Shamil R Sunyaev

List of Publications by Year
in descending order

134	55,705	12330	11939
papers	citations	69 h-index	134 g-index

161	161	161	82020
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	A method and server for predicting damaging missense mutations. Nature Methods, 2010, 7, 248-249.	19.0	11,491
2	Integrative analysis of 111 reference human epigenomes. Nature, 2015, 518, 317-330.	27.8	5,653
3	Mutational heterogeneity in cancer and the search for new cancer-associated genes. Nature, 2013, 499, 214-218.	27.8	4,761
4	Identification and analysis of functional elements in 1% of the human genome by the ENCODE pilot project. Nature, 2007, 447, 799-816.	27.8	4,709
5	Systematic Localization of Common Disease-Associated Variation in Regulatory DNA. Science, 2012, 337, 1190-1195.	12.6	3,129
6	The accessible chromatin landscape of the human genome. Nature, 2012, 489, 75-82.	27.8	2,434
7	Human non-synonymous SNPs: server and survey. Nucleic Acids Research, 2002, 30, 3894-3900.	14.5	1,995
8	Patterns and rates of exonic de novo mutations in autism spectrum disorders. Nature, 2012, 485, 242-245.	27.8	1,597
9	Evolution and Functional Impact of Rare Coding Variation from Deep Sequencing of Human Exomes. Science, 2012, 337, 64-69.	12.6	1,535
10	The mystery of missing heritability: Genetic interactions create phantom heritability. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, 1193-1198.	7.1	1,322
11	Genomic variation landscape of the human gut microbiome. Nature, 2013, 493, 45-50.	27.8	783
12	Pooled Association Tests for Rare Variants in Exon-Resequencing Studies. American Journal of Human Genetics, 2010, 86, 832-838.	6.2	715
13	Assessing the Evolutionary Impact of Amino Acid Mutations in the Human Genome. PLoS Genetics, 2008, 4, e1000083.	3.5	586
14	Exome sequencing identifies rare LDLR and APOA5 alleles conferring risk for myocardial infarction. Nature, 2015, 518, 102-106.	27.8	581
15	Searching for missing heritability: Designing rare variant association studies. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, E455-64.	7.1	570
16	Charting the Proteomes of Organisms with Unsequenced Genomes by MALDI-Quadrupole Time-of-Flight Mass Spectrometry and BLAST Homology Searching. Analytical Chemistry, 2001, 73, 1917-1926.	6.5	569
17	Most Rare Missense Alleles Are Deleterious in Humans: Implications for Complex Disease and Association Studies. American Journal of Human Genetics, 2007, 80, 727-739.	6.2	547
18	Genome sequencing reveals insights into physiology and longevity of the naked mole rat. Nature, 2011, 479, 223-227.	27.8	517

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19	Cell-of-origin chromatin organization shapes the mutational landscape of cancer. Nature, 2015, 518, 360-364.	27.8	491
20	Widespread Macromolecular Interaction Perturbations in Human Genetic Disorders. Cell, 2015, 161, 647-660.	28.9	482
21	Genome-wide patterns and properties of de novo mutations in humans. Nature Genetics, 2015, 47, 822-826.	21.4	384
22	Human mutation rate associated with DNA replication timing. Nature Genetics, 2009, 41, 393-395.	21.4	371
23	Proportionally more deleterious genetic variation in European than in African populations. Nature, 2008, 451, 994-997.	27.8	365
24	Exome sequencing and the genetic basis of complex traits. Nature Genetics, 2012, 44, 623-630.	21.4	340
25	Towards a structural basis of human non-synonymous single nucleotide polymorphisms. Trends in Genetics, 2000, 16, 198-200.	6.7	333
26	Shifting Paradigm of Association Studies: Value of Rare Single-Nucleotide Polymorphisms. American Journal of Human Genetics, 2008, 82, 100-112.	6.2	292
27	A universal trend of amino acid gain and loss in protein evolution. Nature, 2005, 433, 633-638.	27.8	282
28	Dobzhansky-Muller incompatibilities in protein evolution. Proceedings of the National Academy of Sciences of the United States of America, 2002, 99, 14878-14883.	7.1	281
29	Polygenic adaptation on height is overestimated due to uncorrected stratification in genome-wide association studies. ELife, 2019, 8, .	6.0	276
30	Impact of deleterious passenger mutations on cancer progression. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 2910-2915.	7.1	274
31	Extremely low-coverage sequencing and imputation increases power for genome-wide association studies. Nature Genetics, 2012, 44, 631-635.	21.4	239
32	Sequencing studies in human genetics: design and interpretation. Nature Reviews Genetics, 2013, 14, 460-470.	16.3	236
33	Differential Relationship of DNA Replication Timing to Different Forms of Human Mutation and Variation. American Journal of Human Genetics, 2012, 91, 1033-1040.	6.2	220
34	Increase of functional diversity by alternative splicing. Trends in Genetics, 2003, 19, 124-128.	6.7	208
35	Limited statistical evidence for shared genetic effects of eQTLs and autoimmune-disease-associated loci in three major immune-cell types. Nature Genetics, 2017, 49, 600-605.	21.4	205
36	Medical Sequencing at the Extremes of Human Body Mass. American Journal of Human Genetics, 2007, 80, 779-791.	6.2	199

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37	Gain-of-function mutations in the mechanically activated ion channel PIEZO2 cause a subtype of Distal Arthrogryposis. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 4667-4672.	7.1	193
38	The interface of protein structure, protein biophysics, and molecular evolution. Protein Science, 2012, 21, 769-785.	7.6	188
39	No evidence that selection has been less effective at removing deleterious mutations in Europeans than in Africans. Nature Genetics, 2015, 47, 126-131.	21.4	182
40	Identification of cancer driver genes based on nucleotide context. Nature Genetics, 2020, 52, 208-218.	21.4	170
41	Widespread non-additive and interaction effects within HLA loci modulate the risk of autoimmune diseases. Nature Genetics, 2015, 47, 1085-1090.	21.4	164
42	A Model for Statistical Significance of Local Similarities in Structure. Journal of Molecular Biology, 2003, 326, 1307-1316.	4.2	161
43	Estimating the selective effects of heterozygous protein-truncating variants from human exome data. Nature Genetics, 2017, 49, 806-810.	21.4	157
44	Large-Scale Identification and Evolution Indexing of Tyrosine Phosphorylation Sites from Murine Brain. Journal of Proteome Research, 2008, 7, 311-318.	3.7	153
45	Power of deep, all-exon resequencing for discovery of human trait genes. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 3871-3876.	7.1	147
46	Evaluating empirical bounds on complex disease genetic architecture. Nature Genetics, 2013, 45, 1418-1427.	21.4	147
47	Dynamic incorporation of multiple in silico functional annotations empowers rare variant association analysis of large whole-genome sequencing studies at scale. Nature Genetics, 2020, 52, 969-983.	21.4	146
48	The Power and the Limitations of Cross-Species Protein Identification by Mass Spectrometry-driven Sequence Similarity Searches. Molecular and Cellular Proteomics, 2004, 3, 238-249.	3.8	143
49	Clinical Sequencing Exploratory Research Consortium: Accelerating Evidence-Based Practice of Genomic Medicine. American Journal of Human Genetics, 2016, 98, 1051-1066.	6.2	137
50	Progressive Multifocal Leukoencephalopathy (PML) Development Is Associated With Mutations in JC Virus Capsid Protein VP1 That Change Its Receptor Specificity. Journal of Infectious Diseases, 2011, 204, 103-114.	4.0	135
51	Metabolites: a helping hand for pathway evolution?. Trends in Biochemical Sciences, 2003, 28, 336-341.	7.5	133
52	Functional architecture of low-frequency variants highlights strength of negative selection across coding and non-coding annotations. Nature Genetics, 2018, 50, 1600-1607.	21.4	132
53	Mutation mapping and identification by whole-genome sequencing. Genome Research, 2012, 22, 1541-1548.	5.5	126
54	Reduced local mutation density in regulatory DNA of cancer genomes is linked to DNA repair. Nature Biotechnology, 2014, 32, 71-75.	17.5	120

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55	MultiTag:Â Multiple Error-Tolerant Sequence Tag Search for the Sequence-Similarity Identification of Proteins by Mass Spectrometry. Analytical Chemistry, 2003, 75, 1307-1315.	6.5	118
56	Computational and statistical approaches to analyzing variants identified by exome sequencing. Genome Biology, 2011, 12, 227.	9.6	116
57	Adaptive Mutations in the JC Virus Protein Capsid Are Associated with Progressive Multifocal Leukoencephalopathy (PML). PLoS Genetics, 2009, 5, e1000368.	3.5	114
58	Homology-based fold predictions for Mycoplasma genitalium proteins 1 1Edited by G. Von Heijne. Journal of Molecular Biology, 1998, 280, 323-326.	4.2	112
59	GWAS for quantitative resistance phenotypes in Mycobacterium tuberculosis reveals resistance genes and regulatory regions. Nature Communications, 2019, 10, 2128.	12.8	111
60	Genetic Associations with Obstructive Sleep Apnea Traits in Hispanic/Latino Americans. American Journal of Respiratory and Critical Care Medicine, 2016, 194, 886-897.	5.6	107
61	Identification of cis-suppression of human disease mutations by comparative genomics. Nature, 2015, 524, 225-229.	27.8	106
62	A limited role for balancing selection. Trends in Genetics, 2005, 21, 30-32.	6.7	103
63	Negative selection in humans and fruit flies involves synergistic epistasis. Science, 2017, 356, 539-542.	12.6	103
64	Sequence similarityâ€driven proteomics in organisms with unknown genomes by LCâ€MS/MS and automated de novo sequencing. Proteomics, 2007, 7, 2318-2329.	2.2	98
65	Quantification of frequency-dependent genetic architectures in 25 UK Biobank traits reveals action of negative selection. Nature Communications, 2019, 10, 790.	12.8	98
66	Bayesian inference of negative and positive selection in human cancers. Nature Genetics, 2017, 49, 1785-1788.	21.4	90
67	Widely distributed noncoding purifying selection in the human genome. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 12410-12415.	7.1	84
68	Rare, Low-Frequency, and Common Variants in the Protein-Coding Sequence of Biological Candidate Genes from GWASs Contribute to Risk of Rheumatoid Arthritis. American Journal of Human Genetics, 2013, 92, 15-27.	6.2	83
69	Genes with monoallelic expression contribute disproportionately to genetic diversity in humans. Nature Genetics, 2016, 48, 231-237.	21.4	83
70	Calibration of Multiple In Silico Tools for Predicting Pathogenicity of Mismatch Repair Gene Missense Substitutions. Human Mutation, 2013, 34, 255-265.	2.5	80
71	APOBEC-Induced Cancer Mutations Are Uniquely Enriched in Early-Replicating, Gene-Dense, and Active Chromatin Regions. Cell Reports, 2015, 13, 1103-1109.	6.4	80
72	SNP frequencies in human genes. Trends in Genetics, 2000, 16, 335-337.	6.7	78

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73	Dominance of Deleterious Alleles Controls the Response to a Population Bottleneck. PLoS Genetics, 2015, 11, e1005436.	3.5	78
74	Leveraging Distant Relatedness to Quantify Human Mutation and Gene-Conversion Rates. American Journal of Human Genetics, 2015, 97, 775-789.	6.2	77
75	Protein identification pipeline for the homology-driven proteomics. Journal of Proteomics, 2008, 71, 346-356.	2.4	76
76	Small fitness effect of mutations in highly conserved non-coding regions. Human Molecular Genetics, 2005, 14, 2221-2229.	2.9	74
77	Development and Validation of a Computational Method for Assessment of Missense Variants in Hypertrophic Cardiomyopathy. American Journal of Human Genetics, 2011, 88, 183-192.	6.2	73
78	Positive selection at sites of multiple amino acid replacements since ratâ€“mouse divergence. Nature, 2004, 429, 558-562.	27.8	70
79	Automated typing of red blood cell and platelet antigens: a whole-genome sequencing study. Lancet Haematology,the, 2018, 5, e241-e251.	4.6	70
80	Population-specific causal disease effect sizes in functionally important regions impacted by selection. Nature Communications, 2021, 12, 1098.	12.8	68
81	Homology-based functional proteomics by mass spectrometry: Application to theXenopus microtubule-associated proteome. Proteomics, 2004, 4, 2707-2721.	2.2	67
82	Analysis of Sequence Conservation at Nucleotide Resolution. PLoS Computational Biology, 2007, 3, e254.	3.2	65
83	Multiethnic Meta-Analysis Identifies RAI1 as a Possible Obstructive Sleep Apneaâ€“related Quantitative Trait Locus in Men. American Journal of Respiratory Cell and Molecular Biology, 2018, 58, 391-401.	2.9	65
84	Human allelic variation: perspective from protein function, structure, and evolution. Current Opinion in Structural Biology, 2010, 20, 342-350.	5.7	63
85	Deleterious Alleles in the Human Genome Are on Average Younger Than Neutral Alleles of the Same Frequency. PLoS Genetics, 2013, 9, e1003301.	3.5	63
86	Multiplex padlock targeted sequencing reveals human hypermutable CpG variations. Genome Research, 2009, 19, 1606-1615.	5.5	62
87	Excess of Deleterious Mutations around HLA Genes Reveals Evolutionary Cost of Balancing Selection. Molecular Biology and Evolution, 2016, 33, 2555-2564.	8.9	55
88	Evolutionary constraints in conserved nongenic sequences of mammals. Genome Research, 2005, 15, 1373-1378.	5.5	50
89	Indel-Based Evolutionary Distance and Mouse-Human Divergence. Genome Research, 2004, 14, 1610-1616.	5.5	48
90	Inferring causality and functional significance of human coding DNA variants. Human Molecular Genetics, 2012, 21, R10-R17.	2.9	47

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91	Mutations in RABL3 alter KRAS prenylation and are associated with hereditary pancreatic cancer. Nature Genetics, 2019, 51, 1308-1314.	21.4	47
92	Population sequencing data reveal a compendium of mutational processes in the human germ line. Science, 2021, 373, 1030-1035.	12.6	43
93	Prediction of nonsynonymous single nucleotide polymorphisms in human disease-associated genes. Journal of Molecular Medicine, 1999, 77, 754-760.	3.9	42
94	Admixture mapping identifies novel loci for obstructive sleep apnea in Hispanic/Latino Americans. Human Molecular Genetics, 2019, 28, 675-687.	2.9	41
95	Common Single-Nucleotide Polymorphisms Act in Concert to Affect Plasma Levels of High-Density Lipoprotein Cholesterol. American Journal of Human Genetics, 2007, 81, 1298-1303.	6.2	38
96	Separating the Wheat from the Chaff: Unbiased Filtering of Background Tandem Mass Spectra Improves Protein Identification. Journal of Proteome Research, 2008, 7, 3382-3395.	3.7	37
97	Triplet repeat length bias and variation in the human transcriptome. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 17095-17100.	7.1	33
98	Genome-wide analysis of somatic noncoding mutation patterns in cancer. Science, 2022, 376, eabg5601.	12.6	33
99	Impact of selection, mutation rate and genetic drift on human genetic variation. Human Molecular Genetics, 2003, 12, 3325-3330.	2.9	32
100	Hypermutable Non-Synonymous Sites Are under Stronger Negative Selection. PLoS Genetics, 2008, 4, e1000281.	3.5	32
101	Non-parametric Polygenic Risk Prediction via Partitioned GWAS Summary Statistics. American Journal of Human Genetics, 2020, 107, 46-59.	6.2	30
102	StrVCTVRE: A supervised learning method to predict the pathogenicity of human genome structural variants. American Journal of Human Genetics, 2022, 109, 195-209.	6.2	29
103	PINES: phenotype-informed tissue weighting improves prediction of pathogenic noncoding variants. Genome Biology, 2018, 19, 173.	8.8	28
104	Associations of variants In the hexokinase 1 and interleukin 18 receptor regions with oxyhemoglobin saturation during sleep. PLoS Genetics, 2019, 15, e1007739.	3.5	28
105	Error-prone bypass of DNA lesions during lagging-strand replication is a common source of germline and cancer mutations. Nature Genetics, 2019, 51, 36-41.	21.4	28
106	Applicability of the Mutationâ€“Selection Balance Model to Population Genetics of Heterozygous Protein-Truncating Variants in Humans. Molecular Biology and Evolution, 2019, 36, 1701-1710.	8.9	27
107	Fine-Scale Haplotype Structure Reveals Strong Signatures of Positive Selection in a Recombining Bacterial Pathogen. Molecular Biology and Evolution, 2020, 37, 417-428.	8.9	27
108	Balancing Selection on a Regulatory Region Exhibiting Ancient Variation That Predates Humanâ€“Neandertal Divergence. PLoS Genetics, 2013, 9, e1003404.	3.5	26

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109	Increasing Generality and Power of Rare-Variant Tests by Utilizing Extended Pedigrees. American Journal of Human Genetics, 2016, 99, 846-859.	6.2	26
110	The origin of human mutation in light of genomic data. Nature Reviews Genetics, 2021, 22, 672-686.	16.3	26
111	An integrated clinical program and crowdsourcing strategy for genomic sequencing and Mendelian disease gene discovery. Npj Genomic Medicine, 2018, 3, 21.	3.8	24
112	Maintenance of Adaptive Dynamics and No Detectable Load in a Range-Edge Outcrossing Plant Population. Molecular Biology and Evolution, 2021, 38, 1820-1836.	8.9	24
113	Identifying DNase I hypersensitive sites as driver distal regulatory elements in breast cancer. Nature Communications, 2017, 8, 436.	12.8	22
114	Evidence for secondary-variant genetic burden and non-random distribution across biological modules in a recessive ciliopathy. Nature Genetics, 2020, 52, 1145-1150.	21.4	22
115	Polygenic adaptation of rosette growth in Arabidopsis thaliana. PLoS Genetics, 2021, 17, e1008748.	3.5	22
116	Variants in angiopoietin-2 (ANGPT2) contribute to variation in nocturnal oxyhaemoglobin saturation level. Human Molecular Genetics, 2016, 25, ddw324.	2.9	21
117	Error-tolerant EST database searches by tandem mass spectrometry and multiTag software. Proteomics, 2005, 5, 4118-4122.	2.2	20
118	From analysis of protein structural alignments toward a novel approach to align protein sequences. Proteins: Structure, Function and Bioinformatics, 2004, 54, 569-582.	2.6	16
119	Commonalities across computational workflows for uncovering explanatory variants in undiagnosed cases. Genetics in Medicine, 2021, 23, 1075-1085.	2.4	16
120	Replicate sequencing libraries are important for quantification of allelic imbalance. Nature Communications, 2021, 12, 3370.	12.8	13
121	Individual variation in protein-coding sequences of human genome. Advances in Protein Chemistry, 2000, 54, 409-437.	4.4	11
122	Inherited CHST11/MIR3922 deletion is associated with a novel recessive syndrome presenting with skeletal malformation and malignant lymphoproliferative disease. Molecular Genetics & Genomic Medicine, 2015, 3, 413-423.	1.2	11
123	Maintenance of Complex Trait Variation: Classic Theory and Modern Data. Frontiers in Genetics, 2021, 12, 763363.	2.3	11
124	SNP2RFLP: a computational tool to facilitate genetic mapping using benchtop analysis of SNPs. Mammalian Genome, 2008, 19, 687-690.	2.2	8
125	An argument for early genomic sequencing in atypical cases: aWISP3variant leads to diagnosis of progressive pseudorheumatoid arthropathy of childhood. Rheumatology, 2016, 55, kev367.	1.9	6
126	Lessons from the CAGIâ€4 Hopkins clinical panel challenge. Human Mutation, 2017, 38, 1155-1168.	2.5	6

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127	Reply to â€˜Selective effects of heterozygous protein-truncating variantsâ€™. Nature Genetics, 2019, 51, 3-4.	21.4	6
128	Purifying selection on noncoding deletions of human regulatory loci detected using their cellular pleiotropy. Genome Research, 2021, 31, 935-946.	5.5	5
129	Overcoming constraints on the detection of recessive selection in human genes from population frequency data. American Journal of Human Genetics, 2022, 109, 33-49.	6.2	5
130	Systems biology and the analysis of genetic variation. Current Opinion in Genetics and Development, 2013, 23, 599-601.	3.3	3
131	Shared associations identify causal relationships between gene expression and immune cell phenotypes. Communications Biology, 2021, 4, 279.	4.4	3
132	A literature review at genome scale: improving clinical variant assessment. Genetics in Medicine, 2018, 20, 936-941.	2.4	1
133	Session Introduction. , 2004, , .		1
134	SEQuencing a baby for an optimal outcome: a genomic future for newborn screening. Molecular Genetics and Metabolism, 2021, 132, S138.	1.1	0