

# Alexis Brice

## List of Publications by Year in descending order

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830  
papers

95,931  
citations

255

142  
h-index

515

267  
g-index

866  
all docs

866  
docs citations

866  
times ranked

57946  
citing authors

#	ARTICLE	IF	CITATIONS
1	Meta-analysis of 74,046 individuals identifies 11 new susceptibility loci for Alzheimer's disease. <i>Nature Genetics</i> , 2013, 45, 1452-1458.	9.4	3,741
2	Friedreich's Ataxia: Autosomal Recessive Disease Caused by an Intronic GAA Triplet Repeat Expansion. <i>Science</i> , 1996, 271, 1423-1427.	6.0	2,642
3	Mutations in the DJ-1 Gene Associated with Autosomal Recessive Early-Onset Parkinsonism. <i>Science</i> , 2003, 299, 256-259.	6.0	2,467
4	Genetic meta-analysis of diagnosed Alzheimer's disease identifies new risk loci and implicates A $\beta$ , tau, immunity and lipid processing. <i>Nature Genetics</i> , 2019, 51, 414-430.	9.4	1,962
5	Multicenter Analysis of Glucocerebrosidase Mutations in Parkinson's Disease. <i>New England Journal of Medicine</i> , 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. <i>Nature Genetics</i> , 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. <i>Nature Genetics</i> , 2014, 46, 989-993.	9.4	1,685
8	Mutations of the X-linked genes encoding neuroligins NLGN3 and NLGN4 are associated with autism. <i>Nature Genetics</i> , 2003, 34, 27-29.	9.4	1,612
9	Association between Early-Onset Parkinson's Disease and Mutations in the Parkin Gene. <i>New England Journal of Medicine</i> , 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. <i>Lancet Neurology</i> , 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. <i>Lancet Neurology</i> , The, 2008, 7, 583-590.	4.9	1,340
12	APP locus duplication causes autosomal dominant early-onset Alzheimer disease with cerebral amyloid angiopathy. <i>Nature Genetics</i> , 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. <i>Lancet Neurology</i> , The, 2012, 11, 323-330.	4.9	1,039
14	Clinical and Genetic Abnormalities in Patients with Friedreich's Ataxia. <i>New England Journal of Medicine</i> , 1996, 335, 1169-1175.	13.9	1,015
15	Causal relation between $\alpha$ -synuclein locus duplication as a cause of familial Parkinson's disease. <i>Lancet</i> , The, 2004, 364, 1169-1171.	6.3	987
16	Mutations of SCN1A, encoding a neuronal sodium channel, in two families with GEFS+2. <i>Nature Genetics</i> , 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. <i>Nature Genetics</i> , 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. <i>Lancet</i> , The, 2011, 377, 641-649.	6.3	845

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19	Parkinson's disease: from monogenic forms to genetic susceptibility factors. <i>Human Molecular Genetics</i> , 2009, 18, R48-R59.	1.4	816
20	Rare coding variants in <i>PLCG2</i> , <i>ABI3</i> , and <i>TREM2</i> implicate microglial-mediated innate immunity in Alzheimer's disease. <i>Nature Genetics</i> , 2017, 49, 1373-1384.	9.4	783
21	Cloning of the <i>SCA7</i> gene reveals a highly unstable CAG repeat expansion. <i>Nature Genetics</i> , 1997, 17, 65-70.	9.4	758
22	First genetic evidence of GABAA receptor dysfunction in epilepsy: a mutation in the $\beta 2$ -subunit gene. <i>Nature Genetics</i> , 2001, 28, 46-48.	9.4	701
23	Early-Onset Autosomal Dominant Alzheimer Disease: Prevalence, Genetic Heterogeneity, and Mutation Spectrum. <i>American Journal of Human Genetics</i> , 1999, 65, 664-670.	2.6	696
24	Polyglutamine expansion as a pathological epitope in Huntington's disease and four dominant cerebellar ataxias. <i>Nature</i> , 1995, 378, 403-406.	13.7	632
25	G51D $\alpha$ -synuclein mutation causes a novel Parkinsonian "pyramidal syndrome. <i>Annals of Neurology</i> , 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. <i>Nature Genetics</i> , 1999, 23, 296-303.	9.4	575
27	A Wide Variety of Mutations in the <i>Parkin</i> Gene Are Responsible for Autosomal Recessive Parkinsonism in Europe. <i>Human Molecular Genetics</i> , 1999, 8, 567-574.	1.4	571
28	<i>LRRK2</i> G2019S as a Cause of Parkinson's Disease in North African Arabs. <i>New England Journal of Medicine</i> , 2006, 354, 422-423.	13.9	521
29	Genome-wide Analyses Identify <i>KIF5A</i> as a Novel ALS Gene. <i>Neuron</i> , 2018, 97, 1268-1283.e6.	3.8	517
30	What Genetics Tells us About the Causes and Mechanisms of Parkinson's Disease. <i>Physiological Reviews</i> , 2011, 91, 1161-1218.	13.1	515
31	Meta-analysis of <i>SHANK</i> Mutations in Autism Spectrum Disorders: A Gradient of Severity in Cognitive Impairments. <i>PLoS Genetics</i> , 2014, 10, e1004580.	1.5	501
32	Comprehensive Research Synopsis and Systematic Meta-Analyses in Parkinson's Disease Genetics: The PDGene Database. <i>PLoS Genetics</i> , 2012, 8, e1002548.	1.5	495
33	Genome-wide association analyses identify new risk variants and the genetic architecture of amyotrophic lateral sclerosis. <i>Nature Genetics</i> , 2016, 48, 1043-1048.	9.4	494
34	Exome Sequencing Links Corticospinal Motor Neuron Disease to Common Neurodegenerative Disorders. <i>Science</i> , 2014, 343, 506-511.	6.0	466
35	<i>Parkin</i> gene inactivation alters behaviour and dopamine neurotransmission in the mouse. <i>Human Molecular Genetics</i> , 2003, 12, 2277-2291.	1.4	462
36	Senataxin, the ortholog of a yeast RNA helicase, is mutant in ataxia-ocular apraxia 2. <i>Nature Genetics</i> , 2004, 36, 225-227.	9.4	454

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37	Genome-Wide Scan for Autism Susceptibility Genes. <i>Human Molecular Genetics</i> , 1999, 8, 805-812.	1.4	453
38	Spinocerebellar ataxia 3 and machado-joseph disease: Clinical, molecular, and neuropathological features. <i>Annals of Neurology</i> , 1996, 39, 490-499.	2.8	401
39	A mitochondrial origin for frontotemporal dementia and amyotrophic lateral sclerosis through CHCHD10 involvement. <i>Brain</i> , 2014, 137, 2329-2345.	3.7	377
40	A Multicenter Study of Glucocerebrosidase Mutations in Dementia With Lewy Bodies. <i>JAMA Neurology</i> , 2013, 70, 727.	4.5	374
41	Friedreich's ataxia: Point mutations and clinical presentation of compound heterozygotes. <i>Annals of Neurology</i> , 1999, 45, 200-206.	2.8	371
42	DJ-1( PARK7), a novel gene for autosomal recessive, early onset parkinsonism. <i>Neurological Sciences</i> , 2003, 24, 159-160.	0.9	363
43	Parkin prevents mitochondrial swelling and cytochrome c release in mitochondria-dependent cell death. <i>Human Molecular Genetics</i> , 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. <i>American Journal of Human Genetics</i> , 2002, 70, 1328-1332.	2.6	347
45	Spectrin mutations cause spinocerebellar ataxia type 5. <i>Nature Genetics</i> , 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. <i>American Journal of Human Genetics</i> , 2016, 98, 500-513.	2.6	333
47	Phenotype variability in progranulin mutation carriers: a clinical, neuropsychological, imaging and genetic study. <i>Brain</i> , 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)K Akt signalling. <i>Nature Cell Biology</i> , 2006, 8, 834-842.	4.6	325
49	Excessive burden of lysosomal storage disorder gene variants in Parkinson's disease. <i>Brain</i> , 2017, 140, 3191-3203.	3.7	323
50	The natural history of degenerative ataxia: a retrospective study in 466 patients. <i>Brain</i> , 1998, 121, 589-600.	3.7	316
51	Lentiviral vector delivery of parkin prevents dopaminergic degeneration in an $\alpha$ -synuclein rat model of Parkinson's disease. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2004, 101, 17510-17515.	3.3	310
52	Spinocerebellar ataxia type 7 (SCA7): a neurodegenerative disorder with neuronal intranuclear inclusions. <i>Human Molecular Genetics</i> , 1998, 7, 913-918.	1.4	308
53	Mutations in <i>COQ2</i> in Familial and Sporadic Multiple-System Atrophy. <i>New England Journal of Medicine</i> , 2013, 369, 233-244.	13.9	308
54	Sporadic Infantile Epileptic Encephalopathy Caused by Mutations in PCDH19 Resembles Dravet Syndrome but Mainly Affects Females. <i>PLoS Genetics</i> , 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. <i>Nature Genetics</i> , 2007, 39, 366-372.	9.4	303
56	Frontotemporal dementia and its subtypes: a genome-wide association study. <i>Lancet Neurology</i> , The, 2014, 13, 686-699.	4.9	302
57	Chromosomal assignment of the second locus for autosomal dominant cerebellar ataxia (SCA2) to chromosome 12q23-q24.1. <i>Nature Genetics</i> , 1993, 4, 295-299.	9.4	298
58	Association of LRRK2 exonic variants with susceptibility to Parkinson's disease: a case-control study. <i>Lancet Neurology</i> , The, 2011, 10, 898-908.	4.9	294
59	A repeat expansion in the gene encoding junctophilin-3 is associated with Huntington disease-like 2. <i>Nature Genetics</i> , 2001, 29, 377-378.	9.4	288
60	Clinical and pathologic abnormalities in a family with parkinsonism and <i>parkin</i> gene mutations. <i>Neurology</i> , 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. <i>Brain</i> , 1996, 119, 1497-1505.	3.7	285
62	Loss of function of C9orf72 causes motor deficits in a zebrafish model of amyotrophic lateral sclerosis. <i>Annals of Neurology</i> , 2013, 74, 180-187.	2.8	284
63	Parkin mutations are frequent in patients with isolated early-onset parkinsonism. <i>Brain</i> , 2003, 126, 1271-1278.	3.7	279
64	Mutations in voltage-gated potassium channel KCNC3 cause degenerative and developmental central nervous system phenotypes. <i>Nature Genetics</i> , 2006, 38, 447-451.	9.4	278
65	<i>TARDBP</i> mutations in motoneuron disease with frontotemporal lobar degeneration. <i>Annals of Neurology</i> , 2009, 65, 470-473.	2.8	278
66	Clinical Correlations With Lewy Body Pathology in <i>LRRK2</i> -Related Parkinson Disease. <i>JAMA Neurology</i> , 2015, 72, 100.	4.5	272
67	How much phenotypic variation can be attributed to parkin genotype?. <i>Annals of Neurology</i> , 2003, 54, 176-185.	2.8	271
68	Molecular and Clinical Correlations in Spinocerebellar Ataxia 2: A Study of 32 Families. <i>Human Molecular Genetics</i> , 1997, 6, 709-715.	1.4	270
69	Mutations of the presenilin I gene in families with early-onset Alzheimer's disease. <i>Human Molecular Genetics</i> , 1995, 4, 2373-2377.	1.4	268
70	SOD1, ANG, VAPB, TARDBP, and FUS mutations in familial amyotrophic lateral sclerosis: genotype-phenotype correlations. <i>Journal of Medical Genetics</i> , 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. <i>American Journal of Human Genetics</i> , 2003, 72, 1141-1153.	2.6	263
72	Alzheimer's disease associated with mutations in presenilin 2 is rare and variably penetrant. <i>Human Molecular Genetics</i> , 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. <i>Human Molecular Genetics</i> , 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. <i>Human Molecular Genetics</i> , 2011, 20, 202-210.	1.4	258
75	Parkinson's disease age at onset genome-wide association study: Defining heritability, genetic loci, and $\alpha$ -synuclein mechanisms. <i>Movement Disorders</i> , 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. <i>PLoS ONE</i> , 2012, 7, e36458.	1.1	256
77	Spectrum of SPC4 mutations in autosomal dominant spastic paraplegia. <i>Human Molecular Genetics</i> , 2000, 9, 637-644.	1.4	255
78	A Two-Stage Meta-Analysis Identifies Several New Loci for Parkinson's Disease. <i>PLoS Genetics</i> , 2011, 7, e1002142.	1.5	247
79	Genome-wide Pleiotropy Between Parkinson Disease and Autoimmune Diseases. <i>JAMA Neurology</i> , 2017, 74, 780.	4.5	245
80	Title is missing!. <i>Nature Genetics</i> , 2001, 28, 46-48.	9.4	241
81	Molecular and clinical correlations in autosomal dominant cerebellar ataxia with progressive macular dystrophy (SCA7). <i>Human Molecular Genetics</i> , 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. <i>Brain</i> , 1997, 120, 813-823.	3.7	226
83	Specifically neuropathic Gaucher's mutations accelerate cognitive decline in Parkinson's. <i>Annals of Neurology</i> , 2016, 80, 674-685.	2.8	226
84	Alpha-synuclein and Parkinson's disease. <i>Cellular and Molecular Life Sciences</i> , 2000, 57, 1894-1908.	2.4	225
85	Levodopa-responsive dystonia. <i>Brain</i> , 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. <i>Nature Genetics</i> , 2021, 53, 1636-1648.	9.4	223
87	FXTAS. <i>Neurology</i> , 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. <i>Brain</i> , 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. <i>Human Molecular Genetics</i> , 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. <i>Lancet Neurology</i> , The, 2015, 14, 1101-1108.	4.9	213

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91	Cerebellar ataxia with oculomotor apraxia type 1: clinical and genetic studies. <i>Brain</i> , 2003, 126, 2761-2772.	3.7	212
92	Biochemical analysis of Parkinson's disease-causing variants of Parkin, an E3 ubiquitin-protein ligase with monoubiquitylation capacity. <i>Human Molecular Genetics</i> , 2006, 15, 2059-2075.	1.4	212
93	Autosomal dominant cerebellar ataxia type I in Martinique (French West Indies). <i>Brain</i> , 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. <i>American Journal of Human Genetics</i> , 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. <i>Brain</i> , 2008, 131, 772-784.	3.7	206
96	Penetrance of Parkinson disease in glucocerebrosidase gene mutation carriers. <i>Neurology</i> , 2012, 78, 417-420.	1.5	203
97	Homozygous deletions in parkin gene in European and North African families with autosomal recessive juvenile parkinsonism. <i>Lancet, The</i> , 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. <i>Human Molecular Genetics</i> , 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. <i>Nature Genetics</i> , 2021, 53, 294-303.	9.4	198
100	G2019S LRRK2 mutation in French and North African families with Parkinson's disease. <i>Annals of Neurology</i> , 2005, 58, 784-787.	2.8	196
101	Complex relationship between Parkin mutations and Parkinson disease. <i>American Journal of Medical Genetics Part A</i> , 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. <i>American Journal of Human Genetics</i> , 2008, 82, 992-1002.	2.6	192
103	De novo mutations in HCN1 cause early infantile epileptic encephalopathy. <i>Nature Genetics</i> , 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. <i>Human Molecular Genetics</i> , 2001, 10, 415-421.	1.4	188
105	Clinical and molecular advances in autosomal dominant cerebellar ataxias: from genotype to phenotype and physiopathology. <i>European Journal of Human Genetics</i> , 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. <i>Journal of Neuroscience Research</i> , 1989, 23, 266-273.	1.3	185
107	Huntington's disease-like phenotype due to trinucleotide repeat expansions in the TBP and JPH3 genes. <i>Brain</i> , 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. <i>Brain</i> , 2004, 127, 759-767.	3.7	184

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109	Unusual phenotypic alteration of $\beta$ amyloid precursor protein ( $\beta$ AAPP) maturation by a new Val-715 -> Met $\beta$ AAPP-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 Huntington's disease-like 4 (HDL4). <i>Cerebellum</i> , 2008, 7, 170-178.	1.4	164
128	Mutations in the GIGYF2 (TNRC15) Gene at the PARK11 Locus in Familial Parkinson Disease. <i>American Journal of Human Genetics</i> , 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. <i>PLoS Biology</i> , 2010, 8, e1000408.	2.6	164
130	Spectrum of clinical and electrophysiologic features in HNPP patients with the 17p11.2 deletion. <i>Neurology</i> , 1999, 52, 1440-1440.	1.5	163
131	Progranulin null mutations in both sporadic and familial frontotemporal dementia. <i>Human Mutation</i> , 2007, 28, 846-855.	1.1	162
132	Demographic, neurological and behavioural characteristics and brain perfusion SPECT in frontal variant of frontotemporal dementia. <i>Brain</i> , 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. <i>Brain</i> , 1998, 121, 1687-1693.	3.7	157
134	Guadeloupean parkinsonism: a cluster of progressive supranuclear palsy-like tauopathy. <i>Brain</i> , 2002, 125, 801-811.	3.7	157
135	Phenotype difference between ALS patients with expanded repeats in <i>C9ORF72</i> and patients with mutations in other ALS-related genes. <i>Journal of Medical Genetics</i> , 2012, 49, 258-263.	1.5	157
136	Akt is altered in an animal model of Huntington's disease and in patients. <i>European Journal of Neuroscience</i> , 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. <i>Human Molecular Genetics</i> , 2011, 20, 615-627.	1.4	155
138	Gene-Wide Analysis Detects Two New Susceptibility Genes for Alzheimer's Disease. <i>PLoS ONE</i> , 2014, 9, e94661.	1.1	155
139	CAG repeat expansion in the TATA box-binding protein gene causes autosomal dominant cerebellar ataxia. <i>Brain</i> , 2001, 124, 1939-1947.	3.7	154
140	Are interrupted SCA2 CAG repeat expansions responsible for parkinsonism?. <i>Neurology</i> , 2007, 69, 1970-1975.	1.5	154
141	Loss of Function of Glucocerebrosidase GBA2 Is Responsible for Motor Neuron Defects in Hereditary Spastic Paraplegia. <i>American Journal of Human Genetics</i> , 2013, 92, 238-244.	2.6	154
142	<i>SQSTM1</i> Mutations in French Patients With Frontotemporal Dementia or Frontotemporal Dementia With Amyotrophic Lateral Sclerosis. <i>JAMA Neurology</i> , 2013, 70, 1403-10.	4.5	153
143	Autosomal dominant cerebellar ataxia type I: oculomotor abnormalities in families with SCA1, SCA2, and SCA3. <i>Journal of Neurology</i> , 1999, 246, 789-797.	1.8	152
144	Implication of the Immune System in Alzheimer's Disease: Evidence from Genome-Wide Pathway Analysis. <i>Journal of Alzheimer's Disease</i> , 2010, 20, 1107-1118.	1.2	152

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145	Brain white matter oedema due to CIC-2 chloride channel deficiency: an observational analytical study. <i>Lancet Neurology</i> , 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. <i>Nature Genetics</i> , 1995, 10, 84-88.	9.4	151
147	Alteration of Ganglioside Biosynthesis Responsible for Complex Hereditary Spastic Paraplegia. <i>American Journal of Human Genetics</i> , 2013, 93, 118-123.	2.6	151
148	Proteomic analysis of parkin knockout mice: alterations in energy metabolism, protein handling and synaptic function. <i>Journal of Neurochemistry</i> , 2005, 95, 1259-1276.	2.1	149
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