De novo mutations in KIF1A cause progressive encephalopathy and brain atrophy

DOI: 10.1002/acn3.198
Annals of Clinical and Translational Neurology, 2015, 2, 623-3

#	Paper	IF	Citations
86	Variants in KIF1A gene in dominant and sporadic forms of hereditary spastic paraparesis. *Journal of Neurology*, **2015**, 262, 2684-90	5.5	42
85	Sending Mixed Signals: The Expanding Role of Molecular Cascade Mutations in Malformations of Cortical Development and Epilepsy. *Epilepsy Currents*, **2016**, 16, 158-63	1.3	5
84	The Drosophila KIF1A Homolog unc-104 Is Important for Site-Specific Synapse Maturation. *Frontiers in Cellular Neuroscience*, **2016**, 10, 207	6.1	11
83	Novel De Novo Mutations in KIF1A as a Cause of Hereditary Spastic Paraplegia With Progressive Central Nervous System Involvement. *Journal of Child Neurology*, **2016**, 31, 1114-9	2.5	31
82	Emerging roles for motor proteins in progenitor cell behavior and neuronal migration during brain development. *Cytoskeleton*, **2016**, 73, 566-576	2.4	15
81	Neurometabolic disorders: Five new things. *Neurology: Clinical Practice*, **2016**, 6, 348-357	1.7	7
80	Utility of Whole Exome Sequencing for Genetic Diagnosis of Previously Undiagnosed Pediatric Neurology Patients. *Journal of Child Neurology*, **2016**, 31, 1534-1539	2.5	38
79	The importance of de novo mutations for pediatric neurological disease--It is not all in utero or birth trauma. *Mutation Research - Reviews in Mutation Research*, **2016**, 767, 42-58	7	7
78	CCDC88A mutations cause PEHO-like syndrome in humans and mouse. *Brain*, **2016**, 139, 1036-44	11.2	17
77	Monoallelic and Biallelic Variants in EMC1 Identified in Individuals with Global Developmental Delay, Hypotonia, Scoliosis, and Cerebellar Atrophy. *American Journal of Human Genetics*, **2016**, 98, 562-570	11	45
76	KIF1A inhibition immortalizes brain stem cells but blocks BDNF-mediated neuronal migration. *Nature Neuroscience*, **2016**, 19, 253-62	25.5	28
75	Neuroimaging in Aicardi-Goutières syndrome: Biomarkers for a progressive encephalopathy. *Neurology*, **2016**, 86, 15-6	6.5	2
74	De novo dominant variants affecting the motor domain of KIF1A are a cause of PEHO syndrome. *European Journal of Human Genetics*, **2016**, 24, 949-53	5.3	30
73	Genetic control of postnatal human brain growth. *Current Opinion in Neurology*, **2017**, 30, 114-124	7.1	59
72	Optic nerve hypoplasia in a patient with a de novo KIF1A heterozygous mutation. *Canadian Journal of Ophthalmology*, **2017**, 52, e169-e171	1.4	10
71	Hereditary spastic paraplegia caused by compound heterozygous mutations outside the motor domain of the KIF1A gene. *European Journal of Neurology*, **2017**, 24, 741-747	6	17
70	Autosomal dominant transmission of complicated hereditary spastic paraplegia due to a dominant negative mutation of KIF1A, SPG30 gene. *Scientific Reports*, **2017**, 7, 12527	4.9	28

69	A de novo dominant mutation in KIF1A associated with axonal neuropathy, spasticity and autism spectrum disorder. *Journal of the Peripheral Nervous System*, **2017**, 22, 460-463	4.7	18
68	The KIF1A homolog Unc-104 is important for spontaneous release, postsynaptic density maturation and perisynaptic scaffold organization. *Scientific Reports*, **2017**, 7, 38172	4.9	9
67	Co-existence of spastic paraplegia-30 with novel KIF1A mutation and spinocerebellar ataxia 31 with intronic expansion of BEAN and TK2 in a family. *Journal of the Neurological Sciences*, **2017**, 372, 128-130	3.2	6
66	Potential Role of Microtubule Stabilizing Agents in Neurodevelopmental Disorders. *International Journal of Molecular Sciences*, **2017**, 18,	6.3	18
65	Defective kinesin binding of TUBB2A causes progressive spastic ataxia syndrome resembling sacsinopathy. *Human Molecular Genetics*, **2018**, 27, 1892-1904	5.6	18
64	Nuclear migration in mammalian brain development. *Seminars in Cell and Developmental Biology*, **2018**, 82, 57-66	7.5	23
63	Progressive cerebello-cerebral atrophy and progressive encephalopathy with edema, hypsarrhythmia and optic atrophy may be allelic syndromes. *European Journal of Paediatric Neurology*, **2018**, 22, 1133-1138	3.8	12
62	De novo variant in KIF26B is associated with pontocerebellar hypoplasia with infantile spinal muscular atrophy. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2623-2629	2.5	13
61	Genetic heterogeneity in infantile spasms. *Epilepsy Research*, **2019**, 156, 106181	3	20
60	Axonal transport: Driving synaptic function. *Science*, **2019**, 366,	33.3	92
59	Rett and Rett-like syndrome: Expanding the genetic spectrum to KIF1A and GRIN1 gene. *Molecular Genetics & Genomic Medicine*, **2019**, 7, e968	2.3	18
58	Disease-associated mutations hyperactivate KIF1A motility and anterograde axonal transport of synaptic vesicle precursors. *Proceedings of the National Academy of Sciences of the United States of America*, **2019**, 116, 18429-18434	11.5	32
57	Going Too Far Is the Same as Falling Short: Kinesin-3 Family Members in Hereditary Spastic Paraplegia. *Frontiers in Cellular Neuroscience*, **2019**, 13, 419	6.1	15
56	Defects in Axonal Transport in Inherited Neuropathies. *Journal of Neuromuscular Diseases*, **2019**, 6, 401-419	5	14
55	Next-generation sequencing study reveals the broader variant spectrum of hereditary spastic paraplegia and related phenotypes. *Neurogenetics*, **2019**, 20, 27-38	3	22
54	Targeted resequencing identifies genes with recurrent variation in cerebral palsy. *Npj Genomic Medicine*, **2019**, 4, 27	6.2	15
53	PEHO syndrome: KIF1A mutation and decreased activity of mitochondrial respiratory chain complex. *Journal of Clinical Neuroscience*, **2019**, 61, 298-301	2.2	13
52	KIF1A variants are a frequent cause of autosomal dominant hereditary spastic paraplegia. *European Journal of Human Genetics*, **2020**, 28, 40-49	5.3	30

51	Overlapping spectrums: The clinicogenetic commonalities between Charcot-Marie-Tooth and other neurodegenerative diseases. *Brain Research*, **2020**, 1727, 146532	3.7	1
50	Mobility Characteristics of Children with Spastic Paraplegia Due to a Mutation in the KIF1A Gene. *Neuropediatrics*, **2020**, 51, 146-153	1.6	6
49	A Novel de novo KIF1A Mutation in a Patient with Autism, Hyperactivity, Epilepsy, Sensory Disturbance, and Spastic Paraplegia. *Internal Medicine*, **2020**, 59, 839-842	1.1	8
48	Postnatal Role of the Cytoskeleton in Adult Epileptogenesis. *Cerebral Cortex Communications*, **2020**, 1, tgaa024	1.9	2
47	Tubulin mutations in neurodevelopmental disorders as a tool to decipher microtubule function. *FEBS Letters*, **2020**, 594, 3409-3438	3.8	7
46	Clinical and molecular characterization of hereditary spastic paraplegia in a spanish southern region. *International Journal of Neuroscience*, **2020**, 1-12	2	2
45	Anterograde Axonal Transport in Neuronal Homeostasis and Disease. *Frontiers in Molecular Neuroscience*, **2020**, 13, 556175	6.1	16
44	Usefulness of exome sequencing in the study of spastic paraparesis and cerebellar atrophy: de novo mutation of the KIF1A gene, a new hope in prognosis. *Neurología (English Edition)*, **2020**, 35, 535-538	0.4	
43	KIF1A-related autosomal dominant spastic paraplegias (SPG30) in Russian families. *BMC Neurology*, **2020**, 20, 290	3.1	3
42	Heterozygous variants underlie a wide spectrum of neurodevelopmental and neurodegenerative disorders. *Journal of Medical Genetics*, **2021**, 58, 475-483	5.8	5
41	Phenotypic expansion in KIF1A-related dominant disorders: A description of novel variants and review of published cases. *Human Mutation*, **2020**, 41, 2094-2104	4.7	1
40	De novo variants in cause neurodevelopmental disorders associated with corpus callosum abnormalities. *Journal of Medical Genetics*, **2020**, 57, 461-465	5.8	4
39	Synaptic Vesicle Precursors and Lysosomes Are Transported by Different Mechanisms in the Axon of Mammalian Neurons. *Cell Reports*, **2020**, 31, 107775	10.6	18
38	A Rare Missense Mutation Enhances Synaptic Function and Increases Seizure Activity. *Frontiers in Genetics*, **2020**, 11, 61	4.5	14
37	Advances in the evaluation and management of cortical/cerebral visual impairment in children. *Survey of Ophthalmology*, **2020**, 65, 708-724	6.1	20
36	Nucleokinesis. **2020**, 305-322		0
35	Congenital and postnatal microcephalies. **2020**, 377-408		
34	Usefulness of exome sequencing in the study of spastic paraparesis and cerebellar atrophy: De novo mutation of the KIF1A gene, a new hope in prognosis. *Neurología*, **2020**, 35, 535-538	1.4	3

33	Expansion of the phenotypic spectrum of de novo missense variants in kinesin family member 1A (KIF1A). *Human Mutation*, **2020**, 41, 1761-1774	4.7	10
32	KIF1A-related disorders in children: A wide spectrum of central and peripheral nervous system involvement. *Journal of the Peripheral Nervous System*, **2020**, 25, 117-124	4.7	17
31	The neurodevelopmental spectrum of synaptic vesicle cycling disorders. *Journal of Neurochemistry*, **2021**, 157, 208-228	6	14
30	Dominant and sporadic de novo disorders. **2021**, 117-135		
29	Pathogenic variants in CDH11 impair cell adhesion and cause Teebi hypertelorism syndrome. *Human Genetics*, **2021**, 140, 1061-1076	6.3	1
28	A highly conserved 3 helix within the kinesin motor domain is critical for kinesin function and human health. *Science Advances*, **2021**, 7,	14.3	4
27	Genotype and defects in microtubule-based motility correlate with clinical severity in -associated neurological disorder. *Human Genetics and Genomics Advances*, **2021**, 2,	0.8	9
26	The Novel KIF1A Missense Variant (R169T) Strongly Reduces Microtubule Stimulated ATPase Activity and Is Associated With NESCAV Syndrome. *Frontiers in Neuroscience*, **2021**, 15, 618098	5.1	4
25	Neuronal Cytoskeleton in Intellectual Disability: From Systems Biology and Modeling to Therapeutic Opportunities. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	2
24	De novo disease-associated mutations in KIF1A dominant negatively inhibit axonal transport of synaptic vesicle precursors.		1
23	Regulation Of Microtubule: Current Concepts And Relevance To Neurodegenerative Diseases. *CNS and Neurological Disorders - Drug Targets*, **2021**,	2.6	
22	Monoallelic KIF1A-related disorders: a multicenter cross sectional study and systematic literature review. *Journal of Neurology*, **2021**, 1	5.5	3
21	Genotype and defects in microtubule-based motility correlate with clinical severity in KIF1A Associated Neurological Disorder.		1
20	ER Morphology in the Pathogenesis of Hereditary Spastic Paraplegia. *Cells*, **2021**, 10,	7.9	1
19	Here comes the sun: the era of genetics. *Arquivos De Neuro-Psiquiatria*, **2015**, 73, 895-6	1.6	
18	Disease-associated mutations hyperactivate KIF1A motility and anterograde axonal transport of synaptic vesicle precursors.		
17	A Highly Conserved 310-Helix Within the Kinesin Motor Domain is Critical for Kinesin Function and Human Health.		1
16	Hemorrhagic shock and encephalopathy syndrome in a patient with a de novo heterozygous variant in KIF1A.. *Brain and Development*, **2021**,	2.2	0

15	A neuropathy-associated kinesin KIF1A mutation hyper-stabilizes the motor-neck interaction during the ATPase cycle.. *EMBO Journal*, **2022**, e108899	13	2
14	Image_1.tif. **2020**,		
13	Table_1.docx. **2020**,		
12	Table_1.docx. **2019**,		
11	De novo mutations in KIF1A-associated neuronal disorder (KAND) dominant-negatively inhibit motor activity and axonal transport of synaptic vesicle precursors. **2022**, 119,		1
10	The net charge of the K-loop regulates KIF1A superprocessivity by enhancing microtubule affinity in the one-head-bound state.		
9	Kinesin-3 motors are fine-tuned at the molecular level to endow distinct mechanical outputs. **2022**, 20,		0
8	Single-Molecule Studies on the Motion and Force Generation of the Kinesin-3 Motor KIF1A. **2022**, 585-608		0
7	Dodecyl Creatine Ester Improves Cognitive Function and Identifies Drivers of Creatine Deficiency.		0
6	Positive charge in the K-loop of the kinesin-3 motor KIF1A regulates superprocessivity by enhancing microtubule affinity in the one-head-bound state. **2022**, 102818		0
5	KIF1A-Associated Neurological Disorder: An Overview of a Rare Mutational Disease. **2023**, 16, 147		0
4	Insight into the regulation of axonal transport from the study of KIF1A-associated neurological disorder. **2023**, 136,		0
3	High-Resolution structures of microtubule-bound KIF1A and its pathogenic variant P305L.		0
2	Dodecyl creatine ester improves cognitive function and identifies key protein drivers including KIF1A and PLCB1 in a mouse model of creatine transporter deficiency. 16,		0
1	Expanding the Knowledge of KIF1A-Dependent Disorders to a Group of Polish Patients. **2023**, 14, 972		0