European Joint Programme on Rare Diseases

(EJP RD)

Call for Proposals 2022

Recommendations for full proposal submission and practical information

Recommendations for full proposal

Writing of the full proposal

- The full proposal should have a clear lead hypothesis for the presented proposal and should not be a fishing expedition.
- The aims of the full proposal should be clearly described and addressed in the work plan.
- The full proposal should show clear coordination and organisation of the work performed by the different partners. Proposals in which the whole is greater than the sum of their parts are usually the most competitive.
- The full proposals should a) highlight innovation (if any) and b) demonstrate feasibility.
- The full proposal should show preliminary data that justify and support the proposed studies.
- The full proposal should give a strong rationale for the relevance or justification of the cell and/or animal models if they are used.
- The impact of the results to be gained in the project should be clear and compelling.
- The abstract of the full proposal should reflect the content of the proposal and be well written as this abstract will be published on the EJP RD website if the project will be funded.
- Respect of the relevant European and international standards is mandatory (e.g. the new EC Regulation (EC 2016/679) on the protection of natural persons with regard to the processing of personal data and on the free movement of such data; e.g. the ARRIVE Guidelines for animal research (See the Call text and the instructions in the full proposal form).

Recommendations for (bios-statistical and bio-informatics) methodology

- The full proposal should give a good and sound description of statistics with clear methodology articulated, including power calculations where warranted.
- The sample size should be justified for the research undertaken.
- Where statistical or bioinformatics analysis is a prominent component of the proposal it is expected, as is the case for any other critical methodological component, that investigators with relevant expertise will be among the PI's listed in the project. The panel will pay close attention to the demonstrated expertise among the applicants not just in generating 'omics data, but also in integration of multidimensional data and in application of machine learning/AI and related approaches.
- Be specific on

a. Methods and feasibility of data collection (e.g. strategies and timelines for patient/sample recruitment, measurement instruments to be used).
b. Harmonization of information and structured data across project partners
c. Predefined outcomes in patients during disease

- Be specific which methods will be used to
  a. Describe natural history
  b. Discover new disease aspects / clinical features / biomarkers etc.
  c. Validate new disease aspects / clinical phenomenon / biomarkers etc.
- Provide transparent justifications on precision or robustness of the expected results of the project (this should follow from the planned study size, expected number of relevant outcomes in patients, etc.). Justification that the expected sample size will be sufficient for main research questions is of special importance.
- If work packages (WPs) cover different study methods, provide for each WP specific and properly detailed information on (biostatistical and bioinformatics) methodology.
- Be specific which project partners are competent to conduct and take responsibility for the points listed above. There should be a PI assigned for each mission critical deliverable, and expertise of the PI in providing the deliverable should be clearly demonstrated in their CVs particularly for the following technologies as applicable:
  a. bio-banking - clinical sample handling & storing & databasing skills,
  b. biomarker technology (-omic and imaging ability and equipment),
  c. biostatistics - (for epidemiological data analysis and sample size calculation),
  d. bioinformatics - for analysis of -omic and or image data and integration of such data,
  e. artificial intelligence / neural ne-work /machine learning.

  Preliminary data supporting that the mission critical deliverables are feasible is always helpful.

Recommendations for the involvement of patient advocacy organizations (PAOs) and patient representatives

- 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 detailed justifications of allocated resources. Consider using tools as INVOLVE cost calculator.
- Active patient participation is encouraged in the EJP Rare Diseases program.
- If a project doesn´t involve patients or PAOs, this absence should be clearly explained (i.e., why it is not relevant) in order to ensure that researchers have thought about patient involvement but did not deem it relevant in the proposed studies for specific reasons.
- Patient advisory boards should be established to discuss: the research idea; possible outcomes; patient reported outcome measures; informed consent; acceptability of the research and its design; integration of the needs and priorities of patients within the project; review of the data collected etc.
- Sources of where to find patient representatives and PAOs willing to be involved in research are: Orphanet, EURORDIS, European Reference Networks (ERNs) and EUPATI. Regarding the ERNs, all have integrated patient representatives, the ePAG representatives, within their governing boards, disease-specific committees and transversal working groups.

The consortium is encouraged to involve PAOs as active partners in their research project. The funding conditions for the PAOs are set out for each country in the guidelines for applicants.

Data management and FAIR principles

As the data management plan is mandatory in EJP RD Calls we would like to give you some additional information.

The full proposal should address the different aspects of "FAIR" data explicitly (see also https://www.go-fair.org/fair-principles/ and https://www.go-fair.org/technology/):
Findable: How the information on the data and the place where they are stored will be promoted (Publication, website of Consortium); When will they be findable (requirement: ultimately 1 year after the EJP RD funding period of the project).
Accessible: Is there information on the way how to access the data in a technical and organisational, administrative way? What is the technical process? Is there a Data Use and Access Committee which will review demands on the use of data from an external group? Is there a limit to the time allowed for this Committee to provide the answers? When will secured deposition of data in (what type of) databases occur?
Interoperable: Describe the Metadata, what information on the data will be provided? Are the experimental conditions described, are there quality measures of the data provided? Etc.
Reproducible/reusable: Which analysis script (representing software specific statistical code which allows to replicate the analysis) is provided to reproduce the main results of the project, what are the scripts you may submit to get your requests analysed? Will the code eventually be made public (e.g. on GitHub, see https://guides.github.com/activities/hello-world/) and when?

Practical information

Please read carefully the following important information. Your pre-proposal has been re-opened in PT Outline for the submission of your full proposal. Please do **not only** upload your new full proposal form, but **also review and adjust your previous input in all PT Outline sections.** Changes are made in **all** sections, so please read everything carefully and adjust your answers if necessary. Please pay extra attention to the explanation on the budget for the project coordinator *and* project partners.

- The guidelines for applicants contain instructions for the full proposal submission and can be downloaded from the EJP RD website (https://www.ejprarediseases.org/documents-jtc2022/).
- The information given in the pre-proposal is binding. Thus, any fundamental changes between the pre- and full proposals, e.g., composition of the partners of the consortium, objectives of the project, must be communicated to the Joint Call Secretariat (EJP RD JTC 2022 Call secretariat) with detailed justification and will only be allowed by the Call Steering Committee (CSC) under exceptional circumstances.
- A single pdf document (digitally converted from Word, not a scanned file) containing all the information solicited in the full proposal template, and the adjustments for the full proposal in all PT Outline sections, must be uploaded in the electronic submission system not later than **15 June 2022 at 14:00 CEST**.
- Only the full proposal template will be accepted.
- The account you have used for the pre-proposal registration is activated and you will be able to enter additional data for the full proposal submission.

- **Incomplete full proposals, proposals using a different format or exceeding length limitations of any sections will be rejected without further review.**
- In the literature reference list, it is essential to use Vancouver Style (see: International Committee of Medical Journal Editors. Uniform Requirements for Manuscripts submitted to Biomedical Journals. NEJM 1997;336:309-15) or Harvard referencing style (see: https://www.mendeley.com/guides/harvard-citation-guide) and include PUBMED, WoS or SCOPUS IDs. Apply the chosen style consistently throughout the whole proposal.
- Please note that project coordinators will be provided with the opportunity of studying the assessments of external reviewers and commenting on their evaluations of full proposals (for details see point 5.2. Rebuttal stage in the "Call text"). You will have up to one week (deadline 28 July 2022) for this optional response to the reviewers' comments (max 1 page).

We would like to advise you to read again the Guidelines, the Call text and the recommendations above carefully before completing the full proposal form.

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