Ana Osorio

List of Publications by Citations

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	Risks of Breast, Ovarian, and Contralateral Breast Cancer for BRCA1 and BRCA2 Mutation Carriers. *JAMA - Journal of the American Medical Association*, **2017**, 317, 2402-2416	27.4	1140
161	Pathology of breast and ovarian cancers among BRCA1 and BRCA2 mutation carriers: results from the Consortium of Investigators of Modifiers of BRCA1/2 (CIMBA). *Cancer Epidemiology Biomarkers and Prevention*, **2012**, 21, 134-47	4	411
160	Association of type and location of BRCA1 and BRCA2 mutations with risk of breast and ovarian cancer. *JAMA - Journal of the American Medical Association*, **2015**, 313, 1347-61	27.4	286
159	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. *Nature Genetics*, **2010**, 42, 885-92	36.3	276
158	Common breast cancer-predisposition alleles are associated with breast cancer risk in BRCA1 and BRCA2 mutation carriers. *American Journal of Human Genetics*, **2008**, 82, 937-48	11	218
157	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
156	RAD51 135G-->C modifies breast cancer risk among BRCA2 mutation carriers: results from a combined analysis of 19 studies. *American Journal of Human Genetics*, **2007**, 81, 1186-200	11	204
155	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. *Nature Genetics*, **2017**, 49, 680-691	36.3	190
154	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. *Nature Genetics*, **2017**, 49, 1767-1778	36.3	186
153	Identification of six new susceptibility loci for invasive epithelial ovarian cancer. *Nature Genetics*, **2015**, 47, 164-71	36.3	177
152	Dose-Response Association of CD8+ Tumor-Infiltrating Lymphocytes and Survival Time in High-Grade Serous Ovarian Cancer. *JAMA Oncology*, **2017**, 3, e173290	13.4	152
151	Common breast cancer susceptibility alleles and the risk of breast cancer for BRCA1 and BRCA2 mutation carriers: implications for risk prediction. *Cancer Research*, **2010**, 70, 9742-54	10.1	147
150	Rare mutations in XRCC2 increase the risk of breast cancer. *American Journal of Human Genetics*, **2012**, 90, 734-9	11	143
149	Breast Cancer Risk Genes - Association Analysis in More than 113,000 Women. *New England Journal of Medicine*, **2021**, 384, 428-439	59.2	143
148	Mutational spectrum in a worldwide study of 29,700 families with BRCA1 or BRCA2 mutations. *Human Mutation*, **2018**, 39, 593-620	4.7	138
147	Haplotype and phenotype analysis of nine recurrent BRCA2 mutations in 111 families: results of an international study. *American Journal of Human Genetics*, **1998**, 62, 1381-8	11	138
146	Phenotypic characterization of BRCA1 and BRCA2 tumors based in a tissue microarray study with 37 immunohistochemical markers. *Breast Cancer Research and Treatment*, **2005**, 90, 5-14	4.4	135

145	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. *Human Mutation*, **2003**, 22, 301-12	4.7	133
144	BRCA1 CpG island hypermethylation predicts sensitivity to poly(adenosine diphosphate)-ribose polymerase inhibitors. *Journal of Clinical Oncology*, **2010**, 28, e563-4; author reply e565-6	2.2	129
143	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. *Clinical Cancer Research*, **2003**, 9, 3606-14	12.9	123
142	Reproductive and hormonal factors, and ovarian cancer risk for BRCA1 and BRCA2 mutation carriers: results from the International BRCA1/2 Carrier Cohort Study. *Cancer Epidemiology Biomarkers and Prevention*, **2009**, 18, 601-10	4	110
141	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
140	Breast cancer risk variants at 6q25 display different phenotype associations and regulate ESR1, RMND1 and CCDC170. *Nature Genetics*, **2016**, 48, 374-86	36.3	93
139	Common variants in LSP1, 2q35 and 8q24 and breast cancer risk for BRCA1 and BRCA2 mutation carriers. *Human Molecular Genetics*, **2009**, 18, 4442-56	5.6	91
138	Identification of a BRCA2-specific modifier locus at 6p24 related to breast cancer risk. *PLoS Genetics*, **2013**, 9, e1003173	6	90
137	Pathology and gene expression of hereditary breast tumors associated with BRCA1, BRCA2 and CHEK2 gene mutations. *Oncogene*, **2006**, 25, 5837-45	9.2	87
136	Loss of heterozygosity analysis at the BRCA loci in tumor samples from patients with familial breast cancer. *International Journal of Cancer*, **2002**, 99, 305-9	7.5	86
135	Refined histopathological predictors of BRCA1 and BRCA2 mutation status: a large-scale analysis of breast cancer characteristics from the BCAC, CIMBA, and ENIGMA consortia. *Breast Cancer Research* , **2014**, 16, 3419	8.3	82
134	Whole exome sequencing suggests much of non-BRCA1/BRCA2 familial breast cancer is due to moderate and low penetrance susceptibility alleles. *PLoS ONE*, **2013**, 8, e55681	3.7	77
133	The average cumulative risks of breast and ovarian cancer for carriers of mutations in BRCA1 and BRCA2 attending genetic counseling units in Spain. *Clinical Cancer Research*, **2008**, 14, 2861-9	12.9	77
132	Analysis of FANCB and FANCN/PALB2 fanconi anemia genes in BRCA1/2-negative Spanish breast cancer families. *Breast Cancer Research and Treatment*, **2009**, 113, 545-51	4.4	75
131	Common genetic variants and modification of penetrance of BRCA2-associated breast cancer. *PLoS Genetics*, **2010**, 6, e1001183	6	74
130	Interplay between BRCA1 and RHAMM regulates epithelial apicobasal polarization and may influence risk of breast cancer. *PLoS Biology*, **2011**, 9, e1001199	9.7	73
129	Common variants at 12p11, 12q24, 9p21, 9q31.2 and in ZNF365 are associated with breast cancer risk for BRCA1 and/or BRCA2 mutation carriers. *Breast Cancer Research*, **2012**, 14, R33	8.3	70
128	Predominance of pathogenic missense variants in the RAD51C gene occurring in breast and ovarian cancer families. *Human Molecular Genetics*, **2012**, 21, 2889-98	5.6	70

127	Molecular analysis of the BRCA1 and BRCA2 genes in 32 breast and/or ovarian cancer Spanish families. *British Journal of Cancer*, **2000**, 82, 1266-70	8.7	70
126	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
125	Genetic anticipation is associated with telomere shortening in hereditary breast cancer. *PLoS Genetics*, **2011**, 7, e1002182	6	68
124	Distinct genomic aberration patterns are found in familial breast cancer associated with different immunohistochemical subtypes. *Oncogene*, **2008**, 27, 3165-75	9.2	68
123	Deregulated miRNAs in hereditary breast cancer revealed a role for miR-30c in regulating KRAS oncogene. *PLoS ONE*, **2012**, 7, e38847	3.7	66
122	BRCA2 Polymorphic Stop Codon K3326X and the Risk of Breast, Prostate, and Ovarian Cancers. *Journal of the National Cancer Institute*, **2016**, 108,	9.7	65
121	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. *Nature Communications*, **2016**, 7, 11375	17.4	64
120	The breast cancer low-penetrance allele 1100delC in the CHEK2 gene is not present in Spanish familial breast cancer population. *International Journal of Cancer*, **2004**, 108, 54-6	7.5	64
119	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. *Proceedings of the National Academy of Sciences of the United States of America*, **2002**, 99, 827-31	11.5	64
118	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. *Breast Cancer Research*, **2011**, 13, R110	8.3	62
117	Common alleles at 6q25.1 and 1p11.2 are associated with breast cancer risk for BRCA1 and BRCA2 mutation carriers. *Human Molecular Genetics*, **2011**, 20, 3304-21	5.6	62
116	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
115	Age at menarche and menopause and breast cancer risk in the International BRCA1/2 Carrier Cohort Study. *Cancer Epidemiology Biomarkers and Prevention*, **2007**, 16, 740-6	4	56
114	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. *Nature Genetics*, **2020**, 52, 56-73	36.3	56
113	Genomic rearrangements at the BRCA1 locus in Spanish families with breast/ovarian cancer. *Clinical Chemistry*, **2006**, 52, 1480-5	5.5	54
112	Functional mechanisms underlying pleiotropic risk alleles at the 19p13.1 breast-ovarian cancer susceptibility locus. *Nature Communications*, **2016**, 7, 12675	17.4	53
111	Immunohistochemical expression of DNA repair proteins in familial breast cancer differentiate BRCA2-associated tumors. *Journal of Clinical Oncology*, **2005**, 23, 7503-11	2.2	53
110	Association between BRCA1 and BRCA2 mutations and cancer phenotype in Spanish breast/ovarian cancer families: implications for genetic testing. *International Journal of Cancer*, **2002**, 97, 466-71	7.5	52

109	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
108	Hypomorphic Missense Variants Confer Moderate Risks of Breast Cancer. *Cancer Research*, **2017**, 77, 2789-2799	10.1	49
107	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. *Clinical Cancer Research*, **2005**, 11, 1146-53	12.9	49
106	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
105	Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. *Nature Communications*, **2019**, 10, 1741	17.4	47
104	Associations of common breast cancer susceptibility alleles with risk of breast cancer subtypes in BRCA1 and BRCA2 mutation carriers. *Breast Cancer Research*, **2014**, 16, 3416	8.3	46
103	Shared heritability and functional enrichment across six solid cancers. *Nature Communications*, **2019** , 10, 431	17.4	45
102	The role of KRAS rs61764370 in invasive epithelial ovarian cancer: implications for clinical testing. *Clinical Cancer Research*, **2011**, 17, 3742-50	12.9	45
101	Individuals with FANCM biallelic mutations do not develop Fanconi anemia, but show risk for breast cancer, chemotherapy toxicity and may display chromosome fragility. *Genetics in Medicine*, **2018**, 20, 452-457	8.1	44
100	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
99	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. *Cancer Epidemiology Biomarkers and Prevention*, **2012**, 21, 645-57	4	44
98	Immunohistochemical classification of non-BRCA1/2 tumors identifies different groups that demonstrate the heterogeneity of BRCAX families. *Modern Pathology*, **2007**, 20, 1298-306	9.8	44
97	Classification of missense variants of unknown significance in BRCA1 based on clinical and tumor information. *Human Mutation*, **2007**, 28, 477-85	4.7	40
96	Genetic variation at 9p22.2 and ovarian cancer risk for BRCA1 and BRCA2 mutation carriers. *Journal of the National Cancer Institute*, **2011**, 103, 105-16	9.7	37
95	Prevalence of BRCA1 and BRCA2 Jewish mutations in Spanish breast cancer patients. *British Journal of Cancer*, **1999**, 79, 1302-3	8.7	36
94	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
93	Hypermethylation of p15/ink4b/MTS2 gene is differentially implicated among non-Hodgkin© lymphomas. *Leukemia*, **1998**, 12, 937-41	10.7	35
92	Loss of the actin regulator HSPC300 results in clear cell renal cell carcinoma protection in Von Hippel-Lindau patients. *Human Mutation*, **2007**, 28, 613-21	4.7	35

91	Polygenic risk scores and breast and epithelial ovarian cancer risks for carriers of BRCA1 and BRCA2 pathogenic variants. *Genetics in Medicine*, **2020**, 22, 1653-1666	8.1	34
90	DNA glycosylases involved in base excision repair may be associated with cancer risk in BRCA1 and BRCA2 mutation carriers. *PLoS Genetics*, **2014**, 10, e1004256	6	33
89	A Transcriptome-Wide Association Study Among 97,898 Women to Identify Candidate Susceptibility Genes for Epithelial Ovarian Cancer Risk. *Cancer Research*, **2018**, 78, 5419-5430	10.1	32
88	MicroRNA expression signatures for the prediction of BRCA1/2 mutation-associated hereditary breast cancer in paraffin-embedded formalin-fixed breast tumors. *International Journal of Cancer*, **2015**, 136, 593-602	7.5	32
87	Association of the variants CASP8 D302H and CASP10 V410I with breast and ovarian cancer risk in BRCA1 and BRCA2 mutation carriers. *Cancer Epidemiology Biomarkers and Prevention*, **2010**, 19, 2859-68	4	32
86	Accurate prediction of BRCA1 and BRCA2 heterozygous genotype using expression profiling after induced DNA damage. *Clinical Cancer Research*, **2006**, 12, 3896-901	12.9	32
85	Ovarian cancer susceptibility alleles and risk of ovarian cancer in BRCA1 and BRCA2 mutation carriers. *Human Mutation*, **2012**, 33, 690-702	4.7	31
84	Loss of heterozygosity at 11q23.1 and survival in breast cancer: Results of a large European study. *Genes Chromosomes and Cancer*, **1999**, 25, 212-221	5	31
83	A haplotype containing the p53 polymorphisms Ins16bp and Arg72Pro modifies cancer risk in BRCA2 mutation carriers. *Human Mutation*, **2006**, 27, 242-8	4.7	30
82	Estrogen receptor status could modulate the genomic pattern in familial and sporadic breast cancer. *Clinical Cancer Research*, **2007**, 13, 7305-13	12.9	28
81	Re: Germline BRCA1 mutations and a basal epithelial phenotype in breast cancer. *Journal of the National Cancer Institute*, **2004**, 96, 712-4; author reply 714	9.7	28
80	Assessing associations between the AURKA-HMMR-TPX2-TUBG1 functional module and breast cancer risk in BRCA1/2 mutation carriers. *PLoS ONE*, **2015**, 10, e0120020	3.7	26
79	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). *JAMA Oncology*, **2020**, 6, 1218-1230	13.4	25
78	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. *British Journal of Cancer*, **2012**, 106, 2016-24	8.7	25
77	Identification of independent association signals and putative functional variants for breast cancer risk through fine-scale mapping of the 12p11 locus. *Breast Cancer Research*, **2016**, 18, 64	8.3	25
76	DNA repair capacity is impaired in healthy BRCA1 heterozygous mutation carriers. *Breast Cancer Research and Treatment*, **2015**, 152, 271-82	4.4	23
75	Evaluation of rare variants in the new fanconi anemia gene ERCC4 (FANCQ) as familial breast/ovarian cancer susceptibility alleles. *Human Mutation*, **2013**, 34, 1615-8	4.7	23
74	Genomewide high-density SNP linkage analysis of non-BRCA1/2 breast cancer families identifies various candidate regions and has greater power than microsatellite studies. *BMC Genomics*, **2007**, 8, 299	4.5	23

73	Association of Genomic Domains in and with Prostate Cancer Risk and Aggressiveness. *Cancer Research*, **2020**, 80, 624-638	10.1	22
72	MicroRNA-based molecular classification of non-BRCA1/2 hereditary breast tumours. *British Journal of Cancer*, **2013**, 109, 2724-34	8.7	22
71	Impact of chemotherapy on telomere length in sporadic and familial breast cancer patients. *Breast Cancer Research and Treatment*, **2015**, 149, 385-94	4.4	22
70	The variant E233G of the RAD51D gene could be a low-penetrance allele in high-risk breast cancer families without BRCA1/2 mutations. *International Journal of Cancer*, **2004**, 110, 845-9	7.5	22
69	Development and Validation of the Gene Expression Predictor of High-grade Serous Ovarian Carcinoma Molecular SubTYPE (PrOTYPE). *Clinical Cancer Research*, **2020**, 26, 5411-5423	12.9	21
68	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
67	Population distribution and ancestry of the cancer protective MDM2 SNP285 (rs117039649). *Oncotarget*, **2014**, 5, 8223-34	3.3	21
66	Candidate genetic modifiers for breast and ovarian cancer risk in BRCA1 and BRCA2 mutation carriers. *Cancer Epidemiology Biomarkers and Prevention*, **2015**, 24, 308-16	4	20
65	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. *Carcinogenesis*, **2009**, 30, 1898-902	4.6	20
64	A nonsynonymous polymorphism in IRS1 modifies risk of developing breast and ovarian cancers in BRCA1 and ovarian cancer in BRCA2 mutation carriers. *Cancer Epidemiology Biomarkers and Prevention*, **2012**, 21, 1362-70	4	20
63	Assessing the RNA effect of 26 DNA variants in the BRCA1 and BRCA2 genes. *Breast Cancer Research and Treatment*, **2012**, 132, 979-92	4.4	19
62	Evaluation of the BRCA1 interacting genes RAP80 and CCDC98 in familial breast cancer susceptibility. *Breast Cancer Research and Treatment*, **2009**, 113, 371-6	4.4	19
61	Molecular analysis of the six most recurrent mutations in the BRCA1 gene in 87 Spanish breast/ovarian cancer families. *Cancer Letters*, **1998**, 123, 153-8	9.9	19
60	The TP53 Arg72Pro and MDM2 309G>T polymorphisms are not associated with breast cancer risk in BRCA1 and BRCA2 mutation carriers. *British Journal of Cancer*, **2009**, 101, 1456-60	8.7	17
59	Clinical and pathological associations of PTEN expression in ovarian cancer: a multicentre study from the Ovarian Tumour Tissue Analysis Consortium. *British Journal of Cancer*, **2020**, 123, 793-802	8.7	16
58	An original phylogenetic approach identified mitochondrial haplogroup T1a1 as inversely associated with breast cancer risk in BRCA2 mutation carriers. *Breast Cancer Research*, **2015**, 17, 61	8.3	16
57	Exploring the link between MORF4L1 and risk of breast cancer. *Breast Cancer Research*, **2011**, 13, R40	8.3	16
56	Over-representation of two specific haplotypes among chromosomes harbouring BRCA1 mutations. *European Journal of Human Genetics*, **2003**, 11, 489-92	5.3	16

55	The accumulation of specific amplifications characterizes two different genomic pathways of evolution of familial breast tumors. *Clinical Cancer Research*, **2005**, 11, 8577-84	12.9	16
54	No clinical utility of KRAS variant rs61764370 for ovarian or breast cancer. *Gynecologic Oncology*, **2016**, 141, 386-401	4.9	15
53	Association of breast cancer risk in BRCA1 and BRCA2 mutation carriers with genetic variants showing differential allelic expression: identification of a modifier of breast cancer risk at locus 11q22.3. *Breast Cancer Research and Treatment*, **2017**, 161, 117-134	4.4	15
52	Haplotype structure in Ashkenazi Jewish BRCA1 and BRCA2 mutation carriers. *Human Genetics*, **2011**, 130, 685-99	6.3	15
51	Deletion at 6q24.2-26 predicts longer survival of high-grade serous epithelial ovarian cancer patients. *Molecular Oncology*, **2015**, 9, 422-36	7.9	14
50	sigma(54) Promoters control expression of genes encoding the hook and basal body complex in Rhodobacter sphaeroides. *Journal of Bacteriology*, **2000**, 182, 5787-92	3.5	14
49	Genetic and clinical analysis in 10 Spanish patients with multiple endocrine neoplasia type 1. *European Journal of Human Genetics*, **1999**, 7, 585-9	5.3	14
48	DNA copy number profiling reveals extensive genomic loss in hereditary BRCA1 and BRCA2 ovarian carcinomas. *British Journal of Cancer*, **2013**, 108, 1732-42	8.7	13
47	Gene expression profiling integrated into network modelling reveals heterogeneity in the mechanisms of BRCA1 tumorigenesis. *British Journal of Cancer*, **2009**, 101, 1469-80	8.7	13
46	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). *British Journal of Cancer*, **2009**, 101, 2048-54	8.7	13
45	The rs12975333 variant in the miR-125a and breast cancer risk in Germany, Italy, Australia and Spain. *Journal of Medical Genetics*, **2011**, 48, 703-4	5.8	13
44	Haplotype analysis of the BRCA2 9254delATCAT recurrent mutation in breast/ovarian cancer families from Spain. *Human Mutation*, **2003**, 21, 452	4.7	13
43	Molecular insights into the OGG1 gene, a cancer risk modifier in BRCA1 and BRCA2 mutations carriers. *Oncotarget*, **2016**, 7, 25815-25	3.3	13
42	Oral Contraceptive Use and Breast Cancer Risk: Retrospective and Prospective Analyses From a BRCA1 and BRCA2 Mutation Carrier Cohort Study. *JNCI Cancer Spectrum*, **2018**, 2, pky023	4.6	13
41	Almost 2% of Spanish breast cancer families are associated to germline pathogenic mutations in the ATM gene. *Breast Cancer Research and Treatment*, **2017**, 161, 597-604	4.4	12
40	The :p.Arg658* truncating variant is associated with risk of triple-negative breast cancer. *Npj Breast Cancer*, **2019**, 5, 38	7.8	12
39	Pathological features of breast and ovarian cancers in RAD51C germline mutation carriers. *Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin*, **2014**, 465, 365-9	5.1	12
38	An evaluation of the polymorphisms Ins16bp and Arg72Pro in p53 as breast cancer risk modifiers in BRCA1 and BRCA2 mutation carriers. *British Journal of Cancer*, **2008**, 99, 974-7	8.7	12

37	European multicenter study on LOH of APOC3 at 11q23 in 766 breast cancer patients: relation to clinical variables. Breast Cancer Somatic Genetics Consortium. *British Journal of Cancer*, **1999**, 80, 879-82	8.7	12
36	Evidence for a link between TNFRSF11A and risk of breast cancer. *Breast Cancer Research and Treatment*, **2011**, 129, 947-54	4.4	11
35	Transcriptional characteristics of familial non-BRCA1/BRCA2 breast tumors. *International Journal of Cancer*, **2011**, 128, 2635-44	7.5	11
34	The highly prevalent BRCA2 mutation c.2808_2811del (3036delACAA) is located in a mutational hotspot and has multiple origins. *Carcinogenesis*, **2013**, 34, 2505-11	4.6	10
33	RECQL5: Another DNA helicase potentially involved in hereditary breast cancer susceptibility. *Human Mutation*, **2019**, 40, 566-577	4.7	10
32	The Influence of Number and Timing of Pregnancies on Breast Cancer Risk for Women With or Mutations. *JNCI Cancer Spectrum*, **2018**, 2, pky078	4.6	10
31	Transcriptome-wide association study of breast cancer risk by estrogen-receptor status. *Genetic Epidemiology*, **2020**, 44, 442-468	2.6	9
30	Alcohol Consumption, Cigarette Smoking, and Risk of Breast Cancer for and Mutation Carriers: Results from The BRCA1 and BRCA2 Cohort Consortium. *Cancer Epidemiology Biomarkers and Prevention*, **2020**, 29, 368-378	4	9
29	Hypermethylation of P16ink4a and P15ink4b genes as a marker of disease in the follow-up of non-Hodgkin© lymphomas. *British Journal of Haematology*, **2000**, 109, 97-103	4.5	9
28	Oral contraceptive use and ovarian cancer risk for BRCA1/2 mutation carriers: an international cohort study. *American Journal of Obstetrics and Gynecology*, **2021**, 225, 51.e1-51.e17	6.4	9
27	A rapid and easy method for multiple endocrine neoplasia type 1 mutation detection using conformation-sensitive gel electrophoresis. *Journal of Human Genetics*, **2002**, 47, 190-5	4.3	8
26	Cross-Cancer Genome-Wide Association Study of Endometrial Cancer and Epithelial Ovarian Cancer Identifies Genetic Risk Regions Associated with Risk of Both Cancers. *Cancer Epidemiology Biomarkers and Prevention*, **2021**, 30, 217-228	4	7
25	The Spectrum of Protein Truncating Variants in European Breast Cancer Cases. *Cancers*, **2020**, 12,	6.6	7
24	BRCA1 and BRCA2 mutations in males with familial breast and ovarian cancer syndrome. Results of a Spanish multicenter study. *Familial Cancer*, **2015**, 14, 505-13	3	7
23	Genetic variation in the DNA glycosylase gene is associated with oxidative DNA damage in mutation carriers. *Oncotarget*, **2017**, 8, 114626-114636	3.3	7
22	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). *Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin*, **2020**, 476, 195-207	5.1	7
21	Fine-Scale Mapping at 9p22.2 Identifies Candidate Causal Variants That Modify Ovarian Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. *PLoS ONE*, **2016**, 11, e0158801	3.7	7
20	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. *British Journal of Cancer*, **2011**, 104, 1356-61	8.7	6

19	Mutation analysis of the BRCA2 gene in breast/ovarian cancer Spanish families: identification of two new mutations. *Cancer Letters*, **1997**, 121, 115-8	9.9	6
18	Clustering of cancer-related mutations in a subset of BRCA1 alleles: a study in the Spanish population. *International Journal of Cancer*, **2002**, 100, 618-9	7.5	6
17	No mutations in the XRCC2 gene in BRCA1/2-negative high-risk breast cancer families. *International Journal of Cancer*, **2003**, 103, 136-7	7.5	6
16	Mutational analysis of telomere genes in BRCA1/2-negative breast cancer families with very short telomeres. *Breast Cancer Research and Treatment*, **2012**, 134, 1337-43	4.4	5
15	First international workshop of the ATM and cancer risk group (4-5 December 2019). *Familial Cancer* , **2021**, 1	3	5
14	Characterisation of the novel deleterious RAD51C p.Arg312Trp variant and prioritisation criteria for functional analysis of RAD51C missense changes. *British Journal of Cancer*, **2017**, 117, 1048-1062	8.7	4
13	The 12 base pair duplication/insertion alteration could be a regulatory mutation. *Journal of Medical Genetics*, **1997**, 34, 592-3	5.8	4
12	Pathology of Tumors Associated With Pathogenic Germline Variants in 9 Breast Cancer Susceptibility Genes.. *JAMA Oncology*, **2022**,	13.4	4
11	Small molecule inhibitor of OGG1 blocks oxidative DNA damage repair at telomeres and potentiates methotrexate anticancer effects. *Scientific Reports*, **2021**, 11, 3490	4.9	4
10	A case-only study to identify genetic modifiers of breast cancer risk for BRCA1/BRCA2 mutation carriers. *Nature Communications*, **2021**, 12, 1078	17.4	4
9	Evaluation of chromosome 6p22 as a breast cancer risk modifier locus in a follow-up study of BRCA2 mutation carriers. *Breast Cancer Research and Treatment*, **2012**, 136, 295-302	4.4	3
8	Molecular analysis of the BRCA2 gene in 16 breast/ovarian cancer Spanish families. *Clinical Genetics*, **1998**, 54, 142-7	4	3
7	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. *Human Molecular Genetics*, **2016**, 25, 5287-5299	5.6	2
6	Mutant BRCA1 alleles transmission: different approaches and different biases. *International Journal of Cancer*, **2005**, 113, 166-7	7.5	2
5	Identification by comparative genomic hybridization of genetic changes involved in tumoral progression of a T-cell non-Hodgkin lymphoma. *Cancer Genetics and Cytogenetics*, **2000**, 117, 41-4		2
4	Fine-mapping of 150 breast cancer risk regions identifies 178 high confidence target genes		2
3	A common SNP in the UNG gene decreases ovarian cancer risk in BRCA2 mutation carriers. *Molecular Oncology*, **2019**, 13, 1110-1120	7.9	2
2	Polygenic Risk Modelling for Prediction of Epithelial Ovarian Cancer Risk		1

1	A Collaborative Effort to Define Classification Criteria for ATM Variants in Hereditary Cancer Patients. *Clinical Chemistry*, **2021**, 67, 518-533	5.5	1