# Christine Van Broeckhoven

List of Publications by Year
in descending order

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| 797 | 76,635 | 599 128 | 1056 241 |
|---|---|---|---|
| papers | citations | h-index | g-index |

| 872 | 872 | 872 | 56352 |
|---|---|---|---|
| all docs | docs citations | times ranked | citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Meta-analysis of 74,046 individuals identifies 11 new susceptibility loci for Alzheimer's disease. Nature Genetics, 2013, 45, 1452-1458. | 9.4 | 3,741 |
| 2 | Genome-wide association study identifies variants at CLU and PICALM associated with Alzheimer's disease. Nature Genetics, 2009, 41, 1088-1093. | 9.4 | 2,697 |
| 3 | Genome-wide association study identifies variants at CLU and CR1 associated with Alzheimer's disease. Nature Genetics, 2009, 41, 1094-1099. | 9.4 | 2,155 |
| 4 | Genetic meta-analysis of diagnosed Alzheimerâ€™s disease identifies new risk loci and implicates AÎ², tau, immunity and lipid processing. Nature Genetics, 2019, 51, 414-430. | 9.4 | 1,962 |
| 5 | NanoPack: visualizing and processing long-read sequencing data. Bioinformatics, 2018, 34, 2666-2669. | 1.8 | 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. | 9.4 | 1,708 |
| 7 | Null mutations in progranulin cause ubiquitin-positive frontotemporal dementia linked to chromosome 17q21. Nature, 2006, 442, 920-924. | 13.7 | 1,386 |
| 8 | Atherosclerosis, apolipoprotein E, and prevalence of dementia and Alzheimer's disease in the Rotterdam Study. Lancet, The, 1997, 349, 151-154. | 6.3 | 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. | 2.6 | 1,111 |
| 10 | The C9orf72 GGGGCC Repeat Is Translated into Aggregating Dipeptide-Repeat Proteins in FTLD/ALS. Science, 2013, 339, 1335-1338. | 6.0 | 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. | 9.4 | 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. | 9.4 | 783 |
| 13 | Mutation of POLG is associated with progressive external ophthalmoplegia characterized by mtDNA deletions. Nature Genetics, 2001, 28, 211-212. | 9.4 | 748 |
| 14 | Extra-pair paternity results from female preference for high-quality males in the blue tit. Nature, 1992, 357, 494-496. | 13.7 | 720 |
| 15 | Presenile dementia and cerebral haemorrhage linked to a mutation at codon 692 of the Î²â€“amyloid precursor protein gene. Nature Genetics, 1992, 1, 218-221. | 9.4 | 715 |
| 16 | New insights into the genetic etiology of Alzheimerâ€™s disease and related dementias. Nature Genetics, 2022, 54, 412-436. | 9.4 | 700 |
| 17 | The genetic landscape of Alzheimer disease: clinical implications and perspectives. Genetics in Medicine, 2016, 18, 421-430. | 1.1 | 695 |
| 18 | TREM2 mutations implicated in neurodegeneration impair cell surface transport and phagocytosis. Science Translational Medicine, 2014, 6, 243ra86. | 5.8 | 600 |

| # | Article | IF | Citations |
|---|---|---|---|
| 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. Lancet Neurology, The, 2012, 11, 54-65. | 4.9 | 565 |
| 20 | APOE and Alzheimer disease: a major gene with semi-dominant inheritance. Molecular Psychiatry, 2011, 16, 903-907. | 4.1 | 529 |
| 21 | 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, 3858-3863. | 3.3 | 491 |
| 22 | Common variants at 7p21 are associated with frontotemporal lobar degeneration with TDP-43 inclusions. Nature Genetics, 2010, 42, 234-239. | 9.4 | 479 |
| 23 | Smoking and risk of dementia and Alzheimer's disease in a population-based cohort study: the Rotterdam Study. Lancet, The, 1998, 351, 1840-1843. | 6.3 | 475 |
| 24 | Collaborative Analysis of Î±-Synuclein Gene Promoter Variability and Parkinson Disease. JAMA - Journal of the American Medical Association, 2006, 296, 661. | 3.8 | 467 |
| 25 | Apolipoprotein E4 allele in a populationâ€“based study of earlyâ€“onset Alzheimer's disease. Nature Genetics, 1994, 7, 74-78. | 9.4 | 460 |
| 26 | Amyloid beta protein precursor gene and hereditary cerebral hemorrhage with amyloidosis (Dutch). Science, 1990, 248, 1120-1122. | 6.0 | 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. Blood, 2001, 98, 49-56. | 0.6 | 438 |
| 28 | Genetic etiology of Parkinson disease associated with mutations in the SNCA, PARK2, PINK1, PARK7, and LRRK2 genes: a mutation update. Human Mutation, 2010, 31, 763-780. | 1.1 | 428 |
| 29 | Bidirectional transcripts of the expanded C9orf72 hexanucleotide repeat are translated into aggregating dipeptide repeat proteins. Acta Neuropathologica, 2013, 126, 881-893. | 3.9 | 427 |
| 30 | Genetic Association of Apolipoprotein E with Age-Related Macular Degeneration. American Journal of Human Genetics, 1998, 63, 200-206. | 2.6 | 425 |
| 31 | Locusâ€specific mutation databases for neurodegenerative brain diseases. Human Mutation, 2012, 33, 1340-1344. | 1.1 | 414 |
| 32 | Molecular genetics of earlyâ€onset Alzheimer's disease revisited. Alzheimer's and Dementia, 2016, 12, 733-748. | 0.4 | 409 |
| 33 | Genetic linkage studies suggest that Alzheimer's disease is not a single homogeneous disorder. Nature, 1990, 347, 194-197. | 13.7 | 407 |
| 34 | The peripheral myelin protein gene PMPâ€“22 is contained within the Charcotâ€“Marieâ€“Tooth disease type 1A duplication. Nature Genetics, 1992, 1, 171-175. | 9.4 | 404 |
| 35 | Genome Scan Meta-Analysis of Schizophrenia and Bipolar Disorder, Part III: Bipolar Disorder. American Journal of Human Genetics, 2003, 73, 49-62. | 2.6 | 400 |
| 36 | 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. | 1.4 | 396 |

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|---|---|---|---|
| 37 | Hot-spot residue in small heat-shock protein 22 causes distal motor neuropathy. Nature Genetics, 2004, 36, 597-601. | 9.4 | 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. EMBO Molecular Medicine, 2016, 8, 466-476. | 3.3 | 392 |
| 39 | Risk Estimates of Dementia by Apolipoprotein E Genotypes From a Population-Based Incidence Study: The Rotterdam Study. Archives of Neurology, 1998, 55, 964. | 4.9 | 378 |
| 40 | A Multicenter Study of Glucocerebrosidase Mutations in Dementia With Lewy Bodies. JAMA Neurology, 2013, 70, 727. | 4.5 | 374 |
| 41 | Clinical Phenotypes of Different MPZ (P0) Mutations May Include Charcotâ€“Marieâ€“Tooth Type 1B, Dejerineâ€“Sottas, and Congenital Hypomyelination. Neuron, 1996, 17, 451-460. | 3.8 | 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. European Journal of Human Genetics, 1996, 4, 25-33. | 1.4 | 371 |
| 43 | APP duplication is sufficient to cause early onset Alzheimer's dementia with cerebral amyloid angiopathy. Brain, 2006, 129, 2977-2983. | 3.7 | 337 |
| 44 | Phenotype variability in progranulin mutation carriers: a clinical, neuropsychological, imaging and genetic study. Brain, 2008, 131, 732-746. | 3.7 | 331 |
| 45 | The role of tau (MAPT) in frontotemporal dementia and related tauopathies. Human Mutation, 2004, 24, 277-295. | 1.1 | 323 |
| 46 | Mapping of a gene predisposing to earlyâ€“onset Alzheimer's disease to chromosome 14q24.3. Nature Genetics, 1992, 2, 335-339. | 9.4 | 321 |
| 47 | Increased expression of BIN1 mediates Alzheimer genetic risk by modulating tau pathology. Molecular Psychiatry, 2013, 18, 1225-1234. | 4.1 | 321 |
| 48 | Genetic insights in Alzheimer's disease. Lancet Neurology, The, 2013, 12, 92-104. | 4.9 | 310 |
| 49 | Mean age-of-onset of familial alzheimer disease caused by presenilin mutations correlates with both increased AÎ²42 and decreased AÎ²40. Human Mutation, 2006, 27, 686-695. | 1.1 | 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. Acta Neuropathologica, 2013, 125, 413-423. | 3.9 | 302 |
| 51 | Frontotemporal dementia and its subtypes: a genome-wide association study. Lancet Neurology, The, 2014, 13, 686-699. | 4.9 | 302 |
| 52 | Association of LRRK2 exonic variants with susceptibility to Parkinson's disease: a caseâ€“control study. Lancet Neurology, The, 2011, 10, 898-908. | 4.9 | 294 |
| 53 | A novel Alzheimer disease locus located near the gene encoding tau protein. Molecular Psychiatry, 2016, 21, 108-117. | 4.1 | 260 |
| 54 | novoSNP, a novel computational tool for sequence variation discovery. Genome Research, 2005, 15, 436-442. | 2.4 | 254 |

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|---|---|---|---|
| 55 | POLG mutations in neurodegenerative disorders with ataxia but no muscle involvement. Neurology, 2004, 63, 1251-1257. | 1.5 | 252 |
| 56 | Apolipoprotein E epsilon4 and the risk of dementia with stroke. A population-based investigation. JAMA - Journal of the American Medical Association, 1997, 277, 818-821. | 3.8 | 252 |
| 57 | A Panâ€<scp>E</scp>uropean Study of theC9orf72Repeat Associated with<scp>FTLD</scp>: Geographic Prevalence, Genomic Instability, and Intermediate Repeats. Human Mutation, 2013, 34, 363-373. | 1.1 | 247 |
| 58 | Potent amyloidogenicity and pathogenicity of AÎ²43. Nature Neuroscience, 2011, 14, 1023-1032. | 7.1 | 245 |
| 59 | The genetics and neuropathology of frontotemporal lobar degeneration. Acta Neuropathologica, 2012, 124, 353-372. | 3.9 | 242 |
| 60 | Drosophila screen connects nuclear transport genes to DPR pathology in c9ALS/FTD. Scientific Reports, 2016, 6, 20877. | 1.6 | 239 |
| 61 | Mutations in DNAJC5, Encoding Cysteine-String Protein Alpha, Cause Autosomal-Dominant Adult-Onset Neuronal Ceroid Lipofuscinosis. American Journal of Human Genetics, 2011, 89, 241-252. | 2.6 | 236 |
| 62 | Mutations in the peripheral myelin genes and associated genes in inherited peripheral neuropathies. Human Mutation, 1999, 13, 11-28. | 1.1 | 232 |
| 63 | ALS Genes in the Genomic Era and their Implications for FTD. Trends in Genetics, 2018, 34, 404-423. | 2.9 | 229 |
| 64 | Mutations in SEPT9 cause hereditary neuralgic amyotrophy. Nature Genetics, 2005, 37, 1044-1046. | 9.4 | 222 |
| 65 | FUS pathology defines the majority of tau- and TDP-43-negative frontotemporal lobar degeneration. Acta Neuropathologica, 2010, 120, 33-41. | 3.9 | 222 |
| 66 | Recessive POLG mutations presenting with sensory and ataxic neuropathy in compound heterozygote patients with progressive external ophthalmoplegia. Neuromuscular Disorders, 2003, 13, 133-142. | 0.3 | 216 |
| 67 | The Thr124Met mutation in the peripheral myelin protein zero (MPZ) gene is associated with a clinically distinct Charcotâ€“Marieâ€“Tooth phenotype. Brain, 1999, 122, 281-290. | 3.7 | 215 |
| 68 | Pathogenesis of polyglutamine disorders: aggregation revisited. Human Molecular Genetics, 2003, 12, R173-R186. | 1.4 | 212 |
| 69 | A novel presenilin 1 mutation associated with Pick's disease but not ?-amyloid plaques. Annals of Neurology, 2004, 55, 617-626. | 2.8 | 210 |
| 70 | Genetic variation in DPP6 is associated with susceptibility to amyotrophic lateral sclerosis. Nature Genetics, 2008, 40, 29-31. | 9.4 | 205 |
| 71 | Presenilins and Alzheimer disease. Nature Genetics, 1995, 11, 230-232. | 9.4 | 204 |
| 72 | Germline mosaicism and Duchenne muscular dystrophy mutations. Nature, 1987, 329, 554-556. | 13.7 | 201 |

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|---|---|---|---|
| 73 | The C9orf72 repeat size correlates with onset age of disease, DNA methylation and transcriptional downregulation of the promoter. Molecular Psychiatry, 2016, 21, 1112-1124. | 4.1 | 201 |
| 74 | ?-Synuclein promoter confers susceptibility to Parkinson's disease. Annals of Neurology, 2004, 56, 591-595. | 2.8 | 200 |
| 75 | Genetic contribution of FUS to frontotemporal lobar degeneration. Neurology, 2010, 74, 366-371. | 1.5 | 197 |
| 76 | Molecular biological characterization of an azole-resistant Candida glabrata isolate. Antimicrobial Agents and Chemotherapy, 1997, 41, 2229-2237. | 1.4 | 196 |
| 77 | Molecular genetics of Alzheimer's disease: An update. Annals of Medicine, 2008, 40, 562-583. | 1.5 | 196 |
| 78 | ITPR2 as a susceptibility gene in sporadic amyotrophic lateral sclerosis: a genome-wide association study. Lancet Neurology, The, 2007, 6, 869-877. | 4.9 | 195 |
| 79 | Serum biomarker for progranulinâ€associated frontotemporal lobar degeneration. Annals of Neurology, 2009, 65, 603-609. | 2.8 | 195 |
| 80 | Progress in unraveling the genetic etiology of Parkinson disease in a genomic era. Trends in Genetics, 2015, 31, 140-149. | 2.9 | 193 |
| 81 | Further evidence that neurofilament light chain gene mutations can cause Charcot-Marie-Tooth disease type 2E. Annals of Neurology, 2001, 49, 245-249. | 2.8 | 188 |
| 82 | Germinal mosaicism increases the recurrence risk for 'new' Duchenne muscular dystrophy mutations.. Journal of Medical Genetics, 1989, 26, 553-559. | 1.5 | 187 |
| 83 | Alzheimer risk associated with a copy number variation in the complement receptor 1 increasing C3b/C4b binding sites. Molecular Psychiatry, 2012, 17, 223-233. | 4.1 | 179 |
| 84 | Pathogenic APP mutations near the gamma-secretase cleavage site differentially affect Abeta secretion and APP C-terminal fragment stability. Human Molecular Genetics, 2001, 10, 1665-1671. | 1.4 | 178 |
| 85 | De-novo mutation in hereditary motor and sensory neuropathy type I. Lancet, The, 1992, 339, 1081-1082. | 6.3 | 177 |
| 86 | Promoter Mutations That Increase Amyloid Precursor-Protein Expression Are Associated with Alzheimer Disease. American Journal of Human Genetics, 2006, 78, 936-946. | 2.6 | 173 |
| 87 | Genetic variation in the KIF1B locus influences susceptibility to multiple sclerosis. Nature Genetics, 2008, 40, 1402-1403. | 9.4 | 173 |
| 88 | Convergent genetic and expression data implicate immunity in Alzheimer's disease. Alzheimer's and Dementia, 2015, 11, 658-671. | 0.4 | 173 |
| 89 | Molecular genetic analysis of familial early-onset Alzheimer's disease linked to chromosome 14q24.3. Human Molecular Genetics, 1995, 4, 2363-2371. | 1.4 | 171 |
| 90 | De novoSCN1Amutations are a major cause of severe myoclonic epilepsy of infancy. Human Mutation, 2003, 21, 615-621. | 1.1 | 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. | 1.4 | 169 |
| 92 | Presenilin mutations in Alzheimer's disease. , 1998, 11, 183-190. | | 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. | 2.6 | 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. | 1.9 | 168 |
| 95 | Disruption of endocytic trafficking in frontotemporal dementia with CHMP2B mutations. Human Molecular Genetics, 2010, 19, 2228-2238. | 1.4 | 163 |
| 96 | Progranulin null mutations in both sporadic and familial frontotemporal dementia. Human Mutation, 2007, 28, 846-855. | 1.1 | 162 |
| 97 | Genetic variability in progranulin contributes to risk for clinically diagnosed Alzheimer disease. Neurology, 2008, 71, 656-664. | 1.5 | 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. | 1.5 | 158 |
| 99 | Flemish and Dutch Mutations in Amyloid Î² Precursor Protein Have Different Effects on Amyloid Î² Secretion. Neurobiology of Disease, 1998, 5, 281-286. | 2.1 | 157 |
| 100 | The molecular basis of the frontotemporal lobar degenerationâ€“amyotrophic lateral sclerosis spectrum. Annals of Medicine, 2012, 44, 817-828. | 1.5 | 157 |
| 101 | High-density SNP haplotyping suggests altered regulation of tau gene expression in progressive supranuclear palsy. Human Molecular Genetics, 2005, 14, 3281-3292. | 1.4 | 156 |
| 102 | Sequencing an Ashkenazi reference panel supports population-targeted personal genomics and illuminates Jewish and European origins. Nature Communications, 2014, 5, 4835. | 5.8 | 156 |
| 103 | Common pathobiochemical hallmarks of progranulin-associated frontotemporal lobar degeneration and neuronal ceroid lipofuscinosis. Acta Neuropathologica, 2014, 127, 845-60. | 3.9 | 156 |
| 104 | The dopamine D4 receptor gene 48-base-pair-repeat polymorphism and mood disorders: A meta-analysis. Biological Psychiatry, 2005, 57, 999-1003. | 0.7 | 155 |
| 105 | Gene-Wide Analysis Detects Two New Susceptibility Genes for Alzheimer's Disease. PLoS ONE, 2014, 9, e94661. | 1.1 | 155 |
| 106 | CAG repeat expansion in the TATA box-binding protein gene causes autosomal dominant cerebellar ataxia. Brain, 2001, 124, 1939-1947. | 3.7 | 154 |
| 107 | Altered brain white matter integrity in healthy carriers of the APOE Îµ4 allele. Neurology, 2006, 66, 1029-1033. | 1.5 | 153 |
| 108 | Meta-analysis of the Association Between Variants in SORL1 and Alzheimer Disease. Archives of Neurology, 2011, 68, 99. | 4.9 | 153 |

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|---|---|---|---|
| 109 | Two divergent types of nerve pathology in patients with different P sub 0 mutations in Charcot-Marie-Tooth disease. Neurology, 1996, 47, 761-765. | 1.5 | 152 |
| 110 | Mutations in GDAP1. Neurology, 2002, 59, 1865-1872. | 1.5 | 152 |
| 111 | The gene for autosomal dominant cerebellar ataxia with pigmentary macular dystrophy maps to chromosome 3p12â€“p21.1. Nature Genetics, 1995, 10, 84-88. | 9.4 | 151 |
| 112 | Current insights into the C9orf72 repeat expansion diseases of the FTLD/ALS spectrum. Trends in Neurosciences, 2013, 36, 450-459. | 4.2 | 151 |
| 113 | Loss of TBK1 is a frequent cause of frontotemporal dementia in a Belgian cohort. Neurology, 2015, 85, 2116-2125. | 1.5 | 151 |
| 114 | Novel missense mutation in the early growth response 2 gene associated with Dejerineâ€“Sottas syndrome phenotype. Neurology, 1999, 52, 1827-1827. | 1.5 | 150 |
| 115 | Variability of 5-HT2C receptor cys23ser polymorphism among European populations and vulnerability to affective disorder. Molecular Psychiatry, 2001, 6, 579-585. | 4.1 | 150 |
| 116 | A deletion in SCN1B is associated with febrile seizures and early-onset absence epilepsy. Neurology, 2003, 61, 854-856. | 1.5 | 150 |
| 117 | Detection of expanded CAG repeats in Bipolar Affective Disorder using the repeat expansion detection (RED) method. Neurobiology of Disease, 1995, 2, 55-62. | 2.1 | 148 |
| 118 | Progranulin genetic variability contributes to amyotrophic lateral sclerosis. Neurology, 2008, 71, 253-259. | 1.5 | 148 |
| 119 | Novel POLG mutations in progressive external ophthalmoplegia mimicking mitochondrial neurogastrointestinal encephalomyopathy. European Journal of Human Genetics, 2003, 11, 547-549. | 1.4 | 145 |
| 120 | A Genomewide Screen for Late-Onset Alzheimer Disease in a Genetically Isolated Dutch Population. American Journal of Human Genetics, 2007, 81, 17-31. | 2.6 | 145 |
| 121 | Alzheimer and Parkinson Diagnoses in Progranulin Null Mutation Carriers in an Extended Founder Family. Archives of Neurology, 2007, 64, 1436. | 4.9 | 143 |
| 122 | Structural variants identified by Oxford Nanopore PromethION sequencing of the human genome. Genome Research, 2019, 29, 1178-1187. | 2.4 | 143 |
| 123 | A novel GABRG2 mutation associated with febrile seizures. Neurology, 2006, 67, 687-690. | 1.5 | 142 |
| 124 | Earlyâ€onset Alzheimer's disease in 2 large Belgian families. Neurology, 1991, 41, 62-62. | 1.5 | 141 |
| 125 | Common variants in Alzheimerâ€™s disease and risk stratification by polygenic risk scores. Nature Communications, 2021, 12, 3417. | 5.8 | 140 |
| 126 | Glucocorticoid Receptor Gene-Based SNP Analysis in Patients with Recurrent Major Depression. Neuropsychopharmacology, 2006, 31, 620-627. | 2.8 | 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. | 1.4 | 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. | 1.8 | 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-? concentrations. Annals of Neurology, 2000, 48, 806-808. | 2.8 | 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. | 1.4 | 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. | 4.1 | 134 |
| 132 | Reduced functional brain activity response in cognitively intact apolipoprotein E Îµ4 carriers. Brain, 2006, 129, 1240-1248. | 3.7 | 133 |
| 133 | Lrrk2 R1441C parkinsonism is clinically similar to sporadic Parkinson disease. Neurology, 2008, 70, 1456-1460. | 1.5 | 132 |
| 134 | The role of ABCA7 in Alzheimerâ€™s disease: evidence from genomics, transcriptomics and methylomics. Acta Neuropathologica, 2019, 138, 201-220. | 3.9 | 132 |
| 135 | Molecular genetics of Alzheimer's disease. Annals of Medicine, 1998, 30, 560-565. | 1.5 | 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. | 1.4 | 131 |
| 137 | Origin of the de novo duplication in Charcot â€” Marie â€” Tooth disease type 1A: unequal nonsister chromatid exchange during spermatogenesis. Human Molecular Genetics, 1993, 2, 2031-2035. | 1.4 | 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. | 1.0 | 130 |
| 139 | The apolipoprotein E ?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. | 2.8 | 129 |
| 140 | Granulin mutations associated with frontotemporal lobar degeneration and related disorders: An update. Human Mutation, 2008, 29, 1373-1386. | 1.1 | 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. | 3.3 | 126 |
| 142 | Spinocerebellar ataxia type 7 associated with pigmentary retinal dystrophy. European Journal of Human Genetics, 2004, 12, 2-15. | 1.4 | 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. | 4.9 | 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. | 1.5 | 123 |

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|---|---|---|---|
| 145 | TMEM106B is a genetic modifier of frontotemporal lobar degeneration with C9orf72 hexanucleotide repeat expansions. Acta Neuropathologica, 2014, 127, 407-418. | 3.9 | 123 |
| 146 | The pursuit of susceptibility genes for Alzheimer's disease: progress and prospects. Trends in Genetics, 2010, 26, 84-93. | 2.9 | 122 |
| 147 | Pathological mechanisms underlying TDP-43 driven neurodegeneration in FTLD-ALS spectrum disorders. Human Molecular Genetics, 2013, 22, R77-R87. | 1.4 | 122 |
| 148 | Autosornal recessive hereditary motor and sensory neuropathy with focally folded myelin sheaths. Neurology, 1996, 46, 1318-1318. | 1.5 | 121 |
| 149 | Rescue of Progranulin Deficiency Associated with Frontotemporal Lobar Degeneration by Alkalizing Reagents and Inhibition of Vacuolar ATPase. Journal of Neuroscience, 2011, 31, 1885-1894. | 1.7 | 121 |
| 150 | Large-scale replication and heterogeneity in Parkinson disease genetic loci. Neurology, 2012, 79, 659-667. | 1.5 | 119 |
| 151 | A blinded international study on the reliability of genetic testing for GGGGCC-repeat expansions inC9orf72reveals marked differences in results among 14 laboratories. Journal of Medical Genetics, 2014, 51, 419-424. | 1.5 | 118 |
| 152 | A Genome Wide Search for Susceptibility Loci in Three European Malignant Hyperthermia Pedigrees. Human Molecular Genetics, 1997, 6, 953-961. | 1.4 | 117 |
| 153 | Câ€terminal neurogranin is increased in cerebrospinal fluid but unchanged in plasma in Alzheimer's disease. Alzheimer's and Dementia, 2015, 11, 1461-1469. | 0.4 | 117 |
| 154 | Mutations other than null mutations producing a pathogenic loss of progranulin in frontotemporal dementia. Human Mutation, 2007, 28, 416-416. | 1.1 | 116 |
| 155 | Transmission and ageâ€atâ€onset patterns in familial Alzheimer's disease. Neurology, 1990, 40, 395-395. | 1.5 | 115 |
| 156 | Estrogen use and early onset Alzheimer's disease: a population-based study. Journal of Neurology, Neurosurgery and Psychiatry, 1999, 67, 779-781. | 0.9 | 114 |
| 157 | Relationship between C9orf72 repeat size and clinical phenotype. Current Opinion in Genetics and Development, 2017, 44, 117-124. | 1.5 | 114 |
| 158 | WT1 MUTATION IN MALIGNANT MESOTHELIOMA AND WT1 IMMUNOREACTIVITY IN RELATION TOp53 AND GROWTH FACTOR RECEPTOR EXPRESSION, CELL-TYPE TRANSITION, AND PROGNOSIS. , 1997, 181, 67-74. | | 112 |
| 159 | The influence of apoe status on episodic and semantic memory: Data from a population-based study.. Neuropsychology, 2006, 20, 645-657. | 1.0 | 112 |
| 160 | Reduced hippocampal volume in non-demented carriers of the apolipoprotein E É›4: Relation to chronological age and recognition memory. Neuroscience Letters, 2006, 396, 23-27. | 1.0 | 112 |
| 161 | Amyloid precursor protein gene mutation in early-onset Alzheimer's disease. Lancet, The, 1991, 337, 978. | 6.3 | 110 |
| 162 | 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, 653-657. | 1.8 | 110 |

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|---|---|---|---|
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