Shaun M Purcell

List of Publications by Year
in descending order

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78 papers	73,307 citations	50276 46 h-index	71685 76 g-index
95 all docs	95 docs citations	95 times ranked	92421 citing authors

#	Article	IF	Citations
1	PLINK: A Tool Set for Whole-Genome Association and Population-Based Linkage Analyses. American Journal of Human Genetics, 2007, 81, 559-575.	6.2	26,761
2	Analysis of protein-coding genetic variation in 60,706 humans. Nature, 2016, 536, 285-291.	27.8	9,051
3	Second-generation PLINK: rising to the challenge of larger and richer datasets. GigaScience, 2015, 4, 7.	6.4	8,062
4	Clonal Hematopoiesis and Blood-Cancer Risk Inferred from Blood DNA Sequence. New England Journal of Medicine, 2014, 371, 2477-2487.	27.0	2,669
5	Association analyses of 249,796 individuals reveal 18 new loci associated with body mass index. Nature Genetics, 2010, 42, 937-948.	21.4	2,634
6	Synaptic, transcriptional and chromatin genes disrupted in autism. Nature, 2014, 515, 209-215.	27.8	2,254
7	Genome-wide association analyses identify 44 risk variants and refine the genetic architecture of major depression. Nature Genetics, 2018, 50, 668-681.	21.4	2,224
8	Genetic relationship between five psychiatric disorders estimated from genome-wide SNPs. Nature Genetics, 2013, 45, 984-994.	21.4	2,067
9	Partitioning heritability by functional annotation using genome-wide association summary statistics. Nature Genetics, 2015, 47, 1228-1235.	21.4	2,045
10	De novo mutations in schizophrenia implicate synaptic networks. Nature, 2014, 506, 179-184.	27.8	1,510
11	A polygenic burden of rare disruptive mutations in schizophrenia. Nature, 2014, 506, 185-190.	27.8	1,305
12	Genome-wide association study identifies 30 loci associated with bipolar disorder. Nature Genetics, 2019, 51, 793-803.	21.4	1,191
13	Modeling Linkage Disequilibrium Increases Accuracy of Polygenic Risk Scores. American Journal of Human Genetics, 2015, 97, 576-592.	6.2	1,098
14	The genetic architecture of type 2 diabetes. Nature, 2016, 536, 41-47.	27.8	952
15	Gene expression elucidates functional impact of polygenic risk for schizophrenia. Nature Neuroscience, 2016, 19, 1442-1453.	14.8	952
16	A framework for the interpretation of de novo mutation in human disease. Nature Genetics, 2014, 46, 944-950.	21.4	943
17	Contribution of copy number variants to schizophrenia from a genome-wide study of 41,321 subjects. Nature Genetics, 2017, 49, 27-35.	21.4	838
18	Integrated genotype calling and association analysis of SNPs, common copy number polymorphisms and rare CNVs. Nature Genetics, 2008, 40, 1253-1260.	21.4	712

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19	Partitioning Heritability of Regulatory and Cell-Type-Specific Variants across 11 Common Diseases. American Journal of Human Genetics, 2014, 95, 535-552.	6.2	569
20	Statistical power and significance testing in large-scale genetic studies. Nature Reviews Genetics, 2014, 15, 335-346.	16.3	484
21	Increased burden of ultra-rare protein-altering variants among 4,877 individuals with schizophrenia. Nature Neuroscience, 2016, 19, 1433-1441.	14.8	427
22	Rare loss-of-function variants in SETD1A are associated with schizophrenia and developmental disorders. Nature Neuroscience, 2016, 19, 571-577.	14.8	388
23	Genome-wide association study identifies genetic loci for self-reported habitual sleep duration supported by accelerometer-derived estimates. Nature Communications, 2019, 10, 1100.	12.8	369
24	Quantifying prion disease penetrance using large population control cohorts. Science Translational Medicine, 2016, 8, 322ra9.	12.4	289
25	Genome-wide association analyses of sleep disturbance traits identify new loci and highlight shared genetics with neuropsychiatric and metabolic traits. Nature Genetics, 2017, 49, 274-281.	21.4	280
26	Effect of predicted protein-truncating genetic variants on the human transcriptome. Science, 2015, 348, 666-669.	12.6	252
27	Biological and clinical insights from genetics of insomnia symptoms. Nature Genetics, 2019, 51, 387-393.	21.4	250
28	A Role for Noncoding Variation in Schizophrenia. Cell Reports, 2014, 9, 1417-1429.	6.4	225
29	zCall: a rare variant caller for array-based genotyping. Bioinformatics, 2012, 28, 2543-2545.	4.1	195
30	Reduced Sleep Spindles in Schizophrenia: A Treatable Endophenotype That Links Risk Genes to Impaired Cognition?. Biological Psychiatry, 2016, 80, 599-608.	1.3	171
31	Patterns of genic intolerance of rare copy number variation in 59,898 human exomes. Nature Genetics, 2016, 48, 1107-1111.	21.4	167
32	Gene expression imputation across multiple brain regions provides insights into schizophrenia risk. Nature Genetics, 2019, 51, 659-674.	21.4	154
33	Landscape of Conditional eQTL in Dorsolateral Prefrontal Cortex and Co-localization with Schizophrenia GWAS. American Journal of Human Genetics, 2018, 102, 1169-1184.	6.2	128
34	Polygenic overlap between schizophrenia risk and antipsychotic response: a genomic medicine approach. Lancet Psychiatry,the, 2016, 3, 350-357.	7.4	107
35	GENOME-WIDE ASSOCIATION STUDY (GWAS) AND GENOME-WIDE BY ENVIRONMENT INTERACTION STUDY (GWEIS) OF DEPRESSIVE SYMPTOMS IN AFRICAN AMERICAN AND HISPANIC/LATINA WOMEN. Depression and Anxiety, 2016, 33, 265-280.	4.1	99
36	Exome Sequencing of Familial Bipolar Disorder. JAMA Psychiatry, 2016, 73, 590.	11.0	97

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37	Ultra-rare disruptive and damaging mutations influence educational attainment in the general population. Nature Neuroscience, 2016, 19, 1563-1565.	14.8	90
38	Sex differences in obstructive sleep apnea phenotypes, the multi-ethnic study of atherosclerosis. Sleep, 2020, 43, .	1.1	87
39	Integrated Bayesian analysis of rare exonic variants to identify risk genes for schizophrenia and neurodevelopmental disorders. Genome Medicine, 2017, 9, 114.	8.2	86
40	An Analysis of Two Genome-wide Association Meta-analyses Identifies a New Locus for Broad Depression Phenotype. Biological Psychiatry, 2017, 82, 322-329.	1.3	84
41	Arc Requires PSD95 for Assembly into Postsynaptic Complexes Involved with Neural Dysfunction and Intelligence. Cell Reports, 2017, 21, 679-691.	6.4	79
42	Macro and micro sleep architecture and cognitive performance in older adults. Nature Human Behaviour, 2021, 5, 123-145.	12.0	75
43	De novo CNVs in bipolar affective disorder and schizophrenia. Human Molecular Genetics, 2014, 23, 6677-6683.	2.9	70
44	Genetic modifiers and subtypes in schizophrenia: Investigations of age at onset, severity, sex and family history. Schizophrenia Research, 2014, 154, 48-53.	2.0	68
45	Environmental Mediation and The Twin Design. Behavior Genetics, 2005, 35, 491-498.	2.1	52
46	Genetic Association Analysis of 300 Genes Identifies a Risk Haplotype in SLC18A2 for Post-traumatic Stress Disorder in Two Independent Samples. Neuropsychopharmacology, 2014, 39, 1872-1879.	5.4	49
47	Common Variants on Xq28 Conferring Risk of Schizophrenia in Han Chinese. Schizophrenia Bulletin, 2014, 40, 777-786.	4.3	49
48	A Low-Frequency Inactivating AKT2 Variant Enriched in the Finnish Population Is Associated With Fasting Insulin Levels and Type 2 Diabetes Risk. Diabetes, 2017, 66, 2019-2032.	0.6	47
49	Contribution of Rare Copy Number Variants toÂBipolar Disorder Risk Is Limited to Schizoaffective Cases. Biological Psychiatry, 2019, 86, 110-119.	1.3	45
50	Admixture mapping identifies novel loci for obstructive sleep apnea in Hispanic/Latino Americans. Human Molecular Genetics, 2019, 28, 675-687.	2.9	41
51	The relationship between sleep quality and neurocognition in bipolar disorder. Journal of Affective Disorders, 2015, 187, 156-162.	4.1	33
52	Sequence data and association statistics from 12,940 type 2 diabetes cases and controls. Scientific Data, 2017, 4, 170179.	5.3	31
53	Associations of variants In the hexokinase 1 and interleukin 18 receptor regions with oxyhemoglobin saturation during sleep. PLoS Genetics, 2019, 15, e1007739.	3.5	28
54	Epigenome-wide association analysis of daytime sleepiness in the Multi-Ethnic Study of Atherosclerosis reveals African-American-specific associations. Sleep, 2019, 42, .	1.1	27

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55	Effects of a patient-derived de novo coding alteration of CACNA1I in mice connect a schizophrenia risk gene with sleep spindle deficits. Translational Psychiatry, 2020, 10, 29.	4.8	25
56	No evidence for rare recessive and compound heterozygous disruptive variants in schizophrenia. European Journal of Human Genetics, 2015, 23, 555-557.	2.8	21
57	The Genetics of Endophenotypes of Neurofunction to Understand Schizophrenia (GENUS) consortium: A collaborative cognitive and neuroimaging genetics project. Schizophrenia Research, 2018, 195, 306-317.	2.0	17
58	Statistics for Xâ€chromosome associations. Genetic Epidemiology, 2018, 42, 539-550.	1.3	16
59	Whole-genome association analyses of sleep-disordered breathing phenotypes in the NHLBI TOPMed program. Genome Medicine, 2021, 13, 136.	8.2	16
60	Investigating rare pathogenic/likely pathogenic exonic variation in bipolar disorder. Molecular Psychiatry, 2021, 26, 5239-5250.	7.9	15
61	Sequencing Analysis at 8p23 Identifies Multiple Rare Variants in DLC1 Associated with Sleep-Related Oxyhemoglobin Saturation Level. American Journal of Human Genetics, 2019, 105, 1057-1068.	6.2	10
62	Characterization of Single Gene Copy Number Variants in Schizophrenia. Biological Psychiatry, 2020, 87, 736-744.	1.3	10
63	mTADA is a framework for identifying risk genes from de novo mutations in multiple traits. Nature Communications, 2020, 11, 2929.	12.8	10
64	Sleep and Big Data: harnessing data, technology, and analytics for monitoring sleep and improving diagnostics, prediction, and interventionsâ€”an era for Sleep-Omics?. Sleep, 2021, 44, .	1.1	10
65	Non-rapid eye movement sleep and wake neurophysiology in schizophrenia. ELife, 2022, 11, .	6.0	9
66	Statistical Power and the Classical Twin Design. Twin Research and Human Genetics, 2020, 23, 87-89.	0.6	8
67	Sleep and circadian informatics data harmonization: a workshop report from the Sleep Research Society and Sleep Research Network. Sleep, 2022, 45, .	1.1	8
68	Evaluation of an automated pipeline for large-scale EEG spectral analysis: the National Sleep Research Resource. Sleep Medicine, 2018, 47, 126-136.	1.6	7
69	Targeted Genome Sequencing Identifies Multiple Rare Variants in Caveolin-1 Associated with Obstructive Sleep Apnea. American Journal of Respiratory and Critical Care Medicine, 0, , .	5.6	5
70	Impact of chronic sleep restriction on sleep continuity, sleep structure, and neurobehavioral performance. Sleep, 2022, 45, .	1.1	4
71	Spectral sleep electroencephalographic correlates of sleep efficiency, and discrepancies between actigraphy and selfâ€reported measures, in older men. Journal of Sleep Research, 2021, 30, e13033.	3.2	3
72	Populationâ€based identityâ€byâ€descent mapping combined with exome sequencing to detect rare risk variants for schizophrenia. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2019, 180, 223-231.	1.7	2

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73	Antidepressant Response and Polygenes. Biological Psychiatry, 2013, 73, 600-601.	1.3	1
74	Pleiotropy in complex traits: challenges and strategies. , 0, .		1
75	250 AI-Supported Sleep Staging from Activity and Heart Rate. Sleep, 2021, 44, A101-A101.	1.1	0
76	Linking Sleep and Racial Health Disparities: Characterizing Sleep in the National Sleep Research Resource. Innovation in Aging, 2021, 5, 627-627.	0.1	0
77	0194 Sources of variation in the spectral slope of the sleep EEG. Sleep, 2022, 45, A89-A89.	1.1	0
78	0285 Excessive daytime sleepiness with long sleep duration increases myocardial infarction risk. Sleep, 2022, 45, A129-A129.	1.1	0