Steven A Mccarroll

List of Publications by Year
in descending order

159 papers	111,990 citations	3531 90 h-index	6300 158 g-index
208 all docs	208 docs citations	208 times ranked	123567 citing authors

#	Article	IF	Citations
1	Whole-genome analysis of human embryonic stem cells enables rational line selection based on genetic variation. Cell Stem Cell, 2022, 29, 472-486.e7.	11.1	27
2	Mapping genomic loci implicates genes and synaptic biology in schizophrenia. Nature, 2022, 604, 502-508.	27.8	929
3	Rare coding variants in ten genes confer substantial risk for schizophrenia. Nature, 2022, 604, 509-516.	27.8	326
4	The 22q11.2 region regulates presynaptic gene-products linked to schizophrenia. Nature Communications, 2022, 13, .	12.8	22
5	Chromosomal phase improves aneuploidy detection in non-invasive prenatal testing at low fetal DNA fractions. Scientific Reports, 2022, 12, .	3.3	1
6	Genome-Wide Association Studies of Schizophrenia and Bipolar Disorder in a Diverse Cohort of US Veterans. Schizophrenia Bulletin, 2021, 47, 517-529.	4.3	48
7	Overexpression of schizophrenia susceptibility factor human complement C4A promotes excessive synaptic loss and behavioral changes in mice. Nature Neuroscience, 2021, 24, 214-224.	14.8	158
8	Human DDK rescues stalled forks and counteracts checkpoint inhibition at unfired origins to complete DNA replication. Molecular Cell, 2021, 81, 426-441.e8.	9.7	21
9	Genome-wide association study of more than 40,000 bipolar disorder cases provides new insights into the underlying biology. Nature Genetics, 2021, 53, 817-829.	21.4	629
10	Early role for a Na ⁺ ,K ⁺ -ATPase (ATP1A3) in brain development. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	7.1	20
11	Hematopoietic mosaic chromosomal alterations increase the risk for diverse types of infection. Nature Medicine, 2021, 27, 1012-1024.	30.7	109
12	Anterior thalamic dysfunction underlies cognitive deficits in a subset of neuropsychiatric disease models. Neuron, 2021, 109, 2590-2603.e13.	8.1	34
13	Single cell analysis of DNA in more than 10,000 individual sperm from men with abnormal reproductive outcomes. Journal of Assisted Reproduction and Genetics, 2021, 38, 2975-2983.	2.5	2
14	Protein-coding repeat polymorphisms strongly shape diverse human phenotypes. Science, 2021, 373, 1499-1505.	12.6	96
15	Prognostic value of polygenic risk scores for adults with psychosis. Nature Medicine, 2021, 27, 1576-1581.	30.7	31
16	Investigating rare pathogenic/likely pathogenic exonic variation in bipolar disorder. Molecular Psychiatry, 2021, 26, 5239-5250.	7.9	15
17	Comparative cellular analysis of motor cortex in human, marmoset and mouse. Nature, 2021, 598, 111-119.	27.8	361
18	A multimodal cell census and atlas of the mammalian primary motor cortex. Nature, 2021, 598, 86-102.	27.8	316

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19	Contributions of common genetic variants to risk of schizophrenia among individuals of African and Latino ancestry. Molecular Psychiatry, 2020, 25, 2455-2467.	7.9	82
20	Exome sequencing in schizophrenia-affected parentâ€“offspring trios reveals risk conferred by protein-coding de novo mutations. Nature Neuroscience, 2020, 23, 185-193.	14.8	125
21	Differential excitatory vs inhibitory SCN expression at single cell level regulates brain sodium channel function in neurodevelopmental disorders. European Journal of Paediatric Neurology, 2020, 24, 129-133.	1.6	18
22	Innovations present in the primate interneuron repertoire. Nature, 2020, 586, 262-269.	27.8	206
23	Publicly Available hiPSC Lines with Extreme Polygenic Risk Scores for Modeling Schizophrenia. Complex Psychiatry, 2020, 6, 68-82.	0.9	18
24	Complement genes contribute sex-biased vulnerability in diverse disorders. Nature, 2020, 582, 577-581.	27.8	158
25	Insights into variation in meiosis from 31,228 human sperm genomes. Nature, 2020, 583, 259-264.	27.8	73
26	The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 2020, 581, 434-443.	27.8	6,140
27	Chromosomal alterations among age-related haematopoietic clones in Japan. Nature, 2020, 584, 130-135.	27.8	102
28	Monogenic and polygenic inheritance become instruments for clonal selection. Nature, 2020, 584, 136-141.	27.8	119
29	Single-cell RNA sequencing reveals compromised immune microenvironment in precursor stages of multiple myeloma. Nature Cancer, 2020, 1, 493-506.	13.2	209
30	Ultra-Rare Genetic Variation in the Epilepsies: A Whole-Exome Sequencing Study of 17,606 Individuals. American Journal of Human Genetics, 2019, 105, 267-282.	6.2	237
31	SA122STRUCTURAL VARIATIONS OF SCHIZOPHRENIA RISK GENE COMPLEMENT COMPONENT 4 (C4) AND BRAIN MRI PHENOTYPES. European Neuropsychopharmacology, 2019, 29, S1255-S1256.	0.7	0
32	THE GENOMICS OF BIPOLAR AND SCHIZOPHRENIC DISORDERS IN A LARGE PEDIGREE FROM A NORTHERN SWEDISH ISOLATE. European Neuropsychopharmacology, 2019, 29, S902-S903.	0.7	0
33	Multi-platform discovery of haplotype-resolved structural variation in human genomes. Nature Communications, 2019, 10, 1784.	12.8	636
34	Phenotypic Landscape of Schizophrenia-Associated Genes Defines Candidates and Their Shared Functions. Cell, 2019, 177, 478-491.e20.	28.9	159
35	169. New Technology for Learning About Genetic EffectsÂon Brain Cells and Brain Tissue. Biological Psychiatry, 2019, 85, S70.	1.3	0
36	Comparative genetic architectures of schizophrenia in East Asian and European populations. Nature Genetics, 2019, 51, 1670-1678.	21.4	440

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37	Genetic predisposition to mosaic Y chromosome loss in blood. Nature, 2019, 575, 652-657.	27.8	198
38	Single-cell RNA sequencing reveals compromised immune microenvironment in precursor stages of multiple myeloma. Clinical Lymphoma, Myeloma and Leukemia, 2019, 19, e27.	0.4	0
39	Single-Cell RNA Sequencing of Microglia throughout the Mouse Lifespan and in the Injured Brain Reveals Complex Cell-State Changes. Immunity, 2019, 50, 253-271.e6.	14.3	1,351
40	Contribution of Rare Copy Number Variants toÂBipolar Disorder Risk Is Limited to Schizoaffective Cases. Biological Psychiatry, 2019, 86, 110-119.	1.3	45
41	Common schizophrenia alleles are enriched in mutation-intolerant genes and in regions under strong background selection. Nature Genetics, 2018, 50, 381-389.	21.4	1,332
42	Heritability enrichment of specifically expressed genes identifies disease-relevant tissues and cell types. Nature Genetics, 2018, 50, 621-629.	21.4	807
43	Transcriptome-wide association study of schizophrenia and chromatin activity yields mechanistic disease insights. Nature Genetics, 2018, 50, 538-548.	21.4	406
44	Increased neutrophil extracellular trap formation promotes thrombosis in myeloproliferative neoplasms. Science Translational Medicine, 2018, 10, .	12.4	299
45	Polygenic risk for schizophrenia and measured domains of cognition in individuals with psychosis and controls. Translational Psychiatry, 2018, 8, 78.	4.8	49
46	Using Droplet Digital PCR to Analyze Allele-Specific RNA Expression. Methods in Molecular Biology, 2018, 1768, 401-422.	0.9	8
47	Analyzing Copy Number Variation with Droplet Digital PCR. Methods in Molecular Biology, 2018, 1768, 143-160.	0.9	37
48	An analytical framework for whole-genome sequence association studies and its implications for autism spectrum disorder. Nature Genetics, 2018, 50, 727-736.	21.4	235
49	The Genetic Landscape of Diamond-Blackfan Anemia. American Journal of Human Genetics, 2018, 103, 930-947.	6.2	184
50	A whole-genome sequence study identifies genetic risk factors for neuromyelitis optica. Nature Communications, 2018, 9, 1929.	12.8	73
51	Deep coverage whole genome sequences and plasma lipoprotein(a) in individuals of European and African ancestries. Nature Communications, 2018, 9, 2606.	12.8	79
52	Insights into clonal haematopoiesis from 8,342 mosaic chromosomal alterations. Nature, 2018, 559, 350-355.	27.8	279
53	Molecular Diversity and Specializations among the Cells of the Adult Mouse Brain. Cell, 2018, 174, 1015-1030.e16.	28.9	1,231
54	Common Î±-globin variants modify hematologic and other clinical phenotypes in sickle cell trait and disease. PLoS Genetics, 2018, 14, e1007293.	3.5	45

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55	Single-Cell RNA Sequencing Reveals Compromised Immune Microenvironment in Precursor Stages of Multiple Myeloma. Blood, 2018, 132, 2603-2603.	1.4	1
56	A molecular census of arcuate hypothalamus and median eminence cell types. Nature Neuroscience, 2017, 20, 484-496.	14.8	635
57	Cell diversity and network dynamics in photosensitive human brain organoids. Nature, 2017, 545, 48-53.	27.8	933
58	Human pluripotent stem cells recurrently acquire and expand dominant negative P53 mutations. Nature, 2017, 545, 229-233.	27.8	409
59	Genetically Distinct Parallel Pathways in the Entopeduncular Nucleus for Limbic and Sensorimotor Output of the Basal Ganglia. Neuron, 2017, 94, 138-152.e5.	8.1	146
60	Damaging Missense De Novo Coding Mutations Contribute To Schizophrenia Risk. European Neuropsychopharmacology, 2017, 27, S427-S428.	0.7	0
61	A Loss-of-Function Splice Acceptor Variant in IGF2 Is Protective for Type 2 Diabetes. Diabetes, 2017, 66, 2903-2914.	0.6	52
62	Ultra-Rare Protein-Altering Variants Among 4,877 Swedish Individuals with Schizophrenia. European Neuropsychopharmacology, 2017, 27, S426-S427.	0.7	0
63	Mutation intolerant genes and targets of FMRP are enriched for nonsynonymous alleles in schizophrenia. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2017, 174, 724-731.	1.7	19
64	Whole genome sequencing in psychiatric disorders: the WGSPD consortium. Nature Neuroscience, 2017, 20, 1661-1668.	14.8	122
65	Contribution of copy number variants to schizophrenia from a genome-wide study of 41,321 subjects. Nature Genetics, 2017, 49, 27-35.	21.4	838
66	Punctuated bursts in human male demography inferred from 1,244 worldwide Y-chromosome sequences. Nature Genetics, 2016, 48, 593-599.	21.4	273
67	Increased burden of ultra-rare protein-altering variants among 4,877 individuals with schizophrenia. Nature Neuroscience, 2016, 19, 1433-1441.	14.8	427
68	Ultra-rare disruptive and damaging mutations influence educational attainment in the general population. Nature Neuroscience, 2016, 19, 1563-1565.	14.8	90
69	A reference panel of 64,976 haplotypes for genotype imputation. Nature Genetics, 2016, 48, 1279-1283.	21.4	2,421
70	Comprehensive Classification of Retinal Bipolar Neurons by Single-Cell Transcriptomics. Cell, 2016, 166, 1308-1323.e30.	28.9	1,010
71	Analysis of protein-coding genetic variation in 60,706 humans. Nature, 2016, 536, 285-291.	27.8	9,051
72	Schizophrenia risk from complex variation of complement component 4. Nature, 2016, 530, 177-183.	27.8	1,915

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73	Quantifying prion disease penetrance using large population control cohorts. Science Translational Medicine, 2016, 8, 322ra9.	12.4	289
74	Polygenic risk for type 2 diabetes mellitus among individuals with psychosis and their relatives. Journal of Psychiatric Research, 2016, 77, 52-58.	3.1	22
75	Evidence of Common Genetic Overlap Between Schizophrenia and Cognition. Schizophrenia Bulletin, 2016, 42, 832-842.	4.3	102
76	Recurring exon deletions in the HP (haptoglobin) gene contribute to lower blood cholesterol levels. Nature Genetics, 2016, 48, 359-366.	21.4	93
77	Genome-wide association study identifies SESTD1 as a novel risk gene for lithium-responsive bipolar disorder. Molecular Psychiatry, 2016, 21, 1290-1297.	7.9	69
78	A Rapid Molecular Approach for Chromosomal Phasing. PLoS ONE, 2015, 10, e0118270.	2.5	58
79	Highly Parallel Genome-wide Expression Profiling of Individual Cells Using Nanoliter Droplets. Cell, 2015, 161, 1202-1214.	28.9	5,908
80	Novel Findings from CNVs Implicate Inhibitory and Excitatory Signaling Complexes in Schizophrenia. Neuron, 2015, 86, 1203-1214.	8.1	173
81	Genetic studies of body mass index yield new insights for obesity biology. Nature, 2015, 518, 197-206.	27.8	3,823
82	Large multiallelic copy number variations in humans. Nature Genetics, 2015, 47, 296-303.	21.4	357
83	No evidence for rare recessive and compound heterozygous disruptive variants in schizophrenia. European Journal of Human Genetics, 2015, 23, 555-557.	2.8	21
84	Structural forms of the human amylase locus and their relationships to SNPs, haplotypes and obesity. Nature Genetics, 2015, 47, 921-925.	21.4	120
85	Lymphatic and Other Vascular Malformative/Overgrowth Disorders AreÂCaused by Somatic Mutations in PIK3CA. Journal of Pediatrics, 2015, 166, 1048-1054.e5.	1.8	429
86	A global reference for human genetic variation. Nature, 2015, 526, 68-74.	27.8	13,998
87	An integrated map of structural variation in 2,504 human genomes. Nature, 2015, 526, 75-81.	27.8	1,994
88	Clonal Hematopoiesis and Blood-Cancer Risk. New England Journal of Medicine, 2015, 372, 1071-1072.	27.0	57
89	Complex and multi-allelic copy number variation in human disease. Briefings in Functional Genomics, 2015, 14, 329-338.	2.7	50
90	New data and an old puzzle: the negative association between schizophrenia and rheumatoid arthritis. International Journal of Epidemiology, 2015, 44, 1706-1721.	1.9	53

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91	SnapShot-Seq: A Method for Extracting Genome-Wide, In Vivo mRNA Dynamics from a Single Total RNA Sample. PLoS ONE, 2014, 9, e89673.	2.5	53
92	Analysis of copy number variations at 15 schizophrenia-associated loci. British Journal of Psychiatry, 2014, 204, 108-114.	2.8	380
93	Clonal Hematopoiesis and Blood-Cancer Risk Inferred from Blood DNA Sequence. New England Journal of Medicine, 2014, 371, 2477-2487.	27.0	2,669
94	CNV analysis in a large schizophrenia sample implicates deletions at 16p12.1 and SLC1A1 and duplications at 1p36.33 and CGNL1. Human Molecular Genetics, 2014, 23, 1669-1676.	2.9	82
95	De novo CNVs in bipolar affective disorder and schizophrenia. Human Molecular Genetics, 2014, 23, 6677-6683.	2.9	70
96	The Penetrance of Copy Number Variations for Schizophrenia and Developmental Delay. Biological Psychiatry, 2014, 75, 378-385.	1.3	321
97	Pathways Disrupted in Human ALS Motor Neurons Identified through Genetic Correction of Mutant SOD1. Cell Stem Cell, 2014, 14, 781-795.	11.1	392
98	Genome-scale neurogenetics: methodology and meaning. Nature Neuroscience, 2014, 17, 756-763.	14.8	82
99	De novo mutations in schizophrenia implicate synaptic networks. Nature, 2014, 506, 179-184.	27.8	1,510
100	A polygenic burden of rare disruptive mutations in schizophrenia. Nature, 2014, 506, 185-190.	27.8	1,305
101	Random replication of the inactive X chromosome. Genome Research, 2014, 24, 64-69.	5.5	65
102	Partitioning Heritability of Regulatory and Cell-Type-Specific Variants across 11 Common Diseases. American Journal of Human Genetics, 2014, 95, 535-552.	6.2	569
103	Genetic Variation in Human DNA Replication Timing. Cell, 2014, 159, 1015-1026.	28.9	149
104	Cryptic and Complex Chromosomal Aberrations in Early-Onset Neuropsychiatric Disorders. American Journal of Human Genetics, 2014, 95, 454-461.	6.2	45
105	Of Rats and Men. Cell, 2013, 154, 481-483.	28.9	4
106	Genome-wide association analysis identifies 13 new risk loci for schizophrenia. Nature Genetics, 2013, 45, 1150-1159.	21.4	1,395
107	Mapping the Human Reference Genomeâ€™s Missing Sequence by Three-Way Admixture in Latino Genomes. American Journal of Human Genetics, 2013, 93, 411-421.	6.2	36
108	Our Fallen Genomes. Science, 2013, 342, 564-565.	12.6	8

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109	Genetic relationship between five psychiatric disorders estimated from genome-wide SNPs. Nature Genetics, 2013, 45, 984-994.	21.4	2,067
110	Progress in the Genetics of Polygenic Brain Disorders: Significant New Challenges for Neurobiology. Neuron, 2013, 80, 578-587.	8.1	74
111	Using population admixture to help complete maps of the human genome. Nature Genetics, 2013, 45, 406-414.	21.4	61
112	Identification of heart rateâ€“associated loci and their effects on cardiac conduction and rhythm disorders. Nature Genetics, 2013, 45, 621-631.	21.4	282
113	Mutational heterogeneity in cancer and the search for new cancer-associated genes. Nature, 2013, 499, 214-218.	27.8	4,761
114	Population Perspectives on Genome Variation and Complex Disease. , 2013, , 41-49.		0
115	Genome-wide association analysis of red blood cell traits in African Americans: the COGENT Network. Human Molecular Genetics, 2013, 22, 2529-2538.	2.9	57
116	zCall: a rare variant caller for array-based genotyping. Bioinformatics, 2012, 28, 2543-2545.	4.1	195
117	Exploring the variation within. Nature Genetics, 2012, 44, 614-616.	21.4	21
118	Differential Relationship of DNA Replication Timing to Different Forms of Human Mutation and Variation. American Journal of Human Genetics, 2012, 91, 1033-1040.	6.2	220
119	Whole-Genome Sequencing in Autism Identifies Hot Spots for De Novo Germline Mutation. Cell, 2012, 151, 1431-1442.	28.9	501
120	Discovery and Statistical Genotyping of Copy-Number Variation from Whole-Exome Sequencing Depth. American Journal of Human Genetics, 2012, 91, 597-607.	6.2	513
121	Structural haplotypes and recent evolution of the human 17q21.31 region. Nature Genetics, 2012, 44, 881-885.	21.4	124
122	A Systematic Survey of Loss-of-Function Variants in Human Protein-Coding Genes. Science, 2012, 335, 823-828.	12.6	1,095
123	Discovery and genotyping of genome structural polymorphism by sequencing on a population scale. Nature Genetics, 2011, 43, 269-276.	21.4	299
124	Mapping copy number variation by population-scale genome sequencing. Nature, 2011, 470, 59-65.	27.8	991
125	Genetic association analysis highlights new loci that modulate hematological trait variation in Caucasians and African Americans. Human Genetics, 2011, 129, 307-317.	3.8	81
126	Single-Tissue and Cross-Tissue Heritability of Gene Expression Via Identity-by-Descent in Related or Unrelated Individuals. PLoS Genetics, 2011, 7, e1001317.	3.5	173

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127	Mapping a New Spontaneous Preterm Birth Susceptibility Gene, IGF1R, Using Linkage, Haplotype Sharing, and Association Analysis. PLoS Genetics, 2011, 7, e1001293.	3.5	61
128	Meta-analysis identifies 13 new loci associated with waist-hip ratio and reveals sexual dimorphism in the genetic basis of fat distribution. Nature Genetics, 2010, 42, 949-960.	21.4	836
129	Association analyses of 249,796 individuals reveal 18 new loci associated with body mass index. Nature Genetics, 2010, 42, 937-948.	21.4	2,634
130	Copy number variation and human genome maps. Nature Genetics, 2010, 42, 365-366.	21.4	20
131	New genetic loci implicated in fasting glucose homeostasis and their impact on type 2 diabetes risk. Nature Genetics, 2010, 42, 105-116.	21.4	1,982
132	Accurately Assessing the Risk of Schizophrenia Conferred by Rare Copy-Number Variation Affecting Genes with Brain Function. PLoS Genetics, 2010, 6, e1001097.	3.5	134
133	Genome Variation and Donor-Recipient Compatibility in Graft-Versus-Host Disease. Blood, 2010, 116, SCI-12-SCI-12.	1.4	0
134	On the level: IRGM gene function is all about expression. Autophagy, 2009, 5, 96-99.	9.1	10
135	Finding the missing heritability of complex diseases. Nature, 2009, 461, 747-753.	27.8	7,490
136	Mapping duplicated sequences. Nature Biotechnology, 2009, 27, 1001-1002.	17.5	5
137	Six new loci associated with body mass index highlight a neuronal influence on body weight regulation. Nature Genetics, 2009, 41, 25-34.	21.4	1,572
138	Genome-wide association of early-onset myocardial infarction with single nucleotide polymorphisms and copy number variants. Nature Genetics, 2009, 41, 334-341.	21.4	990
139	De novo copy number variants identify new genes and loci in isolated sporadic tetralogy of Fallot. Nature Genetics, 2009, 41, 931-935.	21.4	373
140	Donor-recipient mismatch for common gene deletion polymorphisms in graft-versus-host disease. Nature Genetics, 2009, 41, 1341-1344.	21.4	91
141	Improved detection of global copy number variation using high density, non-polymorphic oligonucleotide probes. BMC Genetics, 2008, 9, 27.	2.7	27
142	Mapping and sequencing of structural variation from eight human genomes. Nature, 2008, 453, 56-64.	27.8	983
143	Deletion polymorphism upstream of IRGM associated with altered IRGM expression and Crohn's disease. Nature Genetics, 2008, 40, 1107-1112.	21.4	604
144	Integrated genotype calling and association analysis of SNPs, common copy number polymorphisms and rare CNVs. Nature Genetics, 2008, 40, 1253-1260.	21.4	712

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145	Integrated detection and population-genetic analysis of SNPs and copy number variation. Nature Genetics, 2008, 40, 1166-1174.	21.4	838
146	Copy-number analysis goes more than skin deep. Nature Genetics, 2008, 40, 5-6.	21.4	35
147	Extending genome-wide association studies to copy-number variation. Human Molecular Genetics, 2008, 17, R135-R142.	2.9	154
148	Evaluation of Common Variants in the Six Known Maturity-Onset Diabetes of the Young (MODY) Genes for Association With Type 2 Diabetes. Diabetes, 2007, 56, 685-693.	0.6	178
149	Copy-number variation and association studies of human disease. Nature Genetics, 2007, 39, S37-S42.	21.4	531
150	Genome-wide detection and characterization of positive selection in human populations. Nature, 2007, 449, 913-918.	27.8	1,788
151	Linkage Disequilibrium and Heritability of Copy-Number Polymorphisms within Duplicated Regions of the Human Genome. American Journal of Human Genetics, 2006, 79, 275-290.	6.2	283
152	Common deletion polymorphisms in the human genome. Nature Genetics, 2006, 38, 86-92.	21.4	656
153	New insights into the biological basis of genomic disorders. Nature Genetics, 2006, 38, 1363-1364.	21.4	12
154	DNA sequence of human chromosome 17 and analysis of rearrangement in the human lineage. Nature, 2006, 440, 1045-1049.	27.8	130
155	Copy number variation: New insights in genome diversity. Genome Research, 2006, 16, 949-961.	5.5	697
156	Identification of Transcriptional Regulatory Elements in Chemosensory Receptor Genes by Probabilistic Segmentation. Current Biology, 2005, 15, 347-352.	3.9	42
157	Comparing genomic expression patterns across species identifies shared transcriptional profile in aging. Nature Genetics, 2004, 36, 197-204.	21.4	434
158	Genes that act downstream of DAF-16 to influence the lifespan of Caenorhabditis elegans. Nature, 2003, 424, 277-283.	27.8	1,998
159	Thy-1 Is a Component Common to Multiple Populations of Synaptic Vesicles. Journal of Cell Biology, 1998, 140, 685-698.	5.2	51