

# Paolo Peterlongo

## List of Publications by Year in descending order

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Version: 2024-02-01

189  
papers

17,248  
citations

19657

61  
h-index

17592

121  
g-index

205  
all docs

205  
docs citations

205  
times ranked

19271  
citing authors

#	ARTICLE	IF	CITATIONS
1	Association analysis identifies 65 new breast cancer risk loci. <i>Nature</i> , 2017, 551, 92-94.	27.8	1,099
2	Large-scale genotyping identifies 41 new loci associated with breast cancer risk. <i>Nature Genetics</i> , 2013, 45, 353-361.	21.4	960
3	Breast-Cancer Risk in Families with Mutations in <i>PALB2</i> . <i>New England Journal of Medicine</i> , 2014, 371, 497-506.	27.0	745
4	Polygenic Risk Scores for Prediction of Breast Cancer and Breast Cancer Subtypes. <i>American Journal of Human Genetics</i> , 2019, 104, 21-34.	6.2	711
5	Parent-of-origin-specific allelic associations among 106 genomic loci for age at menarche. <i>Nature</i> , 2014, 514, 92-97.	27.8	548
6	Breast Cancer Risk Genes Association Analysis in More than 113,000 Women. <i>New England Journal of Medicine</i> , 2021, 384, 428-439.	27.0	532
7	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.	21.4	513
8	Multiple independent variants at the TERT locus are associated with telomere length and risks of breast and ovarian cancer. <i>Nature Genetics</i> , 2013, 45, 371-384.	21.4	493
9	Prediction of Breast Cancer Risk Based on Profiling With Common Genetic Variants. <i>Journal of the National Cancer Institute</i> , 2015, 107, .	6.3	428
10	Genome-wide association studies identify four ER negative-specific breast cancer risk loci. <i>Nature Genetics</i> , 2013, 45, 392-398.	21.4	374
11	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.	21.4	357
12	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. <i>Nature Genetics</i> , 2017, 49, 680-691.	21.4	356
13	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.	21.4	309
14	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. <i>Nature Genetics</i> , 2017, 49, 1767-1778.	21.4	289
15	C-kit mutations in core binding factor leukemias. <i>Blood</i> , 2000, 95, 726-728.	1.4	271
16	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.	1.6	270
17	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.	21.4	265
18	Common Breast Cancer-Predisposition Alleles Are Associated with Breast Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. <i>American Journal of Human Genetics</i> , 2008, 82, 937-948.	6.2	257

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19	Genome-wide association analysis identifies three new breast cancer susceptibility loci. <i>Nature Genetics</i> , 2012, 44, 312-318.	21.4	256
20	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.	3.5	244
21	Evaluation of Polygenic Risk Scores for Breast and Ovarian Cancer Risk Prediction in BRCA1 and BRCA2 Mutation Carriers. <i>Journal of the National Cancer Institute</i> , 2017, 109, .	6.3	242
22	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.	2.5	224
23	Identification of six new susceptibility loci for invasive epithelial ovarian cancer. <i>Nature Genetics</i> , 2015, 47, 164-171.	21.4	221
24	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.	6.2	201
25	TLR1 and TLR6 Polymorphisms Are Associated with Susceptibility to Invasive Aspergillosis after Allogeneic Stem Cell Transplantation. <i>Annals of the New York Academy of Sciences</i> , 2005, 1062, 95-103.	3.8	186
26	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.	21.4	184
27	<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.	3.2	174
28	Common Breast Cancer Susceptibility Alleles and the Risk of Breast Cancer for <i>BRCA1</i> and <i>BRCA2</i> Mutation Carriers: Implications for Risk Prediction. <i>Cancer Research</i> , 2010, 70, 9742-9754.	0.9	169
29	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.	9.4	157
30	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.	1.6	152
31	Evaluation of SNPs in <i>miR-146a</i> , <i>miR196a2</i> and <i>miR-499</i> as low-penetrance alleles in German and Italian familial breast cancer cases. <i>Human Mutation</i> , 2010, 31, E1052-E1057.	2.5	147
32	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.	21.4	125
33	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. <i>Nature Genetics</i> , 2020, 52, 56-73.	21.4	120
34	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.	8.4	118
35	The contribution of pathogenic variants in breast cancer susceptibility genes to familial breast cancer risk. <i>Npj Breast Cancer</i> , 2017, 3, 22.	5.2	108
36	Identification of a <i>BRCA2</i> -Specific Modifier Locus at 6p24 Related to Breast Cancer Risk. <i>PLoS Genetics</i> , 2013, 9, e1003173.	3.5	105

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37	Evidence that breast cancer risk at the 2q35 locus is mediated through IGFBP5 regulation. <i>Nature Communications</i> , 2014, 5, 4999.	12.8	105
38	19p13.1 Is a Triple-Negative-Specific Breast Cancer Susceptibility Locus. <i>Cancer Research</i> , 2012, 72, 1795-1803.	0.9	100
39	Common variants in LSP1, 2q35 and 8q24 and breast cancer risk for BRCA1 and BRCA2 mutation carriers. <i>Human Molecular Genetics</i> , 2009, 18, 4442-4456.	2.9	99
40	Height and Breast Cancer Risk: Evidence From Prospective Studies and Mendelian Randomization. <i>Journal of the National Cancer Institute</i> , 2015, 107, djv219.	6.3	99
41	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.	6.2	98
42	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.	5.0	97
43	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.	2.5	95
44	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.	3.2	94
45	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. <i>Nature Communications</i> , 2016, 7, 11375.	12.8	93
46	Interplay between BRCA1 and RHAMM Regulates Epithelial Apicobasal Polarization and May Influence Risk of Breast Cancer. <i>PLoS Biology</i> , 2011, 9, e1001199.	5.6	91
47	<i>FANCM</i> c.5791C>T nonsense mutation (rs144567652) induces exon skipping, affects DNA repair activity and is a familial breast cancer risk factor. <i>Human Molecular Genetics</i> , 2015, 24, 5345-5355.	2.9	91
48	Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. <i>Nature Communications</i> , 2019, 10, 1741.	12.8	90
49	Male breast cancer in BRCA1 and BRCA2 mutation carriers: pathology data from the Consortium of Investigators of Modifiers of BRCA1/2. <i>Breast Cancer Research</i> , 2016, 18, 15.	5.0	88
50	TGFB1*6A and Cancer: A Meta-Analysis of 12 Case-Control Studies. <i>Journal of Clinical Oncology</i> , 2004, 22, 756-758.	1.6	85
51	Common Genetic Variants and Modification of Penetrance of BRCA2-Associated Breast Cancer. <i>PLoS Genetics</i> , 2010, 6, e1001183.	3.5	85
52	Polygenic risk scores and breast and epithelial ovarian cancer risks for carriers of BRCA1 and BRCA2 pathogenic variants. <i>Genetics in Medicine</i> , 2020, 22, 1653-1666.	2.4	82
53	The role of genetic breast cancer susceptibility variants as prognostic factors. <i>Human Molecular Genetics</i> , 2012, 21, 3926-3939.	2.9	80
54	Common variants at 12p11, 12q24, 9p21, 9q31.2 and in ZNF365 are associated with breast cancer risk for BRCA1 and/or BRCA2 mutation carriers. <i>Breast Cancer Research</i> , 2012, 14, R33.	5.0	78

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55	Functional mechanisms underlying pleiotropic risk alleles at the 19p13.1 breast-ovarian cancer susceptibility locus. Nature Communications, 2016, 7, 12675.	12.8	78
56	BRCA2 Polymorphic Stop Codon K3326X and the Risk of Breast, Prostate, and Ovarian Cancers. Journal of the National Cancer Institute, 2016, 108, djv315.	6.3	77
57	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.	6.2	76
58	BRCA2 Hypomorphic Missense Variants Confer Moderate Risks of Breast Cancer. Cancer Research, 2017, 77, 2789-2799.	0.9	75
59	Clinical and pathologic characteristics of BRCA-positive and BRCA-negative male breast cancer patients: results from a collaborative multicenter study in Italy. Breast Cancer Research and Treatment, 2012, 134, 411-418.	2.5	73
60	Prospective study of breast tomosynthesis as a triage to assessment in screening. Breast Cancer Research and Treatment, 2012, 133, 267-271.	2.5	73
61	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.	5.0	71
62	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, 4693-4706.	2.9	71
63	Common alleles at 6q25.1 and 1p11.2 are associated with breast cancer risk for BRCA1 and BRCA2 mutation carriers. Human Molecular Genetics, 2011, 20, 3304-3321.	2.9	68
64	Individuals with FANCM biallelic mutations do not develop Fanconi anemia, but show risk for breast cancer, chemotherapy toxicity and may display chromosome fragility. Genetics in Medicine, 2018, 20, 452-457.	2.4	59
65	Associations of common breast cancer susceptibility alleles with risk of breast cancer subtypes in BRCA1 and BRCA2 mutation carriers. Breast Cancer Research, 2014, 16, 3416.	5.0	57
66	Colorectal cancer risk in individuals with biallelic or monoallelic mutations of MYH. International Journal of Cancer, 2005, 114, 505-507.	5.1	56
67	Identification of Novel Genetic Markers of Breast Cancer Survival. Journal of the National Cancer Institute, 2015, 107, .	6.3	56
68	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.	0.9	54
69	MSH6 germline mutations are rare in colorectal cancer families. International Journal of Cancer, 2003, 107, 571-579.	5.1	53
70	Common non-synonymous SNPs associated with breast cancer susceptibility: findings from the Breast Cancer Association Consortium. Human Molecular Genetics, 2014, 23, 6096-6111.	2.9	53
71	Genome-wide association study of germline variants and breast cancer-specific mortality. British Journal of Cancer, 2019, 120, 647-657.	6.4	52
72	Fine-scale mapping of 8q24 locus identifies multiple independent risk variants for breast cancer. International Journal of Cancer, 2016, 139, 1303-1317.	5.1	51

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73	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.	2.5	51
74	Pathology of Tumors Associated With Pathogenic Germline Variants in 9 Breast Cancer Susceptibility Genes. JAMA Oncology, 2022, 8, e216744.	7.1	51
75	Rare variants in XRCC2 as breast cancer susceptibility alleles: Table 1. Journal of Medical Genetics, 2012, 49, 618-620.	3.2	49
76	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, 1665-1675.	2.5	49
77	MicroRNA Related Polymorphisms and Breast Cancer Risk. PLoS ONE, 2014, 9, e109973.	2.5	49
78	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.	2.5	47
79	DNA Glycosylases Involved in Base Excision Repair May Be Associated with Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. PLoS Genetics, 2014, 10, e1004256.	3.5	47
80	TGFBR1 $\Delta$ 6A May Contribute to Hereditary Colorectal Cancer. Journal of Clinical Oncology, 2005, 23, 3074-3078.	1.6	45
81	Body mass index and breast cancer survival: a Mendelian randomization analysis. International Journal of Epidemiology, 2017, 46, 1814-1822.	1.9	45
82	Increased frequency of disease-causing MYH mutations in colon cancer families. Carcinogenesis, 2006, 27, 2243-2249.	2.8	44
83	Genetic predisposition to ductal carcinoma in situ of the breast. Breast Cancer Research, 2016, 18, 22.	5.0	43
84	Reproductive profiles and risk of breast cancer subtypes: a multi-center case-only study. Breast Cancer Research, 2017, 19, 119.	5.0	43
85	Genetic Variation at 9p22.2 and Ovarian Cancer Risk for BRCA1 and BRCA2 Mutation Carriers. Journal of the National Cancer Institute, 2011, 103, 105-116.	6.3	40
86	Fine-mapping identifies two additional breast cancer susceptibility loci at 9q31.2. Human Molecular Genetics, 2015, 24, 2966-2984.	2.9	40
87	Insight into genetic susceptibility to male breast cancer by multigene panel testing: Results from a multicenter study in Italy. International Journal of Cancer, 2019, 145, 390-400.	5.1	40
88	Genetic Predisposition to In Situ and Invasive Lobular Carcinoma of the Breast. PLoS Genetics, 2014, 10, e1004285.	3.5	39
89	Homologous recombination DNA repair defects in PALB2-associated breast cancers. Npj Breast Cancer, 2019, 5, 23.	5.2	39
90	Association of Genomic Domains in BRCA1 and BRCA2 with Prostate Cancer Risk and Aggressiveness. Cancer Research, 2020, 80, 624-638.	0.9	39

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91	Identification and characterization of novel associations in the CASP8/ALS2CR12 region on chromosome 2 with breast cancer risk. Human Molecular Genetics, 2015, 24, 285-298.	2.9	38
92	PALB2 germline mutations in familial breast cancer cases with personal and family history of pancreatic cancer. Breast Cancer Research and Treatment, 2011, 126, 825-828.	2.5	37
93	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.	6.2	37
94	COMPLEXO: identifying the missing heritability of breast cancer via next generation collaboration. Breast Cancer Research, 2013, 15, 402.	5.0	36
95	11q13 is a susceptibility locus for hormone receptor positive breast cancer. Human Mutation, 2012, 33, 1123-1132.	2.5	35
96	Ovarian cancer susceptibility alleles and risk of ovarian cancer in <i>BRCA1</i> and <i>BRCA2</i> mutation carriers. Human Mutation, 2012, 33, 690-702.	2.5	34
97	Assessing Associations between the AURKA-HMMR-TPX2-TUBG1 Functional Module and Breast Cancer Risk in <i>BRCA1/2</i> Mutation Carriers. PLoS ONE, 2015, 10, e0120020.	2.5	34
98	The spectrum of <i>BRCA1</i> and <i>BRCA2</i> pathogenic sequence variants in Middle Eastern, North African, and South European countries. Human Mutation, 2019, 40, e1-e23.	2.5	34
99	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.	2.9	33
100	Decapping protein EDC4 regulates DNA repair and phenocopies <i>BRCA1</i> . Nature Communications, 2018, 9, 967.	12.8	33
101	Common variants of the <i>BRCA1</i> wild-type allele modify the risk of breast cancer in <i>BRCA1</i> mutation carriers. Human Molecular Genetics, 2011, 20, 4732-4747.	2.9	32
102	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, 1934-1946.	2.9	32
103	Transcriptome-wide association study of breast cancer risk by estrogen receptor status. Genetic Epidemiology, 2020, 44, 442-468.	1.3	32
104	Identification of independent association signals and putative functional variants for breast cancer risk through fine-scale mapping of the 12p11 locus. Breast Cancer Research, 2016, 18, 64.	5.0	31
105	Whole-exome sequencing and targeted gene sequencing provide insights into the role of <i>PALB2</i> as a male breast cancer susceptibility gene. Cancer, 2017, 123, 210-218.	4.1	31
106	Height and Body Mass Index as Modifiers of Breast Cancer Risk in <i>BRCA1/2</i> Mutation Carriers: A Mendelian Randomization Study. Journal of the National Cancer Institute, 2019, 111, 350-364.	6.3	30
107	A network analysis to identify mediators of germline-driven differences in breast cancer prognosis. Nature Communications, 2020, 11, 312.	12.8	30
108	Germline mutations in BRIP1 and PALB2 in Jewish high cancer risk families. Familial Cancer, 2012, 11, 483-491.	1.9	29



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109	The SNP rs895819 in miR-27a is not associated with familial breast cancer risk in Italians. <i>Breast Cancer Research and Treatment</i> , 2012, 133, 805-807.	2.5	28
110	The FANCM:p.Arg658* truncating variant is associated with risk of triple-negative breast cancer. <i>Npj Breast Cancer</i> , 2019, 5, 38.	5.2	28
111	Confirmation of 5p12 As a Susceptibility Locus for Progesterone-Receptorâ€“Positive, Lower Grade Breast Cancer. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2011, 20, 2222-2231.	2.5	27
112	An original phylogenetic approach identified mitochondrial haplogroup T1a1 as inversely associated with breast cancer risk in BRCA2 mutation carriers. <i>Breast Cancer Research</i> , 2015, 17, 61.	5.0	26
113	Common germline polymorphisms associated with breast cancer-specific survival. <i>Breast Cancer Research</i> , 2015, 17, 58.	5.0	26
114	RAD51B in Familial Breast Cancer. <i>PLoS ONE</i> , 2016, 11, e0153788.	2.5	26
115	PALB2 sequencing in Italian familial breast cancer cases reveals a high-risk mutation recurrent in the province of Bergamo. <i>Genetics in Medicine</i> , 2014, 16, 688-694.	2.4	25
116	Novel and known genetic variants for male breast cancer risk at 8q24.21, 9p21.3, 11q13.3 and 14q24.1: Results from a multicenter study in Italy. <i>European Journal of Cancer</i> , 2015, 51, 2289-2295.	2.8	25
117	Contribution of MUTHYH Variants to Male Breast Cancer Risk: Results From a Multicenter Study in Italy. <i>Frontiers in Oncology</i> , 2018, 8, 583.	2.8	25
118	Fine-Scale Mapping of the 4q24 Locus Identifies Two Independent Loci Associated with Breast Cancer Risk. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2015, 24, 1680-1691.	2.5	24
119	SNPs in ultraconserved elements and familial breast cancer risk. <i>Carcinogenesis</i> , 2009, 30, 544-545.	2.8	23
120	Exploring the link between MORF4L1 and risk of breast cancer. <i>Breast Cancer Research</i> , 2011, 13, R40.	5.0	23
121	A Nonsynonymous Polymorphism in <i>IRS1</i> Modifies Risk of Developing Breast and Ovarian Cancers in <i>BRCA1</i> and Ovarian Cancer in <i>BRCA2</i> Mutation Carriers. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2012, 21, 1362-1370.	2.5	23
122	A possible role of FANCM mutations in male breast cancer susceptibility: Results from a multicenter study in Italy. <i>Breast</i> , 2018, 38, 92-97.	2.2	23
123	Polygenic risk modeling for prediction of epithelial ovarian cancer risk. <i>European Journal of Human Genetics</i> , 2022, 30, 349-362.	2.8	23
124	Candidate Genetic Modifiers for Breast and Ovarian Cancer Risk in <i>BRCA1</i> and <i>BRCA2</i> Mutation Carriers. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2015, 24, 308-316.	2.5	22
125	Population distribution and ancestry of the cancer protective MDM2 SNP285 (rs117039649). <i>Oncotarget</i> , 2014, 5, 8223-8234.	1.8	22
126	Association of genetic susceptibility variants for type 2 diabetes with breast cancer risk in women of European ancestry. <i>Cancer Causes and Control</i> , 2016, 27, 679-693.	1.8	21



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127	The p53 Arg72Pro and Ins16bp polymorphisms and their haplotypes are not associated with breast cancer risk in BRCA-mutation negative familial cases. <i>Cancer Detection and Prevention</i> , 2008, 32, 140-143.	2.1	20
128	Evidences for association of the CASP8 -652 6N del promoter polymorphism with age at diagnosis in familial breast cancer cases. <i>Breast Cancer Research and Treatment</i> , 2009, 113, 607-608.	2.5	20
129	Fine scale mapping of the 17q22 breast cancer locus using dense SNPs, genotyped within the Collaborative Oncological Gene-Environment Study (COGs). <i>Scientific Reports</i> , 2016, 6, 32512.	3.3	19
130	The <i>BRCA2</i> c.68-7T&gt;AA variant is not pathogenic: A model for clinical calibration of spliceogenicity. <i>Human Mutation</i> , 2018, 39, 729-741.	2.5	19
131	Mendelian randomisation study of height and body mass index as modifiers of ovarian cancer risk in 22,588 BRCA1 and BRCA2 mutation carriers. <i>British Journal of Cancer</i> , 2019, 121, 180-192.	6.4	19
132	A case-only study to identify genetic modifiers of breast cancer risk for BRCA1/BRCA2 mutation carriers. <i>Nature Communications</i> , 2021, 12, 1078.	12.8	19
133	Breast and Prostate Cancer Risks for Male<i>BRCA1</i>and<i>BRCA2</i>Pathogenic Variant Carriers Using Polygenic Risk Scores. <i>Journal of the National Cancer Institute</i> , 2022, 114, 109-122.	6.3	19
134	Meta-Analysis of Mismatch Repair Polymorphisms within the Cogent Consortium for Colorectal Cancer Susceptibility. <i>PLoS ONE</i> , 2013, 8, e72091.	2.5	19
135	Breast cancer risks associated with missense variants in breast cancer susceptibility genes. <i>Genome Medicine</i> , 2022, 14, 51.	8.2	19
136	Haplotype structure in Ashkenazi Jewish BRCA1 and BRCA2 mutation carriers. <i>Human Genetics</i> , 2011, 130, 685-699.	3.8	18
137	No clinical utility of KRAS variant rs61764370 for ovarian or breast cancer. <i>Gynecologic Oncology</i> , 2016, 141, 386-401.	1.4	18
138	Association of breast cancer risk in BRCA1 and BRCA2 mutation carriers with genetic variants showing differential allelic expression: identification of a modifier of breast cancer risk at locus 11q22.3. <i>Breast Cancer Research and Treatment</i> , 2017, 161, 117-134.	2.5	18
139	Toll-Like Receptor 4 Polymorphisms and Risk of Gram-Negative Bacteremia after Allogeneic Stem Cell Transplantation. A Prospective Pilot Study. <i>Biology of Blood and Marrow Transplantation</i> , 2009, 15, 1130-1133.	2.0	17
140	9q31.2-rs865686 as a Susceptibility Locus for Estrogen Receptor-Positive Breast Cancer: Evidence from the Breast Cancer Association Consortium. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2012, 21, 1783-1791.	2.5	17
141	No evidence for an association between the earwax-associated polymorphism in ABCC11 and breast cancer risk in Caucasian women. <i>Breast Cancer Research and Treatment</i> , 2011, 126, 235-239.	2.5	16
142	The predictive ability of the 313 variant-based polygenic risk score for contralateral breast cancer risk prediction in women of European ancestry with a heterozygous BRCA1 or BRCA2 pathogenic variant. <i>Genetics in Medicine</i> , 2021, 23, 1726-1737.	2.4	16
143	Beneficial Effect of Fluoxetine in a Case of Sporadic Hyperekplexia. <i>Clinical Neuropharmacology</i> , 2000, 23, 161-163.	0.7	15
144	<i>BRCA1</i> and <i>BRCA2</i> 5' noncoding region variants identified in breast cancer patients alter promoter activity and protein binding. <i>Human Mutation</i> , 2018, 39, 2025-2039.	2.5	15

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145	Germline mutations of AXIN2 are not associated with nonsyndromic colorectal cancer. <i>Human Mutation</i> , 2005, 25, 498-500.	2.5	14
146	BMP2 / BMP4 colorectal cancer susceptibility loci in northern and southern European populations. <i>Carcinogenesis</i> , 2013, 34, 314-318.	2.8	14
147	Genetic variation at CYP3A is associated with age at menarche and breast cancer risk: a case-control study. <i>Breast Cancer Research</i> , 2014, 16, R51.	5.0	14
148	Inherited variants in the inner centromere protein (INCENP) gene of the chromosomal passenger complex contribute to the susceptibility of ER-negative breast cancer. <i>Carcinogenesis</i> , 2015, 36, 256-271.	2.8	14
149	The rs12975333 variant in the miR-125a and breast cancer risk in Germany, Italy, Australia and Spain. <i>Journal of Medical Genetics</i> , 2011, 48, 703-704.	3.2	13
150	Localization of Cancer Susceptibility Genes by Genome-wide Single-Nucleotide Polymorphism Linkage-Disequilibrium Mapping. <i>Cancer Research</i> , 2004, 64, 8116-8125.	0.9	12
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