Peter H St George-Hyslop

List of Publications by Citations

268 papers	29,787 citations	78 h-index	168 g-index
282 ext. papers	34,559 ext. citations	10.3 avg, IF	6.06 L-index

#	Paper	IF	Citations
268	Meta-analysis of 74,046 individuals identifies 11 new susceptibility loci for Alzheimer's disease. *Nature Genetics*, **2013**, 45, 1452-8	36.3	2714
267	TREM2 variants in Alzheimer's disease. *New England Journal of Medicine*, **2013**, 368, 117-27	59.2	1805
266	Common variants at MS4A4/MS4A6E, CD2AP, CD33 and EPHA1 are associated with late-onset Alzheimer's disease. *Nature Genetics*, **2011**, 43, 436-41	36.3	1367
265	A beta peptide immunization reduces behavioural impairment and plaques in a model of Alzheimer's disease. *Nature*, **2000**, 408, 979-82	50.4	1308
264	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
263	The neuronal sortilin-related receptor SORL1 is genetically associated with Alzheimer disease. *Nature Genetics*, **2007**, 39, 168-77	36.3	888
262	Nicastrin modulates presenilin-mediated notch/glp-1 signal transduction and betaAPP processing. *Nature*, **2000**, 407, 48-54	50.4	829
261	Early-onset amyloid deposition and cognitive deficits in transgenic mice expressing a double mutant form of amyloid precursor protein 695. *Journal of Biological Chemistry*, **2001**, 276, 21562-70	5.4	691
260	Analysis of shared heritability in common disorders of the brain. *Science*, **2018**, 360,	33.3	666
259	Functional variants of OCTN cation transporter genes are associated with Crohn disease. *Nature Genetics*, **2004**, 36, 471-5	36.3	661
258	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
257	ALS/FTD Mutation-Induced Phase Transition of FUS Liquid Droplets and Reversible Hydrogels into Irreversible Hydrogels Impairs RNP Granule Function. *Neuron*, **2015**, 88, 678-90	13.9	503
256	Presenilin-1 mutations downregulate the signalling pathway of the unfolded-protein response. *Nature Cell Biology*, **1999**, 1, 479-85	23.4	463
255	gamma-Secretase, Notch, Abeta and Alzheimer's disease: where do the presenilins fit in?. *Nature Reviews Neuroscience*, **2002**, 3, 281-90	13.5	461
254	FUS Phase Separation Is Modulated by a Molecular Chaperone and Methylation of Arginine Cation- Interactions. *Cell*, **2018**, 173, 720-734.e15	56.2	409
253	Candidate single-nucleotide polymorphisms from a genomewide association study of Alzheimer disease. *Archives of Neurology*, **2008**, 65, 45-53		362
252	Rare coding variants in the phospholipase D3 gene confer risk for Alzheimer's disease. *Nature*, **2014** , 505, 550-554	50.4	345

251	Reversal of autophagy dysfunction in the TgCRND8 mouse model of Alzheimer's disease ameliorates amyloid pathologies and memory deficits. *Brain*, **2011**, 134, 258-77	11.2	345
250	Meta-analysis confirms CR1, CLU, and PICALM as alzheimer disease risk loci and reveals interactions with APOE genotypes. *Archives of Neurology*, **2010**, 67, 1473-84		330
249	The presenilin 1 protein is a component of a high molecular weight intracellular complex that contains beta-catenin. *Journal of Biological Chemistry*, **1998**, 273, 16470-5	5.4	326
248	Cyclohexanehexol inhibitors of Abeta aggregation prevent and reverse Alzheimer phenotype in a mouse model. *Nature Medicine*, **2006**, 12, 801-8	50.5	306
247	Presenilin proteins undergo heterogeneous endoproteolysis between Thr291 and Ala299 and occur as stable N- and C-terminal fragments in normal and Alzheimer brain tissue. *Neurobiology of Disease* , **1997**, 3, 325-37	7.5	288
246	Presenilin 1 controls gamma-secretase processing of amyloid precursor protein in pre-golgi compartments of hippocampal neurons. *Journal of Cell Biology*, **1999**, 147, 277-94	7.3	285
245	TMP21 is a presenilin complex component that modulates gamma-secretase but not epsilon-secretase activity. *Nature*, **2006**, 440, 1208-12	50.4	260
244	Wild-type PINK1 prevents basal and induced neuronal apoptosis, a protective effect abrogated by Parkinson disease-related mutations. *Journal of Biological Chemistry*, **2005**, 280, 34025-32	5.4	249
243	Molecular genetics of Alzheimer's disease. *Biological Psychiatry*, **2000**, 47, 183-99	7.9	249
242	Phosphorylation, subcellular localization, and membrane orientation of the Alzheimer's disease-associated presenilins. *Journal of Biological Chemistry*, **1997**, 272, 3590-8	5.4	234
241	Frontotemporal dementia and its subtypes: a genome-wide association study. *Lancet Neurology, The*, **2014**, 13, 686-99	24.1	207
240	Microbleed topography, leukoaraiosis, and cognition in probable Alzheimer disease from the Sunnybrook dementia study. *Archives of Neurology*, **2008**, 65, 790-5		205
239	In vivo reduction of amyloid-beta by a mutant copper transporter. *Proceedings of the National Academy of Sciences of the United States of America*, **2003**, 100, 14193-8	11.5	204
238	Early-onset Parkinson's disease caused by a compound heterozygous DJ-1 mutation. *Annals of Neurology*, **2003**, 54, 271-4	9.4	202
237	SPATACSIN mutations cause autosomal recessive juvenile amyotrophic lateral sclerosis. *Brain*, **2010** , 133, 591-8	11.2	184
236	Defective membrane interactions of familial Parkinson's disease mutant A30P alpha-synuclein. *Journal of Molecular Biology*, **2002**, 315, 799-807	6.5	178
235	Novel splicing mutation in the progranulin gene causing familial corticobasal syndrome. *Brain*, **2006** , 129, 3115-23	11.2	162
234	RNA Granules Hitchhike on Lysosomes for Long-Distance Transport, Using Annexin A11 as a Molecular Tether. *Cell*, **2019**, 179, 147-164.e20	56.2	158

233	Genetic analysis implicates APOE, SNCA and suggests lysosomal dysfunction in the etiology of dementia with Lewy bodies. *Human Molecular Genetics*, **2014**, 23, 6139-46	5.6	152
232	The genetics of adult-onset neuropsychiatric disease: complexities and conundra?. *Science*, **2003**, 302, 822-6	33.3	147
231	Convergent genetic and expression data implicate immunity in Alzheimer's disease. *Alzheimer's and Dementia*, **2015**, 11, 658-71	1.2	146
230	Presenilin-dependent gamma-secretase-mediated control of p53-associated cell death in Alzheimer's disease. *Journal of Neuroscience*, **2006**, 26, 6377-85	6.6	143
229	Immunotherapy for Alzheimer's disease. *Proceedings of the National Academy of Sciences of the United States of America*, **2004**, 101 Suppl 2, 14657-62	11.5	142
228	C-terminal calcium binding of α-synuclein modulates synaptic vesicle interaction. *Nature Communications*, **2018**, 9, 712	17.4	140
227	Disturbed activation of endoplasmic reticulum stress transducers by familial Alzheimer's disease-linked presenilin-1 mutations. *Journal of Biological Chemistry*, **2001**, 276, 43446-54	5.4	139
226	Molecular biology and genetics of Alzheimer's disease. *Comptes Rendus - Biologies*, **2005**, 328, 119-30	1.4	136
225	Identification of novel loci for Alzheimer disease and replication of CLU, PICALM, and BIN1 in Caribbean Hispanic individuals. *Archives of Neurology*, **2011**, 68, 320-8		135
224	Meta-analysis of the association between variants in SORL1 and Alzheimer disease. *Archives of Neurology*, **2011**, 68, 99-106		135
223	APH-1 interacts with mature and immature forms of presenilins and nicastrin and may play a role in maturation of presenilin.nicastrin complexes. *Journal of Biological Chemistry*, **2003**, 278, 7374-80	5.4	133
222	Bilineal disease and trans-heterozygotes in autosomal dominant polycystic kidney disease. *American Journal of Human Genetics*, **2001**, 68, 355-63	11	132
221	Analysis of the PINK1 gene in a large cohort of cases with Parkinson disease. *Archives of Neurology*, **2004**, 61, 1898-904		131
220	Effects of multiple genetic loci on age at onset in late-onset Alzheimer disease: a genome-wide association study. *JAMA Neurology*, **2014**, 71, 1394-404	17.2	129
219	Mature glycosylation and trafficking of nicastrin modulate its binding to presenilins. *Journal of Biological Chemistry*, **2002**, 277, 28135-42	5.4	129
218	The in vivo brain interactome of the amyloid precursor protein. *Molecular and Cellular Proteomics*, **2008**, 7, 15-34	7.6	126
217	Coding mutations in SORL1 and Alzheimer disease. *Annals of Neurology*, **2015**, 77, 215-27	9.4	125
216	SORL1 is genetically associated with late-onset Alzheimer's disease in Japanese, Koreans and Caucasians. *PLoS ONE*, **2013**, 8, e58618	3.7	122

215	ALS mutations in FUS cause neuronal dysfunction and death in Caenorhabditis elegans by a dominant gain-of-function mechanism. *Human Molecular Genetics*, **2012**, 21, 1-9	5.6	121
214	Abeta-degrading endopeptidase, neprilysin, in mouse brain: synaptic and axonal localization inversely correlating with Abeta pathology. *Neuroscience Research*, **2002**, 43, 39-56	2.9	121
213	Investigating the genetic architecture of dementia with Lewy bodies: a two-stage genome-wide association study. *Lancet Neurology, The*, **2018**, 17, 64-74	24.1	121
212	Assessment of the genetic variance of late-onset Alzheimer's disease. *Neurobiology of Aging*, **2016**, 41, 200.e13-200.e20	5.6	119
211	Genotype-renal function correlation in type 2 autosomal dominant polycystic kidney disease. *Journal of the American Society of Nephrology: JASN*, **2003**, 14, 1164-74	12.7	116
210	Identification of Alzheimer disease-associated variants in genes that regulate retromer function. *Neurobiology of Aging*, **2012**, 33, 2231.e15-2231.e30	5.6	115
209	Extracellular monomeric tau protein is sufficient to initiate the spread of tau protein pathology. *Journal of Biological Chemistry*, **2014**, 289, 956-67	5.4	111
208	An alpha-2-macroglobulin insertion-deletion polymorphism in Alzheimer disease. *Nature Genetics*, **1999**, 22, 19-22	36.3	107
207	The human NACP/alpha-synuclein gene: chromosome assignment to 4q21.3-q22 and TaqI RFLP analysis. *Genomics*, **1995**, 26, 425-7	4.3	107
206	Transethnic genome-wide scan identifies novel Alzheimer's disease loci. *Alzheimers and Dementia*, **2017**, 13, 727-738	1.2	106
205	Mutations of PKD1 in ADPKD2 cysts suggest a pathogenic effect of trans-heterozygous mutations. *Nature Genetics*, **2000**, 25, 143-4	36.3	105
204	Rare coding mutations identified by sequencing of Alzheimer disease genome-wide association studies loci. *Annals of Neurology*, **2015**, 78, 487-98	9.4	102
203	A comparison of neurological, metabolic, structural, and genetic evaluations in persons at risk for Huntington's disease. *Annals of Neurology*, **1990**, 28, 614-21	9.4	100
202	Genome-wide linkage scan of a large family with IgA nephropathy localizes a novel susceptibility locus to chromosome 2q36. *Journal of the American Society of Nephrology: JASN*, **2007**, 18, 2408-15	12.7	99
201	Elevated plasma triglyceride levels precede amyloid deposition in Alzheimer's disease mouse models with abundant A beta in plasma. *Neurobiology of Disease*, **2006**, 24, 114-27	7.5	96
200	Analysis of the glucocerebrosidase gene in Parkinson's disease. *Movement Disorders*, **2005**, 20, 367-70	7	93
199	Increased production of beta-amyloid and vulnerability to endoplasmic reticulum stress by an aberrant spliced form of presenilin 2. *Journal of Biological Chemistry*, **2001**, 276, 2108-14	5.4	93
198	Presenilin 1 mutations activate gamma 42-secretase but reciprocally inhibit epsilon-secretase cleavage of amyloid precursor protein (APP) and S3-cleavage of notch. *Journal of Biological Chemistry*, **2002**, 277, 36521-6	5.4	93

197	Frontotemporal dementia with novel tau pathology and a Glu342Val tau mutation. *Annals of Neurology*, **2000**, 48, 850-858	9.4	91
196	Gene-wide analysis detects two new susceptibility genes for Alzheimer's disease. *PLoS ONE*, **2014**, 9, e94661	3.7	90
195	Pyroglutamate-3 amyloid-□deposition in the brains of humans, non-human primates, canines, and Alzheimer disease-like transgenic mouse models. *American Journal of Pathology*, **2013**, 183, 369-81	5.8	84
194	Investigation of c9orf72 in 4 neurodegenerative disorders. *Archives of Neurology*, **2012**, 69, 1583-90		83
193	Benign hereditary chorea: clinical, genetic, and pathological findings. *Annals of Neurology*, **2003**, 54, 244-7	9.4	81
192	Therapeutic effects of remediating autophagy failure in a mouse model of Alzheimer disease by enhancing lysosomal proteolysis. *Autophagy*, **2011**, 7, 788-9	10.2	80
191	SORCS1 alters amyloid precursor protein processing and variants may increase Alzheimer's disease risk. *Annals of Neurology*, **2011**, 69, 47-64	9.4	79
190	Association between SORL1 and Alzheimer's disease in a genome-wide study. *NeuroReport*, **2007**, 18, 1761-4	1.7	76
189	Dissociated phenotypes in presenilin transgenic mice define functionally distinct gamma-secretases. *Proceedings of the National Academy of Sciences of the United States of America*, **2005**, 102, 8972-7	11.5	76
188	Presenilin structure, function and role in Alzheimer disease. *Biochimica Et Biophysica Acta - Molecular Basis of Disease*, **2000**, 1502, 1-15	6.9	75
187	Brain levels of CDK5 activator p25 are not increased in Alzheimer's or other neurodegenerative diseases with neurofibrillary tangles. *Journal of Neurochemistry*, **2003**, 86, 572-81	6	74
186	Progressive loss of renal function is an age-dependent heritable trait in type 1 autosomal dominant polycystic kidney disease. *Journal of the American Society of Nephrology: JASN*, **2005**, 16, 755-62	12.7	73
185	Homozygous inheritance of the Machado-Joseph disease gene. *Annals of Neurology*, **1994**, 36, 443-7	9.4	73
184	ATP-binding cassette transporter A7 (ABCA7) loss of function alters Alzheimer amyloid processing. *Journal of Biological Chemistry*, **2015**, 290, 24152-65	5.4	71
183	Hereditary spastic paraplegia: clinical genetic study of 15 families. *Archives of Neurology*, **2004**, 61, 849-55		71
182	TREM2 shedding by cleavage at the H157-S158 bond is accelerated for the Alzheimer's disease-associated H157Y variant. *EMBO Molecular Medicine*, **2017**, 9, 1366-1378	12	70
181	Rare individual amyloid-□oligomers act on astrocytes to initiate neuronal damage. *Biochemistry*, **2014**, 53, 2442-53	3.2	68
180	Hypermethylation of the CpG-island near the C9orf72 G□□repeat expansion in FTLD patients. *Human Molecular Genetics*, **2014**, 23, 5630-7	5.6	68

179	Single molecule characterization of the interactions between amyloid-β peptides and the membranes of hippocampal cells. *Journal of the American Chemical Society*, **2013**, 135, 1491-8	16.4	68
178	The effect of alcohol and tobacco consumption, and apolipoprotein E genotype, on the age of onset in Alzheimer's disease. *International Journal of Geriatric Psychiatry*, **2010**, 25, 511-8	3.9	68
177	Reentrant liquid condensate phase of proteins is stabilized by hydrophobic and non-ionic interactions. *Nature Communications*, **2021**, 12, 1085	17.4	68
176	Mutation analysis of CHCHD10 in different neurodegenerative diseases. *Brain*, **2015**, 138, e380	11.2	67
175	Familial Alzheimer's disease: progress and problems. *Neurobiology of Aging*, **1989**, 10, 417-25	5.6	67
174	Appoptosin-Mediated Caspase Cleavage of Tau Contributes to Progressive Supranuclear Palsy Pathogenesis. *Neuron*, **2015**, 87, 963-75	13.9	66
173	Both common variations and rare non-synonymous substitutions and small insertion/deletions in CLU are associated with increased Alzheimer risk. *Molecular Neurodegeneration*, **2012**, 7, 3	19	66
172	Group II metabotropic glutamate receptor stimulation triggers production and release of Alzheimer's amyloid(beta)42 from isolated intact nerve terminals. *Journal of Neuroscience*, **2010**, 30, 3870-5	6.6	66
171	Carboxyl-terminal fragments of Alzheimer beta-amyloid precursor protein accumulate in restricted and unpredicted intracellular compartments in presenilin 1-deficient cells. *Journal of Biological Chemistry*, **2000**, 275, 36794-802	5.4	65
170	Apolipoprotein E genotype in patients with Alzheimer's disease: implications for the risk of dementia among relatives. *Annals of Neurology*, **1995**, 38, 797-808	9.4	65
169	ALS5/SPG11/KIAA1840 mutations cause autosomal recessive axonal Charcot-Marie-Tooth disease. *Brain*, **2016**, 139, 73-85	11.2	64
168	Genetic association of CR1 with Alzheimer's disease: a tentative disease mechanism. *Neurobiology of Aging*, **2012**, 33, 2949.e5-2949.e12	5.6	64
167	Cloning and characterization of the Drosophila presenilin homologue. *NeuroReport*, **1997**, 8, 1025-9	1.7	64
166	Will anti-amyloid therapies work for Alzheimer's disease?. *Lancet, The*, **2008**, 372, 180-2	40	64
165	The presenilin proteins are components of multiple membrane-bound complexes that have different biological activities. *Journal of Biological Chemistry*, **2004**, 279, 31329-36	5.4	64
164	Drug Repositioning for Alzheimer's Disease Based on Systematic 'omics' Data Mining. *PLoS ONE*, **2016**, 11, e0168812	3.7	62
163	Analysis of IFT74 as a candidate gene for chromosome 9p-linked ALS-FTD. *BMC Neurology*, **2006**, 6, 44	3.1	61
162	Molecular genetics of Alzheimer's disease: the role of beta-amyloid and the presenilins. *Current Opinion in Neurology*, **2000**, 13, 377-84	7.1	60

161	Three different mutations of presenilin 1 gene in early-onset Alzheimer's disease families. *Neuroscience Letters*, **1996**, 208, 195-8	3.3	60
160	Molecular genetic approaches to Alzheimer's disease. *Trends in Neurosciences*, **1989**, 12, 152-8	13.3	60
159	Association studies of cholesterol metabolism genes (CH25H, ABCA1 and CH24H) in Alzheimer's disease. *Neuroscience Letters*, **2006**, 391, 142-6	3.3	58
158	Crowdsourced estimation of cognitive decline and resilience in Alzheimer's disease. *Alzheimers and Dementia*, **2016**, 12, 645-53	1.2	58
157	Structural interactions between inhibitor and substrate docking sites give insight into mechanisms of human PS1 complexes. *Structure*, **2014**, 22, 125-35	5.2	55
156	Cortical neuronal and glial pathology in TgTauP301L transgenic mice: neuronal degeneration, memory disturbance, and phenotypic variation. *American Journal of Pathology*, **2006**, 169, 1365-75	5.8	55
155	Intracellular oligomeric amyloid-beta rapidly regulates GluA1 subunit of AMPA receptor in the hippocampus. *Scientific Reports*, **2015**, 5, 10934	4.9	54
154	Loss of gamma-secretase function impairs endocytosis of lipoprotein particles and membrane cholesterol homeostasis. *Journal of Neuroscience*, **2008**, 28, 12097-106	6.6	54
153	Conversion to dementia among two groups with cognitive impairment. A preliminary report. *Dementia and Geriatric Cognitive Disorders*, **2004**, 18, 307-13	2.6	54
152	Association of distinct variants in SORL1 with cerebrovascular and neurodegenerative changes related to Alzheimer disease. *Archives of Neurology*, **2008**, 65, 1640-8		53
151	Vps10 family proteins and the retromer complex in aging-related neurodegeneration and diabetes. *Journal of Neuroscience*, **2012**, 32, 14080-6	6.6	52
150	Current and future implications of basic and translational research on amyloid-□peptide production and removal pathways. *Molecular and Cellular Neurosciences*, **2015**, 66, 3-11	4.8	50
149	Both the sequence and length of the C terminus of PEN-2 are critical for intermolecular interactions and function of presenilin complexes. *Journal of Biological Chemistry*, **2004**, 279, 46455-63	5.4	50
148	Genome-wide analysis of genetic correlation in dementia with Lewy bodies, Parkinson's and Alzheimer's diseases. *Neurobiology of Aging*, **2016**, 38, 214.e7-214.e10	5.6	49
147	Genetic complexity of Alzheimer's disease: successes and challenges. *Journal of Alzheimers Disease*, **2006**, 9, 381-7	4.3	46
146	Impaired conditioned taste aversion learning in APP transgenic mice. *Neurobiology of Aging*, **2004**, 25, 1213-9	5.6	46
145	mutations in early- and late-onset Alzheimer disease. *Neurology: Genetics*, **2016**, 2, e116	3.8	45
144	The physiological and pathological biophysics of phase separation and gelation of RNA binding proteins in amyotrophic lateral sclerosis and fronto-temporal lobar degeneration. *Brain Research*, **2018**, 1693, 11-23	3.7	44

143	Genome-wide survey of large rare copy number variants in Alzheimer's disease among Caribbean hispanics. *G3: Genes, Genomes, Genetics*, **2012**, 2, 71-8	3.2	43
142	p53-Dependent transcriptional control of cellular prion by presenilins. *Journal of Neuroscience*, **2009**, 29, 6752-60	6.6	42
141	Genetic variability in CHMP2B and frontotemporal dementia. *Neurodegenerative Diseases*, **2006**, 3, 129-33	2.3	42
140	Catenin is genetically and biologically associated with cortical cataract and future Alzheimer-related structural and functional brain changes. *PLoS ONE*, **2012**, 7, e43728	3.7	42
139	Cytosolic proteins regulate alpha-synuclein dissociation from presynaptic membranes. *Journal of Biological Chemistry*, **2006**, 281, 32148-55	5.4	40
138	The gene encoding nicastrin, a major gamma-secretase component, modifies risk for familial early-onset Alzheimer disease in a Dutch population-based sample. *American Journal of Human Genetics*, **2002**, 70, 1568-74	11	39
137	TET2 Regulates the Neuroinflammatory Response in Microglia. *Cell Reports*, **2019**, 29, 697-713.e8	10.6	38
136	Identification of the genetic locus for keratosis palmaris et plantaris on chromosome 17 near the RARA and keratin type I genes. *Nature Genetics*, **1993**, 5, 158-62	36.3	38
135	Random mutagenesis of presenilin-1 identifies novel mutants exclusively generating long amyloid beta-peptides. *Journal of Biological Chemistry*, **2005**, 280, 19070-7	5.4	36
134	Aberrant presenilin-1 expression downregulates LDL receptor-related protein (LRP): is LRP central to Alzheimer's disease pathogenesis?. *Molecular and Cellular Neurosciences*, **1999**, 14, 129-40	4.8	36
133	Alzheimer amyloid peptide a42 regulates gene expression of transcription and growth factors. *Journal of Alzheimer's Disease*, **2015**, 44, 613-24	4.3	35
132	Single-molecule imaging reveals that small amyloid-1-42 oligomers interact with the cellular prion protein (PrP(C)). *ChemBioChem*, **2014**, 15, 2515-21	3.8	35
131	Epidemiology and genetics of frontotemporal dementia: a door-to-door survey in southern Italy. *Neurobiology of Aging*, **2012**, 33, 2948.e1-2948.e10	5.6	35
130	The gamma/epsilon-secretase-derived APP intracellular domain fragments regulate p53. *Current Alzheimer Research*, **2007**, 4, 423-6	3	35
129	F-box/LRR-repeat protein 7 is genetically associated with Alzheimer's disease. *Annals of Clinical and Translational Neurology*, **2015**, 2, 810-20	5.3	34
128	No association between apolipoprotein E genotype and late-onset depression in Alzheimer's disease. *Biological Psychiatry*, **1997**, 41, 246-8	7.9	34
127	Evidence of recessive Alzheimer disease loci in a Caribbean Hispanic data set: genome-wide survey of runs of homozygosity. *JAMA Neurology*, **2013**, 70, 1261-7	17.2	33
126	The levels of mature glycosylated nicastrin are regulated and correlate with gamma-secretase processing of amyloid beta-precursor protein. *Journal of Neurochemistry*, **2002**, 83, 1065-71	6	33

125	Biomolecular condensates undergo a generic shear-mediated liquid-to-solid transition. *Nature Nanotechnology*, **2020**, 15, 841-847	28.7	33
124	Expanded genomewide scan implicates a novel locus at 3q28 among Caribbean hispanics with familial Alzheimer disease. *Archives of Neurology*, **2006**, 63, 1591-8		32
123	Rarity of the Alzheimer disease-protective APP A673T variant in the United States. *JAMA Neurology* , **2015**, 72, 209-16	17.2	31
122	Family reunion--the ZIP/prion gene family. *Progress in Neurobiology*, **2011**, 93, 405-20	10.9	31
121	Splicing mutation of presenilin-1 gene for early-onset familial Alzheimer's disease. *Human Mutation*, **1998**, Suppl 1, S91-4	4.7	31
120	Presenilin 1 and presenilin 2 have differential effects on the stability and maturation of nicastrin in Mammalian brain. *Journal of Biological Chemistry*, **2003**, 278, 19974-9	5.4	31
119	Inhibiting amyloid precursor protein C-terminal cleavage promotes an interaction with presenilin 1. *Journal of Biological Chemistry*, **2000**, 275, 20794-8	5.4	31
118	Genome sequencing analysis identifies new loci associated with Lewy body dementia and provides insights into its genetic architecture. *Nature Genetics*, **2021**, 53, 294-303	36.3	31
117	A novel pathogenic mutation (Leu262Phe) found in the presenilin 1 gene in early-onset Alzheimer's disease. *Neuroscience Letters*, **1997**, 234, 3-6	3.3	30
116	Structural biology of presenilin 1 complexes. *Molecular Neurodegeneration*, **2014**, 9, 59	19	29
115	Proteolytic processing of presenilin-1 (PS-1) is not associated with Alzheimer's disease with or without PS-1 mutations. *FEBS Letters*, **1997**, 418, 162-6	3.8	29
114	Recurrent fetal loss associated with bilineal inheritance of type 1 autosomal dominant polycystic kidney disease. *American Journal of Kidney Diseases*, **2002**, 40, 16-20	7.4	28
113	Genetic association study of PINK1 coding polymorphisms in Parkinson's disease. *Neuroscience Letters*, **2004**, 372, 226-9	3.3	28
112	Association of Long Runs of Homozygosity With Alzheimer Disease Among African American Individuals. *JAMA Neurology*, **2015**, 72, 1313-23	17.2	27
111	New locus for hereditary spastic paraplegia maps to chromosome 1p31.1-1p21.1. *Annals of Neurology*, **2005**, 58, 423-9	9.4	27
110	Clinical and genetic study of a large SPG4 Italian family. *Movement Disorders*, **2005**, 20, 1055-9	7	27
109	Gerstmann-Str□ssler-Scheinker disease (PRNP P102L): amyloid deposits are best recognized by antibodies directed to epitopes in PrP region 90-165. *Journal of Neuropathology and Experimental Neurology*, **1995**, 54, 790-801	3.1	27
108	APH1 polar transmembrane residues regulate the assembly and activity of presenilin complexes. *Journal of Biological Chemistry*, **2009**, 284, 16298-16307	5.4	26

107	IL5RA and TNFRSF6B gene variants are associated with sporadic IgA nephropathy. *Journal of the American Society of Nephrology: JASN*, **2008**, 19, 1025-33	12.7	26
106	Integrative approach to sporadic Alzheimer's disease: deficiency of TYROBP in a tauopathy mouse model reduces C1q and normalizes clinical phenotype while increasing spread and state of phosphorylation of tau. *Molecular Psychiatry*, **2019**, 24, 1383-1397	15.1	26
105	APOE-ε4 associates with hippocampal volume, learning, and memory across the spectrum of Alzheimer's disease and dementia with Lewy bodies. *Alzheimer's and Dementia*, **2018**, 14, 1137-1147	1.2	26
104	Early fear memory defects are associated with altered synaptic plasticity and molecular architecture in the TgCRND8 Alzheimer's disease mouse model. *Journal of Comparative Neurology*, **2014**, 522, 2319-35	3.4	25
103	Clinical and pathologic evidence of corticobasal degeneration and progressive supranuclear palsy in familial tauopathy. *Archives of Neurology*, **2005**, 62, 1453-7		25
102	Biology of presenilins as causative molecules for Alzheimer disease. *Clinical Genetics*, **1999**, 55, 219-25	4	25
101	Mutation analysis of patients with neurodegenerative disorders using NeuroX array. *Neurobiology of Aging*, **2015**, 36, 545.e9-14	5.6	24
100	Beta amyloid aggregates induce sensitised TLR4 signalling causing long-term potentiation deficit and rat neuronal cell death. *Communications Biology*, **2020**, 3, 79	6.7	24
99	Presenilin-1 holoprotein is an interacting partner of sarco endoplasmic reticulum calcium-ATPase and confers resistance to endoplasmic reticulum stress. *Journal of Alzheimer's Disease*, **2010**, 20, 261-73	4.3	24
98	An exploration of cognitive subgroups in Alzheimer's disease. *Journal of the International Neuropsychological Society*, **2010**, 16, 233-43	3.1	24
97	Age-at-onset linkage analysis in Caribbean Hispanics with familial late-onset Alzheimer's disease. *Neurogenetics*, **2008**, 9, 51-60	3	24
96	Presenilin function: connections to Alzheimer's disease and signal transduction. *Biochemical Society Symposia*, **2001**, 89-100		24
95	Mutation analysis of the MS4A and TREM gene clusters in a case-control Alzheimer's disease data set. *Neurobiology of Aging*, **2016**, 42, 217.e7-217.e13	5.6	24
94	Amyloid-beta-protein isoforms in brain of subjects with PS1-linked, beta APP-linked and sporadic Alzheimer disease. *Molecular Brain Research*, **1998**, 56, 178-85		23
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