KIF1A

Mutations in the kinesin family member 1A (KIF1A) gene have been associated with a wide range of phenotypes including recessive mutations causing hereditary sensory neuropathy and hereditary spastic paraplegia and de novo dominant mutations causing a more complex neurological disorder affecting both the central and peripheral nervous system. We identified by exome sequencing a de novo dominant missense variant, (c.38G>A, p.R13H), within an ATP binding site of the kinesin motor domain in a patient manifesting a complex phenotype characterized by autism spectrum disorder, spastic paraplegia and axonal neuropathy. The presence of autism spectrum disorder distinguishes this case from previously reported patients with de novo dominant mutations in KIF1A [1].

1)
Tomaselli PJ, Rossor AM, Horga A, Laura M, Blake JC, Houlden H, Reilly MM. A de novo dominant mutation in KIF1A associated with axonal neuropathy, spasticity and autism spectrum disorder. J Peripher Nerv Syst. 2017 Aug 23. doi: 10.1111/jns.12235. [Epub ahead of print] PubMed PMID: 28834584.