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	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
2	Rare coding variants in ten genes confer substantial risk for schizophrenia. Nature, 2022, 604, 509-516.	27.8	326
3	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
4	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
5	Addendum: The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 2021, 597, E3-E4.	27.8	45
6	Human genetic analyses of organelles highlight the nucleus in age-related trait heritability. ELife, 2021, 10, .	6.0	20
7	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
8	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
9	Evaluating drug targets through human loss-of-function genetic variation. Nature, 2020, 581, 459-464.	27.8	115
10	The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 2020, 581, 434-443.	27.8	6,140
11	Characterising the loss-of-function impact of 5â€™ untranslated region variants in 15,708 individuals. Nature Communications, 2020, 11, 2523.	12.8	99
12	Landscape of multi-nucleotide variants in 125,748 human exomes and 15,708 genomes. Nature Communications, 2020, 11, 2539.	12.8	98
13	A structural variation reference for medical and population genetics. Nature, 2020, 581, 444-451.	27.8	614
14	Transcript expression-aware annotation improves rare variant interpretation. Nature, 2020, 581, 452-458.	27.8	142
15	The effect of LRRK2 loss-of-function variants in humans. Nature Medicine, 2020, 26, 869-877.	30.7	79
16	Analytic and Translational Genetics. Annual Review of Biomedical Data Science, 2020, 3, 217-241.	6.5	4
17	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
18	Integrative omics for health and disease. Nature Reviews Genetics, 2018, 19, 299-310.	16.3	676

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19	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
20	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
21	Base-specific mutational intolerance near splice sites clarifies the role of nonessential splice nucleotides. Genome Research, 2018, 28, 968-974.	5.5	41
22	Insights into the genetic epidemiology of Crohn's and rare diseases in the Ashkenazi Jewish population. PLoS Genetics, 2018, 14, e1007329.	3.5	66
23	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
24	The ExAC browser: displaying reference data information from over 60 000 exomes. Nucleic Acids Research, 2017, 45, D840-D845.	14.5	587
25	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
26	Human knockouts and phenotypic analysis in a cohort with a high rate of consanguinity. Nature, 2017, 544, 235-239.	27.8	292
27	METHODS TO ENSURE THE REPRODUCIBILITY OF BIOMEDICAL RESEARCH. , 2017, 22, 117-119.		5
28	Improving genetic diagnosis in Mendelian disease with transcriptome sequencing. Science Translational Medicine, 2017, 9, .	12.4	516
29	Using high-resolution variant frequencies to empower clinical genome interpretation. Genetics in Medicine, 2017, 19, 1151-1158.	2.4	355
30	Landscape of X chromosome inactivation across human tissues. Nature, 2017, 550, 244-248.	27.8	764
31	Using ALoFT to determine the impact of putative loss-of-function variants in protein-coding genes. Nature Communications, 2017, 8, 382.	12.8	40
32	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
33	Ten Simple Rules to Enable Multi-site Collaborations through Data Sharing. PLoS Computational Biology, 2017, 13, e1005278.	3.2	29
34	Analysis of protein-coding genetic variation in 60,706 humans. Nature, 2016, 536, 285-291.	27.8	9,051
35	Patterns of genic intolerance of rare copy number variation in 59,898 human exomes. Nature Genetics, 2016, 48, 1107-1111.	21.4	167
36	Populationâ€specific singleâ€nucleotide polymorphism confersÂincreased risk of venous thromboembolism in African Americans. Molecular Genetics & Genomic Medicine, 2016, 4, 513-520.	1.2	7

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37	Quantifying unobserved protein-coding variants in human populations provides a roadmap for large-scale sequencing projects. Nature Communications, 2016, 7, 13293.	12.8	35
38	Quantifying prion disease penetrance using large population control cohorts. Science Translational Medicine, 2016, 8, 322ra9.	12.4	289
39	Health and population effects of rare gene knockouts in adult humans with related parents. Science, 2016, 352, 474-477.	12.6	272
40	Efficient genotype compression and analysis of large genetic-variation data sets. Nature Methods, 2016, 13, 63-65.	19.0	57
41	Concept and design of a genome-wide association genotyping array tailored for transplantation-specific studies. Genome Medicine, 2015, 7, 90.	8.2	49
42	De novo mutations in congenital heart disease with neurodevelopmental and other congenital anomalies. Science, 2015, 350, 1262-1266.	12.6	646
43	Mendelian randomization of blood lipids for coronary heart disease. European Heart Journal, 2015, 36, 539-550.	2.2	567
44	Effect of predicted protein-truncating genetic variants on the human transcriptome. Science, 2015, 348, 666-669.	12.6	252
45	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
46	The UK10K project identifies rare variants in health and disease. Nature, 2015, 526, 82-90.	27.8	1,014
47	Coherent Functional Modules Improve Transcription Factor Target Identification, Cooperativity Prediction, and Disease Association. PLoS Genetics, 2014, 10, e1004122.	3.5	29
48	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
49	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
50	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
51	Genetic variant in folate homeostasis is associated with lower warfarin dose in African Americans. Blood, 2014, 124, 2298-2305.	1.4	57
52	STORMSeq: An Open-Source, User-Friendly Pipeline for Processing Personal Genomics Data in the Cloud. PLoS ONE, 2014, 9, e84860.	2.5	25
53	The future of genomic medicine is here. Genome Biology, 2013, 14, 304.	9.6	3
54	Progress in genomics according to bingo: 2013 edition. Genome Biology, 2013, 14, 143.	9.6	1

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55	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
56	A New System for Comparative Functional Genomics of Saccharomyces Yeasts. Genetics, 2013, 195, 275-287.	2.9	27
57	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
58	Evidence That Personal Genome Testing Enhances Student Learning in a Course on Genomics and Personalized Medicine. PLoS ONE, 2013, 8, e68853.	2.5	84
59	Chapter 7: Pharmacogenomics. PLoS Computational Biology, 2012, 8, e1002817.	3.2	57
60	Annotation of functional variation in personal genomes using RegulomeDB. Genome Research, 2012, 22, 1790-1797.	5.5	2,335
61	Quantifying supercoiling-induced denaturation bubbles in DNA. Soft Matter, 2012, 8, 8651.	2.7	50
62	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
63	Personal Omics Profiling Reveals Dynamic Molecular and Medical Phenotypes. Cell, 2012, 148, 1293-1307.	28.9	1,134
64	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
65	Bioinformatics challenges for personalized medicine. Bioinformatics, 2011, 27, 1741-1748.	4.1	223
66	Regulatory Variation Within and Between Species. Annual Review of Genomics and Human Genetics, 2011, 12, 327-346.	6.2	70
67	Performance comparison of exome DNA sequencing technologies. Nature Biotechnology, 2011, 29, 908-914.	17.5	464
68	Phased Whole-Genome Genetic Risk in a Family Quartet Using a Major Allele Reference Sequence. PLoS Genetics, 2011, 7, e1002280.	3.5	137
69	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
70	Variation in Transcription Factor Binding Among Humans. Science, 2010, 328, 232-235.	12.6	521