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
Phone: (314) 362-4379
Email: jin810@wustl.edu

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, 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	Bioinformatician Mount Sinai
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
Iyiyol, Tugce	8/2022 –	Role of transposable elements in rare pediatric movement disorders		BS Student WUSTL
Ruttenberg, Andrew	8/2022 –	Role of structural variation in rare pediatric movement disorders		BS Student WUSTL

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 cerebral palsy	R00HL143036 + R01NS127108	Postdoctoral 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

Washington University's T32 Genome Analysis Training Program
Agency: Washington University School of Medicine
Postdoc(s)/Student(s): Wendy Dong
Role: Co-sponsor
Duration: 09/2023 – 08/2024
Amount: $34,500/year

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: Co-sponsor
Duration: 10/2022 – 09/2024
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
Gervette M. Penny	DBBS Molecular Genetics and Genomics
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)
Paul Lee	WashU MSTP (Chair)
Juanru Guo	DBBS Computational and Systems Biology
Mariam Khanfar	DBBS Human and Statistical Genetics (Chair)
Chia-Jung Lee	DBBS Computational and Systems Biology
Chien-Wei Peng	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.
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, 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.
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.
23. 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***, 99(2):302-314.e4.
24. 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***, 119:441-443.
25. 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***, 103(5):666-678.
26. 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 (2019). ***Neuron***, 101(3):429-443.e4.
27. 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***, 15(4):e1007704.
28. **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 (2019). ***Molecular Genetics & Genomic Medicine***, 7(9):e892.
29. 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. Recessive inheritance of congenital hydrocephalus with other structural brain abnormalities caused by compound heterozygous mutations in *ATP1A3* (2019). ***Frontiers Cellular Neuroscience***, 13:425.
30. 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***, 116(28):14049-14054.
31. 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***, 116(30):15116-15121.
32. **Jin SC***, Lewis SA*, Bakhtiari S*, Zeng X*, Sierant MC, Shetty S, Nordlie SM, Elie A, Corbett MA, Norton BY, van Eyk CL, Haider S, Guida BS, Magee H, Liu J, Pastore S, Vincent JB, Brunstrom-Hernandez J, Papavasileiou A, Fahey MC, Berry JG, Harper K, Zhou C, Zhang J, Li B, Heim J, Webber DL, Frank MSB, Xia L, Xu Y, Zhu D, Zhang B, Sheth AH, Knight JR, Castaldi C, Tikhonova IR, López-Giráldez F, Keren B, Whalen S, Buratti J, Doummar D, Cho M, Retterer K, Millan F, Wang Y, Waugh JL, Rodan L, Cohen JS, Fatemi A, Lin AE, Phillips JP, Feyma T, MacLennan SC, Vaughan S, Crompton KE, Reid SM, Reddihough DS, Shang Q, Gao C, Novak I, Badawi N, Wilson YA, McIntyre SJ, Mane SM, Wang X, Amor DJ, Zarnescu DC, Lu Q, Xing Q, Zhu C, Bilguvar K, Padilla-Lopez S, Lifton RP, Gecz J, MacLennan AH, Kruer MC. Mutations disrupting neuritogenesis genes confer risk for cerebral palsy (2020). ***Nature Genetics***, 52(10):1046-1056.
33. Wagner M, Lévy J, Jung-Klawitter S, Bakhtiari S, Monteiro F, Maroofian R, Bierhals T, Hempel M, Elmaleh-Bergès M, Kitajima JP, Kim CA, Salomao JG, Amor DJ, Cooper MS, Perrin L, Pipiras E, Neu A, Doosti M, Karimiani EG, Toosi MB, Houlden H, **Jin SC**, Si YC, Rodan LH, Venselaar H, Kruer MC, Kok F, Hoffmann GF, Strom TM, Wortmann SB, Tabet AC, Opladen T. Loss of TNR causes a nonprogressive neurodevelopmental disorder with spasticity and transient opisthotonos (2020). ***Genetics in Medicine***, 22(6):1061-1068.
34. Dong W*, **Jin SC***, Allocco A*, Zeng X*, Sheth AH, Panchagnula S, Castonguay A, Lorenzo LÉ, Islam B, Brindle G, Bachand K, Hu J, Sularz A, Gaillard J, Choi J, Dunbar A, Nelson-Williams C, Kiziltug E, Furey CG, Conine S, Duy PQ, Kundishora AJ, Loring E, Li B, Lu Q, Zhou G, Liu W, Li X, Sierant MC, Mane S, Castaldi C, López-Giráldez F,

Knight JR, Sekula RF Jr, Simard JM, Eskandar EN, Gottschalk C, Moliterno J, Günel M, Gerrard JL, Dib-Hajj S, Waxman SG, Barker FG 2nd, Alper SL, Chahine M, Haider S, De Koninck Y, Lifton RP, Kahle KT. Exome Sequencing Implicates Impaired GABA Signaling and Neuronal Ion Transport in Trigeminal Neuralgia (2020). ***iScience***, 23(10):101552.

35. **Jin SC***, Dong W*, Kundishora AJ*, Panchagnula S*, Moreno-De-Luca A*, Furey CG, Allocco AA, Walker RL, Nelson-Williams C, Smith H, Dunbar A, Conine S, Lu Q, Zeng X, Sierant MC, Knight JR, Sullivan W, Duy PQ, DeSpenza T, Reeves BC, Karimy JK, Marlier A, Castaldi C, Tikhonova IR, Li B, Peña HP, Broach JR, Kabachelor EM, Ssenyonga P, Hehnly C, Ge L, Keren B, Timberlake AT, Goto J, Mangano FT, Johnston JM, Butler WE, Warf BC, Smith ER, Schiff SJ, Limbrick DD Jr, Heuer G, Jackson EM, Iskandar BJ, Mane S, Haider S, Guclu B, Bayri Y, Sahin Y, Duncan CC, Apuzzo MLJ, DiLuna ML, Hoffman EJ, Sestan N, Ment LR, Alper SL, Bilguvar K, Geschwind DH, Günel M, Lifton RP, Kahle KT. Exome sequencing implicates genetic disruption of prenatal neuro-gliogenesis in sporadic congenital hydrocephalus (2020). ***Nature Medicine***, 26(11):1754-1765.
36. Morton SU, Shimamura A, Newburger PE, Opotowsky AR, Quiat D, Pereira AC, **Jin SC**, Gurvitz M, Brueckner M, Chung WK, Shen Y, Bernstein D, Gelb BD, Giardini A, Goldmuntz E, Kim RW, Lifton RP, Porter GA Jr, Srivastava D, Tristani-Firouzi M, Newburger JW, Seidman JG, Seidman CE. Association of Damaging Variants in Genes With Increased Cancer Risk Among Patients With Congenital Heart Disease (2020). ***JAMA Cardiology***, e204947.
37. Sullivan W, Reeves BC, Duy PQ, Nelson-Williams C, Dong W, **Jin SC**, Kahle KT. Exome Sequencing as a Potential Diagnostic Adjunct in Sporadic Congenital Hydrocephalus (2020). ***JAMA Pediatrics***, 1;175(3):310-313.
38. Diab N*, **King S***, Dong W*, Allington G, Sheth A, **Peters ST**, Kahle KT#, **Jin SC**#. Analysis workflow to assess *de novo* genetic variants from whole-exome sequencing (2021). ***STAR Protocols***, 10;2(1):100383.
39. Dzinovic I, Skorvanek M, Pavelekova P, Zhao C, Keren B, Whalen S, Bakhtiari S, **Jin SC**, Kruer MC, Jech R, Winkelmann J, Zech M. Variant recurrence confirms the existence of a FBXO31-related spastic-dystonic cerebral palsy syndrome (2021). ***Annals of Clinical and Translational Neurology***, 8(4):951-955.
40. Alsharhan H, He M, Edmondson AC, Chen J, Donald T, Bakhtiari S, Amor D, Jones EA, Vassallo G, Vincent G, Cogné B, Deb W, Werners AH, **Jin SC**, Bilguvar K, Christodoulou J, Webster RI, Yearwood KR, Ng BG, Freeze HN, Kruer MC, Li D, Raymond KM, Bhoj EJ, Sobering AK. ALG13 X-linked intellectual disability: new variants, Glycosylation analysis, and expansion of the phenotype (2021). ***Journal of Inherited Metabolic Disease***, doi: 10.1002/jimd.12378.
41. Bakhtiari S, Tafakhori A, **Jin SC**, Guida BS, Alehabib E, Firouzbadi S, Bilguvar K, Darvish H, Kruer MC. Recessive *COL4A2* mutation leads to intellectual disability, epilepsy, and spastic cerebral palsy (2021). ***Neurology: Genetics***, 22;7(3):e583.
42. Wiessner M, Maroofian R, Ni MY, Pedroni A, Müller JS, Stucka R, Beetz C, Efthymiou S, Santorelli FM, Alfares AA, Zhu C, Uhrova Meszarosova A, Alehabib E, Bakhtiari S, Janecke AR, Otero MG, Chen JYH, Peterson JT, Strom TM, De Jonghe P, Deconinck T, De Ridder W, De Winter J, Pasquariello R, Ricca I, Alfadhel M, van de Warrenburg BP,

Portier R, Bergmann C, Ghasemi Firouzabadi S, **Jin SC**, Bilguvar K, Hamed S, Abdelhameed M, Haridy NA, Maqbool S, Rahman F, Anwar N, Carmichael J, Pagnamenta A, Wood NW, Tran Mau-Them F, Haack T; Genomics England Research Consortium, PREPARE network, Di Rocco M, Ceccherini I, Iacomino M, Zara F, Salpietro V, Scala M, Rusmini M, Xu Y, Wang Y, Suzuki Y, Koh K, Nan H, Ishiura H, Tsuji S, Lambert L, Schmitt E, Lacaze E, Küpper H, Dredge D, Skraban C, Goldstein A, Willis MJH, Grand K, Graham JM, Lewis RA, Millan F, Duman Ö, Dündar N, Uyanik G, Schöls L, Nürnberg P, Nürnberg G, Catala Bordes A, Seeman P, Kuchar M, Darvish H, Rebelo A, Bouçanova F, Medard JJ, Chrast R, Auer-Grumbach M, Alkuraya FS, Shamseldin H, Al Tala S, Rezazadeh Varaghchi J, Najafi M, Deschner S, Gläser D, Hüttel W, Kruer MC, Kamsteeg EJ, Takiyama Y, Züchner S, Baets J, Synofzik M, Schüle R, Horvath R, Houlden H, Bartesaghi L, Lee HJ, Ampatzis K, Pierson TM, Senderek J. Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia (2021). ***Brain***, 144(5):1422-1434.

43. Omer S, **Jin SC**, Koumangoye R, Robert SM, Duran D, Nelson-Williams C, Huttner A, DiLuna M, Kahle KT, Delpire E. Protein kinase D1 variant associated with human epilepsy and peripheral nerve hypermyelination (2021). ***Clinical Genetics***, doi: 10.1111/cge.13973.
44. Kundishora AJ*, **Peters ST***, Pinard A, Duran D, Panchagnula S, Barak T, Miyagishima DF, Dong W, Smith H, Ocken J, Dunbar A, Nelson-Williams C, Haider S, Walker RL, Li B, Zhao H, Thumkeo D, Marlier A, Duy PQ, Diab NS, Reeves BC, Robert SM, Sujijantarat N, Stratman AN, Chen Y-H, **Zhao S**, Roszko I, Lu Q, Zhang B, Mane S, Castaldi C, López-Giráldez F, Knight JR, Bamshad MJ, Nickerson DA, Geschwind DH, Lang Chen S-S, Storm PB, Diluna ML, Matouk C, Orbach DB, Alper SL, Smith ER, Lifton RP, Gunel M, Milewicz DM**, Jin SC**#, Kahle KT#. *DIAPH1* mutations in non-East Asian patients with sporadic moyamoya disease (2021). ***JAMA Neurology***, Jun 14:e211681. doi: 10.1001/jamaneurol.2021.1681.
45. Li M*, Zeng X*, Jin L*, **Jin SC**, Dong W, Brueckner M, Lifton RP, Lu Q, Zhao H. Integrative modeling of transmitted and *de novo* variants identifies novel risk genes for congenital heart disease (2021). ***Quantitative Biology***, 9(2): 216-227.
46. Lewis SA, Bakhtiari S, Heim J, Liu J, Huaang AJ, Musmacker A, **Jin SC**, Bilguvar K, Padilla-Lopez S, Kruer MC. Mutation in *ZDHHC15* leads to hypotonic cerebral palsy, autism, epilepsy, and intellectual disability (2021). ***Neurology: Genetics***, Jul 29;7(4):e602. doi: 10.1212/NXG.0000000000000602.
47. Calame DG*, Bakhtiari S*, Logan R, Coban-Akdemir Z, Du H, Mitani T, Fatih JM, Hunter JV, Herman I, Pehlivan D, Jhangiani SN, Person R, Schnur RE, **Jin SC**, Bilguvar K, Posey JE, Koh S, Firouzabadi SG, Alehabib E, Tafakhori A, Esmkhani S, Gibbs RA, Noureldeen MM, Zaki MS, Marafi D, Darvish H#, Kruer MC#, Lupski JR#. Biallelic loss-of-function variants in the splicing regulator *NSRP1* cause a severe neurodevelopmental disorder with spastic cerebral palsy and epilepsy (2021). ***Genetics in Medicine***. doi: 10.1038/s41436-021-01291-x.

48. Richard EM*, Bakhtiari S*, Marsh APL*, Kaiyrzhanov R*, Wagner M*, Shetty S, Pagnozzi A, Nordlie SM, Guida BS, Cornejo P, Magee H, Liu J, Norton BY, Webster RI, Worgan L, Hakonarson H, Li J, Guo Y, Jain M, Blesson A, Rodan LH, Abbott M, Comi A, Cohen JS, Alhaddad B, Meitinger T, Lenz D, Ziegler A, Kotzaeridou U, Brunet T, Chassevent A, Smith-Hicks C, Smith-Hicks J, Weiden T, Hahn A, Zharkinbekova N, Turnpenny P, Tucci A, Yelton M, Horvath R, Gungor S, Hiz S, Oktay Y, Lochmuller H, Zollino M, Morleo M, Marangi G, Nigro V, Torella A, Pinelli M, Amenta S, TUDP Study Group, Husain RA, Grossmann B, Rapp M, Steen C, Marquardt I, Grimmel M, Grasshoff U, Korenke GC, Owczarek-Lipska M, Neidhardt J, Radio FC, Mancini C, Sepulveda DJC, McWalter K, Begtrup A, Crunk A, Guillen Sacoto MJ, Person R, Schnur RE, Mancardi MM, Kreuder F, Striano P, Zara F, Chung WK, Marks WA, Tartaglia M, Striano V, Christodoulou J, Kaslin J, Padilla-Lopez S, Bilguvar K, Munchau A, Ahmed ZM, Hufnagel RB, Fahey MC, Maroofian R, Houlden H, Sticht H, Mane SM, Rad A, Vona B, **Jin SC**, Haack TB, Makowski C, Hirsch Y, Riazuddin S#, Kruer MC#. Biallelic variants in *SPATA5L1* lead to intellectual disability, spastic-dystonic cerebral palsy, epilepsy and hearing loss (2021). ***American Journal of Human Genetics***. Oct 7;108(10):2006-2016.
49. Barak T*, Ristori E*, Ercan-Sencicek AG, Miyagishima DF, Nelson-Williams C, Dong W, **Jin SC**, Prendergast A, Henegariu O, Erson-Omay EZ, Harmancı AS, Guy M, Gültekin B, Kilic D, Rai DK, Goc N, Aguilera SM, Gülez B, Altinok S, Ozcan K, Yarman Y, Coskun S, Sempou E, Deniz E, Hintzen J, Cox A, Fomchenko E, Jung SW, Ozturk AK, Louvi A, Bilgüvar K, Connolly Jr. ES, Khokha MK, Kahle KT, Yasuno K, Lifton RP, Mishra-Gorur K#, Nicoli S#, Günel M#. *PPIL4* is essential for brain angiogenesis and mutated in intracranial aneurysm patients (2021). ***Nature Medicine.*** Dec;27(12):2165-2175.
50. Zech M, Kumar KR, Reining S, Reunert J, Tchan M, Riley LG, Drew AP, Adam RJ, Berutti R, Biskup S, Derive N, Bakhtiari S, **Jin SC**, Kruer MC, Bardakjian T, Gonzales-Aalegre P, Sarmiento IJK, Mencacci NE, Lubbe SJ, Kurian MA, Cclot F, Menereett A, de Sainte Agathe J-M, Fung VSC, Vidailhet M, Baumann M, Marquardt T, Winkelmann J, Boesch S. Biallelic AOPEP loss-of-function variants linked to progressive dystonia with prominent limb involvement (2022)***. Movement Disorders***. Jan;37(1):137-147.
51. Fazeli W, Bamborschke D, Moawia A, Bakhtiari S, Tafakhori A, Giersdorf M, Hahn A, Weik A, Kolzter K, Shafiee S, **Jin SC**, Körber F, Lee-Kirsch MA, Cirak S, Darvish H, Kruer MC, Koy A. The phenotypic spectrum of PCDH12 associated disorders - five new cases and review of the literature (2022). ***European Journal of Paediatric*** Neurology. Jan;36:7-13.
52. Zech M*, Kopajtich R&, Steinbrücker K*, Bris C, Gueguen N, Feichtinger RG, Achleitner MT, Duzkale N, Périvier M, Koch J, Engelhardt H, Freisinger P, Wagner M, Brunet T, Berutti R, Smirnov D, Navaratnarajah T, Rodenburg RJT, Pais LS, Austin-Tse C, O'Leary M, Boesch S, Jech R, Bakhtiari S, **Jin SC**, Wilbert F, Kruer MC,

Wortmann SB, Eckenweiler M, Mayr JA, Distelmaier F, Steinfeld R, Winkelmann J, Prokisch H. Variants in mitochondrial ATP synthase cause variable neurologic phenotypes (2022). ***Annals of Neurology***. Feb;91(2):225-237.

53. Tang CSM*, Mononen M*, Lam W-Y, **Jin SC**, Zhuang X, Garcia-Barcelo M-M, Lin Q, Yang Y, Sahara M, Eroglu E, Chien K#, Hong H#, Tma PKH#, Gruber P#. Sequencing of a Chinese Tetralogy of Fallot cohort reveals clustering mutations in myogenic heart progenitors (2022). ***JCI Insight***. Jan 25;7(2):e152198.
54. Duy PQ*, Weise SC*, Marini C, Li X, Liang D, Dahl P, Ma S, Spajic A, Dong W, Juusola J, Kiziltug E, Kundishora AJ, Koundal S, Pedram MZ, Torres-Fernández LZ, Händler K, Domenico ED, Becker M, Ulas T, Juranek SA, Cuevas E, Hao LT, Jux B, Sousa AM, Kim S-K, Li M, Yang Y, Takeo Y, Duque A, Nelson-Williams C, Ha Y, Selvaganesan K, Robert SM, Singh AK, Allington G, Furey CG, Timberlake AT, Reeves BC, Smith H, Dunbar A, DeSpenza Jr. T, Goto J, Marlier A, Moreno-De-Luc A, Yu X, Butler WE, Carter BS, Lake EM, Constable RT, Rakic P, Lin H, Deniz E, Benveniste H, Malvankar N, Estrada-Veras JI, Walsh CA, Alper SL, Schultze J, Paeschke K, Doetzlhofer A, Wulczyn FG, **Jin SC**, Lifton RP, Sestan N, Kolanus W, Kahle KT. Impaired neurogenesis alters brain biomechanics in a neuroprogenitor-based genetic subtype of congenital hydrocephalus (2022). ***Nature Neuroscience.*** Apr;25(4):458-473.
55. Dong W*, Kaymakcalan H*, **Jin SC***, Diab NS*, Tanıdır C, Yalcin ASY, Ercan-Sencicek AG, Mane S, Gunel M, Lifton RP, Bilguvar K, Brueckner M. Mutation spectrum of congenital heart disease in a consanguineous Turkish population (2022). ***Molecular Genetics & Genomic Medicine***. June; 10(6):e1944.
56. Calame DG*, Herman I*, Marshall AE, Maroofian R, Donis KC, Fatih JM, Mitani T, Du H, Grochowski CM, Sousa S, Bakhtiari S, Io YA, Rocca C, Hunter JV, Sutton VR, Emrick LT, Boycott KM, Lossos A, Fellig Y, Prus E, Kalish Y, Meiner V, Suerink M, Ruivenkamp T, Muirhead K, Saadi NW, Zaki MS, Bouman A, Barakat TS, Skidmore DL, Osmond M, Silva TO, Houlden H, Murphy D, Karimiani EG, Jamshidi Y, Jaddoa AG, Tajsharghi H, **Jin SC**, Abbaszadegan MR, Ebrahimzadeh-Vesal R, Hosseini S, Alavi S, Bahreini A, Zarean E, Salehi MM, Robson S, Coban-Akdemir Z, Travaglini L, Nicita F, Jhangiani SN, Gibbs RA, Posey JE, Kruer MC, Kernohan KD, Saute JAM, Vanderver A, Pehlivan D, Marafi D, Lupski JR. Biallelic variants in the ectonucleotidase ENTPD1 cause a complex neurodevelopmental disorder with intellectual disability, distinct white matter abnormalities, and spastic paraplegia (2022). ***Annals of Neurology.*** Apr 26. Aug;92(2):304-321.
57. Dong W*, Wong KHY*, Liu Y*, Levy-Sakin M*, Hung W-C*, Li M, Li B, **Jin SC**, Choi J, Lopez-Giraldez F, Vaka D, Poon A, Chu C, Lao R, Balamir M, Movsesyan I, Malloy MJ, Zhao H, Kwok P-Y, Kane JP, Lifton RP, Pullinger CR. Whole exome sequencing reveals damaging gene variants associated with hypoalphalipoproteinemia (2022). ***Journal of Lipid Research***. Apr 20:100209. doi: 10.1016/j.jlr.2022.100209.

58. Xie Y, Jiang W, Li H, **Jin SC**, Brueckner M, Zhao H. Network assisted analysis of *de novo* variants using protein-protein interaction information identified 46 candidate genes for congenital heart disease (2022). ***PLoS Genetics***. Jun 7;18(6):e1010252. doi: 10.1371/journal.pgen.1010252
59. Guo H, Hou L, Shi Y, **Jin SC**, Zeng X, Li B, Lifton RP, Brueckner M, Zhao H, Lu Q. Quantifying concordant genetic effects of *de novo* mutations on multiple disorders (2022). ***eLife***. Jun 6;11:e75551. doi: 10.7554/eLife.75551.
60. Timberlake AT, Kiziltug E, **Jin SC**, Nelson-Williams C, Loring E, Yale Center for Genome Analysis, Allocco AA, Marlier A, Banka S, Stuart H, Passos-Buenos MR, Rosa R, Rogatto SR, Tonne E, Stiegler AL, Boggon TJ, Alperovich M, Steinbacher D, Flores RL, Persing JA, Kahle KT, Lifton RP. *De novo* mutations in the BMP signaling pathway in lambdoid craniosynostosis (2022). ***Human Genetics***. Aug 23. doi: 10.1007/s00439-022-02477-2.
61. Mishra-Gorur K, Barak T, Kaulen L, Henegariu O, **Jin SC**, Aguilera SM, Goles G, Yalbir E, Nishimura S, Miyagishima D, Djenoune L, Altinok S, Rai DK, Viviano S, Prendergast A, Zerillo C, Ozcan K, Baran B, Sencar L, Goc N, Yarman Y, Ercan-Sencicek AG, Bilguvar K, Lifton RP, Moliterno J, Louvi A, Yuan S, Deniz E, Brueckner B, Gunel M. *Pleiotropic role of TRAF7 in skull-base meningiomas and congenital heart disease* (2023). ***PNAS***. Apr 18;120(16):e2214997120.
62. Timberlake AT, McGee S, Allington G, Kiziltug E, Wolfe EM, Stiegler AL, Boggon TJ, Sanyoura M, Morrow M, Wenger TL, Fernandes EM, Caluseriu O, Persing JA, **Jin SC**, Lifton RP, Kahle KT, Kruszka P. De novo variants implicate chromatin modification, transcriptional regulation, and retinoic acid signaling in syndromic craniosynostosis (2023). ***American Journal of Human Genetics***. Apr 20:S0002-9297(23)00101
63. Kundishora AJ*, Allington G*, McGee S*, Mekbib KY*, Gainullin V, Timberlake AT, Nelson-Williams C, Kiziltug E, Smith H, Ocken J, Shohfi J, Allocco AA, Duy PQ, Elsamadicy A, Dong W, **Zhao S**, **Wang Y-C**, Qureshi H, Diluna ML, Mane S, Tikhonova IR, **Fu P-Y**, Castaldi C, López-Giráldez F, Knight JR, Furey CG, Carter BS, Haider S, Moreno-De-Luca A, Alper SL, Gunel M, Millan F, Lifton RP, Torene RI*, **Jin SC***, Kahle KT*. Multi-omic analyses implicate a neurodevelopmental program in the pathogenesis of cerebral arachnoid cysts (2023). ***Nature Medicine***. Mar;29(3):667-678.
64. Pinard A, Ye W, Fraser SM, Rosenfeld JA, Pichurin P, Hickey SE, Guo D, Cecchi AC, Guey S, Aloui C, Lee K, Kraemer M, Alyemni SO, University of Washington Center for Mendelian Genomics, Bamshad MJ, Nickerson DA, Tournier-Lasserve E, Haider S, **Jin SC**, Smith ER, Kahle KT, Jan LY, He M, Boerio ML, Milewicz DM. Rare variants in ANO1, encoding a Calcium-activated chloride channel, predispose to Moyamoya disease (2023). ***Brain***. May 30; awad172.
65. Ahmad N, Fazeli W, Schließke S, Lesca G, Gokce-Samar Z, Mekbib KY, **Jin SC**, Burton J, Hoganson G, Petersen A, Gracie S, Granger L, Bartels, E, Oppermann H, Kundishora

AJ, Till M, Milleret-Pignot C, Dangerfield S, Viskochil D, Anderson KJ, Palculict TB, Schnur RE, Wentzensen IM, Tiller GE, Kahle KT, Kunz WS, Burkart S, Simons M, Sticht H, Jamra RA, Neuser S. *De novo* variants in RAB11B cause various degrees of global developmental delay and intellectual disability in children (2023). ***Pediatric Neurology*****.** DOI:https://doi.org/10.1016/j.pediatrneurol.2023.08.023

66. Almousa H, Lewis SA, Bakhtiari S, Nordlie SH, Pagnozzi A, Magee H, Efthymiou S, Heim JA, Cornejo P, Zaki MS, Anwar N, Maqbool S, Rahman F, Neilson DE, Vemuri A, **Jin SC**, Yang XR, Heidari A, van Gassen K, Trimouille A, Thauvin-Robinet C, Liu J, Bruel AL, Tomoum H, Shata MO, Hashem MO, Toosi MB, Ghayoor Karimiani E, Yeşil G, Lingappa L, Baruah D, Ebrahimzadeh F, Van-Gils J, Faivre L, Zamani M, Galehdari H, Sadeghian S, Shariati G, Mohammad R, van der Smagt J, Qari A, Vincent JB, Innes AM, Dursun A, Özgül RK, Akar HT, Bilguvar K, Mignot C, Keren B, Raveli C, Burglen L, Afenjar A, Donker Kaat L, van Slegtenhorst M, Alkuraya F, Houlden H, Padilla-Lopez S, Maroofian R, Sacher M, Kruer MC. TRAPPC6B biallelic variants cause a neurodevelopmental disorder with TRAPP II and trafficking disruptions (2023). ***Brain***. DOI: 10.1093/brain/awad301

Book Chapters

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). ***Methods in Molecular Biology***. 1303:299-314.

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***. 25(4):265-286.
2. Lewis SA, Shetty S, Wilson B, Huang AJ, **Jin SC**, Smithers-Sheedy H, Fahey MC, Kruer MC. Insights from genetic studies of cerebral palsy (2021). ***Frontiers in Neurology***. 11:625428.
3. Kundishora AJ, Singh AK, Allington G, Dunbar AM, Duy PQ, **Ryou J**, Alper SL, **Jin SC**, Kahle KT. Genomics of human congenital hydrocephalus (2021). ***Child's Nervous System***. doi: 10.1007/s00381-021-05230-8.
4. Diab NS*, Barish S*, Dong W*, **Zhao S***, Allington G, **Yu X**, Kahle KT, Brueckner M#, **Jin SC**#. Molecular genetics and complex inheritance of congenital heart disease (2021). ***Genes***. 12(7):1020.
5. DeSpenza Jr T*, Carlson M*, Panchagnula S, Robert S, Duy PQ, Mermin-Bunnell N, Reeves BC, Kundishora AJ, Elsamadicy AA, Smith H, Ocken J, Alper SL, Jin SC, Hoffman EJ#, Kahle KT#. *PTEN* mutations in autism spectrum disorder and congenital hydrocephalus: developmental pleiotropy and therapeutic targets (2021). ***Trends in Neurosciences.*** 44(12):961-976.

6. Allington G, Duy PQ, **Ryou J**, Singh A, Kiziltug E, Robert SM, Kundishora AJ, **King S**, Haider S, Kahle KT, **Jin SC**. Genomic approaches to improve the clinical diagnosis and management of patients with congenital hydrocephalus (2021). ***Journal of Neurosurgery: Pediatrics***. 29:1-10.
7. **Wang Y-C***, Wu Y*, **Choi J***, Allington G*, **Zhao S***, **Khanfar M***, **Yang K***, **Fu P-Y**, **Wrubel M**, **Yu X**, Mekbib KY, Ocken J, Smith H, Shohfi J, Kahle KT, Lu Q#, **Jin SC**#. Computational genomics in the era of precision medicine: applications to variant analysis and gene therapy (2022). ***Journal of Personalized Medicine***. Jan 27;12(2):175.
8. Qureshi HQ*, Mekbib KY*, Allington G*, Elsamadicy AA, Duy PQ, Kundishora AJ, **Jin SC**, Kahle KT. Familial and syndromic forms of arachnoid cyst implicate genetic factors in disease pathogenesis (2022). ***Cerebral Cortex***. Jul 18:bhac257.
9. Mekbib KY, Muñoz W, Allington G, McGee S, Mehta NH, Shofi JP, Fortes C, Le HT, Nelson-Williams C, Nanda P, Dennis E, Kundishora AJ, Khanna A, Smith H, Ocken J, Greenberg ABW, Wu R, Moreno-De-Luca A, DeSpenza T Jr, Zhao S, Marlier A**, Jin SC**, Alper SL, Butler WE, Kahle KT. Human genetics and molecular genomics of Chiari malformation type 1 (2023). ***Trends in Molecular Medicine***. Oct 4:S1471-4914(23)00215-0.

- **Spotlight**

1. Duy PQ, Rakic P, Alper SL, Butler WE, Walsh CA, Sestan N, Geschwind DH, **Jin SC**, Kahle KT. Brain ventricles as windows into brain development and disease (2022). ***Neuron***. 110(1):12-15.