Sheng Chih Jin, Ph.D.

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EDUCATION

2014	Ph.D., Human and Statistical Genetics, Washington University in St. Louis
2008	ScM, Biostatistics, Johns Hopkins Bloomberg School of Public Health
2004	B.S., Applied Mathematics, National Chiao Tung University, Taiwan

RESEARCH POSITIONS

Apr. 2020- **Tenure-track Assistant Professor, Department of Genetics at Washington University School of Medicine**

June 2018-Mar. 2020 Postdoctoral Fellow, Rockefeller University

Sep. 2014-May 2018 Postdoctoral Fellow, Yale University School of Medicine

Prof. Richard P. Lifton (advisor), Department of Genetics
Prof. Martina Brueckner (co-advisor), Department of Pediatrics (Cardiology)
Prof. Hongyu Zhao (co-advisor), Department of Biostatistics

- **Congenital Heart Disease:** Lead computational biologist for the whole exome sequencing (WES) and array genotyping analysis working groups in the Pediatric Cardiac Genomics Consortium. I developed a novel statistical framework using the expected frequency of *de novo* mutation, controlling for local sequence context, to estimate the expected frequency of extremely rare transmitted variants. This enables comparison of the observed frequency of damaging variants in each gene to the expected value, completely independent of control subjects. I applied this methodology to a WES dataset of 2,871 congenital heart disease (CHD) probands to demonstrate that ~1.8% of cases are attributed to rare transmitted mutations (Jin SC, et al., ***Nature Genetics*** 2017). Novel genes were implicated, including dominant loss of function mutations in the VEGF receptor *FLT4* in tetralogy of Fallot, recessive mutations in *MYH6* in Shone syndrome, and discovery of a recessive founder mutation in *GDF1* Ashkenazim that accounts for ~5% of CHD in this population. In addition, heterozygous *de novo* mutations in 46 different genes encoding chromatin modifiers account for 3% of probands. Sixty-six different genes had two or more *de novo* damaging mutations, with more than 70% of these being disease-causing. There is very high locus heterogeneity, with an estimated 400 different genes causing CHD by *de novo* mutation.
- **Cerebral Palsy:** Co-lead the Bioinformatics group with Drs. Michael Kruer, Jozef Gecz, and Richard Wintle to perform large-scale whole exome/genome sequencing analyses for the International Cerebral Palsy Genomics Consortium. We identified significant enrichment of *de novo* mutations in 249 cerebral palsy cases, which could account for ~12% of patients and revealed significant enrichment of a novel pathway, GTPase signal transduction, in the cerebral palsy pathogenesis via pathway and network analyses (Jin SC, et al., *In revision* ***Nature Genetics***).
- **Rare Mendelian Diseases**: Lead genomic analysis and methodology development in several genetic studies of complex disease, including congenital hydrocephalus, Vein of Galen malformation, Chiari malformation, trigeminal neuralgia, Dent disease, arachnoid cyst, nephrotic syndrome, and congenital hemangioma, for the Yale Center for Mendelian Genomics.

Mar. 2011-Sept. 2014 Graduate Research Assistant, Washington University School of Medicine

Prof. Alison M. Goate (advisor), Department of Genetics
Prof. Carlos Cruchaga (co-advisor), Department of Psychiatry

- Conducted pooled deep-resequencing experiments, and performed genomic/statistical analyses for known Alzheimer's and Parkinson's associated genes to demonstrate that: (1) rare variants in a novel gene *PLD3* significantly confer risk for Alzheimer's disease (AD); (2) rare variants in *TREM2* increase risk for late-onset AD; (3) variants in *TREML2*, a gene adjacent to *TREM2*, protect against late-onset AD; (4) novel causal mutations in *PSEN1*, GRN, and *MAPT* are found in familial AD cases; (5) the effect of *PSEN1*-p.E318G on AD susceptibility is dependent on an interaction

with *APOE*-ε4 alleles and mediated by Aβ deposition and: (6) novel variants in the primary Parkinson's Mendelian and genome-wide association study (GWAS) identified genes that contribute to the Parkinson's disease.

- Performed GWAS using cerebrospinal fluid biomarkers, disease age at onset, brain gene expression, and myeloid cell expression as disease endophenotypes to identify common risk variants in *SPI1* associated with AD.
- Performed in-vitro cell-based functional experiments to show that risk variants in *TREM2* may impart effects by modulating ligand-binding efficiency.

Jan. 2008-Sept. 2008 Research Assistant, Johns Hopkins School of Medicine
Prof. Leslie Cope (advisor), Department of Oncology

- Analyzed data from several microarray experiments using Bioconductor packages in R to implement statistical analyses, including *t*-tests and Cox proportional hazards regression.

Aug. 2007-June 2008 Research Assistant, National Cancer Institute
Prof. Nilanjan Chatterjee (advisor), Division of Cancer Epidemiology and Genetics

- Performed GWAS and developed a novel Omnibus test, which combined diverse but correlated smoking phenotypes for single-marker analysis.

Sept. 2003-June 2004 Undergraduate Research Volunteer, National Chiao Tung University
Prof. Ming-Chih Lao (advisor), Department of Applied Mathematics

- Used theoretical and numerical methods and performed simulations to solve boundary value problems.

PROFESSIONAL EXPERIENCE

July 2008-Aug. 2010 Senior Biostatistician, Johns Hopkins School of Medicine
Prof. Terri H. Beaty (advisor), Department of Epidemiology
Prof. Nae-Yuh Wang (co-advisor), Division of General Internal Medicine
Prof. Thomas A. Louis (co-advisor), Department of Biostatistics

- Served as a statistical lead on experimental design, grant submission, and data analysis for several research studies. Conducted GWAS and gene-environment interaction analyses in over 2,000 parent-offspring trios for the International Oral Cleft Consortium using the Illumina Human genotyping array.

Sept. 2004-Feb. 2006 Lieutenant, Republic of China (Taiwanese) Army

- Served as a CM-11 tank leader for the mandatary military service.

FUNDING AND FELLOWSHIPS

2019- 2024	K99/R00 Pathway to Independence Award, NHLBI, NIH
2019	Postdoctoral Association Career Development Award, Rockefeller University Postdoctoral Association
2018	American Heart Association Postdoctoral Fellowship
2015	James Hudson Brown – Alexander B. Coxe Fellowship, Yale School of Medicine
2014	Howard Hughes Medical Institute Postdoctoral Fellowship, Howard Hughes Medical Institute at Yale School of Medicine
2012	Alzheimer's Association International Conference Travel Fellowship, Alzheimer's Association International Conference
2011-2013	Fellow, Lucille P. Markey Special Emphasis Pathway in Human Pathobiology, Markey Foundation, Washington University School of Medicine

AWARDS

2012	Best Oral Presentation Award, Human & Statistical Genetics Program, Washington University School of Medicine
2007-2008	Departmental Scholarship, Department of Biostatistics, Johns Hopkins
2007	Cancer Research Training Award, National Cancer Institute, NIH
2006	Merica Institute Scholarship
2004	Dean's List (one semester)

PUBLICATIONS

Selected Articles (*Equal contribution)

1. **Jin SC***, Furey CG*, Zeng X, Alloco A, Nelson-Williams C, Karimy JK, Dong W, Ma S, Delpire E, Kahle KT. SLC12A ion transporter mutations in sporadic and familial human congenital hydrocephalus. *In revision* ***Molecular Genetics & Genomic Medicine.*** PMID: 31393094.
2. Duran D*, Zeng X*, **Jin SC***, Choi J*, Nelson-Williams C, Yatsula B, Gaillard J, Furey CG, Lu Q, Timberlake AT, Dong W, Sorscher MA, Loring E, Klein J, Allocco A, Hunt A, Conine S, Karimy JK, Youngblood MW, Zhang J, DiLuna ML, Matouk CC, Mane SM, Tikhonova IR, Castaldi C, López-Giráldez F, Knight J, Haider S, Soban M, Alper SL, Komiyama M, Ducruet AF, Zabramski JM, Dardik A, Walcott BP, Stapleton CJ, Aagaard-Kienitz B, Rodesch G, Jackson E, Smith ER, Orbach DB, Berenstein A, Bilguvar K, Gunel M, Lifton RP, Kahle KT. Mutation in epigenetic modifiers and signaling regulators of neurovascular development in Vein of Galen malformation (2018). ***Neuron***. PMID: 30578106.
3. **Jin SC***, Homsy J*, Zaidi S*, Lu Q, Morton S, DePalma S, Zeng X, Qi H, Chang W, Hung W, Sierant M, Haider S, Zhang J, Knight J, Bjornson R, Castaldi C, Tikhonoa I, Bilguvar K, Mane S, Sanders S, Mital S, Russell M, Gaynor W, Deanfield J, Giardini A, Porter G, Srivastava D, Lo C, Shen Y, Watkins S, Yandell M, Yost J, Tristani-Firouzi M, Newburger J, Roberts A, Kim R, Zhao H, Kaltman J, Goldmuntz E, Chung W, Seidman J, Gelb B, Seidman C, Lifton RP, Brueckner M. (2017). Contribution of rare transmitted and de novo variants among 2,871 congenital heart disease probands. ***Nature Genetics***, 49(11): 1593-1601. PMID: 28991257.
4. Duran D*, **Jin SC***, DeSpenza T Jr*, Nelson-Williams C, Cogal AG, Abrash EW, Harris PC, Lieske JC, Shimshak SJ, Mane S, Bilguvar K, DiLuna ML, Günel M, Lifton RP, Kahle KT. (2016). Digenic mutations of human OCRL paralogs in Dent's disease type 2 associated with Chiari malformation. ***Human Genome Variation***, 3:16042. PMID: 28018608.
5. **Jin SC*,** Carrasquillo MM*, Benitez BA, Skorupa T, Carrell D, Patel D, Lincoln S, Krishnan S, Kachadoorian M, Reitz C, Mayeux R, Wingo TS, Lah JJ, Levey AI, Murrell AI, Hendrie H, Foroud T, Graff-Radford NR, Goate AM, Cruchaga C, Ertekin-Taner N. (2015). *TERM2* is associated with increased risk for Alzheimer's disease in African Americans. ***Molecular Neurodegeneration***, 10; 10:19. PMID: 25886450.
6. **Jin SC**, Benitez BA*, Karch CM*, Cooper B, Skorupa T, Carrell D, Norton JB, Hsu S, Harari O, Cai Y, Bertelsen S, Goate AM, Cruchaga C. (2014). Coding variants in *TREM2* increase risk for Alzheimer's disease. ***Human Molecular Genetics***, 23(21): 5838-5846. PMID: 24899047.
7. Benitez BA*, **Jin SC***, Guerreiro R, Graham R, Lord J, Harold D, Sims R, Lambert JC, Gibbs JR, Bras J, Sassi C, Harari O, Bertelsen S, Lupton MK, Powell J, Bellenguez C, Brown K, Medway C, Haddick PC, 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; 3C Study Group; EADI consortium; Alzheimer's Disease Genetic Consortium; Alzheimer's Disease Neuroimaging Initiative; GERAD Consortium, Williams J, Kauwe JS, Amouyel P, Morgan K, Singleton A, Hardy J, Goate AM, Cruchaga C. (2014). Missense variants in *TREML2* protects against Alzheimer's disease. ***Neurobiology of Aging***, 35(6): 1510.e19-1510.e26. PMID: 24439484.
8. **Jin SC**, Pastor P, Cooper B, Cervantes S, Benitez BA, Razquin C, Goate AM, Ibero-American Alzheimer's Disease Genetics Group Researchers, Cruchaga C. (2012). Poole-DNA sequencing identifies novel causative variants in *PSEN1*, *GRN*, and *MAPT* in a clinical early-onset and familial Alzheimer's disease Ibero-American cohort. ***Alzheimer's Research & Therapy.*** 4(4):34. PMID: 22906081.

Other Published Work (*Equal contribution)

9. **Jin SC***, Lewis S*, Bakhtiari S*, Zeng X*, Sierant MC, Norton B, Corbett M, Elie A, Norton B, Eyk CV, Elie A, Brunstrom-Hernandez J, Papavasileiou A, Fahey MC, Berry JG, Harper K, Zhou C, Magee H, Liu J, Guida BS, Junhui Zhang, Magee H, Li B, Heim J, Webber DL, Frank MS, Xia L, Xu, Y, Castaldi C, Tikhonoa IR, Cho M, Retterer K, Milan F, Wang Y, Waugh JL, Rodan L, Cohen JS, Fatemi A, Lin A, Phillips J, Feyma T, MacLennan SC, Vaughan S, Crompton KE, Reid SM, Reddihough DS, Shang Q, Gao C, Wilson Y, Badawi N, Novak I, McIntyre S, Mane SM, Wang X, Amor D, Zarnescu DC, Lu Q, Xing Q, Zhu C, Bilguvar K, Padilla-Lopez S, Lifton RP, Gecz J, MacLennan AH, Kruer MC. Mutations in genes crucial for neuritogenesis and maturation are a major risk factor for cerebral palsy. *In revision* ***Nature Genetics***

10. Alloco A*, **Jin SC***, Duy PQ*, Furey CG, Zeng X, Dong W, Nelson-Williams C, Karimy JK, DeSpenza T, Hao LT, Reeves B, Haider S, Gunel M, Lifton RP, Kahle KT. Evidence for a recessive inheritance of congenital hydrocephalus with aqueductal stenosis caused by *ATP1A3* compound heterozygous mutations. *Under review* ***Frontiers Cellular Neuroscience***

11. Timberlake AT, **Jin SC**, Nelson-Williams C, Wu R, Furey CG, Islam B, Haider S, Loring E, Galm A, Yale Center for Genome Analysis, Steinbacher D, Larysz D, Staffenberg D, Flores R, Rodriguez E, Boggon TJ, Persing JA, Lifton RP. Damaging *de novo* and transmitted mutations in *TFAP2B* and genes of the BMP, WNT and Hedgehog pathways in syndromic craniosynostosis (2019). ***PNAS.*** PMID: 31292255.

12. Robson A, Makova S, Barish S, Zaidi S, Mehta S, Drozd J, **Jin SC**, Gelb B, Seidman C, Chung WK, Lifton RP, Khokha M, Brueckner M. Core components of the Histone H2B monoubiquitination complex regulate heart development via transcriptional control of cilia motility (2019). ***PNAS***. PMID: 31235600.
13. Chang SJ, **Jin SC**, Jiao X, Galán JE.. Unique features in the intracellular transport of typhoid toxin revealed by a genome-wide screen (2019). ***PLoS Pathogens.*** PMID: 30951565.
14. Helbig K, Laurerer R, Bahr J, Souza I, Myers C, Uysal B, Schwarz N, Gandini M, Huang S, Keren B, Mignot C, Afenjar A, Billette de Villemeur T, Heron D, Nava C, Valence S, Buratti J, Fagerberg C, Soerensen K, Kibaek M, Kamsteeg EJ, Koolen D, Gunning B, Schelhaas HJ, Kruer M, Fox Jordana, Bakhtiari S, Jarrar R, Padilla-Lopez SR, Lindstrom K, **Jin SC**, Zeng X, Bilguvar K, Papavasileiou A, Xing Q, Zhu C, Boysen K, Vairo F, Lanpher B, Klee E, Tilema JM, Payne E, Baker J, Haan E, Smith N, Corbett M, MacLennan A, Gecz J, Biskup S, Goldmann E, Rodan L, Kichula E, Segal E, Jackson K, Asamoah A, Dimmock D, McCarrier J, Botto L, Filloux F, Tvrdik T, Cascino G, Klingerman S, Neumann C, Wang R, Jacobsen J, Nolan M, Snell R, Lehnert K, Sadleir L, Guerrini R, Friez M, Lyons M, Achkar CE, Smith L, Rotenberg A, Poduri A, Sanchis-Juan A, Carss K, Rankin J, Zeman A, Raymond F, Hurles M, Blyth M, Kerr B, Ruiz K, Urquhart J, Hughes I, Banka S, Hedrich U, Scheffer I, Helbig I, Zamponi G, Lerche H, Mefford H. *De novo* pathogenic variants in *CACNA1E* cause developmental and epileptic encephalopathy with congenital contractures, macrocephaly, and dyskinesias. (2018). ***American Journal of Human Genetics***. PMID: 30343943.
15. Furey CG, Zeng X*, Dong W*, **Jin SC**, Choi J, Timberlake AT, Dunbar AM, Allocco AA, Gunel M, Lifton RP, Kahle KT. (2018). Human genetics and molecular mechanisms of congenital hydrocephalus. ***World Neurosurgery***. PMID: 30205212.
16. Furey CG*, Choi J*, **Jin SC**, Zeng X, Timberlake AT, Nelson-Williams C, Mansuri MS, Lu Q, Duran D, Panchagnula S, Alloco A, Karimy JK, Gaillard J, Antwi P, Khanna A, Loring E, Butler WE, Smith ER, Warf BC, Limbrick DD, Storm PB, Heuer G, Iskandar BJ, Johnston JM, Bilguvar K, Mane S, Tikhonova I, Castaldi C, Lopez-Giraldez F, Knight J, Alper SL, Haider S, Guclu B, Bayri Y, Sahin Y, Duncan CC, DiLuna ML, Gunel M, Lifton RP, Kahle KT. (2018). *De novo* mutation in genes regulating neural stem cell fate in human congenital hydrocephalus. ***Neuron.*** PMID: 29983323.
17. Fomchenko E*, Duran D*, **Jin SC**, Dong W, Erson-Omay EZ, Allocco A, Gaillard J, Cord B, Huttner A, Gunel M, DiLuna M, Kahle K. (2018). *De Novo MYH9* mutation in congenital scalp hemangioma. ***Cold Spring Harbor Molecular Case Studies***. PMID: 29903892.
18. Antwi P, Hong CS, Duran D, **Jin SC**, Dong W, DiLuna M, Kahle, K. (2018). A novel association of campomelic dysplasia with hydrocephalus due to an unbalanced chromosomal translocation upstream of SOX9. ***Cold Spring Harbor Molecular Case Studies***. PMID: 29695406.
19. Scholl UI, Stölting G*, Schewe J*, Thiel A, Tan H, Nelson-Williams C, Vichot AA**, Jin SC**, Loring E, Untiet V, Yoo T, Choi J, Xu S, Wu A, Kirchner M, Mertins P, Rump LC, Onder AM, Gamble C, McKenney D, Lash RW, Jones DP, Chune G, Gagliardi P, Choi M, Gordon R, Stowasser M, Fahlke C, Lifton RP. (2018). *CLCN2* chloride channel mutations in familial hyperaldosteronism type II. ***Nature Genetics*****,** 50(3):349-354. PMID: 29403011.
20. Huang KL*, Marcora E*, Pimenova AA, Di Narzo AF, Kapoor M, **Jin SC**, Harari O, Bertelsen S, Fairfax BP, Czajkowski J, Chouraki V, Grenier-Boley B, Bellenguez C, Deming Y, McKenzie A, Raj T, Renton AE, Budde J, Smith A, Fitzpatrick A, Bis JC, DeStefano A, Adams HHH, Ikram MA, van der Lee S, Del-Aguila JL, Fernandez MV, Ibañez L; International Genomics of Alzheimer's Project; Alzheimer's Disease Neuroimaging Initiative, Sims R, Escott-Price V, Mayeux R, Haines JL, Farrer LA, Pericak-Vance MA, Lambert JC, van Duijn C, Launer L, Seshadri S, Williams J, Amouyel P, Schellenberg GD, Zhang B, Borecki I, Kauwe JSK, Cruchaga C, Hao K, Goate AM. (2017). A

common haplotype lowers PU.1 expression in myeloid cells and delays onset of Alzheimer's disease. ***Nature Neuroscience***, 20(8): 1052-1061. PMID: 28628103.

21. Benitez BA, Davis AA, **Jin SC**, Ibanez L, Ortega-Cubero S, Pastor P, Choi J, Cooper B, Perlmutter JS, Cruchaga C. (2016). Resequencing analysis of five Mendelian genes and the top genes from genome-wide association studies in Parkinson's Disease. ***Molecular Neurodegeneration***, 19; 11:29. PMID: 27094865.
22. Song W, Hooli B, Mullin K, **Jin SC**, Cella M, Ulland TK, Wang Y, Tanzi RE, Colonna M. (2016). Alzheimer's disease-associated *TREM2* variants exhibit either decreased or increased ligand-dependent activation. ***Alzheimer's & Dementia***, 13(4): 381-387. PMID:27520774
23. Homsy J*, Zaidi S*, Shen Y*, Ware JS*, Samocha KE, Karczewski KJ, DePalma SR, McKean D, Wakimoto H, Gorham J, **Jin SC**, Deanfield J, Giardini A, Porter GA Jr, Kim R, Bilguvar K, López-Giráldez F, Tikhonova I, Mane S, Romano-Adesman A, Qi H, Vardarajan B, Ma L, Daly M, Roberts AE, Russell MW, Mital S, Newburger JW, Gaynor JW, Breitbart RE, Iossifov I, Ronemus M, Sanders SJ, Kaltman JR, Seidman JG, Brueckner M, Gelb BD, Goldmuntz E, Lifton RP, Seidman CE, Chung WK. (2015). *De novo* mutations in congenital heart disease with neurodevelopmental and other congenital anomalies. ***Science***, 350(6265):1262-1266. PMID: 26785492.
24. Cruchaga C, Karch CM*, **Jin SC***, Benitez BA, Cai Y, Guerreiro R, Harari O, Norton J, Budde J, Bertelsen S, Jeng AT, Cooper B, Skorupa T, Carrell D, Levitch D, Hsu S, Choi J, Ryten M; UK Brain Expression Consortium, Hardy J, Ryten M, Trabzuni D, Weale ME, Ramasamy A, Smith C, Sassi C, Bras J, Gibbs JR, Hernandez DG, Lupton MK, Powell J, Forabosco P, Ridge PG, Corcoran CD, Tschanz JT, Norton MC, Munger RG, Schmutz C, Leary M, Demirci FY, Bamne MN, Wang X, Lopez OL, Ganguli M, Medway C, Turton J, Lord J, Braae A, Barber I, Brown K; Alzheimer's Research UK Consortium, Pastor P, Lorenzo-Betancor O, Brkanac Z, Scott E, Topol E, Morgan K, Rogaeva E, Singleton AB, Hardy J, Kamboh MI, St George-Hyslop P, Cairns N, Morris JC, Kauwe JS, Goate AM. (2014). Rare coding variants in the phospholipase D3 gene confer risk for Alzheimer's disease. ***Nature***, 505(7484): 550-554. PMID: 24336208.
25. 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 Alzheimer's Disease Genetic Consortium, Goate AM. (2013). GWAS of cerebrospinal fluid tau levels identifies risk variants for Alzheimer's disease. ***Neuron***, 78(2):256-268. PMID: 23562540.
26. Benitez BA, Karch CM, Cai Y, **Jin SC**, Cooper B, Carrell D, Bertelsen S, Chibnik L, Schneider JA, Bennett DA; Alzheimer's Disease Neuroimaging Initiative; Genetic and Environmental Risk for Alzheimer's Disease Consortium, Fagan AM, Holtzman DM, Morris JC, Goate AM, Cruchaga C. (2013). The PSEN1, p.E318G variant increases the risk of Alzheimer's disease in *APOE*-ε4 carriers. ***PLoS Genetics***, 9(8): e1003685. PMID: 23990795.
27. Benitez BA, Cooper B, Pastor P, **Jin SC**, Lorenzo E, Cervantes S, Cruchaga C. (2013). *TERM2* is associated with the risk of Alzheimer's disease in Spanish population. ***Neurobiology of Aging***, 34(6): 1711.e15-1711.e17. PMID: 23391427.
28. Patel PJ, Beaty TH, Ruczinski I, Murray JC, Marazita ML, Munger RG, Hetmanski JB, Wu T, Murray T, Rose M, Redett RJ, **Jin SC**, Lie RT, Su-Chou YH, Wang H, Ye X, Yeow V, Chong S, Jee SH, Shi B, Scott AF. (2013). X-linked Markers in the Duchenne Muscular Dystrophy Gene Associated with Oral Clefts. ***European Journal of Oral Sciences***, 121(2): 63-68. PMID: 23489894.
29. Wang H, Zhang T, Wu T, Hetmanski JB, Ruczinski I, Schwender H, Murray T, Fallin MD, Redett RJ, Raymond GV, **Jin SC**, Wu-Chou YH, Chen PK, Yeow V, Chong SS, Cheah FS, Jee SH, Jabs EW, Liang KY, Scott A, Beaty TH. (2013). The *FGF*&*FGFR* gene family and risk of cleft lip with or without cleft palate. ***The Cleft Palate-Craniofacial Journal***, 50(1):96-103. PMID: 22074045.
30. Beaty TH, Ruczinski I, Murray JC, Marazita ML, Munger RG, Hetmanski JB, Murray T, Redett RJ, Fallin MD, Liang KY, Wu T, Patel PJ, **Jin SC**, Zhang TX, Schwender H, Wu-Chou YH, Chen PK, Chong SS, Cheah F, Yeow V, Ye X, Wang H, Huang S, Jabs EW, Shi B, Wilcox AJ, Lie RT, Jee SH, Christensen K, Doheny KF, Pugh EW, Ling H, Scott AF. (2011). Evidence for gene-environment

interaction in a genome wide study of isolated, nonsyndromic cleft palate. ***Genetic Epidemiology***, 35(6):469-478. PMID: 21618603.

31. Beaty TH, Murray JC, Marazita ML**,** Munger RG, Ruczinski I, Hetmanski JB, Liang KY, Wu T, Murray T, Fallin MD, Redett RA, Raymond G, Schwender H, **Jin SC**, Cooper ME, Dunnwald M, Mansilla MA, Leslie E, Bullard S, Lidral AC, Moreno LM, Menezes R, Vieira AR, Petrin A, Wilcox AJ, Lie RT, Jabs EW, Wu-Chou YH, Chen PK,.Wang H, Ye X, Huang S, Yeow V, Chong SS, Jee SH, Shi B, Christensen K, Melbye M, Doheny KF, Pugh EW, Ling H, Castilla EE, Czeizel AE, Ma L, Field LL, Brody L. Pangilinan F, Mills JL, Molloy AM, Kirke PN, Scott JM, Arcos-Burgos M, Scott AF. (2010). A genome-wide association study of cleft lip with and without cleft palate identifies risk variants near *MAFB* and *ABCA4*. ***Nature Genetics***, 42(6):525-529. PMID: 20436469.
32. Caporaso N*, Gu F*, Chatterjee N*, **Jin SC**, Yu K, Yeager M, Chen C, Jacobs K, Wheeler W, Landi MT, Ziegler RG, Hunter DJ, Chanock S, Hankinson S, Kraft P, Bergen AW. (2009). Genome-wide and candidate gene association study of cigarette smoking behavior. ***PLoS ONE***, 4(2):e4653. PMID: 19247474.

Book Chapter

1. **Jin SC**, Benitez BA, Deming Y, Cruchaga C. Pooled-DNA sequencing for elucidation of genomic risk factors/rare variants underlying Alzheimer's disease (2016). ***The Systems Biology of Alzheimer's Disease: Methods and Protocols***. Springer.

Reviews

1. Zeng X, Hunt A, **Jin SC**, Duran D, Gaillard J, Kahle KT. EphrinB2-EphB4-RASA1 signaling in human cerebrovascular development and disease (2019). ***Trends in Molecular Medicine.***

SELECTED/INVITED TALKS

Jan. 2019	**Invited talk**, Waisman Center, University of Wisconsin - Madison
Jan. 2019	**Invited talk**, Mindich Child Health and Development Institute, Icahn School of Medicine at Mount Sinai
Dec. 2018	**Invited talk**, National Taiwan University College of Medical Institute of Medical Genomics and Proteomics
Nov. 2018	**Invited talk**, Institute for Genomic Medicine, Nationwide Children's Hospital
Nov. 2018	**Invited talk**, Eugene McDermott Center for Human Growth and Development, University of Texas Southwestern Medical Center
Sept. 2018	**Invited talk**, Department of Genetics, Washington University in St. Louis
June 2018	**Invited webinar**, Rare Disease Seminar Series
April 2018	**Invited keynote presentation**, The 2nd International Cerebral Palsy Genomics Consortium Conference, Zhengzhou, China
Dec. 2017	**Invited talk**, Institute of Biomedical Sciences, Academia Sinica, Taiwan
Oct. 2017	**Selected talk**, NHLBI Bench to Bassinet Program Annual Face-To-Face Conference, Rockville, MD
Dec. 2016	**Selected talk**, NHLBI Bench to Bassinet Program Annual Face-To-Face Conference, Rockville, MD
Feb. 2016	**Invited talk**, Institute of Biomedical Sciences, Academia Sinica, Taiwan
July 2014	**Selected talk**, Alzheimer's Association International Conference, Copenhagen, Denmark
July 2012	**Selected talk**, Alzheimer's Association International Conference, Vancouver, Canada

TEACHING AND MENTORING

Sept. 2014-Present	While Dr. Richard Lifton moved on September 1, 2016 to assume the presidency of Rockefeller University, I trained and helped supervise 3 graduate students, 3 medical students, and 2 postdoctoral fellows in the Lifton lab to conduct research projects, perform whole exome/genome sequencing genomic analyses, downstream statistical algorithms, and mentored them on writing proposals and career development.
Sept. 2011–Feb 2012	**Teaching Assistant**, Washington University in St. Louis • Presented lectures; assisted in quiz and exam preparation for a graduate course "Human Linkage and Association"

Sept. 2007–May 2008 **Teaching Assistant**, Johns Hopkins University

- Presented lectures; assisted in quiz and exam preparation for a graduate course “Statistical Methods in Public Health"

PROFESSIONAL SERVICE

2013-Present Ad hoc reviewer for *BMC Neurology*, *Journal of Alzheimer’s Disease, Molecular Neurodegeneration, Alzheimer’s & Dementia*, *European Heart Journal*

2013-Present Review editor for *Frontiers in Genetics, section Neurogenomic*