

From Bench to Bedside: Translational Approaches to Heat Shock Protein B8-Related Myopathy

Dr. Lan Weiss, scientist at the University of California, USA

Abstract

Heat Shock Protein B8 (HSPB8) plays a crucial role in chaperone-assisted selective autophagy (CASA), promoting the removal of misfolded proteins via autophagy. Autosomal dominant myopathy associated with HSPB8 is primarily caused by frameshift (fs) mutations, with affected patients often presenting distal myopathy in their thirties, progressing to generalized muscle weakness. Our research has shown that mutant HSPB8 increases protein aggregation and leads to a secondary loss of function in the wild-type HSPB8 protein. The goal of our work is to establish reliable disease models to study the underlying mechanisms and explore translational treatment strategies. In this talk, you will hear about our investigation of the Adeno-Associated Virus (AAV) dual vector strategy as a potential gene therapy approach for HSPB8-associated myopathy.



About the speaker

Dr. Lan Weiss is a scientist at the University of California, Irvine, USA, with a background in Medicine from Hue University, Vietnam, and a Ph.D. in Human Genetics from Nagasaki University, Japan. Previously, she worked in the Microbiology and Immunology department at the Pasteur Institute, Ho Chi Minh City, Vietnam, focusing on infectious diseases and outbreak response. Since relocating to the USA in 2016, her research has centered on hereditary diseases such as Myopathy associated with mutations in the Heat Shock Protein B8 (HSPB8), VCP multisystem proteinopathy, and Pompe disease. She is particularly interested in using Adeno-Associated Viruses (AAV) and antisense oligonucleotides for gene therapy, leveraging iPSC-derived muscle cells and mouse disease models to advance treatments for these debilitating conditions.