

CMTA Awards \$225,483 for Study on Using ASOs to Treat CMT2E

FOR IMMEDIATE RELEASE

GLENOLDEN, PA, UNITED STATES, SEPTEMBER 28, 2023—The Charcot-Marie-Tooth Association (CMTA) Board of Directors announced Sept. 28 that it has approved a \$225,483 award to Dr. Mario Saporta at the University of Miami for a study on the use of antisense oligonucleotides (ASOs) to treat CMT2E.

CMT is one of the most commonly inherited peripheral neuropathies, affecting the long nerves that go to the feet and hands of one in 2,500 people, or 3 million people, worldwide. As the nerves die, the muscles around them wither. Despite its prevalence, it's often misdiagnosed or not diagnosed at all.

CMT2E is caused by single mutations in one copy of the Neurofilament light chain gene (NfL). Saporta's approach uses antisense oligonucleotides to target the mutant NfL copy, leaving the normal gene copy intact to perform its function.

ASOs are a type of medication that works by altering how specific genes in the body are expressed, or turned from DNA into a functional protein. They are short man-made pieces of genetic material designed to bind to a specific target messenger, RNA (mRNA).

The Food and Drug Administration has approved ASO-based treatments for several neuromuscular conditions, including spinal muscular atrophy (SMA) and familial amyloid neuropathy. The study uses the same chemistry as the SMA treatment (Nusinersen) to accelerate its potential use in patients. Nusinersen has been used since 2016 and demonstrates a very favorable safety profile.

The study will provide further understanding on how neurofilament levels impact axonal health, which may have implications for other forms of CMT. If it is feasible and effective, this approach should support the development of treatments for several forms of CMT type 2, using patient-derived cells and customized genetic treatments to treat patients with ultra-rare forms of CMT.

Dr. Saporta said, “We are very excited to continue this long-standing collaboration with the CMTA. This project has been supported by the CMTA-STAR initiative since day one and foundational work to characterize the human cellular model we are now using to validate our treatment strategy was a product of previous grants received from the CMTA. We look forward to seeing this treatment approach advance into animal models and then patients.”

The CMTA is a patient-led nonprofit that is aggressively searching out a treatment for CMT by putting together a group of top-flight researchers and experts and directly funding their work. The CMTA has invested \$23.5 million in its Strategy to Accelerate Research (STAR) since it was implemented in 2008, making it the world’s largest funder of CMT research.

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For Media Inquiries:

Kenny Raymond

Head of Communications

The Charcot-Marie-Tooth Association

Email: kenny@cmtausa.org

Phone: +1 734-862-8702