Thilo DÃ¶rk

List of Publications by Year
in descending order

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		11235	9346
355	27,003	73	148
papers	citations	h-index	g-index

378	378	378	28828
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Genome-wide association study identifies novel breast cancer susceptibility loci. Nature, 2007, 447, 1087-1093.	13.7	2,165
2	Association analysis identifies 65 new breast cancer risk loci. Nature, 2017, 551, 92-94.	13.7	1,099
3	Large-scale genotyping identifies 41 new loci associated with breast cancer risk. Nature Genetics, 2013, 45, 353-361.	9.4	960
4	Polygenic Risk Scores for Prediction of Breast Cancer and Breast Cancer Subtypes. American Journal of Human Genetics, 2019, 104, 21-34.	2.6	711
5	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.	3.0	596
6	A common coding variant in CASP8 is associated with breast cancer risk. Nature Genetics, 2007, 39, 352-358.	9.4	591
7	Breast Cancer Risk Genes â€” Association Analysis in More than 113,000 Women. New England Journal of Medicine, 2021, 384, 428-439.	13.9	532
8	Nuclear factor TDP-43 and SR proteins promote in vitro and in vivo CFTR exon 9 skipping. EMBO Journal, 2001, 20, 1774-1784.	3.5	531
9	Genome-wide association analysis of more than 120,000 individuals identifies 15 new susceptibility loci for breast cancer. Nature Genetics, 2015, 47, 373-380.	9.4	513
10	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.	9.4	493
11	Newly discovered breast cancer susceptibility loci on 3p24 and 17q23.2. Nature Genetics, 2009, 41, 585-590.	9.4	434
12	Prediction of Breast Cancer Risk Based on Profiling With Common Genetic Variants. Journal of the National Cancer Institute, 2015, 107, .	3.0	428
13	Identification of seven new prostate cancer susceptibility loci through a genome-wide association study. Nature Genetics, 2009, 41, 1116-1121.	9.4	389
14	Genome-wide association studies identify four ER negativeâ€“specific breast cancer risk loci. Nature Genetics, 2013, 45, 392-398.	9.4	374
15	Large-scale genomic analyses link reproductive aging to hypothalamic signaling, breast cancer susceptibility and BRCA1-mediated DNA repair. Nature Genetics, 2015, 47, 1294-1303.	9.4	357
16	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. Nature Genetics, 2017, 49, 680-691.	9.4	356
17	GWAS meta-analysis and replication identifies three new susceptibility loci for ovarian cancer. Nature Genetics, 2013, 45, 362-370.	9.4	326
18	A genome-wide association study identifies susceptibility loci for ovarian cancer at 2q31 and 8q24. Nature Genetics, 2010, 42, 874-879.	9.4	321

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19	Heterogeneity of Breast Cancer Associations with Five Susceptibility Loci by Clinical and Pathological Characteristics. PLoS Genetics, 2008, 4, e1000054.	1.5	315
20	Germline Mutations in the BRIP1, BARD1, PALB2, and NBN Genes in Women With Ovarian Cancer. Journal of the National Cancer Institute, 2015, 107, .	3.0	311
21	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. Nature Genetics, 2017, 49, 1767-1778.	9.4	289
22	Missense mutations but not allelic variants alter the function of ATM by dominant interference in patients with breast cancer. Proceedings of the National Academy of Sciences of the United States of America, 2002, 99, 925-930.	3.3	287
23	A genome-wide association study identifies a new ovarian cancer susceptibility locus on 9p22.2. Nature Genetics, 2009, 41, 996-1000.	9.4	276
24	Seven prostate cancer susceptibility loci identified by a multi-stage genome-wide association study. Nature Genetics, 2011, 43, 785-791.	9.4	265
25	Genome-wide association study identifies 32 novel breast cancer susceptibility loci from overall and subtype-specific analyses. Nature Genetics, 2020, 52, 572-581.	9.4	265
26	Genome-wide association analysis identifies three new breast cancer susceptibility loci. Nature Genetics, 2012, 44, 312-318.	9.4	256
27	Distinct spectrum of CFTR gene mutations in congenital absence of vas deferens. Human Genetics, 1997, 100, 365-377.	1.8	242
28	Identification of six new susceptibility loci for invasive epithelial ovarian cancer. Nature Genetics, 2015, 47, 164-171.	9.4	221
29	Dominant Negative ATM Mutations in Breast Cancer Families. Journal of the National Cancer Institute, 2002, 94, 205-215.	3.0	217
30	Human RAD50 Deficiency in a Nijmegen Breakage Syndrome-like Disorder. American Journal of Human Genetics, 2009, 84, 605-616.	2.6	217
31	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.	2.6	201
32	A new type of mutation causes a splicing defect in ATM. Nature Genetics, 2002, 30, 426-429.	9.4	200
33	Genetic predisposition to mosaic Y chromosome loss in blood. Nature, 2019, 575, 652-657.	13.7	198
34	Characterization of ATM gene mutations in 66 ataxia telangiectasia families. Human Molecular Genetics, 1999, 8, 69-79.	1.4	191
35	A transcriptome-wide association study of 229,000 women identifies new candidate susceptibility genes for breast cancer. Nature Genetics, 2018, 50, 968-978.	9.4	184
36	Testis-specific protein, Y-encoded (TSPY) expression in testicular tissues. Human Molecular Genetics, 1996, 5, 1801-1807.	1.4	183

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37	Identification of nine new susceptibility loci for endometrial cancer. Nature Communications, 2018, 9, 3166.	5.8	178
38	<i>PALB2</i>,<i>CHEK2</i>and<i>ATM</i>rare variants and cancer risk: data from COGS. Journal of Medical Genetics, 2016, 53, 800-811.	1.5	174
39	<i>CHEK2</i>*1100delC Heterozygosity in Women With Breast Cancer Associated With Early Death, Breast Cancerâ€"Specific Death, and Increased Risk of a Second Breast Cancer. Journal of Clinical Oncology, 2012, 30, 4308-4316.	0.8	162
40	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, 1052-1067.	7.7	157
41	Low penetrance breast cancer susceptibility loci are associated with specific breast tumor subtypes: findings from the Breast Cancer Association Consortium. Human Molecular Genetics, 2011, 20, 3289-3303.	1.4	152
42	Age- and Tumor Subtypeâ€"Specific Breast Cancer Risk Estimates for <i>CH</i><i>EK</i><i>2</i>*1100delC Carriers. Journal of Clinical Oncology, 2016, 34, 2750-2760.	0.8	152
43	Multiple Novel Prostate Cancer Predisposition Loci Confirmed by an International Study: The PRACTICAL Consortium. Cancer Epidemiology Biomarkers and Prevention, 2008, 17, 2052-2061.	1.1	148
44	Epigenetic analysis leads to identification of HNF1B as a subtype-specific susceptibility gene for ovarian cancer. Nature Communications, 2013, 4, 1628.	5.8	144
45	Nonclassical splicing mutations in the coding and noncoding regions of the ATM Gene: Maximum entropy estimates of splice junction strengths. Human Mutation, 2004, 23, 67-76.	1.1	133
46	Characterization of a novel 21-kb deletion, CFTRdele2,3(21 kb), in the CFTR gene: a cystic fibrosis mutation of Slavic origin common in Central and East Europe. Human Genetics, 2000, 106, 259-268.	1.8	129
47	Breast cancer risk variants at 6q25 display different phenotype associations and regulate ESR1, RMND1 and CCDC170. Nature Genetics, 2016, 48, 374-386.	9.4	125
48	Nijmegen Breakage Syndrome mutations and risk of breast cancer. International Journal of Cancer, 2008, 122, 802-806.	2.3	120
49	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. Nature Genetics, 2020, 52, 56-73.	9.4	120
50	A meta-analysis of genome-wide association studies to identify prostate cancer susceptibility loci associated with aggressive and non-aggressive disease. Human Molecular Genetics, 2013, 22, 408-415.	1.4	118
51	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.	3.9	118
52	Immaturity, perinatal inflammation, and retinopathy of prematurity: A multi-hit hypothesis. Early Human Development, 2009, 85, 325-329.	0.8	113
53	Association of vitamin D levels and risk of ovarian cancer: a Mendelian randomization study. International Journal of Epidemiology, 2016, 45, 1619-1630.	0.9	111
54	Identification of a BRCA2-Specific Modifier Locus at 6p24 Related to Breast Cancer Risk. PLoS Genetics, 2013, 9, e1003173.	1.5	105

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55	Evidence that breast cancer risk at the 2q35 locus is mediated through IGFBP5 regulation. Nature Communications, 2014, 5, 4999.	5.8	105
56	Genetic determinants of airways' colonisation with Pseudomonas aeruginosa in cystic fibrosis. Lancet, The, 1993, 341, 189-193.	6.3	100
57	Detection of more than 50 different CFTR mutations in a large group of German cystic fibrosis patients. Human Genetics, 1994, 94, 533-542.	1.8	100
58	Risk of Estrogen Receptorâ€"Positive and â€"Negative Breast Cancer and Singleâ€"Nucleotide Polymorphism 2q35-rs13387042. Journal of the National Cancer Institute, 2009, 101, 1012-1018.	3.0	99
59	Height and Breast Cancer Risk: Evidence From Prospective Studies and Mendelian Randomization. Journal of the National Cancer Institute, 2015, 107, djv219.	3.0	99
60	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.	2.6	98
61	Identification and molecular characterization of a new ovarian cancer susceptibility locus at 17q21.31. Nature Communications, 2013, 4, 1627.	5.8	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.	2.2	97
63	CFTR gene mutations and male infertility. Andrologia, 2000, 32, 71-83.	1.0	95
64	Indicators of late normal tissue response after radiotherapy for head and neck cancer: fibroblasts, lymphocytes, genetics, DNA repair, and chromosome aberrations. Radiotherapy and Oncology, 2002, 64, 141-152.	0.3	94
65	No evidence that protein truncating variants in<i>BRIP1</i>are associated with breast cancer risk: implications for gene panel testing. Journal of Medical Genetics, 2016, 53, 298-309.	1.5	94
66	Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. Nature Communications, 2019, 10, 1741.	5.8	90
67	Shared heritability and functional enrichment across six solid cancers. Nature Communications, 2019, 10, 431.	5.8	88
68	Association of ESR1 gene tagging SNPs with breast cancer risk. Human Molecular Genetics, 2009, 18, 1131-1139.	1.4	84
69	ATM polymorphisms as risk factors for prostate cancer development. British Journal of Cancer, 2004, 91, 783-787.	2.9	82
70	Association of two mutations in theCHEK2 gene with breast cancer. International Journal of Cancer, 2005, 116, 263-266.	2.3	82
71	Associations of obesity and circulating insulin and glucose with breast cancer risk: a Mendelian randomization analysis. International Journal of Epidemiology, 2019, 48, 795-806.	0.9	81
72	Do MDM2 SNP309 and TP53 R72P Interact in Breast Cancer Susceptibility? A Large Pooled Series from the Breast Cancer Association Consortium. Cancer Research, 2007, 67, 9584-9590.	0.4	80

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73	The role of genetic breast cancer susceptibility variants as prognostic factors. Human Molecular Genetics, 2012, 21, 3926-3939.	1.4	80
74	Genome-wide significant risk associations for mucinous ovarian carcinoma. Nature Genetics, 2015, 47, 888-897.	9.4	78
75	Functional mechanisms underlying pleiotropic risk alleles at the 19p13.1 breastâ€"ovarian cancer susceptibility locus. Nature Communications, 2016, 7, 12675.	5.8	78
76	Five endometrial cancer risk loci identified through genome-wide association analysis. Nature Genetics, 2016, 48, 667-674.	9.4	77
77	BRCA2 Polymorphic Stop Codon K3326X and the Risk of Breast, Prostate, and Ovarian Cancers. Journal of the National Cancer Institute, 2016, 108, djv315.	3.0	77
78	Functional consequences ofATMsequence variants for chromosomal radiosensitivity. Genes Chromosomes and Cancer, 2004, 40, 109-119.	1.5	76
79	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.	2.6	76
80	<i>BRCA2</i> Hypomorphic Missense Variants Confer Moderate Risks of Breast Cancer. Cancer Research, 2017, 77, 2789-2799.	0.4	75
81	ATM Protein-dependent Phosphorylation of Rad50 Protein Regulates DNA Repair and Cell Cycle Control. Journal of Biological Chemistry, 2011, 286, 31542-31556.	1.6	74
82	Intra- and extragenic marker haplotypes of CFTR mutations in cystic fibrosis families. Human Genetics, 1992, 88, 417-425.	1.8	73
83	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.	1.4	71
84	Adult body mass index and risk of ovarian cancer by subtype: a Mendelian randomization study. International Journal of Epidemiology, 2016, 45, 884-895.	0.9	71
85	Characterization of a Novel Third-Generation Anti-CD24-CAR against Ovarian Cancer. International Journal of Molecular Sciences, 2019, 20, 660.	1.8	70
86	Radiosensitivity of Ataxia Telangiectasia and Nijmegen Breakage Syndrome Homozygotes and Heterozygotes as Determined by Three-Color FISH Chromosome Painting. Radiation Research, 2002, 157, 312-321.	0.7	69
87	Shared genetics underlying epidemiological association between endometriosis and ovarian cancer. Human Molecular Genetics, 2015, 24, 5955-5964.	1.4	68
88	Genetic modifiers of CHEK2*1100delC-associated breast cancer risk. Genetics in Medicine, 2017, 19, 599-603.	1.1	67
89	Aberrant overexpression of miR-421 downregulates ATM and leads to a pronounced DSB repair defect and clinical hypersensitivity in SKX squamous cell carcinoma. Radiotherapy and Oncology, 2013, 106, 147-154.	0.3	66
90	Improved Killing of Ovarian Cancer Stem Cells by Combining a Novel Chimeric Antigen Receptorâ€"Based Immunotherapy and Chemotherapy. Human Gene Therapy, 2017, 28, 886-896.	1.4	65

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91	Genetic Risk Score Mendelian Randomization Shows that Obesity Measured as Body Mass Index, but not Waist:Hip Ratio, Is Causal for Endometrial Cancer. Cancer Epidemiology Biomarkers and Prevention, 2016, 25, 1503-1510.	1.1	64
92	Cis-eQTL analysis and functional validation of candidate susceptibility genes for high-grade serous ovarian cancer. Nature Communications, 2015, 6, 8234.	5.8	63
93	CYP19A1 fine-mapping and Mendelian randomization: estradiol is causal for endometrial cancer. Endocrine-Related Cancer, 2016, 23, 77-91.	1.6	62
94	Genetic overlap between endometriosis and endometrial cancer: evidence from crossâ€disease genetic correlation and GWAS metaâ€analyses. Cancer Medicine, 2018, 7, 1978-1987.	1.3	62
95	Frequency of BRCA1 Mutation 5382insC in German Breast Cancer Patients. Gynecologic Oncology, 1999, 72, 402-406.	0.6	60
96	Clinical radiosensitivity in breast cancer patients carrying pathogenic ATM gene mutations: no observation of increased radiation-induced acute or late effects. Radiotherapy and Oncology, 2003, 69, 155-160.	0.3	60
97	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.	2.6	59
98	A Single UDP-galactofuranose Transporter Is Required for Galactofuranosylation in Aspergillus fumigatus. Journal of Biological Chemistry, 2009, 284, 33859-33868.	1.6	58
99	Five Polymorphisms and Breast Cancer Risk: Results from the Breast Cancer Association Consortium. Cancer Epidemiology Biomarkers and Prevention, 2009, 18, 1610-1616.	1.1	57
100	Nonsense mutation p.Q548X in BLM, the gene mutated in Bloomâ€™s syndrome, is associated with breast cancer in Slavic populations. Breast Cancer Research and Treatment, 2013, 137, 533-539.	1.1	56
101	Risk Analysis of Prostate Cancer in PRACTICAL, a Multinational Consortium, Using 25 Known Prostate Cancer Susceptibility Loci. Cancer Epidemiology Biomarkers and Prevention, 2015, 24, 1121-1129.	1.1	56
102	Polymorphisms of the humanÎ²-defensin-1gene. Molecular and Cellular Probes, 1998, 12, 171-173.	0.9	55
103	Resveratrol modulates DNA double-strand break repair pathways in an ATM/ATRâ€“p53- and â€“Nbs1-dependent manner. Carcinogenesis, 2008, 29, 519-527.	1.3	54
104	Functional characterization connects individual patient mutations in <i>ataxia telangiectasia mutated (ATM)</i> with dysfunction of specific DNA doubleâ€strand breakâ€repair signaling pathways. FASEB Journal, 2011, 25, 3849-3860.	0.2	54
105	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.	0.4	54
106	Cystic-fibrosis-like disease unrelated to the cystic fibrosis transmembrane conductance regulator. Human Genetics, 1998, 102, 582-586.	1.8	53
107	Common non-synonymous SNPs associated with breast cancer susceptibility: findings from the Breast Cancer Association Consortium. Human Molecular Genetics, 2014, 23, 6096-6111.	1.4	53
108	Transcript analysis of CFTR nonsense mutations in lymphocytes and nasal epithelial cells from cystic fibrosis patients. Human Mutation, 1995, 5, 210-220.	1.1	52

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109	Genome-wide association study of germline variants and breast cancer-specific mortality. British Journal of Cancer, 2019, 120, 647-657.	2.9	52
110	Mutations of the CFTR gene in Turkish patients with congenital bilateral absence of the vas deferens. Human Reproduction, 2004, 19, 1094-1100.	0.4	51
111	Double heterozygotes among breast cancer patients analyzed for BRCA1, CHEK2, ATM, NBN/NBS1, and BLM germ-line mutations. Breast Cancer Research and Treatment, 2014, 145, 553-562.	1.1	51
112	Fineâ€scale mapping of 8q24 locus identifies multiple independent risk variants for breast cancer. International Journal of Cancer, 2016, 139, 1303-1317.	2.3	51
113	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.	1.1	51
114	Pathology of Tumors Associated With Pathogenic Germline Variants in 9 Breast Cancer Susceptibility Genes. JAMA Oncology, 2022, 8, e216744.	3.4	51
115	Fine-mapping of the HNF1B multicancer locus identifies candidate variants that mediate endometrial cancer risk. Human Molecular Genetics, 2015, 24, 1478-1492.	1.4	50
116	MicroRNA Related Polymorphisms and Breast Cancer Risk. PLoS ONE, 2014, 9, e109973.	1.1	49
117	Genetic Data from Nearly 63,000 Women of European Descent Predicts DNA Methylation Biomarkers and Epithelial Ovarian Cancer Risk. Cancer Research, 2019, 79, 505-517.	0.4	49
118	Functional Polymorphisms in the TERT Promoter Are Associated with Risk of Serous Epithelial Ovarian and Breast Cancers. PLoS ONE, 2011, 6, e24987.	1.1	48
119	Hereditary breast cancer: ever more pieces to the polygenic puzzle. Hereditary Cancer in Clinical Practice, 2013, 11, 12.	0.6	48
120	Risk of Ovarian Cancer and the NF-Î°B Pathway: Genetic Association with <i>IL1A</i> and <i>TNFSF10</i>. Cancer Research, 2014, 74, 852-861.	0.4	48
121	Family History, Genetic Testing, and Clinical Risk Prediction: Pooled Analysis of CHEK2*1100delC in 1,828 Bilateral Breast Cancers and 7,030 Controls. Cancer Epidemiology Biomarkers and Prevention, 2009, 18, 230-234.	1.1	47
122	A role for XRCC2 gene polymorphisms in breast cancer risk and survival. Journal of Medical Genetics, 2011, 48, 477-484.	1.5	47
123	PALB2 mutations in German and Russian patients with bilateral breast cancer. Breast Cancer Research and Treatment, 2011, 126, 545-550.	1.1	47
124	The Role of KRAS rs61764370 in Invasive Epithelial Ovarian Cancer: Implications for Clinical Testing. Clinical Cancer Research, 2011, 17, 3742-3750.	3.2	47
125	Novel and characteristic CFTR mutations in Saudi Arab children with severe cystic fibrosis.. Journal of Medical Genetics, 1997, 34, 996-999.	1.5	46
126	Body mass index and breast cancer survival: a Mendelian randomization analysis. International Journal of Epidemiology, 2017, 46, 1814-1822.	0.9	45

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127	Exon 9 of the CFTR gene: splice site haplotypes and cystic fibrosis mutations. Human Genetics, 1994, 93, 67-73.	1.8	44
128	Fanconiâ€™s Anemia and Clinical Radiosensitivity. Strahlentherapie Und Onkologie, 2003, 179, 748-753.	1.0	44
129	Mutation Analysis of BRCA1, BRCA2, PALB2 and BRD7 in a Hospital-Based Series of German Patients with Triple-Negative Breast Cancer. PLoS ONE, 2012, 7, e47993.	1.1	44
130	Common Genetic Variation In Cellular Transport Genes and Epithelial Ovarian Cancer (EOC) Risk. PLoS ONE, 2015, 10, e0128106.	1.1	44
131	Genetic predisposition to ductal carcinoma in situ of the breast. Breast Cancer Research, 2016, 18, 22.	2.2	43
132	<i>PPM1D</i>Mosaic Truncating Variants in Ovarian Cancer Cases May Be Treatment-Related Somatic Mutations. Journal of the National Cancer Institute, 2016, 108, djv347.	3.0	43
133	Germline whole exome sequencing and large-scale replication identifies FANCM as a likely high grade serous ovarian cancer susceptibility gene. Oncotarget, 2017, 8, 50930-50940.	0.8	43
134	Haplotype analysis of 94 cystic fibrosis mutations with seven polymorphicCFTR DNA markers. , 1996, 8, 149-159.		42
135	Mutations of the BRCA1 and BRCA2 genes in patients with bilateral breast cancer. British Journal of Cancer, 2001, 85, 850-858.	2.9	42
136	A nonsense mutation (E1978X) in the ATM gene is associated with breast cancer. Breast Cancer Research and Treatment, 2009, 118, 207-211.	1.1	42
137	ATM missense variant P1054R predisposes to prostate cancer. Radiotherapy and Oncology, 2007, 83, 283-288.	0.3	40
138	High frequency and alleleâ€specific differences of <i>BRCA1</i> founder mutations in breast cancer and ovarian cancer patients from Belarus. Clinical Genetics, 2010, 78, 364-372.	1.0	40
139	Cell-type-specific enrichment of risk-associated regulatory elements at ovarian cancer susceptibility loci. Human Molecular Genetics, 2015, 24, 3595-3607.	1.4	40
140	Fine-mapping identifies two additional breast cancer susceptibility loci at 9q31.2. Human Molecular Genetics, 2015, 24, 2966-2984.	1.4	40
141	A cystic fibrosis allele encoding missense mutations in both nucleotide binding folds of the cystic fibrosis transmembrane conductance regulator. Human Mutation, 1992, 1, 204-210.	1.1	39
142	Genetic Predisposition to In Situ and Invasive Lobular Carcinoma of the Breast. PLoS Genetics, 2014, 10, e1004285.	1.5	39
143	Patient survival and tumor characteristics associated with CHEK2:p.I157T â€“ findings from the Breast Cancer Association Consortium. Breast Cancer Research, 2016, 18, 98.	2.2	39
144	Recreational physical inactivity and mortality in women with invasive epithelial ovarian cancer: evidence from the Ovarian Cancer Association Consortium. British Journal of Cancer, 2016, 115, 95-101.	2.9	39

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145	Breast Cancer Polygenic Risk Score and Contralateral Breast Cancer Risk. American Journal of Human Genetics, 2020, 107, 837-848.	2.6	39
146	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.	1.4	38
147	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.	2.6	37
148	Evidence of a genetic link between endometriosis and ovarian cancer. Fertility and Sterility, 2016, 105, 35-43.e10.	0.5	37
149	Interleukin-10 High Producer Allele and Ultrasound-Defined Periventricular White Matter Abnormalities in Preterm Infants: A Preliminary Study. Neuropediatrics, 2006, 37, 130-136.	0.3	36
150	Low prevalence of SPINK1 gene mutations in adult patients with chronic idiopathic pancreatitis. Journal of Medical Genetics, 2001, 38, 243-244.	1.5	36
151	Combined effects of single nucleotide polymorphisms TP53 R72P and MDM2 SNP309, and p53 expression on survival of breast cancer patients. Breast Cancer Research, 2009, 11, R89.	2.2	35
152	11q13 is a susceptibility locus for hormone receptor positive breast cancer. Human Mutation, 2012, 33, 1123-1132.	1.1	35
153	Meta-analysis of genome-wide association studies identifies common susceptibility polymorphisms for colorectal and endometrial cancer near SH2B3 and TSHZ1. Scientific Reports, 2015, 5, 17369.	1.6	35
154	Genetically Predicted Levels of DNA Methylation Biomarkers and Breast Cancer Risk: Data From 228â€‰951 Women of European Descent. Journal of the National Cancer Institute, 2020, 112, 295-304.	3.0	35
155	Mendelian randomization analyses suggest a role for cholesterol in the development of endometrial cancer. International Journal of Cancer, 2021, 148, 307-319.	2.3	35
156	Evidence for a Common Ethnic Origin of Cystic Fibrosis Mutation 3120+1Gâ†'A in Diverse Populations. American Journal of Human Genetics, 1998, 63, 656-662.	2.6	34
157	Breast cancer in female carriers of ATM gene alterations: outcome of adjuvant radiotherapy. Radiotherapy and Oncology, 2004, 72, 319-323.	0.3	34
158	<i>SFRP1</i>CpG island methylation locus is associated with renal cell cancer susceptibility and disease recurrence. Epigenetics, 2012, 7, 447-457.	1.3	34
159	Candidate locus analysis of the TERTâ€"CLPTM1L cancer risk region on chromosome 5p15 identifies multiple independent variants associated with endometrial cancer risk. Human Genetics, 2015, 134, 231-245.	1.8	34
160	Missense Variants in <i>ATM</i> in 26,101 Breast Cancer Cases and 29,842 Controls. Cancer Epidemiology Biomarkers and Prevention, 2010, 19, 2143-2151.	1.1	33
161	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.	1.4	33
162	Slow progression of ataxia-telangiectasia with double missense and in frame splice mutations. , 2004, 126A, 272-277.		32

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163	Diversity of the basic defect of homozygous CFTR mutation genotypes in humans. Journal of Medical Genetics, 2007, 45, 47-54.	1.5	32
164	Genome-Wide Association Study Identifies a Possible Susceptibility Locus for Endometrial Cancer. Cancer Epidemiology Biomarkers and Prevention, 2012, 21, 980-987.	1.1	32
165	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.	1.4	32
166	Towards controlled terminology for reporting germline cancer susceptibility variants: an ENIGMA report. Journal of Medical Genetics, 2019, 56, 347-357.	1.5	32
167	Genetic Susceptibility to Endometrial Cancer: Risk Factors and Clinical Management. Cancers, 2020, 12, 2407.	1.7	32
168	Transcriptomeâ€wide association study of breast cancer risk by estrogenâ€receptor status. Genetic Epidemiology, 2020, 44, 442-468.	0.6	32
169	A novel exon in the cystic fibrosis transmembrane conductance regulator gene activated by the nonsense mutation E92X in airway epithelial cells of patients with cystic fibrosis.. Journal of Clinical Investigation, 1994, 93, 1852-1859.	3.9	31
170	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.	0.8	31
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