Guy A Rouleau

List of Publications by Year
in descending order

320 papers	36,600 citations	8755 77 h-index	4414 178 g-index
408 all docs	408 docs citations	408 times ranked	38431 citing authors

#	Article	IF	Citations
1	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.	1.1	10
2	Diagnostic Yield of Whole Exome Sequencing for Adults with Ataxia: a Brazilian Perspective. Cerebellum, 2022, 21, 49-54.	1.4	6
3	Dissecting the Shared Genetic Architecture of Suicide Attempt, Psychiatric Disorders, and Known Risk Factors. Biological Psychiatry, 2022, 91, 313-327.	0.7	114
4	Rare PSAP Variants and Possible Interaction with GBA in REM Sleep Behavior Disorder. Journal of Parkinson's Disease, 2022, 12, 333-340.	1.5	3
5	Heterozygous De Novo <scp>KPNA3</scp> Mutations Cause Complex Hereditary Spastic Paraplegia. Annals of Neurology, 2022, 91, 730-732.	2.8	1
6	Moyamoya Disease Susceptibility Gene RNF213 Regulates Endothelial Barrier Function. Stroke, 2022, 53, 1263-1275.	1.0	26
7	Association of Essential Tremor With Novel Risk Loci. JAMA Neurology, 2022, 79, 185.	4.5	17
8	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.	1.8	7
9	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.	1.7	11
10	Genome-wide study of DNA methylation shows alterations in metabolic, inflammatory, and cholesterol pathways in ALS. Science Translational Medicine, 2022, 14, eabj0264.	5.8	38
11	Whole-Exome Sequencing in Congenital Hypothyroidism Due to Thyroid Dysgenesis. Thyroid, 2022, 32, 486-495.	2.4	3
12	Transcriptome-wide association study reveals increased neuronal FLT3 expression is associated with Tourette's syndrome. Communications Biology, 2022, 5, 289.	2.0	4
13	Lack of association of TP73 with amyotrophic lateral sclerosis in a large cohort of cases. Neurobiology of Aging, 2022, 115, 109-111.	1.5	2
14	Life-threatening viral disease in a novel form of autosomal recessive IFNAR2 deficiency in the Arctic. Journal of Experimental Medicine, 2022, 219, .	4.2	33
15	Genetic, structural and clinical analysis of spastic paraplegia 4. Parkinsonism and Related Disorders, 2022, 98, 62-69.	1.1	7
16	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.	1.7	4
17	Questioning the Association of the STMN2 Dinucleotide Repeat With Amyotrophic Lateral Sclerosis. Neurology: Genetics, 2022, 8, e678.	0.9	1
18	Association of polygenic score for major depression with response to lithium in patients with bipolar disorder. Molecular Psychiatry, 2021, 26, 2457-2470.	4.1	44

#	Article	IF	Citations
19	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.	2.2	7
20	Comprehensive Analysis of Familial Parkinsonism Genes in Rapidâ€Eyeâ€Movement Sleep Behavior Disorder. Movement Disorders, 2021, 36, 235-240.	2.2	11
21	Analysis of Heterozygous <scp>PRKN</scp> Variants and Copyâ€Number Variations in Parkinson's Disease. Movement Disorders, 2021, 36, 178-187.	2.2	39
22	A potential role for zinc in restless legs syndrome. Sleep, 2021, 44, .	0.6	8
23	Targeted sequencing of Parkinsonâ€™s disease loci genes highlights SYT11, FGF20 and other associations. Brain, 2021, 144, 462-472.	3.7	31
24	Exome-wide rare variant analysis in familial essential tremor. Parkinsonism and Related Disorders, 2021, 82, 109-116.	1.1	11
25	Exemplar scoring identifies genetically separable phenotypes of lithium responsive bipolar disorder. Translational Psychiatry, 2021, 11, 36.	2.4	16
26	Synaptic processes and immune-related pathways implicated in Tourette syndrome. Translational Psychiatry, 2021, 11, 56.	2.4	31
27	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
28	Genome-Wide Association Study Meta-Analysis for Parkinson Disease Motor Subtypes. Neurology: Genetics, 2021, 7, e557.	0.9	25
29	Evidence for Nonâ€Mendelian Inheritance in Spastic Paraplegia 7. Movement Disorders, 2021, 36, 1664-1675.	2.2	11
30	<scp>GCH1</scp> mutations in hereditary spastic paraplegia. Clinical Genetics, 2021, 100, 51-58.	1.0	5
31	Association study of DNAJC13, UCHL1, HTRA2, GIGYF2, and EIF4G1 with Parkinson's disease. Neurobiology of Aging, 2021, 100, 119.e7-119.e13.	1.5	19
32	Is Persistent Motor or Vocal Tic Disorder a Milder Form of Tourette Syndrome?. Movement Disorders, 2021, 36, 1899-1910.	2.2	21
33	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.	9.4	629
34	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.	1.4	17
35	Lack of Causal Effects or Genetic Correlation between Restless Legs Syndrome and Parkinson's Disease. Movement Disorders, 2021, 36, 1967-1972.	2.2	3
36	Occurrence of Amyotrophic Lateral Sclerosis in Type 1 Gaucher Disease. Neurology: Genetics, 2021, 7, e600.	0.9	3

#	Article	IF	Citations
37	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.	3.1	6
38	Characterisation of age and polarity at onset in bipolar disorder. British Journal of Psychiatry, 2021, 219, 659-669.	1.7	20
39	Association of Variants in the SPTLC1 Gene With Juvenile Amyotrophic Lateral Sclerosis. JAMA Neurology, 2021, 78, 1236.	4.5	46
40	HLA-DRB1 and HLA-DQB1 genetic diversity modulates response to lithium in bipolar affective disorders. Scientific Reports, 2021, 11, 17823.	1.6	10
41	Novel Associations of BST1 and LAMP3 With REM Sleep Behavior Disorder. Neurology, 2021, 96, e1402-e1412.	1.5	12
42	Influence of polygenic risk scores for schizophrenia and resilience on the cognition of individuals at-risk for psychosis. Translational Psychiatry, 2021, 11, 518.	2.4	15
43	Hereditary spastic paraplegia initially diagnosed as cerebral palsy. Clinical Parkinsonism & Related Disorders, 2021, 5, 100114.	0.5	5
44	Effect of preexamination conditions in a centralized-testing model of non-invasive prenatal screening. Clinical Chemistry and Laboratory Medicine, 2021, .	1.4	0
45	Enrichment of SARM1 alleles encoding variants with constitutively hyperactive NADase in patients with ALS and other motor nerve disorders. ELife, 2021, 10, .	2.8	44
46	Combining schizophrenia and depression polygenic risk scores improves the genetic prediction of lithium response in bipolar disorder patients. Translational Psychiatry, 2021, 11, 606.	2.4	25
47	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.	9.4	223
48	Missense variants in ATP1A3 and FXYD gene family are associated with childhood-onset schizophrenia. Molecular Psychiatry, 2020, 25, 821-830.	4.1	32
49	Mechanisms Underlying the Hyperexcitability of CA3 and Dentate Gyrus Hippocampal Neurons Derived From Patients With Bipolar Disorder. Biological Psychiatry, 2020, 88, 139-149.	0.7	39
50	Genetic and epidemiological characterization of restless legs syndrome in QuÃ©bec. Sleep, 2020, 43, .	0.6	9
51	Genetic, Structural, and Functional Evidence Link TMEM175 to Synucleinopathies. Annals of Neurology, 2020, 87, 139-153.	2.8	65
52	Transcriptome-wide association study for restless legs syndrome identifies new susceptibility genes. Communications Biology, 2020, 3, 373.	2.0	12
53	Genome-wide association study of intracranial aneurysms identifies 17 risk loci and genetic overlap with clinical risk factors. Nature Genetics, 2020, 52, 1303-1313.	9.4	163
54	Evolution of a Human-Specific Tandem Repeat Associated with ALS. American Journal of Human Genetics, 2020, 107, 445-460.	2.6	39

#	Article	IF	Citations
55	Assessing non-Mendelian inheritance in inherited axonopathies. Genetics in Medicine, 2020, 22, 2114-2119.	1.1	15
56	Transcriptomic Changes Resulting From STK32B Overexpression Identify Pathways Potentially Relevant to Essential Tremor. Frontiers in Genetics, 2020, 11, 813.	1.1	11
57	Assessing the NOTCH2NLC GGC expansion in European patients with essential tremor. Brain, 2020, 143, e89-e89.	3.7	12
58	Characterization of human iPSC-derived astrocytes with potential for disease modeling and drug discovery. Neuroscience Letters, 2020, 731, 135028.	1.0	40
59	Oligogenicity, C9orf72 expansion, and variant severity in ALS. Neurogenetics, 2020, 21, 227-242.	0.7	13
60	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.	1.4	11
61	GBA variants in REM sleep behavior disorder. Neurology, 2020, 95, e1008-e1016.	1.5	45
62	A Physiological Instability Displayed in Hippocampal Neurons Derived From Lithium-Nonresponsive Bipolar Disorder Patients. Biological Psychiatry, 2020, 88, 150-158.	0.7	28
63	Machine learning analysis of exome trios to contrast the genomic architecture of autism and schizophrenia. BMC Psychiatry, 2020, 20, 92.	1.1	7
64	Reliability and correlation of mixture cell correction in methylomic and transcriptomic blood data. BMC Research Notes, 2020, 13, 74.	0.6	4
65	Fineâ€Mapping of SNCA in Rapid Eye Movement Sleep Behavior Disorder and Overt Synucleinopathies. Annals of Neurology, 2020, 87, 584-598.	2.8	39
66	The Quebec Parkinson Network: A Researcher-Patient Matching Platform and Multimodal Biorepository. Journal of Parkinson's Disease, 2020, 10, 301-313.	1.5	35
67	Clinical and genetic analysis of ATP13A2 in hereditary spastic paraplegia expands the phenotype. Molecular Genetics & Genomic Medicine, 2020, 8, e1052.	0.6	20
68	Multiomics Analyses Identify Genes and Pathways Relevant to Essential Tremor. Movement Disorders, 2020, 35, 1153-1162.	2.2	11
69	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.	1.5	13
70	Analysis of common and rare VPS13C variants in late-onset Parkinson disease. Neurology: Genetics, 2020, 6, 385.	0.9	19
71	Characterization of the phenotype with cognitive impairment and protein mislocalization in SCA34. Neurology: Genetics, 2020, 6, e403.	0.9	21
72	SMPD1 variants do not have a major role in rapid eye movement sleep behavior disorder. Neurobiology of Aging, 2020, 93, 142.e5-142.e7.	1.5	4

#	Article	IF	Citations
73	Genetics of primary lateral sclerosis. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2020, 21, 28-34.	1.1	13
74	Exome sequencing in genetic disease: recent advances and considerations. F1000Research, 2020, 9, 336.	0.8	22
75	Genome-wide association study identifies genetic factors that modify age at onset in Machado-Joseph disease. Aging, 2020, 12, 4742-4756.	1.4	10
76	Stress, Cortisol and NR3C1 in At-Risk Individuals for Psychosis: A Mendelian Randomization Study. Frontiers in Psychiatry, 2020, 11, 680.	1.3	3
77	Evolutionary Design and Experimental Evaluation of Selective Hammerhead Ribozymes. , 2020, , .		2
78	Genetic architecture and adaptations of Nunavik Inuit. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 16012-16017.	3.3	14
79	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.	1.4	14
80	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.	4.9	1,414
81	Mineral absorption is an enriched pathway in a brain region of restless legs syndrome patients with reduced MEIS1 expression. PLoS ONE, 2019, 14, e0225186.	1.1	9
82	SPTAN1 variants as a potential cause for autosomal recessive hereditary spastic paraplegia. Journal of Human Genetics, 2019, 64, 1145-1151.	1.1	15
83	Transcriptome-wide association study of attention deficit hyperactivity disorder identifies associated genes and phenotypes. Nature Communications, 2019, 10, 4450.	5.8	56
84	MEIS1 and Restless Legs Syndrome: A Comprehensive Review. Frontiers in Neurology, 2019, 10, 935.	1.1	27
85	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.	2.6	29
86	Neural function in DCC mutation carriers with and without mirror movements. Annals of Neurology, 2019, 85, 433-442.	2.8	12
87	Genome-wide estimates of heritability and genetic correlations in essential tremor. Parkinsonism and Related Disorders, 2019, 64, 262-267.	1.1	10
88	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.	1.4	38
89	Somatic expansion of the C9orf72 hexanucleotide repeat does not occur in ALS spinal cord tissues. Neurology: Genetics, 2019, 5, e317.	0.9	8
90	Cognitive and Psychiatric Evaluation in SYNE1 Ataxia. Cerebellum, 2019, 18, 731-737.	1.4	6

#	Article	IF	Citations
91	Genome-wide association study identifies 30 loci associated with bipolar disorder. Nature Genetics, 2019, 51, 793-803.	9.4	1,191
92	RNA-Based Therapy Utilizing Oculopharyngeal Muscular Dystrophy Transcript Knockdown and Replacement. Molecular Therapy - Nucleic Acids, 2019, 15, 12-25.	2.3	14
93	Interrogating the Genetic Determinants of Tourette's Syndrome and Other Tic Disorders Through Genome-Wide Association Studies. American Journal of Psychiatry, 2019, 176, 217-227.	4.0	242
94	Truncating Mutations in UBAP1 Cause Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2019, 104, 767-773.	2.6	39
95	A new approach for rare variation collapsing on functional protein domains implicates specific genic regions in ALS. Genome Research, 2019, 29, 809-818.	2.4	21
96	SMPD1 mutations, activity, and Î±â€synuclein accumulation in Parkinson's disease. Movement Disorders, 2019, 34, 526-535.	2.2	81
97	Investigation of the RFC1 Repeat Expansion in a Canadian and a Brazilian Ataxia Cohort: Identification of Novel Conformations. Frontiers in Genetics, 2019, 10, 1219.	1.1	51
98	Genomic Relationships, Novel Loci, and Pleiotropic Mechanisms across Eight Psychiatric Disorders. Cell, 2019, 179, 1469-1482.e11.	13.5	935
99	CAPN1 mutations: Expanding the CAPN1-related phenotype: From hereditary spastic paraparesis to spastic ataxia. European Journal of Medical Genetics, 2019, 62, 103605.	0.7	21
100	Investigating the association and causal relationship between restless legs syndrome and essential tremor. Parkinsonism and Related Disorders, 2019, 61, 238-240.	1.1	9
101	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.	1.1	8
102	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
103	Genetics of REM Sleep Behavior Disorder. , 2019, , 589-609.		2
104	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.	6.0	102
105	CYP2C19 variant mitigates Alzheimer disease pathophysiology in vivo and postmortem. Neurology: Genetics, 2018, 4, e216.	0.9	8
106	Recessive mutations in VPS13D cause childhood onset movement disorders. Annals of Neurology, 2018, 83, 1089-1095.	2.8	104
107	Sleep disorders and Parkinson disease; lessons from genetics. Sleep Medicine Reviews, 2018, 41, 101-112.	3.8	35
108	Genetics of Intracranial Aneurysms. Stroke, 2018, 49, 780-787.	1.0	60

#	Article	IF	Citations
109	TOX3 Variants Are Involved in Restless Legs Syndrome and Parkinsonâ€™s Disease with Opposite Effects. Journal of Molecular Neuroscience, 2018, 64, 341-345.	1.1	11
110	POLR3A variants in hereditary spastic paraplegia and ataxia. Brain, 2018, 141, e1-e1.	3.7	17
111	Association study of essential tremor genetic loci in Parkinson'sÂdisease. Neurobiology of Aging, 2018, 66, 178.e13-178.e15.	1.5	9
112	Exome sequencing reveals a novel PLP1 mutation in a Moroccan family with connatal Pelizaeus-Merzbacher disease: a case report. BMC Pediatrics, 2018, 18, 90.	0.7	2
113	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.	1.5	23
114	Genome-wide association analysis identifies new candidate risk loci for familial intracranial aneurysm in the French-Canadian population. Scientific Reports, 2018, 8, 4356.	1.6	12
115	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. Neuron, 2018, 97, 1268-1283.e6.	3.8	517
116	LRRK2 protective haplotype and full sequencing study in REM sleep behavior disorder. Parkinsonism and Related Disorders, 2018, 52, 98-101.	1.1	25
117	Identification of a rare BMP pathway mutation in a non-syndromic human brain arteriovenous malformation via exome sequencing. Human Genome Variation, 2018, 5, 18001.	0.4	20
118	DCCmutation update: Congenital mirror movements, isolated agenesis of the corpus callosum, and developmental split brain syndrome. Human Mutation, 2018, 39, 23-39.	1.1	41
119	Screening of novel restless legs syndromeâ€“associated genes in French-Canadian families. Neurology: Genetics, 2018, 4, e296.	0.9	7
120	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.	2.9	91
121	Triple A syndrome presenting as complicated hereditary spastic paraplegia. Molecular Genetics & Genomic Medicine, 2018, 6, 1134-1139.	0.6	11
122	Reassessing GWAS findings for the shared genetic basis of insomnia and restless legs syndrome. Sleep, 2018, 41, .	0.6	16
123	Full sequencing and haplotype analysis of MAPT in Parkinson's disease and rapid eye movement sleep behavior disorder. Movement Disorders, 2018, 33, 1016-1020.	2.2	31
124	Global characterization of copy number variants in epilepsy patients from whole genome sequencing. PLoS Genetics, 2018, 14, e1007285.	1.5	50
125	Sequencing of the GBA coactivator, Saposin C, in Parkinson disease. Neurobiology of Aging, 2018, 72, 187.e1-187.e3.	1.5	16
126	Non-invasive prenatal aneuploidy testing: Critical diagnostic performance parameters predict sample z-score values. Clinical Biochemistry, 2018, 59, 69-77.	0.8	7

#	Article	IF	Citations
127	Multimodal neuroimaging analysis in patients with SYNE1 Ataxia. Journal of the Neurological Sciences, 2018, 390, 227-230.	0.3	11
128	Analysis of the Influence of microRNAs in Lithium Response in Bipolar Disorder. Frontiers in Psychiatry, 2018, 9, 207.	1.3	28
129	Analysis of shared heritability in common disorders of the brain. Science, 2018, 360, .	6.0	1,085
130	Valproic acid is protective in cellular and worm models of oculopharyngeal muscular dystrophy. Neurology, 2018, 91, e551-e561.	1.5	8
131	A direct interaction between two Restless Legs Syndrome predisposing genes: MEIS1 and SKOR1. Scientific Reports, 2018, 8, 12173.	1.6	23
132	Teneurin transmembrane protein 4 is not a cause for essential tremor in a Canadian population. Movement Disorders, 2017, 32, 292-295.	2.2	29
133	Mutations in DCC cause isolated agenesis of the corpus callosum with incomplete penetrance. Nature Genetics, 2017, 49, 511-514.	9.4	69
134	RIC3 variants are not associated with Parkinson's disease in French-Canadians and French. Neurobiology of Aging, 2017, 53, 194.e9-194.e11.	1.5	5
135	Systematic review of autosomal recessive ataxias and proposal for a classification. Cerebellum and Ataxias, 2017, 4, 3.	1.9	49
136	Dysfunction of the Cerebral Glucose Transporter SLC45A1 in Individuals with Intellectual Disability and Epilepsy. American Journal of Human Genetics, 2017, 100, 824-830.	2.6	22
137	De Novo Coding Variants Are Strongly Associated with Tourette Disorder. Neuron, 2017, 94, 486-499.e9.	3.8	155
138	Rare Copy Number Variants in NRXN1 and CNTN6 Increase Risk for Tourette Syndrome. Neuron, 2017, 94, 1101-1111.e7.	3.8	137
139	CPT1A Missense Mutation Associated With Fatty Acid Metabolism and Reduced Height in Greenlanders. Circulation: Cardiovascular Genetics, 2017, 10, .	5.1	37
140	Heterozygous PINK1 p.G411S in rapid eye movement sleep behaviour disorder. Brain, 2017, 140, e32-e32.	3.7	5
141	Genetics of restless legs syndrome. Sleep Medicine, 2017, 31, 18-22.	0.8	40
142	<scp>KCNA2</scp> mutations are rare in hereditary spastic paraplegia. Annals of Neurology, 2017, 81, 325-326.	2.8	0
143	Clinical and genetic study of hereditary spastic paraplegia in Canada. Neurology: Genetics, 2017, 3, e122.	0.9	82
144	Clinical Spectrum of Amyotrophic Lateral Sclerosis (ALS). Cold Spring Harbor Perspectives in Medicine, 2017, 7, a024117.	2.9	149

#	Article	IF	Citations
145	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	2.6	337
146	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.	4.9	191
147	The Tanenbaum Open Science Institute: Leading a Paradigm Shift at the Montreal Neurological Institute. Neuron, 2017, 95, 1002-1006.	3.8	25
148	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.	0.6	2
149	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.	2.6	112
150	KCC3 loss-of-function contributes to Andermann syndrome by inducing activity-dependent neuromuscular junction defects. Neurobiology of Disease, 2017, 106, 35-48.	2.1	8
151	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.	1.5	25
152	[P1â€“156]: FAMILIAL AGGREGATION OF ATYPICAL DEMENTIA IN A LARGE CANADIAN FAMILY: THE MISSING GENE. Alzheimer's and Dementia, 2017, 13, P302.	0.4	0
153	No rare deleterious variants from STK32B , PPARGC1A , and CTNNA3 are associated with essential tremor. Neurology: Genetics, 2017, 3, e195.	0.9	5
154	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.	0.8	1
155	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.	0.7	36
156	KCC3 axonopathy: neuropathological features in the central and peripheral nervous system. Modern Pathology, 2016, 29, 962-976.	2.9	8
157	The role of the melanoma gene MC1R in Parkinson disease and REM sleep behavior disorder. Neurobiology of Aging, 2016, 43, 180.e7-180.e13.	1.5	12
158	Mutations in CAPN1 Cause Autosomal-Recessive Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2016, 98, 1038-1046.	2.6	96
159	De novo FUS P525L mutation in Juvenile amyotrophic lateral sclerosis with dysphonia and diplopia. Neurology: Genetics, 2016, 2, e63.	0.9	28
160	ALS: Recent Developments from Genetics Studies. Current Neurology and Neuroscience Reports, 2016, 16, 59.	2.0	55
161	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.	0.7	7
162	GBA p.T369M substitution in Parkinson disease: Polymorphism or association? A meta-analysis. Neurology: Genetics, 2016, 2, e104.	0.9	74

#	Article	IF	Citations
163	Toward Precision Medicine: TBC1D4 Disruption Is Common Among the Inuit and Leads to Underdiagnosis of Type 2 Diabetes. Diabetes Care, 2016, 39, 1889-1895.	4.3	33
164	Calpain 1 in neurodegeneration: a therapeutic target?. Lancet Neurology, The, 2016, 15, 1118.	4.9	8
165	The Puzzle of Huntington Disease Phenocopies. JAMA Neurology, 2016, 73, 1056.	4.5	1
166	NEK1 variants confer susceptibility to amyotrophic lateral sclerosis. Nature Genetics, 2016, 48, 1037-1042.	9.4	218
167	SYNE1 mutations cause autosomalâ€recessive ataxia with retained reflexes in Brazilian patients. Movement Disorders, 2016, 31, 1754-1756.	2.2	11
168	RNF213 Is Associated with Intracranial Aneurysms in the French-Canadian Population. American Journal of Human Genetics, 2016, 99, 1072-1085.	2.6	49
169	Inhibition of the kinase WNK1/HSN2 ameliorates neuropathic pain by restoring GABA inhibition. Science Signaling, 2016, 9, ra32.	1.6	43
170	CCNF mutations in amyotrophic lateral sclerosis and frontotemporal dementia. Nature Communications, 2016, 7, 11253.	5.8	174
171	Genome-wide association study in essential tremor identifies three new loci. Brain, 2016, 139, 3163-3169.	3.7	78
172	Molecular, Cellular, and Genetic Determinants of Sporadic Brain Arteriovenous Malformations. Neurosurgery, 2016, 63, 37-42.	0.6	17
173	FET proteins regulate lifespan and neuronal integrity. Scientific Reports, 2016, 6, 25159.	1.6	16
174	SCARB2 variants and glucocerebrosidase activity in Parkinsonâ€™s disease. Npj Parkinson's Disease, 2016, 2, .	2.5	36
175	Analysis of DNAJC13 mutations in French-Canadian/French cohort of Parkinson's disease. Neurobiology of Aging, 2016, 45, 212.e13-212.e17.	1.5	38
176	Genome-wide association study of 40,000 individuals identifies two novel loci associated with bipolar disorder. Human Molecular Genetics, 2016, 25, 3383-3394.	1.4	182
177	Genetic and Clinical Predictors of Deep Brain Stimulation in Youngâ€Onset Parkinson's Disease. Movement Disorders Clinical Practice, 2016, 3, 465-471.	0.8	37
178	Genetic variants associated with response to lithium treatment in bipolar disorder: a genome-wide association study. Lancet, The, 2016, 387, 1085-1093.	6.3	306
179	SPG7 mutations explain a significant proportion of French Canadian spastic ataxia cases. European Journal of Human Genetics, 2016, 24, 1016-1021.	1.4	46
180	Retention of hexanucleotide repeat-containing intron in C9orf72 mRNA: implications for the pathogenesis of ALS/FTD. Acta Neuropathologica Communications, 2016, 4, 18.	2.4	46

#	Article	IF	Citations
181	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.	1.5	40
182	Conserved pharmacological rescue of hereditary spastic paraplegia-related phenotypes across model organisms. Human Molecular Genetics, 2016, 25, 1088-1099.	1.4	27
183	De novo variants in sporadic cases of childhood onset schizophrenia. European Journal of Human Genetics, 2016, 24, 944-948.	1.4	77
184	Replication study of MATR3 in familial and sporadic amyotrophic lateral sclerosis. Neurobiology of Aging, 2016, 37, 209.e17-209.e21.	1.5	53
185	Exome sequencing identifies recessive CDK5RAP2 variants in patients with isolated agenesis of corpus callosum. European Journal of Human Genetics, 2016, 24, 607-610.	1.4	22
186	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.	1.4	77
187	Quantitative Analysis of Climbing Defects in a Drosophila Model of Neurodegenerative Disorders. Journal of Visualized Experiments, 2015, , e52741.	0.2	60
188	Epistasis analysis links immune cascades and cerebral amyloidosis. Journal of Neuroinflammation, 2015, 12, 227.	3.1	10
189	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.	1.8	86
190	Comparison of Sequencing Based CNV Discovery Methods Using Monozygotic Twin Quartets. PLoS ONE, 2015, 10, e0122287.	1.1	22
191	Increased Missense Mutation Burden of Fatty Acid Metabolism Related Genes in Nunavik Inuit Population. PLoS ONE, 2015, 10, e0128255.	1.1	15
192	Mutation Burden of Rare Variants in Schizophrenia Candidate Genes. PLoS ONE, 2015, 10, e0128988.	1.1	17
193	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.	1.6	25
194	GBA mutations are associated with Rapid Eye Movement Sleep Behavior Disorder. Annals of Clinical and Translational Neurology, 2015, 2, 941-945.	1.7	117
195	Exome sequencing in amyotrophic lateral sclerosis identifies risk genes and pathways. Science, 2015, 347, 1436-1441.	6.0	823
196	Preclinical target validation using patient-derived cells. Nature Reviews Drug Discovery, 2015, 14, 149-150.	21.5	46
197	Genetic perspective on the role of the autophagy-lysosome pathway in Parkinson disease. Autophagy, 2015, 11, 1443-1457.	4.3	217
198	Analysis of functional GLO1 variants in the BTBD9 locus and restless legs syndrome. Sleep Medicine, 2015, 16, 1151-1155.	0.8	20

#	Article	IF	Citations
199	Alteration of ornithine metabolism leads to dominant and recessive hereditary spastic paraplegia. Brain, 2015, 138, 2191-2205.	3.7	88
200	Glucocerebrosidase activity in Parkinsonâ€™s disease with and withoutGBAmutations. Brain, 2015, 138, 2648-2658.	3.7	326
201	Identification of rare protein disulfide isomerase gene variants in amyotrophic lateral sclerosis patients. Gene, 2015, 566, 158-165.	1.0	70
202	LRRK2 mutations in Parkinson disease; a sex effect or lack thereof? A meta-analysis. Parkinsonism and Related Disorders, 2015, 21, 778-782.	1.1	30
203	PMPCAmutations cause abnormal mitochondrial protein processing in patients with non-progressive cerebellar ataxia. Brain, 2015, 138, 1505-1517.	3.7	58
204	Genetic markers of Restless Legs Syndrome in Parkinson disease. Parkinsonism and Related Disorders, 2015, 21, 582-585.	1.1	20
205	Defining the genetic connection linking amyotrophic lateral sclerosis (ALS) with frontotemporal dementia (FTD). Trends in Genetics, 2015, 31, 263-273.	2.9	106
206	Joubert Syndrome in French Canadians and Identification of Mutations in CEP104. American Journal of Human Genetics, 2015, 97, 744-753.	2.6	56
207	A NewELOVL4Mutation in a Case of Spinocerebellar Ataxia With Erythrokeratodermia. JAMA Neurology, 2015, 72, 942.	4.5	34
208	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.	0.7	6
209	The emerging role of SMPD1 mutations in Parkinson's disease: Implications for future studies. Parkinsonism and Related Disorders, 2015, 21, 1294-1295.	1.1	33
210	Clinical features associated with an early onset in chronic tic disorders. Psychiatry Research, 2015, 230, 745-748.	1.7	4
211	Novel SIL1 mutations cause cerebellar ataxia and atrophy in a French-Canadian family. Neurogenetics, 2015, 16, 315-318.	0.7	5
212	Deleterious mutations in the essential mRNA metabolism factor, hGle1, in amyotrophic lateral sclerosis. Human Molecular Genetics, 2015, 24, 1363-1373.	1.4	122
213	Genome-Wide Association Study of Intracranial Aneurysm Identifies a New Association on Chromosome 7. Stroke, 2014, 45, 3194-3199.	1.0	52
214	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
215	Dopamine transporter SLC6A3 genotype affects cortico-striatal activity of set-shifts in Parkinsonâ€™s disease. Brain, 2014, 137, 3025-3035.	3.7	28
216	Expanding the Clinical Phenotype Associated WithELOVL4Mutation. JAMA Neurology, 2014, 71, 470.	4.5	110

#	Article	IF	Citations
217	Genome-wide association study reveals two new risk loci for bipolar disorder. Nature Communications, 2014, 5, 3339.	5.8	294
218	Dissection of genetic factors associated with amyotrophic lateral sclerosis. Experimental Neurology, 2014, 262, 91-101.	2.0	145
219	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.	1.5	118
220	Genetics of essential tremor: From phenotype to genes, insights from both human and mouse studies. Progress in Neurobiology, 2014, 119-120, 1-19.	2.8	27
221	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.	1.1	23
222	Exome-wide Rare Variant Analysis Identifies TUBA4A Mutations Associated with Familial ALS. Neuron, 2014, 84, 324-331.	3.8	308
223	A Novel Nonsense Mutation in SCN9A in a Moroccan Child With Congenital Insensitivity to Pain. Pediatric Neurology, 2014, 51, 741-744.	1.0	19
224	Genetically encoded impairment of neuronal <scp>KCC</scp> 2 cotransporter function in human idiopathic generalized epilepsy. EMBO Reports, 2014, 15, 766-774.	2.0	163
225	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.	1.8	33
226	Loss of Association of REEP2 with Membranes Leads to Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2014, 94, 268-277.	2.6	83
227	Vanishing White Matter Disease in French-Canadian Patients From Quebec. Pediatric Neurology, 2014, 51, 225-232.	1.0	6
228	Genome-wide association study in FTD: divide to conquer. Lancet Neurology, The, 2014, 13, 643-644.	4.9	4
229	Molecular aspects of hereditary spastic paraplegia. Experimental Cell Research, 2014, 325, 18-26.	1.2	46
230	IC-P-176: EPISTASIS ANALYSES INDICATE ASSOCIATION BETWEEN CEREBRAL AMYLOID DEPOSITION AND GENES INVOLVED IN IMMUNO-RESPONSE. , 2014, 10, P98-P99.		0
231	P1-049: EPISTASIS ANALYSES INDICATE ASSOCIATION BETWEEN CEREBRAL AMYLOID DEPOSITION AND GENES INVOLVED IN IMMUNORESPONSE. , 2014, 10, P321-P321.		0
232	ERBB4 Mutations that Disrupt the Neuregulin-ErbB4 Pathway Cause Amyotrophic Lateral Sclerosis Type 19. American Journal of Human Genetics, 2013, 93, 900-905.	2.6	123
233	C9orf72 repeat expansions are a rare genetic cause of parkinsonism. Brain, 2013, 136, 385-391.	3.7	143
234	Partitioning the Heritability of Tourette Syndrome and Obsessive Compulsive Disorder Reveals Differences in Genetic Architecture. PLoS Genetics, 2013, 9, e1003864.	1.5	241

#	Article	IF	Citations
235	The Impact of Phenotypic and Genetic Heterogeneity on Results of Genome Wide Association Studies of Complex Diseases. PLoS ONE, 2013, 8, e76295.	1.1	177
236	Deletion of C9ORF72 Results in Motor Neuron Degeneration and Stress Sensitivity in C. elegans. PLoS ONE, 2013, 8, e83450.	1.1	158
237	Assessment of Response to Lithium Maintenance Treatment in Bipolar Disorder: A Consortium on Lithium Genetics (ConLiGen) Report. PLoS ONE, 2013, 8, e65636.	1.1	156
238	Expanded ATXN3 frameshifting events are toxic in Drosophila and mammalian neuron models. Human Molecular Genetics, 2012, 21, 2211-2218.	1.4	40
239	Exome sequencing reveals SPG11 mutations causing juvenile ALS. Neurobiology of Aging, 2012, 33, 839.e5-839.e9.	1.5	87
240	Genome-Wide Association Study of Intracranial Aneurysms Confirms Role of Anril and SOX17 in Disease Risk. Stroke, 2012, 43, 2846-2852.	1.0	106
241	Exome Sequencing Identifies FUS Mutations as a Cause of Essential Tremor. American Journal of Human Genetics, 2012, 91, 313-319.	2.6	176
242	Increased exonic de novo mutation rate in individuals with schizophrenia. Nature Genetics, 2011, 43, 860-863.	9.4	392
243	Identification of novelFUSmutations in sporadic cases of amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2011, 12, 113-117.	2.3	28
244	Genome-Wide Association Study Identifies Novel Restless Legs Syndrome Susceptibility Loci on 2p14 and 16q12.1. PLoS Genetics, 2011, 7, e1002171.	1.5	163
245	LINGO1 Variants in the French-Canadian Population. PLoS ONE, 2011, 6, e16254.	1.1	23
246	Family Study of Restless Legs Syndrome in Quebec, Canada. Archives of Neurology, 2010, 67, 617-22.	4.9	63
247	Direct Measure of the De Novo Mutation Rate in Autism and Schizophrenia Cohorts. American Journal of Human Genetics, 2010, 87, 316-324.	2.6	222
248	Strategies for studying the epilepsy genome. Epilepsia, 2010, 51, 58-58.	2.6	0
249	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.	0.9	134
250	MEIS1 intronic risk haplotype associated with restless legs syndrome affects its mRNA and protein expression levels. Human Molecular Genetics, 2009, 18, 1065-1074.	1.4	85
251	TARDBP mutations in individuals with sporadic and familial amyotrophic lateral sclerosis. Nature Genetics, 2008, 40, 572-574.	9.4	1,371
252	PABPN1 polyalanine tract deletion and long expansions modify its aggregation pattern and expression. Experimental Cell Research, 2008, 314, 1652-1666.	1.2	38

#	Article	IF	Citations
253	A Locus for Primary Lateral Sclerosis on Chromosome 4ptel-4p16.1. Archives of Neurology, 2008, 65, 383-6.	4.9	12
254	Three Families With Amyotrophic Lateral Sclerosis and Frontotemporal Dementia With Evidence of Linkage to Chromosome 9p. Archives of Neurology, 2007, 64, 240.	4.9	111
255	Autosomal dominant primary lateral sclerosis. Neurology, 2007, 68, 1156-1157.	1.5	28
256	Asian Origin for the Worldwide-Spread Mutational Event in Machado-Joseph Disease. Archives of Neurology, 2007, 64, 1502.	4.9	65
257	Genome-wide association study of restless legs syndrome identifies common variants in three genomic regions. Nature Genetics, 2007, 39, 1000-1006.	9.4	633
258	Genetics of familial and sporadic amyotrophic lateral sclerosis. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2006, 1762, 956-972.	1.8	214
259	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.	1.1	111
260	Ribosomal frameshifting on MJD-1 transcripts with long CAG tracts. Human Molecular Genetics, 2005, 14, 2649-2660.	1.4	54
261	Transgenic expression of an expanded (GCG)13 repeat PABPN1 leads to weakness and coordination defects in mice. Neurobiology of Disease, 2005, 18, 528-536.	2.1	48
262	A Frameshift Deletion in Peripherin Gene Associated with Amyotrophic Lateral Sclerosis. Journal of Biological Chemistry, 2004, 279, 45951-45956.	1.6	163
263	The 14q restless legs syndrome locus in the French Canadian population. Annals of Neurology, 2004, 55, 887-891.	2.8	71
264	A mutation in theHSN2gene causes sensory neuropathy type II in a Lebanese family. Annals of Neurology, 2004, 56, 572-575.	2.8	39
265	A novel locus for pure recessive hereditary spastic paraplegia maps to 10q22.1-10q24.1. Annals of Neurology, 2004, 56, 579-582.	2.8	52
266	Mutational analysis of neurotensin in familial restless legs syndrome. Movement Disorders, 2004, 19, 90-94.	2.2	29
267	Mutation screening ofFOXP2 in individuals diagnosed with autistic disorder. American Journal of Medical Genetics Part A, 2003, 118A, 172-175.	2.4	46
268	AnALS2gene mutation causes hereditary spastic paraplegia in a Pakistani kindred. Annals of Neurology, 2003, 53, 144-145.	2.8	97
269	Hereditary motor and sensory neuropathy with agenesis of the corpus callosum. Annals of Neurology, 2003, 54, 9-18.	2.8	67
270	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.	0.8	45

#	Article	IF	Citations
271	Polymorphism, shared functions and convergent evolution of genes with sequences coding for polyalanine domains. Human Molecular Genetics, 2003, 12, 2967-2979.	1.4	103
272	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
273	A gene encoding a putative GTPase regulator is mutated in familial amyotrophic lateral sclerosis 2. Nature Genetics, 2001, 29, 166-173.	9.4	635
274	Compound heterozygous D90A and D96N SOD1 mutations in a recessive amyotrophic lateral sclerosis family. Annals of Neurology, 2001, 49, 267-271.	2.8	80
275	A Founder Mutation in French-Canadian Families with X-linked Hereditary Neuropathy. Canadian Journal of Neurological Sciences, 2001, 28, 51-55.	0.3	4
276	Alopecia Areata Universalis in an Infant. Journal of Cutaneous Medicine and Surgery, 2001, 5, 131-134.	0.6	2
277	Alopecia areata universalis in an infant. Journal of Cutaneous Medicine and Surgery, 2001, 5, 131-134.	0.6	12
278	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
279	High resolution deletion analysis of constitutional DNA from neurofibromatosis type 2 (NF2) patients using microarray-CGH. Human Molecular Genetics, 2001, 10, 271-282.	1.4	147
280	Oligomerization of polyalanine expanded PABPN1 facilitates nuclear protein aggregation that is associated with cell death. Human Molecular Genetics, 2001, 10, 2341-2351.	1.4	105
281	Association and linkage studies of CRH and PENK genes in bipolar disorder: A collaborative IGSLI study. , 2000, 96, 178-181.		26
282	Dopa-responsive dystonia due to a large deletion in the GTP cyclohydrolase I gene. Annals of Neurology, 2000, 47, 517-520.	2.8	63
283	PABP2 polyalanine tract expansion causes intranuclear inclusions in oculopharyngeal muscular dystrophy. Annals of Neurology, 2000, 48, 798-802.	2.8	40
284	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.	2.2	15
285	Mutations in GJB6 cause hidrotic ectodermal dysplasia. Nature Genetics, 2000, 26, 142-144.	9.4	270
286	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.	1.4	43
287	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.	1.4	31
288	Homozygotes for oculopharyngeal muscular dystrophy have a severe form of the disease. Annals of Neurology, 1999, 46, 115-118.	2.8	92

#	Article	IF	Citations
289	Lack of association between the hSKCa3 channel gene CAG polymorphism and schizophrenia. , 1999, 88, 154-157.		26
290	Analysis of 14 CAG repeat-containing genes in schizophrenia. American Journal of Medical Genetics Part A, 1999, 88, 694-699.	2.4	38
291	Family density of alcoholism and linkage information in the analysis of the COGA data. Genetic Epidemiology, 1999, 17, S361-S366.	0.6	7
292	Mutations in a gene encoding a novel protein tyrosine phosphatase cause progressive myoclonus epilepsy. Nature Genetics, 1998, 20, 171-174.	9.4	499
293	Short GCG expansions in the PABP2 gene cause oculopharyngeal muscular dystrophy. Nature Genetics, 1998, 18, 164-167.	9.4	751
294	Prenatal diagnosis of Machadoâ€“Joseph disease by direct mutation analysis. , 1998, 18, 611-617.		17
295	Reply to Bellivier et al.. American Journal of Medical Genetics Part A, 1998, 81, 351-352.	2.4	0
296	The Neuropathology of CAG Repeat Diseases: Review and Update of Genetic and Molecular Features. Brain Pathology, 1997, 7, 901-926.	2.1	134
297	Schizophrenia and chromosome 6p. American Journal of Medical Genetics Part A, 1997, 74, 195-198.	2.4	45
298	Lack of association between bipolar disorder and tyrosine hydroxylase. , 1997, 74, 348-352.		30
299	Modeling the phenotype in parametric linkage analysis of bipolar disorder. Genetic Epidemiology, 1997, 14, 687-691.	0.6	3
300	Schizophrenia and chromosome 6p. , 1997, 74, 195.		1
301	SOD1 mutation is assosiated with accumulation of neurofilaments in amyotrophic lateral scelaries. Annals of Neurology, 1996, 39, 128-131.	2.8	227
302	Somatic mosaicism in the central nervous system in spinocerebellar ataxia type 1 and machado-joseph disease. Annals of Neurology, 1996, 40, 199-206.	2.8	59
303	Moderate expansion of a normally biallelic trinucleotide repeat in spinocerebellar ataxia type 2. Nature Genetics, 1996, 14, 269-276.	9.4	1,092
304	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
305	Screening for germ-line mutations in theNF2 Gene. Genes Chromosomes and Cancer, 1995, 12, 117-127.	1.5	128
306	Gender equality in Machadoâ€“Joseph disease. Nature Genetics, 1995, 11, 118-119.	9.4	12

#	Article	IF	Citations
307	Evidence for the complete inactivation of the NF2 gene in the majority of sporadic meningiomas. Nature Genetics, 1994, 6, 180-184.	9.4	514
308	Variants of the heavy neurofilament subunit are associated with the development of amyotrophic lateral sclerosis. Human Molecular Genetics, 1994, 3, 1757-1761.	1.4	452
309	Newborn Apnea Caused by a Neurofibroma at the Craniocervical Junction. Canadian Journal of Neurological Sciences, 1994, 21, 64-66.	0.3	7
310	Alteration in a new gene encoding a putative membrane-organizing protein causes neuro-fibromatosis type 2. Nature, 1993, 363, 515-521.	13.7	1,351
311	Reduced transcriptional regulatory competence of the androgen receptor in Xâ€“linked spinal and bulbar muscular atrophy. Nature Genetics, 1993, 5, 184-188.	9.4	344
312	Mutations in Cu/Zn superoxide dismutase gene are associated with familial amyotrophic lateral sclerosis. Nature, 1993, 362, 59-62.	13.7	6,331
313	Loss of heterozygosity on the long arm of chromosome 22 in pheochromocytoma. Genes Chromosomes and Cancer, 1992, 5, 399-403.	1.5	43
314	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.	13.9	407
315	Molecular Genetics of Neurofibromatosis 2 and Related Tumors (Acoustic Neuroma and Meningioma). Annals of the New York Academy of Sciences, 1991, 615, 338-343.	1.8	36
316	Glucocorticoid excess induces preferential depletion of myosin in denervated skeletal muscle fibers. Muscle and Nerve, 1987, 10, 428-438.	1.0	118
317	Diagnosing Zygosity in Infant Twins: Physical Similarity, Genotyping, and Chorionicity. , 0, .		6
318	A polyalanine antibody for the diagnosis of oculopharyngeal muscular dystrophy and polyalanine-related diseases. MNI Open Research, 0, 1, 1.	1.0	0
319	RABENOSYN separation-of-function mutations uncouple endosomal recycling from lysosomal degradation, causing a distinct Mendelian Disorder. Human Molecular Genetics, 0, , .	1.4	0
320	Progress in the genetics of restless legs syndrome: the path ahead in the era of whole-genome sequencing. Sleep, 0, , .	0.6	0