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	Precise genomic deletions using paired prime editing. Nature Biotechnology, 2022, 40, 218-226.	9.4	117
2	High-content CRISPR screening. Nature Reviews Methods Primers, 2022, 2, .	11.8	155
3	Systematic reconstruction of cellular trajectories across mouse embryogenesis. Nature Genetics, 2022, 54, 328-341.	9.4	73
4	The Seattle Flu Study: when regulations hinder pandemic surveillance. Nature Medicine, 2022, 28, 7-8.	15.2	3
5	Simultaneous brain cell type and lineage determined by scRNA-seq reveals stereotyped cortical development. Cell Systems, 2022, 13, 438-453.e5.	2.9	2
6	A hybrid open-top light-sheet microscope for versatile multi-scale imaging of cleared tissues. Nature Methods, 2022, 19, 613-619.	9.0	54
7	A time-resolved, multi-symbol molecular recorder via sequential genome editing. Nature, 2022, 608, 98-107.	13.7	59
8	Results of genetic analysis of 11â€‰341 participants enrolled in the My Life, Our Future hemophilia genotyping initiative in the United States. Journal of Thrombosis and Haemostasis, 2022, 20, 2022-2034.	1.9	10
9	Incidence of Medically Attended Acute Respiratory Illnesses Due to Respiratory Viruses Across the Life Course During the 2018/19 Influenza Season. Clinical Infectious Diseases, 2021, 73, 802-807.	2.9	8
10	CADD-Spliceâ€”improving genome-wide variant effect prediction using deep learning-derived splice scores. Genome Medicine, 2021, 13, 31.	3.6	375
11	Comprehensive characterization of tissue-specific chromatin accessibility in L2 Caenorhabditis elegans nematodes. Genome Research, 2021, 31, 1952-1969.	2.4	8
12	Comparable Specimen Collection from Both Ends of At-Home Midturbinate Swabs. Journal of Clinical Microbiology, 2021, 59, .	1.8	2
13	Genome-wide strand asymmetry in massively parallel reporter activity favors genic strands. Genome Research, 2021, 31, 866-876.	2.4	1
14	A continuous model of early mammalian development. Nature, 2021, 593, 200-201.	13.7	0
15	Viral genomes reveal patterns of the SARS-CoV-2 outbreak in Washington State. Science Translational Medicine, 2021, 13, .	5.8	58
16	SwabExpress: An End-to-End Protocol for Extraction-Free COVID-19 Testing. Clinical Chemistry, 2021, 68, 143-152.	1.5	24
17	Embryo-scale, single-cell spatial transcriptomics. Science, 2021, 373, 111-117.	6.0	149
18	The landscape of alternative polyadenylation in single cells of the developing mouse embryo. Nature Communications, 2021, 12, 5101.	5.8	33

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19	Benchmarked approaches for reconstruction of inÂvitro cell lineages and in silico models of C. elegans and M. musculus developmental trees. Cell Systems, 2021, 12, 810-826.e4.	2.9	36
20	Single-cell lineage tracing of metastatic cancer reveals selection of hybrid EMT states. Cancer Cell, 2021, 39, 1150-1162.e9.	7.7	160
21	Single-cell landscape of nuclear configuration and gene expression during stem cell differentiation and X inactivation. Genome Biology, 2021, 22, 279.	3.8	11
22	The glucose-sensing transcription factor MLX balances metabolism and stress to suppress apoptosis and maintain spermatogenesis. PLoS Biology, 2021, 19, e3001085.	2.6	7
23	Adaptations in Hippo-Yap signaling and myofibroblast fate underlie scar-free ear appendage wound healing in spiny mice. Developmental Cell, 2021, 56, 2722-2740.e6.	3.1	31
24	Sci-Hi-C: A single-cell Hi-C method for mapping 3D genome organization in large number of single cells. Methods, 2020, 170, 61-68.	1.9	53
25	A brief history of human disease genetics. Nature, 2020, 577, 179-189.	13.7	441
26	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
27	Massively multiplex chemical transcriptomics at single-cell resolution. Science, 2020, 367, 45-51.	6.0	196
28	Capturing cell type-specific chromatin compartment patterns by applying topic modeling to single-cell Hi-C data. PLoS Computational Biology, 2020, 16, e1008173.	1.5	59
29	A systematic evaluation of the design and context dependencies of massively parallel reporter assays. Nature Methods, 2020, 17, 1083-1091.	9.0	111
30	The Seattle Flu Study: a multiarm community-based prospective study protocol for assessing influenza prevalence, transmission and genomic epidemiology. BMJ Open, 2020, 10, e037295.	0.8	25
31	A human cell atlas of fetal chromatin accessibility. Science, 2020, 370, .	6.0	265
32	A human cell atlas of fetal gene expression. Science, 2020, 370, .	6.0	436
33	Cryptic transmission of SARS-CoV-2 in Washington state. Science, 2020, 370, 571-575.	6.0	217
34	Trans- and cis-acting effects of Firre on epigenetic features of the inactive X chromosome. Nature Communications, 2020, 11, 6053.	5.8	33
35	Predicting mRNA Abundance Directly from Genomic Sequence Using Deep Convolutional Neural Networks. Cell Reports, 2020, 31, 107663.	2.9	144
36	Elevated exopolysaccharide levels in Pseudomonas aeruginosa flagellar mutants have implications for biofilm growth and chronic infections. PLoS Genetics, 2020, 16, e1008848.	1.5	52

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37	Genomic surveillance reveals multiple introductions of SARS-CoV-2 into Northern California. Science, 2020, 369, 582-587.	6.0	253
38	lentiMPRA and MPRAflow for high-throughput functional characterization of gene regulatory elements. Nature Protocols, 2020, 15, 2387-2412.	5.5	65
39	Multimodal singleâ€cell analysis reveals distinct radioresistant stemâ€like and progenitor cell populations in murine glioma. Glia, 2020, 68, 2486-2502.	2.5	8
40	Towards a comprehensive catalogue of validated and target-linked human enhancers. Nature Reviews Genetics, 2020, 21, 292-310.	7.7	229
41	Early Detection of Covid-19 through a Citywide Pandemic Surveillance Platform. New England Journal of Medicine, 2020, 383, 185-187.	13.9	97
42	Sci-fate characterizes the dynamics of gene expression in single cells. Nature Biotechnology, 2020, 38, 980-988.	9.4	89
43	Before the Flood. Clinical Infectious Diseases, 2020, 71, 2513-2515.	2.9	0
44	Suppressor mutations in Mecp2-null mice implicate the DNA damage response in Rett syndrome pathology. Genome Research, 2020, 30, 540-552.	2.4	13
45	Unsupervised manifold alignment for single-cell multi-omics data. , 2020, 2020, 1-10.		33
46	Saturation mutagenesis of twenty disease-associated regulatory elements at single base-pair resolution. Nature Communications, 2019, 10, 3583.	5.8	152
47	Chromatin compartment dynamics in a haploinsufficient model of cardiac laminopathy. Journal of Cell Biology, 2019, 218, 2919-2944.	2.3	46
48	The Rhododendron Genome and Chromosomal Organization Provide Insight into Shared Whole-Genome Duplications across the Heath Family (Ericaceae). Genome Biology and Evolution, 2019, 11, 3353-3371.	1.1	47
49	MaveDB: an open-source platform to distribute and interpret data from multiplexed assays of variant effect. Genome Biology, 2019, 20, 223.	3.8	130
50	High-Throughput Single-Cell Sequencing with Linear Amplification. Molecular Cell, 2019, 76, 676-690.e10.	4.5	82
51	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
52	Supervised classification enables rapid annotation of cell atlases. Nature Methods, 2019, 16, 983-986.	9.0	332
53	Mechanisms of Interplay between Transcription Factors and the 3D Genome. Molecular Cell, 2019, 76, 306-319.	4.5	140
54	Expanding the single-cell genomics toolkit. Nature Genetics, 2019, 51, 931-932.	9.4	3

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55	Functional testing of thousands of osteoarthritis-associated variants for regulatory activity. Nature Communications, 2019, 10, 2434.	5.8	71
56	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
57	Integration of multiple epigenomic marks improves prediction of variant impact in saturation mutagenesis reporter assay. Human Mutation, 2019, 40, 1280-1291.	1.1	46
58	Genomic Medicineâ€“Progress, Pitfalls, and Promise. Cell, 2019, 177, 45-57.	13.5	143
59	Mutations in the transloconâ€associated protein complex subunit SSR3 cause a novel congenital disorder of glycosylation. Journal of Inherited Metabolic Disease, 2019, 42, 993-997.	1.7	18
60	High Sensitivity Profiling of Chromatin Structure by MNase-SSP. Cell Reports, 2019, 26, 2465-2476.e4.	2.9	31
61	The single-cell transcriptional landscape of mammalian organogenesis. Nature, 2019, 566, 496-502.	13.7	2,292
62	Voices in methods development. Nature Methods, 2019, 16, 945-951.	9.0	5
63	LB21. The Seattle Flu Study: A Community-Based Study of Influenza. Open Forum Infectious Diseases, 2019, 6, S1002-S1002.	0.4	8
64	Recommendations for the collection and use of multiplexed functional data for clinical variant interpretation. Genome Medicine, 2019, 11, 85.	3.6	47
65	Condensin-Dependent Chromatin Compaction Represses Transcription Globally during Quiescence. Molecular Cell, 2019, 73, 533-546.e4.	4.5	83
66	A Genome-wide Framework for Mapping Gene Regulation via Cellular Genetic Screens. Cell, 2019, 176, 377-390.e19.	13.5	379
67	CADD: predicting the deleteriousness of variants throughout the human genome. Nucleic Acids Research, 2019, 47, D886-D894.	6.5	2,360
68	A combination of transcription factors mediates inducible interchromosomal contacts. ELife, 2019, 8, .	2.8	16
69	Highly scalable generation of DNA methylation profiles in single cells. Nature Biotechnology, 2018, 36, 428-431.	9.4	215
70	On the design of CRISPR-based single-cell molecular screens. Nature Methods, 2018, 15, 271-274.	9.0	170
71	Using DNase Hi-C techniques to map global and local three-dimensional genome architecture at high resolution. Methods, 2018, 142, 59-73.	1.9	20
72	Quantitative Missense Variant Effect Prediction Using Large-Scale Mutagenesis Data. Cell Systems, 2018, 6, 116-124.e3.	2.9	176

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73	Identifying Novel Enhancer Elements with CRISPR-Based Screens. ACS Chemical Biology, 2018, 13, 326-332.	1.6	25
74	The cis-regulatory dynamics of embryonic development at single-cell resolution. Nature, 2018, 555, 538-542.	13.7	323
75	Simultaneous single-cell profiling of lineages and cell types in the vertebrate brain. Nature Biotechnology, 2018, 36, 442-450.	9.4	478
76	Identification of a novel interspecific hybrid yeast from a metagenomic spontaneously inoculated beer sample using Hiâ€€. Yeast, 2018, 35, 71-84.	0.8	31
77	Tagmentation-Based Library Preparation for Low DNA Input Whole Genome Bisulfite Sequencing. Methods in Molecular Biology, 2018, 1708, 105-122.	0.4	10
78	New insights into structural features and optimal detection of circulating tumor DNA determined by single-strand DNA analysis. Npj Genomic Medicine, 2018, 3, 31.	1.7	71
79	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
80	Prediction of Susceptibility to First-Line Tuberculosis Drugs by DNA Sequencing. New England Journal of Medicine, 2018, 379, 1403-1415.	13.9	405
81	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
82	Accurate classification of BRCA1 variants with saturation genome editing. Nature, 2018, 562, 217-222.	13.7	570
83	Joint profiling of chromatin accessibility and gene expression in thousands of single cells. Science, 2018, 361, 1380-1385.	6.0	683
84	Multiplex assessment of protein variant abundance by massively parallel sequencing. Nature Genetics, 2018, 50, 874-882.	9.4	323
85	A Single-Cell Atlas of InÂVivo Mammalian Chromatin Accessibility. Cell, 2018, 174, 1309-1324.e18.	13.5	620
86	Cicero Predicts cis-Regulatory DNA Interactions from Single-Cell Chromatin Accessibility Data. Molecular Cell, 2018, 71, 858-871.e8.	4.5	572
87	Functional characterization of enhancer evolution in the primate lineage. Genome Biology, 2018, 19, 99.	3.8	38
88	FlashFry: a fast and flexible tool for large-scale CRISPR target design. BMC Biology, 2018, 16, 74.	1.7	93
89	DPAGT1 Deficiency with Encephalopathy (DPAGT1-CDG): Clinical and Genetic Description of 11 New Patients. JIMD Reports, 2018, 44, 85-92.	0.7	16
90	High-resolution comparative analysis of great ape genomes. Science, 2018, 360, .	6.0	304

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91	Massively multiplex single-cell Hi-C. Nature Methods, 2017, 14, 263-266.	9.0	441
92	BRCA Testing by Single-Molecule Molecular Inversion Probes. Clinical Chemistry, 2017, 63, 503-512.	1.5	46
93	Activation of a cryptic splice site in the mitochondrial elongation factor GFM1 causes combined OXPHOS deficiency. Mitochondrion, 2017, 34, 84-90.	1.6	21
94	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
95	The State of Whole-Genome Sequencing. , 2017, , 45-62.		2
96	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
97	DNA sequencing at 40: past, present and future. Nature, 2017, 550, 345-353.	13.7	729
98	Variant Interpretation: Functional Assays to the Rescue. American Journal of Human Genetics, 2017, 101, 315-325.	2.6	275
99	The 4D nucleome project. Nature, 2017, 549, 219-226.	13.7	579
100	Encephalopathy caused by novel mutations in the CMPâ€sialic acid transporter, SLC35A1. American Journal of Medical Genetics, Part A, 2017, 173, 2906-2911.	0.7	26
101	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
102	Comprehensive single-cell transcriptional profiling of a multicellular organism. Science, 2017, 357, 661-667.	6.0	1,067
103	Haplotype phasing of whole human genomes using bead-based barcode partitioning in a single tube. Nature Biotechnology, 2017, 35, 852-857.	9.4	42
104	Quantification of differential gene expression by multiplexed targeted resequencing of cDNA. Nature Communications, 2017, 8, 15190.	5.8	19
105	A systematic comparison reveals substantial differences in chromosomal versus episomal encoding of enhancer activity. Genome Research, 2017, 27, 38-52.	2.4	244
106	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
107	Targeted Capture and High-Throughput Sequencing Using Molecular Inversion Probes (MIPs). Methods in Molecular Biology, 2017, 1492, 95-106.	0.4	17
108	The dynamic three-dimensional organization of the diploid yeast genome. ELife, 2017, 6, .	2.8	57

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109	Fragment Length of Circulating Tumor DNA. PLoS Genetics, 2016, 12, e1006162.	1.5	502
110	Recurrent somatic loss of <scp>TNFRSF14</scp> in classical Hodgkin lymphoma. Genes Chromosomes and Cancer, 2016, 55, 278-287.	1.5	23
111	ALG1-CDG: Clinical and Molecular Characterization of 39 Unreported Patients. Human Mutation, 2016, 37, 653-660.	1.1	40
112	Genome sequencing in a case of Niemannâ€“Pick type C. Journal of Physical Education and Sports Management, 2016, 2, a001222.	0.5	10
113	Expanding the clinical and genetic heterogeneity of hereditary disorders of connective tissue. Human Genetics, 2016, 135, 525-540.	1.8	89
114	Long-read sequence assembly of the gorilla genome. Science, 2016, 352, aae0344.	6.0	368
115	Whole-organism lineage tracing by combinatorial and cumulative genome editing. Science, 2016, 353, aaf7907.	6.0	570
116	LOX Mutations Predispose to Thoracic Aortic Aneurysms and Dissections. Circulation Research, 2016, 118, 928-934.	2.0	180
117	Association ofMTORMutations With Developmental Brain Disorders, Including Megalencephaly, Focal Cortical Dysplasia, and Pigmentary Mosaicism. JAMA Neurology, 2016, 73, 836.	4.5	234
118	Mapping 3D genome architecture through in situ DNase Hi-C. Nature Protocols, 2016, 11, 2104-2121.	5.5	106
119	Classification and characterization of microsatellite instability across 18 cancer types. Nature Medicine, 2016, 22, 1342-1350.	15.2	726
120	The power of multiplexed functional analysis of genetic variants. Nature Protocols, 2016, 11, 1782-1787.	5.5	115
121	A deep dive into genetic variation. Nature, 2016, 536, 277-278.	13.7	8
122	Genome evolution in the allotetraploid frog Xenopus laevis. Nature, 2016, 538, 336-343.	13.7	849
123	Novel mutations in the genes <scp>TGM</scp>1 and <scp>ALOXE</scp>3 underlying autosomal recessive congenital ichthyosis. International Journal of Dermatology, 2016, 55, 524-530.	0.5	6
124	Massively Parallel Genetics. Genetics, 2016, 203, 617-619.	1.2	24
125	Cilia gene mutations cause atrioventricular septal defects by multiple mechanisms. Human Molecular Genetics, 2016, 25, ddw155.	1.4	37
126	Mitral regurgitation as a phenotypic manifestation of nonphotosensitive trichothiodystrophy due to a splice variant in MPLKIP. BMC Medical Genetics, 2016, 17, 13.	2.1	5

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127	Rare variant associations with waist-to-hip ratio in European-American and African-American women from the NHLBI-Exome Sequencing Project. European Journal of Human Genetics, 2016, 24, 1181-1187.	1.4	5
128	Cell-free DNA Comprises an InÂVivo Nucleosome Footprint that Informs Its Tissues-Of-Origin. Cell, 2016, 164, 57-68.	13.5	1,039
129	Accelerating matchmaking of novel dysmorphology syndromes through clinical and genomic characterization of a large cohort. Genetics in Medicine, 2016, 18, 686-695.	1.1	55
130	Multiplex pairwise assembly of array-derived DNA oligonucleotides. Nucleic Acids Research, 2016, 44, e43-e43.	6.5	48
131	Understanding Spatial Genome Organization: Methods and Insights. Genomics, Proteomics and Bioinformatics, 2016, 14, 7-20.	3.0	54
132	Substantial interindividual and limited intraindividual genomic diversity among tumors from men with metastatic prostate cancer. Nature Medicine, 2016, 22, 369-378.	15.2	572
133	Expansion of the spectrum of ITGB6-related disorders to adolescent alopecia, dentogingival abnormalities and intellectual disability. European Journal of Human Genetics, 2016, 24, 1223-1227.	1.4	20
134	Large-scale targeted sequencing comparison highlights extreme genetic heterogeneity in nephronophthisis-related ciliopathies. Journal of Medical Genetics, 2016, 53, 208-214.	1.5	39
135	PIK3CA-associated developmental disorders exhibit distinct classes of mutations with variable expression and tissue distribution. JCI Insight, 2016, 1, .	2.3	134
136	FOXE3 mutations predispose to thoracic aortic aneurysms and dissections. Journal of Clinical Investigation, 2016, 126, 948-961.	3.9	84
137	Novel Approach to and Results of Genetic Analysis of 3000 Hemophilia Patients Enrolled in the MyLifeOurFuture Initiative. Blood, 2016, 128, 205-205.	0.6	1
138	Complex Minigene Library Vaccination for Discovery of Pre-Erythrocytic Plasmodium T Cell Antigens. PLoS ONE, 2016, 11, e0153449.	1.1	7
139	Identification of genes escaping X inactivation by allelic expression analysis in a novel hybrid mouse model. Data in Brief, 2015, 5, 761-769.	0.5	10
140	Bipartite structure of the inactive mouse X chromosome. Genome Biology, 2015, 16, 152.	3.8	211
141	KIAA0586is Mutated in Joubert Syndrome. Human Mutation, 2015, 36, 831-835.	1.1	62
142	Expanding the Molecular and Clinical Phenotype of SSR4-CDG. Human Mutation, 2015, 36, 1048-1051.	1.1	22
143	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
144	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

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145	PI3K/AKT pathway mutations cause a spectrum of brain malformations from megalencephaly to focal cortical dysplasia. Brain, 2015, 138, 1613-1628.	3.7	286
146	Whole genome prediction for preimplantation genetic diagnosis. Genome Medicine, 2015, 7, 35.	3.6	29
147	An essential cell cycle regulation gene causes hybrid inviability in Drosophila. Science, 2015, 350, 1552-1555.	6.0	59
148	Massively parallel single-amino-acid mutagenesis. Nature Methods, 2015, 12, 203-206.	9.0	153
149	Exome Sequencing in Suspected Monogenic Dyslipidemias. Circulation: Cardiovascular Genetics, 2015, 8, 343-350.	5.1	45
150	MAT2A Mutations Predispose Individuals to Thoracic Aortic Aneurysms. American Journal of Human Genetics, 2015, 96, 170-177.	2.6	92
151	Mammalian target of rapamycin pathway mutations cause hemimegalencephaly and focal cortical dysplasia. Annals of Neurology, 2015, 77, 720-725.	2.8	235
152	MIPSTR: a method for multiplex genotyping of germline and somatic STR variation across many individuals. Genome Research, 2015, 25, 750-761.	2.4	37
153	Mutation of ATF6 causes autosomal recessive achromatopsia. Human Genetics, 2015, 134, 941-950.	1.8	69
154	Running spell-check to identify regulatory variants. Nature Genetics, 2015, 47, 853-855.	9.4	5
155	High-throughput determination of RNA structure by proximity ligation. Nature Biotechnology, 2015, 33, 980-984.	9.4	104
156	Rare A2ML1 variants confer susceptibility to otitis media. Nature Genetics, 2015, 47, 917-920.	9.4	38
157	The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities. American Journal of Human Genetics, 2015, 97, 199-215.	2.6	574
158	A homozygous missense variant in type I keratin KRT25 causes autosomal recessive woolly hair. Journal of Medical Genetics, 2015, 52, 676-680.	1.5	23
159	Accurate identification of centromere locations in yeast genomes using Hi-C. Nucleic Acids Research, 2015, 43, 5331-5339.	6.5	61
160	Massively Parallel Functional Analysis of BRCA1 RING Domain Variants. Genetics, 2015, 200, 413-422.	1.2	272
161	An siRNA-based functional genomics screen for theÂidentification of regulators of ciliogenesis and ciliopathyÂgenes. Nature Cell Biology, 2015, 17, 1074-1087.	4.6	215
162	Escape from X Inactivation Varies in Mouse Tissues. PLoS Genetics, 2015, 11, e1005079.	1.5	224

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163	Exome sequencing reveals pathogenic mutations in 91 strains of mice with Mendelian disorders. Genome Research, 2015, 25, 948-957.	2.4	54
164	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
165	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
166	Multiplex single-cell profiling of chromatin accessibility by combinatorial cellular indexing. Science, 2015, 348, 910-914.	6.0	1,045
167	Haplotype-resolved genome sequencing: experimental methods and applications. Nature Reviews Genetics, 2015, 16, 344-358.	7.7	156
168	Autosomal-Dominant Multiple Pterygium Syndrome Is Caused by Mutations in MYH3. American Journal of Human Genetics, 2015, 96, 841-849.	2.6	55
169	Copy-Number Variation and False Positive Prenatal Aneuploidy Screening Results. New England Journal of Medicine, 2015, 372, 1639-1645.	13.9	118
170	The origins, determinants, and consequences of human mutations. Science, 2015, 349, 1478-1483.	6.0	143
171	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
172	Learning the Sequence Determinants of Alternative Splicing from Millions of Random Sequences. Cell, 2015, 163, 698-711.	13.5	223
173	Decoding Long Nanopore Reads of Bacteriophage Phi X 174. Biophysical Journal, 2015, 108, 630a.	0.2	0
174	Experimental Evolution Identifies Vaccinia Virus Mutations in A24R and A35R That Antagonize the Protein Kinase R Pathway and Accompany Collapse of an Extragenic Gene Amplification. Journal of Virology, 2015, 89, 9986-9997.	1.5	28
175	Regional Isolation Drives Bacterial Diversification within Cystic Fibrosis Lungs. Cell Host and Microbe, 2015, 18, 307-319.	5.1	278
176	Challenges and solutions for gene identification in the presence of familial locus heterogeneity. European Journal of Human Genetics, 2015, 23, 1207-1215.	1.4	35
177	Fine-scale chromatin interaction maps reveal the cis-regulatory landscape of human lincRNA genes. Nature Methods, 2015, 12, 71-78.	9.0	177
178	Large-scale genomic sequencing of extraintestinal pathogenic Escherichia coli strains. Genome Research, 2015, 25, 119-128.	2.4	158
179	Whole-Genome Sequencing of Individuals from a Founder Population Identifies Candidate Genes for Asthma. PLoS ONE, 2014, 9, e104396.	1.1	42
180	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

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181	Deep sequencing of multiple regions of glial tumors reveals spatial heterogeneity for mutations in clinically relevant genes. Genome Biology, 2014, 15, 530.	3.8	64
182	Systematic Dissection of Coding Exons at Single Nucleotide Resolution Supports an Additional Role in Cell-Specific Transcriptional Regulation. PLoS Genetics, 2014, 10, e1004592.	1.5	36
183	Adaptive Gene Amplification As an Intermediate Step in the Expansion of Virus Host Range. PLoS Pathogens, 2014, 10, e1004002.	2.1	51
184	Complex MSH2 and MSH6 mutations in hypermutated microsatellite unstable advanced prostate cancer. Nature Communications, 2014, 5, 4988.	5.8	219
185	RNF213 Rare Variants in an Ethnically Diverse Population With Moyamoya Disease. Stroke, 2014, 45, 3200-3207.	1.0	129
186	Genetic Variation Meets Replication Origins. Cell, 2014, 159, 973-974.	13.5	2
187	In vitro, long-range sequence information for de novo genome assembly via transposase contiguity. Genome Research, 2014, 24, 2041-2049.	2.4	150
188	Detection of minimal residual disease in NPM1-mutated acute myeloid leukemia by next-generation sequencing. Modern Pathology, 2014, 27, 1438-1446.	2.9	49
189	Life after genetics. Genome Medicine, 2014, 6, 86.	3.6	6
190	Adenylate cyclase 1 (ADCY1) mutations cause recessive hearing impairment in humans and defects in hair cell function and hearing in zebrafish. Human Molecular Genetics, 2014, 23, 3289-3298.	1.4	48
191	Species-Level Deconvolution of Metagenome Assemblies with Hi-Câ€“Based Contact Probability Maps. G3: Genes, Genomes, Genetics, 2014, 4, 1339-1346.	0.8	177
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