Curriculum Vitae
Sheng Chih Jin, Ph.D.

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Personal Information

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Present Position

2020 – Present	Assistant Professor of Genetics, Washington University School of School of Medicine, St. Louis, MO

Education

2000 – 2004	B.S. (Applied Mathematics), National Chiao Tung University, Hsinchu, Taiwan
2006 – 2008	ScM (Biostatistics), Johns Hopkins Bloomberg School of Public Health, Baltimore, MD, USA
2010 – 2014	Ph.D. (Human & Statistical Genetics), Washington University School of Medicine, St. Louis, MO, USA Advisors: Alison Goate and Carlos Cruchaga

Academic Positions/Employment

2014 – 2018	**Postdoctoral Fellow**, Yale School of Medicine, New Haven, CT, USA Advisors: Richard P. Lifton, Martina Brueckner, and Hongyu Zhao
2018 – 2020	**Postdoctoral Fellow**, Rockefeller University, New York, NY, USA Advisors: Richard P. Lifton, Martina Brueckner, and Hongyu Zhao

2020 – Present	**Assistant Professor,** Department of Genetics, Washington University School of Medicine, St. Louis, MO

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
2022	**Pediatric Cardiac Genomics Consortium and Cardiovascular Development Data Resource Center Challenge Prize**, Bench to Bassinet Program, NHLBI

Editorial/Reviewer Responsibilities

Ad Hoc Reviewer:
Journal of the American College of Cardiology (2024), Trends in Genetics (2023), Genome Research (2022, 2025), European Heart Journal (2018), Nature Communications (2024, 2025), eBioMedicine (2024, 2025), PLoS Genetics (2024), npj Genomic Medicine (2020), Brain (2022), Molecular Neurodegeneration (2014), Proteomics and Bioinformatics (2024), Human Genetics (2024), BMC Neurology (2013), Journal of Alzheimer's Disease (2014), Alzheimer's & Dementia (2016-2018), Genes (2020), Journal of Medical Genetics (2021), STAR Protocols (2022), Journal of Personalized Medicine (2022), Human Molecular Genetics (2025), Communications Medicine (2025)

Editorial Board Memberships:

2013 – Present **Review Editor**, Frontiers in Genetics, Neurogenomics Section

NIH Study Sections Ad hoc Reviewer:

2023, 2024 **Ad Hoc Reviewer**, NIH, Cardiovascular and Respiratory Diseases (CRD) Study Section
2023 **Ad Hoc Reviewer**, NIH ZMH1 ERB-S (02) S - Data Analysis and Coordination Center for the PsychENCODE Consortium (U24)
2024, 2025 **Ad Hoc Reviewer**, NIH, Genetics of Health and Diseases (GHD) Study Section

Institutional Grant Reviews Ad Hoc Reviewer:

2021, 2022 **Ad Hoc Reviewer**, Clinical and Translational Research Funding Program, Washington University Institute of Clinical and Translational Sciences
2022, 2024 **Ad Hoc Reviewer**, NGI Pilot Awards, Washington University NeuroGenomics and Informatics Center

Other Granting Agencies Ad Hoc Mail Reviewer:

2022, 2023, 2025 **Grant Reviewer**, Hydrocephalus Association Innovator Award
2023 **Grant Reviewer**, Sidra Medicine Precision Medicine Challenge Award (IRF 24)
2024, 2025 **Abstract Reviewer**, American Society of Human Genetics Meeting

University, School of Medicine and Hospital Appointments and Committees

2020 – 2024 **DBBS Admissions Committee B Member**, Washington University School of Medicine
2025 – Present **Cross-Program DBBS Admissions Committee Member**, Washington University School of Medicine
2025 – Present **Steering Committee Member**, Washington University in St. Louis Molecular Genetics & Genomics and Computational & Systems Biology Graduate Programs

National Panels, Committees, Boards

2023 **Planning Committee**, Hydrocephalus Association & Rudi Schulte Research Institute Research Workshop
2023 – 2026 **Elected Member**, American Society of Human Genetics Digital Learning Committee

2024	**Moderator**, Platform Session on Machine Learning and AI Applications in Human Genetics, Annual Meeting of the American Society of Human Genetics
2023 – 2025	**Co-Chair**, NIH PRECISION Human Pain Data Subcommittee

Community Service Contributions

Participation In departmental activities:

2023 – Present	**Co-organizer**, Hope Center Monday Noon Seminars

Professional Society Memberships:

2011 – Present	**Member** of the American Society of Human Genetics
2015 – Present	**Member** of the American Heart Association

Major Invited Professorships and Lectureships

External Seminars:

2016	Unraveling the Genetic Basis of Congenital Heart Disease. Institute of Biomedical Sciences Seminar Series, Academia Sinica, Taiwan
2017	Integrated Genomics Characterization of Complex Inheritance in Congenital Heart Disease. Institute of Biomedical Sciences Seminar Series, Academia Sinica, Taiwan
2018	Mutations in GTPase Signal Transduction Genes in Cerebral Palsy. 2nd International Cerebral Palsy Genomics Consortium Conference, Zhengzhou, China (Invited Keynote Presentation)
2018	Integrated Genomics Characterization of Complex Inheritance in Congenital Heart Disease. Institute of Medical Genomics and Proteomics Seminar Series, National Taiwan University College of Medical, Taiwan
2018	Genomics Approaches to Understand the Genetic Architecture of Congenital Heart Disease and Neurodevelopmental Disorders. Eugene McDermott Center for Human Growth and Development Department Seminar Series, University of Texas Southwestern Medical Center, Dallas, TX
2018	Genomics Approaches to Understand the Genetic Architecture of Congenital Heart Disease and Neurodevelopmental Disorders. Institute for Genomic Medicine Seminar Series, Nationwide Children's Hospital, Columbus, OH
2019	Genomics Approaches to Understand the Genetic Architecture of Congenital Heart Disease and Neurodevelopmental Disorders. Waisman Center Seminar Series, University of Wisconsin – Madison, Madison, WI

2019	Genomics Approaches to Understand the Genetic Architecture of Congenital Heart Disease and Neurodevelopmental Disorders. Mindich Child Health and Development Institute Seminar Series, Icahn School of Medicine at Mount Sinai, New York, NY
2019	A Major Role for Genes that Control Developmental Neuritogenesis in Cerebral Palsy. 3rd International Cerebral Palsy Genomics Consortium Conference**,** Anaheim, CA
2021	Genes and Mechanisms of Cerebral Palsy and Other Pediatric Movement Disorders. Boston Taiwanese Biotechnology Association Monthly Seminar Series, Boston Taiwanese Biotechnology Association (Virtual)
2022	Integrated analysis of genome sequencing, exome sequencing, and transcriptome profiling in congenital hydrocephalus. Hydrocephalus Association Network for Discovery Science Webinar Series, HA CONNECT (Virtual)
2023	Molecular Genetics and Complex Inheritance of Congenital Hydrocephalus. Hydrocephalus Association & Rudi Schulte Research Institute Research Workshop, Dallas, TX
2024	Revealing Molecular and Cellular Mechanisms of Congenital Anomalies Using Integrative Genomics. Institute of Medical Genomics and Proteomics Seminar Series, National Taiwan University College of Medicine, Taiwan
2024	Revealing Molecular and Cellular Mechanisms of Congenital Anomalies Using Integrative Genomics. Institute of Molecular Biology Seminar Series, Academia Sinica, Taiwan
2024	Revealing Molecular and Cellular Mechanisms of Congenital Anomalies Using Integrative Genomics. Academic forum, Kaohsiung Medical University
2025	Uniparental Disomy in Congenital Heart Disease. Kids First Long Reads Working Group Meeting (Virtual)

Internal Seminars at Washington University:

2017	Department of Genetics
2021	Pediatric Neurology Research Working Group
2021	Genetics and Genomic Medicine Case Conference
2021	Department of Developmental Biology
2021	Department of Computer Science & Engineering
2022	MSTP Future of Medicine Seminar
2022	Department of Genetics Retreat
2022	Center for Cardiovascular Research
2023	Intellectual and Developmental Disabilities Research Center Inaugural Symposium
2024	Center of Regenerative Medicine Faculty Retreat
2025	I2DB Seminar
2025	Hope Center Monday Noon Seminar

Consulting Relationships and Board Memberships

N/A

Research Support

PRESENT

R01NS131610 Jin (PI) 9/1/2024 – 5/31/2030 2.4CM
NINDS $3,157,725
"Molecular and cellular characterization of congenital hydrocephalus"
The major goal is to establish a diverse cohort of primary congenital hydrocephalus (CH) patients, with a particular emphasis on non-European populations, to ensure broader applicability and inclusivity of our findings. Through the integration of genomics, phenomics, and neuroimaging data, we will comprehensively characterize germline and somatic variants, explore temporal expression patterns of CH risk genes across various cell types, enhance diagnostic precision, and unravel genotype-phenotype correlations. Furthermore, the development of the HYDRO-Seq Genome Browser, integrated with the WashU Epigenome Browser, will streamline data analysis and enable seamless sharing of patient genetic and phenotypic information.
Role: PD/PI

Children's Discovery Institute Faculty Scholar Award 10/1/2021 – 9/30/2026 0.42CM
WashU Children's Discovery Institute Jin (PI) $300,000
"Human Genetics and Molecular Mechanisms of Cerebral Palsy"
The major goal of this project is to utilize an integrative, multi-dimensional omics approach coupled with functional genomics to discover novel cerebral palsy genetic risk factors and provide mechanistic insight into newly identified genetic causes.
Role: PD/PI

Project Grant PRG03121 Jin (PI) 6/1/2022 – 5/31/2027 0.12CM
Cerebral Palsy Alliance Research Foundation $225,000
"Discovery of novel genetic variations in cerebral palsy by whole genome sequencing"
The major goal is to identify novel genetic causes for therapeutic intervention and increase precision in genetic counseling, outcome prognostication, and treatment stratification, and inform future clinical trial design.
Role: PD/PI

R01NS111029 (NCE) Kahle/Jin/Deniz (MPI) 4/1/2020 – 1/31/2026 2.4CM
NINDS $158,028 (Jin)
"Human Genetics and Molecular Mechanisms of Congenital Hydrocephalus"
The goal of this project is to determine the genetic architecture and cellular and molecular mechanisms of human congenital hydrocephalus. The total costs are the amount awarded to WashU by the prime institution.

Role: MPI

U19NS130607 Milbrandt/DiAntonio/Jin (MPI) 9/30/2022–8/31/2027 0.6CM
NINDS $1,214,763 (Jin)
"INTERCEPT: Integrated Research Center for Human Pain Tissues (Project 1)"
In Project 1, we propose to identify additional genes involved in pain pathways via analysis of the genomes of a cohort of patients with idiopathic painful neuropathy. The proposed studies to enumerate, characterize, and spatially map these nerve cells, will provide a foundational resource for the pain community.
Role: Project Lead

U19NS130607 Jin/Zhao (MPI) 9/30/2022–8/31/2027 0.6CM
NINDS $347,161 (Jin)
"INTERCEPT: Integrated Research Center for Human Pain Tissues (Data Core)"
The primary purpose of the Data core is to collect, store, coordinate, and transfer all data, metadata, and analysis strategies generated within the U19 center (the INTERCEPT pain center) to the U24 DCIC and the HEAL Platform. The Data core will conduct multiple inter-disciplinary genomic and statistical analyses to study and integrate the multi-level omics data obtained from the three individual component projects of the INTERCEPT Center. The main objective of the Data core is to serve as a backbone support resource for experimental design refinement, data quality control, management, integration, analyses, and interpretation, and
Role: Project Lead

U24NS132103 Wang/Fulton/Lawson (MPI) 4/15/23 – 3/31/2028 2.4CM
NINDS $345,360 (Jin)
"WashU Somatic Mosaicism across Human Tissues (SMaHT) Program Organizational Center"
The overall goals of the WashU SMaHT-OC are to manage network coordination, lead consortium communications and outreach, and to develop and manage the network's websites, develop and manage engagement, training and collaborative programs. I only receive salary support from this grant.
Role: Co-Investigator

UM1DA058219 Wang/Fulton/Shen (MPI) 5/1/2023 – 4/30/2028 2.4CM
NIDA $629,300 (Jin)
"WashU-VAI Somatic Mosaicism across Human Tissues (SMaHT) Program Genome Characterization Center"
A fundamental goal of our proposed GCC is to generate high throughput, high quality, and high consistency genomic DNA and RNA sequencing data, and to construct a comprehensive catalogue of the human somatic variation together with other members of the Network. I only receive salary support from this grant.
Role: Co-Investigator

R01NS127108 Kruer (PI) 2/1/2023–1/31/2028 1.2CM
NINDS $358,747 (Jin)

"Genomic analysis of the Multiplex, Autozygous Populations in Cerebral Palsy (MAP CP) cohort: a focused approach to a complex disease"
The goal of this project is to discover genes with pathogenic variants detected in consanguineous populations and characterize molecular and developmental pathways that lead to cerebral palsy in neuronal and in vivo models. The total costs are the amount awarded to WashU by the prime institution.
Role: Co-Investigator

RM1NS135283 Haroutounian/Creed/Rodebaugh/Sinha/Lu/Shepherd (MPI)
NINDS 9/18/2024-8/31/2027 0.36CM
$26,918 (Jin)
"Integrated Mechanisms, Phenotypes, and Translational Underpinnings of Chronic Pain after Surgery (IMPETUS)"
The goal is to gain integrated mechanistic insights into peripheral and central biological processes that contribute to Chronic postsurgical pain (CPSP), for developing targeted strategies for its mitigation. I only receive salary support from this grant.
Role: Co-Investigator

PAST

R00HL143036 Jin (PI) 4/1/2020 – 3/31/2023 3.0CM
NHLBI $730,167
"Integrative Genomic Analysis of Congenital Heart Disease"
This project seeks to understand the complex genetics, to evaluate the sex differences in their genetic risk for CHD, and to determine the additive effect of common variants and rare deleterious variants on CHD using novel human genetics, genomics, and statistical approaches.
Role: PD/PI

Zebrafish Models for Pediatric Research Services Cooperative 2/5/2021 – 6/30/2023
WashU Children's Discovery Institute Jin (PI) $10,000
"Functional characterization of the Diaph1 gene using the zebrafish knock model"
The major goal of this project is to provide new insight into MMD pathophysiology with potentially immediate implications for targeted therapy for MMD patients.
Role: PD/PI

Clinical and Translational Research Funding Program 3/1/2021 – 2/28/2022
Washington University ICTS Jin (PI) $50,000
"Long-Read Genome Sequencing and Integrative Genomic Analysis for Cerebral Palsy"
The major goal of this project is to utilize a multi-omics approach coupled with advanced statistical methods to discover novel CP risk genes
Role: PD/PI

Hydrocephalus Association Innovator Award 12/31/2021 – 3/31/2023
Hydrocephalus Association Jin (PI) $50,000
"A Genome-Wide Assessment of Noncoding Risk Variants in Congenital Hydrocephalus"

The major goal of this project is to leverage whole-genome sequencing with recent advances in transcriptomics, computational genetics, integrative genomics, and artificial intelligence-aided phenomics, in a convergent, systems biology approach that seeks to translate genomic discoveries into pathophysiological insights that improve congenital hydrocephalus risk stratification and patient outcomes.
Role: PD/PI

R01NS106298 Kruer (PI) 4/1/2019–12/31/2023 0.12CM
NINDS $20,292 (Jin)
"Genomic Insights into the Neurobiology of Cerebral Palsy"
The goal of this application is to extend our preliminary findings to encompass a much larger cohort, providing the power required to define fundamental aspects of the genetic basis of CP.
Role: Subaward Co-Investigator

R01NS117609 Kahle/Boggon (MPI) 7/1/2020 – 6/30/2025 0.6CM
NINDS $191,213 (Jin)
"Human Genetics and Molecular Mechanisms of Vein of Galen Aneurysmal Malformation"
The goal of this project is to use a multidisciplinary approach that combines cutting-edge, next-generation DNA sequencing and bioinformatics with biochemistry and structural biology to elucidate the genetic architecture and molecular mechanisms of Vein of Galen aneurysmal malformation. The total costs are the amount awarded to WashU by the prime institution
Role: Subaward Co-Investigator

R01AR067715 Dobbs/Gurnett (MPI) 8/1/2020–7/31/2025 0.24CM
NIAMS $52,870 (Jin)
"Genetic Risk Factors for Severe Scoliosis"
The goal of this project is to study scoliosis in diverse patient populations and to comprehensively assess variant pathogenicity using our newly developed functional genomics methods. I only received salary support from this grant.
Role: Co-Investigator

Trainees/Mentees/Sponsorship Record

Postdoctoral Fellows

- **Yung-Chun Wang, PhD** (2021 – 2024). Now Instructor in the Jin Lab

Graduate Students:

- **Shujuan Zhao**, DBBS Molecular Genetics and Genomics (2020 – 2025), co-mentored with Kris Kahle, Doctoral (Ph.D.) thesis successfully defended June 6th, 2025. Now looking for postdoc
- **Nahyun Kong**, DBBS Human & Statistical Genetics (2022 – Present)

- **Julie Choi**, DBBS Molecular Genetics and Genomics (2022 – 2025), co-mentored with Jeffrey Milbrandt, Doctoral (Ph.D.) thesis successfully defended June 27th, 2025. Now looking for industrial job
- **Zitian Tang**, DBBS Biomedical Informatics & Data Science (2023 – Present)
- **Jenna Ulibarri**, DBBS Molecular Genetics and Genomics (2023 – Present)
- **Wendy Dong**, MSTP specialized in Computational & Systems Biology (2023 – Present), co-mentored with Jeffrey Milbrandt
- **Emma Casey**, DBBS Molecular Genetics and Genomics (2024 – Present)
- **Purva Patel**, DBBS Computational & Systems Biology (2024 – Present)
- **Zefan (Vivien) Li**, DBBS Molecular Cell Biology (2024 – Present)

Master's Students:

- **Samuel Peters**, SLU Bioinformatics and Computational Biology (2020 – 2021). Now Research Specialist at WashU's McDonnell Genome Institute
- **Spencer King**, WashU Computer Science (2020 – 2021). Now Bioinformatician at Geneoscopy
- **Xiaobing Yu**, WashU Computer Science (2021 – 2022). Now PhD student in Imaging Science at WashU

Postbaccalaureate trainees:

- **Max Wrubel**, Opportunities in Genomic Research Scholar (2021 – 2022). Now Bioinformatician in Alison Goate's lab at Mount Sinai

Undergraduate Students:

- **Kareena Joshipura**, Opportunities in Genomic Research Scholar (2021). Now Software Engineer in Capgemini
- **Athziri Marcial-Rodriguez**, Opportunities in Genomic Research Scholar (2022). Now Kornfeld Post-Bac Scholar in Jeffrey Gordon's Lab at WashU
- **Cabria Shelton**, Opportunities in Genomic Research Scholar (2022). Now master's Student at Wake Forest U
- **Andrew Ruttenberg**, WashU Computer Science on the Pre-Med Track (2022 – 2025). Now PhD Student at U of Toronto
- **Tuğçe Iyiyol**, WashU Biology on the Pre-Med Track (2022 – 2024). Now applying to Med Schools
- **Brian Yu**, U Chicago Computer Science (2024 – Present).
- **Aria Ma**, Opportunities in Genomic Research Scholar (2024).
- **Owen Limbrick**, WashU Biology on the Pre-Med Track (2024 – 2025).

Rotation Students:

- **Jian Ryou,** DBBS Human & Statistical Genetics (2020)
- **Changfeng Chen,** DBBS Molecular Cell Biology (2021)
- **Prashant Kumar Kuntala,** DBBS Computational & Systems Biology (2021)
- **Kuangying Yang,** DBBS Human & Statistical Genetics (2021)
- **Mariam Khanfar,** DBBS Human & Statistical Genetics (2021)
- **Lei Lu,** WashU Computer Science (2022)
- **Yuxiao Yu,** WashU MSTP (2022)
- **Vincent Gillespie,** DBBS Molecular Genetics and Genomics (2022)
- **Ai Zhang,** DBBS Human & Statistical Genetics (2022)
- **Yu-Liang Yeh,** DBBS Biomedical Informatics & Data Science (2023)
- **Justin Chen,** DBBS Computational & Systems Biology (2023)
- **Qichen Fu,** DBBS Molecular Genetics and Genomics (2023)
- **Tingkuan Chu,** DBBS Molecular Genetics and Genomics (2024)
- **Yu Liu,** DBBS Molecular Cell Biology (2024)
- **Sam Greenberg,** DBBS Molecular Genetics and Genomics (2024)
- **Wilber Palma,** DBBS Molecular Genetics and Genomics (2024)

Fellowships/Scholarships/Grants to Trainees/Mentees:

NIH TL1 Predoctoral Clinical Research Training Fellowship
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: 6/1/2021 – 5/31/2022
Amount: $2,110/month (Declined)

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 Wang
Role: Sponsor
Duration: 7/1/2021
Amount: $10,000 (signing bonus)

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: 7/2022
Amount: $1,800

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: 7/2022
Amount: $900

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: 8/2022 – 8/2024
Amount: $4,000 (one-time stipend supplement)

Maximizing Student Development (IMSD) Program
Agency: Washington University School of Medicine
Postdoc(s)/Student(s): Jenna Ulibarri
Role: Sponsor
Duration: 9/2022 – 9/2023
Amount: $27,144

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 – 9/2025
Amount: $81,720 total

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)

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: 7/2023
Amount: $500

Maximizing Student Development (IMSD) Program

Agency: Washington University School of Medicine
Postdoc(s)/Student(s): Emma Casey
Role: Sponsor
Duration: 9/2023 – 9/2024
Amount: $27,144

Washington University's T32 Genome Analysis Training Program
Agency: Washington University School of Medicine
Postdoc(s)/Student(s): Wendy Dong
Role: Co-sponsor
Duration: 9/2023 – 8/2026
Amount: $84,156 total

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 Cellular & Molecular Biology Training Program
Agency: Washington University School of Medicine
Postdoc(s)/Student(s): Jenna Ulibarri
Role: Sponsor
Duration: 11/2023 – 6/2025
Amount: $49,764 total

Annual Hope Center Retreat Poster Award
Agency: Arts & Sciences at Washington University in St. Louis
Postdoc(s)/Student(s): Julie Choi
Role: Co-sponsor
Duration: 4/2024
Amount: $1,000 (one-time allowance)

Washington University Summer Undergraduate Research Fellowship Program
Agency: Arts & Sciences at Washington University in St. Louis
Postdoc(s)/Student(s): Brian Yu
Role: Sponsor
Duration: 5/2024 – 8/2024
Amount: $2,500 (Declined)

Scholarships to attend Jackson Laboratory's Human and Mammalian Genetics and Genomics: The McKusick Short Course
Agency: Jackson Laboratory
Postdoc(s)/Student(s): Emma Casey

Role: Sponsor
Duration: 08/2024
Amount: $1,750

Scholarships to attend Jackson Laboratory's Human and Mammalian Genetics and Genomics: The McKusick Short Course
Agency: Jackson Laboratory
Postdoc(s)/Student(s): Purva Patel
Role: Sponsor
Duration: 8/2024
Amount: $500

Scholarships to attend Jackson Laboratory's Human and Mammalian Genetics and Genomics: The McKusick Short Course
Agency: Jackson Laboratory
Postdoc(s)/Student(s): Zefan (Vivien) Li
Role: Sponsor
Duration: 8/2024
Amount: $500

Predoctoral semifinalist for the 2024 Trainee Research Excellence Awards
Agency: American Society of Human Genetics
Postdoc(s)/Student(s): Nahyun Kong
Role: Sponsor
Duration: 8/2024
Amount: Complimentary registration to the 2024 ASHG Annual Meeting and $750

Human Cells, Tissues, and Organoids Core Microgrant
Agency: Center of Regenerative Medicine
Postdoc(s)/Student(s): Yung-Chun Wang
Role: Sponsor
Duration: 12/2024 – 12/2025
Amount: $2,500

Research Supplements to Promote Diversity in Health-Related Research
Agency: National Institute of Neurological Disorders and Stroke
Postdoc(s)/Student(s): Emma Casey
Role: Sponsor
Duration: 1/2025 – 12/2027
Amount: $225,981 (Rescinded due to President's Executive Order)

Full Scholarship to attend the Bruce Weir Summer Institute in Statistical Genetics
Agency: Bruce Weir Summer Institute in Statistical Genetics
Postdoc(s)/Student(s): Zefan (Vivien) Li
Role: Sponsor

Duration: 6/4/2025 - 6/13/2025
Amount: $3,640

Washington University's T32 Cellular & Molecular Biology Training Program
Agency: Washington University School of Medicine
Postdoc(s)/Student(s): Emma Casey
Role: Sponsor
Duration: 9/2025 – 8/2026
Amount: $33,838 total

Teaching Title and Responsibilities

Courses Taught:

2021, 2022	**Lecturer**, Bio5488: Genomics, Washington University School of Medicine (four lectures, totaling six hours)
2021, 2022	**Study Section Co-Leader**, Bio5491: Advanced Genetics, Washington University School of Medicine (one study section, totaling 1.5 hours)
2022	**Lecturer**, Bio5487: Genetics & Genomics of Disease, Washington University School of Medicine (one lecture, totaling 1.5 hours)
2022 – Present	**Co-director**, Bio5488: Genomics, Washington University School of Medicine (six lectures, totaling 9 hours)
2023	**Immersion Program Co-Leader**, Washington University School of Medicine (3 full days, totaling 24 hours)
2023	**Lecturer**, Bio5285: Current Topics in Human and Mammalian Genetics, Washington University School of Medicine (one lecture, totaling 1.5 hours)
2024, 2025	**Lecturer**, M65 Peds 511: Clinical Genetics & Genomics I, Washington University School of Medicine (one lecture, totaling 1.5 hours)
2025	**Lecturer**, Cancer Genomics, Washington University Continuing & Professional Studies Prison Education Project (two lectures, totaling 6 hours)

Thesis Examination Committee:

- **Ciyang Wang**, DBBS Molecular Genetics & Genomics Graduate Student in Laboratory of Dr. Carlos Cruchaga. (2021 – Present)
- **Chengran Yang**, DBBS Human & Statistical Genetics Graduate Student in Laboratory of Dr. Carlos Cruchaga. (2021)
- **Tong Wu**, WashU Biomedical Engineering Graduate Student in Laboratory of Dr. Jeffrey Milbrandt. (2021 – Present)
- **Caitlin Dingwall**, WashU MSTP Student in Laboratory of Dr. Jeffrey Milbrandt. (2023)

- **Kuangying Yang**, DBBS Human & Statistical Genetics Graduate Student in Laboratory of Dr. Angela Hirbe. (2023 – Present)
- **Gervette Penny**, DBBS Molecular Genetics & Genomics Graduate Student in Laboratory of Dr. Susan Dutcher. (2023)
- **Kangwen Xiao**, DBBS Molecular Genetics & Genomics Graduate Student in Laboratory of Dr. Angela Hirbe. (2025 – Present)

Qualifying Exam Committee:

- **Ji-Sun Kwon**, DBBS Computational & Systems Biology (2021)
- **Evelyn Craigen**, DBBS Molecular Genetics and Genomics (Chair; 2021)
- **Dan Western**, DBBS Human & Statistical Genetics (2023)
- **Kuangying Yang**, DBBS Human & Statistical Genetics (2023)
- **Grace Cooper**, DBBS Human & Statistical Genetics (Chair; 2023)
- **Juanru Guo**, DBBS Computational & Systems Biology (2023)
- **Mariam Khanfar**, DBBS Human & Statistical Genetics (Chair; 2023)
- **Chia-Jung Lee**, DBBS Computational & Systems Biology (2023)
- **Chien-Wei Peng**, DBBS Human & Statistical Genetics (2024)
- **Paul Lee**, WashU MSTP (Chair; 2024)
- **Arnold Federico**, DBBS Molecular Genetics & Genomics (2024)
- **Joey Nichols**, WashU MSTP (Chair; 2025)
- **Lloyd Tripp**, DBBS Molecular Genetics & Genomics (2025)
- **Qichen Fu**, DBBS Molecular Genetics & Genomics (2025)
- **Bart Olszowy**, DBBS Computational & Systems Biology (Chair; 2025)
- **Yuchen Cheng**, DBBS Molecular Genetics & Genomics (Chair; 2025)
- **Yu Liu**, DBBS Molecular Cell Biology (2025)
- **Wilber Palma**, DBBS Molecular Genetics & Genomics

Patents

N/A

Bibliography

- Corresponding author preprints and papers are indicated with a hash sign (**#**)
- Lab members are shown in **bold**
- Equal-contribution preprints and papers are indicated with an asterisk (*****)

A. Original, peer reviewed articles in refereed journals

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.

Genome-wide and candidate gene association study of cigarette smoking behavior. ***PLoS ONE***, 2009;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. A genome-wide association study of cleft lip with and without cleft palate identifies risk variants near MAFB and ABCA4. ***Nature Genetics***, 2010 Jun;42(6):525-9.
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. Evidence for gene-environment interaction in a genome wide study of isolated, nonsyndromic cleft palate. ***Genetic Epidemiology***, 2011 Sep;35(6):469-78.
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. 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.*** 2012 Aug 20;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. The FGF&FGFR gene family and risk of cleft lip with or without cleft palate. ***The Cleft Palate-Craniofacial Journal***, 2013 Jan;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. X-linked Markers in the Duchenne Muscular Dystrophy Gene Associated with Oral Clefts. ***European Journal of Oral Sciences***, 2013 Apr;121(2):63-8.
7. Benitez BA, Cooper B, Pastor P, **Jin SC**, Lorenzo E, Cervantes S, Cruchaga C. TERM2 is associated with the risk of Alzheimer's disease in Spanish population. ***Neurobiology of Aging***, 2013 Jun;34(6):1711.e15-7.
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. The PSEN1, p.E318G variant increases the risk of Alzheimer's disease in APOE-ε4 carriers. ***PLoS Genetics***, 2013;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. GWAS of cerebrospinal fluid tau levels identifies risk variants for Alzheimer's disease. ***Neuron***, 2013 Apr 24;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. Missense variants in *TREML2* protects against Alzheimer's disease. ***Neurobiology of Aging***, 2014 Jun;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. Coding variants in *TREM2* increase risk for Alzheimer's disease. ***Human Molecular Genetics***, 2014 Nov 1;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. Rare coding variants in the phospholipase D3 gene confer risk for Alzheimer's disease. ***Nature***, 2014 Jan 23;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. *TERM2* is associated with increased risk for Alzheimer's disease in African Americans. ***Molecular Neurodegeneration***, 2015 Apr;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. *De novo* mutations in congenital heart disease with neurodevelopmental and other congenital anomalies. ***Science***, 2015 Dec 4;350(6265):1262-1266.
15. Benitez BA, Davis AA, **Jin SC**, Ibanez L, Ortega-Cubero S, Pastor P, Choi J, Cooper B, Perlmutter JS, Cruchaga C. Resequencing analysis of five Mendelian genes and the top genes from genome-wide association studies in Parkinson's Disease. ***Molecular Neurodegeneration***, 2016 Apr 19; 11:29.

16. 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. Digenic mutations of human OCRL paralogs in Dent's disease type 2 associated with Chiari malformation. ***Human Genome Variation***, 2016 Dec 8;3:16042.
17. Song W, Hooli B, Mullin K, **Jin SC**, Cella M, Ulland TK, Wang Y, Tanzi RE, Colonna M. Alzheimer's disease-associated TREM2 variants exhibit either decreased or increased ligand-dependent activation. ***Alzheimer's & Dementia***, 2017 Apr;13(4):381-387.
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. Contribution of rare transmitted and *de novo* variants among 2,871 congenital heart disease probands. ***Nature Genetics***, 2017 Nov;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. A common haplotype lowers PU.1 expression in myeloid cells and delays onset of Alzheimer's disease. ***Nature Neuroscience***, 2017 Aug;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. CLCN2 chloride channel mutations in familial hyperaldosteronism type II. ***Nature Genetics***, 2018 Mar;50(3):349-354.
21. Antwi P, Hong CS, Duran D, **Jin SC**, Dong W, DiLuna M, Kahle, K. A novel association of campomelic dysplasia with hydrocephalus due to an unbalanced chromosomal translocation upstream of SOX9. ***Cold Spring Harbor Molecular Case Studies***, 2018 Jun 1;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. *De Novo MYH9* mutation in congenital scalp hemangioma. ***Cold Spring Harbor Molecular Case Studies***, 2018 Aug 1;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. *De novo* mutation in genes regulating neural stem cell fate in human congenital hydrocephalus. ***Neuron***, 2018 Jul 25;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. Human genetics and molecular mechanisms of congenital hydrocephalus. ***World Neurosurgery***, 2018 Nov;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. ***American Journal of Human Genetics***, 2018 Nov 1;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. ***Neuron***, 2019 Feb 6;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. ***PLoS Pathogens***, 2019 Apr 5;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. ***Molecular Genetics & Genomic Medicine***, 2019 Sep;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*. ***Frontiers Cellular Neuroscience***, 2019 Sep 26;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. ***PNAS***, 2019 Jul 9;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. ***PNAS***, 2019 Jul 23;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. ***Nature Genetics***, 2020 Oct;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. ***Genetics in Medicine***, 2020 Jun;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. ***iScience***, 2020 Sep 11;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. ***Nature Medicine***, 2020 Nov;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. ***JAMA Cardiology***, 2021 Apr 1;6(4):457-462.
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. ***JAMA Pediatrics***, 2021 Mar 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. ***STAR Protocols***, 2021 Mar 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. ***Annals of Clinical and Translational Neurology***, 2021 Apr;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. ***Journal of Inherited Metabolic Disease***, 2021 Apr 22;7(3):e583.
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. ***Neurology: Genetics***, 2021 Apr 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. ***Brain***, 2021 Jun 22;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. ***Clinical Genetics***, 2021 Aug;100(2):176-186.

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. ***JAMA Neurology***, 2021 Aug 1;78(8):993-1003.
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. ***Quantitative Biology***, 2021 Jun;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. ***Neurology: Genetics***, 2021 Jul 29;7(4):e602.
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. ***Genetics in Medicine***, 2021 Dec;23(12):2455-2460.
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. ***American Journal of Human Genetics***, 2021 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. ***Nature Medicine***, 2021 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***. Movement Disorders***. 2022 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. ***European Journal of Paediatric Neurology***, 2022 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. ***Annals of Neurology***, 2022 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. ***JCI Insight***, 2022 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. ***Nature Neuroscience***, 2022 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. ***Molecular Genetics & Genomic Medicine***, 2022 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. ***Annals of Neurology,*** 2022 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. ***Journal of Lipid Research***, 2022 Jun;63(6):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. ***PLoS Genetics***, 2022 Jun 7;18(6):e1010252.
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. ***eLife***, 2022 Jun 6;11:e75551.
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. ***Human Genetics***, 2023 Jan;142(1):21-32.
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. ***PNAS***, 2023 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. ***American Journal of Human Genetics***, 2023 May 4;110(5):846-862.
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. ***Nature Medicine***, 2023 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. ***Brain***, 2023 Sep 1;146(9):3616-3623.
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. ***Pediatric Neurology*****,** 2023 Nov;148:164-171.
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. ***Brain***, 2023 Sep 15:awad301.

67. Greenberg ABW, Mehta N, Allington G, **Jin SC**, Moreno-De-Luca A, Kahle KT. Molecular diagnostic yield of exome sequencing in patients with congenital hydrocephalus: a systematic review and meta-analysis. ***JAMA Network Open***, 2023 Nov 1;6(11):e2343384.
68. Singh AK*, Garrett Allington*, Viviano S, McGee S, Kiziltug E, Ma S, **Zhao S**, Mekbib KY, Shohfi JP, Duy PQ, DeSpenza Jr T, Furey CG, Reeves BC, Smith H, Sousa AM, Cherskov A, Allocco A, Nelson-Williams C, Haider S, Rizvi SRA, Alper SL, Sestan N, Shimelis H, Walsh LK, Lifton RP, Moreno-De-Luca A, **Jin SC**, Kruszka P, Deniz E#, Kahle KT#. A novel *SMARCC1* BAFopathy implicates neural progenitor epigenetic dysregulation in human hydrocephalus. ***Brain***, 2023 Dec 21:awad405.
69. **#Zhao S***, Mekbib KY*, van der Ent MA*, Allington G*, Prendergast A, Chau JE, Smith H, Shohfi J, Ocken J, Duran D, Furey CG, Hao LT, Duy PQ, Reeves BC, Zhang J, Nelson-Williams C, Chen D, Li B, Nottoli T, Bai S, Rolle M, Zeng X, Dong W, **Fu PY**, **Wang YC**, Mane S, Piwowarczyk P, Fehnel KP, See AP, Iskandar BJ, Aagaard-Kienitz B, Moyer QJ, Dennis E, Kiziltug E, Kundishora AJ, DeSpenza T Jr, Greenberg ABW, Kidanemariam SM, Hale AT, Johnston JM, Jackson EM, Storm PB, Lang SS, Butler WE, Carter BS, Chapman P, Stapleton CJ, Patel AB, Rodesch G, Smajda S, Berenstein A, Barak T, Erson-Omay EZ, Zhao H, Moreno-De-Luca A, Proctor MR, Smith ER, Orbach DB, Alper SL, Nicoli S, Boggon TJ, Lifton RP, Gunel M, King PD#, **Jin SC**#, Kahle KT#. Mutation of key signaling regulators of cerebrovascular development in vein of Galen malformations. ***Nature Communications***, 2023 Nov 17;14(1):7452.
70. Tafaleng EN, Li J, **Wang Y**, Hidvegi T, Soto-Gutierrez A, Locke AE, Nicholas TJ, Wang Y-C, Pak S, Cho MH, Silverman EK, Silverman GA, **Jin SC**, Fox IJ, Perlmutter DH. Variants in autophagy genes *MTMR12* and *FAM134A* are putative modifiers of the hepatic phenotype in α1-antitrypsin deficiency. ***Hepatology***, 2024 Apr 1. doi: 10.1097/HEP.0000000000000865.
71. #Duy PQ*, Jux B*, **Zhao S***, Mekbib KY, Dennis E, Dong W, Nelson-Williams C, Mehta NH, Shohfi JP, Juusola J, Allington G, Smith H, Marlin S, Belhous K, Monteleone B, Schaefer GB, Pisarska MD, Vásquez J, Estrada-Veras JI, Keren B, Mignot C, Flore LA, Palafoll IV, Alper SL, Lifton RP, Haider S, Moreno-De-Luca A, **Jin SC**#, Kolanus W#, Kahle KT#. *TRIM71* mutations cause a neurodevelopmental syndrome featuring ventriculomegaly and hydrocephalus. ***Brain***, 2024 Jun 4:awae175.
72. DeSpenza T Jr*, Singh A*, Allington G*, **Zhao S***, Lee J*, Kizlitug E*, Prina ML, Desmet N, Dang HQ, Fields J, Nelson-Williams C, Zhang J, Mekbib KY, Dennis E, Mehta NH, Duy PQ, Shimelis H, Walsh LK, Marlier A, Deniz E, Lake EMR, Constable RT, Hoffman EJ, Lifton RP, Gulledge A, Fiering S, Moreno-De-Luca A, Haider S, Alper SL, **Jin SC**, Kahle K#, Luikart BW#. Pathogenic variants in autism gene *KATNAL2* cause hydrocephalus and disrupt neuronal connectivity by impairing ciliary microtubule dynamics. ***PNAS***, 2024 Jul 2;121(27):e2314702121.

73. #Allington G*, Mehta NH*, Dennis E*, Mekbib KY*, Reeves B, Kiziltug E, Chen S, **Zhao S**, Duy PQ, Saleh M, Ang LC, Fan B, Nelson-Williams C, Moreno-de-Luca A, Haider S, Lifton RP, Alper SL, McGee S#, **Jin SC#**, Kahle KT#. *De novo* variants disrupt an LDB1-regulated transcriptional network in congenital ventriculomegaly. ***Brain***, 2024 Dec 16:awae395.
74. DeSpenza T Jr*, Kiziltug E*, Allington G*, Barson DG*, McGee S, O'Connor D, Robert SM, Mekbib KY, Nanda P, Greenberg ABW, Singh A, Duy PQ, Mandino F, **Zhao S**, Lynn A, Reeves BC, Marlier A, Getz SA, Nelson-Williams C, Shimelis H, Walsh LK, Zhang J, Wang W, Prina ML, OuYang A, Abdulkareem AF, Smith H, Shohfi J, Mehta NH, Dennis E, Reduron LR, Hong J, Butler W, Carter BS, Deniz E, Lake EMR, Constable RT, Sahin M, Srivastava S, Winden K, Hoffman EJ, Carlson M, Gunel M, Lifton RP, Alper SL, **Jin SC**, Crair MC, Moreno-De-Luca A, Luikart BW#, Kahle KT#. PTEN mutations impair CSF dynamics and cortical networks by dysregulating periventricular neural progenitors. ***Nature Neuroscience***, 2025 Feb 24. doi: 10.1038/s41593-024-01865-3.
75. Dong W, **Jin SC**, Sierant MC, Lu Z, Li B, Lu Q, Morton SU, Zhang J, López-Giráldez F, Nelson-Williams C, Knight J, Zhao H, Cao J, Mane S, Gruber PJ, Lek M, Goldmuntz E, Deanfield J, Giardini A, Mital S, Russell M, Gaynor JW, Cnota JF, Wagner M, Srivastava D, Bernstein D, Porter, Jr GA, Newburger J, Roberts AE, Yandell M, Yost HJ, Tristani-Firouzi M, Kim R, Seidman J, Chung WK, Gelb BD, Seidman CE, Lifton RP#, Brueckner M#. Recessive genetic contribution to congenital heart disease in 5,424 probands. ***PNAS***, 2025 Mar 11;122(10):e2419992122.
76. Sierant MC, **Jin SC**, Bilguvar K, Morton SU, Dong W, Jiang W, Lu Z, Li B, López-Giráldez F, Tikhonova I, Zeng X, Lu Q, Choi J, Zhang J, Nelson-Williams C, Knight JR, Zhao H, Cao J, Mane S, Sedore SC, Gruber PJ, Lek M, Goldmuntz E, Deanfield J, Giardini A, Mital S, Russell M, Gaynor JW, King E, Wagner M, Srivastava D, Shen Y, Bernstein D, Porter Jr GA, Newburger JW, Seidman JG, Roberts AE, Yandell M, Yost HJ, Tristani-Firouzi M, Kim R, Chun WK, Gelb BD, Seidman CE, Brueckner M, Lifton RP. Genomic analysis of 11,555 probands identifies 60 dominant congenital heart disease genes, ***PNAS***, 2025 Apr; 122(13):e2420343122.
77. **Choi J**, Strickland A, Loo HQ, Dong W, Barbar L, Bloom AJ, Sasaki Y, **Jin SC**, DiAntonio A, Milbrandt J. Diverse cell types establish a pathogenic immune environment in peripheral neuropathy, ***Journal of Neuroinflammation***, 2025 May 23;22(1):138.

B. Reviews, Chapters, and Editorials:

- Corresponding author work is indicated with a hash sign (**#**)

1. **Jin SC**, Benitez BA, Deming Y, Cruchaga C. Pooled-DNA sequencing for elucidation of genomic risk factors, rare variants underlying Alzheimer's disease. ***Methods in Molecular Biology***. 2016:1303:299-314.

2. Zeng X, Hunt A, **Jin SC**, Duran D, Gaillard J, Kahle KT. EphrinB2-EphB4-RASA1 signaling in human cerebrovascular development and disease. ***Trends in Molecular Medicine***. 2019 Apr;25(4):265-286.
3. Lewis SA, Shetty S, Wilson B, Huang AJ, **Jin SC**, Smithers-Sheedy H, Fahey MC, Kruer MC. Insights from genetic studies of cerebral palsy. ***Frontiers in Neurology***. 2021 Jan 21:11:625428.
4. Kundishora AJ, Singh AK, Allington G, Dunbar AM, Duy PQ, **Ryou J**, Alper SL, **Jin SC**, Kahle KT. Genomics of human congenital hydrocephalus. ***Child's Nervous System***. 2021 Nov;37(11):3325-3340.
5. **#**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. ***Genes***. 2021 Jun 30;12(7):1020.
6. 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. ***Trends in Neurosciences.*** 2021 Dec; 44(12):961-976.
7. **#**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. ***Journal of Neurosurgery: Pediatrics***. 2021 Oct 29:29(2):168-177.
8. **#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. ***Journal of Personalized Medicine***. 2022 Jan 27;12(2):175.
9. 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. ***Neuron***. 2022 Jan 5;110(1):12-15.
10. 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. ***Cerebral Cortex***. 2023 Mar 10;33(6):3012-3025.
11. 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. ***Trends in Molecular Medicine***. 2023 Dec;29(12):1059-1075.
12. **#**Lewis SA*, **Ruttenberg A*, Iyiyol T, Kong N, Jin SC**[#], Kruer MC[#]. Potential clinical applications of advanced genomic analysis in cerebral palsy. ***EBioMedicine***. 2024 Jul 5;106:105229.
13. **#Choi J*, Tang Z*, Dong W, Ulibarri J, Mehinovic E**, Thomas S, Höke A, **Jin SC**. Unleashing the power of multi-omics: unraveling the molecular landscape of peripheral neuropathy. ***Annals of Clinical and Translational Neurology***. 2025 March 24. doi:10.1002/acn3.70019.

C. Abstracts

- Corresponding author abstracts are indicated with a hash sign (#)

1. **Selected Oral Presentation:** Deep Resequencing of GWAS Loci Associated with Alzheimer's Disease. 2012 Alzheimer's Association International Conference, July 19, 2012, Vancouver, Canada
2. **Selected Oral Presentation:** Novel Coding Variants in *TREM2* increase Risk for Alzheimer's Disease. 2014 Alzheimer's Association International Conference, July 13, 2014, Copenhagen, Denmark
3. **Selected Platform Talk**: Inherited and De Novo Variant Analysis of 2871 WES probands. 2016 NHLBI Bench to Bassinet Research Program Annual Face-To-Face Conference, December 9, 2016, Rockville, MD
4. **Selected Platform Talk:** Expanded Whole Exome Sequencing Cohort Reveals Additional Novel CHD Genes. 2017 NHLBI Bench to Bassinet Research Program Annual Face-To-Face Conference, October 12, 2017, Rockville, MD
5. **Selected Platform Talk:** Trio-Based SNP Array Analysis in Congenital Heart Disease. 2019 NHLBI Bench to Bassinet Research Program Annual Face-To-Face Conference, October 10, 2019, Rockville, MD
6. **Selected Platform Talk:** Exome Sequencing Implicates Genetic Disruption of Prenatal Neuro-Gliogenesis in Sporadic Congenital Hydrocephalus. 2020 Annual Meeting of the American Society of Human Genetics, October 28, 2020, Virtual
7. **#Selected Platform Talk:** Discovery of Uniparental Disomy in 3,694 Congenital Heart Disease Trios. 2023 NHLBI Bench to Bassinet Research Program Annual Face-To-Face Conference, October 12, 2023, Rockville, MD
8. **#Poster:** Investigating Shared Genetic Causes of Heart and Brain Developmental Abnormalities. 2023 NHLBI Bench to Bassinet Research Program Annual Face-To-Face Conference, October 12, 2023, Rockville, MD
9. **#Featured Plenary Abstract Session:** Large-Scale Genomic Analysis and Targeted Functional Studies Uncover Disease-Associated Uniparental Disomy in Congenital Heart Disease. 2024 Annual Meeting of the American Society of Human Genetics, November 5, 2024, Denver, CO
10. **#Selected Platform Talk:** Contribution of Uniparental Disomy to Congenital Heart Disease in 3,869 Trios. 2024 International Joint Conference with the Korean Society for Medical Genetics and Genomics and the East Asian Union of Human Genetics Societies, December 5, 2024, Seoul, Korea
11. **#Poster:** Uncovering the molecular signatures of idiopathic peripheral neuropathy. 2025 Annual Hope Center Retreat, May 13, 2025, St. Louis, MO
12. **#Selected Lighting Talk**: A Comprehensive Benchmarking Resource for Somatic Variant Detection in HapMap Mixtures using Human Pangenome Graphs: 2025 SMaHT Annual Meeting, June 9, 2025, Bethesda, MD
13. **#Poster**: A Comprehensive Benchmarking Resource for Somatic Variant Detection in HapMap Mixtures using Human Pangenome Graphs. 2025 SMaHT Annual Meeting, June 9, 2025, Bethesda, MD
14. **#Poster:** Integrative Genomic Analysis Reveals Candidate Mitochondrial-Localized DNA variants in Idiopathic Peripheral Neuropathy. 2025UMDF's Mitochondrial Medicine Conference, June 18, 2025, St. Louis, MO

15. **#Poster:** Whole-Genome Identification of Short Tandem Repeats Underlying Idiopathic Peripheral Neuropathy Using Long-Read and Short-Read Sequencing: 2024 Annual Meeting of the American Society of Human Genetics, November 7, 2024, Denver, CO
16. **#Poster:** A Comprehensive Benchmarking Resource for Somatic Variant Detection in HapMap Mixtures using Human Pangenome Graphs. 2025 Annual Meeting of the American Society of Human Genetics, Boston, MA
17. **#Poster:** Repeat expansions in RFC1, BEAN1 and STARD7 contribute to idiopathic peripheral neuropathy. 2025 Annual Meeting of the American Society of Human Genetics, Boston, MA
18. **#Poster:** High-Throughput Proteomic Analysis Reveals Actin-Depolymerizing Factor and Complement Proteins as Biomarkers in Idiopathic Peripheral Neuropathy. 2025 Annual Meeting of the American Society of Human Genetics, Boston, MA

D. Preprints and Submitted Manuscripts

- Corresponding author preprints are indicated with a hash sign (#)
- Lab members are shown in **bold**
- Equal-contribution preprints and papers are indicated with an asterisk (*)

1. **#Tang Z***, Ovunc SS*, **Mehinovic E**, Thomas S, **Ulibarri J**, **Li Z**, Baldridge D, Cruchaga C, Johnson M, Milbrandt J, Callaghan B, PNRR Study Group, Höke A#, Todd PK#, **Jin SC#**. Heterozygous and homozygous *RFC1* AAGGG repeat expansions are common in idiopathic peripheral neuropathy. **medRxiv** (https://www.medrxiv.org/content/10.1101/2025.04.18.25325809v2) 2025
2. **#Kong N***, **Tang Z***, **Ruttenberg A***, Macias-Velasco JF, **Li Z**, Zhang W, Miao B, Xin Z, Fu Q, Park H, Zhou X, **Mehinovic E**, Belter Jr E, Tomlinson C, Garza JE, Dong S, **Casey E**, Johnson B, Majewski MF, Palmer T, Cheng Y, Lindsay T, Schedl T, Li D, Shen H, Fulton R, SMaHT Network Assembly/Pangenome Working Group, Wang T#, **Jin SC#**. A pangenomic method for establishing a somatic variant detection resource in HapMap mixtures. **bioRxiv**. https://www.biorxiv.org/content/10.1101/2025.09.29.679336v2.abstract) 2025
3. Tan J, Wu Y, Barve R, Li F, Payne P, **Kong N**, **Jin SC**, Head R, Sun Y. SnakeAltPromoter facilitates differential alternative promoter analysis. **bioRxiv** (https://www.biorxiv.org/content/10.1101/2025.08.16.669128v1.abstract) 2025
4. The Somatic Mosaicism across Human Tissues Network (SMaHT). Comprehensive benchmarking of somatic mutation detection by the SMaHT Network. **bioRxiv** (https://www.biorxiv.org/content/10.1101/2025.10.09.678885v1) 2025
5. #Bisarad P*, **Wang Y-C***, Skidmor PT, Galaz-Montoya CI, Lewis SA, Alhaddad B, **Kong N**, Julian D, Magee H, Kruer TN, Xie Y, Zheng W, Li B, Rajabpour FV, Liu J, Revanur A, Bakur K, Firouzabadi SG, Sharbatkhori S, Tafakhori A, Taghiabadi E, Nezaminargabad E, Vosoogh S, Jamshidi J, Arefnia S, Hosseini SA, Khajehmirzaei A, Jamali F, Ahmadifard A, Khodadadi H, Daneshmand P, Bohlega S, Maddirevula S, Nadeef SS, Hashem MO, Salih MA, Mohmed IN, Sticht H, Morias SP, Damásio J, Santos M, Loureiro JL, Rodrigues R, Stevanin G, Benkirane M, Dauriat B, Head N, Baptista J,

Shahhosseini S, Mohammad F, Zhao H, Padilla-Lopez S, Alkuraya F, Bakhtiari S, Kruer MC#, **Jin SC#**, Darvish H#. Recessive genomic and phenotypic variation in consanguineous families with cerebral palsy. **medRxiv** (https://www.medrxiv.org/content/10.1101/2025.11.04.25339178v1) 2025

6. Su H#, Huang Y, Durham T, **Kong N**, **Casey E**, Benjamin D, **Jin SC**, Garimella K#. Himito: A graph-based toolkit for mitochondrial genome analysis using long reads. **bioRxiv** (https://www.biorxiv.org/content/10.1101/2025.11.03.686348v1) 2025
7. Allington G*, Dennis E*, Li Q*, McGee S, Mehta NH, Mekbib KY, Hatada I, Weston MC, DeSpenza T, Singh A, Miyagishima D, Kiziltug E, Hale AT, Duy PQ, Fan B, Nelson-Williams C, Moreno-de-Luca A, Smith H, Davalan WC, Kundishora AJ, Liukart BW, Butler W, Carter BS, Haider S, Alper SL, Lifton RP, **Jin SC**, & Kahle KT. Genetically regulated co-development of the human cerebrospinal fluid-ventricular system and cerebral cortex. **Under revision**