Francis S Collins

List of Publications by Citations

125 papers	59,979 citations	69 h-index	141 g-index
141 ext. papers	70,487 ext. citations	22.2 avg, IF	6.67 L-index

#	Paper	IF	Citations
125	Initial sequencing and analysis of the human genome. *Nature*, **2001**, 409, 860-921	50.4	17366
124	Identification and analysis of functional elements in 1% of the human genome by the ENCODE pilot project. *Nature*, **2007**, 447, 799-816	50.4	4121
123	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, 9362-7	11.5	3120
122	Biological, clinical and population relevance of 95 loci for blood lipids. *Nature*, **2010**, 466, 707-13	50.4	2742
121	Genetic studies of body mass index yield new insights for obesity biology. *Nature*, **2015**, 518, 197-206	50.4	2687
120	Discovery and refinement of loci associated with lipid levels. *Nature Genetics*, **2013**, 45, 1274-1283	36.3	1904
119	Recurrent de novo point mutations in lamin A cause Hutchinson-Gilford progeria syndrome. *Nature*, **2003**, 423, 293-8	50.4	1586
118	Genetic variants in novel pathways influence blood pressure and cardiovascular disease risk. *Nature* , **2011**, 478, 103-9	50.4	1564
117	Large-scale association analysis provides insights into the genetic architecture and pathophysiology of type 2 diabetes. *Nature Genetics*, **2012**, 44, 981-90	36.3	1482
116	Twelve type 2 diabetes susceptibility loci identified through large-scale association analysis. *Nature Genetics*, **2010**, 42, 579-89	36.3	1449
115	A reference panel of 64,976 haplotypes for genotype imputation. *Nature Genetics*, **2016**, 48, 1279-83	36.3	1447
114	Newly identified loci that influence lipid concentrations and risk of coronary artery disease. *Nature Genetics*, **2008**, 40, 161-9	36.3	1304
113	The path to personalized medicine. *New England Journal of Medicine*, **2010**, 363, 301-4	59.2	1294
112	Common variants at 30 loci contribute to polygenic dyslipidemia. *Nature Genetics*, **2009**, 41, 56-65	36.3	1095
111	New genetic loci link adipose and insulin biology to body fat distribution. *Nature*, **2015**, 518, 187-196	50.4	920
110	Accumulation of mutant lamin A causes progressive changes in nuclear architecture in Hutchinson-Gilford progeria syndrome. *Proceedings of the National Academy of Sciences of the United States of America*, **2004**, 101, 8963-8	11.5	812
109	Genome-wide trans-ancestry meta-analysis provides insight into the genetic architecture of type 2 diabetes susceptibility. *Nature Genetics*, **2014**, 46, 234-44	36.3	784

108	The Human Genome Project: lessons from large-scale biology. *Science*, **2003**, 300, 286-90	33.3	717
107	The genetic architecture of type 2 diabetes. *Nature*, **2016**, 536, 41-47	50.4	704
106	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	36.3	675
105	Large-scale association analyses identify new loci influencing glycemic traits and provide insight into the underlying biological pathways. *Nature Genetics*, **2012**, 44, 991-1005	36.3	621
104	A genome-wide approach accounting for body mass index identifies genetic variants influencing fasting glycemic traits and insulin resistance. *Nature Genetics*, **2012**, 44, 659-69	36.3	615
103	Phenotype and course of Hutchinson-Gilford progeria syndrome. *New England Journal of Medicine*, **2008**, 358, 592-604	59.2	484
102	Toxicology. Transforming environmental health protection. *Science*, **2008**, 319, 906-7	33.3	478
101	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, 17921-6	11.5	477
100	Human laminopathies: nuclei gone genetically awry. *Nature Reviews Genetics*, **2006**, 7, 940-52	30.1	414
99	Rare and low-frequency coding variants alter human adult height. *Nature*, **2017**, 542, 186-190	50.4	412
98	Genetic analysis of over 1 million people identifies 535 new loci associated with blood pressure traits. *Nature Genetics*, **2018**, 50, 1412-1425	36.3	386
97	Genome-wide association study identifies six new loci influencing pulse pressure and mean arterial pressure. *Nature Genetics*, **2011**, 43, 1005-11	36.3	338
96	Reengineering translational science: the time is right. *Science Translational Medicine*, **2011**, 3, 90cm17	17.5	337
95	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	6	326
94	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, 2624-34	0.9	285
93	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, 12879-84	11.5	285
92	Cardiovascular pathology in Hutchinson-Gilford progeria: correlation with the vascular pathology of aging. *Arteriosclerosis, Thrombosis, and Vascular Biology*, **2010**, 30, 2301-9	9.4	263
91	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	3.7	259

90	The genetics of blood pressure regulation and its target organs from association studies in 342,415 individuals. *Nature Genetics*, **2016**, 48, 1171-1184	36.3	251
89	Super-enhancers delineate disease-associated regulatory nodes in T cells. *Nature*, **2015**, 520, 558-62	50.4	247
88	Rapamycin reverses cellular phenotypes and enhances mutant protein clearance in Hutchinson-Gilford progeria syndrome cells. *Science Translational Medicine*, **2011**, 3, 89ra58	17.5	241
87	Impact of type 2 diabetes susceptibility variants on quantitative glycemic traits reveals mechanistic heterogeneity. *Diabetes*, **2014**, 63, 2158-71	0.9	235
86	Refining the accuracy of validated target identification through coding variant fine-mapping in type 2 diabetes. *Nature Genetics*, **2018**, 50, 559-571	36.3	221
85	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	6	220
84	The case for a US prospective cohort study of genes and environment. *Nature*, **2004**, 429, 475-7	50.4	217
83	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, 3250-5	11.5	211
82	A lamin A protein isoform overexpressed in Hutchinson-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, 4949-54	11.5	193
81	Has the revolution arrived?. *Nature*, **2010**, 464, 674-5	50.4	192
80	Protein-altering variants associated with body mass index implicate pathways that control energy intake and expenditure in obesity. *Nature Genetics*, **2018**, 50, 26-41	36.3	186
79	Trans-ancestry meta-analyses identify rare and common variants associated with blood pressure and hypertension. *Nature Genetics*, **2016**, 48, 1151-1161	36.3	181
78	Systematic comparison of three genomic enrichment methods for massively parallel DNA sequencing. *Genome Research*, **2010**, 20, 1420-31	9.7	175
77	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, 20788-93	11.5	157
76	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, 15902-7	11.5	155
75	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	11.5	132
74	Exome sequencing of 20,791 cases of type 2 diabetes and 24,440 controls. *Nature*, **2019**, 570, 71-76	50.4	129
73	Patient-Centered Outcomes Research Institute: the intersection of science and health care. *Science Translational Medicine*, **2010**, 2, 37cm18	17.5	120

72	Directional dominance on stature and cognition in diverse human populations. *Nature*, **2015**, 523, 459-462	50.4	119
71	Genome-wide meta-analysis of 241,258 adults accounting for smoking behaviour identifies novel loci for obesity traits. *Nature Communications*, **2017**, 8, 14977	17.4	105
70	Genome-wide physical activity interactions in adiposity - A meta-analysis of 200,452 adults. *PLoS Genetics*, **2017**, 13, e1006528	6	103
69	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	6	94
68	In vivo base editing rescues Hutchinson-Gilford progeria syndrome in mice. *Nature*, **2021**, 589, 608-614	50.4	92
67	Discovery of active enhancers through bidirectional expression of short transcripts. *Genome Biology* , **2011**, 12, R113	18.3	91
66	Large meta-analysis of genome-wide association studies identifies five loci for lean body mass. *Nature Communications*, **2017**, 8, 80	17.4	88
65	Genetic Regulation of Adipose Gene Expression and Cardio-Metabolic Traits. *American Journal of Human Genetics*, **2017**, 100, 428-443	11	87
64	Novel Blood Pressure Locus and Gene Discovery Using Genome-Wide Association Study and Expression Data Sets From Blood and the Kidney. *Hypertension*, **2017**,	8.5	85
63	The genetic regulatory signature of type 2 diabetes in human skeletal muscle. *Nature Communications*, **2016**, 7, 11764	17.4	82
62	Using science to improve the nation\x{2019}s health system: NIH\x{2019}s commitment to comparative effectiveness research. *JAMA - Journal of the American Medical Association*, **2010**, 303, 2182-3	27.4	80
61	Research agenda. Opportunities for research and NIH. *Science*, **2010**, 327, 36-7	33.3	79
60	The Metabolic Syndrome in Men study: a resource for studies of metabolic and cardiovascular diseases. *Journal of Lipid Research*, **2017**, 58, 481-493	6.3	77
59	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	6	76
58	Mining for therapeutic gold. *Nature Reviews Drug Discovery*, **2011**, 10, 397	64.1	72
57	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	0.9	69
56	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	11	59
55	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	11.5	58

54	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	11.5	54
53	Biotinylation by antibody recognition-a method for proximity labeling. *Nature Methods*, **2018**, 15, 127-133	21.6	54
52	A defect in myoblast fusion underlies Carey-Fineman-Ziter syndrome. *Nature Communications*, **2017**, 8, 16077	17.4	51
51	Linkage disequilibrium between microsatellite markers extends beyond 1 cM on chromosome 20 in Finns. *Genome Research*, **2001**, 11, 1221-6	9.7	51
50	Genome-Wide Association Study of the Modified Stumvoll Insulin Sensitivity Index Identifies BCL2 and FAM19A2 as Novel Insulin Sensitivity Loci. *Diabetes*, **2016**, 65, 3200-11	0.9	47
49	A principal component meta-analysis on multiple anthropometric traits identifies novel loci for body shape. *Nature Communications*, **2016**, 7, 13357	17.4	46
48	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	8.8	46
47	The trans-ancestral genomic architecture of glycemic traits. *Nature Genetics*, **2021**, 53, 840-860	36.3	44
46	Protein-coding variants implicate novel genes related to lipid homeostasis contributing to body-fat distribution. *Nature Genetics*, **2019**, 51, 452-469	36.3	44
45	Re-sequencing expands our understanding of the phenotypic impact of variants at GWAS loci. *PLoS Genetics*, **2014**, 10, e1004147	6	42
44	Associations of autozygosity with a broad range of human phenotypes. *Nature Communications*, **2019**, 10, 4957	17.4	40
43	Multiple Hepatic Regulatory Variants at the GALNT2 GWAS Locus Associated with High-Density Lipoprotein Cholesterol. *American Journal of Human Genetics*, **2015**, 97, 801-15	11	40
42	A Type 2 Diabetes-Associated Functional Regulatory Variant in a Pancreatic Islet Enhancer at the Locus. *Diabetes*, **2017**, 66, 2521-2530	0.9	37
41	Use of microarray hybrid capture and next-generation sequencing to identify the anatomy of a transgene. *Nucleic Acids Research*, **2013**, 41, e70	20.1	36
40	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	6	33
39	Progeria: translational insights from cell biology. *Journal of Cell Biology*, **2012**, 199, 9-13	7.3	30
38	Genetic variant effects on gene expression in human pancreatic islets and their implications for T2D. *Nature Communications*, **2020**, 11, 4912	17.4	30
37	A Low-Frequency Inactivating Variant Enriched in the Finnish Population Is Associated With Fasting Insulin Levels and Type 2 Diabetes Risk. *Diabetes*, **2017**, 66, 2019-2032	0.9	29

36	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	11.5	29
35	Hydroa vacciniforme-like lymphoproliferative disorder: an EBV disease with a low risk of systemic illness in whites. *Blood*, **2019**, 133, 2753-2764	2.2	28
34	Multiomic Profiling Identifies cis-Regulatory Networks Underlying Human Pancreatic Cell Identity and Function. *Cell Reports*, **2019**, 26, 788-801.e6	10.6	28
33	Affirming NIH's commitment to addressing structural racism in the biomedical research enterprise. *Cell*, **2021**, 184, 3075-3079	56.2	23
32	Sequence data and association statistics from 12,940 type 2 diabetes cases and controls. *Scientific Data*, **2017**, 4, 170179	8.2	22
31	Motif signatures in stretch enhancers are enriched for disease-associated genetic variants. *Epigenetics and Chromatin*, **2015**, 8, 23	5.8	22
30	Adipose Tissue Gene Expression Associations Reveal Hundreds of Candidate Genes for Cardiometabolic Traits. *American Journal of Human Genetics*, **2019**, 105, 773-787	11	20
29	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	5.6	20
28	Basic science: Bedrock of progress. *Science*, **2016**, 351, 1405	33.3	20
27	Funding transdisciplinary research. NIH Roadmap/Common Fund at 10 years. *Science*, **2014**, 345, 274-6	33.3	19
26	A targeted antisense therapeutic approach for Hutchinson-Gilford progeria syndrome. *Nature Medicine*, **2021**, 27, 536-545	50.5	17
25	A novel somatic mutation achieves partial rescue in a child with Hutchinson-Gilford progeria syndrome. *Journal of Medical Genetics*, **2017**, 54, 212-216	5.8	14
24	A multi-ancestry genome-wide study incorporating gene-smoking interactions identifies multiple new loci for pulse pressure and mean arterial pressure. *Human Molecular Genetics*, **2019**, 28, 2615-2633	5.6	14
23	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	11.5	13
22	Scientists need a shorter path to research freedom. *Nature*, **2010**, 467, 635	50.4	12
21	Steering a new course for stem cell research: NIH's intramural Center for Regenerative Medicine. *Stem Cells Translational Medicine*, **2012**, 1, 15-7	6.9	11
20	Influence of genetic variants on gene expression in human pancreatic islets Implications for type 2 diabetes		9
19	Interactions between genetic variation and cellular environment in skeletal muscle gene expression. *PLoS ONE*, **2018**, 13, e0195788	3.7	9

18	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	11.5	7
17	Biomedical research: strength from diversity. *Science*, **2013**, 342, 798	33.3	7
16	Biomedical Research: Strength from Diversity. *Science*, **2013**, 342, 798-798	33.3	7
15	Multi-ancestry genetic study of type 2 diabetes highlights the power of diverse populations for discovery and translation.. *Nature Genetics*, **2022**,	36.3	7
14	Single-cell transcriptomics from human pancreatic islets: sample preparation matters. *Biology Methods and Protocols*, **2020**, 5, bpz019	2.4	6
13	Genetic reduction of mTOR extends lifespan in a mouse model of Hutchinson-Gilford Progeria syndrome. *Aging Cell*, **2021**, 20, e13457	9.9	5
12	Genome-wide association studies of metabolites in Finnish men identify disease-relevant loci.. *Nature Communications*, **2022**, 13, 1644	17.4	5
11	ACE2 expression in adipose tissue is associated with COVID-19 cardio-metabolic risk factors and cell type composition **2020**,		4
10	Brain phenotyping in Moebius syndrome and other congenital facial weakness disorders by diffusion MRI morphometry. *Brain Communications*, **2020**, 2, fcaa014	4.5	3
9	Addendum: Biotinylation by antibody recognition-a method for proximity labeling. *Nature Methods*, **2018**, 15, 749	21.6	3
8	Analysis of somatic mutations identifies signs of selection during in vitro aging of primary dermal fibroblasts. *Aging Cell*, **2019**, 18, e13010	9.9	3
7	Update on and Future Directions for Use of Anti-SARS-CoV-2 Antibodies: National Institutes of Health Summit on Treatment and Prevention of COVID-19. *Annals of Internal Medicine*, **2021**,	8	3
6	Adiponectin GWAS loci harboring extensive allelic heterogeneity exhibit distinct molecular consequences. *PLoS Genetics*, **2020**, 16, e1009019	6	3
5	Differentiating Moebius syndrome and other congenital facial weakness disorders with electrodiagnostic studies. *Muscle and Nerve*, **2021**, 63, 516-524	3.4	2
4	Change, change, change: heeding the call. *Molecular Biology of the Cell*, **2010**, 21, 3793-4	3.5	1
3	TUBB3 Arg262His causes a recognizable syndrome including CFEOM3, facial palsy, joint contractures, and early-onset peripheral neuropathy. *Human Genetics*, **2021**, 140, 1709-1731	6.3	1
2	Single cell ATAC-seq in human pancreatic islets and deep learning upscaling of rare cells reveals cell-specific type 2 diabetes regulatory signatures		1
1	A Transcription Start Site Map in Human Pancreatic Islets Reveals Functional Regulatory Signatures. *Diabetes*, **2021**, 70, 1581-1591	0.9	1