Guy A Rouleau

List of Publications by Year in Descending Order

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	Moyamoya Disease Susceptibility Gene Regulates Endothelial Barrier Function.. *Stroke*, **2022**, STROKEAHA120032691	6.7	3
320	Association of Essential Tremor With Novel Risk Loci: A Genome-Wide Association Study and Meta-analysis.. *JAMA Neurology*, **2022**,	17.2	3
319	Using polygenic scores and clinical data for bipolar disorder patient stratification and lithium response prediction: machine learning approach.. *British Journal of Psychiatry*, **2022**, 1-10	5.4	1
318	Genome-wide study of DNA methylation shows alterations in metabolic, inflammatory, and cholesterol pathways in ALS.. *Science Translational Medicine*, **2022**, 14, eabj0264	17.5	4
317	Transcriptome-wide association study reveals increased neuronal FLT3 expression is associated with Tourette's syndrome.. *Communications Biology*, **2022**, 5, 289	6.7	0
316	Lack of association of TP73 with amyotrophic lateral sclerosis in a large cohort of cases.. *Neurobiology of Aging*, **2022**, 115, 109-111	5.6	0
315	Genetic, structural and clinical analysis of spastic paraplegia 4.. *Parkinsonism and Related Disorders*, **2022**, 98, 62-69	3.6	0
314	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
313	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
312	Hereditary spastic paraplegia initially diagnosed as cerebral palsy. *Clinical Parkinsonism & Related Disorders*, **2021**, 5, 100114	0.9	0
311	Enrichment of alleles encoding variants with constitutively hyperactive NADase in patients with ALS and other motor nerve disorders. *ELife*, **2021**, 10,	8.9	8
310	Combining schizophrenia and depression polygenic risk scores improves the genetic prediction of lithium response in bipolar disorder patients. *Translational Psychiatry*, **2021**, 11, 606	8.6	1
309	Heterozygous de novo KPNA3 Mutations Cause Complex Hereditary Spastic Paraplegia. *Annals of Neurology*, **2021**,	9.4	1
308	Influence of polygenic risk scores for schizophrenia and resilience on the cognition of individuals at-risk for psychosis. *Translational Psychiatry*, **2021**, 11, 518	8.6	1
307	Rare PSAP Variants and Possible Interaction with GBA in REM Sleep Behavior Disorder. *Journal of Parkinson's Disease*, **2021**,	5.3	2
306	GCH1 mutations in hereditary spastic paraplegia. *Clinical Genetics*, **2021**, 100, 51-58	4	3
305	Genetic Basis of ALS **2021**, 17-34		0

304	Association study of DNAJC13, UCHL1, HTRA2, GIGYF2, and EIF4G1 with Parkinson's disease. *Neurobiology of Aging*, **2021**, 100, 119.e7-119.e13	5.6	8
303	Is Persistent Motor or Vocal Tic Disorder a Milder Form of Tourette Syndrome?. *Movement Disorders*, **2021**, 36, 1899-1910	7	7
302	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
301	Chronic lithium treatment alters the excitatory/ inhibitory balance of synaptic networks and reduces mGluR5-PKC signalling in mouse cortical neurons. *Journal of Psychiatry and Neuroscience*, **2021**, 46, E402-E414	4.5	2
300	Diagnostic Yield of Whole Exome Sequencing for Adults with Ataxia: a Brazilian Perspective. *Cerebellum*, **2021**, 1	4.3	0
299	Lack of Causal Effects or Genetic Correlation between Restless Legs Syndrome and Parkinson's Disease. *Movement Disorders*, **2021**, 36, 1967-1972	7	0
298	Occurrence of Amyotrophic Lateral Sclerosis in Type 1 Gaucher Disease. *Neurology: Genetics*, **2021**, 7, e600	3.8	1
297	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	7.9	1
296	Expanded CAG Repeats in ATXN1, ATXN2, ATXN3, and HTT in the 1000 Genomes Project. *Movement Disorders*, **2021**, 36, 514-518	7	2
295	Comprehensive Analysis of Familial Parkinsonism Genes in Rapid-Eye-Movement Sleep Behavior Disorder. *Movement Disorders*, **2021**, 36, 235-240	7	7
294	Analysis of Heterozygous PRKN Variants and Copy-Number Variations in Parkinson's Disease. *Movement Disorders*, **2021**, 36, 178-187	7	11
293	Targeted sequencing of Parkinson's disease loci genes highlights SYT11, FGF20 and other associations. *Brain*, **2021**, 144, 462-472	11.2	11
292	Exome-wide rare variant analysis in familial essential tremor. *Parkinsonism and Related Disorders*, **2021**, 82, 109-116	3.6	5
291	Exemplar scoring identifies genetically separable phenotypes of lithium responsive bipolar disorder. *Translational Psychiatry*, **2021**, 11, 36	8.6	5
290	Synaptic processes and immune-related pathways implicated in Tourette syndrome. *Translational Psychiatry*, **2021**, 11, 56	8.6	11
289	Genetic and Epidemiological Study of Adult Ataxia and Spastic Paraplegia in Eastern Quebec. *Canadian Journal of Neurological Sciences*, **2021**, 48, 655-665	1	0
288	Genome-Wide Association Study Meta-Analysis for Parkinson Disease Motor Subtypes. *Neurology: Genetics*, **2021**, 7, e557	3.8	4
287	Evidence for Non-Mendelian Inheritance in Spastic Paraplegia 7. *Movement Disorders*, **2021**, 36, 1664-1675	7	7

286	Characterisation of age and polarity at onset in bipolar disorder.. *British Journal of Psychiatry*, **2021**, 219, 659-669	5.4	2
285	Association of Variants in the SPTLC1 Gene With Juvenile Amyotrophic Lateral Sclerosis. *JAMA Neurology*, **2021**, 78, 1236-1248	17.2	5
284	HLA-DRB1 and HLA-DQB1 genetic diversity modulates response to lithium in bipolar affective disorders. *Scientific Reports*, **2021**, 11, 17823	4.9	1
283	Dissecting the Shared Genetic Architecture of Suicide Attempt, Psychiatric Disorders, and Known Risk Factors. *Biological Psychiatry*, **2021**,	7.9	11
282	Novel Associations of and With REM Sleep Behavior Disorder. *Neurology*, **2021**, 96, e1402-e1412	6.5	3
281	Assessing the NOTCH2NLC GGC expansion in European patients with essential tremor. *Brain*, **2020**, 143, e89	11.2	7
280	Characterization of human iPSC-derived astrocytes with potential for disease modeling and drug discovery. *Neuroscience Letters*, **2020**, 731, 135028	3.3	20
279	Oligogenicity, C9orf72 expansion, and variant severity in ALS. *Neurogenetics*, **2020**, 21, 227-242	3	6
278	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	5.3	3
277	variants in REM sleep behavior disorder: A multicenter study. *Neurology*, **2020**, 95, e1008-e1016	6.5	18
276	A polymorphism in the glutamate metabotropic receptor 7 is associated with cognitive deficits in the early phases of psychosis. *Schizophrenia Research*, **2020**,	3.6	4
275	A Physiological Instability Displayed in Hippocampal Neurons Derived From Lithium-Nonresponsive Bipolar Disorder Patients. *Biological Psychiatry*, **2020**, 88, 150-158	7.9	5
274	Machine learning analysis of exome trios to contrast the genomic architecture of autism and schizophrenia. *BMC Psychiatry*, **2020**, 20, 92	4.2	1
273	Reliability and correlation of mixture cell correction in methylomic and transcriptomic blood data. *BMC Research Notes*, **2020**, 13, 74	2.3	4
272	Fine-Mapping of SNCA in Rapid Eye Movement Sleep Behavior Disorder and Overt Synucleinopathies. *Annals of Neurology*, **2020**, 87, 584-598	9.4	24
271	The Quebec Parkinson Network: A Researcher-Patient Matching Platform and Multimodal Biorepository. *Journal of Parkinson's Disease*, **2020**, 10, 301-313	5.3	17
270	Clinical and genetic analysis of ATP13A2 in hereditary spastic paraplegia expands the phenotype. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1052	2.3	11
269	Multiomics Analyses Identify Genes and Pathways Relevant to Essential Tremor. *Movement Disorders*, **2020**, 35, 1153-1162	7	5

268	Variants in the Niemann-Pick type C gene NPC1 are not associated with Parkinson's disease. *Neurobiology of Aging*, **2020**, 93, 143.e1-143.e4	5.6	7
267	Exome sequencing in genetic disease: recent advances and considerations. *F1000Research*, **2020**, 9,	3.6	12
266	Genome-wide association study identifies genetic factors that modify age at onset in Machado-Joseph disease. *Aging*, **2020**, 12, 4742-4756	5.6	6
265	Stress, Cortisol and NR3C1 in At-Risk Individuals for Psychosis: A Mendelian Randomization Study. *Frontiers in Psychiatry*, **2020**, 11, 680	5	2
264	SMPD1 variants do not have a major role in rapid eye movement sleep behavior disorder. *Neurobiology of Aging*, **2020**, 93, 142.e5-142.e7	5.6	1
263	Genetics of primary lateral sclerosis. *Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration* , **2020**, 21, 28-34	3.6	5
262	Genetic and epidemiological characterization of restless legs syndrome in Québec. *Sleep*, **2020**, 43,	1.1	4
261	Genetic, Structural, and Functional Evidence Link TMEM175 to Synucleinopathies. *Annals of Neurology*, **2020**, 87, 139-153	9.4	40
260	Transcriptome-wide association study for restless legs syndrome identifies new susceptibility genes. *Communications Biology*, **2020**, 3, 373	6.7	2
259	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
258	Evolution of a Human-Specific Tandem Repeat Associated with ALS. *American Journal of Human Genetics*, **2020**, 107, 445-460	11	15
257	Assessing non-Mendelian inheritance in inherited axonopathies. *Genetics in Medicine*, **2020**, 22, 2114-2118	8.1	7
256	Transcriptomic Changes Resulting From Overexpression Identify Pathways Potentially Relevant to Essential Tremor. *Frontiers in Genetics*, **2020**, 11, 813	4.5	3
255	Missense variants in ATP1A3 and FXYD gene family are associated with childhood-onset schizophrenia. *Molecular Psychiatry*, **2020**, 25, 821-830	15.1	13
254	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
253	Analysis of common and rare variants in late-onset Parkinson disease. *Neurology: Genetics*, **2020**, 6, 385	3.8	13
252	Characterization of the phenotype with cognitive impairment and protein mislocalization in SCA34. *Neurology: Genetics*, **2020**, 6, e403	3.8	11
251	SPTAN1 variants as a potential cause for autosomal recessive hereditary spastic paraplegia. *Journal of Human Genetics*, **2019**, 64, 1145-1151	4.3	6

250	Transcriptome-wide association study of attention deficit hyperactivity disorder identifies associated genes and phenotypes. *Nature Communications*, **2019**, 10, 4450	17.4	25
249	and Restless Legs Syndrome: A Comprehensive Review. *Frontiers in Neurology*, **2019**, 10, 935	4.1	20
248	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
247	Neural function in DCC mutation carriers with and without mirror movements. *Annals of Neurology*, **2019**, 85, 433-442	9.4	5
246	Genome-wide estimates of heritability and genetic correlations in essential tremor. *Parkinsonism and Related Disorders*, **2019**, 64, 262-267	3.6	8
245	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
244	Somatic expansion of the hexanucleotide repeat does not occur in ALS spinal cord tissues. *Neurology: Genetics*, **2019**, 5, e317	3.8	7
243	Cognitive and Psychiatric Evaluation in SYNE1 Ataxia. *Cerebellum*, **2019**, 18, 731-737	4.3	2
242	Genome-wide association study identifies 30 loci associated with bipolar disorder. *Nature Genetics*, **2019**, 51, 793-803	36.3	662
241	RNA-Based Therapy Utilizing Oculopharyngeal Muscular Dystrophy Transcript Knockdown and Replacement. *Molecular Therapy - Nucleic Acids*, **2019**, 15, 12-25	10.7	6
240	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
239	Truncating Mutations in UBAP1 Cause Hereditary Spastic Paraplegia. *American Journal of Human Genetics*, **2019**, 104, 767-773	11	20
238	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
237	SMPD1 mutations, activity, and □synuclein accumulation in Parkinson's disease. *Movement Disorders*, **2019**, 34, 526-535	7	41
236	Genetic architecture and adaptations of Nunavik Inuit. *Proceedings of the National Academy of Sciences of the United States of America*, **2019**, 116, 16012-16017	11.5	5
235	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	5.3	4
234	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
233	Mineral absorption is an enriched pathway in a brain region of restless legs syndrome patients with reduced MEIS1 expression. *PLoS ONE*, **2019**, 14, e0225186	3.7	7

232	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
231	Genomic Relationships, Novel Loci, and Pleiotropic Mechanisms across Eight Psychiatric Disorders. *Cell*, **2019**, 179, 1469-1482.e11	56.2	402
230	CAPN1 mutations: Expanding the CAPN1-related phenotype: From hereditary spastic paraparesis to spastic ataxia. *European Journal of Medical Genetics*, **2019**, 62, 103605	2.6	13
229	Investigating the association and causal relationship between restless legs syndrome and essential tremor. *Parkinsonism and Related Disorders*, **2019**, 61, 238-240	3.6	4
228	Exome sequencing of sporadic childhood-onset schizophrenia suggests the contribution of X-linked genes in males. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2019**, 180, 335-340	3.5	3
227	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	6.2	2
226	Genetics of REM Sleep Behavior Disorder **2019**, 589-609		1
225	variant mitigates Alzheimer disease pathophysiology in vivo and postmortem. *Neurology: Genetics*, **2018**, 4, e216	3.8	5
224	Recessive mutations in VPS13D cause childhood onset movement disorders. *Annals of Neurology*, **2018**, 83, 1089-1095	9.4	61
223	Sleep disorders and Parkinson disease; lessons from genetics. *Sleep Medicine Reviews*, **2018**, 41, 101-112	10.2	24
222	Genetics of Intracranial Aneurysms. *Stroke*, **2018**, 49, 780-787	6.7	33
221	TOX3 Variants Are Involved in Restless Legs Syndrome and Parkinson's Disease with Opposite Effects. *Journal of Molecular Neuroscience*, **2018**, 64, 341-345	3.3	7
220	POLR3A variants in hereditary spastic paraplegia and ataxia. *Brain*, **2018**, 141, e1	11.2	14
219	Association study of essential tremor genetic loci in Parkinson's´disease. *Neurobiology of Aging*, **2018**, 66, 178.e13-178.e15	5.6	8
218	Exome sequencing reveals a novel PLP1 mutation in a Moroccan family with connatal Pelizaeus-Merzbacher disease: a case report. *BMC Pediatrics*, **2018**, 18, 90	2.6	0
217	A rare variant in MLKL confers susceptibility to ApoE e4-negative Alzheimer's disease in Hong Kong Chinese population. *Neurobiology of Aging*, **2018**, 68, 160.e1-160.e7	5.6	13
216	Genome-wide association analysis identifies new candidate risk loci for familial intracranial aneurysm in the French-Canadian population. *Scientific Reports*, **2018**, 8, 4356	4.9	10
215	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. *Neuron*, **2018**, 97, 1268-1283.e6	13.9	296

214	LRRK2 protective haplotype and full sequencing study in REM sleep behavior disorder. *Parkinsonism and Related Disorders*, **2018**, 52, 98-101	3.6	16
213	Identification of a rare pathway mutation in a non-syndromic human brain arteriovenous malformation via exome sequencing. *Human Genome Variation*, **2018**, 5, 18001	1.8	6
212	Non-invasive prenatal aneuploidy testing: Critical diagnostic performance parameters predict sample z-score values. *Clinical Biochemistry*, **2018**, 59, 69-77	3.5	4
211	Multimodal neuroimaging analysis in patients with SYNE1 Ataxia. *Journal of the Neurological Sciences*, **2018**, 390, 227-230	3.2	9
210	Analysis of the Influence of microRNAs in Lithium Response in Bipolar Disorder. *Frontiers in Psychiatry*, **2018**, 9, 207	5	15
209	Analysis of shared heritability in common disorders of the brain. *Science*, **2018**, 360,	33.3	666
208	Valproic acid is protective in cellular and worm models of oculopharyngeal muscular dystrophy. *Neurology*, **2018**, 91, e551-e561	6.5	4
207	A direct interaction between two Restless Legs Syndrome predisposing genes: MEIS1 and SKOR1. *Scientific Reports*, **2018**, 8, 12173	4.9	16
206	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
205	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
204	Screening of novel restless legs syndrome-associated genes in French-Canadian families. *Neurology: Genetics*, **2018**, 4, e296	3.8	4
203	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
202	Triple A syndrome presenting as complicated hereditary spastic paraplegia. *Molecular Genetics & Genomic Medicine*, **2018**, 6, 1134-1139	2.3	10
201	Reassessing GWAS findings for the shared genetic basis of insomnia and restless legs syndrome. *Sleep*, **2018**, 41,	1.1	6
200	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
199	Global characterization of copy number variants in epilepsy patients from whole genome sequencing. *PLoS Genetics*, **2018**, 14, e1007285	6	29
198	Sequencing of the GBA coactivator, Saposin C, in Parkinson disease. *Neurobiology of Aging*, **2018**, 72, 187.e1-187.e3	5.6	12
197	Teneurin transmembrane protein 4 is not a cause for essential tremor in a Canadian population. *Movement Disorders*, **2017**, 32, 292-295	7	11

196	Mutations in DCC cause isolated agenesis of the corpus callosum with incomplete penetrance. *Nature Genetics*, **2017**, 49, 511-514	36.3	54
195	RIC3 variants are not associated with Parkinson's disease in French-Canadians and French. *Neurobiology of Aging*, **2017**, 53, 194.e9-194.e11	5.6	4
194	Systematic review of autosomal recessive ataxias and proposal for a classification. *Cerebellum and Ataxias*, **2017**, 4, 3	1.7	42
193	Dysfunction of the Cerebral Glucose Transporter SLC45A1 in Individuals with Intellectual Disability and Epilepsy. *American Journal of Human Genetics*, **2017**, 100, 824-830	11	13
192	De Novo Coding Variants Are Strongly Associated with Tourette Disorder. *Neuron*, **2017**, 94, 486-499.e9	13.9	89
191	Rare Copy Number Variants in NRXN1 and CNTN6 Increase Risk for Tourette Syndrome. *Neuron*, **2017**, 94, 1101-1111.e7	13.9	103
190	Missense Mutation Associated With Fatty Acid Metabolism and Reduced Height in Greenlanders. *Circulation: Cardiovascular Genetics*, **2017**, 10,		19
189	Heterozygous PINK1 p.G411S in rapid eye movement sleep behaviour disorder. *Brain*, **2017**, 140, e32	11.2	5
188	Genetics of restless legs syndrome. *Sleep Medicine*, **2017**, 31, 18-22	4.6	25
187	KCNA2 mutations are rare in hereditary spastic paraplegia. *Annals of Neurology*, **2017**, 81, 325-326	9.4	
186	Clinical and genetic study of hereditary spastic paraplegia in Canada. *Neurology: Genetics*, **2017**, 3, e122	3.8	54
185	Clinical Spectrum of Amyotrophic Lateral Sclerosis (ALS). *Cold Spring Harbor Perspectives in Medicine* , **2017**, 7,	5.4	66
184	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. *American Journal of Human Genetics*, **2017**, 101, 664-685	11	214
183	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
182	The Tanenbaum Open Science Institute: Leading a Paradigm Shift at the Montreal Neurological Institute. *Neuron*, **2017**, 95, 1002-1006	13.9	19
181	Post-concussion symptoms and chronic pain after mild traumatic brain injury are modulated by multiple locus effect in the gene through the expression of antisense: A pilot prospective control study.. *Canadian Journal of Pain*, **2017**, 1, 112-126	1.5	2
180	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
179	KCC3 loss-of-function contributes to Andermann syndrome by inducing activity-dependent neuromuscular junction defects. *Neurobiology of Disease*, **2017**, 106, 35-48	7.5	7

178	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
177	[P1156]: FAMILIAL AGGREGATION OF ATYPICAL DEMENTIA IN A LARGE CANADIAN FAMILY: THE MISSING GENE **2017**, 13, P302-P303		
176	No rare deleterious variants from , , and are associated with essential tremor. *Neurology: Genetics*, **2017**, 3, e195	3.8	2
175	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
174	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
173	Calpain 1 in neurodegeneration: a therapeutic target?. *Lancet Neurology, The*, **2016**, 15, 1118	24.1	4
172	NEK1 variants confer susceptibility to amyotrophic lateral sclerosis. *Nature Genetics*, **2016**, 48, 1037-42	36.3	149
171	SYNE1 mutations cause autosomal-recessive ataxia with retained reflexes in Brazilian patients. *Movement Disorders*, **2016**, 31, 1754-1756	7	10
170	RNF213 Is Associated with Intracranial Aneurysms in the French-Canadian Population. *American Journal of Human Genetics*, **2016**, 99, 1072-1085	11	38
169	Inhibition of the kinase WNK1/HSN2 ameliorates neuropathic pain by restoring GABA inhibition. *Science Signaling*, **2016**, 9, ra32	8.8	30
168	CCNF mutations in amyotrophic lateral sclerosis and frontotemporal dementia. *Nature Communications*, **2016**, 7, 11253	17.4	126
167	Genome-wide association study in essential tremor identifies three new loci. *Brain*, **2016**, 139, 3163-3169	11.2	57
166	Molecular, Cellular, and Genetic Determinants of Sporadic Brain Arteriovenous Malformations. *Neurosurgery*, **2016**, 63 Suppl 1, 37-42	3.2	11
165	FET proteins regulate lifespan and neuronal integrity. *Scientific Reports*, **2016**, 6, 25159	4.9	9
164	variants and glucocerebrosidase activity in Parkinson's disease. *Npj Parkinson's Disease*, **2016**, 2,	9.7	22
163	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
162	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
161	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

160	Genetic variants associated with response to lithium treatment in bipolar disorder: a genome-wide association study. *Lancet, The*, **2016**, 387, 1085-1093	40	216
159	SPG7 mutations explain a significant proportion of French Canadian spastic ataxia cases. *European Journal of Human Genetics*, **2016**, 24, 1016-21	5.3	38
158	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
157	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
156	Conserved pharmacological rescue of hereditary spastic paraplegia-related phenotypes across model organisms. *Human Molecular Genetics*, **2016**, 25, 1088-99	5.6	16
155	De novo variants in sporadic cases of childhood onset schizophrenia. *European Journal of Human Genetics*, **2016**, 24, 944-8	5.3	56
154	Replication study of MATR3 in familial and sporadic amyotrophic lateral sclerosis. *Neurobiology of Aging*, **2016**, 37, 209.e17-209.e21	5.6	41
153	Case-Control and Family-Based Association Study of Specific Variants in Restless Legs Syndrome. *Movement Disorders Clinical Practice*, **2016**, 3, 460-464	2.2	
152	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
151	KCC3 axonopathy: neuropathological features in the central and peripheral nervous system. *Modern Pathology*, **2016**, 29, 962-76	9.8	8
150	The role of the melanoma gene MC1R in Parkinson disease and REM sleep behavior disorder. *Neurobiology of Aging*, **2016**, 43, 180.e7-180.e13	5.6	10
149	Mutations in CAPN1 Cause Autosomal-Recessive Hereditary Spastic Paraplegia. *American Journal of Human Genetics*, **2016**, 98, 1038-1046	11	70
148	De novo FUS P525L mutation in Juvenile amyotrophic lateral sclerosis with dysphonia and diplopia. *Neurology: Genetics*, **2016**, 2, e63	3.8	24
147	ALS: Recent Developments from Genetics Studies. *Current Neurology and Neuroscience Reports*, **2016**, 16, 59	6.6	45
146	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	2.6	6
145	GBA p.T369M substitution in Parkinson disease: Polymorphism or association? A meta-analysis. *Neurology: Genetics*, **2016**, 2, e104	3.8	51
144	Preclinical target validation using patient-derived cells. *Nature Reviews Drug Discovery*, **2015**, 14, 149-50	64.1	40
143	Genetic perspective on the role of the autophagy-lysosome pathway in Parkinson disease. *Autophagy*, **2015**, 11, 1443-57	10.2	168

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2	Genome-wide association study of REM sleep behavior disorder identifies novel loci with distinct polygenic and brain expression effects		1
1	Molecular Genetics of Essential Tremor1-8		0