

# ADSP Publication List

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 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**

7. Osterman MD et al. [The genetic architecture of Alzheimer disease risk in the Ohio and Indiana Amish](#). *HGG Adv.* 2022 Apr 27;3(3):100114. doi: 10.1016/j.xhgg.2022.100114. PMID: 35599847 PMCID: PMC9114685.
8. Ramos J et al. [Genetic variants in the SHISA6 gene are associated with delayed cognitive impairment in two family datasets](#). *Alzheimers Dement.* 2022 May 1. doi: 10.1002/alz.12686. PMID: 35490390.

## ADSP Publication List

### India

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

- Smith J et al. [Association between episodic memory and genetic risk factors for Alzheimer's Disease in South Asians from the Longitudinal Aging Study in India – Diagnostic Assessment of Dementia \(LASI-DAD\)](#). *Journal of the American Geriatrics Society*, 2020, 68: S45 – S53. doi:10.1111/jgs.16735. PMID:32815605 PMCID:PMC7507858.

### Korea

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

- Ahn H, Yi D, Chu K, Joung H, Lee Y, Jung G, Sung K, Han D, Lee JH, Byun MS, Lee DY. [Functional Neural Correlates of Semantic Fluency Task Performance in Mild Cognitive Impairment and Alzheimer's Disease: An FDG- PET Study](#). *J Alzheimers Dis*. 2022;85(4):1689-1700. doi: 10.3233/JAD-215292. PubMed PMID: 34958036; PubMed Central PMCID: PMC9210291.
- Grothe MJ, Moscoso A, Silva-Rodríguez J, Lange C, Nho K, Saykin AJ, Nelson PT, Schöll M, Buchert R, Teipel S. [Differential diagnosis of amnesic dementia patients based on an FDG-PET signature of autopsy-confirmed LATE-NC](#). *Alzheimers Dement*. 2022 Aug 15;. doi: 10.1002/alz.12763. [Epub ahead of print] PubMed PMID: 35971593.
- Kim JW, Byun MS, Yi D, Lee JH, Kim MJ, Jung G, Lee JY, Kang KM, Sohn CH, Lee YS, Kim YK, Lee DY. [Serum zinc levels and in vivo beta-amyloid deposition in the human brain](#). *Alzheimers Res Ther*. 2021 Nov 19;13(1):190. doi: 10.1186/s13195-021-00931-3. PubMed PMID: 34798903; PubMed Central PMCID: PMC8605596.
- Kim JP, Kim BH, Bice PJ, Seo SW, Bennett DA, Saykin AJ, Nho K. [BMI1 is associated with CS8F amyloid-β and rates of cognitive decline in Alzheimer's disease](#). *Alzheimers Res Ther*. 2021 Oct 5;13(1):164. doi: 10.1186/s13195-021-00906-4. PubMed PMID: 34610832; PubMed Central PMCID: PMC8493672.
- Ko K, Yi D, Byun MS, Lee JH, Jeon SY, Kim WJ, Byeon G, Sung K, Han D, Lee Y, Joung H, Jung G, Lee JY, Kim H, Kim YK, Kang KM, Sohn CH, Lee DY. [Cognitive reserve proxies, Alzheimer pathologies, and cognition](#). *Neurobiol Aging*. 2022 Feb;110:88-95. doi: 10.1016/j.neurobiolaging.2021.10.005. Epub 2021 Oct 14. PubMed PMID: 34879329; PubMed Central PMCID: PMC9234822.
- Park YH, Pyun JM, Hodges A, Jang JW, Bice PJ, Kim S, Saykin AJ, Nho K. [Dysregulated expression levels of APOE4 in peripheral blood are associated with brain atrophy and amyloid-β deposition in Alzheimer's disease](#). *Alzheimers Res Ther*. 2021 Nov 3;13(1):183. doi: 10.1186/s13195-021-00919-z. PubMed PMID: 34732252; PubMed Central PMCID: PMC8567578.
- 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-β 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.

## ADSP Publication List

17. Wells LF, Risacher SL, McDonald BC, Farlow MR, Brosch J, Gao S, Apostolova LG, Saykin AJ. [Measuring Subjective Cognitive Decline in Older Adults: Harmonization Between the Cognitive Change Index and the Measurement of Everyday Cognition Instruments](#). J Alzheimers Dis. 2022;87(2):761-769. doi: 10.3233/JAD-215388. PubMed PMID: 35367962; PubMed Central PMCID: PMC9169561.

### U01 AG062602 - Genetic Studies of Alzheimer Disease in Koreans

18. Park J, Lee D, Lee JJ et al. [A novel missense mutation in SHARPIN is associated with Alzheimer's disease](#). Transl Psychiatry 2021; 11:590. PMID: 34785643.

### **ADSP Collaborations with Other Consortia/International Groups**

19. Bellenguez et al. [New insights into the genetic etiology of Alzheimer's disease and related dementias](#). Nature Genetics. 2022 Apr;54(4):412-436. doi: 10.1038/s41588-022-01024-z. PMID: 35379992 PMCID: PMC9005347.
20. Belloy ME, Le Guen Y, Eger SJ et al. [A fast and robust strategy to remove variant level artifacts in Alzheimer's Disease Sequencing Project data](#). Neurol Genet, 8(5), e200012. PMID: 35966919.
21. Belloy ME, Eger SJ, Le Guen Y et al. [Challenges at the APOE locus: A robust quality control approach for accurate APOE genotyping](#). Alzheimers Res Ther, 14:22 (2022). PMID: 35120553.
22. Chen H-H et al. [Genetically regulated expression in late-onset Alzheimer's disease implicates risk genes within known and novel loci](#). Transl Psychiatry. 2021 Dec 6;11(1):618. doi: 10.1038/s41398-021-01677-0. PMID: 34873149 PMCID: PMC8648734.
23. Eger SJ, Le Guen Y, Khan RR et al. [Confirming pathogenicity of the F386L PSEN1 variant in a South Asian family with early-onset Alzheimer's disease](#). Neurol Genet, 8(1):e647 (2021). PMID: 34901437.
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 .
25. He Z, Le Guen Y, Liu L et al. [Genome-wide analysis of common and rare variants via multiple knockoffs at biobank-scale, with an application to Alzheimer's disease genetics](#). Am J Hum Genet, 108(12):2336-2353 (2021). PMID: 34767756.
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.
27. Le Guen Y, Belloy ME, Grenier-Boley B et al; Members of the EADB, GR@ACE, DEGESCO, DemGene, GERAD, and EADI Groups et al. [Association of Rare APOE Missense Variants V236E and R251G With Risk of Alzheimer Disease](#). JAMA Neurol. 2022 Jul 1;79(7):652-663. doi: 10.1001/jamaneurol.2022.1166. PMID: 35639372; PMCID: PMC9157381.
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.

## ADSP Publication List

30. Sexton C, Snyder H, Beher 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.

## **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.
38. Cosacak MI et al. [Single Cell/Nucleus Transcriptomics Comparison in Zebrafish and Humans Reveals Common and Distinct Molecular Responses to Alzheimer's Disease](#). *Cells*. 2022 May 31;11(11):1807. doi: 10.3390/cells11111807. PMID: 35681503 PMCID: PMC9180693.
39. Dressman D et al. [Genotype-Phenotype Correlation of T Cell Subtypes Reveals Senescent and Cytotoxic Genets in Alzheimer's Disease](#). *Hum Mol Genet*. 2022 May 28;ddac126. doi: 10.1093/hmg/ddac126. PMID: 35640154.
40. Frank B et al. [Plasma p-tau181 shows stronger network association to Alzheimer's disease dementia than neurofilament light and total tau](#). *Alzheimers Dement*. 2022 Aug;18(8):1523-1536. doi: 10.1002/alz.12508. PMID: 34854549 PMCID: PMC9160800.

## ADSP Publication List

41. Lao PJ et al. [Amyloid, cerebrovascular disease, and neurodegeneration biomarkers are associated with cognitive trajectories in a racially and ethnically diverse, community-based sample](#). Neurobiol Aging. 2022 Sep;117:83-96. doi: 10.1016/j.neurobiolaging.2022.05.004. PMID: 35679806.
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.
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.
48. Siddiqui T et al. [KYNA/Ahr Signaling Suppresses Neural Stem Cell Plasticity and Neurogenesis in Adult Zebrafish Model of Alzheimer's Disease](#). Cells. 2021 Oct 14;10(10):2748. doi: 10.3390/cells10102748. PMID: 34685728 PMCID: PMC8534484.
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.
50. Silva TC et al. [Distinct sex-specific DNA methylation differences in Alzheimer's disease](#). Alzheimers Res Ther. 2022 Sep 15;14(1):133. doi: 10.1186/s13195-022-01070-z. PMID: 36109771 PMCID: PMC9479371.
51. Silva TC, Young JI, Martin ER et al. (2022) [MethReg: estimating the regulatory potential of DNA methylation in gene transcription](#). Nucleic Acids Res. 2022 May 20;50(9):e51. doi: 10.1093/nar/gkac030. PMID: 35100398; PMCID: PMC9122535.
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.
54. Zahodne LB et al. [Longitudinal associations between racial discrimination and hippocampal and white matter hyperintensity volumes among older Black adults](#). Soc Sci Med. 2022 Feb 7;114789. doi: 10.1016/j.socscimed.2022.114789. PMID: 35164975.

## ADSP Publication List

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  \$\beta\$  levels are driven by genetic variants near APOE, BACE1, APP, PSEN2: A genome-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.
60. Hou J, Hess J, Armstrong N, Bis JC, Grenier-Boley B, Karlsson IK et al. [Polygenic resilience scores capture protective genetic effects for Alzheimer's disease](#). *Transl Psychiatry*. 2022. 12(1):296. doi: 10.1038/s41398-022-02055-0. PMID: 35879306.
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.
62. Le Grand Q, Satizabal CL, Sargurupremraj M, Mishra A, Soumaré A, Laurent A et al. [Genomic studies across the lifespan point to early mechanisms determining subcortical volumes](#). *Biol Psychiatry Cogn Neurosci Neuroimaging*. 2022. 7(6):616-628. doi: 10.1016/j.bpsc.2021.10.011. PMID: 34700051.
63. Madrid L, Moreno-Grau S, Ahmad S, González-Pérez A, de Rojas I, Xia R et al. [Multiomics integrative analysis identifies APOE allele-specific blood biomarkers associated to Alzheimer's disease etiopathogenesis](#). *Aging (Albany NY)*. 2021. 13(7):9277-9329. doi: 10.18632/aging.202950. PMID: 33846280.
64. Mishra A, Malik R, Hachiya T, Jürgenson T, Namba S, Posner D et al., [Stroke genetics informs drug discovery and risk prediction across ancestries](#). *Nature* (2022). <https://doi.org/10.1038/s41586-022-05165-3> (published Sept 30, 2022 PMID pending)
65. Pinheiro A, Demissie S, Scruton A, Charidimou A, Parva P, DeCarli C et al. [Association of Apolipoprotein E  \$\epsilon\$ 4 Allele with Enlarged Perivascular Spaces](#). *Ann Neurol*. 2022. 92(1):23-31. doi: 10.1002/ana.26364. PMID: 35373386.
66. Sarnowski C, Ghanbari M, Bis JC, Logue M, Fornage M, Mishra A et al. [Meta-analysis of genome-wide association studies identifies ancestry-specific associations underlying circulating total tau levels](#). *Commun Biol*. 2022. 5(1):336. doi: 10.1038/s42003-022-03287-y PMID: 35396452.

## ADSP Publication List

67. Sliz E, Shin J, Ahmad S, Williams DM, Frenzel S, Gauß F et al. [Circulating metabolome and white matter hyperintensities in women and men](#). *Circulation*. 2022. 145(14):1040-1052. doi: 10.1161/CIRCULATIONAHA.121.056892. PMID: 35050683.

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

68. Chung J, Das A, Sun X et al. [Genome-wide association and multi-omics studies identify \*MGMT\* as a novel risk gene for Alzheimer disease among women](#). *Alzheimer Dement*. 2022. Online ahead of print. PMID: 35770850.
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 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.

## ADSP Publication List

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**

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.

## **ADSP: Functional Genomics Consortium**

### **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**

83. Moulton MJ, Barish S, Ralhan I. [Neuronal ROS-induced glial lipid droplet formation is altered by loss of Alzheimer's disease-associated genes](#). Proc Natl Acad Sci U S A. 2021 Dec 28;118(52):e2112095118. doi: 10.1073/pnas.2112095118. PMID: 34949639; PMCID: PMC8719885.



## ADSP Publication List

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

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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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### *AI4AD (U01AG068057) - Ultrascale Machine Learning to Empower Discovery in Alzheimers Disease Biobanks*

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### R01s and other mechanisms

#### *RF1AG044546*

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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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PMC7018232 and has served as a consultant to AbbVie, Biogen, Eisai, Illumina, and GSK. Other authors declare no competing financial interests.

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## **U01 Awards**

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