

# Judith Balmaña

## List of Publications by Year in descending order

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150  
papers

10,028  
citations

53660

45  
h-index

39575

94  
g-index

157  
all docs

157  
docs citations

157  
times ranked

13814  
citing authors

#	ARTICLE	IF	CITATIONS
1	Olaparib Monotherapy in Patients With Advanced Cancer and a Germline <i>BRCA1/2</i> Mutation. <i>Journal of Clinical Oncology</i> , 2015, 33, 244-250.	0.8	1,473
2	Adjuvant Olaparib for Patients with <i>BRCA1</i> - or <i>BRCA2</i> -Mutated Breast Cancer. <i>New England Journal of Medicine</i> , 2021, 384, 2394-2405.	13.9	764
3	Combining a PI3K Inhibitor with a PARP Inhibitor Provides an Effective Therapy for <i>BRCA1</i> -Related Breast Cancer. <i>Cancer Discovery</i> , 2012, 2, 1048-1063.	7.7	384
4	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. <i>Nature Genetics</i> , 2017, 49, 680-691.	9.4	356
5	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. <i>Nature Genetics</i> , 2017, 49, 1767-1778.	9.4	289
6	<i>RAD51</i> foci as a functional biomarker of homologous recombination repair and PARP inhibitor resistance in germline <i>BRCA</i> -mutated breast cancer. <i>Annals of Oncology</i> , 2018, 29, 1203-1210.	0.6	280
7	Cancer Risks Associated With Germline <i>PALB2</i> Pathogenic Variants: An International Study of 524 Families. <i>Journal of Clinical Oncology</i> , 2020, 38, 674-685.	0.8	270
8	Efficacy and safety of olaparib monotherapy in germline <i>BRCA1 / 2</i> mutation carriers with advanced ovarian cancer and three or more lines of prior therapy. <i>Gynecologic Oncology</i> , 2016, 140, 199-203.	0.6	252
9	Genome-Wide Association Study in <i>BRCA1</i> Mutation Carriers Identifies Novel Loci Associated with Breast and Ovarian Cancer Risk. <i>PLoS Genetics</i> , 2013, 9, e1003212.	1.5	244
10	Mutational spectrum in a worldwide study of 29,700 families with <i>BRCA1</i> or <i>BRCA2</i> mutations. <i>Human Mutation</i> , 2018, 39, 593-620.	1.1	224
11	<i>CDK12</i> Inhibition Reverses De Novo and Acquired PARP Inhibitor Resistance in <i>BRCA</i> Wild-Type and Mutated Models of Triple-Negative Breast Cancer. <i>Cell Reports</i> , 2016, 17, 2367-2381.	2.9	215
12	A Phase II Study of the Oral PARP Inhibitor Rucaparib in Patients with Germline <i>BRCA1/2</i> -Mutated Ovarian Carcinoma or Other Solid Tumors. <i>Clinical Cancer Research</i> , 2017, 23, 4095-4106.	3.2	213
13	The <i>BRCA1</i> 11q Alternative Splice Isoform Bypasses Germline Mutations and Promotes Therapeutic Resistance to PARP Inhibition and Cisplatin. <i>Cancer Research</i> , 2016, 76, 2778-2790.	0.4	208
14	The <i>PREMM1,2,6</i> Model Predicts Risk of <i>MLH1</i> , <i>MSH2</i> , and <i>MSH6</i> Germline Mutations Based on Cancer History. <i>Gastroenterology</i> , 2011, 140, 73-81.e5.	0.6	171
15	Local therapy in <i>BRCA1</i> and <i>BRCA2</i> mutation carriers with operable breast cancer: comparison of breast conservation and mastectomy. <i>Breast Cancer Research and Treatment</i> , 2010, 121, 389-398.	1.1	170
16	A <i>RAD51</i> assay feasible in routine tumor samples calls PARP inhibitor response beyond <i>BRCA</i> mutation. <i>EMBO Molecular Medicine</i> , 2018, 10, .	3.3	169
17	Prediction of <i>MLH1</i> and <i>MSH2</i> Mutations in Lynch Syndrome. <i>JAMA - Journal of the American Medical Association</i> , 2006, 296, 1469.	3.8	160
18	Prediction of Breast and Prostate Cancer Risks in Male <i>BRCA1</i> and <i>BRCA2</i> Mutation Carriers Using Polygenic Risk Scores. <i>Journal of Clinical Oncology</i> , 2017, 35, 2240-2250.	0.8	152

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19	Conflicting Interpretation of Genetic Variants and Cancer Risk by Commercial Laboratories as Assessed by the Prospective Registry of Multiplex Testing. <i>Journal of Clinical Oncology</i> , 2016, 34, 4071-4078.	0.8	147
20	Breast cancer phenotype in women with TP53 germline mutations: a Li-Fraumeni syndrome consortium effort. <i>Breast Cancer Research and Treatment</i> , 2012, 133, 1125-1130.	1.1	144
21	Phase I trial of olaparib in combination with cisplatin for the treatment of patients with advanced breast, ovarian and other solid tumors. <i>Annals of Oncology</i> , 2014, 25, 1656-1663.	0.6	144
22	Moving From Poly (ADP-Ribose) Polymerase Inhibition to Targeting DNA Repair and DNA Damage Response in Cancer Therapy. <i>Journal of Clinical Oncology</i> , 2019, 37, 2257-2269.	0.8	135
23	Ovarian and Breast Cancer Risks Associated With Pathogenic Variants in <i>RAD51C</i> and <i>RAD51D</i> . <i>Journal of the National Cancer Institute</i> , 2020, 112, 1242-1250.	3.0	106
24	BRCA1185delAG tumors may acquire therapy resistance through expression of RING-less BRCA1. <i>Journal of Clinical Investigation</i> , 2016, 126, 2903-2918.	3.9	105
25	Large scale multifactorial likelihood quantitative analysis of <i>BRCA1</i> and <i>BRCA2</i> variants: An ENIGMA resource to support clinical variant classification. <i>Human Mutation</i> , 2019, 40, 1557-1578.	1.1	102
26	A Phase II Study of Talazoparib after Platinum or Cytotoxic Nonplatinum Regimens in Patients with Advanced Breast Cancer and Germline <i>BRCA1/2</i> Mutations (ABRAZO). <i>Clinical Cancer Research</i> , 2019, 25, 2717-2724.	3.2	102
27	<i>FANCM</i> c.5791C>T nonsense mutation (rs144567652) induces exon skipping, affects DNA repair activity and is a familial breast cancer risk factor. <i>Human Molecular Genetics</i> , 2015, 24, 5345-5355.	1.4	91
28	Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. <i>Nature Communications</i> , 2019, 10, 1741.	5.8	90
29	Male breast cancer in BRCA1 and BRCA2 mutation carriers: pathology data from the Consortium of Investigators of Modifiers of BRCA1/2. <i>Breast Cancer Research</i> , 2016, 18, 15.	2.2	88
30	Role of POLE and POLD1 in familial cancer. <i>Genetics in Medicine</i> , 2020, 22, 2089-2100.	1.1	76
31	Microsatellite instability and immunostaining for MSH-2 and MLH-1 in cutaneous and internal tumors from patients with the Muir-Torre syndrome. <i>Journal of Cutaneous Pathology</i> , 2002, 29, 415-420.	0.7	72
32	Prevalence of germline MUTYH mutations among Lynch-like syndrome patients. <i>European Journal of Cancer</i> , 2014, 50, 2241-2250.	1.3	66
33	Phenotype Comparison of MLH1 and MSH2 Mutation Carriers in a Cohort of 1,914 Individuals Undergoing Clinical Genetic Testing in the United States. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2008, 17, 2044-2051.	1.1	65
34	Development and Validation of a Colon Cancer Risk Assessment Tool for Patients Undergoing Colonoscopy. <i>American Journal of Gastroenterology</i> , 2009, 104, 1508-1518.	0.2	62
35	Opinion about reproductive decision making among individuals undergoing BRCA1/2 genetic testing in a multicentre Spanish cohort. <i>Human Reproduction</i> , 2008, 24, 1000-1006.	0.4	61
36	Screening and surveillance in hereditary gastrointestinal cancers: Recommendations from the European Society of Digestive Oncology (ESDO) expert discussion at the 20th European Society for Medical Oncology (ESMO)/World Congress on Gastrointestinal Cancer, Barcelona, June 2018. <i>European Journal of Cancer</i> , 2018, 104, 91-103.	1.3	60

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37	Validation and Extension of the PREMM1,2 Model in a Population-Based Cohort of Colorectal Cancer Patients. <i>Gastroenterology</i> , 2008, 134, 39-46.	0.6	57
38	Prevalence and Characteristics of <i>MUTYH</i> -Associated Polyposis in Patients with Multiple Adenomatous and Serrated Polyps. <i>Clinical Cancer Research</i> , 2014, 20, 1158-1168.	3.2	57
39	Parity and the risk of breast and ovarian cancer in BRCA1 and BRCA2 mutation carriers. <i>Breast Cancer Research and Treatment</i> , 2010, 119, 221-232.	1.1	56
40	Characterization of BRCA1 and BRCA2 splicing variants: a collaborative report by ENIGMA consortium members. <i>Breast Cancer Research and Treatment</i> , 2012, 132, 1009-1023.	1.1	56
41	Computational Tools for Splicing Defect Prediction in Breast/Ovarian Cancer Genes: How Efficient Are They at Predicting RNA Alterations?. <i>Frontiers in Genetics</i> , 2018, 9, 366.	1.1	53
42	Multigene panel testing beyond BRCA1/2 in breast/ovarian cancer Spanish families and clinical actionability of findings. <i>Journal of Cancer Research and Clinical Oncology</i> , 2018, 144, 2495-2513.	1.2	53
43	Detection of a large rearrangement in PALB2 in Spanish breast cancer families with male breast cancer. <i>Breast Cancer Research and Treatment</i> , 2012, 132, 307-315.	1.1	50
44	Comparison of the clinical prediction model PREMM <sub>1,2,6</sub> and molecular testing for the systematic identification of Lynch syndrome in colorectal cancer. <i>Gut</i> , 2013, 62, 272-279.	6.1	49
45	Controversies in oncology: are genomic tests quantifying homologous recombination repair deficiency (HRD) useful for treatment decision making?. <i>ESMO Open</i> , 2019, 4, e000480.	2.0	47
46	Selecting Patients with Ovarian Cancer for Germline BRCA Mutation Testing: Findings from Guidelines and a Systematic Literature Review. <i>Advances in Therapy</i> , 2016, 33, 129-150.	1.3	46
47	Attitudes Toward Prenatal Genetic Testing in Patients With Familial Adenomatous Polyposis. <i>American Journal of Gastroenterology</i> , 2007, 102, 1284-1290.	0.2	45
48	Role of MDH2 pathogenic variant in pheochromocytoma and paraganglioma patients. <i>Genetics in Medicine</i> , 2018, 20, 1652-1662.	1.1	45
49	BRCA1 intronic Alu elements drive gene rearrangements and PARP inhibitor resistance. <i>Nature Communications</i> , 2019, 10, 5661.	5.8	45
50	Analysis of PALB2 Gene in BRCA1/BRCA2 Negative Spanish Hereditary Breast/Ovarian Cancer Families with Pancreatic Cancer Cases. <i>PLoS ONE</i> , 2013, 8, e67538.	1.1	44
51	The Molecular Tumor Board Portal supports clinical decisions and automated reporting for precision oncology. <i>Nature Cancer</i> , 2022, 3, 251-261.	5.7	44
52	Stumbling Blocks on the Path to Personalized Medicine in Breast Cancer: The Case of PARP Inhibitors for <i>BRCA1/2</i> -Associated Cancers. <i>Cancer Discovery</i> , 2011, 1, 29-34.	7.7	43
53	Multicenter Phase II Study of Lurbinectedin in <i>BRCA</i> -Mutated and Unselected Metastatic Advanced Breast Cancer and Biomarker Assessment Substudy. <i>Journal of Clinical Oncology</i> , 2018, 36, 3134-3143.	0.8	43
54	Genetic counseling program in familial breast cancer: Analysis of its effectiveness, cost and cost-effectiveness ratio. <i>International Journal of Cancer</i> , 2004, 112, 647-652.	2.3	41

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55	Performance of PREMM1,2,6, MMRpredict, and MMRpro in detecting Lynch syndrome among endometrial cancer cases. <i>Genetics in Medicine</i> , 2012, 14, 670-680.	1.1	40
56	<i>POLE</i> and <i>POLD1</i> screening in 155 patients with multiple polyps and early-onset colorectal cancer. <i>Oncotarget</i> , 2017, 8, 26732-26743.	0.8	40
57	Preclinical <i>In Vivo</i> Validation of the RAD51 Test for Identification of Homologous Recombination-Deficient Tumors and Patient Stratification. <i>Cancer Research</i> , 2022, 82, 1646-1657.	0.4	40
58	Association of Genomic Domains in <i>BRCA1</i> and <i>BRCA2</i> with Prostate Cancer Risk and Aggressiveness. <i>Cancer Research</i> , 2020, 80, 624-638.	0.4	39
59	Identification of genetic test results with conflicting interpretations in prospective registry of multiplex testing (PROMPT).. <i>Journal of Clinical Oncology</i> , 2016, 34, 1510-1510.	0.8	38
60	Molecular features of the basal-like breast cancer subtype based on BRCA1 mutation status. <i>Breast Cancer Research and Treatment</i> , 2014, 147, 185-191.	1.1	37
61	Factors Associated with Enrollment in Cancer Genetics Research. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2006, 15, 1355-1359.	1.1	36
62	What factors may influence psychological well being at three months and one year post BRCA genetic result disclosure?. <i>Breast</i> , 2012, 21, 755-760.	0.9	36
63	Clinical Subtypes and Molecular Characteristics of Serrated Polyposis Syndrome. <i>Clinical Gastroenterology and Hepatology</i> , 2013, 11, 705-711.	2.4	36
64	SOLTI NeoPARP: a phase II randomized study of two schedules of iniparib plus paclitaxel versus paclitaxel alone as neoadjuvant therapy in patients with triple-negative breast cancer. <i>Breast Cancer Research and Treatment</i> , 2015, 154, 351-357.	1.1	35
65	Management of individuals with germline variants in PALB2: a clinical practice resource of the American College of Medical Genetics and Genomics (ACMG). <i>Genetics in Medicine</i> , 2021, 23, 1416-1423.	1.1	34
66	RNA analysis of eight BRCA1 and BRCA2 unclassified variants identified in breast/ovarian cancer families from Spain. <i>Human Mutation</i> , 2003, 22, 337-337.	1.1	33
67	Uptake of predictive testing among relatives of BRCA1 and BRCA2 families: a multicenter study in northeastern Spain. <i>Familial Cancer</i> , 2010, 9, 297-304.	0.9	32
68	Opportunistic testing of <i>BRCA1</i>, <i>BRCA2</i> and mismatch repair genes improves the yield of phenotype driven hereditary cancer gene panels. <i>International Journal of Cancer</i> , 2019, 145, 2682-2691.	2.3	30
69	<i>MLH1</i> Founder Mutations with Moderate Penetrance in Spanish Lynch Syndrome Families. <i>Cancer Research</i> , 2010, 70, 7379-7391.	0.4	29
70	RAD51C germline mutations found in Spanish site-specific breast cancer and breast-ovarian cancer families. <i>Breast Cancer Research and Treatment</i> , 2014, 147, 133-143.	1.1	29
71	Comparison of Prediction Models for Lynch Syndrome Among Individuals With Colorectal Cancer. <i>Journal of the National Cancer Institute</i> , 2016, 108, .	3.0	29
72	Increased Risk of Colorectal Cancer in Patients With Multiple Serrated Polyps and Their First-Degree Relatives. <i>Gastroenterology</i> , 2017, 153, 106-112.e2.	0.6	28

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73	The FANCM:p.Arg658* truncating variant is associated with risk of triple-negative breast cancer. <i>Npj Breast Cancer</i> , 2019, 5, 38.	2.3	28
74	International distribution and age estimation of the Portuguese BRCA2 c.156_157insAlu founder mutation. <i>Breast Cancer Research and Treatment</i> , 2011, 127, 671-679.	1.1	27
75	<i>BRCA1</i> and <i>BRCA2</i> pathogenic sequence variants in women of African origin or ancestry. <i>Human Mutation</i> , 2019, 40, 1781-1796.	1.1	26
76	Screening of <i>BRCA1/2</i> deep intronic regions by targeted gene sequencing identifies the first germline <i>BRCA1</i> variant causing pseudoexon activation in a patient with breast/ovarian cancer. <i>Journal of Medical Genetics</i> , 2019, 56, 63-74.	1.5	26
77	Germline mutations in NF1 and BRCA1 in a family with neurofibromatosis type 1 and early-onset breast cancer. <i>Breast Cancer Research and Treatment</i> , 2013, 139, 597-602.	1.1	25
78	Genetic counselling and testing of susceptibility genes for therapeutic decision-making in breast cancer—an European consensus statement and expert recommendations. <i>European Journal of Cancer</i> , 2019, 106, 54-60.	1.3	25
79	Niraparib for Advanced Breast Cancer with Germline <i>BRCA1</i> and <i>BRCA2</i> Mutations: the EORTC 1307-BCG/BIG5-13/TESARO PR-30-50-10-C BRAVO Study. <i>Clinical Cancer Research</i> , 2021, 27, 5482-5491.	3.2	25
80	About 1% of the breast and ovarian Spanish families testing negative for <i>BRCA1</i> and <i>BRCA2</i> are carriers of <i>RAD51D</i> pathogenic variants. <i>International Journal of Cancer</i> , 2014, 134, 2088-2097.	2.3	24
81	Heterogeneous prevalence of recurrent BRCA1 and BRCA2 mutations in Spain according to the geographical area: implications for genetic testing. <i>Familial Cancer</i> , 2010, 9, 187-191.	0.9	23
82	Challenges to the Development of New Agents for Molecularly Defined Patient Subsets: Lessons From <i>BRCA1/2</i>-Associated Breast Cancer. <i>Journal of Clinical Oncology</i> , 2011, 29, 4224-4226.	0.8	23
83	Mammographic density and breast cancer in women from high risk families. <i>Breast Cancer Research</i> , 2015, 17, 93.	2.2	22
84	Overview of hereditary breast and ovarian cancer (HBOC) guidelines across Europe. <i>European Journal of Medical Genetics</i> , 2021, 64, 104350.	0.7	22
85	Antitumor activity of the poly(ADP-ribose) polymerase inhibitor rucaparib as monotherapy in patients with platinum-sensitive, relapsed, <i>BRCA</i>-mutated, high-grade ovarian cancer, and an update on safety. <i>International Journal of Gynecological Cancer</i> , 2019, 29, 1396-1404.	1.2	19
86	Breast and Prostate Cancer Risks for Male <i>BRCA1</i> and <i>BRCA2</i> Pathogenic Variant Carriers Using Polygenic Risk Scores. <i>Journal of the National Cancer Institute</i> , 2022, 114, 109-122.	3.0	19
87	Haplotype analysis of the BRCA29254delATCAT recurrent mutation in breast/ovarian cancer families from Spain. <i>Human Mutation</i> , 2003, 21, 452-452.	1.1	18
88	Comprehensive genetic and endoscopic evaluation may be necessary to distinguish sporadic versus familial adenomatous polyposis-associated abdominal desmoid tumors. <i>Surgery</i> , 2004, 135, 683-689.	1.0	18
89	Germline BRCA testing is moving from cancer risk assessment to a predictive biomarker for targeting cancer therapeutics. <i>Clinical and Translational Oncology</i> , 2016, 18, 981-987.	1.2	18
90	Optimised molecular genetic diagnostics of Fanconi anaemia by whole exome sequencing and functional studies. <i>Journal of Medical Genetics</i> , 2020, 57, 258-268.	1.5	18

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91	Chromosome 12p Amplification in Triple-Negative/BRCA1-Mutated Breast Cancer Associates with Emergence of Docetaxel Resistance and Carboplatin Sensitivity. <i>Cancer Research</i> , 2019, 79, 4258-4270.	0.4	17
92	Role of Splicing Regulatory Elements and In Silico Tools Usage in the Identification of Deep Intronic Splicing Variants in Hereditary Breast/Ovarian Cancer Genes. <i>Cancers</i> , 2021, 13, 3341.	1.7	17
93	Olaparib monotherapy in patients with advanced cancer and a germ-line BRCA1/2 mutation: An open-label phase II study. <i>Journal of Clinical Oncology</i> , 2013, 31, 11024-11024.	0.8	17
94	Prediction models in Lynch syndrome. <i>Familial Cancer</i> , 2013, 12, 217-228.	0.9	16
95	Activity of HSP90 Inhibitor in a Metastatic Lung Cancer Patient With a Germline BRCA1 Mutation. <i>Journal of the National Cancer Institute</i> , 2018, 110, 914-917.	3.0	16
96	Association of premenopausal risk-reducing salpingo-oophorectomy with breast cancer risk in BRCA1/2 mutation carriers: Maximising bias-reduction. <i>European Journal of Cancer</i> , 2020, 132, 53-60.	1.3	16
97	Caution Should Be Used When Interpreting Alterations Affecting the Exon 3 of the BRCA2 Gene in Breast/Ovarian Cancer Families. <i>Journal of Clinical Oncology</i> , 2007, 25, 5035-5036.	0.8	15
98	Analysis of Lynch Syndrome Mismatch Repair Genes in Women with Endometrial Cancer. <i>Oncology</i> , 2016, 91, 171-176.	0.9	15
99	European Breast Cancer Council manifesto 2018: Genetic risk prediction testing in breast cancer. <i>European Journal of Cancer</i> , 2019, 106, 45-53.	1.3	15
100	Comprehensive Constitutional Genetic and Epigenetic Characterization of Lynch-Like Individuals. <i>Cancers</i> , 2020, 12, 1799.	1.7	15
101	Quality of life with talazoparib after platinum or multiple cytotoxic non-platinum regimens in patients with advanced breast cancer and germline BRCA1/2 mutations: patient-reported outcomes from the ABRAZO phase 2 trial. <i>European Journal of Cancer</i> , 2018, 104, 160-168.	1.3	14
102	Chromosome fragility in the buccal epithelium in patients with Fanconi anemia. <i>Cancer Letters</i> , 2020, 472, 1-7.	3.2	14
103	Characterization of four novel BRCA2 large genomic rearrangements in Spanish breast/ovarian cancer families: review of the literature, and reevaluation of the genetic mechanisms involved in their origin. <i>Breast Cancer Research and Treatment</i> , 2012, 133, 273-283.	1.1	13
104	Clinical behavior and outcomes of breast cancer in young women with germline BRCA pathogenic variants. <i>Npj Breast Cancer</i> , 2021, 7, 16.	2.3	13
105	Evidence for a link between TNFRSF11A and risk of breast cancer. <i>Breast Cancer Research and Treatment</i> , 2011, 129, 947-954.	1.1	12
106	A phase II open-label, multicenter study of single-agent rucaparib in the treatment of patients with relapsed ovarian cancer and a deleterious BRCA mutation. <i>Journal of Clinical Oncology</i> , 2015, 33, 5513-5513.	0.8	12
107	Incorporation of semi-quantitative analysis of splicing alterations for the clinical interpretation of variants in BRCA1 and BRCA2 genes. <i>Human Mutation</i> , 2019, 40, 2296-2317.	1.1	11
108	BARD1 Pathogenic Variants Are Associated with Triple-Negative Breast Cancer in a Spanish Hereditary Breast and Ovarian Cancer Cohort. <i>Genes</i> , 2021, 12, 150.	1.0	11

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109	Data reduction for prediction: A case study on robust coding of age and family history for the risk of having a genetic mutation. <i>Statistics in Medicine</i> , 2007, 26, 5545-5556.	0.8	10
110	Detection of the CHEK2 1100delC mutation by MLPA BRCA1/2 analysis: a worthwhile strategy for its clinical applicability in 1100delC low-frequency populations?. <i>Breast Cancer Research and Treatment</i> , 2008, 107, 455-457.	1.1	10
111	Evolving Landscape of Molecular Prescreening Strategies for Oncology Early Clinical Trials. <i>JCO Precision Oncology</i> , 2020, 4, 505-513.	1.5	10
112	Paired Somatic-Germline Testing of 15 Polyposis and Colorectal Cancer Predisposing Genes Highlights the Role of APC Mosaicism in de Novo Familial Adenomatous Polyposis. <i>Journal of Molecular Diagnostics</i> , 2021, 23, 1452-1459.	1.2	10
113	A novel de novo BRCA2 mutation of paternal origin identified in a Spanish woman with early onset bilateral breast cancer. <i>Breast Cancer Research and Treatment</i> , 2010, 121, 221-225.	1.1	9
114	Novel BRCA1 deleterious mutation (c.1949_1950delTA) in a woman of Senegalese descent with triple-negative early-onset breast cancer. <i>Oncology Letters</i> , 2011, 2, 1287-1289.	0.8	9
115	The variants BRCA1 IVS6-1G>A and BRCA2 IVS15+1G>A lead to aberrant splicing of the transcripts. <i>Breast Cancer Research and Treatment</i> , 2009, 117, 461-465.	1.1	8
116	A phase III randomized trial of niraparib versus physician's choice in previously treated, HER2-negative, germline-BRCA mutated breast cancer patients: Intergroup study EORTC-1307-BCG and BIG5-13.. <i>Journal of Clinical Oncology</i> , 2014, 32, TPS659-TPS659.	0.8	7
117	Germline ATM mutational analysis in BRCA1/BRCA2 negative hereditary breast cancer families by MALDI-TOF mass spectrometry. <i>Breast Cancer Research and Treatment</i> , 2011, 128, 573-579.	1.1	6
118	Mutation analysis of the SHFM1 gene in breast/ovarian cancer families. <i>Journal of Cancer Research and Clinical Oncology</i> , 2013, 139, 529-532.	1.2	6
119	Alternative transcript imbalance underlying breast cancer susceptibility in a family carrying PALB2 c.3201+5G>T. <i>Breast Cancer Research and Treatment</i> , 2019, 174, 543-550.	1.1	6
120	Phase I, open-label study of olaparib plus cisplatin in patients with advanced solid tumors.. <i>Journal of Clinical Oncology</i> , 2012, 30, 1009-1009.	0.8	6
121	BRIP1 as an ovarian cancer susceptibility gene: ready for the clinic?. <i>Journal of the National Cancer Institute</i> , 2015, 107, djv262.	3.0	5
122	SOLTI NeoPARP: A phase II, randomized study of two schedules of niraparib plus paclitaxel and paclitaxel alone as neoadjuvant therapy in patients with triple-negative breast cancer (TNBC).. <i>Journal of Clinical Oncology</i> , 2012, 30, 1011-1011.	0.8	5
123	Sex Ratio Distortion in Offspring of Families with BRCA1 or BRCA2 Mutant Alleles: An Ascertainment Bias Phenomenon?. <i>Breast Cancer Research and Treatment</i> , 2005, 92, 273-277.	1.1	4
124	Mutation analysis of the BCCIP gene for breast cancer susceptibility in breast/ovarian cancer families. <i>Gynecologic Oncology</i> , 2013, 131, 460-463.	0.6	4
125	BRCA1 and BRCA2 whole cDNA analysis in unsolved hereditary breast/ovarian cancer patients. <i>Cancer Genetics</i> , 2021, 258-259, 10-17.	0.2	4
126	Clinical consequences of BRCA2 hypomorphism. <i>Npj Breast Cancer</i> , 2021, 7, 117.	2.3	3



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127	PI3K pathway (PI3Kp) dysregulation and response to pan-PI3K/AKT/mTOR/dual PI3K-mTOR inhibitors (PI3Kpi) in metastatic breast cancer (MBC) patients (pts).. Journal of Clinical Oncology, 2012, 30, 509-509.	0.8	3
128	Prospective registry of multiplex testing (PROMPT): A web-based platform to assess cancer risk of genetic variants.. Journal of Clinical Oncology, 2016, 34, 1518-1518.	0.8	3
129	Prospective Registry of Multiplex Testing (PROMPT): Feasible and sustainable.. Journal of Clinical Oncology, 2018, 36, 1543-1543.	0.8	3
130	Screening for Lynch Syndrome among Patients with Newly Diagnosed Endometrial Cancer: A Comprehensive Review. Tumori, 2016, 102, P1-7P.	0.6	2
131	Prevalence of mutations in BRCA and homologous recombination repair genes and real-world standard of care of Asian patients with HER2-negative metastatic breast cancer starting first-line systemic cytotoxic chemotherapy: subgroup analysis of the global BREAKOUT study. Breast Cancer, 2022, 29, 92-102.	1.3	2
132	A phase 2 study (2-stage, 2-cohort) of the oral PARP inhibitor talazoparib (BMN 673) in patients with germline BRCA mutation and locally advanced and/or metastatic breast cancer (ABRAZO).. Journal of Clinical Oncology, 2015, 33, TPS1108-TPS1108.	0.8	2
133	Efficacy and safety of olaparib monotherapy in a subgroup of patients with a germline BRCA1/2 mutation and advanced ovarian cancer from a Phase II open-label study.. Journal of Clinical Oncology, 2015, 33, 5529-5529.	0.8	2
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