Davide Pareyson

List of Publications by Year
in descending order

284	10,814	34016 52	48187 88
papers	citations	h-index	g-index

293	293	293	9909
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Hereditary transthyretin amyloidosis overview. Neurological Sciences, 2022, 43, 595-604.	0.9	39
2	CMT2CC associated withNEFHmutations: a predominantly motor neuronopathy. Journal of Neurology, Neurosurgery and Psychiatry, 2022, 93, 1.2-1.	0.9	3
3	Autophagy and Lysosomal Functionality in CMT2B Fibroblasts Carrying the RAB7K126R Mutation. Cells, 2022, 11, 496.	1.8	10
4	DNAJB2â€related Charcotâ€Marieâ€Tooth disease type 2: Pathomechanism insights and phenotypic spectrum widening. European Journal of Neurology, 2022, 29, 2056-2065.	1.7	7
5	Charcotâ€Marieâ€Tooth disease in Africa. Journal of the Peripheral Nervous System, 2022, 27, 98-99.	1.4	0
6	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
7	European Academy of Neurology guidance for developing and reporting clinical practice guidelines on rare neurological diseases. European Journal of Neurology, 2022, 29, 1571-1586.	1.7	5
8	Optical coherence tomography in adult adrenoleukodystrophy: a cross-sectional and longitudinal study. Neurological Sciences, 2021, 42, 235-241.	0.9	3
9	Hypomyelinating leukodystrophies in adults: Clinical and genetic features. European Journal of Neurology, 2021, 28, 934-944.	1.7	14
10	Dysregulation of myelin synthesis and actomyosin function underlies aberrant myelin in CMT4B1 neuropathy. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	3.3	17
11	Psychosocial burden and professional and social support in patients with hereditary transthyretin amyloidosis (ATTRv) and their relatives in Italy. Orphanet Journal of Rare Diseases, 2021, 16, 163.	1.2	8
12	Neuro-telehealth for fragile patients in a tertiary referral neurological institute during the COVID-19 pandemic in Milan, Lombardy. Neurological Sciences, 2021, 42, 2637-2644.	0.9	18
13	Dysregulation of Muscle-Specific MicroRNAs as Common Pathogenic Feature Associated with Muscle Atrophy in ALS, SMA and SBMA: Evidence from Animal Models and Human Patients. International Journal of Molecular Sciences, 2021, 22, 5673.	1.8	14
14	Charcotâ€“Marieâ€“Tooth disease type 2F associated with biallelic HSPB1 mutations. Annals of Clinical and Translational Neurology, 2021, 8, 1158-1164.	1.7	4
15	Updated review of therapeutic strategies for Charcot-Marie-Tooth disease and related neuropathies. Expert Review of Neurotherapeutics, 2021, 21, 701-713.	1.4	16
16	Association Between Body Mass Index and Disability in Children With Charcot-Marie-Tooth Disease. Neurology, 2021, 97, e1727-e1736.	1.5	2
17	Challenges in Treating Charcot-Marie-Tooth Disease and Related Neuropathies: Current Management and Future Perspectives. Brain Sciences, 2021, 11, 1447.	1.1	22
18	Hereditary neuropathy with liability to pressure palsies. Journal of Neurology, 2020, 267, 2198-2206.	1.8	31

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19	Treadmill training in patients affected by Charcotâ€“Marieâ€“Tooth neuropathy: results of a multicenter, prospective, randomized, singleâ€blind, controlled study. European Journal of Neurology, 2020, 27, 280-287.	1.7	19
20	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
21	Myelin protein zero gene dose dependent axonal ion-channel dysfunction in a family with Charcot-Marie-Tooth disease. Clinical Neurophysiology, 2020, 131, 2440-2451.	0.7	7
22	ATTRv amyloidosis Italian Registry: clinical and epidemiological data. Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis, 2020, 27, 259-265.	1.4	51
23	hATTR Pathology: Nerve Biopsy Results from Italian Referral Centers. Brain Sciences, 2020, 10, 780.	1.1	24
24	Reliability of the <scp>Charcotâ€Marieâ€Tooth</scp> functional outcome measure. Journal of the Peripheral Nervous System, 2020, 25, 288-291.	1.4	8
25	Pregnancy in Charcot-Marie-Tooth disease. Neurology, 2020, 95, e3180-e3189.	1.5	11
26	Recommendations for pre-symptomatic genetic testing for hereditary transthyretin amyloidosis in the era of effective therapy: a multicenter Italian consensus. Orphanet Journal of Rare Diseases, 2020, 15, 348.	1.2	22
27	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
28	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
29	MYORG -related disease is associated with central pontine calcifications and atypical parkinsonism. Neurology: Genetics, 2020, 6, e399.	0.9	13
30	Validation of the Italian version of the <scp>Charcotâ€Marieâ€Tooth</scp> Health Index. Journal of the Peripheral Nervous System, 2020, 25, 292-296.	1.4	3
31	Discovering the Italian phenotype of cerebral amyloid angiopathy (CAA): the SENECA project. Neurological Sciences, 2020, 41, 2193-2200.	0.9	3
32	A longitudinal study of CMT1A using Rasch analysis based CMT neuropathy and examination scores. Neurology, 2020, 94, e884-e896.	1.5	29
33	Severe worsening of adult-onset Alexander disease after minor head trauma: Report of two patients and review of the literature. Journal of Clinical Neuroscience, 2020, 75, 221-223.	0.8	6
34	Charcot-Marie-Tooth Type 2B: A New Phenotype Associated with a Novel RAB7A Mutation and Inhibited EGFR Degradation. Cells, 2020, 9, 1028.	1.8	20
35	ATPase Domain <scp>AFG3L2</scp> Mutations Alter <scp>OPA1</scp> Processing and Cause Optic Neuropathy. Annals of Neurology, 2020, 88, 18-32.	2.8	31
36	Asymptomatic adrenoleukodystrophy in elderly males. Journal of Neurology, 2020, 267, 1849-1851.	1.8	0

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37	Natural history of Charcot-Marie-Tooth disease type 2A: a large international multicentre study. Brain, 2020, 143, 3589-3602.	3.7	39
38	Outcome measures in the clinical evaluation of ambulatory Charcot-Marie-Tooth 1A subjects. European Journal of Physical and Rehabilitation Medicine, 2019, 55, 47-55.	1.1	9
39	Mutation update for myelin protein zero-related neuropathies and the increasing role of variants causing a late-onset phenotype. Journal of Neurology, 2019, 266, 2629-2645.	1.8	23
40	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
41	Expanding the spectrum of genes responsible for hereditary motor neuropathies. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, 1171-1179.	0.9	30
42	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
43	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
44	Neuropsychological features of adult form of Alexander disease. Journal of the Neurological Sciences, 2019, 401, 87-89.	0.3	7
45	Balance impairment in pediatric charcotâ€“marieâ€“tooth disease. Muscle and Nerve, 2019, 60, 242-249.	1.0	22
46	Biopsy-proven primary angiitis of the central nervous system mimicking leukodystrophy: A case report and review of the literature. Journal of Clinical Neuroscience, 2019, 64, 42-44.	0.8	9
47	Longitudinal quantitative magnetic resonance imaging in adrenomyeloneuropathy. European Journal of Neurology, 2019, 26, 1341-1344.	1.7	1
48	Are novel outcome measures for Charcotâ€“Marieâ€“Tooth disease sensitive to change? The 6-minute walk test and StepWatchâ„¢ Activity Monitor in a 12-month longitudinal study. Neuromuscular Disorders, 2019, 29, 310-316.	0.3	6
49	Saposin B deficiency as a cause of adult-onset metachromatic leukodystrophy. Neurology, 2019, 93, 310-312.	1.5	6
50	Leukoencephalopathy With Predominant Infratentorial Involvement Caused by a Novel ABCD1 Mutation. Neurologist, 2019, 24, 194-197.	0.4	2
51	PFN2 and GAMT as common molecular determinants of axonal Charcot-Marie-Tooth disease. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 870-878.	0.9	16
52	Neurofilament light, biomarkers, and Charcot-Marie-Tooth disease. Neurology, 2018, 90, 257-259.	1.5	8
53	Prevalence and orthopedic management of foot and ankle deformities in Charcotâ€“Marieâ€“Tooth disease. Muscle and Nerve, 2018, 57, 255-259.	1.0	39
54	Mitochondrial disorders of the retinal ganglion cells and the optic nerve. Mitochondrion, 2018, 42, 1-10.	1.6	18

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55	Altered <scp>TDP</scp>â€43â€dependent splicing in <scp>HSPB</scp>8â€related distal hereditary motor neuropathy and myofibrillar myopathy. European Journal of Neurology, 2018, 25, 154-163.	1.7	24
56	Adult leukoencephalopathies with prominent infratentorial involvement can be caused by Erdheimâ€“Chester disease. Journal of Neurology, 2018, 265, 273-284.	1.8	17
57	Different nerve ultrasound patterns in charcotâ€marieâ€tooth types and hereditary neuropathy with liability to pressure palsies. Muscle and Nerve, 2018, 57, E18-E23.	1.0	28
58	Development and validation of the Charcot-Marie-Tooth Disease Infant Scale. Brain, 2018, 141, 3319-3330.	3.7	25
59	The Italian neuromuscular registry: a coordinated platform where patient organizations and clinicians collaborate for data collection and multiple usage. Orphanet Journal of Rare Diseases, 2018, 13, 176.	1.2	31
60	Electromyographic and biomechanical analysis of step negotiation in Charcot Marie Tooth subjects whose level walk is not impaired. Gait and Posture, 2018, 62, 497-504.	0.6	6
61	Repeatability, consistency, and accuracy of handâ€held dynamometry with and without fixation for measuring ankle plantarflexion strength in healthy adolescents and adults. Muscle and Nerve, 2017, 56, 896-900.	1.0	14
62	Mutations in noncoding regions of GJB1 are a major cause of X-linked CMT. Neurology, 2017, 88, 1445-1453.	1.5	45
63	2017 Peripheral Nerve Society Meeting July 8â€“12, 2017 Sitges, Barcelona, Spain. Journal of the Peripheral Nervous System, 2017, 22, 226-414.	1.4	14
64	Lack of benefit of acetyl- dl -leucine in patients with multiple system atrophy of the cerebellar type. Journal of the Neurological Sciences, 2017, 379, 12-13.	0.3	3
65	Hereditary gelsolin amyloidosis (<scp>HGA</scp>): a neglected cause of bilateral progressive or recurrent facial palsy. Journal of the Peripheral Nervous System, 2017, 22, 59-63.	1.4	8
66	A novel <scp>NDRG1</scp> mutation in a nonâ€Romani patient with <scp>CMT4D</scp>/<scp>HMSN</scp>â€Lom. Journal of the Peripheral Nervous System, 2017, 22, 47-50.	1.4	6
67	221st ENMC International Workshop:. Neuromuscular Disorders, 2017, 27, 1138-1142.	0.3	10
68	Biomarkers predict outcome in Charcot-Marie-Tooth disease 1A. Journal of Neurology, Neurosurgery and Psychiatry, 2017, 88, 941-952.	0.9	20
69	Responsiveness of gait analysis parameters in a cohort of 71 CMT subjects. Neuromuscular Disorders, 2017, 27, 1029-1037.	0.3	10
70	Posterior column ataxia with retinitis pigmentosa coexisting with sensoryâ€autonomic neuropathy and leukemia due to the homozygous p.Pro221Ser FLVCR1 mutation. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2017, 174, 732-739.	1.1	21
71	Peripheral neuropathy in complex inherited diseases: an approach to diagnosis. Journal of Neurology, Neurosurgery and Psychiatry, 2017, 88, 846-863.	0.9	51
72	Cross-sectional analysis of a large cohort with X-linked Charcot-Marie-Tooth disease (CMTX1). Neurology, 2017, 89, 927-935.	1.5	44

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73	Natural history of Charcotâ€Marieâ€Tooth disease during childhood. Annals of Neurology, 2017, 82, 353-359.	2.8	50
74	New developments in Charcotâ€“Marieâ€“Tooth neuropathy and related diseases. Current Opinion in Neurology, 2017, 30, 471-480.	1.8	145
75	Transcriptional role of androgen receptor in the expression of long non-coding RNA Sox2OT in neurogenesis. PLoS ONE, 2017, 12, e0180579.	1.1	19
76	Novel outcome measures for Charcotâ€™Marieâ€™Tooth disease: validation and reliability of the 6â€min walk test and StepWatch^{â„¢} Activity Monitor and identification of the walking features related to higher quality of life. European Journal of Neurology, 2016, 23, 1343-1350.	1.7	26
77	Screening for SH3TC2 gene mutations in a series of demyelinating recessive Charcotâ€Marieâ€Tooth disease (CMT4). Journal of the Peripheral Nervous System, 2016, 21, 142-149.	1.4	32
78	No effect of <scp>AR</scp> polyG polymorphism on spinal and bulbar muscular atrophy phenotype. European Journal of Neurology, 2016, 23, 1134-1136.	1.7	8
79	Laryngeal and phrenic nerve involvement in a patient with hereditary neuropathy with liability to pressure palsies (HNPP). Neuromuscular Disorders, 2016, 26, 455-458.	0.3	3
80	A novel synonymous mutation in the MPZ gene causing an aberrant splicing pattern and Charcot-Marie-Tooth disease type 1b. Neuromuscular Disorders, 2016, 26, 516-520.	0.3	18
81	Phenotypic Variability of Childhood Charcot-Marie-Tooth Disease. JAMA Neurology, 2016, 73, 645.	4.5	71
82	Monitoring effectiveness and safety of Tafamidis in transthyretin amyloidosis in Italy: a longitudinal multicenter study in a non-endemic area. Journal of Neurology, 2016, 263, 916-924.	1.8	76
83	Quantitative MRI of the spinal cord and brain in adrenomyeloneuropathy:in vivoassessment of structural changes. Brain, 2016, 139, 1735-1746.	3.7	44
84	Validation of the Italian version of the SBMA Functional Rating Scale as outcome measure. Neurological Sciences, 2016, 37, 1815-1821.	0.9	9
85	Nerve conduction velocity in <scp>CMT</scp>1A: what else can we tell?. European Journal of Neurology, 2016, 23, 1566-1571.	1.7	45
86	Neurofascin-155 as a putative antigen in combined central and peripheral demyelination. Neurology: Neuroimmunology and NeuroInflammation, 2016, 3, e238.	3.1	32
87	Early-onset progressive spastic paraplegia caused by a novel TUBB4A mutation: brain MRI and FDG-PET findings. Journal of Neurology, 2016, 263, 591-593.	1.8	17
88	Towards a European Registry and Biorepository for Patients with Spinal and Bulbar Muscular Atrophy. Journal of Molecular Neuroscience, 2016, 58, 394-400.	1.1	10
89	Clinical Trials in Spinal and Bulbar Muscular Atrophyâ€”Past, Present, and Future. Journal of Molecular Neuroscience, 2016, 58, 379-387.	1.1	15
90	Non-neural phenotype of spinal and bulbar muscular atrophy: results from a large cohort of Italian patients. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, 810-816.	0.9	59

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91	Alternative Splicing in the HumanPMP22Gene: Implications in CMT1A Neuropathy. Human Mutation, 2016, 37, 98-109.	1.1	10
92	Combined central and peripheral demyelination: Clinical features, diagnostic findings, and treatment. Journal of the Neurological Sciences, 2016, 363, 182-187.	0.3	55
93	The role of androgen receptor gene variants on SBMA phenotype. Journal of the Neurological Sciences, 2015, 357, e231.	0.3	0
94	Novel lossâ€ofâ€function mutation of the HINT1 gene in a patient with distal motor axonal neuropathy without neuromyotonia. Muscle and Nerve, 2015, 52, 688-689.	1.0	14
95	Mutational mechanisms in <scp>MFN2</scp>â€related neuropathy: compound heterozygosity for recessive and semidominant mutations. Journal of the Peripheral Nervous System, 2015, 20, 380-386.	1.4	23
96	Responsiveness of clinical outcome measures in Charcotâ€Marieâ€Tooth disease. European Journal of Neurology, 2015, 22, 1556-1563.	1.7	47
97	Monitoring safety and effectiveness of Tafamidis in transthyretin amyloidosis in Italy: a 3-year longitudinal multicenter study in a non-endemic area. Orphanet Journal of Rare Diseases, 2015, 10, .	1.2	0
98	Ascorbic acid for the treatment of Charcot-Marie-Tooth disease. The Cochrane Library, 2015, 2015, CD011952.	1.5	36
99	CMT subtypes and disease burden in patients enrolled in the Inherited Neuropathies Consortium natural history study: a cross-sectional analysis. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, 873-878.	0.9	249
100	210th ENMC International Workshop: Research and clinical management of patients with spinal and bulbar muscular atrophy, 27â€“29 March, 2015, Naarden, The Netherlands. Neuromuscular Disorders, 2015, 25, 802-812.	0.3	16
101	Leukodystrophies. , 2015, , 163-175.		0
102	The influence of somatosensory and muscular deficits on postural stabilization: Insights from an instrumented analysis of subjects affected by different types of Charcotâ€“Marieâ€“Tooth disease. Neuromuscular Disorders, 2015, 25, 640-645.	0.3	16
103	Frequency and time to relapse after discontinuing 6-month therapy with IVIg or pulsed methylprednisolone in CIDP. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, 729-734.	0.9	70
104	A slowly progressive mitochondrial encephalomyopathy widens the spectrum of AIFM1 disorders. Neurology, 2015, 84, 2193-2195.	1.5	47
105	Mitochondrial dynamics and inherited peripheral nerve diseases. Neuroscience Letters, 2015, 596, 66-77.	1.0	103
106	Spinal and bulbar muscular atrophy and Charcotâ€“Marieâ€“Tooth type 1A: Co-existence of two rare neuromuscular genetic diseases in the same patient. Neuromuscular Disorders, 2015, 25, 800-801.	0.3	3
107	Genotypeâ€“phenotype characteristics and baseline natural history of heritable neuropathies caused by mutations in theMPZgene. Brain, 2015, 138, 3180-3192.	3.7	80
108	CSF and Blood Levels of GFAP in Alexander Disease. ENeuro, 2015, 2, ENEURO.0080-15.2015.	0.9	30

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109	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
110	Truncating and Missense Mutations in IGHMBP2 Cause Charcot-Marie Tooth Disease Type 2. American Journal of Human Genetics, 2014, 95, 590-601.	2.6	75
111	A new mutation in GJC2 associated with subclinical leukodystrophy. Journal of Neurology, 2014, 261, 1929-1938.	1.8	23
112	Expanding sialidosis spectrum by genome-wide screening. Neurology, 2014, 82, 2003-2006.	1.5	37
113	Brain fluorodeoxyglucose PET in adrenoleukodystrophy. Neurology, 2014, 83, 981-989.	1.5	15
114	PMP22 messenger RNA levels in skin biopsies: testing the effectiveness of a Charcot-Marie-Tooth 1A biomarker. Brain, 2014, 137, 1614-1620.	3.7	33
115	Overlapping phenotypes in complex spastic paraplegias SPG11, SPG15, SPG35 and SPG48. Brain, 2014, 137, 1907-1920.	3.7	133
116	Xâ€linked Charcotâ€Marieâ€Tooth type 1: strokeâ€like presentation of a novel <scp>GJB1</scp> mutation. Journal of the Peripheral Nervous System, 2014, 19, 183-186.	1.4	20
117	Adult polyglucosan body disease in a patient originally diagnosed with Fabryâ€™s disease. Neuromuscular Disorders, 2014, 24, 272-276.	0.3	11
118	Memory loss: do not forget the mammillary bodies. Neurological Sciences, 2014, 35, 473-474.	0.9	0
119	Recurrent generalized seizures, visual loss, and palinopsia as phenotypic features of neuronal ceroid lipofuscinosis due to progranulin gene mutation. Epilepsia, 2014, 55, e56-9.	2.6	50
120	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
121	Pain and small fiber function in charcotâ€“marieâ€“tooth disease type 1A. Muscle and Nerve, 2014, 50, 366-371.	1.0	26
122	Is overwork weakness relevant in Charcotâ€“Marieâ€“Tooth disease?. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 1354-1358.	0.9	23
123	Subclinical leukodystrophy and infertility in a man with a novel homozygous CLCN2 mutation. Neurology, 2014, 83, 1217-1218.	1.5	42
124	Postural stabilization and balance assessment in Charcotâ€“Marieâ€“Tooth 1A subjects. Gait and Posture, 2014, 40, 481-486.	0.6	29
125	Different mutations at V363 MAPT codon are associated with atypical clinical phenotypes and show unusual structural and functional features. Neurobiology of Aging, 2014, 35, 408-417.	1.5	36
126	An update on clinical trials in Charcot-Marie-Tooth Disease. Clinical Investigation, 2014, 4, 221-225.	0.0	4

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127	Human Adipose-Derived Mesenchymal Stem Cells as a New Model of Spinal and Bulbar Muscular Atrophy. PLoS ONE, 2014, 9, e112746.	1.1	15
128	Charcot-Marie-Tooth Disease and Related Hereditary Neuropathies: From Gene Function to Associated Phenotypes. Current Molecular Medicine, 2014, 14, 1009-1033.	0.6	24
129	An autoinflammatory neurological disease due to interleukin 6 hypersecretion. Journal of Neuroinflammation, 2013, 10, 29.	3.1	11
130	Changes of gait pattern in children with Charcot-Marie-Tooth disease type 1A: a 18 months follow-up study. Journal of NeuroEngineering and Rehabilitation, 2013, 10, 65.	2.4	19
131	Selective theory of mind impairment and cerebellar atrophy: a case report. Journal of Neurology, 2013, 260, 2166-2169.	1.8	11
132	Adult-onset leukodystrophies from respiratory chain disorders: do they exist?. Journal of Neurology, 2013, 260, 1617-1623.	1.8	5
133	Dominant Charcotâ€“Marieâ€“Tooth syndrome and cognate disorders. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2013, 115, 817-845.	1.0	7
134	Vitamin C and Charcotâ€“Marieâ€“Tooth 1A: Pharmacokinetic considerations. PharmaNutrition, 2013, 1, 10-12.	0.8	8
135	Inherited neuropathies: an update. Journal of Neurology, 2013, 260, 2684-2690.	1.8	3
136	SETX mutations are a frequent genetic cause of juvenile and adult onset cerebellar ataxia with neuropathy and elevated serum alpha-fetoprotein. Orphanet Journal of Rare Diseases, 2013, 8, 123.	1.2	31
137	Peripheral neuropathy in mitochondrial disorders. Lancet Neurology, The, 2013, 12, 1011-1024.	4.9	101
138	Pendular nystagmus in hypomyelinating leukodystrophy. Journal of Clinical Neuroscience, 2013, 20, 1443-1445.	0.8	6
139	Authorsâ€™ response to Rahman and Fontes. PharmaNutrition, 2013, 1, 101.	0.8	0
140	Influence of comorbidities on the phenotype of patients affected by Charcotâ€“Marieâ€“Tooth neuropathy type 1A. Neuromuscular Disorders, 2013, 23, 902-906.	0.3	15
141	An essential role of MAG in mediating axonâ€“myelin attachment in Charcotâ€“Marieâ€“Tooth 1A disease. Neurobiology of Disease, 2013, 49, 221-231.	2.1	29
142	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
143	Coexistence of Charcotâ€Marieâ€Tooth disease type 1A and antiâ€MAG neuropathy. Journal of the Peripheral Nervous System, 2013, 18, 185-188.	1.4	1
144	Pachymeningeal involvement in POEMS syndrome: MRI and histopathological study. Journal of Neurology, Neurosurgery and Psychiatry, 2012, 83, 33-37.	0.9	24

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145	Clinical neurogenetics: recent advances. Journal of Neurology, 2012, 259, 2255-2260.	1.8	1
146	Effectiveness of valproate for the treatment of manic-like behavior in X-linked adrenoleukodystrophy. Neurological Sciences, 2012, 33, 1197-1199.	0.9	6
147	Vertical supranuclear gaze palsy in Niemann-Pick type C disease. Neurological Sciences, 2012, 33, 1225-1232.	0.9	60
148	Symmetry of foot alignment and ankle flexibility in paediatric Charcotâ€“Marieâ€“Tooth disease. Clinical Biomechanics, 2012, 27, 744-747.	0.5	18
149	Gait pattern classification in children with Charcotâ€“Marieâ€“Tooth disease type 1A. Gait and Posture, 2012, 35, 131-137.	0.6	72
150	Myelin protein zero <scp>Arg36Gly</scp> mutation with very late onset and rapidly progressive painful neuropathy. Journal of the Peripheral Nervous System, 2012, 17, 422-425.	1.4	9
151	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
152	Mutations in CTC1 , Encoding the CTS Telomere Maintenance Complex Component 1, Cause Cerebroretinal Microangiopathy with Calcifications and Cysts. American Journal of Human Genetics, 2012, 90, 540-549.	2.6	141
153	Strikingly Different Clinicopathological Phenotypes Determined by Progranulin-Mutation Dosage. American Journal of Human Genetics, 2012, 90, 1102-1107.	2.6	414
154	Intravenous immunoglobulin versus intravenous methylprednisolone for chronic inflammatory demyelinating polyradiculoneuropathy: a randomised controlled trial. Lancet Neurology, The, 2012, 11, 493-502.	4.9	185
155	Preferential expression of mutant ABCD1 allele is common in adrenoleukodystrophy female carriers but unrelated to clinical symptoms. Orphanet Journal of Rare Diseases, 2012, 7, 10.	1.2	29
156	Mitochondrial dementia: A sporadic case of progressive cognitive and behavioral decline with hearing loss due to the rare m.3291T>C MELAS mutation. Journal of the Neurological Sciences, 2011, 300, 165-168.	0.3	26
157	Lower limb areflexia without central and peripheral conduction abnormalities is highly suggestive of Gerstmannâ€“StrÃ¤usslerâ€“Scheinker disease Pro102Leu. Journal of the Neurological Sciences, 2011, 302, 85-88.	0.3	6
158	Reliability of instrumented movement analysis as outcome measure in Charcotâ€“Marieâ€“Tooth disease: Results from a multitask locomotor protocol. Gait and Posture, 2011, 34, 36-43.	0.6	25
159	Co-occurrence of amyotrophic lateral sclerosis and Charcot-Marie-Tooth disease type 2A in a patient with a novel mutation in the mitofusin-2 gene. Neuromuscular Disorders, 2011, 21, 129-131.	0.3	19
160	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
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