List of Publications by Year in descending order

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#	Article	IF	CITATIONS
1	Polygenic Risk Scores for Prediction of Breast Cancer and Breast Cancer Subtypes. American Journal of Human Genetics, 2019, 104, 21-34.	6.2	711
2	Association of Type and Location of <i>BRCA1</i> and <i>BRCA2</i> Mutations With Risk of Breast and Ovarian Cancer. JAMA - Journal of the American Medical Association, 2015, 313, 1347.	7.4	390
3	Germline Mutation Status, Pathological Complete Response, and Disease-Free Survival in Triple-Negative Breast Cancer. JAMA Oncology, 2017, 3, 1378.	7.1	300
4	Cancer Risks Associated With Germline <i>PALB2</i> Pathogenic Variants: An International Study of 524 Families. Journal of Clinical Oncology, 2020, 38, 674-685.	1.6	270
5	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
6	Clinical and molecular characteristics of HER2-low-positive breast cancer: pooled analysis of individual patient data from four prospective, neoadjuvant clinical trials. Lancet Oncology, The, 2021, 22, 1151-1161.	10.7	248
7	Mutational spectrum in a worldwide study of 29,700 families with <i>BRCA1</i> or <i>BRCA2</i> mutations. Human Mutation, 2018, 39, 593-620.	2.5	224
8	Prevalence of <i>BRCA1/2</i> germline mutations in 21â€401 families with breast and ovarian cancer. Journal of Medical Genetics, 2016, 53, 465-471.	3.2	179
9	In vitro and ex vivo evaluation of second-generation histone deacetylase inhibitors for the treatment of spinal muscular atrophy. Journal of Neurochemistry, 2006, 98, 193-202.	3.9	140
10	Gene panel testing of 5589 <i><scp>BRCA</scp>1/2</i> â€negative index patients with breast cancer in a routine diagnostic setting: results of the German Consortium for Hereditary Breast and Ovarian Cancer. Cancer Medicine, 2018, 7, 1349-1358.	2.8	126
11	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. Nature Genetics, 2020, 52, 56-73.	21.4	120
12	Ovarian and Breast Cancer Risks Associated With Pathogenic Variants in <i>RAD51C</i> and <i>RAD51D</i> . Journal of the National Cancer Institute, 2020, 112, 1242-1250.	6.3	106
13	Prevalence of deleterious germline variants in risk genes including BRCA1/2 in consecutive ovarian cancer patients (AGO-TR-1). PLoS ONE, 2017, 12, e0186043.	2.5	105
14	Large scale multifactorial likelihood quantitative analysis of <i>BRCA1</i> and <i>BRCA2</i> variants: An ENIGMA resource to support clinical variant classification. Human Mutation, 2019, 40, 1557-1578.	2.5	102
15	Cancer Risks Associated With <i>BRCA1</i> and <i>BRCA2</i> Pathogenic Variants. Journal of Clinical Oncology, 2022, 40, 1529-1541.	1.6	90
16	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.	5.0	88
17	Prevalence of pathogenic BRCA1/2 germline mutations among 802 women with unilateral triple-negative breast cancer without family cancer history. BMC Cancer, 2018, 18, 265.	2.6	84
18	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

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19	Performance of in silico prediction tools for the classification of rare BRCA1/2 missense variants in clinical diagnostics. BMC Medical Genomics, 2018, 11, 35.	1.5	78
20	BRIP1 loss-of-function mutations confer high risk for familial ovarian cancer, but not familial breast cancer. Breast Cancer Research, 2018, 20, 7.	5.0	78
21	Association Between Loss-of-Function Mutations Within the <i>FANCM</i> Gene and Early-Onset Familial Breast Cancer. JAMA Oncology, 2017, 3, 1245.	7.1	74
22	Germline Mutations in Triple-Negative Breast Cancer. Breast Care, 2017, 12, 15-19.	1.4	59
23	Individuals with FANCM biallelic mutations do not develop Fanconi anemia, but show risk for breast cancer, chemotherapy toxicity and may display chromosome fragility. Genetics in Medicine, 2018, 20, 452-457.	2.4	59
24	Personalized Risk Assessment for Prevention and Early Detection of Breast Cancer: Integration and Implementation (PERSPECTIVE I&I). Journal of Personalized Medicine, 2021, 11, 511.	2.5	59
25	Pathology of Tumors Associated With Pathogenic Germline Variants in 9 Breast Cancer Susceptibility Genes. JAMA Oncology, 2022, 8, e216744.	7.1	51
26	Characterization of the Cancer Spectrum in Men With Germline <i>BRCA1</i> and <i>BRCA2</i> Pathogenic Variants. JAMA Oncology, 2020, 6, 1218.	7.1	48
27	Germline loss-of-function variants in the BARD1 gene are associated with early-onset familial breast cancer but not ovarian cancer. Breast Cancer Research, 2019, 21, 55.	5.0	44
28	Inheritance of deleterious mutations at both BRCA1 and BRCA2 in an international sample of 32,295 women. Breast Cancer Research, 2016, 18, 112.	5.0	42
29	Association of Germline Variant Status With Therapy Response in High-risk Early-Stage Breast Cancer. JAMA Oncology, 2020, 6, 744.	7.1	42
30	Association of Genomic Domains in <i>BRCA1</i> and <i>BRCA2</i> with Prostate Cancer Risk and Aggressiveness. Cancer Research, 2020, 80, 624-638.	0.9	39
31	Benchmarking of a checklist for the identification of familial risk for breast and ovarian cancers in a prospective cohort. Breast Journal, 2019, 25, 455-460.	1.0	35
32	Deleterious somatic variants in 473 consecutive individuals with ovarian cancer: results of the observational AGO-TR1 study (NCT02222883). Journal of Medical Genetics, 2019, 56, 574-580.	3.2	34
33	Breast cancer risk in <i>BRCA1/2</i> mutation carriers and noncarriers under prospective intensified surveillance. International Journal of Cancer, 2020, 146, 999-1009.	5.1	32
34	RAD51Cdeletion screening identifies a recurrent gross deletion in breast cancer and ovarian cancer families. Breast Cancer Research, 2013, 15, R120.	5.0	28
35	An evaluation of the challenges to developing tumor BRCA1 and BRCA2 testing methodologies for clinical practice. Human Mutation, 2018, 39, 394-405.	2.5	24
36	Polygenic risk modeling for prediction of epithelial ovarian cancer risk. European Journal of Human Genetics, 2022, 30, 349-362.	2.8	23

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37	Biallelic germline <i>BRCA1</i> mutations in a patient with early onset breast cancer, mild Fanconi anemiaâ€like phenotype, and no chromosome fragility. Molecular Genetics & Genomic Medicine, 2019, 7, e863.	1.2	22
38	Performance of Breast Cancer Polygenic Risk Scores in 760 Female <i>CHEK2</i> Germline Mutation Carriers. Journal of the National Cancer Institute, 2021, 113, 893-899.	6.3	21
39	Diagnosis of Li-Fraumeni Syndrome: Differentiating <i>TP53</i> germline mutations from clonal hematopoiesis. Human Mutation, 2018, 39, 2040-2046.	2.5	20
40	A case-only study to identify genetic modifiers of breast cancer risk for BRCA1/BRCA2 mutation carriers. Nature Communications, 2021, 12, 1078.	12.8	19
41	Breast and Prostate Cancer Risks for Male <i>BRCA1</i> and <i>BRCA2</i> Pathogenic Variant Carriers Using Polygenic Risk Scores. Journal of the National Cancer Institute, 2022, 114, 109-122.	6.3	19
42	Breast cancer risks associated with missense variants in breast cancer susceptibility genes. Genome Medicine, 2022, 14, 51.	8.2	19
43	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.	2.5	18
44	Clonal Hematopoiesis–Associated Gene Mutations in a Clinical Cohort of 448 Patients With Ovarian Cancer. Journal of the National Cancer Institute, 2022, 114, 565-570.	6.3	17
45	The predictive ability of the 313 variant–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.	2.4	16
46	Effect of <scp>HIPEC</scp> according to <scp>HRD</scp> / <scp><i>BRCA</i>wt</scp> genomic profile in stage <scp>III</scp> ovarian cancer: Results from the phase <scp>III OVHIPEC</scp> trial. International Journal of Cancer, 2022, 151, 1394-1404.	5.1	15
47	Consensus Recommendations of the German Consortium for Hereditary Breast and Ovarian Cancer. Breast Care, 2022, 17, 199-207.	1.4	12
48	Survival analysis of the randomised phase III GeparOcto trial comparing neoadjuvant chemotherapy of intense dose-dense epirubicin, paclitaxel, cyclophosphamide versus weekly paclitaxel, liposomal doxorubicin (plus carboplatin in triple-negative breast cancer) for patients with high-risk early breast cancer. European Journal of Cancer, 2022, 160, 100-111.	2.8	12
49	Prevalence of Cancer Predisposition Germline Variants in Male Breast Cancer Patients: Results of the German Consortium for Hereditary Breast and Ovarian Cancer. Cancers, 2022, 14, 3292.	3.7	11
50	Non-small cell neuroendocrine carcinoma of the ovary in a BRCA2-germline mutation carrier: A case report and brief review of the literature. Oncology Letters, 2018, 15, 4093-4096.	1.8	10
51	Investigating the effects of additional truncating variants in DNA-repair genes on breast cancer risk in BRCA1-positive women. BMC Cancer, 2019, 19, 787.	2.6	10
52	Mendelian randomisation study of smoking exposure in relation to breast cancer risk. British Journal of Cancer, 2021, 125, 1135-1145.	6.4	9
53	Ovarian Cancer–Specific <i>BRCA</i> -like Copy-Number Aberration Classifiers Detect Mutations Associated with Homologous Recombination Deficiency in the AGO-TR1 Trial. Clinical Cancer Research, 2021, 27, 6559-6569.	7.0	9
54	Evaluation of the association of heterozygous germline variants in NTHL1 with breast cancer predisposition: an international multi-center study of 47,180 subjects. Npj Breast Cancer, 2021, 7, 52.	5.2	7

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55	Association of germline genetic variants with breast cancer-specific survival in patient subgroups defined by clinic-pathological variables related to tumor biology and type of systemic treatment. Breast Cancer Research, 2021, 23, 86.	5.0	7
56	Functional annotation of the 2q35 breast cancer risk locus implicates a structural variant in influencing activity of a long-range enhancer element. American Journal of Human Genetics, 2021, 108, 1190-1203.	6.2	6
57	Rare germline copy number variants (CNVs) and breast cancer risk. Communications Biology, 2022, 5, 65.	4.4	6
58	Association of germline variation with the survival of women with BRCA1/2 pathogenic variants and breast cancer. Npj Breast Cancer, 2020, 6, 44.	5.2	5
59	CYP3A7*1C allele: linking premenopausal oestrone and progesterone levels with risk of hormone receptor-positive breast cancers. British Journal of Cancer, 2021, 124, 842-854.	6.4	5
60	Drug discovery for spinal muscular atrophy. Expert Opinion on Drug Discovery, 2007, 2, 437-451.	5.0	4
61	Performance of In Silico Prediction Tools for the Detection of Germline Copy Number Variations in Cancer Predisposition Genes in 4208 Female Index Patients with Familial Breast and Ovarian Cancer. Cancers, 2021, 13, 118.	3.7	4
62	Sensitivity and specificity of loss of heterozygosity analysis for the classification of rare germline variants in BRCA1/2: results of the observational AGO-TR1 study (NCT02222883). Journal of Medical Genetics, 2020, , jmedgenet-2020-107353.	3.2	3
63	Pathological complete response rate and survival in patients with <i>BRCA</i> -associated triple-negative breast cancer after 12 weeks of de-escalated neoadjuvant chemotherapy: Translational results of the WSG-ADAPT TN randomized phase II trial (NCT01815242) Journal of Clinical Oncology, 2021. 39. 579-579.	1.6	3
64	<i>BRCA</i> -like classification in ovarian cancer: Results from the AGO-TR1-trial Journal of Clinical Oncology, 2017, 35, 5546-5546.	1.6	3
65	The <i>GPRC5A</i> frameshift variant c.183del is not associated with increased breast cancer risk in <i>BRCA1</i> mutation carriers. International Journal of Cancer, 2019, 144, 1761-1763.	5.1	2
66	Incidence of germline mutations in risk genes including <i>BRCA1/2</i> in consecutive ovarian cancer (OC) patients (AGO TR-1) Journal of Clinical Oncology, 2016, 34, 5538-5538.	1.6	2
67	Germline variants and breast cancer survival in patients with distant metastases at primary breast cancer diagnosis. Scientific Reports, 2021, 11, 19787.	3.3	2
68	Uncovering the Contribution of Moderate-Penetrance Susceptibility Genes to Breast Cancer by Whole-Exome Sequencing and Targeted Enrichment Sequencing of Candidate Genes in Women of European Ancestry. Cancers, 2022, 14, 3363.	3.7	2