PRESS RELEASE

December 29, 2023

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SP-Foundation Awarding Over $1 Million For HSP and PLS Research in 2023

[O'Fallon, Missouri] – The Spastic Paraplegia Foundation (SP-Foundation) announces more than $1 Million has been awarded to medical researchers and investigators for 2023. The SP-Foundation completed its annual grant award process, choosing the highest-ranking medical research proposals studying HSP (Hereditary Spastic Paraplegia) and PLS (Primary Lateral Sclerosis). Following a peer review of the SPF Scientific Advisory Board, the SP-Foundation commits $1,114,438 to the following research projects for 2023:

SP-CoE / SP-CERN (Spastic Paraplegia Centers of Excellence Research Network), A collaborative research initiative into HSP and PLS, building a comprehensive program for diagnostic progress and clinical trial readiness to support the development of novel therapeutic approaches. This award will highlight the urgency of a consortium and international collaboration across the scientific community. This initiative will support the development of a registry and natural history study across the whole age span, a biobank, and a genome archive for a cohort of individuals with HSP or PLS identified and recruited, along with establishing a platform for molecular testing for those with no genetic cause(s) yet identified.

Dr. Peter Baas, Ph.D., Professor Department of Neurobiology & Anatomy, Drexel University College of Medicine. Antisense Oligonucleotide Therapy for SPG4-HSP New Treatment Strategy for SPG4-HSP. This grant application proposes to utilize contemporary antisense oligonucleotides (ASOs) generated by Ionis Pharmaceuticals to curtail the expression of mutant spastin proteins in an animal model for SPG4 Hereditary Spastic Paraplegia. Functional recovery will be augmented by exercise therapy in addition to the ASOs. Specific Goal 1. Test various incarnations of SPAST antisense oligonucleotide therapy on a mouse model for SPG4-HSP. Specific Goal 2. Ascertain whether exercise significantly promotes functional recovery in SPG4-HSP with advanced disease symptoms after antisense oligonucleotide therapy.

Stefan Barakat, M.D., Ph.D, Clinical Genetics, Erasmus University, Rotterdam, The Netherlands. "Exploring therapeutic avenues for a new type of HSP caused by mutations in AMFR: a pilot towards therapy." Finding a potential therapy for a rare new form of Hereditary Spastic Paraplegia. Aim-1: in-depth characterization of amfra-/- zebrafish, a new model system for a rare form of HSP, to explore disease mechanisms and potential therapy interventions, Aim-2: creating new human cell models to study HSP caused by AMFR mutations and test disease modifying compounds in human cells.

Laura Civiero, Ph.D. Associate Professor of Physiology, University of Padova, Italy. Finding novel approaches to rescue pathological phenotypes in ATP13A2-linked Hereditary Spastic Paraplegia. Finding novel therapeutic approaches in Hereditary Spastic Paraplegia. The main goal of this one-year trampoline project is to test a series of anti-inflammatory drugs with potential therapeutic effects on rare ATP13A2 loss of function disorders.

Matthias Kneussel, Ph.D., University Medical Center Hamburg-Eppendorf, Germany. Investigating Connections between Tubulin Posttranslational Modifications, Tubular ER Network Integrity and Hereditary Spastic Paraplegia-Related Proteins in Axons. How Hereditary Spastic Paraplegia Proteins May Regulate Microtubule-ER Crosstalk for Axonal Health. The proposed project aims to investigate the role of microtubules (MTs) and posttranslational tubulin modifications (PTMs) in the regulation of tubular endoplasmic reticulum (ER) network continuity in axons.

Claire Pujol, Ph.D, CNRS researcher, Pasteur Institute, Paris, France. Molecular dissection of mitochondrial dysfunction in HSP by unbiased imaging-based pharmacological and genetic screening. Mitochondrial Side Story of HSP disease. This proposal has two interconnected aims: - Drug discovery of mitochondrial-based modulators by a pharmacological screen to provide promising HSP therapeutic compounds. - Identification of new genetic modifiers in HSPs by a genetic screen to better understand the molecular mechanisms of HSP and to try to elucidate the mechanism(s) underpinning the great disparity of phenotypic variations observed in HSP. Together, the proposed project will uncover molecular mechanism that drive mitochondrial dysfunction in HSP and will identify repurposed drugs that will restore mitochondrial and cellular health in models of HSP.

Mukesh Gautam, Ph.D, Northwestern University. "Revealing ultrastructural defects in the motor cortex of PLS patients with and without TDP-43 pathology." Revealing cellular problems in the PLS brain. Specific Aim 1: To reveal the ultrastructural defects within the motor cortex of primary lateral sclerosis (PLS) patients with and without TDP-43 pathology. Specific Aim 2: To investigate organelle-specific defects in motor cortex of PLS patients with respect to TDP-43 pathology.

For more information contact Norma Pruitt at Information@SP-Foundation.org or to provide a tax-deductible donation to the Spastic Paraplegia Foundation visit SP-Foundation.org.

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