

## Call for research projects 2025

### " Research on PACS1 syndrome Schuurs-Hoeijmakers "

Maximum budget: **50 000 €** - Maximum duration: **12 months**

Deadline: **June 12, 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 rare diseases, the Fondation Maladies Rares (Foundation For Rare Diseases - FFRD) and the Association Syndrome PACS1 – Schuurs-Hoeijmakers (PACS1) have agreed on a **partnership to support and stimulate biomedical research on PACS1 syndrome.**

**The Association Syndrome PACS1** is a France-based charity that was created in March 2021 by patients' families. It is composed entirely of volunteers. Its purpose is to:

- o Support families affected by the PACS1 syndrome and promote the link between families.
- o Inform the public, medical profession and health and social authorities, to improve the state of scientific and medical knowledge, facilitate diagnosis and make PACS1 syndrome known and recognized.
- o Contribute to the medical research effort and to the improvement of care practices related to PACS1 syndrome.
- o Establish, develop, and maintain links with other associations and foundations working on the same purposes.

The Association Syndrome PACS1 is working closely with the PACS1 Syndrome Research Foundation (USA).

**FFRD**<sup>1</sup> is a private non-profit organization, founded in 2012<sup>2</sup> by five members<sup>3</sup>, 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

PACS1 syndrome or Schuurs-Hoeijmakers syndrome is a neurodevelopmental disorder, whose features may include distinct facial morphology, developmental delay (motor skills, language acquisition, global delay), as well as various other symptoms to varying degrees (e.g., epilepsy, orality disorder, various sensory difficulties, vision problems, autism spectrum disorder). Almost all patients exhibit the same mutation, i.e., the single nucleotide substitution c.607C>T. The disease is not associated with haploinsufficiency but more probably with a dominant negative impact of the mutated protein.

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<sup>1</sup> <https://fondation-maladiesrares.org/en/>

<sup>2</sup> PNMR2 : <http://www.sante.gouv.fr/le-plan-national-maladies-rares-2011-14-une-ambition-renouvelee.html>

<sup>3</sup> AFM-Telethon, Rare Diseases Alliance, Inserm, Conference of University Hospital Directors, Conference of University Presidents

This call for projects aims to develop **innovative research program to better understand PACS1 syndrome and develop new therapeutic approaches. All biomedical disciplines are eligible.**

Association Syndrome PACS1 – Schuurs-Hoeijmakers is interested in supporting research that advances understanding of the **cellular, molecular, genetic, and systems-level mechanisms of PACS1-related disorders**. However, priority will be given to **innovative projects which could potentially lead to treatments or a cure for those with PACS1 syndrome**.

### Eligibility criteria

The project must explicitly formulate a **research question**, addressing issues specifically related to PACS1 syndrome.

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<sup>4</sup> public research organization. It is expected that the bearer will be statutory and hold a permanent position. Otherwise, the applicant will have to provide proof of an employment contract with the organization managing the allocated funds. The said employment contract must run for the entire duration of the research project plus 6 months.

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 **50 000 €**, for a maximum duration of **12 months**.

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.

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

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.

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<sup>4</sup> **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.

## Submission Process, Selection and Scheduled

The application form, in English, must be completed at <https://ffrd.syntosolution.com/> before **12/06/2025 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 11/06/2025.*

The joint selection by PACS1 association and FFRD will take place on the basis of:

- validation of the eligibility criteria by FFRD,
- of the priorities of PACS1 association,
- evaluations by independent external experts (see Annex);
- of the final scientific opinion, issued by the scientific committee (SC) of PACS1 association.

The members of the board of PACS1 Association 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 and of the SC of PACS1 Association are confidential.

## Scientific and financial follow-up

*The PACS1 Association 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. 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 in French, 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 the PACS1 association.

## **Annex**

### **Evaluation criteria**

#### **1. Interest and importance of the research project to meet the missions of the association.**

*Does this study address an important problem? Innovative potential of the expected results for exploitation and for future relevant applications? Is there a sufficient body of evidence for high quality research that supports the rational? How is the project positioned regarding international competition in the field?*

#### **2. Quality and feasibility**

*Soundness of the concept, clarity, originality, and pertinence of the objectives. Are the conceptual framework, design, methods techniques, and analyses adequately developed, and appropriate for the aims of the projects?*

*Coherence and effectiveness of the work plan, including appropriateness of the timeframe, allocation of task and resources (financial, human, and technological resources linked to the laboratory's infrastructure).*

*Does the applicant acknowledge potential problem areas and consider alternative strategies?*

*Effectiveness of the proposed measures to exploit and disseminate the project results.*

#### **3. Quality of the applicant**

*Ability of the applicant to conduct the research based on the quality of the CV, on its publications and on the quality of the laboratory. Competence and experience of participating research partners. Does the applicant have the right network of collaborators to undertake this type of project? If not, is it important for the Association to attract this researcher because of their skills or previous work on related subjects?*

#### **4. Budget**

*Budget and cost-effectiveness of the project (rational distribution of resources in relation to project's activities, partner responsibilities, and time frame).*