Fergus J Couch

List of Publications by Year in descending order

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397 papers

41,534 citations

95 h-index 186

406 all docs

406 docs citations

406 times ranked 36181 citing authors

g-index

#	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 of Risk-Reducing Surgery in <emph type="ital">BRCA1</emph> or <emph type="ital">BRCA2</emph> Mutation Carriers With Cancer Risk and Mortality. JAMA - Journal of the American Medical Association, 2010, 304, 967.	3.8	1,241
3	Association analysis identifies 65 new breast cancer risk loci. Nature, 2017, 551, 92-94.	13.7	1,099
4	Large-scale genotyping identifies 41 new loci associated with breast cancer risk. Nature Genetics, 2013, 45, 353-361.	9.4	960
5	Secondary mutations as a mechanism of cisplatin resistance in BRCA2-mutated cancers. Nature, 2008, 451, 1116-1120.	13.7	934
6	Gene-Panel Sequencing and the Prediction of Breast-Cancer Risk. New England Journal of Medicine, 2015, 372, 2243-2257.	13.9	764
7	Breast-Cancer Risk in Families with Mutations in <i>PALB2</i> . New England Journal of Medicine, 2014, 371, 497-506.	13.9	745
8	Control of BRCA2 Cellular and Clinical Functions by a Nuclear Partner, PALB2. Molecular Cell, 2006, 22, 719-729.	4.5	724
9	Polygenic Risk Scores for Prediction of Breast Cancer and Breast Cancer Subtypes. American Journal of Human Genetics, 2019, 104, 21-34.	2.6	711
10	BRCA1Mutations in Women Attending Clinics That Evaluate the Risk of Breast Cancer. New England Journal of Medicine, 1997, 336, 1409-1415.	13.9	660
11	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
12	A common coding variant in CASP8 is associated with breast cancer risk. Nature Genetics, 2007, 39, 352-358.	9.4	591
13	Parent-of-origin-specific allelic associations among 106 genomic loci for age at menarche. Nature, 2014, 514, 92-97.	13.7	548
14	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
15	Inherited Mutations in 17 Breast Cancer Susceptibility Genes Among a Large Triple-Negative Breast Cancer Cohort Unselected for Family History of Breast Cancer. Journal of Clinical Oncology, 2015, 33, 304-311.	0.8	521
16	Pathology of Breast and Ovarian Cancers among <i>BRCA1</i> and <i>BRCA2</i> Mutation Carriers: Results from the Consortium of Investigators of Modifiers of <i brca1<="" i="">/<i>2</i> (CIMBA). Cancer Epidemiology Biomarkers and Prevention, 2012, 21, 134-147.</i>	1.1	513
17	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
18	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

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19	Associations Between Cancer Predisposition Testing Panel Genes and Breast Cancer. JAMA Oncology, 2017, 3, 1190.	3.4	472
20	Newly discovered breast cancer susceptibility loci on 3p24 and 17q23.2. Nature Genetics, 2009, 41, 585-590.	9.4	434
21	Prediction of Breast Cancer Risk Based on Profiling With Common Genetic Variants. Journal of the National Cancer Institute, 2015, 107, .	3.0	428
22	Genomic analyses identify hundreds of variants associated with age at menarche and support a role for puberty timing in cancer risk. Nature Genetics, 2017, 49, 834-841.	9.4	426
23	A Systematic Genetic Assessment of 1,433 Sequence Variants of Unknown Clinical Significance in the BRCA1 and BRCA2 Breast Cancer–Predisposition Genes. American Journal of Human Genetics, 2007, 81, 873-883.	2.6	416
24	A Population-Based Study of Genes Previously Implicated in Breast Cancer. New England Journal of Medicine, 2021, 384, 440-451.	13.9	414
25	Association of Type and Location of <i> BRCA1 < /i > and <i> BRCA2 < /i > Mutations With Risk of Breast and Ovarian Cancer. JAMA - Journal of the American Medical Association, 2015, 313, 1347.</i></i>	3.8	390
26	Mutations in the BRCA1 gene in families with early-onset breast and ovarian cancer. Nature Genetics, 1994, 8, 387-391.	9.4	384
27	Association Between Inherited Germline Mutations in Cancer Predisposition Genes and Risk of Pancreatic Cancer. JAMA - Journal of the American Medical Association, 2018, 319, 2401.	3.8	375
28	Genome-wide association studies identify four ER negative–specific breast cancer risk loci. Nature Genetics, 2013, 45, 392-398.	9.4	374
29	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
30	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. Nature Genetics, 2017, 49, 680-691.	9.4	356
31	Integrated Evaluation of DNA Sequence Variants of Unknown Clinical Significance: Application to BRCA1 and BRCA2. American Journal of Human Genetics, 2004, 75, 535-544.	2.6	351
32	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
33	Heterogeneity of Breast Cancer Associations with Five Susceptibility Loci by Clinical and Pathological Characteristics. PLoS Genetics, 2008, 4, e1000054.	1.5	315
34	Recommendations for application of the functional evidence PS3/BS3 criterion using the ACMG/AMP sequence variant interpretation framework. Genome Medicine, 2020, 12, 3.	3.6	312
35	A locus on 19p13 modifies risk of breast cancer in BRCA1 mutation carriers and is associated with hormone receptor–negative breast cancer in the general population. Nature Genetics, 2010, 42, 885-892.	9.4	309
36	International variation in rates of uptake of preventive options in <i>BRCA1</i> and <i>BRCA2</i> mutation carriers. International Journal of Cancer, 2008, 122, 2017-2022.	2.3	306

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37	Two Decades After <i>BRCA:</i> Setting Paradigms in Personalized Cancer Care and Prevention. Science, 2014, 343, 1466-1470.	6.0	300
38	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. Nature Genetics, 2017, 49, 1767-1778.	9.4	289
39	A common variant at the TERT-CLPTM1L locus is associated with estrogen receptor–negative breast cancer. Nature Genetics, 2011, 43, 1210-1214.	9.4	279
40	Cancer Risks Associated With Germline <i>PALB2</i> Pathogenic Variants: An International Study of 524 Families. Journal of Clinical Oncology, 2020, 38, 674-685.	0.8	270
41	ENIGMA-Evidence-based network for the interpretation of germline mutant alleles: An international initiative to evaluate risk and clinical significance associated with sequence variation in BRCA1 and BRCA2 genes. Human Mutation, 2012, 33, 2-7.	1.1	269
42	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
43	Counselling framework for moderate-penetrance cancer-susceptibility mutations. Nature Reviews Clinical Oncology, 2016, 13, 581-588.	12.5	258
44	Common Breast Cancer-Predisposition Alleles Are Associated with Breast Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. American Journal of Human Genetics, 2008, 82, 937-948.	2.6	257
45	Genome-wide association analysis identifies three new breast cancer susceptibility loci. Nature Genetics, 2012, 44, 312-318.	9.4	256
46	The Prevalence of BRCA2 Mutations in Familial Pancreatic Cancer. Cancer Epidemiology Biomarkers and Prevention, 2007, 16, 342-346.	1.1	255
47	Genome-Wide Association Study in BRCA1 Mutation Carriers Identifies Novel Loci Associated with Breast and Ovarian Cancer Risk. PLoS Genetics, 2013, 9, e1003212.	1.5	244
48	Evaluation of Polygenic Risk Scores for Breast and Ovarian Cancer Risk Prediction in BRCA1 and BRCA2 Mutation Carriers. Journal of the National Cancer Institute, 2017, 109, .	3.0	242
49	Fibroblast growth factor receptor 2 translocations in intrahepatic cholangiocarcinoma. Human Pathology, 2014, 45, 1630-1638.	1.1	235
50	Triple-Negative Breast Cancer Risk Genes Identified by Multigene Hereditary Cancer Panel Testing. Journal of the National Cancer Institute, 2018, 110, 855-862.	3.0	225
51	Identification of six new susceptibility loci for invasive epithelial ovarian cancer. Nature Genetics, 2015, 47, 164-171.	9.4	221
52	RAD51 135Gâ†'C Modifies Breast Cancer Risk among BRCA2 Mutation Carriers: Results from a Combined Analysis of 19 Studies. American Journal of Human Genetics, 2007, 81, 1186-1200.	2.6	217
53	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
54	A review of a multifactorial probability-based model for classification of BRCA1 and BRCA2 variants of uncertain significance (VUS). Human Mutation, 2012, 33, 8-21.	1.1	190

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55	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
56	Genetic insights into biological mechanisms governing human ovarian ageing. Nature, 2021, 596, 393-397.	13.7	183
57	Identification of nine new susceptibility loci for endometrial cancer. Nature Communications, 2018, 9, 3166.	5.8	178
58	The Contributions of Breast Density and Common Genetic Variation to Breast Cancer Risk. Journal of the National Cancer Institute, 2015 , 107 , .	3.0	174
59	<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
60	Common Breast Cancer Susceptibility Alleles and the Risk of Breast Cancer for <i>BRCA1</i> and <i>BRCA2</i> Mutation Carriers: Implications for Risk Prediction. Cancer Research, 2010, 70, 9742-9754.	0.4	169
61	A meta-analysis of genome-wide association studies of breast cancer identifies two novel susceptibility loci at 6q14 and 20q11. Human Molecular Genetics, 2012, 21, 5373-5384.	1.4	168
62	Genetic Alterations Associated With Progression From Pancreatic Intraepithelial Neoplasia to Invasive Pancreatic Tumor. Gastroenterology, 2013, 145, 1098-1109.e1.	0.6	166
63	<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
64	Exome sequencing identifies FANCM as a susceptibility gene for triple-negative breast cancer. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 15172-15177.	3.3	162
65	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
66	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
67	Age- and Tumor Subtype–Specific Breast Cancer Risk Estimates for <i>CHEK2</i> *1100delC Carriers. Journal of Clinical Oncology, 2016, 34, 2750-2760.	0.8	152
68	Prediction of Breast and Prostate Cancer Risks in Male <i>BRCA1</i> and <ibrca2< i=""> Mutation Carriers Using Polygenic Risk Scores. Journal of Clinical Oncology, 2017, 35, 2240-2250.</ibrca2<>	0.8	152
69	BRCA Challenge: BRCA Exchange as a global resource for variants in BRCA1 and BRCA2. PLoS Genetics, 2018, 14, e1007752.	1.5	148
70	Conflicting Interpretation of Genetic Variants and Cancer Risk by Commercial Laboratories as Assessed by the Prospective Registry of Multiplex Testing. Journal of Clinical Oncology, 2016, 34, 4071-4078.	0.8	147
71	Genome-wide association study identifies 25 known breast cancer susceptibility loci as risk factors for triple-negative breast cancer. Carcinogenesis, 2014, 35, 1012-1019.	1.3	145
72	An international initiative to identify genetic modifiers of cancer risk in BRCA1 and BRCA2 mutation carriers: the Consortium of Investigators of Modifiers of BRCA1 and BRCA2 (CIMBA). Breast Cancer Research, 2007, 9, 104.	2,2	136

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73	Evidence of Gene–Environment Interactions between Common Breast Cancer Susceptibility Loci and Established Environmental Risk Factors. PLoS Genetics, 2013, 9, e1003284.	1.5	136
74	A clinical guide to hereditary cancer panel testing: evaluation of gene-specific cancer associations and sensitivity of genetic testing criteria in a cohort of 165,000 high-risk patients. Genetics in Medicine, 2020, 22, 407-415.	1.1	136
75	Genetic heterogeneity in Peutz-Jeghers syndrome. Human Mutation, 2000, 16, 23-30.	1.1	125
76	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
77	Biallelic Deleterious <i>BRCA1</i> Mutations in a Woman with Early-Onset Ovarian Cancer. Cancer Discovery, 2013, 3, 399-405.	7.7	124
78	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. Nature Genetics, 2020, 52, 56-73.	9.4	120
79	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
80	A guide for functional analysis of <i>BRCA1</i> variants of uncertain significance. Human Mutation, 2012, 33, 1526-1537.	1.1	117
81	Prevalence of Pathogenic Mutations in Cancer Predisposition Genes among Pancreatic Cancer Patients. Cancer Epidemiology Biomarkers and Prevention, 2016, 25, 207-211.	1.1	116
82	Male breast cancer in a multi-gene panel testing cohort: insights and unexpected results. Breast Cancer Research and Treatment, 2017, 161, 575-586.	1.1	116
83	Evaluation of ACMG-Guideline-Based Variant Classification of Cancer Susceptibility and Non-Cancer-Associated Genes in Families Affected by Breast Cancer. American Journal of Human Genetics, 2016, 98, 801-817.	2.6	113
84	Determination of Cancer Risk Associated with Germ Line BRCA1 Missense Variants by Functional Analysis. Cancer Research, 2007, 67, 1494-1501.	0.4	110
85	Common variants in ZNF365 are associated with both mammographic density and breast cancer risk. Nature Genetics, 2011, 43, 185-187.	9.4	109
86	Common Breast Cancer Susceptibility Loci Are Associated with Triple-Negative Breast Cancer. Cancer Research, 2011, 71, 6240-6249.	0.4	109
87	Genome-wide association study identifies multiple loci associated with both mammographic density and breast cancer risk. Nature Communications, 2014, 5, 5303.	5.8	109
88	Prediction and assessment of splicing alterations: implications for clinical testing. Human Mutation, 2008, 29, 1304-1313.	1.1	108
89	Functional Assays for Classification of <i>BRCA2</i> Variants of Uncertain Significance. Cancer Research, 2008, 68, 3523-3531.	0.4	108
90	The contribution of pathogenic variants in breast cancer susceptibility genes to familial breast cancer risk. Npj Breast Cancer, 2017, 3, 22.	2.3	108

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91	Common Genetic Variation and Breast Cancer Riskâ€"Past, Present, and Future. Cancer Epidemiology Biomarkers and Prevention, 2018, 27, 380-394.	1.1	108
92	Functional Assays for Analysis of Variants of Uncertain Significance in <i>BRCA2</i> Luman Mutation, 2014, 35, 151-164.	1.1	107
93	Combined genetic and splicing analysis of BRCA1 c.[594-2A>C; 641A>G] highlights the relevance of naturally occurring in-frame transcripts for developing disease gene variant classification algorithms. Human Molecular Genetics, 2016, 25, 2256-2268.	1.4	106
94	Ovarian and Breast Cancer Risks Associated With Pathogenic Variants in <i>RAD51C</i> and <i>RAD51D</i> Journal of the National Cancer Institute, 2020, 112, 1242-1250.	3.0	106
95	Identification of a BRCA2-Specific Modifier Locus at 6p24 Related to Breast Cancer Risk. PLoS Genetics, 2013, 9, e1003173.	1.5	105
96	Evidence that breast cancer risk at the 2q35 locus is mediated through IGFBP5 regulation. Nature Communications, 2014, 5, 4999.	5.8	105
97	Frequency of mutations in a large series of clinically ascertained ovarian cancer cases tested on multi-gene panels compared to reference controls. Gynecologic Oncology, 2017, 147, 375-380.	0.6	105
98	A Classification Model for <i>BRCA2</i> DNA Binding Domain Missense Variants Based on Homology-Directed Repair Activity. Cancer Research, 2013, 73, 265-275.	0.4	103
99	Large scale multifactorial likelihood quantitative analysis of <i>BRCA1</i> and <i>BRCA2</i> variants: An ENIGMA resource to support clinical variant classification. Human Mutation, 2019, 40, 1557-1578.	1.1	102
100	Common Breast Cancer Susceptibility Variants in <i>LSP1</i> and <i>RAD51L1</i> Are Associated with Mammographic Density Measures that Predict Breast Cancer Risk. Cancer Epidemiology Biomarkers and Prevention, 2012, 21, 1156-1166.	1.1	101
101	19p13.1 Is a Triple-Negative–Specific Breast Cancer Susceptibility Locus. Cancer Research, 2012, 72, 1795-1803.	0.4	100
102	Common variants in LSP1, 2q35 and 8q24 and breast cancer risk for BRCA1 and BRCA2 mutation carriers. Human Molecular Genetics, 2009, 18, 4442-4456.	1.4	99
103	Height and Breast Cancer Risk: Evidence From Prospective Studies and Mendelian Randomization. Journal of the National Cancer Institute, 2015, 107, djv219.	3.0	99
104	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
105	Identification and molecular characterization of a new ovarian cancer susceptibility locus at 17q21.31. Nature Communications, 2013, 4, 1627.	5.8	98
106	BRCA1 R1699Q variant displaying ambiguous functional abrogation confers intermediate breast and ovarian cancer risk. Journal of Medical Genetics, 2012, 49, 525-532.	1.5	97
107	Functional evaluation and cancer risk assessment of BRCA2 unclassified variants. Cancer Research, 2005, 65, 417-26.	0.4	97
108	No evidence that protein truncating variants in <i>BRIP1</i> i>are associated with breast cancer risk: implications for gene panel testing. Journal of Medical Genetics, 2016, 53, 298-309.	1.5	94

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109	Assessment of functional effects of unclassified genetic variants. Human Mutation, 2008, 29, 1314-1326.	1.1	93
110	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. Nature Communications, 2016, 7, 11375.	5.8	93
111	<i>FANCM</i> c.5791C>T nonsense mutation (rs144567652) induces exon skipping, affects DNA repair activity and is a familial breast cancer risk factor. Human Molecular Genetics, 2015, 24, 5345-5355.	1.4	91
112	The Landscape of Somatic Genetic Alterations in Breast Cancers From ATM Germline Mutation Carriers. Journal of the National Cancer Institute, 2018, 110, 1030-1034.	3.0	90
113	Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. Nature Communications, 2019, 10, 1741.	5.8	90
114	Germ line Fanconi anemia complementation group C mutations and pancreatic cancer. Cancer Research, 2005, 65, 383-6.	0.4	89
115	Male breast cancer in BRCA1 and BRCA2 mutation carriers: pathology data from the Consortium of Investigators of Modifiers of BRCA1/2. Breast Cancer Research, 2016, 18, 15.	2.2	88
116	Joint associations of a polygenic risk score and environmental risk factors for breast cancer in the Breast Cancer Association Consortium. International Journal of Epidemiology, 2018, 47, 526-536.	0.9	88
117	Shared heritability and functional enrichment across six solid cancers. Nature Communications, 2019, 10, 431.	5.8	88
118	Detection of splicing aberrations caused by BRCA1 and BRCA2 sequence variants encoding missense substitutions: implications for prediction of pathogenicity. Human Mutation, 2010, 31, E1484-E1505.	1.1	86
119	BRCA1/2 Mutations and Bevacizumab in the Neoadjuvant Treatment of Breast Cancer: Response and Prognosis Results in Patients With Triple-Negative Breast Cancer From the GeparQuinto Study. Journal of Clinical Oncology, 2018, 36, 2281-2287.	0.8	86
120	The Role of Tbx2 and Tbx3 in Mammary Development and Tumorigenesis. Journal of Mammary Gland Biology and Neoplasia, 2004, 9, 109-118.	1.0	85
121	Common Genetic Variants and Modification of Penetrance of BRCA2-Associated Breast Cancer. PLoS Genetics, 2010, 6, e1001183.	1.5	85
122	Male breast cancer in the United States: Treatment patterns and prognostic factors in the 21st century. Cancer, 2020, 126, 26-36.	2.0	82
123	Polygenic risk scores and breast and epithelial ovarian cancer risks for carriers of BRCA1 and BRCA2 pathogenic variants. Genetics in Medicine, 2020, 22, 1653-1666.	1.1	82
124	The role of genetic breast cancer susceptibility variants as prognostic factors. Human Molecular Genetics, 2012, 21, 3926-3939.	1.4	80
125	Common variants at 12p11, 12q24, 9p21, 9q31.2 and in ZNF365 are associated with breast cancer risk for BRCA1 and/or BRCA2mutation carriers. Breast Cancer Research, 2012, 14, R33.	2.2	78
126	Functional mechanisms underlying pleiotropic risk alleles at the 19p13.1 breast–ovarian cancer susceptibility locus. Nature Communications, 2016, 7, 12675.	5.8	78

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127	Clinically Applicable Models to Characterize <i>BRCA1</i> Significance. Journal of Clinical Oncology, 2008, 26, 5393-5400.	0.8	77
128	Five endometrial cancer risk loci identified through genome-wide association analysis. Nature Genetics, 2016, 48, 667-674.	9.4	77
129	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
130	BRCA1/2 Sequence Variants of Uncertain Significance: A Primer for Providers to Assist in Discussions and in Medical Management. Oncologist, 2013, 18, 518-524.	1.9	76
131	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
132	<i>BRCA2</i> Hypomorphic Missense Variants Confer Moderate Risks of Breast Cancer. Cancer Research, 2017, 77, 2789-2799.	0.4	75
133	Splicing and multifactorial analysis of intronic BRCA1 and BRCA2 sequence variants identifies clinically significant splicing aberrations up to 12 nucleotides from the intron/exon boundary. Human Mutation, 2011, 32, 678-687.	1.1	74
134	BRCA2 Localization to the Midbody by Filamin A Regulates CEP55 Signaling and Completion of Cytokinesis. Developmental Cell, 2012, 23, 137-152.	3.1	74
135	Impact of histopathology, tumor-infiltrating lymphocytes, and adjuvant chemotherapy on prognosis of triple-negative breast cancer. Breast Cancer Research and Treatment, 2018, 167, 89-99.	1.1	74
136	Common breast cancer susceptibility alleles are associated with tumour subtypes in BRCA1 and BRCA2 mutation carriers: results from the Consortium of Investigators of Modifiers of BRCA1/2. Breast Cancer Research, 2011, 13, R110.	2.2	71
137	Functional assays provide a robust tool for the clinical annotation of genetic variants of uncertain significance. Npj Genomic Medicine, $2016,1,.$	1.7	70
138	Strong Evidence of a Genetic Determinant for Mammographic Density, a Major Risk Factor for Breast Cancer. Cancer Research, 2007, 67, 8412-8418.	0.4	69
139	TBX2 is preferentially amplified in BRCA1- and BRCA2-related breast tumors. Cancer Research, 2002, 62, 3587-91.	0.4	69
140	Increased prevalence of the BRCA2 polymorphic stop codon K3326X among individuals with familial pancreatic cancer. Oncogene, 2005, 24, 3652-3656.	2.6	68
141	Common alleles at $6q25.1$ and $1p11.2$ are associated with breast cancer risk for BRCA1 and BRCA2 mutation carriers. Human Molecular Genetics, $2011, 20, 3304-3321$.	1.4	68
142	Association of Genetic Variation in Genes Implicated in the \hat{l}^2 -Catenin Destruction Complex with Risk of Breast Cancer. Cancer Epidemiology Biomarkers and Prevention, 2008, 17, 2101-2108.	1.1	67
143	Genetic modifiers of CHEK2*1100delC-associated breast cancer risk. Genetics in Medicine, 2017, 19, 599-603.	1.1	67
144	Inactivation of Brca2 Promotes Trp53-Associated but Inhibits KrasG12D-Dependent Pancreatic Cancer Development in Mice. Gastroenterology, 2011, 140, 1303-1313.e3.	0.6	65

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145	Classification of missense substitutions in the BRCA genes: A database dedicated to Ex-UVs. Human Mutation, 2012, 33, 22-28.	1.1	65
146	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
147	Assessment of the Clinical Relevance of BRCA2 Missense Variants by Functional and Computational Approaches. American Journal of Human Genetics, 2018, 102, 233-248.	2.6	64
148	Evaluation of Germline Genetic Testing Criteria in a Hospital-Based Series of Women With Breast Cancer. Journal of Clinical Oncology, 2020, 38, 1409-1418.	0.8	64
149	Clinical Decision-Making in Patients with Variant of Uncertain Significance in BRCA1 or BRCA2 Genes. Annals of Surgical Oncology, 2017, 24, 3067-3072.	0.7	63
150	A contemporary review of male breast cancer: current evidence and unanswered questions. Cancer and Metastasis Reviews, 2018, 37, 599-614.	2.7	63
151	CYP19A1 fine-mapping and Mendelian randomization: estradiol is causal for endometrial cancer. Endocrine-Related Cancer, 2016, 23, 77-91.	1.6	62
152	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
153	The Contribution of Germline Predisposition Gene Mutations to Clinical Subtypes of Invasive Breast Cancer From a Clinical Genetic Testing Cohort. Journal of the National Cancer Institute, 2020, 112, 1231-1241.	3.0	61
154	Common variants associated with breast cancer in genome-wide association studies are modifiers of breast cancer risk in BRCA1 and BRCA2 mutation carriers. Human Molecular Genetics, 2010, 19, 2886-2897.	1.4	60
155	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
156	Identification of BRCA1 missense substitutions that confer partial functional activity: potential moderate risk variants?. Breast Cancer Research, 2007, 9, R82.	2.2	58
157	Associations of common breast cancer susceptibility alleles with risk of breast cancer subtypes in BRCA1 and BRCA2 mutation carriers. Breast Cancer Research, 2014, 16, 3416.	2.2	57
158	Crowdsourcing the General Public for Large Scale Molecular Pathology Studies in Cancer. EBioMedicine, 2015, 2, 681-689.	2.7	56
159	Identification of Novel Genetic Markers of Breast Cancer Survival. Journal of the National Cancer Institute, 2015, 107, .	3.0	56
160	Prognostic value of automated KI67 scoring in breast cancer: a centralised evaluation of 8088 patients from 10 study groups. Breast Cancer Research, 2016, 18, 104.	2.2	56
161	Novel Associations between Common Breast Cancer Susceptibility Variants and Risk-Predicting Mammographic Density Measures. Cancer Research, 2015, 75, 2457-2467.	0.4	55
162	A Computational Method to Classify Variants of Uncertain Significance Using Functional Assay Data with Application to <i>BRCA1</i> . Cancer Epidemiology Biomarkers and Prevention, 2011, 20, 1078-1088.	1.1	54

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