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## **CMTA Seed Money Draws NIH Support for Type 2 Gene Editing, Neurofilament Projects**

### **FOR IMMEDIATE RELEASE**

**GLENOLDEN, PA, UNITED STATES, DECEMBER 14, 2021**—The Charcot-Marie-Tooth Association-Strategy to Accelerate Research (CMTA-STAR) today announced that the National Institutes of Health (NIH) awarded additional funding to two Type 2 projects initiated with CMTA seed money.

In 2020, the CMTA awarded Drs. Bruce Conklin and Luke Judge of the Gladstone Institutes and the University of California San Francisco Departments of Medicine and Pediatrics \$653,000 to develop the gene-editing technique known as CRISPR for CMT2A, 2E and 2F.

Based on that work, the NIH awarded the researchers an R01 grant for their proposal to develop and validate a therapeutic gene editing platform for dominant CMT2E mutations using human iPSC-based models. They will test mutation-specific editing for two different NEFL gene mutations and develop rigorous phenotypic assays for therapeutic effect in human iPSC-derived motor neurons. Additionally, they will identify sites of common human genetic variation that can be targeted to excise protein coding or critical regulatory regions and inactivate the disease allele in the majority of patients, regardless of their individual mutations. These studies will also provide proof-of-concept for a strategic approach that can be applied to other forms of dominant CMT2.

Also in 2020, the CMTA awarded the Ohio State University labs of Anthony Brown, PhD; Arthur Burghes, PhD; Kathrin Meyer, PhD and W. David Arnold, MD, \$265,000 to evaluate gene therapy strategies for restoring neurofilaments to diseased neurons in Type 2 mice.

Brown, a professor of neuroscience, was able to leverage pilot data obtained with the CMTA's grant to secure NIH funding for a project on restoring neurofilaments to axons in a mouse model of CMT2E. The NIH award will enable Brown and Arnold, his co-principal investigator and an associate professor of neurology, to establish "proof of principle" of a gene therapy strategy in a mouse model of recessive CMT2E. The work will inform a more general therapeutic strategy for dominant CMT2E.

CMTA CEO Amy Gray said the NIH grants embody a key component of the CMTA's Strategy to Accelerate Research (STAR)—providing seed money for projects that can then be leveraged to raise funds from other entities.

## **ABOUT CMT**

CMT is a group of diseases caused by inherited genetic mutations that damage the peripheral nerves outside of the brain and spinal cord. Scientists have identified over 100 different gene mutations causing CMT. Most people (90 percent) have one of four types of CMT: CMT 1A (PMP 22); CMT 1B (MPZ); CMT 2A (MFN2) and CMT 1X (GJB1). It is estimated that CMT affects more than 3 million people worldwide, regardless of gender, race, or ethnicity. <https://www.cmtausa.org/understanding-cmt/what-is-cmt/>

## **ABOUT THE CMTA**

The CMTA is the largest philanthropic funder of CMT research worldwide. The CMTA's Strategy to Accelerate Research (STAR) brings the best CMT researchers, clinicians, and experts in therapy development together with pharmaceutical and biotechnology companies and patients to expedite the development of treatments for CMT. The CMTA is also actively working to help improve the quality of life for all families living with CMT by offering educational programs and materials, hosting patient and professional conferences, providing support to families through its nationwide branch system and more. More information can be found at [www.cmtausa.org](http://www.cmtausa.org).

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