Alexis Brice

List of Publications by Citations

809 papers	75,143 citations	131 h-index	245 g-index
864 ext. papers	86,546 ext. citations	9 avg, IF	7.05 L-index

#	Paper	IF	Citations
809	Meta-analysis of 74,046 individuals identifies 11 new susceptibility loci for Alzheimer's disease. *Nature Genetics*, **2013**, 45, 1452-8	36.3	2714
808	Friedreich's ataxia: autosomal recessive disease caused by an intronic GAA triplet repeat expansion. *Science*, **1996**, 271, 1423-7	33.3	2281
807	Mutations in the DJ-1 gene associated with autosomal recessive early-onset parkinsonism. *Science*, **2003**, 299, 256-9	33.3	2093
806	Common variants at ABCA7, MS4A6A/MS4A4E, EPHA1, CD33 and CD2AP are associated with Alzheimer's disease. *Nature Genetics*, **2011**, 43, 429-35	36.3	1421
805	Multicenter analysis of glucocerebrosidase mutations in Parkinson's disease. *New England Journal of Medicine*, **2009**, 361, 1651-61	59.2	1351
804	Mutations of the X-linked genes encoding neuroligins NLGN3 and NLGN4 are associated with autism. *Nature Genetics*, **2003**, 34, 27-9	36.3	1335
803	Large-scale meta-analysis of genome-wide association data identifies six new risk loci for Parkinson's disease. *Nature Genetics*, **2014**, 46, 989-93	36.3	1261
802	Association between early-onset Parkinson's disease and mutations in the parkin gene. *New England Journal of Medicine*, **2000**, 342, 1560-7	59.2	1242
801	Phenotype, genotype, and worldwide genetic penetrance of LRRK2-associated Parkinson's disease: a case-control study. *Lancet Neurology, The*, **2008**, 7, 583-90	24.1	1075
800	APP locus duplication causes autosomal dominant early-onset Alzheimer disease with cerebral amyloid angiopathy. *Nature Genetics*, **2006**, 38, 24-6	36.3	928
799	Genetic meta-analysis of diagnosed Alzheimer's disease identifies new risk loci and implicates A tau, immunity and lipid processing. *Nature Genetics*, **2019**, 51, 414-430	36.3	917
798	Clinical and genetic abnormalities in patients with Friedreich's ataxia. *New England Journal of Medicine*, **1996**, 335, 1169-75	59.2	872
797	Causal relation between alpha-synuclein gene duplication and familial Parkinson's disease. *Lancet, The*, **2004**, 364, 1169-71	40	869
796	Frequency of the C9orf72 hexanucleotide repeat expansion in patients with amyotrophic lateral sclerosis and frontotemporal dementia: a cross-sectional study. *Lancet Neurology, The*, **2012**, 11, 323-30	24.1	830
795	Mutations of SCN1A, encoding a neuronal sodium channel, in two families with GEFS+2. *Nature Genetics*, **2000**, 24, 343-5	36.3	794
794	Cloning of the gene for spinocerebellar ataxia 2 reveals a locus with high sensitivity to expanded CAG/glutamine repeats. *Nature Genetics*, **1996**, 14, 285-91	36.3	765
793	Imputation of sequence variants for identification of genetic risks for Parkinson's disease: a meta-analysis of genome-wide association studies. *Lancet, The*, **2011**, 377, 641-9	40	733

792	Cloning of the SCA7 gene reveals a highly unstable CAG repeat expansion. *Nature Genetics*, **1997**, 17, 65-70	36.3	694
791	Parkinson's disease: from monogenic forms to genetic susceptibility factors. *Human Molecular Genetics*, **2009**, 18, R48-59	5.6	668
790	First genetic evidence of GABA(A) receptor dysfunction in epilepsy: a mutation in the gamma2-subunit gene. *Nature Genetics*, **2001**, 28, 46-8	36.3	618
789	Polyglutamine expansion as a pathological epitope in Huntington's disease and four dominant cerebellar ataxias. *Nature*, **1995**, 378, 403-6	50.4	582
788	Early-onset autosomal dominant Alzheimer disease: prevalence, genetic heterogeneity, and mutation spectrum. *American Journal of Human Genetics*, **1999**, 65, 664-70	11	574
787	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
786	A wide variety of mutations in the parkin gene are responsible for autosomal recessive parkinsonism in Europe. French Parkinson's Disease Genetics Study Group and the European Consortium on Genetic Susceptibility in Parkinson's Disease. *Human Molecular Genetics*, **1999**, 8, 567-74	5.6	524
785	Rare coding variants in PLCG2, ABI3, and TREM2 implicate microglial-mediated innate immunity in Alzheimer's disease. *Nature Genetics*, **2017**, 49, 1373-1384	36.3	508
784	Spastin, a new AAA protein, is altered in the most frequent form of autosomal dominant spastic paraplegia. *Nature Genetics*, **1999**, 23, 296-303	36.3	501
783	G51D □synuclein mutation causes a novel parkinsonian-pyramidal syndrome. *Annals of Neurology*, **2013**, 73, 459-71	9.4	462
782	LRRK2 G2019S as a cause of Parkinson's disease in North African Arabs. *New England Journal of Medicine*, **2006**, 354, 422-3	59.2	435
781	What genetics tells us about the causes and mechanisms of Parkinson's disease. *Physiological Reviews*, **2011**, 91, 1161-218	47.9	422
780	Comprehensive research synopsis and systematic meta-analyses in Parkinson's disease genetics: The PDGene database. *PLoS Genetics*, **2012**, 8, e1002548	6	420
779	Genome-wide scan for autism susceptibility genes. Paris Autism Research International Sibpair Study. *Human Molecular Genetics*, **1999**, 8, 805-12	5.6	406
778	Parkin gene inactivation alters behaviour and dopamine neurotransmission in the mouse. *Human Molecular Genetics*, **2003**, 12, 2277-91	5.6	403
777	Senataxin, the ortholog of a yeast RNA helicase, is mutant in ataxia-ocular apraxia 2. *Nature Genetics*, **2004**, 36, 225-7	36.3	385
776	Exome sequencing links corticospinal motor neuron disease to common neurodegenerative disorders. *Science*, **2014**, 343, 506-511	33.3	374
775	Meta-analysis of SHANK Mutations in Autism Spectrum Disorders: a gradient of severity in cognitive impairments. *PLoS Genetics*, **2014**, 10, e1004580	6	340

774	Genome-wide association analyses identify new risk variants and the genetic architecture of amyotrophic lateral sclerosis. *Nature Genetics*, **2016**, 48, 1043-8	36.3	328
773	Friedreich's ataxia: point mutations and clinical presentation of compound heterozygotes. *Annals of Neurology*, **1999**, 45, 200-6	9.4	327
772	Spinocerebellar ataxia 3 and Machado-Joseph disease: clinical, molecular, and neuropathological features. *Annals of Neurology*, **1996**, 39, 490-9	9.4	318
771	DJ-1(PARK7), a novel gene for autosomal recessive, early onset parkinsonism. *Neurological Sciences*, **2003**, 24, 159-60	3.5	313
770	Parkin prevents mitochondrial swelling and cytochrome c release in mitochondria-dependent cell death. *Human Molecular Genetics*, **2003**, 12, 517-26	5.6	310
769	Hereditary spastic paraplegia SPG13 is associated with a mutation in the gene encoding the mitochondrial chaperonin Hsp60. *American Journal of Human Genetics*, **2002**, 70, 1328-32	11	308
768	Spectrin mutations cause spinocerebellar ataxia type 5. *Nature Genetics*, **2006**, 38, 184-90	36.3	300
767	A regulated interaction with the UIM protein Eps15 implicates parkin in EGF receptor trafficking and PI(3)K-Akt signalling. *Nature Cell Biology*, **2006**, 8, 834-42	23.4	299
766	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. *Neuron*, **2018**, 97, 1268-1283.e6	13.9	296
765	A mitochondrial origin for frontotemporal dementia and amyotrophic lateral sclerosis through CHCHD10 involvement. *Brain*, **2014**, 137, 2329-45	11.2	292
764	A multicenter study of glucocerebrosidase mutations in dementia with Lewy bodies. *JAMA Neurology*, **2013**, 70, 727-35	17.2	285
763	Phenotype variability in progranulin mutation carriers: a clinical, neuropsychological, imaging and genetic study. *Brain*, **2008**, 131, 732-46	11.2	275
762	Mutations in SPG11, encoding spatacsin, are a major cause of spastic paraplegia with thin corpus callosum. *Nature Genetics*, **2007**, 39, 366-72	36.3	268
761	Clinical and pathologic abnormalities in a family with parkinsonism and parkin gene mutations. *Neurology*, **2001**, 56, 555-7	6.5	268
760	Lentiviral vector delivery of parkin prevents dopaminergic degeneration in an alpha-synuclein rat model of Parkinson's disease. *Proceedings of the National Academy of Sciences of the United States of America*, **2004**, 101, 17510-5	11.5	263
759	Chromosomal assignment of the second locus for autosomal dominant cerebellar ataxia (SCA2) to chromosome 12q23-24.1. *Nature Genetics*, **1993**, 4, 295-9	36.3	257
758	Autosomal dominant cerebellar ataxia type I clinical features and MRI in families with SCA1, SCA2 and SCA3. *Brain*, **1996**, 119 (Pt 5), 1497-505	11.2	252
757	Mutations in COQ2 in familial and sporadic multiple-system atrophy. *New England Journal of Medicine*, **2013**, 369, 233-44	59.2	251

756	A repeat expansion in the gene encoding junctophilin-3 is associated with Huntington disease-like 2. *Nature Genetics*, **2001**, 29, 377-8	36.3	250
755	Spinocerebellar ataxia type 7 (SCA7): a neurodegenerative disorder with neuronal intranuclear inclusions. *Human Molecular Genetics*, **1998**, 7, 913-8	5.6	249
754	Molecular and clinical correlations in spinocerebellar ataxia 2: a study of 32 families. *Human Molecular Genetics*, **1997**, 6, 709-15	5.6	248
753	Parkin mutations are frequent in patients with isolated early-onset parkinsonism. *Brain*, **2003**, 126, 1271-8	11.2	245
752	TARDBP mutations in motoneuron disease with frontotemporal lobar degeneration. *Annals of Neurology*, **2009**, 65, 470-3	9.4	240
751	The natural history of degenerative ataxia: a retrospective study in 466 patients. *Brain*, **1998**, 121 (Pt 4), 589-600	11.2	239
750	Loss of function of C9orf72 causes motor deficits in a zebrafish model of amyotrophic lateral sclerosis. *Annals of Neurology*, **2013**, 74, 180-7	9.4	238
749	Association of LRRK2 exonic variants with susceptibility to Parkinson's disease: a case-control study. *Lancet Neurology, The*, **2011**, 10, 898-908	24.1	237
748	Mutations in voltage-gated potassium channel KCNC3 cause degenerative and developmental central nervous system phenotypes. *Nature Genetics*, **2006**, 38, 447-51	36.3	236
747	Sporadic infantile epileptic encephalopathy caused by mutations in PCDH19 resembles Dravet syndrome but mainly affects females. *PLoS Genetics*, **2009**, 5, e1000381	6	232
746	Mutations in MTMR13, a new pseudophosphatase homologue of MTMR2 and Sbf1, in two families with an autosomal recessive demyelinating form of Charcot-Marie-Tooth disease associated with early-onset glaucoma. *American Journal of Human Genetics*, **2003**, 72, 1141-53	11	232
745	Mutations of the presenilin I gene in families with early-onset Alzheimer's disease. *Human Molecular Genetics*, **1995**, 4, 2373-7	5.6	227
744	SOD1, ANG, VAPB, TARDBP, and FUS mutations in familial amyotrophic lateral sclerosis: genotype-phenotype correlations. *Journal of Medical Genetics*, **2010**, 47, 554-60	5.8	226
743	Loss of VPS13C Function in Autosomal-Recessive Parkinsonism Causes Mitochondrial Dysfunction and Increases PINK1/Parkin-Dependent Mitophagy. *American Journal of Human Genetics*, **2016**, 98, 500-513	11	225
742	How much phenotypic variation can be attributed to parkin genotype?. *Annals of Neurology*, **2003**, 54, 176-85	9.4	224
741	Segregation of a missense mutation in the microtubule-associated protein tau gene with familial frontotemporal dementia and parkinsonism. *Human Molecular Genetics*, **1998**, 7, 1825-9	5.6	223
740	Excessive burden of lysosomal storage disorder gene variants in Parkinson's disease. *Brain*, **2017**, 140, 3191-3203	11.2	209
739	A two-stage meta-analysis identifies several new loci for Parkinson's disease. *PLoS Genetics*, **2011**, 7, e1002142	6	209

738	Frontotemporal dementia and its subtypes: a genome-wide association study. *Lancet Neurology, The*, **2014**, 13, 686-99	24.1	207
737	Large-scale screening of the Gaucher's disease-related glucocerebrosidase gene in Europeans with Parkinson's disease. *Human Molecular Genetics*, **2011**, 20, 202-10	5.6	206
736	Levodopa-responsive dystonia. GTP cyclohydrolase I or parkin mutations?. *Brain*, **2000**, 123 (Pt 6), 1112-21	11.2	206
735	A deleterious mutation in DNAJC6 encoding the neuronal-specific clathrin-uncoating co-chaperone auxilin, is associated with juvenile parkinsonism. *PLoS ONE*, **2012**, 7, e36458	3.7	203
734	The p38 subunit of the aminoacyl-tRNA synthetase complex is a Parkin substrate: linking protein biosynthesis and neurodegeneration. *Human Molecular Genetics*, **2003**, 12, 1427-37	5.6	198
733	Alzheimer's disease associated with mutations in presenilin 2 is rare and variably penetrant. *Human Molecular Genetics*, **1996**, 5, 985-8	5.6	198
732	Spectrum of SPG4 mutations in autosomal dominant spastic paraplegia. *Human Molecular Genetics*, **2000**, 9, 637-44	5.6	196
731	Autosomal dominant cerebellar ataxia type I in Martinique (French West Indies). Clinical and neuropathological analysis of 53 patients from three unrelated SCA2 families. *Brain*, **1995**, 118 (Pt 6), 1573-81	11.2	193
730	Clinical correlations with Lewy body pathology in LRRK2-related Parkinson disease. *JAMA Neurology*, **2015**, 72, 100-5	17.2	191
729	Biochemical analysis of Parkinson's disease-causing variants of Parkin, an E3 ubiquitin-protein ligase with monoubiquitylation capacity. *Human Molecular Genetics*, **2006**, 15, 2059-75	5.6	191
728	Molecular and clinical correlations in autosomal dominant cerebellar ataxia with progressive macular dystrophy (SCA7). *Human Molecular Genetics*, **1998**, 7, 165-70	5.6	189
727	Charcot-Marie-Tooth disease type 1A with 17p11.2 duplication. Clinical and electrophysiological phenotype study and factors influencing disease severity in 119 cases. *Brain*, **1997**, 120 (Pt 5), 813-23	11.2	188
726	Close associations between prevalences of dominantly inherited spinocerebellar ataxias with CAG-repeat expansions and frequencies of large normal CAG alleles in Japanese and Caucasian populations. *American Journal of Human Genetics*, **1998**, 63, 1060-6	11	187
725	. *Nature Genetics*, **2001**, 28, 46-48	36.3	185
724	Complete sequence of a cDNA encoding an active rat choline acetyltransferase: a tool to investigate the plasticity of cholinergic phenotype expression. *Journal of Neuroscience Research*, **1989**, 23, 266-73	4.4	182
723	Evidence for a role of the rare p.A152T variant in MAPT in increasing the risk for FTD-spectrum and Alzheimer's diseases. *Human Molecular Genetics*, **2012**, 21, 3500-12	5.6	174
722	Ataxia with oculomotor apraxia type 2: clinical, biological and genotype/phenotype correlation study of a cohort of 90 patients. *Brain*, **2009**, 132, 2688-98	11.2	173
721	Cerebellar ataxia with oculomotor apraxia type 1: clinical and genetic studies. *Brain*, **2003**, 126, 2761-72	11.2	173

720	Unusual phenotypic alteration of beta amyloid precursor protein (betaAPP) maturation by a new Val-715 --> Met betaAPP-770 mutation responsible for probable early-onset Alzheimer's disease. *Proceedings of the National Academy of Sciences of the United States of America*, **1999**, 96, 4119-24	11.5	173
719	G2019S LRRK2 mutation in French and North African families with Parkinson's disease. *Annals of Neurology*, **2005**, 58, 784-7	9.4	172
718	Homozygous deletions in parkin gene in European and North African families with autosomal recessive juvenile parkinsonism. The European Consortium on Genetic Susceptibility in Parkinson's Disease and the French Parkinson's Disease Genetics Study Group. *Lancet, The*, **1998**, 352, 1355-6	40	171
717	Mutations in SPG11 are frequent in autosomal recessive spastic paraplegia with thin corpus callosum, cognitive decline and lower motor neuron degeneration. *Brain*, **2008**, 131, 772-84	11.2	170
716	Alpha-synuclein and Parkinson's disease. *Cellular and Molecular Life Sciences*, **2000**, 57, 1894-908	10.3	170
715	Identification of the SPG15 gene, encoding spastizin, as a frequent cause of complicated autosomal-recessive spastic paraplegia, including Kjellin syndrome. *American Journal of Human Genetics*, **2008**, 82, 992-1002	11	167
714	Clinical and molecular advances in autosomal dominant cerebellar ataxias: from genotype to phenotype and physiopathology. *European Journal of Human Genetics*, **2000**, 8, 4-18	5.3	165
713	Penetrance of Parkinson disease in glucocerebrosidase gene mutation carriers. *Neurology*, **2012**, 78, 417-20	6.5	159
712	A second locus for familial generalized epilepsy with febrile seizures plus maps to chromosome 2q21-q33. *American Journal of Human Genetics*, **1999**, 65, 1078-85	11	157
711	X-linked Charcot-Marie-Tooth disease with connexin 32 mutations: clinical and electrophysiologic study. *Neurology*, **1998**, 50, 1074-82	6.5	156
710	Specifically neuropathic Gaucher's mutations accelerate cognitive decline in Parkinson's. *Annals of Neurology*, **2016**, 80, 674-685	9.4	154
709	Complex relationship between Parkin mutations and Parkinson disease. *American Journal of Medical Genetics Part A*, **2002**, 114, 584-91		154
708	Frequency and phenotypic spectrum of ataxia with oculomotor apraxia 2: a clinical and genetic study in 18 patients. *Brain*, **2004**, 127, 759-67	11.2	152
707	Mapping of spinocerebellar ataxia 13 to chromosome 19q13.3-q13.4 in a family with autosomal dominant cerebellar ataxia and mental retardation. *American Journal of Human Genetics*, **2000**, 67, 229-35	11	151
706	Genome-wide Pleiotropy Between Parkinson Disease and Autoimmune Diseases. *JAMA Neurology*, **2017**, 74, 780-792	17.2	150
705	Alteration of fatty-acid-metabolizing enzymes affects mitochondrial form and function in hereditary spastic paraplegia. *American Journal of Human Genetics*, **2012**, 91, 1051-64	11	150
704	A mutation in periaxin is responsible for CMT4F, an autosomal recessive form of Charcot-Marie-Tooth disease. *Human Molecular Genetics*, **2001**, 10, 415-21	5.6	148
703	Transcriptional repression of p53 by parkin and impairment by mutations associated with autosomal recessive juvenile Parkinson's disease. *Nature Cell Biology*, **2009**, 11, 1370-5	23.4	147

702	Hereditary spastic paraplegias: an update. *Current Opinion in Neurology*, **2007**, 20, 674-80	7.1	147
701	Convergent genetic and expression data implicate immunity in Alzheimer's disease. *Alzheimer's and Dementia*, **2015**, 11, 658-71	1.2	146
700	De novo mutations in HCN1 cause early infantile epileptic encephalopathy. *Nature Genetics*, **2014**, 46, 640-5	36.3	145
699	Using genome-wide complex trait analysis to quantify 'missing heritability' in Parkinson's disease. *Human Molecular Genetics*, **2012**, 21, 4996-5009	5.6	145
698	Progranulin null mutations in both sporadic and familial frontotemporal dementia. *Human Mutation* , **2007**, 28, 846-55	4.7	143
697	Huntington's disease-like phenotype due to trinucleotide repeat expansions in the TBP and JPH3 genes. *Brain*, **2003**, 126, 1599-603	11.2	142
696	Autosomal dominant cerebellar ataxia type I. MRI-based volumetry of posterior fossa structures and basal ganglia in spinocerebellar ataxia types 1, 2 and 3. *Brain*, **1998**, 121 (Pt 9), 1687-93	11.2	142
695	Diagnosis of Parkinson's disease on the basis of clinical and genetic classification: a population-based modelling study. *Lancet Neurology, The*, **2015**, 14, 1002-9	24.1	141
694	Akt is altered in an animal model of Huntington's disease and in patients. *European Journal of Neuroscience*, **2005**, 21, 1478-88	3.5	140
693	Spectrum of clinical and electrophysiologic features in HNPP patients with the 17p11.2 deletion. *Neurology*, **1999**, 52, 1440-6	6.5	140
692	Long-term disease progression in spinocerebellar ataxia types 1, 2, 3, and 6: a longitudinal cohort study. *Lancet Neurology, The*, **2015**, 14, 1101-8	24.1	139
691	Genome-wide association study confirms BST1 and suggests a locus on 12q24 as the risk loci for Parkinson's disease in the European population. *Human Molecular Genetics*, **2011**, 20, 615-27	5.6	139
690	Guadeloupean parkinsonism: a cluster of progressive supranuclear palsy-like tauopathy. *Brain*, **2002** , 125, 801-11	11.2	137
689	Parkinson's disease age at onset genome-wide association study: Defining heritability, genetic loci, and □synuclein mechanisms. *Movement Disorders*, **2019**, 34, 866-875	7	136
688	Mutations in the GIGYF2 (TNRC15) gene at the PARK11 locus in familial Parkinson disease. *American Journal of Human Genetics*, **2008**, 82, 822-33	11	135
687	Are interrupted SCA2 CAG repeat expansions responsible for parkinsonism?. *Neurology*, **2007**, 69, 1970-5	6.5	135
686	Linkage of a new locus for autosomal dominant familial spastic paraplegia to chromosome 2p. *Human Molecular Genetics*, **1994**, 3, 1569-73	5.6	135
685	DNA repair pathways underlie a common genetic mechanism modulating onset in polyglutamine diseases. *Annals of Neurology*, **2016**, 79, 983-90	9.4	135

684	The gene for autosomal dominant cerebellar ataxia with pigmentary macular dystrophy maps to chromosome 3p12-p21.1. *Nature Genetics*, **1995**, 10, 84-8	36.3	134
683	Loss of function of glucocerebrosidase GBA2 is responsible for motor neuron defects in hereditary spastic paraplegia. *American Journal of Human Genetics*, **2013**, 92, 238-44	11	133
682	A genome-scale DNA repair RNAi screen identifies SPG48 as a novel gene associated with hereditary spastic paraplegia. *PLoS Biology*, **2010**, 8, e1000408	9.7	133
681	Demographic, neurological and behavioural characteristics and brain perfusion SPECT in frontal variant of frontotemporal dementia. *Brain*, **2006**, 129, 3051-65	11.2	132
680	Autosomal dominant cerebellar ataxia type I: oculomotor abnormalities in families with SCA1, SCA2, and SCA3. *Journal of Neurology*, **1999**, 246, 789-97	5.5	132
679	Phenotype difference between ALS patients with expanded repeats in C9ORF72 and patients with mutations in other ALS-related genes. *Journal of Medical Genetics*, **2012**, 49, 258-63	5.8	131
678	SQSTM1 mutations in French patients with frontotemporal dementia or frontotemporal dementia with amyotrophic lateral sclerosis. *JAMA Neurology*, **2013**, 70, 1403-10	17.2	131
677	Molecular diagnosis of autosomal dominant early onset Alzheimer's disease: an update. *Journal of Medical Genetics*, **2005**, 42, 793-5	5.8	131
676	Genetic variants of the alpha-synuclein gene SNCA are associated with multiple system atrophy. *PLoS ONE*, **2009**, 4, e7114	3.7	130
675	FXTAS: new insights and the need for revised diagnostic criteria. *Neurology*, **2012**, 79, 1898-907	6.5	129
674	Mutational analysis of the PINK1 gene in early-onset parkinsonism in Europe and North Africa. *Brain* , **2006**, 129, 686-94	11.2	129
673	Proteomic analysis of parkin knockout mice: alterations in energy metabolism, protein handling and synaptic function. *Journal of Neurochemistry*, **2005**, 95, 1259-76	6	129
672	C9orf72 repeat expansions are a rare genetic cause of parkinsonism. *Brain*, **2013**, 136, 385-91	11.2	128
671	Spastic paraplegia gene 7 in patients with spasticity and/or optic neuropathy. *Brain*, **2012**, 135, 2980-93	11.2	128
670	Parkinson's disease in GTP cyclohydrolase 1 mutation carriers. *Brain*, **2014**, 137, 2480-92	11.2	127
669	Alteration of ganglioside biosynthesis responsible for complex hereditary spastic paraplegia. *American Journal of Human Genetics*, **2013**, 93, 118-23	11	124
668	Longitudinal analysis of impulse control disorders in Parkinson disease. *Neurology*, **2018**, 91, e189-e201	6.5	123
667	Ultrastructural PMP22 expression in inherited demyelinating neuropathies. *Annals of Neurology*, **1996**, 39, 813-7	9.4	122

666	CAG repeat expansion in the TATA box-binding protein gene causes autosomal dominant cerebellar ataxia. *Brain*, **2001**, 124, 1939-47	11.2	119
665	Brain white matter oedema due to ClC-2 chloride channel deficiency: an observational analytical study. *Lancet Neurology, The*, **2013**, 12, 659-68	24.1	115
664	Alpha-synuclein gene rearrangements in dominantly inherited parkinsonism: frequency, phenotype, and mechanisms. *Archives of Neurology*, **2009**, 66, 102-8		115
663	LRRK2 haplotype analyses in European and North African families with Parkinson disease: a common founder for the G2019S mutation dating from the 13th century. *American Journal of Human Genetics*, **2005**, 77, 330-2	11	115
662	Myoclonus-dystonia syndrome: epsilon-sarcoglycan mutations and phenotype. *Annals of Neurology*, **2002**, 52, 489-92	9.4	114
661	Screening for genomic rearrangements and methylation abnormalities of the 15q11-q13 region in autism spectrum disorders. *Biological Psychiatry*, **2009**, 66, 349-59	7.9	113
660	Mutations in KCND3 cause spinocerebellar ataxia type 22. *Annals of Neurology*, **2012**, 72, 859-69	9.4	111
659	Clinical and molecular features of spinocerebellar ataxia type 6. *Neurology*, **1997**, 49, 1243-6	6.5	111
658	Implication of the immune system in Alzheimer's disease: evidence from genome-wide pathway analysis. *Journal of Alzheimer's Disease*, **2010**, 20, 1107-18	4.3	109
657	Modulation of the age at onset in spinocerebellar ataxia by CAG tracts in various genes. *Brain*, **2014** , 137, 2444-55	11.2	108
656	Expanded CAG repeats in Swedish spinocerebellar ataxia type 7 (SCA7) patients: effect of CAG repeat length on the clinical manifestation. *Human Molecular Genetics*, **1998**, 7, 171-6	5.6	108
655	Genetic overlap between Alzheimer's disease and Parkinson's disease at the MAPT locus. *Molecular Psychiatry*, **2015**, 20, 1588-95	15.1	107
654	Large-scale replication and heterogeneity in Parkinson disease genetic loci. *Neurology*, **2012**, 79, 659-67	6.5	106
653	Exhaustive analysis of BH4 and dopamine biosynthesis genes in patients with Dopa-responsive dystonia. *Brain*, **2009**, 132, 1753-63	11.2	106
652	Parkin-mediated monoubiquitination of the PDZ protein PICK1 regulates the activity of acid-sensing ion channels. *Molecular Biology of the Cell*, **2007**, 18, 3105-18	3.5	106
651	Dementia with prominent frontotemporal features associated with L113P presenilin 1 mutation. *Neurology*, **2000**, 55, 1577-8	6.5	105
650	Ancestral origins of the Machado-Joseph disease mutation: a worldwide haplotype study. *American Journal of Human Genetics*, **2001**, 68, 523-8	11	103
649	SCA12 is a rare locus for autosomal dominant cerebellar ataxia: A study of an Indian family. *Annals of Neurology*, **2001**, 49, 117-121	9.4	102

648	Functional interplay between Parkin and Drp1 in mitochondrial fission and clearance. *Biochimica Et Biophysica Acta - Molecular Cell Research*, **2014**, 1843, 2012-26	4.9	101
647	Are cognitive changes the first symptoms of Huntington's disease? A study of gene carriers. *Journal of Neurology, Neurosurgery and Psychiatry*, **1998**, 64, 172-7	5.5	101
646	Complicated forms of autosomal dominant hereditary spastic paraplegia are frequent in SPG10. *Human Mutation*, **2009**, 30, E376-85	4.7	100
645	Parkinson's disease patients show reduced cortical-subcortical sensorimotor connectivity. *Movement Disorders*, **2013**, 28, 447-54	7	99
644	Prediction of cognition in Parkinson's disease with a clinical-genetic score: a longitudinal analysis of nine cohorts. *Lancet Neurology, The*, **2017**, 16, 620-629	24.1	98
643	Role of mendelian genes in "sporadic" Parkinson's disease. *Parkinsonism and Related Disorders*, **2012**, 18 Suppl 1, S66-70	3.6	98
642	Targeted next-generation sequencing of a 12.5 Mb homozygous region reveals ANO10 mutations in patients with autosomal-recessive cerebellar ataxia. *American Journal of Human Genetics*, **2010**, 87, 813-9	11	98
641	Consensus paper: pathological mechanisms underlying neurodegeneration in spinocerebellar ataxias. *Cerebellum*, **2014**, 13, 269-302	4.3	97
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