Zhiyu Peng

List of Publications by Year
in descending order

46 papers	3,719 citations	331670 21 h-index	233421 45 g-index
59 all docs	59 docs citations	59 times ranked	6892 citing authors

#	Article	IF	Citations
1	Next generation sequencing is a highly reliable method to analyze exon 7 deletion of survival motor neuron 1 (SMN1) gene. Scientific Reports, 2022, 12, 223.	3.3	9
2	Utility of Whole Genome Sequencing for Population Screening of Deafness-Related Genetic Variants and Cytomegalovirus Infection in Newborns. Frontiers in Genetics, 2022, 13, 883617.	2.3	1
3	The effect of hemolysis on quality control metrics for noninvasive prenatal testing. BMC Medical Genomics, 2022, 15, .	1.5	1
4	Comprehensive genetic testing improves the clinical diagnosis and medical management of pediatric patients with isolated hearing loss. BMC Medical Genomics, 2022, 15, .	1.5	4
5	NGS-based spinal muscular atrophy carrier screening of 10,585 diverse couples in China: a pan-ethnic study. European Journal of Human Genetics, 2021, 29, 194-204.	2.8	17
6	Identification of Two Novel Thalassemia Variants, HBA1: c.263delA and HBA2: c.376dupC, in Chinese Individuals. Hemoglobin, 2021, 45, 49-51.	0.8	0
7	Report of Two Novel Thalassemia Variants, HBB: c.181delG and HBA1: c.121_126delAAGACC, in Chinese Individuals. Hemoglobin, 2021, 45, 52-55.	0.8	0
8	A multiplex PCR amplicon sequencing assay to screen genetic hearing loss variants in newborns. BMC Medical Genomics, 2021, 14, 61.	1.5	5
9	Noninvasive prenatal testing of Î±-thalassemia and Î²-thalassemia through population-based parental haplotyping. Genome Medicine, 2021, 13, 18.	8.2	16
10	Molecular diagnosis of non-syndromic hearing loss patients using a stepwise approach. Scientific Reports, 2021, 11, 4036.	3.3	7
11	Characterizing sensitivity and coverage of clinical WGS as a diagnostic test for genetic disorders. BMC Medical Genomics, 2021, 14, 102.	1.5	16
12	VIPâ€HL: Semiâ€automated ACMG/AMP variant interpretation platform for genetic hearing loss. Human Mutation, 2021, 42, 1567-1575.	2.5	10
13	AutoCNV: a semiautomatic CNV interpretation system based on the 2019 ACMG/ClinGen Technical Standards for CNVs. BMC Genomics, 2021, 22, 721.	2.8	6
14	Noninvasive prenatal diagnosis of monogenic disorders based on direct haplotype phasing through targeted linked-read sequencing. BMC Medical Genomics, 2021, 14, 244.	1.5	6
15	Applications of Noninvasive Prenatal Testing for Subchromosomal Copy Number Variations Using Cell-Free DNA. Advances in Molecular Pathology, 2021, 4, 17-25.	0.4	1
16	Five novel globin gene mutations identified in five Chinese families by nextâ€generation sequencing. Molecular Genetics & Genomic Medicine, 2021, , e1835.	1.2	2
17	Impact of ultrasonography on identifying noninvasive prenatal screening falseâ€negative aneuploidy. Molecular Genetics & Genomic Medicine, 2020, 8, e1213.	1.2	1
18	Reinterpretation of common pathogenic variants in ClinVar revealed a high proportion of downgrades. Scientific Reports, 2020, 10, 331.	3.3	34

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19	AutoPVS1: An automatic classification tool for PVS1 interpretation of null variants. Human Mutation, 2020, 41, 1488-1498.	2.5	34
20	Concurrent hearing and genetic screening in a general newborn population. Human Genetics, 2020, 139, 521-530.	3.8	24
21	Hemoglobin, 2019, 43, 7-11.	0.8	11
22	Increased diagnostic yield by reanalysis of data from a hearing loss gene panel. BMC Medical Genomics, 2019, 12, 76.	1.5	16
23	A comprehensive assessment of Nextâ€Generation Sequencing variants validation using a secondary technology. Molecular Genetics & Genomic Medicine, 2019, 7, e00748.	1.2	17
24	Nationwide population genetic screening improves outcomes of newborn screening for hearing loss in China. Genetics in Medicine, 2019, 21, 2231-2238.	2.4	40
25	Pilot study of expanded carrier screening for 11 recessive diseases in China: results from 10,476 ethnically diverse couples. European Journal of Human Genetics, 2019, 27, 254-262.	2.8	33
26	Identification of a novel breast cancerâ€‘causing mutation in the BRCA1 gene by targeted next generation sequencing: A case report. Oncology Letters, 2018, 16, 3913-3916.	1.8	1
27	The mutational landscape of MYCN, Lin28b and ALKF1174L driven murine neuroblastoma mimics human disease. Oncotarget, 2018, 9, 8334-8349.	1.8	6
28	Next-generation sequencing improves thalassemia carrier screening among premarital adults in a high prevalence population: the Dai nationality, China. Genetics in Medicine, 2017, 19, 1022-1031.	2.4	82
29	Î²-thalassemia caused by compound heterozygous mutations and cured by bone marrow transplantation: A case report. Molecular Medicine Reports, 2017, 16, 6552-6557.	2.4	1
30	Rapid Targeted Next-Generation Sequencing Platform for Molecular Screening and Clinical Genotyping in Subjects with Hemoglobinopathies. EBioMedicine, 2017, 23, 150-159.	6.1	138
31	Clinical and genomic evaluation of a Chinese patient with a novel deletion associated with Phelan-McDermid syndrome. Oncotarget, 2016, 7, 80327-80335.	1.8	3
32	Hypothesis: Artifacts, Including Spurious Chimeric RNAs with a Short Homologous Sequence, Caused by Consecutive Reverse Transcriptions and Endogenous Random Primers. Journal of Cancer, 2015, 6, 555-567.	2.5	32
33	Comparison of RNA-seq and microarray-based models for clinical endpoint prediction. Genome Biology, 2015, 16, 133.	8.8	325
34	A systems genetics study of swine illustrates mechanisms underlying human phenotypic traits. BMC Genomics, 2015, 16, 88.	2.8	28
35	Telomerase activation by genomic rearrangements in high-risk neuroblastoma. Nature, 2015, 526, 700-704.	27.8	478
36	Genome-wide identification of RNA editing in hepatocellular carcinoma. Genomics, 2015, 105, 76-82.	2.9	40

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37	RNA Editome in Rhesus Macaque Shaped by Purifying Selection. PLoS Genetics, 2014, 10, e1004274.	3.5	71
38	Caste-specific RNA editomes in the leaf-cutting ant Acromyrmex echinatior. Nature Communications, 2014, 5, 4943.	12.8	60
39	Genome-wide association study dissects the genetic architecture of oil biosynthesis in maize kernels. Nature Genetics, 2013, 45, 43-50.	21.4	764
40	SOAPfusion: a robust and effective computational fusion discovery tool for RNA-seq reads. Bioinformatics, 2013, 29, 2971-2978.	4.1	23
41	RNA sequencing reveals the complex regulatory network in the maize kernel. Nature Communications, 2013, 4, 2832.	12.8	252
42	Lack of evidence for existence of noncanonical RNA editing. Nature Biotechnology, 2013, 31, 19-20.	17.5	87
43	RNA-seq analysis of prostate cancer in the Chinese population identifies recurrent gene fusions, cancer-associated long noncoding RNAs and aberrant alternative splicings. Cell Research, 2012, 22, 806-821.	12.0	352
44	Comprehensive analysis of RNA-Seq data reveals extensive RNA editing in a human transcriptome. Nature Biotechnology, 2012, 30, 253-260.	17.5	461
45	SOAPsplice: Genome-Wide ab initio Detection of Splice Junctions from RNA-Seq Data. Frontiers in Genetics, 2011, 2, 46.	2.3	89
46	A platform of high-density INDEL/CAPS markers for map-based cloning in Arabidopsis. Plant Journal, 2010, 63, 880-888.	5.7	72