

Jonathan K Pritchard

List of Publications by Year in descending order

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118
papers

90,636
citations

3919

88
h-index

18606

119
g-index

155
all docs

155
docs citations

155
times ranked

82795
citing authors

#	ARTICLE	IF	CITATIONS
1	Inference of Population Structure Using Multilocus Genotype Data. <i>Genetics</i> , 2000, 155, 945-959.	1.2	28,015
2	Inference of Population Structure Using Multilocus Genotype Data: Linked Loci and Correlated Allele Frequencies. <i>Genetics</i> , 2003, 164, 1567-1587.	1.2	6,870
3	The Genotype-Tissue Expression (GTEx) pilot analysis: Multitissue gene regulation in humans. <i>Science</i> , 2015, 348, 648-660.	6.0	4,659
4	Inferring weak population structure with the assistance of sample group information. <i>Molecular Ecology Resources</i> , 2009, 9, 1322-1332.	2.2	2,931
5	Inference of population structure using multilocus genotype data: dominant markers and null alleles. <i>Molecular Ecology Notes</i> , 2007, 7, 574-578.	1.7	2,900
6	Genetic Structure of Human Populations. <i>Science</i> , 2002, 298, 2381-2385.	6.0	2,434
7	An Expanded View of Complex Traits: From Polygenic to Omnigenic. <i>Cell</i> , 2017, 169, 1177-1186.	13.5	2,336
8	A Map of Recent Positive Selection in the Human Genome. <i>PLoS Biology</i> , 2006, 4, e72.	2.6	2,329
9	Association Mapping in Structured Populations. <i>American Journal of Human Genetics</i> , 2000, 67, 170-181.	2.6	1,827
10	fastSTRUCTURE: Variational Inference of Population Structure in Large SNP Data Sets. <i>Genetics</i> , 2014, 197, 573-589.	1.2	1,429
11	Convergent adaptation of human lactase persistence in Africa and Europe. <i>Nature Genetics</i> , 2007, 39, 31-40.	9.4	1,375
12	Are Rare Variants Responsible for Susceptibility to Complex Diseases?. <i>American Journal of Human Genetics</i> , 2001, 69, 124-137.	2.6	1,336
13	Understanding mechanisms underlying human gene expression variation with RNA sequencing. <i>Nature</i> , 2010, 464, 768-772.	13.7	1,200
14	Linkage Disequilibrium in Humans: Models and Data. <i>American Journal of Human Genetics</i> , 2001, 69, 1-14.	2.6	1,166
15	Use of Unlinked Genetic Markers to Detect Population Stratification in Association Studies. <i>American Journal of Human Genetics</i> , 1999, 65, 220-228.	2.6	1,112
16	A Systematic Survey of Loss-of-Function Variants in Human Protein-Coding Genes. <i>Science</i> , 2012, 335, 823-828.	6.0	1,095
17	Lineage-specific and single-cell chromatin accessibility charts human hematopoiesis and leukemia evolution. <i>Nature Genetics</i> , 2016, 48, 1193-1203.	9.4	952
18	Traces of Human Migrations in <i>Helicobacter pylori</i> Populations. <i>Science</i> , 2003, 299, 1582-1585.	6.0	922

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19	The Genetics of Human Adaptation: Hard Sweeps, Soft Sweeps, and Polygenic Adaptation. <i>Current Biology</i> , 2010, 20, R208-R215.	1.8	853
20	DNA methylation patterns associate with genetic and gene expression variation in HapMap cell lines. <i>Genome Biology</i> , 2011, 12, R10.	3.8	754
21	Signals of recent positive selection in a worldwide sample of human populations. <i>Genome Research</i> , 2009, 19, 826-837.	2.4	658
22	Using Environmental Correlations to Identify Loci Underlying Local Adaptation. <i>Genetics</i> , 2010, 185, 1411-1423.	1.2	624
23	DNase-sensitivity QTLs are a major determinant of human expression variation. <i>Nature</i> , 2012, 482, 390-394.	13.7	608
24	Informativeness of Genetic Markers for Inference of Ancestry*. <i>American Journal of Human Genetics</i> , 2003, 73, 1402-1422.	2.6	600
25	The allelic architecture of human disease genes: common disease-common variant... or not?. <i>Human Molecular Genetics</i> , 2002, 11, 2417-2423.	1.4	599
26	A high-resolution survey of deletion polymorphism in the human genome. <i>Nature Genetics</i> , 2006, 38, 75-81.	9.4	595
27	Large-scale cis- and trans-eQTL analyses identify thousands of genetic loci and polygenic scores that regulate blood gene expression. <i>Nature Genetics</i> , 2021, 53, 1300-1310.	9.4	590
28	RNA splicing is a primary link between genetic variation and disease. <i>Science</i> , 2016, 352, 600-604.	6.0	574
29	Tracing the peopling of the world through genomics. <i>Nature</i> , 2017, 541, 302-310.	13.7	562
30	Sequencing and Analysis of Neanderthal Genomic DNA. <i>Science</i> , 2006, 314, 1113-1118.	6.0	547
31	Haplotype blocks and linkage disequilibrium in the human genome. <i>Nature Reviews Genetics</i> , 2003, 4, 587-597.	7.7	522
32	Annotation-free quantification of RNA splicing using LeafCutter. <i>Nature Genetics</i> , 2018, 50, 151-158.	9.4	520
33	High-Resolution Mapping of Expression-QTLs Yields Insight into Human Gene Regulation. <i>PLoS Genetics</i> , 2008, 4, e1000214.	1.5	510
34	Accurate inference of transcription factor binding from DNA sequence and chromatin accessibility data. <i>Genome Research</i> , 2011, 21, 447-455.	2.4	501
35	A worldwide survey of haplotype variation and linkage disequilibrium in the human genome. <i>Nature Genetics</i> , 2006, 38, 1251-1260.	9.4	474
36	WASP: allele-specific software for robust molecular quantitative trait locus discovery. <i>Nature Methods</i> , 2015, 12, 1061-1063.	9.0	474

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37	Clines, Clusters, and the Effect of Study Design on the Inference of Human Population Structure. PLoS Genetics, 2005, 1, e70.	1.5	473
38	Effect of read-mapping biases on detecting allele-specific expression from RNA-sequencing data. Bioinformatics, 2009, 25, 3207-3212.	1.8	472
39	Revealing the architecture of gene regulation: the promise of eQTL studies. Trends in Genetics, 2008, 24, 408-415.	2.9	463
40	Identification of Genetic Variants That Affect Histone Modifications in Human Cells. Science, 2013, 342, 747-749.	6.0	429
41	Adaptation “not by sweeps alone. Nature Reviews Genetics, 2010, 11, 665-667.	7.7	410
42	DNA Sequence-Dependent Compartmentalization and Silencing of Chromatin at the Nuclear Lamina. Cell, 2012, 149, 1474-1487.	13.5	405
43	Impact of regulatory variation from RNA to protein. Science, 2015, 347, 664-667.	6.0	399
44	Trans Effects on Gene Expression Can Drive Omnigenic Inheritance. Cell, 2019, 177, 1022-1034.e6.	13.5	385
45	Genetics of 35 blood and urine biomarkers in the UK Biobank. Nature Genetics, 2021, 53, 185-194.	9.4	377
46	Clonal Origin and Evolution of a Transmissible Cancer. Cell, 2006, 126, 477-487.	13.5	375
47	Case-Control Studies of Association in Structured or Admixed Populations. Theoretical Population Biology, 2001, 60, 227-237.	0.5	369
48	The Role of Geography in Human Adaptation. PLoS Genetics, 2009, 5, e1000500.	1.5	358
49	Overcoming the Winner’s Curse: Estimating Penetrance Parameters from Case-Control Data. American Journal of Human Genetics, 2007, 80, 605-615.	2.6	341
50	High-Resolution Mapping of Crossovers Reveals Extensive Variation in Fine-Scale Recombination Patterns Among Humans. Science, 2008, 319, 1395-1398.	6.0	340
51	Detection of human adaptation during the past 2000 years. Science, 2016, 354, 760-764.	6.0	336
52	Genetic Control of Chromatin States in Humans Involves Local and Distal Chromosomal Interactions. Cell, 2015, 162, 1051-1065.	13.5	304
53	Abundant contribution of short tandem repeats to gene expression variation in humans. Nature Genetics, 2016, 48, 22-29.	9.4	291
54	Reduced signal for polygenic adaptation of height in UK Biobank. ELife, 2019, 8, .	2.8	283

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55	The deleterious mutation load is insensitive to recent population history. <i>Nature Genetics</i> , 2014, 46, 220-224.	9.4	279
56	Batch effects and the effective design of single-cell gene expression studies. <i>Scientific Reports</i> , 2017, 7, 39921.	1.6	275
57	Variable prediction accuracy of polygenic scores within an ancestry group. <i>ELife</i> , 2020, 9, .	2.8	268
58	Controls of Nucleosome Positioning in the Human Genome. <i>PLoS Genetics</i> , 2012, 8, e1003036.	1.5	255
59	Methylation QTLs Are Associated with Coordinated Changes in Transcription Factor Binding, Histone Modifications, and Gene Expression Levels. <i>PLoS Genetics</i> , 2014, 10, e1004663.	1.5	255
60	Noisy Splicing Drives mRNA Isoform Diversity in Human Cells. <i>PLoS Genetics</i> , 2010, 6, e1001236.	1.5	254
61	Effect of predicted protein-truncating genetic variants on the human transcriptome. <i>Science</i> , 2015, 348, 666-669.	6.0	252
62	Adaptations to Climate-Mediated Selective Pressures in Humans. <i>PLoS Genetics</i> , 2011, 7, e1001375.	1.5	247
63	ENCODE explained. <i>Nature</i> , 2012, 489, 52-54.	13.7	245
64	Adaptations to Climate in Candidate Genes for Common Metabolic Disorders. <i>PLoS Genetics</i> , 2008, 4, e32.	1.5	238
65	Primate Transcript and Protein Expression Levels Evolve Under Compensatory Selection Pressures. <i>Science</i> , 2013, 342, 1100-1104.	6.0	215
66	A Genome-Wide Study of DNA Methylation Patterns and Gene Expression Levels in Multiple Human and Chimpanzee Tissues. <i>PLoS Genetics</i> , 2011, 7, e1001316.	1.5	196
67	Landscape of stimulation-responsive chromatin across diverse human immune cells. <i>Nature Genetics</i> , 2019, 51, 1494-1505.	9.4	196
68	Confounding from Cryptic Relatedness in Case-Control Association Studies. <i>PLoS Genetics</i> , 2005, 1, e32.	1.5	193
69	Dissecting the regulatory architecture of gene expression QTLs. <i>Genome Biology</i> , 2012, 13, R7.	13.9	188
70	The Functional Consequences of Variation in Transcription Factor Binding. <i>PLoS Genetics</i> , 2014, 10, e1004226.	1.5	187
71	Chromatin accessibility dynamics in a model of human forebrain development. <i>Science</i> , 2020, 367, .	6.0	187
72	Completing the map of human genetic variation. <i>Nature</i> , 2007, 447, 161-165.	13.7	178

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73	The Genetic Architecture of Adaptations to High Altitude in Ethiopia. <i>PLoS Genetics</i> , 2012, 8, e1003110.	1.5	178
74	Admixture facilitates genetic adaptations to high altitude in Tibet. <i>Nature Communications</i> , 2014, 5, 3281.	5.8	172
75	Functional Genetic Variants Revealed by Massively Parallel Precise Genome Editing. <i>Cell</i> , 2018, 175, 544-557.e16.	13.5	166
76	Coregulation of tandem duplicate genes slows evolution of subfunctionalization in mammals. <i>Science</i> , 2016, 352, 1009-1013.	6.0	164
77	Ancient Rome: A genetic crossroads of Europe and the Mediterranean. <i>Science</i> , 2019, 366, 708-714.	6.0	164
78	Genetic variation in MHC proteins is associated with T cell receptor expression biases. <i>Nature Genetics</i> , 2016, 48, 995-1002.	9.4	151
79	Statistical Tests for Admixture Mapping with Case-Control and Cases-Only Data. <i>American Journal of Human Genetics</i> , 2004, 75, 771-789.	2.6	148
80	Comparative RNA sequencing reveals substantial genetic variation in endangered primates. <i>Genome Research</i> , 2012, 22, 602-610.	2.4	145
81	Rapid evolution of the human mutation spectrum. <i>ELife</i> , 2017, 6, .	2.8	144
82	The Genetic and Mechanistic Basis for Variation in Gene Regulation. <i>PLoS Genetics</i> , 2015, 11, e1004857.	1.5	142
83	Large-Scale Clonal Analysis Resolves Aging of the Mouse Hematopoietic Stem Cell Compartment. <i>Cell Stem Cell</i> , 2018, 22, 600-607.e4.	5.2	132
84	Assessing the Performance of the Haplotype Block Model of Linkage Disequilibrium. <i>American Journal of Human Genetics</i> , 2003, 73, 502-515.	2.6	131
85	Thousands of novel translated open reading frames in humans inferred by ribosome footprint profiling. <i>ELife</i> , 2016, 5, .	2.8	122
86	Genome-wide association study of behavioral, physiological and gene expression traits in outbred CFW mice. <i>Nature Genetics</i> , 2016, 48, 919-926.	9.4	119
87	Characterizing natural variation using next-generation sequencing technologies. <i>Trends in Genetics</i> , 2009, 25, 463-471.	2.9	116
88	Impact of regulatory variation across human iPSCs and differentiated cells. <i>Genome Research</i> , 2018, 28, 122-131.	2.4	114
89	Evidence for Extensive Transmission Distortion in the Human Genome. <i>American Journal of Human Genetics</i> , 2004, 74, 62-72.	2.6	111
90	Coalescent-Based Association Mapping and Fine Mapping of Complex Trait Loci. <i>Genetics</i> , 2005, 169, 1071-1092.	1.2	111

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91	Genetic Variation, Not Cell Type of Origin, Underlies the Majority of Identifiable Regulatory Differences in iPSCs. <i>PLoS Genetics</i> , 2016, 12, e1005793.	1.5	111
92	The Contribution of RNA Decay Quantitative Trait Loci to Inter-Individual Variation in Steady-State Gene Expression Levels. <i>PLoS Genetics</i> , 2012, 8, e1003000.	1.5	104
93	Inferring Relevant Cell Types for Complex Traits by Using Single-Cell Gene Expression. <i>American Journal of Human Genetics</i> , 2017, 101, 686-699.	2.6	102
94	Gene Expression Levels Are a Target of Recent Natural Selection in the Human Genome. <i>Molecular Biology and Evolution</i> , 2008, 26, 649-658.	3.5	96
95	Determining the genetic basis of anthracycline-cardiotoxicity by molecular response QTL mapping in induced cardiomyocytes. <i>ELife</i> , 2018, 7, .	2.8	94
96	Interpreting polygenic scores, polygenic adaptation, and human phenotypic differences. <i>Evolution, Medicine and Public Health</i> , 2019, 2019, 26-34.	1.1	90
97	Evolutionary Persistence of DNA Methylation for Millions of Years after Ancient Loss of a De Novo Methyltransferase. <i>Cell</i> , 2020, 180, 263-277.e20.	13.5	87
98	GWAS of three molecular traits highlights core genes and pathways alongside a highly polygenic background. <i>ELife</i> , 2021, 10, .	2.8	77
99	Mutation Rate Variation is a Primary Determinant of the Distribution of Allele Frequencies in Humans. <i>PLoS Genetics</i> , 2016, 12, e1006489.	1.5	63
100	High-resolution mapping of cancer cell networks using co-functional interactions. <i>Molecular Systems Biology</i> , 2018, 14, e8594.	3.2	61
101	Quantification of transplant-derived circulating cell-free DNA in absence of a donor genotype. <i>PLoS Computational Biology</i> , 2017, 13, e1005629.	1.5	60
102	Shared heritability of human face and brain shape. <i>Nature Genetics</i> , 2021, 53, 830-839.	9.4	57
103	Evidence for Weak Selective Constraint on Human Gene Expression. <i>Genetics</i> , 2019, 211, 757-772.	1.2	48
104	A natural mutator allele shapes mutation spectrum variation in mice. <i>Nature</i> , 2022, 605, 497-502.	13.7	38
105	Frequent nonallelic gene conversion on the human lineage and its effect on the divergence of gene duplicates. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2017, 114, 12779-12784.	3.3	37
106	msCentipede: Modeling Heterogeneity across Genomic Sites and Replicates Improves Accuracy in the Inference of Transcription Factor Binding. <i>PLoS ONE</i> , 2015, 10, e0138030.	1.1	37
107	Post-translational buffering leads to convergent protein expression levels between primates. <i>Genome Biology</i> , 2018, 19, 83.	3.8	33
108	Systematic discovery and perturbation of regulatory genes in human T cells reveals the architecture of immune networks. <i>Nature Genetics</i> , 2022, 54, 1133-1144.	9.4	31

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109	Genetic interactions drive heterogeneity in causal variant effect sizes for gene expression and complex traits. <i>American Journal of Human Genetics</i> , 2022, 109, 1286-1297.	2.6	30
110	Reprogramming LCLs to iPSCs Results in Recovery of Donor-Specific Gene Expression Signature. <i>PLoS Genetics</i> , 2015, 11, e1005216.	1.5	29
111	The Effect of Freeze-Thaw Cycles on Gene Expression Levels in Lymphoblastoid Cell Lines. <i>PLoS ONE</i> , 2014, 9, e107166.	1.1	25
112	Remodeling the Specificity of an Endosomal CORVET Tether Underlies Formation of Regulated Secretory Vesicles in the Ciliate <i>Tetrahymena thermophila</i> . <i>Current Biology</i> , 2018, 28, 697-710.e13.	1.8	25
113	Response to Comment on "Genetic Structure of Human Populations". <i>Science</i> , 2003, 300, 1877c-1877.	6.0	20
114	Public Discussion Affects Question Asking at Academic Conferences. <i>American Journal of Human Genetics</i> , 2019, 105, 189-197.	2.6	17
115	Whole Genome Sequencing Identifies a Novel Factor Required for Secretory Granule Maturation in <i>Tetrahymena thermophila</i> . <i>G3: Genes, Genomes, Genetics</i> , 2016, 6, 2505-2516.	0.8	10
116	A Bibliometric History of the Journal <i>GENETICS</i> . <i>Genetics</i> , 2016, 204, 1337-1342.	1.2	7
117	Four makes a party. <i>Nature</i> , 2014, 505, 32-33.	13.7	4
118	Adaptive evolution of conserved non-coding elements in mammals. <i>PLoS Genetics</i> , 2005, preprint, e147.	1.5	2