Marc Tischkowitz

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/6166443/publications.pdf

Version: 2024-02-01

238 papers	23,379 citations	12597 71 h-index	10679 143 g-index
255 all docs	255 docs citations	255 times ranked	28887 citing authors

#	Article	IF	Citations
1	UK recommendations forSDHAgermline genetic testing and surveillance in clinical practice. Journal of Medical Genetics, 2023, 60, 107-111.	1.5	4
2	Breast and Prostate Cancer Risks for MaleBRCA1andBRCA2Pathogenic Variant Carriers Using Polygenic Risk Scores. Journal of the National Cancer Institute, 2022, 114, 109-122.	3.0	19
3	Quantifying prediction of pathogenicity for within-codon concordance (PM5) using 7541 functional classifications of BRCA1 and MSH2 missense variants. Genetics in Medicine, 2022, 24, 552-563.	1.1	5
4	Comprehensive epithelial tubo-ovarian cancer risk prediction model incorporating genetic and epidemiological risk factors. Journal of Medical Genetics, 2022, 59, 632-643.	1.5	33
5	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.	1.1	10
6	Quantifying evidence toward pathogenicity for rare phenotypes: The case of succinate dehydrogenase genes, SDHB and SDHD. Genetics in Medicine, 2022, 24, 41-50.	1.1	5
7	Cancer Risks Associated With BRCA1 and BRCA2 Pathogenic Variants. Journal of Clinical Oncology, 2022, 40, 1529-1541.	0.8	90
8	Polygenic risk modeling for prediction of epithelial ovarian cancer risk. European Journal of Human Genetics, 2022, 30, 349-362.	1.4	23
9	Multilocus Inherited Neoplasia Allele Syndrome (MINAS): an update. European Journal of Human Genetics, 2022, 30, 265-270.	1.4	12
10	BRCA1 and BRCA2 pathogenic variants and prostate cancer risk: systematic review and meta-analysis. British Journal of Cancer, 2022, 126, 1067-1081.	2.9	18
11	Targeted long-read sequencing identifies missing pathogenic variants in unsolved Werner syndrome cases. Journal of Medical Genetics, 2022, 59, 1087-1094.	1.5	14
12	Personalised Risk Prediction in Hereditary Breast and Ovarian Cancer: A Protocol for a Multi-Centre Randomised Controlled Trial. Cancers, 2022, 14, 2716.	1.7	10
13	UKCGG Consensus Group guidelines for the management of patients with constitutional TP53 pathogenic variants. Journal of Medical Genetics, 2021, 58, 135-139.	1.5	23
14	Combining evidence for and against pathogenicity for variants in cancer susceptibility genes: CanVIG-UK consensus recommendations. Journal of Medical Genetics, 2021, 58, 297-304.	1.5	28
15	Hereditary Diffuse Gastric Cancer: Approaches to Screening, Surveillance, and Treatment. Annual Review of Medicine, 2021, 72, 263-280.	5.0	15
16	A no-deal Brexit will be detrimental to people with rare diseases. Lancet, The, 2021, 397, 20.	6.3	4
17	Pathogenic germline variants in patients with features of hereditary renal cell carcinoma: Evidence for further locus heterogeneity. Genes Chromosomes and Cancer, 2021, 60, 5-16.	1.5	10
18	A case-only study to identify genetic modifiers of breast cancer risk for BRCA1/BRCA2 mutation carriers. Nature Communications, 2021, 12, 1078.	5.8	19

#	Article	IF	Citations
19	Update: variable implementation of the 2018 UKCGG/UKGTN guidelines for breast cancer gene panel tests offered by UK genetics services. Journal of Medical Genetics, 2021, 58, 579-580.	1.5	1
20	Quantitative evidence evaluation for singleton rare missense variants in rare distinctive adult-onset phenotypes: the exemplar of SDHB and SDHD. Molecular Genetics and Metabolism, 2021, 132, S37.	0.5	0
21	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.	1.1	34
22	Clonal hematopoiesis and therapy-related myeloid neoplasms following neuroblastoma treatment. Blood, 2021, 137, 2992-2997.	0.6	19
23	A mosaic PIK3CA variant in a young adult with diffuse gastric cancer: case report. European Journal of Human Genetics, 2021, 29, 1354-1358.	1.4	9
24	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.	1.1	16
25	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.	0.9	19
26	Investigating the clinical, pathological and molecular profile of oncocytic adrenocortical neoplasms: a case series and literature review. Endocrine Oncology, 2021, 1, 33-44.	0.1	0
27	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.	1.1	98
28	Overview of hereditary breast and ovarian cancer (HBOC) guidelines across Europe. European Journal of Medical Genetics, 2021, 64, 104350.	0.7	22
29	Assessing BRCA1 activity in DNA damage repair using human induced pluripotent stem cells as an approach to assist classification of BRCA1 variants of uncertain significance. PLoS ONE, 2021, 16, e0260852.	1.1	2
30	A functionally impaired missense variant identified in French Canadian families implicates FANCI as a candidate ovarian cancer-predisposing gene. Genome Medicine, 2021, 13, 186.	3.6	12
31	Association of Genomic Domains in BRCA1 and BRCA2 with Prostate Cancer Risk and Aggressiveness. Cancer Research, 2020, 80, 624-638.	0.4	39
32	A systematic review of predicted pathogenic PALB2 variants: an analysis of mutational overlap between epithelial cancers. Journal of Human Genetics, 2020, 65, 199-205.	1.1	12
33	Prostate Cancer Risks for Male BRCA1 and BRCA2 Mutation Carriers: A Prospective Cohort Study. European Urology, 2020, 77, 24-35.	0.9	124
34	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. Nature Genetics, 2020, 52, 56-73.	9.4	120
35	Cancer Risks Associated With Germline PALB2 Pathogenic Variants: An International Study of 524 Families. Journal of Clinical Oncology, 2020, 38, 674-685.	0.8	270
36	Whole Exome Sequencing Identifies Candidate Genes Associated with Hereditary Predisposition to Uveal Melanoma. Ophthalmology, 2020, 127, 668-678.	2.5	27

#	Article	IF	Citations
37	Sporadic implementation of UK familial mammographic surveillance guidelines 15 years after original publication. British Journal of Cancer, 2020, 122, 329-332.	2.9	4
38	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.	1.1	82
39	Hereditary diffuse gastric cancer: updated clinical practice guidelines. Lancet Oncology, The, 2020, 21, e386-e397.	5.1	237
40	Association of germline variation with the survival of women with BRCA1/2 pathogenic variants and breast cancer. Npj Breast Cancer, 2020, 6, 44.	2.3	5
41	A case-control study of the joint effect of reproductive factors and radiation treatment for first breast cancer and risk of contralateral breast cancer in the WECARE study. Breast, 2020, 54, 62-69.	0.9	3
42	Genome-wide association study identifies 32 novel breast cancer susceptibility loci from overall and subtype-specific analyses. Nature Genetics, 2020, 52, 572-581.	9.4	265
43	Cancer Surveillance Guideline for individuals with PTEN hamartoma tumour syndrome. European Journal of Human Genetics, 2020, 28, 1387-1393.	1.4	63
44	Cancer Variant Interpretation Group UK (CanVIG-UK): an exemplar national subspecialty multidisciplinary network. Journal of Medical Genetics, 2020, 57, 829-834.	1.5	30
45	Personalized early detection and prevention of breast cancer: ENVISION consensus statement. Nature Reviews Clinical Oncology, 2020, 17, 687-705.	12.5	178
46	Prostate Cancer Risk by BRCA2 Genomic Regions. European Urology, 2020, 78, 494-497.	0.9	6
47	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.	1.1	40
48	Characterization of the Cancer Spectrum in Men With GermlineBRCA1andBRCA2Pathogenic Variants. JAMA Oncology, 2020, 6, 1218.	3.4	48
49	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.	3.0	21
50	Ovarian and Breast Cancer Risks Associated With Pathogenic Variants in RAD51C and RAD51D. Journal of the National Cancer Institute, 2020, 112, 1242-1250.	3.0	106
51	Transcriptomeâ€wide association study of breast cancer risk by estrogenâ€receptor status. Genetic Epidemiology, 2020, 44, 442-468.	0.6	32
52	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.	1.1	24
53	Characterization of renal cell carcinomaâ€associated constitutional chromosome abnormalities by genome sequencing. Genes Chromosomes and Cancer, 2020, 59, 333-347.	1.5	10
54	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.	2.2	41

#	Article	IF	Citations
55	Small-Cell Carcinoma of the Ovary, Hypercalcemic Typeâ€“Genetics, New Treatment Targets, and Current Management Guidelines. Clinical Cancer Research, 2020, 26, 3908-3917.	3.2	82
56	Title is missing!. , 2020, 15, e0229999.		0
57	Title is missing!. , 2020, 15, e0229999.		0
58	Title is missing!. , 2020, 15, e0229999.		0
59	Title is missing!. , 2020, 15, e0229999.		0
60	Genotype, extrapyramidal features, and severity of variant ataxiaâ€telangiectasia. Annals of Neurology, 2019, 85, 170-180.	2.8	58
61	Psychosocial impact of undergoing prostate cancer screening for men with <scp>BRCA</scp>1 or <scp>BRCA</scp>2 mutations. BJU International, 2019, 123, 284-292.	1.3	9
62	Homologous recombination DNA repair defects in PALB2-associated breast cancers. Npj Breast Cancer, 2019, 5, 23.	2.3	39
63	Caveat Emptor: The Perils of Panel Testing in Hereditary Breast Cancer. Journal of Clinical Oncology, 2019, 37, 2176-2177.	0.8	2
64	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.	0.7	46
65	The FANCM:p.Arg658* truncating variant is associated with risk of triple-negative breast cancer. Npj Breast Cancer, 2019, 5, 38.	2.3	28
66	Malta (MYH9 Associated Elastin Aggregation) Syndrome: Germline Variants in MYH9 Cause RareÂSweat Duct Proliferations and Irregular ElastinÂAggregations. Journal of Investigative Dermatology, 2019, 139, 2238-2241.e6.	0.3	5
67	Interim Results from the IMPACT Study: Evidence for Prostate-specific Antigen Screening in BRCA2 Mutation Carriers. European Urology, 2019, 76, 831-842.	0.9	148
68	Association of a Pathway-Specific Genetic Risk Score With Risk of Radiation-Associated Contralateral Breast Cancer. JAMA Network Open, 2019, 2, e1912259.	2.8	5
69	Shared heritability and functional enrichment across six solid cancers. Nature Communications, 2019, 10, 431.	5.8	88
70	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.	1.1	102
71	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.	2.9	19
72	Flexible and scalable diagnostic filtering of genomic variants using G2P with Ensembl VEP. Nature Communications, 2019, 10, 2373.	5.8	86

#	Article	IF	Citations
73	BRCA1andBRCA2pathogenic sequence variants in women of African origin or ancestry. Human Mutation, 2019, 40, 1781-1796.	1.1	26
74	The clinical presentation caused by truncating CHD8 variants. Clinical Genetics, 2019, 96, 72-84.	1.0	32
75	Towards controlled terminology for reporting germline cancer susceptibility variants: an ENIGMA report. Journal of Medical Genetics, 2019, 56, 347-357.	1.5	32
76	Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. Nature Communications, 2019, 10, 1741.	5.8	90
77	The Manchester International Consensus Group recommendations for the management of gynecological cancers in Lynch syndrome. Genetics in Medicine, 2019, 21, 2390-2400.	1.1	153
78	Letter from the New Editor in Chief. Genetical Research, 2019, 101, e2.	0.3	0
79	Clinical implications of germline mutations in breast cancer genes: RECQL. Breast Cancer Research and Treatment, 2019, 174, 553-560.	1.1	14
80	BOADICEA: a comprehensive breast cancer risk prediction model incorporating genetic and nongenetic risk factors. Genetics in Medicine, 2019, 21, 1708-1718.	1.1	415
81	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.	3.0	30
82	Cliniciansâ€™ use of breast cancer risk assessment tools according to their perceived importance of breast cancer risk factors: an international survey. Journal of Community Genetics, 2019, 10, 61-71.	0.5	7
83	Penetrance estimates for BRCA1, BRCA2 (also applied to Lynch syndrome) based on presymptomatic testing: a new unbiased method to assess risk?. Journal of Medical Genetics, 2018, 55, 442-448.	1.5	1
84	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.	1.5	88
85	Mutational spectrum in a worldwide study of 29,700 families withBRCA1orBRCA2mutations. Human Mutation, 2018, 39, 593-620.	1.1	224
86	Evaluation of universal immunohistochemical screening of sebaceous neoplasms in a service setting. Clinical and Experimental Dermatology, 2018, 43, 410-415.	0.6	13
87	Histone Lysine Methylases and Demethylases in the Landscape of Human Developmental Disorders. American Journal of Human Genetics, 2018, 102, 175-187.	2.6	204
88	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.	3.7	87
89	pedigreejs: a web-based graphical pedigree editor. Bioinformatics, 2018, 34, 1069-1071.	1.8	9
90	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.	1.1	15

#	Article	IF	Citations
91	Use of the BOADICEA Web Application in clinical practice: appraisals by clinicians from various countries. Familial Cancer, 2018, 17, 31-41.	0.9	9
92	Comparative study of endoscopic surveillance in hereditary diffuse gastric cancer according to CDH1 mutation status. Gastrointestinal Endoscopy, 2018, 87, 408-418.	0.5	85
93	Clinical implications of germline mutations in breast cancer: TP53. Breast Cancer Research and Treatment, 2018, 167, 417-423.	1.1	112
94	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.	0.8	44
95	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.	1.8	39
96	De Novo Truncating Mutations in WASF1 Cause Intellectual Disability with Seizures. American Journal of Human Genetics, 2018, 103, 144-153.	2.6	36
97	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.	0.4	54
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.	2.6	46
99	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.	2.6	26
100	Genetics of gynaecological cancers. Best Practice and Research in Clinical Obstetrics and Gynaecology, 2017, 42, 114-124.	1.4	13
101	Prevalence and architecture of de novo mutations in developmental disorders. Nature, 2017, 542, 433-438.	13.7	1,211
102	Pathology update to the Manchester Scoring System based on testing in over 4000 families. Journal of Medical Genetics, 2017, 54, 674-681.	1.5	51
103	Risks of Breast, Ovarian, and Contralateral Breast Cancer for BRCA1 and BRCA2 Mutation Carriers. JAMA - Journal of the American Medical Association, 2017, 317, 2402.	3.8	1,898
104	Universal BRCA1/BRCA2 Testing for Ovarian Cancer Patients is Welcomed, but with Care: How Women and Staff Contextualize Experiences of Expanded Access. Journal of Genetic Counseling, 2017, 26, 1280-1291.	0.9	14
105	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. Nature Genetics, 2017, 49, 680-691.	9.4	356
106	Compromised BRCA1â€“PALB2 interaction is associated with breast cancer risk. Oncogene, 2017, 36, 4161-4170.	2.6	71
107	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.	2.6	343
108	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

#	Article	IF	Citations
109	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. Nature Genetics, 2017, 49, 1767-1778.	9.4	289
110	Jejunal atresia, periodic fevers and psoriatic arthropathy in Baraitserâ€“Winter malformation syndrome. Clinical Dysmorphology, 2017, 26, 235-237.	0.1	2
111	When to Consider Riskâ€Reducing Mastectomy in BRCA1/BRCA2 Mutation Carriers with Advanced Stage Ovarian Cancer: a Case Study Illustrating the Genetic Counseling Challenges. Journal of Genetic Counseling, 2017, 26, 1173-1178.	0.9	4
112	The use of panel testing in familial breast and ovarian cancer. Clinical Medicine, 2017, 17, 568-572.	0.8	6
113	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.	1.1	18
114	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.	2.2	27
115	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.	0.8	152
116	PARTNER: Randomised, phase II/III trial to evaluate the safety and efficacy of the addition of olaparib to platinum-based neoadjuvant chemotherapy in triple negative and/or germline BRCA mutated breast cancer patients.. Journal of Clinical Oncology, 2017, 35, TPS591-TPS591.	0.8	7
117	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.	0.8	43
118	PALB2,CHEK2andATMrare variants and cancer risk: data from COGS. Journal of Medical Genetics, 2016, 53, 800-811.	1.5	174
119	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.	2.2	31
120	A comparison of the yield from endoscopic surveillance in detecting early gastric cancer in CDH1+ve versus CDH1â^'ve HDGC families. European Journal of Surgical Oncology, 2016, 42, S246-S247.	0.5	0
121	Incorporating truncating variants in PALB2, CHEK2, and ATM into the BOADICEA breast cancer risk model. Genetics in Medicine, 2016, 18, 1190-1198.	1.1	80
122	An international survey of surveillance schemes for unaffected BRCA1 and BRCA2 mutation carriers. Breast Cancer Research and Treatment, 2016, 157, 319-327.	1.1	26
123	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.	1.3	22
124	Inheritance of deleterious mutations at both BRCA1 and BRCA2 in an international sample of 32,295 women. Breast Cancer Research, 2016, 18, 112.	2.2	42
125	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.	1.5	57
126	Multilocus Inherited Neoplasia Alleles Syndrome. JAMA Oncology, 2016, 2, 373.	3.4	43

#	Article	IF	Citations
127	Breast cancer risk variants at 6q25 display different phenotype associations and regulate ESR1, RMND1 and CCDC170. Nature Genetics, 2016, 48, 374-386.	9.4	125
128	BRCA2 Polymorphic Stop Codon K3326X and the Risk of Breast, Prostate, and Ovarian Cancers. Journal of the National Cancer Institute, 2016, 108, djv315.	3.0	77
129	No clinical utility of KRAS variant rs61764370 for ovarian or breast cancer. Gynecologic Oncology, 2016, 141, 386-401.	0.6	18
130	Abstract 134: Mutational landscape of breast cancers from PALB2 germline mutation carriers. , 2016, , .		0
131	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.	2.2	26
132	Gene-Panel Sequencing and the Prediction of Breast-Cancer Risk. New England Journal of Medicine, 2015, 372, 2243-2257.	13.9	764
133	A homozygousPMS2founder mutation with an attenuated constitutional mismatch repair deficiency phenotype. Journal of Medical Genetics, 2015, 52, 348-352.	1.5	30
134	Germline Mutation in BRCA1 or BRCA2 and Ten-Year Survival for Women Diagnosed with Epithelial Ovarian Cancer. Clinical Cancer Research, 2015, 21, 652-657.	3.2	138
135	Identification of six new susceptibility loci for invasive epithelial ovarian cancer. Nature Genetics, 2015, 47, 164-171.	9.4	221
136	Hereditary diffuse gastric cancer: updated clinical guidelines with an emphasis on germlineCDH1mutation carriers. Journal of Medical Genetics, 2015, 52, 361-374.	1.5	479
137	Breast cancer risk in women with PALB2 mutations in different populations. Lancet Oncology, The, 2015, 16, e375-e376.	5.1	9
138	Association of Type and Location ofBRCA1andBRCA2Mutations With Risk of Breast and Ovarian Cancer. JAMA - Journal of the American Medical Association, 2015, 313, 1347.	3.8	390
139	Malignant Peripheral Nerve Sheath Tumor in Cowden Syndrome. Journal of Neuropathology and Experimental Neurology, 2015, 74, 288-292.	0.9	4
140	Heimler Syndrome Is Caused by Hypomorphic Mutations in the Peroxisome-Biogenesis Genes PEX1 and PEX6. American Journal of Human Genetics, 2015, 97, 535-545.	2.6	103
141	Risk reducing salpingectomy and delayed oophorectomy in high risk women: views of cancer geneticists, genetic counsellors and gynaecological oncologists in the UK. Familial Cancer, 2015, 14, 521-530.	0.9	14
142	Biallelic somaticSMARCA4mutations in small cell carcinoma of the ovary, hypercalcemic type (SCCOHT). Pediatric Blood and Cancer, 2015, 62, 728-730.	0.8	16
143	Fanconi anaemia: genetics, molecular biology, and cancerÂâ€“Âimplications for clinical management in children and adults. Clinical Genetics, 2015, 88, 13-24.	1.0	69
144	Characterization of a novel founder <scp>MSH6</scp> mutation causing Lynch syndrome in the French Canadian population. Clinical Genetics, 2015, 87, 536-542.	1.0	13

#	Article	IF	Citations
145	Effect of BRCA Mutations on Metastatic Relapse and Cause-specific Survival After Radical Treatment for Localised Prostate Cancer. European Urology, 2015, 68, 186-193.	0.9	279
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.	1.1	22
147	Breast-Cancer Risk in Families with Mutations in PALB2. New England Journal of Medicine, 2014, 371, 1650-1652.	13.9	68
148	DNA Glycosylases Involved in Base Excision Repair May Be Associated with Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. PLoS Genetics, 2014, 10, e1004256.	1.5	47
149	Serum levels of mature microRNAs in DICER1-mutated pleuropulmonary blastoma. Oncogenesis, 2014, 3, e87-e87.	2.1	40
150	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.	0.6	19
151	Breast-Cancer Risk in Families With Mutations in PALB2. Obstetrical and Gynecological Survey, 2014, 69, 659-660.	0.2	1
152	Lymphocyte Telomere Length Is Long in BRCA1 and BRCA2 Mutation Carriers Regardless of Cancer-Affected Status. Cancer Epidemiology Biomarkers and Prevention, 2014, 23, 1018-1024.	1.1	13
153	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.	2.2	57
154	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.	0.5	75
155	Informed decision-making is the key in women at high risk of breast cancer. European Journal of Surgical Oncology, 2014, 40, 667-669.	0.5	3
156	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.	0.9	195
157	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.	1.5	48
158	Germline and somatic SMARCA4 mutations characterize small cell carcinoma of the ovary, hypercalcemic type. Nature Genetics, 2014, 46, 438-443.	9.4	383
159	Breast-Cancer Risk in Families with Mutations in PALB2. New England Journal of Medicine, 2014, 371, 497-506.	13.9	745
160	Abstract LB-89: Germ-line and somatic SMARCA4 mutations characterize small cell carcinoma of the ovary, hypercalcemic type. , 2014, , .		0
161	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.	0.8	641
162	Cancer Risks for BRCA1 and BRCA2 Mutation Carriers: Results From Prospective Analysis of EMBRACE. Journal of the National Cancer Institute, 2013, 105, 812-822.	3.0	753

#	Article	IF	Citations
163	Hi-Plex for high-throughput mutation screening: application to the breast cancer susceptibility gene PALB2. BMC Medical Genomics, 2013, 6, 48.	0.7	13
164	A germline mosaic <scp>BRCA1</scp> exon deletion in a woman with bilateral basalâ€like breast cancer. Clinical Genetics, 2013, 84, 297-299.	1.0	8
165	Analysis of the Novel Fanconi Anemia GeneSLX4/FANCPin Familial Breast Cancer Cases. Human Mutation, 2013, 34, 70-73.	1.1	21
166	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
167	Heterozygous mutations in the PALB2 hereditary breast cancer predisposition gene impact on the threeâ€dimensional nuclear organization of patientâ€derived cell lines. Genes Chromosomes and Cancer, 2013, 52, 480-494.	1.5	6
168	Identification of a BRCA2-Specific Modifier Locus at 6p24 Related to Breast Cancer Risk. PLoS Genetics, 2013, 9, e1003173.	1.5	105
169	Genome-Wide Association Study in BRCA1 Mutation Carriers Identifies Novel Loci Associated with Breast and Ovarian Cancer Risk. PLoS Genetics, 2013, 9, e1003212.	1.5	244
170	Biallelic Deleterious BRCA1 Mutations in a Woman with Early-Onset Ovarian Cancer. Cancer Discovery, 2013, 3, 399-405.	7.7	124
171	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.	1.1	23
172	Mutation analysis of RAD51D in non-BRCA1/2 ovarian and breast cancer families. British Journal of Cancer, 2012, 106, 1460-1463.	2.9	43
173	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.	1.1	47
174	Alagille Syndrome and Wilson Disease in Siblings: A Diagnostic Conundrum. Canadian Journal of Gastroenterology & Hepatology, 2012, 26, 330-332.	1.8	2
175	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.	3.1	25
176	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.	1.1	23
177	Rare germline mutations inPALB2and breast cancer risk: A population-based study. Human Mutation, 2012, 33, 674-680.	1.1	74
178	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.	1.1	16
179	Germline PALB2 mutation analysis in breast-pancreas cancer families. Journal of Medical Genetics, 2011, 48, 523-525.	1.5	28
180	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.	5.1	1,028

#	Article	IF	Citations
181	Characterization of two Ashkenazi Jewish founder mutations in MSH6 gene causing Lynch syndrome. Clinical Genetics, 2011, 79, 512-522.	1.0	27
182	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.	1.3	83
183	Dermatological manifestations of inherited cancer syndromes in children. British Journal of Dermatology, 2011, 164, 245-256.	1.4	46
184	Mutation analysis of the PALB2 cancer predisposition gene in familial melanoma. Familial Cancer, 2011, 10, 315-317.	0.9	9
185	A survey of APC mutations in Quebec. Familial Cancer, 2011, 10, 659-665.	0.9	11
186	Acinic cell carcinoma of the retromolar trigone region: expanding the tumor phenotype in Cowden syndrome?. Familial Cancer, 2011, 10, 691-694.	0.9	11
187	International distribution and age estimation of the Portuguese BRCA2 c.156_157insAlu founder mutation. Breast Cancer Research and Treatment, 2011, 127, 671-679.	1.1	27
188	Using mouse models to investigate the biological and physiological consequences of defects in the Fanconi anaemia/breast cancer DNA repair signalling pathway. Journal of Pathology, 2011, 224, 301-305.	2.1	10
189	Extending the phenotypes associated withDICER1mutations. Human Mutation, 2011, 32, 1381-1384.	1.1	173
190	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.	1.4	68
191	miRNA Processing and Human Cancer: DICER1 Cuts the Mustard. Science Translational Medicine, 2011, 3, 111ps46.	5.8	60
192	<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.	3.8	284
193	PALB2/FANCN: Recombining Cancer and Fanconi Anemia. Cancer Research, 2010, 70, 7353-7359.	0.4	187
194	Mutation analysis of the gene encoding the PALB2-binding protein MRG15 in BRCA1/2-negative breast cancer families. Journal of Human Genetics, 2010, 55, 842-843.	1.1	6
195	HomozygousBUB1BMutation and Susceptibility to Gastrointestinal Neoplasia. New England Journal of Medicine, 2010, 363, 2628-2637.	13.9	82
196	Germline DICER1 mutations and familial cystic nephroma. Journal of Medical Genetics, 2010, 47, 863-866.	1.5	113
197	A PALB2 mutation associated with high risk of breast cancer. Breast Cancer Research, 2010, 12, R109.	2.2	102
198	RAD51C germline mutations in breast and ovarian cancer patients. Breast Cancer Research, 2010, 12, 404.	2.2	50

#	Article	IF	Citations
199	Abstract 2975: Two putative founder MSH6 mutations associated with inherited cancer susceptibility in Ashkenazi Jewish population. , 2010, , .		0
200	Spinal Hamartoma with Severe Kyphoscoliosis â€“ A Rare Case of Spinal Mass in a Fetus. Fetal Diagnosis and Therapy, 2009, 25, 11-14.	0.6	6
201	A comparison of models used to predict MLH1, MSH2 and MSH6 mutation carriers. Annals of Oncology, 2009, 20, 681-688.	0.6	26
202	High frequency of exon deletions and putative founder effects in French Canadian Lynch syndrome families. Human Mutation, 2009, 30, E797-E812.	1.1	14
203	Analysis of the genes coding for the BRCA1-interacting proteins, RAP80 and Abraxas (CCDC98), in high-risk, non-BRCA1/2, multiethnic breast cancer cases. Breast Cancer Research and Treatment, 2009, 117, 453-459.	1.1	14
204	Hereditary breast cancer: new genetic developments, new therapeutic avenues. Human Genetics, 2008, 124, 31-42.	1.8	276
205	Analysis of the gene coding for the BRCA2â€‐interacting protein PALB2 in hereditary prostate cancer. Prostate, 2008, 68, 675-678.	1.2	36
206	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.	1.4	42
207	Cancer incidence in relatives of British Fanconi Anaemia patients. BMC Cancer, 2008, 8, 257.	1.1	23
208	Identification and characterization of novel SNPs in CHEK2 in Ashkenazi Jewish men with prostate cancer. Cancer Letters, 2008, 270, 173-180.	3.2	19
209	Large Genomic Deletions inAIPin Pituitary Adenoma Predisposition. Journal of Clinical Endocrinology and Metabolism, 2008, 93, 4146-4151.	1.8	74
210	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.	3.3	192
211	Germline CDKN1B/p27Kip1 Mutation in Multiple Endocrine Neoplasia. Journal of Clinical Endocrinology and Metabolism, 2007, 92, 3321-3325.	1.8	262
212	Founder and Recurrent CDH1 Mutations in Families With Hereditary Diffuse Gastric Cancer. JAMA - Journal of the American Medical Association, 2007, 297, 2360.	3.8	394
213	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.	2.2	126
214	Biallelic mutations in PALB2 cause Fanconi anemia subtype FA-N and predispose to childhood cancer. Nature Genetics, 2007, 39, 162-164.	9.4	556
215	Use of immunohistochemical markers can refine prognosis in triple negative breast cancer. BMC Cancer, 2007, 7, 134.	1.1	316
216	Muir Torre syndrome and MSH2 mutations: the importance of dermatological awareness. British Journal of Cancer, 2006, 95, 243-244.	2.9	2

#	Article	IF	Citations
217	The Basal Phenotype of BRCA1-Related Breast Cancer: Past, Present and Future. Cell Cycle, 2006, 5, 963-967.	1.3	46
218	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.	0.6	89
219	A common Fanconi anemia mutation in black populations of sub-Saharan Africa. Blood, 2005, 105, 3542-3544.	0.6	53
220	Nijmegen breakage syndrome diagnosed as Fanconi anaemia. Pediatric Blood and Cancer, 2005, 44, 494-499.	0.8	29
221	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.	1.3	90
222	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.	0.6	21
223	Autosomal dominant B-cell immunodeficiency, distal limb anomalies and urogenital malformations (BILU syndrome) - report of a second family. Clinical Genetics, 2004, 66, 550-555.	1.0	4
224	Fanconi anaemia and leukaemia - clinical and molecular aspects. British Journal of Haematology, 2004, 126, 176-191.	1.2	124
225	Deletion and reduced expression of the Fanconi anemia FANCA gene in sporadic acute myeloid leukemia. Leukemia, 2004, 18, 420-425.	3.3	78
226	Inherited cancer in children: practical/ethical problems and challenges. European Journal of Cancer, 2004, 40, 2459-2470.	1.3	32
227	Medulloblastoma as a First Presentation of Fanconi Anemia. Journal of Pediatric Hematology/Oncology, 2004, 26, 52-55.	0.3	14
228	Screening Those at High Risk for Colorectal Cancer. Medical Radiology, 2004, , 13-23.	0.0	0
229	Bi-allelic silencing of the Fanconi anaemia gene FANCF in acute myeloid leukaemia. British Journal of Haematology, 2003, 123, 469-471.	1.2	65
230	Disruption of the Fanconi anemiaâ€“BRCA pathway in cisplatin-sensitive ovarian tumors. Nature Medicine, 2003, 9, 568-574.	15.2	508
231	Mutations in BRCA1 and BRCA2 and predisposition to prostate cancer. Lancet, The, 2003, 362, 80.	6.3	9
232	Fanconi anaemia. Journal of Medical Genetics, 2003, 40, 1-10.	1.5	243
233	Acquired FANCA dysfunction and cytogenetic instability in adult acute myelogenous leukemia. Blood, 2003, 102, 7-16.	0.6	56
234	Phase II trial of liposomal daunorubicin in malignant pleural mesothelioma. Annals of Oncology, 2001, 12, 497-499.	0.6	35

#	Article	IF	Citations
235	Phase II Study of Vinorelbine in Patients With Malignant Pleural Mesothelioma. Journal of Clinical Oncology, 2000, 18, 3912-3917.	0.8	192
236	A comparison of methods currently used in clinical practice to estimate familial breast cancer risks. Annals of Oncology, 2000, 11, 451-454.	0.6	16
237	Randomized Trial of a Specialist Genetic Assessment Service for Familial Breast Cancer. Journal of the National Cancer Institute, 2000, 92, 1345-1351.	3.0	89
238	Familial gonadal tumours.. Journal of Medical Genetics, 1998, 35, 84-84.	1.5	0