Xiangdong Kong

List of Publications by Year
in descending order

68 papers	749 citations	567281 15 h-index	642732 23 g-index
82 all docs	82 docs citations	82 times ranked	925 citing authors

#	Article	IF	Citations
1	Elf3 drives Î²-catenin transactivation and associates with poor prognosis in colorectal cancer. Cell Death and Disease, 2014, 5, e1263-e1263.	6.3	69
2	PBX1 expression in uterine natural killer cells drives fetal growth. Science Translational Medicine, 2020, 12, .	12.4	54
3	Jumonji domain-containing protein 2B silencing induces DNA damage response via STAT3 pathway in colorectal cancer. British Journal of Cancer, 2014, 110, 1014-1026.	6.4	37
4	Targeted Next-Generation Sequencing for Clinical Diagnosis of 561 Mendelian Diseases. PLoS ONE, 2015, 10, e0133636.	2.5	33
5	Spectrum of PAH gene variants among a population of Han Chinese patients with phenylketonuria from northern China. BMC Medical Genetics, 2017, 18, 108.	2.1	32
6	Molecular genetic characterization of cblC defects in 126 pedigrees and prenatal genetic diagnosis of pedigrees with combined methylmalonic aciduria and homocystinuria. BMC Medical Genetics, 2018, 19, 154.	2.1	31
7	Novel mutations of COL4A3, COL4A4, and COL4A5 genes in Chinese patients with Alport Syndrome using next generation sequence technique. Molecular Genetics & Genomic Medicine, 2019, 7, e653.	1.2	28
8	Co-expression network analysis of Down's syndrome based on microarray data. Experimental and Therapeutic Medicine, 2016, 12, 1503-1508.	1.8	25
9	Incorporation of exomeâ€based CNV analysis makes trioâ€WES a more powerful tool for clinical diagnosis in neurodevelopmental disorders: A retrospective study. Human Mutation, 2021, 42, 990-1004.	2.5	25
10	DJ-1 deficiency attenuates expansion of liver progenitor cells through modulating the inflammatory and fibrogenic niches. Cell Death and Disease, 2016, 7, e2257-e2257.	6.3	23
11	Altered gut microbiota in RA: implications for treatment. Zeitschrift Fur Rheumatologie, 2017, 76, 451-457.	1.0	23
12	Non-invasive prenatal testing of pregnancies at risk for phenylketonuria. Archives of Disease in Childhood: Fetal and Neonatal Edition, 2019, 104, F24-F29.	2.8	20
13	Rapid prenatal diagnosis of Facioscapulohumeral Muscular Dystrophy 1 by combined Bionano optical mapping and karyomapping. Prenatal Diagnosis, 2020, 40, 317-323.	2.3	20
14	The potential of expanded noninvasive prenatal screening for detection of microdeletion and microduplication syndromes. Prenatal Diagnosis, 2021, 41, 1332-1342.	2.3	19
15	Development of coupling controlled polymerizations by adapter-ligation in mate-pair sequencing for detection of various genomic variants in one single assay. DNA Research, 2019, 26, 313-325.	3.4	17
16	Exploring the cause of early miscarriage with SNP-array analysis and karyotyping. Journal of Maternal-Fetal and Neonatal Medicine, 2019, 32, 1-10.	1.5	16
17	Deciphering the complexity of simple chromosomal insertions by genome sequencing. Human Genetics, 2021, 140, 361-380.	3.8	15
18	A Novel Five-Gene Signature for Prognosis Prediction in Hepatocellular Carcinoma. Frontiers in Oncology, 2021, 11, 642563.	2.8	14

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19	Rapid Diagnosis of Aneuploidy Using Segmental Duplication Quantitative Fluorescent PCR. PLoS ONE, 2014, 9, e88932.	2.5	12
20	Evaluation of optical genome mapping for detecting chromosomal translocation in clinical cytogenetics. Molecular Genetics & Genomic Medicine, 2022, 10, e1936.	1.2	12
21	Prognostic Significance of Pregnancy Zone Protein and Its Correlation with Immune Infiltrates in Hepatocellular Carcinoma . Cancer Management and Research, 2020, Volume 12, 9883-9891.	1.9	11
22	Investigation of the genetic etiology in male infertility with apparently balanced chromosomal structural rearrangements by genome sequencing. Asian Journal of Andrology, 2022, 24, 248.	1.6	11
23	Prenatal diagnosis using genetic sequencing and identification of a novel mutation in MMACHC. BMC Medical Genetics, 2015, 16, 48.	2.1	10
24	Mutation analysis of 419 family and prenatal diagnosis of 339 cases of spinal muscular atrophy in China. BMC Medical Genetics, 2020, 21, 133.	2.1	10
25	Recent advances of glucocorticoids in the treatment of Duchenne muscular dystrophy (Review). Experimental and Therapeutic Medicine, 2021, 21, 447.	1.8	10
26	Influence of validating the parental origin on the clinical interpretation of fetal copy number variations in 141 core family cases. Molecular Genetics & Genomic Medicine, 2019, 7, e00944.	1.2	9
27	A case of prenatal diagnosis of 18p deletion syndrome following noninvasive prenatal testing. Molecular Cytogenetics, 2019, 12, 53.	0.9	9
28	Clinical and Genetic Characteristics of 17 Î±-Hydroxylase/17, 20-Lyase Deficiency: c.985_987delTACinsAA Mutation of CYP17A1 Prevalent in the Chinese Han Population. Endocrine Practice, 2021, 27, 137-145.	2.1	9
29	Clinical Application of Noninvasive Prenatal Testing for Sex Chromosome Aneuploidies in Central China. Frontiers in Medicine, 2021, 8, 672211.	2.6	9
30	Development of CRISPR-Mediated Systems in the Study of Duchenne Muscular Dystrophy. Human Gene Therapy Methods, 2019, 30, 71-80.	2.1	8
31	Mutation analysis in the F8 gene in 485 families with haemophilia A and prenatal diagnosis in China. Haemophilia, 2021, 27, e88-e92.	2.1	8
32	Novel mutations of the CYP17A1 gene in four Chinese 46,XX cases with partial 17a-hydroxylase/17,20-lyase deficiency. Steroids, 2021, 173, 108873.	1.8	8
33	Extension of the mutation spectrum of <scp>PAX</scp>6 from three Chinese congenital aniridia families and identification of male gonadal mosaicism. Molecular Genetics & Genomic Medicine, 2018, 6, 1053-1067.	1.2	7
34	Clinical Significance of Non-Invasive Prenatal Screening for Trisomy 7: Cohort Study and Literature Review. Genes, 2021, 12, 11.	2.4	7
35	A novel NAPB splicing mutation identified by Trio-based exome sequencing is associated with early-onset epileptic encephalopathy. European Journal of Medical Genetics, 2021, 64, 104101.	1.3	6
36	A de novo and novel nonsense variants in ASXL2 gene is associated with Shashiâ€“Pena syndrome. European Journal of Medical Genetics, 2022, 65, 104454.	1.3	6

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37	Contiguous 22.1-kb deletion embracing AVPR2 and ARHGAP4 genes at novel breakpoints leads to nephrogenic diabetes insipidus in a Chinese pedigree. BMC Nephrology, 2018, 19, 26.	1.8	5
38	A novel PCDH19 missense mutation, c.812G>A (p.Gly271Asp), identified using wholeâ€exome sequencing in a Chinese family with epilepsy female restricted mental retardation syndrome. Molecular Genetics & Genomic Medicine, 2020, 8, e1234.	1.2	5
39	Noninvasive prenatal diagnosis of duchenne muscular dystrophy in five Chinese families based on relative mutation dosage approach. BMC Medical Genomics, 2021, 14, 275.	1.5	5
40	Genetic Surveillance of Five SARS-CoV-2 Clinical Samples in Henan Province Using Nanopore Sequencing. Frontiers in Immunology, 2022, 13, 814806.	4.8	5
41	Haplotype-Based Noninvasive Prenatal Diagnosis of 21 Families With Duchenne Muscular Dystrophy: Real-World Clinical Data in China. Frontiers in Genetics, 2021, 12, 791856.	2.3	5
42	Targeted nextâ€generation sequencing identifies nine novel filaggrin gene variants in Chinese Han patients with ichthyosis vulgaris. British Journal of Dermatology, 2017, 177, e202-e203.	1.5	4
43	Identification of two novel SMN1 point mutations associated with a very severe SMA-I phenotype. European Journal of Medical Genetics, 2020, 63, 104006.	1.3	4
44	Identification of six novel variants in Waardenburg syndrome type II by nextâ€generation sequencing. Molecular Genetics & Genomic Medicine, 2020, 8, e1128.	1.2	4
45	Identification of four novel mutations in the. Biomedical Reports, 2020, 13, 4.	2.0	4
46	Molecular analysis and novel variation identification of Chinese pedigrees with mucopolysaccharidosis using targeted next-generation sequencing. Clinica Chimica Acta, 2022, 524, 194-200.	1.1	4
47	Novel Partial Exon 51 Deletion in the Duchenne Muscular Dystrophy Gene Identified via Whole Exome Sequencing and Long-Read Whole-Genome Sequencing. Frontiers in Genetics, 2021, 12, 762987.	2.3	4
48	Transcatheter Closure of Secundum Atrial Septal Defect with a New Self-Expanding Nitinol Double Disk Device (Amplatzer Device): Experience in Nanjing. Journal of Interventional Cardiology, 2001, 14, 193-196.	1.2	3
49	Genetic investigation of 211 Chinese families expands the mutational and phenotypical spectra of hereditary retinopathy genes through targeted sequencing technology. BMC Medical Genomics, 2021, 14, 92.	1.5	3
50	Identification of a Rare Case With Nagashima-Type Palmoplantar Keratoderma and 18q Deletion Syndrome via Exome Sequencing and Low-Coverage Whole-Genome Sequencing. Frontiers in Genetics, 2021, 12, 707411.	2.3	3
51	Noninvasive prenatal testing of Duchenne muscular dystrophy in a twin gestation. Prenatal Diagnosis, 2022, , .	2.3	3
52	Identification of 4 novel mutations of androgen receptor gene in 8 Chinese families with complete androgen insensitivity syndrome. Clinical Genetics, 2018, 94, 269-270.	2.0	2
53	Hermanskyâ€“Pudlak syndrome: Five Chinese patients with novel variants in HPS1 and HPS6. European Journal of Medical Genetics, 2021, 64, 104228.	1.3	2
54	DMD/BMD prenatal diagnosis and treatment expectation in a single centre in China for 15Âyears. BMC Medical Genomics, 2021, 14, 181.	1.5	2

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55	Genetic analysis and identification of novel variations in Chinese patients with pediatric epilepsy by whole-exome sequencing. Neurological Sciences, 2022, 43, 4439-4451.	1.9	2
56	The genotype analysis and prenatal genetic diagnosis among 244 pedigrees with methylmalonic aciduria in China. Taiwanese Journal of Obstetrics and Gynecology, 2022, 61, 290-298.	1.3	2
57	Three novel variants (p.Glu178Lys, p.Val245Met, p.Ser250Phe) of the phenylalanine hydroxylase (PAH) gene impair protein expression and function in vitro. Gene, 2018, 668, 135-139.	2.2	1
58	Generation of ZZUi008-A, a transgene-free, induced pluripotent stem cell line derived from chorionic villi cells of a fetus with Duchenne muscular dystrophy. Stem Cell Research, 2018, 32, 47-50.	0.7	1
59	Usefulness of copy number variant detection following monogenic disease exclusion in prenatal diagnosis. Journal of Obstetrics and Gynaecology Research, 2021, 47, 1002-1008.	1.3	1
60	Prenatal and postnatal diagnoses and phenotype of 8p23.3p22 duplication in one family. BMC Medical Genomics, 2021, 14, 88.	1.5	1
61	A novel compound heterozygous variant in SMARCAL1 leading to mild Schimke immune-osseous dysplasia identified using whole-exome sequencing. Journal of International Medical Research, 2021, 49, 030006052110106.	1.0	1
62	Molecular study and genotypeâ€“phenotype in Chinese female patients with 46, XY disorders of sex development. Gynecological Endocrinology, 2021, 37, 934-940.	1.7	1
63	Two females presenting primary amenorrhea diagnosed with Kallmann syndrome caused by novel FGFR1 variants. Journal of Obstetrics and Gynaecology Research, 2021, 47, 3727-3731.	1.3	1
64	Genetic analysis by targeted next-generation sequencing and novel variation identification of maple syrup urine disease in Chinese Han population. Scientific Reports, 2021, 11, 18939.	3.3	1
65	Case Report: A Novel Homozygous Mutation in MYF5 Due to Paternal Uniparental Isodisomy of Chromosome 12 in a Case of External Ophthalmoplegia With Rib and Vertebral Anomalies. Frontiers in Genetics, 2021, 12, 780363.	2.3	1
66	A novel variant in <scp>UBE3A</scp> in a family with multigenerational intellectual disability and developmental delay. Molecular Genetics & Genomic Medicine, 2022, , e1883.	1.2	1
67	A Chinese child with hyperpigmentation diagnosed with familial glucocorticoid deficiency type 1 using whole-exome sequencing. Pediatrics and Neonatology, 2021, 62, 229-230.	0.9	0
68	FVIII inhibitor risk correlated with F8 gene variants in 296 unrelated male Chinese patients with haemophilia A. Haemophilia, 2021, 27, e274-e277.	2.1	0