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Unraveling the genetic basis of peripheral neurodegeneration – is there a hint?

Inherited peripheral neuropathies are frequent neuromuscular disorders knownfor their clinical and genetic heterogeneity. In 33 families we identified eightmutations in histidine triad nucleotide binding protein 1 (HINT1) by combining linkage analyses with next generation sequencing and subsequent patient cohort screening. Our study indicates that HINT1 mutations are a frequent cause of autosomal recessive Charcot-Marie-Tooth disease and provides evidence that loss of functional protein results in a distinct phenotype of axonal neuropathy with neuromyotonia.