Konrad J Karczewski

List of Publications by Year
in descending order

70	31,108	53794 45	79698 73
papers	citations	h-index	g-index

108	108	108	56935
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Analysis of protein-coding genetic variation in 60,706 humans. Nature, 2016, 536, 285-291.	27.8	9,051
2	The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 2020, 581, 434-443.	27.8	6,140
3	Annotation of functional variation in personal genomes using RegulomeDB. Genome Research, 2012, 22, 1790-1797.	5.5	2,335
4	Personal Omics Profiling Reveals Dynamic Molecular and Medical Phenotypes. Cell, 2012, 148, 1293-1307.	28.9	1,134
5	The UK10K project identifies rare variants in health and disease. Nature, 2015, 526, 82-90.	27.8	1,014
6	Landscape of X chromosome inactivation across human tissues. Nature, 2017, 550, 244-248.	27.8	764
7	Integrative omics for health and disease. Nature Reviews Genetics, 2018, 19, 299-310.	16.3	676
8	De novo mutations in congenital heart disease with neurodevelopmental and other congenital anomalies. Science, 2015, 350, 1262-1266.	12.6	646
9	A structural variation reference for medical and population genetics. Nature, 2020, 581, 444-451.	27.8	614
10	The ExAC browser: displaying reference data information from over 60 000 exomes. Nucleic Acids Research, 2017, 45, D840-D845.	14.5	587
11	Mendelian randomization of blood lipids for coronary heart disease. European Heart Journal, 2015, 36, 539-550.	2.2	567
12	Variation in Transcription Factor Binding Among Humans. Science, 2010, 328, 232-235.	12.6	521
13	Improving genetic diagnosis in Mendelian disease with transcriptome sequencing. Science Translational Medicine, 2017, 9, .	12.4	516
14	Performance comparison of exome DNA sequencing technologies. Nature Biotechnology, 2011, 29, 908-914.	17.5	464
15	Using high-resolution variant frequencies to empower clinical genome interpretation. Genetics in Medicine, 2017, 19, 1151-1158.	2.4	355
16	Rare coding variants in ten genes confer substantial risk for schizophrenia. Nature, 2022, 604, 509-516.	27.8	326
17	Refining the role of de novo protein-truncating variants in neurodevelopmental disorders by using population reference samples. Nature Genetics, 2017, 49, 504-510.	21.4	298
18	Human knockouts and phenotypic analysis in a cohort with a high rate of consanguinity. Nature, 2017, 544, 235-239.	27.8	292

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19	Quantifying prion disease penetrance using large population control cohorts. Science Translational Medicine, 2016, 8, 322ra9.	12.4	289
20	Health and population effects of rare gene knockouts in adult humans with related parents. Science, 2016, 352, 474-477.	12.6	272
21	Effect of predicted protein-truncating genetic variants on the human transcriptome. Science, 2015, 348, 666-669.	12.6	252
22	Bioinformatics challenges for personalized medicine. Bioinformatics, 2011, 27, 1741-1748.	4.1	223
23	The origin, evolution, and functional impact of short insertionâ€“deletion variants identified in 179 human genomes. Genome Research, 2013, 23, 749-761.	5.5	206
24	Patterns of genic intolerance of rare copy number variation in 59,898 human exomes. Nature Genetics, 2016, 48, 1107-1111.	21.4	167
25	Gene-centric Meta-analysis in 87,736 Individuals of European Ancestry Identifies Multiple Blood-Pressure-Related Loci. American Journal of Human Genetics, 2014, 94, 349-360.	6.2	158
26	Transcript expression-aware annotation improves rare variant interpretation. Nature, 2020, 581, 452-458.	27.8	142
27	Phased Whole-Genome Genetic Risk in a Family Quartet Using a Major Allele Reference Sequence. PLoS Genetics, 2011, 7, e1002280.	3.5	137
28	Extensive genetic variation in somatic human tissues. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, 18018-18023.	7.1	136
29	Tractor uses local ancestry to enable the inclusion of admixed individuals in GWAS and to boost power. Nature Genetics, 2021, 53, 195-204.	21.4	125
30	Evaluating drug targets through human loss-of-function genetic variation. Nature, 2020, 581, 459-464.	27.8	115
31	Quantifying the Impact of Rare and Ultra-rare Coding Variation across the Phenotypic Spectrum. American Journal of Human Genetics, 2018, 102, 1204-1211.	6.2	102
32	Characterising the loss-of-function impact of 5â€™ untranslated region variants in 15,708 individuals. Nature Communications, 2020, 11, 2523.	12.8	99
33	Landscape of multi-nucleotide variants in 125,748 human exomes and 15,708 genomes. Nature Communications, 2020, 11, 2539.	12.8	98
34	Systematic functional regulatory assessment of disease-associated variants. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 9607-9612.	7.1	85
35	Evidence That Personal Genome Testing Enhances Student Learning in a Course on Genomics and Personalized Medicine. PLoS ONE, 2013, 8, e68853.	2.5	84
36	The effect of LRRK2 loss-of-function variants in humans. Nature Medicine, 2020, 26, 869-877.	30.7	79

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37	Regulatory Variation Within and Between Species. Annual Review of Genomics and Human Genetics, 2011, 12, 327-346.	6.2	70
38	Insights into the genetic epidemiology of Crohn's and rare diseases in the Ashkenazi Jewish population. PLoS Genetics, 2018, 14, e1007329.	3.5	66
39	Transcriptome Sequencing of a Large Human Family Identifies the Impact of Rare Noncoding Variants. American Journal of Human Genetics, 2014, 95, 245-256.	6.2	63
40	Chapter 7: Pharmacogenomics. PLoS Computational Biology, 2012, 8, e1002817.	3.2	57
41	Genetic variant in folate homeostasis is associated with lower warfarin dose in African Americans. Blood, 2014, 124, 2298-2305.	1.4	57
42	Efficient genotype compression and analysis of large genetic-variation data sets. Nature Methods, 2016, 13, 63-65.	19.0	57
43	Haplotype Sharing Provides Insights into Fine-Scale Population History and Disease in Finland. American Journal of Human Genetics, 2018, 102, 760-775.	6.2	57
44	Cooperative transcription factor associations discovered using regulatory variation. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 13353-13358.	7.1	53
45	Quantifying supercoiling-induced denaturation bubbles in DNA. Soft Matter, 2012, 8, 8651.	2.7	50
46	Concept and design of a genome-wide association genotyping array tailored for transplantation-specific studies. Genome Medicine, 2015, 7, 90.	8.2	49
47	Addendum: The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 2021, 597, E3-E4.	27.8	45
48	Non-coding region variants upstream of MEF2C cause severe developmental disorder through three distinct loss-of-function mechanisms. American Journal of Human Genetics, 2021, 108, 1083-1094.	6.2	42
49	Base-specific mutational intolerance near splice sites clarifies the role of nonessential splice nucleotides. Genome Research, 2018, 28, 968-974.	5.5	41
50	Using ALoFT to determine the impact of putative loss-of-function variants in protein-coding genes. Nature Communications, 2017, 8, 382.	12.8	40
51	Quantifying unobserved protein-coding variants in human populations provides a roadmap for large-scale sequencing projects. Nature Communications, 2016, 7, 13293.	12.8	35
52	Coherent Functional Modules Improve Transcription Factor Target Identification, Cooperativity Prediction, and Disease Association. PLoS Genetics, 2014, 10, e1004122.	3.5	29
53	Ten Simple Rules to Enable Multi-site Collaborations through Data Sharing. PLoS Computational Biology, 2017, 13, e1005278.	3.2	29
54	Gene-centric meta-analyses for central adiposity traits in up to 57 412 individuals of European descent confirm known loci and reveal several novel associations. Human Molecular Genetics, 2014, 23, 2498-2510.	2.9	28

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55	A New System for Comparative Functional Genomics of Saccharomyces Yeasts. Genetics, 2013, 195, 275-287.	2.9	27
56	Whole-genome analysis of human embryonic stem cells enables rational line selection based on genetic variation. Cell Stem Cell, 2022, 29, 472-486.e7.	11.1	27
57	STORMSeq: An Open-Source, User-Friendly Pipeline for Processing Personal Genomics Data in the Cloud. PLoS ONE, 2014, 9, e84860.	2.5	25
58	Human genetic analyses of organelles highlight the nucleus in age-related trait heritability. ELife, 2021, 10, .	6.0	20
59	Small RNA Sequencing in Cells and Exosomes Identifies eQTLs and 14q32 as a Region of Active Export. G3: Genes, Genomes, Genetics, 2017, 7, 31-39.	1.8	16
60	Using High-Resolution Variant Frequencies Empowers Clinical Genome Interpretation and Enables Investigation of Genetic Architecture. American Journal of Human Genetics, 2019, 104, 187-190.	6.2	15
61	Population based frequency of naturally occurring lossâ€ofâ€function variants in genes associated with platelet disorders. Journal of Thrombosis and Haemostasis, 2021, 19, 248-254.	3.8	13
62	Interpretome: a freely available, modular, and secure personal genome interpretation engine. Pacific Symposium on Biocomputing Pacific Symposium on Biocomputing, 2012, , 339-350.	0.7	12
63	Genetic meta-analysis of 15,901 African Americans identifies variation in EXOC3L1 is associated with HDL concentration. Journal of Lipid Research, 2015, 56, 1781-1786.	4.2	11
64	Human mutational constraint as a tool to understand biology of rare and emerging bone marrow failure syndromes. Blood Advances, 2020, 4, 5232-5245.	5.2	8
65	Populationâ€specific singleâ€nucleotide polymorphism confersÂincreased risk of venous thromboembolism in African Americans. Molecular Genetics & Genomic Medicine, 2016, 4, 513-520.	1.2	7
66	METHODS TO ENSURE THE REPRODUCIBILITY OF BIOMEDICAL RESEARCH. , 2017, 22, 117-119.		5
67	Analytic and Translational Genetics. Annual Review of Biomedical Data Science, 2020, 3, 217-241.	6.5	4
68	The future of genomic medicine is here. Genome Biology, 2013, 14, 304.	9.6	3
69	Progress in genomics according to bingo: 2013 edition. Genome Biology, 2013, 14, 143.	9.6	1
70	Analysis of the Frequency of Spontaneous, Functionally-Significant Mutations in Genes Associated with Platelet Disorders in >120,000 Healthy Individuals. Blood, 2018, 132, 2438-2438.	1.4	1