Alexis Brice

List of Publications by Year
in descending order

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830	95,931	311	597
papers	citations	142 h-index	267 g-index

866	866	866	63381
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Meta-analysis of 74,046 individuals identifies 11 new susceptibility loci for Alzheimer's disease. Nature Genetics, 2013, 45, 1452-1458.	9.4	3,741
2	Friedreich's Ataxia: Autosomal Recessive Disease Caused by an Intronic GAA Triplet Repeat Expansion. Science, 1996, 271, 1423-1427.	6.0	2,642
3	Mutations in the DJ-1 Gene Associated with Autosomal Recessive Early-Onset Parkinsonism. Science, 2003, 299, 256-259.	6.0	2,467
4	Genetic meta-analysis of diagnosed Alzheimerâ€™s disease identifies new risk loci and implicates AÎ², tau, immunity and lipid processing. Nature Genetics, 2019, 51, 414-430.	9.4	1,962
5	Multicenter Analysis of Glucocerebrosidase Mutations in Parkinson's Disease. New England Journal of Medicine, 2009, 361, 1651-1661.	13.9	1,747
6	Common variants at ABCA7, MS4A6A/MS4A4E, EPHA1, CD33 and CD2AP are associated with Alzheimer's disease. Nature Genetics, 2011, 43, 429-435.	9.4	1,708
7	Large-scale meta-analysis of genome-wide association data identifies six new risk loci for Parkinson's disease. Nature Genetics, 2014, 46, 989-993.	9.4	1,685
8	Mutations of the X-linked genes encoding neuroligins NLGN3 and NLGN4 are associated with autism. Nature Genetics, 2003, 34, 27-29.	9.4	1,612
9	Association between Early-Onset Parkinson's Disease and Mutations in theParkinGene. New England Journal of Medicine, 2000, 342, 1560-1567.	13.9	1,448
10	Identification of novel risk loci, causal insights, and heritable risk for Parkinson's disease: a meta-analysis of genome-wide association studies. Lancet Neurology, The, 2019, 18, 1091-1102.	4.9	1,414
11	Phenotype, genotype, and worldwide genetic penetrance of LRRK2-associated Parkinson's disease: a case-control study. Lancet Neurology, The, 2008, 7, 583-590.	4.9	1,340
12	APP locus duplication causes autosomal dominant early-onset Alzheimer disease with cerebral amyloid angiopathy. Nature Genetics, 2006, 38, 24-26.	9.4	1,087
13	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, 323-330.	4.9	1,039
14	Clinical and Genetic Abnormalities in Patients with Friedreich's Ataxia. New England Journal of Medicine, 1996, 335, 1169-1175.	13.9	1,015
15	Causal relation between Î±-synuclein locus duplication as a cause of familial Parkinson's disease. Lancet, The, 2004, 364, 1169-1171.	6.3	987
16	Mutations of SCN1A, encoding a neuronal sodium channel, in two families with GEFS+2. Nature Genetics, 2000, 24, 343-345.	9.4	910
17	Cloning of the gene for spinocerebellar ataxia 2 reveals a locus with high sensitivity to expanded CAG/glutamine repeats. Nature Genetics, 1996, 14, 285-291.	9.4	857
18	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, 641-649.	6.3	845

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19	Parkinson's disease: from monogenic forms to genetic susceptibility factors. Human Molecular Genetics, 2009, 18, R48-R59.	1.4	816
20	Rare coding variants in PLCG2, ABI3, and TREM2 implicate microglial-mediated innate immunity in Alzheimer's disease. Nature Genetics, 2017, 49, 1373-1384.	9.4	783
21	Cloning of the SCA7 gene reveals a highly unstable CAG repeat expansion. Nature Genetics, 1997, 17, 65-70.	9.4	758
22	First genetic evidence of GABAA receptor dysfunction in epilepsy: a mutation in the Î³2-subunit gene. Nature Genetics, 2001, 28, 46-48.	9.4	701
23	Early-Onset Autosomal Dominant Alzheimer Disease: Prevalence, Genetic Heterogeneity, and Mutation Spectrum. American Journal of Human Genetics, 1999, 65, 664-670.	2.6	696
24	Polyglutamine expansion as a pathological epitope in Huntington's disease and four dominant cerebellar ataxias. Nature, 1995, 378, 403-406.	13.7	632
25	G51D Î±â€synuclein mutation causes a novel Parkinsonianâ€“pyramidal syndrome. Annals of Neurology, 2013, 73, 459-471.	2.8	580
26	Spastin, a new AAA protein, is altered in the most frequent form of autosomal dominant spastic paraplegia. Nature Genetics, 1999, 23, 296-303.	9.4	575
27	A Wide Variety of Mutations in the Parkin Gene Are Responsible for Autosomal Recessive Parkinsonism in Europe. Human Molecular Genetics, 1999, 8, 567-574.	1.4	571
28	LRRK2G2019S as a Cause of Parkinson's Disease in North African Arabs. New England Journal of Medicine, 2006, 354, 422-423.	13.9	521
29	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. Neuron, 2018, 97, 1268-1283.e6.	3.8	517
30	What Genetics Tells us About the Causes and Mechanisms of Parkinson's Disease. Physiological Reviews, 2011, 91, 1161-1218.	13.1	515
31	Meta-analysis of SHANK Mutations in Autism Spectrum Disorders: A Gradient of Severity in Cognitive Impairments. PLoS Genetics, 2014, 10, e1004580.	1.5	501
32	Comprehensive Research Synopsis and Systematic Meta-Analyses in Parkinson's Disease Genetics: The PDGene Database. PLoS Genetics, 2012, 8, e1002548.	1.5	495
33	Genome-wide association analyses identify new risk variants and the genetic architecture of amyotrophic lateral sclerosis. Nature Genetics, 2016, 48, 1043-1048.	9.4	494
34	Exome Sequencing Links Corticospinal Motor Neuron Disease to Common Neurodegenerative Disorders. Science, 2014, 343, 506-511.	6.0	466
35	Parkin gene inactivation alters behaviour and dopamine neurotransmission in the mouse. Human Molecular Genetics, 2003, 12, 2277-2291.	1.4	462
36	Senataxin, the ortholog of a yeast RNA helicase, is mutant in ataxia-ocular apraxia 2. Nature Genetics, 2004, 36, 225-227.	9.4	454

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37	Genome-Wide Scan for Autism Susceptibility Genes. Human Molecular Genetics, 1999, 8, 805-812.	1.4	453
38	Spinocerebellar ataxia 3 and machado-joseph disease: Clinical, molecular, and neuropathological features. Annals of Neurology, 1996, 39, 490-499.	2.8	401
39	A mitochondrial origin for frontotemporal dementia and amyotrophic lateral sclerosis through CHCHD10 involvement. Brain, 2014, 137, 2329-2345.	3.7	377
40	A Multicenter Study of Glucocerebrosidase Mutations in Dementia With Lewy Bodies. JAMA Neurology, 2013, 70, 727.	4.5	374
41	Friedreich's ataxia: Point mutations and clinical presentation of compound heterozygotes. Annals of Neurology, 1999, 45, 200-206.	2.8	371
42	DJ-1(PARK7), a novel gene for autosomal recessive, early onset parkinsonism. Neurological Sciences, 2003, 24, 159-160.	0.9	363
43	Parkin prevents mitochondrial swelling and cytochrome c release in mitochondria-dependent cell death. Human Molecular Genetics, 2003, 12, 517-526.	1.4	352
44	Hereditary Spastic Paraplegia SPG13 Is Associated with a Mutation in the Gene Encoding the Mitochondrial Chaperonin Hsp60. American Journal of Human Genetics, 2002, 70, 1328-1332.	2.6	347
45	Spectrin mutations cause spinocerebellar ataxia type 5. Nature Genetics, 2006, 38, 184-190.	9.4	346
46	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.	2.6	333
47	Phenotype variability in progranulin mutation carriers: a clinical, neuropsychological, imaging and genetic study. Brain, 2008, 131, 732-746.	3.7	331
48	A regulated interaction with the UIM protein Eps15 implicates parkin in EGF receptor trafficking and PI(3)Kâ€“Akt signalling. Nature Cell Biology, 2006, 8, 834-842.	4.6	325
49	Excessive burden of lysosomal storage disorder gene variants in Parkinsonâ€™s disease. Brain, 2017, 140, 3191-3203.	3.7	323
50	The natural history of degenerative ataxia: a retrospective study in 466 patients. Brain, 1998, 121, 589-600.	3.7	316
51	Lentiviral vector delivery of parkin prevents dopaminergic degeneration in an Â-synuclein rat model of Parkinson's disease. Proceedings of the National Academy of Sciences of the United States of America, 2004, 101, 17510-17515.	3.3	310
52	Spinocerebellar ataxia type 7 (SCA7): a neurodegenerative disorder with neuronal intranuclear inclusions. Human Molecular Genetics, 1998, 7, 913-918.	1.4	308
53	Mutations in COQ2 in Familial and Sporadic Multiple-System Atrophy. New England Journal of Medicine, 2013, 369, 233-244.	13.9	308
54	Sporadic Infantile Epileptic Encephalopathy Caused by Mutations in PCDH19 Resembles Dravet Syndrome but Mainly Affects Females. PLoS Genetics, 2009, 5, e1000381.	1.5	304

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55	Mutations in SPG11, encoding spatacsin, are a major cause of spastic paraplegia with thin corpus callosum. Nature Genetics, 2007, 39, 366-372.	9.4	303
56	Frontotemporal dementia and its subtypes: a genome-wide association study. Lancet Neurology, The, 2014, 13, 686-699.	4.9	302
57	Chromosomal assignment of the second locus for autosomal dominant cerebellar ataxia (SCA2) to chromosome 12q23â€“24.1. Nature Genetics, 1993, 4, 295-299.	9.4	298
58	Association of LRRK2 exonic variants with susceptibility to Parkinson's disease: a caseâ€“control study. Lancet Neurology, The, 2011, 10, 898-908.	4.9	294
59	A repeat expansion in the gene encoding junctophilin-3 is associated with Huntington diseaseâ€“like 2. Nature Genetics, 2001, 29, 377-378.	9.4	288
60	Clinical and pathologic abnormalities in a family with parkinsonism and parkin gene mutations. Neurology, 2001, 56, 555-557.	1.5	288
61	Autosomal dominant cerebellar ataxia type I Clinical features and MRI in families with SCA1, SCA2 and SCA3. Brain, 1996, 119, 1497-1505.	3.7	285
62	Loss of function of C9orf72 causes motor deficits in a zebrafish model of amyotrophic lateral sclerosis. Annals of Neurology, 2013, 74, 180-187.	2.8	284
63	Parkin mutations are frequent in patients with isolated earlyâ€onset parkinsonism. Brain, 2003, 126, 1271-1278.	3.7	279
64	Mutations in voltage-gated potassium channel KCNC3 cause degenerative and developmental central nervous system phenotypes. Nature Genetics, 2006, 38, 447-451.	9.4	278
65	TARDBP mutations in motoneuron disease with frontotemporal lobar degeneration. Annals of Neurology, 2009, 65, 470-473.	2.8	278
66	Clinical Correlations With Lewy Body Pathology inLRRK2-Related Parkinson Disease. JAMA Neurology, 2015, 72, 100.	4.5	272
67	How much phenotypic variation can be attributed toparkingenotype?. Annals of Neurology, 2003, 54, 176-185.	2.8	271
68	Molecular and Clinical Correlations in Spinocerebellar Ataxia 2: A Study of 32 Families. Human Molecular Genetics, 1997, 6, 709-715.	1.4	270
69	Mutations of the presenilin I gene in families with early-onset Alzheimer's disease. Human Molecular Genetics, 1995, 4, 2373-2377.	1.4	268
70	SOD1, ANG, VAPB, TARDBP, and FUS mutations in familial amyotrophic lateral sclerosis: genotype-phenotype correlations. Journal of Medical Genetics, 2010, 47, 554-560.	1.5	266
71	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, 1141-1153.	2.6	263
72	Alzheimer's disease associated with mutations in presenilin 2 is rare and variably penetrant. Human Molecular Genetics, 1996, 5, 985-988.	1.4	259

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73	Segregation of a Missense Mutation in the Microtubule-Associated Protein Tau Gene with Familial Frontotemporal Dementia and Parkinsonism. Human Molecular Genetics, 1998, 7, 1825-1829.	1.4	258
74	Large-scale screening of the Gaucher's disease-related glucocerebrosidase gene in Europeans with Parkinson's disease. Human Molecular Genetics, 2011, 20, 202-210.	1.4	258
75	Parkinson's disease age at onset genomeâ€wide association study: Defining heritability, genetic loci, and Î±â€synuclein mechanisms. Movement Disorders, 2019, 34, 866-875.	2.2	258
76	A Deleterious Mutation in DNAJC6 Encoding the Neuronal-Specific Clathrin-Uncoating Co-Chaperone Auxilin, Is Associated with Juvenile Parkinsonism. PLoS ONE, 2012, 7, e36458.	1.1	256
77	Spectrum of SPG4 mutations in autosomal dominant spastic paraplegia. Human Molecular Genetics, 2000, 9, 637-644.	1.4	255
78	A Two-Stage Meta-Analysis Identifies Several New Loci for Parkinson's Disease. PLoS Genetics, 2011, 7, e1002142.	1.5	247
79	Genome-wide Pleiotropy Between Parkinson Disease and Autoimmune Diseases. JAMA Neurology, 2017, 74, 780.	4.5	245
80	Title is missing!. Nature Genetics, 2001, 28, 46-48.	9.4	241
81	Molecular and clinical correlations in autosomal dominant cerebellar ataxia with progressive macular dystrophy (SCA7). Human Molecular Genetics, 1998, 7, 165-170.	1.4	235
82	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, 813-823.	3.7	226
83	Specifically neuropathic Gaucher's mutations accelerate cognitive decline in Parkinson's. Annals of Neurology, 2016, 80, 674-685.	2.8	226
84	Alpha-synuclein and Parkinson's disease. Cellular and Molecular Life Sciences, 2000, 57, 1894-1908.	2.4	225
85	Levodopa-responsive dystonia. Brain, 2000, 123, 1112-1121.	3.7	225
86	Common and rare variant association analyses in amyotrophic lateral sclerosis identify 15 risk loci with distinct genetic architectures and neuron-specific biology. Nature Genetics, 2021, 53, 1636-1648.	9.4	223
87	FXTAS. Neurology, 2012, 79, 1898-1907.	1.5	221
88	Ataxia with oculomotor apraxia type 2: clinical, biological and genotype/phenotype correlation study of a cohort of 90 patients. Brain, 2009, 132, 2688-2698.	3.7	218
89	The p38 subunit of the aminoacyl-tRNA synthetase complex is a Parkin substrate: linking protein biosynthesis and neurodegeneration. Human Molecular Genetics, 2003, 12, 1427-1437.	1.4	217
90	Long-term disease progression in spinocerebellar ataxia types 1, 2, 3, and 6: a longitudinal cohort study. Lancet Neurology, The, 2015, 14, 1101-1108.	4.9	213

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91	Cerebellar ataxia with oculomotor apraxia type 1: clinical and genetic studies. Brain, 2003, 126, 2761-2772.	3.7	212
92	Biochemical analysis of Parkinson's disease-causing variants of Parkin, an E3 ubiquitinâ€“protein ligase with monoubiquitylation capacity. Human Molecular Genetics, 2006, 15, 2059-2075.	1.4	212
93	Autosomal dominant cerebellar ataxia type I in Martinique (French West Indies). Brain, 1995, 118, 1573-1581.	3.7	211
94	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, 1060-1066.	2.6	209
95	Mutations in SPG11 are frequent in autosomal recessive spastic paraplegia with thin corpus callosum, cognitive decline and lower motor neuron degeneration. Brain, 2008, 131, 772-784.	3.7	206
96	Penetrance of Parkinson disease in glucocerebrosidase gene mutation carriers. Neurology, 2012, 78, 417-420.	1.5	203
97	Homozygous deletions in parkin gene in European and North African families with autosomal recessive juvenile parkinsonism. Lancet, The, 1998, 352, 1355-1356.	6.3	199
98	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, 3500-3512.	1.4	198
99	Genome sequencing analysis identifies new loci associated with Lewy body dementia and provides insights into its genetic architecture. Nature Genetics, 2021, 53, 294-303.	9.4	198
100	G2019S LRRK2 mutation in French and North African families with Parkinson's disease. Annals of Neurology, 2005, 58, 784-787.	2.8	196
101	Complex relationship between Parkin mutations and Parkinson disease. American Journal of Medical Genetics Part A, 2002, 114, 584-591.	2.4	193
102	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.	2.6	192
103	De novo mutations in HCN1 cause early infantile epileptic encephalopathy. Nature Genetics, 2014, 46, 640-645.	9.4	192
104	A mutation in periaxin is responsible for CMT4F, an autosomal recessive form of Charcot-Marie-Tooth disease. Human Molecular Genetics, 2001, 10, 415-421.	1.4	188
105	Clinical and molecular advances in autosomal dominant cerebellar ataxias: from genotype to phenotype and physiopathology. European Journal of Human Genetics, 2000, 8, 4-18.	1.4	186
106	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, 266-273.	1.3	185
107	Huntington's disease-like phenotype due to trinucleotide repeat expansions in the TBP and JPH3 genes. Brain, 2003, 126, 1599-1603.	3.7	184
108	Frequency and phenotypic spectrum of ataxia with oculomotor apraxia 2: a clinical and genetic study in 18 patients. Brain, 2004, 127, 759-767.	3.7	184

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109	Unusual phenotypic alteration of Â amyloid precursor protein (ÂAPP) maturation by a new Val-715 -> Met ÂAPP-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, 4119-4124.	3.3	183
110	DNA repair pathways underlie a common genetic mechanism modulating onset in polyglutamine diseases. Annals of Neurology, 2016, 79, 983-990.	2.8	183
111	A Second Locus for Familial Generalized Epilepsy with Febrile Seizures Plus Maps to Chromosome 2q21-q33. American Journal of Human Genetics, 1999, 65, 1078-1085.	2.6	182
112	Clinical, electrophysiological and molecular genetic characteristics of 93 patients with X-linked Charcot-Marie-Tooth disease. Brain, 2001, 124, 1958-1967.	3.7	179
113	Myoclonusâ€“dystonia. Neurology, 2008, 70, 1010-1016.	1.5	179
114	Alteration of Fatty-Acid-Metabolizing Enzymes Affects Mitochondrial Form and Function in Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2012, 91, 1051-1064.	2.6	179
115	Diagnosis of Parkinson's disease on the basis of clinical and genetic classification: a population-based modelling study. Lancet Neurology, The, 2015, 14, 1002-1009.	4.9	179
116	X-linked Charcot-Marie-Tooth disease with connexin 32 mutations. Neurology, 1998, 50, 1074-1082.	1.5	176
117	Using genome-wide complex trait analysis to quantify 'missing heritability' in Parkinson's disease. Human Molecular Genetics, 2012, 21, 4996-5009.	1.4	176
118	The genetic landscape of Parkinson's disease. Revue Neurologique, 2018, 174, 628-643.	0.6	176
119	Longitudinal analysis of impulse control disorders in Parkinson disease. Neurology, 2018, 91, e189-e201.	1.5	175
120	Age at symptom onset and death and disease duration in genetic frontotemporal dementia: an international retrospective cohort study. Lancet Neurology, The, 2020, 19, 145-156.	4.9	175
121	Hereditary spastic paraplegias: an update. Current Opinion in Neurology, 2007, 20, 674-680.	1.8	174
122	Transcriptional repression of p53 by parkin and impairment by mutations associated with autosomal recessive juvenile Parkinson's disease. Nature Cell Biology, 2009, 11, 1370-1375.	4.6	173
123	Convergent genetic and expression data implicate immunity in Alzheimer's disease. Alzheimer's and Dementia, 2015, 11, 658-671.	0.4	173
124	Parkinsonâ€™s disease in GTP cyclohydrolase 1 mutation carriers. Brain, 2014, 137, 2480-2492.	3.7	169
125	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, 229-235.	2.6	166
126	Molecular diagnosis of autosomal dominant early onset Alzheimer's disease: an update. Journal of Medical Genetics, 2005, 42, 793-795.	1.5	165

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127	Spinocerebellar ataxia 17 (SCA17) and Huntingtonâ€™s disease-like 4 (HDL4). Cerebellum, 2008, 7, 170-178.	1.4	164
128	Mutations in the GIGYF2 (TNRC15) Gene at the PARK11 Locus in Familial Parkinson Disease. American Journal of Human Genetics, 2008, 82, 822-833.	2.6	164
129	A Genome-Scale DNA Repair RNAi Screen Identifies SPG48 as a Novel Gene Associated with Hereditary Spastic Paraplegia. PLoS Biology, 2010, 8, e1000408.	2.6	164
130	Spectrum of clinical and electrophysiologic features in HNPP patients with the 17p11.2 deletion. Neurology, 1999, 52, 1440-1440.	1.5	163
131	Progranulin null mutations in both sporadic and familial frontotemporal dementia. Human Mutation, 2007, 28, 846-855.	1.1	162
132	Demographic, neurological and behavioural characteristics and brain perfusion SPECT in frontal variant of frontotemporal dementia. Brain, 2006, 129, 3051-3065.	3.7	158
133	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, 1687-1693.	3.7	157
134	Guadeloupean parkinsonism: a cluster of progressive supranuclear palsyâ€like tauopathy. Brain, 2002, 125, 801-811.	3.7	157
135	Phenotype difference between ALS patients with expanded repeats inC9ORF72and patients with mutations in other ALS-related genes. Journal of Medical Genetics, 2012, 49, 258-263.	1.5	157
136	Akt is altered in an animal model of Huntington's disease and in patients. European Journal of Neuroscience, 2005, 21, 1478-1488.	1.2	156
137	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, 615-627.	1.4	155
138	Gene-Wide Analysis Detects Two New Susceptibility Genes for Alzheimer's Disease. PLoS ONE, 2014, 9, e94661.	1.1	155
139	CAG repeat expansion in the TATA box-binding protein gene causes autosomal dominant cerebellar ataxia. Brain, 2001, 124, 1939-1947.	3.7	154
140	Are interruptedSCA2CAG repeat expansions responsible for parkinsonism?. Neurology, 2007, 69, 1970-1975.	1.5	154
141	Loss of Function of Glucocerebrosidase GBA2 Is Responsible for Motor Neuron Defects in Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2013, 92, 238-244.	2.6	154
142	SQSTM1Mutations in French Patients With Frontotemporal Dementia or Frontotemporal Dementia With Amyotrophic Lateral Sclerosis. JAMA Neurology, 2013, 70, 1403-10.	4.5	153
143	Autosomal dominant cerebellar ataxia type I: oculomotor abnormalities in families with SCA1, SCA2, and SCA3. Journal of Neurology, 1999, 246, 789-797.	1.8	152
144	Implication of the Immune System in Alzheimer's Disease: Evidence from Genome-Wide Pathway Analysis. Journal of Alzheimer's Disease, 2010, 20, 1107-1118.	1.2	152

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145	Brain white matter oedema due to ClC-2 chloride channel deficiency: an observational analytical study. Lancet Neurology, The, 2013, 12, 659-668.	4.9	152
146	The gene for autosomal dominant cerebellar ataxia with pigmentary macular dystrophy maps to chromosome 3p12â€“p21.1. Nature Genetics, 1995, 10, 84-88.	9.4	151
147	Alteration of Ganglioside Biosynthesis Responsible for Complex Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2013, 93, 118-123.	2.6	151
148	Proteomic analysis of parkin knockout mice: alterations in energy metabolism, protein handling and synaptic function. Journal of Neurochemistry, 2005, 95, 1259-1276.	2.1	149
149	Genetic modifiers of risk and age at onset in GBA associated Parkinsonâ€™s disease and Lewy body dementia. Brain, 2020, 143, 234-248.	3.7	149
150	Linkage of a new locus for autosomal dominant familial spastic paraplegia to chromosome 2p. Human Molecular Genetics, 1994, 3, 1569-1573.	1.4	148
151	Spastic paraplegia gene 7 in patients with spasticity and/or optic neuropathy. Brain, 2012, 135, 2980-2993.	3.7	148
152	Genetic Analysis of Inherited Leukodystrophies. JAMA Neurology, 2013, 70, 875.	4.5	147
153	Mutational analysis of the PINK1 gene in early-onset parkinsonism in Europe and North Africa. Brain, 2006, 129, 686-694.	3.7	146
154	Genetic Variants of the Î±-Synuclein Gene SNCA Are Associated with Multiple System Atrophy. PLoS ONE, 2009, 4, e7114.	1.1	144
155	Modulation of the age at onset in spinocerebellar ataxia by CAG tracts in various genes. Brain, 2014, 137, 2444-2455.	3.7	144
156	Myoclonus-dystonia syndrome: Îµ-sarcoglycan mutations and phenotype. Annals of Neurology, 2002, 52, 489-492.	2.8	143
157	C9orf72 repeat expansions are a rare genetic cause of parkinsonism. Brain, 2013, 136, 385-391.	3.7	143
158	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, 171-176.	1.4	141
159	Mutations in KCND3 cause spinocerebellar ataxia type 22. Annals of Neurology, 2012, 72, 859-869.	2.8	138
160	Autosomal recessive cortical myoclonic tremor and epilepsy: association with a mutation in the potassium channel associated gene CNTN2. Brain, 2013, 136, 1155-1160.	3.7	137
161	Ultrastructural PMP22 expression in inherited demyelinating neuropathies. Annals of Neurology, 1996, 39, 813-817.	2.8	136
162	Functional interplay between Parkin and Drp1 in mitochondrial fission and clearance. Biochimica Et Biophysica Acta - Molecular Cell Research, 2014, 1843, 2012-2026.	1.9	134

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163	Screening for Genomic Rearrangements and Methylation Abnormalities of the 15q11-q13 Region in Autism Spectrum Disorders. Biological Psychiatry, 2009, 66, 349-359.	0.7	133
164	Î±-Synuclein Gene Rearrangements in Dominantly Inherited Parkinsonism. Archives of Neurology, 2009, 66, 102.	4.9	133
165	Genetic overlap between Alzheimerâ€™s disease and Parkinsonâ€™s disease at the MAPT locus. Molecular Psychiatry, 2015, 20, 1588-1595.	4.1	133
166	Prediction of cognition in Parkinson's disease with a clinicalâ€“genetic score: a longitudinal analysis of nine cohorts. Lancet Neurology, The, 2017, 16, 620-629.	4.9	131
167	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, 330-332.	2.6	130
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