

Jay Shendure

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.

358 papers	61,480 citations	123 h-index	246 g-index
400 ext. papers	77,423 ext. citations	20.5 avg, IF	7.91 L-index

#	Paper	IF	Citations
358	High-content CRISPR screening. <i>Nature Reviews Methods Primers</i> , 2022 , 2,		10
357	Systematic reconstruction of cellular trajectories across mouse embryogenesis.. <i>Nature Genetics</i> , 2022 , 54, 328-341	36.3	3
356	A hybrid open-top light-sheet microscope for versatile multi-scale imaging of cleared tissues.. <i>Nature Methods</i> , 2022 , 19, 613-619	21.6	4
355	The glucose-sensing transcription factor MLX balances metabolism and stress to suppress apoptosis and maintain spermatogenesis. <i>PLoS Biology</i> , 2021 , 19, e3001085	9.7	0
354	Adaptations in Hippo-Yap signaling and myofibroblast fate underlie scar-free ear appendage wound healing in spiny mice. <i>Developmental Cell</i> , 2021 , 56, 2722-2740.e6	10.2	6
353	Precise genomic deletions using paired prime editing. <i>Nature Biotechnology</i> , 2021 ,	44.5	14
352	SwabExpress: An end-to-end protocol for extraction-free COVID-19 testing 2021 ,		25
351	Comprehensive characterization of tissue-specific chromatin accessibility in L2 nematodes. <i>Genome Research</i> , 2021 , 31, 1952-1969	9.7	2
350	Comparable Specimen Collection from Both Ends of At-Home Midturbinate Swabs. <i>Journal of Clinical Microbiology</i> , 2021 , 59,	9.7	2
349	Genome-wide strand asymmetry in massively parallel reporter activity favors genic strands. <i>Genome Research</i> , 2021 , 31, 866-876	9.7	0
348	Viral genomes reveal patterns of the SARS-CoV-2 outbreak in Washington State. <i>Science Translational Medicine</i> , 2021 , 13,	17.5	18
347	SwabExpress: An end-to-end protocol for extraction-free covid-19 testing. <i>Clinical Chemistry</i> , 2021 ,	5.5	8
346	Embryo-scale, single-cell spatial transcriptomics. <i>Science</i> , 2021 , 373, 111-117	33.3	29
345	Incidence of Medically Attended Acute Respiratory Illnesses Due to Respiratory Viruses Across the Life Course During the 2018/19 Influenza Season. <i>Clinical Infectious Diseases</i> , 2021 , 73, 802-807	11.6	3
344	CADD-Splice-improving genome-wide variant effect prediction using deep learning-derived splice scores. <i>Genome Medicine</i> , 2021 , 13, 31	14.4	64
343	The landscape of alternative polyadenylation in single cells of the developing mouse embryo. <i>Nature Communications</i> , 2021 , 12, 5101	17.4	4
342	Benchmarked approaches for reconstruction of in vitro cell lineages and in silico models of C. elegans and M. musculus developmental trees. <i>Cell Systems</i> , 2021 , 12, 810-826.e4	10.6	4

341	Single-cell lineage tracing of metastatic cancer reveals selection of hybrid EMT states. <i>Cancer Cell</i> , 2021 , 39, 1150-1162.e9	24.3	23
340	Single-cell landscape of nuclear configuration and gene expression during stem cell differentiation and X inactivation. <i>Genome Biology</i> , 2021 , 22, 279	18.3	2
339	The Seattle Flu Study: when regulations hinder pandemic surveillance.. <i>Nature Medicine</i> , 2021 ,	50.5	0
338	Trans- and cis-acting effects of Firre on epigenetic features of the inactive X chromosome. <i>Nature Communications</i> , 2020 , 11, 6053	17.4	15
337	Predicting mRNA Abundance Directly from Genomic Sequence Using Deep Convolutional Neural Networks. <i>Cell Reports</i> , 2020 , 31, 107663	10.6	40
336	Elevated exopolysaccharide levels in <i>Pseudomonas aeruginosa</i> flagellar mutants have implications for biofilm growth and chronic infections. <i>PLoS Genetics</i> , 2020 , 16, e1008848	6	24
335	Genomic surveillance reveals multiple introductions of SARS-CoV-2 into Northern California. <i>Science</i> , 2020 , 369, 582-587	33.3	162
334	lentiMPRA and MPRAflow for high-throughput functional characterization of gene regulatory elements. <i>Nature Protocols</i> , 2020 , 15, 2387-2412	18.8	17
333	Multimodal single-cell analysis reveals distinct radioresistant stem-like and progenitor cell populations in murine glioma. <i>Glia</i> , 2020 , 68, 2486-2502	9	4
332	Towards a comprehensive catalogue of validated and target-linked human enhancers. <i>Nature Reviews Genetics</i> , 2020 , 21, 292-310	30.1	99
331	Early Detection of Covid-19 through a Citywide Pandemic Surveillance Platform. <i>New England Journal of Medicine</i> , 2020 , 383, 185-187	59.2	64
330	Sci-fate characterizes the dynamics of gene expression in single cells. <i>Nature Biotechnology</i> , 2020 , 38, 980-988	44.5	34
329	Before the Flood. <i>Clinical Infectious Diseases</i> , 2020 , 71, 2513-2515	11.6	
328	Unsupervised manifold alignment for single-cell multi-omics data 2020 , 2020, 1-10		5
327	Viral genomes reveal patterns of the SARS-CoV-2 outbreak in Washington State 2020 ,		7
326	A brief history of human disease genetics. <i>Nature</i> , 2020 , 577, 179-189	50.4	181
325	Single-cell ATAC-Seq in human pancreatic islets and deep learning upscaling of rare cells reveals cell-specific type 2 diabetes regulatory signatures. <i>Molecular Metabolism</i> , 2020 , 32, 109-121	8.8	46
324	Massively multiplex chemical transcriptomics at single-cell resolution. <i>Science</i> , 2020 , 367, 45-51	33.3	67

323	Capturing cell type-specific chromatin compartment patterns by applying topic modeling to single-cell Hi-C data. <i>PLoS Computational Biology</i> , 2020 , 16, e1008173	5	20
322	A systematic evaluation of the design and context dependencies of massively parallel reporter assays. <i>Nature Methods</i> , 2020 , 17, 1083-1091	21.6	28
321	The Seattle Flu Study: a multiarm community-based prospective study protocol for assessing influenza prevalence, transmission and genomic epidemiology. <i>BMJ Open</i> , 2020 , 10, e037295	3	11
320	A human cell atlas of fetal chromatin accessibility. <i>Science</i> , 2020 , 370,	33.3	75
319	A human cell atlas of fetal gene expression. <i>Science</i> , 2020 , 370,	33.3	130
318	Cryptic transmission of SARS-CoV-2 in Washington state. <i>Science</i> , 2020 , 370, 571-575	33.3	135
317	Sci-Hi-C: A single-cell Hi-C method for mapping 3D genome organization in large number of single cells. <i>Methods</i> , 2020 , 170, 61-68	4.6	30
316	Suppressor mutations in -null mice implicate the DNA damage response in Rett syndrome pathology. <i>Genome Research</i> , 2020 , 30, 540-552	9.7	6
315	High-Throughput Single-Cell Sequencing with Linear Amplification. <i>Molecular Cell</i> , 2019 , 76, 676-690.e10	17.6	45
314	A pooled single-cell genetic screen identifies regulatory checkpoints in the continuum of the epithelial-to-mesenchymal transition. <i>Nature Genetics</i> , 2019 , 51, 1389-1398	36.3	79
313	Supervised classification enables rapid annotation of cell atlases. <i>Nature Methods</i> , 2019 , 16, 983-986	21.6	157
312	Mechanisms of Interplay between Transcription Factors and the 3D Genome. <i>Molecular Cell</i> , 2019 , 76, 306-319	17.6	70
311	Expanding the single-cell genomics toolkit. <i>Nature Genetics</i> , 2019 , 51, 931-932	36.3	1
310	Functional testing of thousands of osteoarthritis-associated variants for regulatory activity. <i>Nature Communications</i> , 2019 , 10, 2434	17.4	36
309	Massively parallel profiling and predictive modeling of the outcomes of CRISPR/Cas9-mediated double-strand break repair. <i>Nucleic Acids Research</i> , 2019 , 47, 7989-8003	20.1	74
308	Integration of multiple epigenomic marks improves prediction of variant impact in saturation mutagenesis reporter assay. <i>Human Mutation</i> , 2019 , 40, 1280-1291	4.7	19
307	Genomic Medicine-Progress, Pitfalls, and Promise. <i>Cell</i> , 2019 , 177, 45-57	56.2	75
306	Mutations in the translocon-associated protein complex subunit SSR3 cause a novel congenital disorder of glycosylation. <i>Journal of Inherited Metabolic Disease</i> , 2019 , 42, 993-997	5.4	10

305	DPAGT1 Deficiency with Encephalopathy (DPAGT1-CDG): Clinical and Genetic Description of 11 New Patients. <i>JIMD Reports</i> , 2019 , 44, 85-92	1.9	11
304	Saturation mutagenesis of twenty disease-associated regulatory elements at single base-pair resolution. <i>Nature Communications</i> , 2019 , 10, 3583	17.4	71
303	Chromatin compartment dynamics in a haploinsufficient model of cardiac laminopathy. <i>Journal of Cell Biology</i> , 2019 , 218, 2919-2944	7.3	24
302	The Rhododendron Genome and Chromosomal Organization Provide Insight into Shared Whole-Genome Duplications across the Heath Family (Ericaceae). <i>Genome Biology and Evolution</i> , 2019 , 11, 3353-3371	3.9	22
301	MaveDB: an open-source platform to distribute and interpret data from multiplexed assays of variant effect. <i>Genome Biology</i> , 2019 , 20, 223	18.3	42
300	A combination of transcription factors mediates inducible interchromosomal contacts. <i>ELife</i> , 2019 , 8,	8.9	8
299	High Sensitivity Profiling of Chromatin Structure by MNase-SSP. <i>Cell Reports</i> , 2019 , 26, 2465-2476.e4	10.6	15
298	The single-cell transcriptional landscape of mammalian organogenesis. <i>Nature</i> , 2019 , 566, 496-502	50.4	826
297	LB21. The Seattle Flu Study: A Community-Based Study of Influenza. <i>Open Forum Infectious Diseases</i> , 2019 , 6, S1002-S1002	1	6
296	Recommendations for the collection and use of multiplexed functional data for clinical variant interpretation. <i>Genome Medicine</i> , 2019 , 11, 85	14.4	23
295	Condensin-Dependent Chromatin Compaction Represses Transcription Globally during Quiescence. <i>Molecular Cell</i> , 2019 , 73, 533-546.e4	17.6	42
294	A Genome-wide Framework for Mapping Gene Regulation via Cellular Genetic Screens. <i>Cell</i> , 2019 , 176, 377-390.e19	56.2	188
293	CADD: predicting the deleteriousness of variants throughout the human genome. <i>Nucleic Acids Research</i> , 2019 , 47, D886-D894	20.1	1165
292	Highly scalable generation of DNA methylation profiles in single cells. <i>Nature Biotechnology</i> , 2018 , 36, 428-431	44.5	125
291	On the design of CRISPR-based single-cell molecular screens. <i>Nature Methods</i> , 2018 , 15, 271-274	21.6	96
290	Using DNase Hi-C techniques to map global and local three-dimensional genome architecture at high resolution. <i>Methods</i> , 2018 , 142, 59-73	4.6	16
289	Quantitative Missense Variant Effect Prediction Using Large-Scale Mutagenesis Data. <i>Cell Systems</i> , 2018 , 6, 116-124.e3	10.6	100
288	Identifying Novel Enhancer Elements with CRISPR-Based Screens. <i>ACS Chemical Biology</i> , 2018 , 13, 326-332	12.9	16

287	The cis-regulatory dynamics of embryonic development at single-cell resolution. <i>Nature</i> , 2018 , 555, 538-542	54.2	199
286	Simultaneous single-cell profiling of lineages and cell types in the vertebrate brain. <i>Nature Biotechnology</i> , 2018 , 36, 442-450	44.5	299
285	Identification of a novel interspecific hybrid yeast from a metagenomic spontaneously inoculated beer sample using Hi-C. <i>Yeast</i> , 2018 , 35, 71-84	3.4	25
284	A Single-Cell Atlas of In Vivo Mammalian Chromatin Accessibility. <i>Cell</i> , 2018 , 174, 1309-1324.e18	56.2	331
283	Cicero Predicts cis-Regulatory DNA Interactions from Single-Cell Chromatin Accessibility Data. <i>Molecular Cell</i> , 2018 , 71, 858-871.e8	17.6	247
282	Functional characterization of enhancer evolution in the primate lineage. <i>Genome Biology</i> , 2018 , 19, 99	18.3	16
281	FlashFry: a fast and flexible tool for large-scale CRISPR target design. <i>BMC Biology</i> , 2018 , 16, 74	7.3	40
280	High-resolution comparative analysis of great ape genomes. <i>Science</i> , 2018 , 360,	33.3	178
279	Tagmentation-Based Library Preparation for Low DNA Input Whole Genome Bisulfite Sequencing. <i>Methods in Molecular Biology</i> , 2018 , 1708, 105-122	1.4	7
278	New insights into structural features and optimal detection of circulating tumor DNA determined by single-strand DNA analysis. <i>Npj Genomic Medicine</i> , 2018 , 3, 31	6.2	45
277	Genomic Analyses from Non-invasive Prenatal Testing Reveal Genetic Associations, Patterns of Viral Infections, and Chinese Population History. <i>Cell</i> , 2018 , 175, 347-359.e14	56.2	123
276	Prediction of Susceptibility to First-Line Tuberculosis Drugs by DNA Sequencing. <i>New England Journal of Medicine</i> , 2018 , 379, 1403-1415	59.2	243
275	A Multiplex Homology-Directed DNA Repair Assay Reveals the Impact of More Than 1,000 BRCA1 Missense Substitution Variants on Protein Function. <i>American Journal of Human Genetics</i> , 2018 , 103, 498-508	11	62
274	Accurate classification of BRCA1 variants with saturation genome editing. <i>Nature</i> , 2018 , 562, 217-222	50.4	308
273	Joint profiling of chromatin accessibility and gene expression in thousands of single cells. <i>Science</i> , 2018 , 361, 1380-1385	33.3	381
272	Multiplex assessment of protein variant abundance by massively parallel sequencing. <i>Nature Genetics</i> , 2018 , 50, 874-882	36.3	163
271	Massively multiplex single-cell Hi-C. <i>Nature Methods</i> , 2017 , 14, 263-266	21.6	308
270	BRCA Testing by Single-Molecule Molecular Inversion Probes. <i>Clinical Chemistry</i> , 2017 , 63, 503-512	5.5	33

269	Activation of a cryptic splice site in the mitochondrial elongation factor GFM1 causes combined OXPHOS deficiency. <i>Mitochondrion</i> , 2017 , 34, 84-90	4.9	16
268	Single-molecule sequencing and chromatin conformation capture enable de novo reference assembly of the domestic goat genome. <i>Nature Genetics</i> , 2017 , 49, 643-650	36.3	323
267	The State of Whole-Genome Sequencing 2017 , 45-62		2
266	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. <i>American Journal of Human Genetics</i> , 2017 , 101, 768-788	11	81
265	DNA sequencing at 40: past, present and future. <i>Nature</i> , 2017 , 550, 345-353	50.4	486
264	Variant Interpretation: Functional Assays to the Rescue. <i>American Journal of Human Genetics</i> , 2017 , 101, 315-325	11	171
263	The 4D nucleome project. <i>Nature</i> , 2017 , 549, 219-226	50.4	332
262	Encephalopathy caused by novel mutations in the CMP-sialic acid transporter, SLC35A1. <i>American Journal of Medical Genetics, Part A</i> , 2017 , 173, 2906-2911	2.5	16
261	CRISPR/Cas9-Mediated Scanning for Regulatory Elements Required for HPRT1 Expression via Thousands of Large, Programmed Genomic Deletions. <i>American Journal of Human Genetics</i> , 2017 , 101, 192-205	11	87
260	Comprehensive single-cell transcriptional profiling of a multicellular organism. <i>Science</i> , 2017 , 357, 661-663	67.3	645
259	Haplotype phasing of whole human genomes using bead-based barcode partitioning in a single tube. <i>Nature Biotechnology</i> , 2017 , 35, 852-857	44.5	30
258	Quantification of differential gene expression by multiplexed targeted resequencing of cDNA. <i>Nature Communications</i> , 2017 , 8, 15190	17.4	8
257	A systematic comparison reveals substantial differences in chromosomal versus episomal encoding of enhancer activity. <i>Genome Research</i> , 2017 , 27, 38-52	9.7	148
256	Novel approach to genetic analysis and results in 3000 hemophilia patients enrolled in the My Life, Our Future initiative. <i>Blood Advances</i> , 2017 , 1, 824-834	7.8	52
255	The dynamic three-dimensional organization of the diploid yeast genome. <i>ELife</i> , 2017 , 6,	8.9	40
254	Targeted Capture and High-Throughput Sequencing Using Molecular Inversion Probes (MIPs). <i>Methods in Molecular Biology</i> , 2017 , 1492, 95-106	1.4	8
253	The power of multiplexed functional analysis of genetic variants. <i>Nature Protocols</i> , 2016 , 11, 1782-7	18.8	69
252	Human genomics: A deep dive into genetic variation. <i>Nature</i> , 2016 , 536, 277-8	50.4	7

251	Genome evolution in the allotetraploid frog <i>Xenopus laevis</i> . <i>Nature</i> , 2016 , 538, 336-343	50.4	510
250	Novel mutations in the genes TGM1 and ALOXE3 underlying autosomal recessive congenital ichthyosis. <i>International Journal of Dermatology</i> , 2016 , 55, 524-30	1.7	4
249	Massively Parallel Genetics. <i>Genetics</i> , 2016 , 203, 617-9	4	21
248	Cilia gene mutations cause atrioventricular septal defects by multiple mechanisms. <i>Human Molecular Genetics</i> , 2016 , 25, 3011-3028	5.6	27
247	Mitral regurgitation as a phenotypic manifestation of nonphotosensitive trichothiodystrophy due to a splice variant in MPLKIP. <i>BMC Medical Genetics</i> , 2016 , 17, 13	2.1	4
246	Rare variant associations with waist-to-hip ratio in European-American and African-American women from the NHLBI-Exome Sequencing Project. <i>European Journal of Human Genetics</i> , 2016 , 24, 1181-573	5.3	2
245	Cell-free DNA Comprises an In Vivo Nucleosome Footprint that Informs Its Tissues-Of-Origin. <i>Cell</i> , 2016 , 164, 57-68	56.2	664
244	Accelerating matchmaking of novel dysmorphology syndromes through clinical and genomic characterization of a large cohort. <i>Genetics in Medicine</i> , 2016 , 18, 686-95	8.1	35
243	Multiplex pairwise assembly of array-derived DNA oligonucleotides. <i>Nucleic Acids Research</i> , 2016 , 44, e43	20.1	31
242	Understanding Spatial Genome Organization: Methods and Insights. <i>Genomics, Proteomics and Bioinformatics</i> , 2016 , 14, 7-20	6.5	34
241	Substantial interindividual and limited intraindividual genomic diversity among tumors from men with metastatic prostate cancer. <i>Nature Medicine</i> , 2016 , 22, 369-78	50.5	425
240	Expansion of the spectrum of ITGB6-related disorders to adolescent alopecia, dentogingival abnormalities and intellectual disability. <i>European Journal of Human Genetics</i> , 2016 , 24, 1223-7	5.3	12
239	Large-scale targeted sequencing comparison highlights extreme genetic heterogeneity in nephronophthisis-related ciliopathies. <i>Journal of Medical Genetics</i> , 2016 , 53, 208-14	5.8	33
238	-associated developmental disorders exhibit distinct classes of mutations with variable expression and tissue distribution. <i>JCI Insight</i> , 2016 , 1,	9.9	90
237	FOXE3 mutations predispose to thoracic aortic aneurysms and dissections. <i>Journal of Clinical Investigation</i> , 2016 , 126, 948-61	15.9	65
236	Novel Approach to and Results of Genetic Analysis of 3000 Hemophilia Patients Enrolled in the MyLifeOurFuture Initiative. <i>Blood</i> , 2016 , 128, 205-205	2.2	1
235	Complex Minigene Library Vaccination for Discovery of Pre-Erythrocytic Plasmodium T Cell Antigens. <i>PLoS ONE</i> , 2016 , 11, e0153449	3.7	4
234	Fragment Length of Circulating Tumor DNA. <i>PLoS Genetics</i> , 2016 , 12, e1006162	6	349

233	Recurrent somatic loss of TNFRSF14 in classical Hodgkin lymphoma. <i>Genes Chromosomes and Cancer</i> , 2016 , 55, 278-87	5	22
232	ALG1-CDG: Clinical and Molecular Characterization of 39 Unreported Patients. <i>Human Mutation</i> , 2016 , 37, 653-60	4.7	30
231	Genome sequencing in a case of Niemann-Pick type C. <i>Journal of Physical Education and Sports Management</i> , 2016 , 2, a001222	2.8	9
230	Expanding the clinical and genetic heterogeneity of hereditary disorders of connective tissue. <i>Human Genetics</i> , 2016 , 135, 525-540	6.3	61
229	Long-read sequence assembly of the gorilla genome. <i>Science</i> , 2016 , 352, aae0344	33.3	282
228	Whole-organism lineage tracing by combinatorial and cumulative genome editing. <i>Science</i> , 2016 , 353, aaf7907	33.3	409
227	LOX Mutations Predispose to Thoracic Aortic Aneurysms and Dissections. <i>Circulation Research</i> , 2016 , 118, 928-34	15.7	122
226	Association of MTOR Mutations With Developmental Brain Disorders, Including Megalencephaly, Focal Cortical Dysplasia, and Pigmentary Mosaicism. <i>JAMA Neurology</i> , 2016 , 73, 836-845	17.2	166
225	Mapping 3D genome architecture through in situ DNase Hi-C. <i>Nature Protocols</i> , 2016 , 11, 2104-21	18.8	66
224	Classification and characterization of microsatellite instability across 18 cancer types. <i>Nature Medicine</i> , 2016 , 22, 1342-1350	50.5	432
223	Mammalian target of rapamycin pathway mutations cause hemimegalencephaly and focal cortical dysplasia. <i>Annals of Neurology</i> , 2015 , 77, 720-5	9.4	183
222	MIPSTR: a method for multiplex genotyping of germline and somatic STR variation across many individuals. <i>Genome Research</i> , 2015 , 25, 750-61	9.7	27
221	Mutation of ATF6 causes autosomal recessive achromatopsia. <i>Human Genetics</i> , 2015 , 134, 941-50	6.3	51
220	Running spell-check to identify regulatory variants. <i>Nature Genetics</i> , 2015 , 47, 853-5	36.3	5
219	High-throughput determination of RNA structure by proximity ligation. <i>Nature Biotechnology</i> , 2015 , 33, 980-4	44.5	78
218	Rare A2ML1 variants confer susceptibility to otitis media. <i>Nature Genetics</i> , 2015 , 47, 917-20	36.3	29
217	The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities. <i>American Journal of Human Genetics</i> , 2015 , 97, 199-215	11	432
216	A homozygous missense variant in type I keratin KRT25 causes autosomal recessive woolly hair. <i>Journal of Medical Genetics</i> , 2015 , 52, 676-80	5.8	17

215	Accurate identification of centromere locations in yeast genomes using Hi-C. <i>Nucleic Acids Research</i> , 2015 , 43, 5331-9	20.1	38
214	Massively Parallel Functional Analysis of BRCA1 RING Domain Variants. <i>Genetics</i> , 2015 , 200, 413-22	4	190
213	An siRNA-based functional genomics screen for the identification of regulators of ciliogenesis and ciliopathyl genes. <i>Nature Cell Biology</i> , 2015 , 17, 1074-1087	23.4	140
212	Escape from X inactivation varies in mouse tissues. <i>PLoS Genetics</i> , 2015 , 11, e1005079	6	142
211	Exome sequencing reveals pathogenic mutations in 91 strains of mice with Mendelian disorders. <i>Genome Research</i> , 2015 , 25, 948-57	9.7	38
210	The lncRNA Firre anchors the inactive X chromosome to the nucleolus by binding CTCF and maintains H3K27me3 methylation. <i>Genome Biology</i> , 2015 , 16, 52	18.3	170
209	A germline homozygous mutation in the base-excision repair gene NTHL1 causes adenomatous polyposis and colorectal cancer. <i>Nature Genetics</i> , 2015 , 47, 668-71	36.3	229
208	Multiplex single cell profiling of chromatin accessibility by combinatorial cellular indexing. <i>Science</i> , 2015 , 348, 910-4	33.3	668
207	Haplotype-resolved genome sequencing: experimental methods and applications. <i>Nature Reviews Genetics</i> , 2015 , 16, 344-58	30.1	120
206	Autosomal-Dominant Multiple Pterygium Syndrome Is Caused by Mutations in MYH3. <i>American Journal of Human Genetics</i> , 2015 , 96, 841-9	11	36
205	Copy-number variation and false positive prenatal aneuploidy screening results. <i>New England Journal of Medicine</i> , 2015 , 372, 1639-45	59.2	90
204	The origins, determinants, and consequences of human mutations. <i>Science</i> , 2015 , 349, 1478-83	33.3	111
203	Characterisation of mutations of the phosphoinositide-3-kinase regulatory subunit, PIK3R2, in perisylvian polymicrogyria: a next-generation sequencing study. <i>Lancet Neurology, The</i> , 2015 , 14, 1182-95 ^{24.1}	24.1	58
202	Learning the sequence determinants of alternative splicing from millions of random sequences. <i>Cell</i> , 2015 , 163, 698-711	56.2	136
201	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. <i>Journal of Virology</i> , 2015 , 89, 9986-97	6.6	17
200	Regional Isolation Drives Bacterial Diversification within Cystic Fibrosis Lungs. <i>Cell Host and Microbe</i> , 2015 , 18, 307-19	23.4	194
199	Challenges and solutions for gene identification in the presence of familial locus heterogeneity. <i>European Journal of Human Genetics</i> , 2015 , 23, 1207-15	5.3	29
198	Fine-scale chromatin interaction maps reveal the cis-regulatory landscape of human lincRNA genes. <i>Nature Methods</i> , 2015 , 12, 71-8	21.6	147

197	Large-scale genomic sequencing of extraintestinal pathogenic Escherichia coli strains. <i>Genome Research</i> , 2015 , 25, 119-28	9.7	116
196	Identification of genes escaping X inactivation by allelic expression analysis in a novel hybrid mouse model. <i>Data in Brief</i> , 2015 , 5, 761-9	1.2	3
195	Bipartite structure of the inactive mouse X chromosome. <i>Genome Biology</i> , 2015 , 16, 152	18.3	161
194	KIAA0586 is Mutated in Joubert Syndrome. <i>Human Mutation</i> , 2015 , 36, 831-5	4.7	44
193	Expanding the Molecular and Clinical Phenotype of SSR4-CDG. <i>Human Mutation</i> , 2015 , 36, 1048-51	4.7	15
192	A Year of Infection in the Intensive Care Unit: Prospective Whole Genome Sequencing of Bacterial Clinical Isolates Reveals Cryptic Transmissions and Novel Microbiota. <i>PLoS Genetics</i> , 2015 , 11, e1005413 ⁶		125
191	De novo mutations in NALCN cause a syndrome characterized by congenital contractures of the limbs and face, hypotonia, and developmental delay. <i>American Journal of Human Genetics</i> , 2015 , 96, 462-73 ¹¹		91
190	PI3K/AKT pathway mutations cause a spectrum of brain malformations from megalencephaly to focal cortical dysplasia. <i>Brain</i> , 2015 , 138, 1613-28	11.2	211
189	Whole genome prediction for preimplantation genetic diagnosis. <i>Genome Medicine</i> , 2015 , 7, 35	14.4	23
188	An essential cell cycle regulation gene causes hybrid inviability in Drosophila. <i>Science</i> , 2015 , 350, 1552-5 ^{33.3}		37
187	Massively parallel single-amino-acid mutagenesis. <i>Nature Methods</i> , 2015 , 12, 203-6, 4 p following 206	21.6	101
186	Exome sequencing in suspected monogenic dyslipidemias. <i>Circulation: Cardiovascular Genetics</i> , 2015 , 8, 343-50		36
185	MAT2A mutations predispose individuals to thoracic aortic aneurysms. <i>American Journal of Human Genetics</i> , 2015 , 96, 170-7	11	68
184	The complete genome sequence of a Neanderthal from the Altai Mountains. <i>Nature</i> , 2014 , 505, 43-9	50.4	1339
183	Rare-variant extensions of the transmission disequilibrium test: application to autism exome sequence data. <i>American Journal of Human Genetics</i> , 2014 , 94, 33-46	11	48
182	A new congenital disorder of glycosylation caused by a mutation in SSR4, the signal sequence receptor 4 protein of the TRAP complex. <i>Human Molecular Genetics</i> , 2014 , 23, 1602-5	5.6	33
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38	On the design of CRISPR-based single cell molecular screens		4
37	Functional Testing of Thousands of Osteoarthritis-Associated Variants for Regulatory Activity		1
36	Saturation mutagenesis of disease-associated regulatory elements		2

35	An open-source platform to distribute and interpret data from multiplexed assays of variant effect	2
34	Whole organism lineage tracing by combinatorial and cumulative genome editing	4
33	Single-molecule sequencing and conformational capture enable de novo mammalian reference genomes	12
32	Massively multiplex single-cell Hi-C	5
31	The 4D Nucleome Project	1
30	Comprehensive single cell transcriptional profiling of a multicellular organism by combinatorial indexing	8
29	Chromatin accessibility dynamics of myogenesis at single cell resolution	12
28	Scalable and efficient single-cell DNA methylation sequencing by combinatorial indexing	5
27	Using DNase Hi-C techniques to map global and local three-dimensional genome architecture at high resolution	2
26	FlashFry: a fast and flexible tool for large-scale CRISPR target design	2
25	The Seattle Flu Study: a multi-arm community-based prospective study protocol for assessing influenza prevalence, transmission, and genomic epidemiology	2
24	Unsupervised manifold alignment for single-cell multi-omics data	4
23	Single-cell lineage and transcriptome reconstruction of metastatic cancer reveals selection of aggressive hybrid EMT states	2
22	Simultaneous single-cell profiling of lineages and cell types in the vertebrate brain by scGESTALT	7
21	Multiplex Assessment of Protein Variant Abundance by Massively Parallel Sequencing	5
20	Dynamic reorganization of nuclear architecture during human cardiogenesis	6
19	Massively parallel dissection of human accelerated regions in human and chimpanzee neural progenitors	18
18	Functional Characterization of Enhancer Evolution in the Primate Lineage	2

17	Accurate functional classification of thousands of BRCA1 variants with saturation genome editing	12
16	A multiplexed homology-directed DNA repair assay reveals the impact of ~1,700 BRCA1 variants on protein function	2
15	crisprQTL mapping as a genome-wide association framework for cellular genetic screens	3
14	High-throughput mapping of meiotic crossover and chromosome mis-segregation events in interspecific hybrid mice	4
13	Predicting mRNA abundance directly from genomic sequence using deep convolutional neural networks	3
12	Massively parallel profiling and predictive modeling of the outcomes of CRISPR/Cas9-mediated double-strand break repair	6
11	Capturing cell type-specific chromatin structural patterns by applying topic modeling to single-cell Hi-C data	2
10	Supervised classification enables rapid annotation of cell atlases	15
9	Chromatin compartment dynamics in a haploinsufficient model of cardiac laminopathy	2
8	A systematic evaluation of the design, orientation, and sequence context dependencies of massively parallel reporter assays	8
7	Characterizing the temporal dynamics of gene expression in single cells with sci-fate	5
6	Trans- and cis-acting effects of the lncRNA Firre on epigenetic and structural features of the inactive X chromosome	7
5	A systematic comparison reveals substantial differences in chromosomal versus episomal encoding of enhancer activity	1
4	Paired CRISPR/Cas9 guide-RNAs enable high-throughput deletion scanning (ScanDel) of a Mendelian disease locus for functionally critical non-coding elements	1
3	Identification of a novel interspecific hybrid yeast from a metagenomic spontaneously inoculated beer sample using Hi-C	4
2	The landscape of alternative polyadenylation in single cells of the developing mouse embryo	1
1	Precise genomic deletions using paired prime editing	5