



Call for Proposals 2020
**"PRE-CLINICAL RESEARCH TO DEVELOP EFFECTIVE THERAPIES
FOR RARE DISEASES"**

Call Text

**Submission deadline for pre-proposals: 2 p.m. (CET), February 18th,
2020**

For further information, please visit us on the web:

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

Or contact:

Joint Call Secretariat (ANR, France)

EJPRDcall@anr.fr

Florence Guillot

florence.guillot@agencerecherche.fr

+33 (0) 1 78 09 80 01

Kiri Couchman

kiri.couchman@agencerecherche.fr

+33 (0) 1 78 09 81 29

Table of contents

1. Background	3
2. Participating Organisations	3
2.1. Management and Evaluation Structures	4
3. Aim of the Call	5
4. Application	9
4.1. Eligibility	9
4.2. Country and Region-Specific Guidelines	10
4.3. Consortium Makeup	10
4.4. Patient Advocacy Organisations and Patient Involvement	11
4.5. Early Career Researchers	12
5. Registration and Submission	13
6. Evaluation Process	13
6.1. Evaluation Criteria	14
6.2. Pre-proposal Review	15
6.3. Full proposal Review	16
6.4. Funding decision	17
7. Responsibilities, Reporting Requirements, and Dissemination	18
8. Contact and Further Information	18
9. National/Regional Contacts	19

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 and better care of 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 standardised, (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 ERA-Net E-Rare has successfully implemented ten Joint Transnational Calls for rare disease research projects since 2006. This effort is now continued in the frame of the **European Joint Programme on Rare Diseases (EJP RD)** that has been established 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).

2. Participating Organisations

A number of national and regional funding organisations will participate in the **EJP RD Joint Transnational Call (JTC) 2020** and will fund multilateral research projects on rare diseases **together with the European Commission (EC) under the EJP-COFUND action**. The call opens simultaneously with the involvement of the following funding organisations in their respective countries/regions:

- Austrian Science Fund (FWF), Austria
- Research Foundation Flanders (FWO), Belgium, Flanders
- Fund for Scientific Research - FNRS (F.R.S.-FNRS), Belgium, French-speaking community

- Canadian Institutes of Health Research – Institute of Genetics (CIHR-IG), Canada
- Fonds de recherche du Québec-Santé (FRQS), Québec (Canada)
- Ministry of Education, Youth and Sports (MEYS), Czech Republic
- Academy of Finland (AKA), Finland
- French National Research Agency (ANR), France
- French Foundation for Rare Diseases (FFRD), France
- INSERM, France (Patient Advocacy Organisations)
- **Decision Pending:** Federal Ministry of Education and Research (BMBF), Germany
- **Decision Pending:** German Research Foundation (DFG), Germany
- General Secretariat for Research and Technology (GSRT), Ministry of Development & Investments, Greece
- 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 (MoH-IT), Italy
- Ministry of Education, Universities and Research (MIUR), Italy
- Fondazione Regionale per la Ricerca Biomedica (FRRB) Lombardy, Italy
- Tuscany Region (RT/TuscReg), Tuscany (Italy)
- Research Council of Lithuania (RCL), Lithuania
- National Research Fund (FNR), Luxembourg
- National Centre for Research and Development (NCBR), Poland
- **Decision Pending:** The Foundation for Science and Technology (FCT), Portugal
- Slovak Academy of Sciences (SAS), Slovakia
- National Institute of Health Carlos III (ISCIII), Spain
- Swedish Research Council (SRC), Sweden
- Swiss National Science Foundation (SNSF), Switzerland
- Netherlands Organization for Health Research and Development (ZonMw), The Netherlands
- The Scientific and Technological Research Council of Turkey (TUBITAK), Turkey

2.1. 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 and the final selection and award of research projects.

The Call Steering Committee (CSC) is composed of a single representative from each country/region funding organisation that joins the JTC2020. 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 recognised, 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.

3. 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 a clear future benefit for patients.

Topic: PRE-CLINICAL RESEARCH TO DEVELOP EFFECTIVE THERAPIES FOR RARE DISEASES

Research proposals must cover at least one of the following areas:

1. Development of novel therapies in a preclinical setting (including small molecules, repurposing drugs, cell and gene advanced therapies) focusing on condition(s) with unmet medical needs
2. Use of disease models suitable for medicinal product's development according to [EMA guidelines](#)
3. Development of predictive and pharmacodynamics (PD) biomarkers (with appropriate analytical methods e.g. OMICS) in a preclinical setting (e.g. in the validated model or in pre-collected human samples) for monitoring the efficiency of the therapy. The model chosen must mimic the human diseases and be transposable so that the biomarker identified in animals can be valid for humans
4. Proof of principle studies fostering an early (pre-clinical) stage of drug development (excluding interventional clinical trials of phase 1-4).

The following approaches and topics are excluded from the scope of the call:

- a) Therapeutic approaches concerning rare infectious diseases, rare cancers and rare adverse drug events in treatments of common diseases
- b) Interventional clinical trials
- c) Surgery or radiation therapies
- d) Studies that focus on research to accelerate diagnosis or to set up new registry/cohort studies to explore disease progression and mechanisms as these were the focus of JTC 2019.
- e) Rare neurodegenerative diseases which are within the main focus of the Joint Programming Initiative on Neurodegenerative Disease Research (JPND; <http://www.neurodegenerationresearch.eu/>). These are: Alzheimer's disease

and other dementias; Parkinson's disease (PD) and PD-related disorders; Prion disease; Motor Neuron Diseases; Huntington's disease; Spinal Muscular Atrophy and dominant forms of Spinocerebellar Ataxia. Interested researchers should refer to the relevant JPND calls.

Childhood dementias/neurodegenerative diseases are not excluded.

Projects shall involve **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 solid criteria and commonalities if this leverages added value in sharing resources or expertise and has the capacity to elucidate common disease mechanisms and therapeutic targets.

It is highly encouraged that the research will focus on diseases without approved treatment options to contribute to the aims of IRDiRC in this area (for information see [list of EMA approved orphan medical products](#)).

Translatability into humans should be one of the key focuses of the project, and applicants should demonstrate access to relevant scientific or regulatory expertise (e.g. through innovation task forces or competent national authorities).

Consortia performing preclinical development of therapeutics are strongly advised to engage or consult experts in the various stages of product development, with the aim to establish one or more of the following:

- a) **Target validity:** Strong link between target and disease, differentiated efficacy, available and predictive biomarkers.
- b) **Right Tissue:** Adequate bioavailability and tissue exposure, definition of pharmacodynamics biomarkers, clear understanding of preclinical pharmacokinetics.
- c) **Right safety profile:** Differentiated and clear safety margins (in models), understanding of secondary pharmacology risk which consist in evaluating the potential off-target or unintentional effects of a drug, including understanding of reactive metabolites, genotoxicity, drug–drug interactions, and off-target liability. These studies are important in predicting potential toxicities and demonstrating safety of a therapy.
- d) **Right patient:** Identification of the most responsive patient population, with a risk–benefit analysis.

For the development of novel therapies or proof-of-principle studies, the following issues should be addressed in the proposal:

- Orphan drug designation (ODD) planning: has an ODD been granted? If not, the path to ODD development should be described (including target product profile for therapy development).
- Exploration of scale-up feasibility for clinical trials and manufacturing.

- For projects developing a new target (not extensively validated in the literature), target revalidation in preclinical models should be a first step in project.

For validation or development of predictive and pharmacodynamics biomarkers (predictive biomarkers are important to help guide patient selection, pharmacodynamics biomarkers can provide information on the pharmacologic effects of a drug on its target), the following issues should be addressed in the proposal:

- Ensure in the first stage that the biomarker (signature) undergoes analytical validation using high quality samples from an independent collection (different from the collection in which the signal was discovered), which have been collected and stored under quality controlled conditions and following international standards.

Samples used in validation should be sourced from certified biobanks (e.g. <http://www.eurobiobank.org/>). Upon sample provision biobanks should provide a report including information on:

- Identification and specific properties of the materials
- Relevant quality information of the materials and clinical data
- Method used for identification and characterisation of materials
- Method used for testing of the materials
- Method used for sample collection, preparation, preservation, storage
- Accreditation of the lab performing the analytical validation of the biomarker for the method used (e.g. ISO 17025 or 15189).
- Validation should follow a risk-based approach wherein depending on potential confounding variables such as genetic diversity, multiple biobanks from multiple regions may be utilised. Sample size and number should reflect such risk.

Applicants should **describe and justify the use of any disease models** (animal or otherwise) described in the proposal:

- Describe how the model replicates the pathology or human condition (aetiology, pathophysiology, symptomatology and response to therapeutic intervention),
- Whether the model duplicates aspects of the therapy target including expression, distribution and primary structure, pharmacodynamics, metabolism and other pharmacokinetic aspects,
- If the project involves the use of animals, provide sound scientific justification for their use, explain why there are no realistic alternatives, and demonstrate that the numbers proposed will allow meaningful results to be obtained from the research. Please also specify the sex of the animals, and rationale for the numbers of each sex,
- Describe how the proposed pre-clinical work correlates and aligns with any planned future stages of the research in humans.

Furthermore, the following **additional elements need to be considered in all proposals**:

- The design of the study (sample collection, statistical power, interpretation, relevant models for hypothesis validation) must be well justified and should be part of the proposal.
- Appropriate bioinformatics and statistical methods should constitute, whenever justified, an integral part of the proposal, and the relevant personnel should be clearly specified.
- Preliminary data should be described in a manner that would allow a skilled peer to replicate the data, including positive and negative controls, and suitable n values for statistical analysis. All data points should be included in the analysis and presented with error bars where relevant.
- Risk management should be considered including the identification of possible bottlenecks and go/no go contingencies.
- Feasibility of the project given requested resources (budget) and schedule must be demonstrated: timelines should be realistic, and lead times should be accounted for (e.g. regulatory or scientific advice).
- If relevant, the consortium will identify technology transfer officer responsible for intellectual property management. Project plan should include innovation management activities (e.g. ongoing monitoring, expert panels to identify high potential results), and may describe follow-on funding and/or draft study plans past the grant end (e.g. natural history studies with relevant stakeholders including patient groups, or approaching companies for potential in-licensing or co-development).
- Applicants should include information about other ongoing development work on the target/indication, and explain why their approach should be supported.
- Study design and preclinical models (vectors, reagents etc.) may be selected to facilitate approval in human trials and future clinical grade manufacturing.
- To make research data findable, accessible, interoperable and re-usable (FAIR), a data management strategy for the proposed project is mandatory in the full proposal stage. Some countries involved in this call will also require a data management plan at the full proposal stage or upon granting of the project.
- To ensure that the needs and priorities of rare disease patients are adequately addressed, they or their representatives must be appropriately involved in all projects wherever possible (see section 4.4).

The use of **existing European health research infrastructures** and/or **IRDiRC recognized resources** is strongly encouraged when appropriate: e.g. research infrastructures established as a European Research Infrastructure Consortium (ERIC) or identified on the roadmap of the European Strategy Forum on Research Infrastructures (ESFRI). Projects are invited to identify the existing European research data infrastructures that may be used and how these may be mobilised, in particular for long-term data curation and preservation (in accordance with EU and [IRDiRC recommendations](#)).

The following ESFRI European Research Infrastructures and European/international projects or their results may be of use to consortia:

- [BBMRI](#) Biobanking and Biomolecular Resources Research Infrastructure
- [ELIXIR](#) The European Life Sciences Infrastructure for Biological Information
- [INFRAFRONTIER](#) European Infrastructure for Phenotyping, Archiving and Distribution of Mouse Models
- [INSTRUCT](#) Integrated Structural Biology Infrastructure for Europe
- [ECRIN](#) European Clinical Research Infrastructure Network
- [EATRIS](#) European Infrastructure for Translational Medicine
- [EU-OPENSREEN](#) European high-capacity screening network
- [RD-Connect](#) An integrated platform connecting databases, registries, biobanks and clinical bioinformatics for rare disease research
- [Matchmaker Exchange](#) A federated platform to facilitate the matching of cases with similar phenotypic and genotypic profiles
- [IRDiRC recognized resources](#)
- [Orphanet Rare Disease Ontology](#)
- [Human Phenotype Ontology](#)
- [Horizon 2020 FAIR Data Management Plan](#) Annex 1 in:
- [Recommendations for Improving the Quality of Rare Disease Registries](#)

The aim of the call is 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](#).

4. Application

4.1. Eligibility

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 are eligible to obtain funding for their participation in research projects; see section 4.4).

The maximum duration of the project is three years.

4.2. Country 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 submitting an application. **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.

4.3. Consortium Makeup

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 limits may apply, see “Guidelines for Applicants”). This limit also applies to early career researchers and partners from underrepresented and undersubscribed countries (see below). PAOs requesting funding do not count toward this total.

In order to be considered as an eligible partner, each partner (with their respective research groups) must contribute substantially to at least one of the project work packages. If the only role of a group is to provide patient 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 research project must be described (where relevant a CV should 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. There is no limit on the number of collaborators per country, however, the added value of the collaboration must be clearly described and the number of collaborators must remain manageable within the limits of the project.

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 2020 funding country/region. The project coordinator will represent the consortium externally and to the JCS and CSC, and will be responsible for its internal scientific management (such as controlling, reporting, and intellectual

property rights issues). This workload should be taken into account 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.

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

1. The inclusion of partners from participating countries usually underrepresented in projects (Czech Republic, Slovakia, Hungary, Lithuania, Poland, and Turkey).
2. The inclusion of Early Career Researchers as full partners (see section 4.5).

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, EuroNanoMed, ERA PerMed etc.) and national calls. Partners may not apply for funding for the same research activities in different calls.

Consortia of projects funded in previous Joint Transnational Calls of the 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.

4.4. Patient Advocacy Organisations and Patient Involvement

Consortia are strongly advised to include patient representatives and patient advocacy organizations (PAOs), which are **eligible to receive funding for their activities. If patient involvement is not deemed appropriate within a research project, this should be explained and justified.** The included PAOs will not count towards partner limits, and therefore their inclusion does not influence the partner restriction criteria described above. If there is no PAO for a specific rare disease, the consortia could investigate whether an umbrella PAO or a PAO for a similar rare disease may be involved.

The consortia should clearly present the role and responsibilities of the PAO, how they will operate, at what levels and stages of the research, and provide justifications for allocated resources. PAOs can be involved in all levels of the proposed work, including in project design, by advising on prioritisation, sitting on advisory groups, being a member of the consortium steering group or the governance group of a registry. 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, especially patient reported outcome measures,
- informed consent,

- patient input on appropriate clinical outcome measures,
- possible patient intervention in the project,
- review of the data collected,
- dissemination of research findings.

Consortia should also consider training of PAOs and representatives on biomedical knowledge via the attendance of international congress or via specific programs organized for instance by [Eurordis](#).

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

- [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](#).

From an early stage in proposal development, applicants should consult relevant disease-specific patient organisations when possible 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](#)).

Funding for PAOs is limited to a total of 50,000 € over 3 years and per project regardless of the number of participating PAOs (see "Guidelines for Applicants" for eligibility rules). Besides this funding, PAOs can also be involved through national/regional funding or subcontracting depending on the proposed tasks and national/regional funding rules.

4.5. 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. Further information including a definition of ECRs according to European Research Council criteria is provided in the "Guidelines for Applicants" (section 3.1). **Please note that national/regional definitions and time limits might differ.** Therefore, please refer to national guidelines and contact your national/regional funder. Please see the "Guidelines for Applicants" document for further information.

5. 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/ejprd20>. 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. The proposals must strictly follow the instructions in the proposal form.

Call Timeline

18 th February 2020	Pre-proposal submission deadline
End of April 2020	Invitation to full proposal
16 th June 2020	Full proposal submission deadline
28 th July 2020	Deadline for rebuttals
November 2020	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 translatability 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 the national/regional funding organisation.

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

6. Evaluation Process

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

end-point of the study (e.g. proof of principle in a preclinical study). Pre-proposals will be evaluated by scientific/clinical experts.

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. 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).

6.1. 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.

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.

1. Excellence (0-5)

- a. Clarity and pertinence of the objectives,
- b. Credibility of the proposed approach and methodology,
- c. Soundness of the concept (supporting data should be robust),
- d. Innovative potential: description of existing development landscape, relationships with technology transfer offices, plan for ongoing development,
- e. Feasibility of the project (adequate requested resources, time schedule, access to and engagement of patients, data, and material, translatability of medicinal products to patient treatment),
- f. Competence and experience of participating research partners in the field(s) of the proposal (previous work in the field, specific technical 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 commercial exploitation and for future clinical, public health and/or other socio-economic health relevant applications, and preferably for diseases without approved treatment options,
- b. *Added-value of transnational collaboration: gathering a critical mass of patients/biological material, sharing of expertise and resources (models, databases, diagnosis, etc.), and harmonization of data,
- c. Involvement of industry (when appropriate/applicable/available),
- d. Inclusion of Early Career Researchers as partners,
- e. **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,
- f. **Benefit to patients, their families, and carers developed through the involvement of patient organisations and patient representatives where possible,

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 time-frame,
- b. Complementarity of the participants within the consortium, including the integration of PAOs 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 and 2b will be prioritized for assessing the impact of proposals (pre- and full proposal stage).

**Sub-criteria 2c, 3c, 3d and 3e will be taken into account only for the full proposal evaluation step.

6.2. 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/PAO 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 to ensure it falls within the scope of the call, 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. The summary review report and eventual recommendations of the SEC will be forwarded to all applicants.

At this stage research teams of underrepresented or undersubscribed countries may join successful pre-proposals (see 3.2 in Guidelines for Applicants for more details).

6.3. 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 2020) for this **optional** response to the reviewers' comments.

SEC Meeting Evaluation

Four groups of reviewers will be present at the SEC meeting to evaluate projects:

1. SEC 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 discussions**, the SEC will assign final scores (taking into account patient reviewer comments), 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.

2. Patient reviewer evaluation

Proposals will be evaluated by expert patient reviewers according to the evaluation criteria listed above (see section 6.1). These reviewers will be present at the SEC meeting to discuss proposals and provide their feedback on the scores from SEC members for ranking of the proposals.

3. Statistical evaluation

Proposals will be evaluated by experts in biostatistics. These reviewers will not provide a score for the proposals, but will be there to assist in evaluating the feasibility of the projects with respect to bio-statistical methods.

4. Ethical evaluation

After the second SEC meeting, full proposals 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.

6.4. 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:

- Availability of national funding;
- Maximization of use of national funding;
- Proposals with participation of underrepresented or undersubscribed countries;
- Proposals that address diseases not otherwise covered by more highly-ranked proposals.

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

7. Responsibilities, Reporting Requirements, and Dissemination

The Joint Call Secretariat (JCS) is located at the French National Research Agency (ANR) 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 consortia 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 a **brief annual scientific project report** (due on the 28th of February 2022 and subsequent years) **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.

8. Contact 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
------	--------------	-----------------

Joint Call Secretariat	ANR (France)	EJPRDcall@anr.fr Florence Guillot Phone: +33 (0) 1 78 09 80 01 florence.guillot@agencerecherche.fr Kiri Couchman Phone: +33 (0) 1 78 09 81 29 kiri.couchman@agencerecherche.fr
Multinational, for funding of PAO	INSERM (France)	Coordination EJP RD pao@ejprarediseases.org
Monitoring	CSO-MOH (Israel)	Irit Allon Phone: +972-2-5082167 irit.allon@moh.health.gov.il
	FNRS (Belgium)	Florence Quist Phone: +32 2 504 93 51 florence.quist@frs-fnrs.be

9. National/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
Austria	Austrian Science Fund (FWF) www.fwf.ac.at	Stephanie Resch Phone: +43 (1) 505 67 40-8201 mailto:stephanie.resch@fwf.ac.at Anita Stürtz Phone: +43 (1) 505 67 40-8206 mailto:anita.stuertz@fwf.ac.at

Belgium: Flanders	Research Foundation Flanders (FWO) www.fwo.be	Alain Deleener Phone: +32 2 550 15 95 mailto:eranel@fwo.be Toon Monbaliu Phone: +32 2 550 15 70 mailto:eranel@fwo.be
Belgium: French- speaking community	Fund for Scientific Research - FNRS (F.R.S.-FNRS) www.frs-fnrs.be/	Florence Quist Phone: +32 2 504 93 51 mailto:florence.quist@frs-fnrs.be Joël Groeneveld Phone: +32 2 504 92 70 mailto:joel.groeneveld@frs-fnrs.be
Canada	Canadian Institutes of Health Research – Institute of Genetics (CIHR-IG) www.cihr-irsc.gc.ca	Jennifer Vineham Phone: +1 613 941-0796 mailto:jennifer.vineham@cihr-irsc.gc.ca
Canada: Québec	Fonds de recherche du Québec-Santé (FRQS) www.frqs.gouv.qc.ca	Maxime Beaudoin Phone: +1 514 873 2114, ext 1369 mailto:maxime.beaudoin@frq.gouv.qc.ca
Czech Republic	Ministry of Education, Youth and Sports (MEYS) www.msmt.cz	Judita Klosaková (MSMT) Phone: +420 234 811 504 mailto:judita.klosakova@msmt.cz
Finland	Academy of Finland (AKA) www.aka.fi	Heikki Vilen Phone: +358 29 5335 135 mailto:heikki.vilen@aka.fi
France	French National Research Agency (ANR) www.agence-nationale- recherche.fr	Florence Guillot Phone: + 33 (0)1 78 09 80 01 Kiri Couchman Phone: + 33 (0)1 78 09 81 29 mailto:EJPRDcall@anr.fr
France	French Foundation for Rare Diseases (FFRD) https://fondationmaladiesra res.org/eng/	Ingrid Zwaenepoel Phone : + 33 (0) 1 58 14 22 85 Diana Désir-Parseille Phone : + 33 (0) 1 58 14 22 81 mailto:aap-bio@fondation- maladiesrares.com
Germany	Federal Ministry of Education and Research (BMBF) / Project	Dr. Katarzyna Saedler Phone: +49 (0)228 3821 1947

	<p>Management Agency of the German Aerospace Centre (BMBF/ PT-DLR) www.gesundheitsforschung-ung-bmbf.de</p>	<p>mailto:Katarzyna.Saedler@dlr.de</p> <p>Dr. Michaela Fersch Phone: +49 (0)228 3821 1268 mailto:Michaela.Fersch@dlr.de</p> <p>Dr. Ralph Schuster Phone: +49 (0)228 3821 1233 mailto:Ralph.Schuster@dlr.de</p>
Germany	<p>German Research Foundation (DFG) www.dfg.de</p>	<p>Dr. Katja Großmann Phone: +49 (0) 228 885 2565 Fax: +49 (0) 228 885 2777 mailto:katja.grossmann@dfg.de</p>
Greece	<p>General Secretariat for Research and Technology (GSRT) www.gsrt.gr</p>	<p>Sofia DIMITROPOULOU Phone: +30 2131300187 mailto:s.dimitropoulou@gsrt.gr</p>
Hungary	<p>National Research, Development and Innovation Office (NKFIH) www.nkfi.gov.hu</p>	<p>Előd Nemerkenyi Phone: +36 1 8963987 mailto:elod.nemerkenyi@nkfi.gov.hu</p> <p>Gábor Tóth Phone: +36 1 8961727 mailto:gabor.toth@nkfi.gov.hu</p>
Ireland	<p>Health Research Board (HRB) https://www.hrb.ie/</p>	<p>Louise Drudy mailto:ldrudy@hrb.ie</p>
Israel	<p>Chief Scientist Office of the Ministry of Health (CSO-MOH) www.health.gov.il</p>	<p>Irit Allon Phone: +972-2-5082167 mailto:irit.allon@moh.health.gov.il</p>
Italy	<p>Italian Ministry of Health (MoH-IT) www.salute.gov.it</p>	<p>Dr. Gaetano Guglielmi Phone : + 39 06 5994 2197 mailto:g.guglielmi@sanita.it mailto:research.EU.dgric@sanita.it</p> <p>Dr. Monica Paganelli Phone : +39 06 5994 2408 mailto:m.paganelli@sanita.it</p>
Italy	<p>Ministry of Education, Universities and Research (MIUR) http://www.ricercainter nazionale.miur.it/</p>	<p>Aldo Covello Phone: +39 06.9722.6465 mailto:aldo.covello@miur.it</p> <p>Maria Bianco Phone: +39 06.9722.7146 mailto:maria.bianco@miur.it</p>

Italy: Lombardy	Fondazione Regionale per la Ricerca Biomedica (FRRB) www.frrb.it	Miss Paola Bello Mrs. Carmen De Francesco Dr. Paola Larghi, PhD mailto:bandi@frrb.it
Italy: Tuscany	Tuscany Region (RT/TuscReg) www.regione.toscana.it	Donatella Tanini Phone: +39 055 4383256 Teresa Vieri Phone: +39 055 4383289 mailto:ejprare@regione.toscana.it
Lithuania	Research Council of Lithuania (RCL) www.lmt.lt	Dr. Živilė Ruželė Phone: +370 676 14383 mailto:zivile.ruzele@lmt.lt
Luxembourg	National Research Fund (FNR) www.fnr.lu	Dr. Sean Sapcariu Phone: +352 261 925 33 mailto:sean.sapcariu@fnr.lu
Poland	National Centre for Research and Development (NCBR) www.ncbr.gov.pl/en/	Marcin Chmielewski Phone: +48 22 39 07 109 mailto:marcin.chmielewski@ncbr.gov.pl
Portugal	The Foundation for Science and Technology (FCT) https://www.fct.pt/index.html.en	Anabela Isidro Phone: +351 213 911 552 mailto:anabela.isidro@fct.pt Rita Cavaleiro Phone: +351 213 911 541 mailto:rita.cavaleiro@fct.pt
Slovakia	Slovak Academy of Sciences (SAS) https://www.sav.sk/?&lang_change=en	Zuzana Cernakova, PhD. Phone: +421257510118 mailto:cernakova@up.upsav.sk
Spain	National Institute of Health Carlos III (ISCIII) www.isciii.es	María Druet Phone: +34 9182 22530 E-mail: mdruet@isciii.es
Sweden	Swedish Research Council (SRC) www.vr.se	Sverker Lundin Phone: +46 (0) 8 546 12315 E-mail : sverker.lundin@vr.se
Switzerland	Swiss National Science Foundation (SNSF) www.snf.ch	Christoph Meier Phone: +41 31 308 23 62 mailto:christoph.meier@snf.ch

The Netherlands	Netherlands Organization for Health Research and Development (ZonMw) www.zonmw.nl	Harald Moonen Phone: +31-(0)70 349 53 49 mailto:moonen@zonmw.nl Sonja van Weely mailto:weely@zonmw.nl
Turkey	The Scientific and Technological Research Council of Turkey (TUBITAK) www.tubitak.gov.tr	Jale şahin Phone: +90- 312- 298 17 96 mailto:jale.sahin@tubitak.gov.tr
Multinational, for funding of PAO	The French National Institute of Health and Medical Research (INSERM) www.inserm.fr	Coordination EJP RD mailto:pao@ejprarediseases.org