

# Eleni Giannoulatou

## List of Publications by Year in descending order

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Version: 2024-02-01

85  
papers

10,116  
citations

126907

33  
h-index

58581

82  
g-index

91  
all docs

91  
docs citations

91  
times ranked

22579  
citing authors

#	ARTICLE	IF	CITATIONS
1	Whole genome sequencing in transposition of the great arteries and associations with clinically relevant heart, brain and laterality genes. <i>American Heart Journal</i> , 2022, 244, 1-13.	2.7	10
2	Benchmarking the Effectiveness and Accuracy of Multiple Mitochondrial DNA Variant Callers: Practical Implications for Clinical Application. <i>Frontiers in Genetics</i> , 2022, 13, 692257.	2.3	6
3	CHDgene: A Curated Database for Congenital Heart Disease Genes. <i>Circulation Genomic and Precision Medicine</i> , 2022, 15, 101161CIRCGEN121003539.	3.6	4
4	Exploring the Genetic Architecture of Spontaneous Coronary Artery Dissection Using Whole-Genome Sequencing. <i>Circulation Genomic and Precision Medicine</i> , 2022, 15, 101161CIRCGEN121003527.	3.6	14
5	Spontaneous Coronary Artery Dissection and Fibromuscular Dysplasia: Vasculopathies With a Predilection for Women. <i>Heart Lung and Circulation</i> , 2021, 30, 27-35.	0.4	15
6	Heterozygous loss of <i>WBP11</i> function causes multiple congenital defects in humans and mice. <i>Human Molecular Genetics</i> , 2021, 29, 3662-3678.	2.9	14
7	Genotype-phenotype associations in colorectal adenocarcinomas and their matched metastases. <i>Human Pathology</i> , 2021, 107, 104-116.	2.0	6
8	Transposon clusters as substrates for aberrant splice-site activation. <i>RNA Biology</i> , 2021, 18, 354-367.	3.1	8
9	Genotyping data of routinely processed matched primary/metastatic tumor samples. <i>Data in Brief</i> , 2021, 34, 106646.	1.0	2
10	Maternal iron deficiency perturbs embryonic cardiovascular development in mice. <i>Nature Communications</i> , 2021, 12, 3447.	12.8	17
11	Tumor Genotyping and Homologous Recombination Repair Gene Variants in Patients With Epithelial Ovarian Cancer: Is Pathogenic Enough?. <i>Frontiers in Oncology</i> , 2021, 11, 683057.	2.8	1
12	Functional characterization of a novel <i>PBX1</i> de novo missense variant identified in a patient with syndromic congenital heart disease. <i>Human Molecular Genetics</i> , 2020, 29, 1068-1082.	2.9	26
13	Is There an Independent Role of <i>TERT</i> and <i>NF1</i> in High Grade Gliomas?. <i>Translational Oncology</i> , 2020, 13, 346-354.	3.7	11
14	Functional genomics and gene-environment interaction highlight the complexity of congenital heart disease caused by Notch pathway variants. <i>Human Molecular Genetics</i> , 2020, 29, 566-579.	2.9	32
15	Dynamics of Transforming Growth Factor (TGF)- $\beta$ 2 Superfamily Cytokine Induction During HIV-1 Infection Are Distinct From Other Innate Cytokines. <i>Frontiers in Immunology</i> , 2020, 11, 596841.	4.8	15
16	Spontaneous Coronary Artery Dissection. <i>Circulation Genomic and Precision Medicine</i> , 2020, 13, e003030.	3.6	43
17	dv-trio: a family-based variant calling pipeline using DeepVariant. <i>Bioinformatics</i> , 2020, 36, 3549-3551.	4.1	9
18	Tumor Mutational Patterns and Infiltrating Lymphocyte Density in Young and Elderly Patients With Breast Cancer. <i>Cancer Genomics and Proteomics</i> , 2020, 17, 181-193.	2.0	2

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19	Prognostic Biomarkers in Early-stage Gastric Adenocarcinoma Treated With Adjuvant Chemoradiotherapy. <i>Cancer Genomics and Proteomics</i> , 2020, 17, 277-290.	2.0	4
20	Multi-omic profiling reveals associations between the gut mucosal microbiome, the metabolome, and host DNA methylation associated gene expression in patients with colorectal cancer. <i>BMC Microbiology</i> , 2020, 20, 83.	3.3	36
21	Pathogenic mutations and overall survival in 3,084 patients with cancer: the Hellenic Cooperative Oncology Group Precision Medicine Initiative. <i>Oncotarget</i> , 2020, 11, 1-14.	1.8	1
22	A gene-centric strategy for identifying disease-causing rare variants in dilated cardiomyopathy. <i>Genetics in Medicine</i> , 2019, 21, 133-143.	2.4	25
23	Host and microbiome multi-omics integration: applications and methodologies. <i>Biophysical Reviews</i> , 2019, 11, 55-65.	3.2	66
24	Big data: the elements of good questions, open data, and powerful software. <i>Biophysical Reviews</i> , 2019, 11, 1-3.	3.2	11
25	Spliceogen: an integrative, scalable tool for the discovery of splice-altering variants. <i>Bioinformatics</i> , 2019, 35, 4405-4407.	4.1	7
26	Opposite Prognostic Impact of Single PTEN-loss and PIK3CA Mutations in Early High-risk Breast Cancer. <i>Cancer Genomics and Proteomics</i> , 2019, 16, 195-206.	2.0	13
27	Population-based identity-by-descent mapping combined with exome sequencing to detect rare risk variants for schizophrenia. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2019, 180, 223-231.	1.7	2
28	VPOT: A Customizable Variant Prioritization Ordering Tool for Annotated Variants. <i>Genomics, Proteomics and Bioinformatics</i> , 2019, 17, 540-545.	6.9	10
29	Comparison of somatic variant detection algorithms using Ion Torrent targeted deep sequencing data. <i>BMC Medical Genomics</i> , 2019, 12, 181.	1.5	9
30	Relapsed and De Novo Metastatic HER2-positive Breast Cancer Treated With Trastuzumab: Tumor Genotypes and Clinical Measures Associated With Patient Outcome. <i>Clinical Breast Cancer</i> , 2019, 19, 113-125.e4.	2.4	8
31	Pathogenic BRCA1 mutations may be necessary but not sufficient for tissue genomic heterogeneity: Deep sequencing data from ovarian cancer patients. <i>Gynecologic Oncology</i> , 2019, 152, 375-386.	1.4	7
32	Association of the PHACTR1/EDN1 Genetic Locus With Spontaneous Coronary Artery Dissection. <i>Journal of the American College of Cardiology</i> , 2019, 73, 58-66.	2.8	147
33	Antiviral activity of bone morphogenetic proteins and activins. <i>Nature Microbiology</i> , 2019, 4, 339-351.	13.3	39
34	Identification of clinically actionable variants from genome sequencing of families with congenital heart disease. <i>Genetics in Medicine</i> , 2019, 21, 1111-1120.	2.4	54
35	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. <i>Inflammatory Bowel Diseases</i> , 2018, 24, 136-148.	1.9	11
36	Genetic burden and associations with adverse neurodevelopment in neonates with congenital heart disease. <i>American Heart Journal</i> , 2018, 201, 33-39.	2.7	19

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37	Genetic variation in VAC14 is associated with bacteremia secondary to diverse pathogens in African children. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2018, 115, E3601-E3603.	7.1	12
38	A Screening Approach to Identify Clinically Actionable Variants Causing Congenital Heart Disease in Exome Data. <i>Circulation Genomic and Precision Medicine</i> , 2018, 11, e001978.	3.6	65
39	Prevalent somatic <i>BRCA1</i> mutations shape clinically relevant genomic patterns of nasopharyngeal carcinoma in Southeast Europe. <i>International Journal of Cancer</i> , 2018, 142, 66-80.	5.1	13
40	Survival of Idiopathic Pulmonary Arterial Hypertension Patients in the Modern Era in Australia and New Zealand. <i>Heart Lung and Circulation</i> , 2018, 27, 1368-1375.	0.4	26
41	Selfish mutations dysregulating RAS-MAPK signaling are pervasive in aged human testes. <i>Genome Research</i> , 2018, 28, 1779-1790.	5.5	56
42	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. <i>Medical Oncology</i> , 2018, 35, 101.	2.5	6
43	Genomic Dissection of Bipolar Disorder and Schizophrenia, Including 28 Subphenotypes. <i>Cell</i> , 2018, 173, 1705-1715.e16.	28.9	623
44	Advances in the Genetics of Congenital Heart Disease. <i>Journal of the American College of Cardiology</i> , 2017, 69, 859-870.	2.8	115
45	SVPV: a structural variant prediction viewer for paired-end sequencing datasets. <i>Bioinformatics</i> , 2017, 33, 2032-2033.	4.1	9
46	Epidemiology and treatment of pulmonary arterial hypertension. <i>Nature Reviews Cardiology</i> , 2017, 14, 603-614.	13.7	310
47	NAD Deficiency, Congenital Malformations, and Niacin Supplementation. <i>New England Journal of Medicine</i> , 2017, 377, 544-552.	27.0	177
48	Isogenic mice exhibit sexually-dimorphic DNA methylation patterns across multiple tissues. <i>BMC Genomics</i> , 2017, 18, 966.	2.8	26
49	Evaluation of the immunogenicity and impact on the latent HIV-1 reservoir of a conserved region vaccine, MVA.HIVconsv, in antiretroviral therapy-treated subjects. <i>Journal of the International AIDS Society</i> , 2017, 20, 21171.	3.0	36
50	The miR-200 family is increased in dysplastic lesions in ulcerative colitis patients. <i>PLoS ONE</i> , 2017, 12, e0173664.	2.5	14
51	Whole-genome sequencing of spermatocytic tumors provides insights into the mutational processes operating in the male germline. <i>PLoS ONE</i> , 2017, 12, e0178169.	2.5	36
52	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. <i>Oncotarget</i> , 2016, 7, 32731-32753.	1.8	30
53	Tumor Infiltrating Lymphocytes Affect the Outcome of Patients with Operable Triple-Negative Breast Cancer in Combination with Mutated Amino Acid Classes. <i>PLoS ONE</i> , 2016, 11, e0163138.	2.5	8
54	Familial adenomatous patients with desmoid tumours show increased expression of miR-34a in serum and high levels in tumours. <i>Oncoscience</i> , 2016, 3, 173-185.	2.2	9

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55	Effects of TP53 and PIK3CA mutations in early breast cancer: a matter of co-mutation and tumor-infiltrating lymphocytes. <i>Breast Cancer Research and Treatment</i> , 2016, 158, 307-321.	2.5	16
56	A Method to Exploit the Structure of Genetic Ancestry Space to Enhance Case-Control Studies. <i>American Journal of Human Genetics</i> , 2016, 98, 857-868.	6.2	21
57	A cloud-based framework for applying metamorphic testing to a bioinformatics pipeline. , 2016, , .		4
58	Male-lineage transmission of an acquired metabolic phenotype induced by grand-paternal obesity. <i>Molecular Metabolism</i> , 2016, 5, 699-708.	6.5	154
59	Polymorphism in a lincRNA Associates with a Doubled Risk of Pneumococcal Bacteremia in Kenyan Children. <i>American Journal of Human Genetics</i> , 2016, 98, 1092-1100.	6.2	39
60	Visualizing the origins of selfish de novo mutations in individual seminiferous tubules of human testes. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2016, 113, 2454-2459.	7.1	45
61	Binding of transcription factor GabR to DNA requires recognition of DNA shape at a location distinct from its cognate binding site. <i>Nucleic Acids Research</i> , 2016, 44, 1411-1420.	14.5	35
62	Disease evolution and heterogeneity in bilateral breast cancer. <i>American Journal of Cancer Research</i> , 2016, 6, 2611-2630.	1.4	5
63	How to test bioinformatics software?. <i>Biophysical Reviews</i> , 2015, 7, 343-352.	3.2	16
64	Myelodysplastic Syndromes Are Propagated by Rare and Distinct Human Cancer Stem Cells In Vivo. <i>Cancer Cell</i> , 2015, 27, 603-605.	16.8	0
65	Decoding the complex genetic causes of heart diseases using systems biology. <i>Biophysical Reviews</i> , 2015, 7, 141-159.	3.2	0
66	Rapidly Escalating Hepcidin and Associated Serum Iron Starvation Are Features of the Acute Response to Typhoid Infection in Humans. <i>PLoS Neglected Tropical Diseases</i> , 2015, 9, e0004029.	3.0	38
67	Targeted Next-Generation Sequencing Identifies Pathogenic Variants in Familial Congenital Heart Disease. <i>Journal of the American College of Cardiology</i> , 2014, 64, 2498-2506.	2.8	85
68	An inherited duplication at the gene p21 Protein-Activated Kinase 7 (PAK7) is a risk factor for psychosis. <i>Human Molecular Genetics</i> , 2014, 23, 3316-3326.	2.9	37
69	Myelodysplastic Syndromes Are Propagated by Rare and Distinct Human Cancer Stem Cells In Vivo. <i>Cancer Cell</i> , 2014, 25, 794-808.	16.8	272
70	Analysis of hundreds of cis-regulatory landscapes at high resolution in a single, high-throughput experiment. <i>Nature Genetics</i> , 2014, 46, 205-212.	21.4	417
71	Distinct patterns of hepcidin and iron regulation during HIV-1, HBV, and HCV infections. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2014, 111, 12187-12192.	7.1	79
72	Verification and validation of bioinformatics software without a gold standard: a case study of BWA and Bowtie. <i>BMC Bioinformatics</i> , 2014, 15, S15.	2.6	58

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73	Genome-wide association analysis identifies 13 new risk loci for schizophrenia. <i>Nature Genetics</i> , 2013, 45, 1150-1159.	21.4	1,395
74	Common variants in the HLA-DRB1-HLA-DQA1 HLA class II region are associated with susceptibility to visceral leishmaniasis. <i>Nature Genetics</i> , 2013, 45, 208-213.	21.4	86
75	Early dynamic fate changes in haemogenic endothelium characterized at the single-cell level. <i>Nature Communications</i> , 2013, 4, 2924.	12.8	158
76	Contributions of intrinsic mutation rate and selfish selection to levels of de novo HRAS mutations in the paternal germline. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2013, 110, 20152-20157.	7.1	70
77	Cellular interference in craniofrontonasal syndrome: males mosaic for mutations in the X-linked EFNB1 gene are more severely affected than true hemizygotes. <i>Human Molecular Genetics</i> , 2013, 22, 1654-1662.	2.9	66
78	Interferon-induced transmembrane protein-3 genetic variant rs12252-C is associated with severe influenza in Chinese individuals. <i>Nature Communications</i> , 2013, 4, 1418.	12.8	228
79	Pneumococcal genome sequencing tracks a vaccine escape variant formed through a multi-fragment recombination event. <i>Nature Genetics</i> , 2012, 44, 352-355.	21.4	144
80	Identification of 15 new psoriasis susceptibility loci highlights the role of innate immunity. <i>Nature Genetics</i> , 2012, 44, 1341-1348.	21.4	848
81	Genome-Wide Association Study Implicates HLA-C*01:02 as a Risk Factor at the Major Histocompatibility Complex Locus in Schizophrenia. <i>Biological Psychiatry</i> , 2012, 72, 620-628.	1.3	156
82	Smchd1-Dependent and -Independent Pathways Determine Developmental Dynamics of CpG Island Methylation on the Inactive X Chromosome. <i>Developmental Cell</i> , 2012, 23, 265-279.	7.0	160
83	Genetic risk and a primary role for cell-mediated immune mechanisms in multiple sclerosis. <i>Nature</i> , 2011, 476, 214-219.	27.8	2,400
84	Genome-wide association study of CNVs in 16,000 cases of eight common diseases and 3,000 shared controls. <i>Nature</i> , 2010, 464, 713-720.	27.8	737
85	GenoSNP: a variational Bayes within-sample SNP genotyping algorithm that does not require a reference population. <i>Bioinformatics</i> , 2008, 24, 2209-2214.	4.1	65