Douglas R Stewart

List of Publications by Year
in descending order

80 papers	4,084 citations	117625 34 h-index	128289 60 g-index
83 all docs	83 docs citations	83 times ranked	5166 citing authors

#	Article	IF	Citations
1	ACMG SF v3.0 list for reporting of secondary findings in clinical exome and genome sequencing: a policy statement of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2021, 23, 1381-1390.	2.4	356
2	Histopathologic evaluation of atypical neurofibromatous tumors and their transformation into malignant peripheral nerve sheath tumor in patients with neurofibromatosis 1â€"a consensus overview. Human Pathology, 2017, 67, 1-10.	2.0	275
3	DICER1 and Associated Conditions: Identification of At-risk Individuals and Recommended Surveillance Strategies. Clinical Cancer Research, 2018, 24, 2251-2261.	7.0	260
4	Pleuropulmonary blastoma: A report on 350 central pathologyâ€"confirmed pleuropulmonary blastoma cases by the <scp>I</scp>nternational <scp>P</scp>leuropulmonary <scp>B</scp>lastoma <scp>R</scp>egistry. Cancer, 2015, 121, 276-285.	4.1	242
5	Recommendations for reporting of secondary findings in clinical exome and genome sequencing, 2021 update: a policy statement of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2021, 23, 1391-1398.	2.4	145
6	Care of adults with neurofibromatosis type 1: a clinical practice resource of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2018, 20, 671-682.	2.4	128
7	Temporal order of RNase IIIb and loss-of-function mutations during development determines phenotype in DICER1 syndrome: a unique variant of the two-hit tumor suppression model. F1000Research, 2015, 4, 214.	1.6	125
8	Glomus Tumors in Neurofibromatosis Type 1: Genetic, Functional, and Clinical Evidence of a Novel Association. Cancer Research, 2009, 69, 7393-7401.	0.9	122
9	Quantification of Thyroid Cancer and Multinodular Goiter Risk in the DICER1 Syndrome: A Family-Based Cohort Study. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 1614-1622.	3.6	120
10	Neoplasm Risk Among Individuals With a Pathogenic Germline Variant in DICER1. Journal of Clinical Oncology, 2019, 37, 668-676.	1.6	107
11	DICER1-Pleuropulmonary Blastoma Familial Tumor Predisposition Syndrome. , 2014, 19, 90-100.		97
12	DICER1-related Sertoli-Leydig cell tumor and gynandroblastoma: Clinical and genetic findings from the International Ovarian and Testicular Stromal Tumor Registry. Gynecologic Oncology, 2017, 147, 521-527.	1.4	87
13	Malignant Peripheral Nerve Sheath Tumors State of the Science: Leveraging Clinical and Biological Insights into Effective Therapies. Sarcoma, 2017, 2017, 1-10.	1.3	84
14	Nasal chondromesenchymal hamartomas arise secondary to germline and somatic mutations of DICER1 in the pleuropulmonary blastoma tumor predisposition disorder. Human Genetics, 2014, 133, 1443-1450.	3.8	82
15	Genomic and evolutionary classification of lung cancer in never smokers. Nature Genetics, 2021, 53, 1348-1359.	21.4	81
16	Neurofibromatosis Type 1â€"Associated MPNST State of the Science: Outlining a Research Agenda for the Future. Journal of the National Cancer Institute, 2017, 109, .	6.3	80
17	The chromosome 9q subtelomere deletion syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2007, 145C, 383-392.	1.6	79
18	Temporal order of RNase IIIb and loss-of-function mutations during development determines phenotype in pleuropulmonary blastoma / DICER1 syndrome: a unique variant of the two-hit tumor suppression model. F1000Research, 2015, 4, 214.	1.6	78

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19	The prevalence of DICER1 pathogenic variation in population databases. International Journal of Cancer, 2017, 141, 2030-2036.	5.1	75
20	Low mutation burden and frequent loss of CDKN2A/B and SMARCA2, but not PRC2, define premalignant neurofibromatosis type 1â€“associated atypical neurofibromas. Neuro-Oncology, 2019, 21, 981-992.	1.2	69
21	Variable population prevalence estimates of germline TP53 variants: A gnomAD-based analysis. Human Mutation, 2019, 40, 97-105.	2.5	66
22	DICER1 tumor predisposition syndrome: an evolving story initiated with the pleuropulmonary blastoma. Modern Pathology, 2022, 35, 4-22.	5.5	65
23	Diagnosis, management, and complications of glomus tumours of the digits in neurofibromatosis type 1. Journal of Medical Genetics, 2010, 47, 525-532.	3.2	61
24	Mitotic recombination as evidence of alternative pathogenesis of gastrointestinal stromal tumours in neurofibromatosis type 1. Journal of Medical Genetics, 2006, 44, e61-e61.	3.2	59
25	Points to consider for reporting of germline variation in patients undergoing tumor testing: a statement of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2020, 22, 1142-1148.	2.4	59
26	Is Pulmonary Arterial Hypertension in Neurofibromatosis Type 1 Secondary to a Plexogenic Arteriopathy?. Chest, 2007, 132, 798-808.	0.8	56
27	The Cyclic AMP Pathway Is a Sex-Specific Modifier of Glioma Risk in Type I Neurofibromatosis Patients. Cancer Research, 2015, 75, 16-21.	0.9	56
28	Germline Cancer Predisposition Variants inâ€, Pediatric Rhabdomyosarcoma: A Report From the Childrenâ€™s Oncology Group. Journal of the National Cancer Institute, 2021, 113, 875-883.	6.3	55
29	Germline mutations in Protection of Telomeres 1 in two families with Hodgkin lymphoma. British Journal of Haematology, 2018, 181, 372-377.	2.5	48
30	Evidence of polyclonality in neurofibromatosis type 2â€“associated multilobulated vestibular schwannomas. Neuro-Oncology, 2015, 17, 566-573.	1.2	45
31	Association of Inherited Pathogenic Variants in Checkpoint Kinase 2 (CHEK2) With Susceptibility to Testicular Germ Cell Tumors. JAMA Oncology, 2019, 5, 514.	7.1	43
32	Whole exome sequencing reveals a C-terminal germline variant in CEBPA-associated acute myeloid leukemia: 45-year follow up of a large family. Haematologica, 2016, 101, 846-852.	3.5	42
33	Macrocephaly associated with the DICER1 syndrome. Genetics in Medicine, 2017, 19, 244-248.	2.4	42
34	Higher-than-expected population prevalence of potentially pathogenic germlineTP53variants in individuals unselected for cancer history. Human Mutation, 2017, 38, 1723-1730.	2.5	40
35	Advancing <scp>RAS/RASopathy</scp> therapies: An NCIâ€sponsored intramural and extramural collaboration for the study of <scp>RASopathies</scp>. American Journal of Medical Genetics, Part A, 2020, 182, 866-876.	1.2	40
36	Mosaic chromosome Y loss and testicular germ cell tumor risk. Journal of Human Genetics, 2017, 62, 637-640.	2.3	34

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37	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
38	Jaffeâ€“Campanacci syndrome, revisited: detailed clinical and molecular analyses determine whether patients have neurofibromatosis type 1, coincidental manifestations, or a distinct disorder. Genetics in Medicine, 2014, 16, 448-459.	2.4	33
39	Genetic Modifiers of Neurofibromatosis Type 1-Associated CafÃ©-au-Lait Macule Count Identified Using Multi-platform Analysis. PLoS Genetics, 2014, 10, e1004575.	3.5	31
40	Dubowitz Syndrome Is a Complex Comprised of Multiple, Genetically Distinct and Phenotypically Overlapping Disorders. PLoS ONE, 2014, 9, e98686.	2.5	29
41	Cephalometry in adults and children with neurofibromatosis type 1: Implications for the pathogenesis of sphenoid wing dysplasia and the â€œNF1 facies. European Journal of Medical Genetics, 2015, 58, 584-590.	1.3	29
42	Mitotic recombination of chromosome arm 17q as a cause of loss of heterozygosity of NF1 in neurofibromatosis type 1â€associated glomus tumors. Genes Chromosomes and Cancer, 2012, 51, 429-437.	2.8	27
43	Pathogenic Germline Variants in Cancer Susceptibility Genes in Children and Young Adults With Rhabdomyosarcoma. JCO Precision Oncology, 2021, 5, 75-87.	3.0	27
44	A Genome-First Approach to Characterize DICER1 Pathogenic Variant Prevalence, Penetrance, and Phenotype. JAMA Network Open, 2021, 4, e210112.	5.9	25
45	Rare inactivating PDE11A variants associated with testicular germ cell tumors. Endocrine-Related Cancer, 2015, 22, 909-917.	3.1	24
46	Research participant interest in primary, secondary, and incidental genomic findings. Genetics in Medicine, 2016, 18, 1218-1225.	2.4	24
47	The prevalence of germline DICER1 pathogenic variation in cancer populations. Molecular Genetics & Genomic Medicine, 2019, 7, e555.	1.2	24
48	Pleuropulmonary blastoma-like peritoneal sarcoma: a newly described malignancy associated with biallelic DICER1 pathogenic variation. Modern Pathology, 2020, 33, 1922-1929.	5.5	24
49	Presacral malignant teratoid neoplasm in association with pathogenic DICER1 variation. Modern Pathology, 2019, 32, 1744-1750.	5.5	22
50	Juvenile myelomonocytic leukemia due to a germline CBL Y371C mutation: 35-year follow-up of a large family. Human Genetics, 2015, 134, 775-787.	3.8	21
51	DICER1 Syndrome. Ophthalmology, 2019, 126, 296-304.	5.2	20
52	Comparative clinical and genomic analysis of neurofibromatosis type 2-associated cranial and spinal meningiomas. Scientific Reports, 2020, 10, 12563.	3.3	16
53	A genotype-first approach to exploring Mendelian cardiovascular traits with clear external manifestations. Genetics in Medicine, 2021, 23, 94-102.	2.4	16
54	Prevalence of pathogenic/likely pathogenic variants in the 24 cancer genes of the ACMG Secondary Findings v2.0 list in a large cancer cohort and ethnicity-matched controls. Genome Medicine, 2018, 10, 99.	8.2	15

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55	Structural renal abnormalities in the DICER1 syndrome: a family-based cohort study. Pediatric Nephrology, 2018, 33, 2281-2288.	1.7	14
56	First insight into the somatic mutation burden of neurofibromatosis type 2-associated grade I and grade II meningiomas: a case report comprehensive genomic study of two cranial meningiomas with vastly different clinical presentation. BMC Cancer, 2017, 17, 127.	2.6	13
57	Frequency of Pathogenic Germline Variants in Cancer-Susceptibility Genes in the Childhood Cancer Survivor Study. JNCI Cancer Spectrum, 2021, 5, pkab007.	2.9	11
58	Incidence of Benign Meningiomas in the United States: Current and Future Trends. JNCI Cancer Spectrum, 2021, 5, pkab035.	2.9	11
59	Gynecologic and reproductive health in patients with pathogenic germline variants in DICER1. Gynecologic Oncology, 2020, 156, 647-653.	1.4	10
60	Hematologic indices in individuals with pathogenic germline DICER1 variants. Blood Advances, 2021, 5, 216-223.	5.2	6
61	Endemic Burkitt Lymphoma in second-degree relatives in Northern Uganda: in-depth genome-wide analysis suggests clues about genetic susceptibility. Leukemia, 2021, 35, 1209-1213.	7.2	5
62	Unusual phenotypes in patients with a pathogenic germline variant in DICER1. Familial Cancer, 2021, , 1.	1.9	5
63	Nasal chondromesenchymal hamartomas in a cohort with pathogenic germline variation in DICER1. Rhinology, 2020, 3, 15-24.	0.3	5
64	Prospectively Identified Incident Testicular Cancer Risk in a Familial Testicular Cancer Cohort. Cancer Epidemiology Biomarkers and Prevention, 2015, 24, 1614-1621.	2.5	4
65	Dental abnormalities in individuals with pathogenic germline variation in DICER1. American Journal of Medical Genetics, Part A, 2019, 179, 1820-1825.	1.2	4
66	Stomaching Multigene Panel Testing: What to Do About CDH1?. Journal of the National Cancer Institute, 2020, 112, 325-326.	6.3	4
67	Genomeâ€wide association study of cafÃ©â€auâ€lait macule number in neurofibromatosis type 1. Molecular Genetics & Genomic Medicine, 2020, 8, e1400.	1.2	4
68	Testicular Sertoli cell tumour and potentially testicular Leydig cell tumour are features of DICER1 syndrome. Journal of Medical Genetics, 2022, 59, 346-350.	3.2	4
69	In search of genetic factors predisposing to familial hairy cell leukemia (HCL): exome-sequencing of four multiplex HCL pedigrees. Leukemia, 2020, 34, 1934-1938.	7.2	3
70	Quantification of Discordant Variant Interpretations in a Large Family-Based Study of Li-Fraumeni Syndrome. JCO Precision Oncology, 2021, 5, 1727-1737.	3.0	3
71	Identification of Genetic Risk Factors for Familial Urinary Bladder Cancer: An Exome Sequencing Study. JCO Precision Oncology, 2021, 5, 1830-1839.	3.0	3
72	Comment on: DICER1â€Negative Pleuropulmonary Blastoma in a Patient With Selective IgA Deficiency. Pediatric Blood and Cancer, 2016, 63, 1869-1870.	1.5	2

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73	Response to: Concern regarding classification of germline TP53 variants as likely pathogenic. Human Mutation, 2019, 40, 832-833.	2.5	1
74	Novel MAPK/AKT-impairing germline NRAS variant identified in a melanoma-prone family. Familial Cancer, 2022, 21, 347-355.	1.9	1
75	Another Weak Link in the Electron Transport Chain: Variants in SUCLG2 and Risk for Pheochromocytoma and Paraganglioma. Journal of the National Cancer Institute, 2021, , .	6.3	1
76	Response to Hannah-Shmouni and Stratakis. Genetics in Medicine, 2019, 21, 1256.	2.4	0
77	Lack of pathogenic germline DICER1 variants in males with testicular germ-cell tumors. Cancer Genetics, 2020, 248-249, 49-56.	0.4	0
78	Reply: pleuropulmonary blastoma-like peritoneal sarcoma and DICER1-associated sarcomas: toward a unified nomenclature. Modern Pathology, 2021, 34, 1229-1230.	5.5	0
79	Whole exome sequencing and copy-number variation analysis of 20 NF2-associated spinal and cranial meningiomas.. Journal of Clinical Oncology, 2017, 35, 2051-2051.	1.6	0
80	Insights Into Immune-Mediated Disease and Cancer Riskâ€"Delivering on the Promise of UK Biobank Big Data. JAMA Oncology, 2021, , .	7.1	0