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	Origins and functional impact of copy number variation in the human genome. Nature, 2010, 464, 704-712.	27.8	1,721
2	Guidelines for investigating causality of sequence variants in human disease. Nature, 2014, 508, 469-476.	27.8	1,130
3	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
4	Recurrent 16p11.2 microdeletions in autism. Human Molecular Genetics, 2007, 17, 628-638.	2.9	614
5	Variation in genome-wide mutation rates within and between human families. Nature Genetics, 2011, 43, 712-714.	21.4	525
6	Dynamic landscape and regulation of RNA editing in mammals. Nature, 2017, 550, 249-254.	27.8	495
7	The impact of structural variation on human gene expression. Nature Genetics, 2017, 49, 692-699.	21.4	334
8	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
9	Cis and Trans Effects of Human Genomic Variants on Gene Expression. PLoS Genetics, 2014, 10, e1004461.	3.5	117
10	Unified single-cell analysis of testis gene regulation and pathology in five mouse strains. ELife, 2019, 8, .	6.0	102
11	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
12	Population-scale tissue transcriptomics maps long non-coding RNAs to complex disease. Cell, 2021, 184, 2633-2648.e19.	28.9	94
13	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
14	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
15	Variant PNLDC1, Defective piRNA Processing, and Azoospermia. New England Journal of Medicine, 2021, 385, 707-719.	27.0	54
16	Genetic intersection of male infertility and cancer. Fertility and Sterility, 2018, 109, 20-26.	1.0	50
17	Genome-wide significance testing of variation from single case exomes. Nature Genetics, 2016, 48, 1455-1461.	21.4	43
18	Defining the Phenotype and Assessing Severity in Phosphoglucomutase-1ÂDeficiency. Journal of Pediatrics, 2016, 175, 130-136.e8.	1.8	43

#	Article	IF	Citations
19	A de novo paradigm for male infertility. Nature Communications, 2022, 13, 154.	12.8	38
20	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
21	A framework for high-resolution phenotyping of candidate male infertility mutants: from human to mouse. Human Genetics, 2021, 140, 155-182.	3.8	33
22	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
23	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
24	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
25	Comparative single-cell analysis of biopsies clarifies pathogenic mechanisms in Klinefelter syndrome. American Journal of Human Genetics, 2021, 108, 1924-1945.	6.2	29
26	Identification of genetic variants in CFAP221 as a cause of primary ciliary dyskinesia. Journal of Human Genetics, 2020, 65, 175-180.	2.3	27
27	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
28	Rare mutations in the complement regulatory gene CSMD1 are associated with male and female infertility. Nature Communications, 2019, 10, 4626.	12.8	24
29	Rare double sex and mab-3-related transcription factor 1 regulatory variants in severe spermatogenic failure. Andrology, 2015, 3, 825-833.	3.5	17
30	Estimating error models for whole genome sequencing using mixtures of Dirichlet-multinomial distributions. Bioinformatics, 2017, 33, 2322-2329.	4.1	16
31	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
32	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
33	Lack of evidence for a role of PIWIL1 variants in human male infertility. Cell, 2021, 184, 1941-1942.	28.9	11
34	Multispecies Purification of Testicular Germ Cells. Biology of Reproduction, 2016, 95, 85-85.	2.7	10
35	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
36	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

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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	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
39	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
40	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
41	Meeting on big mutations addresses big questions in human genetics. Genome Medicine, 2011, 3, 12.	8.2	1
42	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
43	The long and short of translational control in male germ cellsâ€. Biology of Reproduction, 2017, 97, 2-4.	2.7	0