NEW YORK, Oct. 13, 2021 /PRNewswire/ -- The SynGAP Research Fund (SRF) announces grants to Drs. April Levin, Mustafa Sahin and Annapurna Poduri of Boston Children's Hospital (BCH) to advance SYNGAP1 biomarker characterization and genotype-phenotype analysis.

SYNGAP1-related intellectual disability is a rare genetic disorder caused by a variation on the SYNGAP1 gene, with 883 diagnosed patients accounted for globally as of October 1, 2021. It leads to several neurological issues in patients, including mild-to-severe intellectual disability, epilepsy, autism, sleep issues, hypotonia (low muscle tone), apraxia (delayed/no speech), impulsivity and aggression.

Drs. Levin and Sahin will analyze research-grade EEG recordings and biofluid and neuropsychological assessment data from a cohort of SYNGAP1 patients leveraging the Rare Diseases Clinical Research Network Developmental Synaptopathies Consortium (DSC). The primary goal of the study is to identify predictable biomarkers in SYNGAP1 patients that can be targeted by future therapies.
Dr. Levin says, "We are thrilled to be working with the SynGAP Research Fund on a vitally important effort to identify biomarkers for SYNGAP1-related disorders. The identification of clinically relevant biomarkers will provide tools needed for the development and trials of future therapies."

Dr. Poduri's research group will analyze data collected from over 100 patients in the Citizen SYNGAP1 study and aim to correlate different variants seen in patients to their clinical symptoms. This first-of-its-kind study will shed light on why the disease presents with such a wide spectrum and help set the foundation for more scientific work based on the Citizen dataset.

Dr. Poduri says, "We are delighted to have the unique opportunity to analyze data collected by the SynGAP Research Fund and Citizen. Our team is excited to use this rich dataset to delineate the relationship between the type and location of SYNGAP1 variants and the spectrum of SYNGAP1-related epilepsy and neurodevelopmental features. This is a necessary step in understanding the natural history of SYNGAP1 and developing meaningful clinical outcomes to measure the success of future therapies through clinical trials."

All collected samples and data will be stored in the BCH central repository and made available to the research community upon request. Data will be securely linked to anonymized Citizen records to enable a more comprehensive anonymized dataset for future research use.

The grant was made possible by fund-raising efforts supported by families affected by SYNGAP1, ranging from Facebook birthday fundraisers to Ironman events. SRF is particularly grateful for significant support from the Hillside Foundation.

Michael Graglia, Managing Director of SRF says, "This work is critical to better understanding SYNGAP1 disorder and improving clinical trial design and effectiveness. We are tracking therapeutic development in various biopharma and academic labs, and expect to see trials in the coming years. SRF is laser focused on de-risking trials for industry via this grant, iPSC generation and preparing patient data.

This project underscores the importance of our partnership with Citizen, now part of Invitae. The rich data we have and continue to collect is a key input to this study."

ABOUT SYNGAP RESEARCH FUND
SRF, incorporated in 2018, is a 501(c)(3) 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 $1.8M in grants. The founders cover all operational costs, allowing 100% of donations to go to research. SRF is a member of the Personalized Medicine Coalition, COMBINEDbrain, Global Genes Foundation Alliance and the Everylife Foundation Community Congress. Visit SyngapResearchFund.org

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