

**BIOGRAPHICAL SKETCH**

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NAME: Jin, Sheng Chih

eRA COMMONS USER NAME: PETERJIN

POSITION TITLE: Assistant Professor of Genetics

**EDUCATION/TRAINING**

INSTITUTION AND LOCATION	DEGREE	Completion Date	FIELD OF STUDY
National Chiao Tung University, Taiwan	B.S.	06/2004	Applied Mathematics
Johns Hopkins University	ScM	05/2008	Biostatistics
Washington University in St. Louis	Ph.D.	08/2014	Human & Statistical Genetics
Yale University	Postdoctoral	06/2018	Human Genetics & Genomics
Rockefeller University	Postdoctoral	03/2020	Human Genetics & Genomics

**A. Personal Statement**

I am a human geneticist with an independent research lab at the Washington University School of Medicine in the Department of Genetics. My prior scientific training includes my Ph.D. work with advisors Drs. Alison Goate and Carlos Cruchaga, in which I performed deep sequencing in candidate Alzheimer's disease (AD) genes, analyzed AD-related endophenotypes, and performed *in vitro* cell-based experiments to identify and functionally characterize novel genetic variants affecting AD risk [a]. As a postdoctoral fellow in the lab of Richard Lifton, I developed novel statistical models and bioinformatics pipelines to reveal the significant contribution of rare transmitted and *de novo* mutations on congenital heart disease risk [b]. More recently, I shifted my focus to reveal genetic etiologies of neurodevelopmental disorders. Working with multi-site genomics consortia, we identified novel genes and biological pathways contributing to Vein of Galen malformation [c], congenital hydrocephalus [d], and idiopathic cerebral palsy [Jin et al. under re-revision in *Nature Genetics*]. My current research is devoted to understanding the complex genetic models driving neurodevelopmental and cardiovascular disorders and to characterizing genetic networks of disease via multi-dimensional omics data, biobank sources, and electronic health record data. I utilize a multidisciplinary approach that reflects my training in biostatistics, human genetics, and genomics. My overall goal is to translate advances in basic science into novel targeted therapeutics for congenital and neurodegenerative diseases.

- a. **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. PMCID: [PMC4189899](#) (Citations: >150)
- b. **Jin SC\***, J, Homsy J\*, Zaidi S\*, Lu Q, Morton S, DePalma S, Zeng X, Qi H, Chang W, Hung ., Sierant M, Haider S, Zhang J, Knight ., Bjornson R, Castaldi C, Tikhonova I, Bilguvar K, Mane S, Sanders S, Mital S, Russell M, Gaynor W, Deanfield J, Giardini A, Porter G, Srivastava D, Lo C, Shen Y, Watkins S, Yandell M, Yost J, Tristani-Firouzi M, Newburger J, Roberts A, Kim R, Zhao H, Kaltman J, Goldmuntz E, Chung W, Seidman J, Gelb B, Seidman C<sup>†</sup>, Lifton R<sup>†</sup>, Brueckner M<sup>†</sup>. Contribution of rare inherited and *de novo* variants in 2,871 congenital heart disease probands. **Nature Genetics**. PMCID: [PMC5675000](#). \*Co-first authors (Citations: >170)
- c. Duran D\*, Zeng X\*, **Jin SC\***, Choi J\*, Nelson-Williams C, Yatsula B, Gaillard J, Furey CG, Lu Q, Timberlake AT, Mansuri MS, Sorscher MA, Loring E, Klein J, Montejo JD, Vera A, Hu JK, Allocco A, Karimy JK, Panchagnula S, Youngblood MW, DiLuna ML, Matouk CC, Mane SM, Tikhonova IR, Castaldi C, López-Giráldez F, Knight J, Haider S, Alper SL, Komiyama M, Ducruet AF, Zabramski JM, Dardik A, 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*. 2018. PMID: [30578106](#). \*Co-first authors

- d. 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*. PMCID: [PMC29983323](#).

## B. Positions and Honors

### Positions and Employment

Sept 2004 – Feb. 2006	Corporal, R.O.C. Army, Taiwan
June 2007 – May 2008	Research Assistant, National Cancer Institute, NIH
June 2008 – May 2010	Senior Biostatistician, Johns Hopkins School of Medicine
June 2010 – Aug 2014	Graduate Student, Washington University in St. Louis
Sept 2014 – June 2018	Postdoctoral Fellow, Yale University School of Medicine
July 2018 – Mar. 2020	Postdoctoral Fellow, Rockefeller University
Apr. 2020 – Present	Assistant Professor, Department of Genetics, Washington University in St. Louis

### Other Experience and Professional Memberships

2008 – 2010	Member, Eastern North American Region/International Biometric Society
2011 – Present	Member, American Society of Human Genetics
2013 – Present	<i>Ad hoc</i> reviewer, <i>BMC Neurology</i> , <i>Journal of Alzheimer's Disease</i> , <i>Molecular Neurodegeneration</i> , <i>Alzheimer's &amp; Dementia</i> , <i>European Heart Journal</i>
2013 – Present	Review editor, <i>Frontiers in Genetics</i> , <i>Neurogenomics Section</i>
2015 – Present	Member, American Heart Association

### Honors

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

## C. Contributions to Science

### 1. Determination of the genetic architecture of congenital heart diseases

As the pathogenesis of most congenital heart disease patients remains unknown, the Pediatric Cardiac Genomics Consortium has applied novel genomic techniques to a recruited cohort of over ~11,000 CHD probands in order to uncover the molecular basis of CHD. Our recent study revealed that protein damaging *de novo* mutations account for ~20% patients with CHD and neurodevelopmental disorders (NDD). Furthermore, children with both CHD and NDD share certain genetic mutations that disrupt normal development in the heart and brain [a]. I further developed a novel and robust statistical framework for

comprehensive analysis of rare inherited variants [b]. I identified a founder mutation in *GDF1* (growth and differentiation factor) that accounts for 5% of severe CHD in Ashkenazim, a discovery that is immediately clinically useful. I identify very rare recessive genotypes in *MYH6* (myosin heavy chain 6) in ~11% of Shone complex probands, a particularly severe type of CHD involving multiple levels of obstruction to left ventricular outflow. I also revealed that dominant loss-of-function mutations in *FLT4* (aka vegf receptor3) accounts for 2.3% of patients with tetralogy of Fallot. In aggregate, at least 1.8% of CHD cases can be attributed to rare transmitted variants [b]. Using functional genomics and xenopus as a model system, we demonstrate how histone 2B monoubiquitination pathway modulation during development leads to abnormal cardiac left–right patterning [c].

- a. 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, 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. PMID: [PMC4890146](#).
- b. **Jin SC\***, J, Homsy J\*, Zaidi S\*, Lu Q, Morton S, DePalma S, Zeng X, Qi H, Chang W, Hung ., Sierant M, Haider S, Zhang J, Knight ., Bjornson R, Castaldi C, Tikhonova I, Bilguvar K, Mane S, Sanders S, Mital S, Russell M, Gaynor W, Deanfield J, Giardini A, Porter G, Srivastava D, Lo C, Shen Y, Watkins S, Yandell M, Yost J, Tristani-Firouzi M, Newburger J, Roberts A, Kim R, Zhao H, Kaltman J, Goldmuntz E, Chung W, Seidman J, Gelb B, Seidman C<sup>†</sup>, Lifton R<sup>†</sup>, Brueckner M<sup>†</sup>. Contribution of rare inherited and *de novo* variants in 2,871 congenital heart disease probands. **Nature Genetics**. 2017. PMID: [PMC5675000](#). **\*Co-first authors; <sup>†</sup>Co-corresponding authors**
- c. 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. PMID: [PMC6628794](#)

## 2. Disease gene discovery using genetics, genomics, and statistical approaches for neurological disorders

During my graduate studies at Washington University and postdoctoral training at Yale/Rockefeller, I have established collaborative partnerships with my colleagues on research projects. This strategy has been proven to be fruitful. The first study showed that mutations in chromatin modifier and Ephrin genes cause Vein of Galen malformation and, in aggregate, account for ~30% of patients [a]. The second study demonstrated that *de novo* mutations in genes regulating neural stem cell fate contribute to congenital hydrocephalus [b]. The third study identify two novel *SLC12A* mutations associated with C and implicate genetically encoded impairments in ion transport for the first time in CH pathogenesis [c]. The last study used expression quantitative trait loci data from brain, myeloid cells, and cerebrospinal fluid to fine-map some of the GWAS loci for AD in order to pinpoint strong candidate genes expressed in microglia [d]. These studies have improved our understanding of the genetic etiology underlying complex neurological disorders and providing new insight into mechanisms that can drive both rare and common disease pathogenesis.

- a. Duran D\*, Zeng X\*, **Jin SC\***, Choi J\*, Nelson-Williams C, Yatsula B, Gaillard J, Furey CG, Lu Q, Timberlake AT, Mansuri MS, Sorscher MA, Loring E, Klein J, Montejó JD, Vera A, Hu JK, Allocco A, Karimy JK, Panchagnula S, Youngblood MW, DiLuna ML, Matouk CC, Mane SM, Tikhonova IR, Castaldi C, López-Giráldez F, Knight J, Haider S, Alper SL, Komiyama M, Ducruet AF, Zabramski JM, Dardik A, 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**. 2018. PMID: [30578106](#). **\*Co-first authors.**
- b. Furey CG\*, Choi JC\*, **Jin SC**, Zeng X, Timberlake AT, Nelson-Williams C, Mansuri MS, Lu Q, Duran D, Panchagnula S, Allocco 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, López-Giráldez F, Knight J, Alper SL, Haoder S, Guclu B, Bayri Y, Sahin Y, Duncan CC, DiLuna ML, Günel M, Lifton RP, Kahle KT. *De novo* mutation in genes regulating neural stem cell fate in human congenital hydrocephalus. **Neuron**. 2018. PMID: [29983323](#).

- c. **Jin SC\***, Furey CG\*, Zeng X, Allocco A, Nelson-Williams C, Dong W, Karimy JK, Wang K, Ma S, Delpire E, Kahle KT. SLC12A ion transporter mutations in sporadic and familial human congenital hydrocephalus. **Molecular Genetics & Genomics Medicine**. 2019. PMCID: [PMC6732308](#). \*Co-first authors
- d. 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. PMCID: [PMC5759334](#).

### 3. Identification and functional characterization of novel genes associated with Alzheimer's

For my Ph.D. dissertation research, I performed deep sequencing of candidate AD genes and found: (1) rare variants in a novel gene *PLD3* significantly confer risk for late-onset AD [a]; (2) rare variants in *TREM2* increase risk for late-onset AD [b,c]. I then performed *in vitro* experiments to characterize the effects of *TREM2* variants on *TREM2* cell surface transport [d]. We demonstrated that *TREM2* variants identified in AD patients decreased or markedly increased binding to *TREM2* ligands [d]. We also showed that *TREM2* expression in human monocytes is minimum compared to monocyte-derived dendritic cells [d]. These works have uncovered novel genes/variants that affects AD risk and provided significant understanding of disease pathogenesis that could lead to identification of novel targets for therapeutic development.

- a. 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, Passmore P, Craig D, Johnston J, McGuinness B, Todd S, Heun R, Kölsch H, Kehoe PG, Hooper NM, Vardy ER, Mann DM, Pickering-Brown S, Brown K, Kalsheker N, Lowe J, Morgan K, David Smith A, Wilcock G, Warden D, Holmes C, 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. PMCID: [PMC4050701](#). \*Co-second authors
  - b. **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. PMCID: [PMC4189899](#).
  - c. **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. PMCID: [PMC4426167](#). \*Co-first authors
  - d. 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. PMCID: [PMC5299056](#).
- ### 4. Development and application of statistical methods for genome-wide association studies and clinical trials
- During a summer internship with Dr. Nilanjan Chatterjee at NCI, I developed novel statistical methods to combine diverse but correlated smoking phenotypes for single-marker analysis [a]. As a master's student and senior biostatistician at Hopkins, my research focused on statistical analyses of several randomized clinical trials and genetic epidemiology studies. I worked with Dr. Nae-Yuh Wang and Tom Louis to apply mix-effects and pattern-mixture models to robustly infer missing data in clinical trials. Meanwhile, I worked with Dr. Terri Beaty in the Oral Cleft Consortium GWAS analysis [b-d]. These results identified genetic susceptibility loci and evidence of gene-environment interaction for cleft lip and cleft palate.

- a. Caporaso N, Gu F, Chatterjee N, **Jin SC**, Yu K, Yeager M, Chen C, Jacobs K, Wheeler W, Landi MT, Zsieglar 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. PMCID: [PMC2644817](#)
- b. 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. PMCID: [PMC2941216](#).
- c. 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. PMCID: [PMC3180858](#).
- d. 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. PMCID: [PMC3600648](#).

**Full Bibliography:** <https://www.ncbi.nlm.nih.gov/pubmed/?term=Sheng+Chih+Jin+OR+Sheng+C.+Jin>

#### **D. Additional Information: Research Support and/or Scholastic Performance** **Ongoing Research Support**

NIH/NHLBI 4R00HL143036-02

Jin (PI) 4/1/2020-3/31/2023

*Integrative Genomic Analysis of Congenital Heart Disease*

This project seeks to understand the complex genetic, 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: PI

NIH/NINDS 1R01NS111029-01A1

Kahle (PI) 4/1/2020-1/31/2025

*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.

Role: Subaward Co-Investigator

#### **Completed Research Support**

NIH/NHLBI 1K99HL143036-01A1

Jin (PI) 4/1/2019-3/31/2021

*Integrative Genomic Analysis of Congenital Heart Disease*

The goal of this proposal is to investigate genetic modifiers and mutation spectrum of *FLT4* mutations in CHD, to perform the integrative genomic analysis of *de novo* and inherited variants for the identification of novel CHD risk genes, and to determine the additive effect of common polygenic and rare *de novo* variants on CHD using genomics and bioinformatics.

Role: PI

American Heart Association Postdoctoral Fellowship

Jin (PI) 7/1/2018 – 6/30/2020

*Integrated Genomic Characterization of Complex Inheritance in Congenital Heart Disease*

The major goal of this project is to determine pairwise genetic interaction in chromatin modifier genes on congenital heart disease risk in combination with induced pluripotent stem cell lines to functionally characterize the modifier genes that modulate the expressivity of chromatin modifier genes.

Role: PI

James Hudson Brown – Alexander Brown Coxe Postdoctoral Fellowship in the Medical Sciences

Jin (PI) 07/01/2015 – 06/30/2016

*Unraveling the Genetic Basis of Congenital Heart Disease*

The major goal of this project is to investigate the role of *de novo* single nucleotide variations and *de novo* copy number variations in the congenital heart disease by analyzing the whole exome sequencing data of the PCGC cohort

Role: PI

### **Pending Research Support**

NIH/NINDS 1R01NS117609-01

Kahle/Boggon (MPI) 7/1/2020-6/30/2025

*Human genetics and molecular mechanisms of Vein of Galen 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.

Role: Subaward Co-Investigator

NIH/NIAMS 2R01AR067715-01A1

Dobbs/Gurnett (MPI) 7/1/2020-6/30/2025

*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.

Role: Co-Investigator