

## Alzheimer's Disease Genetics Consortium (ADGC) NIA grant U01AG032984 Collaboration and Data Sharing Summary

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The ADGC was formed to collaboratively use the collective resources of the AD research community to identify Alzheimer's disease (AD) genes. These genes include those that cause AD, increase or decrease AD risk, and protect against AD. The ADGC includes investigators with the clinical, neuropathological, molecular and statistical expertise needed to analyze the complex phenotype of AD.

The ADGC assembled genetic and phenotype data for a large number of AD cases and cognitively normal elderly controls. For non-Hispanic whites (NHW), subjects include 17,876 cases and 19,440 controls (Table 1A). For other ethnic groups (African Americans, Caribbean Hispanic, non-Caribbean Hispanics, and Asians), the subjects include 9,392 AD cases and 15,125 controls (Table 1B). Through a variety of approaches, the ADGC continues to add to this collection of cohorts. The following describes the collaborations established and the data sharing activities of the ADGC.

**I. ADGC collaborative network.** The ADGC collaborates with 21 different cohorts in the US (Table 1A, B). The largest cohort is from the 31 NIA-funded Alzheimer's Disease Centers (ADC's) which have contributed 7,428 cases and 7,342 controls to ADGC studies. While the majority of subjects are non-Hispanic Caucasians (Table 1A), the ADGC also assembled and analyzed genetic and phenotype data for Hispanics, African Americans, and Asians (Table 1B). Each of these cohorts has extensive phenotype data that can be used for genetic analysis.

#### **A. The ADGC national collaborative network.**

1. The 31 ADCs, characterize AD subjects, MCI, and normal controls, and collect blood/DNA for these subjects. The ADCs have provided over 24,267 subjects for ADGC studies including non-Hispanic Whites, Hispanics, African Americans, Native American, and Asian subjects (Table 1A, B).
2. The National Alzheimer's Coordinating Center (NACC) collects phenotype data from the ADCs. NACC works with the ADGC to identify subjects appropriate for genetic studies and provides phenotype data for these genetic studies.
3. The National Cell Repository for AD (NCRAD) which, in collaboration with the ADGC and NACC, collects blood and DNA from ADCs and other studies. NCRAD then provides these samples to the ADGC for genotyping and sequencing.
4. The National Institute on Alzheimer's Genetics of Alzheimer's Disease Storage (NIAGADS) site provides qualified access data sharing for ADGC data.
5. The Alzheimer's Disease Sequence Project (ADSP) is generating and analyzing DNA sequence data for AD, ADRD, and normal control subjects. The ADGC provides samples, genotype data, and phenotype data to the ADSP for samples undergoing whole exome (WES) and whole-genome sequencing (WGS).

**Table 1A. ADGC non-Hispanic White cohorts**

Cohort	Cases	Controls	Other	Total
ACT	532	1571		2,103
ACT3	pending	pending		1,551
ADC1	1549	512		2,061
ADC2	727	156		883
ADC3	894	586		1,480
ADC4	304	377		681
ADC5	286	505		791
ADC6	213	338		551
ADC7	566	878		1,444
ADC8	517	664		1,181
ADC9	728	896		1,624
ADC10	457	724		1,181
ADC11	883	1265		2,148
ADC12	304	441	377	1,122
ADNI	268	173		441
BIOCARD	6	122		128
CHAP	27	144		171
EAS	9	141		150
GSK	666	712		1,378
MAYO	658	1046		1,704
MIRAGE	491	738		1,229
MTV	256	189		445
NBB	80	48		128
NIA-LOAD	1788	1568		3,356
OHSU	132	153		285
PFIZER	696	762		1,458
RMAYO	13	233		246
ROSMAP	354	986		1,340
TARCC	323	181		504
TGEN	668	365		1,033
UCSD ADCS	pending	pending		1,319
UKS	596	170		766
UMVUMSSM	1177	1126		2,303
UPITT	1255	829		2,084
WASHU	339	187		526
WASHU2	38	94		132
WHICAP	76	560		636
<b>TOTAL</b>	<b>17,876</b>	<b>19,440</b>	<b>377</b>	<b>40,563</b>

6. The ADGC generated WES data for ~3,200 African Americans and will be generating WGS for cohorts from Iceland, Mexico, and US Hispanics. These data will be contributed to the ADSP for joint analyses.

7. The Principle Investigators of the cohorts listed in Table 1A are ADGC collaborators. These cohorts provide the ADGC with samples, genetic data, and phenotype data on a collaborative basis.

In addition to these collaborators, the ADGC uses data from multiple public databases such as AMP-AD (SAGE), ENCODE, PSYCENCODE, eQTL databases, multiple expression data sets, MAGMA, dbGaP, and numerous other sources.

**B. The ADGC and international collaborations.** In 2011<sup>1</sup>, an international collaboration in AD genetics called the International Genomics Project (IGAP), was founded by the ADGC (Schellenberg) with investigators from Cohorts for Heart and Aging Research in Genomic Epidemiology (CHARGE, Seshadri), European Alzheimer's Disease Initiative (EADI, Amouyel), and Genetic and Environmental Risk in Alzheimer's Disease (GERAD, Williams). IGAP was formed to assemble the largest possible sample to increase power for AD genetics studies. ADGC with IGAP published a number of seminal manuscripts on AD genetics<sup>2-4</sup> including the largest genome-wide association study of AD performed to date (35,274 cases and 59,163 controls)<sup>2</sup>. In all, the ADGC/IGAP collaborations produced 13 primary manuscripts on AD genetics (included in Appendix A, pages 1-33). In addition, non-ADGC/IGAP investigators used ADGC/IGAP summary statistics in 47 publications (Appendix A pp 27-33). IGAP is expanding, adding the EADI (Lambert), representing European collaborators, and the GRA@CE group from Spain. In November 2018 and December 2019, IGAP group meetings were held in Paris and London, respectively. The following is a summary of these meetings with detailed information in Appendices B and C.

1. There were 52 attendees at the 2018 Paris meeting (Appendix B), and 58 attendees at the 2019 London meeting (Appendix C). For both meetings, there were attendees from 8 countries, representing 9 global collaborations

2. Countries represented: Belgium, Denmark, France, Germany, Netherlands, Spain, United Kingdom, United States

3. Genetic data from 26 countries: Argentina, Australia, Austria, Belgium, Chile, China, Czech Republic, Denmark, Dominican Republic, Finland, France, Germany, Greece, Iceland, Italy, Japan, Korea, Mexico, Netherlands, Norway, Puerto Rico, Spain, Sweden, Taiwan, United Kingdom, United States

The ADGC has added additional international collaborations with investigators from Korea (Kunho Lee), India (Jinkook Lee, LASI-DAD), Iceland (AGES-RS, Vilmundur Guðnason) (Appendix D), multiple Caribbean nations, Mexico, Peru, and other South American countries (Pericak-Vance, Mayeux). These new cohorts will be part of future IGAP projects.

**II. Data Sharing.** The ADGC encourages the use of data and attempts to make data available to researchers as soon as possible. In addition to accessing data through dbGaP and NIAGADS, ADGC members can submit analysis proposals to use unpublished data (Special Analysis Groups - SAGs). The ADGC approved 129 data requests through this process, 12 during the past year. Data provided is the latest data set including imputed genotypes, and cleaned phenotype data appropriate for the proposed studies. The genetic data provided has been extensively cleaned and analyzed to account for population stratification, and genotype batch effects. Appendix E lists approved SAGs (pages 1-17). ADGC SAGs include;

- Total approved SAGs = 129
- Average SAGs per year = 14.3
- Number of represented institutions = 43
- Number of lead investigators = 72

The ADGC data is also available *via* qualified access from NIAGADS. The GWAS datasets are available as well as secondary analysis results and summary statistics (Appendix F). NIAGADS created a designated page to collate the ADGC data to facilitate finding these data [<https://www.niagads.org/resources/related-projects/alzheimers-disease-genetics-consortium-adgc-collection>].

Summary statistics are also available for the latest ADGC-IGAP publication through NIAGADS (Appendix F). While individual-level data for ADGC studies are available through NIAGADS, due to the General Data Protection Rule (GDPR) governing European data, individual-level data are not available for the European data used in IGAP publications. IGAP summary statistics were used in 47 publications that are not from IGAP or the ADGC and do not have ADGC authorship (Appendix A, pp 27-33).

The ADGC generated whole exome sequence data on 3,159 subjects. These data were analyzed by GCAD and are part of GCAD release 2. Release 2 including the ADGC African American subjects will be released by NIAGADS February 2020 after QC is completed.

**III. Publications.** The core analyses, secondary analyses, and SAGs have resulted in 140 ADGC publications, 18 in the past year (Appendix A, pp 1-23). Analyses of genetic data funded by the ADGC account for an additional 31 publications (Appendix A, pp 23-27), and 47 publications state the use of IGAP summary statistics which include ADGC data (Appendix A, pp 27-33).

- (2010-2019) ADGC Publications – 140 (average of 15 publications per year)
- (2012-2019) Additional Publications using ADGC Supported Studies - 31
- (2016-2019) Additional Publications using IGAP Summary Statistics - 47

**Table 1B.** Ethnicity of ADGC cohorts

ADGC subjects				
Ethnic group	AD	MCI	NC	Total
NHW	17,876	2,333	19,440	39,649
AA	2,845	418	5,271	8,143
CH	3,859	-	6,365	10,224
NCH	351	194	216	761
Asian	2,337	90	3,273	5,700

**IV. Member involvement.** To support coordination of efforts within the ADGC, the ADGC holds monthly conference calls, open to all members, to update each other on the status of projects and plan the next steps. The ADGC also holds an annual meeting with an external advisory board. The ADGC held its 10<sup>th</sup> annual meeting in 2019 with 75 members attending, representing 24 institutions, including investigators from Germany, Canada, and South Korea.

- 2009 – 31 participants from 18 institutions
- 2014 – 56 participants from 22 institutions
- 2019 – 75 participants from 24 institutions

In 2019, the ADGC held the ADGC 10-Year Alzheimer's Disease Genetics Symposium (Appendix G). The ADGC and Penn Neurodegeneration Genomics Center (PNGC) hosted a 2-day Alzheimer's Disease Genetics Symposium, to review progress achieved in the past 10 years and to plan future studies. The symposium included 21 speakers from 18 institutions within the US and 3 international speakers from Spain, France, and Iceland. The ADGC 10-Year symposium provided a time and space to discuss additional collaborations. The focus of the talks brought together leaders in genetics, as well as pharmaceutical and technology representatives. (Appendix G).

- Total number of attendees: 194
- Number of Institutions: 45
- 21 speakers from 18 institutions
- Countries represented: 4

**V. Studies Contributing Data to the ADGC.** Cohort contributions are listed in Table 1A with the ethnic breakdown in Table 1B. Study abbreviations are in Appendix H.

### References

1. Holmes, D. Mind the IGAP. *Lancet Neurol* **10**, 502-503 (2011).
2. Kunkle, B.W., *et al.* Genetic meta-analysis of diagnosed Alzheimer's disease identifies new risk loci and implicates Abeta, tau, immunity and lipid processing. *Nat Genet* **51**, 414-430 (2019).
3. Sims, R., *et al.* Rare coding variants in PLCG2, ABI3, and TREM2 implicate microglial-mediated innate immunity in Alzheimer's disease. *Nat Genet* **49**, 1373-1384 (2017).
4. Lambert, J.C., *et al.* Meta-analysis of 74,046 individuals identifies 11 new susceptibility loci for Alzheimer's disease. *Nat Genet* **45**, 1452-1458 (2013).

## Appendix A

### ADGC Publications

1. Shen L, Qi Y, Kim S, Nho K, Wan J, Risacher SL, Saykin AJ; ADNI. **Sparse bayesian learning for identifying imaging biomarkers in AD prediction.** Med Image Comput Assist Interv. 2010 Sept;13(Pt 3):611-8. PMID: 20879451. PMCID: PMC2951627.
2. Jun G, Naj AC, Beecham GW, Wang LS, Buros J, Gallins PJ, Buxbaum JD, Ertekin-Taner N, Fallin MD, Friedland R, Inzelberg R, Kramer P, Rogaeva E, St George-Hyslop P; Alzheimer's Disease Genetics Consortium, Cantwell LB, Dombroski BA, Saykin AJ, Reiman EM, Bennett DA, Morris JC, Lunetta KL, Martin ER, Montine TJ, Goate AM, Blacker D, Tsuang DW, Beekly D, Cupples LA, Hakonarson H, Kukull W, Foroud TM, Haines J, Mayeux R, Farrer LA, Pericak-Vance MA, Schellenberg GD. **Meta-analysis confirms CR1, CLU, and PICALM as Alzheimer disease risk loci and reveals interactions with APOE genotypes.** Arch Neurol. 2010 Dec;67(12):1473-84. doi: 10.1001/archneurol.2010.201. Epub 2010 Aug 9. Erratum in: Arch Neurol. 2011 Feb;68(2):159. PMID: 20697030. PMCID: PMC3048805.
3. Sherva R, Farrer LA. **Power and pitfalls of the genome-wide association study approach to identify genes for Alzheimer's disease.** Curr Psychiatry Rep. 2011 Apr;13(2):138-46. doi: 10.1007/s11920-011-0184-4. PMID: 2132009. PMCID: PMC3154249.
4. Naj AC, Jun G, Beecham GW, Wang LS, Vardarajan BN, Buros J, Gallins PJ, Buxbaum JD, Jarvik GP, Crane PK, Larson EB, Bird TD, Boeve BF, Graff-Radford NR, De Jager PL, Evans D, Schneider JA, Carrasquillo MM, Ertekin-Taner N, Younkin SG, Cruchaga C, Kauwe JS, Nowotny P, Kramer P, Hardy J, Huentelman MJ, Myers AJ, Barmada MM, Demirci FY, Baldwin CT, Green RC, Rogaeva E, St George-Hyslop P, Arnold SE, Barber R, Beach T, Bigio EH, Bowen JD, Boxer A, Burke JR, Cairns NJ, Carlson CS, Carney RM, Carroll SL, Chui HC, Clark DG, Corneveaux J, Cotman CW, Cummings JL, DeCarli C, DeKosky ST, Diaz-Arrastia R, Dick M, Dickson DW, Ellis WG, Faber KM, Fallon KB, Farlow MR, Ferris S, Frosch MP, Galasko DR, Ganguli M, Gearing M, Geschwind DH, Ghetti B, Gilbert JR, Gilman S, Giordani B, Glass JD, Growdon JH, Hamilton RL, Harrell LE, Head E, Honig LS, Hulette CM, Hyman BT, Jicha GA, Jin LW, Johnson N, Karlawish J, Karydas A, Kaye JA, Kim R, Koo EH, Kowall NW, Lah JJ, Levey AI, Lieberman AP, Lopez OL, Mack WJ, Marson DC, Martiniuk F, Mash DC, Masliah E, McCormick WC, McCurry SM, McDavid AN, McKee AC, Mesulam M, Miller BL, Miller CA, Miller JW, Parisi JE, Perl DP, Peskind E, Petersen RC, Poon WW, Quinn JF, Rajbhandary RA, Raskind M, Reisberg B, Ringman JM, Roberson ED, Rosenberg RN, Sano M, Schneider LS, Seeley W, Shelanski ML, Slifer MA, Smith CD, Sonnen JA, Spina S, Stern RA, Tanzi RE, Trojanowski JQ, Troncoso JC, Van Deerlin VM, Vinters HV, Vonsattel JP, Weintraub S, Welsh-Bohmer KA, Williamson J, Woltjer RL, Cantwell LB, Dombroski BA, Beekly D, Lunetta KL, Martin ER, Kamboh MI, Saykin AJ, Reiman EM, Bennett DA, Morris JC, Montine TJ, Goate AM, Blacker D, Tsuang DW, Hakonarson H, Kukull WA, Foroud TM, Haines JL, Mayeux R, Pericak-Vance MA, Farrer LA, Schellenberg GD. **Common variants at MS4A4/MS4A6E, CD2AP, CD33 and EPHA1 are associated with late-onset Alzheimer's disease.** Nat Genet. 2011 May;43(5):436-41. doi: 10.1038/ng.801. Epub 2011 Apr 3. PMID: 21460841. PMCID: PMC3090745.
5. Liu N, Landreh M, Cao K, Abe M, Hendriks GJ, Kennerdell JR, Zhu Y, Wang LS, Bonini NM. **The microRNA miR-34 modulates ageing and neurodegeneration in Drosophila.** Nature. 2012 Feb 15;482(7386):519-23. doi: 10.1038/nature10810. PMID: 2234898. PMCID: PMC3326599.
6. Barral S, Bird T, Goate A, Farlow MR, Diaz-Arrastia R, Bennett DA, Graff-Radford N, Boeve BF, Sweet RA, Stern Y, Wilson RS, Foroud T, Ott J, Mayeux R; National Institute on Aging Late-Onset Alzheimer's Disease Genetics Study. **Genotype patterns at PICALM, CR1, BIN1, CLU, and APOE genes are associated with episodic memory.** Neurology. 2012 May 8;78(19):1464-71. doi: 10.1212/WNL.0b013e3182553c48. Epub 2012 Apr 25. PMID: 22439578. PMCID: PMC3345618.

7. Zou F, Chai HS, Younkin CS, Allen M, Crook J, Pankratz VS, Carrasquillo MM, Rowley CN, Nair AA, Middha S, Maharjan S, Nguyen T, Ma L, Malphrus KG, Palusak R, Lincoln S, Bisceglia G, Georgescu C, Kouri N, Kolbert CP, Jen J, Haines JL, Mayeux R, Pericak-Vance MA, Farrer LA, Schellenberg GD; Alzheimer's Disease Genetics Consortium, Petersen RC, Graff-Radford NR, Dickson DW, Younkin SG, Ertekin-Taner N. **Brain expression genome-wide association study (eGWAS) identifies human disease-associated variants.** PLoS Genet. 2012 June;8(6):e1002707. doi: 10.1371/journal.pgen.1002707. Epub 2012 Jun 7. PMID: 22685416. PMCID: PMC3369937.
8. Allen M, Zou F, Chai HS, Younkin CS, Crook J, Pankratz VS, Carrasquillo MM, Rowley CN, Nair AA, Middha S, Maharjan S, Nguyen T, Ma L, Malphrus KG, Palusak R, Lincoln S, Bisceglia G, Georgescu C, Schultz D, Rakhshan F, Kolbert CP, Jen J, Haines JL, Mayeux R, Pericak-Vance MA, Farrer LA, Schellenberg GD; Alzheimer's Disease Genetics Consortium, Petersen RC, Graff-Radford NR, Dickson DW, Younkin SG, Ertekin-Taner N. **Novel late-onset Alzheimer disease loci variants associate with brain gene expression.** Neurology. 2012 Jul 17;79(3):221-8. doi: 10.1212/WNL.0b013e3182605801. Epub 2012 Jun 20. PMID: 22722634. PMCID: PMC3398432.
9. Ramanan VK, Shen L, Moore JH, Saykin AJ. **Pathway analysis of genomic data: concepts, methods, and prospects for future development.** Trends Genet. 2012 Jul;28(7):323-32. doi: 10.1016/j.tig.2012.03.004. Epub 2012 Apr 3. PMID: 22480918. PMCID: PMC3378813.
10. Coppola G, Chinnathambi S, Lee JJ, Dombroski BA, Baker MC, Soto-Ortolaza AI, Lee SE, Klein E, Huang AY, Sears R, Lane JR, Karydas AM, Kenet RO, Biernat J, Wang LS, Cotman CW, Decarli CS, Levey AI, Ringman JM, Mendez MF, Chui HC, Le Ber I, Brice A, Lupton MK, Preza E, Lovestone S, Powell J, Graff-Radford N, Petersen RC, Boeve BF, Lippa CF, Bigio EH, Mackenzie I, Finger E, Kertesz A, Caselli RJ, Gearing M, Juncos JL, Ghetti B, Spina S, Bordelon YM, Tourtellotte WW, Frosch MP, Vonsattel JP, Zarow C, Beach TG, Albin RL, Lieberman AP, Lee VM, Trojanowski JQ, Van Deerlin VM, Bird TD, Galasko DR, Masliah E, White CL, Troncoso JC, Hannequin D, Boxer AL, Geschwind MD, Kumar S, Mandelkow EM, Wszolek ZK, Uitti RJ, Dickson DW, Haines JL, Mayeux R, Pericak-Vance MA, Farrer LA; Alzheimer's Disease Genetics Consortium, Ross OA, Rademakers R, Schellenberg GD, Miller BL, Mandelkow E, Geschwind DH. **Evidence for a role of the rare p.A152T variant in MAPT in increasing the risk for FTD-spectrum and Alzheimer's diseases.** Hum Mol Genet. 2012 Aug 1;21(15):3500-12. doi: 10.1093/hmg/dds161. Epub 2012 May 3. PMID: 22556362. PMCID: PMC3392107.
11. Vardarajan BN, Bruesegem SY, Harbour ME, Inzelberg R, Friedland R, St George-Hyslop P, Seaman MN, Farrer LA. **Identification of Alzheimer disease-associated variants in genes that regulate retromer function.** Neurobiol Aging. 2012 Sep;33(9):2231.e15-2231.e30. doi: 10.1016/j.neurobiolaging.2012.04.020. Epub 2012 Jun 5. PMID: 22673115. PMCID: PMC3391348.
12. Schellenberg GD, Montine TJ. (2012) **The Genetics and Neuropathology of Alzheimer's Disease.** Acta Neuropathol. 2012 Sep;124(3):305-23. doi: 10.1007/s00401-012-0996-2. Epub 2012 May 23. Review. PMID: 22618995, PMCID: PMC3708460.
13. Jun G, Vardarajan BN, Buross J, Yu CE, Hawk MV, Dombroski BA, Crane PK, Larson EB; Alzheimer's Disease Genetics Consortium, Mayeux R, Haines JL, Lunetta KL, Pericak-Vance MA, Schellenberg GD, Farrer LA. **Comprehensive search for Alzheimer disease susceptibility loci in the APOE region.** Arch Neurol. 2012 Oct;69(10):1270-9. PMID: 22869155. PMCID: PMC3579659.
14. Whitcomb DC, LaRusch J, Krasinskas AM, Klei L, Smith JP, Brand RE, Neoptolemos JP, Lerch MM, Tector M, Sandhu BS, Guda NM, Orlichenko L; Alzheimer's Disease Genetics Consortium, Alkaade S, Amann ST, Anderson MA, Baillie J, Banks PA, Conwell D, Coté GA, Cotton PB, DiSario J, Farrer LA,

- Forsmark CE, Johnstone M, Gardner TB, Gelrud A, Greenhalf W, Haines JL, Hartman DJ, Hawes RA, Lawrence C, Lewis M, Mayerle J, Mayeux R, Melhem NM, Money ME, Muniraj T, Papachristou GI, Pericak-Vance MA, Romagnuolo J, Schellenberg GD, Sherman S, Simon P, Singh VP, Slivka A, Stolz D, Sutton R, Weiss FU, Wilcox CM, Zarnescu NO, Wisniewski SR, O'Connell MR, Kienholz ML, Roeder K, Barmada MM, Yadav D, Devlin B. **Common genetic variants in the *CLDN2* and *PRSS1-PRSS2* loci alter risk for alcohol-related and sporadic pancreatitis.** *Nat Genet.* 2012 Dec;44(12):1349-54. doi: 10.1038/ng.2466. Epub 2012 Nov 11. PMID: 23143602. PMCID: PMC3510344.
15. Holton P, Ryten M, Nalls M, Trabzuni D, Weale ME, Hernandez D, Crehan H, Gibbs JR, Mayeux R, Haines JL, Farrer LA, Pericak-Vance MA, Schellenberg GD; Alzheimer's Disease Genetics Consortium, Ramirez-Restrepo M, Engel A, Myers AJ, Corneveaux JJ, Huentelman MJ, Dillman A, Cookson MR, Reiman EM, Singleton A, Hardy J, Guerreiro R. **Initial assessment of the pathogenic mechanisms of the recently identified Alzheimer risk Loci.** *Ann Hum Genet.* Ann Hum Genet. 2013 Mar;77(2):85-105. doi: 10.1111/ahg.12000. Epub 2013 Jan 30. PMID: 23360175. PMCID: PMC3578142.
16. Miyashita A, Koike A, Jun G, Wang LS, Takahashi S, Matsubara E, Kawarabayashi T, Shoji M, Tomita N, Arai H, Asada T, Harigaya Y, Ikeda M, Amari M, Hanyu H, Higuchi S, Ikeuchi T, Nishizawa M, Suga M, Kawase Y, Akatsu H, Kosaka K, Yamamoto T, Imagawa M, Hamaguchi T, Yamada M, Moriaha T, Takeda M, Takao T, Nakata K, Fujisawa Y, Sasaki K, Watanabe K, Nakashima K, Urakami K, Ooya T, Takahashi M, Yuzuriha T, Serikawa K, Yoshimoto S, Nakagawa R, Kim JW, Ki CS, Won HH, Na DL, Seo SW, Mook-Jung I; Alzheimer Disease Genetics Consortium, St George-Hyslop P, Mayeux R, Haines JL, Pericak-Vance MA, Yoshida M, Nishida N, Tokunaga K, Yamamoto K, Tsuji S, Kanazawa I, Ihara Y, Schellenberg GD, Farrer LA, Kuwano R. ***SORL1* is genetically associated with late-onset Alzheimer's disease in Japanese, Koreans and Caucasians.** *PLoS One.* 2013 Apr 2;8(4):e58618. doi: 10.1371/journal.pone.0058618. Epub 2013 Apr 2. Erratum in: *PLoS One.* 2013;8(7). doi:10.1371/annotation/fcb56ea7-d32a-4e45-818d-39cef330c731. PMID: 23565137. PMCID: PMC3614978.
17. Reitz C, Jun G, Naj A, Rajbhandary R, Vardarajan BN, Wang LS, Valladares O, Lin CF, Larson EB, Graff-Radford NR, Evans D, De Jager PL, Crane PK, Buxbaum JD, Murrell JR, Raj T, Ertekin-Taner N, Logue M, Baldwin CT, Green RC, Barnes LL, Cantwell LB, Fallin MD, Go RC, Griffith P, Obisesan TO, Manly JJ, Lunetta KL, Kamboh MI, Lopez OL, Bennett DA, Hendrie H, Hall KS, Goate AM, Byrd GS, Kukull WA, Foroud TM, Haines JL, Farrer LA, Pericak-Vance MA, Schellenberg GD, Mayeux R; Alzheimer Disease Genetics Consortium. **Variants in the *ATP-binding cassette transporter (ABCA7)*, *apolipoprotein E ε4*, and the risk of late-onset Alzheimer disease in African Americans.** *JAMA.* 2013 Apr 10;309(14):1483-92. doi: 10.1001/jama.2013.2973. PMID: 23571587. PMCID: PMC3667653.
18. Cruchaga C, Kauwe JS, Harari O, Jin SC, Cai Y, Karch CM, Benitez BA, Jeng AT, Skorupa T, Carrell D, Bertelsen S, Bailey M, McKean D, Shulman JM, De Jager PL, Chibnik L, Bennett DA, Arnold SE, Harold D, Sims R, Gerrish A, Williams J, Van Deerlin VM, Lee VM, Shaw LM, Trojanowski JQ, Haines JL, Mayeux R, Pericak-Vance MA, Farrer LA, Schellenberg GD, Peskind ER, Galasko D, Fagan AM, Holtzman DM, Morris JC, GERAD Consortium, Alzheimer's Disease Neuroimaging Initiative (ADNI), Alzheimer Disease Genetic Consortium (ADGC), and Goate AM. **GWAS of cerebrospinal fluid tau levels identifies risk variants for Alzheimer's disease.** *Neuron.* 2013 Apr 24;78(2):256-68. doi: 10.1016/j.neuron.2013.02.026. Epub 2013 Apr 4. PMID: 23562540. PMCID: PMC3664945.
19. Reitz C, Tosto G, Vardarajan B, Rogaeva E, Ghani M, Rogers RS, Conrad C, Haines JL, Pericak-Vance MA, Fallin MD, Foroud T, Farrer LA, Schellenberg GD, George-Hyslop PS, Mayeux R. **Independent and epistatic effects of variants in *VPS10-d* receptors on Alzheimer disease risk and processing of the amyloid precursor protein (APP).** *Transl Psychiatry.* 2013 May 14;3:e256. doi: 10.1038/tp.2013.13. PMID: 23673467. PMCID: PMC3669917.

20. Kohli MA1, John-Williams K, Rajbhandary R, Naj A, Whitehead P, Hamilton K, Carney RM, Wright C, Crocco E, Gwirtzman HE, Lang R, Beecham G, Martin ER, Gilbert J, Benatar M, Small GW, Mash D, Byrd G, Haines JL, Pericak-Vance MA, Züchner S. **Repeat expansions in the C9ORF72 gene contribute to Alzheimer's disease in Caucasians.** *Neurobiol Aging*. 2013 May;34(5):1519.e5-12. doi: 10.1016/j.neurobiolaging.2012.10.003. Epub 2012 Oct 27. PMID: 23107433. PMCID: PMC3586789.
21. Hibar DP, Stein JL, Ryles AB, Kohannim O, Jahanshad N, Medland SE, Hansell NK, McMahon KL, de Zubicaray GI, Montgomery GW, Martin NG, Wright MJ, Saykin AJ, Jack CR Jr, Weiner MW, Toga AW, Thompson PM; Alzheimer's Disease Neuroimaging Initiative. **Genome-wide association identifies genetic variants associated with lentiform nucleus volume in N = 1345 young and elderly subjects.** *Brain Imaging Behav*. 2013 Jun;7(2):102-15. doi: 10.1007/s11682-012-9199-7. PMID: 22903471. PMCID: PMC3779070.
22. Leung YY, Ryvkin P, Ungar LH, Gregory BD, Wang LS. **CoRAL: predicting non-coding RNAs from small RNA-sequencing data.** *Nucleic Acids Res*. 2013 Aug;41(14):e137. doi: 10.1093/nar/gkt426. Epub 2013 May 21. PMID: 23700308. PMCID: PMC3737537.
23. Nuytemans K, Bademci G, Kohli MM, Beecham GW, Wang L, Young JI, Nahab F, Martin ER, Gilbert JR, Benatar M, Haines JL, Scott WK, Züchner S, Pericak-Vance MA, Vance JM. **C9ORF72 intermediate repeat copies are a significant risk factor for Parkinson disease.** *Ann Hum Genet*. 2013 Sep;77(5):351-63. doi: 10.1111/ahg.12033. Epub 2013 Jul 12. PMID: 23845100. PMCID: PMC3815478.
24. Lin C, Valladares O, Childress DM, Klevak E, Geller ET, Hwang YC, Tsai EA, Schelleberg GD, Wang LS. **DRAW+SneakPeek: Analysis workflow and quality metric management for DNA-seq experiments.** *Bioinformatics*. 2013 Oct 1;29(19):2498-500. doi:10.1093 Epub 2013 Aug 13. PMCID: PMC3777113.
25. Cao K, Ryvkin P, Hwang YC, Johnson FB, Wang LS. **Analysis of nonlinear gene expression progression reveals extensive pathway and age-specific transitions in aging human brains.** *PLoS One*. 2013 Oct 3;8(10):e74578. doi: 10.1371/journal.pone.0074578. eCollection 2013. PMID: 24098339. PMCID: PMC3789733.
26. Reitz C, Mayeux R, Alzheimer's Disease Genetics Consortium (2013), **TREM2 and Neurodegenerative Disease.** *N Engl J Med*. 2013 Oct 17;369(16):1564-5. doi: 10.1056/NEJMc1306509. PMID: 24131184. PMCID: PMC3980568.
27. Lin CF, Naj AC, Wang LS. **Analyzing copy number variation using SNP array data: protocols for calling CNV and association tests.** *Curr Protoc Hum Genet*. 2013 Oct 18;79:Unit 1.27.. doi: 10.1002/0471142905.hg0127s79. PMID: 24510649. PMCID: PMC4015338.
28. Ridge PG, Mukherjee S, Crane PK, Kauwe JSK, Alzheimer's Disease Genetics Consortium. **Alzheimer's disease: analyzing the missing heritability.** *PLoS One*. 2013 Nov 7;8(11):e79771. doi: 10.1371/journal.pone.0079771. eCollection 2013. PMID:D 24244562. PMCID: PMC3820606
29. Barral S, Cosentino S, Costa R, Andersen SL, Christensen K, Eckfeldt JH, Newman AB, Perls TT, Province MA, Hadley EC, Rossi WK, Mayeux R; Long Life Family Study. **Exceptional memory performance in the Long Life Family Study.** *Neurobiol Aging*. 2013 Nov;34(11):2445-8. doi: 10.1016/j.neurobiolaging.2013.05.002. Epub 2013 Jun 4. PMID: 23759147. PMCID: PMC4095822.
30. Lambert JC, Ibrahim-Verbaas CA, Harold D, Naj AC, Sims R, Bellenguez C, DeStafano AL, Bis JC, Beecham GW, Grenier-Boley B, Russo G, Thornton-Wells TA, Jones N, Smith AV, Chouraki V, Thomas C, Ikram MA, Zelenika D, Vardarajan BN, Kamatani Y, Lin CF, Gerrish A, Schmidt H, Kunkle B, Dunstan ML,



- Ruiz A, Bihoreau MT, Choi SH, Reitz C, Pasquier F, Cruchaga C, Craig D, Amin N, Berr C, Lopez OL, De Jager PL, Deramecourt V, Johnston JA, Evans D, Lovestone S, Letenneur L, Morón FJ, Rubinsztein DC, Eiriksdottir G, Sleegers K, Goate AM, Fiévet N, Huentelman MW, Gill M, Brown K, Kamboh MI, Keller L, Barberger-Gateau P, McGuinness B, Larson EB, Green R, Myers AJ, Dufouil C, Todd S, Wallon D, Love S, Rogava E, Gallacher J, St George-Hyslop P, Clarimon J, Lleo A, Bayer A, Tsuang DW, Yu L, Tsolaki M, Bossù P, Spalletta G, Proitsi P, Collinge J, Sorbi S, Sanchez-Garcia F, Fox NC, Hardy J, Deniz Naranjo MC, Bosco P, Clarke R, Brayne C, Galimberti D, Mancuso M, Matthews F; European Alzheimer's Disease Initiative (EADI); Genetic and Environmental Risk in Alzheimer's Disease; Alzheimer's Disease Genetic Consortium; Cohorts for Heart and Aging Research in Genomic Epidemiology, Moebus S, Mecocci P, Del Zompo M, Maier W, Hampel H, Pilotto A, Bullido M, Panza F, Caffarra P, Nacmias B, Gilbert JR, Mayhaus M, Lannefelt L, Hakonarson H, Pichler S, Carrasquillo MM, Ingelsson M, Beekly D, Alvarez V, Zou F, Valladares O, Younkin SG, Coto E, Hamilton-Nelson KL, Gu W, Razquin C, Pastor P, Mateo I, Owen MJ, Faber KM, Jonsson PV, Combarros O, O'Donovan MC, Cantwell LB, Soinen H, Blacker D, Mead S, Mosley TH Jr, Bennett DA, Harris TB, Fratiglioni L, Holmes C, de Bruijn RF, Passmore P, Montine TJ, Bettens K, Rotter JI, Brice A, Morgan K, Foroud TM, Kukull WA, Hannequin D, Powell JF, Nalls MA, Ritchie K, Lunetta KL, Kauwe JS, Boerwinkle E, Riemenschneider M, Boada M, Hiltunen M, Martin ER, Schmidt R, Rujescu D, Wang LS, Dartigues JF, Mayeux R, Tzourio C, Hofman A, Nöthen MM, Graff C, Psaty BM, Jones L, Haines JL, Holmans PA, Lathrop M, Pericak-Vance MA, Launer LJ, Farrer LA, van Duijn CM, Van Broeckhoven C, Moskvina V, Seshadri S, Williams J, Schellenberg GD, Amouyel P. **Extended meta-analysis of 74,046 individuals identifies 11 new susceptibility loci for Alzheimer's disease.** *Nat. Genet.* 2013 Dec;45(12):1452-8. Doi: 10.1038/ng.2802. Epub 2013 Oct 27. PMID: 24162737. PMCID: PMC3896259.
31. Sherva R, Tripodis Y, Bennett DA, Chibnik LB, Crane PK, de Jager PL, Farrer LA, Saykin AJ, Shulman JM, Naj A, Green RC, The GENAROAD Consortium, The Alzheimer's Disease Neuroimaging Initiative, and The Alzheimer's Disease Genetics Consortium. **Genome-wide association study of the rate of cognitive decline in Alzheimer's disease.** *Alzheimers Dement.* 2014 Jan;10(1):45-52. doi: 10.1016/j.jalz.2013.01.008. Epub 2013 Mar 25. PMID: 23535033. PMCID: PMC3760995.
32. Ruiz A, Heilmann S, Becker T, Hernández I, Wagner H, Thelen M, Mauleón A, Rosende-Roca M, Bellenguez C, Bis JC, Harold D, Gerrish A, Sims R, Sotolongo-Grau O, Espinosa A, Alegret M, Arrieta JL, Lacour A, Leber M, Becker J, Lafuente A, Ruiz S, Vargas L, Rodríguez O, Ortega G, Dominguez MA; IGAP, Mayeux R, Haines JL, Pericak-Vance MA, Farrer LA, Schellenberg GD, Chouraki V, Launer LJ, van Duijn C, Seshadri S, Antúnez C, Breteler MM, Serrano-Ríos M, Jessen F, Tárrega L, Nöthen MM, Maier W, Boada M, Ramírez A. (2014) **Follow-up of loci from the International Genomics of Alzheimer's Disease Project identifies TRIP4 as a novel susceptibility gene.** *Transl Psychiatry.* 2014 Feb 4;4:e358. doi: 10.1038/tp.2014.2. PMID: 24495969. PMCID: PMC3944635.
33. Ryvkin P, Leung YY, Ungar LH, Gregory BD, Wang LS. **Using machine learning and high-throughput RNA sequencing to classify the precursors of small non-coding RNAs.** *Methods.* 2014 May 1;67(1):28-35. doi: 10.1016/j.ymeth.2013.10.002. Epub 2013 Oct 18. PMID: 24145223. PMCID: PMC3991776.
34. Carney RM, Kohli MA, Kunkle BW, Naj AC, Gilbert JR, Züchner S, Pericak-Vance MA. **Parkinsonism and distinct dementia patterns in a family with the MAPT R406W mutation.** *Alzheimers Dement.* 2014 May;10(3):360-5. doi: 10.1016/j.jalz.2013.02.011. Epub 2013 May 30. PMID: 23727082. PMCID: PMC3762928.
35. Escott-Price V, Bellenguez C, Wang LS, Choi SH, Harold D, Jones L, Holmans P, Gerrish A, Vedernikov A, Richards A, DeStefano AL, Lambert JC, Ibrahim-Verbaas CA, Naj AC, Sims R, Jun G, Bis JC, Beecham GW, Grenier-Boley B, Russo G, Thornton-Wells TA, Denning N, Smith AV, Chouraki V, Thomas

- C, Ikram MA, Zelenika D, Vardarajan BN, Kamatani Y, Lin CF, Schmidt H, Kunkle B, Dunstan ML, Vronskaya M; United Kingdom Brain Expression Consortium, Johnson AD, Ruiz A, Bihoreau MT, Reitz C, Pasquier F, Hollingworth P, Hanon O, Fitzpatrick AL, Buxbaum JD, Campion D, Crane PK, Baldwin C, Becker T, Gudnason V, Cruchaga C, Craig D, Amin N, Berr C, Lopez OL, De Jager PL, Deramecourt V, Johnston JA, Evans D, Lovestone S, Letenneur L, Hernández I, Rubinsztein DC, Eiriksdottir G, Sleegers K, Goate AM, Fiévet N, Huentelman MJ, Gill M, Brown K, Kamboh MI, Keller L, Barberger-Gateau P, McGuinness B, Larson EB, Myers AJ, Dufouil C, Todd S, Wallon D, Love S, Rogaeva E, Gallacher J, George-Hyslop PS, Clarimon J, Lleo A, Bayer A, Tsuang DW, Yu L, Tsolaki M, Bossù P, Spalletta G, Proitsi P, Collinge J, Sorbi S, Garcia FS, Fox NC, Hardy J, Naranjo MC, Bosco P, Clarke R, Brayne C, Galimberti D, Scarpini E, Bonuccelli U, Mancuso M, Siciliano G, Moebus S, Mecocci P, Zompo MD, Maier W, Hampel H, Pilotto A, Frank-García A, Panza F, Solfrizzi V, Caffarra P, Nacmias B, Perry W, Mayhaus M, Lannfelt L, Hakonarson H, Pichler S, Carrasquillo MM, Ingelsson M, Beekly D, Alvarez V, Zou F, Valladares O, Younkin SG, Coto E, Hamilton-Nelson KL, Gu W, Razquin C, Pastor P, Mateo I, Owen MJ, Faber KM, Jonsson PV, Combarros O, O'Donovan MC, Cantwell LB, Soyninen H, Blacker D, Mead S, Mosley TH Jr, Bennett DA, Harris TB, Fratiglioni L, Holmes C, de Bruijn RF, Passmore P, Montine TJ, Bettens K, Rotter JI, Brice A, Morgan K, Foroud TM, Kukull WA, Hannequin D, Powell JF, Nalls MA, Ritchie K, Lunetta KL, Kauwe JS, Boerwinkle E, Riemenschneider M, Boada M, Hiltunen M, Martin ER, Schmidt R, Rujescu D, Dartigues JF, Mayeux R, Tzourio C, Hofman A, Nöthen MM, Graff C, Psaty BM, Haines JL, Lathrop M, Pericak-Vance MA, Launer LJ, Van Broeckhoven C, Farrer LA, van Duijn CM, Ramirez A, Seshadri S, Schellenberg GD, Amouyel P, Williams J; Cardiovascular Health Study (CHS). **Gene-wide analysis detects two new susceptibility genes for Alzheimer's disease.** *PLoS One.* 2014 Jun 12;9(6):e94661. doi: 10.1371/journal.pone.0094661. eCollection 2014. PMID: 24922517. PMCID: PMC4055488.
36. Logue MW, Schu M, Vardarajan BN, Farrell J, Lunetta KL, Jun G, Baldwin CT, Deangelis MM, Farrer LA. **Search for age-related macular degeneration risk variants in Alzheimer disease genes and pathways.** *Neurobiol Aging.* 2014 Jun;35(6):1510.e7-18. doi: 10.1016/j.neurobiolaging.2013.12.007. Epub 2013 Dec 19. PMID: 24439028. PMCID: PMC3961547.
37. Benitez BA, Jin SC, Guerreiro R, Graham R, Lord J, Harold D, Sims R, Lambert J-C, Gibbs JR, Bras J, Sassi C, Harari O, Bertelsen S, Lupton MK, Powell J, Bellenguez C, Brown K, Medway C, Haddick PCG, van der Brug MP, Bhangale T, Ortmann W, Behrens T, Mayeux R, Pericak-Vance MA, Farrer LA, Schellenberg GD, Haines JL, Turton J, Braae A, Barber I, Fagan AM, Holtzman DM, Morris JC, The 3C Study Group, the EADI consortium, the Alzheimer Disease Genetic Consortium (ADGC), Alzheimer's Disease Neuroimaging Initiative (ADNI), the GERAD Consortium, Williams J, Kauwe J, Amouyel P, Morgan K, Singleton A, Hardy J, Goate AM, Cruchaga C, (2014) **Missense variant in *TREML2* protects against Alzheimer's Disease.** *Neurobiol Aging.* 2014 Jun;35(6):1510.e19-26. doi: 10.1016/j.neurobiolaging.2013.12.010. Epub 2013 Dec 21. PMID: 24439484. PMCID: PMC3961557.
38. Nelson PT1, Estus S, Abner EL, Parikh I, Malik M, Neltner JH, Ighodaro E, Wang WX, Wilfred BR, Wang LS, Kukull WA, Nandakumar K, Farman ML, Poon WW, Corrada MM, Kawas CH, Cribbs DH, Bennett DA, Schneider JA, Larson EB, Crane PK, Valladares O, Schmitt FA, Kryscio RJ, Jicha GA, Smith CD, Scheff SW, Sonnen JA, Haines JL, Pericak-Vance MA, Mayeux R, Farrer LA, Van Eldik LJ, Horbinski C, Green RC, Gearing M, Poon LW, Kramer PL, Woltjer RL, Montine TJ, Partch AB, Rajic AJ, Richmire K, Monsell SE; Alzheimer' Disease Genetic Consortium, Schellenberg GD, Fardo DW. **ABCC9 gene polymorphism is associated with hippocampal sclerosis of aging pathology.** *Acta Neuropathol.* 2014 June;127(6):825-43. doi: 10.1007/s00401-014-1282-2. Epub 2014 Apr 27. PMID: 24770881. PMCID: PMC4113197.
39. Allen M, Kachadoorian M, Quicksall Z, Zou F, Chai HS, Younkin C, Crook J, Pankratz S, Carrasquillo MM, Siddharth Krishnan S, Nguyen T, Ma L, Malphrus K, Lincoln S, Bisceglia G, Kolbert CP, Jen J, Mukherjee

- S, Kauwe JK, Crane P, Schellenberg GD, Alzheimer's Disease Genetics Consortium, Petersen RK, Graff-Radford NR, Dickson DW, Younkin SG, Ertekin-Taner N. (2014) **Association of *MAPT* haplotypes with Alzheimer's disease risk and *MAPT* brain gene expression levels.** *Alzheimers Res Ther.* 2014 Jul 1;6(4):39. doi: 10.1186/alzrt268. eCollection 2014. PMID: 25324900. PMCID: PMC4198935.
40. Beecham GW, Hamilton K, Naj AC, Martin ER, Huentelman M, Myers AJ, Corneveaux JJ, Hardy J, Vonsattel JP, Younkin SG, Bennett DA, De Jager PL, Larson EB, Crane PK, Kamboh MI, Kofler JK, Mash DC, Duque L, Gilbert JR, Gwirtsman H, Buxbaum JD, Kramer P, Dickson DW, Farrer LA, Frosch MP, Ghetti B, Haines JL, Hyman BT, Kukull WA, Mayeux RP, Pericak-Vance MA, Schneider JA, Trojanowski JQ, Reiman EM; Alzheimer's Disease Genetics Consortium (ADGC), Schellenberg GD, Montine TJ. **Genome-wide association meta-analysis of neuropathologic features of Alzheimer's disease and related dementias.** *PLoS Genet.* 2014 Sep 4;10(9):e1004606. doi: 10.1371/journal.pgen.1004606. eCollection 2014 Sep. Erratum in: *PLoS Genet.* 2014 Nov;10(11):e1004867. PMID: 25188341. PMCID: PMC4154667
41. Jun G, Asai H, Zeldich E, Drapeau E, Chung J, Park J-H, Chen CD, Kim S, Haroutunian V, Foroud T, Kuwano R, Haines JL, Pericak-Vance MA, Schellenberg GD, Lunetta KL, Kim J-W, Buxbaum JD, Mayeux R, Ikezu T, Abraham CR, Farrer LA. (2014) ***PLXNA4* is Associated With Alzheimer Disease Risk and Modulates Tau Protein Phosphorylation.** *Ann Neurol.* 2014 Sep;76(3):379-92. doi: 10.1002/ana.24219. Epub 2014 Jul 29. PMID: 25043464. PMCID: PMC4830273.
42. Kauwe JS, Bailey MH, Ridge PG, Perry R, Wadsworth ME, Hoyt KL, Staley LA, Karch CM, Harari O, Cruchaga C, Ainscough BJ, Bales K, Pickering EH, Bertelsen S; Alzheimer's Disease Neuroimaging Initiative, Fagan AM, Holtzman DM, Morris JC, Goate AM. **Genome-wide association study of CSF levels of 59 Alzheimer's disease candidate proteins: significant associations with proteins involved in amyloid processing and inflammation.** *PLoS Genet.* 2014 Oct 23;10(10):e1004758. doi: 10.1371/journal.pgen.1004758. eCollection 2014 Oct. PMID: 25340798. PMCID: PMC4207667.
43. Naj AC, Jun G, Reitz C, Kunkle BW, Perry WR, Park YS, Beecham GW, Rajbhandary R, Hamilton-Nelson KL, Wang L-S, Kauwe JSK, Huentelman MJ, Myers AJ, Bird TD, Boeve BF, Baldwin CT, Jarvik GP, Crane PK, Rogaeva E, Barmada MM, Demirci FY, Cruchaga C, Nowotny P, Kramer PL, Alzheimer's Disease Genetics Consortium, Ertekin-Taner N, Hardy J, Graff-Radford NR, Green RC, Larson EB, St. George-Hyslop P, Buxbaum JD, Evans D, Schneider JA, Lunetta KL, Kamboh MI, Saykin AJ, Reiman EM, De Jager PL, Bennett DA, Morris JC, Montine TJ, Goate AM, Blacker D, Tsuang DW, Hakonarson H, Kukull WA, Foroud TM, Martin ER, Haines JL, Mayeux R, Farrer LA, Schellenberg GD, Pericak-Vance MA. **Effects of Multiple Genetic Loci on Age at Onset in Late-Onset Alzheimer Disease: A Genome-Wide Association Study.** *JAMA Neurol.* 2014 Nov;71(11):1394-404. doi: 10.1001/jamaneurol.2014.1491. Erratum in: *JAMA Neurol.* 2014 Nov;71(11):1457. PMID: 25199842. PMCID: PMC4314944.
44. Logue MW, Schu M, Vardarajan BN, Farrell J, Bennett DA, Buxbaum JD, Byrd GS, Ertekin-Taner N, Evans D, Foroud T, Goate A, Graff-Radford NR, Kamboh MI, Kukull WA, Manly JJ, Alzheimer Disease Genetics Consortium, Haines JL, Mayeux R, Pericak-Vance MA, Schellenberg GD, Lunetta KL, Baldwin CT, Fallin MD, and Farrer LA. **Two Rare *AKAP9* Missense Variants Are Associated with Alzheimer Disease in African Americans.** *Alzheimers Dement.* 2014 Nov;10(6):609-618.e11. doi: 10.1016/j.jalz.2014.06.010. Epub 2014 Aug 27. PMID: 25172201. PMCID: PMC4253055.
45. Ramirez A, van der Flier WM, Herold C, Ramonet D, Heilmann S, Lewczuk P, Popp J, Lacour A, Drichel D, Louwersheimer E, Kummer MP, Cruchaga C, Hoffmann P, Teunissen C, Holstege H, Kornhuber J, Peters O, Naj AC, Chouraki V, Bellenguez C, Gerrish A; International Genomics of Alzheimer's Project (IGAP); Alzheimer's Disease Neuroimaging Initiative (ADNI), Heun R, Frölich L, Hüll M, Buscemi L, Herms S, Kölsch H, Scheltens P, Breteler MM, Rübter E, Wiltfang J, Goate A, Jessen F, Maier W, Heneka MT,

- Becker T, Nöthen MM. **SUCLG2 identified as both a determinant of CSF A $\beta$ 1-42-levels and an attenuator of cognitive decline in Alzheimer's disease.** Hum Mol Genet. 2014 Dec 15;23(24):6644-58. doi: 10.1093/hmg/ddu372. Epub 2014 Jul 15. PMID: 25027320. PMCID: PMC4240204.
46. Barral S, Reitz C, Small SA, Mayeux R. **Genetic variants in a 'cAMP element binding protein' (CREB)-dependent histone acetylation pathway influence memory performance in cognitively healthy elderly individuals.** Neurobiol Aging. 2014 Dec;35(12):2881.e7-2881.e10. doi: 10.1016/j.neurobiolaging.2014.06.024. Epub 2014 Jun 28. PMID: 25150575. PMCID: PMC4253058.
47. Barral, S.; Cosentino, S.; Christensen, K3.; Newman, A.N.; Perls, T.T.; Province, M.A.; Hadley, E.C.; Pericak-Vance MA, Haines JL, Farrer L, Schellenberg GD, Mayeux R for the Alzheimer' Disease Genetic Consortium, Mayeux, R. for the Long Life Family Study (LLFS). **Common genetic variants on 6q24 associated with exceptional episodic memory performance in the elderly.** JAMA Neurol. 2014 Dec;71(12):1514-9. doi: 10.1001/jamaneurol.2014.1663. PMID:25317765. PMCID: PMC4355405.
48. Wetzel-Smith MK, Hunkapiller J, Bhangale TR, Maloney JA, Atwal JK, Ortmann W, Rathore N, Tessier-Lavigne M, Alzheimer's Disease Genetics Consortium (ADGC), Mayeux R, Pericak-Vance MA, Haines J, Farrer L, Schellenberg GD, Goate A, Behrens TW, Cruchaga C, Watts RJ, Graham RR. **A rare mutation in UNC5C predisposes to late-onset Alzheimer's disease and increases neuronal cell death.** Nat Med. 2014 Dec;20(12):1452-7. doi: 10.1038/nm.3736. Epub 2014 Nov 24. PMID: 25419706. PMCID: PMC4301587.
49. Schreiber M, Bird TD, Tsuang DW. **Alzheimer's Disease Genetics.** Curr Behav Neurosci Rep 1: 191. December 2014, Volume 1, Issue 4, pp 191–196. <https://doi.org/10.1007/s40473-014-0026-x>.
50. Nelson PT1, Wang WX, Partch AB, Monsell SE, Valladares O, Ellingson SR, Wilfred BR, Naj AC, Wang LS, Kukull WA, Fardo DW. **Reassessment of risk genotypes (GRN, TMEM106B, and ABCC9 variants) associated with hippocampal sclerosis of aging pathology.** J Neuropathol Exp Neurol. 2015 Jan;74(1):75-84. doi: 10.1097/NEN.000000000000151. PMID: 25470345. PMCID: PMC4270894.
51. Wang L-S, Naj AC, Graham RR, Crane PK, Kunkle BW, Cruchaga C, Murcia JDG, Cannon-Albright L, Baldwin CT, Zetterberg H, Blennow K, Kukull WA, Faber KM, Schupf N, Norton MC, Tschanz JT, Munger RG, Corcoran CD, Rogaeva E, Alzheimer's Disease Genetics Consortium, NIA-LOAD Family Study, Lin C-F, Dombroski BA, Cantwell LB, Partch A, Valladares O, Hakonarson H, St George-Hyslop P, Green RC, Goate AM, Foroud TM, Carney RM, Larson EB, Behrens TW, Kauwe JSK, Haines JL, Farrer LA, Pericak-Vance MA, Mayeux R, Schellenberg GD. **Rarity of the Alzheimer disease-protective APP A673T variant in the United States.** JAMA Neurol. 2015 Feb;72(2):209-16. doi: 10.1001/jamaneurol.2014.2157. PMID: 25531812. PMCID: PMC4324097.
52. Beecham GW, Dickson DW, Scott WK, Martin ER, Schellenberg GD on behalf of the Alzheimer Disease Genetics Consortium, Nuytemans K, Larson EB, Buxbaum JD, Trojanowski JQ, Van Deerlin VM, Hurtig HI, Mash DC, Beach TG, Troncoso JC, Pletnikova O, Frosch MP, Ghetti B, Foroud TM, Honig LS, Marder K, Vonsattel JP, Goldman SM, Vinters HV, Ross OA, Wszolek ZK, Wang L, Dykxhoorn DM, Pericak-Vance MA, Montine TJ, Leverenz JB, Dawson TM, Vance JM. **PARK10 is a major locus for sporadic, neuropathologically-confirmed Parkinson disease.** Neurology. 2015 Mar 10;84(10):972-80. doi: 10.1212/WNL.0000000000001332. Epub 2015 Feb 6. PMID: 25663231. PMCID: PMC4352096.
53. Desikan RS, Schork AJ, Wang Y, Thompson WK, Dehghan A, Ridker PM, Chasman DI, McEvoy LK, Holland D, Chen CH, Karow DS, Brewer JB, Hess CP, Williams J, Sims R, O'Donovan MC, Choi SH, Bis JC, Ikram MA, Gudnason V, DeStefano AL, van der Lee SJ, Psaty BM, van Duijn CM, Launer L, Seshadri S, Pericak-Vance MA, Mayeux R, Haines JL, Farrer LA, Hardy J, Ulstein ID, Aarsland D, Fladby T, White

- LR, Sando SB, Rongve A, Witoelar A, Djurovic S, Hyman BT, Snaedal J, Steinberg S, Stefansson H, Stefansson K, Schellenberg GD, Andreassen OA, Dale AM; Inflammation Working Group and International Genomics of Alzheimer's Disease Project (IGAP) and DemGene Investigators. **Polygenic overlap between C-Reactive protein, plasma lipids and Alzheimer's disease.** *Circulation*. 2015 Jun 9;131(23):2061-2069. doi: 10.1161/CIRCULATIONAHA.115.015489. Epub 2015 Apr 10. PMID: 25862742. PMCID: PMC4677995.
54. Malik M, Chiles J 3rd, Xi HS, Medway C, Simpson J, Potluri S, Howard D, Liang Y, Paumi CM, Mukherjee S, Crane P, Younkin S, Fardo DW, Estus S. **Genetics of CD33 Splicing in Human Brain and Acute Myeloid Leukemia.** *Hum Mol Genet*. 2015 Jun 15;24(12):3557-70. doi: 10.1093/hmg/ddv092. Epub 2015 Mar 11. PMID: 25762156. PMCID: PMC4498153.
55. Østergaard SD, Mukherjee S, Sharp SJ, Proitsi P, Lotta LA, Day F, Boehme KL, Walter S, Kauwe JSK, Gibbons LE, Alzheimer's Disease Genetics Consortium, The Genetic and Environmental Risk in AD Consortium, The EPIC-InterAct consortium, Larson EB, Glymour MM, Powell JF, Langenberg C, Crane PK, Wareham NJ, Scott RA. **Associations between potentially modifiable risk factors and Alzheimer's disease: A Mendelian randomization study.** *PLoS Med*. 2015 Jun 16;12(6):e1001841; discussion e1001841. doi: 10.1371/journal.pmed.1001841. eCollection 2015 Jun. PMID: 26079503. PMCID: PMC4469461.
56. Jones L, Bellenguez C, Wang L-S, Choi S-H, Harold D, Vedernikov A, Escott-Price V, Stone T, Richards A, Lambert JC, Ibrahim-Verbaas CA, Naj AC, Sims R, Gerrish A, Jun G, DeStefano AL, Bis JC, Beecham GW, Grenier-Boley B, Russo G, Thornton-Wells TA, Jones N, Smith AV, Chouraki V, Thomas C, Ikram MA, Zelenika D, Vardarajan BN, Kamatani Y, Lin C-F, Schmidt H, Kunkle B, Dunstan ML, Ruiz A, Bihoreau M-T, Reitz C, Pasquier F, Hollingworth P, Hanon O, Fitzpatrick AI, Buxbaum JD, Campion D, Crane PK, Becker T, Gudnason V, Cruchaga C, Craig D, Amin N, Berr C, Lopez OL, de Jager PL, Deramecourt V, Johnston JA, Evans D, Lovestone S, Letteneur L, Morón FJ, Rubinsztein DC, Eiriksdottir G, Sleegers K, Goate AM, Fiévet N, Huentelman MJ, Gill M, Brown K, Kamboh MI, Keller L, Barberger-Gateau P, McGuinness B, Larson EB, Myers AJ, Dufouil C, Todd S, Wallon D, Love S, Rogaeva E, Gallacher J, St George-Hyslop P, Clarimon J, Lleó A, Bayer A, Tsuang DW, Yu L, Tsolaki M, Bossù P, Spalletta G, Proitsi P, Collinge J, Sorbi S, Garcia FS, Fox N, Hardy J, Naranjo MCD, Razquin C, Bosco P, Clarke R, Brayne C, Galimberti D, Mancuso M, MRC CFAS, Moebus S, Mecocci P, del Zompo M, Maier W, Hampel H, Pilotto A, Bullido M, Panza F, Caffarra P, Nacmias B, Gilbert JR, Mayhaus M, Jessen F, Dichgans M, Lannfelt L, Hakonarson H, Pichler S, Carrasquillo MM, Ingelsson M, Beekly D, Alavarez V, Zou F, Valladares O, Younkin SG, Coto E, Hamilton-Nelson KL, Mateo I, Owen MJ, Faber KM, Jonsson PV, Combarros O, O'Donovan MC, Cantwell LB, Soininen H, Blacker D, Mead S, Mosley TH, Bennett DA, Harris TB, Fratiglioni L, Holmes C, de Bruijn RFAG, Passmore P, Montine TJ, Bettens K, Rotter JI, Brice A, Morgan K, Foroud TM, Kukull WA, Hannequin D, Powell JF, Nalls MA, Ritchie K, Lunetta KL, Kauwe JSK, Boerwinkle E, Riemenschneider M, Boada M, Hiltunen M, Martin ER, Pastor P, Schmidt R, Rujescu D, Dartigues J-F, Mayeux R, Tzourio C, Hofman A, Nöthen MM, Graff C, Psaty BM, Haines JL, Lathrop M, Pericak-Vance MA, Launer LJ, Farrer LA, van Duijn CM, Van Broekhoven C, Ramirez A, Schellenberg GD, Seshadri S, Amouyel P, Williams J, Holmans PA. **Convergent genetic and expression data implicate immunity in Alzheimer's disease.** *Alzheimers Dement*. 2015 Jun;11(6):658-71. doi: 10.1016/j.jalz.2014.05.1757. Epub 2014 Dec 20. PMID: 25533204. PMCID: PMC4672734.
57. Ghani M, Reitz C, Cheng R, Vardarajan BN, Jun G, Sato C, Naj A, Rajbhandary R, Wang L-S, Valladares O, Lin C-F, Larson EB, Graff-Radford NR, Evans D, De Jager PL, Crane PK, Buxbaum JD, Murrell JR, Raj T, Ertekin-Taner N, Logue M, Baldwin CT, Green RC, Barnes LL, Cantwell LB, Fallin MD, Go RCP, Griffith P, Obisesan TO, Manly JJ, Lunetta KL, Kamboh MI, Lopez OL, Bennett DA, Hendrie H, Hall KS, Goate AM, Byrd GS, Kukull WA, Foroud TM, Haines JL, Farrer LA, Pericak-Vance MA, Lee JH, Schellenberg GD, St George-Hyslop P, Mayeux R, Rogaeva E & the Alzheimer Disease Genetics

- Consortium. **Association of Long Runs of Homozygosity with Alzheimer's Disease among African American Individuals.** *JAMA Neurol.* 2015 Nov;72(11):1313-23. doi: 10.1001/jamaneurol.2015.1700. PMID: 26366463. PMCID: PMC4641052.
58. Mukherjee S, Walter S, Kauwe JSK, Saykin AJ, Bennett DA, Larson EB, Crane PK, Glymour MM, for the Adult Changes in Thought, Religious Orders Study / Memory and Aging Project, and Alzheimer's Disease Genetics Consortium. **Genetically predicted body mass index and Alzheimer's disease-related phenotypes in three large samples: Mendelian randomization analyses.** *Alzheimers Dement.* 2015 Dec;11(12):1439-1451. doi: 10.1016/j.jalz.2015.05.015. Epub 2015 Jun 12. PMID: 26079416. PMCID: PMC4676945.
59. Desikan RS, Schork AJ, Wang Y, Witoelar A, Sharma M, McEvoy LK, Holland D, Brewer JB, Chen C-H, Thompson WK, Harold D, Williams J, Owen MJ, O'Donovan MC, Pericak-Vance MA, Mayeux R, Haines JL, Farrer LA, Schellenberg GD, Heutink P, Singleton AB, Brice A, Wood NW, Hardy J, Martinez M, Choi SH, DeStefano A, Ikram MA, Bis JC, Smith A, Fitzpatrick AL, Launer L, van Duijn C, Seshadri S, Ulstein ID, Aarsland D, Fladby T, Djurovic S, Hyman BT, Snaedal J, Stefansson H, Stefansson K, Gasser T, Andreassen OA and Dale AM for the ADNI, ADGC, GERAD, CHARGE and IPDGC Investigators. **Genetic overlap between Alzheimer's disease and Parkinson's disease at the MAPT locus.** *Mol Psychiatry.* 2015 Dec;20(12):1588-95. doi: 10.1038/mp.2015.6. Epub 2015 Feb 17. PMID: 25687773. PMCID: PMC4539304.
60. Escott-Price V, Sims R, Bannister C, Harold D, Vronskaya M, Majounie E, Badarinarayan N; GERAD/PERADES; IGAP consortia, Morgan K, Passmore P, Holmes C, Powell J, Brayne C, Gill M, Mead S, Goate A, Cruchaga C, Lambert JC, van Duijn C, Maier W, Ramirez A, Holmans P, Jones L, Hardy J, Seshadri S, Schellenberg GD, Amouyel P, Williams J. **Common polygenic variation enhances risk prediction for Alzheimer's disease.** *Brain.* 2015 Dec;138(Pt 12):3673-84. doi: 10.1093/brain/awv268. Epub 2015 Oct 21. PMID: 26490334. PMCID: PMC5006219.
61. Barral S, Cheng R, Reitz C, Vardarajan B, Lee J, Kunkle B, Beecham G, Cantwell LB, Pericak-Vance MA, Farrer LA, Haines JL, Goate AM, Foroud T, Boerwinkle E, Schellenberg GD, Mayeux R. **Linkage analyses in Caribbean Hispanic families identifies novel loci associated with familial late-onset Alzheimer's disease.** *Alzheimers Dement.* 2015 Dec;11(12):1397-1406. doi: 10.1016/j.jalz.2015.07.487. Epub 2015 Oct 1. PMID: 26433351. PMCID: PMC4690771.
62. Kohli MA, Cukier HN, Hamilton-Nelson KL, Rolati S, Kunkle BW, Whitehead PL, Züchner SL, Farrer LA, Martin ER, Beecham GW, Haines JL, Vance JM, Cuccaro ML, Gilbert JR, Schellenberg GD, Carney RM, Pericak-Vance MA. **Segregation of a rare TTC3 variant in an extended family with late-onset Alzheimer disease.** *Neurol Genet.* 2016 Jan 14;2(1):e41. doi: 10.1212/NXG.0000000000000041. eCollection 2016 Feb. PMID: 27066578. PMCID: PMC4817909.
63. Jun G, Ibrahim-Verbaas CA, Vronskaya M, Lambert J-C, Chung J, Naj AC, Kunkle BW, Wang L-S, Bis JC, Bellenguez C, Harold D, Lunetta KL, Destefano AL, Grenier-Boley B, Sims R, Beecham GW, Smith AV, Chouraki V, Hamilton-Nelson KL, Ikram MA, Fievet N, Denning N, Martin ER, Schmidt H, Kamatani Y, Dunstan ML, Valladares O, Ruiz Laza A, Zelenikax D, Ramirez A, Foroud TM, Choi S-H, Boland A, Becker T, Kukull WA, van der Lee SJ, Pasquier F, Cruchaga C, Beekly D, Fitzpatrick AL, Hanon O, Gill M, Barber R, Gudnason V, Champion D, Love S, Bennett DA, Amin N, Berr C, Tzolaki M, Buxbaum JD, Lopez OL, Deramecourt V, Fox NC, Cantwell LB, Tarraga L, Dufouil C, Hardy J, Crane PK, Eiriksdottir G, Hannequin D, Clarke R, Evans D, Mosley TH, Letenneur L, Brayne C, Maier W, De Jager P, Emilsson V, Dartigues J-F, Hampel H, Kamboh MI, de Bruijn RAFG, Tzourio C, Pastor P, Larson EB, Rotter JI, O'Donovan MC, Montine TJ, Nalls MA, Mead S, Reiman EM, Jonsson PV, Holmes C, St. George-Hyslop PH, Boada M, Passmore P, Wendland JR, Schmidt R, Morgan K, Winslow AR, Powell JF, Carasquillo M,

- Younkin SG, Jakobsdóttir J, Kauwe JSK, Wilhelmsen KC, Rujescu D, Nöthen MM, Hofman A, Jones L, IGAP Consortium, Haines JL, Psaty BM, Van Broeckhoven C, Holmans P, Launer LJ, Mayeux R, Lathrop M, Goate AM, Escott-Price V, Seshadri S, Pericak-Vance MA, Amouyel P, Williams J, van Duijn CM, Schellenberg GD, Farrer LA. **A novel Alzheimer disease locus located near the gene encoding tau protein.** *Mol Psychiatry*. 2016 Jan;21(1):108-17. doi: 10.1038/mp.2015.23. Epub 2015 Mar 17. PMID: 25778476. PMCID: PMC4573764.
64. Kunkle BW, Jaworski J, Barral S, Vardarajan B, Beecham GW, Martin ER, Cantwell LB, Partch A, Bird TD, Raskind WH, DeStefano AL, Carney RM, Cuccaro ML, Vance JM, Farrer LA, Goate AM, Foroud T, Mayeux RP, Schellenberg GD, Haines JL, Pericak-Vance M. **Genome-wide linkage analyses of non-Hispanic white families identifies novel loci for familial late-onset Alzheimer's disease.** *Alzheimers Dement*. 2016 Jan;12(1):2-10. doi: 10.1016/j.jalz.2015.05.020. Epub 2015 Sep 11. PMID: 26365416. PMCID: PMC4717829.
65. Karch CM, Ezerskiy LA, Berelsen S, ADGC, Goate AM. **Alzheimer's disease risk polymorphisms regulate gene expression in the ZCWPW1 and the CELF loci.** *PLoS One*. 2016 Feb 26;11(2):e0148717. doi: 10.1371/journal.pone.0148717. eCollection 2016. PMID: 26919393. PMCID: PMC4769299.
66. Hohman, T.J., Bush, W.S., Jiang, L., Brown-Gentry, K.D., Torstenson, E.S., Dudek, S.M., Mukherjee, S., Naj, A., Kunkle, B.W., Ritchie, M.D., Martin, E.R., Schellenberg, G.D., Mayeux, R., Farrer, L.A., Pericak-Vance, M. A., Haines, J.L., Thornton-Wells, T.A., for the Alzheimer's Disease Genetics Consortium. **Discovery of gene-gene interactions across multiple independent datasets of late onset Alzheimer disease from the Alzheimer Disease Genetics Consortium.** *Neurobiol Aging*. 2016 Feb;38:141-150. doi: 10.1016/j.neurobiolaging.2015.10.031. Epub 2015 Nov 6. PMID: 26827652. PMCID: PMC4735733.
67. Ebbert MT, Boehme KL, Wadsworth ME, Staley LA; Alzheimer's Disease Neuroimaging Initiative; Alzheimer's Disease Genetics Consortium, Mukherjee S, Crane PK, Ridge PG, Kauwe JS. **Interaction between variants in CLU and MS4A4E modulates Alzheimer's disease risk.** *Alzheimers Dement*. 2016 Feb;12(2):121-129. doi: 10.1016/j.jalz.2015.08.163. Epub 2015 Oct 9. PMID: 26449541. PMCID: PMC4744542.
68. Hohman TJ, Cooke-Bailey J, Reitz C, Jun G, Naj A, Beecham G, Liu Z, Rajbhandary R, Vardarajan BN, Wang L-S, Valladares O, Lin C-F, Larson EB, Graff-Radford NR, Evans D, De Jager PL, Crane PK, Buxbaum JD, Murrell JR, Raj T, Ertekin-Taner N, Logue M, Baldwin CT, Green RC, Barnes LL, Cantwell LB, Fallin MD, Go RCP, Griffith P, Obisesan TO, Manly JJ, Lunetta KL, Kamboh MI, Lopez OL, Bennett DA, Hendrie HC, Hall KS, Goate AM, Lang-Walker R, Byrd GS, Kukull WA, Foroud TM, Farrer LA, Martin E, Pericak-Vance MA, Schellenberg GD, Mayeux R, Haines JL, Thornton-Wells TA for the Alzheimer Disease Genetics Consortium. **Global and Local Ancestry in African Americans: Implications for Alzheimer's Disease Risk.** *Alzheimers Dement*. 2016 Mar;12(3):233-43. doi: 10.1016/j.jalz.2015.02.012. Epub 2015 Jun 16. PMID 26092349. PMCID: PMC4681680.
69. Cukier HN, Kunkle BW, Vardarajan BN, Rolati S, Hamilton-Nelson KL, Kohli MA, Whitehead PL, Van Booven D, Lang R, Dykxhoorn DM, Farrer LA, Cuccaro ML, Vance JM, Gilbert JR, Beecham GW, Martin ER, Carney RM, Mayeux R, Schellenberg GD, Byrd GS, Haines JL, Pericak-Vance MA, Alzheimer Disease Genetics Consortium. **ABCA7 Frameshift Deletion Associated with Alzheimer's Disease in African Americans.** *Neurol Genet*. 2016 May 17;2(3):e79. doi: 10.1212/NXG.0000000000000079. eCollection 2016 Jun. PMID: 27231719. PMCID: PMC4871806.
70. Ridge PG, Hoyt KB, Boehme K, Mukherjee S, Crane PK, Haines JL, Mayeux R, Farrer LA, Pericak-Vance MA, Schellenberg GD, Kauwe JSK, Alzheimer's Disease Genetics Consortium. **Assessment of the**

- genetic variance of late-onset Alzheimer's disease.** *Neurobiol Aging.* 2016 May;41:200.e13-200.e20. doi: 10.1016/j.neurobiolaging.2016.02.024. Epub 2016 Mar 3. PMID: 27036079. PMCID: PMC4948179.
71. Mez J, Mukherjee S, Thornton T, Fardo DW, Trittschuh E, Sutti S, Sherva R, Kauwe JS, Naj AC, Beecham GW, Gross A, Saykin AJ, Green RC, Crane PK, for Executive Prominent Alzheimer's Disease: Genetics and Risk Factors (EPAD:GRF), the Alzheimer's Disease Neuroimaging Initiative (ADNI1), and the Alzheimer's Disease Genetics Consortium (ADGC). **The executive prominent / memory prominent spectrum in Alzheimer's disease is highly heritable.** *Neurobiol Aging.* 2016 May;41:115-121. doi: 10.1016/j.neurobiolaging.2016.02.015. Epub 2016 Feb 21. PMID: 27103524. PMCID: PMC4843522.
72. Yokoyama JS, Wang Y, Schork AJ, Thompson WK, Karch CM, Cruchaga C, McEvoy LK, Witoelar A, Chen CH, Holland D, Brewer JB, Franke A, Dillon WP, Wilson DM, Mukherjee P, Hess CP, Miller Z, Bonham LW, Shen J, Rabinovici GD, Rosen HJ, Miller BL, Hyman BT, Schellenberg GD, Karlsen TH, Andreassen OA, Dale AM, Desikan RS; Alzheimer's Disease Neuroimaging Initiative. **Association between genetic traits for immune-mediated diseases and Alzheimer Disease.** *JAMA Neurol.* 2016 Jun 1;73(6):691-7. doi: 10.1001/jamaneurol.2016.0150. PMID: 27088644. PMCID: PMC4905783.
73. Bonham LW, Geier EG, Fan CC, Leong JK, Besser L, Kukull WA, Kornak J, Andreassen OA, Schellenberg GD, Rosen HJ, Dillon WP, Hess CP, Miller BL, Dale AM, Desikan RS, Yokoyama JS. **Age-dependent effects of APOE  $\epsilon$ 4 in preclinical Alzheimer's disease.** *Ann Clin Transl Neurol.* 2016 Aug 26;3(9):668-77. doi: 10.1002/acn3.333. eCollection 2016 Sep. PMID: 27648456. PMCID: PMC5018579.
74. Jakobsdottir J, van der Lee SJ, Bis C, Chouraki, Li-Kroeger D, Yamamoto S, Grove ML, Na a, Vronskaya M, Salazar JL, DeStefano AL, Brody JA, Smith AV, Amin N, Sims R, Ibrahim-Verbaas CA, Choi S-H, Satizabal CL, Lopez OL, Beiser A, Ikram MA, Garcia ME, Hayward C, Varga TV, Ripatti S, Franks PW, Hallmans G, Rolandsson O, Jansson J-H, Porteous DJ, Salomaa V, Eiriksdottir B, Rice KM, Bellen HJ, Levy D, Uitterlinden AG, Emilsson V, Rotter JI, Aspelund T, CHARGE, ADGC, GERAD, O'Donnell CJ, Fitzpatrick AL, Launer LJ, Hofman A, Wang L-S, Williams J, Schellenberg GD, Boerwinkle E, Psaty BM, Seshadri S, Shulman JM, Gudnason V, van Duijn CM. **Rare functional variant in *TM2D3* is associated with late-onset Alzheimer's disease.** *PLoS Genet.* 2016 Oct 20;12(10):e1006327. doi: 10.1371/journal.pgen.1006327. eCollection 2016 Oct. Erratum in: *PLoS Genet.* 2016 Nov 28;12(11):e1006456. PMID: 27764101. PMCID: PMC5072721.
75. Nelson PT, Katsumata Y, Nho K, Artiushin SC, Jicha GA, Wang W-X, Abner EL, Saykin AJ, Kukull WA, Alzheimer's Disease Neuroimaging Initiative (ADNI), Fardo DW. **Genomics and CSF analyses implicate thyroid hormone in hippocampal sclerosis of aging.** *Acta Neuropathol.* 2016 Dec;132(6):841-858. Epub 2016 Nov 4. PMID: 27815632. PMCID: PMC5154615.
76. Naj AC, Schellenberg GD; Alzheimer's Disease Genetics Consortium (ADGC). **Genomic variants, genes, and pathways of Alzheimer's disease: An overview.** *Am J Med Genet B Neuropsychiatr Genet.* 2017 Jan;174(1):5-26. doi: 10.1002/ajmg.b.32499. Review. PMID: 27943641.
77. Ighodaro ET, Abner EL, Fardo DW, Lin A-L, Katsumata Y, Schmitt FA, Kryscio RJ, Jicha GA, Neltner JH, Monsell SE, Kukull WA, Appiah F, Bachstetter AD, Van Eldik LJ, Nelson PT. **Risk Factors and Global Cognitive Status Related to Brain Arteriolosclerosis in Elderly Individuals.** *J Cereb Blood Flow Metab.* 2017 Jan;37(1):201-216. Epub 2016 Jan 6. PMID: 26738751. PMCID: PMC5363738.
78. Haddick PC, Larson JL, Rathore N, Bhangale TR, Phung QT, Srinivasan K, Hansen DV, Lill JR; Alzheimer's Disease Genetic Consortium (ADGC), Alzheimer's Disease Neuroimaging Initiative (ADNI)., Pericak-Vance MA, Haines J, Farrer LA, Kauwe JS, Schellenberg GD, Cruchaga C, Goate AM, Behrens TW, Watts RJ, Graham RR, Kaminker JS, van der Brug M. **A common variant of *IL-6R* is associated**



- with elevated IL-6 pathway activity in Alzheimer's Disease brains.** *J Alzheimers Dis.* 2017 Feb 3;56(3):1037-1054. doi: 10.3233/JAD-160524. PMID: 28106546. PMCID: PMC5667357.
79. Mez J, Chung J, Jun G, Kriegel J, Bourlas AP, Sherva R, Logue MW, Bennett DA, Buxbaum JD, Byrd GS, Ertekin-Taner N, Evans D, Fallin MD, Foroud T, Goate A, Graff-Radford NR, Kamboh MI, Kukull WA, Manly JJ, Alzheimer Disease Genetics Consortium, Haines JL, Mayeux R, Pericak-Vance MA, Schellenberg GD, Lunetta KL, and Farrer LA. **Two novel loci, COBL and SLC10A2, for Alzheimer's disease in African Americans.** *Alzheimers Dement.* 2017 Feb;13(2):119-129. doi: 10.1016/j.jalz.2016.09.002. Epub 2016 Oct 20. PMID: 27770636. PMCID: PMC5318231.
80. Ferrari R, Wang Y, Vandrovcova J, Guelfi S, Witeolar A, Karch CM, Schork AJ, Fan CC, Brewer JB; International FTD-Genomics Consortium (IFGC); International Parkinson's Disease Genomics Consortium (IPDGC); International Genomics of Alzheimer's Project (IGAP), Momeni P, Schellenberg GD, Dillon WP, Sugrue LP, Hess CP, Yokoyama JS, Bonham LW, Rabinovici GD, Miller BL, Andreassen OA, Dale AM, Hardy J, Desikan RS. **Genetic architecture of sporadic frontotemporal dementia and overlap with Alzheimer's and Parkinson's diseases.** *J Neurol Neurosurg Psychiatry.* 2017 Feb;88(2):152-164. doi: 10.1136/jnnp-2016-314411. Epub 2016 Nov 29. PMID: 27899424. PMCID: PMC5237405.
81. Desikan RS, Fan CC, Wang Y, Schork AJ, Cabral HJ, Cupples LA, Thompson WK, Besser L, Kukull WA, Holland D, Chen C-H, Brewer JB, Galasko DR, Karch CM, Bonham LW, Yokoyama JS, Miller BL, Dillon WP, Wilson DM, Hess CP, Pericak-Vance MA, Haines JL, Farrer LA, Mayeux R, Hardy J, Goate AM, Hyman BT, Schellenberg GD, McEvoy LK, Andreassen OA, Dale AM. **Genetic assessment of age-associated Alzheimer's disease risk: development and validation of a polygenic hazard score.** *PLoS Med.* 2017 Mar 21;14(3):e1002258. doi: 10.1371/journal.pmed.1002258. eCollection 2017 Mar. Erratum in: *PLoS Med.* 2017 Mar 28;14(3):e1002289. PMID: 28323831. PMCID: PMC5370097.
82. Steele NZ, Carr JS, Bonham LW, Geier EG, Damotte V, Miller ZA, Desikan RS, Boehme KL, Mukherjee S, Crane PK, Kauwe JS, Kramer JH, Miller BL, Coppola G, Hollenbach JA, Huang Y, Yokoyama JS. **Fine-mapping of the human leukocyte antigen locus as a risk factor for Alzheimer disease: A case-control study.** *PLoS Med.* 2017 Mar 28;14(3):e1002272. doi: 10.1371/journal.pmed.1002272. eCollection 2017 Mar. PMID: 28350795. PMCID: PMC5369701.
83. Butkiewicz M, Haines JL, Bush WS. **Introducing COCOS: codon consequence scanner for annotating reading frame changes induced by stop-lost and frame shift variants.** *Bioinformatics.* 2017 May 15;33(10):1561-1562. doi: 10.1093/bioinformatics/btw820. PMID: 28122779. PMCID: PMC5423451.
84. Deming Y, Harari O, Li Z, Black K, Carrell D, Cai Y, Del-Aguila JL, Fernandez MV, Budde J, Ma S, Saef B, Howells B, Huang K-I, Bertelsen S, Fagan AM, Holtzman DM, Morris JC, Kim S, Saykin AJ, De Jager PL, Albert M, Moghekar A, O'Brien R, Riemenschneider M, Petersen RC, Blennow K, Zetterberg H, Minthon L, Van Deerlin VM, Lee VM, Shaw LM, Trojanowski JQ, Peskind ER, Li G, Alzheimer's Disease Neuroimaging Initiative (ADNI), The Alzheimer Disease Genetic Consortium (ADGC), Kauwe J, Goate A, Cruchaga C. **Genome-wide association study identifies four novel loci associated with Alzheimer's endophenotypes and disease modifiers.** *Acta Neuropathol.* 2017 May;133(5):839-856. doi: 10.1007/s00401-017-1685-y. Epub 2017 Feb 28. PMID: 28247064. PMCID: PMC5613285.
85. Chapuis J, Flaig A, Grenier-Boley B, Eysert F, Pottiez V, Deloison G, Vandeputte A, Ayrat AM, Mendes T, Desai S, Goate AM, Kauwe JS, Leroux F, Herledan A, Demiautte F, Bauer C, Checler F, Petersen RC, Blennow K, Zetterberg H, Minthon L, Van Deerlin VM, Lee VM, Shaw LM, Trojanowski JQ, Albert M, Moghekar A, O'Brien R, Peskind ER, Malmanche N, Schellenberg GD, Dourlen P, Song OR, Cruchaga C, Amouyel P, Deprez B, Brodin P, Lambert JC; ADGC, Alzheimer's Disease Neuroimaging Initiative. **Genome-wide, high-content siRNA screening identifies the Alzheimer's genetic risk factor FERMT2**

- as a major modulator of APP metabolism.** *Acta Neuropathol.* 2017 Jun;133(6):955-966. doi: 10.1007/s00401-016-1652-z. Epub 2016 Dec 8. PMID: 27933404. PMCID: PMC5427165.
86. Jun G, Chung J, Mez J, Barber R, Beecham GW, Bennett DA, Buxbaum JD, Byrd GS, Carasquillo M, Crane PK, Cruchaga C, De Jager P, Ertekin-Taner N, Evans D, Fallin MD, Foroud RM, Friedland RP, Goate AM, Graff-Radford NR, Hendrie H, Hall KS, Hamilton-Nelson KL, Inzelberg R, Kamboh MI, Kauwe JSK, Kukull WA, Kunkle BW, Kuwano R, Logue MW, Manly JJ, Larson EB, Martin ER, Montine TJ, Naj A, Reiman EM, Reitz C, Sherva R, St. George-Hyslop PH, Younkin SG, Vardarajan BN, Wang L-S, Wendlund JR, Winslow AR, Alzheimer Disease Genetics Consortium\*, Haines J, Mayeux R, Pericak-Vance MA, Schellenberg G, Lunetta K, and Farrer LA. **Transethnic Genome-Wide Scan Identifies Novel Alzheimer Disease Loci.** *Alzheimers Dement.* 2017 Jul;13(7):727-738. doi: 10.1016/j.jalz.2016.12.012. Epub 2017 Feb 7. PMID: 28183528. PMCID: PMC5496797.
87. Monsell SE, Mock C, Fardo DW, Berelsen S, Cairns NJ, Roe CM, Ellingson S, Morris JC, Goate AM, Kukull WA. **Genetic comparison of symptomatic and asymptomatic persons with Alzheimer disease neuropathology.** *Alzheimer Dis Assoc Disord.* 2017 Jul-Sep;31(3):232-238. doi: 10.1097/WAD.000000000000179. PMID: 27849641. PMCID: PMC5432419.
88. Huang K, Marcora E, Pimenova AA, Di Narzo AF, Kapoor M, Jin SC, Harari O, Bertelsen S, Fairfax B, Czajkowski J, Lambert JC, Chouraki V, Bellenguez C, Grenier-Boley B, Deming Y, McKenzie A, Raj T, Renton AE, Budde J, Del-Aguila JL, Fernandez MV, Ibañez L, The International Genomics of Alzheimer's Project, The Alzheimer's Disease Neuroimaging Initiative, Sims R, Escott-Price, Mayeux R, Haines JL, Farrer LA, Pericak-Vance MA, Seshadri S, Williams J, Amouyel P, Schellenberg GD, Zhang B, Borecki I, Kauwe JSK, Cruchaga C, Hao K, Goate AM. **A common haplotype lowers *PU.1* expression in myeloid cells and delays onset of Alzheimer's disease.** *Nat Neurosci.* 2017 Aug;20(8):1052-1061. doi: 10.1038/nn.4587. Epub 2017 Jun 19. PMID: 28628103. PMCID: PMC5759334.
89. Cukier HN, Kunkle BW, Hamilton-Nelson KL, Rolati S, Kohli MA, Whitehead PL, Vance JM, Cuccaro M, Carney RM, Gilbert JR, Farrer LA, Martin ER, Beecham GW, Haines JL, Pericak-Vance MA. **Exome sequencing of extended families with Alzheimer's disease identifies novel genes implicated in cell immunity and neuronal function.** *J Alzheimers Dis Parkinsonism.* 2017 Aug;7(4). pii: 355. doi: 10.4172/2161-0460.1000355. Epub 2017 Jul 31. PMID: 29177109. PMCID: PMC5698805.
90. Kunkle BW, Vardarajan B, Naj AC, Whitehead PL, Rolati S, Slifer S, Carney RM, Cuccaro ML, Vance JM, Gilbert JR, The Alzheimer's Disease Genetics Consortium, Wang LS, Farrer LA, Reitz C, Haines JL, Beecham GW, Martin ER, Schellenberg GD, Mayeux RP, Pericak-Vance MA. **Early-onset Alzheimer's disease and candidate risk genes involved in endolysosomal transport.** *JAMA Neurol.* 2017 Sep 1;74(9):1113-1122. doi: 10.1001/jamaneurol.2017.1518. PMID: 28738127. PMCID: PMC5691589.
91. Tan CH, Hyman BT, Tan JJX, Hess CP, Dillon WP, Schellenberg GD, Besser LM, Kukull WA, Kauppi K, McEvoy LK, Andreassen OA, Dale AM, Fan CC, Desikan RS (2017) **Polygenic hazard scores in preclinical Alzheimer's disease.** *Ann Neurol.* 2017 Sep;82(3):484-488. doi: 10.1002/ana.25029. PMID: 28940650. PMCID: PMC5758043.
92. Sims R, van der Lee SJ, Naj AC, Bellenguez C, Badarinarayan N, Jakobsdottir J, Kunkle BW, Boland A, Raybould R, Bis JC, Martin ER, Grenier-Boley B, Heilmann-Heimbach S, Chouraki V, Kuzma AB, Sleegers K, Vronskaya M, Ruiz A, Graham RR, Olaso R, Hoffmann P, Grove ML, Vardarajan BN, Hiltunen M, Nöthen MM, White CC, Hamilton-Nelson KL, Epelbaum J, Maier W, Choi SH, Beecham GW, Dulary C, Herms S, Smith AV, Funk CC, Derbois C, Forstner AJ, Ahmad S, Li H, Bacq D, Harold D, Satizabal CL, Valladares O, Squassina A, Thomas R, Brody JA, Qu L, Sánchez-Juan P, Morgan T, Wolters FJ, Zhao Y, Garcia FS, Denning N, Fornage M, Malamon J, Naranjo MCD, Majounie E, Mosley TH, Dombroski B,

Wallon D, Lupton MK, Dupuis J, Whitehead P, Fratiglioni L, Medway C, Jian X, Mukherjee S, Keller L, Brown K, Lin H, Cantwell LB, Panza F, McGuinness B, Moreno-Grau S, Burgess JD, Solfrizzi V, Proitsi P, Adams HH, Allen M, Seripa D, Pastor P, Cupples LA, Price ND, Hannequin D, Frank-García A, Levy D, Chakrabarty P, Caffarra P, Giegling I, Beiser AS, Giedraitis V, Hampel H, Garcia ME, Wang X, Lannfelt L, Mecocci P, Eiriksdottir G, Crane PK, Pasquier F, Boccardi V, Henández I, Barber RC, Scherer M, Tarraga L, Adams PM, Leber M, Chen Y, Albert MS, Riedel-Heller S, Emilsson V, Beekly D, Braae A, Schmidt R, Blacker D, Masullo C, Schmidt H, Doody RS, Spalletta G, Longstreth WT Jr, Fairchild TJ, Bossù P, Lopez OL, Frosch MP, Sacchinelli E, Ghetti B, Yang Q, Huebinger RM, Jessen F, Li S, Kamboh MI, Morris J, Sotolongo-Grau O, Katz MJ, Corcoran C, Dunstan M, Braddel A, Thomas C, Meggy A, Marshall R, Gerrish A, Chapman J, Aguilar M, Taylor S, Hill M, Fairén MD, Hodges A, Vellas B, Soininen H, Kloszewska I, Daniilidou M, Uphill J, Patel Y, Hughes JT, Lord J, Turton J, Hartmann AM, Cecchetti R, Fenoglio C, Serpente M, Arcaro M, Caltagirone C, Orfei MD, Ciaramella A, Pichler S, Mayhaus M, Gu W, Lleó A, Fortea J, Blesa R, Barber IS, Brookes K, Cupidi C, Maletta RG, Carrell D, Sorbi S, Moebus S, Urbano M, Pilotto A, Kornhuber J, Bosco P, Todd S, Craig D, Johnston J, Gill M, Lawlor B, Lynch A, Fox NC, Hardy J; ARUK Consortium, Albin RL, Apostolova LG, Arnold SE, Asthana S, Atwood CS, Baldwin CT, Barnes LL, Barral S, Beach TG, Becker JT, Bigio EH, Bird TD, Boeve BF, Bowen JD, Boxer A, Burke JR, Burns JM, Buxbaum JD, Cairns NJ, Cao C, Carlson CS, Carlsson CM, Carney RM, Carrasquillo MM, Carroll SL, Diaz CC, Chui HC, Clark DG, Cribbs DH, Crocco EA, DeCarli C, Dick M, Duara R, Evans DA, Faber KM, Fallon KB, Fardo DW, Farlow MR, Ferris S, Foroud TM, Galasko DR, Gearing M, Geschwind DH, Gilbert JR, Graff-Radford NR, Green RC, Growdon JH, Hamilton RL, Harrell LE, Honig LS, Huentelman MJ, Hulette CM, Hyman BT, Jarvik GP, Abner E, Jin LW, Jun G, Karydas A, Kaye JA, Kim R, Kowall NW, Kramer JH, LaFerla FM, Lah JJ, Leverenz JB, Levey AI, Li G, Lieberman AP, Lunetta KL, Lyketsos CG, Marson DC, Martiniuk F, Mash DC, Masliah E, McCormick WC, McCurry SM, McDavid AN, McKee AC, Mesulam M, Miller BL, Miller CA, Miller JW, Morris JC, Murrell JR, Myers AJ, O'Bryant S, Olichney JM, Pankratz VS, Parisi JE, Paulson HL, Perry W, Peskind E, Pierce A, Poon WW, Potter H, Quinn JF, Raj A, Raskind M, Reisberg B, Reitz C, Ringman JM, Roberson ED, Rogaeva E, Rosen HJ, Rosenberg RN, Sager MA, Saykin AJ, Schneider JA, Schneider LS, Seeley WW, Smith AG, Sonnen JA, Spina S, Stern RA, Swerdlow RH, Tanzi RE, Thornton-Wells TA, Trojanowski JQ, Troncoso JC, Van Deerlin VM, Van Eldik LJ, Vinters HV, Vonsattel JP, Weintraub S, Welsh-Bohmer KA, Wilhelmsen KC, Williamson J, Wingo TS, Woltjer RL, Wright CB, Yu CE, Yu L, Garzia F, Golamaully F, Septier G, Engelborghs S, Vandenberghe R, De Deyn PP, Fernandez CM, Benito YA, Thonberg H, Forsell C, Lilius L, Kinhult-Ståhlbom A, Kilander L, Brundin R, Concari L, Helisalmi S, Koivisto AM, Haapasalo A, Dermecourt V, Fievet N, Hanon O, Dufouil C, Brice A, Ritchie K, Dubois B, Himali JJ, Keene CD, Tschanz J, Fitzpatrick AL, Kukull WA, Norton M, Aspelund T, Larson EB, Munger R, Rotter JI, Lipton RB, Bullido MJ, Hofman A, Montine TJ, Coto E, Boerwinkle E, Petersen RC, Alvarez V, Rivadeneira F, Reiman EM, Gallo M, O'Donnell CJ, Reisch JS, Bruni AC, Royall DR, Dichgans M, Sano M, Galimberti D, St George-Hyslop P, Scarpini E, Tsuang DW, Mancuso M, Bonuccelli U, Winslow AR, Daniele A, Wu CK; GERAD/PERADES, CHARGE, ADGC, EADI, Peters O, Nacmias B, Riemenschneider M, Heun R, Brayne C, Rubinsztein DC, Bras J, Guerreiro R, Al-Chalabi A, Shaw CE, Collinge J, Mann D, Tsolaki M, Clarimón J, Sussams R, Lovestone S, O'Donovan MC, Owen MJ, Behrens TW, Mead S, Goate AM, Uitterlinden AG, Holmes C, Cruchaga C, Ingelsson M, Bennett DA, Powell J, Golde TE, Graff C, De Jager PL, Morgan K, Ertekin-Taner N, Combarros O, Psaty BM, Passmore P, Younkin SG, Berr C, Gudnason V, Rujescu D, Dickson DW, Dartigues JF, DeStefano AL, Ortega-Cubero S, Hakonarson H, Campion D, Boada M, Kauwe JK, Farrer LA, Van Broeckhoven C, Ikram MA, Jones L, Haines JL, Tzourio C, Launer LJ, Escott-Price V, Mayeux R, Deleuze JF, Amin N, Holmans PA, Pericak-Vance MA, Amouyel P, van Duijn CM, Ramirez A, Wang LS, Lambert JC, Seshadri S, Williams J, Schellenberg GD. **Rare coding variants in *PLCG2*, *ABI3* and *TREM2* implicate microglial-mediated innate immunity in Alzheimer's disease.** Nat Genet. 2017 Sep;49(9):1373-1384. doi: 10.1038/ng.3916. Epub 2017 Jul 17. PMID: PMC5669039.

93. Rehker J, Rodhe J, Nesbitt RR, Boyle EA, Martin BK, Lord J, Karaca I, Naj A, Jessen F, Helisalmi S, Soininen H, Hiltunen M, Ramirez A, Scherer M, Farrer LA, Haines JL, Pericak-Vance MA, Raskind WH, G

- Schellenberg, Cruchaga C, Joseph B, Brkanac Z. **Caspase-8, association with Alzheimer's disease and functional analysis of rare variants.** PLoS One. 2017 Oct 6;12(10):e0185777. doi: 10.1371/journal.pone.0185777. eCollection 2017. PMID: 28985224. PMCID: PMC5630132.
94. Beecham GW, Bis JC, Martin ER, Choi S-H, DeStefano A, van Duijn CM, Fornage M, Gabriel SB, Koldt DC, Larson DE, Naj AC, Psaty BM, Salerno W, Bush WS, Foroud TM, Wijsman E, Farrer L, Goate A, Haines JL, Pericak-Vance MA, Boerwinkle E, Mayeux R, Seshadri S, Schellenberg GD. **The Alzheimer's Disease Sequencing Project: study design and sample selection.** Neurol Genet. 2017 Oct 13;3(5):e194. doi: 10.1212/NXG.000000000000194. eCollection 2017 Oct. PMID: 29184913. PMCID: PMC5646177.
95. Tan CH, Fan CC, Mormino EC, Sugrue LP, Broce IJ, Hess CP, Dillon WP, Bonham LW, Yokoyama JS, Karch CM, Brewer JB, Rabinovici GD, Miller BL, Schellenberg GD, Kauppi K, Feldman HA, Holland D, McEvoy LK, Hyman BT, Bennett DA, Andreassen OA, Dale AM, Desikan R. (2018) **Polygenic hazard score: an enrichment marker for Alzheimer's associated amyloid and tau deposition.** Acta Neuropathol. 2018 Jan;135(1):85-93. doi: 10.1007/s00401-017-1789-4. Epub 2017 Nov 24. PMID:29177679, PMCID: PMC5758038.
96. Chung J, Zhang X, Allen M, Wang X, Ma Y, Beecham G, Montine TJ, Younkin SG, Dickson DW, Golde TE, Price ND, Ertekin-Taner N, Lunetta KL, Mez J; Alzheimer's Disease Genetics Consortium, Mayeux R, Haines JL, Pericak-Vance MA, Schellenberg G, Jun GR, Farrer LA. **Genome-wide pleiotropy analysis of neuropathological traits related to Alzheimer's disease.** Alzheimers Res Ther. 2018 Feb 20;10(1):22. doi: 10.1186/s13195-018-0349-z. PMID: 29458411. PMCID: PMC5819208.
97. Vardarajan BN, Barral S, Jaworski J, Beecham GW, Blue E, Tosto G, Reyes-Dumeyer D, Medrano M, Lantigua R, Naj A, Thornton T, DeStefano A, Martin E, Wang L-S, Brown L, Bush W, Van Duijn C, Goate A, Farrer L, Haines JL, Boerwinkle E, Schellenberg G, Wijsman E, Pericak-Vance MA, and Mayeux R for The Alzheimer's Disease Sequencing Project. **Whole Genome Sequencing of Caribbean Hispanic Families with Late-onset Alzheimer's Disease.** Ann Clin Transl Neurol. 2018 Mar 13;5(4):406-417. doi: 10.1002/acn3.537. eCollection 2018 Apr. PMID: 29688227. PMCID: PMC5899906.
98. Teslovich TM, Kim DS, Yin X, Stančáková A, Jackson AU, Wielscher M, Naj A, Perry JRB, Huyghe JR, Stringham HM, Davis JP, Raulerson CK, Welch RP, Fuchsberger C, Locke AE, Sim X, Chines PS, Narisu N, AK, Soininen, Genetics of Obesity-related Liver Disease Consortium (GOLD), The Alzheimer's Disease Genetics Consortium (ADGC) , The DIAbetes Genetics Replication And Meta-analysis (DIAGRAM), Ala-Korpela M, Jarvelin M-J, Schellenberg GD, Gudnason V, Musani SK, Speliotes EK, Kuusisto J, Collins FS, Boehnke M, Laakso M, Mohlke KL (2018) **Identification of seven novel loci associated with amino acid levels using single variant and gene-based tests in 8,545 Finnish men from the METSIM study.** Hum Mol Genet. 2018 May 1;27(9):1664-1674. doi: 10.1093/hmg/ddy067. PMID: 29481666. PMCID: PMC5905595.
99. Lancour D, Naj A, Mayeux R, Haines JL, Pericak-Vance MA, Schellenberg GC, Crovella M, Farrer LA, Kasif S. **One for all and all for one: improving replication of genetic studies through network diffusion.** PLoS Genet. 2018 Apr 23;14(4):e1007306. doi: 10.1371/journal.pgen.1007306. eCollection 2018 Apr. PMID: 29684019. PMCID: PMC5933817.
100. Raghavan NS, Brickman AM, Andrews H, Manly JJ, Schupf N, Lantigua R, Wolock CJ, Kamalakaran S, Petrovski S, Tosto G, Vardarajan BN, Goldstein DB, Mayeux R; Alzheimer's Disease Sequencing Project. **Whole-exome sequencing in 20,197 persons for rare variants in Alzheimer's disease.** Ann Clin Transl Neurol. 2018 May 24;5(7):832-842. doi: 10.1002/acn3.582. eCollection 2018 Jul. PMID: 30009200. PMCID: PMC6043775.

101. Chung J, Wang X, Maruyama T, Ma Y, Zhang X, Mez J, Sherva R, Takeyama H, The Alzheimer's Disease Neuroimaging Initiative, Lunetta KL, Farrer LA, Jun GR. **Genome-wide association study of Alzheimer's disease endophenotypes at prediagnosis stages.** *Alzheimers Dement.* 2018 May;14(5):623-633. doi: 10.1016/j.jalz.2017.11.006. Epub 2017 Dec 20. PMID: 29274321. PMCID: PMC5938137.
102. Blue EE, Bis JC, Dorschner MO, Tsuang DW, Barral SM, Beecham G, Below JE, Bush WS, Butkiewicz M, Cruchaga C, DeStefano A, Farrer LA, Goate A, Haines J, Jaworski J, Jun G, Kunkle B, Kuzma A, Lee JJ, Lunetta KL, Ma Y, Martin E, Naj A, Nato AQ, Navas P, Nguyen H, Reitz C, Reyes D, Salerno W, Schellenberg GD, Seshadri S, Sohi H, Thornton TA, Valadares O, van Duijn C, Vardarajan BN, Wang LS, Boerwinkle E, Dupuis J, Pericak-Vance MA, Mayeux R, Wijsman EM; on behalf of the Alzheimer's Disease Sequencing Project. **Genetic Variation in Genes Underlying Diverse Dementias May Explain a Small Proportion of Cases in the Alzheimer's Disease Sequencing Project.** *Dement Geriatr Cogn Disord.* 2018 May;45(1-2):1-17. doi: 10.1159/000485503. Epub 2018 Feb 27. PMID: 29486463. PMCID: PMC5971141.
103. Karch CM, Wen N, Fan CC, Yokoyama JS, Kouri N, Ross OA, Höglinger G, Müller U, Ferrari R, Hardy J, Schellenberg GD, International FTD-Genomics Consortium (IFGC), International Parkinson's Disease Genomics Consortium (IPDGC), International Genomics of Alzheimer's Project (IGAP), Miller BL, Sharma M, Van Deerlin V, Smeland OB, Andreassen OA, Dale AM, Veldink JH, Desikan RS. (2018) **Selective genetic overlap between amyotrophic lateral sclerosis and diseases of the frontotemporal dementia spectrum.** *JAMA Neurol.* 2018 Jul 1;75(7):860-875. doi: 10.1001/jamaneurol.2018.0372. PMID: 29630712. PMCID: PMC6043387
104. Zhou Z, Wang W, Wang LS, Zhang NR. **Integrative DNA copy number detection and genotyping from sequencing and array-based platforms.** *Bioinformatics.* 2018 Jul 15;34(14):2349-2355. doi: 10.1093/bioinformatics/bty104. PMID: 29992253. PMCID: PMC6041760.
105. Hohman TJ, Dumitrescu L, Barnes LL, Thambisetty M, Beecham G, Kunkle B, Gifford K, Bush WS, Chibnik LB, Mukherjee S, De Jager PL, Kukull W, Crane PK, Resnick SM, Keene CD, Montine TJ, Schellenberg GD, Haines JL, Zetterberg H, Blennow K, Larson EB, Johnson SC, Albert M, Bennett DA, Schneider JA, Jefferson AL for the Alzheimer's for the Alzheimer's Disease Genetics Consortium and the Alzheimer's Disease Neuroimaging **Sex-specific Association of Apolipoprotein E With Cerebrospinal Fluid Levels of Tau.** *JAMA Neurol.* 2018 Aug 1;75(8):989-998. doi: 10.1001/jamaneurol.2018.0821. PMID: 29801024. PMCID: PMC6142927.
106. Butkiewicz M, Blue EE, Leung YY, Jian X, Marcora E, Renton AE, Kuzma A, Wang LS, Koboldt DC, Haines JL, Bush WS. **Functional annotation of genomic variants in studies of late-onset Alzheimer's disease.** *Bioinformatics.* 2018 Aug 15;34(16):2724-2731. doi: 10.1093/bioinformatics/bty177. PMID: 29590295. PMCID: PMC6084586.
107. Lobach I, Sampson J, Alekseyenko A, Lobach S, Zhang L. **Case-Control Studies Of Gene-Environment Interactions. When A Case Might Not Be The Case.** *PLoS One.* 2018 Aug 22;13(8):e0201140. doi: 10.1371/journal.pone.0201140. eCollection 2018. PMID: 30133451. PMCID: PMC6104951
108. Naj AC, Lin H, Vardarajan BN, White S, Lancour D, Ma Y, Schmidt M, Sun F, Butkiewicz M, Bush WS, Kunkle BW, Malamon J, Amin N, Choi SH, Hamilton-Nelson KL, van der Lee SJ, Gupta N, Koboldt DC, Saad M, Wang B, Nato AQ, Sohi HK, Kuzma A, Wang LS, Adrienne Cupples L, van Duijn C, Seshadri S, Schellenberg GD, Boerwinkle E, Bis JC, Dupuis J, Salerno W, Wijsman EM, Martin ER, DeStefano AL. **Quality control and integration of genotypes from two calling pipelines for whole genome sequence data in the Alzheimer's disease sequencing project.** *Genomics.* 2018 May 29. pii: S0888-

- 7543(18)30281-7. doi: 10.1016/j.ygeno.2018.05.004. [Epub ahead of print]. PMID: 29857119. PMCID: PMC6397097.
109. Lee S, Zhou X, Vardarajan B, Rajan, Wilson RS, Evans D, ADGC, NACC, Bennett DA, NIA-LOAD, Schupf N, Manly J, Mayeux R, Barral S. **Episodic Memory Performance in a Multiethnic Longitudinal Study of 13,979 Elderly.** PLoS One. 2018 Nov 21;13(11):e0206803. doi: 10.1371/journal.pone.0206803. PMID: 30462667 PMCID: PMC6248922.
110. Deming Y, Dumitrescu L, Barnes LL, Thambisetty M, Kunkle B, Gifford KA, Bush WS, Chibnik LB, Mukherjee S, De Jager PL, Kukull W, Huentelman M, Crane PK, Resnick SM, Keene CD, Montine TJ, Schellenberg GD, Haines JL, Zetterberg H, Blennow K, Larson EB, Johnson SC, Albert M, Moghekar A, del Aguila JL, Fernandez MV, Budde J, Hassenstab J, Fagan AM, Riemenschneider M, Petersen RC, Minthon L, Van Deerlin VM, Lee V M-Y, Shaw LM, Trojanowski JQ, Peskind ER, Li G, Cox NJ, for the Alzheimer's Disease Neuroimaging Initiative, for the Alzheimer's Disease Genetics Consortium, Goate AM, Bennett DA, Schneider JA, Jefferson AL, Cruchaga C, Hohman TJ. **Sex-Specific Genetic Predictors of Alzheimer's Disease Biomarkers.** Acta Neuropathol. 2018 Dec;136(6):857-872. doi: 10.1007/s00401-018-1881-4. Epub 2018 Jul 2. PMID: PMID: 29967939. PMCID: PMC6280657.
111. Amlie-Wolf A, Tang M, Mlynarski EE, Kuksa PP, Valladares O, Katanic Z, Tsuang D, Brown CD, Schellenberg GD, Li-San Wang I-S (2018) **INFERNO: INFERRing the molecular mechanisms of Noncoding genetic variants.** Nucleic Acids Res. 2018 Sep 28;46(17):8740-8753. doi: 10.1093/nar/gky686. PMID: 30113658. PMCID: PMC6158604.
112. Beecham G, Vardarajan B, Blue E, Bush WS, Jaworski J, Barral S, DeStefano A, Hamilton-Nelson K, Kunkle B, Martin E, Naj A, Rajabli F, Reitz C, Thornton T, van Duijn CM, Goate A, Seshadri S, Farrer L, Boerwinkle E, Schellenberg GD, Haines J, Wijsman E, Mayeux R, Pericak-Vance MA. **Rare genetic variation implicated in Non-Hispanic White families with Alzheimer disease.** Neurol Genet. 2018 Nov 21;4(6):e286. doi: 10.1212/NXG.0000000000000286. eCollection 2018 Dec. PMID: 30569016. PMCID: PMC6278241.
113. Gusareva ES, Twizere JC, Slegers K, Dourlen P, Abisambra JF, Meier S, Cloyd R, Weiss B, Dermaut B, Bessonov K, van der Lee SJ, Carrasquillo MM, Katsumata Y, Cherkaoui M, Asselbergh B, Ikram MA, Mayeux R, Farrer LA, Haines JL, Pericak-Vance MA, Schellenberg GD, Sims R, Williams J, Amouyel P, van Duijn CM, Ertekin-Taner N, Van Broeckhoven C, Dequiedt F, Fardo DW, Lambert JC, Van Steen K. **Male-specific epistasis between WWC1 and TLN2 genes is associated with Alzheimer's disease.** Neurobiol Aging. 2018 Dec;72:188.e3-188.e12. doi: 10.1016/j.neurobiolaging.2018.08.001. Epub 2018 Aug 9. PMID: 30201328. PMCID: PMC6769421.
114. Bis JC, Jian X, Kunkle BW, Chen Y, Hamilton-Nelson KL, Bush WS, Salerno WJ, Lancour D, Ma Y, Renton AE, Marcora E, Farrell JJ, Zhao Y, Qu L, Ahmad S, Amin N, Amouyel P, Beecham GW, Below JE, Champion D, Cantwell L, Charbonnier C, Chung J, Crane PK, Cruchaga C, Cupples LA, Dartigues JF, Debette S, Deleuze JF, Fulton L, Gabriel SB, Genin E, Gibbs RA, Goate A, Grenier-Boley B, Gupta N, Haines JL, Havulinna AS, Helisalmi S, Hiltunen M, Howrigan DP, Ikram MA, Kaprio J, Konrad J, Kuzma A, Lander ES, Lathrop M, Lehtimäki T, Lin H, Mattila K, Mayeux R, Muzny DM, Nasser W, Neale B, Nho K, Nicolas G, Patel D, Pericak-Vance MA, Perola M, Psaty BM, Quenez O, Rajabli F, Redon R, Reitz C, Remes AM, Salomaa V, Sarnowski C, Schmidt H, Schmidt M, Schmidt R, Soininen H, Thornton TA, Tosto G, Tzourio C, van der Lee SJ, van Duijn CM, Valladares O, Vardarajan B, Wang LS, Wang W, Wijsman E, Wilson RK, Witten D, Worley KC, Zhang X; Alzheimer's Disease Sequencing Project, Bellenguez C, Lambert JC, Kurki MI, Palotie A, Daly M, Boerwinkle E, Lunetta KL, Destefano AL, Dupuis J, Martin ER, Schellenberg GD, Seshadri S, Naj AC, Fornage M, Farrer LA. **Correction: Whole exome sequencing study identifies novel rare and common Alzheimer's-associated variants involved in immune**

- response and transcriptional regulation.** *Mol Psychiatry*. 2019 Oct 21. doi: 10.1038/s41380-019-0529-7. [Epub ahead of print]. PMID:31636380. PMCID: PMC6375806.
115. Lobach I. **Bias in parameter estimates due to omitting gene-environment interaction terms in case-control studies.** *Genet Epidemiol*. 2018 Dec;42(8):838-845. doi: 10.1002/gepi.22154. Epub 2018 Oct 9. PMID: 30302820. PMCID: PMC6239944.
116. Nafikov RA, Nato AQ, Sohi H, Wang B, Brown L, Horimoto AR, Varadarajan B, Barral SM, Tosto G, Mayeux R, Thornton TA, Blue E, Wijsman E. **Analysis of Pedigree Data in Populations with Multiple Ancestries: Strategies for Dealing with Admixture in Caribbean Hispanic Families from the ADSP.** *Genet Epidemiol*. 2018 Sep;42(6):500-515. doi: 10.1002/gepi.22133. Epub 2018 Jun 3. PMID: 29862559. PMCID: PMC6160322.
117. Guerreiro R, Orme T, Naj AC, Kuzma AB, Schellenberg GD, Bras J. **Is APOE  $\epsilon$ 4 required for Alzheimer's disease to develop in TREM2 p.R47H variant carriers?** *Neuropathol Appl Neurobiol*. 2019 Feb;45(2):187-189. doi: 10.1111/nan.12517. Epub 2018 Oct 24. PMID: 30229991. PMCID: PMC6380937.
118. Rajabli F, Feliciano BE, Celis K, Hamilton-Nelson KL, Whitehead PL, Adams LD, Bussies PL, Manrique CP, Rodriguez A, Rodriguez V, Starks T, Byfield GE, Sierra Lopez CB, McCauley JL, Acosta H, Chinae A, Kunkle BW, Reitz C, Farrer LA, Schellenberg GD, Vardarajan BN, Vance JM, Cuccaro ML, Martin ER, Haines JL, Byrd GS, Beecham GW, Pericak-Vance MA. **Ancestral origin of ApoE  $\epsilon$ 4 Alzheimer disease risk in Puerto Rican and African American populations.** *PLoS Genet*. 2018 Dec 5;14(12):e1007791. doi: 10.1371/journal.pgen.1007791. eCollection 2018 Dec. PMID: 30517106. PMCID: PMC6281216.
119. Mukherjee S, Mez J, Trittschuh E, Saykin AJ, Gibbons LE, Fardo DW, Wessels M, Bauman J, Moore M, Choi W-E, Gross AL, Rich J, Loudon DK, Sanders RE, Grabowski TJ, Bird TD, McCurry SM, Snitz BE, Kamboh MI, Lopez OL, De Jager PL, Bennett DA, Keene CD, Larson EB, Crane PK. **Genetic data and cognitively defined late onset Alzheimer's disease subgroups.** *Mol Psychiatry*. 2018 Dec 4. doi: 10.1038/s41380-018-0298-8. [Epub ahead of print]. PMID: 30514930. PMCID: PMC6548676.
120. Zhang X, Zhu C, Beecham G, Vardarajan BN, Ma Y, Lancour D, Farrell JJ, Chung J; Alzheimer's Disease Sequencing Project, Mayeux R, Haines JL, Schellenberg GD, Pericak-Vance MA, Lunetta KL, Farrer LA. **A rare missense variant in CASP7 is associated with familial late-onset Alzheimer disease.** *Alzheimers Dement*. 2019 Mar;15(3):441-452. doi: 10.1016/j.jalz.2018.10.005. Epub 2019 Jan 3. PMID: 30503768. PMCID: PMC6408965.
121. Katsumata Y, Nelson PT, Estus S, the Alzheimer's Disease Neuroimaging Initiative (ADNI), Fardo DW. **Translating Alzheimer's disease-associated polymorphisms into functional candidates: a survey of IGAP genes and SNPs.** *Neurobiol Aging*. 2019 Feb; 74:135-146. doi: 10.1016/j.neurobiolaging.2018.10.017. Epub 2018 Oct 23. PMID: 30448613. PMCID: PMC6331247.
122. Tan CH, Fan C, Mormino E, Sugrue L, Broce I, Hess C, Yokoyama J, Rabinovici G, Miller B, Yaffe K, Schellenberg GD, Kauppi K, Holland D, McEvoy L, Kukull W, Tosun D, Weiner M, Sperling R, Bennett D, Hyman B, Andreassen O, Dale A, Desikan R. **Polygenic Hazard Score, Amyloid Deposition and Alzheimer's Neurodegeneration.** *Brain*. 2019 Feb 1;142(2):460-470. doi: 10.1093/brain/awy327. PMID: 30689776. PMCID: PMC6351776.
123. Leung YY, Valladares O, Chou Y-F, Lin H-J, Kuzma AB, Cantwell L, Qu L, Gangadharan P, Alzheimer's Disease Sequencing Project (ADSP), Salerno WJ, Schellenberg GD and Wang L-S. **VCPA: genomic variant calling pipeline and data management tool for Alzheimer's Disease Sequencing Project.**

- Bioinformatics. 2019 May 15;35(10):1768-1770. doi: 10.1093/bioinformatics/bty894. PMID: 31004159. PMCID: PMC6546126.
124. Broce IJ, Tan CH, Fan CC, Jansen I, Savage JE, Witoelar A, Wen N, Hess CP, Dillon WP, Glastonbury CM, Glymour M, Yokoyama JS, Elahi FM, Rabinovici GD, Miller BL, Mormino EC, Sperling RA, Bennett DA, McEvoy LK, Brewer JB, Feldman HH, Hyman BT, Pericak-Vance M, Haines JL, Farrer LA, Mayeux R, Schellenberg GD, Yaffe K, Sugrue LP, Dale AM, Posthuma D, Andreassen OA, Karch CM, Desikan RS. **Dissecting the genetic relationship between cardiovascular risk factors and Alzheimer's disease.** *Acta Neuropathologica*. 2019 Feb;137(2):209-226. doi: 10.1007/s00401-018-1928-6. Epub 2018 Nov 9. PMID: 30413934. PMCID: PMC6358498.
125. Hu Y, Li M, Lu Q, Weng H, Wang J, Zekavat SM, Yu Z, Li B, Muchnik S, Shi Y, Kunkle BW, Mukherjee S, Natarajan P, Crane PK, Zhao H. **A statistical framework for cross-tissue transcriptome-wide association analysis.** *Nature Genetics*. 2019 Mar;51(3):568-576. doi: 10.1038/s41588-019-0345-7. Epub 2019 Feb 25. PMID: 30804563. PMCID: PMC6788740.
126. Patel D, Mez J, Vardarajan BN, Staley L, Chung J, Zhang X, Farrell JJ, Rynkiewicz MJ, Cannon-Albright LA, Teerlink CC, Stevens J, Corcoran C, Gonzalez Murcia JD, Lopez OL, Mayeux R, Haines JL, Pericak-Vance MA, Schellenberg G, Kauwe JSK, Lunetta KL, Farrer LA; Alzheimer's Disease Sequencing Project. **Association of Rare Coding Mutations With Alzheimer Disease and Other Dementias Among Adults of European Ancestry.** *JAMA Netw Open*. 2019 Mar 1;2(3):e191350. doi: 10.1001/jamanetworkopen.2019.1350. PMID: 30924900. PMCID: PMC6450321.
127. Lo M-T, Kauppi K, Fan C-C, Sanyal N, Reas E, Farrer LA, Haines JL, Mayeux R, Pericak-Vance M, Schellenberg GD, Lee W-C, Desikan RS, McEvoy LK, Chen C-H, Alzheimer's Disease Genetics Consortium. **Identification of Genetic Heterogeneity of Alzheimer's Disease across Age.** *Neurobiol Aging*. 2019 Mar 12. pii: S0197-4580(19)30072-7. doi: 10.1016/j.neurobiolaging.2019.02.022. [Epub ahead of print]. PMID: 30979435. PMCID: PMC6783343.
128. Lobach I, Kim I, Alekseyenko A, Lobach S, Zhang L. **A simple approximation to bias in the genetic effect estimates when multiple disease states share a clinical diagnosis.** *Front Genet*. 2019 Oct 9;10:886. doi: 10.3389/fgene.2019.00886. eCollection 2019. PMID: 31681402 PMCID: PMC6812609.
129. Kunkle BW, Grenier-Boley B, Sims R, Bis JC, Damotte V, Naj AC, Boland A, Vronskaya M, van der Lee SJ, Amlie-Wolf A, Bellenguez C, Frizatti A, Chouraki V, Martin ER, Sleegers K, Badarinarayan N, Jakobsdottir J, Hamilton-Nelson KL, Moreno-Grau S, Olsaso R, Raybould R, Chen Y, Kuzma AB, Hiltunen M, Morgan T, Ahmad S, Vardarajan BN, Epelbaum J, Hoffmann P, Boada M, Beecham GW, Garnier JG, Harold D, Fitzpatrick AL, Valladares O, Moutet ML, Gerrish A, Smith AV, Qu L, Bacq D, Denning N, Jian X, Zhao Y, Del Zompo M, Fox NC, Choi SH, Mateo I, Hughes JT, Adams HH, Malamon J, Sanchez-Garcia F, Patel Y, Brody JA, Dombroski BA, Naranjo MCD, Daniilidou M, Eiriksdottir G, Mukherjee S, Wallon D, Uphill J, Aspelund T, Cantwell LB, Garzia F, Galimberti D, Hofer E, Butkiewicz M, Fin B, Scarpini E, Sarnowski C, Bush WS, Meslage S, Kornhuber J, White CC, Song Y, Barber RC, Engelborghs S, Sordon S, Vojnovic D, Adams PM, Vandenberghe R, Mayhaus M, Cupples LA, Albert MS, De Deyn PP, Gu W, Himali JJ, Beekly D, Squassina A, Hartmann AM, Orellana A, Blacker D, Rodriguez-Rodriguez E, Lovestone S, Garcia ME, Doody RS, Munoz-Fernandez C, Sussams R, Lin H, Fairchild TJ, Benito YA, Holmes C, Karamujić-Čomić H, Frosch MP, Thonberg H, Maier W, Roschupkin G, Ghetti B, Giedraitis V, Kawalia A, Li S, Huebinger RM, Kilander L, Moebus S, Hernández I, Kamboh MI, Brundin R, Turton J, Yang Q, Katz MJ, Concaro L, Lord J, Beiser AS, Keene CD, Helisalmi S, Kloszewska I, Kukull WA, Koivisto AM, Lynch A, Tarraga L, Larson EB, Haapasalo A, Lawlor B, Mosley TH, Lipton RB, Solfrizzi V, Gill M, Longstreth WT Jr, Montine TJ, Frisardi V, Diez-Fairen M, Rivadeneira F, Petersen RC, Deramecourt V, Alvarez I, Salani F, Ciaramella A, Boerwinkle E, Reiman EM, Fievet N, Rotter JI, Reisch



- JS, Hanon O, Cupidi C, Andre Uitterlinden AG, Royall DR, Dufouil C, Maletta RG, de Rojas I, Sano M, Brice A, Cecchetti R, George-Hyslop PS, Ritchie K, Tsolaki M, Tsuang DW, Dubois B, Craig D, Wu CK, Soininen H, Avramidou D, Albin RL, Fratiglioni L, Germanou A, Apostolova LG, Keller L, Koutroumani M, Arnold SE, Panza F, Gkatzima O, Asthana S, Hannequin D, Whitehead P, Atwood CS, Caffarra P, Hampel H, Quintela I, Carracedo Á, Lannfelt L, Rubinsztein DC, Barnes LL, Pasquier F, Frölich L, Barral S, McGuinness B, Beach TG, Johnston JA, Becker JT, Passmore P, Bigio EH, Schott JM, Bird TD, Warren JD, Boeve BF, Lupton MK, Bowen JD, Proitsi P, Boxer A, Powell JF, Burke JR, Kauwe JSK, Burns JM, Mancuso M, Buxbaum JD, Bonuccelli U, Cairns NJ, McQuillin A, Cao C, Livingston G, Carlson CS, Bass NJ, Carlsson CM, Hardy J, Carney RM, Bras J, Carrasquillo MM, Guerreiro R, Allen M, Chui HC, Fisher E, Masullo C, Crocco EA, DeCarli C, Bisceglia G, Dick M, Ma L, Duara R, Graff-Radford NR, Evans DA, Hodges A, Faber KM, Scherer M, Fallon KB, Riemenschneider M, Fardo DW, Heun R, Farlow MR, Kölsch H, Ferris S, Leber M, Foroud TM, Heuser I, Galasko DR, Giegling I, Gearing M, Hüll M, Geschwind DH, Gilbert JR, Morris J, Green RC, Mayo K, Growdon JH, Feulner T, Hamilton RL, Harrell LE, Driche D, Honig LS, Cushion TD, Huentelman MJ, Hollingworth P, Hulette CM, Hyman BT, Marshall R, Jarvik GP, Meggy A, Abner E, Menzies GE, Jin LW, Leonenko G, Real LM, Jun GR, Baldwin CT, Grozeva D, Karydas A, Russo G, Kaye JA, Kim R, Jessen F, Kowall NW, Vellas B, Kramer JH, Vardy E, LaFerla FM, Jöckel KH, Lah JJ, Dichgans M, Leverenz JB, Mann D, Levey AI, Pickering-Brown S, Lieberman AP, Klopp N, Lunetta KL, Wichmann HE, Lyketsos CG, Morgan K, Marson DC, Brown K, Martiniuk F, Medway C, Mash DC, Nöthen MM, Masliah E, Hooper NM, McCormick WC, Daniele A, McCurry SM, Bayer A, McDavid AN, Gallacher J, McKee AC, van den Bussche H, Mesulam M, Brayne C, Miller BL, Riedel-Heller S, Miller CA, Miller JW, Al-Chalabi A, Morris JC, Shaw CE, Myers AJ, Wiltfang J, O'Bryant S, Olichney JM, Alvarez V, Parisi JE, Singleton AB, Paulson HL, Collinge J, Perry WR, Mead S, Peskind E, Cribbs DH, Rossor M, Pierce A, Ryan NS, Poon WW, Nacmias B, Potter H, Sorbi S, Quinn JF, Sacchinelli E, Raj A, Spalletta G, Raskind M, Caltagirone C, Bossù P, Orfei MD, Reisberg B, Clarke R, Reitz C, Smith AD, Ringman JM, Warden D, Roberson ED, Wilcock G, Rogoewa E, Bruni AC, Rosen HJ, Gallo M, Rosenberg RN, Ben-Shlomo Y, Sager MA, Mecocci P, Saykin AJ, Pastor P, Cuccaro ML, Vance JM, Schneider JA, Schneider LS, Slifer S, Seeley WW, Smith AG, Sonnen JA, Spina S, Stern RA, Swerdlow RH, Tang M, Tanzi RE, Trojanowski JQ, Troncoso JC, Van Deerlin VM, Van Eldik LJ, Vinters HV, Vonsattel JP, Weintraub S, Welsh-Bohmer KA, Wilhelmsen KC, Williamson J, Wingo TS, Woltjer RL, Wright CB, Yu CE, Yu L, Saba Y; Alzheimer Disease Genetics Consortium (ADGC); European Alzheimer's Disease Initiative (EADI); Cohorts for Heart and Aging Research in Genomic Epidemiology Consortium (CHARGE); Genetic and Environmental Risk in AD/Defining Genetic, Polygenic and Environmental Risk for Alzheimer's Disease Consortium (GERAD/PERADES),, Pilotto A, Bullido MJ, Peters O, Crane PK, Bennett D, Bosco P, Coto E, Boccardi V, De Jager PL, Lleo A, Warner N, Lopez OL, Ingelsson M, Deloukas P, Cruchaga C, Graff C, Gwilliam R, Fornage M, Goate AM, Sanchez-Juan P, Kehoe PG, Amin N, Ertekin-Taner N, Berr C, Dobbie S, Love S, Launer LJ, Younkin SG, Dartigues JF, Corcoran C, Ikram MA, Dickson DW, Nicolas G, Campion D, Tschanz J, Schmidt H, Hakonarson H, Clarimon J, Munger R, Schmidt R, Farrer LA, Van Broeckhoven C, C O'Donovan M, DeStefano AL, Jones L, Haines JL, Deleuze JF, Owen MJ, Gudnason V, Mayeux R, Escott-Price V, Psaty BM, Ramirez A, Wang LS, Ruiz A, van Duijn CM, Holmans PA, Seshadri S, Williams J, Amouyel P, Schellenberg GD, Lambert JC, Pericak-Vance MA. **Genetic meta-analysis of diagnosed Alzheimer's disease identifies new risk loci and implicates A $\beta$ , tau, immunity and lipid processing.** *Nat Genet.* 2019 Sep;51(9):1423-1424. doi: 10.1038/s41588-019-0495-7. PMID: 31417202.
130. Ma Y, Jun GR, Chung J, Zhang X, Kunkle B, Naj A, White C, Bennett D, De Jager P, Mayeux R, Haines J, Pericak-Vance M, Schellenberg GD, Farrer L, Lunetta K. **CpG-related SNPs in the MS4A Region have a Dose-Dependent Effect on Risk of Late-onset Alzheimer Disease.** *Aging Cell.* 2019 Aug;18(4):e12964. doi: 10.1111/accel.12964. Epub 2019 May 29. PMID: 31144443. PMCID: PMC6612647.
131. Dumitrescu L, Barnes LL, Thambisetty M, Beecham G, Kunkle B, Bush WS, Gifford KA, Chibnik LB, Mukherjee S, De Jager PL, Kukull W, Crane PK, Resnick SM, Keene CD, Montine TJ, Schellenberg GD,

- Deming Y, Chao MJ, Huentelman M, Martin ER, Hamilton-Nelson K, Shaw LM, Trojanowski JQ, Peskind ER, Cruchaga C, Pericak-Vance MA, Goate AM, Cox NJ, Haines JL, Zetterberg H, Blennow K, Larson EB, Johnson SC, Albert M, for the Alzheimer's Disease Genetics Consortium and the Alzheimer Disease Neuroimaging Initiative, Bennett DA, Schneider JA, Jefferson AL, Hohman TJ. **Sex differences in the genetic predictors of Alzheimer's pathology.** *Brain*. 2019 Sep 1;142(9):2581-2589. doi: 10.1093/brain/awz206. PMID: 31497858.
132. Amlie-Wolf A, Tang M, Way J, Dombroski B, Jiang M, Vrettos N, Chou YF, Zhao Y, Kuzma A, Mlynarski EE, Leung YY, Brown CD, Wang LS, Schellenberg GD. **Inferring the molecular mechanisms of noncoding Alzheimer's disease-associated genetic variants.** *J Alzheimers Dis*. 2019;72(1):301-318. doi: 10.3233/JAD-190568. PMID: 31561366.
133. Baker E, Sims R, Leonenko G, Frizzati A, Harwood JC, Grozeva D; GERAD/PERADES; CHARGE; ADGC; EADI; IGAP consortia, Morgan K, Passmore P, Holmes C, Powell J, Brayne C, Gill M, Mead S, Bossù P, Spalletta G, Goate AM, Cruchaga C, Maier W, Heun R, Jessen F, Peters O, Dichgans M, Frölich L, Ramirez A, Jones L, Hardy J, Ivanov D, Hill M, Holmans P, Allen ND, Morgan BP, Seshadri S, Schellenberg GD, Amouyel P, Williams J, Escott-Price V. **Gene-based analysis in HRC imputed genome wide association data identifies three novel genes for Alzheimer's disease.** *PLoS One*. 2019 Jul 8;14(7):e0218111. doi: 10.1371/journal.pone.0218111. eCollection 2019. PMID: 31283791.
134. Ma Y, Jun GR, Zhang X, Chung J, Naj AC, Chen Y, Bellenguez C, Hamilton-Nelson K, Martin ER, Kunkle BW, Bis JC, Debette S, DeStefano AL, Fornage M, Nicolas G, van Duijn C, Bennett DA, De Jager PL, Mayeux R, Haines JL, Pericak-Vance MA, Seshadri S, Lambert JC, Schellenberg GD, Lunetta KL, Farrer LA; Alzheimer's Disease Sequencing Project and Alzheimer's Disease Exome Sequencing–France Project. **Analysis of Whole-Exome Sequencing Data for Alzheimer Disease Stratified by APOE Genotype.** *JAMA Neurol*. 2019 Jun 10. doi: 10.1001/jamaneurol.2019.1456. [Epub ahead of print] PMID: 31180460. PMCID: PMC6563544.
135. Gardner OK, Wang L, Van Booven D, Whitehead PL, Hamilton-Nelson KL, Adams LD, Starks TD, Hofmann NK, Vance JM, Cuccaro ML, Martin ER, Byrd GS, Haines JL, Bush WS, Beecham GW, Pericak-Vance MA, Griswold AJ. **RNA editing alterations in a multi-ethnic Alzheimer disease cohort converge on immune and endocytic molecular pathways.** *Hum Mol Genet*. 2019 Jun 4. pii: ddz110. doi: 10.1093/hmg/ddz110. [Epub ahead of print] PMID: 31162550.
136. Tosto G, Vardarajan B, Sariya S, Brickman AM, Andrews H, Manly JJ, Schupf N, Reyes-Dumeyer D, Lantigua R, Bennett DA, De Jager PL, Mayeux R. **Association of Variants in PINX1 and TREM2 With Late-Onset Alzheimer Disease.** *JAMA Neurol*. 2019 May 6. doi: 10.1001/jamaneurol.2019.1066. [Epub ahead of print] PubMed PMID: 31058951; PubMed Central PMCID: PMC6503572.
137. Sariya S, Lee JH, Mayeux R, Vardarajan BN, Reyes-Dumeyer D, Manly JJ, Brickman AM, Lantigua R, Medrano M, Jimenez-Velazquez IZ, Tosto G. **Rare Variants Imputation in Admixed Populations: Comparison Across Reference Panels and Bioinformatics Tools.** *Front Genet*. 2019 Apr 3;10:239. doi: 10.3389/fgene.2019.00239. eCollection 2019. PubMed PMID: 31001313; PubMed Central PMCID: PMC6456789.
138. Choi KY, Lee JJ, Gunasekaran TI, Kang S, Lee W, Jeong J, Lim HJ, Zhang X, Zhu C, Won SY, Choi YY, Seo EH, Lee SC, Gim J, Chung JY, Chong A, Byun MS, Seo S, Ko PW, Han JW, McLean C, Farrell J, Lunetta KL, Miyashita A, Hara N, Won S, Choi SM, Ha JM, Jeong JH, Kuwano R, Song MK, An SSA, Lee YM, Park KW, Lee HW, Choi SH, Rhee S, Song WK, Lee JS, Mayeux R, Haines JL, Pericak-Vance MA, Choo ILH, Nho K, Kim KW, Lee DY, Kim S, Kim BC, Kim H, Jun GR, Schellenberg GD, Ikeuchi T, Farrer LA, Lee KH, Neuroimaging Initiative AD. **APOE Promoter Polymorphism-219T/G is an Effect Modifier**

**of the Influence of APOE  $\epsilon$ 4 on Alzheimer's Disease Risk in a Multiracial Sample.** J Clin Med. 2019 Aug 16;8(8). pii: E1236. doi: 10.3390/jcm8081236. PMID: 31426376.

139. Cali CP, Patino M, Tai YK, Ho WY, McLean CA, Morris CM, Seeley WW, Miller BL, Gaig C, Vonsattel JPG, White CL 3rd, Roeber S, Kretzschmar H, Troncoso JC, Troakes C, Gearing M, Ghetti B, Van Deerlin VM, Lee VM, Trojanowski JQ, Mok KY, Ling H, Dickson DW, Schellenberg GD, Ling SC, Lee EB. **C9orf72 intermediate repeats are associated with corticobasal degeneration, increased C9orf72 expression and disruption of autophagy.** Acta Neuropathol. 2019 Nov;138(5):795-811. doi: 10.1007/s00401-019-02045-5. Epub 2019 Jul 20. PMID: 31327044.
140. Eric M. Reiman, Joseph F. Arboleda-Velasquez, Yakeel T. Quiroz, Matthew J. Huentelman, Thomas G. Beach, Richard J. Caselli, Yinghua Chen, Yi Su, Amanda J. Myers, John Hardy, Jean, Paul Vonsattel, Steven G. Younkin, David A. Bennett, Philip L. De Jager, Eric B. Larson, Paul K. Crane, C. Dirk Keene, M. Ilyas Kamboh, Julia K. Kofler, Linda Duque, John R. Gilbert, Harry E. Gwirtsman, Joseph D. Buxbaum, Dennis W. Dickson, Matthew P. Frosch, Bernardino Ghetti, Kathryn L. Lunetta, Li-San Wang, Bradley T. Hyman, Walter A. Kukull, Tatiana Foroud, Jonathan L. Haines, Richard P. Mayeux, Margaret A. Pericak-Vance, Julie A. Schneider, John Q. Trojanowski, Lindsay A. Farrer, Gerard D. Schellenberg, Gary W. Beecham, Thomas J. Montine, and Gyungah R. Jun for the Alzheimer's Disease Genetics Consortium. **Exceptionally low likelihood of Alzheimer's dementia in APOE2 homozygotes.** Nat Commun. 2020 Feb 3;11(1):667. doi: 10.1038/s41467-019-14279-8. PMID: 32015339.

#### Additional Publications Using ADGC Supported Studies

1. Cruchaga C, Kauwe JS, Nowotny P, Bales K, Pickering EH, Mayo K, Bertelsen S, Hinrichs A; Alzheimer's Disease Neuroimaging Initiative, Fagan AM, Holtzman DM, Morris JC, Goate AM. **Cerebrospinal fluid APOE levels: an endophenotype for genetic studies for Alzheimer's disease.** Hum Mol Genet. 2012 Oct 15;21(20):4558-71. Epub 2012 Jul 20. PMID: 22821396. PMCID: PMC3459471.
2. Leung YY, Chang CQ, Hung YS. **An integrated approach for identifying wrongly labelled samples when performing classification in microarray data.** PLoS One. 2012;7(10):e46700. doi: 10.1371/journal.pone.0046700. Epub 2012 Oct 17. PMID: 23082127. PMCID: PMC3474777.
3. Honea RA, Cruchaga C, Perea RD, Saykin AJ, Burns JM, Weinberger DR, Goate AM; Alzheimer's Disease Neuroimaging Initiative (ADNI). **Characterizing the role of brain derived neurotrophic factor genetic variation in Alzheimer's disease neurodegeneration.** PLoS One. 2013 Sep 26;8(9):e76001. doi: 10.1371/journal.pone.0076001. eCollection 2013. PMID: 24086677. PMCID: PMC3784423.
4. Ryvkin P, Leung YY, Silverman IM, Childress M, Valladares O, Dragomir I, Gregory BD, Wang LS. **HAMR: high-throughput annotation of modified ribonucleotides.** RNA. 2013 Dec;19(12):1684-92. doi: 10.1261/rna.036806.112. Epub 2013 Oct 22. PMID: 24149843. PMCID: PMC3884653.
5. Bennett DA, Yu L, De Jager PL. **Building a pipeline to discover and validate novel therapeutic targets and lead compounds for Alzheimer's disease.** Biochem Pharmacol. 2014 Apr 15;88(4):617-30. doi: 10.1016/j.bcp.2014.01.037. Epub 2014 Feb 6. PMID: 24508835. PMCID: PMC4054869.
6. Bailey JN, Pericak-Vance MA, Haines JL. **The impact of the human genome project on complex disease.** Genes (Basel). 2014 Jul 16;5(3):518-35. doi: 10.3390/genes5030518. PMID: 25032678. PMCID: PMC4198915.
7. Gross AL, Sherva R, Mukherjee S, Newhouse S, Kauwe JS, Munsie LM, Waterston LB, Bennett DA, Jones RN, Green RC, Crane PK; Alzheimer's Disease Neuroimaging Initiative; GENAROAD Consortium;

- AD Genetics Consortium. **Calibrating longitudinal cognition in Alzheimer's disease across diverse test batteries and datasets.** *Neuroepidemiology*. 2014;43(3-4):194-205. doi: 10.1159/000367970. Epub 2014 Nov 13. PMID: 25402421. PMCID: PMC4297570.
8. Nho K, Kim S, Risacher SL, Shen L, Corneveaux JJ, Swaminathan S, Lin H, Ramanan VK, Liu Y, Foroud TM, Inlow MH, Siniard AL, Reiman RA, Aisen PS, Petersen RC, Green RC, Jack CR Jr, Weiner MW, Baldwin CT, Lunetta KL, Farrer LA; MIRAGE (Multi-Institutional Research on Alzheimer Genetic Epidemiology) Study, Furney SJ, Lovestone S, Simmons A, Mecocci P, Vellas B, Tsolaki M, Kloszewska I, Soininen H; AddNeuroMed Consortium, McDonald BC, Farlow MR, Ghetti B; Indiana Memory and Aging Study, Huentelman MJ, Saykin AJ; Alzheimer's Disease Neuroimaging Initiative. **Protective variant for hippocampal atrophy identified by whole exome sequencing.** *Ann Neurol*. 2015 Mar;77(3):547-52. doi: 10.1002/ana.24349. Epub 2015 Feb 14. PMID: 25559091. PMCID: PMC4387567.
  9. Deming Y, Xia J, Cai Y, Lord J, Holmans P, Bertelsen S, Holtzman D, Morris JC, Bales K, Pickering EH, Kauwe J, Goate A, Cruchaga C; Alzheimer's Disease Neuroimaging Initiative (ADNI). **A potential endophenotype for Alzheimer's disease: cerebrospinal fluid clusterin.** *Neurobiol Aging*. 2016 Jan;37:208.e1-208.e9. doi: 10.1016/j.neurobiolaging.2015.09.009. Epub 2015 Sep 25. PMID: 26545630. PMCID: PMC5118651.
  10. Leung YY, Kuksa PP, Amlie-Wolf A, Valladares O, Ungar LH, Kannan S, Gregory BD, Wang LS. **DASHR: database of small human noncoding RNAs.** *Nucleic Acids Res*. 2016 Jan 4;44(D1):D216-22. doi: 10.1093/nar/gkv1188. Epub 2015 Nov 8. PMID: 26553799. PMCID: PMC4702848.
  11. LoBue C, Denney D, Hynan LS, Rossetti HC, Lacritz LH, Hart J, Womack KB, Woon FL, Cullum CM. **Self-Reported Traumatic Brain Injury and Mild Cognitive Impairment: Increased Risk and Earlier Age of Diagnosis.** *J Alzheimers Dis*. 2016;51(3):727-36. doi: 10.3233/JAD-150895. PMID: 26890760. PMCID: PMC4853649.
  12. Karch CM, Ezerskiy LA, Bertelsen S; Alzheimer's Disease Genetics Consortium (ADGC), Goate AM. **Alzheimer's Disease Risk Polymorphisms Regulate Gene Expression in the ZCWPW1 and the CELF1 Loci.** *PLoS One*. 2016 Feb 26;11(2):e0148717. doi: 10.1371/journal.pone.0148717. eCollection 2016. PMID: 26919393. PMCID: PMC4769299.
  13. Allen GI, Amoroso N, Anghel C, Balagurusamy V, Bare CJ, Beaton D, Bellotti R, Bennett DA, Boehme KL, Boutros PC, Caberlotto L, Caloian C, Campbell F, Chaibub Neto E, Chang YC, Chen B, Chen CY, Chien TY, Clark T, Das S, Davatzikos C, Deng J, Dillenberger D, Dobson RJ, Dong Q, Doshi J, Duma D, Errico R, Erus G, Everett E, Fardo DW, Friend SH, Fröhlich H, Gan J, St George-Hyslop P, Ghosh SS, Glaab E, Green RC, Guan Y, Hong MY, Huang C, Hwang J, Ibrahim J, Inglese P, Iyappan A, Jiang Q, Katsumata Y, Kauwe JS, Klein A, Kong D, Krause R, Lalonde E, Lauria M, Lee E, Lin X, Liu Z, Livingstone J, Logsdon BA, Lovestone S, Ma TW, Malhotra A, Mangravite LM, Maxwell TJ, Merrill E, Nagorski J, Namasivayam A, Narayan M, Naz M, Newhouse SJ, Norman TC, Nurtdinov RN, Oyang YJ, Pawitan Y, Peng S, Peters MA, Piccolo SR, Praveen P, Priami C, Sabelnykova VY, Senger P, Shen X, Simmons A, Sotiras A, Stolovitzky G, Tangaro S, Tateo A, Tung YA, Tustison NJ, Varol E, Vradenburg G, Weiner MW, Xiao G, Xie L, Xie Y, Xu J, Yang H, Zhan X, Zhou Y, Zhu F, Zhu H, Zhu S; Alzheimer's Disease Neuroimaging Initiative. **Crowdsourced estimation of cognitive decline and resilience in Alzheimer's disease.** *Alzheimers Dement*. 2016 Jun;12(6):645-53. doi: 10.1016/j.jalz.2016.02.006. Epub 2016 Apr 11. PMID: 27079753. PMCID: PMC5474755.
  14. Scott RA, Freitag DF, Li L, Chu AY, Surendran P, Young R, Grarup N, Stancáková A, Chen Y, Varga TV, Yaghootkar H, Luan J, Zhao JH, Willems SM, Wessel J, Wang S, Maruthur N, Michailidou K, Pirie A, van der Lee SJ, Gillson C, Al Olama AA, Amouyel P, Arriola L, Arveiler D, Aviles-Olmos I, Balkau B, Barricarte

- A, Barroso I, Garcia SB, Bis JC, Blankenberg S, Boehnke M, Boeing H, Boerwinkle E, Borecki IB, Bork-Jensen J, Bowden S, Caldas C, Caslake M; CVD50 consortium, Cupples LA, Cruchaga C, Czajkowski J, den Hoed M, Dunn JA, Earl HM, Ehret GB, Ferrannini E, Ferrieres J, Foltynie T, Ford I, Forouhi NG, Gianfagna F, Gonzalez C, Grioni S, Hiller L, Jansson JH, Jørgensen ME, Jukema JW, Kaaks R, Kee F, Kerrison ND, Key TJ, Kontto J, Kote-Jarai Z, Kraja AT, Kuulasmaa K, Kuusisto J, Linneberg A, Liu C, Marenne G, Mohlke KL, Morris AP, Muir K, Müller-Nurasyid M, Munroe PB, Navarro C, Nielsen SF, Nilsson PM, Nordestgaard BG, Packard CJ, Palli D, Panico S, Peloso GM, Perola M, Peters A, Poole CJ, Quirós JR, Rolandsson O, Sacerdote C, Salomaa V, Sánchez MJ, Sattar N, Sharp SJ, Sims R, Slimani N, Smith JA, Thompson DJ, Trompet S, Tumino R, van der A DL, van der Schouw YT, Virtamo J, Walker M, Walter K; GERAD\_EC Consortium; Neurology Working Group of the Cohorts for Heart; Aging Research in Genomic Epidemiology (CHARGE); Alzheimer's Disease Genetics Consortium; Pancreatic Cancer Cohort Consortium; European Prospective Investigation into Cancer and Nutrition–Cardiovascular Disease (EPIC-CVD); EPIC-InterAct, Abraham JE, Amundadottir LT, Aponte JL, Butterworth AS, Dupuis J, Easton DF, Eeles RA, Erdmann J, Franks PW, Frayling TM, Hansen T, Howson JM, Jørgensen T, Kooner J, Laakso M, Langenberg C, McCarthy MI, Pankow JS, Pedersen O, Riboli E, Rotter JI, Saleheen D, Samani NJ, Schunkert H, Vollenweider P, O'Rahilly S; CHARGE consortium; CHD Exome+ Consortium; CARDIOGRAM Exome Consortium, Deloukas P, Danesh J, Goodarzi MO, Kathiresan S, Meigs JB, Ehm MG, Wareham NJ, Waterworth DM. **A genomic approach to therapeutic target validation identifies a glucose-lowering GLP1R variant protective for coronary heart disease.** *Sci Transl Med.* 2016 Jun 1;8(341):341ra76. doi: 10.1126/scitranslmed.aad3744. PMID: 27252175. PMCID: PMC5219001.
15. Staley LA, Ebbert MT, Parker S, Bailey M; Alzheimer's Disease Neuroimaging Initiative, Ridge PG, Goate AM, Kauwe JS. **Genome-wide association study of prolactin levels in blood plasma and cerebrospinal fluid.** *BMC Genomics.* 2016 Jun 29;17 Suppl 3:436. doi: 10.1186/s12864-016-2785-0. PMID: 27357110. PMCID: PMC4943503.
16. Staley LA, Ebbert MT, Bunker D, Bailey M; Alzheimer's Disease Neuroimaging Initiative, Ridge PG, Goate AM, Kauwe JS. **Variants in ACPP are associated with cerebrospinal fluid Prostatic Acid Phosphatase levels.** *BMC Genomics.* 2016 Jun 29;17 Suppl 3:439. doi: 10.1186/s12864-016-2787-y. PMID: 27357282. PMCID: PMC4943489.
17. Ebbert MT, Staley LA, Parker J, Parker S, Bailey M; Alzheimer's Disease Neuroimaging Initiative, Ridge PG, Goate AM, Kauwe JS. **Variants in CCL16 are associated with blood plasma and cerebrospinal fluid CCL16 protein levels.** *BMC Genomics.* 2016 Jun 29;17 Suppl 3:437. doi: 10.1186/s12864-016-2788-x. PMID: 27357396. PMCID: PMC4943476.
18. Kenna KP, van Doormaal PT, Dekker AM, Ticozzi N, Kenna BJ, Diekstra FP, van Rheenen W, van Eijk KR, Jones AR, Keagle P, Shatunov A, Sproviero W, Smith BN, van Es MA, Topp SD, Kenna A, Miller JW, Fallini C, Tiloca C, McLaughlin RL, Vance C, Troakes C, Colombrita C, Mora G, Calvo A, Verde F, Al-Sarraj S, King A, Calini D, de Bellerocche J, Baas F, van der Kooij AJ, de Visser M, Ten Asbroek AL, Sapp PC, McKenna-Yasek D, Polak M, Asress S, Muñoz-Blanco JL, Strom TM, Meitinger T, Morrison KE; SLAGEN Consortium, Lauria G, Williams KL, Leigh PN, Nicholson GA, Blair IP, Leblond CS, Dion PA, Rouleau GA, Pall H, Shaw PJ, Turner MR, Talbot K, Taroni F, Boylan KB, Van Blitterswijk M, Rademakers R, Esteban-Pérez J, García-Redondo A, Van Damme P, Robberecht W, Chio A, Gellera C, Drepper C, Sendtner M, Ratti A, Glass JD, Mora JS, Basak NA, Hardiman O, Ludolph AC, Andersen PM, Weishaupt JH, Brown RH Jr, Al-Chalabi A, Silani V, Shaw CE, van den Berg LH, Veldink JH, Landers JE. **NEK1 variants confer susceptibility to amyotrophic lateral sclerosis.** *Nat Genet.* 2016 Sep;48(9):1037-42. doi: 10.1038/ng.3626. Epub 2016 Jul 25. PMID: 27455347. PMCID: PMC5560030.
19. Deming Y, Black K, Carrell D, Cai Y, Del-Aguila JL, Fernandez MV, Budde J, Ma S, Saef B, Howells B, Bertelsen S, Huang KL, Sutphen CL, Tarawneh R, Fagan AM, Holtzman DM, Morris JC, Goate AM,

- Dougherty JD, Cruchaga C. **Chitinase-3-like 1 protein (CHI3L1) locus influences cerebrospinal fluid levels of YKL-40.** BMC Neurol. 2016 Nov 10;16(1):217. PMID: 27832767. PMCID: PMC5105244.
20. LoBue C, Wadsworth H, Wilmoth K, Clem M, Hart J Jr, Womack KB, Didehbani N, Lacritz LH, Rossetti HC, Cullum CM. **Traumatic brain injury history is associated with earlier age of onset of Alzheimer disease.** Clin Neuropsychol. 2017 Jan;31(1):85-98. doi: 10.1080/13854046.2016.1257069. Epub 2016 Nov 18. PMID: 27855547. PMCID: PMC5224912.
  21. Sennik S, Schweizer TA, Fischer CE, Munoz DG. **Risk Factors and Pathological Substrates Associated with Agitation/Aggression in Alzheimer's Disease: A Preliminary Study using NACC Data.** J Alzheimers Dis. 2017;55(4):1519-1528. doi: 10.3233/JAD-160780. PMID: 27911311. PMCID: PMC5607738.
  22. Rehker J, Rodhe J, Nesbitt RR, Boyle EA, Martin BK, Lord J, Karaca I, Naj A, Jessen F, Helisalmi S, Soininen H, Hiltunen M, Ramirez A, Scherer M, Farrer LA, Haines JL, Pericak-Vance MA, Raskind WH, Cruchaga C, Schellenberg GD, Joseph B, Brkanac Z. **Caspase-8, association with Alzheimer's Disease and functional analysis of rare variants.** PLoS One. 2017 Oct 6;12(10):e0185777. doi: 10.1371/journal.pone.0185777. eCollection 2017. PMID: 28985224. PMCID: PMC5630132.
  23. Larsson SC, Markus HS. **Branched-chain amino acids and Alzheimer's disease: a Mendelian randomization analysis.** Sci Rep. 2017 Oct 19;7(1):13604. doi: 10.1038/s41598-017-12931-1. PMID: 29051501. PMCID: PMC5648806.
  24. Miller JA, Guillozet-Bongaarts A, Gibbons LE, Postupna N, Renz A, Beller AE, Sunkin SM, Ng L, Rose SE, Smith KA, Szafer A, Barber C, Bertagnolli D, Bickley K, Brouner K, Caldejon S, Chapin M, Chua ML, Coleman NM, Cudaback E, Cuhaciyani C, Dalley RA, Dee N, Desta T, Dolbeare TA, Dotson NI, Fisher M, Gaudreault N, Gee G, Gilbert TL, Goldy J, Griffin F, Habel C, Haradon Z, Hejazinia N, Hellstern LL, Horvath S, Howard K, Howard R, Johal J, Jorstad NL, Josephsen SR, Kuan CL, Lai F, Lee E, Lee F, Lemon T, Li X, Marshall DA, Melchor J, Mukherjee S, Nyhus J, Pendergraft J, Potekhina L, Rha EY, Rice S, Rosen D, Sapru A, Schantz A, Shen E, Sherfield E, Shi S, Sodt AJ, Thatra N, Tieu M, Wilson AM, Montine TJ, Larson EB, Bernard A, Crane PK, Ellenbogen RG, Keene CD, Lein E. **Neuropathological and transcriptomic characteristics of the aged brain.** Elife. 2017 Nov 9;6. pii: e31126. doi: 10.7554/eLife.31126. PMID: 29120328. PMCID: PMC5679757.
  25. Bennett RE, Robbins AB, Hu M, Cao X, Betensky RA, Clark T, Das S, Hyman BT. **Tau induces blood vessel abnormalities and angiogenesis-related gene expression in P301L transgenic mice and human Alzheimer's disease.** Proc Natl Acad Sci U S A. 2018 Feb 6;115(6):E1289-E1298. doi: 10.1073/pnas.1710329115. Epub 2018 Jan 22. PMID: 29358399. PMCID: PMC5819390.
  26. Zhou X, Chen Y, Mok KY, Zhao Q, Chen K, Chen Y, Hardy J, Li Y, Fu AKY, Guo Q, Ip NY; Alzheimer's Disease Neuroimaging Initiative. **Identification of genetic risk factors in the Chinese population implicates a role of immune system in Alzheimer's disease pathogenesis.** Proc Natl Acad Sci U S A. 2018 Feb 20;115(8):1697-1706. doi: 10.1073/pnas.1715554115. Epub 2018 Feb 5. PMID: 29432188. PMCID: PMC5828602.
  27. Ikezu T, Chen C, DeLeo AM, Zeldich E, Fallin MD, Kanaan NM, Lunetta KL, Abraham CR, Logue MW, Farrer LA. **Tau Phosphorylation is Impacted by Rare AKAP9 Mutations Associated with Alzheimer Disease in African Americans.** J Neuroimmune Pharmacol. 2018 Jun;13(2):254-264. doi: 10.1007/s11481-018-9781-x. Epub 2018 Mar 7. PMID: 29516269. PMCID: PMC5928172.

28. Dobbyn A, Huckins LM, Boocock J, Sloofman LG, Glicksberg BS, Giambartolomei C, Hoffman GE, Perumal TM, Girdhar K, Jiang Y, Raj T, Ruderfer DM, Kramer RS, Pinto D; CommonMind Consortium, Akbarian S, Roussos P, Domenici E, Devlin B, Sklar P, Stahl EA, Sieberts SK. **Landscape of Conditional eQTL in Dorsolateral Prefrontal Cortex and Co-localization with Schizophrenia GWAS.** *Am J Hum Genet.* 2018 Jun 7;102(6):1169-1184. doi: 10.1016/j.ajhg.2018.04.011. Epub 2018 May 24. PMID: 29805045. PMCID: PMC5993513.
29. Mukherjee S, Mez J, Trittschuh EH, Saykin AJ, Gibbons LE, Fardo DW, Wessels M, Bauman J, Moore M, Choi SE, Gross AL, Rich J, Loudon DKN, Sanders RE, Grabowski TJ, Bird TD, McCurry SM, Snitz BE, Kamboh MI, Lopez OL, De Jager PL, Bennett DA, Keene CD, Larson EB; EPAD Study Group; Investigators from ACT; Investigators from ROS; Investigators from MAP; Investigators from ADNI; Investigators from the University of Pittsburgh ADRC, Crane PK. **Genetic data and cognitively defined late-onset Alzheimer's disease subgroups.** *Mol Psychiatry.* 2018 Dec 4. doi: 10.1038/s41380-018-0298-8. PMID: 30514930. PMCID: PMC6548676.
30. Pathak GA, Zhou Z, Silzer TK, Barber RC, Phillips NR; Alzheimer's Disease Neuroimaging Initiative, Breast and Prostate Cancer Cohort Consortium, and Alzheimer's Disease Genetics Consortium. **Two-stage Bayesian GWAS of 9576 individuals identifies SNP regions that are targeted by miRNAs inversely expressed in Alzheimer's and cancer.** *Alzheimers Dement.* 2020 Jan;16(1):162-177. doi: 10.1002/alz.12003. PMID: 31914222.
31. Ji Z, Liao Y, Zheng L, Wu L, Yu M, Feng Y. **An Assembled Detector Based on Geometrical Constraint for Power Component Recognition.** *Sensors (Basel).* 2019 Aug 11;19(16). pii: E3517. doi: 10.3390/s19163517. PMID: 31405244.

#### Additional Publications Using IGAP Summary Statistics

1. Mormino EC, Sperling RA, Holmes AJ, Buckner RL, De Jager PL, Smoller JW, Sabuncu MR; Alzheimer's Disease Neuroimaging Initiative. **Polygenic risk of Alzheimer disease is associated with early- and late-life processes.** *Neurology.* 2016 Aug 2;87(5):481-8. PMID: 27385740. PMCID: PMC4970660.
2. Zhang DF, Fan Y, Wang D, Bi R, Zhang C, Fang Y, Yao YG. **PLD3 in Alzheimer's Disease: a Modest Effect as Revealed by Updated Association and Expression Analyses.** *Mol Neurobiol.* 2016 Aug;53(6):4034-4045. PMID: 26189833. PMCID: PMC26189833.
3. Heck A, Fastenrath M, Coynel D, Auschra B, Bickel H, Freytag V, Gschwind L, Hartmann F, Jessen F, Kaduszkiewicz H, Maier W, Milnik A, Pentzek M, Riedel-Heller SG, Spalek K, Vogler C, Wagner M, Weyerer S, Wolfsgruber S, de Quervain DJ, Papassotiropoulos A. **Genetic Analysis of Association Between Calcium Signaling and Hippocampal Activation, Memory Performance in the Young and Old, and Risk for Sporadic Alzheimer Disease.** *JAMA Psychiatry.* 2015 Oct;72(10):1029-36. PMID: 26332608. PMCID: PMC5291164.
4. Srinivasan S, Bettella F, Mattingsdal M, Wang Y, Witoelar A, Schork AJ, Thompson WK, Zuber V; Schizophrenia Working Group of the Psychiatric Genomics Consortium, The International Headache Genetics Consortium, Winsvold BS, Zwart JA, Collier DA, Desikan RS, Melle I, Werge T, Dale AM, Djurovic S, Andreassen OA. **Genetic Markers of Human Evolution Are Enriched in Schizophrenia.** *Biol Psychiatry.* 2016 Aug 15;80(4):284-292. PMID: 26681495. PMCID: PMC5397584.
5. Lupton MK, Strike L, Hansell NK, Wen W, Mather KA, Armstrong NJ, Thalamuthu A, McMahon KL, de Zubicaray GI, Assareh AA, Simmons A, Proitsi P, Powell JF, Montgomery GW, Hibar DP, Westman E, Tsolaki M, Kloszewska I, Soininen H, Mecocci P, Velas B, Lovestone S; Alzheimer's Disease

- Neuroimaging Initiative, Brodaty H, Ames D, Trollor JN, Martin NG, Thompson PM, Sachdev PS, Wright MJ. **The effect of increased genetic risk for Alzheimer's disease on hippocampal and amygdala volume.** *Neurobiol Aging*. 2016 Apr;40:68-77. PMID: 26973105. PMCID: PMC4883003.
6. Allen M, Kachadoorian M, Carrasquillo MM, Karhade A, Manly L, Burgess JD, Wang C, Serie D, Wang X, Siuda J, Zou F, Chai HS, Younkin C, Crook J, Medway C, Nguyen T, Ma L, Malphrus K, Lincoln S, Petersen RC, Graff-Radford NR, Asmann YW, Dickson DW, Younkin SG, Ertekin-Taner N. **Late-onset Alzheimer disease risk variants mark brain regulatory loci.** *Neurol Genet*. 2015 Jul 23;1(2):e15. PMID: 27066552. PMCID: PMC4807909.
  7. Fryland T, Christensen JH, Pallesen J, Mattheisen M, Palmfeldt J, Bak M, Grove J, Demontis D, Blechinger J, Ooi HS, Nyegaard M, Hauberg ME, Tommerup N, Gregersen N, Mors O, Corydon TJ, Nielsen AL, Børglum AD. **Identification of the BRD1 interaction network and its impact on mental disorder risk.** *Genome Med*. 2016 May 3;8(1):53. PMID: 27142060. PMCID: PMC4855718.
  8. Mokry LE, Ross S, Morris JA, Manousaki D, Forgetta V, Richards JB. **Genetically decreased vitamin D and risk of Alzheimer disease.** *Neurology*. 2016 Dec 13;87(24):2567-2574. PMID: 27856775. PMCID: PMC5207000.
  9. Pickrell JK, Berisa T, Liu JZ, Séguirel L, Tung JY, Hinds DA. **Detection and interpretation of shared genetic influences on 42 human traits.** *Nat Genet*. 2016 Jul;48(7):709-17. Epub 2016 May 16. Erratum in: *Nat Genet*. 2016 Sep 28;48(10):1296. PMID: 27182965. PMCID: PMC5207801.
  10. Tosto G, Reitz C. **Genomics of Alzheimer's disease: Value of high-throughput genomic technologies to dissect its etiology.** *Mol Cell Probes*. 2016 Dec;30(6):397-403. Epub 2016 Sep 13. Review. PMID: 27618776. DOI: 10.1016/j.mcp.2016.09.001.
  11. Adams HH, Hibar DP, Chouraki V, Stein JL, Nyquist PA, Rentería ME, Trompet S, Arias-Vasquez A, Seshadri S, Desrivières S, Beecham AH, Jahanshad N, Wittfeld K, Van der Lee SJ, Abramovic L, Alhusaini S, Amin N, Andersson M, Arfanakis K, Aribisala BS, Armstrong NJ, Athanasiu L, Axelsson T, Beiser A, Bernard M, Bis JC, Blanken LM, Blanton SH, Bohlken MM, Boks MP, Bralten J, Brickman AM, Carmichael O, Chakravarty MM, Chauhan G, Chen Q, Ching CR, Cuellar-Partida G, Braber AD, Doan NT, Ehrlich S, Filippi I, Ge T, Giddaluru S, Goldman AL, Gottesman RF, Greven CU, Grimm O, Griswold ME, Guadalupe T, Hass J, Haukvik UK, Hilal S, Hofer E, Hoehn D, Holmes AJ, Hoogman M, Janowitz D, Jia T, Kasperaviciute D, Kim S, Klein M, Kraemer B, Lee PH, Liao J, Liewald DC, Lopez LM, Luciano M, Macare C, Marquand A, Matarin M, Mather KA, Mattheisen M, Mazoyer B, McKay DR, McWhirter R, Milaneschi Y, Mirza-Schreiber N, Muetzel RL, Maniega SM, Nho K, Nugent AC, Loohuis LM, Oosterlaan J, Pappmeyer M, Pappa I, Pirpamer L, Pudas S, Pütz B, Rajan KB, Ramasamy A, Richards JS, Risacher SL, Roiz-Santiañez R, Rommelse N, Rose EJ, Royle NA, Rundek T, Sämann PG, Satizabal CL, Schmaal L, Schork AJ, Shen L, Shin J, Shumskaya E, Smith AV, Sprooten E, Strike LT, Teumer A, Thomson R, Tordesillas-Gutierrez D, Toro R, Trabzuni D, Vaidya D, Van der Grond J, Van der Meer D, Van Donkelaar MM, Van Eijk KR, Van Erp TG, Van Rooij D, Walton E, Westlye LT, Whelan CD, Windham BG, Winkler AM, Woldehawariat G, Wolf C, Wolfers T, Xu B, Yanek LR, Yang J, Zijdenbos A, Zwiens MP, Agartz I, Aggarwal NT, Almasry L, Ames D, Amouyel P, Andreassen OA, Arepalli S, Assareh AA, Barral S, Bastin ME, Becker DM, Becker JT, Bennett DA, Blangero J, van Bokhoven H, Boomsma DI, Brodaty H, Brouwer RM, Brunner HG, Buckner RL, Buitelaar JK, Bulayeva KB, Cahn W, Calhoun VD, Cannon DM, Cavalleri GL, Chen C, Cheng CY, Cichon S, Cookson MR, Corvin A, Crespo-Facorro B, Curran JE, Czisch M, Dale AM, Davies GE, De Geus EJ, De Jager PL, de Zubicaray GI, Delanty N, Depondt C, DeStefano AL, Dillman A, Djurovic S, Donohoe G, Drevets WC, Duggirala R, Dyer TD, Erk S, Espeseth T, Evans DA, Fedko IO, Fernández G, Ferrucci L, Fisher SE, Fleischman DA, Ford I, Foroud TM, Fox PT, Francks C, Fukunaga M, Gibbs JR, Glahn DC, Gollub RL, Göring HH, Grabe HJ, Green RC, Gruber O, Gudnason V, Guelfi S, Hansell NK, Hardy J,



Hartman CA, Hashimoto R, Hegenscheid K, Heinz A, Le Hellard S, Hernandez DG, Heslenfeld DJ, Ho BC, Hoekstra PJ, Hoffmann W, Hofman A, Holsboer F, Homuth G, Hosten N, Hottenga JJ, Hulshoff Pol HE, Ikeda M, Ikram MK, Jack CR Jr, Jenkinson M, Johnson R, Jönsson EG, Jukema JW, Kahn RS, Kanai R, Kloszewska I, Knopman DS, Kochunov P, Kwok JB, Lawrie SM, Lemaître H, Liu X, Longo DL, Longstreth WT Jr, Lopez OL, Lovestone S, Martinez O, Martinot JL, Mattay VS, McDonald C, McIntosh AM, McMahon KL, McMahon FJ, Mecocci P, Melle I, Meyer-Lindenberg A, Mohnke S, Montgomery GW, Morris DW, Mosley TH, Mühleisen TW, Müller-Myhsok B, Nalls MA, Nauck M, Nichols TE, Niessen WJ, Nöthen MM, Nyberg L, Ohi K, Olvera RL, Ophoff RA, Pandolfo M, Paus T, Pausova Z, Penninx BW, Pike GB, Potkin SG, Psaty BM, Reppermund S, Rietschel M, Roffman JL, Romanczuk-Seiferth N, Rotter JI, Ryten M, Sacco RL, Sachdev PS, Saykin AJ, Schmidt R, Schofield PR, Sigurdsson S, Simmons A, Singleton A, Sisodiya SM, Smith C, Smoller JW, Soininen H, Srikanth V, Steen VM, Stott DJ, Susmann JE, Thalamuthu A, Tiemeier H, Toga AW, Traynor BJ, Troncoso J, Turner JA, Tzourio C, Uitterlinden AG, Hernández MC, Van der Brug M, Van der Lugt A, Van der Wee NJ, Van Duijn CM, Van Haren NE, Van T Ent D, Van Tol MJ, Vardarajan BN, Veltman DJ, Vernooij MW, Völzke H, Walter H, Wardlaw JM, Wassink TH, Weale ME, Weinberger DR, Weiner MW, Wen W, Westman E, White T, Wong TY, Wright CB, Zielke HR, Zonderman AB, Deary IJ, DeCarli C, Schmidt H, Martin NG, De Craen AJ, Wright MJ, Launer LJ, Schumann G, Fornage M, Franke B, Dobbie S, Medland SE, Ikram MA, Thompson PM. **Novel genetic loci underlying human intracranial volume identified through genome-wide association.** *Nat Neurosci.* 2016 Dec;19(12):1569-1582. PMID: 27694991. PMCID: PMC5227112.

12. Lacour A, Espinosa A, Louwersheimer E, Heilmann S, Hernández I, Wolfsgruber S, Fernández V, Wagner H, Rosende-Roca M, Mauleón A, Moreno-Grau S, Vargas L, Pijnenburg YA, Koene T, Rodríguez-Gómez O, Ortega G, Ruiz S, Holstege H, Sotolongo-Grau O, Kornhuber J, Peters O, Frölich L, Hüll M, Rütger E, Wilfang J, Scherer M, Riedel-Heller S, Alegret M, Nöthen MM, Scheltens P, Wagner M, Tárraga L, Jessen F, Boada M, Maier W, van der Flier WM, Becker T, Ramirez A, Ruiz A. **Genome-wide significant risk factors for Alzheimer's disease: role in progression to dementia due to Alzheimer's disease among subjects with mild cognitive impairment.** *Mol Psychiatry.* 2017 Jan;22(1):153-160. PMID: 26976043. PMCID: PMC5414086.
13. Qvist P, Christensen JH, Vardya I, Rajkumar AP, Mørk A, Paternoster V, Füchtbauer EM, Pallesen J, Fryland T, Dyrvig M, Hauberg ME, Lundsberg B, Fejgin K, Nyegaard M, Jensen K, Nyengaard JR, Mors O, Didriksen M, Børghlum AD. **The Schizophrenia-Associated BRD1 Gene Regulates Behavior, Neurotransmission, and Expression of Schizophrenia Risk Enriched Gene Sets in Mice.** *Biol Psychiatry.* 2017 Jul 1;82(1):62-76. PMID: 27837920. DOI: 10.1016/j.biopsych.2016.08.037.
14. Morgan AR, Touchard S, O'Hagan C, Sims R, Majounie E, Escott-Price V, Jones L, Williams J, Morgan BP. **The Correlation between Inflammatory Biomarkers and Polygenic Risk Score in Alzheimer's Disease.** *J Alzheimers Dis.* 2017;56(1):25-36. PMID: 27911318. DOI: 10.3233/JAD-160889.
15. McLaughlin RL, Schijven D, van Rheenen W, van Eijk KR, O'Brien M, Kahn RS, Ophoff RA, Goris A, Bradley DG, Al-Chalabi A, van den Berg LH, Luykx JJ, Hardiman O, Veldink JH; Project MinE GWAS Consortium; Schizophrenia Working Group of the Psychiatric Genomics Consortium. **Genetic correlation between amyotrophic lateral sclerosis and schizophrenia.** *Nat Commun.* 2017 Mar 21;8:14774. PMID: 28322246. PMCID: PMC5364411.
16. Karlsson IK, Ploner A, Song C, Gatz M, Pedersen NL, Hägg S. **Genetic susceptibility to cardiovascular disease and risk of dementia.** *Transl Psychiatry.* 2017 May 30;7(5):e1142. PMID: 28556832. PMCID: PMC5534941.
17. Myrum C, Nikolaienko O, Bramham CR, Haavik J, Zayats T. **Implication of the APP Gene in Intellectual Abilities.** *J Alzheimers Dis.* 2017;59(2):723-735. PMID: 28671113. PMCID: PMC5523840.

18. Larsson SC, Traylor M, Burgess S, Markus HS. **Genetically-Predicted Adult Height and Alzheimer's Disease.** *J Alzheimers Dis.* 2017;60(2):691-698. PMID: 28869480. PMCID: PMC6037293.
19. Hinney A, Albayrak O, Antel J, Volckmar AL, Sims R, Chapman J, Harold D, Gerrish A, Heid IM, Winkler TW, Scherag A, Wiltfang J, Williams J, Hebebrand J; GERAD Consortium; IGAP Consortium; GIANT Consortium. **Genetic variation at the CELF1 (CUGBP, elav-like family member 1 gene) locus is genome-wide associated with Alzheimer's disease and obesity.** *Am J Med Genet B Neuropsychiatr Genet.* 2014 Jun;165B(4):283-93. PMID: 24788522. PMCID: PMC4879683.
20. Walter S, Marden JR, Kubzansky LD, Mayeda ER, Crane PK, Chang SC, Cornelis M, Rehkopf DH, Mukherjee S, Glymour MM. **Diabetic Phenotypes and Late-Life Dementia Risk: A Mechanism-specific Mendelian Randomization Study.** *Alzheimer Dis Assoc Disord.* 2016 Jan-Mar;30(1):15-20. PMID: 26650880. PMCID: PMC4879683.
21. Traylor M, Adib-Samii P, Harold D; Alzheimer's Disease Neuroimaging Initiative; International Stroke Genetics Consortium (ISGC), UK Young Lacunar Stroke DNA resource, Dichgans M, Williams J, Lewis CM, Markus HS; METASTROKE; International Genomics of Alzheimer's Project (IGAP), investigators. **Shared genetic contribution to Ischaemic Stroke and Alzheimer's Disease.** *Ann Neurol.* 2016 Feb 23. PMID: 26913989. PMCID: PMC4864940.
22. Chouraki V, Reitz C, Maury F, Bis JC, Bellenguez C, Yu L, Jakobsdottir J, Mukherjee S, Adams HH, Choi SH, Larson EB, Fitzpatrick A, Uitterlinden AG, de Jager PL, Hofman A, Gudnason V, Vardarajan B, Ibrahim-Verbaas C, van der Lee SJ, Lopez O, Dartigues JF, Berr C, Amouyel P, Bennett DA, van Duijn C, DeStefano AL, Launer LJ, Ikram MA, Crane PK, Lambert JC, Mayeux R, Seshadri S; International Genomics of Alzheimer's Project. **Evaluation of a Genetic Risk Score to Improved Risk Prediction for Alzheimer's Disease.** *J Alzheimers Dis.* 2016 Jun 18;53(3):921-32. PMID: 27340842. PMCID: PMC5036102.
23. Carrasquillo MM, Allen M, Burgess JD, Wang X, Strickland SL, Aryal S, Siuda J, Kachadoorian ML, Medway C, Younkin CS, Nair A, Wang C, Chanana P, Serie D, Nguyen T, Lincoln S, Malphrus KG, Morgan K, Golde TE, Price ND, White CC, De Jager PL, Bennett DA, Asmann YW, Crook JE, Petersen RC, Graff-Radford NR, Dickson DW, Younkin SG, Ertekin-Taner N. **A candidate regulatory variant at the TREM gene cluster associates with decreased Alzheimer's disease risk and increased TREM1 and TREM2 brain gene expression.** *Alzheimers Dement.* 2017 Jun;13(6):663-673. PMID: 27939925. PMCID: PMC5462884.
24. Benn M, Nordestgaard BG, Frikke-Schmidt R, Tybjaerg-Hansen A. **Low LDL cholesterol, PCSK9 and HMGCR genetic variation, and risk of Alzheimer's disease and Parkinson's disease: Mendelian randomisation study.** *BMJ.* 2017 Apr 24;357:j1648. Erratum in: *BMJ.* 2017 Jun 29;357:j3170. PMID: 28438747. PMCID: PMC5421439.
25. Gibson J, Russ TC, Adams MJ, Clarke TK, Howard DM, Hall LS, Fernandez-Pujals AM, Wigmore EM, Hayward C, Davies G, Murray AD, Smith BH, Porteous DJ, Deary IJ, McIntosh AM. **Assessing the presence of shared genetic architecture between Alzheimer's disease and major depressive disorder using genome-wide association data.** *Transl Psychiatry.* 2017 Apr 18;7(4). PMID: 28418403. PMCID: PMC5416691.

26. Nordestgaard LT, Tybjaerg-Hansen A, Nordestgaard BG, Frikke-Schmidt R. **Body Mass Index and Risk of Alzheimer's Disease: A Mendelian Randomization Study of 399,536 Individuals.** J Clin Endocrinol Metab. 2017 Jul 1;102(7):2310-2320. PMID: 28609829. PMCID: PMC5505195.
27. Feng YA, Cho K, Lindstrom S, Kraft P, Cormack J; IGAP Consortium, Colorectal Transdisciplinary Study (CORECT); Discovery, Biology, and Risk of Inherited Variants in Breast Cancer (DRIVE); Elucidating Loci Involved in Prostate Cancer Susceptibility (ELLIPSE); Transdisciplinary Research in Cancer of the Lung (TRICL), Liang L, Driver JA. **Investigating the genetic relationship between Alzheimer's disease and cancer using GWAS summary statistics.** Hum Genet. 2017 Oct;136(10):1341-1351. PMID: 28780673. PMCID: PMC5693762.
28. Larsson SC, Traylor M, Burgess S, Markus HS. **Genetically-Predicted Adult Height and Alzheimer's Disease.** J Alzheimers Dis. 2017;60(2):691-698. PMID: 28869480. PMCID: PMC6037293.
29. Wang XF, Lin X, Li DY, Zhou R, Greenbaum J, Chen YC, Zeng CP, Peng LP, Wu KH, Ao ZX, Lu JM, Guo YF, Shen J, Deng HW. **Linking Alzheimer's disease and type 2 diabetes: Novel shared susceptibility genes detected by cFDR approach.** J Neurol Sci. 2017 Sep 15;380:262-272. PMID: 28870582. PMCID: PMC6693589.
30. Larsson SC, Markus HS. **Branched-chain amino acids and Alzheimer's disease: a Mendelian randomization analysis.** Sci Rep. 2017 Oct 19;7(1):13604. PMID: 29051501. PMCID: PMC5648806.
31. Chaudhury S, Patel T, Barber IS, Guetta-Baranes T, Brookes KJ, Chappell S, Turton J, Guerreiro R, Bras J, Hernandez D, Singleton A, Hardy J, Mann D; ARUK Consortium, Morgan K. **Polygenic risk score in postmortem diagnosed sporadic early-onset Alzheimer's disease.** Neurobiol Aging. 2018 Feb;62:244. PMID: 29103623. PMCID: PMC5995122.
32. McKenzie AT, Moyon S, Wang M, Katsyv I, Song WM, Zhou X, Dammer EB, Duong DM, Aaker J, Zhao Y, Beckmann N, Wang P, Zhu J, Lah JJ, Seyfried NT, Levey AI, Katsel P, Haroutunian V, Schadt EE, Popko B, Casaccia P, Zhang B. **Multiscale network modeling of oligodendrocytes reveals molecular components of myelin dysregulation in Alzheimer's disease.** Mol Neurodegener. 2017 Nov 6;12(1):82. PMID: 29110684. PMCID: PMC5674813.
33. Patel T, Brookes KJ, Turton J, Chaudhury S, Guetta-Baranes T, Guerreiro R, Bras J, Hernandez D, Singleton A, Francis PT, Hardy J, Morgan K. **Whole-exome sequencing of the BDR cohort: evidence to support the role of the PILRA gene in Alzheimer's disease.** Neuropathol Appl Neurobiol. 2017 Nov 27. PMID: 29181857. PMCID: PMC6005734.
34. Larsson SC, Traylor M, Malik R, Dichgans M, Burgess S, Markus HS; CoSTREAM Consortium, on behalf of the International Genomics of Alzheimer's Project. **Modifiable pathways in Alzheimer's disease: Mendelian randomisation analysis.** BMJ. 2017 Dec 6;359:j5375. PMID: 29212772. PMCID: PMC5717765.
35. Williams DM, Karlsson IK, Pedersen NL, Hägg S. **Circulating insulin-like growth factors and Alzheimer disease: A mendelian randomization study.** Neurology. 2018 Jan 23;90(4):e291-e297. PMID: 29282328. PMCID: PMC5798653.
36. Del-Aguila JL, Fernández MV, Schindler S, Ibanez L, Deming Y, Ma S, Saef B, Black K, Budde J, Norton J, Chasse R; Alzheimer's Disease Neuroimaging Initiative (ADNI), Harari O, Goate A, Xiong C, Morris JC,

- Cruchaga C. **Assessment of the Genetic Architecture of Alzheimer's Disease Risk in Rate of Memory Decline.** *J Alzheimers Dis.* 2018;62(2):745-756. PMID: 29480181. PMCID: PMC5989565.
37. Nordestgaard LT, Tybjaerg-Hansen A, Rasmussen KL, Nordestgaard BG, Frikke-Schmidt R. **Genetic variation in clusterin and risk of dementia and ischemic vascular disease in the general population: cohort studies and meta-analyses of 362,338 individuals.** *BMC Med.* 2018 Mar 14;16(1):39. PMID: 29534716. PMCID: PMC5851250.
38. Bonham LW, Karch CM, Fan CC, Tan C, Geier EG, Wang Y, Wen N, Broce IJ, Li Y, Barkovich MJ, Ferrari R, Hardy J, Momeni P, Höglinger G, Müller U, Hess CP, Sugrue LP, Dillon WP, Schellenberg GD, Miller BL, Andreassen OA, Dale AM, Barkovich AJ, Yokoyama JS, Desikan RS; International FTD-Genomics Consortium (IFGC); International Parkinson's Disease Genetics Consortium (IPDGC); International Genomics of Alzheimer's Project (IGAP). **CXCR4 involvement in neurodegenerative diseases.** *Transl Psychiatry.* 2018 Apr 11;8(1):73. doi: 10.1038/s41398-017-0049-7. PMID: 29636460. PMCID: PMC5893558.
39. Grace C, Clarke R, Goel A, Farrall M, Watkins H, Hopewell JC. **Lack of genetic support for shared aetiology of Coronary Artery Disease and Late-onset Alzheimer's disease.** *Sci Rep.* 2018 May 8;8(1):7102. PMID: 29740026. PMCID: PMC5940751.
40. Kunutsor SK, Laukkanen JA, Burgess S. **Genetically elevated gamma-glutamyltransferase and Alzheimer's disease.** *Exp Gerontol.* 2018 Jun;106:61-66. PMID: 29505811. PMCID: PMC5915296.
41. Ni H, Xu M, Zhan GL, Fan Y, Zhou H, Jiang HY, Lu WH, Tan L, Zhang DF, Yao YG, Zhang C. **The GWAS Risk Genes for Depression May Be Actively Involved in Alzheimer's Disease.** *J Alzheimers Dis.* 2018 Jul 12. [Epub ahead of print] PMID: 30010129. DOI: 10.3233/JAD-180276
42. Girdhar K, Hoffman GE, Jiang Y, Brown L, Kundakovic M, Hauberg ME, Francoeur NJ, Wang Y-C, Shah H, Kavanagh DH, Zharovsky E, Jacobov R, Wiseman JR, Park R, Johnson JS, Kassim BS, Sloofman L, Mattei E, Weng Z, Sieberts SK, Peters MA, Harris BT, Lipska BK, Sklar P, Roussos P, Akbarian S. **Cell-specific histone modification maps in the human frontal lobe link schizophrenia risk to the neuronal epigenome.** *Nat Neurosci.* 2018 Aug;21(8):1126-1136. PMID: 30038276. PMCID: PMC6063773.
43. Chauhan G, Adams HHH, Satizabal CL, Bis JC, Teumer A, Sargurupremraj M, Hofer E, Trompet S, Hilal S, Smith AV, Jian X, Malik R, Traylor M, Pulit SL, Amouyel P, Mazoyer B, Zhu YC, Kaffashian S, Schilling S, Beecham GW, Montine TJ, Schellenberg GD, Kjartansson O, Guðnason V, Knopman DS, Griswold ME, Windham BG, Gottesman RF, Mosley TH, Schmidt R, Saba Y, Schmidt H, Takeuchi F, Yamaguchi S, Nabika T, Kato N, Rajan KB, Aggarwal NT, De Jager PL, Evans DA, Psaty BM, Rotter JI, Rice K, Lopez OL, Liao J, Chen C, Cheng CY, Wong TY, Ikram MK, van der Lee SJ, Amin N, Chouraki V, DeStefano AL, Aparicio HJ, Romero JR, Maillard P, DeCarli C, Wardlaw JM, Hernández MDCV, Luciano M, Liewald D, Deary IJ, Starr JM, Bastin ME, Muñoz Maniega S, Slagboom PE, Beekman M, Deelen J, Uh HW, Lemmens R, Brodaty H, Wright MJ, Ames D, Boncoraglio GB, Hopewell JC, Beecham AH, Blanton SH, Wright CB, Sacco RL, Wen W, Thalamuthu A, Armstrong NJ, Chong E, Schofield PR, Kwok JB, van der Grond J, Stott DJ, Ford I, Jukema JW, Vernooij MW, Hofman A, Uitterlinden AG, van der Lugt A, Wittfeld K, Grabe HJ, Hosten N, von Sarnowski B, Völker U, Levi C, Jimenez-Conde J, Sharma P, Sudlow CLM, Rosand J, Woo D, Cole JW, Meschia JF, Slowik A, Thijs V, Lindgren A, Melander O, Grewal RP, Rundek T, Rexrode K, Rothwell PM, Arnett DK, Jern C, Johnson JA, Benavente OR, Wassertheil-Smoller S, Lee JM, Wong Q, Mitchell BD, Rich SS, McArdle PF, Geerlings MI, van der Graaf Y, de Bakker PIW, Asselbergs FW, Srikanth V, Thomson R, McWhirter R, Moran C, Callisaya M, Phan T, Rutten-Jacobs LCA, Bevan S, Tzourio C, Mather KA, Sachdev PS, van Duijn CM, Worrall BB, Dichgans M, Kittner SJ, Markus HS, Ikram MA, Fornage M, Launer LJ, Seshadri S, Longstreth WT Jr, DeBette S; Stroke Genetics Network

(SiGN), the International Stroke Genetics Consortium (ISGC), METASTROKE, Alzheimer's Disease Genetics Consortium (ADGC), and the Neurology Working Group of the Cohorts for Heart and Aging Research in Genomic Epidemiology (CHARGE) Consortium. **Genetic and lifestyle risk factors for MRI-defined brain infarcts in a population-based setting.** *Neurology*. 2019 Jan 16. pii: 10.1212/WNL.0000000000006851. doi: 10.1212/WNL.0000000000006851. [Epub ahead of print]. PMID: 30651383. PMCID: PMC6369905.

44. Leonenko G, Sims R, Shoai M, Frizzati A, Bossù P, Spalletta G, Fox NC, Williams J; GERAD consortium, Hardy J, Escott-Price V. **Polygenic risk and hazard scores for Alzheimer's disease prediction.** *Ann Clin Transl Neurol*. 2019 Feb 18;6(3):456-465. doi: 10.1002/acn3.716. eCollection 2019 Mar. PMID: 30911569.
45. Chaudhury S, Brookes KJ, Patel T, Fallows A, Guetta-Baranes T, Turton JC, Guerreiro R, Bras J, Hardy J, Francis PT, Croucher R, Holmes C, Morgan K, Thomas AJ. **Alzheimer's disease polygenic risk score as a predictor of conversion from mild-cognitive impairment.** *Transl Psychiatry*. 2019 May 24;9(1):154. doi: 10.1038/s41398-019-0485-7. Erratum in: *Transl Psychiatry*. 2019 Jun 11;9(1):167. PMID: 31127079.
46. Zhou Y, Sun X, Zhou M. **Body Shape and Alzheimer's Disease: A Mendelian Randomization Analysis.** *Front Neurosci*. 2019 Oct 10;13:1084. doi: 10.3389/fnins.2019.01084. eCollection 2019. PMID: 31649504.
47. Wang L, Qiao Y, Zhang H, Zhang Y, Hua J, Jin S, Liu G. **Circulating Vitamin D Levels and Alzheimer's Disease: A Mendelian Randomization Study in the IGAP and UK Biobank.** *J Alzheimers Dis*. 2019 Dec 2. doi: 10.3233/JAD-190713. [Epub ahead of print] PMID: 3181569.

## Appendix B

### IGAP Meeting 2018, Paris

#### Participant list

##### **Alzheimer's Disease Exome Sequencing Consortia – ADES**

Jordi Clarimon, Sant Pau Memory Unit, Barcelona, Spain  
Ruth Frikke-Schmidt, Copenhagen University Hospital, Denmark  
John Hardy, University College London, UK  
Henne Holstege, Amsterdam University Medical Center, Netherlands  
Marc Hulsman, Amsterdam University Medical Center, Netherlands  
Iris Jansen, Amsterdam University Medical Center, Netherlands  
Camille Le Clezio, Normandie University, France  
Kin Mok, University College London, UK  
Gael Nicolas, Normandie University, France  
Olivier Quenez, Normandie University, France  
Maryam Shoai, University College London, UK  
Kristel Slegers, University of Antwerp, Belgium

##### **Alzheimer's Disease Genetic Consortium – ADGC**

Gary Beecham, University of Miami, Florida, USA  
Will Bush, Case Western Reserve University, Cleveland, OH, USA  
Laura Cantwell, University of Pennsylvania, Philadelphia, PA, USA  
Carlos Cruchaga, Washington University St Louis, MS, USA  
Lindsay Farrer, Boston University, Boston, MA, USA  
Tony Griswold, University of Miami, Florida, USA  
Jonathan Haines, Case Western Reserve University, Cleveland, OH, USA  
Gyungah Jun, Boston University, Boston, MA, USA  
Amanda Kuzma, University of Pennsylvania, Philadelphia, PA, USA  
Adam Naj, University of Pennsylvania, Philadelphia, PA, USA  
Margaret Pericak-Vance, University of Miami, Florida, USA  
Alan Renton, Mt Sinai School of Medicine, New York, NY, USA  
Gerard Schellenberg, University of Pennsylvania, Philadelphia, PA, USA  
Giuseppe Tosto, Columbia University University, New York, NY, USA  
Badri Vardarajan, Columbia University University, New York, NY, USA  
Li-San Wang, University of Pennsylvania, Philadelphia, PA, USA

##### **Cohorts for Heart and Aging Research in Genomic Epidemiology – CHARGE**

Josh Bis, University of Washington, Seattle, WA, USA  
Mercé Boada, Fundació ACE, Barcelona, Spain  
Anita DeStefano, Boston University, Boston, MA, USA  
Sonia Moreno, Fundació ACE, Barcelona, Spain  
Claudia Satizabal, University of Texas Health Science Center, San Antonio, TX, USA  
Sudha Seshadri, University of Texas Health Science Center, San Antonio, TX, USA  
Sven van der Lee, Amsterdam University Medical Center, Netherlands

Cornelia Van Duijn, University of Oxford, UK

Habil Zare, University of Texas Health Science Center, San Antonio, TX, USA

**European Alzheimer's Disease Initiative – EADI**

Philippe Amouyel, Institute Pasteur, Lille, France

Céline Bellenguez, Institute Pasteur, Lille, France

Vincent Chouraki, Institute Pasteur, Lille, France

Vincent Damotte, Institute Pasteur, Lille, France

Benjamin Grenier-Boley, Institute Pasteur, Lille, France

Jean-Charles Lambert, Institute Pasteur, Lille, France

Alfredo Ramirez, University of Cologne, Germany

**Genetic and Environmental Risk in Alzheimer's Disease – GERAD**

Valentina Escott-Price, Cardiff University, Wales, UK

Detelina Grozeva, Cardiff University, Wales, UK

Janet Harwood, Cardiff University, Wales, UK

Peter Holmans, Cardiff University, Wales, UK

Ioanna Katzourou, Cardiff University, Wales, UK

Ganna Leonenko, Cardiff University, Wales, UK

Rebecca Sims, Cardiff University, Wales, UK

Julie Williams, Cardiff University, Wales, UK

**Appendix C**  
**IGAP Meeting 2019, London**  
**Participant List**

**Alzheimer's Disease Exome Sequencing Consortia – ADES**

Benedetta Nacmias, University of Florence, Italy  
Bernard Fongang, University of Milan, Italy  
Henne Holstege, Amsterdam University Medical Center, Netherlands  
Marc Hulsman, Amsterdam University Medical Center, Netherlands  
Valentina Bessi, University of Florence, Italy  
Niccolo Tesi, Amsterdam University Medical Center, Netherlands

**Alzheimer's Disease Genetic Consortium – ADGC**

Adam Naj, University of Pennsylvania, Philadelphia, PA, USA  
Amanda Kuzma, University of Pennsylvania, Philadelphia, PA, USA  
Brian Kunkle, University of Miami, USA  
Carlos Cruchaga, Washington University St Louis, MS, USA  
Christiane Reitz, Columbia University, USA  
Eden Martin, University of Miami, USA  
Ekaterina Rogaeva, University of Toronto, Canada  
Fanny Leung, University of Pennsylvania, USA  
Gary Beecham, University of Miami, Florida, USA  
Gerard Schellenberg, University of Pennsylvania, Philadelphia, PA, USA  
Giuseppe Tosto, Columbia University University, New York, NY, USA  
Gyungah Jun, Boston University, Boston, MA, USA  
John Farrell, Boston University, USA  
Jonathan Haines, Case Western Reserve University, Cleveland, OH, USA  
Laura Cantwell, University of Pennsylvania, Philadelphia, PA, USA  
Lindsay Farrer, Boston University, Boston, MA, USA  
Li-San Wang, University of Pennsylvania, Philadelphia, PA, USA  
Margaret Pericak-Vance, University of Miami, Florida, USA  
Peter St. George Hyslop, University of Toronto, Canada  
Regina Carney, University of Miami, USA  
Richard Mayeux, Columbia University, USA  
Tony Griswold, University of Miami, Florida, USA  
Will Bush, Case Western Reserve University, Cleveland, OH, USA  
Brian Fulton-Howard, Mt Sinai School of Medicine, USA  
Nora Matelis, University of Miami, USA

**Cohorts for Heart and Aging Research in Genomic Epidemiology – CHARGE**

Anita DeStefano, Boston University, Boston, MA, USA  
Claudia Satizabal, University of Texas Health Science Center, San Antonio, TX, USA  
Cornelia Van Duijn, University of Oxford, UK  
Josh Bis, University of Washington, Seattle, WA, USA



Mercé Boada, Fundació ACE, Barcelona, Spain  
Sonia Moreno, Fundació ACE, Barcelona, Spain  
Sudha Seshadri, University of Texas Health Science Center, San Antonio, TX, USA  
Sven van der Lee, Amsterdam University Medical Center, Netherlands  
Shahzad Amad, Erasmus Medical Center, Netherlands  
Xueqiu Jian, University Texas Health Science Center, USA  
Honghuang Lin, Boston University, USA  
Agustín Ruiz, Fundacio ACE, Spain  
Chloe Sarnowski, Boston University, USA

**European Alzheimer's Disease Initiative – EADI**

Alfredo Ramirez, University of Cologne, Germany  
Benjamin Grenier-Boley, Institute Pasteur, Lille, France  
Céline Bellenguez, Institute Pasteur, Lille, France  
Jean-Charles Lambert, Institute Pasteur, Lille, France  
Vincent Damotte, Institute Pasteur, Lille, France  
Chiara Fenoglio, Boston University, USA  
Juan Pascual Sanchez, University Hospital Marqués de Valdecilla, Spain

**Genetic and Environmental Risk in Alzheimer's Disease – GERAD**

Detelina Grozeva, Cardiff University, Wales, UK  
Ganna Leonenko, Cardiff University, Wales, UK  
Janet Harwood, Cardiff University, Wales, UK  
Julie Williams, Cardiff University, Wales, UK  
Peter Holmans, Cardiff University, Wales, UK  
Rebecca Sims, Cardiff University, Wales, UK  
Valentina Escott-Price, Cardiff University, Wales, UK

**Appendix D**  
**Approved Foreign Collaborations**

List of foreign collaborations (with State Department Clearance):

<b>Collaborator</b>	<b>Institution</b>	<b>Country</b>
Julie Williams	Cardiff University	United Kingdom
Philippe Amouyel	Institut Pasteur de Lille	France
Matthias Riemenschneider	Saarland University	Germany
Alfredo Ramirez	University of Bonn	Germany
Ryozo Kuwano	Niigata University	Japan
Jong-Won Kim	Samsung Medical Center	Korea
Kunho Lee	Chosun University	Korea
Vilmundur Gudnason	Icelandic Heart Association	Iceland

## Appendix E

### ADGC Analysis Proposals – Special Analysis Groups (SAGs)

#### Year 1 SAGs (private funding)

1. Washington University
  - a. Topic – ***Use of biological and evolutionary information to prioritize SNPs from the ADGC genome-wide association studies for follow-up***
  - b. Investigators – Alison Goate (PI), Keoni Kauwe, Carlos Cruchaga, Sarah Bertelsen
  - c. Ended because project as originally constructed no longer relevant (12/14/2015)
2. Vanderbilt University
  - a. Topic – ***Detection of Gene x Gene Interactions in LOAD GWAS Data***
  - b. Investigators – Jonathan Haines (PI), Marilyn Ritchie, Kim Luci, Kristin Brown, Scott Dudek
  - c. Published: NIHMS ID 736058.
3. NIA and University College London
  - a. Topic - ***Epigenetic and Expression Modifications Associated with AD Risk SNPs***
  - b. Investigators – John Hardy, Andrew Singleton, Michael Nalls, J Raphael Gibbs, Dena Hernandez, Rita Guerreiro
  - c. Published: Epub 2013 Jan 30. PMID: 23360175. PMCID: PMC3578142.
4. OHSU
  - a. Topic – ***Utilization of Genetic Architecture to identify candidate genes for Alzheimer's Disease***
  - b. Investigators – Shannon McWeeney (PI), Jeffrey Kaye, Beth Wilmot, Guanming Wu
  - c. NOT FUNDED
5. Mayo Clinic
  - a. Topic – ***Analysis of the ADGC GWAS Datasets to Identify Alzheimer's Disease Susceptibility Factors Influencing Neuropathology Endophenotypes***
  - b. Investigators – Nilufer Ertekin-Taner (PI), Steven Younkin

#### Year 2 SAGs (U01 funded) – All approved and funded at \$20k December 9, 2010

1. University of Kentucky
  - a. Topic – ***GWAS to identify SNPs associated with pathologically-verified hippocampal sclerosis***
  - b. Investigators - Peter Nelson MD PhD (PI), Steven Estus PhD, and David Fardo PhD
  - c. Funds requested – \$24,000
2. Mass General (and University of Washington)
  - a. Topic – ***A genome-wide association study of Alzheimer's disease with psychosis (AD+P)***
  - b. Investigators – Deborah Blacker MD (PI), Alan Fung MD, Debby Tsuang MD, Rebecca Betensky PhD
  - c. Funds requested – \$30,000
3. Columbia University
  - a. Topic – ***Identification of mitochondrial DNA variants associated with Alzheimer's disease***

- b. Investigators - Sandra Barral Rodriguez PhD (PI), Rong Cheng PhD, Joseph Lee Dr PH, Christiane Reitz MD PhD
- c. Funds requested – \$24,788
- 4. University of Washington (and Boston University, Indiana University)
  - a. Topic – ***Psychometrically sophisticated GWAS: Cognitive decline among AD cases in the ADGC***
  - b. Investigators – Paul Crane MD (PI), S McKay Curtis MD, Robert C Green MD, Shubhabrata Mukherjee PhD, Andrew Saykin PsyD, Li Shen PhD, Richard Sherva PhD
  - c. Funds requested – \$37,230

### Analysis proposals

- 1. University of Pennsylvania
  - a. Topic - ***CNV Analysis of the ADGC Phase 1+2 Dataset***
  - b. Investigators - Li-San Wang PhD, Chiao-Feng Lin PhD, Gerard Schellenberg PhD
  - c. Submitted November 3, 2009 - Approved December 8, 2009
- 2. Mayo Clinic
  - a. Topic – ***Analysis of variants in the chromosome 10 VR22/LRRTM3 region in the combined ADGC and Mayo Clinic Datasets***
  - b. Investigators - Nilufer Ertekin-Taner MD PhD
  - c. Submitted July 31, 2010 - Approved August 13, 2010
- 3. University of Pennsylvania
  - a. Topic - ***Pathway and eSNP Analysis of AD and Cognitive decline***
  - b. Investigators – Li-San Wang PhD, Chiao-Feng Lin PhD, Otto Valladares, Gerard Schellenberg PhD
  - c. Submitted December 28, 2009 – Approved November 3, 2010
- 4. Washington University
  - a. Topic – ***Analysis of ADGC GWAS data for association with cerebrospinal fluid (CSF) biomarkers for LOAD***
  - b. Investigators – Alison Goate PhD, Carlos Cruchaga PhD, Tony Hinrichs PhD, Sarah Bertelsen, John Kauwe PhD
  - c. Submitted January 22, 2010 – Approved November 3, 2010
- 5. Boston University
  - a. Topic - ***Analysis of Gene Interactions for AD Risk Using a Cooperative Game Theory Approach***
  - b. Investigators - Badri Vardarajan, Lindsay Farrer PhD, Adrienne Cupples PhD, Kathryn Lunetta PhD
  - c. Submitted September 26, 2010 – Approved November 12, 2010
- 6. New York University
  - a. Topic – ***Analysis of Gene-Environment Interactions Involved in AD***
  - b. Investigators – Iryna Lobach PhD (PI), Tatiyana Apanasovich PhD, Hongyu Zhao PhD, Steven Ferris PhD, Barry Reisberg MD
  - c. Submitted December 2, 2010 – Approved March 1, 2011 – Resubmitted April 05, 2011 – Approved May 3, 2011
- 7. University of Pennsylvania

- a. Topic – **Evaluation of Mitochondrial Heteroplasmy in AD GWAS datasets**
  - b. Investigators – Neal Sondheimer MD PhD (PI), Jack Tirone, Li-San Wang PhD, Gerard Schellenberg PhD
  - c. Submitted December 10, 2010 – Approved March 1, 2011
8. ROS/MAP
- a. Topic – **Validation of a CR1 susceptibility allele**
  - b. Investigators – Philip L. De Jager MD PhD (PI), Eric Reiman MD, David A. Bennett MD
  - c. Submitted December 16, 2010 – Approved March 1, 2011
9. Mayo Clinic
- a. Topic – **Combined GWAS for brain gene expression levels and AD risk**
  - b. Investigators - Dr. Nilufer Ertekin-Taner (PI), Dr. Steven G. Younkin, Dr. Fanggeng Zou and Ms. Mariet Allen, Drs. Shane Pankratz, Julia Crook and High-Seng Chai
  - c. Submitted January 18, 2011 – Approved March 1, 2011
10. ROS/MAP
- a. Topic –**Replication of Genome-wide Scans for AD-related Intermediate Phenotypes and Functional Validation in Drosophila**
  - b. Investigators – Philip L. De Jager MD PhD (PI), Mel Feany MD PhD, David A. Bennett MD
  - c. Submitted February 7, 2011 – Approved March 16, 2011
11. Columbia University
- a. Topic - **Multi-locus genotype patterns associated with episodic memory in the NIA-LOAD/NCRAD Study**
  - b. Investigators – Sandra Barral PhD (PI), Richard Mayeux MD
  - c. Submitted February 11, 2011 - Approved March 16, 2011
12. University of Washington
- a. Topic - **Pathway-wide association study (PWAS) of AD**
  - b. Investigators – Paul Crane MD (PI), Shubhadbrata Mukherjee PhD, S. McKay Curtis PhD, Emily Tittschuh PhD, Robert Green MD, Rick Sherva PhD, Andy Saykin PhD, Li Shen PhD, Keoni Kauwe PhD
  - c. Submitted March 7, 2011 – Approved April 21, 2011
13. University of Pennsylvania
- a. Topic - **Alzheimer's Disease and toxicity exposure: a gene-environment Interactions analysis**
  - b. Investigators – Shin-Yi Chou (PI) and Rhea Bhatta (Lehigh University) and Laura Cantwell, Li-San Wang and Gerard Schellenberg (University of Pennsylvania)
  - c. Submitted May 6, 2011 – Approved June 9, 2011
14. Columbia University
- a. Topic - **SORCS2, SORCS3 and SORT1 and risk of Alzheimer's disease**
  - b. Investigators – Chistine Reitz, MD PhD, Lindsay Farrer, PhD, and Richard Mayeux, MD MSc
  - c. Submitted May 19, 2011 – Approved June 9, 2011
15. Mayo Clinic
- a. Topic - **Determine risk associated with TMEM106B SNP rs1990622 in AD**
  - b. Investigators – Dr. Rosa Rademakers, Dr. Minerva Carrasquillo and Dr. Steven Younkin
  - c. Submitted May 27, 2011 – Approved June 9, 2011
16. Mayo Clinic

- a. Topic - **Joint Analysis of Genomic and Transcriptomic Data to Identify Novel AD Risk Genes**
  - b. Investigators – Dr. Nilufer Ertekin-Taner (PI), Dr. Steven G. Younkin, Dr. Fanggeng Zou and Ms. Mariet Allen (Department of Neuroscience), Drs. Shane Pankratz, Julia Crook and High-Seng Chai (Department of Biostatistics), from Mayo Clinic. Dr. John Blangero (Texas Biomedical Research Institute).
  - c. Submitted June 4, 2011 – Approved June 13, 2011
17. Oregon Health & Sciences University
- a. Topic – **Replication of a GWAS for autopsy-confirmed AD cases and controls**
  - b. Investigators – Patricia Kramer, Matthew Huentelman, Jason Corneveaux, Shawn Westaway, and Jurg Ott
  - c. Submitted June 1, 2011 – Approved June 11, 2011
18. Autopsy-confirmed Parkinson Disease GWAS Consortium (APDGC)
- a. Topic – **ADGC Controls for Parkinson Disease GWAS**
  - b. Investigators – Dr. William Scott, Dr. Gary Beecham and Cherylyn Jauregui
  - c. Submitted August 12, 2011 – Approved August 26, 2011
19. Brigham Young University
- a. Topic – **Pleiotropy and Interactions in CSF Biomarkers for AD**
  - b. Investigators – Keoni Kauwe, PhD, Alison Goate, PhD, Taylor Maxwell PhD, and Perry Ridge, MS
  - c. Submitted August 2, 2011 – Resubmitted September 16, 2011 – Approved October 4, 2011
20. Genentech
- a. Topic – **Whole-genome sequence analysis of early-onset Alzheimer's disease in APOE 3/4 and 4/4 subjects**
  - b. Investigators – Timothy Behrens, Ward Ortmann, and Robert C. Gentleman
  - c. Submitted September 15, 2011 – Approved September 21, 2011
21. Emory University
- a. Topic – **The proportion of common SNPs that explain late-onset Alzheimer's disease**
  - b. Investigators – Thomas Wingo, MD and David Cutler, PhD
  - c. Submitted September 8, 2011 – Revisions requested September 22, 2011 – Resubmitted October 10, 2011 – Approved November 15, 2011
22. North American Pancreatic Study Group (NAPS)
- a. Topic – **Genetic risk in recurrent acute and chronic pancreatitis**
  - b. Investigators – David Whitcomb, MD, PhD, Bernie Devlin, MD
  - c. Submitted December 28, 2011 – Approved January 30, 2012
23. Brigham Young University
- a. Topic – **Remaining genetic variance in clinical diagnosis of Alzheimer's disease**
  - b. Investigators – Keoni Kauwe, PhD, and Perry Ridge, MS
  - c. Submitted August 2, 2011 – Resubmitted January 5, 2012 – Approved January 30, 2012
24. University of San Francisco
- a. Topic – **Investigation for genetic loci underlying depressive symptomatology in Alzheimer's Disease**

- b. Investigators – Jennifer Yokoyama (PI), Howard Rosen, MD (mentor), Bruce Miller, MD, Kristine Yaffe, MD, Andrew Saykin, PsyD, Steven Hamilton, MD, PhD, Dena Dubal, MD, PhD, Amy L. Byers, PhD, MPH, Lindsay A. Farrer, PhD
  - c. Submitted December 23, 2011 – Resubmitted January 23, 2012 – Approved February 10, 2012
25. University of California, Los Angeles
- a. Topic – **Analysis of genetic association of dementia and handedness**
  - b. Investigators - John Ringman (PI), Gerard Schellenberg, PhD, Li-San Wang, PhD
  - c. Submitted March 1, 2012 – Resubmitted April 20, 2015 – Approved May 11, 2015
26. University of California San Diego
- a. Topic – **Leveraging annotation and genetic pleiotropy methods to improve gene discovery in Alzheimer's disease**
  - b. Investigators – Rahul Desikan (PI), Anders Dale, Nicholas Schork, Ole Andreassen
  - c. Submitted March 25, 2012 – Revisions requested April 20, 2012– Resubmitted May 21, 2012 – Review June 6, 2012 - Executive Committee objection June 11, 2012 – Re-review June 20, 2012 – Approved June 20, 2012
27. University of Washington
- a. Topic – **Bayesian genome-wide association study (BGWAS) of Alzheimer's Disease**
  - b. Investigators – Su-In Lee, PhD (PI), Santosh Srivastava, PhD, Paul Crane, MD, Shubhabrata Mukherjee, PhD
  - c. Submitted May 14, 2012 – Approved July 5, 2012
28. University of Washington/Crane Pathway SAG
- a. Topic – **Comparison of approaches to gene-wide association studies for AD**
  - b. Investigators – Crane Pathway SAG, Valentina Moskvina
  - c. Submitted July 25, 2012 – Approved September 19, 2012
29. University of Washington
- a. Topic – **Mendelian randomization in the ADGC: obesity and AD risk**
  - b. M. Maria Glymour, Stefan Walter, PhD, Gail Jarvik, MD PhD, David Crosslin, PhD, Su-In Lee, PhD, John “Keoni” Kauwe, PhD, from Brigham Young University; Andy Saykin, ScD, from Indiana University, Dave Fardo, PhD, from the University of Kentucky, Robert Green, MD, from Harvard University, Rick Sherva, PhD, from Boston University, Shubhabrata (Joey) Mukherjee, PhD, from the University of Washington, and Paul Crane, MD MPH, from the University of Washington
  - c. Submitted August 7, 2012 – Approved September 19, 2012 – Addendum to add autopsy variable submitted September 27 – Approved October 3, 2012
30. Indiana University
- a. Topic – **Identification of rare variants contributing to Parkinson disease: Request for control data**
  - b. Tatiana Foroud, PhD, Daniel Koller, PhD, Dongbing Lai, MS, Janice Farlow, MD PhD
  - c. Submitted October 1, 2012 – Approved October 8, 2012
31. University of Washington/NACC
- a. Topic - **Cure-rate Model for Dementia with the combined ADC and UDS**
  - b. Bud Kukull , Andrew Zhou, Yue Ma, and Sarah Monsell
  - c. Submitted October 19, 2012 – Revisions requested November 1, 2012 – Resubmitted December 10, 2012 – Approved February 18, 2013

32. Pfizer
  - a. Topic - **Association between lipids (HDL-c, LDL-c, and TG) and AD**
  - b. Hyuna Yang, Eve H Pickering and Jens Wendland
  - c. Submitted November 2012 - Approved November 19, 2012
33. University of Washington
  - a. Topic - **Analyses of SNP associations with AD from the combined ADGC data set**
  - b. Shubhabrata Mukherjee, PhD, Adam Naj, PhD, Sungeun Kim, PhD, Paul Crane, MD MPH, Timothy Thornton, PhD, Ellen Wijsman, PhD, David Fardo, PhD, John S K Kauwe, PhD, Otto Valladares, MS
  - c. Submitted November 2, 2012 and Amendment December 10, 2012 – Approved February 18, 2013
34. University of Kentucky /University of Washington/Indiana University
  - a. Topic - **Inflammation and Alzheimer's Disease: A Genome-wide Investigation of Interrelated Pathways**
  - b. David Fardo, PhD, Lorri Morford, PhD, Shubhabrata Mukherjee, PhD, Andy Saykin, PsyD, and Paul Crane, MD MPH
  - c. Submitted November 1, 2012 – Approved February 18, 2013
35. Columbia University
  - a. Topic - **Interaction between environmental factors and genetic variants in late onset Alzheimer's disease**
  - b. Christiane Reitz, MD PhD, Richard Mayeux, MD MSc
  - c. Approved February 19, 2013
36. Washington University
  - a. Topic - **Using CSF biomarker levels to identify potential functional mechanisms of the GWAS hits**
  - b. Alison Goate, PhD., Carlos Cruchaga, PhD. and John Kauwe, PhD., Sheng Chih Jin, ScM
  - c. Submitted December 18, 2012 – Approved February 18, 2013
37. Brigham Young University
  - a. Topic - **Remaining genetic variance in clinical diagnosis of Alzheimer's disease**
  - b. John Kauwe, PhD, Perry Ridge, MS
  - c. Submitted February 12, 2013 – Approved February 18, 2013
38. Mayo Clinic
  - a. Topic - **MAPT haplotype analyses in ADGC dataset**
  - b. Dr. Nilufer Ertekin-Taner Drs. Mariet Allen and Minerva Carrasquillo, Drs. Shane Pankratz and Julia Crook
  - c. Submitted February 18, 2013 – Approved February 25, 2013
39. King's College London
  - a. Topic - **CSF1R and association with late onset Alzheimer's Disease**
  - b. Angela Hodges and Simon Lovestone (King's College London), Gerard Schellenberg (University of Pennsylvania), Adam Naj (University of Pennsylvania)
  - c. Submitted February 26 – Approved March 1, 2013
40. Columbia University
  - a. Topic – **Memory performance in ADC/NACC data**
  - b. Sandra Barral, Richard Mayeux



- c. Submitted April 25, 2013 – Resubmitted May 30, 2013 – Approved June 3, 2013
41. University of Washington
    - a. Topic – ***Genetic differences between symptomatic and asymptomatic persons with Alzheimer's Disease neuropathologic change***
    - b. Sarah Monsell, MS, Charles Mock, MD, PhD, Alison Goate, PhD, Catherine Roe, PhD, John Morris, MD, Nigel Cairns, PhD, FRCPATH, Walter Kukull, PhD
    - c. Submitted April 26, 2013 – Approved June 24, 2013
  42. Stanford University
    - a. Topic - ***Interaction between APOE and gender in functional imaging biomarkers***
    - b. Michael D Greicius, Andre Altmann, Richard Mayeux
    - c. Submitted July 23, 2013 – Revised October 2, 2013 – Approved October 9, 2013
  43. University of Washington
    - a. Topic - ***Genetics of neuropathologically defined and CSF defined resilience phenotypes***
    - b. Paul Crane, Rich Jones, Shubhabrata Mukherjee, Keoni Kauwe, Carlos Cruchaga, Gary Beecham, Tom Montine, Dave Fardo, Josh Sonnen, Bruce Reed, Yaakov Stern, Eric Larson, Phil De Jager, Josh Shulman, Carol Brayne, et al.
    - c. Submitted July 26, 2013 – Approved September 27, 2013
  44. Columbia University
    - a. Topic - ***Confirmation of the APOE effector genes using the combined ADGC data set***
    - b. Joseph Lee, Rong Cheng, Badri Vardarajan, Sandra Barral, Christiane Reitz, Giuseppe Tosto, Herve Rhinn, Asa Abeliovich, Richard Mayeux
    - c. Submitted August 8, 2013 – Revised September 26, 2013 – Approved October 9, 2013
  45. Vanderbilt University
    - a. Topic – ***Leveraging biomarker and autopsy measures of pathology to identify novel genetic interactions***
    - b. Tricia Thornton-Wells, Timothy Hohman, Jonathan Haines, Alison Goate, John Kauwe, Lori Chibnik
    - c. Submitted August 28, 2013 – Approved September 27, 2013
  46. Washington University in St. Louis
    - a. Topic - ***Use of censored data to identify genetic modulators of age at onset of AD in the entire ADGC dataset and in subsets stratified by APOE genotype***
    - b. Alison Goate, Carlos Cruchaga, John Kauwe and Oscar Harari
    - c. Submitted September 10, 2013 – Revised October 7, 2013 – Approved October 23, 2013
  47. Harvard University
    - a. Topic – ***Request for summary statistics of Late-Onset Alzheimer Disease GWAS In Europeans and African Americans***
    - b. Philip De Jager, Kathleen Hall, Denis Evans
    - c. Submitted September 16, 2013 – Approved September 30, 2013
  48. Columbia University
    - a. Topic – ***Analysis of genetic susceptibility loci underlying Alzheimer's disease with parkinsonism and hallucinations***
    - b. Guiseppe Tosto, Richard Mayeux
    - c. Submitted October 7, 2013 – Approved October 22, 2013
  49. Toronto University

- a. Topic – **Genome-wide survey of runs of homozygosity in African Americans with late-onset Alzheimer Disease**
  - b. Ekaterina Rogava, Peter St. George-Hyslop, Richard Mayeux, Christiane Reitz, Guiseppe Tosto, Joseph Lee, Badri Vardarajan
  - c. Submitted October 8, 2013 – Approved October 22, 2013
50. Washington University
- a. Topic - **Analysis of ADGC GWAS data for association with CSF biomarkers for LOAD**
  - b. Alison Goate, Carlos Cruchaga, Keoni Kauwe
  - c. Submitted November 12, 2013 – Approved November 22, 2013
51. Columbia University
- a. Topic – **Re-Analysis of IGAP Mega-Meta loci and Sortilin pathway genes in African Americans**
  - b. Christiane Reitz, Richard Mayeux
  - c. Submitted November 13, 2013 – Approved November 30, 2013
52. Columbia University
- a. Topic – **Dentate Gyrus differential expressed genes and memory performance in ADGC data**
  - b. Sandra Barral, Richard Mayeux
  - c. Submitted November 25, 2013 – Approved December 11, 2013
53. University of Pennsylvania
- a. Topic – **Mendelian randomization analysis of the relationship between depression and Alzheimer's disease**
  - b. Li-San Wang, Adam Naj, Tianyan Hu, Fanny Leung
  - c. Submitted November 25, 2013 – Approved December 11, 2013
54. Boston University
- a. Topic - **A liability model to increase power in genome-wide association**
  - b. Jesse Mez, Jaeyoon Chung, Guyungah June, Mark Logue, Matthew Schu, Kathryn Lunetta, Lindsay Farrer
  - c. Submitted January 2, 2014 – Approved January 22, 2014
55. University of Washington
- a. Topic – **Interaction of TAU haplotypes and Head Injury in conferring risk of Alzheimer's disease**
  - b. Debby Tsuang, MD, Brad Boeve, MD, Nilufer Taner, MD, PhD, Dan Schaid, PhD, Badri Vardarajan, PhD, Richard Mayeux, MD
  - c. Submitted January 13, 2014 – Approved January 27, 2014
56. University of Kentucky
- a. Topic – **Replication and Exploration of Alzheimer's Disease Epistasis Signals**
  - b. David Fardo, Elena Gusareva, Kristel Van Steen
  - c. Submitted February 3, 2014 – Approved February 26, 2014
57. Columbia University
- a. Topic – **Using GWAS results to identify the types of functional genomic information most relevant for interpreting loci that influence Alzheimer's disease**
  - b. Joseph Pikrell, Christiane Reitz
  - c. Submitted April 28, 2014 – Approved May 20, 2014

58. University of Washington
  - a. Topic - ***A systems-biology approach to identify candidate genes for Alzheimer's Disease by integrating protein-protein interaction network and subsequent in vivo validation of candidate genes using a C. elegans model of A $\beta$  toxicity***
  - b. Shubhabrata Mukherjee, PhD; Matt Kaeberlein, PhD; Kevin Boehme; John SK. Kauwe, PhD; David Fardo, PhD; Adam Naj, PhD; and Paul Crane, MD MPH
  - c. Submitted May 29, 2014 – Approved July 22, 2014
59. University of California San Diego
  - a. Topic - ***Leveraging genetic overlap to detect novel Alzheimer's disease susceptibility loci***
  - b. Dr. Rahul Desikan, Dr. Yunpeng Wang, Andrew Schork, Professor Wes Thompson, Dr. Ole Andreassen and Dr. Anders Dale
  - c. Submitted July 29, 2014 – Approved September 26, 2014
60. University of Washington
  - a. Topic – ***Conditional and joint multiple-SNP analysis of GWAS summary statistics from the IGAP 1000 genomes-imputed meta-analysis***
  - b. Shubhabrata Mukherjee, Paul Crane, Brian Kunkle, Kevin Boehme, John Kauwe, Dave Fardo
  - c. Submitted October 28, 2014 – Approved November 10, 2014
61. Boston University
  - a. Topic - ***Investigation of the Association of CpG-related SNPs (CGSs) with AD***
  - b. Yiyi Ma, Kathryn Lunetta, Gyungah Jun, Lindsay A. Farrer
  - c. Submitted December 1, 2014 – Approved December 8, 2014
62. University of Miami
  - a. Topic - ***Association Analysis of ABCA7 deletion in ADGC African American GWAS Datasets***
  - b. Brian Kunkle, Margaret Pericak-Vance
  - c. Submitted December 15, 2014 – Approved December 22, 2014
63. Children's Hospital of Philadelphia
  - a. Topic – ***The role of vitamin D in prevention of cognitive decline in Alzheimer's Disease: a Mendelian Randomization study***
  - b. Hakon Hakonarson, Jeff Roizen, Alex Casella
  - c. Submitted December 22, 2014 – Approved March 9, 2015
64. University of California San Diego
  - a. Topic - ***Identifying Polygenic Basis of the Human Brain and Brain Disorders***
  - b. Chi-Hua Chen, Anders Dale, Min-Tzu Lo
  - c. Submitted January 26, 2015 – Approved March 9, 2015
65. University of Pennsylvania
  - a. Topic - ***Evaluating a Potential Independent Association with AD at ABCA7***
  - b. Mitchell Tang, Li-San Wang, Adam Naj, and Gerard Schellenberg
  - c. Submitted March 4, 2015 – Approved March 9, 2015
66. Children's Hospital of Philadelphia
  - a. Topic - ***Comparative analysis of the sex specific association results for AD against results for fronto-temporal dementia (FTD) to identify shared loci / pathways***
  - b. Hakon Hakonarson, Patrick Sleiman
  - c. Submitted May 11, 2015 – Approved May 21, 2015

67. University of Southern California
  - a. Topic - ***Pathways, effect of genetic interaction on age of onset, ventricular dilation rate and amyloid- $\beta$  level, and AD risk prediction***
  - b. Kaida Ning, Arthur Toga, Fengzhu Sun
  - c. Submitted June 18, 2015 – Approved August 24, 2015
68. Vanderbilt University
  - a. Topic – ***Disentangling the Association between Genetic Ancestry and Socioeconomic Status***
  - b. Tim Hohman, Jonathan Haines
  - c. Submitted July 31, 2015 – Approved August 21, 2015
69. TGen
  - a. Topic – ***Disease association analysis of variants with differential effects dependent on APOE status***
  - b. Matthew Huentelman
  - c. Submitted August 13, 2015 – Approved August 20, 2015
70. University of Washington
  - a. Topic – ***Conditional and joint multiple-SNP analysis of GWAS summary statistics from the ADGC neuropathological analysis***
  - b. Joey Mukherjee, Paul Crane, Kevin Boehme, John Kauwe, David Fardo, Thomas Montine, Gary Beecham, C. Dirk Keene
  - c. Submitted August 19, 2015 – Approved September 4, 2015
71. University of Washington
  - a. Topic – ***Post-GWAS prioritization of Alzheimer's disease loci based on genomic functional annotation***
  - b. Joey Mukherjee, Qionshi Lu, Brian Kunkle, Paul Crane, Hongyu Zhao
  - c. Submitted August 21, 2015 – Approved September 4, 2015
72. University of Pennsylvania
  - a. Topic – ***Comorbid Lewy Body Dementia among AD cases in the ADGC***
  - b. John Toledo, Adam Naj, Li-San Wang, Gerard Schellenberg, Virginia Lee, John Trojanowski
  - c. Submitted August 23, 2015 – Revision received October 21, 2015 – Approved November 23, 2015
73. Boston University
  - a. Topic – ***Multivariate phenotypes association study of neuropathological features of Alzheimer's disease and related dementias***
  - b. Jaeyoon Chung, Yiyi Ma, Kathryn Lunetta, Jesse Mez, Gyungah Jun, Lindsay Farrer
  - c. Submitted August 24, 2015 – Approved September 28, 2015
74. University of Pennsylvania
  - a. Topic – ***Genetic Influence and Clinical Expression of Tauopathy across AD/FTD***
  - b. David Irwin, Adam Naj, Jeffery Philips, Felicia Cooper, Murray Grossman, John Trojanowski, Vivianna Van Deerlin
  - c. Submitted October 7, 2015 – Approved December 1, 2015
75. University of California San Diego
  - a. Topic – ***Application of Novel Methods for Accommodating Heterogeneity***
  - b. Nicholas Schork, Chi-Hua Chen, Qian Peng, Danjuma Quarless

- c. Submitted November 18, 2015 – Approved December 4, 2015
76. Columbia University
- a. Topic – **Identification of endocytosis-related variants associated with entorhinal cortex volume in AD**
  - b. Julia Ciampa, Christiane Reitz, Scott Small
  - c. Submitted December 14, 2015 – Approved January 11, 2016
77. University of Southern California
- a. Topic – **The relationship of the KL-VS variants to phenotypic traits of Alzheimer's disease in humans**
  - b. Investigators – Priya Bhatt, Arthur Toga
  - c. Submitted January 25, 2016 – Approved February 24, 2016
78. Yale University
- a. Topic - **Improving genetic risk prediction for Alzheimer's disease through integrating genomic functional annotation**
  - b. Investigators - Yiming Hu, Qiongshi Lu, Shubhabrata Mukherjee, Paul Crane, Hongyu Zhao
  - c. Submitted March 4, 2016 – Revised March 31, 2016 – Approved April 14, 2016
79. University of San Francisco
- a. Topic – **Investigation of HLA contributions to risk for Alzheimer's Disease**
  - b. Investigators: Natasha Rabinowitz, MPH, Jill Hollenbach, PhD, Vincent Damotte, PhD, Luke Bonham, Ethan Geier, PhD, Bruce Miller, MD
  - c. Submitted June 7, 2016 – Approved June 20, 2016
80. Case Western – Will Bush
- a. Topic - **Nuclear-Mitochondrial Interactions that Influence Neurocognitive Impairment**
  - b. Investigators – Will Bush, Sandra Smieszek, Sandra Barral, Asha Kallianpur
  - c. Submitted June 24, 2016 – Approved July 11, 2016
81. Columbia University
- a. Topic – **Genetic Variation Underlying Traumatic Brain Injury and Late-Onset Alzheimer's Disease**
  - b. Investigators – Sandra Barral, Badri Vardarajan, Yaakov Stern
  - c. Submitted July 18, 2016 – Approved August 22, 2016
82. University of Pennsylvania and University of Texas
- a. Topic – **Tissue-specific analysis of genetically-regulated expression in Alzheimer's Disease (AD)**
  - b. Investigators – Jennifer “Piper” Below, Adam Naj, Li-San Wang, Jerry Schellenberg
  - c. Submitted August 16, 2016 – Approved August 22, 2016 – *Ongoing with goal of publication*
83. University Miami
- a. Topic – **IGAP rare-variant GWAS Pathway analysis proposal**
  - b. Investigators – Peter Holmans, Brian Kunkle, Mariusz Butkiewicz
  - c. Submitted August 2016 – Approved August 31, 2016
84. Boston University
- a. Topic – **Identifying Functional AD Genes by Integrating Large-Scale eQTL and Alzheimer's Disease Genetic Association Data**
  - b. Investigators – Dan Lancour, Xiaoling Zhang, Jaeyoon Chung, Kathryn Lunetta, and Lindsay Farrer

- c. Submitted August 18, 2016 – Approved September 19, 2016
85. Boston University
  - a. Topic – **Age Stratified Analysis**
  - b. Investigators – Yiyi Ma, Jaeyoon Chung, Kathryn Lunetta, Gyungah Jun, Xiaoling Zhang, Lindsay A. Farrer
  - c. Submitted August 18, 2016 – Approved September 19, 2016
86. Vanderbilt University
  - a. Topic – **Sex-specific drivers of Alzheimer's Disease**
  - b. Investigators – Timothy Hohman, Brian Kunkle, Joey Mukherjee, Gary Beecham
  - c. Submitted August 30, 2016 – Approved September 19, 2016
87. Boston University
  - a. Topic - **Genetics of Rate of Cognitive Decline in AD Cases**
  - b. Investigators – Rick Sherva, Paul Crane, Joey Mukherjee, Alden Gross
  - c. Submitted August 30, 2016 – Approved September 19, 2016
88. Boston University
  - a. Topic - **Evaluation of Pleiotropy between Cancer and Alzheimer Disease as a Means to Detect Novel AD Susceptibility Loci**
  - b. Investigators – Jaeyoon Chung, Yiyi Ma, Kathryn Lunetta, Fergus Couch, Gyungah Jun, Lindsay Farrer
  - c. Submitted September 1, 2016 – Approved September 19, 2016
89. University of Pennsylvania
  - a. Topic – **Analysis of common and rare variants in progressive supranuclear palsy using ADGC controls**
  - b. Investigators – Gerard Schellenberg, Li-San Wang, Adam Naj, Weixin Wang, Laura Cantwell, Otto Valladares, Amanda Partch
  - c. Submitted October 6, 2016 – Approved November 18, 2016
90. University of Pennsylvania
  - a. Topic – **Computational analysis of regulatory effects of IGAP-identified noncoding genetic variants**
  - b. Investigators – Alexandre Amlie-Wolf, Li-San Wang, Gerard Schellenberg, Mitchell Tang, Beth Dombroski, Jessica King
  - c. Submitted October 6, 2016 – Approved November 18, 2016
91. Case Western
  - a. Topic - **Loss of the Y chromosome in Alzheimer's Disease**
  - b. Investigators – Will Bush, Lars Fosberg, Adam Naj, Sandra Smieszek
  - c. Submitted February 21, 2017 – Approved March 6, 2017
92. Boston University
  - a. Topic - **Identification of mitochondrial variants associated with late-onset Alzheimer's disease**
  - b. Investigators – Xiaoling Zhang, John Farrell, Thomas Zhou, Jaeyoon Chung, Adam Naj, Kathryn Lunetta, Lindsay Farrer
  - c. Submitted February 23, 2017 – Approved March 6, 2017
93. Indiana University

- a. Topic - ***Apolipoprotein E gene polymorphism and Alzheimer's disease in different populations***
  - b. Investigators – Kwangsik Nho, Kun Ho Lee (ADGC PI – Lindsay Farrer)
  - c. Submitted March 3, 2017 – sent response and questions April 24, 2017
94. Indiana University
- a. Topic - ***Testing a Model of Cognitive Function with APOE and BDNF Genes and Heart Failure among Persons with Normal Cognition, MCI, and AD***
  - b. Investigators – Miyeon Jung, Susan Pressler, Liana Apostolova, Andrew Saykin (ADGC PI - Schellenberg)
  - c. Submitted April 4, 2017 – Approved April 24, 2017
95. Columbia University
- a. Topic - ***Epistasis in late-onset Alzheimer's disease***
  - b. Investigators – Giuseppe Tosto, Badri Vardarajan, Richard Mayeux
  - c. Submitted April 10, 2017 – Approved April 24, 2017
96. University of Washington
- a. Topic - ***Higher density imputation panel samples for the EPAD project***
  - b. Investigators – Paul Crane, Shubhabrata Mukherjee, Gary Beecham, Adam Naj, David Fardo, Keoni Kauwe, Laura Gibbons, Jesse Mez, Emily Trittschuh, Andy Saykin
  - c. Submitted May 17, 2017 – Approved July 3, 2017
97. University of California San Diego
- a. Topic - ***Cross-Ethnic prediction on the age-at-onset of Alzheimer's disease with polygenic hazard scores***
  - b. Investigators – Rahul Desikan, Chun-Chieh Fan, Ole Andreassen, Anders Dale
  - c. Submitted June 16, 2017 – Approved August 14, 2017
98. University of Pennsylvania/Columbia University
- a. Topic - ***Analysis of Gene-Environment Interactions in Alzheimer's Disease: Education and Intelligence***
  - b. Investigators – Rhea Bhatta, Li-San Wang, Gerard Schellenberg, Shin-Yi Chou, Richard Mayeux, Badri Vardarajan, Neha Raghavan
  - c. Submitted June 29, 2017 – Approved August 14, 2017
99. University of Miami
- a. Topic - ***Sex-specific Analysis of the ADSP Case-control and ADGC Datasets***
  - b. Investigators – Brian Kunkle, Kara Hamilton, Eden martin, Gary Beecham, Adam Naj, Will Bush, Tim Hohman, Margaret Pericak-Vance
  - c. Submitted June 29, 2017 – Approved August 14, 2017
100. University of Washington
- a. Topic - ***Gene-level association mapping for late-onset Alzheimer's disease through cross-tissue imputation of gene expression***
  - b. Investigators – Qiongshi Lu, Mo Li, Yiming Hu, Hongyu Zhao, Shubhabrata Mukherjee, Brian Kunkle, Paul Crane
  - c. Submitted July 5, 2017 – Revision submitted August 9, 2017
101. University of Washington
- a. Topic - ***Analysis of TOMM40 gene promoter haplotype in Alzheimer's disease***
  - b. Investigators – Chang-En Yu, Li-San Wang

- c. Submitted August 3, 2017
102. Boston University
  - a. Topic - ***Disease Specific Imputation Panel for Testing Association of Late-Onset Alzheimer Disease***
  - b. Investigators – John Farrell, Gyungah Jun, Yiyi Ma, Devanshi Patel, Jaeyoon Chung, Kathryn Lunetta, Lindsay Farrer
  - c. Submitted August 7, 2017 – Approved September 14, 2017
103. University of Miami
  - a. Topic - ***ADGC African American Haplotype Reference Consortium Genome-wide Association Study***
  - b. Investigators – Brian Kunkle, Christiane Reitz, Kara Hamilton, Richard Mayeux, Margaret Pericak-Vance
  - c. Submitted August 16, 2017 – Approved October 12, 2017
104. University of Pittsburgh
  - a. Topic – ***Alzheimer's Disease + Psychosis***
  - b. Investigators - Robert A. Sweet MD, Bernie Devlin PhD, M. Ilyas Kamboh PhD, Lambertus Klei PhD, Mikhail Bamne PhD, Howard Seltman MD, PhD, Mary Ann DeMichele-Sweet PhD, Elise Weamer MPH
  - c. Submitted August 31, 2017 – Approved October 12, 2017
105. University of Kentucky
  - a. Topic - ***Examining AD-related endophenotypes, partitioned genetic covariance, and cognitive decline***
  - b. Investigators – David Fardo, Teresa Filshtein, Timothy Hohman, Qiongshi Lu, Beth Mormino, Shubhabrata Mukherjee, Dan Mungas
  - c. Submitted September 18, 2017 – Approved October 31, 2017
106. University of Kentucky
  - a. Topic - ***Exploration of Alzheimer's Disease SNPs and Candidate Causal Proxies***
  - b. Investigators – David Fardo, Yuriko Katsumata
  - c. Submitted September 22, 2017 – Approved November 9, 2017
107. Mayo Jacksonville
  - a. Topic – ***Meta-analyses of ADGC African American GWAS and Mayo AA WES datasets***
  - b. Investigators – Nilufer Ertekin-Taner, Minerva Carrasquillo, Mariet Allen, Joseph Reddy, Jerry Schellenberg
  - c. Submitted October 21, 2017 – Approved December 22, 2017
108. Icahn School of Medicine at Mount Sinai
  - a. Topic – ***Discovery of Rare Variation and Non-Alzheimer's Phenotypes Associated with Polygenic AD Risk***
  - b. Investigators – Edoardo Marcora, Brian Fulton-Howard, Alan Renton, Alison Goate
  - c. Submitted October 30, 2017 – Approved January 5, 2018
109. Massachusetts General Hospital
  - a. Topic - ***Genetic association between Alzheimer's disease and cerebral small/large vessel diseases in subgroups stratified by APOE genotypes***



- b. Investigators - Jaeyoon Chung, Sandro Marini, Matthew Traylor, Rainer Malik, Braxton D Mitchell, Gyungah R Jun, Kathryn L. Lunetta, Lindsay A Farrer, Christopher D Anderson, Jonathan Rosand
  - c. Submitted January 10, 2018 – Approved May 9<sup>th</sup> 2018
110. Washington University
- a. Topic – **Identification of the genetic architecture common to Alzheimer's disease risk and rate of progression**
  - b. Investigators – Carlos Cruchaga, Oscar Harari, Jorge Del-Aguila
  - c. Submitted January 19, 2018 – Revision requested February 19, Received March 6 – Approved March 30, 2018
111. University of California San Diego
- a. Topic - **Identification of Genetic Heterogeneity of Alzheimer's Disease (AD) by Age, Sex, and APOE Status**
  - b. Investigators – Chi-Hua Chen, Min-Tzu Lo, Linda McEvoy
  - c. Submitted March 23, 2018 – Approved May 9, 2018
112. Case Western and Columbia University
- a. Topic - **Assessment of Genomic-based Stroke Risk in the ADGC Cohorts**
  - b. Investigators – Will Bush, Giuseppe Tosto, Tim Hohman, Richard Mayeux, Adam Naj, Penny Bencheck, Akila Nappallan
  - c. Submitted March 28, 2018 – Approved April 13, 2018
113. University of Miami
- a. Topic - **Sex-specific Analysis of the ADGC AND IGAP Datasets**
  - b. Investigators – Brian Kunkle, Eden Martin, Lily Wang, Kara Hamilton, Adam Naj, Will Bush, Tim Hohman, and Margaret Pericak-Vance
  - c. Submitted April 4, 2018 – Approved May 2, 2018
114. University of San Francisco
- a. Topic - **Post-GWAS protein interaction network analysis to identify novel biological processes in Alzheimer's disease**
  - b. Investigators – Luke Bonham, Ethan Geier, Jessie Carr, Patricia Castruita, Rahul Desikan, Sergio Baranzini, Noah Zaitlen, Hao Li, Jialiang Gu, Zachary Miller
  - c. Submitted April 4, 2018 – Approved May 2, 2018
115. Icahn School of Medicine at Mount Sinai and University of Pennsylvania
- a. Topic - **APOE  $\epsilon$ 4 and  $\epsilon$ 3/ $\epsilon$ 3 Extremes Meta-analysis using ADGC HRC-Imputed data with ADSP sequence data**
  - b. Investigators – Alan Renton, Edoardo Marcora, Yi Zhao, Adam Naj
  - c. Submitted April 24, 2018 – Approved May 23, 2018
116. University of Washington
- a. Topic – **Genetically-informed association analysis tests associations between risk factors and late-onset Alzheimer's disease**
  - b. Investigators – Qiongshi Lu, Donghui Yan, Hermione Wang, Bowen Hu, Shubhabrata Mukherjee, Paul Crane, Brian Kunkle
  - c. Submitted June 20, 2018 – Approved August 6, 2018
117. University of California San Francisco
- a. Topic – **Cardiovascular mediated polygenic enrichment in Alzheimer's disease**

- b. Investigators – Rahul Desikan, Iris Broce, Chin Hong Tan
  - c. Submitted July 19, 2018 – Approved August 6, 2018
118. University of Miami
- a. Topic - ***Genetic Basis for Age-at-Onset of AD***
  - b. Investigators – Eden Martin, Yi-Ju Li, Xiaoyi Raymond Gao, Mike Schmidt, Adam Naj
  - c. Submitted July 23, 2018 – Approved August 6, 2018
119. University of Pennsylvania and Columbia University
- a. Topic – ***Studying the regulatory effects of population-specific noncoding genetic variants identified in AD GWAS studies***
  - b. Investigators – Fanny Leung, Alexandre Amlie-Wolf, Pavel Kuksa, Yi Zhao, Yi-Fan Chou, Badri Varadarajan, Li-San Wang, Gerard Schellenberg
  - c. Submitted September 10, 2018 – Approved October 17, 2018
120. Boston University and Banner Health
- a. Topic – ***Comprehensive assessment for effect sizes of APOE genotypes for Alzheimer's disease risk and neuropathological traits***
  - b. Investigators – Gyungah Jun, Lindsay A. Farrer, Gerard D. Schellenberg, Eric M. Reiman,
  - c. Submitted January 5, 2019 – Approved January 23, 2019
121. University of Pennsylvania
- a. Topic – ***PART Analysis***
  - b. Investigators – Kyra Jefferson-George, David Wolk, Edward Lee, Laura Wisse, Sandhitsu Das, Adam Naj, Corey McMillan
  - c. Submitted January 20, 2019 – Approved March 13, 2019
122. Icahn School of Medicine at Mount Sinai
- a. Topic – ***Identify and genotype copy number variants (CNVs) and perform CNV based GWAS and eQTL analyses for Alzheimer's Disease***
  - b. Investigators – Alison Goate, Ke Hao, Zhongyang Zhang, Haoxiang Cheng
  - c. Submitted February 15, 2019 – Approved June 3, 2019
123. University of Munich and Mayo Clinic
- a. Topic – ***MSA GWAS***
  - b. Investigators – Owen Ross, Franziska Hopfner, Gregor Kuhlenbäumer, Ulrich Müller, Günter Höglinger, Gerard Schellenberg
  - c. Submitted March 22, 2019 – Approved April 28, 2019
124. Massachusetts General Hospital
- a. Topic – ***Understanding the Protective Effect of an APOE ε4-linked haplotype using Integrative Bioinformatics Analysis***
  - b. Investigators – Sudeshna Das, Deborah Blacker, Lori Chibnik, Brad Hyman, Colin Magdamo, Li-San Wang, Gerard Schellenberg
  - c. Submitted April 5, 2019 – Approved June 13, 2019
125. University of Southern Denmark and University of California San Diego
- a. Topic – ***Analysis of genetic variants in Alzheimer's disease using a hierarchical model of the cell***
  - b. Investigators – Trey Ideker and Daniel Carlin (University of California San Diego), Jan Baumbach (University of Munich), Simon Larsen (University of Southern Denmark)
  - c. Submitted April 30, 2019 – Approved May 15, 2019

126. Van Andel Institute and the University of Pennsylvania
  - a. Topic – ***Lewy Body Dementia Genetics***
  - b. Investigators – Jose Bras, Rita Guerriero, Adam Naj
  - c. Submitted May 8, 2019 – Approved June 13, 2019
127. Boston University
  - a. Topic - ***APOE Stratified Polygenic Risk Score Analysis using Machine Learning and Mendelian Randomization Approach***
  - b. Investigators – Gyungah Jun, Concong Zhu, Rebecca Panitch, Jaeyoon Chung, Lindsay Farrer
  - c. Submitted June 18, 2019 – Approved September 27, 2019
128. University of Miami
  - a. Topic - ***Estimating Bias-free Polygenic Risk Scores for Alzheimer Disease in Admixed Populations***
  - b. Investigators - Farid Rajabli, Gary W. Beecham, Kara Hamilton-Nelson, , James Jaworski, Brian Kunkle, Eden R Martin, Margaret A. Pericak-Vance
  - c. Submitted June 27, 2019, - Approved September 17, 2019
129. Boston University
  - a. Topic - ***Genetic Association Studies for Identifying Novel AD Risk Factors Using Polygenic Structure of Cerebral Small Vessel Disease***
  - b. Investigators –
  - c. Submitted July 24<sup>th</sup>, 2019 – Approved August 28, 2019

## Appendix F

### ADGC data sets available through NIAGADS

#### ADGC datasets

Cohort	Platform	Cases / Controls	Total	NIAGADS dataset ID
<b>Discovery (Stage 1)</b>				
ACT	Illumina Human660	365 / 94	2171	NG00034
ADC1	Illumina Human660	1985 / 523	2731	NG00022
ADC2	Illumina Human660	688 / 116	926	NG00023
ADNI	Illumina Human660	268 / 173	441	Via ADNI
GenADA	Affymetrix 500	669 / 713	1382	Contact PI
Genetic Differences	Illumina Human660	213 / 0	213	NG00034
UMVUMSSM	Illumina Human660/1M Duo/Affymetrix 6.0	1177 / 1123	2462	NG00042
MIRAGE	Illumina Human610/330	491 / 738	1502	NG00031
NIA-LOAD	Illumina 610	1798 / 1568	4410	NG00032
OSHU	Illumina Human CNV370v1_C	647	647	NG00017
TGEN2	Affymetrix 6.0	668 / 365	1511	NG00028
<b>Replication (Stage 2)</b>				
ADC3	Illumina OmniExpress	843 / 430	1526	NG00024
MAYO	Illumina HumanHap300	728 / 1173	2099	NG00043
ROSMAP	Affymetrix 6.0	294 / 763	1668	NG00029
UPitt	Illumina Human Omni1-Quad	1255 / 829	2219	NG00026
WashU	Illumina Human610	259 / 186	576	NG00030
<b>Additional datasets</b>				
ADC4	Illumina OmniExpress	430 / 372	1054	NG00068
ADC5	Illumina OmniExpress	400 / 539	1224	NG00069
ADC6	Illumina OmniExpress	563 / 298	1333	NG00070
ADC7	Illumina OmniExpress+exome	529 / 706	1462	NG00071
ADC Exome Chip - NorthShore	Illumina HumanExome	3776 / 2222	5998	NG00079
Miami Exome Chip	Illumina HumanExome	931 / 992	1923	NG00080
ADGC Exome Chip - CHOP	Illumina HumanExome	2792 / 2388	5180	NG00081
Wash U Exome Chip	Illumina HumanExome	519 / 349	868	NG00085
ADGC AA WES	Illumina HiSeq	1373 / 1781	3200	In process
ADC8	Illumina OmniExpress+exome	517 / 664	1181	In process
ADC9	Illumina Global Screening Array	728 / 896	1624	In process
ADC10	Illumina Global Screening Array	457 / 724	1181	In process
ADC11	Illumina Global Screening Array	883 / 1265	2148	Genotyping

## ADGC/IGAP Summary Statistics

<b>Dataset</b>	<b>NIAGADS dataset ID</b>
ADGC Summary Statistics- Naj et al. (2011)	NG00027
ADGC African American Summary Statistics- Reitz et al. (2013)	NG00039
ADGC Neuropath Summary Stats and Phenotypes- Beecham et al. (2014)	NG00041
ADGC Age at Onset Summary Statistics- Naj et al. (2014)	NG00048
IGAP Summary Statistics ADGC Subset- Lambert et al. (2013)	NG00053
Transethnic GWAS Summary Statistics - Jun et al. (2017)	NG00056
IGAP Rare Variant Summary Statistics- Kunkle et al. (2019)	NG00075
ADGC case-control summary statistics on 7050 samples not included in the IGAP-2013 discovery stage	NG00076
IGAP APOE-Stratified Analysis Summary Statistics- Jun et al. (2015)	NG00078
GWAS Summary Statistics of informed conditioning analysis in African Americans	NG00088

## Appendix G

### 2019 Alzheimer's Disease Genetics Symposium

#### Attendees

Total Number of Attendees: 194

Number of Institutions: 45

<b>Institution</b>	<b>Number of Attendees</b>
Albert Einstein College of Medicine	1
Boston University School of Medicine	5
BrightFocus Foundation	1
Carlos Albizu University	1
Case Western Reserve University	4
Children's Hospital of Philadelphia	6
Chosun University	1
Cleveland Clinic Lerner Research Institute	1
Columbia University University	8
Drexel University	1
Eli Lilly and Company	1
Envoy At Home LLC	1
Fundacio ACE Barcelona	1
Icahn School of Medicine at Mount Sinai	8
Indiana University School of Medicine	2
Institut Pasteur de Lille	1
Institute for Biomedical Informatics	1
Johns Hopkins	1
Lehigh University	1
Massachusetts General Hospital, Harvard Medical School	2
Mayo Clinic	1
National Centralized Repository for Alzheimer's Disease	1
National Institute on Aging Genetics of Alzheimer's Disease Data Storage Site	2
National Institutes of Health – National Institute on Aging	6
Pfizer	1
Rush University Medical Center	1
Stanford University School of Medicine	1
University of California San Diego	1
University of Florida	2
University of Kansas	2
University of Kentucky	1
University of Miami	11
University of Michigan	1
University of North Texas Health Science Center	1
University of Pennsylvania	93
University of Pittsburgh	1
University of Texas Health Sciences Center, San Antonio	1
University of Toronto	1
University of Toronto and University of Cambridge	1

University of Utah	1
University of Washington	4
University of Washington/VA Puget Sound	1
Vanderbilt University Medical Center	5
Wake Forest School of Medicine	1
Washington University St. Louis	5

	<b>Speaker</b>	<b>Affiliation/Institution</b>
1	Gerard Schellenberg	University of Pennsylvania
2	Eliezer Masliah	NIH-NIA
3	Chun Chieh Fan	University of California San Diego
4	Dennis Dickson	Mayo Clinic
5	Adam Naj	University of Pennsylvania
6	Lindsay Farrer	Boston University
7	Jean-Charles Lambert	Institut Pasteur de Lille
8	Alessandra Chesi	University of Pennsylvania
9	Alison Goate	Icahn School of Medicine at Mount Sinai
10	Brian Kraemer	University of Washington
11	Phil de Jager	Columbia University
12	David Collier	Eli Lilly
13	Virginia Lee	University of Pennsylvania
14	Augustin Ruiz	Fundacio ACE Barcelona
15	Goldie Byrd	Wake Forest School of Medicine
16	Richard Mayeux	Columbia University
17	Jonathan Haines	Case Western Reserve University
18	Farid Rajabli	University of Miami
19	David Bennett	Rush University
20	Todd Golde	University of Florida
21	Hakon Hakonarson	Children's Hospital of Philadelphia

## Appendix H

### List of Studies Contributing to the ADGC

Short	Study	PI/Contact
<b>ACT</b>	Adult Changes in Thought	Eric Larson
<b>ADC</b>	NIA Alzheimer's Disease Center	Walter Kukull/Gerard Schellenberg
<b>ADNI</b>	Alzheimer's Disease Neuroimaging Initiative	Andy Saykin
<b>AGES</b>	Age Gene/Environment Susceptibility Reykjavik study	Vilmundur Gudnason
<b>BIOCARD</b>	Biomarkers of Cognitive Decline among Normal Individuals	Marilyn Albert
<b>CHAP</b>	Chicago Health and Aging Project	Denis Evans
<b>CHOSUN</b>	Chosun University National Research Center for Dementia	Kun Ho Lee
<b>EAS</b>	Einstein Aging Study	Richard Lipton
<b>EFIGA</b>	Estudio Familiar de la Influencia de la Genética del Alzheimer	Richard Mayeux
<b>GenADA</b>	Genetic Alzheimer's Disease Associations study	Peter St George-Hyslop/GSK
<b>GENDIFF</b>	Genetic Differences in Alzheimers Cases and Controls	Walter Kukull/Gerard Schellenberg
<b>GenerAAtions</b>	Genetic and Environmental Risk Factors for Alzheimer Disease Among African Americans	M. Daniele Fallin
<b>INDY</b>	Indianapolis African Americans and Indianapolis-Ibaden Dementia cohorts	Kathleen Hall
<b>JGSCAD</b>	Japanese Genetic Study Consortium for AD	Ryozo Kuwano
<b>Manly</b>	African American AD Genetics Study	Jennifer Manly
<b>MAP</b>	Rush Memory and Aging Project	David Bennett
<b>MARS/CORE</b>	Minority Aging Research Study	Lisa Barnes
<b>MAYO</b>	Jacksonville Mayo Clinic	Nilufer Ertekin-Taner, Neill Graff-Radford
<b>MESA</b>	Multi-Ethnic Study of Atherosclerosis	Stephen Rich
<b>Mexico 10/66</b>	10/66 Dementia Research Group, Mexico	Ana Luisa Sosa
<b>MHAS</b>	Mexican Health and Aging Study	Rebeca Wong
<b>MIRAGE</b>	Multi-Institutional Research on Alzheimer Genetic Epidemiology	Lindsay Farrer
<b>MSSM</b>	Mt Sinai School of Medicine	Joseph Buxbaum
<b>NBB</b>	Netherlands Brain Bank	Mathias Riemenschneider
<b>NC</b>	North Carolina A&T University	Goldie Byrd
<b>NCRAD</b>	National Cell Repository for Alzheimer's Disease	Tatiana Foroud
<b>NIA-LOAD</b>	NIA Late-Onset Alzheimer's Disease Family Study	Richard Mayeux
<b>OHSU</b>	Oregon Health Sciences University	Patricia Kramer
<b>PFIZER</b>	Pfizer LEADe, Vitamin E, and A3041005 clinical trials	Laszlo Kiss
<b>REGARDS</b>	Reasons for Geographic and Racial Differences in Stroke	
<b>RMAYO</b>	Rochester Mayo	Ronald Petersen



<b>ROS</b>	Rush Religious Orders Study	David Bennett
<b>SAMSUNG</b>	Samsung Medical Center	Jong-Won Kim
<b>TARCC</b>	Texas Alzheimer's Research and Care Consortium	Robert Barber
<b>TGEN</b>	Translational Genomics Research Institute	Eric Reiman
<b>UKS</b>	Saarland University	Matthias Riemenschneider
<b>UM</b>	University of Miami/Duke University	Margaret Pericak-Vance
<b>UPITT</b>	University of Pittsburgh	M. Ilyas Kamboh
<b>VU</b>	Vanderbilt University/Case Western Reserve University	Jonathan Haines
<b>WADI ARA</b>	Wadi Ara Israeli-Arab prevalence study of dementia	Lindsay Farrer
<b>WASHU</b>	Washington University St. Louis	Alison Goate/Carlos Cruchaga
<b>WHICAP</b>	Washington Heights-Inwood Community Aging Project	Richard Mayeux