

# Alicia Oshlack

## List of Publications by Year in descending order

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Version: 2024-02-01

110  
papers

23,530  
citations

38660

50  
h-index

27345

106  
g-index

145  
all docs

145  
docs citations

145  
times ranked

43956  
citing authors

#	ARTICLE	IF	CITATIONS
1	A scaling normalization method for differential expression analysis of RNA-seq data. <i>Genome Biology</i> , 2010, 11, R25.	13.9	6,234
2	Gene ontology analysis for RNA-seq: accounting for selection bias. <i>Genome Biology</i> , 2010, 11, R14.	13.9	5,824
3	A comparison of background correction methods for two-colour microarrays. <i>Bioinformatics</i> , 2007, 23, 2700-2707.	1.8	829
4	SWAN: Subset-quantile Within Array Normalization for Illumina Infinium HumanMethylation450 BeadChips. <i>Genome Biology</i> , 2012, 13, R44.	13.9	697
5	Splatter: simulation of single-cell RNA sequencing data. <i>Genome Biology</i> , 2017, 18, 174.	3.8	626
6	From RNA-seq reads to differential expression results. <i>Genome Biology</i> , 2010, 11, 220.	13.9	603
7	missMethyl: an R package for analyzing data from Illumina's HumanMethylation450 platform. <i>Bioinformatics</i> , 2016, 32, 286-288.	1.8	573
8	Clustering trees: a visualization for evaluating clusterings at multiple resolutions. <i>GigaScience</i> , 2018, 7, .	3.3	447
9	Corset: enabling differential gene expression analysis for de novo assembled transcriptomes. <i>Genome Biology</i> , 2014, 15, 410.	3.8	427
10	Transcript length bias in RNA-seq data confounds systems biology. <i>Biology Direct</i> , 2009, 4, 14.	1.9	422
11	A prospective evaluation of whole-exome sequencing as a first-tier molecular test in infants with suspected monogenic disorders. <i>Genetics in Medicine</i> , 2016, 18, 1090-1096.	1.1	332
12	Expression profiling in primates reveals a rapid evolution of human transcription factors. <i>Nature</i> , 2006, 440, 242-245.	13.7	283
13	Diagnostic Impact and Cost-effectiveness of Whole-Exome Sequencing for Ambulant Children With Suspected Monogenic Conditions. <i>JAMA Pediatrics</i> , 2017, 171, 855.	3.3	252
14	ChIP-seq analysis reveals distinct H3K27me3 profiles that correlate with transcriptional activity. <i>Nucleic Acids Research</i> , 2011, 39, 7415-7427.	6.5	250
15	Disorders of sex development: insights from targeted gene sequencing of a large international patient cohort. <i>Genome Biology</i> , 2016, 17, 243.	3.8	241
16	Array-Based Gene Discovery with Three Unrelated Subjects Shows SCARB2/LIMP-2 Deficiency Causes Myoclonus Epilepsy and Glomerulosclerosis. <i>American Journal of Human Genetics</i> , 2008, 82, 673-684.	2.6	230
17	Exploring the single-cell RNA-seq analysis landscape with the scRNA-tools database. <i>PLoS Computational Biology</i> , 2018, 14, e1006245.	1.5	222
18	Optimizing the noise versus bias trade-off for Illumina whole genome expression BeadChips. <i>Nucleic Acids Research</i> , 2010, 38, e204-e204.	6.5	202

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19	Natural selection on gene expression. Trends in Genetics, 2006, 22, 456-461.	2.9	187
20	Evaluation of variability in human kidney organoids. Nature Methods, 2019, 16, 79-87.	9.0	176
21	3D organoid-derived human glomeruli for personalised podocyte disease modelling and drug screening. Nature Communications, 2018, 9, 5167.	5.8	175
22	Single-cell analysis reveals congruence between kidney organoids and human fetal kidney. Genome Medicine, 2019, 11, 3.	3.6	158
23	Patient-iPSC-Derived Kidney Organoids Show Functional Validation of a Ciliopathic Renal Phenotype and Reveal Underlying Pathogenetic Mechanisms. American Journal of Human Genetics, 2018, 102, 816-831.	2.6	157
24	Differentiation of human embryonic stem cells to HOXA+ hemogenic vasculature that resembles the aorta-gonad-mesonephros. Nature Biotechnology, 2016, 34, 1168-1179.	9.4	150
25	Bpipe: a tool for running and managing bioinformatics pipelines. Bioinformatics, 2012, 28, 1525-1526.	1.8	145
26	Gene Regulation in Primates Evolves under Tissue-Specific Selection Pressures. PLoS Genetics, 2008, 4, e1000271.	1.5	143
27	JAFFA: High sensitivity transcriptome-focused fusion gene detection. Genome Medicine, 2015, 7, 43.	3.6	132
28	Single cell analysis of the developing mouse kidney provides deeper insight into marker gene expression and ligand-receptor crosstalk. Development (Cambridge), 2019, 146, .	1.2	123
29	STRetch: detecting and discovering pathogenic short tandem repeat expansions. Genome Biology, 2018, 19, 121.	3.8	117
30	Genome-wide DNA methylation analysis identifies hypomethylated genes regulated by FOXP3 in human regulatory T cells. Blood, 2013, 122, 2823-2836.	0.6	114
31	Identification of recurrent <i>FGFR3</i> fusion genes in lung cancer through RNA sequencing. Journal of Pathology, 2013, 230, 270-276.	2.1	113
32	The polycomb repressive complex 2 governs life and death of peripheral T cells. Blood, 2014, 124, 737-749.	0.6	111
33	A cross-package Bioconductor workflow for analysing methylation array data. F1000Research, 2016, 5, 1281.	0.8	103
34	Analysis of epigenetic changes in survivors of preterm birth reveals the effect of gestational age and evidence for a long term legacy. Genome Medicine, 2013, 5, 96.	3.6	101
35	RNA sequencing reveals sexually dimorphic gene expression before gonadal differentiation in chicken and allows comprehensive annotation of the W-chromosome. Genome Biology, 2013, 14, R26.	13.9	98
36	Kidney micro-organoids in suspension culture as a scalable source of human pluripotent stem cell-derived kidney cells. Development (Cambridge), 2019, 146, .	1.2	97

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37	A cross-package Bioconductor workflow for analysing methylation array data. F1000Research, 0, 5, 1281.	0.8	97
38	A cross-package Bioconductor workflow for analysing methylation array data. F1000Research, 2016, 5, 1281.	0.8	93
39	DiffVar: a new method for detecting differential variability with application to methylation in cancer and aging. Genome Biology, 2014, 15, 465.	3.8	84
40	SuperTranscripts: a data driven reference for analysis and visualisation of transcriptomes. Genome Biology, 2017, 18, 148.	3.8	79
41	Cpipe: a shared variant detection pipeline designed for diagnostic settings. Genome Medicine, 2015, 7, 68.	3.6	78
42	NKX2-5 regulates human cardiomyogenesis via a HEY2 dependent transcriptional network. Nature Communications, 2018, 9, 1373.	5.8	77
43	Gene length and detection bias in single cell RNA sequencing protocols. F1000Research, 2017, 6, 595.	0.8	76
44	Removing unwanted variation in a differential methylation analysis of Illumina HumanMethylation450 array data. Nucleic Acids Research, 2015, 43, e106-e106.	6.5	73
45	A Very Radio Loud Narrowâ€Line Seyfert 1: PKS 2004â€447. Astrophysical Journal, 2001, 558, 578-582.	1.6	72
46	Expression discordance of monozygotic twins at birth: Effect of intrauterine environment and a possible mechanism for fetal programming. Epigenetics, 2011, 6, 579-592.	1.3	70
47	Condensin I associates with structural and gene regulatory regions in vertebrate chromosomes. Nature Communications, 2013, 4, 2537.	5.8	70
48	Gene set enrichment analysis for genome-wide DNA methylation data. Genome Biology, 2021, 22, 173.	3.8	68
49	Black Hole Mass Estimates of Radioâ€selected Quasars. Astrophysical Journal, 2002, 576, 81-88.	1.6	66
50	Normalization of boutique two-color microarrays with a high proportion of differentially expressed probes. Genome Biology, 2007, 8, R2.	13.9	61
51	Using DNA microarrays to study gene expression in closely related species. Bioinformatics, 2007, 23, 1235-1242.	1.8	60
52	Sierra: discovery of differential transcript usage from polyA-captured single-cell RNA-seq data. Genome Biology, 2020, 21, 167.	3.8	59
53	The spectral energy distribution of PKS 2004-447: a compact steep-spectrum source and possible radio-loud narrow-line Seyfert 1 galaxy. Monthly Notices of the Royal Astronomical Society, 2006, 370, 245-254.	1.6	54
54	Identification of candidate gonadal sex differentiation genes in the chicken embryo using RNA-seq. BMC Genomics, 2015, 16, 704.	1.2	54

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55	The application of RNA sequencing for the diagnosis and genomic classification of pediatric acute lymphoblastic leukemia. <i>Blood Advances</i> , 2020, 4, 930-942.	2.5	52
56	Purification and Transcriptomic Analysis of Mouse Fetal Leydig Cells Reveals Candidate Genes for Specification of Gonadal Steroidogenic Cells. <i>Biology of Reproduction</i> , 2015, 92, 145.	1.2	51
57	Nephron progenitor commitment is a stochastic process influenced by cell migration. <i>ELife</i> , 2019, 8, .	2.8	47
58	Sex-Specific Control of Human Heart Maturation by the Progesterone Receptor. <i>Circulation</i> , 2021, 143, 1614-1628.	1.6	42
59	Jarid2 regulates hematopoietic stem cell function by acting with polycomb repressive complex 2. <i>Blood</i> , 2015, 125, 1890-1900.	0.6	41
60	A Combination of Genomic Approaches Reveals the Role of FOXO1a in Regulating an Oxidative Stress Response Pathway. <i>PLoS ONE</i> , 2008, 3, e1670.	1.1	41
61	SuperFreq: Integrated mutation detection and clonal tracking in cancer. <i>PLoS Computational Biology</i> , 2020, 16, e1007603.	1.5	37
62	Diagnostic and cost utility of whole exome sequencing in peripheral neuropathy. <i>Annals of Clinical and Translational Neurology</i> , 2017, 4, 318-325.	1.7	36
63	Ximmer: a system for improving accuracy and consistency of CNV calling from exome data. <i>GigaScience</i> , 2018, 7, .	3.3	32
64	A comparison of control samples for CHIP-seq of histone modifications. <i>Frontiers in Genetics</i> , 2014, 5, 329.	1.1	31
65	MCM3AP in recessive Charcot-Marie-Tooth neuropathy and mild intellectual disability. <i>Brain</i> , 2017, 140, 2093-2103.	3.7	31
66	Identification, Expression, and Regulation of Anti-Müllerian Hormone Type-II Receptor in the Embryonic Chicken Gonad. <i>Biology of Reproduction</i> , 2014, 90, 106.	1.2	28
67	Haploinsufficiency for the Six2 gene increases nephron progenitor proliferation promoting branching and nephron number. <i>Kidney International</i> , 2018, 93, 589-598.	2.6	27
68	Segmental Duplications Contribute to Gene Expression Differences Between Humans and Chimpanzees. <i>Genetics</i> , 2009, 182, 627-630.	1.2	26
69	Cell-Type-Specific Transcriptional Profiles of the Dimorphic Pathogen <i>Penicillium marneffei</i> Reflect Distinct Reproductive, Morphological, and Environmental Demands. <i>G3: Genes, Genomes, Genetics</i> , 2013, 3, 1997-2014.	0.8	25
70	ALLSorts: an RNA-Seq subtype classifier for B-cell acute lymphoblastic leukemia. <i>Blood Advances</i> , 2022, 6, 4093-4097.	2.5	25
71	A clinically driven variant prioritization framework outperforms purely computational approaches for the diagnostic analysis of singleton WES data. <i>European Journal of Human Genetics</i> , 2017, 25, 1268-1272.	1.4	24
72	Transcriptional profiles for distinct aggregation states of mutant Huntingtin exon 1 protein unmask new Huntington's disease pathways. <i>Molecular and Cellular Neurosciences</i> , 2017, 83, 103-112.	1.0	23

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73	Accuracy of short tandem repeats genotyping tools in whole exome sequencing data. <i>F1000Research</i> , 2020, 9, 200.	0.8	23
74	miRNA-Seq normalization comparisons need improvement. <i>Rna</i> , 2013, 19, 733-734.	1.6	22
75	Co-option of the cardiac transcription factor Nkx2.5 during development of the emu wing. <i>Nature Communications</i> , 2017, 8, 132.	5.8	21
76	Direct reprogramming to human nephron progenitor-like cells using inducible piggyBac transposon expression of SNAI2-EYA1-SIX1. <i>Kidney International</i> , 2019, 95, 1153-1166.	2.6	21
77	JAFAL: detecting fusion genes with long-read transcriptome sequencing. <i>Genome Biology</i> , 2022, 23, 10.	3.8	20
78	Enhancer retargeting of <i>CDX2</i> and <i>UBTF::ATXN7L3</i> define a subtype of high-risk B-progenitor acute lymphoblastic leukemia. <i>Blood</i> , 2022, 139, 3519-3531.	0.6	20
79	Statistical analysis of an RNA titration series evaluates microarray precision and sensitivity on a whole-array basis. <i>BMC Bioinformatics</i> , 2006, 7, 511.	1.2	19
80	Clinker: visualizing fusion genes detected in RNA-seq data. <i>GigaScience</i> , 2018, 7, .	3.3	17
81	Susceptibility to Acute Rheumatic Fever Based on Differential Expression of Genes Involved in Cytotoxicity, Chemotaxis, and Apoptosis. <i>Infection and Immunity</i> , 2014, 82, 753-761.	1.0	16
82	MINTIE: identifying novel structural and splice variants in transcriptomes using RNA-seq data. <i>Genome Biology</i> , 2021, 22, 296.	3.8	16
83	Human yolk sac-like haematopoiesis generates <i>RUNX1</i> - and <i>GFI1/1B</i> -dependent blood and <i>SOX17</i> -positive endothelium. <i>Development (Cambridge)</i> , 2020, 147, .	1.2	15
84	Bazam: a rapid method for read extraction and realignment of high-throughput sequencing data. <i>Genome Biology</i> , 2019, 20, 78.	3.8	13
85	Limb patterning genes and heterochronic development of the emu wing bud. <i>EvoDevo</i> , 2016, 7, 26.	1.3	12
86	Cord Blood CD8+ T Cells Have a Natural Propensity to Express IL-4 in a Fatty Acid Metabolism and Caspase Activation-Dependent Manner. <i>Frontiers in Immunology</i> , 2018, 9, 879.	2.2	11
87	STRipy: A graphical application for enhanced genotyping of pathogenic short tandem repeats in sequencing data. <i>Human Mutation</i> , 2022, 43, 859-868.	1.1	11
88	Genotyping microsatellites in next-generation sequencing data. <i>BMC Bioinformatics</i> , 2015, 16, .	1.2	9
89	Necklace: combining reference and assembled transcriptomes for more comprehensive RNA-Seq analysis. <i>GigaScience</i> , 2018, 7, .	3.3	9
90	Targeted therapy and disease monitoring in CNTRL-FCFR1-driven leukaemia. <i>Pediatric Blood and Cancer</i> , 2019, 66, e27897.	0.8	8

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91	Detecting copy number alterations in RNA-Seq using SuperFreq. <i>Bioinformatics</i> , 2021, 37, 4023-4032.	1.8	8
92	Using equivalence class counts for fast and accurate testing of differential transcript usage. <i>F1000Research</i> , 2019, 8, 265.	0.8	8
93	Sixty years of genome biology. <i>Genome Biology</i> , 2013, 14, 113.	13.9	6
94	Differential Expression for RNA Sequencing (RNA-Seq) Data: Mapping, Summarization, Statistical Analysis, and Experimental Design. , 2012, , 169-190.		4
95	MLL-TFE3: a novel and aggressive KMT2A fusion identified in infant leukemia. <i>Blood Advances</i> , 2020, 4, 4918-4923.	2.5	4
96	SFPQ-ABL1 and BCR-ABL1 use different signaling networks to drive B-cell acute lymphoblastic leukemia. <i>Blood Advances</i> , 2022, 6, 2373-2387.	2.5	4
97	splatPop: simulating population scale single-cell RNA sequencing data. <i>Genome Biology</i> , 2021, 22, 341.	3.8	4
98	Near Infrared Micro-variability of Radio-loud Quasars. <i>Publications of the Astronomical Society of Australia</i> , 2002, 19, 222-227.	1.3	3
99	Shifts in ovine cardiopulmonary microRNA expression in late gestation and the perinatal period. <i>PLoS ONE</i> , 2018, 13, e0204038.	1.1	3
100	The role of cardiac transcription factor NKX2-5 in regulating the human cardiac miRNAome. <i>Scientific Reports</i> , 2019, 9, 15928.	1.6	3
101	Fast and accurate differential transcript usage by testing equivalence class counts. <i>F1000Research</i> , 2019, 8, 265.	0.8	3
102	A 10-step guide to party conversation for bioinformaticians. <i>Genome Biology</i> , 2013, 14, 104.	13.9	2
103	Slinker: Visualising novel splicing events in RNA-Seq data. <i>F1000Research</i> , 2021, 10, 1255.	0.8	2
104	DNA Methylation Profiles of Purified Cell Types in Bronchoalveolar Lavage: Applications for Mixed Cell Paediatric Pulmonary Studies. <i>Frontiers in Immunology</i> , 2021, 12, 788705.	2.2	2
105	As we come to the end of 2011, several members of the <i>Genome Biology</i> Editorial Board give their views on the state of play in genomics. <i>Genome Biology</i> , 2011, 12, 137.	13.9	0
106	Whole exome sequencing in systemic juvenile idiopathic arthritis. <i>Pathology</i> , 2016, 48, S43.	0.3	0
107	THE NATURE OF THE OPTICAL EMISSION IN RADIO-SELECTED AGN. , 2004, , .		0
108	The Dose-Dependent Effects of Microrna-155 in Acute Myeloid Leukemia. <i>Blood</i> , 2016, 128, 2841-2841.	0.6	0

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109	Different Classes of ABL1 Fusions Activate Different Downstream Signalling Nodes. Blood, 2018, 132, 2628-2628.	0.6	0
110	Diagnostic Utility of Multimodal Genomic Profiling for Molecular Classification and MRD Assessment in Adult B-Cell Acute Lymphoblastic Leukemia. Blood, 2021, 138, 274-274.	0.6	0