

Paolo Peterlongo

List of Publications by Citations

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184
papers

12,376
citations

58
h-index

107
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205
ext. papers

15,228
ext. citations

9.7
avg, IF

4.11
L-index

#	Paper	IF	Citations
184	Large-scale genotyping identifies 41 new loci associated with breast cancer risk. <i>Nature Genetics</i> , 2013 , 45, 353-61, 361e1-2	36.3	813
183	Association analysis identifies 65 new breast cancer risk loci. <i>Nature</i> , 2017 , 551, 92-94	50.4	643
182	Breast-cancer risk in families with mutations in PALB2. <i>New England Journal of Medicine</i> , 2014 , 371, 497-506	50.6	576
181	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, 371-84, 384e1-2	36.3	422
180	Genome-wide association analysis of more than 120,000 individuals identifies 15 new susceptibility loci for breast cancer. <i>Nature Genetics</i> , 2015 , 47, 373-80	36.3	406
179	Parent-of-origin-specific allelic associations among 106 genomic loci for age at menarche. <i>Nature</i> , 2014 , 514, 92-97	50.4	401
178	Polygenic Risk Scores for Prediction of Breast Cancer and Breast Cancer Subtypes. <i>American Journal of Human Genetics</i> , 2019 , 104, 21-34	11	363
177	Genome-wide association studies identify four ER negative-specific breast cancer risk loci. <i>Nature Genetics</i> , 2013 , 45, 392-8, 398e1-2	36.3	327
176	Prediction of breast cancer risk based on profiling with common genetic variants. <i>Journal of the National Cancer Institute</i> , 2015 , 107,	9.7	324
175	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, 885-92	36.3	276
174	C-kit mutations in core binding factor leukemias. <i>Blood</i> , 2000 , 95, 726-728	2.2	255
173	Genome-wide association analysis identifies three new breast cancer susceptibility loci. <i>Nature Genetics</i> , 2012 , 44, 312-8	36.3	237
172	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	36.3	226
171	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, 937-48	11	218
170	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	6	209
169	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. <i>Nature Genetics</i> , 2017 , 49, 680-691	36.3	190
168	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. <i>Nature Genetics</i> , 2017 , 49, 1767-1778	36.3	186

167	Identification of six new susceptibility loci for invasive epithelial ovarian cancer. <i>Nature Genetics</i> , 2015 , 47, 164-71	36.3	177
166	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	6.5	169
165	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	11	167
164	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,	9.7	153
163	Common breast cancer susceptibility alleles and the risk of breast cancer for BRCA1 and BRCA2 mutation carriers: implications for risk prediction. <i>Cancer Research</i> , 2010 , 70, 9742-54	10.1	147
162	Breast Cancer Risk Genes - Association Analysis in More than 113,000 Women. <i>New England Journal of Medicine</i> , 2021 , 384, 428-439	59.2	143
161	Mutational spectrum in a worldwide study of 29,700 families with BRCA1 or BRCA2 mutations. <i>Human Mutation</i> , 2018 , 39, 593-620	4.7	138
160	Evaluation of SNPs in miR-146a, miR196a2 and miR-499 as low-penetrance alleles in German and Italian familial breast cancer cases. <i>Human Mutation</i> , 2010 , 31, E1052-7	4.7	138
159	Cancer Risks Associated With Germline Pathogenic Variants: An International Study of 524 Families. <i>Journal of Clinical Oncology</i> , 2020 , 38, 674-685	2.2	133
158	PALB2, CHEK2 and ATM rare variants and cancer risk: data from COGS. <i>Journal of Medical Genetics</i> , 2016 , 53, 800-811	5.8	121
157	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, 1052-67	24.4	104
156	Prediction of Breast and Prostate Cancer Risks in Male BRCA1 and BRCA2 Mutation Carriers Using Polygenic Risk Scores. <i>Journal of Clinical Oncology</i> , 2017 , 35, 2240-2250	2.2	101
155	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	36.3	101
154	Breast cancer risk variants at 6q25 display different phenotype associations and regulate ESR1, RMND1 and CCDC170. <i>Nature Genetics</i> , 2016 , 48, 374-86	36.3	93
153	19p13.1 is a triple-negative-specific breast cancer susceptibility locus. <i>Cancer Research</i> , 2012 , 72, 1795-803	30.1	93
152	Common variants in LSP1, 2q35 and 8q24 and breast cancer risk for BRCA1 and BRCA2 mutation carriers. <i>Human Molecular Genetics</i> , 2009 , 18, 4442-56	5.6	91
151	Identification of a BRCA2-specific modifier locus at 6p24 related to breast cancer risk. <i>PLoS Genetics</i> , 2013 , 9, e1003173	6	90
150	Evidence that breast cancer risk at the 2q35 locus is mediated through IGFBP5 regulation. <i>Nature Communications</i> , 2014 , 4, 4999	17.4	87

149	No evidence that protein truncating variants in BRIP1 are associated with breast cancer risk: implications for gene panel testing. <i>Journal of Medical Genetics</i> , 2016 , 53, 298-309	5.8	83
148	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	8.3	82
147	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, 1046-60	11	80
146	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	11.6	80
145	The contribution of pathogenic variants in breast cancer susceptibility genes to familial breast cancer risk. <i>Npj Breast Cancer</i> , 2017 , 3, 22	7.8	78
144	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	3.7	77
143	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	36.3	76
142	The role of genetic breast cancer susceptibility variants as prognostic factors. <i>Human Molecular Genetics</i> , 2012 , 21, 3926-39	5.6	75
141	TGFB1*6A and cancer: a meta-analysis of 12 case-control studies. <i>Journal of Clinical Oncology</i> , 2004 , 22, 756-8	2.2	75
140	Height and Breast Cancer Risk: Evidence From Prospective Studies and Mendelian Randomization. <i>Journal of the National Cancer Institute</i> , 2015 , 107,	9.7	74
139	Common genetic variants and modification of penetrance of BRCA2-associated breast cancer. <i>PLoS Genetics</i> , 2010 , 6, e1001183	6	74
138	Interplay between BRCA1 and RHAMM regulates epithelial apicobasal polarization and may influence risk of breast cancer. <i>PLoS Biology</i> , 2011 , 9, e1001199	9.7	73
137	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	8.3	70
136	FANCM 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, 5345-55	5.6	68
135	Prospective study of breast tomosynthesis as a triage to assessment in screening. <i>Breast Cancer Research and Treatment</i> , 2012 , 133, 267-71	4.4	66
134	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. <i>Human Molecular Genetics</i> , 2011 , 20, 4693-706	5.6	66
133	BRCA2 Polymorphic Stop Codon K3326X and the Risk of Breast, Prostate, and Ovarian Cancers. <i>Journal of the National Cancer Institute</i> , 2016 , 108,	9.7	65
132	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. <i>Nature Communications</i> , 2016 , 7, 11375	17.4	64

131	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. <i>Breast Cancer Research</i> , 2011 , 13, R110	8.3	62
130	Common alleles at 6q25.1 and 1p11.2 are associated with breast cancer risk for BRCA1 and BRCA2 mutation carriers. <i>Human Molecular Genetics</i> , 2011 , 20, 3304-21	5.6	62
129	Fine-scale mapping of the 5q11.2 breast cancer locus reveals at least three independent risk variants regulating MAP3K1. <i>American Journal of Human Genetics</i> , 2015 , 96, 5-20	11	59
128	Clinical and pathologic characteristics of BRCA-positive and BRCA-negative male breast cancer patients: results from a collaborative multicenter study in Italy. <i>Breast Cancer Research and Treatment</i> , 2012 , 134, 411-8	4.4	58
127	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	8.3	58
126	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. <i>Nature Genetics</i> , 2020 , 52, 56-73	36.3	56
125	Functional mechanisms underlying pleiotropic risk alleles at the 19p13.1 breast-ovarian cancer susceptibility locus. <i>Nature Communications</i> , 2016 , 7, 12675	17.4	53
124	Colorectal cancer risk in individuals with biallelic or monoallelic mutations of MYH. <i>International Journal of Cancer</i> , 2005 , 114, 505-7	7.5	52
123	Hypomorphic Missense Variants Confer Moderate Risks of Breast Cancer. <i>Cancer Research</i> , 2017 , 77, 2789-2799	10.1	49
122	MSH6 germline mutations are rare in colorectal cancer families. <i>International Journal of Cancer</i> , 2003 , 107, 571-9	7.5	49
121	Comparison of 6q25 breast cancer hits from Asian and European Genome Wide Association Studies in the Breast Cancer Association Consortium (BCAC). <i>PLoS ONE</i> , 2012 , 7, e42380	3.7	49
120	Common non-synonymous SNPs associated with breast cancer susceptibility: findings from the Breast Cancer Association Consortium. <i>Human Molecular Genetics</i> , 2014 , 23, 6096-111	5.6	48
119	Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. <i>Nature Communications</i> , 2019 , 10, 1741	17.4	47
118	Associations of common breast cancer susceptibility alleles with risk of breast cancer subtypes in BRCA1 and BRCA2 mutation carriers. <i>Breast Cancer Research</i> , 2014 , 16, 3416	8.3	46
117	Individuals with FANCM biallelic mutations do not develop Fanconi anemia, but show risk for breast cancer, chemotherapy toxicity and may display chromosome fragility. <i>Genetics in Medicine</i> , 2018 , 20, 452-457	8.1	44
116	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. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2012 , 21, 645-57	4	44
115	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. <i>Human Mutation</i> , 2012 , 33, 1665-75	4.7	42
114	TGFBR1*6A may contribute to hereditary colorectal cancer. <i>Journal of Clinical Oncology</i> , 2005 , 23, 3074-8.2	4.2	41

113	Increased frequency of disease-causing MYH mutations in colon cancer families. <i>Carcinogenesis</i> , 2006 , 27, 2243-9	4.6	39
112	Identification of novel genetic markers of breast cancer survival. <i>Journal of the National Cancer Institute</i> , 2015 , 107,	9.7	38
111	Genetic predisposition to in situ and invasive lobular carcinoma of the breast. <i>PLoS Genetics</i> , 2014 , 10, e1004285	6	38
110	MicroRNA related polymorphisms and breast cancer risk. <i>PLoS ONE</i> , 2014 , 9, e109973	3.7	37
109	Rare variants in XRCC2 as breast cancer susceptibility alleles. <i>Journal of Medical Genetics</i> , 2012 , 49, 618-208	4.8	37
108	Genetic variation at 9p22.2 and ovarian cancer risk for BRCA1 and BRCA2 mutation carriers. <i>Journal of the National Cancer Institute</i> , 2011 , 103, 105-16	9.7	37
107	Fine-mapping identifies two additional breast cancer susceptibility loci at 9q31.2. <i>Human Molecular Genetics</i> , 2015 , 24, 2966-84	5.6	36
106	Identification and characterization of novel associations in the CASP8/ALS2CR12 region on chromosome 2 with breast cancer risk. <i>Human Molecular Genetics</i> , 2015 , 24, 285-98	5.6	35
105	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	8.1	34
104	DNA glycosylases involved in base excision repair may be associated with cancer risk in BRCA1 and BRCA2 mutation carriers. <i>PLoS Genetics</i> , 2014 , 10, e1004256	6	33
103	11q13 is a susceptibility locus for hormone receptor positive breast cancer. <i>Human Mutation</i> , 2012 , 33, 1123-32	4.7	33
102	A Transcriptome-Wide Association Study Among 97,898 Women to Identify Candidate Susceptibility Genes for Epithelial Ovarian Cancer Risk. <i>Cancer Research</i> , 2018 , 78, 5419-5430	10.1	32
101	PALB2 germline mutations in familial breast cancer cases with personal and family history of pancreatic cancer. <i>Breast Cancer Research and Treatment</i> , 2011 , 126, 825-8	4.4	32
100	Ovarian cancer susceptibility alleles and risk of ovarian cancer in BRCA1 and BRCA2 mutation carriers. <i>Human Mutation</i> , 2012 , 33, 690-702	4.7	31
99	Genetic predisposition to ductal carcinoma in situ of the breast. <i>Breast Cancer Research</i> , 2016 , 18, 22	8.3	31
98	COMPLEXO: identifying the missing heritability of breast cancer via next generation collaboration. <i>Breast Cancer Research</i> , 2013 , 15, 402	8.3	30
97	Genome-wide association study of germline variants and breast cancer-specific mortality. <i>British Journal of Cancer</i> , 2019 , 120, 647-657	8.7	28
96	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. <i>Human Molecular Genetics</i> , 2014 , 23, 1934-46	5.6	28

95	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, 805-7	4.4	28
94	Body mass index and breast cancer survival: a Mendelian randomization analysis. <i>International Journal of Epidemiology</i> , 2017 , 46, 1814-1822	7.8	27
93	Germline mutations in BRIP1 and PALB2 in Jewish high cancer risk families. <i>Familial Cancer</i> , 2012 , 11, 483-91	3	27
92	Confirmation of 5p12 as a susceptibility locus for progesterone-receptor-positive, lower grade breast cancer. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2011 , 20, 2222-31	4	27
91	Polymorphisms in a Putative Enhancer at the 10q21.2 Breast Cancer Risk Locus Regulate NRBF2 Expression. <i>American Journal of Human Genetics</i> , 2015 , 97, 22-34	11	26
90	Reproductive profiles and risk of breast cancer subtypes: a multi-center case-only study. <i>Breast Cancer Research</i> , 2017 , 19, 119	8.3	26
89	Assessing associations between the AURKA-HMMR-TPX2-TUBG1 functional module and breast cancer risk in BRCA1/2 mutation carriers. <i>PLoS ONE</i> , 2015 , 10, e0120020	3.7	26
88	Fine-scale mapping of 8q24 locus identifies multiple independent risk variants for breast cancer. <i>International Journal of Cancer</i> , 2016 , 139, 1303-1317	7.5	26
87	Identification of independent association signals and putative functional variants for breast cancer risk through fine-scale mapping of the 12p11 locus. <i>Breast Cancer Research</i> , 2016 , 18, 64	8.3	25
86	An intergenic risk locus containing an enhancer deletion in 2q35 modulates breast cancer risk by deregulating IGFBP5 expression. <i>Human Molecular Genetics</i> , 2016 , 25, 3863-3876	5.6	24
85	Common germline polymorphisms associated with breast cancer-specific survival. <i>Breast Cancer Research</i> , 2015 , 17, 58	8.3	24
84	Decapping protein EDC4 regulates DNA repair and phenocopies BRCA1. <i>Nature Communications</i> , 2018 , 9, 967	17.4	23
83	Association of Genomic Domains in and with Prostate Cancer Risk and Aggressiveness. <i>Cancer Research</i> , 2020 , 80, 624-638	10.1	22
82	Whole-exome sequencing and targeted gene sequencing provide insights into the role of PALB2 as a male breast cancer susceptibility gene. <i>Cancer</i> , 2017 , 123, 210-218	6.4	22
81	SNPs in ultraconserved elements and familial breast cancer risk. <i>Carcinogenesis</i> , 2009 , 30, 544-5; author reply 546	4.6	22
80	Insight into genetic susceptibility to male breast cancer by multigene panel testing: Results from a multicenter study in Italy. <i>International Journal of Cancer</i> , 2019 , 145, 390-400	7.5	22
79	Height and Body Mass Index as Modifiers of Breast Cancer Risk in BRCA1/2 Mutation Carriers: A Mendelian Randomization Study. <i>Journal of the National Cancer Institute</i> , 2019 , 111, 350-364	9.7	22
78	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, 688-94	8.1	21

77	Common variants of the BRCA1 wild-type allele modify the risk of breast cancer in BRCA1 mutation carriers. <i>Human Molecular Genetics</i> , 2011 , 20, 4732-47	5.6	21
76	Population distribution and ancestry of the cancer protective MDM2 SNP285 (rs117039649). <i>Oncotarget</i> , 2014 , 5, 8223-34	3.3	21
75	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, 2289-95	7.5	20
74	Candidate genetic modifiers for breast and ovarian cancer risk in BRCA1 and BRCA2 mutation carriers. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2015 , 24, 308-16	4	20
73	A network analysis to identify mediators of germline-driven differences in breast cancer prognosis. <i>Nature Communications</i> , 2020 , 11, 312	17.4	20
72	Homologous recombination DNA repair defects in associated breast cancers. <i>Npj Breast Cancer</i> , 2019 , 5, 23	7.8	20
71	A nonsynonymous polymorphism in IRS1 modifies risk of developing breast and ovarian cancers in BRCA1 and ovarian cancer in BRCA2 mutation carriers. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2012 , 21, 1362-70	4	20
70	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, 607-8	4.4	18
69	Meta-analysis of mismatch repair polymorphisms within the cogent consortium for colorectal cancer susceptibility. <i>PLoS ONE</i> , 2013 , 8, e72091	3.7	18
68	RAD51B in Familial Breast Cancer. <i>PLoS ONE</i> , 2016 , 11, e0153788	3.7	18
67	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, 1680-91	4	17
66	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, 1783-91	4	17
65	The BRCA2 c.68-7T>A variant is not pathogenic: A model for clinical calibration of spliceogenicity. <i>Human Mutation</i> , 2018 , 39, 729-741	4.7	16
64	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	4.9	16
63	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	8.3	16
62	Exploring the link between MORF4L1 and risk of breast cancer. <i>Breast Cancer Research</i> , 2011 , 13, R40	8.3	16
61	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, 235-9	4.4	16
60	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, 140-3		16

59	No clinical utility of KRAS variant rs61764370 for ovarian or breast cancer. <i>Gynecologic Oncology</i> , 2016 , 141, 386-401	4.9	15
58	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	4.4	15
57	Haplotype structure in Ashkenazi Jewish BRCA1 and BRCA2 mutation carriers. <i>Human Genetics</i> , 2011 , 130, 685-99	6.3	15
56	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, 679-93	2.8	15
55	The spectrum of BRCA1 and BRCA2 pathogenic sequence variants in Middle Eastern, North African, and South European countries. <i>Human Mutation</i> , 2019 , 40, e1-e23	4.7	14
54	Contribution of Variants to Male Breast Cancer Risk: Results From a Multicenter Study in Italy. <i>Frontiers in Oncology</i> , 2018 , 8, 583	5.3	14
53	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	8.7	13
52	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	3.6	13
51	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, 1130-34	4.7	13
50	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, 703-4	5.8	13
49	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, 256-71	4.6	12
48	The :p.Arg658* truncating variant is associated with risk of triple-negative breast cancer. <i>Npj Breast Cancer</i> , 2019 , 5, 38	7.8	12
47	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	8.3	12
46	BMP2/BMP4 colorectal cancer susceptibility loci in northern and southern European populations. <i>Carcinogenesis</i> , 2013 , 34, 314-8	4.6	12
45	Localization of cancer susceptibility genes by genome-wide single-nucleotide polymorphism linkage-disequilibrium mapping. <i>Cancer Research</i> , 2004 , 64, 8116-25	10.1	12
44	Beneficial effect of fluoxetine in a case of sporadic hyperekplexia. <i>Clinical Neuropharmacology</i> , 2000 , 23, 161-3	1.4	12
43	BRCA1 and BRCA2 5'noncoding region variants identified in breast cancer patients alter promoter activity and protein binding. <i>Human Mutation</i> , 2018 , 39, 2025-2039	4.7	12
42	Genetic variation in mitotic regulatory pathway genes is associated with breast tumor grade. <i>Human Molecular Genetics</i> , 2014 , 23, 6034-46	5.6	11

41	Evidence for a link between TNFRSF11A and risk of breast cancer. <i>Breast Cancer Research and Treatment</i> , 2011 , 129, 947-54	4.4	11
40	Germline mutations of AXIN2 are not associated with nonsyndromic colorectal cancer. <i>Human Mutation</i> , 2005 , 25, 498-500	4.7	11
39	Constitutional de novo deletion of the FBXW7 gene in a patient with focal segmental glomerulosclerosis and multiple primitive tumors. <i>Scientific Reports</i> , 2015 , 5, 15454	4.9	10
38	The CASP8 rs3834129 polymorphism and breast cancer risk in BRCA1 mutation carriers. <i>Breast Cancer Research and Treatment</i> , 2011 , 125, 855-60	4.4	10
37	Transcriptome-wide association study of breast cancer risk by estrogen-receptor status. <i>Genetic Epidemiology</i> , 2020 , 44, 442-468	2.6	9
36	The PALB2 p.Leu939Trp mutation is not associated with breast cancer risk. <i>Breast Cancer Research</i> , 2016 , 18, 111	8.3	9
35	Sequencing analysis of SLX4/FANCP gene in Italian familial breast cancer cases. <i>PLoS ONE</i> , 2012 , 7, e31038	3.7	9
34	Two Missense Variants Detected in Breast Cancer Probands Preventing BRCA2-PALB2 Protein Interaction. <i>Frontiers in Oncology</i> , 2018 , 8, 480	5.3	8
33	The Spectrum of Protein Truncating Variants in European Breast Cancer Cases. <i>Cancers</i> , 2020 , 12,	6.6	7
32	Haplotype analyses of the c.1027C>T and c.2167_2168delAT recurrent truncating mutations in the breast cancer-predisposing gene PALB2. <i>Breast Cancer Research and Treatment</i> , 2016 , 160, 121-129	4.4	7
31	Association between CASP8 -652 6N del polymorphism (rs3834129) and colorectal cancer risk: results from a multi-centric study. <i>PLoS ONE</i> , 2014 , 9, e85538	3.7	7
30	The KL-VS sequence variant of Klotho and cancer risk in BRCA1 and BRCA2 mutation carriers. <i>Breast Cancer Research and Treatment</i> , 2012 , 132, 1119-26	4.4	7
29	Fine-Scale Mapping at 9p22.2 Identifies Candidate Causal Variants That Modify Ovarian Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. <i>PLoS ONE</i> , 2016 , 11, e0158801	3.7	7
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