FONDATION Maladies Rares

CALL FOR PROPOSALS 2025

- OMICS -

Omics approaches to study the genetic and molecular basis of rare diseases and improve their diagnosis

Deadline for applications: **February 25th, 2025, 5:00 pm (CET)**

CONTEXT AND OBJECTIVES

Rare disease diagnosis is a real challenge for patients affected by a rare disease, with an average diagnostic delay of 1.5 years, but this can be up to 5 years for 25% of patients. Deciphering the genetic and molecular basis of rare diseases is therefore a real need to reduce diagnostic delays and improve patient care. In line with this observation, Goal 1 of the International Rare Diseases Research Consortium (IRDiRC) is that "All patients presenting with a suspected rare disease will be diagnosed within one year if their disease is known from the medical literature; all currently undiagnosable individuals will enter a globally coordinated diagnostic and research pipeline".

In France, the "Plan France Médecine Genomique" (PFMG), in collaboration with the "Filières de Santé Maladies Rares" (FSMR) , aims to change the way patients are diagnosed, prevented and treated by ensuring that all patients affected by a rare disease have equal access to whole genome sequencing.

For many years, the Fondation Maladies Rares / Foundation For Rare Diseases (FMR/FFRD) has contributed to this effort through its "GenOmics" call for proposals by funding research projects aimed at deciphering the genetic and molecular basis of rare diseases.

For the 2025 call, the Fondation Maladies Rares / Foundation for Rare Diseases has decided, following the deliberations of its Scientific Advisory Board, to broaden the scope of its call to include not only genomics, but also proteomics and metabolomics, with the same objective of reducing the diagnostic gap in rare diseases.

PROGRAM DESCRIPTION

The objective of this call is to support hypothesis-driven research projects aimed at elucidating the genetic and molecular basis of rare diseases with a view to improving diagnosis using omics approaches (i.e. genomics, proteomics and metabolomics). The choice of technology used must be appropriate to the biological question and clearly justified in the submitted project, in particular the choice of whole-exome sequencing.

Expected projects:

- Projects must focus on **human diseases with a well-characterised clinical phenotype** and for which there is **clear evidence of a genetic origin**.
- Projects must demonstrate **near-term potential leading to molecular diagnosis**,
- Applicants must demonstrate the availability of an expert **bioinformatics hub** for data analysis,
- Physiopathological projects aiming at a better understanding of rare diseases are not considered eligible,
- Projects focusing on disease modifying genes are considered out of scope,
- The FFRD is not intended to fund short read WGS for which there is a pre-indication from the PFMG. Such projects are not eligible for this call. The list of pre-indications is available here: https://pfmg2025.aviesan.fr/professionnels/preindications-et-mise-en-place/.

For rare cancers, the French National Cancer Institute (INCa) and the FFRD have jointly defined the following criteria:

- High throughput sequencing projects concerning primary malignant tumours should be addressed to the INCa,
- Projects concerning benign tumours as well as systemic rare diseases involving tumour development will be evaluated within this call.

WORK WITH TECHNOLOGY PLATFORMS

FFRD has established partnerships with several technology platforms that offer an outstanding range of expertise, skills and services.
The list of partner platforms is available on the FFRD website: https://fondation-maladiesrares.org/en/plateformes-partenariats/.

Applicants are strongly encouraged to use one of FFRD's partner platforms for their project, but the use of another platform is permitted. Please note that FFRD may direct the applicant to another platform if the experimental conditions are similar and the price is lower.

Please note that a quotation from the platform must be submitted with the application (whether or not the platform is a partner of the Foundation). The application will be considered ineligible if the quotation is not submitted on time. Please allow for the time platforms will need to provide you with their quotation.

SAMPLES

This program is intended for **qualified existing biosamples** analysis only. It will not support participant enrolment, consent or biosample collection. **Consents for research purpose sample analysis must be obtained before application.**

For all approaches:

- Samples must be available in the laboratory at the time of submission,
- Quality control of samples must be available at the time of submission.

All this information must be detailed in the application.

ELIGIBILITY

The applicant of the study must be part of a French research team affiliated to the academic sector (research team working in universities, other higher education institutions or research institutes) and/or the clinical/public health sector (research team working in hospitals/public health organisations).
Early career researchers are encouraged to apply as principal investigators.

EVALUATION

Applications will be peer-reviewed by at least two national or international academic experts in the field and selected by a dedicated Scientific Committee composed of members of the FFRD Scientific Advisory Board and experts in the field, based on the following criteria:

- Relevance and significance of the project,
- Project quality and scientific soundness,
- Feasibility of the project,

- Innovation,
- Quality of the applicant and quality of the laboratory.

FUNDING

Funding will only cover the costs of the technological platform (services and consumables) based on the quote provided in the application.
Funding is not intended to cover the costs of the applicant's laboratory.
Overheads are not allowed by the FFRD. FFRD grants are not subject to VAT.

For information only, projects ranging from 1 k€ to 60 k€ have been funded through this call in the last 6 years, with an average funding of 20 k€ per project.

Only one project per research team will be funded for the current call.

SUBMISSION AND SCHEDULE

Applications can only be submitted on the FFRD Synto online platform: https://ffrd.syntosolution.com/.

Provisional schedule:

Launch of the call	January 7th, 2025
Submission deadline for application	February 25th, 2025 - 5:00 pm (CET)
Notification of the results	June 2025

Results will be communicated by e-mail to the applicant.

Applicants resubmitting projects must provide a detailed response to the comments made by the FFRD Scientific Committee at the previous call, highlighting the changes in the revised version.
Applicants who are part of a research team already funded by the FFRD since 2017 must have provided a detailed report on the results and impact of any completed project(s). For ongoing projects, a progress and/or preliminary data report is required.
Report forms are available in the Applicant Portal ("Documentation" tab) or upon request by e-mail at aap-bio@fondation-maladiesrares.com. Please attach all reports to the proposal in the appropriate section.

FAIR POLICY / IRDIRC POLICIES AND GUIDELINES

By submitting a project to this call, applicants agree to comply with the following requirements: FAIR guiding principles for scientific data management and stewardship.

The objectives of the call are in line with the objectives of the International Rare Diseases Research Consortium (IRDiRC). Applicants are expected to follow IRDiRC policies and guidelines.

COMMUNICATION

Applicants must agree that the title and non-confidential abstract of funded projects, as well as the name and affiliation(s) of the Principal Investigator(s), will be made public and published on the FFRD website: http://fondation-maladiesrares.org.

ACKNOLEDGEMENT POLICY

Applicants must acknowledge the FFRD as the source of funding in all communications related to the project (posters, oral presentations, scientific publications, etc.) using the terms "Foundation For Rare Diseases" or "Fondation Maladies Rares" and/or the FFRD logo (available upon request). Reference(s) to the publication(s) must be sent to the FFRD by email to aap-bio@fondation-maladiesrares.com

CONTACT

Please contact aap-bio@fondation-maladiesrares.com with any questions relating to this call.