Ozge Ceyhan-Birsoy

List of Publications by Year
in descending order

31 papers	1,595 citations	430874 18 h-index	434195 31 g-index
33 all docs	33 docs citations	33 times ranked	2959 citing authors

#	Article	IF	Citations
1	ATM Germline-Mutated Gastroesophageal Junction Adenocarcinomas: Clinical Descriptors, Molecular Characteristics, and Potential Therapeutic Implications. Journal of the National Cancer Institute, 2022, 114, 761-770.	6.3	3
2	Early age of onset and broad cancer spectrum persist in MSH6- and PMS2-associated Lynch syndrome. Genetics in Medicine, 2022, 24, 1187-1195.	2.4	7
3	Cancer-Causative Mutations Occurring in Early Embryogenesis. Cancer Discovery, 2022, 12, 949-957.	9.4	21
4	Prospective pan-cancer germline testing using MSK-IMPACT informs clinical translation in 751 patients with pediatric solid tumors. Nature Cancer, 2021, 2, 357-365.	13.2	74
5	Prevalence and Characterization of Biallelic and Monoallelic NTHL1 and MSH3 Variant Carriers From a Pan-Cancer Patient Population. JCO Precision Oncology, 2021, 5, 455-465.	3.0	10
6	Discordant results between conventional newborn screening and genomic sequencing in the BabySeq Project. Genetics in Medicine, 2021, 23, 1372-1375.	2.4	47
7	Paired Tumor-Normal Sequencing Provides Insights into TP53-Related Cancer Spectrum in Li-Fraumeni Patients. Journal of the National Cancer Institute, 2021, , .	6.3	6
8	Psychosocial Effect of Newborn Genomic Sequencing on Families in the BabySeq Project. JAMA Pediatrics, 2021, 175, 1132.	6.2	35
9	A Comprehensive Comparison of Early-Onset and Average-Onset Colorectal Cancers. Journal of the National Cancer Institute, 2021, 113, 1683-1692.	6.3	66
10	Therapeutic Implications of Germline Testing in Patients With Advanced Cancers. Journal of Clinical Oncology, 2021, 39, 2698-2709.	1.6	83
11	Approaches to the comprehensive interpretation of genome-scale sequencing. , 2021, , 237-250.		0
12	Germline RAD51B variants confer susceptibility to breast and ovarian cancers deficient in homologous recombination. Npj Breast Cancer, 2021, 7, 135.	5.2	9
13	Germline Variants Identified in Patients with Early-onset Renal Cell Carcinoma Referred for Germline Genetic Testing. European Urology Oncology, 2021, 4, 993-1000.	5.4	16
14	The context-specific role of germline pathogenicity in tumorigenesis. Nature Genetics, 2021, 53, 1577-1585.	21.4	44
15	Fumarate hydrataseFHc.1431_1433dupAAA (p.Lys477dup) variant is not associated with cancer including renal cell carcinoma. Human Mutation, 2020, 41, 103-109.	2.5	25
16	Evolving Significance of Tumor-Normal Sequencing in Cancer Care. Trends in Cancer, 2020, 6, 31-39.	7.4	30
17	A Clinical Approach to Detecting Germline Pathogenic Variants From Tumor-Only Sequencing. JNCI Cancer Spectrum, 2020, 4, pkaa019.	2.9	5
18	Fumarate hydratase c.914TÂ>ÂC (p.Phe305Ser) is a pathogenic variant associated with hereditary leiomyomatosis and renal cell cancer syndrome. Molecular Genetics & Genomic Medicine, 2020, 8, e1293.	1.2	1

#	Article	IF	Citations
19	Quantifying Downstream Healthcare Utilization in Studies of Genomic Testing. Value in Health, 2020, 23, 559-565.	0.3	6
20	Tumour lineage shapes BRCA-mediated phenotypes. Nature, 2019, 571, 576-579.	27.8	295
21	Analyzing and Reanalyzing the Genome: Findings from the MedSeq Project. American Journal of Human Genetics, 2019, 105, 177-188.	6.2	38
22	The Landscape of Somatic Genetic Alterations in Breast Cancers from CHEK2 Germline Mutation Carriers. JNCI Cancer Spectrum, 2019, 3, pkz027.	2.9	20
23	Activating mutations in CSF1R and additional receptor tyrosine kinases in histiocytic neoplasms. Nature Medicine, 2019, 25, 1839-1842.	30.7	122
24	Interpretation of Genomic Sequencing Results in Healthy and Ill Newborns: Results from the BabySeq Project. American Journal of Human Genetics, 2019, 104, 76-93.	6.2	176
25	A synonymous germline variant PALB2 c.18G>T (p.Gly6=) disrupts normal splicing in a family with pancreatic and breast cancers. Breast Cancer Research and Treatment, 2019, 173, 79-86.	2.5	4
26	Prevalence of Germline Mutations in Cancer Susceptibility Genes in Patients With Advanced Renal Cell Carcinoma. JAMA Oncology, 2018, 4, 1228.	7.1	132
27	The BabySeq project: implementing genomic sequencing in newborns. BMC Pediatrics, 2018, 18, 225.	1.7	115
28	Reconciling newborn screening and a novel splice variant in BTD associated with partial biotinidase deficiency: a BabySeq Project case report. Journal of Physical Education and Sports Management, 2018, 4, a002873.	1.2	7
29	Germline SDHA mutations in children and adults with cancer. Journal of Physical Education and Sports Management, 2018, 4, a002584.	1.2	33
30	A curated gene list for reporting results of newborn genomic sequencing. Genetics in Medicine, 2017, 19, 809-818.	2.4	79
31	A systematic approach to the reporting of medically relevant findings from whole genome sequencing. BMC Medical Genetics, 2014, 15, 134.	2.1	84