Marc Tischkowitz

List of Publications by Year in descending order

238 papers	23,379 citations	10986 71 h-index	9345 143 g-index
255 all docs	255 docs citations	255 times ranked	26986 citing authors

#	Article	IF	Citations
1	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
2	Prevalence and architecture of de novo mutations in developmental disorders. Nature, 2017, 542, 433-438.	27.8	1,211
3	Olaparib in patients with recurrent high-grade serous or poorly differentiated ovarian carcinoma or triple-negative breast cancer: a phase 2, multicentre, open-label, non-randomised study. Lancet Oncology, The, 2011, 12, 852-861.	10.7	1,028
4	Gene-Panel Sequencing and the Prediction of Breast-Cancer Risk. New England Journal of Medicine, 2015, 372, 2243-2257.	27.0	764
5	Cancer Risks for BRCA1 and BRCA2 Mutation Carriers: Results From Prospective Analysis of EMBRACE. Journal of the National Cancer Institute, 2013, 105, 812-822.	6.3	753
6	Breast-Cancer Risk in Families with Mutations in PALB2. New England Journal of Medicine, 2014, 371, 497-506.	27.0	745
7	Germline BRCA Mutations Are Associated With Higher Risk of Nodal Involvement, Distant Metastasis, and Poor Survival Outcomes in Prostate Cancer. Journal of Clinical Oncology, 2013, 31, 1748-1757.	1.6	641
8	Biallelic mutations in PALB2 cause Fanconi anemia subtype FA-N and predispose to childhood cancer. Nature Genetics, 2007, 39, 162-164.	21.4	556
9	Disruption of the Fanconi anemiaâ€“BRCA pathway in cisplatin-sensitive ovarian tumors. Nature Medicine, 2003, 9, 568-574.	30.7	508
10	Hereditary diffuse gastric cancer: updated clinical guidelines with an emphasis on germlineCDH1mutation carriers. Journal of Medical Genetics, 2015, 52, 361-374.	3.2	479
11	BOADICEA: a comprehensive breast cancer risk prediction model incorporating genetic and nongenetic risk factors. Genetics in Medicine, 2019, 21, 1708-1718.	2.4	415
12	Founder and Recurrent CDH1 Mutations in Families With Hereditary Diffuse Gastric Cancer. JAMA - Journal of the American Medical Association, 2007, 297, 2360.	7.4	394
13	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
14	Germline and somatic SMARCA4 mutations characterize small cell carcinoma of the ovary, hypercalcemic type. Nature Genetics, 2014, 46, 438-443.	21.4	383
15	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. Nature Genetics, 2017, 49, 680-691.	21.4	356
16	Comprehensive Rare Variant Analysis via Whole-Genome Sequencing to Determine the Molecular Pathology of Inherited Retinal Disease. American Journal of Human Genetics, 2017, 100, 75-90.	6.2	343
17	Use of immunohistochemical markers can refine prognosis in triple negative breast cancer. BMC Cancer, 2007, 7, 134.	2.6	316
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	<emph type="ital">DICER1</emph> Mutations in Familial Multinodular Goiter With and Without Ovarian Sertoli-Leydig Cell Tumors. JAMA - Journal of the American Medical Association, 2011, 305, 68.	7.4	284
20	Effect of BRCA Mutations on Metastatic Relapse and Cause-specific Survival After Radical Treatment for Localised Prostate Cancer. European Urology, 2015, 68, 186-193.	1.9	279
21	Hereditary breast cancer: new genetic developments, new therapeutic avenues. Human Genetics, 2008, 124, 31-42.	3.8	276
22	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
23	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
24	Germline CDKN1B/p27Kip1 Mutation in Multiple Endocrine Neoplasia. Journal of Clinical Endocrinology and Metabolism, 2007, 92, 3321-3325.	3.6	262
25	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
26	Fanconi anaemia. Journal of Medical Genetics, 2003, 40, 1-10.	3.2	243
27	Hereditary diffuse gastric cancer: updated clinical practice guidelines. Lancet Oncology, The, 2020, 21, e386-e397.	10.7	237
28	Mutational spectrum in a worldwide study of 29,700 families withBRCA1orBRCA2mutations. Human Mutation, 2018, 39, 593-620.	2.5	224
29	Identification of six new susceptibility loci for invasive epithelial ovarian cancer. Nature Genetics, 2015, 47, 164-171.	21.4	221
30	Histone Lysine Methylases and Demethylases in the Landscape of Human Developmental Disorders. American Journal of Human Genetics, 2018, 102, 175-187.	6.2	204
31	Targeted Prostate Cancer Screening in BRCA1 and BRCA2 Mutation Carriers: Results from the Initial Screening Round of the IMPACT Study. European Urology, 2014, 66, 489-499.	1.9	195
32	Phase II Study of Vinorelbine in Patients With Malignant Pleural Mesothelioma. Journal of Clinical Oncology, 2000, 18, 3912-3917.	1.6	192
33	Analysis of PALB2/FANCN-associated breast cancer families. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 6788-6793.	7.1	192
34	PALB2/FANCN: Recombining Cancer and Fanconi Anemia. Cancer Research, 2010, 70, 7353-7359.	0.9	187
35	Personalized early detection and prevention of breast cancer: ENVISION consensus statement. Nature Reviews Clinical Oncology, 2020, 17, 687-705.	27.6	178
36	PALB2,CHEK2andATMrare variants and cancer risk: data from COGS. Journal of Medical Genetics, 2016, 53, 800-811.	3.2	174

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37	Extending the phenotypes associated withDICER1mutations. Human Mutation, 2011, 32, 1381-1384.	2.5	173
38	The Manchester International Consensus Group recommendations for the management of gynecological cancers in Lynch syndrome. Genetics in Medicine, 2019, 21, 2390-2400.	2.4	153
39	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
40	Interim Results from the IMPACT Study: Evidence for Prostate-specific Antigen Screening in BRCA2 Mutation Carriers. European Urology, 2019, 76, 831-842.	1.9	148
41	Germline Mutation in BRCA1 or BRCA2 and Ten-Year Survival for Women Diagnosed with Epithelial Ovarian Cancer. Clinical Cancer Research, 2015, 21, 652-657.	7.0	138
42	Identification of a novel truncating PALB2mutation and analysis of its contribution to early-onset breast cancer in French-Canadian women. Breast Cancer Research, 2007, 9, R83.	5.0	126
43	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
44	Fanconi anaemia and leukaemia â€“ clinical and molecular aspects. British Journal of Haematology, 2004, 126, 176-191.	2.5	124
45	Biallelic Deleterious BRCA1 Mutations in a Woman with Early-Onset Ovarian Cancer. Cancer Discovery, 2013, 3, 399-405.	9.4	124
46	Prostate Cancer Risks for Male BRCA1 and BRCA2 Mutation Carriers: A Prospective Cohort Study. European Urology, 2020, 77, 24-35.	1.9	124
47	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. Nature Genetics, 2020, 52, 56-73.	21.4	120
48	Germline DICER1 mutations and familial cystic nephroma. Journal of Medical Genetics, 2010, 47, 863-866.	3.2	113
49	Clinical implications of germline mutations in breast cancer: TP53. Breast Cancer Research and Treatment, 2018, 167, 417-423.	2.5	112
50	Phenotypic Characterization of EIF2AK4 Mutation Carriers in a Large Cohort of Patients Diagnosed Clinically With Pulmonary Arterial Hypertension. Circulation, 2017, 136, 2022-2033.	1.6	111
51	Ovarian and Breast Cancer Risks Associated With Pathogenic Variants in RAD51C and RAD51D. Journal of the National Cancer Institute, 2020, 112, 1242-1250.	6.3	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	Heimler Syndrome Is Caused by Hypomorphic Mutations in the Peroxisome-Biogenesis Genes PEX1 and PEX6. American Journal of Human Genetics, 2015, 97, 535-545.	6.2	103
54	A PALB2 mutation associated with high risk of breast cancer. Breast Cancer Research, 2010, 12, R109.	5.0	102

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55	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
56	CanRisk Toolâ€"A Web Interface for the Prediction of Breast and Ovarian Cancer Risk and the Likelihood of Carrying Genetic Pathogenic Variants. Cancer Epidemiology Biomarkers and Prevention, 2021, 30, 469-473.	2.5	98
57	An assessment of the efficacy of cancer genetic counselling using real-time videoconferencing technology (telemedicine) compared to face-to-face consultations. European Journal of Cancer, 2005, 41, 2257-2261.	2.8	90
58	Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. Nature Communications, 2019, 10, 1741.	12.8	90
59	Cancer Risks Associated With BRCA1 and BRCA2 Pathogenic Variants. Journal of Clinical Oncology, 2022, 40, 1529-1541.	1.6	90
60	Randomized Trial of a Specialist Genetic Assessment Service for Familial Breast Cancer. Journal of the National Cancer Institute, 2000, 92, 1345-1351.	6.3	89
61	A common founder mutation in FANCA underlies the world's highest prevalence of Fanconi anemia in Gypsy families from Spain. Blood, 2005, 105, 1946-1949.	1.4	89
62	Consensus for genes to be included on cancer panel tests offered by UK genetics services: guidelines of the UK Cancer Genetics Group. Journal of Medical Genetics, 2018, 55, 372-377.	3.2	88
63	Shared heritability and functional enrichment across six solid cancers. Nature Communications, 2019, 10, 431.	12.8	88
64	Germline pathogenic variants in PALB2 and other cancer-predisposing genes in families with hereditary diffuse gastric cancer without CDH1 mutation: a whole-exome sequencing study. The Lancet Gastroenterology and Hepatology, 2018, 3, 489-498.	8.1	87
65	Flexible and scalable diagnostic filtering of genomic variants using G2P with Ensembl VEP. Nature Communications, 2019, 10, 2373.	12.8	86
66	Comparative study of endoscopic surveillance in hereditary diffuse gastric cancer according to CDH1 mutation status. Gastrointestinal Endoscopy, 2018, 87, 408-418.	1.0	85
67	Targeted prostate cancer screening in men with mutations in BRCA1 and BRCA2 detects aggressive prostate cancer: preliminary analysis of the results of the IMPACT study. BJU International, 2011, 107, 28-39.	2.5	83
68	HomozygousBUB1BMutation and Susceptibility to Gastrointestinal Neoplasia. New England Journal of Medicine, 2010, 363, 2628-2637.	27.0	82
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	Small-Cell Carcinoma of the Ovary, Hypercalcemic Typeâ€"Genetics, New Treatment Targets, and Current Management Guidelines. Clinical Cancer Research, 2020, 26, 3908-3917.	7.0	82
71	Incorporating truncating variants in PALB2, CHEK2, and ATM into the BOADICEA breast cancer risk model. Genetics in Medicine, 2016, 18, 1190-1198.	2.4	80
72	Deletion and reduced expression of the Fanconi anemia FANCA gene in sporadic acute myeloid leukemia. Leukemia, 2004, 18, 420-425.	7.2	78

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73	BRCA2 Polymorphic Stop Codon K3326X and the Risk of Breast, Prostate, and Ovarian Cancers. Journal of the National Cancer Institute, 2016, 108, djv315.	6.3	77
74	Prospective cohort study assessing outcomes of patients from families fulfilling criteria for hereditary diffuse gastric cancer undergoing endoscopic surveillance. Gastrointestinal Endoscopy, 2014, 80, 78-87.	1.0	75
75	Large Genomic Deletions inAIPin Pituitary Adenoma Predisposition. Journal of Clinical Endocrinology and Metabolism, 2008, 93, 4146-4151.	3.6	74
76	Rare germline mutations inPALB2and breast cancer risk: A population-based study. Human Mutation, 2012, 33, 674-680.	2.5	74
77	Compromised BRCA1â€“PALB2 interaction is associated with breast cancer risk. Oncogene, 2017, 36, 4161-4170.	5.9	71
78	Fanconi anaemia: genetics, molecular biology, and cancerÂâ€“Âimplications for clinical management in children and adults. Clinical Genetics, 2015, 88, 13-24.	2.0	69
79	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, 3304-3321.	2.9	68
80	Breast-Cancer Risk in Families with Mutations in PALB2. New England Journal of Medicine, 2014, 371, 1650-1652.	27.0	68
81	Biâ€allelic silencing of the Fanconi anaemia gene FANCF in acute myeloid leukaemia. British Journal of Haematology, 2003, 123, 469-471.	2.5	65
82	Cancer Surveillance Guideline for individuals with PTEN hamartoma tumour syndrome. European Journal of Human Genetics, 2020, 28, 1387-1393.	2.8	63
83	miRNA Processing and Human Cancer: DICER1 Cuts the Mustard. Science Translational Medicine, 2011, 3, 111ps46.	12.4	60
84	Genotype, extrapyramidal features, and severity of variant ataxiaâ€telangiectasia. Annals of Neurology, 2019, 85, 170-180.	5.3	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	New paradigms forBRCA1/BRCA2testing in women with ovarian cancer: results of the Genetic Testing in Epithelial Ovarian Cancer (GTEOC) study. Journal of Medical Genetics, 2016, 53, 655-661.	3.2	57
87	Acquired FANCA dysfunction and cytogenetic instability in adult acute myelogenous leukemia. Blood, 2003, 102, 7-16.	1.4	56
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	A common Fanconi anemia mutation in black populations of sub-Saharan Africa. Blood, 2005, 105, 3542-3544.	1.4	53
90	Pathology update to the Manchester Scoring System based on testing in over 4000 families. Journal of Medical Genetics, 2017, 54, 674-681.	3.2	51

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91	RAD51C germline mutations in breast and ovarian cancer patients. Breast Cancer Research, 2010, 12, 404.	5.0	50
92	Fanconi anaemia,BRCA2mutations and childhood cancer: a developmental perspective from clinical and epidemiological observations with implications for genetic counselling. Journal of Medical Genetics, 2014, 51, 71-75.	3.2	48
93	Characterization of the Cancer Spectrum in Men With GermlineBRCA1andBRCA2Pathogenic Variants. JAMA Oncology, 2020, 6, 1218.	7.1	48
94	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, 645-657.	2.5	47
95	DNA Glycosylases Involved in Base Excision Repair May Be Associated with Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. PLoS Genetics, 2014, 10, e1004256.	3.5	47
96	The Basal Phenotype of BRCA1-Related Breast Cancer: Past, Present and Future. Cell Cycle, 2006, 5, 963-967.	2.6	46
97	Dermatological manifestations of inherited cancer syndromes in children. British Journal of Dermatology, 2011, 164, 245-256.	1.5	46
98	Comprehensive Cancer-Predisposition Gene Testing in an Adult Multiple Primary Tumor Series Shows a Broad Range of Deleterious Variants and Atypical Tumor Phenotypes. American Journal of Human Genetics, 2018, 103, 3-18.	6.2	46
99	PTEN Hamartoma tumor syndrome in childhood: A review of the clinical literature. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 591-610.	1.6	46
100	Breast Cancer Family History and Contralateral Breast Cancer Risk in Young Women: An Update From the Womenâ€™s Environmental Cancer and Radiation Epidemiology Study. Journal of Clinical Oncology, 2018, 36, 1513-1520.	1.6	44
101	Mutation analysis of RAD51D in non-BRCA1/2 ovarian and breast cancer families. British Journal of Cancer, 2012, 106, 1460-1463.	6.4	43
102	Multilocus Inherited Neoplasia Alleles Syndrome. JAMA Oncology, 2016, 2, 373.	7.1	43
103	Germline whole exome sequencing and large-scale replication identifies FANCM as a likely high grade serous ovarian cancer susceptibility gene. Oncotarget, 2017, 8, 50930-50940.	1.8	43
104	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
105	Inheritance of deleterious mutations at both BRCA1 and BRCA2 in an international sample of 32,295 women. Breast Cancer Research, 2016, 18, 112.	5.0	42
106	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
107	Serum levels of mature microRNAs in DICER1-mutated pleuropulmonary blastoma. Oncogenesis, 2014, 3, e87-e87.	4.9	40
108	Evaluating clinician acceptability of the prototype CanRisk tool for predicting risk of breast and ovarian cancer: A multi-methods study. PLoS ONE, 2020, 15, e0229999.	2.5	40

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109	p.Val804Met, the Most Frequent Pathogenic Mutation in RET, Confers a Very Low Lifetime Risk of Medullary Thyroid Cancer. Journal of Clinical Endocrinology and Metabolism, 2018, 103, 4275-4282.	3.6	39
110	Homologous recombination DNA repair defects in PALB2-associated breast cancers. Npj Breast Cancer, 2019, 5, 23.	5.2	39
111	Association of Genomic Domains in BRCA1 and BRCA2 with Prostate Cancer Risk and Aggressiveness. Cancer Research, 2020, 80, 624-638.	0.9	39
112	Analysis of the gene coding for the BRCA2â€interacting protein PALB2 in hereditary prostate cancer. Prostate, 2008, 68, 675-678.	2.3	36
113	De Novo Truncating Mutations in WASF1 Cause Intellectual Disability with Seizures. American Journal of Human Genetics, 2018, 103, 144-153.	6.2	36
114	Phase II trial of liposomal daunorubicin in malignant pleural mesothelioma. Annals of Oncology, 2001, 12, 497-499.	1.2	35
115	Management of individuals with germline variants in PALB2: a clinical practice resource of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2021, 23, 1416-1423.	2.4	34
116	Comprehensive epithelial tubo-ovarian cancer risk prediction model incorporating genetic and epidemiological risk factors. Journal of Medical Genetics, 2022, 59, 632-643.	3.2	33
117	Inherited cancer in children: practical/ethical problems and challenges. European Journal of Cancer, 2004, 40, 2459-2470.	2.8	32
118	The clinical presentation caused by truncating CHD8 variants. Clinical Genetics, 2019, 96, 72-84.	2.0	32
119	Towards controlled terminology for reporting germline cancer susceptibility variants: an ENIGMA report. Journal of Medical Genetics, 2019, 56, 347-357.	3.2	32
120	Transcriptomeâ€wide association study of breast cancer risk by estrogenâ€receptor status. Genetic Epidemiology, 2020, 44, 442-468.	1.3	32
121	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
122	A homozygousPMS2founder mutation with an attenuated constitutional mismatch repair deficiency phenotype. Journal of Medical Genetics, 2015, 52, 348-352.	3.2	30
123	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
124	Cancer Variant Interpretation Group UK (CanVIG-UK): an exemplar national subspecialty multidisciplinary network. Journal of Medical Genetics, 2020, 57, 829-834.	3.2	30
125	Nijmegen breakage syndrome diagnosed as Fanconi anaemia. Pediatric Blood and Cancer, 2005, 44, 494-499.	1.5	29
126	Germline PALB2 mutation analysis in breast-pancreas cancer families. Journal of Medical Genetics, 2011, 48, 523-525.	3.2	28

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127	The FANCM:p.Arg658* truncating variant is associated with risk of triple-negative breast cancer. Npj Breast Cancer, 2019, 5, 38.	5.2	28
128	Combining evidence for and against pathogenicity for variants in cancer susceptibility genes: CanVIG-UK consensus recommendations. Journal of Medical Genetics, 2021, 58, 297-304.	3.2	28
129	Characterization of two Ashkenazi Jewish founder mutations in MSH6 gene causing Lynch syndrome. Clinical Genetics, 2011, 79, 512-522.	2.0	27
130	International distribution and age estimation of the Portuguese BRCA2 c.156_157insAlu founder mutation. Breast Cancer Research and Treatment, 2011, 127, 671-679.	2.5	27
131	Hormone receptor status of a first primary breast cancer predicts contralateral breast cancer risk in the WECARE study population. Breast Cancer Research, 2017, 19, 83.	5.0	27
132	Whole Exome Sequencing Identifies Candidate Genes Associated with Hereditary Predisposition to Uveal Melanoma. Ophthalmology, 2020, 127, 668-678.	5.2	27
133	A comparison of models used to predict MLH1, MSH2 and MSH6 mutation carriers. Annals of Oncology, 2009, 20, 681-688.	1.2	26
134	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
135	An international survey of surveillance schemes for unaffected BRCA1 and BRCA2 mutation carriers. Breast Cancer Research and Treatment, 2016, 157, 319-327.	2.5	26
136	Biallelic Mutation of ARHGEF18, Involved in the Determination of Epithelial Apicobasal Polarity, Causes Adult-Onset Retinal Degeneration. American Journal of Human Genetics, 2017, 100, 334-342.	6.2	26
137	BRCA1andBRCA2pathogenic sequence variants in women of African origin or ancestry. Human Mutation, 2019, 40, 1781-1796.	2.5	26
138	Low bone mass and high material bone density in two patients with Loeys-Dietz syndrome caused by transforming growth factor beta receptor 2 mutations. Journal of Bone and Mineral Research, 2012, 27, 713-718.	2.8	25
139	Contribution of the PALB2 c.2323C>T [p.Q775X] Founder mutation in well-defined breast and/or ovarian cancer families and unselected ovarian cancer cases of French Canadian descent. BMC Medical Genetics, 2013, 14, 5.	2.1	25
140	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
141	Cancer incidence in relatives of British Fanconi Anaemia patients. BMC Cancer, 2008, 8, 257.	2.6	23
142	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, 1362-1370.	2.5	23
143	Evaluation of RAD51C as cancer susceptibility gene in a large breast-ovarian cancer patient population referred for genetic testing. Breast Cancer Research and Treatment, 2012, 133, 393-398.	2.5	23
144	UKCGG Consensus Group guidelines for the management of patients with constitutional TP53 pathogenic variants. Journal of Medical Genetics, 2021, 58, 135-139.	3.2	23

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145	Polygenic risk modeling for prediction of epithelial ovarian cancer risk. European Journal of Human Genetics, 2022, 30, 349-362.	2.8	23
146	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
147	Body mass index, weight change, and risk of second primary breast cancer in the <scp>WECARE</scp> study: influence of estrogen receptor status of the first breast cancer. Cancer Medicine, 2016, 5, 3282-3291.	2.8	22
148	Overview of hereditary breast and ovarian cancer (HBOC) guidelines across Europe. European Journal of Medical Genetics, 2021, 64, 104350.	1.3	22
149	Quantitative PCR analysis reveals a high incidence of large intragenic deletions in the FANCA gene in Spanish Fanconi anemia patients. Cytogenetic and Genome Research, 2004, 104, 341-345.	1.1	21
150	Analysis of the Novel Fanconi Anemia GeneSLX4/FANCPin Familial Breast Cancer Cases. Human Mutation, 2013, 34, 70-73.	2.5	21
151	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
152	Identification and characterization of novel SNPs in CHEK2 in Ashkenazi Jewish men with prostate cancer. Cancer Letters, 2008, 270, 173-180.	7.2	19
153	Mutation analysis of PALB2 in BRCA1 and BRCA2-negative breast and/or ovarian cancer families from Eastern Ontario, Canada. Hereditary Cancer in Clinical Practice, 2014, 12, 19.	1.5	19
154	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
155	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
156	Clonal hematopoiesis and therapy-related myeloid neoplasms following neuroblastoma treatment. Blood, 2021, 137, 2992-2997.	1.4	19
157	Surveillance recommendations for DICER1 pathogenic variant carriers: a report from the SIOPE Host Genome Working Group and CanGene-CanVar Clinical Guideline Working Group. Familial Cancer, 2021, 20, 337-348.	1.9	19
158	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
159	No clinical utility of KRAS variant rs61764370 for ovarian or breast cancer. Gynecologic Oncology, 2016, 141, 386-401.	1.4	18
160	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
161	BRCA1 and BRCA2 pathogenic variants and prostate cancer risk: systematic review and meta-analysis. British Journal of Cancer, 2022, 126, 1067-1081.	6.4	18
162	A comparison of methods currently used in clinical practice to estimate familial breast cancer risks. Annals of Oncology, 2000, 11, 451-454.	1.2	16

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163	The BRCA2 c.9004G>A (E2003K) variant is likely pathogenic and recurs in breast and/or ovarian cancer families of French Canadian descent. Breast Cancer Research and Treatment, 2012, 131, 333-340.	2.5	16
164	Biallelic somaticSMARCA4mutations in small cell carcinoma of the ovary, hypercalcemic type (SCCOHT). Pediatric Blood and Cancer, 2015, 62, 728-730.	1.5	16
165	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
166	Risks of breast or ovarian cancer in BRCA1 or BRCA2 predictive test negatives: findings from the EMBRACE study. Genetics in Medicine, 2018, 20, 1575-1582.	2.4	15
167	Hereditary Diffuse Gastric Cancer: Approaches to Screening, Surveillance, and Treatment. Annual Review of Medicine, 2021, 72, 263-280.	12.2	15
168	Medulloblastoma as a First Presentation of Fanconi Anemia. Journal of Pediatric Hematology/Oncology, 2004, 26, 52-55.	0.6	14
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