Tuomo M Mantere

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/8752952/publications.pdf

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22	1,403	687363 13	713466 21
papers	citations	h-index	g-index

25	25	25	3733
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Presence of Genetic Variants Among Young Men With Severe COVID-19. JAMA - Journal of the American Medical Association, 2020, 324, 663.	7.4	626
2	Long-Read Sequencing Emerging in Medical Genetics. Frontiers in Genetics, 2019, 10, 426.	2.3	290
3	Optical genome mapping enables constitutional chromosomal aberration detection. American Journal of Human Genetics, 2021, 108, 1409-1422.	6.2	108
4	Next-generation cytogenetics: Comprehensive assessment of 52 hematological malignancy genomes by optical genome mapping. American Journal of Human Genetics, 2021, 108, 1423-1435.	6.2	85
5	Primary Myocardial Fibrosis as an Alternative Phenotype Pathway of Inherited Cardiac Structural Disorders. Circulation, 2018, 137, 2716-2726.	1.6	41
6	Familial long-read sequencing increases yield of de novo mutations. American Journal of Human Genetics, 2022, 109, 631-646.	6.2	32
7	FANCM mutation c.5791C>T is a risk factor for triple-negative breast cancer in the Finnish population. Breast Cancer Research and Treatment, 2017, 166, 217-226.	2.5	26
8	Optical genome mapping identifies a germline retrotransposon insertion in <scp>SMARCB1</scp> in two siblings with atypical teratoid rhabdoid tumors. Journal of Pathology, 2021, 255, 202-211.	4.5	23
9	Targeted Next-Generation Sequencing Identifies a Recurrent Mutation in MCPH1 Associating with Hereditary Breast Cancer Susceptibility. PLoS Genetics, 2016, 12, e1005816.	3.5	22
10	Mutation of TP53, translocation analysis and immunohistochemical expression of MYC, BCL-2 and BCL-6 in patients with DLBCL treated with R-CHOP. Scientific Reports, 2018, 8, 14814.	3.3	21
11	Case-control analysis of truncating mutations in DNA damage response genes connects TEX15 and FANCD2 with hereditary breast cancer susceptibility. Scientific Reports, 2017, 7, 681.	3.3	20
12	Optical Genome Mapping: A Promising New Tool to Assess Genomic Complexity in Chronic Lymphocytic Leukemia (CLL). Cancers, 2022, 14, 3376.	3.7	18
13	Finnish Fanconi anemia mutations and hereditary predisposition to breast and prostate cancer. Clinical Genetics, 2015, 88, 68-73.	2.0	17
14	Rare missense mutations in RECQL and POLG associate with inherited predisposition to breast cancer. International Journal of Cancer, 2018, 142, 2286-2292.	5.1	15
15	FANCM c.5101C>T mutation associates with breast cancer survival and treatment outcome. International Journal of Cancer, 2016, 139, 2760-2770.	5.1	13
16	Tumor suppressor MCPH1 regulates gene expression profiles related to malignant conversion and chromosomal assembly. International Journal of Cancer, 2019, 145, 2070-2081.	5.1	5
17	Evaluating the role of NTHL1 p.Q90* allele in inherited breast cancer predisposition. Molecular Genetics & Genomic Medicine, 2020, 8, e1493.	1.2	5
18	Long-read technologies identify a hidden inverted duplication in a family with choroideremia. Human Genetics and Genomics Advances, 2021, 2, 100046.	1.7	4

#	Article	IF	Citations
19	Exploring the missing heritability in subjects with hearing loss, enlarged vestibular aqueducts, and a single or no pathogenic SLC26A4 variant. Human Genetics, 2022, 141, 465-484.	3.8	3
20	Mimicking BehÃ§etâ€™s disease: GMâ€CSF gain of function mutation in a family suffering from a BehÃ§etâ€™s diseaseâ€like disorder marked by extreme pathergy. Clinical and Experimental Immunology, 2021, 204, 189-198.	2.6	2
21	Truncating TINF2 p.Tyr312Ter variant and inherited breast cancer susceptibility. Familial Cancer, 2022, , .	1.9	1
22	P3823The role of genetics in apparently acquired non-ischemic cardiomyopathies leading to sudden cardiac death. European Heart Journal, 2018, 39, .	2.2	0