The Relationship Between Elongator Protein 3 (ELP3) and Neurodegenerative Disease: Investigating the Neuromuscular Junction in an Elongator Mouse Model of ALS

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Amyotrophic Lateral Sclerosis (ALS), also called Lou Gehrig's Disease, is an extremely damaging neurodegenerative disorder that attacks the motor system, which gradually weakens and wastes muscle. In both sporadic and hereditary forms of ALS cases, the large degree of phenotypic heterogeneity suggests that multiple genetic modifiers contribute to the progression rate, duration, and site of onset for the disease. A growing body of evidence identifies catalytic subunit 3 of the elongator complex protein (ELP3) as a possible ALS modifier. A verified hallmark of the disease is degeneration of neuromuscular junctions. Neuromuscular junctions are part of a complex system of synapses, which are filled or innervated by motor neurons. Absence of ELP3 was predicted to encourage similar effects to that of ALS, including degeneration of the neuromuscular junctions. Through the process of immunofluorescence, I examined the neuromuscular junctions of tibialis anterior muscle of both a control and an ELP3 knockout mouse sample. Muscles and nerves within the tissue produced a fluorescent hue showing whether or not there was full functionality or full innervation within nerves. Results of this research indicate that the control sample has no disruptions to the function of the neuromuscular junction system. However, in the knockout ELP3 model more than half of the junctions were affected heavily. The similarity of the effects of the absence of ELP3 and ALS, establishes ELP3 as a modifier in the disease. Establishing ELP3 as a modifier allows a more comprehensive understanding of how sensitivity of tRNA is affected in ALS.

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