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List of Publications by Year
in descending order

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83 papers	9,841 citations	87401 40 h-index	64407 83 g-index
83 all docs	83 docs citations	83 times ranked	15068 citing authors

#	Article	IF	Citations
1	Association analysis identifies 65 new breast cancer risk loci. Nature, 2017, 551, 92-94.	13.7	1,099
2	Large-scale genotyping identifies 41 new loci associated with breast cancer risk. Nature Genetics, 2013, 45, 353-361.	9.4	960
3	Polygenic Risk Scores for Prediction of Breast Cancer and Breast Cancer Subtypes. American Journal of Human Genetics, 2019, 104, 21-34.	2.6	711
4	Associations of Breast Cancer Risk Factors With Tumor Subtypes: A Pooled Analysis From the Breast Cancer Association Consortium Studies. Journal of the National Cancer Institute, 2011, 103, 250-263.	3.0	596
5	Breast Cancer Risk Genes â€" Association Analysis in More than 113,000 Women. New England Journal of Medicine, 2021, 384, 428-439.	13.9	532
6	Multiple independent variants at the TERT locus are associated with telomere length and risks of breast and ovarian cancer. Nature Genetics, 2013, 45, 371-384.	9.4	493
7	Prediction of Breast Cancer Risk Based on Profiling With Common Genetic Variants. Journal of the National Cancer Institute, 2015, 107, .	3.0	428
8	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
9	Genome-wide association studies identify four ER negativeâ€"specific breast cancer risk loci. Nature Genetics, 2013, 45, 392-398.	9.4	374
10	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. Nature Genetics, 2017, 49, 1767-1778.	9.4	289
11	Genome-Wide Association Study in BRCA1 Mutation Carriers Identifies Novel Loci Associated with Breast and Ovarian Cancer Risk. PLoS Genetics, 2013, 9, e1003212.	1.5	244
12	RAD51 135Gâ†'C Modifies Breast Cancer Risk among BRCA2 Mutation Carriers: Results from a Combined Analysis of 19 Studies. American Journal of Human Genetics, 2007, 81, 1186-1200.	2.6	217
13	A transcriptome-wide association study of 229,000 women identifies new candidate susceptibility genes for breast cancer. Nature Genetics, 2018, 50, 968-978.	9.4	184
14	Tumor MicroRNA Expression Profiling Identifies Circulating MicroRNAs for Early Breast Cancer Detection. Clinical Chemistry, 2015, 61, 1098-1106.	1.5	183
15	Common Breast Cancer Susceptibility Alleles and the Risk of Breast Cancer for BRCA1 and BRCA2 Mutation Carriers: Implications for Risk Prediction. Cancer Research, 2010, 70, 9742-9754.	0.4	169
16	Phenotypic characterization of BRCA1 and BRCA2 tumors based in a tissue microarray study with 37 immunohistochemical markers. Breast Cancer Research and Treatment, 2005, 90, 5-14.	1.1	147
17	The complex genetic landscape of familial breast cancer. Human Genetics, 2013, 132, 845-863.	1.8	125
18	A mutation in the POT1 gene is responsible for cardiac angiosarcoma in TP53-negative Liâ€"Fraumeni-like families. Nature Communications, 2015, 6, 8383.	5.8	124

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19	Genetically Predicted Body Mass Index and Breast Cancer Risk: Mendelian Randomization Analyses of Data from 145,000 Women of European Descent. PLoS Medicine, 2016, 13, e1002105.	3.9	118
20	Identification of a BRCA2-Specific Modifier Locus at 6p24 Related to Breast Cancer Risk. PLoS Genetics, 2013, 9, e1003173.	1.5	105
21	Height and Breast Cancer Risk: Evidence From Prospective Studies and Mendelian Randomization. Journal of the National Cancer Institute, 2015, 107, djv219.	3.0	99
22	Whole Exome Sequencing Suggests Much of Non-BRCA1/BRCA2 Familial Breast Cancer Is Due to Moderate and Low Penetrance Susceptibility Alleles. PLoS ONE, 2013, 8, e55681.	1.1	95
23	No evidence that protein truncating variants inBRIP1are associated with breast cancer risk: implications for gene panel testing. Journal of Medical Genetics, 2016, 53, 298-309.	1.5	94
24	Male breast cancer in BRCA1 and BRCA2 mutation carriers: pathology data from the Consortium of Investigators of Modifiers of BRCA1/2. Breast Cancer Research, 2016, 18, 15.	2.2	88
25	Analysis of FANCB and FANCN/PALB2 Fanconi Anemia genes in BRCA1/2-negative Spanish breast cancer families. Breast Cancer Research and Treatment, 2009, 113, 545-551.	1.1	83
26	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
27	The role of genetic breast cancer susceptibility variants as prognostic factors. Human Molecular Genetics, 2012, 21, 3926-3939.	1.4	80
28	Functional mechanisms underlying pleiotropic risk alleles at the 19p13.1 breastâ€“ovarian cancer susceptibility locus. Nature Communications, 2016, 7, 12675.	5.8	78
29	BRCA2 Hypomorphic Missense Variants Confer Moderate Risks of Breast Cancer. Cancer Research, 2017, 77, 2789-2799.	0.4	75
30	Exome sequencing identifies ATP4A gene as responsible of an atypical familial type I gastric neuroendocrine tumour. Human Molecular Genetics, 2015, 24, 2914-2922.	1.4	60
31	Evidence that the 5p12 Variant rs10941679 Confers Susceptibility to Estrogen-Receptor-Positive Breast Cancer through FGF10 and MRPS30 Regulation. American Journal of Human Genetics, 2016, 99, 903-911.	2.6	59
32	Identification of Novel Genetic Markers of Breast Cancer Survival. Journal of the National Cancer Institute, 2015, 107, .	3.0	56
33	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
34	Genome-wide association study of germline variants and breast cancer-specific mortality. British Journal of Cancer, 2019, 120, 647-657.	2.9	52
35	Fineâ€scale mapping of 8q24 locus identifies multiple independent risk variants for breast cancer. International Journal of Cancer, 2016, 139, 1303-1317.	2.3	51
36	E-cadherin breast tumor expression, risk factors and survival: Pooled analysis of 5,933 cases from 12 studies in the Breast Cancer Association Consortium. Scientific Reports, 2018, 8, 6574.	1.6	51

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37	Pathology of Tumors Associated With Pathogenic Germline Variants in 9 Breast Cancer Susceptibility Genes. JAMA Oncology, 2022, 8, e216744.	3.4	51
38	Association Between a Germline OCA2 Polymorphism at Chromosome 15q13.1 and Estrogen Receptorâ€“Negative Breast Cancer Survival. Journal of the National Cancer Institute, 2010, 102, 650-662.	3.0	48
39	Characterization of the Cancer Spectrum in Men With GermlineBRCA1andBRCA2Pathogenic Variants. JAMA Oncology, 2020, 6, 1218.	3.4	48
40	A role for XRCC2 gene polymorphisms in breast cancer risk and survival. Journal of Medical Genetics, 2011, 48, 477-484.	1.5	47
41	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
42	Body mass index and breast cancer survival: a Mendelian randomization analysis. International Journal of Epidemiology, 2017, 46, 1814-1822.	0.9	45
43	MicroRNA expression signatures for the prediction of BRCA1/2 mutationâ€associated hereditary breast cancer in paraffinâ€embedded formalinâ€fixed breast tumors. International Journal of Cancer, 2015, 136, 593-602.	2.3	43
44	Genetic predisposition to ductal carcinoma in situ of the breast. Breast Cancer Research, 2016, 18, 22.	2.2	43
45	Reproductive profiles and risk of breast cancer subtypes: a multi-center case-only study. Breast Cancer Research, 2017, 19, 119.	2.2	43
46	MicroRNA deregulation in triple negative breast cancer reveals a role of miR-498 in regulating BRCA1 expression. Oncotarget, 2016, 7, 20068-20079.	0.8	42
47	Analysis of myelodysplastic syndromes with complex karyotypes by high-resolution comparative genomic hybridization and subtelomeric CGH array. Genes Chromosomes and Cancer, 2005, 42, 287-298.	1.5	40
48	Association of Genomic Domains in BRCA1 and BRCA2 with Prostate Cancer Risk and Aggressiveness. Cancer Research, 2020, 80, 624-638.	0.4	39
49	Polymorphisms in a Putative Enhancer at the 10q21.2 Breast Cancer Risk Locus Regulate NRBF2 Expression. American Journal of Human Genetics, 2015, 97, 22-34.	2.6	37
50	The Fanconi anaemia/BRCA pathway and cancer susceptibility. Searching for new therapeutic targets. Clinical and Translational Oncology, 2008, 10, 78-84.	1.2	32
51	Association of breast cancer risk with genetic variants showing differential allelic expression: Identification of a novel breast cancer susceptibility locus at 4q21. Oncotarget, 2016, 7, 80140-80163.	0.8	31
52	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
53	Genomic analysis of the 8p11-12 amplicon in familial breast cancer. International Journal of Cancer, 2007, 120, 714-717.	2.3	30
54	Genetic characterization and structural analysis of VHL Spanish families to define genotype-phenotype correlations. Human Mutation, 2004, 23, 160-169.	1.1	28

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55	Evaluation of Rare Variants in the New Fanconi Anemia GeneERCC4(FANCQ) as Familial Breast/Ovarian Cancer Susceptibility Alleles. Human Mutation, 2013, 34, 1615-1618.	1.1	28
56	An original phylogenetic approach identified mitochondrial haplogroup T1a1 as inversely associated with breast cancer risk in BRCA2 mutation carriers. Breast Cancer Research, 2015, 17, 61.	2.2	26
57	Common germline polymorphisms associated with breast cancer-specific survival. Breast Cancer Research, 2015, 17, 58.	2.2	26
58	DNA repair capacity is impaired in healthy BRCA1 heterozygous mutation carriers. Breast Cancer Research and Treatment, 2015, 152, 271-282.	1.1	26
59	RAD51B in Familial Breast Cancer. PLoS ONE, 2016, 11, e0153788.	1.1	26
60	Fine-Scale Mapping of the 4q24 Locus Identifies Two Independent Loci Associated with Breast Cancer Risk. Cancer Epidemiology Biomarkers and Prevention, 2015, 24, 1680-1691.	1.1	24
61	A 7 Mb region within 11q13 may contain a high penetrance gene for breast cancer. Breast Cancer Research and Treatment, 2009, 118, 151-159.	1.1	23
62	Association of genetic susceptibility variants for type 2 diabetes with breast cancer risk in women of European ancestry. Cancer Causes and Control, 2016, 27, 679-693.	0.8	21
63	Fine scale mapping of the 17q22 breast cancer locus using dense SNPs, genotyped within the Collaborative Oncological Gene-Environment Study (COGs). Scientific Reports, 2016, 6, 32512.	1.6	19
64	Highâ€tthroughput automated scoring of Ki67 in breast cancer tissue microarrays from the Breast Cancer Association Consortium. Journal of Pathology: Clinical Research, 2016, 2, 138-153.	1.3	19
65	The BRCA2 c.68-7TÂ>ÂA variant is not pathogenic: A model for clinical calibration of spliceogenicity. Human Mutation, 2018, 39, 729-741.	1.1	19
66	Etiology of hormone receptor positive breast cancer differs by levels of histologic grade and proliferation. International Journal of Cancer, 2018, 143, 746-757.	2.3	19
67	Whole exome sequencing identifies PLEC, EXO5 and DNAH7 as novel susceptibility genes in testicular cancer. International Journal of Cancer, 2018, 143, 1954-1962.	2.3	19
68	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
69	A knockin mouse model for human ATP4a R703C mutation identified in familial gastric neuroendocrine tumors recapitulates the premalignant condition of the human disease and suggests new therapeutic strategies. DMM Disease Models and Mechanisms, 2016, 9, 975-84.	1.2	18
70	Gene expression analysis of chromosomal regions with gain or loss of genetic material detected by comparative genomic hybridization. Genes Chromosomes and Cancer, 2004, 41, 353-365.	1.5	17
71	RECQL5 : Another DNA helicase potentially involved in hereditary breast cancer susceptibility. Human Mutation, 2019, 40, 566-577.	1.1	16
72	BRCA1 and BRCA2 mutations in males with familial breast and ovarian cancer syndrome. Results of a Spanish multicenter study. Familial Cancer, 2015, 14, 505-513.	0.9	15

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73	Genetic variation at CYP3A is associated with age at menarche and breast cancer risk: a case-control study. Breast Cancer Research, 2014, 16, R51.	2.2	14
74	miRNA expression profiling of formalin-fixed paraffin-embedded (FFPE) hereditary breast tumors. Genomics Data, 2015, 3, 75-79.	1.3	12
75	Fine-Mapping of the 1p11.2 Breast Cancer Susceptibility Locus. PLoS ONE, 2016, 11, e0160316.	1.1	12
76	A cumulative effect involving malfunction of the PTH1R and ATP4A genes explains a familial gastric neuroendocrine tumor with hypothyroidism and arthritis. Gastric Cancer, 2017, 20, 998-1003.	2.7	12
77	The Spectrum of FANCM Protein Truncating Variants in European Breast Cancer Cases. Cancers, 2020, 12, 292.	1.7	11
78	PHIP- a novel candidate breast cancer susceptibility locus on 6q14.1. Oncotarget, 2017, 8, 102769-102782.	0.8	9
79	Genetic variation in the immunosuppression pathway genes and breast cancer susceptibility: a pooled analysis of 42,510 cases and 40,577 controls from the Breast Cancer Association Consortium. Human Genetics, 2016, 135, 137-154.	1.8	8
80	Deep Sequencing of Target Linkage Assay-Identified Regions in Familial Breast Cancer: Methods, Analysis Pipeline and Troubleshooting. PLoS ONE, 2010, 5, e9976.	1.1	6
81	Pharmacogenetic variants and response to neoadjuvant single-agent doxorubicin or docetaxel. Pharmacogenetics and Genomics, 2018, 28, 245-250.	0.7	3
82	rs2735383, located at a microRNA binding site in the 3â€™UTR of NBS1, is not associated with breast cancer risk. Scientific Reports, 2016, 6, 36874.	1.6	2
83	Cuando el cÃ¡ncer es una enfermedad rara. Arbor, 2018, 194, 464.	0.1	2