

Fergus J Couch

List of Publications by Citations

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

387
papers

30,991
citations

86
h-index

167
g-index

406
ext. papers

37,104
ext. citations

9.3
avg, IF

6.04
L-index

#	Paper	IF	Citations
387	Genome-wide association study identifies novel breast cancer susceptibility loci. <i>Nature</i> , 2007 , 447, 1087-93	50.4	1957
386	Association of risk-reducing surgery in BRCA1 or BRCA2 mutation carriers with cancer risk and mortality. <i>JAMA - Journal of the American Medical Association</i> , 2010 , 304, 967-75	27.4	993
385	Large-scale genotyping identifies 41 new loci associated with breast cancer risk. <i>Nature Genetics</i> , 2013 , 45, 353-61, 361e1-2	36.3	813
384	Secondary mutations as a mechanism of cisplatin resistance in BRCA2-mutated cancers. <i>Nature</i> , 2008 , 451, 1116-20	50.4	757
383	Association analysis identifies 65 new breast cancer risk loci. <i>Nature</i> , 2017 , 551, 92-94	50.4	643
382	Control of BRCA2 cellular and clinical functions by a nuclear partner, PALB2. <i>Molecular Cell</i> , 2006 , 22, 719-729	17.6	588
381	Gene-panel sequencing and the prediction of breast-cancer risk. <i>New England Journal of Medicine</i> , 2015 , 372, 2243-57	59.2	587
380	BRCA1 mutations in women attending clinics that evaluate the risk of breast cancer. <i>New England Journal of Medicine</i> , 1997 , 336, 1409-15	59.2	587
379	Breast-cancer risk in families with mutations in PALB2. <i>New England Journal of Medicine</i> , 2014 , 371, 497-506	59.2	576
378	A common coding variant in CASP8 is associated with breast cancer risk. <i>Nature Genetics</i> , 2007 , 39, 352-8	36.3	557
377	Associations of breast cancer risk factors with tumor subtypes: a pooled analysis from the Breast Cancer Association Consortium studies. <i>Journal of the National Cancer Institute</i> , 2011 , 103, 250-63	9.7	513
376	Salpingo-oophorectomy and the risk of ovarian, fallopian tube, and peritoneal cancers in women with a BRCA1 or BRCA2 Mutation. <i>JAMA - Journal of the American Medical Association</i> , 2006 , 296, 185-92	27.4	466
375	Inherited mutations in 17 breast cancer susceptibility genes among a large triple-negative breast cancer cohort unselected for family history of breast cancer. <i>Journal of Clinical Oncology</i> , 2015 , 33, 304-11	11.2	435
374	Multiple independent variants at the TERT locus are associated with telomere length and risks of breast and ovarian cancer. <i>Nature Genetics</i> , 2013 , 45, 371-84, 384e1-2	36.3	422
373	Pathology of breast and ovarian cancers among BRCA1 and BRCA2 mutation carriers: results from the Consortium of Investigators of Modifiers of BRCA1/2 (CIMBA). <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2012 , 21, 134-47	4	411
372	Genome-wide association analysis of more than 120,000 individuals identifies 15 new susceptibility loci for breast cancer. <i>Nature Genetics</i> , 2015 , 47, 373-80	36.3	406
371	Parent-of-origin-specific allelic associations among 106 genomic loci for age at menarche. <i>Nature</i> , 2014 , 514, 92-97	50.4	401

370	Newly discovered breast cancer susceptibility loci on 3p24 and 17q23.2. <i>Nature Genetics</i> , 2009 , 41, 585-906.3	36.3	393
369	Polygenic Risk Scores for Prediction of Breast Cancer and Breast Cancer Subtypes. <i>American Journal of Human Genetics</i> , 2019 , 104, 21-34	11	363
368	A systematic genetic assessment of 1,433 sequence variants of unknown clinical significance in the BRCA1 and BRCA2 breast cancer-predisposition genes. <i>American Journal of Human Genetics</i> , 2007 , 81, 873-83	11	360
367	Mutations in the BRCA1 gene in families with early-onset breast and ovarian cancer. <i>Nature Genetics</i> , 1994 , 8, 387-91	36.3	350
366	Associations Between Cancer Predisposition Testing Panel Genes and Breast Cancer. <i>JAMA Oncology</i> , 2017 , 3, 1190-1196	13.4	349
365	Genome-wide association studies identify four ER negative-specific breast cancer risk loci. <i>Nature Genetics</i> , 2013 , 45, 392-8, 398e1-2	36.3	327
364	Prediction of breast cancer risk based on profiling with common genetic variants. <i>Journal of the National Cancer Institute</i> , 2015 , 107,	9.7	324
363	Integrated evaluation of DNA sequence variants of unknown clinical significance: application to BRCA1 and BRCA2. <i>American Journal of Human Genetics</i> , 2004 , 75, 535-44	11	312
362	Association of type and location of BRCA1 and BRCA2 mutations with risk of breast and ovarian cancer. <i>JAMA - Journal of the American Medical Association</i> , 2015 , 313, 1347-61	27.4	286
361	Heterogeneity of breast cancer associations with five susceptibility loci by clinical and pathological characteristics. <i>PLoS Genetics</i> , 2008 , 4, e1000054	6	280
360	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. <i>Nature Genetics</i> , 2010 , 42, 885-92	36.3	276
359	International variation in rates of uptake of preventive options in BRCA1 and BRCA2 mutation carriers. <i>International Journal of Cancer</i> , 2008 , 122, 2017-22	7.5	268
358	Oral contraceptives and the risk of breast cancer in BRCA1 and BRCA2 mutation carriers. <i>Journal of the National Cancer Institute</i> , 2002 , 94, 1773-9	9.7	266
357	Genomic analyses identify hundreds of variants associated with age at menarche and support a role for puberty timing in cancer risk. <i>Nature Genetics</i> , 2017 , 49, 834-841	36.3	257
356	A common variant at the TERT-CLPTM1L locus is associated with estrogen receptor-negative breast cancer. <i>Nature Genetics</i> , 2011 , 43, 1210-4	36.3	253
355	Genome-wide association analysis identifies three new breast cancer susceptibility loci. <i>Nature Genetics</i> , 2012 , 44, 312-8	36.3	237
354	Two decades after BRCA: setting paradigms in personalized cancer care and prevention. <i>Science</i> , 2014 , 343, 1466-70	33.3	233
353	Large-scale genomic analyses link reproductive aging to hypothalamic signaling, breast cancer susceptibility and BRCA1-mediated DNA repair. <i>Nature Genetics</i> , 2015 , 47, 1294-1303	36.3	226

352	Association Between Inherited Germline Mutations in Cancer Predisposition Genes and Risk of Pancreatic Cancer. <i>JAMA - Journal of the American Medical Association</i> , 2018 , 319, 2401-2409	27.4	222
351	Common breast cancer-predisposition alleles are associated with breast cancer risk in BRCA1 and BRCA2 mutation carriers. <i>American Journal of Human Genetics</i> , 2008 , 82, 937-48	11	218
350	The prevalence of BRCA2 mutations in familial pancreatic cancer. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2007 , 16, 342-6	4	217
349	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. <i>Human Mutation</i> , 2012 , 33, 2-7	4.7	211
348	Genome-wide association study in BRCA1 mutation carriers identifies novel loci associated with breast and ovarian cancer risk. <i>PLoS Genetics</i> , 2013 , 9, e1003212	6	209
347	RAD51 135G-->C modifies breast cancer risk among BRCA2 mutation carriers: results from a combined analysis of 19 studies. <i>American Journal of Human Genetics</i> , 2007 , 81, 1186-200	11	204
346	Counselling framework for moderate-penetrance cancer-susceptibility mutations. <i>Nature Reviews Clinical Oncology</i> , 2016 , 13, 581-8	19.4	200
345	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. <i>Nature Genetics</i> , 2017 , 49, 680-691	36.3	190
344	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. <i>Nature Genetics</i> , 2017 , 49, 1767-1778	36.3	186
343	Fibroblast growth factor receptor 2 translocations in intrahepatic cholangiocarcinoma. <i>Human Pathology</i> , 2014 , 45, 1630-8	3.7	179
342	Identification of six new susceptibility loci for invasive epithelial ovarian cancer. <i>Nature Genetics</i> , 2015 , 47, 164-71	36.3	177
341	Functional variants at the 11q13 risk locus for breast cancer regulate cyclin D1 expression through long-range enhancers. <i>American Journal of Human Genetics</i> , 2013 , 92, 489-503	11	167
340	Evaluation of Polygenic Risk Scores for Breast and Ovarian Cancer Risk Prediction in BRCA1 and BRCA2 Mutation Carriers. <i>Journal of the National Cancer Institute</i> , 2017 , 109,	9.7	153
339	Common breast cancer susceptibility alleles and the risk of breast cancer for BRCA1 and BRCA2 mutation carriers: implications for risk prediction. <i>Cancer Research</i> , 2010 , 70, 9742-54	10.1	147
338	Triple-Negative Breast Cancer Risk Genes Identified by Multigene Hereditary Cancer Panel Testing. <i>Journal of the National Cancer Institute</i> , 2018 , 110, 855-862	9.7	145
337	A review of a multifactorial probability-based model for classification of BRCA1 and BRCA2 variants of uncertain significance (VUS). <i>Human Mutation</i> , 2012 , 33, 8-21	4.7	143
336	A meta-analysis of genome-wide association studies of breast cancer identifies two novel susceptibility loci at 6q14 and 20q11. <i>Human Molecular Genetics</i> , 2012 , 21, 5373-84	5.6	143
335	Low penetrance breast cancer susceptibility loci are associated with specific breast tumor subtypes: findings from the Breast Cancer Association Consortium. <i>Human Molecular Genetics</i> , 2011 , 20, 3289-303	5.6	140

334	CHEK2*1100delC heterozygosity in women with breast cancer associated with early death, breast cancer-specific death, and increased risk of a second breast cancer. <i>Journal of Clinical Oncology</i> , 2012 , 30, 4308-16	2.2	134
333	Cancer Risks Associated With Germline Pathogenic Variants: An International Study of 524 Families. <i>Journal of Clinical Oncology</i> , 2020 , 38, 674-685	2.2	133
332	The contributions of breast density and common genetic variation to breast cancer risk. <i>Journal of the National Cancer Institute</i> , 2015 , 107,	9.7	128
331	Exome sequencing identifies FANCM as a susceptibility gene for triple-negative breast cancer. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2014 , 111, 15172-7	11.5	128
330	Genetic alterations associated with progression from pancreatic intraepithelial neoplasia to invasive pancreatic tumor. <i>Gastroenterology</i> , 2013 , 145, 1098-1109.e1	13.3	126
329	Genome-wide association study identifies 25 known breast cancer susceptibility loci as risk factors for triple-negative breast cancer. <i>Carcinogenesis</i> , 2014 , 35, 1012-9	4.6	121
328	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). <i>Breast Cancer Research</i> , 2007 , 9, 104	8.3	121
327	PALB2, CHEK2 and ATM rare variants and cancer risk: data from COGS. <i>Journal of Medical Genetics</i> , 2016 , 53, 800-811	5.8	121
326	A Population-Based Study of Genes Previously Implicated in Breast Cancer. <i>New England Journal of Medicine</i> , 2021 , 384, 440-451	59.2	115
325	Evidence of gene-environment interactions between common breast cancer susceptibility loci and established environmental risk factors. <i>PLoS Genetics</i> , 2013 , 9, e1003284	6	112
324	Conflicting Interpretation of Genetic Variants and Cancer Risk by Commercial Laboratories as Assessed by the Prospective Registry of Multiplex Testing. <i>Journal of Clinical Oncology</i> , 2016 , 34, 4071-4078	2.7	110
323	Genetic heterogeneity in Peutz-Jeghers syndrome. <i>Human Mutation</i> , 2000 , 16, 23-30	4.7	109
322	Age- and Tumor Subtype-Specific Breast Cancer Risk Estimates for CHEK2*1100delC Carriers. <i>Journal of Clinical Oncology</i> , 2016 , 34, 2750-60	2.2	107
321	Biallelic deleterious BRCA1 mutations in a woman with early-onset ovarian cancer. <i>Cancer Discovery</i> , 2013 , 3, 399-405	24.4	106
320	Recommendations for application of the functional evidence PS3/BS3 criterion using the ACMG/AMP sequence variant interpretation framework. <i>Genome Medicine</i> , 2019 , 12, 3	14.4	106
319	Genome-Wide Meta-Analyses of Breast, Ovarian, and Prostate Cancer Association Studies Identify Multiple New Susceptibility Loci Shared by at Least Two Cancer Types. <i>Cancer Discovery</i> , 2016 , 6, 1052-67	24.4	104
318	Prediction of Breast and Prostate Cancer Risks in Male BRCA1 and BRCA2 Mutation Carriers Using Polygenic Risk Scores. <i>Journal of Clinical Oncology</i> , 2017 , 35, 2240-2250	2.2	101
317	A transcriptome-wide association study of 229,000 women identifies new candidate susceptibility genes for breast cancer. <i>Nature Genetics</i> , 2018 , 50, 968-978	36.3	101

3 ¹⁶	Common breast cancer susceptibility loci are associated with triple-negative breast cancer. <i>Cancer Research</i> , 2011 , 71, 6240-9	10.1	100
3 ¹⁵	Prediction and assessment of splicing alterations: implications for clinical testing. <i>Human Mutation</i> , 2008 , 29, 1304-13	4.7	98
3 ¹⁴	Determination of cancer risk associated with germ line BRCA1 missense variants by functional analysis. <i>Cancer Research</i> , 2007 , 67, 1494-501	10.1	98
3 ¹³	Common variants in ZNF365 are associated with both mammographic density and breast cancer risk. <i>Nature Genetics</i> , 2011 , 43, 185-7	36.3	96
3 ¹²	Functional assays for classification of BRCA2 variants of uncertain significance. <i>Cancer Research</i> , 2008 , 68, 3523-31	10.1	96
3 ¹¹	Functional evaluation and cancer risk assessment of BRCA2 unclassified variants. <i>Cancer Research</i> , 2005 , 65, 417-26	10.1	95
3 ¹⁰	A guide for functional analysis of BRCA1 variants of uncertain significance. <i>Human Mutation</i> , 2012 , 33, 1526-37	4.7	94
3 ⁰⁹	Breast cancer risk variants at 6q25 display different phenotype associations and regulate ESR1, RMND1 and CCDC170. <i>Nature Genetics</i> , 2016 , 48, 374-86	36.3	93
3 ⁰⁸	19p13.1 is a triple-negative-specific breast cancer susceptibility locus. <i>Cancer Research</i> , 2012 , 72, 1795-803	10.1	93
3 ⁰⁷	Common breast cancer susceptibility variants in LSP1 and RAD51L1 are associated with mammographic density measures that predict breast cancer risk. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2012 , 21, 1156-66	4	92
3 ⁰⁶	Common variants in LSP1, 2q35 and 8q24 and breast cancer risk for BRCA1 and BRCA2 mutation carriers. <i>Human Molecular Genetics</i> , 2009 , 18, 4442-56	5.6	91
3 ⁰⁵	Identification of a BRCA2-specific modifier locus at 6p24 related to breast cancer risk. <i>PLoS Genetics</i> , 2013 , 9, e1003173	6	90
3 ⁰⁴	BRCA Challenge: BRCA Exchange as a global resource for variants in BRCA1 and BRCA2. <i>PLoS Genetics</i> , 2018 , 14, e1007752	6	90
3 ⁰³	Prevalence of Pathogenic Mutations in Cancer Predisposition Genes among Pancreatic Cancer Patients. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2016 , 25, 207-11	4	88
3 ⁰²	Evidence that breast cancer risk at the 2q35 locus is mediated through IGFBP5 regulation. <i>Nature Communications</i> , 2014 , 4, 4999	17.4	87
3 ⁰¹	Evaluation of ACMG-Guideline-Based Variant Classification of Cancer Susceptibility and Non-Cancer-Associated Genes in Families Affected by Breast Cancer. <i>American Journal of Human Genetics</i> , 2016 , 98, 801-817	11	86
3 ⁰⁰	Identification and molecular characterization of a new ovarian cancer susceptibility locus at 17q21.31. <i>Nature Communications</i> , 2013 , 4, 1627	17.4	85
299	Assessment of functional effects of unclassified genetic variants. <i>Human Mutation</i> , 2008 , 29, 1314-26	4.7	85

298	Germ line Fanconi anemia complementation group C mutations and pancreatic cancer. <i>Cancer Research</i> , 2005 , 65, 383-6	10.1	85
297	Genome-wide association study identifies multiple loci associated with both mammographic density and breast cancer risk. <i>Nature Communications</i> , 2014 , 5, 5303	17.4	84
296	No evidence that protein truncating variants in BRIP1 are associated with breast cancer risk: implications for gene panel testing. <i>Journal of Medical Genetics</i> , 2016 , 53, 298-309	5.8	83
295	BRCA1 R1699Q variant displaying ambiguous functional abrogation confers intermediate breast and ovarian cancer risk. <i>Journal of Medical Genetics</i> , 2012 , 49, 525-32	5.8	82
294	Male breast cancer in a multi-gene panel testing cohort: insights and unexpected results. <i>Breast Cancer Research and Treatment</i> , 2017 , 161, 575-586	4.4	81
293	Fine-scale mapping of the FGFR2 breast cancer risk locus: putative functional variants differentially bind FOXA1 and E2F1. <i>American Journal of Human Genetics</i> , 2013 , 93, 1046-60	11	80
292	Genetically Predicted Body Mass Index and Breast Cancer Risk: Mendelian Randomization Analyses of Data from 145,000 Women of European Descent. <i>PLoS Medicine</i> , 2016 , 13, e1002105	11.6	80
291	The role of Tbx2 and Tbx3 in mammary development and tumorigenesis. <i>Journal of Mammary Gland Biology and Neoplasia</i> , 2004 , 9, 109-18	2.4	79
290	The contribution of pathogenic variants in breast cancer susceptibility genes to familial breast cancer risk. <i>Npj Breast Cancer</i> , 2017 , 3, 22	7.8	78
289	A classification model for BRCA2 DNA binding domain missense variants based on homology-directed repair activity. <i>Cancer Research</i> , 2013 , 73, 265-75	10.1	77
288	Genome-wide association study identifies 32 novel breast cancer susceptibility loci from overall and subtype-specific analyses. <i>Nature Genetics</i> , 2020 , 52, 572-581	36.3	76
287	Functional assays for analysis of variants of uncertain significance in BRCA2. <i>Human Mutation</i> , 2014 , 35, 151-64	4.7	75
286	The role of genetic breast cancer susceptibility variants as prognostic factors. <i>Human Molecular Genetics</i> , 2012 , 21, 3926-39	5.6	75
285	Height and Breast Cancer Risk: Evidence From Prospective Studies and Mendelian Randomization. <i>Journal of the National Cancer Institute</i> , 2015 , 107,	9.7	74
284	Common genetic variants and modification of penetrance of BRCA2-associated breast cancer. <i>PLoS Genetics</i> , 2010 , 6, e1001183	6	74
283	Clinically applicable models to characterize BRCA1 and BRCA2 variants of uncertain significance. <i>Journal of Clinical Oncology</i> , 2008 , 26, 5393-400	2.2	74
282	Identification of nine new susceptibility loci for endometrial cancer. <i>Nature Communications</i> , 2018 , 9, 3166	17.4	70
281	Common variants at 12p11, 12q24, 9p21, 9q31.2 and in ZNF365 are associated with breast cancer risk for BRCA1 and/or BRCA2 mutation carriers. <i>Breast Cancer Research</i> , 2012 , 14, R33	8.3	70

280	Frequency of mutations in a large series of clinically ascertained ovarian cancer cases tested on multi-gene panels compared to reference controls. <i>Gynecologic Oncology</i> , 2017 , 147, 375-380	4.9	70
279	FANCM c.5791C>T nonsense mutation (rs144567652) induces exon skipping, affects DNA repair activity and is a familial breast cancer risk factor. <i>Human Molecular Genetics</i> , 2015 , 24, 5345-55	5.6	68
278	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. <i>Genetics in Medicine</i> , 2020 , 22, 407-415	8.1	68
277	The Landscape of Somatic Genetic Alterations in Breast Cancers From ATM Germline Mutation Carriers. <i>Journal of the National Cancer Institute</i> , 2018 , 110, 1030-1034	9.7	65
276	Common Genetic Variation and Breast Cancer Risk-Past, Present, and Future. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2018 , 27, 380-394	4	65
275	BRCA2 Polymorphic Stop Codon K3326X and the Risk of Breast, Prostate, and Ovarian Cancers. <i>Journal of the National Cancer Institute</i> , 2016 , 108,	9.7	65
274	BRCA2 localization to the midbody by filamin A regulates cep55 signaling and completion of cytokinesis. <i>Developmental Cell</i> , 2012 , 23, 137-52	10.2	65
273	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. <i>Nature Communications</i> , 2016 , 7, 11375	17.4	64
272	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. <i>Breast Cancer Research</i> , 2011 , 13, R110	8.3	62
271	Common alleles at 6q25.1 and 1p11.2 are associated with breast cancer risk for BRCA1 and BRCA2 mutation carriers. <i>Human Molecular Genetics</i> , 2011 , 20, 3304-21	5.6	62
270	Strong evidence of a genetic determinant for mammographic density, a major risk factor for breast cancer. <i>Cancer Research</i> , 2007 , 67, 8412-8	10.1	62
269	BRCA1/2 sequence variants of uncertain significance: a primer for providers to assist in discussions and in medical management. <i>Oncologist</i> , 2013 , 18, 518-24	5.7	61
268	Increased prevalence of the BRCA2 polymorphic stop codon K3326X among individuals with familial pancreatic cancer. <i>Oncogene</i> , 2005 , 24, 3652-6	9.2	61
267	TBX2 is preferentially amplified in BRCA1- and BRCA2-related breast tumors. <i>Cancer Research</i> , 2002 , 62, 3587-91	10.1	61
266	Association of genetic variation in genes implicated in the beta-catenin destruction complex with risk of breast cancer. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2008 , 17, 2101-8	4	60
265	Fine-scale mapping of the 5q11.2 breast cancer locus reveals at least three independent risk variants regulating MAP3K1. <i>American Journal of Human Genetics</i> , 2015 , 96, 5-20	11	59
264	Male breast cancer in BRCA1 and BRCA2 mutation carriers: pathology data from the Consortium of Investigators of Modifiers of BRCA1/2. <i>Breast Cancer Research</i> , 2016 , 18, 15	8.3	58
263	Classification of missense substitutions in the BRCA genes: a database dedicated to Ex-UVs. <i>Human Mutation</i> , 2012 , 33, 22-8	4.7	56

262	Inactivation of Brca2 promotes Trp53-associated but inhibits KrasG12D-dependent pancreatic cancer development in mice. <i>Gastroenterology</i> , 2011 , 140, 1303-1313.e1-3	13.3	56
261	Common variants associated with breast cancer in genome-wide association studies are modifiers of breast cancer risk in BRCA1 and BRCA2 mutation carriers. <i>Human Molecular Genetics</i> , 2010 , 19, 2886-97	5.6	56
260	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. <i>Nature Genetics</i> , 2020 , 52, 56-73	36.3	56
259	Five endometrial cancer risk loci identified through genome-wide association analysis. <i>Nature Genetics</i> , 2016 , 48, 667-674	36.3	56
258	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. <i>Journal of Clinical Oncology</i> , 2018 , 36, 2281-2287	2.2	56
257	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. <i>Human Molecular Genetics</i> , 2016 , 25, 2256-2268	5.6	55
256	Functional assays provide a robust tool for the clinical annotation of genetic variants of uncertain significance. <i>Npj Genomic Medicine</i> , 2016 , 1,	6.2	55
255	Joint associations of a polygenic risk score and environmental risk factors for breast cancer in the Breast Cancer Association Consortium. <i>International Journal of Epidemiology</i> , 2018 , 47, 526-536	7.8	53
254	Functional mechanisms underlying pleiotropic risk alleles at the 19p13.1 breast-ovarian cancer susceptibility locus. <i>Nature Communications</i> , 2016 , 7, 12675	17.4	53
253	Large scale multifactorial likelihood quantitative analysis of BRCA1 and BRCA2 variants: An ENIGMA resource to support clinical variant classification. <i>Human Mutation</i> , 2019 , 40, 1557-1578	4.7	52
252	Ovarian and Breast Cancer Risks Associated With Pathogenic Variants in RAD51C and RAD51D. <i>Journal of the National Cancer Institute</i> , 2020 , 112, 1242-1250	9.7	51
251	Genetic modifiers of CHEK2*1100delC-associated breast cancer risk. <i>Genetics in Medicine</i> , 2017 , 19, 599-603	6.0	51
250	Identification of BRCA1 missense substitutions that confer partial functional activity: potential moderate risk variants?. <i>Breast Cancer Research</i> , 2007 , 9, R82	8.3	51
249	Mammographic breast density and breast cancer: evidence of a shared genetic basis. <i>Cancer Research</i> , 2012 , 72, 1478-84	10.1	50
248	Hypomorphic Missense Variants Confer Moderate Risks of Breast Cancer. <i>Cancer Research</i> , 2017 , 77, 2789-2799	10.1	49
247	Impact of histopathology, tumor-infiltrating lymphocytes, and adjuvant chemotherapy on prognosis of triple-negative breast cancer. <i>Breast Cancer Research and Treatment</i> , 2018 , 167, 89-99	4.4	48
246	Common non-synonymous SNPs associated with breast cancer susceptibility: findings from the Breast Cancer Association Consortium. <i>Human Molecular Genetics</i> , 2014 , 23, 6096-111	5.6	48
245	Detection of splicing aberrations caused by BRCA1 and BRCA2 sequence variants encoding missense substitutions: implications for prediction of pathogenicity. <i>Human Mutation</i> , 2010 , 31, E1484-503	4.7	48

244	Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. <i>Nature Communications</i> , 2019 , 10, 1741	17.4	47
243	Fine-mapping of the HNF1B multicancer locus identifies candidate variants that mediate endometrial cancer risk. <i>Human Molecular Genetics</i> , 2015 , 24, 1478-92	5.6	46
242	Associations of common breast cancer susceptibility alleles with risk of breast cancer subtypes in BRCA1 and BRCA2 mutation carriers. <i>Breast Cancer Research</i> , 2014 , 16, 3416	8.3	46
241	Shared heritability and functional enrichment across six solid cancers. <i>Nature Communications</i> , 2019 , 10, 431	17.4	45
240	Novel Associations between Common Breast Cancer Susceptibility Variants and Risk-Predicting Mammographic Density Measures. <i>Cancer Research</i> , 2015 , 75, 2457-67	10.1	45
239	Association between a germline OCA2 polymorphism at chromosome 15q13.1 and estrogen receptor-negative breast cancer survival. <i>Journal of the National Cancer Institute</i> , 2010 , 102, 650-62	9.7	45
238	Prognostic value of automated KI67 scoring in breast cancer: a centralised evaluation of 8088 patients from 10 study groups. <i>Breast Cancer Research</i> , 2016 , 18, 104	8.3	44
237	Crowdsourcing the General Public for Large Scale Molecular Pathology Studies in Cancer. <i>EBioMedicine</i> , 2015 , 2, 681-9	8.8	44
236	Modification of BRCA1-Associated Breast and Ovarian Cancer Risk by BRCA1-Interacting Genes. <i>Cancer Research</i> , 2011 , 71, 5792-805	10.1	44
235	Common variants at the 19p13.1 and ZNF365 loci are associated with ER subtypes of breast cancer and ovarian cancer risk in BRCA1 and BRCA2 mutation carriers. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2012 , 21, 645-57	4	44
234	Evidence that the 5p12 Variant rs10941679 Confers Susceptibility to Estrogen-Receptor-Positive Breast Cancer through FGF10 and MRPS30 Regulation. <i>American Journal of Human Genetics</i> , 2016 , 99, 903-911	11	43
233	Genetic Risk Score Mendelian Randomization Shows that Obesity Measured as Body Mass Index, but not Waist:Hip Ratio, Is Causal for Endometrial Cancer. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2016 , 25, 1503-1510	4	42
232	A computational method to classify variants of uncertain significance using functional assay data with application to BRCA1. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2011 , 20, 1078-88	4	42
231	CYP19A1 fine-mapping and Mendelian randomization: estradiol is causal for endometrial cancer. <i>Endocrine-Related Cancer</i> , 2016 , 23, 77-91	5.7	41
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