Nicholas Pachter

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/6292691/publications.pdf

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20	686	932766 10	752256 20
papers	citations	h-index	g-index

20	20	20	1684
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Clinical Validity of Genes for Heritable Thoracic Aortic Aneurysm and Dissection. Journal of the American College of Cardiology, 2018, 72, 605-615.	1.2	190
2	Interim Results from the IMPACT Study: Evidence for Prostate-specific Antigen Screening in BRCA2 Mutation Carriers. European Urology, 2019, 76, 831-842.	0.9	148
3	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.	1.1	102
4	Identification of clinically actionable variants from genome sequencing of families with congenital heart disease. Genetics in Medicine, 2019, 21, 1111-1120.	1.1	54
5	A prospective prostate cancer screening programme for men with pathogenic variants in mismatch repair genes (IMPACT): initial results from an international prospective study. Lancet Oncology, The, 2021, 22, 1618-1631.	5.1	48
6	Clinical significance of circulating microRNAs as markers in detecting and predicting congenital heart defects in children. Journal of Translational Medicine, 2018, 16, 42.	1.8	34
7	Impact of Clinical Genetics Attendance at a Gynecologic Oncology Tumor Board on Referrals for Genetic Counseling and BRCA Mutation Testing. International Journal of Gynecological Cancer, 2016, 26, 892-897.	1.2	23
8	MCM complex members MCM3 and MCM7 are associated with a phenotypic spectrum from Meier-Gorlin syndrome to lipodystrophy and adrenal insufficiency. European Journal of Human Genetics, 2021, 29, 1110-1120.	1.4	16
9	Prostate-specific antigen velocity in a prospective prostate cancer screening study of men with genetic predisposition. British Journal of Cancer, 2018, 118, 266-276.	2.9	12
10	Current mismatch repair deficiency tumor testing practices and capabilities: A survey of Australian pathology providers. Asia-Pacific Journal of Clinical Oncology, 2018, 14, 417-425.	0.7	11
11	Comparing theory and non-theory based implementation approaches to improving referral practices in cancer genetics: a cluster randomised trial protocol. Trials, 2019, 20, 373.	0.7	10
12	Population-based estimates of breast cancer risk for carriers of pathogenic variants identified by gene-panel testing. Npj Breast Cancer, 2021, 7, 153.	2.3	10
13	Uptake of testing for germline BRCA mutations in patients with non-mucinous epithelial ovarian cancers in Western Australia: a comparison of different genetic counseling methods. International Journal of Gynecological Cancer, 2019, 29, 1038-1042.	1.2	8
14	A case of vascular Ehlers-Danlos Syndrome with a cardiomyopathy and multi-system involvement. Cardiovascular Pathology, 2018, 35, 48-51.	0.7	6
15	Study protocol of a multicentre cohort pilot study implementing an expanded preconception carrier-screening programme in metropolitan and regional Western Australia. BMJ Open, 2019, 9, e028209.	0.8	3
16	Implementing gene curation for hereditary cancer susceptibility in Australia: achieving consensus on genes with clinical utility. Journal of Medical Genetics, 2021, 58, 853-858.	1.5	3
17	TRACEBACK: Testing of Historical Tubo-Ovarian Cancer Patients for Hereditary Risk Genes as a Cancer Prevention Strategy in Family Members. Journal of Clinical Oncology, 2022, , JCO2102108.	0.8	3
18	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.	1.2	2

#	Article	IF	Citations
19	Lynch syndrome testing of colorectal cancer patients in a high-income country with universal healthcare: a retrospective study of current practice and gaps in seven australian hospitals. Hereditary Cancer in Clinical Practice, 2022, 20, 18.	0.6	2
20	Incidence of germline BRCA1/2 mutations in women with tubo-ovarian high-grade serous carcinomas with and without serous tubal intra-epithelial carcinomas. International Journal of Gynecological Cancer, 2020, 30, 94-99.	1.2	1