

## John “Keoni” S.K. Kauwe, Ph.D.

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### Education

2007 Ph.D. Evol, Ecol and Population Biology, Washington University, St. Louis, MO  
2003 M.S. Population Genetics, Brigham Young University, Provo, UT  
1999 B.S. Molecular Biology, Brigham Young University, Provo, UT

### Research Experience

2008 – Pres **Assistant Professor**– Brigham Young University, Dept. of Biology.  
Identification and functional characterization of disease associated genetic variation

2007 – 2008 **Postdoctoral Research Scholar**– Washington University School of Medicine. Identification and functional characterization of disease associated genetic variation

2004 – 2007 **Ph.D. Thesis**– Lab of Dr. Alison Goate, Washington University in St. Louis.  
“Using cerebrospinal fluid protein levels as quantitative endophenotypes to identify novel genetic risk factors for Alzheimer’s disease”

2004 **Graduate Research Assistant**– Lab of Dr. Brian Suarez, Washington University. Analyzed data for the Genetics Analysis Workshop 14, comparing the utility of SNPs from the Affymetrix and Illumina genotyping chips and microsatellite markers for genome-wide linkage scans and studies of population stratification.

2004 **Graduate Research Assistant**– Lab of Dr. Alan Templeton, Washington University. Implemented novel, evolution-based methods of analyzing haplotype data for association with both quantitative and qualitative phenotypes.

2003 **Graduate Research Assistant**– Lab of Dr. Alison Goate, Washington University. Performed genotyping and analysis for association studies of Alzheimer’s disease candidate genes.

2001 – 2003 **M.S. Thesis**– Lab of Dr. Dennis Shiozawa, Brigham Young University.  
“Phylogenetic and nested clade analysis of the stonefly *Pteronarcys californica* in the Western United States.”

1998 – 1999 **Undergraduate Research Assistant**, Brigham Young University  
Assisted in the Fish DNA Laboratory

1996 – 1998 **Undergraduate Research Assistant**, Brigham Young University  
Maintained freshwater aquariums

### **Teaching Experience**

2010 **Instructor**, Brigham Young University  
Designed and presented course materials for Biology 463: Genetics of Human Disease

2009 **Instructor**, Brigham Young University  
Designed and presented course materials for Physiology and Developmental Biology 120: Science of Biology

2008 **Instructor**, Washington University in St. Louis  
Designed and presented course materials for “Research Focus”, a course designed to prepare freshman for pursuing a career in research

2008 **Lecturer**, Washington University School of Medicine  
Presented a lecture on “Genetics and Model Organisms” for the Medical School course entitled “Medical Genetics”

2005 – 2006 **Teaching Assistant**, Washington University in St. Louis  
Presented lectures and assisted in exam grading and preparation for courses in “Population Genetics” and “Evolution”

2002 – 2003 **Adjunct Faculty**, Utah Valley University.  
Designed and presented lectures, wrote tests, and evaluated papers for “Basic Biology” course

2001 – 2002 **Instructor**, Brigham Young University.  
Designed class activities and taught field methods for “Methods in Ecology” course

2001 **Teaching Assistant**, Brigham Young University.  
Presented lectures; assisted in quiz and exam preparation for “Molecular Cell Biology” course

### **Other Experience**

2008 – Pres. **Consultant**, King’s View LLC  
Evaluate research and technology and provide scientific advice relating to investment decisions.

2005 – 2008 **Coordinator**, Biomedical Research Apprenticeship Program Lecture Series. Organized and moderated the summer lecture series.

2002 – 2003 **Vice President**, Brigham Young University Graduate Student Association.  
Directed publicity and organized events.

### **Research Awards, Fellowships and Grants**

2011 Mentored Research Environment Grant, Brigham Young University  
2011 BYU Gerontology Program Grant  
2010 NIH R01 AG035083 Subcontract PI  
2010 Mentored Research Environment Grant, Brigham Young University  
2009 Excellence in Research Oral Presentation Award, NIDDK NMRI  
2009 NIH R0302309: Subcontract PI  
2009 Human Genome Variation and Complex Genome Analysis Meeting Grant  
2008 Human Genome Variation and Complex Genome Analysis Meeting Grant  
2008 Ford Foundation Postdoctoral Fellowship (declined)  
2008 International Conference on Alzheimer's Disease Travel Fellowship  
2008 Hope Center for Neurological Disorders Postdoctoral Fellowship  
2007 Poletsky Award for Research in Alzheimer's disease  
2007 NIH Training Grant Fellow T32 MH14677  
2007 Human Genome Variation 2007 Poster Presentation Contest Winner  
2007 Human Genome Variation and Complex Genome Analysis Meeting Grant  
2007 Alzheimer's Disease Biomarkers Meeting Outstanding Poster Award  
2007 O'Leary Award for Research in Neuroscience Finalist  
2005 Ford Foundation Pre-Doctoral Fellowship  
2004 NIH Training Grant Fellow "Genome Analysis Training Program"  
2003 Brigham Young University Research Presentation Award  
2003 Brigham Young University Integrative Biology Department Travel Grant  
2002 Brigham Young University Research Presentation Award  
2002 Brigham Young University Integrative Biology Department Travel Grant  
2002 D. Elden Beck Natural History Award  
1996 Brigham Young University Full Tuition Academic Scholarship  
1996 Robert C. Byrd Federal Academic Merit Scholarship

### **Other Awards and Honors**

2008 Featured in [The Record "Gallery of Graduates"](#)  
2008 Washington University Commencement Student Marshall  
2003 Utah Valley State College Adjunct Faculty Excellence Award Finalist

### **Editorial Positions**

Ad Hoc Reviewer:

**American Journal of Medical Genetics Part B: Neuropsychiatric Genetics**  
**Archives of Neurology**  
**Bioinformatics**  
**Dementia and Geriatric Cognitive Disorders**  
**Molecular Ecology**  
**Neurobiology of Aging**  
**PLoS ONE**

**PLoS Genetics**  
**The International Journal of Neuroscience**  
**The Journal of Alzheimer's Disease**  
**The Journal of the Neurological Sciences**

**Grant Review**

Ad Hoc Reviewer:

**Knight Alzheimer's Disease Research Center**  
**NIH National Institute on Aging Neuroscience Study Section**  
**Alzheimer's Association**  
**Nathan Shock Center for Excellence in the Basic Biology of Aging**  
**Department of Veteran's Affairs**

**Publications (11 first/last author papers, 31 total)**

1. Li Y, Nowotny P, Holmans P, Smemo S, **Kauwe JSK**, Hinrichs AL, Tacey K, et al. (2004) Association of late-onset Alzheimer's disease with genetic variation in multiple members of the GAPD gene family. *Proc Natl Acad Sci U S A* 101:15688–15693
2. **Kauwe JSK**, Shiozawa DK, Evans RP (2004) Phylogenetic and nested clade analysis of the stonefly *Pteronarcys californica* in the Western United States. *Journal of the North American Benthological Society* 23:824–838
3. Nowotny P, Hinrichs AL, Smemo S, **Kauwe JSK**, Maxwell T, Holmans P, Hamshere M, et al. (2005) Association studies between risk for late-onset Alzheimer's disease and variants in insulin degrading enzyme. *Am J Med Genet B Neuropsychiatr Genet* 136:62–68
4. Dunn G, Hinrichs A, Bertelsen S, Jin C, **Kauwe JSK**, Suarez B, Bierut L (2005) Microsatellites versus single-nucleotide polymorphisms in linkage analysis for quantitative and qualitative measures. *BMC Genetics* 6:S122
5. Hinrichs A, Bertelsen S, Bierut L, Dunn G, Jin C, **Kauwe JSK**, Suarez B (2005) Multipoint identity-by-descent computations for single-point polymorphism and microsatellite maps. *BMC Genetics* 6:S34
6. **Kauwe JSK**, Bertelsen S, Bierut L, Dunn G, Hinrichs A, Jin C, Suarez B (2005) The efficacy of short tandem repeat polymorphisms versus single-nucleotide polymorphisms for resolving population structure. *BMC Genetics* 6:S84
7. Suarez B, Taylor C, Bertelsen S, Bierut L, Dunn G, Jin C, **Kauwe JSK**, Paterson A, Hinrichs A (2005) An analysis of identical single-nucleotide polymorphisms genotyped by two different platforms. *BMC Genetics* 6:S152
8. Smemo S, Nowotny P, Hinrichs AL, **Kauwe JSK**, Cherny S, Erickson K, Myers AJ, Kaleem M, Marlowe L, Gibson AM, Hollingworth P, O'Donovan M C, Morris CM, Holmans P, Lovestone S, Morris JC, Thal L, Li Y, Grupe A, Hardy J, Owen MJ, Williams J, Goate A (2006) Ubiquilin 1 polymorphisms are not associated with late-onset Alzheimer's disease. *Ann Neurol* 59:21–26
9. Grupe A, Li Y, Rowland C, Nowotny P, Hinrichs AL, Smemo S, **Kauwe JSK**, et al. (2006) A scan of chromosome 10 identifies a novel locus showing strong association with late-onset Alzheimer disease. *Am J Hum Genet* 78:78–88

10. Li Y, Grupe A, Rowland C, Nowotny P, **Kauwe JSK**, Smemo S, Hinrichs A, et al. (2006) DAPK1 variants are associated with Alzheimer's disease and allele-specific expression. *Hum Mol Genet.* 15:2560–2568.
11. Mukherjee O, Pastor P, Cairns NJ, Chakraverty S, **Kauwe JSK**, et al. (2006) HDDD2 is a familial frontotemporal lobar degeneration with ubiquitin-positive, tau negative inclusions caused by a missense mutation in the signal peptide of progranulin. *Ann Neurol.* 60:314–322.
12. Mukherjee O, **Kauwe JSK**, Mayo K, Morris JC, Goate AM. (2007) Haplotype-based association analysis of the MAPT locus in Late-onset Alzheimer's disease. *BMC Genetics.* 8:3.
13. Wahrle SE, Shah AR, Fagan AM, Smemo S, **Kauwe JSK**, et al. (2007) Apolipoprotein E levels in cerebrospinal fluid and the effects of ABCA1 polymorphisms. *Mol Neurodegener* 2:7.
14. **Kauwe JSK**, Jacquart S, Chakraverty S, Wang J, Mayo K, Fagan AM, Holtzman D, Morris JC, Goate AM. (2007) Extreme cerebrospinal fluid amyloid beta levels identify family with late-onset Alzheimer's disease presenilin 1 mutation. *Ann Neurol* 61:446-53.
15. Nowotny P, Simcock X, Bertelsen S, Hinrichs AL, **Kauwe JSK**, et al. (2007) Association studies testing for risk for late-onset Alzheimer's disease with common variants in the beta-amyloid precursor protein (APP). *Am J Med Genet B Neuropsychiatr Genet.* 144:469–474.
16. Nowotny P, Bertelsen S, Hinrichs AL, **Kauwe JSK**, Mayo K, Morris JC, Goate AM. (2007) Association studies between common variants in prolyl isomerase PIN1 and risk for late-onset Alzheimer's disease. *Neurosci Lett* 419:15–7.
17. Hu Y, **Kauwe JSK**, Gross J, Cairns NJ, Goate AM, Fagan AM, Townsend RR, Holtzman DM. (2007) Identification and validation of novel CSF biomarkers for early stages of Alzheimer's disease. *Prot. Clin. Appl.* 1, 1373-1384.
18. Mukherjee O, Wang J, Gitcho M, Chakraverty S, Taylor-Reinwald L, Shears S, **Kauwe JSK**, et al. (2008) Molecular characterization of novel progranulin (GRN) mutations in frontotemporal dementia. *Hum Mutat.* 29:512-521.
19. **Kauwe JSK**, Wang J, Chakraverty S, Goate AM, Henao-Martinez AF (2008) Novel presenilin 1 variant (P117A) causing Alzheimer's disease in the fourth decade of life. *Neurosci Lett* 438:257-259
20. **Kauwe JSK**, Cruchaga C, Mayo K, Fenoglio C, Bertelsen S, Nowotny P, Galimberti D, Scarpini E, Morris JC, Fagan AM, Holtzman DM, Goate AM (2008) Variation in MAPT is associated with cerebrospinal fluid tau levels in the presence of amyloid-beta deposition. *Proc Natl Acad Sci U S A* 105:8050-8054
21. **Kauwe JSK**, Wang J, Mayo K, Morris JC, Fagan AM, Holtzman DM, Goate AM (2009). Alzheimer's disease risk variants show association with cerebrospinal fluid amyloid beta. *Neurogenetics* 10: 13-7.
22. Harold, D, Abraham R, Hollingworth P et al. (Including **Kauwe JSK**) (2009). "Genome-wide association study identifies variants at CLU and PICALM associated with Alzheimer's disease." *Nat Genet* 41(10): 1088-93.
23. Clifford DB, Fagan AM, Holtzman DM, Morris JC, Teshome M, Shah AR, **Kauwe JSK** (2009) CSF biomarkers of Alzheimer disease in HIV-associated neurologic disease. *Neurology* 73:1982-1987.
24. Fenoglio C, Galimberti D, Cortini F, **Kauwe JSK**, Cruchaga C, Venturelli E, Villa C,

- Serpente M, Scalabrini D, Mayo K, Piccio LM, Clerici F, Albani D, Mariani C, Forloni G, Bresolin N, Goate AM, Scarpini E (2009) Rs5848 Variant Influences GRN mRNA Levels in Brain and Peripheral Mononuclear Cells in Patients with Alzheimer's Disease. *J Alzheimers Dis.* 18:603-12.
25. **Kauwe JSK**, Bertelsen S, Mayo K, Cruchaga C, Abraham R, Hollingworth P, Harold D, Owen MJ, Williams J, Lovestone S, Morris JC, Goate AM (2010) Suggestive synergy between genetic variants in TF and HFE as risk factors for Alzheimer's disease. *Am J Med Genet B Neuropsychiatr Genet.* 153B(4): 955-9.
26. \***Kauwe, JSK**, \*Cruchaga C, Bertelsen S, Mayo K, Latu W, Nowotny P, Hinrichs AL, Fagan AM, Holtzman DM, Alzheimer's Disease Neuroimaging Initiative and Goate AM (2010) "Validating Predicted Biological Effects of Alzheimer's Disease Associated SNPs Using CSF Biomarker Levels." *J Alzheimers Dis.* Epub date 2010/07/17.
27. \*Cruchaga C, \***Kauwe JSK**, Mayo K, Spiegel N, Bertelsen S, Nowotny P, Shah AR, Abraham R, Hollingworth P, Harold D, Owen MM, Williams J, Lovestone S, Peskind ER, Li G, Leverenz JB, Galasko D, Morris JC, Fagan AM, Holtzman DM, Goate AM, the Alzheimer's Disease Neuroimaging Initiative (2010) SNPs Associated with Cerebrospinal Fluid Phospho-Tau Levels Influence Rate of Decline in Alzheimer's Disease. *PLoS Genetics* 6:e1001101
28. Jones L, Holmans PA, Hamshere ML, Harold D, Moskvina V, Ivanov D, Pocklington A, et al. (Including **Kauwe JSK**) (2010) Genetic Evidence Implicates the Immune System and Cholesterol Metabolism in the Aetiology of Alzheimer's Disease. *PLoS ONE* 5:e13950
29. **Kauwe JSK**, Cruchaga C, Karch CM, Sadler B, Lee M, Mayo K, Latu W, Su'a M, Fagan AM, Holtzman DM, Morris JC, ADNI and Goate AM. (2011). Fine Mapping of Genetic Variants in BIN1, CLU, CR1 and PICALM for Association with Cerebrospinal Fluid Biomarkers for Alzheimer's Disease" *PLoS ONE* 6:e15918.
30. Hollingworth et al (including **Kauwe JSK**) (2011) Common variants at ABCA7, MS4A6A/MS4A4E, EPHA1, CD33 and CD2AP are associated with Alzheimer's disease. *Nature Genetics* 43:429-435
31. Naj et al (including **Kauwe JSK**) (2011) Common Variants at MS4A4/MS4A6E, CD2AP, CD33 and EPHA1 are associated with late-onset Alzheimer's disease. *Nature Genetics* 43:436-41

\*Co-first authors who contributed equally

### **Book Chapters**

1. **Kauwe JSK**, Goate AM. Molecular genetics of Alzheimer's disease. In: Dawburn D, Allen SJ, eds. *Neurobiology of Alzheimer's Disease*. 3<sup>rd</sup> ed. Oxford; New York: Oxford University Press, 2007:59–79

**Invited Seminars and Oral Presentations**

**American Academy of Neurologists National Meeting.** Honolulu, HI 2011. Fluid biomarkers for early detection and diagnosis of Alzheimer's disease.

**Molecular Life Sciences Symposium.** Brigham Young University, Provo, UT 2011. Progressing from genetic association to causation in complex human disease.

**Epidemiology Seminar.** Utah State University, Logan, UT 2011. Aggregate genetic risk for AD in the Cache County Study.

**Research Revolution.** Orem Public Library, Orem, UT 2011. The genetics basis of Alzheimer's disease.

**LDS Life Science Research Symposium.** Park City, UT 2010. An endophenotype-based approach to understanding the genetics of Alzheimer's disease.

**Epidemiology Seminar.** Utah State University, Logan, UT 2010. Using biomarkers to generate biological hypotheses for disease risk associations.

**Upward Bound.** Utah Valley University, Orem, UT 2010. Paths to a Career in Science.

**Research Revolution.** Orem Public Library, Orem, UT 2010. Understanding the Genetics of Alzheimer's disease.

**Human Genetics Series.** University of Texas, Houston, Texas Medical Center School of Public Health, Houston TX 2010. SNPs associated with cerebrospinal fluid tau levels influence rate of cognitive decline in Alzheimer's disease.

**AP Biology and Chemistry Seminar.** Walden High School, Provo UT 2009. Bioinformatics: Applications and Career Opportunities.

**Network of Minority Research Investigators National Institute of Diabetes and Digestive and Kidney Disease West Regional Meeting.** San Diego, CA 2009. Using cerebrospinal fluid amyloid-beta levels to understand genetic risk for Alzheimer's disease.

**Biology Department Seminar.** University of Hawaii, Honolulu, HI 2008. Alzheimer's disease, Genome-wide association studies and endophenotypes.

**Biology Colloquium.** California State University, Northridge, Northridge, CA 2008. Issues and progress in genetic studies of Alzheimer's disease.

**Brain Institute Seminar.** University of Utah, Salt Lake City, UT 2008. Use of cerebrospinal fluid biomarkers as endophenotypes for genetic studies of Alzheimer's disease.

**Biology Department Seminar.** Brigham Young University, Provo, UT 2008. Variation in *MAPT* shows association with CSF tau levels, tau expression and age at onset of Alzheimer's disease.

**Research Staff Seminar.** Banner Alzheimer's Institute, Phoenix, AZ 2007. Genetic and Proteomic approaches for using cerebrospinal fluid biomarkers to study Alzheimer's disease.

**Neurogenomics Group Meeting.** Translational Genomics, Phoenix, AZ 2007. SNPs previously reported to show association with risk for AD are associated CSF A $\beta$  levels.

**Psychiatry Department Seminar.** Washington University School of Medicine, St. Louis, MO 2007. Use of cerebrospinal fluid A $\beta$  levels as endophenotype for late-onset Alzheimer's disease.

**BioMedRAP Lecture Series.** Washington University, St. Louis, MO 2007. Candidate gene screen identifies polymorphisms associated with AD and CSF A $\beta$  levels.

**O'Leary Award for Research in Neuroscience Finalists Seminar.** Washington University, St. Louis, MO 2007. Use of cerebrospinal fluid A $\beta$  levels as endophenotype for late-onset Alzheimer's disease.

**Public Education Seminar.** Phoenix, AZ 2007. Clinical implications of genetic research on Alzheimer's disease.

**Microbiology and Molecular Biology Department Seminar.** Brigham Young University, Provo, UT 2006. Use of cerebrospinal fluid A $\beta$  levels as endophenotype for late-onset Alzheimer's disease leads to the identification of a causal mutation in PSEN1.

**Biology Department Seminar.** Utah Valley State College, Orem, UT 2006. The use of cerebrospinal fluid amyloid- $\beta$  levels as an endophenotype to study the genetics of late-onset Alzheimer's disease.

**BioMedRAP Lecture Series.** Washington University, St. Louis, MO 2006. Effects of rare variation in PSEN1 on CSF A $\beta$  levels.

**North American Benthological Society Annual Meetings.** Pittsburgh, PA 2002. Using DNA Markers to Investigate Inter-basin Relationships and Dispersal of Stoneflies.

### **Oral Presentations**

**International Conference on Alzheimer's Disease.** Honolulu, HI 2010. Validating predicted biological effects of Alzheimer's disease associated SNPs using CSF biomarker levels.

**International Conference on Alzheimer's Disease.** Chicago, IL 2008. Variation in MAPT is associated with cerebrospinal fluid tau levels in the presence of amyloid deposition.

**American Society of Human Genetics Annual Meetings.** New Orleans, LA 2006. Association study of cerebrospinal fluid amyloid- $\beta$  levels with late-onset Alzheimer's disease associated polymorphisms.

**American Society of Human Genetics Annual Meetings.** Salt Lake City, UT 2005. A Scan of Chromosome 10 Identifies Novel Candidate Genes Showing Strong Association to Late-Onset Alzheimer's Disease.

**Conference of Ford Fellows.** Arlington, VA 2005. Evolution-based Haplotype Analysis Identifies Haplotype Groups with Strong Association to Late-Onset Alzheimer's Disease.

**Genetics Analysis Workshop 14.** Noordwijkerhout, Netherlands 2004. The efficacy of microsatellites vs. SNPs for resolving population structure.

**North American Benthological Society Annual Meetings.** Athens, GA 2003. Phylogenetic and Nested Clade Analysis of the Stonefly *Pteronarcys californica* in the Western United States.

### **Institutional Service Activities**

2010	Undergraduate Curriculum and Mentoring Committee Chair
2010	Computational Biology Faculty Search Committee Chair
2009	Science Education Faculty Search Committee
2009	Freshman Admissions Application Reader
2009	Departmental Curriculum Committee
2009	College Research Committee

### **Other Service Activities**

2010	Advising session for students at Molokai High School, Ho'olehua, HI
2009	Queen's Center (for Pacific Islander Health) Steering Committee
2008	Managing Mentoring Relationships Seminar at WUSM, St. Louis, MO
2008	Advising session for students at Molokai High School, Ho'olehua, HI
2008	Advising with Minority Access to Research Careers students at UH
2008	Advising with Minority Access to Research Careers students at CSUN
2007	NIH Genetics of Alzheimer's Disease Planning Meeting Participant
2004–2008	Washington University DBBS Diversity Steering Committee
2006	Representative of Washington University at the WCBSURC
2006	Poster Judge at the WCBSURC
2005	Representative of Washington University at CIRT conference

2003–2004 Young Scientist Program Evolution Teaching Team Member  
2002–2003 BYUGSA Grant Review Board  
2002 Utah High School Association Science Fair Judge

**Languages**

English Fluent, native speaker  
Japanese Competent speaker, basic level of reading and writing