

# Stephan J Sanders

## List of Publications by Year in descending order

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Version: 2024-02-01

130  
papers

33,303  
citations

15495

65  
h-index

15716

125  
g-index

162  
all docs

162  
docs citations

162  
times ranked

33910  
citing authors

#	ARTICLE	IF	CITATIONS
1	Synaptic, transcriptional and chromatin genes disrupted in autism. <i>Nature</i> , 2014, 515, 209-215.	13.7	2,254
2	The contribution of de novo coding mutations to autism spectrum disorder. <i>Nature</i> , 2014, 515, 216-221.	13.7	2,188
3	Genetic relationship between five psychiatric disorders estimated from genome-wide SNPs. <i>Nature Genetics</i> , 2013, 45, 984-994.	9.4	2,067
4	De novo mutations revealed by whole-exome sequencing are strongly associated with autism. <i>Nature</i> , 2012, 485, 237-241.	13.7	1,863
5	Large-Scale Exome Sequencing Study Implicates Both Developmental and Functional Changes in the Neurobiology of Autism. <i>Cell</i> , 2020, 180, 568-584.e23.	13.5	1,422
6	Predicting Splicing from Primary Sequence with Deep Learning. <i>Cell</i> , 2019, 176, 535-548.e24.	13.5	1,305
7	Insights into Autism Spectrum Disorder Genomic Architecture and Biology from 71 Risk Loci. <i>Neuron</i> , 2015, 87, 1215-1233.	3.8	1,219
8	Multiple Recurrent De Novo CNVs, Including Duplications of the 7q11.23 Williams Syndrome Region, Are Strongly Associated with Autism. <i>Neuron</i> , 2011, 70, 863-885.	3.8	1,146
9	Most genetic risk for autism resides with common variation. <i>Nature Genetics</i> , 2014, 46, 881-885.	9.4	977
10	A framework for the interpretation of de novo mutation in human disease. <i>Nature Genetics</i> , 2014, 46, 944-950.	9.4	943
11	Genomic Relationships, Novel Loci, and Pleiotropic Mechanisms across Eight Psychiatric Disorders. <i>Cell</i> , 2019, 179, 1469-1482.e11.	13.5	935
12	Coexpression Networks Implicate Human Midfetal Deep Cortical Projection Neurons in the Pathogenesis of Autism. <i>Cell</i> , 2013, 155, 997-1007.	13.5	825
13	Transcriptome-wide isoform-level dysregulation in ASD, schizophrenia, and bipolar disorder. <i>Science</i> , 2018, 362, .	6.0	805
14	De novo mutations in histone-modifying genes in congenital heart disease. <i>Nature</i> , 2013, 498, 220-223.	13.7	798
15	Psychiatric genome-wide association study analyses implicate neuronal, immune and histone pathways. <i>Nature Neuroscience</i> , 2015, 18, 199-209.	7.1	701
16	De novo mutations in congenital heart disease with neurodevelopmental and other congenital anomalies. <i>Science</i> , 2015, 350, 1262-1266.	6.0	646
17	Contribution of rare inherited and de novo variants in 2,871 congenital heart disease probands. <i>Nature Genetics</i> , 2017, 49, 1593-1601.	9.4	624
18	Comprehensive functional genomic resource and integrative model for the human brain. <i>Science</i> , 2018, 362, .	6.0	618

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19	Integrative functional genomic analysis of human brain development and neuropsychiatric risks. <i>Science</i> , 2018, 362, .	6.0	516
20	Meta-analysis of GWAS of over 16,000 individuals with autism spectrum disorder highlights a novel locus at 10q24.32 and a significant overlap with schizophrenia. <i>Molecular Autism</i> , 2017, 8, 21.	2.6	495
21	Whole-exome sequencing identifies recessive WDR62 mutations in severe brain malformations. <i>Nature</i> , 2010, 467, 207-210.	13.7	457
22	Sex and gender differences in autism spectrum disorder: summarizing evidence gaps and identifying emerging areas of priority. <i>Molecular Autism</i> , 2015, 6, 36.	2.6	413
23	Polygenic transmission disequilibrium confirms that common and rare variation act additively to create risk for autism spectrum disorders. <i>Nature Genetics</i> , 2017, 49, 978-985.	9.4	401
24	Using Whole-Exome Sequencing to Identify Inherited Causes of Autism. <i>Neuron</i> , 2013, 77, 259-273.	3.8	383
25	Common genetic variants, acting additively, are a major source of risk for autism. <i>Molecular Autism</i> , 2012, 3, 9.	2.6	357
26	Genetic risk for autism spectrum disorders and neuropsychiatric variation in the general population. <i>Nature Genetics</i> , 2016, 48, 552-555.	9.4	326
27	The autism-associated chromatin modifier CHD8 regulates other autism risk genes during human neurodevelopment. <i>Nature Communications</i> , 2015, 6, 6404.	5.8	316
28	L-Histidine Decarboxylase and Tourette's Syndrome. <i>New England Journal of Medicine</i> , 2010, 362, 1901-1908.	13.9	304
29	Refining the role of de novo protein-truncating variants in neurodevelopmental disorders by using population reference samples. <i>Nature Genetics</i> , 2017, 49, 504-510.	9.4	298
30	Deletion 17q12 Is a Recurrent Copy Number Variant that Confers High Risk of Autism and Schizophrenia. <i>American Journal of Human Genetics</i> , 2010, 87, 618-630.	2.6	282
31	Mutations in <i>BCKD-kinase</i> Lead to a Potentially Treatable Form of Autism with Epilepsy. <i>Science</i> , 2012, 338, 394-397.	6.0	272
32	Integrated Model of De Novo and Inherited Genetic Variants Yields Greater Power to Identify Risk Genes. <i>PLoS Genetics</i> , 2013, 9, e1003671.	1.5	253
33	Rare Complete Knockouts in Humans: Population Distribution and Significant Role in Autism Spectrum Disorders. <i>Neuron</i> , 2013, 77, 235-242.	3.8	242
34	An analytical framework for whole-genome sequence association studies and its implications for autism spectrum disorder. <i>Nature Genetics</i> , 2018, 50, 727-736.	9.4	235
35	Genome-wide de novo risk score implicates promoter variation in autism spectrum disorder. <i>Science</i> , 2018, 362, .	6.0	234
36	Increased Frequency of De Novo Copy Number Variants in Congenital Heart Disease by Integrative Analysis of Single Nucleotide Polymorphism Array and Exome Sequence Data. <i>Circulation Research</i> , 2014, 115, 884-896.	2.0	229

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37	Joint Analysis of Psychiatric Disorders Increases Accuracy of Risk Prediction for Schizophrenia, Bipolar Disorder, and Major Depressive Disorder. <i>American Journal of Human Genetics</i> , 2015, 96, 283-294.	2.6	225
38	Transcriptome and epigenome landscape of human cortical development modeled in organoids. <i>Science</i> , 2018, 362, .	6.0	220
39	Progress in Understanding and Treating SCN2A-Mediated Disorders. <i>Trends in Neurosciences</i> , 2018, 41, 442-456.	4.2	210
40	Opposing Effects on Na V 1.2 Function Underlie Differences Between SCN2A Variants Observed in Individuals With Autism Spectrum Disorder or Infantile Seizures. <i>Biological Psychiatry</i> , 2017, 82, 224-232.	0.7	208
41	Copy-Number Disorders Are a Common Cause of Congenital Kidney Malformations. <i>American Journal of Human Genetics</i> , 2012, 91, 987-997.	2.6	201
42	Rare Inherited and De Novo CNVs Reveal Complex Contributions to ASD Risk in Multiplex Families. <i>American Journal of Human Genetics</i> , 2016, 99, 540-554.	2.6	179
43	Modeling non-syndromic autism and the impact of TRPC6 disruption in human neurons. <i>Molecular Psychiatry</i> , 2015, 20, 1350-1365.	4.1	175
44	Genotype to phenotype relationships in autism spectrum disorders. <i>Nature Neuroscience</i> , 2015, 18, 191-198.	7.1	168
45	Rare Copy Number Variants in Tourette Syndrome Disrupt Genes in Histaminergic Pathways and Overlap with Autism. <i>Biological Psychiatry</i> , 2012, 71, 392-402.	0.7	167
46	Genome-wide Transcriptome Profiling Reveals the Functional Impact of Rare De Novo and Recurrent CNVs in Autism Spectrum Disorders. <i>American Journal of Human Genetics</i> , 2012, 91, 38-55.	2.6	160
47	Defining the diverse spectrum of inversions, complex structural variation, and chromothripsis in the morbid human genome. <i>Genome Biology</i> , 2017, 18, 36.	3.8	159
48	De Novo Coding Variants Are Strongly Associated with Tourette Disorder. <i>Neuron</i> , 2017, 94, 486-499.e9.	3.8	155
49	De Novo Insertions and Deletions of Predominantly Paternal Origin Are Associated with Autism Spectrum Disorder. <i>Cell Reports</i> , 2014, 9, 16-23.	2.9	151
50	The Autism-Associated Gene Scn2a Contributes to Dendritic Excitability and Synaptic Function in the Prefrontal Cortex. <i>Neuron</i> , 2019, 103, 673-685.e5.	3.8	148
51	Loss of $\beta$ -catenin function in severe autism. <i>Nature</i> , 2015, 520, 51-56.	13.7	145
52	Using large clinical data sets to infer pathogenicity for rare copy number variants in autism cohorts. <i>Molecular Psychiatry</i> , 2013, 18, 1090-1095.	4.1	140
53	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. <i>American Journal of Human Genetics</i> , 2017, 101, 768-788.	2.6	136
54	A Genome-wide Association Study of Autism Using the Simons Simplex Collection: Does Reducing Phenotypic Heterogeneity in Autism Increase Genetic Homogeneity?. <i>Biological Psychiatry</i> , 2015, 77, 775-784.	0.7	133

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55	Whole genome sequencing in psychiatric disorders: the WGSPD consortium. <i>Nature Neuroscience</i> , 2017, 20, 1661-1668.	7.1	122
56	A common X-linked inborn error of carnitine biosynthesis may be a risk factor for nondysmorphic autism. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2012, 109, 7974-7981.	3.3	118
57	High rate of disease-related copy number variations in childhood onset schizophrenia. <i>Molecular Psychiatry</i> , 2014, 19, 568-572.	4.1	116
58	Exome Sequencing for Prenatal Diagnosis in Nonimmune Hydrops Fetalis. <i>New England Journal of Medicine</i> , 2020, 383, 1746-1756.	13.9	114
59	DAWN: a framework to identify autism genes and subnetworks using gene expression and genetics. <i>Molecular Autism</i> , 2014, 5, 22.	2.6	111
60	Use of array CGH to detect exonic copy number variants throughout the genome in autism families detects a novel deletion in TMLHE. <i>Human Molecular Genetics</i> , 2011, 20, 4360-4370.	1.4	101
61	De Novo Sequence and Copy Number Variants Are Strongly Associated with Tourette Disorder and Implicate Cell Polarity in Pathogenesis. <i>Cell Reports</i> , 2018, 24, 3441-3454.e12.	2.9	91
62	Whole-Genome and RNA Sequencing Reveal Variation and Transcriptomic Coordination in the Developing Human Prefrontal Cortex. <i>Cell Reports</i> , 2020, 31, 107489.	2.9	91
63	A framework for the investigation of rare genetic disorders in neuropsychiatry. <i>Nature Medicine</i> , 2019, 25, 1477-1487.	15.2	90
64	Frequency and Complexity of De Novo Structural Mutation in Autism. <i>American Journal of Human Genetics</i> , 2016, 98, 667-679.	2.6	88
65	De Novo Disruption of the Proteasome Regulatory Subunit PSMD12 Causes a Syndromic Neurodevelopmental Disorder. <i>American Journal of Human Genetics</i> , 2017, 100, 352-363.	2.6	86
66	First glimpses of the neurobiology of autism spectrum disorder. <i>Current Opinion in Genetics and Development</i> , 2015, 33, 80-92.	1.5	79
67	Identification of Developmental and Behavioral Markers Associated With Genetic Abnormalities in Autism Spectrum Disorder. <i>American Journal of Psychiatry</i> , 2017, 174, 576-585.	4.0	73
68	Adjusting Head Circumference for Covariates in Autism: Clinical Correlates of a Highly Heritable Continuous Trait. <i>Biological Psychiatry</i> , 2013, 74, 576-584.	0.7	70
69	A Chromatin Accessibility Atlas of the Developing Human Telencephalon. <i>Cell</i> , 2020, 182, 754-769.e18.	13.5	69
70	Biological concepts in human sodium channel epilepsies and their relevance in clinical practice. <i>Epilepsia</i> , 2020, 61, 387-399.	2.6	65
71	Intellectual Disability Is Associated with Increased Runs of Homozygosity in Simplex Autism. <i>American Journal of Human Genetics</i> , 2013, 93, 103-109.	2.6	63
72	Modest Impact on Risk for Autism Spectrum Disorder of Rare Copy Number Variants at 15q11.2, Specifically Breakpoints 1 to 2. <i>Autism Research</i> , 2014, 7, 355-362.	2.1	59

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73	Whole-exome sequencing in obsessive-compulsive disorder identifies rare mutations in immunological and neurodevelopmental pathways. <i>Translational Psychiatry</i> , 2016, 6, e764-e764.	2.4	59
74	Homozygous loss of DIAPH1 is a novel cause of microcephaly in humans. <i>European Journal of Human Genetics</i> , 2015, 23, 165-172.	1.4	57
75	De novo structural mutation rates and gamete-of-origin biases revealed through genome sequencing of 2,396 families. <i>American Journal of Human Genetics</i> , 2021, 108, 597-607.	2.6	57
76	Paradoxical hyperexcitability from NaV1.2 sodium channel loss in neocortical pyramidal cells. <i>Cell Reports</i> , 2021, 36, 109483.	2.9	57
77	Dual Molecular Effects of Dominant RORA Mutations Cause Two Variants of Syndromic Intellectual Disability with Either Autism or Cerebellar Ataxia. <i>American Journal of Human Genetics</i> , 2018, 102, 744-759.	2.6	51
78	The female protective effect in autism spectrum disorder is not mediated by a single genetic locus. <i>Molecular Autism</i> , 2015, 6, 25.	2.6	50
79	Neuronal and glial 3D chromatin architecture informs the cellular etiology of brain disorders. <i>Nature Communications</i> , 2021, 12, 3968.	5.8	48
80	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. <i>PLoS Genetics</i> , 2015, 11, e1004852.	1.5	47
81	Paired-Duplication Signatures Mark Cryptic Inversions and Other Complex Structural Variation. <i>American Journal of Human Genetics</i> , 2015, 97, 170-176.	2.6	45
82	DIXDC1 contributes to psychiatric susceptibility by regulating dendritic spine and glutamatergic synapse density via GSK3 and Wnt/ $\beta$ -catenin signaling. <i>Molecular Psychiatry</i> , 2018, 23, 467-475.	4.1	44
83	Peabody Picture Vocabulary Test: Proxy for Verbal IQ in Genetic Studies of Autism Spectrum Disorder. <i>Journal of Autism and Developmental Disorders</i> , 2017, 47, 1073-1085.	1.7	40
84	Body composition at high altitude: a randomized placebo-controlled trial of dietary carbohydrate supplementation. <i>American Journal of Clinical Nutrition</i> , 2009, 90, 1193-1202.	2.2	39
85	Appositions between cocaine and amphetamine-related transcript- and gonadotropin releasing hormone-immunoreactive neurons in the hypothalamus of the Siberian hamster. <i>Neuroscience Letters</i> , 2001, 314, 111-114.	1.0	35
86	Physiological and Psychological Illness Symptoms at High Altitude and Their Relationship With Acute Mountain Sickness: A Prospective Cohort Study. <i>Journal of Travel Medicine</i> , 2012, 19, 210-219.	1.4	30
87	Children with autism spectrum disorder who improve with fever: Insights from the Simons Simplex Collection. <i>Autism Research</i> , 2018, 11, 175-184.	2.1	30
88	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. <i>American Journal of Human Genetics</i> , 2019, 104, 530-541.	2.6	30
89	The female protective effect against autism spectrum disorder. <i>Cell Genomics</i> , 2022, 2, 100134.	3.0	30
90	A balanced t(10;15) translocation in a male patient with developmental language disorder. <i>European Journal of Medical Genetics</i> , 2012, 55, 128-131.	0.7	22

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91	Cross-Disorder Comparison of Four Neuropsychiatric CNV Loci. <i>Current Genetic Medicine Reports</i> , 2014, 2, 151-161.	1.9	22
92	Developmental dynamics of voltage-gated sodium channel isoform expression in the human and mouse brain. <i>Genome Medicine</i> , 2021, 13, 135.	3.6	19
93	Standardizing the Next Generation of Bioinformatics Software Development with BioHDF (HDF5). <i>Advances in Experimental Medicine and Biology</i> , 2010, 680, 693-700.	0.8	18
94	A Markov random field-based approach to characterizing human brain development using spatial-temporal transcriptome data. <i>Annals of Applied Statistics</i> , 2015, 9, 429-451.	0.5	18
95	Differential excitatory vs inhibitory SCN expression at single cell level regulates brain sodium channel function in neurodevelopmental disorders. <i>European Journal of Paediatric Neurology</i> , 2020, 24, 129-133.	0.7	18
96	Extrathymic Aire-expressing cells support maternal-fetal tolerance. <i>Science Immunology</i> , 2021, 6, .	5.6	17
97	High-throughput characterization of the role of non-B DNA motifs on promoter function. <i>Cell Genomics</i> , 2022, 2, 100111.	3.0	17
98	Intergenerational Neuroimaging of Human Brain Circuitry. <i>Trends in Neurosciences</i> , 2016, 39, 644-648.	4.2	16
99	Searching for Potocki-Lupski syndrome phenotype: A patient with language impairment and no autism. <i>Brain and Development</i> , 2012, 34, 700-703.	0.6	15
100	Clinical impact of splicing in neurodevelopmental disorders. <i>Genome Medicine</i> , 2020, 12, 36.	3.6	15
101	Constructing and optimizing 3D atlases from 2D data with application to the developing mouse brain. <i>ELife</i> , 2021, 10, .	2.8	15
102	A model and test for coordinated polygenic epistasis in complex traits. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2021, 118, .	3.3	15
103	Homeostatic plasticity fails at the intersection of autism-gene mutations and a novel class of common genetic modifiers. <i>ELife</i> , 2020, 9, .	2.8	14
104	Next-Generation Sequencing in Autism Spectrum Disorder. <i>Cold Spring Harbor Perspectives in Medicine</i> , 2019, 9, a026872.	2.9	13
105	Patterns of delay in early gross motor and expressive language milestone attainment in probands with genetic conditions versus idiopathic ASD from SFARI registries. <i>Journal of Child Psychology and Psychiatry and Allied Disciplines</i> , 2021, 62, 1297-1307.	3.1	13
106	Prenatal exposure to paternal smoking and likelihood for autism spectrum disorder. <i>Autism</i> , 2021, 25, 1946-1959.	2.4	12
107	Not All Autism Genes Are Created Equal: A Response to Myers et Al.. <i>American Journal of Human Genetics</i> , 2020, 107, 1000-1003.	2.6	11
108	A complex chromosomal rearrangement involving chromosomes 2, 5, and X in autism spectrum disorder. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2012, 159B, 529-536.	1.1	10

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109	Whole-Brain Image Analysis and Anatomical Atlas 3D Generation Using MagellanMapper. <i>Current Protocols in Neuroscience</i> , 2020, 94, e104.	2.6	9
110	Hereditary hyperferritinaemia-cataract syndrome and differential diagnosis of hereditary haemochromatosis. <i>Postgraduate Medical Journal</i> , 2003, 79, 600-601.	0.9	6
111	Attention Finally Being Paid to Girls at Risk of Autism. <i>Journal of the American Academy of Child and Adolescent Psychiatry</i> , 2016, 55, 159-160.	0.3	6
112	Automating Installation of the Integrating Biology and the Bedside (i2b2) Platform. <i>Biomedical Informatics Insights</i> , 2018, 10, 117822261877774.	4.6	6
113	Exome sequencing of fetuses with congenital diaphragmatic hernia supports a causal role for NR2F2, PTPN11, and WT1 variants. <i>American Journal of Surgery</i> , 2022, 223, 182-186.	0.9	6
114	Building a Precision Medicine Delivery Platform for Clinics: The University of California, San Francisco, BRIDGE Experience. <i>Journal of Medical Internet Research</i> , 2022, 24, e34560.	2.1	6
115	A biomedical open knowledge network harnesses the power of AI to understand deep human biology. <i>AI Magazine</i> , 2022, 43, 46-58.	1.4	5
116	Appreciating the Population-wide Impact of Copy Number Variants on Cognition. <i>Biological Psychiatry</i> , 2017, 82, 78-80.	0.7	4
117	Detection of subtle white matter lesions in MRI through texture feature extraction and boundary delineation using an embedded clustering strategy. <i>Scientific Reports</i> , 2022, 12, 4433.	1.6	4
118	Deletion 17q12 Is a Recurrent Copy Number Variant that Confers High Risk of Autism and Schizophrenia. <i>American Journal of Human Genetics</i> , 2011, 88, 121.	2.6	3
119	Harnessing rare variants in neuropsychiatric and neurodevelopment disorders—a Keystone Symposia report. <i>Annals of the New York Academy of Sciences</i> , 2021, , .	1.8	2
120	Enhancing Clinical Information Display to Improve Patient Encounters: Human-Centered Design and Evaluation of the Parkinson Disease-BRIDGE Platform. <i>JMIR Human Factors</i> , 2022, 9, e33967.	1.0	2
121	Gene coexpression modules in human cognition. <i>Nature Neuroscience</i> , 2016, 19, 173-175.	7.1	1
122	706. Wnt/ $\beta$ -Catenin Pathway Contributions to Dendritic Spine and Glutamatergic Synapse Formation Responsive to Lithium-Mediated GSK3 Inhibition. <i>Biological Psychiatry</i> , 2017, 81, S286.	0.7	1
123	LIMITED CONTRIBUTION OF RARE, NONCODING VARIATION TO AUTISM SPECTRUM DISORDER FROM SEQUENCING OF 2,076 GENOMES IN QUARTET FAMILIES. <i>European Neuropsychopharmacology</i> , 2019, 29, S784-S785.	0.3	1
124	Exome Sequencing for Prenatal Diagnosis in Nonimmune Hydrops Fetalis. <i>Obstetrical and Gynecological Survey</i> , 2021, 76, 139-141.	0.2	1
125	Mental Retardation (Former Term). , 2013, , 1841-1841.		0
126	Mutual Gaze. , 2013, , 1966-1967.		0



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127	The Newly Emerging View of the Genome. , 2016, , 3-26.		0
128	Whole-Exome Sequencing in Fetuses with Congenital Diaphragmatic Hernias: Known and Novel Genetic Mutation. Journal of the American College of Surgeons, 2020, 231, S217.	0.2	0
129	7q11.23 Duplications. , 2021, , 10-10.		0
130	Assessing the utility of electronic measures as a proxy for cognitive ability. Autism Research, 0, , .	2.1	0