

Call for research projects 2024 Bardet-Biedl France

Pigmentary retinopathy

Deadline: April 04, 2024 - 5pm (Paris time)

Budget: 90 000 € - Duration max: 24 months

Contact: aap.asso@fondation-maladiesrares.com

Context

Due to the specificities of rare diseases, the Fondation Maladies Rares (Foundation For Rare Diseases - FFRD) and the association Bardet-Biedl France (BBS France) have agreed on a **partnership to support and stimulate biomedical research on the pigmentary retinopathy for an improvement of Bardet-Biedl syndrome (BBS).**

The Association is governed by the law of July 1, 1901, and its aim is to contribute to medical research and the improvement of care practices relating to Bardet-Biedl syndrome, to bring together and inform people suffering from Bardet-Biedl syndrome and their families of their rights and the progress of research, in particular in conjunction with the scientific committee, to provide a forum for discussion, exchanges between families and solidarity, to offer technical and moral support to families confronted with the syndrome, and to inform the public, the medical profession and the health and social authorities, in order to improve scientific and medical knowledge, and to raise awareness and recognition of Bardet-Biedl syndrome.

FFRD¹ is a private non-profit organization, founded in 2012 by five members²³ with the aim of helping to (i) decipher rare diseases to facilitate diagnosis and accelerate the development of new treatments, (ii) improve the daily lives of sick people and their loved ones. FFRD carries a mission of general interest: to animate, coordinate and support research on rare diseases. It provides free support to patient organizations in their efforts to promote rare diseases research, for example through joint calls for projects such as the one proposed here.

Aim of the call

BBS is a rare genetic disease, characterized by multiple clinical affections: pigmentary retinopathy, post-axial polydactyly, obesity/overweight, kidney and urinary tract abnormalities, developmental delay, and hypogonadism. It is caused by dysfunctions of

¹www.fondation-maladiesrares.org

²MRNP2 :<http://www.sante.gouv.fr/le-plan-national-maladies-rares-2011-14-une-ambition-renouvelee.html>

³AFM-Telethon, Alliance Maladies Rares, Inserm, Conférence Directeurs GChu Nationaux, Conférence of University Presidents

proteins involved in the development and formation of primary cilia. More than 24 genes have been implicated in BBS syndrome (BBS1, 2, 3....).

The pigmentary retinopathy is a degenerative disorder of the retina. BBS gene have been implicated in the ciliogenesis of the RPE cells (Retinal pigment Epithelial cells). The progressive retinal degeneration leads to reduced visual acuity, night blindness, photophobia and loss of central vision and colors in late childhood or early adulthood. (Sources : Orphanet et [BBS](#))

This call for projects aims to develop **innovative and translational research program** to better understand **pigmentary retinopathy and develop new therapeutic approaches for Bardet-Biedl syndrome**. All **biomedical disciplines** are eligible.

BBS France is interested in supporting research that advances **understanding** of the cellular, molecular, genetic, and systems-level mechanisms of pigmentary retinopathy, **improvement** of treatment modalities and/or **development** of therapeutic strategies.

Particular attention will be paid to projects involving:

- Nanoparticles,
- Optogenetics,
- Rescue or restoration of dying cones.

Eligibility criteria

The project will explicitly formulate a **research question**, addressing issues specifically related to pigmentary retinopathy.

The project will have to demonstrate its **novelty**, its **feasibility** and the **expertise(s)** of the researcher(s) involved.

The principal investigator ("lead applicant") must be a **researcher**, belonging to a research organization⁴. It is expected that the lead applicant will have a permanent position.

There must be a clear match between the main methodology used in the project and the expertise of the principal investigator.

Funding

This call for research projects provides for financial support of up to **90,000 euros for a maximum duration of 24 month**.

Funding support can cover staff costs (excluding administrative staff), operations, and missions, provided they are entirely and exclusively dedicated to the project. The use of service providers for the project should only cover the execution of a very limited part of the project. Please note that indirect costs and institutional overhead are not provided.

Funding must be realistic, reasonable, detailed post by post and fully justified.

⁴**Research organization:** is considered a research organization, an entity such as universities or research institutes, or research structures dedicated to research within the health institution associated with a university or research institute, regardless of its legal status (public or private body) or its method of funding, whose primary purpose is to carry out basic research or applied research or experimental development activities and to disseminate their results through teaching, publication, or technology transfer.

An agreement will be established between the research program management organization (the lead applicant's research organization) and FFRD. **The management organization will not be able to charge any management fee on the amount awarded.**

Co-financing of the selected project is possible if there is no conflict of interest.

The lead applicant will manage the allocated funds, including, if necessary, the agreement and allocation to the partners teams.

Submission, selection, and schedule

The proposal form (download [here](#)), in English, is to be sent by email:

- **Before April 04, 2024, 5pm** (Paris time)
- In PDF format (no scan)
- At aap.asso@fondation-maladiesrares.com
- With the following email subject: "BBS24 - Your last name"

The joint selection by BBS France and FFRD will be based on:

- 1 – validation of the eligibility criteria by FFRD
- 2 – adequation of the proposal with patient's needs by BBS France
- 3 – evaluations by independent external expert(s) (cf. appendix)

BBS France members and external experts sign a confidentiality and a non-conflict-of-interest agreement.

Reports, documents and rankings sent to members of BBS France are confidential.

Scientific and financial follow-up

BBS France is committed to this approach with gravity and a strong sense of responsibility. The budget that its Board of Directors will commit to the selected projects comes exclusively from donations. These include donations from members, as well as funds donated by friends and families, sometimes following the death of one of their own, in a gesture of solidarity with those who are waiting and hoping. Laureates are expected to adhere to this commitment.

Half of the financial grant will be paid at the start of the project.

For projects lasting more than one year, the main applicant will be required to send a brief progress report by e-mail every 6 months (a few lines).

A precise scientific and financial report must be produced at the mid-term of the project, in order to justify the correct progress of the work and trigger payment of 40% of the grant.

A final scientific and financial report must be produced by the main applicant 1 month after the end of the project, to trigger payment of the remaining 10% of the grant.

Each report must be accompanied by a comprehensive and readable report for patients, whatever their scientific knowledge.

The work carried out and the results obtained are expected to be valorized in the form of mainstream publications (at least for BBS France website and newsletter) and scientific papers.

The project leader will make himself available to present his results at a meeting organized by BBS France.

Annex

Evaluation criteria

1. Excellence

- Clarity and pertinence of the objectives,
- Credibility of the proposed approach and methodology,
- Feasibility of the project,
- Competence and experience of participating research partners

2. Impact

- Potential of the expected results for exploitation and for future relevant applications
- Effectiveness of the proposed measures to exploit and disseminate the project results
- Innovative potential

3. Quality and efficiency of the implementation

- Coherence and effectiveness of the work plan
- Budget and cost-effectiveness of the project