Melissa C. Southey

List of Publications by Citations

562 papers	34,229 citations	88 h-index	168 g-index
603 ext. papers	41,318 ext. citations	8.7 avg, IF	5.77 L-index

#	Paper	IF	Citations
562	Genome-wide association study identifies novel breast cancer susceptibility loci. *Nature*, **2007**, 447, 1087-93	50.4	1957
561	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
560	Large-scale genotyping identifies 41 new loci associated with breast cancer risk. *Nature Genetics*, **2013**, 45, 353-61, 361e1-2	36.3	813
559	Multiple newly identified loci associated with prostate cancer susceptibility. *Nature Genetics*, **2008**, 40, 316-21	36.3	722
558	Association analysis identifies 65 new breast cancer risk loci. *Nature*, **2017**, 551, 92-94	50.4	643
557	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
556	Gene-panel sequencing and the prediction of breast-cancer risk. *New England Journal of Medicine*, **2015**, 372, 2243-57	59.2	587
555	Breast-cancer risk in families with mutations in PALB2. *New England Journal of Medicine*, **2014**, 371, 497-506	59.2	576
554	A common coding variant in CASP8 is associated with breast cancer risk. *Nature Genetics*, **2007**, 39, 352-8	36.3	557
553	Iron-overload-related disease in HFE hereditary hemochromatosis. *New England Journal of Medicine* , **2008**, 358, 221-30	59.2	516
552	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
551	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
550	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
549	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
548	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
547	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
546	Newly discovered breast cancer susceptibility loci on 3p24 and 17q23.2. *Nature Genetics*, **2009**, 41, 585-90	36.3	393

545	Polygenic Risk Scores for Prediction of Breast Cancer and Breast Cancer Subtypes. *American Journal of Human Genetics*, **2019**, 104, 21-34	11	363
544	Identification of seven new prostate cancer susceptibility loci through a genome-wide association study. *Nature Genetics*, **2009**, 41, 1116-21	36.3	360
543	Association analyses of more than 140,000 men identify 63 new prostate cancer susceptibility loci. *Nature Genetics*, **2018**, 50, 928-936	36.3	340
542	A meta-analysis of 87,040 individuals identifies 23 new susceptibility loci for prostate cancer. *Nature Genetics*, **2014**, 46, 1103-9	36.3	331
541	Genome-wide association studies identify four ER negative-specific breast cancer risk loci. *Nature Genetics*, **2013**, 45, 392-8, 398e1-2	36.3	327
540	Prediction of breast cancer risk based on profiling with common genetic variants. *Journal of the National Cancer Institute*, **2015**, 107,	9.7	324
539	Heterogeneity of breast cancer associations with five susceptibility loci by clinical and pathological characteristics. *PLoS Genetics*, **2008**, 4, e1000054	6	280
538	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
537	GWAS meta-analysis and replication identifies three new susceptibility loci for ovarian cancer. *Nature Genetics*, **2013**, 45, 362-70, 370e1-2	36.3	267
536	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
535	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
534	Multiple loci on 8q24 associated with prostate cancer susceptibility. *Nature Genetics*, **2009**, 41, 1058-60	36.3	252
533	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
532	Seven prostate cancer susceptibility loci identified by a multi-stage genome-wide association study. *Nature Genetics*, **2011**, 43, 785-91	36.3	243
531	A genome-wide association study identifies a new ovarian cancer susceptibility locus on 9p22.2. *Nature Genetics*, **2009**, 41, 996-1000	36.3	240
530	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
529	Genome-wide association analysis identifies three new breast cancer susceptibility loci. *Nature Genetics*, **2012**, 44, 312-8	36.3	237
528	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

527	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
526	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
525	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
524	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. *Nature Genetics*, **2017**, 49, 680-691	36.3	190
523	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
522	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. *Nature Genetics*, **2017**, 49, 1767-1778	36.3	186
521	Identification of six new susceptibility loci for invasive epithelial ovarian cancer. *Nature Genetics*, **2015**, 47, 164-71	36.3	177
520	Discovery of common and rare genetic risk variants for colorectal cancer. *Nature Genetics*, **2019**, 51, 76-87	36.3	177
519	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
518	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
517	Rare variants in the ATM gene and risk of breast cancer. *Breast Cancer Research*, **2011**, 13, R73	8.3	151
516	Hypomethylation of smoking-related genes is associated with future lung cancer in four prospective cohorts. *Nature Communications*, **2015**, 6, 10192	17.4	144
515	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
514	Breast Cancer Risk Genes - Association Analysis in More than 113,000 Women. *New England Journal of Medicine*, **2021**, 384, 428-439	59.2	143
513	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
512	Rare, evolutionarily unlikely missense substitutions in ATM confer increased risk of breast cancer. *American Journal of Human Genetics*, **2009**, 85, 427-46	11	140
511	Mutational spectrum in a worldwide study of 29,700 families with BRCA1 or BRCA2 mutations. *Human Mutation*, **2018**, 39, 593-620	4.7	138
510	Genetic and histopathologic evaluation of BRCA1 and BRCA2 DNA sequence variants of unknown clinical significance. *Cancer Research*, **2006**, 66, 2019-27	10.1	138

509	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
508	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
507	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
506	Multiple novel prostate cancer predisposition loci confirmed by an international study: the PRACTICAL Consortium. *Cancer Epidemiology Biomarkers and Prevention*, **2008**, 17, 2052-61	4	134
505	Cancer Risks Associated With Germline Pathogenic Variants: An International Study of 524 Families. *Journal of Clinical Oncology*, **2020**, 38, 674-685	2.2	133
504	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
503	Epigenetic analysis leads to identification of HNF1B as a subtype-specific susceptibility gene for ovarian cancer. *Nature Communications*, **2013**, 4, 1628	17.4	124
502	Genome-wide association study identifies 25 known breast cancer susceptibility loci as risk factors for triple-negative breast cancer. *Carcinogenesis*, **2014**, 35, 1012-9	4.6	121
501	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
500	PALB2, CHEK2 and ATM rare variants and cancer risk: data from COGS. *Journal of Medical Genetics*, **2016**, 53, 800-811	5.8	121
499	Tamoxifen and risk of contralateral breast cancer for BRCA1 and BRCA2 mutation carriers. *Journal of Clinical Oncology*, **2013**, 31, 3091-9	2.2	118
498	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
497	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
496	Evidence of gene-environment interactions between common breast cancer susceptibility loci and established environmental risk factors. *PLoS Genetics*, **2013**, 9, e1003284	6	112
495	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
494	Genome-wide association study identifies multiple susceptibility loci for diffuse large B cell lymphoma. *Nature Genetics*, **2014**, 46, 1233-8	36.3	108
493	Age- and Tumor Subtype-Specific Breast Cancer Risk Estimates for CHEK2*1100delC Carriers. *Journal of Clinical Oncology*, **2016**, 34, 2750-60	2.2	107
492	Colorectal carcinomas with KRAS mutation are associated with distinctive morphological and molecular features. *Modern Pathology*, **2013**, 26, 825-34	9.8	106

491	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
490	PIK3CA activating mutation in colorectal carcinoma: associations with molecular features and survival. *PLoS ONE*, **2013**, 8, e65479	3.7	102
489	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
488	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
487	A genome wide linkage search for breast cancer susceptibility genes. *Genes Chromosomes and Cancer*, **2006**, 45, 646-55	5	100
486	BRCA1 and BRCA2 mutation carriers, oral contraceptive use, and breast cancer before age 50. *Cancer Epidemiology Biomarkers and Prevention*, **2006**, 15, 1863-70	4	98
485	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
484	Common variants in ZNF365 are associated with both mammographic density and breast cancer risk. *Nature Genetics*, **2011**, 43, 185-7	36.3	96
483	HFE C282Y homozygotes are at increased risk of breast and colorectal cancer. *Hepatology*, **2010**, 51, 1311-8	11.2	95
482	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
481	19p13.1 is a triple-negative-specific breast cancer susceptibility locus. *Cancer Research*, **2012**, 72, 1795-803	10.1	93
480	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
479	Identification of a BRCA2-specific modifier locus at 6p24 related to breast cancer risk. *PLoS Genetics*, **2013**, 9, e1003173	6	90
478	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
477	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
476	A PALB2 mutation associated with high risk of breast cancer. *Breast Cancer Research*, **2010**, 12, R109	8.3	89
475	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
474	Evidence that breast cancer risk at the 2q35 locus is mediated through IGFBP5 regulation. *Nature Communications*, **2014**, 4, 4999	17.4	87

473	Identification and molecular characterization of a new ovarian cancer susceptibility locus at 17q21.31. *Nature Communications*, **2013**, 4, 1627	17.4	85
472	Genome-wide association study identifies multiple loci associated with both mammographic density and breast cancer risk. *Nature Communications*, **2014**, 5, 5303	17.4	84
471	Penetrance analysis of the PALB2 c.1592delT founder mutation. *Clinical Cancer Research*, **2008**, 14, 4667-71	11.9	84
470	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
469	DNA methylation changes measured in pre-diagnostic peripheral blood samples are associated with smoking and lung cancer risk. *International Journal of Cancer*, **2017**, 140, 50-61	7.5	83
468	DNA methylation-based biological aging and cancer risk and survival: Pooled analysis of seven prospective studies. *International Journal of Cancer*, **2018**, 142, 1611-1619	7.5	83
467	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
466	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
465	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
464	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
463	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
462	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
461	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
460	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
459	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
458	Association of ESR1 gene tagging SNPs with breast cancer risk. *Human Molecular Genetics*, **2009**, 18, 1131-9	5.6	75
457	The role of genetic breast cancer susceptibility variants as prognostic factors. *Human Molecular Genetics*, **2012**, 21, 3926-39	5.6	75
456	Height and Breast Cancer Risk: Evidence From Prospective Studies and Mendelian Randomization. *Journal of the National Cancer Institute*, **2015**, 107,	9.7	74

455	Common genetic variants and modification of penetrance of BRCA2-associated breast cancer. *PLoS Genetics*, **2010**, 6, e1001183	6	74
454	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
453	Interplay between BRCA1 and RHAMM regulates epithelial apicobasal polarization and may influence risk of breast cancer. *PLoS Biology*, **2011**, 9, e1001199	9.7	73
452	10-year performance of four models of breast cancer risk: a validation study. *Lancet Oncology, The*, **2019**, 20, 504-517	21.7	73
451	The E211 G>A androgen receptor polymorphism is associated with a decreased risk of metastatic prostate cancer and androgenetic alopecia. *Cancer Epidemiology Biomarkers and Prevention*, **2005**, 14, 993-6	4	71
450	Meta-analysis of genome-wide association studies discovers multiple loci for chronic lymphocytic leukemia. *Nature Communications*, **2016**, 7, 10933	17.4	70
449	Identification of nine new susceptibility loci for endometrial cancer. *Nature Communications*, **2018**, 9, 3166	17.4	70
448	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
447	Double-strand break repair gene polymorphisms and risk of breast or ovarian cancer. *Cancer Epidemiology Biomarkers and Prevention*, **2005**, 14, 319-23	4	70
446	Comparison of DNA- and RNA-based methods for detection of truncating BRCA1 mutations. *Human Mutation*, **2002**, 20, 65-73	4.7	69
445	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
444	Risk of extracolonic cancers for people with biallelic and monoallelic mutations in MUTYH. *International Journal of Cancer*, **2016**, 139, 1557-63	7.5	67
443	Novel Common Genetic Susceptibility Loci for Colorectal Cancer. *Journal of the National Cancer Institute*, **2019**, 111, 146-157	9.7	67
442	Socioeconomic position, lifestyle habits and biomarkers of epigenetic aging: a multi-cohort analysis. *Aging*, **2019**, 11, 2045-2070	5.6	67
441	Cancer risks for monoallelic MUTYH mutation carriers with a family history of colorectal cancer. *International Journal of Cancer*, **2011**, 129, 2256-62	7.5	66
440	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
439	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
438	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

437	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
436	The natural history of serum iron indices for HFE C282Y homozygosity associated with hereditary hemochromatosis. *Gastroenterology*, **2008**, 135, 1945-52	13.3	65
435	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. *Nature Communications*, **2016**, 7, 11375	17.4	64
434	HFE C282Y/H63D compound heterozygotes are at low risk of hemochromatosis-related morbidity. *Hepatology*, **2009**, 50, 94-101	11.2	64
433	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
432	Common genetic variants associated with breast cancer and mammographic density measures that predict disease. *Cancer Research*, **2010**, 70, 1449-58	10.1	63
431	The common variant rs1447295 on chromosome 8q24 and prostate cancer risk: results from an Australian population-based case-control study. *Cancer Epidemiology Biomarkers and Prevention*, **2007**, 16, 610-2	4	63
430	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
429	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
428	Association of DNA Methylation-Based Biological Age With Health Risk Factors and Overall and Cause-Specific Mortality. *American Journal of Epidemiology*, **2018**, 187, 529-538	3.8	61
427	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
426	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
425	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
424	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
423	Fine-mapping of prostate cancer susceptibility loci in a large meta-analysis identifies candidate causal variants. *Nature Communications*, **2018**, 9, 2256	17.4	57
422	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
421	Mutation analysis of BRCA1 and BRCA2 cancer predisposition genes in radiation hypersensitive cancer patients. *International Journal of Radiation Oncology Biology Physics*, **2000**, 48, 959-65	4	57
420	Novel DNA sequence variants in the hHR21 DNA repair gene in radiosensitive cancer patients. *International Journal of Radiation Oncology Biology Physics*, **2001**, 50, 1323-31	4	56

419	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. *Nature Genetics*, **2020**, 52, 56-73	36.3	56
418	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
417	Uncommon CHEK2 mis-sense variant and reduced risk of tobacco-related cancers: case control study. *Human Molecular Genetics*, **2007**, 16, 1794-801	5.6	55
416	Genome-wide association analysis implicates dysregulation of immunity genes in chronic lymphocytic leukaemia. *Nature Communications*, **2017**, 8, 14175	17.4	54
415	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
414	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
413	Associations of alcohol intake, smoking, physical activity and obesity with survival following colorectal cancer diagnosis by stage, anatomic site and tumor molecular subtype. *International Journal of Cancer*, **2018**, 142, 238-250	7.5	53
412	Functional mechanisms underlying pleiotropic risk alleles at the 19p13.1 breast-ovarian cancer susceptibility locus. *Nature Communications*, **2016**, 7, 12675	17.4	53
411	Comparison of mRNA splicing assay protocols across multiple laboratories: recommendations for best practice in standardized clinical testing. *Clinical Chemistry*, **2014**, 60, 341-52	5.5	53
410	Five polymorphisms and breast cancer risk: results from the Breast Cancer Association Consortium. *Cancer Epidemiology Biomarkers and Prevention*, **2009**, 18, 1610-6	4	53
409	Primary cutaneous Ewing's sarcoma/peripheral primitive neuroectodermal tumors in childhood. A molecular, cytogenetic, and immunohistochemical study. *Diagnostic Molecular Pathology*, **1995**, 4, 174-81		53
408	Heritable DNA methylation marks associated with susceptibility to breast cancer. *Nature Communications*, **2018**, 9, 867	17.4	52
407	Targeted massively parallel sequencing of a panel of putative breast cancer susceptibility genes in a large cohort of multiple-case breast and ovarian cancer families. *Journal of Medical Genetics*, **2016**, 53, 34-42	5.8	52
406	Epigenetic supersimilarity of monozygotic twin pairs. *Genome Biology*, **2018**, 19, 2	18.3	52
405	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
404	Genetic modifiers of CHEK2*1100delC-associated breast cancer risk. *Genetics in Medicine*, **2017**, 19, 599-603	8.3	51
403	Causal effect of smoking on DNA methylation in peripheral blood: a twin and family study. *Clinical Epigenetics*, **2018**, 10, 18	7.7	50
402	Mammographic breast density and breast cancer: evidence of a shared genetic basis. *Cancer Research*, **2012**, 72, 1478-84	10.1	50

401	Hypomorphic Missense Variants Confer Moderate Risks of Breast Cancer. *Cancer Research*, **2017**, 77, 2789-2799	10.1	49
400	The use of DNA from archival dried blood spots with the Infinium HumanMethylation450 array. *BMC Biotechnology*, **2013**, 13, 23	3.5	49
399	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
398	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
397	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
396	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
395	Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. *Nature Communications*, **2019**, 10, 1741	17.4	47
394	5alpha-Reductase type 2 gene variant associations with prostate cancer risk, circulating hormone levels and androgenetic alopecia. *International Journal of Cancer*, **2007**, 120, 776-80	7.5	47
393	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
392	A Cost-effectiveness Analysis of Multigene Testing for All Patients With Breast Cancer. *JAMA Oncology*, **2019**,	13.4	46
391	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
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