for Government 2020.

Moreover, 69% (n=11) 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.

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

Country (21 respondents)	Year of approval	Year of expiry	Periodical evaluation	Under development
BULGARIA	2008	2013		
CANADA				✓
CZECH REPUBLIC	2010 (last edition)		✓	
ESTONIA	2014	2017		
FRANCE	2018 (last edition)		✓	
GERMANY	2013		✓	
HUNGARY	2013	2020		
IRELAND	2014	2018	✓	
ISRAEL				✓
ITALY	2013	2016		
LITHUANIA	2013		✓	
LUXEMBOURG	2018		✓	
PORTUGAL	2008 (last edition)		✓	
ROMANIA	2014		✓	
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		✓	

These data on the existence, expiry and update of the NP/NS for RD in EU countries are consistent with the results expressed in D2.21 "First Analysis of national state of play and alignment process with EJP RD". The only difference is related to Hungary that, from the present survey results, seems not to have a NP/NS for RD, whereas from the results of D2.21 a NP/NS for Hungary existed, but expired in 2020.

Alignment process with EJP RD

The following outcomes refer to the 16 countries who declare having a NP/NS for RD, active or expired, 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 (44%): France, Germany, Ireland, Luxembourg, Spain, The Netherlands, UK
No (44%): Bulgaria, Estonia, Italy, Lithuania, Portugal, Romania, Serbia
I don't know (12%): Czech Republic, Slovakia

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

Yes (56%): Czech Republic, France, Germany, Ireland, Luxembourg, Portugal, Spain, The Netherlands, UK
No (38%): Bulgaria, Estonia, Italy, Lithuania, Romania, Serbia
I don't know (6%): Slovakia

In comparison to the promotion of *national* calls (44%), a higher percentage of NP/NS for RD promote *transnational* calls for research projects on RD (56%).

The NP/NS for RD of seven countries (44%) promote both national and transnational calls for research projects on RD (France, Germany, Ireland, Luxembourg, Spain, The Netherlands, UK), while for two countries (13%) the NP/NS for RD promote only transnational calls (Czech Republic, Portugal). No country declares to promote only national calls for research projects.

For 6 Countries (38%) the NP/NS for RD does not support neither national nor transnational calls for research projects on RD (Bulgaria, Estonia, Italy, Lithuania, Romania, Serbia).

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.

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

Yes (44%): Bulgaria, Ireland, Lithuania, Luxembourg, Spain, The Netherlands, UK
No (44%): Czech Republic, Estonia, Germany, Italy, Portugal, Romania, Serbia
I don't know (12%): France, Slovakia

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.

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

Yes (81%): Czech Republic, Estonia, France, Germany, Ireland, Italy, Luxembourg, Portugal, Romania, Slovakia, Spain, The Netherlands, UK
No (19%): Bulgaria, Lithuania, Serbia
I don't know: -

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 are reported, covering mostly the field of translational and basic research.

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

Yes (56%): Bulgaria, France, Germany, Ireland, Italy, Lithuania, Portugal, The Netherlands, UK
No (25%): Czech Republic, Estonia, Luxembourg, Serbia
I don't know (19%): Romania, Slovakia, Spain

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.

Seven 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 (France, Germany, Ireland, Italy, Portugal, The Netherlands, UK), three countries⁹ (19%) of public funding initiatives only (Czech Republic, Estonia, Luxembourg) and one country (6%) of private funding initiatives only (Lithuania).

One country (6%) declares that there are no other public or private funding initiatives for research and/or networking in the field of RD (Serbia). The country answers that the NP/NS for RD does not foresee investments for networking to share knowledge on RD either.

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 (50%) Czech Republic, Estonia, France, Ireland, Italy, Luxembourg, Serbia, Spain
I don't know (25%): Lithuania, Slovakia, The Netherlands, UK
Yes, an advisory body exists specific for RD (12,5%): Portugal, Romania ¹⁰
No (12,5%): Bulgaria, Germany

The presence of an advisory body of national experts for EU Research and Innovation (R&I) is reported in 62% (n=10) of the countries (Czech Republic, Estonia, France, Ireland, Italy, Luxembourg, Portugal, Romania, Serbia, Spain) and in 2 of these countries (Romania and Portugal) the advisory body is specific for RD.

⁹ Romania, Slovakia and Spain answer “Yes” for public funding initiatives but “I don't know” for private funding initiatives.

¹⁰ Romania answers “Yes” both for the existence of an advisory body specific and non-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 (56%): France, Germany, Italy, Lithuania, Luxembourg, Portugal, Serbia, Spain, UK
Promoting the implementation of such data repositories and tools (19%): Czech Republic, Ireland, Romania
The NP/NS for RD does not foresee the support of data repositories and tools in rare disease research (13%): Bulgaria, Estonia
Promoting the development of new data repositories and tools (6%): The Netherlands
I don't know (6%): Slovakia

Globally, in 81% (n=13) of the countries the NP/NS for RD foresees the support of data repositories and tools dedicated to RD research (Czech Republic, France, Germany, Ireland, Italy, Lithuania, Luxembourg, Romania, Portugal, Serbia, Spain, The Netherlands, 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”	Czech Republic, France, Germany, Ireland, Italy, Lithuania, Luxembourg, Romania, Serbia
“Ontologies and codification”	Czech Republic, France, Germany, Lithuania, Luxembourg, Spain, UK
“Support for clinical/translational research”	Czech Republic, France, Ireland, Italy, Portugal, The Netherlands, UK
“Biobanks catalogues”	Czech Republic, France, Ireland, Italy, Lithuania, The Netherlands
“Data deposition & analysis”	France, Ireland, Lithuania, Portugal, UK
“Tools”	France, Lithuania, UK
“Access & privacy control”	France, Lithuania
“OMICS services”	France
“Cell lines”	Italy
“Animal models”	Italy
“Semantic standards”	France
“Other”	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?

No (50%): Bulgaria, Estonia, Germany, Lithuania, Luxembourg, Portugal, Serbia, UK
Yes (25%): Czech Republic, France, Italy, The Netherlands
I don't know (25%): Ireland, Slovakia, Spain, Romania

The table below (Table 3) shows the covered topics in the 4 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”	Czech Republic, France, Italy, The Netherlands
“Support for clinical/translational research”	Czech Republic, Italy, The Netherlands
“Biobanks catalogue”	Italy, The Netherlands
“Animal models”	France, The Netherlands
“Data deposition and analysis”	France; The Netherlands
“Cell lines”	The Netherlands
“Semantic standards”	The Netherlands
“Access & privacy control”	The Netherlands
“Tools”	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?

No (38%): Czech Republic, Estonia, Ireland, Lithuania, Luxembourg, Serbia
Yes (31%): Bulgaria, Italy, Portugal, The Netherlands, UK
I don't know (31%): France, Germany, Romania, Slovakia, Spain

The table below (Table 4) shows the covered topics in the 5 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, Portugal, The Netherlands
“Support for clinical/translational research”	Bulgaria, The Netherlands
“Biobanks catalogue”	Italy, Portugal, The Netherlands
“Animal models”	Portugal, The Netherlands
“Data deposition and analysis”	Bulgaria, The Netherlands
“Cell lines”	Portugal The Netherlands
“Semantic standards”	-
“Access & privacy control”	The Netherlands
“Tools”	The Netherlands
“Other”	UK-Patient Registries

Analysing the results of these last three items of the survey, it appears that, in addition to the support given for data repositories and tools in RD research by the NP/NS for RD, in 2 countries (13%) the support is given also by both other public and private funding initiatives (Italy and The Netherlands), in 2 countries (13%) also by private funding initiatives (Portugal and UK), and in two countries (13%) also by other public funding initiatives (Czech Republic and France¹¹).

Furthermore, in 6 countries (38%) the support is given by the NP/NS for RD only (Germany, Lithuania, Luxembourg, Romania, Serbia, Spain¹²). In one country (6%) the support is given by private funding initiatives only (Bulgaria).

In one country (6%) there is lack for 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)¹³.

Finally, one country 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.

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.

Do the NP/NS for RD or other initiatives for RD support FAIR* data in your country?
[*FAIR: Findable, Accessible, Interoperable, Reusable]

No, NP/NS for RD and no other initiatives support FAIR data (25%): Bulgaria, Lithuania, Romania, Serbia
Yes, both NP/NS for RD and other initiatives for RD (25%): France, Germany, Ireland, Spain
Yes, only other initiatives for RD support FAIR data but not the NP/NS for rare diseases (25%): Czech Republic, Italy, Luxembourg, The Netherlands
I don't know (19%): Estonia, Portugal, Slovakia
Yes, only the NP/NS for RD (6%): UK

Considering the outcomes shown in the table above, globally, in 56% (n=9) of the countries FAIR data are supported by the NP/NS for RD and/or by other initiatives.

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

¹¹ France answers “Yes” for other public funding initiatives but “I don’t know” for private funding initiatives

¹² Germany answers that there are no other public funding initiatives, but “I don’t know” regarding private initiatives and Romania and Spain answer “I don’t know” with regard both to other public and private initiatives. Ireland answers that there are no private funding initiatives and “I don’t know” for other public funding initiatives.

¹³ One country answers not to know if data repositories and tools are supported by the NP/NS for RD and/or by other public or private funding initiatives.

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 or by other initiatives for RD of your country?

Yes, by both, NP/NS for RD and by other initiatives for RD (63%): Bulgaria, France, Germany, Ireland, Italy, Lithuania, Portugal, Romania, Spain, The Netherlands
Yes, only by the NP/NS for RD (19%): Luxembourg, Serbia, UK
Not by NP/NS for RD and not by other initiatives for RD (12%): Czech Republic, Estonia
I don't know (6%): Slovakia
Yes, only by other initiatives for RD: -

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 81% (n=13) of the countries.

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).

Pillar 3 “Capacity building and empowerment”

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

Yes (75%): Bulgaria, Czech Republic, France, Germany, Ireland, Lithuania, Luxembourg, Portugal, Romania, Serbia, Spain, UK
No (19%): Italy, Estonia, The Netherlands
I don't know (6%): Slovakia

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, Czech Republic, France, Ireland, Lithuania, Luxembourg, Portugal, Romania, Serbia, Spain
“Registries”	Bulgaria, Czech Republic, France, Ireland, Lithuania
“Online education courses”	Czech Republic, Ireland, Lithuania, Portugal, Romania
“Data management”	Bulgaria, France, Ireland, Lithuania
“Standard and quality of genetics/genomics data in clinical practice and laboratories”	Bulgaria, Czech Republic, Lithuania, Romania
“Biobanks”	Ireland, Lithuania
“Data quality”	Bulgaria, Lithuania
“Other”	Lithuania, UK, Germany
“FAIR data”	-

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 (75%): Bulgaria, Czech Republic, Germany, Ireland, Italy, Lithuania, Portugal, Serbia, Slovakia, Spain, The Netherlands, UK
I don't know (19%): Estonia, France, Romania
No (6%): Luxembourg

9 countries (56%) promote/support training activities both through the NP/NS for RD and through other national initiatives (Bulgaria, Czech Republic, Germany, Ireland, Lithuania, Portugal, Serbia, Spain, UK).

One country (6%) supports training activities only through the NP/NS for RD (Luxembourg) and 2 countries (13%) only through initiatives other than NP/NS for RD (Italy and The Netherlands).

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?

No (50%): Bulgaria, Czech Republic, Estonia, Germany, Italy, Lithuania, Portugal, Serbia
Yes (37%): France, Ireland, Romania, Spain, The Netherlands, UK
I don't know (13%): Slovakia, Luxembourg

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?

Yes (38%): Bulgaria, Ireland, Italy, Portugal, The Netherlands, UK
No (38%): Czech Republic, Estonia, Germany, Lithuania, Luxembourg, Serbia
I don't know (24%): France, Romania, Slovakia, Spain

These 2 questions are complementary. Globally, 9 countries (56%) 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 (Bulgaria, France, Ireland, Italy, Portugal, Romania, Spain, The Netherlands, UK).

3 countries (19%) 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, The Netherlands, UK). The other 3 countries affirming that their NP/NS for RD promotes the rapid translation of research results in clinical studies and healthcare answer not knowing whether other initiatives do so (France, Romania, Spain).

In 5 countries (31%) the rapid translation of research results in clinical studies and healthcare is not promoted by the NP/NS for RD, nor by other initiatives (Czech Republic, Estonia, Germany, Lithuania, Serbia). Three countries (19%) answer that there

are only initiatives than the NP/NS for RD for the rapid translation of research results in clinical studies and healthcare (Bulgaria, Italy, Portugal).

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.

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.

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

No (56%): Bulgaria, Czech Republic, Estonia, Germany, Italy, Luxembourg, Romania, Serbia, The Netherlands
Yes (25%): Ireland, Lithuania, Portugal, UK
I don't know (19%): France, Slovakia, Spain

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

Seven of the EU-13 Countries answered to the survey (Bulgaria, Czech Republic, Estonia, Hungary, Lithuania, Romania, Slovakia¹⁴). The Bulgarian and the Estonian NP/NS for RD are actually not in force and expired respectively in 2013 and 2017. See Figure 3 for a visualization of the countries that declared to have an active or expired NP/NS for RD. The Hungarian NP/NS for RD is under development¹⁵.

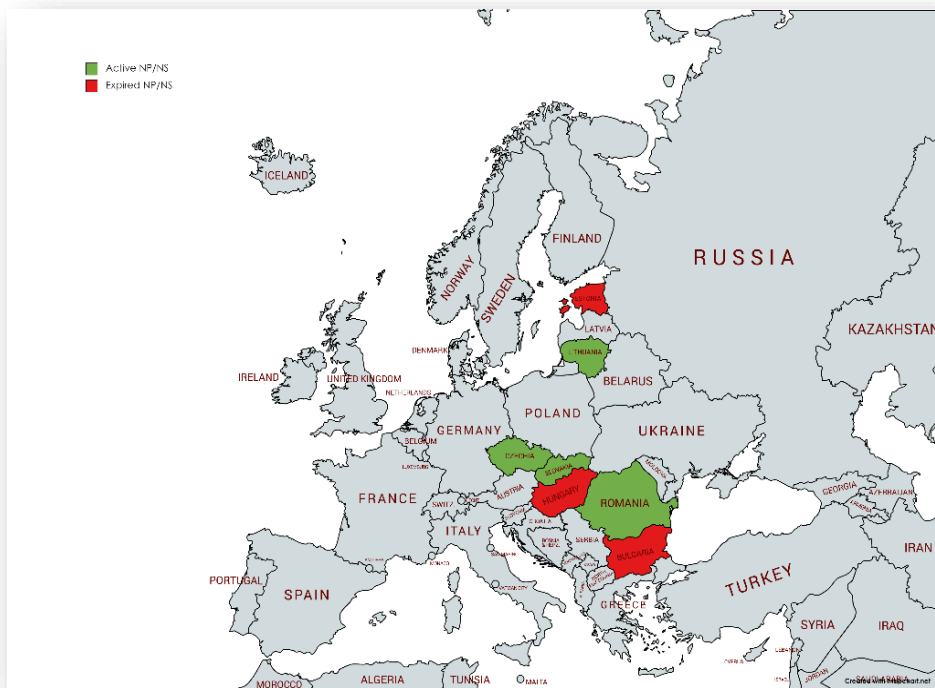


Figure 3. In green the active NP/NS for RD and in red the expired NP/NS for RD of the EU-13 responding countries

As to the questions on the main perceived obstacles and barriers for the development, improvement and translation of RD research results, “Funding” is indicated by 83% (of the participating EU-13 Countries; “Difficulties in accessing to national resources for funding of research and development of RD projects” were mentioned by 67%) of the countries; “Lack of options for exploitation of research results at national level” also by 67%; whereas “Language” and “Other” are pointed out by 17% of the responding countries.

¹⁴ 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.

¹⁵ As declared in the present survey, although it has to be considered that a Hungarian NP for RD exists and expired in 2020;

http://download2.eurordis.org/rdpolicy/National%20Plans/Hungary/2.Hungary_RD%20National%20Plan_2013-2020_English.pdf

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, Czech Republic, Estonia, Lithuania, Romania
"Difficulties in accessing to national resources for funding of research and development of RD projects"	Czech Republic, Lithuania, Romania
"Lack of options for exploitation of research results at national level "	Czech Republic, Estonia, Lithuania
"Language"	Slovakia
"Other"	Slovakia

In the free text comments, the lack of funding is once again 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, 100% 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" (67%), "Bureaucratic application on reporting procedures" (50%), "Quality of support provided by national contact points" (50%), "Limited skills on drafting proposals" (33%), "Irrelevance of programme topics and goals to own research agenda" (17%).

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, Czech Republic, Estonia, Lithuania, Romania, Slovakia
“Lack of information on funding opportunities”	Czech Republic, Estonia, Lithuania, Romania
“Bureaucratic application on reporting procedures”	Czech Republic, Estonia, Romania
“Quality of support provided by national contact points”	Czech Republic, Romania, Slovakia
“Limited skills on drafting proposals”	Czech Republic, Lithuania
“Irrelevance of programme topics and goals to own research agenda”	Lithuania

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 6¹⁶ EU-13 Countries who participated in the survey (Bulgaria, Czech Republic, Estonia, Lithuania, Romania, Slovakia) with the 4 EJP RD Pillars highlighted in points:

¹⁶ Hungary is not included in the present results, as it declared, as stated previously, not to have a NP/NS for RD, and has responded neither to the items investigating the alignment with the 4 EJP RD Pillars, nor the section dedicated to EU 13 Countries

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

- In 4 countries (67%, Bulgaria, Estonia, Lithuania, Romania) the NP/NS for RD do not promote national calls for research projects on RD, and 2 countries, (Czech Republic and Slovakia) answer “I don’t know”
- In one country (17%, Czech Republic) the NP/NS for RD promote transnational calls for research projects. In 4 countries (67%, Bulgaria, Estonia, Lithuania, Romania) they do not promote transnational calls for research projects, and one country answers “I don’t know” (Slovakia).
- In 2 countries (33%, Bulgaria and Lithuania) the NP/NS for RD foresee investments to share knowledge on RD. In 3 countries (50%, Czech Republic, Estonia, Romania) they do not foresee investments to share knowledge on RD, and one country answers “I don’t know” (Slovakia).

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

- 4 countries (67%, Czech Republic, Estonia, Romania, Slovakia) have other public funding initiatives for research and/or networking in the field of RD, and 2 countries (33%, Bulgaria and Lithuania) do not have such initiatives.
- 2 countries (33%, Bulgaria and Lithuania) have private funding initiatives for research and/or networking in the field of RD. 2 countries (33%, Czech Republic and Estonia) do not have these funding initiatives for research and/or networking in the field of RD, and two countries answered “I don’t know” (Romania and Slovakia).

Pillar 2 “Resources and services to foster research on RD”

- In 3 countries (50%, Czech Republic, Estonia, Romania) there is an advisory body of national experts for EU Research and Innovation policy, and in one of these countries (Romania), the advisory body is specific for RD. One country (17%, Bulgaria) declares the absence of an advisory body, and 2 countries answered “I don’t know” (Lithuania and Slovakia).
- In 3 countries (50%, Czech Republic, Lithuania, Romania) the NP/NS for RD foresee the support of data repositories and tools in RD research, in 2 countries (33%, Bulgaria and Estonia) the support is not foreseen, and 1 country answered “I don’t know” (Slovakia).
- The topics supported by the NP/NS for RD of the 3 above cited countries (Czech Republic, Lithuania, Romania) with regard to data repositories and tools for RD research are named in this order: “Registries catalogues”, “Ontologies and codification”, “Support to clinical/translational research”, “Biobanks catalogues”, “Data deposition and analysis”, “Tools”, and “Access & privacy control”.
- With regard to other public funding initiatives for the development of data repositories and tools, these are reported in one country (17%, Czech Republic), and not supported in 3 countries (50%, Bulgaria, Estonia, Lithuania), while 2 countries answered “I don’t know” (Slovakia and Romania).

- The topics covered by the country with other public funding initiatives for the development of data repositories and tools (Czech Republic) are “Registries catalogues” and “Support to clinical/translational research”.
- As for the private funding initiatives for the development of data repositories and tools, these are reported in one country (17%, Bulgaria), and not supported in 3 countries (50%, Czech Republic, Estonia, Lithuania), while 2 countries answered “I don’t know” (Slovakia and 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”.
- FAIR data are supported only by initiatives other than the NP/NS for RD in one country (17%, Czech Republic). 3 countries (50%, Bulgaria, Lithuania, Romania) declare that FAIR data are not supported by the NP/NS for RD, nor by other initiatives, while 2 countries answered “I don’t know” (Estonia and Slovakia).
- The adoption of multidisciplinary holistic approaches for RD diagnostics and therapeutics are promoted by the NP/NS for RD and by other initiatives in 3 countries (50%, Bulgaria, Lithuania, Romania), not promoted at all in 2 countries (33%, Czech Republic, Estonia), and 1 country answers “I don’t know” (Slovakia).

Pillar 3 “Capacity building and empowerment”

- In 4 countries (67%, Bulgaria, Czech Republic, Lithuania, Romania), the NP/NS for RD promote and/or support training activities; in one country (17%, Estonia) it does not endorse this support, and one country answers (Slovakia) “I don’t know”.
- The training activities supported/promoted by the NP/NS for RD of the 4 above listed countries cover all the topics investigated through the dedicated question of the survey (see Table 5), except for trainings on FAIR data, that are not supported by the NP/NS for RD of any of the 16 countries having a NP/NS for RD.
- Trainings on “Standard and quality of genetics/genomics data in clinical practice and laboratories” are supported by the NP/NS for RD of EU-13 Countries only (namely Bulgaria, Czech Republic, Lithuania and Romania) and not by the other EU MS. Overall, the NP/NS of the EU-13 Countries address the training topics investigated through the survey (see Table 5), more frequently than the NP/NS for RD of the other EU MS.
- As to the endorsement of training activities by initiatives other than the NP/NS for RD, 4 countries (67 %, Bulgaria, Czech Republic, Lithuania, Slovakia) declare the presence of other initiatives and 2 countries answer “I don’t know” (Estonia and Romania).

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

- The NP/NS for RD promote the rapid translation of research results into clinical studies and healthcare in one country (17%, Romania), while in 4 countries (67%, Bulgaria, Czech Republic, Estonia, Lithuania), the NP/NS for RD do not back up this promotion, and 1 country answers “I don't know” (Slovakia).
- Initiatives (other than the NP/NS for RD) that promote the rapid translation of research results into clinical studies and healthcare are present in 1 country (17%, Bulgaria), not present in 3 countries (50%, Czech Republic, Estonia, Lithuania), while 2 countries answer “I don't know” (Romania, Slovakia).
- The development of innovative methodologies tailored for clinical trials are reported in the NP/NS for RD of one country (17%, Lithuania), but are not present in the NP/NS of 4 countries (67%, Bulgaria, Czech Republic, Estonia, Romania), while one country answers “I don't know” (Slovakia).

Final open question (to all the countries, including EU-13 Countries)

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 the survey can be considered as a first overview of the current situation and will serve as reference for further assessments and actions. Data obtained through this enquiry, although partial and not reflecting the complete situation in EU and former countries, can nevertheless give important insights of the general state of relevant national RD policies and of their alignment with the EJP RD actions. They can be considered as a first status quo and serve as hints to address the future actions to be promoted within the EJP RD, while an assessment of the impacts of the EJP RD activities on the national policies for RD might be the objective of a forthcoming analysis.

When considering the outcomes of the present document, it has to be taken into account that these reflect a partial situation, as a total of 21 countries (16 EU and 5 Associated/Other countries) participated to the survey. This fact is likely related, at least partially, to the burden faced by health institutions for the force-majeure situation of the COVID-19 pandemic, and partially to the difficulty in identifying target persons for the participation to the survey (or the inability of the person most involved in the NP/NS to respond to it). This latter statement might be confirmed by the fact, on one side, that 24% of the persons who answered to the survey are not directly involved in the NP/NS for RD of their country (so probably did not find anybody else to respond). The contribution of these persons to the survey has been included in the results of the present document, as their profiles refer in any case to the RD community (through universities, National Research Funds, Research Councils, National Institutes of Health, National Academies of Science) and their participation is considered as relevant.

Even if lacking information from several countries, the survey results nonetheless highlight numerous important points with regard to the alignment of the national policies, expressed mainly in terms of NP/NS for RD, with the activities promoted by the four non-transversal Pillars of the EJP RD. Below some details and issues are spotlighted and discussed for each one of the pillars and for the EU-13 Countries.

With things being so, the information gathered is important at least for three reasons: first, because for the responding countries an evaluation of the current situation has been done and is available for further assessments and developments; second, because this serves as reference for positive comparisons and actions in other countries; and third, because a special attention should be put on the non-responding countries, in order to figure out the reasons for the lack of response (especially if the reason is a lack of NP/NS or equivalent initiatives).

Apart from this, the issue that the person(s) more directly involved in the NP/NS in some countries could not be contacted, should be revised with each country in order to target them more directly for future developments.

General information

The general information gathered on the existence, date of approval, expiry and periodical evaluation of NP/NS can suggest some considerations.

Globally, there is a general good presence of NP/NS for RD in the responding countries, with 76% of the countries having adopted a NP/NS for RD at some stage, and 19% of the countries being in the process of developing a NP/NS for RD. Although, it should be considered that some NP/NS for RD already expired and have not been replaced by a new edition (Bulgaria, Estonia and Italy; for Ireland and The Netherlands it is not clear from the collected data whether the NP/NS for RD is under updating, or if it expired and is not in the process of being renewed).

A wider participation to the survey and the development or update of NP/NS for RD of the countries that currently do not have an approved/active NP/NS for RD are therefore important objectives to be envisaged. Besides this, the following specific considerations addressed to the alignment status with the four non transversal Pillars of the EJP RD can be stressed out:

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

The survey results show that in the NP/NS for RD of the responding countries there is a need to direct the efforts for an enhancement of national calls for research projects, as these are promoted only in 44% of the NP/NS. Another outcome is that in 13% of the NP/NS only transnational calls are promoted. Although it is recognized that RD research benefits from the broad collaboration at international level, also national research efforts should be considered by the NP/NS for RD.

Besides of the relatively positive result on the support of the NP/NS for RD for transnational calls for research projects (56% of the NP/NS for RD), it is of crucial importance to consider that in 38% of the NP/NS for RD neither national nor transnational calls for research projects are promoted, highlighting the research area as a critical issue that requires specific intervention. From the survey results, this appears to affect mainly the EU-13 Countries, 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 on the participation to transnational calls, 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.

Furthermore, it emerges that the support toward investments to share knowledge on RD is also an area that needs to be strengthened (endorsed only by 44% of the NP/NS).

Public funding initiatives other than the NP/NS for research and/or networking in the field of RD appear to be well present (81%), with the participation to national/international funded projects. This figure could appear inconsistent with the above-mentioned statement on the need to reinforce the promotion of national/transnational calls for research projects. This discrepancy could be explained by the fact that in many countries research on RD is probably addressed by general health 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 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).

Research activities, and especially national calls for research projects, together with the support of investments to share knowledge on RD, need to be fostered to achieve a better alignment with the activities of Pillar 1.

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

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.

The absence of a dedicated advisory body could mirror the already cited difficulty that emerged in identifying target persons to which forwarding the request to participate to the survey, and the fact that 24% of the responding persons were not directly involved in the NP/NS for RD. 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 better alignment 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 81% of the countries.

Apart from the support given by the NP/NS for RD, data repositories and tools in RD research receive a slightly greater support from private than from other public funding initiatives (31% vs 25%).

The data repositories and tools for RD research supported by the NP/NS for RD and by other public or private funding initiatives cover the topics mostly of “Registries catalogues”, “Support for clinical/translational research”, “Biobanks catalogues”, 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 from the countries’ initiatives.

A general good support to FAIR data is observed globally if considering the NP/NS for RD and/or other national RD initiatives (in 56% of the countries), with 31% of the countries enacting this support through the NP/NS for RD (with or without the addition of other initiatives). Alongside this partly encouraging outcome, it must be pointed out that in 25% of the responding countries the support is enforced only through initiatives other than the NP/NS, and in other 25% of the countries FAIR data are not supported at all (neither through the NP/NS for RD, nor through other initiatives), with EU-13 Countries figuring mainly in this latter option. 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.

The adoption of multidisciplinary holistic approaches for RD diagnostics and therapeutics emerges to receive globally a good endorsement, as only 12 % of the responding countries recognize not to promote such an adoption at all. This appears

as a favourable result, showing an overall good attention to the RD patients' multidisciplinary take in charge through the NP/NS and/or through other initiatives.

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 to enhance the alignment with the actions of Pillar 2 of the EJP RD.

Alignment with Pillar 3 "Capacity building and empowerment"

The attention dedicated to capacity building and empowerment reveals being widespread, with 75 % of the responding countries declaring to endorse training activities in their NP/NS for RD, and 75% 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", "Online education courses", "Data management", "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 not yet endorsed at all by the NP/NS for RD. 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.

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.

When focusing on EU-13 Countries, it can be observed that in these countries training activities are promoted at a certain degree, suggesting that these countries are engaging in efforts to move forward in the RD field.

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.

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

Even if globally there appears to be a relative positive alignment towards the promotion of a rapid translation of clinical studies and healthcare (56% 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 still low (37% of the responding countries) 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 37% of the NP/NS for RD and by 38 % of other initiatives.

Similarly, a high percentage of countries (56%) declares that the NP/NS for RD does not promote the development of innovative methodologies tailored for clinical trials in RD, or this information is not known (19%).

The above-described results could reflect the need to update the NP/NS for RD on these issues and refer both to areas that need strong 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 100% of the 7 responding EU-13 Countries adopted a Plan or Strategy at some stage. Of these, 4 countries have an active NP/NS for RD (Czech Republic, Romania, Slovakia, Lithuania) and 3 have NP/NS for RD that are expired/under update (Bulgaria, Estonia, Hungary).

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", "Lack of options for exploitation of research results at national level" and "Difficulties in accessing to national resources for funding of research and development of RD projects".

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" (indicated by all participating EU-13 Countries), "Lack of information on funding opportunities", "Quality of support provided by national contact points" and "Bureaucratic application on reporting 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 adoption of FAIR data that are not supported by the NP/NS for RD.
- The promotion of the rapid translation of research results into clinical studies and healthcare, that appears almost completely absent, as well as the development of innovative methodologies tailored for clinical trials.

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".

Other countries

A separate analysis for the 5 non-EU MS countries participating to the EJP RD has not been carried out, as only 2 of these have a NP/NS for RD and provided information on the alignment with the EJP RD activities (Serbia and UK), while the other 3 countries (Canada, Israel and Turkey) are developing a NP/NS for RD.

Conclusions and next actions

The results collected through this survey are the first overview of the current situation and will serve as starting point for further assessments and actions.

Comparing these results with the final considerations of D2.21, it can be reaffirmed that there is a **need to continue progressing in the evaluation of both the impact and the implementation status of the national policies for RD**. Hence, this analysis of the national state of play, and its alignment with the actions promoted by the EJP RD, can be considered as an important tool, first to identify the gaps (e.g., lack of promotion of calls at national level; need for clinical trials support) regarding a variety of areas that require to be filled and then to suggest the most important issues that require to be tackled to reach the desired advancements in RD field (e.g., engage with Policy Board (PB) in specific activities addressed to identify mechanisms for national calls or activities of promotion; disseminate the clinical office support at national level to increase engagement). These issues fall in the fields of: (i) the promotion of RD national and transnational calls for research projects, with a dedicated attention to the national level, and the support of investments to share knowledge (Pillar1); (ii) the constitution of dedicated RD advisory bodies for Research and Innovation and the attention to FAIR data; (iii) trainings on FAIR data, to be addressed especially by the NP/NS for RD and trainings on other topics that emerged to be less covered (mainly on Biobanks and Data Quality); and (iv) reinforce the promotion of both the rapid translation of research results in clinical studies and healthcare, and of the development of innovative methodologies tailored for clinical trials.

Besides this, the lining-up of the achievements gained in some countries that could be transferable to different contexts and situations could be of interest for the whole RD community.

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 quality of support provided by national contact points.

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 organization of the WP2 Strategic meeting in July 2021, as planned. The strategic meeting is focused on the presentation of the current state of the EJP RD advancements and on the national RD actions, as highlighted from the survey results that are the objective of the present deliverable. The meeting will also make a zoom on some country experience with regard to the best practices and bottlenecks and will draw some directions to advance in the RD field. The status quo, the highlights on country experiences and the consideration of the direction to move forward will stimulate the discussion on the way to enhance the alignment between the national policies for RD and the EJP RD activities, with the aim to enhance also the national capacities and the international cooperation.
2. For the Third Analysis of national state of play and alignment process with EJP RD, an updated version of the survey will be produced. The updated survey will be addressed both to the countries who participated in the present edition and to the non-participating countries (encouraging them to participate). The countries who already contributed, only the presence of eventual updates will be asked for and assessed, through specific closed questions.

The questions will be asked separately for the NP/NS for RD and for the other national initiatives; moreover, the countries who are not provided with a NP/NS for RD will be asked to fill in the survey, referring to the policies for RD that are present at national level.

Annex 1 Survey “National Plans and Strategies for Rare Diseases”

Items of the survey

Section 1

First name

Last name

Country

Institution

Section 2

NP/NS for RD in your country

Is there an approved National Plan/Strategy for rare diseases in your country?

- Yes
- Yes but not in force
- No
- No but it is under development in my country
- I don't know

If yes, please provide the link to the text of the National Plan/Strategy for rare diseases in your country

When was the National Plan/Strategy for rare diseases approved in your country? (specify year)

Is there a periodical evaluation of the National Plan/Strategy for rare diseases in your country?

- Yes
- No
- I don't have this information

By now, how many editions of National Plans/Strategies has your country adopted?

- One
- More than one
- I don't have this information

If the National Plan/Strategy for rare diseases of your country expired, please specify when it expired

Section 3

Involvement in the NP/NS for RD of your country

Are you directly involved in the development, implementation or review of the National Plan/Strategy for rare diseases in your country?

- Yes
- No

Are you directly involved in the construction of a National Plan/Strategy for rare diseases in your country?

- Yes
- No

If you are not directly involved in the National Plan/Strategy for rare diseases in your country, could you suggest a contact person who is directly involved?

- Yes
- No

Please enter the following details of the contact person directly involved in the National Plan/Strategy for rare diseases in your country

- First name
- Last name
- email address
- Phone number

Section 4

Pillar 1 National and International Investments on research in the field of rare diseases

Does the National Plan/Strategy for rare diseases in your country promote national calls for research projects on rare diseases?

- Yes
- No
- I do not know

Please specify how the National Plan/Strategy of your country promotes national calls for research projects and provide a link to any existing permanent dedicated website/webpage.

Does the National Plan/ Strategy for rare diseases in your country promote transnational calls for research projects?

- Yes
- No
- I do not know

Please specify how the National Plan/Strategy for rare diseases of your country promotes transnational calls for research projects and please, provide a link to any permanent dedicated website/webpage if it exists for calls.

Does the National Plan/ Strategy for rare diseases in your country foresee investments for networking to share knowledge on rare diseases?

- Yes
- No
- I do not know

Please specify how the National Plan/ Strategy for rare diseases foresees investments for networking to share knowledge on rare diseases

Are there public funding initiatives in your country for research and/or networking in the field of rare diseases?

- Yes
- No
- I do not know

Please describe the public funding initiatives in your country for research and/or networking in the field of rare diseases

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

- Yes
- No
- I do not know

Please describe the private funding initiatives in your country for research and/or networking in the field of rare diseases and provide a link to any permanent dedicated website/webpage if it exists.

Section 5

Pillar 2 Resources & Services to foster research on rare diseases

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

- Yes, an advisory body exists, but not specific for rare diseases
- Yes, an advisory body specific for rare diseases exists
- No
- I do not know

How does the National Plan/Strategy for rare diseases of your country foresee the support of data repositories and tools in research on rare diseases?

- The National Plan/Strategy for rare diseases does not foresee the support of data repositories and tools in rare diseases research
- Promoting the implementation of such data repositories and tools
- Promoting the development of new data repositories and tools
- Promoting both their implementation and development
- I do not know

If the National Plan/ Strategy for rare diseases of your country foresees the support of data repositories and tools in research on rare diseases, please specify on which topics (Please select. 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

Do the National Plan/ Strategy for rare diseases or other initiatives for rare diseases support FAIR* data in your country? (*FAIR: Findable, Accessible, Interoperable, Reusable)

- No National Plan/Strategy for rare diseases and no other initiatives support FAIR data
- Yes only the National Plan/Strategy for rare diseases
- Yes only other initiatives for rare diseases support FAIR data but not the National Plan/Strategy for rare diseases
- Yes, both (National Plan/Strategy for rare diseases and other initiatives for rare diseases)
- I do not know

Please describe how the National Plan/Strategy for rare diseases or other initiatives for rare diseases support FAIR data in your country

Is the adoption of multidisciplinary holistic approaches for rare diseases diagnostics and therapeutics promoted by the National Plan/ Strategy for rare diseases or by other initiatives for rare diseases of your country?

- Not by National Plan/Strategy for rare diseases and not by other initiatives for rare diseases
- Yes only by the National Plan/Strategy for rare diseases
- Yes only by other initiatives for rare diseases
- Yes by both, National Plan/Strategy for rare diseases and by other initiatives for rare diseases
- I do not know

Please describe how the adoption of multidisciplinary holistic approaches for rare diseases diagnostics and therapeutics is promoted by the National Plan/ Strategy for rare diseases or other initiatives for rare diseases of your country

Are there other public funding initiatives in your country than National Plan/Strategy for rare diseases, for the development of data repositories and tools to support research on rare diseases?

- Yes
- No
- I do not know

Which topics are covered by other public funding initiatives in your country, others than those foreseen in the National Plan/Strategy for rare diseases for the development of data repositories and tools to support research on rare diseases? (Please select. Possible multiple choice)

- Registries catalogue
- Biobanks catalogue
- Cell lines
- Animal models
- Semantic standards
- Support for clinical/translational research
- Access & privacy control
- Data deposition & analysis
- Tools
- Other

Are there other private funding initiatives in your country than National Plan/Strategy for rare diseases, for the development of data repositories and tools to support research on rare diseases?

- Yes
- No
- I do not know

Which topics are covered by other private funding initiatives in your country, others than those foreseen in the National Plan/Strategy for rare diseases for the development of data repositories and tools to support research on rare diseases? (Please select. Possible multiple choice)

- Registries catalogue
- Biobanks catalogue
- Cell lines
- Animal models
- Semantic standards
- Support for clinical/translational research
- Access & privacy control
- Data deposition & analysis
- Tools
- Other

Section 6

Pillar 3 Capacity building

Does the National Plan/Strategy for rare diseases promote and/or support training activities?

- Yes
- No
- I do not know

On which topics does the National Plan/Strategy for rare diseases promote and/or support training activities? (Please specify. Possible multiple choice)

- Data management
- Data quality
- FAIR data
- Standard and quality of genetics/genomics data in clinical practice and laboratories
- Registries
- Biobanks

- Empowerment of the patients
- Online education courses
- Other

Please describe more in detail how the National Plan/Strategy for rare diseases in your country promotes/supports training activities in the field of rare diseases and please, provide a link to any permanent dedicated website/webpage if it exists.

Apart from the provisions made in the National Plan/Strategy, are there other training, mentoring and coaching activities in the field of rare diseases provided in your country?

- Yes
- No
- I do not know

Please describe the other training, mentoring and coaching activities, apart from the provisions made in the National Plan/Strategy for rare diseases, provided in your country and please, provide a link to any permanent dedicated website/webpage if it exists.

Section 7

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

Does the National Plan/Strategy for rare diseases of your country promote a rapid translation of the research results in clinical studies and healthcare?

- Yes
- No
- I do not know

Please specify how the National Plan/Strategy for rare diseases of your country promotes a rapid translation of the research results in clinical studies and healthcare

Does the National Plan/Strategy for rare diseases of your country promote the development of innovative methodologies tailored for clinical trials in rare diseases?

- Yes
- No
- I do not know

Please specify how the National Plan/Strategy for rare diseases of your country promotes the development of innovative methodologies tailored for clinical trials in rare diseases

Are there other initiatives that promote a rapid translation of the research results in clinical studies and healthcare in the field of rare diseases, other than the National Plan/Strategy for rare diseases in your country?

- Yes
- No
- I do not know

Please specify what other initiatives in the field of rare diseases promote a rapid translation of the research results in clinical studies and healthcare, other than the National Plan/Strategy for rare diseases in your country

Section 8

Additional information

Do you belong to an EU-13 Country?

- Yes
- No

To which EU-13 Country do you belong?

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

- Language
- Funding
- Difficulties in accessing to national resources for funding research and development of RD projects
- Lack of options for exploitation of research results at national level
- Other

Describe more in detail the main obstacles and barriers in your country for the development, improvement and translation of RD research results, based on your experience

Regarding your participation in EU/International projects in the rare diseases field, what do you estimate are the most important obstacles/barriers? (Please select. Possible multiple choice)

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

Please describe more in details what you estimate as the most important obstacles/barriers for the participation of your country in EU/International projects in the rare diseases field

Section 9

Do you want to highlight any other aspect regarding the National Plan/Strategy for rare diseases of your country that was not included in the present survey?

- Yes
- No

If yes please specify

Annex 2 Results for the EU non-responding countries, elaborated from other sources than the survey “National Plans and Strategies for Rare Diseases”

From the analysis of the reports of EUROPLAN, RD-Action, and of the NP/NS for RD that were available through the internet, the following results can be assumed for the countries that did not take part in the survey. It is expected to have more precise and updated results from the interested countries with the next edition of the survey.

Pillar 1 “National and international investments on research in the RD field”-Alignment status with the NP/NS for RD

Country	Promotion of national calls for research projects	Promotion of transnational calls for research projects	Promotion of investments to share knowledge	Promotion of public/private initiatives for research/networking
Austria	✓	✓	✓	Yes, public
Belgium			✓	Yes, public
Denmark			✓	Yes, public
Finland	✓	✓		Yes, public
Greece	✓	✓	✓	
Croatia	✓	✓	✓	
Cyprus	✓	✓	✓	Yes, public and private
Latvia	✓	✓		
Malta	-	-	-	-
Poland	✓	✓	✓	Yes, public
Slovenia	✓		✓	

Pillar 2 “Resources and services to foster solutions in the field of RD” -Alignment status of the NP/NS for RD

Country	Support of data repositories and tools for RD research projects	Support of FAIR data	Support of multidisciplinary holistic approaches
Austria	✓	✓	✓
Belgium	✓		✓
Denmark	✓		✓
Finland			✓
Greece	✓	✓	✓
Croatia			✓
Cyprus	✓		✓
Latvia			✓
Malta	-	-	-
Poland			✓
Slovenia			

Pillar 3” Capacity building and Empowerment” -Alignment Status of the NP/NS for RD

Country	Support of training activities	The majority of the NP/NS for RD seem to support training activities on the empowerment of patients
Austria	✓	
Belgium	✓	
Denmark	✓	
Finland	✓	
Greece	✓	
Croatia	✓	
Cyprus	✓	
Latvia	✓	
Malta	-	
Poland	✓	
Slovenia	✓	

Pillar 4 “Accelerated translation of research projects and improvement of outcomes of clinical studies”-Alignment status with NP/NS for RD

Country	Promotion of rapid translation of research results in clinical studies and healthcare	Promotion of development of innovative methodologies tailored for clinical trials
Austria	✓	
Belgium		
Denmark	✓	
Finland		
Greece	✓	
Croatia	✓	
Cyprus	✓	
Latvia		
Malta	-	-
Poland	✓	
Slovenia		

Form the analysis of the alignment process carried out for the non-responding EU countries based on the results of RD-Action, the following considerations can be proposed:

Pillar 1 “National and international investments on research in the RD field”

- Regarding the promotion of national calls for research projects, these seem to be promoted by the NP/NS for RD of 80% (8/10) of the countries, while 70% (7/10) of the NP/NS for RD seem to support transnational calls.
- The promotion of investments to share knowledge appears to be supported by 70% (8/10) of the countries.
- Public funding initiatives for research/networking appear to be endorsed by the NP/NS for RD of 70% (7/10) of the countries, whereas private funding initiatives only by 20% (2/10).

Pillar 2 “Resources and services to foster solutions in the field of RD”

- Data repositories and tools for RD research projects seem to be supported by the NP/NS for RD of 50% (5/10) of the countries.
- FAIR data appear to be supported by the NP/NS for RD of 20% (2/10) of the countries.
- Multidisciplinary holistic approaches seem to be supported by the NP/NS of 90% (9/10) of the countries.

Pillar 3 “Capacity building and empowerment”

- The support to training activities is endorsed by 100% of the NP/NS for RD with a great presence of trainings for the empowerment of the patients.

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

- 60% of the NP/NS for RD appear to promote a rapid translation of research results in clinical studies and healthcare, while the development of innovative methodologies tailored for clinical trials seem not to be endorsed.

These results show in general better and more optimistic outcomes for nearly all the investigated areas, but it must be considered that this assessment has been carried out also on non-official English versions of the NP/NS for RD and that the advice and collaboration of persons directly involved in the national policies for RD is essential for a more accurate and reliable insight of the drawn national initiatives for RD.

These data should be considered taking into account the percentage of responding countries and will be updated and revised with the outcomes of the next edition on the survey.

Annex 3 Links to the NP/NS for RD

Bulgaria:

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

Czech Republic:

Since the EU Council recommendation on an action in the field of rare diseases Czechia has 1) National Action Strategy (2010-2020; now we are preparing a 2nd National Action Strategy for the upcoming decade 2021-2030) and 3 Action plans

see also from Europlan project -

<http://www.europlanproject.eu/NationalPlans?country=CZ>

Links (some in Czech, with English translations)

http://www.mzcr.cz/dokumenty/narodni-akcni-plan-pro-vzacna-onemocneni-na-leta-2012-2014_6713_1.html

<https://www.databaze-strategie.cz/cz/mzd/strategie/narodni-akcni-plan-pro-vzacna-onemocneni-na-leta-2015-2017>

https://www.mzcr.cz/dokumenty/vlada-schvalila-zpravu-o-plneni-narodni-strategie-pro-vzacna-onemocneni_15601_1.html

<https://www.databaze-strategie.cz/cz/mzd/strategie/narodni-akcni-plan-pro-vzacna-onemocneni-na-leta-2018-2020>

<https://www.databaze-strategie.cz/cz/mzd/strategie/narodni-strategie-pro-vzacna-onemocneni-na-leta-2010-2020>

Estonia:

http://download2.eurordis.org/rdpolicy/National%20Plans/Estonia/Estonia_RD%20National%20Development%20Plan_2014_Estonian.pdf

France:

https://solidarites-sante.gouv.fr/IMG/pdf/plan_national_maladies_rares_2018-2022.pdf

<http://www.bndmr.fr/wp-content/uploads/2018/10/7-ODENT-PNMR3-SEMINAIRE-BNDMR-3-OCT-18.pdf>

Germany: www.namse.de

Hungary:

http://download2.eurordis.org/rdpolicy/National%20Plans/Hungary/2.Hungary_RD%20National%20Plan_2013-2020_English.pdf

Ireland: <https://assets.gov.ie/37342/da70fc6fadd24425b98311e679f4406b.pdf>

Italy: http://www.salute.gov.it/imgs/C_17_publicazioni_2153_allegato.pdf

Lithuania:

https://sam.lrv.lt/uploads/sam/documents/files/Veiklos_sritys/Asmens_sveikatos_prie_ziura/Retos%20ligos/Retu%20lig%C5%B3%20planas.pdf

Luxembourg:

<https://sante.public.lu/fr/politique-sante/plans-action/maladies-rares/index.html>

Romania:

<http://www.bolirareromania.ro/sites/default/files/politici/PNBR%202014-2020.pdf>

Serbia:

<https://www.zdravlje.gov.rs/tekst/343045/program-za-retke-bolesti-i-akcioni-plan.php>

Slovakia: <https://www.health.gov.sk/?narodna-strategia>

Spain:

https://www.mscbs.gob.es/organizacion/sns/planCalidadSNS/pdf/Estrategia_Enfermedades_Raras_SNS_2014.pdf

Portugal:

<https://www.dgs.pt/saude-a-a-z.aspx?v=%3d%3dBAAAAB%2bLCAAAAAABABLszU0AwArk10aBAAAAA%3d%3d#saude-de-a-a-z/doencas-raras>

The Netherlands:

https://www.zonmw.nl/fileadmin/zonmw/documenten/Kwaliteit_van_zorg/NPZZ/NPZeldzame_Ziekten.pdf

UK: <https://www.gov.uk/government/publications/rare-diseases-strategy>