Research Focus Areas:

SCN2A: The FamilieSCN2A Foundation

One $61,280 grant for research to accelerate the development of therapeutic treatments and disease modifying advancements for those living with autism and/or epilepsy due to changes in the SCN2A gene. We are interested in funding innovative projects that will advance development of therapeutic treatments for SCN2A-related disorders (SRDs).

Specific areas of interest include but are not limited to:

1. Exploring safe drug options for treating SCN2A-related disorders, such as repurposing FDA-approved drugs or investigating previously shelved drugs with established clinical safety records.
2. discovery and validation of novel biomarkers for SCN2A-related disorders
3. Discovery of compensatory mechanisms that arise due to SCN2A mutations in development and evaluation of their therapeutic potential
4. Evaluation of mechanisms that lead to phenotypic variability within SCN2A variants.
   A. Screens for genetic modifiers of the SCN2A lof or gof phenotypes
   B. Characterization of autonomic dysfunction in SCN2A animal models (if possible - not sure)
   C. Preclinical efficacy and safety study of any therapeutic (repurposed, "shelved" or novel) in SCN2A animal model or iPSCs

Priority will be given to innovative projects which could potentially lead to therapeutic treatments or cures for those with SCN2A-related disorders.
In addition, applicants are encouraged to collaborate with existing SCN2A researchers and to leverage existing disease models and data (e.g. animal models, Simons Searchlight registry and biobank, CTRS, Ciitizen/Invitae data.)