Robert E Handsaker

List of Publications by Year
in descending order

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	Whole-genome analysis of human embryonic stem cells enables rational line selection based on genetic variation. Cell Stem Cell, 2022, 29, 472-486.e7.	5.2	27
2	Chromosomal phase improves aneuploidy detection in non-invasive prenatal testing at low fetal DNA fractions. Scientific Reports, 2022, 12, .	1.6	1
3	Population-scale tissue transcriptomics maps long non-coding RNAs to complex disease. Cell, 2021, 184, 2633-2648.e19.	13.5	94
4	Protein-coding repeat polymorphisms strongly shape diverse human phenotypes. Science, 2021, 373, 1499-1505.	6.0	96
5	The genetic architecture of DNA replication timing in human pluripotent stem cells. Nature Communications, 2021, 12, 6746.	5.8	26
6	Complement genes contribute sex-biased vulnerability in diverse disorders. Nature, 2020, 582, 577-581.	13.7	158
7	Multi-platform discovery of haplotype-resolved structural variation in human genomes. Nature Communications, 2019, 10, 1784.	5.8	636
8	An analytical framework for whole-genome sequence association studies and its implications for autism spectrum disorder. Nature Genetics, 2018, 50, 727-736.	9.4	235
9	The Genetic Landscape of Diamond-Blackfan Anemia. American Journal of Human Genetics, 2018, 103, 930-947.	2.6	184
10	A whole-genome sequence study identifies genetic risk factors for neuromyelitis optica. Nature Communications, 2018, 9, 1929.	5.8	73
11	Deep coverage whole genome sequences and plasma lipoprotein(a) in individuals of European and African ancestries. Nature Communications, 2018, 9, 2606.	5.8	79
12	Insights into clonal haematopoiesis from 8,342 mosaic chromosomal alterations. Nature, 2018, 559, 350-355.	13.7	279
13	Common Î±-globin variants modify hematologic and other clinical phenotypes in sickle cell trait and disease. PLoS Genetics, 2018, 14, e1007293.	1.5	45
14	Negative selection in humans and fruit flies involves synergistic epistasis. Science, 2017, 356, 539-542.	6.0	103
15	Human pluripotent stem cells recurrently acquire and expand dominant negative P53 mutations. Nature, 2017, 545, 229-233.	13.7	409
16	GenomeVIP: a cloud platform for genomic variant discovery and interpretation. Genome Research, 2017, 27, 1450-1459.	2.4	15
17	Whole genome sequencing in psychiatric disorders: the WGSPD consortium. Nature Neuroscience, 2017, 20, 1661-1668.	7.1	122
18	Punctuated bursts in human male demography inferred from 1,244 worldwide Y-chromosome sequences. Nature Genetics, 2016, 48, 593-599.	9.4	273

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19	Ultra-rare disruptive and damaging mutations influence educational attainment in the general population. Nature Neuroscience, 2016, 19, 1563-1565.	7.1	90
20	Schizophrenia risk from complex variation of complement component 4. Nature, 2016, 530, 177-183.	13.7	1,915
21	Recurring exon deletions in the HP (haptoglobin) gene contribute to lower blood cholesterol levels. Nature Genetics, 2016, 48, 359-366.	9.4	93
22	Genetic studies of body mass index yield new insights for obesity biology. Nature, 2015, 518, 197-206.	13.7	3,823
23	Large multiallelic copy number variations in humans. Nature Genetics, 2015, 47, 296-303.	9.4	357
24	Structural forms of the human amylase locus and their relationships to SNPs, haplotypes and obesity. Nature Genetics, 2015, 47, 921-925.	9.4	120
25	Characteristics of de novo structural changes in the human genome. Genome Research, 2015, 25, 792-801.	2.4	115
26	A global reference for human genetic variation. Nature, 2015, 526, 68-74.	13.7	13,998
27	An integrated map of structural variation in 2,504 human genomes. Nature, 2015, 526, 75-81.	13.7	1,994
28	Clonal Hematopoiesis and Blood-Cancer Risk Inferred from Blood DNA Sequence. New England Journal of Medicine, 2014, 371, 2477-2487.	13.9	2,669
29	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.	13.9	101
30	Pathways Disrupted in Human ALS Motor Neurons Identified through Genetic Correction of Mutant SOD1. Cell Stem Cell, 2014, 14, 781-795.	5.2	392
31	Genetic Variation in Human DNA Replication Timing. Cell, 2014, 159, 1015-1026.	13.5	149
32	Human genomic regions with exceptionally high levels of population differentiation identified from 911 whole-genome sequences. Genome Biology, 2014, 15, R88.	13.9	72
33	Mapping the Human Reference Genomeâ€™s Missing Sequence by Three-Way Admixture in Latino Genomes. American Journal of Human Genetics, 2013, 93, 411-421.	2.6	36
34	Using population admixture to help complete maps of the human genome. Nature Genetics, 2013, 45, 406-414.	9.4	61
35	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.	9.4	237
36	Identification of heart rateâ€“associated loci and their effects on cardiac conduction and rhythm disorders. Nature Genetics, 2013, 45, 621-631.	9.4	282

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37	Genome-wide association analysis of red blood cell traits in African Americans: the COGENT Network. Human Molecular Genetics, 2013, 22, 2529-2538.	1.4	57
38	An integrated map of genetic variation from 1,092 human genomes. Nature, 2012, 491, 56-65.	13.7	7,199
39	Discovery and Statistical Genotyping of Copy-Number Variation from Whole-Exome Sequencing Depth. American Journal of Human Genetics, 2012, 91, 597-607.	2.6	513
40	Structural haplotypes and recent evolution of the human 17q21.31 region. Nature Genetics, 2012, 44, 881-885.	9.4	124
41	A Systematic Survey of Loss-of-Function Variants in Human Protein-Coding Genes. Science, 2012, 335, 823-828.	6.0	1,095
42	The variant call format and VCFtools. Bioinformatics, 2011, 27, 2156-2158.	1.8	11,326
43	Discovery and genotyping of genome structural polymorphism by sequencing on a population scale. Nature Genetics, 2011, 43, 269-276.	9.4	299
44	Mapping copy number variation by population-scale genome sequencing. Nature, 2011, 470, 59-65.	13.7	991
45	A map of human genome variation from population-scale sequencing. Nature, 2010, 467, 1061-1073.	13.7	7,209
46	Genome sequence and analysis of the Irish potato famine pathogen Phytophthora infestans. Nature, 2009, 461, 393-398.	13.7	1,405
47	The Sequence Alignment/Map format and SAMtools. Bioinformatics, 2009, 25, 2078-2079.	1.8	49,124
48	Integrated detection and population-genetic analysis of SNPs and copy number variation. Nature Genetics, 2008, 40, 1166-1174.	9.4	838
49	Genome-Wide Association Analysis Identifies Loci for Type 2 Diabetes and Triglyceride Levels. Science, 2007, 316, 1331-1336.	6.0	2,623