Amanda E Toland

List of Publications by Year in Descending Order

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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.

175 papers	10,299 citations	49 h-index	98 g-index
192 ext. papers	12,584 ext. citations	8.6 avg, IF	5 L-index

#	Paper	IF	Citations
175	Cancer Risks Associated With and Pathogenic Variants.. *Journal of Clinical Oncology*, **2022**, JCO2102112	2.2	7
174	Genetic analysis of a malignant meningioma and associated metastases.. *Acta Neurochirurgica*, **2022**, 1	3	1
173	Differences in somatic TP53 mutation type in breast tumors by race and receptor status.. *Breast Cancer Research and Treatment*, **2022**, 192, 639	4.4	0
172	loss drives aggressive tumor phenotypes in cutaneous squamous cell carcinoma.. *American Journal of Cancer Research*, **2022**, 12, 1309-1322	4.4	
171	Risks of breast and ovarian cancer for women harboring pathogenic missense variants in BRCA1 and BRCA2 compared with those harboring protein truncating variants.. *Genetics in Medicine*, **2021**,	8.1	2
170	The influence of sex, age and sunlight exposure on mutational processes in melanoma. *British Journal of Dermatology*, **2021**, 184, 197-198	4	
169	Machine learning approaches reveal subtle differences in breathing and sleep fragmentation in -derived astrocytes ablated mice. *Journal of Neurophysiology*, **2021**, 125, 1164-1179	3.2	1
168	Oncogenetic network estimation with disjunctive Bayesian networks. *Computational and Systems Oncology*, **2021**, 1, e1027	1	
167	Association Between Smoking and Molecular Subtypes of Colorectal Cancer. *JNCI Cancer Spectrum*, **2021**, 5, pkab056	4.6	2
166	The predictive ability of the 313 variant-based polygenic risk score for contralateral breast cancer risk prediction in women of European ancestry with a heterozygous BRCA1 or BRCA2 pathogenic variant. *Genetics in Medicine*, **2021**, 23, 1726-1737	8.1	2
165	Functional annotation of the 2q35 breast cancer risk locus implicates a structural variant in influencing activity of a long-range enhancer element. *American Journal of Human Genetics*, **2021**, 108, 1190-1203	11	1
164	The p.Ser64Leu and p.Pro104Leu missense variants of PALB2 identified in familial pancreatic cancer patients compromise the DNA damage response. *Human Mutation*, **2021**, 42, 150-163	4.7	
163	A case-only study to identify genetic modifiers of breast cancer risk for BRCA1/BRCA2 mutation carriers. *Nature Communications*, **2021**, 12, 1078	17.4	4
162	Genetic architectures of proximal and distal colorectal cancer are partly distinct. *Gut*, **2021**, 70, 1325-1334	19.2	7
161	Albert de la Chapelle (1933-2020). *American Journal of Human Genetics*, **2021**, 108, 214-216	11	
160	Breast and Prostate Cancer Risks for Male BRCA1 and BRCA2 Pathogenic Variant Carriers Using Polygenic Risk Scores. *Journal of the National Cancer Institute*, **2021**,	9.7	3
159	Polygenic Risk Scores in Prostate Cancer Risk Assessment and Screening. *Urologic Clinics of North America*, **2021**, 48, 387-399	2.9	1

158	Impact of Previous Genetic Counseling and Objective Numeracy on Accurate Interpretation of a Pharmacogenetics Test Report. *Public Health Genomics*, **2021**, 24, 26-32	1.9	4
157	Genome-wide association study identifies 32 novel breast cancer susceptibility loci from overall and subtype-specific analyses. *Nature Genetics*, **2020**, 52, 572-581	36.3	76
156	MicroRNA Expression Profiling of Cutaneous Squamous Cell Carcinomas Arising in Different Sites. *Otolaryngology - Head and Neck Surgery*, **2020**, 163, 538-545	5.5	2
155	Metastatic breast cancer patient perceptions of somatic tumor genomic testing. *BMC Cancer*, **2020**, 20, 389	4.8	2
154	Implementation of Germline Testing for Prostate Cancer: Philadelphia Prostate Cancer Consensus Conference 2019. *Journal of Clinical Oncology*, **2020**, 38, 2798-2811	2.2	80
153	Maternal age at delivery and fertility of the next generation. *Paediatric and Perinatal Epidemiology*, **2020**, 34, 629-636	2.7	2
152	Characterization of the Cancer Spectrum in Men With Germline BRCA1 and BRCA2 Pathogenic Variants: Results From the Consortium of Investigators of Modifiers of BRCA1/2 (CIMBA). *JAMA Oncology*, **2020**, 6, 1218-1230	13.4	25
151	Transcriptome-wide association study of breast cancer risk by estrogen-receptor status. *Genetic Epidemiology*, **2020**, 44, 442-468	2.6	9
150	Association of Genomic Domains in and with Prostate Cancer Risk and Aggressiveness. *Cancer Research*, **2020**, 80, 624-638	10.1	22
149	Genome-wide meta-analysis identifies eight new susceptibility loci for cutaneous squamous cell carcinoma. *Nature Communications*, **2020**, 11, 820	17.4	13
148	Developing risk prediction models for melanoma: balancing better predictive value with ease of clinical implementation. *British Journal of Dermatology*, **2020**, 182, 1089-1090	4	
147	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. *Nature Genetics*, **2020**, 52, 56-73	36.3	56
146	Cumulative Burden of Colorectal Cancer-Associated Genetic Variants Is More Strongly Associated With Early-Onset vs Late-Onset Cancer. *Gastroenterology*, **2020**, 158, 1274-1286.e12	13.3	47
145	Sequencing technology status of 2 testing in Latin American Countries. *Npj Genomic Medicine*, **2020**, 5, 22	6.2	0
144	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
143	Intake of Dietary Fruit, Vegetables, and Fiber and Risk of Colorectal Cancer According to Molecular Subtypes: A Pooled Analysis of 9 Studies. *Cancer Research*, **2020**, 80, 4578-4590	10.1	8
142	Two truncating variants in FANCC and breast cancer risk. *Scientific Reports*, **2019**, 9, 12524	4.9	2
141	Shared heritability and functional enrichment across six solid cancers. *Nature Communications*, **2019**, 10, 431	17.4	45

140	F-Box Protein-Mediated Resistance to PARP Inhibitor Therapy. *Molecular Cell*, **2019**, 73, 195-196	17.6	3
139	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
138	Functional Loss Defines a Targetable Subset in Leiomyosarcoma. *Oncologist*, **2019**, 24, 973-979	5.7	23
137	Mendelian randomisation study of height and body mass index as modifiers of ovarian cancer risk in 22,588 BRCA1 and BRCA2 mutation carriers. *British Journal of Cancer*, **2019**, 121, 180-192	8.7	13
136	BRCA1 and BRCA2 pathogenic sequence variants in women of African origin or ancestry. *Human Mutation*, **2019**, 40, 1781-1796	4.7	16
135	Germline Variants Impact Somatic Events during Tumorigenesis. *Trends in Genetics*, **2019**, 35, 515-526	8.5	16
134	Understanding Mutation CarriersPPreferences for Communication of Genetic Modifiers of Breast Cancer Risk. *Journal of Health Communication*, **2019**, 24, 377-384	2.5	7
133	Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. *Nature Communications*, **2019**, 10, 1741	17.4	47
132	Genetic Testing to Guide Risk-Stratified Screens for Breast Cancer. *Journal of Personalized Medicine*, **2019**, 9,	3.6	12
131	Genome-wide association studies and polygenic risk scores for skin cancer: clinically useful yet?. *British Journal of Dermatology*, **2019**, 181, 1146-1155	4	24
130	Exploring genetic counselorsPperceptions of usefulness and intentions to use refined risk models in clinical care based on the Technology Acceptance Model (TAM). *Journal of Genetic Counseling*, **2019**, 28, 664-672	2.5	2
129	Single Nucleotide Polymorphisms in Carotene Oxygenase 1 are Associated with Plasma Lycopene Responses to a Tomato-Soy Juice Intervention in Men with Prostate Cancer. *Journal of Nutrition*, **2019**, 149, 381-397	4.1	20
128	POT1 pathogenic variants: not all telomere pathway genes are equal in risk of hereditary cutaneous melanoma. *British Journal of Dermatology*, **2019**, 181, 14-15	4	
127	The :p.Arg658* truncating variant is associated with risk of triple-negative breast cancer. *Npj Breast Cancer*, **2019**, 5, 38	7.8	12
126	Keratinocyte Carcinomas: Current Concepts and Future Research Priorities. *Clinical Cancer Research* , **2019**, 25, 2379-2391	12.9	55
125	Lessons learned from two decades of BRCA1 and BRCA2 genetic testing: the evolution of data sharing and variant classification. *Genetics in Medicine*, **2019**, 21, 1476-1480	8.1	2
124	Height and Body Mass Index as Modifiers of Breast Cancer Risk in BRCA1/2 Mutation Carriers: A Mendelian Randomization Study. *Journal of the National Cancer Institute*, **2019**, 111, 350-364	9.7	22
123	Discovery of common and rare genetic risk variants for colorectal cancer. *Nature Genetics*, **2019**, 51, 76-87	36.3	177

122	Polygenic risk scores for prostate cancer: testing considerations. *Canadian Journal of Urology*, **2019**, 26, 17-18	0.8	3
121	The BRCA2 c.68-7T>A variant is not pathogenic: A model for clinical calibration of spliceogenicity. *Human Mutation*, **2018**, 39, 729-741	4.7	16
120	Operationalizing the Reciprocal Engagement Model of Genetic Counseling Practice: a Framework for the Scalable Delivery of Genomic Counseling and Testing. *Journal of Genetic Counseling*, **2018**, 27, 1111-1129	2.5	16
119	Clinical testing of and : a worldwide snapshot of technological practices. *Npj Genomic Medicine*, **2018**, 3, 7	6.2	29
118	Mutational spectrum in a worldwide study of 29,700 families with BRCA1 or BRCA2 mutations. *Human Mutation*, **2018**, 39, 593-620	4.7	138
117	Involvement and Influence of Healthcare Providers, Family Members, and Other Mutation Carriers in the Cancer Risk Management Decision-Making Process of BRCA1 and BRCA2 Mutation Carriers. *Journal of Genetic Counseling*, **2018**, 27, 1291-1301	2.5	3
116	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
115	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
114	Early Outcome Data Assessing Utility of a Post-Test Genomic Counseling Framework for the Scalable Delivery of Precision Health. *Journal of Personalized Medicine*, **2018**, 8,	3.6	3
113	Association of BRCA2 K3326* With Small Cell Lung Cancer and Squamous Cell Cancer of the Skin. *Journal of the National Cancer Institute*, **2018**, 110, 967-974	9.7	16
112	EMR documentation of physician-patient communication following genomic counseling for actionable complex disease and pharmacogenomic results. *Clinical Genetics*, **2017**, 91, 545-556	4	7
111	Variants at the OCA2/HERC2 locus affect time to first cutaneous squamous cell carcinoma in solid organ transplant recipients collected using two different study designs. *British Journal of Dermatology*, **2017**, 177, 1066-1073	4	8
110	Differential mutation frequencies in metastatic cutaneous squamous cell carcinomas versus primary tumors. *Cancer*, **2017**, 123, 1184-1193	6.4	49
109	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. *Nature Genetics*, **2017**, 49, 680-691	36.3	190
108	Outcomes of a Randomized Controlled Trial of Genomic Counseling for Patients Receiving Personalized and Actionable Complex Disease Reports. *Journal of Genetic Counseling*, **2017**, 26, 980-998	2.5	11
107	CounseleesPPerspectives of Genomic Counseling Following Online Receipt of Multiple Actionable Complex Disease and Pharmacogenomic Results: a Qualitative Research Study. *Journal of Genetic Counseling*, **2017**, 26, 738-751	2.5	11
106	Epidemiology of keratinocyte carcinomas after organ transplantation. *British Journal of Dermatology*, **2017**, 177, 1208-1216	4	39
105	Risk prediction tools for keratinocyte carcinoma after solid organ transplantation: a review of the literature. *British Journal of Dermatology*, **2017**, 177, 1202-1207	4	15

104	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. *Nature Genetics*, **2017**, 49, 1767-1778	36.3	186
103	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
102	DNA repair-related functional assays for the classification of BRCA1 and BRCA2 variants: a critical review and needs assessment. *Journal of Medical Genetics*, **2017**, 54, 721-731	5.8	24
101	The pathogenesis of cutaneous squamous cell carcinoma in organ transplant recipients. *British Journal of Dermatology*, **2017**, 177, 1217-1224	4	37
100	Association of breast cancer risk in BRCA1 and BRCA2 mutation carriers with genetic variants showing differential allelic expression: identification of a modifier of breast cancer risk at locus 11q22.3. *Breast Cancer Research and Treatment*, **2017**, 161, 117-134	4.4	15
99	IRF4 Polymorphism Is Associated with Cutaneous Squamous Cell Carcinoma in Organ Transplant Recipients: A Pigment-Independent Phenomenon. *Journal of Investigative Dermatology*, **2017**, 137, 251-253	4.3	11
98	An intergenic risk locus containing an enhancer deletion in 2q35 modulates breast cancer risk by deregulating IGFBP5 expression. *Human Molecular Genetics*, **2016**, 25, 3863-3876	5.6	24
97	Inheritance of deleterious mutations at both BRCA1 and BRCA2 in an international sample of 32,295 women. *Breast Cancer Research*, **2016**, 18, 112	8.3	25
96	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. *Nature Communications*, **2016**, 7, 11375	17.4	64
95	Functional mechanisms underlying pleiotropic risk alleles at the 19p13.1 breast-ovarian cancer susceptibility locus. *Nature Communications*, **2016**, 7, 12675	17.4	53
94	MicroRNA expression profiling in metastatic cutaneous squamous cell carcinoma. *Journal of the European Academy of Dermatology and Venereology*, **2016**, 30, 1043-5	4.6	14
93	Breast cancer risk variants at 6q25 display different phenotype associations and regulate ESR1, RMND1 and CCDC170. *Nature Genetics*, **2016**, 48, 374-86	36.3	93
92	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	11.6	80
91	Fine-Scale Mapping at 9p22.2 Identifies Candidate Causal Variants That Modify Ovarian Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. *PLoS ONE*, **2016**, 11, e0158801	3.7	7
90	Fine-scale mapping of 8q24 locus identifies multiple independent risk variants for breast cancer. *International Journal of Cancer*, **2016**, 139, 1303-1317	7.5	26
89	PALB2, CHEK2 and ATM rare variants and cancer risk: data from COGS. *Journal of Medical Genetics*, **2016**, 53, 800-811	5.8	121
88	Identification of independent association signals and putative functional variants for breast cancer risk through fine-scale mapping of the 12p11 locus. *Breast Cancer Research*, **2016**, 18, 64	8.3	25
87	Variants in an Hdac9 intronic enhancer plasmid impact Twist1 expression in vitro. *Mammalian Genome*, **2016**, 27, 99-110	3.2	3

86	Association of genetic susceptibility variants for type 2 diabetes with breast cancer risk in women of European ancestry. *Cancer Causes and Control*, **2016**, 27, 679-93	2.8	15
85	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	8.3	58
84	High risk cutaneous squamous cell carcinoma of the head and neck. *World Journal of Otorhinolaryngology - Head and Neck Surgery*, **2016**, 2, 136-140	2.6	13
83	Inherited variants in the inner centromere protein (INCENP) gene of the chromosomal passenger complex contribute to the susceptibility of ER-negative breast cancer. *Carcinogenesis*, **2015**, 36, 256-71	4.6	12
82	Identification of six new susceptibility loci for invasive epithelial ovarian cancer. *Nature Genetics*, **2015**, 47, 164-71	36.3	177
81	Genome-wide association analysis of more than 120,000 individuals identifies 15 new susceptibility loci for breast cancer. *Nature Genetics*, **2015**, 47, 373-80	36.3	406
80	Haploinsufficiency for BRCA1 leads to cell-type-specific genomic instability and premature senescence. *Nature Communications*, **2015**, 6, 7505	17.4	74
79	Polymorphisms in a Putative Enhancer at the 10q21.2 Breast Cancer Risk Locus Regulate NRBF2 Expression. *American Journal of Human Genetics*, **2015**, 97, 22-34	11	26
78	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
77	Large-scale genomic analyses link reproductive aging to hypothalamic signaling, breast cancer susceptibility and BRCA1-mediated DNA repair. *Nature Genetics*, **2015**, 47, 1294-1303	36.3	226
76	Height and Breast Cancer Risk: Evidence From Prospective Studies and Mendelian Randomization. *Journal of the National Cancer Institute*, **2015**, 107,	9.7	74
75	Fine-scale mapping of the 4q24 locus identifies two independent loci associated with breast cancer risk. *Cancer Epidemiology Biomarkers and Prevention*, **2015**, 24, 1680-91	4	17
74	Identification and characterization of novel associations in the CASP8/ALS2CR12 region on chromosome 2 with breast cancer risk. *Human Molecular Genetics*, **2015**, 24, 285-98	5.6	35
73	Candidate genetic modifiers for breast and ovarian cancer risk in BRCA1 and BRCA2 mutation carriers. *Cancer Epidemiology Biomarkers and Prevention*, **2015**, 24, 308-16	4	20
72	An original phylogenetic approach identified mitochondrial haplogroup T1a1 as inversely associated with breast cancer risk in BRCA2 mutation carriers. *Breast Cancer Research*, **2015**, 17, 61	8.3	16
71	Functional Analysis of BARD1 Missense Variants in Homology-Directed Repair of DNA Double Strand Breaks. *Human Mutation*, **2015**, 36, 1205-14	4.7	23
70	Allele-specific imbalance mapping at human orthologs of mouse susceptibility to colon cancer (Scc) loci. *International Journal of Cancer*, **2015**, 137, 2323-31	7.5	4
69	Assessing associations between the AURKA-HMMR-TPX2-TUBG1 functional module and breast cancer risk in BRCA1/2 mutation carriers. *PLoS ONE*, **2015**, 10, e0120020	3.7	26

68	Prediction of breast cancer risk based on profiling with common genetic variants. *Journal of the National Cancer Institute*, **2015**, 107,	9.7	324
67	Use of Whole Genome Sequencing for Diagnosis and Discovery in the Cancer Genetics Clinic. *EBioMedicine*, **2015**, 2, 74-81	8.8	43
66	Germline mutation in BRCA1 or BRCA2 and ten-year survival for women diagnosed with epithelial ovarian cancer. *Clinical Cancer Research*, **2015**, 21, 652-7	12.9	107
65	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, 304-11	2.2	435
64	Fine-mapping identifies two additional breast cancer susceptibility loci at 9q31.2. *Human Molecular Genetics*, **2015**, 24, 2966-84	5.6	36
63	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	11	59
62	A large-scale assessment of two-way SNP interactions in breast cancer susceptibility using 46,450 cases and 42,461 controls from the breast cancer association consortium. *Human Molecular Genetics*, **2014**, 23, 1934-46	5.6	28
61	Linking distant relatives with BRCA gene mutations: potential for cost savings. *Clinical Genetics*, **2014**, 85, 54-8	4	4
60	Genome-wide association study identifies 25 known breast cancer susceptibility loci as risk factors for triple-negative breast cancer. *Carcinogenesis*, **2014**, 35, 1012-9	4.6	121
59	Allele-specific imbalance mapping identifies HDAC9 as a candidate gene for cutaneous squamous cell carcinoma. *International Journal of Cancer*, **2014**, 134, 244-8	7.5	14
58	FGF receptor genes and breast cancer susceptibility: results from the Breast Cancer Association Consortium. *British Journal of Cancer*, **2014**, 110, 1088-100	8.7	20
57	Breast-cancer risk in families with mutations in PALB2. *New England Journal of Medicine*, **2014**, 371, 497-506	59.2	576
56	Evidence that breast cancer risk at the 2q35 locus is mediated through IGFBP5 regulation. *Nature Communications*, **2014**, 4, 4999	17.4	87
55	Genetic variation in mitotic regulatory pathway genes is associated with breast tumor grade. *Human Molecular Genetics*, **2014**, 23, 6034-46	5.6	11
54	Genetic variation at CYP3A is associated with age at menarche and breast cancer risk: a case-control study. *Breast Cancer Research*, **2014**, 16, R51	8.3	12
53	MicroRNA related polymorphisms and breast cancer risk. *PLoS ONE*, **2014**, 9, e109973	3.7	37
52	Design and implementation of a randomized controlled trial of genomic counseling for patients with chronic disease. *Journal of Personalized Medicine*, **2014**, 4, 1-19	3.6	17
51	Melanoma incidence rates in active duty military personnel compared with a population-based registry in the United States, 2000-2007. *Military Medicine*, **2014**, 179, 247-53	1.3	13

50	DNA glycosylases involved in base excision repair may be associated with cancer risk in BRCA1 and BRCA2 mutation carriers. *PLoS Genetics*, **2014**, 10, e1004256	6	33
49	Expression of cancer-testis antigens MAGEA1, MAGEA3, ACRBP, PRAME, SSX2, and CTAG2 in myxoid and round cell liposarcoma. *Modern Pathology*, **2014**, 27, 1238-45	9.8	29
48	Common non-synonymous SNPs associated with breast cancer susceptibility: findings from the Breast Cancer Association Consortium. *Human Molecular Genetics*, **2014**, 23, 6096-111	5.6	48
47	mrSNP: software to detect SNP effects on microRNA binding. *BMC Bioinformatics*, **2014**, 15, 73	3.6	45
46	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
45	Salivary gland cancer in BRCA-positive families: a retrospective review. *JAMA Otolaryngology - Head and Neck Surgery*, **2014**, 140, 1213-7	3.9	19
44	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
43	Benchmarking short sequence mapping tools. *BMC Bioinformatics*, **2013**, 14, 184	3.6	140
42	Analysis of BRCA1 variants in double-strand break repair by homologous recombination and single-strand annealing. *Human Mutation*, **2013**, 34, 439-45	4.7	41
41	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	11	167
40	The cancer-testis antigen NY-ESO-1 is highly expressed in myxoid and round cell subset of liposarcomas. *Modern Pathology*, **2013**, 26, 282-8	9.8	33
39	Large-scale genotyping identifies 41 new loci associated with breast cancer risk. *Nature Genetics*, **2013**, 45, 353-61, 361e1-2	36.3	813
38	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
37	The impact of 3′UTR variants on differential expression of candidate cancer susceptibility genes. *PLoS ONE*, **2013**, 8, e58609	3.7	32
36	Differential expression of miR-1, a putative tumor suppressing microRNA, in cancer resistant and cancer susceptible mice. *PeerJ*, **2013**, 1, e68	3.1	18
35	Interaction Between Genetics and Epigenetics in Cancer **2013**, 209-229		
34	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
33	Ovarian cancer susceptibility alleles and risk of ovarian cancer in BRCA1 and BRCA2 mutation carriers. *Human Mutation*, **2012**, 33, 690-702	4.7	31

32	Association between BRCA1 and BRCA2 mutations and survival in women with invasive epithelial ovarian cancer. *JAMA - Journal of the American Medical Association*, **2012**, 307, 382-90	27.4	427
31	A nonsynonymous polymorphism in IRS1 modifies risk of developing breast and ovarian cancers in BRCA1 and ovarian cancer in BRCA2 mutation carriers. *Cancer Epidemiology Biomarkers and Prevention*, **2012**, 21, 1362-70	4	20
30	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
29	The role for oxidative stress in aberrant DNA methylation in Alzheimer's disease. *Current Alzheimer Research*, **2012**, 9, 1077-96	3	20
28	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
27	Evaluation of allele-specific somatic changes of genome-wide association study susceptibility alleles in human colorectal cancers. *PLoS ONE*, **2012**, 7, e37672	3.7	6
26	Aberrant Epigenetic Regulation in Breast Cancer **2012**, 91-122		2
25	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
24	Haplotype structure in Ashkenazi Jewish BRCA1 and BRCA2 mutation carriers. *Human Genetics*, **2011**, 130, 685-99	6.3	15
23	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
22	The combined influence of oral contraceptives and human papillomavirus virus on cutaneous squamous cell carcinoma. *Clinical Medicine Insights: Oncology*, **2011**, 5, 55-75	1.8	3
21	Common variants of the BRCA1 wild-type allele modify the risk of breast cancer in BRCA1 mutation carriers. *Human Molecular Genetics*, **2011**, 20, 4732-47	5.6	21
20	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
19	Germline variation controls the architecture of somatic alterations in tumors. *PLoS Genetics*, **2010**, 6, e1001136	6	31
18	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
17	Identification of breast tumor mutations in BRCA1 that abolish its function in homologous DNA recombination. *Cancer Research*, **2010**, 70, 988-95	10.1	92
16	Common genetic variants and modification of penetrance of BRCA2-associated breast cancer. *PLoS Genetics*, **2010**, 6, e1001183	6	74
15	Characterization of BRCA1 ring finger variants of uncertain significance. *Breast Cancer Research and Treatment*, **2010**, 119, 737-43	4.4	24

14	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
13	Epigenetic alterations in the breast: Implications for breast cancer detection, prognosis and treatment. *Seminars in Cancer Biology*, **2009**, 19, 165-71	12.7	120
12	Chromosomal aberrations in UVB-induced tumors of immunosuppressed mice. *Genes Chromosomes and Cancer*, **2009**, 48, 490-501	5	5
11	Methylation not a frequent "second hit" in tumors with germline BRCA mutations. *Familial Cancer*, **2009**, 8, 339-46	3	49
10	Merkel cell polyomavirus in cutaneous squamous cell carcinoma of immunocompetent individuals. *Journal of Investigative Dermatology*, **2009**, 129, 2868-74	4.3	78
9	Evaluation of a candidate breast cancer associated SNP in ERCC4 as a risk modifier in BRCA1 and BRCA2 mutation carriers. Results from the Consortium of Investigators of Modifiers of BRCA1/BRCA2 (CIMBA). *British Journal of Cancer*, **2009**, 101, 2048-54	8.7	13
8	Sequence divergence of Mus spretus and Mus musculus across a skin cancer susceptibility locus. *BMC Genomics*, **2008**, 9, 626	4.5	15
7	Clinically applicable models to characterize BRCA1 and BRCA2 variants of uncertain significance. *Journal of Clinical Oncology*, **2008**, 26, 5393-400	2.2	74
6	PTPRJ haplotypes and colorectal cancer risk. *Cancer Epidemiology Biomarkers and Prevention*, **2008**, 17, 2782-5	4	15
5	The role of parental and grandparental epigenetic alterations in familial cancer risk. *Cancer Research*, **2008**, 68, 9116-21	10.1	29
4	Lack of germ-line promoter methylation in BRCA1-negative families with familial breast cancer. *Genetic Testing and Molecular Biomarkers*, **2006**, 10, 281-4		22
3	Elastin point mutations cause an obstructive vascular disease, supravalvular aortic stenosis. *Human Molecular Genetics*, **1997**, 6, 1021-8	5.6	188
2	Polygenic Risk Modelling for Prediction of Epithelial Ovarian Cancer Risk		1
1	Genome-wide association study identifies 32 novel breast cancer susceptibility loci from overall and subtype-specific analyses		2