Melissa C Southey

List of Publications by Year
in descending order

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339 papers	31,787 citations	4120 87 h-index	5663 162 g-index
351 all docs	351 docs citations	351 times ranked	28782 citing authors

#	Article	IF	Citations
1	Association analysis identifies 65 new breast cancer risk loci. Nature, 2017, 551, 92-94.	13.7	1,099
2	Large-scale genotyping identifies 41 new loci associated with breast cancer risk. Nature Genetics, 2013, 45, 353-361.	9.4	960
3	Multiple newly identified loci associated with prostate cancer susceptibility. Nature Genetics, 2008, 40, 316-321.	9.4	796
4	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.	3.9	764
5	Breast-Cancer Risk in Families with Mutations in PALB2. New England Journal of Medicine, 2014, 371, 497-506.	13.9	745
6	Polygenic Risk Scores for Prediction of Breast Cancer and Breast Cancer Subtypes. American Journal of Human Genetics, 2019, 104, 21-34.	2.6	711
7	Association analyses of more than 140,000 men identify 63 new prostate cancer susceptibility loci. Nature Genetics, 2018, 50, 928-936.	9.4	652
8	Iron-Overloadâ€“Related Disease inHFEHereditary Hemochromatosis. New England Journal of Medicine, 2008, 358, 221-230.	13.9	649
9	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, 250-263.	3.0	596
10	A common coding variant in CASP8 is associated with breast cancer risk. Nature Genetics, 2007, 39, 352-358.	9.4	591
11	Breast Cancer Risk Genes â€" Association Analysis in More than 113,000 Women. New England Journal of Medicine, 2021, 384, 428-439.	13.9	532
12	A genome-wide association study identifies colorectal cancer susceptibility loci on chromosomes 10p14 and 8q23.3. Nature Genetics, 2008, 40, 623-630.	9.4	514
13	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, 134-147.	1.1	513
14	Genome-wide association analysis of more than 120,000 individuals identifies 15 new susceptibility loci for breast cancer. Nature Genetics, 2015, 47, 373-380.	9.4	513
15	Multiple independent variants at the TERT locus are associated with telomere length and risks of breast and ovarian cancer. Nature Genetics, 2013, 45, 371-384.	9.4	493
16	Identification of 23 new prostate cancer susceptibility loci using the iCOGS custom genotyping array. Nature Genetics, 2013, 45, 385-391.	9.4	492
17	Newly discovered breast cancer susceptibility loci on 3p24 and 17q23.2. Nature Genetics, 2009, 41, 585-590.	9.4	434
18	Prediction of Breast Cancer Risk Based on Profiling With Common Genetic Variants. Journal of the National Cancer Institute, 2015, 107, .	3.0	428

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19	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.	9.4	426
20	Identification of seven new prostate cancer susceptibility loci through a genome-wide association study. Nature Genetics, 2009, 41, 1116-1121.	9.4	389
21	Discovery of common and rare genetic risk variants for colorectal cancer. Nature Genetics, 2019, 51, 76-87.	9.4	377
22	Genome-wide association studies identify four ER negativeâ€“specific breast cancer risk loci. Nature Genetics, 2013, 45, 392-398.	9.4	374
23	Large-scale genomic analyses link reproductive aging to hypothalamic signaling, breast cancer susceptibility and BRCA1-mediated DNA repair. Nature Genetics, 2015, 47, 1294-1303.	9.4	357
24	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. Nature Genetics, 2017, 49, 680-691.	9.4	356
25	GWAS meta-analysis and replication identifies three new susceptibility loci for ovarian cancer. Nature Genetics, 2013, 45, 362-370.	9.4	326
26	Heterogeneity of Breast Cancer Associations with Five Susceptibility Loci by Clinical and Pathological Characteristics. PLoS Genetics, 2008, 4, e1000054.	1.5	315
27	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. Nature Genetics, 2017, 49, 1767-1778.	9.4	289
28	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, 958-964.	0.8	286
29	Breast Cancer Risk From Modifiable and Nonmodifiable Risk Factors Among White Women in the United States. JAMA Oncology, 2016, 2, 1295.	3.4	285
30	A common variant at the TERT-CLPTM1L locus is associated with estrogen receptorâ€“negative breast cancer. Nature Genetics, 2011, 43, 1210-1214.	9.4	279
31	Multiple loci on 8q24 associated with prostate cancer susceptibility. Nature Genetics, 2009, 41, 1058-1060.	9.4	273
32	Seven prostate cancer susceptibility loci identified by a multi-stage genome-wide association study. Nature Genetics, 2011, 43, 785-791.	9.4	265
33	Genome-wide association study identifies 32 novel breast cancer susceptibility loci from overall and subtype-specific analyses. Nature Genetics, 2020, 52, 572-581.	9.4	265
34	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.	9.4	264
35	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.	2.2	255
36	Genome-Wide Association Study in BRCA1 Mutation Carriers Identifies Novel Loci Associated with Breast and Ovarian Cancer Risk. PLoS Genetics, 2013, 9, e1003212.	1.5	244

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37	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.	2.0	243
38	Metachronous colorectal cancer risk for mismatch repair gene mutation carriers: the advantage of more extensive colon surgery. Gut, 2011, 60, 950-957.	6.1	227
39	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.	2.6	201
40	Use of Molecular Tumor Characteristics to Prioritize Mismatch Repair Gene Testing in Early-Onset Colorectal Cancer. Journal of Clinical Oncology, 2005, 23, 6524-6532.	0.8	194
41	Rare variants in the ATMgene and risk of breast cancer. Breast Cancer Research, 2011, 13, R73.	2.2	188
42	A transcriptome-wide association study of 229,000 women identifies new candidate susceptibility genes for breast cancer. Nature Genetics, 2018, 50, 968-978.	9.4	184
43	Genetic insights into biological mechanisms governing human ovarian ageing. Nature, 2021, 596, 393-397.	13.7	183
44	Identification of nine new susceptibility loci for endometrial cancer. Nature Communications, 2018, 9, 3166.	5.8	178
45	PALB2,CHEK2andATMrare variants and cancer risk: data from COGS. Journal of Medical Genetics, 2016, 53, 800-811.	1.5	174
46	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, 5373-5384.	1.4	168
47	Rare, Evolutionarily Unlikely Missense Substitutions in ATM Confer Increased Risk of Breast Cancer. American Journal of Human Genetics, 2009, 85, 427-446.	2.6	165
48	CHEK2*1100delC Heterozygosity in Women With Breast Cancer Associated With Early Death, Breast Cancerâ€“Specific Death, and Increased Risk of a Second Breast Cancer. Journal of Clinical Oncology, 2012, 30, 4308-4316.	0.8	162
49	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, 1052-1067.	7.7	157
50	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, 121-128.	2.1	156
51	Adaptive evolution of the tumour suppressor BRCA1 in humans and chimpanzees. Nature Genetics, 2000, 25, 410-413.	9.4	153
52	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, 3289-3303.	1.4	152
53	Age- and Tumor Subtypeâ€“Specific Breast Cancer Risk Estimates for CHEK2*1100delC Carriers. Journal of Clinical Oncology, 2016, 34, 2750-2760.	0.8	152
54	Cancer Risks For Mismatch Repair Gene Mutation Carriers: A Population-Based Early Onset Case-Family Study. Clinical Gastroenterology and Hepatology, 2006, 4, 489-498.	2.4	151

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55	Familial Risks, Early-Onset Breast Cancer, and BRCA1 and BRCA2 Germline Mutations. Journal of the National Cancer Institute, 2003, 95, 448-457.	3.0	150
56	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.	0.7	147
57	Genome-wide association study identifies multiple susceptibility loci for diffuse large B cell lymphoma. Nature Genetics, 2014, 46, 1233-1238.	9.4	147
58	Epigenetic analysis leads to identification of HNF1B as a subtype-specific susceptibility gene for ovarian cancer. Nature Communications, 2013, 4, 1628.	5.8	144
59	Evidence of Geneâ€“Environment Interactions between Common Breast Cancer Susceptibility Loci and Established Environmental Risk Factors. PLoS Genetics, 2013, 9, e1003284.	1.5	136
60	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.	0.8	134
61	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, 350-356.	1.1	133
62	Colorectal carcinomas with KRAS mutation are associated with distinctive morphological and molecular features. Modern Pathology, 2013, 26, 825-834.	2.9	126
63	Breast cancer risk variants at 6q25 display different phenotype associations and regulate ESR1, RMND1 and CCDC170. Nature Genetics, 2016, 48, 374-386.	9.4	125
64	HFE C282Y homozygotes are at increased risk of breast and colorectal cancer. Hepatology, 2010, 51, 1311-1318.	3.6	123
65	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. Nature Genetics, 2020, 52, 56-73.	9.4	120
66	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, 408-415.	1.4	118
67	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.	3.9	118
68	PIK3CA Activating Mutation in Colorectal Carcinoma: Associations with Molecular Features and Survival. PLoS ONE, 2013, 8, e65479.	1.1	117
69	Cumulative Burden of Colorectal Cancerâ€“Associated Genetic Variants Is More Strongly Associated With Early-Onset vs Late-Onset Cancer. Gastroenterology, 2020, 158, 1274-1286.e12.	0.6	110
70	Common variants in ZNF365 are associated with both mammographic density and breast cancer risk. Nature Genetics, 2011, 43, 185-187.	9.4	109
71	Genome-wide association study identifies multiple loci associated with both mammographic density and breast cancer risk. Nature Communications, 2014, 5, 5303.	5.8	109
72	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.	1.4	106

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73	Identification of a BRCA2-Specific Modifier Locus at 6p24 Related to Breast Cancer Risk. PLoS Genetics, 2013, 9, e1003173.	1.5	105
74	Evidence that breast cancer risk at the 2q35 locus is mediated through IGFBP5 regulation. Nature Communications, 2014, 5, 4999.	5.8	105
75	A PALB2 mutation associated with high risk of breast cancer. Breast Cancer Research, 2010, 12, R109.	2.2	102
76	HFEC282Y/H63D compound heterozygotes are at low risk of hemochromatosis-related morbidity. Hepatology, 2009, 50, 94-101.	3.6	101
77	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, 1156-1166.	1.1	101
78	19p13.1 Is a Triple-Negativeâ€“Specific Breast Cancer Susceptibility Locus. Cancer Research, 2012, 72, 1795-1803.	0.4	100
79	Cross-Cancer Genome-Wide Analysis of Lung, Ovary, Breast, Prostate, and Colorectal Cancer Reveals Novel Pleiotropic Associations. Cancer Research, 2016, 76, 5103-5114.	0.4	100
80	Risk of Estrogen Receptorâ€“Positive and â€“Negative Breast Cancer and Singleâ€“Nucleotide Polymorphism 2q35-rs13387042. Journal of the National Cancer Institute, 2009, 101, 1012-1018.	3.0	99
81	Rare key functional domain missense substitutions in MRE11A, RAD50, and NBNcontribute to breast cancer susceptibility: results from a Breast Cancer Family Registry case-control mutation-screening study. Breast Cancer Research, 2014, 16, R58.	2.2	99
82	Height and Breast Cancer Risk: Evidence From Prospective Studies and Mendelian Randomization. Journal of the National Cancer Institute, 2015, 107, djv219.	3.0	99
83	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, 1046-1060.	2.6	98
84	Identification and molecular characterization of a new ovarian cancer susceptibility locus at 17q21.31. Nature Communications, 2013, 4, 1627.	5.8	98
85	After BRCA1 and BRCA2â€”What Next? Multifactorial Segregation Analyses of Three-Generation, Population-Based Australian Families Affected by Female Breast Cancer. American Journal of Human Genetics, 2001, 68, 420-431.	2.6	97
86	Ethnicity and Risk for Colorectal Cancers Showing Somatic BRAF V600E Mutation or CpG Island Methylator Phenotype. Cancer Epidemiology Biomarkers and Prevention, 2008, 17, 1774-1780.	1.1	96
87	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, 359-365.	1.1	96
88	Causal effect of smoking on DNA methylation in peripheral blood: a twin and family study. Clinical Epigenetics, 2018, 10, 18.	1.8	95
89	No evidence that protein truncating variants inBRIP1are associated with breast cancer risk: implications for gene panel testing. Journal of Medical Genetics, 2016, 53, 298-309.	1.5	94
90	Epigenome-wide methylation in DNA from peripheral blood as a marker of risk for breast cancer. Breast Cancer Research and Treatment, 2014, 148, 665-673.	1.1	93

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91	Penetrance Analysis of the PALB2 c.1592delT Founder Mutation. Clinical Cancer Research, 2008, 14, 4667-4671.	3.2	90
92	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, 6616-6633.	1.4	90
93	Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. Nature Communications, 2019, 10, 1741.	5.8	90
94	Cancer Risks Associated With BRCA1 and BRCA2 Pathogenic Variants. Journal of Clinical Oncology, 2022, 40, 1529-1541.	0.8	90
95	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.	0.9	88
96	Fine-mapping of prostate cancer susceptibility loci in a large meta-analysis identifies candidate causal variants. Nature Communications, 2018, 9, 2256.	5.8	88
97	Shared heritability and functional enrichment across six solid cancers. Nature Communications, 2019, 10, 431.	5.8	88
98	The Natural History of Serum Iron Indices for HFE C282Y Homozygosity Associated With Hereditary Hemochromatosis. Gastroenterology, 2008, 135, 1945-1952.	0.6	86
99	Common Genetic Variants and Modification of Penetrance of BRCA2-Associated Breast Cancer. PLoS Genetics, 2010, 6, e1001183.	1.5	85
100	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, 4795-4800.	0.4	84
101	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.	2.2	82
102	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.	1.1	82
103	Associations of obesity and circulating insulin and glucose with breast cancer risk: a Mendelian randomization analysis. International Journal of Epidemiology, 2019, 48, 795-806.	0.9	81
104	The role of genetic breast cancer susceptibility variants as prognostic factors. Human Molecular Genetics, 2012, 21, 3926-3939.	1.4	80
105	Functional mechanisms underlying pleiotropic risk alleles at the 19p13.1 breastâ€“ovarian cancer susceptibility locus. Nature Communications, 2016, 7, 12675.	5.8	78
106	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, 658-669.	1.1	77
107	BRCA2 Polymorphic Stop Codon K3326X and the Risk of Breast, Prostate, and Ovarian Cancers. Journal of the National Cancer Institute, 2016, 108, djv315.	3.0	77
108	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.	2.6	76

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109	Heritable DNA methylation marks associated with susceptibility to breast cancer. Nature Communications, 2018, 9, 867.	5.8	76
110	BRCA2 Hypomorphic Missense Variants Confer Moderate Risks of Breast Cancer. Cancer Research, 2017, 77, 2789-2799.	0.4	75
111	Comparison of DNA- and RNA-Based Methods for Detection of TruncatingBRCA1 Mutations. Human Mutation, 2002, 20, 65-73.	1.1	74
112	Common Genetic Variants Associated with Breast Cancer and Mammographic Density Measures That Predict Disease. Cancer Research, 2010, 70, 1449-1458.	0.4	74
113	Rare, evolutionarily unlikely missense substitutions in CHEK2contribute to breast cancer susceptibility: results from a breast cancer family registry case-control mutation-screening study. Breast Cancer Research, 2011, 13, R6.	2.2	74
114	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.	2.2	71
115	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 Consortiumâ€. Human Molecular Genetics, 2011, 20, 4693-4706.	1.4	71
116	Adult body mass index and risk of ovarian cancer by subtype: a Mendelian randomization study. International Journal of Epidemiology, 2016, 45, 884-895.	0.9	71
117	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.	2.2	70
118	Multiple novel prostate cancer susceptibility signals identified by fine-mapping of known risk loci among Europeans. Human Molecular Genetics, 2015, 24, 5589-5602.	1.4	67
119	Genetic modifiers of CHEK2*1100delC-associated breast cancer risk. Genetics in Medicine, 2017, 19, 599-603.	1.1	67
120	Body size and risk for colorectal cancers showing BRAF mutations or microsatellite instability: a pooled analysis. International Journal of Epidemiology, 2012, 41, 1060-1072.	0.9	65
121	Cis-eQTL analysis and functional validation of candidate susceptibility genes for high-grade serous ovarian cancer. Nature Communications, 2015, 6, 8234.	5.8	63
122	Refinement of the basis and impact of common 11q23.1 variation to the risk of developing colorectal cancer. Human Molecular Genetics, 2008, 17, 3720-3727.	1.4	61
123	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, 140-149.	1.2	61
124	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.	2.6	59
125	Is uptake of genetic testing for colorectal cancer influenced by knowledge of insurance implications?. Medical Journal of Australia, 2009, 191, 255-258.	0.8	58
126	Five Polymorphisms and Breast Cancer Risk: Results from the Breast Cancer Association Consortium. Cancer Epidemiology Biomarkers and Prevention, 2009, 18, 1610-1616.	1.1	57

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127	Identification of Novel Genetic Markers of Breast Cancer Survival. Journal of the National Cancer Institute, 2015, 107, .	3.0	56
128	Risk Analysis of Prostate Cancer in PRACTICAL, a Multinational Consortium, Using 25 Known Prostate Cancer Susceptibility Loci. Cancer Epidemiology Biomarkers and Prevention, 2015, 24, 1121-1129.	1.1	56
129	Novel Associations between Common Breast Cancer Susceptibility Variants and Risk-Predicting Mammographic Density Measures. Cancer Research, 2015, 75, 2457-2467.	0.4	55
130	Mammographic Breast Density and Breast Cancer: Evidence of a Shared Genetic Basis. Cancer Research, 2012, 72, 1478-1484.	0.4	54
131	Prediction of individual genetic risk to prostate cancer using a polygenic score. Prostate, 2015, 75, 1467-1474.	1.2	54
132	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.	0.4	54
133	ELAC2/HPC2 Polymorphisms, Prostate-Specific Antigen Levels, and Prostate Cancer. Journal of the National Cancer Institute, 2003, 95, 818-824.	3.0	53
134	Common non-synonymous SNPs associated with breast cancer susceptibility: findings from the Breast Cancer Association Consortium. Human Molecular Genetics, 2014, 23, 6096-6111.	1.4	53
135	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.	1.1	53
136	BRCA1 and BRCA2 mutation carriers in the Breast Cancer Family Registry: an open resource for collaborative research. Breast Cancer Research and Treatment, 2009, 116, 379-386.	1.1	52
137	Genetically predicted longer telomere length is associated with increased risk of B-cell lymphoma subtypes. Human Molecular Genetics, 2016, 25, 1663-1676.	1.4	52
138	Genome-wide association study of germline variants and breast cancer-specific mortality. British Journal of Cancer, 2019, 120, 647-657.	2.9	52
139	A high-plex PCR approach for massively parallel sequencing. BioTechniques, 2013, 55, 69-74.	0.8	51
140	Fineâ€scale mapping of 8q24 locus identifies multiple independent risk variants for breast cancer. International Journal of Cancer, 2016, 139, 1303-1317.	2.3	51
141	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.	1.1	51
142	Pathology of Tumors Associated With Pathogenic Germline Variants in 9 Breast Cancer Susceptibility Genes. JAMA Oncology, 2022, 8, e216744.	3.4	51
143	Fine-mapping of the HNF1B multicancer locus identifies candidate variants that mediate endometrial cancer risk. Human Molecular Genetics, 2015, 24, 1478-1492.	1.4	50
144	Atlas of prostate cancer heritability in European and African-American men pinpoints tissue-specific regulation. Nature Communications, 2016, 7, 10979.	5.8	50

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145	Identification of fifteen novel germline variants in theBRCA13â€²UTR reveals a variant in a breast cancer case that introduces a functionalmiR-103target site. Human Mutation, 2012, 33, 1665-1675.	1.1	49
146	MicroRNA Related Polymorphisms and Breast Cancer Risk. PLoS ONE, 2014, 9, e109973.	1.1	49
147	Association Between a Germline OCA2 Polymorphism at Chromosome 15q13.1 and Estrogen Receptorâ€“Negative Breast Cancer Survival. Journal of the National Cancer Institute, 2010, 102, 650-662.	3.0	48
148	A role for XRCC2 gene polymorphisms in breast cancer risk and survival. Journal of Medical Genetics, 2011, 48, 477-484.	1.5	47
149	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, 645-657.	1.1	47
150	Tumor testing to identify lynch syndrome in two Australian colorectal cancer cohorts. Journal of Gastroenterology and Hepatology (Australia), 2017, 32, 427-438.	1.4	47
151	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.	3.0	45
152	Rare Mutations in RINT1 Predispose Carriers to Breast and Lynch Syndromeâ€“Spectrum Cancers. Cancer Discovery, 2014, 4, 804-815.	7.7	44
153	Common Genetic Variation In Cellular Transport Genes and Epithelial Ovarian Cancer (EOC) Risk. PLoS ONE, 2015, 10, e0128106.	1.1	44
154	Estrogen Receptor Polymorphism at Codon 325 and Risk of Breast Cancer in Women Before Age Forty. Journal of the National Cancer Institute, 1998, 90, 532-536.	3.0	43
155	AfterhMSH2 andhMLH1?what next? Analysis of three-generational, population-based, early-onset colorectal cancer families. International Journal of Cancer, 2002, 102, 166-171.	2.3	43
156	Genetic predisposition to ductal carcinoma in situ of the breast. Breast Cancer Research, 2016, 18, 22.	2.2	43
157	Reproductive profiles and risk of breast cancer subtypes: a multi-center case-only study. Breast Cancer Research, 2017, 19, 119.	2.2	43
158	Prevalence of PALB2 mutations in Australasian multiple-case breast cancer families. Breast Cancer Research, 2013, 15, R17.	2.2	42
159	Log odds of carrying an Ancestral Mutation in BRCA1 or BRCA2for a Defined personal and family history in an Ashkenazi Jewish woman (LAMBDA). Breast Cancer Research, 2003, 5, R206-16.	2.2	40
160	PALB2 and breast cancer: ready for clinical translation!. The Application of Clinical Genetics, 2013, 6, 43.	1.4	40
161	Cell-type-specific enrichment of risk-associated regulatory elements at ovarian cancer susceptibility loci. Human Molecular Genetics, 2015, 24, 3595-3607.	1.4	40
162	Fine-mapping identifies two additional breast cancer susceptibility loci at 9q31.2. Human Molecular Genetics, 2015, 24, 2966-2984.	1.4	40

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163	Role of tumour molecular and pathology features to estimate colorectal cancer risk for first-degree relatives. Gut, 2015, 64, 101-110.	6.1	40
164	Germline Sequencing DNA Repair Genes in 5545 Men With Aggressive and Nonaggressive Prostate Cancer. Journal of the National Cancer Institute, 2021, 113, 616-625.	3.0	40
165	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, 769-774.	1.1	39
166	Genetic Predisposition to In Situ and Invasive Lobular Carcinoma of the Breast. PLoS Genetics, 2014, 10, e1004285.	1.5	39
167	Homologous recombination DNA repair defects in PALB2-associated breast cancers. Npj Breast Cancer, 2019, 5, 23.	2.3	39
168	Breast Cancer Polygenic Risk Score and Contralateral Breast Cancer Risk. American Journal of Human Genetics, 2020, 107, 837-848.	2.6	39
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170	Rare Germline Variants in ATM Predispose to Prostate Cancer: A PRACTICAL Consortium Study. European Urology Oncology, 2021, 4, 570-579.	2.6	38
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