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List of Publications by Year
in descending order

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75	16,427	87843 38	76872 74
papers	citations	h-index	g-index

89	89	89	25390
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	In Vivo Modeling of Patient Genetic Heterogeneity Identifies New Ways to Target Cholangiocarcinoma. Cancer Research, 2022, 82, 1548-1559.	0.4	8
2	Signatures of TOP1 transcription-associated mutagenesis in cancer and germline. Nature, 2022, 602, 623-631.	13.7	38
3	The contribution of evolutionarily volatile promoters to molecular phenotypes and human trait variation. Genome Biology, 2022, 23, 89.	3.8	4
4	Mutational bias in spermatogonia impacts the anatomy of regulatory sites in the human genome. Genome Research, 2021, 31, 1994-2007.	2.4	4
5	Codon Usage and Splicing Jointly Influence mRNA Localization. Cell Systems, 2020, 10, 351-362.e8.	2.9	61
6	Functional annotation of human long noncoding RNAs via molecular phenotyping. Genome Research, 2020, 30, 1060-1072.	2.4	109
7	Comparative transcriptomics of primary cells in vertebrates. Genome Research, 2020, 30, 951-961.	2.4	29
8	Evolutionary dependencies show paths to cancer development. Nature Genetics, 2020, 52, 1135-1136.	9.4	0
9	Pervasive lesion segregation shapes cancer genome evolution. Nature, 2020, 583, 265-270.	13.7	36
10	Identification of a localized nonsense-mediated decay pathway at the endoplasmic reticulum. Genes and Development, 2020, 34, 1075-1088.	2.7	37
11	Increased ultra-rare variant load in an isolated Scottish population impacts exonic and regulatory regions. PLoS Genetics, 2019, 15, e1008480.	1.5	17
12	Manipulation of Dipeptidylpeptidase 10 in mouse and human in vivo and in vitro models indicates a protective role in asthma. DMM Disease Models and Mechanisms, 2018, 11, .	1.2	11
13	Aberrant ribonucleotide incorporation and multiple deletions in mitochondrial DNA of the murine MPV17 disease model. Nucleic Acids Research, 2017, 45, 12808-12815.	6.5	43
14	Bidirectional transcription initiation marks accessible chromatin and is not specific to enhancers. Genome Biology, 2017, 18, 242.	3.8	52
15	Mutational Biases Drive Elevated Rates of Substitution at Regulatory Sites across Cancer Types. PLoS Genetics, 2016, 12, e1006207.	1.5	75
16	Homozygous loss-of-function variants in European cosmopolitan and isolate populations. Human Molecular Genetics, 2015, 24, 5464-5474.	1.4	27
17	Lagging-strand replication shapes the mutational landscape of the genome. Nature, 2015, 518, 502-506.	13.7	213
18	The frequent evolutionary birth and death of functional promoters in mouse and human. Genome Research, 2015, 25, 1546-1557.	2.4	55

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19	Exome Sequencing to Detect Rare Variants Associated With General Cognitive Ability: A Pilot Study. Twin Research and Human Genetics, 2015, 18, 117-125.	0.3	7
20	Genome-wide mapping of embedded ribonucleotides and other noncanonical nucleotides using emRiboSeq and EndoSeq. Nature Protocols, 2015, 10, 1433-1444.	5.5	42
21	Genetic heterogeneity in Cornelia de Lange syndrome (CdLS) and CdLS-like phenotypes with observed and predicted levels of mosaicism. Journal of Medical Genetics, 2014, 51, 659-668.	1.5	141
22	Functional analysis of a novel ENU-induced PHD finger 11 (Phf11) mouse mutant. Mammalian Genome, 2014, 25, 573-582.	1.0	7
23	FRA2A Is a CGG Repeat Expansion Associated with Silencing of AFF3. PLoS Genetics, 2014, 10, e1004242.	1.5	41
24	Quantification of epigenetic biomarkers: an evaluation of established and emerging methods for DNA methylation analysis. BMC Genomics, 2014, 15, 1174.	1.2	40
25	Loss of <scp>ALDH</scp>18A1 function is associated with a cellular lipid droplet phenotype suggesting a link between autosomal recessive cutis laxa type 3A and Warburg Micro syndrome. Molecular Genetics & Genomic Medicine, 2014, 2, 319-325.	0.6	19
26	SuRFing the genomics wave: an R package for prioritising SNPs by functionality. Genome Medicine, 2014, 6, 79.	3.6	15
27	A promoter-level mammalian expression atlas. Nature, 2014, 507, 462-470.	13.7	1,838
28	Variant detection sensitivity and biases in whole genome and exome sequencing. BMC Bioinformatics, 2014, 15, 247.	1.2	197
29	Heterozygous Loss-of-Function Mutations in YAP1 Cause Both Isolated and Syndromic Optic Fissure Closure Defects. American Journal of Human Genetics, 2014, 94, 295-302.	2.6	93
30	Monoallelic and Biallelic Mutations in MAB21L2 Cause a Spectrum of Major Eye Malformations. American Journal of Human Genetics, 2014, 94, 915-923.	2.6	79
31	A CGG-Repeat Expansion Mutation inZNF713Causes FRA7A: Association with Autistic Spectrum Disorder in two Families. Human Mutation, 2014, 35, n/a-n/a.	1.1	28
32	Quantifying single nucleotide variant detection sensitivity in exome sequencing. BMC Bioinformatics, 2013, 14, 195.	1.2	74
33	Application of next generation qPCR and sequencing platforms to mRNA biomarker analysis. Methods, 2013, 59, 89-100.	1.9	55
34	Evolution of the human-specific microRNA miR-941. Nature Communications, 2012, 3, 1145.	5.8	103
35	Conservation and divergence in Toll-like receptor 4-regulated gene expression in primary human versus mouse macrophages. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, E944-53.	3.3	332
36	Enzymatic Removal of Ribonucleotides from DNA Is Essential for Mammalian Genome Integrity and Development. Cell, 2012, 149, 1008-1022.	13.5	397

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37	Pervasive haplotypic variation in the spliceo-transcriptome of the human major histocompatibility complex. Genome Research, 2011, 21, 1042-1054.	2.4	63
38	CEP152 is a genome maintenance protein disrupted in Seckel syndrome. Nature Genetics, 2011, 43, 23-26.	9.4	201
39	The (non)malignancy of cancerous amino acidic substitutions. Proteins: Structure, Function and Bioinformatics, 2010, 78, 518-529.	1.5	15
40	Dynamic and Physical Clustering of Gene Expression during Epidermal Barrier Formation in Differentiating Keratinocytes. PLoS ONE, 2009, 4, e7651.	1.1	26
41	High resolution mapping of expression QTLs in heterogeneous stock mice in multiple tissues. Genome Research, 2009, 19, 1133-1140.	2.4	69
42	The Structure of Change. Science, 2009, 323, 347-348.	6.0	7
43	The transcriptional network that controls growth arrest and differentiation in a human myeloid leukemia cell line. Nature Genetics, 2009, 41, 553-562.	9.4	408
44	Comparative Genomics and Mammalian Promoter Evolution. , 2009, , 209-226.		0
45	Expression of the fras1/frem gene family during zebrafish development and fin morphogenesis. Developmental Dynamics, 2008, 237, 3295-3304.	0.8	20
46	Rapidly evolving human promoter regions. Nature Genetics, 2008, 40, 1262-1263.	9.4	18
47	Singleton SNPs in the human genome and implications for genome-wide association studies. European Journal of Human Genetics, 2008, 16, 506-515.	1.4	39
48	Development and evaluation of a real-time PCR assay for detection of Pneumocystis jirovecii DNA in bronchoalveolar lavage fluid of HIV-infected patients. Thorax, 2007, 63, 154-159.	2.7	110
49	Identification of Common Genetic Variation That Modulates Alternative Splicing. PLoS Genetics, 2007, 3, e99.	1.5	139
50	Management, presentation and interpretation of genome scans using GSCANDB. Bioinformatics, 2007, 23, 1545-1549.	1.8	4
51	Evidence of a Large-Scale Functional Organization of Mammalian Chromosomes: Authors' Reply. PLoS Biology, 2007, 5, e128.	2.6	1
52	Problems of Developing Molecular Diagnostic Tests for Opportunistic Pathogens: The Example of Pneumocystis jirovecii. Journal of Eukaryotic Microbiology, 2006, 53, S85-S86.	0.8	4
53	Genome-wide analysis of mammalian promoter architecture and evolution. Nature Genetics, 2006, 38, 626-635.	9.4	1,201
54	Genome-wide genetic association of complex traits in heterogeneous stock mice. Nature Genetics, 2006, 38, 879-887.	9.4	508

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55	A High-Resolution Single Nucleotide Polymorphism Genetic Map of the Mouse Genome. PLoS Biology, 2006, 4, e395.	2.6	243
56	Heterotachy in Mammalian Promoter Evolution. PLoS Genetics, 2006, 2, e30.	1.5	102
57	Genomic anatomy of the Tyrp1 (brown) deletion complex. Proceedings of the National Academy of Sciences of the United States of America, 2006, 103, 3704-3709.	3.3	30
58	Identification of a new gene mutated in Fraser syndrome and mouse myelencephalic blebs. Nature Genetics, 2005, 37, 520-525.	9.4	148
59	The Transcriptional Landscape of the Mammalian Genome. Science, 2005, 309, 1559-1563.	6.0	3,227
60	Occurrence and Consequences of Coding Sequence Insertions and Deletions in Mammalian Genomes. Genome Research, 2004, 14, 555-566.	2.4	114
61	The extracellular matrix gene Frem1 is essential for the normal adhesion of the embryonic epidermis. Proceedings of the National Academy of Sciences of the United States of America, 2004, 101, 13560-13565.	3.3	108
62	Genetic Analysis of Pathways Regulated by the von Hippel-Lindau Tumor Suppressor in Caenorhabditis elegans. PLoS Biology, 2004, 2, e289.	2.6	137
63	Genome sequence of the Brown Norway rat yields insights into mammalian evolution. Nature, 2004, 428, 493-521.	13.7	1,943
64	Sequence Characterization of Teleost Fish Melanocortin Receptors. Annals of the New York Academy of Sciences, 2003, 994, 319-330.	1.8	30
65	Evolutionary constraints on the Disrupted in Schizophrenia locus. Genomics, 2003, 81, 67-77.	1.3	83
66	The structure and evolution of the melanocortin and MCH receptors in fish and mammals. Genomics, 2003, 81, 184-191.	1.3	139
67	Interaction of the Anaphase-promoting Complex/Cyclosome and Proteasome Protein Complexes with Multiubiquitin Chain-binding Proteins. Journal of Biological Chemistry, 2003, 278, 16791-16796.	1.6	60
68	The severe G480C cystic fibrosis mutation, when replicated in the mouse, demonstrates mistrafficking, normal survival and organ-specific bioelectrics. Human Molecular Genetics, 2002, 11, 243-251.	1.4	27
69	Sushi gets serious: the draft genome sequence of the pufferfish Fugu rubripes. Genome Biology, 2002, 3, reviews1025.1.	13.9	1
70	Analysis of the mouse transcriptome based on functional annotation of 60,770 full-length cDNAs. Nature, 2002, 420, 563-573.	13.7	1,548
71	Characterization and comparative analysis of the EGLN gene family. Gene, 2001, 275, 125-132.	1.0	130
72	Isolation and characterization of the mouse translin-associated protein X (Trax) gene. Mammalian Genome, 2000, 11, 395-398.	1.0	12

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73	Genomic Sequence Analysis of Fugu rubripes CFTR and Flanking Genes in a 60 kb Region Conserving Synteny with 800 kb of Human Chromosome 7. Genome Research, 2000, 10, 1194-1203.	2.4	26
74	Disruption of two novel genes by a translocation co-segregating with schizophrenia. Human Molecular Genetics, 2000, 9, 1415-1423.	1.4	1,135
75	Comparative Genomics. , 0, , 105-144.		0