Stephan Zuchner

List of Publications by Year
in descending order

Version: 2024-02-01

147 papers	13,039 citations	24978 57 h-index	25716 108 g-index
155 all docs	155 docs citations	155 times ranked	17918 citing authors

#	Article	IF	Citations
1	Mutations in the mitochondrial GTPase mitofusin 2 cause Charcot-Marie-Tooth neuropathy type 2A. Nature Genetics, 2004, 36, 449-451.	9.4	1,391
2	The Human Phenotype Ontology in 2017. Nucleic Acids Research, 2017, 45, D865-D876.	6.5	699
3	dSarm/Sarm1 Is Required for Activation of an Injury-Induced Axon Death Pathway. Science, 2012, 337, 481-484.	6.0	558
4	TIA1 Mutations in Amyotrophic Lateral Sclerosis and Frontotemporal Dementia Promote Phase Separation and Alter Stress Granule Dynamics. Neuron, 2017, 95, 808-816.e9.	3.8	493
5	The Matchmaker Exchange: A Platform for Rare Disease Gene Discovery. Human Mutation, 2015, 36, 915-921.	1.1	390
6	Axonal neuropathy with optic atrophy is caused by mutations in mitofusin 2. Annals of Neurology, 2006, 59, 276-281.	2.8	380
7	MFN2 mutation distribution and genotype/phenotype correlation in Charcot-Marie-Tooth type 2. Brain, 2006, 129, 2093-2102.	3.7	351
8	Biallelic expansion of an intronic repeat in RFC1 is a common cause of late-onset ataxia. Nature Genetics, 2019, 51, 649-658.	9.4	338
9	Mutations in the pleckstrin homology domain of dynamin 2 cause dominant intermediate Charcot-Marie-Tooth disease. Nature Genetics, 2005, 37, 289-294.	9.4	324
10	Genome-wide Studies of Copy Number Variation and Exome Sequencing Identify Rare Variants in BAG3 as a Cause of Dilated Cardiomyopathy. American Journal of Human Genetics, 2011, 88, 273-282.	2.6	320
11	Hereditary spastic paraplegia: Clinicogenetic lessons from 608 patients. Annals of Neurology, 2016, 79, 646-658.	2.8	218
12	Mutations in the Novel Mitochondrial Protein REEP1 Cause Hereditary Spastic Paraplegia Type 31. American Journal of Human Genetics, 2006, 79, 365-369.	2.6	209
13	Mutations in ANKRD11 Cause KBG Syndrome, Characterized by Intellectual Disability, Skeletal Malformations, and Macrodontia. American Journal of Human Genetics, 2011, 89, 289-294.	2.6	205
14	Genetics of Charcot-Marie-Tooth (CMT) Disease within the Frame of the Human Genome Project Success. Genes, 2014, 5, 13-32.	1.0	203
15	PNPLA6 mutations cause Boucher-NeuhÃuser and Gordon Holmes syndromes as part of a broad neurodegenerative spectrum. Brain, 2014, 137, 69-77.	3.7	189
16	Mutations in a Gene Encoding a Novel SH3/TPR Domain Protein Cause Autosomal Recessive Charcot-Marie-Tooth Type 4C Neuropathy. American Journal of Human Genetics, 2003, 73, 1106-1119.	2.6	185
17	Alteration of Fatty-Acid-Metabolizing Enzymes Affects Mitochondrial Form and Function in Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2012, 91, 1051-1064.	2.6	179
18	Multiple rare SAPAP3 missense variants in trichotillomania and OCD. Molecular Psychiatry, 2009, 14, 6-9.	4.1	166

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19	Mutations in SLC25A46, encoding a UGO1-like protein, cause an optic atrophy spectrum disorder. Nature Genetics, 2015, 47, 926-932.	9.4	166
20	REEP1 mutation spectrum and genotype/phenotype correlation in hereditary spastic paraplegia type 31. Brain, 2008, 131, 1078-1086.	3.7	163
21	Mutations in BICD2 Cause Dominant Congenital Spinal Muscular Atrophy and Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2013, 92, 965-973.	2.6	156
22	Loss of Function of Glucocerebrosidase GBA2 Is Responsible for Motor Neuron Defects in Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2013, 92, 238-244.	2.6	154
23	Alteration of Ganglioside Biosynthesis Responsible for Complex Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2013, 93, 118-123.	2.6	151
24	Mutations in the ER-shaping protein reticulon 2 cause the axon-degenerative disorder hereditary spastic paraplegia type 12. Journal of Clinical Investigation, 2012, 122, 538-544.	3.9	149
25	Whole-Exome Sequencing Efficiently Detects Rare Mutations in Autosomal Recessive Nonsyndromic Hearing Loss. PLoS ONE, 2012, 7, e50628.	1.1	143
26	Loss-of-function mutations in theATP13A2/PARK9 gene cause complicated hereditary spastic paraplegia (SPG78). Brain, 2017, 140, 287-305.	3.7	135
27	A novel mutation in VCP causes Charcotâ€“Marieâ€“Tooth Type 2 disease. Brain, 2014, 137, 2897-2902.	3.7	116
28	Evaluating Pathogenicity of Rare Variants From Dilated Cardiomyopathy in the Exome Era. Circulation: Cardiovascular Genetics, 2012, 5, 167-174.	5.1	112
29	Exome sequencing identifies a significant variant in methionyl-tRNA synthetase (MARS) in a family with late-onset CMT2. Journal of Neurology, Neurosurgery and Psychiatry, 2013, 84, 1247-1249.	0.9	112
30	Exome sequencing allows for rapid gene identification in a Charcotâ€Marieâ€Tooth family. Annals of Neurology, 2011, 69, 464-470.	2.8	107
31	Loss-of-function mutations in HINT1 cause axonal neuropathy with neuromyotonia. Nature Genetics, 2012, 44, 1080-1083.	9.4	102
32	Absence of BiP Co-chaperone DNAJC3 Causes Diabetes Mellitus and Multisystemic Neurodegeneration. American Journal of Human Genetics, 2014, 95, 689-697.	2.6	100
33	Biallelic mutations in SORD cause a common and potentially treatable hereditary neuropathy with implications for diabetes. Nature Genetics, 2020, 52, 473-481.	9.4	97
34	Synaptotagmin 2 Mutations Cause an Autosomal-Dominant Form of Lambert-Eaton Myasthenic Syndrome and Nonprogressive Motor Neuropathy. American Journal of Human Genetics, 2014, 95, 332-339.	2.6	96
35	Extended phenotypic spectrum of KIF5A mutations. Neurology, 2014, 83, 612-619.	1.5	92
36	Innovative Genomic Collaboration Using the GENESIS (GEM.app) Platform. Human Mutation, 2015, 36, 950-956.	1.1	92

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37	Mechanisms of Disease: a molecular genetic update on hereditary axonal neuropathies. Nature Clinical Practice Neurology, 2006, 2, 45-53.	2.7	88
38	Alteration of ornithine metabolism leads to dominant and recessive hereditary spastic paraplegia. Brain, 2015, 138, 2191-2205.	3.7	88
39	SYNE1 ataxia is a common recessive ataxia with major non-cerebellar features: a large multi-centre study. Brain, 2016, 139, 1378-1393.	3.7	87
40	Hypomorphic mutations in POLR3A are a frequent cause of sporadic and recessive spastic ataxia. Brain, 2017, 140, 1561-1578.	3.7	85
41	Developmental Consequences of Defective ATG7-Mediated Autophagy in Humans. New England Journal of Medicine, 2021, 384, 2406-2417.	13.9	84
42	Loss of Association of REEP2 with Membranes Leads to Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2014, 94, 268-277.	2.6	83
43	Phenotypic and molecular insights into spinal muscular atrophy due to mutations in BICD2. Brain, 2015, 138, 293-310.	3.7	82
44	Hereditary Spastic Paraplegia Type 43 (SPG43) is Caused by Mutation inC19orf12. Human Mutation, 2013, 34, 1357-1360.	1.1	79
45	Mitochondrial Disease Sequence Data Resource (MSeqDR): A global grass-roots consortium to facilitate deposition, curation, annotation, and integrated analysis of genomic data for the mitochondrial disease clinical and research communities. Molecular Genetics and Metabolism, 2015, 114, 388-396.	0.5	76
46	Truncating and Missense Mutations in IGHMBP2 Cause Charcot-Marie Tooth Disease Type 2. American Journal of Human Genetics, 2014, 95, 590-601.	2.6	75
47	A recurrent WARS mutation is a novel cause of autosomal dominant distal hereditary motor neuropathy. Brain, 2017, 140, 1252-1266.	3.7	75
48	Loss of function mutations inHARScause a spectrum of inherited peripheral neuropathies. Brain, 2015, 138, 2161-2172.	3.7	71
49	Mutations in the Gene DNAJC5 Cause Autosomal Dominant Kufs Disease in a Proportion of Cases: Study of the Parry Family and 8 Other Families. PLoS ONE, 2012, 7, e29729.	1.1	70
50	Rapid in vivo forward genetic approach for identifying axon death genes in Drosophila. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 9965-9970.	3.3	70
51	FAHN/SPG35: a narrow phenotypic spectrum across disease classifications. Brain, 2019, 142, 1561-1572.	3.7	70
52	GEnomes Management Application (GEM.app): A New Software Tool for Large-Scale Collaborative Genome Analysis. Human Mutation, 2013, 34, 842-846.	1.1	69
53	TYROBP genetic variants in early-onset Alzheimer's disease. Neurobiology of Aging, 2016, 48, 222.e9-222.e15.	1.5	69
54	Exome Sequencing of a Multigenerational Human Pedigree. PLoS ONE, 2009, 4, e8232.	1.1	69

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55	Molecular genetics of autosomal-dominant axonal Charcot-Marie-Tooth disease. NeuroMolecular Medicine, 2006, 8, 63-74.	1.8	66
56	Myopathy associated BAG3 mutations lead to protein aggregation by stalling Hsp70 networks. Nature Communications, 2018, 9, 5342.	5.8	65
57	A new locus for X-linked dominant Charcot-Marie-Tooth disease (CMTX6) is caused by mutations in the pyruvate dehydrogenase kinase isoenzyme 3 (PDK3) gene. Human Molecular Genetics, 2013, 22, 1404-1416.	1.4	64
58	Mutations in phospholipase DDHD2 cause autosomal recessive hereditary spastic paraplegia (SPG54). European Journal of Human Genetics, 2013, 21, 1214-1218.	1.4	63
59	Sequencing of <scp>C</scp>harcotâ€“<scp>M</scp>arieâ€“<scp>T</scp>ooth disease genes in a toxic polyneuropathy. Annals of Neurology, 2014, 76, 727-737.	2.8	63
60	Pure and syndromic optic atrophy explained by deep intronic OPA1 mutations and an intralocus modifier. Brain, 2014, 137, 2164-2177.	3.7	62
61	Mutation screen reveals novel variants and expands the phenotypes associated with DYNC1H1. Journal of Neurology, 2015, 262, 2124-2134.	1.8	59
62	Mutations in ATP1A1 Cause Dominant Charcot-Marie-Tooth Type 2. American Journal of Human Genetics, 2018, 102, 505-514.	2.6	59
63	Motor protein mutations cause a new form of hereditary spastic paraplegia. Neurology, 2014, 82, 2007-2016.	1.5	56
64	STUB1/CHIP mutations cause Gordon Holmes syndrome as part of a widespread multisystemic neurodegeneration: evidence from four novel mutations. Orphanet Journal of Rare Diseases, 2017, 12, 31.	1.2	56
65	Phenotype and frequency of STUB1 mutations: next-generation screenings in Caucasian ataxia and spastic paraplegia cohorts. Orphanet Journal of Rare Diseases, 2014, 9, 57.	1.2	54
66	Cryptic Amyloidogenic Elements in the 3â€² UTRs of Neurofilament Genes Trigger Axonal Neuropathy. American Journal of Human Genetics, 2016, 98, 597-614.	2.6	53
67	Disruptive SCYL1 Mutations Underlie a Syndrome Characterized by Recurrent Episodes of Liver Failure, Peripheral Neuropathy, Cerebellar Atrophy, and Ataxia. American Journal of Human Genetics, 2015, 97, 855-861.	2.6	52
68	Impaired Function is a Common Feature of Neuropathy-Associated Glycyl-tRNA Synthetase Mutations. Human Mutation, 2014, 35, n/a-n/a.	1.1	51
69	Linkage and Association Study of Lateâ€Onset Alzheimer Disease Families Linked to 9p21.3. Annals of Human Genetics, 2008, 72, 725-731.	0.3	49
70	Characterization of the mitofusin 2 <scp>R94W</scp> mutation in a knockâ€in mouse model. Journal of the Peripheral Nervous System, 2014, 19, 152-164.	1.4	48
71	Electrophysiologic features of SYT2 mutations causing a treatable neuromuscular syndrome. Neurology, 2015, 85, 1964-1971.	1.5	47
72	Rare Variants in MME, Encoding Metalloprotease Neprilysin, Are Linked to Late-Onset Autosomal-Dominant Axonal Polyneuropathies. American Journal of Human Genetics, 2016, 99, 607-623.	2.6	47

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73	First de novo KCND3 mutation causes severe Kv4.3 channel dysfunction leading to early onset cerebellar ataxia, intellectual disability, oral apraxia and epilepsy. BMC Medical Genetics, 2015, 16, 51.	2.1	46
74	SCO2 mutations cause early-onset axonal Charcot-Marie-Tooth disease associated with cellular copper deficiency. Brain, 2018, 141, 662-672.	3.7	46
75	Mutation screening of spastin, atlastin, and REEP1 in hereditary spastic paraplegia. Clinical Genetics, 2011, 79, 523-530.	1.0	45
76	<scp>MORC</scp>2 mutations cause axonal <scp>C</scp>harcotâ€“<scp>M</scp>arieâ€“<scp>T</scp>ooth disease with pyramidal signs. Annals of Neurology, 2016, 79, 419-427.	2.8	44
77	Enrichment of SARM1 alleles encoding variants with constitutively hyperactive NADase in patients with ALS and other motor nerve disorders. ELife, 2021, 10, .	2.8	44
78	MSeqDR: A Centralized Knowledge Repository and Bioinformatics Web Resource to Facilitate Genomic Investigations in Mitochondrial Disease. Human Mutation, 2016, 37, 540-548.	1.1	42
79	Novel mutations in dystonin provide clues to the pathomechanisms of HSAN-VI. Neurology, 2017, 88, 2132-2140.	1.5	41
80	Association of the Charcotâ€“Marieâ€“Tooth disease gene ARHGEF10 with paclitaxel induced peripheral neuropathy in NCCTG N08CA (Alliance). Journal of the Neurological Sciences, 2015, 357, 35-40.	0.3	40
81	Absence of Dystrophin Related Protein-2 disrupts Cajal bands in a patient with Charcotâ€“Marieâ€“Tooth disease. Neuromuscular Disorders, 2015, 25, 786-793.	0.3	40
82	Truncating Mutations in UBAP1 Cause Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2019, 104, 767-773.	2.6	39
83	Genetic compensation in a stable slc25a46 mutant zebrafish: A case for using F0 CRISPR mutagenesis to study phenotypes caused by inherited disease. PLoS ONE, 2020, 15, e0230566.	1.1	39
84	Natural history of Charcot-Marie-Tooth disease type 2A: a large international multicentre study. Brain, 2020, 143, 3589-3602.	3.7	39
85	De novo PMP2mutations in families with type 1 Charcotâ€“Marieâ€“Tooth disease. Brain, 2016, 139, 1649-1656.	3.7	37
86	The human motor neuron axonal transcriptome is enriched for transcripts related to mitochondrial function and microtubule-based axonal transport. Experimental Neurology, 2018, 307, 155-163.	2.0	35
87	Variation in SIPA1L2 is correlated with phenotype modification in Charcotâ€“ Marieâ€“ Tooth disease type 1A. Annals of Neurology, 2019, 85, 316-330.	2.8	33
88	Mutation screening of mitofusin 2 in Charcot-Marie-Tooth disease type 2. Journal of Neurology, 2011, 258, 1234-1239.	1.8	32
89	Genetic background of the hereditary spastic paraplegia phenotypes in Hungary â€” An analysis of 58 probands. Journal of the Neurological Sciences, 2016, 364, 116-121.	0.3	32
90	De novo ITPR1 variants are a recurrent cause of early-onset ataxia, acting via loss of channel function. European Journal of Human Genetics, 2018, 26, 1623-1634.	1.4	32

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91	Substrate interaction defects in histidyl-tRNA synthetase linked to dominant axonal peripheral neuropathy. Human Mutation, 2018, 39, 415-432.	1.1	30
92	Beyond ALS and FTD: the phenotypic spectrum of TBK1 mutations includes PSP-like and cerebellar phenotypes. Neurobiology of Aging, 2018, 62, 244.e9-244.e13.	1.5	30
93	Insights into the genotype-phenotype correlation and molecular function of SLC25A46. Human Mutation, 2018, 39, 1995-2007.	1.1	30
94	Emerging pathways for hereditary axonopathies. Journal of Molecular Medicine, 2005, 83, 935-943.	1.7	29
95	Severe axonal Charcot-Marie-Tooth disease with proximal weakness caused byde novomutation in theMORC2gene. Brain, 2016, 139, e26-e26.	3.7	28
96	Genetic modifiers and non-Mendelian aspects of CMT. Brain Research, 2020, 1726, 146459.	1.1	28
97	Uniparental disomy of chromosome 16 unmasks recessive mutations of FA2H /SPG35 in 4 families. Neurology, 2016, 87, 186-191.	1.5	27
98	Cryptic amyloidogenic elements in mutant NEFH causing Charcot-Marie-Tooth 2 trigger aggresome formation and neuronal death. Acta Neuropathologica Communications, 2017, 5, 55.	2.4	25
99	Copy number variations are a rare cause of non-CMT1A Charcot-Marie-Tooth disease. Journal of Neurology, 2010, 257, 735-741.	1.8	24
100	Adult-onset painful axonal polyneuropathy caused by a dominantNAGLUmutation. Brain, 2015, 138, 1477-1483.	3.7	24
101	CNTNAP1 mutations cause CNS hypomyelination and neuropathy with or without arthrogryposis. Neurology: Genetics, 2017, 3, e144.	0.9	24
102	Uniparental disomy determined by wholeâ€exome sequencing in a spectrum of rare motoneuron diseases and ataxias. Molecular Genetics & Genomic Medicine, 2017, 5, 280-286.	0.6	23
103	The genetics of hereditary spastic paraplegia and implications for drug therapy. Expert Opinion on Pharmacotherapy, 2007, 8, 1433-1439.	0.9	22
104	Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia. Brain, 2021, 144, 1422-1434.	3.7	22
105	A Novel p.Leu(381)Phe Mutation in Presenilin 1 is Associated with Very Early Onset and Unusually Fast Progressing Dementia as well as Lysosomal Inclusions Typically Seen in Kufs Disease. Journal of Alzheimer's Disease, 2014, 39, 23-27.	1.2	21
106	Perspectives on the Genomics of HSP Beyond Mendelian Inheritance. Frontiers in Neurology, 2018, 9, 958.	1.1	21
107	Whole Genome Sequencing Identifies a 78 kb Insertion from Chromosome 8 as the Cause of Charcot-Marie-Tooth Neuropathy CMTX3. PLoS Genetics, 2016, 12, e1006177.	1.5	20
108	Identification of a new SYT2 variant validates an unusual distal motor neuropathy phenotype. Neurology: Genetics, 2018, 4, e282.	0.9	19

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109	GDAP2 mutations implicate susceptibility to cellular stress in a new form of cerebellar ataxia. Brain, 2018, 141, 2592-2604.	3.7	19
110	Modifier Gene Candidates in Charcot-Marie-Tooth Disease Type 1A: A Case-Only Genome-Wide Association Study. Journal of Neuromuscular Diseases, 2019, 6, 201-211.	1.1	19
111	The genetic landscape of axonal neuropathies in the middle-aged and elderly. Neurology, 2020, 95, e3163-e3179.	1.5	19
112	Genotype and phenotype distribution of 435 patients with Charcotâ€“Marieâ€“Tooth disease from central south China. European Journal of Neurology, 2021, 28, 3774-3783.	1.7	19
113	Abnormal Paraplegin Expression in Swollen Neurites, Ï„- and Î±-Synuclein Pathology in a Case of Hereditary Spastic Paraplegia SPG7 with an Ala510Val Mutation. International Journal of Molecular Sciences, 2015, 16, 25050-25066.	1.8	18
114	A novel missense mutation of <scp>CMT2P</scp> alters transcription machinery. Annals of Neurology, 2016, 80, 834-845.	2.8	18
115	A network biology approach to unraveling inherited axonopathies. Scientific Reports, 2019, 9, 1692.	1.6	18
116	Biallelic loss-of-function variations in PRDX3 cause cerebellar ataxia. Brain, 2021, 144, 1467-1481.	3.7	18
117	Rare Manifestation of a c.290 C>T, p.Gly97GluVCPMutation. Case Reports in Genetics, 2015, 2015, 1-5.	0.1	16
118	Commonalities across computational workflows for uncovering explanatory variants in undiagnosed cases. Genetics in Medicine, 2021, 23, 1075-1085.	1.1	16
119	Assessing non-Mendelian inheritance in inherited axonopathies. Genetics in Medicine, 2020, 22, 2114-2119.	1.1	15
120	The ARCA Registry: A Collaborative Global Platform for Advancing Trial Readiness in Autosomal Recessive Cerebellar Ataxias. Frontiers in Neurology, 2021, 12, 677551.	1.1	15
121	Hereditary spastic paraplegia is a novel phenotype for germline de novo ATP1A1 mutation. Clinical Genetics, 2020, 97, 521-526.	1.0	14
122	De Novo and Inherited Variants in GBF1 are Associated with Axonal Neuropathy Caused by Golgi Fragmentation. American Journal of Human Genetics, 2020, 107, 763-777.	2.6	14
123	RFC1 repeat expansions: A recurrent cause of sensory and autonomic neuropathy with cough and ataxia. European Journal of Neurology, 2022, 29, 2156-2161.	1.7	14
124	Variant pathogenicity evaluation in the community-driven Inherited Neuropathy Variant Browser. Human Mutation, 2018, 39, 635-642.	1.1	13
125	Molecular Genetics of Autosomal-Dominant Axonal Charcot-Marie-Tooth Disease. NeuroMolecular Medicine, 2006, 8, 63-74.	1.8	12
126	Update on psychiatric genetics. Genetics in Medicine, 2007, 9, 332-340.	1.1	12

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127	Large scale in silico characterization of repeat expansion variation in human genomes. Scientific Data, 2020, 7, 294.	2.4	12
128	Whole genome sequencing identifies causal variants in CMT. Nature Reviews Neurology, 2010, 6, 424-425.	4.9	11
129	A neuropathyâ€associated kinesin KIF1A mutation hyperâ€stabilizes the motorâ€neck interaction during the ATPase cycle. EMBO Journal, 2022, 41, e108899.	3.5	11
130	Whole Genome Sequencing and a New Bioinformatics Platform Allow for Rapid Gene Identification in D. melanogaster EMS Screens. Biology, 2012, 1, 766-777.	1.3	10
131	A CADM3 variant causes Charcot-Marie-Tooth disease with marked upper limb involvement. Brain, 2021, 144, 1197-1213.	3.7	10
132	Restoring Shank3 in the rostral brainstem of shank3abâ^'/â^' zebrafish autism models rescues sensory deficits. Communications Biology, 2021, 4, 1411.	2.0	10
133	Isoform-specific loss of dystonin causes hereditary motor and sensory neuropathy. Neurology: Genetics, 2020, 6, e496.	0.9	9
134	Functional Network Profiles in ARSACS Disclosed by Aptamer-Based Proteomic Technology. Frontiers in Neurology, 2020, 11, 603774.	1.1	9
135	Rare mutations in ATL3, SPTLC2 and SCN9A explaining hereditary sensory neuropathy and congenital insensitivity to pain in a Brazilian cohort. Journal of the Neurological Sciences, 2021, 427, 117498.	0.3	9
136	De Novo and Dominantly Inherited <scp>SPTAN1</scp> Mutations Cause Spastic Paraplegia and Cerebellar Ataxia. Movement Disorders, 2022, 37, 1175-1186.	2.2	9
137	Confirmation of TACO1 as a Leigh Syndrome Disease Gene in Two Additional Families. Journal of Neuromuscular Diseases, 2020, 7, 301-308.	1.1	8
138	POLG mutations presenting as Charcotâ€Marieâ€Tooth disease. Journal of the Peripheral Nervous System, 2019, 24, 213-218.	1.4	6
139	A mutation in the heptad repeat 2 domain of MFN2 in a large CMT2A family. Journal of the Peripheral Nervous System, 2018, 23, 36-39.	1.4	5
140	A novel MFN2 mutation causes variable clinical severity in a multi-generational CMT2 family. Neuromuscular Disorders, 2019, 29, 134-137.	0.3	5
141	De Novo ATP1A1 Variants in an Early-Onset Complex Neurodevelopmental Syndrome. Neurology, 2022, 98, 440-445.	1.5	5
142	Insights into the pathogenesis of ATP1A1 â€related CMT disease using patientâ€specific iPSCs. Journal of the Peripheral Nervous System, 2019, 24, 330-339.	1.4	4
143	Expanding PRDX3 disease: broad range of onset age and infratentorial MRI signal changes. Brain, 2022, 145, e95-e98.	3.7	3
144	Translesion DNA synthesis-driven mutagenesis in very early embryogenesis of fast cleaving embryos. Nucleic Acids Research, 2022, 50, 885-898.	6.5	2

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145	Schwann cell gene therapies in sight. Gene Therapy, 2021, 28, 618-619.	2.3	1
146	Introduction to Applications of Genomic Sequencing. , 2016, , 427-433.		0
147	Charcotâ€Marieâ€Tooth disease in Africa. Journal of the Peripheral Nervous System, 2022, 27, 98-99.	1.4	0