David E Goldgar

List of Publications by Year
in descending order

166 papers	28,216 citations	10389 72 h-index	5829 161 g-index
174 all docs	174 docs citations	174 times ranked	22126 citing authors

#	Article	IF	Citations
1	A Strong Candidate for the Breast and Ovarian Cancer Susceptibility Gene BRCA1. Science, 1994, 266, 66-71.	12.6	5,747
2	Risks of Breast, Ovarian, and Contralateral Breast Cancer for BRCA1 and BRCA2 Mutation Carriers. JAMA - Journal of the American Medical Association, 2017, 317, 2402.	7.4	1,898
3	Localization of a Breast Cancer Susceptibility Gene, BRCA2 , to Chromosome 13q12-13. Science, 1994, 265, 2088-2090.	12.6	1,725
4	Association analysis identifies 65 new breast cancer risk loci. Nature, 2017, 551, 92-94.	27.8	1,099
5	Systematic Population-Based Assessment of Cancer Risk in First-Degree Relatives of Cancer Probands. Journal of the National Cancer Institute, 1994, 86, 1600-1608.	6.3	923
6	Gene-Panel Sequencing and the Prediction of Breast-Cancer Risk. New England Journal of Medicine, 2015, 372, 2243-2257.	27.0	764
7	Breast-Cancer Risk in Families with Mutations in PALB2. New England Journal of Medicine, 2014, 371, 497-506.	27.0	745
8	Polygenic Risk Scores for Prediction of Breast Cancer and Breast Cancer Subtypes. American Journal of Human Genetics, 2019, 104, 21-34.	6.2	711
9	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, 134-147.	2.5	513
10	Multiple independent variants at the TERT locus are associated with telomere length and risks of breast and ovarian cancer. Nature Genetics, 2013, 45, 371-384.	21.4	493
11	Associations Between Cancer Predisposition Testing Panel Genes and Breast Cancer. JAMA Oncology, 2017, 3, 1190.	7.1	472
12	A Systematic Genetic Assessment of 1,433 Sequence Variants of Unknown Clinical Significance in the BRCA1 and BRCA2 Breast Cancerâ€“Predisposition Genes. American Journal of Human Genetics, 2007, 81, 873-883.	6.2	416
13	A Population-Based Study of Genes Previously Implicated in Breast Cancer. New England Journal of Medicine, 2021, 384, 440-451.	27.0	414
14	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.	21.4	410
15	Association of Type and Location ofBRCA1andBRCA2Mutations With Risk of Breast and Ovarian Cancer. JAMA - Journal of the American Medical Association, 2015, 313, 1347.	7.4	390
16	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. Nature Genetics, 2017, 49, 680-691.	21.4	356
17	Integrated Evaluation of DNA Sequence Variants of Unknown Clinical Significance: Application to BRCA1 and BRCA2. American Journal of Human Genetics, 2004, 75, 535-544.	6.2	351
18	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. Nature Genetics, 2017, 49, 1767-1778.	21.4	289

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19	The OncoArray Consortium: A Network for Understanding the Genetic Architecture of Common Cancers. Cancer Epidemiology Biomarkers and Prevention, 2017, 26, 126-135.	2.5	278
20	Cancer Risks Associated With Germline PALB2 Pathogenic Variants: An International Study of 524 Families. Journal of Clinical Oncology, 2020, 38, 674-685.	1.6	270
21	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.	2.5	269
22	Genome-wide association study identifies 32 novel breast cancer susceptibility loci from overall and subtype-specific analyses. Nature Genetics, 2020, 52, 572-581.	21.4	265
23	Genome-Wide Association Study in BRCA1 Mutation Carriers Identifies Novel Loci Associated with Breast and Ovarian Cancer Risk. PLoS Genetics, 2013, 9, e1003212.	3.5	244
24	Triple-Negative Breast Cancer Risk Genes Identified by Multigene Hereditary Cancer Panel Testing. Journal of the National Cancer Institute, 2018, 110, 855-862.	6.3	225
25	Mutational spectrum in a worldwide study of 29,700 families withBRCA1orBRCA2mutations. Human Mutation, 2018, 39, 593-620.	2.5	224
26	Identification of six new susceptibility loci for invasive epithelial ovarian cancer. Nature Genetics, 2015, 47, 164-171.	21.4	221
27	Classification of rare missense substitutions, using risk surfaces, with genetic- and molecular-epidemiology applications. Human Mutation, 2008, 29, 1342-1354.	2.5	209
28	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.	6.2	201
29	A review of a multifactorial probability-based model for classification of BRCA1 and BRCA2 variants of uncertain significance (VUS). Human Mutation, 2012, 33, 8-21.	2.5	190
30	Rare variants in the ATMgene and risk of breast cancer. Breast Cancer Research, 2011, 13, R73.	5.0	188
31	A transcriptome-wide association study of 229,000 women identifies new candidate susceptibility genes for breast cancer. Nature Genetics, 2018, 50, 968-978.	21.4	184
32	Ovarian cancer risk in BRCA1 carriers is modified by the HRAS1 variable number of tandem repeat (VNTR) locus. Nature Genetics, 1996, 12, 309-311.	21.4	183
33	Genetic evidence and integration of various data sources for classifying uncertain variants into a single model. Human Mutation, 2008, 29, 1265-1272.	2.5	169
34	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.	0.9	169
35	Genetic and Histopathologic Evaluation ofBRCA1andBRCA2DNA Sequence Variants of Unknown Clinical Significance. Cancer Research, 2006, 66, 2019-2027.	0.9	153
36	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.	1.6	152

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37	BRCA Challenge: BRCA Exchange as a global resource for variants in BRCA1 and BRCA2. PLoS Genetics, 2018, 14, e1007752.	3.5	148
38	Familial male breast cancer is not linked to the BRCA1 locus on chromosome 17q. Nature Genetics, 1994, 7, 103-107.	21.4	146
39	Multiple Loci within the Major Histocompatibility Complex Confer Risk of Psoriasis. PLoS Genetics, 2009, 5, e1000606.	3.5	141
40	A weighted cohort approach for analysing factors modifying disease risks in carriers of highâ€risk susceptibility genes. Genetic Epidemiology, 2005, 29, 1-11.	1.3	136
41	An international initiative to identify genetic modifiers of cancer risk in BRCA1 and BRCA2 mutation carriers: the Consortium of Investigators of Modifiers of BRCA1 and BRCA2 (CIMBA). Breast Cancer Research, 2007, 9, 104.	5.0	136
42	A clinical guide to hereditary cancer panel testing: evaluation of gene-specific cancer associations and sensitivity of genetic testing criteria in a cohort of 165,000 high-risk patients. Genetics in Medicine, 2020, 22, 407-415.	2.4	136
43	A Full-Likelihood Method for the Evaluation of Causality of Sequence Variants from Family Data. American Journal of Human Genetics, 2003, 73, 652-655.	6.2	130
44	Breast cancer risk variants at 6q25 display different phenotype associations and regulate ESR1, RMND1 and CCDC170. Nature Genetics, 2016, 48, 374-386.	21.4	125
45	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. Nature Genetics, 2020, 52, 56-73.	21.4	120
46	Male breast cancer in a multi-gene panel testing cohort: insights and unexpected results. Breast Cancer Research and Treatment, 2017, 161, 575-586.	2.5	116
47	10-year performance of four models of breast cancer risk: a validation study. Lancet Oncology, The, 2019, 20, 504-517.	10.7	116
48	A genome wide linkage search for breast cancer susceptibility genes. Genes Chromosomes and Cancer, 2006, 45, 646-655.	2.8	111
49	Determination of Cancer Risk Associated with Germ Line BRCA1 Missense Variants by Functional Analysis. Cancer Research, 2007, 67, 1494-1501.	0.9	110
50	Functional Assays for Classification ofBRCA2Variants of Uncertain Significance. Cancer Research, 2008, 68, 3523-3531.	0.9	108
51	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.	2.9	106
52	Identification of a BRCA2-Specific Modifier Locus at 6p24 Related to Breast Cancer Risk. PLoS Genetics, 2013, 9, e1003173.	3.5	105
53	Frequency of mutations in a large series of clinically ascertained ovarian cancer cases tested on multi-gene panels compared to reference controls. Gynecologic Oncology, 2017, 147, 375-380.	1.4	105
54	A Classification Model for BRCA2 DNA Binding Domain Missense Variants Based on Homology-Directed Repair Activity. Cancer Research, 2013, 73, 265-275.	0.9	103

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55	A PALB2 mutation associated with high risk of breast cancer. Breast Cancer Research, 2010, 12, R109.	5.0	102
56	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.	2.5	102
57	Rare key functional domain missense substitutions in MRE11A, RAD50, and NBNcontribute to breast cancer susceptibility: results from a Breast Cancer Family Registry case-control mutation-screening study. Breast Cancer Research, 2014, 16, R58.	5.0	99
58	BRCA1 R1699Q variant displaying ambiguous functional abrogation confers intermediate breast and ovarian cancer risk. Journal of Medical Genetics, 2012, 49, 525-532.	3.2	97
59	Refined histopathological predictors of BRCA1 and BRCA2mutation status: a large-scale analysis of breast cancer characteristics from the BCAC, CIMBA, and ENIGMA consortia. Breast Cancer Research, 2014, 16, 3419.	5.0	97
60	Functional evaluation and cancer risk assessment of BRCA2 unclassified variants. Cancer Research, 2005, 65, 417-26.	0.9	97
61	No evidence that protein truncating variants inBRIP1are associated with breast cancer risk: implications for gene panel testing. Journal of Medical Genetics, 2016, 53, 298-309.	3.2	94
62	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. Nature Communications, 2016, 7, 11375.	12.8	93
63	Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. Nature Communications, 2019, 10, 1741.	12.8	90
64	Cancer Risks Associated With BRCA1 and BRCA2 Pathogenic Variants. Journal of Clinical Oncology, 2022, 40, 1529-1541.	1.6	90
65	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.	5.0	88
66	Shared heritability and functional enrichment across six solid cancers. Nature Communications, 2019, 10, 431.	12.8	88
67	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.	2.5	86
68	Risk of Pancreatic Cancer in Breast Cancer Families from the Breast Cancer Family Registry. Cancer Epidemiology Biomarkers and Prevention, 2013, 22, 803-811.	2.5	83
69	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.	2.4	82
70	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, 200-209.	2.5	81
71	Calibration of Multiple In Silico Tools for Predicting Pathogenicity of Mismatch Repair Gene Missense Substitutions. Human Mutation, 2013, 34, 255-265.	2.5	80
72	Assessing pathogenicity: overview of results from the IARC Unclassified Genetic Variants Working Group. Human Mutation, 2008, 29, 1261-1264.	2.5	79

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73	Functional mechanisms underlying pleiotropic risk alleles at the 19p13.1 breastâ€“ovarian cancer susceptibility locus. Nature Communications, 2016, 7, 12675.	12.8	78
74	BRCA1/2 Sequence Variants of Uncertain Significance: A Primer for Providers to Assist in Discussions and in Medical Management. Oncologist, 2013, 18, 518-524.	3.7	76
75	BRCA2 Hypomorphic Missense Variants Confer Moderate Risks of Breast Cancer. Cancer Research, 2017, 77, 2789-2799.	0.9	75
76	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.	2.5	74
77	Clinical Classification ofBRCA1andBRCA2DNA Sequence Variants: The Value of Cytokeratin Profiles and Evolutionary Analysisâ€"A Report From the kConFab Investigators. Journal of Clinical Oncology, 2008, 26, 1657-1663.	1.6	72
78	Functional assays provide a robust tool for the clinical annotation of genetic variants of uncertain significance. Npj Genomic Medicine, 2016, 1, .	3.8	70
79	Classification of missense substitutions in the BRCA genes: A database dedicated to Ex-UVs. Human Mutation, 2012, 33, 22-28.	2.5	65
80	Assessment of the Clinical Relevance of BRCA2 Missense Variants by Functional and Computational Approaches. American Journal of Human Genetics, 2018, 102, 233-248.	6.2	64
81	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. Journal of Medical Genetics, 2016, 53, 34-42.	3.2	63
82	The Contribution of Germline Predisposition Gene Mutations to Clinical Subtypes of Invasive Breast Cancer From a Clinical Genetic Testing Cohort. Journal of the National Cancer Institute, 2020, 112, 1231-1241.	6.3	61
83	ClassifyingMLH1andMSH2variants using bioinformatic prediction, splicing assays, segregation, and tumor characteristics. Human Mutation, 2009, 30, 757-770.	2.5	60
84	Identification of BRCA1 missense substitutions that confer partial functional activity: potential moderate risk variants?. Breast Cancer Research, 2007, 9, R82.	5.0	58
85	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.	5.0	57
86	Characterization of BRCA1 and BRCA2 splicing variants: a collaborative report by ENIGMA consortium members. Breast Cancer Research and Treatment, 2012, 132, 1009-1023.	2.5	56
87	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.	2.5	54
88	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.	0.9	54
89	Breast cancer risk prediction using a polygenic risk score in the familial setting: a prospective study from the Breast Cancer Family Registry and kConFab. Genetics in Medicine, 2017, 19, 30-35.	2.4	53
90	Adding In Silico Assessment of Potential Splice Aberration to the Integrated Evaluation of BRCA Gene Unclassified Variants. Human Mutation, 2016, 37, 627-639.	2.5	52

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91	Comprehensive annotation of BRCA1 and BRCA2 missense variants by functionally validated sequence-based computational prediction models. Genetics in Medicine, 2019, 21, 71-80.	2.4	52
92	Genome-wide association study of germline variants and breast cancer-specific mortality. British Journal of Cancer, 2019, 120, 647-657.	6.4	52
93	Contribution of Germline Predisposition Gene Mutations to Breast Cancer Risk in African American Women. Journal of the National Cancer Institute, 2020, 112, 1213-1221.	6.3	51
94	The BRCA1 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.	3.2	50
95	Cohort Profile: The Breast Cancer Prospective Family Study Cohort (ProF-SC). International Journal of Epidemiology, 2016, 45, 683-692.	1.9	48
96	Characterization of the Cancer Spectrum in Men With GermlineBRCA1andBRCA2Pathogenic Variants. JAMA Oncology, 2020, 6, 1218.	7.1	48
97	Risk of Breast Cancer Among Carriers of Pathogenic Variants in Breast Cancer Predisposition Genes Varies by Polygenic Risk Score. Journal of Clinical Oncology, 2021, 39, 2564-2573.	1.6	47
98	Rare Mutations in RINT1 Predispose Carriers to Breast and Lynch Syndromeâ€“Spectrum Cancers. Cancer Discovery, 2014, 4, 804-815.	9.4	44
99	Pathogenicity of the BRCA1 missense variant M1775K is determined by the disruption of the BRCT phosphopeptide-binding pocket: a multi-modal approach. European Journal of Human Genetics, 2008, 16, 820-832.	2.8	42
100	Prevalence of PALB2 mutations in Australasian multiple-case breast cancer families. Breast Cancer Research, 2013, 15, R17.	5.0	42
101	Risk-reducing salpingo-oophorectomy, natural menopause, and breast cancer risk: an international prospective cohort of BRCA1 and BRCA2 mutation carriers. Breast Cancer Research, 2020, 22, 8.	5.0	41
102	Association of Genomic Domains in BRCA1 and BRCA2 with Prostate Cancer Risk and Aggressiveness. Cancer Research, 2020, 80, 624-638.	0.9	39
103	Breast Cancer Polygenic Risk Score and Contralateral Breast Cancer Risk. American Journal of Human Genetics, 2020, 107, 837-848.	6.2	39
104	G1738R is a BRCA1 founder mutation in Greek breast/ovarian cancer patients: evaluation of its pathogenicity and inferences on its genealogical history. Breast Cancer Research and Treatment, 2008, 110, 377-385.	2.5	37
105	A functional assayâ€“based procedure to classify mismatch repair gene variants in Lynch syndrome. Genetics in Medicine, 2019, 21, 1486-1496.	2.4	36
106	Assessing Associations between the AURKA-HMMR-TPX2-TUBG1 Functional Module and Breast Cancer Risk in BRCA1/2 Mutation Carriers. PLoS ONE, 2015, 10, e0120020.	2.5	34
107	Oral contraceptive use and ovarian cancer risk for BRCA1/2 mutation carriers: an international cohort study. American Journal of Obstetrics and Gynecology, 2021, 225, 51.e1-51.e17.	1.3	34
108	Oral Contraceptive Use and Breast Cancer Risk: Retrospective and Prospective Analyses From a BRCA1 and BRCA2 Mutation Carrier Cohort Study. JNCI Cancer Spectrum, 2018, 2, pky023.	2.9	33

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109	Report of a novel OCA2 gene mutation and an investigation of OCA2 variants on melanoma risk in a familial melanoma pedigree. Journal of Dermatological Science, 2013, 69, 30-37.	1.9	32
110	Transcriptomeâ€wide association study of breast cancer risk by estrogenâ€receptor status. Genetic Epidemiology, 2020, 44, 442-468.	1.3	32
111	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.	5.0	31
112	Strong functional data for pathogenicity or neutrality classify BRCA2 DNA-binding-domain variants of uncertain significance. American Journal of Human Genetics, 2021, 108, 458-468.	6.2	31
113	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.	6.3	30
114	A network analysis to identify mediators of germline-driven differences in breast cancer prognosis. Nature Communications, 2020, 11, 312.	12.8	30
115	Pancreatic cancer as a sentinel for hereditary cancer predisposition. BMC Cancer, 2018, 18, 697.	2.6	29
116	Likelihood ratios to assess genetic evidence for clinical significance of uncertain variants: Hereditary hemorrhagic telangiectasia as a model. Experimental and Molecular Pathology, 2008, 85, 45-49.	2.1	28
117	The FANCM:p.Arg658* truncating variant is associated with risk of triple-negative breast cancer. Npj Breast Cancer, 2019, 5, 38.	5.2	28
118	Classification of variants of uncertain significance in BRCA1 and BRCA2 using personal and family history of cancer from individuals in a large hereditary cancer multigene panel testing cohort. Genetics in Medicine, 2020, 22, 701-708.	2.4	28
119	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.	5.0	26
120	Rate of Recurrence of Lentigo Maligna Treated With Off-Label Neoadjuvant Topical Imiquimod, 5%, Cream Prior to Conservatively Staged Excision. JAMA Dermatology, 2018, 154, 885.	4.1	24
121	Alcohol Consumption, Cigarette Smoking, and Risk of Breast Cancer for BRCA1 and BRCA2 Mutation Carriers: Results from The BRCA1 and BRCA2 Cohort Consortium. Cancer Epidemiology Biomarkers and Prevention, 2020, 29, 368-378.	2.5	24
122	Functional Assessment of Genetic Variants with Outcomes Adapted to Clinical Decision-Making. PLoS Genetics, 2016, 12, e1006096.	3.5	24
123	Multigene Hereditary Cancer Panels Reveal High-Risk Pancreatic Cancer Susceptibility Genes. JCO Precision Oncology, 2018, 2, 1-28.	3.0	23
124	Candidate Genetic Modifiers for Breast and Ovarian Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. Cancer Epidemiology Biomarkers and Prevention, 2015, 24, 308-316.	2.5	22
125	Germline Pathogenic Variants in Cancer Predisposition Genes Among Women With Invasive Lobular Carcinoma of the Breast. Journal of Clinical Oncology, 2021, 39, 3918-3926.	1.6	22
126	The Influence of Number and Timing of Pregnancies on Breast Cancer Risk for Women With BRCA1 or BRCA2 Mutations. JNCI Cancer Spectrum, 2018, 2, pky078.	2.9	21

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127	Radiation Treatment, ATM, BRCA1/2, and CHEK2*1100delC Pathogenic Variants and Risk of Contralateral Breast Cancer. Journal of the National Cancer Institute, 2020, 112, 1275-1279.	6.3	21
128	Integration of functional assay data results provides strong evidence for classification of hundreds of BRCA1 variants of uncertain significance. Genetics in Medicine, 2021, 23, 306-315.	2.4	21
129	Risk of Late-Onset Breast Cancer in Genetically Predisposed Women. Journal of Clinical Oncology, 2021, 39, 3430-3440.	1.6	21
130	Design Considerations for Massively Parallel Sequencing Studies of Complex Human Disease. PLoS ONE, 2011, 6, e23221.	2.5	20
131	The BRCA2 c.68-7TÂ>ÂA variant is not pathogenic: A model for clinical calibration of spliceogenicity. Human Mutation, 2018, 39, 729-741.	2.5	19
132	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. JCO Precision Oncology, 2018, 2, 1-42.	3.0	19
133	Assessment of blind predictions of the clinical significance of BRCA1 and BRCA2 variants. Human Mutation, 2019, 40, 1546-1556.	2.5	19
134	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.	6.4	19
135	A case-only study to identify genetic modifiers of breast cancer risk for BRCA1/BRCA2 mutation carriers. Nature Communications, 2021, 12, 1078.	12.8	19
136	Breast and Prostate Cancer Risks for MaleBRCA1andBRCA2Pathogenic Variant Carriers Using Polygenic Risk Scores. Journal of the National Cancer Institute, 2022, 114, 109-122.	6.3	19
137	Mutation prevalence tables for hereditary cancer derived from multigene panel testing. Human Mutation, 2020, 41, e1-e6.	2.5	19
138	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.	2.5	18
139	Racial and Ethnic Differences in Multigene Hereditary Cancer Panel Test Results for Women With Breast Cancer. Journal of the National Cancer Institute, 2021, 113, 1429-1433.	6.3	18
140	Multifactorial Likelihood Assessment of BRCA1 and BRCA2 Missense Variants Confirms That BRCA1:c.122A>G(p.His41Arg) Is a Pathogenic Mutation. PLoS ONE, 2014, 9, e86836.	2.5	17
141	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.	2.5	16
142	Panel sequencing of 264 candidate susceptibility genes and segregation analysis in a cohort of non-BRCA1, non-BRCA2 breast cancer families. Breast Cancer Research and Treatment, 2017, 166, 937-949.	2.5	16
143	The predictive ability of the 313 variantâ€“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.	2.4	16
144	Considerations in assessing germline variant pathogenicity using cosegregation analysis. Genetics in Medicine, 2020, 22, 2052-2059.	2.4	15

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145	Comparing 5-Year and Lifetime Risks of Breast CancerÂusing the Prospective Family Study Cohort. Journal of the National Cancer Institute, 2021, 113, 785-791.	6.3	13
146	DNA methylation profiling to assess pathogenicity of BRCA1 unclassified variants in breast cancer. Epigenetics, 2015, 10, 1121-1132.	2.7	12
147	Evaluation of a typology of reading disability. Neuropsychology, Development and Cognition Section A: Journal of Clinical and Experimental Neuropsychology, 1988, 10, 432-450.	1.1	11
148	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.	2.5	10
149	First international workshop of the ATM and cancer risk group (4-5 December 2019). Familial Cancer, 2022, 21, 211-227.	1.9	10
150	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, 2022, 24, 119-129.	2.4	10
151	Mendelian randomisation study of smoking exposure in relation to breast cancer risk. British Journal of Cancer, 2021, 125, 1135-1145.	6.4	9
152	FAVR (Filtering and Annotation of Variants that are Rare): methods to facilitate the analysis of rare germline genetic variants from massively parallel sequencing datasets. BMC Bioinformatics, 2013, 14, 65.	2.6	8
153	An updated quantitative model to classify missense variants in the TP53 gene: A novel multifactorial strategy. Human Mutation, 2021, 42, 1351-1361.	2.5	7
154	Oral Contraceptive Use in BRCA1 and BRCA2 Mutation Carriers: Absolute Cancer Risks and Benefits. Journal of the National Cancer Institute, 2022, 114, 540-552.	6.3	7
155	Is RNASEL:p.Glu265* a modifier of early-onset breast cancer risk for carriers of high-risk mutations?. BMC Cancer, 2018, 18, 165.	2.6	6
156	Comprehensive evaluation and efficient classification of BRCA1 RING domain missense substitutions. American Journal of Human Genetics, 2022, 109, 1153-1174.	6.2	6
157	Two truncating variants in FANCC and breast cancer risk. Scientific Reports, 2019, 9, 12524.	3.3	5
158	Differences in patient ascertainment affect the use of geneâ€specified ACMG/AMP phenotypeâ€related variant classification criteria: Evidence for TP53. Human Mutation, 2020, 41, 537-542.	2.5	5
159	An integrative model for the comprehensive classification of BRCA1 and BRCA2 variants of uncertain clinical significance. Npj Genomic Medicine, 2022, 7, .	3.8	4
160	Germline HOXB13 mutations p.G84E and p.R217C do not confer an increased breast cancer risk. Scientific Reports, 2020, 10, 9688.	3.3	2
161	Considerations When Using Breast Cancer Risk Models for Women with Negative BRCA1/BRCA2 Mutation Results. Journal of the National Cancer Institute, 2020, 112, 418-422.	6.3	1
162	Suggested actions from the Melbourne HVP Information Seminar. Nature Precedings, 0, , .	0.1	1

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163	Screening for linkage using a multipoint identity-by-descent method. Genetic Epidemiology, 1995, 12, 777-782.	1.3	0
164	Suggested actions from the Melbourne HVP Information Seminar. Nature Precedings, 2008, , .	0.1	0
165	Suggested actions from the Melbourne HVP Information Seminar. Nature Precedings, 2008, , .	0.1	0
166	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.	1.6	0