Rocco Liguori

List of Publications by Year
in descending order

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259	10,037	38742	53230
papers	citations	50 h-index	85 g-index

261	261	261	10802
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Chromatic Pupillometry in Isolated Rapid Eye Movement Sleep Behavior Disorder. Movement Disorders, 2022, 37, 205-210.	3.9	9
2	Molecular biomarkers correlate with brain grey and white matter changes in patients with mitochondrial m.3243AÂ>ÂG mutation. Molecular Genetics and Metabolism, 2022, 135, 72-81.	1.1	3
3	Presence of Skin Î±-Synuclein Deposits Discriminates Parkinsonâ€™s Disease from Progressive Supranuclear Palsy and Corticobasal Syndrome. Journal of Parkinson's Disease, 2022, 12, 585-591.	2.8	9
4	L-Acetylcarnitine causes analgesia in mice modeling Fabry disease by up-regulating type-2 metabotropic glutamate receptors. Molecular Pain, 2022, 18, 174480692210870.	2.1	4
5	Predicting functional impairment trajectories in amyotrophic lateral sclerosis: a probabilistic, multifactorial model of disease progression. Journal of Neurology, 2022, 269, 3858-3878.	3.6	7
6	Epidemiological, Clinical and Genetic Features of ALS in the Last Decade: A Prospective Population-Based Study in the Emilia Romagna Region of Italy. Biomedicines, 2022, 10, 819.	3.2	10
7	Identifying and predicting amyotrophic lateral sclerosis clinical subgroups: a population-based machine-learning study. The Lancet Digital Health, 2022, 4, e359-e369.	12.3	19
8	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.	2.0	2
9	The Effect of Curcumin on Idiopathic Parkinson Disease: A Clinical and Skin Biopsy Study. Journal of Neuropathology and Experimental Neurology, 2022, 81, 545-552.	1.7	11
10	Clinical characteristics of a large cohort of patients with narcolepsy candidate for pitolisant: a cross-sectional study from the Italian PASS WakixÂ® Cohort. Neurological Sciences, 2022, 43, 5563-5574.	1.9	7
11	<scp>TWNK</scp> in Parkinson's Disease: A Movement Disorder and Mitochondrial Disease Center Perspective Study. Movement Disorders, 2022, 37, 1938-1943.	3.9	10
12	Intravenous immunoglobulin therapy in COVID-19-related encephalopathy. Journal of Neurology, 2021, 268, 2671-2675.	3.6	35
13	Characterization of novel progranulin gene variants in Italian patients with neurodegenerative diseases. Neurobiology of Aging, 2021, 97, 145.e7-145.e15.	3.1	4
14	Brain metabolic correlates of apathy in amyotrophic lateral sclerosis: An 18Fâ€FDGâ€positron emission tomography stud. European Journal of Neurology, 2021, 28, 745-753.	3.3	10
15	Neuronal surface antibodies are common in children with narcolepsy and active movement disorders. Journal of Neurology, Neurosurgery and Psychiatry, 2021, 92, 111-112.	1.9	2
16	Skin Î±-Synuclein Aggregation Seeding Activity as a Novel Biomarker for Parkinson Disease. JAMA Neurology, 2021, 78, 30.	9.0	125
17	Targeted sequencing panels in Italian ALS patients support different etiologies in the ALS/FTD continuum. Journal of Neurology, 2021, 268, 3766-3776.	3.6	12
18	In Vivo Diagnosis of Synucleinopathies. Neurology, 2021, 96, e2513-e2524.	1.1	63

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19	Brain MRS correlates with mitochondrial dysfunction biomarkers in MELASâ€associated mtDNA mutations. Annals of Clinical and Translational Neurology, 2021, 8, 1200-1211.	3.7	10
20	Consistent skin Î±-synuclein positivity in REM sleep behavior disorder â€“ A two center two-to-four-year follow-up study. Parkinsonism and Related Disorders, 2021, 86, 108-113.	2.2	16
21	Reader Response: In Vivo Distribution of Î±-Synuclein in Multiple Tissues and Biofluids in Parkinson Disease. Neurology, 2021, 96, 964-965.	1.1	4
22	Reviewing the Clinical Implications of Treating Narcolepsy as an Autoimmune Disorder. Nature and Science of Sleep, 2021, Volume 13, 557-577.	2.7	10
23	<scp>RTâ€QuIC</scp> Detection of Pathological Î±â€Synuclein in Skin Punches of Patients with Lewy Body Disease. Movement Disorders, 2021, 36, 2173-2177.	3.9	56
24	The In Vivo Diagnosis of Concomitant Alzheimer and Lewy Body Pathology: A Case Report. Journal of Neuropathology and Experimental Neurology, 2021, 80, 1085-1087.	1.7	1
25	The clinical spectrum of multisystem proteinopathy: Data from a neurodegenerative cohort. Journal of the Neurological Sciences, 2021, 426, 117478.	0.6	3
26	Presynaptic Paraneoplastic Disorders of the Neuromuscular Junction: An Update. Brain Sciences, 2021, 11, 1035.	2.3	6
27	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.	1.7	1
28	The m.3890G>A/MT-ND1 mtDNA rare pathogenic variant: Expanding clinical and MRI phenotypes. Mitochondrion, 2021, 60, 142-149.	3.4	4
29	Nociceptive behavior and central neuropeptidergic dysregulations in male and female mice of a Fabry disease animal model. Brain Research Bulletin, 2021, 175, 158-167.	3.0	3
30	Plasma and CSF Neurofilament Light Chain in Amyotrophic Lateral Sclerosis: A Cross-Sectional and Longitudinal Study. Frontiers in Aging Neuroscience, 2021, 13, 753242.	3.4	33
31	Cutaneous Sensory and Autonomic Small Fiber Neuropathy in HTRA1-Related Cerebral Small Vessel Disease. Journal of Neuropathology and Experimental Neurology, 2021, 80, 713-716.	1.7	2
32	Comparison of 123I-MIBG scintigraphy and phosphorylated Î±-synuclein skin deposits in synucleinopathies. Parkinsonism and Related Disorders, 2020, 81, 48-53.	2.2	22
33	Idiopathic <scp>Nonâ€taskâ€Specific</scp> Upper Limb Dystonia, a Neglected Form of Dystonia. Movement Disorders, 2020, 35, 2038-2045.	3.9	21
34	Chromatic Pupillometry Findings in Alzheimerâ€™s Disease. Frontiers in Neuroscience, 2020, 14, 780.	2.8	15
35	Motor and Sensory Features of Cervical Dystonia Subtypes: Data From the Italian Dystonia Registry. Frontiers in Neurology, 2020, 11, 906.	2.4	6
36	Movement Disorders Associated with GABA A Receptor Encephalitis: A Video Case Report. Movement Disorders Clinical Practice, 2020, 7, 681-683.	1.5	3

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37	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.	1.7	7
38	Skin Biopsy May Help to Distinguish Multiple System Atrophyâ€"Parkinsonism from Parkinson's Disease With Orthostatic Hypotension. Movement Disorders, 2020, 35, 1649-1657.	3.9	50
39	Calcium mishandling in absence of primary mitochondrial dysfunction drives cellular pathology in Wolfram Syndrome. Scientific Reports, 2020, 10, 4785.	3.3	33
40	Clinical Reasoning: Young woman with orbital pain and diplopia. Neurology, 2020, 94, e752-e757.	1.1	1
41	DNMT1 mutations leading to neurodegeneration paradoxically reflect on mitochondrial metabolism. Human Molecular Genetics, 2020, 29, 1864-1881.	2.9	19
42	Immunotherapy in Narcolepsy. Current Treatment Options in Neurology, 2020, 22, 2.	1.8	9
43	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.	9.1	41
44	Biomarkers for REM sleep behavior disorder in idiopathic and narcoleptic patients. Annals of Clinical and Translational Neurology, 2019, 6, 1872-1876.	3.7	34
45	Report of a novel ATP7A mutation causing distal motor neuropathy. Neuromuscular Disorders, 2019, 29, 776-785.	0.6	15
46	Combined brain positron emission tomography/magnetic resonance imaging in GABA A receptor encephalitis. European Journal of Neurology, 2019, 26, e88-e89.	3.3	7
47	Subcutaneous immunoglobulin treatment and leucopenia in acquired demyelinating peripheral neuropathies. European Journal of Neurology, 2019, 26, e80-e81.	3.3	1
48	Broadening the Spectrum of Adulthood X-Linked Adrenoleukodystrophy: A Report of Two Atypical Cases. Frontiers in Neurology, 2019, 10, 70.	2.4	5
49	Abnormal Î±â€synuclein deposits in skin nerves: intraâ€and interâ€laboratory reproducibility. European Journal of Neurology, 2019, 26, 1245-1251.	3.3	38
50	Reader response: Diffuse Lewy body disease manifesting as corticobasal syndrome: A rare form of Lewy body disease. Neurology, 2019, 93, 411-412.	1.1	1
51	The autonomic innervation of hairy skin in humans: an in vivo confocal study. Scientific Reports, 2019, 9, 16982.	3.3	28
52	Diagnostic criteria for amyotrophic lateral sclerosis: A multicentre study of inter-rater variation and sensitivity. Clinical Neurophysiology, 2019, 130, 307-314.	1.5	46
53	Loss of Swallow Tail Sign on Susceptibility-Weighted Imaging in Dementia with Lewy Bodies. Journal of Alzheimer's Disease, 2019, 67, 61-65.	2.6	15
54	Altered globotriaosylceramide accumulation and mucosal neuronal fiber density in the colon of the Fabry disease mouse model. Neurogastroenterology and Motility, 2019, 31, e13529.	3.0	4

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55	Persistence of limb dystonia and myoclonus during sleep in corticobasal syndrome: a case series. Sleep Medicine, 2019, 59, 107-109.	1.6	2
56	SSBP1 mutations cause mtDNA depletion underlying a complex optic atrophy disorder. Journal of Clinical Investigation, 2019, 130, 108-125.	8.2	65
57	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.	2.6	8
58	The role of skin biopsy in differentiating smallâ€fiber neuropathy from ganglionopathy. European Journal of Neurology, 2018, 25, 848-853.	3.3	42
59	Type 1 narcolepsy in anti-Hu antibodies mediated encephalitis: a case report. Sleep Medicine, 2018, 52, 23-25.	1.6	9
60	Riluzole and other prognostic factors in ALS: a population-based registry study in Italy. Journal of Neurology, 2018, 265, 817-827.	3.6	29
61	Subcutaneous immunoglobulin treatment and thromboembolic risk. Annals of Allergy, Asthma and Immunology, 2018, 120, 433-435.	1.0	3
62	Mitochondrial dysfunction in myotonic dystrophy type 1. Neuromuscular Disorders, 2018, 28, 144-149.	0.6	29
63	DGUOK recessive mutations in patients with CPEO, mitochondrial myopathy, parkinsonism and mtDNA deletions. Brain, 2018, 141, e3-e3.	7.6	15
64	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.	0.6	7
65	Non-specific gastrointestinal features: Could it be Fabry disease?. Digestive and Liver Disease, 2018, 50, 429-437.	0.9	28
66	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.	1.9	106
67	Skin Î±-synuclein deposits differ in clinical variants of synucleinopathy: an in vivo study. Scientific Reports, 2018, 8, 14246.	3.3	75
68	Cerebrospinal Fluid Biomarkers in Patients with Frontotemporal Dementia Spectrum: A Single-Center Study. Journal of Alzheimer's Disease, 2018, 66, 551-563.	2.6	46
69	High frequency somatosensory stimulation in dystonia: Evidence fordefective inhibitory plasticity. Movement Disorders, 2018, 33, 1902-1909.	3.9	43
70	Skin Nerve Phosphorylated Î±-Synuclein Deposits in Parkinson Disease With Orthostatic Hypotension. Journal of Neuropathology and Experimental Neurology, 2018, 77, 942-949.	1.7	40
71	Clonal expansion of mtDNA deletions: different disease models assessed by digital droplet PCR in single muscle cells. Scientific Reports, 2018, 8, 11682.	3.3	21
72	Peculiar combinations of individually non-pathogenic missense mitochondrial DNA variants cause low penetrance Leberâ€™s hereditary optic neuropathy. PLoS Genetics, 2018, 14, e1007210.	3.5	47

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73	Nutraceutical Approach to Peripheral Neuropathies: Evidence from Clinical Trials. Current Drug Metabolism, 2018, 19, 460-468.	1.2	9
74	The Italian Dystonia Registry: rationale, design and preliminary findings. Neurological Sciences, 2017, 38, 819-825.	1.9	35
75	Added value of electromyography in the diagnosis of myopathy: A consensus exercise. Clinical Neurophysiology, 2017, 128, 697-701.	1.5	12
76	Spine Topographical Distribution of Skin Î±-Synuclein Deposits in Idiopathic Parkinson Disease. Journal of Neuropathology and Experimental Neurology, 2017, 76, 384-389.	1.7	36
77	Skin nerve phosphorylated Î±-synuclein deposits in idiopathic REM sleep behavior disorder. Neurology, 2017, 88, 2128-2131.	1.1	113
78	The spectrum of REM sleep-related episodes in children with type 1 narcolepsy. Brain, 2017, 140, 1669-1679.	7.6	56
79	High frequency somatosensory stimulation increases sensori-motor inhibition and leads to perceptual improvement in healthy subjects. Clinical Neurophysiology, 2017, 128, 1015-1025.	1.5	45
80	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.	3.6	27
81	Modulation of the Muscle Activity During Sleep in Cervical Dystonia. Sleep, 2017, 40, .	1.1	22
82	Antibodies Against Hypocretin Receptor 2 Are Rare in Narcolepsy. Sleep, 2017, 40, .	1.1	32
83	Post-ganglionic autonomic neuropathy associated with anti-glutamic acid decarboxylase antibodies. Clinical Autonomic Research, 2017, 27, 51-55.	2.5	7
84	Paraneoplastic cerebellar degeneration and lambertâ€eaton myasthenia in a patient with merkel cell carcinoma and voltageâ€gated calcium channel antibodies. Muscle and Nerve, 2017, 56, 998-1000.	2.2	11
85	O96 Large inter- and intra-rater variation on diagnostic criteria for amyotrophic lateral sclerosis. Clinical Neurophysiology, 2017, 128, e209-e210.	1.5	0
86	Absent cardiac and muscle sympathetic nerve activities involvement in Ross syndrome: A follow-up study. Autonomic Neuroscience: Basic and Clinical, 2017, 208, 161-164.	2.8	6
87	A new potential biomarker for dementia with Lewy bodies. Neurology, 2017, 89, 318-326.	1.1	92
88	Skin globotriaosylceramide 3 deposits are specific to Fabry disease with classical mutations and associated with small fibre neuropathy. PLoS ONE, 2017, 12, e0180581.	2.5	21
89	LOPED study: looking for an early diagnosis in a late-onset Pompe disease high-risk population. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, jnnp-2014-310164.	1.9	50
90	Cervical demyelinating lesion presenting with choreoathetoid movements and dystonia. Journal of the Neurological Sciences, 2016, 368, 203-205.	0.6	4

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91	Skin nerve misfolded Î±â€synuclein in pure autonomic failure and <scp>P</scp>arkinson disease. Annals of Neurology, 2016, 79, 306-316.	5.3	118
92	A longitudinal study of a family with adult-onset autosomal dominant leukodystrophy: Clinical, autonomic and neuropsychological findings. Autonomic Neuroscience: Basic and Clinical, 2016, 195, 20-26.	2.8	10
93	Parkinsonian tremor persisting during cataplexy. Sleep Medicine, 2016, 17, 174-176.	1.6	6
94	Relationship of white and gray matter abnormalities to clinical and genetic features in myotonic dystrophy type 1. NeuroImage: Clinical, 2016, 11, 678-685.	2.7	55
95	Pain in Fabry Disease: Practical Recommendations for Diagnosis and Treatment. CNS Neuroscience and Therapeutics, 2016, 22, 568-576.	3.9	75
96	Characterization of Human Dermal Fibroblasts in Fabry Disease. Journal of Cellular Physiology, 2016, 231, 192-203.	4.1	6
97	Melanopsin retinal ganglion cell loss in <scp>A</scp>lzheimer disease. Annals of Neurology, 2016, 79, 90-109.	5.3	299
98	Increased expression of Trpv1 in peripheral terminals mediates thermal nociception in Fabry disease mouse model. Molecular Pain, 2016, 12, 174480691666372.	2.1	28
99	A multiâ€center, multinational ageâ€and genderâ€adjusted normative dataset for immunofluorescent intraepidermal nerve fiber density at the distal leg. European Journal of Neurology, 2016, 23, 333-338.	3.3	107
100	Skin biopsy and microneurography disclose selective noradrenergic dysfunction due to dopamine-Î²-hydroxylase deficiency. Autonomic Neuroscience: Basic and Clinical, 2016, 197, 56-59.	2.8	5
101	ALDH18A1gene mutations cause dominant spastic paraplegia SPG9: loss of function effect and plausibility of a dominant negative mechanism. Brain, 2016, 139, e3-e3.	7.6	42
102	Reply: Both mitochondrial DNA and mitonuclear gene mutations cause hearing loss through cochlear dysfunction. Brain, 2016, 139, e34-e34.	7.6	7
103	Cutaneous sensory and autonomic denervation in CADASIL. Neurology, 2016, 86, 1039-1044.	1.1	13
104	From state dissociation to status dissociatus. Sleep Medicine Reviews, 2016, 28, 5-17.	8.5	56
105	Intermittent head drops: the differential spectrum. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, 414-419.	1.9	11
106	Homozygous <scp>NOTCH</scp> 3 null mutation and impaired <scp>NOTCH</scp> 3 signaling in recessive earlyâ€onset arteriopathy and cavitating leukoencephalopathy. EMBO Molecular Medicine, 2015, 7, 848-858.	6.9	48
107	Nocturnal Sleep Dynamics Identify Narcolepsy Type 1. Sleep, 2015, 38, 1277-1284.	1.1	76
108	Microneurographic recording from unmyelinated nerve fibers in neurological disorders: An update. Clinical Neurophysiology, 2015, 126, 437-445.	1.5	29

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109	Mutations in SLC25A46, encoding a UGO1-like protein, cause an optic atrophy spectrum disorder. Nature Genetics, 2015, 47, 926-932.	21.4	166
110	Skin biopsy and Iâ€123 MIBG scintigraphy findings in idiopathic Parkinson's disease and parkinsonism: A comparative study. Movement Disorders, 2015, 30, 986-989.	3.9	22
111	Syndromic parkinsonism and dementia associated with <scp>OPA</scp>1 missense mutations. Annals of Neurology, 2015, 78, 21-38.	5.3	154
112	Nonâ€paraneoplastic ataxia in a patient with contactinâ€associated proteinâ€2 antibodies and benign course. European Journal of Neurology, 2015, 22, e62-3.	3.3	5
113	Heterogeneity in ALSFRS-R decline and survival: a population-based study in Italy. Neurological Sciences, 2015, 36, 2243-2252.	1.9	41
114	OPA1-related auditory neuropathy: site of lesion and outcome of cochlear implantation. Brain, 2015, 138, 563-576.	7.6	86
115	Immunotherapy of oneiric stupor in Morvan syndrome: Efficacy documented by actigraphy. Neurology, 2015, 84, 2457-2459.	1.1	12
116	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, 2588-2595.	1.6	16
117	An inflammatory myopathy unmasks a case of leprosy in an Italian patient. Journal of Neurology, 2015, 262, 2179-2181.	3.6	5
118	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.	2.5	44
119	Primary progressive narcolepsy type 1: The other side of the coin. Neurology, 2014, 83, 2189-2190.	1.1	46
120	Narcolepsy is a common phenotype in HSAN IE and ADCA-DN. Brain, 2014, 137, 1643-1655.	7.6	49
121	Lower wake resting sympathetic and cardiovascular activities in narcolepsy with cataplexy. Neurology, 2014, 83, 1080-1086.	1.1	47
122	Pearls & Oy-sters: Rapidly progressive dementia. Neurology, 2014, 82, e149-52.	1.1	3
123	Iodineâ€123 Metaiodobenzylguanidine Scintigraphy and Iodineâ€123 Ioflupane Single Photon Emission Computed Tomography in Lewy Body Diseases: Complementary or Alternative Techniques?. Journal of Neuroimaging, 2014, 24, 149-154.	2.0	43
124	Small nerve fiber involvement in patients referred for fibromyalgia. Muscle and Nerve, 2014, 49, 757-759.	2.2	90
125	Italian recommendations for Lambertâ€“Eaton myasthenic syndrome (LEMS) management. Neurological Sciences, 2014, 35, 515-520.	1.9	18
126	Quality of life in patients with craniocervical dystonia: Italian validation of the â€œCervical Dystonia Impact Profile (CDIP-58)â€•and the â€œCraniocervical Dystonia Questionnaire (CDQ-24)â€• Neurological Sciences, 2014, 35, 1053-1058.	1.9	3

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127	Sympathetic and cardiovascular changes during sleep in narcolepsy with cataplexy patients. Sleep Medicine, 2014, 15, 315-321.	1.6	39
128	Efficient mitochondrial biogenesis drives incomplete penetrance in Leberâ€™s hereditary optic neuropathy. Brain, 2014, 137, 335-353.	7.6	229
129	Subcutaneous immunoglobulin in CIDP and MMN: a short-term nationwide study. Journal of Neurology, 2014, 261, 2159-2164.	3.6	59
130	Epidemiology of amyotrophic lateral sclerosis in Emilia Romagna Region (Italy): A population based study. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2014, 15, 262-268.	1.7	46
131	Skin nerve Î±-synuclein deposits. Neurology, 2014, 82, 1362-1369.	1.1	247
132	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, 1789-1793.	3.6	25
133	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, 1825-1827.	3.6	12
134	Hypnic jerks: neurophysiological characterization of a new motor pattern. Sleep Medicine, 2014, 15, 725-727.	1.6	19
135	Cataplectic attacks during rapid eye movement sleep behavior disorder episodes in a narcoleptic patient. Sleep Medicine, 2014, 15, 273-275.	1.6	6
136	A Novel in-Frame 18-bp Microdeletion inMT-CYBCauses a Multisystem Disorder with Prominent Exercise Intolerance. Human Mutation, 2014, 35, 954-958.	2.5	38
137	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, 582-585.	1.6	6
138	Loss of temporal retinal nerve fibers in Parkinson disease: a mitochondrial pattern?. European Journal of Neurology, 2013, 20, 198-201.	3.3	92
139	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, 445-452.	3.8	17
140	Oct-1 recruitment to the nuclear envelope in adult-onset autosomal dominant leukodystrophy. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2013, 1832, 411-420.	3.8	25
141	Brachial amyotrophic diplegia associated with the a140a superoxide dismutase 1 mutation. Neurogenetics, 2013, 14, 255-256.	1.4	3
142	A novel pedigree with familial cortical myoclonic tremor and epilepsy (<scp>FCMTE</scp>): Clinical characterization, refinement of the <scp>FCMTE</scp>2 locus, and confirmation of a founder haplotype. Epilepsia, 2013, 54, 1298-1306.	5.1	23
143	Tremor in primary adult-onset dystonia: prevalence and associated clinical features. Journal of Neurology, Neurosurgery and Psychiatry, 2013, 84, 404-408.	1.9	71
144	Sleep disorders in patients with spinal cord injury. Sleep Medicine Reviews, 2013, 17, 399-409.	8.5	62

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145	Skin sympathetic fiber Î±-synuclein deposits. Neurology, 2013, 80, 725-732.	1.1	72
146	Acute rhabdomyolysis induced by tonicâ€“clonic epileptic seizures in a patient with glucose-6-phosphate dehydrogenase deficiency. Journal of Neurology, 2013, 260, 2669-2671.	3.6	6
147	Idebenone treatment in patients with OPA1-mutant dominant optic atrophy. Brain, 2013, 136, e231-e231.	7.6	62
148	A Novel Null Homozygous Mutation Confirms CACNA2D2 as a Gene Mutated in Epileptic Encephalopathy. PLoS ONE, 2013, 8, e82154.	2.5	67
149	Revisiting the issue of mitochondrial DNA content in optic mitochondriopathies. Neurology, 2012, 79, 1517-1519.	1.1	13
150	Peripheral Autonomic Neuropathy: Diagnostic Contribution of Skin Biopsy. Journal of Neuropathology and Experimental Neurology, 2012, 71, 1000-1008.	1.7	40
151	Phenotypic overlap in familial and sporadic primary adult-onset extracranial dystonia. Journal of Neurology, 2012, 259, 2414-2418.	3.6	7
152	Muscle and skin sympathetic activities in Ross syndrome. Clinical Neurophysiology, 2012, 123, 1639-1643.	1.5	15
153	Age at onset and symptom spread in primary adultâ€onset blepharospasm and cervical dystonia. Movement Disorders, 2012, 27, 1447-1450.	3.9	46
154	Muscle sympathetic response to arousal predicts neurovascular reactivity during mental stress. Journal of Physiology, 2012, 590, 2885-2896.	2.9	23
155	Pseudomyotonia in Romagnola cattle caused by novel ATP2A1mutations. BMC Veterinary Research, 2012, 8, 186.	1.9	13
156	The empowerment of translational research: lessons from laminopathies. Orphanet Journal of Rare Diseases, 2012, 7, 37.	2.7	7
157	Rare Primary Mitochondrial DNA Mutations and Probable Synergistic Variants in Leberâ€™s Hereditary Optic Neuropathy. PLoS ONE, 2012, 7, e42242.	2.5	73
158	Development of a disability scale for myotonic dystrophy type 1. Acta Neurologica Scandinavica, 2012, 125, 431-438.	2.1	14
159	Eye symptoms in relatives of patients with primary adultâ€onset dystonia. Movement Disorders, 2012, 27, 305-307.	3.9	26
160	Microneurographic evaluation of sympathetic activity in small fiber neuropathy. Clinical Neurophysiology, 2011, 122, 1854-1859.	1.5	15
161	Impact of medical audit on electrodiagnostic medicine in polyneuropathy. Clinical Neurophysiology, 2011, 122, 2523-2529.	1.5	3
162	Isolated noradrenergic failure in adult-onset autosomal dominant leukodystrophy. Autonomic Neuroscience: Basic and Clinical, 2011, 159, 123-126.	2.8	21

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163	Autonomic disturbances in narcolepsy. Sleep Medicine Reviews, 2011, 15, 187-196.	8.5	73
164	Somatic and autonomic small fiber neuropathy induced by bortezomib therapy: an immunofluorescence study. Neurological Sciences, 2011, 32, 361-363.	1.9	50
165	Methods of sudomotor innervation quantification. Muscle and Nerve, 2011, 43, 920-921.	2.2	1
166	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.	4.5	36
167	Idebenone Treatment In Leber's Hereditary Optic Neuropathy. Brain, 2011, 134, e188-e188.	7.6	192
168	A clinically complex form of dominant optic atrophy (OPA8) maps on chromosome 16. Human Molecular Genetics, 2011, 20, 1893-1905.	2.9	36
169	Environmental risk factors and clinical phenotype in familial and sporadic primary blepharospasm. Neurology, 2011, 77, 631-637.	1.1	42
170	Small fiber neuropathy in female patients with fabry disease. Muscle and Nerve, 2010, 41, 409-412.	2.2	50
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