Vincent Timmerman

List of Publications by Year
in descending order

188 papers	16,439 citations	16451 64 h-index	17592 121 g-index
194 all docs	194 docs citations	194 times ranked	15625 citing authors

#	Article	IF	Citations
1	De Novo and Dominantly Inherited <scp>SPTAN1</scp> Mutations Cause Spastic Paraplegia and Cerebellar Ataxia. Movement Disorders, 2022, 37, 1175-1186.	3.9	9
2	NCAM1 and GDF15 are biomarkers of Charcot-Marie-Tooth disease in patients and mice. Brain, 2022, 145, 3999-4015.	7.6	12
3	Oligodendroglia-derived extracellular vesicles activate autophagy via LC3B/BAG3 to protect against oxidative stress with an enhanced effect for HSPB8 enriched vesicles. Cell Communication and Signaling, 2022, 20, 58.	6.5	10
4	Genetic pain loss disorders. Nature Reviews Disease Primers, 2022, 8, .	30.5	18
5	246th ENMC International Workshop: Protein aggregate myopathies 24â€“26 May 2019, Hoofddorp, The Netherlands. Neuromuscular Disorders, 2021, 31, 158-166.	0.6	5
6	A weakened interface in the P182L variant of HSP27 associated with severe Charcotâ€Marieâ€Tooth neuropathy causes aberrant binding to interacting proteins. EMBO Journal, 2021, 40, e103811.	7.8	14
7	Induced pluripotent stem cell-derived motor neurons of CMT type 2 patients reveal progressive mitochondrial dysfunction. Brain, 2021, 144, 2471-2485.	7.6	27
8	Microglial derived extracellular vesicles activate autophagy and mediate multiâ€target signaling to maintain cellular homeostasis. Journal of Extracellular Vesicles, 2020, 10, e12022.	12.2	28
9	BAG3 Pro209 mutants associated with myopathy and neuropathy relocate chaperones of the CASA-complex to aggresomes. Scientific Reports, 2020, 10, 8755.	3.3	32
10	Profiling peripheral nerve macrophages reveals two macrophage subsets with distinct localization, transcriptome and response to injury. Nature Neuroscience, 2020, 23, 676-689.	14.8	148
11	Small heat shock proteins in neurodegenerative diseases. Cell Stress and Chaperones, 2020, 25, 679-699.	2.9	57
12	The CMT1A duplication. Medizinische Genetik, 2020, 32, 195-205.	0.2	0
13	Challenges in modelling the Charcot-Marie-Tooth neuropathies for therapy development. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, 58-67.	1.9	61
14	Report of a novel ATP7A mutation causing distal motor neuropathy. Neuromuscular Disorders, 2019, 29, 776-785.	0.6	15
15	Defects in Axonal Transport in Inherited Neuropathies. Journal of Neuromuscular Diseases, 2019, 6, 401-419.	2.6	23
16	Neuropathy-causing mutations in HSPB1 impair autophagy by disturbing the formation of SQSTM1/p62 bodies. Autophagy, 2019, 15, 1051-1068.	9.1	56
17	Loss of Neurological Disease HSAN-I-Associated Gene SPTLC2 Impairs CD8+ T Cell Responses to Infection by Inhibiting T Cell Metabolic Fitness. Immunity, 2019, 50, 1218-1231.e5.	14.3	30
18	Biallelic VARS variants cause developmental encephalopathy with microcephaly that is recapitulated in vars knockout zebrafish. Nature Communications, 2019, 10, 708.	12.8	40

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19	Altered interplay between endoplasmic reticulum and mitochondria in Charcotâ€“Marieâ€“Tooth type 2A neuropathy. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 2328-2337.	7.1	73
20	Sensory neuropathy-causing mutations in ATL3 affect ERâ€“mitochondria contact sites and impair axonal mitochondrial distribution. Human Molecular Genetics, 2019, 28, 615-627.	2.9	31
21	PFN2 and GAMT as common molecular determinants of axonal Charcot-Marie-Tooth disease. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 870-878.	1.9	16
22	A knock-in/knock-out mouse model of HSPB8-associated distal hereditary motor neuropathy and myopathy reveals toxic gain-of-function of mutant Hspb8. Acta Neuropathologica, 2018, 135, 131-148.	7.7	58
23	Sensory-Neuropathy-Causing Mutations in ATL3 Cause Aberrant ER Membrane Tethering. Cell Reports, 2018, 23, 2026-2038.	6.4	29
24	Axonal Neuropathies due to Mutations in Small Heat Shock Proteins: Clinical, Genetic, and Functional Insights into Novel Mutations. Human Mutation, 2017, 38, 556-568.	2.5	54
25	Novel insights in the disease biology of mutant small heat shock proteins in neuromuscular diseases. Brain, 2017, 140, 2541-2549.	7.6	32
26	Mutant HSPB1 causes loss of translational repression by binding to PCBP1, an RNA binding protein with a possible role in neurodegenerative disease. Acta Neuropathologica Communications, 2017, 5, 5.	5.2	29
27	HSPB1 facilitates ERK-mediated phosphorylation and degradation of BIM to attenuate endoplasmic reticulum stress-induced apoptosis. Cell Death and Disease, 2017, 8, e3026-e3026.	6.3	33
28	Autophagy as an Emerging Common Pathomechanism in Inherited Peripheral Neuropathies. Frontiers in Molecular Neuroscience, 2017, 10, 143.	2.9	31
29	Characterization of New Transgenic Mouse Models for Two Charcot-Marie-Tooth-Causing HspB1 Mutations using the Rosa26 Locus. Journal of Neuromuscular Diseases, 2016, 3, 183-200.	2.6	9
30	The hnRNP family: insights into their role in health and disease. Human Genetics, 2016, 135, 851-867.	3.8	720
31	Charcotâ€“Marieâ€“Tooth disease type 2G redefined by a novel mutation in LRSAM1. Annals of Neurology, 2016, 80, 823-833.	5.3	13
32	Molecular Chaperones in the Pathogenesis of Amyotrophic Lateral Sclerosis: The Role of HSPB1. Human Mutation, 2016, 37, 1202-1208.	2.5	45
33	Mitochondria-associated membranes as hubs for neurodegeneration. Acta Neuropathologica, 2016, 131, 505-523.	7.7	172
34	Nlrp6 promotes recovery after peripheral nerve injury independently of inflammasomes. Journal of Neuroinflammation, 2015, 12, 143.	7.2	42
35	Novel Mutations in the DYNC1H1 Tail Domain Refine the Genetic and Clinical Spectrum of Dyneinopathies. Human Mutation, 2015, 36, 287-291.	2.5	36
36	Recessive mutations inSLC13A5result in a loss of citrate transport and cause neonatal epilepsy, developmental delay and teeth hypoplasia. Brain, 2015, 138, 3238-3250.	7.6	96

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37	Unraveling the genetic landscape of autosomal recessive Charcot-Marie-Tooth neuropathies using a homozygosity mapping approach. Neurogenetics, 2015, 16, 33-42.	1.4	29
38	Promising riboflavin treatment for motor neuron disorder. Brain, 2014, 137, 2-3.	7.6	6
39	Sensory neuropathy with bone destruction due to a mutation in the membrane-shaping atlastin GTPase 3. Brain, 2014, 137, 683-692.	7.6	80
40	Recent advances in Charcotâ€“Marieâ€“Tooth disease. Current Opinion in Neurology, 2014, 27, 532-540.	3.6	60
41	Genetics of Charcot-Marie-Tooth (CMT) Disease within the Frame of the Human Genome Project Success. Genes, 2014, 5, 13-32.	2.4	203
42	Whole-exome sequencing in patients with inherited neuropathies: outcome and challenges. Journal of Neurology, 2014, 261, 970-982.	3.6	50
43	CMT-associated mutations in glycyl- and tyrosyl-tRNA synthetases exhibit similar pattern of toxicity and share common genetic modifiers in Drosophila. Neurobiology of Disease, 2014, 68, 180-189.	4.4	34
44	Human Rab7 mutation mimics features of Charcotâ€“Marieâ€“Tooth neuropathy type 2B in Drosophila. Neurobiology of Disease, 2014, 65, 211-219.	4.4	28
45	Molecular Defects in the Motor Adaptor BICD2 Cause Proximal Spinal Muscular Atrophy with Autosomal-Dominant Inheritance. American Journal of Human Genetics, 2013, 92, 955-964.	6.2	112
46	Charcotâ€“Marieâ€“Tooth causing HSPB1 mutations increase Cdk5-mediated phosphorylation of neurofilaments. Acta Neuropathologica, 2013, 126, 93-108.	7.7	43
47	Mutations at Ser331 in the HSN type I gene SPTLC1 are associated with a distinct syndromic phenotype. European Journal of Medical Genetics, 2013, 56, 266-269.	1.3	35
48	Biopsy in a patient with PMP22 exon 2 mutation recapitulates pathology of Trembler-J mouse. Neuromuscular Disorders, 2013, 23, 345-348.	0.6	8
49	A de novo gain-of-function mutation in SCN11A causes loss of pain perception. Nature Genetics, 2013, 45, 1399-1404.	21.4	264
50	Animal models and therapeutic prospects for Charcotâ€“Marieâ€“Tooth disease. Annals of Neurology, 2013, 74, 391-396.	5.3	30
51	The neuroinflammatory role of Schwann cells in disease. Neurobiology of Disease, 2013, 55, 95-103.	4.4	97
52	Overlapping molecular pathological themes link Charcotâ€“Marieâ€“Tooth neuropathies and hereditary spastic paraplegias. Experimental Neurology, 2013, 246, 14-25.	4.1	64
53	HSPB1 Facilitates the Formation of Non-Centrosomal Microtubules. PLoS ONE, 2013, 8, e66541.	2.5	14
54	Loss-of-function mutations in HINT1 cause axonal neuropathy with neuromyotonia. Nature Genetics, 2012, 44, 1080-1083.	21.4	102

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55	Polymorphisms in the GluR2 gene are not associated with amyotrophic lateral sclerosis. Neurobiology of Aging, 2012, 33, 418-420.	3.1	8
56	Mutant HSPB8 causes protein aggregates and a reduced mitochondrial membrane potential in dermal fibroblasts from distal hereditary motor neuropathy patients. Neuromuscular Disorders, 2012, 22, 699-711.	0.6	30
57	Exome Sequencing Identifies a REEP1 Mutation Involved in Distal Hereditary Motor Neuropathy Type V. American Journal of Human Genetics, 2012, 91, 139-145.	6.2	83
58	Acute injury in the peripheral nervous system triggers an alternative macrophage response. Journal of Neuroinflammation, 2012, 9, 176.	7.2	134
59	Neurofilament phosphorylation and their prolineâ€directed kinases in health and disease. Journal of the Peripheral Nervous System, 2012, 17, 365-376.	3.1	38
60	G.O.7 Distal myopathy with upper limb predominance caused by filamin C haploinsufficiency. Neuromuscular Disorders, 2012, 22, 874-875.	0.6	0
61	Mechanisms of disease in hereditary sensory and autonomic neuropathies. Nature Reviews Neurology, 2012, 8, 73-85.	10.1	140
62	Drosophila as a platform to predict the pathogenicity of novel aminoacyl-tRNA synthetase mutations in CMT. Amino Acids, 2012, 42, 1661-1668.	2.7	11
63	Inherited peripheral neuropathies: a myriad of genes and complex phenotypes. Brain, 2011, 134, 1587-1590.	7.6	5
64	Increased axonal ribosome numbers in CMT diseases. Journal of the Peripheral Nervous System, 2011, 16, 71-73.	3.1	3
65	HDAC6 inhibitors reverse axonal loss in a mouse model of mutant HSPB1â€“induced Charcot-Marie-Tooth disease. Nature Medicine, 2011, 17, 968-974.	30.7	405
66	Microtubule dynamics in the peripheral nervous system. Bioarchitecture, 2011, 1, 267-270.	1.5	32
67	KIF1A, an Axonal Transporter of Synaptic Vesicles, Is Mutated in Hereditary Sensory and Autonomic Neuropathy Type 2. American Journal of Human Genetics, 2011, 89, 219-230.	6.2	172
68	Reduced penetrance in hereditary motor neuropathy caused by TRPV4 Arg269Cys mutation. Journal of Neurology, 2011, 258, 1413-1421.	3.6	24
69	Characterization of two mutations in the SPTLC1 subunit of serine palmitoyltransferase associated with hereditary sensory and autonomic neuropathy type I. Human Mutation, 2011, 32, E2211-E2225.	2.5	37
70	Genetic spectrum of hereditary neuropathies with onset in the first year of life. Brain, 2011, 134, 2664-2676.	7.6	112
71	Small Heat-Shock Protein HSPB1 Mutants Stabilize Microtubules in Charcot-Marie-Tooth Neuropathy. Journal of Neuroscience, 2011, 31, 15320-15328.	3.6	95
72	Autosomal-Dominant Striatal Degeneration Is Caused by a Mutation in the Phosphodiesterase 8B Gene. American Journal of Human Genetics, 2010, 86, 83-87.	6.2	35

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73	Missense Mutations in the Copper Transporter Gene ATP7A Cause X-Linked Distal Hereditary Motor Neuropathy. American Journal of Human Genetics, 2010, 86, 343-352.	6.2	170
74	Mechanisms for Nonrecurrent Genomic Rearrangements Associated with CMT1A or HNPP: Rare CNVs as a Cause for Missing Heritability. American Journal of Human Genetics, 2010, 86, 892-903.	6.2	125
75	Mutations in the SPTLC2 Subunit of Serine Palmitoyltransferase Cause Hereditary Sensory and Autonomic Neuropathy Type I. American Journal of Human Genetics, 2010, 87, 513-522.	6.2	159
76	L239F founder mutation in GDAP1 is associated with a mild Charcotâ€“Marieâ€“Tooth type 4C4 (CMT4C4) phenotype. Neurogenetics, 2010, 11, 357-366.	1.4	15
77	Tollâ€like receptor expression in the peripheral nerve. Glia, 2010, 58, 1701-1709.	4.9	121
78	Clinical utility gene card for: HMSN/HNPP HMSN types 1, 2, 3, 6 (CMT1,2,4, DSN, CHN, GAN, CCFDN, HNA); HNPP. European Journal of Human Genetics, 2010, 18, 1071-1071.	2.8	2
79	Mutant HSPB8 causes motor neuron-specific neurite degeneration. Human Molecular Genetics, 2010, 19, 3254-3265.	2.9	83
80	Cell Deathâ€“Mediated Cleavage of the Attraction Signal p43 in Human Atherosclerosis. Arteriosclerosis, Thrombosis, and Vascular Biology, 2010, 30, 1415-1422.	2.4	8
81	Dominant mutations in the cation channel gene transient receptor potential vanilloid 4 cause an unusual spectrum of neuropathies. Brain, 2010, 133, 1798-1809.	7.6	113
82	N88S mutation in the BSCL2 gene in a Serbian family with distal hereditary motor neuropathy type V or Silver syndrome. Journal of the Neurological Sciences, 2010, 296, 107-109.	0.6	9
83	Increased Monomerization of Mutant HSPB1 Leads to Protein Hyperactivity in Charcot-Marie-Tooth Neuropathy. Journal of Biological Chemistry, 2010, 285, 12778-12786.	3.4	95
84	Dominant mutations in the tyrosyl-tRNA synthetase gene recapitulate in Drosophila features of human Charcotâ€“Marieâ€“Tooth neuropathy. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 11782-11787.	7.1	96
85	Phenotypic spectrum of dynamin 2 mutations in Charcot-Marie-Tooth neuropathy. Brain, 2009, 132, 1741-1752.	7.6	134
86	Humoral immunodeficiency in congenital insensitivity to pain with anhidrosis. Neurogenetics, 2009, 10, 161-165.	1.4	23
87	A systematic comparison of all mutations in hereditary sensory neuropathy type I (HSAN I) reveals that the G387A mutation is not disease associated. Neurogenetics, 2009, 10, 135-143.	1.4	26
88	Mutations in FAM134B, encoding a newly identified Golgi protein, cause severe sensory and autonomic neuropathy. Nature Genetics, 2009, 41, 1179-1181.	21.4	205
89	Peripheral neuropathy and 46XY gonadal dysgenesis: A heterogeneous entity. Neuromuscular Disorders, 2009, 19, 172-175.	0.6	11
90	Genes for hereditary sensory and autonomic neuropathies: a genotype-phenotype correlation. Brain, 2009, 132, 2699-2711.	7.6	202

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91	Magnetic resonance imaging findings of leg musculature in Charcot-Marie-Tooth disease type 2 due to dynamin 2 mutation. Journal of Neurology, 2008, 255, 986-992.	3.6	35
92	Charcotâ€Marieâ€Tooth Disease: A Clinicoâ€genetic Confrontation. Annals of Human Genetics, 2008, 72, 416-441.	0.8	136
93	Mutation Scanning the GJB1 Gene with High-Resolution Melting Analysis: Implications for Mutation Scanning of Genes for Charcot-Marie-Tooth Disease. Clinical Chemistry, 2007, 53, 349-352.	3.2	53
94	Astrocytes regulate GluR2 expression in motor neurons and their vulnerability to excitotoxicity. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 14825-14830.	7.1	193
95	Genotypeâ€“phenotype analysis in patients with giant axonal neuropathy (GAN). Neuromuscular Disorders, 2007, 17, 624-630.	0.6	29
96	Clinical and Electrophysiological Features in Charcot-Marie-Tooth Disease With Mutations in the NEFL Gene. Archives of Neurology, 2007, 64, 966.	4.5	37
97	Peripheral Nerve Demyelination Caused by a Mutant Rho GTPase Guanine Nucleotide Exchange Factor, Frabin/FGD4. American Journal of Human Genetics, 2007, 81, 158-164.	6.2	128
98	Relative contribution of mutations in genes for autosomal dominant distal hereditary motor neuropathies: a genotype-phenotype correlation study. Brain, 2007, 131, 1217-1227.	7.6	113
99	Genetic variant in theHSPB1 promoter region impairs the HSP27 stress response. Human Mutation, 2007, 28, 830-830.	2.5	47
100	Molecular genetics, biology, and therapy for inherited peripheral neuropathies. NeuroMolecular Medicine, 2006, 8, 1-2.	3.4	6
101	Unraveling the genetics of distal hereditary motor neuronopathies. NeuroMolecular Medicine, 2006, 8, 131-146.	3.4	57
102	Novel frameshift and splice site mutations in the neurotrophic tyrosine kinase receptor type 1 gene (NTRK1) associated with hereditary sensory neuropathy type IV. Neuromuscular Disorders, 2006, 16, 19-25.	0.6	30
103	G.O.1 In vitro expression of small heat shock protein HSPB8 and HSPB1 mutations causing axonal neuropathy. Neuromuscular Disorders, 2006, 16, 645-646.	0.6	1
104	N.P.3 05 Mitofusin 2 mutations are a major cause for autosomal dominant axonal CMT neuropathy. Neuromuscular Disorders, 2006, 16, 665-666.	0.6	0
105	Recent advances in hereditary sensory and autonomic neuropathies. Current Opinion in Neurology, 2006, 19, 474-480.	3.6	56
106	Spastin gene mutations in Bulgarian patients with hereditary spastic paraplegia. Clinical Genetics, 2006, 70, 490-495.	2.0	6
107	Disrupted function and axonal distribution of mutant tyrosyl-tRNA synthetase in dominant intermediate Charcot-Marie-Tooth neuropathy. Nature Genetics, 2006, 38, 197-202.	21.4	323
108	Genetics of motor neuron disease. Current Neurology and Neuroscience Reports, 2006, 6, 423-431.	4.2	20

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109	Disease mechanisms in hereditary sensory and autonomic neuropathies. Neurobiology of Disease, 2006, 21, 247-255.	4.4	35
110	Axonal neuropathy with optic atrophy is caused by mutations in mitofusin 2. Annals of Neurology, 2006, 59, 276-281.	5.3	380
111	MFN2 mutation distribution and genotype/phenotype correlation in Charcot-Marie-Tooth type 2. Brain, 2006, 129, 2093-2102.	7.6	351
112	A "nerve" ending story in the identification of mutations in Charcot-Marie-Tooth neuropathy. Neurology, 2006, 67, 1114-1115.	1.1	4
113	The CMT1A Duplication. , 2006, , 3-17.		1
114	The CMT1A Duplication and HNPP Deletion. , 2006, , 169-178.		1
115	Mutations in the pleckstrin homology domain of dynamin 2 cause dominant intermediate Charcot-Marie-Tooth disease. Nature Genetics, 2005, 37, 289-294.	21.4	324
116	Mutations in SEPT9 cause hereditary neuralgic amyotrophy. Nature Genetics, 2005, 37, 1044-1046.	21.4	222
117	Experimental Charcotâ€“Marieâ€“Tooth type 1A: A cDNA microarrays analysis. Molecular and Cellular Neurosciences, 2005, 28, 703-714.	2.2	39
118	Synaptopodin and 4 novel genes identified in primary sensory neurons. Molecular and Cellular Neurosciences, 2005, 30, 316-325.	2.2	3
119	Absence of mutations in the prion-protein gene in a large cohort of HMSN patients. Neuromuscular Disorders, 2005, 15, 549-551.	0.6	4
120	Small heat shock proteins in inherited peripheral neuropathies. Annals of Medicine, 2005, 37, 413-422.	3.8	49
121	Autosomal dominant axonal Charcot-Marie-Tooth disease type 2 (CMT2G) maps to chromosome 12q12-q13.3. Journal of Medical Genetics, 2004, 41, 193-197.	3.2	27
122	The phenotype of motor neuropathies associated with BSCL2 mutations is broader than Silver syndrome and distal HMN type V. Brain, 2004, 127, 2124-2130.	7.6	146
123	Clinicopathological and genetic study of early-onset demyelinating neuropathy. Brain, 2004, 127, 2540-2550.	7.6	76
124	Heterozygous missense mutations in BSCL2 are associated with distal hereditary motor neuropathy and Silver syndrome. Nature Genetics, 2004, 36, 271-276.	21.4	349
125	Hot-spot residue in small heat-shock protein 22 causes distal motor neuropathy. Nature Genetics, 2004, 36, 597-601.	21.4	395
126	Mutations in the mitochondrial GTPase mitofusin 2 cause Charcot-Marie-Tooth neuropathy type 2A. Nature Genetics, 2004, 36, 449-451.	21.4	1,391

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127	Mutant small heat-shock protein 27 causes axonal Charcot-Marie-Tooth disease and distal hereditary motor neuropathy. Nature Genetics, 2004, 36, 602-606.	21.4	541
128	SIMPLE mutation in demyelinating neuropathy and distribution in sciatic nerve. Annals of Neurology, 2004, 55, 713-720.	5.3	67
129	Genomic organization and mutation analysis of three candidate genes for hereditary neuralgic amyotrophy. Muscle and Nerve, 2004, 29, 601-604.	2.2	1
130	Molecular genetics of distal hereditary motor neuropathies. Human Molecular Genetics, 2004, 13, R195-R202.	2.9	71
131	DNA/RNA Helicase Gene Mutations in a Form of Juvenile Amyotrophic Lateral Sclerosis (ALS4). American Journal of Human Genetics, 2004, 74, 1128-1135.	6.2	717
132	Anti-steroid takes aim at neuropathy. Nature Medicine, 2003, 9, 1457-1458.	30.7	4
133	Andermann Syndrome in a Turkish Patient. Journal of Child Neurology, 2003, 18, 76-79.	1.4	4
134	Mutations in the Small GTP-ase Late Endosomal Protein RAB7 Cause Charcot-Marie-Tooth Type 2B Neuropathy. American Journal of Human Genetics, 2003, 72, 722-727.	6.2	415
135	Slowed Conduction and Thin Myelination of Peripheral Nerves Associated with Mutant Rho Guanine-Nucleotide Exchange Factor 10. American Journal of Human Genetics, 2003, 73, 926-932.	6.2	107
136	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.	6.2	185
137	Dominant Intermediate Charcot-Marie-Tooth Type C Maps to Chromosome 1p34-p35. American Journal of Human Genetics, 2003, 73, 1423-1430.	6.2	58
138	Identification of novel GDAP1 mutations causing autosomal recessive Charcot-Marie-Tooth disease. Neuromuscular Disorders, 2003, 13, 720-728.	0.6	49
139	Autosomal Dominant Inherited Neuropathies With Prominent Sensory Loss and Mutilations. Archives of Neurology, 2003, 60, 329.	4.5	74
140	Hereditary sensory neuropathy is caused by a mutation in the delta subunit of the cytosolic chaperonin-containing t-complex peptide-1 (Cct4) gene. Human Molecular Genetics, 2003, 12, 1917-1925.	2.9	51
141	Mutations in the neurofilament light chain gene (NEFL) cause early onset severe Charcot-Marie-Tooth disease. Brain, 2003, 126, 590-597.	7.6	259
142	Autosomal dominant juvenile amyotrophic lateral sclerosis and distal hereditary motor neuronopathy with pyramidal tract signs: synonyms for the same disorder?. Brain, 2002, 125, 1320-1325.	7.6	74
143	Exclusion of serine palmitoyltransferase long chain base subunit 2 (SPTLC2) as a common cause for hereditary sensory neuropathy. Neuromuscular Disorders, 2002, 12, 656-658.	0.6	23
144	A novel homozygous missense mutation in the myotubularin-related protein 2 gene associated with recessive Charcotâ€“Marieâ€“Tooth disease with irregularly folded myelin sheaths. Neuromuscular Disorders, 2002, 12, 869-873.	0.6	36

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145	Periaxin mutations cause a broad spectrum of demyelinating neuropathies. Annals of Neurology, 2002, 51, 709-715.	5.3	106
146	Mutation analysis of 12 candidate genes for distal hereditary motor neuropathy type II (distal HMN II) linked to 12q24.3. Journal of the Peripheral Nervous System, 2002, 7, 87-95.	3.1	5
147	Search for mutations in the EGR2 corepressor proteins, NAB1 and NAB2, in human peripheral neuropathies. Neurogenetics, 2002, 4, 37-41.	1.4	9
148	Localization of the Gene for the Intermediate Form of Charcot-Marie-Tooth to Chromosome 10q24.1-q25.1. American Journal of Human Genetics, 2001, 69, 889-894.	6.2	60
149	A novel 3â€²-splice site mutation in peripheral myelin protein 22 causing hereditary neuropathy with liability to pressure palsies. Neuromuscular Disorders, 2001, 11, 400-403.	0.6	27
150	Caspr1/Paranodin/Neurexin IV is most likely not a common disease-causing gene for inherited peripheral neuropathies. NeuroReport, 2001, 12, 2609-2614.	1.2	3
151	Hereditary neuralgic amyotrophy. Neurogenetics, 2001, 3, 115-118.	1.4	11
152	Hereditary Neuralgic Amyotrophy (HNA) is genetically heterogeneous. Journal of Neurology, 2001, 248, 861-865.	3.6	26
153	A novel connexin 32 missense mutation (E208G) causing Charcot-Marie-Tooth disease. Human Mutation, 2001, 17, 157-157.	2.5	0
154	Further evidence that neurofilament light chain gene mutations can cause Charcot-Marie-Tooth disease type 2E. Annals of Neurology, 2001, 49, 245-249.	5.3	188
155	Mutation analysis of 4 candidate genes for hereditary neuralgic amyotrophy (HNA). Human Genetics, 2001, 108, 390-393.	3.8	13
156	A new locus for autosomal dominant Charcot-Marie-Tooth disease type 2 (CMT2F) maps to chromosome 7q11-q21. European Journal of Human Genetics, 2001, 9, 646-650.	2.8	66
157	Screening for mutations in the peripheral myelin genesPMP22,MPZ andCx32 (GJB1) in Russian Charcot-Marie-Tooth neuropathy patients. Human Mutation, 2000, 15, 340-347.	2.5	78
158	Of giant axons and curly hair. Nature Genetics, 2000, 26, 254-255.	21.4	11
159	Charcotâ€Marieâ€Tooth Neuropathy Type 2 and P0 Point Mutations: Two Novel Amino Acid Substitutions (Asp61Gly; Tyr119Cys) and a Possible â€œHotspotâ€•on Thr124Met. Brain Pathology, 2000, 10, 235-248.	4.1	74
160	Fine mapping of the neurally expressed gene SOX14 to human 3q23, relative to three congenital diseases. Human Genetics, 2000, 106, 432-439.	3.8	19
161	PMP22 Thr118Met is not a clinically relevant CMT1 marker. Journal of Neurology, 2000, 247, 696-700.	3.6	22
162	A Clone Contig of 12q24.3 Encompassing the Distal Hereditary Motor Neuropathy Type II Gene. Genomics, 2000, 65, 34-43.	2.9	11

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163	Diagnostic guidelines for hereditary neuralgic amyotrophy or heredofamilial neuritis with brachial plexus predilection. Neuromuscular Disorders, 2000, 10, 515-517.	0.6	30
164	71st ENMC International Workshop, 6th Workshop of the European Charcotâ€“Marieâ€“Tooth Disease Consortium: Hereditary recurrent focal neuropathies, 24â€“25 September 1999, Soestduinen, The Netherlands. Neuromuscular Disorders, 2000, 10, 518-524.	0.6	2
165	Construction of a PAC Contig within the Distal Hereditary Motor Neuropathy Type II Candidate Region at 12q24. Annals of the New York Academy of Sciences, 1999, 883, 463-465.	3.8	0
166	Genetic refinement of the hereditary neuralgic amyotrophy (HNA) locus at chromosome 17q25. European Journal of Human Genetics, 1999, 7, 920-927.	2.8	34
167	Molecular genetics and biology of inherited peripheral neuropathies: a fast-moving field. Neurogenetics, 1999, 2, 137-148.	1.4	34
168	A Sequence-Ready BAC/PAC Contig and Partial Transcript Map of Approximately 1.5 Mb in Human Chromosome 17q25 Comprising Multiple Disease Genes. Genomics, 1999, 62, 242-250.	2.9	15
169	A Novel Type of Hereditary Motor and Sensory Neuropathy Characterized by a Mild Phenotype. Archives of Neurology, 1999, 56, 1283.	4.5	16
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