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Keywords

Alternative splicing - mRNA transport - motor neurons - axon development and survival

Science

Spinal muscular atrophy (SMA) is a neurodegenerative disorder characterized by the selective degeneration of α-motoneurons in the spinal cord. SMA is caused by the loss of the Survival of Motor Neurons gene (SMN1) that encodes a house-keeping protein required by all cell types. Humans have a second copy of the gene, SMN2. Both genes encode identical proteins, but an alternative splicing event predominantly skips exon 7 of SMN2. SMA cells thus produce less functional SMN protein. Why reduction in the levels of a house-keeping factor selectively damages α-motoneurons remains completely enigmatic. At physiological level, the first impairments occur during axonal development level which fails to establish mature connections with the muscular endplate, causing the eventual death of the motor neuron.

My project aims to investigate the alternative splicing of SMN2 gene and the causes of impaired axonal connection on the muscle giving particular attention to mRNA transport and local translation events.