Manuel A Rivas

List of Publications by Year
in descending order

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53	32,963	136950	168389
papers	citations	32	53
		h-index	g-index

75	75	75	68295
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	A framework for variation discovery and genotyping using next-generation DNA sequencing data. Nature Genetics, 2011, 43, 491-498.	21.4	10,018
2	Analysis of protein-coding genetic variation in 60,706 humans. Nature, 2016, 536, 285-291.	27.8	9,051
3	The Genotype-Tissue Expression (GTEx) pilot analysis: Multitissue gene regulation in humans. Science, 2015, 348, 648-660.	12.6	4,659
4	Transcriptome and genome sequencing uncovers functional variation in humans. Nature, 2013, 501, 506-511.	27.8	1,857
5	The genetic architecture of type 2 diabetes. Nature, 2016, 536, 41-47.	27.8	952
6	Landscape of X chromosome inactivation across human tissues. Nature, 2017, 550, 244-248.	27.8	764
7	Deep resequencing of GWAS loci identifies independent rare variants associated with inflammatory bowel disease. Nature Genetics, 2011, 43, 1066-1073.	21.4	698
8	Exome sequencing identifies rare LDLR and APOA5 alleles conferring risk for myocardial infarction. Nature, 2015, 518, 102-106.	27.8	581
9	Rare and low-frequency coding variants alter human adult height. Nature, 2017, 542, 186-190.	27.8	544
10	Testing for an Unusual Distribution of Rare Variants. PLoS Genetics, 2011, 7, e1001322.	3.5	530
11	Protein-altering variants associated with body mass index implicate pathways that control energy intake and expenditure in obesity. Nature Genetics, 2018, 50, 26-41.	21.4	286
12	Effect of predicted protein-truncating genetic variants on the human transcriptome. Science, 2015, 348, 666-669.	12.6	252
13	Mosaic PPM1D mutations are associated with predisposition to breast and ovarian cancer. Nature, 2013, 493, 406-410.	27.8	218
14	The landscape of genomic imprinting across diverse adult human tissues. Genome Research, 2015, 25, 927-936.	5.5	216
15	Genetic Adaptation of Fatty-Acid Metabolism: A Human-Specific Haplotype Increasing the Biosynthesis of Long-Chain Omega-3 and Omega-6 Fatty Acids. American Journal of Human Genetics, 2012, 90, 809-820.	6.2	205
16	Deep Resequencing of GWAS Loci Identifies Rare Variants in CARD9, IL23R and RNF186 That Are Associated with Ulcerative Colitis. PLoS Genetics, 2013, 9, e1003723.	3.5	185
17	Choice of transcripts and software has a large effect on variant annotation. Genome Medicine, 2014, 6, 26.	8.2	158
18	Association of Genetic Variants in NUDT15 With Thiopurine-Induced Myelosuppression in Patients With Inflammatory Bowel Disease. JAMA - Journal of the American Medical Association, 2019, 321, 773.	7.4	129

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19	The Power of Gene-Based Rare Variant Methods to Detect Disease-Associated Variation and Test Hypotheses About Complex Disease. PLoS Genetics, 2015, 11, e1005165.	3.5	124
20	Quantifying the Impact of Rare and Ultra-rare Coding Variation across the Phenotypic Spectrum. American Journal of Human Genetics, 2018, 102, 1204-1211.	6.2	102
21	Medical relevance of protein-truncating variants across 337,205 individuals in the UK Biobank study. Nature Communications, 2018, 9, 1612.	12.8	95
22	Assessing allele-specific expression across multiple tissues from RNA-seq read data. Bioinformatics, 2015, 31, 2497-2504.	4.1	90
23	Protein-coding variants implicate novel genes related to lipid homeostasis contributing to body-fat distribution. Nature Genetics, 2019, 51, 452-469.	21.4	89
24	Rare, Low-Frequency, and Common Variants in the Protein-Coding Sequence of Biological Candidate Genes from GWASs Contribute to Risk of Rheumatoid Arthritis. American Journal of Human Genetics, 2013, 92, 15-27.	6.2	83
25	Global Biobank Engine: enabling genotype-phenotype browsing for biobank summary statistics. Bioinformatics, 2019, 35, 2495-2497.	4.1	79
26	A fast and scalable framework for large-scale and ultrahigh-dimensional sparse regression with application to the UK Biobank. PLoS Genetics, 2020, 16, e1009141.	3.5	75
27	Insights into the genetic epidemiology of Crohn's and rare diseases in the Ashkenazi Jewish population. PLoS Genetics, 2018, 14, e1007329.	3.5	66
28	Phenome-wide Burden of Copy-Number Variation in the UK Biobank. American Journal of Human Genetics, 2019, 105, 373-383.	6.2	55
29	A protein-truncating R179X variant in RNF186 confers protection against ulcerative colitis. Nature Communications, 2016, 7, 12342.	12.8	50
30	Components of genetic associations across 2,138 phenotypes in the UK Biobank highlight adipocyte biology. Nature Communications, 2019, 10, 4064.	12.8	48
31	A Low-Frequency Inactivating AKT2 Variant Enriched in the Finnish Population Is Associated With Fasting Insulin Levels and Type 2 Diabetes Risk. Diabetes, 2017, 66, 2019-2032.	0.6	47
32	TMEM258 Is a Component of the Oligosaccharyltransferase Complex Controlling ER Stress and Intestinal Inflammation. Cell Reports, 2016, 17, 2955-2965.	6.4	42
33	Assessing Digital Phenotyping to Enhance Genetic Studies of Human Diseases. American Journal of Human Genetics, 2020, 106, 611-622.	6.2	42
34	Base-specific mutational intolerance near splice sites clarifies the role of nonessential splice nucleotides. Genome Research, 2018, 28, 968-974.	5.5	41
35	Discovery of rare variants for complex phenotypes. Human Genetics, 2016, 135, 625-634.	3.8	40
36	Significant sparse polygenic risk scores across 813 traits in UK Biobank. PLoS Genetics, 2022, 18, e1010105.	3.5	40

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37	Sequence data and association statistics from 12,940 type 2 diabetes cases and controls. Scientific Data, 2017, 4, 170179.	5.3	31
38	Rare protein-altering variants in ANGPTL7 lower intraocular pressure and protect against glaucoma. PLoS Genetics, 2020, 16, e1008682.	3.5	31
39	Sequence variation between 462 human individuals fine-tunes functional sites of RNA processing. Scientific Reports, 2016, 6, 32406.	3.3	28
40	Evaluating the contribution of rare variants to type 2 diabetes and related traits using pedigrees. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, 379-384.	7.1	28
41	Whole exome sequencing analyses reveal geneâ€“microbiota interactions in the context of IBD. Gut, 2021, 70, gutjnl-2019-319706.	12.1	26
42	Pooled Resequencing of 122 Ulcerative Colitis Genes in a Large Dutch Cohort Suggests Population-Specific Associations of Rare Variants in MUC2. PLoS ONE, 2016, 11, e0159609.	2.5	21
43	Nonsense-mediated decay is highly stable across individuals and tissues. American Journal of Human Genetics, 2021, 108, 1401-1408.	6.2	15
44	A Flexible Approach for the Analysis of Rare Variants Allowing for a Mixture of Effects on Binary or Quantitative Traits. PLoS Genetics, 2013, 9, e1003694.	3.5	14
45	Rare and common variant discovery in complex disease: the IBD case study. Human Molecular Genetics, 2019, 28, R162-R169.	2.9	13
46	Assessing association between protein truncating variants and quantitative traits. Bioinformatics, 2013, 29, 2419-2426.	4.1	12
47	Pooled DNA Resequencing of 68 Myocardial Infarction Candidate Genes in French Canadians. Circulation: Cardiovascular Genetics, 2012, 5, 547-554.	5.1	10
48	Mosaic mutations in blood DNA sequence are associated with solid tumor cancers. Npj Genomic Medicine, 2017, 2, 22.	3.8	10
49	A null mutation in ANGPTL8 does not associate with either plasma glucose or type 2 diabetes in humans. BMC Endocrine Disorders, 2016, 16, 7.	2.2	9
50	A Protein Domain and Family Based Approach to Rare Variant Association Analysis. PLoS ONE, 2016, 11, e0153803.	2.5	6
51	Survival analysis on rare events using group-regularized multi-response Cox regression. Bioinformatics, 2021, 37, 4437-4443.	4.1	3
52	Bayesian model comparison for rare-variant association studies. American Journal of Human Genetics, 2021, 108, 2354-2367.	6.2	2
53	Power of Rare Variant Aggregate Tests. , 2015, , 185-199.		1