Judith Balmaña

List of Publications by Year in descending order

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53660 39575 10,028 150 45 94 citations h-index g-index papers 157 157 157 13814 docs citations times ranked citing authors all docs

#	Article	IF	CITATIONS
1	Breast and Prostate Cancer Risks for Male <i>BRCA1</i> Alond <i>BRCA2</i> Pathogenic Variant Carriers Using Polygenic Risk Scores. Journal of the National Cancer Institute, 2022, 114, 109-122.	3.0	19
2	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
3	Abstract PD5-06: Safety of assisted reproductive technologies (ART) following treatment completion in young women with germline <i>BRCA</i> pathogenic variants having a pregnancy after breast cancer. Cancer Research, 2022, 82, PD5-06-PD5-06.	0.4	O
4	The Molecular Tumor Board Portal supports clinical decisions and automated reporting for precision oncology. Nature Cancer, 2022, 3, 251-261.	5.7	44
5	Abstract OT2-24-02: ZEST: Randomized phase III study evaluating efficacy and safety of niraparib in patients with HER2-negative BRCA-mutated or triple-negative breast cancer with detectable circulating tumor DNA after definitive therapy. Cancer Research, 2022, 82, OT2-24-02-OT2-24-02.	0.4	2
6	Preclinical <i>In Vivo</i> Validation of the RAD51 Test for Identification of Homologous Recombination-Deficient Tumors and Patient Stratification. Cancer Research, 2022, 82, 1646-1657.	0.4	40
7	Current Systemic Treatments for the Hereditary Cancer Syndromes: Drug Development in Light of Genomic Defects. American Society of Clinical Oncology Educational Book / ASCO American Society of Clinical Oncology Meeting, 2022, , 808-824.	1.8	2
8	BARD1 Pathogenic Variants Are Associated with Triple-Negative Breast Cancer in a Spanish Hereditary Breast and Ovarian Cancer Cohort. Genes, 2021, 12, 150.	1.0	11
9	Clinical behavior and outcomes of breast cancer in young women with germline BRCA pathogenic variants. Npj Breast Cancer, 2021, 7, 16.	2.3	13
10	Patients' and professionals' perspective of non-in-person visits in hereditary cancer: predictors and impact of the COVID-19 pandemic. Genetics in Medicine, 2021, 23, 1450-1457.	1.1	1
11	Management of individuals with germline variants in PALB2: a clinical practice resource of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2021, 23, 1416-1423.	1.1	34
12	Adjuvant Olaparib for Patients with <i>BRCA1</i> - or <i>BRCA2</i> -Mutated Breast Cancer. New England Journal of Medicine, 2021, 384, 2394-2405.	13.9	764
13	Role of Splicing Regulatory Elements and In Silico Tools Usage in the Identification of Deep Intronic Splicing Variants in Hereditary Breast/Ovarian Cancer Genes. Cancers, 2021, 13, 3341.	1.7	17
14	Niraparib for Advanced Breast Cancer with Germline <i>BRCA1</i> and <i>BRCA2</i> Mutations: the EORTC 1307-BCG/BIG5â€"13/TESARO PR-30â€"50â€"10-C BRAVO Study. Clinical Cancer Research, 2021, 27, 5482-5491.	3.2	25
15	Paired Somatic-Germline Testing of 15 Polyposis and Colorectal Cancer–Predisposing Genes Highlights the Role of APC Mosaicism in de Novo Familial Adenomatous Polyposis. Journal of Molecular Diagnostics, 2021, 23, 1452-1459.	1.2	10
16	Clinical consequences of BRCA2 hypomorphism. Npj Breast Cancer, 2021, 7, 117.	2.3	3
17	BRCA1 and BRCA2 whole cDNA analysis in unsolved hereditary breast/ovarian cancer patients. Cancer Genetics, 2021, 258-259, 10-17.	0.2	4
18	Overview of hereditary breast and ovarian cancer (HBOC) guidelines across Europe. European Journal of Medical Genetics, 2021, 64, 104350.	0.7	22

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19	Association of Genomic Domains in <i>BRCA1</i> and <i>BRCA2</i> with Prostate Cancer Risk and Aggressiveness. Cancer Research, 2020, 80, 624-638.	0.4	39
20	Cancer Risks Associated With Germline <i>PALB2</i> Pathogenic Variants: An International Study of 524 Families. Journal of Clinical Oncology, 2020, 38, 674-685.	0.8	270
21	Chromosome fragility in the buccal epithelium in patients with Fanconi anemia. Cancer Letters, 2020, 472, 1-7.	3.2	14
22	Comprehensive Constitutional Genetic and Epigenetic Characterization of Lynch-Like Individuals. Cancers, 2020, 12, 1799.	1.7	15
23	Role of POLE and POLD1 in familial cancer. Genetics in Medicine, 2020, 22, 2089-2100.	1.1	76
24	Evolving Landscape of Molecular Prescreening Strategies for Oncology Early Clinical Trials. JCO Precision Oncology, 2020, 4, 505-513.	1.5	10
25	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.	3.0	106
26	Optimised molecular genetic diagnostics of Fanconi anaemia by whole exome sequencing and functional studies. Journal of Medical Genetics, 2020, 57, 258-268.	1.5	18
27	Association of premenopausal risk-reducing salpingo-oophorectomy with breast cancer risk in BRCA1/2 mutation carriers: Maximising bias-reduction. European Journal of Cancer, 2020, 132, 53-60.	1.3	16
28	Incorporation of semiâ€quantitative analysis of splicing alterations for the clinical interpretation of variants in <i>BRCA1</i> BRCA2Senes. Human Mutation, 2019, 40, 2296-2317.	1.1	11
29	The FANCM:p.Arg658* truncating variant is associated with risk of triple-negative breast cancer. Npj Breast Cancer, 2019, 5, 38.	2.3	28
30	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.	1.1	102
31	Chromosome 12p Amplification in Triple-Negative/ <i>BRCA1-</i> Nutated Breast Cancer Associates with Emergence of Docetaxel Resistance and Carboplatin Sensitivity. Cancer Research, 2019, 79, 4258-4270.	0.4	17
32	Controversies in oncology: are genomic tests quantifying homologous recombination repair deficiency (HRD) useful for treatment decision making? ESMO Open, 2019, 4, e000480.	2.0	47
33	<i>BRCA1</i> and <i>BRCA2</i> pathogenic sequence variants in women of African origin or ancestry. Human Mutation, 2019, 40, 1781-1796.	1.1	26
34	Moving From Poly (ADP-Ribose) Polymerase Inhibition to Targeting DNA Repair and DNA Damage Response in Cancer Therapy. Journal of Clinical Oncology, 2019, 37, 2257-2269.	0.8	135
35	Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. Nature Communications, 2019, 10, 1741.	5. 8	90
36	Opportunistic testing of <i>BRCA1</i> , <i>BRCA2</i> and mismatch repair genes improves the yield of phenotype driven hereditary cancer gene panels. International Journal of Cancer, 2019, 145, 2682-2691.	2.3	30

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37	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. International Journal of Gynecological Cancer, 2019, 29, 1396-1404.	1.2	19
38	BRCA1 intronic Alu elements drive gene rearrangements and PARP inhibitor resistance. Nature Communications, 2019, 10, 5661.	5.8	45
39	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). Clinical Cancer Research, 2019, 25, 2717-2724.	3.2	102
40	Alternative transcript imbalance underlying breast cancer susceptibility in a family carrying PALB2 c.3201+5G>T. Breast Cancer Research and Treatment, 2019, 174, 543-550.	1.1	6
41	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. Journal of Medical Genetics, 2019, 56, 63-74.	1.5	26
42	European Breast Cancer Council manifesto 2018: GeneticÂrisk prediction testing in breast cancer. European Journal of Cancer, 2019, 106, 45-53.	1.3	15
43	Genetic counselling and testing of susceptibility genes for therapeutic decision-making in breast cancer—an European consensus statement and expert recommendations. European Journal of Cancer, 2019, 106, 54-60.	1.3	25
44	Prospective Registry of Multiplex Testing (PROMPT): Follow-up Journal of Clinical Oncology, 2019, 37, 1527-1527.	0.8	1
45	Activity of HSP90 Inhibiton in a Metastatic Lung Cancer Patient With a Germline BRCA1 Mutation. Journal of the National Cancer Institute, 2018, 110, 914-917.	3.0	16
46	RAD51 foci as a functional biomarker of homologous recombination repair and PARP inhibitor resistance in germline BRCA-mutated breast cancer. Annals of Oncology, 2018, 29, 1203-1210.	0.6	280
47	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.	1.1	224
48	Multicenter Phase II Study of Lurbinectedin in <i>BRCA</i> -Mutated and Unselected Metastatic Advanced Breast Cancer and Biomarker Assessment Substudy. Journal of Clinical Oncology, 2018, 36, 3134-3143.	0.8	43
49	Computational Tools for Splicing Defect Prediction in Breast/Ovarian Cancer Genes: How Efficient Are They at Predicting RNA Alterations?. Frontiers in Genetics, 2018, 9, 366.	1.1	53
50	Multigene panel testing beyond BRCA1/2 in breast/ovarian cancer Spanish families and clinical actionability of findings. Journal of Cancer Research and Clinical Oncology, 2018, 144, 2495-2513.	1.2	53
51	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. European Journal of Cancer, 2018, 104, 160-168.	1.3	14
52	A <scp>RAD</scp> 51 assay feasible in routine tumor samples calls <scp>PARP</scp> inhibitor response beyond <scp>BRCA</scp> mutation. EMBO Molecular Medicine, 2018, 10, .	3.3	169
53	Screening and surveillance in hereditary gastrointestinal cancers: Recommendations from the European Society of Digestive Oncology (ESDO)Âexpert discussion at the 20th European Society for Medical Oncology (ESMO)/World Congress on Gastrointestinal Cancer, Barcelona, June 2018. European lournal of Cancer. 2018. 104. 91-103.	1.3	60
54	Surveillance Guidelines for Hereditary Colorectal Cancer Syndromes. , 2018, , 305-326.		0

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55	Role of MDH2 pathogenic variant in pheochromocytoma and paraganglioma patients. Genetics in Medicine, 2018, 20, 1652-1662.	1.1	45
56	Prospective Registry of Multiplex Testing (PROMPT): Feasible and sustainable Journal of Clinical Oncology, 2018, 36, 1543-1543.	0.8	3
57	Increased Risk of Colorectal Cancer in Patients With Multiple Serrated Polyps and Their First-Degree Relatives. Gastroenterology, 2017, 153, 106-112.e2.	0.6	28
58	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. Nature Genetics, 2017, 49, 680-691.	9.4	356
59	A Phase l–II Study of the Oral PARP Inhibitor Rucaparib in Patients with Germline <i>BRCA1/2</i> -Mutated Ovarian Carcinoma or Other Solid Tumors. Clinical Cancer Research, 2017, 23, 4095-4106.	3.2	213
60	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. Nature Genetics, 2017, 49, 1767-1778.	9.4	289
61	<i>POLE</i> and <i>POLD1</i> screening in 155 patients with multiple polyps and early-onset colorectal cancer. Oncotarget, 2017, 8, 26732-26743.	0.8	40
62	Reply to R.L. Nussbaum et al and J.S. Dolinsky et al. Journal of Clinical Oncology, 2017, 35, 1262-1263.	0.8	1
63	Prediction of Breast and Prostate Cancer Risks in Male <i>BRCA1</i> and <i>BRCA2</i> Mutation Carriers Using Polygenic Risk Scores. Journal of Clinical Oncology, 2017, 35, 2240-2250.	0.8	152
64	The BRCA1-Î"11q Alternative Splice Isoform Bypasses Germline Mutations and Promotes Therapeutic Resistance to PARP Inhibition and Cisplatin. Cancer Research, 2016, 76, 2778-2790.	0.4	208
65	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
66	Conflicting Interpretation of Genetic Variants and Cancer Risk by Commercial Laboratories as Assessed by the Prospective Registry of Multiplex Testing. Journal of Clinical Oncology, 2016, 34, 4071-4078.	0.8	147
67	CDK12 Inhibition Reverses De Novo and Acquired PARP Inhibitor Resistance in BRCA Wild-Type and Mutated Models of Triple-Negative Breast Cancer. Cell Reports, 2016, 17, 2367-2381.	2.9	215
68	Analysis of Lynch Syndrome Mismatch Repair Genes in Women with Endometrial Cancer. Oncology, 2016, 91, 171-176.	0.9	15
69	Screening for Lynch Syndrome among Patients with Newly Diagnosed Endometrial Cancer: A Comprehensive Review. Tumori, 2016, 102, P1-7P.	0.6	2
70	Selecting Patients with Ovarian Cancer for Germline BRCA Mutation Testing: Findings from Guidelines and a Systematic Literature Review. Advances in Therapy, 2016, 33, 129-150.	1.3	46
71	Efficacy and safety of olaparib monotherapy in germline BRCA1 / 2 mutation carriers with advanced ovarian cancer and three or more lines of prior therapy. Gynecologic Oncology, 2016, 140, 199-203.	0.6	252
72	Germline BRCA testing is moving from cancer risk assessment to a predictive biomarker for targeting cancer therapeutics. Clinical and Translational Oncology, 2016, 18, 981-987.	1.2	18

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73	Comparison of Prediction Models for Lynch Syndrome Among Individuals With Colorectal Cancer. Journal of the National Cancer Institute, 2016, 108, .	3.0	29
74	BRCA1185delAG tumors may acquire therapy resistance through expression of RING-less BRCA1. Journal of Clinical Investigation, 2016, 126, 2903-2918.	3.9	105
75	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
76	Identification of genetic test results with conflicting interpretations in prospective registry of multiplex testing (PROMPT) Journal of Clinical Oncology, 2016, 34, 1510-1510.	0.8	38
77	Mammographic density and breast cancer in women from high risk families. Breast Cancer Research, 2015, 17, 93.	2.2	22
78	PARP inhibitors in ovarian cancer. British Journal of Cancer, 2015, 113, S1-S2.	2.9	1
79	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. Breast Cancer Research and Treatment, 2015, 154, 351-357.	1.1	35
80	Olaparib Monotherapy in Patients With Advanced Cancer and a Germline <i>BRCA1/2</i> Mutation. Journal of Clinical Oncology, 2015, 33, 244-250.	0.8	1,473
81	<i>FANCM</i> c.5791C>T nonsense mutation (rs144567652) induces exon skipping, affects DNA repair activity and is a familial breast cancer risk factor. Human Molecular Genetics, 2015, 24, 5345-5355.	1.4	91
82	BRIP1as an ovarian cancer susceptibility gene: ready for the clinic?. Journal of the National Cancer Institute, 2015, 107, djv262.	3.0	5
83	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 Journal of Clinical Oncology, 2015, 33, 5513-5513.	0.8	12
84	New contributions in the process of breast cancer patient participation in clinical trials in Spain: A qualitative multicenter study Journal of Clinical Oncology, 2015, 33, e20570-e20570.	0.8	0
85	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
86	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
87	Multiplex testing in high risk BRCA1/2-negative families Journal of Clinical Oncology, 2015, 33, e12557-e12557.	0.8	0
88	Prevalence and Characteristics of <i>MUTYH</i> -Associated Polyposis in Patients with Multiple Adenomatous and Serrated Polyps. Clinical Cancer Research, 2014, 20, 1158-1168.	3.2	57
89	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. International Journal of Cancer, 2014, 134, 2088-2097.	2.3	24
90	Prevalence of germline MUTYH mutations among Lynch-like syndrome patients. European Journal of Cancer, 2014, 50, 2241-2250.	1.3	66

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91	Molecular features of the basal-like breast cancer subtype based on BRCA1 mutation status. Breast Cancer Research and Treatment, 2014, 147, 185-191.	1.1	37
92	RAD51C germline mutations found in Spanish site-specific breast cancer and breast-ovarian cancer families. Breast Cancer Research and Treatment, 2014, 147, 133-143.	1.1	29
93	Phase I trial of olaparib in combination with cisplatin for the treatment of patients with advanced breast, ovarian and other solid tumors. Annals of Oncology, 2014, 25, 1656-1663.	0.6	144
94	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 Journal of Clinical Oncology, 2014, 32, TPS659-TPS659.	0.8	7
95	The Role of Platinum Compounds for the Treatment of Breast Cancer. Current Breast Cancer Reports, 2013, 5, 11-22.	0.5	1
96	Mutation analysis of the SHFM1 gene in breast/ovarian cancer families. Journal of Cancer Research and Clinical Oncology, 2013, 139, 529-532.	1.2	6
97	Prediction models in Lynch syndrome. Familial Cancer, 2013, 12, 217-228.	0.9	16
98	Germline mutations in NF1 and BRCA1 in a family with neurofibromatosis type 1 and early-onset breast cancer. Breast Cancer Research and Treatment, 2013, 139, 597-602.	1.1	25
99	Clinical Subtypes and Molecular Characteristics of Serrated Polyposis Syndrome. Clinical Gastroenterology and Hepatology, 2013, 11, 705-711.	2.4	36
100	Mutation analysis of the BCCIP gene for breast cancer susceptibility in breast/ovarian cancer families. Gynecologic Oncology, 2013, 131, 460-463.	0.6	4
101	A 10-year step forward in hereditary cancer in Spain. Clinical and Translational Oncology, 2013, 15, 1-2.	1.2	1
102	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
103	Comparison of the clinical prediction model PREMM _{1,2,6} and molecular testing for the systematic identification of Lynch syndrome in colorectal cancer. Gut, 2013, 62, 272-279.	6.1	49
104	Analysis of PALB2 Gene in BRCA1/BRCA2 Negative Spanish Hereditary Breast/Ovarian Cancer Families with Pancreatic Cancer Cases. PLoS ONE, 2013, 8, e67538.	1.1	44
105	Olaparib monotherapy in patients with advanced cancer and a germ-line <i>BRCA1/2</i> mutation: An open-label phase II study Journal of Clinical Oncology, 2013, 31, 11024-11024.	0.8	17
106	Preliminary analysis of risk factors associated with peritoneal carcinomatosis (PC) after prophylactic bilateral salpingoophorectomy (PBSO) in patients with a BRCA mutation Journal of Clinical Oncology, 2013, 31, 1509-1509.	0.8	0
107	Performance of PREMM1,2,6, MMRpredict, and MMRpro in detecting Lynch syndrome among endometrial cancer cases. Genetics in Medicine, 2012, 14, 670-680.	1.1	40
108	Combining a PI3K Inhibitor with a PARP Inhibitor Provides an Effective Therapy for BRCA1-Related Breast Cancer. Cancer Discovery, 2012, 2, 1048-1063.	7.7	384

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109	What factors may influence psychological well being at three months and one year post BRCA genetic result disclosure?. Breast, 2012, 21, 755-760.	0.9	36
110	Characterization of BRCA1 and BRCA2 splicing variants: a collaborative report by ENIGMA consortium members. Breast Cancer Research and Treatment, 2012, 132, 1009-1023.	1.1	56
111	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. Breast Cancer Research and Treatment, 2012, 133, 273-283.	1.1	13
112	Breast cancer phenotype in women with TP53 germline mutations: a Li-Fraumeni syndrome consortium effort. Breast Cancer Research and Treatment, 2012, 133, 1125-1130.	1.1	144
113	Detection of a large rearrangement in PALB2 in Spanish breast cancer families with male breast cancer. Breast Cancer Research and Treatment, 2012, 132, 307-315.	1.1	50
114	Phase I, open-label study of olaparib plus cisplatin in patients with advanced solid tumors Journal of Clinical Oncology, 2012, 30, 1009-1009.	0.8	6
115	SOLTI NeoPARP: A phase II, randomized study of two schedules of iniparib plus paclitaxel and paclitaxel alone as neoadjuvant therapy in patients with triple-negative breast cancer (TNBC) Journal of Clinical Oncology, 2012, 30, 1011-1011.	0.8	5
116	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
117	Presentation and treatment of HER2-positive metastatic breast cancer patients already treated with adjuvant trastuzumab Journal of Clinical Oncology, 2012, 30, 619-619.	0.8	0
118	Prognostic significance of PI3K pathway (PI3Kp) dysregulation in metastatic breast cancer (MBC) patients (pts) Journal of Clinical Oncology, 2012, 30, 566-566.	0.8	0
119	Stumbling Blocks on the Path to Personalized Medicine in Breast Cancer: The Case of PARP Inhibitors for <i>BRCA1/2</i> -Associated Cancers. Cancer Discovery, 2011, 1, 29-34.	7.7	43
120	The PREMM1,2,6 Model Predicts Risk of MLH1, MSH2, and MSH6 Germline Mutations Based on Cancer History. Gastroenterology, 2011, 140, 73-81.e5.	0.6	171
121	Novel BRCA1 deleterious mutation (c.1949_1950delTA) in a woman of Senegalese descent with triple-negative early-onset breast cancer. Oncology Letters, 2011, 2, 1287-1289.	0.8	9
122	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
123	Germline ATM mutational analysis in BRCA1/BRCA2 negative hereditary breast cancer families by MALDI-TOF mass spectrometry. Breast Cancer Research and Treatment, 2011, 128, 573-579.	1.1	6
124	Evidence for a link between TNFRSF11A and risk of breast cancer. Breast Cancer Research and Treatment, 2011, 129, 947-954.	1.1	12
125	Challenges to the Development of New Agents for Molecularly Defined Patient Subsets: Lessons From <i>BRCA1/2</i> -Associated Breast Cancer. Journal of Clinical Oncology, 2011, 29, 4224-4226.	0.8	23
126	Parity and the risk of breast and ovarian cancer in BRCA1 and BRCA2 mutation carriers. Breast Cancer Research and Treatment, 2010, 119, 221-232.	1.1	56

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127	A novel de novo BRCA2 mutation of paternal origin identified in a Spanish woman with early onset bilateral breast cancer. Breast Cancer Research and Treatment, 2010, 121, 221-225.	1.1	9
128	Local therapy in BRCA1 and BRCA2 mutation carriers with operable breast cancer: comparison of breast conservation and mastectomy. Breast Cancer Research and Treatment, 2010, 121, 389-398.	1.1	170
129	Heterogeneous prevalence of recurrent BRCA1 and BRCA2 mutations in Spain according to the geographical area: implications for genetic testing. Familial Cancer, 2010, 9, 187-191.	0.9	23
130	Uptake of predictive testing among relatives of BRCA1 and BRCA2 families: a multicenter study in northeastern Spain. Familial Cancer, 2010, 9, 297-304.	0.9	32
131	<i>MLH1</i> Founder Mutations with Moderate Penetrance in Spanish Lynch Syndrome Families. Cancer Research, 2010, 70, 7379-7391.	0.4	29
132	Development and Validation of a Colon Cancer Risk Assessment Tool for Patients Undergoing Colonoscopy. American Journal of Gastroenterology, 2009, 104, 1508-1518.	0.2	62
133	The variants BRCA1 IVS6-1G>A and BRCA2 IVS15+1G>A lead to aberrant splicing of the transcripts. Breast Cancer Research and Treatment, 2009, 117, 461-465.	1.1	8
134	Detection of the CHEK2 1100delC mutation by MLPA BRCA1/2 analysis: a worthwhile strategy for its clinical applicability in 1100delC low-frequency populations?. Breast Cancer Research and Treatment, 2008, 107, 455-457.	1.1	10
135	Validation and Extension of the PREMM1,2 Model in a Population-Based Cohort of Colorectal Cancer Patients. Gastroenterology, 2008, 134, 39-46.	0.6	57
136	Phenotype Comparison of MLH1 and MSH2 Mutation Carriers in a Cohort of 1,914 Individuals Undergoing Clinical Genetic Testing in the United States. Cancer Epidemiology Biomarkers and Prevention, 2008, 17, 2044-2051.	1.1	65
137	Opinion about reproductive decision making among individuals undergoing BRCA1/2 genetic testing in a multicentre Spanish cohort. Human Reproduction, 2008, 24, 1000-1006.	0.4	61
138	Caution Should Be Used When Interpreting Alterations Affecting the Exon 3 of the BRCA2 Gene in Breast/Ovarian Cancer Families. Journal of Clinical Oncology, 2007, 25, 5035-5036.	0.8	15
139	Attitudes Toward Prenatal Genetic Testing in Patients With Familial Adenomatous Polyposis. American Journal of Gastroenterology, 2007, 102, 1284-1290.	0.2	45
140	Data reduction for prediction: A case study on robust coding of age and family history for the risk of having a genetic mutation. Statistics in Medicine, 2007, 26, 5545-5556.	0.8	10
141	BRAF mutations in colorectal carcinoma suggest two entities of microsatellite-unstable tumors. Cancer, 2006, 106, 2528-2529.	2.0	0
142	Prediction of MLH1 and MSH2 Mutations in Lynch Syndrome. JAMA - Journal of the American Medical Association, 2006, 296, 1469.	3.8	160
143	Factors Associated with Enrollment in Cancer Genetics Research. Cancer Epidemiology Biomarkers and Prevention, 2006, 15, 1355-1359.	1.1	36
144	Sex Ratio Distortion in Offspring of Families with BRCA1 or BRCA2 Mutant Alleles: An Ascertainment Bias Phenomenon?. Breast Cancer Research and Treatment, 2005, 92, 273-277.	1.1	4

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145	Comprehensive genetic and endoscopic evaluation may be necessary to distinguish sporadic versus familial adenomatous polyposis–associated abdominal desmoid tumors. Surgery, 2004, 135, 683-689.	1.0	18
146	Genetic counseling program in familial breast cancer: Analysis of its effectiveness, cost and cost-effectiveness ratio. International Journal of Cancer, 2004, 112, 647-652.	2.3	41
147	Haplotype analysis of the BRCA 29254 del ATCAT recurrent mutation in breast/ovarian cancer families from Spain. Human Mutation, 2003, 21, 452-452.	1.1	18
148	RNA analysis of eightBRCA1andBRCA2unclassified variants identified in breast/ovarian cancer families from Spain. Human Mutation, 2003, 22, 337-337.	1.1	33
149	Microsatellite instability and immunostaining for MSH-2 and MLH-1 in cutaneous and internal tumors from patients with the Muir-Torre syndrome. Journal of Cutaneous Pathology, 2002, 29, 415-420.	0.7	72
150	What is the hereditary non-polyposis colorectal cancer syndrome?., 2000, 2, 191-201.		1