

# Emmanouil Dermitzakis

## List of Publications by Year in descending order

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147  
papers

40,335  
citations

9264

74  
h-index

10445

139  
g-index

168  
all docs

168  
docs citations

168  
times ranked

59421  
citing authors

#	ARTICLE	IF	CITATIONS
1	2021 Curt Stern Award: Studying the biology of “junk” American Journal of Human Genetics, 2022, 109, 387-389.	6.2	0
2	Integrated GWAS and Gene Expression Suggest ORM1 as a Potential Regulator of Plasma Levels of Cell-Free DNA and Thrombosis Risk. Thrombosis and Haemostasis, 2022, 122, 1027-1039.	3.4	6
3	Ether lipids, sphingolipids and toxic “deoxyceramides as hallmarks for lean and obese type 2 diabetic patients. Acta Physiologica, 2021, 232, e13610.	3.8	29
4	Sex-dimorphic genetic effects and novel loci for fasting glucose and insulin variability. Nature Communications, 2021, 12, 24.	12.8	87
5	The Christian Orthodox Church Fasting Diet Is Associated with Lower Levels of Depression and Anxiety and a Better Cognitive Performance in Middle Life. Nutrients, 2021, 13, 627.	4.1	10
6	Gene regulation contributes to explain the impact of early life socioeconomic disadvantage on adult inflammatory levels in two cohort studies. Scientific Reports, 2021, 11, 3100.	3.3	15
7	Single-cell transcriptomics reveal temporal dynamics of critical regulators of germ cell fate during mouse sex determination. FASEB Journal, 2021, 35, e21452.	0.5	36
8	Specific Transcriptomic Signatures and Dual Regulation of Steroidogenesis Between Fetal and Adult Mouse Leydig Cells. Frontiers in Cell and Developmental Biology, 2021, 9, 695546.	3.7	19
9	The molecular basis, genetic control and pleiotropic effects of local gene co-expression. Nature Communications, 2021, 12, 4842.	12.8	18
10	Identification of tissue-specific and common methylation quantitative trait loci in healthy individuals using MAGAR. Epigenetics and Chromatin, 2021, 14, 44.	3.9	3
11	Large-scale cis- and trans-eQTL analyses identify thousands of genetic loci and polygenic scores that regulate blood gene expression. Nature Genetics, 2021, 53, 1300-1310.	21.4	590
12	A brief history of human disease genetics. Nature, 2020, 577, 179-189.	27.8	441
13	Extensive fragmentation and re-organization of transcription in Systemic Lupus Erythematosus. Scientific Reports, 2020, 10, 16648.	3.3	8
14	P0359CROSS-TISSUE AND MURINE-HUMAN COMPARATIVE TRANSCRIPTOME ANALYSES IDENTIFY TARGETABLE GENES FOR HUMAN SYSTEMIC LUPUS ERYTHEMATOUS AND LUPUS NEPHRITIS. Nephrology Dialysis Transplantation, 2020, 35, .	0.7	0
15	Cell type-“specific genetic regulation of gene expression across human tissues. Science, 2020, 369, .	12.6	210
16	The impact of sex on gene expression across human tissues. Science, 2020, 369, .	12.6	329
17	A vast resource of allelic expression data spanning human tissues. Genome Biology, 2020, 21, 234.	8.8	68
18	The significant effect on musculoskeletal metabolism and bone density of the Eastern Mediterranean Christian Orthodox Church fasting. European Journal of Clinical Nutrition, 2020, 74, 1736-1742.	2.9	4

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19	MethCORR modelling of methylomes from formalin-fixed paraffin-embedded tissue enables characterization and prognostication of colorectal cancer. Nature Communications, 2020, 11, 2025.	12.8	5
20	Genetic variant effects on gene expression in human pancreatic islets and their implications for T2D. Nature Communications, 2020, 11, 4912.	12.8	89
21	Title is missing!. , 2020, 17, e1003149.		0
22	Title is missing!. , 2020, 17, e1003149.		0
23	Title is missing!. , 2020, 17, e1003149.		0
24	Title is missing!. , 2020, 17, e1003149.		0
25	Title is missing!. , 2020, 17, e1003149.		0
26	Cellular circadian period length inversely correlates with HbA1c levels in individuals with type 2 diabetes. Diabetologia, 2019, 62, 1453-1462.	6.3	13
27	Combined genetic and transcriptome analysis of patients with SLE: distinct, targetable signatures for susceptibility and severity. Annals of the Rheumatic Diseases, 2019, 78, 1079-1089.	0.9	109
28	Expression estimation and eQTL mapping for HLA genes with a personalized pipeline. PLoS Genetics, 2019, 15, e1008091.	3.5	75
29	Chromatin three-dimensional interactions mediate genetic effects on gene expression. Science, 2019, 364, .	12.6	163
30	Dissecting Cell Lineage Specification and Sex Fate Determination in Gonadal Somatic Cells Using Single-Cell Transcriptomics. Cell Reports, 2019, 26, 3272-3283.e3.	6.4	137
31	OP0277&œ...RNA SEQUENCING AND MACHINE LEARNING TECHNIQUES PREDICT MAJOR ORGAN INVOLVEMENT IN PATIENTS WITH SYSTEMIC LUPUS ERYTHEMATOSUS. , 2019, , .		0
32	Accurate, scalable and integrative haplotype estimation. Nature Communications, 2019, 10, 5436.	12.8	336
33	Deciphering Cell Lineage Specification during Male Sex Determination with Single-Cell RNA Sequencing. Cell Reports, 2018, 22, 1589-1599.	6.4	126
34	Contribution of allelic imbalance to colorectal cancer. Nature Communications, 2018, 9, 3664.	12.8	25
35	Transcriptomic analyses reveal rhythmic and CLOCK-driven pathways in human skeletal muscle. ELife, 2018, 7, .	6.0	87
36	Using an atlas of gene regulation across 44 human tissues to inform complex disease- and trait-associated variation. Nature Genetics, 2018, 50, 956-967.	21.4	389

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37	Mapping eQTLs with RNA-seq reveals novel susceptibility genes, non-coding RNAs and alternative-splicing events in systemic lupus erythematosus. Human Molecular Genetics, 2017, 26, ddw417.	2.9	39
38	The non-coding variant rs1800734 enhances DCLK3 expression through long-range interaction and promotes colorectal cancer progression. Nature Communications, 2017, 8, 14418.	12.8	48
39	<i>i&gt;MBV&lt;/i&gt;: a method to solve sample mislabeling and detect technical bias in large combined genotype and sequencing assay datasets. Bioinformatics, 2017, 33, 1895-1897.</i>	4.1	43
40	A complete tool set for molecular QTL discovery and analysis. Nature Communications, 2017, 8, 15452.	12.8	247
41	Time-dependent genetic effects on gene expression implicate aging processes. Genome Research, 2017, 27, 545-552.	5.5	31
42	Predicting causal variants affecting expression by using whole-genome sequencing and RNA-seq from multiple human tissues. Nature Genetics, 2017, 49, 1747-1751.	21.4	88
43	Estimating the causal tissues for complex traits and diseases. Nature Genetics, 2017, 49, 1676-1683.	21.4	166
44	Genetic correlations reveal the shared genetic architecture of transcription in human peripheral blood. Nature Communications, 2017, 8, 483.	12.8	22
45	The effect of genetic variation on promoter usage and enhancer activity. Nature Communications, 2017, 8, 1358.	12.8	50
46	Integrative genomic analysis implicates limited peripheral adipose storage capacity in the pathogenesis of human insulin resistance. Nature Genetics, 2017, 49, 17-26.	21.4	452
47	The genomic landscape of human cellular circadian variation points to a novel role for the signalosome. ELife, 2017, 6, .	6.0	9
48	SNHG16 is regulated by the Wnt pathway in colorectal cancer and affects genes involved in lipid metabolism. Molecular Oncology, 2016, 10, 1266-1282.	4.6	151
49	Adiposity-Dependent Regulatory Effects on Multi-tissue Transcriptomes. American Journal of Human Genetics, 2016, 99, 567-579.	6.2	26
50	Genetic Drivers of Epigenetic and Transcriptional Variation in Human Immune Cells. Cell, 2016, 167, 1398-1414.e24.	28.9	573
51	A functional circadian clock is required for proper insulin secretion by human pancreatic islet cells. Diabetes, Obesity and Metabolism, 2016, 18, 355-365.	4.4	77
52	Fast and efficient QTL mapper for thousands of molecular phenotypes. Bioinformatics, 2016, 32, 1479-1485.	4.1	426
53	Sequential transcriptional waves direct the differentiation of newborn neurons in the mouse neocortex. Science, 2016, 351, 1443-1446.	12.6	264
54	Genome-wide association analysis of self-reported events in 6135 individuals and 252 827 controls identifies 8 loci associated with thrombosis. Human Molecular Genetics, 2016, 25, 1867-1874.	2.9	103

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55	IL-13 improves beta-cell survival and protects against IL-1beta-induced beta-cell death. <i>Molecular Metabolism</i> , 2016, 5, 122-131.	6.5	25
56	The Transcriptome of Equine Peripheral Blood Mononuclear Cells. <i>PLoS ONE</i> , 2015, 10, e0122011.	2.5	17
57	Transcript Expression Data from Human Islets Links Regulatory Signals from Genome-Wide Association Studies for Type 2 Diabetes and Glycemic Traits to Their Downstream Effectors. <i>PLoS Genetics</i> , 2015, 11, e1005694.	3.5	178
58	Impaired Cell Cycle Regulation in a Natural Equine Model of Asthma. <i>PLoS ONE</i> , 2015, 10, e0136103.	2.5	24
59	Integrated analysis of mRNA and miRNA expression in response to interleukin-6 in hepatocytes. <i>Genomics</i> , 2015, 106, 107-115.	2.9	13
60	Alternative Splicing QTLs in European and African Populations. <i>American Journal of Human Genetics</i> , 2015, 97, 567-575.	6.2	55
61	Biased Allelic Expression in Human Primary Fibroblast Single Cells. <i>American Journal of Human Genetics</i> , 2015, 96, 70-80.	6.2	117
62	Short Term Exposure of Beta Cells to Low Concentrations of Interleukin-1 $\beta$ Improves Insulin Secretion through Focal Adhesion and Actin Remodeling and Regulation of Gene Expression. <i>Journal of Biological Chemistry</i> , 2015, 290, 6653-6669.	3.4	28
63	Tissue-Specific Effects of Genetic and Epigenetic Variation on Gene Regulation and Splicing. <i>PLoS Genetics</i> , 2015, 11, e1004958.	3.5	185
64	The human transcriptome across tissues and individuals. <i>Science</i> , 2015, 348, 660-665.	12.6	1,127
65	The Genotype-Tissue Expression (GTEx) pilot analysis: Multitissue gene regulation in humans. <i>Science</i> , 2015, 348, 648-660.	12.6	4,659
66	Effect of predicted protein-truncating genetic variants on the human transcriptome. <i>Science</i> , 2015, 348, 666-669.	12.6	252
67	Assessing allele-specific expression across multiple tissues from RNA-seq read data. <i>Bioinformatics</i> , 2015, 31, 2497-2504.	4.1	90
68	Population Variation and Genetic Control of Modular Chromatin Architecture in Humans. <i>Cell</i> , 2015, 162, 1039-1050.	28.9	210
69	Integrated analysis of mRNA and miRNA expression in response to interleukin-6 in hepatocytes. <i>Data in Brief</i> , 2015, 4, 226-228.	1.0	3
70	Gene-gene and gene-environment interactions detected by transcriptome sequence analysis in twins. <i>Nature Genetics</i> , 2015, 47, 88-91.	21.4	215
71	Tandem RNA Chimeras Contribute to Transcriptome Diversity in Human Population and Are Associated with Intronic Genetic Variants. <i>PLoS ONE</i> , 2014, 9, e104567.	2.5	31
72	Allelic mapping bias in RNA-sequencing is not a major confounder in eQTL studies. <i>Genome Biology</i> , 2014, 15, 467.	8.8	67

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73	Cigarette smoking reduces DNA methylation levels at multiple genomic loci but the effect is partially reversible upon cessation. <i>Epigenetics</i> , 2014, 9, 1382-1396.	2.7	285
74	Cis and Trans Effects of Human Genomic Variants on Gene Expression. <i>PLoS Genetics</i> , 2014, 10, e1004461.	3.5	117
75	Fractalkine (CX3CL1), a new factor protecting $\beta^2$ -cells against TNF $\alpha$ . <i>Molecular Metabolism</i> , 2014, 3, 731-741.	6.5	31
76	Gene Age Predicts the Strength of Purifying Selection Acting on Gene Expression Variation in Humans. <i>American Journal of Human Genetics</i> , 2014, 95, 660-674.	6.2	35
77	Whole Exome Sequencing of a Dominant Retinitis Pigmentosa Family Identifies a Novel Deletion in <i>PRPF31</i> . , 2014, 55, 2121.		26
78	Putative cis-regulatory drivers in colorectal cancer. <i>Nature</i> , 2014, 512, 87-90.	27.8	136
79	Genetic interactions affecting human gene expression identified by variance association mapping. <i>ELife</i> , 2014, 3, e01381.	6.0	137
80	Coordinated Effects of Sequence Variation on DNA Binding, Chromatin Structure, and Transcription. <i>Science</i> , 2013, 342, 744-747.	12.6	364
81	Cell-type, allelic, and genetic signatures in the human pancreatic beta cell transcriptome. <i>Genome Research</i> , 2013, 23, 1554-1562.	5.5	161
82	Genetic and Epigenetic Regulation of Human lincRNA Gene Expression. <i>American Journal of Human Genetics</i> , 2013, 93, 1015-1026.	6.2	65
83	Expression quantitative trait loci: present and future. <i>Philosophical Transactions of the Royal Society B: Biological Sciences</i> , 2013, 368, 20120362.	4.0	363
84	Reproducibility of high-throughput mRNA and small RNA sequencing across laboratories. <i>Nature Biotechnology</i> , 2013, 31, 1015-1022.	17.5	251
85	Transcriptome and genome sequencing uncovers functional variation in humans. <i>Nature</i> , 2013, 501, 506-511.	27.8	1,857
86	Examination of the relationship between variation at 17q21 and childhood wheeze phenotypes. <i>Journal of Allergy and Clinical Immunology</i> , 2013, 131, 685-694.	2.9	66
87	Global Analysis of DNA Methylation Variation in Adipose Tissue from Twins Reveals Links to Disease-Associated Variants in Distal Regulatory Elements. <i>American Journal of Human Genetics</i> , 2013, 93, 876-890.	6.2	330
88	Sex-stratified Genome-wide Association Studies Including 270,000 Individuals Show Sexual Dimorphism in Genetic Loci for Anthropometric Traits. <i>PLoS Genetics</i> , 2013, 9, e1003500.	3.5	371
89	Congruence of Additive and Non-Additive Effects on Gene Expression Estimated from Pedigree and SNP Data. <i>PLoS Genetics</i> , 2013, 9, e1003502.	3.5	79
90	Gene expression changes with age in skin, adipose tissue, blood and brain. <i>Genome Biology</i> , 2013, 14, R75.	9.6	263

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91	The Genotype-Tissue Expression (GTEx) project. <i>Nature Genetics</i> , 2013, 45, 580-585.	21.4	6,815
92	Passive and active DNA methylation and the interplay with genetic variation in gene regulation. <i>ELife</i> , 2013, 2, e00523.	6.0	374
93	Immune response is a personal matter. <i>ELife</i> , 2013, 2, e00899.	6.0	1
94	Extent, Causes, and Consequences of Small RNA Expression Variation in Human Adipose Tissue. <i>PLoS Genetics</i> , 2012, 8, e1002704.	3.5	48
95	Patterns of Cis Regulatory Variation in Diverse Human Populations. <i>PLoS Genetics</i> , 2012, 8, e1002639.	3.5	439
96	Sex-biased genetic effects on gene regulation in humans. <i>Genome Research</i> , 2012, 22, 2368-2375.	5.5	92
97	Genetic control of gene expression in whole blood and lymphoblastoid cell lines is largely independent. <i>Genome Research</i> , 2012, 22, 456-466.	5.5	75
98	Genetic and epigenetic contribution to complex traits. <i>Human Molecular Genetics</i> , 2012, 21, R24-R28.	2.9	60
99	Epigenome-Wide Scans Identify Differentially Methylated Regions for Age and Age-Related Phenotypes in a Healthy Ageing Population. <i>PLoS Genetics</i> , 2012, 8, e1002629.	3.5	620
100	Mapping cis- and trans-regulatory effects across multiple tissues in twins. <i>Nature Genetics</i> , 2012, 44, 1084-1089.	21.4	701
101	Meta-analysis of genome-wide association studies identifies three new risk loci for atopic dermatitis. <i>Nature Genetics</i> , 2012, 44, 187-192.	21.4	311
102	The Brisbane Systems Genetics Study: Genetical Genomics Meets Complex Trait Genetics. <i>PLoS ONE</i> , 2012, 7, e35430.	2.5	83
103	Tandem repeat sequence variation as causative Cis-eQTLs for protein-coding gene expression variation: The case of CSTB. <i>Human Mutation</i> , 2012, 33, 1302-1309.	2.5	34
104	A Systematic Survey of Loss-of-Function Variants in Human Protein-Coding Genes. <i>Science</i> , 2012, 335, 823-828.	12.6	1,095
105	Insights into hominid evolution from the gorilla genome sequence. <i>Nature</i> , 2012, 483, 169-175.	27.8	663
106	Cellular genomics for complex traits. <i>Nature Reviews Genetics</i> , 2012, 13, 215-220.	16.3	37
107	Extensive Natural Variation for Cellular Hydrogen Peroxide Release Is Genetically Controlled. <i>PLoS ONE</i> , 2012, 7, e43566.	2.5	5
108	Genome-Wide Association Identifies Nine Common Variants Associated With Fasting Proinsulin Levels and Provides New Insights Into the Pathophysiology of Type 2 Diabetes. <i>Diabetes</i> , 2011, 60, 2624-2634.	0.6	335

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109	Genome-wide association study identifies a common variant associated with risk of endometrial cancer. <i>Nature Genetics</i> , 2011, 43, 451-454.	21.4	141
110	Identification of an imprinted master trans regulator at the KLF14 locus related to multiple metabolic phenotypes. <i>Nature Genetics</i> , 2011, 43, 561-564.	21.4	289
111	From expression QTLs to personalized transcriptomics. <i>Nature Reviews Genetics</i> , 2011, 12, 277-282.	16.3	148
112	Epistatic Selection between Coding and Regulatory Variation in Human Evolution and Disease. <i>American Journal of Human Genetics</i> , 2011, 89, 459-463.	6.2	73
113	Identification of <i>cis</i> - and <i>trans</i> -regulatory variation modulating microRNA expression levels in human fibroblasts. <i>Genome Research</i> , 2011, 21, 68-73.	5.5	70
114	The Architecture of Gene Regulatory Variation across Multiple Human Tissues: The MuTHER Study. <i>PLoS Genetics</i> , 2011, 7, e1002003.	3.5	392
115	Rare and Common Regulatory Variation in Population-Scale Sequenced Human Genomes. <i>PLoS Genetics</i> , 2011, 7, e1002144.	3.5	98
116	Genome Literacy. <i>Science</i> , 2011, 331, 689-690.	12.6	1
117	Data analysis issues for allele-specific expression using Illumina's GoldenGate assay. <i>BMC Bioinformatics</i> , 2010, 11, 280.	2.6	4
118	High-throughput analysis of candidate imprinted genes and allele-specific gene expression in the human term placenta. <i>BMC Genetics</i> , 2010, 11, 25.	2.7	64
119	Transcriptome genetics using second generation sequencing in a Caucasian population. <i>Nature</i> , 2010, 464, 773-777.	27.8	782
120	Genome-wide association study of migraine implicates a common susceptibility variant on 8q22.1. <i>Nature Genetics</i> , 2010, 42, 869-873.	21.4	332
121	Meta-analysis of three genome-wide association studies identifies susceptibility loci for colorectal cancer at 1q41, 3q26.2, 12q13.13 and 20q13.33. <i>Nature Genetics</i> , 2010, 42, 973-977.	21.4	335
122	Evolutionary history of regulatory variation in human populations. <i>Human Molecular Genetics</i> , 2010, 19, R197-R203.	2.9	10
123	Independent and population-specific association of risk variants at the IRGM locus with Crohn's disease. <i>Human Molecular Genetics</i> , 2010, 19, 1828-1839.	2.9	93
124	An Immune Response Network Associated with Blood Lipid Levels. <i>PLoS Genetics</i> , 2010, 6, e1001113.	3.5	112
125	Candidate Causal Regulatory Effects by Integration of Expression QTLs with Complex Trait Genetic Associations. <i>PLoS Genetics</i> , 2010, 6, e1000895.	3.5	434
126	Genevar: a database and Java application for the analysis and visualization of SNP-gene associations in eQTL studies. <i>Bioinformatics</i> , 2010, 26, 2474-2476.	4.1	282



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127	Long- and Short-Term Selective Forces on Malaria Parasite Genomes. PLoS Genetics, 2010, 6, e1001099.	3.5	30
128	The resolution of the genetics of gene expression. Human Molecular Genetics, 2009, 18, R211-R215.	2.9	20
129	Common Regulatory Variation Impacts Gene Expression in a Cell Type-Dependent Manner. Science, 2009, 325, 1246-1250.	12.6	694
130	Meta-Analysis of Genome-Wide Scans for Human Adult Stature Identifies Novel Loci and Associations with Measures of Skeletal Frame Size. PLoS Genetics, 2009, 5, e1000445.	3.5	237
131	A genome-wide association study of testicular germ cell tumor. Nature Genetics, 2009, 41, 807-810.	21.4	317
132	Life After GWA Studies. Science, 2009, 326, 239-240.	12.6	32
133	Genetic variation of regulatory systems. Current Opinion in Genetics and Development, 2009, 19, 586-590.	3.3	12
134	Williams-Beuren syndrome TRIM50 encodes an E3 ubiquitin ligase. European Journal of Human Genetics, 2008, 16, 1038-1049.	2.8	43
135	Common variants near MC4R are associated with fat mass, weight and risk of obesity. Nature Genetics, 2008, 40, 768-775.	21.4	1,179
136	From gene expression to disease risk. Nature Genetics, 2008, 40, 492-493.	21.4	85
137	The functional impact of structural variation in humans. Trends in Genetics, 2008, 24, 238-245.	6.7	163
138	Assaying the regulatory potential of mammalian conserved non-coding sequences in human cells. Genome Biology, 2008, 9, R168.	9.6	18
139	Using gene expression to investigate the genetic basis of complex disorders. Human Molecular Genetics, 2008, 17, R129-R134.	2.9	101
140	Gene Expression Levels Are a Target of Recent Natural Selection in the Human Genome. Molecular Biology and Evolution, 2008, 26, 649-658.	8.9	96
141	High-Resolution Mapping of Expression-QTLs Yields Insight into Human Gene Regulation. PLoS Genetics, 2008, 4, e1000214.	3.5	510
142	Modifier Effects between Regulatory and Protein-Coding Variation. PLoS Genetics, 2008, 4, e1000244.	3.5	33
143	Genetic Analysis of Human Traits In Vitro: Drug Response and Gene Expression in Lymphoblastoid Cell Lines. PLoS Genetics, 2008, 4, e1000287.	3.5	200
144	Chapter 11 Regulatory Variation and Evolution: Implications for Disease. Advances in Genetics, 2008, 61, 295-306.	1.8	2

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145	Breaking the waves: improved detection of copy number variation from microarray-based comparative genomic hybridization. <i>Genome Biology</i> , 2007, 8, R228.	9.6	120
146	Genome-wide detection and characterization of positive selection in human populations. <i>Nature</i> , 2007, 449, 913-918.	27.8	1,788
147	Correction: Passive and active DNA methylation and the interplay with genetic variation in gene regulation. <i>ELife</i> , 0, 2, .	6.0	15