Robert E Handsaker

List of Publications by Year
in descending order

49 papers	112,287 citations	61984 43 h-index	168389 53 g-index
65 all docs	65 docs citations	65 times ranked	146508 citing authors

#	Article	IF	Citations
1	The Sequence Alignment/Map format and SAMtools. Bioinformatics, 2009, 25, 2078-2079.	4.1	49,124
2	A global reference for human genetic variation. Nature, 2015, 526, 68-74.	27.8	13,998
3	The variant call format and VCFtools. Bioinformatics, 2011, 27, 2156-2158.	4.1	11,326
4	A map of human genome variation from population-scale sequencing. Nature, 2010, 467, 1061-1073.	27.8	7,209
5	An integrated map of genetic variation from 1,092 human genomes. Nature, 2012, 491, 56-65.	27.8	7,199
6	Genetic studies of body mass index yield new insights for obesity biology. Nature, 2015, 518, 197-206.	27.8	3,823
7	Clonal Hematopoiesis and Blood-Cancer Risk Inferred from Blood DNA Sequence. New England Journal of Medicine, 2014, 371, 2477-2487.	27.0	2,669
8	Genome-Wide Association Analysis Identifies Loci for Type 2 Diabetes and Triglyceride Levels. Science, 2007, 316, 1331-1336.	12.6	2,623
9	An integrated map of structural variation in 2,504 human genomes. Nature, 2015, 526, 75-81.	27.8	1,994
10	Schizophrenia risk from complex variation of complement component 4. Nature, 2016, 530, 177-183.	27.8	1,915
11	Genome sequence and analysis of the Irish potato famine pathogen Phytophthora infestans. Nature, 2009, 461, 393-398.	27.8	1,405
12	A Systematic Survey of Loss-of-Function Variants in Human Protein-Coding Genes. Science, 2012, 335, 823-828.	12.6	1,095
13	Mapping copy number variation by population-scale genome sequencing. Nature, 2011, 470, 59-65.	27.8	991
14	Integrated detection and population-genetic analysis of SNPs and copy number variation. Nature Genetics, 2008, 40, 1166-1174.	21.4	838
15	Multi-platform discovery of haplotype-resolved structural variation in human genomes. Nature Communications, 2019, 10, 1784.	12.8	636
16	Discovery and Statistical Genotyping of Copy-Number Variation from Whole-Exome Sequencing Depth. American Journal of Human Genetics, 2012, 91, 597-607.	6.2	513
17	Human pluripotent stem cells recurrently acquire and expand dominant negative P53 mutations. Nature, 2017, 545, 229-233.	27.8	409
18	Pathways Disrupted in Human ALS Motor Neurons Identified through Genetic Correction of Mutant SOD1. Cell Stem Cell, 2014, 14, 781-795.	11.1	392

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19	Large multiallelic copy number variations in humans. Nature Genetics, 2015, 47, 296-303.	21.4	357
20	Discovery and genotyping of genome structural polymorphism by sequencing on a population scale. Nature Genetics, 2011, 43, 269-276.	21.4	299
21	Identification of heart rateâ€"associated loci and their effects on cardiac conduction and rhythm disorders. Nature Genetics, 2013, 45, 621-631.	21.4	282
22	Insights into clonal haematopoiesis from 8,342 mosaic chromosomal alterations. Nature, 2018, 559, 350-355.	27.8	279
23	Punctuated bursts in human male demography inferred from 1,244 worldwide Y-chromosome sequences. Nature Genetics, 2016, 48, 593-599.	21.4	273
24	Mutations causing medullary cystic kidney disease type 1 lie in a large VNTR in MUC1 missed by massively parallel sequencing. Nature Genetics, 2013, 45, 299-303.	21.4	237
25	An analytical framework for whole-genome sequence association studies and its implications for autism spectrum disorder. Nature Genetics, 2018, 50, 727-736.	21.4	235
26	The Genetic Landscape of Diamond-Blackfan Anemia. American Journal of Human Genetics, 2018, 103, 930-947.	6.2	184
27	Complement genes contribute sex-biased vulnerability in diverse disorders. Nature, 2020, 582, 577-581.	27.8	158
28	Genetic Variation in Human DNA Replication Timing. Cell, 2014, 159, 1015-1026.	28.9	149
29	Structural haplotypes and recent evolution of the human 17q21.31 region. Nature Genetics, 2012, 44, 881-885.	21.4	124
30	Whole genome sequencing in psychiatric disorders: the WGSPD consortium. Nature Neuroscience, 2017, 20, 1661-1668.	14.8	122
31	Structural forms of the human amylase locus and their relationships to SNPs, haplotypes and obesity. Nature Genetics, 2015, 47, 921-925.	21.4	120
32	Characteristics of de novo structural changes in the human genome. Genome Research, 2015, 25, 792-801.	5.5	115
33	Negative selection in humans and fruit flies involves synergistic epistasis. Science, 2017, 356, 539-542.	12.6	103
34	An international effort towards developing standards for best practices in analysis, interpretation and reporting of clinical genome sequencing results in the CLARITY Challenge. Genome Biology, 2014, 15, R53.	9.6	101
35	Protein-coding repeat polymorphisms strongly shape diverse human phenotypes. Science, 2021, 373, 1499-1505.	12.6	96
36	Population-scale tissue transcriptomics maps long non-coding RNAs to complex disease. Cell, 2021, 184, 2633-2648.e19.	28.9	94

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37	Recurring exon deletions in the HP (haptoglobin) gene contribute to lower blood cholesterol levels. Nature Genetics, 2016, 48, 359-366.	21.4	93
38	Ultra-rare disruptive and damaging mutations influence educational attainment in the general population. Nature Neuroscience, 2016, 19, 1563-1565.	14.8	90
39	Deep coverage whole genome sequences and plasma lipoprotein(a) in individuals of European and African ancestries. Nature Communications, 2018, 9, 2606.	12.8	79
40	A whole-genome sequence study identifies genetic risk factors for neuromyelitis optica. Nature Communications, 2018, 9, 1929.	12.8	73
41	Human genomic regions with exceptionally high levels of population differentiation identified from 911 whole-genome sequences. Genome Biology, 2014, 15, R88.	9.6	72
42	Using population admixture to help complete maps of the human genome. Nature Genetics, 2013, 45, 406-414.	21.4	61
43	Genome-wide association analysis of red blood cell traits in African Americans: the COGENT Network. Human Molecular Genetics, 2013, 22, 2529-2538.	2.9	57
44	Common Î±-globin variants modify hematologic and other clinical phenotypes in sickle cell trait and disease. PLoS Genetics, 2018, 14, e1007293.	3.5	45
45	Mapping the Human Reference Genomeâ€™s Missing Sequence by Three-Way Admixture in Latino Genomes. American Journal of Human Genetics, 2013, 93, 411-421.	6.2	36
46	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
47	The genetic architecture of DNA replication timing in human pluripotent stem cells. Nature Communications, 2021, 12, 6746.	12.8	26
48	GenomeVIP: a cloud platform for genomic variant discovery and interpretation. Genome Research, 2017, 27, 1450-1459.	5.5	15
49	Chromosomal phase improves aneuploidy detection in non-invasive prenatal testing at low fetal DNA fractions. Scientific Reports, 2022, 12, .	3.3	1