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List of Publications by Year in descending order

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78
papers

10,508
citations

74677

40
h-index

70222

77
g-index

132
all docs

132
docs citations

132
times ranked

19357
citing authors

#	ARTICLE	IF	CITATIONS
1	The complete sequence of a human genome. <i>Science</i> , 2022, 376, 44-53.	20.9	1,565
2	Accurate circular consensus long-read sequencing improves variant detection and assembly of a human genome. <i>Nature Biotechnology</i> , 2019, 37, 1155-1162.	20.8	1,123
3	Integrating human sequence data sets provides a resource of benchmark SNP and indel genotype calls. <i>Nature Biotechnology</i> , 2014, 32, 246-251.	20.8	736
4	Extensive sequencing of seven human genomes to characterize benchmark reference materials. <i>Scientific Data</i> , 2016, 3, 160025.	5.4	614
5	Assuring the quality of next-generation sequencing in clinical laboratory practice. <i>Nature Biotechnology</i> , 2012, 30, 1033-1036.	20.8	438
6	Nanopore sequencing and the Shasta toolkit enable efficient de novo assembly of eleven human genomes. <i>Nature Biotechnology</i> , 2020, 38, 1044-1053.	20.8	390
7	A draft human pangenome reference. <i>Nature</i> , 2023, 617, 312-324.	36.2	318
8	Best practices for benchmarking germline small-variant calls in human genomes. <i>Nature Biotechnology</i> , 2019, 37, 555-560.	20.8	304
9	An open resource for accurately benchmarking small variant and reference calls. <i>Nature Biotechnology</i> , 2019, 37, 561-566.	20.8	294
10	A robust benchmark for detection of germline large deletions and insertions. <i>Nature Biotechnology</i> , 2020, 38, 1347-1355.	20.8	267
11	Complete genomic and epigenetic maps of human centromeres. <i>Science</i> , 2022, 376, eabl4178.	20.9	265
12	Stable nanoparticle aggregates/agglomerates of different sizes and the effect of their size on hemolytic cytotoxicity. <i>Nanotoxicology</i> , 2011, 5, 517-530.	3.0	226
13	Measuring silver nanoparticle dissolution in complex biological and environmental matrices using UV-visible absorbance. <i>Analytical and Bioanalytical Chemistry</i> , 2011, 401, 1993-2002.	3.9	194
14	A complete reference genome improves analysis of human genetic variation. <i>Science</i> , 2022, 376, eabl3533.	20.9	185
15	Best practices for evaluating single nucleotide variant calling methods for microbial genomics. <i>Frontiers in Genetics</i> , 2015, 6, 235.	2.3	168
16	Effects of temperature, acyl chain length, and flow-rate ratio on liposome formation and size in a microfluidic hydrodynamic focusing device. <i>Soft Matter</i> , 2010, 6, 1352.	2.8	136
17	Good laboratory practice for clinical next-generation sequencing informatics pipelines. <i>Nature Biotechnology</i> , 2015, 33, 689-693.	20.8	135
18	Chromosome-scale, haplotype-resolved assembly of human genomes. <i>Nature Biotechnology</i> , 2021, 39, 309-312.	20.8	129

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19	Medical implications of technical accuracy in genome sequencing. <i>Genome Medicine</i> , 2016, 8, 24.	8.5	126
20	The complete sequence of a human Y chromosome. <i>Nature</i> , 2023, 621, 344-354.	36.2	124
21	Curated variation benchmarks for challenging medically relevant autosomal genes. <i>Nature Biotechnology</i> , 2022, 40, 672-680.	20.8	110
22	Measuring Agglomerate Size Distribution and Dependence of Localized Surface Plasmon Resonance Absorbance on Gold Nanoparticle Agglomerate Size Using Analytical Ultracentrifugation. <i>ACS Nano</i> , 2011, 5, 8070-8079.	15.3	104
23	svclassify: a method to establish benchmark structural variant calls. <i>BMC Genomics</i> , 2016, 17, 64.	2.9	104
24	Genome-wide reconstruction of complex structural variants using read clouds. <i>Nature Methods</i> , 2017, 14, 915-920.	19.6	101
25	PrecisionFDA Truth Challenge V2: Calling variants from short and long reads in difficult-to-map regions. <i>Cell Genomics</i> , 2022, 2, 100129.	7.1	92
26	Benchmarking challenging small variants with linked and long reads. <i>Cell Genomics</i> , 2022, 2, 100128.	7.1	91
27	An analytical framework for optimizing variant discovery from personal genomes. <i>Nature Communications</i> , 2015, 6, 6275.	13.2	88
28	Theory of nematic backbone polymer phases and conformations. <i>Journal of Physics A</i> , 1986, 19, 2215-2227.	1.6	84
29	Statistical analysis of fractal-based brain tumor detection algorithms. <i>Magnetic Resonance Imaging</i> , 2005, 23, 671-678.	1.9	79
30	Performance assessment of DNA sequencing platforms in the ABRF Next-Generation Sequencing Study. <i>Nature Biotechnology</i> , 2021, 39, 1129-1140.	20.8	77
31	A diploid assembly-based benchmark for variants in the major histocompatibility complex. <i>Nature Communications</i> , 2020, 11, 4794.	13.2	62
32	A Rigorous Interlaboratory Examination of the Need to Confirm Next-Generation Sequencing-detected Variants with an Orthogonal Method in Clinical Genetic Testing. <i>Journal of Molecular Diagnostics</i> , 2019, 21, 318-329.	2.9	56
33	Chasing perfection: validation and polishing strategies for telomere-to-telomere genome assemblies. <i>Nature Methods</i> , 2022, 19, 687-695.	19.6	56
34	One in seven pathogenic variants can be challenging to detect by NGS: an analysis of 450,000 patients with implications for clinical sensitivity and genetic test implementation. <i>Genetics in Medicine</i> , 2021, 23, 1673-1680.	2.4	52
35	Synthetic Spike-in Standards Improve Run-Specific Systematic Error Analysis for DNA and RNA Sequencing. <i>PLoS ONE</i> , 2012, 7, e41356.	2.5	52
36	svviz: a read viewer for validating structural variants. <i>Bioinformatics</i> , 2015, 31, 3994-3996.	4.2	50

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37	Achieving high-sensitivity for clinical applications using augmented exome sequencing. <i>Genome Medicine</i> , 2015, 7, 71.	8.5	46
38	Assembly and annotation of an Ashkenazi human reference genome. <i>Genome Biology</i> , 2020, 21, 129.	9.2	46
39	Search for low mass vector resonances decaying into quark-antiquark pairs in proton-proton collisions at $s = 13 \sqrt{s} = 13 \text{ TeV}$. <i>Journal of High Energy Physics</i> , 2018, 2018, 1.	4.8	45
40	Electrochemical methods for the determination of the diffusion coefficient of ionophores and ionophore-ion complexes in plasticized PVC membranes. <i>Analyst</i> , 2008, 133, 635.	3.5	44
41	Magnetic connectors for microfluidic applications. <i>Lab on A Chip</i> , 2010, 10, 246-249.	6.1	44
42	Disentangling the effects of polymer coatings on silver nanoparticle agglomeration, dissolution, and toxicity to determine mechanisms of nanotoxicity. <i>Journal of Nanoparticle Research</i> , 2012, 14, 1.	2.0	44
43	Variant calling and benchmarking in an era of complete human genome sequences. <i>Nature Reviews Genetics</i> , 2023, 24, 464-483.	16.7	39
44	A research roadmap for next-generation sequencing informatics. <i>Science Translational Medicine</i> , 2016, 8, 335ps10.	13.4	37
45	Mathematical Model of Current-Polarized Ionophore-Based Ion-Selective Membranes. <i>Journal of Physical Chemistry B</i> , 2008, 112, 2008-2015.	2.7	30
46	Tools for annotation and comparison of structural variation. <i>F1000Research</i> , 2017, 6, 1795.	1.6	27
47	Chronopotentiometric method for the assessment of ionophore diffusion coefficients in solvent polymeric membranes. <i>Journal of Solid State Electrochemistry</i> , 2009, 13, 171-179.	2.6	25
48	Assessing reproducibility of inherited variants detected with short-read whole genome sequencing. <i>Genome Biology</i> , 2022, 23, 2.	9.2	23
49	Current-polarized ion-selective membranes: The influence of plasticizer and lipophilic background electrolyte on concentration profiles, resistance, and voltage transients. <i>Sensors and Actuators B: Chemical</i> , 2009, 136, 410-418.	8.0	21
50	Principles and Recommendations for Standardizing the Use of the Next-Generation Sequencing Variant File in Clinical Settings. <i>Journal of Molecular Diagnostics</i> , 2017, 19, 417-426.	2.9	21
51	Systematic analysis of the caridean shrimp superfamily Pandaloidea (Crustacea: Decapoda) based on molecular and morphological evidence. <i>Molecular Phylogenetics and Evolution</i> , 2019, 134, 200-210.	2.9	19
52	Reverse Current Pulse Method To Restore Uniform Concentration Profiles in Ion-Selective Membranes. 1. Galvanostatic Pulse Methods with Decreased Cycle Time. <i>Analytical Chemistry</i> , 2009, 81, 5146-5154.	6.8	18
53	High-coverage, long-read sequencing of Han Chinese trio reference samples. <i>Scientific Data</i> , 2019, 6, 91.	5.4	16
54	A strategy for building and using a human reference pangenome. <i>F1000Research</i> , 2019, 8, 1751.	1.6	14

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55	Assessment of Ionophore Complex Diffusion Coefficients in Solvent Polymeric Membranes. <i>Electroanalysis</i> , 2009, 21, 1923-1930.	3.0	13
56	Interpretation of chronopotentiometric transients of ion-selective membranes with two transition times. <i>Journal of Electroanalytical Chemistry</i> , 2010, 638, 254-261.	3.9	13
57	Recommendations for the Use of in Silico Approaches for Next-Generation Sequencing Bioinformatic Pipeline Validation. <i>Journal of Molecular Diagnostics</i> , 2023, 25, 3-16.	2.9	13
58	Prevalence and short-term outcome of acute kidney injury in patients with acute-on-chronic liver failure: A meta-analysis. <i>Journal of Viral Hepatitis</i> , 2020, 27, 810-817.	2.1	12
59	Nanomaterial Toxicity: Emerging Standards and Efforts to Support Standards Development. <i>Nanostructure Science and Technology</i> , 2011, , 179-208.	0.0	10
60	Serine/Threonine-Protein Kinase PFTK1 Modulates Oligodendrocyte Differentiation via PI3K/AKT Pathway. <i>Journal of Molecular Neuroscience</i> , 2015, 55, 977-984.	2.4	10
61	Determining Performance Metrics for Targeted Next-Generation Sequencing Panels Using Reference Materials. <i>Journal of Molecular Diagnostics</i> , 2018, 20, 583-590.	2.9	10
62	Multiscale analysis of pangenomes enables improved representation of genomic diversity for repetitive and clinically relevant genes. <i>Nature Methods</i> , 2023, 20, 1213-1221.	19.6	10
63	Reverse Current Pulse Method To Restore Uniform Concentration Profiles in Ion-Selective Membranes. 2. Comparison of the Efficiency of the Different Protocols. <i>Analytical Chemistry</i> , 2009, 81, 5155-5164.	6.8	9
64	A crowdsourced set of curated structural variants for the human genome. <i>PLoS Computational Biology</i> , 2020, 16, e1007933.	3.1	8
65	Challenging a bioinformatic tool's ability to detect microbial contaminants using in silico whole genome sequencing data. <i>PeerJ</i> , 2017, 5, e3729.	2.0	8
66	Advancing Benchmarks for Genome Sequencing. <i>Cell Systems</i> , 2015, 1, 176-177.	6.2	6
67	Unleashing novel STRs via characterization of genome in a bottle reference samples. <i>Forensic Science International: Genetics Supplement Series</i> , 2019, 7, 218-220.	0.3	6
68	Immobilization of fibrinogen antibody on self-assembled gold monolayers for immunosensor applications. <i>Tissue Engineering and Regenerative Medicine</i> , 2014, 11, 10-15.	3.8	5
69	Genomic Reference Materials for Clinical Applications. , 2015, , 393-402.		5
70	PEPR: pipelines for evaluating prokaryotic references. <i>Analytical and Bioanalytical Chemistry</i> , 2016, 408, 2975-2983.	3.9	5
71	A strategy for building and using a human reference pangenome. <i>F1000Research</i> , 2019, 8, 1751.	1.6	5
72	International interlaboratory study comparing single organism 16S rRNA gene sequencing data: Beyond consensus sequence comparisons. <i>Biomolecular Detection and Quantification</i> , 2015, 3, 17-24.	6.6	4

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73	Challenges of Accuracy in Germline Clinical Sequencing Data. JAMA - Journal of the American Medical Association, 2021, 326, 268.	7.0	4
74	Treatment of Older Patients, 40 to 70 Years of Age, with Acute Lymphoblastic Leukemia According to a Chemotherapy Regimen That Includes a Novel Pre-Phase for Rapid Tumor Load Reduction. Results of the Dutch-Belgian HOVON-71 Study.. Blood, 2009, 114, 2028-2028.	1.4	0
75	The Role of the National Institute of Standards in Measurement Assurance for Cell Therapies. , 2022, , 609-625.		0
76	On Materials Which Allow Students to Find Out Mathematical Propositions Using Snapping on GeoGebra. , 2020, 27, 13-17.		0
77	LongTR: genome-wide profiling of genetic variation at tandem repeats from long reads. Genome Biology, 2024, 25, .	9.2	0
78	High-coverage nanopore sequencing of samples from the 1000 Genomes Project to build a comprehensive catalog of human genetic variation. Genome Research, 0, , gr.279273.124.	5.6	0