

Michael E Shy

List of Publications by Year in descending order

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Version: 2024-02-01

238
papers

11,891
citations

24978

57
h-index

35952

97
g-index

245
all docs

245
docs citations

245
times ranked

8983
citing authors

#	ARTICLE	IF	CITATIONS
1	Charcot-Marie-Tooth neuropathy score and ambulation index are both predictors of orthotic need for patients with CMT. <i>Neurological Sciences</i> , 2022, 43, 2759-2764.	0.9	2
2	A longitudinal and cross-sectional study of plasma neurofilament light chain concentration in Charcot-Marie-Tooth disease. <i>Journal of the Peripheral Nervous System</i> , 2022, 27, 50-57.	1.4	16
3	Phase 2 Trial of Rituximab in Acetylcholine Receptor Antibody-Positive Generalized Myasthenia Gravis. <i>Neurology</i> , 2022, 98, .	1.5	51
4	Clinical practice guideline for the management of paediatric Charcot-Marie-Tooth disease. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2022, 93, 530-538.	0.9	10
5	A neuropathy-associated kinesin KIF1A mutation hyperstabilizes the motor-neck interaction during the ATPase cycle. <i>EMBO Journal</i> , 2022, 41, e108899.	3.5	11
6	Mitochondrial Phenotypes in Genetically Diverse Neurodegenerative Diseases and Their Response to Mitofusin Activation. <i>Cells</i> , 2022, 11, 1053.	1.8	11
7	The Rare Disease Research Scholars Program: A training curriculum for clinical researchers with mixed methods evaluation study. <i>Translational Science of Rare Diseases</i> , 2022, 6, 1-11.	1.6	1
8	Validation of the Italian version of the pediatric CMT quality of life outcome measure. <i>Journal of the Peripheral Nervous System</i> , 2022, 27, 127-130.	1.4	3
9	Treatment with IFB-088 Improves Neuropathy in CMT1A and CMT1B Mice. <i>Molecular Neurobiology</i> , 2022, 59, 4159-4178.	1.9	14
10	Randomized Phase 2 Study of ACE-083 in Patients With Charcot-Marie-Tooth Disease. <i>Neurology</i> , 2022, 98, .	1.5	10
11	Satisfaction with ankle foot orthoses in individuals with Charcot-Marie-Tooth disease. <i>Muscle and Nerve</i> , 2021, 63, 40-45.	1.0	21
12	Development and Validation of the Pediatric Charcot-Marie-Tooth Disease Quality of Life Outcome Measure. <i>Annals of Neurology</i> , 2021, 89, 369-379.	2.8	13
13	A <i>CADM3</i> variant causes Charcot-Marie-Tooth disease with marked upper limb involvement. <i>Brain</i> , 2021, 144, 1197-1213.	3.7	10
14	Loss of function MPZ mutation causes milder CMT1B neuropathy. <i>Journal of the Peripheral Nervous System</i> , 2021, 26, 177-183.	1.4	15
15	MicroRNAs as Biomarkers of Charcot-Marie-Tooth Disease Type 1A. <i>Neurology</i> , 2021, 97, e489-e500.	1.5	14
16	Association Between Body Mass Index and Disability in Children With Charcot-Marie-Tooth Disease. <i>Neurology</i> , 2021, 97, e1727-e1736.	1.5	2
17	Hereditary neuropathies: A pathological perspective. <i>Journal of the Peripheral Nervous System</i> , 2021, 26, S42-S60.	1.4	1
18	Regulating PMP22 expression as a dosage sensitive neuropathy gene. <i>Brain Research</i> , 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. <i>NeuroMolecular Medicine</i> , 2020, 22, 68-72.	1.8	8
20	Transmembrane protease serine 5: a novel Schwann cell plasma marker for CMT1A. <i>Annals of Clinical and Translational Neurology</i> , 2020, 7, 69-82.	1.7	25
21	Refining clinical trial inclusion criteria to optimize the standardized response mean of the CMTPedS. <i>Annals of Clinical and Translational Neurology</i> , 2020, 7, 1713-1715.	1.7	5
22	Assessing non-Mendelian inheritance in inherited axonopathies. <i>Genetics in Medicine</i> , 2020, 22, 2114-2119.	1.1	15
23	Reliability of the <scp>Charcotâ€“Marieâ€“Tooth</scp> functional outcome measure. <i>Journal of the Peripheral Nervous System</i> , 2020, 25, 288-291.	1.4	8
24	Biallelic mutations in SORD cause a common and potentially treatable hereditary neuropathy with implications for diabetes. <i>Nature Genetics</i> , 2020, 52, 473-481.	9.4	97
25	Validation of the Italian version of the <scp>Charcotâ€“Marieâ€“Tooth</scp> disease Pediatric Scale. <i>Journal of the Peripheral Nervous System</i> , 2020, 25, 138-142.	1.4	5
26	A longitudinal study of CMT1A using Rasch analysis based CMT neuropathy and examination scores. <i>Neurology</i> , 2020, 94, e884-e896.	1.5	29
27	Targeted next-generation sequencing panels in the diagnosis of Charcot-Marie-Tooth disease. <i>Neurology</i> , 2020, 94, e51-e61.	1.5	71
28	Natural history of Charcot-Marie-Tooth disease type 2A: a large international multicentre study. <i>Brain</i> , 2020, 143, 3589-3602.	3.7	39
29	Genetics and adult-onset chronic idiopathic axonal neuropathy. <i>Neurology</i> , 2020, 95, 1071-1073.	1.5	1
30	Burst mitofusin activation reverses neuromuscular dysfunction in murine CMT2A. <i>ELife</i> , 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. <i>International Journal of Audiology</i> , 2019, 58, 902-912.	0.9	12
32	A recurrent GARS mutation causes distal hereditary motor neuropathy. <i>Journal of the Peripheral Nervous System</i> , 2019, 24, 320-323.	1.4	12
33	Variation in <i>SIPA1L2</i> is correlated with phenotype modification in Charcotâ€“ Marieâ€“ Tooth disease type 1A. <i>Annals of Neurology</i> , 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). <i>Annals of Neurology</i> , 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. <i>Journal of Neuromuscular Diseases</i> , 2019, 6, 201-211.	1.1	19
36	Balance impairment in pediatric charcotâ€“marieâ€“tooth disease. <i>Muscle and Nerve</i> , 2019, 60, 242-249.	1.0	22

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37	Schwann cell transcript biomarkers for hereditary neuropathy skin biopsies. <i>Annals of Neurology</i> , 2019, 85, 887-898.	2.8	25
38	MFN2 mutations in Charcot-Marie-Tooth disease alter mitochondria-associated ER membrane function but do not impair bioenergetics. <i>Human Molecular Genetics</i> , 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. <i>Human Molecular Genetics</i> , 2019, 28, 124-132.	1.4	12
40	Variant pathogenicity evaluation in the community-driven Inherited Neuropathy Variant Browser. <i>Human Mutation</i> , 2018, 39, 635-642.	1.1	13
41	Neuropathy. <i>Handbook of Clinical Neurology</i> / Edited By P J Vinken and G W Bruyn, 2018, 148, 653-665.	1.0	45
42	Myelin abnormality in Charcot-Marie-Tooth type 4J recapitulates features of acquired demyelination. <i>Annals of Neurology</i> , 2018, 83, 756-770.	2.8	28
43	Mutations in ATP1A1 Cause Dominant Charcot-Marie-Tooth Type 2. <i>American Journal of Human Genetics</i> , 2018, 102, 505-514.	2.6	59
44	SCO2 mutations cause early-onset axonal Charcot-Marie-Tooth disease associated with cellular copper deficiency. <i>Brain</i> , 2018, 141, 662-672.	3.7	46
45	Neurofilament light, biomarkers, and Charcot-Marie-Tooth disease. <i>Neurology</i> , 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). <i>Journal of Neuromuscular Diseases</i> , 2018, 5, 99-104.	1.1	1
47	Myelin protein zero mutations and the unfolded protein response in Charcot Marie Tooth disease type 1B. <i>Annals of Clinical and Translational Neurology</i> , 2018, 5, 445-455.	1.7	39
48	Prevalence and orthopedic management of foot and ankle deformities in Charcot-Marie-Tooth disease. <i>Muscle and Nerve</i> , 2018, 57, 255-259.	1.0	39
49	Carpal tunnel syndrome in inherited neuropathies: A retrospective survey. <i>Muscle and Nerve</i> , 2018, 57, 388-394.	1.0	14
50	Substrate interaction defects in histidyl-tRNA synthetase linked to dominant axonal peripheral neuropathy. <i>Human Mutation</i> , 2018, 39, 415-432.	1.1	30
51	Mutations in BAC3 cause adult-onset Charcot-Marie-Tooth disease. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2018, 89, 313-315.	0.9	28
52	Development and validation of the Charcot-Marie-Tooth Disease Infant Scale. <i>Brain</i> , 2018, 141, 3319-3330.	3.7	25
53	The Charcot-Marie-Tooth Functional Outcome Measure (CMT-FOM). <i>Neurology</i> , 2018, 91, e1381-e1384.	1.5	25
54	The <sc>C</sc>harcot-Marie-Tooth <sc>M</sc>arie-Tooth <sc>T</sc>ooth <sc>H</sc>ealth <sc>I</sc>ndex: Evaluation of a Patient-Reported Outcome. <i>Annals of Neurology</i> , 2018, 84, 225-233.	2.8	24

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55	Charcot-Marie-Tooth Disease type 4C: Novel mutations, clinical presentations, and diagnostic challenges. <i>Muscle and Nerve</i> , 2018, 57, 749-755.	1.0	12
56	Validation of MRC Centre MRI calf muscle fat fraction protocol as an outcome measure in CMT1A. <i>Neurology</i> , 2018, 91, e1125-e1129.	1.5	43
57	Reply: The classification of Charcot-Marie-Tooth diseases, a never-ending story: CMT4?. <i>Brain</i> , 2018, 141, e71-e71.	3.7	2
58	Charcot-Marie-Tooth disease type 1C: Clinical and electrophysiological findings for the c.334G>a (p.Gly112Ser) mutation. <i>Muscle and Nerve</i> , 2017, 56, 1092-1095.	1.0	8
59	Genetic and clinical characteristics of NEFL-related Charcot-Marie-Tooth disease. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2017, 88, 575-585.	0.9	34
60	Hypomorphic mutations in POLR3A are a frequent cause of sporadic and recessive spastic ataxia. <i>Brain</i> , 2017, 140, 1561-1578.	3.7	85
61	Novel mutations in dystonin provide clues to the pathomechanisms of HSAN-VI. <i>Neurology</i> , 2017, 88, 2132-2140.	1.5	41
62	PMP22 exon 4 deletion causes ER retention of PMP22 and a gain-of-function allele in CMT1E. <i>Annals of Clinical and Translational Neurology</i> , 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. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2017, 88, 941-952.	0.9	20
65	Charcot-Marie-Tooth Disease Type 1A. <i>Journal of Clinical Neurophysiology</i> , 2017, 34, 508-511.	0.9	2
66	Peripheral neuropathy in complex inherited diseases: an approach to diagnosis. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2017, 88, 846-863.	0.9	51
67	Cross-sectional analysis of a large cohort with X-linked Charcot-Marie-Tooth disease (CMTX1). <i>Neurology</i> , 2017, 89, 927-935.	1.5	44
68	Natural history of Charcot-Marie-Tooth disease during childhood. <i>Annals of Neurology</i> , 2017, 82, 353-359.	2.8	50
69	A human cellular model to study peripheral myelination and demyelinating neuropathies. <i>Brain</i> , 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. <i>Journal of Urology</i> , 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. <i>Journal of Clinical Investigation</i> , 2017, 128, 110-112.	3.9	9

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73	Phenotypic Variability of Childhood Charcot-Marie-Tooth Disease. <i>JAMA Neurology</i> , 2016, 73, 645.	4.5	71
74	Gene therapy, CMT1X, and the inherited neuropathies. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2016, 113, 4552-4554.	3.3	0
75	Rydel-Seiffer fork revisited: Beyond a simple case of black and white. <i>Neurology</i> , 2016, 87, 738-740.	1.5	14
76	LRSAM1 lessons. <i>Annals of Neurology</i> , 2016, 80, 821-822.	2.8	1
77	Charcot-marie-tooth disease type 1X in women: Electrodiagnostic findings. <i>Muscle and Nerve</i> , 2016, 54, 728-732.	1.0	10
78	New and emerging treatments of Charcotâ€“Marieâ€“Tooth disease. <i>Expert Opinion on Orphan Drugs</i> , 2015, 3, 151-164.	0.5	0
79	Prospective study of muscle cramps in Charcotâ€“Marieâ€“Tooth disease. <i>Muscle and Nerve</i> , 2015, 51, 485-488.	1.0	18
80	Update on Charcotâ€“Marieâ€“Tooth disease. <i>Current Opinion in Neurology</i> , 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. <i>Muscle and Nerve</i> , 2015, 52, 1110-1113.	1.0	19
82	Coexistence of a T118M <i>PMP22</i> missense mutation and chromosome 17 (17p11.2â€“p12) deletion. <i>Muscle and Nerve</i> , 2015, 52, 905-908.	1.0	4
83	Progressive Lower Extremity Weakness and Axonal Sensorimotor Polyneuropathy from a Mutation in <i>KIF5A</i> (c.611G>A;p.Arg204Gln). <i>Case Reports in Genetics</i> , 2015, 2015, 1-5.	0.1	4
84	Rare Manifestation of a c.290 C>T, p.Gly97Glu VCP Mutation. <i>Case Reports in Genetics</i> , 2015, 2015, 1-5.	0.1	16
85	Reduced neurofilament expression in cutaneous nerve fibers of patients with CMT2E. <i>Neurology</i> , 2015, 85, 228-234.	1.5	21
86	Exome Sequence Analysis Suggests that Genetic Burden Contributes to Phenotypic Variability and Complex Neuropathy. <i>Cell Reports</i> , 2015, 12, 1169-1183.	2.9	211
87	Demyelinating CMTâ€“whatâ€™s known, whatâ€™s new and whatâ€™s in store?. <i>Neuroscience Letters</i> , 2015, 596, 14-26.	1.0	54
88	Inclusion body myositis and sarcoid myopathy: Coincidental occurrence or associated diseases. <i>Neuromuscular Disorders</i> , 2015, 25, 297-300.	0.3	17
89	Ultrasound: the future for evaluating the PNS in humans?. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2015, 86, 362-362.	0.9	5
90	Small nerve fiber involvement in CMT1A. <i>Neurology</i> , 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. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 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. <i>Journal of Neurology</i> , 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. <i>Neuromuscular Disorders</i> , 2015, 25, 786-793.	0.3	40
95	Genotypeâ€“phenotype characteristics and baseline natural history of heritable neuropathies caused by mutations in the <i>MPZ</i> gene. <i>Brain</i> , 2015, 138, 3180-3192.	3.7	80
96	Charcot-Marie-Tooth disease. <i>Neurology</i> , 2015, 85, 1202-1208.	1.5	33
97	Peripheral Neuropathies. , 2015, , 167-188.		1
98	Detection of Copy Number Variation by SNP-Allelotyping. <i>Journal of Neurogenetics</i> , 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. <i>Experimental Neurology</i> , 2015, 263, 190-199.	2.0	97
100	Hereditary motor and sensory neuropathies: Understanding molecular pathogenesis could lead to future treatment strategies. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2015, 1852, 667-678.	1.8	67
101	Psychometrics evaluation of Charcotâ€“Marieâ€“Tooth Neuropathy Score (<sc>CMTNSv2</sc>) second version, using Rasch analysis. <i>Journal of the Peripheral Nervous System</i> , 2014, 19, 192-196.	1.4	59
102	Effect of pain in pediatric inherited neuropathies. <i>Neurology</i> , 2014, 82, 793-797.	1.5	17
103	Genetic testing practices for Charcot-Marie-Tooth type 1A disease. <i>Muscle and Nerve</i> , 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. <i>Human Molecular Genetics</i> , 2014, 23, 5171-5187.	1.4	21
105	Functional Magnetic Resonance Imaging during Urodynamic Testing Identifies Brain Structures Initiating Micturition. <i>Journal of Urology</i> , 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. <i>Neuromuscular Disorders</i> , 2014, 24, 1003-1017.	0.3	25
107	Quality-of-life in Charcotâ€“Marieâ€“Tooth disease: The patientâ€™s perspective. <i>Neuromuscular Disorders</i> , 2014, 24, 1018-1023.	0.3	71
108	A novel mutation in VCP causes Charcotâ€“Marieâ€“Tooth Type 2 disease. <i>Brain</i> , 2014, 137, 2897-2902.	3.7	116

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109	Inherited Peripheral Neuropathies. <i>Neurologic Clinics</i> , 2013, 31, 597-619.	0.8	88
110	Inherited neuropathies: Clinical overview and update. <i>Muscle and Nerve</i> , 2013, 48, 604-622.	1.0	62
111	Objective Evaluation of Overactive Bladder: Which Surveys Should I Use?. <i>Current Bladder Dysfunction Reports</i> , 2013, 8, 45-50.	0.2	18
112	Dynein mutations associated with hereditary motor neuropathies impair mitochondrial morphology and function with age. <i>Neurobiology of Disease</i> , 2013, 58, 220-230.	2.1	40
113	High-Dosage Ascorbic Acid Treatment in Charcot-Marie-Tooth Disease Type 1A. <i>JAMA Neurology</i> , 2013, 70, 981.	4.5	121
114	Transitioning outcome measures: relationship between the CMTPedS and CMTNSv2 in children, adolescents, and young adults with Charcot-Marie-Tooth disease. <i>Journal of the Peripheral Nervous System</i> , 2013, 18, 177-180.	1.4	15
115	Anterior tibialis cmap amplitude correlations with impairment in CMT1A. <i>Muscle and Nerve</i> , 2013, 47, 493-496.	1.0	17
116	Meeting Report: 2013 Peripheral Nerve Society Biennial Meeting, Saint-Malo, France, June 29-July 3, 2013. <i>Journal of the Peripheral Nervous System</i> , 2013, 18, 197-198.	1.4	1
117	Unfolded protein response, treatment and CMT1B. <i>Rare Diseases (Austin, Tex)</i> , 2013, 1, e24049.	1.8	13
118	Lessons from London. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2012, 83, 767-768.	0.9	2
119	MpzR98C arrests Schwann cell development in a mouse model of early-onset Charcot-Marie-Tooth disease type 1B. <i>Brain</i> , 2012, 135, 2032-2047.	3.7	61
120	Charcot-Marie-Tooth Disease and Related Genetic Neuropathies. <i>CONTINUUM Lifelong Learning in Neurology</i> , 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. <i>Brain</i> , 2012, 135, 3551-3566.	3.7	90
123	X inactivation in females with X-linked Charcot-Marie-Tooth disease. <i>Neuromuscular Disorders</i> , 2012, 22, 617-621.	0.3	22
124	Symmetry of foot alignment and ankle flexibility in paediatric Charcot-Marie-Tooth disease. <i>Clinical Biomechanics</i> , 2012, 27, 744-747.	0.5	18
125	Gain of glycosylation: A new pathomechanism of myelin protein zero mutations. <i>Annals of Neurology</i> , 2012, 71, 427-431.	2.8	20
126	Validation of the Charcot-Marie-Tooth disease pediatric scale as an outcome measure of disability. <i>Annals of Neurology</i> , 2012, 71, 642-652.	2.8	137

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127	The relationship between anogenital distance and azoospermia in adult men. <i>Journal of Developmental and Physical Disabilities</i> , 2012, 35, 726-730.	3.6	48
128	Phenotypic presentation of the Ser63Del MPZ mutation. <i>Journal of the Peripheral Nervous System</i> , 2012, 17, 197-200.	1.4	19
129	The relationship between anogenital distance and the efficacy of varicocele repair. <i>BJU International</i> , 2012, 110, E927-30.	1.3	10
130	Inherited Peripheral Neuropathies. <i>CONTINUUM Lifelong Learning in Neurology</i> , 2011, 17, 294-315.	0.4	5
131	Axonal Charcot-Marie-Tooth disease. <i>Current Opinion in Neurology</i> , 2011, 24, 475-483.	1.8	91
132	Distinct pathogenic processes between Fig4-deficient motor and sensory neurons. <i>European Journal of Neuroscience</i> , 2011, 33, 1401-1410.	1.2	34
133	Phenotype expression in women with CMT1X. <i>Journal of the Peripheral Nervous System</i> , 2011, 16, 102-107.	1.4	42
134	<i>In vivo</i> confocal microscopy of Meissner corpuscles as a novel sensory measure in CMT1A. <i>Journal of the Peripheral Nervous System</i> , 2011, 16, 169-174.	1.4	20
135	Reliability of the CMT neuropathy score (second version) in Charcot-Marie-Tooth disease. <i>Journal of the Peripheral Nervous System</i> , 2011, 16, 191-198.	1.4	269
136	Update on Charcot-Marie-Tooth Disease. <i>Current Neurology and Neuroscience Reports</i> , 2011, 11, 78-88.	2.0	128
137	Charcot-Marie-Tooth disease subtypes and genetic testing strategies. <i>Annals of Neurology</i> , 2011, 69, 22-33.	2.8	494
138	Exome sequencing allows for rapid gene identification in a Charcot-Marie-Tooth family. <i>Annals of Neurology</i> , 2011, 69, 464-470.	2.8	107
139	Genetics of Neuropathies. <i>Seminars in Neurology</i> , 2011, 31, 494-505.	0.5	19
140	Neuropathy in a Human Without the PMP22 Gene. <i>Archives of Neurology</i> , 2011, 68, 814-21.	4.9	21
141	Missense Mutations in the Copper Transporter Gene ATP7A Cause X-Linked Distal Hereditary Motor Neuropathy. <i>American Journal of Human Genetics</i> , 2010, 86, 343-352.	2.6	170
142	Copy number variations are a rare cause of non-CMT1A Charcot-Marie-Tooth disease. <i>Journal of Neurology</i> , 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. <i>Journal of the Peripheral Nervous System</i> , 2010, 15, 161-163.	1.4	0
144	Conduction Block in PMP22 Deficiency. <i>Journal of Neuroscience</i> , 2010, 30, 600-608.	1.7	49

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145	Reply: Internodal length variability of dermal myelinated fibres. <i>Brain</i> , 2010, 133, e143-e143.	3.7	2
146	PMP22 expression in dermal nerve myelin from patients with CMT1A. <i>Brain</i> , 2009, 132, 1734-1740.	3.7	68
147	Shortened internodal length of dermal myelinated nerve fibres in Charcot-Marie-Tooth disease type 1A. <i>Brain</i> , 2009, 132, 3263-3273.	3.7	85
148	Ascorbic acid for treatment of CMT1A: the jury is still out. <i>Lancet Neurology</i> , The, 2009, 8, 505-507.	4.9	6
149	Quality of life in children with CMT type 1A. <i>Lancet Neurology</i> , The, 2009, 8, 880-881.	4.9	10
150	Persistent CNS dysfunction in a boy with CMT1X. <i>Journal of the Neurological Sciences</i> , 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. <i>Advances in Experimental Medicine and Biology</i> , 2009, 652, 201-206.	0.8	1
152	Biology of Peripheral Inherited Neuropathies: Schwann Cell Axonal Interactions. <i>Advances in Experimental Medicine and Biology</i> , 2009, 652, 171-181.	0.8	9
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