CMTA Fuels Breakthrough Imaging Research to Address CMT2A's Cellular Weakness

CMTA funds $90,860 research using AI-driven imaging to track cellular defects in CMT2A, accelerating therapeutic discovery for this axonal form of CMT.

GLENOLDEN, PA, UNITED STATES, March 18, 2025 /EINPresswire.com/ -- The Charcot-Marie-Tooth Association (CMTA), the largest philanthropic funder of Charcot-Marie-Tooth disease (CMT) research, announced a $90,860 investment in a two-year research project at the University of California, San Diego (UC San Diego) to investigate the cellular mechanisms of CMT2A and create new tools to accelerate drug development. Led by Uri Manor, PhD, Assistant Professor in the Department of Cell & Developmental Biology, and Director of the Goeddel Family Technology Sandbox, the study will use deep learning-based microscopy to analyze cellular defects in CMT2A.

> By leveraging cutting-edge imaging tools and AI-driven microscopy, we aim to develop a platform for identifying disease-related defects and screening potential therapies."
>
> *Dr. Uri Manor*

CMT2A, the most common axonal form of CMT, is caused by MFN2 mutations that disrupt cellular transport in peripheral nerves, with symptoms typically beginning in early childhood. Proper organelle movement is critical for neuronal function, and disruptions lead to nerve degeneration and worsening symptoms. This project will develop high-resolution imaging assays to track organelle movement in neurons derived from CMT2A patients and a genetically engineered animal model carrying the MFN2 R364W mutation. These assays will allow direct visualization of the impact of a therapeutic molecule on these key CMT2A models, creating a rapid system to screen their potential utility in patients.

"Our research is designed to uncover the cellular mechanisms underlying CMT2A and lay the groundwork for targeted therapeutic strategies," said Dr. Manor. "By leveraging cutting-edge imaging tools and artificial intelligence-driven microscopy, we aim to develop a platform for identifying disease-related defects and screening potential therapies. This work has the potential to reshape CMT research and open new possibilities for future treatments."

"Understanding how MFN2 mutations disrupt cellular transport in CMT2A is a critical piece of the puzzle," said Katherine Forsey, PhD, CMTA's Chief Research Officer. "CMTA's Strategy To Accelerate Research (CMTA-STAR) goes beyond funding. It provides scientists with the resources to develop tools that visualize these disruptions and supports them to identify potential therapeutic targets. Dr. Manor's imaging approach could open new avenues for treatment development for CMT2A, and once established, the approach could be replicated for other axonal types of CMT. CMTA is proud to support research that moves us closer to real treatment solutions across the CMT community."

About CMT

Named after the three doctors who first described it in 1886: Charcot, Marie, and Tooth, CMT affects one in every 2,500 people. This rare disease has multiple subtypes, each with a lower prevalence. People with CMT experience progressive sensory loss, muscle weakness, and atrophy in the arms and legs, along with impaired balance, mobility, hand function, and more. There is currently no treatment or cure for this debilitating disease.

About The Charcot-Marie-Tooth Association

CMTA is a community-led, community-driven 501(c)(3) nonprofit organization with a mission to support the development of new treatments for CMT, to improve the quality of life for people with CMT, and, ultimately, to find a cure. As the leading global philanthropic funder of CMT research, CMTA unites the community with clinicians and industry experts to accelerate the advancement of treatments, with investments of more than $30 million since 2008. For more information, visit cmtausa.org

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