

Omics approaches to improve the diagnosis of rare diseases

Deadline for applications: **February 27th, 2026, 5:00 pm (CET)**

OPEN TO ALL OMICS TECHNOLOGIES
RESTRICTED TO PROJECTS WITH A SHORT-TERM IMPACT ON DIAGNOSTIC IMPROVEMENT

CONTEXT AND OBJECTIVES

Diagnosing rare diseases remains a major challenge: while the average time to diagnosis is around 1.5 years, 25% of patients wait up to five years. Unraveling the genetic and molecular mechanisms underlying rare diseases is therefore essential to shorten diagnostic delays and improve patient care. In this context, Goal 1 of the International Rare Diseases Research Consortium ([IRDiRC](#)) states 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 Génomique" ([PFMG](#)), in collaboration with the "Filières de Santé Maladies Rares" ([FSMR](#)), seeks to transform how patients are diagnosed, monitored, and treated by ensuring equitable access to whole-genome sequencing for everyone affected by a rare disease.

For many years, the Fondation Maladies Rares / Foundation for Rare Diseases ([FMR/FFRD](#)) has supported this effort through its "GenOmics" call for proposals, funding research aimed at elucidating the genetic and molecular basis of rare diseases.

Following the recommendations of its Scientific Advisory Board, the Foundation has decided to expand the scope of the call to include all omics approaches from 2025 onwards. The goal of reducing the diagnostic gap in rare diseases will remain the same.

To ensure that your project is eligible, please carefully read the entire call text.

PROGRAM DESCRIPTION

The objective of this call is to support hypothesis-driven research projects that aim to improve diagnosis of rare genetic disease using omics approaches.

Expected projects

Please note that only projects meeting the following prerequisites will be eligible for evaluation:

- Projects must focus on **rare human diseases that have a clearly defined phenotype** and for which there is compelling **evidence of a genetic origin**. This has to be discussed in the application.
- Projects must demonstrate **short-term potential for improving molecular diagnosis**.
- Applicants must demonstrate that an expert **bioinformatics hub** is available for data analysis.
- All 'omics' technologies are eligible, but the **choice of technology must be justified**, particularly for short-read whole-exome sequencing. For a long-read genome sequencing project, short-read genome sequencing must have been performed beforehand. If not, the reasons must be explained.
- Projects focusing on the study of different family members should include a **phylogenetic tree** and specify the phenotype of each subject and for which subjects biological samples are available.

Ineligible projects

The following projects will be deemed ineligible and will not be evaluated:

- Physiopathological projects aimed at improving our understanding of rare diseases.
- Projects that aim to improve the prognosis or progression of an identified disease.
- Projects focusing on disease-modifying genes.
- The FFRD will not fund short-read whole-genome sequencing projects for which the PFMG has given a pre-indication. 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

Please note that FFRD funding is only intended to cover the costs of the technological platform, as set out in the provided quote.

The FFRD has established partnerships with several technology platforms offering an exceptional range of expertise, skills, and services.

A full list of these platforms can be found on the FFRD website: <https://fondation-maladiesrares.org/en/plateformes-partenariats/>.

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

A quotation from the platform must be submitted with the application, whether or not the platform is a FFRD partner. Applications will be considered ineligible if the quotation is not submitted on time. Please allow time for the platform to provide their quotation.

SAMPLES

This programme is intended for the analysis of **qualified existing biosamples** only. Consent for the analysis of samples for research purposes must be obtained before application.

For all approaches:

- **Samples must be available** in the laboratory at the time of submission.
- Quality control of the samples must also be available at this time (unless the technology requires material to be extracted from biological samples on an ad hoc basis).

All of this information must be detailed in the application.

ELIGIBILITY

Applicants must be part of a French research team affiliated with the academic (universities, other higher education institutions, or research institutes) or clinical/public health (hospitals/public health organisations) sectors.

Early career researchers are encouraged to apply as principal investigators.

If the project has been entirely or partially prepared using artificial intelligence, this must be clearly stated in the proposal.

FUNDING

Funding will only cover the costs of the technological platform (including services and consumables), as set out in the quote provided in the application.

As a guide, projects ranging from €1k to €60k have been funded through this call over the last six years, with an average funding of €20k per project.

Please note that overhead costs are not eligible for FFRD funding. Furthermore, FFRD grants are exempt from VAT.

For the current call, only one project per research team will be funded.

SUBMISSION AND SCHEDULE

Proposals must be submitted exclusively through the FFRD Synto online platform: <https://ffrd.syntosolution.com/>. The application form and the documents to be provided are detailed on the online portal.

Please note that:

- Prior to submitting their project, candidates and their directors are required to create and complete their profiles on the Synto platform. For guidance, please refer to the tutorials available in the 'Documentation' section of the Synto platform.*
- Technical support is only available from the moment the call is initiated until 24 hours before the submission deadline. Candidates are responsible for identifying any potential issues in good time, as no extensions will be granted after the call for proposals has closed.*
- All communications will henceforth be sent via email through the FFRD Synto online platform. Please ensure that emails from 'FFRD by Synto' are received in a timely manner. Candidates are advised to regularly check their Synto account to monitor the progress of their application.*

Provisional schedule:

Launch of the call	January 8 th , 2026
Submission deadline for application	February 27 th , 2026 - 5:00 pm (CET)
Notification of the results	June 2026

Please note that projects should start within nine months of the funding decision being announced.

Applicants resubmitting projects must provide a detailed response to comments received from the FFRD scientific committee in the previous session, clearly highlighting any changes made in the revised version.

In order to be eligible to submit new funding applications, applicants who have already received funding from the FFRD must have submitted the interim and final financial and scientific reports requested.

EVALUATION

Firstly, the FFRD will assess the administrative and budgetary eligibility of the submitted projects, as well as their suitability within the scope of the call for proposals. Projects that do not meet these criteria may be declared ineligible.

All eligible proposals will be evaluated by at least two academic experts from the relevant field, either nationally or internationally (external reviewers). Funded proposals will then be selected by a dedicated scientific committee comprising internal experts. The evaluation and selection of funded proposals will be 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.

ADMINISTRATIVE AND FINANCIAL MONITORING

Successful applicants must ensure that a managing institution is available for contracting purposes; otherwise, funding will be lost. A research agreement will indeed be signed between the managing institution of the selected applicants and the FFRD. This agreement outlines the scientific and financial monitoring requirements for the project, and it should be signed within six months of the funding decision being made. Implementation of this policy is subject to approval by the relevant ethical bodies, where applicable.

Successful applicants will be required to submit project monitoring reports as requested by the FFRD, using the provided templates, and to report on project progress when asked to do so.

FAIR POLICY / IRDIRC POLICIES AND GUIDELINES

By submitting a project to this call, applicants confirm that they will adhere to the [FAIR guiding principles for scientific data management and stewardship](#).

The objectives of this call are in alignment with the goals set out by the International Rare Diseases Research Consortium ([IRDiRC](#)). Applicants are expected to adhere to the [IRDiRC policies and guidelines](#).

COMMUNICATION

Applicants must agree to the publication of the title and non-confidential abstract of funded projects, along with the name and affiliation(s) of the principal investigator, on the FFRD website: <https://fondation-maladiesrares.org/projets-de-recherche-laureats/>.

ACKNOWLEDGEMENT POLICY

Applicants must acknowledge the Foundation for Rare Diseases (FFRD) as the funding source in all project-related communications, such as posters, oral presentations and scientific publications. This can be done by using the terms 'Foundation for Rare Diseases' or 'Fondation Maladies Rares', or by including the appropriate logo (available upon request).

CONTACT

For any questions related to this call, please contact aap-bio@fondation-maladiesrares.com.