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List of Publications by Year
in descending order

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74 papers	36,976 citations	57631 44 h-index	79541 73 g-index
91 all docs	91 docs citations	91 times ranked	60638 citing authors

#	Article	IF	Citations
1	The variant call format and VCFtools. Bioinformatics, 2011, 27, 2156-2158.	1.8	11,326
2	A map of human genome variation from population-scale sequencing. Nature, 2010, 467, 1061-1073.	13.7	7,209
3	An integrated map of genetic variation from 1,092 human genomes. Nature, 2012, 491, 56-65.	13.7	7,199
4	A Systematic Survey of Loss-of-Function Variants in Human Protein-Coding Genes. Science, 2012, 335, 823-828.	6.0	1,095
5	Stampy: A statistical algorithm for sensitive and fast mapping of Illumina sequence reads. Genome Research, 2011, 21, 936-939.	2.4	1,044
6	Integrating mapping-, assembly- and haplotype-based approaches for calling variants in clinical sequencing applications. Nature Genetics, 2014, 46, 912-918.	9.4	937
7	Insights into hominid evolution from the gorilla genome sequence. Nature, 2012, 483, 169-175.	13.7	663
8	Functionality or transcriptional noise? Evidence for selection within long noncoding RNAs. Genome Research, 2007, 17, 556-565.	2.4	632
9	Comparative and demographic analysis of orang-utan genomes. Nature, 2011, 469, 529-533.	13.7	541
10	The bonobo genome compared with the chimpanzee and human genomes. Nature, 2012, 486, 527-531.	13.7	445
11	The Human Pancreatic Islet Transcriptome: Expression of Candidate Genes for Type 1 Diabetes and the Impact of Pro-Inflammatory Cytokines. PLoS Genetics, 2012, 8, e1002552.	1.5	398
12	Dindel: Accurate indel calls from short-read data. Genome Research, 2011, 21, 961-973.	2.4	383
13	Genomic and Transcriptional Co-Localization of Protein-Coding and Long Non-Coding RNA Pairs in the Developing Brain. PLoS Genetics, 2009, 5, e1000617.	1.5	354
14	Factors influencing success of clinical genome sequencing across a broad spectrum of disorders. Nature Genetics, 2015, 47, 717-726.	9.4	310
15	A Fine-Scale Chimpanzee Genetic Map from Population Sequencing. Science, 2012, 336, 193-198.	6.0	273
16	Accelerated Evolution of the Prdm9 Speciation Gene across Diverse Metazoan Taxa. PLoS Genetics, 2009, 5, e1000753.	1.5	256
17	GAT: a simulation framework for testing the association of genomic intervals. Bioinformatics, 2013, 29, 2046-2048.	1.8	221
18	8.2% of the Human Genome Is Constrained: Variation in Rates of Turnover across Functional Element Classes in the Human Lineage. PLoS Genetics, 2014, 10, e1004525.	1.5	213

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19	The origin, evolution, and functional impact of short insertionâ€“deletion variants identified in 179 human genomes. Genome Research, 2013, 23, 749-761.	2.4	206
20	Bayesian coestimation of phylogeny and sequence alignment. BMC Bioinformatics, 2005, 6, 83.	1.2	169
21	Genome-Wide Identification of Human Functional DNA Using a Neutral Indel Model. PLoS Computational Biology, 2006, 2, e5.	1.5	157
22	Rare variants in PPARG with decreased activity in adipocyte differentiation are associated with increased risk of type 2 diabetes. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 13127-13132.	3.3	152
23	Improved workflows for high throughput library preparation using the transposome-based nextera system. BMC Biotechnology, 2013, 13, 104.	1.7	141
24	Sequencing of human genomes with nanopore technology. Nature Communications, 2019, 10, 1869.	5.8	140
25	Novel Type IV Secretion System Involved in Propagation of Genomic Islands. Journal of Bacteriology, 2007, 189, 761-771.	1.0	138
26	Uncertainty in homology inferences: Assessing and improving genomic sequence alignment. Genome Research, 2008, 18, 298-309.	2.4	128
27	Identification of Antigen-Specific B Cell Receptor Sequences Using Public Repertoire Analysis. Journal of Immunology, 2015, 194, 252-261.	0.4	115
28	scrm: efficiently simulating long sequences using the approximated coalescent with recombination. Bioinformatics, 2015, 31, 1680-1682.	1.8	112
29	DeepC: predicting 3D genome folding using megabase-scale transfer learning. Nature Methods, 2020, 17, 1118-1124.	9.0	109
30	A "Long Indel" Model For Evolutionary Sequence Alignment. Molecular Biology and Evolution, 2003, 21, 529-540.	3.5	106
31	A New Isolation with Migration Model along Complete Genomes Infers Very Different Divergence Processes among Closely Related Great Ape Species. PLoS Genetics, 2012, 8, e1003125.	1.5	102
32	Recessive Mutations in SPTBN2 Implicate Î²-III Spectrin in Both Cognitive and Motor Development. PLoS Genetics, 2012, 8, e1003074.	1.5	94
33	In-Depth Assessment of Within-Individual and Inter-Individual Variation in the B Cell Receptor Repertoire. Frontiers in Immunology, 2015, 6, 531.	2.2	92
34	Analysis of B Cell Repertoire Dynamics Following Hepatitis B Vaccination in Humans, and Enrichment of Vaccine-specific Antibody Sequences. EBioMedicine, 2015, 2, 2070-2079.	2.7	92
35	Massive turnover of functional sequence in human and other mammalian genomes. Genome Research, 2010, 20, 1335-1343.	2.4	86
36	BCR repertoire sequencing: different patterns of Bâ€cell activation after two Meningococcal vaccines. Immunology and Cell Biology, 2015, 93, 885-895.	1.0	83

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37	Accurate clinical detection of exon copy number variants in a targeted NGS panel using DECoN. Wellcome Open Research, 2016, 1, 20.	0.9	79
38	Estimating Divergence Time and Ancestral Effective Population Size of Bornean and Sumatran Orangutan Subspecies Using a Coalescent Hidden Markov Model. PLoS Genetics, 2011, 7, e1001319.	1.5	79
39	A nucleotide substitution model with nearest-neighbour interactions. Bioinformatics, 2004, 20, i216-i223.	1.8	72
40	The Diversity and Molecular Evolution of B-Cell Receptors during Infection. Molecular Biology and Evolution, 2016, 33, 1147-1157.	3.5	72
41	Repertoire-wide phylogenetic models of B cell molecular evolution reveal evolutionary signatures of aging and vaccination. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 22664-22672.	3.3	71
42	An Efficient Algorithm for Statistical Multiple Alignment on Arbitrary Phylogenetic Trees. Journal of Computational Biology, 2003, 10, 869-889.	0.8	67
43	B-cell repertoire dynamics after sequential hepatitis B vaccination and evidence for cross-reactive B-cell activation. Genome Medicine, 2016, 8, 68.	3.6	64
44	A Phylogenetic Codon Substitution Model for Antibody Lineages. Genetics, 2017, 206, 417-427.	1.2	56
45	A unified haplotype-based method for accurate and comprehensive variant calling. Nature Biotechnology, 2021, 39, 885-892.	9.4	56
46	CSN and CAVA: variant annotation tools for rapid, robust next-generation sequencing analysis in the clinical setting. Genome Medicine, 2015, 7, 76.	3.6	54
47	Probabilistic whole-genome alignments reveal high indel rates in the human and mouse genomes. Bioinformatics, 2007, 23, i289-i296.	1.8	52
48	Signatures of adaptive evolution within human non-coding sequence. Human Molecular Genetics, 2006, 15, R170-R175.	1.4	45
49	Genome assembly quality: Assessment and improvement using the neutral indel model. Genome Research, 2010, 20, 675-684.	2.4	44
50	Unlocking the Bottleneck in Forward Genetics Using Whole-Genome Sequencing and Identity by Descent to Isolate Causative Mutations. PLoS Genetics, 2013, 9, e1003219.	1.5	44
51	Erythrocytosis associated with a novel missense mutation in the BPGM gene. Haematologica, 2014, 99, e201-e204.	1.7	35
52	HMMoCâ€”a compiler for hidden Markov models. Bioinformatics, 2007, 23, 2485-2487.	1.8	28
53	Accurate Reconstruction of Insertion-Deletion Histories by Statistical Phylogenetics. PLoS ONE, 2012, 7, e34572.	1.1	28
54	Dog as an Outgroup to Human and Mouse. PLoS Computational Biology, 2007, 3, e74.	1.5	27

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55	Resonances in a spring-pendulum: algorithms for equivariant singularity theory. Nonlinearity, 1998, 11, 1569-1605.	0.6	24
56	Simulation of Finnish Population History, Guided by Empirical Genetic Data, to Assess Power of Rare-Variant Tests in Finland. American Journal of Human Genetics, 2014, 94, 710-720.	2.6	24
57	Neuropeptide S receptor 1 is a nonhormonal treatment target in endometriosis. Science Translational Medicine, 2021, 13, .	5.8	23
58	Recurrence and survival after laparoscopy versus laparotomy without lymphadenectomy in early-stage endometrial cancer: Long-term outcomes of a randomised trial. Gynecologic Oncology, 2022, 164, 265-270.	0.6	18
59	Evolution of Primate Gene Expression: Drift and Corrective Sweeps?. Genetics, 2008, 180, 1379-1389.	1.2	17
60	Haplotype matching in large cohorts using the Li and Stephens model. Bioinformatics, 2019, 35, 798-806.	1.8	17
61	A high throughput screen for active human transposable elements. BMC Genomics, 2018, 19, 115.	1.2	14
62	Short and long-read genome sequencing methodologies for somatic variant detection; genomic analysis of a patient with diffuse large B-cell lymphoma. Scientific Reports, 2021, 11, 6408.	1.6	14
63	Respiratory Syncytial Virus, Human Metapneumovirus, and Parainfluenza Virus Infections in Lung Transplant Recipients: A Systematic Review of Outcomes and Treatment Strategies. Clinical Infectious Diseases, 2022, 74, 2252-2260.	2.9	14
64	OpEx - a validated, automated pipeline optimised for clinical exome sequence analysis. Scientific Reports, 2016, 6, 31029.	1.6	10
65	An equivariant Bayesian convolutional network predicts recombination hotspots and accurately resolves binding motifs. Bioinformatics, 2019, 35, 2177-2184.	1.8	10
66	Human brain gene wins genome race. Nature, 2006, 443, 149-150.	13.7	9
67	Investigating selection on viruses: a statistical alignment approach. BMC Bioinformatics, 2008, 9, 304.	1.2	8
68	Inferring B cell specificity for vaccines using a Bayesian mixture model. BMC Genomics, 2020, 21, 176.	1.2	8
69	New Proofs and a Generalization of Inequalities of Fan, Taussky, and Todd. Journal of Mathematical Analysis and Applications, 1994, 185, 464-476.	0.5	6
70	Multi Locus View: an extensible web-based tool for the analysis of genomic data.. Communications Biology, 2021, 4, 623.	2.0	4
71	Benchmarking small-variant genotyping in polyploids. Genome Research, 2022, 32, 403-408.	2.4	4
72	High-throughput DNA Sequencing Identifies Novel CtIP (RBBP8) Variants in Muscle-invasive Bladder Cancer Patients. Bladder Cancer, 2015, 1, 31-44.	0.2	2

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73	Demographic inference from multiple whole genomes using a particle filter for continuous Markov jump processes. PLoS ONE, 2021, 16, e0247647.	1.1	2
74	Efficient inference in state-space models through adaptive learning in online Monte Carlo expectation maximization. Computational Statistics, 2020, 35, 1319-1344.	0.8	0