

Christine Van Broeckhoven

List of Publications by Year in descending order

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799
papers

76,635
citations

492
129
h-index

911
241
g-index

872
all docs

872
docs citations

872
times ranked

51248
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.	21.4	3,741
2	Genome-wide association study identifies variants at CLU and PICALM associated with Alzheimer's disease. Nature Genetics, 2009, 41, 1088-1093.	21.4	2,697
3	Genome-wide association study identifies variants at CLU and CR1 associated with Alzheimer's disease. Nature Genetics, 2009, 41, 1094-1099.	21.4	2,155
4	Genetic meta-analysis of diagnosed Alzheimer's disease identifies new risk loci and implicates A β , tau, immunity and lipid processing. Nature Genetics, 2019, 51, 414-430.	21.4	1,962
5	NanoPack: visualizing and processing long-read sequencing data. Bioinformatics, 2018, 34, 2666-2669.	4.1	1,713
6	Common variants at ABCA7, MS4A6A/MS4A4E, EPHA1, CD33 and CD2AP are associated with Alzheimer's disease. Nature Genetics, 2011, 43, 429-435.	21.4	1,708
7	Null mutations in progranulin cause ubiquitin-positive frontotemporal dementia linked to chromosome 17q21. Nature, 2006, 442, 920-924.	27.8	1,386
8	Atherosclerosis, apolipoprotein E, and prevalence of dementia and Alzheimer's disease in the Rotterdam Study. Lancet, The, 1997, 349, 151-154.	13.7	1,304
9	De Novo Mutations in the Sodium-Channel Gene SCN1A Cause Severe Myoclonic Epilepsy of Infancy. American Journal of Human Genetics, 2001, 68, 1327-1332.	6.2	1,111
10	The <i>C9orf72</i> GGGGCC Repeat Is Translated into Aggregating Dipeptide-Repeat Proteins in FTL/ALS. Science, 2013, 339, 1335-1338.	12.6	1,095
11	VEGF is a modifier of amyotrophic lateral sclerosis in mice and humans and protects motoneurons against ischemic death. Nature Genetics, 2003, 34, 383-394.	21.4	794
12	Rare coding variants in PLCG2, ABI3, and TREM2 implicate microglial-mediated innate immunity in Alzheimer's disease. Nature Genetics, 2017, 49, 1373-1384.	21.4	783
13	Mutation of POLG is associated with progressive external ophthalmoplegia characterized by mtDNA deletions. Nature Genetics, 2001, 28, 211-212.	21.4	748
14	Extra-pair paternity results from female preference for high-quality males in the blue tit. Nature, 1992, 357, 494-496.	27.8	720
15	Presenile dementia and cerebral haemorrhage linked to a mutation at codon 692 of the β -amyloid precursor protein gene. Nature Genetics, 1992, 1, 218-221.	21.4	715
16	New insights into the genetic etiology of Alzheimer's disease and related dementias. Nature Genetics, 2022, 54, 412-436.	21.4	700
17	The genetic landscape of Alzheimer disease: clinical implications and perspectives. Genetics in Medicine, 2016, 18, 421-430.	2.4	695
18	TREM2 mutations implicated in neurodegeneration impair cell surface transport and phagocytosis. Science Translational Medicine, 2014, 6, 243ra86.	12.4	600

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19	A C9orf72 promoter repeat expansion in a Flanders-Belgian cohort with disorders of the frontotemporal lobar degeneration-amyotrophic lateral sclerosis spectrum: a gene identification study. <i>Lancet Neurology</i> , The, 2012, 11, 54-65.	10.2	565
20	APOE and Alzheimer disease: a major gene with semi-dominant inheritance. <i>Molecular Psychiatry</i> , 2011, 16, 903-907.	7.9	529
21	TDP-43 transgenic mice develop spastic paralysis and neuronal inclusions characteristic of ALS and frontotemporal lobar degeneration. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2010, 107, 3858-3863.	7.1	491
22	Common variants at 7p21 are associated with frontotemporal lobar degeneration with TDP-43 inclusions. <i>Nature Genetics</i> , 2010, 42, 234-239.	21.4	479
23	Smoking and risk of dementia and Alzheimer's disease in a population-based cohort study: the Rotterdam Study. <i>Lancet</i> , The, 1998, 351, 1840-1843.	13.7	475
24	Collaborative Analysis of Î±-Synuclein Gene Promoter Variability and Parkinson Disease. <i>JAMA - Journal of the American Medical Association</i> , 2006, 296, 661.	7.4	467
25	Apolipoprotein E4 allele in a population-based study of early-onset Alzheimer's disease. <i>Nature Genetics</i> , 1994, 7, 74-78.	21.4	460
26	Amyloid beta protein precursor gene and hereditary cerebral hemorrhage with amyloidosis (Dutch). <i>Science</i> , 1990, 248, 1120-1122.	12.6	445
27	Highly efficient gene delivery by mRNA electroporation in human hematopoietic cells: superiority to lipofection and passive pulsing of mRNA and to electroporation of plasmid cDNA for tumor antigen loading of dendritic cells. <i>Blood</i> , 2001, 98, 49-56.	1.4	438
28	Genetic etiology of Parkinson disease associated with mutations in the SNCA, PARK2, PINK1, PARK7, and LRRK2 genes: a mutation update. <i>Human Mutation</i> , 2010, 31, 763-780.	2.5	428
29	Bidirectional transcripts of the expanded C9orf72 hexanucleotide repeat are translated into aggregating dipeptide repeat proteins. <i>Acta Neuropathologica</i> , 2013, 126, 881-893.	7.7	427
30	Genetic Association of Apolipoprotein E with Age-Related Macular Degeneration. <i>American Journal of Human Genetics</i> , 1998, 63, 200-206.	6.2	425
31	Locus-specific mutation databases for neurodegenerative brain diseases. <i>Human Mutation</i> , 2012, 33, 1340-1344.	2.5	414
32	Molecular genetics of early-onset Alzheimer's disease revisited. <i>Alzheimer's and Dementia</i> , 2016, 12, 733-748.	0.8	409
33	Genetic linkage studies suggest that Alzheimer's disease is not a single homogeneous disorder. <i>Nature</i> , 1990, 347, 194-197.	27.8	407
34	The peripheral myelin protein gene PMP22 is contained within the Charcot-Marie-Tooth disease type 1A duplication. <i>Nature Genetics</i> , 1992, 1, 171-175.	21.4	404
35	Genome Scan Meta-Analysis of Schizophrenia and Bipolar Disorder, Part III: Bipolar Disorder. <i>American Journal of Human Genetics</i> , 2003, 73, 49-62.	6.2	400
36	Estimation of the Genetic Contribution of Presenilin-1 and -2 Mutations in a Population-Based Study of Presenile Alzheimer Disease. <i>Human Molecular Genetics</i> , 1998, 7, 43-51.	2.9	396

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37	Hot-spot residue in small heat-shock protein 22 causes distal motor neuropathy. <i>Nature Genetics</i> , 2004, 36, 597-601.	21.4	395
38	<scp>sTREM</scp> 2 cerebrospinal fluid levels are a potential biomarker for microglia activity in earlyâ€stage Alzheimer's disease and associate with neuronal injury markers. <i>EMBO Molecular Medicine</i> , 2016, 8, 466-476.	6.9	392
39	Risk Estimates of Dementia by Apolipoprotein E Genotypes From a Population-Based Incidence Study: The Rotterdam Study. <i>Archives of Neurology</i> , 1998, 55, 964.	4.5	378
40	A Multicenter Study of Glucocerebrosidase Mutations in Dementia With Lewy Bodies. <i>JAMA Neurology</i> , 2013, 70, 727.	9.0	374
41	Clinical Phenotypes of Different MPZ (P0) Mutations May Include Charcotâ€Marieâ€Tooth Type 1B, Dejerineâ€Sottas, and Congenital Hypomyelination. <i>Neuron</i> , 1996, 17, 451-460.	8.1	372
42	Estimation of the Mutation Frequencies in Charcot-Marie-Tooth Disease Type 1 and Hereditary Neuropathy with Liability to Pressure Palsies: A European Collaborative Study. <i>European Journal of Human Genetics</i> , 1996, 4, 25-33.	2.8	371
43	APP duplication is sufficient to cause early onset Alzheimer's dementia with cerebral amyloid angiopathy. <i>Brain</i> , 2006, 129, 2977-2983.	7.6	337
44	Phenotype variability in progranulin mutation carriers: a clinical, neuropsychological, imaging and genetic study. <i>Brain</i> , 2008, 131, 732-746.	7.6	331
45	The role of tau (MAPT) in frontotemporal dementia and related tauopathies. <i>Human Mutation</i> , 2004, 24, 277-295.	2.5	323
46	Mapping of a gene predisposing to earlyâ€onset Alzheimer's disease to chromosome 14q24.3. <i>Nature Genetics</i> , 1992, 2, 335-339.	21.4	321
47	Increased expression of BIN1 mediates Alzheimer genetic risk by modulating tau pathology. <i>Molecular Psychiatry</i> , 2013, 18, 1225-1234.	7.9	321
48	Genetic insights in Alzheimer's disease. <i>Lancet Neurology</i> , The, 2013, 12, 92-104.	10.2	310
49	Mean age-of-onset of familial alzheimer disease caused by presenilin mutations correlates with both increased A β 42 and decreased A β 40. <i>Human Mutation</i> , 2006, 27, 686-695.	2.5	306
50	hnRNP A3 binds to GGGGCC repeats and is a constituent of p62-positive/TDP43-negative inclusions in the hippocampus of patients with C9orf72 mutations. <i>Acta Neuropathologica</i> , 2013, 125, 413-423.	7.7	302
51	Frontotemporal dementia and its subtypes: a genome-wide association study. <i>Lancet Neurology</i> , The, 2014, 13, 686-699.	10.2	302
52	Association of LRRK2 exonic variants with susceptibility to Parkinson's disease: a caseâ€control study. <i>Lancet Neurology</i> , The, 2011, 10, 898-908.	10.2	294
53	A novel Alzheimer disease locus located near the gene encoding tau protein. <i>Molecular Psychiatry</i> , 2016, 21, 108-117.	7.9	260
54	novoSNP, a novel computational tool for sequence variation discovery. <i>Genome Research</i> , 2005, 15, 436-442.	5.5	254

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55	<i>POLG</i> mutations in neurodegenerative disorders with ataxia but no muscle involvement. <i>Neurology</i> , 2004, 63, 1251-1257.	1.1	252
56	Apolipoprotein E epsilon4 and the risk of dementia with stroke. A population-based investigation. <i>JAMA - Journal of the American Medical Association</i> , 1997, 277, 818-821.	7.4	252
57	A Pan-European Study of the <i>C9orf72</i> Repeat Associated with <i>FTLD</i> : Geographic Prevalence, Genomic Instability, and Intermediate Repeats. <i>Human Mutation</i> , 2013, 34, 363-373.	2.5	247
58	Potent amyloidogenicity and pathogenicity of A β 43. <i>Nature Neuroscience</i> , 2011, 14, 1023-1032.	14.8	245
59	The genetics and neuropathology of frontotemporal lobar degeneration. <i>Acta Neuropathologica</i> , 2012, 124, 353-372.	7.7	242
60	<i>Drosophila</i> screen connects nuclear transport genes to DPR pathology in c9ALS/FTD. <i>Scientific Reports</i> , 2016, 6, 20877.	3.3	239
61	Mutations in <i>DNAJC5</i> , Encoding Cysteine-String Protein Alpha, Cause Autosomal-Dominant Adult-Onset Neuronal Ceroid Lipofuscinosis. <i>American Journal of Human Genetics</i> , 2011, 89, 241-252.	6.2	236
62	Mutations in the peripheral myelin genes and associated genes in inherited peripheral neuropathies. <i>Human Mutation</i> , 1999, 13, 11-28.	2.5	232
63	ALS Genes in the Genomic Era and their Implications for FTD. <i>Trends in Genetics</i> , 2018, 34, 404-423.	6.7	229
64	Mutations in <i>SEPT9</i> cause hereditary neuralgic amyotrophy. <i>Nature Genetics</i> , 2005, 37, 1044-1046.	21.4	222
65	<i>FUS</i> pathology defines the majority of tau- and TDP-43-negative frontotemporal lobar degeneration. <i>Acta Neuropathologica</i> , 2010, 120, 33-41.	7.7	222
66	Recessive <i>POLG</i> mutations presenting with sensory and ataxic neuropathy in compound heterozygote patients with progressive external ophthalmoplegia. <i>Neuromuscular Disorders</i> , 2003, 13, 133-142.	0.6	216
67	The Thr124Met mutation in the peripheral myelin protein zero (MPZ) gene is associated with a clinically distinct Charcot-Marie-Tooth phenotype. <i>Brain</i> , 1999, 122, 281-290.	7.6	215
68	Pathogenesis of polyglutamine disorders: aggregation revisited. <i>Human Molecular Genetics</i> , 2003, 12, R173-R186.	2.9	212
69	A novel presenilin 1 mutation associated with Pick's disease but not β -amyloid plaques. <i>Annals of Neurology</i> , 2004, 55, 617-626.	5.3	210
70	Genetic variation in <i>DPP6</i> is associated with susceptibility to amyotrophic lateral sclerosis. <i>Nature Genetics</i> , 2008, 40, 29-31.	21.4	205
71	Presenilins and Alzheimer disease. <i>Nature Genetics</i> , 1995, 11, 230-232.	21.4	204
72	Germline mosaicism and Duchenne muscular dystrophy mutations. <i>Nature</i> , 1987, 329, 554-556.	27.8	201

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73	The C9orf72 repeat size correlates with onset age of disease, DNA methylation and transcriptional downregulation of the promoter. <i>Molecular Psychiatry</i> , 2016, 21, 1112-1124.	7.9	201
74	Δ5 synuclein promoter confers susceptibility to Parkinson's disease. <i>Annals of Neurology</i> , 2004, 56, 591-595.	5.3	200
75	Genetic contribution of <i>FUS</i> to frontotemporal lobar degeneration. <i>Neurology</i> , 2010, 74, 366-371.	1.1	197
76	Molecular biological characterization of an azole-resistant <i>Candida glabrata</i> isolate. <i>Antimicrobial Agents and Chemotherapy</i> , 1997, 41, 2229-2237.	3.2	196
77	Molecular genetics of Alzheimer's disease: An update. <i>Annals of Medicine</i> , 2008, 40, 562-583.	3.8	196
78	ITPR2 as a susceptibility gene in sporadic amyotrophic lateral sclerosis: a genome-wide association study. <i>Lancet Neurology</i> , The, 2007, 6, 869-877.	10.2	195
79	Serum biomarker for progranulin-associated frontotemporal lobar degeneration. <i>Annals of Neurology</i> , 2009, 65, 603-609.	5.3	195
80	Progress in unraveling the genetic etiology of Parkinson disease in a genomic era. <i>Trends in Genetics</i> , 2015, 31, 140-149.	6.7	193
81	Further evidence that neurofilament light chain gene mutations can cause Charcot-Marie-Tooth disease type 2E. <i>Annals of Neurology</i> , 2001, 49, 245-249.	5.3	188
82	Germinal mosaicism increases the recurrence risk for 'new' Duchenne muscular dystrophy mutations.. <i>Journal of Medical Genetics</i> , 1989, 26, 553-559.	3.2	187
83	Alzheimer risk associated with a copy number variation in the complement receptor 1 increasing C3b/C4b binding sites. <i>Molecular Psychiatry</i> , 2012, 17, 223-233.	7.9	179
84	Pathogenic APP mutations near the gamma-secretase cleavage site differentially affect Aβ secretion and APP C-terminal fragment stability. <i>Human Molecular Genetics</i> , 2001, 10, 1665-1671.	2.9	178
85	De-novo mutation in hereditary motor and sensory neuropathy type I. <i>Lancet</i> , The, 1992, 339, 1081-1082.	13.7	177
86	Promoter Mutations That Increase Amyloid Precursor-Protein Expression Are Associated with Alzheimer Disease. <i>American Journal of Human Genetics</i> , 2006, 78, 936-946.	6.2	173
87	Genetic variation in the KIF1B locus influences susceptibility to multiple sclerosis. <i>Nature Genetics</i> , 2008, 40, 1402-1403.	21.4	173
88	Convergent genetic and expression data implicate immunity in Alzheimer's disease. <i>Alzheimer's and Dementia</i> , 2015, 11, 658-671.	0.8	173
89	Molecular genetic analysis of familial early-onset Alzheimer's disease linked to chromosome 14q24.3. <i>Human Molecular Genetics</i> , 1995, 4, 2363-2371.	2.9	171
90	De novo SCN1A mutations are a major cause of severe myoclonic epilepsy of infancy. <i>Human Mutation</i> , 2003, 21, 615-621.	2.5	170

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91	The presenilin genes: a new gene family involved in Alzheimer disease pathology. Human Molecular Genetics, 1996, 5, 1449-1455.	2.9	169
92	Presenilin mutations in Alzheimer's disease. Human Mutation, 1998, 11, 183-190.	2.5	169
93	Human Meiotic Recombination Products Revealed by Sequencing a Hotspot for Homologous Strand Exchange in Multiple HNPP Deletion Patients. American Journal of Human Genetics, 1998, 62, 1023-1033.	6.2	168
94	Dense-Core Plaques in Tg2576 and PSAPP Mouse Models of Alzheimer's Disease Are Centered on Vessel Walls. American Journal of Pathology, 2005, 167, 527-543.	3.8	168
95	Disruption of endocytic trafficking in frontotemporal dementia with CHMP2B mutations. Human Molecular Genetics, 2010, 19, 2228-2238.	2.9	163
96	Progranulin null mutations in both sporadic and familial frontotemporal dementia. Human Mutation, 2007, 28, 846-855.	2.5	162
97	Genetic variability in <i>progranulin</i> contributes to risk for clinically diagnosed Alzheimer disease. Neurology, 2008, 71, 656-664.	1.1	158
98	Investigating the role of rare heterozygous TREM2 variants in Alzheimer's disease and frontotemporal dementia. Neurobiology of Aging, 2014, 35, 726.e11-726.e19.	3.1	158
99	Flemish and Dutch Mutations in Amyloid β Precursor Protein Have Different Effects on Amyloid β Secretion. Neurobiology of Disease, 1998, 5, 281-286.	4.4	157
100	The molecular basis of the frontotemporal lobar degeneration—amyotrophic lateral sclerosis spectrum. Annals of Medicine, 2012, 44, 817-828.	3.8	157
101	High-density SNP haplotyping suggests altered regulation of tau gene expression in progressive supranuclear palsy. Human Molecular Genetics, 2005, 14, 3281-3292.	2.9	156
102	Sequencing an Ashkenazi reference panel supports population-targeted personal genomics and illuminates Jewish and European origins. Nature Communications, 2014, 5, 4835.	12.8	156
103	Common pathobiochemical hallmarks of progranulin-associated frontotemporal lobar degeneration and neuronal ceroid lipofuscinosis. Acta Neuropathologica, 2014, 127, 845-60.	7.7	156
104	The dopamine D4 receptor gene 48-base-pair-repeat polymorphism and mood disorders: A meta-analysis. Biological Psychiatry, 2005, 57, 999-1003.	1.3	155
105	Gene-Wide Analysis Detects Two New Susceptibility Genes for Alzheimer's Disease. PLoS ONE, 2014, 9, e94661.	2.5	155
106	CAG repeat expansion in the TATA box-binding protein gene causes autosomal dominant cerebellar ataxia. Brain, 2001, 124, 1939-1947.	7.6	154
107	Altered brain white matter integrity in healthy carriers of the <i>APOE</i> ϵ 4 allele. Neurology, 2006, 66, 1029-1033.	1.1	153
108	Meta-analysis of the Association Between Variants in SORL1 and Alzheimer Disease. Archives of Neurology, 2011, 68, 99.	4.5	153

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109	Two divergent types of nerve pathology in patients with different P sub O mutations in Charcot-Marie-Tooth disease. <i>Neurology</i> , 1996, 47, 761-765.	1.1	152
110	Mutations in <i>GDAP1</i> . <i>Neurology</i> , 2002, 59, 1865-1872.	1.1	152
111	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.	21.4	151
112	Current insights into the C9orf72 repeat expansion diseases of the FTL/ALS spectrum. <i>Trends in Neurosciences</i> , 2013, 36, 450-459.	8.6	151
113	Loss of <i>TBK1</i> is a frequent cause of frontotemporal dementia in a Belgian cohort. <i>Neurology</i> , 2015, 85, 2116-2125.	1.1	151
114	Novel missense mutation in the early growth response 2 gene associated with Dejerineâ€“Sottas syndrome phenotype. <i>Neurology</i> , 1999, 52, 1827-1827.	1.1	150
115	Variability of 5-HT2C receptor cys23ser polymorphism among European populations and vulnerability to affective disorder. <i>Molecular Psychiatry</i> , 2001, 6, 579-585.	7.9	150
116	A deletion in <i>SCN1B</i> is associated with febrile seizures and early-onset absence epilepsy. <i>Neurology</i> , 2003, 61, 854-856.	1.1	150
117	Detection of expanded CAG repeats in Bipolar Affective Disorder using the repeat expansion detection (RED) method. <i>Neurobiology of Disease</i> , 1995, 2, 55-62.	4.4	148
118	<i>Progranulin</i> genetic variability contributes to amyotrophic lateral sclerosis. <i>Neurology</i> , 2008, 71, 253-259.	1.1	148
119	Novel POLG mutations in progressive external ophthalmoplegia mimicking mitochondrial neurogastrointestinal encephalomyopathy. <i>European Journal of Human Genetics</i> , 2003, 11, 547-549.	2.8	145
120	A Genomewide Screen for Late-Onset Alzheimer Disease in a Genetically Isolated Dutch Population. <i>American Journal of Human Genetics</i> , 2007, 81, 17-31.	6.2	145
121	Alzheimer and Parkinson Diagnoses in Progranulin Null Mutation Carriers in an Extended Founder Family. <i>Archives of Neurology</i> , 2007, 64, 1436.	4.5	143
122	Structural variants identified by Oxford Nanopore PromethION sequencing of the human genome. <i>Genome Research</i> , 2019, 29, 1178-1187.	5.5	143
123	A novel GABRG2 mutation associated with febrile seizures. <i>Neurology</i> , 2006, 67, 687-690.	1.1	142
124	Early-onset Alzheimer's disease in 2 large Belgian families. <i>Neurology</i> , 1991, 41, 62-62.	1.1	141
125	Common variants in Alzheimer's disease and risk stratification by polygenic risk scores. <i>Nature Communications</i> , 2021, 12, 3417.	12.8	140
126	Glucocorticoid Receptor Gene-Based SNP Analysis in Patients with Recurrent Major Depression. <i>Neuropsychopharmacology</i> , 2006, 31, 620-627.	5.4	139

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127	Current status on Alzheimer disease molecular genetics: from past, to present, to future. Human Molecular Genetics, 2010, 19, R4-R11.	2.9	138
128	Mutational analysis of the MPZ, PMP22 and Cx32 genes in patients of Spanish ancestry with Charcot-Marie-Tooth disease and hereditary neuropathy with liability to pressure palsies. Human Genetics, 1997, 99, 746-754.	3.8	137
129	Variant Alzheimer's disease with spastic paraparesis and cotton wool plaques is caused by PS-1 mutations that lead to exceptionally high amyloid- β concentrations. Annals of Neurology, 2000, 48, 806-808.	5.3	135
130	Nonfibrillar diffuse amyloid deposition due to a gamma42-secretase site mutation points to an essential role for N-truncated Abeta42 in Alzheimer's disease. Human Molecular Genetics, 2000, 9, 2589-2598.	2.9	135
131	Association between COMT (Val158Met) functional polymorphism and early onset in patients with major depressive disorder in a European multicenter genetic association study. Molecular Psychiatry, 2005, 10, 598-605.	7.9	134
132	Reduced functional brain activity response in cognitively intact apolipoprotein E ϵ 4 carriers. Brain, 2006, 129, 1240-1248.	7.6	133
133	Lrrk2 R1441C parkinsonism is clinically similar to sporadic Parkinson disease. Neurology, 2008, 70, 1456-1460.	1.1	132
134	The role of ABCA7 in Alzheimer's disease: evidence from genomics, transcriptomics and methylomics. Acta Neuropathologica, 2019, 138, 201-220.	7.7	132
135	Molecular genetics of Alzheimer's disease. Annals of Medicine, 1998, 30, 560-565.	3.8	131
136	CHMP2B C-truncating mutations in frontotemporal lobar degeneration are associated with an aberrant endosomal phenotype in vitro. Human Molecular Genetics, 2008, 17, 313-322.	2.9	131
137	Origin of the de novo duplication in Charcot-Marie-Tooth disease type 1A: unequal nonsister chromatid exchange during spermatogenesis. Human Molecular Genetics, 1993, 2, 2031-2035.	2.9	130
138	APOE genotype does not modulate age of onset in families with chromosome 14 encoded Alzheimer's disease. Neuroscience Letters, 1994, 169, 179-180.	2.1	130
139	The apolipoprotein E ϵ 2 allele is associated with an increased risk of early-onset Alzheimer's disease and a reduced survival. Annals of Neurology, 1995, 37, 605-610.	5.3	129
140	Granulin mutations associated with frontotemporal lobar degeneration and related disorders: An update. Human Mutation, 2008, 29, 1373-1386.	2.5	126
141	Loss of ALS-associated TDP-43 in zebrafish causes muscle degeneration, vascular dysfunction, and reduced motor neuron axon outgrowth. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 4986-4991.	7.1	126
142	Spinocerebellar ataxia type 7 associated with pigmentary retinal dystrophy. European Journal of Human Genetics, 2004, 12, 2-15.	2.8	124
143	Mutations in ABCA7 in a Belgian cohort of Alzheimer's disease patients: a targeted resequencing study. Lancet Neurology, The, 2015, 14, 814-822.	10.2	124
144	Estimation of the size of the chromosome 17p11.2 duplication in Charcot-Marie-Tooth neuropathy type 1a (CMT1a). HMSN Collaborative Research Group.. Journal of Medical Genetics, 1992, 29, 5-11.	3.2	123

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145	TMEM106B is a genetic modifier of frontotemporal lobar degeneration with C9orf72 hexanucleotide repeat expansions. <i>Acta Neuropathologica</i> , 2014, 127, 407-418.	7.7	123
146	The pursuit of susceptibility genes for Alzheimer's disease: progress and prospects. <i>Trends in Genetics</i> , 2010, 26, 84-93.	6.7	122
147	Pathological mechanisms underlying TDP-43 driven neurodegeneration in FTL-ALS spectrum disorders. <i>Human Molecular Genetics</i> , 2013, 22, R77-R87.	2.9	122
148	Autosomal recessive hereditary motor and sensory neuropathy with focally folded myelin sheaths. <i>Neurology</i> , 1996, 46, 1318-1318.	1.1	121
149	Rescue of Progranulin Deficiency Associated with Frontotemporal Lobar Degeneration by Alkalizing Reagents and Inhibition of Vacuolar ATPase. <i>Journal of Neuroscience</i> , 2011, 31, 1885-1894.	3.6	121
150	Large-scale replication and heterogeneity in Parkinson disease genetic loci. <i>Neurology</i> , 2012, 79, 659-667.	1.1	119
151	A blinded international study on the reliability of genetic testing for GGGGCC-repeat expansions in C9orf72 reveals marked differences in results among 14 laboratories. <i>Journal of Medical Genetics</i> , 2014, 51, 419-424.	3.2	118
152	A Genome Wide Search for Susceptibility Loci in Three European Malignant Hyperthermia Pedigrees. <i>Human Molecular Genetics</i> , 1997, 6, 953-961.	2.9	117
153	Terminal neurogranin is increased in cerebrospinal fluid but unchanged in plasma in Alzheimer's disease. <i>Alzheimer's and Dementia</i> , 2015, 11, 1461-1469.	0.8	117
154	Mutations other than null mutations producing a pathogenic loss of progranulin in frontotemporal dementia. <i>Human Mutation</i> , 2007, 28, 416-416.	2.5	116
155	Transmission and age-at-onset patterns in familial Alzheimer's disease. <i>Neurology</i> , 1990, 40, 395-395.	1.1	115
156	Estrogen use and early onset Alzheimer's disease: a population-based study. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 1999, 67, 779-781.	1.9	114
157	Relationship between C9orf72 repeat size and clinical phenotype. <i>Current Opinion in Genetics and Development</i> , 2017, 44, 117-124.	3.3	114
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