Juan Du

List of Publications by Year
in descending order

62 papers	1,347 citations	394421 19 h-index	414414 32 g-index
72 all docs	72 docs citations	72 times ranked	1533 citing authors

#	Article	IF	Citations
1	BiallelicCFAP61variants cause male infertility in humans and mice with severe oligoasthenoteratozoospermia. Journal of Medical Genetics, 2023, 60, 144-153.	3.2	12
2	Sperm flagellar 2 (SPEF2) is essential for sperm flagellar assembly in humans. Asian Journal of Andrology, 2022, 24, 359.	1.6	13
3	Bi-allelic variants in DNHD1 cause flagellar axoneme defects and asthenoteratozoospermia in humans and mice. American Journal of Human Genetics, 2022, 109, 157-171.	6.2	17
4	RNA splicing analysis contributes to reclassifying variants of uncertain significance and improves the diagnosis of monogenic disorders. Journal of Medical Genetics, 2022, 59, 1010-1016.	3.2	5
5	A novel <scp>FAM83H</scp> variant causes familial amelogenesis imperfecta with incomplete penetrance. Molecular Genetics & Genomic Medicine, 2022, 10, e1902.	1.2	3
6	Meiotic recombination: insights into its mechanisms and its role in human reproduction with a special focus on non-obstructive azoospermia. Human Reproduction Update, 2022, 28, 763-797.	10.8	27
7	Bi-allelic variants in SHOC1 cause non-obstructive azoospermia with meiosis arrest in humans and mice. Molecular Human Reproduction, 2022, 28, .	2.8	7
8	Homozygous variants in SYCP2L cause premature ovarian insufficiency. Journal of Medical Genetics, 2021, 58, 168-172.	3.2	16
9	Novel homozygous truncating variants in ZMYND15 causing severe oligozoospermia and their implications for male infertility. Human Mutation, 2021, 42, 31-36.	2.5	5
10	Bi-allelic BRWD1 variants cause male infertility with asthenoteratozoospermia and likely primary ciliary dyskinesia. Human Genetics, 2021, 140, 761-773.	3.8	23
11	A novel homozygous frameshift mutation in MNS1 associated with severe oligoasthenoteratozoospermia in humans. Asian Journal of Andrology, 2021, 23, 197.	1.6	8
12	Improving native human sperm freezing protection by using a modified vitrification method. Asian Journal of Andrology, 2021, 23, 91.	1.6	6
13	Deleterious variants in X-linked CFAP47 induce asthenoteratozoospermia and primary male infertility. American Journal of Human Genetics, 2021, 108, 309-323.	6.2	74
14	Reproductive risks and preimplantation genetic testing intervention for Xâ€“autosome translocation carriers. Reproductive BioMedicine Online, 2021, 43, 73-80.	2.4	3
15	TDRD7 participates in lens development and spermiogenesis by mediating autophagosome maturation. Autophagy, 2021, 17, 3848-3864.	9.1	19
16	RNF216 regulates meiosis and PKA stability in the testes. FASEB Journal, 2021, 35, e21460.	0.5	6
17	Clinical and genetic analysis of classical Ehlersâ€Danlos syndrome patient caused by synonymous mutation in COL5A2. Molecular Genetics & Genomic Medicine, 2021, 9, e1632.	1.2	4
18	Nextâ€generation sequenceâ€based preimplantation genetic testing for monogenic disease resulting from maternal mosaicism. Molecular Genetics & Genomic Medicine, 2021, 9, e1662.	1.2	6

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19	CFAP65 is required in the acrosome biogenesis and mitochondrial sheath assembly during spermiogenesis. Human Molecular Genetics, 2021, 30, 2240-2254.	2.9	18
20	Loss-of-function missense variant ofAKAP4induced male infertility through reduced interaction with QRICH2 during sperm flagella development. Human Molecular Genetics, 2021, 31, 219-231.	2.9	19
21	Novel variants of the PCCB gene in Chinese patients with propionic acidemia. Clinica Chimica Acta, 2021, 519, 18-25.	1.1	1
22	Bi-allelic mutations of DNAH10 cause primary male infertility with asthenoteratozoospermia in humans and mice. American Journal of Human Genetics, 2021, 108, 1466-1477.	6.2	50
23	A recurrent mutation in TBPL2 causes diminished ovarian reserve and female infertility. Journal of Genetics and Genomics, 2020, 47, 785-788.	3.9	5
24	Genetic underpinnings of asthenozoospermia. Best Practice and Research in Clinical Endocrinology and Metabolism, 2020, 34, 101472.	4.7	37
25	Whole-genome mate-pair sequencing of apparently balanced chromosome rearrangements reveals complex structural variations: two case studies. Molecular Cytogenetics, 2020, 13, 15.	0.9	7
26	Novel DNAAF6 variants identified by whole-exome sequencing cause male infertility and primary ciliary dyskinesia. Journal of Assisted Reproduction and Genetics, 2020, 37, 811-820.	2.5	9
27	A homozygous RPL10L missense mutation associated with maleÂfactorÂinfertility and severe oligozoospermia. Fertility and Sterility, 2020, 113, 561-568.	1.0	10
28	Novel lossâ€ofâ€function mutation in MCM8 causes premature ovarian insufficiency. Molecular Genetics & Genomic Medicine, 2020, 8, e1165.	1.2	22
29	Novel mutations in SPEF2 causing different defects between flagella and cilia bridge: the phenotypic link between MMAF and PCD. Human Genetics, 2020, 139, 257-271.	3.8	46
30	An M1AP homozygous spliceâ€site mutation associated with severe oligozoospermia in a consanguineous family. Clinical Genetics, 2020, 97, 741-746.	2.0	15
31	Insight on multiple morphological abnormalities of sperm flagella in male infertility: what is new?. Asian Journal of Andrology, 2020, 22, 236.	1.6	42
32	ZP2 pathogenic variantsÂcause in vitro fertilization failure and female infertility. Genetics in Medicine, 2019, 21, 431-440.	2.4	69
33	Next-generation sequencing identified a novel SPTB frameshift insertion causing hereditary spherocytosis in China. Annals of Hematology, 2019, 98, 223-226.	1.8	4
34	Analysis of molecular cytogenetic features and PGT-SR for two infertile patients with small supernumerary marker chromosomes. Journal of Assisted Reproduction and Genetics, 2019, 36, 2533-2539.	2.5	5
35	Xâ€chromosome inactivation pattern of amniocytes predicts the risk of dystrophinopathy in fetal carriers of DMD mutations. Prenatal Diagnosis, 2019, 39, 603-608.	2.3	7
36	Advances in Identification of Susceptibility Gene Defects of Hereditary Colorectal Cancer. Journal of Cancer, 2019, 10, 643-653.	2.5	15

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37	Identification of DNAH6 mutations in infertile men with multiple morphological abnormalities of the sperm flagella. Scientific Reports, 2019, 9, 15864.	3.3	42
38	In-Frame Variants in STAG3 Gene Cause Premature Ovarian Insufficiency. Frontiers in Genetics, 2019, 10, 1016.	2.3	26
39	Novel inactivating mutations in the FSH receptor cause premature ovarian insufficiency with resistant ovary syndrome. Reproductive BioMedicine Online, 2019, 38, 397-406.	2.4	23
40	XRCC2 mutation causes premature ovarian insufficiency as well as nonâ€obstructive azoospermia in humans. Clinical Genetics, 2019, 95, 442-443.	2.0	35
41	Loss-of-function mutations in TDRD7 lead to a rare novel syndrome combining congenital cataract and nonobstructive azoospermia in humans. Genetics in Medicine, 2019, 21, 1209-1217.	2.4	70
42	Biallelic mutations in CFAP65 lead to severe asthenoteratospermia due to acrosome hypoplasia and flagellum malformations. Journal of Medical Genetics, 2019, 56, 750-757.	3.2	61
43	Novel <scp>FOXL</scp>2 mutations cause blepharophimosisâ€ptosisâ€epicanthus inversus syndrome with premature ovarian insufficiency. Molecular Genetics & Genomic Medicine, 2018, 6, 261-267.	1.2	14
44	DMC1 mutation that causes human non-obstructive azoospermia and premature ovarian insufficiency identified by whole-exome sequencing. Journal of Medical Genetics, 2018, 55, 198-204.	3.2	91
45	Maternal interchromosomal insertional translocation leading to 1q43-q44 deletion and duplication in two siblings. Molecular Cytogenetics, 2018, 11, 24.	0.9	11
46	Wholeâ€exome sequencing identifies a homozygous donor spliceâ€site mutation in <scp>STAG3</scp> that causes primary ovarian insufficiency. Clinical Genetics, 2018, 93, 340-344.	2.0	42
47	Long-read sequencing identified a causal structural variant in an exome-negative case and enabled preimplantation genetic diagnosis. Hereditas, 2018, 155, 32.	1.4	47
48	Novel mutations of PKD genes in Chinese patients suffering from autosomal dominant polycystic kidney disease and seeking assisted reproduction. BMC Medical Genetics, 2018, 19, 186.	2.1	9
49	Expanded carrier screening and preimplantation genetic diagnosis in a couple who delivered a baby affected with congenital factor VII deficiency. BMC Medical Genetics, 2018, 19, 15.	2.1	3
50	Rare partial octosomy and hexasomy of 15q11-q13 associated with intellectual impairment and development delay: report of two cases and review of literature. Molecular Cytogenetics, 2018, 11, 15.	0.9	5
51	Inner cell mass incarceration in 8-shaped blastocysts does not increase monozygotic twinning in preimplantation genetic diagnosis and screening patients. PLoS ONE, 2018, 13, e0190776.	2.5	13
52	A rare polypyrimidine tract mutation in the androgen receptor gene results in complete androgen insensitivity syndrome. Asian Journal of Andrology, 2018, 20, 308.	1.6	4
53	Genotype-phenotype correlation and identification of two novel SRD5A2 mutations in 33 Chinese patients with hypospadias. Steroids, 2017, 125, 61-66.	1.8	13
54	Noninvasive prenatal testing (NIPT) in twin pregnancies with treatment of assisted reproductive techniques (ART) in a single center. Prenatal Diagnosis, 2016, 36, 672-679.	2.3	49

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55	The first patient with a pure 1p36 microtriplication associated with severe clinical phenotypes. Molecular Cytogenetics, 2014, 7, 64.	0.9	10
56	Single-nucleotide polymorphism microarray-based preimplantation genetic diagnosis is likely to improve the clinical outcome for translocation carriers. Human Reproduction, 2013, 28, 2581-2592.	0.9	86
57	Study of the Sperm Chromosomal Aneuploidies of Isolated Teratozoospermic Men. Reproduction and Contraception, 2013, 24, 1-9.	0.1	1
58	Dppa2 knockdown-induced differentiation and repressed proliferation of mouse embryonic stem cells. Journal of Biochemistry, 2010, 147, 265-271.	1.7	35
59	Mutation Analysis and Prenatal Exclusion of Fibrodysplasia Ossificans Progressiva in a Chinese Fetus. Genetic Testing and Molecular Biomarkers, 2010, , 110306133116090.	0.7	3
60	Non-invasive prenatal molecular detection of a fetal point mutation for congenital adrenal hyperplasia using co-amplification at lower denaturation temperature PCR. Chinese Medical Journal, 2010, 123, 3343-6.	2.3	4
61	Prenatal diagnosis of nonmosaic tetrasomy 9p by microdissection and FISH: case report. Chinese Medical Journal, 2007, 120, 1281-3.	2.3	1
62	Genetic Changes in Human Fetuses from Spontaneous Abortion after In Vitro Fertilization Detected by Comparative Genomic Hybridization1. Biology of Reproduction, 2004, 70, 495-499.	2.7	11