

Tuomo M Mantere

List of Publications by Year in descending order

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

Version: 2024-02-01

22
papers

1,403
citations

687363

13
h-index

713466

21
g-index

25
all docs

25
docs citations

25
times ranked

3733
citing authors

#	ARTICLE	IF	CITATIONS
1	Exploring the missing heritability in subjects with hearing loss, enlarged vestibular aqueducts, and a single or no pathogenic SLC26A4 variant. <i>Human Genetics</i> , 2022, 141, 465-484.	3.8	3
2	Familial long-read sequencing increases yield of de novo mutations. <i>American Journal of Human Genetics</i> , 2022, 109, 631-646.	6.2	32
3	Truncating TINF2 p.Tyr312Ter variant and inherited breast cancer susceptibility. <i>Familial Cancer</i> , 2022, , .	1.9	1
4	Optical Genome Mapping: A Promising New Tool to Assess Genomic Complexity in Chronic Lymphocytic Leukemia (CLL). <i>Cancers</i> , 2022, 14, 3376.	3.7	18
5	Mimicking Behçet's disease: GM-CSF gain of function mutation in a family suffering from a Behçet's disease-like disorder marked by extreme pathergy. <i>Clinical and Experimental Immunology</i> , 2021, 204, 189-198.	2.6	2
6	Optical genome mapping identifies a germline retrotransposon insertion in <i>SMARCB1</i> in two siblings with atypical teratoid rhabdoid tumors. <i>Journal of Pathology</i> , 2021, 255, 202-211.	4.5	23
7	Optical genome mapping enables constitutional chromosomal aberration detection. <i>American Journal of Human Genetics</i> , 2021, 108, 1409-1422.	6.2	108
8	Next-generation cytogenetics: Comprehensive assessment of 52 hematological malignancy genomes by optical genome mapping. <i>American Journal of Human Genetics</i> , 2021, 108, 1423-1435.	6.2	85
9	Long-read technologies identify a hidden inverted duplication in a family with choroideremia. <i>Human Genetics and Genomics Advances</i> , 2021, 2, 100046.	1.7	4
10	Presence of Genetic Variants Among Young Men With Severe COVID-19. <i>JAMA - Journal of the American Medical Association</i> , 2020, 324, 663.	7.4	626
11	Evaluating the role of <i>NTHL1</i> p.Q90* allele in inherited breast cancer predisposition. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1493.	1.2	5
12	Long-Read Sequencing Emerging in Medical Genetics. <i>Frontiers in Genetics</i> , 2019, 10, 426.	2.3	290
13	Tumor suppressor MCPH1 regulates gene expression profiles related to malignant conversion and chromosomal assembly. <i>International Journal of Cancer</i> , 2019, 145, 2070-2081.	5.1	5
14	Rare missense mutations in <i>RECQL</i> and <i>POLG</i> associate with inherited predisposition to breast cancer. <i>International Journal of Cancer</i> , 2018, 142, 2286-2292.	5.1	15
15	The role of genetics in apparently acquired non-ischemic cardiomyopathies leading to sudden cardiac death. <i>European Heart Journal</i> , 2018, 39, .	2.2	0
16	Mutation of TP53, translocation analysis and immunohistochemical expression of MYC, BCL-2 and BCL-6 in patients with DLBCL treated with R-CHOP. <i>Scientific Reports</i> , 2018, 8, 14814.	3.3	21
17	Primary Myocardial Fibrosis as an Alternative Phenotype Pathway of Inherited Cardiac Structural Disorders. <i>Circulation</i> , 2018, 137, 2716-2726.	1.6	41
18	Case-control analysis of truncating mutations in DNA damage response genes connects TEX15 and FANCD2 with hereditary breast cancer susceptibility. <i>Scientific Reports</i> , 2017, 7, 681.	3.3	20

#	ARTICLE	IF	CITATIONS
19	FANCM mutation c.5791C>T is a risk factor for triple-negative breast cancer in the Finnish population. <i>Breast Cancer Research and Treatment</i> , 2017, 166, 217-226.	2.5	26
20	Targeted Next-Generation Sequencing Identifies a Recurrent Mutation in MCPH1 Associating with Hereditary Breast Cancer Susceptibility. <i>PLoS Genetics</i> , 2016, 12, e1005816.	3.5	22
21	FANCM c.5101C>T mutation associates with breast cancer survival and treatment outcome. <i>International Journal of Cancer</i> , 2016, 139, 2760-2770.	5.1	13
22	Finnish Fanconi anemia mutations and hereditary predisposition to breast and prostate cancer. <i>Clinical Genetics</i> , 2015, 88, 68-73.	2.0	17