

# Stacey Gabriel

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/10705386/publications.pdf>

Version: 2024-02-01

73  
papers

90,275  
citations

17440

63  
h-index

76900

74  
g-index

77  
all docs

77  
docs citations

77  
times ranked

120030  
citing authors

| #  | ARTICLE   | IF   | CITATIONS |
|----|---|------|-----------|
| 1  | The Genome Analysis Toolkit: A MapReduce framework for analyzing next-generation DNA sequencing data. <i>Genome Research</i> , 2010, 20, 1297-1303.                                       | 5.5  | 21,358    |
| 2  | <i>EGFR</i> Mutations in Lung Cancer: Correlation with Clinical Response to Gefitinib Therapy. <i>Science</i> , 2004, 304, 1497-1500.   | 12.6 | 9,038     |
| 3  | Integrated Genomic Analysis Identifies Clinically Relevant Subtypes of Glioblastoma Characterized by Abnormalities in PDGFRA, IDH1, EGFR, and NF1. <i>Cancer Cell</i> , 2010, 17, 98-110. | 16.8 | 6,138     |
| 4  | From FastQ Data to High-Confidence Variant Calls: The Genome Analysis Toolkit Best Practices Pipeline. <i>Current Protocols in Bioinformatics</i> , 2013, 43, 11.10.1-11.10.33.           | 25.8 | 4,796     |
| 5  | The Somatic Genomic Landscape of Glioblastoma. <i>Cell</i> , 2013, 155, 462-477.  | 28.9 | 3,979     |
| 6  | Sensitive detection of somatic point mutations in impure and heterogeneous cancer samples. <i>Nature Biotechnology</i> , 2013, 31, 213-219.   | 17.5 | 3,934     |
| 7  | Age-Related Clonal Hematopoiesis Associated with Adverse Outcomes. <i>New England Journal of Medicine</i> , 2014, 371, 2488-2498.   | 27.0 | 3,474     |
| 8  | The landscape of somatic copy-number alteration across human cancers. <i>Nature</i> , 2010, 463, 899-905.   | 27.8 | 3,331     |
| 9  | Activating mutation in the tyrosine kinase JAK2 in polycythemia vera, essential thrombocythemia, and myeloid metaplasia with myelofibrosis. <i>Cancer Cell</i> , 2005, 7, 387-397.        | 16.8 | 2,695     |
| 10 | Comprehensive, Integrative Genomic Analysis of Diffuse Lower-Grade Gliomas. <i>New England Journal of Medicine</i> , 2015, 372, 2481-2498.  | 27.0 | 2,582     |
| 11 | A reference panel of 64,976 haplotypes for genotype imputation. <i>Nature Genetics</i> , 2016, 48, 1279-1283.   | 21.4 | 2,421     |
| 12 | Genomic correlates of response to CTLA-4 blockade in metastatic melanoma. <i>Science</i> , 2015, 350, 207-211.  | 12.6 | 2,275     |
| 13 | Patterns and rates of exonic de novo mutations in autism spectrum disorders. <i>Nature</i> , 2012, 485, 242-245.  | 27.8 | 1,597     |
| 14 | A polygenic burden of rare disruptive mutations in schizophrenia. <i>Nature</i> , 2014, 506, 185-190.   | 27.8 | 1,305     |
| 15 | Solution hybrid selection with ultra-long oligonucleotides for massively parallel targeted sequencing. <i>Nature Biotechnology</i> , 2009, 27, 182-189.                                   | 17.5 | 1,267     |
| 16 | Evolution and Impact of Subclonal Mutations in Chronic Lymphocytic Leukemia. <i>Cell</i> , 2013, 152, 714-726.  | 28.9 | 1,202     |
| 17 | Sequencing of 53,831 diverse genomes from the NHLBI TOPMed Program. <i>Nature</i> , 2021, 590, 290-299.   | 27.8 | 1,069     |
| 18 | <i>SF3B1</i> and Other Novel Cancer Genes in Chronic Lymphocytic Leukemia. <i>New England Journal of Medicine</i> , 2011, 365, 2497-2506.   | 27.0 | 1,021     |

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|----|---|------|-----------|
| 19 | Advances in understanding cancer genomes through second-generation sequencing. <i>Nature Reviews Genetics</i> , 2010, 11, 685-696.  | 16.3 | 1,014     |
| 20 | The genetic architecture of type 2 diabetes. <i>Nature</i> , 2016, 536, 41-47.  | 27.8 | 952       |
| 21 | High-throughput oncogene mutation profiling in human cancer. <i>Nature Genetics</i> , 2007, 39, 347-351.  | 21.4 | 927       |
| 22 | Genomic Characterization of Brain Metastases Reveals Branched Evolution and Potential Therapeutic Targets. <i>Cancer Discovery</i> , 2015, 5, 1164-1177.  | 9.4  | 821       |
| 23 | Alzheimer's disease: early alterations in brain DNA methylation at ANK1, BIN1, RHBDF2 and other loci. <i>Nature Neuroscience</i> , 2014, 17, 1156-1163.   | 14.8 | 800       |
| 24 | Diagnostic Yield and Clinical Utility of Sequencing Familial Hypercholesterolemia Genes in Patients With Severe Hypercholesterolemia. <i>Journal of the American College of Cardiology</i> , 2016, 67, 2578-2589. | 2.8  | 723       |
| 25 | A structural variation reference for medical and population genetics. <i>Nature</i> , 2020, 581, 444-451.   | 27.8 | 614       |
| 26 | Exome sequencing identifies rare LDLR and APOA5 alleles conferring risk for myocardial infarction. <i>Nature</i> , 2015, 518, 102-106.  | 27.8 | 581       |
| 27 | A scalable, fully automated process for construction of sequence-ready human exome targeted capture libraries. <i>Genome Biology</i> , 2011, 12, R1.  | 9.6  | 547       |
| 28 | Whole-exome sequencing and clinical interpretation of formalin-fixed, paraffin-embedded tumor samples to guide precision cancer medicine. <i>Nature Medicine</i> , 2014, 20, 682-688.                             | 30.7 | 508       |
| 29 | Somatic ERCC2 Mutations Correlate with Cisplatin Sensitivity in Muscle-Invasive Urothelial Carcinoma. <i>Cancer Discovery</i> , 2014, 4, 1140-1153.   | 9.4  | 506       |
| 30 | Prognostically relevant gene signatures of high-grade serous ovarian carcinoma. <i>Journal of Clinical Investigation</i> , 2013, 123, 517-25.   | 8.2  | 462       |
| 31 | Integrated Proteogenomic Characterization of Clear Cell Renal Cell Carcinoma. <i>Cell</i> , 2019, 179, 964-983.e31.   | 28.9 | 430       |
| 32 | Loss-of-function mutations in SLC30A8 protect against type 2 diabetes. <i>Nature Genetics</i> , 2014, 46, 357-363.  | 21.4 | 428       |
| 33 | Proteogenomic Characterization Reveals Therapeutic Vulnerabilities in Lung Adenocarcinoma. <i>Cell</i> , 2020, 182, 200-225.e35.  | 28.9 | 410       |
| 34 | Inherited causes of clonal haematopoiesis in 97,691 whole genomes. <i>Nature</i> , 2020, 586, 763-768.  | 27.8 | 376       |
| 35 | Distribution and Medical Impact of Loss-of-Function Variants in the Finnish Founder Population. <i>PLoS Genetics</i> , 2014, 10, e1004494.  | 3.5  | 351       |
| 36 | ANGPTL3 Deficiency and Protection Against Coronary Artery Disease. <i>Journal of the American College of Cardiology</i> , 2017, 69, 2054-2063.  | 2.8  | 348       |

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|----|---|------|-----------|
| 37 | Whole-genome and multisector exome sequencing of primary and post-treatment glioblastoma reveals patterns of tumor evolution. <i>Genome Research</i> , 2015, 25, 316-327.         | 5.5  | 343       |
| 38 | Drug-sensitive <i>FGFR2</i> mutations in endometrial carcinoma. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2008, 105, 8713-8717.    | 7.1  | 329       |
| 39 | Proteogenomic and metabolomic characterization of human glioblastoma. <i>Cancer Cell</i> , 2021, 39, 509-528.e20.   | 16.8 | 327       |
| 40 | Epidermal Growth Factor Receptor Activation in Glioblastoma through Novel Missense Mutations in the Extracellular Domain. <i>PLoS Medicine</i> , 2006, 3, e485.                   | 8.4  | 298       |
| 41 | Proteogenomic Characterization of Endometrial Carcinoma. <i>Cell</i> , 2020, 180, 729-748.e26.  | 28.9 | 296       |
| 42 | Human knockouts and phenotypic analysis in a cohort with a high rate of consanguinity. <i>Nature</i> , 2017, 544, 235-239.  | 27.8 | 292       |
| 43 | Systematic identification of personal tumor-specific neoantigens in chronic lymphocytic leukemia. <i>Blood</i> , 2014, 124, 453-462.  | 1.4  | 286       |
| 44 | Clonal evolution in patients with chronic lymphocytic leukaemia developing resistance to BTK inhibition. <i>Nature Communications</i> , 2016, 7, 11589.                           | 12.8 | 285       |
| 45 | Genomic sequencing of colorectal adenocarcinomas identifies a recurrent <i>VTI1A-TCF7L2</i> fusion. <i>Nature Genetics</i> , 2011, 43, 964-968.                                   | 21.4 | 270       |
| 46 | Genetic Interleukin 6 Signaling Deficiency Attenuates Cardiovascular Risk in Clonal Hematopoiesis. <i>Circulation</i> , 2020, 141, 124-131.                                       | 1.6  | 270       |
| 47 | Structural Alterations Driving Castration-Resistant Prostate Cancer Revealed by Linked-Read Genome Sequencing. <i>Cell</i> , 2018, 174, 433-447.e19.                              | 28.9 | 258       |
| 48 | Genetic and Clonal Dissection of Murine Small Cell Lung Carcinoma Progression by Genome Sequencing. <i>Cell</i> , 2014, 156, 1298-1311.   | 28.9 | 241       |
| 49 | Proteogenomic characterization of pancreatic ductal adenocarcinoma. <i>Cell</i> , 2021, 184, 5031-5052.e26.   | 28.9 | 236       |
| 50 | Characterization and remediation of sample index swaps by non-redundant dual indexing on massively parallel sequencing platforms. <i>BMC Genomics</i> , 2018, 19, 332.            | 2.8  | 201       |
| 51 | Proteogenomic insights into the biology and treatment of HPV-negative head and neck squamous cell carcinoma. <i>Cancer Cell</i> , 2021, 39, 361-379.e16.                          | 16.8 | 189       |
| 52 | Phenotypic Characterization of Genetically Lowered Human Lipoprotein(a) Levels. <i>Journal of the American College of Cardiology</i> , 2016, 68, 2761-2772.                       | 2.8  | 186       |
| 53 | Association of Rare and Common Variation in the Lipoprotein Lipase Gene With Coronary Artery Disease. <i>JAMA - Journal of the American Medical Association</i> , 2017, 317, 937. | 7.4  | 148       |
| 54 | Association Between Titin Loss-of-Function Variants and Early-Onset Atrial Fibrillation. <i>JAMA - Journal of the American Medical Association</i> , 2018, 320, 2354.             | 7.4  | 144       |

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|----|---|------|-----------|
| 55 | Genomic evolution and chemoresistance in germ-cell tumours. <i>Nature</i> , 2016, 540, 114-118.   | 27.8 | 139       |
| 56 | Pangenomics enables genotyping of known structural variants in 5202 diverse genomes. <i>Science</i> , 2021, 374, abg8871.   | 12.6 | 132       |
| 57 | Molecular Genetic Anatomy and Risk Profile of Hirschsprung's Disease. <i>New England Journal of Medicine</i> , 2019, 380, 1421-1432.  | 27.0 | 131       |
| 58 | A missense variant in Mitochondrial Amidoxime Reducing Component 1 gene and protection against liver disease. <i>PLoS Genetics</i> , 2020, 16, e1008629.  | 3.5  | 101       |
| 59 | Harmonizing Clinical Sequencing and Interpretation for the eMERGE III Network. <i>American Journal of Human Genetics</i> , 2019, 105, 588-605.  | 6.2  | 99        |
| 60 | Identification and Functional Characterization of G6PC2 Coding Variants Influencing Glycemic Traits Define an Effector Transcript at the G6PC2-ABCB11 Locus. <i>PLoS Genetics</i> , 2015, 11, e1004876.     | 3.5  | 95        |
| 61 | Analysis commons, a team approach to discovery in a big-data environment for genetic epidemiology. <i>Nature Genetics</i> , 2017, 49, 1560-1563.  | 21.4 | 93        |
| 62 | Comparing strategies to fine-map the association of common SNPs at chromosome 9p21 with type 2 diabetes and myocardial infarction. <i>Nature Genetics</i> , 2011, 43, 801-805.                              | 21.4 | 79        |
| 63 | Analysis of predicted loss-of-function variants in UK Biobank identifies variants protective for disease. <i>Nature Communications</i> , 2018, 9, 1613.   | 12.8 | 78        |
| 64 | Advances in genetics show the need for extending screening strategies for autosomal dominant hypercholesterolaemia. <i>European Heart Journal</i> , 2012, 33, 1360-1366.                                    | 2.2  | 76        |
| 65 | Phenotypic extremes in rare variant study designs. <i>European Journal of Human Genetics</i> , 2016, 24, 924-930.   | 2.8  | 65        |
| 66 | A Low-Frequency Inactivating <i>AKT2</i> Variant Enriched in the Finnish Population Is Associated With Fasting Insulin Levels and Type 2 Diabetes Risk. <i>Diabetes</i> , 2017, 66, 2019-2032.              | 0.6  | 47        |
| 67 | Sequence data and association statistics from 12,940 type 2 diabetes cases and controls. <i>Scientific Data</i> , 2017, 4, 170179.  | 5.3  | 31        |
| 68 | DNA Sequence Variation in <i>ACVR1C</i> Encoding the Activin Receptor-Like Kinase 7 Influences Body Fat Distribution and Protects Against Type 2 Diabetes. <i>Diabetes</i> , 2019, 68, 226-234.             | 0.6  | 31        |
| 69 | Combined tumor and immune signals from genomes or transcriptomes predict outcomes of checkpoint inhibition in melanoma. <i>Cell Reports Medicine</i> , 2022, 3, 100500.                                     | 6.5  | 13        |
| 70 | Association of Exome Sequences With Cardiovascular Traits Among Blacks in the Jackson Heart Study. <i>Circulation: Cardiovascular Genetics</i> , 2016, 9, 368-374.  | 5.1  | 8         |
| 71 | Variation in the Human Genome and the Inherited Basis of Common Disease. <i>Seminars in Oncology</i> , 2006, 33, 46-49.   | 2.2  | 7         |
| 72 | Rare variant associations with waist-to-hip ratio in European-American and African-American women from the NHLBI-Exome Sequencing Project. <i>European Journal of Human Genetics</i> , 2016, 24, 1181-1187. | 2.8  | 5         |

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|----|---|-----|-----------|
| 73 | Population Genetic Tools: Application to Cancer. <i>Seminars in Oncology</i> , 2007, 34, S21-S24. | 2.2 | 1         |