

Stephen B Montgomery

List of Publications by Citations

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

110
papers

14,978
citations

46
h-index

116
g-index

116
ext. papers

19,467
ext. citations

18.5
avg, IF

5.94
L-index

#	Paper	IF	Citations
110	Integrating common and rare genetic variation in diverse human populations. <i>Nature</i> , 2010 , 467, 52-8	50.4	2135
109	Genetic effects on gene expression across human tissues. <i>Nature</i> , 2017 , 550, 204-213	50.4	2086
108	Transcriptome and genome sequencing uncovers functional variation in humans. <i>Nature</i> , 2013 , 501, 506-514	41.4	1323
107	Population genomics of human gene expression. <i>Nature Genetics</i> , 2007 , 39, 1217-24	36.3	936
106	A systematic survey of loss-of-function variants in human protein-coding genes. <i>Science</i> , 2012 , 335, 823-833	33.3	880
105	Transcriptome genetics using second generation sequencing in a Caucasian population. <i>Nature</i> , 2010 , 464, 773-7	50.4	678
104	Common regulatory variation impacts gene expression in a cell type-dependent manner. <i>Science</i> , 2009 , 325, 1246-50	33.3	607
103	Characterizing the genetic basis of transcriptome diversity through RNA-sequencing of 922 individuals. <i>Genome Research</i> , 2014 , 24, 14-24	9.7	371
102	Patterns of cis regulatory variation in diverse human populations. <i>PLoS Genetics</i> , 2012 , 8, e1002639	6	361
101	Candidate causal regulatory effects by integration of expression QTLs with complex trait genetic associations. <i>PLoS Genetics</i> , 2010 , 6, e1000895	6	339
100	Passive and active DNA methylation and the interplay with genetic variation in gene regulation. <i>ELife</i> , 2013 , 2, e00523	8.9	295
99	RNA Sequencing and Analysis. <i>Cold Spring Harbor Protocols</i> , 2015 , 2015, 951-69	1.2	293
98	Directed evolution using dCas9-targeted somatic hypermutation in mammalian cells. <i>Nature Methods</i> , 2016 , 13, 1036-1042	21.6	288
97	ORegAnno: an open-access community-driven resource for regulatory annotation. <i>Nucleic Acids Research</i> , 2008 , 36, D107-13	20.1	199
96	Overexpression of the Cytokine BAFF and Autoimmunity Risk. <i>New England Journal of Medicine</i> , 2017 , 376, 1615-1626	59.2	198
95	Atheroprotective roles of smooth muscle cell phenotypic modulation and the TCF21 disease gene as revealed by single-cell analysis. <i>Nature Medicine</i> , 2019 , 25, 1280-1289	50.5	198
94	The impact of structural variation on human gene expression. <i>Nature Genetics</i> , 2017 , 49, 692-699	36.3	182

93	Human genomics. Effect of predicted protein-truncating genetic variants on the human transcriptome. <i>Science</i> , 2015 , 348, 666-9	33.3	170
92	The origin, evolution, and functional impact of short insertion-deletion variants identified in 179 human genomes. <i>Genome Research</i> , 2013 , 23, 749-61	9.7	150
91	The impact of rare variation on gene expression across tissues. <i>Nature</i> , 2017 , 550, 239-243	50.4	146
90	Tissue-specific effects of genetic and epigenetic variation on gene regulation and splicing. <i>PLoS Genetics</i> , 2015 , 11, e1004958	6	140
89	The landscape of genomic imprinting across diverse adult human tissues. <i>Genome Research</i> , 2015 , 25, 927-36	9.7	139
88	Genetic conflict reflected in tissue-specific maps of genomic imprinting in human and mouse. <i>Nature Genetics</i> , 2015 , 47, 544-9	36.3	129
87	Long-read genome sequencing identifies causal structural variation in a Mendelian disease. <i>Genetics in Medicine</i> , 2018 , 20, 159-163	8.1	127
86	From expression QTLs to personalized transcriptomics. <i>Nature Reviews Genetics</i> , 2011 , 12, 277-82	30.1	122
85	Identification of rare-disease genes using blood transcriptome sequencing and large control cohorts. <i>Nature Medicine</i> , 2019 , 25, 911-919	50.5	116
84	The impact of sex on gene expression across human tissues. <i>Science</i> , 2020 , 369,	33.3	100
83	Molecular Choreography of Acute Exercise. <i>Cell</i> , 2020 , 181, 1112-1130.e16	56.2	96
82	Cis and trans effects of human genomic variants on gene expression. <i>PLoS Genetics</i> , 2014 , 10, e1004461	6	92
81	ORegAnno 3.0: a community-driven resource for curated regulatory annotation. <i>Nucleic Acids Research</i> , 2016 , 44, D126-32	20.1	89
80	Rare and common regulatory variation in population-scale sequenced human genomes. <i>PLoS Genetics</i> , 2011 , 7, e1002144	6	82
79	Allele-specific expression reveals interactions between genetic variation and environment. <i>Nature Methods</i> , 2017 , 14, 699-702	21.6	81
78	The GTEx Consortium atlas of genetic regulatory effects across human tissues		81
77	Abundant associations with gene expression complicate GWAS follow-up. <i>Nature Genetics</i> , 2019 , 51, 768-769	36.3	77
76	Uganda Genome Resource Enables Insights into Population History and Genomic Discovery in Africa. <i>Cell</i> , 2019 , 179, 984-1002.e36	56.2	76

75	Systematic functional regulatory assessment of disease-associated variants. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2013 , 110, 9607-12	11.5	75
74	Integrative functional genomics identifies regulatory mechanisms at coronary artery disease loci. <i>Nature Communications</i> , 2016 , 7, 12092	17.4	70
73	Quantifying RNA allelic ratios by microfluidic multiplex PCR and sequencing. <i>Nature Methods</i> , 2014 , 11, 51-4	21.6	70
72	Sex-biased genetic effects on gene regulation in humans. <i>Genome Research</i> , 2012 , 22, 2368-75	9.7	68
71	Impact of the X Chromosome and sex on regulatory variation. <i>Genome Research</i> , 2016 , 26, 768-77	9.7	59
70	Single-cell epigenomic analyses implicate candidate causal variants at inherited risk loci for Alzheimer's and Parkinson's diseases. <i>Nature Genetics</i> , 2020 , 52, 1158-1168	36.3	58
69	Identification of cis- and trans-regulatory variation modulating microRNA expression levels in human fibroblasts. <i>Genome Research</i> , 2011 , 21, 68-73	9.7	57
68	Transcriptome analysis reveals differential splicing events in IPF lung tissue. <i>PLoS ONE</i> , 2014 , 9, e92111	3.7	49
67	An Efficient Multiple-Testing Adjustment for eQTL Studies that Accounts for Linkage Disequilibrium between Variants. <i>American Journal of Human Genetics</i> , 2016 , 98, 216-24	11	48
66	Transcriptome sequencing of a large human family identifies the impact of rare noncoding variants. <i>American Journal of Human Genetics</i> , 2014 , 95, 245-56	11	48
65	A Bioinformatic Analysis of Integrative Mobile Genetic Elements Highlights Their Role in Bacterial Adaptation. <i>Cell Host and Microbe</i> , 2020 , 27, 140-153.e9	23.4	47
64	Integrating GWAS and expression data for functional characterization of disease-associated SNPs: an application to follicular lymphoma. <i>American Journal of Human Genetics</i> , 2013 , 92, 126-30	11	45
63	Biallelic Mutations in ATP5F1D, which Encodes a Subunit of ATP Synthase, Cause a Metabolic Disorder. <i>American Journal of Human Genetics</i> , 2018 , 102, 494-504	11	44
62	Allelic expression of deleterious protein-coding variants across human tissues. <i>PLoS Genetics</i> , 2014 , 10, e1004304	6	43
61	DNA Methylation Profiling of Uniparental Disomy Subjects Provides a Map of Parental Epigenetic Bias in the Human Genome. <i>American Journal of Human Genetics</i> , 2016 , 99, 555-566	11	41
60	Genetic Regulatory Mechanisms of Smooth Muscle Cells Map to Coronary Artery Disease Risk Loci. <i>American Journal of Human Genetics</i> , 2018 , 103, 377-388	11	41
59	Exploiting the GTEx resources to decipher the mechanisms at GWAS loci. <i>Genome Biology</i> , 2021 , 22, 49	18.3	38
58	Transcriptomic signatures across human tissues identify functional rare genetic variation. <i>Science</i> , 2020 , 369,	33.3	36

57	Transcriptome sequencing from diverse human populations reveals differentiated regulatory architecture. <i>PLoS Genetics</i> , 2014 , 10, e1004549	6	35
56	High-resolution transcriptome analysis with long-read RNA sequencing. <i>PLoS ONE</i> , 2014 , 9, e108095	3.7	33
55	Ubiquitination of ABCE1 by NOT4 in Response to Mitochondrial Damage Links Co-translational Quality Control to PINK1-Directed Mitophagy. <i>Cell Metabolism</i> , 2018 , 28, 130-144.e7	24.6	31
54	Large-Scale Phenome-Wide Association Study of Variants Demonstrates Protection Against Ischemic Stroke. <i>Circulation Genomic and Precision Medicine</i> , 2018 , 11, e002162	5.2	27
53	Diagnosing rare diseases after the exome. <i>Journal of Physical Education and Sports Management</i> , 2018 , 4,	2.8	26
52	Population- and individual-specific regulatory variation in Sardinia. <i>Nature Genetics</i> , 2017 , 49, 700-707	36.3	24
51	Whole-transcriptome sequencing in blood provides a diagnosis of spinal muscular atrophy with progressive myoclonic epilepsy. <i>Human Mutation</i> , 2017 , 38, 611-614	4.7	22
50	Properties of structural variants and short tandem repeats associated with gene expression and complex traits. <i>Nature Communications</i> , 2020 , 11, 2927	17.4	22
49	Local genetic effects on gene expression across 44 human tissues		22
48	Exploiting the GTEx resources to decipher the mechanisms at GWAS loci		21
47	A TNFRSF14-FcεRI-mast cell pathway contributes to development of multiple features of asthma pathology in mice. <i>Nature Communications</i> , 2016 , 7, 13696	17.4	21
46	Identification of 22 novel loci associated with urinary biomarkers of albumin, sodium, and potassium excretion. <i>Kidney International</i> , 2019 , 95, 1197-1208	9.9	20
45	Population-scale tissue transcriptomics maps long non-coding RNAs to complex disease. <i>Cell</i> , 2021 , 184, 2633-2648.e19	56.2	20
44	Impact of admixture and ancestry on eQTL analysis and GWAS colocalization in GTEx. <i>Genome Biology</i> , 2020 , 21, 233	18.3	19
43	Proficiency Testing of Standardized Samples Shows Very High Interlaboratory Agreement for Clinical Next-Generation Sequencing-Based Oncology Assays. <i>Archives of Pathology and Laboratory Medicine</i> , 2019 , 143, 463-471	5	19
42	Genetic regulation of gene expression and splicing during a 10-year period of human aging. <i>Genome Biology</i> , 2019 , 20, 230	18.3	18
41	FAM13A affects body fat distribution and adipocyte function. <i>Nature Communications</i> , 2020 , 11, 1465	17.4	17
40	Transcriptome analysis reveals differential splicing events in IPF lung tissue. <i>PLoS ONE</i> , 2014 , 9, e97550	3.7	17

39	Identification of rare and common regulatory variants in pluripotent cells using population-scale transcriptomics. <i>Nature Genetics</i> , 2021 , 53, 313-321	36.3	16
38	Recurrently Mutated Genes Differ between Leptomeningeal and Solid Lung Cancer Brain Metastases. <i>Journal of Thoracic Oncology</i> , 2018 , 13, 1022-1027	8.9	15
37	Functional regulatory mechanism of smooth muscle cell-restricted LMOD1 coronary artery disease locus. <i>PLoS Genetics</i> , 2018 , 14, e1007755	6	15
36	Incorporation of Biological Knowledge Into the Study of Gene-Environment Interactions. <i>American Journal of Epidemiology</i> , 2017 , 186, 771-777	3.8	13
35	Diverse transcriptomic signatures across human tissues identify functional rare genetic variation		13
34	PML nuclear bodies contribute to the basal expression of the mTOR inhibitor DDIT4. <i>Scientific Reports</i> , 2017 , 7, 45038	4.9	12
33	Pathologic gene network rewiring implicates PPP1R3A as a central regulator in pressure overload heart failure. <i>Nature Communications</i> , 2019 , 10, 2760	17.4	11
32	Discovery and quality analysis of a comprehensive set of structural variants and short tandem repeats. <i>Nature Communications</i> , 2020 , 11, 2928	17.4	11
31	Genetic analyses of human fetal retinal pigment epithelium gene expression suggest ocular disease mechanisms. <i>Communications Biology</i> , 2019 , 2, 186	6.7	10
30	Dissecting the causal genetic mechanisms of coronary heart disease. <i>Current Atherosclerosis Reports</i> , 2014 , 16, 406	6	10
29	Small RNA Sequencing in Cells and Exosomes Identifies eQTLs and 14q32 as a Region of Active Export. <i>G3: Genes, Genomes, Genetics</i> , 2017 , 7, 31-39	3.2	10
28	The impact of rare variation on gene expression across tissues		9
27	Single-cell epigenomic identification of inherited risk loci in Alzheimer's and Parkinson's disease		9
26	Systematic assessment of regulatory effects of human disease variants in pluripotent cells		9
25	Identifying causal variants and genes using functional genomics in specialized cell types and contexts. <i>Human Genetics</i> , 2020 , 139, 95-102	6.3	9
24	Non-Coding Loss-of-Function Variation in Human Genomes. <i>Human Heredity</i> , 2016 , 81, 78-87	1.1	8
23	Genome-wide functional screen of 3'UTR variants uncovers causal variants for human disease and evolution. <i>Cell</i> , 2021 , 184, 5247-5260.e19	56.2	7
22	Genotype-based test in mapping cis-regulatory variants from allele-specific expression data. <i>PLoS ONE</i> , 2012 , 7, e38667	3.7	6

21	Cohort-specific imputation of gene expression improves prediction of warfarin dose for African Americans. <i>Genome Medicine</i> , 2017 , 9, 98	14.4	5
20	Nonsense-mediated decay is highly stable across individuals and tissues. <i>American Journal of Human Genetics</i> , 2021 , 108, 1401-1408	11	5
19	Functional and structural analysis of cytokine-selective IL6ST defects that cause recessive hyper-IgE syndrome. <i>Journal of Allergy and Clinical Immunology</i> , 2021 , 148, 585-598	11.5	5
18	Identification of rare-disease genes in diverse undiagnosed cases using whole blood transcriptome sequencing and large control cohorts		4
17	Allele-specific expression reveals interactions between genetic variation and environment		3
16	Genetic dysregulation of gene expression and splicing during a ten-year period of human aging		3
15	Evaluating the Genomic Parameters Governing rAAV-Mediated Homologous Recombination. <i>Molecular Therapy</i> , 2021 , 29, 1028-1046	11.7	3
14	Identification of putative causal loci in whole-genome sequencing data via knockoff statistics. <i>Nature Communications</i> , 2021 , 12, 3152	17.4	3
13	An integrated approach to identify environmental modulators of genetic risk factors for complex traits. <i>American Journal of Human Genetics</i> , 2021 , 108, 1866-1879	11	3
12	Population-specific imputation of gene expression improves prediction of pharmacogenomic traits for African Americans		2
11	Ocular disease mechanisms elucidated by genetics of human fetal retinal pigment epithelium gene expression		2
10	The impact of structural variation on human gene expression		2
9	The role of Sp140 revealed in IgE and mast cell responses in Collaborative Cross mice. <i>JCI Insight</i> , 2021 , 6,	9.9	2
8	Genome-wide functional screen of 3UTR variants uncovers causal variants for human disease and evolution		2
7	Lymphoid blast transformation in an MPN with BCR-JAK2 treated with ruxolitinib: putative mechanisms of resistance. <i>Blood Advances</i> , 2021 , 5, 3492-3496	7.8	2
6	Multiple causal variants underlie genetic associations in humans.. <i>Science</i> , 2022 , 375, 1247-1254	33.3	2
5	PATH-SCAN: A REPORTING TOOL FOR IDENTIFYING CLINICALLY ACTIONABLE VARIANTS 2013 ,		1
4	Integration of rare large-effect expression variants improves polygenic risk prediction		1

3	Compound heterozygous variants in progressive myoclonus epilepsy. <i>Journal of Neurogenetics</i> , 2021 , 35, 74-83	1.6	1
2	Nonsense-mediated decay is highly stable across individuals and tissues		1
1	Integration of genetic colocalizations with physiological and pharmacological perturbations identifies cardiometabolic disease genes.. <i>Genome Medicine</i> , 2022 , 14, 31	14.4	0