Aaron R Quinlan

List of Publications by Year
in descending order

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78 papers	34,273 citations	76326 40 h-index	76900 74 g-index
113 all docs	113 docs citations	113 times ranked	62727 citing authors

#	Article	IF	Citations
1	BEDTools: a flexible suite of utilities for comparing genomic features. Bioinformatics, 2010, 26, 841-842.	4.1	20,367
2	BEDTools: The Swissâ€Army Tool for Genome Feature Analysis. Current Protocols in Bioinformatics, 2014, 47, 11.12.1-34.	25.8	1,836
3	Nanopore sequencing and assembly of a human genome with ultra-long reads. Nature Biotechnology, 2018, 36, 338-345.	17.5	1,443
4	LUMPY: a probabilistic framework for structural variant discovery. Genome Biology, 2014, 15, R84.	9.6	1,199
5	BamTools: a C++ API and toolkit for analyzing and managing BAM files. Bioinformatics, 2011, 27, 1691-1692.	4.1	843
6	Mosdepth: quick coverage calculation for genomes and exomes. Bioinformatics, 2018, 34, 867-868.	4.1	638
7	Fine mapping of type 1 diabetes susceptibility loci and evidence for colocalization of causal variants with lymphoid gene enhancers. Nature Genetics, 2015, 47, 381-386.	21.4	589
8	Copy number variation detection and genotyping from exome sequence data. Genome Research, 2012, 22, 1525-1532.	5.5	550
9	SpeedSeq: ultra-fast personal genome analysis and interpretation. Nature Methods, 2015, 12, 966-968.	19.0	515
10	Poretools: a toolkit for analyzing nanopore sequence data. Bioinformatics, 2014, 30, 3399-3401.	4.1	404
11	Pybedtools: a flexible Python library for manipulating genomic datasets and annotations. Bioinformatics, 2011, 27, 3423-3424.	4.1	402
12	Whole-genome sequencing and variant discovery in C. elegans. Nature Methods, 2008, 5, 183-188.	19.0	380
13	GEMINI: Integrative Exploration of Genetic Variation and Genome Annotations. PLoS Computational Biology, 2013, 9, e1003153.	3.2	377
14	Genome-wide mapping and assembly of structural variant breakpoints in the mouse genome. Genome Research, 2010, 20, 623-635.	5.5	257
15	An analytical framework for whole-genome sequence association studies and its implications for autism spectrum disorder. Nature Genetics, 2018, 50, 727-736.	21.4	235
16	Genome-wide de novo risk score implicates promoter variation in autism spectrum disorder. Science, 2018, 362, .	12.6	234
17	Rapid whole-genome mutational profiling using next-generation sequencing technologies. Genome Research, 2008, 18, 1638-1642.	5.5	225
18	Settling the score: variant prioritization and Mendelian disease. Nature Reviews Genetics, 2017, 18, 599-612.	16.3	213

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19	A reference bacterial genome dataset generated on the MinIONâ„¢ portable single-molecule nanopore sequencer. GigaScience, 2014, 3, 22.	6.4	208
20	A map of constrained coding regions in the human genome. Nature Genetics, 2019, 51, 88-95.	21.4	201
21	Cas9-chromatin binding information enables more accurate CRISPR off-target prediction. Nucleic Acids Research, 2015, 43, e118-e118.	14.5	187
22	Pyrobayes: an improved base caller for SNP discovery in pyrosequences. Nature Methods, 2008, 5, 179-181.	19.0	180
23	Whoâ€™s Who? Detecting and Resolving Sample Anomalies in Human DNA Sequencing Studies with Peddy. American Journal of Human Genetics, 2017, 100, 406-413.	6.2	173
24	Vcfanno: fast, flexible annotation of genetic variants. Genome Biology, 2016, 17, 118.	8.8	157
25	Breakpoint profiling of 64 cancer genomes reveals numerous complex rearrangements spawned by homology-independent mechanisms. Genome Research, 2013, 23, 762-776.	5.5	155
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.	7.1	155
27	GIGGLE: a search engine for large-scale integrated genome analysis. Nature Methods, 2018, 15, 123-126.	19.0	154
28	Extending reference assembly models. Genome Biology, 2015, 16, 13.	8.8	139
29	Combating subclonal evolution of resistant cancer phenotypes. Nature Communications, 2017, 8, 1231.	12.8	124
30	Fine-mapping and functional studies highlight potential causal variants for rheumatoid arthritis and type 1 diabetes. Nature Genetics, 2018, 50, 1366-1374.	21.4	122
31	Large, three-generation human families reveal post-zygotic mosaicism and variability in germline mutation accumulation. ELife, 2019, 8, .	6.0	116
32	Genome Sequencing of Mouse Induced Pluripotent Stem Cells Reveals Retroelement Stability and Infrequent DNA Rearrangement during Reprogramming. Cell Stem Cell, 2011, 9, 366-373.	11.1	102
33	Characterizing complex structural variation in germline and somatic genomes. Trends in Genetics, 2012, 28, 43-53.	6.7	93
34	cyvcf2: fast, flexible variant analysis with Python. Bioinformatics, 2017, 33, 1867-1869.	4.1	66
35	Whole-genome analysis for effective clinical diagnosis and gene discovery in early infantile epileptic encephalopathy. Npj Genomic Medicine, 2018, 3, 22.	3.8	64
36	A Novel IFITM5 Mutation in Severe Atypical Osteogenesis Imperfecta Type VI Impairs Osteoblast Production of Pigment Epithelium-Derived Factor. Journal of Bone and Mineral Research, 2014, 29, 1402-1411.	2.8	63

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37	Population Genomic Inferences from Sparse High-Throughput Sequencing of Two Populations of Drosophila melanogaster. Genome Biology and Evolution, 2009, 1, 449-465.	2.5	60
38	Binary Interval Search: a scalable algorithm for counting interval intersections. Bioinformatics, 2013, 29, 1-7.	4.1	58
39	Efficient genotype compression and analysis of large genetic-variation data sets. Nature Methods, 2016, 13, 63-65.	19.0	57
40	De novo structural mutation rates and gamete-of-origin biases revealed through genome sequencing of 2,396 families. American Journal of Human Genetics, 2021, 108, 597-607.	6.2	57
41	Samplot: a platform for structural variant visual validation and automated filtering. Genome Biology, 2021, 22, 161.	8.8	52
42	Effective variant filtering and expected candidate variant yield in studies of rare human disease. Npj Genomic Medicine, 2021, 6, 60.	3.8	51
43	Rare and Coding Region Genetic Variants Associated With Risk of Ischemic Stroke. JAMA Neurology, 2015, 72, 781.	9.0	49
44	Somalier: rapid relatedness estimation for cancer and germline studies using efficient genome sketches. Genome Medicine, 2020, 12, 62.	8.2	48
45	Genetics of systemic lupus erythematosus: immune responses and end organ resistance to damage. Current Opinion in Immunology, 2014, 31, 87-96.	5.5	47
46	Duphold: scalable, depth-based annotation and curation of high-confidence structural variant calls. GigaScience, 2019, 8, .	6.4	45
47	Coexpression patterns define epigenetic regulators associated with neurological dysfunction. Genome Research, 2019, 29, 532-542.	5.5	42
48	De novo EIF2AK1 and EIF2AK2 Variants Are Associated with Developmental Delay, Leukoencephalopathy, and Neurologic Decompensation. American Journal of Human Genetics, 2020, 106, 570-583.	6.2	37
49	Indexcov: fast coverage quality control for whole-genome sequencing. GigaScience, 2017, 6, 1-6.	6.4	36
50	SV-plaudit: A cloud-based framework for manually curating thousands of structural variants. GigaScience, 2018, 7, .	6.4	30
51	Primer-site SNPs mask mutations. Nature Methods, 2007, 4, 192-192.	19.0	29
52	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.	6.2	29
53	hts-nim: scripting high-performance genomic analyses. Bioinformatics, 2018, 34, 3387-3389.	4.1	28
54	Targeted Deep Sequencing in Multiple-Affected Sibships of European Ancestry Identifies Rare Deleterious Variants in PTPN22 That Confer Risk for Type 1 Diabetes. Diabetes, 2016, 65, 794-802.	0.6	24

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55	Coloc-stats: a unified web interface to perform colocalization analysis of genomic features. Nucleic Acids Research, 2018, 46, W186-W193.	14.5	23
56	Long read sequencing reveals poxvirus evolution through rapid homogenization of gene arrays. ELife, 2018, 7, .	6.0	23
57	Identification of ATIC as a Novel Target for Chemoradiosensitization. International Journal of Radiation Oncology Biology Physics, 2018, 100, 162-173.	0.8	22
58	Population-based structural variation discovery with Hydra-Multi. Bioinformatics, 2015, 31, 1286-1289.	4.1	19
59	CaBagE: A Cas9-based Background Elimination strategy for targeted, long-read DNA sequencing. PLoS ONE, 2021, 16, e0241253.	2.5	17
60	Germline mutation rates in young adultsÂpredict longevity and reproductive lifespan. Scientific Reports, 2020, 10, 10001.	3.3	16
61	XPRESSyourself: Enhancing, standardizing, and automating ribosome profiling computational analyses yields improved insight into data. PLoS Computational Biology, 2020, 16, e1007625.	3.2	15
62	Detection and Interpretation of Genomic Structural Variation in Mammals. Methods in Molecular Biology, 2012, 838, 225-248.	0.9	14
63	Detection of a mosaic CDKL5 deletion and inversion by optical genome mapping ends an exhaustive diagnostic odyssey. Molecular Genetics & Genomic Medicine, 2021, 9, e1665.	1.2	11
64	Go Get Data (GGD) is a framework that facilitates reproducible access to genomic data. Nature Communications, 2021, 12, 2151.	12.8	9
65	Searching thousands of genomes to classify somatic and novel structural variants using STIX. Nature Methods, 2022, 19, 445-448.	19.0	8
66	Regulatory sharing between estrogen receptor Î± bound enhancers. Nucleic Acids Research, 2020, 48, 6597-6610.	14.5	7
67	Comprehensive variant calling from wholeâ€genome sequencing identifies a complex inversion that disrupts <scp>ZFPM2</scp> in familial congenital diaphragmatic hernia. Molecular Genetics & Genomic Medicine, 2022, 10, e1888.	1.2	6
68	Unfazed: parent-of-origin detection for large and small de novo variants. Bioinformatics, 2021, 37, 4860-4861.	4.1	4
69	Balancing efficient analysis and storage of quantitative genomics data with the D4 format and d4tools. Nature Computational Science, 2021, 1, 441-447.	8.0	4
70	A Parallel Algorithm for <formula><tex>N</tex></formula>-Way Interval Set Intersection. Proceedings of the IEEE, 2015, 105, 1-10.	21.3	3
71	A Systematic Review to Guide Future Efforts in the Determination of Genetic Causes of Pregnancy Loss. Frontiers in Reproductive Health, 2021, 3, .	1.9	1
72	OncoGEMINI: software for investigating tumor variants from multiple biopsies with integrated cancer annotations. Genome Medicine, 2021, 13, 46.	8.2	0

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73	Genes affecting ionizing radiation survival identified through combined exome sequencing and functional screening. Human Mutation, 2021, 42, 1124-1138.	2.5	0
74	trfermikit: a tool to discover VNTR-associated deletions. Bioinformatics, 2022, 38, 1231-1234.	4.1	0
75	Title is missing!. , 2020, 16, e1007625.		0
76	Title is missing!. , 2020, 16, e1007625.		0
77	Title is missing!. , 2020, 16, e1007625.		0
78	Title is missing!. , 2020, 16, e1007625.		0