BIOGRAPHICAL SKETCH

NAME **Stephan J. Sanders**	POSITION TITLE Assistant Professor
eRA COMMONS USER NAME (credential, e.g., agency login) STEPHANS	

EDUCATION/TRAINING *(Begin with baccalaureate or other initial professional education, such as nursing, include postdoctoral training and residency training if applicable.)*

INSTITUTION AND LOCATION	DEGREE *(if applicable)*	MM/YY	FIELD OF STUDY
Nottingham University Medical School, UK	BMedSci	02/01	Medicine
Nottingham University Medical School, UK	BMBS (MD equivalent)	06/03	Medicine
Yale University, School of Medicine, New Haven, CT, USA	Postdoc	09/11	Genetics
Yale University, School of Medicine, New Haven, CT, USA	Ph.D.	05/14	Genetics

A. Personal Statement

Dr Sanders trained as a pediatric physician in the UK before pursuing a research career in genomics and bioinformatics. His work has helped characterize the role of de novo mutation in the etiology of ASD and identified multiple ASD risk loci including de novo duplications of the William's Syndrome region (7q11.23) and de novo loss of function mutations in the sodium channel gene SCN2A. Working with numerous collaborators, he helped implement this approach to gene discovery to analyze over 25,000 samples. Fifty ASD-associated genes were identified and these were recently published in two companion articles in Nature (2014). Finally, he worked as part of a group that integrated spatiotemporal gene expression data from the human brain with these ASD-associated genes, including SCN2A. This approach has implicated deep layer glutamatergic neurons in the frontal cortex during mid-fetal development in the causation of ASD.

B. Positions and Honors

Positions and Employment

2003-2004	Pre-registration House Officer, Internal Medicine, General Surgery, Paediatrics, Queens Medical Center, Nottingham, UK
2004-2005	Senior House Officer, Emergency Medicine, Derbyshire Royal Infirmary, Derby, UK
2005-2006	Senior House Officer, Paediatrics and Neonatal Intensive Care, Northwick Park Hospital, Harrow, London, UK
2006-2007	Senior House Officer, Paediatrics and Neonatal Intensive Care, Chelsea and Westminster Hospital, London, UK
2008-2011	Postdoctoral Research Associate, Child Study Center and Genetics, Yale University Child Study Center, New Haven, CT
2011-2014	Graduate Student, Genetics, Yale University, New Haven, CT
2014-present	Assistant Professor, Psychiatry, University of California, San Francisco (UCSF), San Francisco, CA, USA

Other Experience and Professional Memberships

1998-2008	British Medical Association
2002-2003	Elected Representative, British Medical Association, Medical Academic Staff Committee
2004-present	British Society of Human Genetics
2009-present	American Society for Human Genetics
2013-present	International Society for Autism Research

Honors

2000	Sir Basil Blackwell Prize for highest mark in the first two years of medical school, Nottingham University, UK
2001	First Class Honours (highest) in BMedSci degree, Nottingham University, UK
2001	Bart Prize for outstanding performance, Nottingham University, UK

2001 Distinction in paediatrics, Nottingham University, UK
2001 Distinction in obstetrics and gynaecology, Nottingham University, UK
2003 Honours (highest) in Medical degree, Nottingham University, UK
2003 Wellcome Trust Elective Prize, Wellcome Trust, London, UK
2003 Vandervell research grant, Vandervell Foundation, London, UK
2003 Grade A research award, British Medical and Dental Student's Trust, London, UK
2003 Cochrane Prize for best student in obstetrics and gynaecology, Nottingham University, UK
2004 Student Prize for pediatrics, Royal College of Paediatrics and Child Health, London, UK
2009 Highly commended for Oxford Handbook of the Foundation Programme, 2e, BMA book awards, British Medical Association, London, UK
2011 Gruber Science Fellowship, Gruber Foundation, New Haven, CT, USA
2012 Top 10 scientific contributions to autism for identifying the role of de novo single nucleotide variants in autism and the means to identify autism associated genes, Autism Speaks, New York, NY, USA
2013 Howard Hughes Medical Institute (HHMI) International Student Research Fellowship, Howard Hughes Medical Institute (HHMI), MD, USA

C. Selected Peer-reviewed Publications (selected from 44 peer-reviewed publications**)**

Most relevant to the current application

1. **Sanders SJ**, Ercan-Sencicek AG, Hus V, Luo R, Murtha MT, Moreno-De-Luca D, Chu SH, Moreau MP, Gupta AR, Thomson SA, Mason CE, Bilguvar K, Celestino-Soper PB, Choi M, Crawford EL, Davis L, Wright NR, Dhodapkar RM, DiCola M, DiLullo NM, Fernandez TV, Fielding-Singh V, Fishman DO, Frahm S, Garagaloyan R, Goh GS, Kammela S, Klei L, Lowe JK, Lund SC, McGrew AD, Meyer KA, Moffat WJ, Murdoch JD, O'Roak BJ, Ober GT, Pottenger RS, Raubeson MJ, Song Y, Wang Q, Yaspan BL, Yu TW, Yurkiewicz IR, Beaudet AL, Cantor RM, Curland M, Grice DE, Günel M, Lifton RP, Mane SM, Martin DM, Shaw CA, Sheldon M, Tischfield JA, Walsh CA, Morrow EM, Ledbetter DH, Fombonne E, Lord C, Martin CL, Brooks AI, Sutcliffe JS, Cook EH, Geschwind D, Roeder K, Devlin B, State MW. Multiple recurrent de novo CNVs, including duplications of the 7q11.23 Williams syndrome region, are strongly associated with autism. **Neuron**. 2011 Jun 9; 70(5):863-85. PMID: 21658581. PMCID: PMC3939065.
2. **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 R, Choi M, Overton JD, Bjornson RD, Carrierio NJ, Meyer KA, Bilguvar K, Mane SM, Sestan N, Lifton RP, Günel M, Roeder K, Geschwind D, Devlin B, State MW. (2012) De novo mutations revealed by whole-exome sequencing are strongly associated with autism. **Nature**. 485(7397):237-41. PMID: 22495306. PMCID: PMC3667984.
3. 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. PMCID: PMC2978962
4. 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. PMCID: PMC3939065
5. 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. PMCID: PMC1742851. *** Equal contribution**

Additional recent publications of importance to the field

6. Fernandez TV, **Sanders SJ**, Yurkiewicz IR, Ercan-Sencicek AG, Kim YS, Fishman DO, Raubeson MJ, Song Y, Yasuno K, Ho WS, Bilguvar K, Glessner J, Chu SH, Leckman JF, King RA, Gilbert DL, Heiman GA, Tischfield JA, Hoekstra PJ, Devlin B, Hakonarson H, Mane SM, Günel M, State MW. Rare copy number variants in tourette syndrome disrupt genes in histaminergic pathways and overlap with autism. **Biol Psychiatry**. 2012 Mar 1; 71(5):392-402. PMID: 22169095. PMCID: PMC3282144.
7. 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. PMCID: PMC3282144.
8. He X, **Sanders SJ**, Liu L, De Rubeis S, Lim ET, Sutcliffe JS, Schellenberg GD, Gibbs RA, Daly MJ, Buxbaum JD, State MW, Devlin B, Roeder K. Integrated model of de novo and inherited genetic variants yields greater power to identify risk genes. **PLoS Genet**. 2013; 9(8):e1003671. PMID: 23966865. PMCID: PMC1742851.
9. 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. PMCID: PMC3361440.
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. Moreno-De-Luca D, Moreno-De-Luca A, Cubells JF, **Sanders SJ**. Cross-Disorder Comparison of Four Neuropsychiatric CNV Loci. **Curr Genet Med Rep**. 2014; 2(3):151-161.
12. Gaugler T, Klei L, **Sanders SJ**, Bodea CA, Goldberg AP, Lee AB, Mahajan M, Manaa D, Pawitan Y, Reichert J, Ripke S, Sandin S, Sklar P, Svantesson O, Reichenberg A, Hultman CM, Devlin B, Roeder K, Buxbaum JD. Most genetic risk for autism resides with common variation. **Nat Genet**. 2014 Aug; 46(8):881-5. PMID: 25038753. PMCID: PMC3343226.
13. Samocha KE, Robinson EB, **Sanders SJ**, Stevens C, Sabo A, McGrath LM, Kosmicki JA, Rehnström K, Mallick S, Kirby A, Wall DP, MacArthur DG, Gabriel SB, DePristo M, Purcell SM, Palotie A, Boerwinkle E, Buxbaum JD, Cook EH, Gibbs RA, Schellenberg GD, Sutcliffe JS, Devlin B, Roeder K, Neale BM, Daly MJ. A framework for the interpretation of de novo mutation in human disease. **Nat Genet**. 2014 Sep; 46(9):944-50. PMID: 25086666. PMCID: PMC3282144.
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, Jeremy Willsey A, Yu TW, Yuen RK. Synaptic, transcriptional and chromatin genes disrupted in autism. **Nature**. 2014 Oct 29. PMID: 25363760. PMCID: PMC2978962.
15. Chang J, Gilman SR, Chiang AH, **Sanders SJ**, Vitkup D. Genotype to phenotype relationships in autism spectrum disorders. **Nat Neurosci**. 2014 Dec 22. PMID: 25531569. PMCID: PMC2894694.

D. Research Support: Stephan, J Sanders, BMBS, PhD

Ongoing Research Support

307705 Sanders, SJ (PI) 09/01/2014-08/31/2016
Simons Foundation Autism Research Initiative (SFARI)
Sexually dimorphic gene-expression and regulation to evaluate autism spectrum disorder sex bias
This proposal seeks to: 1) create a map of sexually dimorphic gene expression in the human brain, and 2) identify ASD genes and networks within this map that may explain the strong male sex bias observed in ASD
Role: PI

274624 State, M (PI) 08/01/2013-07/31/2016
Simons Foundation
A gene-driven systems biological approach to ASD (Autism Spectrum Disorder) pathology
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.
Role: Investigator

R01 MH100027 Geschwind, D (PI) 02/01/2013-01/31/2018
National Institute of Mental Health
Autism Genetics Network, Phase II: Increasing Representation of Human Diversity
The purpose of this grant is to undertake a comprehensive genomic analysis of African American children with autism spectrum disorders and to study the barriers to participation in autism genomics research.
Role: Investigator

U01 MH103339 Sestan, N / State, M (PI) 06/15/2014–05/31/2017
NIH/NIMH
Transcriptional and Epigenetic Signatures of Human Brain Development and Autism
The goal of the grant is to provide integrated transcriptional and epigenetic analysis of normal human brain development and autism.
Role: Bioinformatician

Completed Research Support

Howard Hughes Medical Institute Sanders, SJ (PI) 09/01/2013-05/31/2014
Howard Hughes Medical Institute International Student Research Fellowship
This fellowship program supports outstanding international predoctoral students studying in the United States
Role: Fellow

#124827 R08741 State, MW (PI) 07/01/2008-12/31/2012
Simons Foundation
Simons Simplex Collection Genetic Consortium (SSCGC)
The central aim of this proposal is to pursue a multi-site genome-wide scan of Simons Simplex Collection (SSC) to identify and confirm sequence and structural variations contributing to Autism. Currently on NCE.
Role: Investigator

M144095 R11154 State (PI) 12/01/2010-12/31/2013
Simons Foundation
Whole Exome Sequencing in Autism Spectrum Disorders
We propose to complete whole-exome sequencing on 400 trios from the Simons Simplex Collection (SSC), focusing on the detection of sequence and structural variation with particular emphasis on de novo mutations.
Role: Investigator