Ian P M Tomlinson

List of Publications by Year
in descending order

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232 papers	26,286 citations	7568 77 h-index	7518 151 g-index
260 all docs	260 docs citations	260 times ranked	30784 citing authors

#	Article	IF	Citations
1	Germline mutations in FH predispose to dominantly inherited uterine fibroids, skin leiomyomata and papillary renal cell cancer. Nature Genetics, 2002, 30, 406-410.	21.4	1,426
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	Germline mutations affecting the proofreading domains of POLE and POLD1 predispose to colorectal adenomas and carcinomas. Nature Genetics, 2013, 45, 136-144.	21.4	851
5	A genome-wide association scan of tag SNPs identifies a susceptibility variant for colorectal cancer at 8q24.21. Nature Genetics, 2007, 39, 984-988.	21.4	754
6	Polygenic Risk Scores for Prediction of Breast Cancer and Breast Cancer Subtypes. American Journal of Human Genetics, 2019, 104, 21-34.	6.2	711
7	Genome-wide association scan identifies a colorectal cancer susceptibility locus on 11q23 and replicates risk loci at 8q24 and 18q21. Nature Genetics, 2008, 40, 631-637.	21.4	542
8	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
9	A genome-wide association study identifies colorectal cancer susceptibility loci on chromosomes 10p14 and 8q23.3. Nature Genetics, 2008, 40, 623-630.	21.4	514
10	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
11	Meta-analysis of genome-wide association data identifies four new susceptibility loci for colorectal cancer. Nature Genetics, 2008, 40, 1426-1435.	21.4	498
12	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
13	The common colorectal cancer predisposition SNP rs6983267 at chromosome 8q24 confers potential to enhanced Wnt signaling. Nature Genetics, 2009, 41, 885-890.	21.4	463
14	Prediction of Breast Cancer Risk Based on Profiling With Common Genetic Variants. Journal of the National Cancer Institute, 2015, 107, .	6.3	428
15	Genome-wide association studies identify four ER negativeâ€"specific breast cancer risk loci. Nature Genetics, 2013, 45, 392-398.	21.4	374
16	Deep learning for prediction of colorectal cancer outcome: a discovery and validation study. Lancet, The, 2020, 395, 350-360.	13.7	364
17	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
18	The type of somatic mutation at APC in familial adenomatous polyposis is determined by the site of the germline mutation: a new facet to Knudson's 'two-hit' hypothesis. Nature Medicine, 1999, 5, 1071-1075.	30.7	339

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19	Meta-analysis of three genome-wide association studies identifies susceptibility loci for colorectal cancer at 1q41, 3q26.2, 12q13.13 and 20q13.33. Nature Genetics, 2010, 42, 973-977.	21.4	335
20	A genome-wide association study identifies susceptibility loci for ovarian cancer at 2q31 and 8q24. Nature Genetics, 2010, 42, 874-879.	21.4	321
21	DNA polymerase É› and Î´ exonuclease domain mutations in endometrial cancer. Human Molecular Genetics, 2013, 22, 2820-2828.	2.9	319
22	Factors influencing success of clinical genome sequencing across a broad spectrum of disorders. Nature Genetics, 2015, 47, 717-726.	21.4	310
23	Microsatellite instability and the clinicopathological features of sporadic colorectal cancer. Gut, 2001, 48, 821-829.	12.1	308
24	A panoply of errors: polymerase proofreading domain mutations in cancer. Nature Reviews Cancer, 2016, 16, 71-81.	28.4	292
25	Selection, the mutation rate and cancer: Ensuring that the tail does not wag the dog. Nature Medicine, 1999, 5, 11-12.	30.7	289
26	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. Nature Genetics, 2017, 49, 1767-1778.	21.4	289
27	Common genetic variants at the CRAC1 (HMPS) locus on chromosome 15q13.3 influence colorectal cancer risk. Nature Genetics, 2008, 40, 26-28.	21.4	277
28	Clinical relevance of DPYD variants c.1679T>G, c.1236G>A/HapB3, and c.1601G>A as predictors of severe fluoropyrimidine-associated toxicity: a systematic review and meta-analysis of individual patient data. Lancet Oncology, The, 2015, 16, 1639-1650.	10.7	277
29	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
30	Guidelines for the management of hereditary colorectal cancer from the British Society of Gastroenterology (BSG)/Association of Coloproctology of Great Britain and Ireland (ACPGBI)/United Kingdom Cancer Genetics Group (UKCGG). Gut, 2020, 69, 411-444.	12.1	263
31	British Society of Gastroenterology position statement on serrated polyps in the colon and rectum. Gut, 2017, 66, 1181-1196.	12.1	250
32	Germline and somatic polymerase Ïµ and Î´ mutations define a new class of hypermutated colorectal and endometrial cancers. Journal of Pathology, 2013, 230, 148-153.	4.5	242
33	Exome sequencing of osteosarcoma reveals mutation signatures reminiscent of BRCA deficiency. Nature Communications, 2015, 6, 8940.	12.8	242
34	Prognostic Significance of POLE Proofreading Mutations in Endometrial Cancer. Journal of the National Cancer Institute, 2015, 107, 402.	6.3	229
35	Somatic POLE proofreading domain mutation, immune response, and prognosis in colorectal cancer: a retrospective, pooled biomarker study. The Lancet Gastroenterology and Hepatology, 2016, 1, 207-216.	8.1	227
36	Hereditary mixed polyposis syndrome is caused by a 40-kb upstream duplication that leads to increased and ectopic expression of the BMP antagonist GREM1. Nature Genetics, 2012, 44, 699-703.	21.4	222

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37	Multiple ways of silencing E-cadherin gene expression in lobular carcinoma of the breast. International Journal of Cancer, 2001, 92, 404-408.	5.1	217
38	Aberrant epithelial GREM1 expression initiates colonic tumorigenesis from cells outside the stem cell niche. Nature Medicine, 2015, 21, 62-70.	30.7	213
39	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
40	Genetic predisposition to mosaic Y chromosome loss in blood. Nature, 2019, 575, 652-657.	27.8	198
41	Multiple Common Susceptibility Variants near BMP Pathway Loci GREM1, BMP4, and BMP2 Explain Part of the Missing Heritability of Colorectal Cancer. PLoS Genetics, 2011, 7, e1002105.	3.5	188
42	Identification of nine new susceptibility loci for endometrial cancer. Nature Communications, 2018, 9, 3166.	12.8	178
43	PALB2,CHEK2andATMrare variants and cancer risk: data from COGS. Journal of Medical Genetics, 2016, 53, 800-811.	3.2	174
44	Association analyses identify 31 new risk loci for colorectal cancer susceptibility. Nature Communications, 2019, 10, 2154.	12.8	172
45	FBXW7 influences murine intestinal homeostasis and cancer, targeting Notch, Jun, and DEK for degradation. Journal of Experimental Medicine, 2011, 208, 295-312.	8.5	159
46	Age- and Tumor Subtypeâ€“Specific Breast Cancer Risk Estimates for CHEK2*1100delC Carriers. Journal of Clinical Oncology, 2016, 34, 2750-2760.	1.6	152
47	Image-based consensus molecular subtype (imCMS) classification of colorectal cancer using deep learning. Gut, 2021, 70, 544-554.	12.1	148
48	Mutations in DPC4 (SMAD4) cause juvenile polyposis syndrome, but only account for a minority of cases. Human Molecular Genetics, 1998, 7, 1907-1912.	2.9	142
49	Genome-wide association study identifies a common variant associated with risk of endometrial cancer. Nature Genetics, 2011, 43, 451-454.	21.4	141
50	Putative cis-regulatory drivers in colorectal cancer. Nature, 2014, 512, 87-90.	27.8	136
51	Genome-wide association studies in oesophageal adenocarcinoma and Barrett's oesophagus: a large-scale meta-analysis. Lancet Oncology, The, 2016, 17, 1363-1373.	10.7	133
52	The TCA cycle and tumorigenesis: the examples of fumarate hydratase and succinate dehydrogenase. Annals of Medicine, 2003, 35, 634-635.	3.8	131
53	Adjuvant capecitabine plus bevacizumab versus capecitabine alone in patients with colorectal cancer (QUASAR 2): an open-label, randomised phase 3 trial. Lancet Oncology, The, 2016, 17, 1543-1557.	10.7	129
54	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

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55	Detecting repeated cancer evolution from multi-region tumor sequencing data. Nature Methods, 2018, 15, 707-714.	19.0	124
56	Expression of Idh1R132H in the Murine Subventricular Zone Stem Cell Niche Recapitulates Features of Early Gliomagenesis. Cancer Cell, 2016, 30, 578-594.	16.8	122
57	Rare disruptive mutations and their contribution to the heritable risk of colorectal cancer. Nature Communications, 2016, 7, 11883.	12.8	122
58	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. Nature Genetics, 2020, 52, 56-73.	21.4	120
59	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.	8.4	118
60	F-box and WD Repeat Domain-Containing 7 Regulates Intestinal Cell Lineage Commitment and Is a Haploinsufficient Tumor Suppressor. Gastroenterology, 2010, 139, 929-941.	1.3	114
61	How Many Mutations in a Cancer?. American Journal of Pathology, 2002, 160, 755-758.	3.8	110
62	Evidence that breast cancer risk at the 2q35 locus is mediated through IGFBP5 regulation. Nature Communications, 2014, 5, 4999.	12.8	105
63	The mini-driver model of polygenic cancer evolution. Nature Reviews Cancer, 2015, 15, 680-685.	28.4	104
64	Recurrent chromosomal gains and heterogeneous driver mutations characterise papillary renal cancer evolution. Nature Communications, 2015, 6, 6336.	12.8	100
65	Clinical management of hereditary colorectal cancer syndromes. Nature Reviews Gastroenterology and Hepatology, 2015, 12, 88-97.	17.8	99
66	Height and Breast Cancer Risk: Evidence From Prospective Studies and Mendelian Randomization. Journal of the National Cancer Institute, 2015, 107, djv219.	6.3	99
67	The evolutionary landscape of colorectal tumorigenesis. Nature Ecology and Evolution, 2018, 2, 1661-1672.	7.8	99
68	An FBXW7-ZEB2 axis links EMT and tumour microenvironment to promote colorectal cancer stem cells and chemoresistance. Oncogenesis, 2019, 8, 13.	4.9	99
69	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
70	Evolutionary history of human colitis-associated colorectal cancer. Gut, 2019, 68, 985-995.	12.1	97
71	Does MSI-low exist?. Journal of Pathology, 2002, 197, 6-13.	4.5	95
72	No evidence that protein truncating variants inBRIP1are associated with breast cancer risk: implications for gene panel testing. Journal of Medical Genetics, 2016, 53, 298-309.	3.2	94

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73	Polymorphisms Near TBX5 and GDF7 Are Associated With Increased Risk for Barrettâ€™s Esophagus. Gastroenterology, 2015, 148, 367-378.	1.3	93
74	A candidate gene study of capecitabine-related toxicity in colorectal cancer identifies new toxicity variants atDPYDand a putative role forENOSF1rather thanTYMS. Gut, 2015, 64, 111-120.	12.1	93
75	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. Nature Communications, 2016, 7, 11375.	12.8	93
76	Replicative DNA polymerase mutations in cancer. Current Opinion in Genetics and Development, 2014, 24, 107-113.	3.3	92
77	The effects of mutational processes and selection on driver mutations across cancer types. Nature Communications, 2018, 9, 1857.	12.8	91
78	Allelic imbalance at theLKB1 (STK11) locus in tumours from patients with Peutz-Jeghers' syndrome provides evidence for a hamartoma-(adenoma)-carcinoma sequence. , 1999, 188, 9-13.		85
79	Increased somatic mutation burdens in normal human cells due to defective DNA polymerases. Nature Genetics, 2021, 53, 1434-1442.	21.4	85
80	Differential clonal evolution in oesophageal cancers in response to neo-adjuvant chemotherapy. Nature Communications, 2016, 7, 11111.	12.8	83
81	Validation of Recently Proposed Colorectal Cancer Susceptibility Gene Variants in an Analysis of Families and Patientsâ€”a Systematic Review. Gastroenterology, 2017, 152, 75-77.e4.	1.3	80
82	Modifiable pathways for colorectal cancer: a mendelian randomisation analysis. The Lancet Gastroenterology and Hepatology, 2020, 5, 55-62.	8.1	79
83	Functional mechanisms underlying pleiotropic risk alleles at the 19p13.1 breastâ€“ovarian cancer susceptibility locus. Nature Communications, 2016, 7, 12675.	12.8	78
84	Five endometrial cancer risk loci identified through genome-wide association analysis. Nature Genetics, 2016, 48, 667-674.	21.4	77
85	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
86	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
87	BRCA2 Hypomorphic Missense Variants Confer Moderate Risks of Breast Cancer. Cancer Research, 2017, 77, 2789-2799.	0.9	75
88	Assessment of polygenic architecture and risk prediction based on common variants across fourteen cancers. Nature Communications, 2020, 11, 3353.	12.8	75
89	Loss of heterozygosity analysis: Practically and conceptually flawed?. Genes Chromosomes and Cancer, 2002, 34, 349-353.	2.8	74
90	Putative direct and indirect Wnt targets identified through consistent gene expression changes in APC-mutant intestinal adenomas from humans and mice. Human Molecular Genetics, 2008, 17, 3864-3875.	2.9	73

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91	Critical research gaps and recommendations to inform research prioritisation for more effective prevention and improved outcomes in colorectal cancer. Gut, 2018, 67, 179-193.	12.1	73
92	Multilevel genomics of colorectal cancers with microsatellite instabilityâ€"clinical impact of JAK1 mutations and consensus molecular subtype 1. Genome Medicine, 2017, 9, 46.	8.2	71
93	Somatic POLE exonuclease domain mutations are early events in sporadic endometrial and colorectal carcinogenesis, determining driver mutational landscape, clonal neoantigen burden and immune response. Journal of Pathology, 2018, 245, 283-296.	4.5	71
94	Tissue, cell and stage specificity of (epi)mutations in cancers. Nature Reviews Cancer, 2005, 5, 649-655.	28.4	67
95	Genetic modifiers of CHEK2*1100delC-associated breast cancer risk. Genetics in Medicine, 2017, 19, 599-603.	2.4	67
96	Chromatin organisation and cancer prognosis: a pan-cancer study. Lancet Oncology, The, 2018, 19, 356-369.	10.7	67
97	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.	2.5	64
98	CYP19A1 fine-mapping and Mendelian randomization: estradiol is causal for endometrial cancer. Endocrine-Related Cancer, 2016, 23, 77-91.	3.1	62
99	Genetic overlap between endometriosis and endometrial cancer: evidence from crossâ€disease genetic correlation and GWAS metaâ€analyses. Cancer Medicine, 2018, 7, 1978-1987.	2.8	62
100	Mutation burden and other molecular markers of prognosis in colorectal cancer treated with curative intent: results from the QUASAR 2 clinical trial and an Australian community-based series. The Lancet Gastroenterology and Hepatology, 2018, 3, 635-643.	8.1	60
101	Clinical applicability and cost of a 46-gene panel for genomic analysis of solid tumours: Retrospective validation and prospective audit in the UK National Health Service. PLoS Medicine, 2017, 14, e1002230.	8.4	60
102	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
103	A systematic review and meta-analysis of somatic and germline DNA sequence biomarkers of esophageal cancer survival, therapy response and stage. Annals of Oncology, 2015, 26, 624-644.	1.2	58
104	Robust RNA-based in situ mutation detection delineates colorectal cancer subclonal evolution. Nature Communications, 2017, 8, 1998.	12.8	57
105	In situ analysis ofLKB1/STK11 mRNA expression in human normal tissues and tumours. Journal of Pathology, 2000, 192, 203-206.	4.5	56
106	Identification of Novel Genetic Markers of Breast Cancer Survival. Journal of the National Cancer Institute, 2015, 107, .	6.3	56
107	Prognostic markers for colorectal cancer: estimating ploidy and stroma. Annals of Oncology, 2018, 29, 616-623.	1.2	56
108	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

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109	Management and 5-year outcomes in 9938 women with screen-detected ductal carcinoma in situ: the UK Sloane Project. European Journal of Cancer, 2018, 101, 210-219.	2.8	52
110	Genome-wide association study of germline variants and breast cancer-specific mortality. British Journal of Cancer, 2019, 120, 647-657.	6.4	52
111	Exploiting differential Wnt target gene expression to generate a molecular biomarker for colorectal cancer stratification. Gut, 2020, 69, 1092-1103.	12.1	52
112	Fineâ€scale mapping of 8q24 locus identifies multiple independent risk variants for breast cancer. International Journal of Cancer, 2016, 139, 1303-1317.	5.1	51
113	CRISPR-Cas9 Causes Chromosomal Instability and Rearrangements in Cancer Cell Lines, Detectable by Cytogenetic Methods. CRISPR Journal, 2019, 2, 406-416.	2.9	51
114	Pathology of Tumors Associated With Pathogenic Germline Variants in 9 Breast Cancer Susceptibility Genes. JAMA Oncology, 2022, 8, e216744.	7.1	51
115	Investigation of the atypicalFBXW7mutation spectrum in human tumours by conditional expression of a heterozygous propellor tip missense allele in the mouse intestines. Gut, 2014, 63, 792-799.	12.1	50
116	Fine-mapping of the HNF1B multicancer locus identifies candidate variants that mediate endometrial cancer risk. Human Molecular Genetics, 2015, 24, 1478-1492.	2.9	50
117	Adjuvant Treatment for POLE Proofreading Domainâ€“Mutant Cancers: Sensitivity to Radiotherapy, Chemotherapy, and Nucleoside Analogues. Clinical Cancer Research, 2018, 24, 3197-3203.	7.0	50
118	MicroRNA Related Polymorphisms and Breast Cancer Risk. PLoS ONE, 2014, 9, e109973.	2.5	49
119	A Polymorphic Enhancer near GREM1 Influences Bowel Cancer Risk through Differential CDX2 and TCF7L2 Binding. Cell Reports, 2014, 8, 983-990.	6.4	45
120	Body mass index and breast cancer survival: a Mendelian randomization analysis. International Journal of Epidemiology, 2017, 46, 1814-1822.	1.9	45
121	Homozygous PMS2 Deletion Causes a Severe Colorectal Cancer and Multiple Adenoma Phenotype Without Extraintestinal Cancer. Gastroenterology, 2007, 132, 527-530.	1.3	44
122	POLE mutations in families predisposed to cutaneous melanoma. Familial Cancer, 2015, 14, 621-628.	1.9	43
123	Genetic predisposition to ductal carcinoma in situ of the breast. Breast Cancer Research, 2016, 18, 22.	5.0	43
124	Much of the Genetic Risk of Colorectal Cancer Is Likely to Be Mediated Through Susceptibility to Adenomas. Gastroenterology, 2013, 144, 53-55.	1.3	41
125	Measuring single cell divisions in human tissues from multi-region sequencing data. Nature Communications, 2020, 11, 1035.	12.8	41
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	Genetic Predisposition to In Situ and Invasive Lobular Carcinoma of the Breast. PLoS Genetics, 2014, 10, e1004285.	3.5	39
128	Mendelian randomisation: A powerful and inexpensive method for identifying and excluding non-genetic risk factors for colorectal cancer. Molecular Aspects of Medicine, 2019, 69, 41-47.	6.4	39
129	Breast Cancer Polygenic Risk Score and Contralateral Breast Cancer Risk. American Journal of Human Genetics, 2020, 107, 837-848.	6.2	39
130	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
131	Germline variation in inflammation-related pathways and risk of Barrett's oesophagus and oesophageal adenocarcinoma. Gut, 2017, 66, 1739-1747.	12.1	38
132	FBXW7-mutated colorectal cancer cells exhibit aberrant expression of phosphorylated-p53 at Serine-15. Oncotarget, 2015, 6, 9240-9256.	1.8	38
133	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
134	Bone morphogenetic protein and Notch signalling crosstalk in poorâ€prognosis, mesenchymalâ€subtype colorectal cancer. Journal of Pathology, 2017, 242, 178-192.	4.5	36
135	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.	3.3	35
136	Mendelian randomization analyses suggest a role for cholesterol in the development of endometrial cancer. International Journal of Cancer, 2021, 148, 307-319.	5.1	35
137	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.	3.8	34
138	Premalignant SOX2 overexpression in the fallopian tubes of ovarian cancer patients: Discovery and validation studies. EBioMedicine, 2016, 10, 137-149.	6.1	34
139	Features of Patients With Hereditary Mixed Polyposis Syndrome Caused by Duplication of GREM1 and Implications for Screening and Surveillance. Gastroenterology, 2017, 152, 1876-1880.e1.	1.3	34
140	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
141	Oncogenic BRAF, unrestrained by TGFÎ²-receptor signalling, drives right-sided colonic tumorigenesis. Nature Communications, 2021, 12, 3464.	12.8	33
142	A Common Variant at the 14q32 Endometrial Cancer Risk Locus Activates AKT1 through YY1 Binding. American Journal of Human Genetics, 2016, 98, 1159-1169.	6.2	32
143	The HABP2 G534E Variant Is an Unlikely Cause of Familial Nonmedullary Thyroid Cancer. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 1098-1103.	3.6	32
144	Germline and Somatic Genetic Variants in the p53 Pathway Interact to Affect Cancer Risk, Progression, and Drug Response. Cancer Research, 2021, 81, 1667-1680.	0.9	32

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145	A sensitive and affordable multiplex RT-qPCR assay for SARS-CoV-2 detection. PLoS Biology, 2020, 18, e3001030.	5.6	32
146	Association analysis using next-generation sequence data from publicly available control groups: the robust variance score statistic. Bioinformatics, 2014, 30, 2179-2188.	4.1	31
147	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
148	The clinical features of polymerase proof-reading associated polyposis (PPAP) and recommendations for patient management. Familial Cancer, 2022, 21, 197-209.	1.9	31
149	Target gene mutational pattern in Lynch syndrome colorectal carcinomas according to tumour location and germline mutation. British Journal of Cancer, 2015, 113, 686-692.	6.4	30
150	A network analysis to identify mediators of germline-driven differences in breast cancer prognosis. Nature Communications, 2020, 11, 312.	12.8	30
151	Investigation of the effects of DNA repair gene polymorphisms on the risk of colorectal cancer. Mutagenesis, 2012, 27, 219-223.	2.6	29
152	'Toxgnostics': an unmet need in cancer medicine. Nature Reviews Cancer, 2014, 14, 440-445.	28.4	29
153	Serum- and Glucocorticoid-induced Kinase Sgk1 Directly Promotes the Differentiation of Colorectal Cancer Cells and Restrains Metastasis. Clinical Cancer Research, 2019, 25, 629-640.	7.0	28
154	The use of Mendelian randomisation to identify causal cancer risk factors: promise and limitations. Journal of Pathology, 2020, 250, 541-554.	4.5	28
155	Genetic Biomarkers of Barrettâ€™s Esophagus Susceptibility and Progression to Dysplasia and Cancer: A Systematic Review and Meta-Analysis. Digestive Diseases and Sciences, 2016, 61, 25-38.	2.3	27
156	Common germline polymorphisms associated with breast cancer-specific survival. Breast Cancer Research, 2015, 17, 58.	5.0	26
157	RAD51B in Familial Breast Cancer. PLoS ONE, 2016, 11, e0153788.	2.5	26
158	Comprehensive genetic assessment of the ESR1 locus identifies a risk region for endometrial cancer. Endocrine-Related Cancer, 2015, 22, 851-861.	3.1	25
159	FBXW7 mutations typically found in human cancers are distinct from null alleles and disrupt lung development. Journal of Pathology, 2011, 224, 180-189.	4.5	24
160	Recurrent Coding Sequence Variation Explains Only A Small Fraction of the Genetic Architecture of Colorectal Cancer. Scientific Reports, 2015, 5, 16286.	3.3	24
161	Analyses of 7,635 Patients with Colorectal Cancer Using Independent Training and Validation Cohorts Show That rs9929218 in CDH1 Is a Prognostic Marker of Survival. Clinical Cancer Research, 2015, 21, 3453-3461.	7.0	24
162	Fine-Scale Mapping of the 4q24 Locus Identifies Two Independent Loci Associated with Breast Cancer Risk. Cancer Epidemiology Biomarkers and Prevention, 2015, 24, 1680-1691.	2.5	24

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163	Germline MBD4 deficiency causes a multi-tumor predisposition syndrome. American Journal of Human Genetics, 2022, 109, 953-960.	6.2	23
164	Association of genetic susceptibility variants for type 2 diabetes with breast cancer risk in women of European ancestry. Cancer Causes and Control, 2016, 27, 679-693.	1.8	21
165	Systematic meta-analyses and field synopsis of genetic association studies in colorectal adenomas. International Journal of Epidemiology, 2016, 45, 186-205.	1.9	21
166	Clinically actionable mutation profiles in patients with cancer identified by whole-genome sequencing. Journal of Physical Education and Sports Management, 2018, 4, a002279.	1.2	21
167	Urgent improvements needed to diagnose and manage Lynch syndrome. BMJ: British Medical Journal, 2017, 356, j1388.	2.3	20
168	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.	3.3	19
169	GWAS meta-analysis of 16 852 women identifies new susceptibility locus for endometrial cancer. Human Molecular Genetics, 2016, 25, ddw092.	2.9	19
170	The BRCA2 c.68-7TÂ>ÂA variant is not pathogenic: A model for clinical calibration of spliceogenicity. Human Mutation, 2018, 39, 729-741.	2.5	19
171	Breast Cancer Risk Factors and Survival by Tumor Subtype: Pooled Analyses from the Breast Cancer Association Consortium. Cancer Epidemiology Biomarkers and Prevention, 2021, 30, 623-642.	2.5	19
172	Genetic analyses of gynecological disease identify genetic relationships between uterine fibroids and endometrial cancer, and a novel endometrial cancer genetic risk region at the WNT4 1p36.12 locus. Human Genetics, 2021, 140, 1353-1365.	3.8	18
173	GREM1 germline mutation screening in Ashkenazi Jewish patients with familial colorectal cancer. Genetical Research, 2015, 97, e11.	0.9	17
174	Common colorectal cancer risk alleles contribute to the multiple colorectal adenoma phenotype, but do not influence colonic polyposis in FAP. European Journal of Human Genetics, 2015, 23, 260-263.	2.8	17
175	Histological phenotypic subtypes predict recurrence risk and response to adjuvant chemotherapy in patients with stage III colorectal cancer. Journal of Pathology: Clinical Research, 2020, 6, 283-296.	3.0	17
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