CMTA Invests $423K in Transformative Genomic Research to Advance CMT Diagnostics

CMTA invests $423K in genomic research to close CMT diagnostic gaps, advance gene discovery, and expand the GENESIS platform, driving progress toward a cure.

GLENOLDEN, PA, UNITED STATES, February 4, 2025 /EINPresswire.com/ -- The Charcot-Marie-Tooth Association (CMTA), the largest philanthropic funder of Charcot-Marie-Tooth (CMT) disease research, announced today a $423,000 investment in "Solve-CMT," an advanced CMT genomic science initiative designed to address diagnostic gaps and drive new CMT gene discoveries. Led by CMTA Strategy To Accelerate Research (STAR) Advisory Board Member Stephan Züchner, MD, PhD, at The GENESIS Project Foundation, this three-year project demonstrates CMTA's commitment to accelerating precision diagnostics and understanding CMT.

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> *Dr. Katherine Forsey, CMTA Chief Research Officer*

Despite significant progress in this area, up to 50% of individuals with CMT still lack a genetic diagnosis, with the underlying causes remaining unknown. Through this project, the GENESIS team will address this critical diagnostic gap by incorporating thousands of new genomic datasets into the GENESIS platform, including long-read genomes and exomes. Long-read sequencing enables researchers to uncover complex genetic variations that are challenging to detect with traditional methods. These datasets will be analyzed with advanced computational tools, including artificial intelligence (AI), accelerating gene discovery and improving diagnostic outcomes.

"Data sharing and advanced genomics have been transformative for CMT research, but there is still much work to do," said Dr. Züchner. "CMTA has been an incredible partner in supporting the GENESIS database, a game-changer for CMT genetics over the past decade. This project will

expand GENESIS, improve diagnostic success for undiagnosed CMT, and empower researchers worldwide. Together, we aim to continue delivering new CMT gene discoveries while driving progress toward better treatments."

"This CMTA-STAR initiative is the next step in our partnership with GENESIS, which has already led to the discovery of 30 CMT genes, including the transformative SORD gene discovery," said Katherine Forsey, PhD, CMTA Chief Research Officer. "Expanding this resource isn't just about solving diagnostic puzzles; it's about uncovering the causes of CMT, delivering answers to families, and equipping the community with knowledge that brings us closer to cures. None of this would be possible without the unwavering support of the CMT community, whose commitment to CMTA-STAR drives every breakthrough and brings us closer to a world without CMT."

This latest investment reflects CMTA-STAR's collaborative research model, which unites scientists, clinicians, and the patient community to drive breakthroughs in CMT research. By advancing precision diagnostics and accelerating gene discovery, CMTA continues to address urgent diagnostic gaps while building the foundation for a future with effective treatments and progress toward a cure.

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—a rare disease subdivided into multiple subtypes, each with a lower prevalence. Due to the degradation of their nerves, people with CMT suffer lifelong progressive muscle weakness and atrophy of the arms and legs, and can affect other parts of the body. This leads to problems with balance, walking, hand use, and more. There currently is 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 nearly $30 million since 2008. For more information, visit cmtausa.org

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