

Researchers untangle molecular pathology of giant axonal neuropathy

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Giant axonal neuropathy (GAN) is a rare genetic disorder that causes central and peripheral nervous system dysfunction. GAN is known to be caused by mutations in the gigaxonin gene and is characterized by tangling and aggregation of neural projections, but the mechanistic link between the genetic mutation and the effects on neurons is unclear.

In this issue of the *Journal of Clinical Investigation*, Robert Goldman and colleagues at Northwestern University uncover how mutations in gigaxonin contribute to neural aggregation. They demonstrated that gigaxonin regulates the degradation of neurofilament proteins, which help to guide outgrowth and morphology of neural projections.

Loss of gigaxonin in either GAN patient cells or [transgenic mice](#) increased levels of neurofilament proteins, causing tangling and aggregation of neural projections. Importantly, expression of gigaxonin allowed for clearance of neurofilament proteins in neurons.

These findings demonstrate that mutations in gigaxonin cause accumulation of neurofilament proteins and shed light on the molecular pathology of GAN.

More information: Giant axonal neuropathy–associated gigaxonin mutations impair intermediate filament protein degradation, *J Clin Invest.* [doi:10.1172/JCI66387](https://doi.org/10.1172/JCI66387)

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