



Call for Proposals 2023

"Natural History Studies addressing unmet needs in Rare Diseases"

Call Text

Submission deadline for pre-proposals: February 15th, 2023 at 2 PM (CET)

For further information,

An **information webinar** will be held on **December 15th, 2022**, 14.30-16.00 (CET). You will need to register to participate in the webinar here:

<https://forms.office.com/r/CLYFTuzRyw>

Visit us on the web:

<http://www.ejprarediseases.org/>

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1. Background

There are at least 7000 distinct rare diseases, the great majority being of genetic origin. Although individually rare, taken together rare diseases affect at least 26-30 million people in Europe. Moreover, they represent a major issue in health care: a large number of these diseases have an early or very early onset and/or lead to a significant decrease of life expectancy. Moreover, most of them cause chronic illnesses with a large impact on quality of life and the health care system.

Therefore, research on rare diseases is needed to provide knowledge for prevention, diagnosis, better care and everyday life improvement for patients. Yet, research is hampered by lack of resources at several levels: (1) Few scientists work on any given specific disease, (2) There are few patients per disease and they are scattered over large geographic areas, causing difficulties to assemble the necessary cohorts, (3) Existing databases and bio-material collections are usually local, small, and not accessible or standardized, (4) The complex clinical phenotypes of these diseases require interdisciplinary cooperation to improve research and treatment.

The specificities of rare diseases - limited number of patients per disease, scarcity of relevant knowledge and expertise, and fragmentation of research - single them out as a distinctive domain of very high European added-value. **Rare diseases are therefore a prime example of a research area that necessitates collaboration/coordination on a transnational scale.**

In this context, the **European Joint Programme on Rare Diseases (EJP RD)** has successfully implemented four Joint Transnational Calls since 2019 to further help in coordinating the research efforts of European, Associated and non-European countries in the field of rare diseases and implement the objectives of the International Rare Disease Research Consortium (IRDiRC). These actions are following the ten Joint Transnational Calls for rare diseases research projects launched previously by the ERA-Net E-Rare since 2006. The aim of the call has to be in compliance with the vision and goals set by the International Rare Diseases Research Consortium (IRDiRC) which fosters international collaboration in rare diseases research. For more information, visit the [IRDiRC website](#).

2. Participating organisations

A number of national and regional funding organisations will participate in the **EJP RD Joint Transnational Call (JTC) 2023** and will fund research projects on rare diseases. The call opens simultaneously with the involvement of the following funding organisations in their respective countries/regions:

- Fund for Scientific Research - FNRS (F.R.S.-FNRS), Belgium, French-speaking community
- Canadian Institutes of Health Research – Institute of Genetics (CIHR-IG), Canada
- French National Research Agency (ANR), France
- German Ministry of Education and Research (BMBF), Germany
- German Research Foundation (DFG), Germany
- National Research, Development and Innovation Office (NKFIH), Hungary*
- Health Research Board (HRB), Ireland
- Chief Scientist Office of the Ministry of Health (CSO-MOH), Israel
- Italian Ministry of Health (IT-MoH), Italy
- Fondazione Telethon (FTELE), Italy
- Regional Foundation for Biomedical Research (FRRB), Lombardy - Italy

- Tuscany Region (RT/TuscReg), Tuscany-Italy
 - Research Council of Lithuania (LMT), Lithuania
 - National Research Fund (FNR), Luxembourg
 - National Centre for Research and Development (NCBR), Poland
 - Slovak Academy of Sciences (SAS), Slovakia
 - National Institute of Health Carlos III (ISCIII), Spain
 - Swedish Research Council (SRC), Sweden
 - Vinnova, Sweden
 - Swiss National Science Foundation (SNSF), Switzerland
 - The Scientific and Technological Research Council of Türkiye (TUBITAK), Türkiye*
- * (decision still pending)

3. Management and Evaluation Structures

Two boards, the Call Steering Committee (**CSC**) and the Scientific Evaluation Committee (**SEC**), will manage the evaluation process of the call with support of the Joint Call Secretariat (**JCS**) (ANR, France). SEC and CSC members are not allowed to submit or participate in proposals within this call. The process includes the evaluation procedure of pre- and full proposals, the final selection and the award of research projects.

The Call Steering Committee (CSC) is composed of a single representative from each country/region funding organisation that participates in the JTC2023. The CSC will supervise the progress of the call and the evaluation of proposals. The CSC will make the final funding recommendation to the national/regional funding organisations on the proposals to be funded, based on the final ranking list provided by the SEC. All decisions concerning the call procedures will be taken by the CSC.

The Scientific Evaluation Committee (SEC) is a panel of internationally recognized, independent, scientific experts responsible for the evaluation of submitted proposals. SEC members must sign a confidentiality form and a statement to confirm that they do not have any conflicts of interest.

4. Aim of the call

The aim of the call is to enable scientists in different countries to build an effective collaboration on a common interdisciplinary research project based on complementarities and sharing of expertise, with expected impact to use the results in the future for benefit of patients.

Projects shall focus on **a group of rare diseases or a single rare disease following the European definition** i.e. a disease affecting not more than **five in 10.000 persons** in the European Community, EC associated states, and Canada. Applicants are encouraged to assemble groups of rare diseases based on relevant criteria and commonalities if these leverages added value in sharing resources or expertise.

Topic: “Natural History Studies addressing unmet needs in Rare Diseases”

❖ 4.1 Topics list

The objective of this call is to conduct efficient, innovative and high-quality natural history studies which will facilitate understanding of the disease's or group of disorders' progression throughout the lifespan of a patient. The goal of these studies is to collect and analyze comprehensive patient data to define targets for future therapies, taking into consideration innovation, safety, and efficacy.

Research proposals should cover **at least one** of the following areas:

- Estimation of disease prevalence;
- Identification of biomarkers/companions for the diagnosis/prognosis of a RD;
- Identification of biomarkers/indicators/predictors of a rare disease or group of disorders (e.g. having the same aetiology) onset/progression (including collection of genetic, physiological, environmental data or variables....);
- Identification of relevant endpoints for future studies that include potential biomarkers, querying patient-reported outcomes (PROs) and quality-of-life measures;
- Identification of biomarkers/variables for therapeutic approaches (pharmacology, drug repurposing, gene therapy, RNA therapy, cell therapy, medical devices...).

It is possible to use cellular and animal models for validation of endpoints and biomarkers uncovered in the natural history studies where relevant.

Furthermore, additional elements need to be considered in the application:

- Strategies and timelines for patient recruitment, retention, assessment, and analysis must be included. Data supporting the proposed recruitment numbers is mandatory. The study design and objectives should take into consideration what information regarding the rare disease population would be needed in order to pursue clinical trials or other health care related studies in the specific rare disease/group of rare diseases studied. There always need to be clear research questions that are addressed in the study/registry. Clear plans for sustainability of the resources must be described. Consideration of common data elements as outlined in the recent publication “[Set of Common Data Elements for RD Registration](#)”;
- Integration of appropriate bioinformatics and statistical skills should constitute, whenever justified, an integral part of the proposal, and the relevant personnel should be clearly specified;
- Proposals are expected to consider how sex and/or gender might shape research activities. Applicants are encouraged to visit European Commission, Directorate-General for Research and Innovation, [Horizon Europe, gender equality](#), and [CIHR's Sex, Gender and Health Research resource page](#) for more information on key considerations for the appropriate integration of sex and gender in their proposal;

- Providing harmonized/standardized data, collected using innovative means (AI, simulations, modelling...) are highly encouraged. Modelling and simulation allow for organisation of diverse data sets for optimisation of the possible future therapeutic product based on individual physiology and genetics. Innovative methods for data collection as well as data dissemination which can serve as a model for future studies are highly encouraged;
- The new research data resulting from the project should be treated permissible according to the Findable, Accessible, Interoperable, Reusable for humans and machines conforming to FAIR principles (for more information, see [The FAIR Guiding Principles](#)), and deposited and shared, according to the national/regional rules of the countries involved. It is strongly advised to make data accessible through RD-Connect or Elixir. It is strongly advised to make use of IRDiRC recognized resources and European and global resources for making data, tools, workflows, data models, (e.g. see the [EJP RD resource map](#), and the [ELIXIR Research Data Management Toolkit](#)). To make research data FAIR, a data management strategy for the proposed full project is mandatory in the full proposal stage. It should contain a timeline, budget, and designated consortium partners to comprise a FAIR data steward team. If FAIR expertise is not present in the project consortium, then a FAIR partner can be added later, possibly identified via the EJP RD helpdesk. The FAIR expert group guides the FAIR publication process, including alignment with the [EJP RD Virtual Platform](#) specifications, and helps identify goals for exploiting FAIR data. Research including Indigenous people should also adhere to the [CARE \(Collective benefits, Authority to control, Responsibility, Ethics\) Principles for Indigenous Data Governance](#). Some countries involved in EJP RD JTC 2023 will also ask for a data management plan (DMP) at national level at the stage of the full proposal or after granting of the project;
- Insights into defining genotype/phenotype correlations, identifying appropriate subpopulations or stratification of patients for a trial are of importance to characterise newly discovered disease, a new subgroup of patients etc.;
- Inclusion of patient and caregiver perspectives from the RD community is strongly encouraged. Patients living with a RD or a family member who cares for them, have experiences and knowledge that can contribute to generating data about the natural progression of the disease. Patients should be involved in all steps of planning and implementation of the study;
- For the small group of well-characterized rare diseases with approved treatments or improved standard of care, prospective studies can help to better define the altered disease progression under the current medical setting. Thereby, studies collecting data regarding adverse events and providing reference/data for development of a more effective or safer treatment can be considered for complementing the natural history study.

❖ 4.2 Excluded approaches and topics

The following approaches and topics will be excluded from the scope of the JTC2023:

- Interventional clinical trials to prove efficacy of drugs, treatments, surgical procedures, medical procedures. This also includes studies comparing efficacy, e.g. two surgical techniques or therapies. Projects whose main objective is the implementation of a clinical phase IV pharmacovigilance study cannot be funded either.
- Studies on the exclusive testing of the safety of medical devices and drugs.
- Development of new therapies as covered in EJP RD JTC 2020.
- Development of new analytic tools and pathways to accelerate diagnosis and facilitate diagnostic monitoring of rare diseases as covered in EJP RD JTC 2022.
- Projects focusing only on rare neurodegenerative diseases which are within the main focus of the Joint Programming Initiative on Neurodegenerative Disease Research (JPND). These are: Alzheimer's disease and other dementias; Parkinson's disease (PD) and PD-related disorders; Prion diseases; Motor Neuron Diseases; Huntington's disease; Spinal Muscular Atrophy and dominant forms of Spinocerebellar Ataxia. Interested researchers should refer to the relevant JPND calls. **However, childhood dementias/neurodegenerative diseases are not excluded.**
- Rare infectious diseases, rare cancers and rare adverse drug events in treatments of common diseases. **Rare diseases with a predisposition to cancer are not excluded.**

❖ 4.3 Project description

Applicants will describe and justify the following elements (**see section 3 – Project Description of the Guidelines for Applicants** for complete information on the content of pre and full proposal templates):

- Background, present state of the art in the research field
- Objectives and hypothesis
- Soundness and pertinence
- Workplan & methodology (*highlighting feasibility*)
- Impact
- Valorization, translation to practice
- PAOs engagement/involvement
- Ethical and legal issues, data management
- Work packages, timeline and budget
- Responsibilities and workloads

5. Funding and eligibility criteria

❖ 5.1 Funding

The maximum duration of the project is three years.

Double funding of research projects is not permitted. The JCS and national/regional funding organisations will perform cross-checks of submissions against other joint transnational (e.g. NEURON, JPND, ERA4Health, ERA PerMed etc.) and national calls.

Partners may not apply for funding for the same research activities in different calls. In addition, there can be no double funding for activities already funded by EC H2020 and Horizon Europe calls.

Consortia of [projects funded in previous Joint Transnational Calls of the EJP RD](#) or ERA-Net E-Rare can apply for funding for an extension of their cooperation. These consortia must clearly demonstrate the success of the current project and innovative scientific aims for their future collaboration. Their applications will compete with applications for new research projects.

❖ 5.2 Categories of partners

Partners belonging to one of the following categories may request funding under a joint research proposal (according to country/regional regulations):

- Academia (research teams working in universities, other higher education institutions or research institutes),
- Clinical/public health sector (research teams working in hospitals/public health and/or other health care settings and health organisations),
- Enterprises (all sizes of private companies). Participation of small and medium-sized enterprises (SMEs) is encouraged when allowed by national/regional regulations,
- Patient advocacy organisations (PAOs).

❖ 5.3 Countries and region-specific guidelines

Although applications will be submitted jointly by applicants from several countries, individual groups will be funded by their respective regional/national funding organisation. **Applicants therefore must contact their respective funding organisations and confirm eligibility in advance** of applying (see section 10). **The adherence to the national/regional regulations in the “Guidelines for Applicants” document is mandatory.** The inclusion of a non-eligible partner in a proposal will **lead to the rejection of the entire proposal without further review.** If you need additional information, please contact the JCS. Note that a parallel proposal submission is required by some regional/national funding organisations.

❖ 5.4 Consortium Makeup

The **use of the matchmaking tool is strongly encouraged** to build multidisciplinary research projects:

<https://connect.eventia.com/en/public/events/jtc2023matchmaking/registration/at-tendees>

5.4.1 Limit number of partners

Only transnational projects will be funded. Each consortium submitting a proposal must involve **four to six eligible principal investigator partners (referred to as partners below)** from **at least four different participating countries** (see list in section 2). In specific cases this can be increased to eight partners (see below). No more than two eligible partners from the same country can be present in each consortium; **further national/regional limits may apply, see “Guidelines for Applicants”**. PAOs requesting funding do not count toward the total.

The number of partners can be increased to 8 in two cases:

1. The inclusion of partners from participating countries usually underrepresented in projects (Hungary, Lithuania, Poland, Slovakia, Sweden and Türkiye).
2. The inclusion of Early Career Researchers as full partners (see section 5.6).

5.4.2 What is a partner? a collaborator? a sub-contractor?

In order to be **considered as an eligible partner**, a group must contribute substantially to at least one of the project's work packages. If the only role of a group is to provide patient access, data or samples for the study, they will not be considered as partners of the consortium, but can be included otherwise, via cooperation agreements or subcontracting.

Consortia may include **collaborators** that secure their own funding. Collaborators cannot be work package leaders, and their contribution to the consortium must be described (where relevant a CV can be included in the proposal). As they do not receive funding as part of this call, they do not count toward the limit of 8 partners requesting research funding (nor is there a limitation of collaborators per country, as long as their participation is justified). Collaborators must supply a letter of intent in the proposal.

If necessary, to implement the action, consortia may also include **sub-contractors, according to country/regional regulations**. Sub-contractors may cover only a limited part of the action, and their contribution to the consortium must be described. They do not count toward the limit of 8 partners requesting research funding (nor is there a limitation of subcontractors per country, as long as their participation is justified and if subcontracting is possible according to national/regional funding rules).

5.4.3 Consortium organisation

Each transnational proposal must nominate a **project consortium coordinator** among the project partner principal investigators. The coordinator must be an eligible project partner from an EJP RD JTC 2023 funding country/region. The project coordinator will **represent the consortium externally**, to the JCS and to CSC, and will be **responsible for its internal scientific management** (such as controlling, reporting, and intellectual property rights issues). This workload should be considered in the estimation of the budget of the coordinator. A single principal investigator will represent each project partner. Within a joint proposal, the principal investigator of each project partner will be the contact person for the relevant country/regional funding organisation.

❖ 5.5 Patient Advocacy Organisations and Patient Involvement

Consortia are strongly advised to include patient representatives and patient advocacy organisations (PAOs).

From an early stage in proposal development, applicants should consult relevant disease-specific patient organisations and/or alliances of rare disease patient organisations. For information on where to find patient representatives and PAOs willing to be involved in research, please see:

- Orphanet portal for rare diseases and drugs [patient organisation directory](#)
- Rare Diseases Europe ([EURORDIS](#))
- European Reference Networks ([ERNs](#))
- European Patient's Academy on Therapeutic Innovation ([EUPATI](#))
- European Patients' Forum ([EU-Patient](#))

The consortia will clearly present the role and responsibilities of the patient representatives and PAOs, how they will operate, at what levels and stages of the research, and provide justifications for allocated resources. It is highly encouraged that Patient representatives and PAOs are involved in all levels of the proposed work, including in project design, by advising on prioritization, sitting on advisory groups, being a member of the consortium steering group or the governance group. Patient representatives and PAOs may be part of institutional scientific boards to discuss the proposal and subsequent study on issues such as:

- the research idea, for relevance to patient concerns,
- possible outcomes,
- informed consent,
- patient input on appropriate outcome measures,
- possible patient intervention in the project,
- review of the data collected,
- dissemination of research findings,
- lay language summaries,
- review of data use conditions and access procedures within and outside of the project consortium, within and beyond the runtime of the project.

For more information on patient-centered care and strategies to involve patient representatives and PAOs in your research project, please consult:

- [EJP RD Short guide on patient partnerships in rare diseases research projects](#)
- [INVOLVE Briefing Notes for Researchers](#) and [cost calculator](#),
- [Recommendations for Successful Patient Involvement in Scientific Research](#) (de Witt et al., 2016),
- [Measuring what matters to rare disease patients](#) (Morel & Cano, 2017),
- [CIHR's Patient Engagement resources](#).

The funding conditions for the PAOs will be set out for each country in the guidelines for applicants.

❖ 5.6 Early Career Researchers

Early Career Researchers (ECRs) are encouraged to join consortia as full research partners and are therefore subject to the same eligibility criteria as other partners. ECRs must demonstrate independence and scientific excellence, and should be clearly identified in the proposal and their CV. A definition of ECRs according to European Research Council criteria is provided in the "Guidelines for Applicants", section 4.1. **Please note that national/regional definitions and time limits might differ.** Therefore, please refer to national guidelines and contact your national/regional funder. Please refer to the "Guidelines for Applicants", section 4.2 for requirements for the identification of ECRs.

6. Registration and submission

Research consortia who intend to submit a transnational project proposal should **register as soon as possible** via the electronic proposal system: <https://ptoutline.eu/app/ejprd23>. Please fill in the data sheet in the system. The same data sheet can be used for the submission of pre-proposals and full proposals (if invited).

There will be a **two-stage submission procedure for joint applications**: a pre- and full proposal stage. In both cases, one joint proposal document (in English) shall be prepared by the partners of a joint transnational proposal and must be submitted by the coordinator only to the JCS via the electronic submission system. Proposals must

be prepared using the templates provided on the EJP RD web page (www.ejprarediseases.org). Proposals not conforming to template instructions (including length and format) will be rejected.

Call Timeline

12 th December 2022	Launch of the call
15 th December 2022	Information webinar for potential applicants
15 th February 2023	Pre-proposal submission deadline
End of April 2023	Invitation to full proposal
May 2 nd 2023	Information webinar for applicants invited to submit a full-proposal
14 th June 2023	Full proposal submission deadline
26 th July 2023	Deadline for rebuttals
December 2023	Notification of funding decision

Full proposals will be accepted only from those applicants who were explicitly invited by the JCS to submit them.

In general, no fundamental changes between the pre- and full proposals concerning the composition of the consortia, objectives of the project, or requested budget will be accepted. In order to make such a change, a detailed justification must be provided to the JCS for consideration by the CSC. One justification can be that because of additional advice gathered on the feasibility of the project, additional expertise or resources are needed. However, the national/regional regulations on budget caps will still apply and the budget change needs to be pre-approved by relevant national/regional funding organisation(s)

Further information on how to submit pre-proposals and full proposals electronically (including Guidelines for Applicants and submission templates) is available at the EJP RD website (<http://www.ejprarediseases.org/>).

7. Evaluation process

At the pre-proposal stage, applicants should focus on presenting the scientific idea/hypothesis and supporting preliminary results, studies or data. The proposal should describe the project, starting from an unmet need, and follow through to the expected end-point of the study.

At the full proposal stage, in addition to the scientific content, a full description of patient engagement (or a justification if this is not applicable), data management, statistical methods, and ethical and legal issues will be required in compliance with [EC requirements](#). Applicants should anticipate this requirement and ensure that they have consulted with relevant experts to verify the feasibility of the project, and that the proposal can be completed within the defined budget (taking into account budget limits listed in the Guidelines for Applicants).

❖ 7.1 Evaluation Criteria

Evaluation scores will be awarded according to specific evaluation criteria that are in line with Horizon 2020 rules (see below), using a common evaluation form. Each criterion will be scored out of five, for a maximum overall score of 15 points. The threshold for an individual criterion is three, with an overall threshold of 12 points.

7.1.1 Scoring system

0: Failure: The proposal fails to address the criterion in question or cannot be judged because of missing or incomplete information.

1: Poor: The proposal shows serious weaknesses in relation to the criterion in question.

2: Fair: The proposal generally addresses the criterion, but there are significant weaknesses that need corrections.

3: Good: The proposal addresses the criterion in question well, but certain improvements are necessary.

4: Very good: The proposal addresses the criterion very well, but small improvements are possible.

5: Excellent: The proposal successfully addresses all aspects of the criterion in question.

7.1.2 Criteria

1. Excellence (0-5)

- a. Clarity and pertinence of the objectives,
- b. Credibility of the proposed approach and methodology,
- c. Soundness of the concept,
- d. Innovative potential,
- e. Feasibility of the project (adequate requested resources, time schedule, access to and engagement of patients, data and material),
- f. Competence and experience of participating research partners in the field(s) of the proposal (previous work in the field, specific expertise),
- g. Active and meaningful participation of PAOs and patient representatives in the project (including where possible in the design and definition of research priorities, interpretation and implementation of results, their dissemination, and communication).

2. Impact (0-5)

- a. *Potential of the expected results for exploitation and for future clinical, public health and/or other socio-economic health relevant applications, including patient's needs,
- b. *Benefit to patients, their families, and carers with an active and meaningful involvement of patient organisations and patient representatives,
- c. *Added value of transnational collaboration: gathering a critical mass of patients/ material, sharing of expertise and resources, harmonisation of data, sharing of specific know-how and/or innovative solutions,
- d. **Effectiveness of the proposed measures to exploit and disseminate the project results (including management of IPR), to communicate the project, and to manage research data. A data management strategy in the full proposal is mandatory,
- e. Inclusion of Early Career Researchers as full partners (PI/research team leaders),
- f. Involvement of industry (for a socio-economic impact and/or an acceleration of research development, access to innovation and application for patients).

3. Quality and efficiency of the implementation (0-5)

- a. Coherence and effectiveness of the work plan, including appropriateness of the allocation of tasks, resources and timeframe,
- b. Complementarity of the participants within the consortium, including the integration of PAOs or patient representatives where possible,

- c. ******Appropriateness of the management structures and procedures, including risk management, contingency plans and innovation management,
- d. ******Plan for sustainability of infrastructures or resources initiated by the project,
- e. ******Budget and cost-effectiveness of the project (rational distribution of resources in relation to project's activities, partner responsibilities, and time frame).

*Sub-criteria 2a, 2b and 2c will be prioritized for assessing the impact of proposals (pre- and full proposal stage).

**Sub-criteria 2d, 2e, 3c, 3d and 3e will be only assessed as part of the full proposal evaluation step.

❖ 7.2 Pre-proposal Review

Eligibility check

The JCS will check all pre-proposals to ensure that they meet the call's formal criteria. The JCS will forward the proposals to the CSC members who will perform a check for compliance to country/regional eligibility rules. **Please note that proposals not meeting the formal criteria or the national/regional eligibility criteria and requirements will be declined without further review.**

Peer review of pre-proposals

Pre-proposals passing the eligibility check will be forwarded to the SEC members for a first evaluation (see evaluation criteria above). The SEC members will perform the assessment of the pre-proposal and fill the evaluation forms with scores and comments for each criterion. Each pre-proposal will be assessed by 2 SEC members. The SEC members will then meet to establish a ranking of the pre-proposals. This ranking will be used by the CSC to decide which pre-proposals will be accepted for full proposal submission. General recommendations from the SEC will be forwarded to applicants invited for the second step of the evaluation process. The summary review report will only be forwarded to applicants not invited for the second step.

Widening

At this stage research teams of underrepresented or undersubscribed countries may join successful pre-proposals (see 5.2 in Guidelines for Applicants for more details).

EJP RD Clinical support services

Applicants that are invited to submit a second stage proposal are strongly encouraged to make use of the EJP Rare Diseases [Clinical Support Services](#) (see 5.3 in the Guidelines for Applicants for more details).

❖ 7.3 Full proposal Review

Formal criteria check

The JCS will check the full proposals to ensure that they meet the call's formal criteria.

External reviewer evaluation

Each proposal will be allocated to at least two external scientific reviewers with expertise relevant to the application.

Rebuttal stage

Before the SEC members see the reviews from external reviewers, each project coordinator will be provided with the opportunity to read and provide a written

response to the evaluations of the external reviewers. The scores will not be given at this stage. This step allows applicants to correct factual errors or misunderstandings in the review, and to reply to reviewers' questions. Issues which are not related with reviewers' comments cannot be addressed and the work plan cannot be modified at this stage.

The applicants will have up to one week (in late July 2023) for this **optional** response to the reviewers' comments.

SEC Meeting Evaluation

The JCS will send full proposals, reviews and rebuttals to the SEC members. The SEC will meet to discuss each proposal and, **after consideration of the evaluation criteria, external reviews, rebuttals, and their own reviews and discussions**, the SEC will assign final scores, make a classification of the proposals, and rank proposals recommended for funding. The final summary review report prepared by the SEC members will be sent to all applicants.

Two additional groups of reviewers will be present at the second SEC meeting to evaluate projects:

1. Patient representatives

Proposals will be evaluated by expert patient reviewers according to the relevant evaluation criteria listed above (subcriteria 1g, 2b, 3b; see section 7.1.2) with a 3-levels scoring system. These reviewers will be present at the SEC meeting to discuss proposals and provide their feedback.

2. Statistical / methodological experts

Proposals will be evaluated by experts in methodology or statistics according to the relevant evaluation criteria listed above (subcriteria 1b, 1f, 1g; see section 7.1.2) with a 3-levels scoring system. These reviewers will be present at the SEC meeting to discuss proposals and provide their feedback.

Each proposal will be evaluated by 3 biomedical experts, one patient expert and one statistical/methodological expert.

Ethical evaluation

After the second SEC meeting, full proposals recommended for funding by the SEC will be remotely evaluated by independent experts in ethics. These experts will report on the feasibility of a given proposal to comply with the ethical requirements. If necessary, it will list those tasks that need to be done and documents that need to be submitted by the consortium in order to receive approval for funding from an ethics standpoint.

Only those proposals approved by both the scientific and ethical evaluations (complying with all central Horizon 2020 and regional/national ethical requirements), will be funded.

❖ 7.4 Funding decision

Based on the ranking list established by the SEC and on available funding, the CSC will suggest the projects to be funded to the national/regional funding organisations. Final decisions will be made by the national/regional funding organisations and will be subject to budgetary considerations.

If necessary, the CSC will determine a priority order for proposals which have been awarded the same score within a ranked list. This will be based on (in descending order):

- Availability of national/regional funding;
- Maximization of use of national/regional funding;

- Proposals with participation of underrepresented or undersubscribed countries;
- Proposals that address diseases not otherwise covered by more highly ranked proposals;
- Proposals with meaningful engagement/involvement of PAOs;
- Gender balance within the consortium's partners in order to reduce any inequalities.

The JCS will notify all project coordinators of the final funding decision and disseminate the SEC consensus report.

8. Responsibilities, Reporting requirements and Dissemination

The Joint Call Secretariat (JCS) is the French National Agency for research (ANR, France) to assist the CSC and the national/regional funding bodies during the implementation of the call. The JCS will be responsible for the administrative management of the call. It will be the primary contact point between the research consortia, the funding organisations, and peer reviewers with regard to call procedures. **The project coordinator is the point of contact for consortium during the application procedure and is responsible for forwarding relevant information from the JCS to their consortium members.** CSO-MOH, Israel, will be responsible for the monitoring phase until the funded research projects have ended.

The **coordinators** of all funded projects must submit an **annual scientific project report and a final scientific project report** (due within six months of the end of the project). All reports must be in English and must use the reporting templates provided. The research partners are jointly responsible for delivery of the reports. Only reports delivered on behalf of the consortium, via the project coordinator, will be accepted.

If required, each beneficiary should submit financial and scientific reports to their **national/regional funding organisations**, according to national/regional regulations. The progress and final results of each individual contract/letter of grant will be monitored by the respective national/regional funding organisations.

The coordinators and national/regional group leaders will be asked to present the results of their projects at an **intermediate status symposium** organized by EJP RD. The presence of at least one representative (coordinator and/or partner) per project will be mandatory. Therefore, **the coordinator and respective partners must budget a sufficient amount for the expenses related to these events.**

Please read the “Guidelines for Applicants” document for further information including national/regional information and eligibility requirements.

9. Contacts and further information

Further information on the EJP RD, the Call, and follow-up is available at the EJP RD website (<http://www.ejprarediseases.org/>).

Call Contacts

Role	Organisation	Contact Details
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Joint Call Secretariat	ANR	Florence Guillot Camille de Almeida Ingrid Pfeifer EJPRDcall@anr.fr
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10. National and regional contacts

Applicants should refer to the guidelines document for country-specific information including national/regional rules that may apply. Applicants are strongly advised to contact the national/regional contact person to ensure eligibility before submitting their projects.

Country/Region	Funding Organisation	Contact Details
Belgium	Fund for Scientific Research – FNRS (F.R.S.-FNRS)	Dr. Florence Quist +32 (0)2 504 9351 Joël Groeneveld +32 (0)2 504 9270 International@frs-fnrs.be
Canada	Canadian Institutes of Health Research – Institute of Genetics (CIHR-IG) www.cihr-irsc.gc.ca	Jennifer Vineham jennifer.vineham@cihr-irsc.gc.ca +1 343 552-2760
France	The French National Agency for Research - ANR	Dr. Florence Guillot Dr. Camille de Almeida Dr. Ingrid Pfeifer EJPRDcall@anr.fr
Germany	Federal Ministry of Education and Research (BMBF) / Project Management Agency of the German Aerospace Centre (BMBF/ PT-DLR)	Dr. Katarzyna Saedler Dr. Michaela Fersch Dr. Ralph Schuster +49228-38212453 SelteneErkrankungen@dlr.de
Germany	German Research Foundation (DFG)	Dr. Katja Hartig Dr. Annika Ottersbach Julia Höller +49228-885 2044

Hungary	National Research, Development and Innovation Office (NKFIH)	Dr. Előd Nemerkenyi Phone: +36 1 8963987 elod.nemerkenyi@nkfi.gov.hu Dr. Gábor Tóth Phone: +36 1 8961727 gabor.toth@nkfi.gov.hu
Ireland	Health Research Board (HRB)	Amanda Daly Amanda.Daly@hrb.ie
Israel	Chief Scientist Office Israeli Ministry of Health (CSO-MOH)	Dr. Irit Allon IRIT.ALLON@moh.gov.il +972-2-5082167
Italy	Italian Ministry of Health, IT-MOH	Chiara Ciccarelli +39 06 5994 3919 c.ciccarelli@sanita.it Simona Bifulchi s.bifulchi@sanita.it
Italy	Fondazione Telethon (FTELE)	Carmen Fotino +39 02 202217526 cfotino@telethon.it
Lombardy (Italy)	Regional Foundation for Biomedical Research (FRRB)	General inquiries: bandi@frrb.it Paola Bello Tel: + 39 02 67650174 paola.bello@frrb.it Marcello De Amico Tel: + 39 02 67650186 marcello.deamico@frrb.it
Italy	Tuscany Region	Donatella Tanini Phone: +39 055 4383256 Teresa Vieri Phone: +39 055 4383289 ejprare@regione.toscana.it
Lithuania	Research Council of Lithuania	Dr. Živilė Ruželė zivile.ruzele@lmt.lt (+370) 676 14383
Luxembourg	Luxembourg National Research Fund (FNR)	Dr. Sean Sapcariu sean.sapcariu@fnr.lu +352 691 362

Poland	National Centre for Research and Development	Dr Marcin Chmielewski marcin.chmielewski@ncbr.gov.pl (+48) 0 22 39 07 109, (+48) 571 226 666
Slovakia	Slovak Academy of Sciences (SAS)	Dr. Silvia Kecerova kecerova@up.upsav.sk +421 (0) 2 5751 0 118
Spain	National Institute of Health Carlos III	Mauricio Garcia-Franco mauriciog@isciii.es +34 91822 2885
Sweden	Swedish Research Council (SRC)	Louise Rügheimer Louise.rugheimer@vr.se
Sweden	Vinnova	Gunnar Sandberg Gunnar.sandberg@vinnova.se +46 8 4546445
Switzerland	Swiss National Science Foundation (SNSF)	Dr. Tobias Braun tobias.braun@snf.ch +41 31 308 21 67
Türkiye	The Scientific and Technological Research Council of Türkiye (TUBITAK)	Dr. Emine Derebay Yıldız emine.derebay@tubitak.gov.tr +90(312)298 1195