European Joint Programme on Rare Diseases (EJP RD)

Call for Proposals 2022

"Development of new analytic tools and pathways to accelerate diagnosis and facilitate diagnostic monitoring of rare diseases”

Preliminary Announcement

The content and procedures of the call described in this pre-announcement may be subject to change and are not legally binding.

The **European Joint Programme on Rare Diseases (EJP RD)** has successfully implemented three 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.
A number of national and regional funding organisations will participate in the **EJP RD Joint Transnational Call (JTC) 2022** and will fund multilateral research projects on rare diseases **under the EJP-COFUND action**. Final decision on participating funding agencies will be issued before the launch of the call. The call is expected to be opened simultaneously by the following funding organisations in their respective countries/regions:

- Medical Research Future Fund (MRFF), Australia
- 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
- Foundation For Rare Diseases (FFRD), France
- German Ministry of Education and Research (BMBF), 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 (MoH-IT), 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
- Netherlands Organization for Health Research and Development (ZonMw)The Netherlands*
- The Scientific and Technological Research Council of Turkey (TUBITAK), Turkey*

**Decision is still pending for JTC2022*

❖ 1. 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.

Topic: Development of new analytic tools and pathways to accelerate diagnosis and facilitate diagnostic monitoring of rare diseases

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

1. Phenotype-driven diagnosis: integration across different ontologies, integration of shared pathways, digital phenotyping, development of artificial intelligence approaches/applications to extract health related data in aid of diagnosis;
2. Prognostic markers/biomarkers investigations for early diagnosis and monitoring;
3. Methodologies for solving cases that are currently difficult to analyze due to different underlying mechanisms (e.g. mosaicism, genomic (non-coding) alterations, gene regulation, complex inheritance), including new genomics / functional genomics technologies, multi-omics, mathematics, biostatistics, bioinformatics and artificial intelligence approaches;
4. Functional strategies to globally stratify variants of unknown significance (VUS) for clinical use; setting up of (in vitro) systems to distinguish between VUS and pathogenic variants (e.g. confirming disruption of splicing for deep intronic variants, loss of protein function, and gain of toxic protein function);

5. Development of pathway models to enable diagnosis, especially for newly discovered diseases that may share underlying molecular mechanisms with already known diseases.

It is possible to use cellular and animal models for validation of the new diagnostic approaches in the subtopics listed above where relevant.

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

• The design of the study (sample collection, statistical power, interpretation, relevant models for hypothesis validation) must be well justified and has to be part of the proposal;

• For natural history studies and patient registries: 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 that rare disease. 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;

• The new research data resulting from the project should be treated permissible according to the FAIR[1] 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 and through Elixir - compiling a list of resources for the deposition of experimental, biomolecular data). To make research data findable, accessible, interoperable and re-usable (FAIR), a data management strategy for the proposed full project is mandatory in the full proposal stage. Some countries involved in EJP RD JTC 2022 will also ask for a data management plan (DMP) at national level at the stage of full proposal or after granting of the project.

The following approaches and topics are __excluded__ from the scope of the call JTC2022:

- Interventional clinical trials to prove efficacy of drugs, treatments, surgical procedures, medical technology procedures. This also includes studies comparing efficacy, e.g. two surgical techniques or therapies. Clinical phase IV pharmacovigilance studies cannot be funded either.

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[1] FAIR: Findable, Accessible, Interoperable, Reusable (for more information: see “The FAIR Guiding Principles for scientific data management and stewardship”

- Studies on the exclusive testing of the safety of medical devices.
- Development of new therapies as covered in EJP RD JTC 2020.
- 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.**

❖ 2. Consortium Makeup

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 these leverages added value in sharing resources or expertise and has the capacity to elucidate common disease mechanisms and therapeutic targets.

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 organizations),
- enterprises (all sizes of private companies) when allowed by national/regional regulations,
- patient advocacy organizations.

The maximum duration of the project is three years.

Only transnational projects will be funded. Each consortium submitting a proposal must involve **three to six eligible** principal investigator partners from **at least three different participating countries. No more than two eligible partners from the same country can be present in each consortium** (further national limits may apply). This limit applies to Early Career Researchers and partners from underrepresented countries (see below).

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

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

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 2022 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 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 organization.

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.

The **use of JTC2022 matchmaking tool is strongly encouraged** to build multidisciplinary research projects:
https://live.eventtia.com/en/jtc2022matchmaking
(The platform will be open ten days after this publication)

❖ 3. PRELIMINARY TIMETABLE

There will be a **two-stage submission procedure** for joint applications: pre-proposals and full proposals. The call is scheduled to open on **December 14, 2021**.

An information webinar will be held on December 16, 14.00-15.30 (CET). You will need to register to participate in the webinar here:
https://forms.office.com/r/P7cYnbLLYG

An independent international Scientific Evaluation Committee will carry out a scientific evaluation according to specific evaluation criteria.

16th December 2021	Information webinar for potential applicants
16th February 2022	Pre-proposal submission deadline
End of April 2022	Invitation to full proposal
15th June 2022	Full proposal submission deadline
28th July 2022	Deadline for rebuttals
December 2022	Notification of funding decision

Further information and the official publication of the call will be available on the website of the EJP RD
http://www.ejprarediseases.org

For general questions regarding the joint call please contact the Joint Call Secretariat at the ISCIII, Spain:

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For questions regarding national eligibility criteria and requirements please contact the national contact person listed below
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❖ 4. NATIONAL CONTACT POINTS

Country/ Region	**Funding Organization**	**Contact Details**
Australia	Medical Research Future Fund (MRFF)	Elspeth Langford Elspeth.LANGFORD@Health.gov.au
Austria	Austrian Science Fund (FWF)	Stephanie Resch Phone: +43 (1) 505 67 40-8201 stephanie.resch@fwf.ac.at Anita Stürtz Phone: +43 (1) 505 67 40-8206 anita.stuertz@fwf.ac.at
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Belgium	Fund for Scientific Research – FNRS (F.R.S.-FNRS)	Dr. Florence Quist Florence.quist@frs-fnrs.be +32 (0)2 504 9351 Joël Groeneveld Joel.groeneveld@frs-fnrs.be +32 (0)2 504 9270

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
Québec (Canada)	Fonds de recherche du Québec - Santé, (FRQS),	Maxime Beaudoin maxime.beaudoin@frq.gouv.qc.ca +1 514 873-2114 ext. 4369
Czech Republic	Ministry of Education, Youth and Sports (MEYS)	Judita Klosaková judita.klosakova@msmt.cz + 420 234 811 504
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France	The French National Agency for Research - ANR	Dr. Florence Guillot florence.guillot@anr.fr EJPRDcall@anr.fr
France	Foundation For Rare Diseases (FFRD)	Diana Desir-Parseille Diana.desir-parseille@fondation-maladiesrares.com aap-bio@fondation-maladiesrares.com
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
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Ireland	Health Research Board (HRB)	Amanda Daly Amanda.Daly@hrb.ie
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