# Supplemental Material

## A1: GWAS Catalog study information

Table 5 Overview of studies used for gathering transcriptomic data AD = Alzheimer’s disease, PD = Parkinson’s disease, (s)ALS =(sporadic) Amyotrophic Lateral Sclerosis, HD = Huntington’s disease, ctrl = control, CSF = cerebrospinal fluid, BA = Brodman Area

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| **study** | **Participants** | **comparison** | **Paper** | **disease** |
| (Nazarian, Yashin and Kulminski, 2019) | 952 European ancestry male cases, 1,789 European ancestry female cases, 6,337 European ancestry male controls, 8,402 European ancestry female controls | NA | Genome-wide analysis of genetic predisposition to Alzheimer’s disease and related sex disparities. | Alzheimer’s disease |
| (Nazarian *et al.*, 2019) | 796 European ancestry cases, 4,010 European ancestry controls | NA | Genetic heterogeneity of Alzheimer’s disease in subjects with and without hypertension. | Alzheimer’s disease |
| (Beecham *et al.*, 2009) | 492 European ancestry cases, 496 European ancestry controls | 238 European ancestry cases, 220 European ancestry controls | Genome-wide association study implicates a chromosome 12 risk locus for late-onset Alzheimer disease. | Alzheimer’s disease |
| (Carrasquillo *et al.*, 2009) | 844 European ancestry cases, 1,255 European ancestry controls | 1,547 European ancestry cases, 1,209 European ancestry controls | Genetic variation in *PCDH11X* is associated with susceptibility to late-onset Alzheimer’s disease. | Alzheimer’s disease |
| (Abraham *et al.*, 2008) | 1,082 European ancestry cases, 1,239 European ancestry controls | 1,400 controls | A genome-wide association study for late-onset Alzheimer’s disease using DNA pooling. | Alzheimer’s disease |
| (Webster *et al.*, 2007) | 664 cases, 422 controls | NA | *SORL1* as an Alzheimer’s disease predisposition gene? | Alzheimer’s disease |
| (Coon *et al.*, 2007) | 664 European ancestry cases, 422 European ancestry controls | NA | A high-density whole-genome association study reveals that *APOE* is the major susceptibility gene for sporadic late-onset Alzheimer’s disease. | Alzheimer’s disease |
| (Heinzen *et al.*, 2010) | 331 European ancestry cases, 368 European ancestry controls | NA | Genome-wide scan of copy number variation in late-onset Alzheimer’s disease. | Alzheimer’s disease |
| (Naj *et al.*, 2010) | 931 cases, 1,104 controls | 1,338 cases, 2,003 controls | Dementia revealed: novel chromosome 6 locus for late-onset Alzheimer disease provides genetic evidence for folate-pathway abnormalities. | Alzheimer’s disease |
| (Logue *et al.*, 2011) | 513 African American cases, 496 African American controls | NA | A comprehensive genetic association study of Alzheimer disease in African Americans. | Alzheimer’s disease |
| (Hu *et al.*, 2011) | 1,831 European ancestry cases, 1,764 European ancestry controls | 751 cases, 751 controls | Meta-analysis for genome-wide association study identifies multiple variants at the BIN1 locus associated with late-onset Alzheimer’s disease. | Alzheimer’s disease |
| (Sherva *et al.*, 2014) | 303 European ancestry cases |  | Genome-wide association study of the rate of cognitive decline in Alzheimer’s disease. | Alzheimer’s disease |
| (Lo *et al.*, 2019) | 2,399 European ancestry cases, 4,160 European ancestry controls | NA | Identification of genetic heterogeneity of Alzheimer’s disease across age. | Alzheimer’s disease |
| (Zhu *et al.*, 2019) | 54,162 European ancestry cases, 58,047 European ancestry individuals | NA | Shared genetic architecture between metabolic traits and Alzheimer’s disease: a large-scale genome-wide cross-trait analysis. | Alzheimer’s disease |
| (Feulner *et al.*, 2010) | 491 European ancestry cases, 479 European ancestry controls | NA | Examination of the current top candidate genes for AD in a genome-wide association study. | Alzheimer’s disease |
| (Kramer *et al.*, 2011) | 185 European ancestry low NFT individuals, 114 European ancestry high NFT individuals | NA | Alzheimer disease pathology in cognitively healthy elderly: a genome-wide study. | Alzheimer’s disease |
| (Meda *et al.*, 2012) | 367 European ancestry individuals with mild cognitive impairment, 181 European ancestry individuals with mild early-stage LOAD, 209 European ancestry controls | NA | A large scale multivariate parallel ICA method reveals novel imaging-genetic relationships for Alzheimer’s disease in the ADNI cohort. | Alzheimer’s disease |
| (Kamboh, Barmada, *et al.*, 2012) | 1,190 European ancestry cases, 1,032 cases | NA | Genome-wide association analysis of age-at-onset in Alzheimer’s disease. | Alzheimer’s disease |
| (Chung *et al.*, 2018) | 190 European ancestry Alzheimer’s disease dementia cases | NA | Genome-wide association study of Alzheimer’s disease endophenotypes at prediagnosis stages. | Alzheimer’s disease |
| (Nazarian *et al.*, 2019) | 1,262 European ancestry cases, 9,608 European ancestry controls | NA | Genetic heterogeneity of Alzheimer’s disease in subjects with and without hypertension. | Alzheimer’s disease |
| (Herold *et al.*, 2016) | 2,478 European ancestry cases, 979 ancestry controls both from the same 1,070 families | NA | Family-based association analyses of imputed genotypes reveal genome-wide significant association of Alzheimer’s disease with OSBPL6, PTPRG, and PDCL3. | Alzheimer’s disease |
| (Cummings *et al.*, 2012) | 109 Amish cases, 689 Amish controls | NA | Genome-wide association and linkage study in the Amish detects a novel candidate late-onset Alzheimer disease gene. | Alzheimer’s disease |
| (Tosto *et al.*, 2015) | 2,451 Caribbean Hispanic cases, 2,063 Caribbean Hispanic controls | 550 Caribbean Hispanic cases, 236 Caribbean Hispanic controls | F-box/LRR-repeat protein 7 is genetically associated with Alzheimer’s disease. | Alzheimer’s disease |
| (Pérez-Palma *et al.*, 2014) | 2,540 European ancestry cases, 2,029 European ancestry controls | NA | Overrepresentation of glutamate signaling in Alzheimer’s disease: network-based pathway enrichment using meta-analysis of genome-wide association studies. | Alzheimer’s disease |
| (Wang *et al.*, 2015) | 983 cases | NA | Genetic Determinants of Survival in Patientswith Alzheimer’s Disease. | Alzheimer’s disease |
| (Yashin *et al.*, 2018) | 12,169 European ancestry individuals, 2,078 Black individuals, 4,053 unknown ancestry individuals | NA | Hidden heterogeneity in Alzheimer’s disease: Insights from genetic association studies and other analyses. | Alzheimer’s disease |
| (Reiman *et al.*, 2007) | 446 cases, 290 controls | 415 cases, 260 controls | GAB2 alleles modify Alzheimer’s risk in APOE epsilon4 carriers. | Alzheimer’s disease |
| (Kim *et al.*, 2011) | 96 European ancestry Alzheimer disease cases, 176 European ancestry individuals with mild cognitive impairment, 102 European ancestry controls | NA | Genome-wide association study of CSF biomarkers Abeta1-42, t-tau, and p-tau181p in the ADNI cohort. | Alzheimer’s disease |
| (Kamboh, Demirci, *et al.*, 2012) | 1,291 European ancestry cases, 938 European ancestry controls | 509 European ancestry cases, 753 European ancestry controls, 2,218 cases, 2,583 controls | Genome-wide association study of Alzheimer’s disease. | Alzheimer’s disease |
| (Stein *et al.*, 2010) | 173 European ancestry Alzheimer’s disease cases, 361 European ancestry Mild Cognitive Impairment cases, 208 European ancestry controls | NA | Genome-wide analysis reveals novel genes influencing temporal lobe structure with relevance to neurodegeneration in Alzheimer’s disease. | Alzheimer’s disease |
| (Martinelli-Boneschi *et al.*, 2013) | 92 European ancestry cases, 77 European ancestry controls | 94 European ancestry cases, 74 European ancestry controls | Pharmacogenomics in Alzheimer’s disease: a genome-wide association study of response to cholinesterase inhibitors. | Alzheimer’s disease |
| (Bertram *et al.*, 2008) | 941 European ancestry cases and 404 European ancestry controls from 410 families | 1,767 European ancestry cases and 838 European ancestry controls from 875 families | Genome-wide association analysis reveals putative Alzheimer’s disease susceptibility loci in addition to APOE. | Alzheimer’s disease |
| (Lo *et al.*, 2019) | 7,316 European ancestry cases, 7,579 European ancestry controls | NA | Identification of genetic heterogeneity of Alzheimer’s disease across age. | Alzheimer’s disease |
| (Ramanan *et al.*, 2014) | 555 European ancestry individuals | NA | APOE and BCHE as modulators of cerebral amyloid deposition: a florbetapir PET genome-wide association study. | Alzheimer’s disease |
| (Chung *et al.*, 2018) | 190 European ancestry AD dementia cases | NA | Genome-wide association study of Alzheimer’s disease endophenotypes at prediagnosis stages. | Alzheimer’s disease |
| (Jonsson *et al.*, 2013) | 3,550 European ancestry cases, 8,888 European ancestry controls | 694 European ancestry cases, 4,375 European ancestry controls, 1,343 cases, 5,352 controls | Variant of TREM2 associated with the risk of Alzheimer’s disease. | Alzheimer’s disease |
| (Lee *et al.*, 2017) | 242 European ancestry individuals | NA | Single-nucleotide polymorphisms are associated with cognitive decline at Alzheimer’s disease conversion within mild cognitive impairment patients. | Alzheimer’s disease |
| (Marioni *et al.*, 2018) | up to 42,034 British ancestry individuals with parental history of Alzheimer’s disease, at least 272,244 British ancestry individuals with no parental history of Alzheimer’s disease, 25,580 Alzheimer’s disease cases, 48,466 controls | NA | GWAS on family history of Alzheimer’s disease. | Alzheimer’s disease |
| (Traylor *et al.*, 2016) | 17,008 European ancestry Alzheimer’s disease cases, 3,651 European ancestry small vessel stroke cases, 95,811 European ancestry controls | NA | Shared genetic contribution to Ischaemic Stroke and Alzheimer’s Disease. | Alzheimer’s disease |
| (Furney *et al.*, 2011) | 424 European ancestry mild cognitive impairment cases, 236 European ancestry Alzheimer’s disease cases, 279 European ancestry controls | NA | Genome-wide association with MRI atrophy measures as a quantitative trait locus for Alzheimer’s disease. | Alzheimer’s disease |
| (Hirano *et al.*, 2015) | 489 Japanese ancestry APOE-ε4 noncarrier cases, 6,463 Japanese ancestry APOE-ε4 noncarrier controls, 323 Japanese ancestry APOE-ε4 carrier cases, 1,484 Japanese ancestry APOE-ε4 carrier controls, 4 Japanese ancestry cases, 45 Japanese ancestry controls | 528 Japanese ancestry APOE-ε4 noncarrier cases, 5,824 Japanese ancestry APOE-ε4 noncarrier controls, 480 Japanese ancestry APOE-ε4 carrier cases, 1,364 Japanese ancestry APOE-ε4 carrier controls, 3 Japanese ancestry cases, 24 Japanese ancestry controls | A genome-wide association study of late-onset Alzheimer’s disease in a Japanese population. | Alzheimer’s disease |
| (Wijsman *et al.*, 2011) | 1,848 European ancestry affected individuals, 1,991 European ancestry unaffected individuals | 231 Caribbean Hispanic cases, 187 Caribbean Hispanic controls, 386 European ancestry cases, 386 European ancestry controls | Genome-wide association of familial late-onset Alzheimer’s disease replicates BIN1 and CLU and nominates CUGBP2 in interaction with APOE. | Alzheimer’s disease |
| (Kauwe *et al.*, 2014) | 574 individuals | NA | Genome-wide association study of CSF levels of 59 alzheimer’s disease candidate proteins: significant associations with proteins involved in amyloid processing and inflammation. | Alzheimer’s disease |
| (Deming *et al.*, 2016) | 123 Alzheimer’s disease cases, 270 controls | NA | A potential endophenotype for Alzheimer’s disease: cerebrospinal fluid clusterin. | Alzheimer’s disease |
| (Mukherjee *et al.*, 2018) | Up to 1,107 European ancestry cases, 3,447 European ancestry controls | NA | Genetic data and cognitively defined late-onset Alzheimer’s disease subgroups. | Alzheimer’s disease |
| (Antúnez *et al.*, 2011) | 319 European ancestry cases, 769 European ancestry controls, 2,690 cases, 2,237 controls | 4,982 European ancestry cases, 7,961 European ancestry controls, 2,190 cases, 3,374 controls | The membrane-spanning 4-domains, subfamily A (MS4A) gene cluster contains a common variant associated with Alzheimer’s disease. | Alzheimer’s disease |
| (Lambert *et al.*, 2009) | 2,032 European ancestry cases, 5,328 European ancestry controls | 3,978 European ancestry cases, 3,297 European ancestry controls | Genome-wide association study identifies variants at CLU and CR1 associated with Alzheimer’s disease. | Alzheimer’s disease |
| (Li *et al.*, 2008) | 753 European ancestry cases, 736 European ancestry controls | 418 European ancestry cases, 249 European ancestry controls | Candidate single-nucleotide polymorphisms from a genomewide association study of Alzheimer disease. | Alzheimer’s disease |
| (Miyashita *et al.*, 2013) | 891 Japanese ancestry cases, 844 Japanese ancestry controls | 1,224 East Asian ancestry cases, 2,114 East Asian ancestry controls, 11,840 European ancestry cases, 10,931 European ancestry controls | SORL1 is genetically associated with late-onset Alzheimer’s disease in Japanese, Koreans and Caucasians. | Alzheimer’s disease |
| (Cruchaga *et al.*, 2013) | 591 European ancestry cases, 687 European ancestry controls |  | GWAS of cerebrospinal fluid tau levels identifies risk variants for Alzheimer’s disease. | Alzheimer’s disease |
| (Reitz *et al.*, 2013) | 1,968 African American cases, 3,928 African American controls |  | Variants in the ATP-binding cassette transporter (ABCA7), apolipoprotein E ϵ4,and the risk of late-onset Alzheimer disease in African Americans. | Alzheimer’s disease |
| (Hollingworth *et al.*, 2012) | 1,039 European ancestry cases with psychosis, 5,659 European ancestry controls, 260 European, African American and Native American ancestry cases with psychosis from 264 families | NA | Genome-wide association study of Alzheimer’s disease with psychotic symptoms. | Alzheimer’s disease |
| (Seshadri *et al.*, 2010) | 973 incident AD cases, 2,033 prevalent AD cases, 22,604 controls of European and Hispanic ancestry | 6,505 European ancestry cases, 13,532 European ancestry controls | Genome-wide analysis of genetic loci associated with Alzheimer disease. | Alzheimer’s disease |
| (Gusareva *et al.*, 2018) | 788 male cases, 1,455 female cases, 2,362 male controls, 3,655 female controls | 3,836 male cases, 6,244 female cases, 8,618 male controls, 11,624 female controls | Male-specific epistasis between WWC1 and TLN2 genes is associated with Alzheimer’s disease. | Alzheimer’s disease |
| (Nelson *et al.*, 2014) | 1443 cases and 99 controls | NA | ABCC9 gene polymorphism is associated with hippocampal sclerosis of aging pathology. | Alzheimer’s disease |
| (Jansen *et al.*, 2019) | 24,087 European ancestry late-onset Alzheimer’s disease cases, 47,793 European ancestry individuals with family history of Alzheimer’s disease, 383,378 European ancestry controls | NA | Genome-wide meta-analysis identifies new loci and functional pathways influencing Alzheimer’s disease risk. | Alzheimer’s disease |
| (Moreno-Grau *et al.*, 2019) | 4,120 European ancestry cases, 3,289 European ancestry controls | NA | Genome-wide association analysis of dementia and its clinical endophenotypes reveal novel loci associated with Alzheimer’s disease and three causality networks: The GR@ACE project. | Alzheimer’s disease |
| (Schott *et al.*, 2016) | 293 cases, 10,547 healthy controls | NA | Genetic risk factors for the posterior cortical atrophy variant of Alzheimer’s disease. | Alzheimer’s disease |
| (Ramirez *et al.*, 2014) | 363 European ancestry individuals | 515 individuals | SUCLG2 identified as both a determinator of CSF Aβ1-42 levels and an attenuator of cognitive decline in Alzheimer’s disease. | Alzheimer’s disease |
| (Jun *et al.*, 2017) | 13,100 European ancestry cases, 13,220 European ancestry controls, 1,472 African American cases, 3,511 African American controls, 951 Japanese ancestry cases, 894 Japanese ancestry controls, 51 Israeli-Arab ancestry cases, 64 Israeli-Arab ancestry controls | 5,813 European ancestry cases, 20,474 European ancestry controls | Transethnic genome-wide scan identifies novel Alzheimer’s disease loci. | Alzheimer’s disease |
| (Mukherjee *et al.*, 2018) | Up to 510 European ancestry cases, 3,447 European ancestry controls | NA | Genetic data and cognitively defined late-onset Alzheimer’s disease subgroups. | Alzheimer’s disease |
| (Harold *et al.*, 2009) | 3,941 European ancestry cases, 7,848 European ancestry controls | 2,023 European ancestry cases, 2,340 European ancestry controls | Genome-wide association study identifies variants at CLU and PICALM associated with Alzheimer’s disease. | Alzheimer’s disease |
| (J. C. Lambert *et al.*, 2013) | 2,025 European ancestry cases, 5,328 European ancestry controls | 7,913 European ancestry cases, 10,417 European ancestry controls | Genome-wide haplotype association study identifies the FRMD4A gene as a risk locus for Alzheimer’s disease. | Alzheimer’s disease |
| (Jansen *et al.*, 2019) | 24,087 European ancestry late-onset Alzheimer’s disease cases, 47,793 European ancestry individuals with family history of Alzheimer’s disease, 383,378 European ancestry controls | NA | Genome-wide meta-analysis identifies new loci and functional pathways influencing Alzheimer’s disease risk. | Alzheimer’s disease |
| (Jun *et al.*, 2016) | 7,184 cases, 26,968 controls | 718 European ancestry cases, 1,699 European ancestry controls | A novel Alzheimer disease locus located near the gene encoding tau protein. | Alzheimer’s disease |
| (Hollingworth *et al.*, 2011) | 6,688 European ancestry cases, 13,685 European ancestry controls | 13,182 European ancestry cases, 26,161 European ancestry controls | Common variants at ABCA7, MS4A6A/MS4A4E, EPHA1, CD33 and CD2AP are associated with Alzheimer’s disease. | Alzheimer’s disease |
| (Naj *et al.*, 2011) | 8,309 European ancestry cases, 7,366 European ancestry controls | 10,523 European ancestry cases, 28,231 European ancestry controls | Common variants at MS4A4/MS4A6E, CD2AP, CD33 and EPHA1 are associated with late-onset Alzheimer’s disease. | Alzheimer’s disease |
| (Jean Charles Lambert *et al.*, 2013) | 17,008 European ancestry cases, 37,154 European ancestry controls | 8,572 European ancestry cases, 11,312 European ancestry controls | Meta-analysis of 74,046 individuals identifies 11 new susceptibility loci for Alzheimer’s disease. | Alzheimer’s disease |
| (Chung *et al.*, 2012) | 443 European ancestry cases | NA | Genomic determinants of motor and cognitive outcomes in Parkinson’s disease. | Parkinson’s disease |
| (Wallen *et al.*, 2018) | 1,950 European ancestry cases | 726 European ancestry cases | Plasticity-related gene 3 (LPPR1) and age at diagnosis of Parkinson disease. | Parkinson’s disease |
| (Hu *et al.*, 2016) | 250 Han Chinese ancestry cases, 250 Han Chinese ancestry controls | NA | A Pooling Genome-Wide Association Study Combining a Pathway Analysis for Typical Sporadic Parkinson’s Disease in the Han Population of Chinese Mainland. | Parkinson’s disease |
| (Pickrell *et al.*, 2016) | 9,619 European ancestry cases, 324,522 European ancestry controls | NA | Detection and interpretation of shared genetic influences on 42 human traits. | Parkinson’s disease |
| (Nalls *et al.*, 2011) | 5,333 European ancestry cases, 12,019 European ancestry controls | 7,053 cases, 9,007 controls | Imputation of sequence variants for identification of genetic risks for Parkinson’s disease: a meta-analysis of genome-wide association studies. | Parkinson’s disease |
| (Hamza *et al.*, 2010) | 2,000 European ancestry cases, 1,986 European ancestry controls | Up to 1,447 cases, 1,468 controls | Common genetic variation in the HLA region is associated with late-onset sporadic Parkinson’s disease. | Parkinson’s disease |
| (Maraganore *et al.*, 2005) | 381 European ancestry cases, 363 European ancestry controls, 62 cases,79 controls, 1 Asian ancestry control from 443 sibships | 269 European ancestry cases, 272 European ancestry controls, 62 cases, 60 controls, 1 Asian ancestry case | High-resolution whole-genome association study of Parkinson disease. | Parkinson’s disease |
| (Fung *et al.*, 2006) | 267 European ancestry cases, 270 European ancestry controls | NA | Genome-wide genotyping in Parkinson’s disease and neurologically normal controls: first stage analysis and public release of data. | Parkinson’s disease |
| (Satake *et al.*, 2009) | 988 Japanese ancestry cases, 2,521 Japanese ancestry controls | 933 Japanese ancestry cases, 15,753 Japanese ancestry controls | Genome-wide association study identifies common variants at four loci as genetic risk factors for Parkinson’s disease. | Parkinson’s disease |
| (Pankratz *et al.*, 2009) | 1,119 European ancestry cases, 1,127 European ancestry controls | NA | Genomewide association study for susceptibility genes contributing to familial Parkinson disease. | Parkinson’s disease |
| (Edwards *et al.*, 2010) | 1,752 European ancestry cases, 1,745 European ancestry controls | NA | Genome-wide association study confirms SNPs in SNCA and the MAPT region as common risk factors for Parkinson disease. | Parkinson’s disease |
| (Do *et al.*, 2011) | 3,426 European ancestry cases, 29,624 European ancestry controls | NA | Web-based genome-wide association study identifies two novel loci and a substantial genetic component for Parkinson’s disease. | Parkinson’s disease |
| (Liu *et al.*, 2011) | 268 Ashkenazi Jewish cases, 178 Ashkenazi Jewish controls | 1,782 European ancestry cases, 1,658 European ancestry controls | Genome-wide association study identifies candidate genes for Parkinson’s disease in an Ashkenazi Jewish population. | Parkinson’s disease |
| (Hill-Burns *et al.*, 2014) | 1,565 European ancestry sproadic Parkinson’s disease cases, 435 European ancestry familial Parkinson’s disease cases, 1,986 European ancestry controls | 1,528 European ancestry sporadic Parkinson’s disease cases, 707 European ancestry familial Parkinson’s disease cases, 796 European ancestry controls | Identification of a novel Parkinson’s disease locus via stratified genome-wide association study. | Parkinson’s disease |
| (Davis *et al.*, 2013) | 31 Amish cases, 767 Amish controls | NA | Parkinson disease loci in the mid-western Amish. | Parkinson’s disease |
| (Biernacka *et al.*, 2016) | 364 cases, 364 sibling controls | NA | Genome-wide gene-environment interaction analysis of pesticide exposure and risk of Parkinson’s disease. | Parkinson’s disease |
| (Chang *et al.*, 2017) | 20,184 European ancestry cases, 397,324 European ancestry controls | 5,851 European ancestry cases, 5,866 European ancestry controls | A meta-analysis of genome-wide association studies identifies 17 new Parkinson’s disease risk loci. | Parkinson’s disease |
| (Logue *et al.*, 2011) | 250 Han Chinese ancestry cases, 250 Han Chinese ancestry controls | NA | A Pooling Genome-Wide Association Study Combining a Pathway Analysis for Typical Sporadic Parkinson’s Disease in the Han Population of Chinese Mainland. | Parkinson’s disease |
| (Hill-Burns *et al.*, 2016) | 431 European ancestry cases | 737 European ancestry cases | Identification of genetic modifiers of age-at-onset for familial Parkinson’s disease. | Parkinson’s disease |
| (Nalls *et al.*, 2011) | 5,333 European ancestry cases, 12,019 European ancestry controls | 7,053 cases, 9,007 controls | Imputation of sequence variants for identification of genetic risks for Parkinson’s disease: a meta-analysis of genome-wide association studies. | Parkinson’s disease |
| (Saad *et al.*, 2011) | 1,039 European ancestry cases, 1,984 European ancestry controls | 3,232 European ancestry cases, 7,064 European ancestry controls | Genome-wide association study confirms BST1 and suggests a locus on 12q24 as the risk loci for Parkinson’s disease in the European population. | Parkinson’s disease |
| (Hamza *et al.*, 2010) | 2,000 European ancestry cases, 1,986 European ancestry controls | Up to 1,447 cases, 1,468 controls | Common genetic variation in the HLA region is associated with late-onset sporadic Parkinson’s disease. | Parkinson’s disease |
| (Pankratz *et al.*, 2012) | 4,238 European ancestry cases, 4,239 European ancestry controls | 3,738 European ancestry cases, 2,111 European ancestry controls | Meta-analysis of Parkinson’s disease: identification of a novel locus, RIT2. | Parkinson’s disease |
| (Latourelle *et al.*, 2009) | 857 European ancestry familial cases, 440 idiopathic cases | 747 European ancestry idiopathic cases | Genomewide association study for onset age in Parkinson disease. | Parkinson’s disease |
| (Vacic *et al.*, 2014) | 1,130 Ashkenazi Jewish cases, 2,611 Ashkenazi Jewish controls | 306 Ashkenazi Jewish cases, 2,583 Ashkenazi Jewish controls | Genome-wide mapping of IBD segments in an Ashkenazi PD cohort identifies associated haplotypes. | Parkinson’s disease |
| (Foo *et al.*, 2017) | 779 Han Chinese ancestry cases, 13,227 Han Chinese ancestry controls | 5,125 Asian ancestry cases, 17,604 Asian ancestry controls | Genome-wide association study of Parkinson’s disease in East Asians. | Parkinson’s disease |
| (Beecham *et al.*, 2015) | 484 European ancestry cases, 1,145 European ancestry controls | NA | PARK10 is a major locus for sporadic neuropathologically confirmed Parkinson disease. | Parkinson’s disease |
| (Simón-Sánchez *et al.*, 2009) | 1,713 European ancestry cases, 3,978 European ancestry controls | 3,361 European ancestry cases, 4,573 European ancestry controls | Genome-wide association study reveals genetic risk underlying Parkinson’s disease. | Parkinson’s disease |
| (Nalls *et al.*, 2019) | 15,056 European ancestry cases, 18,618 European ancestry proxy cases, 449,056 European ancestry controls | 22,632 European ancestry cases, 968,735 European ancestry controls | Identification of novel risk loci, causal insights, and heritable risk for Parkinson’s disease: a meta-analysis of genome-wide association studies. | Parkinson’s disease |
| (Blauwendraat *et al.*, 2020) | 1,588 European ancestry cases, 7,584 European ancestry controls | up to 1,194 European ancestry cases, up to 13,901 European ancestry controls | Genetic modifiers of risk and age at onset in GBA associated Parkinson’s disease and Lewy body dementia. | Parkinson’s disease |
| (Spencer *et al.*, 2011) | 1,705 European ancestry cases, 5,175 European ancestry controls | 1,039 European ancestry cases, 1,984 European ancestry controls | Dissection of the genetics of Parkinson’s disease identifies an additional association 5&apos; of SNCA and multiple associated haplotypes at 17q21. | Parkinson’s disease |
| (Lill *et al.*, 2012) | 2,197 cases, 2,061 controls | Up to 98,080 European and Asian ancestry individuals | Comprehensive research synopsis and systematic meta-analyses in Parkinson’s disease genetics: The PDGene database. | Parkinson’s disease |
| (Blauwendraat *et al.*, 2019) | 17,996 cases | 10,572 cases | Parkinson’s disease age at onset genome-wide association study: Defining heritability, genetic loci, and α-synuclein mechanisms. | Parkinson’s disease |
| (Nalls *et al.*, 2014) | 13,708 European ancestry cases, 95,282 European ancestry controls | 5,353 European ancestry cases, 5,551 European ancestry controls | Large-scale meta-analysis of genome-wide association data identifies six new risk loci for Parkinson’s disease. | Parkinson’s disease |
| (Bandres‐Ciga *et al.*, 2019) | 3,997 Spanish ancestry individuals | NA | The Genetic Architecture of Parkinson Disease in Spain: Characterizing Population-Specific Risk, Differential Haplotype Structures, and Providing Etiologic Insight. | Parkinson’s disease |
| (Pottier *et al.*, 2018) | 184 European ancestry cases, 198 European ancestry controls | NA | Potential genetic modifiers of disease risk and age at onset in patients with frontotemporal lobar degeneration and GRN mutations: a genome-wide association study. | Parkinson’s disease |
| (Chao *et al.*, 2018) | 374 Venezuelan ancestry individuals, 4,061 European ancestry individuals | NA | Population-specific genetic modification of Huntington’s disease in Venezuela. | Huntington’s disease |
| (Moss *et al.*, 2017) | 1,989 European and unknown ancestry mutation carriers | NA | Identification of genetic variants associated with Huntington’s disease progression: a genome-wide association study. | Huntington’s disease |
| (Cronin *et al.*, 2009) | 958 European ancestry cases, 932 European ancestry controls | 309 European ancestry cases, 404 European ancestry controls | Screening for replication of genome-wide SNP associations in sporadic ALS. | Amyotrophic lateral sclerosis |
| (Chen *et al.*, 2016) | 94 Taiwanese Han ancestry cases, 376 Taiwanese Han ancestry controls | NA | A genome-wide association study on amyotrophic lateral sclerosis in the Taiwanese Han population. | Amyotrophic lateral sclerosis |
| (McLaughlin *et al.*, 2015) | 25 European ancestry C9orf72-positive cases, 1,179 European ancestry controls | NA | Second-generation Irish genome-wide association study for amyotrophic lateral sclerosis. | Amyotrophic lateral sclerosis |
| (Schymick *et al.*, 2007) | 276 European ancestry cases, 271 European ancestry controls | NA | Genome-wide genotyping in amyotrophic lateral sclerosis and neurologically normal controls: first stage analysis and public release of data. | Amyotrophic lateral sclerosis |
| (Cronin *et al.*, 2008) | 221 Genetically Homogenous Irish cases, 211 Genetically Homogenous Irish controls | 737 European ancestry cases, 721 European ancestry controls | A genome-wide association study of sporadic ALS in a homogenous Irish population. | Amyotrophic lateral sclerosis |
| (van Es *et al.*, 2007) | 461 European ancestry cases, 450 European ancestry controls | 876 European ancestry cases, 906 European ancestry controls | ITPR2 as a susceptibility gene in sporadic amyotrophic lateral sclerosis: a genome-wide association study. | Amyotrophic lateral sclerosis |
| (Laaksovirta *et al.*, 2010) | 405 European ancestry cases, 497 European ancestry controls | NA | Chromosome 9p21 in amyotrophic lateral sclerosis in Finland: a genome-wide association study. | Amyotrophic lateral sclerosis |
| (Kwee *et al.*, 2012) | Up to 639 European ancestry cases, 6,257 European ancestry controls | Up to 183 European ancestry cases, 961 European ancestry controls | A high-density genome-wide association screen of sporadic ALS in US veterans. | Amyotrophic lateral sclerosis |
| (Van Es *et al.*, 2008) | 737 European ancestry cases, 721 European ancestry controls | 1,030 European ancestry cases, 1,195 European ancestry controls | Genetic variation in DPP6 is associated with susceptibility to amyotrophic lateral sclerosis. | Amyotrophic lateral sclerosis |
| (Wei *et al.*, 2019) | 666 Han Chinese ancestry cases, 3,988 Han Chinese ancestry controls | up to 884 Han Chinese ancestry cases, up to 5,329 Han Chinese ancestry controls | Identification of TYW3/CRYZ and FGD4 as susceptibility genes for amyotrophic lateral sclerosis. | Amyotrophic lateral sclerosis |
| (Dekker *et al.*, 2019) | 4,244 European ancestry cases, 3,106 European ancestry controls | NA | Exome array analysis of rare and low frequency variants in amyotrophic lateral sclerosis. | Amyotrophic lateral sclerosis |
| (Shatunov *et al.*, 2010) | 4,857 European ancestry cases, 8,987 European ancestry controls | NA | Chromosome 9p21 in sporadic amyotrophic lateral sclerosis in the UK and seven other countries: a genome-wide association study. | Amyotrophic lateral sclerosis |
| (Van Es *et al.*, 2009) | 2,323 European ancestry cases, 9,013 European ancestry controls | 2,532 European ancestry cases, 5,940 European ancestry controls | Genome-wide association study identifies 19p13.3 (UNC13A) and 9p21.2 as susceptibility loci for sporadic amyotrophic lateral sclerosis. | Amyotrophic lateral sclerosis |
| (Langefeld, 2013) | 4,243 European ancestry cases | NA | Age of onset of amyotrophic lateral sclerosis is modulated by a locus on 1p34.1. | Amyotrophic lateral sclerosis |
| (Deng *et al.*, 2013) | 506 Han Chinese ancestry cases, 1,859 Han Chinese ancestry controls | 706 Han Chinese ancestry cases, 1,777 Han Chinese ancestry controls | Genome-wide association analyses in Han Chinese identify two new susceptibility loci for amyotrophic lateral sclerosis. | Amyotrophic lateral sclerosis |
| (Goris *et al.*, 2014) | 4,088 Multiple sclerosis cases, 3,762 Amyotrophic lateralsclerosis cases, 12,030 controls | NA | No evidence for shared genetic basis of common variants in multiple sclerosis and amyotrophic lateral sclerosis. | Amyotrophic lateral sclerosis |
| (Diekstra *et al.*, 2014) | 4,377 European ancestry ALS cases, 435 European ancestry FTD cases, 14,431 European ancestry controls | 4,056 European ancestry ALS cases, 3,958 European ancestry controls | C9orf72 and UNC13A are shared risk loci for amyotrophic lateral sclerosis and frontotemporal dementia: a genome-wide meta-analysis. | Amyotrophic lateral sclerosis |
| (Landers *et al.*, 2009) | 1,821 cases, 2,258 controls | 538 cases, 556 controls | Reduced expression of the Kinesin-Associated Protein 3 (KIFAP3) gene increases survival in sporadic amyotrophic lateral sclerosis. | Amyotrophic lateral sclerosis |
| (Van Rheenen *et al.*, 2016) | 12,577 European ancestry cases, 23,475 European ancestry controls | 2,579 European ancestry cases, 2,767 European ancestry controls | Genome-wide association analyses identify new risk variants and the genetic architecture of amyotrophic lateral sclerosis. | Amyotrophic lateral sclerosis |
| (Nicolas *et al.*, 2018) | 20,806 European ancestry cases, 59,804 European ancestry controls | 4,159 cases, 18,650 controls | Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. | Amyotrophic lateral sclerosis |

## A2: Transcriptome study information

Table 6 Overview of studies used for gathering transcriptomic data AD = Alzheimer’s disease, PD = Parkinson’s disease, (s)ALS =(sporadic) amyotrophic lateral sclerosis, HD = Huntington’s disease, ctrl = control, CSF = cerebrospinal fluid, BA = Brodman Area

|  |  |  |
| --- | --- | --- |
| **study** | **Participants** | **Tissue** |
| **E-MTAB-6094\_Leandro\_2017\_AD** | 22 AD (7 severe, 15 mild), 13 ctrl | Peripheral Blood Mononuclear Cells |
| (Berchtold *et al.*, 2013) | 26 AD, 33 old ctrl, 22 young ctrl | Entorhinal Cortex, Hippocampus, Postcentral Gyrus, Superior Frontal Cortex |
| (Hokama *et al.*, 2014) | FC: 15 AD, 18 ctrl;  Temporal Cortex: 10 AD, 19 ctrl;  Hippocampus: 7 AD, 10 ctrl | Frontal Cortex, Temporal Cortex, Hippocampus |
| (Fischer *et al.*, 2013) | 3 AD, 3 ctrl (3 MS, 3 TB) | Cortex |
| (Blalock *et al.*, 2004) | 22 AD, 9 ctrl | Hippocampus |
| (Blalock *et al.*, 2011) | 22 AD (7 incipient, 8 moderate, 7 severe), 8 ctrl | CA1 hippocampal gray matter |
| **GSE 18309\_Chen\_2009\_AD** | 4 AD, 5 mild cognitive impairment, 4 ctrl | Peripheral Blood Mononuclear Cells |
| (Liang *et al.*, 2007) | EC: 10 AD, 13 ctrl; HIP: 10 AD, 13 ctrl; MTG: 16 AD, 12 ctrl; PC: 9 AD, 13 ctrl; SFG: 23 AD, 11 ctrl; VCX: 19 AD, 12 ctrl | EC=Entorhinal Cortex; HIP=Hippocampus; MTG=Medial temporal gyrus; PC=Posterior Singulate; SFG=Superior Frontal Gyrus; VCX=Primary visual cortex |
| (Magistri *et al.*, 2015) | 4 LOAD, 4 ctrl | Hippocampus |
| (Dunckley *et al.*, 2006) | 19 AD, 14 ctrl | Entorhinal cortex |
| (Scheckel *et al.*, 2016) | 9 AD, 8 ctrl | Brain |
| (Meyer *et al.*, 2019) | 5 AD,5 ctrl | iPSC induced neurons |
| (Mathys *et al.*, 2019) | 24 AD, 24 ctrl | 80660 sc transcriptomes from prefrontal cortex, 6 different celltypes: astrocytes, exitatory neurons, inhibitory neurons, microglia, oligodendrocyte progenitor cells, oligodendrocytes |
| (Stopa *et al.*, 2018) | 7 AD, 3 HD, 6 ctrl | choroid plexus |
| (Simunovic *et al.*, 2009) | 10 PD, 9 ctrl | neurons of Substantia nigra |
| (Elstner *et al.*, 2011) | 11 PD, 11 old ctrl, 8 young ctrl | neurons of Substantia nigra |
| (Riley *et al.*, 2014) | 16 PD-20 ctrl (FC), 17 PD-17 ctrl (striatum), 16 PD-14 ctrl (SN) | Cortex, Striatum, Substantia nigra |
| (Dijkstra *et al.*, 2015) | 9 PD, 8 ctrl | Substantia nigra |
| (Dumitriu *et al.*, 2012) | 27 PD, 26 ctrl | prefrontal cortex (BA9) |
| (Dumitriu *et al.*, 2016) | 29 PD, 44 ctrl | prefrontal cortex (BA9) |
| (Scherzer *et al.*, 2007) | 50 PD, 22 ctrl | whole blood |
| (Lesnick *et al.*, 2007) | 16 PD, 9 ctrl | Substantia nigra |
| (Zhang *et al.*, 2005) | 15 PD, 15 ctrl | Prefrontal cortex (BA9) |
| (Zhang *et al.*, 2005) | 15 PD, 15 ctrl | Substantia nigra |
| **GSE20333\_Edna\_2010\_PD** | 6 PD, 6 ctrl | Substantia nigra |
| (Calligaris *et al.*, 2015) | 40 PD, 20 ctrl | Blood |
| (Ring *et al.*, 2015) | 16 HD, 16 ctrl | Transcriptomic analysis of HD iPSCs and HD NSCs compared to isogenic controls using RNA-Seq. |
| (Świtońska *et al.*, 2019) | 2HD, 2 ctrl | HD and control iPSC lines |
| (Al-Dalahmah *et al.*, 2020) | 6 HD Grad III/IV, 6 ctrl | astrocytes of singulate cortex |
| (Moss *et al.*, 2017) | 186 HD, 49 ctrl | Blood |
| (Labadorf *et al.*, 2015) | 20 HD, 49 ctrl, male | prefrontal cortex, Brodman Area 9 |
| (Mastrokolias *et al.*, 2015) | Blood 27 präHD, 64 HD, 33 ctrl | Blood |
| (Lin *et al.*, 2016) | 7 HD, 7 ctrl | motor cortex |
| (Feyeux *et al.*, 2012) | 6 HD, 4 ctrl | embryonic stem cells |
| (Lim, Salazar, *et al.*, 2017) | 6 HD, 4 ctrl | iPSC derived neural cells |
| (Lim, Quan, *et al.*, 2017) | 4 HD, 2 ctrl | iPSC induced brain microvascular endothelial cells |
| (Mehta *et al.*, 2018) | 3 HD, 3 ctrl | iPSC derived cortical neurons |
| (Stopa *et al.*, 2018) | 7 AD, 3 HD, 6 ctrl | choroid plexus |
| (Cox *et al.*, 2010) | 3 ALS, 7 ctrl | cervical spinal cord |
| (Otake, Kamiguchi and Hirozane, 2019) | 4 ALS, 4 ctrl | exosomale mRNA in CSF |
| (Gagliardi *et al.*, 2018) | 11 sALS, je 2 fALS(FUS,TARDBP,SOD1), 3 ctrl | blood\_monocytes |
| (Swindell *et al.*, 2019) | 397 ALS, 645 ctrl | blood |
| (Prudencio *et al.*, 2015) | 19 ALS (10 sALS, 9 c9ALS), 9 ctrl | Cerebellum (BA9/44), Frontal cortex (BA4) |
| (Raman *et al.*, 2015) | 6 sALS, 6 ctrl | Fibroblasts from skin cells |
| (Kapeli *et al.*, 2016) | 13 sALS, 9 control | human stem cell-derived motor neurons |
| (Dols-Icardo *et al.*, 2020) | 11 ALS, 8 ctrl | motor cortex, BA 4 |

## A3: Proteome study information

Table 7 Overview of studies used for gathering proteomic data AD = Alzheimer’s disease, PD = Parkinson’s disease, ALS = amyotrophic lateral sclerosis, HD = Huntington’s disease, ctrl = control, CSF = cerebrospinal fluid, BA = Brodman Area

|  |  |  |
| --- | --- | --- |
| **study** | **Participants** | **Tissue** |
| (Johnson *et al.*, 2020) | 268A D, 104 asymptomatic AD, 99 ctrl | dorsolateral prefrontal cortex |
| (Bader *et al.*, 2020) | ca. 90 AD, 150 ctrl | CSF |
| (Zhang *et al.*, 2018) | 8 AD, 8 ctrl | frontal cortex |
| (Seyfried *et al.*, 2017) | Emory: each 8 AD, PD, ALS, ctrl;  BLSA: 20 AD (BA7), 20 AD (BA9), 15 ctrl (BA7), 13 ctrl (BA9) | Emory: prefrontal cortex;  BLSA: prefrontal cortex (BA9), Precuneus (BA7) |
| (Higginbotham, Dammer, *et al.*, 2019) | 6 AD, 6 ctrl | middle frontal gyrus BA 8, BA 9 |
| (Higginbotham, Ping, *et al.*, 2019) | CSF1: 20 AD, 20 ctrl; Brain1: 10 AD, 10 ctrl; Brain2: 9 AD, 10 ctrl; CSF2: 33 AD, 32 ctrl | dorsolateral prefrontal cortex |
| (Wingo *et al.*, 2020) | 383 AD, 375 ctrl | dorsolateral prefrontal cortex (BA9) |
| (Lachén-Montes *et al.*, 2017) | each 5 AD (3 states), 5 ctrl | olfactory bulb |
| (Hondius *et al.*, 2016) | 35 AD, 5 ctrl | hippocampus: CA1 + subiculum |
| (Dumitriu *et al.*, 2016) | 12 PD, 12 ctrl | prefrontal cortex BA9 |
| (Rotunno *et al.*, 2020) | Cohort 1: 53 PD, 72 ctrl  Cohort 2: 28 PD, 43 ctrl | CSF |
| (Riley *et al.*, 2014) | 3 PD, 3 ctrl (striatum), 5 PD, 5 ctrl (Cortex) | Striatum, cortex |
| (Seyfried *et al.*, 2017) | 8 PD, 8 ctrl | Prefrontal cortex |
| (Lachén-Montes *et al.*, 2019) | 21 PD, 8 ctrl | olfactory bulb |
| (Higginbotham, Ping, *et al.*, 2019) | 10 PD, 10 ctrl | dorsolateral prefrontal cortex |
| (van Dijk *et al.*, 2012) | 6 PD, 6 ctrl | locus ceruleus |
| (Umoh *et al.*, 2018) | 19 ALS, 10 ctrl | frontal cortex |
| (Oeckl *et al.*, 2020) | 26 ALS, 16 ctrl (CSF), 8ALS, 7ctrl (spinal cord tissue) | CSF, spinal cord tissue |
| (Iridoy *et al.*, 2019) | 9 ALS, 8 ctrl | anterior horn of spinal cord |
| (Collins *et al.*, 2015) | 90 sALS, 80 ctrl, pooled to 9 sALS, 8 ctrl | CSF |
| (Varghese *et al.*, 2013) | 10 ALS, 10 ctrl | CSF |
| (Ratovitski *et al.*, 2016) | 12 HD, 12 ctrl | cortex |
| (Fang *et al.*, 2009) | 10 earlyHD, 10 midHD, 10 ctrl | CSF |
| (McQuade *et al.*, 2014) | hESC: 3 HD, 3 ctrl  NSC: 4 HD, 4 ctrl | hESC, NSC |