Ana Peixoto

List of Publications by Year
in descending order

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58 papers	3,130 citations	257450 24 h-index	175258 52 g-index
61 all docs	61 docs citations	61 times ranked	6040 citing authors

#	Article	IF	Citations
1	Association of Type and Location ofBRCA1andBRCA2Mutations With Risk of Breast and Ovarian Cancer. JAMA - Journal of the American Medical Association, 2015, 313, 1347.	7.4	390
2	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. Nature Genetics, 2017, 49, 680-691.	21.4	356
3	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. Nature Genetics, 2017, 49, 1767-1778.	21.4	289
4	Genome-wide association study identifies 32 novel breast cancer susceptibility loci from overall and subtype-specific analyses. Nature Genetics, 2020, 52, 572-581.	21.4	265
5	TMPRSS2-ERG Gene Fusion Causing ERG Overexpression Precedes Chromosome Copy Number Changes in Prostate Carcinomas, Paired HGPIN Lesions. Neoplasia, 2006, 8, 826-832.	5.3	225
6	Mutational spectrum in a worldwide study of 29,700 families withBRCA1orBRCA2mutations. Human Mutation, 2018, 39, 593-620.	2.5	224
7	Genome-Wide Meta-Analyses of Breast, Ovarian, and Prostate Cancer Association Studies Identify Multiple New Susceptibility Loci Shared by at Least Two Cancer Types. Cancer Discovery, 2016, 6, 1052-1067.	9.4	157
8	Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. Nature Communications, 2019, 10, 1741.	12.8	90
9	Polygenic risk scores and breast and epithelial ovarian cancer risks for carriers of BRCA1 and BRCA2 pathogenic variants. Genetics in Medicine, 2020, 22, 1653-1666.	2.4	82
10	BRCA2 Polymorphic Stop Codon K3326X and the Risk of Breast, Prostate, and Ovarian Cancers. Journal of the National Cancer Institute, 2016, 108, djv315.	6.3	77
11	The c.156_157insAlu BRCA2 rearrangement accounts for more than one-fourth of deleterious BRCA mutations in northern/central Portugal. Breast Cancer Research and Treatment, 2009, 114, 31-38.	2.5	52
12	Targeted next generation sequencing identifies functionally deleterious germline mutations in novel genes in early-onset/familial prostate cancer. PLoS Genetics, 2018, 14, e1007355.	3.5	50
13	High resolution melting analysis of KRAS, BRAF and PIK3CA in KRASexon 2 wild-type metastatic colorectal cancer. BMC Cancer, 2013, 13, 169.	2.6	44
14	<scp>POLE</scp> somatic mutations in advanced colorectal cancer. Cancer Medicine, 2017, 6, 2966-2971.	2.8	43
15	Feasibility of differential diagnosis of kidney tumors by comparative genomic hybridization of fine needle aspiration biopsies. Genes Chromosomes and Cancer, 2010, 49, 935-947.	2.8	41
16	Association of Genomic Domains in BRCA1 and BRCA2 with Prostate Cancer Risk and Aggressiveness. Cancer Research, 2020, 80, 624-638.	0.9	39
17	Comparison of methodologies for KRAS mutation detection in metastatic colorectal cancer. Cancer Genetics, 2011, 204, 439-446.	0.4	37
18	Identification of Two Novel HOXB13 Germline Mutations in Portuguese Prostate Cancer Patients. PLoS ONE, 2015, 10, e0132728.	2.5	34

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19	Transcriptomeâ€wide association study of breast cancer risk by estrogenâ€receptor status. Genetic Epidemiology, 2020, 44, 442-468.	1.3	32
20	BRCA1 and BRCA2 germline mutational spectrum and evidence for genetic anticipation in Portuguese breast/ovarian cancer families. Familial Cancer, 2006, 5, 379-387.	1.9	30
21	EGFR exon mutation distribution and outcome in non-small-cell lung cancer: a Portuguese retrospective study. Tumor Biology, 2012, 33, 2061-2068.	1.8	30
22	Target gene mutational pattern in Lynch syndrome colorectal carcinomas according to tumour location and germline mutation. British Journal of Cancer, 2015, 113, 686-692.	6.4	30
23	Contribution of <scp>MLH</scp>1 constitutional methylation for Lynch syndrome diagnosis in patients with tumor <scp>MLH</scp>1 downregulation. Cancer Medicine, 2018, 7, 433-444.	2.8	28
24	The FANCM:p.Arg658* truncating variant is associated with risk of triple-negative breast cancer. Npj Breast Cancer, 2019, 5, 38.	5.2	28
25	International distribution and age estimation of the Portuguese BRCA2 c.156_157insAlu founder mutation. Breast Cancer Research and Treatment, 2011, 127, 671-679.	2.5	27
26	The role of germline mutations in the BRCA1/2 and mismatch repair genes in men ascertained for early-onset and/or familial prostate cancer. Familial Cancer, 2016, 15, 111-121.	1.9	26
27	Li-Fraumeni-like syndrome associated with a large BRCA1 intragenic deletion. BMC Cancer, 2012, 12, 237.	2.6	25
28	Genomic characterization of two large Alu-mediated rearrangements of the BRCA1 gene. Journal of Human Genetics, 2013, 58, 78-83.	2.3	24
29	Pathogenicity Evaluation of BRCA1 and BRCA2 Unclassified Variants Identified in Portuguese Breast/Ovarian Cancer Families. Journal of Molecular Diagnostics, 2014, 16, 324-334.	2.8	24
30	Full in-frame exon 3 skipping of BRCA2 confers high risk of breast and/or ovarian cancer. Oncotarget, 2018, 9, 17334-17348.	1.8	24
31	CSF1R copy number changes, point mutations, and RNA and protein overexpression in renal cell carcinomas. Modern Pathology, 2009, 22, 744-752.	5.5	23
32	Implementation of next-generation sequencing for molecular diagnosis of hereditary breast and ovarian cancer highlights its genetic heterogeneity. Breast Cancer Research and Treatment, 2016, 159, 245-256.	2.5	23
33	Polygenic risk modeling for prediction of epithelial ovarian cancer risk. European Journal of Human Genetics, 2022, 30, 349-362.	2.8	23
34	BRCA1 and BRCA2 rearrangements in Brazilian individuals with Hereditary Breast and Ovarian Cancer Syndrome. Genetics and Molecular Biology, 2016, 39, 223-231.	1.3	22
35	A novel exonic rearrangement affecting MLH1 and the contiguous LRRFIP2 is a founder mutation in Portuguese Lynch syndrome families. Genetics in Medicine, 2011, 13, 895-902.	2.4	21
36	Potential clinical applications of circulating cell-free DNA in ovarian cancer patients. Expert Reviews in Molecular Medicine, 2018, 20, e6.	3.9	20

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37	The CHEK2 Variant C.349A>G Is Associated with Prostate Cancer Risk and Carriers Share a Common Ancestor. Cancers, 2020, 12, 3254.	3.7	16
38	Analysis of Founder Mutations in Rare Tumors Associated With Hereditary Breast/Ovarian Cancer Reveals a Novel Association of BRCA2 Mutations with Ampulla of Vater Carcinomas. PLoS ONE, 2016, 11, e0161438.	2.5	15
39	TP53 germline mutations in Portugal and genetic modifiers of age at cancer onset. Familial Cancer, 2009, 8, 383-390.	1.9	14
40	Intraepidermal epidermotropic metastatic melanoma: a clinical and histopathological mimicker of melanoma in situ occurring in multiplicity. Journal of Cutaneous Pathology, 2011, 38, 514-520.	1.3	14
41	Validation of a Next-Generation Sequencing Pipeline for the Molecular Diagnosis of Multiple Inherited Cancer Predisposing Syndromes. Journal of Molecular Diagnostics, 2017, 19, 502-513.	2.8	13
42	Haplotype and quantitative transcript analyses of Portuguese breast/ovarian cancer families with the BRCA1 R71G founder mutation of Galician origin. Familial Cancer, 2009, 8, 203-208.	1.9	11
43	Tumor Testing for Somatic and Germline BRCA1/BRCA2 Variants in Ovarian Cancer Patients in the Context of Strong Founder Effects. Frontiers in Oncology, 2020, 10, 1318.	2.8	11
44	The Spectrum of FANCM Protein Truncating Variants in European Breast Cancer Cases. Cancers, 2020, 12, 292.	3.7	11
45	Portuguese c.156_157insAlu BRCA2 founder mutation: gastrointestinal and tongue neoplasias may be part of the phenotype. Familial Cancer, 2012, 11, 657-660.	1.9	10
46	Molecular diagnosis of the Portuguese founder mutation BRCA2 c.156_157insAlu. Breast Cancer Research and Treatment, 2009, 117, 215-217.	2.5	8
47	Co-occurrence of nonsense mutations in MSH6 and MSH2 in Lynch syndrome families evidencing that not all truncating mutations are equal. Journal of Human Genetics, 2016, 61, 151-156.	2.3	8
48	The Brazilian Founder Mutation<i>TP53</i>p.R337H is Uncommon in Portuguese Women Diagnosed with Breast Cancer. Breast Journal, 2014, 20, 534-536.	1.0	6
49	Screening and characterization of BRCA2 c.156_157insAlu in Brazil: Results from 1380 individuals from the South and Southeast. Cancer Genetics, 2018, 228-229, 93-97.	0.4	6
50	Gene Panel Tumor Testing in Ovarian Cancer Patients Significantly Increases the Yield of Clinically Actionable Germline Variants beyond BRCA1/BRCA2. Cancers, 2020, 12, 2834.	3.7	6
51	Next Generation Sequencing of Tumor and Matched Plasma Samples: Identification of Somatic Variants in ctDNA From Ovarian Cancer Patients. Frontiers in Oncology, 2021, 11, 754094.	2.8	5
52	The nonsense mutation <i>MSH2</i> c.2152C>T shows a founder effect in Portuguese Lynch syndrome families. Genes Chromosomes and Cancer, 2019, 58, 657-664.	2.8	3
53	Expression Profiling in Ovarian Cancer Reveals Coordinated Regulation of BRCA1/2 and Homologous Recombination Genes. Biomedicines, 2022, 10, 199.	3.2	3
54	KRAS and NRAS mutational analysis in plasma ctDNA from patients with metastatic colorectal cancer by real-time PCR and digital PCR. International Journal of Colorectal Disease, 2022, 37, 895-905.	2.2	3

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55	Molecular characterization of a rare MLLâ€“AF4 (MLLâ€“AFF1) fusion rearrangement in infant leukemia. Cancer Genetics and Cytogenetics, 2007, 178, 61-64.	1.0	2
56	Performance of Lynch syndrome predictive models in quantifying the likelihood of germline mutations in patients with abnormal MLH1 immunoexpression. Familial Cancer, 2017, 16, 73-81.	1.9	2
57	The role of TP53 pathogenic variants in early-onset HER2-positive breast cancer. Familial Cancer, 2021, 20, 173-180.	1.9	2
58	Pathogenicity reclassification of two BRCA1/BRCA2 exonic duplications after identification of genomic breakpoints and tandem orientation. Cancer Genetics, 2020, 248-249, 18-24.	0.4	0