

# Francis S Collins

## List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

162 papers	88,881 citations	94 h-index	168 g-index
168 ext. papers	100,142 ext. citations	23.2 avg, IF	7.22 L-index

#	Paper	IF	Citations
162	Subcutaneous adipose tissue splice quantitative trait loci reveal differences in isoform usage associated with cardiometabolic traits.. <i>American Journal of Human Genetics</i> , <b>2022</b> , 109, 66-80	11	0
161	Genetic effects on liver chromatin accessibility identify disease regulatory variants. <i>American Journal of Human Genetics</i> , <b>2021</b> , 108, 1169-1189	11	3
160	Adipose Tissue Gene Expression Associations Reveal Hundreds of Candidate Genes for Cardiometabolic Traits. <i>American Journal of Human Genetics</i> , <b>2019</b> , 105, 773-787	11	20
159	Colocalization of GWAS and eQTL signals at loci with multiple signals identifies additional candidate genes for body fat distribution. <i>Human Molecular Genetics</i> , <b>2019</b> , 28, 4161-4172	5.6	12
158	Protein-coding variants implicate novel genes related to lipid homeostasis contributing to body-fat distribution. <i>Nature Genetics</i> , <b>2019</b> , 51, 452-469	36.3	44
157	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. <i>Human Molecular Genetics</i> , <b>2018</b> , 27, 1664-1674	5.6	20
156	Refining the accuracy of validated target identification through coding variant fine-mapping in type 2 diabetes. <i>Nature Genetics</i> , <b>2018</b> , 50, 559-571	36.3	221
155	A Large-Scale Multi-ancestry Genome-wide Study Accounting for Smoking Behavior Identifies Multiple Significant Loci for Blood Pressure. <i>American Journal of Human Genetics</i> , <b>2018</b> , 102, 375-400	11	59
154	Protein-altering variants associated with body mass index implicate pathways that control energy intake and expenditure in obesity. <i>Nature Genetics</i> , <b>2018</b> , 50, 26-41	36.3	186
153	Fine-mapping type 2 diabetes loci to single-variant resolution using high-density imputation and islet-specific epigenome maps. <i>Nature Genetics</i> , <b>2018</b> , 50, 1505-1513	36.3	675
152	The Metabolic Syndrome in Men study: a resource for studies of metabolic and cardiovascular diseases. <i>Journal of Lipid Research</i> , <b>2017</b> , 58, 481-493	6.3	77
151	Genetic Regulation of Adipose Gene Expression and Cardio-Metabolic Traits. <i>American Journal of Human Genetics</i> , <b>2017</b> , 100, 428-443	11	87
150	Genome-wide meta-analysis of 241,258 adults accounting for smoking behaviour identifies novel loci for obesity traits. <i>Nature Communications</i> , <b>2017</b> , 8, 14977	17.4	105
149	Causal Effect of Plasminogen Activator Inhibitor Type 1 on Coronary Heart Disease. <i>Journal of the American Heart Association</i> , <b>2017</b> , 6,	6	65
148	An Expanded Genome-Wide Association Study of Type 2 Diabetes in Europeans. <i>Diabetes</i> , <b>2017</b> , 66, 2888-2902	29.02	414
147	Genetic risk scores in the prediction of plasma glucose, impaired insulin secretion, insulin resistance and incident type 2 diabetes in the METSIM study. <i>Diabetologia</i> , <b>2017</b> , 60, 1722-1730	10.3	15
146	-ancestry Fine Mapping and Molecular Assays Identify Regulatory Variants at the HDL-C GWAS Locus. <i>G3: Genes, Genomes, Genetics</i> , <b>2017</b> , 7, 3217-3227	3.2	14

145	Sequence data and association statistics from 12,940 type 2 diabetes cases and controls. <i>Scientific Data</i> , <b>2017</b> , 4, 170179	8.2	22
144	Genome-wide physical activity interactions in adiposity - A meta-analysis of 200,452 adults. <i>PLoS Genetics</i> , <b>2017</b> , 13, e1006528	6	103
143	Common, low-frequency, and rare genetic variants associated with lipoprotein subclasses and triglyceride measures in Finnish men from the METSIM study. <i>PLoS Genetics</i> , <b>2017</b> , 13, e1007079	6	33
142	A principal component meta-analysis on multiple anthropometric traits identifies novel loci for body shape. <i>Nature Communications</i> , <b>2016</b> , 7, 13357	17.4	46
141	Trans-ancestry meta-analyses identify rare and common variants associated with blood pressure and hypertension. <i>Nature Genetics</i> , <b>2016</b> , 48, 1151-1161	36.3	181
140	The genetics of blood pressure regulation and its target organs from association studies in 342,415 individuals. <i>Nature Genetics</i> , <b>2016</b> , 48, 1171-1184	36.3	251
139	Rare variant in scavenger receptor BI raises HDL cholesterol and increases risk of coronary heart disease. <i>Science</i> , <b>2016</b> , 351, 1166-71	33.3	325
138	The genetic architecture of type 2 diabetes. <i>Nature</i> , <b>2016</b> , 536, 41-47	50.4	704
137	A new initiative on precision medicine. <i>New England Journal of Medicine</i> , <b>2015</b> , 372, 793-5	59.2	3119
136	Directional dominance on stature and cognition in diverse human populations. <i>Nature</i> , <b>2015</b> , 523, 459-462	50.4	119
135	Genetic fine mapping and genomic annotation defines causal mechanisms at type 2 diabetes susceptibility loci. <i>Nature Genetics</i> , <b>2015</b> , 47, 1415-25	36.3	292
134	Human Genome Project: Twenty-five years of big biology. <i>Nature</i> , <b>2015</b> , 526, 29-31	50.4	122
133	The Influence of Age and Sex on Genetic Associations with Adult Body Size and Shape: A Large-Scale Genome-Wide Interaction Study. <i>PLoS Genetics</i> , <b>2015</b> , 11, e1005378	6	220
132	Multiple Hepatic Regulatory Variants at the GALNT2 GWAS Locus Associated with High-Density Lipoprotein Cholesterol. <i>American Journal of Human Genetics</i> , <b>2015</b> , 97, 801-15	11	40
131	New genetic loci link adipose and insulin biology to body fat distribution. <i>Nature</i> , <b>2015</b> , 518, 187-196	50.4	920
130	Genetic studies of body mass index yield new insights for obesity biology. <i>Nature</i> , <b>2015</b> , 518, 197-206	50.4	2687
129	Defining the role of common variation in the genomic and biological architecture of adult human height. <i>Nature Genetics</i> , <b>2014</b> , 46, 1173-86	36.3	1339
128	Genome-wide trans-ancestry meta-analysis provides insight into the genetic architecture of type 2 diabetes susceptibility. <i>Nature Genetics</i> , <b>2014</b> , 46, 234-44	36.3	784

127	Inheritance of rare functional GCKR variants and their contribution to triglyceride levels in families. <i>Human Molecular Genetics</i> , <b>2014</b> , 23, 5570-8	5.6	18
126	Re-sequencing expands our understanding of the phenotypic impact of variants at GWAS loci. <i>PLoS Genetics</i> , <b>2014</b> , 10, e1004147	6	42
125	MicroRNA-27b is a regulatory hub in lipid metabolism and is altered in dyslipidemia. <i>Hepatology</i> , <b>2013</b> , 57, 533-42	11.2	164
124	A Drosophila functional evaluation of candidates from human genome-wide association studies of type 2 diabetes and related metabolic traits identifies tissue-specific roles for dHHEX. <i>BMC Genomics</i> , <b>2013</b> , 14, 136	4.5	27
123	Discovery and refinement of loci associated with lipid levels. <i>Nature Genetics</i> , <b>2013</b> , 45, 1274-1283	36.3	1904
122	Common variants associated with plasma triglycerides and risk for coronary artery disease. <i>Nature Genetics</i> , <b>2013</b> , 45, 1345-52	36.3	597
121	Genome-wide meta-analysis identifies 11 new loci for anthropometric traits and provides insights into genetic architecture. <i>Nature Genetics</i> , <b>2013</b> , 45, 501-12	36.3	437
120	Trans-ethnic fine-mapping of lipid loci identifies population-specific signals and allelic heterogeneity that increases the trait variance explained. <i>PLoS Genetics</i> , <b>2013</b> , 9, e1003379	6	94
119	Sex-stratified genome-wide association studies including 270,000 individuals show sexual dimorphism in genetic loci for anthropometric traits. <i>PLoS Genetics</i> , <b>2013</b> , 9, e1003500	6	277
118	Large-scale association analysis provides insights into the genetic architecture and pathophysiology of type 2 diabetes. <i>Nature Genetics</i> , <b>2012</b> , 44, 981-90	36.3	1482
117	FTO genotype is associated with phenotypic variability of body mass index. <i>Nature</i> , <b>2012</b> , 490, 267-72	50.4	304
116	A genome-wide association search for type 2 diabetes genes in African Americans. <i>PLoS ONE</i> , <b>2012</b> , 7, e29202	3.7	138
115	A genome-wide approach accounting for body mass index identifies genetic variants influencing fasting glycemic traits and insulin resistance. <i>Nature Genetics</i> , <b>2012</b> , 44, 659-69	36.3	615
114	Stratifying type 2 diabetes cases by BMI identifies genetic risk variants in LAMA1 and enrichment for risk variants in lean compared to obese cases. <i>PLoS Genetics</i> , <b>2012</b> , 8, e1002741	6	162
113	Novel loci for adiponectin levels and their influence on type 2 diabetes and metabolic traits: a multi-ethnic meta-analysis of 45,891 individuals. <i>PLoS Genetics</i> , <b>2012</b> , 8, e1002607	6	326
112	Large-scale association analyses identify new loci influencing glycemic traits and provide insight into the underlying biological pathways. <i>Nature Genetics</i> , <b>2012</b> , 44, 991-1005	36.3	621
111	No interactions between previously associated 2-hour glucose gene variants and physical activity or BMI on 2-hour glucose levels. <i>Diabetes</i> , <b>2012</b> , 61, 1291-6	0.9	21
110	Correlation of rare coding variants in the gene encoding human glucokinase regulatory protein with phenotypic, cellular, and kinetic outcomes. <i>Journal of Clinical Investigation</i> , <b>2012</b> , 122, 205-17	15.9	31

109	Genetic variants in novel pathways influence blood pressure and cardiovascular disease risk. <i>Nature</i> , <b>2011</b> , 478, 103-9	50.4	1564
108	Discovery of active enhancers through bidirectional expression of short transcripts. <i>Genome Biology</i> , <b>2011</b> , 12, R113	18.3	91
107	Genome-wide association identifies nine common variants associated with fasting proinsulin levels and provides new insights into the pathophysiology of type 2 diabetes. <i>Diabetes</i> , <b>2011</b> , 60, 2624-34	0.9	285
106	Effects of 34 risk loci for type 2 diabetes or hyperglycemia on lipoprotein subclasses and their composition in 6,580 nondiabetic Finnish men. <i>Diabetes</i> , <b>2011</b> , 60, 1608-16	0.9	65
105	Biological, clinical and population relevance of 95 loci for blood lipids. <i>Nature</i> , <b>2010</b> , 466, 707-13	50.4	2742
104	Hundreds of variants clustered in genomic loci and biological pathways affect human height. <i>Nature</i> , <b>2010</b> , 467, 832-8	50.4	1514
103	Genetic variation in GIPR influences the glucose and insulin responses to an oral glucose challenge. <i>Nature Genetics</i> , <b>2010</b> , 42, 142-8	36.3	527
102	Twelve type 2 diabetes susceptibility loci identified through large-scale association analysis. <i>Nature Genetics</i> , <b>2010</b> , 42, 579-89	36.3	1449
101	Meta-analysis identifies 13 new loci associated with waist-hip ratio and reveals sexual dimorphism in the genetic basis of fat distribution. <i>Nature Genetics</i> , <b>2010</b> , 42, 949-60	36.3	724
100	Association analyses of 249,796 individuals reveal 18 new loci associated with body mass index. <i>Nature Genetics</i> , <b>2010</b> , 42, 937-48	36.3	2267
99	Genomic medicine--an updated primer. <i>New England Journal of Medicine</i> , <b>2010</b> , 362, 2001-11	59.2	354
98	Genome-wide association scan meta-analysis identifies three Loci influencing adiposity and fat distribution. <i>PLoS Genetics</i> , <b>2009</b> , 5, e1000508	6	393
97	The completion of the Mammalian Gene Collection (MGC). <i>Genome Research</i> , <b>2009</b> , 19, 2324-33	9.7	98
96	Finding the missing heritability of complex diseases. <i>Nature</i> , <b>2009</b> , 461, 747-53	50.4	6084
95	Six new loci associated with body mass index highlight a neuronal influence on body weight regulation. <i>Nature Genetics</i> , <b>2009</b> , 41, 25-34	36.3	1368
94	Variants in MTNR1B influence fasting glucose levels. <i>Nature Genetics</i> , <b>2009</b> , 41, 77-81	36.3	584
93	Common variants at 30 loci contribute to polygenic dyslipidemia. <i>Nature Genetics</i> , <b>2009</b> , 41, 56-65	36.3	1095
92	Association of 18 confirmed susceptibility loci for type 2 diabetes with indices of insulin release, proinsulin conversion, and insulin sensitivity in 5,327 nondiabetic Finnish men. <i>Diabetes</i> , <b>2009</b> , 58, 2129-36	0.9	149

91	The HapMap and genome-wide association studies in diagnosis and therapy. <i>Annual Review of Medicine</i> , <b>2009</b> , 60, 443-56	17.4	155
90	The Scientific Foundation for personal genomics: recommendations from a National Institutes of Health-Centers for Disease Control and Prevention multidisciplinary workshop. <i>Genetics in Medicine</i> , <b>2009</b> , 11, 559-67	8.1	186
89	Tissue-specific alternative splicing of TCF7L2. <i>Human Molecular Genetics</i> , <b>2009</b> , 18, 3795-804	5.6	89
88	Potential etiologic and functional implications of genome-wide association loci for human diseases and traits. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2009</b> , 106, 9362-7	11.5	3120
87	Common variants near MC4R are associated with fat mass, weight and risk of obesity. <i>Nature Genetics</i> , <b>2008</b> , 40, 768-75	36.3	1048
86	Common variants in the GDF5-UQC region are associated with variation in human height. <i>Nature Genetics</i> , <b>2008</b> , 40, 198-203	36.3	315
85	Newly identified loci that influence lipid concentrations and risk of coronary artery disease. <i>Nature Genetics</i> , <b>2008</b> , 40, 161-9	36.3	1304
84	MicroRNA target site polymorphisms and human disease. <i>Trends in Genetics</i> , <b>2008</b> , 24, 489-97	8.5	285
83	Meta-analysis of genome-wide association data and large-scale replication identifies additional susceptibility loci for type 2 diabetes. <i>Nature Genetics</i> , <b>2008</b> , 40, 638-45	36.3	1496
82	Comprehensive association study of type 2 diabetes and related quantitative traits with 222 candidate genes. <i>Diabetes</i> , <b>2008</b> , 57, 3136-44	0.9	82
81	A HapMap harvest of insights into the genetics of common disease. <i>Journal of Clinical Investigation</i> , <b>2008</b> , 118, 1590-605	15.9	683
80	Variations in the G6PC2/ABCB11 genomic region are associated with fasting glucose levels. <i>Journal of Clinical Investigation</i> , <b>2008</b> , 118, 2620-8	15.9	127
79	A genome-wide association study of type 2 diabetes in Finns detects multiple susceptibility variants. <i>Science</i> , <b>2007</b> , 316, 1341-5	33.3	2269
78	Replicating genotype-phenotype associations. <i>Nature</i> , <b>2007</b> , 447, 655-60	50.4	1363
77	New models of collaboration in genome-wide association studies: the Genetic Association Information Network. <i>Nature Genetics</i> , <b>2007</b> , 39, 1045-51	36.3	258
76	Identification and analysis of functional elements in 1% of the human genome by the ENCODE pilot project. <i>Nature</i> , <b>2007</b> , 447, 799-816	50.4	4121
75	A second generation human haplotype map of over 3.1 million SNPs. <i>Nature</i> , <b>2007</b> , 449, 851-61	50.4	3647
74	Validity of reported genetic risk factors for acute coronary syndrome. <i>JAMA - Journal of the American Medical Association</i> , <b>2007</b> , 298, 1757; author reply 1759	27.4	

73	Distant conserved sequences flanking endothelial-specific promoters contain tissue-specific DNase-hypersensitive sites and over-represented motifs. <i>Human Molecular Genetics</i> , <b>2006</b> , 15, 2098-105	5.6	17
72	2005 William Allan Award address. No longer just looking under the lamppost. <i>American Journal of Human Genetics</i> , <b>2006</b> , 79, 421-6	11	10
71	Genome-wide mapping of DNase hypersensitive sites using massively parallel signature sequencing (MPSS). <i>Genome Research</i> , <b>2006</b> , 16, 123-31	9.7	363
70	Nurses and the genomic revolution. <i>Journal of Nursing Scholarship</i> , <b>2005</b> , 37, 98-101	3.6	32
69	Realizing the promise of genomics in biomedical research. <i>JAMA - Journal of the American Medical Association</i> , <b>2005</b> , 294, 1399-402	27.4	105
68	Race and ethnicity in the genome era: the complexity of the constructs. <i>American Psychologist</i> , <b>2005</b> , 60, 9-15	9.5	77
67	Identifying gene regulatory elements by genome-wide recovery of DNase hypersensitive sites. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2004</b> , 101, 992-7	11.5	150
66	The status, quality, and expansion of the NIH full-length cDNA project: the Mammalian Gene Collection (MGC). <i>Genome Research</i> , <b>2004</b> , 14, 2121-7	9.7	404
65	The knockout mouse project. <i>Nature Genetics</i> , <b>2004</b> , 36, 921-4	36.3	490
64	What we do and don't know about Race & ethnicity, genetics and health at the dawn of the genome era. <i>Nature Genetics</i> , <b>2004</b> , 36, S13-5	36.3	193
63	A large set of Finnish affected sibling pair families with type 2 diabetes suggests susceptibility loci on chromosomes 6, 11, and 14. <i>Diabetes</i> , <b>2004</b> , 53, 821-9	0.9	64
62	Welcome to the genomic era. <i>New England Journal of Medicine</i> , <b>2003</b> , 349, 996-8	59.2	221
61	Psychiatry in the genomics era. <i>American Journal of Psychiatry</i> , <b>2003</b> , 160, 616-20	11.9	57
60	A vision for the future of genomics research. <i>Nature</i> , <b>2003</b> , 422, 835-47	50.4	1390
59	Initial sequencing and comparative analysis of the mouse genome. <i>Nature</i> , <b>2002</b> , 420, 520-62	50.4	5376
58	Genomic medicine--a primer. <i>New England Journal of Medicine</i> , <b>2002</b> , 347, 1512-20	59.2	468
57	Generation and initial analysis of more than 15,000 full-length human and mouse cDNA sequences. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2002</b> , 99, 16899-903	11.5	1457
56	Linkage disequilibrium between microsatellite markers extends beyond 1 cM on chromosome 20 in Finns. <i>Genome Research</i> , <b>2001</b> , 11, 1221-6	9.7	51



55	The human genome project and the future of medicine. <i>Annals of the New York Academy of Sciences</i> , <b>1999</b> , 882, 42-55; discussion 56-65	6.5	39
54	The mammalian gene collection. <i>Science</i> , <b>1999</b> , 286, 455-7	33.3	237
53	A DNA polymorphism discovery resource for research on human genetic variation. <i>Genome Research</i> , <b>1998</b> , 8, 1229-31	9.7	627
52	Strategies for mutational analysis of the large multiexon ATM gene using high-density oligonucleotide arrays. <i>Genome Research</i> , <b>1998</b> , 8, 1245-58	9.7	99
51	Variations on a theme: cataloging human DNA sequence variation. <i>Science</i> , <b>1997</b> , 278, 1580-1	33.3	849
50	Gene therapy for cystic fibrosis. <i>Chest</i> , <b>1996</b> , 109, 241-52	5.3	39
49	Novel missense mutation (G314R) in a cystic fibrosis patient with hepatic failure. <i>Human Mutation</i> , <b>1996</b> , 7, 151-4	4.7	2
48	The geneticist's approach to complex disease. <i>Annual Review of Medicine</i> , <b>1996</b> , 47, 333-53	17.4	55
47	Transcript identification in the BRCA1 candidate region. <i>Breast Cancer Research and Treatment</i> , <b>1995</b> , 33, 115-24	4.4	2
46	A high-density microsatellite map of the ataxia-telangiectasia locus. <i>Human Genetics</i> , <b>1995</b> , 95, 451-4	6.3	54
45	TEL1, an <i>S. cerevisiae</i> homolog of the human gene mutated in ataxia telangiectasia, is functionally related to the yeast checkpoint gene MEC1. <i>Cell</i> , <b>1995</b> , 82, 831-40	56.2	331
44	A single ataxia telangiectasia gene with a product similar to PI-3 kinase. <i>Science</i> , <b>1995</b> , 268, 1749-53	33.3	2334
43	Nonsense mutations at Arg-1947 in two cases of familial neurofibromatosis type 1 in Japanese. <i>Human Genetics</i> , <b>1994</b> , 93, 81-3	6.3	20
42	Familial breast cancer. Approaching the isolation of a susceptibility gene. <i>Cancer</i> , <b>1994</b> , 74, 1013-20	6.4	10
41	Loss of neurofibromin in adrenal gland tumors from patients with neurofibromatosis type I. <i>Genes Chromosomes and Cancer</i> , <b>1994</b> , 10, 55-8	5	57
40	A YAC contig spanning the ataxia-telangiectasia locus (groups A and C) at 11q22-q23. <i>Genomics</i> , <b>1994</b> , 24, 234-42	4.3	7
39	Molecular basis of defective anion transport in L cells expressing recombinant forms of CFTR. <i>Human Molecular Genetics</i> , <b>1993</b> , 2, 1253-61	5.6	76
38	Expression of an abundant alternatively spliced form of the cystic fibrosis transmembrane conductance regulator (CFTR) gene is not associated with a cAMP-activated chloride conductance. <i>Human Molecular Genetics</i> , <b>1993</b> , 2, 225-30	5.6	88



37	Localization of the cystic fibrosis transmembrane conductance regulator in human bile duct epithelial cells. <i>Gastroenterology</i> , <b>1993</b> , 105, 1857-64	13.3	289
36	Molecular biology of cystic fibrosis. <i>Molecular Genetic Medicine</i> , <b>1993</b> , 3, 33-68		26
35	An African-American cystic fibrosis patient homozygous for a novel frameshift mutation associated with reduced CFTR mRNA levels. <i>Human Mutation</i> , <b>1993</b> , 2, 148-51	4.7	8
34	The human NME2 gene lies within 18kb of NME1 in chromosome 17. <i>Genes Chromosomes and Cancer</i> , <b>1993</b> , 6, 245-8	5	21
33	Mutations in the neurofibromatosis 1 gene in sporadic malignant melanoma cell lines. <i>Nature Genetics</i> , <b>1993</b> , 3, 118-21	36.3	133
32	Somatic deletion of the neurofibromatosis type 1 gene in a neurofibrosarcoma supports a tumour suppressor gene hypothesis. <i>Nature Genetics</i> , <b>1993</b> , 3, 122-6	36.3	347
31	A cosmid contig and high resolution restriction map of the 2 megabase region containing the Huntington <sup>®</sup> disease gene. <i>Nature Genetics</i> , <b>1993</b> , 4, 181-6	36.3	93
30	Magnetic bead capture of expressed sequences encoded within large genomic segments. <i>Nature</i> , <b>1993</b> , 361, 751-3	50.4	92
29	Sequencing and analysis of genomic fragments from the NF1 locus. <i>DNA Sequence</i> , <b>1992</b> , 3, 237-43		11
28	Aberrant regulation of ras proteins in malignant tumour cells from type 1 neurofibromatosis patients. <i>Nature</i> , <b>1992</b> , 356, 713-5	50.4	589
27	The Huntington <sup>®</sup> disease candidate region exhibits many different haplotypes. <i>Nature Genetics</i> , <b>1992</b> , 1, 99-103	36.3	142
26	Characterization of an intron 12 splice donor mutation in the cystic fibrosis transmembrane conductance regulator (CFTR) gene. <i>Human Mutation</i> , <b>1992</b> , 1, 380-7	4.7	25
25	Recent progress toward understanding the molecular biology of von Recklinghausen neurofibromatosis. <i>Annals of Neurology</i> , <b>1992</b> , 31, 555-61	9.4	44
24	A de novo Alu insertion results in neurofibromatosis type 1. <i>Nature</i> , <b>1991</b> , 353, 864-6	50.4	390
23	Cystic fibrosis gene mutation in two sisters with mild disease and normal sweat electrolyte levels. <i>New England Journal of Medicine</i> , <b>1991</b> , 325, 1630-4	59.2	97
22	cDNA cloning of the type 1 neurofibromatosis gene: complete sequence of the NF1 gene product. <i>Genomics</i> , <b>1991</b> , 11, 931-40	4.3	350
21	The Genome Project and human health. <i>FASEB Journal</i> , <b>1991</b> , 5, 77	0.9	10
20	Molecular genetics of von Recklinghausen neurofibromatosis. <i>Advances in Human Genetics</i> , <b>1991</b> , 20, 267-307		9

19	The cystic fibrosis gene: isolation and significance. <i>Hospital Practice (1995)</i> , <b>1990</b> , 25, 47-57	2.2	24
18	A chromosome jump crosses a translocation breakpoint in the von Recklinghausen neurofibromatosis region. <i>Genes Chromosomes and Cancer</i> , <b>1990</b> , 2, 271-7	5	20
17	Mutation analysis for heterozygote detection and the prenatal diagnosis of cystic fibrosis. <i>New England Journal of Medicine</i> , <b>1990</b> , 322, 291-6	59.2	197
16	Type 1 neurofibromatosis gene: identification of a large transcript disrupted in three NF1 patients. <i>Science</i> , <b>1990</b> , 249, 181-6	33.3	1279
15	Type 1 neurofibromatosis gene: correction. <i>Science</i> , <b>1990</b> , 250, 1749-1749	33.3	
14	Reverse genetics and cystic fibrosis. <i>American Journal of Respiratory Cell and Molecular Biology</i> , <b>1990</b> , 2, 309-16	5.7	9
13	Approaches to localizing disease genes as applied to cystic fibrosis. <i>Nucleic Acids Research</i> , <b>1990</b> , 18, 345-50	20.1	21
12	Correction of the cystic fibrosis defect in vitro by retrovirus-mediated gene transfer. <i>Cell</i> , <b>1990</b> , 62, 1227-33	5.3	528
11	Response : Type 1 Neurofibromatosis Gene: Correction. <i>Science</i> , <b>1990</b> , 250, 1749-1749	33.3	
10	Response : Type 1 Neurofibromatosis Gene: Correction. <i>Science</i> , <b>1990</b> , 250, 1749-1749	33.3	
9	Mutations in the p53 gene occur in diverse human tumour types. <i>Nature</i> , <b>1989</b> , 342, 705-8	50.4	2418
8	Identification of the cystic fibrosis gene: chromosome walking and jumping. <i>Science</i> , <b>1989</b> , 245, 1059-65	33.3	2745
7	Two patients with ring chromosome 15 syndrome. <i>American Journal of Medical Genetics Part A</i> , <b>1988</b> , 29, 149-54		53
6	Physical mapping of the cystic fibrosis region by pulsed-field gel electrophoresis. <i>Genomics</i> , <b>1988</b> , 2, 346-54	4.4	43
5	Adult Turner syndrome associated with chylous ascites and vascular anomalies. <i>Clinical Genetics</i> , <b>1987</b> , 31, 218-23	4	1
4	Molecular analysis of deletion and nondeletion hereditary persistence of fetal hemoglobin and identification of a new mutation causing beta-thalassemia. <i>Annals of the New York Academy of Sciences</i> , <b>1985</b> , 445, 159-69	6.5	8
3	Refining the accuracy of validated target identification through coding variant fine-mapping in type 2 diabetes		4
2	Fine-mapping of an expanded set of type 2 diabetes loci to single-variant resolution using high-density imputation and islet-specific epigenome maps		18

1	Protein-Coding Variants Implicate Novel Genes Related to Lipid Homeostasis Contributing to Body Fat Distribution	1
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