

# Len A Pennacchio

## 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.

157  
papers

26,492  
citations

78  
h-index

162  
g-index

177  
ext. papers

31,346  
ext. citations

19.3  
avg, IF

6.36  
L-index

#	Paper	IF	Citations
157	Characterization of Mammalian In Vivo Enhancers Using Mouse Transgenesis and CRISPR Genome Editing.. <i>Methods in Molecular Biology</i> , <b>2022</b> , 2403, 147-186	1.4	0
156	Differential Etv2 threshold requirement for endothelial and erythropoietic development. <i>Cell Reports</i> , <b>2022</b> , 39, 110881	10.6	0
155	Long-read metagenomics of soil communities reveals phylum-specific secondary metabolite dynamics. <i>Communications Biology</i> , <b>2021</b> , 4, 1302	6.7	2
154	Perfect and imperfect views of ultraconserved sequences. <i>Nature Reviews Genetics</i> , <b>2021</b> ,	30.1	1
153	Ultraconserved enhancer function does not require perfect sequence conservation. <i>Nature Genetics</i> , <b>2021</b> , 53, 521-528	36.3	10
152	Deletion of a non-canonical regulatory sequence causes loss of Scn1a expression and epileptic phenotypes in mice. <i>Genome Medicine</i> , <b>2021</b> , 13, 69	14.4	5
151	Reactivation of a developmentally silenced embryonic globin gene. <i>Nature Communications</i> , <b>2021</b> , 12, 4439	17.4	5
150	Coding and noncoding variants in EBF3 are involved in HADDs and simplex autism. <i>Human Genomics</i> , <b>2021</b> , 15, 44	6.8	3
149	HAND transcription factors cooperatively specify the aorta and pulmonary trunk. <i>Developmental Biology</i> , <b>2021</b> , 476, 1-10	3.1	1
148	Transcriptional network orchestrating regional patterning of cortical progenitors.. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2021</b> , 118,	11.5	2
147	Presynaptic Homeostasis Opposes Disease Progression in Mouse Models of ALS-Like Degeneration: Evidence for Homeostatic Neuroprotection. <i>Neuron</i> , <b>2020</b> , 107, 95-111.e6	13.9	18
146	Comprehensive InVivo Interrogation Reveals Phenotypic Impact of Human Enhancer Variants. <i>Cell</i> , <b>2020</b> , 180, 1262-1271.e15	56.2	34
145	ATAC-Seq Reveals an Enhancer That Regulates Sinoatrial Node Development and Function. <i>Circulation Research</i> , <b>2020</b> , 127, 1502-1518	15.7	11
144	An atlas of dynamic chromatin landscapes in mouse fetal development. <i>Nature</i> , <b>2020</b> , 583, 744-751	50.4	76
143	Spatiotemporal DNA methylome dynamics of the developing mouse fetus. <i>Nature</i> , <b>2020</b> , 583, 752-759	50.4	35
142	Expanded encyclopaedias of DNA elements in the human and mouse genomes. <i>Nature</i> , <b>2020</b> , 583, 699-710	50.4	360
141	The changing mouse embryo transcriptome at whole tissue and single-cell resolution. <i>Nature</i> , <b>2020</b> , 583, 760-767	50.4	39

140	Supervised enhancer prediction with epigenetic pattern recognition and targeted validation. <i>Nature Methods</i> , <b>2020</b> , 17, 807-814	21.6	24
139	Noncoding deletions reveal a gene that is critical for intestinal function. <i>Nature</i> , <b>2019</b> , 571, 107-111	50.4	12
138	TIMELESS mutation alters phase responsiveness and causes advanced sleep phase. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2019</b> , 116, 12045-12053	11.5	30
137	Dynamic BAF chromatin remodeling complex subunit inclusion promotes temporally distinct gene expression programs in cardiogenesis. <i>Development (Cambridge)</i> , <b>2019</b> , 146,	6.6	17
136	Genomic Resolution of DLX-Orchestrated Transcriptional Circuits Driving Development of Forebrain GABAergic Neurons. <i>Cell Reports</i> , <b>2019</b> , 28, 2048-2063.e8	10.6	29
135	Stable enhancers are active in development, and fragile enhancers are associated with evolutionary adaptation. <i>Genome Biology</i> , <b>2019</b> , 20, 140	18.3	3
134	Single-nucleus analysis of accessible chromatin in developing mouse forebrain reveals cell-type-specific transcriptional regulation. <i>Nature Neuroscience</i> , <b>2018</b> , 21, 432-439	25.5	172
133	Enhancer redundancy provides phenotypic robustness in mammalian development. <i>Nature</i> , <b>2018</b> , 554, 239-243	50.4	275
132	Ultraconserved Enhancers Are Required for Normal Development. <i>Cell</i> , <b>2018</b> , 172, 491-499.e15	56.2	101
131	Relationship between genetic variation at PPP1R3B and levels of liver glycogen and triglyceride. <i>Hepatology</i> , <b>2018</b> , 67, 2182-2195	11.2	37
130	Parkinson-Associated SNCA Enhancer Variants Revealed by Open Chromatin in Mouse Dopamine Neurons. <i>American Journal of Human Genetics</i> , <b>2018</b> , 103, 874-892	11	15
129	Improved regulatory element prediction based on tissue-specific local epigenomic signatures. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2017</b> , 114, E1633-E1640	11.5	60
128	Genomic Patterns of De Novo Mutation in Simplex Autism. <i>Cell</i> , <b>2017</b> , 171, 710-722.e12	56.2	189
127	Limb-Enhancer Genie: An accessible resource of accurate enhancer predictions in the developing limb. <i>PLoS Computational Biology</i> , <b>2017</b> , 13, e1005720	5	12
126	Germline Chd8 haploinsufficiency alters brain development in mouse. <i>Nature Neuroscience</i> , <b>2017</b> , 20, 1062-1073	25.5	136
125	52 Genetic Loci Influencing Myocardial Mass. <i>Journal of the American College of Cardiology</i> , <b>2016</b> , 68, 1435-1448	15.1	76
124	Genome-wide compendium and functional assessment of in vivo heart enhancers. <i>Nature Communications</i> , <b>2016</b> , 7, 12923	17.4	51
123	Progressive Loss of Function in a Limb Enhancer during Snake Evolution. <i>Cell</i> , <b>2016</b> , 167, 633-642.e11	56.2	160

122	The Epigenomic Landscape of Prokaryotes. <i>PLoS Genetics</i> , <b>2016</b> , 12, e1005854	6	198
121	A Cryptochrome 2 mutation yields advanced sleep phase in humans. <i>ELife</i> , <b>2016</b> , 5,	8.9	78
120	Genetic dissection of the $\beta$ globin super-enhancer in vivo. <i>Nature Genetics</i> , <b>2016</b> , 48, 895-903	36.3	204
119	Enhancer Variants Synergistically Drive Dysfunction of a Gene Regulatory Network In Hirschsprung Disease. <i>Cell</i> , <b>2016</b> , 167, 355-368.e10	56.2	80
118	A large genomic deletion leads to enhancer adoption by the lamin B1 gene: a second path to autosomal dominant adult-onset demyelinating leukodystrophy (ADLD). <i>Human Molecular Genetics</i> , <b>2015</b> , 24, 3143-54	5.6	93
117	Brg1 modulates enhancer activation in mesoderm lineage commitment. <i>Development (Cambridge)</i> , <b>2015</b> , 142, 1418-30	6.6	61
116	Activating Mutations Affecting the Dbl Homology Domain of SOS2 Cause Noonan Syndrome. <i>Human Mutation</i> , <b>2015</b> , 36, 1080-7	4.7	51
115	Occupancy by key transcription factors is a more accurate predictor of enhancer activity than histone modifications or chromatin accessibility. <i>Epigenetics and Chromatin</i> , <b>2015</b> , 8, 16	5.8	71
114	Brg1 coordinates multiple processes during retinogenesis and is a tumor suppressor in retinoblastoma. <i>Development (Cambridge)</i> , <b>2015</b> , 142, 4092-106	6.6	20
113	Function-based identification of mammalian enhancers using site-specific integration. <i>Nature Methods</i> , <b>2014</b> , 11, 566-71	21.6	59
112	An etiologic regulatory mutation in IRF6 with loss- and gain-of-function effects. <i>Human Molecular Genetics</i> , <b>2014</b> , 23, 2711-20	5.6	39
111	Tissue-specific SMARCA4 binding at active and repressed regulatory elements during embryogenesis. <i>Genome Research</i> , <b>2014</b> , 24, 920-9	9.7	47
110	Principles of regulatory information conservation between mouse and human. <i>Nature</i> , <b>2014</b> , 515, 371-375	50.4	190
109	Functional importance of cardiac enhancer-associated noncoding RNAs in heart development and disease. <i>Journal of Molecular and Cellular Cardiology</i> , <b>2014</b> , 76, 55-70	5.8	98
108	Identification of novel craniofacial regulatory domains located far upstream of SOX9 and disrupted in Pierre Robin sequence. <i>Human Mutation</i> , <b>2014</b> , 35, 1011-20	4.7	54
107	Dynamic GATA4 enhancers shape the chromatin landscape central to heart development and disease. <i>Nature Communications</i> , <b>2014</b> , 5, 4907	17.4	102
106	Tissue-specific RNA expression marks distant-acting developmental enhancers. <i>PLoS Genetics</i> , <b>2014</b> , 10, e1004610	6	80
105	Transcriptional regulation of enhancers active in protodomains of the developing cerebral cortex. <i>Neuron</i> , <b>2014</b> , 82, 989-1003	13.9	73

104	Evolution of extreme resistance to ionizing radiation via genetic adaptation of DNA repair. <i>ELife</i> , <b>2014</b> , 3, e01322	8.9	53
103	Single site-specific integration targeting coupled with embryonic stem cell differentiation provides a high-throughput alternative to in vivo enhancer analyses. <i>Biology Open</i> , <b>2013</b> , 2, 1229-38	2.2	8
102	Rapid and pervasive changes in genome-wide enhancer usage during mammalian development. <i>Cell</i> , <b>2013</b> , 155, 1521-31	56.2	256
101	Dlx1&2-dependent expression of Zfhx1b (Sip1, Zeb2) regulates the fate switch between cortical and striatal interneurons. <i>Neuron</i> , <b>2013</b> , 77, 83-98	13.9	122
100	Enhancers: five essential questions. <i>Nature Reviews Genetics</i> , <b>2013</b> , 14, 288-95	30.1	316
99	A high-resolution enhancer atlas of the developing telencephalon. <i>Cell</i> , <b>2013</b> , 152, 895-908	56.2	189
98	Fine tuning of craniofacial morphology by distant-acting enhancers. <i>Science</i> , <b>2013</b> , 342, 1241006	33.3	157
97	Chromatin stretch enhancer states drive cell-specific gene regulation and harbor human disease risk variants. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2013</b> , 110, 17921-6	11.5	477
96	Congenital heart defects in patients with deletions upstream of SOX9. <i>Human Mutation</i> , <b>2013</b> , 34, 1628-31	3.7	27
95	Use of "MGE enhancers" for labeling and selection of embryonic stem cell-derived medial ganglionic eminence (MGE) progenitors and neurons. <i>PLoS ONE</i> , <b>2013</b> , 8, e61956	3.7	25
94	Genome resequencing reveals multiscale geographic structure and extensive linkage disequilibrium in the forest tree <i>Populus trichocarpa</i> . <i>New Phytologist</i> , <b>2012</b> , 196, 713-725	9.8	134
93	Differences in enhancer activity in mouse and zebrafish reporter assays are often associated with changes in gene expression. <i>BMC Genomics</i> , <b>2012</b> , 13, 713	4.5	16
92	Massively parallel functional dissection of mammalian enhancers in vivo. <i>Nature Biotechnology</i> , <b>2012</b> , 30, 265-70	44.5	366
91	Generation of long insert pairs using a Cre-LoxP Inverse PCR approach. <i>PLoS ONE</i> , <b>2012</b> , 7, e29437	3.7	23
90	Large-scale discovery of enhancers from human heart tissue. <i>Nature Genetics</i> , <b>2011</b> , 44, 89-93	36.3	197
89	Metagenomic discovery of biomass-degrading genes and genomes from cow rumen. <i>Science</i> , <b>2011</b> , 331, 463-7	33.3	893
88	A genetic signature of spina bifida risk from pathway-informed comprehensive gene-variant analysis. <i>PLoS ONE</i> , <b>2011</b> , 6, e28408	3.7	21
87	Genetic and functional analyses identify DISC1 as a novel callosal agenesis candidate gene. <i>American Journal of Medical Genetics, Part A</i> , <b>2011</b> , 155A, 1865-76	2.5	34

86	Targeted deletion of the 9p21 non-coding coronary artery disease risk interval in mice. <i>Nature</i> , <b>2010</b> , 464, 409-12	50.4	380
85	A restricted spectrum of NRAS mutations causes Noonan syndrome. <i>Nature Genetics</i> , <b>2010</b> , 42, 27-9	36.3	232
84	ChIP-Seq identification of weakly conserved heart enhancers. <i>Nature Genetics</i> , <b>2010</b> , 42, 806-10	36.3	343
83	Homotypic clusters of transcription factor binding sites are a key component of human promoters and enhancers. <i>Genome Research</i> , <b>2010</b> , 20, 565-77	9.7	167
82	Massively parallel sequencing identifies the gene Megf8 with ENU-induced mutation causing heterotaxy. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2009</b> , 106, 3219-24	11.5	50
81	Association of functionally significant Melanocortin-4 but not Melanocortin-3 receptor mutations with severe adult obesity in a large North American case-control study. <i>Human Molecular Genetics</i> , <b>2009</b> , 18, 1140-7	5.6	102
80	Directed evolution of ionizing radiation resistance in Escherichia coli. <i>Journal of Bacteriology</i> , <b>2009</b> , 191, 5240-52	3.5	100
79	Contrasting patterns of sequence evolution at the functionally redundant bric-1/brac paralogs in Drosophila melanogaster. <i>Journal of Molecular Evolution</i> , <b>2009</b> , 69, 194-202	3.1	4
78	The INSIG2 rs7566605 genetic variant does not play a major role in obesity in a sample of 24,722 individuals from four cohorts. <i>BMC Medical Genetics</i> , <b>2009</b> , 10, 56	2.1	12
77	ChIP-seq accurately predicts tissue-specific activity of enhancers. <i>Nature</i> , <b>2009</b> , 457, 854-8	50.4	1301
76	Genomic views of distant-acting enhancers. <i>Nature</i> , <b>2009</b> , 461, 199-205	50.4	451
75	Mutation of SHOC2 promotes aberrant protein N-myristoylation and causes Noonan-like syndrome with loose anagen hair. <i>Nature Genetics</i> , <b>2009</b> , 41, 1022-6	36.3	291
74	Functional autonomy of distant-acting human enhancers. <i>Genomics</i> , <b>2009</b> , 93, 509-13	4.3	49
73	Rare loss-of-function mutations in ANGPTL family members contribute to plasma triglyceride levels in humans. <i>Journal of Clinical Investigation</i> , <b>2009</b> , 119, 70-9	15.9	277
72	The amphioxus genome and the evolution of the chordate karyotype. <i>Nature</i> , <b>2008</b> , 453, 1064-71	50.4	1266
71	Ultraconservation identifies a small subset of extremely constrained developmental enhancers. <i>Nature Genetics</i> , <b>2008</b> , 40, 158-60	36.3	253
70	Disruption of an AP-2alpha binding site in an IRF6 enhancer is associated with cleft lip. <i>Nature Genetics</i> , <b>2008</b> , 40, 1341-7	36.3	338
69	Genetic variation in PNPLA3 confers susceptibility to nonalcoholic fatty liver disease. <i>Nature Genetics</i> , <b>2008</b> , 40, 1461-5	36.3	2115

68	Dicer, Drosha, and outcomes in patients with ovarian cancer. <i>New England Journal of Medicine</i> , <b>2008</b> , 359, 2641-50	59.2	573
67	Human-specific gain of function in a developmental enhancer. <i>Science</i> , <b>2008</b> , 321, 1346-50	33.3	260
66	Glucose regulates the expression of the apolipoprotein A5 gene. <i>Journal of Molecular Biology</i> , <b>2008</b> , 380, 789-98	6.5	16
65	Combinatorial regulation of endothelial gene expression by ets and forkhead transcription factors. <i>Cell</i> , <b>2008</b> , 135, 1053-64	56.2	245
64	The amphioxus genome illuminates vertebrate origins and cephalochordate biology. <i>Genome Research</i> , <b>2008</b> , 18, 1100-11	9.7	387
63	Association of common variants in the Joubert syndrome gene (AHI1) with autism. <i>Human Molecular Genetics</i> , <b>2008</b> , 17, 3887-96	5.6	65
62	Integrated analysis of homozygous deletions, focal amplifications, and sequence alterations in breast and colorectal cancers. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2008</b> , 105, 16224-9	11.5	230
61	Gain-of-function SOS1 mutations cause a distinctive form of Noonan syndrome. <i>Nature Genetics</i> , <b>2007</b> , 39, 75-9	36.3	440
60	Population-based resequencing of ANGPTL4 uncovers variations that reduce triglycerides and increase HDL. <i>Nature Genetics</i> , <b>2007</b> , 39, 513-6	36.3	423
59	Gain-of-function RAF1 mutations cause Noonan and LEOPARD syndromes with hypertrophic cardiomyopathy. <i>Nature Genetics</i> , <b>2007</b> , 39, 1007-12	36.3	523
58	VISTA Enhancer Browser--a database of tissue-specific human enhancers. <i>Nucleic Acids Research</i> , <b>2007</b> , 35, D88-92	20.1	700
57	Deletion of ultraconserved elements yields viable mice. <i>PLoS Biology</i> , <b>2007</b> , 5, e234	9.7	217
56	Predicting tissue-specific enhancers in the human genome. <i>Genome Research</i> , <b>2007</b> , 17, 201-11	9.7	107
55	Enhancer identification through comparative genomics. <i>Seminars in Cell and Developmental Biology</i> , <b>2007</b> , 18, 140-52	7.5	81
54	In vivo characterization of human APOA5 haplotypes. <i>Genomics</i> , <b>2007</b> , 90, 674-9	4.3	22
53	Medical sequencing at the extremes of human body mass. <i>American Journal of Human Genetics</i> , <b>2007</b> , 80, 779-91	11	180
52	Most rare missense alleles are deleterious in humans: implications for complex disease and association studies. <i>American Journal of Human Genetics</i> , <b>2007</b> , 80, 727-39	11	461
51	A common allele on chromosome 9 associated with coronary heart disease. <i>Science</i> , <b>2007</b> , 316, 1488-91	33.3	1415

50	Gain-of-function R225W mutation in human AMPKgamma(3) causing increased glycogen and decreased triglyceride in skeletal muscle. <i>PLoS ONE</i> , <b>2007</b> , 2, e903	3.7	62
49	Comparative genomics: a tool to functionally annotate human DNA. <i>Methods in Molecular Biology</i> , <b>2007</b> , 366, 229-51	1.4	3
48	A PYY Q62P variant linked to human obesity. <i>Human Molecular Genetics</i> , <b>2006</b> , 15, 387-91	5.6	23
47	Close sequence comparisons are sufficient to identify human cis-regulatory elements. <i>Genome Research</i> , <b>2006</b> , 16, 855-63	9.7	154
46	Human cathepsin L rescues the neurodegeneration and lethality in cathepsin B/L double-deficient mice. <i>Biological Chemistry</i> , <b>2006</b> , 387, 885-91	4.5	43
45	In vivo enhancer analysis of human conserved non-coding sequences. <i>Nature</i> , <b>2006</b> , 444, 499-502	50.4	911
44	Array comparative genomic hybridization in patients with congenital diaphragmatic hernia: mapping of four CDH-critical regions and sequencing of candidate genes at 15q26.1-15q26.2. <i>European Journal of Human Genetics</i> , <b>2006</b> , 14, 999-1008	5.3	89
43	Transcriptional regulation of apolipoprotein A5 gene expression by the nuclear receptor RORalpha. <i>Arteriosclerosis, Thrombosis, and Vascular Biology</i> , <b>2005</b> , 25, 1186-92	9.4	45
42	In vivo characterization of a vertebrate ultraconserved enhancer. <i>Genomics</i> , <b>2005</b> , 85, 774-81	4.3	66
41	Cathepsin L is required for endothelial progenitor cell-induced neovascularization. <i>Nature Medicine</i> , <b>2005</b> , 11, 206-13	50.5	261
40	Comparative genomic analysis reveals a distant liver enhancer upstream of the COUP-TFII gene. <i>Mammalian Genome</i> , <b>2005</b> , 16, 91-5	3.2	16
39	Lack of support for the association between GAD2 polymorphisms and severe human obesity. <i>PLoS Biology</i> , <b>2005</b> , 3, e315	9.7	38
38	Insulin-mediated down-regulation of apolipoprotein A5 gene expression through the phosphatidylinositol 3-kinase pathway: role of upstream stimulatory factor. <i>Molecular and Cellular Biology</i> , <b>2005</b> , 25, 1537-48	4.8	79
37	Apolipoprotein AV accelerates plasma hydrolysis of triglyceride-rich lipoproteins by interaction with proteoglycan-bound lipoprotein lipase. <i>Journal of Biological Chemistry</i> , <b>2005</b> , 280, 21553-60	5.4	225
36	Apolipoprotein A-V deficiency results in marked hypertriglyceridemia attributable to decreased lipolysis of triglyceride-rich lipoproteins and removal of their remnants. <i>Arteriosclerosis, Thrombosis, and Vascular Biology</i> , <b>2005</b> , 25, 2573-9	9.4	107
35	Lack of MEF2A mutations in coronary artery disease. <i>Journal of Clinical Investigation</i> , <b>2005</b> , 115, 1016-20	15.9	87
34	Apoa5 Q139X truncation predisposes to late-onset hyperchylomicronemia due to lipoprotein lipase impairment. <i>Journal of Clinical Investigation</i> , <b>2005</b> , 115, 2862-9	15.9	134
33	The liver X receptor ligand T0901317 down-regulates APOA5 gene expression through activation of SREBP-1c. <i>Journal of Biological Chemistry</i> , <b>2004</b> , 279, 45462-9	5.4	69



32	Haplotypes in the APOA1-C3-A4-A5 gene cluster affect plasma lipids in both humans and baboons. <i>Human Molecular Genetics</i> , <b>2004</b> , 13, 1049-56	5.6	24
31	Analysis of apolipoprotein A5, c3, and plasma triglyceride concentrations in genetically engineered mice. <i>Arteriosclerosis, Thrombosis, and Vascular Biology</i> , <b>2004</b> , 24, 1297-302	9.4	63
30	Linkage and association between distinct variants of the APOA1/C3/A4/A5 gene cluster and familial combined hyperlipidemia. <i>Arteriosclerosis, Thrombosis, and Vascular Biology</i> , <b>2004</b> , 24, 167-74	9.4	90
29	The DNA sequence and biology of human chromosome 19. <i>Nature</i> , <b>2004</b> , 428, 529-35	50.4	216
28	The DNA sequence and comparative analysis of human chromosome 5. <i>Nature</i> , <b>2004</b> , 431, 268-74	50.4	86
27	The sequence and analysis of duplication-rich human chromosome 16. <i>Nature</i> , <b>2004</b> , 432, 988-94	50.4	129
26	Comparative genomic analysis as a tool for biological discovery. <i>Journal of Physiology</i> , <b>2004</b> , 554, 31-9	3.9	83
25	Identification of a novel enhancer of brain expression near the apoE gene cluster by comparative genomics. <i>Biochimica Et Biophysica Acta Gene Regulatory Mechanisms</i> , <b>2004</b> , 1676, 41-50		20
24	Mechanism of triglyceride lowering in mice expressing human apolipoprotein A5. <i>Biochemical and Biophysical Research Communications</i> , <b>2004</b> , 319, 397-404	3.4	163
23	Haplotype analysis of the apolipoprotein gene cluster on human chromosome 11. <i>Genomics</i> , <b>2004</b> , 83, 912-23	4.3	89
22	Apolipoprotein AIV gene variant S347 is associated with increased risk of coronary heart disease and lower plasma apolipoprotein AIV levels. <i>Circulation Research</i> , <b>2003</b> , 92, 969-75	15.7	63
21	Apolipoprotein A5, a crucial determinant of plasma triglyceride levels, is highly responsive to peroxisome proliferator-activated receptor alpha activators. <i>Journal of Biological Chemistry</i> , <b>2003</b> , 278, 17982-5	5.4	161
20	Insights from human/mouse genome comparisons. <i>Mammalian Genome</i> , <b>2003</b> , 14, 429-36	3.2	57
19	Cathepsin B but not cathepsins L or S contributes to the pathogenesis of Unverricht-Lundborg progressive myoclonus epilepsy (EPM1). <i>Journal of Neurobiology</i> , <b>2003</b> , 56, 315-27		94
18	Apolipoprotein A5, a newly identified gene that affects plasma triglyceride levels in humans and mice. <i>Arteriosclerosis, Thrombosis, and Vascular Biology</i> , <b>2003</b> , 23, 529-34	9.4	137
17	Comparative genomic tools and databases: providing insights into the human genome. <i>Journal of Clinical Investigation</i> , <b>2003</b> , 111, 1099-1106	15.9	43
16	Comparative genomic tools and databases: providing insights into the human genome. <i>Journal of Clinical Investigation</i> , <b>2003</b> , 111, 1099-106	15.9	18
15	Comparative and functional analysis of cardiovascular-related genes. <i>Pharmacogenomics</i> , <b>2003</b> , 4, 571-82.6		1

14	Relative contribution of variation within the APOC3/A4/A5 gene cluster in determining plasma triglycerides. <i>Human Molecular Genetics</i> , <b>2002</b> , 11, 3039-46	5.6	299
13	Neuropathological changes in a mouse model of progressive myoclonus epilepsy: cystatin B deficiency and Unverricht-Lundborg disease. <i>Journal of Neuropathology and Experimental Neurology</i> , <b>2002</b> , 61, 1085-91	3.1	62
12	Two independent apolipoprotein A5 haplotypes influence human plasma triglyceride levels. <i>Human Molecular Genetics</i> , <b>2002</b> , 11, 3031-8	5.6	316
11	Genomic strategies to identify mammalian regulatory sequences. <i>Nature Reviews Genetics</i> , <b>2001</b> , 2, 100-9	30.1	312
10	Progressive ataxia, myoclonic epilepsy and cerebellar apoptosis in cystatin B-deficient mice. <i>Nature Genetics</i> , <b>1998</b> , 20, 251-8	36.3	293
9	Characterization of the human neurocan gene, CSPG3. <i>Gene</i> , <b>1998</b> , 221, 199-205	3.8	17
8	Unstable minisatellite expansion causing recessively inherited myoclonus epilepsy, EPM1. <i>Nature Genetics</i> , <b>1997</b> , 15, 393-6	36.3	186
7	Structure, sequence and location of the UQCRFS1 gene for the human Rieske Fe-S protein. <i>Gene</i> , <b>1995</b> , 155, 207-11	3.8	9
6	Single nucleus analysis of the chromatin landscape in mouse forebrain development		2
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4	Spatiotemporal DNA Methylome Dynamics of the Developing Mammalian Fetus		11
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