

Martin Kircher

List of Publications by Citations

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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.

76
papers

20,725
citations

43
h-index

90
g-index

90
ext. papers

26,381
ext. citations

14.3
avg, IF


6.9
L-index

#	Paper	IF	Citations
76	A general framework for estimating the relative pathogenicity of human genetic variants. <i>Nature Genetics</i> , 2014 , 46, 310-5	36.3	3626
75	A draft sequence of the Neandertal genome. <i>Science</i> , 2010 , 328, 710-722	33.3	2599
74	The complete genome sequence of a Neanderthal from the Altai Mountains. <i>Nature</i> , 2014 , 505, 43-9	50.4	1339
73	A high-coverage genome sequence from an archaic Denisovan individual. <i>Science</i> , 2012 , 338, 222-6	33.3	1276
72	Illumina sequencing library preparation for highly multiplexed target capture and sequencing. <i>Cold Spring Harbor Protocols</i> , 2010 , 2010, pdb.prot5448	1.2	1195
71	Genetic history of an archaic hominin group from Denisova Cave in Siberia. <i>Nature</i> , 2010 , 468, 1053-60	50.4	1169
70	CADD: predicting the deleteriousness of variants throughout the human genome. <i>Nucleic Acids Research</i> , 2019 , 47, D886-D894	20.1	1165
69	The evolution of gene expression levels in mammalian organs. <i>Nature</i> , 2011 , 478, 343-8	50.4	787
68	Deep proteome and transcriptome mapping of a human cancer cell line. <i>Molecular Systems Biology</i> , 2011 , 7, 548	12.2	723
67	Double indexing overcomes inaccuracies in multiplex sequencing on the Illumina platform. <i>Nucleic Acids Research</i> , 2012 , 40, e3	20.1	681
66	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
65	The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities. <i>American Journal of Human Genetics</i> , 2015 , 97, 199-215	11	432
64	High-throughput DNA sequencing--concepts and limitations. <i>BioEssays</i> , 2010 , 32, 524-36	4.1	397
63	Denisova admixture and the first modern human dispersals into Southeast Asia and Oceania. <i>American Journal of Human Genetics</i> , 2011 , 89, 516-28	11	390
62	Removal of deaminated cytosines and detection of in vivo methylation in ancient DNA. <i>Nucleic Acids Research</i> , 2010 , 38, e87	20.1	283
61	Ancient gene flow from early modern humans into Eastern Neanderthals. <i>Nature</i> , 2016 , 530, 429-33	50.4	269
60	Somatic mutations in cerebral cortical malformations. <i>New England Journal of Medicine</i> , 2014 , 371, 733-43	59.2	265

59	Characterization of ancient and modern genomes by SNP detection and phylogenomic and metagenomic analysis using PALEOMIX. <i>Nature Protocols</i> , 2014 , 9, 1056-82	18.8	231
58	Biallelic mutations in BRCA1 cause a new Fanconi anemia subtype. <i>Cancer Discovery</i> , 2015 , 5, 135-42	24.4	215
57	Transcriptomes of germinal zones of human and mouse fetal neocortex suggest a role of extracellular matrix in progenitor self-renewal. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2012 , 109, 11836-41	11.5	215
56	A complete mtDNA genome of an early modern human from Kostenki, Russia. <i>Current Biology</i> , 2010 , 20, 231-6	6.3	213
55	Improved base calling for the Illumina Genome Analyzer using machine learning strategies. <i>Genome Biology</i> , 2009 , 10, R83	18.3	188
54	Patterns of coding variation in the complete exomes of three Neandertals. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2014 , 111, 6666-71	11.5	175
53	Multiplex assessment of protein variant abundance by massively parallel sequencing. <i>Nature Genetics</i> , 2018 , 50, 874-882	36.3	163
52	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
51	Mutations in RSPH1 cause primary ciliary dyskinesia with a unique clinical and ciliary phenotype. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2014 , 189, 707-17	10.2	139
50	Analysis of high-throughput ancient DNA sequencing data. <i>Methods in Molecular Biology</i> , 2012 , 840, 197-228	12.9	129
49	The earliest transcribed zygotic genes are short, newly evolved, and different across species. <i>Cell Reports</i> , 2014 , 6, 285-92	10.6	121
48	Addressing challenges in the production and analysis of illumina sequencing data. <i>BMC Genomics</i> , 2011 , 12, 382	4.5	99
47	Mosaicism of the UDP-galactose transporter SLC35A2 causes a congenital disorder of glycosylation. <i>American Journal of Human Genetics</i> , 2013 , 92, 632-6	11	93
46	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	11.5	91
45	-associated developmental disorders exhibit distinct classes of mutations with variable expression and tissue distribution. <i>JCI Insight</i> , 2016 , 1,	9.9	90
44	Saturation mutagenesis of twenty disease-associated regulatory elements at single base-pair resolution. <i>Nature Communications</i> , 2019 , 10, 3583	17.4	71
43	freelbis: an efficient basecaller with calibrated quality scores for Illumina sequencers. <i>Bioinformatics</i> , 2013 , 29, 1208-9	7.2	68
42	CADD-Splice-improving genome-wide variant effect prediction using deep learning-derived splice scores. <i>Genome Medicine</i> , 2021 , 13, 31	14.4	64

41	Expanding the clinical and genetic heterogeneity of hereditary disorders of connective tissue. <i>Human Genetics</i> , 2016 , 135, 525-540	6.3	61
40	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
39	Whole exome sequencing identifies de novo heterozygous CAV1 mutations associated with a novel neonatal onset lipodystrophy syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2015 , 167A, 1796-806	8.5	49
38	Opsins in onychophora (velvet worms) suggest a single origin and subsequent diversification of visual pigments in arthropods. <i>Molecular Biology and Evolution</i> , 2012 , 29, 3451-8	8.3	49
37	GGC Repeat Expansion and Exon 1 Methylation of XYLT1 Is a Common Pathogenic Variant in Baratela-Scott Syndrome. <i>American Journal of Human Genetics</i> , 2019 , 104, 35-44	11	47
36	Structural conservation versus functional divergence of maternally expressed microRNAs in the Dlk1/Gtl2 imprinting region. <i>BMC Genomics</i> , 2008 , 9, 346	4.5	45
35	Evaluating intra- and inter-individual variation in the human placental transcriptome. <i>Genome Biology</i> , 2015 , 16, 54	18.3	43
34	Road blocks on paleogenomes—polymerase extension profiling reveals the frequency of blocking lesions in ancient DNA. <i>Nucleic Acids Research</i> , 2010 , 38, e161	20.1	43
33	Primate iPS cells as tools for evolutionary analyses. <i>Stem Cell Research</i> , 2014 , 12, 622-9	1.6	41
32	Exome sequencing reveals pathogenic mutations in 91 strains of mice with Mendelian disorders. <i>Genome Research</i> , 2015 , 25, 948-57	9.7	38
31	Autosomal-Dominant Multiple Pterygium Syndrome Is Caused by Mutations in MYH3. <i>American Journal of Human Genetics</i> , 2015 , 96, 841-9	11	36
30	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
29	The evolutionary and phylogeographic history of woolly mammoths: a comprehensive mitogenomic analysis. <i>Scientific Reports</i> , 2017 , 7, 44585	4.9	34
28	Comparative population genomics of the ejaculate in humans and the great apes. <i>Molecular Biology and Evolution</i> , 2013 , 30, 964-76	8.3	34
27	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
26	ALG1-CDG: Clinical and Molecular Characterization of 39 Unreported Patients. <i>Human Mutation</i> , 2016 , 37, 653-60	4.7	30
25	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
24	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

23	Bi-allelic POLR3A Loss-of-Function Variants Cause Autosomal-Recessive Wiedemann-Rautenstrauch Syndrome. <i>American Journal of Human Genetics</i> , 2018 , 103, 968-975	11	28
22	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
21	Massively parallel dissection of human accelerated regions in human and chimpanzee neural progenitors		18
20	lentiMPRA and MPRAflow for high-throughput functional characterization of gene regulatory elements. <i>Nature Protocols</i> , 2020 , 15, 2387-2412	18.8	17
19	Activation of a cryptic splice site in the mitochondrial elongation factor GFM1 causes combined OXPPOS deficiency. <i>Mitochondrion</i> , 2017 , 34, 84-90	4.9	16
18	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
17	SRD5A3-CDG: Expanding the phenotype of a congenital disorder of glycosylation with emphasis on adult onset features. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170, 3165-3171	2.5	16
16	Molecular outcomes, clinical consequences, and genetic diagnosis of Oculocutaneous Albinism in Pakistani population. <i>Scientific Reports</i> , 2017 , 7, 44185	4.9	15
15	Expanding the Molecular and Clinical Phenotype of SSR4-CDG. <i>Human Mutation</i> , 2015 , 36, 1048-51	4.7	15
14	Mutations in the fourth propeller domain of LRP4 are associated with isolated syndactyly with fusion of the third and fourth fingers. <i>Human Mutation</i> , 2018 , 39, 811-815	4.7	11
13	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
12	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
11	A systematic evaluation of the design, orientation, and sequence context dependencies of massively parallel reporter assays		8
10	Concurrent genome and epigenome editing by CRISPR-mediated sequence replacement. <i>BMC Biology</i> , 2019 , 17, 90	7.3	6
9	Running spell-check to identify regulatory variants. <i>Nature Genetics</i> , 2015 , 47, 853-5	36.3	5
8	Multiplex Assessment of Protein Variant Abundance by Massively Parallel Sequencing		5
7	Saturation mutagenesis of disease-associated regulatory elements		2
6	A framework to score the effects of structural variants in health and disease.. <i>Genome Research</i> , 2022 ,	9.7	2

5	A systematic comparison reveals substantial differences in chromosomal versus episomal encoding of enhancer activity		1
4	CADD-SV  framework to score the effects of structural variants in health and disease		1
3	HemoMIPs-Automated analysis and result reporting pipeline for targeted sequencing data. <i>PLoS Computational Biology</i> , 2020 , 16, e1007956	5	0
2	Bayesian Optimization Improves Tissue-Specific Prediction of Active Regulatory Regions with Deep Neural Networks. <i>Lecture Notes in Computer Science</i> , 2020 , 600-612	0.9	0
1	The impact of different negative training data on regulatory sequence predictions. <i>PLoS ONE</i> , 2020 , 15, e0237412	3.7	0