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List of Publications by Year in descending order

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Version: 2024-02-01

77
papers

2,502
citations

218381

26
h-index

233125

45
g-index

83
all docs

83
docs citations

83
times ranked

4289
citing authors

#	ARTICLE	IF	CITATIONS
1	Mutations in SYNE1 lead to a newly discovered form of autosomal recessive cerebellar ataxia. <i>Nature Genetics</i> , 2007, 39, 80-85.	9.4	287
2	Deleterious mutations in the essential mRNA metabolism factor, hGle1, in amyotrophic lateral sclerosis. <i>Human Molecular Genetics</i> , 2015, 24, 1363-1373.	1.4	122
3	From animal models to human disease: a genetic approach for personalized medicine in ALS. <i>Acta Neuropathologica Communications</i> , 2016, 4, 70.	2.4	115
4	Mutations in CAPN1 Cause Autosomal-Recessive Hereditary Spastic Paraplegia. <i>American Journal of Human Genetics</i> , 2016, 98, 1038-1046.	2.6	96
5	Misfolded SOD1 pathology in sporadic Amyotrophic Lateral Sclerosis. <i>Scientific Reports</i> , 2018, 8, 14223.	1.6	85
6	Clinical and genetic study of autosomal recessive cerebellar ataxia type 1. <i>Annals of Neurology</i> , 2007, 62, 93-98.	2.8	82
7	Clinical and genetic study of hereditary spastic paraplegia in Canada. <i>Neurology: Genetics</i> , 2017, 3, e122.	0.9	82
8	<i>SMPD1</i> mutations, activity, and α -synuclein accumulation in Parkinson's disease. <i>Movement Disorders</i> , 2019, 34, 526-535.	2.2	81
9	The Classification of Autosomal Recessive Cerebellar Ataxias: a Consensus Statement from the Society for Research on the Cerebellum and Ataxias Task Force. <i>Cerebellum</i> , 2019, 18, 1098-1125.	1.4	80
10	Hereditary motor and sensory neuropathy with agenesis of the corpus callosum. <i>Annals of Neurology</i> , 2003, 54, 9-18.	2.8	67
11	Genetic, Structural, and Functional Evidence Link <i>TMEM175</i> to Synucleinopathies. <i>Annals of Neurology</i> , 2020, 87, 139-153.	2.8	65
12	Pompe Disease: Diagnosis and Management. Evidence-Based Guidelines from a Canadian Expert Panel. <i>Canadian Journal of Neurological Sciences</i> , 2016, 43, 472-485.	0.3	54
13	Replication study of MATR3 in familial and sporadic amyotrophic lateral sclerosis. <i>Neurobiology of Aging</i> , 2016, 37, 209.e17-209.e21.	1.5	53
14	RNF213 Is Associated with Intracranial Aneurysms in the French-Canadian Population. <i>American Journal of Human Genetics</i> , 2016, 99, 1072-1085.	2.6	49
15	Systematic review of autosomal recessive ataxias and proposal for a classification. <i>Cerebellum and Ataxias</i> , 2017, 4, 3.	1.9	49
16	<i>SYNE1</i> Mutations in Autosomal Recessive Cerebellar Ataxia. <i>JAMA Neurology</i> , 2013, 70, 1296-31.	4.5	47
17	Early detection of structural abnormalities and cytoplasmic accumulation of TDP-43 in tissue-engineered skins derived from ALS patients. <i>Acta Neuropathologica Communications</i> , 2015, 3, 5.	2.4	47
18	Clinical Validity of the Mattis Dementia Rating Scale-2 in Parkinson Disease With MCI and Dementia. <i>Journal of Geriatric Psychiatry and Neurology</i> , 2012, 25, 100-106.	1.2	45

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19	Clinical, electrophysiologic, and genetic study of non-dystrophic myotonia in French-Canadians. <i>Neuromuscular Disorders</i> , 2009, 19, 330-334.	0.3	44
20	Canadian best practice recommendations for the management of amyotrophic lateral sclerosis. <i>Cmaj</i> , 2020, 192, E1453-E1468.	0.9	44
21	Evolution of a Human-Specific Tandem Repeat Associated with ALS. <i>American Journal of Human Genetics</i> , 2020, 107, 445-460.	2.6	39
22	Fineâ€œMapping of <i>SNCA</i> in Rapid Eye Movement Sleep Behavior Disorder and Overt Synucleinopathies. <i>Annals of Neurology</i> , 2020, 87, 584-598.	2.8	39
23	Analysis of Heterozygous <sc><i>PRKN</i></sc> Variants and Copyâ€œNumber Variations in Parkinson's Disease. <i>Movement Disorders</i> , 2021, 36, 178-187.	2.2	39
24	Analysis of DNAJC13 mutations in French-Canadian/French cohort of Parkinson's disease. <i>Neurobiology of Aging</i> , 2016, 45, 212.e13-212.e17.	1.5	38
25	The Quebec Parkinson Network: A Researcher-Patient Matching Platform and Multimodal Biorepository. <i>Journal of Parkinson's Disease</i> , 2020, 10, 301-313.	1.5	35
26	Targeted sequencing of Parkinsonâ€œs disease loci genes highlights <i>SYT11</i>, <i>FGF20</i> and other associations. <i>Brain</i> , 2021, 144, 462-472.	3.7	31
27	Transmission of ALS pathogenesis by the cerebrospinal fluid. <i>Acta Neuropathologica Communications</i> , 2020, 8, 65.	2.4	30
28	Linkage to the CCM2 Locus and Genetic Heterogeneity in Familial Cerebral Cavernous Malformation. <i>Canadian Journal of Neurological Sciences</i> , 2003, 30, 122-128.	0.3	29
29	Teneurin transmembrane protein 4 is not a cause for essential tremor in a Canadian population. <i>Movement Disorders</i> , 2017, 32, 292-295.	2.2	29
30	Management of Patients with Cerebellar Ataxia During the COVID-19 Pandemic: Current Concerns and Future Implications. <i>Cerebellum</i> , 2020, 19, 562-568.	1.4	26
31	Moyamoya Disease Susceptibility Gene <i>RNF213</i> Regulates Endothelial Barrier Function. <i>Stroke</i> , 2022, 53, 1263-1275.	1.0	26
32	Cognitive Impairment in ARCA-1, a Newly Discovered Pure Cerebellar Ataxia Syndrome. <i>Cerebellum</i> , 2010, 9, 443-453.	1.4	24
33	Effects of Antiparkinson Medication on Cognition in Parkinsonâ€œs Disease: A Systematic Review. <i>Canadian Journal of Neurological Sciences</i> , 2018, 45, 375-404.	0.3	24
34	Chemosensory Dysfunctions Induced by COVID-19 Can Persist up to 7 Months: A Study of Over 700 Healthcare Workers. <i>Chemical Senses</i> , 2021, 46, .	1.1	24
35	The Occurrence of FUS Mutations in Pediatric Amyotrophic Lateral Sclerosis: A Case Report and Review of the Literature. <i>Journal of Child Neurology</i> , 2020, 35, 556-562.	0.7	23
36	Characterization of the phenotype with cognitive impairment and protein mislocalization in SCA34. <i>Neurology: Genetics</i> , 2020, 6, e403.	0.9	21

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37	Common and rare GCH1 variants are associated with Parkinson's disease. <i>Neurobiology of Aging</i> , 2019, 73, 231.e1-231.e6.	1.5	20
38	Clinical and genetic analysis of <i>ATP13A2</i> in hereditary spastic paraplegia expands the phenotype. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1052.	0.6	20
39	Analysis of common and rare <i>VPS13C</i> variants in late-onset Parkinson disease. <i>Neurology: Genetics</i> , 2020, 6, 385.	0.9	19
40	Association study of DNAJC13, UCHL1, HTRA2, GIGYF2, and EIF4G1 with Parkinson's disease. <i>Neurobiology of Aging</i> , 2021, 100, 119.e7-119.e13.	1.5	19
41	Spastic ataxias. <i>Handbook of Clinical Neurology</i> / Edited By P J Vinken and G W Bruyn, 2018, 155, 191-203.	1.0	18
42	Characterization of a novel SPC3A deletion in a French-Canadian family. <i>Annals of Neurology</i> , 2007, 61, 599-603.	2.8	17
43	POLR3A variants in hereditary spastic paraplegia and ataxia. <i>Brain</i> , 2018, 141, e1-e1.	3.7	17
44	The influence of vascular risk factors on cognitive function in early Parkinson's disease. <i>International Journal of Geriatric Psychiatry</i> , 2018, 33, 288-297.	1.3	17
45	An Optimized Approach to Recover Secreted Proteins from Fibroblast Conditioned-Media for Secretomic Analysis. <i>Frontiers in Cellular Neuroscience</i> , 2016, 10, 70.	1.8	16
46	Physician-assisted death. <i>Neurology</i> , 2016, 87, 1152-1160.	1.5	16
47	Sequencing of the GBA coactivator, Saposin C, in Parkinson disease. <i>Neurobiology of Aging</i> , 2018, 72, 187.e1-187.e3.	1.5	16
48	Smoking history is associated to cognitive impairment in Parkinson's disease. <i>Aging and Mental Health</i> , 2017, 21, 322-326.	1.5	13
49	Medical and Paramedical Care of Patients With Cerebellar Ataxia During the COVID-19 Outbreak: Seven Practical Recommendations of the COVID 19 Cerebellum Task Force. <i>Frontiers in Neurology</i> , 2020, 11, 516.	1.1	13
50	Variants in the Niemann-Pick type C gene NPC1 are not associated with Parkinson's disease. <i>Neurobiology of Aging</i> , 2020, 93, 143.e1-143.e4.	1.5	13
51	Genome-wide association analysis identifies new candidate risk loci for familial intracranial aneurysm in the French-Canadian population. <i>Scientific Reports</i> , 2018, 8, 4356.	1.6	12
52	Recessive cerebellar and afferent ataxias – clinical challenges and future directions. <i>Nature Reviews Neurology</i> , 2022, 18, 257-272.	4.9	12
53	TOX3 Variants Are Involved in Restless Legs Syndrome and Parkinson's Disease with Opposite Effects. <i>Journal of Molecular Neuroscience</i> , 2018, 64, 341-345.	1.1	11
54	Evidence for Non-Mendelian Inheritance in Spastic Paraplegia 7. <i>Movement Disorders</i> , 2021, 36, 1664-1675.	2.2	11

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55	Paresthesia and sensory disturbances associated with 2009 pandemic vaccine receipt: Clinical features and risk factors. <i>Vaccine</i> , 2015, 33, 4464-4471.	1.7	9
56	Association study of essential tremor genetic loci in Parkinson's disease. <i>Neurobiology of Aging</i> , 2018, 66, 178.e13-178.e15.	1.5	9
57	Current and Promising Therapies in Autosomal Recessive Ataxias. <i>CNS and Neurological Disorders - Drug Targets</i> , 2018, 17, 161-171.	0.8	9
58	Genetic testing for amyotrophic lateral sclerosis in Canada – an assessment of current practices. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2021, , 1-8.	1.1	9
59	LRRK2 is not a Significant Cause of Parkinson's Disease in French-Canadians. <i>Canadian Journal of Neurological Sciences</i> , 2007, 34, 333-335.	0.3	8
60	Functional alterations in large-scale resting-state networks of amyotrophic lateral sclerosis: A multi-site study across Canada and the United States. <i>PLoS ONE</i> , 2022, 17, e0269154.	1.1	8
61	Biofabrication of a three dimensional human-based personalized neurofibroma model. <i>Biotechnology Journal</i> , 2021, 16, e2000250.	1.8	7
62	Distinct patterns of progressive gray and white matter degeneration in amyotrophic lateral sclerosis. <i>Human Brain Mapping</i> , 2022, 43, 1519-1534.	1.9	7
63	Genetic, structural and clinical analysis of spastic paraplegia 4. <i>Parkinsonism and Related Disorders</i> , 2022, 98, 62-69.	1.1	7
64	Case Report: Acute Necrotizing Encephalopathy Following COVID-19 Vaccine. <i>Frontiers in Neurology</i> , 2022, 13, 872734.	1.1	7
65	A National Spinal Muscular Atrophy Registry for Real-World Evidence. <i>Canadian Journal of Neurological Sciences</i> , 2020, 47, 810-815.	0.3	6
66	Goal management training and psychoeducation / mindfulness for treatment of executive dysfunction in Parkinson's disease: A feasibility pilot trial. <i>PLoS ONE</i> , 2022, 17, e0263108.	1.1	6
67	Persisting chemosensory impairments in 366 healthcare workers following COVID-19: an 11-month follow-up. <i>Chemical Senses</i> , 2022, 47, .	1.1	6
68	RIC3 variants are not associated with Parkinson's disease in French-Canadians and French. <i>Neurobiology of Aging</i> , 2017, 53, 194.e9-194.e11.	1.5	5
69	No rare deleterious variants from <i>STK32B</i> , <i>PPARGC1A</i> , and <i>CTNNA3</i> are associated with essential tremor. <i>Neurology: Genetics</i> , 2017, 3, e195.	0.9	5
70	Environmental risk factors for amyotrophic lateral sclerosis: a case-control study in Canada and France. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2022, 23, 592-600.	1.1	5
71	Altered Theory of Mind in Parkinson's Disease and Impact on Caregivers: A Pilot Study. <i>Canadian Journal of Neurological Sciences</i> , 2022, 49, 437-440.	0.3	4
72	Goal management training – home-based approach for mild cognitive impairment in Parkinson's disease: a multiple baseline case report. <i>Neurocase</i> , 2018, 24, 276-286.	0.2	3

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73	Genetic and Epidemiological Study of Adult Ataxia and Spastic Paraplegia in Eastern Quebec. Canadian Journal of Neurological Sciences, 2021, 48, 655-665.	0.3	3
74	CAPTURE ALS: the comprehensive analysis platform to understand, remedy and eliminate ALS. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2022, , 1-7.	1.1	3
75	Absence of Mutation Enrichment for Genes Phylogenetically Conserved in the Olivocerebellar Motor Circuitry in a Cohort of Canadian Essential Tremor Cases. Molecular Neurobiology, 2019, 56, 4317-4321.	1.9	2
76	The Puzzle of Huntington Disease Phenocopies. JAMA Neurology, 2016, 73, 1056.	4.5	1
77	<scp> <i>KCNA2</i></scp> mutations are rare in hereditary spastic paraplegia. Annals of Neurology, 2017, 81, 325-326.	2.8	0