Paolo Radice

List of Publications by Year
in descending order

306 papers	28,308 citations	8181 76 h-index	6836 155 g-index
323 all docs	323 docs citations	323 times ranked	26239 citing authors

#	Article	IF	Citations
1	Average Risks of Breast and Ovarian Cancer Associated with BRCA1 or BRCA2 Mutations Detected in Case Series Unselected for Family History: A Combined Analysis of 22 Studies. American Journal of Human Genetics, 2003, 72, 1117-1130.	6.2	3,105
2	Association analysis identifies 65 new breast cancer risk loci. Nature, 2017, 551, 92-94.	27.8	1,099
3	Large-scale genotyping identifies 41 new loci associated with breast cancer risk. Nature Genetics, 2013, 45, 353-361.	21.4	960
4	Breast-Cancer Risk in Families with Mutations in PALB2. New England Journal of Medicine, 2014, 371, 497-506.	27.0	745
5	Polygenic Risk Scores for Prediction of Breast Cancer and Breast Cancer Subtypes. American Journal of Human Genetics, 2019, 104, 21-34.	6.2	711
6	Mutations predisposing to hereditary nonpolyposis colorectal cancer: Database and results of a collaborative study. The International Collaborative Group on Hereditary Nonpolyposis Colorectal Cancer. Gastroenterology, 1997, 113, 1146-1158.	1.3	682
7	Associations of Breast Cancer Risk Factors With Tumor Subtypes: A Pooled Analysis From the Breast Cancer Association Consortium Studies. Journal of the National Cancer Institute, 2011, 103, 250-263.	6.3	596
8	Oral Contraceptives and the Risk of Hereditary Ovarian Cancer. New England Journal of Medicine, 1998, 339, 424-428.	27.0	591
9	Prediction of BRCA1 Status in Patients with Breast Cancer Using Estrogen Receptor and Basal Phenotype. Clinical Cancer Research, 2005, 11, 5175-5180.	7.0	577
10	Parent-of-origin-specific allelic associations among 106 genomic loci for age at menarche. Nature, 2014, 514, 92-97.	27.8	548
11	Breast Cancer Risk Genes â€" Association Analysis in More than 113,000 Women. New England Journal of Medicine, 2021, 384, 428-439.	27.0	532
12	Pathology of Breast and Ovarian Cancers among BRCA1 and BRCA2 Mutation Carriers: Results from the Consortium of Investigators of Modifiers of BRCA1/2 (CIMBA). Cancer Epidemiology Biomarkers and Prevention, 2012, 21, 134-147.	2.5	513
13	Genome-wide association analysis of more than 120,000 individuals identifies 15 new susceptibility loci for breast cancer. Nature Genetics, 2015, 47, 373-380.	21.4	513
14	Multiple independent variants at the TERT locus are associated with telomere length and risks of breast and ovarian cancer. Nature Genetics, 2013, 45, 371-384.	21.4	493
15	Prediction of Breast Cancer Risk Based on Profiling With Common Genetic Variants. Journal of the National Cancer Institute, 2015, 107, .	6.3	428
16	Genomic analyses identify hundreds of variants associated with age at menarche and support a role for puberty timing in cancer risk. Nature Genetics, 2017, 49, 834-841.	21.4	426
17	Association of Type and Location ofBRCA1andBRCA2Mutations With Risk of Breast and Ovarian Cancer. JAMA - Journal of the American Medical Association, 2015, 313, 1347.	7.4	390
18	Genome-wide association studies identify four ER negativeâ€"specific breast cancer risk loci. Nature Genetics, 2013, 45, 392-398.	21.4	374

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19	Single-Nucleotide Polymorphisms Inside MicroRNA Target Sites Influence Tumor Susceptibility. Cancer Research, 2010, 70, 2789-2798.	0.9	365
20	Large-scale genomic analyses link reproductive aging to hypothalamic signaling, breast cancer susceptibility and BRCA1-mediated DNA repair. Nature Genetics, 2015, 47, 1294-1303.	21.4	357
21	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. Nature Genetics, 2017, 49, 680-691.	21.4	356
22	A locus on 19p13 modifies risk of breast cancer in BRCA1 mutation carriers and is associated with hormone receptorâ€“negative breast cancer in the general population. Nature Genetics, 2010, 42, 885-892.	21.4	309
23	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. Nature Genetics, 2017, 49, 1767-1778.	21.4	289
24	Cancer Risks Associated With Germline PALB2 Pathogenic Variants: An International Study of 524 Families. Journal of Clinical Oncology, 2020, 38, 674-685.	1.6	270
25	ENIGMA-Evidence-based network for the interpretation of germline mutant alleles: An international initiative to evaluate risk and clinical significance associated with sequence variation in BRCA1 and BRCA2 genes. Human Mutation, 2012, 33, 2-7.	2.5	269
26	Genome-wide association study identifies 32 novel breast cancer susceptibility loci from overall and subtype-specific analyses. Nature Genetics, 2020, 52, 572-581.	21.4	265
27	Common Breast Cancer-Predisposition Alleles Are Associated with Breast Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. American Journal of Human Genetics, 2008, 82, 937-948.	6.2	257
28	Genome-Wide Association Study in BRCA1 Mutation Carriers Identifies Novel Loci Associated with Breast and Ovarian Cancer Risk. PLoS Genetics, 2013, 9, e1003212.	3.5	244
29	Pathology of Ovarian Cancers in BRCA1 and BRCA2 Carriers. Clinical Cancer Research, 2004, 10, 2473-2481.	7.0	224
30	Mutational spectrum in a worldwide study of 29,700 families withBRCA1orBRCA2mutations. Human Mutation, 2018, 39, 593-620.	2.5	224
31	Identification of six new susceptibility loci for invasive epithelial ovarian cancer. Nature Genetics, 2015, 47, 164-171.	21.4	221
32	Evidence of a founder mutation of BRCA1 in a highly homogeneous population from southern Italy with breast/ovarian cancer. Human Mutation, 2001, 18, 163-164.	2.5	215
33	Genotype and phenotype factors as determinants of desmoid tumors in patients with familial adenomatous polyposis. International Journal of Cancer, 2001, 95, 102-107.	5.1	206
34	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.	6.2	201
35	Different Genetic Features Associated with Colon and Rectal Carcinogenesis. Clinical Cancer Research, 2004, 10, 4015-4021.	7.0	191
36	Multiple Approach to the Exploration of Genotype-Phenotype Correlations in Familial Adenomatous Polyposis. Journal of Clinical Oncology, 2003, 21, 1698-1707.	1.6	184

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37	A transcriptome-wide association study of 229,000 women identifies new candidate susceptibility genes for breast cancer. Nature Genetics, 2018, 50, 968-978.	21.4	184
38	PALB2,CHEK2andATMrare variants and cancer risk: data from COGS. Journal of Medical Genetics, 2016, 53, 800-811.	3.2	174
39	A novel zinc finger gene is fused to EWS in small round cell tumor. Oncogene, 2000, 19, 3799-3804.	5.9	173
40	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, 9742-9754.	0.9	169
41	Prevalence of the Y165C, G382D and 1395delGGA germline mutations of the MYH gene in Italian patients with adenomatous polyposis coli and colorectal adenomas. International Journal of Cancer, 2004, 109, 680-684.	5.1	159
42	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.	1.6	152
43	Evaluation of SNPs inmiR-146a,miR196a2andmiR-499as low-penetrance alleles in German and Italian familial breast cancer cases. Human Mutation, 2010, 31, E1052-E1057.	2.5	147
44	Functional analysis of human MLH1 mutations in Saccharomyces cerevisiae. Nature Genetics, 1998, 19, 384-389.	21.4	136
45	Atypical Epithelial Proliferation in Fallopian Tubes in Prophylactic Salpingo-oophorectomy Specimens from BRCA1 and BRCA2 Germline Mutation Carriers. International Journal of Gynecological Pathology, 2004, 23, 35-40.	1.4	135
46	Incidental Carcinomas in Prophylactic Specimens in BRCA1 and BRCA2 Germ-line Mutation Carriers, With Emphasis on Fallopian Tube Lesions. American Journal of Surgical Pathology, 2006, 30, 1222-1230.	3.7	130
47	Reproductive and Hormonal Factors, and Ovarian Cancer Risk for BRCA1 and BRCA2 Mutation Carriers: Results from the International BRCA1/2 Carrier Cohort Study. Cancer Epidemiology Biomarkers and Prevention, 2009, 18, 601-610.	2.5	130
48	Breast cancer risk variants at 6q25 display different phenotype associations and regulate ESR1, RMND1 and CCDC170. Nature Genetics, 2016, 48, 374-386.	21.4	125
49	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. Nature Genetics, 2020, 52, 56-73.	21.4	120
50	Determination of Cancer Risk Associated with Germ Line BRCA1 Missense Variants by Functional Analysis. Cancer Research, 2007, 67, 1494-1501.	0.9	110
51	Prediction and assessment of splicing alterations: implications for clinical testing. Human Mutation, 2008, 29, 1304-1313.	2.5	108
52	Combined genetic and splicing analysis of BRCA1 c.[594-2A>C; 641A>G] highlights the relevance of naturally occurring in-frame transcripts for developing disease gene variant classification algorithms. Human Molecular Genetics, 2016, 25, 2256-2268.	2.9	106
53	Identification of a BRCA2-Specific Modifier Locus at 6p24 Related to Breast Cancer Risk. PLoS Genetics, 2013, 9, e1003173.	3.5	105
54	Evidence that breast cancer risk at the 2q35 locus is mediated through IGFBP5 regulation. Nature Communications, 2014, 5, 4999.	12.8	105

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55	A Classification Model for BRCA2 DNA Binding Domain Missense Variants Based on Homology-Directed Repair Activity. Cancer Research, 2013, 73, 265-275.	0.9	103
56	Large scale multifactorial likelihood quantitative analysis of BRCA1 and BRCA2 variants: An ENIGMA resource to support clinical variant classification. Human Mutation, 2019, 40, 1557-1578.	2.5	102
57	The Exon 13 Duplication in the BRCA1 Gene Is a Founder Mutation Present in Geographically Diverse Populations. American Journal of Human Genetics, 2000, 67, 207-212.	6.2	100
58	19p13.1 Is a Triple-Negativeâ€“Specific Breast Cancer Susceptibility Locus. Cancer Research, 2012, 72, 1795-1803.	0.9	100
59	Common variants in LSP1, 2q35 and 8q24 and breast cancer risk for BRCA1 and BRCA2 mutation carriers. Human Molecular Genetics, 2009, 18, 4442-4456.	2.9	99
60	Height and Breast Cancer Risk: Evidence From Prospective Studies and Mendelian Randomization. Journal of the National Cancer Institute, 2015, 107, djv219.	6.3	99
61	Fine-Scale Mapping of the FGFR2 Breast Cancer Risk Locus: Putative Functional Variants Differentially Bind FOXA1 and E2F1. American Journal of Human Genetics, 2013, 93, 1046-1060.	6.2	98
62	Refined histopathological predictors of BRCA1 and BRCA2mutation status: a large-scale analysis of breast cancer characteristics from the BCAC, CIMBA, and ENIGMA consortia. Breast Cancer Research, 2014, 16, 3419.	5.0	97
63	Comprehensive annotation of splice junctions supports pervasive alternative splicing at the BRCA1 locus: a report from the ENIGMA consortium. Human Molecular Genetics, 2014, 23, 3666-3680.	2.9	96
64	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.	2.5	95
65	Comparison of mRNA Splicing Assay Protocols across Multiple Laboratories: Recommendations for Best Practice in Standardized Clinical Testing. Clinical Chemistry, 2014, 60, 341-352.	3.2	95
66	Loss of the Inactive X Chromosome and Replication of the Active X in BRCA1-Defective and Wild-type Breast Cancer Cells. Cancer Research, 2005, 65, 2139-2146.	0.9	94
67	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. Nature Communications, 2016, 7, 11375.	12.8	93
68	Lynch Syndromeâ€”Related Endometrial Carcinomas Show a High Frequency of Nonendometrioid Types and of High FIGO Grade Endometrioid Types. International Journal of Surgical Pathology, 2010, 18, 21-26.	0.8	91
69	Interplay between BRCA1 and RHAMM Regulates Epithelial Apicobasal Polarization and May Influence Risk of Breast Cancer. PLoS Biology, 2011, 9, e1001199.	5.6	91
70	FANCMc.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, 5345-5355.	2.9	91
71	Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. Nature Communications, 2019, 10, 1741.	12.8	90
72	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.	5.0	88

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73	Shared heritability and functional enrichment across six solid cancers. Nature Communications, 2019, 10, 431.	12.8	88
74	Common Genetic Variants and Modification of Penetrance of BRCA2-Associated Breast Cancer. PLoS Genetics, 2010, 6, e1001183.	3.5	85
75	Genotype and Phenotype Factors as Determinants for Rectal Stump Cancer in Patients With Familial Adenomatous Polyposis. Annals of Surgery, 2000, 231, 538-543.	4.2	84
76	Mutations of adenomatous polyposis coli (APC) gene are uncommon in sporadic desmoid tumours. British Journal of Cancer, 1998, 78, 582-587.	6.4	82
77	Cyclooxygenase-2 and Platelet-Derived Growth Factor Receptors as Potential Targets in Treating Aggressive Fibromatosis. Clinical Cancer Research, 2007, 13, 5034-5040.	7.0	82
78	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.	2.4	82
79	Associations of obesity and circulating insulin and glucose with breast cancer risk: a Mendelian randomization analysis. International Journal of Epidemiology, 2019, 48, 795-806.	1.9	81
80	Common variants at 12p11, 12q24, 9p21, 9q31.2 and in ZNF365 are associated with breast cancer risk for BRCA1 and/or BRCA2mutation carriers. Breast Cancer Research, 2012, 14, R33.	5.0	78
81	Functional mechanisms underlying pleiotropic risk alleles at the 19p13.1 breastâ€“ovarian cancer susceptibility locus. Nature Communications, 2016, 7, 12675.	12.8	78
82	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
83	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
84	Misbehaviour of XIST RNA in Breast Cancer Cells. PLoS ONE, 2009, 4, e5559.	2.5	75
85	BRCA2 Hypomorphic Missense Variants Confer Moderate Risks of Breast Cancer. Cancer Research, 2017, 77, 2789-2799.	0.9	75
86	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
87	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
88	Associations of common variants at 1p11.2 and 14q24.1 (RAD51L1) with breast cancer risk and heterogeneity by tumor subtype: findings from the Breast Cancer Association Consortiumâ€. Human Molecular Genetics, 2011, 20, 4693-4706.	2.9	71
89	Naturally occurringBRCA2alternative mRNA splicing events in clinically relevant samples. Journal of Medical Genetics, 2016, 53, 548-558.	3.2	69
90	Survival of patients with hereditary colorectal cancer: Comparison of HNPCC and colorectal cancer in FAP patients with sporadic colorectal cancer. , 1999, 80, 183-187.		68

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91	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
92	Evaluation of a 5-Tier Scheme Proposed for Classification of Sequence Variants Using Bioinformatic and Splicing Assay Data: Inter-Reviewer Variability and Promotion of Minimum Reporting Guidelines. Human Mutation, 2013, 34, 1424-1431.	2.5	67
93	Comparative In Vitro and In Silico Analyses of Variants in Splicing Regions of BRCA1 and BRCA2 Genes and Characterization of Novel Pathogenic Mutations. PLoS ONE, 2013, 8, e57173.	2.5	64
94	Age at Menarche and Menopause and Breast Cancer Risk in the International BRCA1/2 Carrier Cohort Study. Cancer Epidemiology Biomarkers and Prevention, 2007, 16, 740-746.	2.5	63
95	Risk of desmoid tumours after open and laparoscopic colectomy in patients with familial adenomatous polyposis. British Journal of Surgery, 2014, 101, 558-565.	0.3	60
96	Evidence that the 5p12 Variant rs10941679 Confers Susceptibility to Estrogen-Receptor-Positive Breast Cancer through FGF10 and MRPS30 Regulation. American Journal of Human Genetics, 2016, 99, 903-911.	6.2	59
97	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
98	miR-342 Regulates BRCA1 Expression through Modulation of ID4 in Breast Cancer. PLoS ONE, 2014, 9, e87039.	2.5	59
99	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
100	Identification of Novel Genetic Markers of Breast Cancer Survival. Journal of the National Cancer Institute, 2015, 107, .	6.3	56
101	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
102	A Human Cell-Based Assay to Evaluate the Effects of Alterations in the MLH1 Mismatch Repair Gene. Cancer Research, 2006, 66, 9036-9044.	0.9	52
103	Genome-wide association study of germline variants and breast cancer-specific mortality. British Journal of Cancer, 2019, 120, 647-657.	6.4	52
104	Recommendations for the implementation of BRCA testing in ovarian cancer patients and their relatives. Critical Reviews in Oncology/Hematology, 2019, 140, 67-72.	4.4	51
105	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
106	Pathology of Tumors Associated With Pathogenic Germline Variants in 9 Breast Cancer Susceptibility Genes. JAMA Oncology, 2022, 8, e216744.	7.1	51
107	Unclassified variants in BRCA genes: guidelines for interpretation. Annals of Oncology, 2011, 22, i18-i23.	1.2	50
108	Rare variants in XRCC2 as breast cancer susceptibility alleles: TableÂ1. Journal of Medical Genetics, 2012, 49, 618-620.	3.2	49

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109	Identification of fifteen novel germline variants in theBRCA13â€²UTR reveals a variant in a breast cancer case that introduces a functionalmiR-103target site. Human Mutation, 2012, 33, 1665-1675.	2.5	49
110	MicroRNA Related Polymorphisms and Breast Cancer Risk. PLoS ONE, 2014, 9, e109973.	2.5	49
111	Characterization of the Cancer Spectrum in Men With GermlineBRCA1andBRCA2Pathogenic Variants. JAMA Oncology, 2020, 6, 1218.	7.1	48
112	The Role of KRAS rs61764370 in Invasive Epithelial Ovarian Cancer: Implications for Clinical Testing. Clinical Cancer Research, 2011, 17, 3742-3750.	7.0	47
113	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
114	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
115	Mean age of tumor onset in hereditary nonpolyposis colorectal cancer (HNPCC) families correlates with the presence of mutations in DNA mismatch repair genes. , 1997, 19, 135-142.		46
116	Mapping of a Putative Tumor Suppressor Locus to Proximal 7p in Wilms Tumors. Genomics, 1996, 37, 310-315.	2.9	45
117	Body mass index and breast cancer survival: a Mendelian randomization analysis. International Journal of Epidemiology, 2017, 46, 1814-1822.	1.9	45
118	Screening for mutations of theAPC gene in 66 Italian familial adenomatous polyposis patients: Evidence for phenotypic differences in cases with and without identified mutation. Human Mutation, 1999, 13, 116-123.	2.5	44
119	Increased frequency of disease-causing MYH mutations in colon cancer families. Carcinogenesis, 2006, 27, 2243-2249.	2.8	44
120	Germline mutations of TP53 and BRCA2 genes in breast cancer/sarcoma families. European Journal of Cancer, 2007, 43, 601-606.	2.8	44
121	BRCA1 p.Val1688del Is a Deleterious Mutation That Recurs in Breast and Ovarian Cancer Families From Northeast Italy. Journal of Clinical Oncology, 2008, 26, 26-31.	1.6	44
122	Genetic predisposition to ductal carcinoma in situ of the breast. Breast Cancer Research, 2016, 18, 22.	5.0	43
123	Involvement of MBD4 inactivation in mismatch repair-deficient tumorigenesis. Oncotarget, 2015, 6, 42892-42904.	1.8	43
124	Inheritance of deleterious mutations at both BRCA1 and BRCA2 in an international sample of 32,295 women. Breast Cancer Research, 2016, 18, 112.	5.0	42
125	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
126	Fine-mapping identifies two additional breast cancer susceptibility loci at 9q31.2. Human Molecular Genetics, 2015, 24, 2966-2984.	2.9	40

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127	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
128	The CHEK2 c.1100delC mutation plays an irrelevant role in breast cancer predisposition in Italy. Human Mutation, 2004, 24, 100-101.	2.5	39
129	Genetic Predisposition to In Situ and Invasive Lobular Carcinoma of the Breast. PLoS Genetics, 2014, 10, e1004285.	3.5	39
130	Association of Genomic Domains in BRCA1 and BRCA2 with Prostate Cancer Risk and Aggressiveness. Cancer Research, 2020, 80, 624-638.	0.9	39
131	Breast Cancer Polygenic Risk Score and Contralateral Breast Cancer Risk. American Journal of Human Genetics, 2020, 107, 837-848.	6.2	39
132	Germline mutations of thePOU6F2 gene in Wilms tumors with loss of heterozygosity on chromosome 7p14. Human Mutation, 2004, 24, 400-407.	2.5	38
133	Analysis of a set of missense, frameshift, and in-frame deletion variants of BRCA1. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2009, 660, 1-11.	1.0	38
134	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
135	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
136	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
137	COMPLEXO: identifying the missing heritability of breast cancer via next generation collaboration. Breast Cancer Research, 2013, 15, 402.	5.0	36
138	11q13 is a susceptibility locus for hormone receptor positive breast cancer. Human Mutation, 2012, 33, 1123-1132.	2.5	35
139	Methylation of O 6-methylguanine-DNA methyltransferase (MGMT) promoter gene in triple-negative breast cancer patients. Breast Cancer Research and Treatment, 2012, 134, 131-137.	2.5	35
140	Ovarian cancer susceptibility alleles and risk of ovarian cancer inBRCA1andBRCA2mutation carriers. Human Mutation, 2012, 33, 690-702.	2.5	34
141	First description of an acinic cell carcinoma of the breast in a BRCA1 mutation carrier: a case report. BMC Cancer, 2013, 13, 46.	2.6	34
142	Assessing Associations between the AURKA-HMMR-TPX2-TUBG1 Functional Module and Breast Cancer Risk in BRCA1/2 Mutation Carriers. PLoS ONE, 2015, 10, e0120020.	2.5	34
143	Analysis of the neurofibromatosis type 2 gene in different human tumors of neuroectodermal origin. Human Genetics, 1996, 97, 638-641.	3.8	33
144	Microsatellite instability in colorectal-cancer patients with suspected genetic predisposition. International Journal of Cancer, 2000, 89, 87-91.	5.1	33

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145	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
146	Decapping protein EDC4 regulates DNA repair and phenocopies BRCA1. Nature Communications, 2018, 9, 967.	12.8	33
147	Common variants of the BRCA1 wild-type allele modify the risk of breast cancer in BRCA1 mutation carriers. Human Molecular Genetics, 2011, 20, 4732-4747.	2.9	32
148	Association of low-penetrance alleles with male breast cancer risk and clinicopathological characteristics: results from a multicenter study in Italy. Breast Cancer Research and Treatment, 2013, 138, 861-868.	2.5	32
149	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
150	Towards controlled terminology for reporting germline cancer susceptibility variants: an ENIGMA report. Journal of Medical Genetics, 2019, 56, 347-357.	3.2	32
151	Transcriptomeâ€wide association study of breast cancer risk by estrogenâ€receptor status. Genetic Epidemiology, 2020, 44, 442-468.	1.3	32
152	Association of breast cancer risk with genetic variants showing differential allelic expression: Identification of a novel breast cancer susceptibility locus at 4q21. Oncotarget, 2016, 7, 80140-80163.	1.8	31
153	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
154	Wholeâ€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.	4.1	31
155	Loss of Heterozygosity Analysis at Different Chromosome Regions in Wilms Tumor Confirms 1p Allelic Loss as a Marker of Worse Prognosis: A Study from the Italian Association of Pediatric Hematology and Oncology. Journal of Urology, 2013, 189, 260-267.	0.4	30
156	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.	6.3	30
157	Germline mutations in BRIP1 and PALB2 in Jewish high cancer risk families. Familial Cancer, 2012, 11, 483-491.	1.9	29
158	The SNP rs895819 in miR-27a is not associated with familial breast cancer risk in Italians. Breast Cancer Research and Treatment, 2012, 133, 805-807.	2.5	28
159	Genomic profiling by wholeâ€genome single nucleotide polymorphism arrays in Wilms tumor and association with relapse. Genes Chromosomes and Cancer, 2012, 51, 644-653.	2.8	28
160	Is Wilms Tumor a Candidate Neoplasia for Treatment with WNT/Î²-Catenin Pathway Modulators?â€"A Report from the Renal Tumors Biology-Driven Drug Development Workshop. Molecular Cancer Therapeutics, 2013, 12, 2619-2627.	4.1	28
161	The FANCM:p.Arg658* truncating variant is associated with risk of triple-negative breast cancer. Npj Breast Cancer, 2019, 5, 38.	5.2	28
162	Four new cases of double heterozygosity for BRCA1 and BRCA2 gene mutations: clinical, pathological, and family characteristics. Breast Cancer Research and Treatment, 2010, 124, 251-258.	2.5	27

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163	Confirmation of 5p12 As a Susceptibility Locus for Progesterone-Receptorâ€"Positive, Lower Grade Breast Cancer. Cancer Epidemiology Biomarkers and Prevention, 2011, 20, 2222-2231.	2.5	27
164	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.	5.0	26
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