Davide Pareyson

List of Publications by Year in descending order

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284 papers

10,814 citations

52 h-index 88 g-index

293 all docs 293 docs citations

times ranked

293

9909 citing authors

#	Article	IF	CITATIONS
1	Diagnosis, natural history, and management of Charcot–Marie–Tooth disease. Lancet Neurology, The, 2009, 8, 654-667.	4.9	475
2	Strikingly Different Clinicopathological Phenotypes Determined by Progranulin-Mutation Dosage. American Journal of Human Genetics, 2012, 90, 1102-1107.	2.6	414
3	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
4	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
5	Ascorbic acid in Charcot–Marie–Tooth disease type 1A (CMT-TRIAAL and CMT-TRAUK): a double-blind randomised trial. Lancet Neurology, The, 2011, 10, 320-328.	4.9	222
6	Axonal swellings predict the degeneration of epidermal nerve fibers in painful neuropathies. Neurology, 2003, 61, 631-636.	1.5	220
7	Polyneuropathy in POEMS syndrome: role of angiogenic factors in the pathogenesis. Brain, 2005, 128, 1911-1920.	3.7	216
8	Intravenous immunoglobulin versus intravenous methylprednisolone for chronic inflammatory demyelinating polyradiculoneuropathy: a randomised controlled trial. Lancet Neurology, The, 2012, 11, 493-502.	4.9	185
9	Adult-onset Alexander disease: a series of eleven unrelated cases with review of the literature. Brain, 2008, 131, 2321-2331.	3.7	169
10	Randomised controlled trial of methotrexate for chronic inflammatory demyelinating polyradiculoneuropathy (RMC trial): a pilot, multicentre study. Lancet Neurology, The, 2009, 8, 158-164.	4.9	155
11	Sensory neuron diseases. Lancet Neurology, The, 2005, 4, 349-361.	4.9	151
12	Clinical and electrophysiological aspects of Charcot-Marie-Tooth disease. NeuroMolecular Medicine, 2006, 8, 3-22.	1.8	149
13	New developments in Charcot–Marie–Tooth neuropathy and related diseases. Current Opinion in Neurology, 2017, 30, 471-480.	1.8	145
14	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
15	Spinal radiological findings in nine patients with spontaneous intracranial hypotension. Neuroradiology, 2002, 44, 143-150.	1.1	139
16	Validation of the Charcot–Marie–Tooth disease pediatric scale as an outcome measure of disability. Annals of Neurology, 2012, 71, 642-652.	2.8	137
17	Hereditary spastic paraplegia is a novel phenotype for GJA12/GJC2 mutations. Brain, 2009, 132, 426-438.	3.7	135
18	Overlapping phenotypes in complex spastic paraplegias SPG11, SPG15, SPG35 and SPG48. Brain, 2014, 137, 1907-1920.	3.7	133

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19	Ataxia with isolated vitamin E deficiency: neurological phenotype, clinical follow-up and novel mutations in TTPAgene in Italian families. Neurological Sciences, 2004, 25, 130-137.	0.9	131
20	Phenotypic heterogeneity in hereditary neuropathy with liability to pressure palsies associated with chromosome 17p11.2-12 deletion. Neurology, 1996, 46, 1133-1137.	1.5	118
21	Expression of capsaicin receptor immunoreactivity in human peripheral nervous system and in painful neuropathies. Journal of the Peripheral Nervous System, 2006, 11, 262-271.	1.4	114
22	Phenotypic manifestations associated with CAG-repeat expansion in the androgen receptor gene in male patients and heterozygous females: a clinical and molecular study of 30 families. Neuromuscular Disorders, 2000, 10, 391-397.	0.3	112
23	A multicentre follow-up study of 1152 patients with myasthenia gravis in Italy. Journal of Neurology, 1990, 237, 339-344.	1.8	108
24	Mitochondrial dynamics and inherited peripheral nerve diseases. Neuroscience Letters, 2015, 596, 66-77.	1.0	103
25	Peripheral neuropathy in mitochondrial disorders. Lancet Neurology, The, 2013, 12, 1011-1024.	4.9	101
26	IgM deposits on skin nerves in anti-myelin-associated glycoprotein neuropathy. Annals of Neurology, 2005, 57, 180-187.	2.8	100
27	Can MR Imaging Diagnose Adult-Onset Alexander Disease?. American Journal of Neuroradiology, 2008, 29, 1190-1196.	1.2	99
28	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
29	Cranial nerve involvement in CMT disease type 1 due to early growth response 2 gene mutation. Neurology, 2000, 54, 1696-1698.	1.5	84
30	Charcot-Marie-Tooth disease and related neuropathies: Molecular basis for distinction and diagnosis. Muscle and Nerve, 1999, 22, 1498-1509.	1.0	83
31	Genotype–phenotype characteristics and baseline natural history of heritable neuropathies caused by mutations in the⟨i⟩MPZ⟨/i⟩gene. Brain, 2015, 138, 3180-3192.	3.7	80
32	Clinical and magnetic resonance imaging findings in chronic sensory ganglionopathies. Annals of Neurology, 2000, 47, 104-109.	2.8	79
33	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
34	Epidermal nerve fiber density in sensory ganglionopathies: Clinical and neurophysiologic correlations. Muscle and Nerve, 2001, 24, 1034-1039.	1.0	75
35	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
36	Recessive axonal Charcot-Marie-Tooth disease due to compound heterozygous mitofusin 2 mutations. Neurology, 2011, 77, 168-173.	1.5	72

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37	Gait pattern classification in children with Charcot–Marie–Tooth disease type 1A. Gait and Posture, 2012, 35, 131-137.	0.6	72
38	Phenotypic Variability of Childhood Charcot-Marie-Tooth Disease. JAMA Neurology, 2016, 73, 645.	4.5	71
39	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
40	Tubule and neurofilament immunoreactivity in human hairy skin: Markers for intraepidermal nerve fibers. Muscle and Nerve, 2004, 30, 310-316.	1.0	66
41	Further evidence for genetic heterogeneity of distal HMN type V, CMT2 with predominant hand involvement and Silver syndrome. Journal of the Neurological Sciences, 2007, 263, 100-106.	0.3	64
42	Clinical and molecular studies of 73 Italian families with autosomal dominant cerebellar ataxia type I: SCA1 and SCA2 are the most common genotypes. Journal of Neurology, 1999, 246, 389-393.	1.8	63
43	Relative Frequencies of CAG Expansions in Spinocerebellar Ataxia and Dentatorubropallidoluysian Atrophy in 116 Italian Families. European Neurology, 2000, 44, 31-36.	0.6	63
44	Activation of the Contact System in Cerebrospinal Fluid of Patients with Alzheimer Disease. Alzheimer Disease and Associated Disorders, 1998, 12, 102-108.	0.6	62
45	Chronic inflammatory demyelinating polyradiculoneuropathy: long-term course and treatment of 60 patients. Neurological Sciences, 2000, 21, 31-37.	0.9	62
46	Vertical supranuclear gaze palsy in Niemann-Pick type C disease. Neurological Sciences, 2012, 33, 1225-1232.	0.9	60
47	Gene dosage effects in hereditary peripheral neuropathy. Neurology, 1997, 49, 1635-1640.	1.5	59
48	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
49	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
50	Excellent response to steroid treatment in anti-GAD cerebellar ataxia. Lancet Neurology, The, 2003, 2, 634-635.	4.9	57
51	Balance control in Sensory Neuron Disease. Clinical Neurophysiology, 2007, 118, 538-550.	0.7	57
52	Reliability of clinical outcome measures in Charcot-Marie-Tooth disease. Neuromuscular Disorders, 2008, 18, 19-26.	0.3	55
53	Combined central and peripheral demyelination: Clinical features, diagnostic findings, and treatment. Journal of the Neurological Sciences, 2016, 363, 182-187.	0.3	55
54	Differential diagnosis of Charcot-Marie-Tooth disease and related neuropathies. Neurological Sciences, 2004, 25, 72-82.	0.9	51

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55	Peripheral neuropathy in complex inherited diseases: an approach to diagnosis. Journal of Neurology, Neurosurgery and Psychiatry, 2017, 88, 846-863.	0.9	51
56	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
57	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
58	Natural history of Charcotâ€Marie‶ooth disease during childhood. Annals of Neurology, 2017, 82, 353-359.	2.8	50
59	Chloroquine myopathy and myasthenia-like syndrome. Muscle and Nerve, 1988, 11, 114-119.	1.0	48
60	Epidural anaesthesia and spinal arachnoiditis. Anaesthesia, 1989, 44, 317-321.	1.8	47
61	A multicenter, randomized, double-blind, placebo-controlled trial of long-term ascorbic acid treatment in Charcot-Marie-Tooth disease type 1A (CMT-TRIAAL): The study protocol [EudraCT no.: 2006-000032-27]. Pharmacological Research, 2006, 54, 436-441.	3.1	47
62	Four novel cases of periaxin-related neuropathy and review of the literature. Neurology, 2010, 75, 1830-1838.	1.5	47
63	Responsiveness of clinical outcome measures in Charcotâ^'Marieâ^'Tooth disease. European Journal of Neurology, 2015, 22, 1556-1563.	1.7	47
64	A slowly progressive mitochondrial encephalomyopathy widens the spectrum of <i>AIFM1</i> disorders. Neurology, 2015, 84, 2193-2195.	1.5	47
65	Neurophysiological Diagnosis of Acquired Sensory Ganglionopathies. European Neurology, 2003, 50, 146-152.	0.6	46
66	Ataxia with oculomotor apraxia type1 (AOA1): novel and recurrent aprataxin mutations, coenzyme Q10 analyses, and clinical findings in Italian patients. Neurogenetics, 2011, 12, 193-201.	0.7	46
67	Nerve conduction velocity in <scp>CMT</scp> 1A: what else can we tell?. European Journal of Neurology, 2016, 23, 1566-1571.	1.7	45
68	Mutations in noncoding regions of $\langle i \rangle GJB1 \langle i \rangle$ are a major cause of X-linked CMT. Neurology, 2017, 88, 1445-1453.	1.5	45
69	Quantitative MRI of the spinal cord and brain in adrenomyeloneuropathy: <i>in vivo</i> assessment of structural changes. Brain, 2016, 139, 1735-1746.	3.7	44
70	Cross-sectional analysis of a large cohort with X-linked Charcot-Marie-Tooth disease (CMTX1). Neurology, 2017, 89, 927-935.	1.5	44
71	Diagnosis of hereditary neuropathies in adult patients. Journal of Neurology, 2003, 250, 148-160.	1.8	43
72	The G389R mutation in the <i>MAPT</i> gene presenting as sporadic corticobasal syndrome. Movement Disorders, 2008, 23, 892-895.	2.2	43

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73	A novel mutation in the mitochondrial tRNAVal gene associated with a complex neurological presentation. Annals of Neurology, 1998, 43, 98-101.	2.8	42
74	Rare association of motor neuron disease and spinocerebellar ataxia type 2 (SCA2): a new case and review of the literature. Journal of Neurology, 2009, 256, 1926-1928.	1.8	42
75	Subclinical leukodystrophy and infertility in a man with a novel homozygous <i>CLCN2</i> mutation. Neurology, 2014, 83, 1217-1218.	1.5	42
76	HMSN III phenotype due to homozygous expression of a dominant HMSN II gene. Neurology, 1992, 42, 2201-2201.	1.5	42
77	Natural history of CMT1A including QoL: A 2-year prospective study. Neuromuscular Disorders, 2008, 18, 199-203.	0.3	40
78	Prevalence and orthopedic management of foot and ankle deformities in Charcot–Marie–Tooth disease. Muscle and Nerve, 2018, 57, 255-259.	1.0	39
79	Hereditary transthyretin amyloidosis overview. Neurological Sciences, 2022, 43, 595-604.	0.9	39
80	Natural history of Charcot-Marie-Tooth disease type 2A: a large international multicentre study. Brain, 2020, 143, 3589-3602.	3.7	39
81	Partial laminin ?2 chain deficiency in a patient with myopathy resembling inclusion body myositis. Annals of Neurology, 2000, 47, 811-816.	2.8	37
82	Imaging and outcome in severe complications of lumbar epidural anaesthesia: report of 16 cases. Neuroradiology, 2000, 42, 564-571.	1.1	37
83	Lack of On-Going Adaptations in the Soleus Muscle Activity During Walking in Patients Affected by Large-Fiber Neuropathy. Journal of Neurophysiology, 2005, 93, 3075-3085.	0.9	37
84	Variables influencing quality of life and disability in Charcot Marie Tooth (CMT) patients: Italian multicentre study. Neurological Sciences, 2006, 27, 417-423.	0.9	37
85	Correlation between clinical/neurophysiological findings and quality of life in Charcotâ€Marieâ€₹ooth type 1A. Journal of the Peripheral Nervous System, 2008, 13, 64-70.	1.4	37
86	Expanding sialidosis spectrum by genome-wide screening. Neurology, 2014, 82, 2003-2006.	1.5	37
87	Neurogenic muscle hypertrophy. Journal of Neurology, 1989, 236, 292-295.	1.8	36
88	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
89	Ascorbic acid for the treatment of Charcot-Marie-Tooth disease. The Cochrane Library, 2015, 2015, CD011952.	1.5	36
90	Very late onset Friedreich's ataxia without cardiomyopathy is associated with limited GAA expansion in the <i>X25</i> gene. Neurology, 1997, 49, 1153-1155.	1.5	35

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91	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
92	Detection of hereditary neuropathy with liability to pressure palsies among patients with acute painless mononeuropathy or plexopathy., 1998, 21, 1686-1691.		34
93	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
94	Variation in <i>SIPA1L2</i> is correlated with phenotype modification in Charcot– Marie– Tooth disease type 1A. Annals of Neurology, 2019, 85, 316-330.	2.8	33
95	Novel mutations in the HSN2 gene causing hereditary sensory and autonomic neuropathy type II. Neurology, 2006, 66, 748-751.	1.5	32
96	Charcot-Marie-Tooth and pain: correlations with neurophysiological, clinical, and disability findings. Neurological Sciences, 2008, 29, 193-194.	0.9	32
97	Clinical/Scientific Notes. Neurology, 2011, 77, 1401-1402.	1.5	32
98	Screening for <i>SH3TC2</i> 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
99	Neurofascin-155 as a putative antigen in combined central and peripheral demyelination. Neurology: Neuroimmunology and NeuroInflammation, 2016, 3, e238.	3.1	32
100	Deletion of the PMP22 gene and hereditary neuropathy with liability to pressure palsies. Current Opinion in Neurology, 1996, 9, 348-354.	1.8	31
101	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
102	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
103	Hereditary neuropathy with liability to pressure palsies. Journal of Neurology, 2020, 267, 2198-2206.	1.8	31
104	ATPase Domain <scp><i>AFG3L2</i></scp> Mutations Alter <scp>OPA1</scp> Processing and Cause Optic Neuropathy. Annals of Neurology, 2020, 88, 18-32.	2.8	31
105	Expanding the spectrum of genes responsible for hereditary motor neuropathies. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, 1171-1179.	0.9	30
106	CSF and Blood Levels of GFAP in Alexander Disease. ENeuro, 2015, 2, ENEURO.0080-15.2015.	0.9	30
107	Rapid progression of late onset axonal Charcot Marie Tooth disease associated with a novel MPZ mutation in the extracellular domain. Journal of Neurology, Neurosurgery and Psychiatry, 2007, 78, 1263-1266.	0.9	29
108	<i>GFAP </i> mutations and polymorphisms in 13 unrelated Italian patients affected by Alexander disease. Clinical Genetics, 2007, 72, 427-433.	1.0	29

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109	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
110	An essential role of MAG in mediating axon–myelin attachment in Charcot–Marie–Tooth 1A disease. Neurobiology of Disease, 2013, 49, 221-231.	2.1	29
111	Postural stabilization and balance assessment in Charcot–Marie–Tooth 1A subjects. Gait and Posture, 2014, 40, 481-486.	0.6	29
112	A longitudinal study of CMT1A using Rasch analysis based CMT neuropathy and examination scores. Neurology, 2020, 94, e884-e896.	1.5	29
113	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
114	Natural History and Treatment of Peripheral Inherited Neuropathies. Advances in Experimental Medicine and Biology, 2009, 652, 207-224.	0.8	26
115	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
116	Pain and small fiber function in charcot–marie–tooth disease type 1A. Muscle and Nerve, 2014, 50, 366-371.	1.0	26
117	Novel outcome measures for Charcotâ 'Marieâ 'Tooth disease: validation and reliability of the 6â€min walk test and StepWatch (sup > â,,¢ (sup > 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
118	F response and somatosensory and brainstem auditory evoked potential studies in HMSN type I and II Journal of Neurology, Neurosurgery and Psychiatry, 1992, 55, 1027-1031.	0.9	25
119	POEMS syndrome: relapse after successful autologous peripheral blood stem cell transplantation. Neuromuscular Disorders, 2007, 17, 980-982.	0.3	25
120	Hereditary predominantly motor neuropathies. Current Opinion in Neurology, 2009, 22, 451-459.	1.8	25
121	Reliability of instrumented movement analysis as outcome measure in Charcot–Marie–Tooth disease: Results from a multitask locomotor protocol. Gait and Posture, 2011, 34, 36-43.	0.6	25
122	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
123	Development and validation of the Charcot-Marie-Tooth Disease Infant Scale. Brain, 2018, 141, 3319-3330.	3.7	25
124	Neurologic symptoms after epidural anaesthesia. Report of three cases. Acta Anaesthesiologica Scandinavica, 1994, 38, 742-743.	0.7	24
125	Pachymeningeal involvement in POEMS syndrome: MRI and histopathological study. Journal of Neurology, Neurosurgery and Psychiatry, 2012, 83, 33-37.	0.9	24
126	Altered <scp>TDP</scp> â€43â€dependent splicing in <i><scp>HSPB</scp>8</i> â€related distal hereditary motor neuropathy and myofibrillar myopathy. European Journal of Neurology, 2018, 25, 154-163.	1.7	24

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127	hATTR Pathology: Nerve Biopsy Results from Italian Referral Centers. Brain Sciences, 2020, 10, 780.	1.1	24
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	Novel mutations in the GDAP1 gene in patients affected with early-onset axonal Charcot-Marie-Tooth type 4A. Neuromuscular Disorders, 2009, 19, 476-480.	0.3	23
130	A new mutation in GJC2 associated with subclinical leukodystrophy. Journal of Neurology, 2014, 261, 1929-1938.	1.8	23
131	Is overwork weakness relevant in Charcot–Marie–Tooth disease?. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 1354-1358.	0.9	23
132	Mutational mechanisms in <i><scp>MFN2</scp></i> àêrelated neuropathy: compound heterozygosity for recessive and semidominant mutations. Journal of the Peripheral Nervous System, 2015, 20, 380-386.	1.4	23
133	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
134	Relationship between clinical examination, Quality of Life, disability and depression in CMT patients: Italian Multicenter study. Neurological Sciences, 2008, 29, 157-162.	0.9	22
135	Balance impairment in pediatric charcot–marie–tooth disease. Muscle and Nerve, 2019, 60, 242-249.	1.0	22
136	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
137	Challenges in Treating Charcot-Marie-Tooth Disease and Related Neuropathies: Current Management and Future Perspectives. Brain Sciences, 2021, 11, 1447.	1.1	22
138	The effects of wallerian degeneration of the optic radiations demonstrated by MRI. Neuroradiology, 1992, 34, 323-325.	1.1	21
139	Does CMT1A homozygosity cause more severe disease with root hypertrophy and higher CSF proteins?. Neurology, 2003, 60, 1721-1722.	1.5	21
140	Late presentation of leucoencephalopathy with calcifications and cysts: report of two cases. Journal of Neurology, Neurosurgery and Psychiatry, 2008, 79, 1303-1304.	0.9	21
141	Posterior column ataxia with retinitis pigmentosa coexisting with sensoryâ€autonomic neuropathy and leukemia due to the homozygous p.Pro221Ser <i>FLVCR1 </i> mutation. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2017, 174, 732-739.	1.1	21
142	Clinical and Electrophysiological Aspects of Charcot-Marie-Tooth Disease. NeuroMolecular Medicine, 0, 8, 3-22.	1.8	21
143	Peripheral nerve involvement in Churg-Strauss syndrome. Journal of Neurology, 1992, 239, 317-321.	1.8	20
144	Mild functional effects of a novel GFAP mutant allele identified in a familial case of adult-onset Alexander disease. European Journal of Human Genetics, 2008, 16, 462-470.	1.4	20

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145	Xâ€linked Charcotâ€Marieâ€Tooth type 1: strokeâ€like presentation of a novel <i><scp>GJB1</scp></i> mutation. Journal of the Peripheral Nervous System, 2014, 19, 183-186.	1.4	20
146	Biomarkers predict outcome in Charcot-Marie-Tooth disease 1A. Journal of Neurology, Neurosurgery and Psychiatry, 2017, 88, 941-952.	0.9	20
147	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
148	The POEMS syndrome: Report of six cases. Italian Journal of Neurological Sciences, 1994, 15, 353-358.	0.1	19
149	Heterozygous Null Mutation in the P O Gene Associated with Mild Charcot-Marie-Tooth Disease. Annals of the New York Academy of Sciences, 1999, 883, 477-480.	1.8	19
150	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
151	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
152	Transcriptional role of androgen receptor in the expression of long non-coding RNA Sox2OT in neurogenesis. PLoS ONE, 2017, 12, e0180579.	1.1	19
153	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
154	Treadmill training in patients affected by Charcot–Marie–Tooth neuropathy: results of a multicenter, prospective, randomized, singleâ€blind, controlled study. European Journal of Neurology, 2020, 27, 280-287.	1.7	19
155	Symmetry of foot alignment and ankle flexibility in paediatric Charcot–Marie–Tooth disease. Clinical Biomechanics, 2012, 27, 744-747.	0.5	18
156	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
157	Mitochondrial disorders of the retinal ganglion cells and the optic nerve. Mitochondrion, 2018, 42, 1-10.	1.6	18
158	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
159	Natural history of Charcot–Marie-Tooth 2: 2-year follow-up of muscle strength, walking ability and quality of life. Neurological Sciences, 2010, 31, 175-178.	0.9	17
160	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
161	Adult leukoencephalopathies with prominent infratentorial involvement can be caused by Erdheim–Chester disease. Journal of Neurology, 2018, 265, 273-284.	1.8	17
162	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

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163	Anti AChR antibody: Relevance to diagnosis and clinical aspects of myasthenia gravis. Italian Journal of Neurological Sciences, 1988, 9, 141-145.	0.1	16
164	Parenchymatous neurosyphilis. Neurological Sciences, 2001, 22, 281-282.	0.9	16
165	Late-onset sporadic ataxia, pontine lesion, and retroperitoneal fibrosis: a case of Erdheim-Chester disease. Neurological Sciences, 2008, 29, 263-267.	0.9	16
166	Outcome measures for Charcotâ€Marieâ€Tooth disease: clinical and neurofunctional assessment in children. Journal of the Peripheral Nervous System, 2011, 16, 237-242.	1.4	16
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