



Call for research projects 2024

"CASK-related disorders"

Maximum budget: **45 000 €** - Maximum duration: **24 months**

Deadline: **09th January 2025 - 5pm** (Paris time)

Your profile must be created on the application platform prior to any application (instructions p3).

Context

Due to the specificities of the field of rare diseases, the AECF (Association Enfants CASK France) and the Fondation Maladies Rares (Foundation For Rare Diseases - FFRD) have agreed on a **partnership to support and stimulate research on** disorders linked to CASK gene (rare diseases group: neurological diseases; Network(s): DefiScience, AnDDI-Rares, EpiCARE, ERN-RND, ITHACA).

AECF¹ is a non-profit organization of general interest that aims to support fundamental, clinical and translational research into CASK-related disorders. The association brings together, supports and provides up-to-date information to families affected by this ultrarare disease, at national and international level, and collaborates with CASK associations abroad on scientific, medical and medico-social issues.

FFRD² is a private non-profit organization, founded in 2012³ by five members⁴, with the aim of helping to (i) deciphering rare diseases to facilitate diagnosis and accelerate the development of new treatments, (ii) improve the daily lives of people with disease and their loved ones. Its mission is of general interest: to lead, 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

What is CASK?

CASK-related intellectual disability is a disorder of brain development that has two major forms: microcephaly with pontine and cerebellar hypoplasia and X-linked intellectual disability. The disorder predominately affects females as the mutation spectrum is so severe that males often die prematurely during childhood except when having specific mutations.

Individuals with CASK-related intellectual disability typically have severe learning impairments, abnormal sleep, exhibit self-biting, hand flapping, abnormal repetitive behaviours, and recurrent seizure activity. Many affected by CASK-related intellectual disability do not develop language skills nor are able to learn to walk, associated or not with vision impairment and/or sensorineural hearing losses.

Currently, treatments are only focused on alleviating symptoms which include medication to control seizure frequency, nutritional support, hearing and vision aids, and physical therapy. While these

¹ <https://www.aecf-france.fr/>

² <https://fondation-maladiesrares.org/en/>

³ PNM2 : <http://www.sante.gouv.fr/le-plan-national-maladies-rares-2011-14-une-ambition-renouvelee.html>

⁴ AFM-Telethon, Rare Diseases Alliance, Inserm, Conference of University Hospital Directors, Conference of University Presidents

therapies can improve the quality of life of the individual when administered by a team of specialist, **there are currently no disease modifying approaches for CASK-related intellectual disabilities, representing a critical unmet medical need.**

This call aims to support **innovative** and **transdisciplinary research** programs, at **any stage** of development (fundamental, translational or clinical) **on CASK-related disorders**. All disciplines in **Biomedical Sciences** are eligible.

The association wishes to support any research aimed at **understanding the pathophysiology** of the disease, improving **diagnosis**, developing **better management modalities** and/or **developing therapeutic strategies**, with priority given to projects that have reached **proof-of-concept**.

Eligibility criteria

The project must explicitly formulate a **research question**, addressing issues specifically linked to CASK-related disorders.

The project must demonstrate its **originality**, feasibility and **the expertise(s) of the researchers** involved.

The principal investigator of the project ("lead applicant" – P.I.) must be a **researcher**, belonging to a French or foreign public research organization⁵. It is expected that the bearer will be statutory and hold a permanent position.

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

Financing

This call for research projects provides for financial support up to **45 000 €**, for a **maximum duration of 24 months**.

Funding support can cover, **if it is entirely and exclusively dedicated to the research experiments**, the costs of:

- **Fixed-term** contract staff (excluding administrative staff).
- Equipment, up to a maximum of €2,000 (any excess to be explained precisely in the response form),
- Consumables,
- Travels, up to a maximum of €2,000 (any excess to be explained precisely in the response form),
- Provision of services, if they relate only to the execution of a very limited part of the project (maximum 10% of the budget).

No environmental costs or management fees are eligible.

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

An agreement will be established between the organisation managing the research programme (organisation of the main leader) and FFRD. **The management organisation will not be able to charge any management fee on the amount granted.**

⁵ **Public research organization:** entity such as a university or research institute, or a structure dedicated to research within a health institution and associated with a university or research institute, 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.

Co-financing of the selected project is possible as long as no conflict of interest exists.

The lead applicant will be responsible for the management of the allocated funds, including, where applicable, the agreement and distribution to the partner teams.

Submission Process, Selection and Scheduled

The application form, in English, must be completed on <https://ffrd.syntosolution.com/> before **January 9th, 2025, 5pm** (Paris time).

You must have already created your profile on the application platform before submitting (a delay may be necessary). Technical support for the platform is available from 9am to 6pm until January 8th, 2025.

The joint selection by AECF and FFRD will take place based on:

- The validation of the eligibility criteria by the FFRD,
- the priorities of AECF,
- the evaluations by independent external experts (see Annex).

The members of the board of AECF and the external experts sign a confidentiality and a non-conflict-of-interest agreement.

The reports, documents and filings sent to the members of the board of AECF, FFRD and external experts/reviewers are confidential.

Scientific and financial follow-up

AECF 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 and fundraising actions. 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. PI are expected to adhere to this commitment.

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

For projects longer than one year, the PI will have to share briefly by email, every 6 months, on the progress of the project (a few lines).

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

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

Each report must be accompanied by a report understandable by all members of the association, regardless of their scientific knowledge.

It is expected that the work carried out and the results obtained at the end of the project will be promoted, in the form of publications, and scientific and mainstream communications. The project leader will make himself available for the presentation of his results at a meeting organized by AECF.

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