Paolo Peterlongo

List of Publications by Year in Descending Order

184 papers	12,376 citations	58 h-index	107 g-index
205 ext. papers	15,228 ext. citations	9.7 avg, IF	4.11 L-index

#	Paper	IF	Citations
184	Rare germline copy number variants (CNVs) and breast cancer risk.. *Communications Biology*, **2022**, 5, 65	6.7	0
183	Pathology of Tumors Associated With Pathogenic Germline Variants in 9 Breast Cancer Susceptibility Genes.. *JAMA Oncology*, **2022**,	13.4	4
182	Breast cancer risks associated with missense variants in breast cancer susceptibility genes.. *Genome Medicine*, **2022**, 14, 51	14.4	0
181	Risks of breast and ovarian cancer for women harboring pathogenic missense variants in BRCA1 and BRCA2 compared with those harboring protein truncating variants.. *Genetics in Medicine*, **2021**,	8.1	2
180	Germline variants and breast cancer survival in patients with distant metastases at primary breast cancer diagnosis. *Scientific Reports*, **2021**, 11, 19787	4.9	0
179	Detecting Variants in the NBN Gene While Testing for Hereditary Breast Cancer: What to Do Next?. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	1
178	Pleiotropy-guided transcriptome imputation from normal and tumor tissues identifies candidate susceptibility genes for breast and ovarian cancer. *Human Genetics and Genomics Advances*, **2021**, 2, 100042-100042	0.8	2
177	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. *Genetics in Medicine*, **2021**, 23, 1726-1737	8.1	2
176	Functional annotation of the 2q35 breast cancer risk locus implicates a structural variant in influencing activity of a long-range enhancer element. *American Journal of Human Genetics*, **2021**, 108, 1190-1203	11	1
175	CYP3A7*1C allele: linking premenopausal oestrone and progesterone levels with risk of hormone receptor-positive breast cancers. *British Journal of Cancer*, **2021**, 124, 842-854	8.7	2
174	A case-only study to identify genetic modifiers of breast cancer risk for BRCA1/BRCA2 mutation carriers. *Nature Communications*, **2021**, 12, 1078	17.4	4
173	Breast Cancer Risk Genes - Association Analysis in More than 113,000 Women. *New England Journal of Medicine*, **2021**, 384, 428-439	59.2	143
172	Breast and Prostate Cancer Risks for Male BRCA1 and BRCA2 Pathogenic Variant Carriers Using Polygenic Risk Scores. *Journal of the National Cancer Institute*, **2021**,	9.7	3
171	Association of germline genetic variants with breast cancer-specific survival in patient subgroups defined by clinic-pathological variables related to tumor biology and type of systemic treatment. *Breast Cancer Research*, **2021**, 23, 86	8.3	1
170	Mendelian randomisation study of smoking exposure in relation to breast cancer risk. *British Journal of Cancer*, **2021**, 125, 1135-1145	8.7	0
169	Protein truncating variants in FANCM and risk for ER-negative/triple negative breast cancer. *Npj Breast Cancer*, **2021**, 7, 130	7.8	0
168	Analysis of Italian Pathogenic Variants Identifies a Private Spectrum in the Population from the Bergamo Province in Northern Italy. *Cancers*, **2021**, 13,	6.6	5

167	Genome-wide association study identifies 32 novel breast cancer susceptibility loci from overall and subtype-specific analyses. *Nature Genetics*, **2020**, 52, 572-581	36.3	76
166	Germline HOXB13 mutations p.G84E and p.R217C do not confer an increased breast cancer risk. *Scientific Reports*, **2020**, 10, 9688	4.9	2
165	Transcriptome-wide association study of breast cancer risk by estrogen-receptor status. *Genetic Epidemiology*, **2020**, 44, 442-468	2.6	9
164	A network analysis to identify mediators of germline-driven differences in breast cancer prognosis. *Nature Communications*, **2020**, 11, 312	17.4	20
163	The Spectrum of Protein Truncating Variants in European Breast Cancer Cases. *Cancers*, **2020**, 12,	6.6	7
162	Association of Genomic Domains in and with Prostate Cancer Risk and Aggressiveness. *Cancer Research*, **2020**, 80, 624-638	10.1	22
161	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. *Nature Genetics*, **2020**, 52, 56-73	36.3	56
160	Cancer Risks Associated With Germline Pathogenic Variants: An International Study of 524 Families. *Journal of Clinical Oncology*, **2020**, 38, 674-685	2.2	133
159	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	8.1	34
158	Mutation Rates in Cancer Susceptibility Genes in Patients With Breast Cancer With Multiple Primary Cancers. *JCO Precision Oncology*, **2020**, 4,	3.6	2
157	Two truncating variants in FANCC and breast cancer risk. *Scientific Reports*, **2019**, 9, 12524	4.9	2
156	The spectrum of BRCA1 and BRCA2 pathogenic sequence variants in Middle Eastern, North African, and South European countries. *Human Mutation*, **2019**, 40, e1-e23	4.7	14
155	Mendelian randomisation study of height and body mass index as modifiers of ovarian cancer risk in 22,588 BRCA1 and BRCA2 mutation carriers. *British Journal of Cancer*, **2019**, 121, 180-192	8.7	13
154	Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. *Nature Communications*, **2019**, 10, 1741	17.4	47
153	Genome-wide association study of germline variants and breast cancer-specific mortality. *British Journal of Cancer*, **2019**, 120, 647-657	8.7	28
152	Homologous recombination DNA repair defects in associated breast cancers. *Npj Breast Cancer*, **2019**, 5, 23	7.8	20
151	The :p.Arg658* truncating variant is associated with risk of triple-negative breast cancer. *Npj Breast Cancer*, **2019**, 5, 38	7.8	12
150	Evaluation of CYP17A1 and CYP1B1 polymorphisms in male breast cancer risk. *Endocrine Connections*, **2019**, 8, 1224-1229	3.5	2

149	Polygenic Risk Scores for Prediction of Breast Cancer and Breast Cancer Subtypes. *American Journal of Human Genetics*, **2019**, 104, 21-34	11	363
148	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	7.5	22
147	Height and Body Mass Index as Modifiers of Breast Cancer Risk in BRCA1/2 Mutation Carriers: A Mendelian Randomization Study. *Journal of the National Cancer Institute*, **2019**, 111, 350-364	9.7	22
146	The BRCA2 c.68-7T□□A variant is not pathogenic: A model for clinical calibration of spliceogenicity. *Human Mutation*, **2018**, 39, 729-741	4.7	16
145	Decapping protein EDC4 regulates DNA repair and phenocopies BRCA1. *Nature Communications*, **2018**, 9, 967	17.4	23
144	Mutational spectrum in a worldwide study of 29,700 families with BRCA1 or BRCA2 mutations. *Human Mutation*, **2018**, 39, 593-620	4.7	138
143	A possible role of FANCM mutations in male breast cancer susceptibility: Results from a multicenter study in Italy. *Breast*, **2018**, 38, 92-97	3.6	13
142	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	8.1	44
141	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	10.1	32
140	A transcriptome-wide association study of 229,000 women identifies new candidate susceptibility genes for breast cancer. *Nature Genetics*, **2018**, 50, 968-978	36.3	101
139	Contribution of Variants to Male Breast Cancer Risk: Results From a Multicenter Study in Italy. *Frontiers in Oncology*, **2018**, 8, 583	5.3	14
138	Two Missense Variants Detected in Breast Cancer Probands Preventing BRCA2-PALB2 Protein Interaction. *Frontiers in Oncology*, **2018**, 8, 480	5.3	8
137	BRCA1 and BRCA2 5Pnoncoding region variants identified in breast cancer patients alter promoter activity and protein binding. *Human Mutation*, **2018**, 39, 2025-2039	4.7	12
136	Hypomorphic Missense Variants Confer Moderate Risks of Breast Cancer. *Cancer Research*, **2017**, 77, 2789-2799	10.1	49
135	The contribution of pathogenic variants in breast cancer susceptibility genes to familial breast cancer risk. *Npj Breast Cancer*, **2017**, 3, 22	7.8	78
134	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. *Nature Genetics*, **2017**, 49, 680-691	36.3	190
133	Association analysis identifies 65 new breast cancer risk loci. *Nature*, **2017**, 551, 92-94	50.4	643
132	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. *Nature Genetics*, **2017**, 49, 1767-1778	36.3	186

131	Reproductive profiles and risk of breast cancer subtypes: a multi-center case-only study. *Breast Cancer Research*, **2017**, 19, 119	8.3	26
130	Prediction of Breast and Prostate Cancer Risks in Male BRCA1 and BRCA2 Mutation Carriers Using Polygenic Risk Scores. *Journal of Clinical Oncology*, **2017**, 35, 2240-2250	2.2	101
129	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. *Breast Cancer Research and Treatment*, **2017**, 161, 117-134	4.4	15
128	Whole-exome sequencing and targeted gene sequencing provide insights into the role of PALB2 as a male breast cancer susceptibility gene. *Cancer*, **2017**, 123, 210-218	6.4	22
127	Evaluation of Polygenic Risk Scores for Breast and Ovarian Cancer Risk Prediction in BRCA1 and BRCA2 Mutation Carriers. *Journal of the National Cancer Institute*, **2017**, 109,	9.7	153
126	Body mass index and breast cancer survival: a Mendelian randomization analysis. *International Journal of Epidemiology*, **2017**, 46, 1814-1822	7.8	27
125	No clinical utility of KRAS variant rs61764370 for ovarian or breast cancer. *Gynecologic Oncology*, **2016**, 141, 386-401	4.9	15
124	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	5.6	24
123	Haplotype analyses of the c.1027C>T and c.2167_2168delAT recurrent truncating mutations in the breast cancer-predisposing gene PALB2. *Breast Cancer Research and Treatment*, **2016**, 160, 121-129	4.4	7
122	Genome-Wide Meta-Analyses of Breast, Ovarian, and Prostate Cancer Association Studies Identify Multiple New Susceptibility Loci Shared by at Least Two Cancer Types. *Cancer Discovery*, **2016**, 6, 1052-67	24.4	104
121	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. *Nature Communications*, **2016**, 7, 11375	17.4	64
120	Functional mechanisms underlying pleiotropic risk alleles at the 19p13.1 breast-ovarian cancer susceptibility locus. *Nature Communications*, **2016**, 7, 12675	17.4	53
119	Fine scale mapping of the 17q22 breast cancer locus using dense SNPs, genotyped within the Collaborative Oncological Gene-Environment Study (COGs). *Scientific Reports*, **2016**, 6, 32512	4.9	16
118	The PALB2 p.Leu939Trp mutation is not associated with breast cancer risk. *Breast Cancer Research*, **2016**, 18, 111	8.3	9
117	No evidence that protein truncating variants in BRIP1 are associated with breast cancer risk: implications for gene panel testing. *Journal of Medical Genetics*, **2016**, 53, 298-309	5.8	83
116	Breast cancer risk variants at 6q25 display different phenotype associations and regulate ESR1, RMND1 and CCDC170. *Nature Genetics*, **2016**, 48, 374-86	36.3	93
115	Genetic variation in the immunosuppression pathway genes and breast cancer susceptibility: a pooled analysis of 42,510 cases and 40,577 controls from the Breast Cancer Association Consortium. *Human Genetics*, **2016**, 135, 137-54	6.3	6
114	BRCA2 Polymorphic Stop Codon K3326X and the Risk of Breast, Prostate, and Ovarian Cancers. *Journal of the National Cancer Institute*, **2016**, 108,	9.7	65

113	RAD51B in Familial Breast Cancer. *PLoS ONE*, **2016**, 11, e0153788	3.7	18
112	Genetically Predicted Body Mass Index and Breast Cancer Risk: Mendelian Randomization Analyses of Data from 145,000 Women of European Descent. *PLoS Medicine*, **2016**, 13, e1002105	11.6	80
111	Fine-Scale Mapping at 9p22.2 Identifies Candidate Causal Variants That Modify Ovarian Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. *PLoS ONE*, **2016**, 11, e0158801	3.7	7
110	Fine-scale mapping of 8q24 locus identifies multiple independent risk variants for breast cancer. *International Journal of Cancer*, **2016**, 139, 1303-1317	7.5	26
109	PALB2, CHEK2 and ATM rare variants and cancer risk: data from COGS. *Journal of Medical Genetics*, **2016**, 53, 800-811	5.8	121
108	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	8.3	25
107	Genetic predisposition to ductal carcinoma in situ of the breast. *Breast Cancer Research*, **2016**, 18, 22	8.3	31
106	Association of genetic susceptibility variants for type 2 diabetes with breast cancer risk in women of European ancestry. *Cancer Causes and Control*, **2016**, 27, 679-93	2.8	15
105	Male breast cancer in BRCA1 and BRCA2 mutation carriers: pathology data from the Consortium of Investigators of Modifiers of BRCA1/2. *Breast Cancer Research*, **2016**, 18, 15	8.3	58
104	Inherited variants in the inner centromere protein (INCENP) gene of the chromosomal passenger complex contribute to the susceptibility of ER-negative breast cancer. *Carcinogenesis*, **2015**, 36, 256-71	4.6	12
103	Identification of six new susceptibility loci for invasive epithelial ovarian cancer. *Nature Genetics*, **2015**, 47, 164-71	36.3	177
102	Genome-wide association analysis of more than 120,000 individuals identifies 15 new susceptibility loci for breast cancer. *Nature Genetics*, **2015**, 47, 373-80	36.3	406
101	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. *European Journal of Cancer*, **2015**, 51, 2289-95	7.5	20
100	FANCM c.5791C>T nonsense mutation (rs144567652) induces exon skipping, affects DNA repair activity and is a familial breast cancer risk factor. *Human Molecular Genetics*, **2015**, 24, 5345-55	5.6	68
99	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	11	26
98	Identification of novel genetic markers of breast cancer survival. *Journal of the National Cancer Institute*, **2015**, 107,	9.7	38
97	Large-scale genomic analyses link reproductive aging to hypothalamic signaling, breast cancer susceptibility and BRCA1-mediated DNA repair. *Nature Genetics*, **2015**, 47, 1294-1303	36.3	226
96	Height and Breast Cancer Risk: Evidence From Prospective Studies and Mendelian Randomization. *Journal of the National Cancer Institute*, **2015**, 107,	9.7	74

95	Fine-scale mapping of the 4q24 locus identifies two independent loci associated with breast cancer risk. *Cancer Epidemiology Biomarkers and Prevention*, **2015**, 24, 1680-91	4	17
94	Identification and characterization of novel associations in the CASP8/ALS2CR12 region on chromosome 2 with breast cancer risk. *Human Molecular Genetics*, **2015**, 24, 285-98	5.6	35
93	Candidate genetic modifiers for breast and ovarian cancer risk in BRCA1 and BRCA2 mutation carriers. *Cancer Epidemiology Biomarkers and Prevention*, **2015**, 24, 308-16	4	20
92	Constitutional de novo deletion of the FBXW7 gene in a patient with focal segmental glomerulosclerosis and multiple primitive tumors. *Scientific Reports*, **2015**, 5, 15454	4.9	10
91	An original phylogenetic approach identified mitochondrial haplogroup T1a1 as inversely associated with breast cancer risk in BRCA2 mutation carriers. *Breast Cancer Research*, **2015**, 17, 61	8.3	16
90	Common germline polymorphisms associated with breast cancer-specific survival. *Breast Cancer Research*, **2015**, 17, 58	8.3	24
89	Large-Scale Genomic Analyses Link Reproductive Aging to Hypothalamic Signaling, Breast Cancer Susceptibility, and BRCA1-Mediated DNA Repair. *Obstetrical and Gynecological Survey*, **2015**, 70, 758-762	2.4	
88	Assessing associations between the AURKA-HMMR-TPX2-TUBG1 functional module and breast cancer risk in BRCA1/2 mutation carriers. *PLoS ONE*, **2015**, 10, e0120020	3.7	26
87	Prediction of breast cancer risk based on profiling with common genetic variants. *Journal of the National Cancer Institute*, **2015**, 107,	9.7	324
86	Fine-mapping identifies two additional breast cancer susceptibility loci at 9q31.2. *Human Molecular Genetics*, **2015**, 24, 2966-84	5.6	36
85	Fine-scale mapping of the 5q11.2 breast cancer locus reveals at least three independent risk variants regulating MAP3K1. *American Journal of Human Genetics*, **2015**, 96, 5-20	11	59
84	PALB2 sequencing in Italian familial breast cancer cases reveals a high-risk mutation recurrent in the province of Bergamo. *Genetics in Medicine*, **2014**, 16, 688-94	8.1	21
83	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, 1934-46	5.6	28
82	Parent-of-origin-specific allelic associations among 106 genomic loci for age at menarche. *Nature*, **2014**, 514, 92-97	50.4	401
81	Breast-cancer risk in families with mutations in PALB2. *New England Journal of Medicine*, **2014**, 371, 497-506	59.2	576
80	Evidence that breast cancer risk at the 2q35 locus is mediated through IGFBP5 regulation. *Nature Communications*, **2014**, 4, 4999	17.4	87
79	Genetic variation in mitotic regulatory pathway genes is associated with breast tumor grade. *Human Molecular Genetics*, **2014**, 23, 6034-46	5.6	11
78	Genetic variation at CYP3A is associated with age at menarche and breast cancer risk: a case-control study. *Breast Cancer Research*, **2014**, 16, R51	8.3	12

77	Association between CASP8 -652 6N del polymorphism (rs3834129) and colorectal cancer risk: results from a multi-centric study. *PLoS ONE*, **2014**, 9, e85538	3.7	7
76	MicroRNA related polymorphisms and breast cancer risk. *PLoS ONE*, **2014**, 9, e109973	3.7	37
75	Genetic predisposition to in situ and invasive lobular carcinoma of the breast. *PLoS Genetics*, **2014**, 10, e1004285	6	38
74	DNA glycosylases involved in base excision repair may be associated with cancer risk in BRCA1 and BRCA2 mutation carriers. *PLoS Genetics*, **2014**, 10, e1004256	6	33
73	Common non-synonymous SNPs associated with breast cancer susceptibility: findings from the Breast Cancer Association Consortium. *Human Molecular Genetics*, **2014**, 23, 6096-111	5.6	48
72	Refined histopathological predictors of BRCA1 and BRCA2 mutation status: a large-scale analysis of breast cancer characteristics from the BCAC, CIMBA, and ENIGMA consortia. *Breast Cancer Research* , **2014**, 16, 3419	8.3	82
71	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	8.3	46
70	The MLH1 c.1852_1853delinsGC (p.K618A) variant in colorectal cancer: genetic association study in 18,723 individuals. *PLoS ONE*, **2014**, 9, e95022	3.7	6
69	Population distribution and ancestry of the cancer protective MDM2 SNP285 (rs117039649). *Oncotarget*, **2014**, 5, 8223-34	3.3	21
68	COMPLEXO: identifying the missing heritability of breast cancer via next generation collaboration. *Breast Cancer Research*, **2013**, 15, 402	8.3	30
67	Fine-scale mapping of the FGFR2 breast cancer risk locus: putative functional variants differentially bind FOXA1 and E2F1. *American Journal of Human Genetics*, **2013**, 93, 1046-60	11	80
66	Multiple independent variants at the TERT locus are associated with telomere length and risks of breast and ovarian cancer. *Nature Genetics*, **2013**, 45, 371-84, 384e1-2	36.3	422
65	Functional variants at the 11q13 risk locus for breast cancer regulate cyclin D1 expression through long-range enhancers. *American Journal of Human Genetics*, **2013**, 92, 489-503	11	167
64	Genome-wide association studies identify four ER negative-specific breast cancer risk loci. *Nature Genetics*, **2013**, 45, 392-8, 398e1-2	36.3	327
63	Large-scale genotyping identifies 41 new loci associated with breast cancer risk. *Nature Genetics*, **2013**, 45, 353-61, 361e1-2	36.3	813
62	Identification of a BRCA2-specific modifier locus at 6p24 related to breast cancer risk. *PLoS Genetics*, **2013**, 9, e1003173	6	90
61	Genome-wide association study in BRCA1 mutation carriers identifies novel loci associated with breast and ovarian cancer risk. *PLoS Genetics*, **2013**, 9, e1003212	6	209
60	BMP2/BMP4 colorectal cancer susceptibility loci in northern and southern European populations. *Carcinogenesis*, **2013**, 34, 314-8	4.6	12

59	Whole exome sequencing suggests much of non-BRCA1/BRCA2 familial breast cancer is due to moderate and low penetrance susceptibility alleles. *PLoS ONE*, **2013**, 8, e55681	3.7	77
58	Meta-analysis of mismatch repair polymorphisms within the cogent consortium for colorectal cancer susceptibility. *PLoS ONE*, **2013**, 8, e72091	3.7	18
57	Common variants at 12p11, 12q24, 9p21, 9q31.2 and in ZNF365 are associated with breast cancer risk for BRCA1 and/or BRCA2 mutation carriers. *Breast Cancer Research*, **2012**, 14, R33	8.3	70
56	Ovarian cancer susceptibility alleles and risk of ovarian cancer in BRCA1 and BRCA2 mutation carriers. *Human Mutation*, **2012**, 33, 690-702	4.7	31
55	Genome-wide association analysis identifies three new breast cancer susceptibility loci. *Nature Genetics*, **2012**, 44, 312-8	36.3	237
54	Rare variants in XRCC2 as breast cancer susceptibility alleles. *Journal of Medical Genetics*, **2012**, 49, 618-23	5.8	37
53	9q31.2-rs865686 as a susceptibility locus for estrogen receptor-positive breast cancer: evidence from the Breast Cancer Association Consortium. *Cancer Epidemiology Biomarkers and Prevention*, **2012**, 21, 1783-91	4	17
52	Germline mutations in BRIP1 and PALB2 in Jewish high cancer risk families. *Familial Cancer*, **2012**, 11, 483-91	3	27
51	Sequencing analysis of SLX4/FANCP gene in Italian familial breast cancer cases. *PLoS ONE*, **2012**, 7, e31038	3.7	9
50	11q13 is a susceptibility locus for hormone receptor positive breast cancer. *Human Mutation*, **2012**, 33, 1123-32	4.7	33
49	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, 1665-75	4.7	42
48	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, 411-8	4.4	58
47	The KL-VS sequence variant of Klotho and cancer risk in BRCA1 and BRCA2 mutation carriers. *Breast Cancer Research and Treatment*, **2012**, 132, 1119-26	4.4	7
46	Prospective study of breast tomosynthesis as a triage to assessment in screening. *Breast Cancer Research and Treatment*, **2012**, 133, 267-71	4.4	66
45	The SNP rs895819 in miR-27a is not associated with familial breast cancer risk in Italians. *Breast Cancer Research and Treatment*, **2012**, 133, 805-7	4.4	28
44	A nonsynonymous polymorphism in IRS1 modifies risk of developing breast and ovarian cancers in BRCA1 and ovarian cancer in BRCA2 mutation carriers. *Cancer Epidemiology Biomarkers and Prevention*, **2012**, 21, 1362-70	4	20
43	19p13.1 is a triple-negative-specific breast cancer susceptibility locus. *Cancer Research*, **2012**, 72, 1795-803	13.1	93
42	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, 645-57	4	44

41	The role of genetic breast cancer susceptibility variants as prognostic factors. *Human Molecular Genetics*, **2012**, 21, 3926-39	5.6	75
40	Comparison of 6q25 breast cancer hits from Asian and European Genome Wide Association Studies in the Breast Cancer Association Consortium (BCAC). *PLoS ONE*, **2012**, 7, e42380	3.7	49
39	Exploring the link between MORF4L1 and risk of breast cancer. *Breast Cancer Research*, **2011**, 13, R40	8.3	16
38	Common breast cancer susceptibility alleles are associated with tumour subtypes in BRCA1 and BRCA2 mutation carriers: results from the Consortium of Investigators of Modifiers of BRCA1/2. *Breast Cancer Research*, **2011**, 13, R110	8.3	62
37	The CASP8 rs3834129 polymorphism and breast cancer risk in BRCA1 mutation carriers. *Breast Cancer Research and Treatment*, **2011**, 125, 855-60	4.4	10
36	No evidence for an association between the earwax-associated polymorphism in ABCC11 and breast cancer risk in Caucasian women. *Breast Cancer Research and Treatment*, **2011**, 126, 235-9	4.4	16
35	PALB2 germline mutations in familial breast cancer cases with personal and family history of pancreatic cancer. *Breast Cancer Research and Treatment*, **2011**, 126, 825-8	4.4	32
34	Evidence for a link between TNFRSF11A and risk of breast cancer. *Breast Cancer Research and Treatment*, **2011**, 129, 947-54	4.4	11
33	Haplotype structure in Ashkenazi Jewish BRCA1 and BRCA2 mutation carriers. *Human Genetics*, **2011**, 130, 685-99	6.3	15
32	Confirmation of 5p12 as a susceptibility locus for progesterone-receptor-positive, lower grade breast cancer. *Cancer Epidemiology Biomarkers and Prevention*, **2011**, 20, 2222-31	4	27
31	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, 3304-21	5.6	62
30	Associations of common variants at 1p11.2 and 14q24.1 (RAD51L1) with breast cancer risk and heterogeneity by tumor subtype: findings from the Breast Cancer Association Consortium. *Human Molecular Genetics*, **2011**, 20, 4693-706	5.6	66
29	The rs12975333 variant in the miR-125a and breast cancer risk in Germany, Italy, Australia and Spain. *Journal of Medical Genetics*, **2011**, 48, 703-4	5.8	13
28	Common variants of the BRCA1 wild-type allele modify the risk of breast cancer in BRCA1 mutation carriers. *Human Molecular Genetics*, **2011**, 20, 4732-47	5.6	21
27	Genetic variation at 9p22.2 and ovarian cancer risk for BRCA1 and BRCA2 mutation carriers. *Journal of the National Cancer Institute*, **2011**, 103, 105-16	9.7	37
26	Interplay between BRCA1 and RHAMM regulates epithelial apicobasal polarization and may influence risk of breast cancer. *PLoS Biology*, **2011**, 9, e1001199	9.7	73
25	A locus on 19p13 modifies risk of breast cancer in BRCA1 mutation carriers and is associated with hormone receptor-negative breast cancer in the general population. *Nature Genetics*, **2010**, 42, 885-92	36.3	276
24	Common breast cancer susceptibility alleles and the risk of breast cancer for BRCA1 and BRCA2 mutation carriers: implications for risk prediction. *Cancer Research*, **2010**, 70, 9742-54	10.1	147

23	A BRCA1 promoter variant (rs11655505) and breast cancer risk. *Journal of Medical Genetics*, **2010**, 47, 268-70	5.8	5
22	Common genetic variants and modification of penetrance of BRCA2-associated breast cancer. *PLoS Genetics*, **2010**, 6, e1001183	6	74
21	Evaluation of SNPs in miR-146a, miR196a2 and miR-499 as low-penetrance alleles in German and Italian familial breast cancer cases. *Human Mutation*, **2010**, 31, E1052-7	4.7	138
20	Common variants in LSP1, 2q35 and 8q24 and breast cancer risk for BRCA1 and BRCA2 mutation carriers. *Human Molecular Genetics*, **2009**, 18, 4442-56	5.6	91
19	SNPs in ultraconserved elements and familial breast cancer risk. *Carcinogenesis*, **2009**, 30, 544-5; author reply 546	4.6	22
18	Evidences for association of the CASP8 -652 6N del promoter polymorphism with age at diagnosis in familial breast cancer cases. *Breast Cancer Research and Treatment*, **2009**, 113, 607-8	4.4	18
17	Toll-like receptor 4 polymorphisms and risk of gram-negative bacteremia after allogeneic stem cell transplantation. A prospective pilot study. *Biology of Blood and Marrow Transplantation*, **2009**, 15, 1130-3	4.7	13
16	The p53 Arg72Pro and Ins16bp polymorphisms and their haplotypes are not associated with breast cancer risk in BRCA-mutation negative familial cases. *Cancer Detection and Prevention*, **2008**, 32, 140-3		16
15	Common breast cancer-predisposition alleles are associated with breast cancer risk in BRCA1 and BRCA2 mutation carriers. *American Journal of Human Genetics*, **2008**, 82, 937-48	11	218
14	Increased frequency of disease-causing MYH mutations in colon cancer families. *Carcinogenesis*, **2006**, 27, 2243-9	4.6	39
13	TLR1 and TLR6 polymorphisms are associated with susceptibility to invasive aspergillosis after allogeneic stem cell transplantation. *Annals of the New York Academy of Sciences*, **2005**, 1062, 95-103	6.5	169
12	Germline mutations of AXIN2 are not associated with nonsyndromic colorectal cancer. *Human Mutation*, **2005**, 25, 498-500	4.7	11
11	Colorectal cancer risk in individuals with biallelic or monoallelic mutations of MYH. *International Journal of Cancer*, **2005**, 114, 505-7	7.5	52
10	TGFBR1*6A may contribute to hereditary colorectal cancer. *Journal of Clinical Oncology*, **2005**, 23, 3074-8	2.2	41
9	TGFBR1*6A and cancer: a meta-analysis of 12 case-control studies. *Journal of Clinical Oncology*, **2004**, 22, 756-8	2.2	75
8	Localization of cancer susceptibility genes by genome-wide single-nucleotide polymorphism linkage-disequilibrium mapping. *Cancer Research*, **2004**, 64, 8116-25	10.1	12
7	MSH6 germline mutations are rare in colorectal cancer families. *International Journal of Cancer*, **2003**, 107, 571-9	7.5	49
6	Beneficial effect of fluoxetine in a case of sporadic hyperekplexia. *Clinical Neuropharmacology*, **2000**, 23, 161-3	1.4	12

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4	Polygenic Risk Modelling for Prediction of Epithelial Ovarian Cancer Risk		1
3	Combining genome-wide studies of breast, prostate, ovarian and endometrial cancers maps cross-cancer susceptibility loci and identifies new genetic associations		2
2	Fine-mapping of 150 breast cancer risk regions identifies 178 high confidence target genes		2
1	Genome-wide association study identifies 32 novel breast cancer susceptibility loci from overall and subtype-specific analyses		2