Guy A Rouleau

List of Publications by Citations

321 papers	26,702 citations	69 h-index	160 g-index
408 ext. papers	32,277 ext. citations	9.3 avg, IF	6 L-index

#	Paper	IF	Citations
321	Mutations in Cu/Zn superoxide dismutase gene are associated with familial amyotrophic lateral sclerosis. *Nature*, **1993**, 362, 59-62	50.4	5355
320	Alteration in a new gene encoding a putative membrane-organizing protein causes neuro-fibromatosis type 2. *Nature*, **1993**, 363, 515-21	50.4	1205
319	TARDBP mutations in individuals with sporadic and familial amyotrophic lateral sclerosis. *Nature Genetics*, **2008**, 40, 572-4	36.3	1171
318	Moderate expansion of a normally biallelic trinucleotide repeat in spinocerebellar ataxia type 2. *Nature Genetics*, **1996**, 14, 269-76	36.3	963
317	Analysis of shared heritability in common disorders of the brain. *Science*, **2018**, 360,	33.3	666
316	Genome-wide association study identifies 30 loci associated with bipolar disorder. *Nature Genetics*, **2019**, 51, 793-803	36.3	662
315	Short GCG expansions in the PABP2 gene cause oculopharyngeal muscular dystrophy. *Nature Genetics*, **1998**, 18, 164-7	36.3	656
314	Exome sequencing in amyotrophic lateral sclerosis identifies risk genes and pathways. *Science*, **2015**, 347, 1436-41	33.3	642
313	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	24.1	562
312	A gene encoding a putative GTPase regulator is mutated in familial amyotrophic lateral sclerosis 2. *Nature Genetics*, **2001**, 29, 166-73	36.3	552
311	Genome-wide association study of restless legs syndrome identifies common variants in three genomic regions. *Nature Genetics*, **2007**, 39, 1000-6	36.3	545
310	Evidence for the complete inactivation of the NF2 gene in the majority of sporadic meningiomas. *Nature Genetics*, **1994**, 6, 180-4	36.3	450
309	Mutations in a gene encoding a novel protein tyrosine phosphatase cause progressive myoclonus epilepsy. *Nature Genetics*, **1998**, 20, 171-4	36.3	440
308	Genomic Relationships, Novel Loci, and Pleiotropic Mechanisms across Eight Psychiatric Disorders. *Cell*, **2019**, 179, 1469-1482.e11	56.2	402
307	Variants of the heavy neurofilament subunit are associated with the development of amyotrophic lateral sclerosis. *Human Molecular Genetics*, **1994**, 3, 1757-61	5.6	398
306	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, 1381-4	59.2	355
305	Increased exonic de novo mutation rate in individuals with schizophrenia. *Nature Genetics*, **2011**, 43, 860-3	36.3	333

304	Reduced transcriptional regulatory competence of the androgen receptor in X-linked spinal and bulbar muscular atrophy. *Nature Genetics*, **1993**, 5, 184-8	36.3	322
303	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. *Neuron*, **2018**, 97, 1268-1283.e6	13.9	296
302	Genome-wide association study reveals two new risk loci for bipolar disorder. *Nature Communications*, **2014**, 5, 3339	17.4	248
301	Glucocerebrosidase activity in Parkinson's disease with and without GBA mutations. *Brain*, **2015**, 138, 2648-58	11.2	234
300	Exome-wide rare variant analysis identifies TUBA4A mutations associated with familial ALS. *Neuron*, **2014**, 84, 324-31	13.9	229
299	Mutations in GJB6 cause hidrotic ectodermal dysplasia. *Nature Genetics*, **2000**, 26, 142-4	36.3	227
298	Genetic variants associated with response to lithium treatment in bipolar disorder: a genome-wide association study. *Lancet, The*, **2016**, 387, 1085-1093	40	216
297	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. *American Journal of Human Genetics*, **2017**, 101, 664-685	11	214
296	SOD1 mutation is associated with accumulation of neurofilaments in amyotrophic lateral sclerosis. *Annals of Neurology*, **1996**, 39, 128-31	9.4	210
295	Partitioning the heritability of Tourette syndrome and obsessive compulsive disorder reveals differences in genetic architecture. *PLoS Genetics*, **2013**, 9, e1003864	6	189
294	Direct measure of the de novo mutation rate in autism and schizophrenia cohorts. *American Journal of Human Genetics*, **2010**, 87, 316-24	11	181
293	Genetics of familial and sporadic amyotrophic lateral sclerosis. *Biochimica Et Biophysica Acta - Molecular Basis of Disease*, **2006**, 1762, 956-72	6.9	179
292	Genetic perspective on the role of the autophagy-lysosome pathway in Parkinson disease. *Autophagy*, **2015**, 11, 1443-57	10.2	168
291	NEK1 variants confer susceptibility to amyotrophic lateral sclerosis. *Nature Genetics*, **2016**, 48, 1037-42	36.3	149
290	Exome sequencing identifies FUS mutations as a cause of essential tremor. *American Journal of Human Genetics*, **2012**, 91, 313-9	11	147
289	A frameshift deletion in peripherin gene associated with amyotrophic lateral sclerosis. *Journal of Biological Chemistry*, **2004**, 279, 45951-6	5.4	137
288	Deletion of C9ORF72 results in motor neuron degeneration and stress sensitivity in C. elegans. *PLoS ONE*, **2013**, 8, e83450	3.7	135
287	Genome-wide association study identifies novel restless legs syndrome susceptibility loci on 2p14 and 16q12.1. *PLoS Genetics*, **2011**, 7, e1002171	6	135

286	C9orf72 repeat expansions are a rare genetic cause of parkinsonism. *Brain*, **2013**, 136, 385-91	11.2	128
285	CCNF mutations in amyotrophic lateral sclerosis and frontotemporal dementia. *Nature Communications*, **2016**, 7, 11253	17.4	126
284	Genome-wide association study of 40,000 individuals identifies two novel loci associated with bipolar disorder. *Human Molecular Genetics*, **2016**, 25, 3383-3394	5.6	125
283	The neuropathology of CAG repeat diseases: review and update of genetic and molecular features. *Brain Pathology*, **1997**, 7, 901-26	6	124
282	Genetically encoded impairment of neuronal KCC2 cotransporter function in human idiopathic generalized epilepsy. *EMBO Reports*, **2014**, 15, 766-74	6.5	123
281	The impact of phenotypic and genetic heterogeneity on results of genome wide association studies of complex diseases. *PLoS ONE*, **2013**, 8, e76295	3.7	122
280	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	24.1	121
279	Dissection of genetic factors associated with amyotrophic lateral sclerosis. *Experimental Neurology*, **2014**, 262 Pt B, 91-101	5.7	116
278	High resolution deletion analysis of constitutional DNA from neurofibromatosis type 2 (NF2) patients using microarray-CGH. *Human Molecular Genetics*, **2001**, 10, 271-82	5.6	115
277	Assessment of Response to Lithium Maintenance Treatment in Bipolar Disorder: A Consortium on Lithium Genetics (ConLiGen) Report. *PLoS ONE*, **2013**, 8, e65636	3.7	113
276	Screening for germ-line mutations in the NF2 gene. *Genes Chromosomes and Cancer*, **1995**, 12, 117-27	5	110
275	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, 72-8	4	109
274	Glucocorticoid excess induces preferential depletion of myosin in denervated skeletal muscle fibers. *Muscle and Nerve*, **1987**, 10, 428-38	3.4	106
273	Rare Copy Number Variants in NRXN1 and CNTN6 Increase Risk for Tourette Syndrome. *Neuron*, **2017**, 94, 1101-1111.e7	13.9	103
272	Three families with amyotrophic lateral sclerosis and frontotemporal dementia with evidence of linkage to chromosome 9p. *Archives of Neurology*, **2007**, 64, 240-5		101
271	Deleterious mutations in the essential mRNA metabolism factor, hGle1, in amyotrophic lateral sclerosis. *Human Molecular Genetics*, **2015**, 24, 1363-73	5.6	98
270	A blinded international study on the reliability of genetic testing for GGGGCC-repeat expansions in C9orf72 reveals marked differences in results among 14 laboratories. *Journal of Medical Genetics*, **2014**, 51, 419-24	5.8	96
269	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	11.9	95

268	ERBB4 mutations that disrupt the neuregulin-ErbB4 pathway cause amyotrophic lateral sclerosis type 19. *American Journal of Human Genetics*, **2013**, 93, 900-5	11	95
267	GBA mutations are associated with Rapid Eye Movement Sleep Behavior Disorder. *Annals of Clinical and Translational Neurology*, **2015**, 2, 941-5	5.3	95
266	Polymorphism, shared functions and convergent evolution of genes with sequences coding for polyalanine domains. *Human Molecular Genetics*, **2003**, 12, 2967-79	5.6	94
265	NLGN3/NLGN4 gene mutations are not responsible for autism in the Quebec population. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2005**, 132B, 74-5	3.5	94
264	Genome-wide association study of intracranial aneurysms confirms role of Anril and SOX17 in disease risk. *Stroke*, **2012**, 43, 2846-52	6.7	90
263	Oligomerization of polyalanine expanded PABPN1 facilitates nuclear protein aggregation that is associated with cell death. *Human Molecular Genetics*, **2001**, 10, 2341-51	5.6	90
262	De Novo Coding Variants Are Strongly Associated with Tourette Disorder. *Neuron*, **2017**, 94, 486-499.e9	13.9	89
261	Defining the genetic connection linking amyotrophic lateral sclerosis (ALS) with frontotemporal dementia (FTD). *Trends in Genetics*, **2015**, 31, 263-73	8.5	88
260	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	36.3	83
259	An ALS2 gene mutation causes hereditary spastic paraplegia in a Pakistani kindred. *Annals of Neurology*, **2003**, 53, 144-5	9.4	81
258	Expanding the clinical phenotype associated with ELOVL4 mutation: study of a large French-Canadian family with autosomal dominant spinocerebellar ataxia and erythrokeratodermia. *JAMA Neurology*, **2014**, 71, 470-5	17.2	78
257	Homozygotes for oculopharyngeal muscular dystrophy have a severe form of the disease. *Annals of Neurology*, **1999**, 46, 115-118	9.4	78
256	Association of Polygenic Score for Schizophrenia and HLA Antigen and Inflammation Genes With Response to Lithium in Bipolar Affective Disorder: A Genome-Wide Association Study. *JAMA Psychiatry*, **2018**, 75, 65-74	14.5	75
255	Exome sequencing reveals SPG11 mutations causing juvenile ALS. *Neurobiology of Aging*, **2012**, 33, 839.e5-9	5.0	73
254	MEIS1 intronic risk haplotype associated with restless legs syndrome affects its mRNA and protein expression levels. *Human Molecular Genetics*, **2009**, 18, 1065-74	5.6	73
253	Mutations in CAPN1 Cause Autosomal-Recessive Hereditary Spastic Paraplegia. *American Journal of Human Genetics*, **2016**, 98, 1038-1046	11	70
252	Compound heterozygous D90A and D96N SOD1 mutations in a recessive amyotrophic lateral sclerosis family. *Annals of Neurology*, **2001**, 49, 267-71	9.4	67
251	Clinical Spectrum of Amyotrophic Lateral Sclerosis (ALS). *Cold Spring Harbor Perspectives in Medicine* , **2017**, 7,	5.4	66

250	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	11	66
249	Alteration of ornithine metabolism leads to dominant and recessive hereditary spastic paraplegia. *Brain*, **2015**, 138, 2191-205	11.2	64
248	Loss of association of REEP2 with membranes leads to hereditary spastic paraplegia. *American Journal of Human Genetics*, **2014**, 94, 268-77	11	63
247	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	6.1	62
246	Recessive mutations in VPS13D cause childhood onset movement disorders. *Annals of Neurology*, **2018**, 83, 1089-1095	9.4	61
245	Hereditary motor and sensory neuropathy with agenesis of the corpus callosum. *Annals of Neurology*, **2003**, 54, 9-18	9.4	61
244	The 14q restless legs syndrome locus in the French Canadian population. *Annals of Neurology*, **2004**, 55, 887-91	9.4	60
243	Identification of rare protein disulfide isomerase gene variants in amyotrophic lateral sclerosis patients. *Gene*, **2015**, 566, 158-65	3.8	59
242	Genome-wide association study in essential tremor identifies three new loci. *Brain*, **2016**, 139, 3163-3169	11.2	57
241	De novo variants in sporadic cases of childhood onset schizophrenia. *European Journal of Human Genetics*, **2016**, 24, 944-8	5.3	56
240	A homozygous loss-of-function variant in MYH11 in a case with megacystis-microcolon-intestinal hypoperistalsis syndrome. *European Journal of Human Genetics*, **2015**, 23, 1266-8	5.3	55
239	Dopa-responsive dystonia due to a large deletion in the GTP cyclohydrolase I gene. *Annals of Neurology*, **2000**, 47, 517-520	9.4	55
238	Somatic mosaicism in the central nervous system in spinocerebellar ataxia type 1 and Machado-Joseph disease. *Annals of Neurology*, **1996**, 40, 199-206	9.4	55
237	Mutations in DCC cause isolated agenesis of the corpus callosum with incomplete penetrance. *Nature Genetics*, **2017**, 49, 511-514	36.3	54
236	Clinical and genetic study of hereditary spastic paraplegia in Canada. *Neurology: Genetics*, **2017**, 3, e122	3.8	54
235	GBA p.T369M substitution in Parkinson disease: Polymorphism or association? A meta-analysis. *Neurology: Genetics*, **2016**, 2, e104	3.8	51
234	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	10.6	51
233	Family study of restless legs syndrome in Quebec, Canada: clinical characterization of 671 familial cases. *Archives of Neurology*, **2010**, 67, 617-22		50

232	Asian origin for the worldwide-spread mutational event in Machado-Joseph disease. *Archives of Neurology*, **2007**, 64, 1502-8		50
231	Ribosomal frameshifting on MJD-1 transcripts with long CAG tracts. *Human Molecular Genetics*, **2005**, 14, 2649-60	5.6	49
230	A novel locus for pure recessive hereditary spastic paraplegia maps to 10q22.1-10q24.1. *Annals of Neurology*, **2004**, 56, 579-82	9.4	47
229	ALS: Recent Developments from Genetics Studies. *Current Neurology and Neuroscience Reports*, **2016**, 16, 59	6.6	45
228	PMPCA mutations cause abnormal mitochondrial protein processing in patients with non-progressive cerebellar ataxia. *Brain*, **2015**, 138, 1505-17	11.2	43
227	Genome-wide association study of intracranial aneurysms identifies 17 risk loci and genetic overlap with clinical risk factors. *Nature Genetics*, **2020**, 52, 1303-1313	36.3	43
226	Systematic review of autosomal recessive ataxias and proposal for a classification. *Cerebellum and Ataxias*, **2017**, 4, 3	1.7	42
225	Mutation screening of FOXP2 in individuals diagnosed with autistic disorder. *American Journal of Medical Genetics Part A*, **2003**, 118A, 172-5		42
224	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, 372-80	5.3	42
223	SMPD1 mutations, activity, and -synuclein accumulation in Parkinson's disease. *Movement Disorders*, **2019**, 34, 526-535	7	41
222	Joubert Syndrome in French Canadians and Identification of Mutations in CEP104. *American Journal of Human Genetics*, **2015**, 97, 744-53	11	41
221	Replication study of MATR3 in familial and sporadic amyotrophic lateral sclerosis. *Neurobiology of Aging*, **2016**, 37, 209.e17-209.e21	5.6	41
220	Genome-wide association study of intracranial aneurysm identifies a new association on chromosome 7. *Stroke*, **2014**, 45, 3194-9	6.7	41
219	Transgenic expression of an expanded (GCG)13 repeat PABPN1 leads to weakness and coordination defects in mice. *Neurobiology of Disease*, **2005**, 18, 528-36	7.5	41
218	Preclinical target validation using patient-derived cells. *Nature Reviews Drug Discovery*, **2015**, 14, 149-50	64.1	40
217	Schizophrenia and chromosome 6p. *American Journal of Medical Genetics Part A*, **1997**, 74, 195-198		40
216	Functional characterization of the D188V mutation in neuronal voltage-gated sodium channel causing generalized epilepsy with febrile seizures plus (GEFS). *Epilepsy Research*, **2003**, 53, 107-17	3	40
215	Genetic, Structural, and Functional Evidence Link TMEM175 to Synucleinopathies. *Annals of Neurology*, **2020**, 87, 139-153	9.4	40

214	RNF213 Is Associated with Intracranial Aneurysms in the French-Canadian Population. *American Journal of Human Genetics*, **2016**, 99, 1072-1085	11	38
213	SPG7 mutations explain a significant proportion of French Canadian spastic ataxia cases. *European Journal of Human Genetics*, **2016**, 24, 1016-21	5.3	38
212	PABP2 polyalanine tract expansion causes intranuclear inclusions in oculopharyngeal muscular dystrophy. *Annals of Neurology*, **2000**, 48, 798-802	9.4	38
211	Loss of heterozygosity on the long arm of chromosome 22 in pheochromocytoma. *Genes Chromosomes and Cancer*, **1992**, 5, 399-403	5	37
210	Molecular aspects of hereditary spastic paraplegia. *Experimental Cell Research*, **2014**, 325, 18-26	4.2	36
209	Expanded ATXN3 frameshifting events are toxic in Drosophila and mammalian neuron models. *Human Molecular Genetics*, **2012**, 21, 2211-8	5.6	36
208	Analysis of 14 CAG repeat-containing genes in schizophrenia. *American Journal of Medical Genetics Part A*, **1999**, 88, 694-699		36
207	Retention of hexanucleotide repeat-containing intron in C9orf72 mRNA: implications for the pathogenesis of ALS/FTD. *Acta Neuropathologica Communications*, **2016**, 4, 18	7.3	34
206	Genetics of Intracranial Aneurysms. *Stroke*, **2018**, 49, 780-787	6.7	33
205	Molecular genetics of neurofibromatosis 2 and related tumors (acoustic neuroma and meningioma). *Annals of the New York Academy of Sciences*, **1991**, 615, 338-43	6.5	33
204	A mutation in the HSN2 gene causes sensory neuropathy type II in a Lebanese family. *Annals of Neurology*, **2004**, 56, 572-5	9.4	32
203	Analysis of DNAJC13 mutations in French-Canadian/French cohort of Parkinson's disease. *Neurobiology of Aging*, **2016**, 45, 212.e13-212.e17	5.6	31
202	Gain-of-function missense variant in SLC12A2, encoding the bumetanide-sensitive NKCC1 cotransporter, identified in human schizophrenia. *Journal of Psychiatric Research*, **2016**, 77, 22-6	5.2	31
201	Quantitative Analysis of Climbing Defects in a Drosophila Model of Neurodegenerative Disorders. *Journal of Visualized Experiments*, **2015**, e52741	1.6	31
200	Inhibition of the kinase WNK1/HSN2 ameliorates neuropathic pain by restoring GABA inhibition. *Science Signaling*, **2016**, 9, ra32	8.8	30
199	Modifiers of (CAG)(n) instability in Machado-Joseph disease (MJD/SCA3) transmissions: an association study with DNA replication, repair and recombination genes. *Human Genetics*, **2014**, 133, 1311-8	6.3	29
198	PABPN1 polyalanine tract deletion and long expansions modify its aggregation pattern and expression. *Experimental Cell Research*, **2008**, 314, 1652-66	4.2	29
197	A de novo frameshift mutation in chromodomain helicase DNA-binding domain 8 (CHD8): A case report and literature review. *American Journal of Medical Genetics, Part A*, **2016**, 170A, 1225-35	2.5	29

196	Investigation of the Repeat Expansion in a Canadian and a Brazilian Ataxia Cohort: Identification of Novel Conformations. *Frontiers in Genetics*, **2019**, 10, 1219	4.5	29
195	Global characterization of copy number variants in epilepsy patients from whole genome sequencing. *PLoS Genetics*, **2018**, 14, e1007285	6	29
194	A New ELOVL4 Mutation in a Case of Spinocerebellar Ataxia With Erythrokeratodermia. *JAMA Neurology*, **2015**, 72, 942-3	17.2	27
193	Identification of novel FUS mutations in sporadic cases of amyotrophic lateral sclerosis. *Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders*, **2011**, 12, 113-7		27
192	Lack of association between bipolar disorder and tyrosine hydroxylase: a meta-analysis. *American Journal of Medical Genetics Part A*, **1997**, 74, 348-52		27
191	The emerging role of SMPD1 mutations in Parkinson's disease: Implications for future studies. *Parkinsonism and Related Disorders*, **2015**, 21, 1294-5	3.6	26
190	Identification of three polymorphisms in the translated region of PLC-gamma1 and their investigation in lithium responsive bipolar disorder. *American Journal of Medical Genetics Part A*, **2001**, 105, 301-5		26
189	DCC mutation update: Congenital mirror movements, isolated agenesis of the corpus callosum, and developmental split brain syndrome. *Human Mutation*, **2018**, 39, 23-39	4.7	26
188	Genetics of restless legs syndrome. *Sleep Medicine*, **2017**, 31, 18-22	4.6	25
187	Transcriptome-wide association study of attention deficit hyperactivity disorder identifies associated genes and phenotypes. *Nature Communications*, **2019**, 10, 4450	17.4	25
186	Fine-Mapping of SNCA in Rapid Eye Movement Sleep Behavior Disorder and Overt Synucleinopathies. *Annals of Neurology*, **2020**, 87, 584-598	9.4	24
185	Sleep disorders and Parkinson disease; lessons from genetics. *Sleep Medicine Reviews*, **2018**, 41, 101-112	10.2	24
184	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, 147-56	5.3	24
183	De novo FUS P525L mutation in Juvenile amyotrophic lateral sclerosis with dysphonia and diplopia. *Neurology: Genetics*, **2016**, 2, e63	3.8	24
182	Genetics of essential tremor: from phenotype to genes, insights from both human and mouse studies. *Progress in Neurobiology*, **2014**, 119-120, 1-19	10.9	23
181	Association and linkage studies of CRH and PENK genes in bipolar disorder: a collaborative IGSLI study. *American Journal of Medical Genetics Part A*, **2000**, 96, 178-81		23
180	Lack of association between the hSKCa3 channel gene CAG polymorphism and schizophrenia **1999**, 88, 154-157		23
179	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	6.8	22

178	LRRK2 mutations in Parkinson disease; a sex effect or lack thereof? A meta-analysis. *Parkinsonism and Related Disorders*, **2015**, 21, 778-82	3.6	22
177	Toward Precision Medicine: TBC1D4 Disruption Is Common Among the Inuit and Leads to Underdiagnosis of Type 2 Diabetes. *Diabetes Care*, **2016**, 39, 1889-1895	14.6	22
176	variants and glucocerebrosidase activity in Parkinson's disease. *Npj Parkinson's Disease*, **2016**, 2,	9.7	22
175	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, 13948-57	5.4	22
174	Mutational analysis of neurotensin in familial restless legs syndrome. *Movement Disorders*, **2004**, 19, 90-4	7	22
173	LINGO1 variants in the French-Canadian population. *PLoS ONE*, **2011**, 6, e16254	3.7	22
172	Genetic and Clinical Predictors of Deep Brain Stimulation in Young-Onset Parkinson's Disease. *Movement Disorders Clinical Practice*, **2016**, 3, 465-471	2.2	21
171	and Restless Legs Syndrome: A Comprehensive Review. *Frontiers in Neurology*, **2019**, 10, 935	4.1	20
170	Truncating Mutations in UBAP1 Cause Hereditary Spastic Paraplegia. *American Journal of Human Genetics*, **2019**, 104, 767-773	11	20
169	Characterization of human iPSC-derived astrocytes with potential for disease modeling and drug discovery. *Neuroscience Letters*, **2020**, 731, 135028	3.3	20
168	The dementia-associated APOE ε4 allele is not associated with rapid eye movement sleep behavior disorder. *Neurobiology of Aging*, **2017**, 49, 218.e13-218.e15	5.6	20
167	Dopamine transporter SLC6A3 genotype affects cortico-striatal activity of set-shifts in Parkinson's disease. *Brain*, **2014**, 137, 3025-35	11.2	20
166	Autosomal dominant primary lateral sclerosis. *Neurology*, **2007**, 68, 1156-7	6.5	20
165	Exome sequencing identifies recessive CDK5RAP2 variants in patients with isolated agenesis of corpus callosum. *European Journal of Human Genetics*, **2016**, 24, 607-10	5.3	19
164	Missense Mutation Associated With Fatty Acid Metabolism and Reduced Height in Greenlanders. *Circulation: Cardiovascular Genetics*, **2017**, 10,		19
163	The Tanenbaum Open Science Institute: Leading a Paradigm Shift at the Montreal Neurological Institute. *Neuron*, **2017**, 95, 1002-1006	13.9	19
162	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	36.3	19
161	Full sequencing and haplotype analysis of MAPT in Parkinson's disease and rapid eye movement sleep behavior disorder. *Movement Disorders*, **2018**, 33, 1016-1020	7	19

160	variants in REM sleep behavior disorder: A multicenter study. *Neurology*, **2020**, 95, e1008-e1016	6.5	18
159	Adenylosuccinate lyase (ADSL) and infantile autism: absence of previously reported point mutation. *American Journal of Medical Genetics Part A*, **1995**, 60, 554-7		18
158	De Novo Pathogenic Variants in N-cadherin Cause a Syndromic Neurodevelopmental Disorder with Corpus Collosum, Axon, Cardiac, Ocular, and Genital Defects. *American Journal of Human Genetics*, **2019**, 105, 854-868	11	17
157	Association of polygenic score for major depression with response to lithium in patients with bipolar disorder. *Molecular Psychiatry*, **2021**, 26, 2457-2470	15.1	17
156	The Quebec Parkinson Network: A Researcher-Patient Matching Platform and Multimodal Biorepository. *Journal of Parkinson's Disease*, **2020**, 10, 301-313	5.3	17
155	Genetic markers of Restless Legs Syndrome in Parkinson disease. *Parkinsonism and Related Disorders*, **2015**, 21, 582-5	3.6	16
154	LRRK2 protective haplotype and full sequencing study in REM sleep behavior disorder. *Parkinsonism and Related Disorders*, **2018**, 52, 98-101	3.6	16
153	Conserved pharmacological rescue of hereditary spastic paraplegia-related phenotypes across model organisms. *Human Molecular Genetics*, **2016**, 25, 1088-99	5.6	16
152	A direct interaction between two Restless Legs Syndrome predisposing genes: MEIS1 and SKOR1. *Scientific Reports*, **2018**, 8, 12173	4.9	16
151	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	3.6	16
150	Analysis of the Influence of microRNAs in Lithium Response in Bipolar Disorder. *Frontiers in Psychiatry*, **2018**, 9, 207	5	15
149	Mutation burden of rare variants in schizophrenia candidate genes. *PLoS ONE*, **2015**, 10, e0128988	3.7	15
148	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	7	15
147	Evolution of a Human-Specific Tandem Repeat Associated with ALS. *American Journal of Human Genetics*, **2020**, 107, 445-460	11	15
146	Mechanisms Underlying the Hyperexcitability of CA3 and Dentate Gyrus Hippocampal Neurons Derived From Patients With Bipolar Disorder. *Biological Psychiatry*, **2020**, 88, 139-149	7.9	15
145	A new approach for rare variation collapsing on functional protein domains implicates specific genic regions in ALS. *Genome Research*, **2019**, 29, 809-818	9.7	14
144	Analysis of functional GLO1 variants in the BTBD9 locus and restless legs syndrome. *Sleep Medicine*, **2015**, 16, 1151-5	4.6	14
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