

Lucio Santoro

List of Publications by Year in descending order

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Version: 2024-02-01

164
papers

6,926
citations

57758

44
h-index

74163

75
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167
all docs

167
docs citations

167
times ranked

8197
citing authors

#	ARTICLE	IF	CITATIONS
1	Human mitochondrial DNA deletions associated with mutations in the gene encoding Twinkle, a phage T7 gene 4-like protein localized in mitochondria. <i>Nature Genetics</i> , 2001, 28, 223-231.	21.4	803
2	Sensory deficit in Parkinson's disease: evidence of a cutaneous denervation. <i>Brain</i> , 2008, 131, 1903-1911.	7.6	326
3	Ascorbic acid in Charcot-Marie-Tooth disease type 1A (CMT-TRIAAL and CMT-TRAIUK): a double-blind randomised trial. <i>Lancet Neurology</i> , The, 2011, 10, 320-328.	10.2	222
4	Intravenous immunoglobulin versus intravenous methylprednisolone for chronic inflammatory demyelinating polyradiculoneuropathy: a randomised controlled trial. <i>Lancet Neurology</i> , The, 2012, 11, 493-502.	10.2	185
5	Quantification of myelinated endings and mechanoreceptors in human digital skin. <i>Annals of Neurology</i> , 2003, 54, 197-205.	5.3	184
6	The Heterogeneity of Early Parkinson's Disease: A Cluster Analysis on Newly Diagnosed Untreated Patients. <i>PLoS ONE</i> , 2013, 8, e70244.	2.5	150
7	Myelinated nerve endings in human skin. <i>Muscle and Nerve</i> , 2007, 35, 767-775.	2.2	133
8	Ross syndrome: a rare or a misknown disorder of thermoregulation? A skin innervation study on 12 subjects. <i>Brain</i> , 2006, 129, 2119-2131.	7.6	123
9	Functional involvement of central cholinergic circuits and visual hallucinations in Parkinson's disease. <i>Brain</i> , 2009, 132, 2350-2355.	7.6	115
10	Neuropathy and levodopa in Parkinson's disease: Evidence from a multicenter study. <i>Movement Disorders</i> , 2013, 28, 1391-1397.	3.9	114
11	A standardized clinical evaluation of patients affected by facioscapulohumeral muscular dystrophy: The FSHD clinical score. <i>Muscle and Nerve</i> , 2010, 42, 213-217.	2.2	113
12	Predictors of recovery of responsiveness in prolonged anoxic vegetative state. <i>Neurology</i> , 2013, 80, 464-470.	1.1	111
13	Atypical CIDP: diagnostic criteria, progression and treatment response. Data from the Italian CIDP Database. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2019, 90, 125-132.	1.9	108
14	Large-Scale Population Analysis Challenges the Current Criteria for the Molecular Diagnosis of Facioscapulohumeral Muscular Dystrophy. <i>American Journal of Human Genetics</i> , 2012, 90, 628-635.	6.2	104
15	Biallelic mutations in SORD cause a common and potentially treatable hereditary neuropathy with implications for diabetes. <i>Nature Genetics</i> , 2020, 52, 473-481.	21.4	97
16	Skin sympathetic adrenergic innervation: An immunofluorescence confocal study. <i>Annals of Neurology</i> , 2006, 59, 376-381.	5.3	93
17	Large scale genotype-phenotype analyses indicate that novel prognostic tools are required for families with facioscapulohumeral muscular dystrophy. <i>Brain</i> , 2013, 136, 3408-3417.	7.6	85
18	Novel ATP13A2 (PARK9) homozygous mutation in a family with marked phenotype variability. <i>Neurogenetics</i> , 2011, 12, 33-39.	1.4	84

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19	Gender differences in non-motor symptoms in early, drug naïve Parkinson's disease. Journal of Neurology, 2013, 260, 2849-2855.	3.6	83
20	Blockade of glutamate mGlu5 receptors in a rat model of neuropathic pain prevents early over-expression of pro-apoptotic genes and morphological changes in dorsal horn lamina II. Neuropharmacology, 2004, 46, 468-479.	4.1	78
21	Small fibers involvement in Friedreich's ataxia. Annals of Neurology, 2001, 50, 17-25.	5.3	77
22	Tremor in primary adult-onset dystonia: prevalence and associated clinical features. Journal of Neurology, Neurosurgery and Psychiatry, 2013, 84, 404-408.	1.9	71
23	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.	1.9	70
24	Interpreting Genetic Variants in Titin in Patients With Muscle Disorders. JAMA Neurology, 2018, 75, 557.	9.0	69
25	Loss of cutaneous large and small fibers in naïve and L-DOPA-treated PD patients. Neurology, 2017, 89, 776-784.	1.1	66
26	Evolution of Gastric Electrical Features and Gastric Emptying in Children with Duchenne and Becker Muscular Dystrophy. American Journal of Gastroenterology, 2005, 100, 695-702.	0.4	65
27	Charcot-Marie-Tooth disease: frequency of genetic subtypes in a Southern Italy population. Journal of the Peripheral Nervous System, 2014, 19, 292-298.	3.1	64
28	RFC1 expansions are a common cause of idiopathic sensory neuropathy. Brain, 2021, 144, 1542-1550.	7.6	63
29	Link between non-motor symptoms and cognitive dysfunctions in de novo, drug-naïve PD patients. Journal of Neurology, 2012, 259, 1808-1813.	3.6	60
30	Clinical expression of facioscapulohumeral muscular dystrophy in carriers of 1-3 D4Z4 reduced alleles: experience of the FSHD Italian National Registry. BMJ Open, 2016, 6, e007798.	1.9	60
31	Brain MRI features of congenital- and adult-form myotonic dystrophy type 1: case-control study. Neuromuscular Disorders, 2002, 12, 476-483.	0.6	58
32	Clinical and Neuropsychological Long-Term Outcomes After Late Recovery of Responsiveness: A Case Series. Archives of Physical Medicine and Rehabilitation, 2014, 95, 711-716.	0.9	57
33	The glycopeptide CSF114(Glc) detects serum antibodies in multiple sclerosis. Journal of Neuroimmunology, 2005, 167, 131-137.	2.3	56
34	A novel clinical tool to classify facioscapulohumeral muscular dystrophy phenotypes. Journal of Neurology, 2016, 263, 1204-1214.	3.6	55
35	Facioscapulohumeral muscular dystrophy: new insights from compound heterozygotes and implication for prenatal genetic counselling. Journal of Medical Genetics, 2012, 49, 171-178.	3.2	53
36	Centronuclear myopathy related to dynamin 2 mutations: Clinical, morphological, muscle imaging and genetic features of an Italian cohort. Neuromuscular Disorders, 2013, 23, 229-238.	0.6	53

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37	Cutaneous innervation of the human face as assessed by skin biopsy. <i>Journal of Anatomy</i> , 2013, 222, 161-169.	1.5	53
38	Validation of the Italian version of the Neuropathic Pain Symptom Inventory in peripheral nervous system diseases. <i>Neurological Sciences</i> , 2009, 30, 99-106.	1.9	51
39	Centronuclear myopathies: genotype-phenotype correlation and frequency of defined genetic forms in an Italian cohort. <i>Journal of Neurology</i> , 2015, 262, 1728-1740.	3.6	51
40	ATTRv amyloidosis Italian Registry: clinical and epidemiological data. <i>Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis</i> , 2020, 27, 259-265.	3.0	51
41	Pattern and significance of white matter abnormalities in myotonic dystrophy type 1: an MRI study. <i>Journal of Neurology</i> , 2002, 249, 1175-1182.	3.6	48
42	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]. <i>Pharmacological Research</i> , 2006, 54, 436-441.	7.1	47
43	Dilated Virchow-Robin Spaces in Myotonic Dystrophy: Frequency, Extent and Significance. <i>European Neurology</i> , 2001, 46, 131-139.	1.4	46
44	Age at onset and symptom spread in primary adult-onset blepharospasm and cervical dystonia. <i>Movement Disorders</i> , 2012, 27, 1447-1450.	3.9	46
45	Serum epidermal growth factor predicts cognitive functions in early, drug-naïve Parkinson's disease patients. <i>Journal of Neurology</i> , 2013, 260, 438-444.	3.6	46
46	Postganglionic sudomotor denervation in patients with multiple system atrophy. <i>Neurology</i> , 2014, 82, 2223-2229.	1.1	45
47	Friedreich's ataxia: electrophysiological and histological findings. <i>Acta Neurologica Scandinavica</i> , 1983, 67, 26-40.	2.1	44
48	Postexercise facilitation of motor evoked potentials following transcranial magnetic stimulation: A study in normal subjects. <i>Muscle and Nerve</i> , 2002, 25, 448-452.	2.2	44
49	Novel mutations in <i>dystonin</i> provide clues to the pathomechanisms of HSAN-VI. <i>Neurology</i> , 2017, 88, 2132-2140.	1.1	41
50	Autosomal dominant cerebellar ataxia type I. Clinical and molecular study in 36 Italian families including a comparison between SCA1 and SCA2 phenotypes. <i>Journal of the Neurological Sciences</i> , 1996, 142, 140-147.	0.6	40
51	Influence of GAA expansion size and disease duration on central nervous system impairment in Friedreich's ataxia: contribution to the understanding of the pathophysiology of the disease. <i>Clinical Neurophysiology</i> , 2000, 111, 1023-1030.	1.5	40
52	Anodal transcranial direct current stimulation of motor cortex does not ameliorate spasticity in multiple sclerosis. <i>Restorative Neurology and Neuroscience</i> , 2015, 33, 487-492.	0.7	39
53	Brain damage in glycogen storage disease type I. <i>Journal of Pediatrics</i> , 2004, 144, 637-642.	1.8	38
54	Electrophysiological and histological follow-up study in 15 Friedreich's ataxia patients. <i>Muscle and Nerve</i> , 1990, 13, 536-540.	2.2	37

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55	The Effect of Cerebellar Degeneration on Human Sensori-motor Plasticity. Brain Stimulation, 2015, 8, 1144-1150.	1.6	37
56	Autonomic dysfunction in children with Hirschsprung's disease. Digestive Diseases and Sciences, 1999, 44, 960-965.	2.3	36
57	Why do some Friedreich's ataxia patients retain tendon reflexes?. Journal of Neurology, 1999, 246, 353-357.	3.6	36
58	Motor cortex cholinergic dysfunction in CADASIL: A transcranial magnetic demonstration. Clinical Neurophysiology, 2008, 119, 351-355.	1.5	35
59	Subclinical neurological involvement does not develop if Wilson's disease is treated early. Parkinsonism and Related Disorders, 2016, 24, 15-19.	2.2	34
60	PMP22 messenger RNA levels in skin biopsies: testing the effectiveness of a Charcot-Marie-Tooth 1A biomarker. Brain, 2014, 137, 1614-1620.	7.6	33
61	Charcot-Marie-Tooth disease. Neurology, 2015, 85, 1202-1208.	1.1	33
62	Upper gastrointestinal tract motility in children with progressive muscular dystrophy. Journal of Pediatrics, 1992, 121, 720-724.	1.8	32
63	Is serum uric acid related to non-motor symptoms in de-novo Parkinson's disease patients?. Parkinsonism and Related Disorders, 2014, 20, 772-775.	2.2	32
64	The R495W mutation in SPG3A causes spastic paraplegia associated with axonal neuropathy. Journal of Neurology, 2005, 252, 901-903.	3.6	31
65	Small fiber involvement in spinobulbar muscular atrophy (Kennedy's disease). Muscle and Nerve, 2007, 36, 816-820.	2.2	31
66	Electrophysiological characterisation in hereditary spastic paraplegia type 5. Clinical Neurophysiology, 2011, 122, 819-822.	1.5	31
67	Epidermal innervation morphometry by immunofluorescence and bright-field microscopy. Journal of the Peripheral Nervous System, 2015, 20, 387-391.	3.1	30
68	Small nerve fiber involvement in CMT1A. Neurology, 2015, 84, 407-414.	1.1	30
69	Premature aging in Werner's syndrome spares the central nervous system. Neurobiology of Aging, 1996, 17, 325-330.	3.1	29
70	Neurophysiological evidence of corticospinal tract abnormality in patients with Parkin mutations. Journal of Neurology, 2006, 253, 275-279.	3.6	29
71	Anhidrosis in multiple system atrophy: A preganglionic sudomotor dysfunction?. Movement Disorders, 2008, 23, 885-888.	3.9	29
72	Adult-onset Alexander disease. Journal of Neurology, 2008, 255, 24-30.	3.6	28

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73	Increased peptidergic fibers as a potential cutaneous marker of pain in diabetic small fiber neuropathy. <i>Pain</i> , 2021, 162, 778-786.	4.2	28
74	Neuropathy in idiopathic Parkinson disease: An Iatrogenic problem?. <i>Annals of Neurology</i> , 2011, 69, 427-428.	5.3	27
75	A new Italian FHM2 family: Clinical aspects and functional analysis of the disease-associated mutation. <i>Cephalalgia</i> , 2011, 31, 808-819.	3.9	27
76	Executive functions are impaired in heterozygote patients with oculopharyngeal muscular dystrophy. <i>Journal of Neurology</i> , 2012, 259, 833-837.	3.6	27
77	Alterations of autophagy in the peripheral neuropathy Charcot-Marie-Tooth type 2B. <i>Autophagy</i> , 2018, 14, 1-12.	9.1	27
78	Alteration of the late endocytic pathway in Charcot-Marie-Tooth type 2B disease. <i>Cellular and Molecular Life Sciences</i> , 2021, 78, 351-372.	5.4	27
79	Modifications of brain tissue volumes in facioscapulohumeral dystrophy. <i>NeuroImage</i> , 2006, 32, 1237-1242.	4.2	26
80	Eye symptoms in relatives of patients with primary adult-onset dystonia. <i>Movement Disorders</i> , 2012, 27, 305-307.	3.9	26
81	The combined treatment with orbital and pretarsal botulinum toxin injections in the management of poorly responsive blepharospasm. <i>Neurological Sciences</i> , 2014, 35, 397-400.	1.9	26
82	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.	0.6	25
83	Elevated TGF- β 2 serum levels in Emery-Dreifuss Muscular Dystrophy: Implications for myocyte and tenocyte differentiation and fibrogenic processes. <i>Nucleus</i> , 2018, 9, 337-349.	2.2	25
84	Phenotypic Variability Among Patients With D4Z4 Reduced Allele Facioscapulohumeral Muscular Dystrophy. <i>JAMA Network Open</i> , 2020, 3, e204040.	5.9	25
85	Central cholinergic dysfunction in the adult form of Niemann Pick disease type C: a further link with Alzheimer's disease?. <i>Journal of Neurology</i> , 2014, 261, 804-808.	3.6	24
86	Estimating the impact of COVID-19 pandemic on services provided by Italian Neuromuscular Centers: an Italian Association of Myology survey of the acute phase. <i>Acta Myologica</i> , 2020, 39, 57-66.	1.5	24
87	Anti-GAD antibody ocular flutter: expanding the spectrum of autoimmune ocular motor disorders. <i>Journal of Neurology</i> , 2013, 260, 2675-2677.	3.6	23
88	Characterization of nigrostriatal dysfunction in spinocerebellar ataxia 17. <i>Movement Disorders</i> , 2006, 21, 872-875.	3.9	22
89	Short-latency afferent inhibition in patients with Parkinson's disease and freezing of gait. <i>Journal of Neural Transmission</i> , 2015, 122, 1533-1540.	2.8	22
90	Electrophysiological characterization of adult-onset Niemann-Pick type C disease. <i>Journal of the Neurological Sciences</i> , 2015, 348, 262-265.	0.6	22

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91	Neuropsychologic Assessment and Cognitive Rehabilitation in a Patient With Locked-In Syndrome and Left Neglect. Archives of Physical Medicine and Rehabilitation, 2010, 91, 498-502.	0.9	21
92	Autonomic nervous system involvement in a new CMT2B family. Journal of the Peripheral Nervous System, 2012, 17, 361-364.	3.1	20
93	Postural instability in Charcot-Marie-Tooth 1A disease. Gait and Posture, 2016, 49, 353-357.	1.4	20
94	Hirayama's disease: an Italian single center experience and review of the literature. Quantitative Imaging in Medicine and Surgery, 2016, 6, 364-373.	2.0	20
95	Early detection of biventricular involvement in myotonic dystrophy by tissue Doppler. International Journal of Cardiology, 2007, 118, 227-232.	1.7	19
96	Autoimmune Autonomic Ganglionopathy. Archives of Neurology, 2011, 68, 504.	4.5	19
97	Somatosensory Temporal Discrimination Threshold Is Increased in Patients with Cerebellar Atrophy. Cerebellum, 2013, 12, 456-459.	2.5	19
98	Atypical clinical and radiological presentation of cryptococcal choroid plexitis in an immunocompetent woman. Journal of the Neurological Sciences, 2013, 334, 180-182.	0.6	19
99	Small fiber pathology parallels disease progression in Parkinson disease: a longitudinal study. Acta Neuropathologica, 2018, 136, 501-503.	7.7	19
100	Interpretation of the Epigenetic Signature of Facioscapulohumeral Muscular Dystrophy in Light of Genotype-Phenotype Studies. International Journal of Molecular Sciences, 2020, 21, 2635.	4.1	18
101	Intrafamilial phenotype variation in Friedreich's disease: possible exceptions to diagnostic criteria. Journal of Neurology, 1991, 238, 147-150.	3.6	17
102	Amiodarone-induced experimental acute neuropathy in rats. Muscle and Nerve, 1992, 15, 788-795.	2.2	17
103	A refined physical and transcriptional map of the SPG9 locus on 10q23.3-q24.2. European Journal of Human Genetics, 2000, 8, 777-782.	2.8	17
104	Neurophysiological evaluation of motor corticospinal pathways by TMS in idiopathic early-onset Parkinson's disease. Clinical Neurophysiology, 2011, 122, 546-549.	1.5	17
105	Muscle pain syndromes and fibromyalgia: the role of muscle biopsy. Current Opinion in Supportive and Palliative Care, 2018, 12, 382-387.	1.3	17
106	A quantitative polymerase chain reaction (PCR) assay completely discriminates between Duchenne and Becker muscular dystrophy deletion carriers and normal females. Molecular and Cellular Probes, 1996, 10, 129-137.	2.1	16
107	A rare mutation in MYH7 gene occurs with overlapping phenotype. Biochemical and Biophysical Research Communications, 2015, 457, 262-266.	2.1	16
108	Large genotype-phenotype study in carriers of D4Z4 borderline alleles provides guidance for facioscapulohumeral muscular dystrophy diagnosis. Scientific Reports, 2020, 10, 21648.	3.3	16

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109	Familial aggregation of white matter lesions in myotonic dystrophy type 1. <i>Neuromuscular Disorders</i> , 2008, 18, 299-305.	0.6	15
110	Case of acute motor conduction block neuropathy (AMCBN). <i>Muscle and Nerve</i> , 2009, 39, 224-226.	2.2	15
111	Influence of comorbidities on the phenotype of patients affected by Charcot-Marie-Tooth neuropathy type 1A. <i>Neuromuscular Disorders</i> , 2013, 23, 902-906.	0.6	15
112	Differential trigeminal myelinated and unmyelinated nerve fiber involvement in FOSMN syndrome. <i>Neurology</i> , 2015, 84, 540-542.	1.1	15
113	Relevance of diagnostic investigations in chronic inflammatory demyelinating polyradiculoneuropathy: Data from the Italian CIDP database. <i>Journal of the Peripheral Nervous System</i> , 2020, 25, 152-161.	3.1	15
114	A 5-year clinical follow-up study from the Italian National Registry for FSHD. <i>Journal of Neurology</i> , 2021, 268, 356-366.	3.6	15
115	Chronic inflammatory demyelinating polyradiculoneuropathy: can a diagnosis be made in patients not fulfilling electrodiagnostic criteria?. <i>European Journal of Neurology</i> , 2021, 28, 620-629.	3.3	15
116	A novel autosomal dominant <i>GDAP1</i> mutation in an Italian CMT2 family. <i>Journal of the Peripheral Nervous System</i> , 2012, 17, 351-355.	3.1	13
117	Cutaneous sensory and autonomic denervation in CADASIL. <i>Neurology</i> , 2016, 86, 1039-1044.	1.1	13
118	Cervical dystonia patients display subclinical gait changes. <i>Parkinsonism and Related Disorders</i> , 2017, 43, 97-100.	2.2	13
119	Excitatory and inhibitory mechanisms in Wilson's disease: investigation with magnetic motor cortex stimulation. <i>Journal of the Neurological Sciences</i> , 2001, 192, 35-40.	0.6	12
120	Ross syndrome. <i>Neurology</i> , 2013, 80, 417-418.	1.1	12
121	Differential myelinated and unmyelinated sensory and autonomic skin nerve fiber involvement in patients with ophthalmic postherpetic neuralgia. <i>Frontiers in Neuroanatomy</i> , 2015, 9, 105.	1.7	12
122	An altered lipid metabolism characterizes Charcot-Marie-Tooth type 2B peripheral neuropathy. <i>Biochimica Et Biophysica Acta - Molecular and Cell Biology of Lipids</i> , 2020, 1865, 158805.	2.4	12
123	IN VIVO CONFOCAL MICROSCOPY OF MEISSNER CORPUSCLES AS A MEASURE OF SENSORY NEUROPATHY. <i>Neurology</i> , 2008, 71, 536-537.	1.1	11
124	Six-minute walk test is reliable and sensitive in detecting response to therapy in CIDP. <i>Journal of Neurology</i> , 2019, 266, 860-865.	3.6	11
125	Pregnancy in Charcot-Marie-Tooth disease. <i>Neurology</i> , 2020, 95, e3180-e3189.	1.1	11
126	Electrophysiological comparison between males and females in HNPP. <i>Neurological Sciences</i> , 2013, 34, 1429-1432.	1.9	10

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127	Intraepidermal nerve fiber analysis using immunofluorescence with and without confocal microscopy. Muscle and Nerve, 2015, 51, 501-504.	2.2	10
128	Long-term therapy with miglustat and cognitive decline in the adult form of Niemann-Pick disease type C: a case report. Neurological Sciences, 2018, 39, 1015-1019.	1.9	10
129	GDAP1 mutation in autosomal recessive Charcot-Marie-Tooth with pyramidal features. Journal of Neurology, 2006, 253, 1234-1235.	3.6	9
130	Two families with novel <i>PMP22</i> point mutations: genotype-phenotype correlation. Journal of the Peripheral Nervous System, 2009, 14, 208-212.	3.1	9
131	Perioral skin biopsy to study skeletal muscle protein expression. Muscle and Nerve, 2010, 41, 392-398.	2.2	9
132	In vivo evidence of cortical amyloid deposition in the adult form of Niemann Pick type C. Heliyon, 2019, 5, e02776.	3.2	9
133	Specific impairment of BAERs in Friedreich's ataxia. Journal of the Neurological Sciences, 1984, 65, 111-120.	0.6	8
134	Autosomal recessive progressive myoclonus epilepsy with ataxia and mental retardation. Journal of Neurology, 2005, 252, 897-900.	3.6	8
135	Nine-year case history of monofocal motor neuropathy. Muscle and Nerve, 2008, 38, 927-929.	2.2	8
136	Teaching Video Neuro Images : Clonus of the lower jaw. Neurology, 2014, 82, e96.	1.1	8
137	Postganglionic Sudomotor Assessment in Early Stage of Multiple System Atrophy and Parkinson Disease. Neurology, 2022, 98, .	1.1	8
138	Two new mutations in the myophosphorylase gene in Italian patients with McArdle's disease. Neuromuscular Disorders, 2002, 12, 498-500.	0.6	7
139	Phenotypic overlap in familial and sporadic primary adult-onset extracranial dystonia. Journal of Neurology, 2012, 259, 2414-2418.	3.6	7
140	Muscle fiber type disproportion (FTD) in a family with mutations in the <i>LMNA</i> gene. Muscle and Nerve, 2015, 51, 604-608.	2.2	7
141	Disruption of GABA(A)-mediated intracortical inhibition in patients with chorea-acanthocytosis. Neuroscience Letters, 2017, 654, 107-110.	2.1	7
142	RYR1 Sequence Variants in Myopathies: Expression and Functional Studies in Two Families. BioMed Research International, 2019, 2019, 1-13.	1.9	7
143	A novel family with axonal Charcot-Marie-Tooth disease caused by a mutation in the <i>EGR2</i> gene. Journal of the Peripheral Nervous System, 2019, 24, 219-223.	3.1	7
144	Cutaneous sensory and autonomic denervation in progressive supranuclear palsy. Neuropathology and Applied Neurobiology, 2021, 47, 653-663.	3.2	7

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145	Electrodiagnosis of Guillain-Barre syndrome in the International GBS Outcome Study: Differences in methods and reference values. <i>Clinical Neurophysiology</i> , 2022, 138, 231-240.	1.5	7
146	Cerebellar vermis aplasia: Patient report and exclusion of the candidate genes <i>EN2</i> and <i>ZIC1</i> . <i>American Journal of Medical Genetics, Part A</i> , 2005, 136A, 198-200.	1.2	6
147	Are novel outcome measures for Charcot-Marie-Tooth disease sensitive to change? The 6-minute walk test and StepWatch [®] Activity Monitor in a 12-month longitudinal study. <i>Neuromuscular Disorders</i> , 2019, 29, 310-316.	0.6	6
148	Different cortical excitability profiles in hereditary brain iron and copper accumulation. <i>Neurological Sciences</i> , 2020, 41, 679-685.	1.9	6
149	Possible contribution of vascular innervation to somatic sensory function. <i>Pain</i> , 2010, 151, 552-553.	4.2	5
150	Early changes of myocardial deformation properties in patients with dystrophia myotonica type 1: A three-dimensional Speckle Tracking echocardiographic study. <i>International Journal of Cardiology</i> , 2014, 176, 1094-1096.	1.7	5
151	Isolated intracranial <i>Mycobacterium avium</i> complex granulomas in an immune-competent man. <i>Journal of the Neurological Sciences</i> , 2015, 349, 264-265.	0.6	4
152	Insights into the pathogenesis of <i>ATP1A1</i> -related CMT disease using patient-specific iPSCs. <i>Journal of the Peripheral Nervous System</i> , 2019, 24, 330-339.	3.1	4
153	A Model to Study Myelinated Fiber Degeneration and Regeneration in Human Skin. <i>Annals of Neurology</i> , 2020, 87, 456-465.	5.3	4
154	Impact of environmental factors and physical activity on disability and quality of life in CIDP. <i>Journal of Neurology</i> , 2020, 267, 2683-2691.	3.6	4
155	Internodal length variability of dermal myelinated fibres. <i>Brain</i> , 2010, 133, e142-e142.	7.6	3
156	The occurrence of lateral shift in cervical dystonia. <i>Neurological Sciences</i> , 2017, 38, 683-686.	1.9	3
157	The impact of symptoms on daily life as perceived by patients with Charcot-Marie-Tooth type 1A disease. <i>Neurological Sciences</i> , 2022, 43, 559-563.	1.9	3
158	The neurophysiological lesson from the Italian CIDP database. <i>Neurological Sciences</i> , 2021, , 1.	1.9	3
159	A compound score to screen patients with hereditary transthyretin amyloidosis. <i>Journal of Neurology</i> , 2022, , .	3.6	3
160	Diabetes mellitus in Kearns-Sayre Syndrome: a case with a 10-year follow-up. <i>Diabetes Research and Clinical Practice</i> , 1995, 30, 233-235.	2.8	2
161	Thermosensitive hereditary neuropathy with liability to pressure palsy. <i>Muscle and Nerve</i> , 2011, 43, 448-449.	2.2	2
162	A case of congenital cataracts, facial dysmorphisms, neuropathy, and hyperkinetic movement disorder. <i>Movement Disorders</i> , 2013, 28, 559-560.	3.9	1

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163	Chronic inflammatory demyelinating polyneuropathy mimicking an acute painful diabetic neuropathy. Neurological Sciences, 2015, 36, 1509-1510.	1.9	1
164	Prolonged distal motor latency of median nerve does not improve diagnostic accuracy for CIDP. Journal of Neurology, 2021, , 1.	3.6	1