

Motor Control Science Club, November 4, 2022, 3:00 PM CET (online) The lecture is open to everybody

## Repeat Associated Non-ATG (RAN) in neurological and neuromuscular disease

Prof. Laura Ranum Department of Molecular Genetics and Microbiology, University of Florida, USA

Host: Prof. Brunhilde Wirth Institute of Human Genetics, University of Cologne



The goal of the Ranum laboratory is to perform cutting edge research that will lead to improvements in diagnosis and treatments for neurological and neuromuscular disease. Current research in the Ranum lab focuses on the role of Repeat Associated Non-ATG (RAN) translation, RNA gain of function and protein gain of function in repeat expansion disorders including amyotrophic lateral sclerosis (ALS), spinocerebellar ataxia (SCA) types 5 and 8, myotonic dystrophy (DM) types 1 and 2 and Huntington's disease (HD). She is investigating the mechanism by which RAN translation occurs in these diseases and the toxic effects of RAN proteins on the brain and other organs. She has shown that RAN proteins accumulate in brain tissue from patients diagnosed with SCA8, ALS and Huntington's disease and is using mouse models of these diseases to better understand the impact of these proteins and to develop therapeutic strategies. Additionally, the Ranum laboratory continues to search for novel human disease genes. She is using high-throughput sequencing strategies to look for single-gene mutations that cause novel forms of ataxia, ALS and neuropsychiatric diseases. Because RAN translation has now been shown to occur across multiple diseases, and greater than 30 diseases are caused by repeat expansion mutations, these studies are likely to contribute to the understanding and development of urgently needed therapies for a large category of diseases.



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