

Melissa C. Southey

List of Publications by Year in descending order

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Version: 2024-02-01

572
papers

46,452
citations

2426

97
h-index

3031

188
g-index

603
all docs

603
docs citations

603
times ranked

37269
citing authors

| # | ARTICLE | IF | CITATIONS |
|----|---|------|-----------|
| 1 | Genome-wide association study identifies novel breast cancer susceptibility loci. <i>Nature</i> , 2007, 447, 1087-1093. | 13.7 | 2,165 |
| 2 | Risks of Breast, Ovarian, and Contralateral Breast Cancer for <i>BRCA1</i> and <i>BRCA2</i> Mutation Carriers. <i>JAMA - Journal of the American Medical Association</i> , 2017, 317, 2402. | 3.8 | 1,898 |
| 3 | Association analysis identifies 65 new breast cancer risk loci. <i>Nature</i> , 2017, 551, 92-94. | 13.7 | 1,099 |
| 4 | Large-scale genotyping identifies 41 new loci associated with breast cancer risk. <i>Nature Genetics</i> , 2013, 45, 353-361. | 9.4 | 960 |
| 5 | Multiple newly identified loci associated with prostate cancer susceptibility. <i>Nature Genetics</i> , 2008, 40, 316-321. | 9.4 | 796 |
| 6 | 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. <i>PLoS Medicine</i> , 2010, 7, e1000279. | 3.9 | 764 |
| 7 | Gene-Panel Sequencing and the Prediction of Breast-Cancer Risk. <i>New England Journal of Medicine</i> , 2015, 372, 2243-2257. | 13.9 | 764 |
| 8 | Breast-Cancer Risk in Families with Mutations in <i>PALB2</i> . <i>New England Journal of Medicine</i> , 2014, 371, 497-506. | 13.9 | 745 |
| 9 | Polygenic Risk Scores for Prediction of Breast Cancer and Breast Cancer Subtypes. <i>American Journal of Human Genetics</i> , 2019, 104, 21-34. | 2.6 | 711 |
| 10 | Association analyses of more than 140,000 men identify 63 new prostate cancer susceptibility loci. <i>Nature Genetics</i> , 2018, 50, 928-936. | 9.4 | 652 |
| 11 | Iron-Overload-Related Disease in <i>HFE</i> Hereditary Hemochromatosis. <i>New England Journal of Medicine</i> , 2008, 358, 221-230. | 13.9 | 649 |
| 12 | Associations of Breast Cancer Risk Factors With Tumor Subtypes: A Pooled Analysis From the Breast Cancer Association Consortium Studies. <i>Journal of the National Cancer Institute</i> , 2011, 103, 250-263. | 3.0 | 596 |
| 13 | A common coding variant in <i>CASP8</i> is associated with breast cancer risk. <i>Nature Genetics</i> , 2007, 39, 352-358. | 9.4 | 591 |
| 14 | Breast Cancer Risk Genes Association Analysis in More than 113,000 Women. <i>New England Journal of Medicine</i> , 2021, 384, 428-439. | 13.9 | 532 |
| 15 | A genome-wide association study identifies colorectal cancer susceptibility loci on chromosomes 10p14 and 8q23.3. <i>Nature Genetics</i> , 2008, 40, 623-630. | 9.4 | 514 |
| 16 | Pathology of Breast and Ovarian Cancers among <i>BRCA1</i> and <i>BRCA2</i> Mutation Carriers: Results from the Consortium of Investigators of Modifiers of <i>BRCA1/2</i> (CIMBA). <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2012, 21, 134-147. | 1.1 | 513 |
| 17 | Genome-wide association analysis of more than 120,000 individuals identifies 15 new susceptibility loci for breast cancer. <i>Nature Genetics</i> , 2015, 47, 373-380. | 9.4 | 513 |
| 18 | Multiple independent variants at the <i>TERT</i> locus are associated with telomere length and risks of breast and ovarian cancer. <i>Nature Genetics</i> , 2013, 45, 371-384. | 9.4 | 493 |

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|----|--|-----|-----------|
| 19 | Identification of 23 new prostate cancer susceptibility loci using the iCOGS custom genotyping array. <i>Nature Genetics</i> , 2013, 45, 385-391. | 9.4 | 492 |
| 20 | Newly discovered breast cancer susceptibility loci on 3p24 and 17q23.2. <i>Nature Genetics</i> , 2009, 41, 585-590. | 9.4 | 434 |
| 21 | Prediction of Breast Cancer Risk Based on Profiling With Common Genetic Variants. <i>Journal of the National Cancer Institute</i> , 2015, 107, . | 3.0 | 428 |
| 22 | Genomic analyses identify hundreds of variants associated with age at menarche and support a role for puberty timing in cancer risk. <i>Nature Genetics</i> , 2017, 49, 834-841. | 9.4 | 426 |
| 23 | A meta-analysis of 87,040 individuals identifies 23 new susceptibility loci for prostate cancer. <i>Nature Genetics</i> , 2014, 46, 1103-1109. | 9.4 | 408 |
| 24 | Identification of seven new prostate cancer susceptibility loci through a genome-wide association study. <i>Nature Genetics</i> , 2009, 41, 1116-1121. | 9.4 | 389 |
| 25 | Discovery of common and rare genetic risk variants for colorectal cancer. <i>Nature Genetics</i> , 2019, 51, 76-87. | 9.4 | 377 |
| 26 | Genome-wide association studies identify four ER negative-specific breast cancer risk loci. <i>Nature Genetics</i> , 2013, 45, 392-398. | 9.4 | 374 |
| 27 | Large-scale genomic analyses link reproductive aging to hypothalamic signaling, breast cancer susceptibility and BRCA1-mediated DNA repair. <i>Nature Genetics</i> , 2015, 47, 1294-1303. | 9.4 | 357 |
| 28 | Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. <i>Nature Genetics</i> , 2017, 49, 680-691. | 9.4 | 356 |
| 29 | CWAS meta-analysis and replication identifies three new susceptibility loci for ovarian cancer. <i>Nature Genetics</i> , 2013, 45, 362-370. | 9.4 | 326 |
| 30 | Heterogeneity of Breast Cancer Associations with Five Susceptibility Loci by Clinical and Pathological Characteristics. <i>PLoS Genetics</i> , 2008, 4, e1000054. | 1.5 | 315 |
| 31 | A locus on 19p13 modifies risk of breast cancer in BRCA1 mutation carriers and is associated with hormone receptor-negative breast cancer in the general population. <i>Nature Genetics</i> , 2010, 42, 885-892. | 9.4 | 309 |
| 32 | Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. <i>Nature Genetics</i> , 2017, 49, 1767-1778. | 9.4 | 289 |
| 33 | Colorectal and Other Cancer Risks for Carriers and Noncarriers From Families With a DNA Mismatch Repair Gene Mutation: A Prospective Cohort Study. <i>Journal of Clinical Oncology</i> , 2012, 30, 958-964. | 0.8 | 286 |
| 34 | Breast Cancer Risk From Modifiable and Nonmodifiable Risk Factors Among White Women in the United States. <i>JAMA Oncology</i> , 2016, 2, 1295. | 3.4 | 285 |
| 35 | A common variant at the TERT-CLPTM1L locus is associated with estrogen receptor-negative breast cancer. <i>Nature Genetics</i> , 2011, 43, 1210-1214. | 9.4 | 279 |
| 36 | A genome-wide association study identifies a new ovarian cancer susceptibility locus on 9p22.2. <i>Nature Genetics</i> , 2009, 41, 996-1000. | 9.4 | 276 |

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|----|---|------|-----------|
| 37 | Multiple loci on 8q24 associated with prostate cancer susceptibility. <i>Nature Genetics</i> , 2009, 41, 1058-1060. | 9.4 | 273 |
| 38 | Cancer Risks Associated With Germline <i>PALB2</i> Pathogenic Variants: An International Study of 524 Families. <i>Journal of Clinical Oncology</i> , 2020, 38, 674-685. | 0.8 | 270 |
| 39 | Seven prostate cancer susceptibility loci identified by a multi-stage genome-wide association study. <i>Nature Genetics</i> , 2011, 43, 785-791. | 9.4 | 265 |
| 40 | Genome-wide association study identifies 32 novel breast cancer susceptibility loci from overall and subtype-specific analyses. <i>Nature Genetics</i> , 2020, 52, 572-581. | 9.4 | 265 |
| 41 | Trans-ancestry genome-wide association meta-analysis of prostate cancer identifies new susceptibility loci and informs genetic risk prediction. <i>Nature Genetics</i> , 2021, 53, 65-75. | 9.4 | 264 |
| 42 | Genome-wide association analysis identifies three new breast cancer susceptibility loci. <i>Nature Genetics</i> , 2012, 44, 312-318. | 9.4 | 256 |
| 43 | The Breast Cancer Family Registry: an infrastructure for cooperative multinational, interdisciplinary and translational studies of the genetic epidemiology of breast cancer. <i>Breast Cancer Research</i> , 2004, 6, R375-89. | 2.2 | 255 |
| 44 | Genome-Wide Association Study in BRCA1 Mutation Carriers Identifies Novel Loci Associated with Breast and Ovarian Cancer Risk. <i>PLoS Genetics</i> , 2013, 9, e1003212. | 1.5 | 244 |
| 45 | The histologic phenotypes of breast carcinoma occurring before age 40 years in women with and without BRCA1 or BRCA2 germline mutations. <i>Cancer</i> , 1998, 83, 2335-2345. | 2.0 | 243 |
| 46 | Metachronous colorectal cancer risk for mismatch repair gene mutation carriers: the advantage of more extensive colon surgery. <i>Gut</i> , 2011, 60, 950-957. | 6.1 | 227 |
| 47 | Mutational spectrum in a worldwide study of 29,700 families with <i>BRCA1</i> or <i>BRCA2</i> mutations. <i>Human Mutation</i> , 2018, 39, 593-620. | 1.1 | 224 |
| 48 | Identification of six new susceptibility loci for invasive epithelial ovarian cancer. <i>Nature Genetics</i> , 2015, 47, 164-171. | 9.4 | 221 |
| 49 | Functional Variants at the 11q13 Risk Locus for Breast Cancer Regulate Cyclin D1 Expression through Long-Range Enhancers. <i>American Journal of Human Genetics</i> , 2013, 92, 489-503. | 2.6 | 201 |
| 50 | Hypomethylation of smoking-related genes is associated with future lung cancer in four prospective cohorts. <i>Nature Communications</i> , 2015, 6, 10192. | 5.8 | 197 |
| 51 | Use of Molecular Tumor Characteristics to Prioritize Mismatch Repair Gene Testing in Early-Onset Colorectal Cancer. <i>Journal of Clinical Oncology</i> , 2005, 23, 6524-6532. | 0.8 | 194 |
| 52 | Rare variants in the ATM gene and risk of breast cancer. <i>Breast Cancer Research</i> , 2011, 13, R73. | 2.2 | 188 |
| 53 | A transcriptome-wide association study of 229,000 women identifies new candidate susceptibility genes for breast cancer. <i>Nature Genetics</i> , 2018, 50, 968-978. | 9.4 | 184 |
| 54 | Genetic insights into biological mechanisms governing human ovarian ageing. <i>Nature</i> , 2021, 596, 393-397. | 13.7 | 183 |

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|----|---|-----|-----------|
| 55 | Identification of nine new susceptibility loci for endometrial cancer. <i>Nature Communications</i> , 2018, 9, 3166. | 5.8 | 178 |
| 56 | <i>PALB2</i> , <i>CHEK2</i> and <i>ATM</i> rare variants and cancer risk: data from COGS. <i>Journal of Medical Genetics</i> , 2016, 53, 800-811. | 1.5 | 174 |
| 57 | A meta-analysis of genome-wide association studies of breast cancer identifies two novel susceptibility loci at 6q14 and 20q11. <i>Human Molecular Genetics</i> , 2012, 21, 5373-5384. | 1.4 | 168 |
| 58 | Rare, Evolutionarily Unlikely Missense Substitutions in <i>ATM</i> Confer Increased Risk of Breast Cancer. <i>American Journal of Human Genetics</i> , 2009, 85, 427-446. | 2.6 | 165 |
| 59 | <i>CHEK2</i> *1100delC Heterozygosity in Women With Breast Cancer Associated With Early Death, Breast Cancer-Specific Death, and Increased Risk of a Second Breast Cancer. <i>Journal of Clinical Oncology</i> , 2012, 30, 4308-4316. | 0.8 | 162 |
| 60 | Genome-Wide Meta-Analyses of Breast, Ovarian, and Prostate Cancer Association Studies Identify Multiple New Susceptibility Loci Shared by at Least Two Cancer Types. <i>Cancer Discovery</i> , 2016, 6, 1052-1067. | 7.7 | 157 |
| 61 | <i>BRCA2</i> Mutation-associated Breast Cancers Exhibit a Distinguishing Phenotype Based on Morphology and Molecular Profiles From Tissue Microarrays. <i>American Journal of Surgical Pathology</i> , 2007, 31, 121-128. | 2.1 | 156 |
| 62 | Adaptive evolution of the tumour suppressor <i>BRCA1</i> in humans and chimpanzees. <i>Nature Genetics</i> , 2000, 25, 410-413. | 9.4 | 153 |
| 63 | Genetic and Histopathologic Evaluation of <i>BRCA1</i> and <i>BRCA2</i> DNA Sequence Variants of Unknown Clinical Significance. <i>Cancer Research</i> , 2006, 66, 2019-2027. | 0.4 | 153 |
| 64 | DNA methylation-based biological aging and cancer risk and survival: Pooled analysis of seven prospective studies. <i>International Journal of Cancer</i> , 2018, 142, 1611-1619. | 2.3 | 153 |
| 65 | Low penetrance breast cancer susceptibility loci are associated with specific breast tumor subtypes: findings from the Breast Cancer Association Consortium. <i>Human Molecular Genetics</i> , 2011, 20, 3289-3303. | 1.4 | 152 |
| 66 | Age- and Tumor Subtype-Specific Breast Cancer Risk Estimates for <i>CH</i> <i>EK</i> <i>2</i> *1100delC Carriers. <i>Journal of Clinical Oncology</i> , 2016, 34, 2750-2760. | 0.8 | 152 |
| 67 | Prediction of Breast and Prostate Cancer Risks in Male <i>BRCA1</i> and <i>BRCA2</i> Mutation Carriers Using Polygenic Risk Scores. <i>Journal of Clinical Oncology</i> , 2017, 35, 2240-2250. | 0.8 | 152 |
| 68 | Cancer Risks For Mismatch Repair Gene Mutation Carriers: A Population-Based Early Onset Case-Family Study. <i>Clinical Gastroenterology and Hepatology</i> , 2006, 4, 489-498. | 2.4 | 151 |
| 69 | Familial Risks, Early-Onset Breast Cancer, and <i>BRCA1</i> and <i>BRCA2</i> Germline Mutations. <i>Journal of the National Cancer Institute</i> , 2003, 95, 448-457. | 3.0 | 150 |
| 70 | Multiple Novel Prostate Cancer Predisposition Loci Confirmed by an International Study: The PRACTICAL Consortium. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2008, 17, 2052-2061. | 1.1 | 148 |
| 71 | Tamoxifen and Risk of Contralateral Breast Cancer for <i>BRCA1</i> and <i>BRCA2</i> Mutation Carriers. <i>Journal of Clinical Oncology</i> , 2013, 31, 3091-3099. | 0.8 | 148 |
| 72 | Constitutional Methylation of the <i>BRCA1</i> Promoter Is Specifically Associated with <i>BRCA1</i> Mutation-Associated Pathology in Early-Onset Breast Cancer. <i>Cancer Prevention Research</i> , 2011, 4, 23-33. | 0.7 | 147 |

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|----|--|-----|-----------|
| 73 | Genome-wide association study identifies multiple susceptibility loci for diffuse large B cell lymphoma. <i>Nature Genetics</i> , 2014, 46, 1233-1238. | 9.4 | 147 |
| 74 | Genome-wide association study identifies 25 known breast cancer susceptibility loci as risk factors for triple-negative breast cancer. <i>Carcinogenesis</i> , 2014, 35, 1012-1019. | 1.3 | 145 |
| 75 | Epigenetic analysis leads to identification of HNF1B as a subtype-specific susceptibility gene for ovarian cancer. <i>Nature Communications</i> , 2013, 4, 1628. | 5.8 | 144 |
| 76 | Within-sibship genome-wide association analyses decrease bias in estimates of direct genetic effects. <i>Nature Genetics</i> , 2022, 54, 581-592. | 9.4 | 142 |
| 77 | Socioeconomic position, lifestyle habits and biomarkers of epigenetic aging: a multi-cohort analysis. <i>Aging</i> , 2019, 11, 2045-2070. | 1.4 | 137 |
| 78 | Evidence of Gene-Environment Interactions between Common Breast Cancer Susceptibility Loci and Established Environmental Risk Factors. <i>PLoS Genetics</i> , 2013, 9, e1003284. | 1.5 | 136 |
| 79 | Breast Cancer Prognosis in <i>BRCA1</i> and <i>BRCA2</i> Mutation Carriers: An International Prospective Breast Cancer Family Registry Population-Based Cohort Study. <i>Journal of Clinical Oncology</i> , 2012, 30, 19-26. | 0.8 | 134 |
| 80 | Oral Contraceptive Use and Risk of Early-Onset Breast Cancer in Carriers and Noncarriers of <i>BRCA1</i> and <i>BRCA2</i> Mutations. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2005, 14, 350-356. | 1.1 | 133 |
| 81 | Novel Common Genetic Susceptibility Loci for Colorectal Cancer. <i>Journal of the National Cancer Institute</i> , 2019, 111, 146-157. | 3.0 | 129 |
| 82 | Colorectal carcinomas with <i>KRAS</i> mutation are associated with distinctive morphological and molecular features. <i>Modern Pathology</i> , 2013, 26, 825-834. | 2.9 | 126 |
| 83 | Breast cancer risk variants at 6q25 display different phenotype associations and regulate <i>ESR1</i> , <i>RMND1</i> and <i>CCDC170</i> . <i>Nature Genetics</i> , 2016, 48, 374-386. | 9.4 | 125 |
| 84 | HFE C282Y homozygotes are at increased risk of breast and colorectal cancer. <i>Hepatology</i> , 2010, 51, 1311-1318. | 3.6 | 123 |
| 85 | Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. <i>Nature Genetics</i> , 2020, 52, 56-73. | 9.4 | 120 |
| 86 | A meta-analysis of genome-wide association studies to identify prostate cancer susceptibility loci associated with aggressive and non-aggressive disease. <i>Human Molecular Genetics</i> , 2013, 22, 408-415. | 1.4 | 118 |
| 87 | Genetically Predicted Body Mass Index and Breast Cancer Risk: Mendelian Randomization Analyses of Data from 145,000 Women of European Descent. <i>PLoS Medicine</i> , 2016, 13, e1002105. | 3.9 | 118 |
| 88 | <i>PIK3CA</i> Activating Mutation in Colorectal Carcinoma: Associations with Molecular Features and Survival. <i>PLoS ONE</i> , 2013, 8, e65479. | 1.1 | 117 |
| 89 | 10-year performance of four models of breast cancer risk: a validation study. <i>Lancet Oncology</i> , The, 2019, 20, 504-517. | 5.1 | 116 |
| 90 | <i>BRCA1</i> and <i>BRCA2</i> Mutation Carriers, Oral Contraceptive Use, and Breast Cancer Before Age 50. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2006, 15, 1863-1870. | 1.1 | 115 |

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|-----|---|-----|-----------|
| 91 | DNA methylation changes measured in pre-diagnostic peripheral blood samples are associated with smoking and lung cancer risk. <i>International Journal of Cancer</i> , 2017, 140, 50-61. | 2.3 | 115 |
| 92 | A genome wide linkage search for breast cancer susceptibility genes. <i>Genes Chromosomes and Cancer</i> , 2006, 45, 646-655. | 1.5 | 111 |
| 93 | Cumulative Burden of Colorectal Cancer-Associated Genetic Variants Is More Strongly Associated With Early-Onset vs Late-Onset Cancer. <i>Gastroenterology</i> , 2020, 158, 1274-1286.e12. | 0.6 | 110 |
| 94 | Common variants in ZNF365 are associated with both mammographic density and breast cancer risk. <i>Nature Genetics</i> , 2011, 43, 185-187. | 9.4 | 109 |
| 95 | Genome-wide association study identifies multiple loci associated with both mammographic density and breast cancer risk. <i>Nature Communications</i> , 2014, 5, 5303. | 5.8 | 109 |
| 96 | Risk of extracolonic cancers for people with biallelic and monoallelic mutations in <i>MUTYH</i> . <i>International Journal of Cancer</i> , 2016, 139, 1557-1563. | 2.3 | 107 |
| 97 | 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. <i>Human Molecular Genetics</i> , 2016, 25, 2256-2268. | 1.4 | 106 |
| 98 | Association of DNA Methylation-Based Biological Age With Health Risk Factors and Overall and Cause-Specific Mortality. <i>American Journal of Epidemiology</i> , 2018, 187, 529-538. | 1.6 | 106 |
| 99 | Identification of a BRCA2-Specific Modifier Locus at 6p24 Related to Breast Cancer Risk. <i>PLoS Genetics</i> , 2013, 9, e1003173. | 1.5 | 105 |
| 100 | Evidence that breast cancer risk at the 2q35 locus is mediated through IGFBP5 regulation. <i>Nature Communications</i> , 2014, 5, 4999. | 5.8 | 105 |
| 101 | A PALB2 mutation associated with high risk of breast cancer. <i>Breast Cancer Research</i> , 2010, 12, R109. | 2.2 | 102 |
| 102 | <i>HFE</i> C282Y/H63D compound heterozygotes are at low risk of hemochromatosis-related morbidity. <i>Hepatology</i> , 2009, 50, 94-101. | 3.6 | 101 |
| 103 | Common Breast Cancer Susceptibility Variants in <i>LSP1</i> and <i>RAD51L1</i> Are Associated with Mammographic Density Measures that Predict Breast Cancer Risk. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2012, 21, 1156-1166. | 1.1 | 101 |
| 104 | 19p13.1 Is a Triple-Negative-Specific Breast Cancer Susceptibility Locus. <i>Cancer Research</i> , 2012, 72, 1795-1803. | 0.4 | 100 |
| 105 | Cross-Cancer Genome-Wide Analysis of Lung, Ovary, Breast, Prostate, and Colorectal Cancer Reveals Novel Pleiotropic Associations. <i>Cancer Research</i> , 2016, 76, 5103-5114. | 0.4 | 100 |
| 106 | Risk of Estrogen Receptor-Positive and -Negative Breast Cancer and Single-Nucleotide Polymorphism 2q35-rs13387042. <i>Journal of the National Cancer Institute</i> , 2009, 101, 1012-1018. | 3.0 | 99 |
| 107 | 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. <i>Breast Cancer Research</i> , 2014, 16, R58. | 2.2 | 99 |
| 108 | Height and Breast Cancer Risk: Evidence From Prospective Studies and Mendelian Randomization. <i>Journal of the National Cancer Institute</i> , 2015, 107, djv219. | 3.0 | 99 |

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|-----|--|-----|-----------|
| 109 | Fine-Scale Mapping of the FGFR2 Breast Cancer Risk Locus: Putative Functional Variants Differentially Bind FOXA1 and E2F1. <i>American Journal of Human Genetics</i> , 2013, 93, 1046-1060. | 2.6 | 98 |
| 110 | Identification and molecular characterization of a new ovarian cancer susceptibility locus at 17q21.31. <i>Nature Communications</i> , 2013, 4, 1627. | 5.8 | 98 |
| 111 | After BRCA1 and BRCA2â€”What Next? Multifactorial Segregation Analyses of Three-Generation, Population-Based Australian Families Affected by Female Breast Cancer. <i>American Journal of Human Genetics</i> , 2001, 68, 420-431. | 2.6 | 97 |
| 112 | Refined histopathological predictors of BRCA1 and BRCA2 mutation status: a large-scale analysis of breast cancer characteristics from the BCAC, CIMBA, and ENIGMA consortia. <i>Breast Cancer Research</i> , 2014, 16, 3419. | 2.2 | 97 |
| 113 | Ethnicity and Risk for Colorectal Cancers Showing Somatic <i>BRAF</i> V600E Mutation or CpG Island Methylator Phenotype. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2008, 17, 1774-1780. | 1.1 | 96 |
| 114 | Breast Cancer Risk Prediction Using Clinical Models and 77 Independent Risk-Associated SNPs for Women Aged Under 50 Years: Australian Breast Cancer Family Registry. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2016, 25, 359-365. | 1.1 | 96 |
| 115 | Whole Exome Sequencing Suggests Much of Non-BRCA1/BRCA2 Familial Breast Cancer Is Due to Moderate and Low Penetrance Susceptibility Alleles. <i>PLoS ONE</i> , 2013, 8, e55681. | 1.1 | 95 |
| 116 | Comparison of mRNA Splicing Assay Protocols across Multiple Laboratories: Recommendations for Best Practice in Standardized Clinical Testing. <i>Clinical Chemistry</i> , 2014, 60, 341-352. | 1.5 | 95 |
| 117 | Causal effect of smoking on DNA methylation in peripheral blood: a twin and family study. <i>Clinical Epigenetics</i> , 2018, 10, 18. | 1.8 | 95 |
| 118 | Meta-analysis of genome-wide association studies discovers multiple loci for chronic lymphocytic leukemia. <i>Nature Communications</i> , 2016, 7, 10933. | 5.8 | 94 |
| 119 | No evidence that protein truncating variants in <i>BRIP1</i> are associated with breast cancer risk: implications for gene panel testing. <i>Journal of Medical Genetics</i> , 2016, 53, 298-309. | 1.5 | 94 |
| 120 | Cancer risks for monoallelic <i>MUTYH</i> mutation carriers with a family history of colorectal cancer. <i>International Journal of Cancer</i> , 2011, 129, 2256-2262. | 2.3 | 93 |
| 121 | Epigenome-wide methylation in DNA from peripheral blood as a marker of risk for breast cancer. <i>Breast Cancer Research and Treatment</i> , 2014, 148, 665-673. | 1.1 | 93 |
| 122 | Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. <i>Nature Communications</i> , 2016, 7, 11375. | 5.8 | 93 |
| 123 | Interplay between BRCA1 and RHAMM Regulates Epithelial Apicobasal Polarization and May Influence Risk of Breast Cancer. <i>PLoS Biology</i> , 2011, 9, e1001199. | 2.6 | 91 |
| 124 | A Cost-effectiveness Analysis of Multigene Testing for All Patients With Breast Cancer. <i>JAMA Oncology</i> , 2019, 5, 1718. | 3.4 | 91 |
| 125 | Penetrance Analysis of the <i>PALB2</i> c.1592delT Founder Mutation. <i>Clinical Cancer Research</i> , 2008, 14, 4667-4671. | 3.2 | 90 |
| 126 | Imputation and subset-based association analysis across different cancer types identifies multiple independent risk loci in the TERT-CLPTM1L region on chromosome 5p15.33. <i>Human Molecular Genetics</i> , 2014, 23, 6616-6633. | 1.4 | 90 |

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|-----|---|-----|-----------|
| 127 | Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. <i>Nature Communications</i> , 2019, 10, 1741. | 5.8 | 90 |
| 128 | Cancer Risks Associated With <i>BRCA1</i> and <i>BRCA2</i> Pathogenic Variants. <i>Journal of Clinical Oncology</i> , 2022, 40, 1529-1541. | 0.8 | 90 |
| 129 | Epigenetic supersimilarity of monozygotic twin pairs. <i>Genome Biology</i> , 2018, 19, 2. | 3.8 | 89 |
| 130 | Male breast cancer in <i>BRCA1</i> and <i>BRCA2</i> mutation carriers: pathology data from the Consortium of Investigators of Modifiers of <i>BRCA1/2</i> . <i>Breast Cancer Research</i> , 2016, 18, 15. | 2.2 | 88 |
| 131 | Joint associations of a polygenic risk score and environmental risk factors for breast cancer in the Breast Cancer Association Consortium. <i>International Journal of Epidemiology</i> , 2018, 47, 526-536. | 0.9 | 88 |
| 132 | Fine-mapping of prostate cancer susceptibility loci in a large meta-analysis identifies candidate causal variants. <i>Nature Communications</i> , 2018, 9, 2256. | 5.8 | 88 |
| 133 | Shared heritability and functional enrichment across six solid cancers. <i>Nature Communications</i> , 2019, 10, 431. | 5.8 | 88 |
| 134 | The Natural History of Serum Iron Indices for <i>HFE C282Y</i> Homozygosity Associated With Hereditary Hemochromatosis. <i>Gastroenterology</i> , 2008, 135, 1945-1952. | 0.6 | 86 |
| 135 | Common Genetic Variants and Modification of Penetrance of <i>BRCA2</i> -Associated Breast Cancer. <i>PLoS Genetics</i> , 2010, 6, e1001183. | 1.5 | 85 |
| 136 | Association of <i>ESR1</i> gene tagging SNPs with breast cancer risk. <i>Human Molecular Genetics</i> , 2009, 18, 1131-1139. | 1.4 | 84 |
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