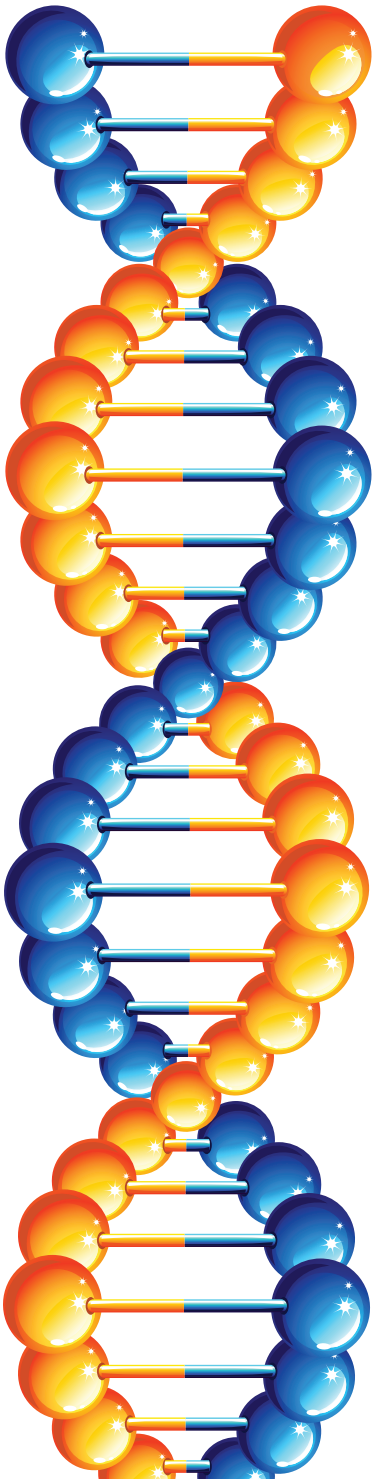
CMTA
Charcot-Marie-Tooth Association
CMTAUSA.ORG

TYPE 2
RESEARCH DEVELOPMENTS



GENE THERAPY

Progress: One of the most exciting clinical developments has been the development of gene therapy for Spinal Muscular Atrophy (SMA), which affects the same motor neurons that are affected in CMT2. The CMTA has supported pilot studies of gene therapy in CMT mouse models and convened a meeting of gene therapy experts to outline the next steps in bringing this therapeutic strategy to CMT2. We have recruited leaders in the gene therapy field to our scientific advisory board, who are guiding our efforts in this area. We are also partnering with an eminent expert in the new technology of genome editing to explore the application of this therapeutic approach to CMT2A and CMT2E.

Next Phase: Support the first single-patient clinical trial of gene therapy for CMT2 in 2020, and extend this therapy to more common forms of CMT2 by the end of 2021.

INHIBITION OF AXON DEGENERATION

Progress: The progression of all types of CMT occurs as the longest axons are lost in a process termed axon degeneration. CMTA partners are working on developing chemical inhibitors of recently identified biochemical triggers of axon degeneration. The CMTA plans to explore the applicability of this recent technology to the multiple forms of CMT2 using the many models of CMT that have been generated within our network.

Next Phase: Test the applicability to CMT2A and CMT2E by the end of 2019, leading to phase 1 clinical trials by 2021.

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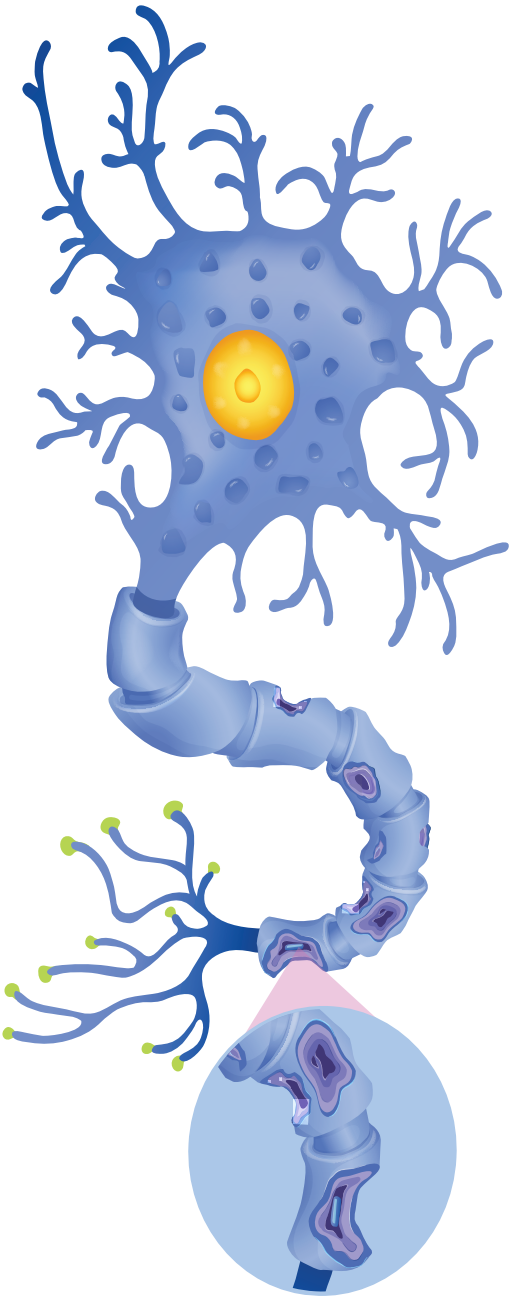

SMALL MOLECULE THERAPIES

Progress: The most common cause of CMT2A is mutation of the Mitofusin 2 gene. Researchers have recently identified custom-designed molecules that can stimulate the activity of mitofusin proteins. We are seeking to promote development of this therapy by testing for efficacy in our recently developed rat models of CMT2A.

Next Phase: Complete the first evaluations of these compounds in our CMT2A rats by 2020, leading to phase 1 clinical trial as early as 2021.

IDENTIFICATION OF NEW GENES & UNDIAGNOSED TYPE 2*s*

Approximately 50 percent of CMT2 patients do not yet have a definitive genetic diagnosis. Dr. Stephan Züchner at the University of Miami is working to change that, spearheading an ambitious project to identify new disease-causing mutations in patients seen in the Inherited Neuropathies Consortium.

CROSSOVER EFFORTS

Stem cell studies are needed to make human neurons based on cells obtained from CMT2A and CMT2E patients.

Biomarkers and outcome measures are needed for clinical trials that will make CMT an ideal target for therapeutic development.

In total, the CMTA will need $5 million in the next four years to implement our Type 2 strategy. If you or a loved one has Type 2, we are asking you to help us as we keep reaching for our goals.

Donate at: cmtausa.org/wearefamily

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