Gill Bejerano

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	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
2	Avoiding genetic racial profiling in criminal DNA profile databases. Nature Computational Science, 2021, 1, 272-279.	3.8	1
3	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
4	InpherNet accelerates monogenic disease diagnosis using patientsâ€™ candidate genesâ€™ neighbors. Genetics in Medicine, 2021, 23, 1984-1992.	1.1	1
5	The Effect of Population Structure on Murine Genome-Wide Association Studies. Frontiers in Genetics, 2021, 12, 745361.	1.1	7
6	AVADA: toward automated pathogenic variant evidence retrieval directly from the full-text literature. Genetics in Medicine, 2020, 22, 362-370.	1.1	24
7	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
8	Morphogenesis is transcriptionally coupled to neurogenesis during peripheral olfactory organ development. Development (Cambridge), 2020, 147, .	1.2	6
9	AMELIE speeds Mendelian diagnosis by matching patient phenotype and genotype to primary literature. Science Translational Medicine, 2020, 12, .	5.8	60
10	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
11	Phrank measures phenotype sets similarity to greatly improve Mendelian diagnostic disease prioritization. Genetics in Medicine, 2019, 21, 464-470.	1.1	33
12	Components of genetic associations across 2,138 phenotypes in the UK Biobank highlight adipocyte biology. Nature Communications, 2019, 10, 4064.	5.8	48
13	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
14	CRISPR/Cas9 Genome Engineering in Engraftable Human Brain-Derived Neural Stem Cells. IScience, 2019, 15, 524-535.	1.9	27
15	Darwin-WGA: A Co-processor Provides Increased Sensitivity in Whole Genome Alignments with High Speedup. , 2019, , .		17
16	Identification of rare-disease genes using blood transcriptome sequencing and large control cohorts. Nature Medicine, 2019, 25, 911-919.	15.2	221
17	S-CAP extends pathogenicity prediction to genetic variants that affect RNA splicing. Nature Genetics, 2019, 51, 755-763.	9.4	56
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

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19	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
20	Darwin. , 2018, , .		66
21	A sequence-based, deep learning model accurately predicts RNA splicing branchpoints. Rna, 2018, 24, 1647-1658.	1.6	59
22	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
23	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
24	Darwin. ACM SIGPLAN Notices, 2018, 53, 199-213.	0.2	41
25	Mutations of AKT3 are associated with a wide spectrum of developmental disorders including extreme megalencephaly. Brain, 2017, 140, 2610-2622.	3.7	102
26	MicroRNA-9 Couples Brain Neurogenesis and Angiogenesis. Cell Reports, 2017, 20, 1533-1542.	2.9	90
27	Deriving genomic diagnoses without revealing patient genomes. Science, 2017, 357, 692-695.	6.0	110
28	Systematic reanalysis of clinical exome data yields additional diagnoses: implications for providers. Genetics in Medicine, 2017, 19, 209-214.	1.1	261
29	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
30	TBR1 regulates autism risk genes in the developing neocortex. Genome Research, 2016, 26, 1013-1022.	2.4	71
31	â€œReverse Genomicsâ€•Predicts Function of Human Conserved Noncoding Elements. Molecular Biology and Evolution, 2016, 33, 1358-1369.	3.5	55
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	Erosion of Conserved Binding Sites in Personal Genomes Points to Medical Histories. PLoS Computational Biology, 2016, 12, e1004711.	1.5	7
34	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
35	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
36	A family of transposable elements co-opted into developmental enhancers in the mouse neocortex. Nature Communications, 2015, 6, 6644.	5.8	88

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37	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
38	Microbiota modulate transcription in the intestinal epithelium without remodeling the accessible chromatin landscape. Genome Research, 2014, 24, 1504-1516.	2.4	119
39	Enhancers: five essential questions. Nature Reviews Genetics, 2013, 14, 288-295.	7.7	455
40	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
41	PRISM offers a comprehensive genomic approach to transcription factor function prediction. Genome Research, 2013, 23, 889-904.	2.4	32
42	The Enhancer Landscape during Early Neocortical Development Reveals Patterns of Dense Regulation and Co-option. PLoS Genetics, 2013, 9, e1003728.	1.5	33
43	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
44	Hundreds of conserved non-coding genomic regions are independently lost in mammals. Nucleic Acids Research, 2012, 40, 11463-11476.	6.5	48
45	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
46	Coding exons function as tissue-specific enhancers of nearby genes. Genome Research, 2012, 22, 1059-1068.	2.4	202
47	Human Developmental Enhancers Conserved between Deuterostomes and Protostomes. PLoS Genetics, 2012, 8, e1002852.	1.5	55
48	Human-specific loss of regulatory DNA and the evolution of human-specific traits. Nature, 2011, 471, 216-219.	13.7	439
49	GREAT improves functional interpretation of cis-regulatory regions. Nature Biotechnology, 2010, 28, 495-501.	9.4	3,789
50	Dispensability of mammalian DNA. Genome Research, 2008, 18, 1743-1751.	2.4	42
51	A distal enhancer and an ultraconserved exon are derived from a novel retroposon. Nature, 2006, 441, 87-90.	13.7	452
52	Computational screening of conserved genomic DNA in search of functional noncoding elements. Nature Methods, 2005, 2, 535-545.	9.0	52
53	Evolutionarily conserved elements in vertebrate, insect, worm, and yeast genomes. Genome Research, 2005, 15, 1034-1050.	2.4	3,517
54	Ultraconserved Elements in the Human Genome. Science, 2004, 304, 1321-1325.	6.0	1,496