

# Alicia Oshlack

## List of Publications by Year in descending order

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Version: 2024-02-01

110  
papers

23,530  
citations

44444

50  
h-index

31191

106  
g-index

145  
all docs

145  
docs citations

145  
times ranked

48090  
citing authors

#	ARTICLE	IF	CITATIONS
1	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
2	JAFFAL: detecting fusion genes with long-read transcriptome sequencing. <i>Genome Biology</i> , 2022, 23, 10.	3.8	20
3	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
4	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
5	ALLSorts: an RNA-Seq subtype classifier for B-cell acute lymphoblastic leukemia. <i>Blood Advances</i> , 2022, 6, 4093-4097.	2.5	25
6	Sex-Specific Control of Human Heart Maturation by the Progesterone Receptor. <i>Circulation</i> , 2021, 143, 1614-1628.	1.6	42
7	Detecting copy number alterations in RNA-Seq using SuperFreq. <i>Bioinformatics</i> , 2021, 37, 4023-4032.	1.8	8
8	Gene set enrichment analysis for genome-wide DNA methylation data. <i>Genome Biology</i> , 2021, 22, 173.	3.8	68
9	MINTIE: identifying novel structural and splice variants in transcriptomes using RNA-seq data. <i>Genome Biology</i> , 2021, 22, 296.	3.8	16
10	Diagnostic Utility of Multimodal Genomic Profiling for Molecular Classification and MRD Assessment in Adult B-Cell Acute Lymphoblastic Leukemia. <i>Blood</i> , 2021, 138, 274-274.	0.6	0
11	Slinker: Visualising novel splicing events in RNA-Seq data. <i>F1000Research</i> , 2021, 10, 1255.	0.8	2
12	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
13	splatPop: simulating population scale single-cell RNA sequencing data. <i>Genome Biology</i> , 2021, 22, 341.	3.8	4
14	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
15	MLL-TFE3: a novel and aggressive KMT2A fusion identified in infant leukemia. <i>Blood Advances</i> , 2020, 4, 4918-4923.	2.5	4
16	Sierra: discovery of differential transcript usage from polyA-captured single-cell RNA-seq data. <i>Genome Biology</i> , 2020, 21, 167.	3.8	59
17	SuperFreq: Integrated mutation detection and clonal tracking in cancer. <i>PLoS Computational Biology</i> , 2020, 16, e1007603.	1.5	37
18	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

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19	Accuracy of short tandem repeats genotyping tools in whole exome sequencing data. F1000Research, 2020, 9, 200.	0.8	23
20	Targeted therapy and disease monitoring in CNTRLâ€FGFR1â€ driven leukaemia. Pediatric Blood and Cancer, 2019, 66, e27897.	0.8	8
21	The role of cardiac transcription factor NKX2-5 in regulating the human cardiac miRNAome. Scientific Reports, 2019, 9, 15928.	1.6	3
22	Single-cell analysis reveals congruence between kidney organoids and human fetal kidney. Genome Medicine, 2019, 11, 3.	3.6	158
23	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
24	Bazam: a rapid method for read extraction and realignment of high-throughput sequencing data. Genome Biology, 2019, 20, 78.	3.8	13
25	Direct reprogramming to human nephron progenitor-like cells using inducible piggyBac transposon expression of SNAI2-EYA1-SIX1. Kidney International, 2019, 95, 1153-1166.	2.6	21
26	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
27	Evaluation of variability in human kidney organoids. Nature Methods, 2019, 16, 79-87.	9.0	176
28	Nephron progenitor commitment is a stochastic process influenced by cell migration. ELife, 2019, 8, .	2.8	47
29	Fast and accurate differential transcript usage by testing equivalence class counts. F1000Research, 2019, 8, 265.	0.8	3
30	Using equivalence class counts for fast and accurate testing of differential transcript usage. F1000Research, 2019, 8, 265.	0.8	8
31	NKX2-5 regulates human cardiomyogenesis via a HEY2 dependent transcriptional network. Nature Communications, 2018, 9, 1373.	5.8	77
32	Necklace: combining reference and assembled transcriptomes for more comprehensive RNA-Seq analysis. GigaScience, 2018, 7, .	3.3	9
33	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
34	Haploinsufficiency for the Six2 gene increases nephron progenitor proliferation promoting branching and nephron number. Kidney International, 2018, 93, 589-598.	2.6	27
35	3D organoid-derived human glomeruli for personalised podocyte disease modelling and drug screening. Nature Communications, 2018, 9, 5167.	5.8	175
36	Clinker: visualizing fusion genes detected in RNA-seq data. GigaScience, 2018, 7, .	3.3	17

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37	Shifts in ovine cardiopulmonary microRNA expression in late gestation and the perinatal period. PLoS ONE, 2018, 13, e0204038.	1.1	3
38	Ximmer: a system for improving accuracy and consistency of CNV calling from exome data. GigaScience, 2018, 7, .	3.3	32
39	Exploring the single-cell RNA-seq analysis landscape with the scRNA-tools database. PLoS Computational Biology, 2018, 14, e1006245.	1.5	222
40	Cord Blood CD8+ T Cells Have a Natural Propensity to Express IL-4 in a Fatty Acid Metabolism and Caspase Activation-Dependent Manner. Frontiers in Immunology, 2018, 9, 879.	2.2	11
41	Clustering trees: a visualization for evaluating clusterings at multiple resolutions. GigaScience, 2018, 7, .	3.3	447
42	STretch: detecting and discovering pathogenic short tandem repeat expansions. Genome Biology, 2018, 19, 121.	3.8	117
43	Different Classes of ABL1 Fusions Activate Different Downstream Signalling Nodes. Blood, 2018, 132, 2628-2628.	0.6	0
44	Diagnostic and cost utility of whole exome sequencing in peripheral neuropathy. Annals of Clinical and Translational Neurology, 2017, 4, 318-325.	1.7	36
45	MCM3AP in recessive Charcot-Marie-Tooth neuropathy and mild intellectual disability. Brain, 2017, 140, 2093-2103.	3.7	31
46	A clinically driven variant prioritization framework outperforms purely computational approaches for the diagnostic analysis of singleton WES data. European Journal of Human Genetics, 2017, 25, 1268-1272.	1.4	24
47	Transcriptional profiles for distinct aggregation states of mutant Huntingtin exon 1 protein unmask new Huntington's disease pathways. Molecular and Cellular Neurosciences, 2017, 83, 103-112.	1.0	23
48	Diagnostic Impact and Cost-effectiveness of Whole-Exome Sequencing for Ambulant Children With Suspected Monogenic Conditions. JAMA Pediatrics, 2017, 171, 855.	3.3	252
49	Co-option of the cardiac transcription factor Nkx2.5 during development of the emu wing. Nature Communications, 2017, 8, 132.	5.8	21
50	SuperTranscripts: a data driven reference for analysis and visualisation of transcriptomes. Genome Biology, 2017, 18, 148.	3.8	79
51	Splatter: simulation of single-cell RNA sequencing data. Genome Biology, 2017, 18, 174.	3.8	626
52	Gene length and detection bias in single cell RNA sequencing protocols. F1000Research, 2017, 6, 595.	0.8	76
53	Disorders of sex development: insights from targeted gene sequencing of a large international patient cohort. Genome Biology, 2016, 17, 243.	3.8	241
54	Limb patterning genes and heterochronic development of the emu wing bud. EvoDevo, 2016, 7, 26.	1.3	12

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55	Differentiation of human embryonic stem cells to HOXA+ hemogenic vasculature that resembles the aorta-gonad-mesonephros. <i>Nature Biotechnology</i> , 2016, 34, 1168-1179.	9.4	150
56	Whole exome sequencing in systemic juvenile idiopathic arthritis. <i>Pathology</i> , 2016, 48, S43.	0.3	0
57	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
58	missMethyl: an R package for analyzing data from Illumina's HumanMethylation450 platform. <i>Bioinformatics</i> , 2016, 32, 286-288.	1.8	573
59	A cross-package Bioconductor workflow for analysing methylation array data. <i>F1000Research</i> , 2016, 5, 1281.	0.8	93
60	A cross-package Bioconductor workflow for analysing methylation array data. <i>F1000Research</i> , 2016, 5, 1281.	0.8	103
61	The Dose-Dependent Effects of Microna-155 in Acute Myeloid Leukemia. <i>Blood</i> , 2016, 128, 2841-2841.	0.6	0
62	Genotyping microsatellites in next-generation sequencing data. <i>BMC Bioinformatics</i> , 2015, 16, .	1.2	9
63	Jarid2 regulates hematopoietic stem cell function by acting with polycomb repressive complex 2. <i>Blood</i> , 2015, 125, 1890-1900.	0.6	41
64	Identification of candidate gonadal sex differentiation genes in the chicken embryo using RNA-seq. <i>BMC Genomics</i> , 2015, 16, 704.	1.2	54
65	JAFFA: High sensitivity transcriptome-focused fusion gene detection. <i>Genome Medicine</i> , 2015, 7, 43.	3.6	132
66	Cpipe: a shared variant detection pipeline designed for diagnostic settings. <i>Genome Medicine</i> , 2015, 7, 68.	3.6	78
67	Removing unwanted variation in a differential methylation analysis of Illumina HumanMethylation450 array data. <i>Nucleic Acids Research</i> , 2015, 43, e106-e106.	6.5	73
68	Purification and Transcriptomic Analysis of Mouse Fetal Leydig Cells Reveals Candidate Genes for Specification of Gonadal Steroidogenic Cells1. <i>Biology of Reproduction</i> , 2015, 92, 145.	1.2	51
69	DiffVar: a new method for detecting differential variability with application to methylation in cancer and aging. <i>Genome Biology</i> , 2014, 15, 465.	3.8	84
70	A comparison of control samples for CHIP-seq of histone modifications. <i>Frontiers in Genetics</i> , 2014, 5, 329.	1.1	31
71	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
72	Identification, Expression, and Regulation of Anti-Müllerian Hormone Type-II Receptor in the Embryonic Chicken Gonad1. <i>Biology of Reproduction</i> , 2014, 90, 106.	1.2	28

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73	Corset: enabling differential gene expression analysis for de novo assembled transcriptomes. <i>Genome Biology</i> , 2014, 15, 410.	3.8	427
74	The polycomb repressive complex 2 governs life and death of peripheral T cells. <i>Blood</i> , 2014, 124, 737-749.	0.6	111
75	RNA sequencing reveals sexually dimorphic gene expression before gonadal differentiation in chicken and allows comprehensive annotation of the W-chromosome. <i>Genome Biology</i> , 2013, 14, R26.	13.9	98
76	Condensin I associates with structural and gene regulatory regions in vertebrate chromosomes. <i>Nature Communications</i> , 2013, 4, 2537.	5.8	70
77	Sixty years of genome biology. <i>Genome Biology</i> , 2013, 14, 113.	13.9	6
78	A 10-step guide to party conversation for bioinformaticians. <i>Genome Biology</i> , 2013, 14, 104.	13.9	2
79	Identification of recurrent <i>FGFR3</i> fusion genes in lung cancer through RNA sequencing. <i>Journal of Pathology</i> , 2013, 230, 270-276.	2.1	113
80	Analysis of epigenetic changes in survivors of preterm birth reveals the effect of gestational age and evidence for a long term legacy. <i>Genome Medicine</i> , 2013, 5, 96.	3.6	101
81	miRNA-Seq normalization comparisons need improvement. <i>Rna</i> , 2013, 19, 733-734.	1.6	22
82	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
83	Genome-wide DNA methylation analysis identifies hypomethylated genes regulated by FOXP3 in human regulatory T cells. <i>Blood</i> , 2013, 122, 2823-2836.	0.6	114
84	Bpipe: a tool for running and managing bioinformatics pipelines. <i>Bioinformatics</i> , 2012, 28, 1525-1526.	1.8	145
85	SWAN: Subset-quantile Within Array Normalization for Illumina Infinium HumanMethylation450 BeadChips. <i>Genome Biology</i> , 2012, 13, R44.	13.9	697
86	Differential Expression for RNA Sequencing (RNA-Seq) Data: Mapping, Summarization, Statistical Analysis, and Experimental Design. , 2012, , 169-190.		4
87	As we come to the end of 2011, several members of the Genome Biology Editorial Board give their views on the state of play in genomics. <i>Genome Biology</i> , 2011, 12, 137.	13.9	0
88	Expression discordance of monozygotic twins at birth: Effect of intrauterine environment and a possible mechanism for fetal programming. <i>Epigenetics</i> , 2011, 6, 579-592.	1.3	70
89	ChIP-seq analysis reveals distinct H3K27me3 profiles that correlate with transcriptional activity. <i>Nucleic Acids Research</i> , 2011, 39, 7415-7427.	6.5	250
90	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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91	A scaling normalization method for differential expression analysis of RNA-seq data. <i>Genome Biology</i> , 2010, 11, R25.	13.9	6,234
92	From RNA-seq reads to differential expression results. <i>Genome Biology</i> , 2010, 11, 220.	13.9	603
93	Gene ontology analysis for RNA-seq: accounting for selection bias. <i>Genome Biology</i> , 2010, 11, R14.	13.9	5,824
94	Segmental Duplications Contribute to Gene Expression Differences Between Humans and Chimpanzees. <i>Genetics</i> , 2009, 182, 627-630.	1.2	26
95	Transcript length bias in RNA-seq data confounds systems biology. <i>Biology Direct</i> , 2009, 4, 14.	1.9	422
96	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
97	Gene Regulation in Primates Evolves under Tissue-Specific Selection Pressures. <i>PLoS Genetics</i> , 2008, 4, e1000271.	1.5	143
98	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
99	A comparison of background correction methods for two-colour microarrays. <i>Bioinformatics</i> , 2007, 23, 2700-2707.	1.8	829
100	Using DNA microarrays to study gene expression in closely related species. <i>Bioinformatics</i> , 2007, 23, 1235-1242.	1.8	60
101	Normalization of boutique two-color microarrays with a high proportion of differentially expressed probes. <i>Genome Biology</i> , 2007, 8, R2.	13.9	61
102	The spectral energy distribution of PKS 2004-447: a compact steep-spectrum source and possible radio-loud narrow-line Seyfert 1 galaxy. <i>Monthly Notices of the Royal Astronomical Society</i> , 2006, 370, 245-254.	1.6	54
103	Expression profiling in primates reveals a rapid evolution of human transcription factors. <i>Nature</i> , 2006, 440, 242-245.	13.7	283
104	Natural selection on gene expression. <i>Trends in Genetics</i> , 2006, 22, 456-461.	2.9	187
105	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
106	THE NATURE OF THE OPTICAL EMISSION IN RADIO-SELECTED AGN. , 2004, , .		0
107	Near Infrared Micro-variability of Radio-loud Quasars. <i>Publications of the Astronomical Society of Australia</i> , 2002, 19, 222-227.	1.3	3
108	Black Hole Mass Estimates of Radio-selected Quasars. <i>Astrophysical Journal</i> , 2002, 576, 81-88.	1.6	66

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109	A Very Radio Loud Narrow-Line Seyfert 1: PKS 2004+47. <i>Astrophysical Journal</i> , 2001, 558, 578-582.	1.6	72
110	A cross-package Bioconductor workflow for analysing methylation array data. <i>F1000Research</i> , 0, 5, 1281.	0.8	97