

David E Goldgar

List of Publications by Year in descending order

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Version: 2024-02-01

166
papers

28,216
citations

11908

72
h-index

6686

161
g-index

174
all docs

174
docs citations

174
times ranked

23873
citing authors

#	ARTICLE	IF	CITATIONS
1	First international workshop of the ATM and cancer risk group (4-5 December 2019). <i>Familial Cancer</i> , 2022, 21, 211-227.	0.9	10
2	Breast and Prostate Cancer Risks for Male <i>BRCA1</i> and <i>BRCA2</i> Pathogenic Variant Carriers Using Polygenic Risk Scores. <i>Journal of the National Cancer Institute</i> , 2022, 114, 109-122.	3.0	19
3	Risks of breast and ovarian cancer for women harboring pathogenic missense variants in <i>BRCA1</i> and <i>BRCA2</i> compared with those harboring protein truncating variants. <i>Genetics in Medicine</i> , 2022, 24, 119-129.	1.1	10
4	Cancer Risks Associated With <i>BRCA1</i> and <i>BRCA2</i> Pathogenic Variants. <i>Journal of Clinical Oncology</i> , 2022, 40, 1529-1541.	0.8	90
5	Oral Contraceptive Use in <i>BRCA1</i> and <i>BRCA2</i> Mutation Carriers: Absolute Cancer Risks and Benefits. <i>Journal of the National Cancer Institute</i> , 2022, 114, 540-552.	3.0	7
6	An integrative model for the comprehensive classification of <i>BRCA1</i> and <i>BRCA2</i> variants of uncertain clinical significance. <i>Npj Genomic Medicine</i> , 2022, 7, .	1.7	4
7	Comprehensive evaluation and efficient classification of <i>BRCA1</i> RING domain missense substitutions. <i>American Journal of Human Genetics</i> , 2022, 109, 1153-1174.	2.6	6
8	Comparing 5-Year and Lifetime Risks of Breast Cancer Using the Prospective Family Study Cohort. <i>Journal of the National Cancer Institute</i> , 2021, 113, 785-791.	3.0	13
9	Integration of functional assay data results provides strong evidence for classification of hundreds of <i>BRCA1</i> variants of uncertain significance. <i>Genetics in Medicine</i> , 2021, 23, 306-315.	1.1	21
10	A case-only study to identify genetic modifiers of breast cancer risk for <i>BRCA1/BRCA2</i> mutation carriers. <i>Nature Communications</i> , 2021, 12, 1078.	5.8	19
11	A Population-Based Study of Genes Previously Implicated in Breast Cancer. <i>New England Journal of Medicine</i> , 2021, 384, 440-451.	13.9	414
12	Strong functional data for pathogenicity or neutrality classify <i>BRCA2</i> DNA-binding-domain variants of uncertain significance. <i>American Journal of Human Genetics</i> , 2021, 108, 458-468.	2.6	31
13	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 <i>BRCA1</i> or <i>BRCA2</i> pathogenic variant. <i>Genetics in Medicine</i> , 2021, 23, 1726-1737.	1.1	16
14	Risk of Late-Onset Breast Cancer in Genetically Predisposed Women. <i>Journal of Clinical Oncology</i> , 2021, 39, 3430-3440.	0.8	21
15	Oral contraceptive use and ovarian cancer risk for <i>BRCA1/2</i> mutation carriers: an international cohort study. <i>American Journal of Obstetrics and Gynecology</i> , 2021, 225, 51.e1-51.e17.	0.7	34
16	An updated quantitative model to classify missense variants in the <i>TP53</i> gene: A novel multifactorial strategy. <i>Human Mutation</i> , 2021, 42, 1351-1361.	1.1	7
17	Risk of Breast Cancer Among Carriers of Pathogenic Variants in Breast Cancer Predisposition Genes Varies by Polygenic Risk Score. <i>Journal of Clinical Oncology</i> , 2021, 39, 2564-2573.	0.8	47
18	Mendelian randomisation study of smoking exposure in relation to breast cancer risk. <i>British Journal of Cancer</i> , 2021, 125, 1135-1145.	2.9	9

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19	Racial and Ethnic Differences in Multigene Hereditary Cancer Panel Test Results for Women With Breast Cancer. <i>Journal of the National Cancer Institute</i> , 2021, 113, 1429-1433.	3.0	18
20	Germline Pathogenic Variants in Cancer Predisposition Genes Among Women With Invasive Lobular Carcinoma of the Breast. <i>Journal of Clinical Oncology</i> , 2021, 39, 3918-3926.	0.8	22
21	Association of Genomic Domains in <i>BRCA1</i> and <i>BRCA2</i> with Prostate Cancer Risk and Aggressiveness. <i>Cancer Research</i> , 2020, 80, 624-638.	0.4	39
22	Considerations When Using Breast Cancer Risk Models for Women with Negative <i>BRCA1/BRCA2</i> Mutation Results. <i>Journal of the National Cancer Institute</i> , 2020, 112, 418-422.	3.0	1
23	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.	1.1	136
24	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. <i>Nature Genetics</i> , 2020, 52, 56-73.	9.4	120
25	Cancer Risks Associated With Germline <i>PALB2</i> Pathogenic Variants: An International Study of 524 Families. <i>Journal of Clinical Oncology</i> , 2020, 38, 674-685.	0.8	270
26	Classification of variants of uncertain significance in <i>BRCA1</i> and <i>BRCA2</i> using personal and family history of cancer from individuals in a large hereditary cancer multigene panel testing cohort. <i>Genetics in Medicine</i> , 2020, 22, 701-708.	1.1	28
27	Differences in patient ascertainment affect the use of gene-specific ACMG/AMP phenotype-related variant classification criteria: Evidence for <i>TP53</i> . <i>Human Mutation</i> , 2020, 41, 537-542.	1.1	5
28	Polygenic risk scores and breast and epithelial ovarian cancer risks for carriers of <i>BRCA1</i> and <i>BRCA2</i> pathogenic variants. <i>Genetics in Medicine</i> , 2020, 22, 1653-1666.	1.1	82
29	Considerations in assessing germline variant pathogenicity using cosegregation analysis. <i>Genetics in Medicine</i> , 2020, 22, 2052-2059.	1.1	15
30	Breast Cancer Polygenic Risk Score and Contralateral Breast Cancer Risk. <i>American Journal of Human Genetics</i> , 2020, 107, 837-848.	2.6	39
31	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.	9.4	265
32	Contribution of Germline Predisposition Gene Mutations to Breast Cancer Risk in African American Women. <i>Journal of the National Cancer Institute</i> , 2020, 112, 1213-1221.	3.0	51
33	Germline <i>HOXB13</i> mutations p.G84E and p.R217C do not confer an increased breast cancer risk. <i>Scientific Reports</i> , 2020, 10, 9688.	1.6	2
34	Characterization of the Cancer Spectrum in Men With Germline <i>BRCA1</i> and <i>BRCA2</i> Pathogenic Variants. <i>JAMA Oncology</i> , 2020, 6, 1218.	3.4	48
35	Radiation Treatment, <i>ATM</i> , <i>BRCA1/2</i> , and <i>CHEK2</i> *1100delC Pathogenic Variants and Risk of Contralateral Breast Cancer. <i>Journal of the National Cancer Institute</i> , 2020, 112, 1275-1279.	3.0	21
36	The Contribution of Germline Predisposition Gene Mutations to Clinical Subtypes of Invasive Breast Cancer From a Clinical Genetic Testing Cohort. <i>Journal of the National Cancer Institute</i> , 2020, 112, 1231-1241.	3.0	61

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37	Transcriptome-wide association study of breast cancer risk by estrogen receptor status. <i>Genetic Epidemiology</i> , 2020, 44, 442-468.	0.6	32
38	Alcohol Consumption, Cigarette Smoking, and Risk of Breast Cancer for <i>BRCA1</i> and <i>BRCA2</i> Mutation Carriers: Results from The <i>BRCA1</i> and <i>BRCA2</i> Cohort Consortium. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2020, 29, 368-378.	1.1	24
39	A network analysis to identify mediators of germline-driven differences in breast cancer prognosis. <i>Nature Communications</i> , 2020, 11, 312.	5.8	30
40	Risk-reducing salpingo-oophorectomy, natural menopause, and breast cancer risk: an international prospective cohort of <i>BRCA1</i> and <i>BRCA2</i> mutation carriers. <i>Breast Cancer Research</i> , 2020, 22, 8.	2.2	41
41	Mutation prevalence tables for hereditary cancer derived from multigene panel testing. <i>Human Mutation</i> , 2020, 41, e1-e6.	1.1	19
42	Comprehensive annotation of <i>BRCA1</i> and <i>BRCA2</i> missense variants by functionally validated sequence-based computational prediction models. <i>Genetics in Medicine</i> , 2019, 21, 71-80.	1.1	52
43	Assessment of blind predictions of the clinical significance of <i>BRCA1</i> and <i>BRCA2</i> variants. <i>Human Mutation</i> , 2019, 40, 1546-1556.	1.1	19
44	The <i>FANCM</i> :p.Arg658* truncating variant is associated with risk of triple-negative breast cancer. <i>Npj Breast Cancer</i> , 2019, 5, 38.	2.3	28
45	Two truncating variants in <i>FANCC</i> and breast cancer risk. <i>Scientific Reports</i> , 2019, 9, 12524.	1.6	5
46	Shared heritability and functional enrichment across six solid cancers. <i>Nature Communications</i> , 2019, 10, 431.	5.8	88
47	Large scale multifactorial likelihood quantitative analysis of <i>BRCA1</i> and <i>BRCA2</i> variants: An ENIGMA resource to support clinical variant classification. <i>Human Mutation</i> , 2019, 40, 1557-1578.	1.1	102
48	Mendelian randomisation study of height and body mass index as modifiers of ovarian cancer risk in 22,588 <i>BRCA1</i> and <i>BRCA2</i> mutation carriers. <i>British Journal of Cancer</i> , 2019, 121, 180-192.	2.9	19
49	Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. <i>Nature Communications</i> , 2019, 10, 1741.	5.8	90
50	Genome-wide association study of germline variants and breast cancer-specific mortality. <i>British Journal of Cancer</i> , 2019, 120, 647-657.	2.9	52
51	10-year performance of four models of breast cancer risk: a validation study. <i>Lancet Oncology</i> , The, 2019, 20, 504-517.	5.1	116
52	Polygenic Risk Scores for Prediction of Breast Cancer and Breast Cancer Subtypes. <i>American Journal of Human Genetics</i> , 2019, 104, 21-34.	2.6	711
53	Height and Body Mass Index as Modifiers of Breast Cancer Risk in <i>BRCA1</i> / <i>BRCA2</i> Mutation Carriers: A Mendelian Randomization Study. <i>Journal of the National Cancer Institute</i> , 2019, 111, 350-364.	3.0	30
54	A functional assay-based procedure to classify mismatch repair gene variants in Lynch syndrome. <i>Genetics in Medicine</i> , 2019, 21, 1486-1496.	1.1	36

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55	The <i>BRCA2</i> c.68-7T> variant is not pathogenic: A model for clinical calibration of spliceogenicity. <i>Human Mutation</i> , 2018, 39, 729-741.	1.1	19
56	Mutational spectrum in a worldwide study of 29,700 families with <i>BRCA1</i> or <i>BRCA2</i> mutations. <i>Human Mutation</i> , 2018, 39, 593-620.	1.1	224
57	Assessment of the Clinical Relevance of BRCA2 Missense Variants by Functional and Computational Approaches. <i>American Journal of Human Genetics</i> , 2018, 102, 233-248.	2.6	64
58	Is RNASEL:p.Glu265* a modifier of early-onset breast cancer risk for carriers of high-risk mutations?. <i>BMC Cancer</i> , 2018, 18, 165.	1.1	6
59	The <i>BRCA1</i> c. 5096G>A p.Arg1699Gln (R1699Q) intermediate risk variant: breast and ovarian cancer risk estimation and recommendations for clinical management from the ENIGMA consortium. <i>Journal of Medical Genetics</i> , 2018, 55, 15-20.	1.5	50
60	Genetic Testing and Clinical Management Practices for Variants in Non-BRCA1/2 Breast (and) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 552 for the Interpretation of Germline Mutant Alleles (ENIGMA) Clinical Working Group. <i>JCO Precision Oncology</i> , 2018, 2, 1-42.	1.5	19
61	The Influence of Number and Timing of Pregnancies on Breast Cancer Risk for Women With BRCA1 or BRCA2 Mutations. <i>JNCI Cancer Spectrum</i> , 2018, 2, pky078.	1.4	21
62	Multigene Hereditary Cancer Panels Reveal High-Risk Pancreatic Cancer Susceptibility Genes. <i>JCO Precision Oncology</i> , 2018, 2, 1-28.	1.5	23
63	BRCA Challenge: BRCA Exchange as a global resource for variants in BRCA1 and BRCA2. <i>PLoS Genetics</i> , 2018, 14, e1007752.	1.5	148
64	Oral Contraceptive Use and Breast Cancer Risk: Retrospective and Prospective Analyses From a BRCA1 and BRCA2 Mutation Carrier Cohort Study. <i>JNCI Cancer Spectrum</i> , 2018, 2, pky023.	1.4	33
65	Rate of Recurrence of Lentigo Maligna Treated With Off-Label Neoadjuvant Topical Imiquimod, 5%, Cream Prior to Conservatively Staged Excision. <i>JAMA Dermatology</i> , 2018, 154, 885.	2.0	24
66	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.	3.0	225
67	A Transcriptome-Wide Association Study Among 97,898 Women to Identify Candidate Susceptibility Genes for Epithelial Ovarian Cancer Risk. <i>Cancer Research</i> , 2018, 78, 5419-5430.	0.4	54
68	Pancreatic cancer as a sentinel for hereditary cancer predisposition. <i>BMC Cancer</i> , 2018, 18, 697.	1.1	29
69	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.	9.4	184
70	Breast cancer risk prediction using a polygenic risk score in the familial setting: a prospective study from the Breast Cancer Family Registry and kConFab. <i>Genetics in Medicine</i> , 2017, 19, 30-35.	1.1	53
71	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.	1.1	116
72	<i>BRCA2</i> Hypomorphic Missense Variants Confer Moderate Risks of Breast Cancer. <i>Cancer Research</i> , 2017, 77, 2789-2799.	0.4	75

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73	Associations Between Cancer Predisposition Testing Panel Genes and Breast Cancer. <i>JAMA Oncology</i> , 2017, 3, 1190.	3.4	472
74	Risks of Breast, Ovarian, and Contralateral Breast Cancer for <i>BRCA1</i> and <i>BRCA2</i> Mutation Carriers. <i>JAMA - Journal of the American Medical Association</i> , 2017, 317, 2402.	3.8	1,898
75	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. <i>Nature Genetics</i> , 2017, 49, 680-691.	9.4	356
76	Association analysis identifies 65 new breast cancer risk loci. <i>Nature</i> , 2017, 551, 92-94.	13.7	1,099
77	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. <i>Nature Genetics</i> , 2017, 49, 1767-1778.	9.4	289
78	Panel sequencing of 264 candidate susceptibility genes and segregation analysis in a cohort of non- <i>BRCA1</i> , non- <i>BRCA2</i> breast cancer families. <i>Breast Cancer Research and Treatment</i> , 2017, 166, 937-949.	1.1	16
79	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.	0.6	105
80	Association of breast cancer risk in <i>BRCA1</i> and <i>BRCA2</i> mutation carriers with genetic variants showing differential allelic expression: identification of a modifier of breast cancer risk at locus 11q22.3. <i>Breast Cancer Research and Treatment</i> , 2017, 161, 117-134.	1.1	18
81	The OncoArray Consortium: A Network for Understanding the Genetic Architecture of Common Cancers. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2017, 26, 126-135.	1.1	278
82	Prediction of Breast and Prostate Cancer Risks in Male <i>BRCA1</i> and <i>BRCA2</i> Mutation Carriers Using Polygenic Risk Scores. <i>Journal of Clinical Oncology</i> , 2017, 35, 2240-2250.	0.8	152
83	Fine-Scale Mapping at 9p22.2 Identifies Candidate Causal Variants That Modify Ovarian Cancer Risk in <i>BRCA1</i> and <i>BRCA2</i> Mutation Carriers. <i>PLoS ONE</i> , 2016, 11, e0158801.	1.1	10
84	Adding In Silico Assessment of Potential Splice Aberration to the Integrated Evaluation of <i>BRCA</i> Gene Unclassified Variants. <i>Human Mutation</i> , 2016, 37, 627-639.	1.1	52
85	Functional assays provide a robust tool for the clinical annotation of genetic variants of uncertain significance. <i>Npj Genomic Medicine</i> , 2016, 1, .	1.7	70
86	Identification of independent association signals and putative functional variants for breast cancer risk through fine-scale mapping of the 12p11 locus. <i>Breast Cancer Research</i> , 2016, 18, 64.	2.2	31
87	Point Mutations in Exon 1B of APC Reveal Gastric Adenocarcinoma and Proximal Polyposis of the Stomach as a Familial Adenomatous Polyposis Variant. <i>American Journal of Human Genetics</i> , 2016, 98, 830-842.	2.6	201
88	Male breast cancer in <i>BRCA1</i> and <i>BRCA2</i> mutation carriers: pathology data from the Consortium of Investigators of Modifiers of <i>BRCA1/2</i> . <i>Breast Cancer Research</i> , 2016, 18, 15.	2.2	88
89	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. <i>Nature Communications</i> , 2016, 7, 11375.	5.8	93
90	Functional mechanisms underlying pleiotropic risk alleles at the 19p13.1 breast-ovarian cancer susceptibility locus. <i>Nature Communications</i> , 2016, 7, 12675.	5.8	78

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91	Cohort Profile: The Breast Cancer Prospective Family Study Cohort (ProF-SC). <i>International Journal of Epidemiology</i> , 2016, 45, 683-692.	0.9	48
92	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.	1.4	106
93	No evidence that protein truncating variants in <i>BRIP1</i> are associated with breast cancer risk: implications for gene panel testing. <i>Journal of Medical Genetics</i> , 2016, 53, 298-309.	1.5	94
94	Breast cancer risk variants at 6q25 display different phenotype associations and regulate ESR1, RMND1 and CCDC170. <i>Nature Genetics</i> , 2016, 48, 374-386.	9.4	125
95	Targeted massively parallel sequencing of a panel of putative breast cancer susceptibility genes in a large cohort of multiple-case breast and ovarian cancer families. <i>Journal of Medical Genetics</i> , 2016, 53, 34-42.	1.5	63
96	Functional Assessment of Genetic Variants with Outcomes Adapted to Clinical Decision-Making. <i>PLoS Genetics</i> , 2016, 12, e1006096.	1.5	24
97	An original phylogenetic approach identified mitochondrial haplogroup T1a1 as inversely associated with breast cancer risk in BRCA2 mutation carriers. <i>Breast Cancer Research</i> , 2015, 17, 61.	2.2	26
98	Assessing Associations between the AURKA-HMMR-TPX2-TUBG1 Functional Module and Breast Cancer Risk in BRCA1/2 Mutation Carriers. <i>PLoS ONE</i> , 2015, 10, e0120020.	1.1	34
99	Gene-Panel Sequencing and the Prediction of Breast-Cancer Risk. <i>New England Journal of Medicine</i> , 2015, 372, 2243-2257.	13.9	764
100	DNA methylation profiling to assess pathogenicity of BRCA1 unclassified variants in breast cancer. <i>Epigenetics</i> , 2015, 10, 1121-1132.	1.3	12
101	Identification of six new susceptibility loci for invasive epithelial ovarian cancer. <i>Nature Genetics</i> , 2015, 47, 164-171.	9.4	221
102	Association of Type and Location of <i>BRCA1</i> and <i>BRCA2</i> Mutations With Risk of Breast and Ovarian Cancer. <i>JAMA - Journal of the American Medical Association</i> , 2015, 313, 1347.	3.8	390
103	Candidate Genetic Modifiers for Breast and Ovarian Cancer Risk in <i>BRCA1</i> and <i>BRCA2</i> Mutation Carriers. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2015, 24, 308-316.	1.1	22
104	Refined histopathological predictors of BRCA1 and BRCA2 mutation status: a large-scale analysis of breast cancer characteristics from the BCAC, CIMBA, and ENIGMA consortia. <i>Breast Cancer Research</i> , 2014, 16, 3419.	2.2	97
105	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.	2.2	57
106	Application of a 5-tiered scheme for standardized classification of 2,360 unique mismatch repair gene variants in the InSiGHT locus-specific database. <i>Nature Genetics</i> , 2014, 46, 107-115.	9.4	410
107	Rare Mutations in <i>RINT1</i> Predispose Carriers to Breast and Lynch Syndrome—“Spectrum Cancers. <i>Cancer Discovery</i> , 2014, 4, 804-815.	7.7	44
108	Breast-Cancer Risk in Families with Mutations in <i>PALB2</i> . <i>New England Journal of Medicine</i> , 2014, 371, 497-506.	13.9	745

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109	Rare key functional domain missense substitutions in MRE11A, RAD50, and NBN contribute to breast cancer susceptibility: results from a Breast Cancer Family Registry case-control mutation-screening study. <i>Breast Cancer Research</i> , 2014, 16, R58.	2.2	99
110	Multifactorial Likelihood Assessment of BRCA1 and BRCA2 Missense Variants Confirms That BRCA1:c.122A>G(p.His41Arg) Is a Pathogenic Mutation. <i>PLoS ONE</i> , 2014, 9, e86836.	1.1	17
111	Prevalence of PALB2 mutations in Australasian multiple-case breast cancer families. <i>Breast Cancer Research</i> , 2013, 15, R17.	2.2	42
112	FAVR (Filtering and Annotation of Variants that are Rare): methods to facilitate the analysis of rare germline genetic variants from massively parallel sequencing datasets. <i>BMC Bioinformatics</i> , 2013, 14, 65.	1.2	8
113	A Classification Model for BRCA2 DNA Binding Domain Missense Variants Based on Homology-Directed Repair Activity. <i>Cancer Research</i> , 2013, 73, 265-275.	0.4	103
114	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, 371-384.	9.4	493
115	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. <i>Human Mutation</i> , 2013, 34, 200-209.	1.1	81
116	Calibration of Multiple In Silico Tools for Predicting Pathogenicity of Mismatch Repair Gene Missense Substitutions. <i>Human Mutation</i> , 2013, 34, 255-265.	1.1	80
117	Report of a novel OCA2 gene mutation and an investigation of OCA2 variants on melanoma risk in a familial melanoma pedigree. <i>Journal of Dermatological Science</i> , 2013, 69, 30-37.	1.0	32
118	Identification of a BRCA2-Specific Modifier Locus at 6p24 Related to Breast Cancer Risk. <i>PLoS Genetics</i> , 2013, 9, e1003173.	1.5	105
119	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.	1.5	244
120	Risk of Pancreatic Cancer in Breast Cancer Families from the Breast Cancer Family Registry. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2013, 22, 803-811.	1.1	83
121	BRCA1/2 Sequence Variants of Uncertain Significance: A Primer for Providers to Assist in Discussions and in Medical Management. <i>Oncologist</i> , 2013, 18, 518-524.	1.9	76
122	BRCA1 R1699Q variant displaying ambiguous functional abrogation confers intermediate breast and ovarian cancer risk. <i>Journal of Medical Genetics</i> , 2012, 49, 525-532.	1.5	97
123	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, 134-147.	1.1	513
124	Characterization of BRCA1 and BRCA2 splicing variants: a collaborative report by ENIGMA consortium members. <i>Breast Cancer Research and Treatment</i> , 2012, 132, 1009-1023.	1.1	56
125	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.	1.1	190
126	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.	1.1	269

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127	Classification of missense substitutions in the BRCA genes: A database dedicated to Ex-UVs. Human Mutation, 2012, 33, 22-28.	1.1	65
128	Rare variants in the ATM gene and risk of breast cancer. Breast Cancer Research, 2011, 13, R73.	2.2	188
129	Design Considerations for Massively Parallel Sequencing Studies of Complex Human Disease. PLoS ONE, 2011, 6, e23221.	1.1	20
130	FAN1 variants identified in multiple-case early-onset breast cancer families via exome sequencing: no evidence for association with risk for breast cancer. Breast Cancer Research and Treatment, 2011, 130, 1043-1049.	1.1	16
131	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
132	A Computational Method to Classify Variants of Uncertain Significance Using Functional Assay Data with Application to BRCA1. Cancer Epidemiology Biomarkers and Prevention, 2011, 20, 1078-1088.	1.1	54
133	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
134	71: The Breast Cancer Genes IARC Database: a tool to improve evaluation of BRCA1 and BRCA2 uncertain sequence variants. Bulletin Du Cancer, 2010, 97, S61.	0.6	0
135	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, 9742-9754.	0.4	169
136	A PALB2 mutation associated with high risk of breast cancer. Breast Cancer Research, 2010, 12, R109.	2.2	102
137	Multiple Loci within the Major Histocompatibility Complex Confer Risk of Psoriasis. PLoS Genetics, 2009, 5, e1000606.	1.5	141
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