William D Foulkes

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/1162012/publications.pdf

Version: 2024-02-01

650 papers	46,524 citations	1463 107 h-index	2895 190 g-index
674 all docs	674 docs citations	674 times ranked	42207 citing authors

#	Article	IF	Citations
1	Triple-Negative Breast Cancer. New England Journal of Medicine, 2010, 363, 1938-1948.	27.0	3,233
2	REVEL: An Ensemble Method for Predicting the Pathogenicity of Rare Missense Variants. American Journal of Human Genetics, 2016, 99, 877-885.	6.2	1,555
3	BRCA1 and BRCA2: 1994 and beyond. Nature Reviews Cancer, 2004, 4, 665-676.	28.4	824
4	Germline BRCA1 Mutations and a Basal Epithelial Phenotype in Breast Cancer. Journal of the National Cancer Institute, 2003, 95, 1482-1485.	6.3	807
5	Sequence variant classification and reporting: recommendations for improving the interpretation of cancer susceptibility genetic test results. Human Mutation, 2008, 29, 1282-1291.	2.5	782
6	Subtyping of Breast Cancer by Immunohistochemistry to Investigate a Relationship between Subtype and Short and Long Term Survival: A Collaborative Analysis of Data for 10,159 Cases from 12 Studies. PLoS Medicine, 2010, 7, e1000279.	8.4	764
7	Gene-Panel Sequencing and the Prediction of Breast-Cancer Risk. New England Journal of Medicine, 2015, 372, 2243-2257.	27.0	764
8	Breast-Cancer Risk in Families with Mutations in PALB2. New England Journal of Medicine, 2014, 371, 497-506.	27.0	745
9	Contralateral Breast Cancer in BRCA1 and BRCA2 Mutation Carriers. Journal of Clinical Oncology, 2004, 22, 2328-2335.	1.6	595
10	Familial Adenomatous Polyposis. American Journal of Gastroenterology, 2006, 101, 385-398.	0.4	575
11	Salpingo-oophorectomy and the Risk of Ovarian, Fallopian Tube, and Peritoneal Cancers in Women With a <EMPH TYPE="ITAL">BRCA1</EMPH> or <EMPH TYPE="ITAL">BRCA2</EMPH> Mutation. JAMA - Journal of the American Medical Association, 2006, 296, 185.	7.4	544
12	Impact of Oophorectomy on Cancer Incidence and Mortality in Women With a BRCA1 or BRCA2 Mutation. Journal of Clinical Oncology, 2014, 32, 1547-1553.	1.6	523
13	Inherited Susceptibility to Common Cancers. New England Journal of Medicine, 2008, 359, 2143-2153.	27.0	462
14	Is Breast Cancer the Same Disease in Asian and Western Countries?. World Journal of Surgery, 2010, 34, 2308-2324.	1.6	460
15	A mutational signature reveals alterations underlying deficient homologous recombination repair in breast cancer. Nature Genetics, 2017, 49, 1476-1486.	21.4	427
16	Triple-Negative Breast Cancer: Distinguishing between Basal and Nonbasal Subtypes. Clinical Cancer Research, 2009, 15, 2302-2310.	7.0	422
17	Breast Cancer Risk Following Bilateral Oophorectomy in BRCA1 and BRCA2 Mutation Carriers: An International Case-Control Study. Journal of Clinical Oncology, 2005, 23, 7491-7496.	1.6	408
18	DICER1: mutations, microRNAs and mechanisms. Nature Reviews Cancer, 2014, 14, 662-672.	28.4	404

#	Article	IF	Citations
19	Recurrent SomaticDICER1Mutations in Nonepithelial Ovarian Cancers. New England Journal of Medicine, 2012, 366, 234-242.	27.0	401
20	Identification of seven new prostate cancer susceptibility loci through a genome-wide association study. Nature Genetics, 2009, 41, 1116-1121.	21.4	389
21	Germline and somatic SMARCA4 mutations characterize small cell carcinoma of the ovary, hypercalcemic type. Nature Genetics, 2014, 46, 438-443.	21.4	383
22	Triple-Negative Breast Cancer: Risk Factors to Potential Targets. Clinical Cancer Research, 2008, 14, 8010-8018.	7.0	380
23	CD8+ lymphocyte infiltration is an independent favorable prognostic indicator in basal-like breast cancer. Breast Cancer Research, 2012, 14, R48.	5.0	376
24	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. Nature Genetics, 2017, 49, 680-691.	21.4	356
25	Intrinsic Breast Tumor Subtypes, Race, and Long-Term Survival in the Carolina Breast Cancer Study. Clinical Cancer Research, 2010, 16, 6100-6110.	7.0	351
26	The Prognostic Implication of the Basal-Like (Cyclin) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 467 Td (Ehigh/p27low/p53+/Glomeruloid-Micr-Related Breast Cancer. Cancer Research, 2004, 64, 830-835.	0.9	345
27	Oral Contraceptives and the Risk of Breast Cancer in BRCA1 and BRCA2 Mutation Carriers. Journal of the National Cancer Institute, 2002, 94, 1773-1779.	6.3	318
28	Use of immunohistochemical markers can refine prognosis in triple negative breast cancer. BMC Cancer, 2007, 7, 134.	2.6	316
29	International variation in rates of uptake of preventive options in BRCA1 and BRCA2 mutation carriers. International Journal of Cancer, 2008, 122, 2017-2022.	5.1	306
30	Frequency of recurrent BRCA1 and BRCA2 mutations in Ashkenazi Jewish breast cancer families. Nature Medicine, 1996, 2, 1179-1183.	30.7	294
31	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. Nature Genetics, 2017, 49, 1767-1778.	21.4	289
32	<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
33	Hereditary breast cancer: new genetic developments, new therapeutic avenues. Human Genetics, 2008, 124, 31-42.	3.8	276
34	Estrogen Receptor Status in BRCA1 - and BRCA2 -Related Breast Cancer. Clinical Cancer Research, 2004, 10, 2029-2034.	7.0	270
35	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
36	A combined analysis of outcome following breast cancer: differences in survival based on BRCA1/BRCA2 mutation status and administration of adjuvant treatment. Breast Cancer Research, 2003, 6, R8-R17.	5.0	262

#	Article	IF	Citations
37	Prospective Study of Breast Cancer Incidence in Women With a BRCA1 or BRCA2 Mutation Under Surveillance With and Without Magnetic Resonance Imaging. Journal of Clinical Oncology, 2011, 29, 1664-1669.	1.6	248
38	Tamoxifen and contralateral breast cancer inBRCA1 andBRCA2 carriers: An update. International Journal of Cancer, 2006, 118, 2281-2284.	5.1	246
39	Contralateral mastectomy and survival after breast cancer in carriers of BRCA1 and BRCA2 mutations: retrospective analysis. BMJ, The, 2014, 348, g226-g226.	6.0	238
40	The CDKN2A (p16) Gene and Human Cancer. Molecular Medicine, 1997, 3, 5-20.	4.4	228
41	The incidence of pancreatic cancer in BRCA1 and BRCA2 mutation carriers. British Journal of Cancer, 2012, 107, 2005-2009.	6.4	221
42	Reproductive risk factors for ovarian cancer in carriers of BRCA1 or BRCA2 mutations: a case-control study. Lancet Oncology, The, 2007, 8, 26-34.	10.7	220
43	Tumor necrosis factor and its receptors in human ovarian cancer. Potential role in disease progression.. Journal of Clinical Investigation, 1993, 91, 2194-2206.	8.2	219
44	Pituitary blastoma: a pathognomonic feature of germ-line DICER1 mutations. Acta Neuropathologica, 2014, 128, 111-122.	7.7	211
45	Identification of genes associated with head and neck carcinogenesis by cDNA microarray comparison between matched primary normal epithelial and squamous carcinoma cells. Oncogene, 2002, 21, 2634-2640.	5.9	204
46	Achieving clinical success with BET inhibitors as anti-cancer agents. British Journal of Cancer, 2021, 124, 1478-1490.	6.4	204
47	Familial Nontoxic Multinodular Thyroid Goiter Locus Maps to Chromosome 14q but Does Not Account for Familial Nonmedullary Thyroid Cancer. American Journal of Human Genetics, 1997, 61, 1123-1130.	6.2	203
48	Pregnancy and risk of early breast cancer in carriers of BRCA1 and BRCA2. Lancet, The, 1999, 354, 1846-1850.	13.7	202
49	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
50	Predictors of contralateral breast cancer in BRCA1 and BRCA2 mutation carriers. British Journal of Cancer, 2011, 104, 1384-1392.	6.4	195
51	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
52	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
53	A Basal Epithelial Phenotype Is More Frequent in Interval Breast Cancers Compared with Screen Detected Tumors. Cancer Epidemiology Biomarkers and Prevention, 2005, 14, 1108-1112.	2.5	185
54	Prognostic significance of FOXP3+ tumor-infiltrating lymphocytes in breast cancer depends on estrogen receptor and human epidermal growth factor receptor-2 expression status and concurrent cytotoxic T-cell infiltration. Breast Cancer Research, 2014, 16, 432.	5.0	182

#	Article	IF	Citations
55	Methionine Metabolism Shapes T Helper Cell Responses through Regulation of Epigenetic Reprogramming. Cell Metabolism, 2020, 31, 250-266.e9.	16.2	182
56	PALB2,CHEK2andATMrare variants and cancer risk: data from COGS. Journal of Medical Genetics, 2016, 53, 800-811.	3.2	174
57	Extending the phenotypes associated withDICER1mutations. Human Mutation, 2011, 32, 1381-1384.	2.5	173
58	Breast-feeding and the Risk of Breast Cancer in BRCA1 and BRCA2 Mutation Carriers. Journal of the National Cancer Institute, 2004, 96, 1094-1098.	6.3	172
59	Germline RECQL mutations are associated with breast cancer susceptibility. Nature Genetics, 2015, 47, 643-646.	21.4	168
60	HOXB13 is a susceptibility gene for prostate cancer: results from the International Consortium for Prostate Cancer Genetics (ICPCG). Human Genetics, 2013, 132, 5-14.	3.8	166
61	Retinoblastoma and Neuroblastoma Predisposition and Surveillance. Clinical Cancer Research, 2017, 23, e98-e106.	7.0	166
62	PREDICT Plus: development and validation of a prognostic model for early breast cancer that includes HER2. British Journal of Cancer, 2012, 107, 800-807.	6.4	163
63	Influence of BRCA1 mutations on nuclear grade and estrogen receptor status of breast carcinoma in Ashkenazi Jewish women. Cancer, 1997, 80, 435-441.	4.1	162
64	Predictors of Contralateral Prophylactic Mastectomy in Women With a BRCA1 or BRCA2 Mutation: The Hereditary Breast Cancer Clinical Study Group. Journal of Clinical Oncology, 2008, 26, 1093-1097.	1.6	161
65	Bilateral Oophorectomy and Breast Cancer Risk inBRCA1andBRCA2Mutation Carriers. Journal of the National Cancer Institute, 2017, 109, .	6.3	160
66	Placental Cadherin and the Basal Epithelial Phenotype of BRCA1-Related Breast Cancer. Clinical Cancer Research, 2005, 11, 4003-4011.	7.0	157
67	Founder BRCA1 and BRCA2 Mutations in French Canadian Breast and Ovarian Cancer Families. American Journal of Human Genetics, 1998, 63, 1341-1351.	6.2	156
68	SMARCA4-mutated atypical teratoid/rhabdoid tumors are associated with inherited germline alterations and poor prognosis. Acta Neuropathologica, 2014, 128, 453-456.	7.7	155
69	CARD9 Deficiency and Spontaneous Central Nervous System Candidiasis: Complete Clinical Remission With GM-CSF Therapy. Clinical Infectious Diseases, 2014, 59, 81-84.	5.8	153
70	Effect of pregnancy as a risk factor for breast cancer in BRCA1/BRCA2 mutation carriers. International Journal of Cancer, 2005, 117, 988-991.	5.1	152
71	TP53 mutations in breast cancer associated with BRCA1 or BRCA2 germ-line mutations: distinctive spectrum and structural distribution. Cancer Research, 2001, 61, 4092-7.	0.9	152
72	Germline HAVCR2 mutations altering TIM-3 characterize subcutaneous panniculitis-like T cell lymphomas with hemophagocytic lymphohistiocytic syndrome. Nature Genetics, 2018, 50, 1650-1657.	21.4	151

#	Article	IF	Citations
73	Multiple Novel Prostate Cancer Predisposition Loci Confirmed by an International Study: The PRACTICAL Consortium. Cancer Epidemiology Biomarkers and Prevention, 2008, 17, 2052-2061.	2.5	148
74	BRCA Challenge: BRCA Exchange as a global resource for variants in BRCA1 and BRCA2. PLoS Genetics, 2018, 14, e1007752.	3.5	148
75	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
76	BRCA1 functions as a breast stem cell regulator. Journal of Medical Genetics, 2004, 41, 1-5.	3.2	146
77	Germline and somatic FGFR1 abnormalities in dysembryoplastic neuroepithelial tumors. Acta Neuropathologica, 2016, 131, 847-863.	7.7	143
78	The risk of ovarian cancer after breast cancer in BRCA1 and BRCA2 carriers. Gynecologic Oncology, 2005, 96, 222-226.	1.4	141
79	Cancer Surveillance in Gorlin Syndrome and Rhabdoid Tumor Predisposition Syndrome. Clinical Cancer Research, 2017, 23, e62-e67.	7.0	139
80	A Combined Genomewide Linkage Scan of 1,233 Families for Prostate Cancerâ€“Susceptibility Genes Conducted by the International Consortium for Prostate Cancer Genetics. American Journal of Human Genetics, 2005, 77, 219-229.	6.2	138
81	Germ-line and somatic DICER1 mutations in pineoblastoma. Acta Neuropathologica, 2014, 128, 583-595.	7.7	137
82	A somatic BRCA1 mutation in an ovarian tumour. Nature Genetics, 1995, 9, 343-344.	21.4	135
83	A significant response to neoadjuvant chemotherapy in BRCA1/2 related breast cancer. Journal of Medical Genetics, 2002, 39, 608-610.	3.2	135
84	Analysis of the Gene Coding for the BRCA2-Interacting Protein PALB2 in Familial and Sporadic Pancreatic Cancer. Gastroenterology, 2009, 137, 1183-1186.	1.3	135
85	Increased Risk for Nonmedullary Thyroid Cancer in the First Degree Relatives of Prevalent Cases of Nonmedullary Thyroid Cancer: A Hospital-Based Study. Journal of Clinical Endocrinology and Metabolism, 2001, 86, 5307-5312.	3.6	132
86	Rapid progression of prostate cancer in men with a BRCA2 mutation. British Journal of Cancer, 2008, 99, 371-374.	6.4	132
87	Clinico-pathological characteristics of BRCA1- and BRCA2-related breast cancer. Journal of Surgical Oncology, 2000, 18, 287-295.	1.4	131
88	Extensive DNA methylation in normal colorectal mucosa in hyperplastic polyposis. Gut, 2006, 55, 1467-1474.	12.1	131
89	BRCA1 and BRCA2: Chemosensitivity, Treatment Outcomes and Prognosis. Familial Cancer, 2006, 5, 135-142.	1.9	128
90	PTEN, DICER1, FH, and Their Associated Tumor Susceptibility Syndromes: Clinical Features, Genetics, and Surveillance Recommendations in Childhood. Clinical Cancer Research, 2017, 23, e76-e82.	7.0	128

#	Article	IF	Citations
91	Biallelic <scp>DICER1</scp> mutations occur in Wilms tumours. Journal of Pathology, 2013, 230, 154-164.	4.5	127
92	Familial risks of squamous cell carcinoma of the head and neck: retrospective case-control study. BMJ: British Medical Journal, 1996, 313, 716-721.	2.3	127
93	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
94	SMARCA4 loss is synthetic lethal with CDK4/6 inhibition in non-small cell lung cancer. Nature Communications, 2019, 10, 557.	12.8	125
95	Biallelic Deleterious BRCA1 Mutations in a Woman with Early-Onset Ovarian Cancer. Cancer Discovery, 2013, 3, 399-405.	9.4	124
96	Linkage Analysis of Chromosome 1q Markers in 136 Prostate Cancer Families. American Journal of Human Genetics, 1998, 62, 653-658.	6.2	123
97	Mutational Signature Analysis Reveals NTHL1 Deficiency to Cause a Multi-tumor Phenotype. Cancer Cell, 2019, 35, 256-266.e5.	16.8	123
98	Hormone Replacement Therapy After Oophorectomy and Breast Cancer Risk Among BRCA1 Mutation Carriers. JAMA Oncology, 2018, 4, 1059.	7.1	121
99	Incidence of colorectal cancer in BRCA1 and BRCA2 mutation carriers: results from a follow-up study. British Journal of Cancer, 2014, 110, 530-534.	6.4	120
100	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. Nature Genetics, 2020, 52, 56-73.	21.4	120
101	Effect of Smoking on Breast Cancer in Carriers of Mutant BRCA1 or BRCA2 Genes. Journal of the National Cancer Institute, 1998, 90, 761-765.	6.3	118
102	Germline BRCA1/2 Mutations and p27^{Kip1} Protein Levels Independently Predict Outcome After Breast Cancer. Journal of Clinical Oncology, 2000, 18, 4045-4052.	1.6	118
103	The Founder Mutation MSH2*1906Gâ†'C Is an Important Cause of Hereditary Nonpolyposis Colorectal Cancer in the Ashkenazi Jewish Population. American Journal of Human Genetics, 2002, 71, 1395-1412.	6.2	118
104	No small surpriseÂâ€“Âsmall cell carcinoma of the ovary, hypercalcaemic type, is a malignant rhabdoid tumour. Journal of Pathology, 2014, 233, 209-214.	4.5	117
105	Frequent loss of heterozygosity on chromosome 6 in human ovarian carcinoma. British Journal of Cancer, 1993, 67, 551-559.	6.4	116
106	DICER1 Mutations Are Consistently Present in Moderately and Poorly Differentiated Sertoli-Leydig Cell Tumors. American Journal of Surgical Pathology, 2017, 41, 1178-1187.	3.7	114
107	Germline DICER1 mutations and familial cystic nephroma. Journal of Medical Genetics, 2010, 47, 863-866.	3.2	113
108	Molecular characterization of the spectrum of genomic deletions in the mismatch repair genes MSH2, MLH1, MSH6, and PMS2 responsible for hereditary nonpolyposis colorectal cancer (HNPCC). Genes Chromosomes and Cancer, 2005, 44, 123-138.	2.8	112

#	Article	IF	Citations
109	Loss of heterozygosity and amplification on chromosome 11q in human ovarian cancer. British Journal of Cancer, 1993, 67, 268-273.	6.4	111
110	DICER1 hotspot mutations in non-epithelial gonadal tumours. British Journal of Cancer, 2013, 109, 2744-2750.	6.4	111
111	Exploring the Association BetweenDICER1Mutations and Differentiated Thyroid Carcinoma. Journal of Clinical Endocrinology and Metabolism, 2014, 99, E1072-E1077.	3.6	111
112	Frequency of premature menopause in women who carry a BRCA1 or BRCA2 mutation. Fertility and Sterility, 2013, 99, 1724-1728.	1.0	110
113	The Histomorphology of Lynch Syndromeâ€"associated Ovarian Carcinomas. American Journal of Surgical Pathology, 2014, 38, 1173-1181.	3.7	108
114	E6/E7 proteins of HPV type 16 and ErbB-2 cooperate to induce neoplastic transformation of primary normal oral epithelial cells. Oncogene, 2004, 23, 350-358.	5.9	107
115	On the origin and diffusion of BRCA1 c.5266dupC (5382insC) in European populations. European Journal of Human Genetics, 2011, 19, 300-306.	2.8	107
116	Effect of Oophorectomy on Survival After Breast Cancer inBRCA1andBRCA2Mutation Carriers. JAMA Oncology, 2015, 1, 306.	7.1	107
117	The incidence of endometrial cancer in women with BRCA1 and BRCA2 mutations: An international prospective cohort study. Gynecologic Oncology, 2013, 130, 127-131.	1.4	106
118	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
119	Re: Magnetic Resonance Imaging and Mammography in Women With a Hereditary Risk of Breast Cancer. Journal of the National Cancer Institute, 2001, 93, 1754-1754.	6.3	105
120	Screening mammography and risk of breast cancer in BRCA1 and BRCA2 mutation carriers: a case-control study. Lancet Oncology, The, 2006, 7, 402-406.	10.7	104
121	Tumor size and survival in breast cancerâ€"a reappraisal. Nature Reviews Clinical Oncology, 2010, 7, 348-353.	27.6	104
122	Prognostic significance of CD8+ T lymphocytes in breast cancer depends upon both oestrogen receptor status and histological grade. Histopathology, 2011, 58, no-no.	2.9	104
123	Disruption of the expected positive correlation between breast tumor size and lymph node status inBRCA1-related breast carcinoma. Cancer, 2003, 98, 1569-1577.	4.1	103
124	Changes in body weight and the risk of breast cancer in BRCA1 and BRCA2mutation carriers. Breast Cancer Research, 2005, 7, R833-43.	5.0	103
125	A PALB2 mutation associated with high risk of breast cancer. Breast Cancer Research, 2010, 12, R109.	5.0	102
126	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

#	Article	IF	Citations
127	Pheochromocytoma and paraganglioma syndromes: genetics and management update. Current Oncology, 2014, 21, 8.	2.2	102
128	A novel gene encoding a B-box protein within the BRCA1 region at 17q21.1. Human Molecular Genetics, 1994, 3, 589-594.	2.9	98
129	Family history of cancer is a risk factor for squamous cell carcinoma of the head and neck in Brazil: A caseâ€control study. International Journal of Cancer, 1995, 63, 769-773.	5.1	98
130	Women's attitudes toward preventive strategies for hereditary breast or ovarian carcinoma differ from one country to another. Cancer, 2001, 92, 959-968.	4.1	98
131	Are triple-negative tumours and basal-like breast cancer synonymous?. Breast Cancer Research, 2007, 9, 404; author reply 405.	5.0	98
132	Poorly differentiated thyroid carcinoma of childhood and adolescence: a distinct entity characterized by DICER1 mutations. Modern Pathology, 2020, 33, 1264-1274.	5.5	96
133	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. Nature Communications, 2016, 7, 11375.	12.8	93
134	Tumour predisposition and cancer syndromes as models to study geneâ€“environment interactions. Nature Reviews Cancer, 2020, 20, 533-549.	28.4	93
135	An evaluation of needs of female BRCA1 and BRCA2 carriers undergoing genetic counselling. Journal of Medical Genetics, 2000, 37, 866-874.	3.2	92
136	Breastfeeding and the risk of breast cancer in BRCA1 and BRCA2 mutation carriers. Breast Cancer Research, 2012, 14, R42.	5.0	92
137	Impaired RASGRF1/ERKâ€“mediated GM-CSF response characterizes CARD9 deficiency in French-Canadians. Journal of Allergy and Clinical Immunology, 2016, 137, 1178-1188.e7.	2.9	92
138	Cancer Screening Recommendations and Clinical Management of Inherited Gastrointestinal Cancer Syndromes in Childhood. Clinical Cancer Research, 2017, 23, e107-e114.	7.0	91
139	Family History of Cancer and Cancer Risks in Women with BRCA1 or BRCA2 Mutations. Journal of the National Cancer Institute, 2010, 102, 1874-1878.	6.3	89
140	Very frequent loss of heterozygosity throughout chromosome 17 in sporadic ovarian carcinoma. International Journal of Cancer, 1993, 54, 220-225.	5.1	88
141	Impact of germlineBRCA1mutations and overexpression of p53 on prognosis and response to treatment following breast carcinoma. Cancer, 2003, 97, 527-536.	4.1	87
142	Infertility, treatment of infertility, and the risk of breast cancer among women with BRCA1 and BRCA2 mutations: a caseâ€“control study. Cancer Causes and Control, 2008, 19, 1111-1119.	1.8	87
143	In Brief: BRCA1 and BRCA2. Journal of Pathology, 2013, 230, 347-349.	4.5	87
144	Population genetic testing for cancer susceptibility: founder mutations to genomes. Nature Reviews Clinical Oncology, 2016, 13, 41-54.	27.6	86

#	Article	IF	Citations
145	The influence of clinical and genetic factors on patient outcome in small cell carcinoma of the ovary, hypercalcemic type. Gynecologic Oncology, 2016, 141, 454-460.	1.4	85
146	Germ-line BRCA1 mutation is an adverse prognostic factor in Ashkenazi Jewish women with breast cancer. Clinical Cancer Research, 1997, 3, 2465-9.	7.0	85
147	E6/E7 of HPV Type 16 Promotes Cell Invasion and Metastasis of Human Breast Cancer Cells. Cell Cycle, 2007, 6, 2038-2042.	2.6	83
148	HomozygousBUB1BMutation and Susceptibility to Gastrointestinal Neoplasia. New England Journal of Medicine, 2010, 363, 2628-2637.	27.0	82
149	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
150	Clinical practice guidelines for BRCA1 and BRCA2 genetic testing. European Journal of Cancer, 2021, 146, 30-47.	2.8	81
151	Prognostic importance of glomeruloid microvascular proliferation indicates an aggressive angiogenic phenotype in human cancers. Cancer Research, 2002, 62, 6808-11.	0.9	81
152	Oncogenic role of PDK4 in human colon cancer cells. British Journal of Cancer, 2017, 116, 930-936.	6.4	80
153	DICER1 Mutations Are Frequent in Adolescent-Onset Papillary Thyroid Carcinoma. Journal of Clinical Endocrinology and Metabolism, 2018, 103, 2009-2015.	3.6	79
154	Functional mechanisms underlying pleiotropic risk alleles at the 19p13.1 breastâ€“ovarian cancer susceptibility locus. Nature Communications, 2016, 7, 12675.	12.8	78
155	Breast, ovarian, and endometrial malignancies in systemic lupus erythematosus: a meta-analysis. British Journal of Cancer, 2011, 104, 1478-1481.	6.4	77
156	Primary node negative breast cancer in BRCA1 mutation carriers has a poor outcome. Annals of Oncology, 2000, 11, 307-314.	1.2	76
157	Tumor size is an unreliable predictor of prognosis in basal-like breast cancers and does not correlate closely with lymph node status. Breast Cancer Research and Treatment, 2009, 117, 199-204.	2.5	76
158	Ten years of DICER1 mutations: Provenance, distribution, and associated phenotypes. Human Mutation, 2019, 40, 1939-1953.	2.5	76
159	CDK4/6 inhibitors target SMARCA4-determined cyclin D1 deficiency in hypercalcemic small cell carcinoma of the ovary. Nature Communications, 2019, 10, 558.	12.8	76
160	Pancreatic adenocarcinoma: epidemiology and genetics.. Journal of Medical Genetics, 1996, 33, 889-898.	3.2	75
161	Biallelic NTHL1 Mutations in a Woman with Multiple Primary Tumors. New England Journal of Medicine, 2015, 373, 1985-1986.	27.0	75
162	Association of Rare Pathogenic DNA Variants for Familial Hypercholesterolemia, Hereditary Breast and Ovarian Cancer Syndrome, and Lynch Syndrome With Disease Risk in Adults According to Family History. JAMA Network Open, 2020, 3, e203959.	5.9	75

#	Article	IF	Citations
163	Allele loss and mutation screen at the Peutz-Jeghers (LKB1) locus (19p13.3) in sporadic ovarian tumours. British Journal of Cancer, 1999, 80, 70-72.	6.4	74
164	Rare germline mutations inPALB2and breast cancer risk: A population-based study. Human Mutation, 2012, 33, 674-680.	2.5	74
165	Coffee consumption and breast cancer risk amongBRCA1 andBRCA2 mutation carriers. International Journal of Cancer, 2006, 118, 103-107.	5.1	73
166	Risk of ipsilateral breast cancer in BRCA1 and BRCA2 mutation carriers. Breast Cancer Research and Treatment, 2011, 127, 287-296.	2.5	73
167	Age at menarche and the risk of breast cancer in BRCA1 and BRCA2 mutation carriers. Cancer Causes and Control, 2005, 16, 667-674.	1.8	71
168	Compromised BRCA1â€“PALB2 interaction is associated with breast cancer risk. Oncogene, 2017, 36, 4161-4170.	5.9	71
169	Immunohistochemical Expression of DNA Repair Proteins in Familial Breast Cancer DifferentiateBRCA2-Associated Tumors. Journal of Clinical Oncology, 2005, 23, 7503-7511.	1.6	70
170	Breast-Cancer Risk in Families with Mutations in PALB2. New England Journal of Medicine, 2014, 371, 1650-1652.	27.0	68
171	An update on the central nervous system manifestations of DICER1 syndrome. Acta Neuropathologica, 2020, 139, 689-701.	7.7	68
172	High-sensitivity sequencing reveals multi-organ somatic mosaicism causing DICER1 syndrome. Journal of Medical Genetics, 2016, 53, 43-52.	3.2	67
173	Risk of pancreatic cancer among individuals with a family history of cancer of the pancreas. International Journal of Cancer, 2002, 97, 807-810.	5.1	66
174	The APC I1307K allele and breast cancer risk. Nature Genetics, 1998, 20, 13-14.	21.4	65
175	Genetic mechanisms in squamous cell carcinoma of the head and neck. Oral Oncology, 2001, 37, 115-126.	1.5	65
176	Age-specific ovarian cancer risks among women with a BRCA1 or BRCA2 mutation. Gynecologic Oncology, 2018, 150, 85-91.	1.4	65
177	The CDKN2A (p16) gene and human cancer. Molecular Medicine, 1997, 3, 5-20.	4.4	64
178	Diabetes and breast cancer among women with BRCA1 and BRCA2 mutations. Cancer, 2011, 117, 1812-1818.	4.1	62
179	Risk factors for familial and sporadic ovarian cancer among French Canadians: A case-control study. American Journal of Obstetrics and Gynecology, 1998, 179, 403-410.	1.3	61
180	Haplotype analysis of two recurrentCDKN2A mutations in 10 melanoma families: Evidence for common founders and independent mutations. , 1998, 11, 424-431.		61

#	Article	IF	Citations
181	Meta-analysis and pooled analysis of GSTM1 and CYP1A1 polymorphisms and oral and pharyngeal cancers: a HuGE-GSEC review. Genetics in Medicine, 2008, 10, 369-384.	2.4	60
182	miRNA Processing and Human Cancer: DICER1 Cuts the Mustard. Science Translational Medicine, 2011, 3, 111ps46.	12.4	60
183	Familial rhabdoid tumour 'avant la lettre '-from pathology review to exome sequencing and back again. Journal of Pathology, 2013, 231, 35-43.	4.5	60
184	A wholeâ€genome massively parallel sequencing analysis of BRCA1 mutant oestrogen receptorâ€negative and â€positive breast cancers. Journal of Pathology, 2012, 227, 29-41.	4.5	58
185	Pooled genome linkage scan of aggressive prostate cancer: results from the International Consortium for Prostate Cancer Genetics. Human Genetics, 2006, 120, 471-485.	3.8	57
186	The impact of pregnancy on breast cancer survival in women who carry a BRCA1 or BRCA2 mutation. Breast Cancer Research and Treatment, 2013, 142, 177-185.	2.5	57
187	Diagnostic criteria for constitutional mismatch repair deficiency (CMMRD): recommendations from the international consensus working group. Journal of Medical Genetics, 2022, 59, 318-327.	3.2	57
188	Inherited Mutations in Breast Cancer Genesâ€"Risk and Response. Journal of Mammary Gland Biology and Neoplasia, 2011, 16, 3-15.	2.7	56
189	Risk Analysis of Prostate Cancer in PRACTICAL, a Multinational Consortium, Using 25 Known Prostate Cancer Susceptibility Loci. Cancer Epidemiology Biomarkers and Prevention, 2015, 24, 1121-1129.	2.5	56
190	GermlineDICER1mutation and associated loss of heterozygosity in a pineoblastoma: Figure 1. Journal of Medical Genetics, 2012, 49, 417-419.	3.2	55
191	Hormone replacement therapy after menopause and risk of breast cancer in BRCA1 mutation carriers: a caseâ€"control study. Breast Cancer Research and Treatment, 2016, 155, 365-373.	2.5	55
192	Anaplastic sarcomas of the kidney are characterized by DICER1 mutations. Modern Pathology, 2018, 31, 169-178.	5.5	55
193	No association between P53 codon 72 polymorphism and risk of squamous cell carcinoma of the head and neck. British Journal of Cancer, 2000, 82, 757-759.	6.4	54
194	Tumor characteristics as an analytic tool for classifying genetic variants of uncertain clinical significance. Human Mutation, 2008, 29, 1292-1303.	2.5	54
195	Frequency of the CHEK2 1100delC Mutation among Women with Breast Cancer: An International Study. Cancer Research, 2008, 68, 2154-2157.	0.9	54
196	SWI/SNF-Compromised Cancers Are Susceptible to Bromodomain Inhibitors. Cancer Research, 2019, 79, 2761-2774.	0.9	54
197	Small-Cell Carcinoma of the Ovary of Hypercalcemic Type (Malignant Rhabdoid Tumor of the Ovary). Surgical Pathology Clinics, 2016, 9, 215-226.	1.7	53
198	Increased Risk for Nonmedullary Thyroid Cancer in the First Degree Relatives of Prevalent Cases of Nonmedullary Thyroid Cancer: A Hospital-Based Study. Journal of Clinical Endocrinology and Metabolism, 2001, 86, 5307-5312.	3.6	53

#	Article	IF	Citations
199	Loss of SMARCA4 (BRG1) protein expression as determined by immunohistochemistry in smallâ€cell carcinoma of the ovary, hypercalcaemic type distinguishes these tumours from their mimics. Histopathology, 2016, 69, 727-738.	2.9	52
200	Glomeruloid microvascular proliferation is associated with p53 expression, germline BRCA1 mutations and an adverse outcome following breast cancer. British Journal of Cancer, 2003, 89, 1031-1034.	6.4	51
201	Cyclin E expression in breast cancer: predicting germline BRCA1 mutations, prognosis and response to treatment. Annals of Oncology, 2005, 16, 735-742.	1.2	51
202	Hereditary SWI/SNF complex deficiency syndromes. Seminars in Diagnostic Pathology, 2018, 35, 193-198.	1.5	51
203	RAD51C germline mutations in breast and ovarian cancer patients. Breast Cancer Research, 2010, 12, 404.	5.0	50
204	The G84E mutation of HOXB13 is associated with increased risk for prostate cancer: results from the REDUCE trial. Carcinogenesis, 2013, 34, 1260-1264.	2.8	50
205	Prevention and genetic testing for breast cancer: variations in medical decisions. Social Science and Medicine, 2004, 58, 1085-1096.	3.8	49
206	The influence of familial and hereditary factors on the prognosis of breast cancer. Annals of Oncology, 1999, 10, 1163-1170.	1.2	48
207	Family history as a predictor of uptake of cancer preventive procedures by women with a BRCA1 or BRCA2 mutation. Clinical Genetics, 2008, 73, 474-479.	2.0	48
208	Vascular proliferation is increased in basal-like breast cancer. Breast Cancer Research and Treatment, 2011, 130, 1063-1071.	2.5	48
209	Familial medullary thyroid carcinoma and prominent corneal nerves associated with the germline V804M and V778I mutations on the same allele of RET. Journal of Medical Genetics, 2001, 38, 784-787.	3.2	48
210	MSI-low, a real phenomenon which varies in frequency among cancer types. Journal of Pathology, 2003, 201, 389-394.	4.5	47
211	Smoking and the risk of breast cancer among carriers of BRCA mutations. International Journal of Cancer, 2004, 110, 413-416.	5.1	47
212	Oophorectomy after Menopause and the Risk of Breast Cancer in BRCA1 and BRCA2 Mutation Carriers. Cancer Epidemiology Biomarkers and Prevention, 2012, 21, 1089-1096.	2.5	47
213	Candidate DNA repair susceptibility genes identified by exome sequencing in high-risk pancreatic cancer. Cancer Letters, 2016, 370, 302-312.	7.2	47
214	Atypical teratoid/rhabdoid tumors (ATRTs) with SMARCA4 mutation are molecularly distinct from SMARCB1-deficient cases. Acta Neuropathologica, 2021, 141, 291-301.	7.7	47
215	The Basal Phenotype of BRCA1-Related Breast Cancer: Past, Present and Future. Cell Cycle, 2006, 5, 963-967.	2.6	46
216	Hereditary leiomyomatosis and renal cell cancer: an unusual and aggressive form of hereditary renal carcinoma. Nature Clinical Practice Oncology, 2007, 4, 256-261.	4.3	46

#	Article	IF	Citations
217	Ovarian embryonal rhabdomyosarcoma is a rare manifestation of the DICER1 syndrome. Human Pathology, 2015, 46, 917-922.	2.0	46
218	Uterine Tumor Resembling Ovarian Sex Cord Tumor (UTROSCT) Commonly Exhibits Positivity With Sex Cord Markers FOXL2 and SF-1 but Lacks FOXL2 and DICER1 Mutations. International Journal of Gynecological Pathology, 2016, 35, 301-308.	1.4	46
219	Age at first birth and the risk of breast cancer in BRCA1 and BRCA2 mutation carriers. Breast Cancer Research and Treatment, 2007, 105, 221-228.	2.5	45
220	Malignant teratoid tumor of the thyroid gland: an aggressive primitive multiphenotypic malignancy showing organotypical elements and frequent DICER1 alterationsâ€"is the term â€œthyroblastomaâ€•more appropriate?. Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin, 2020, 477, 787-798.	2.8	45
221	Progesterone receptor variant increases ovarian cancer risk in BRCA1 and BRCA2 mutation carriers who were never exposed to oral contraceptives. Pharmacogenetics and Genomics, 2001, 11, 635-638.	5.7	44
222	A Comparison of Bilateral Breast Cancers in BRCA Carriers. Cancer Epidemiology Biomarkers and Prevention, 2005, 14, 1534-1538.	2.5	44
223	Spontaneous and therapeutic abortions and the risk of breast cancer among BRCAmutation carriers. Breast Cancer Research, 2006, 8, R15.	5.0	44
224	Distinct patterns of germ-line deletions inMLH1 andMSH2: the implication of Alu repetitive element in the genetic etiology of Lynch syndrome (HNPCC). Human Mutation, 2006, 27, 388-388.	2.5	44
225	Clinical testing of BRCA1 and BRCA2: a worldwide snapshot of technological practices. Npj Genomic Medicine, 2018, 3, 7.	3.8	44
226	CDKN2A mutation in a non-FAMMM kindred with cancers at multiple sites results in a functionally abnormal protein. , 1997, 73, 531-536.		43
227	The combination of high cyclin E and Skp2 expression in breast cancer is associated with a poor prognosis and the basal phenotype. Human Pathology, 2008, 39, 1431-1437.	2.0	43
228	Mutation analysis of RAD51D in non-BRCA1/2 ovarian and breast cancer families. British Journal of Cancer, 2012, 106, 1460-1463.	6.4	43
229	Gynecologic Manifestations of the DICER1 Syndrome. Surgical Pathology Clinics, 2016, 9, 227-241.	1.7	43
230	Significantly greater prevalence of DICER1 alterations in uterine embryonal rhabdomyosarcoma compared to adenosarcoma. Modern Pathology, 2020, 33, 1207-1219.	5.5	43
231	ALMNASplicing Mutation in Two Sisters with Severe Dunnigan-Type Familial Partial Lipodystrophy Type 2. Journal of Clinical Endocrinology and Metabolism, 2006, 91, 2689-2695.	3.6	42
232	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
233	Embryonal Rhabdomyosarcoma of the Ovary and Fallopian Tube. American Journal of Surgical Pathology, 2020, 44, 738-747.	3.7	42
234	Molecular analyses reveal close similarities between small cell carcinoma of the ovary, hypercalcemic type and atypical teratoid/rhabdoid tumor. Oncotarget, 2016, 7, 1732-1740.	1.8	42

#	Article	IF	Citations
235	Results of a genome-wide linkage analysis in prostate cancer families ascertained through the ACTANE consortium. Prostate, 2003, 57, 270-279.	2.3	41
236	Significant proportion of breast and/or ovarian cancer families of French Canadian descent harbor 1 of 5BRCA1 andBRCA2 mutations. International Journal of Cancer, 2004, 112, 411-419.	5.1	41
237	Locus-specific databases and recommendations to strengthen their contribution to the classification of variants in cancer susceptibility genes. Human Mutation, 2008, 29, 1273-1281.	2.5	41
238	Life insurance: genomic stratification and risk classification. European Journal of Human Genetics, 2014, 22, 575-579.	2.8	41
239	Ovarian Microcystic Stromal Tumors Are Characterized by Alterations in the Beta-Catenin-APC Pathway and May be an Extracolonic Manifestation of Familial Adenomatous Polyposis. American Journal of Surgical Pathology, 2018, 42, 137-139.	3.7	41
240	FounderBRCA1 andBRCA2 mutations in early-onset French Canadian breast cancer cases unselected for family history. International Journal of Cancer, 2001, 95, 189-193.	5.1	40
241	Germline truncating mutations in both MSH2 and BRCA2 in a single kindred. British Journal of Cancer, 2004, 90, 483-491.	6.4	40
242	The contribution of founder mutations to earlyâ€onset breast cancer in Frenchâ€Canadian women. Clinical Genetics, 2009, 76, 421-426.	2.0	40
243	Identification of gene fusion transcripts by transcriptome sequencing in BRCA1-mutated breast cancers and cell lines. BMC Medical Genomics, 2011, 4, 75.	1.5	40
244	Serum levels of mature microRNAs in DICER1-mutated pleuropulmonary blastoma. Oncogenesis, 2014, 3, e87-e87.	4.9	40
245	Loss of human ICOSL results in combined immunodeficiency. Journal of Experimental Medicine, 2018, 215, 3151-3164.	8.5	40
246	DICER1-associated central nervous system sarcoma in children: comprehensive clinicopathologic and genetic analysis of a newly described rare tumor. Modern Pathology, 2020, 33, 1910-1921.	5.5	40
247	Survival Benefit for Individuals With Constitutional Mismatch Repair Deficiency Undergoing Surveillance. Journal of Clinical Oncology, 2021, 39, 2779-2790.	1.6	40
248	A tale of four syndromes: familial adenomatous polyposis, Gardner syndrome, attenuated APC and Turcot syndrome. QJM - Monthly Journal of the Association of Physicians, 1995, 88, 853-63.	0.5	40
249	Ovarian Sex Cord-Stromal Tumors in Patients With Probable or Confirmed Germline DICER1 Mutations. International Journal of Gynecological Pathology, 2015, 34, 266-274.	1.4	39
250	Homologous recombination DNA repair defects in PALB2-associated breast cancers. Npj Breast Cancer, 2019, 5, 23.	5.2	39
251	Mesenchymal Hamartoma of the Liver and DICER1 Syndrome. New England Journal of Medicine, 2019, 380, 1834-1842.	27.0	39
252	Allele loss on chromosome 17q in sporadic ovarian cancer. Lancet, The, 1991, 338, 444-445.	13.7	38

#	Article	IF	Citations
253	Loss of heterozygosity on chromosome 22 in ovarian carcinoma is distal to and is not accompanied by mutations in NF2 at 22q12. British Journal of Cancer, 1994, 70, 905-907.	6.4	38
254	The risk of breast cancer in women with a BRCA1 mutation from North America and Poland. International Journal of Cancer, 2012, 131, 229-234.	5.1	38
255	Co-amplification of CCND1 and EMSY is associated with an adverse outcome in ER-positive tamoxifen-treated breast cancers. Breast Cancer Research and Treatment, 2010, 121, 347-354.	2.5	37
256	Analysis of the gene coding for the BRCA2â€‐interacting protein PALB2 in hereditary prostate cancer. Prostate, 2008, 68, 675-678.	2.3	36
257	Clinical, morphological and immunohistochemical evidence that smallâ€cell carcinoma of the ovary of hypercalcaemic type (<scp>SCCOHT</scp>) may be a primitive germâ€cell neoplasm. Histopathology, 2017, 70, 1147-1154.	2.9	36
258	Germ-line deletion in DICER1 revealed by a novel MLPA assay using synthetic oligonucleotides. European Journal of Human Genetics, 2014, 22, 564-567.	2.8	35
259	Duration of tamoxifen use and the risk of contralateral breast cancer in BRCA1 and BRCA2 mutation carriers. Breast Cancer Research and Treatment, 2014, 146, 421-427.	2.5	35
260	Epithelial inactivation of Yy1 abrogates lung branching morphogenesis. Development (Cambridge), 2015, 142, 2981-2995.	2.5	35
261	MDM2 overexpression is rare in ovarian carcinoma irrespective of TP53 mutation status. British Journal of Cancer, 1995, 72, 883-888.	6.4	34
262	Triple-Negative and Basal-Like Breast Cancer: Implications for Oncologists. Current Oncology, 2011, 18, 161-164.	2.2	34
263	Rare non-epithelial ovarian neoplasms: Pathology, genetics and treatment. Gynecologic Oncology, 2016, 142, 190-198.	1.4	34
264	Functionally Null RAD51D Missense Mutation Associates Strongly with Ovarian Carcinoma. Cancer Research, 2017, 77, 4517-4529.	0.9	34
265	Imaging of DICER1 syndrome. Pediatric Radiology, 2019, 49, 1488-1505.	2.0	34
266	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
267	Cyclin D1 is essential for neoplastic transformation induced by both E6/E7 and E6/E7/ErbB-2 cooperation in normal cells. Oncogene, 2004, 23, 5252-5256.	5.9	33
268	Germline TP53 mutations in BRCA1 and BRCA2 mutation-negative French Canadian breast cancer families. Breast Cancer Research and Treatment, 2008, 108, 399-408.	2.5	33
269	Vascular proliferation is a prognostic factor in breast cancer. Breast Cancer Research and Treatment, 2012, 133, 501-510.	2.5	33
270	DICER1mutations in an adolescent with cervical embryonal rhabdomyosarcoma (cERMS). Pediatric Blood and Cancer, 2014, 61, 568-569.	1.5	33

#	Article	IF	Citations
271	Risk of breast cancer after a diagnosis of ovarian cancer in BRCA mutation carriers: Is preventive mastectomy warranted?. Gynecologic Oncology, 2017, 145, 346-351.	1.4	33
272	Multiple DICER1â€related tumors in a child with a large interstitial 14q32 deletion. Genes Chromosomes and Cancer, 2018, 57, 223-230.	2.8	33
273	Detection of Pathogenic Variants With Germline Genetic Testing Using Deep Learning vs Standard Methods in Patients With Prostate Cancer and Melanoma. JAMA - Journal of the American Medical Association, 2020, 324, 1957.	7.4	33
274	Mutation analysis of the tumor suppressorPTEN and the glypican 3 (GPC3) gene in patients diagnosed with Proteus syndrome. American Journal of Medical Genetics Part A, 2004, 130A, 123-127.	2.4	32
275	Evolution of Renal Cysts to Anaplastic Sarcoma of Kidney in a Child With DICER1 Syndrome. Pediatric Blood and Cancer, 2016, 63, 1272-1275.	1.5	32
276	Compelling evidence for a prostate cancer gene at 22q12.3 by the International Consortium for Prostate Cancer Genetics. Human Molecular Genetics, 2007, 16, 1271-1278.	2.9	31
277	BRCA1 and BRCA2 families and the risk of skin cancer. Familial Cancer, 2010, 9, 489-493.	1.9	31
278	Should all BRCA1 mutation carriers with stage I breast cancer receive chemotherapy?. Breast Cancer Research and Treatment, 2013, 138, 273-279.	2.5	31
279	Tumor progression in DICER1-mutated cystic nephromaâ€"witnessing the genesis of anaplastic sarcoma of the kidney. Human Pathology, 2016, 53, 114-120.	2.0	31
280	DGCR8 microprocessor defect characterizes familial multinodular goiter with schwannomatosis. Journal of Clinical Investigation, 2020, 130, 1479-1490.	8.2	31
281	Loss of heterozygosity on chromosome 5 in sporadic ovarian carcinoma is a late event and is not associated with mutations in APC at 5q21-22. Human Mutation, 1994, 3, 283-291.	2.5	30
282	A rapid fluorescent multiplexed-PCR analysis (FMPA) for founder mutations in the BRCA1 and BRCA2 genes. Clinical Genetics, 2001, 57, 213-220.	2.0	30
283	Risk of non-medullary thyroid cancer influenced by polymorphic variation in the thyroglobulin gene. Carcinogenesis, 2003, 25, 369-373.	2.8	30
284	Is the E133K allele of VG5Q associated with Klippel-Trenaunay and other overgrowth syndromes?. Journal of Medical Genetics, 2005, 43, 613-614.	3.2	30
285	Expression of epidermal growth factor receptor in relation to BRCA1 status, basal-like markers and prognosis in breast cancer. Journal of Clinical Pathology, 2009, 62, 139-146.	2.0	30
286	A case of Cowden's syndrome presenting with gastric carcinomas and gastrointestinal polyposis. Nature Reviews Gastroenterology and Hepatology, 2009, 6, 184-189.	17.8	30
287	Functional redundancy of exon 12 ofBRCA2revealed by a comprehensive analysis of the c.6853A>G (p.I2285V) variant. Human Mutation, 2009, 30, 1543-1550.	2.5	30
288	Multinodular Goiter in Children: An Important Pointer to a Germline DICER1 Mutation. Journal of Clinical Endocrinology and Metabolism, 2014, 99, 1947-1948.	3.6	30

#	Article	IF	Citations
289	A homozygousPMS2founder mutation with an attenuated constitutional mismatch repair deficiency phenotype. Journal of Medical Genetics, 2015, 52, 348-352.	3.2	30
290	Deep Sequencing Reveals Spatially Distributed Distinct Hot Spot Mutations in DICER1-Related Multinodular Goiter. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 3637-3645.	3.6	30
291	The McGill Interactive Pediatric OncoGenetic Guidelines: An approach to identifying pediatric oncology patients most likely to benefit from a genetic evaluation. Pediatric Blood and Cancer, 2017, 64, e26441.	1.5	30
292	Sequencing of DICER1 in sarcomas identifies biallelic somatic DICER1 mutations in an adult-onset embryonal rhabdomyosarcoma. British Journal of Cancer, 2017, 116, 1621-1626.	6.4	30
293	A sensitive and scalable microsatellite instability assay to diagnose constitutional mismatch repair deficiency by sequencing of peripheral blood leukocytes. Human Mutation, 2019, 40, 649-655.	2.5	30
294	Fortnightly review: Hereditary ovarian carcinoma. BMJ: British Medical Journal, 1999, 318, 786-789.	2.3	29
295	The polymorphic CAG repeat of the androgen receptor gene: a potential role in breast cancer in women over 40. Breast Cancer Research and Treatment, 2001, 70, 109-116.	2.5	29
296	p53 â€" Master and Commander. New England Journal of Medicine, 2007, 357, 2539-2541.	27.0	29
297	Germ-line and somaticDICER1mutations in a pleuropulmonary blastoma. Pediatric Blood and Cancer, 2013, 60, 2091-2092.	1.5	29
298	DICER1 gene mutations in endocrine tumors. Endocrine-Related Cancer, 2018, 25, R197-R208.	3.1	29
299	<scp>DICER1â€associated</scp> embryonal rhabdomyosarcoma and adenosarcoma of the gynecologic tract: Pathology, molecular genetics, and indications for molecular testing. Genes Chromosomes and Cancer, 2021, 60, 217-233.	2.8	29
300	A Comprehensive Analysis of MNG1, TCO1, fPTC, PTEN, TSHR, and TRKA in Familial Nonmedullary Thyroid Cancer: Confirmation of Linkage to TCO1. Journal of Clinical Endocrinology and Metabolism, 2001, 86, 3701-3704.	3.6	29
301	Genetic implications of double primary cancers of the colorectum and endometrium.. Journal of Medical Genetics, 1998, 35, 978-984.	3.2	28
302	Tamoxifen May Be an Effective Adjuvant Treatment for BRCA1-Related Breast Cancer Irrespective of Estrogen Receptor Status. Journal of the National Cancer Institute, 2002, 94, 1504-1506.	6.3	28
303	Polymorphisms and HNPCC: PMS2-MLH1 protein interactions diminished by single nucleotide polymorphisms. Human Mutation, 2002, 19, 108-113.	2.5	28
304	Challenges to cancer control by screening. Nature Reviews Cancer, 2003, 3, 297-303.	28.4	28
305	Clinical and genetic findings in an Ashkenazi Jewish population with colorectal neoplasms. Cancer, 2005, 104, 719-729.	4.1	28
306	Germline PALB2 mutation analysis in breast-pancreas cancer families. Journal of Medical Genetics, 2011, 48, 523-525.	3.2	28

#	Article	IF	Citations
307	Prophylactic oophorectomy for hereditary small cell carcinoma of the ovary, hypercalcemic type. Gynecologic Oncology Reports, 2015, 12, 20-22.	0.6	28
308	Germline and Somatic DICER1 Mutations in a Well-Differentiated Fetal Adenocarcinoma of the Lung. Journal of Thoracic Oncology, 2016, 11, e31-e33.	1.1	28
309	The FANCM:p.Arg658* truncating variant is associated with risk of triple-negative breast cancer. Npj Breast Cancer, 2019, 5, 38.	5.2	28
310	Suggestive evidence for a site specific prostate cancer gene on chromosome 1p36. Journal of Medical Genetics, 2000, 37, 947-949.	3.2	28
311	The Role of Genetic Factors in the Etiology of Pancreatic Adenocarcinoma: An Update. Cancer Investigation, 2001, 19, 65-75.	1.3	27
312	Haplotype Analysis of BRCA2 8765delAG Mutation Carriers in French Canadian and Yemenite Jewish Hereditary Breast Cancer Families. Human Heredity, 2001, 52, 116-120.	0.8	27
313	Relationship Between Angiotensin-Converting Enzyme Gene Polymorphism and Body Composition, Functional Performance, and Blood Biomarkers in Advanced Cancer Patients. Clinical Cancer Research, 2009, 15, 2442-2447.	7.0	27
314	Smoking and the risk of breast cancer in BRCA1 and BRCA2 carriers: an update. Breast Cancer Research and Treatment, 2009, 114, 127-135.	2.5	27
315	Characterization of two Ashkenazi Jewish founder mutations in MSH6 gene causing Lynch syndrome. Clinical Genetics, 2011, 79, 512-522.	2.0	27
316	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
317	Data sharing as a national quality improvement program: reporting on BRCA1 and BRCA2 variant-interpretation comparisons through the Canadian Open Genetics Repository (COGR). Genetics in Medicine, 2018, 20, 294-302.	2.4	27
318	A636P is Associated with Early-Onset Colon Cancer in Ashkenazi Jews. Journal of the American College of Surgeons, 2003, 196, 222-225.	0.5	26
319	Disruption of an exon splicing enhancer in exon 3 of MLH1 is the cause of HNPCC in a Quebec family. Journal of Medical Genetics, 2005, 43, 153-156.	3.2	26
320	Application of BRCA1 and BRCA2 mutation carrier prediction models in breast and/or ovarian cancer families of French Canadian descent. Clinical Genetics, 2006, 70, 320-329.	2.0	26
321	Exploring the endocrine manifestations of DICER1 mutations. Trends in Molecular Medicine, 2012, 18, 503-505.	6.7	26
322	A founder BRCA2 mutation in nonâ€Afrikaner breast cancer patients of the Western Cape of South Africa. Clinical Genetics, 2012, 81, 179-184.	2.0	26
323	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
324	BRCA1andBRCA2pathogenic sequence variants in women of African origin or ancestry. Human Mutation, 2019, 40, 1781-1796.	2.5	26

#	Article	IF	Citations
325	The genetic landscape of choroid plexus tumors in children and adults. Neuro-Oncology, 2021, 23, 650-660.	1.2	26
326	Prevalence and Spectrum of DICER1 Mutations in Adult-onset Thyroid Nodules with Indeterminate Cytology. Journal of Clinical Endocrinology and Metabolism, 2021, 106, e968-e977.	3.6	26
327	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
328	Physical activity during adolescence and young adulthood and the risk of breast cancer in BRCA1 and BRCA2 mutation carriers. Breast Cancer Research and Treatment, 2018, 169, 561-571.	2.5	25
329	eIF4A Inhibitors Suppress Cell-Cycle Feedback Response and Acquired Resistance to CDK4/6 Inhibition in Cancer. Molecular Cancer Therapeutics, 2019, 18, 2158-2170.	4.1	25
330	Oestrogen receptor status and survival in women with BRCA2-associated breast cancer. British Journal of Cancer, 2019, 120, 398-403.	6.4	25
331	Influenza A and rhabdomyolysis. Journal of Infection, 1990, 21, 303-304.	3.3	24
332	A missense mutation in both hMSH2 andAPC in an Ashkenazi Jewish HNPCC kindred: implications for clinical screening. Journal of Medical Genetics, 1999, 36, 792-793.	3.2	24
333	Synchronous occult cancers of the endometrium and fallopian tube in an MSH2 mutation carrier at time of prophylactic surgery. Gynecologic Oncology, 2008, 111, 575-578.	1.4	24
334	Alcohol consumption and the risk of breast cancer among BRCA1 and BRCA2 mutation carriers. Breast, 2010, 19, 479-483.	2.2	24
335	Qualitative thematic analysis of consent forms used in cancer genome sequencing. BMC Medical Ethics, 2011, 12, 14.	2.4	24
336	Association analysis of 9,560 prostate cancer cases from the International Consortium of Prostate Cancer Genetics confirms the role of reported prostate cancer associated SNPs for familial disease. Human Genetics, 2014, 133, 347-356.	3.8	24
337	Mammography screening and the risk of breast cancer in BRCA1 and BRCA2 mutation carriers: a prospective study. Breast Cancer Research and Treatment, 2014, 147, 113-118.	2.5	24
338	Expanding the morphological spectrum of ovarian microcystic stromal tumour. Histopathology, 2019, 74, 443-451.	2.9	24
339	Founder BRCA1/BRCA2/PALB2 pathogenic variants in French-Canadian breast cancer cases and controls. Scientific Reports, 2020, 10, 6491.	3.3	24
340	False family history of breast cancer in the family cancer clinic. European Journal of Surgical Oncology, 1998, 24, 275-279.	1.0	23
341	Constitutional alterations of the ATM gene in early onset sporadic breast cancer. Journal of Medical Genetics, 2002, 39, 751-753.	3.2	23
342	Adult-Onset Cervical Embryonal Rhabdomyosarcoma and DICER1 Mutations. Journal of Lower Genital Tract Disease, 2016, 20, e8-e10.	1.9	23

#	Article	IF	Citations
343	Functional Repair Assay for the Diagnosis of Constitutional Mismatch Repair Deficiency From Non-Neoplastic Tissue. Journal of Clinical Oncology, 2019, 37, 461-470.	1.6	23
344	Clinical Outcomes and Complications of Pituitary Blastoma. Journal of Clinical Endocrinology and Metabolism, 2021, 106, 351-363.	3.6	23
345	Genetic counseling and interpretation of genetic tests in familial adenomatous polyposis and hereditary nonpolyposis colorectal cancer. Diseases of the Colon and Rectum, 2001, 44, 271-279.	1.3	22
346	Genomeâ€wide linkage analysis of 1,233 prostate cancer pedigrees from the International Consortium for prostate cancer Genetics using novel sumLINK and sumLOD analyses. Prostate, 2010, 70, 735-744.	2.3	22
347	Radiographic Screening of Infants and Young Children With Genetic Predisposition for Rare Malignancies: DICER1 Mutations and Pleuropulmonary Blastoma. American Journal of Roentgenology, 2015, 204, W475-W482.	2.2	22
348	Somatic DICER1 mutations in adult-onset pulmonary blastoma. European Respiratory Journal, 2016, 47, 1879-1882.	6.7	22
349	Functional characterization of multiple DICER1 mutations in an adolescent. Endocrine-Related Cancer, 2016, 23, L1-L5.	3.1	22
350	The hereditary nature of small cell carcinoma of the ovary, hypercalcemic type: two new familial cases. Familial Cancer, 2017, 16, 395-399.	1.9	22
351	Clinical and Molecular Characteristics May Alter Treatment Strategies of Thyroid Malignancies in DICER1 Syndrome. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 277-284.	3.6	22
352	An eHealth decisionâ€support tool to prioritize referral practices for genetic evaluation of patients with Wilms tumor. International Journal of Cancer, 2020, 146, 1010-1017.	5.1	22
353	Macrofollicular Variant of Follicular Thyroid Carcinoma: A Rare Underappreciated Pitfall in the Diagnosis of Thyroid Carcinoma. Thyroid, 2020, 30, 72-80.	4.5	22
354	DICER1-associated sarcomas: towards a unified nomenclature. Modern Pathology, 2021, 34, 1226-1228.	5.5	22
355	Performance of the McGill Interactive Pediatric OncoGenetic Guidelines for Identifying Cancer Predisposition Syndromes. JAMA Oncology, 2021, 7, 1806.	7.1	22
356	The impact of oophorectomy on survival after breast cancer in BRCA1 and BRCA2 mutation carriers.. Journal of Clinical Oncology, 2014, 32, 1507-1507.	1.6	22
357	Increased risk of head and neck cancer in association withGSTT1 nullizygosity for individuals with low exposure to tobacco. International Journal of Cancer, 2000, 87, 452-454.	5.1	21
358	Macrophage Scavenger Receptor 1 999C>T (R293X) Mutation and Risk of Prostate Cancer. Cancer Epidemiology Biomarkers and Prevention, 2005, 14, 397-402.	2.5	21
359	How old is this mutation? - a study of three Ashkenazi Jewish founder mutations. BMC Genetics, 2010, 11, 39.	2.7	21
360	Melphalan as a treatment for BRCA-related ovarian carcinoma: can you teach an old drug new tricks?: Figure 1. Journal of Clinical Pathology, 2011, 64, 924-926.	2.0	21

#	Article	IF	Citations
361	Validation of prostate cancer risk-related loci identified from genome-wide association studies using family-based association analysis: evidence from the International Consortium for Prostate Cancer Genetics (ICPCG). Human Genetics, 2012, 131, 1095-1103.	3.8	21
362	In Brief: Picturing the complex world of chromatin remodelling families. Journal of Pathology, 2015, 237, 403-406.	4.5	21
363	Breast cancer in systemic lupus. Lupus, 2017, 26, 311-315.	1.6	21
364	Retrospective evaluation of a decisionâ€support algorithm (MIPOGG) for genetic referrals for children with neuroblastic tumors. Pediatric Blood and Cancer, 2018, 65, e27390.	1.5	21
365	NovelPOLEpathogenic germline variant in a family with multiple primary tumors results in distinct mutational signatures. Human Mutation, 2019, 40, 36-41.	2.5	21
366	Embryonal rhabdomyosarcoma of the uterine corpus: a clinicopathological and molecular analysis of 21 cases highlighting a frequent association with DICER1 mutations. Modern Pathology, 2021, 34, 1750-1762.	5.5	21
367	High risk genes predisposing to prostate cancer developmentâ€"do they exist?. Prostate Cancer and Prostatic Diseases, 2000, 3, 241-247.	3.9	20
368	The HNPCC associated MSH2*1906G->C founder mutation probably originated between 1440 CE and 1715 CE in the Ashkenazi Jewish population. Journal of Medical Genetics, 2005, 42, 766-768.	3.2	20
369	BRCA2: breaks, mistakes and failed separations. Trends in Molecular Medicine, 2005, 11, 145-148.	6.7	20
370	Haplotype analysis suggest common founders in carriers of the recurrent BRCA2mutation, 3398delAAAAG, in French Canadian hereditary breast and/ovarian cancer families. BMC Medical Genetics, 2006, 7, 23.	2.1	20
371	BRCA1 mutations contribute to cell motility and invasion by affecting its main regulators. Cell Cycle, 2008, 7, 3781-3783.	2.6	20
372	Neonatal Gardner Fibroma: A Sentinel Presentation of Severe Familial Adenomatous Polyposis. Pediatrics, 2010, 126, e1599-e1602.	2.1	20
373	The Effect of a DNA Repair Gene on Cellular Invasiveness: Xrcc3 Over-Expression in Breast Cancer Cells. PLoS ONE, 2011, 6, e16394.	2.5	20
374	Germline TP53 mutational spectrum in French Canadians with breast cancer. BMC Medical Genetics, 2015, 16, 24.	2.1	20
375	Familial multinodular goiter and Sertoli-Leydig cell tumors associated with a large intragenic in-frame DICER1 deletion. European Journal of Endocrinology, 2018, 178, K11-K19.	3.7	20
376	A Preclinical Trial and Molecularly Annotated Patient Cohort Identify Predictive Biomarkers in Homologous Recombinationâ€"deficient Pancreatic Cancer. Clinical Cancer Research, 2020, 26, 5462-5476.	7.0	20
377	SMARCA4/2 loss inhibits chemotherapy-induced apoptosis by restricting IP3R3-mediated Ca2+ flux to mitochondria. Nature Communications, 2021, 12, 5404.	12.8	20
378	Penetrance of Mutations in the Familial Wilms Tumor Gene FWT1. Journal of the National Cancer Institute, 2000, 92, 650-652.	6.3	19

#	Article	IF	Citations
379	Identification and characterization of novel SNPs in CHEK2 in Ashkenazi Jewish men with prostate cancer. Cancer Letters, 2008, 270, 173-180.	7.2	19
380	Eleven years disease-free: role of chemotherapy in metastatic BRCA2-related breast cancer. Nature Reviews Clinical Oncology, 2009, 6, 488-492.	27.6	19
381	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
382	Cultural Aspects of Healthy BRCA Carriers From Two Ethnocultural Groups. Qualitative Health Research, 2014, 24, 665-681.	2.1	19
383	Expression of Nestin associates with BRCA1 mutations, a basal-like phenotype and aggressive breast cancer. Scientific Reports, 2017, 7, 1089.	3.3	19
384	Cognitive markers of dementia risk in middle-aged women with bilateral salpingo-oophorectomy prior to menopause. Neurobiology of Aging, 2020, 94, 1-6.	3.1	19
385	The ten genes for breast (and ovarian) cancer susceptibility. Nature Reviews Clinical Oncology, 2021, 18, 259-260.	27.6	19
386	Paediatric ovarian tumours and their associated cancer susceptibility syndromes. Journal of Medical Genetics, 2018, 55, 1-10.	3.2	19
387	Sonographic Features of Breast Carcinoma Presenting as Masses inBRCAGene Mutation Carriers. Journal of Ultrasound in Medicine, 2007, 26, 817-824.	1.7	18
388	Many Mosaic Mutations. Current Oncology, 2013, 20, 85-87.	2.2	18
389	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
390	Reflex Testing for Germline BRCA1, BRCA2, PALB2, and ATM Mutations in Pancreatic Cancer: Mutation Prevalence and Clinical Outcomes From Two Canadian Research Registries. JCO Precision Oncology, 2018, 2, 1-16.	3.0	18
391	Stathmin expression associates with vascular and immune responses in aggressive breast cancer subgroups. Scientific Reports, 2020, 10, 2914.	3.3	18
392	Excess of congenital abnormalities in French-Canadian children with neuroblastoma: A case series study from Montreal. , 1997, 29, 272-279.		17
393	Role of molecular diagnostic testing in familial adenomatous polyposis and hereditary nonpolyposis colorectal cancer families. Diseases of the Colon and Rectum, 2001, 44, 437-446.	1.3	17
394	Tumor Buds Show Reduced Expression of Laminin-5 Gamma 2 Chain in DNA Mismatch Repair Deficient Colorectal Cancer. Diseases of the Colon and Rectum, 2006, 49, 1193-1202.	1.3	17
395	<scp>BRCA1</scp> and <scp>BRCA2</scp>Ââ€“Âupdate and implications on the genetics of breast cancer: a clinical perspective. Clinical Genetics, 2014, 85, 1-4.	2.0	17
396	Recently characterized molecular events in uncommon gynaecological neoplasms and their clinical importance. Histopathology, 2016, 69, 903-913.	2.9	17

#	Article	IF	Citations
397	Variation in rates of uptake of preventive options by Canadian women carrying the BRCA1 or BRCA2 genetic mutation. Open Medicine, 2007, 1, e92-8.	1.5	17
398	Rhabdomyolysis after intramuscular iron-dextran in malabsorption.. Annals of the Rheumatic Diseases, 1991, 50, 184-186.	0.9	16
399	Cutaneous malignant melanoma in women is uncommonly associated with a family history of melanoma in first-degree relatives: a case-control study. Melanoma Research, 1996, 6, 435-440.	1.2	16
400	Haplotype analysis of aBRCA1: 185delAG mutation in a Chilean family supports its Ashkenazi origins. Clinical Genetics, 2002, 62, 151-156.	2.0	16
401	Fallopian tube cancer in a BRCA1 mutation carrier: Rapid development and failure of screening. American Journal of Obstetrics and Gynecology, 2002, 186, 53-54.	1.3	16
402	Identification of a novel CHEK2variant and assessment of its contribution to the risk of breast cancer in French Canadian women. BMC Cancer, 2008, 8, 239.	2.6	16
403	Size Surprise? Tumour Size, Nodal Status, and Outcome after Breast Cancer. Current Oncology, 2012, 19, 241-243.	2.2	16
404	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
405	Psychosocial Impact of Lynch Syndrome on Affected Individuals and Families. Digestive Diseases and Sciences, 2015, 60, 2246-2250.	2.3	16
406	Biallelic somaticSMARCA4mutations in small cell carcinoma of the ovary, hypercalcemic type (SCCOHT). Pediatric Blood and Cancer, 2015, 62, 728-730.	1.5	16
407	Infantile Pulmonary Teratoid Tumor. New England Journal of Medicine, 2018, 378, 2238-2240.	27.0	16
408	Is hereditary site-specific ovarian cancer a distinct genetic condition?. American Journal of Medical Genetics Part A, 1998, 75, 55-58.	2.4	15
409	Survival of patients with breast cancer and BRCA1 mutations. Lancet, The, 1998, 351, 1359.	13.7	15
410	The Importance of a Family History of Breast Cancer in Predicting the Presence of a BRCA Mutation. American Journal of Human Genetics, 1999, 65, 1776-1778.	6.2	15
411	The frequent BRCA1 mutation 1135insA has multiple origins: a haplotype study in different populations. BMC Medical Genetics, 2006, 7, 15.	2.1	15
412	Exome Sequencing in BRCA1- and BRCA2-Negative Greek Families Identifies MDM1 and NBEAL1 as Candidate Risk Genes for Hereditary Breast Cancer. Frontiers in Genetics, 2019, 10, 1005.	2.3	15
413	BRCA testing in women with high-grade serous ovarian cancer: gynecologic oncologist-initiated testing compared with genetics referral. International Journal of Gynecological Cancer, 2020, 30, 1757-1761.	2.5	15
414	Pineoblastoma is uniquely tolerant of mutually exclusive loss of DICER1, DROSHA or DGCR8. Acta Neuropathologica, 2020, 139, 1115-1118.	7.7	15

#	Article	IF	Citations
415	The contribution of inherited factors to the clinicopathological features and behavior of breast cancer. Journal of Mammary Gland Biology and Neoplasia, 2001, 6, 453-465.	2.7	14
416	Transforming growth factor-beta pathway disruption and infiltration of colorectal cancers by intraepithelial lymphocytes. Histopathology, 2006, 49, 371-380.	2.9	14
417	High frequency of exon deletions and putative founder effects in French Canadian Lynch syndrome families. Human Mutation, 2009, 30, E797-E812.	2.5	14
418	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.	2.5	14
419	Chromosomes 4 and 8 implicated in a genome wide SNP linkage scan of 762 prostate cancer families collected by the ICPCG. Prostate, 2012, 72, 410-426.	2.3	14
420	Exome profiling of primary, metastatic and recurrent ovarian carcinomas in a BRCA1-positive patient. BMC Cancer, 2013, 13, 146.	2.6	14
421	Preventing Ovarian Cancer by Salpingectomy. Current Oncology, 2013, 20, 139-142.	2.2	14
422	Data sharing to improve concordance in variant interpretation across laboratories: results from the Canadian Open Genetics Repository. Journal of Medical Genetics, 2022, 59, 571-578.	3.2	14
423	The risk of breast cancer in BRCA1 and BRCA2 mutation carriers without a firstâ€degree relative with breast cancer. Clinical Genetics, 2018, 93, 1063-1068.	2.0	14
424	DICER1-sarcoma: an emerging entity. Modern Pathology, 2021, 34, 2096-2097.	5.5	14
425	Prevalence of founder BRCA1 and BRCA2 mutations in unselected French Canadian women with breast cancer. Clinical Genetics, 2001, 59, 418-423.	2.0	13
426	Double primary cancers of the breast and thyroid in women: molecular analysis and genetic implications. Familial Cancer, 2001, 1, 17-24.	1.9	13
427	Double frameshift mutations in APC and MSH2 in the same individual. International Journal of Colorectal Disease, 2005, 20, 466-470.	2.2	13
428	Clinically Relevant Biology of Hereditary Breast Cancer. Seminars in Oncology, 2007, 34, 379-383.	2.2	13
429	Therapeutic implications of Src independent calcium mobilization in diffuse large B-cell lymphoma. Leukemia Research, 2010, 34, 585-593.	0.8	13
430	Germ-line DICER1 mutations do not make a major contribution to the etiology of familial testicular germ cell tumours. BMC Research Notes, 2013, 6, 127.	1.4	13
431	Hi-Plex for high-throughput mutation screening: application to the breast cancer susceptibility gene PALB2. BMC Medical Genomics, 2013, 6, 48.	1.5	13
432	Characterization of a novel founder <scp>MSH6</scp> mutation causing Lynch syndrome in the French Canadian population. Clinical Genetics, 2015, 87, 536-542.	2.0	13

#	Article	IF	Citations
433	Sarcoma and germ-line DICER1 mutations. Lancet Oncology, The, 2016, 17, e470.	10.7	13
434	Low yield of gastroscopy in patients with Lynch syndrome. Turkish Journal of Gastroenterology, 2017, 28, 434-438.	1.1	13
435	Increased incidence of cancer in first degree relatives of women with double primary carcinomas of the breast and colon.. Journal of Medical Genetics, 1996, 33, 534-539.	3.2	12
436	Variation in prophylactic surgery decisions. Lancet, The, 2000, 356, 1687.	13.7	12
437	An MLH1 haplotype is over-represented on chromosomes carrying an HNPCC predisposing mutation in MLH1. Journal of Medical Genetics, 2002, 39, 323-327.	3.2	12
438	Is there a correlation between the structure of hair and breast cancer or BRCA1/2 mutations?. Physics in Medicine and Biology, 2002, 47, 1623-1632.	3.0	12
439	Validation study of the <scp>lambda</scp> model for predicting the BRCA1 or BRCA2 mutation carrier status of North American Ashkenazi Jewish women. Clinical Genetics, 2007, 72, 87-97.	2.0	12
440	BRCA1â€"sowing the seeds crooked in the furrow. Nature Genetics, 2008, 40, 8-9.	21.4	12
441	Using a Family History Questionnaire to Identify Adult Patients with Increased Genetic Risk for Sarcoma. Current Oncology, 2015, 22, 317-325.	2.2	12
442	Prospective evaluation of alcohol consumption and the risk of breast cancer in BRCA1 and BRCA2 mutation carriers. Breast Cancer Research and Treatment, 2015, 151, 435-441.	2.5	12
443	Establishing a Clinic-Based Pancreatic Cancer and Periampullary Tumour Research Registry in Quebec. Current Oncology, 2015, 22, 113-121.	2.2	12
444	Prostate-specific antigen velocity in a prospective prostate cancer screening study of men with genetic predisposition. British Journal of Cancer, 2018, 118, 266-276.	6.4	12
445	The dilemma of early preventive oophorectomy in familial small cell carcinoma of the ovary of hypercalcemic type. Gynecologic Oncology Reports, 2019, 28, 47-49.	0.6	12
446	Oophorectomy and risk of contralateral breast cancer among BRCA1 and BRCA2 mutation carriers. Breast Cancer Research and Treatment, 2019, 175, 443-449.	2.5	12
447	Etiologic Index â€" A Case-Only Measure of BRCA1/2â€"Associated Cancer Risk. New England Journal of Medicine, 2020, 383, 286-288.	27.0	12
448	Evaluation of molecular analysis in challenging ovarian sex cord-stromal tumours: a review of 50 cases. Pathology, 2020, 52, 686-693.	0.6	12
449	The contribution of large genomic rearrangements in BRCA1 and BRCA2 to South African familial breast cancer. BMC Cancer, 2020, 20, 391.	2.6	12
450	Oncology clinic-based germline genetic testing for exocrine pancreatic cancer enables timely return of results and unveils low uptake of cascade testing. Journal of Medical Genetics, 2022, 59, 793-800.	3.2	12

#	Article	IF	Citations
451	The Value of DICER1 Mutation Analysis in â€œSubtleâ€•Diagnostically Challenging Embryonal Rhabdomyosarcomas of the Uterine Cervix. International Journal of Gynecological Pathology, 2021, 40, 435-440.	1.4	12
452	A functionally impaired missense variant identified in French Canadian families implicates FANCI as a candidate ovarian cancer-predisposing gene. Genome Medicine, 2021, 13, 186.	8.2	12
453	Polymorphisms in P21CIP1/WAF1 are not correlated with TP53 status in sporadic ovarian tumours. European Journal of Cancer, 1996, 32, 2360-2363.	2.8	11
454	The effect of the I1307K APC polymorphism on the clinicopathological features and natural history of breast cancer. British Journal of Cancer, 1999, 81, 850-854.	6.4	11
455	Change in the penetrance of founder BRCA1/2 mutations? A retrospective cohort study. Journal of Medical Genetics, 2002, 39, 407-409.	3.2	11
456	Of mice and women. Cancer Cell, 2002, 1, 11-12.	16.8	11
457	Double frameshift mutations in APC and MSH2 in the same individual. International Journal of Colorectal Disease, 2005, 20, 466.	2.2	11
458	BRCA1:185delAG found in the San Luis Valley probably originated in a Jewish founder. Journal of Medical Genetics, 2005, 42, e27-e27.	3.2	11
459	Invasive breast cancer following bilateral subcutaneous mastectomy in a BRCA2 mutation carrier: a case report and review of the literature. World Journal of Surgical Oncology, 2005, 3, 52.	1.9	11
460	Mixed Ovarian Germ Cell Tumor in a BRCA2 Mutation Carrier. International Journal of Gynecological Pathology, 2007, 26, 160-164.	1.4	11
461	A survey of APC mutations in Quebec. Familial Cancer, 2011, 10, 659-665.	1.9	11
462	A Prior Diagnosis of Breast Cancer is a Risk Factor for Breast Cancer in Brca1 and Brca2 Carriers. Current Oncology, 2014, 21, 64-68.	2.2	11
463	CCR 20th Anniversary Commentary: Triple-Negative Breast Cancer in 2015â€”Still in the Ballpark. Clinical Cancer Research, 2015, 21, 3813-3814.	7.0	11
464	Prospective evaluation of body size and breast cancer risk among BRCA1 and BRCA2 mutation carriers. International Journal of Epidemiology, 2018, 47, 987-997.	1.9	11
465	DICER1 screening in 15 paediatric paratesticular sarcomas unveils an unusual DICER1â€associated sarcoma. Journal of Pathology: Clinical Research, 2020, 6, 185-194.	3.0	11
466	<scp>SMARCB1</scp> loss induces druggable cyclin <scp>D1</scp> deficiency via upregulation of <scp>MIR17HG</scp> in atypical teratoid rhabdoid tumors. Journal of Pathology, 2020, 252, 77-87.	4.5	11
467	Molecular characterization of DICER1-mutated pituitary blastoma. Acta Neuropathologica, 2021, 141, 929-944.	7.7	11
468	DICER1-associated sarcomas at different sites exhibit morphological overlap arguing for a unified nomenclature. Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin, 2021, 479, 431-433.	2.8	11

#	Article	IF	Citations
469	Clinical Implications of Next-Generation Sequencing for Cancer Medicine. Current Oncology, 2010, 17, 39-42.	2.2	11
470	Hereditary breast and ovarian cancer: epidemiology, genetics, screening and predictive testing. Clinical and Investigative Medicine, 1995, 18, 473-83.	0.6	11
471	Multiple primary malignancies in a patient with situs ambiguus. Clinical Genetics, 2006, 69, 528-531.	2.0	10
472	Longâ€term outcome after neoâ€adjuvant chemotherapy for breast cancer in BRCA1/2 carriers. International Journal of Cancer, 2009, 125, 2236-2238.	5.1	10
473	MSI-H colorectal cancers preferentially retain and expand intraepithelial lymphocytes rather than peripherally derived CD8+ T cells. Cancer Immunology, Immunotherapy, 2009, 58, 135-144.	4.2	10
474	Genetic architecture of prostate cancer in the Ashkenazi Jewish population. British Journal of Cancer, 2011, 105, 864-869.	6.4	10
475	Anthropometric Measures and Risk of Ovarian Cancer Among BRCA1 and BRCA2 Mutation Carriers. Obesity, 2012, 20, 1288-1292.	3.0	10
476	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
477	Atypical tuberous sclerosis complex presenting as familial renal cell carcinoma with leiomyomatous stroma. Journal of Pathology: Clinical Research, 2018, 4, 167-174.	3.0	10
478	Age at first full-term birth and breast cancer risk in BRCA1 and BRCA2 mutation carriers. Breast Cancer Research and Treatment, 2018, 171, 421-426.	2.5	10
479	65 YEARS OF THE DOUBLE HELIX: Endocrine tumour syndromes in children and adolescents. Endocrine-Related Cancer, 2018, 25, T221-T244.	3.1	10
480	Breastfeeding and the risk of epithelial ovarian cancer among women with a BRCA1 or BRCA2 mutation. Gynecologic Oncology, 2020, 159, 820-826.	1.4	10
481	Loss of heterozygosity of methylenetetrahydrofolate reductase in colon carcinomas.. Oncology Reports, 1999, 6, 597-9.	2.6	10
482	DGCR8 and the six hit, three-step model of schwannomatosis. Acta Neuropathologica, 2022, 143, 115-117.	7.7	10
483	Hyperplastic polyposis and cancer of the colon with gastrinoma of the duodenum. Nature Clinical Practice Oncology, 2006, 3, 281-284.	4.3	9
484	Traffic Control for BRCA1. New England Journal of Medicine, 2010, 362, 755-756.	27.0	9
485	Breast cancer risk in women with PALB2 mutations in different populations. Lancet Oncology, The, 2015, 16, e375-e376.	10.7	9
486	gsSKAT: Rapid gene set analysis and multiple testing correction for rareâ€variant association studies using weighted linear kernels. Genetic Epidemiology, 2017, 41, 297-308.	1.3	9

#	Article	IF	Citations
487	A case of neuroblastoma in DICER1 syndrome: Chance finding or noncanonical causation?. Pediatric Blood and Cancer, 2018, 65, e26715.	1.5	9
488	A complex DICER1 syndrome phenotype associated with a germline pathogenic variant affecting the RNase IIIa domain of DICER1. Journal of Medical Genetics, 2022, 59, 141-146.	3.2	9
489	Olaparib for Metastatic Castration-Resistant Prostate Cancer. New England Journal of Medicine, 2020, 383, 890-891.	27.0	9
490	Relative contribution should be given after each author's name. BMJ: British Medical Journal, 1996, 312, 1423-1423.	2.3	9
491	Cellular context determines <scp>DNA</scp> methylation profiles in <scp>SWI</scp>/<scp>SNF</scp>â€deficient cancers of the gynecologic tract. Journal of Pathology, 2022, 257, 140-145.	4.5	9
492	Incidence of Occult Breast Cancer in Carriers of BRCA1/2 or Other High-Penetrance Pathogenic Variants Undergoing Prophylactic Mastectomy: When is Sentinel Lymph Node Biopsy Indicated?. Annals of Surgical Oncology, 2022, 29, 6660-6668.	1.5	9
493	Mutation analysis of RASK and the â€˜FLR exonâ€™ of NF1 in sporadic ovarian carcinoma. European Journal of Cancer, 1994, 30, 528-530.	2.8	8
494	BRCA mutations and survival in breast cancer.. Journal of Clinical Oncology, 1998, 16, 3206-3208.	1.6	8
495	Ratio of female to male offspring of women tested for BRCA1 and BRCA2 mutations. Journal of Medical Genetics, 2004, 41, e103-e103.	3.2	8
496	Putative common origin of two MLH1 mutations in Italian-Quebec hereditary non-polyposis colorectal cancer families. Clinical Genetics, 2004, 66, 137-143.	2.0	8
497	Novel genomic insertion-deletion in MLH1: possible mechanistic role for non-homologous end-joining DNA repair. Clinical Genetics, 2005, 68, 234-238.	2.0	8
498	A Practical Guide to Human Cancer Genetics. , 2014, , .		8
499	Genetics meets pathology â€“ an increasingly important relationship. Journal of Pathology, 2017, 241, 119-122.	4.5	8
500	Somatic tumour testing establishes that bilateral DICER1â€associated ovarian Sertoliâ€“Leydig cell tumours represent independent primary neoplasms. Histopathology, 2020, 77, 223-230.	2.9	8
501	Contraceptive use and the risk of ovarian cancer among women with a BRCA1 or BRCA2 mutation. Gynecologic Oncology, 2022, 164, 514-521.	1.4	8
502	I1307K APC and hMLH1 mutations in a non-Jewish family with hereditary non-polyposis colorectal cancer. Clinical Genetics, 1998, 54, 368-370.	2.0	7
503	The Value of Multi-Modal Gene Screening in HNPCC in Quebec: Three Mutations in Mismatch Repair Genes that would have not been Correctly Identified by Genomic DNA Sequencing Alone. Familial Cancer, 2006, 5, 21-28.	1.9	7
504	Reading Between the Lines: A Comparison of Responders and Nonâ€responders to a Family History Questionnaire and Implications for Cancer Genetic Counselling. Journal of Genetic Counseling, 2012, 21, 273-291.	1.6	7

#	Article	IF	Citations
505	Bilateral Tumors â€" Inherited or Acquired?. New England Journal of Medicine, 2020, 383, 280-282.	27.0	7
506	Cancer Immunoprevention: A Case Report Raising the Possibility of â€œImmuno-interceptionâ€• Cancer Prevention Research, 2020, 13, 351-356.	1.5	7
507	BRCA1/2 and Endometrial Cancer Risk: Implications for Management. Journal of the National Cancer Institute, 2021, 113, 1127-1128.	6.3	7
508	DICER1 syndrome in a young adult with pituitary blastoma. Acta Neuropathologica, 2021, 142, 1071-1076.	7.7	7
509	Scene memory and hippocampal volume in middle-aged women with early hormone loss. Neurobiology of Aging, 2022, 117, 97-106.	3.1	7
510	Ovarian cancer risk and family history. Lancet, The, 1997, 349, 878.	13.7	6
511	Family with Graves disease, multinodular goiter, nonmedullary thyroid carcinoma, and alveolar rhabdomyosarcoma. , 1997, 72, 30-33.		6
512	Familial Prostate and Breast Cancer in Men Treated with Prostatectomy for Prostate Cancer: A Population based Case-Control Study. Journal of Urology, 2003, 169, 240-244.	0.4	6
513	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.	2.3	6
514	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.	2.8	6
515	An Assessment of the Shared Allelic Architecture between Type II Diabetes and Prostate Cancer. Cancer Epidemiology Biomarkers and Prevention, 2013, 22, 1473-1475.	2.5	6
516	Primary rhabdoid tumor of the ovary: When large cells become small cells. Gynecologic Oncology Reports, 2015, 12, 64-66.	0.6	6
517	Lynch syndrome: five unanswered questions. Clinical Genetics, 2015, 87, 503-506.	2.0	6
518	Does the age of breast cancer diagnosis in first-degree relatives impact on the risk of breast cancer in BRCA1 and BRCA2 mutation carriers?. Breast Cancer Research and Treatment, 2015, 154, 163-169.	2.5	6
519	Clinical follow-up and breast and ovarian cancer screening of true BRCA1/2 noncarriers: a qualitative investigation. Genetics in Medicine, 2016, 18, 627-634.	2.4	6
520	Genome Sequencing of Multiple Primary Tumors Reveals a Novel PALB2 Variant. Journal of Clinical Oncology, 2016, 34, e61-e67.	1.6	6
521	Revisiting pleuropulmonary blastoma and atypical choroid plexus papilloma in a young child: DICER1 syndrome or not?. Pediatric Blood and Cancer, 2018, 65, e27294.	1.5	6
522	Further evidence that full gene deletions of DICER1 predispose to DICER1 syndrome. Genes Chromosomes and Cancer, 2019, 58, 602-604.	2.8	6

#	Article	IF	Citations
523	MRN Complex and Cancer Risk: Old Bottles, New Wine. Clinical Cancer Research, 2021, 27, 5465-5471.	7.0	6
524	Utility of a Cancer Predisposition Screening Tool for Predicting Subsequent Malignant Neoplasms in Childhood Cancer Survivors. Journal of Clinical Oncology, 2021, 39, JCO.21.00018.	1.6	6
525	Weight Gain and the Risk of Ovarian Cancer in BRCA1 and BRCA2 Mutation Carriers. Cancer Epidemiology Biomarkers and Prevention, 2021, 30, 2038-2043.	2.5	6
526	Neuroectodermal elements are part of the morphological spectrum of DICER1-associated neoplasms. Human Pathology, 2022, 123, 46-58.	2.0	6
527	A decade of RAD51C and RAD51D germline variants in cancer. Human Mutation, 2022, 43, 285-298.	2.5	6
528	Intra-Tumoral CD8+ T-Cell Infiltration and PD-L1 Positivity in Homologous Recombination Deficient Pancreatic Ductal Adenocarcinoma. Frontiers in Oncology, 2022, 12, 860767.	2.8	6
529	Comprehensive evaluation and efficient classification of BRCA1 RING domain missense substitutions. American Journal of Human Genetics, 2022, 109, 1153-1174.	6.2	6
530	The expanded role of NF1. Nature Genetics, 1993, 3, 282-282.	21.4	5
531	BRCA1 and BRCA2: penetrating the clinical arena. Lancet, The, 1998, 352, 1325-1326.	13.7	5
532	An absence of founderBRCA2 mutations in individuals with squamous cell carcinoma of the head and neck. , 1999, 83, 803-804.		5
533	Re: Gynecologic Surgeries and Risk of Ovarian Cancer in Women with BRCA1 and BRCA2 Ashkenazi Founder Mutations: An Israeli Population-Based Case-Control Study. Journal of the National Cancer Institute, 2003, 95, 1640-1640.	6.3	5
534	Analysis of Xq27-28 linkage in the international consortium for prostate cancer genetics (ICPCG) families. BMC Medical Genetics, 2012, 13, 46.	2.1	5
535	Hereditary Colorectal Cancer Registries in Canada: Report from the Colorectal Cancer Association of Canada Consensus Meeting; Montreal, Quebec; October 28, 2011. Current Oncology, 2013, 20, 273-278.	2.2	5
536	ClinGen and Genetic Testing. New England Journal of Medicine, 2015, 373, 1376-1379.	27.0	5
537	Post hoc Analysis for Detecting Individual Rare Variant Risk Associations Using Probit Regression Bayesian Variable Selection Methods in Caseâ€Control Sequencing Studies. Genetic Epidemiology, 2016, 40, 461-469.	1.3	5
538	A novel <scp>DICER</scp>1 mutation in familial multinodular goitre. Clinical Endocrinology, 2018, 89, 110-112.	2.4	5
539	Thorough in silico and in vitro cDNA analysis of 21 putativeBRCA1andBRCA2splice variants and a complex tandem duplication inBRCA2allowing the identification of activated cryptic splice donor sites inBRCA2exon 11. Human Mutation, 2018, 39, 515-526.	2.5	5
540	Does preventive oophorectomy increase the risk of depression in BRCA mutation carriers?. Menopause, 2020, 27, 156-161.	2.0	5

#	Article	IF	Citations
541	Reclassification of a frequent Africanâ€origin variant from PMS2 to the pseudogene PMS2CL. Human Mutation, 2020, 41, 749-752.	2.5	5
542	Reprogramming of Nucleotide Metabolism Mediates Synergy between Epigenetic Therapy and MAP Kinase Inhibition. Molecular Cancer Therapeutics, 2021, 20, 64-75.	4.1	5
543	Investigating the causal role of MRE11A p.E506* in breast and ovarian cancer. Scientific Reports, 2021, 11, 2409.	3.3	5
544	Mismatch Repair Universal Screening of Endometrial Cancers (MUSE) in a Canadian Cohort. Current Oncology, 2021, 28, 509-522.	2.2	5
545	Breast cancer risk after age 60 amongÂBRCA1 andÂBRCA2 mutation carriers. Breast Cancer Research and Treatment, 2021, 187, 515-523.	2.5	5
546	A novel mouse model of PMS2 founder mutation that causes mismatch repair defect due to aberrant splicing. Cell Death and Disease, 2021, 12, 838.	6.3	5
547	Mutations in BRCA-related breast and ovarian cancer in the South African Indian population: A descriptive study. Cancer Genetics, 2021, 258-259, 1-6.	0.4	5
548	Family History and Risk of Ovarian Cancer. JAMA - Journal of the American Medical Association, 1995, 274, 383.	7.4	4
549	Genetic Screening for Breast Cancer. New England Journal of Medicine, 1996, 334, 1200-1201.	27.0	4
550	Re: Population-Based Study of BRCA1 and BRCA2 Mutations in 1035 Unselected Finnish Breast Cancer Patients. Journal of the National Cancer Institute, 2001, 93, 152-153.	6.3	4
551	Re: Potential for Bias in Studies on Efficacy of Prophylactic Surgery for BRCA1 and BRCA2 Mutation Carriers. Journal of the National Cancer Institute, 2003, 95, 1344-1344.	6.3	4
552	Sporadic desmoid tumor in an Ashkenazi patient homozygous for theAPC*I1307Kgene mutation. Acta OncolÃ³gica, 2008, 47, 1158-1161.	1.8	4
553	A genome-wide analysis of loss of heterozygosity and chromosomal copy number variation in Proteus syndrome using high-density SNP microarrays. Journal of Human Genetics, 2010, 55, 627-630.	2.3	4
554	Hereditary leiomyomatosis and renal cell cancer syndrome. Cmaj, 2016, 188, 140-140.	2.0	4
555	GATA2 Deficiency Due to de Novo Complete Monoallelic Deletion in an Adolescent With Myelodysplasia. Journal of Pediatric Hematology/Oncology, 2018, 40, e225-e228.	0.6	4
556	Long-term outcomes following a diagnosis of ovarian cancer at the time of preventive oophorectomy among BRCA1 and BRCA2 mutation carriers. International Journal of Gynecological Cancer, 2020, 30, 825-830.	2.5	4
557	Common clonal origin of chronic myelomonocytic leukemia and B-cell acute lymphoblastic leukemia in a patient with a germline CHEK2 variant. Journal of Physical Education and Sports Management, 2021, 7, a006090.	1.2	4
558	Li-Fraumeni Syndrome in the Cancer Genomics Era. Journal of the National Cancer Institute, 2021, 113, 1615-1617.	6.3	4

#	Article	IF	Citations
559	A full molecular picture of F8 intron 1 inversion created with optical genome mapping. Haemophilia, 2021, 27, e638-e640.	2.1	4
560	Circulating tumor DNA is readily detectable among Ghanaian breast cancer patients supporting non-invasive cancer genomic studies in Africa. Npj Precision Oncology, 2021, 5, 83.	5.4	4
561	The Genetic and Molecular Analyses of RAD51C and RAD51D Identifies Rare Variants Implicated in Hereditary Ovarian Cancer from a Genetically Unique Population. Cancers, 2022, 14, 2251.	3.7	4
562	Nonovarian Pelvic Cancers in BRCA1/2 Mutation Carriers and the BRCAPRO Statistical Model. Journal of Clinical Oncology, 2002, 20, 3936-3937.	1.6	3
563	Overview of the clinical genetics of ovarian cancer. , 2002, , 43-72.		3
564	Familial prostate cancer: the damage done and lessons learnt. Nature Reviews Urology, 2013, 10, 116-122.	3.8	3
565	Made-to-measure medicine: BRCA and gynaecological cancer. Lancet Oncology, The, 2014, 15, 783-785.	10.7	3
566	No Evidence of Excessive Cancer Screening in Female Noncarriers from BRCA1/2 Mutationâ€“Positive Families. Current Oncology, 2017, 24, 352-359.	2.2	3
567	Typical 22q11.2 deletion syndrome appears to confer a reduced risk of schwannoma. Genetics in Medicine, 2021, 23, 1779-1782.	2.4	3
568	Current gene panels account for nearly all homologous recombination repair-associated multiple-case breast cancer families. Npj Breast Cancer, 2021, 7, 109.	5.2	3
569	Deletion of Yy1 in mouse lung epithelium unveils molecular mechanisms governing pleuropulmonary blastoma pathogenesis. DMM Disease Models and Mechanisms, 2020, 13, .	2.4	3
570	<scp>SWI</scp>/<scp>SNF</scp>â€deficient undifferentiated malignancies: where to draw the line^{â€}. Journal of Pathology, 2022, 256, 139-142.	4.5	3
571	<scp>PRAME</scp> protein expression in <scp>DICER1</scp>â€related tumours. Journal of Pathology: Clinical Research, 2022, 8, 294-304.	3.0	3
572	Case Review: Whole-Exome Sequencing Analyses Identify Carriers of a Known Likely Pathogenic Intronic BRCA1 Variant in Ovarian Cancer Cases Clinically Negative for Pathogenic BRCA1 and BRCA2 Variants. Genes, 2022, 13, 697.	2.4	3
573	Bilateral Oophorectomy and the Risk of Breast Cancer in BRCA1 Mutation Carriers: A Reappraisal. Cancer Epidemiology Biomarkers and Prevention, 2022, 31, 1351-1358.	2.5	3
574	Is Biannual Surveillance for Pancreatic Cancer Sufficient in Individuals With Genetic Syndromes or Familial Pancreatic Cancer?. Journal of the National Comprehensive Cancer Network: JNCCN, 2022, 20, 663-673.e12.	4.9	3
575	Genetics of cancer. Lancet, The, 1992, 340, 1402-1403.	13.7	2
576	Molecular Genetics of Ovarian Cancer. Molecular Biotechnology, 2001, 19, 013-028.	2.4	2

#	Article	IF	Citations
577	Re: Estrogen Receptor Status of Primary Breast Cancer Is Predictive of Estrogen Receptor Status of Contralateral Breast Cancer. Journal of the National Cancer Institute, 2004, 96, 1040-1041.	6.3	2
578	Effect of Prior Bilateral Oophorectomy on the Presentation of Breast Cancer in BRCA1 and BRCA2 Mutation Carriers. Hereditary Cancer in Clinical Practice, 2005, 3, 53.	1.5	2
579	Muir Torre syndrome and MSH2 mutations: the importance of dermatological awareness. British Journal of Cancer, 2006, 95, 243-244.	6.4	2
580	Will MRI screening deliver the expected survival advantage in BRCA 1 carriers?. Clinical Radiology, 2009, 64, 1045-1047.	1.1	2
581	Weight Gain After Oophorectomy Among Women with a BRCA1 or BRCA2 Mutation. Women's Health, 2015, 11, 453-459.	1.5	2
582	BRCA2: a grown-up cancer susceptibility gene. Endocrine-Related Cancer, 2016, 23, E1-E3.	3.1	2
583	Analysis of DICER1 in familial and sporadic cases of transposition of the great arteries. Congenital Heart Disease, 2018, 13, 401-406.	0.2	2
584	A Case Report of Syndromic Multinodular Goitre in Adolescence: Exploring the Phenotype Overlap between Cowden and DICER1 Syndromes. European Thyroid Journal, 2018, 7, 44-50.	2.4	2
585	65 YEARS OF THE DOUBLE HELIX: Itâ€™s all in the DNA: understanding and managing endocrine neoplasms. Endocrine-Related Cancer, 2018, 25, E5-E7.	3.1	2
586	Journeyâ€™s end: the quest for BRCA-like hereditary breast cancer genes is nearly over. Annals of Oncology, 2019, 30, 1023-1025.	1.2	2
587	A child with neuroblastoma and metachronous anaplastic sarcoma of the kidney: Underlying DICER1 syndrome?. Pediatric Blood and Cancer, 2020, 67, e28488.	1.5	2
588	Risk Assessment & Genetic Testing. , 2002, 107, 29-59.		2
589	The Great Majority of Homologous Recombination Repair-Deficient Tumors Are Accounted for by Established Causes. Frontiers in Genetics, 0, 13, .	2.3	2
590	Screening for ovarian cancer. Lancet, The, 1999, 354, 509-510.	13.7	1
591	Long-term outcome of invasive breast cancer. Lancet, The, 2000, 355, 1551.	13.7	1
592	Management of BRCA1/2 mutation carriers. , 2002, , 237-274.		1
593	Multimodal Molecular Screening Is Required to Improve the Sensitivity of MLH1 and MSH2 Mutation Analysis. Journal of Clinical Oncology, 2002, 20, 1705-1707.	1.6	1
594	Polygenic or Pollyanna?. Nature Medicine, 2002, 8, 328-330.	30.7	1

#	Article	IF	Citations
595	RESPONSE: Re: Germline BRCA1 Mutations and a Basal Epithelial Phenotype in Breast Cancer. Journal of the National Cancer Institute, 2004, 96, 714-714.	6.3	1
596	The Basal Phenotype of BRCA1-Related Breast Cancer. Breast Diseases, 2006, 17, 22-25.	0.0	1
597	Are we there yet?. Current Opinion in Genetics and Development, 2010, 20, 197-200.	3.3	1
598	Screening Mammography: The Turning of the Tide?. Current Oncology, 2014, 21, 205-207.	2.2	1
599	Breast-Cancer Risk in Families With Mutations in PALB2. Obstetrical and Gynecological Survey, 2014, 69, 659-660.	0.4	1
600	Risky business: getting a grip on BRIP. Journal of Medical Genetics, 2016, 53, 296-297.	3.2	1
601	Ovarian carcinoma histotype in Lynch syndrome. Gynecologic Oncology Reports, 2017, 20, 140-141.	0.6	1
602	SMO Syndrome: A Unifying Molecular Diagnosis That Suggests Therapeutic Opportunities. JCO Precision Oncology, 2018, 2, 1-6.	3.0	1
603	Ovarian small cell carcinoma in one of aÂpair of monozygous twins. Familial Cancer, 2019, 18, 161-163.	1.9	1
604	Survey of primary care physiciansâ€™ views about breast and ovarian cancer screening for true BRCA1/2 non-carriers. Journal of Community Genetics, 2020, 11, 205-213.	1.2	1
605	Endometrial Stem/Progenitor cell (ES/PC) Marker Expression Profile in Adenosarcoma and Endometrial Stromal Sarcoma. Cancer Treatment and Research Communications, 2021, 27, 100363.	1.7	1
606	An evaluation of memory and attention in BRCA mutation carriers using an online cognitive assessment tool. Cancer, 2021, 127, 3183-3193.	4.1	1
607	Letter to the Editor: Prevalence of WWP1 Gene Mutations in Patients with Thyroid Nodules. Thyroid, 2021, 31, 1147-1148.	4.5	1
608	An Unusual Enteric Yolk Sac Tumor. International Journal of Gynecological Pathology, 2021, Publish Ahead of Print, .	1.4	1
609	Long-term tumour dormancy in a BRCA1 heterozygote. Journal of Medical Genetics, 2022, , jmedgenet-2021-108269.	3.2	1
610	Comment on: Consensus recommendations from the EXPeRT/PARTNER groups for the diagnosis and therapy of sex cord stromal tumors in children and adolescents. Pediatric Blood and Cancer, 2022, 69, e29650.	1.5	1
611	Patient Experience with a Gynecologic Oncology-Initiated Genetic Testing Model for Women with Tubo-Ovarian Cancer. Current Oncology, 2022, 29, 3565-3575.	2.2	1
612	Gynecologic Cancer Risk and Genetics: Informing an Ideal Model of Gynecologic Cancer Prevention. Current Oncology, 2022, 29, 4632-4646.	2.2	1

#	Article	IF	Citations
613	Breast cancer, desmold tumours, and familial adenomatous polyposis. Lancet, The, 1993, 342, 1560-1561.	13.7	0
614	Breast cancer genetics. Lancet, The, 1997, 350, 366.	13.7	0
615	The natural history of hereditary breast cancer. , 2002, , 81-107.		0
616	Re: Italian Randomized Trial Among Women With Hysterectomy: Tamoxifen and Hormone-Dependent Breast Cancer in High-Risk Women. Journal of the National Cancer Institute, 2003, 95, 917-917.	6.3	0
617	BRCA Mutations and Ductal Carcinoma In SituBRCA. JAMA - Journal of the American Medical Association, 2005, 294, 553.	7.4	0
618	Colonic or coelomic?. Lancet, The, 2010, 375, 1844.	13.7	0
619	Introducing Countercurrentsâ€"A New Feature of Current Oncology. Current Oncology, 2011, 18, 203-203.	2.2	0
620	Epigenetic modification and cancer: mark or stamp?. Endocrine-Related Cancer, 2012, 19, C23-C27.	3.1	0
621	Case 35-2018: A Woman with Back Pain and a Remote History of Breast Cancer. New England Journal of Medicine, 2019, 380, e22.	27.0	0
622	DICER1 Syndrome. , 2021, , 227-265.		0
623	Likely foregut endoderm origin for a postzygotic mutation affecting the RNase IIIb domain of DICER1. Journal of Medical Genetics, 2021, , jmedgenet-2021-107887.	3.2	0
624	Prostate Cancer in Special Populations. , 2010, , 249-288.		0
625	Abstract 2975: Two putative founder MSH6 mutations associated with inherited cancer susceptibility in Ashkenazi Jewish population. , 2010, , .		0
626	The Role of BRCA1 and BRCA2 in Anticancer Drug Therapy. , 2013, , 143-155.		0
627	Genome sequencing of multiple primary tumors to reveal underlying germline cancer susceptibility.. Journal of Clinical Oncology, 2013, 31, 1552-1552.	1.6	0
628	Reproductive System. , 2014, , 89-136.		0
629	Urinary System. , 2014, , 137-144.		0
630	Abstract LB-89: Germ-line and somatic SMARCA4 mutations characterize small cell carcinoma of the ovary, hypercalcemic type. , 2014, , .		0

#	Article	IF	Citations
631	Abstract IA03: DICER1: From ontogenesis to oncogenesis. , 2014, , .		0
632	Abstract 09: Contribution of known and novel BRCA-mediated DNA repair pathway genes to pancreatic cancer susceptibility. , 2014, , .		0
633	Abstract 2429: Nestin expression in tumor cells is associated with BRCA1 positive genotype and a basal-like phenotype in breast cancer. , 2015, , .		0
634	Abstract AS01: Molecular genetic approaches to understanding ovarian cancer. , 2015, , .		0
635	Abstract 4934: Mosaic RNase IIIb domainDICER1mutations in children with multiple primary tumors. , 2015, , .		0
636	Abstract 3422: The influence of genetic and clinical factors on the outcome following a diagnosis of small cell carcinoma of the ovary, hypercalcemic type. , 2016, , .		0
637	Abstract 134: Mutational landscape of breast cancers from PALB2 germline mutation carriers. , 2016, , .		0
638	Abstract LB-019: FGFR1 abnormalities in seizure-associated familial and sporadic dysembryoplastic neuroepithelial tumors. , 2016, , .		0
639	Abstract 2390: The polyclonal nature of multinodular goitre revealed by DICER1 sequencing. , 2016, , .		0
640	Abstract 2479: A functionally nullRAD51Dmissense mutation is strongly associated with ovarian carcinoma. , 2017, , .		0
641	Abstract 2470: Compromised BRCA1-PALB2 interaction is associated with breast cancer risk. , 2017, , .		0
642	Abstract 3304: The mutation landscape of cancers serves as a record of early malignant transformation. , 2018, , .		0
643	Germline TIM-3 Mutations Characterize Sub-Cutaneous Panniculitis T-Cell Lymphomas with Hemophagocytic Lymphohistiocytic Syndrome. Blood, 2018, 132, 1569-1569.	1.4	0
644	Abstract 2727: The premalignant state captured in the landscape of somatic mutations can reveal the cancer cell-of-origin. , 2019, , .		0
645	Cancer geneticsâ€"one family at a time. Clinical and Investigative Medicine, 2019, 42, E7-E13.	0.6	0
646	Abstract 2335: HeterozygousPALB2mutations cause replication stress and DNA repair defects in carrier derived lymphoblastoid cell lines. , 2020, , .		0
647	RARE-17. SURVIVAL BENEFIT FOR INDIVIDUALS WITH CONSTITUTIONAL MISMATCH REPAIR DEFICIENCY SYNDROME AND BRAIN TUMORS WHO UNDERGO SURVEILLANCE PROTOCOL. A REPORT FROM THE INTERNATIONAL REPLICATION REPAIR CONSORTIUM. Neuro-Oncology, 2020, 22, iii445-iii446.	1.2	0
648	RARE-22. GERMLINE PATHOGENIC VARIANT c.1552G>A;p.E518K IN DGCR8 CONFERS SUSCEPTIBILITY FOR SCHWANNOMATOSIS AND THYROID TUMORS. Neuro-Oncology, 2020, 22, iii447-iii447.	1.2	0

#	Article	IF	Citations
649	Intrathyroidal Thymus (Incidentaloma) Mimicking Thyroid Neoplasia in DICER1 Syndrome. European Thyroid Journal, 2021, 10, 257-261.	2.4	0
650	The risks of breast and ovarian cancer associated with the Ashkenazi Jewish founder allele <scp>BRCA2 6174delT</scp>. Clinical Genetics, 2022, 101, 317-323.	2.0	0