Stacey Gabriel

List of Publications by Year
in descending order

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73 papers	90,275 citations	17440 63 h-index	76898 74 g-index
77 all docs	77 docs citations	77 times ranked	120030 citing authors

#	Article	IF	Citations
1	Combined tumor and immune signals from genomes or transcriptomes predict outcomes of checkpoint inhibition in melanoma. Cell Reports Medicine, 2022, 3, 100500.	6.5	13
2	Sequencing of 53,831 diverse genomes from the NHLBI TOPMed Program. Nature, 2021, 590, 290-299.	27.8	1,069
3	Proteogenomic insights into the biology and treatment of HPV-negative head and neck squamous cell carcinoma. Cancer Cell, 2021, 39, 361-379.e16.	16.8	189
4	Proteogenomic and metabolomic characterization of human glioblastoma. Cancer Cell, 2021, 39, 509-528.e20.	16.8	327
5	Proteogenomic characterization of pancreatic ductal adenocarcinoma. Cell, 2021, 184, 5031-5052.e26.	28.9	236
6	Pangenomics enables genotyping of known structural variants in 5202 diverse genomes. Science, 2021, 374, abg8871.	12.6	132
7	Genetic Interleukin 6 Signaling Deficiency Attenuates Cardiovascular Risk in Clonal Hematopoiesis. Circulation, 2020, 141, 124-131.	1.6	270
8	Inherited causes of clonal haematopoiesis in 97,691 whole genomes. Nature, 2020, 586, 763-768.	27.8	376
9	A structural variation reference for medical and population genetics. Nature, 2020, 581, 444-451.	27.8	614
10	Proteogenomic Characterization Reveals Therapeutic Vulnerabilities in Lung Adenocarcinoma. Cell, 2020, 182, 200-225.e35.	28.9	410
11	Proteogenomic Characterization of Endometrial Carcinoma. Cell, 2020, 180, 729-748.e26.	28.9	296
12	A missense variant in Mitochondrial Amidoxime Reducing Component 1 gene and protection against liver disease. PLoS Genetics, 2020, 16, e1008629.	3.5	101
13	Integrated Proteogenomic Characterization of Clear Cell Renal Cell Carcinoma. Cell, 2019, 179, 964-983.e31.	28.9	430
14	Harmonizing Clinical Sequencing and Interpretation for the eMERGE III Network. American Journal of Human Genetics, 2019, 105, 588-605.	6.2	99
15	Molecular Genetic Anatomy and Risk Profile of Hirschsprungâ€™s Disease. New England Journal of Medicine, 2019, 380, 1421-1432.	27.0	131
16	DNA Sequence Variation in ACVR1C Encoding the Activin Receptor-Like Kinase 7 Influences Body Fat Distribution and Protects Against Type 2 Diabetes. Diabetes, 2019, 68, 226-234.	0.6	31
17	Analysis of predicted loss-of-function variants in UK Biobank identifies variants protective for disease. Nature Communications, 2018, 9, 1613.	12.8	78
18	Association Between Titin Loss-of-Function Variants and Early-Onset Atrial Fibrillation. JAMA - Journal of the American Medical Association, 2018, 320, 2354.	7.4	144

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19	Characterization and remediation of sample index swaps by non-redundant dual indexing on massively parallel sequencing platforms. BMC Genomics, 2018, 19, 332.	2.8	201
20	Structural Alterations Driving Castration-Resistant Prostate Cancer Revealed by Linked-Read Genome Sequencing. Cell, 2018, 174, 433-447.e19.	28.9	258
21	Association of Rare and Common Variation in the Lipoprotein Lipase Gene With Coronary Artery Disease. JAMA - Journal of the American Medical Association, 2017, 317, 937.	7.4	148
22	Human knockouts and phenotypic analysis in a cohort with a high rate of consanguinity. Nature, 2017, 544, 235-239.	27.8	292
23	ANGPTL3 Deficiency and Protection Against Coronary Artery Disease. Journal of the American College of Cardiology, 2017, 69, 2054-2063.	2.8	348
24	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
25	Analysis commons, a team approach to discovery in a big-data environment for genetic epidemiology. Nature Genetics, 2017, 49, 1560-1563.	21.4	93
26	Sequence data and association statistics from 12,940 type 2 diabetes cases and controls. Scientific Data, 2017, 4, 170179.	5.3	31
27	The genetic architecture of type 2 diabetes. Nature, 2016, 536, 41-47.	27.8	952
28	Genomic evolution and chemoresistance in germ-cell tumours. Nature, 2016, 540, 114-118.	27.8	139
29	Phenotypic Characterization of GeneticallyÂLowered Human Lipoprotein(a) Levels. Journal of the American College of Cardiology, 2016, 68, 2761-2772.	2.8	186
30	Diagnostic Yield and Clinical Utility of Sequencing Familial Hypercholesterolemia Genes in Patients With Severe Hypercholesterolemia. Journal of the American College of Cardiology, 2016, 67, 2578-2589.	2.8	723
31	A reference panel of 64,976 haplotypes for genotype imputation. Nature Genetics, 2016, 48, 1279-1283.	21.4	2,421
32	Association of Exome Sequences With Cardiovascular Traits Among Blacks in the Jackson Heart Study. Circulation: Cardiovascular Genetics, 2016, 9, 368-374.	5.1	8
33	Clonal evolution in patients with chronic lymphocytic leukaemia developing resistance to BTK inhibition. Nature Communications, 2016, 7, 11589.	12.8	285
34	Rare variant associations with waist-to-hip ratio in European-American and African-American women from the NHLBI-Exome Sequencing Project. European Journal of Human Genetics, 2016, 24, 1181-1187.	2.8	5
35	Phenotypic extremes in rare variant study designs. European Journal of Human Genetics, 2016, 24, 924-930.	2.8	65
36	Whole-genome and multisector exome sequencing of primary and post-treatment glioblastoma reveals patterns of tumor evolution. Genome Research, 2015, 25, 316-327.	5.5	343

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37	Comprehensive, Integrative Genomic Analysis of Diffuse Lower-Grade Gliomas. New England Journal of Medicine, 2015, 372, 2481-2498.	27.0	2,582
38	Identification and Functional Characterization of G6PC2 Coding Variants Influencing Glycemic Traits Define an Effector Transcript at the G6PC2-ABCB11 Locus. PLoS Genetics, 2015, 11, e1004876.	3.5	95
39	Genomic Characterization of Brain Metastases Reveals Branched Evolution and Potential Therapeutic Targets. Cancer Discovery, 2015, 5, 1164-1177.	9.4	821
40	Genomic correlates of response to CTLA-4 blockade in metastatic melanoma. Science, 2015, 350, 207-211.	12.6	2,275
41	Exome sequencing identifies rare LDLR and APOA5 alleles conferring risk for myocardial infarction. Nature, 2015, 518, 102-106.	27.8	581
42	Somatic ERCC2 Mutations Correlate with Cisplatin Sensitivity in Muscle-Invasive Urothelial Carcinoma. Cancer Discovery, 2014, 4, 1140-1153.	9.4	506
43	Distribution and Medical Impact of Loss-of-Function Variants in the Finnish Founder Population. PLoS Genetics, 2014, 10, e1004494.	3.5	351
44	Age-Related Clonal Hematopoiesis Associated with Adverse Outcomes. New England Journal of Medicine, 2014, 371, 2488-2498.	27.0	3,474
45	Systematic identification of personal tumor-specific neoantigens in chronic lymphocytic leukemia. Blood, 2014, 124, 453-462.	1.4	286
46	Genetic and Clonal Dissection of Murine Small Cell Lung Carcinoma Progression by Genome Sequencing. Cell, 2014, 156, 1298-1311.	28.9	241
47	Whole-exome sequencing and clinical interpretation of formalin-fixed, paraffin-embedded tumor samples to guide precision cancer medicine. Nature Medicine, 2014, 20, 682-688.	30.7	508
48	A polygenic burden of rare disruptive mutations in schizophrenia. Nature, 2014, 506, 185-190.	27.8	1,305
49	Alzheimer's disease: early alterations in brain DNA methylation at ANK1, BIN1, RHBDF2 and other loci. Nature Neuroscience, 2014, 17, 1156-1163.	14.8	800
50	Loss-of-function mutations in SLC30A8 protect against type 2 diabetes. Nature Genetics, 2014, 46, 357-363.	21.4	428
51	The Somatic Genomic Landscape of Glioblastoma. Cell, 2013, 155, 462-477.	28.9	3,979
52	Evolution and Impact of Subclonal Mutations in Chronic Lymphocytic Leukemia. Cell, 2013, 152, 714-726.	28.9	1,202
53	From FastQ Data to Highâ€Confidence Variant Calls: The Genome Analysis Toolkit Best Practices Pipeline. Current Protocols in Bioinformatics, 2013, 43, 11.10.1-11.10.33.	25.8	4,796
54	Sensitive detection of somatic point mutations in impure and heterogeneous cancer samples. Nature Biotechnology, 2013, 31, 213-219.	17.5	3,934

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55	Prognostically relevant gene signatures of high-grade serous ovarian carcinoma. Journal of Clinical Investigation, 2013, 123, 517-25.	8.2	462
56	Advances in genetics show the need for extending screening strategies for autosomal dominant hypercholesterolaemia. European Heart Journal, 2012, 33, 1360-1366.	2.2	76
57	Patterns and rates of exonic de novo mutations in autism spectrum disorders. Nature, 2012, 485, 242-245.	27.8	1,597
58	SF3B1and Other Novel Cancer Genes in Chronic Lymphocytic Leukemia. New England Journal of Medicine, 2011, 365, 2497-2506.	27.0	1,021
59	Comparing strategies to fine-map the association of common SNPs at chromosome 9p21 with type 2 diabetes and myocardial infarction. Nature Genetics, 2011, 43, 801-805.	21.4	79
60	Genomic sequencing of colorectal adenocarcinomas identifies a recurrent VTI1A-TCF7L2 fusion. Nature Genetics, 2011, 43, 964-968.	21.4	270
61	A scalable, fully automated process for construction of sequence-ready human exome targeted capture libraries. Genome Biology, 2011, 12, R1.	9.6	547
62	Integrated Genomic Analysis Identifies Clinically Relevant Subtypes of Glioblastoma Characterized by Abnormalities in PDGFRA, IDH1, EGFR, and NF1. Cancer Cell, 2010, 17, 98-110.	16.8	6,138
63	The landscape of somatic copy-number alteration across human cancers. Nature, 2010, 463, 899-905.	27.8	3,331
64	Advances in understanding cancer genomes through second-generation sequencing. Nature Reviews Genetics, 2010, 11, 685-696.	16.3	1,014
65	The Genome Analysis Toolkit: A MapReduce framework for analyzing next-generation DNA sequencing data. Genome Research, 2010, 20, 1297-1303.	5.5	21,358
66	Solution hybrid selection with ultra-long oligonucleotides for massively parallel targeted sequencing. Nature Biotechnology, 2009, 27, 182-189.	17.5	1,267
67	Drug-sensitiveFGFR2mutations in endometrial carcinoma. Proceedings of the National Academy of Sciences of the United States of America, 2008, 105, 8713-8717.	7.1	329
68	High-throughput oncogene mutation profiling in human cancer. Nature Genetics, 2007, 39, 347-351.	21.4	927
69	Population Genetic Tools: Application to Cancer. Seminars in Oncology, 2007, 34, S21-S24.	2.2	1
70	Variation in the Human Genome and the Inherited Basis of Common Disease. Seminars in Oncology, 2006, 33, 46-49.	2.2	7
71	Epidermal Growth Factor Receptor Activation in Glioblastoma through Novel Missense Mutations in the Extracellular Domain. PLoS Medicine, 2006, 3, e485.	8.4	298
72	Activating mutation in the tyrosine kinase JAK2 in polycythemia vera, essential thrombocythemia, and myeloid metaplasia with myelofibrosis. Cancer Cell, 2005, 7, 387-397.	16.8	2,695

#	Article	IF	Citations
73	EGFR Mutations in Lung Cancer: Correlation with Clinical Response to Gefitinib Therapy. Science, 2004, 304, 1497-1500.	12.6	9,038