Guy A Rouleau

List of Publications by Year
in descending order

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320 papers	36,600 citations	7568 77 h-index	3830 178 g-index
408 all docs	408 docs citations	408 times ranked	35056 citing authors

#	Article	IF	Citations
1	Mutations in Cu/Zn superoxide dismutase gene are associated with familial amyotrophic lateral sclerosis. Nature, 1993, 362, 59-62.	27.8	6,331
2	Identification of novel risk loci, causal insights, and heritable risk for Parkinson's disease: a meta-analysis of genome-wide association studies. Lancet Neurology, The, 2019, 18, 1091-1102.	10.2	1,414
3	TARDBP mutations in individuals with sporadic and familial amyotrophic lateral sclerosis. Nature Genetics, 2008, 40, 572-574.	21.4	1,371
4	Alteration in a new gene encoding a putative membrane-organizing protein causes neuro-fibromatosis type 2. Nature, 1993, 363, 515-521.	27.8	1,351
5	Genome-wide association study identifies 30 loci associated with bipolar disorder. Nature Genetics, 2019, 51, 793-803.	21.4	1,191
6	Moderate expansion of a normally biallelic trinucleotide repeat in spinocerebellar ataxia type 2. Nature Genetics, 1996, 14, 269-276.	21.4	1,092
7	Analysis of shared heritability in common disorders of the brain. Science, 2018, 360, .	12.6	1,085
8	Genomic Relationships, Novel Loci, and Pleiotropic Mechanisms across Eight Psychiatric Disorders. Cell, 2019, 179, 1469-1482.e11.	28.9	935
9	Exome sequencing in amyotrophic lateral sclerosis identifies risk genes and pathways. Science, 2015, 347, 1436-1441.	12.6	823
10	Short GCG expansions in the PABP2 gene cause oculopharyngeal muscular dystrophy. Nature Genetics, 1998, 18, 164-167.	21.4	751
11	A gene encoding a putative GTPase regulator is mutated in familial amyotrophic lateral sclerosis 2. Nature Genetics, 2001, 29, 166-173.	21.4	635
12	Genome-wide association study of restless legs syndrome identifies common variants in three genomic regions. Nature Genetics, 2007, 39, 1000-1006.	21.4	633
13	Genome-wide association study of more than 40,000 bipolar disorder cases provides new insights into the underlying biology. Nature Genetics, 2021, 53, 817-829.	21.4	629
14	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. Neuron, 2018, 97, 1268-1283.e6.	8.1	517
15	Evidence for the complete inactivation of the NF2 gene in the majority of sporadic meningiomas. Nature Genetics, 1994, 6, 180-184.	21.4	514
16	Mutations in a gene encoding a novel protein tyrosine phosphatase cause progressive myoclonus epilepsy. Nature Genetics, 1998, 20, 171-174.	21.4	499
17	Variants of the heavy neurofilament subunit are associated with the development of amyotrophic lateral sclerosis. Human Molecular Genetics, 1994, 3, 1757-1761.	2.9	452
18	Linkage of a Gene Causing Familial Amyotrophic Lateral Sclerosis to Chromosome 21 and Evidence of Genetic-Locus Heterogeneity. New England Journal of Medicine, 1991, 324, 1381-1384.	27.0	407

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19	Increased exonic de novo mutation rate in individuals with schizophrenia. Nature Genetics, 2011, 43, 860-863.	21.4	392
20	Reduced transcriptional regulatory competence of the androgen receptor in Xâ€“linked spinal and bulbar muscular atrophy. Nature Genetics, 1993, 5, 184-188.	21.4	344
21	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	6.2	337
22	Glucocerebrosidase activity in Parkinsonâ€™s disease with and withoutGBAmutations. Brain, 2015, 138, 2648-2658.	7.6	326
23	Exome-wide Rare Variant Analysis Identifies TUBA4A Mutations Associated with Familial ALS. Neuron, 2014, 84, 324-331.	8.1	308
24	Genetic variants associated with response to lithium treatment in bipolar disorder: a genome-wide association study. Lancet, The, 2016, 387, 1085-1093.	13.7	306
25	Genome-wide association study reveals two new risk loci for bipolar disorder. Nature Communications, 2014, 5, 3339.	12.8	294
26	Mutations in GJB6 cause hidrotic ectodermal dysplasia. Nature Genetics, 2000, 26, 142-144.	21.4	270
27	Interrogating the Genetic Determinants of Touretteâ€™s Syndrome and Other Tic Disorders Through Genome-Wide Association Studies. American Journal of Psychiatry, 2019, 176, 217-227.	7.2	242
28	Partitioning the Heritability of Tourette Syndrome and Obsessive Compulsive Disorder Reveals Differences in Genetic Architecture. PLoS Genetics, 2013, 9, e1003864.	3.5	241
29	SOD1 mutation is assosiated with accumulation of neurofilaments in amyotrophic lateral scelaries. Annals of Neurology, 1996, 39, 128-131.	5.3	227
30	Common and rare variant association analyses in amyotrophic lateral sclerosis identify 15 risk loci with distinct genetic architectures and neuron-specific biology. Nature Genetics, 2021, 53, 1636-1648.	21.4	223
31	Direct Measure of the De Novo Mutation Rate in Autism and Schizophrenia Cohorts. American Journal of Human Genetics, 2010, 87, 316-324.	6.2	222
32	NEK1 variants confer susceptibility to amyotrophic lateral sclerosis. Nature Genetics, 2016, 48, 1037-1042.	21.4	218
33	Genetic perspective on the role of the autophagy-lysosome pathway in Parkinson disease. Autophagy, 2015, 11, 1443-1457.	9.1	217
34	Genetics of familial and sporadic amyotrophic lateral sclerosis. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2006, 1762, 956-972.	3.8	214
35	Identification of novel risk loci for restless legs syndrome in genome-wide association studies in individuals of European ancestry: a meta-analysis. Lancet Neurology, The, 2017, 16, 898-907.	10.2	191
36	Genome-wide association study of 40,000 individuals identifies two novel loci associated with bipolar disorder. Human Molecular Genetics, 2016, 25, 3383-3394.	2.9	182

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37	The Impact of Phenotypic and Genetic Heterogeneity on Results of Genome Wide Association Studies of Complex Diseases. PLoS ONE, 2013, 8, e76295.	2.5	177
38	Exome Sequencing Identifies FUS Mutations as a Cause of Essential Tremor. American Journal of Human Genetics, 2012, 91, 313-319.	6.2	176
39	CCNF mutations in amyotrophic lateral sclerosis and frontotemporal dementia. Nature Communications, 2016, 7, 11253.	12.8	174
40	A Frameshift Deletion in Peripherin Gene Associated with Amyotrophic Lateral Sclerosis. Journal of Biological Chemistry, 2004, 279, 45951-45956.	3.4	163
41	Genome-Wide Association Study Identifies Novel Restless Legs Syndrome Susceptibility Loci on 2p14 and 16q12.1. PLoS Genetics, 2011, 7, e1002171.	3.5	163
42	Genetically encoded impairment of neuronal <scp>KCC</scp> 2 cotransporter function in human idiopathic generalized epilepsy. EMBO Reports, 2014, 15, 766-774.	4.5	163
43	Genome-wide association study of intracranial aneurysms identifies 17 risk loci and genetic overlap with clinical risk factors. Nature Genetics, 2020, 52, 1303-1313.	21.4	163
44	Deletion of C9ORF72 Results in Motor Neuron Degeneration and Stress Sensitivity in C. elegans. PLoS ONE, 2013, 8, e83450.	2.5	158
45	Assessment of Response to Lithium Maintenance Treatment in Bipolar Disorder: A Consortium on Lithium Genetics (ConLiGen) Report. PLoS ONE, 2013, 8, e65636.	2.5	156
46	De Novo Coding Variants Are Strongly Associated with Tourette Disorder. Neuron, 2017, 94, 486-499.e9.	8.1	155
47	Clinical Spectrum of Amyotrophic Lateral Sclerosis (ALS). Cold Spring Harbor Perspectives in Medicine, 2017, 7, a024117.	6.2	149
48	High resolution deletion analysis of constitutional DNA from neurofibromatosis type 2 (NF2) patients using microarray-CGH. Human Molecular Genetics, 2001, 10, 271-282.	2.9	147
49	Dissection of genetic factors associated with amyotrophic lateral sclerosis. Experimental Neurology, 2014, 262, 91-101.	4.1	145
50	C9orf72 repeat expansions are a rare genetic cause of parkinsonism. Brain, 2013, 136, 385-391.	7.6	143
51	Rare Copy Number Variants in NRXN1 and CNTN6 Increase Risk for Tourette Syndrome. Neuron, 2017, 94, 1101-1111.e7.	8.1	137
52	The Neuropathology of CAG Repeat Diseases: Review and Update of Genetic and Molecular Features. Brain Pathology, 1997, 7, 901-926.	4.1	134
53	The International Consortium on Lithium Genetics (ConLiGen): An Initiative by the NIMH and IGSLI to Study the Genetic Basis of Response to Lithium Treatment. Neuropsychobiology, 2010, 62, 72-78.	1.9	134
54	Screening for germ-line mutations in theNF2 Gene. Genes Chromosomes and Cancer, 1995, 12, 117-127.	2.8	128

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55	ERBB4 Mutations that Disrupt the Neuregulin-ErbB4 Pathway Cause Amyotrophic Lateral Sclerosis Type 19. American Journal of Human Genetics, 2013, 93, 900-905.	6.2	123
56	Deleterious mutations in the essential mRNA metabolism factor, hGle1, in amyotrophic lateral sclerosis. Human Molecular Genetics, 2015, 24, 1363-1373.	2.9	122
57	Glucocorticoid excess induces preferential depletion of myosin in denervated skeletal muscle fibers. Muscle and Nerve, 1987, 10, 428-438.	2.2	118
58	A blinded international study on the reliability of genetic testing for GGGGCC-repeat expansions inC9orf72reveals marked differences in results among 14 laboratories. Journal of Medical Genetics, 2014, 51, 419-424.	3.2	118
59	GBA mutations are associated with Rapid Eye Movement Sleep Behavior Disorder. Annals of Clinical and Translational Neurology, 2015, 2, 941-945.	3.7	117
60	Dissecting the Shared Genetic Architecture of Suicide Attempt, Psychiatric Disorders, and Known Risk Factors. Biological Psychiatry, 2022, 91, 313-327.	1.3	114
61	A Pentanucleotide ATTTC Repeat Insertion in the Non-coding Region of DAB1, Mapping to SCA37, Causes Spinocerebellar Ataxia. American Journal of Human Genetics, 2017, 101, 87-103.	6.2	112
62	NLGN3/NLGN4 gene mutations are not responsible for autism in the Quebec population. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2005, 132B, 74-75.	1.7	111
63	Three Families With Amyotrophic Lateral Sclerosis and Frontotemporal Dementia With Evidence of Linkage to Chromosome 9p. Archives of Neurology, 2007, 64, 240.	4.5	111
64	Expanding the Clinical Phenotype Associated WithELOVL4Mutation. JAMA Neurology, 2014, 71, 470.	9.0	110
65	Genome-Wide Association Study of Intracranial Aneurysms Confirms Role of Anril and SOX17 in Disease Risk. Stroke, 2012, 43, 2846-2852.	2.0	106
66	Defining the genetic connection linking amyotrophic lateral sclerosis (ALS) with frontotemporal dementia (FTD). Trends in Genetics, 2015, 31, 263-273.	6.7	106
67	Oligomerization of polyalanine expanded PABPN1 facilitates nuclear protein aggregation that is associated with cell death. Human Molecular Genetics, 2001, 10, 2341-2351.	2.9	105
68	Recessive mutations in VPS13D cause childhood onset movement disorders. Annals of Neurology, 2018, 83, 1089-1095.	5.3	104
69	Polymorphism, shared functions and convergent evolution of genes with sequences coding for polyalanine domains. Human Molecular Genetics, 2003, 12, 2967-2979.	2.9	103
70	Association of Polygenic Score for Schizophrenia and HLA Antigen and Inflammation Genes With Response to Lithium in Bipolar Affective Disorder. JAMA Psychiatry, 2018, 75, 65-74.	11.0	102
71	An ALS2 gene mutation causes hereditary spastic paraplegia in a Pakistani kindred. Annals of Neurology, 2003, 53, 144-145.	5.3	97
72	Mutations in CAPN1 Cause Autosomal-Recessive Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2016, 98, 1038-1046.	6.2	96

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73	Homozygotes for oculopharyngeal muscular dystrophy have a severe form of the disease. Annals of Neurology, 1999, 46, 115-118.	5.3	92
74	De Novo Sequence and Copy Number Variants Are Strongly Associated with Tourette Disorder and Implicate Cell Polarity in Pathogenesis. Cell Reports, 2018, 24, 3441-3454.e12.	6.4	91
75	Alteration of ornithine metabolism leads to dominant and recessive hereditary spastic paraplegia. Brain, 2015, 138, 2191-2205.	7.6	88
76	Exome sequencing reveals SPG11 mutations causing juvenile ALS. Neurobiology of Aging, 2012, 33, 839.e5-839.e9.	3.1	87
77	Regulatory domain or CpG site variation in SLC12A5, encoding the chloride transporter KCC2, in human autism and schizophrenia. Frontiers in Cellular Neuroscience, 2015, 9, 386.	3.7	86
78	MEIS1 intronic risk haplotype associated with restless legs syndrome affects its mRNA and protein expression levels. Human Molecular Genetics, 2009, 18, 1065-1074.	2.9	85
79	Loss of Association of REEP2 with Membranes Leads to Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2014, 94, 268-277.	6.2	83
80	Clinical and genetic study of hereditary spastic paraplegia in Canada. Neurology: Genetics, 2017, 3, e122.	1.9	82
81	SMPD1 mutations, activity, and Î±â€synuclein accumulation in Parkinson's disease. Movement Disorders, 2019, 34, 526-535.	3.9	81
82	Compound heterozygous D90A and D96N SOD1 mutations in a recessive amyotrophic lateral sclerosis family. Annals of Neurology, 2001, 49, 267-271.	5.3	80
83	Genome-wide association study in essential tremor identifies three new loci. Brain, 2016, 139, 3163-3169.	7.6	78
84	A homozygous loss-of-function variant in MYH11 in a case with megacystis-microcolon-intestinal hypoperistalsis syndrome. European Journal of Human Genetics, 2015, 23, 1266-1268.	2.8	77
85	De novo variants in sporadic cases of childhood onset schizophrenia. European Journal of Human Genetics, 2016, 24, 944-948.	2.8	77
86	GBA p.T369M substitution in Parkinson disease: Polymorphism or association? A meta-analysis. Neurology: Genetics, 2016, 2, e104.	1.9	74
87	The 14q restless legs syndrome locus in the French Canadian population. Annals of Neurology, 2004, 55, 887-891.	5.3	71
88	Identification of rare protein disulfide isomerase gene variants in amyotrophic lateral sclerosis patients. Gene, 2015, 566, 158-165.	2.2	70
89	Mutations in DCC cause isolated agenesis of the corpus callosum with incomplete penetrance. Nature Genetics, 2017, 49, 511-514.	21.4	69
90	Hereditary motor and sensory neuropathy with agenesis of the corpus callosum. Annals of Neurology, 2003, 54, 9-18.	5.3	67

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91	Asian Origin for the Worldwide-Spread Mutational Event in Machado-Joseph Disease. Archives of Neurology, 2007, 64, 1502.	4.5	65
92	Genetic, Structural, and Functional Evidence Link TMEM175 to Synucleinopathies. Annals of Neurology, 2020, 87, 139-153.	5.3	65
93	Dopa-responsive dystonia due to a large deletion in the GTP cyclohydrolase I gene. Annals of Neurology, 2000, 47, 517-520.	5.3	63
94	Family Study of Restless Legs Syndrome in Quebec, Canada. Archives of Neurology, 2010, 67, 617-22.	4.5	63
95	Quantitative Analysis of Climbing Defects in a Drosophila Model of Neurodegenerative Disorders. Journal of Visualized Experiments, 2015, , e52741.	0.3	60
96	Genetics of Intracranial Aneurysms. Stroke, 2018, 49, 780-787.	2.0	60
97	Somatic mosaicism in the central nervous system in spinocerebellar ataxia type 1 and machado-joseph disease. Annals of Neurology, 1996, 40, 199-206.	5.3	59
98	PMPCAmutations cause abnormal mitochondrial protein processing in patients with non-progressive cerebellar ataxia. Brain, 2015, 138, 1505-1517.	7.6	58
99	Joubert Syndrome in French Canadians and Identification of Mutations in CEP104. American Journal of Human Genetics, 2015, 97, 744-753.	6.2	56
100	Transcriptome-wide association study of attention deficit hyperactivity disorder identifies associated genes and phenotypes. Nature Communications, 2019, 10, 4450.	12.8	56
101	ALS: Recent Developments from Genetics Studies. Current Neurology and Neuroscience Reports, 2016, 16, 59.	4.2	55
102	Ribosomal frameshifting on MJD-1 transcripts with long CAG tracts. Human Molecular Genetics, 2005, 14, 2649-2660.	2.9	54
103	Replication study of MATR3 in familial and sporadic amyotrophic lateral sclerosis. Neurobiology of Aging, 2016, 37, 209.e17-209.e21.	3.1	53
104	A novel locus for pure recessive hereditary spastic paraplegia maps to 10q22.1-10q24.1. Annals of Neurology, 2004, 56, 579-582.	5.3	52
105	Genome-Wide Association Study of Intracranial Aneurysm Identifies a New Association on Chromosome 7. Stroke, 2014, 45, 3194-3199.	2.0	52
106	Investigation of the RFC1 Repeat Expansion in a Canadian and a Brazilian Ataxia Cohort: Identification of Novel Conformations. Frontiers in Genetics, 2019, 10, 1219.	2.3	51
107	Global characterization of copy number variants in epilepsy patients from whole genome sequencing. PLoS Genetics, 2018, 14, e1007285.	3.5	50
108	RNF213 Is Associated with Intracranial Aneurysms in the French-Canadian Population. American Journal of Human Genetics, 2016, 99, 1072-1085.	6.2	49

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109	Systematic review of autosomal recessive ataxias and proposal for a classification. Cerebellum and Ataxias, 2017, 4, 3.	1.9	49
110	Transgenic expression of an expanded (GCG)13 repeat PABPN1 leads to weakness and coordination defects in mice. Neurobiology of Disease, 2005, 18, 528-536.	4.4	48
111	Mutation screening ofFOXP2 in individuals diagnosed with autistic disorder. American Journal of Medical Genetics Part A, 2003, 118A, 172-175.	2.4	46
112	Molecular aspects of hereditary spastic paraplegia. Experimental Cell Research, 2014, 325, 18-26.	2.6	46
113	Preclinical target validation using patient-derived cells. Nature Reviews Drug Discovery, 2015, 14, 149-150.	46.4	46
114	SPG7 mutations explain a significant proportion of French Canadian spastic ataxia cases. European Journal of Human Genetics, 2016, 24, 1016-1021.	2.8	46
115	Retention of hexanucleotide repeat-containing intron in C9orf72 mRNA: implications for the pathogenesis of ALS/FTD. Acta Neuropathologica Communications, 2016, 4, 18.	5.2	46
116	Association of Variants in the SPTLC1 Gene With Juvenile Amyotrophic Lateral Sclerosis. JAMA Neurology, 2021, 78, 1236.	9.0	46
117	Schizophrenia and chromosome 6p. American Journal of Medical Genetics Part A, 1997, 74, 195-198.	2.4	45
118	Functional characterization of the D188V mutation in neuronal voltage-gated sodium channel causing generalized epilepsy with febrile seizures plus (GEFS). Epilepsy Research, 2003, 53, 107-117.	1.6	45
119	GBA variants in REM sleep behavior disorder. Neurology, 2020, 95, e1008-e1016.	1.1	45
120	Association of polygenic score for major depression with response to lithium in patients with bipolar disorder. Molecular Psychiatry, 2021, 26, 2457-2470.	7.9	44
121	Enrichment of SARM1 alleles encoding variants with constitutively hyperactive NADase in patients with ALS and other motor nerve disorders. ELife, 2021, 10, .	6.0	44
122	Loss of heterozygosity on the long arm of chromosome 22 in pheochromocytoma. Genes Chromosomes and Cancer, 1992, 5, 399-403.	2.8	43
123	Clouston hidrotic ectodermal dysplasia (HED): genetic homogeneity, presence of a founder effect in the French Canadian population and fine genetic mapping. European Journal of Human Genetics, 2000, 8, 372-380.	2.8	43
124	Inhibition of the kinase WNK1/HSN2 ameliorates neuropathic pain by restoring GABA inhibition. Science Signaling, 2016, 9, ra32.	3.6	43
125	DCCmutation update: Congenital mirror movements, isolated agenesis of the corpus callosum, and developmental split brain syndrome. Human Mutation, 2018, 39, 23-39.	2.5	41
126	PABP2 polyalanine tract expansion causes intranuclear inclusions in oculopharyngeal muscular dystrophy. Annals of Neurology, 2000, 48, 798-802.	5.3	40

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127	Expanded ATXN3 frameshifting events are toxic in Drosophila and mammalian neuron models. Human Molecular Genetics, 2012, 21, 2211-2218.	2.9	40
128	Gain-of-function missense variant in SLC12A2, encoding the bumetanide-sensitive NKCC1 cotransporter, identified in human schizophrenia. Journal of Psychiatric Research, 2016, 77, 22-26.	3.1	40
129	Genetics of restless legs syndrome. Sleep Medicine, 2017, 31, 18-22.	1.6	40
130	Characterization of human iPSC-derived astrocytes with potential for disease modeling and drug discovery. Neuroscience Letters, 2020, 731, 135028.	2.1	40
131	A mutation in theHSN2gene causes sensory neuropathy type II in a Lebanese family. Annals of Neurology, 2004, 56, 572-575.	5.3	39
132	Truncating Mutations in UBAP1 Cause Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2019, 104, 767-773.	6.2	39
133	Mechanisms Underlying the Hyperexcitability of CA3 and Dentate Gyrus Hippocampal Neurons Derived From Patients With Bipolar Disorder. Biological Psychiatry, 2020, 88, 139-149.	1.3	39
134	Evolution of a Human-Specific Tandem Repeat Associated with ALS. American Journal of Human Genetics, 2020, 107, 445-460.	6.2	39
135	Fineâ€Mapping of SNCA in Rapid Eye Movement Sleep Behavior Disorder and Overt Synucleinopathies. Annals of Neurology, 2020, 87, 584-598.	5.3	39
136	Analysis of Heterozygous <scp>PRKN</scp> Variants and Copyâ€Number Variations in Parkinson's Disease. Movement Disorders, 2021, 36, 178-187.	3.9	39
137	Analysis of 14 CAG repeat-containing genes in schizophrenia. American Journal of Medical Genetics Part A, 1999, 88, 694-699.	2.4	38
138	PABPN1 polyalanine tract deletion and long expansions modify its aggregation pattern and expression. Experimental Cell Research, 2008, 314, 1652-1666.	2.6	38
139	Analysis of DNAJC13 mutations in French-Canadian/French cohort of Parkinson's disease. Neurobiology of Aging, 2016, 45, 212.e13-212.e17.	3.1	38
140	Mutations in ATP13A2 (PARK9) are associated with an amyotrophic lateral sclerosis-like phenotype, implicating this locus in further phenotypic expansion. Human Genomics, 2019, 13, 19.	2.9	38
141	Genome-wide study of DNA methylation shows alterations in metabolic, inflammatory, and cholesterol pathways in ALS. Science Translational Medicine, 2022, 14, eabj0264.	12.4	38
142	Genetic and Clinical Predictors of Deep Brain Stimulation in Youngâ€Onset Parkinson's Disease. Movement Disorders Clinical Practice, 2016, 3, 465-471.	1.5	37
143	CPT1A Missense Mutation Associated With Fatty Acid Metabolism and Reduced Height in Greenlanders. Circulation: Cardiovascular Genetics, 2017, 10, .	5.1	37
144	Molecular Genetics of Neurofibromatosis 2 and Related Tumors (Acoustic Neuroma and Meningioma). Annals of the New York Academy of Sciences, 1991, 615, 338-343.	3.8	36

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145	A de novo frameshift mutation in chromodomain helicase DNAâ€binding domain 8 (CHD8): A case report and literature review. American Journal of Medical Genetics, Part A, 2016, 170, 1225-1235.	1.2	36
146	SCARB2 variants and glucocerebrosidase activity in Parkinsonâ€™s disease. Npj Parkinson's Disease, 2016, 2, .	5.3	36
147	Sleep disorders and Parkinson disease; lessons from genetics. Sleep Medicine Reviews, 2018, 41, 101-112.	8.5	35
148	The Quebec Parkinson Network: A Researcher-Patient Matching Platform and Multimodal Biorepository. Journal of Parkinson's Disease, 2020, 10, 301-313.	2.8	35
149	A NewELOVL4Mutation in a Case of Spinocerebellar Ataxia With Erythrokeratodermia. JAMA Neurology, 2015, 72, 942.	9.0	34
150	Modifiers of (CAG)n instability in Machadoâ€“Joseph disease (MJD/SCA3) transmissions: an association study with DNA replication, repair and recombination genes. Human Genetics, 2014, 133, 1311-1318.	3.8	33
151	The emerging role of SMPD1 mutations in Parkinson's disease: Implications for future studies. Parkinsonism and Related Disorders, 2015, 21, 1294-1295.	2.2	33
152	Toward Precision Medicine: TBC1D4 Disruption Is Common Among the Inuit and Leads to Underdiagnosis of Type 2 Diabetes. Diabetes Care, 2016, 39, 1889-1895.	8.6	33
153	Life-threatening viral disease in a novel form of autosomal recessive IFNAR2 deficiency in the Arctic. Journal of Experimental Medicine, 2022, 219, .	8.5	33
154	Missense variants in ATP1A3 and FXYD gene family are associated with childhood-onset schizophrenia. Molecular Psychiatry, 2020, 25, 821-830.	7.9	32
155	Study of three intragenic polymorphisms in the Machado-Joseph disease gene (MJD1) in relation to genetic instability of the (CAG)n tract. European Journal of Human Genetics, 1999, 7, 147-156.	2.8	31
156	Full sequencing and haplotype analysis of MAPT in Parkinson's disease and rapid eye movement sleep behavior disorder. Movement Disorders, 2018, 33, 1016-1020.	3.9	31
157	Targeted sequencing of Parkinsonâ€™s disease loci genes highlights SYT11, FGF20 and other associations. Brain, 2021, 144, 462-472.	7.6	31
158	Synaptic processes and immune-related pathways implicated in Tourette syndrome. Translational Psychiatry, 2021, 11, 56.	4.8	31
159	Lack of association between bipolar disorder and tyrosine hydroxylase. , 1997, 74, 348-352.		30
160	LRRK2 mutations in Parkinson disease; a sex effect or lack thereof? A meta-analysis. Parkinsonism and Related Disorders, 2015, 21, 778-782.	2.2	30
161	Identification of three polymorphisms in the translated region of PLC-?1 and their investigation in lithium responsive bipolar disorder. American Journal of Medical Genetics Part A, 2001, 105, 301-305.	2.4	29
162	Mutational analysis of neurotensin in familial restless legs syndrome. Movement Disorders, 2004, 19, 90-94.	3.9	29

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163	Teneurin transmembrane protein 4 is not a cause for essential tremor in a Canadian population. Movement Disorders, 2017, 32, 292-295.	3.9	29
164	De Novo Pathogenic Variants in N-cadherin Cause a Syndromic Neurodevelopmental Disorder with Corpus Callosum, Axon, Cardiac, Ocular, and Genital Defects. American Journal of Human Genetics, 2019, 105, 854-868.	6.2	29
165	Autosomal dominant primary lateral sclerosis. Neurology, 2007, 68, 1156-1157.	1.1	28
166	Identification of novelFUSmutations in sporadic cases of amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2011, 12, 113-117.	2.1	28
167	Dopamine transporter SLC6A3 genotype affects cortico-striatal activity of set-shifts in Parkinsonâ€™s disease. Brain, 2014, 137, 3025-3035.	7.6	28
168	De novo FUS P525L mutation in Juvenile amyotrophic lateral sclerosis with dysphonia and diplopia. Neurology: Genetics, 2016, 2, e63.	1.9	28
169	Analysis of the Influence of microRNAs in Lithium Response in Bipolar Disorder. Frontiers in Psychiatry, 2018, 9, 207.	2.6	28
170	A Physiological Instability Displayed in Hippocampal Neurons Derived From Lithium-Nonresponsive Bipolar Disorder Patients. Biological Psychiatry, 2020, 88, 150-158.	1.3	28
171	Genetics of essential tremor: From phenotype to genes, insights from both human and mouse studies. Progress in Neurobiology, 2014, 119-120, 1-19.	5.7	27
172	Conserved pharmacological rescue of hereditary spastic paraplegia-related phenotypes across model organisms. Human Molecular Genetics, 2016, 25, 1088-1099.	2.9	27
173	MEIS1 and Restless Legs Syndrome: A Comprehensive Review. Frontiers in Neurology, 2019, 10, 935.	2.4	27
174	Lack of association between the hSKCa3 channel gene CAG polymorphism and schizophrenia. , 1999, 88, 154-157.		26
175	Association and linkage studies of CRH and PENK genes in bipolar disorder: A collaborative IGSLI study. , 2000, 96, 178-181.		26
176	Moyamoya Disease Susceptibility Gene RNF213 Regulates Endothelial Barrier Function. Stroke, 2022, 53, 1263-1275.	2.0	26
177	A Point Mutation in the Ubiquitin Ligase RNF170 That Causes Autosomal Dominant Sensory Ataxia Destabilizes the Protein and Impairs Inositol 1,4,5-Trisphosphate Receptor-mediated Ca2+ Signaling. Journal of Biological Chemistry, 2015, 290, 13948-13957.	3.4	25
178	The Tanenbaum Open Science Institute: Leading a Paradigm Shift at the Montreal Neurological Institute. Neuron, 2017, 95, 1002-1006.	8.1	25
179	The dementia-associated APOE Îµ4 allele is not associated with rapid eye movement sleep behavior disorder. Neurobiology of Aging, 2017, 49, 218.e13-218.e15.	3.1	25
180	LRRK2 protective haplotype and full sequencing study in REM sleep behavior disorder. Parkinsonism and Related Disorders, 2018, 52, 98-101.	2.2	25

#	Article	IF	Citations
181	Genome-Wide Association Study Meta-Analysis for Parkinson Disease Motor Subtypes. Neurology: Genetics, 2021, 7, e557.	1.9	25
182	Combining schizophrenia and depression polygenic risk scores improves the genetic prediction of lithium response in bipolar disorder patients. Translational Psychiatry, 2021, 11, 606.	4.8	25
183	Family-based association study of common variants, rare mutation study and epistatic interaction detection in HDAC genes in schizophrenia. Schizophrenia Research, 2014, 160, 97-103.	2.0	23
184	A rare variant in MLKL confers susceptibility to ApoE É›4-negative Alzheimer's disease in Hong Kong Chinese population. Neurobiology of Aging, 2018, 68, 160.e1-160.e7.	3.1	23
185	A direct interaction between two Restless Legs Syndrome predisposing genes: MEIS1 and SKOR1. Scientific Reports, 2018, 8, 12173.	3.3	23
186	LINGO1 Variants in the French-Canadian Population. PLoS ONE, 2011, 6, e16254.	2.5	23
187	Comparison of Sequencing Based CNV Discovery Methods Using Monozygotic Twin Quartets. PLoS ONE, 2015, 10, e0122287.	2.5	22
188	Exome sequencing identifies recessive CDK5RAP2 variants in patients with isolated agenesis of corpus callosum. European Journal of Human Genetics, 2016, 24, 607-610.	2.8	22
189	Dysfunction of the Cerebral Glucose Transporter SLC45A1 in Individuals with Intellectual Disability and Epilepsy. American Journal of Human Genetics, 2017, 100, 824-830.	6.2	22
190	Exome sequencing in genetic disease: recent advances and considerations. F1000Research, 2020, 9, 336.	1.6	22
191	Adenylosuccinate lyase (ADSL) and infantile autism: Absence of previously reported point mutation. American Journal of Medical Genetics Part A, 1995, 60, 554-557.	2.4	21
192	A new approach for rare variation collapsing on functional protein domains implicates specific genic regions in ALS. Genome Research, 2019, 29, 809-818.	5.5	21
193	CAPN1 mutations: Expanding the CAPN1-related phenotype: From hereditary spastic paraparesis to spastic ataxia. European Journal of Medical Genetics, 2019, 62, 103605.	1.3	21
194	Characterization of the phenotype with cognitive impairment and protein mislocalization in SCA34. Neurology: Genetics, 2020, 6, e403.	1.9	21
195	Is Persistent Motor or Vocal Tic Disorder a Milder Form of Tourette Syndrome?. Movement Disorders, 2021, 36, 1899-1910.	3.9	21
196	Analysis of functional GLO1 variants in the BTBD9 locus and restless legs syndrome. Sleep Medicine, 2015, 16, 1151-1155.	1.6	20
197	Genetic markers of Restless Legs Syndrome in Parkinson disease. Parkinsonism and Related Disorders, 2015, 21, 582-585.	2.2	20
198	Identification of a rare BMP pathway mutation in a non-syndromic human brain arteriovenous malformation via exome sequencing. Human Genome Variation, 2018, 5, 18001.	0.7	20

#	Article	IF	Citations
199	Clinical and genetic analysis of ATP13A2 in hereditary spastic paraplegia expands the phenotype. Molecular Genetics & Genomic Medicine, 2020, 8, e1052.	1.2	20
200	Characterisation of age and polarity at onset in bipolar disorder. British Journal of Psychiatry, 2021, 219, 659-669.	2.8	20
201	A Novel Nonsense Mutation in SCN9A in a Moroccan Child With Congenital Insensitivity to Pain. Pediatric Neurology, 2014, 51, 741-744.	2.1	19
202	Analysis of common and rare VPS13C variants in late-onset Parkinson disease. Neurology: Genetics, 2020, 6, 385.	1.9	19
203	Association study of DNAJC13, UCHL1, HTRA2, GIGYF2, and EIF4G1 with Parkinson's disease. Neurobiology of Aging, 2021, 100, 119.e7-119.e13.	3.1	19
204	Prenatal diagnosis of Machadoâ€“Joseph disease by direct mutation analysis. , 1998, 18, 611-617.		17
205	Mutation Burden of Rare Variants in Schizophrenia Candidate Genes. PLoS ONE, 2015, 10, e0128988.	2.5	17
206	Molecular, Cellular, and Genetic Determinants of Sporadic Brain Arteriovenous Malformations. Neurosurgery, 2016, 63, 37-42.	1.1	17
207	POLR3A variants in hereditary spastic paraplegia and ataxia. Brain, 2018, 141, e1-e1.	7.6	17
208	Chronic lithium treatment alters the excitatory/inhibitory balance of synaptic networks and reduces mGluR5â€“PKC signalling in mouse cortical neurons. Journal of Psychiatry and Neuroscience, 2021, 46, E402-E414.	2.4	17
209	Association of Essential Tremor With Novel Risk Loci. JAMA Neurology, 2022, 79, 185.	9.0	17
210	FET proteins regulate lifespan and neuronal integrity. Scientific Reports, 2016, 6, 25159.	3.3	16
211	Reassessing GWAS findings for the shared genetic basis of insomnia and restless legs syndrome. Sleep, 2018, 41, .	1.1	16
212	Sequencing of the GBA coactivator, Saposin C, in Parkinson disease. Neurobiology of Aging, 2018, 72, 187.e1-187.e3.	3.1	16
213	Exemplar scoring identifies genetically separable phenotypes of lithium responsive bipolar disorder. Translational Psychiatry, 2021, 11, 36.	4.8	16
214	Abnormal activity of membrane phospholipid synthetic enzymes in the brain of patients with Friedreich's ataxia and spinocerebellar atrophy type-1. Movement Disorders, 2000, 15, 294-300.	3.9	15
215	Increased Missense Mutation Burden of Fatty Acid Metabolism Related Genes in Nunavik Inuit Population. PLoS ONE, 2015, 10, e0128255.	2.5	15
216	SPTAN1 variants as a potential cause for autosomal recessive hereditary spastic paraplegia. Journal of Human Genetics, 2019, 64, 1145-1151.	2.3	15

#	Article	IF	Citations
217	Assessing non-Mendelian inheritance in inherited axonopathies. Genetics in Medicine, 2020, 22, 2114-2119.	2.4	15
218	Influence of polygenic risk scores for schizophrenia and resilience on the cognition of individuals at-risk for psychosis. Translational Psychiatry, 2021, 11, 518.	4.8	15
219	Genetic architecture and adaptations of Nunavik Inuit. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 16012-16017.	7.1	14
220	Prospective head-to-head comparison of accuracy of two sequencing platforms for screening for fetal aneuploidy by cell-free DNA: the PEGASUS study. European Journal of Human Genetics, 2019, 27, 1701-1715.	2.8	14
221	RNA-Based Therapy Utilizing Oculopharyngeal Muscular Dystrophy Transcript Knockdown and Replacement. Molecular Therapy - Nucleic Acids, 2019, 15, 12-25.	5.1	14
222	Oligogenicity, C9orf72 expansion, and variant severity in ALS. Neurogenetics, 2020, 21, 227-242.	1.4	13
223	Variants in the Niemannâ€“Pick type C gene NPC1 are not associated with Parkinson's disease. Neurobiology of Aging, 2020, 93, 143.e1-143.e4.	3.1	13
224	Genetics of primary lateral sclerosis. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2020, 21, 28-34.	1.7	13
225	Gender equality in Machadoâ€“Joseph disease. Nature Genetics, 1995, 11, 118-119.	21.4	12
226	Alopecia areata universalis in an infant. Journal of Cutaneous Medicine and Surgery, 2001, 5, 131-134.	1.2	12
227	A Locus for Primary Lateral Sclerosis on Chromosome 4ptel-4p16.1. Archives of Neurology, 2008, 65, 383-6.	4.5	12
228	The role of the melanoma gene MC1R in Parkinson disease and REM sleep behavior disorder. Neurobiology of Aging, 2016, 43, 180.e7-180.e13.	3.1	12
229	Genome-wide association analysis identifies new candidate risk loci for familial intracranial aneurysm in the French-Canadian population. Scientific Reports, 2018, 8, 4356.	3.3	12
230	Neural function in DCC mutation carriers with and without mirror movements. Annals of Neurology, 2019, 85, 433-442.	5.3	12
231	Transcriptome-wide association study for restless legs syndrome identifies new susceptibility genes. Communications Biology, 2020, 3, 373.	4.4	12
232	Assessing the NOTCH2NLC GGC expansion in European patients with essential tremor. Brain, 2020, 143, e89-e89.	7.6	12
233	Novel Associations of BST1 and LAMP3 With REM Sleep Behavior Disorder. Neurology, 2021, 96, e1402-e1412.	1.1	12
234	SYNE1 mutations cause autosomalâ€recessive ataxia with retained reflexes in Brazilian patients. Movement Disorders, 2016, 31, 1754-1756.	3.9	11

#	Article	IF	Citations
235	TOX3 Variants Are Involved in Restless Legs Syndrome and Parkinsonâ€™s Disease with Opposite Effects. Journal of Molecular Neuroscience, 2018, 64, 341-345.	2.3	11
236	Triple A syndrome presenting as complicated hereditary spastic paraplegia. Molecular Genetics & Genomic Medicine, 2018, 6, 1134-1139.	1.2	11
237	Multimodal neuroimaging analysis in patients with SYNE1 Ataxia. Journal of the Neurological Sciences, 2018, 390, 227-230.	0.6	11
238	Transcriptomic Changes Resulting From STK32B Overexpression Identify Pathways Potentially Relevant to Essential Tremor. Frontiers in Genetics, 2020, 11, 813.	2.3	11
239	SKOR1 has a transcriptional regulatory role on genes involved in pathways related to restless legs syndrome. European Journal of Human Genetics, 2020, 28, 1520-1528.	2.8	11
240	Multiomics Analyses Identify Genes and Pathways Relevant to Essential Tremor. Movement Disorders, 2020, 35, 1153-1162.	3.9	11
241	Comprehensive Analysis of Familial Parkinsonism Genes in Rapidâ€Eyeâ€Movement Sleep Behavior Disorder. Movement Disorders, 2021, 36, 235-240.	3.9	11
242	Exome-wide rare variant analysis in familial essential tremor. Parkinsonism and Related Disorders, 2021, 82, 109-116.	2.2	11
243	Evidence for Nonâ€Mendelian Inheritance in Spastic Paraplegia 7. Movement Disorders, 2021, 36, 1664-1675.	3.9	11
244	Using polygenic scores and clinical data for bipolar disorder patient stratification and lithium response prediction: machine learning approach. British Journal of Psychiatry, 2022, 220, 219-228.	2.8	11
245	Epistasis analysis links immune cascades and cerebral amyloidosis. Journal of Neuroinflammation, 2015, 12, 227.	7.2	10
246	Genome-wide estimates of heritability and genetic correlations in essential tremor. Parkinsonism and Related Disorders, 2019, 64, 262-267.	2.2	10
247	A polymorphism in the glutamate metabotropic receptor 7 is associated with cognitive deficits in the early phases of psychosis. Schizophrenia Research, 2022, 249, 56-62.	2.0	10
248	HLA-DRB1 and HLA-DQB1 genetic diversity modulates response to lithium in bipolar affective disorders. Scientific Reports, 2021, 11, 17823.	3.3	10
249	Genome-wide association study identifies genetic factors that modify age at onset in Machado-Joseph disease. Aging, 2020, 12, 4742-4756.	3.1	10
250	Association study of essential tremor genetic loci in Parkinson'sÂdisease. Neurobiology of Aging, 2018, 66, 178.e13-178.e15.	3.1	9
251	Mineral absorption is an enriched pathway in a brain region of restless legs syndrome patients with reduced MEIS1 expression. PLoS ONE, 2019, 14, e0225186.	2.5	9
252	Investigating the association and causal relationship between restless legs syndrome and essential tremor. Parkinsonism and Related Disorders, 2019, 61, 238-240.	2.2	9

#	Article	IF	Citations
253	Genetic and epidemiological characterization of restless legs syndrome in QuÃ©bec. Sleep, 2020, 43, .	1.1	9
254	KCC3 axonopathy: neuropathological features in the central and peripheral nervous system. Modern Pathology, 2016, 29, 962-976.	5.5	8
255	Calpain 1 in neurodegeneration: a therapeutic target?. Lancet Neurology, The, 2016, 15, 1118.	10.2	8
256	KCC3 loss-of-function contributes to Andermann syndrome by inducing activity-dependent neuromuscular junction defects. Neurobiology of Disease, 2017, 106, 35-48.	4.4	8
257	CYP2C19 variant mitigates Alzheimer disease pathophysiology in vivo and postmortem. Neurology: Genetics, 2018, 4, e216.	1.9	8
258	Valproic acid is protective in cellular and worm models of oculopharyngeal muscular dystrophy. Neurology, 2018, 91, e551-e561.	1.1	8
259	Somatic expansion of the C9orf72 hexanucleotide repeat does not occur in ALS spinal cord tissues. Neurology: Genetics, 2019, 5, e317.	1.9	8
260	Exome sequencing of sporadic childhoodâ€onset schizophrenia suggests the contribution of Xâ€linked genes in males. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2019, 180, 335-340.	1.7	8
261	A potential role for zinc in restless legs syndrome. Sleep, 2021, 44, .	1.1	8
262	Newborn Apnea Caused by a Neurofibroma at the Craniocervical Junction. Canadian Journal of Neurological Sciences, 1994, 21, 64-66.	0.5	7
263	Family density of alcoholism and linkage information in the analysis of the COGA data. Genetic Epidemiology, 1999, 17, S361-S366.	1.3	7
264	A 23 years follow-up study identifies GLUT1 deficiency syndrome initially diagnosed as complicated hereditary spastic paraplegia. European Journal of Medical Genetics, 2016, 59, 564-568.	1.3	7
265	Screening of novel restless legs syndromeâ€“associated genes in French-Canadian families. Neurology: Genetics, 2018, 4, e296.	1.9	7
266	Non-invasive prenatal aneuploidy testing: Critical diagnostic performance parameters predict sample z-score values. Clinical Biochemistry, 2018, 59, 69-77.	1.9	7
267	Machine learning analysis of exome trios to contrast the genomic architecture of autism and schizophrenia. BMC Psychiatry, 2020, 20, 92.	2.6	7
268	Expanded <scp>CAG</scp> Repeats in <scp>ATXN1</scp>, <scp>ATXN2</scp>, <scp>ATXN3</scp>, and <scp>HTT</scp> in the 1000 Genomes Project. Movement Disorders, 2021, 36, 514-518.	3.9	7
269	Rapid Generation of Ventral Spinal Cord-like Astrocytes from Human iPSCs for Modeling Non-Cell Autonomous Mechanisms of Lower Motor Neuron Disease. Cells, 2022, 11, 399.	4.1	7
270	Genetic, structural and clinical analysis of spastic paraplegia 4. Parkinsonism and Related Disorders, 2022, 98, 62-69.	2.2	7

#	Article	IF	Citations
271	Vanishing White Matter Disease in French-Canadian Patients From Quebec. Pediatric Neurology, 2014, 51, 225-232.	2.1	6
272	Identification of a novel homozygous SPG7 mutation by whole exome sequencing in a Greek family with a complicated form of hereditary spastic paraplegia. European Journal of Medical Genetics, 2015, 58, 573-577.	1.3	6
273	Cognitive and Psychiatric Evaluation in SYNE1 Ataxia. Cerebellum, 2019, 18, 731-737.	2.5	6
274	Diagnostic Yield of Whole Exome Sequencing for Adults with Ataxia: a Brazilian Perspective. Cerebellum, 2022, 21, 49-54.	2.5	6
275	Polygenic scores differentially predict developmental trajectories of subtypes of social withdrawal in childhood. Journal of Child Psychology and Psychiatry and Allied Disciplines, 2021, 62, 1320-1329.	5.2	6
276	Diagnosing Zygosity in Infant Twins: Physical Similarity, Genotyping, and Chorionicity. Twin Research and Human Genetics, 2003, 6, 479-485.	1.0	6
277	Novel SIL1 mutations cause cerebellar ataxia and atrophy in a French-Canadian family. Neurogenetics, 2015, 16, 315-318.	1.4	5
278	RIC3 variants are not associated with Parkinson's disease in French-Canadians and French. Neurobiology of Aging, 2017, 53, 194.e9-194.e11.	3.1	5
279	Heterozygous PINK1 p.G411S in rapid eye movement sleep behaviour disorder. Brain, 2017, 140, e32-e32.	7.6	5
280	No rare deleterious variants from STK32B , PPARGC1A , and CTNNA3 are associated with essential tremor. Neurology: Genetics, 2017, 3, e195.	1.9	5
281	<scp>GCH1</scp> mutations in hereditary spastic paraplegia. Clinical Genetics, 2021, 100, 51-58.	2.0	5
282	Hereditary spastic paraplegia initially diagnosed as cerebral palsy. Clinical Parkinsonism & Related Disorders, 2021, 5, 100114.	0.9	5
283	A Founder Mutation in French-Canadian Families with X-linked Hereditary Neuropathy. Canadian Journal of Neurological Sciences, 2001, 28, 51-55.	0.5	4
284	Genome-wide association study in FTD: divide to conquer. Lancet Neurology, The, 2014, 13, 643-644.	10.2	4
285	Clinical features associated with an early onset in chronic tic disorders. Psychiatry Research, 2015, 230, 745-748.	3.3	4
286	Reliability and correlation of mixture cell correction in methylomic and transcriptomic blood data. BMC Research Notes, 2020, 13, 74.	1.4	4
287	SMPD1 variants do not have a major role in rapid eye movement sleep behavior disorder. Neurobiology of Aging, 2020, 93, 142.e5-142.e7.	3.1	4
288	Transcriptome-wide association study reveals increased neuronal FLT3 expression is associated with Touretteâ€™s syndrome. Communications Biology, 2022, 5, 289.	4.4	4

#	Article	IF	Citations
289	The Genetic and Molecular Analyses of RAD51C and RAD51D Identifies Rare Variants Implicated in Hereditary Ovarian Cancer from a Genetically Unique Population. Cancers, 2022, 14, 2251.	3.7	4
290	Modeling the phenotype in parametric linkage analysis of bipolar disorder. Genetic Epidemiology, 1997, 14, 687-691.	1.3	3
291	Genetic and Epidemiological Study of Adult Ataxia and Spastic Paraplegia in Eastern Quebec. Canadian Journal of Neurological Sciences, 2021, 48, 655-665.	0.5	3
292	Lack of Causal Effects or Genetic Correlation between Restless Legs Syndrome and Parkinson's Disease. Movement Disorders, 2021, 36, 1967-1972.	3.9	3
293	Occurrence of Amyotrophic Lateral Sclerosis in Type 1 Gaucher Disease. Neurology: Genetics, 2021, 7, e600.	1.9	3
294	Rare PSAP Variants and Possible Interaction with GBA in REM Sleep Behavior Disorder. Journal of Parkinson's Disease, 2022, 12, 333-340.	2.8	3
295	Stress, Cortisol and NR3C1 in At-Risk Individuals for Psychosis: A Mendelian Randomization Study. Frontiers in Psychiatry, 2020, 11, 680.	2.6	3
296	Whole-Exome Sequencing in Congenital Hypothyroidism Due to Thyroid Dysgenesis. Thyroid, 2022, 32, 486-495.	4.5	3
297	Polymorphism in the cell division cycle 45 like gene and schizophrenia. American Journal of Medical Genetics Part A, 2001, 105, 214-215.	2.4	2
298	Alopecia Areata Universalis in an Infant. Journal of Cutaneous Medicine and Surgery, 2001, 5, 131-134.	1.2	2
299	Exome sequencing revealed PMM2 gene mutations in a French-Canadian family with congenital atrophy of the cerebellum. Cerebellum and Ataxias, 2014, 1, 8.	1.9	2
300	Post-concussion symptoms and chronic pain after mild traumatic brain injury are modulated by multiple locus effect in the BDNF gene through the expression of antisense: A pilot prospective control study. Canadian Journal of Pain, 2017, 1, 112-126.	1.7	2
301	Exome sequencing reveals a novel PLP1 mutation in a Moroccan family with connatal Pelizaeus-Merzbacher disease: a case report. BMC Pediatrics, 2018, 18, 90.	1.7	2
302	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.	4.0	2
303	Genetics of REM Sleep Behavior Disorder. , 2019, , 589-609.		2
304	Evolutionary Design and Experimental Evaluation of Selective Hammerhead Ribozymes. , 2020, , .		2
305	Lack of association of TP73 with amyotrophic lateral sclerosis in a large cohort of cases. Neurobiology of Aging, 2022, 115, 109-111.	3.1	2
306	Caseâ€“Control and Familyâ€Based Association Study of Specific <scp>PTPRD</scp> Variants in Restless Legs Syndrome. Movement Disorders Clinical Practice, 2016, 3, 460-464.	1.5	1

#	Article	IF	Citations
307	The Puzzle of Huntington Disease Phenocopies. JAMA Neurology, 2016, 73, 1056.	9.0	1
308	Schizophrenia and chromosome 6p. American Journal of Medical Genetics Part A, 1997, 74, 195-198.	2.4	1
309	Heterozygous De Novo <scp>KPNA3</scp> Mutations Cause Complex Hereditary Spastic Paraplegia. Annals of Neurology, 2022, 91, 730-732.	5.3	1
310	Questioning the Association of the STMN2 Dinucleotide Repeat With Amyotrophic Lateral Sclerosis. Neurology: Genetics, 2022, 8, e678.	1.9	1
311	Reply to Bellivier et al.. American Journal of Medical Genetics Part A, 1998, 81, 351-352.	2.4	0
312	Strategies for studying the epilepsy genome. Epilepsia, 2010, 51, 58-58.	5.1	0
313	IC-P-176: EPISTASIS ANALYSES INDICATE ASSOCIATION BETWEEN CEREBRAL AMYLOID DEPOSITION AND GENES INVOLVED IN IMMUNO-RESPONSE. , 2014, 10, P98-P99.		0
314	P1-049: EPISTASIS ANALYSES INDICATE ASSOCIATION BETWEEN CEREBRAL AMYLOID DEPOSITION AND GENES INVOLVED IN IMMUNORESPONSE. , 2014, 10, P321-P321.		0
315	<scp>KCNA2</scp> mutations are rare in hereditary spastic paraplegia. Annals of Neurology, 2017, 81, 325-326.	5.3	0
316	[P1â€“156]: FAMILIAL AGGREGATION OF ATYPICAL DEMENTIA IN A LARGE CANADIAN FAMILY: THE MISSING GENE. Alzheimer's and Dementia, 2017, 13, P302.	0.8	0
317	A polyalanine antibody for the diagnosis of oculopharyngeal muscular dystrophy and polyalanine-related diseases. MNI Open Research, 0, 1, 1.	1.0	0
318	Effect of preexamination conditions in a centralized-testing model of non-invasive prenatal screening. Clinical Chemistry and Laboratory Medicine, 2021, .	2.3	0
319	RABENOSYN separation-of-function mutations uncouple endosomal recycling from lysosomal degradation, causing a distinct Mendelian Disorder. Human Molecular Genetics, 0, , .	2.9	0
320	Progress in the genetics of restless legs syndrome: the path ahead in the era of whole-genome sequencing. Sleep, 0, , .	1.1	0