Jan O Korbel

List of Publications by Citations

183 papers	50,647 citations	73 h-index	202 g-index
202 ext. papers	62,692 ext. citations	19.6 avg, IF	6.74 L-index

#	Paper	IF	Citations
183	A global reference for human genetic variation. *Nature*, **2015**, 526, 68-74	50.4	8599
182	A map of human genome variation from population-scale sequencing. *Nature*, **2010**, 467, 1061-73	50.4	6142
181	An integrated map of genetic variation from 1,092 human genomes. *Nature*, **2012**, 491, 56-65	50.4	6049
180	Identification and analysis of functional elements in 1% of the human genome by the ENCODE pilot project. *Nature*, **2007**, 447, 799-816	50.4	4121
179	Driver mutations in histone H3.3 and chromatin remodelling genes in paediatric glioblastoma. *Nature*, **2012**, 482, 226-31	50.4	1655
178	International network of cancer genome projects. *Nature*, **2010**, 464, 993-8	50.4	1613
177	An integrated map of structural variation in 2,504 human genomes. *Nature*, **2015**, 526, 75-81	50.4	1368
176	DELLY: structural variant discovery by integrated paired-end and split-read analysis. *Bioinformatics*, **2012**, 28, i333-i339	7.2	1069
175	Comprehensive genomic profiles of small cell lung cancer. *Nature*, **2015**, 524, 47-53	50.4	1061
174	Paired-end mapping reveals extensive structural variation in the human genome. *Science*, **2007**, 318, 420-6	33.3	895
173	Mapping copy number variation by population-scale genome sequencing. *Nature*, **2011**, 470, 59-65	50.4	833
172	Dissecting the genomic complexity underlying medulloblastoma. *Nature*, **2012**, 488, 100-5	50.4	623
171	The landscape of genomic alterations across childhood cancers. *Nature*, **2018**, 555, 321-327	50.4	603
170	Genome sequencing of pediatric medulloblastoma links catastrophic DNA rearrangements with TP53 mutations. *Cell*, **2012**, 148, 59-71	56.2	600
169	Subgroup-specific structural variation across 1,000 medulloblastoma genomes. *Nature*, **2012**, 488, 49-56	50.4	596
168	Recurrent somatic alterations of FGFR1 and NTRK2 in pilocytic astrocytoma. *Nature Genetics*, **2013**, 45, 927-32	36.3	550
167	The whole-genome landscape of medulloblastoma subtypes. *Nature*, **2017**, 547, 311-317	50.4	472

166	Genome sequencing of SHH medulloblastoma predicts genotype-related response to smoothened inhibition. *Cancer Cell*, **2014**, 25, 393-405	24.3	469
165	Variation in transcription factor binding among humans. *Science*, **2010**, 328, 232-5	33.3	447
164	Toward understanding and exploiting tumor heterogeneity. *Nature Medicine*, **2015**, 21, 846-53	50.5	441
163	Epigenomic alterations define lethal CIMP-positive ependymomas of infancy. *Nature*, **2014**, 506, 445-50	50.4	434
162	What is a gene, post-ENCODE? History and updated definition. *Genome Research*, **2007**, 17, 669-81	9.7	417
161	Assembly and diploid architecture of an individual human genome via single-molecule technologies. *Nature Methods*, **2015**, 12, 780-6	21.6	383
160	Enhancer hijacking activates GFI1 family oncogenes in medulloblastoma. *Nature*, **2014**, 511, 428-34	50.4	377
159	Natural variation in genome architecture among 205 Drosophila melanogaster Genetic Reference Panel lines. *Genome Research*, **2014**, 24, 1193-208	9.7	372
158	Multi-platform discovery of haplotype-resolved structural variation in human genomes. *Nature Communications*, **2019**, 10, 1784	17.4	346
157	Criteria for inference of chromothripsis in cancer genomes. *Cell*, **2013**, 152, 1226-36	56.2	342
156	Phenotypic impact of genomic structural variation: insights from and for human disease. *Nature Reviews Genetics*, **2013**, 14, 125-38	30.1	340
155	Atypical Teratoid/Rhabdoid Tumors Are Comprised of Three Epigenetic Subgroups with Distinct Enhancer Landscapes. *Cancer Cell*, **2016**, 29, 379-393	24.3	319
154	Recurrent mutation of the ID3 gene in Burkitt lymphoma identified by integrated genome, exome and transcriptome sequencing. *Nature Genetics*, **2012**, 44, 1316-20	36.3	317
153	The genetic architecture of Down syndrome phenotypes revealed by high-resolution analysis of human segmental trisomies. *Proceedings of the National Academy of Sciences of the United States of America*, **2009**, 106, 12031-6	11.5	280
152	Eleven grand challenges in single-cell data science. *Genome Biology*, **2020**, 21, 31	18.3	274
151	The genomic and transcriptomic landscape of a HeLa cell line. *G3: Genes, Genomes, Genetics*, **2013**, 3, 1213-24	3.2	269
150	Integrative genomic analyses reveal an androgen-driven somatic alteration landscape in early-onset prostate cancer. *Cancer Cell*, **2013**, 23, 159-70	24.3	259
149	Active medulloblastoma enhancers reveal subgroup-specific cellular origins. *Nature*, **2016**, 530, 57-62	50.4	234

148	Patterns of somatic structural variation in human cancer genomes. *Nature*, **2020**, 578, 112-121	50.4	232
147	Pan-cancer analysis of somatic copy-number alterations implicates IRS4 and IGF2 in enhancer hijacking. *Nature Genetics*, **2017**, 49, 65-74	36.3	220
146	Prediction of effective genome size in metagenomic samples. *Genome Biology*, **2007**, 8, R10	18.3	219
145	A comprehensive map of mobile element insertion polymorphisms in humans. *PLoS Genetics*, **2011**, 7, e1002236	6	218
144	PEMer: a computational framework with simulation-based error models for inferring genomic structural variants from massive paired-end sequencing data. *Genome Biology*, **2009**, 10, R23	18.3	201
143	Intratumor DNA methylation heterogeneity reflects clonal evolution in aggressive prostate cancer. *Cell Reports*, **2014**, 8, 798-806	10.6	177
142	Exome sequencing of osteosarcoma reveals mutation signatures reminiscent of BRCA deficiency. *Nature Communications*, **2015**, 6, 8940	17.4	175
141	Comprehensive analysis of chromothripsis in 2,658 human cancers using whole-genome sequencing. *Nature Genetics*, **2020**, 52, 331-341	36.3	168
140	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	21.7	159
139	High-resolution genomic profiling of chronic lymphocytic leukemia reveals new recurrent genomic alterations. *Blood*, **2012**, 120, 4783-94	2.2	156
138	SHOT: a web server for the construction of genome phylogenies. *Trends in Genetics*, **2002**, 18, 158-62	8.5	156
137	Genomic basis for RNA alterations in cancer. *Nature*, **2020**, 578, 129-136	50.4	148
136	Quantifying environmental adaptation of metabolic pathways in metagenomics. *Proceedings of the National Academy of Sciences of the United States of America*, **2009**, 106, 1374-9	11.5	148
135	Highly rearranged chromosomes reveal uncoupling between genome topology and gene expression. *Nature Genetics*, **2019**, 51, 1272-1282	36.3	145
134	Analysis of genomic context: prediction of functional associations from conserved bidirectionally transcribed gene pairs. *Nature Biotechnology*, **2004**, 22, 911-7	44.5	142
133	Nucleotide-resolution analysis of structural variants using BreakSeq and a breakpoint library. *Nature Biotechnology*, **2010**, 28, 47-55	44.5	136
132	Structured RNAs in the ENCODE selected regions of the human genome. *Genome Research*, **2007**, 17, 852-64	9.7	131
131	Genomics and drug profiling of fatal TCF3-HLF-positive acute lymphoblastic leukemia identifies recurrent mutation patterns and therapeutic options. *Nature Genetics*, **2015**, 47, 1020-1029	36.3	127

130	Pan-cancer analysis of whole genomes identifies driver rearrangements promoted by LINE-1 retrotransposition. *Nature Genetics*, **2020**, 52, 306-319	36.3	122
129	Shadow Enhancers Are Pervasive Features of Developmental Regulatory Networks. *Current Biology*, **2016**, 26, 38-51	6.3	121
128	Genomic deletion of MAP3K7 at 6q12-22 is associated with early PSA recurrence in prostate cancer and absence of TMPRSS2:ERG fusions. *Modern Pathology*, **2013**, 26, 975-83	9.8	121
127	Systematic association of genes to phenotypes by genome and literature mining. *PLoS Biology*, **2005**, 3, e134	9.7	119
126	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. *Genome Research*, **2008**, 18, 1865-74	9.7	114
125	High-resolution mapping of DNA copy alterations in human chromosome 22 using high-density tiling oligonucleotide arrays. *Proceedings of the National Academy of Sciences of the United States of America*, **2006**, 103, 4534-9	11.5	112
124	Positive selection at the protein network periphery: evaluation in terms of structural constraints and cellular context. *Proceedings of the National Academy of Sciences of the United States of America* , **2007**, 104, 20274-9	11.5	111
123	Systematic discovery of analogous enzymes in thiamin biosynthesis. *Nature Biotechnology*, **2003**, 21, 790-5	44.5	109
122	Whole-exome sequencing links caspase recruitment domain 11 (CARD11) inactivation to severe combined immunodeficiency. *Journal of Allergy and Clinical Immunology*, **2013**, 131, 1376-83.e3	11.5	103
121	Haplotype-resolved diverse human genomes and integrated analysis of structural variation. *Science* , **2021**, 372,	33.3	100
120	BAZ2A (TIP5) is involved in epigenetic alterations in prostate cancer and its overexpression predicts disease recurrence. *Nature Genetics*, **2015**, 47, 22-30	36.3	99
119	Analysis of copy number variation in the rhesus macaque genome identifies candidate loci for evolutionary and human disease studies. *Human Molecular Genetics*, **2008**, 17, 1127-36	5.6	96
118	Risk-adapted therapy for young children with medulloblastoma (SJYC07): therapeutic and molecular outcomes from a multicentre, phase 2 trial. *Lancet Oncology, The*, **2018**, 19, 768-784	21.7	95
117	Molecular Evolution of Early-Onset Prostate Cancer Identifies Molecular Risk Markers and Clinical Trajectories. *Cancer Cell*, **2018**, 34, 996-1011.e8	24.3	89
116	A cell-based model system links chromothripsis with hyperploidy. *Molecular Systems Biology*, **2015**, 11, 828	12.2	88
115	High-resolution copy-number variation map reflects human olfactory receptor diversity and evolution. *PLoS Genetics*, **2008**, 4, e1000249	6	87
114	Clinical significance of different types of p53 gene alteration in surgically treated prostate cancer. *International Journal of Cancer*, **2014**, 135, 1369-80	7.5	85
113	Relating CNVs to transcriptome data at fine resolution: assessment of the effect of variant size, type, and overlap with functional regions. *Genome Research*, **2011**, 21, 2004-13	9.7	80

112	The baker's yeast diploid genome is remarkably stable in vegetative growth and meiosis. *PLoS Genetics*, **2010**, 6, e1001109	6	79
111	The current excitement about copy-number variation: how it relates to gene duplications and protein families. *Current Opinion in Structural Biology*, **2008**, 18, 366-74	8.1	78
110	Distinct genomic aberrations associated with ERG rearranged prostate cancer. *Genes Chromosomes and Cancer*, **2009**, 48, 366-80	5	72
109	Data analysis: Create a cloud commons. *Nature*, **2015**, 523, 149-51	50.4	71
108	Systematic prediction and validation of breakpoints associated with copy-number variants in the human genome. *Proceedings of the National Academy of Sciences of the United States of America*, **2007**, 104, 10110-5	11.5	70
107	Pan-cancer analysis of whole genomes		70
106	Primate genome architecture influences structural variation mechanisms and functional consequences. *Proceedings of the National Academy of Sciences of the United States of America*, **2013**, 110, 15764-9	11.5	69
105	Mitochondrial mutations drive prostate cancer aggression. *Nature Communications*, **2017**, 8, 656	17.4	66
104	Global identification and characterization of transcriptionally active regions in the rice genome. *PLoS ONE*, **2007**, 2, e294	3.7	63
103	Comprehensive characterization of complex structural variations in cancer by directly comparing genome sequence reads. *Nature Biotechnology*, **2014**, 32, 1106-12	44.5	62
102	Dense and accurate whole-chromosome haplotyping of individual genomes. *Nature Communications*, **2017**, 8, 1293	17.4	60
101	Analysis of deletion breakpoints from 1,092 humans reveals details of mutation mechanisms. *Nature Communications*, **2015**, 6, 7256	17.4	56
100	Systematic inference of copy-number genotypes from personal genome sequencing data reveals extensive olfactory receptor gene content diversity. *PLoS Computational Biology*, **2010**, 6, e1000988	5	54
99	Impact of genomic structural variation in Drosophila melanogaster based on population-scale sequencing. *Genome Research*, **2013**, 23, 568-79	9.7	53
98	Pediatric T-cell lymphoblastic leukemia evolves into relapse by clonal selection, acquisition of mutations and promoter hypomethylation. *Haematologica*, **2015**, 100, 1442-50	6.6	50
97	Genomic and transcriptomic changes complement each other in the pathogenesis of sporadic Burkitt lymphoma. *Nature Communications*, **2019**, 10, 1459	17.4	49
96	Similar gene expression profiles do not imply similar tissue functions. *Trends in Genetics*, **2006**, 22, 132-8	8.5	49
95	Genetic code expansion for multiprotein complex engineering. *Nature Methods*, **2016**, 13, 997-1000	21.6	48

94	TMPRSS2-ERG fusions are strongly linked to young patient age in low-grade prostate cancer. *European Urology*, **2014**, 66, 978-81	10.2	48
93	Germline Elongator mutations in Sonic Hedgehog medulloblastoma. *Nature*, **2020**, 580, 396-401	50.4	47
92	The molecular landscape of ETMR at diagnosis and relapse. *Nature*, **2019**, 576, 274-280	50.4	46
91	Negative Selection and Chromosome Instability Induced by Mad2 Overexpression Delay Breast Cancer but Facilitate Oncogene-Independent Outgrowth. *Cell Reports*, **2016**, 15, 2679-91	10.6	43
90	Fully phased human genome assembly without parental data using single-cell strand sequencing and long reads. *Nature Biotechnology*, **2021**, 39, 302-308	44.5	42
89	Targeted Perturb-seq enables genome-scale genetic screens in single cells. *Nature Methods*, **2020**, 17, 629-635	21.6	41
88	Comprehensive Analysis of Chromatin States in Atypical Teratoid/Rhabdoid Tumor Identifies Diverging Roles for SWI/SNF and Polycomb in Gene Regulation. *Cancer Cell*, **2019**, 35, 95-110.e8	24.3	40
87	The whole-genome panorama of cancer drivers		38
86	Combining frequency and positional information to predict transcription factor binding sites. *Bioinformatics*, **2001**, 17, 1019-26	7.2	37
85	Leveraging European infrastructures to access 1 million human genomes by 2022. *Nature Reviews Genetics*, **2019**, 20, 693-701	30.1	36
84	The NSL complex maintains nuclear architecture stability via lamin A/C acetylation. *Nature Cell Biology*, **2019**, 21, 1248-1260	23.4	34
83	Pan-cancer analysis distinguishes transcriptional changes of aneuploidy from proliferation. *Genome Research*, **2017**, 27, 501-511	9.7	31
82	A supervised hidden markov model framework for efficiently segmenting tiling array data in transcriptional and chIP-chip experiments: systematically incorporating validated biological knowledge. *Bioinformatics*, **2006**, 22, 3016-24	7.2	31
81	Alterations of microRNA and microRNA-regulated messenger RNA expression in germinal center B-cell lymphomas determined by integrative sequencing analysis. *Haematologica*, **2016**, 101, 1380-1389	6.6	31
80	Challenges in studying genomic structural variant formation mechanisms: the short-read dilemma and beyond. *BioEssays*, **2011**, 33, 840-50	4.1	30
79	Chromatin modifiers Mdm2 and RNF2 prevent RNA:DNA hybrids that impair DNA replication. *Proceedings of the National Academy of Sciences of the United States of America*, **2018**, 115, E11311-E11320	11.5	30
78	MSB: a mean-shift-based approach for the analysis of structural variation in the genome. *Genome Research*, **2009**, 19, 106-17	9.7	29
77	Alfred: interactive multi-sample BAM alignment statistics, feature counting and feature annotation for long- and short-read sequencing. *Bioinformatics*, **2019**, 35, 2489-2491	7.2	29

76	Genomic structural variations lead to dysregulation of important coding and non-coding RNA species in dilated cardiomyopathy. *EMBO Molecular Medicine*, **2018**, 10, 107-120	12	29
75	Sequence diversity analyses of an improved rhesus macaque genome enhance its biomedical utility. *Science*, **2020**, 370,	33.3	28
74	Germline Mutations Predispose to Pediatric Medulloblastoma. *Journal of Clinical Oncology*, **2020**, 38, 43-50	2.2	28
73	Patterns of structural variation in human cancer		26
72	Multi-platform discovery of haplotype-resolved structural variation in human genomes		26
71	Acquisition of chromosome instability is a mechanism to evade oncogene addiction. *EMBO Molecular Medicine*, **2020**, 12, e10941	12	22
70	Identification of ZCCHC8 as fusion partner of ROS1 in a case of congenital glioblastoma multiforme with a t(6;12)(q21;q24.3). *Genes Chromosomes and Cancer*, **2016**, 55, 677-87	5	22
69	The DART classification of unannotated transcription within the ENCODE regions: associating transcription with known and novel loci. *Genome Research*, **2007**, 17, 732-45	9.7	21
68	Deletion lengthening at chromosomes 6q and 16q targets multiple tumor suppressor genes and is associated with an increasingly poor prognosis in prostate cancer. *Oncotarget*, **2017**, 8, 108923-108935	3.3	21
67	A fully phased accurate assembly of an individual human genome		20
66	PDX models recapitulate the genetic and epigenetic landscape of pediatric T-cell leukemia. *EMBO Molecular Medicine*, **2018**, 10,	12	20
65	Heterogeneity of ERG expression in prostate cancer: a large section mapping study of entire prostatectomy specimens from 125 patients. *BMC Cancer*, **2016**, 16, 641	4.8	19
64	Genomics: data sharing needs an international code of conduct. *Nature*, **2020**, 578, 31-33	50.4	17
63	Strand-seq enables reliable separation of long reads by chromosome via expectation maximization. *Bioinformatics*, **2018**, 34, i115-i123	7.2	17
62	Computing patient data in the cloud: practical and legal considerations for genetics and genomics research in Europe and internationally. *Genome Medicine*, **2017**, 9, 58	14.4	17
61	Extracting information from cDNA arrays. *Chaos*, **2001**, 11, 98-107	3.3	17
60	Immortalization capacity of HPV types is inversely related to chromosomal instability. *Oncotarget*, **2016**, 7, 37608-37621	3.3	17
59	Single-cell analysis of structural variations and complex rearrangements with tri-channel processing. *Nature Biotechnology*, **2020**, 38, 343-354	44.5	17

58	A scalable CRISPR/Cas9-based fluorescent reporter assay to study DNA double-strand break repair choice. *Nature Communications*, **2020**, 11, 4077	17.4	15
57	Large-Scale Uniform Analysis of Cancer Whole Genomes in Multiple Computing Environments		14
56	Recurrent inversion toggling and great ape genome evolution. *Nature Genetics*, **2020**, 52, 849-858	36.3	13
55	Use of pathway analysis and genome context methods for functional genomics of Mycoplasma pneumoniae nucleotide metabolism. *Gene*, **2007**, 396, 215-25	3.8	13
54	Germline determinants of the somatic mutation landscape in 2,642 cancer genomes		13
53	Expectations and blind spots for structural variation detection from long-read assemblies and short-read genome sequencing technologies. *American Journal of Human Genetics*, **2021**, 108, 919-928	11	13
52	Identification of a Ninein (NIN) mutation in a family with spondyloepimetaphyseal dysplasia with joint laxity (leptodactylic type)-like phenotype. *Matrix Biology*, **2013**, 32, 387-92	11.4	12
51	Genome-wide Screens Implicate Loss of Cullin Ring Ligase 3 in Persistent Proliferation and Genome Instability in TP53-Deficient Cells. *Cell Reports*, **2020**, 31, 107465	10.6	11
50	Genomic data sharing in Europe is stumbling-Could a code of conduct prevent its fall?. *EMBO Molecular Medicine*, **2020**, 12, e11421	12	11
49	Pangenome-based genome inference		11
48	Pan-cancer analysis of whole genomes reveals driver rearrangements promoted by LINE-1 retrotransposition in human tumours		10
47	Genomic basis for RNA alterations revealed by whole-genome analyses of 27 cancer types		10
46	Prevalence of chromosomal rearrangements involving non-ETS genes in prostate cancer. *International Journal of Oncology*, **2015**, 46, 1637-42	4.4	9
45	AlphaDesign: A de novo protein design framework based on AlphaFold		9
44	Butler enables rapid cloud-based analysis of thousands of human genomes. *Nature Biotechnology*, **2020**, 38, 288-292	44.5	9
43	VISOR: a versatile haplotype-aware structural variant simulator for short- and long-read sequencing. *Bioinformatics*, **2020**, 36, 1267-1269	7.2	8
42	InTAD: chromosome conformation guided analysis of enhancer target genes. *BMC Bioinformatics*, **2019**, 20, 60	3.6	8
41	Next-generation sequencing-based detection of germline L1-mediated transductions. *BMC Genomics*, **2016**, 17, 342	4.5	7

40	Genomes of early onset prostate cancer. *Current Opinion in Urology*, **2017**, 27, 481-487	2.8	7
39	Chromothripsis followed by circular recombination drives oncogene amplification in human cancer. *Nature Genetics*, **2021**, 53, 1673-1685	36.3	7
38	TRiCoLOR: tandem repeat profiling using whole-genome long-read sequencing data. *GigaScience*, **2020**, 9,	7.6	7
37	A 15q24 microdeletion in transient myeloproliferative disease (TMD) and acute megakaryoblastic leukaemia (AMKL) implicates PML and SUMO3 in the leukaemogenesis of TMD/AMKL. *British Journal of Haematology*, **2012**, 157, 180-7	4.5	6
36	The Helmholtz Network for Bioinformatics: an integrative web portal for bioinformatics resources. *Bioinformatics*, **2004**, 20, 268-70	7.2	6
35	Framework for quality assessment of whole genome, cancer sequences		6
34	Transgene methylation in mice reflects copy number but not expression level. *Molecular Biotechnology*, **2004**, 26, 215-20	3	5
33	Systems approaches identify the consequences of monosomy in somatic human cells. *Nature Communications*, **2021**, 12, 5576	17.4	5
32	Single-cell strand sequencing of a macaque genome reveals multiple nested inversions and breakpoint reuse during primate evolution. *Genome Research*, **2020**, 30, 1680-1693	9.7	4
31	Using large-scale genome variation cohorts to decipher the molecular mechanism of cancer. *Comptes Rendus - Biologies*, **2016**, 339, 308-13	1.4	4
30	The genomic and transcriptional landscape of primary central nervous system lymphoma.. *Nature Communications*, **2022**, 13, 2558	17.4	4
29	Expectations and blind spots for structural variation detection from short-read alignment and long-read assembly		3
28	De novo assembly of 64 haplotype-resolved human genomes of diverse ancestry and integrated analysis of structural variation		3
27	Chromatin accessibility landscape of pediatric T-lymphoblastic leukemia and human T-cell precursors. *EMBO Molecular Medicine*, **2020**, 12, e12104	12	3
26	Mutational mechanisms shaping the coding and noncoding genome of germinal center derived B-cell lymphomas. *Leukemia*, **2021**, 35, 2002-2016	10.7	3
25	Automated assembly of high-quality diploid human reference genomes		3
24	Enriched power of disease-concordant twin-case-only design in detecting interactions in genome-wide association studies. *European Journal of Human Genetics*, **2019**, 27, 631-636	5.3	2
23	A common microdeletion affecting a hippocampus- and amygdala-specific isoform of tryptophan hydroxylase 2 is not associated with affective disorders. *Bipolar Disorders*, **2014**, 16, 764-8	3.8	2

22	12 Grand Challenges in Single-Cell Data Science		2
21	Dense and accurate whole-chromosome haplotyping of individual genomes		2
20	Enabling rapid cloud-based analysis of thousands of human genomes via Butler		2
19	Versatile workflow for cell type-resolved transcriptional and epigenetic profiles from cryopreserved human lung. *JCI Insight*, **2021**, 6,	9.9	2
18	ASHLEYS: automated quality control for single-cell Strand-seq data. *Bioinformatics*, **2021**,	7.2	2
17	Systems approaches identify the consequences of monosomy in somatic human cells		2
16	Haplotype-resolved inversion landscape reveals hotspots of mutational recurrence associated with genomic disorders		2
15	Recurrent inversion polymorphisms in humans associate with genetic instability and genomic disorders.. *Cell*, **2022**,	56.2	2
14	Systematic Identification of Determinants for Single-Strand Annealing-Mediated Deletion Formation in. *G3: Genes, Genomes, Genetics*, **2017**, 7, 3269-3279	3.2	1
13	ICGC PedBrain - dissecting the genomic complexity underlying medulloblastoma using whole-genome sequencing. *BMC Proceedings*, **2012**, 6,	2.3	1
12	Somatic structural variant formation is guided by and influences genome architecture		1
11	The X chromosome from telomere to telomere: key achievements and future opportunities.. *Faculty Reviews*, **2021**, 10, 63	1.2	1
10	The Porto European Cancer Research Summit 2021. *Molecular Oncology*, **2021**, 15, 2507-2543	7.9	1
9	Somatic structural variant formation is guided by and influences genome architecture.. *Genome Research*, **2022**,	9.7	1
8	Pangenome-based genome inference allows efficient and accurate genotyping across a wide spectrum of variant classes.. *Nature Genetics*, **2022**,	36.3	1
7	Potenzial und Herausforderungen der personalisierten Genomik und des 1000-Genom-Projekts. *Medizinische Genetik*, **2010**, 22, 242-247	0.5	
6	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	2.2	
5	Targeted Deep Sequencing of Genetic Alterations Identified By Whole Exome Sequencing Reveals Clonal Evolution in Pediatric T-Lymphoblastic Leukemia. *Blood*, **2014**, 124, 491-491	2.2	

4	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	2.2
3	Whole-Exome Sequencing Links CARD11 Inactivation with SCID. *Blood*, **2012**, 120, 258-258	2.2
2	Whole Exome Sequencing Identifies Novel Lyst-Missense Mutations In Incomplete Childhood Chediak-Higashi-Syndrome Presenting As Hemphagocytic Lymphohistiocytosis (HLH). *Blood*, **2013**, 122, 3479-3479	2.2
1	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	2.2