Oliver Stegle

List of Publications by Citations

176 papers	29,431 citations	63 h-index	171 g-index
209 ext. papers	41,593 ext. citations	20 avg, IF	7.68 L-index

#	Paper	IF	Citations
176	A global reference for human genetic variation. *Nature*, **2015**, 526, 68-74	50.4	8599
175	An integrated map of structural variation in 2,504 human genomes. *Nature*, **2015**, 526, 75-81	50.4	1368
174	Transcriptome and genome sequencing uncovers functional variation in humans. *Nature*, **2013**, 501, 506-11	50.4	1323
173	The Human Cell Atlas. *ELife*, **2017**, 6,	8.9	937
172	Pan-cancer analysis of whole genomes. *Nature*, **2020**, 578, 82-93	50.4	840
171	Computational analysis of cell-to-cell heterogeneity in single-cell RNA-sequencing data reveals hidden subpopulations of cells. *Nature Biotechnology*, **2015**, 33, 155-60	44.5	778
170	Whole-genome sequencing of multiple Arabidopsis thaliana populations. *Nature Genetics*, **2011**, 43, 956-63	30.3	737
169	Computational and analytical challenges in single-cell transcriptomics. *Nature Reviews Genetics*, **2015**, 16, 133-45	30.1	736
168	Deep learning for computational biology. *Molecular Systems Biology*, **2016**, 12, 878	12.2	733
167	Single-cell genome-wide bisulfite sequencing for assessing epigenetic heterogeneity. *Nature Methods*, **2014**, 11, 817-820	21.6	673
166	A comprehensive assessment of RNA-seq accuracy, reproducibility and information content by the Sequencing Quality Control Consortium. *Nature Biotechnology*, **2014**, 32, 903-14	44.5	618
165	Lineage-Specific Genome Architecture Links Enhancers and Non-coding Disease Variants to Target Gene Promoters. *Cell*, **2016**, 167, 1369-1384.e19	56.2	556
164	Spontaneous epigenetic variation in the Arabidopsis thaliana methylome. *Nature*, **2011**, 480, 245-9	50.4	533
163	Multiple reference genomes and transcriptomes for Arabidopsis thaliana. *Nature*, **2011**, 477, 419-23	50.4	495
162	Using probabilistic estimation of expression residuals (PEER) to obtain increased power and interpretability of gene expression analyses. *Nature Protocols*, **2012**, 7, 500-7	18.8	460
161	Parallel single-cell sequencing links transcriptional and epigenetic heterogeneity. *Nature Methods*, **2016**, 13, 229-232	21.6	430
160	Patterns of cis regulatory variation in diverse human populations. *PLoS Genetics*, **2012**, 8, e1002639	6	361

159	Genetic Drivers of Epigenetic and Transcriptional Variation in Human Immune Cells. *Cell*, **2016**, 167, 1398-1414.e24	36.2	339
158	Comprehensive Analysis of Alternative Splicing Across Tumors from 8,705 Patients. *Cancer Cell*, **2018**, 34, 211-224.e6	24.3	327
157	scNMT-seq enables joint profiling of chromatin accessibility DNA methylation and transcription in single cells. *Nature Communications*, **2018**, 9, 781	17.4	303
156	DNA methylation in Arabidopsis has a genetic basis and shows evidence of local adaptation. *ELife*, **2015**, 4, e05255	8.9	300
155	A Bayesian framework to account for complex non-genetic factors in gene expression levels greatly increases power in eQTL studies. *PLoS Computational Biology*, **2010**, 6, e1000770	5	295
154	Common genetic variation drives molecular heterogeneity in human iPSCs. *Nature*, **2017**, 546, 370-375	50.4	294
153	Multi-Omics Factor Analysis-a framework for unsupervised integration of multi-omics data sets. *Molecular Systems Biology*, **2018**, 14, e8124	12.2	287
152	Eleven grand challenges in single-cell data science. *Genome Biology*, **2020**, 21, 31	18.3	274
151	Computational assignment of cell-cycle stage from single-cell transcriptome data. *Methods*, **2015**, 85, 54-61	4.6	259
150	Open Targets: a platform for therapeutic target identification and validation. *Nucleic Acids Research* , **2017**, 45, D985-D994	20.1	241
149	Single-cell epigenomics: Recording the past and predicting the future. *Science*, **2017**, 358, 69-75	33.3	237
148	Arabidopsis defense against Botrytis cinerea: chronology and regulation deciphered by high-resolution temporal transcriptomic analysis. *Plant Cell*, **2012**, 24, 3530-57	11.6	233
147	DeepCpG: accurate prediction of single-cell DNA methylation states using deep learning. *Genome Biology*, **2017**, 18, 67	18.3	226
146	Expression Atlas: gene and protein expression across multiple studies and organisms. *Nucleic Acids Research*, **2018**, 46, D246-D251	20.1	222
145	Multi-tissue DNA methylation age predictor in mouse. *Genome Biology*, **2017**, 18, 68	18.3	220
144	Vitamin A-Retinoic Acid Signaling Regulates Hematopoietic Stem Cell Dormancy. *Cell*, **2017**, 169, 807-823.e19	36.2	200
143	Unraveling the polygenic architecture of complex traits using blood eQTL metaanalysis		175
142	Single-cell RNA-seq and computational analysis using temporal mixture modelling resolves Th1/Tfh fate bifurcation in malaria. *Science Immunology*, **2017**, 2,	28	171

141	Human genomics. Effect of predicted protein-truncating genetic variants on the human transcriptome. *Science*, **2015**, 348, 666-9	33.3	170
140	SpatialDE: identification of spatially variable genes. *Nature Methods*, **2018**, 15, 343-346	21.6	153
139	Genomic basis for RNA alterations in cancer. *Nature*, **2020**, 578, 129-136	50.4	148
138	Benchmarking single-cell RNA-sequencing protocols for cell atlas projects. *Nature Biotechnology*, **2020**, 38, 747-755	44.5	142
137	Multi-omics profiling of mouse gastrulation at single-cell resolution. *Nature*, **2019**, 576, 487-491	50.4	137
136	Gaussian process robust regression for noisy heart rate data. *IEEE Transactions on Biomedical Engineering*, **2008**, 55, 2143-51	5	134
135	DNA Methylation and Transcription Patterns in Intestinal Epithelial Cells From Pediatric Patients With Inflammatory Bowel Diseases Differentiate Disease Subtypes and Associate With Outcome. *Gastroenterology*, **2018**, 154, 585-598	13.3	126
134	Century-scale methylome stability in a recently diverged Arabidopsis thaliana lineage. *PLoS Genetics*, **2015**, 11, e1004920	6	104
133	Haplotype-resolved diverse human genomes and integrated analysis of structural variation. *Science* , **2021**, 372,	33.3	100
132	MOFA+: a statistical framework for comprehensive integration of multi-modal single-cell data. *Genome Biology*, **2020**, 21, 111	18.3	97
131	The Polygenic and Monogenic Basis of Blood Traits and Diseases. *Cell*, **2020**, 182, 1214-1231.e11	56.2	96
130	Estimation of Free-Living Energy Expenditure by Heart Rate and Movement Sensing: A Doubly-Labelled Water Study. *PLoS ONE*, **2015**, 10, e0137206	3.7	86
129	GWAS for executive function and processing speed suggests involvement of the CADM2 gene. *Molecular Psychiatry*, **2016**, 21, 189-197	15.1	85
128	Predicting and understanding the stability of G-quadruplexes. *Bioinformatics*, **2009**, 25, i374-82	7.2	81
127	Single-cell RNA-sequencing of differentiating iPS cells reveals dynamic genetic effects on gene expression. *Nature Communications*, **2020**, 11, 810	17.4	76
126	A Lasso multi-marker mixed model for association mapping with population structure correction. *Bioinformatics*, **2013**, 29, 206-14	7.2	76
125	Whole-Genome Sequencing Coupled to Imputation Discovers Genetic Signals for Anthropometric Traits. *American Journal of Human Genetics*, **2017**, 100, 865-884	11	74
124	Joint modelling of confounding factors and prominent genetic regulators provides increased accuracy in genetical genomics studies. *PLoS Computational Biology*, **2012**, 8, e1002330	5	74

123	DNA methylation defines regional identity of human intestinal epithelial organoids and undergoes dynamic changes during development. *Gut*, **2019**, 68, 49-61	19.2	73
122	Genome-wide analysis of differential transcriptional and epigenetic variability across human immune cell types. *Genome Biology*, **2017**, 18, 18	18.3	70
121	Efficient set tests for the genetic analysis of correlated traits. *Nature Methods*, **2015**, 12, 755-8	21.6	70
120	The Kipoi repository accelerates community exchange and reuse of predictive models for genomics. *Nature Biotechnology*, **2019**, 37, 592-600	44.5	69
119	Limited Contribution of DNA Methylation Variation to Expression Regulation in Arabidopsis thaliana. *PLoS Genetics*, **2016**, 12, e1006141	6	68
118	A robust Bayesian two-sample test for detecting intervals of differential gene expression in microarray time series. *Journal of Computational Biology*, **2010**, 17, 355-67	1.7	67
117	Genomic Determinants of Protein Abundance Variation in Colorectal Cancer Cells. *Cell Reports*, **2017**, 20, 2201-2214	10.6	64
116	f-scLVM: scalable and versatile factor analysis for single-cell RNA-seq. *Genome Biology*, **2017**, 18, 212	18.3	63
115	Genotype-environment interactions reveal causal pathways that mediate genetic effects on phenotype. *PLoS Genetics*, **2013**, 9, e1003803	6	63
114	A linear mixed-model approach to study multivariate gene-environment interactions. *Nature Genetics*, **2019**, 51, 180-186	36.3	63
113	Large-scale cis- and trans-eQTL analyses identify thousands of genetic loci and polygenic scores that regulate blood gene expression. *Nature Genetics*, **2021**, 53, 1300-1310	36.3	60
112	Modeling Cell-Cell Interactions from Spatial Molecular Data with Spatial Variance Component Analysis. *Cell Reports*, **2019**, 29, 202-211.e6	10.6	57
111	Robustness and applicability of transcription factor and pathway analysis tools on single-cell RNA-seq data. *Genome Biology*, **2020**, 21, 36	18.3	57
110	Joint genetic analysis of gene expression data with inferred cellular phenotypes. *PLoS Genetics*, **2011**, 7, e1001276	6	57
109	easyGWAS: A Cloud-Based Platform for Comparing the Results of Genome-Wide Association Studies. *Plant Cell*, **2017**, 29, 5-19	11.6	56
108	A Pan-cancer Transcriptome Analysis Reveals Pervasive Regulation through Alternative Promoters. *Cell*, **2019**, 178, 1465-1477.e17	56.2	56
107	LifeTime and improving European healthcare through cell-based interceptive medicine. *Nature*, **2020**, 587, 377-386	50.4	56
106	Genetic Variation in the Social Environment Contributes to Health and Disease. *PLoS Genetics*, **2017**, 13, e1006498	6	53

105	Extensive cis-regulatory variation robust to environmental perturbation in Arabidopsis. *Plant Cell*, **2014**, 26, 4298-310	11.6	53
104	Promoter shape varies across populations and affects promoter evolution and expression noise. *Nature Genetics*, **2017**, 49, 550-558	36.3	51
103	A random forest approach to capture genetic effects in the presence of population structure. *Nature Communications*, **2015**, 6, 7432	17.4	50
102	LIMIX: genetic analysis of multiple traits		50
101	Genome-wide studies of verbal declarative memory in nondemented older people: the Cohorts for Heart and Aging Research in Genomic Epidemiology consortium. *Biological Psychiatry*, **2015**, 77, 749-63	7.9	48
100	Vireo: Bayesian demultiplexing of pooled single-cell RNA-seq data without genotype reference. *Genome Biology*, **2019**, 20, 273	18.3	46
99	Genome-Scale Oscillations in DNA Methylation during Exit from Pluripotency. *Cell Systems*, **2018**, 7, 63-76.e12	6.3	44
98	The Human Cell Atlas **2017**,		41
97	A toolbox for predicting g-quadruplex formation and stability. *Journal of Nucleic Acids*, **2010**, 2010,	2.3	40
96	Genetic variants regulating expression levels and isoform diversity during embryogenesis. *Nature*, **2017**, 541, 402-406	50.4	39
95	Genome-wide mapping of genetic determinants influencing DNA methylation and gene expression in human hippocampus. *Nature Communications*, **2017**, 8, 1511	17.4	37
94	Population-scale single-cell RNA-seq profiling across dopaminergic neuron differentiation. *Nature Genetics*, **2021**, 53, 304-312	36.3	37
93	Combined single-cell profiling of expression and DNA methylation reveals splicing regulation and heterogeneity. *Genome Biology*, **2019**, 20, 30	18.3	36
92	Screening for genes that accelerate the epigenetic aging clock in humans reveals a role for the H3K36 methyltransferase NSD1. *Genome Biology*, **2019**, 20, 146	18.3	36
91	Comprehensive mapping of tissue cell architecture via integrated single cell and spatial transcriptomics		36
90	Warped linear mixed models for the genetic analysis of transformed phenotypes. *Nature Communications*, **2014**, 5, 4890	17.4	35
89	A Palaeolithic-type diet causes strong tissue-specific effects on ectopic fat deposition in obese postmenopausal women. *Journal of Internal Medicine*, **2013**, 274, 67-76	10.8	35
88	Computational principles and challenges in single-cell data integration. *Nature Biotechnology*, **2021**, 39, 1202-1215	44.5	33

87	Platelet function is modified by common sequence variation in megakaryocyte super enhancers. *Nature Communications*, **2017**, 8, 16058	17.4	30
86	Accurate detection of differential RNA processing. *Nucleic Acids Research*, **2013**, 41, 5189-98	20.1	30
85	The Organoid Cell Atlas. *Nature Biotechnology*, **2021**, 39, 13-17	44.5	30
84	A high-content platform to characterise human induced pluripotent stem cell lines. *Methods*, **2016**, 96, 85-96	4.6	28
83	A Single-Cell Transcriptomics CRISPR-Activation Screen Identifies Epigenetic Regulators of the Zygotic Genome Activation Program. *Cell Systems*, **2020**, 11, 25-41.e9	10.6	27
82	The germline genetic component of drug sensitivity in cancer cell lines. *Nature Communications*, **2018**, 9, 3385	17.4	24
81	Structural rearrangements generate cell-specific, gene-independent CRISPR-Cas9 loss of fitness effects. *Genome Biology*, **2019**, 20, 27	18.3	22
80	Properties of structural variants and short tandem repeats associated with gene expression and complex traits. *Nature Communications*, **2020**, 11, 2927	17.4	22
79	Cell2location maps fine-grained cell types in spatial transcriptomics.. *Nature Biotechnology*, **2022**,	44.5	18
78	Cardelino: computational integration of somatic clonal substructure and single-cell transcriptomes. *Nature Methods*, **2020**, 17, 414-421	21.6	17
77	Modelling local gene networks increases power to detect trans-acting genetic effects on gene expression. *Genome Biology*, **2016**, 17, 33	18.3	16
76	Population-scale proteome variation in human induced pluripotent stem cells. *ELife*, **2020**, 9,	8.9	16
75	Naive Pluripotent Stem Cells Exhibit Phenotypic Variability that Is Driven by Genetic Variation. *Cell Stem Cell*, **2020**, 27, 470-481.e6	18	16
74	Identification of rare and common regulatory variants in pluripotent cells using population-scale transcriptomics. *Nature Genetics*, **2021**, 53, 313-321	36.3	16
73	Identifying Extrinsic versus Intrinsic Drivers of Variation in Cell Behavior in Human iPSC Lines from Healthy Donors. *Cell Reports*, **2019**, 26, 2078-2087.e3	10.6	16
72	Genomic Rearrangements in Considered as Quantitative Traits. *Genetics*, **2017**, 205, 1425-1441	4	15
71	GeneCodeq: quality score compression and improved genotyping using a Bayesian framework. *Bioinformatics*, **2016**, 32, 3124-3132	7.2	14
70	Accurate prediction of single-cell DNA methylation states using deep learning		14

69	Cell segmentation-free inference of cell types from in situ transcriptomics data. *Nature Communications*, **2021**, 12, 3545	17.4	14
68	Germline determinants of the somatic mutation landscape in 2,642 cancer genomes		13
67	Kipoi: accelerating the community exchange and reuse of predictive models for genomics		13
66	Benchmarking Single-Cell RNA Sequencing Protocols for Cell Atlas Projects		13
65	Joint genetic analysis using variant sets reveals polygenic gene-context interactions. *PLoS Genetics*, **2017**, 13, e1006693	6	12
64	Personalized medicine: from genotypes, molecular phenotypes and the quantified self, towards improved medicine. *Pacific Symposium on Biocomputing Pacific Symposium on Biocomputing*, **2015**, 342-6	1.3	12
63	Systematic genetic analysis of the MHC region reveals mechanistic underpinnings of HLA type associations with disease. *ELife*, **2019**, 8,	8.9	12
62	Cardelino: Integrating whole exomes and single-cell transcriptomes to reveal phenotypic impact of somatic variants		12
61	Single-cell RNA-sequencing of differentiating iPS cells reveals dynamic genetic effects on gene expression		12
60	Discovery and quality analysis of a comprehensive set of structural variants and short tandem repeats. *Nature Communications*, **2020**, 11, 2928	17.4	11
59	Detecting regulatory gene-environment interactions with unmeasured environmental factors. *Bioinformatics*, **2013**, 29, 1382-9	7.2	10
58	Statistical Tests for Detecting Differential RNA-Transcript Expression from Read Counts. *Nature Precedings*, **2010**,		10
57	Genomic basis for RNA alterations revealed by whole-genome analyses of 27 cancer types		10
56	MOFA+: a probabilistic framework for comprehensive integration of structured single-cell data		10
55	Subclone-specific microenvironmental impact and drug response in refractory multiple myeloma revealed by single-cell transcriptomics. *Nature Communications*, **2021**, 12, 6960	17.4	9
54	Modelling cell-cell interactions from spatial molecular data with spatial variance component analysis		9
53	Systematic assessment of regulatory effects of human disease variants in pluripotent cells		9
52	Cell segmentation-free inference of cell types from in situ transcriptomics data		9

51	Interactions between genetic variation and cellular environment in skeletal muscle gene expression. *PLoS ONE*, **2018**, 13, e0195788	3.7	9
50	A spatial multi-omics atlas of the human lung reveals a novel immune cell survival niche		8
49	Genetic associations at regulatory phenotypes improve fine-mapping of causal variants for twelve immune-mediated diseases		8
48	A linear mixed model approach to study multivariate gene-environment interactions		8
47	Generalized correlation measure using count statistics for gene expression data with ordered samples. *Bioinformatics*, **2018**, 34, 617-624	7.2	7
46	Common genetic variation drives molecular heterogeneity in human IPSCs		7
45	Multi-Omics factor analysis - a framework for unsupervised integration of multi-omic data sets		7
44	IceR improves proteome coverage and data completeness in global and single-cell proteomics. *Nature Communications*, **2021**, 12, 4787	17.4	7
43	Identifying temporal and spatial patterns of variation from multi-modal data using MEFISTO		6
42	Population-scale proteome variation in human induced pluripotent stem cells		6
41	Single cell multi-omics profiling reveals a hierarchical epigenetic landscape during mammalian germ layer specification		6
40	Scalable latent-factor models applied to single-cell RNA-seq data separate biological drivers from confounding effects		6
39	Identifying temporal and spatial patterns of variation from multimodal data using MEFISTO.. *Nature Methods*, **2022**,	21.6	5
38	SpatialDE - Identification of spatially variable genes		5
37	Multi-omics Characterization of Interaction-mediated Control of Human Protein Abundance levels. *Molecular and Cellular Proteomics*, **2019**, 18, S114-S125	7.6	4
36	PERSONALIZED MEDICINE: FROM GENOTYPES, MOLECULAR PHENOTYPES AND THE QUANTIFIED SELF, TOWARDS IMPROVED MEDICINE **2014**,		4
35	ShapePheno: unsupervised extraction of shape phenotypes from biological image collections. *Bioinformatics*, **2012**, 28, 1001-8	7.2	4
34	Integrative genome-wide analysis of the determinants of RNA splicing in kidney renal clear cell carcinoma. *Pacific Symposium on Biocomputing Pacific Symposium on Biocomputing*, **2015**, 44-55	1.3	4

33	Temporal mixture modelling of single-cell RNA-seq data resolves a CD4+ T cell fate bifurcation		4
32	scNMT-seq enables joint profiling of chromatin accessibility DNA methylation and transcription in single cells		4
31	Mitochondrial DNA variants modulate N-formylmethionine, proteostasis and risk of late-onset human diseases. *Nature Medicine*, **2021**, 27, 1564-1575	50.5	4
30	Inference algorithms and learning theory for Bayesian sparse factor analysis. *Journal of Physics: Conference Series*, **2009**, 197, 012002	0.3	3
29	Genomic properties of structural variants and short tandem repeats that impact gene expression and complex traits in humans		3
28	Genetic Analyses of Blood Cell Structure for Biological and Pharmacological Inference		3
27	The Polygenic and Monogenic Basis of Blood Traits and Diseases		3
26	De novo assembly of 64 haplotype-resolved human genomes of diverse ancestry and integrated analysis of structural variation		3
25	Dissecting indirect genetic effects from peers in laboratory mice		3
24	Tandem duplications lead to loss of fitness effects in CRISPR-Cas9 data		3
23	Vireo: Bayesian demultiplexing of pooled single-cell RNA-seq data without genotype reference		3
22	A Pan-Cancer Transcriptome Analysis Reveals Pervasive Regulation through Tumor-Associated Alternative Promoters		3
21	Erosion of human X chromosome inactivation causes major remodeling of the iPSC proteome. *Cell Reports*, **2021**, 35, 109032	10.6	3
20	Author response: DNA methylation in Arabidopsis has a genetic basis and shows evidence of local adaptation **2015**,		2
19	Discovery and Quality Analysis of a Comprehensive Set of Structural Variants and Short Tandem Repeats		2
18	SpatialDE2: Fast and localized variance component analysis of spatial transcriptomics		2
17	LiMMBo: a simple, scalable approach for linear mixed models in high-dimensional genetic association studies		2
16	Muon: multimodal omics analysis framework		2

List of Publications

15	Single-cell Atlas of common variable immunodeficiency shows germinal center-associated epigenetic dysregulation in B-cell responses.. *Nature Communications*, **2022**, 13, 1779	17.4	2
14	Personalized medicine: from genotypes and molecular phenotypes towards therapy- session introduction. *Pacific Symposium on Biocomputing Pacific Symposium on Biocomputing*, **2014**, 19, 224-8	1.3	1
13	Efficient branch-and-bound techniques for two-locus association mapping. *BMC Bioinformatics*, **2011**, 12,	3.6	1
12	MUON: multimodal omics analysis framework.. *Genome Biology*, **2022**, 23, 42	18.3	1
11	FISHFactor: A Probabilistic Factor Model for Spatial Transcriptomics Data with Subcellular Resolution		1
10	Combined single-cell profiling of expression and DNA methylation reveals splicing regulation and heterogeneity		1
9	A Robust Bayesian Two-Sample Test for Detecting Intervals of Differential Gene Expression in Microarray Time Series. *Lecture Notes in Computer Science*, **2009**, 201-216	0.9	1
8	Warped Matrix Factorisation for Multi-view Data Integration. *Lecture Notes in Computer Science*, **2016**, 789-804	0.9	1
7	Mapping interindividual dynamics of innate immune response at single-cell resolution		1
6	CellRegMap: A statistical framework for mapping context-specific regulatory variants using scRNA-seq		1
5	Genetic associations at regulatory phenotypes improve fine-mapping of causal variants for 12 immune-mediated diseases.. *Nature Genetics*, **2022**, 54, 251-262	36.3	1
4	scDALI: modeling allelic heterogeneity in single cells reveals context-specific genetic regulation.. *Genome Biology*, **2022**, 23, 8	18.3	0
3	Dissecting indirect genetic effects from peers in laboratory mice. *Genome Biology*, **2021**, 22, 216	18.3	0
2	Reply. *Gastroenterology*, **2018**, 155, 230-231	13.3	
1	PERSONALIZED MEDICINE: FROM GENOTYPES AND MOLECULAR PHENOTYPES TOWARDS COMPUTED THERAPY. *Pacific Symposium on Biocomputing Pacific Symposium on Biocomputing*, **2013**, 18, 171-174	1.3	