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List of Publications by Year
in descending order

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54 papers	13,403 citations	172207 29 h-index	174990 52 g-index
74 all docs	74 docs citations	74 times ranked	27048 citing authors

#	Article	IF	Citations
1	GREAT improves functional interpretation of cis-regulatory regions. Nature Biotechnology, 2010, 28, 495-501.	9.4	3,789
2	Evolutionarily conserved elements in vertebrate, insect, worm, and yeast genomes. Genome Research, 2005, 15, 1034-1050.	2.4	3,517
3	Ultraconserved Elements in the Human Genome. Science, 2004, 304, 1321-1325.	6.0	1,496
4	M-CAP eliminates a majority of variants of uncertain significance in clinical exomes at high sensitivity. Nature Genetics, 2016, 48, 1581-1586.	9.4	654
5	Enhancers: five essential questions. Nature Reviews Genetics, 2013, 14, 288-295.	7.7	455
6	A distal enhancer and an ultraconserved exon are derived from a novel retroposon. Nature, 2006, 441, 87-90.	13.7	452
7	Human-specific loss of regulatory DNA and the evolution of human-specific traits. Nature, 2011, 471, 216-219.	13.7	439
8	Systematic reanalysis of clinical exome data yields additional diagnoses: implications for providers. Genetics in Medicine, 2017, 19, 209-214.	1.1	261
9	Identification of rare-disease genes using blood transcriptome sequencing and large control cohorts. Nature Medicine, 2019, 25, 911-919.	15.2	221
10	Coding exons function as tissue-specific enhancers of nearby genes. Genome Research, 2012, 22, 1059-1068.	2.4	202
11	A â€œForward Genomicsâ€•Approach Links Genotype to Phenotype using Independent Phenotypic Losses among Related Species. Cell Reports, 2012, 2, 817-823.	2.9	133
12	Microbiota modulate transcription in the intestinal epithelium without remodeling the accessible chromatin landscape. Genome Research, 2014, 24, 1504-1516.	2.4	119
13	Deriving genomic diagnoses without revealing patient genomes. Science, 2017, 357, 692-695.	6.0	110
14	Mutations of AKT3 are associated with a wide spectrum of developmental disorders including extreme megalencephaly. Brain, 2017, 140, 2610-2622.	3.7	102
15	MicroRNA-9 Couples Brain Neurogenesis and Angiogenesis. Cell Reports, 2017, 20, 1533-1542.	2.9	90
16	A family of transposable elements co-opted into developmental enhancers in the mouse neocortex. Nature Communications, 2015, 6, 6644.	5.8	88
17	TBR1 regulates autism risk genes in the developing neocortex. Genome Research, 2016, 26, 1013-1022.	2.4	71
18	ClinPhen extracts and prioritizes patient phenotypes directly from medical records to expedite genetic disease diagnosis. Genetics in Medicine, 2019, 21, 1585-1593.	1.1	67

#	Article	IF	Citations
19	Darwin. , 2018, , .		66
20	AMELIE speeds Mendelian diagnosis by matching patient phenotype and genotype to primary literature. Science Translational Medicine, 2020, 12, .	5.8	60
21	A sequence-based, deep learning model accurately predicts RNA splicing branchpoints. Rna, 2018, 24, 1647-1658.	1.6	59
22	S-CAP extends pathogenicity prediction to genetic variants that affect RNA splicing. Nature Genetics, 2019, 51, 755-763.	9.4	56
23	â€œReverse Genomicsâ€•Predicts Function of Human Conserved Noncoding Elements. Molecular Biology and Evolution, 2016, 33, 1358-1369.	3.5	55
24	Human Developmental Enhancers Conserved between Deuterostomes and Protostomes. PLoS Genetics, 2012, 8, e1002852.	1.5	55
25	Computational screening of conserved genomic DNA in search of functional noncoding elements. Nature Methods, 2005, 2, 535-545.	9.0	52
26	Mx1 and Mx2 key antiviral proteins are surprisingly lost in toothed whales. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, 8036-8040.	3.3	50
27	Hundreds of conserved non-coding genomic regions are independently lost in mammals. Nucleic Acids Research, 2012, 40, 11463-11476.	6.5	48
28	Components of genetic associations across 2,138 phenotypes in the UK Biobank highlight adipocyte biology. Nature Communications, 2019, 10, 4064.	5.8	48
29	Dispensability of mammalian DNA. Genome Research, 2008, 18, 1743-1751.	2.4	42
30	Characterization of TCF21 Downstream Target Regions Identifies a Transcriptional Network Linking Multiple Independent Coronary Artery Disease Loci. PLoS Genetics, 2015, 11, e1005202.	1.5	41
31	Darwin. ACM SIGPLAN Notices, 2018, 53, 199-213.	0.2	41
32	Changes in the enhancer landscape during early placental development uncover a trophoblast invasion gene-enhancer network. Placenta, 2016, 37, 45-55.	0.7	35
33	A functional enrichment test for molecular convergent evolution finds a clear protein-coding signal in echolocating bats and whales. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 21094-21103.	3.3	34
34	The Enhancer Landscape during Early Neocortical Development Reveals Patterns of Dense Regulation and Co-option. PLoS Genetics, 2013, 9, e1003728.	1.5	33
35	Phrank measures phenotype sets similarity to greatly improve Mendelian diagnostic disease prioritization. Genetics in Medicine, 2019, 21, 464-470.	1.1	33
36	PRISM offers a comprehensive genomic approach to transcription factor function prediction. Genome Research, 2013, 23, 889-904.	2.4	32

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37	Structure-aided prediction of mammalian transcription factor complexes in conserved non-coding elements. Philosophical Transactions of the Royal Society B: Biological Sciences, 2013, 368, 20130029.	1.8	30
38	CRISPR/Cas9 Genome Engineering in Engraftable Human Brain-Derived Neural Stem Cells. IScience, 2019, 15, 524-535.	1.9	27
39	AVADA: toward automated pathogenic variant evidence retrieval directly from the full-text literature. Genetics in Medicine, 2020, 22, 362-370.	1.1	24
40	Transcription factor expression defines subclasses of developing projection neurons highly similar to single-cell RNA-seq subtypes. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 25074-25084.	3.3	23
41	Darwin-WGA: A Co-processor Provides Increased Sensitivity in Whole Genome Alignments with High Speedup. , 2019, , .		17
42	A Penile Spine/Vibrissa Enhancer Sequence Is Missing in Modern and Extinct Humans but Is Retained in Multiple Primates with Penile Spines and Sensory Vibrissae. PLoS ONE, 2013, 8, e84258.	1.1	16
43	Biallelic lossâ€ofâ€function WNT5A mutations in an infant with severe and atypical manifestations of Robinow syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 1030-1036.	0.7	15
44	Independent erosion of conserved transcription factor binding sites points to shared hindlimb, vision and external testes loss in different mammals. Nucleic Acids Research, 2018, 46, 9299-9308.	6.5	15
45	An MTF1 binding site disrupted by a homozygous variant in the promoter of ATP7B likely causes Wilson Disease. European Journal of Human Genetics, 2018, 26, 1810-1818.	1.4	15
46	Automated Discovery of Tissue-Targeting Enhancers and Transcription Factors from Binding Motif and Gene Function Data. PLoS Computational Biology, 2014, 10, e1003449.	1.5	11
47	Detection of a mosaic CDKL5 deletion and inversion by optical genome mapping ends an exhaustive diagnostic odyssey. Molecular Genetics & Genomic Medicine, 2021, 9, e1665.	0.6	11
48	A fully-automated method discovers loss of mouse-lethal and human-monogenic disease genes in 58 mammals. Nucleic Acids Research, 2020, 48, e91-e91.	6.5	7
49	The Effect of Population Structure on Murine Genome-Wide Association Studies. Frontiers in Genetics, 2021, 12, 745361.	1.1	7
50	Erosion of Conserved Binding Sites in Personal Genomes Points to Medical Histories. PLoS Computational Biology, 2016, 12, e1004711.	1.5	7
51	Morphogenesis is transcriptionally coupled to neurogenesis during peripheral olfactory organ development. Development (Cambridge), 2020, 147, .	1.2	6
52	Champagne: Automated Whole-Genome Phylogenomic Character Matrix Method Using Large Genomic Indels for Homoplasy-Free Inference. Genome Biology and Evolution, 2022, 14, .	1.1	3
53	Avoiding genetic racial profiling in criminal DNA profile databases. Nature Computational Science, 2021, 1, 272-279.	3.8	1
54	InpherNet accelerates monogenic disease diagnosis using patientsâ€™ candidate genesâ€™ neighbors. Genetics in Medicine, 2021, 23, 1984-1992.	1.1	1