Miao-Xin Li

List of Publications by Year
in descending order

77 papers	3,868 citations	218677 26 h-index	149698 56 g-index
82 all docs	82 docs citations	82 times ranked	8076 citing authors

#	Article	IF	Citations
1	Mapping genomic loci implicates genes and synaptic biology in schizophrenia. Nature, 2022, 604, 502-508.	27.8	929
2	Evaluating the effective numbers of independent tests and significant p-value thresholds in commercial genotyping arrays and public imputation reference datasets. Human Genetics, 2012, 131, 747-756.	3.8	658
3	GATES: A Rapid and Powerful Gene-Based Association Test Using Extended Simes Procedure. American Journal of Human Genetics, 2011, 88, 283-293.	6.2	350
4	A comprehensive framework for prioritizing variants in exome sequencing studies of Mendelian diseases. Nucleic Acids Research, 2012, 40, e53-e53.	14.5	229
5	Predicting Mendelian Disease-Causing Non-Synonymous Single Nucleotide Variants in Exome Sequencing Studies. PLoS Genetics, 2013, 9, e1003143.	3.5	127
6	Heritability of variation in glycaemic response to metformin: a genome-wide complex trait analysis. Lancet Diabetes and Endocrinology,the, 2014, 2, 481-487.	11.4	101
7	HYST: A Hybrid Set-Based Test for Genome-wide Association Studies, with Application to Protein-Protein Interaction-Based Association Analysis. American Journal of Human Genetics, 2012, 91, 478-488.	6.2	96
8	The p.Ser267Phe variant in SLC10A1 is associated with resistance to chronic hepatitis B. Hepatology, 2015, 61, 1251-1260.	7.3	78
9	Uncovering the total heritability explained by all true susceptibility variants in a genome-wide association study. Genetic Epidemiology, 2011, 35, n/a-n/a.	1.3	67
10	Dysfunction of Myosin Lightâ€Chain 4 (MYL4) Leads to Heritable Atrial Cardiomyopathy With Electrical, Contractile, and Structural Components: Evidence From Geneticallyâ€Engineered Rats. Journal of the American Heart Association, 2017, 6, .	3.7	52
11	Comparisons of seven algorithms for pathway analysis using the WTCCC Crohn's Disease dataset. BMC Research Notes, 2011, 4, 386.	1.4	49
12	Common Variants on Xq28 Conferring Risk of Schizophrenia in Han Chinese. Schizophrenia Bulletin, 2014, 40, 777-786.	4.3	49
13	A Knowledge-Based Weighting Framework to Boost the Power of Genome-Wide Association Studies. PLoS ONE, 2010, 5, e14480.	2.5	48
14	Genetic determination and correlation of body mass index and bone mineral density at the spine and hip in Chinese Han ethnicity. Osteoporosis International, 2006, 17, 119-124.	3.1	44
15	Burden of rare variants in ALS genes influences survival in familial and sporadic ALS. Neurobiology of Aging, 2017, 58, 238.e9-238.e15.	3.1	42
16	Increased co-expression of genes harboring the damaging de novo mutations in Chinese schizophrenic patients during prenatal development. Scientific Reports, 2015, 5, 18209.	3.3	40
17	Predicting regulatory variants with composite statistic. Bioinformatics, 2016, 32, 2729-2736.	4.1	40
18	MGAS: a powerful tool for multivariate gene-based genome-wide association analysis. Bioinformatics, 2015, 31, 1007-1015.	4.1	39

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19	Estrogen Receptor Î± Gene Polymorphisms and Peak Bone Density in Chinese Nuclear Families. Journal of Bone and Mineral Research, 2003, 18, 1028-1035.	2.8	36
20	Robust and rapid algorithms facilitate large-scale whole genome sequencing downstream analysis in an integrative framework. Nucleic Acids Research, 2017, 45, gkx019.	14.5	36
21	Bone mineral density in elderly Chinese: effects of age, sex, weight, height, and body mass index. Journal of Bone and Mineral Metabolism, 2004, 22, 71-78.	2.7	34
22	SNPP: automating large-scale SNP genotype data management. Bioinformatics, 2005, 21, 266-268.	4.1	34
23	cepip: context-dependent epigenomic weighting for prioritization of regulatory variants and disease-associated genes. Genome Biology, 2017, 18, 52.	8.8	33
24	An NT-3-releasing bioscaffold supports the formation of TrkC-modified neural stem cell-derived neural network tissue with efficacy in repairing spinal cord injury. Bioactive Materials, 2021, 6, 3766-3781.	15.6	31
25	Rare inborn errors associated with chronic hepatitis B virus infection*. Hepatology, 2012, 56, 1661-1670.	7.3	30
26	<scp>PMCA</scp>4 (<scp>ATP</scp>2B4) mutation in familial spastic paraplegia causes delay in intracellular calcium extrusion. Brain and Behavior, 2015, 5, e00321.	2.2	30
27	The role of gene variants in the pathogenesis of neurodegenerative disorders as revealed by next generation sequencing studies: a review. Translational Neurodegeneration, 2017, 6, 27.	8.0	29
28	PMCA4 (ATP2B4) Mutation in Familial Spastic Paraplegia. PLoS ONE, 2014, 9, e104790.	2.5	28
29	WITER: a powerful method for estimation of cancer-driver genes using a weighted iterative regression modelling background mutation counts. Nucleic Acids Research, 2019, 47, e96-e96.	14.5	28
30	Computational resources associating diseases with genotypes, phenotypes and exposures. Briefings in Bioinformatics, 2019, 20, 2098-2115.	6.5	27
31	A major gene model of adult height is suggested in Chinese. Journal of Human Genetics, 2004, 49, 148-153.	2.3	23
32	A rare variant in MLKL confers susceptibility to ApoE É›4-negative Alzheimer's disease in Hong Kong Chinese population. Neurobiology of Aging, 2018, 68, 160.e1-160.e7.	3.1	23
33	The â^'1997 G/T Polymorphism in the COLIA1 Upstream Regulatory Region is Associated with Hip Bone Mineral Density (BMD) in Chinese Nuclear Families. Calcified Tissue International, 2005, 76, 107-112.	3.1	21
34	Genome scan for QTLs underlying bone size variation at 10 refined skeletal sites: genetic heterogeneity and the significance of phenotype refinement. Physiological Genomics, 2004, 17, 326-331.	2.3	20
35	Dynamic landscape of alternative polyadenylation during retinal development. Cellular and Molecular Life Sciences, 2017, 74, 1721-1739.	5.4	20
36	A powerful conditional gene-based association approach implicated functionally important genes for schizophrenia. Bioinformatics, 2019, 35, 628-635.	4.1	19

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37	Hippocampal transcriptome-wide association study and neurobiological pathway analysis for Alzheimerâ€™s disease. PLoS Genetics, 2021, 17, e1009363.	3.5	18
38	Identification of Genes with Allelic Imbalance on 6p Associated with Nasopharyngeal Carcinoma in Southern Chinese. PLoS ONE, 2011, 6, e14562.	2.5	17
39	Genetic determination of variation and covariation of bone mineral density at the hip and spine in a Chinese population. Journal of Bone and Mineral Metabolism, 2005, 23, 181-185.	2.7	16
40	No major effect of the insulin-like growth factor I gene on bone mineral density in premenopausal Chinese women. Bone, 2005, 36, 694-699.	2.9	16
41	â^'459C>T point mutation in 5â€² nonâ€coding region of human GJB1 gene is linked to Xâ€linked Charcotâ€Marieâ€Tooth neuropathy. Journal of the Peripheral Nervous System, 2009, 14, 14-21.	3.1	16
42	DESE: estimating driver tissues by selective expression of genes associated with complex diseases or traits. Genome Biology, 2019, 20, 233.	8.8	15
43	Interaction effects between estrogen receptor alpha and vitamin D receptor genes on age at menarche in Chinese women. Acta Pharmacologica Sinica, 2005, 26, 860-864.	6.1	14
44	Ultrafast and scalable variant annotation and prioritization with big functional genomics data. Genome Research, 2020, 30, 1789-1801.	5.5	14
45	Lack of association between the Hind III RFLP of the osteocalcin (BGP) gene and bone mineral density (BMD) in healthy pre- and postmenopausal Chinese women. Journal of Bone and Mineral Metabolism, 2004, 22, 264-269.	2.7	13
46	Allele and genotype frequencies of polymorphicFMO3 gene in two genetically distinct populations. Cell Biochemistry and Function, 2007, 25, 443-453.	2.9	13
47	SNPTracker: A Swift Tool for Comprehensive Tracking and Unifying dbSNP rs IDs and Genomic Coordinates of Massive Sequence Variants. G3: Genes, Genomes, Genetics, 2016, 6, 205-207.	1.8	13
48	Cancer gene mutations in congenital pulmonary airway malformation patients. ERJ Open Research, 2019, 5, 00196-2018.	2.6	12
49	Inheritance-mode specific pathogenicity prioritization (ISPP) for human protein coding genes. Bioinformatics, 2016, 32, 3065-3071.	4.1	11
50	FAPI: Fast and accurate P-value Imputation for genome-wide association study. European Journal of Human Genetics, 2016, 24, 761-766.	2.8	11
51	IGG3: a tool to rapidly integrate large genotype datasets for whole-genome imputation and individual-level meta-analysis. Bioinformatics, 2009, 25, 1449-1450.	4.1	10
52	wKGGSeq: A Comprehensive Strategy-Based and Disease-Targeted Online Framework to Facilitate Exome Sequencing Studies of Inherited Disorders. Human Mutation, 2015, 36, 496-503.	2.5	10
53	De novo mutations in Caudal Type Homeo Box transcription Factor 2 (CDX2) in patients with persistent cloaca. Human Molecular Genetics, 2018, 27, 351-358.	2.9	9
54	A powerful approach reveals numerous expression quantitative trait haplotypes in multiple tissues. Bioinformatics, 2018, 34, 3145-3150.	4.1	9

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55	Powerful and robust inference of complex phenotypes' causal genes with dependent expression quantitative loci by a median-based Mendelian randomization. American Journal of Human Genetics, 2022, 109, 838-856.	6.2	8
56	Parathyroid hormone gene with bone phenotypes in Chinese. Biochemical and Biophysical Research Communications, 2003, 307, 666-671.	2.1	7
57	Lack of Evidence for a Major Gene in the Mendelian Transmission of BMI in Chinese. Obesity, 2004, 12, 1967-1973.	4.0	7
58	Association and linkage analyses of interleukin-6 gene 634C/G polymorphism and bone phenotypes in Chinese. Journal of Bone and Mineral Metabolism, 2005, 23, 323-328.	2.7	7
59	Sharing of Genes and Pathways Across Complex Phenotypes: A Multilevel Genome-Wide Analysis. Genetics, 2017, 206, 1601-1609.	2.9	7
60	Knowledge-based analyses reveal new candidate genes associated with risk of hepatitis B virus related hepatocellular carcinoma. BMC Cancer, 2020, 20, 403.	2.6	6
61	Robust Indices of Hardy-Weinberg Disequilibrium for QTL Fine Mapping. Human Heredity, 2003, 56, 160-165.	0.8	5
62	Genetic overlap between epilepsy and schizophrenia: Evidence from cross phenotype analysis in Hong Kong Chinese population. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2018, 177, 86-92.	1.7	5
63	A conditional gene-based association framework integrating isoform-level eQTL data reveals new susceptibility genes for schizophrenia. ELife, 2022, 11, .	6.0	5
64	The genetic, environmental and phenotypic correlations of bone phenotypes at the spine and hip in Chinese. Annals of Human Biology, 2006, 33, 500-509.	1.0	4
65	Multivariate Gene-Based Association Test on Family Data in MGAS. Behavior Genetics, 2016, 46, 718-725.	2.1	4
66	PCGA: a comprehensive web server for phenotype-cell-gene association analysis. Nucleic Acids Research, 2022, 50, W568-W576.	14.5	4
67	An accurate prediction model of digenic interaction for estimating pathogenic gene pairs of human diseases. Computational and Structural Biotechnology Journal, 2022, 20, 3639-3652.	4.1	4
68	Linkage exclusion analysis of two important chromosomal regions for height. Biochemical and Biophysical Research Communications, 2005, 335, 1287-1292.	2.1	3
69	Exclusion mapping of chromosomes 1, 4, 6 and 14 with bone mineral density in 79 Caucasian pedigrees. Bone, 2006, 38, 450-455.	2.9	3
70	SPS: A Simulation Tool for Calculating Power of Setâ€Based Genetic Association Tests. Genetic Epidemiology, 2015, 39, 395-397.	1.3	3
71	Deviation from baseline mutation burden provides powerful and robust rare-variants association test for complex diseases. Nucleic Acids Research, 2022, 50, e34-e34.	14.5	3
72	DEPD: a novel database for differentially expressed proteins. Bioinformatics, 2005, 21, 3694-3696.	4.1	2

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73	De novo mutations as causes of schizophrenia. Psychiatry Research, 2018, 270, 1168-1169.	3.3	1
74	Case report: exome sequencing achieved a definite diagnosis in a Chinese family with muscle atrophy. BMC Neurology, 2021, 21, 96.	1.8	1
75	RNA-SSNV: A Reliable Somatic Single Nucleotide Variant Identification Framework for Bulk RNA-Seq Data. Frontiers in Genetics, 0, 13, .	2.3	1
76	Power of transmission/disequilibrium tests in admixed populations. Genetic Epidemiology, 2008, 32, 434-444.	1.3	0
77	Genome-Wide Association Study Identifies New Risk Loci for Progression of Schistosomiasis Among the Chinese Population. Frontiers in Cellular and Infection Microbiology, 2022, 12, 871545.	3.9	0