

Filipa Carvalho

List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

83
papers

2,137
citations

22
h-index

44
g-index

94
ext. papers

2,544
ext. citations

5.1
avg, IF

4.27
L-index

#	Paper	IF	Citations
83	A de novo paradigm for male infertility.. <i>Nature Communications</i> , 2022 , 13, 154	17.4	2
82	Common Variation in the PIN1 Locus Increases the Genetic Risk to Suffer from Sertoli Cell-Only Syndrome. <i>Journal of Personalized Medicine</i> , 2022 , 12, 932	3.6	
81	Congenital Adrenal Hyperplasia Due to 21-Hydroxylase Deficiency: An Update on Genetic Analysis of CYP21A2 Gene. <i>Experimental and Clinical Endocrinology and Diabetes</i> , 2021 , 129, 477-481	2.3	4
80	Effect and in silico characterization of genetic variants associated with severe spermatogenic disorders in a large Iberian cohort. <i>Andrology</i> , 2021 , 9, 1151-1165	4.2	2
79	Variants in GCNA, X-linked germ-cell genome integrity gene, identified in men with primary spermatogenic failure. <i>Human Genetics</i> , 2021 , 140, 1169-1182	6.3	3
78	ESHRE PGT Consortium data collection XIX-XX: PGT analyses from 2016 to 2017. <i>Human Reproduction Open</i> , 2021 , 2021, hoab024	6.1	8
77	Variant , Defective piRNA Processing, and Azoospermia. <i>New England Journal of Medicine</i> , 2021 , 385, 707-719	59.2	8
76	Mindfulness and self-compassion based intervention program to prevent burnout in medical and dentistry students. <i>European Psychiatry</i> , 2021 , 64, S459-S460	6	
75	Habits and quality of life in portuguese girl adolescents: Association with psychological disturbance distress. <i>European Psychiatry</i> , 2021 , 64, S221-S221	6	
74	Postpartum depression screening scale-7: A valid and reliable short version both for portugal and brasil. <i>European Psychiatry</i> , 2021 , 64, S607-S607	6	
73	Eating disorder examination-questionnaire ¶7: Construct validity in a sample of portuguese overweight women. <i>European Psychiatry</i> , 2021 , 64, S360-S360	6	
72	The role of shame in the relationship between bullying and self-harm in portuguese adolescents. <i>European Psychiatry</i> , 2021 , 64, S221-S221	6	
71	The portuguese version of the big three perfectionism scale ¶further validation with adults from the general population. <i>European Psychiatry</i> , 2021 , 64, S445-S445	6	2
70	Early Gestational Diagnosis of Lethal Skeletal Dysplasias: A 15 Year Retrospective Cohort Reviewing Concordance between Ultrasonographic, Genetic and Morphological Features. <i>Fetal and Pediatric Pathology</i> , 2020 , 1-12	1.7	0
69	ESHRE PGT Consortium good practice recommendations for the organisation of PGT. <i>Human Reproduction Open</i> , 2020 , 2020, hoaa021	6.1	28
68	ESHRE PGT Consortium good practice recommendations for the detection of monogenic disorders. <i>Human Reproduction Open</i> , 2020 , 2020, hoaa018	6.1	20
67	Premature ovarian insufficiency: clinical orientations for genetic testing and genetic counseling. <i>Porto Biomedical Journal</i> , 2020 , 5, e62	1.1	7

66	Evaluation of Male Fertility-Associated Loci in a European Population of Patients with Severe Spermatogenic Impairment. <i>Journal of Personalized Medicine</i> , 2020 , 11,	3.6	2
65	Berry fruits modulate kidney dysfunction and urine metabolome in Dahl salt-sensitive rats. <i>Free Radical Biology and Medicine</i> , 2020 , 154, 119-131	7.8	5
64	ESHRE PGT Consortium data collection XVI-XVIII: cycles from 2013 to 2015. <i>Human Reproduction Open</i> , 2020 , 2020, hoaa043	6.1	13
63	Bi-allelic Mutations in M1AP Are a Frequent Cause of Meiotic Arrest and Severely Impaired Spermatogenesis Leading to Male Infertility. <i>American Journal of Human Genetics</i> , 2020 , 107, 342-351	11	19
62	Intronic variation of the SOHLH2 gene confers risk to male reproductive impairment. <i>Fertility and Sterility</i> , 2020 , 114, 398-406	4.8	3
61	Epimutations in human sperm from patients with impaired spermatogenesis. <i>Clinical Epigenetics</i> , 2020 , 12, 172	7.7	
60	Semen quality is affected by HLA class I alleles together with sexually transmitted diseases. <i>Andrology</i> , 2019 , 7, 867-877	4.2	3
59	Spectrum of CFTR gene sequence variants in a northern Portugal population. <i>Pulmonology</i> , 2018 , 24, 3-9	3.7	
58	Characterization of microbiota in male infertility cases uncovers differences in seminal hyperviscosity and oligoasthenoteratozoospermia possibly correlated with increased prevalence of infectious bacteria. <i>American Journal of Reproductive Immunology</i> , 2018 , 79, e12838	3.8	32
57	The clinical utility of PGD with HLA matching: a collaborative multi-centre ESHRE study. <i>Human Reproduction</i> , 2018 , 33, 520-530	5.7	15
56	Clinical outcomes after preimplantation genetic diagnosis of patients with Corino de Andrade disease (familial amyloid polyneuropathy). <i>Reproductive BioMedicine Online</i> , 2018 , 36, 39-46	4	5
55	Bi-allelic Recessive Loss-of-Function Variants in FANCM Cause Non-obstructive Azoospermia. <i>American Journal of Human Genetics</i> , 2018 , 103, 200-212	11	63
54	DNA methylation imprinting errors in spermatogenic cells from maturation arrest azoospermic patients. <i>Andrology</i> , 2017 , 5, 451-459	4.2	11
53	Sequence variation at KLK and WFDC clusters and its association to semen hyperviscosity and other male infertility phenotypes. <i>Human Reproduction</i> , 2016 , 31, 2881-2891	5.7	8
52	Collagen type IV-related nephropathies in Portugal: pathogenic COL4A3 and COL4A4 mutations and clinical characterization of 25 families. <i>Clinical Genetics</i> , 2015 , 88, 456-61	4	12
51	The mutational spectrum of WT1 in male infertility. <i>Journal of Urology</i> , 2015 , 193, 1709-15	2.5	10
50	Collagen type IV-related nephropathies in Portugal: pathogenic COL4A5 mutations and clinical characterization of 22 families. <i>Clinical Genetics</i> , 2015 , 88, 462-7	4	3
49	Rare double sex and mab-3-related transcription factor 1 regulatory variants in severe spermatogenic failure. <i>Andrology</i> , 2015 , 3, 825-33	4.2	11

48	Relevance of genomic imprinting in intrauterine human growth expression of CDKN1C, H19, IGF2, KCNQ1 and PHLDA2 imprinted genes. <i>Journal of Assisted Reproduction and Genetics</i> , 2014 , 31, 1361-8	3.4	38
47	A novel Alu-mediated microdeletion at 11p13 removes WT1 in a patient with cryptorchidism and azoospermia. <i>Reproductive BioMedicine Online</i> , 2014 , 29, 388-91	4	13
46	Evaluation of PCR-based preimplantation genetic diagnosis applied to monogenic diseases: a collaborative ESHRE PGD consortium study. <i>European Journal of Human Genetics</i> , 2014 , 22, 1012-8	5.3	51
45	Treatment by testicular sperm extraction and intracytoplasmic sperm injection of 65 azoospermic patients with non-mosaic Klinefelter syndrome with birth of 17 healthy children. <i>Andrology</i> , 2014 , 2, 623-31	4.3	52
44	Expression of stem cell markers: OCT4, KIT, ITGA6, and ITGB1 in the male germinal epithelium. <i>Systems Biology in Reproductive Medicine</i> , 2013 , 59, 233-43	2.9	15
43	Squamous upper tract carcinoma presenting as a perinephric abscess. <i>Case Reports in Urology</i> , 2013 , 2013, 789097	0.5	
42	Human spermatogenic failure purges deleterious mutation load from the autosomes and both sex chromosomes, including the gene DMRT1. <i>PLoS Genetics</i> , 2013 , 9, e1003349	6	99
41	Deletion of the 5' exons of COL4A6 is not needed for the development of diffuse leiomyomatosis in patients with Alport syndrome. <i>Journal of Medical Genetics</i> , 2013 , 50, 745-53	5.8	16
40	Comparative study of gene expression in patients with varicocele by microarray technology. <i>Andrologia</i> , 2012 , 44 Suppl 1, 260-5	2.4	9
39	Genetic regulation on ex vivo differentiated natural killer cells from human umbilical cord blood CD34+ cells. <i>Journal of Receptor and Signal Transduction Research</i> , 2012 , 32, 238-49	2.6	5
38	Quantitative analysis of cellular proliferation and differentiation of the human seminiferous epithelium in vitro. <i>Reproductive Sciences</i> , 2012 , 19, 1063-74	3	3
37	Comprehensive genetic analysis and structural characterization of CYP21A2 mutations in CAH patients. <i>Experimental and Clinical Endocrinology and Diabetes</i> , 2012 , 120, 535-9	2.3	9
36	Biomarkers Expression in Human Seminiferous Epithelium. <i>Microscopy and Microanalysis</i> , 2012 , 18, 15-16	0.5	
35	DNA methylation imprinting marks and DNA methyltransferase expression in human spermatogenic cell stages. <i>Epigenetics</i> , 2011 , 6, 1354-61	5.7	94
34	Concordance for bilateral congenital diaphragmatic hernia in a monozygotic dichorionic twin pair - first clinical report. <i>Fetal Diagnosis and Therapy</i> , 2010 , 27, 106-9	2.4	3
33	Mutational characterization of steroid 21-hydroxylase gene in Portuguese patients with congenital adrenal hyperplasia. <i>Experimental and Clinical Endocrinology and Diabetes</i> , 2010 , 118, 505-12	2.3	10
32	Aneuploidies detection in miscarriages and fetal deaths using multiplex ligation-dependent probe amplification: an alternative for speeding up results?. <i>European Journal of Obstetrics, Gynecology and Reproductive Biology</i> , 2010 , 153, 151-5	2.4	24
31	Methylation defects of imprinted genes in human testicular spermatozoa. <i>Fertility and Sterility</i> , 2010 , 94, 585-94	4.8	97

30	Association of cystic fibrosis genetic modifiers with congenital bilateral absence of the vas deferens. <i>Fertility and Sterility</i> , 2010 , 94, 2122-7	4.8	17
29	Gene expression pattern of IGF2, PHLDA2, PEG10 and CDKN1C imprinted genes in spontaneous miscarriages or fetal deaths. <i>Epigenetics</i> , 2010 , 5, 444-50	5.7	39
28	Application of touch FISH in the study of mosaic tetraploidy and maternal cell contamination in pregnancy losses. <i>Journal of Assisted Reproduction and Genetics</i> , 2010 , 27, 657-62	3.4	9
27	Phenotypic expression in the first case of complete trisomy 12: combination of prenatal ultrasound and necropsic examination. <i>Fetal Diagnosis and Therapy</i> , 2009 , 25, 234-8	2.4	3
26	Keratitis-ichthyosis-deafness syndrome caused by GJB2 maternal mosaicism. <i>Journal of Investigative Dermatology</i> , 2009 , 129, 776-9	4.3	19
25	An efficient protocol for the detection of chromosomal abnormalities in spontaneous miscarriages or foetal deaths. <i>European Journal of Obstetrics, Gynecology and Reproductive Biology</i> , 2009 , 147, 144-50	2.4	30
24	Abnormal methylation of imprinted genes in human sperm is associated with oligozoospermia. <i>Molecular Human Reproduction</i> , 2008 , 14, 67-74	4.4	283
23	Cytological and expression studies and quantitative analysis of the temporal and stage-specific effects of follicle-stimulating hormone and testosterone during cocultures of the normal human seminiferous epithelium. <i>Biology of Reproduction</i> , 2008 , 79, 962-75	3.9	21
22	Effect of single-nucleotide polymorphisms of the 5' untranslated region of the human alpha-galactosidase gene on enzyme activity, and their frequencies in Portuguese caucasians. <i>Journal of Inherited Metabolic Disease</i> , 2008 , 31 Suppl 2, S247-53	5.4	14
21	The g.1170C>T polymorphism of the 5' untranslated region of the human alpha-galactosidase gene is associated with decreased enzyme expression--evidence from a family study. <i>Journal of Inherited Metabolic Disease</i> , 2008 , 31 Suppl 2, S405-13	5.4	19
20	Molecular and functional characterization of CBAVD-causing mutations located in CFTR nucleotide-binding domains. <i>Cellular Physiology and Biochemistry</i> , 2008 , 22, 79-92	3.9	8
19	Sequence diversity at the proximal 14q32.1 SERPIN subcluster: evidence for natural selection favoring the pseudogenization of SERPINA2. <i>Molecular Biology and Evolution</i> , 2007 , 24, 587-98	8.3	17
18	Molecular characterization of the cystic fibrosis transmembrane conductance regulator gene in congenital absence of the vas deferens. <i>Genetics in Medicine</i> , 2007 , 9, 163-72	8.1	26
17	Characterization of missense mutations and large deletions in the ALPL gene by sequencing and quantitative multiplex PCR of short fragments. <i>Genetic Testing and Molecular Biomarkers</i> , 2006 , 10, 252-7		22
16	A novel missense mutation P1290S at exon-20 of the CFTR gene in a Portuguese patient with congenital bilateral absence of the vas deferens. <i>Fertility and Sterility</i> , 2005 , 83, 448-51	4.8	12
15	Estudo do imprinting genético em espermatozoides de pacientes com oligozoospermia. <i>Revista Internacional De Andrologia</i> , 2005 , 3, 101-108	0.6	
14	Thomsen-Friedenreich antigen expression in gastric carcinomas is associated with MUC1 mucin VNTR polymorphism. <i>Glycobiology</i> , 2005 , 15, 511-7	5.8	29
13	AZF and DAZ gene copy-specific deletion analysis in maturation arrest and Sertoli cell-only syndrome. <i>Molecular Human Reproduction</i> , 2004 , 10, 755-61	4.4	35

12	Characterization of cystic fibrosis conductance transmembrane regulator gene mutations and IVS8 poly(T) variants in Portuguese patients with congenital absence of the vas deferens. <i>Human Reproduction</i> , 2004 , 19, 2502-8	5.7	38
11	Genomic imprinting in disruptive spermatogenesis. <i>Lancet, The</i> , 2004 , 363, 1700-2	4.0	276
10	MUC1 polymorphism confers increased risk for intestinal metaplasia in a Colombian population with chronic gastritis. <i>European Journal of Human Genetics</i> , 2003 , 11, 380-4	5.3	20
9	Unique (Y;13) translocation in a male with oligozoospermia: cytogenetic and molecular studies. <i>European Journal of Human Genetics</i> , 2002 , 10, 467-74	5.3	52
8	Preimplantation genetic diagnosis for familial amyloidotic polyneuropathy (FAP). <i>Prenatal Diagnosis</i> , 2001 , 21, 1093-9	3.2	22
7	MUC1 gene polymorphism in the gastric carcinogenesis pathway. <i>European Journal of Human Genetics</i> , 2001 , 9, 548-52	5.3	54
6	Immunohistochemical study of the expression of MUC6 mucin and co-expression of other secreted mucins (MUC5AC and MUC2) in human gastric carcinomas. <i>Journal of Histochemistry and Cytochemistry</i> , 2000 , 48, 377-88	3.4	125
5	MUC1 gene polymorphism does not explain the different incidence of gastric cancer in Portugal and Denmark. <i>Annals of Human Genetics</i> , 1999 , 63, 187-91	2.2	10
4	Mucins and mucin-associated carbohydrate antigens expression in gastric carcinoma cell lines. <i>Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin</i> , 1999 , 435, 479-85	5.1	21
3	MUC1 gene polymorphism and gastric cancer--an epidemiological study. <i>Glycoconjugate Journal</i> , 1997 , 14, 107-11	3	77
2	Allele loss in human gastric carcinomas - relation to tumor progression and differentiation. <i>International Journal of Oncology</i> , 1995 , 7, 1159-66	1	1
1	Biallelic mutations in M1AP are a frequent cause of meiotic arrest leading to male infertility		1