Name: **A Jeremy Willsey, PhD**

Position: Assistant Professor In Residence
Psychiatry
School of Medicine

Address: 675 Nelson Rising Ln
Room 416, Bldg 19A (Sandler Neuroscience Center)
University of California, San Francisco
San Francisco, CA 94143
415-502-2503
Fax: 415-476-0526
jeremy.willsey@ucsf.edu
willseylab.ucsf.edu

EDUCATION

2004 - 2010	Simon Fraser University	BSc	Molecular Biology and Biochemistry
2010 - 2014	Yale University	PhD	Genetics
2014 - 2015	University of California, San Francisco	Postdoctoral Scholar	Genetics

PRINCIPAL POSITIONS HELD

2010 - 2014	Yale University	Graduate Student	Genetics
2013 - 2014	University of California, San Francisco	Junior Specialist	Psychiatry
2014 - 2015	University of California, San Francisco	Postdoctoral Scholar	Psychiatry
2015 - 2016	University of California, San Francisco	Assistant Professor	Psychiatry
2016 - present	University of California, San Francisco	Assistant Professor	IND & Psychiatry

HONORS AND AWARDS

2010	Dean's Medal for Undergraduate Studies in the Faculty of Science	Simon Fraser University
2012	Doctoral Foreign Study Award (predoctoral fellowship)	Canadian Institutes of Health Research
2013	Top Ten Advances in Autism Research	Autism Speaks
2013	SFARI Notable Paper	Simons Foundation
2014	The Carolyn Slayman Prize in Genetics (outstanding thesis award)	Yale University
2015	Postdoctoral Fellowship (ranked #1, declined)	Canadian Institutes of Health Research

KEYWORDS/AREAS OF INTEREST

autism spectrum disorders, ASD, tourette syndrome, schizophrenia, childhood-onset schizophrenia, psychiatric disorders, genetics, genomics, bioinformatics, systems biology, network analysis, gene expression, chromatin regulation, gene co-expression, gene regulation, whole-exome sequencing, whole-genome sequencing, high-throughput sequencing, RNA-seq, ChIP-seq, de novo variation, rare variation, SNVs, CNVs, neurodevelopment, brain development, spatiotemporal, induced pluripotent stem cells, iPSCs

MEMBERSHIPS

2009 - present Golden Key International Honour Society

2012 - present American Society of Human Genetics (ASHG)

2012 - present International Society for Autism Research (INSAR)

2013 - present Molecular Psychiatry Association

SERVICE TO PROFESSIONAL PUBLICATIONS

2013 - present Ad hoc reviewer for Biological Psychiatry (2 manuscripts in last 3 years)

2015 - present Ad hoc reviewer for Autism Research (1 manuscript in last 1 year)

2016 - present Ad hoc reviewer for Nature Medicine (2 manuscripts in last 1 year)

2016 - present Ad hoc reviewer for Molecular Autism (1 manuscript in last 1 year)

2016 - present Ad hoc reviewer for Genome Medicine (1 manuscript in last 1 year)

NATIONAL INVITED PRESENTATIONS

2016	Molecular Psychiatry Association	Invited Speaker

REGIONAL AND OTHER INVITED PRESENTATIONS

2013	GENE 760 - Genomic Methods for Genetic Analysis (graduate course), Yale University	Guest Lecturer
2014	UC Irvine Autism Update 2014: The Future of Autism Research, University of California, Irvine	Invited Speaker

CONTINUING EDUCATION AND PROFESSIONAL DEVELOPMENT ACTIVITIES

2014 The Data Scientist's Toolbox - Part of the online Data Science specialization offered by Johns Hopkins University

2014 R Programming - Part of the online Data Science specialization offered by Johns Hopkins University

2015 Getting and Cleaning Data - Part of the online Data Science specialization offered by Johns Hopkins University

2016 Master R Developer Workshop - Hosted by RStudio and led by Hadley Wickham.

UNIVERSITY SERVICE - CAMPUS

2016 - present	Planning Committee for New Neuroscience 23a Building	Committee Member

UNIVERSITY SERVICE - OTHERS

2012 - 2013	Department of Genetics Graduate Education Steering Committee	Yale University

COMMUNITY & PUBLIC SERVICE

2010 - 2012	Graduate and Professional Student Senate (GPSS), Yale University	Senator, Co-chair of Public Service committee
2010 - 2012	Graduate School Public Service Awards Committee, Yale University	Committee Member
2011 - 2012	McDougal Center, Yale University, New Haven	Public Service Fellow
2012 - 2013	McDougal Center, Yale University, New Haven	Technology and Communications Fellow

PREDOC STUDENTS SUPERVISED/MENTORED

Date	Name	Program or School	Mentor Type	Role	Current Position
2011 - 2012	Mack Su	Undergraduate Student, Yale University	Project Mentor	Research / Scholarly Mentor	MD-PhD Candidate, Harvard Medical School
2012 - 2013	Jake Gockley	Graduate Student, Yale University	Research/Scholarly Mentor,Project Mentor	Research/Scholarly Mentor	Graduate Student, Yale University
2014 - 2014	Margaret Cunniff	Rotating Graduate Student, University of California, San Francisco	Project Mentor	Research / Scholarly Mentor	Graduate Student, University of California, San Francisco
2014 - 2014	Manoj Kanagaraj	Summer Student, University of California, San Francisco	Research/Scholarly Mentor,Project Mentor	Research / Scholarly Mentor	Angier B. Duke Scholar at Duke University
2014 - 2015	Wipa Panmontha	Junior Specialist, University of California, San Francisco	Research/Scholarly Mentor,Project Mentor	Research / Scholarly Mentor	Graduate Student, Chulalongkorn University, Bangkok, Thailand
2015 - 2015	Nerissa Hoglen	Rotating Graduate Student, University of California, San Francisco	Project Mentor	Research / Scholarly Mentor	Graduate Student, University of California, San Francisco

Date	Name	Program or School	Mentor Type	Role	Current Position
2016 - 2017	Rebecca Krasnoff	SRA 1 (post-bac)	Research/Scholarly Mentor,Project Mentor	PI	Student, Post-baccalaureate Research Education Program, Johns Hopkins
2016 - present	Montana Morris	SRA 1 (post-bac)	Research/Scholarly Mentor,Project Mentor	PI	SRA 1, UCSF
2016 - present	Sheng Wang	PhD Student (exchange)	Research/Scholarly Mentor,Project Mentor,Career Mentor	PI	Phd Student, UCSF
2017 - present	Nia Teerikorpi	Rotating Graduate Student	Research/Scholarly Mentor,Project Mentor	PI	Graduate Student, UCSF

POSTDOC FELLOWS/RESIDENTS SUPERVISED/MENTORED

Date	Name	Fellow	Mentor Type	Faculty Role	Current Position
2014 - 2015	Rehab Khalil	Postdoctoral Scholar, UCSF	Project Mentor	3-4 times per year. Helping with data analysis and experimental design.	Postdoctoral Scholar, UCSF
2014 - 2016	Aaron Besterman	Psychiatry Resident, UCSF	Research/Scholarly Mentor,Project Mentor	Once per week. Providing general scientific mentorship and teaching. Also helping with data analysis and experimental design.	Child and Adolescent Psychiatry Fellow, UCLA
2014 - present	Vanessa Hus Bal	Postdoctoral Scholar, UCSF	Research/Scholarly Mentor,Project Mentor	Two times per month. Providing general scientific mentorship and teaching.	Assistant Professor, UCSF
2016 - present	Siavash Darbandi	Postdoctoral Scholar, UCSF	Project Mentor	Once every month. Helping with data analysis and experimental design.	Postdoctoral Scholar, UCSF
2016 - present	Eirene Markenscoff-Papadimitriou	Postdoctoral Scholar, UCSF	Project Mentor	Once every month. Helping with data analysis and experimental design.	Postdoctoral Scholar, UCSF
2016 - present	Irina Epstein	Postdoctoral Scholar, UCSF	Research/Scholarly Mentor,Project Mentor,Career Mentor	Co-advisor	Postdoctoral Scholar, UCSF
2017 - present	Nawei Sun	Postdoctoral Scholar, UCSF	Research/Scholarly Mentor,Project Mentor,Career Mentor	PI	Postdoctoral Scholar, UCSF

RESEARCH & CREATIVE ACTIVITIES SUMMARY

My research lies at the interface of genetics/genomics, systems biology, and neuroscience, and I focus on four areas: (**1**) **gene discovery** utilizing genome-wide approaches for identifying rare, large effect size variants, focusing on *de novo* mutations; (**2**) **systems**

biological approaches to translate genetic findings to biological insights and testable hypotheses; (**3**) testing these hypotheses in **model systems**; and (**4**) **generation of 'omics datasets** to aid areas 1-2 and/or as part of 3. Critically, these four areas work together in an iterative and synergistic manner. My early research focused on the genetics and biology of autism spectrum disorder (ASD) but I have recently expanded these approaches to Tourette Syndrome (TS).

Gene Discovery. Working closely with Matthew State and Stephan Sanders (UCSF), we have identified over 65 ASD associated genes by integrating large scale whole exome sequencing (WES) data with *de novo* copy number variant (CNV) data (Sanders, *Neuron* 2015). I am now leading a similar effort in TS. We recently published a manuscript implicating, for the first time, *de novo* variants in TS risk and identifying the first four clear-cut risk genes (Willsey *et al*., Neuron 2017). In collaboration with Drs. State and Sanders, we are also currently conducting a whole genome sequencing (WGS) analysis of 540 ASD quartets (unaffected parents, affected child, unaffected sibling). We have recently published this work in BiorXiv (http://biorxiv.org/content/early/2017/04/13/127043) and submitted it to the journal, *Nature*. I led a pilot analysis of the initial data and demonstrated an enrichment of *de novo* mutations in enhancers active within the human brain. Strikingly, the most deleterious variants, insertion-deletion variants, show the strongest enrichment.

Systems biological approaches. I am particularly interested in designing systems analyses that constrain spatial, temporal, and cell-type specific variables of disease pathology. This is a salient question in neuropsychiatric disorders because the genetic and phenotypic heterogeneity of this disorder has made understanding the neurobiology difficult. Hence, we have limited information with regard to when and where to look in model systems for specific ASD-related mechanisms. To address this challenge, I developed a novel spatiotemporal framework for co-expression network analysis that identifies the brain region(s) and developmental time point(s) showing the greatest degree of convergence of genes implicated in a particular disorder. I applied this methodology to the ASD genes identified by WES and identified deep layer cortical glutamatergic neurons in the prefrontal cortex during midfetal development as a critical nexus of ASD risk (Willsey, *Cell* 2013). Excitingly, this method has contributed to a paradigm shift in the field. We followed up this finding by using gene expression data from these brain regions alongside transmitted and *de novo* variation to increase our power to identify ASD-associated genes (Liu, *Mol. Autism* 2014). I am now applying this methodology to the genes we have identified in TS and I am developing methods to leverage non-coding mutations from WGS in a similar framework that relies on the spatial, temporal, and cell-type specificity of regulatory loci.

Model systems. After delineating the co-expression networks in midfetal prefrontal cortex, we utilized human neural stem cells and embryonic mouse cortex to identify complementary regulatory networks, mediated by the chromatin modifier and ASD gene, CHD8, and used these networks to further increase gene discovery (Cotney, *Nat. Commun*. 2015). We are expanding this result by assessing additional ASD-associated chromatin modifiers. I have recently started several collaborations at UCSF. With Nevan Krogan I will characterize the protein-protein interaction networks of the top ASD genes. This will be conducted in human induced pluripotent stem cells (iPSCs) in collaboration with Martin Kampmann at UCSF and Michael Ward at the NIH. Additionally, in collaboration with Martin and Michael, as well as Sergiu Pasca at Stanford, I aim to use CRISPR-mediated interference to repress expression of candidate ASD genes in iPSC-derived neurons, followed by high-throughput screening with techniques such as RNA-Seq. Once reliable phenotypes are established, we will expand this work to human cortical spheroids (Pasca Lab). Steve Finkbeiner and I have also discussed leveraging his high-throughput system for highly-parallelized cellular phenotyping.

Generation of 'omics datasets. As part of a longstanding collaborative group that includes Drs. State and Sanders, as well as investigators at Yale, UPitt, and CMU, we are generating RNA-Seq and ChIP-Seq data from developing human, primate, and mouse brain. Many 'omics datasets are also being generated in the model systems projects detailed above, e.g. proteomics data generated from elucidating protein-protein interaction networks.

RESEARCH AWARDS - CURRENT

1. R01 MH109901 | Multi-PI | 10 % effort | State/Sanders/Willsey (PI)
   NIH | 03/01/2016 | 02/28/2019
   1/3 Multidimensional investigation of the etiology of autism spectrum disorder | $ 142,630 direct/yr 1 | $ 428,415 total
   In this proposal we aim to: 1) Generate a list of ASD-associated genes; 2) Identify points of convergence between these genes in biological data (e.g. gene regulation, expression, sexual dimorphism, cell-type); and 3) Validate these points of convergence in model systems using CRISPR in mice and human iPSCs. JIT Submitted.

2. UCSF Ref P0512536 | Multi-PI | 8 % effort | State (contact) / Sanders / Willsey (PI)
   NIH/NIMH | 07/01/2016 | 06/30/2020
   3/3 Integrative Genomic Analysis of Human Brain Development and Autism | $ 204,173 direct/yr 1 | $ 818,341 total
   To help determine which parts of the developing brain and molecular processes are involved in ASD, thus improving our understanding of the disease and contributing to the development of diagnostic tests to detect such changes. JIT Submitted.

3. P50 MH106934 | Investigator | 30 % effort | Sestan (PI)
   NIH | 9/19/2014 | 7/31/2019
   Functional Genomics of Human Brain Development | $ 124,505 direct/yr 1 | $ 622,505 total
   The purpose of this grant is to analyze how variation in the regulatory architecture of the genome contributes to pathogenesis of autism spectrum disorder (ASD).

4. P50 MH106934-03S | Co-Investigator | 10 % effort | Sestan (PI)
   NIH/NIMH | 06/01/2016 | 05/31/2018
   Functional Genomics of Human Brain Development | $ 107,795 direct/yr 1 | $ 215,129 total
   To support the discovery and characterization of developmentally regulated and human-specific non-coding functional genomic elements in multiple regions of the developing human brain. JIT Submitted.

5. U01 MH105575-03S1 Co-Investigator 5 % effort State (PI)
NIH/NIMH 06/01/2016 05/31/2018
2/3-Identifying regulatory mutations that influence neuropsychiatric disease (Supplement) $ 49,441 direct/yr 1 $ 98,882 total
The purpose of the supplement is to harmonize phenotype data across the four projects to permit cross-disorder and cross-project genetic analyses; to identify heritable phenotypic features of the combined dataset (symptom level and quantitative trait data) that contribute to our understanding of the genetic architecture of the NIMH Research Domain Criteria (RDoC); and to develop a WGSPD phenotype database that will facilitate the above analyses and encourage data sharing, both within the consortium and with the scientific community broadly. JIT Submitted.

6. U01 MH105575 Investigator (Non-Key) 35 % effort State (PI)
NIH/NIMH 09/18/2014 07/31/2018
2/3-Identifying regulatory mutations that influence neuropsychiatric disease $ 830,233 direct/yr 1 $ 3,332,389 total
This study aims to develop methods for identifying regulatory mutations relevant to neuropsychiatric disorders. In addition to utilizing existing population genetic data and regulatory maps, whole genome sequence data will be analyzed alongside gene expression data from primary and reprogrammed cells from the same patients.

7. 390222 Co-Investigator 5 % effort State (PI)
Simons Foundation 10/01/2015 09/30/2017
Validation of candidate ASD genes by targeted sequencing with molecular inversion probes $ 90,094 direct/yr 1 $ 183,300 total
This project will use molecular inversion probes (MIPs) to sequence 250 candidate autism spectrum disorder (ASD) risk genes in 15,250 individuals, including 6,250 ASD probands, in order to replicate previously associated genes or to identify new ASD risk genes.

RESEARCH AWARDS - PAST

1. 201110DFS-277697-216529 PI (predoctoral fellowship) Willsey (PI)
Canadian Institutes of Health Research 05/01/2012 05/30/2014
Genome wide analysis of de novo copy number variation in families with autism spectrum disorder $ 35,000 direct/yr 1 $ 105,000 total
We proposed to complete a comprehensive analysis of copy number variants (CNVs), with a focus on de novo variants, in approximately 17,000 samples from the Autism Genetic Resource Exchange and Simons Simplex collections.

2. R01 ES021462-05S1 | Co-Investigator | 20 % effort | Kim (PI)
   NIH | | 7/20/2015 | 3/31/2016
   Interaction between environmental factors and germline de novo mutation rate in autism. (Supplement to The Roles of Environmental Risks and GEX in Increasing ASD Prevalence) | $ 284,043 direct/yr 1 | $ 284,043 total
   This proposal seeks supplemental funds for the current NIEHS grant R01ES021462. While the primary aims of this proposal were to examine environmental exposures and risk for Autism Spectrum Disorder (ASD), the 3rd specific aim was to establish a biorepository of ASD samples from Korea for future studies. This supplement will use the stored samples in the context of a new collaboration between epidemiological researcher Kim and three researchers with expertise in genetics (State, Sanders and Willsey): investigation of the effect of pre-conceptional paternal smoking on the rate of de novo mutation in 30 simplex ASD families.

3. U01 MH100239-03S1 | Co-Investigator | 40 % effort | State (PI)
   NIH | | 08/01/2015 | 07/31/2016
   4/4 The Autism Sequencing Consortium: Autism gene discovery in the >20,000 exomes (Supplement) | $ 767,927 direct/yr 1 | $ 767,927 total
   This project is within the scope of the original Autism Sequencing Consortium (ASC) grant. The rationale for the grant was that the identification of genetic variants conferring high risk for ASD and associated neurodevelopmental disorders would form the bases of studies to understand pathogenesis as well as the bases for novel therapies. Moreover, such variants would also have direct implications for patients and their families in terms of etiological diagnosis, genetic counseling and patient care. This supplement aims to form an important extension to the parent grant by increasing the number of samples sequenced as well as our ability to detect additional ASD-associated genes.

4. 385110 | Multi-PI | 5 % effort | State/Sanders/Willsey/Sestan/Goldstein (PI)
   Simons Foundation | | 11/01/2015 | 04/30/2017
   Extending ASD risk locus discovery to the non-coding genome | | $ 138,198 direct/yr 1 | $ 250,000 total
   We propose to identify de novo single nucleotide variants (SNVs) and insertion-deletion variants (indels) in the 500 Simons Simplex Collection (SSC) quartets whole genome sequenced by the Simons Foundation and the New York Genome Center (NYGC). We will integrate multidimensional datasets from the developing human brain in order to identify relevant functional mutations, with the ultimate goal of identifying non-coding regions associated with ASD risk.

5. 274624	Investigator	10 % effort	State/Sestan/Noonan/Roeder (PI)
Simons Foundation		06/01/2013	06/01/2017
A gene-driven systems biological approach to ASD (Autism Spectrum Disorder) pathology		$ 958,437 direct/yr 1	$ 2,875,311 total

Our proposal is aimed at leveraging the tremendous genetic heterogeneity identified by whole-exome sequencing efforts to identify molecular mechanisms shared by individuals with ASD. By integrating multiple biological datasets including, exome sequencing, RNA-Seq, and ChIP-Seq we aim to identify points of convergence that lead to the ASD phenotype.

PEER REVIEWED PUBLICATIONS

1. Sanders SJ, Murtha MT, Gupta AR, Murdoch JD, Raubeson MJ, **Willsey AJ**, Ercan-Sencicek AG, DiLullo NM, Parikshak NN, Stein JL, Walker MF, Ober GT, Teran NA, Song Y, El-Fishawy P, Murtha RC, Choi M, Overton JD, Bjornson RD, Carriero NJ, Meyer KA, Bilguvar K, Mane SM, Sestan N, Lifton RP, Günel M, Roeder K, Geschwind DH, Devlin B, State MW. De novo mutations revealed by whole-exome sequencing are strongly associated with autism. ***Nature***. 2012 May 10; 485(7397):237-41. PMID: 22495306
2. Klei L, Sanders SJ, Murtha MT, Hus V, Lowe JK, **Willsey AJ**, Moreno-De-Luca D, Yu TW, Fombonne E, Geschwind D, Grice DE, Ledbetter DH, Lord C, Mane SM, Martin CL, Martin DM, Morrow EM, Walsh CA, Melhem NM, Chaste P, Sutcliffe JS, State MW, Cook EH, Roeder K, Devlin B. Common genetic variants, acting additively, are a major source of risk for autism. ***Mol Autism***. 2012; 3(1):9. PMID: 23067556.
3. Watson CT, Steinberg KM, Huddleston J, Warren RL, Malig M, Schein J, **Willsey AJ**, Joy JB, Scott JK, Graves TA, Wilson RK, Holt RA, Eichler EE, Breden F. Complete haplotype sequence of the human immunoglobulin heavy-chain variable, diversity, and joining genes and characterization of allelic and copy-number variation. ***Am J Hum Genet***. 2013 Apr 4; 92(4):530-46. PMID: 23541343.
4. Cross-Disorder Working Group of the Psychiatric Genomics Consortium. Genetic relationship between five psychiatric disorders estimated from genome-wide SNPs. ***Nat Genet***. 2013 Sep; 45(9):984-94. PMID: 23933821.
5. Moreno-De-Luca D, Sanders SJ, **Willsey AJ**, Mulle JG, Lowe JK, Geschwind DH, State MW, Martin CL, Ledbetter DH. Using large clinical data sets to infer pathogenicity for rare copy number variants in autism cohorts. ***Mol Psychiatry***. 2013 Oct; 18(10):1090-5. PMID: 23044707.
6. Chaste P, Klei L, Sanders SJ, Murtha MT, Hus V, Lowe JK, **Willsey AJ**, Moreno-De-Luca D, Yu TW, Fombonne E, Geschwind D, Grice DE, Ledbetter DH, Lord C, Mane SM, Lese Martin C, Martin DM, Morrow EM, Walsh CA, Sutcliffe JS, State MW, Devlin B, Cook EH, Kim SJ. Adjusting head circumference for covariates in autism: clinical correlates of a highly heritable continuous trait. ***Biol Psychiatry***. 2013 Oct 15; 74(8):576-84. PMID: 23746936.
7. **Willsey AJ**, Sanders SJ, Li M, Dong S, Tebbenkamp AT, Muhle RA, Reilly SK, Lin L, Fertuzinhos S, Miller JA, Murtha MT, Bichsel C, Niu W, Cotney J, Ercan-Sencicek AG, Gockley J, Gupta AR, Han W, He X, Hoffman EJ, Klei L, Lei J, Liu W, Liu L, Lu C, Xu X, Zhu Y, Mane SM, Lein ES, Wei L, Noonan JP, Roeder K, Devlin B, Sestan N, State MW.

Coexpression networks implicate human midfetal deep cortical projection neurons in the pathogenesis of autism. ***Cell***. 2013 Nov 21; 155(5):997-1007. PMID: 24267886.

8. Liu L, Lei J, Sanders SJ, **Willsey AJ**, Kou Y, Cicek AE, Klei L, Lu C, He X, Li M, Muhle RA, Ma'ayan A, Noonan JP, Sestan N, McFadden KA, State MW, Buxbaum JD, Devlin B, Roeder K. DAWN: a framework to identify autism genes and subnetworks using gene expression and genetics. ***Mol Autism***. 2014; 5(1):22. PMID: 24602502.
9. *Tebbenkamp AT, ***Willsey AJ**, State MW, Sestan N. The developmental transcriptome of the human brain: implications for neurodevelopmental disorders. ***Curr Opin Neurol***. 2014 Apr; 27(2):149-56. PMID: 24565942. *Equal contribution
10. Chaste P, Klei L, Sanders SJ, Hus V, Murtha MT, Lowe JK, **Willsey AJ**, Moreno-De-Luca D, Yu TW, Fombonne E, Geschwind D, Grice DE, Ledbetter DH, Mane SM, Martin DM, Morrow EM, Walsh CA, Sutcliffe JS, Lese Martin C, Beaudet AL, Lord C, State MW, Cook EH, Devlin B. A Genome-wide Association Study of Autism Using the Simons Simplex Collection: Does Reducing Phenotypic Heterogeneity in Autism Increase Genetic Homogeneity? ***Biol Psychiatry***. 2014 Sep 30. PMID: 25534755
11. Chaste P, Sanders SJ, Mohan KN, Klei L, Song Y, Murtha MT, Hus V, Lowe JK, **Willsey AJ**, Moreno-De-Luca D, Yu TW, Fombonne E, Geschwind D, Grice DE, Ledbetter DH, Lord C, Mane SM, Martin DM, Morrow EM, Walsh CA, Sutcliffe JS, State MW, Martin CL, Devlin B, Beaudet AL, Cook EH, Kim SJ. Modest impact on risk for autism spectrum disorder of rare copy number variants at 15q11.2, specifically breakpoints 1 to 2. ***Autism Res***. 2014 Jun; 7(3):355-62. PMID: 24821083.
12. Dong S, Walker MF, Carriero NJ, DiCola M, **Willsey AJ**, Ye AY, Waqar Z, Gonzalez LE, Overton JD, Frahm S, Keaney JF, Teran NA, Dea J, Mandell JD, Hus Bal V, Sullivan CA, DiLullo NM, Khalil RO, Gockley J, Yuksel Z, Sertel SM, Ercan-Sencicek AG, Gupta AR, Mane SM, Sheldon M, Brooks AI, Roeder K, Devlin B, State MW, Wei L, Sanders SJ. De novo insertions and deletions of predominantly paternal origin are associated with autism spectrum disorder. ***Cell Rep***. 2014 Oct 9; 9(1):16-23. PMID: 25284784.
13. Glessner JT, Bick AG, Ito K, Homsy JG, Rodriguez-Murillo L, Fromer M, Mazaika E, Vardarajan B, Italia M, Leipzig J, DePalma SR, Golhar R, Sanders SJ, Yamrom B, Ronemus M, Iossifov I, **Willsey AJ**, State MW, Kaltman JR, White PS, Shen Y, Warburton D, Brueckner M, Seidman C, Goldmuntz E, Gelb BD, Lifton R, Seidman J, Hakonarson H, Chung WK. Increased frequency of de novo copy number variants in congenital heart disease by integrative analysis of single nucleotide polymorphism array and exome sequence data. ***Circ Res***. 2014 Oct 24; 115(10):884-96. PMID: 25205790.
14. De Rubeis S, He X, Goldberg AP, Poultney CS, Samocha K, Ercument Cicek A, Kou Y, Liu L, Fromer M, Walker S, Singh T, Klei L, Kosmicki J, Fu SC, Aleksic B, Biscaldi M, Bolton PF, Brownfeld JM, Cai J, Campbell NG, Carracedo A, Chahrour MH, Chiocchetti AG, Coon H, Crawford EL, Crooks L, Curran SR, Dawson G, Duketis E, Fernandez BA, Gallagher L, Geller E, Guter SJ, Sean Hill R, Ionita-Laza I, Jimenez Gonzalez P, Kilpinen H, Klauck SM, Kolevzon A, Lee I, Lei J, Lehtimäki T, Lin CF, Ma'ayan A, Marshall CR, McInnes AL, Neale B, Owen MJ, Ozaki N, Parellada M, Parr JR, Purcell S, Puura K, Rajagopalan D, Rehnström K, Reichenberg A, Sabo A, Sachse M, Sanders SJ, Schafer C, Schulte-Rüther M, Skuse D, Stevens C, Szatmari P, Tammimies K, Valladares O, Voran A, Wang LS, Weiss LA, **Willsey AJ**, Yu TW, Yuen RK. Synaptic, transcriptional and chromatin genes disrupted in autism. ***Nature***. 2014 Oct 29. PMID: 25363760
15. Iossifov I, O'Roak BJ, Sanders SJ, Ronemus M, Krumm N, Levy D, Stessman HA, Witherspoon KT, Vives L, Patterson KE, Smith JD, Paeper B, Nickerson DA, Dea J, Dong

S, Gonzalez LE, Mandell JD, Mane SM, Murtha MT, Sullivan CA, Walker MF, Waqar Z, Wei L, **Willsey AJ**, Yamrom B, Lee YH, Grabowska E, Dalkic E, Wang Z, Marks S, Andrews P, Leotta A, Kendall J, Hakker I, Rosenbaum J, Ma B, Rodgers L, Troge J, Narzisi G, Yoon S, Schatz MC, Ye K, McCombie WR, Shendure J, Eichler EE, State MW, Wigler M. The contribution of de novo coding mutations to autism spectrum disorder. ***Nature***. 2014 Oct 29. PMID: 25363768

16. Murdoch JD, Gupta AR, Sanders SJ, Walker MF, Keaney J, Fernandez TV, Murtha MT, Anyanwu S, Ober GT, Raubeson MJ, DiLullo NM, Villa N, Waqar Z, Sullivan C, Gonzalez L, **Willsey AJ**, Choe SY, Neale BM, Daly MJ, State MW. No Evidence for Association of Autism with Rare Heterozygous Point Mutations in Contactin-Associated Protein-Like 2 (CNTNAP2), or in Other Contactin-Associated Proteins or Contactins. ***PLoS Genet***. 2015 Jan; 11(1):e1004852. PMID: 25621974.

17. Sanders SJ, He X, **Willsey AJ**, Ercan-Sencicek AG, Samocha KE, Cicek AE, Murtha MT, Bal VH, Bishop SL, Dong S, Goldberg AP, Jinlu C, Keaney JF, Klei L, Mandell JD, Moreno-De-Luca D, Poultney CS, Robinson EB, Smith L, Solli-Nowlan T, Su MY, Teran NA, Walker MF, Werling DM, Beaudet AL, Cantor RM, Fombonne E, Geschwind DH, Grice DE, Lord C, Lowe JK, Mane SM, Martin DM, Morrow EM, Talkowski ME, Sutcliffe JS, Walsh CA, Yu TW. Insights into Autism Spectrum Disorder Genomic Architecture and Biology from 71 Risk Loci. **Neuron**. 2015 Sep 23; 87(6):1215-33. PMID: 26402605.

18. Psychiatric genome-wide association study analyses implicate neuronal, immune and histone pathways. **Nat Neurosci**. 2015 Feb; 18(2):199-209. PMID: 25599223.

19. Akbarian S, Liu C, Knowles JA, Vaccarino FM, Farnham PJ, Crawford GE, Jaffe AE, Pinto D, Dracheva S, Geschwind DH, Mill J, Nairn AC, Abyzov A, Pochareddy S, Prabhakar S, Weissman S, Sullivan PF, State MW, Weng Z, Peters MA, White KP, Gerstein MB, Amiri A, Armoskus C, Ashley-Koch AE, Bae T, Beckel-Mitchener A, Berman BP, Coetzee GA, Coppola G, Francoeur N, Fromer M, Gao R, Grennan K, Herstein J, Kavanagh DH, Ivanov NA, Jiang Y, Kitchen RR, Kozlenkov A, Kundakovic M, Li M, Li Z, Liu S, Mangravite LM, Mattei E, Markenscoff-Papadimitriou E, Navarro FC, North N, Omberg L, Panchision D, Parikshak N, Poschmann J, Price AJ, Purcaro M, Reddy TE, Roussos P, Schreiner S, Scuderi S, Sebra R, Shibata M, Shieh AW, Skarica M, Sun W, Swarup V, Thomas A, Tsuji J, van Bakel H, Wang D, Wang Y, Wang K, Werling DM, **Willsey AJ**, Witt H, Won H, Wong CC, Wray GA, Wu EY, Xu X, Yao L, Senthil G, Lehner T, Sklar P, Sestan N. The PsychENCODE project. **Nat Neurosci**. 2015 Nov 25; 18(12):1707-12. PMID: 26605881.

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