

Proteogenomics of Spinal Muscular Atrophy

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Abstract

Spinal Muscular Atrophy (SMA), a disease caused by the mutations of Survival Motor Neuron 1 (SMN1) gene, is the most common genetic cause of infant mortality. Our preliminary results show that SMN is found in a complex containing an E3 ubiquitin ligase the Anaphase- Promoting Complex (APC) and HuD, a RNA binding protein. The APC targets proteins to the proteasome for degradation whereas HuD stabilizes mRNAs. Furthermore, evidence indicates a role for APC in neuronal survival, axonal growth, and synaptic function, and HuD is involved in the maturation and maintenance of neurons. These data together prompted us to formulate a working model in which: 1) ubiquitination by APC regulates the stability and/or function of SMN or other members of the SMN complex and 2) the putative HuD-SMN-associated mRNAs in axons are important for the growth and survival of motor neurons.

About the Speaker

Dr. Judith Steen is an Associate Professor of Neurobiology at Harvard Medical School, a member of the Harvard Stem Cell Institute and the Director of the Neuroproteomics Laboratory in the F. M. Kirby Neuroscience Center at Boston Children's Hospital. Her laboratory works to understand neuro-regeneration and neurodegenerative diseases using systems biology approaches. The laboratory develops novel qualitative and quantitative methodologies at the interface of proteomics and transcriptomics with special emphasis on computational proteomics approaches.