Stephan J Sanders

List of Publications by Year
in descending order

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	Exome sequencing of fetuses with congenital diaphragmatic hernia supports a causal role for NR2F2, PTPN11, and WT1 variants. American Journal of Surgery, 2022, 223, 182-186.	0.9	6
2	Building a Precision Medicine Delivery Platform for Clinics: The University of California, San Francisco, BRIDGE Experience. Journal of Medical Internet Research, 2022, 24, e34560.	2.1	6
3	Detection of subtle white matter lesions in MRI through texture feature extraction and boundary delineation using an embedded clustering strategy. Scientific Reports, 2022, 12, 4433.	1.6	4
4	High-throughput characterization of the role of non-B DNA motifs on promoter function. Cell Genomics, 2022, 2, 100111.	3.0	17
5	Enhancing Clinical Information Display to Improve Patient Encounters: Human-Centered Design and Evaluation of the Parkinson Disease-BRIDGE Platform. JMIR Human Factors, 2022, 9, e33967.	1.0	2
6	A biomedical open knowledge network harnesses the power of AI to understand deep human biology. AI Magazine, 2022, 43, 46-58.	1.4	5
7	The female protective effect against autism spectrum disorder. Cell Genomics, 2022, 2, 100134.	3.0	30
8	7q11.23 Duplications. , 2021, , 10-10.		0
9	Constructing and optimizing 3D atlases from 2D data with application to the developing mouse brain. ELife, 2021, 10, .	2.8	15
10	Exome Sequencing for Prenatal Diagnosis in Nonimmune Hydrops Fetalis. Obstetrical and Gynecological Survey, 2021, 76, 139-141.	0.2	1
11	A model and test for coordinated polygenic epistasis in complex traits. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	3.3	15
12	Prenatal exposure to paternal smoking and likelihood for autism spectrum disorder. Autism, 2021, 25, 1946-1959.	2.4	12
13	De novo structural mutation rates and gamete-of-origin biases revealed through genome sequencing of 2,396 families. American Journal of Human Genetics, 2021, 108, 597-607.	2.6	57
14	Neuronal and glial 3D chromatin architecture informs the cellular etiology of brain disorders. Nature Communications, 2021, 12, 3968.	5.8	48
15	Extrathymic Aire -expressing cells support maternal-fetal tolerance. Science Immunology, 2021, 6, .	5.6	17
16	Patterns of delay in early gross motor and expressive language milestone attainment in probands with genetic conditions versus idiopathic ASD from SFARI registries. Journal of Child Psychology and Psychiatry and Allied Disciplines, 2021, 62, 1297-1307.	3.1	13
17	Paradoxical hyperexcitability from NaV1.2 sodium channel loss in neocortical pyramidal cells. Cell Reports, 2021, 36, 109483.	2.9	57
18	Harnessing rare variants in neuropsychiatric and neurodevelopment disordersâ€"a Keystone Symposia report. Annals of the New York Academy of Sciences, 2021, , .	1.8	2

#	Article	IF	Citations
19	Developmental dynamics of voltage-gated sodium channel isoform expression in the human and mouse brain. Genome Medicine, 2021, 13, 135.	3.6	19
20	Differential excitatory vs inhibitory SCN expression at single cell level regulates brain sodium channel function in neurodevelopmental disorders. European Journal of Paediatric Neurology, 2020, 24, 129-133.	0.7	18
21	Exome Sequencing for Prenatal Diagnosis in Nonimmune Hydrops Fetalis. New England Journal of Medicine, 2020, 383, 1746-1756.	13.9	114
22	Wholeâ€Brain Image Analysis and Anatomical Atlas 3D Generation Using MagellanMapper. Current Protocols in Neuroscience, 2020, 94, e104.	2.6	9
23	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
24	Not All Autism Genes Are Created Equal: A Response to Myers etÂal.. American Journal of Human Genetics, 2020, 107, 1000-1003.	2.6	11
25	A Chromatin Accessibility Atlas of the Developing Human Telencephalon. Cell, 2020, 182, 754-769.e18.	13.5	69
26	Biological concepts in human sodium channel epilepsies and their relevance in clinical practice. Epilepsia, 2020, 61, 387-399.	2.6	65
27	Large-Scale Exome Sequencing Study Implicates Both Developmental and Functional Changes in the Neurobiology of Autism. Cell, 2020, 180, 568-584.e23.	13.5	1,422
28	Whole-Genome and RNA Sequencing Reveal Variation and Transcriptomic Coordination in the Developing Human Prefrontal Cortex. Cell Reports, 2020, 31, 107489.	2.9	91
29	Clinical impact of splicing in neurodevelopmental disorders. Genome Medicine, 2020, 12, 36.	3.6	15
30	Homeostatic plasticity fails at the intersection of autism-gene mutations and a novel class of common genetic modifiers. ELife, 2020, 9, .	2.8	14
31	The Autism-Associated Gene Scn2a Contributes to Dendritic Excitability and Synaptic Function in the Prefrontal Cortex. Neuron, 2019, 103, 673-685.e5.	3.8	148
32	A framework for the investigation of rare genetic disorders in neuropsychiatry. Nature Medicine, 2019, 25, 1477-1487.	15.2	90
33	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. American Journal of Human Genetics, 2019, 104, 530-541.	2.6	30
34	LIMITED CONTRIBUTION OF RARE, NONCODING VARIATION TO AUTISM SPECTRUM DISORDER FROM SEQUENCING OF 2,076 GENOMES IN QUARTET FAMILIES. European Neuropsychopharmacology, 2019, 29, S784-S785.	0.3	1
35	Genomic Relationships, Novel Loci, and Pleiotropic Mechanisms across Eight Psychiatric Disorders. Cell, 2019, 179, 1469-1482.e11.	13.5	935
36	Next-Generation Sequencing in Autism Spectrum Disorder. Cold Spring Harbor Perspectives in Medicine, 2019, 9, a026872.	2.9	13

#	Article	IF	Citations
37	Predicting Splicing from Primary Sequence with Deep Learning. Cell, 2019, 176, 535-548.e24.	13.5	1,305
38	Progress in Understanding and Treating SCN2A-Mediated Disorders. Trends in Neurosciences, 2018, 41, 442-456.	4.2	210
39	An analytical framework for whole-genome sequence association studies and its implications for autism spectrum disorder. Nature Genetics, 2018, 50, 727-736.	9.4	235
40	Dual Molecular Effects of Dominant RORA Mutations Cause Two Variants of Syndromic Intellectual Disability with Either Autism or Cerebellar Ataxia. American Journal of Human Genetics, 2018, 102, 744-759.	2.6	51
41	DIXDC1 contributes to psychiatric susceptibility by regulating dendritic spine and glutamatergic synapse density via GSK3 and Wnt/Î²-catenin signaling. Molecular Psychiatry, 2018, 23, 467-475.	4.1	44
42	Children with autism spectrum disorder who improve with fever: Insights from the Simons Simplex Collection. Autism Research, 2018, 11, 175-184.	2.1	30
43	Genome-wide de novo risk score implicates promoter variation in autism spectrum disorder. Science, 2018, 362, .	6.0	234
44	Transcriptome and epigenome landscape of human cortical development modeled in organoids. Science, 2018, 362, .	6.0	220
45	Integrative functional genomic analysis of human brain development and neuropsychiatric risks. Science, 2018, 362, .	6.0	516
46	Transcriptome-wide isoform-level dysregulation in ASD, schizophrenia, and bipolar disorder. Science, 2018, 362, .	6.0	805
47	Comprehensive functional genomic resource and integrative model for the human brain. Science, 2018, 362, .	6.0	618
48	De Novo Sequence and Copy Number Variants Are Strongly Associated with Tourette Disorder and Implicate Cell Polarity in Pathogenesis. Cell Reports, 2018, 24, 3441-3454.e12.	2.9	91
49	Automating Installation of the Integrating Biology and the Bedside (i2b2) Platform. Biomedical Informatics Insights, 2018, 10, 117822261877774.	4.6	6
50	De Novo Disruption of the Proteasome Regulatory Subunit PSMD12 Causes a Syndromic Neurodevelopmental Disorder. American Journal of Human Genetics, 2017, 100, 352-363.	2.6	86
51	Opposing Effects on Na V 1.2 Function Underlie Differences Between SCN2A Variants Observed in Individuals With Autism Spectrum Disorder or Infantile Seizures. Biological Psychiatry, 2017, 82, 224-232.	0.7	208
52	Peabody Picture Vocabulary Test: Proxy for Verbal IQ in Genetic Studies of Autism Spectrum Disorder. Journal of Autism and Developmental Disorders, 2017, 47, 1073-1085.	1.7	40
53	Defining the diverse spectrum of inversions, complex structural variation, and chromothripsis in the morbid humanÂgenome. Genome Biology, 2017, 18, 36.	3.8	159
54	Identification of Developmental and Behavioral Markers Associated With Genetic Abnormalities in Autism Spectrum Disorder. American Journal of Psychiatry, 2017, 174, 576-585.	4.0	73

#	Article	IF	Citations
55	Refining the role of de novo protein-truncating variants in neurodevelopmental disorders by using population reference samples. Nature Genetics, 2017, 49, 504-510.	9.4	298
56	Polygenic transmission disequilibrium confirms that common and rare variation act additively to create risk for autism spectrum disorders. Nature Genetics, 2017, 49, 978-985.	9.4	401
57	706. Wnt/Î²-Catenin Pathway Contributions to Dendritic Spine and Glutamatergic Synapse Formation Responsive to Lithium-Mediated GSK3 Inhibition. Biological Psychiatry, 2017, 81, S286.	0.7	1
58	De Novo Coding Variants Are Strongly Associated with Tourette Disorder. Neuron, 2017, 94, 486-499.e9.	3.8	155
59	Appreciating the Population-wide Impact of Copy Number Variants on Cognition. Biological Psychiatry, 2017, 82, 78-80.	0.7	4
60	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. American Journal of Human Genetics, 2017, 101, 768-788.	2.6	136
61	Contribution of rare inherited and de novo variants in 2,871 congenital heart disease probands. Nature Genetics, 2017, 49, 1593-1601.	9.4	624
62	Whole genome sequencing in psychiatric disorders: the WGSPD consortium. Nature Neuroscience, 2017, 20, 1661-1668.	7.1	122
63	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. Molecular Autism, 2017, 8, 21.	2.6	495
64	Whole-exome sequencing in obsessive-compulsive disorder identifies rare mutations in immunological and neurodevelopmental pathways. Translational Psychiatry, 2016, 6, e764-e764.	2.4	59
65	Intergenerational Neuroimaging of Human Brain Circuitry. Trends in Neurosciences, 2016, 39, 644-648.	4.2	16
66	Rare Inherited and De Novo CNVs Reveal Complex Contributions to ASD Risk in Multiplex Families. American Journal of Human Genetics, 2016, 99, 540-554.	2.6	179
67	The Newly Emerging View of the Genome. , 2016, , 3-26.		0
68	Gene coexpression modules in human cognition. Nature Neuroscience, 2016, 19, 173-175.	7.1	1
69	Genetic risk for autism spectrum disorders and neuropsychiatric variation in the general population. Nature Genetics, 2016, 48, 552-555.	9.4	326
70	Frequency and Complexity of De Novo Structural Mutation in Autism. American Journal of Human Genetics, 2016, 98, 667-679.	2.6	88
71	Attention Finally Being Paid to Girls at Risk of Autism. Journal of the American Academy of Child and Adolescent Psychiatry, 2016, 55, 159-160.	0.3	6
72	A Markov random field-based approach to characterizing human brain development using spatialâ€“temporal transcriptome data. Annals of Applied Statistics, 2015, 9, 429-451.	0.5	18

#	Article	IF	Citations
73	De novo mutations in congenital heart disease with neurodevelopmental and other congenital anomalies. Science, 2015, 350, 1262-1266.	6.0	646
74	Joint Analysis of Psychiatric Disorders Increases Accuracy of Risk Prediction for Schizophrenia, Bipolar Disorder, and Major Depressive Disorder. American Journal of Human Genetics, 2015, 96, 283-294.	2.6	225
75	Psychiatric genome-wide association study analyses implicate neuronal, immune and histone pathways. Nature Neuroscience, 2015, 18, 199-209.	7.1	701
76	Paired-Duplication Signatures Mark Cryptic Inversions and Other Complex Structural Variation. American Journal of Human Genetics, 2015, 97, 170-176.	2.6	45
77	Sex and gender differences in autism spectrum disorder: summarizing evidence gaps and identifying emerging areas of priority. Molecular Autism, 2015, 6, 36.	2.6	413
78	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 Genetics, 2015, 11, e1004852.	1.5	47
79	The female protective effect in autism spectrum disorder is not mediated by a single genetic locus. Molecular Autism, 2015, 6, 25.	2.6	50
80	Loss of Î´-catenin function in severe autism. Nature, 2015, 520, 51-56.	13.7	145
81	The autism-associated chromatin modifier CHD8 regulates other autism risk genes during human neurodevelopment. Nature Communications, 2015, 6, 6404.	5.8	316
82	Insights into Autism Spectrum Disorder Genomic Architecture and Biology from 71 Risk Loci. Neuron, 2015, 87, 1215-1233.	3.8	1,219
83	First glimpses of the neurobiology of autism spectrum disorder. Current Opinion in Genetics and Development, 2015, 33, 80-92.	1.5	79
84	Genotype to phenotype relationships in autism spectrum disorders. Nature Neuroscience, 2015, 18, 191-198.	7.1	168
85	Modeling non-syndromic autism and the impact of TRPC6 disruption in human neurons. Molecular Psychiatry, 2015, 20, 1350-1365.	4.1	175
86	A Genome-wide Association Study of Autism Using the Simons Simplex Collection: Does Reducing Phenotypic Heterogeneity in Autism Increase Genetic Homogeneity?. Biological Psychiatry, 2015, 77, 775-784.	0.7	133
87	Homozygous loss of DIAPH1 is a novel cause of microcephaly in humans. European Journal of Human Genetics, 2015, 23, 165-172.	1.4	57
88	De Novo Insertions and Deletions of Predominantly Paternal Origin Are Associated with Autism Spectrum Disorder. Cell Reports, 2014, 9, 16-23.	2.9	151
89	DAWN: a framework to identify autism genes and subnetworks using gene expression and genetics. Molecular Autism, 2014, 5, 22.	2.6	111
90	The contribution of de novo coding mutations to autism spectrum disorder. Nature, 2014, 515, 216-221.	13.7	2,188

#	Article	IF	Citations
91	Synaptic, transcriptional and chromatin genes disrupted in autism. Nature, 2014, 515, 209-215.	13.7	2,254
92	Modest Impact on Risk for Autism Spectrum Disorder of Rare Copy Number Variants at 15<scp>q</scp>11.2, Specifically Breakpoints 1 to 2. Autism Research, 2014, 7, 355-362.	2.1	59
93	Increased Frequency of De Novo Copy Number Variants in Congenital Heart Disease by Integrative Analysis of Single Nucleotide Polymorphism Array and Exome Sequence Data. Circulation Research, 2014, 115, 884-896.	2.0	229
94	A framework for the interpretation of de novo mutation in human disease. Nature Genetics, 2014, 46, 944-950.	9.4	943
95	Most genetic risk for autism resides with common variation. Nature Genetics, 2014, 46, 881-885.	9.4	977
96	Cross-Disorder Comparison of Four Neuropsychiatric CNV Loci. Current Genetic Medicine Reports, 2014, 2, 151-161.	1.9	22
97	High rate of disease-related copy number variations in childhood onset schizophrenia. Molecular Psychiatry, 2014, 19, 568-572.	4.1	116
98	Intellectual Disability Is Associated with Increased Runs of Homozygosity in Simplex Autism. American Journal of Human Genetics, 2013, 93, 103-109.	2.6	63
99	Mental Retardation (Former Term). , 2013, , 1841-1841.		0
100	Adjusting Head Circumference for Covariates in Autism: Clinical Correlates of a Highly Heritable Continuous Trait. Biological Psychiatry, 2013, 74, 576-584.	0.7	70
101	Coexpression Networks Implicate Human Midfetal Deep Cortical Projection Neurons in the Pathogenesis of Autism. Cell, 2013, 155, 997-1007.	13.5	825
102	Genetic relationship between five psychiatric disorders estimated from genome-wide SNPs. Nature Genetics, 2013, 45, 984-994.	9.4	2,067
103	Rare Complete Knockouts in Humans: Population Distribution and Significant Role in Autism Spectrum Disorders. Neuron, 2013, 77, 235-242.	3.8	242
104	Using Whole-Exome Sequencing to Identify Inherited Causes of Autism. Neuron, 2013, 77, 259-273.	3.8	383
105	De novo mutations in histone-modifying genes in congenital heart disease. Nature, 2013, 498, 220-223.	13.7	798
106	Integrated Model of De Novo and Inherited Genetic Variants Yields Greater Power to Identify Risk Genes. PLoS Genetics, 2013, 9, e1003671.	1.5	253
107	Using large clinical data sets to infer pathogenicity for rare copy number variants in autism cohorts. Molecular Psychiatry, 2013, 18, 1090-1095.	4.1	140
108	Mutual Gaze. , 2013, , 1966-1967.		0

#	Article	IF	Citations
109	A common X-linked inborn error of carnitine biosynthesis may be a risk factor for nondysmorphic autism. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, 7974-7981.	3.3	118
110	Copy-Number Disorders Are a Common Cause of Congenital Kidney Malformations. American Journal of Human Genetics, 2012, 91, 987-997.	2.6	201
111	Rare Copy Number Variants in Tourette Syndrome Disrupt Genes in Histaminergic Pathways and Overlap with Autism. Biological Psychiatry, 2012, 71, 392-402.	0.7	167
112	Mutations in BCKD-kinase Lead to a Potentially Treatable Form of Autism with Epilepsy. Science, 2012, 338, 394-397.	6.0	272
113	Genome-wide Transcriptome Profiling Reveals the Functional Impact of Rare De Novo and Recurrent CNVs in Autism Spectrum Disorders. American Journal of Human Genetics, 2012, 91, 38-55.	2.6	160
114	A balanced t(10;15) translocation in a male patient with developmental language disorder. European Journal of Medical Genetics, 2012, 55, 128-131.	0.7	22
115	Searching for Potockiâ€“Lupski syndrome phenotype: A patient with language impairment and no autism. Brain and Development, 2012, 34, 700-703.	0.6	15
116	Common genetic variants, acting additively, are a major source of risk for autism. Molecular Autism, 2012, 3, 9.	2.6	357
117	De novo mutations revealed by whole-exome sequencing are strongly associated with autism. Nature, 2012, 485, 237-241.	13.7	1,863
118	A complex chromosomal rearrangement involving chromosomes 2, 5, and X in autism spectrum disorder. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2012, 159B, 529-536.	1.1	10
119	Physiological and Psychological Illness Symptoms at High Altitude and Their Relationship With Acute Mountain Sickness: A Prospective Cohort Study. Journal of Travel Medicine, 2012, 19, 210-219.	1.4	30
120	Multiple Recurrent De Novo CNVs, Including Duplications of the 7q11.23 Williams Syndrome Region, Are Strongly Associated with Autism. Neuron, 2011, 70, 863-885.	3.8	1,146
121	Deletion 17q12 Is a Recurrent Copy Number Variant that Confers High Risk of Autism and Schizophrenia. American Journal of Human Genetics, 2011, 88, 121.	2.6	3
122	Use of array CGH to detect exonic copy number variants throughout the genome in autism families detects a novel deletion in TMLHE. Human Molecular Genetics, 2011, 20, 4360-4370.	1.4	101
123	Deletion 17q12 Is a Recurrent Copy Number Variant that Confers High Risk of Autism and Schizophrenia. American Journal of Human Genetics, 2010, 87, 618-630.	2.6	282
124	Whole-exome sequencing identifies recessive WDR62 mutations in severe brain malformations. Nature, 2010, 467, 207-210.	13.7	457
125	L-Histidine Decarboxylase and Tourette's Syndrome. New England Journal of Medicine, 2010, 362, 1901-1908.	13.9	304
126	Standardizing the Next Generation of Bioinformatics Software Development with BioHDF (HDF5). Advances in Experimental Medicine and Biology, 2010, 680, 693-700.	0.8	18

#	Article	IF	Citations
127	Body composition at high altitude: a randomized placebo-controlled trial of dietary carbohydrate supplementation. American Journal of Clinical Nutrition, 2009, 90, 1193-1202.	2.2	39
128	Hereditary hyperferritinaemia-cataract syndrome and differential diagnosis of hereditary haemochromatosis. Postgraduate Medical Journal, 2003, 79, 600-601.	0.9	6
129	Appositions between cocaine and amphetamine-related transcript- and gonadotropin releasing hormone-immunoreactive neurons in the hypothalamus of the Siberian hamster. Neuroscience Letters, 2001, 314, 111-114.	1.0	35
130	Assessing the utility of electronic measures as a proxy for cognitive ability. Autism Research, 0, , .	2.1	0