William D Foulkes

List of Publications by Citations

610 papers	35,929 citations	97 h-index	170 g-index
673 ext. papers	41,779 ext. citations	9.3 avg, IF	7.14 L-index

#	Paper	IF	Citations
610	Triple-negative breast cancer. *New England Journal of Medicine*, **2010**, 363, 1938-48	59.2	2458
609	Germline BRCA1 mutations and a basal epithelial phenotype in breast cancer. *Journal of the National Cancer Institute*, **2003**, 95, 1482-5	9.7	737
608	REVEL: An Ensemble Method for Predicting the Pathogenicity of Rare Missense Variants. *American Journal of Human Genetics*, **2016**, 99, 877-885	11	722
607	BRCA1 and BRCA2: 1994 and beyond. *Nature Reviews Cancer*, **2004**, 4, 665-76	31.3	694
606	Sequence variant classification and reporting: recommendations for improving the interpretation of cancer susceptibility genetic test results. *Human Mutation*, **2008**, 29, 1282-91	4.7	622
605	Subtyping of breast cancer by immunohistochemistry to investigate a relationship between subtype and short and long term survival: a collaborative analysis of data for 10,159 cases from 12 studies. *PLoS Medicine*, **2010**, 7, e1000279	11.6	616
604	Gene-panel sequencing and the prediction of breast-cancer risk. *New England Journal of Medicine*, **2015**, 372, 2243-57	59.2	587
603	Breast-cancer risk in families with mutations in PALB2. *New England Journal of Medicine*, **2014**, 371, 497-506	59.2	576
602	Contralateral breast cancer in BRCA1 and BRCA2 mutation carriers. *Journal of Clinical Oncology*, **2004**, 22, 2328-35	2.2	514
601	Salpingo-oophorectomy and the risk of ovarian, fallopian tube, and peritoneal cancers in women with a BRCA1 or BRCA2 Mutation. *JAMA - Journal of the American Medical Association*, **2006**, 296, 185-92	27.4	466
600	Familial adenomatous polyposis. *American Journal of Gastroenterology*, **2006**, 101, 385-98	0.7	456
599	Impact of oophorectomy on cancer incidence and mortality in women with a BRCA1 or BRCA2 mutation. *Journal of Clinical Oncology*, **2014**, 32, 1547-53	2.2	397
598	Inherited susceptibility to common cancers. *New England Journal of Medicine*, **2008**, 359, 2143-53	59.2	379
597	Triple-negative breast cancer: distinguishing between basal and nonbasal subtypes. *Clinical Cancer Research*, **2009**, 15, 2302-10	12.9	371
596	Identification of seven new prostate cancer susceptibility loci through a genome-wide association study. *Nature Genetics*, **2009**, 41, 1116-21	36.3	360
595	Breast cancer risk following bilateral oophorectomy in BRCA1 and BRCA2 mutation carriers: an international case-control study. *Journal of Clinical Oncology*, **2005**, 23, 7491-6	2.2	360
594	Is breast cancer the same disease in Asian and Western countries?. *World Journal of Surgery*, **2010**, 34, 2308-24	3.3	352

593	Triple-negative breast cancer: risk factors to potential targets. *Clinical Cancer Research*, **2008**, 14, 8010-8	12.9	336
592	Recurrent somatic DICER1 mutations in nonepithelial ovarian cancers. *New England Journal of Medicine*, **2012**, 366, 234-42	59.2	332
591	DICER1: mutations, microRNAs and mechanisms. *Nature Reviews Cancer*, **2014**, 14, 662-72	31.3	308
590	The prognostic implication of the basal-like (cyclin E high/p27 low/p53+/glomeruloid-microvascular-proliferation+) phenotype of BRCA1-related breast cancer. *Cancer Research*, **2004**, 64, 830-5	10.1	307
589	Germline and somatic SMARCA4 mutations characterize small cell carcinoma of the ovary, hypercalcemic type. *Nature Genetics*, **2014**, 46, 438-43	36.3	305
588	CD8+ lymphocyte infiltration is an independent favorable prognostic indicator in basal-like breast cancer. *Breast Cancer Research*, **2012**, 14, R48	8.3	290
587	Intrinsic breast tumor subtypes, race, and long-term survival in the Carolina Breast Cancer Study. *Clinical Cancer Research*, **2010**, 16, 6100-10	12.9	286
586	Use of immunohistochemical markers can refine prognosis in triple negative breast cancer. *BMC Cancer*, **2007**, 7, 134	4.8	284
585	International variation in rates of uptake of preventive options in BRCA1 and BRCA2 mutation carriers. *International Journal of Cancer*, **2008**, 122, 2017-22	7.5	268
584	Oral contraceptives and the risk of breast cancer in BRCA1 and BRCA2 mutation carriers. *Journal of the National Cancer Institute*, **2002**, 94, 1773-9	9.7	266
583	A mutational signature reveals alterations underlying deficient homologous recombination repair in breast cancer. *Nature Genetics*, **2017**, 49, 1476-1486	36.3	255
582	Frequency of recurrent BRCA1 and BRCA2 mutations in Ashkenazi Jewish breast cancer families. *Nature Medicine*, **1996**, 2, 1179-83	50.5	253
581	Estrogen receptor status in BRCA1- and BRCA2-related breast cancer: the influence of age, grade, and histological type. *Clinical Cancer Research*, **2004**, 10, 2029-34	12.9	234
580	Hereditary breast cancer: new genetic developments, new therapeutic avenues. *Human Genetics*, **2008**, 124, 31-42	6.3	233
579	A combined analysis of outcome following breast cancer: differences in survival based on BRCA1/BRCA2 mutation status and administration of adjuvant treatment. *Breast Cancer Research*, **2004**, 6, R8-R17	8.3	225
578	DICER1 mutations in familial multinodular goiter with and without ovarian Sertoli-Leydig cell tumors. *JAMA - Journal of the American Medical Association*, **2011**, 305, 68-77	27.4	222
577	Tamoxifen and contralateral breast cancer in BRCA1 and BRCA2 carriers: an update. *International Journal of Cancer*, **2006**, 118, 2281-4	7.5	220
576	Prospective study of breast cancer incidence in women with a BRCA1 or BRCA2 mutation under surveillance with and without magnetic resonance imaging. *Journal of Clinical Oncology*, **2011**, 29, 1664-9	2.2	199

575	Identification of genes associated with head and neck carcinogenesis by cDNA microarray comparison between matched primary normal epithelial and squamous carcinoma cells. *Oncogene*, **2002**, 21, 2634-40	9.2	194
574	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. *Nature Genetics*, **2017**, 49, 680-691	36.3	190
573	Tumor necrosis factor and its receptors in human ovarian cancer. Potential role in disease progression. *Journal of Clinical Investigation*, **1993**, 91, 2194-206	15.9	187
572	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. *Nature Genetics*, **2017**, 49, 1767-1778	36.3	186
571	Reproductive risk factors for ovarian cancer in carriers of BRCA1 or BRCA2 mutations: a case-control study. *Lancet Oncology, The*, **2007**, 8, 26-34	21.7	186
570	Contralateral mastectomy and survival after breast cancer in carriers of BRCA1 and BRCA2 mutations: retrospective analysis. *BMJ, The*, **2014**, 348, g226	5.9	181
569	The incidence of pancreatic cancer in BRCA1 and BRCA2 mutation carriers. *British Journal of Cancer*, **2012**, 107, 2005-9	8.7	179
568	Familial nontoxic multinodular thyroid goiter locus maps to chromosome 14q but does not account for familial nonmedullary thyroid cancer. *American Journal of Human Genetics*, **1997**, 61, 1123-30	11	178
567	Pregnancy and risk of early breast cancer in carriers of BRCA1 and BRCA2. *Lancet, The*, **1999**, 354, 1846-50	40	175
566	The CDKN2A (p16) Gene and Human Cancer. *Molecular Medicine*, **1997**, 3, 5-20	6.2	174
565	Analysis of PALB2/FANCN-associated breast cancer families. *Proceedings of the National Academy of Sciences of the United States of America*, **2007**, 104, 6788-93	11.5	173
564	Predictors of contralateral breast cancer in BRCA1 and BRCA2 mutation carriers. *British Journal of Cancer*, **2011**, 104, 1384-92	8.7	167
563	Pituitary blastoma: a pathognomonic feature of germ-line DICER1 mutations. *Acta Neuropathologica*, **2014**, 128, 111-22	14.3	160
562	A basal epithelial phenotype is more frequent in interval breast cancers compared with screen detected tumors. *Cancer Epidemiology Biomarkers and Prevention*, **2005**, 14, 1108-12	4	159
561	Targeted prostate cancer screening in BRCA1 and BRCA2 mutation carriers: results from the initial screening round of the IMPACT study. *European Urology*, **2014**, 66, 489-99	10.2	156
560	Point Mutations in Exon 1B of APC Reveal Gastric Adenocarcinoma and Proximal Polyposis of the Stomach as a Familial Adenomatous Polyposis Variant. *American Journal of Human Genetics*, **2016**, 98, 830-842	11	153
559	Extending the phenotypes associated with DICER1 mutations. *Human Mutation*, **2011**, 32, 1381-4	4.7	144
558	Influence of BRCA1 mutations on nuclear grade and estrogen receptor status of breast carcinoma in Ashkenazi Jewish women. *Cancer*, **1997**, 80, 435-41	6.4	144

557	Placental cadherin and the basal epithelial phenotype of BRCA1-related breast cancer. *Clinical Cancer Research*, **2005**, 11, 4003-11	12.9	143
556	Predictors of contralateral prophylactic mastectomy in women with a BRCA1 or BRCA2 mutation: the Hereditary Breast Cancer Clinical Study Group. *Journal of Clinical Oncology*, **2008**, 26, 1093-7	2.2	141
555	Prognostic significance of FOXP3+ tumor-infiltrating lymphocytes in breast cancer depends on estrogen receptor and human epidermal growth factor receptor-2 expression status and concurrent cytotoxic T-cell infiltration. *Breast Cancer Research*, **2014**, 16, 432	8.3	137
554	Breast-feeding and the risk of breast cancer in BRCA1 and BRCA2 mutation carriers. *Journal of the National Cancer Institute*, **2004**, 96, 1094-8	9.7	137
553	Founder BRCA1 and BRCA2 mutations in French Canadian breast and ovarian cancer families. *American Journal of Human Genetics*, **1998**, 63, 1341-51	11	136
552	HOXB13 is a susceptibility gene for prostate cancer: results from the International Consortium for Prostate Cancer Genetics (ICPCG). *Human Genetics*, **2013**, 132, 5-14	6.3	134
551	Multiple novel prostate cancer predisposition loci confirmed by an international study: the PRACTICAL Consortium. *Cancer Epidemiology Biomarkers and Prevention*, **2008**, 17, 2052-61	4	134
550	TP53 mutations in breast cancer associated with BRCA1 or BRCA2 germ-line mutations: distinctive spectrum and structural distribution. *Cancer Research*, **2001**, 61, 4092-7	10.1	134
549	Cancer Risks Associated With Germline Pathogenic Variants: An International Study of 524 Families. *Journal of Clinical Oncology*, **2020**, 38, 674-685	2.2	133
548	Effect of pregnancy as a risk factor for breast cancer in BRCA1/BRCA2 mutation carriers. *International Journal of Cancer*, **2005**, 117, 988-91	7.5	131
547	PREDICT Plus: development and validation of a prognostic model for early breast cancer that includes HER2. *British Journal of Cancer*, **2012**, 107, 800-7	8.7	130
546	A combined genomewide linkage scan of 1,233 families for prostate cancer-susceptibility genes conducted by the international consortium for prostate cancer genetics. *American Journal of Human Genetics*, **2005**, 77, 219-29	11	129
545	Germline RECQL mutations are associated with breast cancer susceptibility. *Nature Genetics*, **2015**, 47, 643-6	36.3	123
544	BRCA1 functions as a breast stem cell regulator. *Journal of Medical Genetics*, **2004**, 41, 1-5	5.8	122
543	A significant response to neoadjuvant chemotherapy in BRCA1/2 related breast cancer. *Journal of Medical Genetics*, **2002**, 39, 608-10	5.8	122
542	Analysis of the gene coding for the BRCA2-interacting protein PALB2 in familial and sporadic pancreatic cancer. *Gastroenterology*, **2009**, 137, 1183-6	13.3	121
541	PALB2, CHEK2 and ATM rare variants and cancer risk: data from COGS. *Journal of Medical Genetics*, **2016**, 53, 800-811	5.8	121
540	Extensive DNA methylation in normal colorectal mucosa in hyperplastic polyposis. *Gut*, **2006**, 55, 1467-74	9.2	120

539	A somatic BRCA1 mutation in an ovarian tumour. *Nature Genetics*, **1995**, 9, 343-4	36.3	120
538	The risk of ovarian cancer after breast cancer in BRCA1 and BRCA2 carriers. *Gynecologic Oncology*, **2005**, 96, 222-6	4.9	118
537	Increased risk for nonmedullary thyroid cancer in the first degree relatives of prevalent cases of nonmedullary thyroid cancer: a hospital-based study. *Journal of Clinical Endocrinology and Metabolism*, **2001**, 86, 5307-12	5.6	118
536	Retinoblastoma and Neuroblastoma Predisposition and Surveillance. *Clinical Cancer Research*, **2017**, 23, e98-e106	12.9	117
535	SMARCA4-mutated atypical teratoid/rhabdoid tumors are associated with inherited germline alterations and poor prognosis. *Acta Neuropathologica*, **2014**, 128, 453-6	14.3	116
534	BRCA1 and BRCA2: chemosensitivity, treatment outcomes and prognosis. *Familial Cancer*, **2006**, 5, 135-42	3	114
533	Clinico-pathological characteristics of BRCA1- and BRCA2-related breast cancer. *Journal of Surgical Oncology*, **2000**, 18, 287-95		114
532	Linkage analysis of chromosome 1q markers in 136 prostate cancer families. The Cancer Research Campaign/British Prostate Group U.K. Familial Prostate Cancer Study Collaborators. *American Journal of Human Genetics*, **1998**, 62, 653-8	11	112
531	Rapid progression of prostate cancer in men with a BRCA2 mutation. *British Journal of Cancer*, **2008**, 99, 371-4	8.7	112
530	Identification of a novel truncating PALB2 mutation and analysis of its contribution to early-onset breast cancer in French-Canadian women. *Breast Cancer Research*, **2007**, 9, R83	8.3	110
529	The founder mutation MSH2*1906G-->C is an important cause of hereditary nonpolyposis colorectal cancer in the Ashkenazi Jewish population. *American Journal of Human Genetics*, **2002**, 71, 1395-412	11	109
528	Germline BRCA1/2 mutations and p27(Kip1) protein levels independently predict outcome after breast cancer. *Journal of Clinical Oncology*, **2000**, 18, 4045-52	2.2	108
527	Familial risks of squamous cell carcinoma of the head and neck: retrospective case-control study. *BMJ: British Medical Journal*, **1996**, 313, 716-21		107
526	Biallelic deleterious BRCA1 mutations in a woman with early-onset ovarian cancer. *Cancer Discovery*, **2013**, 3, 399-405	24.4	106
525	Germline and somatic FGFR1 abnormalities in dysembryoplastic neuroepithelial tumors. *Acta Neuropathologica*, **2016**, 131, 847-63	14.3	105
524	Molecular characterization of the spectrum of genomic deletions in the mismatch repair genes MSH2, MLH1, MSH6, and PMS2 responsible for hereditary nonpolyposis colorectal cancer (HNPCC). *Genes Chromosomes and Cancer*, **2005**, 44, 123-38	5	105
523	CARD9 deficiency and spontaneous central nervous system candidiasis: complete clinical remission with GM-CSF therapy. *Clinical Infectious Diseases*, **2014**, 59, 81-4	11.6	104
522	Effect of smoking on breast cancer in carriers of mutant BRCA1 or BRCA2 genes. *Journal of the National Cancer Institute*, **1998**, 90, 761-6	9.7	104

521	Germ-line and somatic DICER1 mutations in pineoblastoma. *Acta Neuropathologica*, **2014**, 128, 583-95	14.3	103
520	Loss of heterozygosity and amplification on chromosome 11q in human ovarian cancer. *British Journal of Cancer*, **1993**, 67, 268-73	8.7	103
519	No small surprise´-´small cell carcinoma of the ovary, hypercalcaemic type, is a malignant rhabdoid tumour. *Journal of Pathology*, **2014**, 233, 209-14	9.4	101
518	Bilateral Oophorectomy and Breast Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. *Journal of the National Cancer Institute*, **2017**, 109,	9.7	99
517	Frequent loss of heterozygosity on chromosome 6 in human ovarian carcinoma. *British Journal of Cancer*, **1993**, 67, 551-9	8.7	99
516	Biallelic DICER1 mutations occur in Wilms tumours. *Journal of Pathology*, **2013**, 230, 154-64	9.4	98
515	Cancer Surveillance in Gorlin Syndrome and Rhabdoid Tumor Predisposition Syndrome. *Clinical Cancer Research*, **2017**, 23, e62-e67	12.9	97
514	, and Their Associated Tumor Susceptibility Syndromes: Clinical Features, Genetics, and Surveillance Recommendations in Childhood. *Clinical Cancer Research*, **2017**, 23, e76-e82	12.9	97
513	Incidence of colorectal cancer in BRCA1 and BRCA2 mutation carriers: results from a follow-up study. *British Journal of Cancer*, **2014**, 110, 530-4	8.7	95
512	Germline DICER1 mutations and familial cystic nephroma. *Journal of Medical Genetics*, **2010**, 47, 863-6	5.8	94
511	DICER1 hotspot mutations in non-epithelial gonadal tumours. *British Journal of Cancer*, **2013**, 109, 2744-50	8.7	93
510	E6/E7 proteins of HPV type 16 and ErbB-2 cooperate to induce neoplastic transformation of primary normal oral epithelial cells. *Oncogene*, **2004**, 23, 350-8	9.2	92
509	Disruption of the expected positive correlation between breast tumor size and lymph node status in BRCA1-related breast carcinoma. *Cancer*, **2003**, 98, 1569-77	6.4	92
508	A novel gene encoding a B-box protein within the BRCA1 region at 17q21.1. *Human Molecular Genetics*, **1994**, 3, 589-94	5.6	92
507	Methionine Metabolism Shapes T Helper Cell Responses through Regulation of Epigenetic Reprogramming. *Cell Metabolism*, **2020**, 31, 250-266.e9	24.6	91
506	Frequency of premature menopause in women who carry a BRCA1 or BRCA2 mutation. *Fertility and Sterility*, **2013**, 99, 1724-8	4.8	90
505	BRCA Challenge: BRCA Exchange as a global resource for variants in BRCA1 and BRCA2. *PLoS Genetics*, **2018**, 14, e1007752	6	90
504	A PALB2 mutation associated with high risk of breast cancer. *Breast Cancer Research*, **2010**, 12, R109	8.3	89

503	The histomorphology of Lynch syndrome-associated ovarian carcinomas: toward a subtype-specific screening strategy. *American Journal of Surgical Pathology*, **2014**, 38, 1173-81	6.7	87
502	Exploring the association Between DICER1 mutations and differentiated thyroid carcinoma. *Journal of Clinical Endocrinology and Metabolism*, **2014**, 99, E1072-7	5.6	86
501	Pheochromocytoma and paraganglioma syndromes: genetics and management update. *Current Oncology*, **2014**, 21, e8-e17	2.8	85
500	Hormone Replacement Therapy After Oophorectomy and Breast Cancer Risk Among BRCA1 Mutation Carriers. *JAMA Oncology*, **2018**, 4, 1059-1065	13.4	84
499	Prognostic significance of CD8+ T lymphocytes in breast cancer depends upon both oestrogen receptor status and histological grade. *Histopathology*, **2011**, 58, 1107-16	7.3	84
498	The incidence of endometrial cancer in women with BRCA1 and BRCA2 mutations: an international prospective cohort study. *Gynecologic Oncology*, **2013**, 130, 127-31	4.9	83
497	Are triple-negative tumours and basal-like breast cancer synonymous?. *Breast Cancer Research*, **2007**, 9, 404; author reply 405	8.3	83
496	Women's attitudes toward preventive strategies for hereditary breast or ovarian carcinoma differ from one country to another: differences among English, French, and Canadian women. *Cancer*, **2001**, 92, 959-68	6.4	83
495	Family history of cancer is a risk factor for squamous cell carcinoma of the head and neck in Brazil: a case-control study. *International Journal of Cancer*, **1995**, 63, 769-73	7.5	83
494	Tumor size and survival in breast cancer--a reappraisal. *Nature Reviews Clinical Oncology*, **2010**, 7, 348-53	19.4	82
493	Effect of Oophorectomy on Survival After Breast Cancer in BRCA1 and BRCA2 Mutation Carriers. *JAMA Oncology*, **2015**, 1, 306-13	13.4	81
492	Changes in body weight and the risk of breast cancer in BRCA1 and BRCA2 mutation carriers. *Breast Cancer Research*, **2005**, 7, R833-43	8.3	80
491	Screening mammography and risk of breast cancer in BRCA1 and BRCA2 mutation carriers: a case-control study. *Lancet Oncology, The*, **2006**, 7, 402-6	21.7	80
490	Very frequent loss of heterozygosity throughout chromosome 17 in sporadic ovarian carcinoma. *International Journal of Cancer*, **1993**, 54, 220-5	7.5	80
489	On the origin and diffusion of BRCA1 c.5266dupC (5382insC) in European populations. *European Journal of Human Genetics*, **2011**, 19, 300-6	5.3	79
488	Impact of germline BRCA1 mutations and overexpression of p53 on prognosis and response to treatment following breast carcinoma: 10-year follow up data. *Cancer*, **2003**, 97, 527-36	6.4	79
487	Interim Results from the IMPACT Study: Evidence for Prostate-specific Antigen Screening in BRCA2 Mutation Carriers. *European Urology*, **2019**, 76, 831-842	10.2	78
486	An evaluation of needs of female BRCA1 and BRCA2 carriers undergoing genetic counselling. *Journal of Medical Genetics*, **2000**, 37, 866-74	5.8	78

485	RARE-17. SURVIVAL BENEFIT FOR INDIVIDUALS WITH CONSTITUTIONAL MISMATCH REPAIR DEFICIENCY SYNDROME AND BRAIN TUMORS WHO UNDERGO SURVEILLANCE PROTOCOL. A REPORT FROM THE INTERNATIONAL REPLICATION REPAIR CONSORTIUM. *Neuro-Oncology*, **2020**, 22, iii445-iii446	1	78
484	RARE-22. GERMLINE PATHOGENIC VARIANT c.1552G>A;p.E518K IN DGCR8 CONFERS SUSCEPTIBILITY FOR SCHWANNOMATOSIS AND THYROID TUMORS. *Neuro-Oncology*, **2020**, 22, iii447-iii447	1	78
483	Germ-line BRCA1 mutation is an adverse prognostic factor in Ashkenazi Jewish women with breast cancer. *Clinical Cancer Research*, **1997**, 3, 2465-9	12.9	76
482	Family history of cancer and cancer risks in women with BRCA1 or BRCA2 mutations. *Journal of the National Cancer Institute*, **2010**, 102, 1874-8	9.7	74
481	Germline HAVCR2 mutations altering TIM-3 characterize subcutaneous panniculitis-like T cell lymphomas with hemophagocytic lymphohistiocytic syndrome. *Nature Genetics*, **2018**, 50, 1650-1657	36.3	74
480	Primary node negative breast cancer in BRCA1 mutation carriers has a poor outcome. *Annals of Oncology*, **2000**, 11, 307-13	10.3	73
479	SMARCA4 loss is synthetic lethal with CDK4/6 inhibition in non-small cell lung cancer. *Nature Communications*, **2019**, 10, 557	17.4	72
478	Mutational Signature Analysis Reveals NTHL1 Deficiency to Cause a Multi-tumor Phenotype. *Cancer Cell*, **2019**, 35, 256-266.e5	24.3	72
477	Tumor size is an unreliable predictor of prognosis in basal-like breast cancers and does not correlate closely with lymph node status. *Breast Cancer Research and Treatment*, **2009**, 117, 199-204	4.4	69
476	Breastfeeding and the risk of breast cancer in BRCA1 and BRCA2 mutation carriers. *Breast Cancer Research*, **2012**, 14, R42	8.3	68
475	Prognostic importance of glomeruloid microvascular proliferation indicates an aggressive angiogenic phenotype in human cancers. *Cancer Research*, **2002**, 62, 6808-11	10.1	68
474	Population genetic testing for cancer susceptibility: founder mutations to genomes. *Nature Reviews Clinical Oncology*, **2016**, 13, 41-54	19.4	67
473	Risk of ipsilateral breast cancer in BRCA1 and BRCA2 mutation carriers. *Breast Cancer Research and Treatment*, **2011**, 127, 287-96	4.4	66
472	E6/E7 of HPV type 16 promotes cell invasion and metastasis of human breast cancer cells. *Cell Cycle* , **2007**, 6, 2038-42	4.7	66
471	In brief: BRCA1 and BRCA2. *Journal of Pathology*, **2013**, 230, 347-9	9.4	65
470	Homozygous BUB1B mutation and susceptibility to gastrointestinal neoplasia. *New England Journal of Medicine*, **2010**, 363, 2628-37	59.2	65
469	Allele loss and mutation screen at the Peutz-Jeghers (LKB1) locus (19p13.3) in sporadic ovarian tumours. *British Journal of Cancer*, **1999**, 80, 70-2	8.7	65
468	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. *Nature Communications*, **2016**, 7, 11375	17.4	64

467	Age at menarche and the risk of breast cancer in BRCA1 and BRCA2 mutation carriers. *Cancer Causes and Control*, **2005**, 16, 667-74	2.8	64
466	Rare germline mutations in PALB2 and breast cancer risk: a population-based study. *Human Mutation*, **2012**, 33, 674-80	4.7	63
465	Infertility, treatment of infertility, and the risk of breast cancer among women with BRCA1 and BRCA2 mutations: a case-control study. *Cancer Causes and Control*, **2008**, 19, 1111-9	2.8	63
464	Coffee consumption and breast cancer risk among BRCA1 and BRCA2 mutation carriers. *International Journal of Cancer*, **2006**, 118, 103-7	7.5	63
463	Cancer Screening Recommendations and Clinical Management of Inherited Gastrointestinal Cancer Syndromes in Childhood. *Clinical Cancer Research*, **2017**, 23, e107-e114	12.9	62
462	Biallelic NTHL1 Mutations in a Woman with Multiple Primary Tumors. *New England Journal of Medicine*, **2015**, 373, 1985-6	59.2	62
461	Breast, ovarian, and endometrial malignancies in systemic lupus erythematosus: a meta-analysis. *British Journal of Cancer*, **2011**, 104, 1478-81	8.7	62
460	The influence of clinical and genetic factors on patient outcome in small cell carcinoma of the ovary, hypercalcemic type. *Gynecologic Oncology*, **2016**, 141, 454-460	4.9	61
459	DICER1 Mutations Are Consistently Present in Moderately and Poorly Differentiated Sertoli-Leydig Cell Tumors. *American Journal of Surgical Pathology*, **2017**, 41, 1178-1187	6.7	60
458	Risk of pancreatic cancer among individuals with a family history of cancer of the pancreas. *International Journal of Cancer*, **2002**, 97, 807-10	7.5	60
457	The APCI1307K allele and breast cancer risk. *Nature Genetics*, **1998**, 20, 13-4	36.3	59
456	Pancreatic adenocarcinoma: epidemiology and genetics. *Journal of Medical Genetics*, **1996**, 33, 889-98	5.8	59
455	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. *Nature Genetics*, **2020**, 52, 56-73	36.3	56
454	Oncogenic role of PDK4 in human colon cancer cells. *British Journal of Cancer*, **2017**, 116, 930-936	8.7	55
453	Pooled genome linkage scan of aggressive prostate cancer: results from the International Consortium for Prostate Cancer Genetics. *Human Genetics*, **2006**, 120, 471-85	6.3	55
452	miRNA processing and human cancer: DICER1 cuts the mustard. *Science Translational Medicine*, **2011**, 3, 111ps46	17.5	54
451	Functional mechanisms underlying pleiotropic risk alleles at the 19p13.1 breast-ovarian cancer susceptibility locus. *Nature Communications*, **2016**, 7, 12675	17.4	53
450	Risk factors for familial and sporadic ovarian cancer among French Canadians: a case-control study. *American Journal of Obstetrics and Gynecology*, **1998**, 179, 403-10	6.4	53

449	Immunohistochemical expression of DNA repair proteins in familial breast cancer differentiate BRCA2-associated tumors. *Journal of Clinical Oncology*, **2005**, 23, 7503-11	2.2	53
448	Genetic mechanisms in squamous cell carcinoma of the head and neck. *Oral Oncology*, **2001**, 37, 115-26	4.4	53
447	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	4.7	52
446	Breast-cancer risk in families with mutations in PALB2. *New England Journal of Medicine*, **2014**, 371, 1651-2	59.2	52
445	Inherited mutations in breast cancer genes--risk and response. *Journal of Mammary Gland Biology and Neoplasia*, **2011**, 16, 3-15	2.4	52
444	Meta-analysis and pooled analysis of GSTM1 and CYP1A1 polymorphisms and oral and pharyngeal cancers: a HuGE-GSEC review. *Genetics in Medicine*, **2008**, 10, 369-84	8.1	52
443	The CDKN2A (p16) gene and human cancer. *Molecular Medicine*, **1997**, 3, 5-20	6.2	52
442	High-sensitivity sequencing reveals multi-organ somatic mosaicism causing DICER1 syndrome. *Journal of Medical Genetics*, **2016**, 53, 43-52	5.8	51
441	Compromised BRCA1-PALB2 interaction is associated with breast cancer risk. *Oncogene*, **2017**, 36, 4161-4170	5.7	51
440	Ovarian and Breast Cancer Risks Associated With Pathogenic Variants in RAD51C and RAD51D. *Journal of the National Cancer Institute*, **2020**, 112, 1242-1250	9.7	51
439	Tumor characteristics as an analytic tool for classifying genetic variants of uncertain clinical significance. *Human Mutation*, **2008**, 29, 1292-303	4.7	51
438	Impaired RASGRF1/ERK-mediated GM-CSF response characterizes CARD9 deficiency in French-Canadians. *Journal of Allergy and Clinical Immunology*, **2016**, 137, 1178-1188.e7	11.5	49
437	Familial rhabdoid tumour Mavant la lettreM-from pathology review to exome sequencing and back again. *Journal of Pathology*, **2013**, 231, 35-43	9.4	49
436	Diabetes and breast cancer among women with BRCA1 and BRCA2 mutations. *Cancer*, **2011**, 117, 1812-8	6.4	49
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