Jay Shendure

List of Publications by Year
in descending order

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326	87,420	518	559
papers	citations	131 h-index	270 g-index

400	400	400	109650
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	A general framework for estimating the relative pathogenicity of human genetic variants. Nature Genetics, 2014, 46, 310-315.	9.4	5,167
2	Next-generation DNA sequencing. Nature Biotechnology, 2008, 26, 1135-1145.	9.4	3,609
3	CADD: predicting the deleteriousness of variants throughout the human genome. Nucleic Acids Research, 2019, 47, D886-D894.	6.5	2,360
4	The single-cell transcriptional landscape of mammalian organogenesis. Nature, 2019, 566, 496-502.	13.7	2,292
5	The contribution of de novo coding mutations to autism spectrum disorder. Nature, 2014, 515, 216-221.	13.7	2,188
6	Sporadic autism exomes reveal a highly interconnected protein network of de novo mutations. Nature, 2012, 485, 246-250.	13.7	1,960
7	The complete genome sequence of a Neanderthal from the Altai Mountains. Nature, 2014, 505, 43-49.	13.7	1,830
8	Exome sequencing identifies the cause of a mendelian disorder. Nature Genetics, 2010, 42, 30-35.	9.4	1,813
9	Targeted capture and massively parallel sequencing of 12 human exomes. Nature, 2009, 461, 272-276.	13.7	1,801
10	A High-Coverage Genome Sequence from an Archaic Denisovan Individual. Science, 2012, 338, 222-226.	6.0	1,695
11	Exome sequencing as a tool for Mendelian disease gene discovery. Nature Reviews Genetics, 2011, 12, 745-755.	7.7	1,484
12	Exome sequencing identifies MLL2 mutations as a cause of Kabuki syndrome. Nature Genetics, 2010, 42, 790-793.	9.4	1,238
13	Accurate Multiplex Polony Sequencing of an Evolved Bacterial Genome. Science, 2005, 309, 1728-1732.	6.0	1,189
14	Integrative genome analyses identify key somatic driver mutations of small-cell lung cancer. Nature Genetics, 2012, 44, 1104-1110.	9.4	1,186
15	Chromosome-scale scaffolding of de novo genome assemblies based on chromatin interactions. Nature Biotechnology, 2013, 31, 1119-1125.	9.4	1,141
16	Multiplex Targeted Sequencing Identifies Recurrently Mutated Genes in Autism Spectrum Disorders. Science, 2012, 338, 1619-1622.	6.0	1,133
17	Exome sequencing in sporadic autism spectrum disorders identifies severe de novo mutations. Nature Genetics, 2011, 43, 585-589.	9.4	1,080
18	Comprehensive single-cell transcriptional profiling of a multicellular organism. Science, 2017, 357, 661-667.	6.0	1,067

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19	Target-enrichment strategies for next-generation sequencing. Nature Methods, 2010, 7, 111-118.	9.0	1,052
20	Multiplex single-cell profiling of chromatin accessibility by combinatorial cellular indexing. Science, 2015, 348, 910-914.	6.0	1,045
21	Cell-free DNA Comprises an InÂVivo Nucleosome Footprint that Informs Its Tissues-Of-Origin. Cell, 2016, 164, 57-68.	13.5	1,039
22	Analysis of Genetic Inheritance in a Family Quartet by Whole-Genome Sequencing. Science, 2010, 328, 636-639.	6.0	979
23	Analysis of 6,515 exomes reveals the recent origin of most human protein-coding variants. Nature, 2013, 493, 216-220.	13.7	898
24	A three-dimensional model of the yeast genome. Nature, 2010, 465, 363-367.	13.7	894
25	Genome evolution in the allotetraploid frog Xenopus laevis. Nature, 2016, 538, 336-343.	13.7	849
26	DNA sequencing at 40: past, present and future. Nature, 2017, 550, 345-353.	13.7	729
27	Classification and characterization of microsatellite instability across 18 cancer types. Nature Medicine, 2016, 22, 1342-1350.	15.2	726
28	Joint profiling of chromatin accessibility and gene expression in thousands of single cells. Science, 2018, 361, 1380-1385.	6.0	683
29	Disruptive CHD8 Mutations Define a Subtype of Autism Early in Development. Cell, 2014, 158, 263-276.	13.5	637
30	De novo germline and postzygotic mutations in AKT3, PIK3R2 and PIK3CA cause a spectrum of related megalencephaly syndromes. Nature Genetics, 2012, 44, 934-940.	9.4	621
31	A Single-Cell Atlas of InÂVivo Mammalian Chromatin Accessibility. Cell, 2018, 174, 1309-1324.e18.	13.5	620
32	Diversity of Human Copy Number Variation and Multicopy Genes. Science, 2010, 330, 641-646.	6.0	609
33	Single-molecule sequencing and chromatin conformation capture enable de novo reference assembly of the domestic goat genome. Nature Genetics, 2017, 49, 643-650.	9.4	600
34	Targeted resequencing in epileptic encephalopathies identifies de novo mutations in CHD2 and SYNGAP1. Nature Genetics, 2013, 45, 825-830.	9.4	589
35	Refining analyses of copy number variation identifies specific genes associated with developmental delay. Nature Genetics, 2014, 46, 1063-1071.	9.4	583
36	Assemblathon 2: evaluating de novo methods of genome assembly in three vertebrate species. GigaScience, 2013, 2, 10.	3.3	582

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37	The 4D nucleome project. Nature, 2017, 549, 219-226.	13.7	579
38	The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities. American Journal of Human Genetics, 2015, 97, 199-215.	2.6	574
39	Substantial interindividual and limited intraindividual genomic diversity among tumors from men with metastatic prostate cancer. Nature Medicine, 2016, 22, 369-378.	15.2	572
40	Cicero Predicts cis-Regulatory DNA Interactions from Single-Cell Chromatin Accessibility Data. Molecular Cell, 2018, 71, 858-871.e8.	4.5	572
41	Whole-organism lineage tracing by combinatorial and cumulative genome editing. Science, 2016, 353, aaf7907.	6.0	570
42	Accurate classification of BRCA1 variants with saturation genome editing. Nature, 2018, 562, 217-222.	13.7	570
43	Needles in stacks of needles: finding disease-causal variants in a wealth of genomic data. Nature Reviews Genetics, 2011, 12, 628-640.	7.7	531
44	Fragment Length of Circulating Tumor DNA. PLoS Genetics, 2016, 12, e1006162.	1.5	502
45	Advanced sequencing technologies: methods and goals. Nature Reviews Genetics, 2004, 5, 335-344.	7.7	499
46	Rapid, low-input, low-bias construction of shotgun fragment libraries by high-density in vitro transposition. Genome Biology, 2010, 11, R119.	13.9	499
47	Simultaneous single-cell profiling of lineages and cell types in the vertebrate brain. Nature Biotechnology, 2018, 36, 442-450.	9.4	478
48	Massively parallel functional dissection of mammalian enhancers in vivo. Nature Biotechnology, 2012, 30, 265-270.	9.4	468
49	Massively multiplex single-cell Hi-C. Nature Methods, 2017, 14, 263-266.	9.0	441
50	A brief history of human disease genetics. Nature, 2020, 577, 179-189.	13.7	441
51	A human cell atlas of fetal gene expression. Science, 2020, 370, .	6.0	436
52	A de novo convergence of autism genetics and molecular neuroscience. Trends in Neurosciences, 2014, 37, 95-105.	4.2	410
53	Prediction of Susceptibility to First-Line Tuberculosis Drugs by DNA Sequencing. New England Journal of Medicine, 2018, 379, 1403-1415.	13.9	405
54	Multiplex amplification of large sets of human exons. Nature Methods, 2007, 4, 931-936.	9.0	392

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55	The million mutation project: A new approach to genetics in Caenorhabditis elegans. Genome Research, 2013, 23, 1749-1762.	2.4	382
56	A Genome-wide Framework for Mapping Gene Regulation via Cellular Genetic Screens. Cell, 2019, 176, 377-390.e19.	13.5	379
57	CADD-Spliceâ€"improving genome-wide variant effect prediction using deep learning-derived splice scores. Genome Medicine, 2021, 13, 31.	3.6	375
58	Long-read sequence assembly of the gorilla genome. Science, 2016, 352, aae0344.	6.0	368
59	Decoding long nanopore sequencing reads of natural DNA. Nature Biotechnology, 2014, 32, 829-833.	9.4	355
60	Noninvasive Whole-Genome Sequencing of a Human Fetus. Science Translational Medicine, 2012, 4, 137ra76.	5.8	348
61	Supervised classification enables rapid annotation of cell atlases. Nature Methods, 2019, 16, 983-986.	9.0	332
62	GRIN2A mutations cause epilepsy-aphasia spectrum disorders. Nature Genetics, 2013, 45, 1073-1076.	9.4	326
63	Somatic Mutations in Cerebral Cortical Malformations. New England Journal of Medicine, 2014, 371, 733-743.	13.9	326
64	The cis-regulatory dynamics of embryonic development at single-cell resolution. Nature, 2018, 555, 538-542.	13.7	323
65	Multiplex assessment of protein variant abundance by massively parallel sequencing. Nature Genetics, 2018, 50, 874-882.	9.4	323
66	High-resolution analysis of DNA regulatory elements by synthetic saturation mutagenesis. Nature Biotechnology, 2009, 27, 1173-1175.	9.4	322
67	TGFB2 mutations cause familial thoracic aortic aneurysms and dissections associated with mild systemic features of Marfan syndrome. Nature Genetics, 2012, 44, 916-921.	9.4	319
68	A germline homozygous mutation in the base-excision repair gene NTHL1 causes adenomatous polyposis and colorectal cancer. Nature Genetics, 2015, 47, 668-671.	9.4	311
69	Global survey of escape from X inactivation by RNA-sequencing in mouse. Genome Research, 2010, 20, 614-622.	2.4	309
70	High-resolution comparative analysis of great ape genomes. Science, 2018, 360, .	6.0	304
71	The haplotype-resolved genome and epigenome of the aneuploid HeLa cancer cell line. Nature, 2013, 500, 207-211.	13.7	302
72	Saturation editing of genomic regions by multiplex homology-directed repair. Nature, 2014, 513, 120-123.	13.7	301

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73	Single molecule molecular inversion probes for targeted, high-accuracy detection of low-frequency variation. Genome Research, 2013, 23, 843-854.	2.4	292
74	The beginning of the end for microarrays?. Nature Methods, 2008, 5, 585-587.	9.0	291
75	Transcriptome-wide miR-155 Binding Map Reveals Widespread Noncanonical MicroRNA Targeting. Molecular Cell, 2012, 48, 760-770.	4.5	290
76	PI3K/AKT pathway mutations cause a spectrum of brain malformations from megalencephaly to focal cortical dysplasia. Brain, 2015, 138, 1613-1628.	3.7	286
77	Poxviruses Deploy Genomic Accordions to Adapt Rapidly against Host Antiviral Defenses. Cell, 2012, 150, 831-841.	13.5	281
78	The expanding scope of DNA sequencing. Nature Biotechnology, 2012, 30, 1084-1094.	9.4	280
79	Regional Isolation Drives Bacterial Diversification within Cystic Fibrosis Lungs. Cell Host and Microbe, 2015, 18, 307-319.	5.1	278
80	Variant Interpretation: Functional Assays to the Rescue. American Journal of Human Genetics, 2017, 101, 315-325.	2.6	275
81	Massively Parallel Functional Analysis of BRCA1 RING Domain Variants. Genetics, 2015, 200, 413-422.	1.2	272
82	A human cell atlas of fetal chromatin accessibility. Science, 2020, 370, .	6.0	265
83	Evidence for compensatory upregulation of expressed X-linked genes in mammals, Caenorhabditis elegans and Drosophila melanogaster. Nature Genetics, 2011, 43, 1179-1185.	9.4	260
84	Exome Sequencing Identifies SMAD3 Mutations as a Cause of Familial Thoracic Aortic Aneurysm and Dissection With Intracranial and Other Arterial Aneurysms. Circulation Research, 2011, 109, 680-686.	2.0	258
85	Genomic surveillance reveals multiple introductions of SARS-CoV-2 into Northern California. Science, 2020, 369, 582-587.	6.0	253
86	A Molecular Pathway Including Id2, Tbx5, and Nkx2-5 Required for Cardiac Conduction System Development. Cell, 2007, 129, 1365-1376.	13.5	248
87	A systematic comparison reveals substantial differences in chromosomal versus episomal encoding of enhancer activity. Genome Research, 2017, 27, 38-52.	2.4	244
88	De novo mutations in the actin genes ACTB and ACTG1 cause Baraitser-Winter syndrome. Nature Genetics, 2012, 44, 440-444.	9.4	237
89	Mammalian target of rapamycin pathway mutations cause hemimegalencephaly and focal cortical dysplasia. Annals of Neurology, 2015, 77, 720-725.	2.8	235
90	Association ofMTORMutations With Developmental Brain Disorders, Including Megalencephaly, Focal Cortical Dysplasia, and Pigmentary Mosaicism. JAMA Neurology, 2016, 73, 836.	4.5	234

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91	Exome sequencing identifies a spectrum of mutation frequencies in advanced and lethal prostate cancers. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 17087-17092.	3.3	233
92	GABRA1 and STXBP1 : Novel genetic causes of Dravet syndrome. Neurology, 2014, 82, 1245-1253.	1.5	229
93	The lncRNA Firre anchors the inactive X chromosome to the nucleolus by binding CTCF and maintains H3K27me3 methylation. Genome Biology, 2015, 16, 52.	3.8	229
94	Towards a comprehensive catalogue of validated and target-linked human enhancers. Nature Reviews Genetics, 2020, 21, 292-310.	7.7	229
95	Massively parallel decoding of mammalian regulatory sequences supports a flexible organizational model. Nature Genetics, 2013, 45, 1021-1028.	9.4	226
96	Escape from X Inactivation Varies in Mouse Tissues. PLoS Genetics, 2015, 11, e1005079.	1.5	224
97	Learning the Sequence Determinants of Alternative Splicing from Millions of Random Sequences. Cell, 2015, 163, 698-711.	13.5	223
98	Complex MSH2 and MSH6 mutations in hypermutated microsatellite unstable advanced prostate cancer. Nature Communications, 2014, 5, 4988.	5.8	219
99	Genome-Scale Identification of Resistance Functions in Pseudomonas aeruginosa Using Tn-seq. MBio, 2011, 2, e00315-10.	1.8	217
100	Cryptic transmission of SARS-CoV-2 in Washington state. Science, 2020, 370, 571-575.	6.0	217
101	Haplotype-resolved genome sequencing of a Gujarati Indian individual. Nature Biotechnology, 2011, 29, 59-63.	9.4	216
102	An siRNA-based functional genomics screen for theÂidentification of regulators of ciliogenesis and ciliopathyÂgenes. Nature Cell Biology, 2015, 17, 1074-1087.	4.6	215
103	Highly scalable generation of DNA methylation profiles in single cells. Nature Biotechnology, 2018, 36, 428-431.	9.4	215
104	Genomic Analyses from Non-invasive Prenatal Testing Reveal Genetic Associations, Patterns of Viral Infections, and Chinese Population History. Cell, 2018, 175, 347-359.e14.	13.5	213
105	Bipartite structure of the inactive mouse X chromosome. Genome Biology, 2015, 16, 152.	3.8	211
106	Estimating the human mutation rate using autozygosity in a founder population. Nature Genetics, 2012, 44, 1277-1281.	9.4	202
107	Recurrent Gain-of-Function Mutation in PRKG1 Causes Thoracic Aortic Aneurysms and Acute Aortic Dissections. American Journal of Human Genetics, 2013, 93, 398-404.	2.6	197
108	Identification of sexâ€“specific quantitative trait loci controlling alcohol preference in C57BL/6 mice. Nature Genetics, 1996, 13, 147-153.	9.4	196

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109	Massively multiplex chemical transcriptomics at single-cell resolution. Science, 2020, 367, 45-51.	6.0	196
110	Mutations in RSPH1 Cause Primary Ciliary Dyskinesia with a Unique Clinical and Ciliary Phenotype. American Journal of Respiratory and Critical Care Medicine, 2014, 189, 707-717.	2.5	191
111	A suppressor screen in Mecp2 mutant mice implicates cholesterol metabolism in Rett syndrome. Nature Genetics, 2013, 45, 1013-1020.	9.4	190
112	Haploinsufficiency of SF3B4, a Component of the Pre-mRNA Spliceosomal Complex, Causes Nager Syndrome. American Journal of Human Genetics, 2012, 90, 925-933.	2.6	188
113	Massively parallel exon capture and library-free resequencing across 16 genomes. Nature Methods, 2009, 6, 315-316.	9.0	186
114	Rapid 16S rRNA Next-Generation Sequencing of Polymicrobial Clinical Samples for Diagnosis of Complex Bacterial Infections. PLoS ONE, 2013, 8, e65226.	1.1	186
115	LOX Mutations Predispose to Thoracic Aortic Aneurysms and Dissections. Circulation Research, 2016, 118, 928-934.	2.0	180
116	Species-Level Deconvolution of Metagenome Assemblies with Hi-Câ€“Based Contact Probability Maps. G3: Genes, Genomes, Genetics, 2014, 4, 1339-1346.	0.8	177
117	Fine-scale chromatin interaction maps reveal the cis-regulatory landscape of human lincRNA genes. Nature Methods, 2015, 12, 71-78.	9.0	177
118	Quantitative Missense Variant Effect Prediction Using Large-Scale Mutagenesis Data. Cell Systems, 2018, 6, 116-124.e3.	2.9	176
119	Mutations in PIEZO2 Cause Gordon Syndrome, Marden-Walker Syndrome, and Distal Arthrogryposis Type 5. American Journal of Human Genetics, 2014, 94, 734-744.	2.6	171
120	On the design of CRISPR-based single-cell molecular screens. Nature Methods, 2018, 15, 271-274.	9.0	170
121	Haplotype-resolved whole-genome sequencing by contiguity-preserving transposition and combinatorial indexing. Nature Genetics, 2014, 46, 1343-1349.	9.4	168
122	A Year of Infection in the Intensive Care Unit: Prospective Whole Genome Sequencing of Bacterial Clinical Isolates Reveals Cryptic Transmissions and Novel Microbiota. PLoS Genetics, 2015, 11, e1005413.	1.5	165
123	Massively parallel sequencing and rare disease. Human Molecular Genetics, 2010, 19, R119-R124.	1.4	163
124	Whole-Exome Capture and Sequencing Identifies HEATR2 Mutation as a Cause of Primary Ciliary Dyskinesia. American Journal of Human Genetics, 2012, 91, 685-693.	2.6	163
125	Single-nucleotide evolutionary constraint scores highlight disease-causing mutations. Nature Methods, 2010, 7, 250-251.	9.0	162
126	Tagmentation-based whole-genome bisulfite sequencing. Nature Protocols, 2013, 8, 2022-2032.	5.5	161

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127	Spectrum of MLL2 (ALR) mutations in 110 cases of Kabuki syndrome. American Journal of Medical Genetics, Part A, 2011, 155, 1511-1516.	0.7	160
128	Single-cell lineage tracing of metastatic cancer reveals selection of hybrid EMT states. Cancer Cell, 2021, 39, 1150-1162.e9.	7.7	160
129	Fluorescent in situ sequencing on polymerase colonies. Analytical Biochemistry, 2003, 320, 55-65.	1.1	159
130	Activity-enhancing mutations in an E3 ubiquitin ligase identified by high-throughput mutagenesis. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, E1263-72.	3.3	158
131	Large-scale genomic sequencing of extraintestinal pathogenic Escherichia coli strains. Genome Research, 2015, 25, 119-128.	2.4	158
132	Haplotype-resolved genome sequencing: experimental methods and applications. Nature Reviews Genetics, 2015, 16, 344-358.	7.7	156
133	High-content CRISPR screening. Nature Reviews Methods Primers, 2022, 2, .	11.8	155
134	Massively parallel single-amino-acid mutagenesis. Nature Methods, 2015, 12, 203-206.	9.0	153
135	Computational discovery of sense-antisense transcription in the human and mouse genomes. Genome Biology, 2002, 3, research0044.1.	13.9	152
136	Saturation mutagenesis of twenty disease-associated regulatory elements at single base-pair resolution. Nature Communications, 2019, 10, 3583.	5.8	152
137	Identification by whole-genome resequencing of gene defect responsible for severe hypercholesterolemia. Human Molecular Genetics, 2010, 19, 4313-4318.	1.4	151
138	megaTALs: a rare-cleaving nuclease architecture for therapeutic genome engineering. Nucleic Acids Research, 2014, 42, 2591-2601.	6.5	151
139	In vitro, long-range sequence information for de novo genome assembly via transposase contiguity. Genome Research, 2014, 24, 2041-2049.	2.4	150
140	A pooled single-cell genetic screen identifies regulatory checkpoints in the continuum of the epithelial-to-mesenchymal transition. Nature Genetics, 2019, 51, 1389-1398.	9.4	150
141	Embryo-scale, single-cell spatial transcriptomics. Science, 2021, 373, 111-117.	6.0	149
142	Parallel, tag-directed assembly of locally derived short sequence reads. Nature Methods, 2010, 7, 119-122.	9.0	144
143	Predicting mRNA Abundance Directly from Genomic Sequence Using Deep Convolutional Neural Networks. Cell Reports, 2020, 31, 107663.	2.9	144
144	The origins, determinants, and consequences of human mutations. Science, 2015, 349, 1478-1483.	6.0	143

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145	Genomic Medicineâ€“Progress, Pitfalls, and Promise. Cell, 2019, 177, 45-57.	13.5	143
146	Digital genotyping and haplotyping with polymerase colonies. Proceedings of the National Academy of Sciences of the United States of America, 2003, 100, 5926-5931.	3.3	141
147	Selection analyses of insertional mutants using subgenic-resolution arrays. Nature Biotechnology, 2001, 19, 1060-1065.	9.4	140
148	Mechanisms of Interplay between Transcription Factors and the 3D Genome. Molecular Cell, 2019, 76, 306-319.	4.5	140
149	Exome Sequencing Identifies Mutations in CCDC114 as a Cause of Primary Ciliary Dyskinesia. American Journal of Human Genetics, 2013, 92, 99-106.	2.6	138
150	MIPgen: optimized modeling and design of molecular inversion probes for targeted resequencing. Bioinformatics, 2014, 30, 2670-2672.	1.8	138
151	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. American Journal of Human Genetics, 2017, 101, 768-788.	2.6	136
152	Mutations in SPAG1 Cause Primary Ciliary Dyskinesia Associated with Defective Outer and Inner Dynein Arms. American Journal of Human Genetics, 2013, 93, 711-720.	2.6	135
153	Massively parallel profiling and predictive modeling of the outcomes of CRISPR/Cas9-mediated double-strand break repair. Nucleic Acids Research, 2019, 47, 7989-8003.	6.5	135
154	PIK3CA-associated developmental disorders exhibit distinct classes of mutations with variable expression and tissue distribution. JCI Insight, 2016, 1, .	2.3	134
155	CRISPR/Cas9-Mediated Scanning for Regulatory Elements Required for HPRT1 Expression via Thousands of Large, Programmed Genomic Deletions. American Journal of Human Genetics, 2017, 101, 192-205.	2.6	133
156	MaveDB: an open-source platform to distribute and interpret data from multiplexed assays of variant effect. Genome Biology, 2019, 20, 223.	3.8	130
157	RNF213 Rare Variants in an Ethnically Diverse Population With Moyamoya Disease. Stroke, 2014, 45, 3200-3207.	1.0	129
158	De Novo Mutations in NALCN Cause a Syndrome Characterized by Congenital Contractures of the Limbs and Face, Hypotonia, and Developmental Delay. American Journal of Human Genetics, 2015, 96, 462-473.	2.6	124
159	Copy-Number Variation and False Positive Prenatal Aneuploidy Screening Results. New England Journal of Medicine, 2015, 372, 1639-1645.	13.9	118
160	Precise genomic deletions using paired prime editing. Nature Biotechnology, 2022, 40, 218-226.	9.4	117
161	The power of multiplexed functional analysis of genetic variants. Nature Protocols, 2016, 11, 1782-1787.	5.5	115
162	Methods for Genomic Partitioning. Annual Review of Genomics and Human Genetics, 2009, 10, 263-284.	2.5	114

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163	Ultra-low-input, tagmentation-based whole-genome bisulfite sequencing. Genome Research, 2012, 22, 1139-1143.	2.4	114
164	Mosaicism of the UDP-Galactose Transporter SLC35A2 Causes a Congenital Disorder of Glycosylation. American Journal of Human Genetics, 2013, 92, 632-636.	2.6	114
165	Trans genomic capture and sequencing of primate exomes reveals new targets of positive selection. Genome Research, 2011, 21, 1686-1694.	2.4	111
166	A systematic evaluation of the design and context dependencies of massively parallel reporter assays. Nature Methods, 2020, 17, 1083-1091.	9.0	111
167	De novo TBR1 mutations in sporadic autism disrupt protein functions. Nature Communications, 2014, 5, 4954.	5.8	109
168	Informed consent for whole genome sequencing: A qualitative analysis of participant expectations and perceptions of risks, benefits, and harms. American Journal of Medical Genetics, Part A, 2012, 158A, 1310-1319.	0.7	106
169	Mapping 3D genome architecture through in situ DNase Hi-C. Nature Protocols, 2016, 11, 2104-2121.	5.5	106
170	Mutations in CSPP1 Cause Primary Cilia Abnormalities and Joubert Syndrome with or without Jeune Asphyxiating Thoracic Dystrophy. American Journal of Human Genetics, 2014, 94, 62-72.	2.6	104
171	High-throughput determination of RNA structure by proximity ligation. Nature Biotechnology, 2015, 33, 980-984.	9.4	104
172	Mammalian X Upregulation Is Associated with Enhanced Transcription Initiation, RNA Half-Life, and MOF-Mediated H4K16 Acetylation. Developmental Cell, 2013, 25, 55-68.	3.1	103
173	Single-cell ATAC-Seq in human pancreatic islets and deep learning upscaling of rare cells reveals cell-specific type 2 diabetes regulatory signatures. Molecular Metabolism, 2020, 32, 109-121.	3.0	103
174	Mutation discovery in mice by whole exome sequencing. Genome Biology, 2011, 12, R86.	13.9	102
175	A Multiplex Homology-Directed DNA Repair Assay Reveals the Impact of More Than 1,000 BRCA1 Missense Substitution Variants on Protein Function. American Journal of Human Genetics, 2018, 103, 498-508.	2.6	99
176	Next generation sequence analysis for mitochondrial disorders. Genome Medicine, 2009, 1, 100.	3.6	98
177	Long-range polony haplotyping of individual human chromosome molecules. Nature Genetics, 2006, 38, 382-387.	9.4	97
178	Early Detection of Covid-19 through a Citywide Pandemic Surveillance Platform. New England Journal of Medicine, 2020, 383, 185-187.	13.9	97
179	Characterization of Apparently Balanced Chromosomal Rearrangements from the Developmental Genome Anatomy Project. American Journal of Human Genetics, 2008, 82, 712-722.	2.6	95
180	Single Molecule Profiling of Alternative Pre-mRNA Splicing. Science, 2003, 301, 836-838.	6.0	93

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181	FlashFry: a fast and flexible tool for large-scale CRISPR target design. BMC Biology, 2018, 16, 74.	1.7	93
182	MAT2A Mutations Predispose Individuals to Thoracic Aortic Aneurysms. American Journal of Human Genetics, 2015, 96, 170-177.	2.6	92
183	Expanding the clinical and genetic heterogeneity of hereditary disorders of connective tissue. Human Genetics, 2016, 135, 525-540.	1.8	89
184	Sci-fate characterizes the dynamics of gene expression in single cells. Nature Biotechnology, 2020, 38, 980-988.	9.4	89
185	Exome-wide DNA capture and next generation sequencing in domestic and wild species. BMC Genomics, 2011, 12, 347.	1.2	88
186	FOXE3 mutations predispose to thoracic aortic aneurysms and dissections. Journal of Clinical Investigation, 2016, 126, 948-961.	3.9	84
187	Novel approach to genetic analysis and results in 3000 hemophilia patients enrolled in the My Life, Our Future initiative. Blood Advances, 2017, 1, 824-834.	2.5	83
188	Condensin-Dependent Chromatin Compaction Represses Transcription Globally during Quiescence. Molecular Cell, 2019, 73, 533-546.e4.	4.5	83
189	High-Throughput Single-Cell Sequencing with Linear Amplification. Molecular Cell, 2019, 76, 676-690.e10.	4.5	82
190	Characterisation of mutations of the phosphoinositide-3-kinase regulatory subunit, PIK3R2, in perisylvian polymicrogyria: a next-generation sequencing study. Lancet Neurology, The, 2015, 14, 1182-1195.	4.9	74
191	Discovering functional transcription-factor combinations in the human cell cycle. Genome Research, 2005, 15, 848-855.	2.4	73
192	Systematic reconstruction of cellular trajectories across mouse embryogenesis. Nature Genetics, 2022, 54, 328-341.	9.4	73
193	Primate evolution of the recombination regulator PRDM9. Nature Communications, 2014, 5, 4370.	5.8	72
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