# Xiangdong Kong

List of Publications by Year
in descending order



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|---|---|---|---|
| 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 | 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 |
| 2 | 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 |
| 3 | 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 |
| 4 | 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 |
| 5 | Noninvasive prenatal testing of Duchenne muscular dystrophy in a twin gestation. Prenatal Diagnosis, 2022, , . | 2.3 | 3 |
| 6 | 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 |
| 7 | Evaluation of optical genome mapping for detecting chromosomal translocation in clinical cytogenetics. Molecular Genetics & Genomic Medicine, 2022, 10, e1936. | 1.2 | 12 |
| 8 | Genetic Surveillance of Five SARS-CoV-2 Clinical Samples in Henan Province Using Nanopore Sequencing. Frontiers in Immunology, 2022, 13, 814806. | 4.8 | 5 |
| 9 | 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 |
| 10 | Deciphering the complexity of simple chromosomal insertions by genome sequencing. Human Genetics, 2021, 140, 361-380. | 3.8 | 15 |
| 11 | 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 |
| 12 | 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 |
| 13 | 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 |
| 14 | 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 |
| 15 | 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 |
| 16 | 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 |
| 17 | Recent advances of glucocorticoids in the treatment of Duchenne muscular dystrophy (Review). Experimental and Therapeutic Medicine, 2021, 21, 447. | 1.8 | 10 |
| 18 | Prenatal and postnatal diagnoses and phenotype of 8p23.3p22 duplication in one family. BMC Medical Genomics, 2021, 14, 88. | 1.5 | 1 |

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|---|---|---|---|
| 19 | 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 |
| 20 | 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 |
| 21 | 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 |
| 22 | Hermanskyâ€“Pudlak syndrome: Five Chinese patients with novel variants in HPS1 and HPS6. European Journal of Medical Genetics, 2021, 64, 104228. | 1.3 | 2 |
| 23 | DMD/BMD prenatal diagnosis and treatment expectation in a single centre in China for 15Âyears. BMC Medical Genomics, 2021, 14, 181. | 1.5 | 2 |
| 24 | The potential of expanded noninvasive prenatal screening for detection of microdeletion and microduplication syndromes. Prenatal Diagnosis, 2021, 41, 1332-1342. | 2.3 | 19 |
| 25 | A Novel Five-Gene Signature for Prognosis Prediction in Hepatocellular Carcinoma. Frontiers in Oncology, 2021, 11, 642563. | 2.8 | 14 |
| 26 | 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 |
| 27 | 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 |
| 28 | 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 |
| 29 | 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 |
| 30 | 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 |
| 31 | Clinical Significance of Non-Invasive Prenatal Screening for Trisomy 7: Cohort Study and Literature Review. Genes, 2021, 12, 11. | 2.4 | 7 |
| 32 | 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 |
| 33 | 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 |
| 34 | Clinical Application of Noninvasive Prenatal Testing for Sex Chromosome Aneuploidies in Central China. Frontiers in Medicine, 2021, 8, 672211. | 2.6 | 9 |
| 35 | 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 |
| 36 | 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 |

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|---|---|---|---|
| 37 | Rapid prenatal diagnosis of Facioscapulohumeral Muscular Dystrophy 1 by combined Bionano optical mapping and karyomapping. Prenatal Diagnosis, 2020, 40, 317-323. | 2.3 | 20 |
| 38 | 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 |
| 39 | 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 |
| 40 | 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 |
| 41 | Identification of six novel variants in Waardenburg syndrome type II by nextâ€generation sequencing. Molecular Genetics & Genomic Medicine, 2020, 8, e1128. | 1.2 | 4 |
| 42 | PBX1 expression in uterine natural killer cells drives fetal growth. Science Translational Medicine, 2020, 12, . | 12.4 | 54 |
| 43 | 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 |
| 44 | Identification of four novel mutations in the. Biomedical Reports, 2020, 13, 4. | 2.0 | 4 |
| 45 | 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 |
| 46 | 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 |
| 47 | Development of CRISPR-Mediated Systems in the Study of Duchenne Muscular Dystrophy. Human Gene Therapy Methods, 2019, 30, 71-80. | 2.1 | 8 |
| 48 | 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 |
| 49 | A case of prenatal diagnosis of 18p deletion syndrome following noninvasive prenatal testing. Molecular Cytogenetics, 2019, 12, 53. | 0.9 | 9 |
| 50 | 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 |
| 51 | 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 |
| 52 | 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 |
| 53 | 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 |
| 54 | 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 |

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|---|---|---|---|
| 55 | 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 |
| 56 | 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 |
| 57 | 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 |
| 58 | 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 |
| 59 | Altered gut microbiota in RA: implications for treatment. Zeitschrift Fur Rheumatologie, 2017, 76, 451-457. | 1.0 | 23 |
| 60 | 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 |
| 61 | Co-expression network analysis of Down's syndrome based on microarray data. Experimental and Therapeutic Medicine, 2016, 12, 1503-1508. | 1.8 | 25 |
| 62 | 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 |
| 63 | Prenatal diagnosis using genetic sequencing and identification of a novel mutation in MMACHC. BMC Medical Genetics, 2015, 16, 48. | 2.1 | 10 |
| 64 | Targeted Next-Generation Sequencing for Clinical Diagnosis of 561 Mendelian Diseases. PLoS ONE, 2015, 10, e0133636. | 2.5 | 33 |
| 65 | Elf3 drives Î²-catenin transactivation and associates with poor prognosis in colorectal cancer. Cell Death and Disease, 2014, 5, e1263-e1263. | 6.3 | 69 |
| 66 | 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 |
| 67 | Rapid Diagnosis of Aneuploidy Using Segmental Duplication Quantitative Fluorescent PCR. PLoS ONE, 2014, 9, e88932. | 2.5 | 12 |
| 68 | 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 |