Michael E Shy

List of Publications by Year
in descending order

238	11,891	28736	40945
papers	citations	57 h-index	97 g-index

245	245	245	9738
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Charcot-Marie-Tooth neuropathy score and ambulation index are both predictors of orthotic need for patients with CMT. Neurological Sciences, 2022, 43, 2759-2764.	0.9	2
2	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.	1.4	16
3	Phase 2 Trial of Rituximab in Acetylcholine Receptor Antibody-Positive Generalized Myasthenia Gravis. Neurology, 2022, 98, .	1.5	51
4	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
5	A neuropathyâ€associated kinesin KIF1A mutation hyperâ€stabilizes the motorâ€neck interaction during the ATPase cycle. EMBO Journal, 2022, 41, e108899.	3.5	11
6	Mitochondrial Phenotypes in Genetically Diverse Neurodegenerative Diseases and Their Response to Mitofusin Activation. Cells, 2022, 11, 1053.	1.8	11
7	The Rare Disease Research Scholars Program: A training curriculum for clinical researchers with mixed methods evaluation study. Translational Science of Rare Diseases, 2022, 6, 1-11.	1.6	1
8	Validation of the Italian version of the pediatric <scp>CMT</scp> quality of life outcome measure. Journal of the Peripheral Nervous System, 2022, 27, 127-130.	1.4	3
9	Treatment with IFB-088 Improves Neuropathy in CMT1A and CMT1B Mice. Molecular Neurobiology, 2022, 59, 4159-4178.	1.9	14
10	Randomized Phase 2 Study of ACE-083 in Patients With Charcot-Marie-Tooth Disease. Neurology, 2022, 98, .	1.5	10
11	Satisfaction with ankle foot orthoses in individuals with <scp>Charcotâ€Marieâ€Tooth disease</scp>. Muscle and Nerve, 2021, 63, 40-45.	1.0	21
12	Development and Validation of the Pediatric Charcotâ€“Marieâ€“Tooth Disease Quality of Life Outcome Measure. Annals of Neurology, 2021, 89, 369-379.	2.8	13
13	A CADM3 variant causes Charcot-Marie-Tooth disease with marked upper limb involvement. Brain, 2021, 144, 1197-1213.	3.7	10
14	Loss of function <scp>MPZ</scp> mutation causes milder <scp>CMT1B</scp> neuropathy. Journal of the Peripheral Nervous System, 2021, 26, 177-183.	1.4	15
15	MicroRNAs as Biomarkers of Charcot-Marie-Tooth Disease Type 1A. Neurology, 2021, 97, e489-e500.	1.5	14
16	Association Between Body Mass Index and Disability in Children With Charcot-Marie-Tooth Disease. Neurology, 2021, 97, e1727-e1736.	1.5	2
17	Hereditary neuropathies: A pathological perspective. Journal of the Peripheral Nervous System, 2021, 26, S42-S60.	1.4	1
18	Regulating PMP22 expression as a dosage sensitive neuropathy gene. Brain Research, 2020, 1726, 146491.	1.1	30

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19	Incidence and Clinical Features of TRPV4-Linked Axonal Neuropathies in a USA Cohort of Charcotâ€“Marieâ€“Tooth Disease Type 2. NeuroMolecular Medicine, 2020, 22, 68-72.	1.8	8
20	Transmembrane protease serine 5: a novel Schwann cell plasma marker for CMT1A. Annals of Clinical and Translational Neurology, 2020, 7, 69-82.	1.7	25
21	Refining clinical trial inclusion criteria to optimize the standardized response mean of the CMTPedS. Annals of Clinical and Translational Neurology, 2020, 7, 1713-1715.	1.7	5
22	Assessing non-Mendelian inheritance in inherited axonopathies. Genetics in Medicine, 2020, 22, 2114-2119.	1.1	15
23	Reliability of the <scp>Charcotâ€Marieâ€Tooth</scp> functional outcome measure. Journal of the Peripheral Nervous System, 2020, 25, 288-291.	1.4	8
24	Biallelic mutations in SORD cause a common and potentially treatable hereditary neuropathy with implications for diabetes. Nature Genetics, 2020, 52, 473-481.	9.4	97
25	Validation of the Italian version of the <scp>Charcotâ€Marieâ€Tooth</scp> disease Pediatric Scale. Journal of the Peripheral Nervous System, 2020, 25, 138-142.	1.4	5
26	A longitudinal study of CMT1A using Rasch analysis based CMT neuropathy and examination scores. Neurology, 2020, 94, e884-e896.	1.5	29
27	Targeted next-generation sequencing panels in the diagnosis of Charcot-Marie-Tooth disease. Neurology, 2020, 94, e51-e61.	1.5	71
28	Natural history of Charcot-Marie-Tooth disease type 2A: a large international multicentre study. Brain, 2020, 143, 3589-3602.	3.7	39
29	Genetics and adult-onset chronic idiopathic axonal neuropathy. Neurology, 2020, 95, 1071-1073.	1.5	1
30	Burst mitofusin activation reverses neuromuscular dysfunction in murine CMT2A. ELife, 2020, 9, .	2.8	34
31	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.	0.9	12
32	A recurrent GARS mutation causes distal hereditary motor neuropathy. Journal of the Peripheral Nervous System, 2019, 24, 320-323.	1.4	12
33	Variation in SIPA1L2 is correlated with phenotype modification in Charcotâ€“ Marieâ€“ Tooth disease type 1A. Annals of Neurology, 2019, 85, 316-330.	2.8	33
34	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.	2.8	35
35	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.	1.1	19
36	Balance impairment in pediatric charcotâ€“marieâ€“tooth disease. Muscle and Nerve, 2019, 60, 242-249.	1.0	22

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37	Schwann cell transcript biomarkers for hereditary neuropathy skin biopsies. Annals of Neurology, 2019, 85, 887-898.	2.8	25
38	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.	1.4	72
39	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.	1.4	12
40	Variant pathogenicity evaluation in the community-driven Inherited Neuropathy Variant Browser. Human Mutation, 2018, 39, 635-642.	1.1	13
41	Neuropathy. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2018, 148, 653-665.	1.0	45
42	Myelin abnormality in Charcotâ€“Marieâ€“Tooth type 4J recapitulates features of acquired demyelination. Annals of Neurology, 2018, 83, 756-770.	2.8	28
43	Mutations in ATP1A1 Cause Dominant Charcot-Marie-Tooth Type 2. American Journal of Human Genetics, 2018, 102, 505-514.	2.6	59
44	SCO2 mutations cause early-onset axonal Charcot-Marie-Tooth disease associated with cellular copper deficiency. Brain, 2018, 141, 662-672.	3.7	46
45	Neurofilament light, biomarkers, and Charcot-Marie-Tooth disease. Neurology, 2018, 90, 257-259.	1.5	8
46	Asymmetric Ataxia, Depression, Memory Loss, Epilepsy, and Axonal Neuropathy Associated with A Heterozygous DNA Polymerase Gamma Variant of Uncertain Significance, c1370G>a (R457Q). Journal of Neuromuscular Diseases, 2018, 5, 99-104.	1.1	1
47	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.	1.7	39
48	Prevalence and orthopedic management of foot and ankle deformities in Charcotâ€“Marieâ€“Tooth disease. Muscle and Nerve, 2018, 57, 255-259.	1.0	39
49	Carpal tunnel syndrome in inherited neuropathies: A retrospective survey. Muscle and Nerve, 2018, 57, 388-394.	1.0	14
50	Substrate interaction defects in histidyl-tRNA synthetase linked to dominant axonal peripheral neuropathy. Human Mutation, 2018, 39, 415-432.	1.1	30
51	Mutations in BAG3 cause adult-onset Charcot-Marie-Tooth disease. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 313-315.	0.9	28
52	Development and validation of the Charcot-Marie-Tooth Disease Infant Scale. Brain, 2018, 141, 3319-3330.	3.7	25
53	The Charcot-Marie-Tooth Functional Outcome Measure (CMT-FOM). Neurology, 2018, 91, e1381-e1384.	1.5	25
54	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.	2.8	24

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55	Charcotâ€“Marieâ€“Tooth Disease type 4C: Novel mutations, clinical presentations, and diagnostic challenges. Muscle and Nerve, 2018, 57, 749-755.	1.0	12
56	Validation of MRC Centre MRI calf muscle fat fraction protocol as an outcome measure in CMT1A. Neurology, 2018, 91, e1125-e1129.	1.5	43
57	Reply: The classification of Charcot-Marie-Tooth diseases, a never-ending story: CMT4?. Brain, 2018, 141, e71-e71.	3.7	2
58	Charcotâ€“Marieâ€“Tooth disease type 1C: Clinical and electrophysiological findings for the c.334G>a (p.Gly112Ser) Litaf/Simple mutation. Muscle and Nerve, 2017, 56, 1092-1095.	1.0	8
59	Genetic and clinical characteristics ofNEFL-related Charcot-Marie-Tooth disease. Journal of Neurology, Neurosurgery and Psychiatry, 2017, 88, 575-585.	0.9	34
60	Hypomorphic mutations in POLR3A are a frequent cause of sporadic and recessive spastic ataxia. Brain, 2017, 140, 1561-1578.	3.7	85
61	Novel mutations in dystonin provide clues to the pathomechanisms of HSAN-VI. Neurology, 2017, 88, 2132-2140.	1.5	41
62	PMP22 exon 4 deletion causes ER retention of PMP22 and a gainâ€ofâ€function allele in CMT1E. Annals of Clinical and Translational Neurology, 2017, 4, 236-245.	1.7	6
63	221st ENMC International Workshop:. Neuromuscular Disorders, 2017, 27, 1138-1142.	0.3	10
64	Biomarkers predict outcome in Charcot-Marie-Tooth disease 1A. Journal of Neurology, Neurosurgery and Psychiatry, 2017, 88, 941-952.	0.9	20
65	Charcotâ€“Marieâ€“Tooth Disease Type 1A. Journal of Clinical Neurophysiology, 2017, 34, 508-511.	0.9	2
66	Peripheral neuropathy in complex inherited diseases: an approach to diagnosis. Journal of Neurology, Neurosurgery and Psychiatry, 2017, 88, 846-863.	0.9	51
67	Cross-sectional analysis of a large cohort with X-linked Charcot-Marie-Tooth disease (CMTX1). Neurology, 2017, 89, 927-935.	1.5	44
68	Natural history of Charcotâ€Marieâ€Tooth disease during childhood. Annals of Neurology, 2017, 82, 353-359.	2.8	50
69	A human cellular model to study peripheral myelination and demyelinating neuropathies. Brain, 2017, 140, 856-859.	3.7	2
70	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.	0.2	42
71	Genetic Peripheral Neuropathies. , 2017, , 1073-1080.		0
72	Antisense oligonucleotides offer hope to patients with Charcot-Marie-Tooth disease type 1A. Journal of Clinical Investigation, 2017, 128, 110-112.	3.9	9

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73	Phenotypic Variability of Childhood Charcot-Marie-Tooth Disease. JAMA Neurology, 2016, 73, 645.	4.5	71
74	Gene therapy, CMT1X, and the inherited neuropathies. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, 4552-4554.	3.3	0
75	Rydel-Seiffer fork revisited: Beyond a simple case of black and white. Neurology, 2016, 87, 738-740.	1.5	14
76	LRSAM1 lessons. Annals of Neurology, 2016, 80, 821-822.	2.8	1
77	Charcot-marie-tooth disease type 1X in women: Electrodiagnostic findings. Muscle and Nerve, 2016, 54, 728-732.	1.0	10
78	New and emerging treatments of Charcotâ€“Marieâ€“Tooth disease. Expert Opinion on Orphan Drugs, 2015, 3, 151-164.	0.5	0
79	Prospective study of muscle cramps in Charcotâ€Marieâ€Tooth disease. Muscle and Nerve, 2015, 51, 485-488.	1.0	18
80	Update on Charcotâ€“Marieâ€“Tooth disease. Current Opinion in Neurology, 2015, 28, 462-467.	1.8	44
81	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.	1.0	19
82	Coexistence of a T118M PMP22 missense mutation and chromosome 17 (17p11.2â€p12) deletion. Muscle and Nerve, 2015, 52, 905-908.	1.0	4
83	Progressive Lower Extremity Weakness and Axonal Sensorimotor Polyneuropathy from a Mutation inKIF5A(c.611G>A;p.Arg204Gln). Case Reports in Genetics, 2015, 2015, 1-5.	0.1	4
84	Rare Manifestation of a c.290 C>T, p.Gly97GluVCPMutation. Case Reports in Genetics, 2015, 2015, 1-5.	0.1	16
85	Reduced neurofilament expression in cutaneous nerve fibers of patients with CMT2E. Neurology, 2015, 85, 228-234.	1.5	21
86	Exome Sequence Analysis Suggests that Genetic Burden Contributes to Phenotypic Variability and Complex Neuropathy. Cell Reports, 2015, 12, 1169-1183.	2.9	211
87	Demyelinating CMTâ€“whatâ€™s known, whatâ€™s new and whatâ€™s in store?. Neuroscience Letters, 2015, 596, 14-26.	1.0	54
88	Inclusion body myositis and sarcoid myopathy: Coincidental occurrence or associated diseases. Neuromuscular Disorders, 2015, 25, 297-300.	0.3	17
89	Ultrasound: the future for evaluating the PNS in humans?. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, 362-362.	0.9	5
90	Small nerve fiber involvement in CMT1A. Neurology, 2015, 84, 407-414.	1.5	30

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91	Defining disability: development and validation of a mobility-Disability Severity Index (mDSI) in Charcot-Marie-tooth disease. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, 635-639.	0.9	7
92	Hereditary Neuropathies in Late Childhood and Adolescence. , 2015, , 319-339.		0
93	Mutation screen reveals novel variants and expands the phenotypes associated with DYNC1H1. Journal of Neurology, 2015, 262, 2124-2134.	1.8	59
94	Absence of Dystrophin Related Protein-2 disrupts Cajal bands in a patient with Charcotâ€“Marieâ€“Tooth disease. Neuromuscular Disorders, 2015, 25, 786-793.	0.3	40
95	Genotypeâ€“phenotype characteristics and baseline natural history of heritable neuropathies caused by mutations in theMPZgene. Brain, 2015, 138, 3180-3192.	3.7	80
96	Charcot-Marie-Tooth disease. Neurology, 2015, 85, 1202-1208.	1.5	33
97	Peripheral Neuropathies. , 2015, , 167-188.		1
98	Detection of Copy Number Variation by SNP-Allelotyping. Journal of Neurogenetics, 2015, 29, 4-7.	0.6	3
99	Axonal Charcotâ€“Marieâ€“Tooth disease patient-derived motor neurons demonstrate disease-specific phenotypes including abnormal electrophysiological properties. Experimental Neurology, 2015, 263, 190-199.	2.0	97
100	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.	1.8	67
101	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.	1.4	59
102	Effect of pain in pediatric inherited neuropathies. Neurology, 2014, 82, 793-797.	1.5	17
103	Genetic testing practices for Charcot-Marie-Tooth type 1A disease. Muscle and Nerve, 2014, 49, 478-482.	1.0	8
104	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.	1.4	21
105	Functional Magnetic Resonance Imaging during Urodynamic Testing Identifies Brain Structures Initiating Micturition. Journal of Urology, 2014, 192, 1149-1154.	0.2	61
106	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.	0.3	25
107	Quality-of-life in Charcotâ€“Marieâ€“Tooth disease: The patientâ€™s perspective. Neuromuscular Disorders, 2014, 24, 1018-1023.	0.3	71
108	A novel mutation in VCP causes Charcotâ€“Marieâ€“Tooth Type 2 disease. Brain, 2014, 137, 2897-2902.	3.7	116

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109	Inherited Peripheral Neuropathies. Neurologic Clinics, 2013, 31, 597-619.	0.8	88
110	Inherited neuropathies: Clinical overview and update. Muscle and Nerve, 2013, 48, 604-622.	1.0	62
111	Objective Evaluation of Overactive Bladder: Which Surveys Should I Use?. Current Bladder Dysfunction Reports, 2013, 8, 45-50.	0.2	18
112	Dynein mutations associated with hereditary motor neuropathies impair mitochondrial morphology and function with age. Neurobiology of Disease, 2013, 58, 220-230.	2.1	40
113	High-Dosage Ascorbic Acid Treatment in Charcot-Marie-Tooth Disease Type 1A. JAMA Neurology, 2013, 70, 981.	4.5	121
114	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.	1.4	15
115	Anterior tibialis cmap amplitude correlations with impairment in CMT1A. Muscle and Nerve, 2013, 47, 493-496.	1.0	17
116	Meeting Report: 2013 Peripheral Nerve Society Biennial Meeting, Saintâ€Malo, France, June 29â€“July 3, 2013. Journal of the Peripheral Nervous System, 2013, 18, 197-198.	1.4	1
117	Unfolded protein response, treatment and CMT1B. Rare Diseases (Austin, Tex), 2013, 1, e24049.	1.8	13
118	Lessons from London. Journal of Neurology, Neurosurgery and Psychiatry, 2012, 83, 767-768.	0.9	2
119	MpzR98C arrests Schwann cell development in a mouse model of early-onset Charcotâ€“Marieâ€“Tooth disease type 1B. Brain, 2012, 135, 2032-2047.	3.7	61
120	Charcot-Marie-Tooth Disease and Related Genetic Neuropathies. CONTINUUM Lifelong Learning in Neurology, 2012, 18, 39-59.	0.4	15
121	Peripheral Neuropathies. , 2012, , 2396-2409.		2
122	Curcumin derivatives promote Schwann cell differentiation and improve neuropathy in R98C CMT1B mice. Brain, 2012, 135, 3551-3566.	3.7	90
123	X inactivation in females with X-linked Charcotâ€“Marieâ€“Tooth disease. Neuromuscular Disorders, 2012, 22, 617-621.	0.3	22
124	Symmetry of foot alignment and ankle flexibility in paediatric Charcotâ€“Marieâ€“Tooth disease. Clinical Biomechanics, 2012, 27, 744-747.	0.5	18
125	Gain of glycosylation: A new pathomechanism of myelin protein zero mutations. Annals of Neurology, 2012, 71, 427-431.	2.8	20
126	Validation of the Charcotâ€“Marieâ€“Tooth disease pediatric scale as an outcome measure of disability. Annals of Neurology, 2012, 71, 642-652.	2.8	137

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127	The relationship between anogenital distance and azoospermia in adult men. Journal of Developmental and Physical Disabilities, 2012, 35, 726-730.	3.6	48
128	Phenotypic presentation of the Ser63Del MPZ mutation. Journal of the Peripheral Nervous System, 2012, 17, 197-200.	1.4	19
129	The relationship between anogenital distance and the efficacy of varicocele repair. BJU International, 2012, 110, E927-30.	1.3	10
130	Inherited Peripheral Neuropathies. CONTINUUM Lifelong Learning in Neurology, 2011, 17, 294-315.	0.4	5
131	Axonal Charcotâ€“Marieâ€“Tooth disease. Current Opinion in Neurology, 2011, 24, 475-483.	1.8	91
132	Distinct pathogenic processes between Fig4-deficient motor and sensory neurons. European Journal of Neuroscience, 2011, 33, 1401-1410.	1.2	34
133	Phenotype expression in women with CMT1X. Journal of the Peripheral Nervous System, 2011, 16, 102-107.	1.4	42
134	In vivo confocal microscopy of Meissner corpuscles as a novel sensory measure in CMT1A. Journal of the Peripheral Nervous System, 2011, 16, 169-174.	1.4	20
135	Reliability of the CMT neuropathy score (second version) in Charcotâ€Marieâ€Tooth disease. Journal of the Peripheral Nervous System, 2011, 16, 191-198.	1.4	269
136	Update on Charcot-Marie-Tooth Disease. Current Neurology and Neuroscience Reports, 2011, 11, 78-88.	2.0	128
137	Charcotâ€marieâ€tooth disease subtypes and genetic testing strategies. Annals of Neurology, 2011, 69, 22-33.	2.8	494
138	Exome sequencing allows for rapid gene identification in a Charcotâ€Marieâ€Tooth family. Annals of Neurology, 2011, 69, 464-470.	2.8	107
139	Genetics of Neuropathies. Seminars in Neurology, 2011, 31, 494-505.	0.5	19
140	Neuropathy in a Human Without the PMP22 Gene. Archives of Neurology, 2011, 68, 814-21.	4.9	21
141	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
142	Copy number variations are a rare cause of non-CMT1A Charcot-Marie-Tooth disease. Journal of Neurology, 2010, 257, 735-741.	1.8	24
143	Report from the Satellite Meeting of the Peripheral Nerve Society in Sydney, Australia, July 5-7, 2010. Journal of the Peripheral Nervous System, 2010, 15, 161-163.	1.4	0
144	Conduction Block in PMP22 Deficiency. Journal of Neuroscience, 2010, 30, 600-608.	1.7	49

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145	Reply: Internodal length variability of dermal myelinated fibres. Brain, 2010, 133, e143-e143.	3.7	2
146	PMP22 expression in dermal nerve myelin from patients with CMT1A. Brain, 2009, 132, 1734-1740.	3.7	68
147	Shortened internodal length of dermal myelinated nerve fibres in Charcot-Marie-Tooth disease type 1A. Brain, 2009, 132, 3263-3273.	3.7	85
148	Ascorbic acid for treatment of CMT1A: the jury is still out. Lancet Neurology, The, 2009, 8, 505-507.	4.9	6
149	Quality of life in children with CMT type 1A. Lancet Neurology, The, 2009, 8, 880-881.	4.9	10
150	Persistent CNS dysfunction in a boy with CMT1X. Journal of the Neurological Sciences, 2009, 279, 109-113.	0.3	48
151	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
152	Biology of Peripheral Inherited Neuropathies: Schwann Cell Axonal Interactions. Advances in Experimental Medicine and Biology, 2009, 652, 171-181.	0.8	9
153	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.	1.4	37
154	Diabetes mellitus exacerbates motor and sensory impairment in CMT1A. Journal of the Peripheral Nervous System, 2008, 13, 299-304.	1.4	34
155	Obstructive sleep apnoea and CMT1A: answers and more questions. Journal of Neurology, Neurosurgery and Psychiatry, 2008, 79, 743-744.	0.9	3
156	Mutation of FIG4 causes a rapidly progressive, asymmetric neuronal degeneration. Brain, 2008, 131, 1990-2001.	3.7	128
157	Hereditary motor neuropathy and heat shock proteins. Neurology, 2008, 71, 1656-1657.	1.5	6
158	Different cellular and molecular mechanisms for early and late-onset myelin protein zero mutations. Human Molecular Genetics, 2008, 17, 1877-1889.	1.4	69
159	Plasticity of Adenylyl Cyclase-Related Signaling Sequelae after Long-Term Morphine Treatment. Molecular Pharmacology, 2008, 73, 868-879.	1.0	14
160	Charcot-Marie-Tooth Neuropathies: Diagnosis and Management. Seminars in Neurology, 2008, 28, 185-194.	0.5	67
161	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.	2.3	22
162	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

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163	Women and men are equally disabled by Charcot-Marie-Tooth disease type 1A. Neurology, 2007, 68, 873-873.	1.5	8
164	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.	2.6	116
165	Myelin Protein Zero and CMT1B: A Tale of Two Phenotypes. , 2007, , 463-474.		0
166	Mutation of FIG4 causes neurodegeneration in the pale tremor mouse and patients with CMT4J. Nature, 2007, 448, 68-72.	13.7	438
167	Disorders of Pulmonary Function, Sleep, and the Upper Airway in Charcot-Marie-Tooth Disease. Lung, 2007, 185, 1-7.	1.4	80
168	Therapeutic strategies for the inherited neuropathies. NeuroMolecular Medicine, 2006, 8, 255-278.	1.8	28
169	Peripheral neuropathies caused by mutations in the myelin protein zero. Journal of the Neurological Sciences, 2006, 242, 55-66.	0.3	80
170	Effect of an R69C Mutation in the Myelin Protein Zero Gene on Myelination and Ion Channel Subtypes. Archives of Neurology, 2006, 63, 1787.	4.9	40
171	Major myelin protein gene (P0) mutation causes a novel form of axonal degeneration. Journal of Comparative Neurology, 2006, 498, 252-265.	0.9	49
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