List of Publications by Year in descending order

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#	Article	IF	CITATIONS
1	The complete sequence of a human genome. Science, 2022, 376, 44-53.	12.6	1,222
2	Accurate circular consensus long-read sequencing improves variant detection and assembly of a human genome. Nature Biotechnology, 2019, 37, 1155-1162.	17.5	1,010
3	Integrating human sequence data sets provides a resource of benchmark SNP and indel genotype calls. Nature Biotechnology, 2014, 32, 246-251.	17.5	722
4	Extensive sequencing of seven human genomes to characterize benchmark reference materials. Scientific Data, 2016, 3, 160025.	5.3	575
5	Assuring the quality of next-generation sequencing in clinical laboratory practice. Nature Biotechnology, 2012, 30, 1033-1036.	17.5	437
6	Nanopore sequencing and the Shasta toolkit enable efficient de novo assembly of eleven human genomes. Nature Biotechnology, 2020, 38, 1044-1053.	17.5	344
7	An open resource for accurately benchmarking small variant and reference calls. Nature Biotechnology, 2019, 37, 561-566.	17.5	277
8	Best practices for benchmarking germline small-variant calls in human genomes. Nature Biotechnology, 2019, 37, 555-560.	17.5	273
9	A robust benchmark for detection of germline large deletions and insertions. Nature Biotechnology, 2020, 38, 1347-1355.	17.5	233
10	Stable nanoparticle aggregates/agglomerates of different sizes and the effect of their size on hemolytic cytotoxicity. Nanotoxicology, 2011, 5, 517-530.	3.0	218
11	Complete genomic and epigenetic maps of human centromeres. Science, 2022, 376, eabl4178.	12.6	204
12	Measuring silver nanoparticle dissolution in complex biological and environmental matrices using UV–visible absorbance. Analytical and Bioanalytical Chemistry, 2011, 401, 1993-2002.	3.7	186
13	Best practices for evaluating single nucleotide variant calling methods for microbial genomics. Frontiers in Genetics, 2015, 6, 235.	2.3	160
14	A complete reference genome improves analysis of human genetic variation. Science, 2022, 376, eabl3533.	12.6	144
15	Good laboratory practice for clinical next-generation sequencing informatics pipelines. Nature Biotechnology, 2015, 33, 689-693.	17.5	134
16	Effects of temperature, acyl chain length, and flow-rate ratio on liposome formation and size in a microfluidic hydrodynamic focusing device. Soft Matter, 2010, 6, 1352.	2.7	129
17	Medical implications of technical accuracy in genome sequencing. Genome Medicine, 2016, 8, 24.	8.2	123
18	Chromosome-scale, haplotype-resolved assembly of human genomes. Nature Biotechnology, 2021, 39, 309-312.	17.5	109

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19	svclassify: a method to establish benchmark structural variant calls. BMC Genomics, 2016, 17, 64.	2.8	98
20	Measuring Agglomerate Size Distribution and Dependence of Localized Surface Plasmon Resonance Absorbance on Gold Nanoparticle Agglomerate Size Using Analytical Ultracentrifugation. ACS Nano, 2011, 5, 8070-8079.	14.6	96
21	Genome-wide reconstruction of complex structural variants using read clouds. Nature Methods, 2017, 14, 915-920.	19.0	96
22	Curated variation benchmarks for challenging medically relevant autosomal genes. Nature Biotechnology, 2022, 40, 672-680.	17.5	90
23	An analytical framework for optimizing variant discovery from personal genomes. Nature Communications, 2015, 6, 6275.	12.8	88
24	Statistical analysis of fractal-based brain tumor detection algorithms. Magnetic Resonance Imaging, 2005, 23, 671-678.	1.8	78
25	Benchmarking challenging small variants with linked and long reads. Cell Genomics, 2022, 2, 100128.	6.5	77
26	PrecisionFDA Truth Challenge V2: Calling variants from short and long reads in difficult-to-map regions. Cell Genomics, 2022, 2, 100129.	6.5	72
27	Performance assessment of DNA sequencing platforms in the ABRF Next-Generation Sequencing Study. Nature Biotechnology, 2021, 39, 1129-1140.	17.5	69
28	A diploid assembly-based benchmark for variants in the major histocompatibility complex. Nature Communications, 2020, 11, 4794.	12.8	56
29	Synthetic Spike-in Standards Improve Run-Specific Systematic Error Analysis for DNA and RNA Sequencing. PLoS ONE, 2012, 7, e41356.	2.5	52
30	A Rigorous Interlaboratory Examination of the Need to Confirm Next-Generation Sequencing–Detected Variants with an Orthogonal MethodÂin Clinical Genetic Testing. Journal of Molecular Diagnostics, 2019, 21, 318-329.	2.8	49
31	svviz: a read viewer for validating structural variants. Bioinformatics, 2015, 31, 3994-3996.	4.1	46
32	Achieving high-sensitivity for clinical applications using augmented exome sequencing. Genome Medicine, 2015, 7, 71.	8.2	46
33	Electrochemical methods for the determination of the diffusion coefficient of ionophores and ionophore–ion complexes in plasticized PVC membranes. Analyst, The, 2008, 133, 635.	3.5	44
34	Disentangling the effects of polymer coatings on silver nanoparticle agglomeration, dissolution, and toxicity to determine mechanisms of nanotoxicity. Journal of Nanoparticle Research, 2012, 14, 1.	1.9	44
35	Magnetic connectors for microfluidic applications. Lab on A Chip, 2010, 10, 246-249.	6.0	43
36	Assembly and annotation of an Ashkenazi human reference genome. Genome Biology, 2020, 21, 129.	8.8	42

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37	Chasing perfection: validation and polishing strategies for telomere-to-telomere genome assemblies. Nature Methods, 2022, 19, 687-695.	19.0	42
38	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. Genetics in Medicine, 2021, 23, 1673-1680.	2.4	40
39	A research roadmap for next-generation sequencing informatics. Science Translational Medicine, 2016, 8, 335ps10.	12.4	37
40	Mathematical Model of Currentâ€Polarized Ionophoreâ€Based Ionâ€Selective Membranes: Large Current Chronopotentiometry. Electroanalysis, 2008, 20, 259-269.	2.9	30
41	Mathematical Model of Current-Polarized Ionophore-Based Ion-Selective Membranes. Journal of Physical Chemistry B, 2008, 112, 2008-2015.	2.6	28
42	Tools for annotation and comparison of structural variation. F1000Research, 2017, 6, 1795.	1.6	26
43	Chronopotentiometric method for the assessment of ionophore diffusion coefficients in solvent polymeric membranes. Journal of Solid State Electrochemistry, 2009, 13, 171-179.	2.5	24
44	Development and Characterization of Reference Materials for Genetic Testing: Focus on Public Partnerships. Annals of Laboratory Medicine, 2016, 36, 513-520.	2.5	21
45	Current-polarized ion-selective membranes: The influence of plasticizer and lipophilic background electrolyte on concentration profiles, resistance, and voltage transients. Sensors and Actuators B: Chemical, 2009, 136, 410-418.	7.8	20
46	Principles and Recommendations for Standardizing the Use of the Next-Generation Sequencing Variant File in Clinical Settings. Journal of Molecular Diagnostics, 2017, 19, 417-426.	2.8	19
47	Assessing reproducibility of inherited variants detected with short-read whole genome sequencing. Genome Biology, 2022, 23, 2.	8.8	18
48	Reverse Current Pulse Method To Restore Uniform Concentration Profiles in Ion-Selective Membranes. 1. Galvanostatic Pulse Methods with Decreased Cycle Time. Analytical Chemistry, 2009, 81, 5146-5154.	6.5	17
49	A strategy for building and using a human reference pangenome. F1000Research, 2019, 8, 1751.	1.6	14
50	Assessment of Ionâ€lonophore Complex Diffusion Coefficients in Solvent Polymeric Membranes. Electroanalysis, 2009, 21, 1923-1930.	2.9	13
51	Interpretation of chronopotentiometric transients of ion-selective membranes with two transition times. Journal of Electroanalytical Chemistry, 2010, 638, 254-261.	3.8	13
52	High-coverage, long-read sequencing of Han Chinese trio reference samples. Scientific Data, 2019, 6, 91.	5.3	13
53	Determining Performance Metrics for Targeted Next-Generation Sequencing Panels Using Reference Materials. Journal of Molecular Diagnostics, 2018, 20, 583-590.	2.8	10
54	Reverse Current Pulse Method To Restore Uniform Concentration Profiles in Ion-Selective Membranes. 2. Comparison of the Efficiency of the Different Protocols. Analytical Chemistry, 2009, 81, 5155-5164.	6.5	9

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55	Nanomaterial Toxicity: Emerging Standards and Efforts to Support Standards Development. Nanostructure Science and Technology, 2011, , 179-208.	0.1	9
56	Challenging a bioinformatic tool's ability to detect microbial contaminants using <i>in silico</i> whole genome sequencing data. PeerJ, 2017, 5, e3729.	2.0	8
57	Advancing Benchmarks for Genome Sequencing. Cell Systems, 2015, 1, 176-177.	6.2	6
58	A crowdsourced set of curated structural variants for the human genome. PLoS Computational Biology, 2020, 16, e1007933.	3.2	6
59	Immobilization of fibrinogen antibody on self-assembled gold monolayers for immunosensor applications. Tissue Engineering and Regenerative Medicine, 2014, 11, 10-15.	3.7	5
60	Genomic Reference Materials for Clinical Applications. , 2015, , 393-402.		5
61	PEPR: pipelines for evaluating prokaryotic references. Analytical and Bioanalytical Chemistry, 2016, 408, 2975-2983.	3.7	5
62	A strategy for building and using a human reference pangenome. F1000Research, 2019, 8, 1751.	1.6	5
63	Unleashing novel STRS via characterization of genome in a bottle reference samples. Forensic Science International: Genetics Supplement Series, 2019, 7, 218-220.	0.3	5
64	Challenges of Accuracy in Germline Clinical Sequencing Data. JAMA - Journal of the American Medical Association, 2021, 326, 268.	7.4	4
65	International interlaboratory study comparing single organism 16S rRNA gene sequencing data: Beyond consensus sequence comparisons. Biomolecular Detection and Quantification, 2015, 3, 17-24.	7.0	3
66	CrowdVariant: a crowdsourcing approach to classify copy number variants. Pacific Symposium on Biocomputing, 2019, 24, 224-235.	0.7	2
67	The Role of the National Institute of Standards in Measurement Assurance for Cell Therapies. , 2022, , 609-625.		0