Jeffrey G Reid

List of Publications by Year
in descending order

49	25,014	87888 38	182427 51
papers	citations	h-index	g-index

59	59	59	45203
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	UGT1A1 genetic variants are associated with increases in bilirubin levels in rheumatoid arthritis patients treated with sarilumab. Pharmacogenomics Journal, 2022, 22, 160-165.	2.0	1
2	Analysis of rare genetic variation underlying cardiometabolic diseases and traits among 200,000 individuals in the UK Biobank. Nature Genetics, 2022, 54, 240-250.	21.4	68
3	Genome-wide analysis provides genetic evidence that ACE2 influences COVID-19 risk and yields risk scores associated with severe disease. Nature Genetics, 2022, 54, 382-392.	21.4	97
4	Within-sibship genome-wide association analyses decrease bias in estimates of direct genetic effects. Nature Genetics, 2022, 54, 581-592.	21.4	142
5	Sparse Project VCF: efficient encoding of population genotype matrices. Bioinformatics, 2021, 36, 5537-5538.	4.1	4
6	Kidney disease genetic risk variants alter lysosomal beta-mannosidase (MANBA) expression and disease severity. Science Translational Medicine, 2021, 13, .	12.4	30
7	Exome-wide evaluation of rare coding variants using electronic health records identifies new geneâ€“phenotype associations. Nature Medicine, 2021, 27, 66-72.	30.7	44
8	Mutation spectrum of NOD2 reveals recessive inheritance as a main driver of Early Onset Crohnâ€™s Disease. Scientific Reports, 2021, 11, 5595.	3.3	29
9	Computationally efficient whole-genome regression for quantitative and binary traits. Nature Genetics, 2021, 53, 1097-1103.	21.4	457
10	Genomeâ€wide association analysis of serum alanine and aspartate aminotransferase, and the modifying effects of BMI in 388kÂEuropean individuals. Genetic Epidemiology, 2021, 45, 664-681.	1.3	9
11	Advancing human genetics research and drug discovery through exome sequencing of the UK Biobank. Nature Genetics, 2021, 53, 942-948.	21.4	234
12	Pan-ancestry exome-wide association analyses of COVID-19 outcomes in 586,157 individuals. American Journal of Human Genetics, 2021, 108, 1350-1355.	6.2	72
13	Sequencing of 640,000 exomes identifies GPR75 variants associated with protection from obesity. Science, 2021, 373, .	12.6	130
14	Exome sequencing and analysis of 454,787 UK Biobank participants. Nature, 2021, 599, 628-634.	27.8	377
15	Gene-level analysis of rare variants in 379,066 whole exome sequences identifies an association of GIGYF1 loss of function with type 2 diabetes. Scientific Reports, 2021, 11, 21565.	3.3	25
16	Clinical and Molecular Prevalence of Lipodystrophy in an Unascertained Large Clinical Care Cohort. Diabetes, 2020, 69, 249-258.	0.6	51
17	Exome sequencing and characterization of 49,960 individuals in the UK Biobank. Nature, 2020, 586, 749-756.	27.8	369
18	Identification of Neuropsychiatric Copy Number Variants in a Health Care System Population. JAMA Psychiatry, 2020, 77, 1276.	11.0	46

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19	Genome-wide association study of alcohol consumption and use disorder in 274,424 individuals from multiple populations. Nature Communications, 2019, 10, 1499.	12.8	346
20	Homozygosity for a mutation affecting the catalytic domain of tyrosyl-tRNA synthetase (YARS) causes multisystem disease. Human Molecular Genetics, 2019, 28, 525-538.	2.9	22
21	Exome Sequencing in Children With Pulmonary Arterial Hypertension Demonstrates Differences Compared With Adults. Circulation Genomic and Precision Medicine, 2018, 11, e001887.	3.6	104
22	Profiling and Leveraging Relatedness in a Precision Medicine Cohort of 92,455 Exomes. American Journal of Human Genetics, 2018, 102, 874-889.	6.2	58
23	A Protein-Truncating HSD17B13 Variant and Protection from Chronic Liver Disease. New England Journal of Medicine, 2018, 378, 1096-1106.	27.0	556
24	Genomic diagnostics within a medically underserved population: efficacy and implications. Genetics in Medicine, 2018, 20, 31-41.	2.4	47
25	The transcription factor POU3F2 regulates a gene coexpression network in brain tissue from patients with psychiatric disorders. Science Translational Medicine, 2018, 10, .	12.4	81
26	Exome Sequencingâ€"Based Screening for BRCA1/2 Expected Pathogenic Variants Among Adult Biobank Participants. JAMA Network Open, 2018, 1, e182140.	5.9	163
27	Loss-of-Function ABCC8 Mutations in Pulmonary Arterial Hypertension. Circulation Genomic and Precision Medicine, 2018, 11, e002087.	3.6	62
28	Rare variants in SOX17 are associated with pulmonary arterial hypertension with congenital heart disease. Genome Medicine, 2018, 10, 56.	8.2	112
29	Genetic inactivation of ANGPTL4 improves glucose homeostasis and is associated with reduced risk of diabetes. Nature Communications, 2018, 9, 2252.	12.8	99
30	Familial Hypercholesterolemia and Type 2 Diabetes in the Old Order Amish. Diabetes, 2017, 66, 2054-2058.	0.6	28
31	Electronic health record phenotype in subjects with genetic variants associated with arrhythmogenic right ventricular cardiomyopathy: a study of 30,716 subjects with exome sequencing. Genetics in Medicine, 2017, 19, 1245-1252.	2.4	43
32	Genetic and Pharmacologic Inactivation of ANGPTL3 and Cardiovascular Disease. New England Journal of Medicine, 2017, 377, 211-221.	27.0	633
33	Distribution and clinical impact of functional variants in 50,726 whole-exome sequences from the DiscovEHR study. Science, 2016, 354, .	12.6	464
34	Genetic identification of familial hypercholesterolemia within a single U.S. health care system. Science, 2016, 354, .	12.6	349
35	Diagnostic Yield of Clinical Tumor and Germline Whole-Exome Sequencing for Children With Solid Tumors. JAMA Oncology, 2016, 2, 616.	7.1	378
36	Inactivating Variants in ANGPTL4 and Risk of Coronary Artery Disease. New England Journal of Medicine, 2016, 374, 1123-1133.	27.0	411

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37	Good laboratory practice for clinical next-generation sequencing informatics pipelines. Nature Biotechnology, 2015, 33, 689-693.	17.5	134
38	A global reference for human genetic variation. Nature, 2015, 526, 68-74.	27.8	13,998
39	Assessing structural variation in a personal genomeâ€"towards a human reference diploid genome. BMC Genomics, 2015, 16, 286.	2.8	153
40	Next-generation sequencing identifies rare variants associated with Noonan syndrome. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 11473-11478.	7.1	158
41	Molecular Findings Among Patients Referred for Clinical Whole-Exome Sequencing. JAMA - Journal of the American Medical Association, 2014, 312, 1870.	7.4	1,171
42	Launching genomics into the cloud: deployment of Mercury, a next generation sequence analysis pipeline. BMC Bioinformatics, 2014, 15, 30.	2.6	199
43	PBHoney: identifying genomic variants via long-read discordance and interrupted mapping. BMC Bioinformatics, 2014, 15, 180.	2.6	146
44	Clinical Whole-Exome Sequencing for the Diagnosis of Mendelian Disorders. New England Journal of Medicine, 2013, 369, 1502-1511.	27.0	1,717
45	Mutations inVRK1Associated With Complex Motor and Sensory Axonal Neuropathy Plus Microcephaly. JAMA Neurology, 2013, 70, 1491-8.	9.0	54
46	Exome sequencing resolves apparent incidental findings and reveals further complexity of SH3TC2 variant alleles causing Charcot-Marie-Tooth neuropathy. Genome Medicine, 2013, 5, 57.	8.2	143
47	FOXO3 Variants Are Associated With Lower Fetal Hemoglobin Levels In Children With Sickle Cell Disease. Blood, 2013, 122, 778-778.	1.4	1
48	Whole-Genome Sequencing in a Patient with Charcotâ€"Marieâ€"Tooth Neuropathy. New England Journal of Medicine, 2010, 362, 1181-1191.	27.0	698
49	MicroRNAs in Human Ovarian Cancer.. Biology of Reproduction, 2008, 78, 200-200.	2.7	2