List of Publications by Year in descending order

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#	Article	IF	CITATIONS
1	EXCAVATOR: detecting copy number variants from whole-exome sequencing data. Genome Biology, 2013, 14, R120.	9.6	213
2	Detection of Genomic Structural Variants from Next-Generation Sequencing Data. Frontiers in Bioengineering and Biotechnology, 2015, 3, 92.	4.1	212
3	Discovering chimeric transcripts in paired-end RNA-seq data by using EricScript. Bioinformatics, 2012, 28, 3232-3239.	4.1	154
4	Nanopore sequencing data analysis: state of the art, applications and challenges. Briefings in Bioinformatics, 2018, 19, 1256-1272.	6.5	91
5	<i>H</i> Â3 Â <i>M</i> Â2 : detection of runs of homozygosity from whole-exome sequencing data. Bioinformatics, 2014, 30, 2852-2859.	4.1	88
6	Enhanced copy number variants detection from whole-exome sequencing data using EXCAVATOR2. Nucleic Acids Research, 2016, 44, gkw695.	14.5	75
7	Read count approach for DNA copy number variants detection. Bioinformatics, 2012, 28, 470-478.	4.1	67
8	Bioinformatics for Next Generation Sequencing Data. Genes, 2010, 1, 294-307.	2.4	65
9	Genetic analysis of 56 polymorphisms in 17 genes involved in methionine metabolism in patients with abdominal aortic aneurysm. Journal of Medical Genetics, 2008, 45, 721-730.	3.2	63
10	Detecting common copy number variants in high-throughput sequencing data by using JointSLM algorithm. Nucleic Acids Research, 2011, 39, e65-e65.	14.5	63
11	Clinical Significance of Rare Copy Number Variations in Epilepsy. Archives of Neurology, 2012, 69, 322.	4.5	61
12	Genetic Bases of Bicuspid Aortic Valve: The Contribution of Traditional and High-Throughput Sequencing Approaches on Research and Diagnosis. Frontiers in Physiology, 2017, 8, 612.	2.8	57
13	A model of anti-angiogenesis: differential transcriptosome profiling of microvascular endothelial cells from diffuse systemic sclerosis patients. Arthritis Research and Therapy, 2006, 8, R115.	3.5	56
14	The antiangiogenic tissue kallikrein pattern of endothelial cells in systemic sclerosis. Arthritis and Rheumatism, 2005, 52, 3618-3628.	6.7	55
15	372Åkb microdeletion in 18q12.3 causing SETBP1 haploinsufficiency associated with mild mental retardation and expressive speech impairment. European Journal of Medical Genetics, 2012, 55, 216-221.	1.3	55
16	Epilepsy with auditory features. Neurology: Genetics, 2015, 1, e5.	1.9	55
17	Characterization of MinION nanopore data for resequencing analyses. Briefings in Bioinformatics, 2017, 18, bbw077.	6.5	55
18	Genetic polymorphisms of antioxidant enzymes as risk factors for oxidative stress-associated complications in preterm infants. Free Radical Research, 2012, 46, 1130-1139.	3.3	43

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19	Gene Expression Profiling of Peripheral Blood in Patients with Abdominal Aortic Aneurysm. European Journal of Vascular and Endovascular Surgery, 2009, 38, 104-112.	1.5	42
20	Early-onset ischaemic stroke: Analysis of 58 polymorphisms in 17 genes involved in methionine metabolism. Thrombosis and Haemostasis, 2010, 104, 231-242.	3.4	35
21	XCAVATOR: accurate detection and genotyping of copy number variants from second and third generation whole-genome sequencing experiments. BMC Genomics, 2017, 18, 747.	2.8	29
22	VISOR: a versatile haplotype-aware structural variant simulator for short- and long-read sequencing. Bioinformatics, 2020, 36, 1267-1269.	4.1	29
23	TRPA1 mediates damage of the retina induced by ischemia and reperfusion in mice. Cell Death and Disease, 2020, 11, 633.	6.3	28
24	EX-HOM (EXome HOMozygosity): A Proof of Principle. Human Heredity, 2011, 72, 45-53.	0.8	27
25	Desmoglein-2-Integrin Beta-8 Interaction Regulates Actin Assembly in Endothelial Cells: Deregulation in Systemic Sclerosis. PLoS ONE, 2013, 8, e68117.	2.5	27
26	Nanopore sequencing from liquid biopsy: analysis of copy number variations from cell-free DNA of lung cancer patients. Molecular Cancer, 2021, 20, 32.	19.2	27
27	A shifting level model algorithm that identifies aberrations in array-CGH data. Biostatistics, 2010, 11, 265-280.	1.5	26
28	RNA sequencing reveals <i>PNN</i> and <i>KCNQ1OT1</i> as predictive biomarkers of clinical outcome in stage III colorectal cancer patients treated with adjuvant chemotherapy. International Journal of Cancer, 2019, 145, 2580-2593.	5.1	26
29	A new hybrid approach for MHC genotyping: high-throughput NGS and long read MinION nanopore sequencing, with application to the non-model vertebrate Alpine chamois (Rupicapra rupicapra). Heredity, 2018, 121, 293-303.	2.6	25
30	Characterization and identification of hidden rare variants in the human genome. BMC Genomics, 2015, 16, 340.	2.8	24
31	Apolipoprotein(a) Kringle-IV Type 2 Copy Number Variation Is Associated with Venous Thromboembolism. PLoS ONE, 2016, 11, e0149427.	2.5	24
32	Sphingosine 1-Phosphate Induces Differentiation of Mesoangioblasts towards Smooth Muscle. A Role for GATA6. PLoS ONE, 2011, 6, e20389.	2.5	23
33	Gene expression profile of rat left ventricles reveals persisting changes following chronic mild exercise protocol: implications for cardioprotection. BMC Genomics, 2009, 10, 342.	2.8	22
34	Detection of Runs of Homozygosity from Whole Exome Sequencing Data: State of the Art and Perspectives for Clinical, Population and Epidemiological Studies. Human Heredity, 2014, 77, 63-72.	0.8	21
35	A novel founder MYO15A frameshift duplication is the major cause of genetic hearing loss in Oman. Journal of Human Genetics, 2017, 62, 259-264.	2.3	21
36	The ion channels and transporters gene expression profile indicates a shift in excitability and metabolisms during malignant progression of Follicular Lymphoma. Scientific Reports, 2019, 9, 8586.	3.3	20

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37	Sanger Validation of High-Throughput Sequencing in Genetic Diagnosis: Still the Best Practice?. Frontiers in Genetics, 2020, 11, 592588.	2.3	20
38	Heterogeneous magnitude of immunological memory to SARSâ€CoVâ€2 in recovered individuals. Clinical and Translational Immunology, 2021, 10, e1281.	3.8	19
39	Exome sequencing of two Italian pedigrees with non-isolated Chiari malformation type I reveals candidate genes for cranio-facial development. European Journal of Human Genetics, 2017, 25, 952-959.	2.8	18
40	NanoR: A user-friendly R package to analyze and compare nanopore sequencing data. PLoS ONE, 2019, 14, e0216471.	2.5	17
41	A very fast and accurate method for calling aberrations in array-CGH data. Biostatistics, 2010, 11, 515-518.	1.5	16
42	Nano-GLADIATOR: real-time detection of copy number alterations from nanopore sequencing data. Bioinformatics, 2019, 35, 4213-4221.	4.1	15
43	TRiCoLOR: tandem repeat profiling using whole-genome long-read sequencing data. GigaScience, 2020, 9, .	6.4	15
44	Xome-Blender: A novel cancer genome simulator. PLoS ONE, 2018, 13, e0194472.	2.5	14
45	A microRNA profile of pediatric glioblastoma: The role of NUCKS1 upregulation. Molecular and Clinical Oncology, 2019, 10, 331-338.	1.0	13
46	Assessment of Fibrinolytic Activity by Measuring the Lysis Time of a Tissue Factor-induced Clot: A Feasibility Evaluation. Clinical and Applied Thrombosis/Hemostasis, 2010, 16, 337-344.	1.7	12
47	Evaluation of Germline Structural Variant Calling Methods for Nanopore Sequencing Data. Frontiers in Genetics, 2021, 12, 761791.	2.3	12
48	Effect of space flight on the behavior of human retinal pigment epithelial ARPE-19 cells and evaluation of coenzyme Q10 treatment. Cellular and Molecular Life Sciences, 2021, 78, 7795-7812.	5.4	11
49	Genome-wide copy number analysis in pediatric glioblastoma multiforme. American Journal of Cancer Research, 2014, 4, 293-303.	1.4	10
50	PyPore: a python toolbox for nanopore sequencing data handling. Bioinformatics, 2019, 35, 4445-4447.	4.1	9
51	Using a calibration experiment to assess gene-specific information: full Bayesian and empirical Bayesian models for two-channel microarray data. Bioinformatics, 2006, 22, 50-57.	4.1	8
52	High-Throughput Multiplex Single-Nucleotide Polymorphism (SNP) Analysis in Genes Involved in Methionine Metabolism. Biochemical Genetics, 2008, 46, 406-423.	1.7	7
53	A systematic analysis of bone marrow cells by flow cytometry defines a specific phenotypic profile beyond GPI deficiency in paroxysmal nocturnal hemoglobinuria. Cytometry Part B - Clinical Cytometry, 2013, 84B, 71-81.	1.5	7
54	Using XCAVATOR and EXCAVATOR2 to Identify CNVs from WGS, WES, and TS Data. Current Protocols in Human Genetics, 2018, 98, e65.	3.5	7

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55	Charting differentially methylated regions in cancer with Rocker-meth. Communications Biology, 2021, 4, 1249.	4.4	7
56	Bicuspid Aortic Valve: Role of Multiple Gene Variants in Influencing the Clinical Phenotype. BioMed Research International, 2018, 2018, 1-9.	1.9	6
57	WNP: A Novel Algorithm for Gene Products Annotation from Weighted Functional Networks. PLoS ONE, 2012, 7, e38767.	2.5	5
58	Long Reads, Short Time: Feasibility of Prenatal Sample Karyotyping by Nanopore Genome Sequencing. Clinical Chemistry, 2019, 65, 1605-1608.	3.2	4
59	Involvement of RUNX1 Pathway Is a Common Event in the Leukemic Transformation of Chronic Myeloproliferative Neoplasms (MPNs). Blood, 2019, 134, 2968-2968.	1.4	4
60	Third-Generation Cytogenetic Analysis. Journal of Molecular Diagnostics, 2022, 24, 711-718.	2.8	4
61	SLMSuite: a suite of algorithms for segmenting genomic profiles. BMC Bioinformatics, 2017, 18, 321.	2.6	3
62	The Coenzyme Q10 (CoQ10) as Countermeasure for Retinal Damage Onboard the International Space Station: the CORM Project. Microgravity Science and Technology, 2018, 30, 925-931.	1.4	3
63	AUDACITY: A comprehensive approach for the detection and classification of Runs of Homozygosity in medical and population genomics. Computational and Structural Biotechnology Journal, 2020, 18, 1956-1967.	4.1	3
64	Carotid Artery Disease: Novel Pathophysiological Mechanisms Identified by Gene-expression Profiling of Peripheral Blood. European Journal of Vascular and Endovascular Surgery, 2010, 40, 549-558.	1.5	2
65	Precision Trial Drawer, a Computational Tool to Assist Planning of Genomics-Driven Trials in Oncology. JCO Precision Oncology, 2018, 2, 1-16.	3.0	2
66	GENE-03. MICRORNAS PROFILE IN PAEDIATRIC GBMS. Neuro-Oncology, 2017, 19, iv18-iv18.	1.2	1
67	Versatile Quality Control Methods for Nanopore Sequencing. Evolutionary Bioinformatics, 2019, 15, 117693431986306.	1.2	1
68	Genetic and nutritional factors determining circulating levels of lipoprotein(a): results of the "Montignoso Study― Internal and Emergency Medicine, 2020, 15, 1239-1245.	2.0	1
69	Moment estimation in discrete shifting level model applied to fast arrayâ€CGH segmentation. Statistica Neerlandica, 2013, 67, 227-262.	1.6	0
70	Editorial: Repetitive Structures in Biological Sequences: Algorithms and Applications. Frontiers in Bioengineering and Biotechnology, 2016, 4, 66.	4.1	0
71	Comparative Genomic and Expression Analysis of Chronic and Blast-Phase Cells in Patients with Myeloproliferative Neoplasms. Blood, 2018, 132, 1777-1777.	1.4	0
72	Large Genomic Alterations Occurring in the Transition from Chronic to Blast Phase of Chronic Myeloproliferative Neoplasms. Blood, 2018, 132, 3028-3028.	1.4	0