

Aaron R Quinlan

List of Publications by Year in descending order

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Version: 2024-02-01

78
papers

34,273
citations

76196

40
h-index

76769

74
g-index

113
all docs

113
docs citations

113
times ranked

62727
citing authors

#	ARTICLE	IF	CITATIONS
1	Comprehensive variant calling from whole-genome sequencing identifies a complex inversion that disrupts <i>ZFPM2</i> in familial congenital diaphragmatic hernia. <i>Molecular Genetics & Genomic Medicine</i> , 2022, 10, e1888.	0.6	6
2	Searching thousands of genomes to classify somatic and novel structural variants using STIX. <i>Nature Methods</i> , 2022, 19, 445-448.	9.0	8
3	trfermikit: a tool to discover VNTR-associated deletions. <i>Bioinformatics</i> , 2022, 38, 1231-1234.	1.8	0
4	OncoGEMINI: software for investigating tumor variants from multiple biopsies with integrated cancer annotations. <i>Genome Medicine</i> , 2021, 13, 46.	3.6	0
5	Go Get Data (GCD) is a framework that facilitates reproducible access to genomic data. <i>Nature Communications</i> , 2021, 12, 2151.	5.8	9
6	CaBagE: A Cas9-based Background Elimination strategy for targeted, long-read DNA sequencing. <i>PLoS ONE</i> , 2021, 16, e0241253.	1.1	17
7	De novo structural mutation rates and gamete-of-origin biases revealed through genome sequencing of 2,396 families. <i>American Journal of Human Genetics</i> , 2021, 108, 597-607.	2.6	57
8	Samplot: a platform for structural variant visual validation and automated filtering. <i>Genome Biology</i> , 2021, 22, 161.	3.8	52
9	Detection of a mosaic <i>CDKL5</i> deletion and inversion by optical genome mapping ends an exhaustive diagnostic odyssey. <i>Molecular Genetics & Genomic Medicine</i> , 2021, 9, e1665.	0.6	11
10	Unfazed: parent-of-origin detection for large and small <i>de novo</i> variants. <i>Bioinformatics</i> , 2021, 37, 4860-4861.	1.8	4
11	Balancing efficient analysis and storage of quantitative genomics data with the D4 format and d4tools. <i>Nature Computational Science</i> , 2021, 1, 441-447.	3.8	4
12	Genes affecting ionizing radiation survival identified through combined exome sequencing and functional screening. <i>Human Mutation</i> , 2021, 42, 1124-1138.	1.1	0
13	Effective variant filtering and expected candidate variant yield in studies of rare human disease. <i>Npj Genomic Medicine</i> , 2021, 6, 60.	1.7	51
14	A Systematic Review to Guide Future Efforts in the Determination of Genetic Causes of Pregnancy Loss. <i>Frontiers in Reproductive Health</i> , 2021, 3, .	0.6	1
15	Somalier: rapid relatedness estimation for cancer and germline studies using efficient genome sketches. <i>Genome Medicine</i> , 2020, 12, 62.	3.6	48
16	Regulatory sharing between estrogen receptor $\hat{\pm}$ bound enhancers. <i>Nucleic Acids Research</i> , 2020, 48, 6597-6610.	6.5	7
17	Germline mutation rates in young adults predict longevity and reproductive lifespan. <i>Scientific Reports</i> , 2020, 10, 10001.	1.6	16
18	De novo EIF2AK1 and EIF2AK2 Variants Are Associated with Developmental Delay, Leukoencephalopathy, and Neurologic Decompensation. <i>American Journal of Human Genetics</i> , 2020, 106, 570-583.	2.6	37

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19	XPRESSyourself: Enhancing, standardizing, and automating ribosome profiling computational analyses yields improved insight into data. PLoS Computational Biology, 2020, 16, e1007625.	1.5	15
20	Title is missing!. , 2020, 16, e1007625.		0
21	Title is missing!. , 2020, 16, e1007625.		0
22	Title is missing!. , 2020, 16, e1007625.		0
23	Title is missing!. , 2020, 16, e1007625.		0
24	De Novo Pathogenic Variants in N-cadherin Cause a Syndromic Neurodevelopmental Disorder with Corpus Callosum, Axon, Cardiac, Ocular, and Genital Defects. American Journal of Human Genetics, 2019, 105, 854-868.	2.6	29
25	Duphold: scalable, depth-based annotation and curation of high-confidence structural variant calls. GigaScience, 2019, 8, .	3.3	45
26	Overlooked roles of DNA damage and maternal age in generating human germline mutations. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 9491-9500.	3.3	155
27	Coexpression patterns define epigenetic regulators associated with neurological dysfunction. Genome Research, 2019, 29, 532-542.	2.4	42
28	A map of constrained coding regions in the human genome. Nature Genetics, 2019, 51, 88-95.	9.4	201
29	Large, three-generation human families reveal post-zygotic mosaicism and variability in germline mutation accumulation. ELife, 2019, 8, .	2.8	116
30	Nanopore sequencing and assembly of a human genome with ultra-long reads. Nature Biotechnology, 2018, 36, 338-345.	9.4	1,443
31	Mosdepth: quick coverage calculation for genomes and exomes. Bioinformatics, 2018, 34, 867-868.	1.8	638
32	GIGGLE: a search engine for large-scale integrated genome analysis. Nature Methods, 2018, 15, 123-126.	9.0	154
33	An analytical framework for whole-genome sequence association studies and its implications for autism spectrum disorder. Nature Genetics, 2018, 50, 727-736.	9.4	235
34	hts-nim: scripting high-performance genomic analyses. Bioinformatics, 2018, 34, 3387-3389.	1.8	28
35	Identification of ATIC as a Novel Target for Chemoradiosensitization. International Journal of Radiation Oncology Biology Physics, 2018, 100, 162-173.	0.4	22
36	Genome-wide de novo risk score implicates promoter variation in autism spectrum disorder. Science, 2018, 362, .	6.0	234

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37	SV-plaudit: A cloud-based framework for manually curating thousands of structural variants. <i>GigaScience</i> , 2018, 7, .	3.3	30
38	Fine-mapping and functional studies highlight potential causal variants for rheumatoid arthritis and type 1 diabetes. <i>Nature Genetics</i> , 2018, 50, 1366-1374.	9.4	122
39	Whole-genome analysis for effective clinical diagnosis and gene discovery in early infantile epileptic encephalopathy. <i>Npj Genomic Medicine</i> , 2018, 3, 22.	1.7	64
40	Coloc-stats: a unified web interface to perform colocalization analysis of genomic features. <i>Nucleic Acids Research</i> , 2018, 46, W186-W193.	6.5	23
41	Long read sequencing reveals poxvirus evolution through rapid homogenization of gene arrays. <i>ELife</i> , 2018, 7, .	2.8	23
42	Who's Who? Detecting and Resolving Sample Anomalies in Human DNA Sequencing Studies with Peddy. <i>American Journal of Human Genetics</i> , 2017, 100, 406-413.	2.6	173
43	cyvcf2: fast, flexible variant analysis with Python. <i>Bioinformatics</i> , 2017, 33, 1867-1869.	1.8	66
44	Combating subclonal evolution of resistant cancer phenotypes. <i>Nature Communications</i> , 2017, 8, 1231.	5.8	124
45	Settling the score: variant prioritization and Mendelian disease. <i>Nature Reviews Genetics</i> , 2017, 18, 599-612.	7.7	213
46	Indexcov: fast coverage quality control for whole-genome sequencing. <i>GigaScience</i> , 2017, 6, 1-6.	3.3	36
47	Vcfanno: fast, flexible annotation of genetic variants. <i>Genome Biology</i> , 2016, 17, 118.	3.8	157
48	Targeted Deep Sequencing in Multiple-Affected Sibships of European Ancestry Identifies Rare Deleterious Variants in <i>PTPN22</i> That Confer Risk for Type 1 Diabetes. <i>Diabetes</i> , 2016, 65, 794-802.	0.3	24
49	Efficient genotype compression and analysis of large genetic-variation data sets. <i>Nature Methods</i> , 2016, 13, 63-65.	9.0	57
50	Fine mapping of type 1 diabetes susceptibility loci and evidence for colocalization of causal variants with lymphoid gene enhancers. <i>Nature Genetics</i> , 2015, 47, 381-386.	9.4	589
51	A Parallel Algorithm for N^2 -Way Interval Set Intersection. <i>Proceedings of the IEEE</i> , 2015, 105, 1-10.	16.4	3
52	Cas9-chromatin binding information enables more accurate CRISPR off-target prediction. <i>Nucleic Acids Research</i> , 2015, 43, e118-e118.	6.5	187
53	Extending reference assembly models. <i>Genome Biology</i> , 2015, 16, 13.	3.8	139
54	Rare and Coding Region Genetic Variants Associated With Risk of Ischemic Stroke. <i>JAMA Neurology</i> , 2015, 72, 781.	4.5	49

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55	SpeedSeq: ultra-fast personal genome analysis and interpretation. <i>Nature Methods</i> , 2015, 12, 966-968.	9.0	515
56	Population-based structural variation discovery with Hydra-Multi. <i>Bioinformatics</i> , 2015, 31, 1286-1289.	1.8	19
57	A Novel <i>IFITM5</i> Mutation in Severe Atypical Osteogenesis Imperfecta Type VI Impairs Osteoblast Production of Pigment Epithelium-Derived Factor. <i>Journal of Bone and Mineral Research</i> , 2014, 29, 1402-1411.	3.1	63
58	Genetics of systemic lupus erythematosus: immune responses and end organ resistance to damage. <i>Current Opinion in Immunology</i> , 2014, 31, 87-96.	2.4	47
59	Poretools: a toolkit for analyzing nanopore sequence data. <i>Bioinformatics</i> , 2014, 30, 3399-3401.	1.8	404
60	A reference bacterial genome dataset generated on the MinION, a portable single-molecule nanopore sequencer. <i>GigaScience</i> , 2014, 3, 22.	3.3	208
61	BEDTools: The Swiss Army Tool for Genome Feature Analysis. <i>Current Protocols in Bioinformatics</i> , 2014, 47, 11.12.1-34.	25.8	1,836
62	LUMPY: a probabilistic framework for structural variant discovery. <i>Genome Biology</i> , 2014, 15, R84.	13.9	1,199
63	Binary Interval Search: a scalable algorithm for counting interval intersections. <i>Bioinformatics</i> , 2013, 29, 1-7.	1.8	58
64	Breakpoint profiling of 64 cancer genomes reveals numerous complex rearrangements spawned by homology-independent mechanisms. <i>Genome Research</i> , 2013, 23, 762-776.	2.4	155
65	GEMINI: Integrative Exploration of Genetic Variation and Genome Annotations. <i>PLoS Computational Biology</i> , 2013, 9, e1003153.	1.5	377
66	Copy number variation detection and genotyping from exome sequence data. <i>Genome Research</i> , 2012, 22, 1525-1532.	2.4	550
67	Detection and Interpretation of Genomic Structural Variation in Mammals. <i>Methods in Molecular Biology</i> , 2012, 838, 225-248.	0.4	14
68	Characterizing complex structural variation in germline and somatic genomes. <i>Trends in Genetics</i> , 2012, 28, 43-53.	2.9	93
69	Genome Sequencing of Mouse Induced Pluripotent Stem Cells Reveals Retroelement Stability and Infrequent DNA Rearrangement during Reprogramming. <i>Cell Stem Cell</i> , 2011, 9, 366-373.	5.2	102
70	BamTools: a C++ API and toolkit for analyzing and managing BAM files. <i>Bioinformatics</i> , 2011, 27, 1691-1692.	1.8	843
71	Pybedtools: a flexible Python library for manipulating genomic datasets and annotations. <i>Bioinformatics</i> , 2011, 27, 3423-3424.	1.8	402
72	Genome-wide mapping and assembly of structural variant breakpoints in the mouse genome. <i>Genome Research</i> , 2010, 20, 623-635.	2.4	257

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73	BEDTools: a flexible suite of utilities for comparing genomic features. <i>Bioinformatics</i> , 2010, 26, 841-842.	1.8	20,367
74	Population Genomic Inferences from Sparse High-Throughput Sequencing of Two Populations of <i>Drosophila melanogaster</i> . <i>Genome Biology and Evolution</i> , 2009, 1, 449-465.	1.1	60
75	Pyrobayes: an improved base caller for SNP discovery in pyrosequences. <i>Nature Methods</i> , 2008, 5, 179-181.	9.0	180
76	Whole-genome sequencing and variant discovery in <i>C. elegans</i> . <i>Nature Methods</i> , 2008, 5, 183-188.	9.0	380
77	Rapid whole-genome mutational profiling using next-generation sequencing technologies. <i>Genome Research</i> , 2008, 18, 1638-1642.	2.4	225
78	Primer-site SNPs mask mutations. <i>Nature Methods</i> , 2007, 4, 192-192.	9.0	29