

# Robert E Handsaker

## List of Publications by Year in descending order

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Version: 2024-02-01

49  
papers

112,287  
citations

61945

43  
h-index

168321

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. <i>Bioinformatics</i> , 2009, 25, 2078-2079.	1.8	49,124
2	A global reference for human genetic variation. <i>Nature</i> , 2015, 526, 68-74.	13.7	13,998
3	The variant call format and VCFtools. <i>Bioinformatics</i> , 2011, 27, 2156-2158.	1.8	11,326
4	A map of human genome variation from population-scale sequencing. <i>Nature</i> , 2010, 467, 1061-1073.	13.7	7,209
5	An integrated map of genetic variation from 1,092 human genomes. <i>Nature</i> , 2012, 491, 56-65.	13.7	7,199
6	Genetic studies of body mass index yield new insights for obesity biology. <i>Nature</i> , 2015, 518, 197-206.	13.7	3,823
7	Clonal Hematopoiesis and Blood-Cancer Risk Inferred from Blood DNA Sequence. <i>New England Journal of Medicine</i> , 2014, 371, 2477-2487.	13.9	2,669
8	Genome-Wide Association Analysis Identifies Loci for Type 2 Diabetes and Triglyceride Levels. <i>Science</i> , 2007, 316, 1331-1336.	6.0	2,623
9	An integrated map of structural variation in 2,504 human genomes. <i>Nature</i> , 2015, 526, 75-81.	13.7	1,994
10	Schizophrenia risk from complex variation of complement component 4. <i>Nature</i> , 2016, 530, 177-183.	13.7	1,915
11	Genome sequence and analysis of the Irish potato famine pathogen <i>Phytophthora infestans</i> . <i>Nature</i> , 2009, 461, 393-398.	13.7	1,405
12	A Systematic Survey of Loss-of-Function Variants in Human Protein-Coding Genes. <i>Science</i> , 2012, 335, 823-828.	6.0	1,095
13	Mapping copy number variation by population-scale genome sequencing. <i>Nature</i> , 2011, 470, 59-65.	13.7	991
14	Integrated detection and population-genetic analysis of SNPs and copy number variation. <i>Nature Genetics</i> , 2008, 40, 1166-1174.	9.4	838
15	Multi-platform discovery of haplotype-resolved structural variation in human genomes. <i>Nature Communications</i> , 2019, 10, 1784.	5.8	636
16	Discovery and Statistical Genotyping of Copy-Number Variation from Whole-Exome Sequencing Depth. <i>American Journal of Human Genetics</i> , 2012, 91, 597-607.	2.6	513
17	Human pluripotent stem cells recurrently acquire and expand dominant negative P53 mutations. <i>Nature</i> , 2017, 545, 229-233.	13.7	409
18	Pathways Disrupted in Human ALS Motor Neurons Identified through Genetic Correction of Mutant SOD1. <i>Cell Stem Cell</i> , 2014, 14, 781-795.	5.2	392

#	ARTICLE	IF	CITATIONS
19	Large multiallelic copy number variations in humans. <i>Nature Genetics</i> , 2015, 47, 296-303.	9.4	357
20	Discovery and genotyping of genome structural polymorphism by sequencing on a population scale. <i>Nature Genetics</i> , 2011, 43, 269-276.	9.4	299
21	Identification of heart rate-associated loci and their effects on cardiac conduction and rhythm disorders. <i>Nature Genetics</i> , 2013, 45, 621-631.	9.4	282
22	Insights into clonal haematopoiesis from 8,342 mosaic chromosomal alterations. <i>Nature</i> , 2018, 559, 350-355.	13.7	279
23	Punctuated bursts in human male demography inferred from 1,244 worldwide Y-chromosome sequences. <i>Nature Genetics</i> , 2016, 48, 593-599.	9.4	273
24	Mutations causing medullary cystic kidney disease type 1 lie in a large VNTR in MUC1 missed by massively parallel sequencing. <i>Nature Genetics</i> , 2013, 45, 299-303.	9.4	237
25	An analytical framework for whole-genome sequence association studies and its implications for autism spectrum disorder. <i>Nature Genetics</i> , 2018, 50, 727-736.	9.4	235
26	The Genetic Landscape of Diamond-Blackfan Anemia. <i>American Journal of Human Genetics</i> , 2018, 103, 930-947.	2.6	184
27	Complement genes contribute sex-biased vulnerability in diverse disorders. <i>Nature</i> , 2020, 582, 577-581.	13.7	158
28	Genetic Variation in Human DNA Replication Timing. <i>Cell</i> , 2014, 159, 1015-1026.	13.5	149
29	Structural haplotypes and recent evolution of the human 17q21.31 region. <i>Nature Genetics</i> , 2012, 44, 881-885.	9.4	124
30	Whole genome sequencing in psychiatric disorders: the WGSPD consortium. <i>Nature Neuroscience</i> , 2017, 20, 1661-1668.	7.1	122
31	Structural forms of the human amylase locus and their relationships to SNPs, haplotypes and obesity. <i>Nature Genetics</i> , 2015, 47, 921-925.	9.4	120
32	Characteristics of de novo structural changes in the human genome. <i>Genome Research</i> , 2015, 25, 792-801.	2.4	115
33	Negative selection in humans and fruit flies involves synergistic epistasis. <i>Science</i> , 2017, 356, 539-542.	6.0	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. <i>Genome Biology</i> , 2014, 15, R53.	13.9	101
35	Protein-coding repeat polymorphisms strongly shape diverse human phenotypes. <i>Science</i> , 2021, 373, 1499-1505.	6.0	96
36	Population-scale tissue transcriptomics maps long non-coding RNAs to complex disease. <i>Cell</i> , 2021, 184, 2633-2648.e19.	13.5	94

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37	Recurring exon deletions in the HP (haptoglobin) gene contribute to lower blood cholesterol levels. <i>Nature Genetics</i> , 2016, 48, 359-366.	9.4	93
38	Ultra-rare disruptive and damaging mutations influence educational attainment in the general population. <i>Nature Neuroscience</i> , 2016, 19, 1563-1565.	7.1	90
39	Deep coverage whole genome sequences and plasma lipoprotein(a) in individuals of European and African ancestries. <i>Nature Communications</i> , 2018, 9, 2606.	5.8	79
40	A whole-genome sequence study identifies genetic risk factors for neuromyelitis optica. <i>Nature Communications</i> , 2018, 9, 1929.	5.8	73
41	Human genomic regions with exceptionally high levels of population differentiation identified from 911 whole-genome sequences. <i>Genome Biology</i> , 2014, 15, R88.	13.9	72
42	Using population admixture to help complete maps of the human genome. <i>Nature Genetics</i> , 2013, 45, 406-414.	9.4	61
43	Genome-wide association analysis of red blood cell traits in African Americans: the COGENT Network. <i>Human Molecular Genetics</i> , 2013, 22, 2529-2538.	1.4	57
44	Common $\beta$ -globin variants modify hematologic and other clinical phenotypes in sickle cell trait and disease. <i>PLoS Genetics</i> , 2018, 14, e1007293.	1.5	45
45	Mapping the Human Reference Genome's Missing Sequence by Three-Way Admixture in Latino Genomes. <i>American Journal of Human Genetics</i> , 2013, 93, 411-421.	2.6	36
46	Whole-genome analysis of human embryonic stem cells enables rational line selection based on genetic variation. <i>Cell Stem Cell</i> , 2022, 29, 472-486.e7.	5.2	27
47	The genetic architecture of DNA replication timing in human pluripotent stem cells. <i>Nature Communications</i> , 2021, 12, 6746.	5.8	26
48	GenomeVIP: a cloud platform for genomic variant discovery and interpretation. <i>Genome Research</i> , 2017, 27, 1450-1459.	2.4	15
49	Chromosomal phase improves aneuploidy detection in non-invasive prenatal testing at low fetal DNA fractions. <i>Scientific Reports</i> , 2022, 12, .	1.6	1