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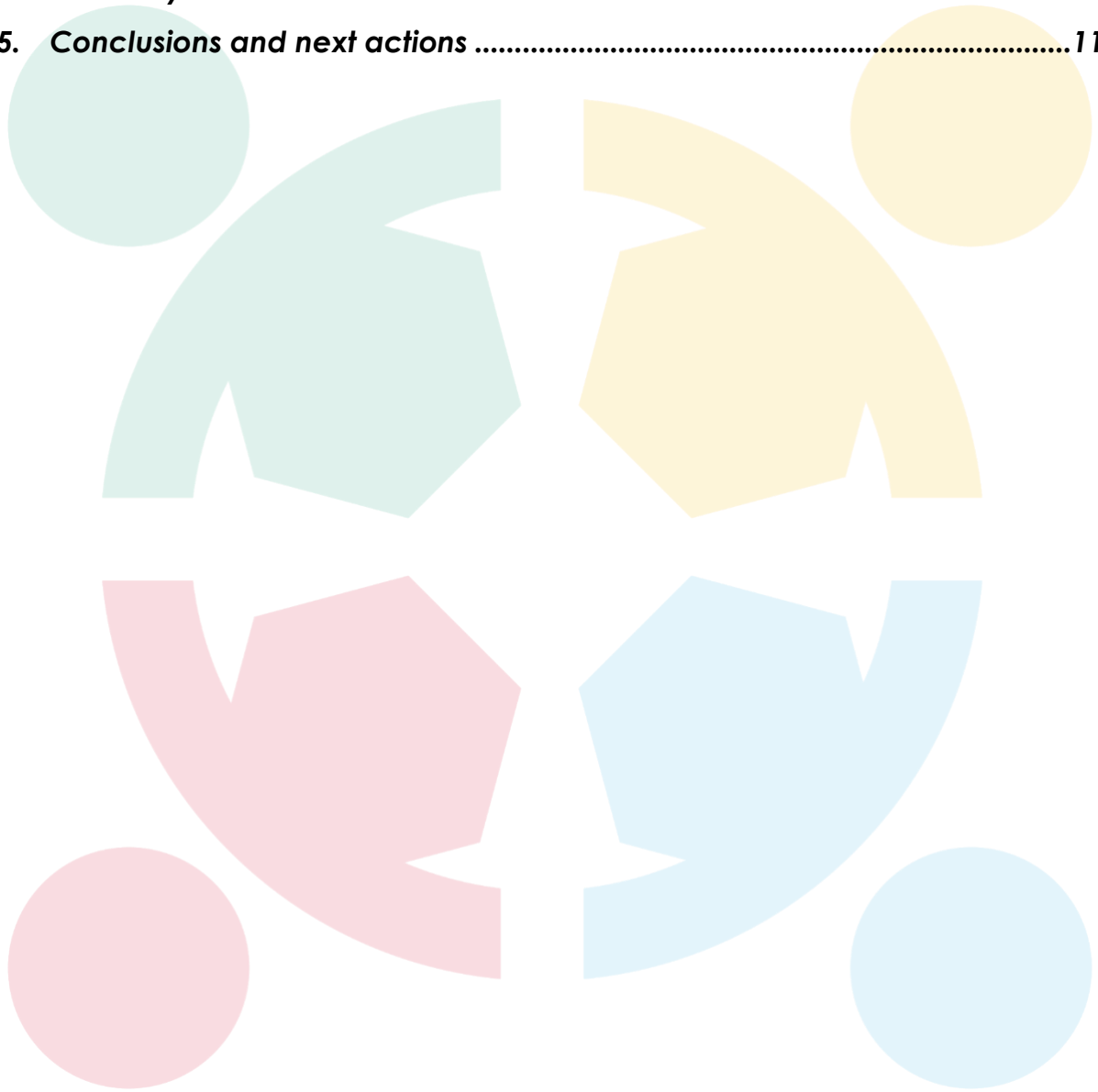
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1. Executive Summary

The Third Scoping paper is intended as an additional document and tool regarding the regular mapping of research and innovation (R&I) needs of the rare diseases (RD) community (including the European Joint Programme on Rare Diseases EJP RD), that the EJP RD performs every year. This allows up-to date and focused actions for the benefits of the RD community. This document will be transmitted to Task 2.3, 2.4 and 2.5 leaders for complementary actions, as indicated in the Description of Action. In addition, the outcomes described in the present document should be promoted and implemented by the National Mirror Groups (NMGs), once constituted, and in the Horizon Europe frame.

For the present document, several and wide-ranging inputs have been taken into account, and it reflects mainly the outcomes of the Deliverable 2.5 (D2.5) “Third Summary document on Research and Innovation (R&I) Needs” of the EJP RD.

The present deliverable reformulates the results of the R&I needs mapping exercise performed within Work Package 2 (WP2) “Integrative Research and Innovation Strategy” that contributes yearly to assure well focused Annual Work Plans as well as a consistent long-term strategy of the EJP RD, and helps moreover to the promotion and implementation of informed RD actions also outside the EJP RD.

Highlights and Next Actions

The most relevant sources for the shaping of the R&I needs are:

- The IRDiRC’s International research strategy, as a guideline for the EJP RD actions and for the achievements of the common goals, for the benefits of the RD community.
- Topics of current and previous Joint Transnational Calls (JTCs)
- The subjects discussed in the R&I Days of 2020 (Directorate-General for Research and Innovation of the European Commission & Horizon Europe).
- The highly relevant patient perspective, collected through the activities carried out by EURORDIS and highlighted also in the Rare2030 pilot project.
- Internal inputs from the EJP RD, in the form of the discussions held during the Policy Meeting of 2021 and of two surveys performed within Subtask 10.2.2 and Task 2.5.
- Other sources considering other domains besides the scientific, such as diagnostics and healthcare, regulatory and ethics, competitiveness and innovation.

Table of abbreviations

COVID-19	Corona Virus Disease-19
D	Deliverable
EC	European Commission
EJP RD	European Joint Programme on Rare Diseases
ERN(s)	European Reference Network(s)
ESFRI	European Forum on Research Infrastructures
EU	European Union
EU-13 Countries	European-13 Countries
ExCom	Executive Committee
FAIR	Findable, Accessible, Interoperable, Reusable
IRDiRC	International Rare Diseases Research Consortium
ISCIII	Instituto de Salud Carlos III
ISS	Istituto Superiore di Sanità
JTC(s)	Joint Transnational Call(s)
MUW	Warszawski Uniwersytet Medyczny
NMG(s)	National Mirror Group(s)
NP/NS for RD	National Plan/Strategy for RD
R&I	Research and innovation
RD	Rare Diseases
RWD	Real World Data
SARS-CoV-2	Severe Acute Respiratory Syndrome caused by Corona Virus-2
WP	Work Package

2. Introduction and Objectives

The main objective of the series of Scoping Papers that are being produced annually in the framework of WP 2 “Integrative Research and Innovation Strategy” of the EJP RD, is to transmit a summary of the mapped research and innovation (R&I) needs both internally and externally to the EJP RD. This transmission allows to contribute to the medium and long-term strategy of the EJP RD with well focused actions that can respond to the real needs.

Internal purpose of the Scoping Paper: The Scoping Paper is meant to reformulate the outcomes of the annual work on the mapping of R&I needs, performed in Task 2.2, to be transmitted to the leaders of Tasks 2.3, 2.4 and 2.5 for complementary actions in their Tasks. The mapping on R&I needs is updated and submitted annually, up to 2023, as a confidential document (longer than the Scoping Paper) for members of the Consortium only (including the Commission Services), through a “Summary document on Mapped Research and Innovation Needs” which helps to feed the annual plans of the EJP RD. To date, the “Third Summary document on Mapped Research and Innovation Needs” (D 2.5) has been elaborated, validated by the Executive Committee and uploaded to the Portal.

External purpose of the Scoping Paper: The Scoping Paper is also intended to have an impact out of the EJP RD itself, by being promoted and implemented by the NMGs (where constituted) and within the current EC Framework Programme, Horizon Europe.

The present deliverable is the third public Scoping Paper, prepared by ISS in close collaboration with Task 2.2 leaders (ISCIII, ISS) and the EJP RD Coordination.

To better place this Scoping Paper within the frame of the EJP RD activities, just to mention, as already stated in the previous Scoping Papers, that WP 2 is dedicated to the development of the EJP RD research and innovation strategy in connection with all related stakeholders, and has specific objectives, recalled below:

- To map the needs for RD research and innovation.
- To define the prioritization model for the EJP RD actions as part of the annual planning and in connection to WP3 (Sustainability) and WP4 (Ethical, Regulatory, Legal and IPR framework).
- To prioritize topics for the joint transnational calls (JTCs) to be implemented within Pillar 1.

- To feed the medium and long-term RD research and innovation strategy in collaboration with IRDiRC.
- To prepare a Scoping Paper to be promoted (and implemented) by the NMGs and within the current and forthcoming EC Framework Programme. This is precisely the objective to which this deliverable is contributing.

All these activities are part of the so-called Pillar 0 (“Coordination and transversal activities”), the central coordination Pillar that assists the actions of the four major Pillars of the EJP RD.

As for the previous deliverables of this series, the Scoping Paper is a summary document based on some deliverables produced within WP2, namely:

- D2.1 (“Final list of prioritization criteria”) (Public Report),
- D2.2 (“Prioritization Scheme for EJP RD actions, including decision-making process-Guidelines for prioritization”) (Public Report),
- D2.5 (“Third Summary document on Mapped Research and Innovation Needs”) (Confidential Report).

WP 2 Task 2.3, 2.4, 2.5 leaders will receive the comprehensive summary included in the present document for complementary actions in their respective tasks, specifically:

- “Scientific programming of joint transnational calls” (Task 2.3),
- “Management of the medium, longer-term research strategy questions and dedicated linkage with Task Forces of IRDiRC” (Task 2.4)
- “Translation/impact of prioritization on national and EU strategies” (Task 2.5).

Recapitulating, the objective of the series of annual Scoping Papers is to transmit a summary of the mapped R&I needs identified through the work carried out in WP 2, and especially in Task 2.2 “Mapping the research and innovation needs” for more focused EJP RD actions.

The general aim of the present Deliverable D2.10 Third Scoping Paper is therefore to support in the EJP RD progresses, contributing to the medium and long-term strategy of the project.

3. Sources for the preparation of the Third Scoping paper

The elaboration of the present document is based mainly on the update of the D2.9 “Second Scoping Paper” and on the D2.5 “Third Summary document on mapped R&I needs”.

The deliverables D2.1 “Final list of prioritization criteria” and D2.2 “Prioritization scheme including decision-making process-Guidelines for prioritization” are shortly recalled, as they have been more extensively described in the previous papers “First Scoping Paper” and “Second Scoping Paper” and constitute a general background that are complementary to the mapping of R&I needs. The activities described under D2.1 and D2.2 are moreover regularly monitored and updated, according to the emerging needs for a tailored planning of the EJP RD’s R&I strategy, to ensure a flexible alignment of the project with the real needs on R&I.

4. Summary Results

The RD research challenges tackled by the EJP RD encompass the fast translation of research results into clinical applications and uptake in healthcare, which require the identification of substantial and up-to-date R&I needs. Besides the regular R&I needs mapping exercise, that is a crucial operation for the accurate construction and information of the Annual Work Plans inside the EJP RD, the definition of R&I needs is also essential for supporting key policy recommendations for addressing RD beyond the EJP RD. Charting of R&I needs is therefore significant for the effects inside and beyond the EJP RD activities.

It is important to underline that the major activities of the EJP RD were already accurately detailed in the proposal of the project approved by the EC, and that the prioritization strategy has to be considered: (i) in the event of exceptional circumstances for major reasons (to cite an example that affected worldwide years 2020 and 2021, the spread of the COVID-19 pandemic caused by SARS-CoV-2 has impacted almost all the activities; however, through the adoption of mitigation strategies inside the EJP RD significant deviations from the proposed activities were prevented); (ii) besides the adoption of mitigation/extraordinary measures to ensure the correct progress in the EJP RD activities; the definition of the Annual Work Plans also envisages a part of adaptability, for which the prioritization strategy might be of interest for Task and Subtask leaders to establish the steps to be taken for moving forward and encountering the real emerging R&I needs.

This flexibility enables an up-to-date alignment of the EJP RD actions with the real current needs in RD research.

Brief summary on D2.1 “Final list of prioritization criteria” and D2.2 “Prioritization Scheme for EJP RD actions, including decision-making process-Guidelines for prioritization”

The aims of the prioritization activities described in D2.1 are: i) supporting and assessing the decision-making process to prioritize mapped needs and actions that contribute to the EJP RD objectives (ii) facilitating the planning of future actions within the Annual Work Plan (iii) prioritizing choices and actions in case of onset or need of deviation from planned activities (iv) further ameliorating the criteria, indicators and methodology used from the process of prioritizing, after assessing the impact of the decisions taken. Additionally, four wide scope prioritization criteria are defined, namely Scientific evidence aspects, Demands of the RD community, Regulatory concerns, Financial and technical feasibility.

These criteria are applicable and particularized to each Pillar of the EJP RD and might be used differently, depending on the Pillar specific activities.

The final list of prioritization criteria is meant to be updated and reviewed before the definition of each Annual Work Plan, if needed, to better approach the real needs of the EJP RD.

The recommendations of D2.1 have to be implemented together with the content of D2.2, that gives details on how to apply the criteria to the prioritization process and guidelines on the prioritization and decision-making process, respecting the principles of transparency and non-arbitrariness, to achieve the goals of the EJP ED for the benefits of patients.

D2.5 “Third Summary document on Research and Innovation Needs”

Summary documents on mapped R&I needs are generated yearly and sent to the Policy Board and ExCom as a basis for the prioritization activity of the R&I needs of the EJP RD. The summary document that has been taken as reference in the present Third Scoping Paper is the “Third Summary document on R&I Needs” (D2.5), a confidential report submitted in 2021. Different sources of information have been taken into account in D2.5 and cover the whole scenario of the RD field relevant for year 2021, comprising both contributions collected inside and beyond the EJP RD.

The different sources are summarized below.

1. **A survey on specific needs arisen for RD patients and their families with the outspread of the COVID-19 pandemic.** RD patients and their families revealed to be especially vulnerable in the pandemic situation, and the survey carried out by the EURORDIS Rare Barometer Programme helped to discover specific needs of patients and caregivers, to be translated in

R&I needs. Recourse to telemedicine/more digital health options on the one hand and the necessity of continuity and maintenance of the healthcare pathway on the other, were confirmed to be essential R&I needs, even and more extensively, in exceptional circumstances (e.g. a pandemic).

2. **The EURORDIS position papers.** The position papers published by EURORDIS meet the requirement to plan and prioritize the active participation of the public and patients to establish R&I needs. The previous position papers on the need for a comprehensive health data strategy have been complemented with position papers on the need of patients for a holistic, patient-centered approach in RD research. The EURORDIS position papers continue thereby to contribute to the mapping exercise of the unmet R&I needs.
3. **The “Rare2030” pilot project.** The project collected contributions from a panel of experts constituted by patients, doctors and key opinion leaders, furnishing information on community needs and summarizing the essential topics in eight knowledge base summaries to fill the gaps of the current RD policies with regard to unfulfilled RD needs. The titles of the eight recommendations are: 1. Longterm, integrated European and national plans and strategies. 2 Earlier, Faster, More Accurate Diagnosis. 3. Access to High quality Healthcare. 4. Integrated and Person- Centred Care. 5. Partnership with Patients. 6. Innovative and Needs-led Research and Development. 7. Optimising Data for Patients and Societal benefit. 8. Available, Accessible and Affordable Treatments.
4. **The ERNs and Researchers’ surveys.** This activity has been carried out inside the EJP RD (Subtask 10.2.2. “Community surveys and structured interviews”). A comparison between the two surveys on the needs of ERNs and researchers has been summarized in D2.4 (“Second Summary document on R&I Needs”), highlighting that researchers and ERNs are engaged in similar efforts (e.g., among others collaboration, data-sharing, education/dissemination of information, adoption of FAIR principles) but may face different priorities. While ERNs seem more concentrated on improving diagnostics and innovative care, researchers appear more focused on developing disease models and understanding disease mechanisms. These results are useful to design specific types of R&I needs for ERNs and researchers.
5. **A survey on National Plans and Strategies (NP/NS) for RD.** The survey has been performed within Task 2.5 of the EJP RD, “Translation/impact of prioritization on national and EU strategies” to assess the alignment process of national RD strategies with the actions promoted by the 4

major Pillars of the EJP RD. The survey had also a specific focus on EU-13 Countries with regard to specific needs, obstacles and advancements. The survey encompasses the areas of: National and International Investments in RD research, Resources and Services to foster RD research, Capacity Building and Empowerment, Accelerated translation of research projects and improvement of outcomes of clinical studies, Specific obstacles and barriers faced by EU-13 Countries for the development, improvement and translation of RD research results and for their participation in EU/International RD research projects. The results of the survey contributed to uncover the main areas that require an increase of efforts inside the EJP RD Pillars and represent a reference for the prioritization of the coming activities of the project.

6. **The discussions held during the session of the EJP RD Policy Meeting of January 2021.** The issues discussed during the EJP RD Policy Meeting were taken into account to feed the mapping of further R&I needs of the EJP RD. ERNs, improvement of data sharing and analysis, the development of standards and interoperability, the importance of Real World Data (RWD) for the advancement in genetic testing and gene therapy can be cited as some of the major topics, together with the national strategies, education and empowerment, social justice and the future of RD initiatives.
7. **The ongoing Task Forces that encounter the three IRDiRC goals.** The three IRDiRC goals for the period 2017-2027 (diagnosis within 1 year for RD patients, 1000 new therapies approved and the development of the methodology to evaluate the impact of diagnosis and therapies for RD) continue to constitute an overall basis for the identification of the R&I needs in the RD field. The focus of the ongoing Task Forces (some delayed due to the Covid-19 pandemic) rely on several topics, e.g., new technologies for diagnosis, shared molecular etiologies research, key factors to attract industry for research and development, research on sustainable economic models for the development and commercialization of orphan drugs. In addition, a Re(ACT) Congress (International Congress of Research on Rare and Orphan Diseases) and an IRDiRC Conference took both place at the beginning of 2021, showing the current progresses and needs in R&I. Overall a trend on projects conducting research on new technologies and artificial intelligence has emerged, and this will be reflected on the formulation of the coming R&I. More extensively, the topics of the Re(ACT) Congress-IRDiRC Conference of January 2021 were: Diagnostics, Whole Genome Sequencing, Artificial Intelligence, New Technologies, Molecular etiology of RD innovative clinical trials precision medicine, Advanced therapies,

Patient engagement in drug development and clinical trial, Access to diagnostic and drug for all, Methodologies to assess the effect of diagnostics and therapeutics in RD pathologies.

8. **Topics approached in the Joint Transnational Calls (JTC)s.** The topics mirror the needs and interest of the RD community, and the 2021 selected topic was “Social Sciences and Humanities Research to improve health care implementation and everyday life of people living with a rare disease”, with specific subtopics (e.g. “Psychological and Social Impact of RD”, “Economic Impact of RD”, “Health & social care services research to improve patient and familial/household health outcomes”). The selection of the next JTCs topic of 2022 will be oriented by the identified R&I needs.
9. **The progresses of the “European Forum on Research Infrastructures (ESFRI) roadmap 2021” and the launching of Horizon Europe 2021-2027.** New research infrastructure projects and the assessment of research performance figure moreover in the list of contributions for the recognition of areas that can suggest R&I needs. Specific areas emerged in the planning of Horizon Europe and in the “Research and Innovation Days” held in September 2020 have been recognized to be of particular relevance for advancing in research, and comprise (as stated above) artificial intelligence, innovations in data sharing and technologies. The next “Research and Innovation Days” of 2021 will be considered, among other sources, for the Fourth Summary document on R&I needs. Moreover, the European partnership turned out to be an essential issue in the framework of Horizon Europe 2021-2027 and the European partnership in Rare Diseases stands out as a possible partnership. The main goal of the RD Partnership (2024) would be to improve the life of patients with rare diseases by developing diagnostics and treatments thanks to multidisciplinary research and coordination of national, local and European R&I programmes.
10. **Other sources.** For the mapping of R&I needs other domains than the scientific have been explored, for a complementary vision on global R&I needs: the diagnosis & healthcare, the Regulatory & ethics and the EU competitiveness & innovation domains.

5. Conclusions and next actions

The EJP RD fosters a concrete and up-to date alignment with the IRDiRC goals for diagnosis, treatment and impact assessment through the yearly mapping of R&I needs, gathering inputs from the research, clinical and policy area, as well as from patients, patient organisations and from other domains, like

regulatory and ethics, pharmaceutical industry or EU competitiveness & innovation.

Globally, the R&I needs that emerged, rely on the necessity to progress in knowledge, understanding, and innovation in rare diseases research and envisage an expanded view into health systems for rare diseases, with a holistic, patient-centred approach.

In the definition of the current R&I needs the topics and subtopics of the JTCs reflect the interests of the RD community and represent an up-to date and key source for the understanding of the RD community needs. The subjects of the Re(ACT) Congress-IRDiRC Conference of January 2021 offer a wide-ranging overview of most relevant R&I needs.

The collaboration and feedback of patients as crucial stakeholders, also with reference to exceptional circumstances as the COVID-19 pandemic (that shed light even on more general needs of persons living with a rare disease), continue to be central for the definition of the R&I needs. Besides the needs for a comprehensive health data strategy, needs for a holistic, patient-centered approach have been identified as relevant focuses. Moreover, the Rare 2030 pilot project delivered 8 Recommendations concentrated on different specific aspects aiming at a future “Investment for Social Justice”, guided by need-led innovation, collective responsibility and outcome-based goals.

Topics of the JTCs and the framework of Horizon Europe 2021-2027 pose further impulses to feed the R&I needs. In particular, the European Partnership on Rare Diseases shall aim at improving the life of patients with rare diseases by developing diagnostics and treatments through multidisciplinary research and coordination on national, local and European R&I programmes, considering training, data related topics and open science as pivotal issues.

Inputs from inside the EJP RD for the definition of the R&I needs derive from the discussions held during the sessions of the EJP RD Policy Meeting of January 2021 and from a survey on National Plans and Strategies for RD, both comprising all major aspects faced by the EJP RD and reflecting the topics constituting the actions of the 4 EJP RD Pillars, as well as a look to the future of RD actions.

Also, a survey on ERNs and researchers needs counts among the inputs gathered inside the EJP RD. The researchers and ERNs R&I needs require each specific and dedicated attentions, with the first appearing to be more focused on improving diagnostics and innovative care and the latter on developing disease models and understanding disease mechanism.

Finally, other domains like diagnosis and healthcare, regulatory and ethics and EU competitiveness are considered for a wider definition of the R&I needs.

The next actions will be the transmission of the present Scoping Paper to the T2.3, T2.4 and T2.5 leaders for complementary actions, to the NMGs (if constituted), and within the forthcoming EC Framework Programme.

In year 4 a Fourth Scoping Paper will be elaborated by MUW, as an additional tool summarizing the efforts of the mapping of R&I needs performed within WP 2 “Integrative research and innovation strategy” of the EJP RD.

