William D Foulkes

List of Publications by Year in Descending Order

610 papers	35,929 citations	97 h-index	170 g-index
673 ext. papers	41,779 ext. citations	9.3 avg, IF	7.14 L-index

#	Paper	IF	Citations
610	Contraceptive use and the risk of ovarian cancer among women with a BRCA1 or BRCA2 mutation.. *Gynecologic Oncology*, **2022**,	4.9	1
609	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**, e29650	3	
608	Cellular context determines DNA methylation profiles in SWI/SNF-deficient cancers of the gynecologic tract.. *Journal of Pathology*, **2022**,	9.4	1
607	Intra-Tumoral CD8+ T-Cell Infiltration and PD-L1 Positivity in Homologous Recombination Deficient Pancreatic Ductal Adenocarcinoma.. *Frontiers in Oncology*, **2022**, 12, 860767	5.3	0
606	Patient Experience with a Gynecologic Oncology-Initiated Genetic Testing Model for Women with Tubo-Ovarian Cancer. *Current Oncology*, **2022**, 29, 3565-3575	2.8	
605	Comprehensive evaluation and efficient classification of BRCA1 RING domain missense substitutions. *American Journal of Human Genetics*, **2022**, 109, 1153-1174	11	1
604	Intrathyroidal Thymus (Incidentaloma) Mimicking Thyroid Neoplasia in DICER1 Syndrome. *European Thyroid Journal*, **2021**, 10, 257-261	4.2	
603	SWI/SNF-deficient undifferentiated malignancies: where to draw the line. *Journal of Pathology*, **2021**, 256, 139	9.4	
602	DGCR8 and the six hit, three-step model of schwannomatosis. *Acta Neuropathologica*, **2021**, 143, 115	14.3	0
601	DICER1 syndrome in a young adult with pituitary blastoma. *Acta Neuropathologica*, **2021**, 142, 1071-1076	14.3	2
600	The Value of DICER1 Mutation Analysis in "Subtle" Diagnostically Challenging Embryonal Rhabdomyosarcomas of the Uterine Cervix. *International Journal of Gynecological Pathology*, **2021**, 40, 435-440	3.2	6
599	Achieving clinical success with BET inhibitors as anti-cancer agents. *British Journal of Cancer*, **2021**, 124, 1478-1490	8.7	40
598	Clinical practice guidelines for BRCA1 and BRCA2 genetic testing. *European Journal of Cancer*, **2021**, 146, 30-47	7.5	15
597	The ten genes for breast (and ovarian) cancer susceptibility. *Nature Reviews Clinical Oncology*, **2021**, 18, 259-260	19.4	3
596	Molecular characterization of DICER1-mutated pituitary blastoma. *Acta Neuropathologica*, **2021**, 141, 929-944	14.3	6
595	Typical 22q11.2 deletion syndrome appears to confer a reduced risk of schwannoma. *Genetics in Medicine*, **2021**, 23, 1779-1782	8.1	2
594	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	5.1	1

593	Data sharing to improve concordance in variant interpretation across laboratories: results from the Canadian Open Genetics Repository. *Journal of Medical Genetics*, **2021**,	5.8	3
592	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	8.1	5
591	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	9.8	2
590	An evaluation of memory and attention in BRCA mutation carriers using an online cognitive assessment tool. *Cancer*, **2021**, 127, 3183-3193	6.4	
589	Li-Fraumeni Syndrome in the Cancer Genomics Era. *Journal of the National Cancer Institute*, **2021**,	9.7	1
588	A full molecular picture of F8 intron 1 inversion created with optical genome mapping. *Haemophilia*, **2021**, 27, e638-e640	3.3	0
587	DICER1-associated sarcomas: towards a unified nomenclature. *Modern Pathology*, **2021**, 34, 1226-1228	9.8	8
586	DICER1-associated embryonal rhabdomyosarcoma and adenosarcoma of the gynecologic tract: Pathology, molecular genetics, and indications for molecular testing. *Genes Chromosomes and Cancer*, **2021**, 60, 217-233	5	7
585	Reprogramming of Nucleotide Metabolism Mediates Synergy between Epigenetic Therapy and MAP Kinase Inhibition. *Molecular Cancer Therapeutics*, **2021**, 20, 64-75	6.1	3
584	The genetic landscape of choroid plexus tumors in children and adults. *Neuro-Oncology*, **2021**, 23, 650-660		3
583	Clinical Outcomes and Complications of Pituitary Blastoma. *Journal of Clinical Endocrinology and Metabolism*, **2021**, 106, 351-363	5.6	9
582	Atypical teratoid/rhabdoid tumors (ATRTs) with SMARCA4 mutation are molecularly distinct from SMARCB1-deficient cases. *Acta Neuropathologica*, **2021**, 141, 291-301	14.3	14
581	Endometrial Stem/Progenitor cell (ES/PC) Marker Expression Profile in Adenosarcoma and Endometrial Stromal Sarcoma. *Cancer Treatment and Research Communications*, **2021**, 27, 100363	2	0
580	Investigating the causal role of MRE11A p.E506* in breast and ovarian cancer. *Scientific Reports*, **2021**, 11, 2409	4.9	3
579	Prevalence and Spectrum of DICER1 Mutations in Adult-onset Thyroid Nodules with Indeterminate Cytology. *Journal of Clinical Endocrinology and Metabolism*, **2021**, 106, 968-977	5.6	3
578	Mismatch Repair Universal Screening of Endometrial Cancers (MUSE) in a Canadian Cohort. *Current Oncology*, **2021**, 28, 509-522	2.8	0
577	Breast cancer risk after age 60 among ́BRCA1 and ́BRCA2 mutation carriers. *Breast Cancer Research and Treatment*, **2021**, 187, 515-523	4.4	1
576	DICER1 Syndrome **2021**, 227-265		

575	Diagnostic criteria for constitutional mismatch repair deficiency (CMMRD): recommendations from the international consensus working group. *Journal of Medical Genetics*, **2021**,	5.8	11
574	Prevalence of Gene Mutations in Patients with Thyroid Nodules. *Thyroid*, **2021**, 31, 1147-1148	6.2	
573	MRN Complex and Cancer Risk: Old Bottles, New Wine. *Clinical Cancer Research*, **2021**, 27, 5465-5471	12.9	1
572	Current gene panels account for nearly all homologous recombination repair-associated multiple-case breast cancer families. *Npj Breast Cancer*, **2021**, 7, 109	7.8	0
571	Utility of a Cancer Predisposition Screening Tool for Predicting Subsequent Malignant Neoplasms in Childhood Cancer Survivors. *Journal of Clinical Oncology*, **2021**, 39, 3207-3216	2.2	0
570	Weight Gain and the Risk of Ovarian Cancer in and Mutation Carriers. *Cancer Epidemiology Biomarkers and Prevention*, **2021**, 30, 2038-2043	4	1
569	Survival Benefit for Individuals With Constitutional Mismatch Repair Deficiency Undergoing Surveillance. *Journal of Clinical Oncology*, **2021**, 39, 2779-2790	2.2	10
568	SMARCA4/2 loss inhibits chemotherapy-induced apoptosis by restricting IP3R3-mediated Ca flux to mitochondria. *Nature Communications*, **2021**, 12, 5404	17.4	3
567	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	9.8	0
566	A novel mouse model of PMS2 founder mutation that causes mismatch repair defect due to aberrant splicing. *Cell Death and Disease*, **2021**, 12, 838	9.8	0
565	Mutations in BRCA-related breast and ovarian cancer in the South African Indian population: A descriptive study. *Cancer Genetics*, **2021**, 258-259, 1-6	2.3	2
564	A functionally impaired missense variant identified in French Canadian families implicates FANCI as a candidate ovarian cancer-predisposing gene. *Genome Medicine*, **2021**, 13, 186	14.4	2
563	Long-term outcomes following a diagnosis of ovarian cancer at the time of preventive oophorectomy among and mutation carriers. *International Journal of Gynecological Cancer*, **2020**, 30, 825-830	3.5	1
562	DICER1 screening in 15 paediatric paratesticular sarcomas unveils an unusual DICER1-associated sarcoma. *Journal of Pathology: Clinical Research*, **2020**, 6, 185-194	5.3	6
561	The contribution of large genomic rearrangements in BRCA1 and BRCA2 to South African familial breast cancer. *BMC Cancer*, **2020**, 20, 391	4.8	4
560	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	10.4	31
559	Tumour predisposition and cancer syndromes as models to study gene-environment interactions. *Nature Reviews Cancer*, **2020**, 20, 533-549	31.3	32
558	Malignant teratoid tumor of the thyroid gland: an aggressive primitive multiphenotypic malignancy showing organotypical elements and frequent DICER1 alterations-is the term "thyroblastoma" more appropriate?. *Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin*, **2020**, 477, 787-798	5.1	19

557	A child with neuroblastoma and metachronous anaplastic sarcoma of the kidney: Underlying DICER1 syndrome?. *Pediatric Blood and Cancer*, **2020**, 67, e28488	3	1
556	SMARCB1 loss induces druggable cyclin D1 deficiency via upregulation of MIR17HG in atypical teratoid rhabdoid tumors. *Journal of Pathology*, **2020**, 252, 77-87	9.4	6
555	Cognitive markers of dementia risk in middle-aged women with bilateral salpingo-oophorectomy prior to menopause. *Neurobiology of Aging*, **2020**, 94, 1-6	5.6	11
554	Stathmin expression associates with vascular and immune responses in aggressive breast cancer subgroups. *Scientific Reports*, **2020**, 10, 2914	4.9	5
553	Ovarian and Breast Cancer Risks Associated With Pathogenic Variants in RAD51C and RAD51D. *Journal of the National Cancer Institute*, **2020**, 112, 1242-1250	9.7	51
552	Pineoblastoma is uniquely tolerant of mutually exclusive loss of DICER1, DROSHA or DGCR8. *Acta Neuropathologica*, **2020**, 139, 1115-1118	14.3	7
551	Poorly differentiated thyroid carcinoma of childhood and adolescence: a distinct entity characterized by DICER1 mutations. *Modern Pathology*, **2020**, 33, 1264-1274	9.8	45
550	Reclassification of a frequent African-origin variant from PMS2 to the pseudogene PMS2CL. *Human Mutation*, **2020**, 41, 749-752	4.7	1
549	Somatic tumour testing establishes that bilateral DICER1-associated ovarian Sertoli-Leydig cell tumours represent independent primary neoplasms. *Histopathology*, **2020**, 77, 223-230	7.3	6
548	DGCR8 microprocessor defect characterizes familial multinodular goiter with schwannomatosis. *Journal of Clinical Investigation*, **2020**, 130, 1479-1490	15.9	15
547	Deletion of in mouse lung epithelium unveils molecular mechanisms governing pleuropulmonary blastoma pathogenesis. *DMM Disease Models and Mechanisms*, **2020**, 13,	4.1	2
546	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	78
545	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	78
544	Methionine Metabolism Shapes T Helper Cell Responses through Regulation of Epigenetic Reprogramming. *Cell Metabolism*, **2020**, 31, 250-266.e9	24.6	91
543	Embryonal Rhabdomyosarcoma of the Ovary and Fallopian Tube: Rare Neoplasms Associated With Germline and Somatic DICER1 Mutations. *American Journal of Surgical Pathology*, **2020**, 44, 738-747	6.7	28
542	Macrofollicular Variant of Follicular Thyroid Carcinoma: A Rare Underappreciated Pitfall in the Diagnosis of Thyroid Carcinoma. *Thyroid*, **2020**, 30, 72-80	6.2	11
541	Survey of primary care physiciansM views about breast and ovarian cancer screening for true BRCA1/2 non-carriers. *Journal of Community Genetics*, **2020**, 11, 205-213	2.5	0
540	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. *Nature Genetics*, **2020**, 52, 56-73	36.3	56

539	Cancer Risks Associated With Germline Pathogenic Variants: An International Study of 524 Families. *Journal of Clinical Oncology*, **2020**, 38, 674-685	2.2	133
538	Does preventive oophorectomy increase the risk of depression in BRCA mutation carriers?. *Menopause*, **2020**, 27, 156-161	2.5	3
537	Significantly greater prevalence of DICER1 alterations in uterine embryonal rhabdomyosarcoma compared to adenosarcoma. *Modern Pathology*, **2020**, 33, 1207-1219	9.8	24
536	Breastfeeding and the risk of epithelial ovarian cancer among women with a BRCA1 or BRCA2 mutation. *Gynecologic Oncology*, **2020**, 159, 820-826	4.9	3
535	Etiologic Index - A Case-Only Measure of -Associated Cancer Risk. *New England Journal of Medicine*, **2020**, 383, 286-288	59.2	9
534	Bilateral Tumors - Inherited or Acquired?. *New England Journal of Medicine*, **2020**, 383, 280-282	59.2	5
533	A complex DICER1 syndrome phenotype associated with a germline pathogenic variant affecting the RNase IIIa domain of DICER1. *Journal of Medical Genetics*, **2020**,	5.8	2
532	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, 1957-1969	27.4	8
531	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	3.5	5
530	Evaluation of molecular analysis in challenging ovarian sex cord-stromal tumours: a review of 50 cases. *Pathology*, **2020**, 52, 686-693	1.6	3
529	A Preclinical Trial and Molecularly Annotated Patient Cohort Identify Predictive Biomarkers in Homologous Recombination-deficient Pancreatic Cancer. *Clinical Cancer Research*, **2020**, 26, 5462-5476	12.9	14
528	Olaparib for Metastatic Castration-Resistant Prostate Cancer. *New England Journal of Medicine*, **2020**, 383, 890	59.2	5
527	An update on the central nervous system manifestations of DICER1 syndrome. *Acta Neuropathologica*, **2020**, 139, 689-701	14.3	34
526	An eHealth decision-support tool to prioritize referral practices for genetic evaluation of patients with Wilms tumor. *International Journal of Cancer*, **2020**, 146, 1010-1017	7.5	12
525	Founder BRCA1/BRCA2/PALB2 pathogenic variants in French-Canadian breast cancer cases and controls. *Scientific Reports*, **2020**, 10, 6491	4.9	14
524	Small-Cell Carcinoma of the Ovary, Hypercalcemic Type-Genetics, New Treatment Targets, and Current Management Guidelines. *Clinical Cancer Research*, **2020**, 26, 3908-3917	12.9	28
523	Cancer Immunoprevention: A Case Report Raising the Possibility of "Immuno-interception". *Cancer Prevention Research*, **2020**, 13, 351-356	3.2	5
522	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	9.8	16

521	eIF4A Inhibitors Suppress Cell-Cycle Feedback Response and Acquired Resistance to CDK4/6 Inhibition in Cancer. *Molecular Cancer Therapeutics*, **2019**, 18, 2158-2170	6.1	14
520	Interim Results from the IMPACT Study: Evidence for Prostate-specific Antigen Screening in BRCA2 Mutation Carriers. *European Urology*, **2019**, 76, 831-842	10.2	78
519	Oestrogen receptor status and survival in women with BRCA2-associated breast cancer. *British Journal of Cancer*, **2019**, 120, 398-403	8.7	13
518	CDK4/6 inhibitors target SMARCA4-determined cyclin D1 deficiency in hypercalcemic small cell carcinoma of the ovary. *Nature Communications*, **2019**, 10, 558	17.4	42
517	SMARCA4 loss is synthetic lethal with CDK4/6 inhibition in non-small cell lung cancer. *Nature Communications*, **2019**, 10, 557	17.4	72
516	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	4.7	52
515	BRCA1 and BRCA2 pathogenic sequence variants in women of African origin or ancestry. *Human Mutation*, **2019**, 40, 1781-1796	4.7	16
514	Mesenchymal Hamartoma of the Liver and DICER1 Syndrome. *New England Journal of Medicine*, **2019**, 380, 1834-1842	59.2	23
513	SWI/SNF-Compromised Cancers Are Susceptible to Bromodomain Inhibitors. *Cancer Research*, **2019**, 79, 2761-2774	10.1	36
512	The dilemma of early preventive oophorectomy in familial small cell carcinoma of the ovary of hypercalcemic type. *Gynecologic Oncology Reports*, **2019**, 28, 47-49	1.3	9
511	Functional Repair Assay for the Diagnosis of Constitutional Mismatch Repair Deficiency From Non-Neoplastic Tissue. *Journal of Clinical Oncology*, **2019**, 37, 461-470	2.2	17
510	Case 35-2018: A Woman with Back Pain and a Remote History of Breast Cancer. *New England Journal of Medicine*, **2019**, 380, e22	59.2	
509	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	4.7	18
508	Mutational Signature Analysis Reveals NTHL1 Deficiency to Cause a Multi-tumor Phenotype. *Cancer Cell*, **2019**, 35, 256-266.e5	24.3	72
507	Homologous recombination DNA repair defects in associated breast cancers. *Npj Breast Cancer*, **2019**, 5, 23	7.8	20
506	Ten years of DICER1 mutations: Provenance, distribution, and associated phenotypes. *Human Mutation*, **2019**, 40, 1939-1953	4.7	37
505	Imaging of DICER1 syndrome. *Pediatric Radiology*, **2019**, 49, 1488-1505	2.8	18
504	The :p.Arg658* truncating variant is associated with risk of triple-negative breast cancer. *Npj Breast Cancer*, **2019**, 5, 38	7.8	12

503	Exome Sequencing in and -Negative Greek Families Identifies and as Candidate Risk Genes for Hereditary Breast Cancer. *Frontiers in Genetics*, **2019**, 10, 1005	4.5	7
502	Cancer genetics-one family at a time. *Clinical and Investigative Medicine*, **2019**, 42, E7-E13	0.9	
501	Oophorectomy and risk of contralateral breast cancer among BRCA1 and BRCA2 mutation carriers. *Breast Cancer Research and Treatment*, **2019**, 175, 443-449	4.4	7
500	Further evidence that full gene deletions of DICER1 predispose to DICER1 syndrome. *Genes Chromosomes and Cancer*, **2019**, 58, 602-604	5	6
499	Novel POLE pathogenic germline variant in a family with multiple primary tumors results in distinct mutational signatures. *Human Mutation*, **2019**, 40, 36-41	4.7	12
498	Expanding the morphological spectrum of ovarian microcystic stromal tumour. *Histopathology*, **2019**, 74, 443-451	7.3	9
497	Ovarian small cell carcinoma in one of a´pair of monozygous twins. *Familial Cancer*, **2019**, 18, 161-163	3	0
496	Clinical and Molecular Characteristics May Alter Treatment Strategies of Thyroid Malignancies in DICER1 Syndrome. *Journal of Clinical Endocrinology and Metabolism*, **2019**, 104, 277-284	5.6	13
495	DICER1 Mutations Are Frequent in Adolescent-Onset Papillary Thyroid Carcinoma. *Journal of Clinical Endocrinology and Metabolism*, **2018**, 103, 2009-2015	5.6	47
494	Clinical testing of and : a worldwide snapshot of technological practices. *Npj Genomic Medicine*, **2018**, 3, 7	6.2	29
493	Analysis of DICER1 in familial and sporadic cases of transposition of the great arteries. *Congenital Heart Disease*, **2018**, 13, 401-406	3.1	2
492	Atypical tuberous sclerosis complex presenting as familial renal cell carcinoma with leiomyomatous stroma. *Journal of Pathology: Clinical Research*, **2018**, 4, 167-174	5.3	7
491	A novel DICER1 mutation in familial multinodular goitre. *Clinical Endocrinology*, **2018**, 89, 110-112	3.4	3
490	Hormone Replacement Therapy After Oophorectomy and Breast Cancer Risk Among BRCA1 Mutation Carriers. *JAMA Oncology*, **2018**, 4, 1059-1065	13.4	84
489	GATA2 Deficiency Due to de Novo Complete Monoallelic Deletion in an Adolescent With Myelodysplasia. *Journal of Pediatric Hematology/Oncology*, **2018**, 40, e225-e228	1.2	2
488	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	4.4	19
487	Hereditary SWI/SNF complex deficiency syndromes. *Seminars in Diagnostic Pathology*, **2018**, 35, 193-198	4.3	39
486	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	4.2	1

485	Thorough in silico and in vitro cDNA analysis of 21 putative BRCA1 and BRCA2 splice variants and a complex tandem duplication in BRCA2 allowing the identification of activated cryptic splice donor sites in BRCA2 exon 11. *Human Mutation*, **2018**, 39, 515-526	4.7	5
484	gene mutations in endocrine tumors. *Endocrine-Related Cancer*, **2018**, 25, R197-R208	5.7	16
483	Multiple DICER1-related tumors in a child with a large interstitial 14q32 deletion. *Genes Chromosomes and Cancer*, **2018**, 57, 223-230	5	29
482	Prostate-specific antigen velocity in a prospective prostate cancer screening study of men with genetic predisposition. *British Journal of Cancer*, **2018**, 118, 266-276	8.7	9
481	Prospective evaluation of body size and breast cancer risk among BRCA1 and BRCA2 mutation carriers. *International Journal of Epidemiology*, **2018**, 47, 987-997	7.8	6
480	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	8.1	20
479	Anaplastic sarcomas of the kidney are characterized by DICER1 mutations. *Modern Pathology*, **2018**, 31, 169-178	9.8	35
478	65 YEARS OF THE DOUBLE HELIX: Endocrine tumour syndromes in children and adolescents. *Endocrine-Related Cancer*, **2018**, 25, T221-T244	5.7	6
477	Retrospective evaluation of a decision-support algorithm (MIPOGG) for genetic referrals for children with neuroblastic tumors. *Pediatric Blood and Cancer*, **2018**, 65, e27390	3	16
476	Infantile Pulmonary Teratoid Tumor. *New England Journal of Medicine*, **2018**, 378, 2238-2240	59.2	12
475	The risk of breast cancer in BRCA1 and BRCA2 mutation carriers without a first-degree relative with breast cancer. *Clinical Genetics*, **2018**, 93, 1063-1068	4	9
474	Paediatric ovarian tumours and their associated cancer susceptibility syndromes. *Journal of Medical Genetics*, **2018**, 55, 1-10	5.8	12
473	Germline TIM-3 Mutations Characterize Sub-Cutaneous Panniculitis T-Cell Lymphomas with Hemophagocytic Lymphohistiocytic Syndrome. *Blood*, **2018**, 132, 1569-1569	2.2	
472	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	6.7	17
471	Familial multinodular goiter and Sertoli-Leydig cell tumors associated with a large intragenic in-frame deletion. *European Journal of Endocrinology*, **2018**, 178, K11-K19	6.5	17
470	A case of neuroblastoma in DICER1 syndrome: Chance finding or noncanonical causation?. *Pediatric Blood and Cancer*, **2018**, 65, e26715	3	6
469	SMO Syndrome: A Unifying Molecular Diagnosis That Suggests Therapeutic Opportunities.. *JCO Precision Oncology*, **2018**, 2, 1-6	3.6	1
468	Reflex Testing for Germline , , , and Mutations in Pancreatic Cancer: Mutation Prevalence and Clinical Outcomes From Two Canadian Research Registries.. *JCO Precision Oncology*, **2018**, 2, 1-16	3.6	10

467	BRCA Challenge: BRCA Exchange as a global resource for variants in BRCA1 and BRCA2. *PLoS Genetics*, **2018**, 14, e1007752	6	90
466	Loss of human ICOSL results in combined immunodeficiency. *Journal of Experimental Medicine*, **2018**, 215, 3151-3164	16.6	21
465	Germline HAVCR2 mutations altering TIM-3 characterize subcutaneous panniculitis-like T cell lymphomas with hemophagocytic lymphohistiocytic syndrome. *Nature Genetics*, **2018**, 50, 1650-1657	36.3	74
464	Age-specific ovarian cancer risks among women with a BRCA1 or BRCA2 mutation. *Gynecologic Oncology*, **2018**, 150, 85-91	4.9	47
463	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	4.4	8
462	Revisiting pleuropulmonary blastoma and atypical choroid plexus papilloma in a young child: DICER1 syndrome or not?. *Pediatric Blood and Cancer*, **2018**, 65, e27294	3	5
461	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	3	19
460	Clinical, morphological and immunohistochemical evidence that small-cell carcinoma of the ovary of hypercalcaemic type (SCCOHT) may be a primitive germ-cell neoplasm. *Histopathology*, **2017**, 70, 1147-1154	7.3	23
459	Oncogenic role of PDK4 in human colon cancer cells. *British Journal of Cancer*, **2017**, 116, 930-936	8.7	55
458	gsSKAT: Rapid gene set analysis and multiple testing correction for rare-variant association studies using weighted linear kernels. *Genetic Epidemiology*, **2017**, 41, 297-308	2.6	5
457	Expression of Nestin associates with BRCA1 mutations, a basal-like phenotype and aggressive breast cancer. *Scientific Reports*, **2017**, 7, 1089	4.9	13
456	Cancer Surveillance in Gorlin Syndrome and Rhabdoid Tumor Predisposition Syndrome. *Clinical Cancer Research*, **2017**, 23, e62-e67	12.9	97
455	, and Their Associated Tumor Susceptibility Syndromes: Clinical Features, Genetics, and Surveillance Recommendations in Childhood. *Clinical Cancer Research*, **2017**, 23, e76-e82	12.9	97
454	Sequencing of DICER1 in sarcomas identifies biallelic somatic DICER1 mutations in an adult-onset embryonal rhabdomyosarcoma. *British Journal of Cancer*, **2017**, 116, 1621-1626	8.7	24
453	Ovarian carcinoma histotype in Lynch syndrome. *Gynecologic Oncology Reports*, **2017**, 20, 140-141	1.3	1
452	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. *Nature Genetics*, **2017**, 49, 680-691	36.3	190
451	Risk of breast cancer after a diagnosis of ovarian cancer in BRCA mutation carriers: Is preventive mastectomy warranted?. *Gynecologic Oncology*, **2017**, 145, 346-351	4.9	23
450	Compromised BRCA1-PALB2 interaction is associated with breast cancer risk. *Oncogene*, **2017**, 36, 4161-4170	5.7	51

449	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. *Nature Genetics*, **2017**, 49, 1767-1778	36.3	186
448	No evidence of excessive cancer screening in female noncarriers from mutation-positive families. *Current Oncology*, **2017**, 24, 352-359	2.8	1
447	A mutational signature reveals alterations underlying deficient homologous recombination repair in breast cancer. *Nature Genetics*, **2017**, 49, 1476-1486	36.3	255
446	Functionally Null Missense Mutation Associates Strongly with Ovarian Carcinoma. *Cancer Research*, **2017**, 77, 4517-4529	10.1	18
445	Retinoblastoma and Neuroblastoma Predisposition and Surveillance. *Clinical Cancer Research*, **2017**, 23, e98-e106	12.9	117
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3	Tumor mutational landscape is a record of the pre-malignant state		8
2	Fine-mapping of 150 breast cancer risk regions identifies 178 high confidence target genes		2
1	PRSS2 stimulates tumor growth by remodeling the TME via repression of Tsp1		1