Eleni Giannoulatou

List of Publications by Year
in descending order

85	10,116	126907	58581
papers	citations	33 h-index	82 g-index

91	91	91	22579
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Genetic risk and a primary role for cell-mediated immune mechanisms in multiple sclerosis. Nature, 2011, 476, 214-219.	27.8	2,400
2	Genome-wide association analysis identifies 13 new risk loci for schizophrenia. Nature Genetics, 2013, 45, 1150-1159.	21.4	1,395
3	Identification of 15 new psoriasis susceptibility loci highlights the role of innate immunity. Nature Genetics, 2012, 44, 1341-1348.	21.4	848
4	Genome-wide association study of CNVs in 16,000 cases of eight common diseases and 3,000 shared controls. Nature, 2010, 464, 713-720.	27.8	737
5	Genomic Dissection of Bipolar Disorder and Schizophrenia, Including 28 Subphenotypes. Cell, 2018, 173, 1705-1715.e16.	28.9	623
6	Analysis of hundreds of cis-regulatory landscapes at high resolution in a single, high-throughput experiment. Nature Genetics, 2014, 46, 205-212.	21.4	417
7	Epidemiology and treatment of pulmonary arterial hypertension. Nature Reviews Cardiology, 2017, 14, 603-614.	13.7	310
8	Myelodysplastic Syndromes Are Propagated by Rare and Distinct Human Cancer Stem Cells InÂVivo. Cancer Cell, 2014, 25, 794-808.	16.8	272
9	Interferon-induced transmembrane protein-3 genetic variant rs12252-C is associated with severe influenza in Chinese individuals. Nature Communications, 2013, 4, 1418.	12.8	228
10	NAD Deficiency, Congenital Malformations, and Niacin Supplementation. New England Journal of Medicine, 2017, 377, 544-552.	27.0	177
11	Smchd1-Dependent and -Independent Pathways Determine Developmental Dynamics of CpG Island Methylation on the Inactive X Chromosome. Developmental Cell, 2012, 23, 265-279.	7.0	160
12	Early dynamic fate changes in haemogenic endothelium characterized at the single-cell level. Nature Communications, 2013, 4, 2924.	12.8	158
13	Genome-Wide Association Study Implicates HLA-C*01:02 as a Risk Factor at the Major Histocompatibility Complex Locus in Schizophrenia. Biological Psychiatry, 2012, 72, 620-628.	1.3	156
14	Male-lineage transmission of an acquired metabolic phenotype induced by grand-paternal obesity. Molecular Metabolism, 2016, 5, 699-708.	6.5	154
15	Association of the PHACTR1/EDN1 Genetic Locus With Spontaneous Coronary Artery Dissection. Journal of the American College of Cardiology, 2019, 73, 58-66.	2.8	147
16	Pneumococcal genome sequencing tracks a vaccine escape variant formed through a multi-fragment recombination event. Nature Genetics, 2012, 44, 352-355.	21.4	144
17	Advances in the Genetics of Congenital HeartÂDisease. Journal of the American College of Cardiology, 2017, 69, 859-870.	2.8	115
18	Common variants in the HLA-DRB1â€“HLA-DQA1 HLA class II region are associated with susceptibility to visceral leishmaniasis. Nature Genetics, 2013, 45, 208-213.	21.4	86

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19	Targeted Next-Generation Sequencing Identifies Pathogenic Variants in Familial Congenital Heart Disease. Journal of the American College of Cardiology, 2014, 64, 2498-2506.	2.8	85
20	Distinct patterns of hepcidin and iron regulation during HIV-1, HBV, and HCV infections. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 12187-12192.	7.1	79
21	Contributions of intrinsic mutation rate and selfish selection to levels of de novo HRAS mutations in the paternal germline. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 20152-20157.	7.1	70
22	Cellular interference in craniofrontonasal syndrome: males mosaic for mutations in the X-linked EFNB1 gene are more severely affected than true hemizygotes. Human Molecular Genetics, 2013, 22, 1654-1662.	2.9	66
23	Host and microbiome multi-omics integration: applications and methodologies. Biophysical Reviews, 2019, 11, 55-65.	3.2	66
24	GenoSNP: a variational Bayes within-sample SNP genotyping algorithm that does not require a reference population. Bioinformatics, 2008, 24, 2209-2214.	4.1	65
25	A Screening Approach to Identify Clinically Actionable Variants Causing Congenital Heart Disease in Exome Data. Circulation Genomic and Precision Medicine, 2018, 11, e001978.	3.6	65
26	Verification and validation of bioinformatics software without a gold standard: a case study of BWA and Bowtie. BMC Bioinformatics, 2014, 15, S15.	2.6	58
27	Selfish mutations dysregulating RAS-MAPK signaling are pervasive in aged human testes. Genome Research, 2018, 28, 1779-1790.	5.5	56
28	Identification of clinically actionable variants from genome sequencing of families with congenital heart disease. Genetics in Medicine, 2019, 21, 1111-1120.	2.4	54
29	Visualizing the origins of selfish de novo mutations in individual seminiferous tubules of human testes. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, 2454-2459.	7.1	45
30	Spontaneous Coronary Artery Dissection. Circulation Genomic and Precision Medicine, 2020, 13, e003030.	3.6	43
31	Polymorphism in a lincRNA Associates with a Doubled Risk of Pneumococcal Bacteremia in Kenyan Children. American Journal of Human Genetics, 2016, 98, 1092-1100.	6.2	39
32	Antiviral activity of bone morphogenetic proteins and activins. Nature Microbiology, 2019, 4, 339-351.	13.3	39
33	Rapidly Escalating Hepcidin and Associated Serum Iron Starvation Are Features of the Acute Response to Typhoid Infection in Humans. PLoS Neglected Tropical Diseases, 2015, 9, e0004029.	3.0	38
34	An inherited duplication at the gene p21 Protein-Activated Kinase 7 (PAK7) is a risk factor for psychosis. Human Molecular Genetics, 2014, 23, 3316-3326.	2.9	37
35	Evaluation of the immunogenicity and impact on the latent HIVâ€1 reservoir of a conserved region vaccine, MVA.HIVconsv, in antiretroviral therapyâ€treated subjects. Journal of the International AIDS Society, 2017, 20, 21171.	3.0	36
36	Multi-omic profiling reveals associations between the gut mucosal microbiome, the metabolome, and host DNA methylation associated gene expression in patients with colorectal cancer. BMC Microbiology, 2020, 20, 83.	3.3	36

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37	Whole-genome sequencing of spermatocytic tumors provides insights into the mutational processes operating in the male germline. PLoS ONE, 2017, 12, e0178169.	2.5	36
38	Binding of transcription factor GabR to DNA requires recognition of DNA shape at a location distinct from its cognate binding site. Nucleic Acids Research, 2016, 44, 1411-1420.	14.5	35
39	Functional genomics and gene-environment interaction highlight the complexity of congenital heart disease caused by Notch pathway variants. Human Molecular Genetics, 2020, 29, 566-579.	2.9	32
40	TP53 mutations and protein immunopositivity may predict for poor outcome but also for trastuzumab benefit in patients with early breast cancer treated in the adjuvant setting. Oncotarget, 2016, 7, 32731-32753.	1.8	30
41	Isogenic mice exhibit sexually-dimorphic DNA methylation patterns across multiple tissues. BMC Genomics, 2017, 18, 966.	2.8	26
42	Survival of Idiopathic Pulmonary Arterial Hypertension Patients in the Modern Era in Australia and New Zealand. Heart Lung and Circulation, 2018, 27, 1368-1375.	0.4	26
43	Functional characterization of a novel PBX1 de novo missense variant identified in a patient with syndromic congenital heart disease. Human Molecular Genetics, 2020, 29, 1068-1082.	2.9	26
44	A gene-centric strategy for identifying disease-causing rare variants in dilated cardiomyopathy. Genetics in Medicine, 2019, 21, 133-143.	2.4	25
45	A Method to Exploit the Structure of Genetic Ancestry Space to Enhance Case-Control Studies. American Journal of Human Genetics, 2016, 98, 857-868.	6.2	21
46	Genetic burden and associations with adverse neurodevelopment in neonates with congenital heart disease. American Heart Journal, 2018, 201, 33-39.	2.7	19
47	Maternal iron deficiency perturbs embryonic cardiovascular development in mice. Nature Communications, 2021, 12, 3447.	12.8	17
48	How to test bioinformatics software?. Biophysical Reviews, 2015, 7, 343-352.	3.2	16
49	Effects of TP53 and PIK3CA mutations in early breast cancer: a matter of co-mutation and tumor-infiltrating lymphocytes. Breast Cancer Research and Treatment, 2016, 158, 307-321.	2.5	16
50	Dynamics of Transforming Growth Factor (TGF)-Î² Superfamily Cytokine Induction During HIV-1 Infection Are Distinct From Other Innate Cytokines. Frontiers in Immunology, 2020, 11, 596841.	4.8	15
51	Spontaneous Coronary Artery Dissection and Fibromuscular Dysplasia: Vasculopathies With a Predilection for Women. Heart Lung and Circulation, 2021, 30, 27-35.	0.4	15
52	Heterozygous loss of WBP11 function causes multiple congenital defects in humans and mice. Human Molecular Genetics, 2021, 29, 3662-3678.	2.9	14
53	The miR-200 family is increased in dysplastic lesions in ulcerative colitis patients. PLoS ONE, 2017, 12, e0173664.	2.5	14
54	Exploring the Genetic Architecture of Spontaneous Coronary Artery Dissection Using Whole-Genome Sequencing. Circulation Genomic and Precision Medicine, 2022, 15, 101161CIRCGEN121003527.	3.6	14

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55	Prevalent somatic BRCA1 mutations shape clinically relevant genomic patterns of nasopharyngeal carcinoma in Southeast Europe. International Journal of Cancer, 2018, 142, 66-80.	5.1	13
56	Opposite Prognostic Impact of Single PTEN-loss andPIK3CAMutations in Early High-risk Breast Cancer. Cancer Genomics and Proteomics, 2019, 16, 195-206.	2.0	13
57	Genetic variation in VAC14 is associated with bacteremia secondary to diverse pathogens in African children. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, E3601-E3603.	7.1	12
58	Separation of Dual Oxidase 2 and Lactoperoxidase Expression in Intestinal Crypts and Species Differences May Limit Hydrogen Peroxide Scavenging During Mucosal Healing in Mice and Humans. Inflammatory Bowel Diseases, 2018, 24, 136-148.	1.9	11
59	Big data: the elements of good questions, open data, and powerful software. Biophysical Reviews, 2019, 11, 1-3.	3.2	11
60	Is There an Independent Role of TERT and NF1 in High Grade Gliomas?. Translational Oncology, 2020, 13, 346-354.	3.7	11
61	VPOT: A Customizable Variant Prioritization Ordering Tool for Annotated Variants. Genomics, Proteomics and Bioinformatics, 2019, 17, 540-545.	6.9	10
62	Whole genome sequencing in transposition of the great arteries and associations with clinically relevant heart, brain and laterality genes. American Heart Journal, 2022, 244, 1-13.	2.7	10
63	Familial adenomatous patients with desmoid tumours show increased expression of miR-34a in serum and high levels in tumours. Oncoscience, 2016, 3, 173-185.	2.2	9
64	SVPV: a structural variant prediction viewer for paired-end sequencing datasets. Bioinformatics, 2017, 33, 2032-2033.	4.1	9
65	Comparison of somatic variant detection algorithms using Ion Torrent targeted deep sequencing data. BMC Medical Genomics, 2019, 12, 181.	1.5	9
66	dv-trio: a family-based variant calling pipeline using DeepVariant. Bioinformatics, 2020, 36, 3549-3551.	4.1	9
67	Tumor Infiltrating Lymphocytes Affect the Outcome of Patients with Operable Triple-Negative Breast Cancer in Combination with Mutated Amino Acid Classes. PLoS ONE, 2016, 11, e0163138.	2.5	8
68	Relapsed and De Novo Metastatic HER2-positive Breast Cancer Treated With Trastuzumab: Tumor Genotypes and Clinical Measures Associated With Patient Outcome. Clinical Breast Cancer, 2019, 19, 113-125.e4.	2.4	8
69	Transposon clusters as substrates for aberrant splice-site activation. RNA Biology, 2021, 18, 354-367.	3.1	8
70	Spliceogen: an integrative, scalable tool for the discovery of splice-altering variants. Bioinformatics, 2019, 35, 4405-4407.	4.1	7
71	Pathogenic BRCA1 mutations may be necessary but not sufficient for tissue genomic heterogeneity: Deep sequencing data from ovarian cancer patients. Gynecologic Oncology, 2019, 152, 375-386.	1.4	7
72	Phase II study of panitumumab combined with capecitabine and oxaliplatin as first-line treatment in metastatic colorectal cancer patients: clinical results including extended tumor genotyping. Medical Oncology, 2018, 35, 101.	2.5	6

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73	Genotype-phenotype associations in colorectal adenocarcinomas and their matched metastases. Human Pathology, 2021, 107, 104-116.	2.0	6
74	Benchmarking the Effectiveness and Accuracy of Multiple Mitochondrial DNA Variant Callers: Practical Implications for Clinical Application. Frontiers in Genetics, 2022, 13, 692257.	2.3	6
75	Disease evolution and heterogeneity in bilateral breast cancer. American Journal of Cancer Research, 2016, 6, 2611-2630.	1.4	5
76	A cloud-based framework for applying metamorphic testing to a bioinformatics pipeline. , 2016, , .		4
77	Prognostic Biomarkers in Early-stage Gastric Adenocarcinoma Treated With Adjuvant Chemoradiotherapy. Cancer Genomics and Proteomics, 2020, 17, 277-290.	2.0	4
78	CHDgene: A Curated Database for Congenital Heart Disease Genes. Circulation Genomic and Precision Medicine, 2022, 15, 101161CIRCGEN121003539.	3.6	4
79	Populationâ€based identityâ€byâ€descent mapping combined with exome sequencing to detect rare risk variants for schizophrenia. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2019, 180, 223-231.	1.7	2
80	Tumor Mutational Patterns and Infiltrating Lymphocyte Density in Young and Elderly Patients With Breast Cancer. Cancer Genomics and Proteomics, 2020, 17, 181-193.	2.0	2
81	Genotyping data of routinely processed matched primary/metastatic tumor samples. Data in Brief, 2021, 34, 106646.	1.0	2
82	Tumor Genotyping and Homologous Recombination Repair Gene Variants in Patients With Epithelial Ovarian Cancer: Is Pathogenic Enough?. Frontiers in Oncology, 2021, 11, 683057.	2.8	1
83	Pathogenic mutations and overall survival in 3,084 patients with cancer: the Hellenic Cooperative Oncology Group Precision Medicine Initiative. Oncotarget, 2020, 11, 1-14.	1.8	1
84	Myelodysplastic Syndromes Are Propagated by Rare and Distinct Human Cancer Stem Cells InÂVivo. Cancer Cell, 2015, 27, 603-605.	16.8	0
85	Decoding the complex genetic causes of heart diseases using systems biology. Biophysical Reviews, 2015, 7, 141-159.	3.2	0