

It's Not a Wrap! 4 Key Proteins Call Their Agents

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Charcot-Marie-Tooth disease (CMT) is the most commonly inherited peripheral neuropathy disorder in humans with an incidence of about 1 in 2500. This disease involves slowly progressive atrophy and muscle weakness in the peripheral appendages. CMT affects many different types of people with varying age-of-onset and progression. There are a few different classes of CMT: CMT1, which entails demyelination and slow nerve conduction velocities (NCV's), and CMT2, which involves axonal atrophy and decreased NCV's and amplitudes. This review will focus on CMT1, which includes mutations in four different genes: PMP22, MPZ, Cx32, and EGR2. PMP22 and MPZ encode the myelin structure proteins, Cx32 encodes a gap junction protein involved in movement of molecules across the myelin sheath, and EGR2 encodes a transcription factor needed for myelin development. Current research has shown that mutations within these genes contribute significantly to symptoms seen in CMT1 patients. Understanding the mechanism behind these mutations may provide researchers with the knowledge to better comprehend the molecular basis of CMT1 in hopes of leading to an eventual cure to this debilitating disease.

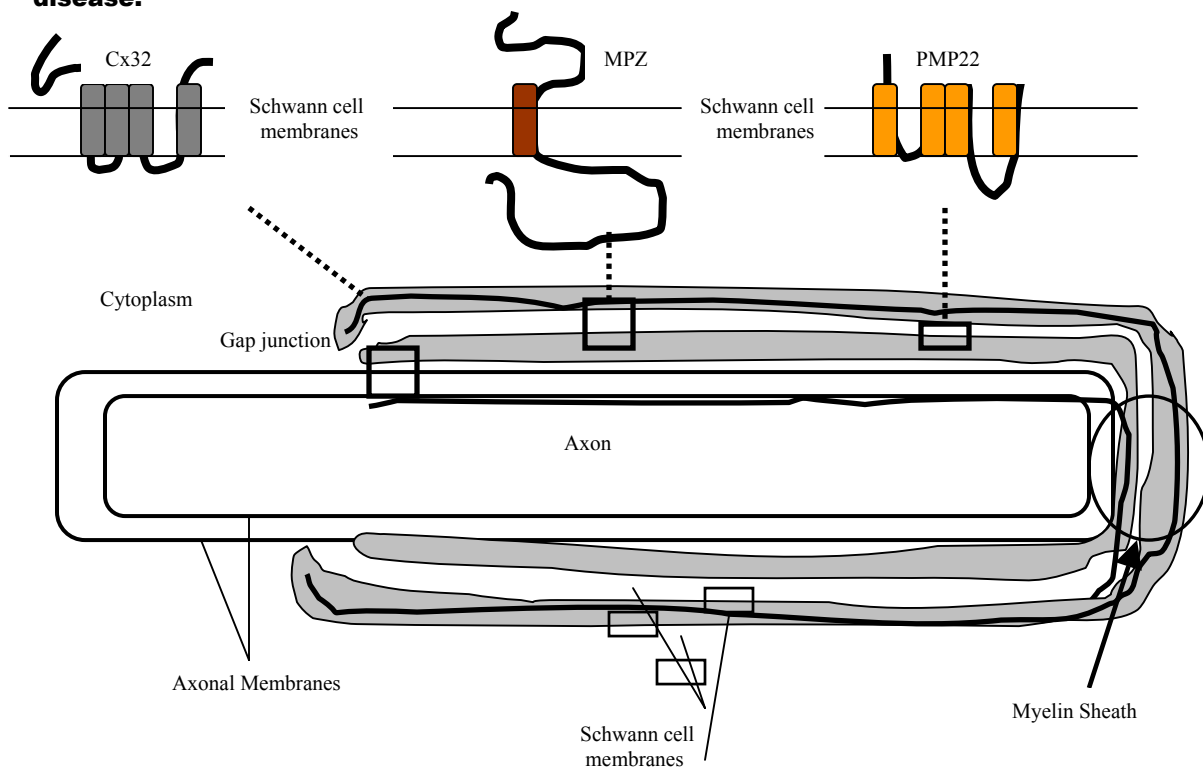


Figure 2: Biological Model of 3 Key Proteins Associated with CMT1 located on a Neuron. Three peripheral proteins involved in CMT1 are connexin 32 (Cx32), which is the X-linked form, myelin protein zero (MPZ), involved in CMT1B, and peripheral myelin protein 22 (PMP22), involved in CMT1A. Cx32 is a gap junction protein that sits in between the schwann cell membranes and allows for passage of small molecules. MPZ is located on the myelin sheath and it serves as a glue for the sheath. PMP22 is also located in the myelin sheath and aids in the structural formation of it. In conclusion, a myelin sheath is constructed of schwann cell membranes that wrap around the axon of a neuron and serve to protect and aid in the transfer of signals throughout the peripheral nervous system.