Lucio Santoro

List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

163 papers	5,447 citations	39 h-index	66 g-index
167 ext. papers	6,307 ext. citations	5.5 avg, IF	4.83 L-index

#	Paper	IF	Citations
163	Postganglionic Sudomotor Assessment in Early Stage of Multiple System Atrophy and Parkinson Disease: A Morpho-functional Study.. *Neurology*, **2022**,	6.5	4
162	Increased peptidergic fibers as a potential cutaneous marker of pain in diabetic small fiber neuropathy. *Pain*, **2021**, 162, 778-786	8	15
161	The impact of symptoms on daily life as perceived by patients with Charcot-Marie-Tooth type 1A disease. *Neurological Sciences*, **2021**, 1	3.5	1
160	The neurophysiological lesson from the Italian CIDP database. *Neurological Sciences*, **2021**, 1	3.5	1
159	RFC1 expansions are a common cause of idiopathic sensory neuropathy. *Brain*, **2021**, 144, 1542-1550	11.2	11
158	Prolonged distal motor latency of median nerve does not improve diagnostic accuracy for CIDP. *Journal of Neurology*, **2021**, 1	5.5	0
157	Alteration of the late endocytic pathway in Charcot-Marie-Tooth type 2B disease. *Cellular and Molecular Life Sciences*, **2021**, 78, 351-372	10.3	10
156	A 5-year clinical follow-up study from the Italian National Registry for FSHD. *Journal of Neurology*, **2021**, 268, 356-366	5.5	9
155	Chronic inflammatory demyelinating polyradiculoneuropathy: can a diagnosis be made in patients not fulfilling electrodiagnostic criteria?. *European Journal of Neurology*, **2021**, 28, 620-629	6	9
154	Cutaneous sensory and autonomic denervation in progressive supranuclear palsy. *Neuropathology and Applied Neurobiology*, **2021**, 47, 653-663	5.2	2
153	Phenotypic Variability Among Patients With D4Z4 Reduced Allele Facioscapulohumeral Muscular Dystrophy. *JAMA Network Open*, **2020**, 3, e204040	10.4	14
152	Biallelic mutations in SORD cause a common and potentially treatable hereditary neuropathy with implications for diabetes. *Nature Genetics*, **2020**, 52, 473-481	36.3	38
151	Impact of environmental factors and physical activity on disability and quality of life in CIDP. *Journal of Neurology*, **2020**, 267, 2683-2691	5.5	3
150	RELEVANCE OF DIAGNOSTIC INVESTIGATIONS IN CHRONIC INFLAMMATORY DEMYELINATING POLIRADICULONEUROPATHY: DATA FROM THE ITALIAN CIDP DATABASE. *Journal of the Peripheral Nervous System*, **2020**, 25, 152	4.7	7
149	Interpretation of the Epigenetic Signature of Facioscapulohumeral Muscular Dystrophy in Light of Genotype-Phenotype Studies. *International Journal of Molecular Sciences*, **2020**, 21,	6.3	4
148	Estimating the impact of COVID-19 pandemic on services provided by Italian Neuromuscular Centers: an Italian Association of Myology survey of the acute phase. *Acta Myologica*, **2020**, 39, 57-66	1.6	15
147	A Model to Study Myelinated Fiber Degeneration and Regeneration in Human Skin. *Annals of Neurology*, **2020**, 87, 456-465	9.4	2

146	Different cortical excitability profiles in hereditary brain iron and copper accumulation. *Neurological Sciences*, **2020**, 41, 679-685	3.5	3
145	An altered lipid metabolism characterizes Charcot-Marie-Tooth type 2B peripheral neuropathy. *Biochimica Et Biophysica Acta - Molecular and Cell Biology of Lipids*, **2020**, 1865, 158805	5	3
144	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	2.7	25
143	Pregnancy in Charcot-Marie-Tooth disease: Data from the Italian CMT national registry. *Neurology*, **2020**, 95, e3180-e3189	6.5	3
142	Large genotype-phenotype study in carriers of D4Z4 borderline alleles provides guidance for facioscapulohumeral muscular dystrophy diagnosis. *Scientific Reports*, **2020**, 10, 21648	4.9	8
141	Six-minute walk test is reliable and sensitive in detecting response to therapy in CIDP. *Journal of Neurology*, **2019**, 266, 860-865	5.5	6
140	Sequence Variants in Myopathies: Expression and Functional Studies in Two Families. *BioMed Research International*, **2019**, 2019, 7638946	3	4
139	A novel family with axonal Charcot-Marie-Tooth disease caused by a mutation in the EGR2 gene. *Journal of the Peripheral Nervous System*, **2019**, 24, 219-223	4.7	5
138	Insights into the pathogenesis of ATP1A1-related CMT disease using patient-specific iPSCs. *Journal of the Peripheral Nervous System*, **2019**, 24, 330-339	4.7	3
137	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. *Neuromuscular Disorders*, **2019**, 29, 310-316	2.9	2
136	In vivo evidence of cortical amyloid deposition in the adult form of Niemann Pick type C. *Heliyon*, **2019**, 5, e02776	3.6	6
135	Atypical CIDP: diagnostic criteria, progression and treatment response. Data from the Italian CIDP Database. *Journal of Neurology, Neurosurgery and Psychiatry*, **2019**, 90, 125-132	5.5	67
134	Interpreting Genetic Variants in Titin in Patients With Muscle Disorders. *JAMA Neurology*, **2018**, 75, 557-565	17.2	42
133	Elevated TGF β serum levels in Emery-Dreifuss Muscular Dystrophy: Implications for myocyte and tenocyte differentiation and fibrogenic processes. *Nucleus*, **2018**, 9, 292-304	3.9	15
132	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	3.5	6
131	Muscle pain syndromes and fibromyalgia: the role of muscle biopsy. *Current Opinion in Supportive and Palliative Care*, **2018**, 12, 382-387	2.6	10
130	Small fiber pathology parallels disease progression in Parkinson disease: a longitudinal study. *Acta Neuropathologica*, **2018**, 136, 501-503	14.3	14
129	Alterations of autophagy in the peripheral neuropathy Charcot-Marie-Tooth type 2B. *Autophagy*, **2018**, 14, 930-941	10.2	22

128	The occurrence of lateral shift in cervical dystonia. *Neurological Sciences*, **2017**, 38, 683-686	3.5	1
127	Novel mutations in provide clues to the pathomechanisms of HSAN-VI. *Neurology*, **2017**, 88, 2132-2140	6.5	23
126	Loss of cutaneous large and small fibers in naive and l-dopa-treated PD patients. *Neurology*, **2017**, 89, 776-784	6.5	49
125	Disruption of GABA(A)-mediated intracortical inhibition in patients with chorea-acanthocytosis. *Neuroscience Letters*, **2017**, 654, 107-110	3.3	6
124	Cervical dystonia patients display subclinical gait changes. *Parkinsonism and Related Disorders*, **2017**, 43, 97-100	3.6	7
123	Postural instability in Charcot-Marie-Tooth 1A disease. *Gait and Posture*, **2016**, 49, 353-357	2.6	15
122	Cutaneous sensory and autonomic denervation in CADASIL. *Neurology*, **2016**, 86, 1039-44	6.5	9
121	Subclinical neurological involvement does not develop if Wilson's disease is treated early. *Parkinsonism and Related Disorders*, **2016**, 24, 15-9	3.6	25
120	Hirayama's disease: an Italian single center experience and review of the literature. *Quantitative Imaging in Medicine and Surgery*, **2016**, 6, 364-373	3.6	13
119	Electromyography **2016**, 21-37		
118	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	3	44
117	A novel clinical tool to classify facioscapulohumeral muscular dystrophy phenotypes. *Journal of Neurology*, **2016**, 263, 1204-14	5.5	39
116	Chronic inflammatory demyelinating polyneuropathy mimicking an acute painful diabetic neuropathy. *Neurological Sciences*, **2015**, 36, 1509-10	3.5	1
115	The Effect of Cerebellar Degeneration on Human Sensori-motor Plasticity. *Brain Stimulation*, **2015**, 8, 1144-50	5.1	27
114	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, 729-34	5.5	57
113	Small nerve fiber involvement in CMT1A. *Neurology*, **2015**, 84, 407-14	6.5	21
112	Differential trigeminal myelinated and unmyelinated nerve fiber involvement in FOSMN syndrome. *Neurology*, **2015**, 84, 540-2	6.5	10
111	Centronuclear myopathies: genotype-phenotype correlation and frequency of defined genetic forms in an Italian cohort. *Journal of Neurology*, **2015**, 262, 1728-40	5.5	42

110	Intraepidermal nerve fiber analysis using immunofluorescence with and without confocal microscopy. *Muscle and Nerve*, **2015**, 51, 501-4	3.4	8
109	Charcot-Marie-Tooth disease: New insights from skin biopsy. *Neurology*, **2015**, 85, 1202-8	6.5	20
108	Anodal transcranial direct current stimulation of motor cortex does not ameliorate spasticity in multiple sclerosis. *Restorative Neurology and Neuroscience*, **2015**, 33, 487-92	2.8	27
107	Epidermal innervation morphometry by immunofluorescence and bright-field microscopy. *Journal of the Peripheral Nervous System*, **2015**, 20, 387-91	4.7	25
106	Muscle fiber type disproportion (FTD) in a family with mutations in the LMNA gene. *Muscle and Nerve*, **2015**, 51, 604-8	3.4	6
105	Differential myelinated and unmyelinated sensory and autonomic skin nerve fiber involvement in patients with ophthalmic postherpetic neuralgia. *Frontiers in Neuroanatomy*, **2015**, 9, 105	3.6	9
104	Short-latency afferent inhibition in patients with Parkinson‗s disease and freezing of gait. *Journal of Neural Transmission*, **2015**, 122, 1533-40	4.3	20
103	Isolated intracranial Mycobacterium avium complex granulomas in an immune-competent man. *Journal of the Neurological Sciences*, **2015**, 349, 264-5	3.2	4
102	A rare mutation in MYH7 gene occurs with overlapping phenotype. *Biochemical and Biophysical Research Communications*, **2015**, 457, 262-6	3.4	11
101	Electrophysiological characterization of adult-onset Niemann-Pick type C disease. *Journal of the Neurological Sciences*, **2015**, 348, 262-5	3.2	18
100	Central cholinergic dysfunction in the adult form of Niemann Pick disease type C: a further link with Alzheimer‗s disease?. *Journal of Neurology*, **2014**, 261, 804-8	5.5	21
99	The combined treatment with orbital and pretarsal botulinum toxin injections in the management of poorly responsive blepharospasm. *Neurological Sciences*, **2014**, 35, 397-400	3.5	21
98	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, 1003-17	2.9	22
97	Early changes of myocardial deformation properties in patients with dystrophia myotonica type 1: a three-dimensional Speckle Tracking echocardiographic study. *International Journal of Cardiology*, **2014**, 176, 1094-6	3.2	5
96	Clinical and neuropsychological long-term outcomes after late recovery of responsiveness: a case series. *Archives of Physical Medicine and Rehabilitation*, **2014**, 95, 711-6	2.8	38
95	Postganglionic sudomotor denervation in patients with multiple system atrophy. *Neurology*, **2014**, 82, 2223-9	6.5	39
94	Is serum uric acid related to non-motor symptoms in de-novo Parkinson‗s disease patients?. *Parkinsonism and Related Disorders*, **2014**, 20, 772-5	3.6	21
93	PMP22 messenger RNA levels in skin biopsies: testing the effectiveness of a Charcot-Marie-Tooth 1A biomarker. *Brain*, **2014**, 137, 1614-20	11.2	23

92	Charcot-Marie-Tooth disease: frequency of genetic subtypes in a Southern Italy population. *Journal of the Peripheral Nervous System*, **2014**, 19, 292-8	4.7	50
91	Teaching video neuroimages: clonus of the lower jaw: an old sign that comes back. *Neurology*, **2014**, 82, e96	6.5	7
90	Serum epidermal growth factor predicts cognitive functions in early, drug-naive Parkinson's disease patients. *Journal of Neurology*, **2013**, 260, 438-44	5.5	39
89	Somatosensory temporal discrimination threshold is increased in patients with cerebellar atrophy. *Cerebellum*, **2013**, 12, 456-9	4.3	18
88	Gender differences in non-motor symptoms in early, drug naïve Parkinson's disease. *Journal of Neurology*, **2013**, 260, 2849-55	5.5	61
87	Neuropathy and levodopa in Parkinson's disease: evidence from a multicenter study. *Movement Disorders*, **2013**, 28, 1391-7	7	86
86	Tremor in primary adult-onset dystonia: prevalence and associated clinical features. *Journal of Neurology, Neurosurgery and Psychiatry*, **2013**, 84, 404-8	5.5	51
85	Centronuclear myopathy related to dynamin 2 mutations: clinical, morphological, muscle imaging and genetic features of an Italian cohort. *Neuromuscular Disorders*, **2013**, 23, 229-38	2.9	45
84	Influence of comorbidities on the phenotype of patients affected by Charcot-Marie-Tooth neuropathy type 1A. *Neuromuscular Disorders*, **2013**, 23, 902-6	2.9	14
83	Atypical clinical and radiological presentation of cryptococcal choroid plexitis in an immunocompetent woman. *Journal of the Neurological Sciences*, **2013**, 334, 180-2	3.2	14
82	Anti-GAD antibody ocular flutter: expanding the spectrum of autoimmune ocular motor disorders. *Journal of Neurology*, **2013**, 260, 2675-7	5.5	22
81	Cutaneous innervation of the human face as assessed by skin biopsy. *Journal of Anatomy*, **2013**, 222, 161-9	2.9	43
80	Electrophysiological comparison between males and females in HNPP. *Neurological Sciences*, **2013**, 34, 1429-32	3.5	8
79	Ross syndrome: a lesson from a monozygotic twin pair. *Neurology*, **2013**, 80, 417-8	6.5	10
78	Predictors of recovery of responsiveness in prolonged anoxic vegetative state. *Neurology*, **2013**, 80, 464-70	6.9	83
77	Large scale genotype-phenotype analyses indicate that novel prognostic tools are required for families with facioscapulohumeral muscular dystrophy. *Brain*, **2013**, 136, 3408-17	11.2	71
76	A case of congenital cataracts, facial dysmorphisms, neuropathy, and hyperkinetic movement disorder. *Movement Disorders*, **2013**, 28, 559-60	7	1
75	The heterogeneity of early Parkinson's disease: a cluster analysis on newly diagnosed untreated patients. *PLoS ONE*, **2013**, 8, e70244	3.7	118

74	Intravenous immunoglobulin versus intravenous methylprednisolone for chronic inflammatory demyelinating polyradiculoneuropathy: a randomised controlled trial. *Lancet Neurology, The*, **2012**, 11, 493-502	24.1	153
73	Eye symptoms in relatives of patients with primary adult-onset dystonia. *Movement Disorders*, **2012**, 27, 305-7	7	21
72	A novel autosomal dominant GDAP1 mutation in an Italian CMT2 family. *Journal of the Peripheral Nervous System*, **2012**, 17, 351-5	4.7	10
71	Autonomic nervous system involvement in a new CMT2B family. *Journal of the Peripheral Nervous System*, **2012**, 17, 361-4	4.7	12
70	Link between non-motor symptoms and cognitive dysfunctions in de novo, drug-naive PD patients. *Journal of Neurology*, **2012**, 259, 1808-13	5.5	47
69	Phenotypic overlap in familial and sporadic primary adult-onset extracranial dystonia. *Journal of Neurology*, **2012**, 259, 2414-8	5.5	6
68	Age at onset and symptom spread in primary adult-onset blepharospasm and cervical dystonia. *Movement Disorders*, **2012**, 27, 1447-50	7	31
67	Executive functions are impaired in heterozygote patients with oculopharyngeal muscular dystrophy. *Journal of Neurology*, **2012**, 259, 833-7	5.5	22
66	Large-scale population analysis challenges the current criteria for the molecular diagnosis of fascioscapulohumeral muscular dystrophy. *American Journal of Human Genetics*, **2012**, 90, 628-35	11	86
65	Facioscapulohumeral muscular dystrophy: new insights from compound heterozygotes and implication for prenatal genetic counselling. *Journal of Medical Genetics*, **2012**, 49, 171-8	5.8	42
64	Neurophysiological evaluation of motor corticospinal pathways by TMS in idiopathic early-onset Parkinson's disease. *Clinical Neurophysiology*, **2011**, 122, 546-549	4.3	12
63	Electrophysiological characterisation in hereditary spastic paraplegia type 5. *Clinical Neurophysiology*, **2011**, 122, 819-22	4.3	28
62	Ascorbic acid in Charcot-Marie-Tooth disease type 1A (CMT-TRIAAL and CMT-TRAUK): a double-blind randomised trial. *Lancet Neurology, The*, **2011**, 10, 320-8	24.1	184
61	Novel ATP13A2 (PARK9) homozygous mutation in a family with marked phenotype variability. *Neurogenetics*, **2011**, 12, 33-9	3	72
60	Thermosensitive hereditary neuropathy with liability to pressure palsy. *Muscle and Nerve*, **2011**, 43, 448-9	3.4	2
59	Neuropathy in idiopathic Parkinson disease: an iatrogenic problem?. *Annals of Neurology*, **2011**, 69, 427-8; author reply 428-9	9.4	24
58	A new Italian FHM2 family: clinical aspects and functional analysis of the disease-associated mutation. *Cephalalgia*, **2011**, 31, 808-19	6.1	19
57	Autoimmune autonomic ganglionopathy: a possible postganglionic neuropathy. *Archives of Neurology*, **2011**, 68, 504-7		14

56	Internodal length variability of dermal myelinated fibres. *Brain*, **2010**, 133, e142; author reply e143	11.2	3
55	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	2.8	19
54	Possible contribution of vascular innervation to somatic sensory function. *Pain*, **2010**, 151, 552-553	8	2
53	Perioral skin biopsy to study skeletal muscle protein expression. *Muscle and Nerve*, **2010**, 41, 392-8	3.4	8
52	A standardized clinical evaluation of patients affected by facioscapulohumeral muscular dystrophy: The FSHD clinical score. *Muscle and Nerve*, **2010**, 42, 213-7	3.4	84
51	Functional involvement of central cholinergic circuits and visual hallucinations in Parkinson's disease. *Brain*, **2009**, 132, 2350-5	11.2	92
50	Case of acute motor conduction block neuropathy (AMCBN). *Muscle and Nerve*, **2009**, 39, 224-6	3.4	10
49	Validation of the Italian version of the Neuropathic Pain Symptom Inventory in peripheral nervous system diseases. *Neurological Sciences*, **2009**, 30, 99-106	3.5	40
48	Two families with novel PMP22 point mutations: genotype-phenotype correlation. *Journal of the Peripheral Nervous System*, **2009**, 14, 208-12	4.7	8
47	Familial aggregation of white matter lesions in myotonic dystrophy type 1. *Neuromuscular Disorders* , **2008**, 18, 299-305	2.9	15
46	Motor cortex cholinergic dysfunction in CADASIL: a transcranial magnetic demonstration. *Clinical Neurophysiology*, **2008**, 119, 351-5	4.3	33
45	Sensory deficit in Parkinson's disease: evidence of a cutaneous denervation. *Brain*, **2008**, 131, 1903-11	11.2	275
44	In vivo confocal microscopy of meissner corpuscles as a measure of sensory neuropathy. *Neurology*, **2008**, 71, 536-7; author reply 537	6.5	9
43	Adult-onset Alexander disease : report on a family. *Journal of Neurology*, **2008**, 255, 24-30	5.5	23
42	Anhidrosis in multiple system atrophy: a preganglionic sudomotor dysfunction?. *Movement Disorders*, **2008**, 23, 885-8	7	26
41	Nine-year case history of monofocal motor neuropathy. *Muscle and Nerve*, **2008**, 38, 927-9	3.4	6
40	Myelinated nerve endings in human skin. *Muscle and Nerve*, **2007**, 35, 767-75	3.4	120
39	Small-fiber involvement in spinobulbar muscular atrophy (Kennedy's disease). *Muscle and Nerve*, **2007**, 36, 816-20	3.4	27

38	Early detection of biventricular involvement in myotonic dystrophy by tissue Doppler. *International Journal of Cardiology*, **2007**, 118, 227-32	3.2	18
37	Characterization of nigrostriatal dysfunction in spinocerebellar ataxia 17. *Movement Disorders*, **2006**, 21, 872-5	7	20
36	Skin sympathetic adrenergic innervation: an immunofluorescence confocal study. *Annals of Neurology*, **2006**, 59, 376-81	9.4	81
35	Modifications of brain tissue volumes in facioscapulohumeral dystrophy. *NeuroImage*, **2006**, 32, 1237-42	7.9	24
34	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, 436-41	10.2	42
33	Ross syndrome: a rare or a misknown disorder of thermoregulation? A skin innervation study on 12 subjects. *Brain*, **2006**, 129, 2119-31	11.2	100
32	Neurophysiological evidence of corticospinal tract abnormality in patients with Parkin mutations. *Journal of Neurology*, **2006**, 253, 275-9	5.5	25
31	GDAP1 mutation in autosomal recessive Charcot-Marie-Tooth with pyramidal features. *Journal of Neurology*, **2006**, 253, 1234-5	5.5	9
30	The glycopeptide CSF114(Glc) detects serum antibodies in multiple sclerosis. *Journal of Neuroimmunology*, **2005**, 167, 131-7	3.5	49
29	Cerebellar vermis aplasia: patient report and exclusion of the candidate genes EN2 and ZIC1. *American Journal of Medical Genetics, Part A*, **2005**, 136, 198-200	2.5	6
28	Autosomal recessive progressive myoclonus epilepsy with ataxia and mental retardation. *Journal of Neurology*, **2005**, 252, 897-900	5.5	5
27	The R495W mutation in SPG3A causes spastic paraplegia associated with axonal neuropathy. *Journal of Neurology*, **2005**, 252, 901-3	5.5	29
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	0.7	50
25	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, 468-79	5.5	71
24	Brain damage in glycogen storage disease type I. *Journal of Pediatrics*, **2004**, 144, 637-42	3.6	26
23	Quantification of myelinated endings and mechanoreceptors in human digital skin. *Annals of Neurology*, **2003**, 54, 197-205	9.4	157
22	Postexercise facilitation of motor evoked potentials following transcranial magnetic stimulation: a study in normal subjects. *Muscle and Nerve*, **2002**, 25, 448-52	3.4	43
21	Pattern and significance of white matter abnormalities in myotonic dystrophy type 1: an MRI study. *Journal of Neurology*, **2002**, 249, 1175-82	5.5	36

20	Two new mutations in the myophosphorylase gene in Italian patients with McArdle's disease. *Neuromuscular Disorders*, **2002**, 12, 498-500	2.9	5
19	Brain MRI features of congenital- and adult-form myotonic dystrophy type 1: case-control study. *Neuromuscular Disorders*, **2002**, 12, 476-83	2.9	48
18	Small fibers involvement in Friedreich's ataxia. *Annals of Neurology*, **2001**, 50, 17-25	9.4	65
17	Human mitochondrial DNA deletions associated with mutations in the gene encoding Twinkle, a phage T7 gene 4-like protein localized in mitochondria. *Nature Genetics*, **2001**, 28, 223-31	36.3	704
16	Dilated Virchow-Robin spaces in myotonic dystrophy: frequency, extent and significance. *European Neurology*, **2001**, 46, 131-9	2.1	41
15	Excitatory and inhibitory mechanisms in Wilson's disease: investigation with magnetic motor cortex stimulation. *Journal of the Neurological Sciences*, **2001**, 192, 35-40	3.2	10
14	A refined physical and transcriptional map of the SPG9 locus on 10q23.3-q24.2. *European Journal of Human Genetics*, **2000**, 8, 777-82	5.3	15
13	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. *Clinical Neurophysiology*, **2000**, 111, 1023-30	4.3	37
12	Autonomic dysfunction in children with Hirschsprung's disease. *Digestive Diseases and Sciences*, **1999**, 44, 960-5	4	33
11	Why do some Friedreich's ataxia patients retain tendon reflexes? A clinical, neurophysiological and molecular study. *Journal of Neurology*, **1999**, 246, 353-7	5.5	32
10	Premature aging in Werner's syndrome spares the central nervous system. *Neurobiology of Aging*, **1996**, 17, 325-30	5.6	28
9	Autosomal dominant cerebellar ataxia type I. Clinical and molecular study in 36 Italian families including a comparison between SCA1 and SCA2 phenotypes. *Journal of the Neurological Sciences*, **1996**, 142, 140-7	3.2	39
8	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, 129-37	3.3	12
7	Diabetes mellitus in Kearns-Sayre syndrome: a case with a 10-year follow-up. *Diabetes Research and Clinical Practice*, **1995**, 30, 233-5	7.4	2
6	Upper gastrointestinal tract motility in children with progressive muscular dystrophy. *Journal of Pediatrics*, **1992**, 121, 720-4	3.6	25
5	Amiodarone-induced experimental acute neuropathy in rats. *Muscle and Nerve*, **1992**, 15, 788-95	3.4	15
4	Intrafamilial phenotype variation in Friedreich's disease: possible exceptions to diagnostic criteria. *Journal of Neurology*, **1991**, 238, 147-50	5.5	16
3	Electrophysiological and histological follow-up study in 15 Friedreich's ataxia patients. *Muscle and Nerve*, **1990**, 13, 536-40	3.4	35

2	Specific impairment of BAERs in Friedreich's ataxia. Auditory evoked responses in clinical evaluation and differential diagnosis. *Journal of the Neurological Sciences*, **1984**, 65, 111-20	3.2	8
1	Friedreich's ataxia: electrophysiological and histological findings. *Acta Neurologica Scandinavica*, **1983**, 67, 26-40	3.8	42