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List of Publications by Year
in descending order

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78	44,859	50170 46	66788 78
papers	citations	h-index	g-index

93	93	93	64903
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	A global reference for human genetic variation. Nature, 2015, 526, 68-74.	13.7	13,998
2	A map of human genome variation from population-scale sequencing. Nature, 2010, 467, 1061-1073.	13.7	7,209
3	An integrated map of genetic variation from 1,092 human genomes. Nature, 2012, 491, 56-65.	13.7	7,199
4	An integrated map of structural variation in 2,504 human genomes. Nature, 2015, 526, 75-81.	13.7	1,994
5	A Systematic Survey of Loss-of-Function Variants in Human Protein-Coding Genes. Science, 2012, 335, 823-828.	6.0	1,095
6	Mapping copy number variation by population-scale genome sequencing. Nature, 2011, 470, 59-65.	13.7	991
7	Classical Nuclear Localization Signals: Definition, Function, and Interaction with Importin Î±*. Journal of Biological Chemistry, 2007, 282, 5101-5105.	1.6	966
8	Multi-platform discovery of haplotype-resolved structural variation in human genomes. Nature Communications, 2019, 10, 1784.	5.8	636
9	Diversity of Human Copy Number Variation and Multicopy Genes. Science, 2010, 330, 641-646.	6.0	609
10	Demographic history and rare allele sharing among human populations. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 11983-11988.	3.3	589
11	An initial map of insertion and deletion (INDEL) variation in the human genome. Genome Research, 2006, 16, 1182-1190.	2.4	548
12	Variation in genome-wide mutation rates within and between human families. Nature Genetics, 2011, 43, 712-714.	9.4	525
13	Natural Mutagenesis of Human Genomes by Endogenous Retrotransposons. Cell, 2010, 141, 1253-1261.	13.5	513
14	CGG Repeat-Associated Translation Mediates Neurodegeneration in Fragile X Tremor Ataxia Syndrome. Neuron, 2013, 78, 440-455.	3.8	422
15	Which transposable elements are active in the human genome?. Trends in Genetics, 2007, 23, 183-191.	2.9	406
16	Comprehensive assessment of array-based platforms and calling algorithms for detection of copy number variants. Nature Biotechnology, 2011, 29, 512-520.	9.4	384
17	Integrating sequence and array data to create an improved 1000 Genomes Project haplotype reference panel. Nature Communications, 2014, 5, 3934.	5.8	364
18	Haplotype-resolved diverse human genomes and integrated analysis of structural variation. Science, 2021, 372, .	6.0	358

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19	Integrative Annotation of Variants from 1092 Humans: Application to Cancer Genomics. Science, 2013, 342, 1235587.	6.0	341
20	Structural variation in the sequencing era. Nature Reviews Genetics, 2020, 21, 171-189.	7.7	337
21	The 1000 Genomes Project: data management and community access. Nature Methods, 2012, 9, 459-462.	9.0	308
22	A highly annotated whole-genome sequence of a Korean individual. Nature, 2009, 460, 1011-1015.	13.7	295
23	Small insertions and deletions (INDELs) in human genomes. Human Molecular Genetics, 2010, 19, R131-R136.	1.4	286
24	The Mobile Element Locator Tool (MELT): population-scale mobile element discovery and biology. Genome Research, 2017, 27, 1916-1929.	2.4	273
25	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, 1022-1032.	2.6	255
26	A robust benchmark for detection of germline large deletions and insertions. Nature Biotechnology, 2020, 38, 1347-1355.	9.4	233
27	Active Alu retrotransposons in the human genome. Genome Research, 2008, 18, 1875-1883.	2.4	230
28	Complex reorganization and predominant non-homologous repair following chromosomal breakage in karyotypically balanced germline rearrangements and transgenic integration. Nature Genetics, 2012, 44, 390-397.	9.4	229
29	Natural genetic variation caused by small insertions and deletions in the human genome. Genome Research, 2011, 21, 830-839.	2.4	212
30	The origin, evolution, and functional impact of short insertionâ€“deletion variants identified in 179 human genomes. Genome Research, 2013, 23, 749-761.	2.4	206
31	Intersection of diverse neuronal genomes and neuropsychiatric disease: The Brain Somatic Mosaicism Network. Science, 2017, 356, .	6.0	206
32	Discovery of common Asian copy number variants using integrated high-resolution array CGH and massively parallel DNA sequencing. Nature Genetics, 2010, 42, 400-405.	9.4	179
33	The functional spectrum of low-frequency coding variation. Genome Biology, 2011, 12, R84.	13.9	173
34	The genomic landscape of polymorphic human nuclear mitochondrial insertions. Nucleic Acids Research, 2014, 42, 12640-12649.	6.5	168
35	Genetics of Combined Pituitary Hormone Deficiency: Roadmap into the Genome Era. Endocrine Reviews, 2016, 37, 636-675.	8.9	147
36	Identification of Proteins Associated with Murine Cytomegalovirus Virions. Journal of Virology, 2004, 78, 11187-11197.	1.5	138

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37	Recently Mobilized Transposons in the Human and Chimpanzee Genomes. American Journal of Human Genetics, 2006, 78, 671-679.	2.6	136
38	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, 529-534.	3.3	102
39	Expanding the Definition of the Classical Bipartite Nuclear Localization Signal. Traffic, 2010, 11, 311-323.	1.3	94
40	Complete Sequence and Comparative Analysis of the Genome of Herpes B Virus (Cercopithecine) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50	1.5	88
41	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, 15764-15769.	3.3	80
42	Prokaryotic Gene Prediction Using GeneMark and GeneMark.hmm. Current Protocols in Bioinformatics, 2003, 1, 4.5.1-4.5.16.	25.8	76
43	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.	7.1	73
44	Human genomic regions with exceptionally high levels of population differentiation identified from 911 whole-genome sequences. Genome Biology, 2014, 15, R88.	13.9	72
45	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.	2.6	72
46	Identification and characterization of occult human-specific LINE-1 insertions using long-read sequencing technology. Nucleic Acids Research, 2020, 48, 1146-1163.	6.5	68
47	Refinement of primate copy number variationhotspots identifies candidate genomic regions evolving under positive selection. Genome Biology, 2011, 12, R52.	3.8	58
48	Analysis of variable retroduplications in human populations suggests coupling of retrotransposition to cell division. Genome Research, 2013, 23, 2042-2052.	2.4	52
49	FusorSV: an algorithm for optimally combining data from multiple structural variation detection methods. Genome Biology, 2018, 19, 38.	3.8	46
50	Improving gene annotation of complete viral genomes. Nucleic Acids Research, 2003, 31, 7041-7055.	6.5	45
51	Cryptic and Complex Chromosomal Aberrations in Early-Onset Neuropsychiatric Disorders. American Journal of Human Genetics, 2014, 95, 454-461.	2.6	45
52	Rapid, ultra low coverage copy number profiling of cell-free DNA as a precision oncology screening strategy. Oncotarget, 2017, 8, 89848-89866.	0.8	45
53	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, 12926-12934.	1.6	43
54	SPECtre: a spectral coherence-Âbased classifier of actively translated transcripts from ribosome profiling sequence data. BMC Bioinformatics, 2016, 17, 482.	1.2	41

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55	Early HPV ctDNA Kinetics and Imaging Biomarkers Predict Therapeutic Response in p16+ Oropharyngeal Squamous Cell Carcinoma. Clinical Cancer Research, 2022, 28, 350-359.	3.2	38
56	Regulatory element copy number differences shape primate expression profiles. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, 12656-12661.	3.3	37
57	Resolving complex structural genomic rearrangements using a randomized approach. Genome Biology, 2016, 17, 126.	3.8	36
58	Cas9 targeted enrichment of mobile elements using nanopore sequencing. Nature Communications, 2021, 12, 3586.	5.8	33
59	SquiggleNet: real-time, direct classification of nanopore signals. Genome Biology, 2021, 22, 298.	3.8	33
60	Translation of upstream open reading frames in a model of neuronal differentiation. BMC Genomics, 2019, 20, 391.	1.2	30
61	Comprehensive identification of somatic nucleotide variants in human brain tissue. Genome Biology, 2021, 22, 92.	3.8	26
62	Somatic mosaicism reveals clonal distributions of neocortical development. Nature, 2022, 604, 689-696.	13.7	26
63	Copy number variation prevalence in known asthma genes and their impact on asthma susceptibility. Clinical and Experimental Allergy, 2013, 43, 455-462.	1.4	25
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.	3.3	23
65	A recurrence-based approach for validating structural variation using long-read sequencing technology. GigaScience, 2017, 6, 1-9.	3.3	22
66	Machine learning reveals bilateral distribution of somatic L1 insertions in human neurons and glia. Nature Neuroscience, 2021, 24, 186-196.	7.1	22
67	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.	1.6	21
68	CodonShuffle: a tool for generating and analyzing synonymously mutated sequences. Virus Evolution, 2015, 1, vev012.	2.2	20
69	Prognostic model for multiple myeloma progression integrating gene expression and clinical features. GigaScience, 2019, 8, .	3.3	17
70	Association of CNVs with methylation variation. Npj Genomic Medicine, 2020, 5, 41.	1.7	17
71	Eukaryotic Gene Prediction Using GeneMark.hmm. Current Protocols in Bioinformatics, 2003, 1, 4.6.1-4.6.12.	25.8	15
72	Constitutively Higher Level of GSTT2 in Esophageal Tissues From African Americans Protects Cells Against DNA Damage. Gastroenterology, 2019, 156, 1404-1415.	0.6	15

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73	Characterization of nuclear mitochondrial insertions in the whole genomes of primates. NAR Genomics and Bioinformatics, 2020, 2, lqaa089.	1.5	14
74	Genome diversity in Ukraine. GigaScience, 2021, 10, .	3.3	9
75	SearcHPV: A novel approach to identify and assemble human papillomavirusâ€“host genomic integration events in cancer. Cancer, 2021, 127, 3531-3540.	2.0	8
76	Copy number variation genotyping using family information. BMC Bioinformatics, 2013, 14, 157.	1.2	7
77	Does germ-line deletion of the PIP gene constitute a widespread risk for cancer?. European Journal of Human Genetics, 2014, 22, 307-309.	1.4	4
78	BAMnostic: an OS-agnostic toolkit for genomic sequence analysis. Journal of Open Source Software, 2018, 3, 826.	2.0	0