Call for research projects 2022
"Research on PACS1 syndrome Schuurs-Hoeijmakers "

Deadline: June 16, 2022 - 5pm (Paris time)

Budget: 50 000 € - Duration: max 12 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 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:

- Support families affected by the PACS1 syndrome and promote the link between families.
- 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.
- Contribute to the medical research effort and to the improvement of care practices related to PACS1 syndrome.
- 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[1] is a private non-profit organization, founded in 2012 by five members[2][3] 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.

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

[1] *www.fondation-maladiesrares.org*
[2] *MRNP2 : http://www.sante.gouv.fr/le-plan-national-maladies-rares-2011-14-une-ambition-renouvelee.html*
[3] *AFM-Telethon, Alliance Maladies Rares, Inserm, Conference Directors GChu Nationals, Conference of University Presidents*

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.

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 will explicitly formulate a research **question** addressing issues specifically related to PACS1 syndrome.
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 organization[4]. It is expected that the lead applicant will have a permanent position. Otherwise, the lead applicant will have to provide proof of an employment contract with the research organization managing the allocated funds. The employment contract will have to run for the entire duration of the research project, plus 6 months.

Funding

This call for research projects provides for financial support of up to **50,000 euros for a maximum duration of 12 month**; exceptionally a financial support up to 100,000 euros for a maximum duration of 24 month may be provided.
Funding support can cover personnel expenses (excluding administrative staff and Master students), 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.

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.

[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.*

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, in English, is to be sent by email:

- Before **June 16, 2022**, 5pm (Paris time)
- At aap.asso@fondation-maladiesrares.com
- With the following email item: "PACS1 AAP2022 - *Your last name*"

The joint selection by PACS1 and FFRD will be based on:
1 – validation of the eligibility criteria by FFRD
2 – adequation of the proposal with patient's needs by PACS1
3 – evaluations by independent external expert(s) (cf. appendix)

Scientific and financial follow-up

A precise scientific and financial report will have to be produced at the mid-term of the project.
A final scientific research report, a comprehensive and readable report for patients, and a summary of the research results will have to be produced by the main applicant 3 months after the end of the project.
The work carried out and the results obtained are expected to be valorized in the form of mainstream publications (at least for PACS1 website and newsletter) and scientific papers.
Laureates may be invited to present their results at a dedicated scientific day.

Annex

Evaluation criteria

1. Excellence

- Clarity and pertinence of the objectives,
- Credibility of the proposed approach and methodology,
- Soundness of the concept,
- 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
- Benefit to patients, their families, and carers

3. Quality and efficiency of the implementation

- Coherence and effectiveness of the work plan
- Complementarity of the participants
- Plan for sustainability of infrastructures or resources initiated by the project,
- Budget and cost-effectiveness of the project