Peripheral Nervous System Alterations in Disease and Injury

Eric Villalon

Dr. Michael L. Garcia, Dissertation Supervisor

Abstract

Charcot-Marie-Tooth (CMT) is the most common inherited neuropathy of the PNS affecting approximately 2.8 million people. CMT type 2E, an axonal form of CMT, has been linked to mutations in the neurofilament light gene (*nefl*) and leads to distal neuropathy characterized by reduced nerve conduction velocity, muscle atrophy and sensory loss. However, the mechanisms of disease pathogenesis are not well understood. My work characterizes a CMT2E mouse model and reveals new insights into disease pathogenesis. Moreover, patients suffering with CMT are susceptible to develop exacerbation of neuropathy following a traumatic event. My work also provides insights into exacerbation of neuropathy in our CMT mouse model. Lastly, following remyelination of peripheral nerves after injury, internode lengths are significantly reduced. My work provides the first molecular basis for the reduction in internode length after remyelination and identifies a potential target for therapeutic intervention aimed at enhancing recovery from demyelinating diseases or nerve injuries.