Donald Conrad

List of Publications by Year
in descending order

43 papers	6,857 citations	279798 23 h-index	265206 42 g-index
54 all docs	54 docs citations	54 times ranked	14139 citing authors

#	Article	IF	Citations
1	A de novo paradigm for male infertility. Nature Communications, 2022, 13, 154.	12.8	38
2	Actionable secondary findings following exome sequencing of 836 non-obstructive azoospermia cases and their value in patient management. Human Reproduction, 2022, 37, 1652-1663.	0.9	3
3	Large-scale analyses of the X chromosome in 2,354 infertile men discover recurrently affected genes associated with spermatogenic failure. American Journal of Human Genetics, 2022, 109, 1458-1471.	6.2	10
4	A framework for high-resolution phenotyping of candidate male infertility mutants: from human to mouse. Human Genetics, 2021, 140, 155-182.	3.8	33
5	Disruption of human meiotic telomere complex genes TERB1, TERB2 and MAJIN in men with non-obstructive azoospermia. Human Genetics, 2021, 140, 217-227.	3.8	31
6	The Sertoli cell expressed gene secerninâ€1 (Scrn1) is dispensable for male fertility in the mouse. Developmental Dynamics, 2021, 250, 922-931.	1.8	12
7	Lack of evidence for a role of PIWIL1 variants in human male infertility. Cell, 2021, 184, 1941-1942.	28.9	11
8	Variants in GCNA, X-linked germ-cell genome integrity gene, identified in men with primary spermatogenic failure. Human Genetics, 2021, 140, 1169-1182.	3.8	27
9	Population-scale tissue transcriptomics maps long non-coding RNAs to complex disease. Cell, 2021, 184, 2633-2648.e19.	28.9	94
10	Variant PNLDC1, Defective piRNA Processing, and Azoospermia. New England Journal of Medicine, 2021, 385, 707-719.	27.0	54
11	Comparative single-cell analysis of biopsies clarifies pathogenic mechanisms in Klinefelter syndrome. American Journal of Human Genetics, 2021, 108, 1924-1945.	6.2	29
12	Identification of genetic variants in CFAP221 as a cause of primary ciliary dyskinesia. Journal of Human Genetics, 2020, 65, 175-180.	2.3	27
13	Bi-allelic Mutations in M1AP Are a Frequent Cause of Meiotic Arrest and Severely Impaired Spermatogenesis Leading to Male Infertility. American Journal of Human Genetics, 2020, 107, 342-351.	6.2	68
14	Mutation of CFAP57, a protein required for the asymmetric targeting of a subset of inner dynein arms in Chlamydomonas, causes primary ciliary dyskinesia. PLoS Genetics, 2020, 16, e1008691.	3.5	36
15	Genetic dissection of spermatogenic arrest through exome analysis: clinical implications for the management of azoospermic men. Genetics in Medicine, 2020, 22, 1956-1966.	2.4	88
16	Rare mutations in the complement regulatory gene CSMD1 are associated with male and female infertility. Nature Communications, 2019, 10, 4626.	12.8	24
17	Unified single-cell analysis of testis gene regulation and pathology in five mouse strains. ELife, 2019, 8, .	6.0	102
18	Genetic intersection of male infertility and cancer. Fertility and Sterility, 2018, 109, 20-26.	1.0	50

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19	Bi-allelic Recessive Loss-of-Function Variants in FANCM Cause Non-obstructive Azoospermia. American Journal of Human Genetics, 2018, 103, 200-212.	6.2	95
20	Multiplex shRNA Screening of Germ Cell Development by in Vivo Transfection of Mouse Testis. G3: Genes, Genomes, Genetics, 2017, 7, 247-255.	1.8	2
21	The impact of structural variation on human gene expression. Nature Genetics, 2017, 49, 692-699.	21.4	334
22	Dynamic landscape and regulation of RNA editing in mammals. Nature, 2017, 550, 249-254.	27.8	495
23	The long and short of translational control in male germ cellsâ€. Biology of Reproduction, 2017, 97, 2-4.	2.7	0
24	A Standardized Approach for Multispecies Purification of Mammalian Male Germ Cells by Mechanical Tissue Dissociation and Flow Cytometry. Journal of Visualized Experiments, 2017, , .	0.3	8
25	How to Map the Genetic Basis for Conditions that are Comorbid with Male Infertility. Seminars in Reproductive Medicine, 2017, 35, 225-230.	1.1	1
26	Estimating error models for whole genome sequencing using mixtures of Dirichlet-multinomial distributions. Bioinformatics, 2017, 33, 2322-2329.	4.1	16
27	Multispecies Purification of Testicular Germ Cells. Biology of Reproduction, 2016, 95, 85-85.	2.7	10
28	Genome-wide significance testing of variation from single case exomes. Nature Genetics, 2016, 48, 1455-1461.	21.4	43
29	Defining the Phenotype and Assessing Severity in Phosphoglucomutase-1ÂDeficiency. Journal of Pediatrics, 2016, 175, 130-136.e8.	1.8	43
30	Improved detection of diseaseâ€associated variation by sexâ€specific characterization and prediction of genes required for fertility. Andrology, 2015, 3, 1140-1149.	3.5	2
31	Rare double sex and mab-3-related transcription factor 1 regulatory variants in severe spermatogenic failure. Andrology, 2015, 3, 825-833.	3.5	17
32	Low-frequency germline variants across 6p22.2â€“6p21.33 are associated with non-obstructive azoospermia in Han Chinese men. Human Molecular Genetics, 2015, 24, 5628-5636.	2.9	12
33	Using whole-genome sequences of the LG/J and SM/J inbred mouse strains to prioritize quantitative trait genes and nucleotides. BMC Genomics, 2015, 16, 415.	2.8	31
34	A Screen for Genomic Disorders of Infertility Identifies MAST2 Duplications Associated with Nonobstructive Azoospermia in Humans1. Biology of Reproduction, 2015, 93, 61.	2.7	30
35	Cis and Trans Effects of Human Genomic Variants on Gene Expression. PLoS Genetics, 2014, 10, e1004461.	3.5	117
36	Guidelines for investigating causality of sequence variants in human disease. Nature, 2014, 508, 469-476.	27.8	1,130

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37	Validating single-cell genomics for the study of renal development. Kidney International, 2014, 86, 1049-1055.	5.2	3
38	Human Spermatogenic Failure Purges Deleterious Mutation Load from the Autosomes and Both Sex Chromosomes, including the Gene DMRT1. PLoS Genetics, 2013, 9, e1003349.	3.5	118
39	Meeting on big mutations addresses big questions in human genetics. Genome Medicine, 2011, 3, 12.	8.2	1
40	Variation in genome-wide mutation rates within and between human families. Nature Genetics, 2011, 43, 712-714.	21.4	525
41	Origins and functional impact of copy number variation in the human genome. Nature, 2010, 464, 704-712.	27.8	1,721
42	Genome-wide association study of CNVs in 16,000 cases of eight common diseases and 3,000 shared controls. Nature, 2010, 464, 713-720.	27.8	737
43	Recurrent 16p11.2 microdeletions in autism. Human Molecular Genetics, 2007, 17, 628-638.	2.9	614