Eleni Giannoulatou

List of Publications by Year in Descending Order

83 papers	7,608 citations	30 h-index	87 g-index
91 ext. papers	9,207 ext. citations	10.7 avg, IF	5 L-index

#	Paper	IF	Citations
83	Benchmarking the Effectiveness and Accuracy of Multiple Mitochondrial DNA Variant Callers: Practical Implications for Clinical Application.. *Frontiers in Genetics*, **2022**, 13, 692257	4.5	1
82	Congenital Heart Disease Gene: a Curated Database for Congenital Heart Disease Genes.. *Circulation Genomic and Precision Medicine*, **2022**, 101161CIRCGEN121003539	5.2	
81	Exploring the Genetic Architecture of Spontaneous Coronary Artery Dissection Using Whole-Genome Sequencing.. *Circulation Genomic and Precision Medicine*, **2022**, 101161CIRCGEN121003527	5.2	1
80	Whole genome sequencing in transposition of the great arteries and associations with clinically relevant heart, brain and laterality genes. *American Heart Journal*, **2021**, 244, 1-13	4.9	1
79	Maternal iron deficiency perturbs embryonic cardiovascular development in mice. *Nature Communications*, **2021**, 12, 3447	17.4	5
78	Tumor Genotyping and Homologous Recombination Repair Gene Variants in Patients With Epithelial Ovarian Cancer: Is Pathogenic Enough?. *Frontiers in Oncology*, **2021**, 11, 683057	5.3	
77	Spontaneous Coronary Artery Dissection and Fibromuscular Dysplasia: Vasculopathies With a Predilection for Women. *Heart Lung and Circulation*, **2021**, 30, 27-35	1.8	1
76	Genotype-phenotype associations in colorectal adenocarcinomas and their matched metastases. *Human Pathology*, **2021**, 107, 104-116	3.7	2
75	Transposon clusters as substrates for aberrant splice-site activation. *RNA Biology*, **2021**, 18, 354-367	4.8	4
74	Genotyping data of routinely processed matched primary/metastatic tumor samples. *Data in Brief*, **2021**, 34, 106646	1.2	1
73	dv-trio: a family-based variant calling pipeline using DeepVariant. *Bioinformatics*, **2020**, 36, 3549-3551	7.2	4
72	Tumor Mutational Patterns and Infiltrating Lymphocyte Density in Young and Elderly Patients With Breast Cancer. *Cancer Genomics and Proteomics*, **2020**, 17, 181-193	3.3	2
71	Prognostic Biomarkers in Early-stage Gastric Adenocarcinoma Treated With Adjuvant Chemoradiotherapy. *Cancer Genomics and Proteomics*, **2020**, 17, 277-290	3.3	1
70	Pathogenic mutations and overall survival in 3,084 patients with cancer: the Hellenic Cooperative Oncology Group Precision Medicine Initiative. *Oncotarget*, **2020**, 11, 1-14	3.3	0
69	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	5.6	13
68	Is There an Independent Role of TERT and NF1 in High Grade Gliomas?. *Translational Oncology*, **2020**, 13, 346-354	4.9	7
67	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	5.6	16

66	Dynamics of Transforming Growth Factor (TGF)-β Superfamily Cytokine Induction During HIV-1 Infection Are Distinct From Other Innate Cytokines. *Frontiers in Immunology*, **2020**, 11, 596841	8.4	3
65	Spontaneous Coronary Artery Dissection: Insights on Rare Genetic Variation From Genome Sequencing. *Circulation Genomic and Precision Medicine*, **2020**, 13, e003030	5.2	14
64	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	4.5	11
63	Heterozygous loss of WBP11 function causes multiple congenital defects in humans and mice. *Human Molecular Genetics*, **2020**, 29, 3662-3678	5.6	3
62	Host and microbiome multi-omics integration: applications and methodologies. *Biophysical Reviews*, **2019**, 11, 55-65	3.7	42
61	Spliceogen: an integrative, scalable tool for the discovery of splice-altering variants. *Bioinformatics*, **2019**, 35, 4405-4407	7.2	3
60	Opposite Prognostic Impact of Single PTEN-loss and Mutations in Early High-risk Breast Cancer. *Cancer Genomics and Proteomics*, **2019**, 16, 195-206	3.3	7
59	Population-based identity-by-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	3.5	2
58	A gene-centric strategy for identifying disease-causing rare variants in dilated cardiomyopathy. *Genetics in Medicine*, **2019**, 21, 133-143	8.1	20
57	VPOT: A Customizable Variant Prioritization Ordering Tool for Annotated Variants. *Genomics, Proteomics and Bioinformatics*, **2019**, 17, 540-545	6.5	2
56	Comparison of somatic variant detection algorithms using Ion Torrent targeted deep sequencing data. *BMC Medical Genomics*, **2019**, 12, 181	3.7	3
55	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	3	5
54	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	4.9	6
53	Association of the PHACTR1/EDN1 Genetic Locus With Spontaneous Coronary Artery Dissection. *Journal of the American College of Cardiology*, **2019**, 73, 58-66	15.1	86
52	Antiviral activity of bone morphogenetic proteins and activins. *Nature Microbiology*, **2019**, 4, 339-351	26.6	19
51	Identification of clinically actionable variants from genome sequencing of families with congenital heart disease. *Genetics in Medicine*, **2019**, 21, 1111-1120	8.1	25
50	Genetic burden and associations with adverse neurodevelopment in neonates with congenital heart disease. *American Heart Journal*, **2018**, 201, 33-39	4.9	15
49	Genetic variation in 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	11.5	8

48	A Screening Approach to Identify Clinically Actionable Variants Causing Congenital Heart Disease in Exome Data. *Circulation Genomic and Precision Medicine*, **2018**, 11, e001978	5.2	37
47	Prevalent somatic BRCA1 mutations shape clinically relevant genomic patterns of nasopharyngeal carcinoma in Southeast Europe. *International Journal of Cancer*, **2018**, 142, 66-80	7.5	11
46	Survival of Idiopathic Pulmonary Arterial Hypertension Patients in the Modern Era in Australia and New Zealand. *Heart Lung and Circulation*, **2018**, 27, 1368-1375	1.8	17
45	Genomic Dissection of Bipolar Disorder and Schizophrenia, Including 28 Subphenotypes. *Cell*, **2018**, 173, 1705-1715.e16	56.2	360
44	Selfish mutations dysregulating RAS-MAPK signaling are pervasive in aged human testes. *Genome Research*, **2018**, 28, 1779-1790	9.7	36
43	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	3.7	6
42	Advances in the Genetics of Congenital Heart Disease: A Clinician's Guide. *Journal of the American College of Cardiology*, **2017**, 69, 859-870	15.1	76
41	SVPV: a structural variant prediction viewer for paired-end sequencing datasets. *Bioinformatics*, **2017**, 33, 2032-2033	7.2	7
40	Epidemiology and treatment of pulmonary arterial hypertension. *Nature Reviews Cardiology*, **2017**, 14, 603-614	14.8	182
39	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*, **2017**, 24, 136-148	4.5	9
38	NAD Deficiency, Congenital Malformations, and Niacin Supplementation. *New England Journal of Medicine*, **2017**, 377, 544-552	59.2	114
37	Isogenic mice exhibit sexually-dimorphic DNA methylation patterns across multiple tissues. *BMC Genomics*, **2017**, 18, 966	4.5	21
36	Evaluation of the immunogenicity and impact on the latent HIV-1 reservoir of a conserved region vaccine, MVA.HIVconsv, in antiretroviral therapy-treated subjects. *Journal of the International AIDS Society*, **2017**, 20, 21171	5.4	26
35	The miR-200 family is increased in dysplastic lesions in ulcerative colitis patients. *PLoS ONE*, **2017**, 12, e0173664	3.7	10
34	Whole-genome sequencing of spermatocytic tumors provides insights into the mutational processes operating in the male germline. *PLoS ONE*, **2017**, 12, e0178169	3.7	24
33	A cloud-based framework for applying metamorphic testing to a bioinformatics pipeline **2016**,		3
32	Male-lineage transmission of an acquired metabolic phenotype induced by grand-paternal obesity. *Molecular Metabolism*, **2016**, 5, 699-708	8.8	104
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	11	30

30	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, 2454-9	11.5	34
29	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, 1411-20	20.1	27
28	Disease evolution and heterogeneity in bilateral breast cancer. *American Journal of Cancer Research* , **2016**, 6, 2611-2630	4.4	4
27	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, 32731-53	3.3	25
26	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	3.7	8
25	Familial adenomatous patients with desmoid tumours show increased expression of miR-34a in serum and high levels in tumours. *Oncoscience*, **2016**, 3, 173-85	0.8	8
24	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, 307-21	4.4	12
23	A Method to Exploit the Structure of Genetic Ancestry Space to Enhance Case-Control Studies. *American Journal of Human Genetics*, **2016**, 98, 857-868	11	14
22	Decoding the complex genetic causes of heart diseases using systems biology. *Biophysical Reviews*, **2015**, 7, 141-159	3.7	
21	How to test bioinformatics software?. *Biophysical Reviews*, **2015**, 7, 343-352	3.7	12
20	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	4.8	25
19	Myelodysplastic syndromes are propagated by rare and distinct human cancer stem cells in vivo. *Cancer Cell*, **2014**, 25, 794-808	24.3	216
18	Analysis of hundreds of cis-regulatory landscapes at high resolution in a single, high-throughput experiment. *Nature Genetics*, **2014**, 46, 205-12	36.3	331
17	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, 12187-92	11.5	56
16	Verification and validation of bioinformatics software without a gold standard: a case study of BWA and Bowtie. *BMC Bioinformatics*, **2014**, 15 Suppl 16, S15	3.6	40
15	Targeted next-generation sequencing identifies pathogenic variants in familial congenital heart disease. *Journal of the American College of Cardiology*, **2014**, 64, 2498-506	15.1	60
14	An inherited duplication at the gene p21 Protein-Activated Kinase 7 (PAK7) is a risk factor for psychosis. *Human Molecular Genetics*, **2014**, 23, 3316-26	5.6	32
13	Genome-wide association analysis identifies 13 new risk loci for schizophrenia. *Nature Genetics*, **2013**, 45, 1150-9	36.3	1153

12	Common variants in the HLA-DRB1-HLA-DQA1 HLA class II region are associated with susceptibility to visceral leishmaniasis. *Nature Genetics*, **2013**, 45, 208-13	36.3	76
11	Early dynamic fate changes in haemogenic endothelium characterized at the single-cell level. *Nature Communications*, **2013**, 4, 2924	17.4	124
10	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, 20152-7	11.5	51
9	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, 1654-62	5.6	50
8	Interferon-induced transmembrane protein-3 genetic variant rs12252-C is associated with severe influenza in Chinese individuals. *Nature Communications*, **2013**, 4, 1418	17.4	174
7	Pneumococcal genome sequencing tracks a vaccine escape variant formed through a multi-fragment recombination event. *Nature Genetics*, **2012**, 44, 352-5	36.3	82
6	Identification of 15 new psoriasis susceptibility loci highlights the role of innate immunity. *Nature Genetics*, **2012**, 44, 1341-8	36.3	681
5	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, 620-8	7.9	130
4	Smchd1-dependent and -independent pathways determine developmental dynamics of CpG island methylation on the inactive X chromosome. *Developmental Cell*, **2012**, 23, 265-79	10.2	128
3	Genetic risk and a primary role for cell-mediated immune mechanisms in multiple sclerosis. *Nature*, **2011**, 476, 214-9	50.4	1948
2	Genome-wide association study of CNVs in 16,000 cases of eight common diseases and 3,000 shared controls. *Nature*, **2010**, 464, 713-20	50.4	639
1	GenoSNP: a variational Bayes within-sample SNP genotyping algorithm that does not require a reference population. *Bioinformatics*, **2008**, 24, 2209-14	7.2	56