Curriculum Vitae WUSM Format Sheng Chih Jin, Ph.D.

Date: 10/11/2023

Name Jin, Sheng Chih

Address, Telephone and email:

Washington University School of Medicine Jin Lab, Department of Genetics 660 South Euclid Avenue, Campus Box 8232 St. Louis MO, 63110-1010

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Present Position:

April 2020 - Present, Assistant Professor of Genetics and Pediatrics

Education:

National Chiao Tung University, Hsinchu, Taiwan, 2000 – 2004 B.S., Applied Mathematics

Johns Hopkins Bloomberg School of Public Health, Baltimore, MD, USA, 2006 – 2008 ScM, Biostatistics

Washington University School of Medicine, St. Louis, MO, USA, 2010 – 2014 Ph.D., Human & Statistical Genetics (Advisors: Alison Goate and Carlos Cruchaga)

Yale School of Medicine, New Haven, CT, USA, 2014 – 2018 Postdoctoral Fellow (Advisors: Richard P. Lifton, Martina Brueckner, and Hongyu Zhao)

Rockefeller University, New York, NY, USA, 2018 – 2020 Postdoctoral Fellow (Advisors: Richard P. Lifton, Martina Brueckner, and Hongyu Zhao)

Academic Positions / Employment:

2020 – Assistant Professor of Genetics and Pediatrics Washington University School of Medicine

Honors and Awards:

2007	Cancer Research Training Award, Biostatistics Branch, Division of Cancer Epidemiology & Genetics, National Cancer Institute, NIH
2007	Departmental Scholarship, Department of Biostatistics, Johns Hopkins University
2011	Lucille P. Markey Special Emphasis Pathway in Human Pathobiology Fellowship, Markey Foundation, Washington University School of Medicine
2012	Alzheimer's Disease International Conference Travel Fellowship, Alzheimer's Association
2012	Best Oral Presentation Award, Human and Statistical Genetics Program 2012 Retreat
2014	Finalist, Fourth Annual Hope Center Retreat Poster Session, Hope Center for Neurological Disorders, Washington University School of Medicine
2014	Howard Hughes Medical Institute Postdoctoral Fellowship, Department of Genetics, Yale University School of Medicine
2015	James Hudson Brown – Alexander Brown Coxe Postdoctoral Fellowship in the Medical Sciences, Yale University School of Medicine
2018	American Heart Association Postdoctoral Fellowship
2019	NIH/NHLBI K99/R00 Pathway to Independence Award
2019	Postdoctoral Association Career Development Award, Rockefeller University
2020	Rockefeller University Nominee, Blavatnik Regional Award for Young Scientists
2021	Children's Discovery Institute Faculty Scholar, St. Louis Children's Hospital
2021	Hydrocephalus Association Innovator Award
2022	Pediatric Cardiac Genomics Consortium and Cardiovascular Development Data Resource Center Challenge Prize

Editorial Responsibilities:

- 2013 Ad Hoc Reviewer, Trends in Genetics, Genome Research, European Heart Journal, npj Genomic Medicine, Brain, Molecular Neurodegeneration, BMC Neurology, Journal of Alzheimer's Disease, Alzheimer's & Dementia, Genes, Journal of Medical Genetics, Biomolecules, STAR Protocols, Journal of Personalized Medicine
- 2013 **Review Editor**, Frontiers in Genetics, Neurogenomics Section

National Panels, Committees, Boards:

N/A

Community Service Contributions:

☐ University Appointments and Committees

- 2020 **Member**, DBBS Admissions Committee B, Washington University School of Medicine
- 2023 **Coordinator**, Hope Center Monday Noon Seminars

☐ Professional Societies and Organizations

- 2011 **Member**, American Society of Human Genetics
- 2015 **Member**, American Heart Association
- 2023 **Planning Committee**, Hydrocephalus Association & Rudi Schulte Research Institute Research Workshop

Major Invited Professorships and Lectureships:

2012	Selected Oral Presentation, Alzheimer's Association International
	Conference (from submitted abstract), Vancouver, Canada
2014	Selected Oral Presentation, Alzheimer's Association International
	Conference (from submitted abstract), Copenhagen, Denmark
2016	Invited Talk, Institute of Biomedical Sciences, Academia Sinica, Taiwan
2017	Selected Oral Presentation, NHLBI Bench to Bassinet Program Annual
	Face-to-Face Meeting, Rockville, MD
2017	Invited Talk, Institute of Biomedical Sciences, Academia Sinica, Taiwan
2017	Invited Keynote Presentation, International Cerebral Palsy Genomics
	Consortium Conference, Zhengzhou, China
2018	Invited Talk, Department of Genetics, Washington University School of
	Medicine
2018	Invited Talk, Eugene McDermott Center for Human Growth and
	Development, University of Texas Southwestern Medical Center
2018	Invited Talk, Institute for Genomic Medicine, Nationwide Children's
	Hospital
2018	Invited Talk, National Taiwan University College of Medical Institute of
	Medical Genomics and Proteomics
2019	Invited Talk, Waisman Center, University of Wisconsin – Madison
2019	Invited Talk, Mindich Child Health and Development Institute, Icahn
	School of Medicine at Mount Sinai
2019	Invited Talk, International Cerebral Palsy Genomics Consortium
	Conference, Anaheim, CA
2020	Selected Oral Presentation , American Society of Human Genetics (from
	submitted abstract), Virtual Meeting
2021	Invited Talk, Mount Sinai x Open Box Science Computational Omics
	Seminar
2021	Invited Talk, Washington University School of Medicine, Pediatric
	Neurology Research Working Group

2021	Invited Talk, Washington University School of Medicine, Division of
	Genetics and Genomic Medicine
2021	Invited Talk, Boston Taiwanese Biotechnology Association Monthly
	Seminar Series
2021	Invited Talk, Washington University Department of Developmental
	Biology Seminar Series
2021	Invited Talk, Washington University Department of Computer Science &
	Engineering Colloquia Series
2022	Invited Talk, Washington University MSTP Future of Medicine Seminar
2022	Invited Talk, Washington University School of Medicine, Center for
	Cardiovascular Research Seminar Series
2022	Invited Talk, Hydrocephalus Association Network for Discovery Science
	Webinar Series
2023	Invited Talk, Genomic Information Commons Working Group Meeting
2023	Invited Talk, Washington University Intellectual and Developmental
	Disabilities Research Center Inaugural Symposium
2023	Invited Talk, Hydrocephalus Association & Rudi Schulte Research
	Institute Research Workshop

Consulting Relationships and Board Memberships:

N/A

Internal Review Work

2021	Ad Hoc Reviewer, Clinical and Translational Research Funding Program,
	Washington University Institute of Clinical and Translational Sciences
2022	Ad Hoc Reviewer, NGI Pilot Awards, Washington University
	NeuroGenomics and Informatics Center
2022	Ad Hoc Reviewer, Clinical and Translational Research Funding Program,
	Washington University Institute of Clinical and Translational Sciences

External Review Work

2022	Grant Reviewer , Hydrocephalus Association Innovator Award
2023	Ad Hoc Reviewer, NIH, Cardiovascular and Respiratory Diseases Study
	Section
2023	Grant Reviewer, Hydrocephalus Association Innovator Award
2023	Grant Reviewer , Sidra Medicine Precision Medicine Challenge Award
	(IRF 24)
2023	Ad Hoc Reviewer, NIH ZMH1 ERB-S (02) S - Data Analysis and
	Coordination Center for the PsychENCODE Consortium (U24)

Research Support

□ Governmental

Title: WashU Somatic Mosaicism across Human Tissues (SMaHT) Program.

Organizational Center Agency: NIH U24NS132103 Role: Co-Investigator

Duration: 4/15/2023-03/31/2028 Amount: \$7.470.939 total costs

Title: WashU-VAI Somatic Mosaicism across Human Tissues (SMaHT) Program.

Genome Characterization Center Agency: NIH UM1DA058219

Role: Co-Investigator

Duration: 5/01/2023-04/30/2028 Amount: \$1,499,999 total costs

Title: INTERCEPT: Integrated Research Center for Human Pain Tissues

Agency: NIH/NINDS U19NS130607

Role: Co Principal Investigator (Project 1: Milbrandt/DiAntonio/Jin)

Duration: 12/01/2022-11/30/2027 Amount: \$3,644,291 Project 1 total costs

Title: INTERCEPT: Integrated Research Center for Human Pain Tissues

Agency: NIH/NINDS U19NS130607

Role: Co Principal Investigator (Data Core: Jin/Zhao)

Duration: 12/01/2022–11/30/2027 Amount: \$694,321 Data Core total costs

Title: Genomic analysis of the Multiplex, Autozygous Populations in Cerebral Palsy

(MAP CP) cohort: a focused approach to a complex disease

Agency: NIH/NINDS R01NS127108

Role: Co-Investigator

Duration: 02/01/2023-01/31/2028

Amount: \$421,321 total costs

Title: Human Genetics and Molecular Mechanisms of Congenital Hydrocephalus

Agency: NIH/NINDS 1R01NS1111029

Role: Co-Investigator

Duration: 04/01/2020 – 01/31/2025 Amount: \$199,706 total sub costs

Title: Human Genetics and Molecular Mechanisms of Vein of Galen Malformation

Agency: NIH/NINDS 1R01NS117609

Role: Co-Investigator (Subaward to Yale University)

Duration: 07/01/2020 – 06/30/2024 Amount: \$172,000 total sub costs

Title: Genetic Risk Factors for Severe Scoliosis

Agency: NIH/NIAMS 2R01AR067715

Role: Co-Investigator

Duration: 07/01/2020 - 06/30/2024

Amount: \$3,248,850 total costs (Salary Support only)

Title: Genomic Insights into the Neurobiology of Cerebral Palsy

Agency: NIH/NINDS 5R01NS106298

Role: Co-Investigator

Duration: 04/01/2019 – 12/31/2023 Amount: \$19,770 total sub costs

Title: Integrative Genomic Analysis of Congenital Heart Disease

Agency: NIH/NHLBI 4R00HL143036

Role: Principal Investigator

Duration: 04/01/2020 - 03/31/2023

Amount: \$730,167 total costs

■ Non-Governmental

Title: Discovery of novel genetic variations in cerebral palsy by whole genome

sequencing

Agency: Cerebral Palsy Alliance Research Foundation

Role: Principal Investigator

Duration: 06/01/2022 - 05/31/2027

Amount: \$225,000

Title: A Genome-Wide Assessment of Noncoding Risk Variants in Congenital

Hydrocephalus

Agency: Hydrocephalus Association

Role: Principal Investigator

Duration: 12/31/2021 - 12/30/2022

Amount: \$50,000

Title: Human Genetics and Molecular Mechanisms of Cerebral Palsy Agency: Children's Discovery Institute - St. Louis Children's Hospital

Role: Principal Investigator

Duration: 10/01/2021 - 09/30/2026

Amount: \$300,000

Title: Long-Read Genome Sequencing and Integrative Genomic Analysis for Cerebral

Palsy

Agency: CTRFP - Washington University Institute of Clinical and Translational Sciences

Role: Principal Investigator

Duration: 03/01/2021 - 02/28/2022

Amount: \$50,000

Title: Functional characterization of the Diaph1 gene using the zebrafish knock

model

Agency: Children's Discovery Institute - St. Louis Children's Hospital

Role: Principal Investigator

Duration: 02/05/2021 - 06/30/2023 (No cost extension)

Amount: \$10,000

Pending

Title: Molecular and cellular characterization of congenital hydrocephalus

Agency: NIH/NINDS 1R01NS131610A

Role: Principal Investigator

Duration: 04/01/2024 - 03/30/2029

Percentile: 9%

Undergraduate Mentoring:

Trainee	Period	Project Title	Support	Current
				Position
Joshipura,	6/2021	Human genetics of	Mount Holyoke	BS Student
Kareena	_	cerebral palsy	College's Lynk	Mount
	8/2021		Fellowship	Holyoke
			_	College
Wrubel, Max	11/2021	Human genetics of	Post-	Bioinformatici
	_	cerebral palsy	Baccalaureate	an
	7/2022		Extensive Study	Mount Sinai
	-		Program	
Marcial-	6/2022	Human genetics of	MGI OGR Summer	BS Student St.
Rodriguez,	_	congenital	Undergraduate	Olaf College
Athziri	8/2022	hydrocephalus	Scholars Program	
Shelton, Cabria	6/2022	Human genetics of	MGI OGR Summer	BS Student
	_	patent ductus	Undergraduate	Rhodes College
	8/2022	arteriosus	Scholars Program	
Iyiyol, Tugce	8/2022	Role of transposable		BS Student
	_	elements in rare		WUSTL
		pediatric movement		
		disorders		
Ruttenberg,	8/2022	Role of structural		BS Student
Andrew	_	variation in rare		WUSTL
		pediatric movement		
		disorders		

Graduate Mentoring:

Trainee	Period	Project Title	Support	Current Position
Ulibarri, Jenna	7/2023	Proteogenomics in peripheral neuropathy	Start-up	PhD Student WUSTL
Tang, Zitian	5/2023	Impact of repeat expansion in peripheral neuropathy	Start-up	PhD Student WUSTL
Dong, Wendy (joint with Jeffrey Milbrandt)	3/2023 -	Functional genetics of peripheral neuropathy	WashU T32	MSTP Candidate WUSTL
Zhao, Shujuan (joint with Kris Kahle at MGH)	9/2020 -	Human genetics of Vein of Galen Malformation	R01NS117609 + R00HL143036 + Markey Pathway	PhD Candidate WUSTL
Choi, Julie (joint with Jeffrey Milbrandt)	4/2022 -	Human genetics of peripheral neuropathy	WashU T32GATP	PhD Candidate WUSTL
Kong, Nahyun	4/2022	Human genetics of rare movement disorders	Start-up + Study Abroad Scholarships from the Mogam Science Scholarship Foundation	PhD Candidate WUSTL
Qiu, Tian (Devin)	7/2022- 12/2022	Human genetics of rare movement disorders	R00HL143036	PhD Student Van Andel Institute
Peters, Samuel	5/2020 - 4/2021	Human genetics of primary Moyamoya disease	R00HL143036	MS Student SLU
King, Spencer	5/2020 - 5/2021	Human genetics of cerebral palsy	R00HL143036	Data Scientist Geneoscopy
Yu, Xiaobing	2/2021 - 11/2021	Single-cell RNA- sequencing analysis for rare neurological disorders	R00HL143036	PhD Student WUSTL
Shaffiey, Shohaib	2/2021 - 5/2021	Whole genome sequencing analysis for rare neurological disorders	R00HL143036	MS Student WUSTL

Postgraduate Mentoring:

Trainee	Period	Project Title	Support	Current Position
Wang, Yung-Chun	6/2021	Human genetics of	R00HL143036 +	Postdoctoral
	_	cerebral palsy	R01NS127108	Fellow

Fellowships/Scholarships/Grants to Postdocs/Students:

Scholarships to attend Cold Spring Harbor Laboratory's Scientific Writing Retreat

Agency: Cold Spring Harbor Laboratory Postdoc(s)/Student(s): Yung-Chun Wang

Role: Sponsor Duration: 10/2023 Amount: \$500

Precision Medicine Pathway

Agency: Washington University School of Medicine

Postdoc(s)/Student(s): Zitian Tang

Role: Sponsor

Duration: 08/2023 - 08/2025

Amount: \$0

Scholarships to attend Jackson Laboratory's Human and Mammalian Genetics and

Genomics: The McKusick Short Course

Agency: Jackson Laboratory

Postdoc(s)/Student(s): Zitian Tang

Role: Sponsor
Duration: 07/2023
Amount: \$500

Study Abroad Scholarships

Agency: Mogam Science Scholarship Foundation

Postdoc(s)/Student(s): Nahyun Kong

Role: Sponsor Duration: 1/2023

Amount: \$10,000 (one-time allowance)

Washington University's T32 Genome Analysis Training Program

Agency: Washington University School of Medicine

Postdoc(s)/Student(s): Julie Choi

Role: Sponsor

Duration: 10/2022 - 09/2023

Amount: \$34,500/year

Lucille P. Markey Special Emphasis Pathway in Human Pathobiology Fellowship

Agency: Washington University School of Medicine

Postdoc(s)/Student(s): Shujuan Zhao

Role: Sponsor

Duration: 08/2022 - 08/2024

Amount: \$4,000 (one-time stipend supplement)

Precision Medicine Pathway

Agency: Washington University School of Medicine

Postdoc(s)/Student(s): Julie Choi

Role: Sponsor

Duration: 08/2022 - 08/2024

Amount: \$0

Precision Medicine Pathway

Agency: Washington University School of Medicine

Postdoc(s)/Student(s): Nahyun Kong

Role: Sponsor

Duration: 08/2022 - 08/2024

Amount: \$0

Scholarships to attend Jackson Laboratory's Human and Mammalian Genetics and

Genomics: The McKusick Short Course

Agency: Jackson Laboratory

Postdoc(s)/Student(s): Shujuan Zhao

Role: Sponsor Duration: 07/2022 Amount: \$900

Scholarships to attend Jackson Laboratory's Human and Mammalian Genetics and

Genomics: The McKusick Short Course

Agency: Jackson Laboratory

Postdoc(s)/Student(s): Max Wrubel

Role: Sponsor Duration: 07/2022 Amount: \$1,800

Center of Regenerative Medicine Postdoctoral Fellowship

Title: Human genomics and molecular mechanisms of Sporadic Moyamoya disease

Agency: Washington University Center of Regenerative Medicine

Postdoc(s)/Student(s): Yung-Chun (David) Wang

Role: Sponsor

Duration: 07/01/2021

Amount: \$10,000 (signing bonus)

NIH TL1 Predoctoral Clinical Research Training Fellowship (Declined)

Title: Integrative Genomic Analysis of Cerebral Palsy and Rare Pediatric Movement

Disorders

Agency: Washington University Clinical Research Training Center

Postdoc(s)/Student(s): Amar Sheth

Role: Sponsor

Duration: 06/01/2021 - 05/31/2022

Amount: \$2,110/month

Thesis Committee Advisees:

Ciyang Wang DBBS Molecular Genetics and Genomics Chengran Yang DBBS Human and Statistical Genetics

Tong Wu Biomedical Engineering

Caitlin Dingwall WashU MSTP

Kuangying Yang
Gervette M. Penny
Travis Law

DBBS Human and Statistical Genetics
DBBS Molecular Genetics and Genomics
DBBS Computational and Systems Biology

Qualifying Exam Committee:

Ji-Sun Kwon DBBS Computational and Systems Biology
Evelyn Craigen DBBS Molecular Genetics and Genomics (Chair)

Dan Western DBBS Human and Statistical Genetics Kuangying Yang DBBS Human and Statistical Genetics

Grace Cooper DBBS Human and Statistical Genetics (Chair)

Paul Lee WashU MSTP (Chair)

Juanru Guo

Mariam Khanfar

Chia-Jung Lee

Chien-Wei Peng

DBBS Computational and Systems Biology

DBBS Human and Statistical Genetics (Chair)

DBBS Computational and Systems Biology

DBBS Human and Statistical Genetics

Patents:

N/A

Teaching Responsibilities:

2021 – 2022 Lecturer, Bio5488: Genomics, Washington University School of Medicine

2021 – 2022 Study Section Co-Leader, Bio5491: Advanced Genetics, Washington University School of Medicine

2022 - Lecturer, Bio5487: Genetics & Genomics of Disease, Washington

University School of Medicine

- 2022 Co-director, Bio5488: Genomics, Washington University School of Medicine
- 2023 Immersion Program Leader, Washington University School of Medicine
- 2023 Lecturer, Bio5285: Current Topics in Human and Mammalian Genetics, Washington University School of Medicine

Bibliography:

□ Peer Reviewed Manuscripts (* Equal contribution; # Co-corresponding; Lab members in bold)

- 1. 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.
- 2. 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.
- 3. 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.
- 4. **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.
- 5. 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.
- 6. 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.
- 7. 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.
- 8. 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.
- 9. 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.
- 10. 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.
- 11. **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.
- 12. 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.
- 13. **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.
- 14. 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.
- 15. 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.
- 16. 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.
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