**REquest for proposals**

**National Organization for Rare Disorders (NORD) with funding from**

**The Founders of the PACS1 Syndrome Research Foundation and Public Donations**

#### *Announces*

# *Research Grant Money (up to $45,000 U.S.)*

*for*

**PACS1-Related Syndrome**

**(Schuurs-Hoeijmakers Syndrome)**

**DEADLINE FOR INITIAL APPLICATIONS: July 16, 2018 (11:59 pm ET)**

NORD, with funding from the founders of the PACS1 Syndrome Research Foundation and public donations, is accepting applications for a total of $45,000 U.S., for scientific and/or clinical research studies on PACS1-related syndrome, also known as Schuurs-Hoeijmakers syndrome. Individuals with PACS1-related syndrome have a de novo mutation (c.607C>T) in the *PACS1* gene, hypothesized to disrupt the PACS1 protein’s role as a membrane traffic regulator. Key features of the syndrome are developmental delay (cognitive, language, motor), intellectual disability, and characteristic facial appearance (including hypertelorism with downslanting palpebral fissures, arched eyebrows, bulbous nasal tip, and thin upper lip). Diagnosis is made via exome sequencing, with additional evaluation recommended to identify commonly associated clinical features, particularly brain, heart, eye, and kidney abnormalities. Treatments are used to manage symptoms and may include gastrostomy tube placement for feeding difficulties, anti-epileptic drugs for seizures, and/or speech therapy for delayed language development.

**Research Objectives**

The purpose of the NORD Rare Disease Research Grant Program is to encourage meritorious scientific and clinical studies designed to improve the diagnosis or therapy of rare “orphan” diseases. Grants will be awarded to academic researchers to initiate small scientific research studies or clinical trials, the results of which could be used to obtain funding from the NIH, FDA, or other funding agencies, or to attract a corporate sponsor. Procedures or proposed therapeutic trials may be new, based on recent biochemical or pharmacological evidence, or in preliminary stages of clinical investigation. Evaluation of proposals will include careful consideration of protocol design, objectiveness of parameters measured, statistical evaluation proposed, and overall impact on the field. Priority will be given to proposals that most effectively leverage recent advances in PACS1-related syndrome research and tools (see next page) while avoiding redundancies.

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1. Schuurs-Hoeijmakers JHM, Oh EC, Lisenka ELM, et al. Recurrent De Novo Mutations in PACS1 Cause Defective Cranial-Neural-Crest Migration and Define a Recognizable Intellectual-Disability Syndrome. Am J Med Gen. 2012;91(6):1122-1127.
2. Schuurs-Hoeijmakers JHM, Landsverk ML, Foulds N, et al. Clinical Delineation of the PACS1-Related Syndrome—Report on 19 Patients. Am J Med Gen Part A. 2016;170A(3):670-675.

**PACS1-RELATED SYNDROME: TOOLS & RESEARCH**

* Fibroblasts (Coriell/NIGMS)
  + Currently Available
  + [www.nigms.nih.gov](http://www.nigms.nih.gov)
* HeLa cells (System Biosciences)
  + Currently Available
  + [www.systembio.com](http://www.systembio.com)
* Mouse model (Jackson Laboratory)
  + Available Soon
  + [www.jax.org](http://www.jax.org)
* IPSC lines (Coriell/Wicell)
  + In Development
  + [www.coriell.org](http://www.coriell.org) (development); [www.wicell.org](http://www.wicell.org) (storage and distribution)
* Clinical history, blood and saliva samples (Simons Foundation)
  + [www.sfari.org/resource/sfari-base](http://www.sfari.org/resource/sfari-base)
* Zebrafish and cell biology studies (Duke University)
* Protein structure and molecular studies (University of Pittsburgh)

For the most current information on PACS1-related syndrome research and tools, please refer to the resources below.

* Current research: <https://pacs1foundation.org/funded-research>
* Tools and reagents: <https://pacs1foundation.org/tools>
* Publications to date: <http://pacs1smiles.org/research>

**Application Procedure**

**Deadline for Initial Applications: July 16, 2018 (11:59 pm ET)**

All of the following application elements must be received by July 16, 2018 (11:59 pm ET). Incomplete applications may not be considered. For electronic submission (preferred), all application elements should be merged into a single PDF file and sent to [research@rarediseases.org](mailto:research@rarediseases.org) with “NORD Abstract Proposal” as the subject line. Alternatively, applications can be mailed to:

**NORD Rare Disease Research Grant Program**

**55 Kenosia Avenue**

**Danbury, CT 06810 USA**

* Application summary (page 4)
* Biographical sketch and bibliography for principal investigator (templates provided on pages 5-6)
* List of co-investigators, if applicable (page 7)
* Reviewer information, optional (page 8)
* Initial letter of intent (maximum one page)
* Abstract of the proposal (maximum one page)
* Brief budget outline
* Cogent reasons why NORD funding is essential (explain applicant’s particular interest in this grant)

**INCOMPLETE APPLICATIONS MAY NOT BE CONSIDERED.**

**FULL PROPOSALS**

* Full proposal invitations will be issued via email in August 2018.
* Application requirements for full proposals will accompany these invitations.

**AWARDING OF GRANT**

* Award announcements will be made via email and posted on NORD’s website in December 2018.
* Funding will begin after all necessary documents (e.g. IRB forms, patient consent forms, signed grant agreements) have been received by NORD.

**FURTHER INFORMATION**

* If the study involves human or animal subjects, copies of governance documents will be required from each site involved in the study before payment can be issued.
* Clinical drug trials must meet requirements established by the U.S. Food & Drug Administration (FDA).
* Duplicate/overlapping funds from any other private or public source are not to be used.

**ABOUT NORD**

The National Organization for Rare Disorders (NORD)® is the leading independent advocacy organization representing all patients and families affected by rare diseases. NORD is committed to the identification, treatment and cure of the 7,000 rare diseases that affect 30 million Americans, or 1 in every 10 people. For more than 30 years, NORD has led the way in voicing the needs of the rare disease community, driving supportive policies and education, advancing medical research, and providing patient and family services for those who need them most. Besides advocating for increased government research funding and referring patients to clinical trials and genetic investigations, NORD funds clinical research grants on new treatments for rare diseases.

**APPLICATION SUMMARY**

**Deadline for Initial Applications: July 16, 2018 (11:59 pm ET)**

|  |  |
| --- | --- |
| **PRINCIPAL INVESTIGATOR INFORMATION** | |
| Name |  |
| Position/Title |  |
| Email |  |
| Mailing Address |  |
| Telephone |  |
| **PROPOSAL INFORMATION** | |
| Project Title |  |
| Project Term | [ ] 1 YEAR  [ ] 2 YEARS |
| Funding Amount Requested ($) *not to exceed $45,000 U.S.* |  |
| Institution(s) where research will be conducted |  |
| City, State/Province, Country of Institution(s) |  |
| Will research involve human subjects? | [ ] YES  [ ] NO |
| Will research involve animals? | [ ] YES  [ ] NO |
| How did you hear about this RFP? | NORD Member Organization Website Posting  Email Subscription Service Referral from Colleague  Medical/Research Publication  Other (please specify below) |
| Please be as specific as possible: |  |
| Principal Investigator Signature  **REQUIRED** |  |

**Biographical Sketch**

Please provide a biographical sketch and bibliography for the principal investigator. Applicant may use this form or the NIH Biosketch form. Please modify the form to include, when applicable, the following:

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name**  Position/Title |  | | | |
| **Education/Training**  Begin with baccalaureate or other initial professional education and include postdoctoral training. | Institution and Location | Degree | Year(s) | Field of Study |
|  |  |  |  |
| **Research and Professional Experience**  Concluding with present position, list in chronological order previous employment, experience, and honors. Include present membership on any advisory committee. |  | | | |
| **Honors and Awards** |  | | | |

**Bibliography**

**Publications:** List in chronological order the titles, all authors, and complete references of all publications in the last three years and representative earlier publications pertinent to this application. If the list of publications exceeds two pages, select the most pertinent publications.

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**Co-Investigator(s)**

|  |  |
| --- | --- |
| Name of Co-Investigator |  |
| Position/Title |  |
| Institution |  |
| Email |  |

|  |  |
| --- | --- |
| Name of Co-Investigator |  |
| Position/Title |  |
| Institution |  |
| Email |  |

|  |  |
| --- | --- |
| Name of Co-Investigator |  |
| Position/Title |  |
| Institution |  |
| Email |  |

**REVIEWer information**

**Please list up to five areas of scientific/medical expertise needed to review this application (optional).**



**Please list below any individuals who should not review this application (optional).**

|  |  |  |
| --- | --- | --- |
| **NAME** | **INSTITUTION** | **JUSTIFICATION** |
|  |  |  |
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**application checklist**

**Deadline for Initial Applications: July 16, 2018 (11:59 pm ET)**

|  |  |
| --- | --- |
| **Application summary** (page 4) |  |
| **Biographical sketch and bibliography** (templates provided on pages 5-6) |  |
| **List of co-investigators, if applicable** (page 7) |  |
| **Reviewer information, optional** (page 8) |  |
| **Initial letter of intent** (maximum one page) |  |
| **Abstract of the proposal** (maximum one page) |  |
| **Brief budget outline.** Do not include PI salary, overhead, or indirect costs. Funding can be used to cover expenses such as staff salary, technical assistance, supplies, and small equipment. |  |
| **Cogent reasons why NORD funding is essential** |  |

**To receive notification of future funding opportunities through NORD,**

**please contact** [**research@rarediseases.org**](mailto:research@rarediseases.org)