# Martin Kircher

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/2049566/publications.pdf

Version: 2024-02-01

| | | 50170 | 79541 |
|---|---|---|---|
| 78 | 29,822 | 46 | 73 |
| papers | citations | h-index | g-index |

| | | | |
|---|---|---|---|
| 90 | 90 | 90 | 47206 |
| all docs | docs citations | times ranked | citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | A general framework for estimating the relative pathogenicity of human genetic variants. Nature Genetics, 2014, 46, 310-315. | 9.4 | 5,167 |
| 2 | A Draft Sequence of the Neandertal Genome. Science, 2010, 328, 710-722. | 6.0 | 3,588 |
| 3 | CADD: predicting the deleteriousness of variants throughout the human genome. Nucleic Acids Research, 2019, 47, D886-D894. | 6.5 | 2,360 |
| 4 | The complete genome sequence of a Neanderthal from the Altai Mountains. Nature, 2014, 505, 43-49. | 13.7 | 1,830 |
| 5 | A High-Coverage Genome Sequence from an Archaic Denisovan Individual. Science, 2012, 338, 222-226. | 6.0 | 1,695 |
| 6 | Illumina Sequencing Library Preparation for Highly Multiplexed Target Capture and Sequencing. Cold Spring Harbor Protocols, 2010, 2010, pdb.prot5448. | 0.2 | 1,690 |
| 7 | Genetic history of an archaic hominin group from Denisova Cave in Siberia. Nature, 2010, 468, 1053-1060. | 13.7 | 1,537 |
| 8 | The evolution of gene expression levels in mammalian organs. Nature, 2011, 478, 343-348. | 13.7 | 1,080 |
| 9 | Cell-free DNA Comprises an InÂVivo Nucleosome Footprint that Informs Its Tissues-Of-Origin. Cell, 2016, 164, 57-68. | 13.5 | 1,039 |
| 10 | Double indexing overcomes inaccuracies in multiplex sequencing on the Illumina platform. Nucleic Acids Research, 2012, 40, e3-e3. | 6.5 | 944 |
| 11 | Deep proteome and transcriptome mapping of a human cancer cell line. Molecular Systems Biology, 2011, 7, 548. | 3.2 | 878 |
| 12 | The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities. American Journal of Human Genetics, 2015, 97, 199-215. | 2.6 | 574 |
| 13 | Denisova Admixture and the First Modern Human Dispersals into Southeast Asia and Oceania. American Journal of Human Genetics, 2011, 89, 516-528. | 2.6 | 525 |
| 14 | Highâ€‰throughput DNA sequencing â€“ concepts and limitations. BioEssays, 2010, 32, 524-536. | 1.2 | 490 |
| 15 | Characterization of ancient and modern genomes by SNP detection and phylogenomic and metagenomic analysis using PALEOMIX. Nature Protocols, 2014, 9, 1056-1082. | 5.5 | 403 |
| 16 | Ancient gene flow from early modern humans into Eastern Neanderthals. Nature, 2016, 530, 429-433. | 13.7 | 392 |
| 17 | CADD-Spliceâ€”improving genome-wide variant effect prediction using deep learning-derived splice scores. Genome Medicine, 2021, 13, 31. | 3.6 | 375 |
| 18 | Removal of deaminated cytosines and detection of in vivo methylation in ancient DNA. Nucleic Acids Research, 2010, 38, e87-e87. | 6.5 | 362 |

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|---|---|---|---|
| 19 | Somatic Mutations in Cerebral Cortical Malformations. New England Journal of Medicine, 2014, 371, 733-743. | 13.9 | 326 |
| 20 | Multiplex assessment of protein variant abundance by massively parallel sequencing. Nature Genetics, 2018, 50, 874-882. | 9.4 | 323 |
| 21 | 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, 11836-11841. | 3.3 | 282 |
| 22 | A Complete mtDNA Genome of an Early Modern Human from Kostenki, Russia. Current Biology, 2010, 20, 231-236. | 1.8 | 252 |
| 23 | Biallelic Mutations in BRCA1 Cause a New Fanconi Anemia Subtype. Cancer Discovery, 2015, 5, 135-142. | 7.7 | 251 |
| 24 | A systematic comparison reveals substantial differences in chromosomal versus episomal encoding of enhancer activity. Genome Research, 2017, 27, 38-52. | 2.4 | 244 |
| 25 | 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, 6666-6671. | 3.3 | 223 |
| 26 | Improved base calling for the Illumina Genome Analyzer using machine learning strategies. Genome Biology, 2009, 10, R83. | 13.9 | 212 |
| 27 | 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 |
| 28 | The Earliest Transcribed Zygotic Genes Are Short, Newly Evolved, and Different across Species. Cell Reports, 2014, 6, 285-292. | 2.9 | 179 |
| 29 | Analysis of High-Throughput Ancient DNA Sequencing Data. Methods in Molecular Biology, 2012, 840, 197-228. | 0.4 | 177 |
| 30 | Saturation mutagenesis of twenty disease-associated regulatory elements at single base-pair resolution. Nature Communications, 2019, 10, 3583. | 5.8 | 152 |
| 31 | PIK3CA-associated developmental disorders exhibit distinct classes of mutations with variable expression and tissue distribution. JCI Insight, 2016, 1, . | 2.3 | 134 |
| 32 | Addressing challenges in the production and analysis of illumina sequencing data. BMC Genomics, 2011, 12, 382. | 1.2 | 126 |
| 33 | 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 |
| 34 | Mosaicism of the UDP-Galactose Transporter SLC35A2 Causes a Congenital Disorder of Glycosylation. American Journal of Human Genetics, 2013, 92, 632-636. | 2.6 | 114 |
| 35 | A systematic evaluation of the design and context dependencies of massively parallel reporter assays. Nature Methods, 2020, 17, 1083-1091. | 9.0 | 111 |
| 36 | Expanding the clinical and genetic heterogeneity of hereditary disorders of connective tissue. Human Genetics, 2016, 135, 525-540. | 1.8 | 89 |

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|---|---|---|---|
| 37 | 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 |
| 38 | 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. | 2.6 | 81 |
| 39 | freelbis: an efficient basecaller with calibrated quality scores for Illumina sequencers. Bioinformatics, 2013, 29, 1208-1209. | 1.8 | 71 |
| 40 | 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, 167, 1796-1806. | 0.7 | 71 |
| 41 | lentiMPRA and MPRAflow for high-throughput functional characterization of gene regulatory elements. Nature Protocols, 2020, 15, 2387-2412. | 5.5 | 65 |
| 42 | Opsins in Onychophora (Velvet Worms) Suggest a Single Origin and Subsequent Diversification of Visual Pigments in Arthropods. Molecular Biology and Evolution, 2012, 29, 3451-3458. | 3.5 | 61 |
| 43 | Primate iPS cells as tools for evolutionary analyses. Stem Cell Research, 2014, 12, 622-629. | 0.3 | 61 |
| 44 | Autosomal-Dominant Multiple Pterygium Syndrome Is Caused by Mutations in MYH3. American Journal of Human Genetics, 2015, 96, 841-849. | 2.6 | 55 |
| 45 | 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 |
| 46 | Exome sequencing reveals pathogenic mutations in 91 strains of mice with Mendelian disorders. Genome Research, 2015, 25, 948-957. | 2.4 | 54 |
| 47 | Structural conservation versus functional divergence of maternally expressed microRNAs in the Dlk1/Gtl2 imprinting region. BMC Genomics, 2008, 9, 346. | 1.2 | 49 |
| 48 | Road blocks on paleogenomesâ€"polymerase extension profiling reveals the frequency of blocking lesions in ancient DNA. Nucleic Acids Research, 2010, 38, e161-e161. | 6.5 | 47 |
| 49 | Integration of multiple epigenomic marks improves prediction of variant impact in saturation mutagenesis reporter assay. Human Mutation, 2019, 40, 1280-1291. | 1.1 | 46 |
| 50 | 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, 1602-1605. | 1.4 | 45 |
| 51 | Evaluating intra- and inter-individual variation in the human placental transcriptome. Genome Biology, 2015, 16, 54. | 3.8 | 45 |
| 52 | Bi-allelic POLR3A Loss-of-Function Variants Cause Autosomal-Recessive Wiedemann-Rautenstrauch Syndrome. American Journal of Human Genetics, 2018, 103, 968-975. | 2.6 | 43 |
| 53 | Comparative Population Genomics of the Ejaculate in Humans and the Great Apes. Molecular Biology and Evolution, 2013, 30, 964-976. | 3.5 | 40 |
| 54 | ALG1-CDG: Clinical and Molecular Characterization of 39 Unreported Patients. Human Mutation, 2016, 37, 653-660. | 1.1 | 40 |

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|---|---|---|---|
| 55 | The evolutionary and phylogeographic history of woolly mammoths: a comprehensive mitogenomic analysis. Scientific Reports, 2017, 7, 44585. | 1.6 | 39 |
| 56 | 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 |
| 57 | 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 |
| 58 | Molecular outcomes, clinical consequences, and genetic diagnosis of Oculocutaneous Albinism in Pakistani population. Scientific Reports, 2017, 7, 44185. | 1.6 | 25 |
| 59 | SRD5A3â€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. | 0.7 | 23 |
| 60 | Expanding the Molecular and Clinical Phenotype of SSR4-CDG. Human Mutation, 2015, 36, 1048-1051. | 1.1 | 22 |
| 61 | Activation of a cryptic splice site in the mitochondrial elongation factor GFM1 causes combined OXPHOS deficiency. Mitochondrion, 2017, 34, 84-90. | 1.6 | 21 |
| 62 | 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 |
| 63 | Mutations in the fourth Î²-propeller domain of LRP4 are associated with isolated syndactyly with fusion of the third and fourth fingers. Human Mutation, 2018, 39, 811-815. | 1.1 | 17 |
| 64 | A framework to score the effects of structural variants in health and disease. Genome Research, 2022, 32, 766-777. | 2.4 | 17 |
| 65 | DPAGT1 Deficiency with Encephalopathy (DPAGT1-CDG): Clinical and Genetic Description of 11 New Patients. JIMD Reports, 2018, 44, 85-92. | 0.7 | 16 |
| 66 | 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 |
| 67 | Concurrent genome and epigenome editing by CRISPR-mediated sequence replacement. BMC Biology, 2019, 17, 90. | 1.7 | 9 |
| 68 | Computational and experimental methods for classifying variants of unknown clinical significance.. Cold Spring Harbor Molecular Case Studies, 2022, 8, . | 0.7 | 7 |
| 69 | Running spell-check to identify regulatory variants. Nature Genetics, 2015, 47, 853-855. | 9.4 | 5 |
| 70 | The impact of different negative training data on regulatory sequence predictions. PLoS ONE, 2020, 15, e0237412. | 1.1 | 4 |
| 71 | HemoMIPsâ€”Automated analysis and result reporting pipeline for targeted sequencing data. PLoS Computational Biology, 2020, 16, e1007956. | 1.5 | 2 |
| 72 | Bayesian Optimization Improves Tissue-Specific Prediction of Active Regulatory Regions with Deep Neural Networks. Lecture Notes in Computer Science, 2020, , 600-612. | 1.0 | 1 |

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|---|---|---|---|
| 73 | The impact of different negative training data on regulatory sequence predictions. , 2020, 15, e0237412. | | 0 |
| 74 | The impact of different negative training data on regulatory sequence predictions. , 2020, 15, e0237412. | | 0 |
| 75 | The impact of different negative training data on regulatory sequence predictions. , 2020, 15, e0237412. | | 0 |
| 76 | The impact of different negative training data on regulatory sequence predictions. , 2020, 15, e0237412. | | 0 |
| 77 | The impact of different negative training data on regulatory sequence predictions. , 2020, 15, e0237412. | | 0 |
| 78 | The impact of different negative training data on regulatory sequence predictions. , 2020, 15, e0237412. | | 0 |