Amanda B Spurdle

List of Publications by Citations

393 papers	22,482 citations	74 h-index	136 g-index
438 ext. papers	26,610 ext. citations	7.2 avg, IF	5.65 L-index

#	Paper	IF	Citations
393	Genome-wide association study identifies novel breast cancer susceptibility loci. *Nature*, **2007**, 447, 1087-93	36.4	1957
392	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
391	A common coding variant in CASP8 is associated with breast cancer risk. *Nature Genetics*, **2007**, 39, 352-8	36.3	557
390	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, 250-63	9.7	513
389	Type I and II endometrial cancers: have they different risk factors?. *Journal of Clinical Oncology*, **2013**, 31, 2607-18	2.2	458
388	Multiple independent variants at the TERT locus are associated with telomere length and risks of breast and ovarian cancer. *Nature Genetics*, **2013**, 45, 371-84, 384e1-2	36.3	422
387	Identification of 23 new prostate cancer susceptibility loci using the iCOGS custom genotyping array. *Nature Genetics*, **2013**, 45, 385-91, 391e1-2	36.3	413
386	Pathology of breast and ovarian cancers among BRCA1 and BRCA2 mutation carriers: results from the Consortium of Investigators of Modifiers of BRCA1/2 (CIMBA). *Cancer Epidemiology Biomarkers and Prevention*, **2012**, 21, 134-47	4	411
385	Newly discovered breast cancer susceptibility loci on 3p24 and 17q23.2. *Nature Genetics*, **2009**, 41, 585-90	36.3	393
384	Identification of seven new prostate cancer susceptibility loci through a genome-wide association study. *Nature Genetics*, **2009**, 41, 1116-21	36.3	360
383	Application of a 5-tiered scheme for standardized classification of 2,360 unique mismatch repair gene variants in the InSiGHT locus-specific database. *Nature Genetics*, **2014**, 46, 107-115	36.3	332
382	A meta-analysis of 87,040 individuals identifies 23 new susceptibility loci for prostate cancer. *Nature Genetics*, **2014**, 46, 1103-9	36.3	331
381	Association of type and location of BRCA1 and BRCA2 mutations with risk of breast and ovarian cancer. *JAMA - Journal of the American Medical Association*, **2015**, 313, 1347-61	27.4	286
380	Heterogeneity of breast cancer associations with five susceptibility loci by clinical and pathological characteristics. *PLoS Genetics*, **2008**, 4, e1000054	6	280
379	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, 885-92	36.3	276
378	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	36.3	257
377	Seven prostate cancer susceptibility loci identified by a multi-stage genome-wide association study. *Nature Genetics*, **2011**, 43, 785-91	36.3	243

376	Association Between Telomere Length and Risk of Cancer and Non-Neoplastic Diseases: A Mendelian Randomization Study. *JAMA Oncology*, **2017**, 3, 636-651	13.4	236
375	Common breast cancer-predisposition alleles are associated with breast cancer risk in BRCA1 and BRCA2 mutation carriers. *American Journal of Human Genetics*, **2008**, 82, 937-48	11	218
374	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	4.7	211
373	Genome-wide association study in BRCA1 mutation carriers identifies novel loci associated with breast and ovarian cancer risk. *PLoS Genetics*, **2013**, 9, e1003212	6	209
372	RAD51 135G-->C modifies breast cancer risk among BRCA2 mutation carriers: results from a combined analysis of 19 studies. *American Journal of Human Genetics*, **2007**, 81, 1186-200	11	204
371	Correlation of tumour BRAF mutations and MLH1 methylation with germline mismatch repair (MMR) gene mutation status: a literature review assessing utility of tumour features for MMR variant classification. *Journal of Medical Genetics*, **2012**, 49, 151-7	5.8	200
370	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. *Nature Genetics*, **2017**, 49, 680-691	36.3	190
369	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. *Nature Genetics*, **2017**, 49, 1767-1778	36.3	186
368	The OncoArray Consortium: A Network for Understanding the Genetic Architecture of Common Cancers. *Cancer Epidemiology Biomarkers and Prevention*, **2017**, 26, 126-135	4	183
367	Dominant negative ATM mutations in breast cancer families. *Journal of the National Cancer Institute* , **2002**, 94, 205-15	9.7	183
366	The geographic distribution of human Y chromosome variation. *Genetics*, **1997**, 145, 787-805	4	182
365	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,	9.7	153
364	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
363	Rare variants in the ATM gene and risk of breast cancer. *Breast Cancer Research*, **2011**, 13, R73	8.3	151
362	Tumor mismatch repair immunohistochemistry and DNA MLH1 methylation testing of patients with endometrial cancer diagnosed at age younger than 60 years optimizes triage for population-level germline mismatch repair gene mutation testing. *Journal of Clinical Oncology*, **2014**, 32, 90-100	2.2	149
361	Common breast cancer susceptibility alleles and the risk of breast cancer for BRCA1 and BRCA2 mutation carriers: implications for risk prediction. *Cancer Research*, **2010**, 70, 9742-54	10.1	147
360	Genetic evidence and integration of various data sources for classifying uncertain variants into a single model. *Human Mutation*, **2008**, 29, 1265-72	4.7	145
359	Breast Cancer Risk Genes - Association Analysis in More than 113,000 Women. *New England Journal of Medicine*, **2021**, 384, 428-439	59.2	143

358	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, 3289-303	5.6	140
357	Mutational spectrum in a worldwide study of 29,700 families with BRCA1 or BRCA2 mutations. *Human Mutation*, **2018**, 39, 593-620	4.7	138
356	Genetic and histopathologic evaluation of BRCA1 and BRCA2 DNA sequence variants of unknown clinical significance. *Cancer Research*, **2006**, 66, 2019-27	10.1	138
355	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
354	Analysis of cancer risk and BRCA1 and BRCA2 mutation prevalence in the kConFab familial breast cancer resource. *Breast Cancer Research*, **2006**, 8, R12	8.3	125
353	BRCA1 and BRCA2 genetic testing-pitfalls and recommendations for managing variants of uncertain clinical significance. *Annals of Oncology*, **2015**, 26, 2057-65	10.3	124
352	Genome-wide association study identifies a common variant associated with risk of endometrial cancer. *Nature Genetics*, **2011**, 43, 451-4	36.3	121
351	PALB2, CHEK2 and ATM rare variants and cancer risk: data from COGS. *Journal of Medical Genetics*, **2016**, 53, 800-811	5.8	121
350	The CYP3A4*1B polymorphism has no functional significance and is not associated with risk of breast or ovarian cancer. *Pharmacogenetics and Genomics*, **2002**, 12, 355-66		117
349	Evidence of gene-environment interactions between common breast cancer susceptibility loci and established environmental risk factors. *PLoS Genetics*, **2013**, 9, e1003284	6	112
348	Polycystic ovary syndrome increases the risk of endometrial cancer in women aged less than 50 years: an Australian case-control study. *Cancer Causes and Control*, **2010**, 21, 2303-8	2.8	110
347	A meta-analysis of genome-wide association studies to identify prostate cancer susceptibility loci associated with aggressive and non-aggressive disease. *Human Molecular Genetics*, **2013**, 22, 408-15	5.6	109
346	Age- and Tumor Subtype-Specific Breast Cancer Risk Estimates for CHEK2*1100delC Carriers. *Journal of Clinical Oncology*, **2016**, 34, 2750-60	2.2	107
345	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, 1052-67	24.4	104
344	Prediction of Breast and Prostate Cancer Risks in Male BRCA1 and BRCA2 Mutation Carriers Using Polygenic Risk Scores. *Journal of Clinical Oncology*, **2017**, 35, 2240-2250	2.2	101
343	Germline pathogenic variants of 11 breast cancer genes in 7,051 Japanese patients and 11,241 controls. *Nature Communications*, **2018**, 9, 4083	17.4	99
342	Prediction and assessment of splicing alterations: implications for clinical testing. *Human Mutation*, **2008**, 29, 1304-13	4.7	98
341	Evidence of a Causal Association Between Insulinemia and Endometrial Cancer: A Mendelian Randomization Analysis. *Journal of the National Cancer Institute*, **2015**, 107,	9.7	96

340	Common variants in LSP1, 2q35 and 8q24 and breast cancer risk for BRCA1 and BRCA2 mutation carriers. *Human Molecular Genetics*, **2009**, 18, 4442-56	5.6	91
339	Identification of a BRCA2-specific modifier locus at 6p24 related to breast cancer risk. *PLoS Genetics*, **2013**, 9, e1003173	6	90
338	Risk of estrogen receptor-positive and -negative breast cancer and single-nucleotide polymorphism 2q35-rs13387042. *Journal of the National Cancer Institute*, **2009**, 101, 1012-8	9.7	90
337	BRCA Challenge: BRCA Exchange as a global resource for variants in BRCA1 and BRCA2. *PLoS Genetics*, **2018**, 14, e1007752	6	90
336	Fine-mapping identifies multiple prostate cancer risk loci at 5p15, one of which associates with TERT expression. *Human Molecular Genetics*, **2013**, 22, 2520-8	5.6	88
335	Polygenic hazard score to guide screening for aggressive prostate cancer: development and validation in large scale cohorts. *BMJ, The*, **2018**, 360, j5757	5.9	85
334	ABCB1 (MDR 1) polymorphisms and progression-free survival among women with ovarian cancer following paclitaxel/carboplatin chemotherapy. *Clinical Cancer Research*, **2008**, 14, 5594-601	12.9	83
333	Opposite effects of androgen receptor CAG repeat length on increased risk of left-handedness in males and females. *Behavior Genetics*, **2005**, 35, 735-44	3.2	83
332	Refined histopathological predictors of BRCA1 and BRCA2 mutation status: a large-scale analysis of breast cancer characteristics from the BCAC, CIMBA, and ENIGMA consortia. *Breast Cancer Research* , **2014**, 16, 3419	8.3	82
331	BRCA1 R1699Q variant displaying ambiguous functional abrogation confers intermediate breast and ovarian cancer risk. *Journal of Medical Genetics*, **2012**, 49, 525-32	5.8	82
330	Incidence, risk factors and estimates of a woman's risk of developing secondary lower limb lymphedema and lymphedema-specific supportive care needs in women treated for endometrial cancer. *Gynecologic Oncology*, **2015**, 136, 87-93	4.9	81
329	Molecular, pathologic, and clinical features of early-onset endometrial cancer: identifying presumptive Lynch syndrome patients. *Clinical Cancer Research*, **2008**, 14, 1692-700	12.9	81
328	Lynch syndrome-associated breast cancers: clinicopathologic characteristics of a case series from the colon cancer family registry. *Clinical Cancer Research*, **2010**, 16, 2214-24	12.9	80
327	Specifications of the ACMG/AMP variant curation guidelines for the analysis of germline CDH1 sequence variants. *Human Mutation*, **2018**, 39, 1553-1568	4.7	80
326	Population-based estimates of breast cancer risks associated with ATM gene variants c.7271T>G and c.1066-6T>G (IVS10-6T>G) from the Breast Cancer Family Registry. *Human Mutation*, **2006**, 27, 1122-8	4.7	78
325	Possible genetic predisposition to lymphedema after breast cancer. *Lymphatic Research and Biology* , **2012**, 10, 2-13	2.3	77
324	Association of ESR1 gene tagging SNPs with breast cancer risk. *Human Molecular Genetics*, **2009**, 18, 1131-9	5.6	75
323	Most common 'sporadic' cancers have a significant germline genetic component. *Human Molecular Genetics*, **2014**, 23, 6112-8	5.6	74

322	Common genetic variants and modification of penetrance of BRCA2-associated breast cancer. *PLoS Genetics*, **2010**, 6, e1001183	6	74
321	Assessing interactions between the associations of common genetic susceptibility variants, reproductive history and body mass index with breast cancer risk in the breast cancer association consortium: a combined case-control study. *Breast Cancer Research*, **2010**, 12, R110	8.3	74
320	Interplay between BRCA1 and RHAMM regulates epithelial apicobasal polarization and may influence risk of breast cancer. *PLoS Biology*, **2011**, 9, e1001199	9.7	73
319	Serum HE4 as a prognostic marker in endometrial cancer--a population based study. *Gynecologic Oncology*, **2014**, 132, 159-65	4.9	72
318	Identification of a novel prostate cancer susceptibility variant in the KLK3 gene transcript. *Human Genetics*, **2011**, 129, 687-94	6.3	72
317	Mutations in the human mannose-binding protein gene: frequencies in several population groups. *European Journal of Human Genetics*, **1996**, 4, 13-9	5.3	72
316	Identification of nine new susceptibility loci for endometrial cancer. *Nature Communications*, **2018**, 9, 3166	17.4	70
315	Common variants at 12p11, 12q24, 9p21, 9q31.2 and in ZNF365 are associated with breast cancer risk for BRCA1 and/or BRCA2 mutation carriers. *Breast Cancer Research*, **2012**, 14, R33	8.3	70
314	A multifactorial likelihood model for MMR gene variant classification incorporating probabilities based on sequence bioinformatics and tumor characteristics: a report from the Colon Cancer Family Registry. *Human Mutation*, **2013**, 34, 200-9	4.7	70
313	Calibration of multiple in silico tools for predicting pathogenicity of mismatch repair gene missense substitutions. *Human Mutation*, **2013**, 34, 255-65	4.7	70
312	Double-strand break repair gene polymorphisms and risk of breast or ovarian cancer. *Cancer Epidemiology Biomarkers and Prevention*, **2005**, 14, 319-23	4	70
311	A single nucleotide polymorphism in the 5' untranslated region of RAD51 and risk of cancer among BRCA1/2 mutation carriers. *Cancer Epidemiology Biomarkers and Prevention*, **2001**, 10, 955-60	4	67
310	Associations of common variants at 1p11.2 and 14q24.1 (RAD51L1) with breast cancer risk and heterogeneity by tumor subtype: findings from the Breast Cancer Association Consortium. *Human Molecular Genetics*, **2011**, 20, 4693-706	5.6	66
309	BRCA2 Polymorphic Stop Codon K3326X and the Risk of Breast, Prostate, and Ovarian Cancers. *Journal of the National Cancer Institute*, **2016**, 108,	9.7	65
308	Clinical classification of BRCA1 and BRCA2 DNA sequence variants: the value of cytokeratin profiles and evolutionary analysis--a report from the kConFab Investigators. *Journal of Clinical Oncology*, **2008**, 26, 1657-63	2.2	65
307	Androgen receptor exon 1 CAG repeat length and breast cancer in women before age forty years. *Journal of the National Cancer Institute*, **1999**, 91, 961-6	9.7	63
306	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	8.3	62
305	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, 3304-21	5.6	62

304	Aspirin, nonsteroidal anti-inflammatory drugs, paracetamol and risk of endometrial cancer: a case-control study, systematic review and meta-analysis. *International Journal of Cancer*, **2013**, 132, 1146-55	7.5	60
303	Comprehensive annotation of splice junctions supports pervasive alternative splicing at the BRCA1 locus: a report from the ENIGMA consortium. *Human Molecular Genetics*, **2014**, 23, 3666-80	5.6	60
302	High-throughput interrogation of PIK3CA, PTEN, KRAS, FBXW7 and TP53 mutations in primary endometrial carcinoma. *Gynecologic Oncology*, **2013**, 128, 327-34	4.9	60
301	Polymorphisms at the glutathione S-transferase GSTM1, GSTT1 and GSTP1 loci: risk of ovarian cancer by histological subtype. *Carcinogenesis*, **2001**, 22, 67-72	4.6	60
300	Association between single-nucleotide polymorphisms in hormone metabolism and DNA repair genes and epithelial ovarian cancer: results from two Australian studies and an additional validation set. *Cancer Epidemiology Biomarkers and Prevention*, **2007**, 16, 2557-65	4	58
299	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, 2886-97	5.6	56
298	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. *Nature Genetics*, **2020**, 52, 56-73	36.3	56
297	Five endometrial cancer risk loci identified through genome-wide association analysis. *Nature Genetics*, **2016**, 48, 667-674	36.3	56
296	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	5.6	55
295	Classifying MLH1 and MSH2 variants using bioinformatic prediction, splicing assays, segregation, and tumor characteristics. *Human Mutation*, **2009**, 30, 757-70	4.7	55
294	Age at last birth in relation to risk of endometrial cancer: pooled analysis in the epidemiology of endometrial cancer consortium. *American Journal of Epidemiology*, **2012**, 176, 269-78	3.8	55
293	Multiple novel prostate cancer susceptibility signals identified by fine-mapping of known risk loci among Europeans. *Human Molecular Genetics*, **2015**, 24, 5589-602	5.6	54
292	Human papillomavirus DNA detected in peripheral blood samples from healthy Australian male blood donors. *Journal of Medical Virology*, **2009**, 81, 1792-6	19.7	54
291	Mismatch repair deficiency endows tumors with a unique mutation signature and sensitivity to DNA double-strand breaks. *ELife*, **2014**, 3, e02725	8.9	54
290	Comparison of mRNA splicing assay protocols across multiple laboratories: recommendations for best practice in standardized clinical testing. *Clinical Chemistry*, **2014**, 60, 341-52	5.5	53
289	Clinical problems of colorectal cancer and endometrial cancer cases with unknown cause of tumor mismatch repair deficiency (suspected Lynch syndrome). *The Application of Clinical Genetics*, **2014**, 7, 183-93	3.1	53
288	Five polymorphisms and breast cancer risk: results from the Breast Cancer Association Consortium. *Cancer Epidemiology Biomarkers and Prevention*, **2009**, 18, 1610-6	4	53
287	A novel corepressor, BCoR-L1, represses transcription through an interaction with CtBP. *Journal of Biological Chemistry*, **2007**, 282, 15248-57	5.4	53

286	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
285	Characterization of BRCA1 and BRCA2 splicing variants: a collaborative report by ENIGMA consortium members. *Breast Cancer Research and Treatment*, **2012**, 132, 1009-23	4.4	51
284	Gynecological conditions and the risk of endometrial cancer. *Gynecologic Oncology*, **2011**, 123, 537-41	4.9	51
283	Tumor characteristics as an analytic tool for classifying genetic variants of uncertain clinical significance. *Human Mutation*, **2008**, 29, 1292-303	4.7	51
282	PSA/KLK3 AREI promoter polymorphism alters androgen receptor binding and is associated with prostate cancer susceptibility. *Carcinogenesis*, **2007**, 28, 1032-9	4.6	51
281	Identification of BRCA1 missense substitutions that confer partial functional activity: potential moderate risk variants?. *Breast Cancer Research*, **2007**, 9, R82	8.3	51
280	Hypomorphic Missense Variants Confer Moderate Risks of Breast Cancer. *Cancer Research*, **2017**, 77, 2789-2799	10.1	49
279	Detection of splicing aberrations caused by BRCA1 and BRCA2 sequence variants encoding missense substitutions: implications for prediction of pathogenicity. *Human Mutation*, **2010**, 31, E1484-503	4.7	48
278	Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. *Nature Communications*, **2019**, 10, 1741	17.4	47
277	Risk Analysis of Prostate Cancer in PRACTICAL, a Multinational Consortium, Using 25 Known Prostate Cancer Susceptibility Loci. *Cancer Epidemiology Biomarkers and Prevention*, **2015**, 24, 1121-9	4	46
276	Fine-mapping of the HNF1B multicancer locus identifies candidate variants that mediate endometrial cancer risk. *Human Molecular Genetics*, **2015**, 24, 1478-92	5.6	46
275	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	8.3	46
274	The obesity-associated polymorphisms FTO rs9939609 and MC4R rs17782313 and endometrial cancer risk in non-Hispanic white women. *PLoS ONE*, **2011**, 6, e16756	3.7	46
273	Shared heritability and functional enrichment across six solid cancers. *Nature Communications*, **2019**, 10, 431	17.4	45
272	Impact of weight change and weight cycling on risk of different subtypes of endometrial cancer. *European Journal of Cancer*, **2013**, 49, 2717-26	7.5	45
271	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. *Cancer Epidemiology Biomarkers and Prevention*, **2012**, 21, 645-57	4	44
270	Molecular characterization and cancer risk associated with BRCA1 and BRCA2 splice site variants identified in multiple-case breast cancer families. *Human Mutation*, **2005**, 26, 495	4.7	44
269	CYP17 promoter polymorphism and breast cancer in Australian women under age forty years. *Journal of the National Cancer Institute*, **2000**, 92, 1674-81	9.7	44

268	Adding In Silico Assessment of Potential Splice Aberration to the Integrated Evaluation of BRCA Gene Unclassified Variants. *Human Mutation*, **2016**, 37, 627-39	4.7	44
267	Prediction of individual genetic risk to prostate cancer using a polygenic score. *Prostate*, **2015**, 75, 1467-74	4.2	43
266	A genetic variant of MDM4 influences regulation by multiple microRNAs in prostate cancer. *Endocrine-Related Cancer*, **2015**, 22, 265-76	5.7	43
265	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	4	42
264	Identification of fifteen novel germline variants in the BRCA1 3'UTR reveals a variant in a breast cancer case that introduces a functional miR-103 target site. *Human Mutation*, **2012**, 33, 1665-75	4.7	42
263	Evaluation of candidate stromal epithelial cross-talk genes identifies association between risk of serous ovarian cancer and TERT, a cancer susceptibility "hot-spot". *PLoS Genetics*, **2010**, 6, e1001016	6	42
262	Validating genetic risk associations for ovarian cancer through the international Ovarian Cancer Association Consortium. *British Journal of Cancer*, **2009**, 100, 412-20	8.7	42
261	CYP19A1 fine-mapping and Mendelian randomization: estradiol is causal for endometrial cancer. *Endocrine-Related Cancer*, **2016**, 23, 77-91	5.7	41
260	A Large-Scale Analysis of Genetic Variants within Putative miRNA Binding Sites in Prostate Cancer. *Cancer Discovery*, **2015**, 5, 368-79	24.4	41
259	Genetic overlap between endometriosis and endometrial cancer: evidence from cross-disease genetic correlation and GWAS meta-analyses. *Cancer Medicine*, **2018**, 7, 1978-1987	4.8	40
258	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, 678-87	4.7	39
257	Locus-specific databases and recommendations to strengthen their contribution to the classification of variants in cancer susceptibility genes. *Human Mutation*, **2008**, 29, 1273-81	4.7	39
256	The role of glutathione-S-transferase polymorphisms in ovarian cancer survival. *European Journal of Cancer*, **2007**, 43, 283-90	7.5	39
255	The androgen receptor CAG repeat polymorphism and modification of breast cancer risk in BRCA1 and BRCA2 mutation carriers. *Breast Cancer Research*, **2005**, 7, R176-83	8.3	39
254	Intrauterine devices and endometrial cancer risk: a pooled analysis of the Epidemiology of Endometrial Cancer Consortium. *International Journal of Cancer*, **2015**, 136, E410-22	7.5	38
253	Association of a common AKAP9 variant with breast cancer risk: a collaborative analysis. *Journal of the National Cancer Institute*, **2008**, 100, 437-42	9.7	38
252	Mutation analysis of FANCD2, BRIP1/BACH1, LMO4 and SFN in familial breast cancer. *Breast Cancer Research*, **2005**, 7, R1005-16	8.3	38
251	Atlas of prostate cancer heritability in European and African-American men pinpoints tissue-specific regulation. *Nature Communications*, **2016**, 7, 10979	17.4	37

250	Novel diagnostic tool for prediction of variant spliceogenicity derived from a set of 395 combined in silico/in vitro studies: an international collaborative effort. *Nucleic Acids Research*, **2018**, 46, 7913-7923	20.1	37
249	Genetic variation at 9p22.2 and ovarian cancer risk for BRCA1 and BRCA2 mutation carriers. *Journal of the National Cancer Institute*, **2011**, 103, 105-16	9.7	37
248	Naturally occurring BRCA2 alternative mRNA splicing events in clinically relevant samples. *Journal of Medical Genetics*, **2016**, 53, 548-58	5.8	37
247	The c. 5096G>A p.Arg1699Gln (R1699Q) intermediate risk variant: breast and ovarian cancer risk estimation and recommendations for clinical management from the ENIGMA consortium. *Journal of Medical Genetics*, **2018**, 55, 15-20	5.8	36
246	Glycemic index, glycemic load and endometrial cancer risk: results from the Australian National Endometrial Cancer study and an updated systematic review and meta-analysis. *European Journal of Nutrition*, **2013**, 52, 705-15	5.2	36
245	Genetic, functional, and histopathological evaluation of two C-terminal BRCA1 missense variants. *Journal of Medical Genetics*, **2006**, 43, 74-83	5.8	36
244	Whole-genome sequencing reveals clinically relevant insights into the aetiology of familial breast cancers. *Annals of Oncology*, **2019**, 30, 1071-1079	10.3	35
243	A systematic approach to analysing gene-gene interactions: polymorphisms at the microsomal epoxide hydrolase EPHX and glutathione S-transferase GSTM1, GSTT1, and GSTP1 loci and breast cancer risk. *Cancer Epidemiology Biomarkers and Prevention*, **2007**, 16, 769-74	4	35
242	Two ATM variants and breast cancer risk. *Human Mutation*, **2005**, 25, 594-5	4.7	35
241	Mutation deep within an intron of MSH2 causes Lynch syndrome. *Familial Cancer*, **2011**, 10, 297-301	3	34
240	BRCA2 Arg372Hispolymorphism and epithelial ovarian cancer risk. *International Journal of Cancer*, **2003**, 103, 427-30	7.5	34
239	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	8.1	34
238	Breastfeeding and Endometrial Cancer Risk: An Analysis From the Epidemiology of Endometrial Cancer Consortium. *Obstetrics and Gynecology*, **2017**, 129, 1059-1067	4.9	33
237	Genome-wide association study of endometrial cancer in E2C2. *Human Genetics*, **2014**, 133, 211-24	6.3	33
236	Clinical relevance of rare germline sequence variants in cancer genes: evolution and application of classification models. *Current Opinion in Genetics and Development*, **2010**, 20, 315-23	4.9	33
235	Germline Pathogenic Variants in 7636 Japanese Patients With Prostate Cancer and 12□366 Controls. *Journal of the National Cancer Institute*, **2020**, 112, 369-376	9.7	33
234	Assessment of polygenic architecture and risk prediction based on common variants across fourteen cancers. *Nature Communications*, **2020**, 11, 3353	17.4	32
233	The association between diabetes, comorbidities, body mass index and all-cause and cause-specific mortality among women with endometrial cancer. *Gynecologic Oncology*, **2018**, 150, 99-105	4.9	32

232	A Transcriptome-Wide Association Study Among 97,898 Women to Identify Candidate Susceptibility Genes for Epithelial Ovarian Cancer Risk. *Cancer Research*, **2018**, 78, 5419-5430	10.1	32
231	Evaluation of CADD Scores in Curated Mismatch Repair Gene Variants Yields a Model for Clinical Validation and Prioritization. *Human Mutation*, **2015**, 36, 712-9	4.7	32
230	Association of the variants CASP8 D302H and CASP10 V410I with breast and ovarian cancer risk in BRCA1 and BRCA2 mutation carriers. *Cancer Epidemiology Biomarkers and Prevention*, **2010**, 19, 2859-68	4	32
229	The microsomal epoxide hydrolase Tyr113His polymorphism: association with risk of ovarian cancer. *Molecular Carcinogenesis*, **2001**, 30, 71-8	5	32
228	Androgen receptor exon 1 cag repeat length and risk of ovarian cancer. *International Journal of Cancer*, **2000**, 87, 637-643	7.5	32
227	Ovarian cancer susceptibility alleles and risk of ovarian cancer in BRCA1 and BRCA2 mutation carriers. *Human Mutation*, **2012**, 33, 690-702	4.7	31
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