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List of Publications by Year in Descending Order

83 papers	32,924 citations	44 h-index	93 g-index
93 ext. papers	40,426 ext. citations	18.6 avg, IF	6.46 L-index

#	Paper	IF	Citations
83	Somatic mosaicism reveals clonal distributions of neocortical development.. *Nature*, **2022**,	50.4	1
82	SquiggleNet: real-time, direct classification of nanopore signals. *Genome Biology*, **2021**, 22, 298	18.3	1
81	Early HPV ctDNA Kinetics and Imaging Biomarkers Predict Therapeutic Response in p16+ Oropharyngeal Squamous Cell Carcinoma. *Clinical Cancer Research*, **2021**,	12.9	2
80	Comprehensive identification of somatic nucleotide variants in human brain tissue. *Genome Biology*, **2021**, 22, 92	18.3	3
79	Haplotype-resolved diverse human genomes and integrated analysis of structural variation. *Science* , **2021**, 372,	33.3	100
78	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
77	SearcHPV: A novel approach to identify and assemble human papillomavirus-host genomic integration events in cancer. *Cancer*, **2021**, 127, 3531-3540	6.4	2
76	Cas9 targeted enrichment of mobile elements using nanopore sequencing. *Nature Communications*, **2021**, 12, 3586	17.4	10
75	Genome diversity in Ukraine. *GigaScience*, **2021**, 10,	7.6	5
74	The landscape of somatic mutation in cerebral cortex of autistic and neurotypical individuals revealed by ultra-deep whole-genome sequencing. *Nature Neuroscience*, **2021**, 24, 176-185	25.5	19
73	Machine learning reveals bilateral distribution of somatic L1 insertions in human neurons and glia. *Nature Neuroscience*, **2021**, 24, 186-196	25.5	9
72	A robust benchmark for detection of germline large deletions and insertions. *Nature Biotechnology*, **2020**, 38, 1347-1355	44.5	98
71	Characterization of nuclear mitochondrial insertions in the whole genomes of primates. *NAR Genomics and Bioinformatics*, **2020**, 2, lqaa089	3.7	1
70	Identification and characterization of occult human-specific LINE-1 insertions using long-read sequencing technology. *Nucleic Acids Research*, **2020**, 48, 1146-1163	20.1	36
69	Structural variation in the sequencing era. *Nature Reviews Genetics*, **2020**, 21, 171-189	30.1	129
68	Association of CNVs with methylation variation. *Npj Genomic Medicine*, **2020**, 5, 41	6.2	4
67	Translation of upstream open reading frames in a model of neuronal differentiation. *BMC Genomics* , **2019**, 20, 391	4.5	16

66	Multi-platform discovery of haplotype-resolved structural variation in human genomes. *Nature Communications*, **2019**, 10, 1784	17.4	346
65	Constitutively Higher Level of GSTT2 in Esophageal Tissues From African Americans Protects Cells Against DNA Damage. *Gastroenterology*, **2019**, 156, 1404-1415	13.3	5
64	RNA ligation precedes the retrotransposition of U6/LINE-1 chimeric RNA. *Proceedings of the National Academy of Sciences of the United States of America*, **2019**, 116, 20612-20622	11.5	14
63	Prognostic model for multiple myeloma progression integrating gene expression and clinical features. *GigaScience*, **2019**, 8,	7.6	9
62	FusorSV: an algorithm for optimally combining data from multiple structural variation detection methods. *Genome Biology*, **2018**, 19, 38	18.3	28
61	BAMnostic: an OS-agnostic toolkit for genomic sequence analysis. *Journal of Open Source Software*, **2018**, 3, 826	5.2	
60	Intersection of diverse neuronal genomes and neuropsychiatric disease: The Brain Somatic Mosaicism Network. *Science*, **2017**, 356,	33.3	152
59	A recurrence-based approach for validating structural variation using long-read sequencing technology. *GigaScience*, **2017**, 6, 1-9	7.6	13
58	Rapid, ultra low coverage copy number profiling of cell-free DNA as a precision oncology screening strategy. *Oncotarget*, **2017**, 8, 89848-89866	3.3	36
57	The Mobile Element Locator Tool (MELT): population-scale mobile element discovery and biology. *Genome Research*, **2017**, 27, 1916-1929	9.7	156
56	Genetics of Combined Pituitary Hormone Deficiency: Roadmap into the Genome Era. *Endocrine Reviews*, **2016**, 37, 636-675	27.2	106
55	SPECtre: a spectral coherence--based classifier of actively translated transcripts from ribosome profiling sequence data. *BMC Bioinformatics*, **2016**, 17, 482	3.6	31
54	Resolving complex structural genomic rearrangements using a randomized approach. *Genome Biology*, **2016**, 17, 126	18.3	24
53	A global reference for human genetic variation. *Nature*, **2015**, 526, 68-74	50.4	8599
52	An integrated map of structural variation in 2,504 human genomes. *Nature*, **2015**, 526, 75-81	50.4	1368
51	CodonShuffle: a tool for generating and analyzing synonymously mutated sequences. *Virus Evolution*, **2015**, 1, vev012	3.7	15
50	Cryptic and complex chromosomal aberrations in early-onset neuropsychiatric disorders. *American Journal of Human Genetics*, **2014**, 95, 454-61	11	37
49	Does germ-line deletion of the PIP gene constitute a widespread risk for cancer?. *European Journal of Human Genetics*, **2014**, 22, 307-9	5.3	4

48	Integrating sequence and array data to create an improved 1000 Genomes Project haplotype reference panel. *Nature Communications*, **2014**, 5, 3934	17.4	253
47	Human genomic regions with exceptionally high levels of population differentiation identified from 911 whole-genome sequences. *Genome Biology*, **2014**, 15, R88	18.3	51
46	Increased genomic integrity of an improved protein-based mouse induced pluripotent stem cell method compared with current viral-induced strategies. *Stem Cells Translational Medicine*, **2014**, 3, 599-609	6.9	20
45	The genomic landscape of polymorphic human nuclear mitochondrial insertions. *Nucleic Acids Research*, **2014**, 42, 12640-9	20.1	105
44	Copy number variation genotyping using family information. *BMC Bioinformatics*, **2013**, 14, 157	3.6	6
43	Integrative annotation of variants from 1092 humans: application to cancer genomics. *Science*, **2013**, 342, 1235587	33.3	281
42	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
41	CGG repeat-associated translation mediates neurodegeneration in fragile X tremor ataxia syndrome. *Neuron*, **2013**, 78, 440-55	13.9	327
40	The origin, evolution, and functional impact of short insertion-deletion variants identified in 179 human genomes. *Genome Research*, **2013**, 23, 749-61	9.7	150
39	Analysis of variable retroduplications in human populations suggests coupling of retrotransposition to cell division. *Genome Research*, **2013**, 23, 2042-52	9.7	41
38	Copy number variation prevalence in known asthma genes and their impact on asthma susceptibility. *Clinical and Experimental Allergy*, **2013**, 43, 455-62	4.1	16
37	Complex reorganization and predominant non-homologous repair following chromosomal breakage in karyotypically balanced germline rearrangements and transgenic integration. *Nature Genetics*, **2012**, 44, 390-7, S1	36.3	190
36	The 1000 Genomes Project: data management and community access. *Nature Methods*, **2012**, 9, 459-62	21.6	202
35	An integrated map of genetic variation from 1,092 human genomes. *Nature*, **2012**, 491, 56-65	50.4	6049
34	Deleterious- and disease-allele prevalence in healthy individuals: insights from current predictions, mutation databases, and population-scale resequencing. *American Journal of Human Genetics*, **2012**, 91, 1022-32	11	221
33	Regulatory element copy number differences shape primate expression profiles. *Proceedings of the National Academy of Sciences of the United States of America*, **2012**, 109, 12656-61	11.5	27
32	A systematic survey of loss-of-function variants in human protein-coding genes. *Science*, **2012**, 335, 823-8	33.3	880
31	Extensive genetic diversity and substructuring among zebrafish strains revealed through copy number variant analysis. *Proceedings of the National Academy of Sciences of the United States of America*, **2012**, 109, 529-34	11.5	87

30	Refinement of primate copy number variation hotspots identifies candidate genomic regions evolving under positive selection. *Genome Biology*, **2011**, 12, R52	18.3	48
29	The functional spectrum of low-frequency coding variation. *Genome Biology*, **2011**, 12, R84	18.3	161
28	Mapping copy number variation by population-scale genome sequencing. *Nature*, **2011**, 470, 59-65	50.4	833
27	Comprehensive assessment of array-based platforms and calling algorithms for detection of copy number variants. *Nature Biotechnology*, **2011**, 29, 512-20	44.5	333
26	Variation in genome-wide mutation rates within and between human families. *Nature Genetics*, **2011**, 43, 712-4	36.3	404
25	Demographic history and rare allele sharing among human populations. *Proceedings of the National Academy of Sciences of the United States of America*, **2011**, 108, 11983-8	11.5	455
24	Natural genetic variation caused by small insertions and deletions in the human genome. *Genome Research*, **2011**, 21, 830-9	9.7	178
23	Expanding the definition of the classical bipartite nuclear localization signal. *Traffic*, **2010**, 11, 311-23	5.7	83
22	A map of human genome variation from population-scale sequencing. *Nature*, **2010**, 467, 1061-73	50.4	6142
21	Discovery of common Asian copy number variants using integrated high-resolution array CGH and massively parallel DNA sequencing. *Nature Genetics*, **2010**, 42, 400-5	36.3	167
20	Small insertions and deletions (INDELs) in human genomes. *Human Molecular Genetics*, **2010**, 19, R131-6	5.6	221
19	Natural mutagenesis of human genomes by endogenous retrotransposons. *Cell*, **2010**, 141, 1253-61	56.2	445
18	Diversity of human copy number variation and multicopy genes. *Science*, **2010**, 330, 641-6	33.3	491
17	A highly annotated whole-genome sequence of a Korean individual. *Nature*, **2009**, 460, 1011-5	50.4	265
16	A PY-NLS nuclear targeting signal is required for nuclear localization and function of the Saccharomyces cerevisiae mRNA-binding protein Hrp1. *Journal of Biological Chemistry*, **2008**, 283, 12926-34	5.4	34
15	Active Alu retrotransposons in the human genome. *Genome Research*, **2008**, 18, 1875-83	9.7	192
14	Which transposable elements are active in the human genome?. *Trends in Genetics*, **2007**, 23, 183-91	8.5	333
13	Classical nuclear localization signals: definition, function, and interaction with importin alpha. *Journal of Biological Chemistry*, **2007**, 282, 5101-5	5.4	824

12	An initial map of insertion and deletion (INDEL) variation in the human genome. *Genome Research*, **2006**, 16, 1182-90	9.7	455
11	Recently mobilized transposons in the human and chimpanzee genomes. *American Journal of Human Genetics*, **2006**, 78, 671-9	11	118
10	Identification of proteins associated with murine cytomegalovirus virions. *Journal of Virology*, **2004**, 78, 11187-97	6.6	121
9	Prokaryotic gene prediction using GeneMark and GeneMark.hmm. *Current Protocols in Bioinformatics*, **2003**, Chapter 4, Unit4.5	24.2	60
8	Eukaryotic gene prediction using GeneMark.hmm. *Current Protocols in Bioinformatics*, **2003**, Chapter 4, Unit4.6	24.2	11
7	Complete sequence and comparative analysis of the genome of herpes B virus (Cercopithecine herpesvirus 1) from a rhesus monkey. *Journal of Virology*, **2003**, 77, 6167-77	6.6	76
6	Improving gene annotation of complete viral genomes. *Nucleic Acids Research*, **2003**, 31, 7041-55	20.1	38
5	Multi-platform discovery of haplotype-resolved structural variation in human genomes		26
4	Expectations and blind spots for structural variation detection from short-read alignment and long-read assembly		3
3	De novo assembly of 64 haplotype-resolved human genomes of diverse ancestry and integrated analysis of structural variation		3
2	A robust benchmark for germline structural variant detection		34
1	Real-Time, Direct Classification of Nanopore Signals with SquiggleNet		1