

# Ana Osorio

## List of Publications by Year in Descending Order

**Source:** <https://exaly.com/author-pdf/1496950/ana-osorio-publications-by-year.pdf>

**Version:** 2024-04-20

This document has been generated based on the publications and citations recorded by exaly.com. For the latest version of this publication list, visit the link given above.

The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

162  
papers

9,149  
citations

52  
h-index

91  
g-index

173  
ext. papers

11,163  
ext. citations

8.6  
avg, IF

4.31  
L-index

#	Paper	IF	Citations
162	Pathology of Tumors Associated With Pathogenic Germline Variants in 9 Breast Cancer Susceptibility Genes.. <i>JAMA Oncology</i> , <b>2022</b> ,	13.4	4
161	Cross-Cancer Genome-Wide Association Study of Endometrial Cancer and Epithelial Ovarian Cancer Identifies Genetic Risk Regions Associated with Risk of Both Cancers. <i>Cancer Epidemiology Biomarkers and Prevention</i> , <b>2021</b> , 30, 217-228	4	7
160	First international workshop of the ATM and cancer risk group (4-5 December 2019). <i>Familial Cancer</i> , <b>2021</b> , 1	3	5
159	A Collaborative Effort to Define Classification Criteria for ATM Variants in Hereditary Cancer Patients. <i>Clinical Chemistry</i> , <b>2021</b> , 67, 518-533	5.5	1
158	Small molecule inhibitor of OGG1 blocks oxidative DNA damage repair at telomeres and potentiates methotrexate anticancer effects. <i>Scientific Reports</i> , <b>2021</b> , 11, 3490	4.9	4
157	A case-only study to identify genetic modifiers of breast cancer risk for BRCA1/BRCA2 mutation carriers. <i>Nature Communications</i> , <b>2021</b> , 12, 1078	17.4	4
156	Breast Cancer Risk Genes - Association Analysis in More than 113,000 Women. <i>New England Journal of Medicine</i> , <b>2021</b> , 384, 428-439	59.2	143
155	Oral contraceptive use and ovarian cancer risk for BRCA1/2 mutation carriers: an international cohort study. <i>American Journal of Obstetrics and Gynecology</i> , <b>2021</b> , 225, 51.e1-51.e17	6.4	9
154	Development and Validation of the Gene Expression Predictor of High-grade Serous Ovarian Carcinoma Molecular SubTYPE (PrOTYPE). <i>Clinical Cancer Research</i> , <b>2020</b> , 26, 5411-5423	12.9	21
153	Clinical and pathological associations of PTEN expression in ovarian cancer: a multicentre study from the Ovarian Tumour Tissue Analysis Consortium. <i>British Journal of Cancer</i> , <b>2020</b> , 123, 793-802	8.7	16
152	Characterization of the Cancer Spectrum in Men With Germline BRCA1 and BRCA2 Pathogenic Variants: Results From the Consortium of Investigators of Modifiers of BRCA1/2 (CIMBA). <i>JAMA Oncology</i> , <b>2020</b> , 6, 1218-1230	13.4	25
151	Ovarian and Breast Cancer Risks Associated With Pathogenic Variants in RAD51C and RAD51D. <i>Journal of the National Cancer Institute</i> , <b>2020</b> , 112, 1242-1250	9.7	51
150	Transcriptome-wide association study of breast cancer risk by estrogen-receptor status. <i>Genetic Epidemiology</i> , <b>2020</b> , 44, 442-468	2.6	9
149	Alcohol Consumption, Cigarette Smoking, and Risk of Breast Cancer for and Mutation Carriers: Results from The BRCA1 and BRCA2 Cohort Consortium. <i>Cancer Epidemiology Biomarkers and Prevention</i> , <b>2020</b> , 29, 368-378	4	9
148	The Spectrum of Protein Truncating Variants in European Breast Cancer Cases. <i>Cancers</i> , <b>2020</b> , 12,	6.6	7
147	Association of Genomic Domains in and with Prostate Cancer Risk and Aggressiveness. <i>Cancer Research</i> , <b>2020</b> , 80, 624-638	10.1	22
146	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. <i>Nature Genetics</i> , <b>2020</b> , 52, 56-73	36.3	56

145	Mutational Screening of BRCA1/2 Genes as a Predictive Factor for Therapeutic Response in Epithelial Ovarian Cancer: A Consensus Guide from the Spanish Society of Pathology (SEAP-IAP) and the Spanish Society of Human Genetics (AEGH). <i>Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin</i> , <b>2020</b> , 476, 195-207	5.1	7
144	Polygenic risk scores and breast and epithelial ovarian cancer risks for carriers of BRCA1 and BRCA2 pathogenic variants. <i>Genetics in Medicine</i> , <b>2020</b> , 22, 1653-1666	8.1	34
143	Shared heritability and functional enrichment across six solid cancers. <i>Nature Communications</i> , <b>2019</b> , 10, 431	17.4	45
142	Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. <i>Nature Communications</i> , <b>2019</b> , 10, 1741	17.4	47
141	The p.Arg658* truncating variant is associated with risk of triple-negative breast cancer. <i>Npj Breast Cancer</i> , <b>2019</b> , 5, 38	7.8	12
140	RECQL5: Another DNA helicase potentially involved in hereditary breast cancer susceptibility. <i>Human Mutation</i> , <b>2019</b> , 40, 566-577	4.7	10
139	A common SNP in the UNG gene decreases ovarian cancer risk in BRCA2 mutation carriers. <i>Molecular Oncology</i> , <b>2019</b> , 13, 1110-1120	7.9	2
138	Mutational spectrum in a worldwide study of 29,700 families with BRCA1 or BRCA2 mutations. <i>Human Mutation</i> , <b>2018</b> , 39, 593-620	4.7	138
137	Individuals with FANCM biallelic mutations do not develop Fanconi anemia, but show risk for breast cancer, chemotherapy toxicity and may display chromosome fragility. <i>Genetics in Medicine</i> , <b>2018</b> , 20, 452-457	8.1	44
136	A Transcriptome-Wide Association Study Among 97,898 Women to Identify Candidate Susceptibility Genes for Epithelial Ovarian Cancer Risk. <i>Cancer Research</i> , <b>2018</b> , 78, 5419-5430	10.1	32
135	The Influence of Number and Timing of Pregnancies on Breast Cancer Risk for Women With or Mutations. <i>JNCI Cancer Spectrum</i> , <b>2018</b> , 2, pky078	4.6	10
134	Oral Contraceptive Use and Breast Cancer Risk: Retrospective and Prospective Analyses From a BRCA1 and BRCA2 Mutation Carrier Cohort Study. <i>JNCI Cancer Spectrum</i> , <b>2018</b> , 2, pky023	4.6	13
133	Hypomorphic Missense Variants Confer Moderate Risks of Breast Cancer. <i>Cancer Research</i> , <b>2017</b> , 77, 2789-2799	10.1	49
132	Almost 2% of Spanish breast cancer families are associated to germline pathogenic mutations in the ATM gene. <i>Breast Cancer Research and Treatment</i> , <b>2017</b> , 161, 597-604	4.4	12
131	Risks of Breast, Ovarian, and Contralateral Breast Cancer for BRCA1 and BRCA2 Mutation Carriers. <i>JAMA - Journal of the American Medical Association</i> , <b>2017</b> , 317, 2402-2416	27.4	1140
130	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. <i>Nature Genetics</i> , <b>2017</b> , 49, 680-691	36.3	190
129	Dose-Response Association of CD8+ Tumor-Infiltrating Lymphocytes and Survival Time in High-Grade Serous Ovarian Cancer. <i>JAMA Oncology</i> , <b>2017</b> , 3, e173290	13.4	152
128	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. <i>Nature Genetics</i> , <b>2017</b> , 49, 1767-1778	36.3	186

127	Prediction of Breast and Prostate Cancer Risks in Male BRCA1 and BRCA2 Mutation Carriers Using Polygenic Risk Scores. <i>Journal of Clinical Oncology</i> , <b>2017</b> , 35, 2240-2250	2.2	101
126	Characterisation of the novel deleterious RAD51C p.Arg312Trp variant and prioritisation criteria for functional analysis of RAD51C missense changes. <i>British Journal of Cancer</i> , <b>2017</b> , 117, 1048-1062	8.7	4
125	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. <i>Breast Cancer Research and Treatment</i> , <b>2017</b> , 161, 117-134	4.4	15
124	Genetic variation in the DNA glycosylase gene is associated with oxidative DNA damage in mutation carriers. <i>Oncotarget</i> , <b>2017</b> , 8, 114626-114636	3.3	7
123	No clinical utility of KRAS variant rs61764370 for ovarian or breast cancer. <i>Gynecologic Oncology</i> , <b>2016</b> , 141, 386-401	4.9	15
122	Germline missense pathogenic variants in the BRCA1 BRCT domain, p.Gly1706Glu and p.Ala1708Glu, increase cellular sensitivity to PARP inhibitor olaparib by a dominant negative effect. <i>Human Molecular Genetics</i> , <b>2016</b> , 25, 5287-5299	5.6	2
121	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. <i>Nature Communications</i> , <b>2016</b> , 7, 11375	17.4	64
120	Functional mechanisms underlying pleiotropic risk alleles at the 19p13.1 breast-ovarian cancer susceptibility locus. <i>Nature Communications</i> , <b>2016</b> , 7, 12675	17.4	53
119	Breast cancer risk variants at 6q25 display different phenotype associations and regulate ESR1, RMND1 and CCDC170. <i>Nature Genetics</i> , <b>2016</b> , 48, 374-86	36.3	93
118	BRCA2 Polymorphic Stop Codon K3326X and the Risk of Breast, Prostate, and Ovarian Cancers. <i>Journal of the National Cancer Institute</i> , <b>2016</b> , 108,	9.7	65
117	Molecular insights into the OGG1 gene, a cancer risk modifier in BRCA1 and BRCA2 mutations carriers. <i>Oncotarget</i> , <b>2016</b> , 7, 25815-25	3.3	13
116	Fine-Scale Mapping at 9p22.2 Identifies Candidate Causal Variants That Modify Ovarian Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. <i>PLoS ONE</i> , <b>2016</b> , 11, e0158801	3.7	7
115	Identification of independent association signals and putative functional variants for breast cancer risk through fine-scale mapping of the 12p11 locus. <i>Breast Cancer Research</i> , <b>2016</b> , 18, 64	8.3	25
114	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> , <b>2016</b> , 18, 15	8.3	58
113	Identification of six new susceptibility loci for invasive epithelial ovarian cancer. <i>Nature Genetics</i> , <b>2015</b> , 47, 164-71	36.3	177
112	FANCM 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> , <b>2015</b> , 24, 5345-55	5.6	68
111	DNA repair capacity is impaired in healthy BRCA1 heterozygous mutation carriers. <i>Breast Cancer Research and Treatment</i> , <b>2015</b> , 152, 271-82	4.4	23
110	Association of type and location of BRCA1 and BRCA2 mutations with risk of breast and ovarian cancer. <i>JAMA - Journal of the American Medical Association</i> , <b>2015</b> , 313, 1347-61	27.4	286

109	Candidate genetic modifiers for breast and ovarian cancer risk in BRCA1 and BRCA2 mutation carriers. <i>Cancer Epidemiology Biomarkers and Prevention</i> , <b>2015</b> , 24, 308-16	4	20
108	MicroRNA expression signatures for the prediction of BRCA1/2 mutation-associated hereditary breast cancer in paraffin-embedded formalin-fixed breast tumors. <i>International Journal of Cancer</i> , <b>2015</b> , 136, 593-602	7.5	32
107	An original phylogenetic approach identified mitochondrial haplogroup T1a1 as inversely associated with breast cancer risk in BRCA2 mutation carriers. <i>Breast Cancer Research</i> , <b>2015</b> , 17, 61	8.3	16
106	Assessing associations between the AURKA-HMMR-TPX2-TUBG1 functional module and breast cancer risk in BRCA1/2 mutation carriers. <i>PLoS ONE</i> , <b>2015</b> , 10, e0120020	3.7	26
105	BRCA1 and BRCA2 mutations in males with familial breast and ovarian cancer syndrome. Results of a Spanish multicenter study. <i>Familial Cancer</i> , <b>2015</b> , 14, 505-13	3	7
104	Impact of chemotherapy on telomere length in sporadic and familial breast cancer patients. <i>Breast Cancer Research and Treatment</i> , <b>2015</b> , 149, 385-94	4.4	22
103	Deletion at 6q24.2-26 predicts longer survival of high-grade serous epithelial ovarian cancer patients. <i>Molecular Oncology</i> , <b>2015</b> , 9, 422-36	7.9	14
102	Pathological features of breast and ovarian cancers in RAD51C germline mutation carriers. <i>Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin</i> , <b>2014</b> , 465, 365-9	5.1	12
101	DNA glycosylases involved in base excision repair may be associated with cancer risk in BRCA1 and BRCA2 mutation carriers. <i>PLoS Genetics</i> , <b>2014</b> , 10, e1004256	6	33
100	About 1% of the breast and ovarian Spanish families testing negative for BRCA1 and BRCA2 are carriers of RAD51D pathogenic variants. <i>International Journal of Cancer</i> , <b>2014</b> , 134, 2088-97	7.5	21
99	Refined histopathological predictors of BRCA1 and BRCA2 mutation status: a large-scale analysis of breast cancer characteristics from the BCAC, CIMBA, and ENIGMA consortia. <i>Breast Cancer Research</i> , <b>2014</b> , 16, 3419	8.3	82
98	Associations of common breast cancer susceptibility alleles with risk of breast cancer subtypes in BRCA1 and BRCA2 mutation carriers. <i>Breast Cancer Research</i> , <b>2014</b> , 16, 3416	8.3	46
97	Population distribution and ancestry of the cancer protective MDM2 SNP285 (rs117039649). <i>Oncotarget</i> , <b>2014</b> , 5, 8223-34	3.3	21
96	MicroRNA-based molecular classification of non-BRCA1/2 hereditary breast tumours. <i>British Journal of Cancer</i> , <b>2013</b> , 109, 2724-34	8.7	22
95	DNA copy number profiling reveals extensive genomic loss in hereditary BRCA1 and BRCA2 ovarian carcinomas. <i>British Journal of Cancer</i> , <b>2013</b> , 108, 1732-42	8.7	13
94	The highly prevalent BRCA2 mutation c.2808_2811del (3036delACAA) is located in a mutational hotspot and has multiple origins. <i>Carcinogenesis</i> , <b>2013</b> , 34, 2505-11	4.6	10
93	Identification of a BRCA2-specific modifier locus at 6p24 related to breast cancer risk. <i>PLoS Genetics</i> , <b>2013</b> , 9, e1003173	6	90
92	Genome-wide association study in BRCA1 mutation carriers identifies novel loci associated with breast and ovarian cancer risk. <i>PLoS Genetics</i> , <b>2013</b> , 9, e1003212	6	209

91	Analysis of PALB2 gene in BRCA1/BRCA2 negative Spanish hereditary breast/ovarian cancer families with pancreatic cancer cases. <i>PLoS ONE</i> , <b>2013</b> , 8, e67538	3.7	35
90	Evaluation of rare variants in the new fanconi anemia gene ERCC4 (FANCO) as familial breast/ovarian cancer susceptibility alleles. <i>Human Mutation</i> , <b>2013</b> , 34, 1615-8	4.7	23
89	Whole exome sequencing suggests much of non-BRCA1/BRCA2 familial breast cancer is due to moderate and low penetrance susceptibility alleles. <i>PLoS ONE</i> , <b>2013</b> , 8, e55681	3.7	77
88	Common variants at 12p11, 12q24, 9p21, 9q31.2 and in ZNF365 are associated with breast cancer risk for BRCA1 and/or BRCA2 mutation carriers. <i>Breast Cancer Research</i> , <b>2012</b> , 14, R33	8.3	70
87	Ovarian cancer susceptibility alleles and risk of ovarian cancer in BRCA1 and BRCA2 mutation carriers. <i>Human Mutation</i> , <b>2012</b> , 33, 690-702	4.7	31
86	Detection of a large rearrangement in PALB2 in Spanish breast cancer families with male breast cancer. <i>Breast Cancer Research and Treatment</i> , <b>2012</b> , 132, 307-15	4.4	44
85	Mutational analysis of telomere genes in BRCA1/2-negative breast cancer families with very short telomeres. <i>Breast Cancer Research and Treatment</i> , <b>2012</b> , 134, 1337-43	4.4	5
84	Evaluation of chromosome 6p22 as a breast cancer risk modifier locus in a follow-up study of BRCA2 mutation carriers. <i>Breast Cancer Research and Treatment</i> , <b>2012</b> , 136, 295-302	4.4	3
83	Deregulated miRNAs in hereditary breast cancer revealed a role for miR-30c in regulating KRAS oncogene. <i>PLoS ONE</i> , <b>2012</b> , 7, e38847	3.7	66
82	Association of PHB 1630 C>T and MTHFR 677 C>T polymorphisms with breast and ovarian cancer risk in BRCA1/2 mutation carriers: results from a multicenter study. <i>British Journal of Cancer</i> , <b>2012</b> , 106, 2016-24	8.7	25
81	Assessing the RNA effect of 26 DNA variants in the BRCA1 and BRCA2 genes. <i>Breast Cancer Research and Treatment</i> , <b>2012</b> , 132, 979-92	4.4	19
80	Rare mutations in XRCC2 increase the risk of breast cancer. <i>American Journal of Human Genetics</i> , <b>2012</b> , 90, 734-9	11	143
79	A nonsynonymous polymorphism in IRS1 modifies risk of developing breast and ovarian cancers in BRCA1 and ovarian cancer in BRCA2 mutation carriers. <i>Cancer Epidemiology Biomarkers and Prevention</i> , <b>2012</b> , 21, 1362-70	4	20
78	Common variants at the 19p13.1 and ZNF365 loci are associated with ER subtypes of breast cancer and ovarian cancer risk in BRCA1 and BRCA2 mutation carriers. <i>Cancer Epidemiology Biomarkers and Prevention</i> , <b>2012</b> , 21, 645-57	4	44
77	Pathology of breast and ovarian cancers among BRCA1 and BRCA2 mutation carriers: results from the Consortium of Investigators of Modifiers of BRCA1/2 (CIMBA). <i>Cancer Epidemiology Biomarkers and Prevention</i> , <b>2012</b> , 21, 134-47	4	411
76	Predominance of pathogenic missense variants in the RAD51C gene occurring in breast and ovarian cancer families. <i>Human Molecular Genetics</i> , <b>2012</b> , 21, 2889-98	5.6	70
75	Exploring the link between MORF4L1 and risk of breast cancer. <i>Breast Cancer Research</i> , <b>2011</b> , 13, R40	8.3	16
74	Common breast cancer susceptibility alleles are associated with tumour subtypes in BRCA1 and BRCA2 mutation carriers: results from the Consortium of Investigators of Modifiers of BRCA1/2. <i>Breast Cancer Research</i> , <b>2011</b> , 13, R110	8.3	62



73	Evaluation of the XRCC1 gene as a phenotypic modifier in BRCA1/2 mutation carriers. Results from the consortium of investigators of modifiers of BRCA1/BRCA2. <i>British Journal of Cancer</i> , <b>2011</b> , 104, 1356-61	8.7	6
72	Evidence for a link between TNFRSF11A and risk of breast cancer. <i>Breast Cancer Research and Treatment</i> , <b>2011</b> , 129, 947-54	4.4	11
71	Haplotype structure in Ashkenazi Jewish BRCA1 and BRCA2 mutation carriers. <i>Human Genetics</i> , <b>2011</b> , 130, 685-99	6.3	15
70	Transcriptional characteristics of familial non-BRCA1/BRCA2 breast tumors. <i>International Journal of Cancer</i> , <b>2011</b> , 128, 2635-44	7.5	11
69	Common alleles at 6q25.1 and 1p11.2 are associated with breast cancer risk for BRCA1 and BRCA2 mutation carriers. <i>Human Molecular Genetics</i> , <b>2011</b> , 20, 3304-21	5.6	62
68	The role of KRAS rs61764370 in invasive epithelial ovarian cancer: implications for clinical testing. <i>Clinical Cancer Research</i> , <b>2011</b> , 17, 3742-50	12.9	45
67	The rs12975333 variant in the miR-125a and breast cancer risk in Germany, Italy, Australia and Spain. <i>Journal of Medical Genetics</i> , <b>2011</b> , 48, 703-4	5.8	13
66	Genetic variation at 9p22.2 and ovarian cancer risk for BRCA1 and BRCA2 mutation carriers. <i>Journal of the National Cancer Institute</i> , <b>2011</b> , 103, 105-16	9.7	37
65	Genetic anticipation is associated with telomere shortening in hereditary breast cancer. <i>PLoS Genetics</i> , <b>2011</b> , 7, e1002182	6	68
64	Interplay between BRCA1 and RHAMM regulates epithelial apicobasal polarization and may influence risk of breast cancer. <i>PLoS Biology</i> , <b>2011</b> , 9, e1001199	9.7	73
63	A locus on 19p13 modifies risk of breast cancer in BRCA1 mutation carriers and is associated with hormone receptor-negative breast cancer in the general population. <i>Nature Genetics</i> , <b>2010</b> , 42, 885-92	36.3	276
62	Common breast cancer susceptibility alleles and the risk of breast cancer for BRCA1 and BRCA2 mutation carriers: implications for risk prediction. <i>Cancer Research</i> , <b>2010</b> , 70, 9742-54	10.1	147
61	BRCA1 CpG island hypermethylation predicts sensitivity to poly(adenosine diphosphate)-ribose polymerase inhibitors. <i>Journal of Clinical Oncology</i> , <b>2010</b> , 28, e563-4; author reply e565-6	2.2	129
60	Association of the variants CASP8 D302H and CASP10 V410I with breast and ovarian cancer risk in BRCA1 and BRCA2 mutation carriers. <i>Cancer Epidemiology Biomarkers and Prevention</i> , <b>2010</b> , 19, 2859-68	4	32
59	Common genetic variants and modification of penetrance of BRCA2-associated breast cancer. <i>PLoS Genetics</i> , <b>2010</b> , 6, e1001183	6	74
58	Parity and the risk of breast and ovarian cancer in BRCA1 and BRCA2 mutation carriers. <i>Breast Cancer Research and Treatment</i> , <b>2010</b> , 119, 221-32	4.4	48
57	Mutational analysis of FANCL, FANCM and the recently identified FANCI suggests that among the 13 known Fanconi Anemia genes, only FANCD1/BRCA2 plays a major role in high-risk breast cancer predisposition. <i>Carcinogenesis</i> , <b>2009</b> , 30, 1898-902	4.6	20
56	Common variants in LSP1, 2q35 and 8q24 and breast cancer risk for BRCA1 and BRCA2 mutation carriers. <i>Human Molecular Genetics</i> , <b>2009</b> , 18, 4442-56	5.6	91

55	Reproductive and hormonal factors, and ovarian cancer risk for BRCA1 and BRCA2 mutation carriers: results from the International BRCA1/2 Carrier Cohort Study. <i>Cancer Epidemiology Biomarkers and Prevention</i> , <b>2009</b> , 18, 601-10	4	110
54	Evaluation of the BRCA1 interacting genes RAP80 and CCDC98 in familial breast cancer susceptibility. <i>Breast Cancer Research and Treatment</i> , <b>2009</b> , 113, 371-6	4.4	19
53	Analysis of FANCB and FANCN/PALB2 fanconi anemia genes in BRCA1/2-negative Spanish breast cancer families. <i>Breast Cancer Research and Treatment</i> , <b>2009</b> , 113, 545-51	4.4	75
52	Gene expression profiling integrated into network modelling reveals heterogeneity in the mechanisms of BRCA1 tumorigenesis. <i>British Journal of Cancer</i> , <b>2009</b> , 101, 1469-80	8.7	13
51	The TP53 Arg72Pro and MDM2 309G>T polymorphisms are not associated with breast cancer risk in BRCA1 and BRCA2 mutation carriers. <i>British Journal of Cancer</i> , <b>2009</b> , 101, 1456-60	8.7	17
50	Evaluation of a candidate breast cancer associated SNP in ERCC4 as a risk modifier in BRCA1 and BRCA2 mutation carriers. Results from the Consortium of Investigators of Modifiers of BRCA1/BRCA2 (CIMBA). <i>British Journal of Cancer</i> , <b>2009</b> , 101, 2048-54	8.7	13
49	Distinct genomic aberration patterns are found in familial breast cancer associated with different immunohistochemical subtypes. <i>Oncogene</i> , <b>2008</b> , 27, 3165-75	9.2	68
48	An evaluation of the polymorphisms Ins16bp and Arg72Pro in p53 as breast cancer risk modifiers in BRCA1 and BRCA2 mutation carriers. <i>British Journal of Cancer</i> , <b>2008</b> , 99, 974-7	8.7	12
47	The average cumulative risks of breast and ovarian cancer for carriers of mutations in BRCA1 and BRCA2 attending genetic counseling units in Spain. <i>Clinical Cancer Research</i> , <b>2008</b> , 14, 2861-9	12.9	77
46	Common breast cancer-predisposition alleles are associated with breast cancer risk in BRCA1 and BRCA2 mutation carriers. <i>American Journal of Human Genetics</i> , <b>2008</b> , 82, 937-48	11	218
45	Classification of missense variants of unknown significance in BRCA1 based on clinical and tumor information. <i>Human Mutation</i> , <b>2007</b> , 28, 477-85	4.7	40
44	Loss of the actin regulator HSPC300 results in clear cell renal cell carcinoma protection in Von Hippel-Lindau patients. <i>Human Mutation</i> , <b>2007</b> , 28, 613-21	4.7	35
43	Immunohistochemical classification of non-BRCA1/2 tumors identifies different groups that demonstrate the heterogeneity of BRCA1 families. <i>Modern Pathology</i> , <b>2007</b> , 20, 1298-306	9.8	44
42	Genomewide high-density SNP linkage analysis of non-BRCA1/2 breast cancer families identifies various candidate regions and has greater power than microsatellite studies. <i>BMC Genomics</i> , <b>2007</b> , 8, 299	4.5	23
41	Age at menarche and menopause and breast cancer risk in the International BRCA1/2 Carrier Cohort Study. <i>Cancer Epidemiology Biomarkers and Prevention</i> , <b>2007</b> , 16, 740-6	4	56
40	Estrogen receptor status could modulate the genomic pattern in familial and sporadic breast cancer. <i>Clinical Cancer Research</i> , <b>2007</b> , 13, 7305-13	12.9	28
39	RAD51 135G-->C modifies breast cancer risk among BRCA2 mutation carriers: results from a combined analysis of 19 studies. <i>American Journal of Human Genetics</i> , <b>2007</b> , 81, 1186-200	11	204
38	A haplotype containing the p53 polymorphisms Ins16bp and Arg72Pro modifies cancer risk in BRCA2 mutation carriers. <i>Human Mutation</i> , <b>2006</b> , 27, 242-8	4.7	30



37	Genomic rearrangements at the BRCA1 locus in Spanish families with breast/ovarian cancer. <i>Clinical Chemistry</i> , <b>2006</b> , 52, 1480-5	5.5	54
36	Accurate prediction of BRCA1 and BRCA2 heterozygous genotype using expression profiling after induced DNA damage. <i>Clinical Cancer Research</i> , <b>2006</b> , 12, 3896-901	12.9	32
35	Pathology and gene expression of hereditary breast tumors associated with BRCA1, BRCA2 and CHEK2 gene mutations. <i>Oncogene</i> , <b>2006</b> , 25, 5837-45	9.2	87
34	Mutant BRCA1 alleles transmission: different approaches and different biases. <i>International Journal of Cancer</i> , <b>2005</b> , 113, 166-7	7.5	2
33	Phenotypic characterization of BRCA1 and BRCA2 tumors based in a tissue microarray study with 37 immunohistochemical markers. <i>Breast Cancer Research and Treatment</i> , <b>2005</b> , 90, 5-14	4.4	135
32	The accumulation of specific amplifications characterizes two different genomic pathways of evolution of familial breast tumors. <i>Clinical Cancer Research</i> , <b>2005</b> , 11, 8577-84	12.9	16
31	Immunohistochemical expression of DNA repair proteins in familial breast cancer differentiate BRCA2-associated tumors. <i>Journal of Clinical Oncology</i> , <b>2005</b> , 23, 7503-11	2.2	53
30	A predictor based on the somatic genomic changes of the BRCA1/BRCA2 breast cancer tumors identifies the non-BRCA1/BRCA2 tumors with BRCA1 promoter hypermethylation. <i>Clinical Cancer Research</i> , <b>2005</b> , 11, 1146-53	12.9	49
29	Re: Germline BRCA1 mutations and a basal epithelial phenotype in breast cancer. <i>Journal of the National Cancer Institute</i> , <b>2004</b> , 96, 712-4; author reply 714	9.7	28
28	The variant E233G of the RAD51D gene could be a low-penetrance allele in high-risk breast cancer families without BRCA1/2 mutations. <i>International Journal of Cancer</i> , <b>2004</b> , 110, 845-9	7.5	22
27	The breast cancer low-penetrance allele 1100delC in the CHEK2 gene is not present in Spanish familial breast cancer population. <i>International Journal of Cancer</i> , <b>2004</b> , 108, 54-6	7.5	64
26	Analysis of BRCA1 and BRCA2 genes in Spanish breast/ovarian cancer patients: a high proportion of mutations unique to Spain and evidence of founder effects. <i>Human Mutation</i> , <b>2003</b> , 22, 301-12	4.7	133
25	Haplotype analysis of the BRCA2 9254delATCAT recurrent mutation in breast/ovarian cancer families from Spain. <i>Human Mutation</i> , <b>2003</b> , 21, 452	4.7	13
24	No mutations in the XRCC2 gene in BRCA1/2-negative high-risk breast cancer families. <i>International Journal of Cancer</i> , <b>2003</b> , 103, 136-7	7.5	6
23	Over-representation of two specific haplotypes among chromosomes harbouring BRCA1 mutations. <i>European Journal of Human Genetics</i> , <b>2003</b> , 11, 489-92	5.3	16
22	Immunohistochemical characteristics defined by tissue microarray of hereditary breast cancer not attributable to BRCA1 or BRCA2 mutations: differences from breast carcinomas arising in BRCA1 and BRCA2 mutation carriers. <i>Clinical Cancer Research</i> , <b>2003</b> , 9, 3606-14	12.9	123
21	Loss of heterozygosity analysis at the BRCA loci in tumor samples from patients with familial breast cancer. <i>International Journal of Cancer</i> , <b>2002</b> , 99, 305-9	7.5	86
20	Clustering of cancer-related mutations in a subset of BRCA1 alleles: a study in the Spanish population. <i>International Journal of Cancer</i> , <b>2002</b> , 100, 618-9	7.5	6

19	Association between BRCA1 and BRCA2 mutations and cancer phenotype in Spanish breast/ovarian cancer families: implications for genetic testing. <i>International Journal of Cancer</i> , <b>2002</b> , 97, 466-71	7.5	52
18	A rapid and easy method for multiple endocrine neoplasia type 1 mutation detection using conformation-sensitive gel electrophoresis. <i>Journal of Human Genetics</i> , <b>2002</b> , 47, 190-5	4.3	8
17	Evaluation of linkage of breast cancer to the putative BRCA3 locus on chromosome 13q21 in 128 multiple case families from the Breast Cancer Linkage Consortium. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2002</b> , 99, 827-31	11.5	64
16	Hypermethylation of P16ink4a and P15ink4b genes as a marker of disease in the follow-up of non-Hodgkin lymphomas. <i>British Journal of Haematology</i> , <b>2000</b> , 109, 97-103	4.5	9
15	Identification by comparative genomic hybridization of genetic changes involved in tumoral progression of a T-cell non-Hodgkin lymphoma. <i>Cancer Genetics and Cytogenetics</i> , <b>2000</b> , 117, 41-4		2
14	Molecular analysis of the BRCA1 and BRCA2 genes in 32 breast and/or ovarian cancer Spanish families. <i>British Journal of Cancer</i> , <b>2000</b> , 82, 1266-70	8.7	70
13	sigma(54) Promoters control expression of genes encoding the hook and basal body complex in <i>Rhodobacter sphaeroides</i> . <i>Journal of Bacteriology</i> , <b>2000</b> , 182, 5787-92	3.5	14
12	Genetic and clinical analysis in 10 Spanish patients with multiple endocrine neoplasia type 1. <i>European Journal of Human Genetics</i> , <b>1999</b> , 7, 585-9	5.3	14
11	Prevalence of BRCA1 and BRCA2 Jewish mutations in Spanish breast cancer patients. <i>British Journal of Cancer</i> , <b>1999</b> , 79, 1302-3	8.7	36
10	European multicenter study on LOH of APOC3 at 11q23 in 766 breast cancer patients: relation to clinical variables. Breast Cancer Somatic Genetics Consortium. <i>British Journal of Cancer</i> , <b>1999</b> , 80, 879-82	8.7	12
9	Loss of heterozygosity at 11q23.1 and survival in breast cancer: Results of a large European study. <i>Genes Chromosomes and Cancer</i> , <b>1999</b> , 25, 212-221	5	31
8	Hypermethylation of p15/ink4b/MTS2 gene is differentially implicated among non-Hodgkin lymphomas. <i>Leukemia</i> , <b>1998</b> , 12, 937-41	10.7	35
7	Molecular analysis of the six most recurrent mutations in the BRCA1 gene in 87 Spanish breast/ovarian cancer families. <i>Cancer Letters</i> , <b>1998</b> , 123, 153-8	9.9	19
6	Haplotype and phenotype analysis of nine recurrent BRCA2 mutations in 111 families: results of an international study. <i>American Journal of Human Genetics</i> , <b>1998</b> , 62, 1381-8	11	138
5	Molecular analysis of the BRCA2 gene in 16 breast/ovarian cancer Spanish families. <i>Clinical Genetics</i> , <b>1998</b> , 54, 142-7	4	3
4	The 12 base pair duplication/insertion alteration could be a regulatory mutation. <i>Journal of Medical Genetics</i> , <b>1997</b> , 34, 592-3	5.8	4
3	Mutation analysis of the BRCA2 gene in breast/ovarian cancer Spanish families: identification of two new mutations. <i>Cancer Letters</i> , <b>1997</b> , 121, 115-8	9.9	6
2	Polygenic Risk Modelling for Prediction of Epithelial Ovarian Cancer Risk		1

1 Fine-mapping of 150 breast cancer risk regions identifies 178 high confidence target genes

2