Judith Balmaa

List of Publications by Year in Descending Order

146 papers	6,659 citations	40 h-index	79 g-index
157 ext. papers	8,510 ext. citations	6.4 avg, IF	5.05 L-index

#	Paper	IF	Citations
146	Abstract PD5-06: Safety of assisted reproductive technologies (ART) following treatment completion in young women with germline BRCA pathogenic variants having a pregnancy after breast cancer. *Cancer Research*, **2022**, 82, PD5-06-PD5-06	10.1	
145	The Molecular Tumor Board Portal supports clinical decisions and automated reporting for precision oncology.. *Nature Cancer*, **2022**, 3, 251-261	15.4	3
144	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	10.1	1
143	Preclinical In Vivo Validation of the RAD51 Test for Identification of Homologous Recombination-Deficient Tumors and Patient Stratification.. *Cancer Research*, **2022**, 82, 1646-1657	10.1	4
142	Overview of hereditary breast and ovarian cancer (HBOC) guidelines across Europe. *European Journal of Medical Genetics*, **2021**, 64, 104350	2.6	5
141	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	8.1	
140	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	8.1	5
139	Adjuvant Olaparib for Patients with - or -Mutated Breast Cancer. *New England Journal of Medicine*, **2021**, 384, 2394-2405	59.2	145
138	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,	6.6	3
137	Pathogenic Variants are Associated with Triple-Negative Breast Cancer in a Spanish Hereditary Breast and Ovarian Cancer Cohort. *Genes*, **2021**, 12,	4.2	5
136	Clinical behavior and outcomes of breast cancer in young women with germline BRCA pathogenic variants. *Npj Breast Cancer*, **2021**, 7, 16	7.8	3
135	Niraparib for Advanced Breast Cancer with Germline and Mutations: the EORTC 1307-BCG/BIG5-13/TESARO PR-30-50-10-C BRAVO Study. *Clinical Cancer Research*, **2021**, 27, 5482-5491	12.9	3
134	Breast and Prostate Cancer Risks for Male BRCA1 and BRCA2 Pathogenic Variant Carriers Using Polygenic Risk Scores. *Journal of the National Cancer Institute*, **2021**,	9.7	3
133	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*, **2021**, 1	3.4	
132	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	5.1	0
131	Clinical consequences of BRCA2 hypomorphism. *Npj Breast Cancer*, **2021**, 7, 117	7.8	0
130	BRCA1 and BRCA2 whole cDNA analysis in unsolved hereditary breast/ovarian cancer patients. *Cancer Genetics*, **2021**, 258-259, 10-17	2.3	1

129	Evolving Landscape of Molecular Prescreening Strategies for Oncology Early Clinical Trials. *JCO Precision Oncology*, **2020**, 4,	3.6	4
128	Ovarian and Breast Cancer Risks Associated With Pathogenic Variants in RAD51C and RAD51D. *Journal of the National Cancer Institute*, **2020**, 112, 1242-1250	9.7	51
127	Optimised molecular genetic diagnostics of Fanconi anaemia by whole exome sequencing and functional studies. *Journal of Medical Genetics*, **2020**, 57, 258-268	5.8	12
126	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	7.5	10
125	Association of Genomic Domains in and with Prostate Cancer Risk and Aggressiveness. *Cancer Research*, **2020**, 80, 624-638	10.1	22
124	Cancer Risks Associated With Germline Pathogenic Variants: An International Study of 524 Families. *Journal of Clinical Oncology*, **2020**, 38, 674-685	2.2	133
123	Chromosome fragility in the buccal epithelium in patients with Fanconi anemia. *Cancer Letters*, **2020**, 472, 1-7	9.9	8
122	Comprehensive Constitutional Genetic and Epigenetic Characterization of Lynch-Like Individuals. *Cancers*, **2020**, 12,	6.6	2
121	Role of POLE and POLD1 in familial cancer. *Genetics in Medicine*, **2020**, 22, 2089-2100	8.1	23
120	Large scale multifactorial likelihood quantitative analysis of BRCA1 and BRCA2 variants: An ENIGMA resource to support clinical variant classification. *Human Mutation*, **2019**, 40, 1557-1578	4.7	52
119	Chromosome 12p Amplification in Triple-Negative/Mutated Breast Cancer Associates with Emergence of Docetaxel Resistance and Carboplatin Sensitivity. *Cancer Research*, **2019**, 79, 4258-4270	10.1	6
118	BRCA1 and BRCA2 pathogenic sequence variants in women of African origin or ancestry. *Human Mutation*, **2019**, 40, 1781-1796	4.7	16
117	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	2.2	69
116	Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. *Nature Communications*, **2019**, 10, 1741	17.4	47
115	Opportunistic testing of BRCA1, BRCA2 and mismatch repair genes improves the yield of phenotype driven hereditary cancer gene panels. *International Journal of Cancer*, **2019**, 145, 2682-2691	7.5	21
114	Incorporation of semi-quantitative analysis of splicing alterations for the clinical interpretation of variants in BRCA1 and BRCA2 genes. *Human Mutation*, **2019**, 40, 2296-2317	4.7	6
113	The :p.Arg658* truncating variant is associated with risk of triple-negative breast cancer. *Npj Breast Cancer*, **2019**, 5, 38	7.8	12
112	Prospective Registry of Multiplex Testing (PROMPT): Follow-up.. *Journal of Clinical Oncology*, **2019**, 37, 1527-1527	2.2	0

111	Antitumor activity of the poly(ADP-ribose) polymerase inhibitor rucaparib as monotherapy in patients with platinum-sensitive, relapsed, -mutated, high-grade ovarian cancer, and an update on safety. *International Journal of Gynecological Cancer*, **2019**, 29, 1396-1404	3.5	11
110	BRCA1 intronic Alu elements drive gene rearrangements and PARP inhibitor resistance. *Nature Communications*, **2019**, 10, 5661	17.4	25
109	A Phase II Study of Talazoparib after Platinum or Cytotoxic Nonplatinum Regimens in Patients with Advanced Breast Cancer and Germline Mutations (ABRAZO). *Clinical Cancer Research*, **2019**, 25, 2717-2724	12.9	65
108	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	4.4	5
107	Screening of deep intronic regions by targeted gene sequencing identifies the first germline variant causing pseudoexon activation in a patient with breast/ovarian cancer. *Journal of Medical Genetics*, **2019**, 56, 63-74	5.8	16
106	European Breast Cancer Council manifesto 2018: Genetic risk prediction testing in breast cancer. *European Journal of Cancer*, **2019**, 106, 45-53	7.5	14
105	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	7.5	13
104	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	9.7	12
103	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	10.3	160
102	Mutational spectrum in a worldwide study of 29,700 families with BRCA1 or BRCA2 mutations. *Human Mutation*, **2018**, 39, 593-620	4.7	138
101	Role of MDH2 pathogenic variant in pheochromocytoma and paraganglioma patients. *Genetics in Medicine*, **2018**, 20, 1652-1662	8.1	33
100	Prospective Registry of Multiplex Testing (PROMPT): Feasible and sustainable.. *Journal of Clinical Oncology*, **2018**, 36, 1543-1543	2.2	2
99	Multicenter Phase II Study of Lurbinectedin in BRCA-Mutated and Unselected Metastatic Advanced Breast Cancer and Biomarker Assessment Substudy. *Journal of Clinical Oncology*, **2018**, 36, 3134-3143	2.2	29
98	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	4.5	35
97	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	4.9	31
96	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	7.5	9
95	A RAD51 assay feasible in routine tumor samples calls PARP inhibitor response beyond BRCA mutation. *EMBO Molecular Medicine*, **2018**, 10,	12	85
94	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. *European Journal of Cancer*, **2018**, 104, 91-103	7.5	37

93	Surveillance Guidelines for Hereditary Colorectal Cancer Syndromes **2018**, 305-326		
92	Increased Risk of Colorectal Cancer in Patients With Multiple Serrated Polyps and Their First-Degree Relatives. *Gastroenterology*, **2017**, 153, 106-112.e2	13.3	22
91	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. *Nature Genetics*, **2017**, 49, 680-691	36.3	190
90	A Phase I-II Study of the Oral PARP Inhibitor Rucaparib in Patients with Germline -Mutated Ovarian Carcinoma or Other Solid Tumors. *Clinical Cancer Research*, **2017**, 23, 4095-4106	12.9	164
89	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. *Nature Genetics*, **2017**, 49, 1767-1778	36.3	186
88	POLE and POLD1 screening in 155 patients with multiple polyps and early-onset colorectal cancer. *Oncotarget*, **2017**, 8, 26732-26743	3.3	29
87	Reply to R.L. Nussbaum et al and J.S. Dolinsky et al. *Journal of Clinical Oncology*, **2017**, 35, 1262-1263	2.2	1
86	Prediction of Breast and Prostate Cancer Risks in Male BRCA1 and BRCA2 Mutation Carriers Using Polygenic Risk Scores. *Journal of Clinical Oncology*, **2017**, 35, 2240-2250	2.2	101
85	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	10.6	146
84	Analysis of Lynch Syndrome Mismatch Repair Genes in Women with Endometrial Cancer. *Oncology*, **2016**, 91, 171-6	3.6	12
83	Screening for Lynch syndrome among patients with newly diagnosed endometrial cancer: a comprehensive review. *Tumori*, **2016**, 102, 548-554	1.7	1
82	Selecting Patients with Ovarian Cancer for Germline BRCA Mutation Testing: Findings from Guidelines and a Systematic Literature Review. *Advances in Therapy*, **2016**, 33, 129-50	4.1	37
81	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	4.9	188
80	Germline BRCA testing is moving from cancer risk assessment to a predictive biomarker for targeting cancer therapeutics. *Clinical and Translational Oncology*, **2016**, 18, 981-7	3.6	16
79	Comparison of Prediction Models for Lynch Syndrome Among Individuals With Colorectal Cancer. *Journal of the National Cancer Institute*, **2016**, 108,	9.7	24
78	BRCA1185delAG tumors may acquire therapy resistance through expression of RING-less BRCA1. *Journal of Clinical Investigation*, **2016**, 126, 2903-18	15.9	80
77	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	2.2	1
76	Identification of genetic test results with conflicting interpretations in prospective registry of multiplex testing (PROMPT).. *Journal of Clinical Oncology*, **2016**, 34, 1510-1510	2.2	

75	The BRCA1-Δ11q Alternative Splice Isoform Bypasses Germline Mutations and Promotes Therapeutic Resistance to PARP Inhibition and Cisplatin. *Cancer Research*, **2016**, 76, 2778-90	10.1	136
74	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	8.3	58
73	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	2.2	110
72	FANCM 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, 5345-55	5.6	68
71	Mammographic density and breast cancer in women from high risk families. *Breast Cancer Research*, **2015**, 17, 93	8.3	15
70	PARP inhibitors in ovarian cancer. *British Journal of Cancer*, **2015**, 113 Suppl 1, S1-2	8.7	1
69	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, 351-7	4.4	25
68	Olaparib monotherapy in patients with advanced cancer and a germline BRCA1/2 mutation. *Journal of Clinical Oncology*, **2015**, 33, 244-50	2.2	1171
67	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	2.2	11
66	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	2.2	
65	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	2.2	2
64	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	2.2	1
63	Multiplex testing in high risk BRCA1/2-negative families.. *Journal of Clinical Oncology*, **2015**, 33, e12557-e12557	2.2	
62	Prevalence of germline MUTYH mutations among Lynch-like syndrome patients. *European Journal of Cancer*, **2014**, 50, 2241-50	7.5	54
61	Molecular features of the basal-like breast cancer subtype based on BRCA1 mutation status. *Breast Cancer Research and Treatment*, **2014**, 147, 185-91	4.4	33
60	RAD51C germline mutations found in Spanish site-specific breast cancer and breast-ovarian cancer families. *Breast Cancer Research and Treatment*, **2014**, 147, 133-43	4.4	23
59	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, 1656-63	10.3	118
58	Prevalence and characteristics of MUTYH-associated polyposis in patients with multiple adenomatous and serrated polyps. *Clinical Cancer Research*, **2014**, 20, 1158-68	12.9	43

57	About 1% of the breast and ovarian Spanish families testing negative for BRCA1 and BRCA2 are carriers of RAD51D pathogenic variants. *International Journal of Cancer*, **2014**, 134, 2088-97	7.5	21
56	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	2.2	7
55	The Role of Platinum Compounds for the Treatment of Breast Cancer. *Current Breast Cancer Reports*, **2013**, 5, 11-22	0.8	1
54	Mutation analysis of the SHFM1 gene in breast/ovarian cancer families. *Journal of Cancer Research and Clinical Oncology*, **2013**, 139, 529-32	4.9	4
53	Prediction models in Lynch syndrome. *Familial Cancer*, **2013**, 12, 217-28	3	15
52	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	4.4	22
51	Clinical subtypes and molecular characteristics of serrated polyposis syndrome. *Clinical Gastroenterology and Hepatology*, **2013**, 11, 705-11; quiz e46	6.9	30
50	Mutation analysis of the BCCIP gene for breast cancer susceptibility in breast/ovarian cancer families. *Gynecologic Oncology*, **2013**, 131, 460-3	4.9	3
49	Genome-wide association study in BRCA1 mutation carriers identifies novel loci associated with breast and ovarian cancer risk. *PLoS Genetics*, **2013**, 9, e1003212	6	209
48	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, 272-9	19.2	42
47	Analysis of PALB2 gene in BRCA1/BRCA2 negative Spanish hereditary breast/ovarian cancer families with pancreatic cancer cases. *PLoS ONE*, **2013**, 8, e67538	3.7	35
46	Olaparib monotherapy in patients with advanced cancer and a germ-line BRCA1/2 mutation: An open-label phase II study.. *Journal of Clinical Oncology*, **2013**, 31, 11024-11024	2.2	10
45	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	2.2	
44	Detection of a large rearrangement in PALB2 in Spanish breast cancer families with male breast cancer. *Breast Cancer Research and Treatment*, **2012**, 132, 307-15	4.4	44
43	Combining a PI3K inhibitor with a PARP inhibitor provides an effective therapy for BRCA1-related breast cancer. *Cancer Discovery*, **2012**, 2, 1048-63	24.4	335
42	What factors may influence psychological well being at three months and one year post BRCA genetic result disclosure?. *Breast*, **2012**, 21, 755-60	3.6	27
41	Characterization of BRCA1 and BRCA2 splicing variants: a collaborative report by ENIGMA consortium members. *Breast Cancer Research and Treatment*, **2012**, 132, 1009-23	4.4	51
40	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, 273-83	4.4	11

39	Breast cancer phenotype in women with TP53 germline mutations: a Li-Fraumeni syndrome consortium effort. *Breast Cancer Research and Treatment*, **2012**, 133, 1125-30	4.4	120
38	Performance of PREMM(1,2,6), MMRpredict, and MMRpro in detecting Lynch syndrome among endometrial cancer cases. *Genetics in Medicine*, **2012**, 14, 670-80	8.1	34
37	Phase I, open-label study of olaparib plus cisplatin in patients with advanced solid tumors.. *Journal of Clinical Oncology*, **2012**, 30, 1009-1009	2.2	5
36	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	2.2	2
35	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	2.2	3
34	Presentation and treatment of HER2-positive metastatic breast cancer patients already treated with adjuvant trastuzumab.. *Journal of Clinical Oncology*, **2012**, 30, 619-619	2.2	
33	Prognostic significance of PI3K pathway (PI3Kp) dysregulation in metastatic breast cancer (MBC) patients (pts).. *Journal of Clinical Oncology*, **2012**, 30, 566-566	2.2	
32	The PREMM(1,2,6) model predicts risk of MLH1, MSH2, and MSH6 germline mutations based on cancer history. *Gastroenterology*, **2011**, 140, 73-81	13.3	146
31	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	2.6	8
30	International distribution and age estimation of the Portuguese BRCA2 c.156_157insAlu founder mutation. *Breast Cancer Research and Treatment*, **2011**, 127, 671-9	4.4	21
29	Germline ATM mutational analysis in BRCA1/BRCA2 negative hereditary breast cancer families by MALDI-TOF mass spectrometry. *Breast Cancer Research and Treatment*, **2011**, 128, 573-9	4.4	6
28	Evidence for a link between TNFRSF11A and risk of breast cancer. *Breast Cancer Research and Treatment*, **2011**, 129, 947-54	4.4	11
27	Stumbling blocks on the path to personalized medicine in breast cancer: the case of PARP inhibitors for BRCA1/2-associated cancers. *Cancer Discovery*, **2011**, 1, 29-34	24.4	39
26	Challenges to the development of new agents for molecularly defined patient subsets: lessons from BRCA1/2-associated breast cancer. *Journal of Clinical Oncology*, **2011**, 29, 4224-6	2.2	21
25	MLH1 founder mutations with moderate penetrance in Spanish Lynch syndrome families. *Cancer Research*, **2010**, 70, 7379-91	10.1	28
24	Parity and the risk of breast and ovarian cancer in BRCA1 and BRCA2 mutation carriers. *Breast Cancer Research and Treatment*, **2010**, 119, 221-32	4.4	48
23	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, 221-5	4.4	7
22	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, 389-98	4.4	139

21	Heterogeneous prevalence of recurrent BRCA1 and BRCA2 mutations in Spain according to the geographical area: implications for genetic testing. *Familial Cancer*, **2010**, 9, 187-91	3	19
20	Uptake of predictive testing among relatives of BRCA1 and BRCA2 families: a multicenter study in northeastern Spain. *Familial Cancer*, **2010**, 9, 297-304	3	19
19	Development and validation of a colon cancer risk assessment tool for patients undergoing colonoscopy. *American Journal of Gastroenterology*, **2009**, 104, 1508-18	0.7	55
18	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, 461-5	4.4	8
17	Opinion about reproductive decision making among individuals undergoing BRCA1/2 genetic testing in a multicentre Spanish cohort. *Human Reproduction*, **2009**, 24, 1000-6	5.7	51
16	Validation and extension of the PREMM1,2 model in a population-based cohort of colorectal cancer patients. *Gastroenterology*, **2008**, 134, 39-46	13.3	49
15	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, 2044-51	4	57
14	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, 455-7	4.4	10
13	Attitudes toward prenatal genetic testing in patients with familial adenomatous polyposis. *American Journal of Gastroenterology*, **2007**, 102, 1284-90	0.7	40
12	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, 5545-56	2.3	7
11	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, 5035-6; author reply 5036-8	2.2	14
10	BRAF mutations in colorectal carcinoma suggest two entities of microsatellite-unstable tumors. *Cancer*, **2006**, 106, 2528-9; author reply 2529	6.4	
9	Prediction of MLH1 and MSH2 mutations in Lynch syndrome. *JAMA - Journal of the American Medical Association*, **2006**, 296, 1469-78	27.4	132
8	Factors associated with enrollment in cancer genetics research. *Cancer Epidemiology Biomarkers and Prevention*, **2006**, 15, 1355-9	4	34
7	Sex ratio distortion in offspring of families with BRCA1 or BRCA2 mutant alleles: an ascertainment bias phenomenon?. *Breast Cancer Research and Treatment*, **2005**, 92, 273-7	4.4	4
6	Comprehensive genetic and endoscopic evaluation may be necessary to distinguish sporadic versus familial adenomatous polyposis-associated abdominal desmoid tumors. *Surgery*, **2004**, 135, 683-9	3.6	13
5	Genetic counseling program in familial breast cancer: analysis of its effectiveness, cost and cost-effectiveness ratio. *International Journal of Cancer*, **2004**, 112, 647-52	7.5	35
4	Haplotype analysis of the BRCA2 9254delATCAT recurrent mutation in breast/ovarian cancer families from Spain. *Human Mutation*, **2003**, 21, 452	4.7	13

3	RNA analysis of eight BRCA1 and BRCA2 unclassified variants identified in breast/ovarian cancer families from Spain. *Human Mutation*, **2003**, 22, 337	4.7	30
2	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, 415-20	1.7	60
1	What is the hereditary non-polyposis colorectal cancer syndrome? **2000**, 2, 191-201		1