Open Opportunities

There are no open opportunities at this time.

Closed Opportunities

Gene Function Studies – January 2019
Gene Function Studies – September 2019
Gene Function Studies – September 2020

Funded Projects

Institution(s)	Principal Investigator(s)	Project Title	Project Start Date
Baylor College of Medicine	Hugo J. Bellen, DVM, PhD	Understanding the role of *IRF2BPL* in neurological disease	July 1, 2019
Baylor College of Medicine	Lindsay Burrage, MD, PhD	ER stress in *TANGO2*-related metabolic encephalopathy and arrhythmias	July 1, 2019
University of Iowa	Lori Wallrath, PhD	Mechanisms of *TMEM43* muscle disease	July 1, 2019
University of Michigan	Paul C. Tang, MD, PhD	Elucidation of the Mechanism of Disease in a *TAX1BP3* gene variant associated with human Arrhythmogenic Right Ventricular Cardiomyopathy	July 1, 2019
University of Oregon	Monte Westerfield, PhD	Undiagnosed Diseases Network Gene Function Study *MAST2*	July 1, 2019
Washington University in St. Louis	Kristen Kroll, PhD	Using human pluripotent stem cell models to evaluate pathogenicity and define disease mechanisms for a *ZNF292* variant found in a UDN participant	July 1, 2019
The University of Texas MD Anderson Cancer Center	Swathi Arur, PhD	To understand the mechanism of action of the damaging *DROSHA* variant p.D1219G using *C. elegans*	July 1, 2020

Institution(s)	Principal Investigator(s)	Project Title	Project Start Date
Baylor College of Medicine	Hugo J. Bellen, DVM, PhD	Exploring the role of *WDR37* in a neurological syndrome with striking similarities to Schuurs-Hoeijmakers syndrome	July 1, 2020
Washington University in St. Louis	John A. Cooper, MD, PhD	Using zebrafish as an animal model to evaluate pathogenicity and define disease mechanisms for a *CARMIL3* variant (p.Gln928Glu)	July 1, 2020
University of California, Davis	Daniel Starr, PhD	A humanized C. elegans model to study *KLC4* kinesin light chain in disease	July 1, 2020
University of Oregon	Monte Westerfield, PhD	Undiagnosed Diseases Network Gene Function Study *PAPSS1*	July 1, 2020
Baylor College of Medicine	Shinya Yamamoto, PhD	Exploring the molecular mechanisms of Glutaminase-related neurological diseases	July 1, 2020
Baylor College of Medicine	Hugo J. Bellen, DVM, PhD and Scott Barish, PhD	Uncovering the molecular mechanisms of the chromatin remodeler *BICRA* in the nervous system	July 1, 2021
Monash University	Robert Bryson-Richardson, PhD	Identifying the mechanism of, and therapies for, *UBA5* epileptic encephalopathy	July 1, 2021
The Henry M. Jackson Foundation for the Advancement of Military Medicine, Inc. and Massachusetts General Hospital	Teresa Dunn, PhD and Florian Eichler, MD	The mechanistic basis of developmental delay due to mutations in *SPTSSA*	July 1, 2021
Washington University in St. Louis	Lilianna Solnica-Krezel, PhD	Using zebrafish and human pluripotent stem cells to evaluate pathogenicity and define disease mechanisms for a *ENY2* variant (p.K30Rfs*6)	July 1, 2021

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University of Alabama at Birmingham	Summer Thyme, PhD	Undiagnosed Diseases Network Gene Function Study *NSD2*	July 1, 2021
Rutgers University	Ching-On Wong, PhD	Delineating the signaling pathways associated with a disease-causing gain-of-function *CLCN7* variant	July 1, 2021