

Juan Du

List of Publications by Year in descending order

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Version: 2024-02-01

62
papers

1,347
citations

393982

19
h-index

414034

32
g-index

72
all docs

72
docs citations

72
times ranked

1533
citing authors

#	ARTICLE	IF	CITATIONS
1	Biallelic <i>CFAP61</i> variants cause male infertility in humans and mice with severe oligoasthenoteratozoospermia. <i>Journal of Medical Genetics</i> , 2023, 60, 144-153.	1.5	12
2	Sperm flagellar 2 (SPEF2) is essential for sperm flagellar assembly in humans. <i>Asian Journal of Andrology</i> , 2022, 24, 359.	0.8	13
3	Bi-allelic variants in <i>DNHD1</i> cause flagellar axoneme defects and asthenoteratozoospermia in humans and mice. <i>American Journal of Human Genetics</i> , 2022, 109, 157-171.	2.6	17
4	RNA splicing analysis contributes to reclassifying variants of uncertain significance and improves the diagnosis of monogenic disorders. <i>Journal of Medical Genetics</i> , 2022, 59, 1010-1016.	1.5	5
5	A novel <i>FAM83H</i> variant causes familial amelogenesis imperfecta with incomplete penetrance. <i>Molecular Genetics & Genomic Medicine</i> , 2022, 10, e1902.	0.6	3
6	Meiotic recombination: insights into its mechanisms and its role in human reproduction with a special focus on non-obstructive azoospermia. <i>Human Reproduction Update</i> , 2022, 28, 763-797.	5.2	27
7	Bi-allelic variants in <i>SHOC1</i> cause non-obstructive azoospermia with meiosis arrest in humans and mice. <i>Molecular Human Reproduction</i> , 2022, 28, .	1.3	7
8	Homozygous variants in <i>SYCP2L</i> cause premature ovarian insufficiency. <i>Journal of Medical Genetics</i> , 2021, 58, 168-172.	1.5	16
9	Novel homozygous truncating variants in <i>ZMYND15</i> causing severe oligozoospermia and their implications for male infertility. <i>Human Mutation</i> , 2021, 42, 31-36.	1.1	5
10	Bi-allelic <i>BRWD1</i> variants cause male infertility with asthenoteratozoospermia and likely primary ciliary dyskinesia. <i>Human Genetics</i> , 2021, 140, 761-773.	1.8	23
11	A novel homozygous frameshift mutation in <i>MNS1</i> associated with severe oligoasthenoteratozoospermia in humans. <i>Asian Journal of Andrology</i> , 2021, 23, 197.	0.8	8
12	Improving native human sperm freezing protection by using a modified vitrification method. <i>Asian Journal of Andrology</i> , 2021, 23, 91.	0.8	6
13	Deleterious variants in X-linked <i>CFAP47</i> induce asthenoteratozoospermia and primary male infertility. <i>American Journal of Human Genetics</i> , 2021, 108, 309-323.	2.6	74
14	Reproductive risks and preimplantation genetic testing intervention for X-autosome translocation carriers. <i>Reproductive BioMedicine Online</i> , 2021, 43, 73-80.	1.1	3
15	<i>TDRD7</i> participates in lens development and spermiogenesis by mediating autophagosome maturation. <i>Autophagy</i> , 2021, 17, 3848-3864.	4.3	19
16	<i>RNF216</i> regulates meiosis and PKA stability in the testes. <i>FASEB Journal</i> , 2021, 35, e21460.	0.2	6
17	Clinical and genetic analysis of classical Ehlers-Danlos syndrome patient caused by synonymous mutation in <i>COL5A2</i> . <i>Molecular Genetics & Genomic Medicine</i> , 2021, 9, e1632.	0.6	4
18	Next-generation sequence-based preimplantation genetic testing for monogenic disease resulting from maternal mosaicism. <i>Molecular Genetics & Genomic Medicine</i> , 2021, 9, e1662.	0.6	6

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19	CFAP65 is required in the acrosome biogenesis and mitochondrial sheath assembly during spermiogenesis. <i>Human Molecular Genetics</i> , 2021, 30, 2240-2254.	1.4	18
20	Loss-of-function missense variant of <i>AKAP4</i> induced male infertility through reduced interaction with QRICH2 during sperm flagella development. <i>Human Molecular Genetics</i> , 2021, 31, 219-231.	1.4	19
21	Novel variants of the PCCB gene in Chinese patients with propionic acidemia. <i>Clinica Chimica Acta</i> , 2021, 519, 18-25.	0.5	1
22	Bi-allelic mutations of DNAH10 cause primary male infertility with asthenoteratozoospermia in humans and mice. <i>American Journal of Human Genetics</i> , 2021, 108, 1466-1477.	2.6	50
23	A recurrent mutation in TBPL2 causes diminished ovarian reserve and female infertility. <i>Journal of Genetics and Genomics</i> , 2020, 47, 785-788.	1.7	5
24	Genetic underpinnings of asthenozoospermia. <i>Best Practice and Research in Clinical Endocrinology and Metabolism</i> , 2020, 34, 101472.	2.2	37
25	Whole-genome mate-pair sequencing of apparently balanced chromosome rearrangements reveals complex structural variations: two case studies. <i>Molecular Cytogenetics</i> , 2020, 13, 15.	0.4	7
26	Novel DNAAF6 variants identified by whole-exome sequencing cause male infertility and primary ciliary dyskinesia. <i>Journal of Assisted Reproduction and Genetics</i> , 2020, 37, 811-820.	1.2	9
27	A homozygous RPL10L missense mutation associated with male factor infertility and severe oligozoospermia. <i>Fertility and Sterility</i> , 2020, 113, 561-568.	0.5	10
28	Novel loss-of-function mutation in <i>MCM8</i> causes premature ovarian insufficiency. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1165.	0.6	22
29	Novel mutations in SPEF2 causing different defects between flagella and cilia bridge: the phenotypic link between MMAF and PCD. <i>Human Genetics</i> , 2020, 139, 257-271.	1.8	46
30	An <i>M1AP</i> homozygous splice-site mutation associated with severe oligozoospermia in a consanguineous family. <i>Clinical Genetics</i> , 2020, 97, 741-746.	1.0	15
31	Insight on multiple morphological abnormalities of sperm flagella in male infertility: what is new?. <i>Asian Journal of Andrology</i> , 2020, 22, 236.	0.8	42
32	ZP2 pathogenic variants cause in vitro fertilization failure and female infertility. <i>Genetics in Medicine</i> , 2019, 21, 431-440.	1.1	69
33	Next-generation sequencing identified a novel SPTB frameshift insertion causing hereditary spherocytosis in China. <i>Annals of Hematology</i> , 2019, 98, 223-226.	0.8	4
34	Analysis of molecular cytogenetic features and PGT-SR for two infertile patients with small supernumerary marker chromosomes. <i>Journal of Assisted Reproduction and Genetics</i> , 2019, 36, 2533-2539.	1.2	5
35	X-chromosome inactivation pattern of amniocytes predicts the risk of dystrophinopathy in fetal carriers of DMD mutations. <i>Prenatal Diagnosis</i> , 2019, 39, 603-608.	1.1	7
36	Advances in Identification of Susceptibility Gene Defects of Hereditary Colorectal Cancer. <i>Journal of Cancer</i> , 2019, 10, 643-653.	1.2	15

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37	Identification of DNAH6 mutations in infertile men with multiple morphological abnormalities of the sperm flagella. <i>Scientific Reports</i> , 2019, 9, 15864.	1.6	42
38	In-Frame Variants in STAG3 Gene Cause Premature Ovarian Insufficiency. <i>Frontiers in Genetics</i> , 2019, 10, 1016.	1.1	26
39	Novel inactivating mutations in the FSH receptor cause premature ovarian insufficiency with resistant ovary syndrome. <i>Reproductive BioMedicine Online</i> , 2019, 38, 397-406.	1.1	23
40	<i>XRCC2</i> mutation causes premature ovarian insufficiency as well as non-obstructive azoospermia in humans. <i>Clinical Genetics</i> , 2019, 95, 442-443.	1.0	35
41	Loss-of-function mutations in TDRD7 lead to a rare novel syndrome combining congenital cataract and nonobstructive azoospermia in humans. <i>Genetics in Medicine</i> , 2019, 21, 1209-1217.	1.1	70
42	Biallelic mutations in <i>CFAP65</i> lead to severe asthenoteratospermia due to acrosome hypoplasia and flagellum malformations. <i>Journal of Medical Genetics</i> , 2019, 56, 750-757.	1.5	61
43	Novel <i>FOXL2</i> mutations cause blepharophimosis-ptosis-epicanthus inversus syndrome with premature ovarian insufficiency. <i>Molecular Genetics & Genomic Medicine</i> , 2018, 6, 261-267.	0.6	14
44	<i>DMC1</i> mutation that causes human non-obstructive azoospermia and premature ovarian insufficiency identified by whole-exome sequencing. <i>Journal of Medical Genetics</i> , 2018, 55, 198-204.	1.5	91
45	Maternal interchromosomal insertional translocation leading to 1q43-q44 deletion and duplication in two siblings. <i>Molecular Cytogenetics</i> , 2018, 11, 24.	0.4	11
46	Whole-exome sequencing identifies a homozygous donor splice-site mutation in <i>STAG3</i> that causes primary ovarian insufficiency. <i>Clinical Genetics</i> , 2018, 93, 340-344.	1.0	42
47	Long-read sequencing identified a causal structural variant in an exome-negative case and enabled preimplantation genetic diagnosis. <i>Hereditas</i> , 2018, 155, 32.	0.5	47
48	Novel mutations of PKD genes in Chinese patients suffering from autosomal dominant polycystic kidney disease and seeking assisted reproduction. <i>BMC Medical Genetics</i> , 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. <i>BMC Medical Genetics</i> , 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. <i>Molecular Cytogenetics</i> , 2018, 11, 15.	0.4	5
51	Inner cell mass incarceration in 8-shaped blastocysts does not increase monozygotic twinning in preimplantation genetic diagnosis and screening patients. <i>PLoS ONE</i> , 2018, 13, e0190776.	1.1	13
52	A rare polypyrimidine tract mutation in the androgen receptor gene results in complete androgen insensitivity syndrome. <i>Asian Journal of Andrology</i> , 2018, 20, 308.	0.8	4
53	Genotype-phenotype correlation and identification of two novel SRD5A2 mutations in 33 Chinese patients with hypospadias. <i>Steroids</i> , 2017, 125, 61-66.	0.8	13
54	Noninvasive prenatal testing (NIPT) in twin pregnancies with treatment of assisted reproductive techniques (ART) in a single center. <i>Prenatal Diagnosis</i> , 2016, 36, 672-679.	1.1	49

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55	The first patient with a pure 1p36 microtriplication associated with severe clinical phenotypes. <i>Molecular Cytogenetics</i> , 2014, 7, 64.	0.4	10
56	Single-nucleotide polymorphism microarray-based preimplantation genetic diagnosis is likely to improve the clinical outcome for translocation carriers. <i>Human Reproduction</i> , 2013, 28, 2581-2592.	0.4	86
57	Study of the Sperm Chromosomal Aneuploidies of Isolated Teratozoospermic Men. <i>Reproduction and Contraception</i> , 2013, 24, 1-9.	0.1	1
58	Dppa2 knockdown-induced differentiation and repressed proliferation of mouse embryonic stem cells. <i>Journal of Biochemistry</i> , 2010, 147, 265-271.	0.9	35
59	Mutation Analysis and Prenatal Exclusion of Fibrodysplasia Ossificans Progressiva in a Chinese Fetus. <i>Genetic Testing and Molecular Biomarkers</i> , 2010, , 110306133116090.	0.3	3
60	Non-invasive prenatal molecular detection of a fetal point mutation for congenital adrenal hyperplasia using co-amplification at lower denaturation temperature PCR. <i>Chinese Medical Journal</i> , 2010, 123, 3343-6.	0.9	4
61	Prenatal diagnosis of nonmosaic tetrasomy 9p by microdissection and FISH: case report. <i>Chinese Medical Journal</i> , 2007, 120, 1281-3.	0.9	1
62	Genetic Changes in Human Fetuses from Spontaneous Abortion after In Vitro Fertilization Detected by Comparative Genomic Hybridization1. <i>Biology of Reproduction</i> , 2004, 70, 495-499.	1.2	11