Garth A Nicholson

List of Publications by Citations

190 papers	14,331 citations	57 h-index	117 g-index
202 ext. papers	16,620 ext. citations	8.2 avg, IF	5.51 L-index

#	Paper	IF	Citations
190	Mutations in FUS, an RNA processing protein, cause familial amyotrophic lateral sclerosis type 6. *Science*, **2009**, 323, 1208-1211	33.3	1890
189	TDP-43 mutations in familial and sporadic amyotrophic lateral sclerosis. *Science*, **2008**, 319, 1668-72	33.3	1877
188	DNA/RNA helicase gene mutations in a form of juvenile amyotrophic lateral sclerosis (ALS4). *American Journal of Human Genetics*, **2004**, 74, 1128-35	11	607
187	Controversies and priorities in amyotrophic lateral sclerosis. *Lancet Neurology, The*, **2013**, 12, 310-22	24.1	377
186	Mutations in SPTLC1, encoding serine palmitoyltransferase, long chain base subunit-1, cause hereditary sensory neuropathy type I. *Nature Genetics*, **2001**, 27, 309-12	36.3	341
185	Genome-wide association analyses identify new risk variants and the genetic architecture of amyotrophic lateral sclerosis. *Nature Genetics*, **2016**, 48, 1043-8	36.3	328
184	Clinical and pathological features of a parkinsonian syndrome in a family with an Ala53Thr α-synuclein mutation. *Annals of Neurology*, **2001**, 49, 313-319	9.4	316
183	Cortical hyperexcitability may precede the onset of familial amyotrophic lateral sclerosis. *Brain*, **2008**, 131, 1540-50	11.2	307
182	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, 20881-90	11.5	302
181	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. *Neuron*, **2018**, 97, 1268-1283.e6	13.9	296
180	Mutations in DNMT1 cause hereditary sensory neuropathy with dementia and hearing loss. *Nature Genetics*, **2011**, 43, 595-600	36.3	284
179	Mutations in the pleckstrin homology domain of dynamin 2 cause dominant intermediate Charcot-Marie-Tooth disease. *Nature Genetics*, **2005**, 37, 289-94	36.3	278
178	A frame shift mutation in the PMP22 gene in hereditary neuropathy with liability to pressure palsies. *Nature Genetics*, **1994**, 6, 263-6	36.3	238
177	Hereditary sensory neuropathy type 1 is caused by the accumulation of two neurotoxic sphingolipids. *Journal of Biological Chemistry*, **2010**, 285, 11178-87	5.4	233
176	Exome-wide rare variant analysis identifies TUBA4A mutations associated with familial ALS. *Neuron*, **2014**, 84, 324-31	13.9	229
175	Evaluating the role of the FUS/TLS-related gene EWSR1 in amyotrophic lateral sclerosis. *Human Molecular Genetics*, **2012**, 21, 2899-911	5.6	207
174	De novo mutation of the myelin P0 gene in Dejerine-Sottas disease (hereditary motor and sensory neuropathy type III). *Nature Genetics*, **1993**, 5, 266-8	36.3	193

173	FUS mutations in amyotrophic lateral sclerosis: clinical, pathological, neurophysiological and genetic analysis. *Journal of Neurology, Neurosurgery and Psychiatry*, **2010**, 81, 639-45	5.5	177
172	Compound heterozygosity for loss-of-function lysyl-tRNA synthetase mutations in a patient with peripheral neuropathy. *American Journal of Human Genetics*, **2010**, 87, 560-6	11	150
171	NEK1 variants confer susceptibility to amyotrophic lateral sclerosis. *Nature Genetics*, **2016**, 48, 1037-42	36.3	149
170	Two novel (M233T and R278T) presenilin-1 mutations in early-onset Alzheimer's disease pedigrees and preliminary evidence for association of presenilin-1 mutations with a novel phenotype. *NeuroReport*, **1997**, 8, 1537-42	1.7	149
169	Tau haplotypes regulate transcription and are associated with Parkinson's disease. *Annals of Neurology*, **2004**, 55, 329-34	9.4	147
168	Missense mutations in the copper transporter gene ATP7A cause X-linked distal hereditary motor neuropathy. *American Journal of Human Genetics*, **2010**, 86, 343-52	11	142
167	KIF1A, an axonal transporter of synaptic vesicles, is mutated in hereditary sensory and autonomic neuropathy type 2. *American Journal of Human Genetics*, **2011**, 89, 219-30	11	136
166	Influence of heterozygosity for parkin mutation on onset age in familial Parkinson disease: the GenePD study. *Archives of Neurology*, **2006**, 63, 826-32		131
165	CCNF mutations in amyotrophic lateral sclerosis and frontotemporal dementia. *Nature Communications*, **2016**, 7, 11253	17.4	126
164	Use of Whole-Exome Sequencing for Diagnosis of Limb-Girdle Muscular Dystrophy: Outcomes and Lessons Learned. *JAMA Neurology*, **2015**, 72, 1424-32	17.2	121
163	The gene for hereditary sensory neuropathy type I (HSN-I) maps to chromosome 9q22.1-q22.3. *Nature Genetics*, **1996**, 13, 101-4	36.3	117
162	Exome sequencing to identify de novo mutations in sporadic ALS trios. *Nature Neuroscience*, **2013**, 16, 851-5	25.5	112
161	Phenotypic spectrum of disorders associated with glycyl-tRNA synthetase mutations. *Brain*, **2005**, 128, 2304-14	11.2	109
160	Peripheral nerve demyelination caused by a mutant Rho GTPase guanine nucleotide exchange factor, frabin/FGD4. *American Journal of Human Genetics*, **2007**, 81, 158-64	11	107
159	Clinical and genetic study of Friedreich ataxia in an Australian population. *American Journal of Medical Genetics Part A*, **1999**, 87, 168-74		107
158	ERBB4 mutations that disrupt the neuregulin-ErbB4 pathway cause amyotrophic lateral sclerosis type 19. *American Journal of Human Genetics*, **2013**, 93, 900-5	11	95
157	UBQLN2/ubiquilin 2 mutation and pathology in familial amyotrophic lateral sclerosis. *Neurobiology of Aging*, **2012**, 33, 2527.e3-10	5.6	95
156	Distinctive genetic and clinical features of CMT4J: a severe neuropathy caused by mutations in the PI(3,5)P□phosphatase FIG4. *Brain*, **2011**, 134, 1959-71	11.2	89

155	A loss-of-function variant in the human histidyl-tRNA synthetase (HARS) gene is neurotoxic in vivo. *Human Mutation*, **2013**, 34, 191-9	4.7	85
154	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, 351-7		84
153	Expanding the clinical, pathological and MRI phenotype of DNM2-related centronuclear myopathy. *Neuromuscular Disorders*, **2010**, 20, 229-37	2.9	82
152	Variable phenotype of Alzheimer's disease with spastic paraparesis. *Annals of Neurology*, **2001**, 49, 125-9	9.4	81
151	Pathophysiological insights into ALS with C9ORF72 expansions. *Journal of Neurology, Neurosurgery and Psychiatry*, **2013**, 84, 931-5	5.5	80
150	Variants associated with Gaucher disease in multiple system atrophy. *Annals of Clinical and Translational Neurology*, **2015**, 2, 417-26	5.3	76
149	The TRK-fused gene is mutated in hereditary motor and sensory neuropathy with proximal dominant involvement. *American Journal of Human Genetics*, **2012**, 91, 320-9	11	76
148	A recurrent loss-of-function alanyl-tRNA synthetase (AARS) mutation in patients with Charcot-Marie-Tooth disease type 2N (CMT2N). *Human Mutation*, **2012**, 33, 244-53	4.7	75
147	The Gly2019Ser mutation in LRRK2 is not fully penetrant in familial Parkinson's disease: the GenePD study. *BMC Medicine*, **2008**, 6, 32	11.4	72
146	Intermediate forms of Charcot-Marie-Tooth neuropathy: a review. *NeuroMolecular Medicine*, **2006**, 8, 123-30	4.6	72
145	Histopathological findings in hereditary motor and sensory neuropathy of axonal type with onset in early childhood associated with mitofusin 2 mutations. *Journal of Neuropathology and Experimental Neurology*, **2008**, 67, 1097-102	3.1	69
144	Spinocerebellar ataxia type 15 (sca15) maps to 3p24.2-3pter: exclusion of the ITPR1 gene, the human orthologue of an ataxic mouse mutant. *Neurobiology of Disease*, **2003**, 13, 147-57	7.5	65
143	Autosomal dominant inherited neuropathies with prominent sensory loss and mutilations: a review. *Archives of Neurology*, **2003**, 60, 329-34		65
142	Alteration of ornithine metabolism leads to dominant and recessive hereditary spastic paraplegia. *Brain*, **2015**, 138, 2191-205	11.2	64
141	Hereditary spastic paraplegia type 43 (SPG43) is caused by mutation in C19orf12. *Human Mutation*, **2013**, 34, 1357-60	4.7	64
140	Pedigree with frontotemporal lobar degeneration--motor neuron disease and Tar DNA binding protein-43 positive neuropathology: genetic linkage to chromosome 9. *BMC Neurology*, **2008**, 8, 32	3.1	64
139	Autosomal dominant juvenile amyotrophic lateral sclerosis and distal hereditary motor neuronopathy with pyramidal tract signs: synonyms for the same disorder?. *Brain*, **2002**, 125, 1320-5	11.2	63
138	Evidence for an X-linked genetic component in familial typical migraine. *Human Molecular Genetics*, **1998**, 7, 459-63	5.6	62

137	Phenotypic spectrum of dynamin 2 mutations in Charcot-Marie-Tooth neuropathy. *Brain*, **2009**, 132, 1741-52	11.2	60
136	Cortical Function in Asymptomatic Carriers and Patients With C9orf72 Amyotrophic Lateral Sclerosis. *JAMA Neurology*, **2015**, 72, 1268-74	17.2	59
135	Hypomorphic mutations in POLR3A are a frequent cause of sporadic and recessive spastic ataxia. *Brain*, **2017**, 140, 1561-1578	11.2	58
134	A locus for hereditary sensory neuropathy with cough and gastroesophageal reflux on chromosome 3p22-p24. *American Journal of Human Genetics*, **2003**, 73, 632-7	11	58
133	The correlation of clinical phenotype in Friedreich ataxia with the site of point mutations in the FRDA gene. *Neurogenetics*, **1998**, 1, 253-7	3	56
132	Autosomal dominant hereditary sensory neuropathy with chronic cough and gastro-oesophageal reflux: clinical features in two families linked to chromosome 3p22-p24. *Brain*, **2005**, 128, 2797-810	11.2	55
131	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, 100-6	3.2	54
130	Homozygous mutations in MFN2 cause multiple symmetric lipomatosis associated with neuropathy. *Human Molecular Genetics*, **2015**, 24, 5109-14	5.6	53
129	Asian origin for the worldwide-spread mutational event in Machado-Joseph disease. *Archives of Neurology*, **2007**, 64, 1502-8		50
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, 349-52	5.5	49
127	DJ[illegible]ine-Sottas neuropathy is associated with a de novo PMP22 mutation. *Human Mutation*, **1995**, 5, 76-80	4.7	49
126	A recurrent WARS mutation is a novel cause of autosomal dominant distal hereditary motor neuropathy. *Brain*, **2017**, 140, 1252-1266	11.2	48
125	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, 1404-16	5.6	48
124	Improved inherited peripheral neuropathy genetic diagnosis by whole-exome sequencing. *Molecular Genetics & Genomic Medicine*, **2015**, 3, 143-54	2.3	47
123	A novel homozygous mutation of the myelin Po gene producing Dejerine-Sottas disease (hereditary motor and sensory neuropathy type III). *Biochemical and Biophysical Research Communications*, **1996**, 222, 107-10	3.4	47
122	TDP-43: a DNA and RNA binding protein with roles in neurodegenerative diseases. *International Journal of Biochemistry and Cell Biology*, **2010**, 42, 1606-9	5.6	46
121	DNMT1 mutation hot spot causes varied phenotypes of HSAN1 with dementia and hearing loss. *Neurology*, **2013**, 80, 824-8	6.5	45
120	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, 193-9		43

119	Mutation screening of the N-myc downstream-regulated gene 1 (NDRG1) in patients with Charcot-Marie-Tooth Disease. *Human Mutation*, **2003**, 22, 129-35	4.7	41
118	Calpain Inhibition Is Protective in Machado-Joseph Disease Zebrafish Due to Induction of Autophagy. *Journal of Neuroscience*, **2017**, 37, 7782-7794	6.6	40
117	Hereditary spastic paraplegia 3A associated with axonal neuropathy. *Archives of Neurology*, **2007**, 64, 706-13		37
116	Non-nuclear Pool of Splicing Factor SFPQ Regulates Axonal Transcripts Required for Normal Motor Development. *Neuron*, **2017**, 94, 322-336.e5	13.9	36
115	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, 1415-8	5.5	34
114	Dominant intermediate Charcot-Marie-Tooth neuropathy maps to chromosome 19p12-p13.2. *American Journal of Human Genetics*, **2001**, 69, 883-8	11	34
113	CYLD is a causative gene for frontotemporal dementia - amyotrophic lateral sclerosis. *Brain*, **2020**, 143, 783-799	11.2	33
112	Three novel mutations and two variants in the gene for Cu/Zn superoxide dismutase in familial amyotrophic lateral sclerosis. *Neuromuscular Disorders*, **1996**, 6, 361-6	2.9	33
111	MORC2 mutations cause axonal Charcot-Marie-Tooth disease with pyramidal signs. *Annals of Neurology*, **2016**, 79, 419-27	9.4	32
110	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, 168-75	6.9	31
109	A Tol2 Gateway-Compatible Toolbox for the Study of the Nervous System and Neurodegenerative Disease. *Zebrafish*, **2017**, 14, 69-72	2	30
108	Stoichiometric alteration of PMP22 protein determines the phenotype of hereditary neuropathy with liability to pressure palsies. *Archives of Neurology*, **2007**, 64, 974-8		30
107	Determination of gene dosage at the PMP22 and androgen receptor loci by quantitative PCR. *Clinical Chemistry*, **1998**, 44, 724-730	5.5	30
106	Novel TBK1 truncating mutation in a familial amyotrophic lateral sclerosis patient of Chinese origin. *Neurobiology of Aging*, **2015**, 36, 3334.e1-3334.e5	5.6	29
105	Replication of association between ELAVL4 and Parkinson disease: the GenePD study. *Human Genetics*, **2008**, 124, 95-9	6.3	28
104	Axonal ion channel dysfunction in c9orf72 familial amyotrophic lateral sclerosis. *JAMA Neurology*, **2015**, 72, 49-57	17.2	27
103	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, 585-9	7	27
102	Fused in sarcoma/translocated in liposarcoma: a multifunctional DNA/RNA binding protein. *International Journal of Biochemistry and Cell Biology*, **2010**, 42, 1408-11	5.6	25

101	The dominantly inherited motor and sensory neuropathies: clinical and molecular advances. *Muscle and Nerve*, **2006**, 33, 589-97	3.4	25
100	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	5.6	24
99	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, 135-43	3	22
98	Novel motor phenotypes in patients with VRK1 mutations without pontocerebellar hypoplasia. *Neurology*, **2016**, 87, 65-70	6.5	22
97	Monozygotic twins and triplets discordant for amyotrophic lateral sclerosis display differential methylation and gene expression. *Scientific Reports*, **2019**, 9, 8254	4.9	21
96	Evaluation of Skin Fibroblasts from Amyotrophic Lateral Sclerosis Patients for the Rapid Study of Pathological Features. *Neurotoxicity Research*, **2015**, 28, 138-46	4.3	21
95	Deletion and nonsense mutations of the connexin 32 gene associated with Charcot-Marie-Tooth disease. *Tohoku Journal of Experimental Medicine*, **1999**, 188, 239-44	2.4	21
94	A novel locus for distal motor neuron degeneration maps to chromosome 7q34-q36. *Human Genetics*, **2007**, 121, 559-64	6.3	20
93	Pathogenic Huntingtin Repeat Expansions in Patients with Frontotemporal Dementia and Amyotrophic Lateral Sclerosis. *Neuron*, **2021**, 109, 448-460.e4	13.9	20
92	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	3.6	19
91	Mutations in the SPTLC1 protein cause mitochondrial structural abnormalities and endoplasmic reticulum stress in lymphoblasts. *DNA and Cell Biology*, **2014**, 33, 399-407	3.6	19
90	Correlation between muscle atrophy on MRI and manual strength testing in hereditary neuropathies. *Journal of Clinical Neuroscience*, **2010**, 17, 874-8	2.2	19
89	Mutational origin of Machado-Joseph disease in the Australian Aboriginal communities of Groote Eylandt and Yirrkala. *Archives of Neurology*, **2012**, 69, 746-51		19
88	Exclusion of serine palmitoyltransferase long chain base subunit 2 (SPTLC2) as a common cause for hereditary sensory neuropathy. *Neuromuscular Disorders*, **2002**, 12, 656-8	2.9	19
87	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	36.3	19
86	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, 135-8		18
85	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	10.6	18
84	The Charcot-Marie-Tooth binary repeat contains a gene transcribed from the opposite strand of a partially duplicated region of the COX10 gene. *Genomics*, **1997**, 46, 61-9	4.3	17

83	Association study on glutathione S-transferase omega 1 and 2 and familial ALS. *Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders*, **2008**, 9, 81-4		17
82	A YAC-based transcript map of human chromosome 9q22.1-q22.3 encompassing the loci for hereditary sensory neuropathy type I and multiple self-healing squamous epithelioma. *Genomics*, **1998**, 51, 277-81	4.3	17
81	Mutation Testing in Charcot-Marie-Tooth Neuropathy. *Annals of the New York Academy of Sciences*, **1999**, 883, 383-388	6.5	17
80	Impaired NHEJ repair in amyotrophic lateral sclerosis is associated with TDP-43 mutations. *Molecular Neurodegeneration*, **2020**, 15, 51	19	17
79	Quantitative muscle ultrasound as a biomarker in Charcot-Marie-Tooth neuropathy. *Clinical Neurophysiology*, **2017**, 128, 227-232	4.3	16
78	Genetic basis of hindlimb loss in a naturally occurring vertebrate model. *Biology Open*, **2016**, 5, 359-66	2.2	16
77	Genetic and Pathological Assessment of hnRNPA1, hnRNPA2/B1, and hnRNPA3 in Familial and Sporadic Amyotrophic Lateral Sclerosis. *Neurodegenerative Diseases*, **2017**, 17, 304-312	2.3	16
76	Mutant human FUS Is ubiquitously mislocalized and generates persistent stress granules in primary cultured transgenic zebrafish cells. *PLoS ONE*, **2014**, 9, e90572	3.7	16
75	Motor cortical dysfunction develops in spinocerebellar ataxia type 3. *Clinical Neurophysiology*, **2016**, 127, 3418-3424	4.3	16
74	Mutation analysis and immunopathological studies of PFN1 in familial and sporadic amyotrophic lateral sclerosis. *Neurobiology of Aging*, **2013**, 34, 2235.e7-10	5.6	15
73	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	6	15
72	Mutation analysis of VCP in familial and sporadic amyotrophic lateral sclerosis. *Neurobiology of Aging*, **2012**, 33, 1488.e15-6	5.6	14
71	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	2.3	13
70	Mutation analysis of the optineurin gene in familial amyotrophic lateral sclerosis. *Neurobiology of Aging*, **2012**, 33, 210.e9-10	5.6	13
69	Intermediate Forms of Charcot-Marie-Tooth Neuropathy: A Review. *NeuroMolecular Medicine*,8, 123-130	4.6	13
68	Mutation analysis of MATR3 in Australian familial amyotrophic lateral sclerosis. *Neurobiology of Aging*, **2015**, 36, 1602.e1-2	5.6	12
67	Late-onset hereditary sensory neuropathy type I due to SPTLC1 mutation: autopsy findings. *Clinical Neurology and Neurosurgery*, **2006**, 108, 780-3	2	12
66	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, 455-60		12

65	Significant out-of-sample classification from methylation profile scoring for amyotrophic lateral sclerosis. *Npj Genomic Medicine*, **2020**, 5, 10	6.2	11
64	A novel TARDBP insertion/deletion mutation in the flail arm variant of amyotrophic lateral sclerosis. *Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders*, **2012**, 13, 465-70		11
63	Age dependent penetrance of three different superoxide dismutase 1 (sod 1) mutations. *International Journal of Neuroscience*, **2005**, 115, 1119-30	2	11
62	A Simple Differentiation Protocol for Generation of Induced Pluripotent Stem Cell-Derived Basal Forebrain-Like Cholinergic Neurons for Alzheimer's Disease and Frontotemporal Dementia Disease Modeling. *Cells*, **2020**, 9,	7.9	11
61	Sarcolemmal excitability in the myotonic dystrophies. *Muscle and Nerve*, **2018**, 57, 595-602	3.4	10
60	Hereditary sensory neuropathy type 1 in a Portuguese family-electrodiagnostic and autonomic nervous system studies. *Journal of the Neurological Sciences*, **2004**, 227, 35-8	3.2	10
59	Pathogenic mechanisms underlying X-linked Charcot-Marie-Tooth neuropathy (CMTX6) in patients with a pyruvate dehydrogenase kinase 3 mutation. *Neurobiology of Disease*, **2016**, 94, 237-44	7.5	10
58	Relationship between physical performance and quality of life in Charcot-Marie-Tooth disease: a pilot study. *Journal of the Peripheral Nervous System*, **2016**, 21, 357-364	4.7	10
57	Characterizing the molecular phenotype of an Atp7a(T985I) conditional knock in mouse model for X-linked distal hereditary motor neuropathy (dHMNX). *Metallomics*, **2016**, 8, 981-92	4.5	9
56	Axonal excitability in X-linked dominant Charcot Marie Tooth disease. *Clinical Neurophysiology*, **2014**, 125, 1261-9	4.3	9
55	Re-analysis of an original CMTX3 family using exome sequencing identifies a known BSCL2 mutation. *Muscle and Nerve*, **2013**, 47, 922-4	3.4	9
54	Biomarkers of disease in a case of familial lower motor neuron ALS. *Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders*, **2010**, 11, 486-9		8
53	Exclusion of NFIL3 as the gene causing hereditary sensory neuropathy type I by mutation analysis. *Human Genetics*, **2000**, 106, 594-6	6.3	8
52	Huntington CAG repeat size does not modify onset age in familial Parkinson's disease: the GenePD study. *Movement Disorders*, **2008**, 23, 1596-601	7	7
51	Establishment of the Australasian paediatric Charcot-Marie-Tooth disease registry. *Neuromuscular Disorders*, **2007**, 17, 349-50	2.9	7
50	Hypoxia causes aggregation of serine palmitoyltransferase followed by non-apoptotic death of human lymphocytes. *Cell Cycle*, **2004**, 3, 1271-7	4.7	7
49	Polygenic risk score analysis for amyotrophic lateral sclerosis leveraging cognitive performance, educational attainment and schizophrenia. *European Journal of Human Genetics*, **2021**,	5.3	7
48	Evidence of a founder haplotype refines the X-linked Charcot-Marie-Tooth (CMTX3) locus to a 2.5 Mb region. *Neurogenetics*, **2008**, 9, 191-5	3	6

47	Refined localization of dominant intermediate Charcot-Marie-Tooth neuropathy and exclusion of seven known candidate genes in the region. *Neurogenetics*, **2003**, 4, 179-83	3	6
46	Genomic structure and physical mapping of C17orf1: a gene associated with the proximal element of the CMT1A-REP binary repeat. *Genomics*, **1998**, 53, 110-2	4.3	6
45	Genome-wide association study identifies genetic factors that modify age at onset in Machado-Joseph disease. *Aging*, **2020**, 12, 4742-4756	5.6	6
44	RFC1 expansions can mimic hereditary sensory neuropathy with cough and Sj□gren syndrome. *Brain* , **2020**, 143, e82	11.2	6
43	Meta-analysis of genome-wide DNA methylation identifies shared associations across neurodegenerative disorders. *Genome Biology*, **2021**, 22, 90	18.3	6
42	Mutation analysis of genes within the dynactin complex in a cohort of hereditary peripheral neuropathies. *Clinical Genetics*, **2016**, 90, 127-33	4	6
41	Clinical and pathological features of a parkinsonian syndrome in a family with an Ala53Thr □synuclein mutation **2001**, 49, 313		6
40	The genetic landscape of axonal neuropathies in the middle-aged and elderly: Focus on. *Neurology*, **2020**, 95, e3163-e3179	6.5	5
39	A 1.35 ́Mb DNA fragment is inserted into the DHMN1 locus on chromosome 7q34-q36.2. *Human Genetics*, **2016**, 135, 1269-1278	6.3	5
38	Inherited Neuropathies. *Seminars in Neurology*, **2019**, 39, 620-639	3.2	5
37	De novo mutation of the myelin Po gene in D□□ine-Sottas disease (hereditary motor and sensory neuropathy type III): two amino acid insertion after Asp 118. *Human Mutation*, **1998**, Suppl 1, S103-5	4.7	5
36	Exclusion of NFIL3 as the gene causing hereditary sensory neuropathy type I by mutation analysis. *Human Genetics*, **2000**, 106, 594-596	6.3	5
35	Motor Neuron Abnormalities Correlate with Impaired Movement in Zebrafish that Express Mutant Superoxide Dismutase 1. *Zebrafish*, **2019**, 16, 8-14	2	5
34	Neuronal cell culture from transgenic zebrafish models of neurodegenerative disease. *Biology Open* , **2018**, 7,	2.2	5
33	Association of Variants in the SPTLC1 Gene With Juvenile Amyotrophic Lateral Sclerosis. *JAMA Neurology*, **2021**, 78, 1236-1248	17.2	5
32	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	3	4
31	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	5.5	4
30	"Dancing feet dyskinesias": a clue to parkin gene mutations. *Movement Disorders*, **2012**, 27, 587-8	7	4

29	A novel MCM3AP mutation in a Lebanese family with recessive Charcot-Marie-Tooth neuropathy. *Brain*, **2018**, 141, e66	11.2	4
28	Identity by descent analysis identifies founder events and links familial and sporadic ALS cases. *Npj Genomic Medicine*, **2020**, 5, 32	6.2	4
27	Genome-wide study of DNA methylation shows alterations in metabolic, inflammatory, and cholesterol pathways in ALS.. *Science Translational Medicine*, **2022**, 14, eabj0264	17.5	4
26	Infantile-Onset Myelin Protein Zero-Related Demyelinating Neuropathy Presenting as an Upper Extremity Monoplegia. *Seminars in Pediatric Neurology*, **2018**, 26, 52-55	2.9	3
25	Analysis of dynein intermediate chains, light intermediate chains and light chains in a cohort of hereditary peripheral neuropathies. *Neurogenetics*, **2014**, 15, 229-35	3	3
24	The MFN2 V705I Variant Is Not a Disease-Causing Mutation: A Segregation Analysis in a CMT2 Family. *Journal of Neurodegenerative Diseases*, **2013**, 2013, 495873		3
23	X-linked CMT: genes and gene loci in an Australian cohort. *Neurogenetics*, **2010**, 11, 267-9	3	3
22	Peripheral neuropathies of infancy. *Developmental Medicine and Child Neurology*, **2003**, 45, 408-14	3.3	3
21	Modelling the pathogenesis of X-linked distal hereditary motor neuropathy using patient-derived iPSCs. *DMM Disease Models and Mechanisms*, **2020**, 13,	4.1	3
20	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	1.6	2
19	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	4.7	2
18	Unique clinical and neurophysiologic profile of a cohort of children with CMTX3. *Neurology*, **2018**, 90, e1706-e1710	6.5	2
17	Apparent anticipation in SOD1 familial amyotrophic lateral sclerosis. *Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration*, **2013**, 14, 452-6	3.6	2
16	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	3	2
15	Genome-Wide Analyses Identify KIF5A as a Novel ALS Gene. *SSRN Electronic Journal*,	1	2
14	IBD analysis of Australian amyotrophic lateral sclerosis SOD1-mutation carriers identifies five founder events and links sporadic cases to existing ALS families		2
13	Genetic analysis of GLT8D1 and ARPP21 in Australian familial and sporadic amyotrophic lateral sclerosis. *Neurobiology of Aging*, **2021**, 101, 297.e9-297.e11	5.6	2
12	Body composition and its association with physical performance, quality of life, and clinical indictors in Charcot-Marie-Tooth disease: a pilot study. *Disability and Rehabilitation*, **2019**, 41, 405-412	2.4	2

11	Sodium valproate increases activity of the sirtuin pathway resulting in beneficial effects for spinocerebellar ataxia-3 in vivo. *Molecular Brain*, **2021**, 14, 128	4.5	2
10	A family with 2 X-linked disorders: Charcot-Marie-Tooth disease and hemophilia A. *Muscle and Nerve* , **2012**, 46, 454-5	3.4	1
9	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, 201-6	3.6	1
8	Genome-wide study of DNA methylation in Amyotrophic Lateral Sclerosis identifies differentially methylated loci and implicates metabolic, inflammatory and cholesterol pathways		1
7	A de novo EGR2 variant, c.1232A > G p.Asp411Gly, causes severe early-onset Charcot-Marie-Tooth Neuropathy Type 3 (Dejerine-Sottas Neuropathy). *Scientific Reports*, **2019**, 9, 19336	4.9	1
6	A novel Parkinson's disease risk variant, p. W378R, in the Gaucher's disease GBA gene. *Movement Disorders*, **2018**, 33, 1662-1664	7	1
5	Charcot-Marie-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**,	5.6	1
4	Long read sequencing overcomes challenges in the diagnosis of SORD neuropathy.. *Journal of the Peripheral Nervous System*, **2022**,	4.7	1
3	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	4.7	
2	Hereditary Sensory Neuropathy **2006**, 329-335		
1	Revisiting the pathogenic mechanism of the GJB1 5PUTR c.-103C > T mutation causing CMTX1. *Neurogenetics*, **2021**, 22, 149-160	3	