

NEWS RELEASE 6-APR-2022

SynGAP Research Fund (SRF) announces grants to Dr. Kurt Haas and Dr. Graziella Di Cristo in partnership with Canada's Rare Diseases: Models and Mechanisms Network (RDMMN)

Grant and Award Announcement

SYNGAP RESEARCH FUND



IMAGE: SRF IS A 501(C)(3) PUBLIC CHARITY INCORPORATED IN 2018. THE MISSION IS TO IMPROVE THE QUALITY OF LIFE OF SYNGAP1 PATIENTS THROUGH THE RESEARCH AND DEVELOPMENT OF TREATMENTS, THERAPIES AND SUPPORT SYSTEMS. COMPLETELY PARENT LED, SRF IS THE LARGEST NON-GOVERNMENT FUNDER OF SYNGAP RESEARCH. THE FOUNDERS COVER ALL OPERATIONAL COSTS, ALLOWING 100% OF DONATIONS TO GO TO RESEARCH. SRF'S MANTRA IS COLLABORATION, TRANSPARENCY & URGENCY. SRF IS A MEMBER OF COMBINEDBRAIN, THE PRECISION MEDICINE COALITION, THE GLOBAL GENES RARE FOUNDATION ALLIANCE & THE EVERYLIFE FOUNDATION COMMUNITY CONGRESS. LEARN MORE AT [SYNGAPRESEARCHFUND.ORG](https://syngapresearchfund.org). [view more >](#)

CREDIT: SYNGAP RESEARCH FUND

[SynGAP Research Fund](#) & [Rare Diseases Models and Mechanisms Network](#) partner to co-fund two SYNGAP1 animal model studies in the labs of Dr. Graziella Di Cristo & Dr. Kurt Haas in Canada.

Christine Oriel of RDMM says, "The Rare Diseases: Models and Mechanisms Network (RDMM) is happy to partner with SRF in co-funding these research projects. We look forward to seeing the research of Drs. Haas and Di Cristo to better understand the disease and identify and develop new therapies to treat SYNGAP1. SRF has done a lot to support SYNGAP1 research and we look forward to continuing with this partnership in the future."

Michael Graglia of SRF says, "SRF is grateful to RDMM for co-funding these critical projects on SYNGAP1. We desperately need to better understand SYNGAP1 and develop therapies for our patients. This partnership allows us to both identify high-impact projects and then leverage funds raised from the families of affected patients. I am especially grateful to the Yassıbaş family who generously supported this grant in honor of their son."

Dr. Graziella Di Cristo, Professor, Department of Neuroscience, Université de Montréal and the CHU Sainte-Justine Research Center says, "Sensory processing disorders, or the difficulty in receiving and responding to information that comes through the senses (hearing, vision, touch, etc.), are common in children with neurodevelopmental disorders. We have recently found alterations on how the brain responds to complex visual and auditory stimulations in children and in mouse models carrying mutations in the gene SYNGAP1. With this study, we hope to understand how specific neuron types contribute to these phenotypes in the mouse models. We hope that better understanding of the cellular underpinning of sensory processing disorders might help developing treatment strategies tailored and optimized towards these symptoms in SYNGAP1 patients."

Dr. Kurt Haas, Professor, Dept of Cellular and Physiological Sciences, Faculty of Medicine, University of British Columbia says, "Our lab recently reported results from our study of 57 missense mutations of SYNGAP1, many identified in individuals with Autism Spectrum Disorder (ASD). In contrast to the conventional model that SYNGAP1 mutations in ASD induce complete loss of protein function leading to haploinsufficiency, my team found that missense mutations induced selective deficits in pathways associated with either long-term potentiation (LTP) or depression (LTD) forms of synaptic plasticity. Here, we will follow up this study by directly testing how these SYNGAP1 mutations impact synaptic plasticity in neurons within intact neural circuits."

SYNGAP1-related intellectual disability (US ICD-10: F78.A1) is a rare genetic disorder caused by a variation on the SYNGAP1 gene, with nearly 1,000 diagnosed patients accounted for globally as of December 2021. It leads to several neurological issues in patients, including intellectual disability, epilepsy, autism, sleep challenges, gastro-intestinal and feeding problems, hypotonia (low muscle tone), apraxia (delayed/no speech), impulsivity and aggression. (Vlaskamp, 2019)

ABOUT SYNGAP RESEARCH FUND

SRF, incorporated in 2018, is a 501(c)(3) US public charity whose mission is to improve the quality of life of SYNGAP1 patients through the research and development of treatments, therapies and support systems. Completely parent-led, SRF is the largest non-government funder of SynGAP research having committed over \$2M in grants. The founders cover all operational costs, allowing 100% of donations to go to research. SRF's grant program awards one or two-year grants to young investigators, physician residents, and clinicians who are interested in studying SYNGAP1. SRF grants are intended to help researchers explore novel ideas and

answer questions related to the clinical aspects, therapies and/or genetic causes of SYNGAP1. SRF is a member of the [Personalized Medicine Coalition](#), [COMBINEDbrain](#), [Global Genes Foundation Alliance](#), [Everylife Foundation Community Congress](#), [Rare Epilepsy Network](#), [Innovation and Value Initiative](#) & the [Epilepsy Leadership Council](#).

Visit [SyngapResearchFund.org](https://syngapresearchfund.org) to learn more.

ABOUT RARE DISEASES MODELS & MECHANISMS NETWORK

The Rare Diseases: Models & Mechanisms Network has been established to catalyze connections between people discovering new genes in patients with rare diseases, and basic scientists who can analyze equivalent genes and pathways in model organisms. Catalyst Grants fund projects that will allow rapid confirmation of potentially disease-causing genes, and fuel pilot studies to improve understanding of how specific gene mutations cause disease. It is intended that collaborations across the Canadian biomedical community will expedite the understanding of disorders, enabling the design of new therapies to the ultimate benefit of those affected by rare diseases.

Visit rare-diseases-catalyst-network.ca/ to learn more.

Contact:

Peter Halliburton, SRF Development Director

peter@syngapresearchfund.org

Disclaimer: AAAS and EurekAlert! are not responsible for the accuracy of news releases posted to EurekAlert! by contributing institutions or for the use of any information through the EurekAlert system.

Media Contact

Michael Graglia
Syngap Research Fund
Mike@syngapresearchfund.org
Office: 650-441-4191