

HNRNP Family Foundation

2023

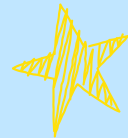
IMPACT REPORT



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INFO@HNRNP.ORG**



As we close our inaugural year, we are thrilled about the strides made possible by our dedicated families, clinicians, researchers, and nonprofit partners!



OUR EXECUTIVE BOARD

Leila Margolis Mom to Sidney, who has SYNCRIP/HNRNPQ-Related Disorder

Dr. Susan Altschuller PhD Mom to Gus, who has HNRNPU-Related Disorder

Dr. Maddie Gillentine PhD Genetic Variant Scientist with published work on HNRNP-Related Disorders

Amanda Reuther Mom to Paeyton, who has HNRNPU-Related Disorder

SCIENTIFIC MEDICAL ADVISORY BOARD

Dr. Billie Au Clinical Geneticist at University of Calgary who is an expert on Au-Kline syndrome

Dr. Jenifer Bain Neurologist at Columbia University Irving Medical Center who is an expert on HNRNPH2-Related Neurodevelopmental Disorder and movement disorders

Dr. Meena Balasubramanain Clinical Geneticist at University of Sheffield who is an expert on HNRNPU-Related Neurodevelopmental Disorder

Dr. Arjun Bouman Clinical Geneticist at Erasmus Medical Center who is an expert on HNRNPC-Related Neurodevelopmental Disorder

Dr. Christopher Ricupero Neuroscientist who is an expert on iPSC modeling of neurological disorders, including HNRNPH2-Related Neurodevelopmental Disorder

INTERNATIONAL CHAPTERS

Kaori Suzuki - HNRNP Japan





PROJECTS



DETAILS




OUTCOME

Community

- HNRNP-Family Foundation Family and Scientific Symposium
- Virtual meet and greets for families for all HNRNP-RNDDs
- Inaugural social media awareness campaign
- In-person at Columbia University Irving Medical Center in New York City, with data and biosample collection
- Families from several US states, the UK, Ireland, France, Norway, Sweden, Portugal, the Netherlands, Australia, Kenya, and Japan connected
- Over 20 families shared their journey on social media
- Expanded natural history studies, fibroblast cell lines, and samples for epigenatures
- Have created community for families to share their stories
- HNRNP.org had a 400% increase in site traffic

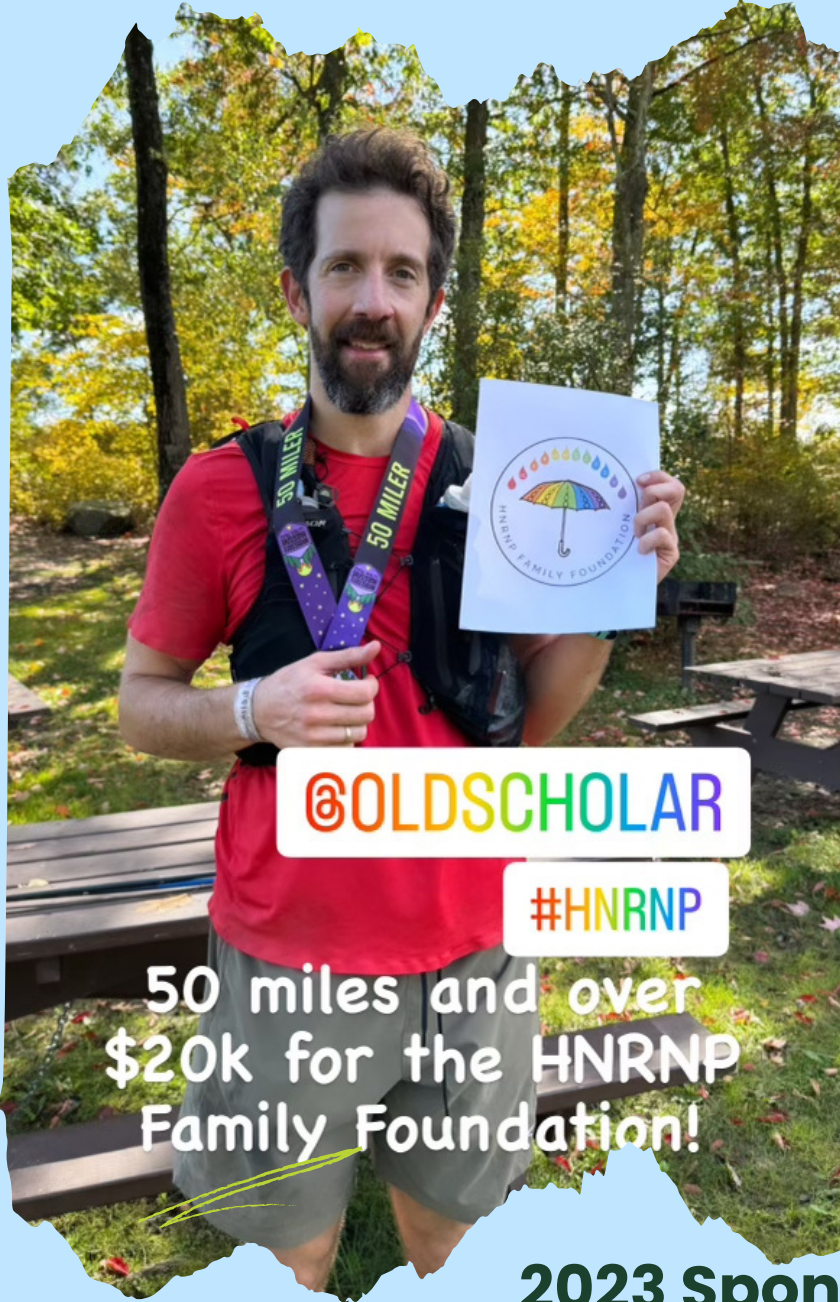
Research

- New disorders formally published
- Methylation Signatures
- Natural history expansion 
- ASO Treatment
- HNRNPC-Related Neurodevelopmental Disorder and RBMX/HNRNPG-Related Neurodevelopmental Disorders
- A new methylation signature for HNRNPU-Related Neurodevelopmental Disorder,
- Additional HNRNPH2-RNDD Rett syndrome-like features were published
- Novel disease mechanism that may allow for compensation by hnRNPH1.
- n of 1 trial for by n-Lorem of an antisense oligonucleotide (ASO) treatment for HNRNPH2-RNDD, thanks to the hard work of the Yellow Brick Road Project and their families!
- 13 HNRNPC-RNDD cases published, multiple large families for RBMX/HNRNPG-RNDDs published
- Second methylation signature after Au-Kline syndrome, will help with diagnostics and understanding disease mechanism
- Proof on concept that the HNRNP genes work together and/or for each other
- Trial started in last Quarter of 2023



Advocacy

- Submission of drafts to the National Organization for Rare Diseases (NORD)
- Conference attendance and presentations
- Addition of all HNRNP-RNDD genes to the Simon's Searchlight Project
- Currently only HNRNPU-RNDD is listed
- Global Genes Rare Advocacy Summit
- Smith Dysmorphology Meeting
- Bio TechX
- American Epilepsy Society
- The Simon's Foundation funds autism research, all HNRNP-RNDD have been formally linked to increased risk of autism
- SYNCRIP/HNRNPQ-RNDD pending
- Will allow more families to be diagnosed through the Simon's Searchlight Project and participate in research opportunities
- Increased awareness and assistance from related networks



@OLDSCHOLAR

#HNRNP

**50 miles and over
\$20k for the HNRNP
Family Foundation!**

FINANCES

Current Assets: ~\$130,000

Liabilities: None

**Operating expenses: <\$400,
most expenses donated by our
board president and everything
is run by volunteers**

Funding Summary

Donations: \$130,000

In Kind hours donated: 100 hours+

Grants/Sponsorships: \$15,900

2023 Sponsors

**SIMONS
SEARCHLIGHT**

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CRANIOFACIAL RESEARCH