Stephen B Montgomery

List of Publications by Year in Descending Order

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	Integration of genetic colocalizations with physiological and pharmacological perturbations identifies cardiometabolic disease genes.. *Genome Medicine*, **2022**, 14, 31	14.4	0
109	Multiple causal variants underlie genetic associations in humans.. *Science*, **2022**, 375, 1247-1254	33.3	2
108	Identification of rare and common regulatory variants in pluripotent cells using population-scale transcriptomics. *Nature Genetics*, **2021**, 53, 313-321	36.3	16
107	Evaluating the Genomic Parameters Governing rAAV-Mediated Homologous Recombination. *Molecular Therapy*, **2021**, 29, 1028-1046	11.7	3
106	Compound heterozygous variants in progressive myoclonus epilepsy. *Journal of Neurogenetics*, **2021**, 35, 74-83	1.6	1
105	Population-scale tissue transcriptomics maps long non-coding RNAs to complex disease. *Cell*, **2021**, 184, 2633-2648.e19	56.2	20
104	Identification of putative causal loci in whole-genome sequencing data via knockoff statistics. *Nature Communications*, **2021**, 12, 3152	17.4	3
103	The role of Sp140 revealed in IgE and mast cell responses in Collaborative Cross mice. *JCI Insight*, **2021**, 6,	9.9	2
102	Exploiting the GTEx resources to decipher the mechanisms at GWAS loci. *Genome Biology*, **2021**, 22, 49	18.3	38
101	Nonsense-mediated decay is highly stable across individuals and tissues. *American Journal of Human Genetics*, **2021**, 108, 1401-1408	11	5
100	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	11.5	5
99	Lymphoid blast transformation in an MPN with BCR-JAK2 treated with ruxolitinib: putative mechanisms of resistance. *Blood Advances*, **2021**, 5, 3492-3496	7.8	2
98	Genome-wide functional screen of 3'UTR variants uncovers causal variants for human disease and evolution. *Cell*, **2021**, 184, 5247-5260.e19	56.2	7
97	An integrated approach to identify environmental modulators of genetic risk factors for complex traits. *American Journal of Human Genetics*, **2021**, 108, 1866-1879	11	3
96	Molecular Choreography of Acute Exercise. *Cell*, **2020**, 181, 1112-1130.e16	56.2	96
95	Discovery and quality analysis of a comprehensive set of structural variants and short tandem repeats. *Nature Communications*, **2020**, 11, 2928	17.4	11
94	Properties of structural variants and short tandem repeats associated with gene expression and complex traits. *Nature Communications*, **2020**, 11, 2927	17.4	22

93	FAM13A affects body fat distribution and adipocyte function. *Nature Communications*, **2020**, 11, 1465	17.4	17
92	A Bioinformatic Analysis of Integrative Mobile Genetic Elements Highlights Their Role in Bacterial Adaptation. *Cell Host and Microbe*, **2020**, 27, 140-153.e9	23.4	47
91	Single-cell epigenomic analyses implicate candidate causal variants at inherited risk loci for Alzheimer's and Parkinson's diseases. *Nature Genetics*, **2020**, 52, 1158-1168	36.3	58
90	Transcriptomic signatures across human tissues identify functional rare genetic variation. *Science*, **2020**, 369,	33.3	36
89	The impact of sex on gene expression across human tissues. *Science*, **2020**, 369,	33.3	100
88	Impact of admixture and ancestry on eQTL analysis and GWAS colocalization in GTEx. *Genome Biology*, **2020**, 21, 233	18.3	19
87	Identifying causal variants and genes using functional genomics in specialized cell types and contexts. *Human Genetics*, **2020**, 139, 95-102	6.3	9
86	Genetic analyses of human fetal retinal pigment epithelium gene expression suggest ocular disease mechanisms. *Communications Biology*, **2019**, 2, 186	6.7	10
85	Pathologic gene network rewiring implicates PPP1R3A as a central regulator in pressure overload heart failure. *Nature Communications*, **2019**, 10, 2760	17.4	11
84	Identification of rare-disease genes using blood transcriptome sequencing and large control cohorts. *Nature Medicine*, **2019**, 25, 911-919	50.5	116
83	Abundant associations with gene expression complicate GWAS follow-up. *Nature Genetics*, **2019**, 51, 768-769	36.3	77
82	Identification of 22 novel loci associated with urinary biomarkers of albumin, sodium, and potassium excretion. *Kidney International*, **2019**, 95, 1197-1208	9.9	20
81	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	50.5	198
80	Uganda Genome Resource Enables Insights into Population History and Genomic Discovery in Africa. *Cell*, **2019**, 179, 984-1002.e36	56.2	76
79	Genetic regulation of gene expression and splicing during a 10-year period of human aging. *Genome Biology*, **2019**, 20, 230	18.3	18
78	Proficiency Testing of Standardized Samples Shows Very High Interlaboratory Agreement for Clinical Next-Generation Sequencing-Based Oncology Assays. *Archives of Pathology and Laboratory Medicine*, **2019**, 143, 463-471	5	19
77	Biallelic Mutations in ATP5F1D, which Encodes a Subunit of ATP Synthase, Cause a Metabolic Disorder. *American Journal of Human Genetics*, **2018**, 102, 494-504	11	44
76	Recurrently Mutated Genes Differ between Leptomeningeal and Solid Lung Cancer Brain Metastases. *Journal of Thoracic Oncology*, **2018**, 13, 1022-1027	8.9	15

75	Long-read genome sequencing identifies causal structural variation in a Mendelian disease. *Genetics in Medicine*, **2018**, 20, 159-163	8.1	127
74	Genetic Regulatory Mechanisms of Smooth Muscle Cells Map to Coronary Artery Disease Risk Loci. *American Journal of Human Genetics*, **2018**, 103, 377-388	11	41
73	Diagnosing rare diseases after the exome. *Journal of Physical Education and Sports Management*, **2018**, 4,	2.8	26
72	Functional regulatory mechanism of smooth muscle cell-restricted LMOD1 coronary artery disease locus. *PLoS Genetics*, **2018**, 14, e1007755	6	15
71	Large-Scale Phenome-Wide Association Study of Variants Demonstrates Protection Against Ischemic Stroke. *Circulation Genomic and Precision Medicine*, **2018**, 11, e002162	5.2	27
70	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	24.6	31
69	Whole-transcriptome sequencing in blood provides a diagnosis of spinal muscular atrophy with progressive myoclonic epilepsy. *Human Mutation*, **2017**, 38, 611-614	4.7	22
68	Population- and individual-specific regulatory variation in Sardinia. *Nature Genetics*, **2017**, 49, 700-707	36.3	24
67	Overexpression of the Cytokine BAFF and Autoimmunity Risk. *New England Journal of Medicine*, **2017**, 376, 1615-1626	59.2	198
66	Allele-specific expression reveals interactions between genetic variation and environment. *Nature Methods*, **2017**, 14, 699-702	21.6	81
65	The impact of structural variation on human gene expression. *Nature Genetics*, **2017**, 49, 692-699	36.3	182
64	PML nuclear bodies contribute to the basal expression of the mTOR inhibitor DDIT4. *Scientific Reports*, **2017**, 7, 45038	4.9	12
63	The impact of rare variation on gene expression across tissues. *Nature*, **2017**, 550, 239-243	50.4	146
62	Genetic effects on gene expression across human tissues. *Nature*, **2017**, 550, 204-213	50.4	2086
61	Cohort-specific imputation of gene expression improves prediction of warfarin dose for African Americans. *Genome Medicine*, **2017**, 9, 98	14.4	5
60	Incorporation of Biological Knowledge Into the Study of Gene-Environment Interactions. *American Journal of Epidemiology*, **2017**, 186, 771-777	3.8	13
59	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	3.2	10
58	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	11	41

57	Integrative functional genomics identifies regulatory mechanisms at coronary artery disease loci. *Nature Communications*, **2016**, 7, 12092	17.4	70
56	Directed evolution using dCas9-targeted somatic hypermutation in mammalian cells. *Nature Methods*, **2016**, 13, 1036-1042	21.6	288
55	ORegAnno 3.0: a community-driven resource for curated regulatory annotation. *Nucleic Acids Research*, **2016**, 44, D126-32	20.1	89
54	An Efficient Multiple-Testing Adjustment for eQTL Studies that Accounts for Linkage Disequilibrium between Variants. *American Journal of Human Genetics*, **2016**, 98, 216-24	11	48
53	Impact of the X Chromosome and sex on regulatory variation. *Genome Research*, **2016**, 26, 768-77	9.7	59
52	A TNFRSF14-FceRI-mast cell pathway contributes to development of multiple features of asthma pathology in mice. *Nature Communications*, **2016**, 7, 13696	17.4	21
51	Non-Coding Loss-of-Function Variation in Human Genomes. *Human Heredity*, **2016**, 81, 78-87	1.1	8
50	The landscape of genomic imprinting across diverse adult human tissues. *Genome Research*, **2015**, 25, 927-36	9.7	139
49	Tissue-specific effects of genetic and epigenetic variation on gene regulation and splicing. *PLoS Genetics*, **2015**, 11, e1004958	6	140
48	Genetic conflict reflected in tissue-specific maps of genomic imprinting in human and mouse. *Nature Genetics*, **2015**, 47, 544-9	36.3	129
47	Human genomics. Effect of predicted protein-truncating genetic variants on the human transcriptome. *Science*, **2015**, 348, 666-9	33.3	170
46	RNA Sequencing and Analysis. *Cold Spring Harbor Protocols*, **2015**, 2015, 951-69	1.2	293
45	Dissecting the causal genetic mechanisms of coronary heart disease. *Current Atherosclerosis Reports* , **2014**, 16, 406	6	10
44	Characterizing the genetic basis of transcriptome diversity through RNA-sequencing of 922 individuals. *Genome Research*, **2014**, 24, 14-24	9.7	371
43	Quantifying RNA allelic ratios by microfluidic multiplex PCR and sequencing. *Nature Methods*, **2014**, 11, 51-4	21.6	70
42	High-resolution transcriptome analysis with long-read RNA sequencing. *PLoS ONE*, **2014**, 9, e108095	3.7	33
41	Cis and trans effects of human genomic variants on gene expression. *PLoS Genetics*, **2014**, 10, e1004461	6	92
40	Transcriptome sequencing from diverse human populations reveals differentiated regulatory architecture. *PLoS Genetics*, **2014**, 10, e1004549	6	35

39	Allelic expression of deleterious protein-coding variants across human tissues. *PLoS Genetics*, **2014**, 10, e1004304	6	43
38	Transcriptome sequencing of a large human family identifies the impact of rare noncoding variants. *American Journal of Human Genetics*, **2014**, 95, 245-56	11	48
37	Transcriptome analysis reveals differential splicing events in IPF lung tissue. *PLoS ONE*, **2014**, 9, e92111	3.7	49
36	Transcriptome analysis reveals differential splicing events in IPF lung tissue. *PLoS ONE*, **2014**, 9, e97550	3.7	17
35	Transcriptome and genome sequencing uncovers functional variation in humans. *Nature*, **2013**, 501, 506-11	50.4	1323
34	Integrating GWAS and expression data for functional characterization of disease-associated SNPs: an application to follicular lymphoma. *American Journal of Human Genetics*, **2013**, 92, 126-30	11	45
33	The origin, evolution, and functional impact of short insertion-deletion variants identified in 179 human genomes. *Genome Research*, **2013**, 23, 749-61	9.7	150
32	PATH-SCAN: A REPORTING TOOL FOR IDENTIFYING CLINICALLY ACTIONABLE VARIANTS **2013**,		1
31	Systematic functional regulatory assessment of disease-associated variants. *Proceedings of the National Academy of Sciences of the United States of America*, **2013**, 110, 9607-12	11.5	75
30	Passive and active DNA methylation and the interplay with genetic variation in gene regulation. *ELife*, **2013**, 2, e00523	8.9	295
29	A systematic survey of loss-of-function variants in human protein-coding genes. *Science*, **2012**, 335, 823-8	33.3	880
28	Patterns of cis regulatory variation in diverse human populations. *PLoS Genetics*, **2012**, 8, e1002639	6	361
27	Sex-biased genetic effects on gene regulation in humans. *Genome Research*, **2012**, 22, 2368-75	9.7	68
26	Genotype-based test in mapping cis-regulatory variants from allele-specific expression data. *PLoS ONE*, **2012**, 7, e38667	3.7	6
25	From expression QTLs to personalized transcriptomics. *Nature Reviews Genetics*, **2011**, 12, 277-82	30.1	122
24	Identification of cis- and trans-regulatory variation modulating microRNA expression levels in human fibroblasts. *Genome Research*, **2011**, 21, 68-73	9.7	57
23	Rare and common regulatory variation in population-scale sequenced human genomes. *PLoS Genetics*, **2011**, 7, e1002144	6	82
22	Transcriptome genetics using second generation sequencing in a Caucasian population. *Nature*, **2010**, 464, 773-7	50.4	678

21	Integrating common and rare genetic variation in diverse human populations. *Nature*, **2010**, 467, 52-8	50.4	2135
20	Candidate causal regulatory effects by integration of expression QTLs with complex trait genetic associations. *PLoS Genetics*, **2010**, 6, e1000895	6	339
19	Common regulatory variation impacts gene expression in a cell type-dependent manner. *Science*, **2009**, 325, 1246-50	33.3	607
18	ORegAnno: an open-access community-driven resource for regulatory annotation. *Nucleic Acids Research*, **2008**, 36, D107-13	20.1	199
17	Population genomics of human gene expression. *Nature Genetics*, **2007**, 39, 1217-24	36.3	936
16	Allele-specific expression reveals interactions between genetic variation and environment		3
15	The impact of rare variation on gene expression across tissues		9
14	Local genetic effects on gene expression across 44 human tissues		22
13	Population-specific imputation of gene expression improves prediction of pharmacogenomic traits for African Americans		2
12	Single-cell epigenomic identification of inherited risk loci in Alzheimer and Parkinson disease		9
11	Integration of rare large-effect expression variants improves polygenic risk prediction		1
10	Identification of rare-disease genes in diverse undiagnosed cases using whole blood transcriptome sequencing and large control cohorts		4
9	Ocular disease mechanisms elucidated by genetics of human fetal retinal pigment epithelium gene expression		2
8	Genetic dysregulation of gene expression and splicing during a ten-year period of human aging		3
7	Systematic assessment of regulatory effects of human disease variants in pluripotent cells		9
6	Diverse transcriptomic signatures across human tissues identify functional rare genetic variation		13
5	The GTEx Consortium atlas of genetic regulatory effects across human tissues		81
4	Exploiting the GTEx resources to decipher the mechanisms at GWAS loci		21

3	The impact of structural variation on human gene expression	2
2	Genome-wide functional screen of 3′UTR variants uncovers causal variants for human disease and evolution	2
1	Nonsense-mediated decay is highly stable across individuals and tissues	1