Martin Kircher

List of Publications by Year in Descending Order

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 framework to score the effects of structural variants in health and disease.. *Genome Research*, **2022**,	9.7	2
75	CADD-Splice-improving genome-wide variant effect prediction using deep learning-derived splice scores. *Genome Medicine*, **2021**, 13, 31	14.4	64
74	HemoMIPs-Automated analysis and result reporting pipeline for targeted sequencing data. *PLoS Computational Biology*, **2020**, 16, e1007956	5	0
73	lentiMPRA and MPRAflow for high-throughput functional characterization of gene regulatory elements. *Nature Protocols*, **2020**, 15, 2387-2412	18.8	17
72	Bayesian Optimization Improves Tissue-Specific Prediction of Active Regulatory Regions with Deep Neural Networks. *Lecture Notes in Computer Science*, **2020**, 600-612	0.9	0
71	The impact of different negative training data on regulatory sequence predictions. *PLoS ONE*, **2020**, 15, e0237412	3.7	0
70	A systematic evaluation of the design and context dependencies of massively parallel reporter assays. *Nature Methods*, **2020**, 17, 1083-1091	21.6	28
69	Integration of multiple epigenomic marks improves prediction of variant impact in saturation mutagenesis reporter assay. *Human Mutation*, **2019**, 40, 1280-1291	4.7	19
68	Mutations in the translocon-associated protein complex subunit SSR3 cause a novel congenital disorder of glycosylation. *Journal of Inherited Metabolic Disease*, **2019**, 42, 993-997	5.4	10
67	DPAGT1 Deficiency with Encephalopathy (DPAGT1-CDG): Clinical and Genetic Description of 11 New Patients. *JIMD Reports*, **2019**, 44, 85-92	1.9	11
66	Saturation mutagenesis of twenty disease-associated regulatory elements at single base-pair resolution. *Nature Communications*, **2019**, 10, 3583	17.4	71
65	Concurrent genome and epigenome editing by CRISPR-mediated sequence replacement. *BMC Biology*, **2019**, 17, 90	7.3	6
64	CADD: predicting the deleteriousness of variants throughout the human genome. *Nucleic Acids Research*, **2019**, 47, D886-D894	20.1	1165
63	GGC Repeat Expansion and Exon 1 Methylation of XYLT1 Is a Common Pathogenic Variant in Baratela-Scott Syndrome. *American Journal of Human Genetics*, **2019**, 104, 35-44	11	47
62	Mutations in the fourth β-propeller domain of LRP4 are associated with isolated syndactyly with fusion of the third and fourth fingers. *Human Mutation*, **2018**, 39, 811-815	4.7	11
61	Bi-allelic POLR3A Loss-of-Function Variants Cause Autosomal-Recessive Wiedemann-Rautenstrauch Syndrome. *American Journal of Human Genetics*, **2018**, 103, 968-975	11	28
60	Multiplex assessment of protein variant abundance by massively parallel sequencing. *Nature Genetics*, **2018**, 50, 874-882	36.3	163

59	Activation of a cryptic splice site in the mitochondrial elongation factor GFM1 causes combined OXPHOS deficiency. *Mitochondrion*, **2017**, 34, 84-90	4.9	16
58	Molecular outcomes, clinical consequences, and genetic diagnosis of Oculocutaneous Albinism in Pakistani population. *Scientific Reports*, **2017**, 7, 44185	4.9	15
57	The evolutionary and phylogeographic history of woolly mammoths: a comprehensive mitogenomic analysis. *Scientific Reports*, **2017**, 7, 44585	4.9	34
56	Encephalopathy caused by novel mutations in the CMP-sialic acid transporter, SLC35A1. *American Journal of Medical Genetics, Part A*, **2017**, 173, 2906-2911	2.5	16
55	A systematic comparison reveals substantial differences in chromosomal versus episomal encoding of enhancer activity. *Genome Research*, **2017**, 27, 38-52	9.7	148
54	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
53	Cell-free DNA Comprises an In Vivo Nucleosome Footprint that Informs Its Tissues-Of-Origin. *Cell*, **2016**, 164, 57-68	56.2	664
52	Accelerating matchmaking of novel dysmorphology syndromes through clinical and genomic characterization of a large cohort. *Genetics in Medicine*, **2016**, 18, 686-95	8.1	35
51	Ancient gene flow from early modern humans into Eastern Neanderthals. *Nature*, **2016**, 530, 429-33	50.4	269
50	-associated developmental disorders exhibit distinct classes of mutations with variable expression and tissue distribution. *JCI Insight*, **2016**, 1,	9.9	90
49	ALG1-CDG: Clinical and Molecular Characterization of 39 Unreported Patients. *Human Mutation*, **2016**, 37, 653-60	4.7	30
48	Expanding the clinical and genetic heterogeneity of hereditary disorders of connective tissue. *Human Genetics*, **2016**, 135, 525-540	6.3	61
47	SRD5A3-CDG: Expanding the phenotype of a congenital disorder of glycosylation with emphasis on adult onset features. *American Journal of Medical Genetics, Part A*, **2016**, 170, 3165-3171	2.5	16
46	Biallelic mutations in BRCA1 cause a new Fanconi anemia subtype. *Cancer Discovery*, **2015**, 5, 135-42	24.4	215
45	Running spell-check to identify regulatory variants. *Nature Genetics*, **2015**, 47, 853-5	36.3	5
44	The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities. *American Journal of Human Genetics*, **2015**, 97, 199-215	11	432
43	Exome sequencing reveals pathogenic mutations in 91 strains of mice with Mendelian disorders. *Genome Research*, **2015**, 25, 948-57	9.7	38
42	Autosomal-Dominant Multiple Pterygium Syndrome Is Caused by Mutations in MYH3. *American Journal of Human Genetics*, **2015**, 96, 841-9	11	36

41	Challenges and solutions for gene identification in the presence of familial locus heterogeneity. *European Journal of Human Genetics*, **2015**, 23, 1207-15	5.3	29
40	Expanding the Molecular and Clinical Phenotype of SSR4-CDG. *Human Mutation*, **2015**, 36, 1048-51	4.7	15
39	Whole exome sequencing identifies de novo heterozygous CAV1 mutations associated with a novel neonatal onset lipodystrophy syndrome. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 1796-806	2.5	49
38	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
37	Evaluating intra- and inter-individual variation in the human placental transcriptome. *Genome Biology*, **2015**, 16, 54	18.3	43
36	Patterns of coding variation in the complete exomes of three Neandertals. *Proceedings of the National Academy of Sciences of the United States of America*, **2014**, 111, 6666-71	11.5	175
35	Characterization of ancient and modern genomes by SNP detection and phylogenomic and metagenomic analysis using PALEOMIX. *Nature Protocols*, **2014**, 9, 1056-82	18.8	231
34	The complete genome sequence of a Neanderthal from the Altai Mountains. *Nature*, **2014**, 505, 43-9	50.4	1339
33	A new congenital disorder of glycosylation caused by a mutation in SSR4, the signal sequence receptor 4 protein of the TRAP complex. *Human Molecular Genetics*, **2014**, 23, 1602-5	5.6	33
32	A general framework for estimating the relative pathogenicity of human genetic variants. *Nature Genetics*, **2014**, 46, 310-5	36.3	3626
31	Somatic mutations in cerebral cortical malformations. *New England Journal of Medicine*, **2014**, 371, 733-43	59.2	265
30	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
29	The earliest transcribed zygotic genes are short, newly evolved, and different across species. *Cell Reports*, **2014**, 6, 285-92	10.6	121
28	Primate iPS cells as tools for evolutionary analyses. *Stem Cell Research*, **2014**, 12, 622-9	1.6	41
27	Mosaicism of the UDP-galactose transporter SLC35A2 causes a congenital disorder of glycosylation. *American Journal of Human Genetics*, **2013**, 92, 632-6	11	93
26	Comparative population genomics of the ejaculate in humans and the great apes. *Molecular Biology and Evolution*, **2013**, 30, 964-76	8.3	34
25	freeIbis: an efficient basecaller with calibrated quality scores for Illumina sequencers. *Bioinformatics*, **2013**, 29, 1208-9	7.2	68
24	Opsins in onychophora (velvet worms) suggest a single origin and subsequent diversification of visual pigments in arthropods. *Molecular Biology and Evolution*, **2012**, 29, 3451-8	8.3	49

23	A high-coverage genome sequence from an archaic Denisovan individual. *Science*, **2012**, 338, 222-6	33.3	1276
22	Analysis of high-throughput ancient DNA sequencing data. *Methods in Molecular Biology*, **2012**, 840, 197-228	1.4	129
21	Double indexing overcomes inaccuracies in multiplex sequencing on the Illumina platform. *Nucleic Acids Research*, **2012**, 40, e3	20.1	681
20	Transcriptomes of germinal zones of human and mouse fetal neocortex suggest a role of extracellular matrix in progenitor self-renewal. *Proceedings of the National Academy of Sciences of the United States of America*, **2012**, 109, 11836-41	11.5	215
19	Deep proteome and transcriptome mapping of a human cancer cell line. *Molecular Systems Biology*, **2011**, 7, 548	12.2	723
18	The evolution of gene expression levels in mammalian organs. *Nature*, **2011**, 478, 343-8	50.4	787
17	Denisova admixture and the first modern human dispersals into Southeast Asia and Oceania. *American Journal of Human Genetics*, **2011**, 89, 516-28	11	390
16	Addressing challenges in the production and analysis of illumina sequencing data. *BMC Genomics*, **2011**, 12, 382	4.5	99
15	Genetic history of an archaic hominin group from Denisova Cave in Siberia. *Nature*, **2010**, 468, 1053-60	50.4	1169
14	Road blocks on paleogenomes--polymerase extension profiling reveals the frequency of blocking lesions in ancient DNA. *Nucleic Acids Research*, **2010**, 38, e161	20.1	43
13	Removal of deaminated cytosines and detection of in vivo methylation in ancient DNA. *Nucleic Acids Research*, **2010**, 38, e87	20.1	283
12	A draft sequence of the Neandertal genome. *Science*, **2010**, 328, 710-722	33.3	2599
11	Illumina sequencing library preparation for highly multiplexed target capture and sequencing. *Cold Spring Harbor Protocols*, **2010**, 2010, pdb.prot5448	1.2	1195
10	A complete mtDNA genome of an early modern human from Kostenki, Russia. *Current Biology*, **2010** , 20, 231-6	6.3	213
9	High-throughput DNA sequencing--concepts and limitations. *BioEssays*, **2010**, 32, 524-36	4.1	397
8	Improved base calling for the Illumina Genome Analyzer using machine learning strategies. *Genome Biology*, **2009**, 10, R83	18.3	188
7	Structural conservation versus functional divergence of maternally expressed microRNAs in the Dlk1/Gtl2 imprinting region. *BMC Genomics*, **2008**, 9, 346	4.5	45
6	Saturation mutagenesis of disease-associated regulatory elements		2

5	Multiplex Assessment of Protein Variant Abundance by Massively Parallel Sequencing	5
4	Massively parallel dissection of human accelerated regions in human and chimpanzee neural progenitors	18
3	A systematic evaluation of the design, orientation, and sequence context dependencies of massively parallel reporter assays	8
2	A systematic comparison reveals substantial differences in chromosomal versus episomal encoding of enhancer activity	1
1	CADD-SV: a framework to score the effects of structural variants in health and disease	1