William D Foulkes

List of Publications by Year
in descending order

649 papers	46,524 citations	1530 106 h-index	2883 190 g-index
674 all docs	674 docs citations	674 times ranked	42207 citing authors

#	Article	IF	Citations
1	Triple-Negative Breast Cancer. New England Journal of Medicine, 2010, 363, 1938-1948.	13.9	3,233
2	REVEL: An Ensemble Method for Predicting the Pathogenicity of Rare Missense Variants. American Journal of Human Genetics, 2016, 99, 877-885.	2.6	1,555
3	BRCA1 and BRCA2: 1994 and beyond. Nature Reviews Cancer, 2004, 4, 665-676.	12.8	824
4	Germline BRCA1 Mutations and a Basal Epithelial Phenotype in Breast Cancer. Journal of the National Cancer Institute, 2003, 95, 1482-1485.	3.0	807
5	Sequence variant classification and reporting: recommendations for improving the interpretation of cancer susceptibility genetic test results. Human Mutation, 2008, 29, 1282-1291.	1.1	782
6	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.	3.9	764
7	Gene-Panel Sequencing and the Prediction of Breast-Cancer Risk. New England Journal of Medicine, 2015, 372, 2243-2257.	13.9	764
8	Breast-Cancer Risk in Families with Mutations in PALB2. New England Journal of Medicine, 2014, 371, 497-506.	13.9	745
9	Contralateral Breast Cancer in BRCA1 and BRCA2 Mutation Carriers. Journal of Clinical Oncology, 2004, 22, 2328-2335.	0.8	595
10	Familial Adenomatous Polyposis. American Journal of Gastroenterology, 2006, 101, 385-398.	0.2	575
11	Salpingo-oophorectomy and the Risk of Ovarian, Fallopian Tube, and Peritoneal Cancers in Women With a <EMPH TYPE="ITAL">BRCA1</EMPH> or <EMPH TYPE="ITAL">BRCA2</EMPH> Mutation. JAMA - Journal of the American Medical Association, 2006, 296, 185.	3.8	544
12	Impact of Oophorectomy on Cancer Incidence and Mortality in Women With a BRCA1 or BRCA2 Mutation. Journal of Clinical Oncology, 2014, 32, 1547-1553.	0.8	523
13	Inherited Susceptibility to Common Cancers. New England Journal of Medicine, 2008, 359, 2143-2153.	13.9	462
14	Is Breast Cancer the Same Disease in Asian and Western Countries?. World Journal of Surgery, 2010, 34, 2308-2324.	0.8	460
15	A mutational signature reveals alterations underlying deficient homologous recombination repair in breast cancer. Nature Genetics, 2017, 49, 1476-1486.	9.4	427
16	Triple-Negative Breast Cancer: Distinguishing between Basal and Nonbasal Subtypes. Clinical Cancer Research, 2009, 15, 2302-2310.	3.2	422
17	Breast Cancer Risk Following Bilateral Oophorectomy in BRCA1 and BRCA2 Mutation Carriers: An International Case-Control Study. Journal of Clinical Oncology, 2005, 23, 7491-7496.	0.8	408
18	DICER1: mutations, microRNAs and mechanisms. Nature Reviews Cancer, 2014, 14, 662-672.	12.8	404

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19	Recurrent SomaticDICER1Mutations in Nonepithelial Ovarian Cancers. New England Journal of Medicine, 2012, 366, 234-242.	13.9	401
20	Identification of seven new prostate cancer susceptibility loci through a genome-wide association study. Nature Genetics, 2009, 41, 1116-1121.	9.4	389
21	Germline and somatic SMARCA4 mutations characterize small cell carcinoma of the ovary, hypercalcemic type. Nature Genetics, 2014, 46, 438-443.	9.4	383
22	Triple-Negative Breast Cancer: Risk Factors to Potential Targets. Clinical Cancer Research, 2008, 14, 8010-8018.	3.2	380
23	CD8+ lymphocyte infiltration is an independent favorable prognostic indicator in basal-like breast cancer. Breast Cancer Research, 2012, 14, R48.	2.2	376
24	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. Nature Genetics, 2017, 49, 680-691.	9.4	356
25	Intrinsic Breast Tumor Subtypes, Race, and Long-Term Survival in the Carolina Breast Cancer Study. Clinical Cancer Research, 2010, 16, 6100-6110.	3.2	351
26	The Prognostic Implication of the Basal-Like (Cyclin) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 467 Td (Ehigh/p27low/p53+/Glomeruloid-Micr Cancer. Cancer Research, 2004, 64, 830-835.	0.4	345
27	Oral Contraceptives and the Risk of Breast Cancer in BRCA1 and BRCA2 Mutation Carriers. Journal of the National Cancer Institute, 2002, 94, 1773-1779.	3.0	318
28	Use of immunohistochemical markers can refine prognosis in triple negative breast cancer. BMC Cancer, 2007, 7, 134.	1.1	316
29	International variation in rates of uptake of preventive options in BRCA1 and BRCA2 mutation carriers. International Journal of Cancer, 2008, 122, 2017-2022.	2.3	306
30	Frequency of recurrent BRCA1 and BRCA2 mutations in Ashkenazi Jewish breast cancer families. Nature Medicine, 1996, 2, 1179-1183.	15.2	294
31	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. Nature Genetics, 2017, 49, 1767-1778.	9.4	289
32	<emph type="ital">DICER1</emph> Mutations in Familial Multinodular Goiter With and Without Ovarian Sertoli-Leydig Cell Tumors. JAMA - Journal of the American Medical Association, 2011, 305, 68.	3.8	284
33	Hereditary breast cancer: new genetic developments, new therapeutic avenues. Human Genetics, 2008, 124, 31-42.	1.8	276
34	Estrogen Receptor Status in BRCA1- and BRCA2-Related Breast Cancer. Clinical Cancer Research, 2004, 10, 2029-2034.	3.2	270
35	Cancer Risks Associated With Germline PALB2 Pathogenic Variants: An International Study of 524 Families. Journal of Clinical Oncology, 2020, 38, 674-685.	0.8	270
36	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, 2003, 6, R8-R17.	2.2	262

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37	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, 1664-1669.	0.8	248
38	Tamoxifen and contralateral breast cancer inBRCA1 andBRCA2 carriers: An update. International Journal of Cancer, 2006, 118, 2281-2284.	2.3	246
39	Contralateral mastectomy and survival after breast cancer in carriers of BRCA1 and BRCA2 mutations: retrospective analysis. BMJ, The, 2014, 348, g226-g226.	3.0	238
40	The CDKN2A (p16) Gene and Human Cancer. Molecular Medicine, 1997, 3, 5-20.	1.9	228
41	The incidence of pancreatic cancer in BRCA1 and BRCA2 mutation carriers. British Journal of Cancer, 2012, 107, 2005-2009.	2.9	221
42	Reproductive risk factors for ovarian cancer in carriers of BRCA1 or BRCA2 mutations: a case-control study. Lancet Oncology, The, 2007, 8, 26-34.	5.1	220
43	Tumor necrosis factor and its receptors in human ovarian cancer. Potential role in disease progression.. Journal of Clinical Investigation, 1993, 91, 2194-2206.	3.9	219
44	Pituitary blastoma: a pathognomonic feature of germ-line DICER1 mutations. Acta Neuropathologica, 2014, 128, 111-122.	3.9	211
45	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, 2634-2640.	2.6	204
46	Achieving clinical success with BET inhibitors as anti-cancer agents. British Journal of Cancer, 2021, 124, 1478-1490.	2.9	204
47	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, 1123-1130.	2.6	203
48	Pregnancy and risk of early breast cancer in carriers of BRCA1 and BRCA2. Lancet, The, 1999, 354, 1846-1850.	6.3	202
49	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.	2.6	201
50	Predictors of contralateral breast cancer in BRCA1 and BRCA2 mutation carriers. British Journal of Cancer, 2011, 104, 1384-1392.	2.9	195
51	Targeted Prostate Cancer Screening in BRCA1 and BRCA2 Mutation Carriers: Results from the Initial Screening Round of the IMPACT Study. European Urology, 2014, 66, 489-499.	0.9	195
52	Analysis of PALB2/FANCN-associated breast cancer families. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 6788-6793.	3.3	192
53	A Basal Epithelial Phenotype Is More Frequent in Interval Breast Cancers Compared with Screen Detected Tumors. Cancer Epidemiology Biomarkers and Prevention, 2005, 14, 1108-1112.	1.1	185
54	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.	2.2	182

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55	Methionine Metabolism Shapes T Helper Cell Responses through Regulation of Epigenetic Reprogramming. Cell Metabolism, 2020, 31, 250-266.e9.	7.2	182
56	PALB2,CHEK2andATMrare variants and cancer risk: data from COGS. Journal of Medical Genetics, 2016, 53, 800-811.	1.5	174
57	Extending the phenotypes associated withDICER1mutations. Human Mutation, 2011, 32, 1381-1384.	1.1	173
58	Breast-feeding and the Risk of Breast Cancer in BRCA1 and BRCA2 Mutation Carriers. Journal of the National Cancer Institute, 2004, 96, 1094-1098.	3.0	172
59	Germline RECQL mutations are associated with breast cancer susceptibility. Nature Genetics, 2015, 47, 643-646.	9.4	168
60	HOXB13 is a susceptibility gene for prostate cancer: results from the International Consortium for Prostate Cancer Genetics (ICPCG). Human Genetics, 2013, 132, 5-14.	1.8	166
61	Retinoblastoma and Neuroblastoma Predisposition and Surveillance. Clinical Cancer Research, 2017, 23, e98-e106.	3.2	166
62	PREDICT Plus: development and validation of a prognostic model for early breast cancer that includes HER2. British Journal of Cancer, 2012, 107, 800-807.	2.9	163
63	Influence of BRCA1 mutations on nuclear grade and estrogen receptor status of breast carcinoma in Ashkenazi Jewish women. Cancer, 1997, 80, 435-441.	2.0	162
64	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, 1093-1097.	0.8	161
65	Bilateral Oophorectomy and Breast Cancer Risk inBRCA1andBRCA2Mutation Carriers. Journal of the National Cancer Institute, 2017, 109, .	3.0	160
66	Placental Cadherin and the Basal Epithelial Phenotype of BRCA1-Related Breast Cancer. Clinical Cancer Research, 2005, 11, 4003-4011.	3.2	157
67	Founder BRCA1 and BRCA2 Mutations in French Canadian Breast and Ovarian Cancer Families. American Journal of Human Genetics, 1998, 63, 1341-1351.	2.6	156
68	SMARCA4-mutated atypical teratoid/rhabdoid tumors are associated with inherited germline alterations and poor prognosis. Acta Neuropathologica, 2014, 128, 453-456.	3.9	155
69	CARD9 Deficiency and Spontaneous Central Nervous System Candidiasis: Complete Clinical Remission With GM-CSF Therapy. Clinical Infectious Diseases, 2014, 59, 81-84.	2.9	153
70	Effect of pregnancy as a risk factor for breast cancer inBRCA1/BRCA2 mutation carriers. International Journal of Cancer, 2005, 117, 988-991.	2.3	152
71	TP53 mutations in breast cancer associated with BRCA1 or BRCA2 germ-line mutations: distinctive spectrum and structural distribution. Cancer Research, 2001, 61, 4092-7.	0.4	152
72	Germline HAVCR2 mutations altering TIM-3 characterize subcutaneous panniculitis-like T cell lymphomas with hemophagocytic lymphohistiocytic syndrome. Nature Genetics, 2018, 50, 1650-1657.	9.4	151

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73	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
74	BRCA Challenge: BRCA Exchange as a global resource for variants in BRCA1 and BRCA2. PLoS Genetics, 2018, 14, e1007752.	1.5	148
75	Interim Results from the IMPACT Study: Evidence for Prostate-specific Antigen Screening in BRCA2 Mutation Carriers. European Urology, 2019, 76, 831-842.	0.9	148
76	BRCA1 functions as a breast stem cell regulator. Journal of Medical Genetics, 2004, 41, 1-5.	1.5	146
77	Germline and somatic FGFR1 abnormalities in dysembryoplastic neuroepithelial tumors. Acta Neuropathologica, 2016, 131, 847-863.	3.9	143
78	The risk of ovarian cancer after breast cancer in BRCA1 and BRCA2 carriers. Gynecologic Oncology, 2005, 96, 222-226.	0.6	141
79	Cancer Surveillance in Gorlin Syndrome and Rhabdoid Tumor Predisposition Syndrome. Clinical Cancer Research, 2017, 23, e62-e67.	3.2	139
80	A Combined Genomewide Linkage Scan of 1,233 Families for Prostate Cancerâ€“Susceptibility Genes Conducted by the International Consortium for Prostate Cancer Genetics. American Journal of Human Genetics, 2005, 77, 219-229.	2.6	138
81	Germ-line and somatic DICER1 mutations in pineoblastoma. Acta Neuropathologica, 2014, 128, 583-595.	3.9	137
82	A somatic BRCA1 mutation in an ovarian tumour. Nature Genetics, 1995, 9, 343-344.	9.4	135
83	A significant response to neoadjuvant chemotherapy in BRCA1/2 related breast cancer. Journal of Medical Genetics, 2002, 39, 608-610.	1.5	135
84	Analysis of the Gene Coding for the BRCA2-Interacting Protein PALB2 in Familial and Sporadic Pancreatic Cancer. Gastroenterology, 2009, 137, 1183-1186.	0.6	135
85	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, 5307-5312.	1.8	132
86	Rapid progression of prostate cancer in men with a BRCA2 mutation. British Journal of Cancer, 2008, 99, 371-374.	2.9	132
87	Clinico-pathological characteristics of BRCA1- and BRCA2-related breast cancer. , 2000, 18, 287-295.		131
88	Extensive DNA methylation in normal colorectal mucosa in hyperplastic polyposis. Gut, 2006, 55, 1467-1474.	6.1	131
89	BRCA1 and BRCA2: Chemosensitivity, Treatment Outcomes and Prognosis. Familial Cancer, 2006, 5, 135-142.	0.9	128
90	PTEN, DICER1, FH, and Their Associated Tumor Susceptibility Syndromes: Clinical Features, Genetics, and Surveillance Recommendations in Childhood. Clinical Cancer Research, 2017, 23, e76-e82.	3.2	128

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91	Biallelic <scp>DICER1</scp> mutations occur in Wilms tumours. Journal of Pathology, 2013, 230, 154-164.	2.1	127
92	Familial risks of squamous cell carcinoma of the head and neck: retrospective case-control study. BMJ: British Medical Journal, 1996, 313, 716-721.	2.4	127
93	Identification of a novel truncating PALB2mutation and analysis of its contribution to early-onset breast cancer in French-Canadian women. Breast Cancer Research, 2007, 9, R83.	2.2	126
94	SMARCA4 loss is synthetic lethal with CDK4/6 inhibition in non-small cell lung cancer. Nature Communications, 2019, 10, 557.	5.8	125
95	Biallelic Deleterious BRCA1 Mutations in a Woman with Early-Onset Ovarian Cancer. Cancer Discovery, 2013, 3, 399-405.	7.7	124
96	Linkage Analysis of Chromosome 1q Markers in 136 Prostate Cancer Families. American Journal of Human Genetics, 1998, 62, 653-658.	2.6	123
97	Mutational Signature Analysis Reveals NTHL1 Deficiency to Cause a Multi-tumor Phenotype. Cancer Cell, 2019, 35, 256-266.e5.	7.7	123
98	Hormone Replacement Therapy After Oophorectomy and Breast Cancer Risk Among BRCA1 Mutation Carriers. JAMA Oncology, 2018, 4, 1059.	3.4	121
99	Incidence of colorectal cancer in BRCA1 and BRCA2 mutation carriers: results from a follow-up study. British Journal of Cancer, 2014, 110, 530-534.	2.9	120
100	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. Nature Genetics, 2020, 52, 56-73.	9.4	120
101	Effect of Smoking on Breast Cancer in Carriers of Mutant BRCA1 or BRCA2 Genes. Journal of the National Cancer Institute, 1998, 90, 761-765.	3.0	118
102	Germline BRCA1/2 Mutations and p27^{Kip1} Protein Levels Independently Predict Outcome After Breast Cancer. Journal of Clinical Oncology, 2000, 18, 4045-4052.	0.8	118
103	The Founder Mutation MSH2*1906Gâ†'C Is an Important Cause of Hereditary Nonpolyposis Colorectal Cancer in the Ashkenazi Jewish Population. American Journal of Human Genetics, 2002, 71, 1395-1412.	2.6	118
104	No small surpriseÂâ€“Âsmall cell carcinoma of the ovary, hypercalcaemic type, is a malignant rhabdoid tumour. Journal of Pathology, 2014, 233, 209-214.	2.1	117
105	Frequent loss of heterozygosity on chromosome 6 in human ovarian carcinoma. British Journal of Cancer, 1993, 67, 551-559.	2.9	116
106	DICER1 Mutations Are Consistently Present in Moderately and Poorly Differentiated Sertoli-Leydig Cell Tumors. American Journal of Surgical Pathology, 2017, 41, 1178-1187.	2.1	114
107	Germline DICER1 mutations and familial cystic nephroma. Journal of Medical Genetics, 2010, 47, 863-866.	1.5	113
108	Molecular characterization of the spectrum of genomic deletions in the mismatch repair genesMSH2,MLH1,MSH6, andPMS2 responsible for hereditary nonpolyposis colorectal cancer (HNPCC). Genes Chromosomes and Cancer, 2005, 44, 123-138.	1.5	112

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109	Loss of heterozygosity and amplification on chromosome 11q in human ovarian cancer. British Journal of Cancer, 1993, 67, 268-273.	2.9	111
110	DICER1 hotspot mutations in non-epithelial gonadal tumours. British Journal of Cancer, 2013, 109, 2744-2750.	2.9	111
111	Exploring the Association BetweenDICER1Mutations and Differentiated Thyroid Carcinoma. Journal of Clinical Endocrinology and Metabolism, 2014, 99, E1072-E1077.	1.8	111
112	Frequency of premature menopause in women who carry a BRCA1 or BRCA2 mutation. Fertility and Sterility, 2013, 99, 1724-1728.	0.5	110
113	The Histomorphology of Lynch Syndromeâ€"associated Ovarian Carcinomas. American Journal of Surgical Pathology, 2014, 38, 1173-1181.	2.1	108
114	E6/E7 proteins of HPV type 16 and ErbB-2 cooperate to induce neoplastic transformation of primary normal oral epithelial cells. Oncogene, 2004, 23, 350-358.	2.6	107
115	On the origin and diffusion of BRCA1 c.5266dupC (5382insC) in European populations. European Journal of Human Genetics, 2011, 19, 300-306.	1.4	107
116	Effect of Oophorectomy on Survival After Breast Cancer inBRCA1andBRCA2Mutation Carriers. JAMA Oncology, 2015, 1, 306.	3.4	107
117	The incidence of endometrial cancer in women with BRCA1 and BRCA2 mutations: An international prospective cohort study. Gynecologic Oncology, 2013, 130, 127-131.	0.6	106
118	Ovarian and Breast Cancer Risks Associated With Pathogenic Variants in RAD51C and RAD51D. Journal of the National Cancer Institute, 2020, 112, 1242-1250.	3.0	106
119	Re: Magnetic Resonance Imaging and Mammography in Women With a Hereditary Risk of Breast Cancer. Journal of the National Cancer Institute, 2001, 93, 1754-1754.	3.0	105
120	Screening mammography and risk of breast cancer in BRCA1 and BRCA2 mutation carriers: a case-control study. Lancet Oncology, The, 2006, 7, 402-406.	5.1	104
121	Tumor size and survival in breast cancerâ€"a reappraisal. Nature Reviews Clinical Oncology, 2010, 7, 348-353.	12.5	104
122	Prognostic significance of CD8+ T lymphocytes in breast cancer depends upon both oestrogen receptor status and histological grade. Histopathology, 2011, 58, no-no.	1.6	104
123	Disruption of the expected positive correlation between breast tumor size and lymph node status inBRCA1-related breast carcinoma. Cancer, 2003, 98, 1569-1577.	2.0	103
124	Changes in body weight and the risk of breast cancer in BRCA1 and BRCA2mutation carriers. Breast Cancer Research, 2005, 7, R833-43.	2.2	103
125	A PALB2 mutation associated with high risk of breast cancer. Breast Cancer Research, 2010, 12, R109.	2.2	102
126	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.	1.1	102

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127	Pheochromocytoma and paraganglioma syndromes: genetics and management update. Current Oncology, 2014, 21, 8.	0.9	102
128	A novel gene encoding a B-box protein within the BRCA1 region at 17q21.1. Human Molecular Genetics, 1994, 3, 589-594.	1.4	98
129	Family history of cancer is a risk factor for squamous cell carcinoma of the head and neck in Brazil: A caseâ€control study. International Journal of Cancer, 1995, 63, 769-773.	2.3	98
130	Women's attitudes toward preventive strategies for hereditary breast or ovarian carcinoma differ from one country to another. Cancer, 2001, 92, 959-968.	2.0	98
131	Are triple-negative tumours and basal-like breast cancer synonymous?. Breast Cancer Research, 2007, 9, 404; author reply 405.	2.2	98
132	Poorly differentiated thyroid carcinoma of childhood and adolescence: a distinct entity characterized by DICER1 mutations. Modern Pathology, 2020, 33, 1264-1274.	2.9	96
133	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. Nature Communications, 2016, 7, 11375.	5.8	93
134	Tumour predisposition and cancer syndromes as models to study geneâ€“environment interactions. Nature Reviews Cancer, 2020, 20, 533-549.	12.8	93
135	An evaluation of needs of female BRCA1 and BRCA2 carriers undergoing genetic counselling. Journal of Medical Genetics, 2000, 37, 866-874.	1.5	92
136	Breastfeeding and the risk of breast cancer in BRCA1 and BRCA2 mutation carriers. Breast Cancer Research, 2012, 14, R42.	2.2	92
137	Impaired RASGRF1/ERKâ€“mediated GM-CSF response characterizes CARD9 deficiency in French-Canadians. Journal of Allergy and Clinical Immunology, 2016, 137, 1178-1188.e7.	1.5	92
138	Cancer Screening Recommendations and Clinical Management of Inherited Gastrointestinal Cancer Syndromes in Childhood. Clinical Cancer Research, 2017, 23, e107-e114.	3.2	91
139	Family History of Cancer and Cancer Risks in Women with BRCA1 or BRCA2 Mutations. Journal of the National Cancer Institute, 2010, 102, 1874-1878.	3.0	89
140	Very frequent loss of heterozygosity throughout chromosome 17 in sporadic ovarian carcinoma. International Journal of Cancer, 1993, 54, 220-225.	2.3	88
141	Impact of germlineBRCA1mutations and overexpression of p53 on prognosis and response to treatment following breast carcinoma. Cancer, 2003, 97, 527-536.	2.0	87
142	Infertility, treatment of infertility, and the risk of breast cancer among women with BRCA1 and BRCA2 mutations: a caseâ€“control study. Cancer Causes and Control, 2008, 19, 1111-1119.	0.8	87
143	In Brief: BRCA1 and BRCA2. Journal of Pathology, 2013, 230, 347-349.	2.1	87
144	Population genetic testing for cancer susceptibility: founder mutations to genomes. Nature Reviews Clinical Oncology, 2016, 13, 41-54.	12.5	86

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145	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.	0.6	85
146	Germ-line BRCA1 mutation is an adverse prognostic factor in Ashkenazi Jewish women with breast cancer. Clinical Cancer Research, 1997, 3, 2465-9.	3.2	85
147	E6/E7 of HPV Type 16 Promotes Cell Invasion and Metastasis of Human Breast Cancer Cells. Cell Cycle, 2007, 6, 2038-2042.	1.3	83
148	HomozygousBUB1BMutation and Susceptibility to Gastrointestinal Neoplasia. New England Journal of Medicine, 2010, 363, 2628-2637.	13.9	82
149	Small-Cell Carcinoma of the Ovary, Hypercalcemic Typeâ€“Genetics, New Treatment Targets, and Current Management Guidelines. Clinical Cancer Research, 2020, 26, 3908-3917.	3.2	82
150	Clinical practice guidelines for BRCA1 and BRCA2 genetic testing. European Journal of Cancer, 2021, 146, 30-47.	1.3	81
151	Prognostic importance of glomeruloid microvascular proliferation indicates an aggressive angiogenic phenotype in human cancers. Cancer Research, 2002, 62, 6808-11.	0.4	81
152	Oncogenic role of PDK4 in human colon cancer cells. British Journal of Cancer, 2017, 116, 930-936.	2.9	80
153	DICER1 Mutations Are Frequent in Adolescent-Onset Papillary Thyroid Carcinoma. Journal of Clinical Endocrinology and Metabolism, 2018, 103, 2009-2015.	1.8	79
154	Functional mechanisms underlying pleiotropic risk alleles at the 19p13.1 breastâ€“ovarian cancer susceptibility locus. Nature Communications, 2016, 7, 12675.	5.8	78
155	Breast, ovarian, and endometrial malignancies in systemic lupus erythematosus: a meta-analysis. British Journal of Cancer, 2011, 104, 1478-1481.	2.9	77
156	Primary node negative breast cancer in BRCA1 mutation carriers has a poor outcome. Annals of Oncology, 2000, 11, 307-314.	0.6	76
157	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.	1.1	76
158	Ten years of DICER1 mutations: Provenance, distribution, and associated phenotypes. Human Mutation, 2019, 40, 1939-1953.	1.1	76
159	CDK4/6 inhibitors target SMARCA4-determined cyclin D1 deficiency in hypercalcemic small cell carcinoma of the ovary. Nature Communications, 2019, 10, 558.	5.8	76
160	Pancreatic adenocarcinoma: epidemiology and genetics.. Journal of Medical Genetics, 1996, 33, 889-898.	1.5	75
161	Biallelic NTHL1 Mutations in a Woman with Multiple Primary Tumors. New England Journal of Medicine, 2015, 373, 1985-1986.	13.9	75
162	Association of Rare Pathogenic DNA Variants for Familial Hypercholesterolemia, Hereditary Breast and Ovarian Cancer Syndrome, and Lynch Syndrome With Disease Risk in Adults According to Family History. JAMA Network Open, 2020, 3, e203959.	2.8	75

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163	Allele loss and mutation screen at the Peutz-Jeghers (LKB1) locus (19p13.3) in sporadic ovarian tumours. British Journal of Cancer, 1999, 80, 70-72.	2.9	74
164	Rare germline mutations inPALB2and breast cancer risk: A population-based study. Human Mutation, 2012, 33, 674-680.	1.1	74
165	Coffee consumption and breast cancer risk amongBRCA1 andBRCA2 mutation carriers. International Journal of Cancer, 2006, 118, 103-107.	2.3	73
166	Risk of ipsilateral breast cancer in BRCA1 and BRCA2 mutation carriers. Breast Cancer Research and Treatment, 2011, 127, 287-296.	1.1	73
167	Age at menarche and the risk of breast cancer in BRCA1 and BRCA2 mutation carriers. Cancer Causes and Control, 2005, 16, 667-674.	0.8	71
168	Compromised BRCA1â€“PALB2 interaction is associated with breast cancer risk. Oncogene, 2017, 36, 4161-4170.	2.6	71
169	Immunohistochemical Expression of DNA Repair Proteins in Familial Breast Cancer Differentiate BRCA2-Associated Tumors. Journal of Clinical Oncology, 2005, 23, 7503-7511.	0.8	70
170	Breast-Cancer Risk in Families with Mutations in PALB2. New England Journal of Medicine, 2014, 371, 1650-1652.	13.9	68
171	An update on the central nervous system manifestations of DICER1 syndrome. Acta Neuropathologica, 2020, 139, 689-701.	3.9	68
172	High-sensitivity sequencing reveals multi-organ somatic mosaicism causing DICER1 syndrome. Journal of Medical Genetics, 2016, 53, 43-52.	1.5	67
173	Risk of pancreatic cancer among individuals with a family history of cancer of the pancreas. International Journal of Cancer, 2002, 97, 807-810.	2.3	66
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