# Francis S Collins

List of Publications by Year
in descending order



| | | | |
|---|---|---|---|
| 129 papers | 77,235 citations | 9264 74 h-index | 11939 134 g-index |
| 141 all docs | 141 docs citations | 141 times ranked | 75410 citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Initial sequencing and analysis of the human genome. Nature, 2001, 409, 860-921. | 27.8 | 21,074 |
| 2 | Identification and analysis of functional elements in 1% of the human genome by the ENCODE pilot project. Nature, 2007, 447, 799-816. | 27.8 | 4,709 |
| 3 | Genetic studies of body mass index yield new insights for obesity biology. Nature, 2015, 518, 197-206. | 27.8 | 3,823 |
| 4 | Potential etiologic and functional implications of genome-wide association loci for human diseases and traits. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 9362-9367. | 7.1 | 3,719 |
| 5 | Biological, clinical and population relevance of 95 loci for blood lipids. Nature, 2010, 466, 707-713. | 27.8 | 3,249 |
| 6 | Discovery and refinement of loci associated with lipid levels. Nature Genetics, 2013, 45, 1274-1283. | 21.4 | 2,641 |
| 7 | A reference panel of 64,976 haplotypes for genotype imputation. Nature Genetics, 2016, 48, 1279-1283. | 21.4 | 2,421 |
| 8 | Recurrent de novo point mutations in lamin A cause Hutchinsonâ€“Gilford progeria syndrome. Nature, 2003, 423, 293-298. | 27.8 | 1,925 |
| 9 | Genetic variants in novel pathways influence blood pressure and cardiovascular disease risk. Nature, 2011, 478, 103-109. | 27.8 | 1,855 |
| 10 | Large-scale association analysis provides insights into the genetic architecture and pathophysiology of type 2 diabetes. Nature Genetics, 2012, 44, 981-990. | 21.4 | 1,748 |
| 11 | Twelve type 2 diabetes susceptibility loci identified through large-scale association analysis. Nature Genetics, 2010, 42, 579-589. | 21.4 | 1,631 |
| 12 | The Path to Personalized Medicine. New England Journal of Medicine, 2010, 363, 301-304. | 27.0 | 1,595 |
| 13 | Newly identified loci that influence lipid concentrations and risk of coronary artery disease. Nature Genetics, 2008, 40, 161-169. | 21.4 | 1,488 |
| 14 | Fine-mapping type 2 diabetes loci to single-variant resolution using high-density imputation and islet-specific epigenome maps. Nature Genetics, 2018, 50, 1505-1513. | 21.4 | 1,331 |
| 15 | New genetic loci link adipose and insulin biology to body fat distribution. Nature, 2015, 518, 187-196. | 27.8 | 1,328 |
| 16 | Common variants at 30 loci contribute to polygenic dyslipidemia. Nature Genetics, 2009, 41, 56-65. | 21.4 | 1,234 |
| 17 | Accumulation of mutant lamin A causes progressive changes in nuclear architecture in Hutchinsonâ€“Gilford progeria syndrome. Proceedings of the National Academy of Sciences of the United States of America, 2004, 101, 8963-8968. | 7.1 | 988 |
| 18 | The Human Genome Project: Lessons from Large-Scale Biology. Science, 2003, 300, 286-290. | 12.6 | 959 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Genome-wide trans-ancestry meta-analysis provides insight into the genetic architecture of type 2 diabetes susceptibility. Nature Genetics, 2014, 46, 234-244. | 21.4 | 959 |
| 20 | The genetic architecture of type 2 diabetes. Nature, 2016, 536, 41-47. | 27.8 | 952 |
| 21 | Genetic analysis of over 1 million people identifies 535 new loci associated with blood pressure traits. Nature Genetics, 2018, 50, 1412-1425. | 21.4 | 924 |
| 22 | A genome-wide approach accounting for body mass index identifies genetic variants influencing fasting glycemic traits and insulin resistance. Nature Genetics, 2012, 44, 659-669. | 21.4 | 762 |
| 23 | Large-scale association analyses identify new loci influencing glycemic traits and provide insight into the underlying biological pathways. Nature Genetics, 2012, 44, 991-1005. | 21.4 | 746 |
| 24 | Phenotype and Course of Hutchinsonâ€“Gilford Progeria Syndrome. New England Journal of Medicine, 2008, 358, 592-604. | 27.0 | 610 |
| 25 | Chromatin stretch enhancer states drive cell-specific gene regulation and harbor human disease risk variants. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 17921-17926. | 7.1 | 606 |
| 26 | Transforming Environmental Health Protection. Science, 2008, 319, 906-907. | 12.6 | 580 |
| 27 | Rare and low-frequency coding variants alter human adult height. Nature, 2017, 542, 186-190. | 27.8 | 544 |
| 28 | Human laminopathies: nuclei gone genetically awry. Nature Reviews Genetics, 2006, 7, 940-952. | 16.3 | 478 |
| 29 | Novel Loci for Adiponectin Levels and Their Influence on Type 2 Diabetes and Metabolic Traits: A Multi-Ethnic Meta-Analysis of 45,891 Individuals. PLoS Genetics, 2012, 8, e1002607. | 3.5 | 419 |
| 30 | Reengineering Translational Science: The Time Is Right. Science Translational Medicine, 2011, 3, 90cm17. | 12.4 | 409 |
| 31 | Genome-wide association study identifies six new loci influencing pulse pressure and mean arterial pressure. Nature Genetics, 2011, 43, 1005-1011. | 21.4 | 403 |
| 32 | The genetics of blood pressure regulation and its target organs from association studies in 342,415 individuals. Nature Genetics, 2016, 48, 1171-1184. | 21.4 | 362 |
| 33 | Refining the accuracy of validated target identification through coding variant fine-mapping in type 2 diabetes. Nature Genetics, 2018, 50, 559-571. | 21.4 | 356 |
| 34 | The trans-ancestral genomic architecture of glycemic traits. Nature Genetics, 2021, 53, 840-860. | 21.4 | 341 |
| 35 | Genome-Wide Association Identifies Nine Common Variants Associated With Fasting Proinsulin Levels and Provides New Insights Into the Pathophysiology of Type 2 Diabetes. Diabetes, 2011, 60, 2624-2634. | 0.6 | 335 |
| 36 | Inhibiting farnesylation of progerin prevents the characteristic nuclear blebbing of Hutchinson-Gilford progeria syndrome. Proceedings of the National Academy of Sciences of the United States of America, 2005, 102, 12879-12884. | 7.1 | 334 |

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|---|---|---|---|
| 37 | Cardiovascular Pathology in Hutchinson-Gilford Progeria: Correlation With the Vascular Pathology of Aging. Arteriosclerosis, Thrombosis, and Vascular Biology, 2010, 30, 2301-2309. | 2.4 | 332 |
| 38 | The Influence of Age and Sex on Genetic Associations with Adult Body Size and Shape: A Large-Scale Genome-Wide Interaction Study. PLoS Genetics, 2015, 11, e1005378. | 3.5 | 331 |
| 39 | Super-enhancers delineate disease-associated regulatory nodes in T cells. Nature, 2015, 520, 558-562. | 27.8 | 323 |
| 40 | The Mutant Form of Lamin A that Causes Hutchinson-Gilford Progeria Is a Biomarker of Cellular Aging in Human Skin. PLoS ONE, 2007, 2, e1269. | 2.5 | 305 |
| 41 | Impact of Type 2 Diabetes Susceptibility Variants on Quantitative Glycemic Traits Reveals Mechanistic Heterogeneity. Diabetes, 2014, 63, 2158-2171. | 0.6 | 297 |
| 42 | Rapamycin Reverses Cellular Phenotypes and Enhances Mutant Protein Clearance in Hutchinson-Gilford Progeria Syndrome Cells. Science Translational Medicine, 2011, 3, 89ra58. | 12.4 | 294 |
| 43 | The case for a US prospective cohort study of genes and environment. Nature, 2004, 429, 475-477. | 27.8 | 290 |
| 44 | 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 |
| 45 | In vivo base editing rescues Hutchinsonâ€“Gilford progeria syndrome in mice. Nature, 2021, 589, 608-614. | 27.8 | 275 |
| 46 | Trans-ancestry meta-analyses identify rare and common variants associated with blood pressure and hypertension. Nature Genetics, 2016, 48, 1151-1161. | 21.4 | 261 |
| 47 | Progressive vascular smooth muscle cell defects in a mouse model of Hutchinson-Gilford progeria syndrome. Proceedings of the National Academy of Sciences of the United States of America, 2006, 103, 3250-3255. | 7.1 | 255 |
| 48 | Multi-ancestry genetic study of type 2 diabetes highlights the power of diverse populations for discovery and translation. Nature Genetics, 2022, 54, 560-572. | 21.4 | 250 |
| 49 | Exome sequencing of 20,791Âcases of type 2 diabetes and 24,440Âcontrols. Nature, 2019, 570, 71-76. | 27.8 | 248 |
| 50 | Has the revolution arrived?. Nature, 2010, 464, 674-675. | 27.8 | 243 |
| 51 | A lamin A protein isoform overexpressed in Hutchinsonâ€“Gilford progeria syndrome interferes with mitosis in progeria and normal cells. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 4949-4954. | 7.1 | 235 |
| 52 | Systematic comparison of three genomic enrichment methods for massively parallel DNA sequencing. Genome Research, 2010, 20, 1420-1431. | 5.5 | 194 |
| 53 | Genetic regulatory signatures underlying islet gene expression and type 2 diabetes. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, 2301-2306. | 7.1 | 189 |
| 54 | A progeria mutation reveals functions for lamin A in nuclear assembly, architecture, and chromosome organization. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 20788-20793. | 7.1 | 185 |

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|---|---|---|---|
| 55 | A farnesyltransferase inhibitor prevents both the onset and late progression of cardiovascular disease in a progeria mouse model. Proceedings of the National Academy of Sciences of the United States of America, 2008, 105, 15902-15907. | 7.1 | 181 |
| 56 | Directional dominance on stature and cognition inÂdiverse human populations. Nature, 2015, 523, 459-462. | 27.8 | 173 |
| 57 | Genome-wide meta-analysis of 241,258 adults accounting for smoking behaviour identifies novel loci for obesity traits. Nature Communications, 2017, 8, 14977. | 12.8 | 169 |
| 58 | Patient-Centered Outcomes Research Institute: The Intersection of Science and Health Care. Science Translational Medicine, 2010, 2, 37cm18. | 12.4 | 168 |
| 59 | Genome-wide physical activity interactions in adiposity â€• A meta-analysis of 200,452 adults. PLoS Genetics, 2017, 13, e1006528. | 3.5 | 158 |
| 60 | The Metabolic Syndrome in Men study: a resource for studies of metabolic and cardiovascular diseases. Journal of Lipid Research, 2017, 58, 481-493. | 4.2 | 147 |
| 61 | Large meta-analysis of genome-wide association studies identifies five loci for lean body mass. Nature Communications, 2017, 8, 80. | 12.8 | 147 |
| 62 | Genetic Regulation of Adipose Gene Expression and Cardio-Metabolic Traits. American Journal of Human Genetics, 2017, 100, 428-443. | 6.2 | 141 |
| 63 | Novel Blood Pressure Locus and Gene Discovery Using Genome-Wide Association Study and Expression Data Sets From Blood and the Kidney. Hypertension, 2017, 70, . | 2.7 | 123 |
| 64 | A Large-Scale Multi-ancestry Genome-wide Study Accounting for Smoking Behavior Identifies Multiple Significant Loci for Blood Pressure. American Journal of Human Genetics, 2018, 102, 375-400. | 6.2 | 123 |
| 65 | Discovery of active enhancers through bidirectional expression of short transcripts. Genome Biology, 2011, 12, R113. | 9.6 | 120 |
| 66 | The genetic regulatory signature of type 2 diabetes in human skeletal muscle. Nature Communications, 2016, 7, 11764. | 12.8 | 114 |
| 67 | Integrative analysis of gene expression, DNA methylation, physiological traits, and genetic variation in human skeletal muscle. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 10883-10888. | 7.1 | 114 |
| 68 | Trans-Ethnic Fine-Mapping of Lipid Loci Identifies Population-Specific Signals and Allelic Heterogeneity That Increases the Trait Variance Explained. PLoS Genetics, 2013, 9, e1003379. | 3.5 | 112 |
| 69 | Biotinylation by antibody recognitionâ€”a method for proximity labeling. Nature Methods, 2018, 15, 127-133. | 19.0 | 107 |
| 70 | Single-cell ATAC-Seq in human pancreatic islets and deep learning upscaling of rare cells reveals cell-specific type 2 diabetes regulatory signatures. Molecular Metabolism, 2020, 32, 109-121. | 6.5 | 103 |
| 71 | A Genome-Wide Association Study of IVGTT-Based Measures of First-Phase Insulin Secretion Refines the Underlying Physiology of Type 2 Diabetes Variants. Diabetes, 2017, 66, 2296-2309. | 0.6 | 102 |
| 72 | Mining for therapeutic gold. Nature Reviews Drug Discovery, 2011, 10, 397-397. | 46.4 | 96 |

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|---|---|---|---|
| 73 | 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 |
| 74 | Using Science to Improve the Nation's Health System. JAMA - Journal of the American Medical Association, 2010, 303, 2182. | 7.4 | 94 |
| 75 | Opportunities for Research and NIH. Science, 2010, 327, 36-37. | 12.6 | 91 |
| 76 | Protein-coding variants implicate novel genes related to lipid homeostasis contributing to body-fat distribution. Nature Genetics, 2019, 51, 452-469. | 21.4 | 89 |
| 77 | Genetic variant effects on gene expression in human pancreatic islets and their implications for T2D. Nature Communications, 2020, 11, 4912. | 12.8 | 89 |
| 78 | Associations of autozygosity with a broad range of human phenotypes. Nature Communications, 2019, 10, 4957. | 12.8 | 84 |
| 79 | Affirming NIHâ€™s commitment to addressing structural racism in the biomedical research enterprise. Cell, 2021, 184, 3075-3079. | 28.9 | 81 |
| 80 | A principal component meta-analysis on multiple anthropometric traits identifies novel loci for body shape. Nature Communications, 2016, 7, 13357. | 12.8 | 74 |
| 81 | A defect in myoblast fusion underlies Carey-Fineman-Ziter syndrome. Nature Communications, 2017, 8, 16077. | 12.8 | 72 |
| 82 | Protein farnesylation inhibitors cause donut-shaped cell nuclei attributable to a centrosome separation defect. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 4997-5002. | 7.1 | 71 |
| 83 | Multiomic Profiling Identifies cis-Regulatory Networks Underlying Human Pancreatic Î² Cell Identity and Function. Cell Reports, 2019, 26, 788-801.e6. | 6.4 | 68 |
| 84 | Genome-Wide Association Study of the Modified Stumvoll Insulin Sensitivity Index Identifies BCL2 and FAM19A2 as Novel Insulin Sensitivity Loci. Diabetes, 2016, 65, 3200-3211. | 0.6 | 67 |
| 85 | Genome-wide association studies of metabolites in Finnish men identify disease-relevant loci. Nature Communications, 2022, 13, 1644. | 12.8 | 63 |
| 86 | Linkage Disequilibrium Between Microsatellite Markers Extends Beyond 1 cM on Chromosome 20 in Finns. Genome Research, 2001, 11, 1221-1226. | 5.5 | 60 |
| 87 | A targeted antisense therapeutic approach for Hutchinsonâ€“Gilford progeria syndrome. Nature Medicine, 2021, 27, 536-545. | 30.7 | 55 |
| 88 | A Type 2 Diabetesâ€“Associated Functional Regulatory Variant in a Pancreatic Islet Enhancer at the ADCY5 Locus. Diabetes, 2017, 66, 2521-2530. | 0.6 | 54 |
| 89 | Re-sequencing Expands Our Understanding of the Phenotypic Impact of Variants at GWAS Loci. PLoS Genetics, 2014, 10, e1004147. | 3.5 | 50 |
| 90 | Multiple Hepatic Regulatory Variants at the GALNT2 GWAS Locus Associated with High-Density Lipoprotein Cholesterol. American Journal of Human Genetics, 2015, 97, 801-815. | 6.2 | 49 |

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|---|---|---|---|
| 91 | Common, low-frequency, and rare genetic variants associated with lipoprotein subclasses and triglyceride measures in Finnish men from the METSIM study. PLoS Genetics, 2017, 13, e1007079. | 3.5 | 49 |
| 92 | 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 |
| 93 | Hydroa vacciniformeâ€“like lymphoproliferative disorder: an EBV disease with a low risk of systemic illness in whites. Blood, 2019, 133, 2753-2764. | 1.4 | 46 |
| 94 | Adipose Tissue Gene Expression Associations Reveal Hundreds of Candidate Genes for Cardiometabolic Traits. American Journal of Human Genetics, 2019, 105, 773-787. | 6.2 | 45 |
| 95 | Everolimus rescues multiple cellular defects in laminopathy-patient fibroblasts. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, 4206-4211. | 7.1 | 43 |
| 96 | Use of microarray hybrid capture and next-generation sequencing to identify the anatomy of a transgene. Nucleic Acids Research, 2013, 41, e70-e70. | 14.5 | 41 |
| 97 | Progeria: Translational insights from cell biology. Journal of Cell Biology, 2012, 199, 9-13. | 5.2 | 37 |
| 98 | Network News: Powering Clinical Research. Science Translational Medicine, 2013, 5, 182fs13. | 12.4 | 33 |
| 99 | Sequence data and association statistics from 12,940 type 2 diabetes cases and controls. Scientific Data, 2017, 4, 170179. | 5.3 | 31 |
| 100 | A multi-ancestry genome-wide study incorporating geneâ€“smoking interactions identifies multiple new loci for pulse pressure and mean arterial pressure. Human Molecular Genetics, 2019, 28, 2615-2633. | 2.9 | 31 |
| 101 | Identification of seven novel loci associated with amino acid levels using single-variant and gene-based tests in 8545 Finnish men from the METSIM study. Human Molecular Genetics, 2018, 27, 1664-1674. | 2.9 | 30 |
| 102 | Motif signatures in stretch enhancers are enriched for disease-associated genetic variants. Epigenetics and Chromatin, 2015, 8, 23. | 3.9 | 28 |
| 103 | A novel somatic mutation achieves partial rescue in a child with Hutchinson-Gilford progeria syndrome. Journal of Medical Genetics, 2017, 54, 212-216. | 3.2 | 28 |
| 104 | Genetic reduction of mTOR extends lifespan in a mouse model of Hutchinsonâ€Gilford Progeria syndrome. Aging Cell, 2021, 20, e13457. | 6.7 | 27 |
| 105 | NIH Roadmap/Common Fund at 10 years. Science, 2014, 345, 274-276. | 12.6 | 24 |
| 106 | Basic science: Bedrock of progress. Science, 2016, 351, 1405-1405. | 12.6 | 24 |
| 107 | NIH Basics. Science, 2012, 337, 503-503. | 12.6 | 20 |
| 108 | Evaluation of musculoskeletal phenotype of the G608G progeria mouse model with lonafarnib, pravastatin, and zoledronic acid as treatment groups. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 12029-12040. | 7.1 | 20 |

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| 109 | Interactions between genetic variation and cellular environment in skeletal muscle gene expression. PLoS ONE, 2018, 13, e0195788. | 2.5 | 18 |
| 110 | Transient induction of telomerase expression mediates senescence and reduces tumorigenesis in primary fibroblasts. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 18983-18993. | 7.1 | 18 |
| 111 | Scientists need a shorter path to research freedom. Nature, 2010, 467, 635-635. | 27.8 | 15 |
| 112 | Seeking a Cure for One of the Rarest Diseases: Progeria. Circulation, 2016, 134, 126-129. | 1.6 | 15 |
| 113 | Single-cell transcriptomics from human pancreatic islets: sample preparation matters. Biology Methods and Protocols, 2019, 4, bpz019. | 2.2 | 15 |
| 114 | COVID-19 lessons for research. Science, 2021, 371, 1081-1081. | 12.6 | 14 |
| 115 | Steering a New Course for Stem Cell Research: NIH's Intramural Center for Regenerative Medicine. Stem Cells Translational Medicine, 2012, 1, 15-17. | 3.3 | 13 |
| 116 | TUBB3 Arg262His causes a recognizable syndrome including CFEOM3, facial palsy, joint contractures, and early-onset peripheral neuropathy. Human Genetics, 2021, 140, 1709-1731. | 3.8 | 13 |
| 117 | Update on and Future Directions for Use of Antiâ€“SARS-CoV-2 Antibodies: National Institutes of Health Summit on Treatment and Prevention of COVID-19. Annals of Internal Medicine, 2022, 175, 119-126. | 3.9 | 13 |
| 118 | Biomedical Research: Strength from Diversity. Science, 2013, 342, 798-798. | 12.6 | 12 |
| 119 | Adiponectin GWAS loci harboring extensive allelic heterogeneity exhibit distinct molecular consequences. PLoS Genetics, 2020, 16, e1009019. | 3.5 | 11 |
| 120 | Biomedical Research: Strength from Diversity. Science, 2013, 342, 798-798. | 12.6 | 10 |
| 121 | Brain phenotyping in Moebius syndrome and other congenital facial weakness disorders by diffusion MRI morphometry. Brain Communications, 2020, 2, fcaa014. | 3.3 | 9 |
| 122 | A Transcription Start Site Map in Human Pancreatic Islets Reveals Functional Regulatory Signatures. Diabetes, 2021, 70, 1581-1591. | 0.6 | 7 |
| 123 | Addendum: Biotinylation by antibody recognitionâ€”a method for proximity labeling. Nature Methods, 2018, 15, 749-749. | 19.0 | 6 |
| 124 | Analysis of somatic mutations identifies signs of selection during in vitro aging of primary dermal fibroblasts. Aging Cell, 2019, 18, e13010. | 6.7 | 6 |
| 125 | Differentiating Moebius syndrome and other congenital facial weakness disorders with electrodiagnostic studies. Muscle and Nerve, 2021, 63, 516-524. | 2.2 | 6 |
| 126 | Base editor treats progeria in mice. Nature, 2021, , . | 27.8 | 4 |

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|---|---|---|---|
| 127 | NIH research: Think globally. Science, 2015, 348, 159-159. | 12.6 | 3 |
| 128 | Change, Change, Change: Heeding the Call. Molecular Biology of the Cell, 2010, 21, 3793-3794. | 2.1 | 1 |
| 129 | Biotinylation by antibody recognition. Protocol Exchange, 0, , . | 0.3 | 1 |