

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

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Name Jin, Sheng Chih

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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 for 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 for Frontiers in Genetics, Neurogenomics Section

National Panels, Committees, Boards:

N/A

Community Service Contributions:

- **University Appointments and Committees**

2020 – DBBS Admissions Committee B, Washington University School of Medicine

- **Professional Societies and Organizations**

2011 – American Society of Human Genetics (Member)

2015 – American Heart Association (Member)

Major Invited Professorships and Lectureships:

2012 Alzheimer's Association International Conference (from submitted abstract), Vancouver, Canada

2014 Alzheimer's Association International Conference (from submitted abstract), Copenhagen, Denmark

2016 Institute of Biomedical Sciences, Academia Sinica, Taiwan

2017 NHLBI Bench to Bassinet Program Annual Face-to-Face Meeting, Rockville, MD

2017 Institute of Biomedical Sciences, Academia Sinica, Taiwan

2017 International Cerebral Palsy Genomics Consortium Conference (invited keynote presentation), Zhengzhou, China

2018 Department of Genetics, Washington University School of Medicine

2018 Eugene McDermott Center for Human Growth and Development, University of Texas Southwestern Medical Center

2018 Institute for Genomic Medicine, Nationwide Children's Hospital

2018 National Taiwan University College of Medical Institute of Medical Genomics and Proteomics

2019 Waisman Center, University of Wisconsin – Madison

2019 Mindich Child Health and Development Institute, Icahn School of Medicine at Mount Sinai

2019 International Cerebral Palsy Genomics Consortium Conference (invited presentation), Anaheim, CA

2020 American Society of Human Genetics (from submitted abstract), Virtual Meeting

2021 Mount Sinai x Open Box Science Computational Omics Seminar

2021 Washington University School of Medicine, Pediatric Neurology Research Working Group

2021 Washington University School of Medicine, Division of Genetics and Genomic Medicine

2021 Boston Taiwanese Biotechnology Association Monthly Seminar Series

2021 Washington University Department of Developmental Biology Seminar Series

2021 Washington University Department of Computer Science & Engineering Colloquia Series

2022	Washington University MSTP Future of Medicine Seminar
2022	Washington University School of Medicine, Center for Cardiovascular Research Seminar Series
2022	Hydrocephalus Association Network for Discovery Science Webinar Series

Consulting Relationships and Board Memberships:

N/A

Internal Review Work

2021	Ad hoc reviewer for the Clinical and Translational Research Funding Program, Washington University Institute of Clinical and Translational Sciences
2022	Ad hoc reviewer for the NGI Pilot Awards, Washington University NeuroGenomics and Informatics Center
2022	Ad hoc reviewer for the Clinical and Translational Research Funding Program, Washington University Institute of Clinical and Translational Sciences

External Review Work

2022	Hydrocephalus Association Innovator Award Grant Reviewer
2023	NIH, Cardiovascular and Respiratory Diseases Study Section, Ad Hoc Reviewer

Research Support

- **Governmental**

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: Integrative Genomic Analysis of Congenital Heart Disease
 Agency: NIH/NHLBI 4R00HL143036
 Role: Principal Investigator

Duration: 04/01/2020 – 03/31/2023
 Amount: \$249,005/year

Title: Human Genetics and Molecular Mechanisms of Congenital Hydrocephalus
 Agency: NIH/NINDS 1R01NS1111029
 Role: Co-Investigator
 Duration: 04/01/2020 – 01/31/2025
 Amount: \$41,678 (Year 3)

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: \$39,321 (Year 3)

Title: Genetic Risk Factors for Severe Scoliosis
 Agency: NIH/NIAMS 2R01AR067715
 Role: Co-Investigator
 Duration: 07/01/2020 – 06/30/2024
 Amount: \$10,966 (Year 3)

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 (Year 5)

- **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

Undergraduate Mentoring:

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

Graduate Mentoring:

Trainee	Period	Project Title	Support	Current Position
Wendy Dong (joint with Jeffrey Milbrandt)	3/2023 –	Functional genetics of peripheral neuropathy	U19NS130607	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
Julie, Choi (joint with Jeffrey Milbrandt)	4/2022 –	Human genetics of peripheral neuropathy	WashU T32GATP	PhD Candidate WUSTL
Nahyun, Kong	4/2022 –	Human genetics of rare movement disorders	R00HL143036 + Study Abroad Scholarships from the Mogam Science Scholarship Foundation	PhD Student WUSTL
Peters, Samuel	5/2020 – 4/2021	Human genetics of primary Moyamoya disease	4R00HL143036-02	MS Student SLU
King, Spencer	5/2020 – 5/2021	Human genetics of cerebral palsy	4R00HL143036-02	Data Scientist at Geneoscopy
Yu, Xiaobing	2/2021 – 11/2021	Single-cell RNA-sequencing analysis for rare neurological disorders	4R00HL143036-02	MS Student WUSTL
Shaffiey, Shohaib	2/2021 – 5/2021	Whole genome sequencing analysis for rare neurological disorders	4R00HL143036-02	MS Student WUSTL

Postgraduate Mentoring:

Trainee	Period	Project Title	Support	Current Position
Wang, Yung-Chun	6/2021 –	Human genetics of cerebral palsy	4R00HL143036-02 + CDI Faculty Scholar	Postdoctoral Fellow

Fellowships/Scholarships/Grants to Postdocs/Students:

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 – 09/2025

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	DBBS Human and Statistical Genetics
Travis Law	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)

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

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- ϵ 4 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.
 17. 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.
 18. 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, Tikhonova 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.
 19. 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.

20. 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.
21. 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*, 4(3):a002766.
22. 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*, 4(4):a002998.
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• Book Chapters

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• Reviews

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- **Spotlight**

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