
EUROPEAN JOINT PROGRAMME
ON RARE DISEASES (EJP RD)
EUROPEAN JOINT PROGRAMME
RARE DISEASES

Joint Transnational Call 2022

Topic JTC 2022: "Development of new analytic tools and pathways to accelerate diagnosis and diagnostic monitoring of rare diseases"

Joint Transnational Call 2022

INFORMATION:

https://www.ejprarediseases.org/joint-transnational-call-2022/

REGISTRATION AND SUBMISSION

https://https://ptoutline.eu/app/ejprd22

Joint Call Secretariat 2022

JCS 2022 is hosted by the National Institute of Health of Spain Carlos III

Maria Druet

mdruet@isciii.es

Documents: CALL TEXT

GUIDELINES FOR APPLICANTS

PRE-PROPOSAL TEMPLATE

- **Funding agencies: 27**
- **Countries: 21**

(Australia, Austria, Belgium, Canada, Czech Republic, Finland, France, Germany, Hungary, Ireland, Israel, Italy, Lithuania, Luxembourg, Poland, Slovakia, Spain, Sweden, Switzerland, The Netherlands and Turkey)

- We expect to fund **15 -20 projects**
- **Budget** committed for projects funding : **24.000.000€**

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Documents Joint Transnational Call 2022

EJP RD - European Joint Programme on Rare Diseases - Documents Joint Transnational Call 2022

Electronic submission system

https://ptoutline.eu/app/ejprd22

EJP RD Joint Transnational Call 2022 Documents:

Call-Text – EJP RD – JTC2022

Guidelines – EJP RD – JTC2022

Pre Proposal Form – JTC 2022

Specific documents for eligibility pre-check available to download:

- Fondazione Regionale per la Ricerca Biomedica (FRRB) Lombardy. EJP pre-eligibility-check-FRRB_2022

The ejp budget tool FRRB 2022 and TUBITAK rules and terms for the national application will be available soon.

Please note that beside the above other regional/national funding organisations may demand submission of specific documents through the regional/national systems. All details can be found in the Guidelines for Applicants.
Please note that the inclusion of a non-eligible research partner (principle investigator) in a proposal **leads to the rejection of the entire proposal without further review**.

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Call for Proposals 2022

MAIN CHARACTERISTICS

- Launched **every year** in December, pre-announcement in November
- 2-stage evaluation process (short pre-proposals + invitation to submit full proposal after 1st round of scientific evaluation)
- Pre-proposal submission stage open for **60 days**
- **Rebuttal stage** included in full proposal evaluation (applicants have possibility to respond to evaluators' comments)
- A minimum of **4** eligible **research teams** and a max. of **6** per project (can be extended to 8 according to specific conditions) **4** different countries.
- Involvement of **under-represented countries** is encouraged
- Involvement of **Patient Advocacy Organisations** is encouraged
- Projects are multinational but **funding is national** (contract is signed by national funding bodies)
- Typical success rate:
  - 1st stage = 10-12%;
  - 2nd stage = 35 -50%

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Subtopics:

- **Phenotype-driven diagnosis**: integration across different ontologies, integration of shared pathways, digital phenotyping, development of artificial intelligence approaches/applications to mine health related data to aid diagnosis;
- **Prognostic markers/biomarkers investigations for early diagnosis and monitoring**;
- **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**;
- **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);
- **Development of pathway models to enable diagnosis**, especially for newly discovered diseases that may share underlying molecular mechanisms with already known diseases.

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

- **The design of the study** must be well justified and has to be part of the proposal;
- 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 , information** regarding the rare disease population to pursue **clinical trials** or other health care.
- Clear plans for **sustainability** of the resources must be described.
- 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.
- The **new research data resulting** from the project should be treated permissible according to the FAIR principles, and deposited and shared, according to the national/regional rules of the countries involved.
- To make data accessible through **RD-Connect** and through **Elixir** - compiling a list of resources for the deposition of experimental, biomolecular data.

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Excluded topics

- **Interventional clinical trials to prove efficacy of drugs, treatments, surgical procedures, medical technology procedures**. 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)**:
  - 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.
  - 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.

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Project description

- 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

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CONSORTIA COMPOSITION

Categories of partners

- **Academia** (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)

- **Patient advocacy organizations** (PAOs)

Consortia Composition

- Each consortium must involve **4 to 6 eligible partners** from at least **4 different participating countries**. The **number** of partners can be increased to **8** in two cases:
  - The inclusion of partners from participating countries usually underrepresented in projects (**Slovakia, Hungary, Lithuania, Poland, and Turkey**).
  - The inclusion of Early Career Researchers as full partners
- **Patient advocacy organizations (PAOs)** requesting funding do not count toward the total number of partners in the consotia.
- No more than **2 eligible partners** from **the same country in a consortium** (Further national/regional limits may apply).

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Consortia Composition

Minimum number partners requesting funding	Maximum number	Conditions
4	6	- At least 4 different participating countries
4	8	- At least 4 different participating countries - The inclusion of partners from participating countries usually underrepresented in projects AND/OR the inclusion of Early Career Researchers

- No more than 2 eligible partners from the same country per consortium
- PAOs requesting funding do not count toward the total
- Collaborators and sub-contractors do not count toward the total

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Matchmacking tool

https://virtual-stage.eventtia.com/en/jtc2022matchmaking/stage/161617

The matchmaking tool aims to:

- help you find teams with the necessary expertise to build multidisciplinary research projets
- help you find a consortium looking for your team's expertise

Do not hesitate to register!
Today **51** persons are registered

Early Career Researchers

To be considered an ECR, these applicants must provide:

- the certificates of both a medical doctor degree and a PhD, two to seven years prior to the pre-proposal submission deadline.
- or proof of an appointment that requires doctoral equivalency (e.g. post-doctoral fellowship or professorship appointment) two to seven years

- Medical Doctor applicants that do not hold a PhD must have been awarded their MD four to nine years prior to the pre-proposal submission deadline.

For medical doctors who have been awarded both an MD and a PhD, the date of the earliest degree that makes the applicant eligible will be used to calculate eligibility.

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CALL TIMELINE

Registration and submission

There will be a two-stage submission procedure for joint applications: a pre- and full proposal stage

1. Register as soon as possible via the electronic proposal system: https://ptoutline.eu/app/ejprd22.

2. Pre-proposal submission deadline: **February 16th** 2022 at 2pm (CET)

3. Full proposals submission deadline: **June 15th** 2022 at 2pm (CEST) (only from those applicants who were explicitly invited by the JCS to submit them)

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Call Timeline

NOV 2021

- Nov, 15 **Pre-announcement of the call**

DEC 2021

- Dec, 14 **Publication of the JTC 2022**
- **Information webinar for potential applicants** December 16, 2021

Feb 2022

Feb 16, Submission deadline for the pre-proposals. 2pm (CET)

APR 2022

Communication selection of`pre-proposals

MAY 2022

- **Communication on "widening principle"**
- **Mentoring service**

JUN 2022

- **Jun, 15 Submission deadline for full proposals**

JUL 2022

- **Jul, 28 Deadline for rebuttal**

SEP 2022

- **September 2022, 2nd SEC meeting, CSC meeting**

NOV 2022

- **Funding decisions**

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Evaluation criteria and procedure

Evaluation scores will be awarded according to specific evaluation criteria that are in line with *Horizon 2020* rules 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.

Evaluation criteria

- **Excellence**

Objectives, methodology, feasibility

- **Impact**

Expected results for relevant application, innovative potential, benefit to patients, their families and carers

- **Quality and efficiency of the implementation**

Coherence and effectiveness of the work plan, complementarity of the participants

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Evaluation Process

Pre- proposals (passing the eligibility check)

SEC Members (Each proposal assessed at least by 2 SEC Members)

First evaluation
Ranking List

CSC decides which pre-proposals will be accepted for full proposal submission

Recommendations from the SEC will be forwarded to applicants

Widening procedure:
Research teams of underrepresented or undersubscribed countries may join successful pre-proposals

Submission Full proposals

Full- proposals (passing the formal criteria checked by the JCS)

Evaluation of the external reviewer
Each proposal will be allocated to at least two external scientific reviewers

Rebuttal stage
Each project coordinator will be provided with the opportunity to provide a written response to the evaluations of the external reviewers. The scores will not be given at this stage

SEC Meeting Evaluation

- The SEC will discuss each proposal considering evaluation criteria, external reviews, rebuttals, and their own discussions.
- **Two additional groups** of reviewers will be present at the second SEC meeting to evaluate projects: **Patient representatives** and **Statistical / methodological experts**

Ethical Evaluation
Full proposals recommended for funding will be remotely evaluated by independent experts in ethics

Final scores, rank proposals recommended for funding
The **final summary review report** will be sent to applicants

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Ethical evaluation

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.

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.**

Thank you for your attention!

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