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FOR IMMEDIATE RELEASE

**STAR™ INITIATIVE UNITES INTERNATIONAL SCIENTIFIC COMMUNITY  
TO SEEK TREATMENT AND CURE FOR LITTLE KNOWN DISORDER**

**--Charcot-Marie-Tooth Association Launches Strategy to Accelerate Research--**

**--\$10 Million Needed to Support STAR Efforts Over Next Five Years--**

**CHESTER, PA**—The Charcot-Marie-Tooth Association (CMTA) is launching an initiative, known as the Strategy to Accelerate Research™ (STAR), that is designed to lead to new treatments for Charcot-Marie-Tooth disorder (CMT), and a cure within ten years.

CMT is the most commonly inherited peripheral neuropathy estimated to affect one in every 2,500 people—approximately 2.6 million people worldwide. CMT is a progressive disorder that causes the nerve cells leading to the body's extremities to slowly degenerate resulting in the loss of normal use of the feet/legs and hands/arms. It may also lead to a loss of sensory nerve function and deformities. CMT strikes people of all ages, genders, races and ethnicities.

Momentum in the field of genetics has increased optimism among researchers seeking to treat and eventually cure this disorder. According to Dr. Michael E. Shy, Director of the CMT Clinic and Co-director of the Neuromuscular Program at Wayne State University, and Chair of the CMTA Medical Advisory Board, "We now are able to replicate this disorder in the laboratory and in doing so, can begin testing new treatments that will ultimately lead to clinical trials in people. In addition, the translational science used in the research could have major implications for the treatment of a host of related genetic disorders." These disorders include Multiple Sclerosis, Muscular Dystrophy and ALS (Lou Gehrig's disease). These current scientific and technological advances have resulted in the collaboration of researchers spearheading the STAR initiative.

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The most common form of CMT is due to an overproduction of the PMP22 protein in Schwann cells. This causes the myelin sheath that surrounds and insulates the nerve fiber to deteriorate. STAR will involve three primary projects:

\*In Project 1, researchers will be able to genetically engineer stable Schwann cell lines that express PMP22, couple them with a fluorescent “reporter” molecule, and test the efficacy of thousands of readily available medicines through a process known as “high-throughput screening.” Medicines that decrease the amount of PMP22 will become candidates for further studies.

\*Project 2 will focus on developing Schwann cell lines that more closely mimic human cells. This new line will improve test results from Project 1, and laboratory models will be used to test other candidate medicines as they are identified.

\*Project 3 seeks to further scientific understanding of how the human PMP22 gene is “turned on,” causing the overexpression of the PMP22 protein.

“Historically, CMT has been a mysterious disease that is not well understood or immediately recognizable by the health care community,” said Dr. Steven Scherer, Associate Professor of neurology at the University of Pennsylvania Medical Center and Co-chair of the CMTA Scientific Advisory Board. “Many of the patients we see tell us that, in hindsight, they had symptoms as a child but blamed it on their own clumsiness, when in fact, the problem was CMT.”

Named after the three physicians who discovered it in 1886, Jean-Martin Charcot, Pierre Marie and Howard Henry Tooth, CMT has not been the focus of much research. However, recent advances in genomic science led researchers from around the globe to begin to unravel the genetic mystery behind CMT. In 1991, scientists identified the specific gene associated with about 60 percent of all CMT cases (known as CMT1A). To date, 33 genes have been identified that play a role in CMT and that form the foundation for new research pathways.

“Now that genes involved in CMT have been identified, we are on the threshold of developing treatments for the millions of people suffering from the disorder,” said Patrick A. Livney, Chairman and President of the CMTA Board of Directors. “We also believe that if we can secure the requisite funding for the STAR initiative, a cure is within our reach.”

Researchers estimate that it will take \$10 million raised over the next 5 years to support the three projects of the STAR initiative. Tax deductible donations can be made to the Charcot-Marie-Tooth Association on their website at [www.charcot-marie-tooth.org](http://www.charcot-marie-tooth.org); by calling the CMTA at 1-800-606-CMTA (2682); or by mailing a check to the Charcot-Marie-Tooth Association, 2700 Chestnut Street, Chester, PA 19013.

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### **About CMT:**

CMT affects people of all ages, genders, races and ethnicities. It usually is not life-threatening and almost never affects intellectual functioning. The first signs may be frequent tripping or clumsiness, often accompanied by an abnormal burning sensation in the extremities. As the condition progresses, symptoms may include:

- Weakness in hands or ankles
- High arches
- Foot drop
- Hammertoes
- Muscle loss in the feet/legs, hands/arms
- Deformities
- Loss of motor nerve control

CMT is inherited in most cases. In dominant forms, one parent has CMT and their children have a 50/50 chance of developing the disorder. Less frequently, both parents carry a recessive CMT gene although they do not show signs of the disorder. In this instance, their children have a 25% chance of developing CMT. As with all genetic disorders, CMT can also appear as a new mutation in people with no prior hereditary history.

At present there is no cure for CMT. Common treatments include physical therapy, occupational therapy, bracing, and custom orthotics. Orthopedic surgical options are also available, but patients should carefully weigh the potential benefits and risks when making this decision.

### **About the Charcot-Marie-Tooth Association (CMTA):**

The CMTA, headquartered in Chester, Pennsylvania, is a 501(C)(3) nonprofit organization founded in 1983. The CMTA goals include providing patient support, public education, promotion of research, and ultimately the improved treatment and cure of CMT. The organization is led by an Executive Director and governed by a voluntary Board of Directors (BOD), and an international Medical Advisory Board (MAB) comprised of over fifty clinical and research professionals in specialties such as neurology, genetics, orthopedic surgery, physiatry, physical therapy and podiatry. The CMTA Scientific Advisory Board (SAB) is comprised of senior scientists with extensive research experience related to CMT. An External Advisory Board, made up of internationally recognized scientists, serves in an advisory capacity to the SAB. The CMTA has more than 15,000 patients and families, supportive friends, and medical

professionals in its member database. For more information, visit [www.charcot-marie-tooth.org](http://www.charcot-marie-tooth.org).

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Editor's note: To arrange an interview with Michael E. Shy, MD; Steven Scherer, MD, PhD; Patrick A. Livney, Chairman of the CMTA, or a CMT patient advocate, please contact Shannon Baker at 412.381.5400 or by e-mail at [sbaker@gmdadv.com](mailto:sbaker@gmdadv.com). Visit the Media Room at [www.charcot-marie-tooth.org](http://www.charcot-marie-tooth.org) for electronic documents, biosketches, medical images, photographs and other pertinent information.