

Lucio Santoro

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163
papers

5,447
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39
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167
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ext. citations

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avg, IF

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L-index

#	Paper	IF	Citations
163	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, 223-31	36.3	704
162	Sensory deficit in Parkinson's disease: evidence of a cutaneous denervation. <i>Brain</i> , 2008 , 131, 1903-11	11.2	275
161	Ascorbic acid in Charcot-Marie-Tooth disease type 1A (CMT-TRIAAL and CMT-TRAUK): a double-blind randomised trial. <i>Lancet Neurology</i> , 2011 , 10, 320-8	24.1	184
160	Quantification of myelinated endings and mechanoreceptors in human digital skin. <i>Annals of Neurology</i> , 2003 , 54, 197-205	9.4	157
159	Intravenous immunoglobulin versus intravenous methylprednisolone for chronic inflammatory demyelinating polyradiculoneuropathy: a randomised controlled trial. <i>Lancet Neurology</i> , 2012 , 11, 493-502	24.1	153
158	Myelinated nerve endings in human skin. <i>Muscle and Nerve</i> , 2007 , 35, 767-75	3.4	120
157	The heterogeneity of early Parkinson's disease: a cluster analysis on newly diagnosed untreated patients. <i>PLoS ONE</i> , 2013 , 8, e70244	3.7	118
156	Ross syndrome: a rare or a misknown disorder of thermoregulation? A skin innervation study on 12 subjects. <i>Brain</i> , 2006 , 129, 2119-31	11.2	100
155	Functional involvement of central cholinergic circuits and visual hallucinations in Parkinson's disease. <i>Brain</i> , 2009 , 132, 2350-5	11.2	92
154	Neuropathy and levodopa in Parkinson's disease: evidence from a multicenter study. <i>Movement Disorders</i> , 2013 , 28, 1391-7	7	86
153	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, 628-35	11	86
152	A standardized clinical evaluation of patients affected by facioscapulohumeral muscular dystrophy: The FSHD clinical score. <i>Muscle and Nerve</i> , 2010 , 42, 213-7	3.4	84
151	Predictors of recovery of responsiveness in prolonged anoxic vegetative state. <i>Neurology</i> , 2013 , 80, 464-70		83
150	Skin sympathetic adrenergic innervation: an immunofluorescence confocal study. <i>Annals of Neurology</i> , 2006 , 59, 376-81	9.4	81
149	Novel ATP13A2 (PARK9) homozygous mutation in a family with marked phenotype variability. <i>Neurogenetics</i> , 2011 , 12, 33-9	3	72
148	Large scale genotype-phenotype analyses indicate that novel prognostic tools are required for families with facioscapulohumeral muscular dystrophy. <i>Brain</i> , 2013 , 136, 3408-17	11.2	71
147	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. <i>Neuropharmacology</i> , 2004 , 46, 468-79	5.5	71

146	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	5.5	67
145	Small fibers involvement in Friedreich's ataxia. <i>Annals of Neurology</i> , 2001 , 50, 17-25	9.4	65
144	Gender differences in non-motor symptoms in early, drug naïve Parkinson's disease. <i>Journal of Neurology</i> , 2013 , 260, 2849-55	5.5	61
143	Frequency and time to relapse after discontinuing 6-month therapy with IVIg or pulsed methylprednisolone in CIDP. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2015 , 86, 729-34	5.5	57
142	Tremor in primary adult-onset dystonia: prevalence and associated clinical features. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2013 , 84, 404-8	5.5	51
141	Charcot-Marie-Tooth disease: frequency of genetic subtypes in a Southern Italy population. <i>Journal of the Peripheral Nervous System</i> , 2014 , 19, 292-8	4.7	50
140	Evolution of gastric electrical features and gastric emptying in children with Duchenne and Becker muscular dystrophy. <i>American Journal of Gastroenterology</i> , 2005 , 100, 695-702	0.7	50
139	Loss of cutaneous large and small fibers in naïve and l-dopa-treated PD patients. <i>Neurology</i> , 2017 , 89, 776-784	6.5	49
138	The glycopeptide CSF114(Glc) detects serum antibodies in multiple sclerosis. <i>Journal of Neuroimmunology</i> , 2005 , 167, 131-7	3.5	49
137	Brain MRI features of congenital- and adult-form myotonic dystrophy type 1: case-control study. <i>Neuromuscular Disorders</i> , 2002 , 12, 476-83	2.9	48
136	Link between non-motor symptoms and cognitive dysfunctions in de novo, drug-naïve PD patients. <i>Journal of Neurology</i> , 2012 , 259, 1808-13	5.5	47
135	Centronuclear myopathy related to dynamin 2 mutations: clinical, morphological, muscle imaging and genetic features of an Italian cohort. <i>Neuromuscular Disorders</i> , 2013 , 23, 229-38	2.9	45
134	Clinical expression of facioscapulohumeral muscular dystrophy in carriers of 1-3 D4Z4 reduced alleles: experience of the FSHD Italian National Registry. <i>BMJ Open</i> , 2016 , 6, e007798	3	44
133	Cutaneous innervation of the human face as assessed by skin biopsy. <i>Journal of Anatomy</i> , 2013 , 222, 161-9	2.9	43
132	Postexercise facilitation of motor evoked potentials following transcranial magnetic stimulation: a study in normal subjects. <i>Muscle and Nerve</i> , 2002 , 25, 448-52	3.4	43
131	Centronuclear myopathies: genotype-phenotype correlation and frequency of defined genetic forms in an Italian cohort. <i>Journal of Neurology</i> , 2015 , 262, 1728-40	5.5	42
130	Interpreting Genetic Variants in Titin in Patients With Muscle Disorders. <i>JAMA Neurology</i> , 2018 , 75, 557-565	5.5	42
129	Facioscapulohumeral muscular dystrophy: new insights from compound heterozygotes and implication for prenatal genetic counselling. <i>Journal of Medical Genetics</i> , 2012 , 49, 171-8	5.8	42

128	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, 436-41	10.2	42
127	Friedreich's ataxia: electrophysiological and histological findings. <i>Acta Neurologica Scandinavica</i> , 1983 , 67, 26-40	3.8	42
126	Dilated Virchow-Robin spaces in myotonic dystrophy: frequency, extent and significance. <i>European Neurology</i> , 2001 , 46, 131-9	2.1	41
125	Validation of the Italian version of the Neuropathic Pain Symptom Inventory in peripheral nervous system diseases. <i>Neurological Sciences</i> , 2009 , 30, 99-106	3.5	40
124	Postganglionic sudomotor denervation in patients with multiple system atrophy. <i>Neurology</i> , 2014 , 82, 2223-9	6.5	39
123	Serum epidermal growth factor predicts cognitive functions in early, drug-naive Parkinson's disease patients. <i>Journal of Neurology</i> , 2013 , 260, 438-44	5.5	39
122	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, 140-7	3.2	39
121	A novel clinical tool to classify facioscapulohumeral muscular dystrophy phenotypes. <i>Journal of Neurology</i> , 2016 , 263, 1204-14	5.5	39
120	Biallelic mutations in SORD cause a common and potentially treatable hereditary neuropathy with implications for diabetes. <i>Nature Genetics</i> , 2020 , 52, 473-481	36.3	38
119	Clinical and neuropsychological long-term outcomes after late recovery of responsiveness: a case series. <i>Archives of Physical Medicine and Rehabilitation</i> , 2014 , 95, 711-6	2.8	38
118	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, 1023-30	4.3	37
117	Pattern and significance of white matter abnormalities in myotonic dystrophy type 1: an MRI study. <i>Journal of Neurology</i> , 2002 , 249, 1175-82	5.5	36
116	Electrophysiological and histological follow-up study in 15 Friedreich's ataxia patients. <i>Muscle and Nerve</i> , 1990 , 13, 536-40	3.4	35
115	Motor cortex cholinergic dysfunction in CADASIL: a transcranial magnetic demonstration. <i>Clinical Neurophysiology</i> , 2008 , 119, 351-5	4.3	33
114	Autonomic dysfunction in children with Hirschsprung's disease. <i>Digestive Diseases and Sciences</i> , 1999 , 44, 960-5	4	33
113	Why do some Friedreich's ataxia patients retain tendon reflexes? A clinical, neurophysiological and molecular study. <i>Journal of Neurology</i> , 1999 , 246, 353-7	5.5	32
112	Age at onset and symptom spread in primary adult-onset blepharospasm and cervical dystonia. <i>Movement Disorders</i> , 2012 , 27, 1447-50	7	31
111	The R495W mutation in SPG3A causes spastic paraplegia associated with axonal neuropathy. <i>Journal of Neurology</i> , 2005 , 252, 901-3	5.5	29

110	Electrophysiological characterisation in hereditary spastic paraplegia type 5. <i>Clinical Neurophysiology</i> , 2011 , 122, 819-22	4.3	28
109	Premature aging in Werner's syndrome spares the central nervous system. <i>Neurobiology of Aging</i> , 1996 , 17, 325-30	5.6	28
108	The Effect of Cerebellar Degeneration on Human Sensori-motor Plasticity. <i>Brain Stimulation</i> , 2015 , 8, 1144-50	5.1	27
107	Anodal transcranial direct current stimulation of motor cortex does not ameliorate spasticity in multiple sclerosis. <i>Restorative Neurology and Neuroscience</i> , 2015 , 33, 487-92	2.8	27
106	Small-fiber involvement in spinobulbar muscular atrophy (Kennedy's disease). <i>Muscle and Nerve</i> , 2007 , 36, 816-20	3.4	27
105	Anhidrosis in multiple system atrophy: a preganglionic sudomotor dysfunction?. <i>Movement Disorders</i> , 2008 , 23, 885-8	7	26
104	Brain damage in glycogen storage disease type I. <i>Journal of Pediatrics</i> , 2004 , 144, 637-42	3.6	26
103	Subclinical neurological involvement does not develop if Wilson's disease is treated early. <i>Parkinsonism and Related Disorders</i> , 2016 , 24, 15-9	3.6	25
102	Epidermal innervation morphometry by immunofluorescence and bright-field microscopy. <i>Journal of the Peripheral Nervous System</i> , 2015 , 20, 387-91	4.7	25
101	Neurophysiological evidence of corticospinal tract abnormality in patients with Parkin mutations. <i>Journal of Neurology</i> , 2006 , 253, 275-9	5.5	25
100	Upper gastrointestinal tract motility in children with progressive muscular dystrophy. <i>Journal of Pediatrics</i> , 1992 , 121, 720-4	3.6	25
99	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	2.7	25
98	Neuropathy in idiopathic Parkinson disease: an iatrogenic problem?. <i>Annals of Neurology</i> , 2011 , 69, 427-8; author reply 428-9	9.4	24
97	Modifications of brain tissue volumes in facioscapulohumeral dystrophy. <i>NeuroImage</i> , 2006 , 32, 1237-42	7.9	24
96	Novel mutations in provide clues to the pathomechanisms of HSAN-VI. <i>Neurology</i> , 2017 , 88, 2132-2140	6.5	23
95	PMP22 messenger RNA levels in skin biopsies: testing the effectiveness of a Charcot-Marie-Tooth 1A biomarker. <i>Brain</i> , 2014 , 137, 1614-20	11.2	23
94	Adult-onset Alexander disease : report on a family. <i>Journal of Neurology</i> , 2008 , 255, 24-30	5.5	23
93	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, 1003-17	2.9	22

92	Anti-GAD antibody ocular flutter: expanding the spectrum of autoimmune ocular motor disorders. <i>Journal of Neurology</i> , 2013 , 260, 2675-7	5.5	22
91	Executive functions are impaired in heterozygote patients with oculopharyngeal muscular dystrophy. <i>Journal of Neurology</i> , 2012 , 259, 833-7	5.5	22
90	Alterations of autophagy in the peripheral neuropathy Charcot-Marie-Tooth type 2B. <i>Autophagy</i> , 2018 , 14, 930-941	10.2	22
89	Small nerve fiber involvement in CMT1A. <i>Neurology</i> , 2015 , 84, 407-14	6.5	21
88	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, 804-8	5.5	21
87	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	3.5	21
86	Is serum uric acid related to non-motor symptoms in de-novo Parkinson's disease patients?. <i>Parkinsonism and Related Disorders</i> , 2014 , 20, 772-5	3.6	21
85	Eye symptoms in relatives of patients with primary adult-onset dystonia. <i>Movement Disorders</i> , 2012 , 27, 305-7	7	21
84	Charcot-Marie-Tooth disease: New insights from skin biopsy. <i>Neurology</i> , 2015 , 85, 1202-8	6.5	20
83	Short-latency afferent inhibition in patients with Parkinson's disease and freezing of gait. <i>Journal of Neural Transmission</i> , 2015 , 122, 1533-40	4.3	20
82	Characterization of nigrostriatal dysfunction in spinocerebellar ataxia 17. <i>Movement Disorders</i> , 2006 , 21, 872-5	7	20
81	Neuropsychologic assessment and cognitive rehabilitation in a patient with locked-in syndrome and left neglect. <i>Archives of Physical Medicine and Rehabilitation</i> , 2010 , 91, 498-502	2.8	19
80	A new Italian FHM2 family: clinical aspects and functional analysis of the disease-associated mutation. <i>Cephalalgia</i> , 2011 , 31, 808-19	6.1	19
79	Somatosensory temporal discrimination threshold is increased in patients with cerebellar atrophy. <i>Cerebellum</i> , 2013 , 12, 456-9	4.3	18
78	Electrophysiological characterization of adult-onset Niemann-Pick type C disease. <i>Journal of the Neurological Sciences</i> , 2015 , 348, 262-5	3.2	18
77	Early detection of biventricular involvement in myotonic dystrophy by tissue Doppler. <i>International Journal of Cardiology</i> , 2007 , 118, 227-32	3.2	18
76	Intrafamilial phenotype variation in Friedreich's disease: possible exceptions to diagnostic criteria. <i>Journal of Neurology</i> , 1991 , 238, 147-50	5.5	16
75	Elevated TGF- β serum levels in Emery-Dreifuss Muscular Dystrophy: Implications for myocyte and tenocyte differentiation and fibrogenic processes. <i>Nucleus</i> , 2018 , 9, 292-304	3.9	15

74	Postural instability in Charcot-Marie-Tooth 1A disease. <i>Gait and Posture</i> , 2016 , 49, 353-357	2.6	15
73	Familial aggregation of white matter lesions in myotonic dystrophy type 1. <i>Neuromuscular Disorders</i> , 2008 , 18, 299-305	2.9	15
72	A refined physical and transcriptional map of the SPG9 locus on 10q23.3-q24.2. <i>European Journal of Human Genetics</i> , 2000 , 8, 777-82	5.3	15
71	Amiodarone-induced experimental acute neuropathy in rats. <i>Muscle and Nerve</i> , 1992 , 15, 788-95	3.4	15
70	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	1.6	15
69	Increased peptidergic fibers as a potential cutaneous marker of pain in diabetic small fiber neuropathy. <i>Pain</i> , 2021 , 162, 778-786	8	15
68	Phenotypic Variability Among Patients With D4Z4 Reduced Allele Facioscapulohumeral Muscular Dystrophy. <i>JAMA Network Open</i> , 2020 , 3, e204040	10.4	14
67	Influence of comorbidities on the phenotype of patients affected by Charcot-Marie-Tooth neuropathy type 1A. <i>Neuromuscular Disorders</i> , 2013 , 23, 902-6	2.9	14
66	Atypical clinical and radiological presentation of cryptococcal choroid plexitis in an immunocompetent woman. <i>Journal of the Neurological Sciences</i> , 2013 , 334, 180-2	3.2	14
65	Autoimmune autonomic ganglionopathy: a possible postganglionic neuropathy. <i>Archives of Neurology</i> , 2011 , 68, 504-7		14
64	Small fiber pathology parallels disease progression in Parkinson disease: a longitudinal study. <i>Acta Neuropathologica</i> , 2018 , 136, 501-503	14.3	14
63	Hirayama's disease: an Italian single center experience and review of the literature. <i>Quantitative Imaging in Medicine and Surgery</i> , 2016 , 6, 364-373	3.6	13
62	Autonomic nervous system involvement in a new CMT2B family. <i>Journal of the Peripheral Nervous System</i> , 2012 , 17, 361-4	4.7	12
61	Neurophysiological evaluation of motor corticospinal pathways by TMS in idiopathic early-onset Parkinson's disease. <i>Clinical Neurophysiology</i> , 2011 , 122, 546-549	4.3	12
60	A quantitative polymerase chain reaction (PCR) assay completely discriminates between Duchenne and Becker muscular dystrophy deletion carriers and normal females. <i>Molecular and Cellular Probes</i> , 1996 , 10, 129-37	3.3	12
59	A rare mutation in MYH7 gene occurs with overlapping phenotype. <i>Biochemical and Biophysical Research Communications</i> , 2015 , 457, 262-6	3.4	11
58	RFC1 expansions are a common cause of idiopathic sensory neuropathy. <i>Brain</i> , 2021 , 144, 1542-1550	11.2	11
57	Differential trigeminal myelinated and unmyelinated nerve fiber involvement in FOSMN syndrome. <i>Neurology</i> , 2015 , 84, 540-2	6.5	10

56	Muscle pain syndromes and fibromyalgia: the role of muscle biopsy. <i>Current Opinion in Supportive and Palliative Care</i> , 2018 , 12, 382-387	2.6	10
55	A novel autosomal dominant GDAP1 mutation in an Italian CMT2 family. <i>Journal of the Peripheral Nervous System</i> , 2012 , 17, 351-5	4.7	10
54	Ross syndrome: a lesson from a monozygotic twin pair. <i>Neurology</i> , 2013 , 80, 417-8	6.5	10
53	Case of acute motor conduction block neuropathy (AMCBN). <i>Muscle and Nerve</i> , 2009 , 39, 224-6	3.4	10
52	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	3.2	10
51	Alteration of the late endocytic pathway in Charcot-Marie-Tooth type 2B disease. <i>Cellular and Molecular Life Sciences</i> , 2021 , 78, 351-372	10.3	10
50	Cutaneous sensory and autonomic denervation in CADASIL. <i>Neurology</i> , 2016 , 86, 1039-44	6.5	9
49	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	3.6	9
48	In vivo confocal microscopy of meissner corpuscles as a measure of sensory neuropathy. <i>Neurology</i> , 2008 , 71, 536-7; author reply 537	6.5	9
47	GDAP1 mutation in autosomal recessive Charcot-Marie-Tooth with pyramidal features. <i>Journal of Neurology</i> , 2006 , 253, 1234-5	5.5	9
46	A 5-year clinical follow-up study from the Italian National Registry for FSHD. <i>Journal of Neurology</i> , 2021 , 268, 356-366	5.5	9
45	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	6	9
44	Intraepidermal nerve fiber analysis using immunofluorescence with and without confocal microscopy. <i>Muscle and Nerve</i> , 2015 , 51, 501-4	3.4	8
43	Electrophysiological comparison between males and females in HNPP. <i>Neurological Sciences</i> , 2013 , 34, 1429-32	3.5	8
42	Two families with novel PMP22 point mutations: genotype-phenotype correlation. <i>Journal of the Peripheral Nervous System</i> , 2009 , 14, 208-12	4.7	8
41	Perioral skin biopsy to study skeletal muscle protein expression. <i>Muscle and Nerve</i> , 2010 , 41, 392-8	3.4	8
40	Specific impairment of BAER's in Friedreich's ataxia. Auditory evoked responses in clinical evaluation and differential diagnosis. <i>Journal of the Neurological Sciences</i> , 1984 , 65, 111-20	3.2	8
39	Large genotype-phenotype study in carriers of D4Z4 borderline alleles provides guidance for facioscapulohumeral muscular dystrophy diagnosis. <i>Scientific Reports</i> , 2020 , 10, 21648	4.9	8

38	RELEVANCE OF DIAGNOSTIC INVESTIGATIONS IN CHRONIC INFLAMMATORY DEMYELINATING POLIRADICULONEUROPATHY: DATA FROM THE ITALIAN CIDP DATABASE. <i>Journal of the Peripheral Nervous System</i> , 2020 , 25, 152	4.7	7
37	Cervical dystonia patients display subclinical gait changes. <i>Parkinsonism and Related Disorders</i> , 2017 , 43, 97-100	3.6	7
36	Teaching video neuroimages: clonus of the lower jaw: an old sign that comes back. <i>Neurology</i> , 2014 , 82, e96	6.5	7
35	Six-minute walk test is reliable and sensitive in detecting response to therapy in CIDP. <i>Journal of Neurology</i> , 2019 , 266, 860-865	5.5	6
34	Long-term therapy with miglustat and cognitive decline in the adult form of Niemann-Pick disease type C: a case report. <i>Neurological Sciences</i> , 2018 , 39, 1015-1019	3.5	6
33	Disruption of GABA(A)-mediated intracortical inhibition in patients with chorea-acanthocytosis. <i>Neuroscience Letters</i> , 2017 , 654, 107-110	3.3	6
32	Muscle fiber type disproportion (FTD) in a family with mutations in the LMNA gene. <i>Muscle and Nerve</i> , 2015 , 51, 604-8	3.4	6
31	Phenotypic overlap in familial and sporadic primary adult-onset extracranial dystonia. <i>Journal of Neurology</i> , 2012 , 259, 2414-8	5.5	6
30	Nine-year case history of monofocal motor neuropathy. <i>Muscle and Nerve</i> , 2008 , 38, 927-9	3.4	6
29	Cerebellar vermis aplasia: patient report and exclusion of the candidate genes EN2 and ZIC1. <i>American Journal of Medical Genetics, Part A</i> , 2005 , 136, 198-200	2.5	6
28	In vivo evidence of cortical amyloid deposition in the adult form of Niemann Pick type C. <i>Heliyon</i> , 2019 , 5, e02776	3.6	6
27	A novel family with axonal Charcot-Marie-Tooth disease caused by a mutation in the EGR2 gene. <i>Journal of the Peripheral Nervous System</i> , 2019 , 24, 219-223	4.7	5
26	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, 1094-6	3.2	5
25	Autosomal recessive progressive myoclonus epilepsy with ataxia and mental retardation. <i>Journal of Neurology</i> , 2005 , 252, 897-900	5.5	5
24	Two new mutations in the myophosphorylase gene in Italian patients with McArdle's disease. <i>Neuromuscular Disorders</i> , 2002 , 12, 498-500	2.9	5
23	Sequence Variants in Myopathies: Expression and Functional Studies in Two Families. <i>BioMed Research International</i> , 2019 , 2019, 7638946	3	4
22	Interpretation of the Epigenetic Signature of Facioscapulohumeral Muscular Dystrophy in Light of Genotype-Phenotype Studies. <i>International Journal of Molecular Sciences</i> , 2020 , 21,	6.3	4
21	Isolated intracranial Mycobacterium avium complex granulomas in an immune-competent man. <i>Journal of the Neurological Sciences</i> , 2015 , 349, 264-5	3.2	4

20	Postganglionic Sudomotor Assessment in Early Stage of Multiple System Atrophy and Parkinson Disease: A Morpho-functional Study.. <i>Neurology</i> , 2022 ,	6.5	4
19	Impact of environmental factors and physical activity on disability and quality of life in CIDP. <i>Journal of Neurology</i> , 2020 , 267, 2683-2691	5.5	3
18	Insights into the pathogenesis of ATP1A1-related CMT disease using patient-specific iPSCs. <i>Journal of the Peripheral Nervous System</i> , 2019 , 24, 330-339	4.7	3
17	Internodal length variability of dermal myelinated fibres. <i>Brain</i> , 2010 , 133, e142; author reply e143	11.2	3
16	Different cortical excitability profiles in hereditary brain iron and copper accumulation. <i>Neurological Sciences</i> , 2020 , 41, 679-685	3.5	3
15	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	5	3
14	Pregnancy in Charcot-Marie-Tooth disease: Data from the Italian CMT national registry. <i>Neurology</i> , 2020 , 95, e3180-e3189	6.5	3
13	Thermosensitive hereditary neuropathy with liability to pressure palsy. <i>Muscle and Nerve</i> , 2011 , 43, 448-454	9.4	2
12	Possible contribution of vascular innervation to somatic sensory function. <i>Pain</i> , 2010 , 151, 552-553	8	2
11	Diabetes mellitus in Kearns-Sayre syndrome: a case with a 10-year follow-up. <i>Diabetes Research and Clinical Practice</i> , 1995 , 30, 233-5	7.4	2
10	A Model to Study Myelinated Fiber Degeneration and Regeneration in Human Skin. <i>Annals of Neurology</i> , 2020 , 87, 456-465	9.4	2
9	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	2.9	2
8	Cutaneous sensory and autonomic denervation in progressive supranuclear palsy. <i>Neuropathology and Applied Neurobiology</i> , 2021 , 47, 653-663	5.2	2
7	The occurrence of lateral shift in cervical dystonia. <i>Neurological Sciences</i> , 2017 , 38, 683-686	3.5	1
6	Chronic inflammatory demyelinating polyneuropathy mimicking an acute painful diabetic neuropathy. <i>Neurological Sciences</i> , 2015 , 36, 1509-10	3.5	1
5	A case of congenital cataracts, facial dysmorphisms, neuropathy, and hyperkinetic movement disorder. <i>Movement Disorders</i> , 2013 , 28, 559-60	7	1
4	The impact of symptoms on daily life as perceived by patients with Charcot-Marie-Tooth type 1A disease. <i>Neurological Sciences</i> , 2021 , 1	3.5	1
3	The neurophysiological lesson from the Italian CIDP database. <i>Neurological Sciences</i> , 2021 , 1	3.5	1

- 2 Prolonged distal motor latency of median nerve does not improve diagnostic accuracy for CIDP.
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- 1 Electromyography **2016**, 21-37