

# Guy A Rouleau

## List of Publications by Year in descending order

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320  
papers

36,600  
citations

7551

77  
h-index

3815

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. <i>Nature</i> , 1993, 362, 59-62.	13.7	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. <i>Lancet Neurology</i> , The, 2019, 18, 1091-1102.	4.9	1,414
3	TARDBP mutations in individuals with sporadic and familial amyotrophic lateral sclerosis. <i>Nature Genetics</i> , 2008, 40, 572-574.	9.4	1,371
4	Alteration in a new gene encoding a putative membrane-organizing protein causes neuro-fibromatosis type 2. <i>Nature</i> , 1993, 363, 515-521.	13.7	1,351
5	Genome-wide association study identifies 30 loci associated with bipolar disorder. <i>Nature Genetics</i> , 2019, 51, 793-803.	9.4	1,191
6	Moderate expansion of a normally biallelic trinucleotide repeat in spinocerebellar ataxia type 2. <i>Nature Genetics</i> , 1996, 14, 269-276.	9.4	1,092
7	Analysis of shared heritability in common disorders of the brain. <i>Science</i> , 2018, 360, .	6.0	1,085
8	Genomic Relationships, Novel Loci, and Pleiotropic Mechanisms across Eight Psychiatric Disorders. <i>Cell</i> , 2019, 179, 1469-1482.e11.	13.5	935
9	Exome sequencing in amyotrophic lateral sclerosis identifies risk genes and pathways. <i>Science</i> , 2015, 347, 1436-1441.	6.0	823
10	Short GCG expansions in the PABP2 gene cause oculopharyngeal muscular dystrophy. <i>Nature Genetics</i> , 1998, 18, 164-167.	9.4	751
11	A gene encoding a putative GTPase regulator is mutated in familial amyotrophic lateral sclerosis 2. <i>Nature Genetics</i> , 2001, 29, 166-173.	9.4	635
12	Genome-wide association study of restless legs syndrome identifies common variants in three genomic regions. <i>Nature Genetics</i> , 2007, 39, 1000-1006.	9.4	633
13	Genome-wide association study of more than 40,000 bipolar disorder cases provides new insights into the underlying biology. <i>Nature Genetics</i> , 2021, 53, 817-829.	9.4	629
14	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. <i>Neuron</i> , 2018, 97, 1268-1283.e6.	3.8	517
15	Evidence for the complete inactivation of the NF2 gene in the majority of sporadic meningiomas. <i>Nature Genetics</i> , 1994, 6, 180-184.	9.4	514
16	Mutations in a gene encoding a novel protein tyrosine phosphatase cause progressive myoclonus epilepsy. <i>Nature Genetics</i> , 1998, 20, 171-174.	9.4	499
17	Variants of the heavy neurofilament subunit are associated with the development of amyotrophic lateral sclerosis. <i>Human Molecular Genetics</i> , 1994, 3, 1757-1761.	1.4	452
18	Linkage of a Gene Causing Familial Amyotrophic Lateral Sclerosis to Chromosome 21 and Evidence of Genetic-Locus Heterogeneity. <i>New England Journal of Medicine</i> , 1991, 324, 1381-1384.	13.9	407

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19	Increased exonic de novo mutation rate in individuals with schizophrenia. <i>Nature Genetics</i> , 2011, 43, 860-863.	9.4	392
20	Reduced transcriptional regulatory competence of the androgen receptor in X-linked spinal and bulbar muscular atrophy. <i>Nature Genetics</i> , 1993, 5, 184-188.	9.4	344
21	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. <i>American Journal of Human Genetics</i> , 2017, 101, 664-685.	2.6	337
22	Glucocerebrosidase activity in Parkinson's disease with and without GBA mutations. <i>Brain</i> , 2015, 138, 2648-2658.	3.7	326
23	Exome-wide Rare Variant Analysis Identifies TUBA4A Mutations Associated with Familial ALS. <i>Neuron</i> , 2014, 84, 324-331.	3.8	308
24	Genetic variants associated with response to lithium treatment in bipolar disorder: a genome-wide association study. <i>Lancet</i> , 2016, 387, 1085-1093.	6.3	306
25	Genome-wide association study reveals two new risk loci for bipolar disorder. <i>Nature Communications</i> , 2014, 5, 3339.	5.8	294
26	Mutations in GJB6 cause hidrotic ectodermal dysplasia. <i>Nature Genetics</i> , 2000, 26, 142-144.	9.4	270
27	Interrogating the Genetic Determinants of Tourette's Syndrome and Other Tic Disorders Through Genome-Wide Association Studies. <i>American Journal of Psychiatry</i> , 2019, 176, 217-227.	4.0	242
28	Partitioning the Heritability of Tourette Syndrome and Obsessive Compulsive Disorder Reveals Differences in Genetic Architecture. <i>PLoS Genetics</i> , 2013, 9, e1003864.	1.5	241
29	SOD1 mutation is associated with accumulation of neurofilaments in amyotrophic lateral sclerosis. <i>Annals of Neurology</i> , 1996, 39, 128-131.	2.8	227
30	Common and rare variant association analyses in amyotrophic lateral sclerosis identify 15 risk loci with distinct genetic architectures and neuron-specific biology. <i>Nature Genetics</i> , 2021, 53, 1636-1648.	9.4	223
31	Direct Measure of the De Novo Mutation Rate in Autism and Schizophrenia Cohorts. <i>American Journal of Human Genetics</i> , 2010, 87, 316-324.	2.6	222
32	NEK1 variants confer susceptibility to amyotrophic lateral sclerosis. <i>Nature Genetics</i> , 2016, 48, 1037-1042.	9.4	218
33	Genetic perspective on the role of the autophagy-lysosome pathway in Parkinson disease. <i>Autophagy</i> , 2015, 11, 1443-1457.	4.3	217
34	Genetics of familial and sporadic amyotrophic lateral sclerosis. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2006, 1762, 956-972.	1.8	214
35	Identification of novel risk loci for restless legs syndrome in genome-wide association studies in individuals of European ancestry: a meta-analysis. <i>Lancet Neurology</i> , 2017, 16, 898-907.	4.9	191
36	Genome-wide association study of 40,000 individuals identifies two novel loci associated with bipolar disorder. <i>Human Molecular Genetics</i> , 2016, 25, 3383-3394.	1.4	182

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37	The Impact of Phenotypic and Genetic Heterogeneity on Results of Genome Wide Association Studies of Complex Diseases. <i>PLoS ONE</i> , 2013, 8, e76295.	1.1	177
38	Exome Sequencing Identifies FUS Mutations as a Cause of Essential Tremor. <i>American Journal of Human Genetics</i> , 2012, 91, 313-319.	2.6	176
39	CCNF mutations in amyotrophic lateral sclerosis and frontotemporal dementia. <i>Nature Communications</i> , 2016, 7, 11253.	5.8	174
40	A Frameshift Deletion in Peripherin Gene Associated with Amyotrophic Lateral Sclerosis. <i>Journal of Biological Chemistry</i> , 2004, 279, 45951-45956.	1.6	163
41	Genome-Wide Association Study Identifies Novel Restless Legs Syndrome Susceptibility Loci on 2p14 and 16q12.1. <i>PLoS Genetics</i> , 2011, 7, e1002171.	1.5	163
42	Genetically encoded impairment of neuronal $\text{KCC}2$ cotransporter function in human idiopathic generalized epilepsy. <i>EMBO Reports</i> , 2014, 15, 766-774.	2.0	163
43	Genome-wide association study of intracranial aneurysms identifies 17 risk loci and genetic overlap with clinical risk factors. <i>Nature Genetics</i> , 2020, 52, 1303-1313.	9.4	163
44	Deletion of C9ORF72 Results in Motor Neuron Degeneration and Stress Sensitivity in <i>C. elegans</i> . <i>PLoS ONE</i> , 2013, 8, e83450.	1.1	158
45	Assessment of Response to Lithium Maintenance Treatment in Bipolar Disorder: A Consortium on Lithium Genetics (ConLiGen) Report. <i>PLoS ONE</i> , 2013, 8, e65636.	1.1	156
46	De Novo Coding Variants Are Strongly Associated with Tourette Disorder. <i>Neuron</i> , 2017, 94, 486-499.e9.	3.8	155
47	Clinical Spectrum of Amyotrophic Lateral Sclerosis (ALS). <i>Cold Spring Harbor Perspectives in Medicine</i> , 2017, 7, a024117.	2.9	149
48	High resolution deletion analysis of constitutional DNA from neurofibromatosis type 2 (NF2) patients using microarray-CGH. <i>Human Molecular Genetics</i> , 2001, 10, 271-282.	1.4	147
49	Dissection of genetic factors associated with amyotrophic lateral sclerosis. <i>Experimental Neurology</i> , 2014, 262, 91-101.	2.0	145
50	C9orf72 repeat expansions are a rare genetic cause of parkinsonism. <i>Brain</i> , 2013, 136, 385-391.	3.7	143
51	Rare Copy Number Variants in NRXN1 and CNTN6 Increase Risk for Tourette Syndrome. <i>Neuron</i> , 2017, 94, 1101-1111.e7.	3.8	137
52	The Neuropathology of CAG Repeat Diseases: Review and Update of Genetic and Molecular Features. <i>Brain Pathology</i> , 1997, 7, 901-926.	2.1	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. <i>Neuropsychobiology</i> , 2010, 62, 72-78.	0.9	134
54	Screening for germ-line mutations in the NF2 Gene. <i>Genes Chromosomes and Cancer</i> , 1995, 12, 117-127.	1.5	128

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55	ERBB4 Mutations that Disrupt the Neuregulin-ErbB4 Pathway Cause Amyotrophic Lateral Sclerosis Type 19. <i>American Journal of Human Genetics</i> , 2013, 93, 900-905.	2.6	123
56	Deleterious mutations in the essential mRNA metabolism factor, hGle1, in amyotrophic lateral sclerosis. <i>Human Molecular Genetics</i> , 2015, 24, 1363-1373.	1.4	122
57	Glucocorticoid excess induces preferential depletion of myosin in denervated skeletal muscle fibers. <i>Muscle and Nerve</i> , 1987, 10, 428-438.	1.0	118
58	A blinded international study on the reliability of genetic testing for GGGGCC-repeat expansions in <i>C9orf72</i> reveals marked differences in results among 14 laboratories. <i>Journal of Medical Genetics</i> , 2014, 51, 419-424.	1.5	118
59	GBA mutations are associated with Rapid Eye Movement Sleep Behavior Disorder. <i>Annals of Clinical and Translational Neurology</i> , 2015, 2, 941-945.	1.7	117
60	Dissecting the Shared Genetic Architecture of Suicide Attempt, Psychiatric Disorders, and Known Risk Factors. <i>Biological Psychiatry</i> , 2022, 91, 313-327.	0.7	114
61	A Pentanucleotide ATTTC Repeat Insertion in the Non-coding Region of DAB1, Mapping to SCA37, Causes Spinocerebellar Ataxia. <i>American Journal of Human Genetics</i> , 2017, 101, 87-103.	2.6	112
62	NLGN3/NLGN4 gene mutations are not responsible for autism in the Quebec population. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2005, 132B, 74-75.	1.1	111
63	Three Families With Amyotrophic Lateral Sclerosis and Frontotemporal Dementia With Evidence of Linkage to Chromosome 9p. <i>Archives of Neurology</i> , 2007, 64, 240.	4.9	111
64	Expanding the Clinical Phenotype Associated With <i>ELOVL4</i> Mutation. <i>JAMA Neurology</i> , 2014, 71, 470.	4.5	110
65	Genome-Wide Association Study of Intracranial Aneurysms Confirms Role of Anril and SOX17 in Disease Risk. <i>Stroke</i> , 2012, 43, 2846-2852.	1.0	106
66	Defining the genetic connection linking amyotrophic lateral sclerosis (ALS) with frontotemporal dementia (FTD). <i>Trends in Genetics</i> , 2015, 31, 263-273.	2.9	106
67	Oligomerization of polyalanine expanded PABPN1 facilitates nuclear protein aggregation that is associated with cell death. <i>Human Molecular Genetics</i> , 2001, 10, 2341-2351.	1.4	105
68	Recessive mutations in <i>VPS13D</i> cause childhood onset movement disorders. <i>Annals of Neurology</i> , 2018, 83, 1089-1095.	2.8	104
69	Polymorphism, shared functions and convergent evolution of genes with sequences coding for polyalanine domains. <i>Human Molecular Genetics</i> , 2003, 12, 2967-2979.	1.4	103
70	Association of Polygenic Score for Schizophrenia and HLA Antigen and Inflammation Genes With Response to Lithium in Bipolar Affective Disorder. <i>JAMA Psychiatry</i> , 2018, 75, 65-74.	6.0	102
71	An <i>ALS2</i> gene mutation causes hereditary spastic paraplegia in a Pakistani kindred. <i>Annals of Neurology</i> , 2003, 53, 144-145.	2.8	97
72	Mutations in <i>CAPN1</i> Cause Autosomal-Recessive Hereditary Spastic Paraplegia. <i>American Journal of Human Genetics</i> , 2016, 98, 1038-1046.	2.6	96

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73	Homozygotes for oculopharyngeal muscular dystrophy have a severe form of the disease. <i>Annals of Neurology</i> , 1999, 46, 115-118.	2.8	92
74	De Novo Sequence and Copy Number Variants Are Strongly Associated with Tourette Disorder and Implicate Cell Polarity in Pathogenesis. <i>Cell Reports</i> , 2018, 24, 3441-3454.e12.	2.9	91
75	Alteration of ornithine metabolism leads to dominant and recessive hereditary spastic paraplegia. <i>Brain</i> , 2015, 138, 2191-2205.	3.7	88
76	Exome sequencing reveals SPG11 mutations causing juvenile ALS. <i>Neurobiology of Aging</i> , 2012, 33, 839.e5-839.e9.	1.5	87
77	Regulatory domain or CpG site variation in SLC12A5, encoding the chloride transporter KCC2, in human autism and schizophrenia. <i>Frontiers in Cellular Neuroscience</i> , 2015, 9, 386.	1.8	86
78	MEIS1 intronic risk haplotype associated with restless legs syndrome affects its mRNA and protein expression levels. <i>Human Molecular Genetics</i> , 2009, 18, 1065-1074.	1.4	85
79	Loss of Association of REEP2 with Membranes Leads to Hereditary Spastic Paraplegia. <i>American Journal of Human Genetics</i> , 2014, 94, 268-277.	2.6	83
80	Clinical and genetic study of hereditary spastic paraplegia in Canada. <i>Neurology: Genetics</i> , 2017, 3, e122.	0.9	82
81	<i>SMPD1</i> mutations, activity, and $\alpha$ -synuclein accumulation in Parkinson's disease. <i>Movement Disorders</i> , 2019, 34, 526-535.	2.2	81
82	Compound heterozygous D90A and D96N SOD1 mutations in a recessive amyotrophic lateral sclerosis family. <i>Annals of Neurology</i> , 2001, 49, 267-271.	2.8	80
83	Genome-wide association study in essential tremor identifies three new loci. <i>Brain</i> , 2016, 139, 3163-3169.	3.7	78
84	A homozygous loss-of-function variant in MYH11 in a case with megacystis-microcolon-intestinal hypoperistalsis syndrome. <i>European Journal of Human Genetics</i> , 2015, 23, 1266-1268.	1.4	77
85	De novo variants in sporadic cases of childhood onset schizophrenia. <i>European Journal of Human Genetics</i> , 2016, 24, 944-948.	1.4	77
86	<i>GBA</i> p.T369M substitution in Parkinson disease: Polymorphism or association? A meta-analysis. <i>Neurology: Genetics</i> , 2016, 2, e104.	0.9	74
87	The 14q restless legs syndrome locus in the French Canadian population. <i>Annals of Neurology</i> , 2004, 55, 887-891.	2.8	71
88	Identification of rare protein disulfide isomerase gene variants in amyotrophic lateral sclerosis patients. <i>Gene</i> , 2015, 566, 158-165.	1.0	70
89	Mutations in DCC cause isolated agenesis of the corpus callosum with incomplete penetrance. <i>Nature Genetics</i> , 2017, 49, 511-514.	9.4	69
90	Hereditary motor and sensory neuropathy with agenesis of the corpus callosum. <i>Annals of Neurology</i> , 2003, 54, 9-18.	2.8	67

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91	Asian Origin for the Worldwide-Spread Mutational Event in Machado-Joseph Disease. Archives of Neurology, 2007, 64, 1502.	4.9	65
92	Genetic, Structural, and Functional Evidence Link <i>TMEM175</i> to Synucleinopathies. Annals of Neurology, 2020, 87, 139-153.	2.8	65
93	Dopa-responsive dystonia due to a large deletion in the GTP cyclohydrolase I gene. Annals of Neurology, 2000, 47, 517-520.	2.8	63
94	Family Study of Restless Legs Syndrome in Quebec, Canada. Archives of Neurology, 2010, 67, 617-22.	4.9	63
95	Quantitative Analysis of Climbing Defects in a Drosophila Model of Neurodegenerative Disorders. Journal of Visualized Experiments, 2015, , e52741.	0.2	60
96	Genetics of Intracranial Aneurysms. Stroke, 2018, 49, 780-787.	1.0	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.	2.8	59
98	<i>PMPCA</i> mutations cause abnormal mitochondrial protein processing in patients with non-progressive cerebellar ataxia. Brain, 2015, 138, 1505-1517.	3.7	58
99	Joubert Syndrome in French Canadians and Identification of Mutations in CEP104. American Journal of Human Genetics, 2015, 97, 744-753.	2.6	56
100	Transcriptome-wide association study of attention deficit hyperactivity disorder identifies associated genes and phenotypes. Nature Communications, 2019, 10, 4450.	5.8	56
101	ALS: Recent Developments from Genetics Studies. Current Neurology and Neuroscience Reports, 2016, 16, 59.	2.0	55
102	Ribosomal frameshifting on MJD-1 transcripts with long CAG tracts. Human Molecular Genetics, 2005, 14, 2649-2660.	1.4	54
103	Replication study of MATR3 in familial and sporadic amyotrophic lateral sclerosis. Neurobiology of Aging, 2016, 37, 209.e17-209.e21.	1.5	53
104	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
105	Genome-Wide Association Study of Intracranial Aneurysm Identifies a New Association on Chromosome 7. Stroke, 2014, 45, 3194-3199.	1.0	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.	1.1	51
107	Global characterization of copy number variants in epilepsy patients from whole genome sequencing. PLoS Genetics, 2018, 14, e1007285.	1.5	50
108	RNF213 Is Associated with Intracranial Aneurysms in the French-Canadian Population. American Journal of Human Genetics, 2016, 99, 1072-1085.	2.6	49

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109	Systematic review of autosomal recessive ataxias and proposal for a classification. <i>Cerebellum and Ataxias</i> , 2017, 4, 3.	1.9	49
110	Transgenic expression of an expanded (GCG) <sub>13</sub> repeat PABPN1 leads to weakness and coordination defects in mice. <i>Neurobiology of Disease</i> , 2005, 18, 528-536.	2.1	48
111	Mutation screening of FOXP2 in individuals diagnosed with autistic disorder. <i>American Journal of Medical Genetics Part A</i> , 2003, 118A, 172-175.	2.4	46
112	Molecular aspects of hereditary spastic paraplegia. <i>Experimental Cell Research</i> , 2014, 325, 18-26.	1.2	46
113	Preclinical target validation using patient-derived cells. <i>Nature Reviews Drug Discovery</i> , 2015, 14, 149-150.	21.5	46
114	SPG7 mutations explain a significant proportion of French Canadian spastic ataxia cases. <i>European Journal of Human Genetics</i> , 2016, 24, 1016-1021.	1.4	46
115	Retention of hexanucleotide repeat-containing intron in C9orf72 mRNA: implications for the pathogenesis of ALS/FTD. <i>Acta Neuropathologica Communications</i> , 2016, 4, 18.	2.4	46
116	Association of Variants in the <i>SPTLC1</i> Gene With Juvenile Amyotrophic Lateral Sclerosis. <i>JAMA Neurology</i> , 2021, 78, 1236.	4.5	46
117	Schizophrenia and chromosome 6p. <i>American Journal of Medical Genetics Part A</i> , 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). <i>Epilepsy Research</i> , 2003, 53, 107-117.	0.8	45
119	<i>GBA</i> variants in REM sleep behavior disorder. <i>Neurology</i> , 2020, 95, e1008-e1016.	1.5	45
120	Association of polygenic score for major depression with response to lithium in patients with bipolar disorder. <i>Molecular Psychiatry</i> , 2021, 26, 2457-2470.	4.1	44
121	Enrichment of SARM1 alleles encoding variants with constitutively hyperactive NADase in patients with ALS and other motor nerve disorders. <i>ELife</i> , 2021, 10, .	2.8	44
122	Loss of heterozygosity on the long arm of chromosome 22 in pheochromocytoma. <i>Genes Chromosomes and Cancer</i> , 1992, 5, 399-403.	1.5	43
123	Clouston hidrotic ectodermal dysplasia (HED): genetic homogeneity, presence of a founder effect in the French Canadian population and fine genetic mapping. <i>European Journal of Human Genetics</i> , 2000, 8, 372-380.	1.4	43
124	Inhibition of the kinase WNK1/HSN2 ameliorates neuropathic pain by restoring GABA inhibition. <i>Science Signaling</i> , 2016, 9, ra32.	1.6	43
125	<i>DCC</i> mutation update: Congenital mirror movements, isolated agenesis of the corpus callosum, and developmental split brain syndrome. <i>Human Mutation</i> , 2018, 39, 23-39.	1.1	41
126	PABP2 polyalanine tract expansion causes intranuclear inclusions in oculopharyngeal muscular dystrophy. <i>Annals of Neurology</i> , 2000, 48, 798-802.	2.8	40



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127	Expanded ATXN3 frameshifting events are toxic in Drosophila and mammalian neuron models. <i>Human Molecular Genetics</i> , 2012, 21, 2211-2218.	1.4	40
128	Gain-of-function missense variant in SLC12A2, encoding the bumetanide-sensitive NKCC1 cotransporter, identified in human schizophrenia. <i>Journal of Psychiatric Research</i> , 2016, 77, 22-26.	1.5	40
129	Genetics of restless legs syndrome. <i>Sleep Medicine</i> , 2017, 31, 18-22.	0.8	40
130	Characterization of human iPSC-derived astrocytes with potential for disease modeling and drug discovery. <i>Neuroscience Letters</i> , 2020, 731, 135028.	1.0	40
131	A mutation in the HSN2 gene causes sensory neuropathy type II in a Lebanese family. <i>Annals of Neurology</i> , 2004, 56, 572-575.	2.8	39
132	Truncating Mutations in UBAP1 Cause Hereditary Spastic Paraplegia. <i>American Journal of Human Genetics</i> , 2019, 104, 767-773.	2.6	39
133	Mechanisms Underlying the Hyperexcitability of CA3 and Dentate Gyrus Hippocampal Neurons Derived From Patients With Bipolar Disorder. <i>Biological Psychiatry</i> , 2020, 88, 139-149.	0.7	39
134	Evolution of a Human-Specific Tandem Repeat Associated with ALS. <i>American Journal of Human Genetics</i> , 2020, 107, 445-460.	2.6	39
135	Fine Mapping of SNCA in Rapid Eye Movement Sleep Behavior Disorder and Overt Synucleinopathies. <i>Annals of Neurology</i> , 2020, 87, 584-598.	2.8	39
136	Analysis of Heterozygous PRKN Variants and Copy Number Variations in Parkinson's Disease. <i>Movement Disorders</i> , 2021, 36, 178-187.	2.2	39
137	Analysis of 14 CAG repeat-containing genes in schizophrenia. <i>American Journal of Medical Genetics Part A</i> , 1999, 88, 694-699.	2.4	38
138	PABPN1 polyalanine tract deletion and long expansions modify its aggregation pattern and expression. <i>Experimental Cell Research</i> , 2008, 314, 1652-1666.	1.2	38
139	Analysis of DNAJC13 mutations in French-Canadian/French cohort of Parkinson's disease. <i>Neurobiology of Aging</i> , 2016, 45, 212.e13-212.e17.	1.5	38
140	Mutations in ATP13A2 (PARK9) are associated with an amyotrophic lateral sclerosis-like phenotype, implicating this locus in further phenotypic expansion. <i>Human Genomics</i> , 2019, 13, 19.	1.4	38
141	Genome-wide study of DNA methylation shows alterations in metabolic, inflammatory, and cholesterol pathways in ALS. <i>Science Translational Medicine</i> , 2022, 14, eabj0264.	5.8	38
142	Genetic and Clinical Predictors of Deep Brain Stimulation in Young-Onset Parkinson's Disease. <i>Movement Disorders Clinical Practice</i> , 2016, 3, 465-471.	0.8	37
143	CPT1A Missense Mutation Associated With Fatty Acid Metabolism and Reduced Height in Greenlanders. <i>Circulation: Cardiovascular Genetics</i> , 2017, 10, .	5.1	37
144	Molecular Genetics of Neurofibromatosis 2 and Related Tumors (Acoustic Neuroma and Meningioma). <i>Annals of the New York Academy of Sciences</i> , 1991, 615, 338-343.	1.8	36

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145	A de novo frameshift mutation in chromodomain helicase DNA-binding domain 8 (CHD8): A case report and literature review. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 1225-1235.	0.7	36
146	SCARB2 variants and glucocerebrosidase activity in Parkinson's disease. <i>Npj Parkinson's Disease</i> , 2016, 2, .	2.5	36
147	Sleep disorders and Parkinson disease; lessons from genetics. <i>Sleep Medicine Reviews</i> , 2018, 41, 101-112.	3.8	35
148	The Quebec Parkinson Network: A Researcher-Patient Matching Platform and Multimodal Biorepository. <i>Journal of Parkinson's Disease</i> , 2020, 10, 301-313.	1.5	35
149	A New <i>ELOVL4</i> Mutation in a Case of Spinocerebellar Ataxia With Erythrokeratodermia. <i>JAMA Neurology</i> , 2015, 72, 942.	4.5	34
150	Modifiers of (CAG) <sub>n</sub> instability in Machado-Joseph disease (MJD/SCA3) transmissions: an association study with DNA replication, repair and recombination genes. <i>Human Genetics</i> , 2014, 133, 1311-1318.	1.8	33
151	The emerging role of SMPD1 mutations in Parkinson's disease: Implications for future studies. <i>Parkinsonism and Related Disorders</i> , 2015, 21, 1294-1295.	1.1	33
152	Toward Precision Medicine: <i>TBC1D4</i> Disruption Is Common Among the Inuit and Leads to Underdiagnosis of Type 2 Diabetes. <i>Diabetes Care</i> , 2016, 39, 1889-1895.	4.3	33
153	Life-threatening viral disease in a novel form of autosomal recessive <i>IFNAR2</i> deficiency in the Arctic. <i>Journal of Experimental Medicine</i> , 2022, 219, .	4.2	33
154	Missense variants in ATP1A3 and FXD gene family are associated with childhood-onset schizophrenia. <i>Molecular Psychiatry</i> , 2020, 25, 821-830.	4.1	32
155	Study of three intragenic polymorphisms in the Machado-Joseph disease gene (MJD1) in relation to genetic instability of the (CAG) <sub>n</sub> tract. <i>European Journal of Human Genetics</i> , 1999, 7, 147-156.	1.4	31
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