Aaron R Quinlan

List of Publications by Year
in descending order

78	34,273	87401 40	87275 74
papers	citations	h-index	g-index

113	113	113	69294
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	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.	0.6	6
2	Searching thousands of genomes to classify somatic and novel structural variants using STIX. Nature Methods, 2022, 19, 445-448.	9.0	8
3	trfermikit: a tool to discover VNTR-associated deletions. Bioinformatics, 2022, 38, 1231-1234.	1.8	0
4	OncoGEMINI: software for investigating tumor variants from multiple biopsies with integrated cancer annotations. Genome Medicine, 2021, 13, 46.	3.6	0
5	Go Get Data (GGD) is a framework that facilitates reproducible access to genomic data. Nature Communications, 2021, 12, 2151.	5.8	9
6	CaBagE: A Cas9-based Background Elimination strategy for targeted, long-read DNA sequencing. PLoS ONE, 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. American Journal of Human Genetics, 2021, 108, 597-607.	2.6	57
8	Samplot: a platform for structural variant visual validation and automated filtering. Genome Biology, 2021, 22, 161.	3.8	52
9	Detection of a mosaic CDKL5 deletion and inversion by optical genome mapping ends an exhaustive diagnostic odyssey. Molecular Genetics & Genomic Medicine, 2021, 9, e1665.	0.6	11
10	Unfazed: parent-of-origin detection for large and small de novo variants. Bioinformatics, 2021, 37, 4860-4861.	1.8	4
11	Balancing efficient analysis and storage of quantitative genomics data with the D4 format and d4tools. Nature Computational Science, 2021, 1, 441-447.	3.8	4
12	Genes affecting ionizing radiation survival identified through combined exome sequencing and functional screening. Human Mutation, 2021, 42, 1124-1138.	1.1	0
13	Effective variant filtering and expected candidate variant yield in studies of rare human disease. Npj Genomic Medicine, 2021, 6, 60.	1.7	51
14	A Systematic Review to Guide Future Efforts in the Determination of Genetic Causes of Pregnancy Loss. Frontiers in Reproductive Health, 2021, 3, .	0.6	1
15	Somalier: rapid relatedness estimation for cancer and germline studies using efficient genome sketches. Genome Medicine, 2020, 12, 62.	3.6	48
16	Regulatory sharing between estrogen receptor Î± bound enhancers. Nucleic Acids Research, 2020, 48, 6597-6610.	6.5	7
17	Germline mutation rates in young adultsÂpredict longevity and reproductive lifespan. Scientific Reports, 2020, 10, 10001.	1.6	16
18	De novo EIF2AK1 and EIF2AK2 Variants Are Associated with Developmental Delay, Leukoencephalopathy, and Neurologic Decompensation. American Journal of Human Genetics, 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. GigaScience, 2018, 7, .	3.3	30
38	Fine-mapping and functional studies highlight potential causal variants for rheumatoid arthritis and type 1 diabetes. Nature Genetics, 2018, 50, 1366-1374.	9.4	122
39	Whole-genome analysis for effective clinical diagnosis and gene discovery in early infantile epileptic encephalopathy. Npj Genomic Medicine, 2018, 3, 22.	1.7	64
40	Coloc-stats: a unified web interface to perform colocalization analysis of genomic features. Nucleic Acids Research, 2018, 46, W186-W193.	6.5	23
41	Long read sequencing reveals poxvirus evolution through rapid homogenization of gene arrays. ELife, 2018, 7, .	2.8	23
42	Whoâ€™s Who? Detecting and Resolving Sample Anomalies in Human DNA Sequencing Studies with Peddy. American Journal of Human Genetics, 2017, 100, 406-413.	2.6	173
43	cyvcf2: fast, flexible variant analysis with Python. Bioinformatics, 2017, 33, 1867-1869.	1.8	66
44	Combating subclonal evolution of resistant cancer phenotypes. Nature Communications, 2017, 8, 1231.	5.8	124
45	Settling the score: variant prioritization and Mendelian disease. Nature Reviews Genetics, 2017, 18, 599-612.	7.7	213
46	Indexcov: fast coverage quality control for whole-genome sequencing. GigaScience, 2017, 6, 1-6.	3.3	36
47	Vcfanno: fast, flexible annotation of genetic variants. Genome Biology, 2016, 17, 118.	3.8	157
48	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.	0.3	24
49	Efficient genotype compression and analysis of large genetic-variation data sets. Nature Methods, 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. Nature Genetics, 2015, 47, 381-386.	9.4	589
51	A Parallel Algorithm for <formula><tex>N</tex></formula>-Way Interval Set Intersection. Proceedings of the IEEE, 2015, 105, 1-10.	16.4	3
52	Cas9-chromatin binding information enables more accurate CRISPR off-target prediction. Nucleic Acids Research, 2015, 43, e118-e118.	6.5	187
53	Extending reference assembly models. Genome Biology, 2015, 16, 13.	3.8	139
54	Rare and Coding Region Genetic Variants Associated With Risk of Ischemic Stroke. JAMA Neurology, 2015, 72, 781.	4.5	49

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

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