Marianthi Georgitsi

List of Publications by Year
in descending order

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50 papers	2,881 citations	304368 22 h-index	189595 50 g-index
51 all docs	51 docs citations	51 times ranked	2888 citing authors

#	Article	IF	Citations
1	Pituitary Adenoma Predisposition Caused by Germline Mutations in the AIP Gene. Science, 2006, 312, 1228-1230.	6.0	557
2	Haploinsufficiency for the erythroid transcription factor KLF1 causes hereditary persistence of fetal hemoglobin. Nature Genetics, 2010, 42, 801-805.	9.4	323
3	Clinical Characteristics and Therapeutic Responses in Patients with Germ-LineAIPMutations and Pituitary Adenomas: An International Collaborative Study. Journal of Clinical Endocrinology and Metabolism, 2010, 95, E373-E383.	1.8	323
4	Germline CDKN1B/p27Kip1 Mutation in Multiple Endocrine Neoplasia. Journal of Clinical Endocrinology and Metabolism, 2007, 92, 3321-3325.	1.8	262
5	Molecular diagnosis of pituitary adenoma predisposition caused by aryl hydrocarbon receptor-interacting protein gene mutations. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 4101-4105.	3.3	173
6	Systematic documentation and analysis of human genetic variation in hemoglobinopathies using the microattribution approach. Nature Genetics, 2011, 43, 295-301.	9.4	142
7	Evidence for association of an ACCN1 gene variant with response to lithium treatment in Sardinian patients with bipolar disorder. Pharmacogenomics, 2011, 12, 1559-1569.	0.6	82
8	Aryl hydrocarbon receptor interacting protein (AIP) gene mutation analysis in children and adolescents with sporadic pituitary adenomas. Clinical Endocrinology, 2008, 69, 621-627.	1.2	80
9	Mice with Inactivation of Aryl Hydrocarbon Receptor-Interacting Protein (Aip) Display Complete Penetrance of Pituitary Adenomas with Aberrant ARNT Expression. American Journal of Pathology, 2010, 177, 1969-1976.	1.9	78
10	Large Genomic Deletions inAIPin Pituitary Adenoma Predisposition. Journal of Clinical Endocrinology and Metabolism, 2008, 93, 4146-4151.	1.8	74
11	Mutation analysis of aryl hydrocarbon receptor interacting protein (AIP) gene in colorectal, breast, and prostate cancers. British Journal of Cancer, 2007, 96, 352-356.	2.9	56
12	MEN-4 and other multiple endocrine neoplasias due to cyclin-dependent kinase inhibitors (p27Kip1 and) Tj ETQq0 0 0 rgBT /Overlock 10 425-437.	2.2	52
13	No evidence of somatic aryl hydrocarbon receptor interacting protein mutations in sporadic endocrine neoplasia. Endocrine-Related Cancer, 2007, 14, 901-906.	1.6	48
14	Aggressive pituitary adenomas occurring in young patients in a large Polynesian kindred with a germline R271W mutation in the AIP gene. European Journal of Endocrinology, 2009, 161, 799-804.	1.9	45
15	The Genetic Etiology of Tourette Syndrome: Large-Scale Collaborative Efforts on the Precipice of Discovery. Frontiers in Neuroscience, 2016, 10, 351.	1.4	45
16	KLF10 gene expression is associated with high fetal hemoglobin levels and with response to hydroxyurea treatment in Î²-hemoglobinopathy patients. Pharmacogenomics, 2012, 13, 1487-1500.	0.6	37
17	European Multicentre Tics in Children Studies (EMTICS): protocol for two cohort studies to assess risk factors for tic onset and exacerbation in children and adolescents. European Child and Adolescent Psychiatry, 2019, 28, 91-109.	2.8	36
18	Population-specific documentation of pharmacogenomic markers and their allelic frequencies in FINDbase. Pharmacogenomics, 2011, 12, 49-58.	0.6	30

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19	Targeted Re-Sequencing Approach of Candidate Genes Implicates Rare Potentially Functional Variants in Tourette Syndrome Etiology. Frontiers in Neuroscience, 2016, 10, 428.	1.4	29
20	Mutation analysis of MEN1, HRPT2, CASR, CDKN1B, and AIP genes in primary hyperparathyroidism patients with features of genetic predisposition. Journal of Endocrinological Investigation, 2009, 32, 512-518.	1.8	28
21	Meta-Analysis of Tourette Syndrome and Attention Deficit Hyperactivity Disorder Provides Support for a Shared Genetic Basis. Frontiers in Neuroscience, 2016, 10, 340.	1.4	26
22	Genomic variation in the MAP3K5 gene is associated with Î²-thalassemia disease severity and hydroxyurea treatment efficacy. Pharmacogenomics, 2013, 14, 469-483.	0.6	25
23	FINDbase: a worldwide database for genetic variation allele frequencies updated. Nucleic Acids Research, 2011, 39, D926-D932.	6.5	22
24	A Single Nucleotide Polymorphism in theHBBP1Gene in the Human Î²-Globin Locus is Associated with a Mild Î²-Thalassemia Disease Phenotype. Hemoglobin, 2012, 36, 433-445.	0.4	22
25	Functional analysis of a novel KLF1 gene promoter variation associated with hereditary persistence of fetal hemoglobin. Annals of Hematology, 2013, 92, 53-58.	0.8	22
26	Genetic recombination as a major cause of mutagenesis in the human globin gene clusters. Clinical Biochemistry, 2009, 42, 1839-1850.	0.8	21
27	6-mercaptopurine influences TPMT gene transcription in a TPMT gene promoter variable number of tandem repeats-dependent manner. Pharmacogenomics, 2012, 13, 283-295.	0.6	21
28	Variant Ranker: a web-tool to rank genomic data according to functional significance. BMC Bioinformatics, 2017, 18, 341.	1.2	21
29	Association of ALOX12 gene polymorphism with all-cause and cardiovascular mortality in diabetic nephropathy. International Urology and Nephrology, 2018, 50, 321-329.	0.6	20
30	Transcriptional regulation and pharmacogenomics. Pharmacogenomics, 2011, 12, 655-673.	0.6	16
31	Antibodies to neuronal surface proteins in Tourette Syndrome: Lack of evidence in a European paediatric cohort. Brain, Behavior, and Immunity, 2019, 81, 665-669.	2.0	15
32	Antiâ€dopamine D2 receptor antibodies in chronic tic disorders. Developmental Medicine and Child Neurology, 2020, 62, 1205-1212.	1.1	15
33	Oxidative Stress Genes in Diabetes Mellitus Type 2: Association with Diabetic Kidney Disease. Oxidative Medicine and Cellular Longevity, 2021, 2021, 1-10.	1.9	15
34	Association of rs11780592 Polymorphism in the Human Soluble Epoxide Hydrolase Gene (EPHX2) with Oxidized LDL and Mortality in Patients with Diabetic Chronic Kidney Disease. Oxidative Medicine and Cellular Longevity, 2021, 2021, 1-8.	1.9	13
35	Aryl hydrocarbon receptor interacting protein mutations seem not to associate with familial non-medullary thyroid cancer. Journal of Endocrinological Investigation, 2009, 32, 426-429.	1.8	12
36	Genetics of Myasthenia Gravis: A Case-Control Association Study in the Hellenic Population. Clinical and Developmental Immunology, 2012, 2012, 1-7.	3.3	12

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37	Population-ethnic group specific genome variation allele frequency data: A querying and visualization journey. Genomics, 2012, 100, 93-101.	1.3	12
38	The Polygenic Nature and Complex Genetic Architecture of Specific Learning Disorder. Brain Sciences, 2021, 11, 631.	1.1	12
39	Impact of ACE and ApoE polymorphisms on myocardial perfusion: correlation with myocardial single photon emission computed tomographic imaging. Journal of Human Genetics, 2009, 54, 595-602.	1.1	10
40	Clinical Profiles and Socio-Demographic Characteristics of Adults with Specific Learning Disorder in Northern Greece. Brain Sciences, 2021, 11, 602.	1.1	8
41	The Genetics of Gilles de la Tourette Syndrome: a Common Aetiological Basis with Comorbid Disorders?. Current Behavioral Neuroscience Reports, 2016, 3, 218-231.	0.6	7
42	Assessment of association between lipoxygenase genes variants in elderly Greek population and type 2 diabetes mellitus. Diabetes and Vascular Disease Research, 2018, 15, 340-343.	0.9	7
43	Association between CUBN gene variants, type 2 diabetes and vitamin D concentrations in an elderly Greek population. Journal of Steroid Biochemistry and Molecular Biology, 2020, 198, 105549.	1.2	7
44	Genetic variation in CARD8, a gene coding for an NLRP3 inflammasome-associated protein, alters the genetic risk for diabetic nephropathy in the context of type 2 diabetes mellitus. Diabetes and Vascular Disease Research, 2020, 17, 147916412097089.	0.9	6
45	Myasthenia gravis genome-wide association study implicates AGRN as a risk locus. Journal of Medical Genetics, 2022, 59, 801-809.	1.5	5
46	Genetic Databases in Pharmacogenomics: The Frequency of Inherited Disorders Database (FINDbase). Methods in Molecular Biology, 2013, 1015, 321-336.	0.4	3
47	First Report of Hb A₂-NYU (HBD:c.39T>A) in the Hellenic Population. Hemoglobin, 2011, 35, 91-95.	0.4	1
48	Lithium-induced differential expression of SAT1 in suicide completers and controls is not correlated with polymorphisms in the promoter region of the gene. Psychiatry Research, 2014, 220, 1167-1168.	1.7	1
49	Neuropathology-driven Whole-genome Sequencing Study Points to Novel Candidate Genes for Healthy Brain Aging. Alzheimer Disease and Associated Disorders, 2019, 33, 7-14.	0.6	1
50	Variants in clock genes could be associated with lower risk of type 2 diabetes in an elderly Greek population. Maturitas, 2021, 152, 20-25.	1.0	1