Michael E Shy

List of Publications by Year
in descending order

239	11,891	25034	36028
papers	citations	57 h-index	97 g-index

245	245	245	8983
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Charcotâ€marieâ€tooth disease subtypes and genetic testing strategies. Annals of Neurology, 2011, 69, 22-33.	5.3	494
2	Mutation of FIG4 causes neurodegeneration in the pale tremor mouse and patients with CMT4J. Nature, 2007, 448, 68-72.	27.8	438
3	Axonal neuropathy with optic atrophy is caused by mutations in mitofusin 2. Annals of Neurology, 2006, 59, 276-281.	5.3	380
4	Neurological dysfunction and axonal degeneration in Charcot-Marie-Tooth disease type 1A. Brain, 2000, 123, 1516-1527.	7.6	359
5	MFN2 mutation distribution and genotype/phenotype correlation in Charcot-Marie-Tooth type 2. Brain, 2006, 129, 2093-2102.	7.6	351
6	Patients lacking the major CNS myelin protein, proteolipid protein 1, develop length-dependent axonal degeneration in the absence of demyelination and inflammation. Brain, 2002, 125, 551-561.	7.6	272
7	Reliability of the CMT neuropathy score (second version) in Charcotâ€Marieâ€Tooth disease. Journal of the Peripheral Nervous System, 2011, 16, 191-198.	3.1	269
8	Charcotâ€Marieâ€Tooth disease and related neuropathies: Mutation distribution and genotypeâ€phenotype correlation. Annals of Neurology, 2002, 51, 190-201.	5.3	257
9	Phenotypic clustering in MPZ mutations. Brain, 2004, 127, 371-384.	7.6	257
10	Vitamin A controls epithelial/mesenchymal interactions through Ret expression. Nature Genetics, 2001, 27, 74-78.	21.4	240
11	Exome Sequence Analysis Suggests that Genetic Burden Contributes to Phenotypic Variability and Complex Neuropathy. Cell Reports, 2015, 12, 1169-1183.	6.4	211
12	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
13	Increased Survival and Function of SOD1 Mice After Glial Cell-Derived Neurotrophic Factor Gene Therapy. Human Gene Therapy, 2002, 13, 1047-1059.	2.7	165
14	GDAP1, the protein causing Charcotâ€“Marieâ€“Tooth disease type 4A, is expressed in neurons and is associated with mitochondria. Human Molecular Genetics, 2005, 14, 1087-1094.	2.9	164
15	Transient central nervous system white matter abnormality in X-linked Charcot-Marie-Tooth disease. Annals of Neurology, 2002, 52, 429-434.	5.3	150
16	Asymmetric flaccid paralysis: A neuromuscular presentation of West Nile virus infection. Annals of Neurology, 2003, 53, 703-710.	5.3	142
17	Proteolipid Protein Is Necessary in Peripheral as Well as Central Myelin. Neuron, 1997, 19, 205-218.	8.1	140
18	Electrophysiological features of inherited demyelinating neuropathies: A reappraisal in the era of molecular diagnosis. Muscle and Nerve, 2000, 23, 1472-1487.	2.2	138

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19	Hereditary neuropathy with liability to pressure palsy. Neurology, 2002, 58, 1769-1773.	1.1	138
20	Validation of the Charcotâ€“Marieâ€“Tooth disease pediatric scale as an outcome measure of disability. Annals of Neurology, 2012, 71, 642-652.	5.3	137
21	The Molecular Pathogenesis of Pelizaeus-Merzbacher Disease. Archives of Neurology, 1999, 56, 1210.	4.5	131
22	Mutation of FIG4 causes a rapidly progressive, asymmetric neuronal degeneration. Brain, 2008, 131, 1990-2001.	7.6	128
23	Update on Charcot-Marie-Tooth Disease. Current Neurology and Neuroscience Reports, 2011, 11, 78-88.	4.2	128
24	High-Dosage Ascorbic Acid Treatment in Charcot-Marie-Tooth Disease Type 1A. JAMA Neurology, 2013, 70, 981.	9.0	121
25	Mutations in PRPS1, Which Encodes the Phosphoribosyl Pyrophosphate Synthetase Enzyme Critical for Nucleotide Biosynthesis, Cause Hereditary Peripheral Neuropathy with Hearing Loss and Optic Neuropathy (CMTX5). American Journal of Human Genetics, 2007, 81, 552-558.	6.2	116
26	A novel mutation in VCP causes Charcotâ€“Marieâ€“Tooth Type 2 disease. Brain, 2014, 137, 2897-2902.	7.6	116
27	Skin biopsies in myelin-related neuropathies: bringing molecular pathology to the bedside. Brain, 2005, 128, 1168-1177.	7.6	113
28	Exome sequencing allows for rapid gene identification in a Charcotâ€Marieâ€Tooth family. Annals of Neurology, 2011, 69, 464-470.	5.3	107
29	Heterozygous P0 Knockout Mice Develop a Peripheral Neuropathy that Resembles Chronic Inflammatory Demyelinating Polyneuropathy (CIDP). Journal of Neuropathology and Experimental Neurology, 1997, 56, 811-821.	1.7	106
30	Charcot-Marie-Tooth disease: an update. Current Opinion in Neurology, 2004, 17, 579-585.	3.6	99
31	Axonal Charcotâ€“Marieâ€“Tooth disease patient-derived motor neurons demonstrate disease-specific phenotypes including abnormal electrophysiological properties. Experimental Neurology, 2015, 263, 190-199.	4.1	97
32	Biallelic mutations in SORD cause a common and potentially treatable hereditary neuropathy with implications for diabetes. Nature Genetics, 2020, 52, 473-481.	21.4	97
33	Axonal Charcotâ€“Marieâ€“Tooth disease. Current Opinion in Neurology, 2011, 24, 475-483.	3.6	91
34	Curcumin derivatives promote Schwann cell differentiation and improve neuropathy in R98C CMT1B mice. Brain, 2012, 135, 3551-3566.	7.6	90
35	Inherited Peripheral Neuropathies. Neurologic Clinics, 2013, 31, 597-619.	1.8	88
36	Measurement and significance of antibodies against GM1 ganglioside Report of a workshop, 18 April 1989, Chicago, IL, U.S.A.. Journal of Neuroimmunology, 1989, 25, 255-259.	2.3	86

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37	SIMPLEmutations in Charcot-Marie-Tooth disease and the potential role of its protein product in protein degradation. Human Mutation, 2005, 25, 372-383.	2.5	86
38	Shortened internodal length of dermal myelinated nerve fibres in Charcot-Marie-Tooth disease type 1A. Brain, 2009, 132, 3263-3273.	7.6	85
39	Hypomorphic mutations in POLR3A are a frequent cause of sporadic and recessive spastic ataxia. Brain, 2017, 140, 1561-1578.	7.6	85
40	Mutations in the cytoplasmic domain of P0 reveal a role for PKC-mediated phosphorylation in adhesion and myelination. Journal of Cell Biology, 2001, 155, 439-446.	5.2	84
41	Peripheral neuropathies caused by mutations in the myelin protein zero. Journal of the Neurological Sciences, 2006, 242, 55-66.	0.6	80
42	Disorders of Pulmonary Function, Sleep, and the Upper Airway in Charcot-Marie-Tooth Disease. Lung, 2007, 185, 1-7.	3.3	80
43	Genotypeâ€“phenotype characteristics and baseline natural history of heritable neuropathies caused by mutations in theMPZgene. Brain, 2015, 138, 3180-3192.	7.6	80
44	Charcotâ€“Marieâ€“Tooth disease type 1. Brain, 2000, 123, 222-233.	7.6	78
45	Hereditary motor and sensory neuropathies: a biological perspective. Lancet Neurology, The, 2002, 1, 110-118.	10.2	74
46	MFN2 mutations in Charcotâ€“Marieâ€“Tooth disease alter mitochondria-associated ER membrane function but do not impair bioenergetics. Human Molecular Genetics, 2019, 28, 1782-1800.	2.9	72
47	Hereditary Motor and Sensory Neuropathies: An Overview of Clinical, Genetic, Electrophysiologic, and Pathologic Features. , 2005, , 1623-1658.		71
48	Quality-of-life in Charcotâ€“Marieâ€“Tooth disease: The patientâ€™s perspective. Neuromuscular Disorders, 2014, 24, 1018-1023.	0.6	71
49	Phenotypic Variability of Childhood Charcot-Marie-Tooth Disease. JAMA Neurology, 2016, 73, 645.	9.0	71
50	Targeted next-generation sequencing panels in the diagnosis of Charcot-Marie-Tooth disease. Neurology, 2020, 94, e51-e61.	1.1	71
51	Antibodies to GM1 and GD1b in patients with motor neuron disease without plasma cell dyscrasia. Annals of Neurology, 1989, 25, 511-513.	5.3	69
52	Different cellular and molecular mechanisms for early and late-onset myelin protein zero mutations. Human Molecular Genetics, 2008, 17, 1877-1889.	2.9	69
53	PMP22 expression in dermal nerve myelin from patients with CMT1A. Brain, 2009, 132, 1734-1740.	7.6	68
54	Charcot-Marie-Tooth Neuropathies: Diagnosis and Management. Seminars in Neurology, 2008, 28, 185-194.	1.4	67

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55	Hereditary motor and sensory neuropathies: Understanding molecular pathogenesis could lead to future treatment strategies. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2015, 1852, 667-678.	3.8	67
56	Absence of P0 leads to the dysregulation of myelin gene expression and myelin morphogenesis. Journal of Neuroscience Research, 2000, 60, 714-724.	2.9	66
57	Loss-of-function phenotype of hereditary neuropathy with liability to pressure palsies. Muscle and Nerve, 2004, 29, 205-210.	2.2	62
58	Inherited neuropathies: Clinical overview and update. Muscle and Nerve, 2013, 48, 604-622.	2.2	62
59	MpzR98C arrests Schwann cell development in a mouse model of early-onset Charcotâ€“Marieâ€“Tooth disease type 1B. Brain, 2012, 135, 2032-2047.	7.6	61
60	Functional Magnetic Resonance Imaging during Urodynamic Testing Identifies Brain Structures Initiating Micturition. Journal of Urology, 2014, 192, 1149-1154.	0.4	61
61	Psychometrics evaluation of Charcotâ€Marieâ€Tooth Neuropathy Score (<scp>CMTNSv2</scp>) second version, using Rasch analysis. Journal of the Peripheral Nervous System, 2014, 19, 192-196.	3.1	59
62	Mutation screen reveals novel variants and expands the phenotypes associated with DYNC1H1. Journal of Neurology, 2015, 262, 2124-2134.	3.6	59
63	Mutations in ATP1A1 Cause Dominant Charcot-Marie-Tooth Type 2. American Journal of Human Genetics, 2018, 102, 505-514.	6.2	59
64	T118M PMP22 mutation causes partial loss of function and HNPPâ€like neuropathy. Annals of Neurology, 2006, 59, 358-364.	5.3	58
65	Schwann cell expression of PLP1 but not DM20 is necessary to prevent neuropathy. Annals of Neurology, 2003, 53, 354-365.	5.3	55
66	Demyelinating CMTâ€“whatâ€™s known, whatâ€™s new and whatâ€™s in store?. Neuroscience Letters, 2015, 596, 14-26.	2.1	54
67	Induced Effects of Backgrounds and Foregrounds in Three-Dimensional Configurations: The Role of T-Junctions. Perception, 1997, 26, 395-408.	1.2	53
68	Peripheral neuropathy in complex inherited diseases: an approach to diagnosis. Journal of Neurology, Neurosurgery and Psychiatry, 2017, 88, 846-863.	1.9	51
69	Phase 2 Trial of Rituximab in Acetylcholine Receptor Antibody-Positive Generalized Myasthenia Gravis. Neurology, 2022, 98, .	1.1	51
70	Protein Zero Is Necessary for E-Cadherin-Mediated Adherens Junction Formation in Schwann Cells. Molecular and Cellular Neurosciences, 2001, 18, 606-618.	2.2	50
71	Regulation of Oleoylâ€CoA Synthesis in the Peripheral Nervous System: Demonstration of a Link with Myelin Synthesis. Journal of Neurochemistry, 1998, 71, 1719-1726.	3.9	50
72	Natural history of Charcotâ€Marieâ€Tooth disease during childhood. Annals of Neurology, 2017, 82, 353-359.	5.3	50

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73	Major myelin protein gene (P0) mutation causes a novel form of axonal degeneration. Journal of Comparative Neurology, 2006, 498, 252-265.	1.6	49
74	Conduction Block in PMP22 Deficiency. Journal of Neuroscience, 2010, 30, 600-608.	3.6	49
75	An adenoviral vector can transfer lacZ expression into schwann cells in culture and in sciatic nerve. Annals of Neurology, 1995, 38, 429-436.	5.3	48
76	Persistent CNS dysfunction in a boy with CMT1X. Journal of the Neurological Sciences, 2009, 279, 109-113.	0.6	48
77	The relationship between anogenital distance and azoospermia in adult men. Journal of Developmental and Physical Disabilities, 2012, 35, 726-730.	3.6	48
78	Motor unit number estimate of distal and proximal muscles in Charcot-Marie-Tooth disease. Muscle and Nerve, 2003, 28, 161-167.	2.2	47
79	SCO2 mutations cause early-onset axonal Charcot-Marie-Tooth disease associated with cellular copper deficiency. Brain, 2018, 141, 662-672.	7.6	46
80	Neuropathy. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2018, 148, 653-665.	1.8	45
81	Update on Charcotâ€“Marieâ€“Tooth disease. Current Opinion in Neurology, 2015, 28, 462-467.	3.6	44
82	Cross-sectional analysis of a large cohort with X-linked Charcot-Marie-Tooth disease (CMTX1). Neurology, 2017, 89, 927-935.	1.1	44
83	Validation of MRC Centre MRI calf muscle fat fraction protocol as an outcome measure in CMT1A. Neurology, 2018, 91, e1125-e1129.	1.1	43
84	Peripheral Neuropathy Caused by Proteolipid Protein Gene Mutations. Annals of the New York Academy of Sciences, 1999, 883, 351-365.	3.8	42
85	Phenotype expression in women with CMT1X. Journal of the Peripheral Nervous System, 2011, 16, 102-107.	3.1	42
86	Functional Magnetic Resonance Imaging with Concurrent Urodynamic Testing Identifies Brain Structures Involved in Micturition Cycle in Patients with Multiple Sclerosis. Journal of Urology, 2017, 197, 438-444.	0.4	42
87	Novel mutations in dystonin provide clues to the pathomechanisms of HSAN-VI. Neurology, 2017, 88, 2132-2140.	1.1	41
88	Effect of an R69C Mutation in the Myelin Protein Zero Gene on Myelination and Ion Channel Subtypes. Archives of Neurology, 2006, 63, 1787.	4.5	40
89	Dynein mutations associated with hereditary motor neuropathies impair mitochondrial morphology and function with age. Neurobiology of Disease, 2013, 58, 220-230.	4.4	40
90	Absence of Dystrophin Related Protein-2 disrupts Cajal bands in a patient with Charcotâ€“Marieâ€“Tooth disease. Neuromuscular Disorders, 2015, 25, 786-793.	0.6	40

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91	Myelin protein zero mutations and the unfolded protein response in Charcot Marie Tooth disease type 1B. Annals of Clinical and Translational Neurology, 2018, 5, 445-455.	3.7	39
92	Prevalence and orthopedic management of foot and ankle deformities in Charcotâ€“Marieâ€“Tooth disease. Muscle and Nerve, 2018, 57, 255-259.	2.2	39
93	Natural history of Charcot-Marie-Tooth disease type 2A: a large international multicentre study. Brain, 2020, 143, 3589-3602.	7.6	39
94	Regulation of Insulin-Like Growth Factor-Binding Protein-5 Expression during Schwann Cell Differentiation1. Endocrinology, 1999, 140, 4478-4485.	2.8	38
95	Correlation between clinical/neurophysiological findings and quality of life in Charcotâ€Marieâ€Tooth type 1A. Journal of the Peripheral Nervous System, 2008, 13, 64-70.	3.1	37
96	Dispersion of compound muscle action potential in hereditary neuropathies and chronic inflammatory demyelinating polyneuropathy. Muscle and Nerve, 2006, 34, 417-422.	2.2	36
97	Stoichiometric Alteration of PMP22 Protein Determines the Phenotype of Hereditary Neuropathy With Liability to Pressure Palsies. Archives of Neurology, 2007, 64, 974.	4.5	35
98	A multicenter retrospective study of charcotâ€marieâ€tooth disease type 4B (CMT4B) associated with mutations in myotubularinâ€related proteins (MTMRs). Annals of Neurology, 2019, 86, 55-67.	5.3	35
99	Diabetes mellitus exacerbates motor and sensory impairment in CMT1A. Journal of the Peripheral Nervous System, 2008, 13, 299-304.	3.1	34
100	Distinct pathogenic processes between Fig4-deficient motor and sensory neurons. European Journal of Neuroscience, 2011, 33, 1401-1410.	2.6	34
101	Genetic and clinical characteristics ofNEFL-related Charcot-Marie-Tooth disease. Journal of Neurology, Neurosurgery and Psychiatry, 2017, 88, 575-585.	1.9	34
102	Burst mitofusin activation reverses neuromuscular dysfunction in murine CMT2A. ELife, 2020, 9, .	6.0	34
103	Charcot-Marie-Tooth disease. Neurology, 2015, 85, 1202-1208.	1.1	33
104	Variation in SIPA1L2 is correlated with phenotype modification in Charcotâ€“ Marieâ€“ Tooth disease type 1A. Annals of Neurology, 2019, 85, 316-330.	5.3	33
105	Charcot-Marie-Tooth disease impairs quality of life: Why? And how do we improve it?. Neurology, 2005, 65, 790-791.	1.1	31
106	Small nerve fiber involvement in CMT1A. Neurology, 2015, 84, 407-414.	1.1	30
107	Substrate interaction defects in histidyl-tRNA synthetase linked to dominant axonal peripheral neuropathy. Human Mutation, 2018, 39, 415-432.	2.5	30
108	Regulating PMP22 expression as a dosage sensitive neuropathy gene. Brain Research, 2020, 1726, 146491.	2.2	30

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109	A longitudinal study of CMT1A using Rasch analysis based CMT neuropathy and examination scores. Neurology, 2020, 94, e884-e896.	1.1	29
110	Therapeutic strategies for the inherited neuropathies. NeuroMolecular Medicine, 2006, 8, 255-278.	3.4	28
111	Myelin abnormality in Charcotâ€“Marieâ€“Tooth type 4J recapitulates features of acquired demyelination. Annals of Neurology, 2018, 83, 756-770.	5.3	28
112	Mutations in BAG3 cause adult-onset Charcot-Marie-Tooth disease. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 313-315.	1.9	28
113	Correlation between Weakness and Axonal Loss in Patients with CMT1A. Annals of the New York Academy of Sciences, 1999, 883, 490-492.	3.8	26
114	Electrodiagnostic Findings in CMTX: A Disorder of the Schwann Cell and Peripheral Nerve Myelin. Annals of the New York Academy of Sciences, 1999, 883, 504-507.	3.8	26
115	Selected items from the Charcot-Marie-Tooth (CMT) Neuropathy Score and secondary clinical outcome measures serve as sensitive clinical markers of disease severity in CMT1A patients. Neuromuscular Disorders, 2014, 24, 1003-1017.	0.6	25
116	Development and validation of the Charcot-Marie-Tooth Disease Infant Scale. Brain, 2018, 141, 3319-3330.	7.6	25
117	The Charcot-Marie-Tooth Functional Outcome Measure (CMT-FOM). Neurology, 2018, 91, e1381-e1384.	1.1	25
118	Schwann cell transcript biomarkers for hereditary neuropathy skin biopsies. Annals of Neurology, 2019, 85, 887-898.	5.3	25
119	Transmembrane protease serine 5: a novel Schwann cell plasma marker for CMT1A. Annals of Clinical and Translational Neurology, 2020, 7, 69-82.	3.7	25
120	Current therapy for Charcot-Marie-Tooth disease. Current Treatment Options in Neurology, 2005, 7, 23-31.	1.8	24
121	Copy number variations are a rare cause of non-CMT1A Charcot-Marie-Tooth disease. Journal of Neurology, 2010, 257, 735-741.	3.6	24
122	The <scp>C</scp>harcotâ€“<scp>M</scp>arieâ€“<scp>T</scp>ooth <scp>H</scp>ealth <scp>I</scp>ndex: Evaluation of a Patientâ€Reported Outcome. Annals of Neurology, 2018, 84, 225-233.	5.3	24
123	Proteolipid protein mRNA stability is regulated by axonal contact in the rodent peripheral nervous system. Journal of Neurobiology, 2000, 44, 7-19.	3.6	23
124	Myelin protein zero/P0 phosphorylation and function require an adaptor protein linking it to RACK1 and PKCÎ±. Journal of Cell Biology, 2007, 177, 707-716.	5.2	22
125	X inactivation in females with X-linked Charcotâ€“Marieâ€“Tooth disease. Neuromuscular Disorders, 2012, 22, 617-621.	0.6	22
126	Balance impairment in pediatric charcotâ€“marieâ€“tooth disease. Muscle and Nerve, 2019, 60, 242-249.	2.2	22

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127	Anti-GM1/GD1b M-proteins damage human spinal cord neurons co-cultured with muscle. Journal of the Neurological Sciences, 1993, 120, 38-45.	0.6	21
128	Developmental expression of P0 mRNA and P0 protein in the sciatic nerve and the spinal nerve roots of the rat. Journal of Neurocytology, 1994, 23, 249-257.	1.5	21
129	Haplotype-specific modulation of a SOX10/CREB response element at the Charcotâ€“Marieâ€“Tooth disease type 4C locus SH3TC2. Human Molecular Genetics, 2014, 23, 5171-5187.	2.9	21
130	Reduced neurofilament expression in cutaneous nerve fibers of patients with CMT2E. Neurology, 2015, 85, 228-234.	1.1	21
131	Satisfaction with ankle foot orthoses in individuals with <scp>Charcotâ€Marieâ€Tooth disease</scp>. Muscle and Nerve, 2021, 63, 40-45.	2.2	21
132	Neuropathy in a Human Without the PMP22 Gene. Archives of Neurology, 2011, 68, 814-21.	4.5	21
133	In vivo confocal microscopy of Meissner corpuscles as a novel sensory measure in CMT1A. Journal of the Peripheral Nervous System, 2011, 16, 169-174.	3.1	20
134	Gain of glycosylation: A new pathomechanism of myelin protein zero mutations. Annals of Neurology, 2012, 71, 427-431.	5.3	20
135	Biomarkers predict outcome in Charcot-Marie-Tooth disease 1A. Journal of Neurology, Neurosurgery and Psychiatry, 2017, 88, 941-952.	1.9	20
136	Characterization of oligosaccharides that bind to human anti-MAG antibodies and to the mouse monoclonal antibody HNK-1. Journal of Neuroimmunology, 1986, 12, 291-298.	2.3	19
137	Genetic testing in neuromuscular disease. Neurologic Clinics, 2004, 22, 481-508.	1.8	19
138	Genetics of Neuropathies. Seminars in Neurology, 2011, 31, 494-505.	1.4	19
139	Phenotypic presentation of the Ser63Del MPZ mutation. Journal of the Peripheral Nervous System, 2012, 17, 197-200.	3.1	19
140	A case of neuromyotonia and axonal motor neuropathy: A report of a HINT1 mutation in the United States. Muscle and Nerve, 2015, 52, 1110-1113.	2.2	19
141	Modifier Gene Candidates in Charcot-Marie-Tooth Disease Type 1A: A Case-Only Genome-Wide Association Study. Journal of Neuromuscular Diseases, 2019, 6, 201-211.	2.6	19
142	Fatty acid synthase expression during peripheral nervous system myelination. Molecular Brain Research, 2002, 101, 52-58.	2.3	18
143	Symmetry of foot alignment and ankle flexibility in paediatric Charcotâ€“Marieâ€“Tooth disease. Clinical Biomechanics, 2012, 27, 744-747.	1.2	18
144	Objective Evaluation of Overactive Bladder: Which Surveys Should I Use?. Current Bladder Dysfunction Reports, 2013, 8, 45-50.	0.5	18

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145	Prospective study of muscle cramps in Charcotâ€Marieâ€Tooth disease. Muscle and Nerve, 2015, 51, 485-488.	2.2	18
146	Regulation of Myelin-Specific Gene Expression: Relevance to CMT1. Annals of the New York Academy of Sciences, 1999, 883, 91-108.	3.8	17
147	Anterior tibialis cmap amplitude correlations with impairment in CMT1A. Muscle and Nerve, 2013, 47, 493-496.	2.2	17
148	Effect of pain in pediatric inherited neuropathies. Neurology, 2014, 82, 793-797.	1.1	17
149	Inclusion body myositis and sarcoid myopathy: Coincidental occurrence or associated diseases. Neuromuscular Disorders, 2015, 25, 297-300.	0.6	17
150	Modulation of Cell-Mediated Immunity Prolongs Adenovirus-Mediated Transgene Expression in Sciatic Nerve. Human Gene Therapy, 1999, 10, 787-800.	2.7	16
151	Rare Manifestation of a c.290 C>T, p.Gly97GluVCPMutation. Case Reports in Genetics, 2015, 2015, 1-5.	0.2	16
152	A longitudinal and crossâ€sectional study of plasma neurofilament light chain concentration in <scp>Charcotâ€Marieâ€Tooth</scp> disease. Journal of the Peripheral Nervous System, 2022, 27, 50-57.	3.1	16
153	Charcot-Marie-Tooth Disease and Related Genetic Neuropathies. CONTINUUM Lifelong Learning in Neurology, 2012, 18, 39-59.	0.8	15
154	Transitioning outcome measures: relationship between the CMTPedS and CMTNSv2 in children, adolescents, and young adults with Charcotâ€Marieâ€Tooth disease. Journal of the Peripheral Nervous System, 2013, 18, 177-180.	3.1	15
155	Assessing non-Mendelian inheritance in inherited axonopathies. Genetics in Medicine, 2020, 22, 2114-2119.	2.4	15
156	Loss of function <scp>MPZ</scp> mutation causes milder <scp>CMT1B</scp> neuropathy. Journal of the Peripheral Nervous System, 2021, 26, 177-183.	3.1	15
157	Plasticity of Adenylyl Cyclase-Related Signaling Sequelae after Long-Term Morphine Treatment. Molecular Pharmacology, 2008, 73, 868-879.	2.3	14
158	Rydel-Seiffer fork revisited: Beyond a simple case of black and white. Neurology, 2016, 87, 738-740.	1.1	14
159	Carpal tunnel syndrome in inherited neuropathies: A retrospective survey. Muscle and Nerve, 2018, 57, 388-394.	2.2	14
160	MicroRNAs as Biomarkers of Charcot-Marie-Tooth Disease Type 1A. Neurology, 2021, 97, e489-e500.	1.1	14
161	Treatment with IFB-088 Improves Neuropathy in CMT1A and CMT1B Mice. Molecular Neurobiology, 2022, 59, 4159-4178.	4.0	14
162	Unfolded protein response, treatment and CMT1B. Rare Diseases (Austin, Tex), 2013, 1, e24049.	1.8	13

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163	Variant pathogenicity evaluation in the community-driven Inherited Neuropathy Variant Browser. Human Mutation, 2018, 39, 635-642.	2.5	13
164	Development and Validation of the Pediatric Charcotâ€“Marieâ€“Tooth Disease Quality of Life Outcome Measure. Annals of Neurology, 2021, 89, 369-379.	5.3	13
165	Appearance of PLP mRNA in Specific Regions of the Developing Rat Lumbosacral Spinal Cord as Revealed by in Situ Hybridization. Experimental Neurology, 1993, 121, 139-147.	4.1	12
166	Elevated expression of messenger RNA for peripheral myelin protein 22 in biopsied peripheral nerves of patients with C Harcot-Marie-Tooth disease type 1A. Annals of Neurology, 1994, 36, 451-452.	5.3	12
167	Charcotâ€“Marieâ€“Tooth Disease type 4C: Novel mutations, clinical presentations, and diagnostic challenges. Muscle and Nerve, 2018, 57, 749-755.	2.2	12
168	The audiologic profile of patients with Charcot-Marie Tooth neuropathy can be characterised by both cochlear and neural deficits. International Journal of Audiology, 2019, 58, 902-912.	1.7	12
169	A recurrent GARS mutation causes distal hereditary motor neuropathy. Journal of the Peripheral Nervous System, 2019, 24, 320-323.	3.1	12
170	A nonsense mutation in myelin protein zero causes congenital hypomyelination neuropathy through altered P0 membrane targeting and gain of abnormal function. Human Molecular Genetics, 2019, 28, 124-132.	2.9	12
171	A molecular basis for hereditary motor and sensory neuropathy disorders. Current Neurology and Neuroscience Reports, 2001, 1, 77-88.	4.2	11
172	Regulation of Insulin-Like Growth Factor-Binding Protein-5 Expression during Schwann Cell Differentiation. Endocrinology, 1999, 140, 4478-4485.	2.8	11
173	A neuropathyâ€associated kinesin KIF1A mutation hyperâ€stabilizes the motorâ€neck interaction during the ATPase cycle. EMBO Journal, 2022, 41, e108899.	7.8	11
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