

# ADSP Publication List

2023

## Diverse Ancestries

### African/African American

#### U01 AG052410- Replication and Extension of ADSP Discoveries in African-Americans

1. Bouzid H, Belk JA, Jan M, et al. Clonal hematopoiesis is associated with protection from Alzheimer's disease. *Nat Med.* 2023 Jul;29(7):1662–1670. doi: 10.1038/s41591-023-02397-2 PMID: 37322115; <https://www.ncbi.nlm.nih.gov/pubmed/37322115> PMCID: PMC10353941
2. Byfield G, Starks TD, Luther R, et al. Leveraging African American family connectors for Alzheimer's disease genomic studies. *Alzheimers Dement.* 2023 May 22; doi: 10.1002/alz.13106 PMID: 37212603; <https://www.ncbi.nlm.nih.gov/pubmed/37212603>
3. Celis K, Moreno MDMM, Rajabli F, et al. Ancestry-related differences in chromatin accessibility and gene expression of APOE  $\epsilon$ 4 are associated with Alzheimer's disease risk. *Alzheimers Dement.* 2023 Apr 10; doi: 10.1002/alz.13075 PMID: 37037656; <https://www.ncbi.nlm.nih.gov/pubmed/37037656>
4. Fan K, Francis L, Aslam MM, et al. Investigation of the independent role of a rare APOE variant (L28P; APOE\*4Pittsburgh) in late-onset Alzheimer disease. *Neurobiol Aging.* 2023 Feb;122:107–111. doi: 10.1016/j.neurobiolaging.2022.11.007 PMID: 36528961; <https://www.ncbi.nlm.nih.gov/pubmed/36528961> PMCID: PMC9839598
5. Horimoto ARVR, Boyken LA, Blue EE, et al. Admixture mapping implicates 13q33.3 as ancestry-of-origin locus for Alzheimer disease in Hispanic and Latino populations. *HGG Adv.* 2023 Jul 13;4(3):100207. doi: 10.1016/j.xhgg.2023.100207 PMID: 37333771; <https://www.ncbi.nlm.nih.gov/pubmed/37333771> PMCID: PMC10276158
6. Rajabli F, Tosto G, Hamilton-Nelson KL, et al. Admixture mapping identifies novel Alzheimer's disease risk regions in African Americans. *Alzheimers Dement.* 2023 Jun;19(6):2538–2548. doi: 10.1002/alz.12865 PMID: 36539198; <https://www.ncbi.nlm.nih.gov/pubmed/36539198> PMCID: PMC10272044
7. Reyes-Dumeyer D, Faber K, Vardarajan B, et al. The National Institute on Aging Late-Onset Alzheimer's Disease Family Based Study: A resource for genetic discovery. *Alzheimers Dement.* 2022 Oct;18(10):1889–1897. doi: 10.1002/alz.12514 PMID: 34978149; <https://www.ncbi.nlm.nih.gov/pubmed/34978149> PMCID: PMC9250549

#### R01AG072547 - The Origins of Alzheimer Disease in African Americans

8. Nuytemans K, Lipkin Vasquez M, Wang L, *et al.* Identifying differential regulatory control of APOE  $\epsilon$ 4 on African versus European haplotypes as potential therapeutic targets. *Alzheimer's Dement.* 2022 Oct;18(10):1930–1942. doi: [10.1002/alz.12534](https://doi.org/10.1002/alz.12534) PMID: 34978147; <https://www.ncbi.nlm.nih.gov/pubmed/34978147> PMCID: PMC9250552
9. Reitz C, Pericak-Vance MA, Foroud T, Mayeux R. A global view of the genetic basis of Alzheimer disease. *Nat Rev Neurol.* 2023 May;19(5):261–277. doi: [10.1038/s41582-023-00789-z](https://doi.org/10.1038/s41582-023-00789-z) PMID: 37024647; <https://www.ncbi.nlm.nih.gov/pubmed/37024647>
10. Rogers A, Chung A, Seixas A, Chung D, Zizi F, Jean-Louis G. Strategies to Engage Blacks in Sleep Medicine: Lessons Learned from Three Studies Applying Community-Based Participatory Research Principles. *J Sleep Disord Ther.* 2023;12(4):425. PMID: 37425370; <https://www.ncbi.nlm.nih.gov/pubmed/37425370> PMCID: PMC10327646

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## Amish

### **R01 AG058066 - Protective Genetic Variants for Alzheimer Disease in the Amish**

11. Prough MB, Zaman A, Caywood LJ, *et al.* Visuospatial and Verbal Memory Differences in Amish Individuals With Alzheimer Disease and Related Dementias. *Alzheimer Dis Assoc Disord.* 2023 Sep 1;37(3):195–199. doi: [10.1097/WAD.0000000000000570](https://doi.org/10.1097/WAD.0000000000000570) PMID: 37561946; <https://www.ncbi.nlm.nih.gov/pubmed/37561946>
12. Ramos J, Caywood LJ, Prough MB, *et al.* Genetic variants in the SHISA6 gene are associated with delayed cognitive impairment in two family datasets. *Alzheimers Dement.* 2023 Feb;19(2):611–620. doi: [10.1002/alz.12686](https://doi.org/10.1002/alz.12686) PMID: 35490390; <https://www.ncbi.nlm.nih.gov/pubmed/35490390> PMCID: PMC9622429
13. Zaman A, Caywood L, Prough M, *et al.* Psychometric approaches to defining cognitive phenotypes in the Old Order Amish. *Int J Geriatr Psychiatry.* 2023 Apr;38(4):e5903. doi: [10.1002/gps.5903](https://doi.org/10.1002/gps.5903) PMID: 36929524; <https://www.ncbi.nlm.nih.gov/pubmed/36929524>

## India

### **U01 AG064948 - Harmonized Diagnostic Assessment of Dementia (DAD) for Longitudinal Aging Study of India (LASI)-Genomic Study**

14. Banerjee J, Petrosyan S, Rao AR, *et al.* Cohort Profile: Real-Time Insights of COVID-19 in India (RTI COVID-India). *BMC Public Health.* 2023 Feb 9;23(1):292. doi: [10.1186/s12889-023-15084-1](https://doi.org/10.1186/s12889-023-15084-1) PMID: 36759802; <https://www.ncbi.nlm.nih.gov/pubmed/36759802> PMCID: PMC9909130
15. Jin H, Crimmins E, Langa KM, Dey AB, Lee J. Estimating the Prevalence of Dementia in India Using a Semi-Supervised Machine Learning Approach. *Neuroepidemiology.* 2023;57(1):43–50. doi: [10.1159/000528904](https://doi.org/10.1159/000528904) PMID: 36617419; <https://www.ncbi.nlm.nih.gov/pubmed/36617419> PMCID: PMC10038923
16. Lee J, Meijer E, Langa KM, *et al.* Prevalence of dementia in India: National and state estimates from a nationwide study. *Alzheimers Dement.* 2023 Jul;19(7):2898–2912. doi: [10.1002/alz.12928](https://doi.org/10.1002/alz.12928) PMID: 36637034; <https://www.ncbi.nlm.nih.gov/pubmed/36637034> PMCID: PMC10338640
17. Lee J, Petrosyan S, Khobragade P, *et al.* Deep phenotyping and genomic data from a nationally representative study on dementia in India. *Sci Data.* 2023 Jan 20;10(1):45. doi: [10.1038/s41597-023-01941-6](https://doi.org/10.1038/s41597-023-01941-6) PMID: 36670106; <https://www.ncbi.nlm.nih.gov/pubmed/36670106> PMCID: PMC9852797
18. Nichols E, Ng DK, Hayat S, *et al.* Measurement differences in the assessment of functional limitations for cognitive impairment classification across geographic locations. *Alzheimers Dement.* 2023 May;19(5):2218–2225. doi: [10.1002/alz.12994](https://doi.org/10.1002/alz.12994) PMID: 36807779; <https://www.ncbi.nlm.nih.gov/pubmed/36807779> PMCID: PMC10182237

## Korea

### **U01 AG072177 - KBASE2: Korean Brain Aging Study, Longitudinal Endophenotypes, and Systems Biology**

19. Byun MS, Chang M, Yi D, *et al.* Association of Central Auditory Processing Dysfunction With Preclinical Alzheimer's Disease. *Otolaryngol Head Neck Surg.* 2023 Jul;169(1):112–119. doi: [10.1002/hlth.12994](https://doi.org/10.1002/hlth.12994)

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- [10.1002/ohn.228](https://pubmed.ncbi.nlm.nih.gov/36939433/) PMID: 36939433; <https://www.ncbi.nlm.nih.gov/pubmed/36939433>
20. Coleman ME, Roessler MEH, Peng S, *et al.* Social enrichment on the job: Complex work with people improves episodic memory, promotes brain reserve, and reduces the risk of dementia. *Alzheimers Dement.* 2023 Jun;19(6):2655–2665. doi: [10.1002/alz.13035](https://doi.org/10.1002/alz.13035) PMID: 37037592; <https://www.ncbi.nlm.nih.gov/pubmed/37037592> PMCID: PMC10272079
  21. Grothe MJ, Moscoso A, Silva-Rodríguez J, *et al.* Differential diagnosis of amnesic dementia patients based on an FDG-PET signature of autopsy-confirmed LATE-NC. *Alzheimers Dement.* 2023 Apr;19(4):1234–1244. doi: [10.1002/alz.12763](https://doi.org/10.1002/alz.12763) PMID: 35971593; <https://www.ncbi.nlm.nih.gov/pubmed/35971593> PMCID: PMC9929029
  22. Hirschfeld LR, Risacher SL, Nho K, Saykin AJ. Myelin repair in Alzheimer’s disease: a review of biological pathways and potential therapeutics. *Transl Neurodegener.* 2022 Oct 26;11(1):47. doi: [10.1186/s40035-022-00321-1](https://doi.org/10.1186/s40035-022-00321-1) PMID: 36284351; <https://www.ncbi.nlm.nih.gov/pubmed/36284351> PMCID: PMC9598036
  23. Hu F, Chen AA, Horng H, *et al.* Image harmonization: A review of statistical and deep learning methods for removing batch effects and evaluation metrics for effective harmonization. *Neuroimage.* 2023 Jul 1;274:120125. doi: [10.1016/j.neuroimage.2023.120125](https://doi.org/10.1016/j.neuroimage.2023.120125) PMID: 37084926; <https://www.ncbi.nlm.nih.gov/pubmed/37084926> PMCID: PMC10257347
  24. Jeon SY, Byun MS, Yi D, *et al.* Circadian rest-activity rhythm and longitudinal brain changes underlying late-life cognitive decline. *Psychiatry Clin Neurosci.* 2023 Apr;77(4):205–212. doi: [10.1111/pcn.13521](https://doi.org/10.1111/pcn.13521) PMID: 36527292; <https://www.ncbi.nlm.nih.gov/pubmed/36527292> PMCID: PMC10360409
  25. Jung JH, Kim G, Byun MS, *et al.* Gut microbiome alterations in preclinical Alzheimer’s disease. *PLoS One.* 2022;17(11):e0278276. doi: [10.1371/journal.pone.0278276](https://doi.org/10.1371/journal.pone.0278276) PMID: 36445883; <https://www.ncbi.nlm.nih.gov/pubmed/36445883> PMCID: PMC9707757
  26. Kang KM, Byun MS, Yi D, *et al.* Enlarged perivascular spaces are associated with decreased brain tau deposition. *CNS Neurosci Ther.* 2023 Feb;29(2):577–586. doi: [10.1111/cns.14040](https://doi.org/10.1111/cns.14040) PMID: 36468423; <https://www.ncbi.nlm.nih.gov/pubmed/36468423> PMCID: PMC9873511
  27. Kim JW, Byun MS, Lee JH, *et al.* Spouse bereavement and brain pathologies: A propensity score matching study. *Psychiatry Clin Neurosci.* 2022 Oct;76(10):490–504. doi: [10.1111/pcn.13439](https://doi.org/10.1111/pcn.13439) PMID: 35751876; <https://www.ncbi.nlm.nih.gov/pubmed/35751876> PMCID: PMC9796777
  28. Kim JW, Byun MS, Yi D, *et al.* Serum Adiponectin and In Vivo Brain Amyloid Deposition in Cognitively Normal Older Adults: A Cohort Study. *Aging Dis.* 2023 Jun 1;14(3):904–918. doi: [10.14336/AD.2022.1118](https://doi.org/10.14336/AD.2022.1118) PMID: 37191420; <https://www.ncbi.nlm.nih.gov/pubmed/37191420> PMCID: PMC10187695
  29. Kim JP, Kim BH, Bice PJ, *et al.* Integrative Co-methylation Network Analysis Identifies Novel DNA Methylation Signatures and Their Target Genes in Alzheimer’s Disease. *Biol Psychiatry.* 2023 May 1;93(9):842–851. doi: [10.1016/j.biopsych.2022.06.020](https://doi.org/10.1016/j.biopsych.2022.06.020) PMID: 36150909; <https://www.ncbi.nlm.nih.gov/pubmed/36150909> PMCID: PMC9789210
  30. Kim T, Yi D, Byun MS, *et al.* Synergistic interaction of high blood pressure and cerebral beta-amyloid on tau pathology. *Alzheimers Res Ther.* 2022 Dec 24;14(1):193. doi: [10.1186/s13195-022-01149-7](https://doi.org/10.1186/s13195-022-01149-7) PMID: 36566225; <https://www.ncbi.nlm.nih.gov/pubmed/36566225> PMCID: PMC9789538
  31. Lee S, Byun MS, Yi D, *et al.* Body mass index and two-year change of in vivo Alzheimer’s disease pathologies in cognitively normal older adults. *Alzheimers Res Ther.* 2023 Jun 13;15(1):108. doi: [10.1186/s13195-023-01259-w](https://doi.org/10.1186/s13195-023-01259-w) PMID: 37312229; <https://www.ncbi.nlm.nih.gov/pubmed/37312229> PMCID: PMC10262499

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32. Moon SW, Byun MS, Yi D, *et al.* Low Ankle-Brachial Index Relates to Alzheimer-Signature Cerebral Glucose Metabolism in Cognitively Impaired Older Adults. *J Alzheimers Dis.* 2023;93(1):87–95. doi: [10.3233/JAD-220911](https://doi.org/10.3233/JAD-220911) PMID: 36938732; <https://www.ncbi.nlm.nih.gov/pubmed/36938732> PMCID: PMC10200182
33. Moutinho M, Coronel I, Tsai AP, *et al.* TREM2 splice isoforms generate soluble TREM2 species that disrupt long-term potentiation. *Genome Med.* 2023 Feb 20;15(1):11. doi: [10.1186/s13073-023-01160-z](https://doi.org/10.1186/s13073-023-01160-z) PMID: 36805764; <https://www.ncbi.nlm.nih.gov/pubmed/36805764> PMCID: PMC9940368
34. Oatman SR, Reddy JS, Quicksall Z, *et al.* Genome-wide association study of brain biochemical phenotypes reveals distinct genetic architecture of Alzheimer’s disease related proteins. *Mol Neurodegener.* 2023 Jan 7;18(1):2. doi: [10.1186/s13024-022-00592-2](https://doi.org/10.1186/s13024-022-00592-2) PMID: 36609403; <https://www.ncbi.nlm.nih.gov/pubmed/36609403> PMCID: PMC9825010
35. Park JC, Lim H, Byun MS, *et al.* Sex differences in the progression of glucose metabolism dysfunction in Alzheimer’s disease. *Exp Mol Med.* 2023 May;55(5):1023–1032. doi: [10.1038/s12276-023-00993-3](https://doi.org/10.1038/s12276-023-00993-3) PMID: 37121979; <https://www.ncbi.nlm.nih.gov/pubmed/37121979> PMCID: PMC10238450
36. Pyun JM, Park YH, Wang J, *et al.* Aberrant GAP43 Gene Expression Is Alzheimer Disease Pathology-Specific. *Ann Neurol.* 2023 May;93(5):1047–1048. doi: [10.1002/ana.26637](https://doi.org/10.1002/ana.26637) PMID: 36897291; <https://www.ncbi.nlm.nih.gov/pubmed/36897291>
37. Pyun JM, Park YH, Wang J, *et al.* Transcriptional risk scores in Alzheimer’s disease: From pathology to cognition. *Alzheimers Dement.* 2023 Aug 10; doi: [10.1002/alz.13406](https://doi.org/10.1002/alz.13406) PMID: 37563770; <https://www.ncbi.nlm.nih.gov/pubmed/37563770>
38. Swinford CG, Risacher SL, Vosmeier A, *et al.* Amyloid and tau pathology are associated with cerebral blood flow in a mixed sample of nondemented older adults with and without vascular risk factors for Alzheimer’s disease. *Neurobiol Aging.* 2023 Oct;130:103–113. doi: [10.1016/j.neurobiolaging.2023.06.014](https://doi.org/10.1016/j.neurobiolaging.2023.06.014) PMID: 37499587; <https://www.ncbi.nlm.nih.gov/pubmed/37499587>
39. Swinford CG, Risacher SL, Wu YC, *et al.* Altered cerebral blood flow in older adults with Alzheimer’s disease: a systematic review. *Brain Imaging Behav.* 2023 Apr;17(2):223–256. doi: [10.1007/s11682-022-00750-6](https://doi.org/10.1007/s11682-022-00750-6) PMID: 36484922; <https://www.ncbi.nlm.nih.gov/pubmed/36484922> PMCID: PMC10117447
40. Tometich DB, Mosher CE, Cyders M, *et al.* An Examination of the Longitudinal Relationship Between Cognitive Function and Physical Activity Among Older Breast Cancer Survivors in the Thinking and Living With Cancer Study. *Ann Behav Med.* 2023 Apr 5;57(3):237–248. doi: [10.1093/abm/kaac048](https://doi.org/10.1093/abm/kaac048) PMID: 36356044; <https://www.ncbi.nlm.nih.gov/pubmed/36356044> PMCID: PMC10074030

### **U01AG062602 - Genetic Studies of Alzheimer Disease in Koreans**

41. Chung J, Sahelijo N, Maruyama T, *et al.* Alzheimer’s disease heterogeneity explained by polygenic risk scores derived from brain transcriptomic profiles. *Alzheimers Dement.* 2023 May 11; doi: [10.1002/alz.13069](https://doi.org/10.1002/alz.13069) PMID: 37166019; <https://www.ncbi.nlm.nih.gov/pubmed/37166019>
42. Jun GR, You Y, Zhu C, *et al.* Protein phosphatase 2A and complement component 4 are linked to the protective effect of APOE ε2 for Alzheimer’s disease. *Alzheimers Dement.* 2022 Nov;18(11):2042–2054. doi: [10.1002/alz.12607](https://doi.org/10.1002/alz.12607) PMID: 35142023; <https://www.ncbi.nlm.nih.gov/pubmed/35142023> PMCID: PMC9360190
43. Kang M, Ang TFA, Devine SA, *et al.* A genome-wide search for pleiotropy in more than 100,000

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- harmonized longitudinal cognitive domain scores. *Mol Neurodegener.* 2023 Jun 22;18(1):40. doi: [10.1186/s13024-023-00633-4](https://doi.org/10.1186/s13024-023-00633-4) PMID: 37349795; <https://www.ncbi.nlm.nih.gov/pubmed/37349795> PMCID: PMC10286470
44. Le Guen Y, Raulin AC, Logue MW, *et al.* Association of African Ancestry-Specific APOE Missense Variant R145C With Risk of Alzheimer Disease. *JAMA.* 2023 Feb 21;329(7):551–560. doi: [10.1001/jama.2023.0268](https://doi.org/10.1001/jama.2023.0268) PMID: 36809323; <https://www.ncbi.nlm.nih.gov/pubmed/36809323> PMCID: PMC9945061
45. Lee Y, Park JY, Lee JJ, *et al.* Heritability of cognitive abilities and regional brain structures in middle-aged to elderly East Asians. *Cereb Cortex.* 2023 May 9;33(10):6051–6062. doi: [10.1093/cercor/bhac483](https://doi.org/10.1093/cercor/bhac483) PMID: 36642501; <https://www.ncbi.nlm.nih.gov/pubmed/36642501> PMCID: PMC10183741
46. Li D, Farrell JJ, Mez J, *et al.* Novel loci for Alzheimer’s disease identified by a genome-wide association study in Ashkenazi Jews. *Alzheimers Dement.* 2023 Jun 1; doi: [10.1002/alz.13117](https://doi.org/10.1002/alz.13117) PMID: 37260021; <https://www.ncbi.nlm.nih.gov/pubmed/37260021>
47. Marini S, Chung J, Han X, *et al.* Pleiotropy analysis between lobar intracerebral hemorrhage and CSF  $\beta$ -amyloid highlights new and established associations. *Int J Stroke.* 2023 Aug;18(7):804–811. doi: [10.1177/17474930231155816](https://doi.org/10.1177/17474930231155816) PMID: 36705426; <https://www.ncbi.nlm.nih.gov/pubmed/36705426>

### **R56AG069130 - Asian Cohort for Alzheimers Disease (ACAD) (FUS2.0)**

48. Gao Y, Jia Z, Zhao L, Han S. The Effect of Activity Participation in Middle-Aged and Older People on the Trajectory of Depression in Later Life: National Cohort Study. *JMIR Public Health Surveill.* 2023 Mar 23;9:e44682. doi: [10.2196/44682](https://doi.org/10.2196/44682) PMID: 36951932; <https://www.ncbi.nlm.nih.gov/pubmed/36951932> PMCID: PMC10131905
49. Lee H, Ha H, Yim S, *et al.* Using community-based geographical information system (GIS) to recruit older Asian Americans in an Alzheimer’s disease study. *BMJ Open.* 2023 Aug 3;13(8):e072761. doi: [10.1136/bmjopen-2023-072761](https://doi.org/10.1136/bmjopen-2023-072761) PMID: 37536975; <https://www.ncbi.nlm.nih.gov/pubmed/37536975> PMCID: PMC10401260
50. Martinez AE, Weissberger G, Kuklenyik Z, *et al.* The small HDL particle hypothesis of Alzheimer’s disease. *Alzheimers Dement.* 2023 Feb;19(2):391–404. doi: [10.1002/alz.12649](https://doi.org/10.1002/alz.12649) PMID: 35416404; <https://www.ncbi.nlm.nih.gov/pubmed/35416404>
51. Miyawaki CE, Garcia JM, Nguyen KN, Park VT, Markides KS. Multiple Chronic Conditions and Disability among Vietnamese Older Adults: Results from the Vietnamese Aging and Care Survey (VACS). *J Racial Ethn Health Disparities.* 2023 May 30; doi: [10.1007/s40615-023-01652-z](https://doi.org/10.1007/s40615-023-01652-z) PMID: 37249829; <https://www.ncbi.nlm.nih.gov/pubmed/37249829>
52. Ta Park VM, Ly Q, von Oppenfeld J, *et al.* A scoping review of dementia caregiving for Korean Americans and recommendations for future research. *Clin Gerontol.* 2023;46(2):223–239. doi: [10.1080/07317115.2022.2133907](https://doi.org/10.1080/07317115.2022.2133907) PMID: 36268979; <https://www.ncbi.nlm.nih.gov/pubmed/36268979> PMCID: PMC9928901

### **CADRE**

#### **U01AG058654 - The Alzheimer Disease Sequence Analysis Collaborative**

53. Bai H, Naj AC, Benchek P, *et al.* A haptoglobin (HP) structural variant alters the effect of APOE alleles on Alzheimer’s disease. *Alzheimer’s Dement.* 2023 Apr 12; doi: [10.1002/alz.13050](https://doi.org/10.1002/alz.13050) PMID: 37051669; <https://www.ncbi.nlm.nih.gov/pubmed/37051669>

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54. Celis K, Zaman A, Adams LD, *et al.* Neuropsychiatric features in a multi-ethnic population with Alzheimer disease and mild cognitive impairment. *Int J Geriatr Psychiatry*. 2023 Sep;38(9):e5992. doi: [10.1002/gps.5992](https://doi.org/10.1002/gps.5992) PMID: 37655494; <https://www.ncbi.nlm.nih.gov/pubmed/37655494>
55. Holstege H, Hulsman M, Charbonnier C, *et al.* Exome sequencing identifies rare damaging variants in ATP8B4 and ABCA1 as risk factors for Alzheimer's disease. *Nat Genet*. 2022 Dec;54(12):1786–1794. doi: [10.1038/s41588-022-01208-7](https://doi.org/10.1038/s41588-022-01208-7) PMID: 36411364; <https://www.ncbi.nlm.nih.gov/pubmed/36411364> PMCID: PMC9729101
56. Tejada M, Farrell J, Zhu C, *et al.* DNA from multiple viral species is associated with Alzheimer's disease risk. *Alzheimer's Dement*. 2023 Aug 14; doi: [10.1002/alz.13414](https://doi.org/10.1002/alz.13414) PMID: 37578203; <https://www.ncbi.nlm.nih.gov/pubmed/37578203>
57. Vardarajan BN, Reyes-Dumeyer D, Piriz AL, *et al.* Progranulin mutations in clinical and neuropathological Alzheimer's disease. *Alzheimer's Dement*. 2022 Dec;18(12):2458–2467. doi: [10.1002/alz.12567](https://doi.org/10.1002/alz.12567) PMID: 35258170; <https://www.ncbi.nlm.nih.gov/pubmed/35258170> PMCID: PMC9360185
58. Walters S, Contreras AG, Eissman JM, *et al.* Associations of Sex, Race, and Apolipoprotein E Alleles With Multiple Domains of Cognition Among Older Adults. *JAMA Neurol*. 2023 Jul 17;e232169. doi: [10.1001/jamaneurol.2023.2169](https://doi.org/10.1001/jamaneurol.2023.2169) PMID: 37459083; <https://www.ncbi.nlm.nih.gov/pubmed/37459083> PMCID: PMC10352930

### **Alzheimer's Disease Genetics Consortium (ADGC)**

#### **U01AG032984**

59. Barendrecht S, Schreurs A, Geissler S, *et al.* A novel human tau knock-in mouse model reveals interaction of Abeta and human tau under progressing cerebral amyloidosis in 5xFAD mice. *Alzheimers Res Ther*. 2023 Jan 14;15(1):16. doi: [10.1186/s13195-022-01144-y](https://doi.org/10.1186/s13195-022-01144-y) PMID: 36641439; <https://www.ncbi.nlm.nih.gov/pubmed/36641439> PMCID: PMC9840277
60. Chen Y, Dai J, Tang L, *et al.* Neuroimmune transcriptome changes in patient brains of psychiatric and neurological disorders. *Mol Psychiatry*. 2023 Feb;28(2):710–721. doi: [10.1038/s41380-022-01854-7](https://doi.org/10.1038/s41380-022-01854-7) PMID: 36424395; <https://www.ncbi.nlm.nih.gov/pubmed/36424395> PMCID: PMC9911365
61. Cui Y, Arnold FJ, Peng F, *et al.* Alternative polyadenylation transcriptome-wide association study identifies APA-linked susceptibility genes in brain disorders. *Nat Commun*. 2023 Feb 3;14(1):583. doi: [10.1038/s41467-023-36311-8](https://doi.org/10.1038/s41467-023-36311-8) PMID: 36737438; <https://www.ncbi.nlm.nih.gov/pubmed/36737438> PMCID: PMC9898543
62. Gao XR, Chiariglione M, Qin K, *et al.* Explainable machine learning aggregates polygenic risk scores and electronic health records for Alzheimer's disease prediction. *Sci Rep*. 2023 Jan 9;13(1):450. doi: [10.1038/s41598-023-27551-1](https://doi.org/10.1038/s41598-023-27551-1) PMID: 36624143; <https://www.ncbi.nlm.nih.gov/pubmed/36624143> PMCID: PMC9829871
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## ADSP U01 Awards

### FUS | Therapeutic Targets

#### U01AG058635 - Genomic approach to identification of microglial networks involved in Alzheimer disease risk

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## **ADSP: Functional Genomics Consortium**

### **U01AG072577 - Circular RNAs and Their Interactions With RNA-Binding Proteins to Modulate AD-Related Neuropathology**

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### **U01AG072439 - Functional Genomic Dissection of Alzheimer's Disease in Humans and Drosophila Models**

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### **U01AG072464 - Investigating the Functional Impact of AD Risk Genes on Neuro-Vascular Interactions**

111. Abi-Ghanem C, Salinero AE, Kordit D, *et al.* Sex differences in the effects of high fat diet on underlying neuropathology in a mouse model of VCID. *Biol Sex Differ.* 2023 May 19;14(1):31. doi: [10.1186/s13293-023-00513-y](https://doi.org/10.1186/s13293-023-00513-y) PMID: 37208759; <https://www.ncbi.nlm.nih.gov/pubmed/37208759> PMCID: PMC10199629
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114. Gannon OJ, Naik JS, Riccio D, *et al.* Menopause causes metabolic and cognitive impairments in a chronic cerebral hypoperfusion model of vascular contributions to cognitive impairment and dementia. *Biol Sex Differ.* 2023 May 23;14(1):34. doi: [10.1186/s13293-023-00518-7](https://doi.org/10.1186/s13293-023-00518-7) PMID: 37221553; <https://www.ncbi.nlm.nih.gov/pubmed/37221553> PMCID: PMC10204285
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### **U01AG072572 - Alzheimer Variants: Propagation of Shared Functional Changes Across Cellular Networks**

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## Discovery

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#### U19AG078109 - The Health & Aging Brain Study - Health Disparities (HABS-HD)

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### Essential Infrastructure

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### NOTES:

- Total ADSP publications related to specific NIA Notices of Funding Opportunities (NOFOs) between October 2022 and September 2023 is 240. Total publications related to specific NIA Notices of Funding Opportunities (NOFOs) for all years 2017-2023 is 705
- A supporting effort to examine genetic mechanisms of AD pathophysiology is <https://grants.nih.gov/grants/guide/notice-files/NOT-AG-18-046.html>, Notice to Specify High-Priority Research Topic for PAR-19-070 and PAR-19-071 “Genetic Underpinnings of Endosomal Trafficking as a Pathological Hub in Alzheimer's Disease and Alzheimer's Disease-Related Dementias (AD/ADRD)”
- The number of publications for this between October 2022 and September 2023 is 109. Total publications for this effort are 710
- The grand total of all ADSP publications and supporting endolysosomal studies is 1655

## 2022

### ADSP Banner Publications

1. Lee W-P et al. [Copy Number Variation Identification on 3,800 Alzheimer's Disease Whole Genome Sequencing Data from the Alzheimer's Disease Sequencing Project.](#) *Front Genet.* 2021

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- Nov 4;12:752390. doi: 10.3389/fgene.2021.752390. PMID: 34804120; PMCID: PMC8599981.
2. Neupane A, Lenny B, Budde JP, Wang F, Norton J, Morris JC et al. [Replication study of AD-associated rare variants](#). *Alzheimers Dement*. 2022. 18(4):858-862. doi: 10.1002/alz.12583. PMID: 35103389.
  3. Xue D, Bush WS, Renton AE et al. [Large-scale sequencing studies expand the known genetic architecture of Alzheimer's disease](#). *Alzheimers Dement*. 2021. 13(1): e12255. PMID: 35005195; PMCID: PMC8720139; doi: 10.1002/dad2.12255.

### **Diverse Ancestries**

#### **African/African American**

##### **U01 AG052410, Replication and Extension of ADSP Discoveries in African-Americans**

4. Jin B et al. [An association test of the spatial distribution of rare missense variants within protein structures identify Alzheimer's disease-related patterns](#). *Genome Research*. 2022 Apr;32(4):778-790. doi: 10.1101/gr.276069.121. PMID: 35210353 PMCID: PMC8997344.

#### ***In Press***

5. Rajabli F, Tosto G, Hamilton-Nelson KL et al. for the Alzheimer's Disease Genetics Consortium (ADGC), Collaboration on Alzheimer's Disease Research (CADRE), and Alzheimer's Disease Sequencing Project (ADSP). Admixture mapping identifies novel Alzheimer disease risk regions in African Americans. *Alz Dis and Dementia*, 2022. In Press.

##### **R01AG072547 - The Origins of Alzheimer Disease in African Americans**

6. Gardner OK et al. [Genetic architecture of RNA editing regulation in Alzheimer's disease across diverse ancestral populations](#). *Hum Mol Genet*. 2022 Aug 25;31(17):2876-2886. doi: 10.1093/hmg/ddac075. PMID: 35383839 PMCID: PMC9433728.

#### **Amish**

##### **R01 AG058066 - Protective Genetic Variants for Alzheimer Disease in the Amish**

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#### **India**

##### **U01 AG064948-03 - Harmonized Diagnostic Assessment of Dementia (DAD) for Longitudinal Aging Study of India (LASI)-Genomic Study**

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#### **Korea**

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### U01 AG072177 - KBASE2: Korean Brain Aging Study, Longitudinal Endophenotypes, and Systems Biology

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16. Sohn BK, Byun MS, Yi D, Jeon SY, Lee JH, Choe YM, Lee DW, Lee JY, Kim YK, Sohn CH, Lee DY. [Late-Life Physical Activities Moderate the Relationship of Amyloid- \$\beta\$  Pathology with Neurodegeneration in Individuals Without Dementia.](#) J Alzheimers Dis. 2022;86(1):441-450. doi: 10.3233/JAD-215258. PubMed PMID: 35068452; PubMed Central PMCID: PMC9210327.
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### U01 AG062602 - Genetic Studies of Alzheimer Disease in Koreans

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### ADSP Collaborations with Other Consortia/International Groups

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24. Farrell K, Kim S, Han N et al. [Genome-wide association study and functional validation implicates JADE1 in tauopathy](#). Acta Neuropathol. 2022 Jan;143(1):33-53. doi: 10.1007/s00401-021-02379-z. Epub 2021 Nov 1. PMID: 34719765 .
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26. Heath L et al. [Manifestations of Alzheimer's disease genetic risk in the blood are evident in a multiomic analysis in healthy adults aged 18 to 90](#). Sci Rep. 2022 Apr 12;12(1):6117. doi: 10.1038/s41598-022-09825-2. PMID: 35413975 PMCID: PMC9005657.
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28. Napolioni V, Fredericks CA, Kim Y et al. [Phenotypic heterogeneity among GBA p.R202X carriers in Lewy body spectrum disorders](#). Biomedicines, 10(1):160 (2022). PMID: 35052839
29. Qiao Fan, Shuming Sun, Yi-Ju Li. [Precisely modeling zero-inflated count phenotype for rare variants](#). Genet Epidemiol. 2022 Feb;46(1):73-86. doi: 10.1002/gepi.22438. Epub 2021 Nov 15. PMID: 34779034.
30. Sexton C, Snyder H, Behr D et al. [Current directions in tau research: Highlights from Tau 2020](#). Alzheimers Dement. 2022 May;18(5):988-1007. doi: 10.1002/alz.12452. Epub 2021 Sep 28. PMID: 34581500 Review.
31. Shade, LM et al. [Genome-wide association study of brain arteriosclerosis](#). J Cereb Blood Flow Metab. 2022 Aug;42(8):1437-1450. doi: 10.1177/0271678X211066299. PMID: 35156446 PMCID: PMC9274864.

### *In Press*

32. Holstege H, Hulsman M, Charbonnier C et al. Exome sequencing identifies rare damaging variants in ATP8B4 and ABCA1 as novel risk factors for Alzheimer's Disease. Nat Genet, In press.

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### U01 AG058654 - CADRE: The Alzheimer Disease Sequence Analysis Collaborative

#### *General*

33. Jun GR, Zhu C, Chung J et al. [Protein phosphatase 2A, complement component 4, and APOE genotype linked to Alzheimer disease using a systems biology approach](#). *Alzheimer Dement* 2022. Online ahead of print. PMID: 35142023.
34. Panitch R, Hu J, Stein TD et al. [Blood and brain transcriptome analysis reveals APOE genotype-mediated and immune-related pathways involved in Alzheimer disease](#). *Alz Res Ther* 2022; 14(1):30. PMID: 35139885.

#### *In Press*

35. Rajabli F, Tosto G, Hamilton-Nelson KL et al. for the Alzheimer's Disease Genetics Consortium (ADGC), Collaboration on Alzheimer's Disease Research (CADRE), and Alzheimer's Disease Sequencing Project (ADSP). Admixture mapping identifies novel Alzheimer disease risk regions in African Americans. *Alz Dis and Dementia*, 2022. In Press.

#### *Publications with Other AD Investigators*

36. Arce Renteria M et al. [Midlife Vascular Factors and Prevalence of Mild Cognitive Impairment in Late-Life in Mexico](#). *J Int Neuropsychol Soc*. 2022 Apr;28(4):351-361. doi: 10.1017/S1355617721000539. PMID: 34376262 PMCID: PMC8831650.
37. Brickman AM et al. [Correlation of plasma and neuroimaging biomarkers in Alzheimer's disease](#). *Ann Clin Transl Neurol*. 2022 May;9(5):756-761. doi: 10.1002/acn3.51529. PMID: 35306760 PMCID: PMC9082382.
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42. Morrison MS et al. [Antemortem plasma phosphorylated tau \(181\) predicts Alzheimer's disease neuropathology and regional tau at autopsy](#). *Brain*. 2022 May 13;awac175. doi: 10.1093/brain/awac175. PMID: 35554506.
43. Nishikawa M et al. [Association of Dietary Prebiotic Consumption with Reduced Risk of Alzheimer's Disease in a Multiethnic Population](#). *Curr Alzheimer Res*. 2021;18(12):984-992. doi: 10.2174/1567205019666211222115142. PMID: 34951365 PMCID: PMC8781223.
44. Oluwatosin AO et al. [Molecular Quantitative Trait Locus Mapping in Human Complex Diseases](#). *Curr Protoc*. 2022 May;2(5):e426. doi: 10.1002/cpz1.426. PMID: 35587224 PMCID: PMC9186089.

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45. Panitch R, Hu J, Chung J et al. [Integrative brain omics analysis links the complement pathway to the APOE ε2 protective effect in Alzheimer disease](#). Mol Psychiatry 2021; 26:6054-6064. PMID: 34480088.
46. Rosen AC et al. [The Advisory Group on Risk Evidence Education for Dementia: Multidisciplinary and Open to All](#). J Alzheimers Dis. 2022 Jul 31. doi: 10.3233/JAD-220458. PMID: 35938255.
47. Seixas AA et al. [Associations of digital neuro-signatures with molecular and neuroimaging measures of brain resilience: The altoida large cohort study](#). Front Psychiatry. 022 Aug 9;13:899080. doi: 10.3389/fpsy.2022.899080. PMID: 36061297 PMCID: PMC9435312.
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49. Silva TC et al. [Cross-tissue analysis of blood and brain epigenome-wide association studies in Alzheimer's disease](#). Nat Commun. 2022 Aug 18;13(1):4852. doi: 10.1038/s41467-022-32475-x. PMID: 35982059 PMCID: PMC9388493.
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52. Tao Q et al. [Different loneliness types, cognitive function, and brain structure in midlife: Findings from the Framingham Heart Study](#). E Clinical Medicine. 2022 Sep 6;53:101643. doi: 10.1016/j.eclinm.2022.101643. PMID: 36105871 PMCID: PMC9465265.
53. Yang Y et al. [Alzheimer's disease associated AKAP9 I2558M mutation alters posttranslational modification and interactome of tau and cellular functions in CRISPR-edited human neuronal cells](#). Aging Cell. 2022 Jun;21(6):e13617. doi: 10.1111/accel.13617. PMID: 35567427 PMCID: PMC9197405.
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55. Zhang X, Farrell JJ, Tong T, et al.; Alzheimer's Disease Sequencing Project,. [Association of mitochondrial variants and haplogroups identified by whole exome sequencing with Alzheimer's disease](#). Alzheimers Dement. 2022 Feb;18(2):294-306. doi: 10.1002/alz.12396. PMID: 34152079.
56. Zhang X et al. [Midlife lipid and glucose levels are associated with Alzheimer's disease](#). Alzheimers Dement. 2022 Mar 23. doi: 10.1002/alz.12641. PMID: 35319157.

## **CHARGE**

### ***Publications with Other AD Investigators***

57. Bressler J, Davies G, Smith AV, Saba Y, Bis JC, Jian X et al. [Association of low-frequency and rare coding variants with information processing speed](#). Transl Psychiatry. 2021. 11(1):613. doi: 10.1038/s41398-021-01736-6. PMID: 34864818.
58. Damotte V, van Der Lee SJ, Chouraki V, Grenier-Boley B, Simino J, Adams H et al. [Plasma amyloid β levels are driven by genetic variants near APOE, BACE1, APP, PSEN2: A genome-](#)

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- [wide association study in over 12,000 non-demented participants](#). *Alzheimers Dement*. 2021. 17(10):1663-1674. doi: 10.1002/alz.12333. PMID: 34002480
59. De Rojas I, Moreno-Grau S, Tesi N, Grenier-Boley B, Andrade V, Jansen IE et al. [Common variants in Alzheimer's disease and risk stratification by polygenic risk scores](#). *Nat Commun*. 2021. 12(1):3417. doi: 10.1038/s41467-021-22491-8. PMID: 34099642.
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61. Lahti J, Tuominen S, Yang Q, Pergola G, Ahmad S, Amin N et al. [Genome-wide meta-analyses reveal novel loci for verbal short-term memory and learning](#). *Mol Psychiatry*. 2022. doi: 10.1038/s41380-022-01710-8. PMID: 35974141.
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### **Alzheimer's Disease Genetics Consortium (ADGC)**

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69. DeMichele-Sweet MAA, Klei L, Creese B, et al; NIA-LOAD Family Based Study Consortium, Alzheimer's Disease Genetics Consortium (ADGC) et al. [Genome-wide association identifies the first risk loci for psychosis in Alzheimer disease](#). *Mol Psychiatry*. 2021 Oct;26(10):5797-5811. doi: 10.1038/s41380-021-01152-8. Epub 2021 Jun 10. PMID: 34112972.
70. Gao Y, Felsky D, Reyes-Dumeyer D, et al; CHAP, UKBB, ADNI, ROSMAP, LLFS, NIA-LOAD and ADGC consortia. [Integration of GWAS and brain transcriptomic analyses in a multiethnic](#)

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[sample of 35,245 older adults identifies DCDC2 gene as predictor of episodic memory maintenance](#). *Alzheimers Dement*. 2021 Dec 7. doi: 10.1002/alz.12524. Epub ahead of print. PMID: 34873813. PMCID: 34873813.

### **ADSP U01 Awards**

#### **U01 AG052411 - Identification and Characterization of AD Risk Networks Using Multi-dimensional Omics Data**

71. Horgusluoglu E, Neff R, Song WM, et al; Alzheimer's Disease Neuroimaging Initiative (ADNI); Alzheimer Disease Metabolomics Consortium. [Integrative metabolomics-genomics approach reveals key metabolic pathways and regulators of Alzheimer's disease](#). *Alzheimers Dement*. 2022 Jun;18(6):1260-1278. doi: 10.1002/alz.12468. Epub 2021 Nov 10. PMID: 34757660

#### **U01 AG052409 – ADSP Follow-up in Multi-Ethnic Cohorts via Endophenotypes, Omics, & Model Systems**

72. Mishra A, Duplaà C, Vojinovic D, Suzuki H, Sargurupremraj M, Zilhão NR et al. [Gene-mapping study of extremes of cerebral small vessel disease reveals TRIM47 as a strong candidate](#). *Brain*. 2022. doi: 10.1093/brain/awab432. PubMed PMID: 35511193.
73. Tin A, Bressler J, Simino J, Sullivan KJ, Mei H, Windham BG et al. [Genetic Risk, Midlife Life's Simple 7, and Incident Dementia in the Atherosclerosis Risk in Communities Study](#). *Neurology*. 2022. doi: 10.1212/WNL.0000000000200520. PubMed PMID: 35613930.
74. Yang Y, Knol MJ, Wang R, Mishra A, Liu D, Luciano M et al. [Epigenetic and integrative cross-omics analyses of cerebral white matter hyperintensities on MRI](#). *Brain*. 2022. doi: 10.1093/brain/awac290. PMID: 35943854.

#### **U01 AG057659 - Whole Genome Sequencing in Ethnically Diverse Cohorts for the ADSP Follow-up Study (FUS)**

75. DeRosa BA et al. [Generation of two iPSC lines \(UMio38-A & UMio39-A\) from siblings bearing an Alzheimer's disease-associated variant in SORL1](#). *Stem Cell Res*. 2022 Jul;62:102823. doi: 10.1016/j.scr.2022.102823. PMID: 35671596.
76. Godrich D, Martin ER, Schellenberg G et al. [Neuropathological lesions and their contribution to dementia and cognitive impairment in a heterogeneous clinical population](#). *Alzheimer's Dement*. 2022 Feb 9. PMID: 35142102. Online ahead of print.

#### **U01 AG058635 – Genomic Approach to Identification of Microglial Networks Involved in Alzheimer Disease Risk**

77. Martens YA, Zhao N, Liu CC et al. [ApoE Cascade Hypothesis in the pathogenesis of Alzheimer's disease and related dementias](#). *Neuron*. 2022 Apr 20;110(8):1304-1317. doi: 10.1016/j.neuron.2022.03.004. Epub 2022 Mar 16. PMID: 35298921 Review.

#### **U01 AG058635 - Genomic Approach to Identification of Microglial Networks Involved in Alzheimer Disease Risk**

78. Saroja SR, Gorbachev K, Julia T et al. [Astrocyte-secreted glypican-4 drives APOE4-dependent tau hyperphosphorylation](#). *Proc Natl Acad Sci U S A*. 2022 Aug 23;119(34):e2108870119. doi: 10.1073/pnas.2108870119. Epub 2022 Aug 15. PMID: 35969759.

#### **U01 AG058589 – Therapeutic Target Discovery in ADSP Data via Comprehensive Whole-Genome Analysis Incorporating Ethnic Diversity and Systems Approaches**

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79. Wang Y, Chen H, Peloso GM et al. [Exploiting family history in aggregation unit-based genetic association tests](#). Eur J Hum Genet. 2021. doi: 10.1038/s41431-021-00980-0. PMID: 34690355.
80. Wang Y, Chen H, Peloso GM et al. [Family history aggregation unit-based tests to detect rare genetic variant associations with application to the Framingham Heart Study](#). Am J Hum Genet. 2022. 109(4):738-749. doi: 10.1016/j.ajhg.2022.03.001. PMID: 35316615.

### Therapeutic Targets

#### **5 U01 AG058635-052022 - Genomic Approach to Identification of Microglial Networks Involved in Alzheimer Disease Risk**

81. TCW J, Qian L, Pipalia NH et al. [Cholesterol and matrisome pathways dysregulated in astrocytes and microglia](#). Cell. 2022 Jun 23;185(13):2213-2233.e25. doi: 10.1016/j.cell.2022.05.017. PMID: 35750033.

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#### **U01 AG072577 - Circular RNAs and Their Interactions With RNA-Binding Proteins to Modulate AD-Related Neuropathology**

82. Olayinka OA, O'Neill NK, Farrer LA et al. [Molecular Quantitative Trait Locus Mapping in Human Complex Diseases](#). Curr Protoc. 2022 May;2(5):e426. doi: 10.1002/cpz1.426. PMID: 35587224.

#### **U01 AG072439 - Functional Genomic Dissection of Alzheimer's Disease in Humans and Drosophila Models**

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84. Ma M, Moulton MJ, Lu S, Bellen HG. ['Fly-ing' from rare to common neurodegenerative disease mechanisms](#). Trends Genet. 2022 Sep;38(9):972-984. doi: 10.1016/j.tig.2022.03.018. Epub 2022 Apr 25. PMID: 35484057; PMCID: PMC9378361.

#### **U01 AG072464 - Investigating the Functional Impact of AD Risk Genes on Neuro-Vascular Interactions**

85. Cooper YA, Teyssier N, Dräger NM et al. [Functional regulatory variants implicate distinct transcriptional networks in dementia](#). Science. 2022 Aug 19;377(6608):eabi8654. doi: 10.1126/science.abi8654. Epub 2022 Aug 19. PMID: 35981026.
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#### **U01 AG072573 Multi-omic Functional Assessment of Novel AD Variants Using High-throughput and Single-cell Technologies**

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88. Smail C, Ferraro NM, Hui Q et al; Million Veteran Program. [Integration of rare expression outlier-associated variants improves polygenic risk prediction](#). Am J Hum Genet. 2022 Jun 2;109(6):1055-1064.

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### **R01 AG067501 - Genetic Epidemiology and Multi-Omics Analyses in Familial and Sporadic Alzheimer's Disease Among Secular Caribbean Hispanics and Religious Order**

89. Kizil C et al. [Admixture Mapping of Alzheimer's disease in Caribbean Hispanics identifies a new locus on 22q13.1](#). Mol Psychiatry. 2022 Jun;27(6):2813-2820. doi: 10.1038/s41380-022-01526-6. PMID: 35365809 PMCID: PMC9167722.
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#### *Genomic approach to identification of microglial networks involved in Alzheimer disease risk*

#### *Identification and characterization of AD risk networks using multi-dimensional omics data*

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## 2020

### **Consortia within the ADSP**

#### *General*

1. Writing Committee for the E-CNVWG, van der Meer D, Sonderby IE, Kaufmann T, Walters GB, Abdellaoui A, et al. [Association of Copy Number Variation of the 15q11.2 BP1-BP2 Region With Cortical and Subcortical Morphology and Cognition](#). JAMA Psychiatry. 2020;77(4):420-30. doi: 10.1001/jamapsychiatry.2019.3779. PubMed PMID: 31665216; PubMed Central PMCID: PMC6822096.
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