List of Publications by Year in descending order

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#	Article	IF	CITATIONS
1	Constitutional PIGA mutations cause a novel subtype of hemochromatosis in patients with neurologic dysfunction. Blood, 2022, 139, 1418-1422.	0.6	8
2	Neutralizing antibody response against the B.1.617.2 (delta) and the B.1.1.529 (omicron) variants after a third mRNA SARS-CoV-2 vaccine dose in kidney transplant recipients. American Journal of Transplantation, 2022, 22, 1873-1883.	2.6	37
3	Pangenome-based genome inference allows efficient and accurate genotyping across a wide spectrum of variant classes. Nature Genetics, 2022, 54, 518-525.	9.4	92
4	Pediatric T-ALL type-1 and type-2 relapses develop along distinct pathways of clonal evolution. Leukemia, 2022, 36, 1759-1768.	3.3	4
5	Haplotype-resolved diverse human genomes and integrated analysis of structural variation. Science, 2021, 372, .	6.0	358
6	Genomic insights into the pathogenesis of Epstein–Barr virus-associated diffuse large B-cell lymphoma by whole-genome and targeted amplicon sequencing. Blood Cancer Journal, 2021, 11, 102.	2.8	28
7	Metagenomic analysis of primary colorectal carcinomas and their metastases identifies potential microbial risk factors. Molecular Oncology, 2021, 15, 3363-3384.	2.1	17
8	Evidence for the Association between the Intronic Haplotypes of Ionotropic Glutamate Receptors and First-Episode Schizophrenia. Journal of Personalized Medicine, 2021, 11, 1250.	1.1	1
9	VISOR: a versatile haplotype-aware structural variant simulator for short- and long-read sequencing. Bioinformatics, 2020, 36, 1267-1269.	1.8	29
10	Single-cell analysis of structural variations and complex rearrangements with tri-channel processing. Nature Biotechnology, 2020, 38, 343-354.	9.4	59
11	TRiCoLOR: tandem repeat profiling using whole-genome long-read sequencing data. GigaScience, 2020, 9, .	3.3	15
12	Chromatin accessibility landscape of pediatric Tâ€lymphoblastic leukemia and human Tâ€cell precursors. EMBO Molecular Medicine, 2020, 12, e12104.	3.3	13
13	Tracy: basecalling, alignment, assembly and deconvolution of sanger chromatogram trace files. BMC Genomics, 2020, 21, 230.	1.2	63
14	Germline Elongator mutations in Sonic Hedgehog medulloblastoma. Nature, 2020, 580, 396-401.	13.7	94
15	Pan-cancer analysis of whole genomes. Nature, 2020, 578, 82-93.	13.7	1,966
16	The effects of common structural variants on 3D chromatin structure. BMC Genomics, 2020, 21, 95.	1.2	23
17	MBCL-21. GERMLINE ELONGATOR MUTATIONS IN SONIC HEDGEHOG MEDULLOBLASTOMA. Neuro-Oncology, 2020, 22, iii392-iii393.	0.6	0
18	MEDU-11. MOLECULAR CHARACTERIZATION OF ETMRs REVEALS A ROLE FOR R-LOOP MEDIATED CHROMOSOMAL INSTABILITY. Neuro-Oncology, 2019, 21, ii105-ii105.	0.6	0

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19	The NSL complex maintains nuclear architecture stability via lamin A/C acetylation. Nature Cell Biology, 2019, 21, 1248-1260.	4.6	61
20	Missense variants in NOX1 and p22phox in a case of very-early-onset inflammatory bowel disease are functionally linked to NOD2. Journal of Physical Education and Sports Management, 2019, 5, a002428.	0.5	13
21	Multi-platform discovery of haplotype-resolved structural variation in human genomes. Nature Communications, 2019, 10, 1784.	5.8	636
22	A highly soluble Sleeping Beauty transposase improves control of gene insertion. Nature Biotechnology, 2019, 37, 1502-1512.	9.4	63
23	The molecular landscape of ETMR at diagnosis and relapse. Nature, 2019, 576, 274-280.	13.7	94
24	<i>JUNB</i> , <i>DUSP2</i> , <i>SGK1</i> , <i>SOCS1</i> and <i>CREBBP</i> are frequently mutated in T-cell/histiocyte-rich large B-cell lymphoma. Haematologica, 2019, 104, 330-337.	1.7	45
25	Alfred: interactive multi-sample BAM alignment statistics, feature counting and feature annotation for long- and short-read sequencing. Bioinformatics, 2019, 35, 2489-2491.	1.8	61
26	Genomic structural variations lead to dysregulation of important coding and non oding RNA species in dilated cardiomyopathy. EMBO Molecular Medicine, 2018, 10, 107-120.	3.3	43
27	<scp>PDX</scp> models recapitulate the genetic and epigenetic landscape of pediatric Tâ€cell leukemia. EMBO Molecular Medicine, 2018, 10, .	3.3	38
28	ToTem: a tool for variant calling pipeline optimization. BMC Bioinformatics, 2018, 19, 243.	1.2	2
29	Coordinated expression and genetic polymorphisms in Grainyhead-like genes in human non-melanoma skin cancers. BMC Cancer, 2018, 18, 23.	1.1	10
30	Spectrum and prevalence of genetic predisposition in medulloblastoma: a retrospective genetic study and prospective validation in a clinical trial cohort. Lancet Oncology, The, 2018, 19, 785-798.	5.1	268
31	EMBR-10. GENOMIC COMPLEXITY AND EVOLUTION OF EMBRYONAL TUMORS WITH MULTILAYERED ROSETTES (ETMR). Neuro-Oncology, 2018, 20, i70-i70.	0.6	0
32	Abstract 3172: Targeting genomic instability in embryonal tumors with multilayered rosettes (ETMR). , 2018, , .		0
33	Longitudinal Multilevel Omic Analysis of Pediatric T-ALL Reveals Distinct Mechanisms for Disease Progression in Type 1 and in Type 2 Relapses. Blood, 2018, 132, 2826-2826.	0.6	0
34	Pediatric T-ALLs Developing into a Type 2 Relapse Originate from Cells That Carry the Potential of Variable Maturation into Subclones with Distinct Chromatin Landscapes. Blood, 2018, 132, 1545-1545.	0.6	0
35	Identification of a genetically defined ultra-high-risk group in relapsed pediatric T-lymphoblastic leukemia. Blood Cancer Journal, 2017, 7, e523-e523.	2.8	69
36	Potential protective role of Grainyheadâ€like genes in the development of clear cell renal cell cell carcinoma. Molecular Carcinogenesis, 2017, 56, 2414-2423.	1.3	11

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37	Characterization of Two Historic Smallpox Specimens from a Czech Museum. Viruses, 2017, 9, 200.	1.5	27
38	Identification of novel follicular dendritic cell sarcoma markers, FDCSP and SRGN, by whole transcriptome sequencing. Oncotarget, 2017, 8, 16463-16472.	0.8	43
39	<i>MCM3AP</i> and <i>POMP</i> Mutations Cause a DNA-Repair and DNA-Damage-Signaling Defect in an Immunodeficient Child. Human Mutation, 2016, 37, 257-268.	1.1	18
40	Personality similarity between teachers and their students influences teacher judgement of student achievement. Educational Psychology, 2016, 36, 863-878.	1.2	22
41	Highly recurrent mutations of SGK1, DUSP2 and JUNB in nodular lymphocyte predominant Hodgkin lymphoma. Leukemia, 2016, 30, 844-853.	3.3	80
42	Identification of an Ultra High-Risk and Targetable Molecular Signature in Relapsed Pediatric T-ALL. Blood, 2016, 128, 1084-1084.	0.6	0
43	Identification of cytokine-induced modulation of microRNA expression and secretion as measured by a novel microRNA specific qPCR assay. Scientific Reports, 2015, 5, 11590.	1.6	55
44	Assembly and diploid architecture of an individual human genome via single-molecule technologies. Nature Methods, 2015, 12, 780-786.	9.0	465
45	A global reference for human genetic variation. Nature, 2015, 526, 68-74.	13.7	13,998
46	An integrated map of structural variation in 2,504 human genomes. Nature, 2015, 526, 75-81.	13.7	1,994
47	Pediatric T-cell lymphoblastic leukemia evolves into relapse by clonal selection, acquisition of mutations and promoter hypomethylation. Haematologica, 2015, 100, 1442-1450.	1.7	65
48	Mit Wissen zu akkurateren Urteilen?. Zeitschrift Fur Entwicklungspsychologie Und Padagogische Psychologie, 2015, 47, 147-158.	0.3	4
49	A novel autosomal recessive TERT T1129P mutation in a dyskeratosis congenita family leads to cellular senescence and loss of CD34+ hematopoietic stem cells not reversible by mTOR-inhibition. Aging, 2015, 7, 911-927.	1.4	13
50	Gene Panel Sequencing of Primary and Relapsed Pediatric T-ALL Shows That Relapse-Specific Mutations Are Diverse and Mostly Non-Recurrent. Blood, 2015, 126, 1428-1428.	0.6	0
51	Genome Sequencing of SHH Medulloblastoma Predicts Genotype-Related Response to Smoothened Inhibition. Cancer Cell, 2014, 25, 393-405.	7.7	627
52	Identification of novel sequence variations in microRNAs in chronic lymphocytic leukemia. Carcinogenesis, 2014, 35, 992-1002.	1.3	18
53	Integrating sequence and array data to create an improved 1000 Genomes Project haplotype reference panel. Nature Communications, 2014, 5, 3934.	5.8	364
54	The activating STAT5B N642H mutation is a common abnormality in pediatric T-cell acute lymphoblastic leukemia and confers a higher risk of relapse. Haematologica, 2014, 99, e188-e192.	1.7	114

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55	Accuracy of Teacher Judgments. , 2014, , 27-43.		17
56	Targeted Deep Sequencing of Genetic Alterations Identified By Whole Exome Sequencing Reveals Clonal Evolution in Pediatric T-Lymphoblastic Leukemia. Blood, 2014, 124, 491-491.	0.6	0
57	Whole-exome sequencing links caspase recruitment domainÂ11 (CARD11) inactivation to severe combined immunodeficiency. Journal of Allergy and Clinical Immunology, 2013, 131, 1376-1383.e3.	1.5	127
58	Identification of a Ninein (NIN) mutation in a family with spondyloepimetaphyseal dysplasia with joint laxity (leptodactylic type)-like phenotype. Matrix Biology, 2013, 32, 387-392.	1.5	15
59	Integrative Genomic Analyses Reveal an Androgen-Driven Somatic Alteration Landscape in Early-Onset Prostate Cancer. Cancer Cell, 2013, 23, 159-170.	7.7	292
60	The Genomic and Transcriptomic Landscape of a HeLa Cell Line. G3: Genes, Genomes, Genetics, 2013, 3, 1213-1224.	0.8	355
61	Impact of genomic structural variation in <i>Drosophila melanogaster</i> based on population-scale sequencing. Genome Research, 2013, 23, 568-579.	2.4	72
62	Whole Exome Sequencing Identifies Novel Lyst-Missense Mutations In Incomplete Childhood Chediak-Higashi-Syndrome Presenting As Hemphagocytic Lymphohistiocytosis (HLH). Blood, 2013, 122, 3479-3479.	0.6	0
63	Whole Exome Sequencing In Relapsed Pediatric T-ALL: Progression Into Relapse Is Characterized By An Increased Number Of Somatic Mutations and a Conservation Of Mutations In Leukemogenic Driver Genes. Blood, 2013, 122, 228-228.	0.6	0
64	DELLY: structural variant discovery by integrated paired-end and split-read analysis. Bioinformatics, 2012, 28, i333-i339.	1.8	1,785
65	Genome Sequencing of Pediatric Medulloblastoma Links Catastrophic DNA Rearrangements with TP53 Mutations. Cell, 2012, 148, 59-71.	13.5	743
66	An integrated map of genetic variation from 1,092 human genomes. Nature, 2012, 491, 56-65.	13.7	7,199
67	Recurrent mutation of the ID3 gene in Burkitt lymphoma identified by integrated genome, exome and transcriptome sequencing. Nature Genetics, 2012, 44, 1316-1320.	9.4	389
68	Driver mutations in histone H3.3 and chromatin remodelling genes in paediatric glioblastoma. Nature, 2012, 482, 226-231.	13.7	2,129
69	ICGC PedBrain - dissecting the genomic complexity underlying medulloblastoma using whole-genome sequencing. BMC Proceedings, 2012, 6, .	1.8	1
70	Dissecting the genomic complexity underlying medulloblastoma. Nature, 2012, 488, 100-105.	13.7	765
71	Abstract 4872: ICGC PedBrain Tumor - Next-generation sequencing identifies novel subgroup-specific mutations and copy number aberrations in medulloblastoma. Cancer Research, 2012, 72, 4872-4872.	0.4	1
72	Sequence Variations in miRNA Genes Are Common and May Affect Their Expression in Patients with Chronic Lymphocytic Leukemia. Blood, 2012, 120, 3895-3895.	0.6	0

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73	Whole-Exome Sequencing Links CARD11 Inactivation with SCID. Blood, 2012, 120, 258-258.	0.6	0
74	Mapping copy number variation by population-scale genome sequencing. Nature, 2011, 470, 59-65.	13.7	991
75	Mutational Analysis of Mir-29 Family Members in Chronic Lymphocytic Leukemia. Blood, 2011, 118, 1770-1770.	0.6	0
76	Practical Multiple Sequence Alignment. , 2010, , 21-43.		0
77	RazerS—fast read mapping with sensitivity control. Genome Research, 2009, 19, 1646-1654.	2.4	125
78	A consistency-based consensus algorithm for <i>de novo</i> and reference-guided sequence assembly of short reads. Bioinformatics, 2009, 25, 1118-1124.	1.8	30
79	A parallel genetic algorithm to discover patterns in genetic markers that indicate predisposition to multifactorial disease. Computers in Biology and Medicine, 2008, 38, 826-836.	3.9	9
80	SeqAn An efficient, generic C++ library for sequence analysis. BMC Bioinformatics, 2008, 9, 11.	1.2	287
81	Robust consensus computation. BMC Bioinformatics, 2008, 9, .	1.2	3
82	Segment-based multiple sequence alignment. Bioinformatics, 2008, 24, i187-i192.	1.8	44
83	Fast and Adaptive Variable Order Markov Chain Construction. Lecture Notes in Computer Science,	1.0	15