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	De Novo ATP1A1 Variants in an Early-Onset Complex Neurodevelopmental Syndrome. Neurology, 2022, 98, 440-445.	1.5	5
2	De Novo and Dominantly Inherited <scp>SPTAN1</scp> Mutations Cause Spastic Paraplegia and Cerebellar Ataxia. Movement Disorders, 2022, 37, 1175-1186.	2.2	9
3	A neuropathyâ€associated kinesin KIF1A mutation hyperâ€stabilizes the motorâ€neck interaction during the ATPase cycle. EMBO Journal, 2022, 41, e108899.	3.5	11
4	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
5	Charcotâ€Marieâ€Tooth disease in Africa. Journal of the Peripheral Nervous System, 2022, 27, 98-99.	1.4	0
6	Translesion DNA synthesis-driven mutagenesis in very early embryogenesis of fast cleaving embryos. Nucleic Acids Research, 2022, 50, 885-898.	6.5	2
7	Expanding PRDX3 disease: broad range of onset age and infratentorial MRI signal changes. Brain, 2022, 145, e95-e98.	3.7	3
8	Commonalities across computational workflows for uncovering explanatory variants in undiagnosed cases. Genetics in Medicine, 2021, 23, 1075-1085.	1.1	16
9	Biallelic loss-of-function variations in PRDX3 cause cerebellar ataxia. Brain, 2021, 144, 1467-1481.	3.7	18
10	A CADM3 variant causes Charcot-Marie-Tooth disease with marked upper limb involvement. Brain, 2021, 144, 1197-1213.	3.7	10
11	Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia. Brain, 2021, 144, 1422-1434.	3.7	22
12	Schwann cell gene therapies in sight. Gene Therapy, 2021, 28, 618-619.	2.3	1
13	Developmental Consequences of Defective ATG7-Mediated Autophagy in Humans. New England Journal of Medicine, 2021, 384, 2406-2417.	13.9	84
14	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
15	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
16	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
17	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
18	Restoring Shank3 in the rostral brainstem of shank3abâ¯/â¯ zebrafish autism models rescues sensory deficits. Communications Biology, 2021, 4, 1411.	2.0	10

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19	Genetic modifiers and non-Mendelian aspects of CMT. Brain Research, 2020, 1726, 146459.	1.1	28
20	Hereditary spastic paraplegia is a novel phenotype for germline de novo ATP1A1 mutation. Clinical Genetics, 2020, 97, 521-526.	1.0	14
21	Isoform-specific loss of dystonin causes hereditary motor and sensory neuropathy. Neurology: Genetics, 2020, 6, e496.	0.9	9
22	Assessing non-Mendelian inheritance in inherited axonopathies. Genetics in Medicine, 2020, 22, 2114-2119.	1.1	15
23	Large scale in silico characterization of repeat expansion variation in human genomes. Scientific Data, 2020, 7, 294.	2.4	12
24	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
25	The genetic landscape of axonal neuropathies in the middle-aged and elderly. Neurology, 2020, 95, e3163-e3179.	1.5	19
26	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
27	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
28	Functional Network Profiles in ARSACS Disclosed by Aptamer-Based Proteomic Technology. Frontiers in Neurology, 2020, 11, 603774.	1.1	9
29	Natural history of Charcot-Marie-Tooth disease type 2A: a large international multicentre study. Brain, 2020, 143, 3589-3602.	3.7	39
30	Confirmation of TACO1 as a Leigh Syndrome Disease Gene in Two Additional Families. Journal of Neuromuscular Diseases, 2020, 7, 301-308.	1.1	8
31	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
32	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
33	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
34	FAHN/SPG35: a narrow phenotypic spectrum across disease classifications. Brain, 2019, 142, 1561-1572.	3.7	70
35	POLG mutations presenting as Charcotâ€Marieâ€Tooth disease. Journal of the Peripheral Nervous System, 2019, 24, 213-218.	1.4	6
36	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

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37	Truncating Mutations in UBAP1 Cause Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2019, 104, 767-773.	2.6	39
38	A network biology approach to unraveling inherited axonopathies. Scientific Reports, 2019, 9, 1692.	1.6	18
39	A novel MFN2 mutation causes variable clinical severity in a multi-generational CMT2 family. Neuromuscular Disorders, 2019, 29, 134-137.	0.3	5
40	Variant pathogenicity evaluation in the community-driven Inherited Neuropathy Variant Browser. Human Mutation, 2018, 39, 635-642.	1.1	13
41	Mutations in ATP1A1 Cause Dominant Charcot-Marie-Tooth Type 2. American Journal of Human Genetics, 2018, 102, 505-514.	2.6	59
42	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
43	SCO2 mutations cause early-onset axonal Charcot-Marie-Tooth disease associated with cellular copper deficiency. Brain, 2018, 141, 662-672.	3.7	46
44	Substrate interaction defects in histidyl-tRNA synthetase linked to dominant axonal peripheral neuropathy. Human Mutation, 2018, 39, 415-432.	1.1	30
45	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
46	Identification of a new SYT2 variant validates an unusual distal motor neuropathy phenotype. Neurology: Genetics, 2018, 4, e282.	0.9	19
47	Myopathy associated BAG3 mutations lead to protein aggregation by stalling Hsp70 networks. Nature Communications, 2018, 9, 5342.	5.8	65
48	Perspectives on the Genomics of HSP Beyond Mendelian Inheritance. Frontiers in Neurology, 2018, 9, 958.	1.1	21
49	Insights into the genotype-phenotype correlation and molecular function of SLC25A46. Human Mutation, 2018, 39, 1995-2007.	1.1	30
50	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
51	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
52	GDAP2 mutations implicate susceptibility to cellular stress in a new form of cerebellar ataxia. Brain, 2018, 141, 2592-2604.	3.7	19
53	The Human Phenotype Ontology in 2017. Nucleic Acids Research, 2017, 45, D865-D876.	6.5	699
54	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

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55	Hypomorphic mutations in POLR3A are a frequent cause of sporadic and recessive spastic ataxia. Brain, 2017, 140, 1561-1578.	3.7	85
56	Novel mutations in dystonin provide clues to the pathomechanisms of HSAN-VI. Neurology, 2017, 88, 2132-2140.	1.5	41
57	CNTNAP1 mutations cause CNS hypomyelination and neuropathy with or without arthrogryposis. Neurology: Genetics, 2017, 3, e144.	0.9	24
58	Loss-of-function mutations in theATP13A2/PARK9 gene cause complicated hereditary spastic paraplegia (SPG78). Brain, 2017, 140, 287-305.	3.7	135
59	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
60	A recurrent WARS mutation is a novel cause of autosomal dominant distal hereditary motor neuropathy. Brain, 2017, 140, 1252-1266.	3.7	75
61	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
62	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
63	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
64	<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
65	Hereditary spastic paraplegia: Clinicogenetic lessons from 608 patients. Annals of Neurology, 2016, 79, 646-658.	2.8	218
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	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
68	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
69	A novel missense mutation of <scp>CMT2P</scp> alters transcription machinery. Annals of Neurology, 2016, 80, 834-845.	2.8	18
70	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
71	TYROBP genetic variants in early-onset Alzheimer's disease. Neurobiology of Aging, 2016, 48, 222.e9-222.e15.	1.5	69
72	Uniparental disomy of chromosome 16 unmasks recessive mutations of FA2H /SPG35 in 4 families. Neurology, 2016, 87, 186-191.	1.5	27

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73	De novo PMP2mutations in families with type 1 Charcotâ€“Marieâ€“Tooth disease. Brain, 2016, 139, 1649-1656.	3.7	37
74	Severe axonal Charcot-Marie-Tooth disease with proximal weakness caused byde novomutation in theMORC2gene. Brain, 2016, 139, e26-e26.	3.7	28
75	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
76	Introduction to Applications of Genomic Sequencing. , 2016, , 427-433.		0
77	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
78	Rare Manifestation of a c.290 C>T, p.Gly97GluVCPMutation. Case Reports in Genetics, 2015, 2015, 1-5.	0.1	16
79	Innovative Genomic Collaboration Using the GENESIS (GEM.app) Platform. Human Mutation, 2015, 36, 950-956.	1.1	92
80	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
81	Alteration of ornithine metabolism leads to dominant and recessive hereditary spastic paraplegia. Brain, 2015, 138, 2191-2205.	3.7	88
82	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
83	Mutations in SLC25A46, encoding a UGO1-like protein, cause an optic atrophy spectrum disorder. Nature Genetics, 2015, 47, 926-932.	9.4	166
84	Loss of function mutations inHARScause a spectrum of inherited peripheral neuropathies. Brain, 2015, 138, 2161-2172.	3.7	71
85	Adult-onset painful axonal polyneuropathy caused by a dominantNAGLUmutation. Brain, 2015, 138, 1477-1483.	3.7	24
86	Mutation screen reveals novel variants and expands the phenotypes associated with DYNC1H1. Journal of Neurology, 2015, 262, 2124-2134.	1.8	59
87	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
88	Electrophysiologic features of SYT2 mutations causing a treatable neuromuscular syndrome. Neurology, 2015, 85, 1964-1971.	1.5	47
89	The Matchmaker Exchange: A Platform for Rare Disease Gene Discovery. Human Mutation, 2015, 36, 915-921.	1.1	390
90	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

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91	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
92	Phenotypic and molecular insights into spinal muscular atrophy due to mutations in BICD2. Brain, 2015, 138, 293-310.	3.7	82
93	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
94	Motor protein mutations cause a new form of hereditary spastic paraplegia. Neurology, 2014, 82, 2007-2016.	1.5	56
95	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
96	Impaired Function is a Common Feature of Neuropathy-Associated Glycyl-tRNA Synthetase Mutations. Human Mutation, 2014, 35, n/a-n/a.	1.1	51
97	Pure and syndromic optic atrophy explained by deep intronic OPA1 mutations and an intralocus modifier. Brain, 2014, 137, 2164-2177.	3.7	62
98	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
99	Absence of BiP Co-chaperone DNAJC3 Causes Diabetes Mellitus and Multisystemic Neurodegeneration. American Journal of Human Genetics, 2014, 95, 689-697.	2.6	100
100	Genetics of Charcot-Marie-Tooth (CMT) Disease within the Frame of the Human Genome Project Success. Genes, 2014, 5, 13-32.	1.0	203
101	PNPLA6 mutations cause Boucher-NeuhÃuser and Gordon Holmes syndromes as part of a broad neurodegenerative spectrum. Brain, 2014, 137, 69-77.	3.7	189
102	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
103	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
104	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
105	Extended phenotypic spectrum of KIF5A mutations. Neurology, 2014, 83, 612-619.	1.5	92
106	A novel mutation in VCP causes Charcotâ€“Marieâ€“Tooth Type 2 disease. Brain, 2014, 137, 2897-2902.	3.7	116
107	Loss of Association of REEP2 with Membranes Leads to Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2014, 94, 268-277.	2.6	83
108	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

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109	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
110	Hereditary Spastic Paraplegia Type 43 (SPG43) is Caused by Mutation inC19orf12. Human Mutation, 2013, 34, 1357-1360.	1.1	79
111	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
112	GEnomes Management Application (GEM.app): A New Software Tool for Large-Scale Collaborative Genome Analysis. Human Mutation, 2013, 34, 842-846.	1.1	69
113	Alteration of Ganglioside Biosynthesis Responsible for Complex Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2013, 93, 118-123.	2.6	151
114	Mutations in phospholipase DDHD2 cause autosomal recessive hereditary spastic paraplegia (SPG54). European Journal of Human Genetics, 2013, 21, 1214-1218.	1.4	63
115	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
116	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
117	Loss-of-function mutations in HINT1 cause axonal neuropathy with neuromyotonia. Nature Genetics, 2012, 44, 1080-1083.	9.4	102
118	Evaluating Pathogenicity of Rare Variants From Dilated Cardiomyopathy in the Exome Era. Circulation: Cardiovascular Genetics, 2012, 5, 167-174.	5.1	112
119	dSarm/Sarm1 Is Required for Activation of an Injury-Induced Axon Death Pathway. Science, 2012, 337, 481-484.	6.0	558
120	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
121	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
122	Whole-Exome Sequencing Efficiently Detects Rare Mutations in Autosomal Recessive Nonsyndromic Hearing Loss. PLoS ONE, 2012, 7, e50628.	1.1	143
123	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
124	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
125	Mutation screening of spastin, atlastin, and REEP1 in hereditary spastic paraplegia. Clinical Genetics, 2011, 79, 523-530.	1.0	45
126	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

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127	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
128	Mutation screening of mitofusin 2 in Charcot-Marie-Tooth disease type 2. Journal of Neurology, 2011, 258, 1234-1239.	1.8	32
129	Exome sequencing allows for rapid gene identification in a Charcotâ€Marieâ€Tooth family. Annals of Neurology, 2011, 69, 464-470.	2.8	107
130	Copy number variations are a rare cause of non-CMT1A Charcot-Marie-Tooth disease. Journal of Neurology, 2010, 257, 735-741.	1.8	24
131	Whole genome sequencing identifies causal variants in CMT. Nature Reviews Neurology, 2010, 6, 424-425.	4.9	11
132	Multiple rare SAPAP3 missense variants in trichotillomania and OCD. Molecular Psychiatry, 2009, 14, 6-9.	4.1	166
133	Exome Sequencing of a Multigenerational Human Pedigree. PLoS ONE, 2009, 4, e8232.	1.1	69
134	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
135	REEP1 mutation spectrum and genotype/phenotype correlation in hereditary spastic paraplegia type 31. Brain, 2008, 131, 1078-1086.	3.7	163
136	Update on psychiatric genetics. Genetics in Medicine, 2007, 9, 332-340.	1.1	12
137	The genetics of hereditary spastic paraplegia and implications for drug therapy. Expert Opinion on Pharmacotherapy, 2007, 8, 1433-1439.	0.9	22
138	Mechanisms of Disease: a molecular genetic update on hereditary axonal neuropathies. Nature Clinical Practice Neurology, 2006, 2, 45-53.	2.7	88
139	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
140	Molecular genetics of autosomal-dominant axonal Charcot-Marie-Tooth disease. NeuroMolecular Medicine, 2006, 8, 63-74.	1.8	66
141	Molecular Genetics of Autosomal-Dominant Axonal Charcot-Marie-Tooth Disease. NeuroMolecular Medicine, 2006, 8, 63-74.	1.8	12
142	Axonal neuropathy with optic atrophy is caused by mutations in mitofusin 2. Annals of Neurology, 2006, 59, 276-281.	2.8	380
143	MFN2 mutation distribution and genotype/phenotype correlation in Charcot-Marie-Tooth type 2. Brain, 2006, 129, 2093-2102.	3.7	351
144	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

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145	Emerging pathways for hereditary axonopathies. Journal of Molecular Medicine, 2005, 83, 935-943.	1.7	29
146	Mutations in the mitochondrial GTPase mitofusin 2 cause Charcot-Marie-Tooth neuropathy type 2A. Nature Genetics, 2004, 36, 449-451.	9.4	1,391
147	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