EJP RD
European Joint Programme on Rare Diseases

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

Grant agreement number 825575

Del 2.21
First Analysis of national state of play and alignment process with EJP RD

Organisation name of lead beneficiary for this deliverable:
Partner N°26 – ISS

Due date of deliverable: month 9

Dissemination level:
Public

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1. Introduction and Objective

As defined in Task 2.5 "Translation/impact of prioritization on national and EU strategies" (M6-60) of the EJP-RD Project, the European dimension of the EJP RD and the involvement of the Policy Board is expected to have impact in two ways: on one side, on the activities of the EJP RD itself, and on the other side, on the undertakings at national and EU level.

The main objective foreseen for the present deliverable is to verify the alignment of the national plans with the actions promoted by the EJP RD at European level.

This falls under the action of WP2 "Integrative research and innovation strategy" (M1-60), that is dedicated to the development of EJP RD research and innovation strategy in connection to all related stakeholders.

Specific objectives of the whole WP2 are:

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

In addition to verifying the alignment of the national policies and national plans with the actions promoted by the EJP RD at European level it is foreseen that 2.5 Task Leaders (TL) are responsible for collection of information from EU Member States (MS) via a survey targeting national mirror groups on the EJP RD relevant/complementary actions performed at national level. Specific focus regards EU-13 countries in respect to their specific needs, obstacles and advancements.

The report analyzing national alignment with EJP RD will be produced annually and presented at the occasion of the Policy Board and ExeCom meetings.

The deliverables already presented for WP 2 at M9 are:

D2.1 Final list of prioritization criteria (Public Report)
D2.2 Prioritization scheme including decision-making process (Public Report)
D2.3 First Summary document on mapped research and innovation needs (Confidential Report)
D2.8 First Scoping paper (Public Report)
D2.13 First List of refined JTC topics (Confidential Report)
D2.16 First List of research and innovation needs requiring medium-or long-term approach and related Task Forces (Public Report).

As defined above, Task 2.5 includes between its objectives the promotion of Task 2.2 Scoping paper to the national mirror groups and within the current and forthcoming EC Framework programme.
It has indeed to be pointed out, that the D2.8 First Scoping Paper is on stand-by and based on the related documents available at the date of July 10, and that the national mirror groups are still under construction.

1.1. Methodology

To elaborate the present deliverable "First Analysis of national state of play and alignment process with EJP RD", after deep assessment and discussion among WP2 leaders on the most useful sources of information regarding the current or most recent national state of play, a review of previous documents published by RD-ACTION, IRDiRC and EUROPLAN and specific publications was carried out.
The present document will focus specifically on an overview regarding the documents that address national plans and strategies in the field of RDs in Member States (MS) of EU, while the broad range of relevant aspects analyzed in the cited documents is publicly consultable at the links listed below.
The analyzed documents are:

- "Overview report on the State of the Art of Rare Disease Activities in Europe" RD-ACTION, Version 2018 (http://www.rd-action.eu/wp-content/uploads/2018/09/Final-Overview-Report-State-of-the-Art-2018-version.pdf)
- "Overview report on the State of the Art of Rare Disease Activities in Europe" RD-ACTION, Version 2016 (http://www.rd-action.eu/wp-content/uploads/2015/10/Overview-Report-State-of-the-Art-2016-Final-for-Portal.pdf)
- International Rare Diseases Research Consortium "State of Play in the Field of RDs 2015-2018" (http://www.irdirc.org/wp-content/uploads/2019/09/IRDiRC_State-of-Play-2018_Final.pdf.)
- EUROPLAN Project (http://www.europlanproject.eu/Content?folder=1).
- "Policies and actions to tackle rare diseases at European level" (Montserrat A, Taruscio D., 2019) (https://www.ncbi.nlm.nih.gov/pubmed/31553326)
- "Exploring the usability of EUCERD core indicators for rare diseases" (Ferrelli RM et al, 2015) (https://www.ncbi.nlm.nih.gov/pubmed/26783222)

- "EUROPLAN: a project to support the development of national plans in Europe" (Taruscio et. al, 2013) (https://www.ncbi.nlm.nih.gov/pubmed/24503588)

- "National plans and strategies on rare diseases in Europe" (Taruscio et al, 2010) (https://www.ncbi.nlm.nih.gov/pubmed/20824461)

The results of this overview of documents are taken into account to establish the state of the art on national plans and strategies in the field of RD in MS of EU, to verify the alignment with EJP RD actions and to identify further actions to be implemented, if it is the case.

1.2. Results

1.2.1. Political framework of the state of the art of RD activities at European level

Since many years, RD have been a policy priority at both European Union (EU) and MS level. Various countries have engaged in activities and initiatives to reach legislative texts, regulations and national strategies, plans and recommendations concerning RD (in all their facets) and orphan drugs.
EU policy about RD was defined through several initiatives (see Annex 1), including the Commission Communication and Council Recommendation, in November 2008, changing significantly the scenario in this field. (https://ec.europa.eu/health/ph_threats/non_com/docs/rare_com_en.pdf)
Many progresses had been achieved by the RD community, at the EU and national levels but still a number of challenges and opportunities have to be faced to continue progressing through the advance in diagnosis, treatment and care for people with RD in Europe.
More precisely, these points can be highlighted:

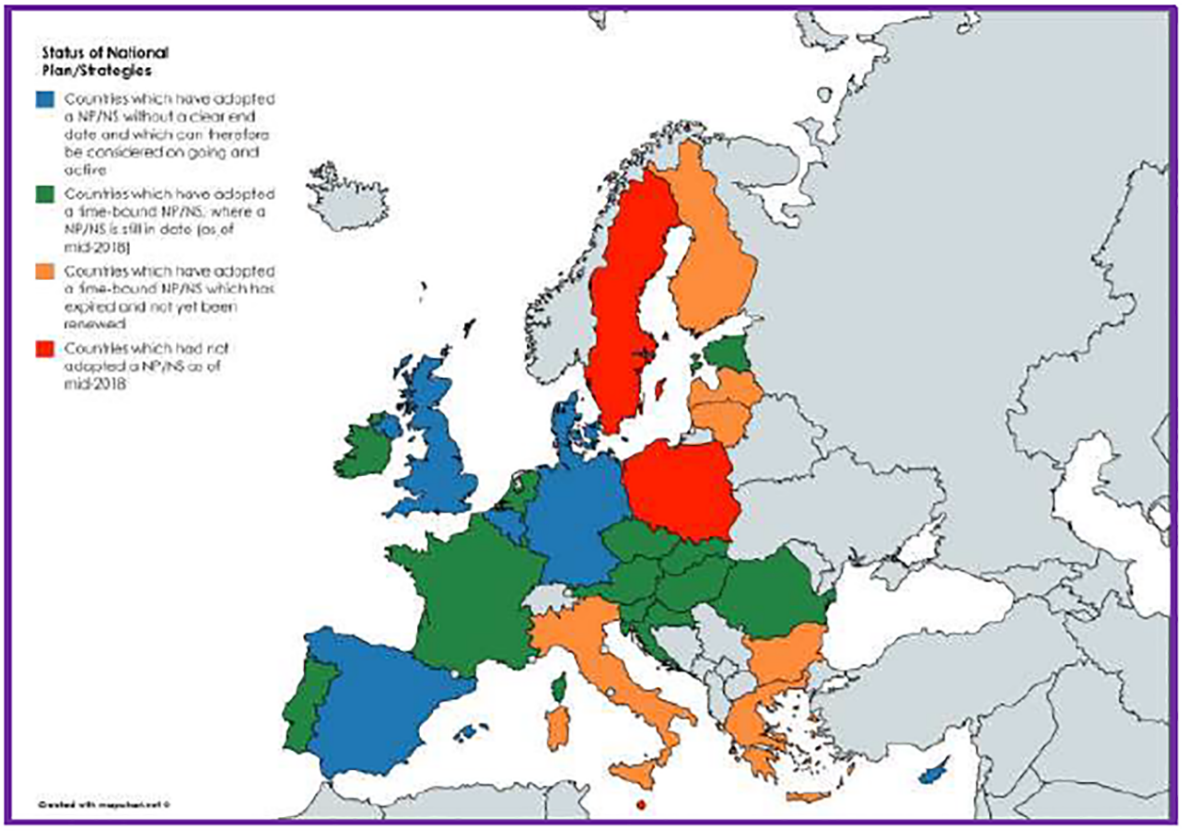
- 2016 saw the approval and official "birth" of the long-awaited European Reference Networks (ERNs) for RD. In 2017, 24 ERNs have been lunched and now are operational.
- In ERNs collaboration between the "healthcare" and "research" domains is increasing, and will continue to grow.
- Patients'organisations, such as EURORDIS, continue to grow and play leading roles in initiatives such as Joint Actions, Tenders and Projects driving forward progress (the EJP RD included).
- EURORDIS and Orphanet celebrated 20 years of activities in 2017.

1.2.2. National Policies and National Plans at European Member State level

By the time of the delivery of RD-ACTION Overview Report of 2018, the progresses made towards the goal to create national plans (NP) or strategies (NS) for RD were:

- 25 countries adopted a NP/NS for RDs at some stage
- 19 of these countries adopted NP/NS which were time-bound (i.e. they were approved covering certain years of activity)

- The following 13 countries have time-bound NP/NS which were still apparently active in July 2018: Austria, Croatia, Czech Republic, Estonia, France, Hungary, Ireland, Luxembourg, Netherlands, Portugal, Romania, Slovak Republic, Slovenia.
- The following 6 countries adopted time-bound NP/NS which had expired by July of 2018 and seemed not to have been replaced/renewed: Bulgaria, Finland, Greece, Italy, Latvia, Lithuania
- The following 6 countries adopted NP/NS which appear to be "ongoing" (i.e. according to the 2016 SoA data received, do not cover specific time periods): Belgium, Cyprus, Denmark, Germany, Spain, UK
- Three EU MS that appear not to have adopted a NP/NS by the end of 2016: Poland, Malta and Sweden.
- By 2018 also Switzerland and Norway had a RD plan or strategy.



From: "Overview report on the State of the Art of Rare Disease Activities in Europe" RD-ACTION, Version 2018 (http://www.rd-action.eu/wp-content/uploads/2018/09/Final-Overview-Report-State-of-the-Art-2018-version.pdf)

In July 2018, France became the first country to adopt a Third National Plan for RDs, becoming an important example to other European countries, as many initial NP/NS are currently either out of date or near to expiration (often with limited evidence of realistic impact and concrete levels of implementation).

The Overview Report 2018 of RD ACTION referred that the NP/NS of 6 countries were technically no longer "active" and some of the 13 of the time bound NP/NS were approaching the end by 2020.

Few months ago the Italian Ministry of Health started the elaboration of the second national plan (2019-)

The renewal of the focus on National Plans and Strategies for Rare Diseases in Europe is therefore strongly stressed out as an imperative priority at this time.

As already mentioned, there is a great heterogeneity in the state of advancement of national policies, plans or strategies for RDs.

The needs are:

- to evaluate the extent to which existing NP/NS have actually been implemented in European countries
- to encourage to adopt their second and third NP/NS to maintain the much-needed national focus and momentum on RD
- to define key objectives and content for this next generation of NP/NS, by identifying good practices which have yielded results in particular countries/regions, assessing their transferability to other countries/situations, and agreeing new issues and topics which should be addressed via robust Plans and Strategies for the coming years.

In conclusion, as stated in the Grant Agreement, "rare diseases are a prime example of a research area that can strongly profit from coordination on a European and international scale. RD research should be improved to overcome fragmentation, leading to efficacious use of data and resources, faster scientific progress and competitiveness, and most importantly decrease unnecessary hardship and prolonged suffering of RD patients".
This was almost fixed by the Council Recommendation of June 8, 2009 on an action in the field of RD (2009/C 151/02), which specifically emphasized that the coordination of Community, national and regional programmes for RD research should be improved. The overarching ambition of the EJP RD is to establish an urgently needed comprehensive strategy covering research, data, tools and clinics leading to optimization and exploitation of results, higher and timely diagnostic rates, faster drug discovery at reduced costs, improved patients' care, empowering all stakeholders as well as giving Europe a leading role in the field of RD in the coming years.

As an expected impact EJP RD foresees to contribute to improve the alignment of national/regional activities and policies in RD. The strength of the EJP RD consortium lies not only in the number and diversity of its partners but also in its capacity to exploit the existing elements, detect their weaknesses, improve, innovate and deliver novel more efficient solutions. To efficiently transcribe EJP RD activities and outcomes at regional, national, EU and international levels the EJP RD already involved and will

maintain strong connection with relevant policy makers. The governance, research and innovation strategy and prioritization process is almost organized around the Policy Board, with ministries of research and health, relevant EC directorates and other key high-level stakeholders. In addition, an urgent and on-going action is to put in place National Mirror Groups (NMG) bridging national partners participating directly in the EJP RD and additional actors of the RD field. NMG are in fact key actors for identifying, discussing and bringing the national needs to the upper level. It is expected that NMG will include representatives of each National Plan for RD, national nodes of the ERNs, relevant national authorities and research institutions (whether participating to the EJP RD or not), as well as relevant national partners of the EJP RD and Governing Board (GB) member that will report NMG's views and positions during GB's meetings.
With specific regard to Task 2.5, the NMG will be in charge of promoting the Task 2.2 Scoping Paper.

1.3. Next actions

As a result of the "First Analysis of national state of play and alignment process with EPJ RD", it emerged that the next actions that need to be implemented to reach the achievements of Task 2.5 are:

- The final constitution of National Mirror Groups (NMG) in most of the involved countries.
- Collect information from EU Member States on the EJP RD relevant/complementary actions, via a survey targeting NMG. For this purpose dedicated questionnaire with be developed. The survey will have a special focus on EU-13 Countries in respect to their specific needs, obstacles and advancements.
- After collection of this data, the alignment with EJP RD will be analyzed through a report, to be updated annually and presented at the occasion of the Policy Board and ExecCom meetings.

2. ANNEX 1

Key RD Policy Documents, Legislation and Regulation

The key RD Policy Documents and Legislation encompass a broad range of aspects regarding the field of RDs. In the present document, they will be mentioned and/or summarized pointing out the issues specifically regarding national plans and strategies, while the complete documents are consultable at the dedicated links. Here, a brief summary on them:

2.1 Regulation on Orphan Medicinal Products

"Regulation (EC) No 141/2000 of the European Parliament and of the Council of 16 December 1999 on Orphan Medicinal Products", that by defining "Orphan Medical Products" defined also what constitutes a RD in the European framework, and remained as base for all further developments.

2.2 Commission Communication on Rare Diseases: Europe's challenges [COM82008) 679 final] and the "Council Recommendation of 8 June 2009 on an action in the field of rare diseases (2009/C 151/02)

These key "foundation documents" (although "soft law" documents), influenced and guided the RD policy sphere at European level. European and Member States activities received a great push towards unification of efforts by these "key policy documents", that helped moreover to shape the work of bodies such as the RD Joint Actions and the Expert Groups/Committees. Recommendations were made to the MS around 7 distinct though inter-related topics:

- Plans and strategies in the field of RD
- Adequate definition, codification and inventorying of RD
- Research on RD
- Centres of expertise and ERNs
- Gathering the expertise on RD at European Level
- Empowerment of patients organisations
- Sustainability

In 2014, the European Commission published an Implementation report (https://ec.europa.eu/health//sites/health/files/rare_diseases/docs/2014_rarediseases_implementationreport_en.pdf) on both the Council Recommendation and the Commission Communication, addressed to the European Parliament, the Council, the European Economic and Social Committee and the Committee of the Regions. The report concludes that overall, the objectives of the Communication and the Council Recommendation have been reached and that both served to strengthen the cooperation between the European Union, the Member States and the relevant stakeholders.

The Implementation Report highlighted various actions to continue supporting Member States, such as:

- Maintain the EU's coordinative role in the development of the EU policy on RD and to support Member States in their activities on the national level.
- Continue to support the development of high quality National Rare Diseases Plans/Strategies in the European Union.
- Provide continued support for the International Rare Disease Research Consortium (IRDiRC) and initiatives developed under its umbrella.
- Continue to ensure proper codification of RD.
- Work further to decrease inequalities between patients with RD and patients suffering from more common disorders and to support initiatives promoting equal access to diagnosis and treatment.
- Continue to promote patients empowerment in all aspects of rare disease policy development.
- Make use of the Directive 2011/24/EU on the application of patient's rights in cross-border healthcare to bring together European Reference Networks on RD, support the development of the tools facilitating cooperation and interoperability of the ERNs for RD.
- Implement and continue support for the European Platform on RD registration.
- Continue playing a global role in the RD initiative and collaborating with important international stakeholders.

2.3 The Cross-Border Healthcare (Directive 2011/24/EU) http://eur-lex.europa.eu/legal-content/EN/ALL/?uri=celex%3A32011L0024,the Directive on the Application of Patient's Rights in Cross-Border Healthcare has played a key role in the development of ERNs. The impact of the Directive is monitored through a series of reports:

https://ec.europa.eu/health/sites/health/files/cross_border_care/docs/2016_msdata_en.pdf);

https://ec.europa.eu/health/sites/health/files/cross_border_care/docs/2015_operation_report_dir201124eu_en.pdf

Former European Expert Groups/Committees for RDs

3.1 The Commission Expert Group on rare diseases (CEGRD 2014-2016)

A Commission Expert Group on rare diseases (CEGRD) was formed in 2013, chaired by the European Commission. The main stakeholders groups represented were:

- Member States' competent authorities.
- Patient organisations in the field of RD.
- European associations of producers or service providers relevant for patients affected by RD.
- European professional associations or scientific societies acting in the field of RD.
- Individuals appointed in a personal capacity as experts having public health or scientific expertise at EU level in the field of RD.

The CEGRD adopted several sets of Recommendations between its first meeting on 11-12 February 2014 and its final meeting on 28-29 November 2016:

- The Recommendation on ways to improve codification for rare diseases in health information systems 7http://ec.europa.eu/health//sites/health/files/rare_diseases/docs/recommendation_coding_cegrd_en.pdf
- Rare Disease European Reference Networks: An Addendum to the EUCERD Recommendations of January 2013 https://ec.europa.eu/health//sites/health/files/rare_diseases/docs/20150610_erns_eucerdaddendum_en.pdf.
- Recommendations on cross-border genetic testing of rare diseases in the European Union (2015).https://ec.europa.eu/health//sites/health/files/rare_diseases/docs/2015_recommendation_crossbordergenetictesting_en.pdf
- Recommendations to support the incorporation of rare diseases into social services (2016) https://ec.europa.eu/health//sites/health/files/rare_diseases/docs/recommendations_socialservices_policies_en.pdf.

The meetings covered a broad range of topics of relevance to the rare disease sphere, delivered by different Directorates General such as: DG RTD (The Directorate-General for Research and Innovation (***DG RTD***)that presented IRDiRChttp://www.irdirc.org/ , RD-Connect http://rd-connect.eu/ , H2020 funding streams, JRC (Joint Research Centre) for the progress relating to rare disease

registration, DG EMPL (Directorate-General for Employment, Social Affairs and Inclusion) for EU Policy on social services.
Additional topics of the meetings were: Structural Funds, reimbursement of orphan medical products (OMPs), pharmaceutical legislation, Medicine Adaptives Pathways to Patients (MAPPs) and rare cancers.
At the final meeting, it was announced that discussions had to take place internally in the European Commission, regarding the future of Expert Groups on RD in general.
The mandate of CERGRD expired and at the time of early January 2017 there were no updates.

3.2 European Union Committee of Experts on Rare Diseases (EUCERD, 2010-2013)
Over its three years of duration, the European Union Commission of Experts on Rare Diseases (EUCERD, 2010-2013) was very successful and prolific in assisting the European Commission to prepare and implement community activities in the rare disease field. With its multi-stakeholders composition, it functioned as an effective platform to discuss and debate key topics and concerns for the whole rare disease community. It garnered the cooperation of Member States representatives, European Commission, patients, experts and representatives from industries and was supported by a dedicated Joint Action that generated a set of recommendations to be further discussed and elaborated by EUCERD. Key topics were:

- Centres of Expertise
- ERNs
- Patient registration and data collection
- Access to Orphan Medicinal products for RD
- RD National Plans /Strategies
- Newborn screening
- Codification of RD
- Cross-border genetic testing

A point that is strictly relevant to the present Deliverable is the adoption by EUCERD of 5 sets of Recommendations over its three years of activity, on the topics of indicators for national plans/strategies, registries, ERNs, centres of expertise and improving informed decisions based on the clinical added value of orphan medical products.
The 5 Recommendations were:

- EUCERD Recommendations on Core Indicators for Rare Disease National Plans/Strategies (2013) http://www.eucerd.eu/wp-content/uploads/2013/06/EUCERD_Recommendations_Indicators_adopted.pdf
- EUCERD CORE Recommendations on Rare Disease Patient Registries (2013) http://www.eucerd.eu/wp-content/uploads/2013/06/EUCERD_Recommendations_RDRegistryDataCollection_adopted.pdf
- EUCERD Recommendations on European Reference Networks for Rare Diseases (2013) http://www.eucerd.eu/?post_type=document&p=2207
- EUCERD Recommendations on the Clinical Added Value of Orphan Medical Products (CAVOMP) Information Flow (2012) http://www.eucerd.eu/?post_type=document&p=1446

- EUCERD Recommendations on Quality Criteria for Centres of Expertise for Rare Diseases in Member States (2011) http://www.eucerd.eu/?post_type=document&p=1224

As reported in the Council Recommendation of 8 June 2009 on an action in the field of rare diseases (2009/C 151/02)- "Member States elaborate and adopt a plan or strategy as soon as possible, preferably by the end of 2013 at the latest, aimed at guiding and structuring relevant actions in the field of rare diseases within the framework of their health and social systems".

In order to promote this process, the three-year project "EUROPLAN" (European Project for Rare Disease National Plans Development, 2008-2010), coordinated by the Italian National Centre for Rare Diseases, IstitutoSuperiore di Sanità, had the aim to develop tools and carry out activities in order to help EU countries to establish and implement national plans or strategies in the field of rare diseases. One of the tools that has been created is the "Report on indicators for monitoring the implementation and evaluating the impact of National Plans or Strategies for rare diseases"(http://www.europlanproject.eu/Documentation?idDocumentationType=2&idDocumentationTypeChild=3).
A set of initial 59 indicators, although all considered as relevant to monitor the implementation of National Plans and Strategies, were further listed in "Core Indicators", organised as process and outcome indicators based on usefulness for patients and carers, feasibility and political usefulness (EUROPLAN I).
Target groups for these Recommendations are the EU Member States and the European Commission. Their dissemination is also addressed to other targets, such as EC initiatives (projects and joint actions, Cross-Border Healthcare Expert Group, EUnetHTA, EPAAC), Centres of Expertise in the field of RD, healthcare providers, RD experts and RD network co-ordinators and partners, and patient organizations.
The Core Indicators for Rare Disease National Plans and Strategies (hereafter Core Indicators) are instrumental for the decision-making process related to the adoption, assessment and further development of public policies for RD.
EU Member States should use these Core Indicators to collect data on an annual basis. The data collected shall be used, according to the document:

- By the Member States as a supporting instrument to follow the policy initiatives integrated in their National Plans and Strategies for RD (adopted in response to the Council Recommendations of 8 June 2009), besides other additional indicators eventually adopted by Member States
- For the annual reporting at Member States Level to elaborate the "Report on the State of the Art on Rare Disease Activities in Europe of the European Union Committee of Experts on RD", published yearly by EUCERD.

The following list of the Core Indicators is proposed to all EU Member States so as to monitor their National Plans or Strategies on Rare Diseases.
For a detailed description of the here listed Core Indicators see http://www.eucerd.eu/wp-content/uploads/2013/06/EUCERD_Recommendations_Indicators_adopted.pdf

List of Core Indicators:

- Background Indicators (Preparation of the Plan/Strategy)
  1. Existence of regulations/laws, or equivalent official national decisions that support the establishment and development of a Rare Diseases (RD) plan
  2. Existence of a RD advisory committee
  3. Permanent and official patient's representation in plan development, monitoring and assessment
  4. Adoption of the EU RD definition

- Content Indicators

  Centres of Expertise
  5. Existence of a national policy for establishing Centres of Expertise on RD
  6. Number of national and regional Centres of Expertise adhering to the national policy
  7. Participation of national or regional Centres of Expertise in ERNs

  Information
  8. NP/NS support to the development of/participation in a comprehensive national and/or regional RD information system
  9. Existence of Help lines for RD

  Knowledge, classification/coding, registries and research
  10. Existence of a national policy on rare disease clinical guideline development and implementation
  11. Type of classification/coding used by the healthcare system
  12. Existence of a national policy on registries or data collection on RD
  13. Existence of RD research programmes and/or projects in the country
  14. Participation in European and international research initiatives

  Therapies
  15. Number of Orphan Medical Products (OMPs) with a European Union marketing authorization and available in the country (i.e priced reimbursed or directly supplied by the national health system)
  16. Existence of a governmental system for compassionate use of medicinal products

  Social Services
  17. Existence of programmes to support the integration of RD patients in their daily life

- **Financial Support Indicators**
  18. Existence of a policy/decision to ensure long-term sustainability of the RD plan/strategy
  19. Amount of public funds allocated to the RD plan/strategy
  20. Specific public funds allocated for RD research
  21. Public funds specifically allocated for RD actions/projects per year since the plan started

More details are available in the Summary of EUCERD achievements, in: http://ojrd.biomedcentral.com/articles/10.1186/1750-1172-9-30

The Steering Group on Health Promotion, Disease Prevention and Management of Non-Communicable Diseases (SGPP)

The SGPP is chaired by the EU Commission and is composed entirely of MS' representatives. Its main goal is to facilitate the implementation of evidence-based best practices by EU countries, in order to ensure that the most up-to-date research findings and knowledge are put into practice.

https://ec.europa.eu/health/sites/health/files/major_chronic_diseases/docs/c2018_4492_en.pdf

Consensus European Recommendations on RD Issues

This is a summary list of European recommendations issued and adopted by consensus (details on the date of adoption and accessing link are also provided in the table):

Recommendation Title	Date Adopted	Link to Document
Quality Criteria for Centres of Expertise for Rare Diseases	October 2011	http://www.eucerd.eu/?post_type=document&p=1224
Improving Decisions based onthe Clinical Added Value ofOrphan Medicinal Products(CAVOMP) Information Flow	September 2012	http://www.eucerd.eu/?post_type=document&p=1446
European Reference Networksfor Rare Diseases **Addendum to EUCERD** **Recommendations on Rare** **Disease ERNs**	January 2013 June 2015	http://www.eucerd.eu/?post_type=document&p=2207 https://ec.europa.eu/health//sites/health/files/rare_diseases/docs/20150610_erns_eucerdaddendum_en.pdf

Rare Disease Patient Registration and Data Collection	June 2013	http://www.eucerd.eu/wp-content/uploads/2013/06/EUCERD_Recommendations_RDRegistryDataCollection_adopted.pdf
Core Indicators for Rare DiseaseNational Plans/Strategies	June 2013	http://www.eucerd.eu/wp-content/uploads/2013/06/EUCERD_Recommendations_Indicators_adopted.pdf
Potential Areas of EuropeanCollaboration in the field of NewBorn Screening (Opinion)	July 2013	http://www.eucerd.eu/wp-content/uploads/2013/07/EUCERD_NBS_Opinion_Adopted.pdf
Ways to Improve Codification ofRare Diseases	November 2014	https://ec.europa.eu/health//sites/health/files/rare_diseases/docs/recommendation_coding_cegrd_en.pdf
Cross-Border Genetic Testing ofRare Diseases in the EU	November 2015	https://ec.europa.eu/health//sites/health/files/rare_diseases/docs/2015_recommendation_crossbordergenetictesting_en.pdf
Support the Incorporation ofRare Diseases into SocialServices and Policies	April 2016	https://ec.europa.eu/health//sites/health/files/rare_diseases/docs/recommen

		dations_socialservices_policies_en.pdf

Joint Actions in the field of RD

EUCERD Joint Action (EJA)

The Joint action's mandate was to assist the European Commission in the formulation and implementation of activities within the rare disease community, and to foster exchanges of relevant experiences, policies and practices between Member States and stakeholders. This Joint Action was primarily prompted by the European Commission Communication "Rare Diseases: Europe's Challenge" (11 November 2008), and the Council Recommendation on an action in the field of rare diseases (8 June 2009).

The EJA had a very broad scope, and was structured around five main work areas, the first of which was the implementation of plans and strategies for RD at national level. The project built upon the achievements of previous European initiatives within the RD field. These included the European Commission RD task Force, Orphanet, the EUROPLAN project and the outputs of several RD networks that had received funding from EU in the past.

Among its main objectives, it was to "Accelerate implementation of the inter-sectoral national action plans for RDs". Key outputs were 4 sets of draft Recommendations on the topics of registries, ERNs, National plans/strategies for RDs, and codification that, at the submission on RD-ACTION Overview 2018 Version, had been presented for EUCERD and CEGRD's revision and adoption.

Besides the draft Recommendations, the EJA created many other outputs, including the following deliverable:

> *Deliverable 5: Capacity Building Report for Rare Diseases National Plans/National Strategies in EU Member States (http://www.eucerd.eu/?page%20id=3029) was one of the numerous outputs of the project, as well as the production of 3 volumes of the State of the Art report. The EJA continued in the scope of RD-ACTION, "Data and Policies for Rare Disease" (2015-2018).*

All the deliverables may be found at http://www.eucerd.eu/?page id=3029

The EJA also produced 3 volumes of the State of the Art Report and delivered 25 national EUROPLAN conferences. The support to RD provided by the EJA continued in the scope of RD-ACTION (2015-2018).

Work Programmes at European Level

At European level, research on RD has been addressed under the EU Framework Programmes for Research and Technological Development (fp) since the early 1990s. In the previous Programme (FP7 2007-2013) the Health Theme for "Cooperation" Specific Programme, was designed to support multinational collaborative research in different forms. FP7 was succeeded by Horizon 2020, the Framework Programme covering the period 2014-2020.

Horizon 2020 will, in turn, be succeeded by Horizon-Europe (2021-2027).