

# Jeffrey G Reid

## List of Publications by Year in descending order

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Version: 2024-02-01

49  
papers

25,014  
citations

87888

38  
h-index

182427

51  
g-index

59  
all docs

59  
docs citations

59  
times ranked

45203  
citing authors

#	ARTICLE	IF	CITATIONS
1	A global reference for human genetic variation. <i>Nature</i> , 2015, 526, 68-74.	27.8	13,998
2	Clinical Whole-Exome Sequencing for the Diagnosis of Mendelian Disorders. <i>New England Journal of Medicine</i> , 2013, 369, 1502-1511.	27.0	1,717
3	Molecular Findings Among Patients Referred for Clinical Whole-Exome Sequencing. <i>JAMA - Journal of the American Medical Association</i> , 2014, 312, 1870.	7.4	1,171
4	Whole-Genome Sequencing in a Patient with Charcotâ€“Marieâ€“Tooth Neuropathy. <i>New England Journal of Medicine</i> , 2010, 362, 1181-1191.	27.0	698
5	Genetic and Pharmacologic Inactivation of ANGPTL3 and Cardiovascular Disease. <i>New England Journal of Medicine</i> , 2017, 377, 211-221.	27.0	633
6	A Protein-Truncating <i>HSD17B13</i> Variant and Protection from Chronic Liver Disease. <i>New England Journal of Medicine</i> , 2018, 378, 1096-1106.	27.0	556
7	Distribution and clinical impact of functional variants in 50,726 whole-exome sequences from the DiscovEHR study. <i>Science</i> , 2016, 354, .	12.6	464
8	Computationally efficient whole-genome regression for quantitative and binary traits. <i>Nature Genetics</i> , 2021, 53, 1097-1103.	21.4	457
9	Inactivating Variants in <i>ANGPTL4</i> and Risk of Coronary Artery Disease. <i>New England Journal of Medicine</i> , 2016, 374, 1123-1133.	27.0	411
10	Diagnostic Yield of Clinical Tumor and Germline Whole-Exome Sequencing for Children With Solid Tumors. <i>JAMA Oncology</i> , 2016, 2, 616.	7.1	378
11	Exome sequencing and analysis of 454,787 UK Biobank participants. <i>Nature</i> , 2021, 599, 628-634.	27.8	377
12	Exome sequencing and characterization of 49,960 individuals in the UK Biobank. <i>Nature</i> , 2020, 586, 749-756.	27.8	369
13	Genetic identification of familial hypercholesterolemia within a single U.S. health care system. <i>Science</i> , 2016, 354, .	12.6	349
14	Genome-wide association study of alcohol consumption and use disorder in 274,424 individuals from multiple populations. <i>Nature Communications</i> , 2019, 10, 1499.	12.8	346
15	Advancing human genetics research and drug discovery through exome sequencing of the UK Biobank. <i>Nature Genetics</i> , 2021, 53, 942-948.	21.4	234
16	Launching genomics into the cloud: deployment of Mercury, a next generation sequence analysis pipeline. <i>BMC Bioinformatics</i> , 2014, 15, 30.	2.6	199
17	Exome Sequencingâ€“Based Screening for <i>BRCA1/2</i> Expected Pathogenic Variants Among Adult Biobank Participants. <i>JAMA Network Open</i> , 2018, 1, e182140.	5.9	163
18	Next-generation sequencing identifies rare variants associated with Noonan syndrome. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2014, 111, 11473-11478.	7.1	158

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19	Assessing structural variation in a personal genome towards a human reference diploid genome. BMC Genomics, 2015, 16, 286.	2.8	153
20	PBHoney: identifying genomic variants via long-read discordance and interrupted mapping. BMC Bioinformatics, 2014, 15, 180.	2.6	146
21	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
22	Within-sibship genome-wide association analyses decrease bias in estimates of direct genetic effects. Nature Genetics, 2022, 54, 581-592.	21.4	142
23	Good laboratory practice for clinical next-generation sequencing informatics pipelines. Nature Biotechnology, 2015, 33, 689-693.	17.5	134
24	Sequencing of 640,000 exomes identifies <i>GPR75</i> variants associated with protection from obesity. Science, 2021, 373, .	12.6	130
25	Rare variants in <i>SOX17</i> are associated with pulmonary arterial hypertension with congenital heart disease. Genome Medicine, 2018, 10, 56.	8.2	112
26	Exome Sequencing in Children With Pulmonary Arterial Hypertension Demonstrates Differences Compared With Adults. Circulation Genomic and Precision Medicine, 2018, 11, e001887.	3.6	104
27	Genetic inactivation of <i>ANGPTL4</i> improves glucose homeostasis and is associated with reduced risk of diabetes. Nature Communications, 2018, 9, 2252.	12.8	99
28	Genome-wide analysis provides genetic evidence that <i>ACE2</i> influences COVID-19 risk and yields risk scores associated with severe disease. Nature Genetics, 2022, 54, 382-392.	21.4	97
29	The transcription factor <i>POU3F2</i> regulates a gene coexpression network in brain tissue from patients with psychiatric disorders. Science Translational Medicine, 2018, 10, .	12.4	81
30	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
31	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
32	Loss-of-Function <i>ABCC8</i> Mutations in Pulmonary Arterial Hypertension. Circulation Genomic and Precision Medicine, 2018, 11, e002087.	3.6	62
33	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
34	Mutations in <i>VRK1</i> Associated With Complex Motor and Sensory Axonal Neuropathy Plus Microcephaly. JAMA Neurology, 2013, 70, 1491-8.	9.0	54
35	Clinical and Molecular Prevalence of Lipodystrophy in an Unascertained Large Clinical Care Cohort. Diabetes, 2020, 69, 249-258.	0.6	51
36	Genomic diagnostics within a medically underserved population: efficacy and implications. Genetics in Medicine, 2018, 20, 31-41.	2.4	47

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37	Identification of Neuropsychiatric Copy Number Variants in a Health Care System Population. <i>JAMA Psychiatry</i> , 2020, 77, 1276.	11.0	46
38	Exome-wide evaluation of rare coding variants using electronic health records identifies new gene-phenotype associations. <i>Nature Medicine</i> , 2021, 27, 66-72.	30.7	44
39	Electronic health record phenotype in subjects with genetic variants associated with arrhythmogenic right ventricular cardiomyopathy: a study of 30,716 subjects with exome sequencing. <i>Genetics in Medicine</i> , 2017, 19, 1245-1252.	2.4	43
40	Kidney disease genetic risk variants alter lysosomal beta-mannosidase ( <i>MANBA</i> ) expression and disease severity. <i>Science Translational Medicine</i> , 2021, 13, .	12.4	30
41	Mutation spectrum of NOD2 reveals recessive inheritance as a main driver of Early Onset Crohn's Disease. <i>Scientific Reports</i> , 2021, 11, 5595.	3.3	29
42	Familial Hypercholesterolemia and Type 2 Diabetes in the Old Order Amish. <i>Diabetes</i> , 2017, 66, 2054-2058.	0.6	28
43	Gene-level analysis of rare variants in 379,066 whole exome sequences identifies an association of GIGYF1 loss of function with type 2 diabetes. <i>Scientific Reports</i> , 2021, 11, 21565.	3.3	25
44	Homozygosity for a mutation affecting the catalytic domain of tyrosyl-tRNA synthetase (YARS) causes multisystem disease. <i>Human Molecular Genetics</i> , 2019, 28, 525-538.	2.9	22
45	Genome-wide association analysis of serum alanine and aspartate aminotransferase, and the modifying effects of BMI in 388k European individuals. <i>Genetic Epidemiology</i> , 2021, 45, 664-681.	1.3	9
46	Sparse Project VCF: efficient encoding of population genotype matrices. <i>Bioinformatics</i> , 2021, 36, 5537-5538.	4.1	4
47	MicroRNAs in Human Ovarian Cancer.. <i>Biology of Reproduction</i> , 2008, 78, 200-200.	2.7	2
48	FOXO3 Variants Are Associated With Lower Fetal Hemoglobin Levels In Children With Sickle Cell Disease. <i>Blood</i> , 2013, 122, 778-778.	1.4	1
49	UGT1A1 genetic variants are associated with increases in bilirubin levels in rheumatoid arthritis patients treated with sarilumab. <i>Pharmacogenomics Journal</i> , 2022, 22, 160-165.	2.0	1