Garth A Nicholson

List of Publications by Year
in descending order

191 papers	18,345 citations	19608 61 h-index	14156 128 g-index
202 all docs	202 docs citations	202 times ranked	17942 citing authors

#	Article	IF	Citations
1	Polygenic risk score analysis for amyotrophic lateral sclerosis leveraging cognitive performance, educational attainment and schizophrenia. European Journal of Human Genetics, 2022, 30, 532-539.	1.4	16
2	Clinical practice guideline for the management of paediatric Charcot-Marie-Tooth disease. Journal of Neurology, Neurosurgery and Psychiatry, 2022, 93, 530-538.	0.9	10
3	Genome-wide study of DNA methylation shows alterations in metabolic, inflammatory, and cholesterol pathways in ALS. Science Translational Medicine, 2022, 14, eabj0264.	5.8	38
4	Long read sequencing overcomes challenges in the diagnosis of <scp>SORD</scp> neuropathy. Journal of the Peripheral Nervous System, 2022, 27, 120-126.	1.4	6
5	Pathogenic Huntingtin Repeat Expansions in Patients with Frontotemporal Dementia and Amyotrophic Lateral Sclerosis. Neuron, 2021, 109, 448-460.e4.	3.8	56
6	Meta-analysis of genome-wide DNA methylation identifies shared associations across neurodegenerative disorders. Genome Biology, 2021, 22, 90.	3.8	49
7	Genetic analysis of GLT8D1 and ARPP21 in Australian familial and sporadic amyotrophic lateral sclerosis. Neurobiology of Aging, 2021, 101, 297.e9-297.e11.	1.5	6
8	Revisiting the pathogenic mechanism of the GJB1 5â€™ UTR c.-103Câ€‰>â€‰T mutation causing CMTX1. Neurogenetics, 2021, 22, 149-160.	0.7	1
9	113â€…Clinical and neurophysiological improvement in Hereditary sensory and autonomic neuropathy type I (HSAN-1) following high dose serine therapy. , 2021, , .		0
10	Sodium valproate increases activity of the sirtuin pathway resulting in beneficial effects for spinocerebellar ataxia-3 in vivo. Molecular Brain, 2021, 14, 128.	1.3	12
11	Association of Variants in the SPTLC1 Gene With Juvenile Amyotrophic Lateral Sclerosis. JAMA Neurology, 2021, 78, 1236.	4.5	46
12	Charcotâ€“Marieâ€“tooth disease causing mutation (p.R158H) in pyruvate dehydrogenase kinase 3 (PDK3) affects synaptic transmission, ATP production and causes neurodegeneration in a CMTX6 C. elegans model. Human Molecular Genetics, 2021, , .	1.4	5
13	Common and rare variant association analyses in amyotrophic lateral sclerosis identify 15 risk loci with distinct genetic architectures and neuron-specific biology. Nature Genetics, 2021, 53, 1636-1648.	9.4	223
14	Genome-wide Meta-analysis Finds the ACSL5-ZDHHC6 Locus Is Associated with ALS and Links Weight Loss to the Disease Genetics. Cell Reports, 2020, 33, 108323.	2.9	41
15	A Simple Differentiation Protocol for Generation of Induced Pluripotent Stem Cell-Derived Basal Forebrain-Like Cholinergic Neurons for Alzheimerâ€™s Disease and Frontotemporal Dementia Disease Modeling. Cells, 2020, 9, 2018.	1.8	27
16	RFC1 expansions can mimic hereditary sensory neuropathy with cough and SjÃ¶gren syndrome. Brain, 2020, 143, e82-e82.	3.7	25
17	Identity by descent analysis identifies founder events and links SOD1 familial and sporadic ALS cases. Npj Genomic Medicine, 2020, 5, 32.	1.7	20
18	Impaired NHEJ repair in amyotrophic lateral sclerosis is associated with TDP-43 mutations. Molecular Neurodegeneration, 2020, 15, 51.	4.4	54

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19	The genetic landscape of axonal neuropathies in the middle-aged and elderly. Neurology, 2020, 95, e3163-e3179.	1.5	19
20	CYLD is a causative gene for frontotemporal dementia â€“ amyotrophic lateral sclerosis. Brain, 2020, 143, 783-799.	3.7	62
21	Significant out-of-sample classification from methylation profile scoring for amyotrophic lateral sclerosis. Npj Genomic Medicine, 2020, 5, 10.	1.7	25
22	Genetic and immunopathological analysis of CHCHD10 in Australian amyotrophic lateral sclerosis and frontotemporal dementia and transgenic TDP-43 mice. Journal of Neurology, Neurosurgery and Psychiatry, 2020, 91, 162-171.	0.9	8
23	Modelling the pathogenesis of X-linked distal hereditary motor neuropathy using patient-derived iPSCs. DMM Disease Models and Mechanisms, 2020, 13, .	1.2	7
24	Genome-wide association study identifies genetic factors that modify age at onset in Machado-Joseph disease. Aging, 2020, 12, 4742-4756.	1.4	10
25	Inherited Neuropathies. Seminars in Neurology, 2019, 39, 620-639.	0.5	8
26	Generation and characterization of a human induced pluripotent stem cell line UOWi005-A from dermal fibroblasts derived from a CCNF familial amyotrophic lateral sclerosis patient using mRNA reprogramming. Stem Cell Research, 2019, 40, 101530.	0.3	6
27	Monozygotic twins and triplets discordant for amyotrophic lateral sclerosis display differential methylation and gene expression. Scientific Reports, 2019, 9, 8254.	1.6	36
28	Linkage analysis and whole exome sequencing reveals AHNAK2 as a novel genetic cause for autosomal recessive CMT in a Malaysian family. Neurogenetics, 2019, 20, 117-127.	0.7	12
29	Hereditary sensory and autonomic neuropathy type IC accompanied by upper motor neuron abnormalities and type II juxtafoveal retinal telangiectasias. Journal of the Peripheral Nervous System, 2019, 24, 224-229.	1.4	5
30	A de novo EGR2 variant, c.1232Aâ€‰>â€‰G p.Asp411Gly, causes severe early-onset Charcot-Marie-Tooth Neuropathy Type 3 (Dejerine-Sottas Neuropathy). Scientific Reports, 2019, 9, 19336.	1.6	4
31	Motor Neuron Abnormalities Correlate with Impaired Movement in Zebrafish that Express Mutant Superoxide Dismutase 1. Zebrafish, 2019, 16, 8-14.	0.5	16
32	Body composition and its association with physical performance, quality of life, and clinical indicators in Charcot-Marie-Tooth disease: a pilot study. Disability and Rehabilitation, 2019, 41, 405-412.	0.9	5
33	Structural variations causing inherited peripheral neuropathies: A paradigm for understanding genomic organization, chromatin interactions, and gene dysregulation. Molecular Genetics & Genomic Medicine, 2018, 6, 422-433.	0.6	17
34	Unique clinical and neurophysiologic profile of a cohort of children with CMTX3. Neurology, 2018, 90, e1706-e1710.	1.5	3
35	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. Neuron, 2018, 97, 1268-1283.e6.	3.8	517
36	Infantile-Onset Myelin Protein Zeroâ€“Related Demyelinating Neuropathy Presenting as an Upper Extremity Monoplegia. Seminars in Pediatric Neurology, 2018, 26, 52-55.	1.0	3

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37	Sarcolemmal excitability in the myotonic dystrophies. Muscle and Nerve, 2018, 57, 595-602.	1.0	12
38	A novel Parkinson's disease risk variant, p. W378R, in the Gaucher's disease GBA gene. Movement Disorders, 2018, 33, 1662-1664.	2.2	1
39	Neuronal cell culture from transgenic zebrafish models of neurodegenerative disease. Biology Open, 2018, 7, .	0.6	8
40	OUP accepted manuscript. Brain, 2018, 141, e66.	3.7	7
41	Quantitative muscle ultrasound as a biomarker in Charcot-Marie-Tooth neuropathy. Clinical Neurophysiology, 2017, 128, 227-232.	0.7	25
42	Non-nuclear Pool of Splicing Factor SFPQ Regulates Axonal Transcripts Required for Normal Motor Development. Neuron, 2017, 94, 322-336.e5.	3.8	61
43	Hypomorphic mutations in POLR3A are a frequent cause of sporadic and recessive spastic ataxia. Brain, 2017, 140, 1561-1578.	3.7	85
44	Expression of ALS/FTD-linked mutant CCNF in zebrafish leads to increased cell death in the spinal cord and an aberrant motor phenotype. Human Molecular Genetics, 2017, 26, 2616-2626.	1.4	44
45	A recurrent WARS mutation is a novel cause of autosomal dominant distal hereditary motor neuropathy. Brain, 2017, 140, 1252-1266.	3.7	75
46	Genetic and Pathological Assessment of hnRNPA1, hnRNPA2/B1, and hnRNPA3 in Familial and Sporadic Amyotrophic Lateral Sclerosis. Neurodegenerative Diseases, 2017, 17, 304-312.	0.8	27
47	Calpain Inhibition Is Protective in Machadoâ€“Joseph Disease Zebrafish Due to Induction of Autophagy. Journal of Neuroscience, 2017, 37, 7782-7794.	1.7	57
48	A novel amyotrophic lateral sclerosis mutation in OPTN induces ER stress and Golgi fragmentation in vitro. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2017, 18, 126-133.	1.1	24
49	A Tol2 Gateway-Compatible Toolbox for the Study of the Nervous System and Neurodegenerative Disease. Zebrafish, 2017, 14, 69-72.	0.5	56
50	Pathogenic mechanisms underlying X-linked Charcot-Marie-Tooth neuropathy (CMTX6) in patients with a pyruvate dehydrogenase kinase 3 mutation. Neurobiology of Disease, 2016, 94, 237-244.	2.1	12
51	Mutation analysis of genes within the dynactin complex in a cohort of hereditary peripheral neuropathies. Clinical Genetics, 2016, 90, 127-133.	1.0	7
52	<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
53	Motor cortical dysfunction develops in spinocerebellar ataxia type 3. Clinical Neurophysiology, 2016, 127, 3418-3424.	0.7	22
54	Relationship between physical performance and quality of life in Charcotâ€Marieâ€Tooth disease: a pilot study. Journal of the Peripheral Nervous System, 2016, 21, 357-364.	1.4	13

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55	Novel motor phenotypes in patients with VRK1 mutations without pontocerebellar hypoplasia. Neurology, 2016, 87, 65-70.	1.5	38
56	A 1.35ÂMb DNA fragment is inserted into the DHMN1 locus on chromosome 7q34â€“q36.2. Human Genetics, 2016, 135, 1269-1278.	1.8	9
57	Genome-wide association analyses identify new risk variants and the genetic architecture of amyotrophic lateral sclerosis. Nature Genetics, 2016, 48, 1043-1048.	9.4	494
58	NEK1 variants confer susceptibility to amyotrophic lateral sclerosis. Nature Genetics, 2016, 48, 1037-1042.	9.4	218
59	CCNF mutations in amyotrophic lateral sclerosis and frontotemporal dementia. Nature Communications, 2016, 7, 11253.	5.8	174
60	Genetic basis of hindlimb loss in a naturally occurring vertebrate model. Biology Open, 2016, 5, 359-366.	0.6	24
61	Characterizing the molecular phenotype of an Atp7a^{T985I}conditional knock in mouse model for X-linked distal hereditary motor neuropathy (dHMNX). Metallomics, 2016, 8, 981-992.	1.0	9
62	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
63	Improved inherited peripheral neuropathy genetic diagnosis by wholeâ€exome sequencing. Molecular Genetics & Genomic Medicine, 2015, 3, 143-154.	0.6	59
64	Axonal Ion Channel Dysfunction inC9orf72Familial Amyotrophic Lateral Sclerosis. JAMA Neurology, 2015, 72, 49.	4.5	35
65	Evaluation of Skin Fibroblasts from Amyotrophic Lateral Sclerosis Patients for the Rapid Study of Pathological Features. Neurotoxicity Research, 2015, 28, 138-146.	1.3	30
66	Homozygous mutations inMFN2cause multiple symmetric lipomatosis associated with neuropathy. Human Molecular Genetics, 2015, 24, 5109-5114.	1.4	61
67	Alteration of ornithine metabolism leads to dominant and recessive hereditary spastic paraplegia. Brain, 2015, 138, 2191-2205.	3.7	88
68	Variants associated with Gaucher disease in multiple system atrophy. Annals of Clinical and Translational Neurology, 2015, 2, 417-426.	1.7	90
69	Mutation analysis of MATR3 in Australian familial amyotrophic lateral sclerosis. Neurobiology of Aging, 2015, 36, 1602.e1-1602.e2.	1.5	13
70	Use of Whole-Exome Sequencing for Diagnosis of Limb-Girdle Muscular Dystrophy. JAMA Neurology, 2015, 72, 1424.	4.5	164
71	Novel TBK1 truncating mutation in a familial amyotrophic lateral sclerosis patient of Chinese origin. Neurobiology of Aging, 2015, 36, 3334.e1-3334.e5.	1.5	35
72	Cortical Function in Asymptomatic Carriers and Patients WithC9orf72Amyotrophic Lateral Sclerosis. JAMA Neurology, 2015, 72, 1268.	4.5	74

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73	Mutations in theSPTLC1Protein Cause Mitochondrial Structural Abnormalities and Endoplasmic Reticulum Stress in Lymphoblasts. DNA and Cell Biology, 2014, 33, 399-407.	0.9	24
74	Analysis of dynein intermediate chains, light intermediate chains and light chains in a cohort of hereditary peripheral neuropathies. Neurogenetics, 2014, 15, 229-235.	0.7	3
75	Exome-wide Rare Variant Analysis Identifies TUBA4A Mutations Associated with Familial ALS. Neuron, 2014, 84, 324-331.	3.8	308
76	Axonal excitability in X-linked dominant Charcot Marie Tooth disease. Clinical Neurophysiology, 2014, 125, 1261-1269.	0.7	12
77	Mutant Human FUS Is Ubiquitously Mislocalized and Generates Persistent Stress Granules in Primary Cultured Transgenic Zebrafish Cells. PLoS ONE, 2014, 9, e90572.	1.1	19
78	ERBB4 Mutations that Disrupt the Neuregulin-ErbB4 Pathway Cause Amyotrophic Lateral Sclerosis Type 19. American Journal of Human Genetics, 2013, 93, 900-905.	2.6	123
79	Controversies and priorities in amyotrophic lateral sclerosis. Lancet Neurology, The, 2013, 12, 310-322.	4.9	454
80	Hereditary Spastic Paraplegia Type 43 (SPG43) is Caused by Mutation inC19orf12. Human Mutation, 2013, 34, 1357-1360.	1.1	79
81	A Loss-of-Function Variant in the Human Histidyl-tRNA Synthetase (HARS) Gene is Neurotoxic In Vivo. Human Mutation, 2013, 34, 191-199.	1.1	104
82	Pathophysiological insights into ALS with C9ORF72 expansions. Journal of Neurology, Neurosurgery and Psychiatry, 2013, 84, 931-935.	0.9	89
83	Mutation analysis and immunopathological studies of PFN1 in familial and sporadic amyotrophic lateral sclerosis. Neurobiology of Aging, 2013, 34, 2235.e7-2235.e10.	1.5	16
84	Apparent anticipation in SOD1 familial amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2013, 14, 452-456.	1.1	2
85	DNMT1 mutation hot spot causes varied phenotypes of HSAN1 with dementia and hearing loss. Neurology, 2013, 80, 824-828.	1.5	60
86	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
87	Reâ€analysis of an original <scp>CMTX3</scp> family using exome sequencing identifies a known <scp>BSCL2</scp> mutation. Muscle and Nerve, 2013, 47, 922-924.	1.0	10
88	The MFN2 V705I Variant Is Not a Disease-Causing Mutation: A Segregation Analysis in a CMT2 Family. Journal of Neurodegenerative Diseases, 2013, 2013, 1-5.	1.1	3
89	Exome sequencing to identify de novo mutations in sporadic ALS trios. Nature Neuroscience, 2013, 16, 851-855.	7.1	129
90	A novelTARDBPinsertion/deletion mutation in the flail arm variant of amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2012, 13, 465-470.	2.3	12

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91	Evaluating the role of the FUS/TLS-related gene EWSR1 in amyotrophic lateral sclerosis. Human Molecular Genetics, 2012, 21, 2899-2911.	1.4	246
92	Mutational Origin of Machado-Joseph Disease in the Australian Aboriginal Communities of Groote Eylandt and Yirrkala. Archives of Neurology, 2012, 69, 746-51.	4.9	25
93	Mutation analysis of the optineurin gene in familial amyotrophic lateral sclerosis. Neurobiology of Aging, 2012, 33, 210.e9-210.e10.	1.5	13
94	Mutation analysis of VCP in familial and sporadic amyotrophic lateral sclerosis. Neurobiology of Aging, 2012, 33, 1488.e15-1488.e16.	1.5	17
95	UBQLN2/ubiquilin 2 mutation and pathology in familial amyotrophic lateral sclerosis. Neurobiology of Aging, 2012, 33, 2527.e3-2527.e10.	1.5	114
96	A family with 2 X-linked disorders: Charcot-Marie-Tooth disease and hemophilia A. Muscle and Nerve, 2012, 46, 454-455.	1.0	1
97	The TRK-Fused Gene Is Mutated in Hereditary Motor and Sensory Neuropathy with Proximal Dominant Involvement. American Journal of Human Genetics, 2012, 91, 320-329.	2.6	98
98	A Recurrent loss-of-function alanyl-tRNA synthetase (AARSâ€‰) mutation in patients with charcot-marie-tooth disease type 2N (CMT2N). Human Mutation, 2012, 33, 244-253.	1.1	90
99	â€œDancing feet dyskinesiasâ€• A clue to parkin gene mutations. Movement Disorders, 2012, 27, 587-588.	2.2	6
100	Distinctive genetic and clinical features of CMT4J: a severe neuropathy caused by mutations in the PI(3,5)P2 phosphatase FIG4. Brain, 2011, 134, 1959-1971.	3.7	107
101	Mutations in DNMT1 cause hereditary sensory neuropathy with dementia and hearing loss. Nature Genetics, 2011, 43, 595-600.	9.4	342
102	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.	2.6	172
103	A yeast functional screen predicts new candidate ALS disease genes. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 20881-20890.	3.3	365
104	Missense Mutations in the Copper Transporter Gene ATP7A Cause X-Linked Distal Hereditary Motor Neuropathy. American Journal of Human Genetics, 2010, 86, 343-352.	2.6	170
105	Compound Heterozygosity for Loss-of-Function Lysyl-tRNA Synthetase Mutations in a Patient with Peripheral Neuropathy. American Journal of Human Genetics, 2010, 87, 560-566.	2.6	169
106	X-linked CMT: genes and gene loci in an Australian cohort. Neurogenetics, 2010, 11, 267-269.	0.7	4
107	Biomarkers of disease in a case of familial lower motor neuron ALS. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2010, 11, 486-489.	2.3	10
108	Hereditary Sensory Neuropathy Type 1 Is Caused by the Accumulation of Two Neurotoxic Sphingolipids. Journal of Biological Chemistry, 2010, 285, 11178-11187.	1.6	320

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109	FUS mutations in amyotrophic lateral sclerosis: clinical, pathological, neurophysiological and genetic analysis. Journal of Neurology, Neurosurgery and Psychiatry, 2010, 81, 639-645.	0.9	205
110	Correlation between muscle atrophy on MRI and manual strength testing in hereditary neuropathies. Journal of Clinical Neuroscience, 2010, 17, 874-878.	0.8	21
111	Fused in sarcoma/translocated in liposarcoma: A multifunctional DNA/RNA binding protein. International Journal of Biochemistry and Cell Biology, 2010, 42, 1408-1411.	1.2	30
112	TDP-43: A DNA and RNA binding protein with roles in neurodegenerative diseases. International Journal of Biochemistry and Cell Biology, 2010, 42, 1606-1609.	1.2	53
113	Expanding the clinical, pathological and MRI phenotype of DNM2-related centronuclear myopathy. Neuromuscular Disorders, 2010, 20, 229-237.	0.3	100
114	Development of a Multiplex Ligation-Dependent Probe Amplification Assay for Diagnosis and Estimation of the Frequency of Spinocerebellar Ataxia Type 15. Clinical Chemistry, 2009, 55, 1415-1418.	1.5	39
115	Phenotypic spectrum of dynamin 2 mutations in Charcot-Marie-Tooth neuropathy. Brain, 2009, 132, 1741-1752.	3.7	134
116	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.	0.7	26
117	Genotypes & Sensory Phenotypes in 2 New X-Linked Neuropathies (CMTX3 and dSMAX) and Dominant CMT/HMN Overlap Syndromes. Advances in Experimental Medicine and Biology, 2009, 652, 201-206.	0.8	1
118	Mutations in FUS, an RNA Processing Protein, Cause Familial Amyotrophic Lateral Sclerosis Type 6. Science, 2009, 323, 1208-1211.	6.0	2,295
119	Evidence of a founder haplotype refines the X-linked Charcotâ€“Marie-Tooth (CMTX3) locus to a 2.5 Mb region. Neurogenetics, 2008, 9, 191-5.	0.7	6
120	Replication of association between ELAVL4 and Parkinson disease: the GenePD study. Human Genetics, 2008, 124, 95-99.	1.8	34
121	Huntington CAG repeat size does not modify onset age in familial Parkinson's disease: The GenePD study. Movement Disorders, 2008, 23, 1596-1601.	2.2	8
122	Pedigree with frontotemporal lobar degeneration â€“ motor neuron disease and Tar DNA binding protein-43 positive neuropathology: genetic linkage to chromosome 9. BMC Neurology, 2008, 8, 32.	0.8	71
123	The Gly2019Ser mutation in LRRK2is not fully penetrant in familial Parkinson's disease: the GenePD study. BMC Medicine, 2008, 6, 32.	2.3	102
124	TDP-43 Mutations in Familial and Sporadic Amyotrophic Lateral Sclerosis. Science, 2008, 319, 1668-1672.	6.0	2,268
125	Association study on glutathione Sâ€transferase omega 1 and 2 and familial ALS. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2008, 9, 81-84.	2.3	19
126	Histopathological Findings in Hereditary Motor and Sensory Neuropathy of Axonal Type With Onset in Early Childhood Associated WithMitofusin 2Mutations. Journal of Neuropathology and Experimental Neurology, 2008, 67, 1097-1102.	0.9	81

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127	Cortical hyperexcitability may precede the onset of familial amyotrophic lateral sclerosis. Brain, 2008, 131, 1540-1550.	3.7	391
128	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.	1.5	53
129	Hereditary Spastic Paraplegia 3A Associated With Axonal Neuropathy. Archives of Neurology, 2007, 64, 706.	4.9	42
130	Asian Origin for the Worldwide-Spread Mutational Event in Machado-Joseph Disease. Archives of Neurology, 2007, 64, 1502.	4.9	65
131	Stoichiometric Alteration of PMP22 Protein Determines the Phenotype of Hereditary Neuropathy With Liability to Pressure Palsies. Archives of Neurology, 2007, 64, 974.	4.9	35
132	Establishment of the Australasian paediatric Charcot-Marie-Tooth disease registry. Neuromuscular Disorders, 2007, 17, 349-350.	0.3	9
133	Peripheral Nerve Demyelination Caused by a Mutant Rho GTPase Guanine Nucleotide Exchange Factor, Frabin/FGD4. American Journal of Human Genetics, 2007, 81, 158-164.	2.6	128
134	Further evidence for genetic heterogeneity of distal HMN type V, CMT2 with predominant hand involvement and Silver syndrome. Journal of the Neurological Sciences, 2007, 263, 100-106.	0.3	64
135	A novel locus for distal motor neuron degeneration maps to chromosome 7q34-q36. Human Genetics, 2007, 121, 559-564.	1.8	23
136	Intermediate forms of Charcot-Marie-Tooth neuropathy. NeuroMolecular Medicine, 2006, 8, 123-130.	1.8	80
137	Night splinting does not increase ankle range of motion in people with Charcot-Marie-Tooth disease: A randomised, cross-over trial. Australian Journal of Physiotherapy, 2006, 52, 193-199.	0.9	49
138	Late-onset hereditary sensory neuropathy type I due to SPTLC1 mutation: Autopsy findings. Clinical Neurology and Neurosurgery, 2006, 108, 780-783.	0.6	16
139	The dominantly inherited motor and sensory neuropathies: Clinical and molecular advances. Muscle and Nerve, 2006, 33, 589-597.	1.0	26
140	Influence of Heterozygosity for Parkin Mutation on Onset Age in Familial Parkinson Disease. Archives of Neurology, 2006, 63, 826.	4.9	147
141	Hereditary Sensory Neuropathy. , 2006, , 329-335.		0
142	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
143	Autosomal dominant hereditary sensory neuropathy with chronic cough and gastro-oesophageal reflux: clinical features in two families linked to chromosome 3p22â€“p24. Brain, 2005, 128, 2797-2810.	3.7	70
144	Phenotypic spectrum of disorders associated with glycyl-tRNA synthetase mutations. Brain, 2005, 128, 2304-2314.	3.7	124

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145	AGE DEPENDENT PENETRANCE OF THREE DIFFERENT SUPEROXIDE DISMUTASE 1 (SOD 1) MUTATIONS. International Journal of Neuroscience, 2005, 115, 1119-1130.	0.8	12
146	Equilibrium Between Cell Division and Apoptosis in Immortal Cells as an Alternative to the G1 Restriction Mechanism in Mammalian Cells. Cell Cycle, 2004, 3, 489-493.	1.3	1
147	Transcript map of the candidate region for HSNI with cough and gastroesophageal reflux on chromosome 3p and exclusion of candidate genes. Neurogenetics, 2004, 5, 197-200.	0.7	2
148	Tau haplotypes regulate transcription and are associated with Parkinson's disease. Annals of Neurology, 2004, 55, 329-334.	2.8	157
149	Hereditary sensory neuropathy type 1 in a Portuguese familyâ€"electrodiagnostic and autonomic nervous system studies. Journal of the Neurological Sciences, 2004, 227, 35-38.	0.3	10
150	Activity of partially inhibited serine palmitoyltransferase is sufficient for normal sphingolipid metabolism and viability of HSN1 patient cells. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2004, 1688, 168-175.	1.8	35
151	DNA/RNA Helicase Gene Mutations in a Form of Juvenile Amyotrophic Lateral Sclerosis (ALS4). American Journal of Human Genetics, 2004, 74, 1128-1135.	2.6	717
152	Hypoxia Causes Aggregation of Serine Palmitoyltransferase followed by Non-Apoptotic Death of Human Lymphocytes. Cell Cycle, 2004, 3, 1271-1277.	1.3	7
153	Refined localization of dominant intermediate Charcot-Marie-Tooth neuropathy and exclusion of seven known candidate genes in the region. Neurogenetics, 2003, 4, 179-183.	0.7	9
154	Mutation screening of the N-myc downstream-regulated gene 1 (NDRG1) in patients with Charcot-Marie-Tooth Disease. Human Mutation, 2003, 22, 129-135.	1.1	61
155	A Locus for Hereditary Sensory Neuropathy with Cough and Gastroesophageal Reflux on Chromosome 3p22-p24. American Journal of Human Genetics, 2003, 73, 632-637.	2.6	69
156	Spinocerebellar ataxia type 15 (sca15) maps to 3p24.2-3pter:. Neurobiology of Disease, 2003, 13, 147-157.	2.1	70
157	A Rapid and Definitive Test for Charcot-Marie-Tooth 1A and Hereditary Neuropathy with Liability to Pressure Palsies Using Multiplexed Real-Time PCR. Genetic Testing and Molecular Biomarkers, 2003, 7, 135-138.	1.7	19
158	Autosomal Dominant Inherited Neuropathies With Prominent Sensory Loss and Mutilations. Archives of Neurology, 2003, 60, 329.	4.9	74
159	Peripheral neuropathies of infancy. Developmental Medicine and Child Neurology, 2003, 45, 408-14.	1.1	13
160	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.	3.7	74
161	GDAP1 mutations in CMT4: Axonal and demyelinating phenotypes?. Neurology, 2002, 59, 1835-1836.	1.5	9
162	Exclusion of serine palmitoyltransferase long chain base subunit 2 (SPTLC2) as a common cause for hereditary sensory neuropathy. Neuromuscular Disorders, 2002, 12, 656-658.	0.3	23

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163	Friedreich's ataxia with chorea and myoclonus caused by a compound heterozygosity for a novel deletion and the trinucleotide GAA expansion. Movement Disorders, 2002, 17, 585-589.	2.2	28
164	Dominant Intermediate Charcot-Marie-Tooth Neuropathy Maps to Chromosome 19p12-p13.2. American Journal of Human Genetics, 2001, 69, 883-888.	2.6	42
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