# CMT Research Foundation Partners to Advance Study of CMT Type 1J by Dr. Stephan Zuchner

*Resulting model will be available to other investigators to study CMT1J*

CMT Research Foundation

ATLANTA, GA, USA, February 20, 2024 /EINPresswire.com/ -- The CMT Research Foundation, a non-profit focused solely on delivering treatments and cures for Charcot-Marie-Tooth disease (CMT)*, has partnered with the 1J Foundation, a 501(c)(3) organization dedicated to finding a cure for patients with 1J, a newly identified subtype of CMT. The two Foundations will co-fund the development of an ITPR3 gene mutation mouse model of CMT1J under the direction of Stephan Zuchner, MD, PhD at the University of Miami, FL.

> This is a big step towards finding a therapy for 1J patients and in alignment with CMTRF's mission to resolve ALL forms of CMT."
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> *Cleary Simpson, CEO of CMTRF*

The ITPR3 gene is responsible for making a protein heavily abundant in the nervous system, especially in Schwann cells. It works as a calcium channel, controlling the flow of calcium ions in the cell. The researchers hypothesize that a specific mutation in the ITPR3 gene in CMT1J patients may interfere with the normal function of the protein, affecting calcium flow and causing issues in Schwann cells. Dr. Zuchner and his team will develop a new mouse model that contains the human ITPR3 mutation(s) which will then be used to study affected biological pathways.

This model will be available to other investigators to study CMT1J.

Other team members on this project include Dr. Mario Saporta, MD, PhD, Associate Professor at the University of Miami, Co-Investigator; Katherina Walz, Ph.D., Associate Professor at the University of Miami, Co-Investigator; Adriana Rebelo, PhD, Scientist at the University of Miami; and Clemer Abad, D.V.D, Manager, Research Support. Dr. Rob Burgess from the Jackson Laboratory has been instrumental in creating the founder animals and will collaborate with the U of Miami group on this project.

“Dr. Zuchner has a well-established track record in neurology, peripheral nerve neurology, and molecular genetics,“ says Cleary Simpson, CEO of CMTRF. “We are pleased to help further his quest to find a cure in partnership with the 1J Foundation. This is a big step towards finding a therapy for 1J patients and in alignment with CMTRF's mission to resolve ALL forms of CMT.”

1J Foundation is a patient-led non-profit to fund treatment for CMT1J, one of the newest types identified. Variability in severity is one of the hallmarks of CMT1J. Symptoms can appear at any age, from infancy to late adulthood, and severity ranges from asymptomatic to fatal. Those who have symptom onset during childhood have more severe disease, so there is an urgent need to find treatment.

CMT Research Foundation (CMTRF) is a patient-led, non-profit focused on delivering treatments and cures for CMT. The foundation identifies significant obstacles or deficiencies impeding progress toward a cure and seeks out collaborators to address these issues. To date, CMTRF has funded 21 projects, of which 6 are completed.  Of those 6 completed projects, 5 have clinical candidates. CMTRF's mission to invest in promising science with high potential of leading to treatments and cures was proven effective and ground-breaking when DTx Pharma with a CMTRF-backed program as its lead candidate was acquired by Novartis for $1 billion. Founded by two patients who are driven to expedite drug delivery to people who live with CMT, the 501(c)(3) federal tax-exempt organization is supported by personal and corporate financial gifts.

*Charcot-Marie-Tooth encompasses a group of inherited, chronic peripheral neuropathies that result in nerve degradation. CMT patients suffer from progressive muscle atrophy of legs and arms, causing walking, running and balance problems as well as abnormal functioning of hands and feet. CMT affects one in 2,500 people (about the same prevalence as cystic fibrosis), including 150,000 Americans and nearly 3 million people worldwide. At the moment, there is no treatment or cure for CMT.

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