CMTA Invests $210K in Long-Read Sequencing to Accelerate CMT Gene Discovery

New CMTA-STAR project aims to uncover elusive mutations in CMT through long-read sequencing, helping more patients reach a genetic diagnosis.

GLENOLDEN, PA, UNITED STATES, April 22, 2025 /EINPresswire.com/ -- The Charcot-Marie-Tooth Association (CMTA), the largest philanthropic funder of Charcot-Marie-Tooth (CMT) disease research, announced today a $210,000 investment in advanced gene discovery. This two-year project, led by Andrea Cortese, MD, PhD, at University College London, leverages cutting-edge long-read genome sequencing to identify complex genetic mutations in undiagnosed axonal CMT patients.

> We're investing in long-read sequencing to empower the CMT community with answers and lay the groundwork for future treatments."
>
> *Katherine Forsey, PhD, CMTA Chief Research Officer*

Axonal forms of CMT, such as CMT2 and distal hereditary motor neuropathy (dHMN and HMN), along with intermediate forms of CMT, are notoriously difficult to diagnose, with up to 50% of patients lacking genetic confirmation. This study aims to address this gap by identifying hidden gene mutations that traditional genetic testing can't detect.

"Despite significant advances in genetic diagnostics, a substantial portion of axonal CMT cases remain undiagnosed," said Dr. Cortese. "Thanks to CMTA's support, we believe this project will dramatically improve gene discovery, access to care, and participation in future clinical trials for these patients and their families."

"This CMTA Strategy To Accelerate Research (CMTA-STAR) initiative demonstrates our commitment to addressing the needs of a significant portion of the CMT community who remain without a genetic diagnosis," said Katherine Forsey, PhD, CMTA Chief Research Officer. "By investing in advanced long-read sequencing technology and supporting a global network of

researchers through this collaborative project, we're empowering the CMT community with answers and laying the groundwork for new treatments."

By accelerating long-read sequencing, this project seeks to improve diagnostic rates and establish this technology as a new standard for CMT genetic testing when conventional methods have not provided definitive answers. The findings could have far-reaching implications for patient care, including enabling more precise treatment approaches and opening the door to new therapeutic options. This project would not be possible without the community's unwavering support that drives every breakthrough.

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 CMTA

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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