# Deconstructing SMA: from RNA Processing to Motor Neuron Disease

## June 6

### Tuesday, 12:30 pm

**Billings Building—Rosedale Room and Zoom**

***SPEAKER:***


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**Host: Rajiv R. Ratan, M.D., Ph.D.**

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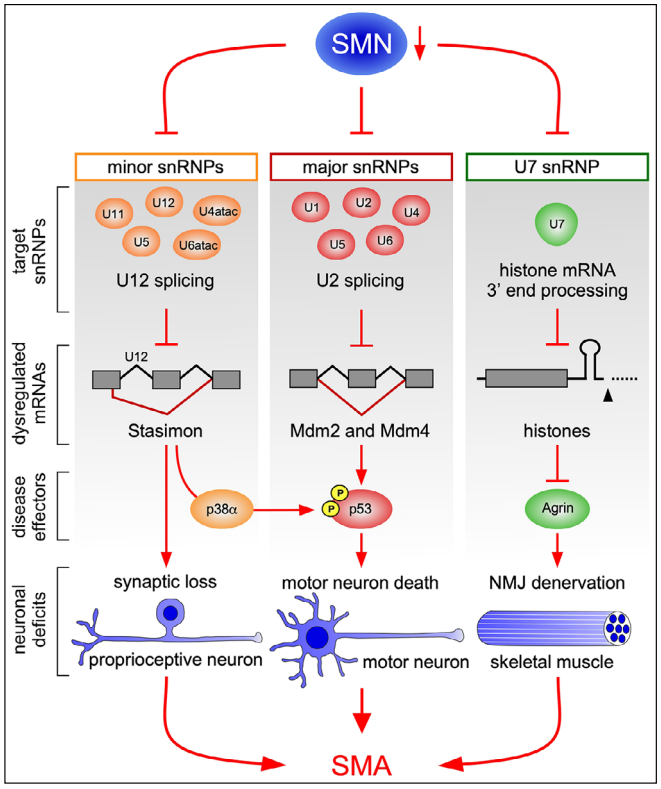
## Abstract


Research in the Pellizzoni laboratory investigates the mechanisms by which RNA binding proteins (RBPs) and molecular chaperones that mediate their assembly into RNA-protein complexes (RNPs) regulate gene expression. The laboratory also focuses on the question of how general perturbations of RNA metabolism cause synaptic dysfunction and neuronal death in neurodegenerative diseases such as spinal muscular atrophy (SMA). In these studies, we employ cellular and animal models as well as a wide range of genomic, biochemical, molecular, and imaging approaches. High-throughput screens are also used to discover chemical and genetic modifiers of disease pathways. On one hand, these efforts are designed to advance our knowledge of how RNA regulation contributes to neural circuit function. On the other, they aim to deconstruct disease mechanisms and identify potential therapeutic targets.


### *Publications*


1. M. Van Alstyne, C.M. Simon, S.P. Sardi, L.S. Shihabuddin, G.Z. Mentis and L. Pellizzoni*. (2018). Dysregulation of Mdm2 and Mdm4 alternative splicing underlies motor neuron death in spinal muscular atrophy. Genes Dev. 32, 1045-1059. https://pubmed.ncbi.nlm.nih.gov/30012555/

2. M. Van Alstyne, I. Tattoli, N. Delestree, Y. Recinos, E. Workman, L.S. Shihabuddin, C. Zhang, G.Z. Mentis and L. Pellizzoni*. (2021). Gain of toxic function by long-term AAV9-mediated SMN overexpression in the sensorimotor circuit. Nat. Neurosci. 24, 930-940. https://pubmed.ncbi.nlm.nih.gov/33795885/

3. S. Tisdale, M. Van Alstyne, C.M. Simon, G.Z. Mentis and L. Pellizzoni*. (2022). SMN controls neuromuscular junction integrity through U7 snRNP. Cell Rep. 40, 111393. https://pubmed.ncbi.nlm.nih.gov/36130491/