

FamilieSCN2A Foundation Announces Recipients of the 2024 Action Potential Awards

Megan Abbot, MD, of Children's Hospital Colorado and Kathryn Salvati, PhD, of UCSF Each Receive \$75,000 SCN2A Research Grants

GETTYSBURG, PENNSYLVANIA, UNITED STATES, August 26, 2024 /EINPresswire.com/ -- The FamilieSCN2A Foundation, the leading nonprofit organization dedicated to advancing research, education, and support for individuals and families affected by [SCN2A](#)-related disorders (SRDs), has awarded its competitive Action Potential (AP) Grant to two young investigators: Megan Abbott, MD, a pediatric neurology specialist at the Children's Hospital of Colorado; and Kathryn Salvati, PhD, a postdoctoral fellow at University of California, San Francisco (UCSF). The winners were announced at the recent FamilieSCN2A Foundation 2024 Summer Conference in Anaheim, CA.

Dr. Abbot's project aims to develop the inaugural set of outcome measures tailored specifically for SRDs. This project will focus on refining a set of clinician and caregiver-reported outcome measures originally created for CDKL5-deficiency disorder and piloting them in patients with SRD. These measures hold the potential to serve as endpoints in future clinical trials for disease-modifying therapies for SRD.

Dr. Salvati is a postdoctoral fellow in the laboratory of Dr. Kevin Bender, Professor of Neurology at Weill Institute for Neuroscience at UCSF. Dr. Salvati's project will examine how SCN2A dysfunction contributes to atypical touch sensitivity and perception in ASD. She will test sensory perception by investigating the ability of SCN2A loss-of-function mice to detect movement of their whiskers, while simultaneously imaging neurons in vivo. This project will address a fundamental gap in understanding mechanisms that lead to sensory issues in SRDs, as reported by SCN2A families, and will potentially inform future clinical trial design for SCN2A therapeutics.



Action Potential Awardee, Kathryn Salvati, PhD

“The FamilieSCN2A Action Potential grant program is designed to support preliminary investigations that may lay the groundwork for subsequent grants from government, industry, or other funding sources, including the FamilieSCN2A Foundation,” said Shawn Egan, Chief Scientific Officer of The Foundation. “This is also an investment in promising early-career SCN2A research professionals, and both of this year’s award winners look to have a very bright future ahead of them in this space”.

As part of the grant, Dr. Abbott and Dr. Salvatti will each receive \$75,000 USD of funding to further their research. The FamilieSCN2A Foundation is confident that these grants will act as a catalyst towards their vision of a world with effective treatments and cures for all SCN2A-related disorders. Previously funded projects have successfully examined various aspects of the cellular mechanisms underlying [epilepsy](#) and cognitive/behavioral deficits, the development of quantitative biomarkers, and novel gene delivery vectors.



Action Potential Awardee, Dr. Megan Abbott

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Shawn Egan, PhD, Chief Scientific Officer for the FamilieSCN2A Foundation

About The FamilieSCN2A Foundation: The mission of The FamilieSCN2A Foundation is to accelerate research, build community, and advocate to improve the lives of those affected by SCN2A-related disorders around the world. Founded in 2015 by parents of children affected by changes in the SCN2A gene, the organization has funded more than \$4.5 million in less than ten years. For more information on The FamilieSCN2A Foundation and its mission, or to donate to this cause, visit www.scn2a.org.

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