Marta Cardoso

List of Publications by Year
in descending order

96 papers	5,995 citations	87723 38 h-index	85405 71 g-index
101 all docs	101 docs citations	101 times ranked	11297 citing authors

#	Article	IF	Citations
1	Breast and Prostate Cancer Risks for MaleBRCA1andBRCA2Pathogenic Variant Carriers Using Polygenic Risk Scores. Journal of the National Cancer Institute, 2022, 114, 109-122.	3.0	19
2	Polygenic risk modeling for prediction of epithelial ovarian cancer risk. European Journal of Human Genetics, 2022, 30, 349-362.	1.4	23
3	Multi-Gene Panel Testing in Gastroenterology: Are We Ready for the Results?. GE Portuguese Journal of Gastroenterology, 2021, 28, 1-7.	0.3	2
4	The predictive ability of the 313 variantâ€“based polygenic risk score for contralateral breast cancer risk prediction in women of European ancestry with a heterozygous BRCA1 or BRCA2 pathogenic variant. Genetics in Medicine, 2021, 23, 1726-1737.	1.1	16
5	Association of Genomic Domains in BRCA1 and BRCA2 with Prostate Cancer Risk and Aggressiveness. Cancer Research, 2020, 80, 624-638.	0.4	39
6	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. Nature Genetics, 2020, 52, 56-73.	9.4	120
7	Pathogenicity reclassification of two BRCA1/BRCA2 exonic duplications after identification of genomic breakpoints and tandem orientation. Cancer Genetics, 2020, 248-249, 18-24.	0.2	0
8	Haplotype analysis of the internationally distributed BRCA1 c.3331_3334delCAAG founder mutation reveals a common ancestral origin in Iberia. Breast Cancer Research, 2020, 22, 108.	2.2	9
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.	1.1	82
10	Hereditary Predisposition to Prostate Cancer: From Genetics to Clinical Implications. International Journal of Molecular Sciences, 2020, 21, 5036.	1.8	38
11	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.	1.3	11
12	Association of germline variation with the survival of women with BRCA1/2 pathogenic variants and breast cancer. Npj Breast Cancer, 2020, 6, 44.	2.3	5
13	The CHEK2 Variant C.349A>G Is Associated with Prostate Cancer Risk and Carriers Share a Common Ancestor. Cancers, 2020, 12, 3254.	1.7	16
14	Genome-wide association study identifies 32 novel breast cancer susceptibility loci from overall and subtype-specific analyses. Nature Genetics, 2020, 52, 572-581.	9.4	265
15	A Genetic Risk Score to Personalize Prostate Cancer Screening, Applied to Population Data. Cancer Epidemiology Biomarkers and Prevention, 2020, 29, 1731-1738.	1.1	27
16	Characterization of the Cancer Spectrum in Men With GermlineBRCA1andBRCA2Pathogenic Variants. JAMA Oncology, 2020, 6, 1218.	3.4	48
17	Transcriptomeâ€wide association study of breast cancer risk by estrogenâ€receptor status. Genetic Epidemiology, 2020, 44, 442-468.	0.6	32
18	A network analysis to identify mediators of germline-driven differences in breast cancer prognosis. Nature Communications, 2020, 11, 312.	5.8	30

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19	Shared heritability and functional enrichment across six solid cancers. Nature Communications, 2019, 10, 431.	5.8	88
20	Mendelian randomisation study of height and body mass index as modifiers of ovarian cancer risk in 22,588 BRCA1 and BRCA2 mutation carriers. British Journal of Cancer, 2019, 121, 180-192.	2.9	19
21	Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. Nature Communications, 2019, 10, 1741.	5.8	90
22	Genome-wide association study of germline variants and breast cancer-specific mortality. British Journal of Cancer, 2019, 120, 647-657.	2.9	52
23	Prostate-specific antigen velocity in a prospective prostate cancer screening study of men with genetic predisposition. British Journal of Cancer, 2018, 118, 266-276.	2.9	12
24	Polygenic hazard score to guide screening for aggressive prostate cancer: development and validation in large scale cohorts. BMJ: British Medical Journal, 2018, 360, j5757.	2.4	153
25	Germline pathogenic variants in PALB2 and other cancer-predisposing genes in families with hereditary diffuse gastric cancer without CDH1 mutation: a whole-exome sequencing study. The Lancet Gastroenterology and Hepatology, 2018, 3, 489-498.	3.7	87
26	Discontinuation of tyrosine kinase inhibitors in CML patients in real-world clinical practice at a single institution. BMC Cancer, 2018, 18, 1245.	1.1	15
27	Ponatinib induces a sustained deep molecular response in a chronic myeloid leukaemia patient with an early relapse with a T315I mutation following allogeneic hematopoietic stem cell transplantation: a case report. BMC Cancer, 2018, 18, 1229.	1.1	11
28	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.	0.2	6
29	Targeted next generation sequencing identifies functionally deleterious germline mutations in novel genes in early-onset/familial prostate cancer. PLoS Genetics, 2018, 14, e1007355.	1.5	50
30	A Transcriptome-Wide Association Study Among 97,898 Women to Identify Candidate Susceptibility Genes for Epithelial Ovarian Cancer Risk. Cancer Research, 2018, 78, 5419-5430.	0.4	54
31	AA9int: SNP interaction pattern search using non-hierarchical additive model set. Bioinformatics, 2018, 34, 4141-4150.	1.8	3
32	Full in-frame exon 3 skipping of BRCA2 confers high risk of breast and/or ovarian cancer. Oncotarget, 2018, 9, 17334-17348.	0.8	24
33	SNP interaction pattern identifier (SIPI): an intensive search for SNPâ€“SNP interaction patterns. Bioinformatics, 2017, 33, 822-833.	1.8	11
34	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.	1.2	13
35	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. Nature Genetics, 2017, 49, 680-691.	9.4	356
36	Germline Mutations in PALB2, BRCA1, and RAD51C, Which Regulate DNA Recombination Repair, in Patients With Gastric Cancer. Gastroenterology, 2017, 152, 983-986.e6.	0.6	98

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37	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. Nature Genetics, 2017, 49, 1767-1778.	9.4	289
38	Association of breast cancer risk in BRCA1 and BRCA2 mutation carriers with genetic variants showing differential allelic expression: identification of a modifier of breast cancer risk at locus 11q22.3. Breast Cancer Research and Treatment, 2017, 161, 117-134.	1.1	18
39	Investigating the possible causal role of coffee consumption with prostate cancer risk and progression using Mendelian randomization analysis. International Journal of Cancer, 2017, 140, 322-328.	2.3	17
40	Alcohol consumption and prostate cancer incidence and progression: A Mendelian randomisation study. International Journal of Cancer, 2017, 140, 75-85.	2.3	28
41	Fine-Scale Mapping at 9p22.2 Identifies Candidate Causal Variants That Modify Ovarian Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. PLoS ONE, 2016, 11, e0158801.	1.1	10
42	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.	1.1	15
43	PALB2,CHEK2andATMrare variants and cancer risk: data from COGS. Journal of Medical Genetics, 2016, 53, 800-811.	1.5	174
44	Identification of independent association signals and putative functional variants for breast cancer risk through fine-scale mapping of the 12p11 locus. Breast Cancer Research, 2016, 18, 64.	2.2	31
45	Prostate Cancer Prognosis Defined by the Combined Analysis of 8q, PTEN and ERG. Translational Oncology, 2016, 9, 575-582.	1.7	12
46	Genetic basis of PD-L1 overexpression in diffuse large B-cell lymphomas. Blood, 2016, 127, 3026-3034.	0.6	168
47	Polyunsaturated fatty acids and prostate cancer risk: a Mendelian randomisation analysis from the PRACTICAL consortium. British Journal of Cancer, 2016, 115, 624-631.	2.9	23
48	Assessing the role of insulinâ€like growth factors and binding proteins in prostate cancer using Mendelian randomization: Genetic variants as instruments for circulating levels. International Journal of Cancer, 2016, 139, 1520-1533.	2.3	26
49	Blood lipids and prostate cancer: a Mendelian randomization analysis. Cancer Medicine, 2016, 5, 1125-1136.	1.3	68
50	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.	1.1	23
51	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.	7.7	157
52	Inheritance of deleterious mutations at both BRCA1 and BRCA2 in an international sample of 32,295 women. Breast Cancer Research, 2016, 18, 112.	2.2	42
53	Prostate cancer risk regions at 8q24 and 17q24 are differentially associated with somaticTMPRSS2:ERGfusion status. Human Molecular Genetics, 2016, 25, ddw349.	1.4	8
54	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. Nature Communications, 2016, 7, 11375.	5.8	93

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55	Functional mechanisms underlying pleiotropic risk alleles at the 19p13.1 breastâ€“ovarian cancer susceptibility locus. Nature Communications, 2016, 7, 12675.	5.8	78
56	Atlas of prostate cancer heritability in European and African-American men pinpoints tissue-specific regulation. Nature Communications, 2016, 7, 10979.	5.8	50
57	Pubertal development and prostate cancer risk: Mendelian randomization study in a population-based cohort. BMC Medicine, 2016, 14, 66.	2.3	42
58	Breast cancer risk variants at 6q25 display different phenotype associations and regulate ESR1, RMND1 and CCDC170. Nature Genetics, 2016, 48, 374-386.	9.4	125
59	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.	1.1	8
60	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.	0.9	26
61	Meta-analysis of genome-wide association studies identifies common susceptibility polymorphisms for colorectal and endometrial cancer near SH2B3 and TSHZ1. Scientific Reports, 2015, 5, 17369.	1.6	35
62	Prediction of individual genetic risk to prostate cancer using a polygenic score. Prostate, 2015, 75, 1467-1474.	1.2	54
63	Assessing Associations between the AURKA-HMMR-TPX2-TUBG1 Functional Module and Breast Cancer Risk in BRCA1/2 Mutation Carriers. PLoS ONE, 2015, 10, e0120020.	1.1	34
64	Identification of Two Novel HOXB13 Germline Mutations in Portuguese Prostate Cancer Patients. PLoS ONE, 2015, 10, e0132728.	1.1	34
65	A Large-Scale Analysis of Genetic Variants within Putative miRNA Binding Sites in Prostate Cancer. Cancer Discovery, 2015, 5, 368-379.	7.7	56
66	Identification of six new susceptibility loci for invasive epithelial ovarian cancer. Nature Genetics, 2015, 47, 164-171.	9.4	221
67	Association of Type and Location ofBRCA1andBRCA2Mutations With Risk of Breast and Ovarian Cancer. JAMA - Journal of the American Medical Association, 2015, 313, 1347.	3.8	390
68	The effects of height and BMI on prostate cancer incidence and mortality: a Mendelian randomization study in 20,848 cases and 20,214 controls from the PRACTICAL consortium. Cancer Causes and Control, 2015, 26, 1603-1616.	0.8	77
69	Multiple novel prostate cancer susceptibility signals identified by fine-mapping of known risk loci among Europeans. Human Molecular Genetics, 2015, 24, 5589-5602.	1.4	67
70	Target gene mutational pattern in Lynch syndrome colorectal carcinomas according to tumour location and germline mutation. British Journal of Cancer, 2015, 113, 686-692.	2.9	30
71	Genome-Wide Association Study of Prostate Cancerâ€“Specific Survival. Cancer Epidemiology Biomarkers and Prevention, 2015, 24, 1796-1800.	1.1	27
72	Fine-mapping of the HNF1B multicancer locus identifies candidate variants that mediate endometrial cancer risk. Human Molecular Genetics, 2015, 24, 1478-1492.	1.4	50

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73	Candidate Genetic Modifiers for Breast and Ovarian Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. Cancer Epidemiology Biomarkers and Prevention, 2015, 24, 308-316.	1.1	22
74	Identification of previously unrecognized FAP in children with Gardner fibroma. European Journal of Human Genetics, 2015, 23, 715-718.	1.4	11
75	Uncovering potential downstream targets of oncogenic GRPR overexpression in prostate carcinomas harboring ETS rearrangements. Oncoscience, 2015, 2, 497-507.	0.9	11
76	Specific and redundant activities ofETV1andETV4in prostate cancer aggressiveness revealed by co-overexpression cellular contexts. Oncotarget, 2015, 6, 5217-5236.	0.8	24
77	DNA Glycosylases Involved in Base Excision Repair May Be Associated with Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. PLoS Genetics, 2014, 10, e1004256.	1.5	47
78	Fine-Mapping the HOXB Region Detects Common Variants Tagging a Rare Coding Allele: Evidence for Synthetic Association in Prostate Cancer. PLoS Genetics, 2014, 10, e1004129.	1.5	34
79	Carcinoma of the Thyroid With Ewing/PNET Family Tumor Elements. International Journal of Surgical Pathology, 2014, 22, 579-581.	0.4	18
80	Constitutional and somatic rearrangement of chromosome 21 in acute lymphoblastic leukaemia. Nature, 2014, 508, 98-102.	13.7	261
81	Targeted Prostate Cancer Screening in BRCA1 and BRCA2 Mutation Carriers: Results from the Initial Screening Round of the IMPACT Study. European Urology, 2014, 66, 489-499.	0.9	195
82	Pathogenicity Evaluation of BRCA1 and BRCA2 Unclassified Variants Identified in Portuguese Breast/Ovarian Cancer Families. Journal of Molecular Diagnostics, 2014, 16, 324-334.	1.2	24
83	A meta-analysis of 87,040 individuals identifies 23 new susceptibility loci for prostate cancer. Nature Genetics, 2014, 46, 1103-1109.	9.4	408
84	Exome sequencing reveals novel mutation targets in diffuse large B-cell lymphomas derived from Chinese patients. Blood, 2014, 124, 2544-2553.	0.6	102
85	Portuguese c.156_157insAlu BRCA2 founder mutation: gastrointestinal and tongue neoplasias may be part of the phenotype. Familial Cancer, 2012, 11, 657-660.	0.9	10
86	Genetic and Clinical Characterization of 45 Acute Leukemia Patients with MLL Gene Rearrangements From a Single Institution.. Blood, 2012, 120, 2477-2477.	0.6	0
87	Prognostic Impact of High Hematogones in Acute Myeloid Leukemia. Blood, 2012, 120, 1435-1435.	0.6	0
88	Cytogenetic Analysis of Tumor Clonality. Advances in Cancer Research, 2011, 112, 127-149.	1.9	21
89	International distribution and age estimation of the Portuguese BRCA2 c.156_157insAlu founder mutation. Breast Cancer Research and Treatment, 2011, 127, 671-679.	1.1	27
90	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.	1.1	52

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91	Overexpression of the Mitotic Checkpoint Genes BUB1 and BUBR1 is Associated with Genomic Complexity in Clear Cell Kidney Carcinomas. Analytical Cellular Pathology, 2008, 30, 389-395.	0.7	3
92	Recurrent Fusion Oncogenes in Carcinomas. Critical Reviews in Oncogenesis, 2006, 12, 257-271.	0.2	45
93	Multiple numerical chromosome aberrations in cancer: what are their causes and what are their consequences?. Seminars in Cancer Biology, 2005, 15, 3-12.	4.3	36
94	Genomic analysis of prostate carcinoma specimens obtained via ultrasound-guided needle biopsy may be of use in preoperative decision-making. Cancer, 2004, 101, 1786-1793.	2.0	28
95	Karyotypic divergence and convergence in two synchronous lung metastases of a clear cell sarcoma of tendons and aponeuroses with t(12;22)(q13;q12) and type 1 EWS/ATF1. Cancer Genetics and Cytogenetics, 2003, 145, 121-125.	1.0	5
96	Cytogenetic clues to breast carcinogenesis. Genes Chromosomes and Cancer, 2002, 33, 1-16.	1.5	61