William D Foulkes

List of Publications by Year
in descending order

649 papers	46,524 citations	1793 106 h-index	3343 190 g-index
674 all docs	674 docs citations	674 times ranked	45447 citing authors

#	Article	IF	Citations
1	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.	1.5	9
2	Diagnostic criteria for constitutional mismatch repair deficiency (CMMRD): recommendations from the international consensus working group. Journal of Medical Genetics, 2022, 59, 318-327.	1.5	57
3	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.	1.5	14
4	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.	1.5	12
5	<scp>SWI</scp>/<scp>SNF</scp>â€deficient undifferentiated malignancies: where to draw the line^{â€}. Journal of Pathology, 2022, 256, 139-142.	2.1	3
6	DGCR8 and the six hit, three-step model of schwannomatosis. Acta Neuropathologica, 2022, 143, 115-117.	3.9	10
7	Contraceptive use and the risk of ovarian cancer among women with a BRCA1 or BRCA2 mutation. Gynecologic Oncology, 2022, 164, 514-521.	0.6	8
8	The risks of breast and ovarian cancer associated with the Ashkenazi Jewish founder allele <scp>BRCA2 6174delT</scp>. Clinical Genetics, 2022, 101, 317-323.	1.0	0
9	Long-term tumour dormancy in a BRCA1 heterozygote. Journal of Medical Genetics, 2022, , jmedgenet-2021-108269.	1.5	1
10	<scp>PRAME</scp> protein expression in <scp>DICER1</scp>â€related tumours. Journal of Pathology: Clinical Research, 2022, 8, 294-304.	1.3	3
11	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.	0.8	1
12	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.	2.1	9
13	Neuroectodermal elements are part of the morphological spectrum of DICER1-associated neoplasms. Human Pathology, 2022, 123, 46-58.	1.1	6
14	A decade of RAD51C and RAD51D germline variants in cancer. Human Mutation, 2022, 43, 285-298.	1.1	6
15	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.	1.0	3
16	Bilateral Oophorectomy and the Risk of Breast Cancer in BRCA1 Mutation Carriers: A Reappraisal. Cancer Epidemiology Biomarkers and Prevention, 2022, 31, 1351-1358.	1.1	3
17	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.	1.7	4
18	Intra-Tumoral CD8+ T-Cell Infiltration and PD-L1 Positivity in Homologous Recombination Deficient Pancreatic Ductal Adenocarcinoma. Frontiers in Oncology, 2022, 12, 860767.	1.3	6

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19	Scene memory and hippocampal volume in middle-aged women with early hormone loss. Neurobiology of Aging, 2022, 117, 97-106.	1.5	7
20	Patient Experience with a Gynecologic Oncology-Initiated Genetic Testing Model for Women with Tubo-Ovarian Cancer. Current Oncology, 2022, 29, 3565-3575.	0.9	1
21	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.	0.7	9
22	Comprehensive evaluation and efficient classification of BRCA1 RING domain missense substitutions. American Journal of Human Genetics, 2022, 109, 1153-1174.	2.6	6
23	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.	2.3	3
24	Gynecologic Cancer Risk and Genetics: Informing an Ideal Model of Gynecologic Cancer Prevention. Current Oncology, 2022, 29, 4632-4646.	0.9	1
25	DICER1-associated sarcomas: towards a unified nomenclature. Modern Pathology, 2021, 34, 1226-1228.	2.9	22
26	<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.	1.5	29
27	Reprogramming of Nucleotide Metabolism Mediates Synergy between Epigenetic Therapy and MAP Kinase Inhibition. Molecular Cancer Therapeutics, 2021, 20, 64-75.	1.9	5
28	The genetic landscape of choroid plexus tumors in children and adults. Neuro-Oncology, 2021, 23, 650-660.	0.6	26
29	Clinical Outcomes and Complications of Pituitary Blastoma. Journal of Clinical Endocrinology and Metabolism, 2021, 106, 351-363.	1.8	23
30	Atypical teratoid/rhabdoid tumors (ATRTs) with SMARCA4 mutation are molecularly distinct from SMARCB1-deficient cases. Acta Neuropathologica, 2021, 141, 291-301.	3.9	47
31	Endometrial Stem/Progenitor cell (ES/PC) Marker Expression Profile in Adenosarcoma and Endometrial Stromal Sarcoma. Cancer Treatment and Research Communications, 2021, 27, 100363.	0.7	1
32	Investigating the causal role of MRE11A p.E506* in breast and ovarian cancer. Scientific Reports, 2021, 11, 2409.	1.6	5
33	Prevalence and Spectrum of DICER1 Mutations in Adult-onset Thyroid Nodules with Indeterminate Cytology. Journal of Clinical Endocrinology and Metabolism, 2021, 106, e968-e977.	1.8	26
34	Mismatch Repair Universal Screening of Endometrial Cancers (MUSE) in a Canadian Cohort. Current Oncology, 2021, 28, 509-522.	0.9	5
35	Breast cancer risk after age 60 amongÂBRCA1 andÂBRCA2 mutation carriers. Breast Cancer Research and Treatment, 2021, 187, 515-523.	1.1	5
36	DICER1 Syndrome. , 2021, , 227-265.		0

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37	BRCA1/2 and Endometrial Cancer Risk: Implications for Management. Journal of the National Cancer Institute, 2021, 113, 1127-1128.	3.0	7
38	Achieving clinical success with BET inhibitors as anti-cancer agents. British Journal of Cancer, 2021, 124, 1478-1490.	2.9	204
39	Clinical practice guidelines for BRCA1 and BRCA2 genetic testing. European Journal of Cancer, 2021, 146, 30-47.	1.3	81
40	The ten genes for breast (and ovarian) cancer susceptibility. Nature Reviews Clinical Oncology, 2021, 18, 259-260.	12.5	19
41	Molecular characterization of DICER1-mutated pituitary blastoma. Acta Neuropathologica, 2021, 141, 929-944.	3.9	11
42	Typical 22q11.2 deletion syndrome appears to confer a reduced risk of schwannoma. Genetics in Medicine, 2021, 23, 1779-1782.	1.1	3
43	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.	1.4	11
44	Management of individuals with germline variants in PALB2: a clinical practice resource of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2021, 23, 1416-1423.	1.1	34
45	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.	2.9	21
46	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.	0.5	4
47	An evaluation of memory and attention in BRCA mutation carriers using an online cognitive assessment tool. Cancer, 2021, 127, 3183-3193.	2.0	1
48	Li-Fraumeni Syndrome in the Cancer Genomics Era. Journal of the National Cancer Institute, 2021, 113, 1615-1617.	3.0	4
49	A full molecular picture of F8 intron 1 inversion created with optical genome mapping. Haemophilia, 2021, 27, e638-e640.	1.0	4
50	Letter to the Editor: Prevalence of WWP1 Gene Mutations in Patients with Thyroid Nodules. Thyroid, 2021, 31, 1147-1148.	2.4	1
51	MRN Complex and Cancer Risk: Old Bottles, New Wine. Clinical Cancer Research, 2021, 27, 5465-5471.	3.2	6
52	Current gene panels account for nearly all homologous recombination repair-associated multiple-case breast cancer families. Npj Breast Cancer, 2021, 7, 109.	2.3	3
53	An Unusual Enteric Yolk Sac Tumor. International Journal of Gynecological Pathology, 2021, Publish Ahead of Print, .	0.9	1
54	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.	0.8	6

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55	Weight Gain and the Risk of Ovarian Cancer in BRCA1 and BRCA2 Mutation Carriers. Cancer Epidemiology Biomarkers and Prevention, 2021, 30, 2038-2043.	1.1	6
56	Survival Benefit for Individuals With Constitutional Mismatch Repair Deficiency Undergoing Surveillance. Journal of Clinical Oncology, 2021, 39, 2779-2790.	0.8	40
57	Likely foregut endoderm origin for a postzygotic mutation affecting the RNase IIIb domain of DICER1. Journal of Medical Genetics, 2021, , jmedgenet-2021-107887.	1.5	0
58	SMARCA4/2 loss inhibits chemotherapy-induced apoptosis by restricting IP3R3-mediated Ca2+ flux to mitochondria. Nature Communications, 2021, 12, 5404.	5.8	20
59	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.	2.3	4
60	A novel mouse model of PMS2 founder mutation that causes mismatch repair defect due to aberrant splicing. Cell Death and Disease, 2021, 12, 838.	2.7	5
61	Mutations in BRCA-related breast and ovarian cancer in the South African Indian population: A descriptive study. Cancer Genetics, 2021, 258-259, 1-6.	0.2	5
62	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.	0.9	12
63	DICER1 syndrome in a young adult with pituitary blastoma. Acta Neuropathologica, 2021, 142, 1071-1076.	3.9	7
64	DICER1-sarcoma: an emerging entity. Modern Pathology, 2021, 34, 2096-2097.	2.9	14
65	Performance of the McGill Interactive Pediatric OncoGenetic Guidelines for Identifying Cancer Predisposition Syndromes. JAMA Oncology, 2021, 7, 1806.	3.4	22
66	Intrathyroidal Thymus (Incidentaloma) Mimicking Thyroid Neoplasia in DICER1 Syndrome. European Thyroid Journal, 2021, 10, 257-261.	1.2	0
67	A functionally impaired missense variant identified in French Canadian families implicates FANCI as a candidate ovarian cancer-predisposing gene. Genome Medicine, 2021, 13, 186.	3.6	12
68	An update on the central nervous system manifestations of DICER1 syndrome. Acta Neuropathologica, 2020, 139, 689-701.	3.9	68
69	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.	2.3	22
70	Macrofollicular Variant of Follicular Thyroid Carcinoma: A Rare Underappreciated Pitfall in the Diagnosis of Thyroid Carcinoma. Thyroid, 2020, 30, 72-80.	2.4	22
71	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.	0.5	1
72	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. Nature Genetics, 2020, 52, 56-73.	9.4	120

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73	Cancer Risks Associated With Germline PALB2 Pathogenic Variants: An International Study of 524 Families. Journal of Clinical Oncology, 2020, 38, 674-685.	0.8	270
74	Does preventive oophorectomy increase the risk of depression in BRCA mutation carriers?. Menopause, 2020, 27, 156-161.	0.8	5
75	Significantly greater prevalence of DICER1 alterations in uterine embryonal rhabdomyosarcoma compared to adenosarcoma. Modern Pathology, 2020, 33, 1207-1219.	2.9	43
76	Breastfeeding and the risk of epithelial ovarian cancer among women with a BRCA1 or BRCA2 mutation. Gynecologic Oncology, 2020, 159, 820-826.	0.6	10
77	Etiologic Index â€" A Case-Only Measure of BRCA1/2â€"Associated Cancer Risk. New England Journal of Medicine, 2020, 383, 286-288.	13.9	12
78	Bilateral Tumors â€" Inherited or Acquired?. New England Journal of Medicine, 2020, 383, 280-282.	13.9	7
79	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.	3.8	33
80	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.	1.2	15
81	Evaluation of molecular analysis in challenging ovarian sex cord-stromal tumours: a review of 50 cases. Pathology, 2020, 52, 686-693.	0.3	12
82	A Preclinical Trial and Molecularly Annotated Patient Cohort Identify Predictive Biomarkers in Homologous Recombinationâ€"deficient Pancreatic Cancer. Clinical Cancer Research, 2020, 26, 5462-5476.	3.2	20
83	Olaparib for Metastatic Castration-Resistant Prostate Cancer. New England Journal of Medicine, 2020, 383, 890-891.	13.9	9
84	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.	1.2	4
85	DICER1 screening in 15 paediatric paratesticular sarcomas unveils an unusual DICER1â€associated sarcoma. Journal of Pathology: Clinical Research, 2020, 6, 185-194.	1.3	11
86	The contribution of large genomic rearrangements in BRCA1 and BRCA2 to South African familial breast cancer. BMC Cancer, 2020, 20, 391.	1.1	12
87	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.	2.8	75
88	Tumour predisposition and cancer syndromes as models to study geneâ€"environment interactions. Nature Reviews Cancer, 2020, 20, 533-549.	12.8	93
89	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.	1.4	45
90	A child with neuroblastoma and metachronous anaplastic sarcoma of the kidney: Underlying DICER1 syndrome?. Pediatric Blood and Cancer, 2020, 67, e28488.	0.8	2

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91	<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.	2.1	11
92	Cognitive markers of dementia risk in middle-aged women with bilateral salpingo-oophorectomy prior to menopause. Neurobiology of Aging, 2020, 94, 1-6.	1.5	19
93	Stathmin expression associates with vascular and immune responses in aggressive breast cancer subgroups. Scientific Reports, 2020, 10, 2914.	1.6	18
94	Ovarian and Breast Cancer Risks Associated With Pathogenic Variants in RAD51C and RAD51D. Journal of the National Cancer Institute, 2020, 112, 1242-1250.	3.0	106
95	Pineoblastoma is uniquely tolerant of mutually exclusive loss of DICER1, DROSHA or DGCR8. Acta Neuropathologica, 2020, 139, 1115-1118.	3.9	15
96	Poorly differentiated thyroid carcinoma of childhood and adolescence: a distinct entity characterized by DICER1 mutations. Modern Pathology, 2020, 33, 1264-1274.	2.9	96
97	Reclassification of a frequent Africanâ€origin variant from PMS2 to the pseudogene PMS2CL. Human Mutation, 2020, 41, 749-752.	1.1	5
98	Somatic tumour testing establishes that bilateral DICER1â€associated ovarian Sertoliâ€“Leydig cell tumours represent independent primary neoplasms. Histopathology, 2020, 77, 223-230.	1.6	8
99	Founder BRCA1/BRCA2/PALB2 pathogenic variants in French-Canadian breast cancer cases and controls. Scientific Reports, 2020, 10, 6491.	1.6	24
100	Small-Cell Carcinoma of the Ovary, Hypercalcemic Typeâ€“Genetics, New Treatment Targets, and Current Management Guidelines. Clinical Cancer Research, 2020, 26, 3908-3917.	3.2	82
101	Cancer Immunoprevention: A Case Report Raising the Possibility of â€œImmuno-interceptionâ€. Cancer Prevention Research, 2020, 13, 351-356.	0.7	7
102	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.	2.9	40
103	Methionine Metabolism Shapes T Helper Cell Responses through Regulation of Epigenetic Reprogramming. Cell Metabolism, 2020, 31, 250-266.e9.	7.2	182
104	Embryonal Rhabdomyosarcoma of the Ovary and Fallopian Tube. American Journal of Surgical Pathology, 2020, 44, 738-747.	2.1	42
105	DGCR8 microprocessor defect characterizes familial multinodular goiter with schwannomatosis. Journal of Clinical Investigation, 2020, 130, 1479-1490.	3.9	31
106	Deletion of Yy1 in mouse lung epithelium unveils molecular mechanisms governing pleuropulmonary blastoma pathogenesis. DMM Disease Models and Mechanisms, 2020, 13, .	1.2	3
107	Abstract 2335: HeterozygousPALB2mutations cause replication stress and DNA repair defects in carrier derived lymphoblastoid cell lines. , 2020, , .		0
108	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.	0.6	0

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109	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.	0.6	0
110	Homologous recombination DNA repair defects in PALB2-associated breast cancers. Npj Breast Cancer, 2019, 5, 23.	2.3	39
111	Ten years of DICER1 mutations: Provenance, distribution, and associated phenotypes. Human Mutation, 2019, 40, 1939-1953.	1.1	76
112	Imaging of DICER1 syndrome. Pediatric Radiology, 2019, 49, 1488-1505.	1.1	34
113	The FANCM:p.Arg658* truncating variant is associated with risk of triple-negative breast cancer. Npj Breast Cancer, 2019, 5, 38.	2.3	28
114	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.	1.1	15
115	eIF4A Inhibitors Suppress Cell-Cycle Feedback Response and Acquired Resistance to CDK4/6 Inhibition in Cancer. Molecular Cancer Therapeutics, 2019, 18, 2158-2170.	1.9	25
116	Interim Results from the IMPACT Study: Evidence for Prostate-specific Antigen Screening in BRCA2 Mutation Carriers. European Urology, 2019, 76, 831-842.	0.9	148
117	Oestrogen receptor status and survival in women with BRCA2-associated breast cancer. British Journal of Cancer, 2019, 120, 398-403.	2.9	25
118	CDK4/6 inhibitors target SMARCA4-determined cyclin D1 deficiency in hypercalcemic small cell carcinoma of the ovary. Nature Communications, 2019, 10, 558.	5.8	76
119	SMARCA4 loss is synthetic lethal with CDK4/6 inhibition in non-small cell lung cancer. Nature Communications, 2019, 10, 557.	5.8	125
120	Large scale multifactorial likelihood quantitative analysis of BRCA1 and BRCA2 variants: An ENIGMA resource to support clinical variant classification. Human Mutation, 2019, 40, 1557-1578.	1.1	102
121	BRCA1andBRCA2pathogenic sequence variants in women of African origin or ancestry. Human Mutation, 2019, 40, 1781-1796.	1.1	26
122	Mesenchymal Hamartoma of the Liver and DICER1 Syndrome. New England Journal of Medicine, 2019, 380, 1834-1842.	13.9	39
123	Journeyâ€™s end: the quest for BRCA-like hereditary breast cancer genes is nearly over. Annals of Oncology, 2019, 30, 1023-1025.	0.6	2
124	SWI/SNF-Compromised Cancers Are Susceptible to Bromodomain Inhibitors. Cancer Research, 2019, 79, 2761-2774.	0.4	54
125	The dilemma of early preventive oophorectomy in familial small cell carcinoma of the ovary of hypercalcemic type. Gynecologic Oncology Reports, 2019, 28, 47-49.	0.3	12
126	Functional Repair Assay for the Diagnosis of Constitutional Mismatch Repair Deficiency From Non-Neoplastic Tissue. Journal of Clinical Oncology, 2019, 37, 461-470.	0.8	23

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127	Case 35-2018: A Woman with Back Pain and a Remote History of Breast Cancer. New England Journal of Medicine, 2019, 380, e22.	13.9	0
128	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.	1.1	30
129	Mutational Signature Analysis Reveals NTHL1 Deficiency to Cause a Multi-tumor Phenotype. Cancer Cell, 2019, 35, 256-266.e5.	7.7	123
130	Oophorectomy and risk of contralateral breast cancer among BRCA1 and BRCA2 mutation carriers. Breast Cancer Research and Treatment, 2019, 175, 443-449.	1.1	12
131	Further evidence that full gene deletions of DICER1 predispose to DICER1 syndrome. Genes Chromosomes and Cancer, 2019, 58, 602-604.	1.5	6
132	NovelPOLEpathogenic germline variant in a family with multiple primary tumors results in distinct mutational signatures. Human Mutation, 2019, 40, 36-41.	1.1	21
133	Expanding the morphological spectrum of ovarian microcystic stromal tumour. Histopathology, 2019, 74, 443-451.	1.6	24
134	Ovarian small cell carcinoma in one of aÂpair of monozygous twins. Familial Cancer, 2019, 18, 161-163.	0.9	1
135	Clinical and Molecular Characteristics May Alter Treatment Strategies of Thyroid Malignancies in DICER1 Syndrome. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 277-284.	1.8	22
136	Abstract 2727: The premalignant state captured in the landscape of somatic mutations can reveal the cancer cell-of-origin. , 2019, , .		0
137	Cancer geneticsâ€"one family at a time. Clinical and Investigative Medicine, 2019, 42, E7-E13.	0.3	0
138	DICER1 Mutations Are Frequent in Adolescent-Onset Papillary Thyroid Carcinoma. Journal of Clinical Endocrinology and Metabolism, 2018, 103, 2009-2015.	1.8	79
139	Clinical testing of BRCA1 and BRCA2: a worldwide snapshot of technological practices. Npj Genomic Medicine, 2018, 3, 7.	1.7	44
140	Analysis of DICER1 in familial and sporadic cases of transposition of the great arteries. Congenital Heart Disease, 2018, 13, 401-406.	0.0	2
141	Atypical tuberous sclerosis complex presenting as familial renal cell carcinoma with leiomyomatous stroma. Journal of Pathology: Clinical Research, 2018, 4, 167-174.	1.3	10
142	A novel <scp>DICER</scp>1 mutation in familial multinodular goitre. Clinical Endocrinology, 2018, 89, 110-112.	1.2	5
143	Hormone Replacement Therapy After Oophorectomy and Breast Cancer Risk Among BRCA1 Mutation Carriers. JAMA Oncology, 2018, 4, 1059.	3.4	121
144	GATA2 Deficiency Due to de Novo Complete Monoallelic Deletion in an Adolescent With Myelodysplasia. Journal of Pediatric Hematology/Oncology, 2018, 40, e225-e228.	0.3	4

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145	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.	1.1	25
146	Hereditary SWI/SNF complex deficiency syndromes. Seminars in Diagnostic Pathology, 2018, 35, 193-198.	1.0	51
147	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.	1.2	2
148	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.	1.1	5
149	DICER1 gene mutations in endocrine tumors. Endocrine-Related Cancer, 2018, 25, R197-R208.	1.6	29
150	Multiple DICER1â€related tumors in a child with a large interstitial 14q32 deletion. Genes Chromosomes and Cancer, 2018, 57, 223-230.	1.5	33
151	Prostate-specific antigen velocity in a prospective prostate cancer screening study of men with genetic predisposition. British Journal of Cancer, 2018, 118, 266-276.	2.9	12
152	Prospective evaluation of body size and breast cancer risk among BRCA1 and BRCA2 mutation carriers. International Journal of Epidemiology, 2018, 47, 987-997.	0.9	11
153	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.	1.1	27
154	Anaplastic sarcomas of the kidney are characterized by DICER1 mutations. Modern Pathology, 2018, 31, 169-178.	2.9	55
155	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.	2.1	41
156	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.	1.9	20
157	A case of neuroblastoma in DICER1 syndrome: Chance finding or noncanonical causation?. Pediatric Blood and Cancer, 2018, 65, e26715.	0.8	9
158	SMO Syndrome: A Unifying Molecular Diagnosis That Suggests Therapeutic Opportunities. JCO Precision Oncology, 2018, 2, 1-6.	1.5	1
159	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.	1.5	18
160	BRCA Challenge: BRCA Exchange as a global resource for variants in BRCA1 and BRCA2. PLoS Genetics, 2018, 14, e1007752.	1.5	148
161	Loss of human ICOSL results in combined immunodeficiency. Journal of Experimental Medicine, 2018, 215, 3151-3164.	4.2	40
162	Germline HAVCR2 mutations altering TIM-3 characterize subcutaneous panniculitis-like T cell lymphomas with hemophagocytic lymphohistiocytic syndrome. Nature Genetics, 2018, 50, 1650-1657.	9.4	151

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