Xtraordinary Joy Foundation Research Award

Xtraordinary Joy Foundation is seeking applications for research projects that would investigate some aspects of rare chromosome deletion Xq27.3-Xq28. One project will be selected to grant $50,000 to the investigators for proposed work. The purpose of this funding is to help drive the highest quality early stage, basic and translational research for patient benefit.

This past year Xtraordinary Joy funded novel research to develop a model for understanding rare X-chromosome disorders. Induced Pluripotent Stem cells (iPSc) were created from one patient with 7.3Mb deletion at Xq27.3-Xq28 (142,271,441-149,561,309). iPSC clones have been validated for both the cells with distinct X chromosome inactivation of either deleted or intact allele (to serve as control). Two UF neuroscientists are currently differentiating these cells into neurons to study deficient pathways and synapses for targeted therapeutic options. Our overarching goal is to understand this multi-gene deletion and discover treatment possibilities. You can read about case studies of Xq27.3-Xq28 deletions here https://www.ncbi.nlm.nih.gov/pubmed/24715853 and about the foundation here http://www.xtraordinaryjoy.org/.

Investigator Eligibility: Any current research faculty at the University of Florida can submit a proposal.

Project Eligibility: We are looking for projects utilizing currently available resources (i.e. iPScs described above) and/or other approaches to make meaningful progress into studying this rare deletion. Eligible project types include by are not limited to:

- Characterization of unstudied genes within the deletion (like novel miRNAs)
- Contributions to disorders within the deletion like Fragile X Syndrome, Hunter Syndrome, FRAXE, or myotubular myopathy.
- Impacting broad symptoms of rare disorders like autism, hypotonia, sensory processing, intellectual disability, psychological disorders and epilepsy, for example.
- Areas of approach like metabolomics, gut microbiota, and gene editing, for instance.

Application process:

Step 1: LOI Submission
Mandatory letters of intent are due January 26, 2018 and should include
A. Project Title
B. PI name(s) and contact information
C. Brief description of proposal and approach
Step 2: Proposal Submission (invitation not required)
A faculty member may apply with multiple applications. Applicants should write a 5 page proposal (B-F; no page limit on A, G-I).

- A. Title, name(s) of applicants, affiliations
- B. Specific aims (1 page)
- C. Significance and impact of your overall project (1 page)
- D. Research plan (2 pages)
- E. Timeline and milestones
- F. Feasibility (up to 0.5 pages)
- G. References
- H. Biosketch (NIH format)
- I. Budget
  - Funds must be used within the 12-month period for the activities detailed in the application
  - Funds cannot be used for equipment purchases, indirect costs or faculty salaries

Important dates:

- December 8, 2018 – RFA announcement
- January 26, 2018 – Letter of intent deadline
- March 2, 2018 – Application deadline
- March 23, 2018 – Announcement of awarded proposal

Pre-submission inquiries, as well as Letters of intent and final applications should be sent electronically (as a single pdf file) to Amy Meacham (meachy23@ufl.edu)

Selection criteria:

- Medical Need and Impact
- Research plan (Experimental Approach)
- Timeline, milestones and feasibility
  - Does the applicant present a clear, logical plan to complete the proposed work within a reasonable timeline?

The reviewed proposals will be ranked by the Xtraordinary Joy Scientific Advisory Board and selection will be made. Decisions will be announced on March 23, 2018.

Funding Period Expectations:
The selected applicants are expected to submit progress reports quarterly and attend biannual meetings with Xtraordinary Joy Scientific Advisory Board for one year. Advancement of the project toward publication will be tracked following the conclusion of funding.