Filipa Carvalho

List of Publications by Citations

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	Abnormal methylation of imprinted genes in human sperm is associated with oligozoospermia. *Molecular Human Reproduction*, **2008**, 14, 67-74	4.4	283
82	Genomic imprinting in disruptive spermatogenesis. *Lancet, The*, **2004**, 363, 1700-2	40	276
81	Immunohistochemical study of the expression of MUC6 mucin and co-expression of other secreted mucins (MUC5AC and MUC2) in human gastric carcinomas. *Journal of Histochemistry and Cytochemistry*, **2000**, 48, 377-88	3.4	125
80	Human spermatogenic failure purges deleterious mutation load from the autosomes and both sex chromosomes, including the gene DMRT1. *PLoS Genetics*, **2013**, 9, e1003349	6	99
79	Methylation defects of imprinted genes in human testicular spermatozoa. *Fertility and Sterility*, **2010**, 94, 585-94	4.8	97
78	DNA methylation imprinting marks and DNA methyltransferase expression in human spermatogenic cell stages. *Epigenetics*, **2011**, 6, 1354-61	5.7	94
77	MUC1 gene polymorphism and gastric cancer--an epidemiological study. *Glycoconjugate Journal*, **1997**, 14, 107-11	3	77
76	Bi-allelic Recessive Loss-of-Function Variants in FANCM Cause Non-obstructive Azoospermia. *American Journal of Human Genetics*, **2018**, 103, 200-212	11	63
75	MUC1 gene polymorphism in the gastric carcinogenesis pathway. *European Journal of Human Genetics*, **2001**, 9, 548-52	5.3	54
74	Treatment by testicular sperm extraction and intracytoplasmic sperm injection of 65 azoospermic patients with non-mosaic Klinefelter syndrome with birth of 17 healthy children. *Andrology*, **2014**, 2, 623-31	4.2	52
73	Unique (Y;13) translocation in a male with oligozoospermia: cytogenetic and molecular studies. *European Journal of Human Genetics*, **2002**, 10, 467-74	5.3	52
72	Evaluation of PCR-based preimplantation genetic diagnosis applied to monogenic diseases: a collaborative ESHRE PGD consortium study. *European Journal of Human Genetics*, **2014**, 22, 1012-8	5.3	51
71	Gene expression pattern of IGF2, PHLDA2, PEG10 and CDKN1C imprinted genes in spontaneous miscarriages or fetal deaths. *Epigenetics*, **2010**, 5, 444-50	5.7	39
70	Relevance of genomic imprinting in intrauterine human growth expression of CDKN1C, H19, IGF2, KCNQ1 and PHLDA2 imprinted genes. *Journal of Assisted Reproduction and Genetics*, **2014**, 31, 1361-8	3.4	38
69	Characterization of cystic fibrosis conductance transmembrane regulator gene mutations and IVS8 poly(T) variants in Portuguese patients with congenital absence of the vas deferens. *Human Reproduction*, **2004**, 19, 2502-8	5.7	38
68	AZF and DAZ gene copy-specific deletion analysis in maturation arrest and Sertoli cell-only syndrome. *Molecular Human Reproduction*, **2004**, 10, 755-61	4.4	35
67	Characterization of microbiota in male infertility cases uncovers differences in seminal hyperviscosity and oligoasthenoteratozoospermia possibly correlated with increased prevalence of infectious bacteria. *American Journal of Reproductive Immunology*, **2018**, 79, e12838	3.8	32

66	An efficient protocol for the detection of chromosomal abnormalities in spontaneous miscarriages or foetal deaths. *European Journal of Obstetrics, Gynecology and Reproductive Biology*, **2009**, 147, 144-50	2.4	30
65	Thomsen-Friedenreich antigen expression in gastric carcinomas is associated with MUC1 mucin VNTR polymorphism. *Glycobiology*, **2005**, 15, 511-7	5.8	29
64	ESHRE PGT Consortium good practice recommendations for the organisation of PGT. *Human Reproduction Open*, **2020**, 2020, hoaa021	6.1	28
63	Molecular characterization of the cystic fibrosis transmembrane conductance regulator gene in congenital absence of the vas deferens. *Genetics in Medicine*, **2007**, 9, 163-72	8.1	26
62	Aneuploidies detection in miscarriages and fetal deaths using multiplex ligation-dependent probe amplification: an alternative for speeding up results?. *European Journal of Obstetrics, Gynecology and Reproductive Biology*, **2010**, 153, 151-5	2.4	24
61	Characterization of missense mutations and large deletions in the ALPL gene by sequencing and quantitative multiplex PCR of short fragments. *Genetic Testing and Molecular Biomarkers*, **2006**, 10, 252-7		22
60	Preimplantation genetic diagnosis for familial amyloidotic polyneuropathy (FAP). *Prenatal Diagnosis* , **2001**, 21, 1093-9	3.2	22
59	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. *Biology of Reproduction*, **2008**, 79, 962-75	3.9	21
58	Mucins and mucin-associated carbohydrate antigens expression in gastric carcinoma cell lines. *Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin*, **1999**, 435, 479-85	5.1	21
57	ESHRE PGT Consortium good practice recommendations for the detection of monogenic disorders. *Human Reproduction Open*, **2020**, 2020, hoaa018	6.1	20
56	MUC1 polymorphism confers increased risk for intestinal metaplasia in a Colombian population with chronic gastritis. *European Journal of Human Genetics*, **2003**, 11, 380-4	5.3	20
55	Keratitis-ichthyosis-deafness syndrome caused by GJB2 maternal mosaicism. *Journal of Investigative Dermatology*, **2009**, 129, 776-9	4.3	19
54	The g.1170C>T polymorphism of the 5Quntranslated region of the human alpha-galactosidase gene is associated with decreased enzyme expression--evidence from a family study. *Journal of Inherited Metabolic Disease*, **2008**, 31 Suppl 2, S405-13	5.4	19
53	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	11	19
52	Association of cystic fibrosis genetic modifiers with congenital bilateral absence of the vas deferens. *Fertility and Sterility*, **2010**, 94, 2122-7	4.8	17
51	Sequence diversity at the proximal 14q32.1 SERPIN subcluster: evidence for natural selection favoring the pseudogenization of SERPINA2. *Molecular Biology and Evolution*, **2007**, 24, 587-98	8.3	17
50	Deletion of the 5@xons of COL4A6 is not needed for the development of diffuse leiomyomatosis in patients with Alport syndrome. *Journal of Medical Genetics*, **2013**, 50, 745-53	5.8	16
49	The clinical utility of PGD with HLA matching: a collaborative multi-centre ESHRE study. *Human Reproduction*, **2018**, 33, 520-530	5.7	15

48	Expression of stem cell markers: OCT4, KIT, ITGA6, and ITGB1 in the male germinal epithelium. *Systems Biology in Reproductive Medicine*, **2013**, 59, 233-43	2.9	15
47	Effect of single-nucleotide polymorphisms of the 5Quntranslated region of the human galactosidase gene on enzyme activity, and their frequencies in Portuguese caucasians. *Journal of Inherited Metabolic Disease*, **2008**, 31 Suppl 2, S247-53	5.4	14
46	A novel Alu-mediated microdeletion at 11p13 removes WT1 in a patient with cryptorchidism and azoospermia. *Reproductive BioMedicine Online*, **2014**, 29, 388-91	4	13
45	ESHRE PGT Consortium data collection XVI-XVIII: cycles from 2013 to 2015. *Human Reproduction Open*, **2020**, 2020, hoaa043	6.1	13
44	Collagen type IV-related nephropathies in Portugal: pathogenic COL4A3 and COL4A4 mutations and clinical characterization of 25 families. *Clinical Genetics*, **2015**, 88, 456-61	4	12
43	A novel missense mutation P1290S at exon-20 of the CFTR gene in a Portuguese patient with congenital bilateral absence of the vas deferens. *Fertility and Sterility*, **2005**, 83, 448-51	4.8	12
42	DNA methylation imprinting errors in spermatogenic cells from maturation arrest azoospermic patients. *Andrology*, **2017**, 5, 451-459	4.2	11
41	Rare double sex and mab-3-related transcription factor 1 regulatory variants in severe spermatogenic failure. *Andrology*, **2015**, 3, 825-33	4.2	11
40	The mutational spectrum of WT1 in male infertility. *Journal of Urology*, **2015**, 193, 1709-15	2.5	10
39	Mutational characterization of steroid 21-hydroxylase gene in Portuguese patients with congenital adrenal hyperplasia. *Experimental and Clinical Endocrinology and Diabetes*, **2010**, 118, 505-12	2.3	10
38	MUC1 gene polymorphism does not explain the different incidence of gastric cancer in Portugal and Denmark. *Annals of Human Genetics*, **1999**, 63, 187-91	2.2	10
37	Comparative study of gene expression in patients with varicocele by microarray technology. *Andrologia*, **2012**, 44 Suppl 1, 260-5	2.4	9
36	Comprehensive genetic analysis and structural characterization of CYP21A2 mutations in CAH patients. *Experimental and Clinical Endocrinology and Diabetes*, **2012**, 120, 535-9	2.3	9
35	Application of touch FISH in the study of mosaic tetraploidy and maternal cell contamination in pregnancy losses. *Journal of Assisted Reproduction and Genetics*, **2010**, 27, 657-62	3.4	9
34	Sequence variation at KLK and WFDC clusters and its association to semen hyperviscosity and other male infertility phenotypes. *Human Reproduction*, **2016**, 31, 2881-2891	5.7	8
33	Molecular and functional characterization of CBAVD-causing mutations located in CFTR nucleotide-binding domains. *Cellular Physiology and Biochemistry*, **2008**, 22, 79-92	3.9	8
32	ESHRE PGT Consortium data collection XIX-XX: PGT analyses from 2016 to 2017. *Human Reproduction Open*, **2021**, 2021, hoab024	6.1	8
31	Variant , Defective piRNA Processing, and Azoospermia. *New England Journal of Medicine*, **2021**, 385, 707-719	59.2	8

30	Premature ovarian insufficiency: clinical orientations for genetic testing and genetic counseling. *Porto Biomedical Journal*, **2020**, 5, e62	1.1	7
29	Clinical outcomes after preimplantation genetic diagnosis of patients with Corino de Andrade disease (familial amyloid polyneuropathy). *Reproductive BioMedicine Online*, **2018**, 36, 39-46	4	5
28	Genetic regulation on ex vivo differentiated natural killer cells from human umbilical cord blood CD34+ cells. *Journal of Receptor and Signal Transduction Research*, **2012**, 32, 238-49	2.6	5
27	Berry fruits modulate kidney dysfunction and urine metabolome in Dahl salt-sensitive rats. *Free Radical Biology and Medicine*, **2020**, 154, 119-131	7.8	5
26	Congenital Adrenal Hyperplasia Due to 21-Hydroxylase Deficiency: An Update on Genetic Analysis of CYP21A2 Gene. *Experimental and Clinical Endocrinology and Diabetes*, **2021**, 129, 477-481	2.3	4
25	Semen quality is affected by HLA class I alleles together with sexually transmitted diseases. *Andrology*, **2019**, 7, 867-877	4.2	3
24	Collagen type IV-related nephropathies in Portugal: pathogenic COL4A5 mutations and clinical characterization of 22 families. *Clinical Genetics*, **2015**, 88, 462-7	4	3
23	Concordance for bilateral congenital diaphragmatic hernia in a monozygotic dichorionic twin pair - first clinical report. *Fetal Diagnosis and Therapy*, **2010**, 27, 106-9	2.4	3
22	Phenotypic expression in the first case of complete trisomy 12: combination of prenatal ultrasound and necropsic examination. *Fetal Diagnosis and Therapy*, **2009**, 25, 234-8	2.4	3
21	Quantitative analysis of cellular proliferation and differentiation of the human seminiferous epithelium in vitro. *Reproductive Sciences*, **2012**, 19, 1063-74	3	3
20	Intronic variation of the SOHLH2 gene confers risk to male reproductive impairment. *Fertility and Sterility*, **2020**, 114, 398-406	4.8	3
19	Variants in GCNA, X-linked germ-cell genome integrity gene, identified in men with primary spermatogenic failure. *Human Genetics*, **2021**, 140, 1169-1182	6.3	3
18	A de novo paradigm for male infertility.. *Nature Communications*, **2022**, 13, 154	17.4	2
17	Evaluation of Male Fertility-Associated Loci in a European Population of Patients with Severe Spermatogenic Impairment. *Journal of Personalized Medicine*, **2020**, 11,	3.6	2
16	Effect and in silico characterization of genetic variants associated with severe spermatogenic disorders in a large Iberian cohort. *Andrology*, **2021**, 9, 1151-1165	4.2	2
15	The portuguese version of the big three perfectionism scale Further validation with adults from the general population. *European Psychiatry*, **2021**, 64, S445-S445	6	2
14	Allele loss in human gastric carcinomas - relation to tumor progression and differentiation. *International Journal of Oncology*, **1995**, 7, 1159-66	1	1
13	Biallelic mutations in M1AP are a frequent cause of meiotic arrest leading to male infertility		1

12	Early Gestational Diagnosis of Lethal Skeletal Dysplasias: A 15 Year Retrospective Cohort Reviewing Concordance between Ultrasonographic, Genetic and Morphological Features. *Fetal and Pediatric Pathology*, **2020**, 1-12	1.7	0
11	Spectrum of CFTR gene sequence variants in a northern Portugal population. *Pulmonology*, **2018**, 24, 3-9	3.7	
10	Squamous upper tract carcinoma presenting as a perinephric abscess. *Case Reports in Urology*, **2013** , 2013, 789097	0.5	
9	Biomarkers Expression in Human Seminiferous Epithelium. *Microscopy and Microanalysis*, **2012**, 18, 15-16	0.5	
8	Estudo do imprinting gen□ico em espermatoz□des de pacientes com oligozoospermia. *Revista Internacional De Androloga*, **2005**, 3, 101-108	0.6	
7	Epimutations in human sperm from patients with impaired spermatogenesis. *Clinical Epigenetics*, **2020**, 12, 172	7.7	
6	Mindfulness and self-compassion based intervention program to prevent burnout in medical and dentistry students. *European Psychiatry*, **2021**, 64, S459-S460	6	
5	Habits and quality of life in portuguese girl adolescents: Association with psychological disturbance distress. *European Psychiatry*, **2021**, 64, S221-S221	6	
4	Postpartum depression screening scale-7: A valid and reliable short version both for portugal and brasil. *European Psychiatry*, **2021**, 64, S607-S607	6	
3	Eating disorder examination-questionnaire □7: Construct validity in a sample of portuguese overweight women. *European Psychiatry*, **2021**, 64, S360-S360	6	
2	The role of shame in the relationship between bullying and self-harm in portuguese adolescents. *European Psychiatry*, **2021**, 64, S221-S221	6	
1	Common Variation in the PIN1 Locus Increases the Genetic Risk to Suffer from Sertoli Cell-Only Syndrome. *Journal of Personalized Medicine*, **2022**, 12, 932	3.6	