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List of Publications by Year
in descending order

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52 papers	62,782 citations	100601 38 h-index	206121 51 g-index
54 all docs	54 docs citations	54 times ranked	82720 citing authors

#	Article	IF	Citations
1	Haplotyping the Vitis collinear core genome with rhAmpSeq improves marker transferability in a diverse genus. Nature Communications, 2020, 11, 413.	5.8	52
2	Thousands of human sequences provide deep insight into single genomes. Nature, 2020, 581, 385-386.	13.7	1
3	A general approach for detecting expressed mutations in AML cells using single cell RNA-sequencing. Nature Communications, 2019, 10, 3660.	5.8	147
4	The emergent landscape of the mouse gut endoderm at single-cell resolution. Nature, 2019, 569, 361-367.	13.7	285
5	Multi-platform discovery of haplotype-resolved structural variation in human genomes. Nature Communications, 2019, 10, 1784.	5.8	636
6	Resolving the full spectrum of human genome variation using Linked-Reads. Genome Research, 2019, 29, 635-645.	2.4	182
7	Genomes for all. Nature Biotechnology, 2018, 36, 815-816.	9.4	5
8	Evaluation of GRCh38 and de novo haploid genome assemblies demonstrates the enduring quality of the reference assembly. Genome Research, 2017, 27, 849-864.	2.4	728
9	Direct determination of diploid genome sequences. Genome Research, 2017, 27, 757-767.	2.4	728
10	Principles and Recommendations for Standardizing the Use of the Next-Generation Sequencing Variant File in Clinical Settings. Journal of Molecular Diagnostics, 2017, 19, 417-426.	1.2	19
11	Assembly: a resource for assembled genomes at NCBI. Nucleic Acids Research, 2016, 44, D73-D80.	6.5	292
12	Mitochondrial Disease Sequence Data Resource (MSeqDR): A global grass-roots consortium to facilitate deposition, curation, annotation, and integrated analysis of genomic data for the mitochondrial disease clinical and research communities. Molecular Genetics and Metabolism, 2015, 114, 388-396.	0.5	76
13	Good laboratory practice for clinical next-generation sequencing informatics pipelines. Nature Biotechnology, 2015, 33, 689-693.	9.4	134
14	A global reference for human genetic variation. Nature, 2015, 526, 68-74.	13.7	13,998
15	ClinVar: public archive of relationships among sequence variation and human phenotype. Nucleic Acids Research, 2014, 42, D980-D985.	6.5	2,270
16	Single haplotype assembly of the human genome from a hydatidiform mole. Genome Research, 2014, 24, 2066-2076.	2.4	133
17	An evidence-based approach to establish the functional and clinical significance of copy number variants in intellectual and developmental disabilities. Genetics in Medicine, 2011, 13, 777-784.	1.1	371
18	Modernizing Reference Genome Assemblies. PLoS Biology, 2011, 9, e1001091.	2.6	458

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19	Consensus Statement: Chromosomal Microarray Is a First-Tier Clinical Diagnostic Test for Individuals with Developmental Disabilities or Congenital Anomalies. American Journal of Human Genetics, 2010, 86, 749-764.	2.6	2,325
20	Public data archives for genomic structural variation. Nature Genetics, 2010, 42, 813-814.	9.4	71
21	Lineage-Specific Biology Revealed by a Finished Genome Assembly of the Mouse. PLoS Biology, 2009, 7, e1000112.	2.6	419
22	Mouse segmental duplication and copy number variation. Nature Genetics, 2008, 40, 909-914.	9.4	209
23	Completing the map of human genetic variation. Nature, 2007, 447, 161-165.	13.7	178
24	Identification and analysis of functional elements in 1% of the human genome by the ENCODE pilot project. Nature, 2007, 447, 799-816.	13.7	4,709
25	WebaCGH. Applied Bioinformatics, 2006, 5, 125-130.	1.7	5
26	Candidate Single Nucleotide Polymorphism Selection using Publicly Available Tools: A Guide for Epidemiologists. American Journal of Epidemiology, 2006, 164, 794-804.	1.6	49
27	A genome-wide comparison of recent chimpanzee and human segmental duplications. Nature, 2005, 437, 88-93.	13.7	353
28	Analysis of Segmental Duplications and Genome Assembly in the Mouse. Genome Research, 2004, 14, 789-801.	2.4	106
29	Shotgun sequence assembly and recent segmental duplications within the human genome. Nature, 2004, 431, 927-930.	13.7	228
30	Cross-Species Sequence Comparisons: A Review of Methods and Available Resources. Genome Research, 2003, 13, 1-12.	2.4	210
31	Sequence variations in the public human genome data reflect a bottlenecked population history. Proceedings of the National Academy of Sciences of the United States of America, 2003, 100, 376-381.	3.3	113
32	Connecting Sequence and Biology in the Laboratory Mouse. Genome Research, 2003, 13, 1505-1519.	2.4	18
33	Database resources of the National Center for Biotechnology. Nucleic Acids Research, 2003, 31, 28-33.	6.5	879
34	Database resources of the National Center for Biotechnology Information: 2002 update. Nucleic Acids Research, 2002, 30, 13-16.	6.5	184
35	Accessing the Human Genome. Current Protocols in Human Genetics, 2002, 34, Unit 6.9.	3.5	0
36	Initial sequencing and comparative analysis of the mouse genome. Nature, 2002, 420, 520-562.	13.7	6,319

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37	A radiation hybrid map of mouse genes. Nature Genetics, 2001, 29, 201-205.	9.4	93
38	Initial sequencing and analysis of the human genome. Nature, 2001, 409, 860-921.	13.7	21,074
39	Spidey: A Tool for mRNA-to-Genomic Alignments. Genome Research, 2001, 11, 1952-1957.	2.4	285
40	Mouse Chromosome 2. Mammalian Genome, 1999, 10, 941-941.	1.0	39
41	[6] Gene identification by exon amplification. Methods in Enzymology, 1999, 303, 83-99.	0.4	14
42	Transcript Mapping of the Human Chromosome 11q12â€“q13.1 Gene-Rich Region Identifies Several Newly Described Conserved Genes. Genomics, 1998, 49, 419-429.	1.3	16
43	A High-Resolution Physical and Transcript Map of the Cri du Chat Region of Human Chromosomeâ€‰5p. Genome Research, 1997, 7, 787-801.	2.4	44
44	A Radiation Hybrid Map of Human Chromosome 5 with Integration of Cytogenetic, Genetic, and Transcriptâ€‰Maps. Genome Research, 1997, 7, 897-909.	2.4	11
45	Exon trapping and sequence-based methods of gene finding in transcript mapping of human 4p 16.3. Somatic Cell and Molecular Genetics, 1997, 23, 413-427.	0.7	4
46	Structure and expression of the Huntington's disease gene: Evidence against simple inactivation due to an expanded CAG repeat. Somatic Cell and Molecular Genetics, 1994, 20, 27-38.	0.7	246
47	Isolation of genes from complex sources of mammalian genomic DNA using exon amplification. Nature Genetics, 1994, 6, 98-105.	9.4	291
48	Mutations in the transmembrane domain of FGFR3 cause the most common genetic form of dwarfism, achondroplasia. Cell, 1994, 78, 335-342.	13.5	1,218
49	Efficiency and specificity of gene isolation by exon amplification. Mammalian Genome, 1993, 4, 466-474.	1.0	18
50	A gene from chromosome 4p 16.3 with similarity to a superfamily of transporter proteins. Human Molecular Genetics, 1993, 2, 673-676.	1.4	28
51	Identification of human chromosome 9 specific genes using exon amplification. Human Molecular Genetics, 1993, 2, 1915-1920.	1.4	30
52	Molecular basis of myotonic dystrophy: Expansion of a trinucleotide (CTG) repeat at the 3â€² end of a transcript encoding a protein kinase family member. Cell, 1992, 68, 799-808.	13.5	2,464