Rocco Liguori

List of Publications by Year in Descending Order

250 papers	7,593 citations	46 h-index	75 g-index
261 ext. papers	8,863 ext. citations	5.1 avg, IF	5.46 L-index

#	Paper	IF	Citations
250	L-Acetylcarnitine causes analgesia in mice modeling Fabry disease by up-regulating type-2 metabotropic glutamate receptors.. *Molecular Pain*, **2022**, 17448069221087033	3.4	
249	Predicting functional impairment trajectories in amyotrophic lateral sclerosis: a probabilistic, multifactorial model of disease progression.. *Journal of Neurology*, **2022**, 1	5.5	1
248	Pilomotor seizures in autoimmune limbic encephalitis: description of two GAD65 antibodies-related cases and literature review.. *Seizure: the Journal of the British Epilepsy Association*, **2022**, 98, 71-78	3.2	0
247	Plasma and CSF Neurofilament Light Chain in Amyotrophic Lateral Sclerosis: A Cross-Sectional and Longitudinal Study. *Frontiers in Aging Neuroscience*, **2021**, 13, 753242	5.3	4
246	Cutaneous Sensory and Autonomic Small Fiber Neuropathy in HTRA1-Related Cerebral Small Vessel Disease. *Journal of Neuropathology and Experimental Neurology*, **2021**, 80, 713-716	3.1	0
245	Intravenous immunoglobulin therapy in COVID-19-related encephalopathy. *Journal of Neurology*, **2021**, 268, 2671-2675	5.5	13
244	Targeted sequencing panels in Italian ALS patients support different etiologies in the ALS/FTD continuum. *Journal of Neurology*, **2021**, 268, 3766-3776	5.5	1
243	In Vivo Diagnosis of Synucleinopathies: A Comparative Study of Skin Biopsy and RT-QuIC. *Neurology* , **2021**, 96, e2513-e2524	6.5	22
242	Brain MRS correlates with mitochondrial dysfunction biomarkers in MELAS-associated mtDNA mutations. *Annals of Clinical and Translational Neurology*, **2021**, 8, 1200-1211	5.3	3
241	Consistent skin □synuclein positivity in REM sleep behavior disorder - A two center two-to-four-year follow-up study. *Parkinsonism and Related Disorders*, **2021**, 86, 108-113	3.6	4
240	Reader Response: In Vivo Distribution of □Synuclein in Multiple Tissues and Biofluids in Parkinson Disease. *Neurology*, **2021**, 96, 964-965	6.5	3
239	Reviewing the Clinical Implications of Treating Narcolepsy as an Autoimmune Disorder. *Nature and Science of Sleep*, **2021**, 13, 557-577	3.6	2
238	RT-QuIC Detection of Pathological □Synuclein in Skin Punches of Patients with Lewy Body Disease. *Movement Disorders*, **2021**, 36, 2173-2177	7	14
237	Characterization of novel progranulin gene variants in Italian patients with neurodegenerative diseases. *Neurobiology of Aging*, **2021**, 97, 145.e7-145.e15	5.6	1
236	Brain metabolic correlates of apathy in amyotrophic lateral sclerosis: An 18F-FDG-positron emission tomography stud. *European Journal of Neurology*, **2021**, 28, 745-753	6	3
235	The In Vivo Diagnosis of Concomitant Alzheimer and Lewy Body Pathology: A Case Report. *Journal of Neuropathology and Experimental Neurology*, **2021**,	3.1	1
234	The clinical spectrum of multisystem proteinopathy: Data from a neurodegenerative cohort. *Journal of the Neurological Sciences*, **2021**, 426, 117478	3.2	2

233	Small Fiber Neuropathy in Patients with Chronic Pain and a Previous Diagnosis of Multiple Chemical Sensitivity Syndrome. *Journal of Neuropathology and Experimental Neurology*, **2021**, 80, 868-874	3.1	0
232	The m.3890G>A/MT-ND1 mtDNA rare pathogenic variant: Expanding clinical and MRI phenotypes. *Mitochondrion*, **2021**, 60, 142-149	4.9	1
231	Nociceptive behavior and central neuropeptidergic dysregulations in male and female mice of a Fabry disease animal model. *Brain Research Bulletin*, **2021**, 175, 158-167	3.9	0
230	Presence of Skin □Synuclein Deposits Discriminates Parkinson□s Disease from Progressive Supranuclear Palsy and Corticobasal Syndrome. *Journal of Parkinson□s Disease*, **2021**,	5.3	1
229	Movement Disorders Associated with GABA Receptor Encephalitis: A Video Case Report. *Movement Disorders Clinical Practice*, **2020**, 7, 681-683	2.2	1
228	A Longitudinal Skin Biopsy Study of Phosphorylated Alpha-Synuclein in a Patient With Parkinson Disease and Orthostatic Hypotension. *Journal of Neuropathology and Experimental Neurology*, **2020**, 79, 813-816	3.1	4
227	Skin Biopsy May Help to Distinguish Multiple System Atrophy-Parkinsonism from Parkinson□s Disease With Orthostatic Hypotension. *Movement Disorders*, **2020**, 35, 1649-1657	7	26
226	Calcium mishandling in absence of primary mitochondrial dysfunction drives cellular pathology in Wolfram Syndrome. *Scientific Reports*, **2020**, 10, 4785	4.9	16
225	Clinical Reasoning: Young woman with orbital pain and diplopia. *Neurology*, **2020**, 94, e752-e757	6.5	1
224	DNMT1 mutations leading to neurodegeneration paradoxically reflect on mitochondrial metabolism. *Human Molecular Genetics*, **2020**, 29, 1864-1881	5.6	10
223	Immunotherapy in Narcolepsy. *Current Treatment Options in Neurology*, **2020**, 22, 2	4.4	4
222	SSBP1 mutations cause mtDNA depletion underlying a complex optic atrophy disorder. *Journal of Clinical Investigation*, **2020**, 130, 108-125	15.9	49
221	Comparison of 123I-MIBG scintigraphy and phosphorylated □Synuclein skin deposits in synucleinopathies. *Parkinsonism and Related Disorders*, **2020**, 81, 48-53	3.6	9
220	Idiopathic Non-task-Specific Upper Limb Dystonia, a Neglected Form of Dystonia. *Movement Disorders*, **2020**, 35, 2038-2045	7	13
219	Chromatic Pupillometry Findings in Alzheimer□s Disease. *Frontiers in Neuroscience*, **2020**, 14, 780	5.1	5
218	Motor and Sensory Features of Cervical Dystonia Subtypes: Data From the Italian Dystonia Registry. *Frontiers in Neurology*, **2020**, 11, 906	4.1	3
217	Neuronal surface antibodies are common in children with narcolepsy and active movement disorders. *Journal of Neurology, Neurosurgery and Psychiatry*, **2020**,	5.5	1
216	Skin □Synuclein Aggregation Seeding Activity as a Novel Biomarker for Parkinson Disease. *JAMA Neurology*, **2020**,	17.2	52

215	Report of a novel ATP7A mutation causing distal motor neuropathy. *Neuromuscular Disorders*, **2019**, 29, 776-785	2.9	10
214	Combined brain positron emission tomography/magnetic resonance imaging in GABA receptor encephalitis. *European Journal of Neurology*, **2019**, 26, e88-e89	6	4
213	Subcutaneous immunoglobulin treatment and leucopenia in acquired demyelinating peripheral neuropathies. *European Journal of Neurology*, **2019**, 26, e80-e81	6	0
212	Broadening the Spectrum of Adulthood X-Linked Adrenoleukodystrophy: A Report of Two Atypical Cases. *Frontiers in Neurology*, **2019**, 10, 70	4.1	2
211	Abnormal □synuclein deposits in skin nerves: intra- and inter-laboratory reproducibility. *European Journal of Neurology*, **2019**, 26, 1245-1251	6	27
210	ZFYVE26/SPASTIZIN and SPG11/SPATACSIN mutations in hereditary spastic paraplegia types AR-SPG15 and AR-SPG11 have different effects on autophagy and endocytosis. *Autophagy*, **2019**, 15, 34-57	10.2	25
209	Biomarkers for REM sleep behavior disorder in idiopathic and narcoleptic patients. *Annals of Clinical and Translational Neurology*, **2019**, 6, 1872-1876	5.3	14
208	Persistence of limb dystonia and myoclonus during sleep in corticobasal syndrome: a case series. *Sleep Medicine*, **2019**, 59, 107-109	4.6	1
207	Reader response: Diffuse Lewy body disease manifesting as corticobasal syndrome: A rare form of Lewy body disease. *Neurology*, **2019**, 93, 411-412	6.5	1
206	The autonomic innervation of hairy skin in humans: an in vivo confocal study. *Scientific Reports*, **2019**, 9, 16982	4.9	17
205	Diagnostic criteria for amyotrophic lateral sclerosis: A multicentre study of inter-rater variation and sensitivity. *Clinical Neurophysiology*, **2019**, 130, 307-314	4.3	21
204	Loss of Swallow Tail Sign on Susceptibility-Weighted Imaging in Dementia with Lewy Bodies. *Journal of Alzheimer's Disease*, **2019**, 67, 61-65	4.3	11
203	Altered globotriaosylceramide accumulation and mucosal neuronal fiber density in the colon of the Fabry disease mouse model. *Neurogastroenterology and Motility*, **2019**, 31, e13529	4	2
202	The First Historically Reported Italian Family with FTD/ALS Teaches a Lesson on C9orf72 RE: Clinical Heterogeneity and Oligogenic Inheritance. *Journal of Alzheimer's Disease*, **2018**, 62, 687-697	4.3	4
201	The role of skin biopsy in differentiating small-fiber neuropathy from ganglionopathy. *European Journal of Neurology*, **2018**, 25, 848-853	6	28
200	Type 1 narcolepsy in anti-Hu antibodies mediated encephalitis: a case report. *Sleep Medicine*, **2018**, 52, 23-25	4.6	6
199	Riluzole and other prognostic factors in ALS: a population-based registry study in Italy. *Journal of Neurology*, **2018**, 265, 817-827	5.5	19
198	Subcutaneous immunoglobulin treatment and thromboembolic risk. *Annals of Allergy, Asthma and Immunology*, **2018**, 120, 433-435	3.2	3

197	Mitochondrial dysfunction in myotonic dystrophy type 1. *Neuromuscular Disorders*, **2018**, 28, 144-149	2.9	15
196	DGUOK recessive mutations in patients with CPEO, mitochondrial myopathy, parkinsonism and mtDNA deletions. *Brain*, **2018**, 141, e3	11.2	9
195	The incidental finding of elevated anti GQ1B antibodies in a patient with selective small fiber neuropathy. *Journal of the Neurological Sciences*, **2018**, 388, 192-194	3.2	5
194	Non-specific gastrointestinal features: Could it be Fabry disease?. *Digestive and Liver Disease*, **2018**, 50, 429-437	3.3	21
193	Accuracy of clinical diagnosis of dementia with Lewy bodies: a systematic review and meta-analysis. *Journal of Neurology, Neurosurgery and Psychiatry*, **2018**, 89, 358-366	5.5	67
192	Clonal expansion of mtDNA deletions: different disease models assessed by digital droplet PCR in single muscle cells. *Scientific Reports*, **2018**, 8, 11682	4.9	16
191	Peculiar combinations of individually non-pathogenic missense mitochondrial DNA variants cause low penetrance Leber's hereditary optic neuropathy. *PLoS Genetics*, **2018**, 14, e1007210	6	24
190	Nutraceutical Approach to Peripheral Neuropathies: Evidence from Clinical Trials. *Current Drug Metabolism*, **2018**, 19, 460-468	3.5	7
189	Skin -synuclein deposits differ in clinical variants of synucleinopathy: an in vivo study. *Scientific Reports*, **2018**, 8, 14246	4.9	50
188	Cerebrospinal Fluid Biomarkers in Patients with Frontotemporal Dementia Spectrum: A Single-Center Study. *Journal of Alzheimer's Disease*, **2018**, 66, 551-563	4.3	29
187	High frequency somatosensory stimulation in dystonia: Evidence fordefective inhibitory plasticity. *Movement Disorders*, **2018**, 33, 1902-1909	7	31
186	Skin Nerve Phosphorylated -Synuclein Deposits in Parkinson Disease With Orthostatic Hypotension. *Journal of Neuropathology and Experimental Neurology*, **2018**, 77, 942-949	3.1	24
185	The Italian Dystonia Registry: rationale, design and preliminary findings. *Neurological Sciences*, **2017** , 38, 819-825	3.5	22
184	Added value of electromyography in the diagnosis of myopathy: A consensus exercise. *Clinical Neurophysiology*, **2017**, 128, 697-701	4.3	9
183	Spine Topographical Distribution of Skin -Synuclein Deposits in Idiopathic Parkinson Disease. *Journal of Neuropathology and Experimental Neurology*, **2017**, 76, 384-389	3.1	26
182	Skin nerve phosphorylated -synuclein deposits in idiopathic REM sleep behavior disorder. *Neurology*, **2017**, 88, 2128-2131	6.5	84
181	The spectrum of REM sleep-related episodes in children with type 1 narcolepsy. *Brain*, **2017**, 140, 1669-1679	11.2	41
180	High frequency somatosensory stimulation increases sensori-motor inhibition and leads to perceptual improvement in healthy subjects. *Clinical Neurophysiology*, **2017**, 128, 1015-1025	4.3	32

179	Multiple variants in families with amyotrophic lateral sclerosis and frontotemporal dementia related to C9orf72 repeat expansion: further observations on their oligogenic nature. *Journal of Neurology*, **2017**, 264, 1426-1433	5.5	22
178	Modulation of the Muscle Activity During Sleep in Cervical Dystonia. *Sleep*, **2017**, 40,	1.1	12
177	Antibodies Against Hypocretin Receptor 2 Are Rare in Narcolepsy. *Sleep*, **2017**, 40,	1.1	24
176	Post-ganglionic autonomic neuropathy associated with anti-glutamic acid decarboxylase antibodies. *Clinical Autonomic Research*, **2017**, 27, 51-55	4.3	7
175	Paraneoplastic cerebellar degeneration and lambert-eaton myasthenia in a patient with merkel cell carcinoma and voltage-gated calcium channel antibodies. *Muscle and Nerve*, **2017**, 56, 998-1000	3.4	6
174	Absent cardiac and muscle sympathetic nerve activities involvement in Ross syndrome: A follow-up study. *Autonomic Neuroscience: Basic and Clinical*, **2017**, 208, 161-164	2.4	5
173	A new potential biomarker for dementia with Lewy bodies: Skin nerve □synuclein deposits. *Neurology*, **2017**, 89, 318-326	6.5	67
172	Skin globotriaosylceramide 3 deposits are specific to Fabry disease with classical mutations and associated with small fibre neuropathy. *PLoS ONE*, **2017**, 12, e0180581	3.7	14
171	Intermittent head drops: the differential spectrum. *Journal of Neurology, Neurosurgery and Psychiatry*, **2016**, 87, 414-9	5.5	8
170	Melanopsin retinal ganglion cell loss in Alzheimer disease. *Annals of Neurology*, **2016**, 79, 90-109	9.4	215
169	Increased expression of Trpv1 in peripheral terminals mediates thermal nociception in Fabry disease mouse model. *Molecular Pain*, **2016**, 12,	3.4	24
168	A multi-center, multinational age- and gender-adjusted normative dataset for immunofluorescent intraepidermal nerve fiber density at the distal leg. *European Journal of Neurology*, **2016**, 23, 333-8	6	77
167	Skin biopsy and microneurography disclose selective noradrenergic dysfunction due to dopamine-□hydroxylase deficiency. *Autonomic Neuroscience: Basic and Clinical*, **2016**, 197, 56-9	2.4	4
166	ALDH18A1 gene mutations cause dominant spastic paraplegia SPG9: loss of function effect and plausibility of a dominant negative mechanism. *Brain*, **2016**, 139, e3	11.2	30
165	Reply: Both mitochondrial DNA and mitonuclear gene mutations cause hearing loss through cochlear dysfunction. *Brain*, **2016**, 139, e34	11.2	4
164	Cutaneous sensory and autonomic denervation in CADASIL. *Neurology*, **2016**, 86, 1039-44	6.5	9
163	From state dissociation to status dissociatus. *Sleep Medicine Reviews*, **2016**, 28, 5-17	10.2	39
162	LOPED study: looking for an early diagnosis in a late-onset Pompe disease high-risk population. *Journal of Neurology, Neurosurgery and Psychiatry*, **2016**, 87, 5-11	5.5	39

161	Cervical demyelinating lesion presenting with choreoathetoid movements and dystonia. *Journal of the Neurological Sciences*, **2016**, 368, 203-5	3.2	2
160	Skin nerve misfolded □synuclein in pure autonomic failure and Parkinson disease. *Annals of Neurology*, **2016**, 79, 306-16	9.4	90
159	A longitudinal study of a family with adult-onset autosomal dominant leukodystrophy: Clinical, autonomic and neuropsychological findings. *Autonomic Neuroscience: Basic and Clinical*, **2016**, 195, 20-6	2.4	8
158	Parkinsonian tremor persisting during cataplexy. *Sleep Medicine*, **2016**, 17, 174-6	4.6	5
157	Relationship of white and gray matter abnormalities to clinical and genetic features in myotonic dystrophy type 1. *NeuroImage: Clinical*, **2016**, 11, 678-685	5.3	39
156	Pain in Fabry Disease: Practical Recommendations for Diagnosis and Treatment. *CNS Neuroscience and Therapeutics*, **2016**, 22, 568-76	6.8	50
155	Characterization of Human Dermal Fibroblasts in Fabry Disease. *Journal of Cellular Physiology*, **2016** , 231, 192-203	7	6
154	Mutations in SLC25A46, encoding a UGO1-like protein, cause an optic atrophy spectrum disorder. *Nature Genetics*, **2015**, 47, 926-32	36.3	131
153	Skin biopsy and I-123 MIBG scintigraphy findings in idiopathic Parkinson□ disease and parkinsonism: a comparative study. *Movement Disorders*, **2015**, 30, 986-9	7	17
152	Syndromic parkinsonism and dementia associated with OPA1 missense mutations. *Annals of Neurology*, **2015**, 78, 21-38	9.4	119
151	Non-paraneoplastic ataxia in a patient with contactin-associated protein-2 antibodies and benign course. *European Journal of Neurology*, **2015**, 22, e62-3	6	4
150	Heterogeneity in ALSFRS-R decline and survival: a population-based study in Italy. *Neurological Sciences*, **2015**, 36, 2243-52	3.5	24
149	OPA1-related auditory neuropathy: site of lesion and outcome of cochlear implantation. *Brain*, **2015**, 138, 563-76	11.2	58
148	Immunotherapy of oneiric stupor in Morvan syndrome: Efficacy documented by actigraphy. *Neurology*, **2015**, 84, 2457-9	6.5	8
147	De novo Diagnosis of Fabry Disease among Italian Adults with Acute Ischemic Stroke or Transient Ischemic Attack. *Journal of Stroke and Cerebrovascular Diseases*, **2015**, 24, 2588-95	2.8	14
146	An inflammatory myopathy unmasks a case of leprosy in an Italian patient. *Journal of Neurology*, **2015**, 262, 2179-81	5.5	3
145	Homozygous NOTCH3 null mutation and impaired NOTCH3 signaling in recessive early-onset arteriopathy and cavitating leukoencephalopathy. *EMBO Molecular Medicine*, **2015**, 7, 848-58	12	33
144	Nocturnal Sleep Dynamics Identify Narcolepsy Type 1. *Sleep*, **2015**, 38, 1277-84	1.1	56

143	Microneurographic recording from unmyelinated nerve fibers in neurological disorders: an update. *Clinical Neurophysiology*, **2015**, 126, 437-45	4.3	22
142	Italian recommendations for Lambert-Eaton myasthenic syndrome (LEMS) management. *Neurological Sciences*, **2014**, 35, 515-20	3.5	16
141	Quality of life in patients with craniocervical dystonia: Italian validation of the "Cervical Dystonia Impact Profile (CDIP-58)" and the "Craniocervical Dystonia Questionnaire (CDQ-24)". *Neurological Sciences*, **2014**, 35, 1053-8	3.5	3
140	Sympathetic and cardiovascular changes during sleep in narcolepsy with cataplexy patients. *Sleep Medicine*, **2014**, 15, 315-21	4.6	33
139	Efficient mitochondrial biogenesis drives incomplete penetrance in Leber's hereditary optic neuropathy. *Brain*, **2014**, 137, 335-53	11.2	186
138	Subcutaneous immunoglobulin in CIDP and MMN: a short-term nationwide study. *Journal of Neurology*, **2014**, 261, 2159-64	5.5	52
137	Epidemiology of amyotrophic lateral sclerosis in Emilia Romagna Region (Italy): A population based study. *Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration*, **2014**, 15, 262-8	3.6	34
136	Skin nerve □-synuclein deposits: a biomarker for idiopathic Parkinson disease. *Neurology*, **2014**, 82, 1362-9	6.5	201
135	Impairment of brain and muscle energy metabolism detected by magnetic resonance spectroscopy in hereditary spastic paraparesis type 28 patients with DDHD1 mutations. *Journal of Neurology*, **2014**, 261, 1789-93	5.5	19
134	Atypical late-onset hereditary spastic paraplegia with thin corpus callosum due to novel compound heterozygous mutations in the SPG11 gene. *Journal of Neurology*, **2014**, 261, 1825-7	5.5	11
133	Hypnic jerks: neurophysiological characterization of a new motor pattern. *Sleep Medicine*, **2014**, 15, 725-7	4.6	16
132	Cataplectic attacks during rapid eye movement sleep behavior disorder episodes in a narcoleptic patient. *Sleep Medicine*, **2014**, 15, 273-5	4.6	6
131	A novel in-frame 18-bp microdeletion in MT-CYB causes a multisystem disorder with prominent exercise intolerance. *Human Mutation*, **2014**, 35, 954-8	4.7	31
130	Polysomnographic and neurometabolic features may mark preclinical autosomal dominant cerebellar ataxia, deafness, and narcolepsy due to a mutation in the DNA (cytosine-5-)-methyltransferase gene, DNMT1. *Sleep Medicine*, **2014**, 15, 582-5	4.6	4
129	Pain related channels are differentially expressed in neuronal and non-neuronal cells of glabrous skin of fabry knockout male mice. *PLoS ONE*, **2014**, 9, e108641	3.7	35
128	Primary progressive narcolepsy type 1: the other side of the coin. *Neurology*, **2014**, 83, 2189-90	6.5	40
127	Narcolepsy is a common phenotype in HSAN IE and ADCA-DN. *Brain*, **2014**, 137, 1643-55	11.2	36
126	Lower wake resting sympathetic and cardiovascular activities in narcolepsy with cataplexy. *Neurology*, **2014**, 83, 1080-6	6.5	37

125	Pearls & Oy-sters: rapidly progressive dementia: prions or immunomediated?. *Neurology*, **2014**, 82, e149-52	8.3	3
124	Iodine-123 metaiodobenzylguanidine scintigraphy and iodine-123 ioflupane single photon emission computed tomography in Lewy body diseases: complementary or alternative techniques?. *Journal of Neuroimaging*, **2014**, 24, 149-54	2.8	36
123	Small nerve fiber involvement in patients referred for fibromyalgia. *Muscle and Nerve*, **2014**, 49, 757-9	3.4	75
122	Loss of temporal retinal nerve fibers in Parkinson disease: a mitochondrial pattern?. *European Journal of Neurology*, **2013**, 20, 198-201	6	76
121	Cybrid studies establish the causal link between the mtDNA m.3890G>A/MT-ND1 mutation and optic atrophy with bilateral brainstem lesions. *Biochimica Et Biophysica Acta - Molecular Basis of Disease*, **2013**, 1832, 445-52	6.9	15
120	Oct-1 recruitment to the nuclear envelope in adult-onset autosomal dominant leukodystrophy. *Biochimica Et Biophysica Acta - Molecular Basis of Disease*, **2013**, 1832, 411-20	6.9	22
119	Brachial amyotrophic diplegia associated with the a140a superoxide dismutase 1 mutation. *Neurogenetics*, **2013**, 14, 255-6	3	3
118	A novel pedigree with familial cortical myoclonic tremor and epilepsy (FCMTE): clinical characterization, refinement of the FCMTE2 locus, and confirmation of a founder haplotype. *Epilepsia*, **2013**, 54, 1298-306	6.4	16
117	Tremor in primary adult-onset dystonia: prevalence and associated clinical features. *Journal of Neurology, Neurosurgery and Psychiatry*, **2013**, 84, 404-8	5.5	51
116	Sleep disorders in patients with spinal cord injury. *Sleep Medicine Reviews*, **2013**, 17, 399-409	10.2	43
115	Skin sympathetic fiber □synuclein deposits: a potential biomarker for pure autonomic failure. *Neurology*, **2013**, 80, 725-32	6.5	56
114	Acute rhabdomyolysis induced by tonic-clonic epileptic seizures in a patient with glucose-6-phosphate dehydrogenase deficiency. *Journal of Neurology*, **2013**, 260, 2669-71	5.5	4
113	Idebenone treatment in patients with OPA1-mutant dominant optic atrophy. *Brain*, **2013**, 136, e231	11.2	53
112	A novel null homozygous mutation confirms CACNA2D2 as a gene mutated in epileptic encephalopathy. *PLoS ONE*, **2013**, 8, e82154	3.7	55
111	Development of a disability scale for myotonic dystrophy type 1. *Acta Neurologica Scandinavica*, **2012**, 125, 431-8	3.8	9
110	Eye symptoms in relatives of patients with primary adult-onset dystonia. *Movement Disorders*, **2012**, 27, 305-7	7	21
109	Phenotypic overlap in familial and sporadic primary adult-onset extracranial dystonia. *Journal of Neurology*, **2012**, 259, 2414-8	5.5	6
108	Muscle and skin sympathetic activities in Ross syndrome. *Clinical Neurophysiology*, **2012**, 123, 1639-43	4.3	12

107	Age at onset and symptom spread in primary adult-onset blepharospasm and cervical dystonia. *Movement Disorders*, **2012**, 27, 1447-50	7	31
106	Muscle sympathetic response to arousal predicts neurovascular reactivity during mental stress. *Journal of Physiology*, **2012**, 590, 2885-96	3.9	18
105	Pseudomyotonia in Romagnola cattle caused by novel ATP2A1 mutations. *BMC Veterinary Research*, **2012**, 8, 186	2.7	11
104	The empowerment of translational research: lessons from laminopathies. *Orphanet Journal of Rare Diseases*, **2012**, 7, 37	4.2	5
103	Rare primary mitochondrial DNA mutations and probable synergistic variants in Leber's hereditary optic neuropathy. *PLoS ONE*, **2012**, 7, e42242	3.7	60
102	Revisiting the issue of mitochondrial DNA content in optic mitochondriopathies. *Neurology*, **2012**, 79, 1517-9	6.5	12
101	Peripheral autonomic neuropathy: diagnostic contribution of skin biopsy. *Journal of Neuropathology and Experimental Neurology*, **2012**, 71, 1000-8	3.1	34
100	Microneurographic evaluation of sympathetic activity in small fiber neuropathy. *Clinical Neurophysiology*, **2011**, 122, 1854-9	4.3	14
99	Impact of medical audit on electrodiagnostic medicine in polyneuropathy. *Clinical Neurophysiology*, **2011**, 122, 2523-9	4.3	2
98	Isolated noradrenergic failure in adult-onset autosomal dominant leukodystrophy. *Autonomic Neuroscience: Basic and Clinical*, **2011**, 159, 123-6	2.4	19
97	Autonomic disturbances in narcolepsy. *Sleep Medicine Reviews*, **2011**, 15, 187-96	10.2	60
96	Somatic and autonomic small fiber neuropathy induced by bortezomib therapy: an immunofluorescence study. *Neurological Sciences*, **2011**, 32, 361-3	3.5	46
95	Methods of sudomotor innervation quantification. *Muscle and Nerve*, **2011**, 43, 920-1; author reply 921	3.4	1
94	Defective mitochondrial adenosine triphosphate production in skeletal muscle from patients with dominant optic atrophy due to OPA1 mutations. *Archives of Neurology*, **2011**, 68, 67-73		32
93	Idebenone treatment in Leber's hereditary optic neuropathy. *Brain*, **2011**, 134, e188	11.2	149
92	A clinically complex form of dominant optic atrophy (OPA8) maps on chromosome 16. *Human Molecular Genetics*, **2011**, 20, 1893-905	5.6	35
91	Environmental risk factors and clinical phenotype in familial and sporadic primary blepharospasm. *Neurology*, **2011**, 77, 631-7	6.5	31
90	Autonomic innervation in multiple system atrophy and pure autonomic failure. *Journal of Neurology, Neurosurgery and Psychiatry*, **2010**, 81, 1327-35	5.5	58

89	Sleep-related periodic respiration with central sleep apnea in Leber Hereditary Optic Neuropathy (LHON). *Sleep Medicine*, **2010**, 11, 426-7	4.6	3
88	Variation in the neurophysiological examination of amyotrophic lateral sclerosis in Europe. *Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders*, **2010**, 11, 443-8		4
87	Small fiber neuropathy in female patients with fabry disease. *Muscle and Nerve*, **2010**, 41, 409-12	3.4	40
86	Lower limb involvement in adult-onset primary dystonia: frequency and clinical features. *European Journal of Neurology*, **2010**, 17, 242-6	6	21
85	Visual system involvement in patients with Friedreich's ataxia. *Brain*, **2009**, 132, 116-23	11.2	117
84	Axial myoclonus in Devic neuromyelitis optica. *Movement Disorders*, **2009**, 24, 1708-9	7	6
83	A case of fatal familial insomnia in Africa. *Journal of Neurology*, **2009**, 256, 1778-9	5.5	10
82	Autosomal recessive hereditary spastic paraplegia with thin corpus callosum: a novel mutation in the SPG11 gene and further evidence for genetic heterogeneity. *European Journal of Neurology*, **2009**, 16, 121-6	6	12
81	Agrypnia Excitata: a microneurographic study of muscle sympathetic nerve activity. *Clinical Neurophysiology*, **2009**, 120, 1139-42	4.3	16
80	A defective SERCA1 protein is responsible for congenital pseudomyotonia in Chianina cattle. *American Journal of Pathology*, **2009**, 174, 565-73	5.8	27
79	Refinement of the SPG9 locus on chromosome 10q23.3-24.2 and exclusion of candidate genes. *European Journal of Neurology*, **2008**, 15, 520-4	6	6
78	Erythematous papules on the parasternal region in a 76-year-old man. *Clinical and Experimental Dermatology*, **2008**, 33, 369-70	1.8	1
77	Sympathetic and cardiovascular activity during cataplexy in narcolepsy. *Journal of Sleep Research*, **2008**, 17, 458-63	5.8	26
76	Differential cerebro spinal fluid proteome investigation of Leber hereditary optic neuropathy (LHON) and multiple sclerosis. *Journal of Neuroimmunology*, **2008**, 193, 156-60	3.5	25
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