Stephen B Montgomery

List of Publications by Year
in descending order

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97 papers	22,226 citations	41258 49 h-index	38300 95 g-index
116 all docs	116 docs citations	116 times ranked	36840 citing authors

#	Article	IF	Citations
1	Genetic effects on gene expression across human tissues. Nature, 2017, 550, 204-213.	13.7	3,500
2	Integrating common and rare genetic variation in diverse human populations. Nature, 2010, 467, 52-58.	13.7	2,625
3	Transcriptome and genome sequencing uncovers functional variation in humans. Nature, 2013, 501, 506-511.	13.7	1,857
4	A Systematic Survey of Loss-of-Function Variants in Human Protein-Coding Genes. Science, 2012, 335, 823-828.	6.0	1,095
5	Population genomics of human gene expression. Nature Genetics, 2007, 39, 1217-1224.	9.4	1,072
6	Transcriptome genetics using second generation sequencing in a Caucasian population. Nature, 2010, 464, 773-777.	13.7	782
7	Common Regulatory Variation Impacts Gene Expression in a Cell Typeâ€“Dependent Manner. Science, 2009, 325, 1246-1250.	6.0	694
8	Characterizing the genetic basis of transcriptome diversity through RNA-sequencing of 922 individuals. Genome Research, 2014, 24, 14-24.	2.4	547
9	RNA Sequencing and Analysis. Cold Spring Harbor Protocols, 2015, 2015, pdb.top084970.	0.2	538
10	Atheroprotective roles of smooth muscle cell phenotypic modulation and the TCF21 disease gene as revealed by single-cell analysis. Nature Medicine, 2019, 25, 1280-1289.	15.2	494
11	Patterns of Cis Regulatory Variation in Diverse Human Populations. PLoS Genetics, 2012, 8, e1002639.	1.5	439
12	Candidate Causal Regulatory Effects by Integration of Expression QTLs with Complex Trait Genetic Associations. PLoS Genetics, 2010, 6, e1000895.	1.5	434
13	Directed evolution using dCas9-targeted somatic hypermutation in mammalian cells. Nature Methods, 2016, 13, 1036-1042.	9.0	378
14	Passive and active DNA methylation and the interplay with genetic variation in gene regulation. ELife, 2013, 2, e00523.	2.8	374
15	The impact of structural variation on human gene expression. Nature Genetics, 2017, 49, 692-699.	9.4	334
16	The impact of sex on gene expression across human tissues. Science, 2020, 369, .	6.0	329
17	Overexpression of the Cytokine BAFF and Autoimmunity Risk. New England Journal of Medicine, 2017, 376, 1615-1626.	13.9	301
18	Molecular Choreography of Acute Exercise. Cell, 2020, 181, 1112-1130.e16.	13.5	261

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19	Effect of predicted protein-truncating genetic variants on the human transcriptome. Science, 2015, 348, 666-669.	6.0	252
20	The impact of rare variation on gene expression across tissues. Nature, 2017, 550, 239-243.	13.7	229
21	ORegAnno: an open-access community-driven resource for regulatory annotation. Nucleic Acids Research, 2007, 36, D107-D113.	6.5	227
22	Genetic conflict reflected in tissue-specific maps of genomic imprinting in human and mouse. Nature Genetics, 2015, 47, 544-549.	9.4	221
23	Identification of rare-disease genes using blood transcriptome sequencing and large control cohorts. Nature Medicine, 2019, 25, 911-919.	15.2	221
24	Single-cell epigenomic analyses implicate candidate causal variants at inherited risk loci for Alzheimerâ€™s and Parkinsonâ€™s diseases. Nature Genetics, 2020, 52, 1158-1168.	9.4	217
25	The landscape of genomic imprinting across diverse adult human tissues. Genome Research, 2015, 25, 927-936.	2.4	216
26	Abundant associations with gene expression complicate GWAS follow-up. Nature Genetics, 2019, 51, 768-769.	9.4	210
27	The origin, evolution, and functional impact of short insertionâ€“deletion variants identified in 179 human genomes. Genome Research, 2013, 23, 749-761.	2.4	206
28	Long-read genome sequencing identifies causal structural variation in a Mendelian disease. Genetics in Medicine, 2018, 20, 159-163.	1.1	189
29	Tissue-Specific Effects of Genetic and Epigenetic Variation on Gene Regulation and Splicing. PLoS Genetics, 2015, 11, e1004958.	1.5	185
30	Uganda Genome Resource Enables Insights into Population History and Genomic Discovery in Africa. Cell, 2019, 179, 984-1002.e36.	13.5	152
31	Exploiting the GTEx resources to decipher the mechanisms at GWAS loci. Genome Biology, 2021, 22, 49.	3.8	150
32	From expression QTLs to personalized transcriptomics. Nature Reviews Genetics, 2011, 12, 277-282.	7.7	148
33	ORegAnno 3.0: a community-driven resource for curated regulatory annotation. Nucleic Acids Research, 2016, 44, D126-D132.	6.5	142
34	Allele-specific expression reveals interactions between genetic variation and environment. Nature Methods, 2017, 14, 699-702.	9.0	135
35	Integrative functional genomics identifies regulatory mechanisms at coronary artery disease loci. Nature Communications, 2016, 7, 12092.	5.8	123
36	A Bioinformatic Analysis of Integrative Mobile Genetic Elements Highlights Their Role in Bacterial Adaptation. Cell Host and Microbe, 2020, 27, 140-153.e9.	5.1	119

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37	Cis and Trans Effects of Human Genomic Variants on Gene Expression. PLoS Genetics, 2014, 10, e1004461.	1.5	117
38	Rare and Common Regulatory Variation in Population-Scale Sequenced Human Genomes. PLoS Genetics, 2011, 7, e1002144.	1.5	98
39	Population-scale tissue transcriptomics maps long non-coding RNAs to complex disease. Cell, 2021, 184, 2633-2648.e19.	13.5	94
40	Sex-biased genetic effects on gene regulation in humans. Genome Research, 2012, 22, 2368-2375.	2.4	92
41	An Efficient Multiple-Testing Adjustment for eQTL Studies that Accounts for Linkage Disequilibrium between Variants. American Journal of Human Genetics, 2016, 98, 216-224.	2.6	91
42	Transcriptomic signatures across human tissues identify functional rare genetic variation. Science, 2020, 369, .	6.0	89
43	Impact of the X Chromosome and sex on regulatory variation. Genome Research, 2016, 26, 768-777.	2.4	88
44	Systematic functional regulatory assessment of disease-associated variants. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 9607-9612.	3.3	85
45	Quantifying RNA allelic ratios by microfluidic multiplex PCR and sequencing. Nature Methods, 2014, 11, 51-54.	9.0	81
46	Genetic Regulatory Mechanisms of Smooth Muscle Cells Map to Coronary Artery Disease Risk Loci. American Journal of Human Genetics, 2018, 103, 377-388.	2.6	76
47	Multiple causal variants underlie genetic associations in humans. Science, 2022, 375, 1247-1254.	6.0	75
48	Transcriptome Analysis Reveals Differential Splicing Events in IPF Lung Tissue. PLoS ONE, 2014, 9, e92111.	1.1	73
49	Identification of cis- and trans-regulatory variation modulating microRNA expression levels in human fibroblasts. Genome Research, 2011, 21, 68-73.	2.4	70
50	Properties of structural variants and short tandem repeats associated with gene expression and complex traits. Nature Communications, 2020, 11, 2927.	5.8	67
51	DNA Methylation Profiling of Uniparental Disomy Subjects Provides a Map of Parental Epigenetic Bias in the Human Genome. American Journal of Human Genetics, 2016, 99, 555-566.	2.6	66
52	Impact of admixture and ancestry on eQTL analysis and GWAS colocalization in GTEx. Genome Biology, 2020, 21, 233.	3.8	64
53	Transcriptome Sequencing of a Large Human Family Identifies the Impact of Rare Noncoding Variants. American Journal of Human Genetics, 2014, 95, 245-256.	2.6	63
54	Genome-wide functional screen of 3â€²UTR variants uncovers causal variants for human disease and evolution. Cell, 2021, 184, 5247-5260.e19.	13.5	62

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55	Ubiquitination of ABCE1 by NOT4 in Response to Mitochondrial Damage Links Co-translational Quality Control to PINK1-Directed Mitophagy. Cell Metabolism, 2018, 28, 130-144.e7.	7.2	61
56	Allelic Expression of Deleterious Protein-Coding Variants across Human Tissues. PLoS Genetics, 2014, 10, e1004304.	1.5	60
57	Biallelic Mutations in ATP5F1D, which Encodes a Subunit of ATP Synthase, Cause a Metabolic Disorder. American Journal of Human Genetics, 2018, 102, 494-504.	2.6	59
58	Genetic regulation of gene expression and splicing during a 10-year period of human aging. Genome Biology, 2019, 20, 230.	3.8	57
59	Integrating GWAS and Expression Data for Functional Characterization of Disease-Associated SNPs: An Application to Follicular Lymphoma. American Journal of Human Genetics, 2013, 92, 126-130.	2.6	49
60	Transcriptome Sequencing from Diverse Human Populations Reveals Differentiated Regulatory Architecture. PLoS Genetics, 2014, 10, e1004549.	1.5	49
61	Diagnosing rare diseases after the exome. Journal of Physical Education and Sports Management, 2018, 4, a003392.	0.5	48
62	Large-Scale Phenome-Wide Association Study of PCSK9 Variants Demonstrates Protection Against Ischemic Stroke. Circulation Genomic and Precision Medicine, 2018, 11, e002162.	1.6	48
63	High-Resolution Transcriptome Analysis with Long-Read RNA Sequencing. PLoS ONE, 2014, 9, e108095.	1.1	47
64	Identification of rare and common regulatory variants in pluripotent cells using population-scale transcriptomics. Nature Genetics, 2021, 53, 313-321.	9.4	42
65	Population- and individual-specific regulatory variation in Sardinia. Nature Genetics, 2017, 49, 700-707.	9.4	38
66	A TNFRSF14-FcÉ›RI-mast cell pathway contributes to development of multiple features of asthma pathology in mice. Nature Communications, 2016, 7, 13696.	5.8	36
67	FAM13A affects body fat distribution and adipocyte function. Nature Communications, 2020, 11, 1465.	5.8	36
68	Identification of 22 novel loci associated withÂurinary biomarkers of albumin, sodium, andÂpotassium excretion. Kidney International, 2019, 95, 1197-1208.	2.6	33
69	Proficiency Testing of Standardized Samples Shows Very High Interlaboratory Agreement for Clinical Next-Generation Sequencingâ€“Based Oncology Assays. Archives of Pathology and Laboratory Medicine, 2019, 143, 463-471.	1.2	32
70	Functional regulatory mechanism of smooth muscle cell-restricted LMOD1 coronary artery disease locus. PLoS Genetics, 2018, 14, e1007755.	1.5	30
71	Whole-transcriptome sequencing in blood provides a diagnosis of spinal muscular atrophy with progressive myoclonic epilepsy. Human Mutation, 2017, 38, 611-614.	1.1	25
72	Incorporation of Biological Knowledge Into the Study of Gene-Environment Interactions. American Journal of Epidemiology, 2017, 186, 771-777.	1.6	23

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73	Pathologic gene network rewiring implicates PPP1R3A as a central regulator in pressure overload heart failure. Nature Communications, 2019, 10, 2760.	5.8	22
74	Discovery and quality analysis of a comprehensive set of structural variants and short tandem repeats. Nature Communications, 2020, 11, 2928.	5.8	22
75	Recurrently Mutated Genes Differ between Leptomeningeal and Solid Lung Cancer Brain Metastases. Journal of Thoracic Oncology, 2018, 13, 1022-1027.	0.5	20
76	Genetic analyses of human fetal retinal pigment epithelium gene expression suggest ocular disease mechanisms. Communications Biology, 2019, 2, 186.	2.0	20
77	Functional and structural analysis of cytokine-selective IL6ST defects that cause recessive hyper-IgE syndrome. Journal of Allergy and Clinical Immunology, 2021, 148, 585-598.	1.5	20
78	Correction: Transcriptome Analysis Reveals Differential Splicing Events in IPF Lung Tissue. PLoS ONE, 2014, 9, e97550.	1.1	20
79	Identification of putative causal loci in whole-genome sequencing data via knockoff statistics. Nature Communications, 2021, 12, 3152.	5.8	17
80	Non-Coding Loss-of-Function Variation in Human Genomes. Human Heredity, 2016, 81, 78-87.	0.4	16
81	Small RNA Sequencing in Cells and Exosomes Identifies eQTLs and 14q32 as a Region of Active Export. G3: Genes, Genomes, Genetics, 2017, 7, 31-39.	0.8	16
82	Identifying causal variants and genes using functional genomics in specialized cell types and contexts. Human Genetics, 2020, 139, 95-102.	1.8	16
83	PML nuclear bodies contribute to the basal expression of the mTOR inhibitor DDIT4. Scientific Reports, 2017, 7, 45038.	1.6	15
84	Nonsense-mediated decay is highly stable across individuals and tissues. American Journal of Human Genetics, 2021, 108, 1401-1408.	2.6	15
85	Lymphoid blast transformation in an MPN with BCR-JAK2 treated with ruxolitinib: putative mechanisms of resistance. Blood Advances, 2021, 5, 3492-3496.	2.5	14
86	Dissecting the Causal Genetic Mechanisms of Coronary Heart Disease. Current Atherosclerosis Reports, 2014, 16, 406.	2.0	11
87	Cohort-specific imputation of gene expression improves prediction of warfarin dose for African Americans. Genome Medicine, 2017, 9, 98.	3.6	11
88	PATH-SCAN: A REPORTING TOOL FOR IDENTIFYING CLINICALLY ACTIONABLE VARIANTS. , 2013, , .		9
89	An integrated approach to identify environmental modulators of genetic risk factors for complex traits. American Journal of Human Genetics, 2021, 108, 1866-1879.	2.6	9
90	TOWARDS TRANSCRIPTOMICS AS A PRIMARY TOOL FOR RARE DISEASE INVESTIGATION. Journal of Physical Education and Sports Management, 2022, , mcs.a006198.	0.5	9

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91	The role of Sp140 revealed in IgE and mast cell responses in Collaborative Cross mice. JCI Insight, 2021, 6, .	2.3	8
92	Integration of rare expression outlier-associated variants improves polygenic risk prediction. American Journal of Human Genetics, 2022, 109, 1055-1064.	2.6	8
93	Genotype-Based Test in Mapping Cis-Regulatory Variants from Allele-Specific Expression Data. PLoS ONE, 2012, 7, e38667.	1.1	7
94	Integration of genetic colocalizations with physiological and pharmacological perturbations identifies cardiometabolic disease genes. Genome Medicine, 2022, 14, 31.	3.6	7
95	Evaluating the Genomic Parameters Governing rAAV-Mediated Homologous Recombination. Molecular Therapy, 2021, 29, 1028-1046.	3.7	6
96	Compound heterozygous KCTD7 variants in progressive myoclonus epilepsy. Journal of Neurogenetics, 2021, 35, 74-83.	0.6	4
97	Abstract 62: Molecular Basis of Regulatory Variation at Coronary Heart Disease-Associated Loci. Arteriosclerosis, Thrombosis, and Vascular Biology, 2015, 35, .	1.1	0