

# Jan O Korbek

## List of Publications by Year in descending order

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168  
papers

70,169  
citations

6613

79  
h-index

5394

164  
g-index

202  
all docs

202  
docs citations

202  
times ranked

90977  
citing authors

#	ARTICLE	IF	CITATIONS
1	A global reference for human genetic variation. <i>Nature</i> , 2015, 526, 68-74.	27.8	13,998
2	A map of human genome variation from population-scale sequencing. <i>Nature</i> , 2010, 467, 1061-1073.	27.8	7,209
3	An integrated map of genetic variation from 1,092 human genomes. <i>Nature</i> , 2012, 491, 56-65.	27.8	7,199
4	Identification and analysis of functional elements in 1% of the human genome by the ENCODE pilot project. <i>Nature</i> , 2007, 447, 799-816.	27.8	4,709
5	Driver mutations in histone H3.3 and chromatin remodelling genes in paediatric glioblastoma. <i>Nature</i> , 2012, 482, 226-231.	27.8	2,129
6	International network of cancer genome projects. <i>Nature</i> , 2010, 464, 993-998.	27.8	2,114
7	An integrated map of structural variation in 2,504 human genomes. <i>Nature</i> , 2015, 526, 75-81.	27.8	1,994
8	DELLY: structural variant discovery by integrated paired-end and split-read analysis. <i>Bioinformatics</i> , 2012, 28, i333-i339.	4.1	1,785
9	Comprehensive genomic profiles of small cell lung cancer. <i>Nature</i> , 2015, 524, 47-53.	27.8	1,634
10	The landscape of genomic alterations across childhood cancers. <i>Nature</i> , 2018, 555, 321-327.	27.8	1,068
11	Paired-End Mapping Reveals Extensive Structural Variation in the Human Genome. <i>Science</i> , 2007, 318, 420-426.	12.6	1,003
12	Mapping copy number variation by population-scale genome sequencing. <i>Nature</i> , 2011, 470, 59-65.	27.8	991
13	The whole-genome landscape of medulloblastoma subtypes. <i>Nature</i> , 2017, 547, 311-317.	27.8	787
14	Dissecting the genomic complexity underlying medulloblastoma. <i>Nature</i> , 2012, 488, 100-105.	27.8	765
15	Subgroup-specific structural variation across 1,000 medulloblastoma genomes. <i>Nature</i> , 2012, 488, 49-56.	27.8	761
16	Genome Sequencing of Pediatric Medulloblastoma Links Catastrophic DNA Rearrangements with TP53 Mutations. <i>Cell</i> , 2012, 148, 59-71.	28.9	743
17	Eleven grand challenges in single-cell data science. <i>Genome Biology</i> , 2020, 21, 31.	8.8	742
18	Recurrent somatic alterations of FGFR1 and NTRK2 in pilocytic astrocytoma. <i>Nature Genetics</i> , 2013, 45, 927-932.	21.4	674

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19	Multi-platform discovery of haplotype-resolved structural variation in human genomes. <i>Nature Communications</i> , 2019, 10, 1784.	12.8	636
20	Genome Sequencing of SHH Medulloblastoma Predicts Genotype-Related Response to Smoothed Inhibition. <i>Cancer Cell</i> , 2014, 25, 393-405.	16.8	627
21	Toward understanding and exploiting tumor heterogeneity. <i>Nature Medicine</i> , 2015, 21, 846-853.	30.7	604
22	Natural variation in genome architecture among 205 <i>Drosophila melanogaster</i> Genetic Reference Panel lines. <i>Genome Research</i> , 2014, 24, 1193-1208.	5.5	565
23	Patterns of somatic structural variation in human cancer genomes. <i>Nature</i> , 2020, 578, 112-121.	27.8	560
24	What is a gene, post-ENCODE? History and updated definition. <i>Genome Research</i> , 2007, 17, 669-681.	5.5	530
25	Variation in Transcription Factor Binding Among Humans. <i>Science</i> , 2010, 328, 232-235.	12.6	521
26	Epigenomic alterations define lethal CIMP-positive ependymomas of infancy. <i>Nature</i> , 2014, 506, 445-450.	27.8	521
27	Enhancer hijacking activates GF11 family oncogenes in medulloblastoma. <i>Nature</i> , 2014, 511, 428-434.	27.8	520
28	Phenotypic impact of genomic structural variation: insights from and for human disease. <i>Nature Reviews Genetics</i> , 2013, 14, 125-138.	16.3	502
29	Assembly and diploid architecture of an individual human genome via single-molecule technologies. <i>Nature Methods</i> , 2015, 12, 780-786.	19.0	465
30	Criteria for Inference of Chromothripsis in Cancer Genomes. <i>Cell</i> , 2013, 152, 1226-1236.	28.9	457
31	Atypical Teratoid/Rhabdoid Tumors Are Comprised of Three Epigenetic Subgroups with Distinct Enhancer Landscapes. <i>Cancer Cell</i> , 2016, 29, 379-393.	16.8	438
32	Comprehensive analysis of chromothripsis in 2,658 human cancers using whole-genome sequencing. <i>Nature Genetics</i> , 2020, 52, 331-341.	21.4	431
33	Recurrent mutation of the ID3 gene in Burkitt lymphoma identified by integrated genome, exome and transcriptome sequencing. <i>Nature Genetics</i> , 2012, 44, 1316-1320.	21.4	389
34	Haplotype-resolved diverse human genomes and integrated analysis of structural variation. <i>Science</i> , 2021, 372, .	12.6	358
35	The Genomic and Transcriptomic Landscape of a HeLa Cell Line. <i>G3: Genes, Genomes, Genetics</i> , 2013, 3, 1213-1224.	1.8	355
36	The genetic architecture of Down syndrome phenotypes revealed by high-resolution analysis of human segmental trisomies. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2009, 106, 12031-12036.	7.1	342

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37	Pan-cancer analysis of somatic copy-number alterations implicates IRS4 and IGF2 in enhancer hijacking. <i>Nature Genetics</i> , 2017, 49, 65-74.	21.4	326
38	Active medulloblastoma enhancers reveal subgroup-specific cellular origins. <i>Nature</i> , 2016, 530, 57-62.	27.8	318
39	Integrative Genomic Analyses Reveal an Androgen-Driven Somatic Alteration Landscape in Early-Onset Prostate Cancer. <i>Cancer Cell</i> , 2013, 23, 159-170.	16.8	292
40	Prediction of effective genome size in metagenomic samples. <i>Genome Biology</i> , 2007, 8, R10.	9.6	281
41	Genomic basis for RNA alterations in cancer. <i>Nature</i> , 2020, 578, 129-136.	27.8	280
42	A Comprehensive Map of Mobile Element Insertion Polymorphisms in Humans. <i>PLoS Genetics</i> , 2011, 7, e1002236.	3.5	278
43	Pan-cancer analysis of whole genomes identifies driver rearrangements promoted by LINE-1 retrotransposition. <i>Nature Genetics</i> , 2020, 52, 306-319.	21.4	275
44	Highly rearranged chromosomes reveal uncoupling between genome topology and gene expression. <i>Nature Genetics</i> , 2019, 51, 1272-1282.	21.4	272
45	Spectrum and prevalence of genetic predisposition in medulloblastoma: a retrospective genetic study and prospective validation in a clinical trial cohort. <i>Lancet Oncology</i> , The, 2018, 19, 785-798.	10.7	268
46	Exome sequencing of osteosarcoma reveals mutation signatures reminiscent of BRCA deficiency. <i>Nature Communications</i> , 2015, 6, 8940.	12.8	242
47	PEMer: a computational framework with simulation-based error models for inferring genomic structural variants from massive paired-end sequencing data. <i>Genome Biology</i> , 2009, 10, R23.	9.6	223
48	Intratumor DNA Methylation Heterogeneity Reflects Clonal Evolution in Aggressive Prostate Cancer. <i>Cell Reports</i> , 2014, 8, 798-806.	6.4	219
49	Shadow Enhancers Are Pervasive Features of Developmental Regulatory Networks. <i>Current Biology</i> , 2016, 26, 38-51.	3.9	212
50	SHOT: a web server for the construction of genome phylogenies. <i>Trends in Genetics</i> , 2002, 18, 158-162.	6.7	193
51	Genomics and drug profiling of fatal TCF3-HLF <sup>+</sup> positive acute lymphoblastic leukemia identifies recurrent mutation patterns and therapeutic options. <i>Nature Genetics</i> , 2015, 47, 1020-1029.	21.4	190
52	Molecular Evolution of Early-Onset Prostate Cancer Identifies Molecular Risk Markers and Clinical Trajectories. <i>Cancer Cell</i> , 2018, 34, 996-1011.e8.	16.8	190
53	High-resolution genomic profiling of chronic lymphocytic leukemia reveals new recurrent genomic alterations. <i>Blood</i> , 2012, 120, 4783-4794.	1.4	179
54	Quantifying environmental adaptation of metabolic pathways in metagenomics. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2009, 106, 1374-1379.	7.1	177

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55	Analysis of genomic context: prediction of functional associations from conserved bidirectionally transcribed gene pairs. <i>Nature Biotechnology</i> , 2004, 22, 911-917.	17.5	166
56	Nucleotide-resolution analysis of structural variants using BreakSeq and a breakpoint library. <i>Nature Biotechnology</i> , 2010, 28, 47-55.	17.5	158
57	Risk-adapted therapy for young children with medulloblastoma (SJYC07): therapeutic and molecular outcomes from a multicentre, phase 2 trial. <i>Lancet Oncology</i> , The, 2018, 19, 768-784.	10.7	151
58	Structured RNAs in the ENCODE selected regions of the human genome. <i>Genome Research</i> , 2007, 17, 852-864.	5.5	150
59	BAZ2A (TIP5) is involved in epigenetic alterations in prostate cancer and its overexpression predicts disease recurrence. <i>Nature Genetics</i> , 2015, 47, 22-30.	21.4	141
60	Targeted Perturb-seq enables genome-scale genetic screens in single cells. <i>Nature Methods</i> , 2020, 17, 629-635.	19.0	139
61	Systematic Association of Genes to Phenotypes by Genome and Literature Mining. <i>PLoS Biology</i> , 2005, 3, e134.	5.6	138
62	Positive selection at the protein network periphery: Evaluation in terms of structural constraints and cellular context. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2007, 104, 20274-20279.	7.1	132
63	Whole-exome sequencing links caspase recruitment domain 11 (CARD11) inactivation to severe combined immunodeficiency. <i>Journal of Allergy and Clinical Immunology</i> , 2013, 131, 1376-1383.e3.	2.9	127
64	Genomic deletion of MAP3K7 at 6q12-22 is associated with early PSA recurrence in prostate cancer and absence of TMPRSS2:ERG fusions. <i>Modern Pathology</i> , 2013, 26, 975-983.	5.5	127
65	Fully phased human genome assembly without parental data using single-cell strand sequencing and long reads. <i>Nature Biotechnology</i> , 2021, 39, 302-308.	17.5	127
66	Analysis of copy number variants and segmental duplications in the human genome: Evidence for a change in the process of formation in recent evolutionary history. <i>Genome Research</i> , 2008, 18, 1865-1874.	5.5	126
67	High-resolution mapping of DNA copy alterations in human chromosome 22 using high-density tiling oligonucleotide arrays. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2006, 103, 4534-4539.	7.1	125
68	Systematic discovery of analogous enzymes in thiamin biosynthesis. <i>Nature Biotechnology</i> , 2003, 21, 790-795.	17.5	121
69	A cell-based model system links chromothripsis with hyperploidy. <i>Molecular Systems Biology</i> , 2015, 11, 828.	7.2	118
70	Relating CNVs to transcriptome data at fine resolution: Assessment of the effect of variant size, type, and overlap with functional regions. <i>Genome Research</i> , 2011, 21, 2004-2013.	5.5	109
71	Sequence diversity analyses of an improved rhesus macaque genome enhance its biomedical utility. <i>Science</i> , 2020, 370, .	12.6	105
72	Analysis of copy number variation in the rhesus macaque genome identifies candidate loci for evolutionary and human disease studies. <i>Human Molecular Genetics</i> , 2008, 17, 1127-1136.	2.9	101

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73	Mitochondrial mutations drive prostate cancer aggression. <i>Nature Communications</i> , 2017, 8, 656.	12.8	100
74	High-Resolution Copy-Number Variation Map Reflects Human Olfactory Receptor Diversity and Evolution. <i>PLoS Genetics</i> , 2008, 4, e1000249.	3.5	99
75	Genomic and transcriptomic changes complement each other in the pathogenesis of sporadic Burkitt lymphoma. <i>Nature Communications</i> , 2019, 10, 1459.	12.8	99
76	Clinical significance of different types of <i>p53</i> gene alteration in surgically treated prostate cancer. <i>International Journal of Cancer</i> , 2014, 135, 1369-1380.	5.1	95
77	The molecular landscape of ETMR at diagnosis and relapse. <i>Nature</i> , 2019, 576, 274-280.	27.8	94
78	Germline Elongator mutations in Sonic Hedgehog medulloblastoma. <i>Nature</i> , 2020, 580, 396-401.	27.8	94
79	The current excitement about copy-number variation: how it relates to gene duplications and protein families. <i>Current Opinion in Structural Biology</i> , 2008, 18, 366-374.	5.7	92
80	Pangenome-based genome inference allows efficient and accurate genotyping across a wide spectrum of variant classes. <i>Nature Genetics</i> , 2022, 54, 518-525.	21.4	92
81	Effects of the COVID-19 pandemic on life scientists. <i>Genome Biology</i> , 2020, 21, 113.	8.8	90
82	The Baker's Yeast Diploid Genome Is Remarkably Stable in Vegetative Growth and Meiosis. <i>PLoS Genetics</i> , 2010, 6, e1001109.	3.5	89
83	Data analysis: Create a cloud commons. <i>Nature</i> , 2015, 523, 149-151.	27.8	89
84	Distinct genomic aberrations associated with <i>ERG</i> rearranged prostate cancer. <i>Genes Chromosomes and Cancer</i> , 2009, 48, 366-380.	2.8	86
85	Dense and accurate whole-chromosome haplotyping of individual genomes. <i>Nature Communications</i> , 2017, 8, 1293.	12.8	83
86	Global Identification and Characterization of Transcriptionally Active Regions in the Rice Genome. <i>PLoS ONE</i> , 2007, 2, e294.	2.5	82
87	Primate genome architecture influences structural variation mechanisms and functional consequences. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2013, 110, 15764-15769.	7.1	80
88	Systematic prediction and validation of breakpoints associated with copy-number variants in the human genome. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2007, 104, 10110-10115.	7.1	78
89	Analysis of deletion breakpoints from 1,092 humans reveals details of mutation mechanisms. <i>Nature Communications</i> , 2015, 6, 7256.	12.8	77
90	Comprehensive characterization of complex structural variations in cancer by directly comparing genome sequence reads. <i>Nature Biotechnology</i> , 2014, 32, 1106-1112.	17.5	74

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91	Impact of genomic structural variation in <i>Drosophila melanogaster</i> based on population-scale sequencing. <i>Genome Research</i> , 2013, 23, 568-579.	5.5	72
92	Expectations and blind spots for structural variation detection from long-read assemblies and short-read genome sequencing technologies. <i>American Journal of Human Genetics</i> , 2021, 108, 919-928.	6.2	72
93	Leveraging European infrastructures to access 1 million human genomes by 2022. <i>Nature Reviews Genetics</i> , 2019, 20, 693-701.	16.3	69
94	Recurrent inversion polymorphisms in humans associate with genetic instability and genomic disorders. <i>Cell</i> , 2022, 185, 1986-2005.e26.	28.9	67
95	Pediatric T-cell lymphoblastic leukemia evolves into relapse by clonal selection, acquisition of mutations and promoter hypomethylation. <i>Haematologica</i> , 2015, 100, 1442-1450.	3.5	65
96	Negative Selection and Chromosome Instability Induced by Mad2 Overexpression Delay Breast Cancer but Facilitate Oncogene-Independent Outgrowth. <i>Cell Reports</i> , 2016, 15, 2679-2691.	6.4	65
97	Comprehensive Analysis of Chromatin States in Atypical Teratoid/Rhabdoid Tumor Identifies Diverging Roles for SWI/SNF and Polycomb in Gene Regulation. <i>Cancer Cell</i> , 2019, 35, 95-110.e8.	16.8	65
98	Genetic code expansion for multiprotein complex engineering. <i>Nature Methods</i> , 2016, 13, 997-1000.	19.0	63
99	The NSL complex maintains nuclear architecture stability via lamin A/C acetylation. <i>Nature Cell Biology</i> , 2019, 21, 1248-1260.	10.3	61
100	Alfred: interactive multi-sample BAM alignment statistics, feature counting and feature annotation for long- and short-read sequencing. <i>Bioinformatics</i> , 2019, 35, 2489-2491.	4.1	61
101	Chromothripsis followed by circular recombination drives oncogene amplification in human cancer. <i>Nature Genetics</i> , 2021, 53, 1673-1685.	21.4	61
102	Similar gene expression profiles do not imply similar tissue functions. <i>Trends in Genetics</i> , 2006, 22, 132-138.	6.7	59
103	Single-cell analysis of structural variations and complex rearrangements with tri-channel processing. <i>Nature Biotechnology</i> , 2020, 38, 343-354.	17.5	59
104	Systematic Inference of Copy-Number Genotypes from Personal Genome Sequencing Data Reveals Extensive Olfactory Receptor Gene Content Diversity. <i>PLoS Computational Biology</i> , 2010, 6, e1000988.	3.2	56
105	TMPRSS2-ERG Fusions Are Strongly Linked to Young Patient Age in Low-grade Prostate Cancer. <i>European Urology</i> , 2014, 66, 978-981.	1.9	54
106	Genome assembly and haplotyping with Hi-C. <i>Nature Biotechnology</i> , 2013, 31, 1099-1101.	17.5	53
107	Pan-cancer analysis distinguishes transcriptional changes of aneuploidy from proliferation. <i>Genome Research</i> , 2017, 27, 501-511.	5.5	52
108	The genomic and transcriptional landscape of primary central nervous system lymphoma. <i>Nature Communications</i> , 2022, 13, 2558.	12.8	52

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109	Germline <i>GPR161</i> Mutations Predispose to Pediatric Medulloblastoma. <i>Journal of Clinical Oncology</i> , 2020, 38, 43-50.	1.6	50
110	Acquisition of chromosome instability is a mechanism to evade oncogene addiction. <i>EMBO Molecular Medicine</i> , 2020, 12, e10941.	6.9	45
111	Chromatin modifiers Mdm2 and RNF2 prevent RNA:DNA hybrids that impair DNA replication. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2018, 115, E11311-E11320.	7.1	44
112	Alterations of microRNA and microRNA-regulated messenger RNA expression in germinal center B-cell lymphomas determined by integrative sequencing analysis. <i>Haematologica</i> , 2016, 101, 1380-1389.	3.5	43
113	Genomic structural variations lead to dysregulation of important coding and non-coding RNA species in dilated cardiomyopathy. <i>EMBO Molecular Medicine</i> , 2018, 10, 107-120.	6.9	43
114	Combining frequency and positional information to predict transcription factor binding sites. <i>Bioinformatics</i> , 2001, 17, 1019-1026.	4.1	42
115	Recurrent inversion toggling and great ape genome evolution. <i>Nature Genetics</i> , 2020, 52, 849-858.	21.4	40
116	<i>PDX</i> models recapitulate the genetic and epigenetic landscape of pediatric T-cell leukemia. <i>EMBO Molecular Medicine</i> , 2018, 10, .	6.9	38
117	Challenges in studying genomic structural variant formation mechanisms: The short-read dilemma and beyond. <i>BioEssays</i> , 2011, 33, 840-850.	2.5	34
118	Mutational mechanisms shaping the coding and noncoding genome of germinal center derived B-cell lymphomas. <i>Leukemia</i> , 2021, 35, 2002-2016.	7.2	34
119	MSB: A mean-shift-based approach for the analysis of structural variation in the genome. <i>Genome Research</i> , 2009, 19, 106-117.	5.5	33
120	Identification of <i>ZCCHC8</i> as fusion partner of <i>ROS1</i> in a case of congenital glioblastoma multiforme with a t(6;12)(q21;q24.3). <i>Genes Chromosomes and Cancer</i> , 2016, 55, 677-687.	2.8	33
121	A scalable CRISPR/Cas9-based fluorescent reporter assay to study DNA double-strand break repair choice. <i>Nature Communications</i> , 2020, 11, 4077.	12.8	33
122	A supervised hidden markov model framework for efficiently segmenting tiling array data in transcriptional and ChIP-chip experiments: systematically incorporating validated biological knowledge. <i>Bioinformatics</i> , 2006, 22, 3016-3024.	4.1	32
123	Familial long-read sequencing increases yield of de novo mutations. <i>American Journal of Human Genetics</i> , 2022, 109, 631-646.	6.2	32
124	Genomics: data sharing needs an international code of conduct. <i>Nature</i> , 2020, 578, 31-33.	27.8	31
125	VISOR: a versatile haplotype-aware structural variant simulator for short- and long-read sequencing. <i>Bioinformatics</i> , 2020, 36, 1267-1269.	4.1	29
126	Systems approaches identify the consequences of monosomy in somatic human cells. <i>Nature Communications</i> , 2021, 12, 5576.	12.8	29



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127	Deletion lengthening at chromosomes 6q and 16q targets multiple tumor suppressor genes and is associated with an increasingly poor prognosis in prostate cancer. <i>Oncotarget</i> , 2017, 8, 108923-108935.	1.8	26
128	The DART classification of unannotated transcription within the ENCODE regions: Associating transcription with known and novel loci. <i>Genome Research</i> , 2007, 17, 732-745.	5.5	25
129	Immortalization capacity of HPV types is inversely related to chromosomal instability. <i>Oncotarget</i> , 2016, 7, 37608-37621.	1.8	25
130	Heterogeneity of ERG expression in prostate cancer: a large section mapping study of entire prostatectomy specimens from 125 patients. <i>BMC Cancer</i> , 2016, 16, 641.	2.6	24
131	Strand-seq enables reliable separation of long reads by chromosome via expectation maximization. <i>Bioinformatics</i> , 2018, 34, i115-i123.	4.1	24
132	Genome-wide Screens Implicate Loss of Cullin Ring Ligase 3 in Persistent Proliferation and Genome Instability in TP53-Deficient Cells. <i>Cell Reports</i> , 2020, 31, 107465.	6.4	24
133	Extracting information from cDNA arrays. <i>Chaos</i> , 2001, 11, 98.	2.5	23
134	Structural Variation in Cancer: Role, Prevalence, and Mechanisms. <i>Annual Review of Genomics and Human Genetics</i> , 2022, 23, 123-152.	6.2	23
135	Genomic data sharing in Europe is stumbling—Could a code of conduct prevent its fall?. <i>EMBO Molecular Medicine</i> , 2020, 12, e11421.	6.9	22
136	Computing patient data in the cloud: practical and legal considerations for genetics and genomics research in Europe and internationally. <i>Genome Medicine</i> , 2017, 9, 58.	8.2	21
137	Single-cell strand sequencing of a macaque genome reveals multiple nested inversions and breakpoint reuse during primate evolution. <i>Genome Research</i> , 2020, 30, 1680-1693.	5.5	16
138	Identification of a Ninein (NIN) mutation in a family with spondyloepimetaphyseal dysplasia with joint laxity (leptodactylic type)-like phenotype. <i>Matrix Biology</i> , 2013, 32, 387-392.	3.6	15
139	TRiCoLOR: tandem repeat profiling using whole-genome long-read sequencing data. <i>GigaScience</i> , 2020, 9, .	6.4	15
140	InTAD: chromosome conformation guided analysis of enhancer target genes. <i>BMC Bioinformatics</i> , 2019, 20, 60.	2.6	14
141	Use of pathway analysis and genome context methods for functional genomics of <i>Mycoplasma pneumoniae</i> nucleotide metabolism. <i>Gene</i> , 2007, 396, 215-225.	2.2	13
142	Prevalence of chromosomal rearrangements involving non-ETS genes in prostate cancer. <i>International Journal of Oncology</i> , 2015, 46, 1637-1642.	3.3	13
143	Chromatin accessibility landscape of pediatric T-lymphoblastic leukemia and human T-cell precursors. <i>EMBO Molecular Medicine</i> , 2020, 12, e12104.	6.9	13
144	Somatic structural variant formation is guided by and influences genome architecture. <i>Genome Research</i> , 2022, 32, 643-655.	5.5	12

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145	Butler enables rapid cloud-based analysis of thousands of human genomes. <i>Nature Biotechnology</i> , 2020, 38, 288-292.	17.5	11
146	A 15q24 microdeletion in transient myeloproliferative disease (<scp>TMD</scp>) and acute megakaryoblastic leukaemia (<scp>AMKL</scp>) implicates <scp>PML</scp> and <scp>SUMO</scp>3 in the leukaemogenesis of <scp>TMD</scp>/<scp>AMKL</scp>. <i>British Journal of Haematology</i> , 2012, 157, 180-187.	2.5	9
147	Genomes of early onset prostate cancer. <i>Current Opinion in Urology</i> , 2017, 27, 481-487.	1.8	9
148	ASHLEYS: automated quality control for single-cell Strand-seq data. <i>Bioinformatics</i> , 2021, 37, 3356-3357.	4.1	9
149	Versatile workflow for cell typeâ€resolved transcriptional and epigenetic profiles from cryopreserved human lung. <i>JCI Insight</i> , 2021, 6, .	5.0	8
150	The Helmholtz Network for Bioinformatics: an integrative web portal for bioinformatics resources. <i>Bioinformatics</i> , 2004, 20, 268-270.	4.1	7
151	Next-generation sequencing-based detection of germline L1-mediated transductions. <i>BMC Genomics</i> , 2016, 17, 342.	2.8	7
152	The Porto European Cancer Research Summit 2021. <i>Molecular Oncology</i> , 2021, 15, 2507-2543.	4.6	7
153	Transgene Methylation in Mice Reflects Copy Number But Not Expression Level. <i>Molecular Biotechnology</i> , 2004, 26, 215-220.	2.4	5
154	The Aging Prostate Is Never â€Normalâ€ Implications from the Genomic Characterization of Multifocal Prostate Cancers. <i>European Urology</i> , 2015, 68, 348-350.	1.9	5
155	Using large-scale genome variation cohorts to decipher the molecular mechanism of cancer. <i>Comptes Rendus - Biologies</i> , 2016, 339, 308-313.	0.2	5
156	Enriched power of disease-concordant twin-case-only design in detecting interactions in genome-wide association studies. <i>European Journal of Human Genetics</i> , 2019, 27, 631-636.	2.8	4
157	Pediatric T-ALL type-1 and type-2 relapses develop along distinct pathways of clonal evolution. <i>Leukemia</i> , 2022, 36, 1759-1768.	7.2	4
158	A common microdeletion affecting a hippocampusâ€and amygdalaâ€specific isoform of tryptophan hydroxylase 2 is not associated with affective disorders. <i>Bipolar Disorders</i> , 2014, 16, 764-768.	1.9	2
159	Rare Germline Variants Are Associated with Rapid Biochemical Recurrence After Radical Prostate Cancer Treatment: A Pan Prostate Cancer Group Study. <i>European Urology</i> , 2022, 82, 201-211.	1.9	2
160	ICGC PedBrain - dissecting the genomic complexity underlying medulloblastoma using whole-genome sequencing. <i>BMC Proceedings</i> , 2012, 6, .	1.6	1
161	Systematic Identification of Determinants for Single-Strand Annealing-Mediated Deletion Formation in <i>Saccharomyces cerevisiae</i> . <i>G3: Genes, Genomes, Genetics</i> , 2017, 7, 3269-3279.	1.8	1
162	The X Chromosome from Telomere to Telomere: Key Achievements and Future Opportunities. <i>Faculty Reviews</i> , 2021, 10, 63.	3.9	1

#	ARTICLE	IF	CITATIONS
163	Whole-Exome Sequencing Links CARD11 Inactivation with SCID. Blood, 2012, 120, 258-258.	1.4	0
164	Whole Exome Sequencing Identifies Novel Lyst-Missense Mutations In Incomplete Childhood Chediak-Higashi-Syndrome Presenting As Hemphagocytic Lymphohistiocytosis (HLH). Blood, 2013, 122, 3479-3479.	1.4	0
165	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.	1.4	0
166	Targeted Deep Sequencing of Genetic Alterations Identified By Whole Exome Sequencing Reveals Clonal Evolution in Pediatric T-Lymphoblastic Leukemia. Blood, 2014, 124, 491-491.	1.4	0
167	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.	1.4	0
168	TP53 and KRAS Variants at Initial Diagnosis Identify an Ultra-High Risk Group of Pediatric T-Lymphoblastic Leukemia (T-ALL). Blood, 2021, 138, 1315-1315.	1.4	0