Xiaowu gai

List of Publications by Year
in descending order

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93 papers	7,785 citations	66343 42 h-index	54911 84 g-index
107 all docs	107 docs citations	107 times ranked	14120 citing authors

#	Article	IF	Citations
1	m.3685T>C is a Novel Mitochondrial DNA Variant That Causes Leigh Syndrome. Journal of Physical Education and Sports Management, 2022, , mcs.a006136.	1.2	0
2	Mitochondrial 1555 G>A variant as a potential risk factor for childhood glioblastoma. Neuro-Oncology Advances, 2022, 4, vdac045.	0.7	1
3	Implementation of a Streamlined SARS-CoV-2 Whole-Genome Sequencing Assay for Expeditious Surveillance during the Emergence of the Omicron Variant. Journal of Clinical Microbiology, 2022, 60, e0256921.	3.9	4
4	Potential of Aqueous Humor as a Liquid Biopsy for Uveal Melanoma. International Journal of Molecular Sciences, 2022, 23, 6226.	4.1	10
5	Abstract 1967: Potential of aqueous humor as a liquid biopsy for uveal melanoma. Cancer Research, 2022, 82, 1967-1967.	0.9	0
6	High Prevalence of SARS-CoV-2 Genetic Variation and D614G Mutation in Pediatric Patients With COVID-19. Open Forum Infectious Diseases, 2021, 8, ofaa551.	0.9	26
7	Rapidly emerging SARS-CoV-2 B.1.1.7 sub-lineage in the United States of America with spike protein D178H and membrane protein V70L mutations. Emerging Microbes and Infections, 2021, 10, 1293-1299.	6.5	18
8	Establishing the Clinical Utility of ctDNA Analysis for Diagnosis, Prognosis, and Treatment Monitoring of Retinoblastoma: The Aqueous Humor Liquid Biopsy. Cancers, 2021, 13, 1282.	3.7	30
9	Utility of viral whole-genome sequencing for institutional infection surveillance during the coronavirus disease 2019 (COVID-19) pandemic. Infection Control and Hospital Epidemiology, 2021, , 1-2.	1.8	6
10	Emerging variants of concern in SARS-CoV-2 membrane protein: a highly conserved target with potential pathological and therapeutic implications. Emerging Microbes and Infections, 2021, 10, 885-893.	6.5	44
11	Increased viral variants in children and young adults with impaired humoral immunity and persistent SARS-CoV-2 infection: A consecutive case series. EBioMedicine, 2021, 67, 103355.	6.1	128
12	The spectrum of mitochondrial DNA (mtDNA) mutations in pediatric central nervous system (CNS) tumors. Neuro-Oncology Advances, 2021, 3, vdab074.	0.7	3
13	Inter-eye genomic heterogeneity in bilateral retinoblastoma via aqueous humor liquid biopsy. Npj Precision Oncology, 2021, 5, 73.	5.4	8
14	Aqueous Humor as a Liquid Biopsy for Retinoblastoma: Clear Corneal Paracentesis and Genomic Analysis. Journal of Visualized Experiments, 2021, , .	0.3	12
15	Whose Data, Whose Risk? Omics Privacy Concerns Should be Defined by Individuals, not Researchers. American Journal of Bioethics, 2021, 21, 67-70.	0.9	2
16	Bilateral Choroidal Neovascularization and Chorioretinal Anastomosis in Autosomal Recessive Bestrophinopathy. Journal of Vitreoretinal Diseases, 2020, 4, 69-74.	0.7	2
17	Comprehensive Genome Analysis of 6,000 USA SARS-CoV-2 Isolates Reveals Haplotype Signatures and Localized Transmission Patterns by State and by Country. Frontiers in Microbiology, 2020, 11, 573430.	3.5	17
18	Specifications of the ACMG/AMP standards and guidelines for mitochondrial DNA variant interpretation. Human Mutation, 2020, 41, 2028-2057.	2.5	84

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19	Simultaneous identification of clinically relevant RB1 mutations and copy number alterations in aqueous humor of retinoblastoma eyes. Ophthalmic Genetics, 2020, 41, 526-532.	1.2	19
20	Variability in retinoblastoma genome stability is driven by age and not heritability. Genes Chromosomes and Cancer, 2020, 59, 584-590.	2.8	17
21	Mitochondrial Disease Genes Compendium: connecting with knowledge in the Mitochondrial Disease Sequence Data Resource (MSeqDR). , 2020, , 17-23.		0
22	AnthOligo: automating the design of oligonucleotides for capture/enrichment technologies. Bioinformatics, 2020, 36, 4353-4356.	4.1	4
23	Detection of mitochondrial DNA variants at low level heteroplasmy in pediatric CNS and extra-CNS solid tumors with three different enrichment methods. Mitochondrion, 2020, 51, 97-103.	3.4	5
24	Clinical Bioinformatics in Precise Diagnosis of Mitochondrial Disease. Clinics in Laboratory Medicine, 2020, 40, 149-161.	1.4	9
25	Abstract S08-01: Highly sensitive and full-genome interrogation of SARS-CoV-2 using multiplexed PCR enrichment followed by next-generation sequencing. , 2020, , .		2
26	Identification of a novel pathogenic missense mutation inPRPF31using whole exome sequencing: a case report. British Journal of Ophthalmology, 2019, 103, 761-767.	3.9	9
27	Utility of In Vitro Mutagenesis of RPE65 Protein for Verification of Mutational Pathogenicity Before Gene Therapy. JAMA Ophthalmology, 2019, 137, 1381.	2.5	11
28	Landscape of Germline and Somatic Mitochondrial DNA Mutations in Pediatric Malignancies. Cancer Research, 2019, 79, 1318-1330.	0.9	32
29	A semiautomated whole-exome sequencing workflow leads to increased diagnostic yield and identification of novel candidate variants. Journal of Physical Education and Sports Management, 2019, 5, a003756.	1.2	41
30	Resistance to autosomal dominant Alzheimerâ€™s disease in an APOE3 Christchurch homozygote: a case report. Nature Medicine, 2019, 25, 1680-1683.	30.7	328
31	MSeqDR mvTool: A mitochondrial DNA Web and API resource for comprehensive variant annotation, universal nomenclature collation, and reference genome conversion. Human Mutation, 2018, 39, 806-810.	2.5	32
32	On variants and disease-causing mutations: Case studies of a SEMA4A variant identified in inherited blindness. Ophthalmic Genetics, 2018, 39, 144-146.	1.2	4
33	OncoKids. Journal of Molecular Diagnostics, 2018, 20, 765-776.	2.8	58
34	USMG5 Ashkenazi Jewish founder mutation impairs mitochondrial complex V dimerization and ATP synthesis. Human Molecular Genetics, 2018, 27, 3305-3312.	2.9	45
35	Identification of RUNX1 as a Mediator of Aberrant Retinal Angiogenesis. Diabetes, 2017, 66, 1950-1956.	0.6	56
36	Whole exome sequencing identification of novel candidate genes in patients with proliferative diabetic retinopathy. Vision Research, 2017, 139, 168-176.	1.4	33

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37	Inherited germline ATRX mutation in two brothers with ATRâ€X syndrome and osteosarcoma. American Journal of Medical Genetics, Part A, 2017, 173, 1390-1395.	1.2	27
38	Copy-number variation is an important contributor to the genetic causality of inherited retinal degenerations. Genetics in Medicine, 2017, 19, 643-651.	2.4	51
39	Clinical utility of the low-density Infinium QC genotyping Array in a genomics-based diagnostics laboratory. BMC Medical Genomics, 2017, 10, 57.	1.5	6
40	Mitochondrial DNA Depletion Syndromes Presenting in Childhood. , 2016, , 187-198.		1
41	MSeqDR: A Centralized Knowledge Repository and Bioinformatics Web Resource to Facilitate Genomic Investigations in Mitochondrial Disease. Human Mutation, 2016, 37, 540-548.	2.5	42
42	Acute and Chronic Ophthalmic Involvement in Stevens-Johnson Syndrome/Toxic Epidermal Necrolysis â€“ A Comprehensive Review and Guide to Therapy. II. Ophthalmic Disease. Ocular Surface, 2016, 14, 168-188.	4.4	163
43	The next generation of target capture technologies - large DNA fragment enrichment and sequencing determines regional genomic variation of high complexity. BMC Genomics, 2016, 17, 486.	2.8	61
44	Incontinence Medication Response Relates to the Female Urinary Microbiota. Obstetrical and Gynecological Survey, 2016, 71, 464-465.	0.4	0
45	NOD2 genetic variants and sarcoidosis-associated uveitis. American Journal of Ophthalmology Case Reports, 2016, 3, 39-42.	0.7	10
46	From case studies to community knowledge base: MSeqDR provides a platform for the curation and genomic analysis of mitochondrial diseases. Journal of Physical Education and Sports Management, 2016, 2, a001065.	1.2	10
47	Incontinence medication response relates to the female urinary microbiota. International Urogynecology Journal, 2016, 27, 723-733.	1.4	213
48	A novelHSD17B10mutation impairing the activities of the mitochondrial RNase P complex causes X-linked intractable epilepsy and neurodevelopmental regression. RNA Biology, 2016, 13, 477-485.	3.1	42
49	Stevens-Johnson Syndrome/Toxic Epidermal Necrolysis â€“ A Comprehensive Review and Guide to Therapy. I. Systemic Disease. Ocular Surface, 2016, 14, 2-19.	4.4	112
50	Innovative Genomic Collaboration Using the GENESIS (GEM.app) Platform. Human Mutation, 2015, 36, 950-956.	2.5	92
51	Mitochondrial Disease Sequence Data Resource (MSeqDR): A global grass-roots consortium to facilitate deposition, curation, annotation, and integrated analysis of genomic data for the mitochondrial disease clinical and research communities. Molecular Genetics and Metabolism, 2015, 114, 388-396.	1.1	76
52	Phy-Mer: a novel alignment-free and reference-independent mitochondrial haplogroup classifier. Bioinformatics, 2015, 31, 1310-1312.	4.1	55
53	Copy-Number Variation of the Glucose Transporter Gene SLC2A3 and Congenital Heart Defects in the 22q11.2 Deletion Syndrome. American Journal of Human Genetics, 2015, 96, 753-764.	6.2	62
54	Genetic Variants That Predispose to DNA Double-Strand Breaks in Lymphocytes From a Subset of Patients With Familial Colorectal Carcinomas. Gastroenterology, 2015, 149, 1872-1883.e9.	1.3	31

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55	The female urinary microbiome in urgency urinary incontinence. American Journal of Obstetrics and Gynecology, 2015, 213, 347.e1-347.e11.	1.3	244
56	Mutations in IFT172 cause isolated retinal degeneration and Bardetâ€“Biedl syndrome. Human Molecular Genetics, 2015, 24, 230-242.	2.9	136
57	Panel-based genetic diagnostic testing for inherited eye diseases is highly accurate and reproducible, and more sensitive for variant detection, than exome sequencing. Genetics in Medicine, 2015, 17, 253-261.	2.4	216
58	Characterization of cells from patient-derived fibrovascular membranes in proliferative diabetic retinopathy. Molecular Vision, 2015, 21, 673-87.	1.1	10
59	Targeted Exon Sequencing in Usher Syndrome Type I. Investigative Ophthalmology and Visual Science, 2014, 55, 8488-8496.	3.3	24
60	The Female Urinary Microbiome: a Comparison of Women with and without Urgency Urinary Incontinence. MBio, 2014, 5, e01283-14.	4.1	562
61	Single nucleotide polymorphism array analysis of bone marrow failure patients reveals characteristic patterns of genetic changes. British Journal of Haematology, 2014, 164, 73-82.	2.5	18
62	Mitochondrial DNA Variant in COX1 Subunit Significantly Alters Energy Metabolism of Geographically Divergent Wild Isolates in Caenorhabditis elegans. Journal of Molecular Biology, 2014, 426, 2199-2216.	4.2	49
63	AGC1 Deficiency Causes Infantile Epilepsy, Abnormal Myelination, and Reduced N-Acetylaspartate. JIMD Reports, 2014, 14, 77-85.	1.5	57
64	Urine Is Not Sterile: Use of Enhanced Urine Culture Techniques To Detect Resident Bacterial Flora in the Adult Female Bladder. Journal of Clinical Microbiology, 2014, 52, 871-876.	3.9	676
65	Germ-Line and Somatic Mutations in Familial Myeloproliferative Neoplasms (MPNs). a Pilot Study. Blood, 2014, 124, 3214-3214.	1.4	0
66	Mutations in FBXL4, Encoding a Mitochondrial Protein, Cause Early-Onset Mitochondrial Encephalomyopathy. American Journal of Human Genetics, 2013, 93, 482-495.	6.2	138
67	Prevalence of rare mitochondrial DNA mutations in mitochondrial disorders. Journal of Medical Genetics, 2013, 50, 704-714.	3.2	95
68	NMNAT1 mutations cause Leber congenital amaurosis. Nature Genetics, 2012, 44, 1040-1045.	21.4	171
69	Reâ€sequencing of ankyrin 3 exon 48 and caseâ€control association analysis of rare variants in bipolar disorder type I. Bipolar Disorders, 2012, 14, 809-821.	1.9	7
70	Rare structural variation of synapse and neurotransmission genes in autism. Molecular Psychiatry, 2012, 17, 402-411.	7.9	151
71	Mitochondrial disease genetic diagnostics: optimized whole-exome analysis for all MitoCarta nuclear genes and the mitochondrial genome. Discovery Medicine, 2012, 14, 389-99.	0.5	47
72	Implementation of high resolution single nucleotide polymorphism array analysis asÂaÂclinical test for patients with hematologic malignancies. Cancer Genetics, 2011, 204, 26-38.	0.4	29

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73	Microdeletions and Microduplications in Patients with Congenital Heart Disease and Multiple Congenital Anomalies. Congenital Heart Disease, 2011, 6, 592-602.	0.2	82
74	Mitochondrial genome sequence analysis: A custom bioinformatics pipeline substantially improves Affymetrix MitoChip v2.0 call rate and accuracy. BMC Bioinformatics, 2011, 12, 402.	2.6	18
75	CNV Workshop: an integrated platform for high-throughput copy number variation discovery and clinical diagnostics. BMC Bioinformatics, 2010, 11, 74.	2.6	50
76	Genomic alterations in biliary atresia suggest region of potential disease susceptibility in 2q37.3. American Journal of Medical Genetics, Part A, 2010, 152A, 886-895.	1.2	64
77	Rare structural variants found in attention-deficit hyperactivity disorder are preferentially associated with neurodevelopmental genes. Molecular Psychiatry, 2010, 15, 637-646.	7.9	470
78	Next-generation sequencing: the solution for high-resolution, unambiguous human leukocyte antigen typing. Human Immunology, 2010, 71, 1033-1042.	2.4	147
79	High-resolution mapping and analysis of copy number variations in the human genome: A data resource for clinical and research applications. Genome Research, 2009, 19, 1682-1690.	5.5	313
80	Genomic Analysis Using High-Density Single Nucleotide Polymorphism-Based Oligonucleotide Arrays and Multiplex Ligation-Dependent Probe Amplification Provides a Comprehensive Analysis of INI1/SMARCB1 in Malignant Rhabdoid Tumors. Clinical Cancer Research, 2009, 15, 1923-1930.	7.0	190
81	Sequence mining and transcript profiling to explore cyst nematode parasitism. BMC Genomics, 2009, 10, 58.	2.8	43
82	SNP array mapping of chromosome 20p deletions: Genotypes, phenotypes, and copy number variation. Human Mutation, 2009, 30, 371-378.	2.5	61
83	Duplication of 7q34 in Pediatric Lowâ€Grade Astrocytomas Detected by Highâ€Density Singleâ€Nucleotide Polymorphismâ€Based Genotype Arrays Results in a Novel BRAF Fusion Gene. Brain Pathology, 2009, 19, 449-458.	4.1	227
84	A 3.1-Mb microdeletion of 3p21.31 associated with cortical blindness, cleft lip, CNS abnormalities, and developmental delay. European Journal of Medical Genetics, 2009, 52, 265-268.	1.3	21
85	Concept, Design and Implementation of a Cardiovascular Gene-Centric 50 K SNP Array for Large-Scale Genomic Association Studies. PLoS ONE, 2008, 3, e3583.	2.5	339
86	A GATA-1-regulated microRNA locus essential for erythropoiesis. Proceedings of the National Academy of Sciences of the United States of America, 2008, 105, 3333-3338.	7.1	309
87	Divergent evolution of arrested development in the dauer stage of Caenorhabditis elegans and the infective stage of Heterodera glycines. Genome Biology, 2007, 8, R211.	9.6	40
88	Ty5 gag Mutations Increase Retrotransposition and Suggest a Role for Hydrogen Bonding in the Function of the Nucleocapsid Zinc Finger. Journal of Virology, 2002, 76, 3240-3247.	3.4	6
89	Comparison of RNA Expression Profiles Based on Maize Expressed Sequence Tag Frequency Analysis and Micro-Array Hybridization. Plant Physiology, 2002, 128, 896-910.	4.8	96
90	A compilation of soybean ESTs: generation and analysis. Genome, 2002, 45, 329-338.	2.0	133

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91	Targeting of the Yeast Ty5 Retrotransposon to Silent Chromatin Is Mediated by Interactions between Integrase and Sir4p. Molecular and Cellular Biology, 2001, 21, 6606-6614.	2.3	143
92	Gene discovery using the maize genome database ZmDB. Nucleic Acids Research, 2000, 28, 94-96.	14.5	58
93	A Single Amino Acid Change in the Yeast Retrotransposon Ty5 Abolishes Targeting to Silent Chromatin. Molecular Cell, 1998, 1, 1051-1055.	9.7	55