Edward D Esplin

List of Publications by Year
in descending order

46 papers	1,645 citations	516561 16 h-index	330025 37 g-index
50 all docs	50 docs citations	50 times ranked	3257 citing authors

#	Article	IF	Citations
1	The Human Tumor Atlas Network: Charting Tumor Transitions across Space and Time at Single-Cell Resolution. Cell, 2020, 181, 236-249.	13.5	334
2	Prevalence of Germline Variants in Prostate Cancer and Implications for Current Genetic Testing Guidelines. JAMA Oncology, 2019, 5, 523.	3.4	240
3	Comparison of Universal Genetic Testing vs Guideline-Directed Targeted Testing for Patients With Hereditary Cancer Syndrome. JAMA Oncology, 2021, 7, 230.	3.4	146
4	Perinatal features of the RASopathies: Noonan syndrome, Cardiofaciocutaneous syndrome and Costello syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 2814-2821.	0.7	78
5	Single-cell analyses define a continuum of cell state and composition changes in the malignant transformation of polyps to colorectal cancer. Nature Genetics, 2022, 54, 985-995.	9.4	77
6	Yield and Utility of Germline Testing Following Tumor Sequencing in Patients With Cancer. JAMA Network Open, 2020, 3, e2019452.	2.8	76
7	Possible precision medicine implications from genetic testing using combined detection of sequence and intragenic copy number variants in a large cohort with childhood epilepsy. Epilepsia Open, 2019, 4, 397-408.	1.3	68
8	Personalized sequencing and the future of medicine: discovery, diagnosis and defeat of disease. Pharmacogenomics, 2014, 15, 1771-1790.	0.6	66
9	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.	1.1	59
10	Expanded Gene Panel Use for Women With Breast Cancer: Identification and Intervention Beyond Breast Cancer Risk. Annals of Surgical Oncology, 2017, 24, 3060-3066.	0.7	54
11	Underdiagnosis of Hereditary Breast and Ovarian Cancer in Medicare Patients: Genetic Testing Criteria Miss the Mark. Annals of Surgical Oncology, 2018, 25, 2925-2931.	0.7	53
12	Prevalence of pathogenic germline cancer risk variants in high-risk urothelial carcinoma. Genetics in Medicine, 2020, 22, 709-718.	1.1	44
13	Nine patients with Xp22.31 microduplication, cognitive deficits, seizures, and talipes anomalies. American Journal of Medical Genetics, Part A, 2014, 164, 2097-2103.	0.7	38
14	RTTN Mutations Cause Primary Microcephaly and Primordial Dwarfism in Humans. American Journal of Human Genetics, 2015, 97, 862-868.	2.6	36
15	Developing and Optimizing Innovative Tools to Address Familial Hypercholesterolemia Underdiagnosis. Circulation Genomic and Precision Medicine, 2021, 14, e003120.	1.6	23
16	Points to consider: is there evidence to support BRCA1/2 and other inherited breast cancer genetic testing for all breast cancer patients? A statement of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2020, 22, 681-685.	1.1	20
17	Democratizing genomics: Leveraging software to make genetics an integral part of routine care. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2021, 187, 14-27.	0.7	20
18	CDH1 germline variants are enriched in patients with colorectal cancer, gastric cancer, and breast cancer. British Journal of Cancer, 2022, 126, 797-803.	2.9	17

#	Article	IF	Citations
19	Trans-ethnic variation in germline variants of patients with renal cell carcinoma. Cell Reports, 2021, 34, 108926.	2.9	16
20	Expanding the phenotype of cardiovascular malformations in Adamsâ€“Oliver syndrome. American Journal of Medical Genetics, Part A, 2013, 161, 1386-1389.	0.7	15
21	Biallelic lossâ€ofâ€function WNT5A mutations in an infant with severe and atypical manifestations of Robinow syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 1030-1036.	0.7	15
22	Misattributed parentage as an unanticipated finding during exome/genome sequencing: current clinical laboratory practices and an opportunity for standardization. Genetics in Medicine, 2019, 21, 861-866.	1.1	14
23	Limited-Variant Screening vs Comprehensive Genetic Testing for Familial Hypercholesterolemia Diagnosis. JAMA Cardiology, 2021, 6, 902.	3.0	14
24	Response to â€œThe use of ACMG secondary findings recommendations for general population screening: a policy statement of the American College of Medical Genetics and Genomics (ACMG)â€• Genetics in Medicine, 2019, 21, 2836-2837.	1.1	11
25	Fumarate hydratase variant prevalence and manifestations among individuals receiving germline testing. Cancer, 2022, 128, 675-684.	2.0	11
26	Multigene Panel Testing in Individuals With Hepatocellular Carcinoma Identifies Pathogenic Germline Variants. JCO Precision Oncology, 2021, 5, 988-1000.	1.5	10
27	Germline hemizygous deletion of CDKN2Aâ€“CDKN2B locus in a patient presenting with Liâ€“Fraumeni syndrome. Npj Genomic Medicine, 2016, 1, 16015.	1.7	9
28	Germline Cancer Susceptibility Gene Testing in Unselected Patients with Hepatobiliary Cancers: A Multi-Center Prospective Study. Cancer Prevention Research, 2022, 15, 121-128.	0.7	9
29	Clinical Implications of Pathogenic Germline Variants in Small Intestine Neuroendocrine Tumors (SI-NETs). JCO Precision Oncology, 2021, 5, 808-816.	1.5	7
30	Underdiagnosis of Hereditary Colorectal Cancers Among Medicare Patients: Genetic Testing Criteria for Lynch Syndrome Miss the Mark. JCO Precision Oncology, 2021, 5, 1103-1111.	1.5	7
31	What Is a Variant of Uncertain Significance in Genetic Testing?. European Urology Focus, 2022, 8, 654-656.	1.6	6
32	The Impact of Proband Indication for Genetic Testing on the Uptake of Cascade Testing Among Relatives. Frontiers in Genetics, 0, 13, .	1.1	6
33	Protein Phosphatase 2A. , 2010, , 1353-1365.		5
34	46,XY disorders of sex development and congenital diaphragmatic hernia: A case with dysmorphic facies, truncus arteriosus, bifid thymus, gut malrotation, rhizomelia, and adactyly. American Journal of Medical Genetics, Part A, 2015, 167, 1360-1364.	0.7	4
35	Secondary findings on virtual panels: opportunities, challenges, and potential for preventive medicine. Genetics in Medicine, 2019, 21, 1250-1251.	1.1	4
36	Universal Genetic Testing to Identify Pathogenic Germline Variants in Patients With Cancerâ€”Reply. JAMA Oncology, 2021, 7, 1071.	3.4	4

#	Article	IF	Citations
37	Comprehensive Genetic Testing for Pediatric Hypertrophic Cardiomyopathy Reveals Clinical Management Opportunities and Syndromic Conditions. Pediatric Cardiology, 2022, 43, 616-623.	0.6	4
38	Germline alterations among Hispanic men with prostate cancer. Prostate Cancer and Prostatic Diseases, 2022, 25, 561-567.	2.0	4
39	Patterns of germline and somatic testing after universal tumor screening for Lynch syndrome: A clinical practice survey of active members of the Collaborative Group of the Americas on Inherited Gastrointestinal Cancer. Journal of Genetic Counseling, 2022, 31, 949-955.	0.9	4
40	Conflicts of interest in genetic counseling: addressing and delivering. Genetics in Medicine, 2018, 20, 1094-1095.	1.1	3
41	Feasibility and Assessment of a Cascade Traceback Screening Program (FACTS): Protocol for a Multisite Study to Implement and Assess an Ovarian Cancer Traceback Cascade Testing Program. Journal of Personalized Medicine, 2021, 11, 543.	1.1	3
42	Clinical Impact of Pathogenic Variants in DNA Damage Repair Genes beyond BRCA1 and BRCA2 in Breast and Ovarian Cancer Patients. Cancers, 2022, 14, 2426.	1.7	3
43	Protein Phosphatase 2A. , 2003, , 405-415.		1
44	Risk categorization for oversight of laboratory-developed tests for inherited conditions: an updated position statement of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2020, 22, 983-985.	1.1	0
45	Addendum: American College of Medical Genetics guideline on the cytogenetic evaluation of the individual with developmental delay or mental retardation. Genetics in Medicine, 2020, 22, 2128-2128.	1.1	0
46	Prevalence of pathogenic germline cancer risk variants in testicular cancer patients: Identifying high risk groups. Urologic Oncology: Seminars and Original Investigations, 2022, , .	0.8	0