Information Webinar
Joint Transnational Call 2023 European Joint Programme on Rare Diseases (EJP RD)

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Information Webinar
December 15th, 2022

EUROPEAN JOINT PROGRAMME
RARE DISEASES

Agenda

1/ Presentation of JTC2023

2/ Clinical support services

3/ PAO engagement/involvement

4/ FAIRification of data

Call Topic

Natural History Studies addressing unmet needs in Rare Diseases

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Participating countries

- **Funding agencies**: 21

- **Countries**: 16

(Belgium (wallonie), Canada, France, Germany, Hungary*, Ireland, Israel, Italy, Lithuania, Luxembourg, Poland, Slovakia, Spain, Sweden, Switzerland, and Türkiye*)

* *decision pending*

- We expect to fund **10-15 projects**

- 3 years projects !

Budget committed for projects funding : **~18 mio€**

At least one of the following area to be covered:

- 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…).

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;
- 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;
- Providing harmonized/standardized data, collected using innovative means (AI, simulations, modelling…) 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;

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Additional elements need to be considered in the application

- 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.

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Excluded topics

- 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.
- Topics covered in EJP RD JTC 2020 and JTC2022.
- **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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Consortium make-up

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Eligibility

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)

➔ All partners shall contact their respective funding organizations and confirm eligibility in advance of submitting an application

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 Türkiye*)
  - The inclusion of Early Career Researchers as full partners
- Patient advocacy organizations (PAOs) do not count toward the total number of partners in the consortium
- No more than 2 eligible partners from the same country in a consortium (Further national/regional limits may apply)

Consortium make-up

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.

Patients' advocacy organisation

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

- From an early stage in proposal development, applicants should consult relevant disease-specific patient organizations and/or alliances of rare disease patient organizations.

Depending on their activities within the consortium and on the specific guidelines from each funding agencies, PAOs from participating countries, can participate:

- as partners
- as collaborators
- as sub-contractors

Collaborator

- Secure their own funding
- Must supply a letter of intent in the proposal and CV
- Cannot be work package leaders
- Do not count in the total number of partners in the consortium
- Can come from a non-participating country

Subcontractor

- May cover only a limited part of the action
- Funding rules according to country/regional rules
- Do not count in the total number of partners in the consortium

Looking for collaborations:

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!

https://connect.eventtia.com/en/public/events/jtc2023matchmaking/registration/attendees

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Evaluation procedure

- **Scientific Evaluation criteria**
  - **Excellence**

Objectives, methodology, feasibility

  - **Impact**

Expected results for relevant application, innovative potential, benefit to patients, their families and carers, inclusion of ECR or company

  - **Quality and efficiency of the implementation**

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

- **Ethics Evaluation at full-proposal stage**

Full proposals recommended for funding by the Scientific Evaluation Committee (SEC) will be remotely evaluated by independent experts in ethics

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INFORMATION

https://www.ejprarediseases.org/joint-transnational-call-2023/

Documents:

CALL TEXT

GUIDELINES: version downloaded

PRE-PROPOSAL TEMPLATE

Specific documents for eligibility pre-check from some funding agencies

General Information

Registration & Submission

To be filled by the coordinator

https://ptoutline.eu/app/ejprd23

- Administrative part on the platform
- Pre-proposal application template to upload (5-pages document)

Timeline

- Pre-proposal submission deadline: **February 15th 2023**, 14h CET;
- Invitation for full-proposal stage: end of April;
- Information webinar in **May**;
- Full-proposal submission deadline: **June 14th 2023**, 14h CET;
- Rebuttal stage included in full proposal evaluation (applicants have possibility to respond to evaluators' comments): end of July

Questions

- Disease eligibility/scope of the call/project description
  - Hereditary Spastic Paraplegias : Yes
  - Friedreich Ataxia: Yes
  - common diseases (e.g. schizophrenia) that are likely due to rare genetic causes (i.e. rare CNV, protein truncating variants) : the definition of a RD is indicated in the call text
  - primary immunodeficiency among secondary immunodeficiency patients (onco-haematological, autoimmune diseases): Yes but rare cancers are excluded
  - Paediatrics cancers: No
  - Hereditary Breast/Ovarian Cancer: No
  - CACNA1A disorders: yes
  - Deciphering the role of genetic markers on RD predisposition: Yes
  - Funding single-cell genomics projects applied to patient-derived primary samples as a valuable way to obtain insights into disease mechanisms and potentially biomarkers: this is a methodology so the evaluation panel will decide whether it is appropriate for a NHS
  - Can the project be set up among cohorts already existing yes (is it encouraged?) not specifically
  - Is the focus on one disease area in conformity with the call? Yes
  - Funding Longitudinal Natural History Study? 3 years project and clinical trials excluded

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Questions

- Consortium make-up
  - balance expected between clinical teams vs preclinical/research academia teams : build it according to the needs for the project
  - Proportion/weight is expected in the project between epidemiological research/and translational research (biomarkers): build it according to the needs for the project
  - company / SME as partner: fully encouraged but rules depends on the funder
  - Can the main applicant be an Assoc Prof? Yes but check the eligibility rules of your funding body
  - Is the call open to a private italian health foundation with a polyclinic? Check pages 30 to 39 of the guidelines

https://www.ejprarediseases.org/wp-content/uploads/2022/12/4-EJP-RD-JTC2023-Guidelines_fv.pdf

  - Deliverables and related deadlines during the project duration: see page 15 of the call text

https://www.ejprarediseases.org/wp-content/uploads/2022/12/3-EJP-RD-JTC2023-Call-text_fv.pdf

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THANK YOU

https://www.ejprarediseases.org/documents-jtc2023/

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