Melissa C Southey

List of Publications by Citations

337 papers	23,606 citations	78 h-index	145 g-index
351 ext. papers	28,352 ext. citations	9.8 avg, IF	5.17 L-index

#	Paper	IF	Citations
337	Large-scale genotyping identifies 41 new loci associated with breast cancer risk. *Nature Genetics*, **2013**, 45, 353-61, 361e1-2	36.3	813
336	Multiple newly identified loci associated with prostate cancer susceptibility. *Nature Genetics*, **2008**, 40, 316-21	36.3	722
335	Association analysis identifies 65 new breast cancer risk loci. *Nature*, **2017**, 551, 92-94	50.4	643
334	Subtyping of breast cancer by immunohistochemistry to investigate a relationship between subtype and short and long term survival: a collaborative analysis of data for 10,159 cases from 12 studies. *PLoS Medicine*, **2010**, 7, e1000279	11.6	616
333	Breast-cancer risk in families with mutations in PALB2. *New England Journal of Medicine*, **2014**, 371, 497-506	59.2	576
332	A common coding variant in CASP8 is associated with breast cancer risk. *Nature Genetics*, **2007**, 39, 352-8	36.3	557
331	Iron-overload-related disease in HFE hereditary hemochromatosis. *New England Journal of Medicine* , **2008**, 358, 221-30	59.2	516
330	Associations of breast cancer risk factors with tumor subtypes: a pooled analysis from the Breast Cancer Association Consortium studies. *Journal of the National Cancer Institute*, **2011**, 103, 250-63	9.7	513
329	A genome-wide association study identifies colorectal cancer susceptibility loci on chromosomes 10p14 and 8q23.3. *Nature Genetics*, **2008**, 40, 623-30	36.3	463
328	Multiple independent variants at the TERT locus are associated with telomere length and risks of breast and ovarian cancer. *Nature Genetics*, **2013**, 45, 371-84, 384e1-2	36.3	422
327	Identification of 23 new prostate cancer susceptibility loci using the iCOGS custom genotyping array. *Nature Genetics*, **2013**, 45, 385-91, 391e1-2	36.3	413
326	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
325	Genome-wide association analysis of more than 120,000 individuals identifies 15 new susceptibility loci for breast cancer. *Nature Genetics*, **2015**, 47, 373-80	36.3	406
324	Newly discovered breast cancer susceptibility loci on 3p24 and 17q23.2. *Nature Genetics*, **2009**, 41, 585-90	36.3	393
323	Polygenic Risk Scores for Prediction of Breast Cancer and Breast Cancer Subtypes. *American Journal of Human Genetics*, **2019**, 104, 21-34	11	363
322	Identification of seven new prostate cancer susceptibility loci through a genome-wide association study. *Nature Genetics*, **2009**, 41, 1116-21	36.3	360
321	Association analyses of more than 140,000 men identify 63 new prostate cancer susceptibility loci. *Nature Genetics*, **2018**, 50, 928-936	36.3	340

320	Genome-wide association studies identify four ER negative-specific breast cancer risk loci. *Nature Genetics*, **2013**, 45, 392-8, 398e1-2	36.3	327
319	Prediction of breast cancer risk based on profiling with common genetic variants. *Journal of the National Cancer Institute*, **2015**, 107,	9.7	324
318	Heterogeneity of breast cancer associations with five susceptibility loci by clinical and pathological characteristics. *PLoS Genetics*, **2008**, 4, e1000054	6	280
317	GWAS meta-analysis and replication identifies three new susceptibility loci for ovarian cancer. *Nature Genetics*, **2013**, 45, 362-70, 370e1-2	36.3	267
316	Genomic analyses identify hundreds of variants associated with age at menarche and support a role for puberty timing in cancer risk. *Nature Genetics*, **2017**, 49, 834-841	36.3	257
315	A common variant at the TERT-CLPTM1L locus is associated with estrogen receptor-negative breast cancer. *Nature Genetics*, **2011**, 43, 1210-4	36.3	253
314	Multiple loci on 8q24 associated with prostate cancer susceptibility. *Nature Genetics*, **2009**, 41, 1058-60	36.3	252
313	Colorectal and other cancer risks for carriers and noncarriers from families with a DNA mismatch repair gene mutation: a prospective cohort study. *Journal of Clinical Oncology*, **2012**, 30, 958-64	2.2	245
312	Seven prostate cancer susceptibility loci identified by a multi-stage genome-wide association study. *Nature Genetics*, **2011**, 43, 785-91	36.3	243
311	The Breast Cancer Family Registry: an infrastructure for cooperative multinational, interdisciplinary and translational studies of the genetic epidemiology of breast cancer. *Breast Cancer Research*, **2004**, 6, R375-89	8.3	239
310	Large-scale genomic analyses link reproductive aging to hypothalamic signaling, breast cancer susceptibility and BRCA1-mediated DNA repair. *Nature Genetics*, **2015**, 47, 1294-1303	36.3	226
309	The histologic phenotypes of breast carcinoma occurring before age 40 years in women with and without BRCA1 or BRCA2 germline mutations. *Cancer*, **1998**, 83, 2335-2345	6.4	211
308	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
307	Metachronous colorectal cancer risk for mismatch repair gene mutation carriers: the advantage of more extensive colon surgery. *Gut*, **2011**, 60, 950-7	19.2	192
306	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. *Nature Genetics*, **2017**, 49, 680-691	36.3	190
305	Breast Cancer Risk From Modifiable and Nonmodifiable Risk Factors Among White Women in the United States. *JAMA Oncology*, **2016**, 2, 1295-1302	13.4	189
304	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. *Nature Genetics*, **2017**, 49, 1767-1778	36.3	186
303	Discovery of common and rare genetic risk variants for colorectal cancer. *Nature Genetics*, **2019**, 51, 76-87	36.3	177

302	Use of molecular tumor characteristics to prioritize mismatch repair gene testing in early-onset colorectal cancer. *Journal of Clinical Oncology*, **2005**, 23, 6524-32	2.2	172
301	Functional variants at the 11q13 risk locus for breast cancer regulate cyclin D1 expression through long-range enhancers. *American Journal of Human Genetics*, **2013**, 92, 489-503	11	167
300	Rare variants in the ATM gene and risk of breast cancer. *Breast Cancer Research*, **2011**, 13, R73	8.3	151
299	A meta-analysis of genome-wide association studies of breast cancer identifies two novel susceptibility loci at 6q14 and 20q11. *Human Molecular Genetics*, **2012**, 21, 5373-84	5.6	143
298	Breast Cancer Risk Genes - Association Analysis in More than 113,000 Women. *New England Journal of Medicine*, **2021**, 384, 428-439	59.2	143
297	Low penetrance breast cancer susceptibility loci are associated with specific breast tumor subtypes: findings from the Breast Cancer Association Consortium. *Human Molecular Genetics*, **2011** , 20, 3289-303	5.6	140
296	Rare, evolutionarily unlikely missense substitutions in ATM confer increased risk of breast cancer. *American Journal of Human Genetics*, **2009**, 85, 427-46	11	140
295	Familial risks, early-onset breast cancer, and BRCA1 and BRCA2 germline mutations. *Journal of the National Cancer Institute*, **2003**, 95, 448-57	9.7	137
294	Cancer risks for mismatch repair gene mutation carriers: a population-based early onset case-family study. *Clinical Gastroenterology and Hepatology*, **2006**, 4, 489-98	6.9	136
293	CHEK2*1100delC heterozygosity in women with breast cancer associated with early death, breast cancer-specific death, and increased risk of a second breast cancer. *Journal of Clinical Oncology*, **2012**, 30, 4308-16	2.2	134
292	BRCA2 mutation-associated breast cancers exhibit a distinguishing phenotype based on morphology and molecular profiles from tissue microarrays. *American Journal of Surgical Pathology*, **2007**, 31, 121-8	6.7	128
291	Epigenetic analysis leads to identification of HNF1B as a subtype-specific susceptibility gene for ovarian cancer. *Nature Communications*, **2013**, 4, 1628	17.4	124
290	Constitutional methylation of the BRCA1 promoter is specifically associated with BRCA1 mutation-associated pathology in early-onset breast cancer. *Cancer Prevention Research*, **2011**, 4, 23-33	3.2	121
289	PALB2, CHEK2 and ATM rare variants and cancer risk: data from COGS. *Journal of Medical Genetics*, **2016**, 53, 800-811	5.8	121
288	Breast cancer prognosis in BRCA1 and BRCA2 mutation carriers: an International Prospective Breast Cancer Family Registry population-based cohort study. *Journal of Clinical Oncology*, **2012**, 30, 19-26	2.2	116
287	Oral contraceptive use and risk of early-onset breast cancer in carriers and noncarriers of BRCA1 and BRCA2 mutations. *Cancer Epidemiology Biomarkers and Prevention*, **2005**, 14, 350-6	4	113
286	Evidence of gene-environment interactions between common breast cancer susceptibility loci and established environmental risk factors. *PLoS Genetics*, **2013**, 9, e1003284	6	112
285	A meta-analysis of genome-wide association studies to identify prostate cancer susceptibility loci associated with aggressive and non-aggressive disease. *Human Molecular Genetics*, **2013**, 22, 408-15	5.6	109

284	Genome-wide association study identifies multiple susceptibility loci for diffuse large B cell lymphoma. *Nature Genetics*, **2014**, 46, 1233-8	36.3	108
283	Age- and Tumor Subtype-Specific Breast Cancer Risk Estimates for CHEK2*1100delC Carriers. *Journal of Clinical Oncology*, **2016**, 34, 2750-60	2.2	107
282	Colorectal carcinomas with KRAS mutation are associated with distinctive morphological and molecular features. *Modern Pathology*, **2013**, 26, 825-34	9.8	106
281	Genome-Wide Meta-Analyses of Breast, Ovarian, and Prostate Cancer Association Studies Identify Multiple New Susceptibility Loci Shared by at Least Two Cancer Types. *Cancer Discovery*, **2016**, 6, 1052-67	24.4	104
280	PIK3CA activating mutation in colorectal carcinoma: associations with molecular features and survival. *PLoS ONE*, **2013**, 8, e65479	3.7	102
279	A transcriptome-wide association study of 229,000 women identifies new candidate susceptibility genes for breast cancer. *Nature Genetics*, **2018**, 50, 968-978	36.3	101
278	Adaptive evolution of the tumour suppressor BRCA1 in humans and chimpanzees. Australian Breast Cancer Family Study. *Nature Genetics*, **2000**, 25, 410-3	36.3	98
277	Common variants in ZNF365 are associated with both mammographic density and breast cancer risk. *Nature Genetics*, **2011**, 43, 185-7	36.3	96
276	HFE C282Y homozygotes are at increased risk of breast and colorectal cancer. *Hepatology*, **2010**, 51, 1311-8	11.2	95
275	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
274	19p13.1 is a triple-negative-specific breast cancer susceptibility locus. *Cancer Research*, **2012**, 72, 1795-803	10.1	93
273	Common breast cancer susceptibility variants in LSP1 and RAD51L1 are associated with mammographic density measures that predict breast cancer risk. *Cancer Epidemiology Biomarkers and Prevention*, **2012**, 21, 1156-66	4	92
272	Identification of a BRCA2-specific modifier locus at 6p24 related to breast cancer risk. *PLoS Genetics*, **2013**, 9, e1003173	6	90
271	Risk of estrogen receptor-positive and -negative breast cancer and single-nucleotide polymorphism 2q35-rs13387042. *Journal of the National Cancer Institute*, **2009**, 101, 1012-8	9.7	90
270	After BRCA1 and BRCA2-what next? Multifactorial segregation analyses of three-generation, population-based Australian families affected by female breast cancer. *American Journal of Human Genetics*, **2001**, 68, 420-31	11	90
269	A PALB2 mutation associated with high risk of breast cancer. *Breast Cancer Research*, **2010**, 12, R109	8.3	89
268	Ethnicity and risk for colorectal cancers showing somatic BRAF V600E mutation or CpG island methylator phenotype. *Cancer Epidemiology Biomarkers and Prevention*, **2008**, 17, 1774-80	4	89
267	Evidence that breast cancer risk at the 2q35 locus is mediated through IGFBP5 regulation. *Nature Communications*, **2014**, 4, 4999	17.4	87

266	Identification and molecular characterization of a new ovarian cancer susceptibility locus at 17q21.31. *Nature Communications*, **2013**, 4, 1627	17.4	85
265	Genome-wide association study identifies multiple loci associated with both mammographic density and breast cancer risk. *Nature Communications*, **2014**, 5, 5303	17.4	84
264	Penetrance analysis of the PALB2 c.1592delT founder mutation. *Clinical Cancer Research*, **2008**, 14, 4667-71	12.9	84
263	No evidence that protein truncating variants in BRIP1 are associated with breast cancer risk: implications for gene panel testing. *Journal of Medical Genetics*, **2016**, 53, 298-309	5.8	83
262	Fine-scale mapping of the FGFR2 breast cancer risk locus: putative functional variants differentially bind FOXA1 and E2F1. *American Journal of Human Genetics*, **2013**, 93, 1046-60	11	80
261	Genetically Predicted Body Mass Index and Breast Cancer Risk: Mendelian Randomization Analyses of Data from 145,000 Women of European Descent. *PLoS Medicine*, **2016**, 13, e1002105	11.6	80
260	Rare key functional domain missense substitutions in MRE11A, RAD50, and NBN contribute to breast cancer susceptibility: results from a Breast Cancer Family Registry case-control mutation-screening study. *Breast Cancer Research*, **2014**, 16, R58	8.3	78
259	Imputation and subset-based association analysis across different cancer types identifies multiple independent risk loci in the TERT-CLPTM1L region on chromosome 5p15.33. *Human Molecular Genetics*, **2014**, 23, 6616-33	5.6	77
258	Genome-wide association study identifies 32 novel breast cancer susceptibility loci from overall and subtype-specific analyses. *Nature Genetics*, **2020**, 52, 572-581	36.3	76
257	Epigenome-wide methylation in DNA from peripheral blood as a marker of risk for breast cancer. *Breast Cancer Research and Treatment*, **2014**, 148, 665-73	4.4	75
256	Population-based estimate of the contribution of TP53 mutations to subgroups of early-onset breast cancer: Australian Breast Cancer Family Study. *Cancer Research*, **2010**, 70, 4795-800	10.1	75
255	The role of genetic breast cancer susceptibility variants as prognostic factors. *Human Molecular Genetics*, **2012**, 21, 3926-39	5.6	75
254	Height and Breast Cancer Risk: Evidence From Prospective Studies and Mendelian Randomization. *Journal of the National Cancer Institute*, **2015**, 107,	9.7	74
253	Common genetic variants and modification of penetrance of BRCA2-associated breast cancer. *PLoS Genetics*, **2010**, 6, e1001183	6	74
252	Assessing interactions between the associations of common genetic susceptibility variants, reproductive history and body mass index with breast cancer risk in the breast cancer association consortium: a combined case-control study. *Breast Cancer Research*, **2010**, 12, R110	8.3	74
251	Identification of nine new susceptibility loci for endometrial cancer. *Nature Communications*, **2018**, 9, 3166	17.4	70
250	Comparison of DNA- and RNA-based methods for detection of truncating BRCA1 mutations. *Human Mutation*, **2002**, 20, 65-73	4.7	69
249	Breast Cancer Risk Prediction Using Clinical Models and 77 Independent Risk-Associated SNPs for Women Aged Under 50 Years: Australian Breast Cancer Family Registry. *Cancer Epidemiology Biomarkers and Prevention*, **2016**, 25, 359-65	4	68

248	Associations of common variants at 1p11.2 and 14q24.1 (RAD51L1) with breast cancer risk and heterogeneity by tumor subtype: findings from the Breast Cancer Association Consortium. *Human Molecular Genetics*, **2011**, 20, 4693-706	5.6	66
247	Cross-Cancer Genome-Wide Analysis of Lung, Ovary, Breast, Prostate, and Colorectal Cancer Reveals Novel Pleiotropic Associations. *Cancer Research*, **2016**, 76, 5103-14	10.1	66
246	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
245	Rare, evolutionarily unlikely missense substitutions in CHEK2 contribute to breast cancer susceptibility: results from a breast cancer family registry case-control mutation-screening study. *Breast Cancer Research*, **2011**, 13, R6	8.3	65
244	The natural history of serum iron indices for HFE C282Y homozygosity associated with hereditary hemochromatosis. *Gastroenterology*, **2008**, 135, 1945-52	13.3	65
243	HFE C282Y/H63D compound heterozygotes are at low risk of hemochromatosis-related morbidity. *Hepatology*, **2009**, 50, 94-101	11.2	64
242	A genome-wide association study of early-onset breast cancer identifies PFKM as a novel breast cancer gene and supports a common genetic spectrum for breast cancer at any age. *Cancer Epidemiology Biomarkers and Prevention*, **2014**, 23, 658-69	4	63
241	Common genetic variants associated with breast cancer and mammographic density measures that predict disease. *Cancer Research*, **2010**, 70, 1449-58	10.1	63
240	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
239	Trans-ancestry genome-wide association meta-analysis of prostate cancer identifies new susceptibility loci and informs genetic risk prediction. *Nature Genetics*, **2021**, 53, 65-75	36.3	62
238	Fine-scale mapping of the 5q11.2 breast cancer locus reveals at least three independent risk variants regulating MAP3K1. *American Journal of Human Genetics*, **2015**, 96, 5-20	11	59
237	Body size and risk for colorectal cancers showing BRAF mutations or microsatellite instability: a pooled analysis. *International Journal of Epidemiology*, **2012**, 41, 1060-72	7.8	59
236	Are the so-called low penetrance breast cancer genes, ATM, BRIP1, PALB2 and CHEK2, high risk for women with strong family histories?. *Breast Cancer Research*, **2008**, 10, 208	8.3	58
235	Fine-mapping of prostate cancer susceptibility loci in a large meta-analysis identifies candidate causal variants. *Nature Communications*, **2018**, 9, 2256	17.4	57
234	Refinement of the basis and impact of common 11q23.1 variation to the risk of developing colorectal cancer. *Human Molecular Genetics*, **2008**, 17, 3720-7	5.6	57
233	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. *Nature Genetics*, **2020**, 52, 56-73	36.3	56
232	Combined genetic and splicing analysis of BRCA1 c.[594-2A>C; 641A>G] highlights the relevance of naturally occurring in-frame transcripts for developing disease gene variant classification algorithms. *Human Molecular Genetics*, **2016**, 25, 2256-2268	5.6	55
231	Multiple novel prostate cancer susceptibility signals identified by fine-mapping of known risk loci among Europeans. *Human Molecular Genetics*, **2015**, 24, 5589-602	5.6	54

230	Joint associations of a polygenic risk score and environmental risk factors for breast cancer in the Breast Cancer Association Consortium. *International Journal of Epidemiology*, **2018**, 47, 526-536	7.8	53
229	Functional mechanisms underlying pleiotropic risk alleles at the 19p13.1 breast-ovarian cancer susceptibility locus. *Nature Communications*, **2016**, 7, 12675	17.4	53
228	Five polymorphisms and breast cancer risk: results from the Breast Cancer Association Consortium. *Cancer Epidemiology Biomarkers and Prevention*, **2009**, 18, 1610-6	4	53
227	Heritable DNA methylation marks associated with susceptibility to breast cancer. *Nature Communications*, **2018**, 9, 867	17.4	52
226	Associations of obesity and circulating insulin and glucose with breast cancer risk: a Mendelian randomization analysis. *International Journal of Epidemiology*, **2019**, 48, 795-806	7.8	52
225	Genetic modifiers of CHEK2*1100delC-associated breast cancer risk. *Genetics in Medicine*, **2017**, 19, 599-603	6.3	51
224	Causal effect of smoking on DNA methylation in peripheral blood: a twin and family study. *Clinical Epigenetics*, **2018**, 10, 18	7.7	50
223	Mammographic breast density and breast cancer: evidence of a shared genetic basis. *Cancer Research*, **2012**, 72, 1478-84	10.1	50
222	Hypomorphic Missense Variants Confer Moderate Risks of Breast Cancer. *Cancer Research*, **2017**, 77, 2789-2799	10.1	49
221	BRCA1 and BRCA2 mutation carriers in the Breast Cancer Family Registry: an open resource for collaborative research. *Breast Cancer Research and Treatment*, **2009**, 116, 379-86	4.4	49
220	A novel association between a SNP in CYBRD1 and serum ferritin levels in a cohort study of HFE hereditary haemochromatosis. *British Journal of Haematology*, **2009**, 147, 140-9	4.5	49
219	Comparison of 6q25 breast cancer hits from Asian and European Genome Wide Association Studies in the Breast Cancer Association Consortium (BCAC). *PLoS ONE*, **2012**, 7, e42380	3.7	49
218	Common non-synonymous SNPs associated with breast cancer susceptibility: findings from the Breast Cancer Association Consortium. *Human Molecular Genetics*, **2014**, 23, 6096-111	5.6	48
217	Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. *Nature Communications*, **2019**, 10, 1741	17.4	47
216	Cumulative Burden of Colorectal Cancer-Associated Genetic Variants Is More Strongly Associated With Early-Onset vs Late-Onset Cancer. *Gastroenterology*, **2020**, 158, 1274-1286.e12	13.3	47
215	Risk Analysis of Prostate Cancer in PRACTICAL, a Multinational Consortium, Using 25 Known Prostate Cancer Susceptibility Loci. *Cancer Epidemiology Biomarkers and Prevention*, **2015**, 24, 1121-9	4	46
214	Fine-mapping of the HNF1B multicancer locus identifies candidate variants that mediate endometrial cancer risk. *Human Molecular Genetics*, **2015**, 24, 1478-92	5.6	46
213	Shared heritability and functional enrichment across six solid cancers. *Nature Communications*, **2019**, 10, 431	17.4	45

212	Novel Associations between Common Breast Cancer Susceptibility Variants and Risk-Predicting Mammographic Density Measures. *Cancer Research*, **2015**, 75, 2457-67	10.1	45
211	Association between a germline OCA2 polymorphism at chromosome 15q13.1 and estrogen receptor-negative breast cancer survival. *Journal of the National Cancer Institute*, **2010**, 102, 650-62	9.7	45
210	ELAC2/HPC2 polymorphisms, prostate-specific antigen levels, and prostate cancer. *Journal of the National Cancer Institute*, **2003**, 95, 818-24	9.7	45
209	Adult body mass index and risk of ovarian cancer by subtype: a Mendelian randomization study. *International Journal of Epidemiology*, **2016**, 45, 884-95	7.8	45
208	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
207	Prediction of individual genetic risk to prostate cancer using a polygenic score. *Prostate*, **2015**, 75, 1467-74	4.2	43
206	Is uptake of genetic testing for colorectal cancer influenced by knowledge of insurance implications?. *Medical Journal of Australia*, **2009**, 191, 255-8	4	43
205	Evidence that the 5p12 Variant rs10941679 Confers Susceptibility to Estrogen-Receptor-Positive Breast Cancer through FGF10 and MRPS30 Regulation. *American Journal of Human Genetics*, **2016**, 99, 903-911	11	43
204	Identification of fifteen novel germline variants in the BRCA1 3'UTR reveals a variant in a breast cancer case that introduces a functional miR-103 target site. *Human Mutation*, **2012**, 33, 1665-75	4.7	42
203	A role for XRCC2 gene polymorphisms in breast cancer risk and survival. *Journal of Medical Genetics*, **2011**, 48, 477-84	5.8	42
202	Estrogen receptor polymorphism at codon 325 and risk of breast cancer in women before age forty. *Journal of the National Cancer Institute*, **1998**, 90, 532-6	9.7	42
201	After hMSH2 and hMLH1--what next? Analysis of three-generational, population-based, early-onset colorectal cancer families. *International Journal of Cancer*, **2002**, 102, 166-71	7.5	41
200	Cis-eQTL analysis and functional validation of candidate susceptibility genes for high-grade serous ovarian cancer. *Nature Communications*, **2015**, 6, 8234	17.4	40
199	Genetically predicted longer telomere length is associated with increased risk of B-cell lymphoma subtypes. *Human Molecular Genetics*, **2016**, 25, 1663-76	5.6	39
198	Rare mutations in RINT1 predispose carriers to breast and Lynch syndrome-spectrum cancers. *Cancer Discovery*, **2014**, 4, 804-15	24.4	39
197	Identification of novel genetic markers of breast cancer survival. *Journal of the National Cancer Institute*, **2015**, 107,	9.7	38
196	Genetic predisposition to in situ and invasive lobular carcinoma of the breast. *PLoS Genetics*, **2014**, 10, e1004285	6	38
195	Atlas of prostate cancer heritability in European and African-American men pinpoints tissue-specific regulation. *Nature Communications*, **2016**, 7, 10979	17.4	37

194	MicroRNA related polymorphisms and breast cancer risk. *PLoS ONE*, **2014**, 9, e109973	3.7	37
193	A high-plex PCR approach for massively parallel sequencing. *BioTechniques*, **2013**, 55, 69-74	2.5	37
192	PALB2 and breast cancer: ready for clinical translation!. *The Application of Clinical Genetics*, **2013**, 6, 43-52	3.1	37
191	Prevalence of PALB2 mutations in Australasian multiple-case breast cancer families. *Breast Cancer Research*, **2013**, 15, R17	8.3	36
190	Fine-mapping identifies two additional breast cancer susceptibility loci at 9q31.2. *Human Molecular Genetics*, **2015**, 24, 2966-84	5.6	36
189	Log odds of carrying an Ancestral Mutation in BRCA1 or BRCA2 for a Defined personal and family history in an Ashkenazi Jewish woman (LAMBDA). *Breast Cancer Research*, **2003**, 5, R206-16	8.3	36
188	Identification and characterization of novel associations in the CASP8/ALS2CR12 region on chromosome 2 with breast cancer risk. *Human Molecular Genetics*, **2015**, 24, 285-98	5.6	35
187	A systematic approach to analysing gene-gene interactions: polymorphisms at the microsomal epoxide hydrolase EPHX and glutathione S-transferase GSTM1, GSTT1, and GSTP1 loci and breast cancer risk. *Cancer Epidemiology Biomarkers and Prevention*, **2007**, 16, 769-74	4	35
186	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
185	11q13 is a susceptibility locus for hormone receptor positive breast cancer. *Human Mutation*, **2012**, 33, 1123-32	4.7	33
184	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
183	Cell-type-specific enrichment of risk-associated regulatory elements at ovarian cancer susceptibility loci. *Human Molecular Genetics*, **2015**, 24, 3595-607	5.6	32
182	Breast cancer risk prediction using a polygenic risk score in the familial setting: a prospective study from the Breast Cancer Family Registry and kConFab. *Genetics in Medicine*, **2017**, 19, 30-35	8.1	31
181	Role of tumour molecular and pathology features to estimate colorectal cancer risk for first-degree relatives. *Gut*, **2015**, 64, 101-10	19.2	31
180	Missense variants in ATM in 26,101 breast cancer cases and 29,842 controls. *Cancer Epidemiology Biomarkers and Prevention*, **2010**, 19, 2143-51	4	31
179	Genetic predisposition to ductal carcinoma in situ of the breast. *Breast Cancer Research*, **2016**, 18, 22	8.3	31
178	Candidate locus analysis of the TERT-CLPTM1L cancer risk region on chromosome 5p15 identifies multiple independent variants associated with endometrial cancer risk. *Human Genetics*, **2015**, 134, 231-45	6.3	30
177	Explaining variance in the cumulus mammographic measures that predict breast cancer risk: a twins and sisters study. *Cancer Epidemiology Biomarkers and Prevention*, **2013**, 22, 2395-403	4	29

176	Genome-wide association study of germline variants and breast cancer-specific mortality. *British Journal of Cancer*, **2019**, 120, 647-657	8.7	28
175	A large-scale assessment of two-way SNP interactions in breast cancer susceptibility using 46,450 cases and 42,461 controls from the breast cancer association consortium. *Human Molecular Genetics*, **2014**, 23, 1934-46	5.6	28
174	Association of breast cancer risk loci with breast cancer survival. *International Journal of Cancer*, **2015**, 137, 2837-45	7.5	28
173	Identification of a novel percent mammographic density locus at 12q24. *Human Molecular Genetics*, **2012**, 21, 3299-305	5.6	28
172	Genetic insights into biological mechanisms governing human ovarian ageing. *Nature*, **2021**, 596, 393-397	50.4	28
171	Tumor testing to identify lynch syndrome in two Australian colorectal cancer cohorts. *Journal of Gastroenterology and Hepatology (Australia)*, **2017**, 32, 427-438	4	27
170	Genome-wide association of familial prostate cancer cases identifies evidence for a rare segregating haplotype at 8q24.21. *Human Genetics*, **2016**, 135, 923-38	6.3	27
169	Confirmation of 5p12 as a susceptibility locus for progesterone-receptor-positive, lower grade breast cancer. *Cancer Epidemiology Biomarkers and Prevention*, **2011**, 20, 2222-31	4	27
168	Polymorphisms in a Putative Enhancer at the 10q21.2 Breast Cancer Risk Locus Regulate NRBF2 Expression. *American Journal of Human Genetics*, **2015**, 97, 22-34	11	26
167	Reproductive profiles and risk of breast cancer subtypes: a multi-center case-only study. *Breast Cancer Research*, **2017**, 19, 119	8.3	26
166	Evidence of a genetic link between endometriosis and ovarian cancer. *Fertility and Sterility*, **2016**, 105, 35-43.e1-10	4.8	26
165	Investigation of gene-environment interactions between 47 newly identified breast cancer susceptibility loci and environmental risk factors. *International Journal of Cancer*, **2015**, 136, E685-96	7.5	26
164	No evidence of MMTV-like env sequences in specimens from the Australian Breast Cancer Family Study. *Breast Cancer Research and Treatment*, **2011**, 125, 229-35	4.4	26
163	Fine-scale mapping of 8q24 locus identifies multiple independent risk variants for breast cancer. *International Journal of Cancer*, **2016**, 139, 1303-1317	7.5	26
162	Using SNP genotypes to improve the discrimination of a simple breast cancer risk prediction model. *Breast Cancer Research and Treatment*, **2013**, 139, 887-96	4.4	25
161	Prostate cancer segregation analyses using 4390 families from UK and Australian population-based studies. *Genetic Epidemiology*, **2010**, 34, 42-50	2.6	25
160	RAD51 and breast cancer susceptibility: no evidence for rare variant association in the Breast Cancer Family Registry study. *PLoS ONE*, **2012**, 7, e52374	3.7	25
159	Network-Based Integration of GWAS and Gene Expression Identifies a HOX-Centric Network Associated with Serous Ovarian Cancer Risk. *Cancer Epidemiology Biomarkers and Prevention*, **2015**, 24, 1574-84	4	24

158	An intergenic risk locus containing an enhancer deletion in 2q35 modulates breast cancer risk by deregulating IGFBP5 expression. *Human Molecular Genetics*, **2016**, 25, 3863-3876	5.6	24
157	Germline mutations in PMS2 and MLH1 in individuals with solitary loss of PMS2 expression in colorectal carcinomas from the Colon Cancer Family Registry Cohort. *BMJ Open*, **2016**, 6, e010293	3	24
156	Identification of new genetic susceptibility loci for breast cancer through consideration of gene-environment interactions. *Genetic Epidemiology*, **2014**, 38, 84-93	2.6	24
155	Common germline polymorphisms associated with breast cancer-specific survival. *Breast Cancer Research*, **2015**, 17, 58	8.3	24
154	Variants in the prostate-specific antigen (PSA) gene and prostate cancer risk, survival, and circulating PSA. *Cancer Epidemiology Biomarkers and Prevention*, **2006**, 15, 1142-7	4	22
153	CYP17 genetic polymorphism, breast cancer, and breast cancer risk factors: Australian Breast Cancer Family Study. *Breast Cancer Research*, **2005**, 7, R513-21	8.3	22
152	Low somatic K-ras mutation frequency in colorectal cancer diagnosed under the age of 45 years. *European Journal of Cancer*, **2006**, 42, 1357-61	7.5	22
151	Common Genetic Variation in Circadian Rhythm Genes and Risk of Epithelial Ovarian Cancer (EOC). *Journal of Genetics and Genome Research*, **2015**, 2,		22
150	Genome-wide association study of subtype-specific epithelial ovarian cancer risk alleles using pooled DNA. *Human Genetics*, **2014**, 133, 481-97	6.3	21
149	Regressive logistic and proportional hazards disease models for within-family analyses of measured genotypes, with application to a CYP17 polymorphism and breast cancer. *Genetic Epidemiology*, **2003**, 24, 161-72	2.6	21
148	Association of breast cancer risk with genetic variants showing differential allelic expression: Identification of a novel breast cancer susceptibility locus at 4q21. *Oncotarget*, **2016**, 7, 80140-80163	3.3	21
147	Lifetime alcohol intake is associated with an increased risk of KRAS+ and BRAF-/KRAS- but not BRAF+ colorectal cancer. *International Journal of Cancer*, **2017**, 140, 1485-1493	7.5	20
146	Blood DNA methylation and breast cancer risk: a meta-analysis of four prospective cohort studies. *Breast Cancer Research*, **2019**, 21, 62	8.3	20
145	Common variants at the CHEK2 gene locus and risk of epithelial ovarian cancer. *Carcinogenesis*, **2015**, 36, 1341-53	4.6	20
144	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
143	A network analysis to identify mediators of germline-driven differences in breast cancer prognosis. *Nature Communications*, **2020**, 11, 312	17.4	20
142	PALB2: research reaching to clinical outcomes for women with breast cancer. *Hereditary Cancer in Clinical Practice*, **2016**, 14, 9	2.3	20
141	Homologous recombination DNA repair defects in associated breast cancers. *Npj Breast Cancer*, **2019**, 5, 23	7.8	20

140	Mutation screening of PALB2 in clinically ascertained families from the Breast Cancer Family Registry. *Breast Cancer Research and Treatment*, **2015**, 149, 547-54	4.4	20
139	Identification of new genetic risk factors for prostate cancer. *Asian Journal of Andrology*, **2009**, 11, 49-55	2.8	20
138	SNP-SNP interaction analysis of NF-B signaling pathway on breast cancer survival. *Oncotarget*, **2015**, 6, 37979-94	3.3	19
137	Joint association of mammographic density adjusted for age and body mass index and polygenic risk score with breast cancer risk. *Breast Cancer Research*, **2019**, 21, 68	8.3	18
136	Mammographic density defined by higher than conventional brightness thresholds better predicts breast cancer risk. *International Journal of Epidemiology*, **2017**, 46, 652-661	7.8	18
135	SNP rs16906252C>T Is an Expression and Methylation Quantitative Trait Locus Associated with an Increased Risk of Developing MGMT-Methylated Colorectal Cancer. *Clinical Cancer Research*, **2016**, 22, 6266-6277	12.9	18
134	HLA Class I and II Diversity Contributes to the Etiologic Heterogeneity of Non-Hodgkin Lymphoma Subtypes. *Cancer Research*, **2018**, 78, 4086-4096	10.1	18
133	Epithelial-Mesenchymal Transition (EMT) Gene Variants and Epithelial Ovarian Cancer (EOC) Risk. *Genetic Epidemiology*, **2015**, 39, 689-97	2.6	18
132	Family history of breast cancer and all-cause mortality after breast cancer diagnosis in the Breast Cancer Family Registry. *Breast Cancer Research and Treatment*, **2009**, 117, 167-76	4.4	18
131	RAD51B in Familial Breast Cancer. *PLoS ONE*, **2016**, 11, e0153788	3.7	18
130	Fine-scale mapping of the 4q24 locus identifies two independent loci associated with breast cancer risk. *Cancer Epidemiology Biomarkers and Prevention*, **2015**, 24, 1680-91	4	17
129	A three-protein biomarker panel assessed in diagnostic tissue predicts death from prostate cancer for men with localized disease. *Cancer Medicine*, **2014**, 3, 1266-74	4.8	17
128	9q31.2-rs865686 as a susceptibility locus for estrogen receptor-positive breast cancer: evidence from the Breast Cancer Association Consortium. *Cancer Epidemiology Biomarkers and Prevention*, **2012**, 21, 1783-91	4	17
127	Genes associated with histopathologic features of triple negative breast tumors predict molecular subtypes. *Breast Cancer Research and Treatment*, **2016**, 157, 117-31	4.4	17
126	The BRCA2 c.68-7T>A variant is not pathogenic: A model for clinical calibration of spliceogenicity. *Human Mutation*, **2018**, 39, 729-741	4.7	16
125	Fine scale mapping of the 17q22 breast cancer locus using dense SNPs, genotyped within the Collaborative Oncological Gene-Environment Study (COGs). *Scientific Reports*, **2016**, 6, 32512	4.9	16
124	The potential value of sibling controls compared with population controls for association studies of lifestyle-related risk factors: an example from the Breast Cancer Family Registry. *International Journal of Epidemiology*, **2011**, 40, 1342-54	7.8	16
123	Genetic overlap between autoimmune diseases and non-Hodgkin lymphoma subtypes. *Genetic Epidemiology*, **2019**, 43, 844-863	2.6	15

122	Common Genetic Variation In Cellular Transport Genes and Epithelial Ovarian Cancer (EOC) Risk. *PLoS ONE*, **2015**, 10, e0128106	3.7	15
121	FAN1 variants identified in multiple-case early-onset breast cancer families via exome sequencing: no evidence for association with risk for breast cancer. *Breast Cancer Research and Treatment*, **2011**, 130, 1043-9	4.4	15
120	Haplotype structure in Ashkenazi Jewish BRCA1 and BRCA2 mutation carriers. *Human Genetics*, **2011**, 130, 685-99	6.3	15
119	Association of genetic susceptibility variants for type 2 diabetes with breast cancer risk in women of European ancestry. *Cancer Causes and Control*, **2016**, 27, 679-93	2.8	15
118	Cirrus: An Automated Mammography-Based Measure of Breast Cancer Risk Based on Textural Features. *JNCI Cancer Spectrum*, **2018**, 2, pky057	4.6	15
117	Tools for translational epigenetic studies involving formalin-fixed paraffin-embedded human tissue: applying the Infinium HumanMethyation450 Beadchip assay to large population-based studies. *BMC Research Notes*, **2015**, 8, 543	2.3	14
116	The SNP rs6500843 in 16p13.3 is associated with survival specifically among chemotherapy-treated breast cancer patients. *Oncotarget*, **2015**, 6, 7390-407	3.3	14
115	Combined Associations of a Polygenic Risk Score and Classical Risk Factors With Breast Cancer Risk. *Journal of the National Cancer Institute*, **2021**, 113, 329-337	9.7	14
114	Germline Sequencing DNA Repair Genes in 5545 Men With Aggressive and Nonaggressive Prostate Cancer. *Journal of the National Cancer Institute*, **2021**, 113, 616-625	9.7	14
113	Genome-wide association study of peripheral blood DNA methylation and conventional mammographic density measures. *International Journal of Cancer*, **2019**, 145, 1768-1773	7.5	13
112	Gene-environment interactions involving functional variants: Results from the Breast Cancer Association Consortium. *International Journal of Cancer*, **2017**, 141, 1830-1840	7.5	13
111	A comprehensive evaluation of interaction between genetic variants and use of menopausal hormone therapy on mammographic density. *Breast Cancer Research*, **2015**, 17, 110	8.3	13
110	2q36.3 is associated with prognosis for oestrogen receptor-negative breast cancer patients treated with chemotherapy. *Nature Communications*, **2014**, 5, 4051	17.4	13
109	Inherited variants in the inner centromere protein (INCENP) gene of the chromosomal passenger complex contribute to the susceptibility of ER-negative breast cancer. *Carcinogenesis*, **2015**, 36, 256-71	4.6	12
108	Evaluating the ovarian cancer gonadotropin hypothesis: a candidate gene study. *Gynecologic Oncology*, **2015**, 136, 542-8	4.9	12
107	The :p.Arg658* truncating variant is associated with risk of triple-negative breast cancer. *Npj Breast Cancer*, **2019**, 5, 38	7.8	12
106	Genetic variation at CYP3A is associated with age at menarche and breast cancer risk: a case-control study. *Breast Cancer Research*, **2014**, 16, R51	8.3	12
105	Breast Cancer Polygenic Risk Score and Contralateral Breast Cancer Risk. *American Journal of Human Genetics*, **2020**, 107, 837-848	11	12

104	Genome-wide DNA methylation assessment of BRCA1-like early-onset breast cancer: Data from the Australian Breast Cancer Family Registry. *Experimental and Molecular Pathology*, **2018**, 105, 404-410	4.4	12
103	Rare Germline Variants in ATM Predispose to Prostate Cancer: A PRACTICAL Consortium Study. *European Urology Oncology*, **2021**, 4, 570-579	6.7	12
102	Integrating DNA methylation measures to improve clinical risk assessment: are we there yet? The case of BRCA1 methylation marks to improve clinical risk assessment of breast cancer. *British Journal of Cancer*, **2020**, 122, 1133-1140	8.7	11
101	Somatic mutations of the coding microsatellites within the beta-2-microglobulin gene in mismatch repair-deficient colorectal cancers and adenomas. *Familial Cancer*, **2018**, 17, 91-100	3	11
100	Genetic variation in mitotic regulatory pathway genes is associated with breast tumor grade. *Human Molecular Genetics*, **2014**, 23, 6034-46	5.6	11
99	Genome wide association study identifies a novel putative mammographic density locus at 1q12-q21. *International Journal of Cancer*, **2015**, 136, 2427-36	7.5	11
98	The AIB1 glutamine repeat polymorphism is not associated with risk of breast cancer before age 40 years in Australian women. *Breast Cancer Research*, **2005**, 7, R353-6	8.3	11
97	A protein-truncating mutation in CYP17A1 in three sisters with early-onset breast cancer. *Human Mutation*, **2005**, 26, 298-302	4.7	11
96	Methylation of Breast Cancer Predisposition Genes in Early-Onset Breast Cancer: Australian Breast Cancer Family Registry. *PLoS ONE*, **2016**, 11, e0165436	3.7	11
95	Fine-Mapping of the 1p11.2 Breast Cancer Susceptibility Locus. *PLoS ONE*, **2016**, 11, e0160316	3.7	11
94	Prediction and clinical utility of a contralateral breast cancer risk model. *Breast Cancer Research*, **2019**, 21, 144	8.3	11
93	Adult height is associated with increased risk of ovarian cancer: a Mendelian randomisation study. *British Journal of Cancer*, **2018**, 118, 1123-1129	8.7	10
92	Breast cancer risk and 6q22.33: combined results from Breast Cancer Association Consortium and Consortium of Investigators on Modifiers of BRCA1/2. *PLoS ONE*, **2012**, 7, e35706	3.7	10
91	No evidence that GATA3 rs570613 SNP modifies breast cancer risk. *Breast Cancer Research and Treatment*, **2009**, 117, 371-9	4.4	10
90	Interval breast cancer risk associations with breast density, family history and breast tissue aging. *International Journal of Cancer*, **2020**, 147, 375-382	7.5	10
89	Transcriptome-wide association study of breast cancer risk by estrogen-receptor status. *Genetic Epidemiology*, **2020**, 44, 442-468	2.6	9
88	The PALB2 p.Leu939Trp mutation is not associated with breast cancer risk. *Breast Cancer Research*, **2016**, 18, 111	8.3	9
87	Variants in genes encoding small GTPases and association with epithelial ovarian cancer susceptibility. *PLoS ONE*, **2018**, 13, e0197561	3.7	9

86	The incidence of PALB2 c.3113G>A in women with a strong family history of breast and ovarian cancer attending familial cancer centres in Australia. *Familial Cancer*, **2013**, 12, 587-95	3	9
85	Risk factors for uncommon histologic subtypes of breast cancer using centralized pathology review in the Breast Cancer Family Registry. *Breast Cancer Research and Treatment*, **2012**, 134, 1209-20	4.4	9
84	Contribution of large genomic BRCA1 alterations to early-onset breast cancer selected for family history and tumour morphology: a report from The Breast Cancer Family Registry. *Breast Cancer Research*, **2011**, 13, R14	8.3	9
83	Dependence of colorectal cancer risk on the parent-of-origin of mutations in DNA mismatch repair genes. *Human Mutation*, **2011**, 32, 207-12	4.7	9
82	Gene panel testing for hereditary breast cancer. *Medical Journal of Australia*, **2016**, 204, 188-90	4	9
81	Evaluation of associations between genetically predicted circulating protein biomarkers and breast cancer risk. *International Journal of Cancer*, **2020**, 146, 2130-2138	7.5	9
80	Two-stage Study of Familial Prostate Cancer by Whole-exome Sequencing and Custom Capture Identifies 10 Novel Genes Associated with the Risk of Prostate Cancer. *European Urology*, **2021**, 79, 353-361	10.2	9
79	Consortium analysis of gene and gene-folate interactions in purine and pyrimidine metabolism pathways with ovarian carcinoma risk. *Molecular Nutrition and Food Research*, **2014**, 58, 2023-35	5.9	8
78	Increased genomic burden of germline copy number variants is associated with early onset breast cancer: Australian breast cancer family registry. *Breast Cancer Research*, **2017**, 19, 30	8.3	8
77	Tumour morphology of early-onset breast cancers predicts breast cancer risk for first-degree relatives: the Australian Breast Cancer Family Registry. *Breast Cancer Research*, **2012**, 14, R122	8.3	8
76	Rationale for, and approach to, studying modifiers of risk in persons with a genetic predisposition to colorectal cancer. *Current Oncology Reports*, **2007**, 9, 202-7	6.3	8
75	The RAD51D E233G variant and breast cancer risk: population-based and clinic-based family studies of Australian women. *Breast Cancer Research and Treatment*, **2008**, 112, 35-9	4.4	8
74	Novel mammogram-based measures improve breast cancer risk prediction beyond an established mammographic density measure. *International Journal of Cancer*, **2021**, 148, 2193-2202	7.5	8
73	Mortality after breast cancer as a function of time since diagnosis by estrogen receptor status and age at diagnosis. *International Journal of Cancer*, **2019**, 145, 3207-3217	7.5	7
72	The Spectrum of Protein Truncating Variants in European Breast Cancer Cases. *Cancers*, **2020**, 12,	6.6	7
71	Prediction of contralateral breast cancer: external validation of risk calculators in 20 international cohorts. *Breast Cancer Research and Treatment*, **2020**, 181, 423-434	4.4	7
70	Rare germline genetic variants and risk of aggressive prostate cancer. *International Journal of Cancer*, **2020**, 147, 2142-2149	7.5	7
69	Obtaining high quality transcriptome data from formalin-fixed, paraffin-embedded diagnostic prostate tumor specimens. *Laboratory Investigation*, **2018**, 98, 537-550	5.9	7

68	A genome-wide linkage study of mammographic density, a risk factor for breast cancer. *Breast Cancer Research*, **2011**, 13, R132	8.3	7
67	Cancer Risks Associated With and Pathogenic Variants.. *Journal of Clinical Oncology*, **2022**, JCO2102112	2.2	7
66	TP53-based interaction analysis identifies cis-eQTL variants for TP53BP2, FBXO28, and FAM53A that associate with survival and treatment outcome in breast cancer. *Oncotarget*, **2017**, 8, 18381-18398	3.3	7
65	Targeted massively parallel sequencing characterises the mutation spectrum of PALB2 in breast and ovarian cancer cases from Poland and Ukraine. *Familial Cancer*, **2018**, 17, 345-349	3	6
64	Genetic variation in the immunosuppression pathway genes and breast cancer susceptibility: a pooled analysis of 42,510 cases and 40,577 controls from the Breast Cancer Association Consortium. *Human Genetics*, **2016**, 135, 137-54	6.3	6
63	DNA methylation-based biological age, genome-wide average DNA methylation, and conventional breast cancer risk factors. *Scientific Reports*, **2019**, 9, 15055	4.9	6
62	Re: Microsatellite instability and BRAF mutation testing in colorectal cancer prognostication. *Journal of the National Cancer Institute*, **2014**, 106,	9.7	6
61	Comparing the frequency of common genetic variants and haplotypes between carriers and non-carriers of BRCA1 and BRCA2 deleterious mutations in Australian women diagnosed with breast cancer before 40 years of age. *BMC Cancer*, **2010**, 10, 466	4.8	6
60	Stochastic Epigenetic Mutations Are Associated with Risk of Breast Cancer, Lung Cancer, and Mature B-cell Neoplasms. *Cancer Epidemiology Biomarkers and Prevention*, **2020**, 29, 2026-2037	4	6
59	Heterogeneity of luminal breast cancer characterised by immunohistochemical expression of basal markers. *British Journal of Cancer*, **2016**, 114, 298-304	8.7	5
58	Height, selected genetic markers and prostate cancer risk: results from the PRACTICAL consortium. *British Journal of Cancer*, **2017**, 117, 734-743	8.7	5
57	Testing for Gene-Environment Interactions Using a Prospective Family Cohort Design: Body Mass Index in Early and Later Adulthood and Risk of Breast Cancer. *American Journal of Epidemiology*, **2017**, 185, 487-500	3.8	5
56	Detecting differential allelic expression using high-resolution melting curve analysis: application to the breast cancer susceptibility gene CHEK2. *BMC Medical Genomics*, **2011**, 4, 39	3.7	5
55	A BRCA1 promoter variant (rs11655505) and breast cancer risk. *Journal of Medical Genetics*, **2010**, 47, 268-70	5.8	5
54	Genetic variants within the hTERT gene and the risk of colorectal cancer in Lynch syndrome. *Genes and Cancer*, **2015**, 6, 445-51	2.9	5
53	First international workshop of the ATM and cancer risk group (4-5 December 2019). *Familial Cancer* , **2021**, 1	3	5
52	The Variant C.349A>G Is Associated with Prostate Cancer Risk and Carriers Share a Common Ancestor. *Cancers*, **2020**, 12,	6.6	4
51	Socio-economic status and survival from breast cancer for young, Australian, urban women. *Australian and New Zealand Journal of Public Health*, **2010**, 34, 200-5	2.3	4

50	Germ-line variation at a functional p53 binding site increases susceptibility to breast cancer development. *The HUGO Journal*, **2009**, 3, 31-40		4
49	Pathology of Tumors Associated With Pathogenic Germline Variants in 9 Breast Cancer Susceptibility Genes.. *JAMA Oncology*, **2022**,	13.4	4
48	Assessment of variation in immunosuppressive pathway genes reveals TGFBR2 to be associated with risk of clear cell ovarian cancer. *Oncotarget*, **2016**, 7, 69097-69110	3.3	4
47	Blood pressure and risk of breast cancer, overall and by subtypes: a prospective cohort study. *Journal of Hypertension*, **2017**, 35, 1371-1380	1.9	4
46	Analysis of the breast cancer methylome using formalin-fixed paraffin-embedded tumour. *Breast Cancer Research and Treatment*, **2016**, 160, 173-180	4.4	4
45	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
44	PALB2 Genetic Variants: Can Functional Assays Assist Translation?. *Trends in Cancer*, **2020**, 6, 263-265	12.5	3
43	Mismatch repair gene pathogenic germline variants in a population-based cohort of breast cancer. *Familial Cancer*, **2020**, 19, 197-202	3	3
42	Is RNASEL:p.Glu265* a modifier of early-onset breast cancer risk for carriers of high-risk mutations?. *BMC Cancer*, **2018**, 18, 165	4.8	3
41	Use of a Novel Nonparametric Version of DEPTH to Identify Genomic Regions Associated with Prostate Cancer Risk. *Cancer Epidemiology Biomarkers and Prevention*, **2016**, 25, 1619-1624	4	3
40	rs495139 in the TYMS-ENOSF1 Region and Risk of Ovarian Carcinoma of Mucinous Histology. *International Journal of Molecular Sciences*, **2018**, 19,	6.3	3
39	Common variants in breast cancer risk loci predispose to distinct tumor subtypes.. *Breast Cancer Research*, **2022**, 24, 2	8.3	3
38	- a novel candidate breast cancer susceptibility locus on 6q14.1. *Oncotarget*, **2017**, 8, 102769-102782	3.3	3
37	Association of germline variation with the survival of women with pathogenic variants and breast cancer. *Npj Breast Cancer*, **2020**, 6, 44	7.8	3
36	Prospective Evaluation of the Addition of Polygenic Risk Scores to Breast Cancer Risk Models. *JNCI Cancer Spectrum*, **2021**, 5, pkab021	4.6	3
35	: Genetic Variation, Heritable Methylation and Disease Association. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	3
34	ABRAXAS (FAM175A) and Breast Cancer Susceptibility: No Evidence of Association in the Breast Cancer Family Registry. *PLoS ONE*, **2016**, 11, e0156820	3.7	3
33	Two truncating variants in FANCC and breast cancer risk. *Scientific Reports*, **2019**, 9, 12524	4.9	2

32	Germline HOXB13 mutations p.G84E and p.R217C do not confer an increased breast cancer risk. *Scientific Reports*, **2020**, 10, 9688	4.9	2
31	rs2735383, located at a microRNA binding site in the 3ℕTR of NBS1, is not associated with breast cancer risk. *Scientific Reports*, **2016**, 6, 36874	4.9	2
30	Is BRCA2 c.9079 G>A a predisposing variant for early onset breast cancer?. *Breast Cancer Research and Treatment*, **2008**, 109, 177-9	4.4	2
29	Rare genetic variants: making the connection with breast cancer susceptibility. *AIMS Genetics*, **2015**, 02, 281-292	2.1	2
28	Genome-wide association study identifies 32 novel breast cancer susceptibility loci from overall and subtype-specific analyses		2
27	Population-Based Estimates of the Age-Specific Cumulative Risk of Breast Cancer for Pathogenic Variants in : Findings from the Australian Breast Cancer Family Registry. *Cancers*, **2021**, 13,	6.6	2
26	CYP3A7*1C allele: linking premenopausal oestrone and progesterone levels with risk of hormone receptor-positive breast cancers. *British Journal of Cancer*, **2021**, 124, 842-854	8.7	2
25	Familial Aspects of Mammographic Density Measures Associated with Breast Cancer Risk.. *Cancers*, **2022**, 14,	6.6	2
24	Genetically Determined Height and Risk of Non-hodgkin Lymphoma. *Frontiers in Oncology*, **2019**, 9, 1539	5.3	1
23	The Role of New Sequencing Technology in Identifying Rare Mutations in New Susceptibility Genes for Cancer. *Current Genetic Medicine Reports*, **2013**, 1, 175-181	2.2	1
22	Common variants in breast cancer risk loci predispose to distinct tumor subtypes		1
21	Polygenic Risk Modelling for Prediction of Epithelial Ovarian Cancer Risk		1
20	Rare Germline Pathogenic Variants Identified by Multigene Panel Testing and the Risk of Aggressive Prostate Cancer. *Cancers*, **2021**, 13,	6.6	1
19	Genomic Risk Prediction for Breast Cancer in Older Women. *Cancers*, **2021**, 13,	6.6	1
18	Functional annotation of the 2q35 breast cancer risk locus implicates a structural variant in influencing activity of a long-range enhancer element. *American Journal of Human Genetics*, **2021**, 108, 1190-1203	11	1
17	DNA methylation and breast cancer risk: value of twin and family studies **2021**, 67-83		1
16	Association of germline genetic variants with breast cancer-specific survival in patient subgroups defined by clinic-pathological variables related to tumor biology and type of systemic treatment. *Breast Cancer Research*, **2021**, 23, 86	8.3	1
15	Population-based estimates of breast cancer risk for carriers of pathogenic variants identified by gene-panel testing. *Npj Breast Cancer*, **2021**, 7, 153	7.8	1

14	Improving breast cancer risk prediction with epigenetic risk factors.. *Nature Reviews Clinical Oncology*, **2022**,	19.4	1
13	Genome-wide and transcriptome-wide association studies of mammographic density phenotypes reveal novel loci.. *Breast Cancer Research*, **2022**, 24, 27	8.3	1
12	Segregation analysis of 17,425 population-based breast cancer families: evidence for genetic susceptibility and risk prediction		1
11	Rare germline copy number variants (CNVs) and breast cancer risk.. *Communications Biology*, **2022**, 5, 65	6.7	0
10	Germline variants and breast cancer survival in patients with distant metastases at primary breast cancer diagnosis. *Scientific Reports*, **2021**, 11, 19787	4.9	0
9	Mendelian randomisation study of smoking exposure in relation to breast cancer risk. *British Journal of Cancer*, **2021**, 125, 1135-1145	8.7	0
8	Population-based estimates of age-specific cumulative risk of breast cancer for pathogenic variants in ATM.. *Breast Cancer Research*, **2022**, 24, 24	8.3	0
7	Breast cancer risks associated with missense variants in breast cancer susceptibility genes.. *Genome Medicine*, **2022**, 14, 51	14.4	0
6	Interpretation of genomic variation and disease association: the great missense mutation challenge!. *Breast Cancer Research and Treatment*, **2015**, 151, 475-6	4.4	
5	Large-Scale Genomic Analyses Link Reproductive Aging to Hypothalamic Signaling, Breast Cancer Susceptibility, and BRCA1-Mediated DNA Repair. *Obstetrical and Gynecological Survey*, **2015**, 70, 758-762	2.4	
4	Rationale for, and approach to, studying modifiers of risk in persons with a genetic predisposition to colorectal cancer. *Current Colorectal Cancer Reports*, **2006**, 2, 173-178	1	
3	Genome-wide homozygosity and risk of four non-Hodgkin lymphoma subtypes. *Journal of Translational Genetics and Genomics*, **2021**, 5, 200-217	1.7	
2	Genome-wide interaction analysis of menopausal hormone therapy use and breast cancer risk among 62,370 women.. *Scientific Reports*, **2022**, 12, 6199	4.9	
1	Genetic Aspects of Mammographic Density Measures Associated with Breast Cancer Risk. *Cancers*, **2022**, 14, 2767	6.6	