CMTA Awards $300,000 for Gene Discovery Work

GLENOLDEN, PENNSYLVANIA, USA, May 19, 2021 /EINPresswire.com/ -- The Charcot-Marie-Tooth Association (CMTA) awarded Dr. Stephan Züchner and his team at the University of Miami $300,000 for a project aimed at increasing understanding of the genes that cause CMT, a critical step in the drug development process.

The three-year project will support data sharing, curation of existing data, and the development of web-based access for CMT patients to participate in genomic studies. In addition, the funding will support the implementation and validation of a machine-learning algorithm for CMT genetic variations.

CMT is a degenerative neuromuscular disease that kills the long, or peripheral, nerves to the hands and feet. As the nerves die, the muscles around them follow suit. All of the CMTA's research efforts are consolidated under the banner of STAR—which brings together the world's largest network of biotech research partners, research scientists, clinicians and patients—and funds more CMT grants than any other philanthropic organization to increase the likelihood of finding a cure. Since 2008, the CMTA has invested more than $17 million in STAR, with plans to invest another $10 million in the next few years.
The first generation of genetic therapies for neuromuscular diseases is now available (i.e. SMA, hereditary amyloidosis with neuropathy) or in clinical trials (i.e. GAN), and a flurry of similar projects will begin soon. There is a new urgency to uncover the complete genetic basis of inherited neuromuscular diseases, a prerequisite to finding treatments. CMT is one of the most common inherited disorders in neurology, affecting one in 2500 individuals, yet less than 50 percent of CMT2 patients receive a genetic diagnosis.

Dr. Züchner's previous work has contributed to the discovery of more than 25 novel, mostly axonal, Type 2 genes. He and his team have built extensive discovery resources, including the largest collection of CMT exomes/genomes and GENESIS, a data analysis platform that allows for real-time data sharing and genetic matchmaking

Building out the CMTA's bioinformatics abilities, which are available to the Inherited Neuropathy Consortium (INC) and other CMT collaborators in GENESIS, will result in a continued high pace of gene/allele discovery. Züchner and his team recently identified mutations in the SORD gene that cause a recessive form of CMT2.

The future focus will be on advanced techniques such as machine learning, non-coding space, complex structural variation and more. These cutting-edge techniques will ultimately allow for better diagnosis of patients with unknown variants and support important gene therapy projects.

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