CSNK2A1 Foundation Awards Grant to TGEN: Dr. Huentelman to Study Patient-Derived Stem Cells to Advance OCNDS Research

SAN FRANCISCO, CA, USA, December 27, 2023 /EINPresswire.com/ -- CSNK2A1 Foundation, a leading rare disease organization, is pleased to announce the award of a research grant to the Translational Genomics Research Institute (TGen), part of City of Hope, to further the understanding of Okur-Chung Neurodevelopmental Syndrome (OCNDS). This partnership aims to improve our understanding of how this rare genetic syndrome affects development and will accelerate the discovery of potential treatments.

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> *Dr. Gabrielle Rushing, the CSNK2A1 Foundation Science Program Director*

Gabrielle Rushing, Ph.D., the CSNK2A1 Foundation Science Program Director, expressed her enthusiasm for the collaboration, stating, "We are thrilled to partner with TGen in pursuit of our shared mission to improve the lives of individuals living with OCNDS. This research grant represents an important step forward in our efforts to unravel the complexities of this syndrome."

First described in 2016, OCNDS is an ultra-rare genetic syndrome caused by a mutation on chromosome 20 in the CSNK2A1 gene. The CSNK2A1 gene produces an important subunit of a protein called CK2 that is present in all cells of the body. OCNDS affects every aspect of life, with symptoms ranging from mild to severe and can include the inability to speak or delayed speech, global developmental delay, epilepsy, autism spectrum disorder traits, behavioral challenges, hypotonia, feeding difficulty, and severely disrupted sleep pattern. To date, there is no treatment for OCNDS.

The research project, led by Matthew Huentelman, Ph.D., Director of TGen's Neurogenomics Division, will focus on characterizing four OCNDS patient-derived induced pluripotent stem cell (iPSC) lines. Huentelman explains, "Our team will analyze various time points, neuronal

precursor cells (NPC), and early/mid/late neuron differentiation stages at 6 weeks, 8 weeks, and 12 weeks. We will employ state-of-the-art techniques, such as phosphoproteomics, to identify the targets of CK2 in the cells. Additionally, RNA sequencing will provide insights into gene expression patterns and potential dysregulations specific to OCNDS."

This research builds upon the Foundation's previous efforts to support OCNDS studies. In 2019, TGen established an OCNDS Research Program to facilitate advancements in understanding and treating OCNDS. TGen has been at the forefront of these research initiatives. "We are dedicated to making tangible progress in the pursuit of effective treatments for OCNDS," says Huentelman. "This collaboration with the CSNK2A1 Foundation allows us to leverage our expertise in genomics and stem cell biology to develop rigorous experiments in a controlled setting."

Vinodh Narayanan, M.D., Medical Director of TGen's Center for Rare Childhood Disorders, also expressed his support for the collaboration, stating, "We are committed to translating scientific discoveries into meaningful interventions for patients. The CSNK2A1 Foundation's support provides critical resources to further our understanding of OCNDS and potentially uncover new therapeutic avenues."

The CSNK2A1 Foundation and TGen's collaboration represents a significant stride towards improving the lives of individuals affected by OCNDS. By combining their expertise and resources, the partnership aims to accelerate research efforts and bring innovative treatment options to those in need.

For more information about CSNK2A1 Foundation and their ongoing initiatives to support OCNDS research, please visit www.csnk2a1foundation.org.

About CSNK2A1 Foundation
CSNK2A1 Foundation is focused on finding a cure for Okur-Chung Neurodevelopmental Syndrome and ensuring affected individuals have the opportunities and supports necessary for happy and full lives. CSNK2A1 Foundation is a 501(c)(3) non-profit organization operated and funded by a committed team of volunteers, advocates, and researchers. To date, the Foundation has registered over 210 individuals with OCNDS in its patient registry worldwide.
www.csnk2a1foundation.org

About TGen, part of City of Hope
Translational Genomics Research Institute (TGen) is a Phoenix, Arizona-based nonprofit organization dedicated to conducting groundbreaking research with life-changing results. TGen is part of City of Hope, a world-renowned independent research and treatment center for cancer, diabetes and other life-threatening diseases. This precision medicine affiliation enables both institutes to complement each other in research and patient care, with City of Hope providing a significant clinical setting to advance scientific discoveries made by TGen. TGen is focused on helping patients with neurological disorders, cancer, diabetes and infectious diseases through cutting-edge translational research (the process of rapidly moving research toward patient

benefit). TGen physicians and scientists work to unravel the genetic components of both common and complex rare diseases in adults and children. Working with collaborators in the scientific and medical communities worldwide, TGen makes a substantial contribution to help patients through efficiency and effectiveness of the translational process. www.TGEN.org

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