Jay Shendure

List of Publications by Citations

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	A general framework for estimating the relative pathogenicity of human genetic variants. *Nature Genetics*, **2014**, 46, 310-5	36.3	3626
357	Next-generation DNA sequencing. *Nature Biotechnology*, **2008**, 26, 1135-45	44.5	3040
356	Sporadic autism exomes reveal a highly interconnected protein network of de novo mutations. *Nature*, **2012**, 485, 246-50	50.4	1587
355	Exome sequencing identifies the cause of a mendelian disorder. *Nature Genetics*, **2010**, 42, 30-5	36.3	1573
354	Targeted capture and massively parallel sequencing of 12 human exomes. *Nature*, **2009**, 461, 272-6	50.4	1573
353	The contribution of de novo coding mutations to autism spectrum disorder. *Nature*, **2014**, 515, 216-21	50.4	1470
352	The complete genome sequence of a Neanderthal from the Altai Mountains. *Nature*, **2014**, 505, 43-9	50.4	1339
351	A high-coverage genome sequence from an archaic Denisovan individual. *Science*, **2012**, 338, 222-6	33.3	1276
350	Exome sequencing as a tool for Mendelian disease gene discovery. *Nature Reviews Genetics*, **2011**, 12, 745-55	30.1	1265
349	CADD: predicting the deleteriousness of variants throughout the human genome. *Nucleic Acids Research*, **2019**, 47, D886-D894	20.1	1165
348	Exome sequencing identifies MLL2 mutations as a cause of Kabuki syndrome. *Nature Genetics*, **2010**, 42, 790-3	36.3	1041
347	Accurate multiplex polony sequencing of an evolved bacterial genome. *Science*, **2005**, 309, 1728-32	33.3	1011
346	Integrative genome analyses identify key somatic driver mutations of small-cell lung cancer. *Nature Genetics*, **2012**, 44, 1104-10	36.3	919
345	Exome sequencing in sporadic autism spectrum disorders identifies severe de novo mutations. *Nature Genetics*, **2011**, 43, 585-9	36.3	899
344	Multiplex targeted sequencing identifies recurrently mutated genes in autism spectrum disorders. *Science*, **2012**, 338, 1619-22	33.3	892
343	Target-enrichment strategies for next-generation sequencing. *Nature Methods*, **2010**, 7, 111-8	21.6	863
342	The single-cell transcriptional landscape of mammalian organogenesis. *Nature*, **2019**, 566, 496-502	50.4	826

341	Analysis of genetic inheritance in a family quartet by whole-genome sequencing. *Science*, **2010**, 328, 636-9	33.3	822
340	Analysis of 6,515 exomes reveals the recent origin of most human protein-coding variants. *Nature*, **2013**, 493, 216-20	50.4	723
339	A three-dimensional model of the yeast genome. *Nature*, **2010**, 465, 363-7	50.4	722
338	Multiplex single cell profiling of chromatin accessibility by combinatorial cellular indexing. *Science*, **2015**, 348, 910-4	33.3	668
337	Cell-free DNA Comprises an In Vivo Nucleosome Footprint that Informs Its Tissues-Of-Origin. *Cell*, **2016**, 164, 57-68	56.2	664
336	Chromosome-scale scaffolding of de novo genome assemblies based on chromatin interactions. *Nature Biotechnology*, **2013**, 31, 1119-25	44.5	650
335	Comprehensive single-cell transcriptional profiling of a multicellular organism. *Science*, **2017**, 357, 661-667	33.3	645
334	De novo germline and postzygotic mutations in AKT3, PIK3R2 and PIK3CA cause a spectrum of related megalencephaly syndromes. *Nature Genetics*, **2012**, 44, 934-40	36.3	521
333	Genome evolution in the allotetraploid frog Xenopus laevis. *Nature*, **2016**, 538, 336-343	50.4	510
332	Targeted resequencing in epileptic encephalopathies identifies de novo mutations in CHD2 and SYNGAP1. *Nature Genetics*, **2013**, 45, 825-30	36.3	500
331	Diversity of human copy number variation and multicopy genes. *Science*, **2010**, 330, 641-6	33.3	491
330	DNA sequencing at 40: past, present and future. *Nature*, **2017**, 550, 345-353	50.4	486
329	Disruptive CHD8 mutations define a subtype of autism early in development. *Cell*, **2014**, 158, 263-276	56.2	467
328	Assemblathon 2: evaluating de novo methods of genome assembly in three vertebrate species. *GigaScience*, **2013**, 2, 10	7.6	461
327	Advanced sequencing technologies: methods and goals. *Nature Reviews Genetics*, **2004**, 5, 335-44	30.1	440
326	The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities. *American Journal of Human Genetics*, **2015**, 97, 199-215	11	432
325	Classification and characterization of microsatellite instability across 18 cancer types. *Nature Medicine*, **2016**, 22, 1342-1350	50.5	432
324	Refining analyses of copy number variation identifies specific genes associated with developmental delay. *Nature Genetics*, **2014**, 46, 1063-71	36.3	429

323	Substantial interindividual and limited intraindividual genomic diversity among tumors from men with metastatic prostate cancer. *Nature Medicine*, **2016**, 22, 369-78	50.5	425
322	Needles in stacks of needles: finding disease-causal variants in a wealth of genomic data. *Nature Reviews Genetics*, **2011**, 12, 628-40	30.1	423
321	Whole-organism lineage tracing by combinatorial and cumulative genome editing. *Science*, **2016**, 353, aaf7907	33.3	409
320	Joint profiling of chromatin accessibility and gene expression in thousands of single cells. *Science*, **2018**, 361, 1380-1385	33.3	381
319	Rapid, low-input, low-bias construction of shotgun fragment libraries by high-density in vitro transposition. *Genome Biology*, **2010**, 11, R119	18.3	377
318	Massively parallel functional dissection of mammalian enhancers in vivo. *Nature Biotechnology*, **2012**, 30, 265-70	44.5	366
317	Multiplex amplification of large sets of human exons. *Nature Methods*, **2007**, 4, 931-6	21.6	357
316	Fragment Length of Circulating Tumor DNA. *PLoS Genetics*, **2016**, 12, e1006162	6	349
315	The 4D nucleome project. *Nature*, **2017**, 549, 219-226	50.4	332
314	A Single-Cell Atlas of In Vivo Mammalian Chromatin Accessibility. *Cell*, **2018**, 174, 1309-1324.e18	56.2	331
313	A de novo convergence of autism genetics and molecular neuroscience. *Trends in Neurosciences*, **2014**, 37, 95-105	13.3	327
312	Single-molecule sequencing and chromatin conformation capture enable de novo reference assembly of the domestic goat genome. *Nature Genetics*, **2017**, 49, 643-650	36.3	323
311	Massively multiplex single-cell Hi-C. *Nature Methods*, **2017**, 14, 263-266	21.6	308
310	Accurate classification of BRCA1 variants with saturation genome editing. *Nature*, **2018**, 562, 217-222	50.4	308
309	Simultaneous single-cell profiling of lineages and cell types in the vertebrate brain. *Nature Biotechnology*, **2018**, 36, 442-450	44.5	299
308	Noninvasive whole-genome sequencing of a human fetus. *Science Translational Medicine*, **2012**, 4, 137ra76	17.5	294
307	Decoding long nanopore sequencing reads of natural DNA. *Nature Biotechnology*, **2014**, 32, 829-33	44.5	291
306	Long-read sequence assembly of the gorilla genome. *Science*, **2016**, 352, aae0344	33.3	282

305	Somatic mutations in cerebral cortical malformations. *New England Journal of Medicine*, **2014**, 371, 733-43	59.2	265
304	Global survey of escape from X inactivation by RNA-sequencing in mouse. *Genome Research*, **2010**, 20, 614-22	9.7	261
303	Transcriptome-wide miR-155 binding map reveals widespread noncanonical microRNA targeting. *Molecular Cell*, **2012**, 48, 760-70	17.6	257
302	TGFB2 mutations cause familial thoracic aortic aneurysms and dissections associated with mild systemic features of Marfan syndrome. *Nature Genetics*, **2012**, 44, 916-21	36.3	257
301	GRIN2A mutations cause epilepsy-aphasia spectrum disorders. *Nature Genetics*, **2013**, 45, 1073-6	36.3	249
300	The million mutation project: a new approach to genetics in Caenorhabditis elegans. *Genome Research*, **2013**, 23, 1749-62	9.7	249
299	Cicero Predicts cis-Regulatory DNA Interactions from Single-Cell Chromatin Accessibility Data. *Molecular Cell*, **2018**, 71, 858-871.e8	17.6	247
298	High-resolution analysis of DNA regulatory elements by synthetic saturation mutagenesis. *Nature Biotechnology*, **2009**, 27, 1173-5	44.5	247
297	Prediction of Susceptibility to First-Line Tuberculosis Drugs by DNA Sequencing. *New England Journal of Medicine*, **2018**, 379, 1403-1415	59.2	243
296	The expanding scope of DNA sequencing. *Nature Biotechnology*, **2012**, 30, 1084-94	44.5	237
295	The haplotype-resolved genome and epigenome of the aneuploid HeLa cancer cell line. *Nature*, **2013**, 500, 207-11	50.4	236
294	A germline homozygous mutation in the base-excision repair gene NTHL1 causes adenomatous polyposis and colorectal cancer. *Nature Genetics*, **2015**, 47, 668-71	36.3	229
293	Saturation editing of genomic regions by multiplex homology-directed repair. *Nature*, **2014**, 513, 120-3	50.4	223
292	Single molecule molecular inversion probes for targeted, high-accuracy detection of low-frequency variation. *Genome Research*, **2013**, 23, 843-54	9.7	221
291	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, 680-6	15.7	221
290	PI3K/AKT pathway mutations cause a spectrum of brain malformations from megalencephaly to focal cortical dysplasia. *Brain*, **2015**, 138, 1613-28	11.2	211
289	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, 17087-92	11.5	211
288	A molecular pathway including Id2, Tbx5, and Nkx2-5 required for cardiac conduction system development. *Cell*, **2007**, 129, 1365-76	56.2	209

287	Evidence for compensatory upregulation of expressed X-linked genes in mammals, Caenorhabditis elegans and Drosophila melanogaster. *Nature Genetics*, **2011**, 43, 1179-85	36.3	206
286	The cis-regulatory dynamics of embryonic development at single-cell resolution. *Nature*, **2018**, 555, 538-542	50.4	199
285	Regional Isolation Drives Bacterial Diversification within Cystic Fibrosis Lungs. *Cell Host and Microbe* , **2015**, 18, 307-19	23.4	194
284	Haplotype-resolved genome sequencing of a Gujarati Indian individual. *Nature Biotechnology*, **2011**, 29, 59-63	44.5	194
283	Massively Parallel Functional Analysis of BRCA1 RING Domain Variants. *Genetics*, **2015**, 200, 413-22	4	190
282	A Genome-wide Framework for Mapping Gene Regulation via Cellular Genetic Screens. *Cell*, **2019**, 176, 377-390.e19	56.2	188
281	Mammalian target of rapamycin pathway mutations cause hemimegalencephaly and focal cortical dysplasia. *Annals of Neurology*, **2015**, 77, 720-5	9.4	183
280	Complex MSH2 and MSH6 mutations in hypermutated microsatellite unstable advanced prostate cancer. *Nature Communications*, **2014**, 5, 4988	17.4	182
279	De novo mutations in the actin genes ACTB and ACTG1 cause Baraitser-Winter syndrome. *Nature Genetics*, **2012**, 44, 440-4, S1-2	36.3	181
278	A brief history of human disease genetics. *Nature*, **2020**, 577, 179-189	50.4	181
277	GABRA1 and STXBP1: novel genetic causes of Dravet syndrome. *Neurology*, **2014**, 82, 1245-53	6.5	180
276	Poxviruses deploy genomic accordions to adapt rapidly against host antiviral defenses. *Cell*, **2012**, 150, 831-41	56.2	180
275	High-resolution comparative analysis of great ape genomes. *Science*, **2018**, 360,	33.3	178
274	Identification of sex-specific quantitative trait loci controlling alcohol preference in C57BL/ 6 mice. *Nature Genetics*, **1996**, 13, 147-53	36.3	175
273	Variant Interpretation: Functional Assays to the Rescue. *American Journal of Human Genetics*, **2017**, 101, 315-325	11	171
272	The lncRNA Firre anchors the inactive X chromosome to the nucleolus by binding CTCF and maintains H3K27me3 methylation. *Genome Biology*, **2015**, 16, 52	18.3	170
271	Association of MTOR Mutations With Developmental Brain Disorders, Including Megalencephaly, Focal Cortical Dysplasia, and Pigmentary Mosaicism. *JAMA Neurology*, **2016**, 73, 836-845	17.2	166
270	Genome-scale identification of resistance functions in Pseudomonas aeruginosa using Tn-seq. *MBio* , **2011**, 2, e00315-10	7.8	165

269	Massively parallel exon capture and library-free resequencing across 16 genomes. *Nature Methods*, **2009**, 6, 315-6	21.6	164
268	Multiplex assessment of protein variant abundance by massively parallel sequencing. *Nature Genetics*, **2018**, 50, 874-882	36.3	163
267	Genomic surveillance reveals multiple introductions of SARS-CoV-2 into Northern California. *Science*, **2020**, 369, 582-587	33.3	162
266	Estimating the human mutation rate using autozygosity in a founder population. *Nature Genetics*, **2012**, 44, 1277-81	36.3	162
265	Bipartite structure of the inactive mouse X chromosome. *Genome Biology*, **2015**, 16, 152	18.3	161
264	Supervised classification enables rapid annotation of cell atlases. *Nature Methods*, **2019**, 16, 983-986	21.6	157
263	Massively parallel decoding of mammalian regulatory sequences supports a flexible organizational model. *Nature Genetics*, **2013**, 45, 1021-1028	36.3	155
262	Recurrent gain-of-function mutation in PRKG1 causes thoracic aortic aneurysms and acute aortic dissections. *American Journal of Human Genetics*, **2013**, 93, 398-404	11	153
261	A systematic comparison reveals substantial differences in chromosomal versus episomal encoding of enhancer activity. *Genome Research*, **2017**, 27, 38-52	9.7	148
260	Fine-scale chromatin interaction maps reveal the cis-regulatory landscape of human lincRNA genes. *Nature Methods*, **2015**, 12, 71-8	21.6	147
259	Massively parallel sequencing and rare disease. *Human Molecular Genetics*, **2010**, 19, R119-24	5.6	147
258	Rapid 16S rRNA next-generation sequencing of polymicrobial clinical samples for diagnosis of complex bacterial infections. *PLoS ONE*, **2013**, 8, e65226	3.7	144
257	Fluorescent in situ sequencing on polymerase colonies. *Analytical Biochemistry*, **2003**, 320, 55-65	3.1	144
256	A suppressor screen in Mecp2 mutant mice implicates cholesterol metabolism in Rett syndrome. *Nature Genetics*, **2013**, 45, 1013-20	36.3	143
255	Escape from X inactivation varies in mouse tissues. *PLoS Genetics*, **2015**, 11, e1005079	6	142
254	An siRNA-based functional genomics screen for the identification of regulators of ciliogenesis and ciliopathy genes. *Nature Cell Biology*, **2015**, 17, 1074-1087	23.4	140
253	Mutations in RSPH1 cause primary ciliary dyskinesia with a unique clinical and ciliary phenotype. *American Journal of Respiratory and Critical Care Medicine*, **2014**, 189, 707-17	10.2	139
252	Single-nucleotide evolutionary constraint scores highlight disease-causing mutations. *Nature Methods*, **2010**, 7, 250-1	21.6	139

251	Learning the sequence determinants of alternative splicing from millions of random sequences. *Cell*, **2015**, 163, 698-711	56.2	136
250	Identification by whole-genome resequencing of gene defect responsible for severe hypercholesterolemia. *Human Molecular Genetics*, **2010**, 19, 4313-8	5.6	136
249	Whole-exome capture and sequencing identifies HEATR2 mutation as a cause of primary ciliary dyskinesia. *American Journal of Human Genetics*, **2012**, 91, 685-93	11	135
248	Haploinsufficiency of SF3B4, a component of the pre-mRNA spliceosomal complex, causes Nager syndrome. *American Journal of Human Genetics*, **2012**, 90, 925-33	11	135
247	Cryptic transmission of SARS-CoV-2 in Washington state. *Science*, **2020**, 370, 571-575	33.3	135
246	Selection analyses of insertional mutants using subgenic-resolution arrays. *Nature Biotechnology*, **2001**, 19, 1060-5	44.5	134
245	Haplotype-resolved whole-genome sequencing by contiguity-preserving transposition and combinatorial indexing. *Nature Genetics*, **2014**, 46, 1343-9	36.3	132
244	Spectrum of MLL2 (ALR) mutations in 110 cases of Kabuki syndrome. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 1511-6	2.5	132
243	Digital genotyping and haplotyping with polymerase colonies. *Proceedings of the National Academy of Sciences of the United States of America*, **2003**, 100, 5926-31	11.5	130
242	A human cell atlas of fetal gene expression. *Science*, **2020**, 370,	33.3	130
241	megaTALs: a rare-cleaving nuclease architecture for therapeutic genome engineering. *Nucleic Acids Research*, **2014**, 42, 2591-601	20.1	128
240	Computational discovery of sense-antisense transcription in the human and mouse genomes. *Genome Biology*, **2002**, 3, RESEARCH0044	18.3	127
239	Highly scalable generation of DNA methylation profiles in single cells. *Nature Biotechnology*, **2018**, 36, 428-431	44.5	125
238	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	6	125
237	Mutations in PIEZO2 cause Gordon syndrome, Marden-Walker syndrome, and distal arthrogryposis type 5. *American Journal of Human Genetics*, **2014**, 94, 734-44	11	124
236	Genomic Analyses from Non-invasive Prenatal Testing Reveal Genetic Associations, Patterns of Viral Infections, and Chinese Population History. *Cell*, **2018**, 175, 347-359.e14	56.2	123
235	LOX Mutations Predispose to Thoracic Aortic Aneurysms and Dissections. *Circulation Research*, **2016**, 118, 928-34	15.7	122
234	Haplotype-resolved genome sequencing: experimental methods and applications. *Nature Reviews Genetics*, **2015**, 16, 344-58	30.1	120

233	Species-level deconvolution of metagenome assemblies with Hi-C-based contact probability maps. *G3: Genes, Genomes, Genetics*, **2014**, 4, 1339-46	3.2	120
232	Large-scale genomic sequencing of extraintestinal pathogenic Escherichia coli strains. *Genome Research*, **2015**, 25, 119-28	9.7	116
231	Parallel, tag-directed assembly of locally derived short sequence reads. *Nature Methods*, **2010**, 7, 119-22	21.6	116
230	In vitro, long-range sequence information for de novo genome assembly via transposase contiguity. *Genome Research*, **2014**, 24, 2041-9	9.7	112
229	The origins, determinants, and consequences of human mutations. *Science*, **2015**, 349, 1478-83	33.3	111
228	Exome sequencing identifies mutations in CCDC114 as a cause of primary ciliary dyskinesia. *American Journal of Human Genetics*, **2013**, 92, 99-106	11	111
227	Mutations in SPAG1 cause primary ciliary dyskinesia associated with defective outer and inner dynein arms. *American Journal of Human Genetics*, **2013**, 93, 711-20	11	109
226	Tagmentation-based whole-genome bisulfite sequencing. *Nature Protocols*, **2013**, 8, 2022-32	18.8	108
225	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	11.5	108
224	RNF213 rare variants in an ethnically diverse population with Moyamoya disease. *Stroke*, **2014**, 45, 3200-6	8.7	103
223	Massively parallel single-amino-acid mutagenesis. *Nature Methods*, **2015**, 12, 203-6, 4 p following 206	21.6	101
222	Quantitative Missense Variant Effect Prediction Using Large-Scale Mutagenesis Data. *Cell Systems*, **2018**, 6, 116-124.e3	10.6	100
221	Towards a comprehensive catalogue of validated and target-linked human enhancers. *Nature Reviews Genetics*, **2020**, 21, 292-310	30.1	99
220	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, 1310-9	2.5	99
219	MIPgen: optimized modeling and design of molecular inversion probes for targeted resequencing. *Bioinformatics*, **2014**, 30, 2670-2	7.2	98
218	Ultra-low-input, tagmentation-based whole-genome bisulfite sequencing. *Genome Research*, **2012**, 22, 1139-43	9.7	98
217	Methods for genomic partitioning. *Annual Review of Genomics and Human Genetics*, **2009**, 10, 263-84	9.7	98
216	On the design of CRISPR-based single-cell molecular screens. *Nature Methods*, **2018**, 15, 271-274	21.6	96

215	Mosaicism of the UDP-galactose transporter SLC35A2 causes a congenital disorder of glycosylation. *American Journal of Human Genetics*, **2013**, 92, 632-6	11	93
214	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, 462-73	11	91
213	Copy-number variation and false positive prenatal aneuploidy screening results. *New England Journal of Medicine*, **2015**, 372, 1639-45	59.2	90
212	-associated developmental disorders exhibit distinct classes of mutations with variable expression and tissue distribution. *JCI Insight*, **2016**, 1,	9.9	90
211	Mutation discovery in mice by whole exome sequencing. *Genome Biology*, **2011**, 12, R86	18.3	89
210	Next generation sequence analysis for mitochondrial disorders. *Genome Medicine*, **2009**, 1, 100	14.4	88
209	Single molecule profiling of alternative pre-mRNA splicing. *Science*, **2003**, 301, 836-8	33.3	88
208	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	11	87
207	Characterization of apparently balanced chromosomal rearrangements from the developmental genome anatomy project. *American Journal of Human Genetics*, **2008**, 82, 712-22	11	84
206	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. *American Journal of Human Genetics*, **2017**, 101, 768-788	11	81
205	De novo TBR1 mutations in sporadic autism disrupt protein functions. *Nature Communications*, **2014**, 5, 4954	17.4	80
204	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	11	80
203	Long-range polony haplotyping of individual human chromosome molecules. *Nature Genetics*, **2006**, 38, 382-7	36.3	80
202	A pooled single-cell genetic screen identifies regulatory checkpoints in the continuum of the epithelial-to-mesenchymal transition. *Nature Genetics*, **2019**, 51, 1389-1398	36.3	79
201	High-throughput determination of RNA structure by proximity ligation. *Nature Biotechnology*, **2015**, 33, 980-4	44.5	78
200	Next-generation human genetics. *Genome Biology*, **2011**, 12,	18.3	78
199	Exome-wide DNA capture and next generation sequencing in domestic and wild species. *BMC Genomics*, **2011**, 12, 347	4.5	77
198	Genomic Medicine-Progress, Pitfalls, and Promise. *Cell*, **2019**, 177, 45-57	56.2	75

197	A human cell atlas of fetal chromatin accessibility. *Science*, **2020**, 370,	33.3	75
196	Massively parallel profiling and predictive modeling of the outcomes of CRISPR/Cas9-mediated double-strand break repair. *Nucleic Acids Research*, **2019**, 47, 7989-8003	20.1	74
195	Trans genomic capture and sequencing of primate exomes reveals new targets of positive selection. *Genome Research*, **2011**, 21, 1686-94	9.7	74
194	Saturation mutagenesis of twenty disease-associated regulatory elements at single base-pair resolution. *Nature Communications*, **2019**, 10, 3583	17.4	71
193	Mechanisms of Interplay between Transcription Factors and the 3D Genome. *Molecular Cell*, **2019**, 76, 306-319	17.6	70
192	The power of multiplexed functional analysis of genetic variants. *Nature Protocols*, **2016**, 11, 1782-7	18.8	69
191	Mammalian X upregulation is associated with enhanced transcription initiation, RNA half-life, and MOF-mediated H4K16 acetylation. *Developmental Cell*, **2013**, 25, 55-68	10.2	69
190	MAT2A mutations predispose individuals to thoracic aortic aneurysms. *American Journal of Human Genetics*, **2015**, 96, 170-7	11	68
189	Massively multiplex chemical transcriptomics at single-cell resolution. *Science*, **2020**, 367, 45-51	33.3	67
188	Mutations in TBC1D24, a gene associated with epilepsy, also cause nonsyndromic deafness DFNB86. *American Journal of Human Genetics*, **2014**, 94, 144-52	11	66
187	Mapping 3D genome architecture through in situ DNase Hi-C. *Nature Protocols*, **2016**, 11, 2104-21	18.8	66
186	FOXE3 mutations predispose to thoracic aortic aneurysms and dissections. *Journal of Clinical Investigation*, **2016**, 126, 948-61	15.9	65
185	Early Detection of Covid-19 through a Citywide Pandemic Surveillance Platform. *New England Journal of Medicine*, **2020**, 383, 185-187	59.2	64
184	Extraordinary molecular evolution in the PRDM9 fertility gene. *PLoS ONE*, **2009**, 4, e8505	3.7	64
183	CADD-Splice-improving genome-wide variant effect prediction using deep learning-derived splice scores. *Genome Medicine*, **2021**, 13, 31	14.4	64
182	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	11	62
181	Expanding the clinical and genetic heterogeneity of hereditary disorders of connective tissue. *Human Genetics*, **2016**, 135, 525-540	6.3	61
180	Discovering functional transcription-factor combinations in the human cell cycle. *Genome Research*, **2005**, 15, 848-55	9.7	60

179	Characterisation of mutations of the phosphoinositide-3-kinase regulatory subunit, PIK3R2, in perisylvian polymicrogyria: a next-generation sequencing study. *Lancet Neurology, The*, **2015**, 14, 1182-95	24.1	58
178	Gorilla genome structural variation reveals evolutionary parallelisms with chimpanzee. *Genome Research*, **2011**, 21, 1640-9	9.7	56
177	Mutations in ECEL1 cause distal arthrogryposis type 5D. *American Journal of Human Genetics*, **2013**, 92, 150-6	11	55
176	Identification of foreign gene sequences by transcript filtering against the human genome. *Nature Genetics*, **2002**, 30, 141-2	36.3	54
175	Primate evolution of the recombination regulator PRDM9. *Nature Communications*, **2014**, 5, 4370	17.4	53
174	Targeted enrichment and high-resolution digital profiling of mitochondrial DNA deletions in human brain. *Aging Cell*, **2014**, 13, 29-38	9.9	53
173	A major influence of sex-specific loci on alcohol preference in C57Bl/6 and DBA/2 inbred mice. *Mammalian Genome*, **1998**, 9, 942-8	3.2	53
172	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	7.8	52
171	Mutation of ATF6 causes autosomal recessive achromatopsia. *Human Genetics*, **2015**, 134, 941-50	6.3	51
170	Computational comparison of two draft sequences of the human genome. *Nature*, **2001**, 409, 856-9	50.4	50
169	Deep sequencing of multiple regions of glial tumors reveals spatial heterogeneity for mutations in clinically relevant genes. *Genome Biology*, **2014**, 15, 530	18.3	49
168	Rare-variant extensions of the transmission disequilibrium test: application to autism exome sequence data. *American Journal of Human Genetics*, **2014**, 94, 33-46	11	48
167	Mutations in KCTD1 cause scalp-ear-nipple syndrome. *American Journal of Human Genetics*, **2013**, 92, 621-6	11	48
166	Accurate gene synthesis with tag-directed retrieval of sequence-verified DNA molecules. *Nature Methods*, **2012**, 9, 913-5	21.6	48
165	Whole-genome analysis reveals that mutations in inositol polyphosphate phosphatase-like 1 cause opsismodysplasia. *American Journal of Human Genetics*, **2013**, 92, 137-43	11	46
164	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	8.8	46
163	High-Throughput Single-Cell Sequencing with Linear Amplification. *Molecular Cell*, **2019**, 76, 676-690.e10	17.6	45
162	Detection of minimal residual disease in NPM1-mutated acute myeloid leukemia by next-generation sequencing. *Modern Pathology*, **2014**, 27, 1438-46	9.8	45

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