Francis S Collins

List of Publications by Year
in descending order

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129 papers	77,235 citations	10650 74 h-index	13635 134 g-index
141 all docs	141 docs citations	141 times ranked	83662 citing authors

#	Article	IF	Citations
1	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.	2.0	13
2	Genome-wide association studies of metabolites in Finnish men identify disease-relevant loci. Nature Communications, 2022, 13, 1644.	5.8	63
3	Multi-ancestry genetic study of type 2 diabetes highlights the power of diverse populations for discovery and translation. Nature Genetics, 2022, 54, 560-572.	9.4	250
4	In vivo base editing rescues Hutchinsonâ€“Gilford progeria syndrome in mice. Nature, 2021, 589, 608-614.	13.7	275
5	COVID-19 lessons for research. Science, 2021, 371, 1081-1081.	6.0	14
6	A targeted antisense therapeutic approach for Hutchinsonâ€“Gilford progeria syndrome. Nature Medicine, 2021, 27, 536-545.	15.2	55
7	A Transcription Start Site Map in Human Pancreatic Islets Reveals Functional Regulatory Signatures. Diabetes, 2021, 70, 1581-1591.	0.3	7
8	The trans-ancestral genomic architecture of glycemic traits. Nature Genetics, 2021, 53, 840-860.	9.4	341
9	Base editor treats progeria in mice. Nature, 2021, , .	13.7	4
10	Affirming NIHâ€™s commitment to addressing structural racism in the biomedical research enterprise. Cell, 2021, 184, 3075-3079.	13.5	81
11	Genetic reduction of mTOR extends lifespan in a mouse model of Hutchinsonâ€Gilford Progeria syndrome. Aging Cell, 2021, 20, e13457.	3.0	27
12	Differentiating Moebius syndrome and other congenital facial weakness disorders with electrodiagnostic studies. Muscle and Nerve, 2021, 63, 516-524.	1.0	6
13	TUBB3 Arg262His causes a recognizable syndrome including CFEOM3, facial palsy, joint contractures, and early-onset peripheral neuropathy. Human Genetics, 2021, 140, 1709-1731.	1.8	13
14	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.	3.0	103
15	Adiponectin GWAS loci harboring extensive allelic heterogeneity exhibit distinct molecular consequences. PLoS Genetics, 2020, 16, e1009019.	1.5	11
16	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.	3.3	20
17	Brain phenotyping in Moebius syndrome and other congenital facial weakness disorders by diffusion MRI morphometry. Brain Communications, 2020, 2, fcaa014.	1.5	9
18	Genetic variant effects on gene expression in human pancreatic islets and their implications for T2D. Nature Communications, 2020, 11, 4912.	5.8	89

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19	Analysis of somatic mutations identifies signs of selection during in vitro aging of primary dermal fibroblasts. Aging Cell, 2019, 18, e13010.	3.0	6
20	Associations of autozygosity with a broad range of human phenotypes. Nature Communications, 2019, 10, 4957.	5.8	84
21	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.	3.3	18
22	Adipose Tissue Gene Expression Associations Reveal Hundreds of Candidate Genes for Cardiometabolic Traits. American Journal of Human Genetics, 2019, 105, 773-787.	2.6	45
23	Exome sequencing of 20,791Âcases of type 2 diabetes and 24,440Âcontrols. Nature, 2019, 570, 71-76.	13.7	248
24	Hydroa vacciniformeâ€“like lymphoproliferative disorder: an EBV disease with a low risk of systemic illness in whites. Blood, 2019, 133, 2753-2764.	0.6	46
25	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.	3.3	114
26	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.	1.4	31
27	Protein-coding variants implicate novel genes related to lipid homeostasis contributing to body-fat distribution. Nature Genetics, 2019, 51, 452-469.	9.4	89
28	Single-cell transcriptomics from human pancreatic islets: sample preparation matters. Biology Methods and Protocols, 2019, 4, bpz019.	1.0	15
29	Multiomic Profiling Identifies cis-Regulatory Networks Underlying Human Pancreatic Î² Cell Identity and Function. Cell Reports, 2019, 26, 788-801.e6.	2.9	68
30	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.	1.4	30
31	Refining the accuracy of validated target identification through coding variant fine-mapping in type 2 diabetes. Nature Genetics, 2018, 50, 559-571.	9.4	356
32	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.	2.6	123
33	Biotinylation by antibody recognitionâ€”a method for proximity labeling. Nature Methods, 2018, 15, 127-133.	9.0	107
34	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.	3.3	43
35	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.	9.4	1,331
36	Genetic analysis of over 1 million people identifies 535 new loci associated with blood pressure traits. Nature Genetics, 2018, 50, 1412-1425.	9.4	924

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37	Interactions between genetic variation and cellular environment in skeletal muscle gene expression. PLoS ONE, 2018, 13, e0195788.	1.1	18
38	Addendum: Biotinylation by antibody recognitionâ€"a method for proximity labeling. Nature Methods, 2018, 15, 749-749.	9.0	6
39	Protein-altering variants associated with body mass index implicate pathways that control energy intake and expenditure in obesity. Nature Genetics, 2018, 50, 26-41.	9.4	286
40	The Metabolic Syndrome in Men study: a resource for studies of metabolic and cardiovascular diseases. Journal of Lipid Research, 2017, 58, 481-493.	2.0	147
41	Rare and low-frequency coding variants alter human adult height. Nature, 2017, 542, 186-190.	13.7	544
42	Genetic Regulation of Adipose Gene Expression and Cardio-Metabolic Traits. American Journal of Human Genetics, 2017, 100, 428-443.	2.6	141
43	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.	3.3	189
44	A novel somatic mutation achieves partial rescue in a child with Hutchinson-Gilford progeria syndrome. Journal of Medical Genetics, 2017, 54, 212-216.	1.5	28
45	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.	0.3	102
46	Genome-wide meta-analysis of 241,258 adults accounting for smoking behaviour identifies novel loci for obesity traits. Nature Communications, 2017, 8, 14977.	5.8	169
47	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.	0.3	47
48	Novel Blood Pressure Locus and Gene Discovery Using Genome-Wide Association Study and Expression Data Sets From Blood and the Kidney. Hypertension, 2017, 70, .	1.3	123
49	Large meta-analysis of genome-wide association studies identifies five loci for lean body mass. Nature Communications, 2017, 8, 80.	5.8	147
50	A Type 2 Diabetesâ€"Associated Functional Regulatory Variant in a Pancreatic Islet Enhancer at the ADCY5 Locus. Diabetes, 2017, 66, 2521-2530.	0.3	54
51	A defect in myoblast fusion underlies Carey-Fineman-Ziter syndrome. Nature Communications, 2017, 8, 16077.	5.8	72
52	Sequence data and association statistics from 12,940 type 2 diabetes cases and controls. Scientific Data, 2017, 4, 170179.	2.4	31
53	Genome-wide physical activity interactions in adiposity â€• A meta-analysis of 200,452 adults. PLoS Genetics, 2017, 13, e1006528.	1.5	158
54	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.	1.5	49

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55	The genetic architecture of type 2 diabetes. Nature, 2016, 536, 41-47.	13.7	952
56	The genetic regulatory signature of type 2 diabetes in human skeletal muscle. Nature Communications, 2016, 7, 11764.	5.8	114
57	A reference panel of 64,976 haplotypes for genotype imputation. Nature Genetics, 2016, 48, 1279-1283.	9.4	2,421
58	A principal component meta-analysis on multiple anthropometric traits identifies novel loci for body shape. Nature Communications, 2016, 7, 13357.	5.8	74
59	Trans-ancestry meta-analyses identify rare and common variants associated with blood pressure and hypertension. Nature Genetics, 2016, 48, 1151-1161.	9.4	261
60	The genetics of blood pressure regulation and its target organs from association studies in 342,415 individuals. Nature Genetics, 2016, 48, 1171-1184.	9.4	362
61	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.	0.3	67
62	Seeking a Cure for One of the Rarest Diseases: Progeria. Circulation, 2016, 134, 126-129.	1.6	15
63	Basic science: Bedrock of progress. Science, 2016, 351, 1405-1405.	6.0	24
64	Motif signatures in stretch enhancers are enriched for disease-associated genetic variants. Epigenetics and Chromatin, 2015, 8, 23.	1.8	28
65	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.	1.5	331
66	Multiple Hepatic Regulatory Variants at the GALNT2 GWAS Locus Associated with High-Density Lipoprotein Cholesterol. American Journal of Human Genetics, 2015, 97, 801-815.	2.6	49
67	New genetic loci link adipose and insulin biology to body fat distribution. Nature, 2015, 518, 187-196.	13.7	1,328
68	Genetic studies of body mass index yield new insights for obesity biology. Nature, 2015, 518, 197-206.	13.7	3,823
69	Super-enhancers delineate disease-associated regulatory nodes in T cells. Nature, 2015, 520, 558-562.	13.7	323
70	Directional dominance on stature and cognition inÂdiverse human populations. Nature, 2015, 523, 459-462.	13.7	173
71	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.	1.5	95
72	NIH research: Think globally. Science, 2015, 348, 159-159.	6.0	3

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73	Impact of Type 2 Diabetes Susceptibility Variants on Quantitative Glycemic Traits Reveals Mechanistic Heterogeneity. Diabetes, 2014, 63, 2158-2171.	0.3	297
74	Re-sequencing Expands Our Understanding of the Phenotypic Impact of Variants at GWAS Loci. PLoS Genetics, 2014, 10, e1004147.	1.5	50
75	NIH Roadmap/Common Fund at 10 years. Science, 2014, 345, 274-276.	6.0	24
76	Genome-wide trans-ancestry meta-analysis provides insight into the genetic architecture of type 2 diabetes susceptibility. Nature Genetics, 2014, 46, 234-244.	9.4	959
77	Discovery and refinement of loci associated with lipid levels. Nature Genetics, 2013, 45, 1274-1283.	9.4	2,641
78	Biomedical Research: Strength from Diversity. Science, 2013, 342, 798-798.	6.0	12
79	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.	1.5	112
80	Use of microarray hybrid capture and next-generation sequencing to identify the anatomy of a transgene. Nucleic Acids Research, 2013, 41, e70-e70.	6.5	41
81	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.	3.3	606
82	Network News: Powering Clinical Research. Science Translational Medicine, 2013, 5, 182fs13.	5.8	33
83	Biomedical Research: Strength from Diversity. Science, 2013, 342, 798-798.	6.0	10
84	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.	1.5	419
85	NIH Basics. Science, 2012, 337, 503-503.	6.0	20
86	Progeria: Translational insights from cell biology. Journal of Cell Biology, 2012, 199, 9-13.	2.3	37
87	Steering a New Course for Stem Cell Research: NIH's Intramural Center for Regenerative Medicine. Stem Cells Translational Medicine, 2012, 1, 15-17.	1.6	13
88	Large-scale association analyses identify new loci influencing glycemic traits and provide insight into the underlying biological pathways. Nature Genetics, 2012, 44, 991-1005.	9.4	746
89	Large-scale association analysis provides insights into the genetic architecture and pathophysiology of type 2 diabetes. Nature Genetics, 2012, 44, 981-990.	9.4	1,748
90	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.	9.4	762

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91	Genetic variants in novel pathways influence blood pressure and cardiovascular disease risk. Nature, 2011, 478, 103-109.	13.7	1,855
92	Discovery of active enhancers through bidirectional expression of short transcripts. Genome Biology, 2011, 12, R113.	13.9	120
93	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.	0.3	335
94	Mining for therapeutic gold. Nature Reviews Drug Discovery, 2011, 10, 397-397.	21.5	96
95	Rapamycin Reverses Cellular Phenotypes and Enhances Mutant Protein Clearance in Hutchinson-Gilford Progeria Syndrome Cells. Science Translational Medicine, 2011, 3, 89ra58.	5.8	294
96	Reengineering Translational Science: The Time Is Right. Science Translational Medicine, 2011, 3, 90cm17.	5.8	409
97	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.	3.3	71
98	Genome-wide association study identifies six new loci influencing pulse pressure and mean arterial pressure. Nature Genetics, 2011, 43, 1005-1011.	9.4	403
99	Biological, clinical and population relevance of 95 loci for blood lipids. Nature, 2010, 466, 707-713.	13.7	3,249
100	Has the revolution arrived?. Nature, 2010, 464, 674-675.	13.7	243
101	Twelve type 2 diabetes susceptibility loci identified through large-scale association analysis. Nature Genetics, 2010, 42, 579-589.	9.4	1,631
102	Using Science to Improve the Nation's Health System. JAMA - Journal of the American Medical Association, 2010, 303, 2182.	3.8	94
103	Patient-Centered Outcomes Research Institute: The Intersection of Science and Health Care. Science Translational Medicine, 2010, 2, 37cm18.	5.8	168
104	Change, Change, Change: Heeding the Call. Molecular Biology of the Cell, 2010, 21, 3793-3794.	0.9	1
105	Opportunities for Research and NIH. Science, 2010, 327, 36-37.	6.0	91
106	Scientists need a shorter path to research freedom. Nature, 2010, 467, 635-635.	13.7	15
107	The Path to Personalized Medicine. New England Journal of Medicine, 2010, 363, 301-304.	13.9	1,595
108	Cardiovascular Pathology in Hutchinson-Gilford Progeria: Correlation With the Vascular Pathology of Aging. Arteriosclerosis, Thrombosis, and Vascular Biology, 2010, 30, 2301-2309.	1.1	332

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109	Systematic comparison of three genomic enrichment methods for massively parallel DNA sequencing. Genome Research, 2010, 20, 1420-1431.	2.4	194
110	Common variants at 30 loci contribute to polygenic dyslipidemia. Nature Genetics, 2009, 41, 56-65.	9.4	1,234
111	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.	3.3	185
112	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.	3.3	3,719
113	Phenotype and Course of Hutchinsonâ€“Gilford Progeria Syndrome. New England Journal of Medicine, 2008, 358, 592-604.	13.9	610
114	Newly identified loci that influence lipid concentrations and risk of coronary artery disease. Nature Genetics, 2008, 40, 161-169.	9.4	1,488
115	Transforming Environmental Health Protection. Science, 2008, 319, 906-907.	6.0	580
116	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.	3.3	181
117	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, 4949-4954.	3.3	235
118	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.	1.1	305
119	Identification and analysis of functional elements in 1% of the human genome by the ENCODE pilot project. Nature, 2007, 447, 799-816.	13.7	4,709
120	Human laminopathies: nuclei gone genetically awry. Nature Reviews Genetics, 2006, 7, 940-952.	7.7	478
121	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.	3.3	255
122	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.	3.3	334
123	The case for a US prospective cohort study of genes and environment. Nature, 2004, 429, 475-477.	13.7	290
124	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.	3.3	988
125	Recurrent de novo point mutations in lamin A cause Hutchinsonâ€“Gilford progeria syndrome. Nature, 2003, 423, 293-298.	13.7	1,925
126	The Human Genome Project: Lessons from Large-Scale Biology. Science, 2003, 300, 286-290.	6.0	959

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127	Initial sequencing and analysis of the human genome. Nature, 2001, 409, 860-921.	13.7	21,074
128	Linkage Disequilibrium Between Microsatellite Markers Extends Beyond 1 cM on Chromosome 20 in Finns. Genome Research, 2001, 11, 1221-1226.	2.4	60
129	Biotinylation by antibody recognition. Protocol Exchange, 0, , .	0.3	1