Tuuli Lappalainen

List of Publications by Year
in descending order

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75 papers	34,561 citations	50170 46 h-index	71532 76 g-index
102 all docs	102 docs citations	102 times ranked	57484 citing authors

#	Article	IF	Citations
1	An integrated map of genetic variation from 1,092 human genomes. Nature, 2012, 491, 56-65.	13.7	7,199
2	The Genotype-Tissue Expression (GTEx) project. Nature Genetics, 2013, 45, 580-585.	9.4	6,815
3	The Genotype-Tissue Expression (GTEx) pilot analysis: Multitissue gene regulation in humans. Science, 2015, 348, 648-660.	6.0	4,659
4	Genetic effects on gene expression across human tissues. Nature, 2017, 550, 204-213.	13.7	3,500
5	Transcriptome and genome sequencing uncovers functional variation in humans. Nature, 2013, 501, 506-511.	13.7	1,857
6	The human transcriptome across tissues and individuals. Science, 2015, 348, 660-665.	6.0	1,127
7	Landscape of X chromosome inactivation across human tissues. Nature, 2017, 550, 244-248.	13.7	764
8	Insights into hominid evolution from the gorilla genome sequence. Nature, 2012, 483, 169-175.	13.7	663
9	Identification of Required Host Factors for SARS-CoV-2 Infection in Human Cells. Cell, 2021, 184, 92-105.e16.	13.5	480
10	Passive and active DNA methylation and the interplay with genetic variation in gene regulation. ELife, 2013, 2, e00523.	2.8	374
11	Coordinated Effects of Sequence Variation on DNA Binding, Chromatin Structure, and Transcription. Science, 2013, 342, 744-747.	6.0	364
12	Distribution and Medical Impact of Loss-of-Function Variants in the Finnish Founder Population. PLoS Genetics, 2014, 10, e1004494.	1.5	351
13	Integrative Annotation of Variants from 1092 Humans: Application to Cancer Genomics. Science, 2013, 342, 1235587.	6.0	341
14	Tools and best practices for data processing in allelic expression analysis. Genome Biology, 2015, 16, 195.	3.8	335
15	The impact of sex on gene expression across human tissues. Science, 2020, 369, .	6.0	329
16	Associating cellular epigenetic models with human phenotypes. Nature Reviews Genetics, 2017, 18, 441-451.	7.7	257
17	Effect of predicted protein-truncating genetic variants on the human transcriptome. Science, 2015, 348, 666-669.	6.0	252
18	Reproducibility of high-throughput mRNA and small RNA sequencing across laboratories. Nature Biotechnology, 2013, 31, 1015-1022.	9.4	251

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19	A Quantitative Proteome Map of the Human Body. Cell, 2020, 183, 269-283.e19.	13.5	243
20	The landscape of genomic imprinting across diverse adult human tissues. Genome Research, 2015, 25, 927-936.	2.4	216
21	Gene-gene and gene-environment interactions detected by transcriptome sequence analysis in twins. Nature Genetics, 2015, 47, 88-91.	9.4	215
22	Cell typeâ€“specific genetic regulation of gene expression across human tissues. Science, 2020, 369, .	6.0	210
23	Genomic Analysis in the Age of Human Genome Sequencing. Cell, 2019, 177, 70-84.	13.5	205
24	Tissue-Specific Effects of Genetic and Epigenetic Variation on Gene Regulation and Splicing. PLoS Genetics, 2015, 11, e1004958.	1.5	185
25	Modified penetrance of coding variants by cis-regulatory variation contributes to disease risk. Nature Genetics, 2018, 50, 1327-1334.	9.4	167
26	Identifying Genes Whose Mutant Transcripts Cause Dominant Disease Traits by Potential Gain-of-Function Alleles. American Journal of Human Genetics, 2018, 103, 171-187.	2.6	160
27	Genetic regulatory effects modified by immune activation contribute to autoimmune disease associations. Nature Communications, 2017, 8, 266.	5.8	157
28	Exploiting the GTEx resources to decipher the mechanisms at GWAS loci. Genome Biology, 2021, 22, 49.	3.8	150
29	Molecular Transducers of Physical Activity Consortium (MoTrPAC): Mapping the Dynamic Responses to Exercise. Cell, 2020, 181, 1464-1474.	13.5	147
30	Genetic interactions affecting human gene expression identified by variance association mapping. ELife, 2014, 3, e01381.	2.8	137
31	Quantifying the regulatory effect size of cis-acting genetic variation using allelic fold change. Genome Research, 2017, 27, 1872-1884.	2.4	114
32	Genome-Wide Analysis of Single Nucleotide Polymorphisms Uncovers Population Structure in Northern Europe. PLoS ONE, 2008, 3, e3519.	1.1	112
33	Rare variant phasing and haplotypic expression from RNA sequencing with phASER. Nature Communications, 2016, 7, 12817.	5.8	105
34	Genetic regulatory variation in populations informs transcriptome analysis in rare disease. Science, 2019, 366, 351-356.	6.0	99
35	Rare and Common Regulatory Variation in Population-Scale Sequenced Human Genomes. PLoS Genetics, 2011, 7, e1002144.	1.5	98
36	Population-scale tissue transcriptomics maps long non-coding RNAs to complex disease. Cell, 2021, 184, 2633-2648.e19.	13.5	94

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37	Sex-biased genetic effects on gene regulation in humans. Genome Research, 2012, 22, 2368-2375.	2.4	92
38	Assessing allele-specific expression across multiple tissues from RNA-seq read data. Bioinformatics, 2015, 31, 2497-2504.	1.8	90
39	Transcriptomic signatures across human tissues identify functional rare genetic variation. Science, 2020, 369, .	6.0	89
40	Distinct Variants at LIN28B Influence Growth in Height from Birth to Adulthood. American Journal of Human Genetics, 2010, 86, 773-782.	2.6	81
41	From variant to function in human disease genetics. Science, 2021, 373, 1464-1468.	6.0	75
42	Epistatic Selection between Coding and Regulatory Variation in Human Evolution and Disease. American Journal of Human Genetics, 2011, 89, 459-463.	2.6	73
43	A vast resource of allelic expression data spanning human tissues. Genome Biology, 2020, 21, 234.	3.8	68
44	Regional differences among the Finns: A Y-chromosomal perspective. Gene, 2006, 376, 207-215.	1.0	67
45	Allelic mapping bias in RNA-sequencing is not a major confounder in eQTL studies. Genome Biology, 2014, 15, 467.	3.8	67
46	DNA Methylation Profiling of Uniparental Disomy Subjects Provides a Map of Parental Epigenetic Bias in the Human Genome. American Journal of Human Genetics, 2016, 99, 555-566.	2.6	66
47	Migration Waves to the Baltic Sea Region. Annals of Human Genetics, 2008, 72, 337-348.	0.3	63
48	Functional genomics bridges the gap between quantitative genetics and molecular biology. Genome Research, 2015, 25, 1427-1431.	2.4	63
49	High Risk Population Isolate Reveals Low Frequency Variants Predisposing to Intracranial Aneurysms. PLoS Genetics, 2014, 10, e1004134.	1.5	55
50	Transcriptome Sequencing from Diverse Human Populations Reveals Differentiated Regulatory Architecture. PLoS Genetics, 2014, 10, e1004549.	1.5	49
51	MBV: a method to solve sample mislabeling and detect technical bias in large combined genotype and sequencing assay datasets. Bioinformatics, 2017, 33, 1895-1897.	1.8	43
52	Swedish Population Substructure Revealed by Genome-Wide Single Nucleotide Polymorphism Data. PLoS ONE, 2011, 6, e16747.	1.1	39
53	Population Structure in Contemporary Swedenâ€"A Yâ€Chromosomal and Mitochondrial DNA Analysis. Annals of Human Genetics, 2009, 73, 61-73.	0.3	36
54	Gene Age Predicts the Strength of Purifying Selection Acting on Gene Expression Variation in Humans. American Journal of Human Genetics, 2014, 95, 660-674.	2.6	35

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55	Whole genome sequence analysis of pulmonary function and COPD in 19,996 multi-ethnic participants. Nature Communications, 2020, 11, 5182.	5.8	32
56	Population substructure in Finland and Sweden revealed by the use of spatial coordinates and a small number of unlinked autosomal SNPs. BMC Genetics, 2008, 9, 54.	2.7	31
57	Genomic landscape of positive natural selection in Northern European populations. European Journal of Human Genetics, 2010, 18, 471-478.	1.4	31
58	Tandem RNA Chimeras Contribute to Transcriptome Diversity in Human Population and Are Associated with Intronic Genetic Variants. PLoS ONE, 2014, 9, e104567.	1.1	31
59	Sequence variation between 462 human individuals fine-tunes functional sites of RNA processing. Scientific Reports, 2016, 6, 32406.	1.6	28
60	Identification and removal of low-complexity sites in allele-specific analysis of ChIP-seq data. Bioinformatics, 2014, 30, 165-171.	1.8	21
61	Genetic and non-genetic factors affecting the expression of COVID-19-relevant genes in the large airway epithelium. Genome Medicine, 2021, 13, 66.	3.6	21
62	Leveraging allelic imbalance to refine fine-mapping for eQTL studies. PLoS Genetics, 2019, 15, e1008481.	1.5	20
63	Arsenic Exposure, Blood DNA Methylation, and Cardiovascular Disease. Circulation Research, 2022, 131, .	2.0	20
64	An autoimmune disease risk variant: A trans master regulatory effect mediated by IRF1 under immune stimulation?. PLoS Genetics, 2021, 17, e1009684.	1.5	17
65	Transcription factor regulation of eQTL activity across individuals and tissues. PLoS Genetics, 2022, 18, e1009719.	1.5	14
66	Protein prediction for trait mapping in diverse populations. PLoS ONE, 2022, 17, e0264341.	1.1	13
67	Evolutionary history of regulatory variation in human populations. Human Molecular Genetics, 2010, 19, R197-R203.	1.4	10
68	SnapShot: Discovering Genetic Regulatory Variants by QTL Analysis. Cell, 2017, 171, 980-980.e1.	13.5	10
69	Polygenic transcriptome risk scores for COPD and lung function improve cross-ethnic portability of prediction in the NHLBI TOPMed program. American Journal of Human Genetics, 2022, 109, 857-870.	2.6	7
70	A polyclonal allelic expression assay for detecting regulatory effects of transcript variants. Genome Medicine, 2020, 12, 79.	3.6	5
71	Concerted Genetic Function in Blood Traits. Cell, 2016, 167, 1167-1169.	13.5	4
72	Voices of biotech. Nature Biotechnology, 2016, 34, 270-275.	9.4	4

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73	Janusâ€”a comprehensive tool investigating the two faces of transcription. Bioinformatics, 2013, 29, 1600-1606.	1.8	2
74	From trainee to tenure-track: ten tips. Genome Biology, 2015, 16, 132.	3.8	2
75	Preeclampsia does not share common risk alleles in 9p21 with coronary artery disease and type 2 diabetes. Annals of Medicine, 2016, 48, 330-336.	1.5	2