Christine Van Broeckhoven

List of Publications by Citations

785 papers	61,028 citations	122 h-index	220 g-index
870 ext. papers	69,229 ext. citations	8.3 avg, IF	7.18 L-index

#	Paper	IF	Citations
785	Meta-analysis of 74,046 individuals identifies 11 new susceptibility loci for Alzheimer's disease. *Nature Genetics*, **2013**, 45, 1452-8	36.3	2714
784	Genome-wide association study identifies variants at CLU and PICALM associated with Alzheimer's disease. *Nature Genetics*, **2009**, 41, 1088-93	36.3	2018
783	Genome-wide association study identifies variants at CLU and CR1 associated with Alzheimer's disease. *Nature Genetics*, **2009**, 41, 1094-9	36.3	1819
782	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
781	Null mutations in progranulin cause ubiquitin-positive frontotemporal dementia linked to chromosome 17q21. *Nature*, **2006**, 442, 920-4	50.4	1212
780	Atherosclerosis, apolipoprotein E, and prevalence of dementia and Alzheimer's disease in the Rotterdam Study. *Lancet, The*, **1997**, 349, 151-4	40	1142
779	De novo mutations in the sodium-channel gene SCN1A cause severe myoclonic epilepsy of infancy. *American Journal of Human Genetics*, **2001**, 68, 1327-32	11	929
778	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
777	The C9orf72 GGGGCC repeat is translated into aggregating dipeptide-repeat proteins in FTLD/ALS. *Science*, **2013**, 339, 1335-8	33.3	879
776	VEGF is a modifier of amyotrophic lateral sclerosis in mice and humans and protects motoneurons against ischemic death. *Nature Genetics*, **2003**, 34, 383-94	36.3	712
775	Mutation of POLG is associated with progressive external ophthalmoplegia characterized by mtDNA deletions. *Nature Genetics*, **2001**, 28, 211-2	36.3	676
774	Presenile dementia and cerebral haemorrhage linked to a mutation at codon 692 of the beta-amyloid precursor protein gene. *Nature Genetics*, **1992**, 1, 218-21	36.3	652
773	Extra-pair paternity results from female preference for high-quality males in the blue tit. *Nature*, **1992**, 357, 494-496	50.4	618
772	NanoPack: visualizing and processing long-read sequencing data. *Bioinformatics*, **2018**, 34, 2666-2669	7.2	616
771	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
770	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. *Lancet Neurology, The*, **2012**, 11, 54-65	24.1	489
769	The genetic landscape of Alzheimer disease: clinical implications and perspectives. *Genetics in Medicine*, **2016**, 18, 421-30	8.1	477

768	TREM2 mutations implicated in neurodegeneration impair cell surface transport and phagocytosis. *Science Translational Medicine*, **2014**, 6, 243ra86	17.5	436
767	TDP-43 transgenic mice develop spastic paralysis and neuronal inclusions characteristic of ALS and frontotemporal lobar degeneration. *Proceedings of the National Academy of Sciences of the United States of America*, **2010**, 107, 3858-63	11.5	417
766	Apolipoprotein E4 allele in a population-based study of early-onset Alzheimer's disease. *Nature Genetics*, **1994**, 7, 74-8	36.3	413
765	Amyloid beta protein precursor gene and hereditary cerebral hemorrhage with amyloidosis (Dutch). *Science*, **1990**, 248, 1120-2	33.3	411
764	Collaborative analysis of alpha-synuclein gene promoter variability and Parkinson disease. *JAMA - Journal of the American Medical Association*, **2006**, 296, 661-70	27.4	403
763	Smoking and risk of dementia and Alzheimer's disease in a population-based cohort study: the Rotterdam Study. *Lancet, The*, **1998**, 351, 1840-3	40	400
762	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. *Blood*, **2001**, 98, 49-56	2.2	393
761	APOE and Alzheimer disease: a major gene with semi-dominant inheritance. *Molecular Psychiatry*, **2011**, 16, 903-7	15.1	391
760	Genetic linkage studies suggest that Alzheimer's disease is not a single homogeneous disorder. *Nature*, **1990**, 347, 194-7	50.4	371
759	Common variants at 7p21 are associated with frontotemporal lobar degeneration with TDP-43 inclusions. *Nature Genetics*, **2010**, 42, 234-9	36.3	361
758	Hot-spot residue in small heat-shock protein 22 causes distal motor neuropathy. *Nature Genetics*, **2004**, 36, 597-601	36.3	358
757	Genetic association of apolipoprotein E with age-related macular degeneration. *American Journal of Human Genetics*, **1998**, 63, 200-6	11	357
756	Estimation of the genetic contribution of presenilin-1 and -2 mutations in a population-based study of presenile Alzheimer disease. *Human Molecular Genetics*, **1998**, 7, 43-51	5.6	356
755	Genome scan meta-analysis of schizophrenia and bipolar disorder, part III: Bipolar disorder. *American Journal of Human Genetics*, **2003**, 73, 49-62	11	353
754	Locus-specific mutation databases for neurodegenerative brain diseases. *Human Mutation*, **2012**, 33, 1340-4	4.7	349
753	Bidirectional transcripts of the expanded C9orf72 hexanucleotide repeat are translated into aggregating dipeptide repeat proteins. *Acta Neuropathologica*, **2013**, 126, 881-93	14.3	342
752	Genetic etiology of Parkinson disease associated with mutations in the SNCA, PARK2, PINK1, PARK7, and LRRK2 genes: a mutation update. *Human Mutation*, **2010**, 31, 763-80	4.7	341
751	The peripheral myelin protein gene PMP-22 is contained within the Charcot-Marie-Tooth disease type 1A duplication. *Nature Genetics*, **1992**, 1, 171-5	36.3	340

750	Clinical phenotypes of different MPZ (P0) mutations may include Charcot-Marie-Tooth type 1B, Dejerine-Sottas, and congenital hypomyelination. *Neuron*, **1996**, 17, 451-60	13.9	336
749	Risk estimates of dementia by apolipoprotein E genotypes from a population-based incidence study: the Rotterdam Study. *Archives of Neurology*, **1998**, 55, 964-8		324
748	Mapping of a gene predisposing to early-onset Alzheimer's disease to chromosome 14q24.3. *Nature Genetics*, **1992**, 2, 335-9	36.3	291
747	APP duplication is sufficient to cause early onset Alzheimer's dementia with cerebral amyloid angiopathy. *Brain*, **2006**, 129, 2977-83	11.2	286
746	A multicenter study of glucocerebrosidase mutations in dementia with Lewy bodies. *JAMA Neurology*, **2013**, 70, 727-35	17.2	285
745	Estimation of the mutation frequencies in Charcot-Marie-Tooth disease type 1 and hereditary neuropathy with liability to pressure palsies: a European collaborative study. *European Journal of Human Genetics*, **1996**, 4, 25-33	5.3	285
744	Phenotype variability in progranulin mutation carriers: a clinical, neuropsychological, imaging and genetic study. *Brain*, **2008**, 131, 732-46	11.2	275
743	Mean age-of-onset of familial alzheimer disease caused by presenilin mutations correlates with both increased Abeta42 and decreased Abeta40. *Human Mutation*, **2006**, 27, 686-95	4.7	259
742	Molecular genetics of early-onset Alzheimer's disease revisited. *Alzheimerjs and Dementia*, **2016**, 12, 733-48	12	258
741	sTREM2 cerebrospinal fluid levels are a potential biomarker for microglia activity in early-stage Alzheimer's disease and associate with neuronal injury markers. *EMBO Molecular Medicine*, **2016**, 8, 466-76	12	256
740	The role of tau (MAPT) in frontotemporal dementia and related tauopathies. *Human Mutation*, **2004**, 24, 277-95	4.7	255
739	Increased expression of BIN1 mediates Alzheimer genetic risk by modulating tau pathology. *Molecular Psychiatry*, **2013**, 18, 1225-34	15.1	251
738	hnRNP A3 binds to GGGGCC repeats and is a constituent of p62-positive/TDP43-negative inclusions in the hippocampus of patients with C9orf72 mutations. *Acta Neuropathologica*, **2013**, 125, 413-23	14.3	250
737	Genetic insights in Alzheimer's disease. *Lancet Neurology, The*, **2013**, 12, 92-104	24.1	248
736	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
735	POLG mutations in neurodegenerative disorders with ataxia but no muscle involvement. *Neurology*, **2004**, 63, 1251-7	6.5	231
734	Apolipoprotein E epsilon4 and the risk of dementia with stroke. A population-based investigation. *JAMA - Journal of the American Medical Association*, **1997**, 277, 818-21	27.4	225
733	A pan-European study of the C9orf72 repeat associated with FTLD: geographic prevalence, genomic instability, and intermediate repeats. *Human Mutation*, **2013**, 34, 363-73	4.7	208

732	Frontotemporal dementia and its subtypes: a genome-wide association study. *Lancet Neurology, The*, **2014**, 13, 686-99	24.1	207
731	Mutations in the peripheral myelin genes and associated genes in inherited peripheral neuropathies. *Human Mutation*, **1999**, 13, 11-28	4.7	207
730	The genetics and neuropathology of frontotemporal lobar degeneration. *Acta Neuropathologica*, **2012**, 124, 353-72	14.3	206
729	Potent amyloidogenicity and pathogenicity of Aβ43. *Nature Neuroscience*, **2011**, 14, 1023-32	25.5	206
728	FUS pathology defines the majority of tau- and TDP-43-negative frontotemporal lobar degeneration. *Acta Neuropathologica*, **2010**, 120, 33-41	14.3	198
727	novoSNP, a novel computational tool for sequence variation discovery. *Genome Research*, **2005**, 15, 436-42	9.7	198
726	The Thr124Met mutation in the peripheral myelin protein zero (MPZ) gene is associated with a clinically distinct Charcot-Marie-Tooth phenotype. *Brain*, **1999**, 122 (Pt 2), 281-90	11.2	194
725	Mutations in DNAJC5, encoding cysteine-string protein alpha, cause autosomal-dominant adult-onset neuronal ceroid lipofuscinosis. *American Journal of Human Genetics*, **2011**, 89, 241-52	11	192
724	Pathogenesis of polyglutamine disorders: aggregation revisited. *Human Molecular Genetics*, **2003**, 12 Spec No 2, R173-86	5.6	189
723	Germline mosaicism and Duchenne muscular dystrophy mutations. *Nature*, **1987**, 329, 554-6	50.4	187
722	Serum biomarker for progranulin-associated frontotemporal lobar degeneration. *Annals of Neurology*, **2009**, 65, 603-9	9.4	185
721	Mutations in SEPT9 cause hereditary neuralgic amyotrophy. *Nature Genetics*, **2005**, 37, 1044-6	36.3	184
720	alpha-Synuclein promoter confers susceptibility to Parkinson's disease. *Annals of Neurology*, **2004**, 56, 591-5	9.4	182
719	A novel presenilin 1 mutation associated with Pick's disease but not beta-amyloid plaques. *Annals of Neurology*, **2004**, 55, 617-26	9.4	181
718	Drosophila screen connects nuclear transport genes to DPR pathology in c9ALS/FTD. *Scientific Reports*, **2016**, 6, 20877	4.9	179
717	Recessive POLG mutations presenting with sensory and ataxic neuropathy in compound heterozygote patients with progressive external ophthalmoplegia. *Neuromuscular Disorders*, **2003**, 13, 133-42	2.9	179
716	Genetic variation in DPP6 is associated with susceptibility to amyotrophic lateral sclerosis. *Nature Genetics*, **2008**, 40, 29-31	36.3	177
715	A novel Alzheimer disease locus located near the gene encoding tau protein. *Molecular Psychiatry*, **2016**, 21, 108-17	15.1	175

714	Molecular genetics of Alzheimer's disease: an update. *Annals of Medicine*, **2008**, 40, 562-83	1.5	175
713	Further evidence that neurofilament light chain gene mutations can cause Charcot-Marie-Tooth disease type 2E. *Annals of Neurology*, **2001**, 49, 245-9	9.4	175
712	Genetic contribution of FUS to frontotemporal lobar degeneration. *Neurology*, **2010**, 74, 366-71	6.5	172
711	Molecular biological characterization of an azole-resistant Candida glabrata isolate. *Antimicrobial Agents and Chemotherapy*, **1997**, 41, 2229-37	5.9	172
710	ITPR2 as a susceptibility gene in sporadic amyotrophic lateral sclerosis: a genome-wide association study. *Lancet Neurology, The*, **2007**, 6, 869-77	24.1	168
709	De-novo mutation in hereditary motor and sensory neuropathy type I. *Lancet, The*, **1992**, 339, 1081-2	40	167
708	Germinal mosaicism increases the recurrence risk for 'new' Duchenne muscular dystrophy mutations. *Journal of Medical Genetics*, **1989**, 26, 553-9	5.8	163
707	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, 1023-33	11	155
706	The C9orf72 repeat size correlates with onset age of disease, DNA methylation and transcriptional downregulation of the promoter. *Molecular Psychiatry*, **2016**, 21, 1112-24	15.1	154
705	Promoter mutations that increase amyloid precursor-protein expression are associated with Alzheimer disease. *American Journal of Human Genetics*, **2006**, 78, 936-46	11	154
704	Molecular genetic analysis of familial early-onset Alzheimer's disease linked to chromosome 14q24.3. *Human Molecular Genetics*, **1995**, 4, 2363-71	5.6	152
703	Genetic variation in the KIF1B locus influences susceptibility to multiple sclerosis. *Nature Genetics*, **2008**, 40, 1402-3	36.3	150
702	Progress in unraveling the genetic etiology of Parkinson disease in a genomic era. *Trends in Genetics*, **2015**, 31, 140-9	8.5	149
701	ALS Genes in the Genomic Era and their Implications for FTD. *Trends in Genetics*, **2018**, 34, 404-423	8.5	149
700	Alzheimer risk associated with a copy number variation in the complement receptor 1 increasing C3b/C4b binding sites. *Molecular Psychiatry*, **2012**, 17, 223-33	15.1	149
699	Dense-core plaques in Tg2576 and PSAPP mouse models of Alzheimer's disease are centered on vessel walls. *American Journal of Pathology*, **2005**, 167, 527-43	5.8	149
698	The presenilin genes: a new gene family involved in Alzheimer disease pathology. *Human Molecular Genetics*, **1996**, 5 Spec No, 1449-55	5.6	148
697	Convergent genetic and expression data implicate immunity in Alzheimer's disease. *Alzheimerjs and Dementia*, **2015**, 11, 658-71	1.2	146

696	Pathogenic APP mutations near the gamma-secretase cleavage site differentially affect Abeta secretion and APP C-terminal fragment stability. *Human Molecular Genetics*, **2001**, 10, 1665-71	5.6	145
695	High-density SNP haplotyping suggests altered regulation of tau gene expression in progressive supranuclear palsy. *Human Molecular Genetics*, **2005**, 14, 3281-92	5.6	144
694	Progranulin null mutations in both sporadic and familial frontotemporal dementia. *Human Mutation* , **2007**, 28, 846-55	4.7	143
693	Novel missense mutation in the early growth response 2 gene associated with Dejerine-Sottas syndrome phenotype. *Neurology*, **1999**, 52, 1827-32	6.5	142
692	The molecular basis of the frontotemporal lobar degeneration-amyotrophic lateral sclerosis spectrum. *Annals of Medicine*, **2012**, 44, 817-28	1.5	141
691	Disruption of endocytic trafficking in frontotemporal dementia with CHMP2B mutations. *Human Molecular Genetics*, **2010**, 19, 2228-38	5.6	140
690	Presenilin mutations in Alzheimer's disease. *Human Mutation*, **1998**, 11, 183-90	4.7	139
689	Genetic variability in progranulin contributes to risk for clinically diagnosed Alzheimer disease. *Neurology*, **2008**, 71, 656-64	6.5	139
688	Altered brain white matter integrity in healthy carriers of the APOE epsilon4 allele: a risk for AD?. *Neurology*, **2006**, 66, 1029-33	6.5	139
687	Variability of 5-HT2C receptor cys23ser polymorphism among European populations and vulnerability to affective disorder. *Molecular Psychiatry*, **2001**, 6, 579-85	15.1	138
686	Flemish and Dutch mutations in amyloid beta precursor protein have different effects on amyloid beta secretion. *Neurobiology of Disease*, **1998**, 5, 281-6	7.5	138
685	The dopamine D4 receptor gene 48-base-pair-repeat polymorphism and mood disorders: a meta-analysis. *Biological Psychiatry*, **2005**, 57, 999-1003	7.9	137
684	De novo SCN1A mutations are a major cause of severe myoclonic epilepsy of infancy. *Human Mutation*, **2003**, 21, 615-21	4.7	137
683	Two divergent types of nerve pathology in patients with different P0 mutations in Charcot-Marie-Tooth disease. *Neurology*, **1996**, 47, 761-5	6.5	137
682	A deletion in SCN1B is associated with febrile seizures and early-onset absence epilepsy. *Neurology*, **2003**, 61, 854-6	6.5	136
681	Common pathobiochemical hallmarks of progranulin-associated frontotemporal lobar degeneration and neuronal ceroid lipofuscinosis. *Acta Neuropathologica*, **2014**, 127, 845-60	14.3	135
680	Meta-analysis of the association between variants in SORL1 and Alzheimer disease. *Archives of Neurology*, **2011**, 68, 99-106		135
679	Progranulin genetic variability contributes to amyotrophic lateral sclerosis. *Neurology*, **2008**, 71, 253-9	6.5	135

678	Detection of expanded CAG repeats in bipolar affective disorder using the repeat expansion detection (RED) method. *Neurobiology of Disease*, **1995**, 2, 55-62	7.5	135
677	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
676	Early-onset Alzheimer's disease in 2 large Belgian families. *Neurology*, **1991**, 41, 62-8	6.5	132
675	Investigating the role of rare heterozygous TREM2 variants in Alzheimer's disease and frontotemporal dementia. *Neurobiology of Aging*, **2014**, 35, 726.e11-9	5.6	131
674	Novel POLG mutations in progressive external ophthalmoplegia mimicking mitochondrial neurogastrointestinal encephalomyopathy. *European Journal of Human Genetics*, **2003**, 11, 547-9	5.3	131
673	Mutations in GDAP1: autosomal recessive CMT with demyelination and axonopathy. *Neurology*, **2002**, 59, 1865-72	6.5	130
672	Current insights into the C9orf72 repeat expansion diseases of the FTLD/ALS spectrum. *Trends in Neurosciences*, **2013**, 36, 450-9	13.3	129
671	Glucocorticoid receptor gene-based SNP analysis in patients with recurrent major depression. *Neuropsychopharmacology*, **2006**, 31, 620-7	8.7	129
670	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, 746-54	6.3	128
669	Current status on Alzheimer disease molecular genetics: from past, to present, to future. *Human Molecular Genetics*, **2010**, 19, R4-R11	5.6	125
668	A novel GABRG2 mutation associated with febrile seizures. *Neurology*, **2006**, 67, 687-90	6.5	125
667	Alzheimer and Parkinson diagnoses in progranulin null mutation carriers in an extended founder family. *Archives of Neurology*, **2007**, 64, 1436-46		124
666	A genomewide screen for late-onset Alzheimer disease in a genetically isolated Dutch population. *American Journal of Human Genetics*, **2007**, 81, 17-31	11	124
665	Reduced functional brain activity response in cognitively intact apolipoprotein E epsilon4 carriers. *Brain*, **2006**, 129, 1240-8	11.2	122
664	Origin of the de novo duplication in Charcot-Marie-Tooth disease type 1A: unequal nonsister chromatid exchange during spermatogenesis. *Human Molecular Genetics*, **1993**, 2, 2031-5	5.6	120
663	Loss of TBK1 is a frequent cause of frontotemporal dementia in a Belgian cohort. *Neurology*, **2015**, 85, 2116-25	6.5	119
662	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	15.1	119
661	CAG repeat expansion in the TATA box-binding protein gene causes autosomal dominant cerebellar ataxia. *Brain*, **2001**, 124, 1939-47	11.2	119

660	APOE genotype does not modulate age of onset in families with chromosome 14 encoded Alzheimer's disease. *Neuroscience Letters*, **1994**, 169, 179-80	3.3	119
659	Granulin mutations associated with frontotemporal lobar degeneration and related disorders: an update. *Human Mutation*, **2008**, 29, 1373-86	4.7	116
658	Sequencing an Ashkenazi reference panel supports population-targeted personal genomics and illuminates Jewish and European origins. *Nature Communications*, **2014**, 5, 4835	17.4	115
657	The apolipoprotein E epsilon 2 allele is associated with an increased risk of early-onset Alzheimer's disease and a reduced survival. *Annals of Neurology*, **1995**, 37, 605-10	9.4	115
656	Variant Alzheimer's disease with spastic paraparesis and cotton wool plaques is caused by PS-1 mutations that lead to exceptionally high amyloid-□ concentrations. *Annals of Neurology*, **2000**, 48, 806-808	9.4	113
655	CHMP2B C-truncating mutations in frontotemporal lobar degeneration are associated with an aberrant endosomal phenotype in vitro. *Human Molecular Genetics*, **2008**, 17, 313-22	5.6	112
654	Nonfibrillar diffuse amyloid deposition due to a gamma(42)-secretase site mutation points to an essential role for N-truncated A beta(42) in Alzheimer's disease. *Human Molecular Genetics*, **2000**, 9, 2589-98	5.6	112
653	Pathological mechanisms underlying TDP-43 driven neurodegeneration in FTLD-ALS spectrum disorders. *Human Molecular Genetics*, **2013**, 22, R77-87	5.6	111
652	Molecular genetics of Alzheimer's disease. *Annals of Medicine*, **1998**, 30, 560-5	1.5	111
651	Rescue of progranulin deficiency associated with frontotemporal lobar degeneration by alkalizing reagents and inhibition of vacuolar ATPase. *Journal of Neuroscience*, **2011**, 31, 1885-94	6.6	109
650	Lrrk2 R1441C parkinsonism is clinically similar to sporadic Parkinson disease. *Neurology*, **2008**, 70, 1456-60	6.5	109
649	Large-scale replication and heterogeneity in Parkinson disease genetic loci. *Neurology*, **2012**, 79, 659-67	6.5	106
648	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	5.8	105
647	Reduced hippocampal volume in non-demented carriers of the apolipoprotein E epsilon4: relation to chronological age and recognition memory. *Neuroscience Letters*, **2006**, 396, 23-7	3.3	104
646	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, 4986-91	11.5	103
645	Loss of progranulin function in frontotemporal lobar degeneration. *Trends in Genetics*, **2008**, 24, 186-94	8.5	101
644	Spinocerebellar ataxia type 7 associated with pigmentary retinal dystrophy. *European Journal of Human Genetics*, **2004**, 12, 2-15	5.3	101
643	Rapid screening of myelin genes in CMT1 patients by SSCP analysis: identification of new mutations and polymorphisms in the P0 gene. *Human Genetics*, **1994**, 94, 653-7	6.3	101

642	Autosomal recessive hereditary motor and sensory neuropathy with focally folded myelin sheaths: clinical, electrophysiologic, and genetic aspects of a large family. *Neurology*, **1996**, 46, 1318-24	6.5	100
641	The pursuit of susceptibility genes for Alzheimer's disease: progress and prospects. *Trends in Genetics*, **2010**, 26, 84-93	8.5	98
640	Tau negative frontal lobe dementia at 17q21: significant finemapping of the candidate region to a 4.8 cM interval. *Molecular Psychiatry*, **2002**, 7, 1064-74	15.1	98
639	TMEM106B is a genetic modifier of frontotemporal lobar degeneration with C9orf72 hexanucleotide repeat expansions. *Acta Neuropathologica*, **2014**, 127, 407-18	14.3	97
638	Genetic variability in the mitochondrial serine protease HTRA2 contributes to risk for Parkinson disease. *Human Mutation*, **2008**, 29, 832-40	4.7	97
637	Twinkle and POLG defects enhance age-dependent accumulation of mutations in the control region of mtDNA. *Nucleic Acids Research*, **2004**, 32, 3053-64	20.1	97
636	A blinded international study on the reliability of genetic testing for GGGGCC-repeat expansions in C9orf72 reveals marked differences in results among 14 laboratories. *Journal of Medical Genetics*, **2014**, 51, 419-24	5.8	96
635	A genome wide search for susceptibility loci in three European malignant hyperthermia pedigrees. *Human Molecular Genetics*, **1997**, 6, 953-61	5.6	96
634	WT1 mutation in malignant mesothelioma and WT1 immunoreactivity in relation to p53 and growth factor receptor expression, cell-type transition, and prognosis. *Journal of Pathology*, **1997**, 181, 67-74	9.4	96
633	Nonviral transfection of distinct types of human dendritic cells: high-efficiency gene transfer by electroporation into hematopoietic progenitor- but not monocyte-derived dendritic cells. *Gene Therapy*, **1998**, 5, 700-7	4	96
632	The influence of APOE status on episodic and semantic memory: data from a population-based study. *Neuropsychology*, **2006**, 20, 645-57	3.8	96
631	Mutations in ABCA7 in a Belgian cohort of Alzheimer's disease patients: a targeted resequencing study. *Lancet Neurology, The*, **2015**, 14, 814-822	24.1	95
630	C-terminal neurogranin is increased in cerebrospinal fluid but unchanged in plasma in Alzheimer's disease. *Alzheimerjs and Dementia*, **2015**, 11, 1461-1469	1.2	95
629	No association of CSF biomarkers with APOEepsilon4, plaque and tangle burden in definite Alzheimer's disease. *Brain*, **2007**, 130, 2320-6	11.2	95
628	Behavioral disturbances without amyloid deposits in mice overexpressing human amyloid precursor protein with Flemish (A692G) or Dutch (E693Q) mutation. *Neurobiology of Disease*, **2000**, 7, 9-22	7.5	94
627	Genetic and clinical features of progranulin-associated frontotemporal lobar degeneration. *Archives of Neurology*, **2011**, 68, 488-97		93
626	Cellular ageing, increased mortality and FTLD-TDP-associated neuropathology in progranulin knockout mice. *Journal of Pathology*, **2012**, 228, 67-76	9.4	92
625	Dense-core senile plaques in the Flemish variant of Alzheimer's disease are vasocentric. *American Journal of Pathology*, **2002**, 161, 507-20	5.8	92

624	Parental origin and germline mosaicism of deletions and duplications of the dystrophin gene: a European study. *Human Genetics*, **1992**, 88, 249-57	6.3	92
623	Mutations other than null mutations producing a pathogenic loss of progranulin in frontotemporal dementia. *Human Mutation*, **2007**, 28, 416	4.7	91
622	Amyloid precursor protein gene mutation in early-onset Alzheimer's disease. *Lancet, The*, **1991**, 337, 978	40	91
621	TMEM106B is associated with frontotemporal lobar degeneration in a clinically diagnosed patient cohort. *Brain*, **2011**, 134, 808-15	11.2	90
620	PRNP Val129 homozygosity increases risk for early-onset Alzheimer's disease. *Annals of Neurology*, **2003**, 53, 409-12	9.4	90
619	Genetics of personality: are we making progress?. *Molecular Psychiatry*, **2003**, 8, 840-52	15.1	90
618	Estrogen use and early onset Alzheimer's disease: a population-based study. *Journal of Neurology, Neurosurgery and Psychiatry*, **1999**, 67, 779-81	5.5	90
617	Gene-wide analysis detects two new susceptibility genes for Alzheimer's disease. *PLoS ONE*, **2014**, 9, e94661	3.7	90
616	Belgian Fabry study: prevalence of Fabry disease in a cohort of 1000 young patients with cerebrovascular disease. *Stroke*, **2010**, 41, 863-8	6.7	88
615	Association of brain-specific tryptophan hydroxylase, TPH2, with unipolar and bipolar disorder in a Northern Swedish, isolated population. *Archives of General Psychiatry*, **2006**, 63, 1103-10		88
614	A major SNP haplotype of the arginine vasopressin 1B receptor protects against recurrent major depression. *Molecular Psychiatry*, **2004**, 9, 287-92	15.1	88
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