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	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
2	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
3	Genome-wide analysis of somatic noncoding mutation patterns in cancer. Science, 2022, 376, eabg5601.	12.6	33
4	Polygenic adaptation of rosette growth in Arabidopsis thaliana. PLoS Genetics, 2021, 17, e1008748.	3.5	22
5	Population-specific causal disease effect sizes in functionally important regions impacted by selection. Nature Communications, 2021, 12, 1098.	12.8	68
6	Commonalities across computational workflows for uncovering explanatory variants in undiagnosed cases. Genetics in Medicine, 2021, 23, 1075-1085.	2.4	16
7	Shared associations identify causal relationships between gene expression and immune cell phenotypes. Communications Biology, 2021, 4, 279.	4.4	3
8	SEQuencing a baby for an optimal outcome: a genomic future for newborn screening. Molecular Genetics and Metabolism, 2021, 132, S138.	1.1	0
9	Purifying selection on noncoding deletions of human regulatory loci detected using their cellular pleiotropy. Genome Research, 2021, 31, 935-946.	5.5	5
10	Replicate sequencing libraries are important for quantification of allelic imbalance. Nature Communications, 2021, 12, 3370.	12.8	13
11	The origin of human mutation in light of genomic data. Nature Reviews Genetics, 2021, 22, 672-686.	16.3	26
12	Population sequencing data reveal a compendium of mutational processes in the human germ line. Science, 2021, 373, 1030-1035.	12.6	43
13	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
14	Maintenance of Complex Trait Variation: Classic Theory and Modern Data. Frontiers in Genetics, 2021, 12, 763363.	2.3	11
15	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
16	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
17	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
18	Non-parametric Polygenic Risk Prediction via Partitioned GWAS Summary Statistics. American Journal of Human Genetics, 2020, 107, 46-59.	6.2	30

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19	Identification of cancer driver genes based on nucleotide context. Nature Genetics, 2020, 52, 208-218.	21.4	170
20	Mutations in RABL3 alter KRAS prenylation and are associated with hereditary pancreatic cancer. Nature Genetics, 2019, 51, 1308-1314.	21.4	47
21	GWAS for quantitative resistance phenotypes in Mycobacterium tuberculosis reveals resistance genes and regulatory regions. Nature Communications, 2019, 10, 2128.	12.8	111
22	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
23	Polygenic adaptation on height is overestimated due to uncorrected stratification in genome-wide association studies. ELife, 2019, 8, .	6.0	276
24	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
25	Quantification of frequency-dependent genetic architectures in 25 UK Biobank traits reveals action of negative selection. Nature Communications, 2019, 10, 790.	12.8	98
26	Reply to â€˜Selective effects of heterozygous protein-truncating variantsâ€™. Nature Genetics, 2019, 51, 3-4.	21.4	6
27	Admixture mapping identifies novel loci for obstructive sleep apnea in Hispanic/Latino Americans. Human Molecular Genetics, 2019, 28, 675-687.	2.9	41
28	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
29	A literature review at genome scale: improving clinical variant assessment. Genetics in Medicine, 2018, 20, 936-941.	2.4	1
30	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
31	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
32	PINES: phenotype-informed tissue weighting improves prediction of pathogenic noncoding variants. Genome Biology, 2018, 19, 173.	8.8	28
33	Automated typing of red blood cell and platelet antigens: a whole-genome sequencing study. Lancet Haematology,the, 2018, 5, e241-e251.	4.6	70
34	An integrated clinical program and crowdsourcing strategy for genomic sequencing and Mendelian disease gene discovery. Npj Genomic Medicine, 2018, 3, 21.	3.8	24
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	Negative selection in humans and fruit flies involves synergistic epistasis. Science, 2017, 356, 539-542.	12.6	103

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37	Estimating the selective effects of heterozygous protein-truncating variants from human exome data. Nature Genetics, 2017, 49, 806-810.	21.4	157
38	Identifying DNase I hypersensitive sites as driver distal regulatory elements in breast cancer. Nature Communications, 2017, 8, 436.	12.8	22
39	Lessons from the CAGIâ€4 Hopkins clinical panel challenge. Human Mutation, 2017, 38, 1155-1168.	2.5	6
40	Bayesian inference of negative and positive selection in human cancers. Nature Genetics, 2017, 49, 1785-1788.	21.4	90
41	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
42	Clinical Sequencing Exploratory Research Consortium: Accelerating Evidence-Based Practice of Genomic Medicine. American Journal of Human Genetics, 2016, 98, 1051-1066.	6.2	137
43	Excess of Deleterious Mutations around HLA Genes Reveals Evolutionary Cost of Balancing Selection. Molecular Biology and Evolution, 2016, 33, 2555-2564.	8.9	55
44	Increasing Generality and Power of Rare-Variant Tests by Utilizing Extended Pedigrees. American Journal of Human Genetics, 2016, 99, 846-859.	6.2	26
45	Variants in angiopoietin-2 (ANGPT2) contribute to variation in nocturnal oxyhaemoglobin saturation level. Human Molecular Genetics, 2016, 25, ddw324.	2.9	21
46	Genes with monoallelic expression contribute disproportionately to genetic diversity in humans. Nature Genetics, 2016, 48, 231-237.	21.4	83
47	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
48	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
49	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
50	Dominance of Deleterious Alleles Controls the Response to a Population Bottleneck. PLoS Genetics, 2015, 11, e1005436.	3.5	78
51	Leveraging Distant Relatedness to Quantify Human Mutation and Gene-Conversion Rates. American Journal of Human Genetics, 2015, 97, 775-789.	6.2	77
52	Integrative analysis of 111 reference human epigenomes. Nature, 2015, 518, 317-330.	27.8	5,653
53	Cell-of-origin chromatin organization shapes the mutational landscape of cancer. Nature, 2015, 518, 360-364.	27.8	491
54	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

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55	Identification of cis-suppression of human disease mutations by comparative genomics. Nature, 2015, 524, 225-229.	27.8	106
56	Widespread Macromolecular Interaction Perturbations in Human Genetic Disorders. Cell, 2015, 161, 647-660.	28.9	482
57	Genome-wide patterns and properties of de novo mutations in humans. Nature Genetics, 2015, 47, 822-826.	21.4	384
58	Widespread non-additive and interaction effects within HLA loci modulate the risk of autoimmune diseases. Nature Genetics, 2015, 47, 1085-1090.	21.4	164
59	Exome sequencing identifies rare LDLR and APOA5 alleles conferring risk for myocardial infarction. Nature, 2015, 518, 102-106.	27.8	581
60	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
61	Reduced local mutation density in regulatory DNA of cancer genomes is linked to DNA repair. Nature Biotechnology, 2014, 32, 71-75.	17.5	120
62	Systems biology and the analysis of genetic variation. Current Opinion in Genetics and Development, 2013, 23, 599-601.	3.3	3
63	Evaluating empirical bounds on complex disease genetic architecture. Nature Genetics, 2013, 45, 1418-1427.	21.4	147
64	Genomic variation landscape of the human gut microbiome. Nature, 2013, 493, 45-50.	27.8	783
65	Calibration of Multiple In Silico Tools for Predicting Pathogenicity of Mismatch Repair Gene Missense Substitutions. Human Mutation, 2013, 34, 255-265.	2.5	80
66	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
67	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
68	Mutational heterogeneity in cancer and the search for new cancer-associated genes. Nature, 2013, 499, 214-218.	27.8	4,761
69	Sequencing studies in human genetics: design and interpretation. Nature Reviews Genetics, 2013, 14, 460-470.	16.3	236
70	Balancing Selection on a Regulatory Region Exhibiting Ancient Variation That Predates Humanâ€“Neandertal Divergence. PLoS Genetics, 2013, 9, e1003404.	3.5	26
71	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
72	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

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73	Inferring causality and functional significance of human coding DNA variants. Human Molecular Genetics, 2012, 21, R10-R17.	2.9	47
74	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
75	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
76	Systematic Localization of Common Disease-Associated Variation in Regulatory DNA. Science, 2012, 337, 1190-1195.	12.6	3,129
77	Patterns and rates of exonic de novo mutations in autism spectrum disorders. Nature, 2012, 485, 242-245.	27.8	1,597
78	The accessible chromatin landscape of the human genome. Nature, 2012, 489, 75-82.	27.8	2,434
79	The interface of protein structure, protein biophysics, and molecular evolution. Protein Science, 2012, 21, 769-785.	7.6	188
80	Evolution and Functional Impact of Rare Coding Variation from Deep Sequencing of Human Exomes. Science, 2012, 337, 64-69.	12.6	1,535
81	Mutation mapping and identification by whole-genome sequencing. Genome Research, 2012, 22, 1541-1548.	5.5	126
82	Extremely low-coverage sequencing and imputation increases power for genome-wide association studies. Nature Genetics, 2012, 44, 631-635.	21.4	239
83	Exome sequencing and the genetic basis of complex traits. Nature Genetics, 2012, 44, 623-630.	21.4	340
84	Computational and statistical approaches to analyzing variants identified by exome sequencing. Genome Biology, 2011, 12, 227.	9.6	116
85	Genome sequencing reveals insights into physiology and longevity of the naked mole rat. Nature, 2011, 479, 223-227.	27.8	517
86	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
87	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
88	Pooled Association Tests for Rare Variants in Exon-Resequencing Studies. American Journal of Human Genetics, 2010, 86, 832-838.	6.2	715
89	Human allelic variation: perspective from protein function, structure, and evolution. Current Opinion in Structural Biology, 2010, 20, 342-350.	5.7	63
90	A method and server for predicting damaging missense mutations. Nature Methods, 2010, 7, 248-249.	19.0	11,491

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91	Multiplex padlock targeted sequencing reveals human hypermutable CpG variations. Genome Research, 2009, 19, 1606-1615.	5.5	62
92	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
93	Adaptive Mutations in the JC Virus Protein Capsid Are Associated with Progressive Multifocal Leukoencephalopathy (PML). PLoS Genetics, 2009, 5, e1000368.	3.5	114
94	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
95	Human mutation rate associated with DNA replication timing. Nature Genetics, 2009, 41, 393-395.	21.4	371
96	SNP2RFLP: a computational tool to facilitate genetic mapping using benchtop analysis of SNPs. Mammalian Genome, 2008, 19, 687-690.	2.2	8
97	Protein identification pipeline for the homology-driven proteomics. Journal of Proteomics, 2008, 71, 346-356.	2.4	76
98	Large-Scale Identification and Evolution Indexing of Tyrosine Phosphorylation Sites from Murine Brain. Journal of Proteome Research, 2008, 7, 311-318.	3.7	153
99	Proportionally more deleterious genetic variation in European than in African populations. Nature, 2008, 451, 994-997.	27.8	365
100	Hypermutable Non-Synonymous Sites Are under Stronger Negative Selection. PLoS Genetics, 2008, 4, e1000281.	3.5	32
101	Shifting Paradigm of Association Studies: Value of Rare Single-Nucleotide Polymorphisms. American Journal of Human Genetics, 2008, 82, 100-112.	6.2	292
102	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
103	Assessing the Evolutionary Impact of Amino Acid Mutations in the Human Genome. PLoS Genetics, 2008, 4, e1000083.	3.5	586
104	Analysis of Sequence Conservation at Nucleotide Resolution. PLoS Computational Biology, 2007, 3, e254.	3.2	65
105	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
106	Medical Sequencing at the Extremes of Human Body Mass. American Journal of Human Genetics, 2007, 80, 779-791.	6.2	199
107	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
108	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

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109	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
110	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
111	A universal trend of amino acid gain and loss in protein evolution. Nature, 2005, 433, 633-638.	27.8	282
112	A limited role for balancing selection. Trends in Genetics, 2005, 21, 30-32.	6.7	103
113	Error-tolerant EST database searches by tandem mass spectrometry and multiTag software. Proteomics, 2005, 5, 4118-4122.	2.2	20
114	Small fitness effect of mutations in highly conserved non-coding regions. Human Molecular Genetics, 2005, 14, 2221-2229.	2.9	74
115	Evolutionary constraints in conserved nongenic sequences of mammals. Genome Research, 2005, 15, 1373-1378.	5.5	50
116	Indel-Based Evolutionary Distance and Mouse-Human Divergence. Genome Research, 2004, 14, 1610-1616.	5.5	48
117	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
118	Positive selection at sites of multiple amino acid replacements since ratâ€“mouse divergence. Nature, 2004, 429, 558-562.	27.8	70
119	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
120	Homology-based functional proteomics by mass spectrometry: Application to theXenopus microtubule-associated proteome. Proteomics, 2004, 4, 2707-2721.	2.2	67
121	Session Introduction. , 2004, , .		1
122	Metabolites: a helping hand for pathway evolution?. Trends in Biochemical Sciences, 2003, 28, 336-341.	7.5	133
123	Increase of functional diversity by alternative splicing. Trends in Genetics, 2003, 19, 124-128.	6.7	208
124	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
125	A Model for Statistical Significance of Local Similarities in Structure. Journal of Molecular Biology, 2003, 326, 1307-1316.	4.2	161
126	Impact of selection, mutation rate and genetic drift on human genetic variation. Human Molecular Genetics, 2003, 12, 3325-3330.	2.9	32

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127	Human non-synonymous SNPs: server and survey. Nucleic Acids Research, 2002, 30, 3894-3900.	14.5	1,995
128	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
129	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
130	Individual variation in protein-coding sequences of human genome. Advances in Protein Chemistry, 2000, 54, 409-437.	4.4	11
131	Towards a structural basis of human non-synonymous single nucleotide polymorphisms. Trends in Genetics, 2000, 16, 198-200.	6.7	333
132	SNP frequencies in human genes. Trends in Genetics, 2000, 16, 335-337.	6.7	78
133	Prediction of nonsynonymous single nucleotide polymorphisms in human disease-associated genes. Journal of Molecular Medicine, 1999, 77, 754-760.	3.9	42
134	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