

Dravet Syndrome Foundation Announces 2023 Research Grant Awardees

Dravet Syndrome Foundation announced their 2023 grant awardees and is pleased to be funding 8 new research grants totaling \$2.475M this year.

New DSF Logo

CHERRY HILL, NJ, UNITED STATES, December 12, 2023 /

EINPresswire.com/ -- <u>Dravet Syndrome Foundation</u> (DSF) announced their 2023 grant awardees at their 14th annual Research Roundtable on November 30. DSF is pleased to be funding 8 new research grants totaling \$2.475M this year, including two that have been funded with support from <u>JAM for Dravet</u> (The Chang Family) and <u>Marlins for Mason</u> (The Prather Family). This brings

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This grant will support generation of an invaluable resource of clinical and genetic data to be made available to the research community to accelerate advances in the field of Dravet syndrome." *Ethan M. Goldberg MD, PhD* the total amount of research funded by DSF since 2009 to over \$9.1M. Said Executive Director, Mary Anne Meskis, "Awarding more than three times the amount of research grants compared to any previous year is a significant achievement. It reflects a strong commitment from our community to advance the field of Dravet syndrome research. Increased funding contributes to more extensive and impactful studies, potentially leading to better understanding, treatment options, and improved outcomes for individuals affected by Dravet syndrome."

Unique to 2023, DSF announced a special request for

applications focused on increasing the understanding of broader genetic impacts on the phenotypic spectrum of Dravet syndrome, with emphasis on also creating databases and bioresources that could be shared to facilitate future research. DSF awarded an unprecedented \$1,000,000, to be distributed over three years, to researchers Ingo Helbig, MD and Ethan M. Goldberg MD, PhD of The Children's Hospital of Philadelphia for their grant titled "Generating a shareable clinico-genomic data resource for Dravet syndrome." They propose to utilize whole-genome sequencing and harmonization of longitudinal clinical data from up to 500 patients with Dravet syndrome to investigate the impact of genetic variation on clinical outcomes, create shareable clinico-genomic datasets, and concurrently create the template for a learning health system approach to Dravet syndrome. "This transformative grant from the Dravet Syndrome Foundation will support generation of an invaluable resource of clinical and genetic data to be

made available to the research community to accelerate advances in the field of Dravet syndrome for patients and their families," said Dr. Goldberg.

The first-ever DSF Transformational Science Grant was funded with support from Marlins for Mason and awarded \$500,000 over three years. The collaborative project entitled "Therapeutic benefit of MGE progenitor cell transplantation in a rabbit model of Dravet syndrome," will bring together the expertise of Lori L. Isom, PhD and Jack M. Parent, MD, both of the University of Michigan, and Scott Baraban, PhD of the University of California San Francisco to test a potentially disease-modifying therapy in a novel animal model of Dravet syndrome.

DSF awarded a 2-year, \$250,000 Research Grant for the project "Abnormal ectopic action potentials in PV-INs: A novel pathophysiological mechanism in Dravet syndrome" that will investigate disruptions in a specific form of neuronal activity in inhibitory interneurons from a mouse model of Dravet syndrome. The investigator, Brian Theyel, MD, PhD is a Psychiatrist at Brown University whose research interest in basic neuroscience is targeted at understanding how unique properties of interneurons relate to neuropsychiatric diseases, with an emphasis on schizophrenia, autism spectrum disorder, and epilepsy.

Two Clinical Research Grants were awarded at \$250,000 over two years. The first entitled "Extended monitoring for cardiac arrhythmias in Dravet syndrome" was from investigators Mary Connolly MB, BCh, FRCPC(C), FRCP(I), FRCP(Edin) and Shubhayan Sanatani, MD, FRCPC of The University of British Columbia. Dr. Connolly noted that "Having subcutaneously implanted cardiac monitors in place for at least 2 years will provide long term information on cardiac rhythm during many seizures and will provide information that could potentially lead to interventions to reduce the risk of SUDEP."

The second Clinical Research Grant was awarded to Cristina Reschke, PhD of RCSI University of Medicine and Health Sciences for her proposal "DS-TIME: a multi-assessment approach to uncover the underlying mechanisms of sleep disturbances in children with Dravet syndrome." Dr. Reschke will investigate molecular markers of the circadian clock in patients with Dravet syndrome and compare them with clinical findings related to the severity of sleep disturbances.

DSF also awarded two Postdoctoral Fellowships at \$75,000 for 1 year. The first was awarded to Sophie Hill, PhD of the Children's Hospital of Philadelphia for their proposal entitled "Mechanistic investigation in a novel mouse model of early-onset SCN1A-related disorders." Under the mentorship of Ethan M. Goldberg, MD, PhD, Dr. Hill will characterize neurological abnormalities and electrical properties of a mouse model with a patient-derived gain-of-function mutation in the SCN1A gene.

The second DSF Postdoctoral Fellowship was awarded to Meiling Zhao, PhD of the University of Michigan for their proposal "Establishing the role of the noradrenergic network during seizures in Dravet syndrome." Dr. Zhao will investigate the changes in activity of neurons in lesser-explored deep brain regions in a mouse model of Dravet syndrome under the mentorship of

Joanna Mattis, MD, PhD and co-mentor Jack Parent, MD.

A Clinician-Researcher Award for \$75,000 for 1 year was awarded to Megan Abbott, MD of the University of Colorado Denver for the project "Increasing clinical trial readiness in Dravet syndrome- Creation and pilot of Dravet-specific clinical outcome measures." With mentorship from Kelly Knupp, MD MSCS and Scott Demarest, MD MSCS, Dr. Abbott will adapt an existing clinician and caregiver severity assessment developed for another rare epilepsy to be relevant for measuring meaningful changes in Dravet syndrome.

Grants are awarded through a competitive application and review process. Said DSF Scientific Director, Dr. Veronica Hood, "DSF is continuing to push the field of research forward by funding projects that can uncover novel insights into the basic neurobiology of Dravet syndrome, provide proof-of-concept for novel disease-modifying therapies, create new research tools, and further characterize the clinical spectrum of Dravet syndrome."

You can read abstracts for each of the 2023 grant awards, as well as see past projects at <u>https://dravetfoundation.org/dsf-funded-research/</u>.

DSF has funded more than 65 research projects, totaling over \$9.1M since 2009, making them the largest non-governmental funder of Dravet syndrome research, worldwide. Of those projects, over 72% have resulted in scientific publications, with DSF contributing to over 60 scientific publications either through direct involvement and/or financial support. DSF-funded researchers have subsequently received over \$30M in NIH-level funding for projects related to Dravet syndrome.

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