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	Comprehensive Genetic Testing for Pediatric Hypertrophic Cardiomyopathy Reveals Clinical Management Opportunities and Syndromic Conditions. Pediatric Cardiology, 2022, 43, 616-623.	0.6	4
2	Fumarate hydratase variant prevalence and manifestations among individuals receiving germline testing. Cancer, 2022, 128, 675-684.	2.0	11
3	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
4	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
5	Germline alterations among Hispanic men with prostate cancer. Prostate Cancer and Prostatic Diseases, 2022, 25, 561-567.	2.0	4
6	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
7	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
8	What Is a Variant of Uncertain Significance in Genetic Testing?. European Urology Focus, 2022, 8, 654-656.	1.6	6
9	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
10	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
11	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
12	Comparison of Universal Genetic Testing vs Guideline-Directed Targeted Testing for Patients With Hereditary Cancer Syndrome. JAMA Oncology, 2021, 7, 230.	3.4	146
13	Developing and Optimizing Innovative Tools to Address Familial Hypercholesterolemia Underdiagnosis. Circulation Genomic and Precision Medicine, 2021, 14, e003120.	1.6	23
14	Trans-ethnic variation in germline variants of patients with renal cell carcinoma. Cell Reports, 2021, 34, 108926.	2.9	16
15	Multigene Panel Testing in Individuals With Hepatocellular Carcinoma Identifies Pathogenic Germline Variants. JCO Precision Oncology, 2021, 5, 988-1000.	1.5	10
16	Clinical Implications of Pathogenic Germline Variants in Small Intestine Neuroendocrine Tumors (SI-NETs). JCO Precision Oncology, 2021, 5, 808-816.	1.5	7
17	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
18	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

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19	Universal Genetic Testing to Identify Pathogenic Germline Variants in Patients With Cancerâ€"Reply. JAMA Oncology, 2021, 7, 1071.	3.4	4
20	Limited-Variant Screening vs Comprehensive Genetic Testing for Familial Hypercholesterolemia Diagnosis. JAMA Cardiology, 2021, 6, 902.	3.0	14
21	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
22	Prevalence of pathogenic germline cancer risk variants in high-risk urothelial carcinoma. Genetics in Medicine, 2020, 22, 709-718.	1.1	44
23	Yield and Utility of Germline Testing Following Tumor Sequencing in Patients With Cancer. JAMA Network Open, 2020, 3, e2019452.	2.8	76
24	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
25	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
26	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
27	The Human Tumor Atlas Network: Charting Tumor Transitions across Space and Time at Single-Cell Resolution. Cell, 2020, 181, 236-249.	13.5	334
28	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
29	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
30	Prevalence of Germline Variants in Prostate Cancer and Implications for Current Genetic Testing Guidelines. JAMA Oncology, 2019, 5, 523.	3.4	240
31	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
32	Secondary findings on virtual panels: opportunities, challenges, and potential for preventive medicine. Genetics in Medicine, 2019, 21, 1250-1251.	1.1	4
33	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
34	Conflicts of interest in genetic counseling: addressing and delivering. Genetics in Medicine, 2018, 20, 1094-1095.	1.1	3
35	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
36	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

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37	Germline hemizygous deletion of CDKN2Aâ€“CDKN2B locus in a patient presenting with Liâ€“Fraumeni syndrome. Npj Genomic Medicine, 2016, 1, 16015.	1.7	9
38	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
39	RTTN Mutations Cause Primary Microcephaly and Primordial Dwarfism in Humans. American Journal of Human Genetics, 2015, 97, 862-868.	2.6	36
40	Personalized sequencing and the future of medicine: discovery, diagnosis and defeat of disease. Pharmacogenomics, 2014, 15, 1771-1790.	0.6	66
41	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
42	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
43	Expanding the phenotype of cardiovascular malformations in Adamsâ€“Oliver syndrome. American Journal of Medical Genetics, Part A, 2013, 161, 1386-1389.	0.7	15
44	Protein Phosphatase 2A. , 2010, , 1353-1365.		5
45	Protein Phosphatase 2A. , 2003, , 405-415.		1
46	The Impact of Proband Indication for Genetic Testing on the Uptake of Cascade Testing Among Relatives. Frontiers in Genetics, 0, 13, .	1.1	6