Garth A Nicholson

List of Publications by Year
in descending order

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191 papers	18,345 citations	18465 62 h-index	14197 128 g-index
202 all docs	202 docs citations	202 times ranked	17942 citing authors

#	Article	IF	Citations
1	Mutations in FUS, an RNA Processing Protein, Cause Familial Amyotrophic Lateral Sclerosis Type 6. Science, 2009, 323, 1208-1211.	6.0	2,295
2	TDP-43 Mutations in Familial and Sporadic Amyotrophic Lateral Sclerosis. Science, 2008, 319, 1668-1672.	6.0	2,268
3	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
4	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. Neuron, 2018, 97, 1268-1283.e6.	3.8	517
5	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
6	Controversies and priorities in amyotrophic lateral sclerosis. Lancet Neurology, The, 2013, 12, 310-322.	4.9	454
7	Mutations in SPTLC1, encoding serine palmitoyltransferase, long chain base subunit-1, cause hereditary sensory neuropathy type I. Nature Genetics, 2001, 27, 309-312.	9.4	402
8	Cortical hyperexcitability may precede the onset of familial amyotrophic lateral sclerosis. Brain, 2008, 131, 1540-1550.	3.7	391
9	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
10	Clinical and pathological features of a parkinsonian syndrome in a family with an Ala53Thr ?-synuclein mutation. Annals of Neurology, 2001, 49, 313-319.	2.8	364
11	Mutations in DNMT1 cause hereditary sensory neuropathy with dementia and hearing loss. Nature Genetics, 2011, 43, 595-600.	9.4	342
12	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
13	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
14	Exome-wide Rare Variant Analysis Identifies TUBA4A Mutations Associated with Familial ALS. Neuron, 2014, 84, 324-331.	3.8	308
15	A frame shift mutation in the PMP22 gene in hereditary neuropathy with liability to pressure palsies. Nature Genetics, 1994, 6, 263-266.	9.4	264
16	Evaluating the role of the FUS/TLS-related gene EWSR1 in amyotrophic lateral sclerosis. Human Molecular Genetics, 2012, 21, 2899-2911.	1.4	246
17	De novo mutation of the myelin Po gene in Dejerineâ€“Sottas disease (hereditary motor and sensory) Tj ETQq1 1 0.784314 rgBT /Ove	9.4	227
18	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

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19	NEK1 variants confer susceptibility to amyotrophic lateral sclerosis. Nature Genetics, 2016, 48, 1037-1042.	9.4	218
20	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
21	CCNF mutations in amyotrophic lateral sclerosis and frontotemporal dementia. Nature Communications, 2016, 7, 11253.	5.8	174
22	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
23	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
24	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
25	Two novel (M233T and Ï278T) presenilin-1 mutations in early-onset AlzheimerÊ¼s disease pedigrees and preliminary evidence for association of presenilin-1 mutations with a novel phenotype. NeuroReport, 1997, 8, 1537-1542.	0.6	165
26	Use of Whole-Exome Sequencing for Diagnosis of Limb-Girdle Muscular Dystrophy. JAMA Neurology, 2015, 72, 1424.	4.5	164
27	Tau haplotypes regulate transcription and are associated with Parkinson's disease. Annals of Neurology, 2004, 55, 329-334.	2.8	157
28	Influence of Heterozygosity for Parkin Mutation on Onset Age in Familial Parkinson Disease. Archives of Neurology, 2006, 63, 826.	4.9	147
29	Phenotypic spectrum of dynamin 2 mutations in Charcot-Marie-Tooth neuropathy. Brain, 2009, 132, 1741-1752.	3.7	134
30	The gene for hereditary sensory neuropathy type I (HSNâ€“I) maps to chromosome 9q22.1â€“q22.3. Nature Genetics, 1996, 13, 101-104.	9.4	130
31	Exome sequencing to identify de novo mutations in sporadic ALS trios. Nature Neuroscience, 2013, 16, 851-855.	7.1	129
32	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
33	Clinical and genetic study of Friedreich ataxia in an Australian population. American Journal of Medical Genetics Part A, 1999, 87, 168-174.	2.4	125
34	Phenotypic spectrum of disorders associated with glycyl-tRNA synthetase mutations. Brain, 2005, 128, 2304-2314.	3.7	124
35	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
36	UBQLN2/ubiquilin 2 mutation and pathology in familial amyotrophic lateral sclerosis. Neurobiology of Aging, 2012, 33, 2527.e3-2527.e10.	1.5	114

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37	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
38	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
39	The Gly2019Ser mutation in LRRK2is not fully penetrant in familial Parkinson's disease: the GenePD study. BMC Medicine, 2008, 6, 32.	2.3	102
40	Expanding the clinical, pathological and MRI phenotype of DNM2-related centronuclear myopathy. Neuromuscular Disorders, 2010, 20, 229-237.	0.3	100
41	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
42	Variable phenotype of Alzheimer's disease with spastic paraparesis. Annals of Neurology, 2001, 49, 125-129.	2.8	90
43	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
44	Variants associated with Gaucher disease in multiple system atrophy. Annals of Clinical and Translational Neurology, 2015, 2, 417-426.	1.7	90
45	Frequency of spinocerebellar ataxia types 1, 2, 3, 6, and 7 in Australian patients with spinocerebellar ataxia. American Journal of Medical Genetics Part A, 2000, 95, 351-358.	2.4	89
46	Pathophysiological insights into ALS with C9ORF72 expansions. Journal of Neurology, Neurosurgery and Psychiatry, 2013, 84, 931-935.	0.9	89
47	Alteration of ornithine metabolism leads to dominant and recessive hereditary spastic paraplegia. Brain, 2015, 138, 2191-2205.	3.7	88
48	Hypomorphic mutations in POLR3A are a frequent cause of sporadic and recessive spastic ataxia. Brain, 2017, 140, 1561-1578.	3.7	85
49	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
50	Intermediate forms of Charcot-Marie-Tooth neuropathy. NeuroMolecular Medicine, 2006, 8, 123-130.	1.8	80
51	Hereditary Spastic Paraplegia Type 43 (SPG43) is Caused by Mutation inC19orf12. Human Mutation, 2013, 34, 1357-1360.	1.1	79
52	Evidence for an X-linked genetic component in familial typical migraine. Human Molecular Genetics, 1998, 7, 459-463.	1.4	77
53	A recurrent WARS mutation is a novel cause of autosomal dominant distal hereditary motor neuropathy. Brain, 2017, 140, 1252-1266.	3.7	75
54	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

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55	Autosomal Dominant Inherited Neuropathies With Prominent Sensory Loss and Mutilations. Archives of Neurology, 2003, 60, 329.	4.9	74
56	Cortical Function in Asymptomatic Carriers and Patients WithC9orf72Amyotrophic Lateral Sclerosis. JAMA Neurology, 2015, 72, 1268.	4.5	74
57	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
58	Spinocerebellar ataxia type 15 (sca15) maps to 3p24.2-3pter:. Neurobiology of Disease, 2003, 13, 147-157.	2.1	70
59	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
60	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
61	Asian Origin for the Worldwide-Spread Mutational Event in Machado-Joseph Disease. Archives of Neurology, 2007, 64, 1502.	4.9	65
62	The correlation of clinical phenotype in Friedreich ataxia with the site of point mutations in the FRDA gene. Neurogenetics, 1998, 1, 253-257.	0.7	64
63	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
64	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
65	CYLD is a causative gene for frontotemporal dementia â€“ amyotrophic lateral sclerosis. Brain, 2020, 143, 783-799.	3.7	62
66	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
67	Homozygous mutations inMFN2cause multiple symmetric lipomatosis associated with neuropathy. Human Molecular Genetics, 2015, 24, 5109-5114.	1.4	61
68	Non-nuclear Pool of Splicing Factor SFPQ Regulates Axonal Transcripts Required for Normal Motor Development. Neuron, 2017, 94, 322-336.e5.	3.8	61
69	DNMT1 mutation hot spot causes varied phenotypes of HSAN1 with dementia and hearing loss. Neurology, 2013, 80, 824-828.	1.5	60
70	DÃ©jÃ©rine-Sottas neuropathy is associated with a de novo PMP22 mutation. Human Mutation, 1995, 5, 76-80.	1.1	59
71	Improved inherited peripheral neuropathy genetic diagnosis by wholeâ€exome sequencing. Molecular Genetics & Genomic Medicine, 2015, 3, 143-154.	0.6	59
72	Calpain Inhibition Is Protective in Machadoâ€“Joseph Disease Zebrafish Due to Induction of Autophagy. Journal of Neuroscience, 2017, 37, 7782-7794.	1.7	57

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73	A Tol2 Gateway-Compatible Toolbox for the Study of the Nervous System and Neurodegenerative Disease. Zebrafish, 2017, 14, 69-72.	0.5	56
74	Pathogenic Huntingtin Repeat Expansions in Patients with Frontotemporal Dementia and Amyotrophic Lateral Sclerosis. Neuron, 2021, 109, 448-460.e4.	3.8	56
75	Impaired NHEJ repair in amyotrophic lateral sclerosis is associated with TDP-43 mutations. Molecular Neurodegeneration, 2020, 15, 51.	4.4	54
76	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
77	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
78	A Novel Homozygous Mutation of the Myelin Po Gene Producing Dejerineâ€“Sottas Disease (Hereditary) Tj ETQq0 0 0 rgBT /Overlock 10 222, 107-110.	1.0	49
79	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
80	Meta-analysis of genome-wide DNA methylation identifies shared associations across neurodegenerative disorders. Genome Biology, 2021, 22, 90.	3.8	49
81	Association of Variants in the SPTLC1 Gene With Juvenile Amyotrophic Lateral Sclerosis. JAMA Neurology, 2021, 78, 1236.	4.5	46
82	Three novel mutations and two variants in the gene for Cu/Zn superoxide dismutase in familial amyotrophic lateral sclerosis. Neuromuscular Disorders, 1996, 6, 361-366.	0.3	45
83	<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
84	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
85	Dominant Intermediate Charcot-Marie-Tooth Neuropathy Maps to Chromosome 19p12-p13.2. American Journal of Human Genetics, 2001, 69, 883-888.	2.6	42
86	Hereditary Spastic Paraplegia 3A Associated With Axonal Neuropathy. Archives of Neurology, 2007, 64, 706.	4.9	42
87	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
88	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
89	Novel motor phenotypes in patients with VRK1 mutations without pontocerebellar hypoplasia. Neurology, 2016, 87, 65-70.	1.5	38
90	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

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91	Monozygotic twins and triplets discordant for amyotrophic lateral sclerosis display differential methylation and gene expression. Scientific Reports, 2019, 9, 8254.	1.6	36
92	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
93	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
94	Axonal Ion Channel Dysfunction inC9orf72Familial Amyotrophic Lateral Sclerosis. JAMA Neurology, 2015, 72, 49.	4.5	35
95	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
96	Replication of association between ELAVL4 and Parkinson disease: the GenePD study. Human Genetics, 2008, 124, 95-99.	1.8	34
97	Determination of gene dosage at the PMP22 and androgen receptor loci by quantitative PCR. Clinical Chemistry, 1998, 44, 724-730.	1.5	30
98	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
99	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
100	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
101	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
102	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
103	The dominantly inherited motor and sensory neuropathies: Clinical and molecular advances. Muscle and Nerve, 2006, 33, 589-597.	1.0	26
104	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
105	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
106	Quantitative muscle ultrasound as a biomarker in Charcot-Marie-Tooth neuropathy. Clinical Neurophysiology, 2017, 128, 227-232.	0.7	25
107	RFC1 expansions can mimic hereditary sensory neuropathy with cough and SjÃ¶gren syndrome. Brain, 2020, 143, e82-e82.	3.7	25
108	Significant out-of-sample classification from methylation profile scoring for amyotrophic lateral sclerosis. Npj Genomic Medicine, 2020, 5, 10.	1.7	25

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109	Deletion and Nonsense Mutations of the Connexin 32 Gene Associated with Charcot-Marie-Tooth Disease.. Tohoku Journal of Experimental Medicine, 1999, 188, 239-244.	0.5	24
110	Mutations in theSPTLC1Protein Cause Mitochondrial Structural Abnormalities and Endoplasmic Reticulum Stress in Lymphoblasts. DNA and Cell Biology, 2014, 33, 399-407.	0.9	24
111	Genetic basis of hindlimb loss in a naturally occurring vertebrate model. Biology Open, 2016, 5, 359-366.	0.6	24
112	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
113	A YAC-Based Transcript Map of Human Chromosome 9q22.1â€“q22.3 Encompassing the Loci for Hereditary Sensory Neuropathy Type I and Multiple Self-Healing Squamous Epithelioma. Genomics, 1998, 51, 277-281.	1.3	23
114	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
115	A novel locus for distal motor neuron degeneration maps to chromosome 7q34-q36. Human Genetics, 2007, 121, 559-564.	1.8	23
116	Motor cortical dysfunction develops in spinocerebellar ataxia type 3. Clinical Neurophysiology, 2016, 127, 3418-3424.	0.7	22
117	Correlation between muscle atrophy on MRI and manual strength testing in hereditary neuropathies. Journal of Clinical Neuroscience, 2010, 17, 874-878.	0.8	21
118	Identity by descent analysis identifies founder events and links SOD1 familial and sporadic ALS cases. Npj Genomic Medicine, 2020, 5, 32.	1.7	20
119	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
120	Mutation Testing in Charcot-Marie-Tooth Neuropathy. Annals of the New York Academy of Sciences, 1999, 883, 383-388.	1.8	19
121	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
122	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
123	The genetic landscape of axonal neuropathies in the middle-aged and elderly. Neurology, 2020, 95, e3163-e3179.	1.5	19
124	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
125	The Charcotâ€“Marieâ€“Tooth Binary Repeat Contains a Gene Transcribed from the Opposite Strand of a Partially Duplicated Region of theCOX10Gene. Genomics, 1997, 46, 61-69.	1.3	17
126	Mutation analysis of VCP in familial and sporadic amyotrophic lateral sclerosis. Neurobiology of Aging, 2012, 33, 1488.e15-1488.e16.	1.5	17

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127	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
128	Late-onset hereditary sensory neuropathy type I due to SPTLC1 mutation: Autopsy findings. Clinical Neurology and Neurosurgery, 2006, 108, 780-783.	0.6	16
129	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
130	Motor Neuron Abnormalities Correlate with Impaired Movement in Zebrafish that Express Mutant Superoxide Dismutase 1. Zebrafish, 2019, 16, 8-14.	0.5	16
131	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
132	Intermediate Forms of Charcot-Marie-Tooth Neuropathy: A Review. NeuroMolecular Medicine, 0, 8, 123-130.	1.8	16
133	Peripheral neuropathies of infancy. Developmental Medicine and Child Neurology, 2003, 45, 408-14.	1.1	13
134	Mutation analysis of the optineurin gene in familial amyotrophic lateral sclerosis. Neurobiology of Aging, 2012, 33, 210.e9-210.e10.	1.5	13
135	Mutation analysis of MATR3 in Australian familial amyotrophic lateral sclerosis. Neurobiology of Aging, 2015, 36, 1602.e1-1602.e2.	1.5	13
136	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
137	Charcot-Marie-Tooth neuropathy type 1A mutation: Apparent crossovers with D17S122 are due to a duplication. American Journal of Medical Genetics Part A, 1992, 44, 455-460.	2.4	12
138	AGE DEPENDENT PENETRANCE OF THREE DIFFERENT SUPEROXIDE DISMUTASE 1 (SOD 1) MUTATIONS. International Journal of Neuroscience, 2005, 115, 1119-1130.	0.8	12
139	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
140	Axonal excitability in X-linked dominant Charcot Marie Tooth disease. Clinical Neurophysiology, 2014, 125, 1261-1269.	0.7	12
141	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
142	Sarcolemmal excitability in the myotonic dystrophies. Muscle and Nerve, 2018, 57, 595-602.	1.0	12
143	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
144	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

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145	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
146	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
147	Reâ€anâ€alysis 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
148	Genome-wide association study identifies genetic factors that modify age at onset in Machado-Joseph disease. Aging, 2020, 12, 4742-4756.	1.4	10
149	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
150	GDAP1 mutations in CMT4: Axonal and demyelinating phenotypes?. Neurology, 2002, 59, 1835-1836.	1.5	9
151	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
152	Establishment of the Australasian paediatric Charcot-Marie-Tooth disease registry. Neuromuscular Disorders, 2007, 17, 349-350.	0.3	9
153	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
154	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
155	Exclusion of NFIL3 as the gene causing hereditary sensory neuropathy typeÂI by mutation analysis. Human Genetics, 2000, 106, 594-596.	1.8	8
156	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
157	Neuronal cell culture from transgenic zebrafish models of neurodegenerative disease. Biology Open, 2018, 7, .	0.6	8
158	Inherited Neuropathies. Seminars in Neurology, 2019, 39, 620-639.	0.5	8
159	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
160	De novo mutation of the myelin Po gene in DÃ©jÃ©rine-Sottas disease (hereditary motor and sensory) Tj ETQq0 0 0 rgBT /Overlock 10	1.1	7
161	Hypoxia Causes Aggregation of Serine Palmitoyltransferase followed by Non-Apoptotic Death of Human Lymphocytes. Cell Cycle, 2004, 3, 1271-1277.	1.3	7
162	Mutation analysis of genes within the dynactin complex in a cohort of hereditary peripheral neuropathies. Clinical Genetics, 2016, 90, 127-133.	1.0	7

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164	OUP accepted manuscript. Brain, 2018, 141, e66.	3.7	7
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