Foundation for USP7 Related Diseases Announces Grant to Neurolentech in Search for Cure to Rare Disease Hao-Fountain Syndrome

FALMOUTH, Maine, Apr. 2, 2021 -- The Foundation for USP7 Related Diseases (usp7.org) today announced a new research grant award to Neurolentech in a search for a cure to the rare disease Hao-Fountain Syndrome.

The Austrian startup Neurolentech, led by Carsten Pfeffer, Ph.D. and co-founded by Gaia Novarino, Professor for Neuroscience at Institute of Science and Technology Austria (IST Austria), will receive $100,000 over two years towards funding studies that will focus on modeling Hao-Fountain Syndrome in cells.

Hao-Fountain Syndrome is a rare genetic disorder caused by mutations in the USP7 gene, with just 80 known patients worldwide. It leads to autism spectrum disorder, increased prevalence of epilepsy, developmental delay/intellectual disability, abnormal brain MRIs, speech impairment, hypotonia, gastrointestinal issues, and eye anomalies.

Neurolentech will develop and characterize neuronal cultures with USP7 gene mutations using transcriptomics, morphology, and electrophysiology, in order to seek biomarkers in those models for potential drug treatments. Dr. Novarino's lab at IST Austria will analyze a haploinsufficient USP7 mouse model to gain a better understanding of the biological mechanisms underlying Hao-Fountain Syndrome.

This grant is the result of years of fundraising from the Foundation for USP7 Related Diseases, including hundreds of donations from all over the world, including from its sister organization in France, Manger La Vie, which also seeks to cure Hao-Fountain Syndrome.

"We are humbled to work with the inspiring families of the Foundation for USP7 Related Diseases towards a better understanding of the molecular and cellular disorder pathology and we fully support their aim of finding treatments for Hao-Fountain Syndrome," said Dr. Pfeffer. Dr. Novarino added, "Basic research into the mechanisms of neurodevelopmental disorders like Hao-Fountain Syndrome will provide the most promising approach to help the patients. We are thrilled to work together with patients and their families in order to advance the best research for USP7 related diseases. The only way to deal with not knowing is to start investigating."

Bo Bigelow, chairman and co-founder of the Foundation for USP7 Related Diseases said, "We started this organization to move quickly in investigating the mechanisms of USP7 and what's causing Hao-Fountain Syndrome, so we can get to a cure or treatment. We are delighted to discover brilliant collaborators like Dr. Novarino and Dr. Pfeffer, who share our sense of urgency, and we're thrilled to partner with their amazing startup."

ABOUT THE FOUNDATION FOR USP7 RELATED DISEASES

The Foundation for USP7 Related Diseases was started in 2017 by a group of parents of affected children. It is a registered 501(c)(3) nonprofit organization. Its mission is to cure Hao-Fountain Syndrome by finding patients and funding research. It is a member of COMBINEDBrain, NORD, The Global Genes RARE Foundation Alliance, and the Rare Epilepsy Network. For more information, visit usp7.org.

ABOUT NEUROLENTECH

Neurolentech is a spinoff company from IST-Austria started by Professor Gaia Novarino and Carsten Pfeffer in December 2020. The startup's goal is to fight neurodevelopmental disorders, through developing patient-specific cellular models for detailed investigation of the disease mechanisms and further development of diagnostics and therapies. The Neurolentech team is supported by funds from the Austrian federal promotional bank (AWS).