List of Publications by Year in descending order

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#	Article	IF	CITATIONS
1	ANNOVAR: functional annotation of genetic variants from high-throughput sequencing data. Nucleic Acids Research, 2010, 38, e164-e164.	14.5	10,960
2	PennCNV: An integrated hidden Markov model designed for high-resolution copy number variation detection in whole-genome SNP genotyping data. Genome Research, 2007, 17, 1665-1674.	5.5	1,586
3	MapSplice: Accurate mapping of RNA-seq reads for splice junction discovery. Nucleic Acids Research, 2010, 38, e178-e178.	14.5	946
4	InterVar: Clinical Interpretation of Genetic Variants by the 2015 ACMG-AMP Guidelines. American Journal of Human Genetics, 2017, 100, 267-280.	6.2	717
5	wANNOVAR: annotating genetic variants for personal genomes via the web. Journal of Medical Genetics, 2012, 49, 433-436.	3.2	366
6	Reverse engineering cellular networks. Nature Protocols, 2006, 1, 662-671.	12.0	345
7	A human Bâ€cell interactome identifies MYB and FOXM1 as master regulators of proliferation in germinal centers. Molecular Systems Biology, 2010, 6, 377.	7.2	336
8	Phenolyzer: phenotype-based prioritization of candidate genes for human diseases. Nature Methods, 2015, 12, 841-843.	19.0	327
9	Improved exome prioritization of disease genes through cross-species phenotype comparison. Genome Research, 2014, 24, 340-348.	5.5	300
10	Long-read sequencing and de novo assembly of a Chinese genome. Nature Communications, 2016, 7, 12065.	12.8	242
11	Detection of DNA base modifications by deep recurrent neural network on Oxford Nanopore sequencing data. Nature Communications, 2019, 10, 2449.	12.8	237
12	Genomic characteristics of cattle copy number variations. BMC Genomics, 2011, 12, 127.	2.8	201
13	TAF1 Variants Are Associated with Dysmorphic Features, Intellectual Disability, and Neurological Manifestations. American Journal of Human Genetics, 2015, 97, 922-932.	6.2	101
14	Deep Phenotyping on Electronic Health Records Facilitates Genetic Diagnosis by Clinical Exomes. American Journal of Human Genetics, 2018, 103, 58-73.	6.2	99
15	Long-read sequencing identified intronic repeat expansions in <i>SAMD12</i> from Chinese pedigrees affected with familial cortical myoclonic tremor with epilepsy. Journal of Medical Genetics, 2019, 56, 265-270.	3.2	82
16	Interrogating the "unsequenceable―genomic trinucleotide repeat disorders by long-read sequencing. Genome Medicine, 2017, 9, 65.	8.2	79
17	Modeling genetic inheritance of copy number variations. Nucleic Acids Research, 2008, 36, e138-e138.	14.5	77
18	Jannovar: A Java Library for Exome Annotation. Human Mutation, 2014, 35, 548-555.	2.5	63

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19	Genome-Wide Association Study of Multiplex Schizophrenia Pedigrees. American Journal of Psychiatry, 2012, 169, 963-973.	7.2	61
20	Deconvolution of transcriptional networks identifies TCF4 as a master regulator in schizophrenia. Science Advances, 2019, 5, eaau4139.	10.3	59
21	Group Lasso Regularized Deep Learning for Cancer Prognosis from Multi-Omics and Clinical Features. Genes, 2019, 10, 240.	2.4	59
22	NanoMod: a computational tool to detect DNA modifications using Nanopore long-read sequencing data. BMC Genomics, 2019, 20, 78.	2.8	50
23	LIQA: long-read isoform quantification and analysis. Genome Biology, 2021, 22, 182.	8.8	49
24	Doc2Hpo: a web application for efficient and accurate HPO concept curation. Nucleic Acids Research, 2019, 47, W566-W570.	14.5	47
25	iCACES: integrated CAncer GEnome Score for comprehensively prioritizing driver genes in personal cancer genomes. Genome Medicine, 2016, 8, 135.	8.2	45
26	Phen2Gene: rapid phenotype-driven gene prioritization for rare diseases. NAR Genomics and Bioinformatics, 2020, 2, Iqaa032.	3.2	45
27	Single-molecule optical mapping enables quantitative measurement of D4Z4 repeats in facioscapulohumeral muscular dystrophy (FSHD). Journal of Medical Genetics, 2020, 57, 109-120.	3.2	43
28	Single-cell multiomics reveals increased plasticity, resistant populations, and stem-cell–like blasts in <i>KMT2A</i> -rearranged leukemia. Blood, 2022, 139, 2198-2211.	1.4	37
29	Variant Interpretation for Cancer (VIC): a computational tool for assessing clinical impacts of somatic variants. Genome Medicine, 2019, 11, 53.	8.2	36
30	A semi-supervised approach for predicting cell-type specific functional consequences of non-coding variation using MPRAs. Nature Communications, 2018, 9, 5199.	12.8	34
31	NanoCaller for accurate detection of SNPs and indels in difficult-to-map regions from long-read sequencing by haplotype-aware deep neural networks. Genome Biology, 2021, 22, 261.	8.8	33
32	Detection of base analogs incorporated during DNA replication by nanopore sequencing. Nucleic Acids Research, 2020, 48, e88-e88.	14.5	31
33	NextSV: a meta-caller for structural variants from low-coverage long-read sequencing data. BMC Bioinformatics, 2018, 19, 180.	2.6	29
34	Gene Expression in Patient-Derived Neural Progenitors Implicates WNT5A Signaling in the Etiology of Schizophrenia. Biological Psychiatry, 2020, 88, 236-247.	1.3	28
35	Multiplex gene and phenotype network to characterize shared genetic pathways of epilepsy and autism. Scientific Reports, 2021, 11, 952.	3.3	27
36	LinkedSV for detection of mosaic structural variants from linked-read exome and genome sequencing data. Nature Communications, 2019, 10, 5585.	12.8	24

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37	LongGF: computational algorithm and software tool for fast and accurate detection of gene fusions by long-read transcriptome sequencing. BMC Genomics, 2020, 21, 793.	2.8	22
38	Detecting differential alternative splicing events in scRNA-seq with or without Unique Molecular Identifiers. PLoS Computational Biology, 2020, 16, e1007925.	3.2	20
39	The proteome and its dynamics: A missing piece for integrative multi-omics in schizophrenia. Schizophrenia Research, 2020, 217, 148-161.	2.0	16
40	DeepRepeat: direct quantification of short tandem repeats on signal data from nanopore sequencing. Genome Biology, 2022, 23, 108.	8.8	16
41	KBG syndrome involving a single-nucleotide duplication in <i>ANKRD11</i> . Journal of Physical Education and Sports Management, 2016, 2, a001131.	1.2	15
42	Ultrafast and scalable variant annotation and prioritization with big functional genomics data. Genome Research, 2020, 30, 1789-1801.	5.5	14
43	CancerVar: An artificial intelligence–empowered platform for clinical interpretation of somatic mutations in cancer. Science Advances, 2022, 8, eabj1624.	10.3	14
44	<i>SCN8A</i> mutation in a child presenting with seizures and developmental delays. Journal of Physical Education and Sports Management, 2016, 2, a001073.	1.2	12
45	Genome-wide detection of short tandem repeat expansions by long-read sequencing. BMC Bioinformatics, 2020, 21, 542.	2.6	11
46	Identification of Copy Number Variants from SNP Arrays Using PennCNV. Methods in Molecular Biology, 2018, 1833, 1-28.	0.9	8
47	PhenCards: a data resource linking human phenotype information to biomedical knowledge. Genome Medicine, 2021, 13, 91.	8.2	6
48	Clinical Phenotypic Spectrum of 4095 Individuals with Down Syndrome from Text Mining of Electronic Health Records. Genes, 2021, 12, 1159.	2.4	6
49	Cell-Type-Specific Proteogenomic Signal Diffusion for Integrating Multi-Omics Data Predicts Novel Schizophrenia Risk Genes. Patterns, 2020, 1, 100091.	5.9	5
50	A computational method for direct imputation of cell type-specific expression profiles and cellular compositions from bulk-tissue RNA-Seq in brain disorders. NAR Genomics and Bioinformatics, 2021, 3, lqab056.	3.2	5
51	Expediting knowledge acquisition by a web framework for Knowledge Graph Exploration and Visualization (KGEV): case studies on COVID-19 and Human Phenotype Ontology. BMC Medical Informatics and Decision Making, 2022, 22, .	3.0	3
52	Evaluation of biological and technical variations in low-input RNA-Seq and single-cell RNA-Seq. International Journal of Computational Biology and Drug Design, 2018, 11, 5.	0.3	2
53	Cell Type-Specific Annotation and Fine Mapping of Variants Associated With Brain Disorders. Frontiers in Genetics, 2020, 11, 575928.	2.3	2
54	Accelerating bioinformatics research with International Conference on Intelligent Biology and Medicine 2020. BMC Bioinformatics, 2020, 21, 563.	2.6	2

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55	Evaluation of biological and technical variations in low-input RNA-Seq and single-cell RNA-Seq. International Journal of Computational Biology and Drug Design, 2018, 11, 5.	0.3	1
56	Polishing high-quality genome assemblies. Nature Methods, 0, , .	19.0	1
57	Innovating Computational Biology and Intelligent Medicine: ICIBM 2019 Special Issue. Genes, 2020, 11, 437.	2.4	Ο
58	Epigenetic Dysregulation in Meningiomas. Neuro-Oncology Advances, 0, , .	0.7	0