Charcot-Marie-Tooth Research Foundation and Muscular Dystrophy Association Announce Joint Research Study Grant

NEW YORK, NY, USA, December 15, 2022 /EINPresswire.com/ -- The Muscular Dystrophy Association (MDA) and Charcot-Marie-Tooth Research Foundation (CMTRF) announce joint funding for a study by Alessandra Bolino, Ph.D., at San Raffaele Hospital (Ospedale San Raffaele), Italy, on restoring membrane trafficking in Charcot-Marie-Tooth (CMT) neuropathies with aberrant myelin. This will be a three-year study, with the grant totaling $263,450.

“We at the Muscular Dystrophy Association welcome this opportunity to join with CMTRF to support this important research,” says Sharon Hesterlee, Ph.D, Chief Research Officer, MDA. “Collaborating with like-minded organizations to fund research that may lead to increasing advancements in treatments and cures for Charcot-Marie-Tooth disease will increase our efficiency and help accelerate MDA’s overall mission to improve the lives of neuromuscular disease patients.”

CMT neuropathies are generally characterized by progressive muscular atrophy and weakness, with an age at onset usually comprised between the first and the second decade of life. Among CMT neuropathies, CMT4B1 is a very severe demyelinating neuropathy with childhood-onset, characterized by myelin outfoldings, redundant loops of myelin in the nerve that degenerate causing axonal problems. This form of aberrant myelin is also a pathological feature of other forms of demyelinating CMT, such as CMT4B2, B3, CMT4C and CMT4H. The laboratory previously demonstrated that loss of MTMR2 (Myotubularin-related 2) phosphatase is the cause of the disease. By investigating why loss of MTMR2 in Schwann cells provokes aberrant myelin, they identified a novel mechanism by which MTMR2 and its lipid substrate coordinate cytoskeleton dynamics and membrane growth within myelin-forming cells. In this project, researchers will further explore this mechanism of relevance in cell biology and test whether pharmacological and/or genetic modulation of these pathways can represent an effective strategy for the therapy of CMT4B with aberrant myelin.

“The CMT Research Foundation is pleased to partner with the Muscular Dystrophy Association on Dr. Bolino’s CMT4B1 project,” says Cleary Simpson CEO, CMTRF. “CMTRF is ready to form alliances with organizations that can get us closer to treatments or a cure for CMT.”

Muscular Dystrophy Association (MDA) is the #1 voluntary health organization in the United States for people living with muscular dystrophy, ALS, and related neuromuscular diseases. For

over 70 years, MDA has led the way in accelerating research, advancing care, and advocating for the support of our families. MDA's mission is to empower the people we serve to live longer, more independent lives.

The CMT Research Foundation (CMTRF) is focused solely on delivering treatments and cures for CMT. Founded by two patients who are driven to expedite drug delivery to people who live with CMT globally, the organization funds research for drug development. The 501(c)(3) federal tax-exempt organization is supported by personal and corporate financial gifts.

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