Mimouni *et al. Rare Dis Orphan Drugs J* 2024;3:17
DOI: 10.20517/rdodj.2024.06

Rare Disease and Orphan Drugs Journal

Perspective

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The European joint programme on rare diseases: building the rare diseases research ecosystem

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How to cite this article: Mimouni Y, Halftermeyer J, Petton Y, Adam P, Moreau C, Rath A, Favresse R, Tumiene B, Julkowska D. The European joint programme on rare diseases: building the rare diseases research ecosystem. *Rare Dis Orphan Drugs J* 2024;3:17. https://dx.doi.org/10.20517/rdodj.2024.06

Received: 2 Feb 2024 **First Decision:** 11 Apr 2024 **Revised:** 7 May 2024 **Accepted:** 14 Jun 2024 **Published:** 24 Jun 2024

Academic Editors: Tao Duan, Jacques S. Beckmann **Copy Editor:** Fangyuan Liu **Production Editor:** Fangyuan Liu

Abstract

The European Joint Programme on Rare Diseases (EJP RD) represents a significant step forward in the rare diseases research ecosystem, integrating over 130 institutions from 35 countries. It focuses on streamlining the process from basic research to clinical application, fostering collaboration at an international scale, and emphasizing patient engagement. The initiative has funded numerous projects across various medical domains, developed the Rare Diseases Virtual Platform, and significantly impacted the rare disease landscape through training, project mentoring, and innovative methodology developments. Expanding beyond, EJP RD collaborates with various initiatives to optimize resource use and is evolving into the European Rare Diseases Research Alliance, aiming to accelerate diagnostics, develop innovative therapies, and assess the rare disease burden to inform policy decisions.

Keywords: Rare diseases, research funding, virtual platform, capacity building, research acceleration, patients' empowerment, innovative methodologies, translational research

WHAT IS EJP RD?

For years, many stakeholders in Europe, with the support of the European Union, have contributed to

building elements of the Rare Diseases (RD) ecosystem. New avenues for the RD community were opened thanks to the emergence of important initiatives, instruments, and organisations, including the creation of the resource platform Orphanet and the patients' organisation EURORDIS (1997), the establishment of the International Rare Diseases Research Consortium (IRDiRC) (2011), the implementation of the European Reference Networks (ERNs) (2017), and the launch of the European Platform for Rare Disease Registration (2018). At the national level, many European countries adopted national plans (NP) and/or national strategies (NS) to tackle rare diseases, including some policies for research. At the EU level, these endeavours were supported by EU co-funding through the so-called ERA-Net scheme (networking of national research funding agencies): E-Rare-1, E-Rare-2, and E-Rare-3. However, the integration of these foundational building blocks into a cohesive structure remained necessary. The launch of the European Joint Programme on Rare Diseases (EJP RD) was a major milestone and a prime example of Member States working together in a coordinated manner beyond joint funding activities. EJP RD (2019-2024) aims to establish an effective research ecosystem for rare diseases (RD), promoting progress and innovation to benefit patients[1]. This initiative unites over 130 institutions from 35 countries, exemplifying cooperative efforts between Member States and a range of (above-mentioned) stakeholders including research funders, ministries, research institutes, universities, European Reference Networks (ERNs), EU research infrastructures, foundations, and patient organizations. The overall budget of EJP RD - 101 million euros - is supported by the EU R&I funding programme Horizon 2020 (55 million euros) and the participating entities.

EJP RD focuses on creating a streamlined process from basic research to clinical application ("bench to bedside") to accelerate the translation of research findings into healthcare improvements. The programme builds upon 15 years of work in the RD field, integrating and expanding existing infrastructures, training programmes, funding mechanisms, and tools, while also developing new, essential components. This approach has resulted in a harmonized, centralised RD research ecosystem, incorporating cross-sectoral and integrative strategies to tackle health challenges.

The programme's structure encompasses Collaborative Research Funding, Coordinated Access to Data, Tools, and Services through a Virtual Platform, Capacity Building through Training and Empowerment, Accelerated Translation of Research Results and Clinical Studies, and Centralised Coordination and Transversal Activities. These latter activities also cover strategy, sustainability, ethics, regulatory issues, and communication, all integral to the programme's success. The EJP RD research support cycle is presented in Supplementary Figure 1.

EJP RD AS PART OF THE OVERALL RD LANDSCAPE

Collaboration is crucial in bolstering the EU's leadership in the RD domain. At international scale, the EJP RD serves as a pivotal tool in fostering stakeholder cooperation within the RD field, promoting enhanced research, policy alignment, as well as the sharing of infrastructure and knowledge across various scales for the benefit of patients.

The international dimension of EJP RD is also reflected by its alignment with the International Rare Diseases Research Consortium's (IRDiRC) objectives[2,3]. EJP RD hosts and supports the Scientific Secretariat of IRDiRC. This cooperation is underscored by a strong, mutually inclusive link in research and policy actions. The chair of IRDiRC brings a global dimension as a member of EJP RD's Policy Board[4]. Independent IRDiRC experts contribute to discussions on topics related to EJP RD's calls for projects. On the other hand, since 2019, 42 experts from EJP RD beneficiary institutions contributed to IRDiRC Task Forces, with 24 participating in its scientific committees[5]. Furthermore, collaborative initiatives include

joint conferences like the 2021 and 2023 RE(ACT) Congress & IRDiRC Conference[6] and jointly produced tools such as the Innovation Management Toolbox[7] and Orphan Drug Development Guidebook[8].

Patient engagement is a vital component of transnational collaboration in all rare diseases research areas. The EJP RD adopts a patient-centred approach, by supporting patient engagement and involvement in its activities and by funding patient organizations in research projects. Moreover, EURORDIS[9], the patient organizations alliance, plays a significant role in the programme. It co-leads the training and empowerment pillar[10], oversees training activities like the one on leadership and communication skills for patient representatives[10], co-develops EJP RD's Massive Open Online Courses[10], and is involved in the governance structures.

The connection with national stakeholders is essential for aligning European and national research policies and activities. The EJP RD facilitates this through the establishment of National Mirror Groups (NMGs) in participating countries[11]. These groups bring together expertise and knowledge from the RD community in a specific country by joining national/regional RD programmes, infrastructures, and centres of expertise. They facilitate the identification of national needs and policy gaps, advocate for reforms conducive to the EJP RD's success, and promote national alignment with the European research strategy. To date, four NMGs have been established in the Netherlands, France, Portugal, and the UK, with an additional ten expected to benefit member countries by the end of the EJP RD.

EJP RD ACTIVITIES AND ACHIEVEMENTS

Research funding

EJP RD Joint Transnational Calls stem from the three successive ERANETs E-Rare: the first one launched in 2006, the second (E-Rare-2) in 2010, and the third in 2014 (E-Rare-3, 2014-2019, H2020), with the major goals being to foster the systematic exchange of information and build a transnational research funding programme on rare diseases. EJP RD has maintained a steady focus of its predecessors on multinational RD funding activities. Taken together, from 2006, 207 projects were funded through 15 Joint Transnational Calls (JTCs)[12], allocating a budget exceeding 223 million euros. The European Commission (EC) co-funded selected research projects in 2015, 2019, and 2020.

The funders within EJP RD allocated EC co-funding to support Patient Advocacy Organisations (PAO) in 2019, 2020, and 2021, resulting in the funding of 44 PAOs - the following JTCs being funded only through national funding, no central EC budget was available to continue this support, but many national funders strived to provide some alternatives. Concurrently, collaborations among patients, funders, and researchers led to the development of a guide on patient partnership in rare diseases research[13]. This initiative was successful, with 100% of the projects funded in the JTC-2021 involving at least one patient organisation. In parallel, EJP RD has laid a strong foundation for patient-centeredness in RD research training activities. A total of 425 patients or patient representatives attended specialised workshops[14]. This engagement also contributed to the increased participation of patient representatives in EJP RD JTCs and NSS calls[15]. It also has a possible longer-term impact on the implication of 150 patients in protocol development at the European Medicines Agency (EMA).

To further strengthen connections within the RD community, EJP RD introduced a new funding model for networking events, the Networking Support Scheme (NSS)[15]. Since 2020, this scheme has supported 69 events conducted in-person, online, or in a hybrid format following the COVID-19 crisis. Significantly, 22 (32%) of these networking events were coordinated by PAOs, and 50 (72%) included participants from under-represented countries (8 being the coordinator of the Networking event), highlighting the scheme's

effectiveness in fostering broader involvement. EJP RD has also made significant efforts to include underrepresented countries through widening activities in JTCs: 19 teams from 6 countries were included in funded consortia in the JTC2019 to 2023 through the widening process. Building on this achievement, the next phase of the programme, as described below, plan to empower under-represented countries and measure their influence on research activities and results uptake.

Additionally, EJP RD launched a novel funding initiative involving industry participation and co-funding. The Rare Diseases Research Challenges Call, implemented in 2020, funded three projects in response to challenges defined by the involved industrial partners[16]. These 30-month projects, started in late 2021, are aiming to deliver tangible solutions to these challenges, with a swift translation of results to patient benefit. The innovation aspects were also addressed within 5 EJP RD-funded projects fostering demonstration and innovation in methodologies for clinical trials[17,18] [Supplementary Table 1].

The projects stemming from the 15 joint transnational calls of the EJP RD and E-Rare programme comprehensively cover various medical domains and stages of the research pipeline. Notably, about 35% of the funded projects focus on neurological diseases, followed by 12% on haematology and immunology, and 7% on metabolic diseases [Figure 1]. The distribution of the funded project per disease area aligns with the distribution of initially submitted proposals, and is not caused by any specific bias in the selection of particular domains. In addition, to keep the balance and avoid over-financing of certain domains, the EJP RD funders systematically benchmark topics of other existing initiatives and, if needed, exclude topics or domains already covered in other similar programmes (i.e., JPND, ERA4Health, *etc.*). The funded projects range from basic research in molecular and pathophysiological studies to clinical studies, diagnostic studies, preclinical and validation studies, natural history studies, and social sciences and humanities [Figure 2]. Notably, for the latter two areas, dedicated JTCs were launched in 2021 and 2023. Importantly, many projects encompass multiple approaches. Both E-Rare programmes and EJP RD mostly focus on fundamental, preclinical, and translational research. The results of the clinically oriented projects are yet to come.

Rare diseases virtual platform

In 2023, EJP RD officially opened a public portal for accessing the Rare Diseases Virtual Platform (VP)[20]. This platform is a growing network of Findable, Accessible, Interoperable, and Reusable (FAIR) resources developed or enhanced by EJP RD to support the rare disease research community. It features a variety of sources, including resource catalogues, registries, biobanks, knowledge bases, animal models and cell line libraries, omics deposition and analysis platforms, and tools adhering to agreed standards[21]. The portal enables real-time, one-stop searching of the VP network's resources, facilitating their discovery and preventing the need to navigate multiple platforms.

Data stay at its source but can be remotely queried from various network points. This setup allows for federated discovery, querying, and analysis while maintaining patient confidentiality and adhering to each resource's access conditions. Certain resources, like patient registries, require authentication for detailed data queries, whereas others, like catalogues and knowledge bases, are freely accessible. Logging in enables the application of specific patient data search filters, providing aggregated results.

The VP's adherence to FAIR principles ensures its viability, enhancing data discovery, indexing, integration, and reuse under clear conditions. FAIR implementation also allows for secure, automated data handling with minimal human involvement. To facilitate this, EJP RD has developed services, guidelines, tools, and training for FAIRification[22].

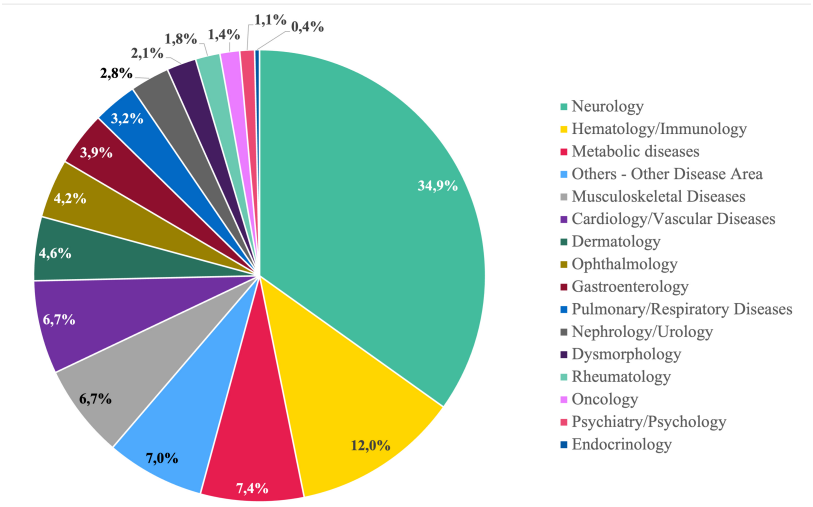

Figure 1. Overview of the distribution of the 284 EJP RD- and E-Rare-funded projects from 2007 to 2023 (including 207 JTCs, 69 NSS, 3 RDRC, and 5 Demonstration/Innovation funded projects) per disease area. The numbers are shown as percentages of the total number of projects.

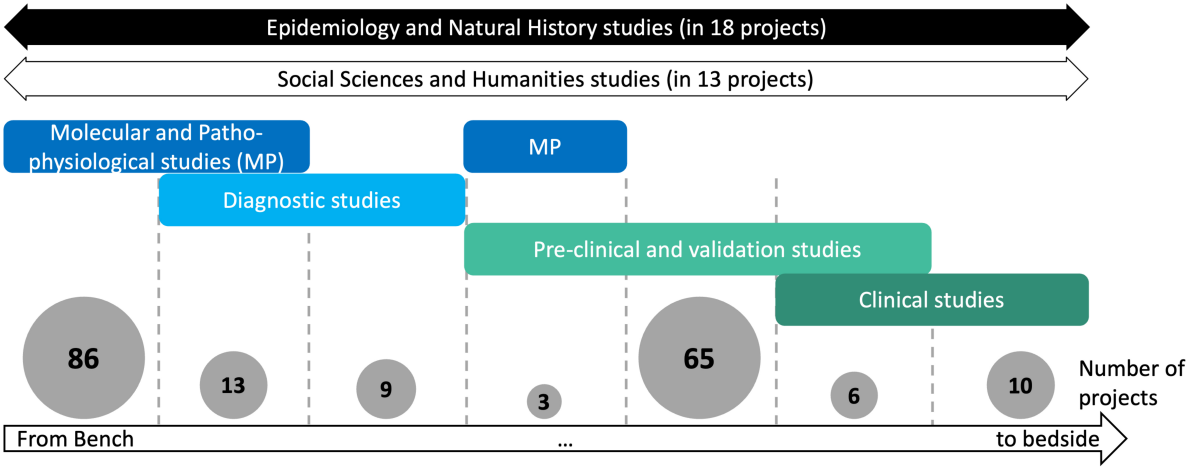

Figure 2. Distribution of 215 projects funded by EJP RD and E-Rare from 2007 to 2023 (including 207 JTCs, 3 RDRC and 5 Demonstration/Innovation funded projects) throughout the research pipeline. All research projects have been analysed to classify their work into categories[19] and grouped into 6 non-exclusive categories: molecular and pathophysiological studies; diagnostic studies; preclinical and validation studies; clinical studies; epidemiology and natural history studies; social sciences and humanities studies. Many of the projects could be assigned to more than one category, which is reflected in the numbers of project distribution. Epidemiology and natural history studies and social sciences and humanities studies have been found to be associated with different steps of the research pipeline in funded projects, and thus, they have been identified as overarching research topics. Numbers of projects in those 2 arrow groups projects focusing only on this topic or in combination with any of the other 4 topics (molecular and pathophysiological studies; diagnostic studies; preclinical and validation studies; clinical studies). MP: Molecular and pathophysiological studies.

The current VP network primarily consists of resources and tools that have collaborated with EJP RD. This work streamlined the process for data producers to submit data to the resources, enhancing discoverability. User-friendly interfaces, utilising relevant ontologies and standards, have been developed for data and metadata collection. Quality assurance mechanisms include manual curation, automatic metric generation, or a combination of both. Enhanced APIs and GUIs allow for effective querying and data access[23-27].

Workflows and tools for phenomic and multi-omics analysis, integration, and sharing, aiding in diagnostic acceleration, were also further developed[28-31] and are being connected to the VP. Additionally, resources for systems biology research on rare diseases have been updated and expanded. This approach mitigates the challenge of limited RD patient and sample numbers for data analysis by incorporating prior knowledge from known molecular interactions and annotated multi-omics data. Various methods have been developed for hypothesis generation relating to diagnostics, drug repurposing, and identifying potential toxic interactions in rare diseases[32-34]. In collaboration with the ELIXIR rare disease community, a comprehensive systems biology service bundle was created, including guides and tutorials on tools and methods for systems biology in rare diseases[35].

Looking ahead, the VP will expand to include new resources that address the ongoing and emerging needs of rare disease research.

Training and empowerment

A significant impact on the RD landscape was achieved through EJP RD support activities, which primarily include training[36] and project mentoring[37]. Stakeholders from widening countries have shown active participation in these initiatives. In data management and quality training, they represented 12% to 50% of the attendees[38]. Moreover, research mobility fellowships[39] and ERN Research training workshops[40] beneficiaries from these countries constituted 20% and 10%, respectively[38]. The Massive Open Online Course (MOOC) on "Diagnosing Rare Diseases" has drawn over 5,800 participants from 150 countries since April 2021, illustrating EJP RD's extensive global influence beyond the EU[41].

The programme has also been instrumental in accompanying researchers by providing highly specialised knowledge. In 2020, 15 projects from 8 countries benefited from EJP RD mentoring services (confidential EJP RD deliverable No 19.1). From 2021 to 2022, this expanded to include a total of 32 projects with Principal Investigators from 12 countries (confidential EJP RD deliverable No 19.2). Mentoring support is essential to accelerate the translation of research results as it empowers researchers with expertise on regulatory, biostatistical, or business development aspects that are key to increasing the success rate of the uptake and follow-on funding. Furthermore, EJP RD implemented the "train the trainers" concept on an international scale, standardizing and disseminating high-quality education programmes. An example of this was the training dedicated to Orphanet nomenclature and RD ontologies, which was adapted in five different countries[42]. The EJP RD training and capacity-building activities helped promote interdisciplinary collaboration among scientists, clinicians, and patient representatives, further enhancing the EJP RD's impact on the RD research field.

EXPANDING BEYOND EJP RD

Avoiding duplication and optimizing resource use in synergy with other initiatives has been a cornerstone of the EJP RD. The sustained collaboration with the European Rare Disease Research Coordination and Support Action consortium (ERICA) has bolstered RD research with ERNs, focusing on patient registries, biobanking, translational research, and clinical trials. The EJP RD plays a pivotal role in the Global Alliance for Genomics and Health (GA4GH), driving the development of standards, products, and policies for genomic data sharing and interoperability[43]. Particularly, EJP RD's contributions to data processing through its RD use-case have been instrumental in contributing to the establishment of the federated ecosystem for genomic data in Europe, as part of the 1 + Million Genomes (1 + MG) initiative[44].

In collaboration with the Critical Path Institute, EJP RD is advancing data standards and ontologies, mapping and transforming data, and developing semantic models for rare diseases[45,46]. Additionally,

EJP RD's alignment with the European Health Data Space ensures interoperability and supports the shared goal of safely utilising health data for research, innovation, and policymaking.

Moreover, the partnership with the Pan-European Paediatric Clinical Trial Network "connect4children", the European Federation of Pharmaceutical Industries and Associations (EFPIA), and the European Medicines Agency is pivotal in progressing toward clinical trial readiness. This is achieved through capacity-building activities that focus on innovative methodologies[47]. Furthermore, the collaboration with EFPIA, the European Confederation of Pharmaceutical Entrepreneurs, and the European Association of Bioindustries contributed to shaping the Rare Disease Moonshot. This initiative scales up public-private partnerships to accelerate scientific discovery and drug development in rare and paediatric diseases for which currently there is no therapeutic option[48].

Building on its achievements and successful collaborations, the EJP RD is set to evolve into the "European Rare Diseases Research Alliance (ERDERA)"[49], encompassing over 170 public and private organizations across more than 36 countries. This programme is secured with research funding for the next seven years, extending into clinical trial sponsorships. At the heart of ERDERA are the European Clinical Research Network and Acceleration Hub, dedicated to accelerating diagnostics, clinical trial readiness, innovative therapies development, and outcome research. ERDERA will support RD research by providing essential services such as data processing, regulatory compliance, innovative methodologies, and training support, ensuring strategic coherence and resource efficiency on a global and national scale.

The RD research ecosystem fostered by EJP RD and expanded by ERDERA aims to significantly accelerate diagnosis establishment and research enrolment for undiagnosed patients on average within six months of medical presentation, contribute to the development of effective RD therapies approved in the EU and internationally, and deepen understanding of RD's societal impact to inform policymaking. From the outset, ERDERA will leverage EJP RD's sustainability plan and an exit strategy to ensure its long-term viability by linking development and delivery efforts. This includes establishing a sustainable vision, operationalising core values like data and expertise infrastructure, clinical research services, and funding support, and incentivizing knowledge sharing and commercial engagement. The international collaboration and information sharing fostered by EJP RD will remain key to ERDERA. Its Strategic Research and Innovation Agenda (SRIA) already lists the pre-identified collaborations with EU programmes, missions, and other initiatives. These collaborations will expand to other entities according to arising needs and interests[50]. Ultimately, ERDERA strives to form a European Federated Rare Disease Research organisation capable of sustaining long-term public and private support, thereby transforming the landscape of RD research.

DECLARATIONS

Acknowledgements

We are deeply thankful to the entire EJP RD consortium for their unwavering commitment and significant contributions toward the development and success of this consolidated rare disease research ecosystem. This work stands on the collective efforts of a diverse group of experts and institutions whose dedication, interdisciplinary collaboration, and innovative thinking have propelled forwards our understanding of rare diseases. Their relentless pursuit of excellence, shared knowledge, and resources have been instrumental in overcoming the challenges associated with rare disease research. We acknowledge the unique role of each member and the synergy that has enabled us to achieve our shared goals. This achievement shows the power of collaboration in advancing scientific discovery and improving medicine and care.

Authors' contributions

Made substantial contributions to the conception of the perspective article: Mimouni Y, Julkowska D, Halftermeyer J, Adam P, Moreau C, Petton Y, Rath A, Favresse R, Tumiene B

Performed data acquisition: Halftermeyer J, Rath A

Performed data analysis and interpretation: Halftermeyer J, Mimouni Y, Rath A, Tumiene B

Availability of data and materials

Not applicable.

Financial support and sponsorship

This work is supported by funding from the European Union's Horizon 2020 research and innovation programme under the EJP RD COFUND-EJP N° 825575.

Conflicts of interest

All authors declared that there are no conflicts of interest.

Ethical approval and consent to participate

Not applicable.

Consent for publication

Not applicable.

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