Jonathan Sebat

List of Publications by Year
in descending order

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75 papers	21,586 citations	76031 42 h-index	81351 76 g-index
92 all docs	92 docs citations	92 times ranked	28843 citing authors

#	Article	IF	Citations
1	Enhancing Discovery of Genetic Variants for Posttraumatic Stress Disorder Through Integration of Quantitative Phenotypes and Trauma Exposure Information. Biological Psychiatry, 2022, 91, 626-636.	0.7	21
2	SNPs, short tandem repeats, and structural variants are responsible for differential gene expression across C57BL/6 and C57BL/10 substrains. Cell Genomics, 2022, 2, 100102.	3.0	9
3	Genes To Mental Health (G2MH): A Framework to Map the Combined Effects of Rare and Common Variants on Dimensions of Cognition and Psychopathology. American Journal of Psychiatry, 2022, 179, 189-203.	4.0	29
4	A phenotypic spectrum of autism is attributable to the combined effects of rare variants, polygenic risk and sex. Nature Genetics, 2022, 54, 1284-1292.	9.4	66
5	Exome sequencing analysis of Japanese autism spectrum disorder case-control sample supports an increased burden of synaptic function-related genes. Translational Psychiatry, 2022, 12, .	2.4	4
6	Autism-linked Cullin3 germline haploinsufficiency impacts cytoskeletal dynamics and cortical neurogenesis through RhoA signaling. Molecular Psychiatry, 2021, 26, 3586-3613.	4.1	26
7	Customized de novo mutation detection for any variant calling pipeline: SynthDNM. Bioinformatics, 2021, 37, 3640-3641.	1.8	3
8	Cortical organoids model early brain development disrupted by 16p11.2 copy number variants in autism. Molecular Psychiatry, 2021, 26, 7560-7580.	4.1	61
9	Developmental and temporal characteristics of clonal sperm mosaicism. Cell, 2021, 184, 4772-4783.e15.	13.5	27
10	Autism risk in offspring can be assessed through quantification of male sperm mosaicism. Nature Medicine, 2020, 26, 143-150.	15.2	76
11	Current progress and future direction in the genetics of PTSD: Focus on the development and contributions of the PGC-PTSD working group. , 2020, , 285-296.		0
12	Inferring the molecular and phenotypic impact of amino acid variants with MutPred2. Nature Communications, 2020, 11, 5918.	5.8	305
13	Identifying schizophrenia patients who carry pathogenic genetic copy number variants using standard clinical assessment: retrospective cohort study. British Journal of Psychiatry, 2020, 216, 275-279.	1.7	12
14	The effects of common structural variants on 3D chromatin structure. BMC Genomics, 2020, 21, 95.	1.2	23
15	Getting to the Cores of Autism. Cell, 2019, 178, 1287-1298.	13.5	204
16	Oligogenic Effects of 16p11.2 Copy-Number Variation on Craniofacial Development. Cell Reports, 2019, 28, 3320-3328.e4.	2.9	34
17	A framework for the investigation of rare genetic disorders in neuropsychiatry. Nature Medicine, 2019, 25, 1477-1487.	15.2	90
18	Pathogenicity and functional impact of non-frameshifting insertion/deletion variation in the human genome. PLoS Computational Biology, 2019, 15, e1007112.	1.5	34

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19	Targeted Treatment of Individuals With Psychosis Carrying a Copy Number Variant Containing a Genomic Triplication of the Glycine Decarboxylase Gene. Biological Psychiatry, 2019, 86, 523-535.	0.7	32
20	Multi-platform discovery of haplotype-resolved structural variation in human genomes. Nature Communications, 2019, 10, 1784.	5.8	636
21	Ranking of non-coding pathogenic variants and putative essential regions of the human genome. Nature Communications, 2019, 10, 5241.	5.8	65
22	Common DNA sequence variation influences 3-dimensional conformation of the human genome. Genome Biology, 2019, 20, 255.	3.8	65
23	Joint Contributions of Rare Copy Number Variants and Common SNPs to Risk for Schizophrenia. American Journal of Psychiatry, 2019, 176, 29-35.	4.0	104
24	Paternally inherited cis-regulatory structural variants are associated with autism. Science, 2018, 360, 327-331.	6.0	174
25	SV2: accurate structural variation genotyping and de novo mutation detection from whole genomes. Bioinformatics, 2018, 34, 1774-1777.	1.8	44
26	Marker chromosome genomic structure and temporal origin implicate a chromoanasynthesis event in a family with pleiotropic psychiatric phenotypes. Human Mutation, 2018, 39, 939-946.	1.1	26
27	Modeling the Interplay Between Neurons and Astrocytes in Autism Using Human Induced Pluripotent Stem Cells. Biological Psychiatry, 2018, 83, 569-578.	0.7	130
28	Divergent Levels of Marker Chromosomes in an hiPSC-Based Model ofÂPsychosis. Stem Cell Reports, 2017, 8, 519-528.	2.3	11
29	FOXP1-related intellectual disability syndrome: a recognisable entity. Journal of Medical Genetics, 2017, 54, 613-623.	1.5	48
30	Contribution of copy number variants to schizophrenia from a genome-wide study of 41,321 subjects. Nature Genetics, 2017, 49, 27-35.	9.4	838
31	When loss-of-function is loss of function: assessing mutational signatures and impact of loss-of-function genetic variants. Bioinformatics, 2017, 33, i389-i398.	1.8	53
32	Genome Tools and Methods. , 2016, , 63-72.		0
33	Frequency and Complexity of De Novo Structural Mutation in Autism. American Journal of Human Genetics, 2016, 98, 667-679.	2.6	88
34	Characterization of molecular and cellular phenotypes associated with a heterozygous CNTNAP2 deletion using patient-derived hiPSC neural cells. NPJ Schizophrenia, 2015, 1, .	2.0	52
35	Spatiotemporal 16p11.2 Protein Network Implicates Cortical Late Mid-Fetal Brain Development and KCTD13-Cul3-RhoA Pathway in Psychiatric Diseases. Neuron, 2015, 85, 742-754.	3.8	139
36	From De Novo Mutations to Personalized Therapeutic Interventions in Autism. Annual Review of Medicine, 2015, 66, 487-507.	5.0	41

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37	An integrated map of structural variation in 2,504 human genomes. Nature, 2015, 526, 75-81.	13.7	1,994
38	The Influence of Microdeletions and Microduplications of 16p11.2 on Global Transcription Profiles. Journal of Child Neurology, 2015, 30, 1947-1953.	0.7	13
39	Protein interaction network of alternatively spliced isoforms from brain links genetic risk factors for autism. Nature Communications, 2014, 5, 3650.	5.8	131
40	Reciprocal Duplication of the Williams-Beuren Syndrome Deletion on Chromosome 7q11.23 Is Associated with Schizophrenia. Biological Psychiatry, 2014, 75, 371-377.	0.7	66
41	Implication of a Rare Deletion at Distal 16p11.2 in Schizophrenia. JAMA Psychiatry, 2013, 70, 253.	6.0	69
42	Formation of Chimeric Genes by Copy-Number Variation as a Mutational Mechanism in Schizophrenia. American Journal of Human Genetics, 2013, 93, 697-710.	2.6	40
43	Fish heads and human disease. Nature, 2012, 485, 318-319.	13.7	4
44	Differential Relationship of DNA Replication Timing to Different Forms of Human Mutation and Variation. American Journal of Human Genetics, 2012, 91, 1033-1040.	2.6	220
45	Whole-Genome Sequencing in Autism Identifies Hot Spots for De Novo Germline Mutation. Cell, 2012, 151, 1431-1442.	13.5	501
46	CNVs: Harbingers of a Rare Variant Revolution in Psychiatric Genetics. Cell, 2012, 148, 1223-1241.	13.5	759
47	forestSV: structural variant discovery through statistical learning. Nature Methods, 2012, 9, 819-821.	9.0	44
48	High Frequencies of De Novo CNVs in Bipolar Disorder and Schizophrenia. Neuron, 2011, 72, 951-963.	3.8	290
49	Duplications of the neuropeptide receptor gene VIPR2 confer significant risk for schizophrenia. Nature, 2011, 471, 499-503.	13.7	296
50	Reduced transcript expression of genes affected by inherited and de novo CNVs in autism. European Journal of Human Genetics, 2011, 19, 727-731.	1.4	109
51	Mapping copy number variation by population-scale genome sequencing. Nature, 2011, 470, 59-65.	13.7	991
52	Modelling schizophrenia using human induced pluripotent stem cells. Nature, 2011, 473, 221-225.	13.7	1,206
53	Inferring Haplotypes of Copy Number Variations From High-Throughput Data With Uncertainty. G3: Genes, Genomes, Genetics, 2011, 1, 35-42.	0.8	4
54	Genomic Duplication and Overexpression of TJP2/ZO-2 Leads to Altered Expression of Apoptosis Genes in Progressive Nonsyndromic Hearing Loss DFNA51. American Journal of Human Genetics, 2010, 87, 101-109.	2.6	95

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55	A recurrent 16p12.1 microdeletion supports a two-hit model for severe developmental delay. Nature Genetics, 2010, 42, 203-209.	9.4	539
56	Reduced NMDAR1 expression in the Sp4 hypomorphic mouse may contribute to endophenotypes of human psychiatric disorders. Human Molecular Genetics, 2010, 19, 3797-3805.	1.4	36
57	Genomewide Association Study of Movement-Related Adverse Antipsychotic Effects. Biological Psychiatry, 2010, 67, 279-282.	0.7	122
58	Sensitive and accurate detection of copy number variants using read depth of coverage. Genome Research, 2009, 19, 1586-1592.	2.4	518
59	Rare structural variants in schizophrenia: one disorder, multiple mutations; one mutation, multiple disorders. Trends in Genetics, 2009, 25, 528-535.	2.9	235
60	Microduplications of 16p11.2 are associated with schizophrenia. Nature Genetics, 2009, 41, 1223-1227.	9.4	646
61	Recurrent Rearrangements of Chromosome 1q21.1 and Variable Pediatric Phenotypes. New England Journal of Medicine, 2008, 359, 1685-1699.	13.9	663
62	Linkage, Association, and Gene-Expression Analyses Identify CNTNAP2 as an Autism-Susceptibility Gene. American Journal of Human Genetics, 2008, 82, 150-159.	2.6	738
63	Rare Structural Variants Disrupt Multiple Genes in Neurodevelopmental Pathways in Schizophrenia. Science, 2008, 320, 539-543.	6.0	1,654
64	Computing Power and Sample Size for Case-Control Association Studies with Copy Number Polymorphism: Application of Mixture-Based Likelihood Ratio Test. PLoS ONE, 2008, 3, e3475.	1.1	15
65	Copy-number variants in patients with a strong family history of pancreatic cancer. Cancer Biology and Therapy, 2007, 6, 1592-1599.	1.5	36
66	A unified genetic theory for sporadic and inherited autism. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 12831-12836.	3.3	284
67	Strong Association of De Novo Copy Number Mutations with Autism. Science, 2007, 316, 445-449.	6.0	2,497
68	Completing the map of human genetic variation. Nature, 2007, 447, 161-165.	13.7	178
69	Major changes in our DNA lead to major changes in our thinking. Nature Genetics, 2007, 39, S3-S5.	9.4	96
70	PROBER: oligonucleotide FISH probe design software. Bioinformatics, 2006, 22, 2437-2438.	1.8	32
71	Application of ROMA (representational oligonucleotide microarray analysis) to patients with cytogenetic rearrangements. Genetics in Medicine, 2005, 7, 111-118.	1.1	32
72	Distribution of short paired duplications in mammalian genomes. Proceedings of the National Academy of Sciences of the United States of America, 2004, 101, 10349-10354.	3.3	23

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73	Large-Scale Copy Number Polymorphism in the Human Genome. Science, 2004, 305, 525-528.	6.0	2,293
74	Representational Oligonucleotide Microarray Analysis: A High-Resolution Method to Detect Genome Copy Number Variation. Genome Research, 2003, 13, 2291-2305.	2.4	376
75	Oligogenic Effects of 16p11.2 Copy Number Variation on Craniofacial Development. SSRN Electronic Journal, 0, , .	0.4	1