Frank TÃ¼ttelmann

List of Publications by Year
in descending order

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101 papers	4,458 citations	126708 33 h-index	123241 61 g-index
128 all docs	128 docs citations	128 times ranked	3987 citing authors

#	Article	IF	Citations
1	Human INHBB Gene Variant (c.1079T>C:p.Met360Thr) Alters Testis Germ Cell Content, but Does Not Impact Fertility in Mice. Endocrinology, 2022, 163, .	1.4	2
2	Machine learning based prediction models in male reproductive health: Development of a proofâ€ofâ€concept model for Klinefelter Syndrome in azoospermic patients. Andrology, 2022, 10, 534-544.	1.9	6
3	A de novo paradigm for male infertility. Nature Communications, 2022, 13, 154.	5.8	38
4	Strukturelle ChromosomenverÃnderungen. Springer Reference Medizin, 2022, , 1-6.	0.0	0
5	Zyto- und molekulargenetische Untersuchungen. Springer Reference Medizin, 2022, , 1-8.	0.0	0
6	Protocol for developing a core outcome set for male infertility research: an international consensus development study. Human Reproduction Open, 2022, 2022, hoac014.	2.3	4
7	A GWAS in Idiopathic/Unexplained Infertile Men Detects a Genomic Region Determining Follicle-Stimulating Hormone Levels. Journal of Clinical Endocrinology and Metabolism, 2022, 107, 2350-2361.	1.8	4
8	O-153â€ƒUnravelling the causes of male infertility: the role of genetics. Human Reproduction, 2022, 37, .	0.4	0
9	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.	2.6	10
10	Disruption of human meiotic telomere complex genes TERB1, TERB2 and MAJIN in men with non-obstructive azoospermia. Human Genetics, 2021, 140, 217-227.	1.8	31
11	European academy of andrology guidelines on Klinefelter Syndrome Endorsing Organization: European Society of Endocrinology. Andrology, 2021, 9, 145-167.	1.9	86
12	The X chromosome and male infertility. Human Genetics, 2021, 140, 203-215.	1.8	40
13	A global approach to addressing the policy, research and social challenges of male reproductive health. Human Reproduction Open, 2021, 2021, hoab009.	2.3	19
14	Defects in the cytoplasmic assembly of axonemal dynein arms cause morphological abnormalities and dysmotility in sperm cells leading to male infertility. PLoS Genetics, 2021, 17, e1009306.	1.5	50
15	Reply: CFTR analysis should not be offered to all patients with unexplained azoospermia in the presence of normal gonadotropin levels. Human Reproduction, 2021, 36, 2067-2068.	0.4	0
16	Pituitary response to GnRH stimulation tests in different FSHB-211 G/T genotypes. Human Reproduction, 2021, 36, 1376-1382.	0.4	4
17	Lack of evidence for a role of PIWIL1 variants in human male infertility. Cell, 2021, 184, 1941-1942.	13.5	11
18	Variants in GCNA, X-linked germ-cell genome integrity gene, identified in men with primary spermatogenic failure. Human Genetics, 2021, 140, 1169-1182.	1.8	27

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19	TRIM71 Deficiency Causes Germ Cell Loss During Mouse Embryogenesis and Is Associated With Human Male Infertility. Frontiers in Cell and Developmental Biology, 2021, 9, 658966.	1.8	17
20	Can Unlikely Neanderthal Chloride Channel CLC-2 Gene Variants Provide Insights in Modern Human Infertility?. Cellular Physiology and Biochemistry, 2021, 55, 301-310.	1.1	0
21	O-089 A Genome Wide Association Study in men with unexplained infertility identifies nine SNPs at the FSHB locus to be associated with Follicle Stimulating Hormone level. Human Reproduction, 2021, 36, .	0.4	0
22	O-118 New insight into the genetic contribution of common variants to the development of extreme phenotypes of unexplained male infertility: a multicenter genome-wide association study. Human Reproduction, 2021, 36, .	0.4	0
23	A systematic review of the validated monogenic causes of human male infertility: 2020 update and a discussion of emerging geneâ€“disease relationships. Human Reproduction Update, 2021, 28, 15-29.	5.2	121
24	Single-cell RNA-seq unravels alterations of the human spermatogonial stem cell compartment in patients with impaired spermatogenesis. Cell Reports Medicine, 2021, 2, 100395.	3.3	33
25	Andrological findings in infertile men with two (biallelic) CFTR mutations: results of a multicentre study in Germany and Austria comprising 71 patients. Human Reproduction, 2021, 36, 551-559.	0.4	19
26	Bi-allelic variants in DNA mismatch repair proteins MutS Homolog MSH4 and MSH5 cause infertility in both sexes. Human Reproduction, 2021, 37, 178-189.	0.4	18
27	Genetic counseling and diagnostic guidelines for couples with infertility and/or recurrent miscarriage. Medizinische Genetik, 2021, 33, 3-12.	0.1	2
28	Effect of Genetic Variants of Gonadotropins and Their Receptors on Ovarian Stimulation Outcomes: A Delphi Consensus. Frontiers in Endocrinology, 2021, 12, 797365.	1.5	9
29	Sequence analysis of 37 candidate genes for male infertility: challenges in variant assessment and validating genes. Andrology, 2020, 8, 434-441.	1.9	40
30	SYCP2 Translocation-Mediated Dysregulation and Frameshift Variants Cause Human Male Infertility. American Journal of Human Genetics, 2020, 106, 41-57.	2.6	66
31	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.	2.6	68
32	The Ca2+ channel CatSper is not activated by cAMP/PKA signaling but directly affected by chemicals used to probe the action of cAMP and PKA. Journal of Biological Chemistry, 2020, 295, 13181-13193.	1.6	27
33	Genetic dissection of spermatogenic arrest through exome analysis: clinical implications for the management of azoospermic men. Genetics in Medicine, 2020, 22, 1956-1966.	1.1	88
34	Testicular blood supply is altered in the 41,XXY* Klinefelter syndrome mouse model. Scientific Reports, 2020, 10, 14369.	1.6	5
35	A germ cellâ€specific ageing pattern in otherwise healthy men. Aging Cell, 2020, 19, e13242.	3.0	27
36	The Male Fertility Gene Atlas: a web tool for collecting and integrating OMICS data in the context of male infertility. Human Reproduction, 2020, 35, 1983-1990.	0.4	13

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37	European Academy of Andrology (EAA) guidelines on investigation, treatment and monitoring of functional hypogonadism in males. Andrology, 2020, 8, 970-987.	1.9	230
38	Rotational motion and rheotaxis of human sperm do not require functional CatSper channels and transmembrane Ca ²⁺ signaling. EMBO Journal, 2020, 39, e102363.	3.5	42
39	Initial experience with [18F]DPA-714 TSPO-PET to image inflammation in primary angiitis of the central nervous system. European Journal of Nuclear Medicine and Molecular Imaging, 2020, 47, 2131-2141.	3.3	14
40	Mutations in the stromal antigen 3 (STAG3) gene cause male infertility due to meiotic arrest. Human Reproduction, 2019, 34, 2112-2119.	0.4	43
41	The ReproGenomics Viewer: a multi-omics and cross-species resource compatible with single-cell studies for the reproductive science community. Bioinformatics, 2019, 35, 3133-3139.	1.8	49
42	FSHB â^'211 G>T Polymorphism as Predictor for TESE Success in Patients With Unexplained Azoospermia. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 2315-2324.	1.8	25
43	Diagnosis and Treatment Before Assisted Reproductive Treatments. Guideline of the DGGG, OEGGG and SGGG (S2k Level, AWMF Register Number 015-085, February 2019) â€“ Part 2, Hemostaseology, Andrology, Genetics and History of Malignant Disease. Geburtshilfe Und Frauenheilkunde, 2019, 79, 1293-1308.	0.8	17
44	Diagnosis and Therapy Before Assisted Reproductive Treatments. Guideline of the DGGG, OEGGG and SGGG (S2k Level, AWMF Register Number 015-085, February 2019) â€“ Part 1, Basic Assessment of the Woman. Geburtshilfe Und Frauenheilkunde, 2019, 79, 1278-1292.	0.8	25
45	Interest in, willingness-to-pay for and willingness-to-recommend genetic testing for prostate cancer among affected men after radical prostatectomy. Familial Cancer, 2019, 18, 221-230.	0.9	4
46	Disorders of spermatogenesis. Medizinische Genetik, 2018, 30, 12-20.	0.1	156
47	Action of steroids and plant triterpenoids on CatSper Ca ²⁺ channels in human sperm. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, E344-E346.	3.3	33
48	Is the FSHR 2039A>G variant associated with susceptibility to testicular germ cell cancer?. Andrology, 2018, 6, 176-183.	1.9	6
49	A no-stop mutation in MAGEB4 is a possible cause of rare X-linked azoospermia and oligozoospermia in a consanguineous Turkish family. Journal of Assisted Reproduction and Genetics, 2017, 34, 683-694.	1.2	38
50	MECHANISMS IN ENDOCRINOLOGY: Aberrations of the X chromosome as cause of male infertility. European Journal of Endocrinology, 2017, 177, R249-R259.	1.9	18
51	Tubulin-related cerebellar dysplasia: definition of a distinct pattern of cerebellar malformation. European Radiology, 2017, 27, 5080-5092.	2.3	36
52	Genetics of Male Infertility. Endocrinology, 2017, , 1-21.	0.1	0
53	Genetics of Male Infertility. Endocrinology, 2017, , 1029-1049.	0.1	0
54	The humanRHOXgene cluster: target genes and functional analysis of gene variants in infertile men. Human Molecular Genetics, 2016, 25, ddw313.	1.4	25

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55	FSHB â€211G>T stratification for follicleâ€stimulating hormone treatment of male infertility patients: making the case for a pharmacogenetic approach in genetic functional secondary hypogonadism. Andrology, 2015, 3, 1050-1053.	1.9	20
56	X-Linked TEX11 Mutations, Meiotic Arrest, and Azoospermia in Infertile Men. New England Journal of Medicine, 2015, 372, 2097-2107.	13.9	279
57	Gene Expression Patterns in Relation to the Clinical Phenotype in Klinefelter Syndrome. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E518-E523.	1.8	80
58	The FSHB â^'211G>T variant attenuates serum FSH levels in the supraphysiological gonadotropin setting of Klinefelter syndrome. European Journal of Human Genetics, 2015, 23, 700-703.	1.4	18
59	Three novel presenilin 1 mutations marking the wide spectrum of age at onset and clinical patterns in familial Alzheimerâ€™s disease. Journal of Neural Transmission, 2015, 122, 1715-1719.	1.4	8
60	Azoospermia and ring chromosome 9â€"a case report. Journal of Assisted Reproduction and Genetics, 2015, 32, 293-296.	1.2	6
61	Restoration of fertility by gonadotropin replacement in a man with hypogonadotropic azoospermia and testicular adrenal rest tumors due to untreated simple virilizing congenital adrenal hyperplasia. European Journal of Endocrinology, 2014, 170, K11-K17.	1.9	19
62	ESHRE Task Force on Ethics and Law 21: genetic screening of gamete donors: ethical issues. Human Reproduction, 2014, 29, 1353-1359.	0.4	49
63	Reply: Y-chromosome microdeletions are not associated with SHOX haploinsufficiency. Human Reproduction, 2014, 29, 1114-1115.	0.4	0
64	Intratesticular testosterone is increased in men with Klinefelter syndrome and may not be released into the bloodstream owing to altered testicular vascularization - a preliminary report. Andrology, 2014, 2, 275-281.	1.9	54
65	Epigenetic regulation of the RHOX homeobox gene cluster and its association with human male infertility. Human Molecular Genetics, 2014, 23, 12-23.	1.4	45
66	<scp>EAA</scp>/<scp>EMQN</scp> best practice guidelines for molecular diagnosis of Yâ€chromosomal microdeletions: stateâ€ofâ€theâ€art 2013. Andrology, 2014, 2, 5-19.	1.9	356
67	Lower Incidence of M2/ANXA5 Carriage in Recurrent Pregnancy Loss Patients With Elevated Lipoprotein(a) Levels. Clinical and Applied Thrombosis/Hemostasis, 2014, 20, 706-709.	0.7	4
68	DMRT1 mutations are rarely associated with male infertility. Fertility and Sterility, 2014, 102, 816-820.e3.	0.5	28
69	TheTRHRGene Is Associated with Hypothalamo-Pituitary Sensitivity to Levothyroxine. European Thyroid Journal, 2014, 3, 101-108.	1.2	10
70	The haplotype M2 of the ANXA5 gene is not associated with antitrophoblast antibodies. Journal of Assisted Reproduction and Genetics, 2013, 30, 711-716.	1.2	7
71	Raman microspectroscopic discrimination of TCam-2 cultures reveals the presence of two sub-populations of cells. Cell and Tissue Research, 2013, 354, 623-632.	1.5	7
72	DNA methylation in spermatozoa as a prospective marker in andrology. Andrology, 2013, 1, 731-740.	1.9	70

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73	Mosaicism for an unbalanced Y;21 translocation in an infertile man: a case report. Journal of Assisted Reproduction and Genetics, 2013, 30, 1553-1558.	1.2	8
74	Further insights into the role of the annexin A5 M2 haplotype as recurrent pregnancy loss factor, assessing timing of miscarriage and partner risk. Fertility and Sterility, 2013, 100, 1321-1325.	0.5	32
75	Comprehensive sequence analysis of the NR5A1 gene encoding steroidogenic factor 1 in a large group of infertile males. European Journal of Human Genetics, 2013, 21, 1012-1015.	1.4	80
76	Independent association of the M2/ANXA5 haplotype with recurrent pregnancy loss (RPL) in PCOS patients. Metabolism: Clinical and Experimental, 2013, 62, 1057-1060.	1.5	13
77	Effects of the FSH-Î²-Subunit Promoter Polymorphism â^'211Gâ†'T on the Hypothalamic-Pituitary-Ovarian Axis in Normally Cycling Women Indicate a Gender-Specific Regulation of Gonadotropin Secretion. Journal of Clinical Endocrinology and Metabolism, 2013, 98, E82-E86.	1.8	36
78	Y-chromosome microdeletions are not associated with SHOX haploinsufficiency. Human Reproduction, 2013, 28, 3155-3160.	0.4	13
79	In Klinefelter men and mouse models reduced testicular vascularization may contribute to low serum testosterone (T) levels despite normal intratesticular T (ITT) concentrations. Experimental and Clinical Endocrinology and Diabetes, 2013, 121, .	0.6	0
80	Influence of variants in the FSHB and FSHR gene on reproductive parameters in males and females. Experimental and Clinical Endocrinology and Diabetes, 2013, 121, .	0.6	0
81	Paternal and maternal carriage ofÂtheÂannexin A5 M2 haplotype areÂequal risk factors for recurrent pregnancy loss: a pilot study. Fertility and Sterility, 2012, 98, 383-388.	0.5	42
82	Combined Effects of the VariantsFSHBâ^'211G>T andFSHR2039A>G on Male Reproductive Parameters. Journal of Clinical Endocrinology and Metabolism, 2012, 97, 3639-3647.	1.8	116
83	The future of testis research is turning 6! Six years of International Network for Young Researchers in Male Fertility. Journal of Developmental and Physical Disabilities, 2012, 35, 211-213.	3.6	4
84	Clinical experience with azoospermia: aetiology and chances for spermatozoa detection upon biopsy. Journal of Developmental and Physical Disabilities, 2011, 34, 291-298.	3.6	132
85	Genetische Aspekte bei SpermatogenesestÃ¶rungen. Medizinische Genetik, 2011, 23, 259-266.	0.1	2
86	Hypogonadotroper Hypogonadismus aufgrund eines IHH oder Kallmann-Syndroms beim Mann. Medizinische Genetik, 2011, 23, 254-258.	0.1	5
87	Copy Number Variants in Patients with Severe Oligozoospermia and Sertoli-Cell-Only Syndrome. PLoS ONE, 2011, 6, e19426.	1.1	129
88	Aquaporins in the human testis and spermatozoa â€“ identification, involvement in sperm volume regulation and clinical relevance. Journal of Developmental and Physical Disabilities, 2010, 33, 629-641.	3.6	50
89	Idiopathic male infertility is strongly associated with aberrant methylation of MEST and IGF2/H19 ICR1. Journal of Developmental and Physical Disabilities, 2010, 33, 642-649.	3.6	185
90	A common haplotype of protamine 1 and 2 genes is associated with higher sperm counts. Journal of Developmental and Physical Disabilities, 2010, 33, e240-8.	3.6	37

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91	Novel genetic aspects of Klinefelter's syndrome. Molecular Human Reproduction, 2010, 16, 386-395.	1.3	136
92	Classification of Andrological Disorders. , 2010, , 87-92.		22
93	Coiled sperm from infertile patients: characteristics, associated factors and biological implication. Human Reproduction, 2009, 24, 1288-1295.	0.4	40
94	Role of syndecan-3 polymorphisms in obesity and female hyperandrogenism. Journal of Molecular Medicine, 2009, 87, 1241-1250.	1.7	12
95	Anti-MÃ¼llerian hormone in men with normal and reduced sperm concentration and men with maldescended testes. Fertility and Sterility, 2009, 91, 1812-1819.	0.5	68
96	Clinical consequences of microdeletions of the Y chromosome: the extended MÃ¼nster experience. Reproductive BioMedicine Online, 2008, 16, 289-303.	1.1	167
97	Polymorphisms of the luteinizing hormone/chorionic gonadotropin receptor gene: association with maldescended testes and male infertility. Pharmacogenetics and Genomics, 2008, 18, 193-200.	0.7	53
98	Gene polymorphisms and male infertility â€“ a meta-analysis and literature review. Reproductive BioMedicine Online, 2007, 15, 643-658.	1.1	205
99	The presence of germ cells in the semen of azoospermic, cryptozoospermic and severe oligozoospermic patients: stringent flow cytometric analysis and correlations with hormonal status. Clinical Endocrinology, 2007, 67, 767-775.	1.2	17
100	Optimising workflow in andrology: a new electronic patient record and database. Asian Journal of Andrology, 2006, 8, 235-241.	0.8	38
101	Inhibition of Muscarinic Potassium Current by the Class III Antiarrhythmic Drug RP58866 in Guineaâ€Pig Atrial Myocytes. PACE - Pacing and Clinical Electrophysiology, 2000, 23, 1812-1815.	0.5	5