Susana Fernandes

List of Publications by Year in descending order

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51 papers	1,840 citations	377584 21 h-index	299063 42 g-index
53 all docs	53 docs citations	53 times ranked	2353 citing authors

#	Article	IF	Citations
1	A de novo paradigm for male infertility. Nature Communications, 2022, 13, 154.	5.8	38
2	Whole-Exome Sequencing Targeting a Gene Panel for Sensorineural Hearing Loss: The First Portuguese Cohort Study. Cytogenetic and Genome Research, 2022, 162, 1-9.	0.6	0
3	Acute respiratory distress due to a bronchogenic cyst submitted to percutaneous drainage followed by thoracoscopic resection. Pulmonology, 2021, 27, 371-373.	1.0	1
4	GJB2: Frequency of the Less Common Variants in a Sample of the Portuguese Population. Acta Medica Portuguesa, 2021, 34, 592.	0.2	0
5	The CDH1 c.1901C>T Variant: A Founder Variant in the Portuguese Population with Severe Impact in mRNA Splicing. Cancers, 2021, 13, 4464.	1.7	7
6	Clinical and molecular characterization of Y microdeletions and Xâ€linked CNV 67 implications in male fertility: a 20â€year experience. Andrology, 2020, 8, 307-314.	1.9	10
7	Acromegaly with congenital generalized lipodystrophy â€“ two rare insulin resistance conditions in one patient: a case report. Journal of Medical Case Reports, 2020, 14, 34.	0.4	2
8	Highly Sensitive Blocker Displacement Amplification and Droplet Digital PCR Reveal Low-Level Parental FOXF1 Somatic Mosaicism in Families with Alveolar Capillary Dysplasia with Misalignment of Pulmonary Veins. Journal of Molecular Diagnostics, 2020, 22, 447-456.	1.2	13
9	<scp>DNA</scp> methylation imprinting errors in spermatogenic cells from maturation arrest azoospermic patients. Andrology, 2017, 5, 451-459.	1.9	15
10	Novel Missense LCAT Gene Mutation Associated with an Atypical Phenotype of Familial LCAT Deficiency in Two Portuguese Brothers. JIMD Reports, 2017, 40, 55-62.	0.7	9
11	Y-chromosome microdeletions in nonobstructive azoospermia and severe oligozoospermia. Asian Journal of Andrology, 2017, 19, 338.	0.8	39
12	Study of Met34Thr variant in nonsyndromic hearing loss in four Portuguese families. Porto Biomedical Journal, 2016, 1, 32-35.	0.4	1
13	MEF2C haploinsufficiency syndrome: Report of a new MEF2C mutation and review. European Journal of Medical Genetics, 2016, 59, 478-482.	0.7	53
14	Thoracoscopy in the management of pediatric empyemas. Revista Portuguesa De Pneumologia, 2016, 22, 157-162.	0.7	4
15	Increased expression of Î±7nAChR in chronic rhinosinusitis: The intranasal cholinergic anti-inflammatory hypothesis. Auris Nasus Larynx, 2016, 43, 176-181.	0.5	4
16	Filaggrin Gene Polymorphism Pro478Ser, but Not Loss-of-Function Mutations Mp.Arg501Ter or C.2282del4, Relates with Atopic Dermatitis Severity and Increased Staphylococcal aureus Colonization in Adult Patients. Journal of Allergy and Clinical Immunology, 2015, 135, AB260.	1.5	0
17	Rare double sex and mab-3-related transcription factor 1 regulatory variants in severe spermatogenic failure. Andrology, 2015, 3, 825-833.	1.9	17
18	Familial partial lipodystrophy type 3: a new mutation on the PPARG gene. Hormones, 2015, 14, 317-320.	0.9	5

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19	X-linked agammaglobulinemia: Experience in a Portuguese hospital. Anales De PediatrÃa (English) Tj ETQq1 1 0.784314 rgBT /Overloc	0.1	2
20	Prevalence of 35delG and Met34Thr GJB2 variants in Portuguese samples. International Journal of Pediatric Otorhinolaryngology, 2015, 79, 2187-2190.	0.4	3
21	Bilateral Frontoparietal Polymicrogyria: A Novel GPR56 Mutation and an Unusual Phenotype. Neuropediatrics, 2015, 46, 134-138.	0.3	15
22	The Mutational Spectrum of WT1 in Male Infertility. Journal of Urology, 2015, 193, 1709-1715.	0.2	11
23	Alsin Related Disorders: Literature Review and Case Study with Novel Mutations. Case Reports in Genetics, 2014, 2014, 1-5.	0.1	11
24	A novel Alu-mediated microdeletion at 11p13 removes WT1 in a patient with cryptorchidism and azoospermia. Reproductive BioMedicine Online, 2014, 29, 388-391.	1.1	18
25	Nonoptical Massive Parallel DNA Sequencing ofBRCA1andBRCA2Genes in a Diagnostic Setting. Human Mutation, 2013, 34, 629-635.	1.1	37
26	Human Spermatogenic Failure Purges Deleterious Mutation Load from the Autosomes and Both Sex Chromosomes, including the Gene DMRT1. PLoS Genetics, 2013, 9, e1003349.	1.5	118
27	A Novel Mutation in <i>FOXF1</i> Gene Associated with Alveolar Capillary Dysplasia with Misalignment of Pulmonary Veins, Intestinal Malrotation and Annular Pancreas. Neonatology, 2013, 103, 241-245.	0.9	13
28	NovelFOXF1Mutations in Sporadic and Familial Cases of Alveolar Capillary Dysplasia with Misaligned Pulmonary Veins Imply a Role for its DNA Binding Domain. Human Mutation, 2013, 34, 801-811.	1.1	97
29	Y-Chromosome Detection in Turner Syndrome. , 2013, 03, .		2
30	Expression Analysis of MLH3, MLH1, and MSH4 in Maturation Arrest. Reproductive Sciences, 2012, 19, 587-596.	1.1	5
31	A novel splicing mutation causes analbuminemia in a Portuguese boy. Molecular Genetics and Metabolism, 2012, 105, 479-483.	0.5	13
32	AZFb microdeletions and oligozoospermiaâ€"which mechanisms?. Fertility and Sterility, 2012, 97, 858-863.	0.5	50
33	Mutational Characterization of Steroid 21-Hydroxylase Gene in Portuguese patients with Congenital Adrenal Hyperplasia. Experimental and Clinical Endocrinology and Diabetes, 2010, 118, 505-512.	0.6	12
34	Gene expression pattern ofIGF2,PHLDA2,PEG10andCDKN1Cimprinted genes in spontaneous miscarriages or fetal deaths. Epigenetics, 2010, 5, 444-450.	1.3	51
35	Abnormal methylation of imprinted genes in human sperm is associated with oligozoospermia. Molecular Human Reproduction, 2008, 14, 67-74.	1.3	330
36	Identification of new breakpoints in AZFb and AZFc. Molecular Human Reproduction, 2008, 14, 251-258.	1.3	39

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37	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 Epithelium1. Biology of Reproduction, 2008, 79, 962-975.	1.2	25
38	DAZ gene copies: evidence of Y chromosome evolution. Molecular Human Reproduction, 2006, 12, 519-523.	1.3	23
39	Unique t(Y;1)(q12;q12) reciprocal translocation with loss of the heterochromatic region of chromosome 1 in a male with azoospermia due to meiotic arrest: a case report. Human Reproduction, 2005, 20, 689-696.	0.4	44
40	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, 448-451.	0.5	14
41	AZF and DAZ gene copy-specific deletion analysis in maturation arrest and Sertoli cell-only syndrome. Molecular Human Reproduction, 2004, 10, 755-761.	1.3	39
42	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, 2502-2508.	0.4	45
43	A Large AZFc Deletion Removes DAZ3/DAZ4 and Nearby Genes from Men in Y Haplogroup N. American Journal of Human Genetics, 2004, 74, 180-187.	2.6	176
44	Are Sequence Family Variants Useful for Identifying Deletions in the Human Y Chromosome?. American Journal of Human Genetics, 2004, 75, 514-517.	2.6	24
45	Reply to Repping et al.. American Journal of Human Genetics, 2004, 75, 517-518.	2.6	8
46	Polymorphic DAZ gene family in polymorphic structure of AZFc locus: Artwork or functional for human spermatogenesis?. Review article. Apmis, 2003, 111, 115-127.	0.9	48
47	Frequent DAZ1/DAZ2 deletions in men with severe oligozoospermia. Andrologia, 2003, 35, 5-6.	1.0	3
48	High frequency of DAZ1/DAZ2 gene deletions in patients with severe oligozoospermia. Molecular Human Reproduction, 2002, 8, 286-298.	1.3	153
49	Preimplantation genetic diagnosis for familial amyloidotic polyneuropathy (FAP). Prenatal Diagnosis, 2001, 21, 1093-1099.	1.1	28
50	High deletion frequency of the complete AZFa sequence in men with Sertoli-cell-only syndrome. Molecular Human Reproduction, 2001, 7, 987-994.	1.3	148
51	Prognostic factors for successful testicle spermatid recover. Molecular and Cellular Endocrinology, 2000, 166, 37-43.	1.6	13