

Francis S Collins

List of Publications by Year in descending order

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Version: 2024-02-01

163
papers

107,283
citations

2565

99
h-index

7234

158
g-index

168
all docs

168
docs citations

168
times ranked

96405
citing authors

#	ARTICLE	IF	CITATIONS
1	Subcutaneous adipose tissue splice quantitative trait loci reveal differences in isoform usage associated with cardiometabolic traits. American Journal of Human Genetics, 2022, 109, 66-80.	2.6	13
2	Genetic effects on liver chromatin accessibility identify disease regulatory variants. American Journal of Human Genetics, 2021, 108, 1169-1189.	2.6	22
3	Realizing the Dream of Molecularly Targeted Therapies for Cystic Fibrosis. New England Journal of Medicine, 2019, 381, 1863-1865.	13.9	34
4	Colocalization of GWAS and eQTL signals at loci with multiple signals identifies additional candidate genes for body fat distribution. Human Molecular Genetics, 2019, 28, 4161-4172.	1.4	41
5	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
6	Protein-coding variants implicate novel genes related to lipid homeostasis contributing to body-fat distribution. Nature Genetics, 2019, 51, 452-469.	9.4	89
7	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
8	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
9	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
10	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
11	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
12	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
13	Genetic Regulation of Adipose Gene Expression and Cardio-Metabolic Traits. American Journal of Human Genetics, 2017, 100, 428-443.	2.6	141
14	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
15	Causal Effect of Plasminogen Activator Inhibitor Type 1 on Coronary Heart Disease. Journal of the American Heart Association, 2017, 6, .	1.6	89
16	An Expanded Genome-Wide Association Study of Type 2 Diabetes in Europeans. Diabetes, 2017, 66, 2888-2902.	0.3	615
17	Genetic risk scores in the prediction of plasma glucose, impaired insulin secretion, insulin resistance and incident type 2 diabetes in the METSIM study. Diabetologia, 2017, 60, 1722-1730.	2.9	26
18	<i>Trans</i>-ancestry Fine Mapping and Molecular Assays Identify Regulatory Variants at the <i>ANGPTL8</i> HDL-C GWAS Locus. G3: Genes, Genomes, Genetics, 2017, 7, 3217-3227.	0.8	19

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19	Sequence data and association statistics from 12,940 type 2 diabetes cases and controls. <i>Scientific Data</i> , 2017, 4, 170179.	2.4	31
20	Genome-wide physical activity interactions in adiposity • A meta-analysis of 200,452 adults. <i>PLoS Genetics</i> , 2017, 13, e1006528.	1.5	158
21	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> , 2017, 13, e1007079.	1.5	49
22	The genetic architecture of type 2 diabetes. <i>Nature</i> , 2016, 536, 41-47.	13.7	952
23	A principal component meta-analysis on multiple anthropometric traits identifies novel loci for body shape. <i>Nature Communications</i> , 2016, 7, 13357.	5.8	74
24	Trans-ancestry meta-analyses identify rare and common variants associated with blood pressure and hypertension. <i>Nature Genetics</i> , 2016, 48, 1151-1161.	9.4	261
25	The genetics of blood pressure regulation and its target organs from association studies in 342,415 individuals. <i>Nature Genetics</i> , 2016, 48, 1171-1184.	9.4	362
26	Rare variant in scavenger receptor BI raises HDL cholesterol and increases risk of coronary heart disease. <i>Science</i> , 2016, 351, 1166-1171.	6.0	438
27	Human Genome Project: Twenty-five years of big biology. <i>Nature</i> , 2015, 526, 29-31.	13.7	184
28	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> , 2015, 11, e1005378.	1.5	331
29	Multiple Hepatic Regulatory Variants at the GALNT2 GWAS Locus Associated with High-Density Lipoprotein Cholesterol. <i>American Journal of Human Genetics</i> , 2015, 97, 801-815.	2.6	49
30	New genetic loci link adipose and insulin biology to body fat distribution. <i>Nature</i> , 2015, 518, 187-196.	13.7	1,328
31	Genetic studies of body mass index yield new insights for obesity biology. <i>Nature</i> , 2015, 518, 197-206.	13.7	3,823
32	A New Initiative on Precision Medicine. <i>New England Journal of Medicine</i> , 2015, 372, 793-795.	13.9	4,081
33	Directional dominance on stature and cognition in diverse human populations. <i>Nature</i> , 2015, 523, 459-462.	13.7	173
34	Genetic fine mapping and genomic annotation defines causal mechanisms at type 2 diabetes susceptibility loci. <i>Nature Genetics</i> , 2015, 47, 1415-1425.	9.4	365
35	Inheritance of rare functional GCKR variants and their contribution to triglyceride levels in families. <i>Human Molecular Genetics</i> , 2014, 23, 5570-5578.	1.4	21
36	Re-sequencing Expands Our Understanding of the Phenotypic Impact of Variants at GWAS Loci. <i>PLoS Genetics</i> , 2014, 10, e1004147.	1.5	50

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37	Defining the role of common variation in the genomic and biological architecture of adult human height. <i>Nature Genetics</i> , 2014, 46, 1173-1186.	9.4	1,818
38	Genome-wide trans-ancestry meta-analysis provides insight into the genetic architecture of type 2 diabetes susceptibility. <i>Nature Genetics</i> , 2014, 46, 234-244.	9.4	959
39	MicroRNA-27b is a regulatory hub in lipid metabolism and is altered in dyslipidemia. <i>Hepatology</i> , 2013, 57, 533-542.	3.6	196
40	A <i>Drosophila</i> 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> , 2013, 14, 136.	1.2	37
41	Discovery and refinement of loci associated with lipid levels. <i>Nature Genetics</i> , 2013, 45, 1274-1283.	9.4	2,641
42	Common variants associated with plasma triglycerides and risk for coronary artery disease. <i>Nature Genetics</i> , 2013, 45, 1345-1352.	9.4	754
43	Genome-wide meta-analysis identifies 11 new loci for anthropometric traits and provides insights into genetic architecture. <i>Nature Genetics</i> , 2013, 45, 501-512.	9.4	578
44	Trans-Ethnic Fine-Mapping of Lipid Loci Identifies Population-Specific Signals and Allelic Heterogeneity That Increases the Trait Variance Explained. <i>PLoS Genetics</i> , 2013, 9, e1003379.	1.5	112
45	Sex-stratified Genome-wide Association Studies Including 270,000 Individuals Show Sexual Dimorphism in Genetic Loci for Anthropometric Traits. <i>PLoS Genetics</i> , 2013, 9, e1003500.	1.5	371
46	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> , 2012, 8, e1002741.	1.5	190
47	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> , 2012, 8, e1002607.	1.5	419
48	Large-scale association analyses identify new loci influencing glycaemic traits and provide insight into the underlying biological pathways. <i>Nature Genetics</i> , 2012, 44, 991-1005.	9.4	746
49	No Interactions Between Previously Associated 2-Hour Glucose Gene Variants and Physical Activity or BMI on 2-Hour Glucose Levels. <i>Diabetes</i> , 2012, 61, 1291-1296.	0.3	23
50	Large-scale association analysis provides insights into the genetic architecture and pathophysiology of type 2 diabetes. <i>Nature Genetics</i> , 2012, 44, 981-990.	9.4	1,748
51	FTO genotype is associated with phenotypic variability of body mass index. <i>Nature</i> , 2012, 490, 267-272.	13.7	383
52	A Genome-Wide Association Search for Type 2 Diabetes Genes in African Americans. <i>PLoS ONE</i> , 2012, 7, e29202.	1.1	197
53	A genome-wide approach accounting for body mass index identifies genetic variants influencing fasting glycaemic traits and insulin resistance. <i>Nature Genetics</i> , 2012, 44, 659-669.	9.4	762
54	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> , 2012, 122, 205-217.	3.9	41

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55	Genetic variants in novel pathways influence blood pressure and cardiovascular disease risk. <i>Nature</i> , 2011, 478, 103-109.	13.7	1,855
56	Discovery of active enhancers through bidirectional expression of short transcripts. <i>Genome Biology</i> , 2011, 12, R113.	13.9	120
57	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> , 2011, 60, 2624-2634.	0.3	335
58	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> , 2011, 60, 1608-1616.	0.3	77
59	Biological, clinical and population relevance of 95 loci for blood lipids. <i>Nature</i> , 2010, 466, 707-713.	13.7	3,249
60	Hundreds of variants clustered in genomic loci and biological pathways affect human height. <i>Nature</i> , 2010, 467, 832-838.	13.7	1,789
61	Genetic variation in GIPR influences the glucose and insulin responses to an oral glucose challenge. <i>Nature Genetics</i> , 2010, 42, 142-148.	9.4	591
62	Twelve type 2 diabetes susceptibility loci identified through large-scale association analysis. <i>Nature Genetics</i> , 2010, 42, 579-589.	9.4	1,631
63	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> , 2010, 42, 949-960.	9.4	836
64	Association analyses of 249,796 individuals reveal 18 new loci associated with body mass index. <i>Nature Genetics</i> , 2010, 42, 937-948.	9.4	2,634
65	Genomic Medicine – An Updated Primer. <i>New England Journal of Medicine</i> , 2010, 362, 2001-2011.	13.9	411
66	Genome-Wide Association Scan Meta-Analysis Identifies Three Loci Influencing Adiposity and Fat Distribution. <i>PLoS Genetics</i> , 2009, 5, e1000508.	1.5	453
67	The completion of the Mammalian Gene Collection (MGC). <i>Genome Research</i> , 2009, 19, 2324-2333.	2.4	125
68	Finding the missing heritability of complex diseases. <i>Nature</i> , 2009, 461, 747-753.	13.7	7,490
69	Six new loci associated with body mass index highlight a neuronal influence on body weight regulation. <i>Nature Genetics</i> , 2009, 41, 25-34.	9.4	1,572
70	Variants in MTNR1B influence fasting glucose levels. <i>Nature Genetics</i> , 2009, 41, 77-81.	9.4	662
71	Common variants at 30 loci contribute to polygenic dyslipidemia. <i>Nature Genetics</i> , 2009, 41, 56-65.	9.4	1,234
72	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> , 2009, 58, 2129-2136.	0.3	161

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73	The HapMap and Genome-Wide Association Studies in Diagnosis and Therapy. <i>Annual Review of Medicine</i> , 2009, 60, 443-456.	5.0	191
74	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> , 2009, 11, 559-567.	1.1	207
75	Tissue-specific alternative splicing of TCF7L2. <i>Human Molecular Genetics</i> , 2009, 18, 3795-3804.	1.4	100
76	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> , 2009, 106, 9362-9367.	3.3	3,719
77	Common variants near MC4R are associated with fat mass, weight and risk of obesity. <i>Nature Genetics</i> , 2008, 40, 768-775.	9.4	1,179
78	Common variants in the GDF5-UQCC region are associated with variation in human height. <i>Nature Genetics</i> , 2008, 40, 198-203.	9.4	369
79	Newly identified loci that influence lipid concentrations and risk of coronary artery disease. <i>Nature Genetics</i> , 2008, 40, 161-169.	9.4	1,488
80	MicroRNA target site polymorphisms and human disease. <i>Trends in Genetics</i> , 2008, 24, 489-497.	2.9	318
81	Meta-analysis of genome-wide association data and large-scale replication identifies additional susceptibility loci for type 2 diabetes. <i>Nature Genetics</i> , 2008, 40, 638-645.	9.4	1,683
82	Comprehensive Association Study of Type 2 Diabetes and Related Quantitative Traits With 222 Candidate Genes. <i>Diabetes</i> , 2008, 57, 3136-3144.	0.3	104
83	A HapMap harvest of insights into the genetics of common disease. <i>Journal of Clinical Investigation</i> , 2008, 118, 1590-1605.	3.9	788
84	Variations in the G6PC2/ABCB11 genomic region are associated with fasting glucose levels. <i>Journal of Clinical Investigation</i> , 2008, 118, 2620-8.	3.9	146
85	Validity of Reported Genetic Risk Factors for Acute Coronary Syndrome. <i>JAMA - Journal of the American Medical Association</i> , 2007, 298, 1757.	3.8	0
86	A Genome-Wide Association Study of Type 2 Diabetes in Finns Detects Multiple Susceptibility Variants. <i>Science</i> , 2007, 316, 1341-1345.	6.0	2,534
87	Replicating genotype-phenotype associations. <i>Nature</i> , 2007, 447, 655-660.	13.7	1,509
88	New models of collaboration in genome-wide association studies: the Genetic Association Information Network. <i>Nature Genetics</i> , 2007, 39, 1045-1051.	9.4	288
89	Identification and analysis of functional elements in 1% of the human genome by the ENCODE pilot project. <i>Nature</i> , 2007, 447, 799-816.	13.7	4,709
90	A second generation human haplotype map of over 3.1 million SNPs. <i>Nature</i> , 2007, 449, 851-861.	13.7	4,137

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91	No Longer Just Looking under the Lamppost**Previously presented at the annual meeting of The American Society of Human Genetics, in Salt Lake City, on October 28, 2005.. American Journal of Human Genetics, 2006, 79, 421-426.	2.6	16
92	Distant conserved sequences flanking endothelial-specific promoters contain tissue-specific DNase-hypersensitive sites and over-represented motifs. Human Molecular Genetics, 2006, 15, 2098-2105.	1.4	20
93	Nurses and the Genomic Revolution. Journal of Nursing Scholarship, 2005, 37, 98-101.	1.1	36
94	Genome-wide mapping of DNase hypersensitive sites using massively parallel signature sequencing (MPSS). Genome Research, 2005, 16, 123-131.	2.4	431
95	Realizing the Promise of Genomics in Biomedical Research. JAMA - Journal of the American Medical Association, 2005, 294, 1399.	3.8	127
96	Race and ethnicity in the genome era: The complexity of the constructs.. American Psychologist, 2005, 60, 9-15.	3.8	106
97	Identifying gene regulatory elements by genome-wide recovery of DNase hypersensitive sites. Proceedings of the National Academy of Sciences of the United States of America, 2004, 101, 992-997.	3.3	166
98	The Status, Quality, and Expansion of the NIH Full-Length cDNA Project: The Mammalian Gene Collection (MGC). Genome Research, 2004, 14, 2121-2127.	2.4	486
99	The Knockout Mouse Project. Nature Genetics, 2004, 36, 921-924.	9.4	556
100	What we do and don't know about 'race', 'ethnicity', genetics and health at the dawn of the genome era. Nature Genetics, 2004, 36, S13-S15.	9.4	249
101	A Large Set of Finnish Affected Sibling Pair Families With Type 2 Diabetes Suggests Susceptibility Loci on Chromosomes 6, 11, and 14. Diabetes, 2004, 53, 821-829.	0.3	73
102	A vision for the future of genomics research. Nature, 2003, 422, 835-847.	13.7	1,650
103	Welcome to the Genomic Era. New England Journal of Medicine, 2003, 349, 996-998.	13.9	251
104	Psychiatry in the Genomics Era. American Journal of Psychiatry, 2003, 160, 616-620.	4.0	71
105	Genomic Medicine â€” A Primer. New England Journal of Medicine, 2002, 347, 1512-1520.	13.9	538
106	Generation and initial analysis of more than 15,000 full-length human and mouse cDNA sequences. Proceedings of the National Academy of Sciences of the United States of America, 2002, 99, 16899-16903.	3.3	1,610
107	Initial sequencing and comparative analysis of the mouse genome. Nature, 2002, 420, 520-562.	13.7	6,319
108	Linkage Disequilibrium Between Microsatellite Markers Extends Beyond 1 cM on Chromosome 20 in Finns. Genome Research, 2001, 11, 1221-1226.	2.4	60

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109	The Human Genome Project and the Future of Medicine. <i>Annals of the New York Academy of Sciences</i> , 1999, 882, 42-55.	1.8	51
110	The Mammalian Gene Collection. <i>Science</i> , 1999, 286, 455-457.	6.0	266
111	A DNA Polymorphism Discovery Resource for Research on Human Genetic Variation: Table 1.. <i>Genome Research</i> , 1998, 8, 1229-1231.	2.4	750
112	Strategies for Mutational Analysis of the Large Multiexon ATM Gene Using High-Density Oligonucleotide Arrays. <i>Genome Research</i> , 1998, 8, 1245-1258.	2.4	116
113	Variations on a Theme: Cataloging Human DNA Sequence Variation. <i>Science</i> , 1997, 278, 1580-1581.	6.0	979
114	Gene Therapy for Cystic Fibrosis. <i>Chest</i> , 1996, 109, 241-252.	0.4	45
115	Novel missense mutation (G314R) in a cystic fibrosis patient with hepatic failure. <i>Human Mutation</i> , 1996, 7, 151-154.	1.1	3
116	THE GENETICIST'S APPROACH TO COMPLEX DISEASE. <i>Annual Review of Medicine</i> , 1996, 47, 333-353.	5.0	62
117	Transcript identification in the BRCA1 candidate region. <i>Breast Cancer Research and Treatment</i> , 1995, 33, 115-124.	1.1	2
118	A high-density microsatellite map of the ataxia-telangiectasia locus. <i>Human Genetics</i> , 1995, 95, 451-454.	1.8	56
119	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> , 1995, 82, 831-840.	13.5	372
120	A single ataxia telangiectasia gene with a product similar to PI-3 kinase. <i>Science</i> , 1995, 268, 1749-1753.	6.0	2,634
121	Nonsense mutations at Arg-1947 in two cases of familial neurofibromatosis type 1 in Japanese. <i>Human Genetics</i> , 1994, 93, 81-3.	1.8	25
122	Familial breast cancer. Approaching the isolation of a susceptibility gene. <i>Cancer</i> , 1994, 74, 1013-1020.	2.0	11
123	Loss of neurofibromin in adrenal gland tumors from patients with neurofibromatosis type I. <i>Genes Chromosomes and Cancer</i> , 1994, 10, 55-58.	1.5	67
124	A YAC Contig Spanning the Ataxia-Telangiectasia Locus (Groups A and C) at 11q22-q23. <i>Genomics</i> , 1994, 24, 234-242.	1.3	8
125	An African-American cystic fibrosis patient homozygous for a novel frameshift mutation associated with reduced CFTR mRNA levels. <i>Human Mutation</i> , 1993, 2, 148-151.	1.1	13
126	The human NME2 gene lies within 18kb of NME1 in chromosome 17. <i>Genes Chromosomes and Cancer</i> , 1993, 6, 245-248.	1.5	23

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127	Mutations in the neurofibromatosis 1 gene in sporadic malignant melanoma cell lines. Nature Genetics, 1993, 3, 118-121.	9.4	147
128	Somatic deletion of the neurofibromatosis type 1 gene in a neurofibrosarcoma supports a tumour suppressor gene hypothesis. Nature Genetics, 1993, 3, 122-126.	9.4	394
129	A cosmid contig and high resolution restriction map of the 2 megabase region containing the Huntington's disease gene. Nature Genetics, 1993, 4, 181-186.	9.4	102
130	Magnetic bead capture of expressed sequences encoded within large genomic segments. Nature, 1993, 361, 751-753.	13.7	97
131	Molecular basis of defective anion transport in L cells expressing recombinant forms of CFTR. Human Molecular Genetics, 1993, 2, 1253-1261.	1.4	83
132	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. Human Molecular Genetics, 1993, 2, 225-230.	1.4	100
133	Localization of the cystic fibrosis transmembrane conductance regulator in human bile duct epithelial cells. Gastroenterology, 1993, 105, 1857-1864.	0.6	330
134	Molecular Biology of Cystic Fibrosis. , 1993, 3, 33-68.		32
135	Sequencing and analysis of genomic fragments from the <i>NF1</i> locus. DNA Sequence, 1992, 3, 237-243.	0.7	11
136	Aberrant regulation of ras proteins in malignant tumour cells from type 1 neurofibromatosis patients. Nature, 1992, 356, 713-715.	13.7	653
137	A welcome animal model. Nature, 1992, 358, 708-709.	13.7	16
138	More from the modellers. Nature, 1992, 359, 195-196.	13.7	13
139	Positional cloning: Let's not call it reverse anymore. Nature Genetics, 1992, 1, 3-6.	9.4	456
140	The Huntington's disease candidate region exhibits many different haplotypes. Nature Genetics, 1992, 1, 99-103.	9.4	157
141	Characterization of an intron 12 splice donor mutation in the cystic fibrosis transmembrane conductance regulator (CFTR) gene. Human Mutation, 1992, 1, 380-387.	1.1	27
142	Recent progress toward understanding the molecular biology of von Recklinghausen neurofibromatosis. Annals of Neurology, 1992, 31, 555-561.	2.8	51
143	cDNA cloning of the type 1 neurofibromatosis gene: Complete sequence of the NF1 gene product. Genomics, 1991, 11, 931-940.	1.3	384
144	The Genome Project and human health. FASEB Journal, 1991, 5, 77-77.	0.2	15

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145	A de novo Alu insertion results in neurofibromatosis type 1. <i>Nature</i> , 1991, 353, 864-866.	13.7	444
146	Cystic Fibrosis Gene Mutation in Two Sisters with Mild Disease and Normal Sweat Electrolyte Levels. <i>New England Journal of Medicine</i> , 1991, 325, 1630-1634.	13.9	112
147	Molecular Genetics of von Recklinghausen Neurofibromatosis. , 1991, 20, 267-307.		9
148	The Cystic Fibrosis Gene: Isolation and Significance. <i>Hospital Practice (1995)</i> , 1990, 25, 47-57.	0.5	32
149	A chromosome jump crosses a translocation breakpoint in the von recklinghausen neurofibromatosis region. <i>Genes Chromosomes and Cancer</i> , 1990, 2, 271-277.	1.5	22
150	Mutation Analysis for Heterozygote Detection and the Prenatal Diagnosis of Cystic Fibrosis. <i>New England Journal of Medicine</i> , 1990, 322, 291-296.	13.9	224
151	Type 1 neurofibromatosis gene: identification of a large transcript disrupted in three NF1 patients. <i>Science</i> , 1990, 249, 181-186.	6.0	1,433
152	Type 1 neurofibromatosis gene: correction. <i>Science</i> , 1990, 250, 1749-1749.	6.0	0
153	Reverse Genetics and Cystic Fibrosis. <i>American Journal of Respiratory Cell and Molecular Biology</i> , 1990, 2, 309-316.	1.4	10
154	Approaches to localizing disease genes as applied to cystic fibrosis. <i>Nucleic Acids Research</i> , 1990, 18, 345-350.	6.5	24
155	Correction of the cystic fibrosis defect in vitro by retrovirus-mediated gene transfer. <i>Cell</i> , 1990, 62, 1227-1233.	13.5	595
156	<i>Response</i> : Type 1 Neurofibromatosis Gene: Correction. <i>Science</i> , 1990, 250, 1749-1749.	6.0	0
157	<i>Response</i> : Type 1 Neurofibromatosis Gene: Correction. <i>Science</i> , 1990, 250, 1749-1749.	6.0	0
158	Mutations in the p53 gene occur in diverse human tumour types. <i>Nature</i> , 1989, 342, 705-708.	13.7	2,702
159	Identification of the cystic fibrosis gene: chromosome walking and jumping. <i>Science</i> , 1989, 245, 1059-1065.	6.0	3,136
160	Two patients with ring chromosome 15 syndrome. <i>American Journal of Medical Genetics Part A</i> , 1988, 29, 149-154.	2.4	63
161	Physical mapping of the cystic fibrosis region by pulsed-field gel electrophoresis. <i>Genomics</i> , 1988, 2, 346-354.	1.3	51
162	Adult Turner syndrome associated with chylous ascites and vascular anomalies. <i>Clinical Genetics</i> , 1987, 31, 218-223.	1.0	3

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163	Molecular Analysis of Deletion and Nondeletion Hereditary Persistence of Fetal Hemoglobin and Identification of a New Mutation Causing β -Thalassemia. <i>Annals of the New York Academy of Sciences</i> , 1985, 445, 159-169.	1.8	11