

Shamil R Sunyaev

List of Publications by Year in descending order

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134
papers

55,705
citations

14124

69
h-index

13635

134
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161
all docs

161
docs citations

161
times ranked

90274
citing authors

#	ARTICLE	IF	CITATIONS
1	Overcoming constraints on the detection of recessive selection in human genes from population frequency data. <i>American Journal of Human Genetics</i> , 2022, 109, 33-49.	2.6	5
2	StrVCTVRE: A supervised learning method to predict the pathogenicity of human genome structural variants. <i>American Journal of Human Genetics</i> , 2022, 109, 195-209.	2.6	29
3	Genome-wide analysis of somatic noncoding mutation patterns in cancer. <i>Science</i> , 2022, 376, eabg5601.	6.0	33
4	Polygenic adaptation of rosette growth in <i>Arabidopsis thaliana</i> . <i>PLoS Genetics</i> , 2021, 17, e1008748.	1.5	22
5	Population-specific causal disease effect sizes in functionally important regions impacted by selection. <i>Nature Communications</i> , 2021, 12, 1098.	5.8	68
6	Commonalities across computational workflows for uncovering explanatory variants in undiagnosed cases. <i>Genetics in Medicine</i> , 2021, 23, 1075-1085.	1.1	16
7	Shared associations identify causal relationships between gene expression and immune cell phenotypes. <i>Communications Biology</i> , 2021, 4, 279.	2.0	3
8	SEQuencing a baby for an optimal outcome: a genomic future for newborn screening. <i>Molecular Genetics and Metabolism</i> , 2021, 132, S138.	0.5	0
9	Purifying selection on noncoding deletions of human regulatory loci detected using their cellular pleiotropy. <i>Genome Research</i> , 2021, 31, 935-946.	2.4	5
10	Replicate sequencing libraries are important for quantification of allelic imbalance. <i>Nature Communications</i> , 2021, 12, 3370.	5.8	13
11	The origin of human mutation in light of genomic data. <i>Nature Reviews Genetics</i> , 2021, 22, 672-686.	7.7	26
12	Population sequencing data reveal a compendium of mutational processes in the human germ line. <i>Science</i> , 2021, 373, 1030-1035.	6.0	43
13	Maintenance of Adaptive Dynamics and No Detectable Load in a Range-Edge Outcrossing Plant Population. <i>Molecular Biology and Evolution</i> , 2021, 38, 1820-1836.	3.5	24
14	Maintenance of Complex Trait Variation: Classic Theory and Modern Data. <i>Frontiers in Genetics</i> , 2021, 12, 763363.	1.1	11
15	Fine-Scale Haplotype Structure Reveals Strong Signatures of Positive Selection in a Recombining Bacterial Pathogen. <i>Molecular Biology and Evolution</i> , 2020, 37, 417-428.	3.5	27
16	Evidence for secondary-variant genetic burden and non-random distribution across biological modules in a recessive ciliopathy. <i>Nature Genetics</i> , 2020, 52, 1145-1150.	9.4	22
17	Dynamic incorporation of multiple in silico functional annotations empowers rare variant association analysis of large whole-genome sequencing studies at scale. <i>Nature Genetics</i> , 2020, 52, 969-983.	9.4	146
18	Non-parametric Polygenic Risk Prediction via Partitioned GWAS Summary Statistics. <i>American Journal of Human Genetics</i> , 2020, 107, 46-59.	2.6	30

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19	Identification of cancer driver genes based on nucleotide context. <i>Nature Genetics</i> , 2020, 52, 208-218.	9.4	170
20	Mutations in RABL3 alter KRAS prenylation and are associated with hereditary pancreatic cancer. <i>Nature Genetics</i> , 2019, 51, 1308-1314.	9.4	47
21	GWAS for quantitative resistance phenotypes in <i>Mycobacterium tuberculosis</i> reveals resistance genes and regulatory regions. <i>Nature Communications</i> , 2019, 10, 2128.	5.8	111
22	Associations of variants in the hexokinase 1 and interleukin 18 receptor regions with oxyhemoglobin saturation during sleep. <i>PLoS Genetics</i> , 2019, 15, e1007739.	1.5	28
23	Polygenic adaptation on height is overestimated due to uncorrected stratification in genome-wide association studies. <i>ELife</i> , 2019, 8, .	2.8	276
24	Applicability of the Mutationâ€“Selection Balance Model to Population Genetics of Heterozygous Protein-Truncating Variants in Humans. <i>Molecular Biology and Evolution</i> , 2019, 36, 1701-1710.	3.5	27
25	Quantification of frequency-dependent genetic architectures in 25 UK Biobank traits reveals action of negative selection. <i>Nature Communications</i> , 2019, 10, 790.	5.8	98
26	Reply to â€“Selective effects of heterozygous protein-truncating variantsâ€™. <i>Nature Genetics</i> , 2019, 51, 3-4.	9.4	6
27	Admixture mapping identifies novel loci for obstructive sleep apnea in Hispanic/Latino Americans. <i>Human Molecular Genetics</i> , 2019, 28, 675-687.	1.4	41
28	Error-prone bypass of DNA lesions during lagging-strand replication is a common source of germline and cancer mutations. <i>Nature Genetics</i> , 2019, 51, 36-41.	9.4	28
29	A literature review at genome scale: improving clinical variant assessment. <i>Genetics in Medicine</i> , 2018, 20, 936-941.	1.1	1
30	Multiethnic Meta-Analysis Identifies <i>RAI1</i> as a Possible Obstructive Sleep Apneaâ€“related Quantitative Trait Locus in Men. <i>American Journal of Respiratory Cell and Molecular Biology</i> , 2018, 58, 391-401.	1.4	65
31	Functional architecture of low-frequency variants highlights strength of negative selection across coding and non-coding annotations. <i>Nature Genetics</i> , 2018, 50, 1600-1607.	9.4	132
32	PINES: phenotype-informed tissue weighting improves prediction of pathogenic noncoding variants. <i>Genome Biology</i> , 2018, 19, 173.	3.8	28
33	Automated typing of red blood cell and platelet antigens: a whole-genome sequencing study. <i>Lancet Haematology</i> , 2018, 5, e241-e251.	2.2	70
34	An integrated clinical program and crowdsourcing strategy for genomic sequencing and Mendelian disease gene discovery. <i>Npj Genomic Medicine</i> , 2018, 3, 21.	1.7	24
35	Limited statistical evidence for shared genetic effects of eQTLs and autoimmune-disease-associated loci in three major immune-cell types. <i>Nature Genetics</i> , 2017, 49, 600-605.	9.4	205
36	Negative selection in humans and fruit flies involves synergistic epistasis. <i>Science</i> , 2017, 356, 539-542.	6.0	103

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37	Estimating the selective effects of heterozygous protein-truncating variants from human exome data. <i>Nature Genetics</i> , 2017, 49, 806-810.	9.4	157
38	Identifying DNase I hypersensitive sites as driver distal regulatory elements in breast cancer. <i>Nature Communications</i> , 2017, 8, 436.	5.8	22
39	Lessons from the CAGI4 Hopkins clinical panel challenge. <i>Human Mutation</i> , 2017, 38, 1155-1168.	1.1	6
40	Bayesian inference of negative and positive selection in human cancers. <i>Nature Genetics</i> , 2017, 49, 1785-1788.	9.4	90
41	An argument for early genomic sequencing in atypical cases: a <i>WISP3</i> variant leads to diagnosis of progressive pseudorheumatoid arthropathy of childhood. <i>Rheumatology</i> , 2016, 55, kev367.	0.9	6
42	Clinical Sequencing Exploratory Research Consortium: Accelerating Evidence-Based Practice of Genomic Medicine. <i>American Journal of Human Genetics</i> , 2016, 98, 1051-1066.	2.6	137
43	Excess of Deleterious Mutations around HLA Genes Reveals Evolutionary Cost of Balancing Selection. <i>Molecular Biology and Evolution</i> , 2016, 33, 2555-2564.	3.5	55
44	Increasing Generality and Power of Rare-Variant Tests by Utilizing Extended Pedigrees. <i>American Journal of Human Genetics</i> , 2016, 99, 846-859.	2.6	26
45	Variants in angiotensin-converting enzyme 2 (<i>ANGPT2</i>) contribute to variation in nocturnal oxyhaemoglobin saturation level. <i>Human Molecular Genetics</i> , 2016, 25, ddw324.	1.4	21
46	Genes with monoallelic expression contribute disproportionately to genetic diversity in humans. <i>Nature Genetics</i> , 2016, 48, 231-237.	9.4	83
47	Genetic Associations with Obstructive Sleep Apnea Traits in Hispanic/Latino Americans. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2016, 194, 886-897.	2.5	107
48	Inherited <i>CHST11/MIR3922</i> deletion is associated with a novel recessive syndrome presenting with skeletal malformation and malignant lymphoproliferative disease. <i>Molecular Genetics & Genomic Medicine</i> , 2015, 3, 413-423.	0.6	11
49	APOBEC-Induced Cancer Mutations Are Uniquely Enriched in Early-Replicating, Gene-Dense, and Active Chromatin Regions. <i>Cell Reports</i> , 2015, 13, 1103-1109.	2.9	80
50	Dominance of Deleterious Alleles Controls the Response to a Population Bottleneck. <i>PLoS Genetics</i> , 2015, 11, e1005436.	1.5	78
51	Leveraging Distant Relatedness to Quantify Human Mutation and Gene-Conversion Rates. <i>American Journal of Human Genetics</i> , 2015, 97, 775-789.	2.6	77
52	Integrative analysis of 111 reference human epigenomes. <i>Nature</i> , 2015, 518, 317-330.	13.7	5,653
53	Cell-of-origin chromatin organization shapes the mutational landscape of cancer. <i>Nature</i> , 2015, 518, 360-364.	13.7	491
54	No evidence that selection has been less effective at removing deleterious mutations in Europeans than in Africans. <i>Nature Genetics</i> , 2015, 47, 126-131.	9.4	182

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55	Identification of cis-suppression of human disease mutations by comparative genomics. <i>Nature</i> , 2015, 524, 225-229.	13.7	106
56	Widespread Macromolecular Interaction Perturbations in Human Genetic Disorders. <i>Cell</i> , 2015, 161, 647-660.	13.5	482
57	Genome-wide patterns and properties of de novo mutations in humans. <i>Nature Genetics</i> , 2015, 47, 822-826.	9.4	384
58	Widespread non-additive and interaction effects within HLA loci modulate the risk of autoimmune diseases. <i>Nature Genetics</i> , 2015, 47, 1085-1090.	9.4	164
59	Exome sequencing identifies rare LDLR and APOA5 alleles conferring risk for myocardial infarction. <i>Nature</i> , 2015, 518, 102-106.	13.7	581
60	Searching for missing heritability: Designing rare variant association studies. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2014, 111, E455-64.	3.3	570
61	Reduced local mutation density in regulatory DNA of cancer genomes is linked to DNA repair. <i>Nature Biotechnology</i> , 2014, 32, 71-75.	9.4	120
62	Systems biology and the analysis of genetic variation. <i>Current Opinion in Genetics and Development</i> , 2013, 23, 599-601.	1.5	3
63	Evaluating empirical bounds on complex disease genetic architecture. <i>Nature Genetics</i> , 2013, 45, 1418-1427.	9.4	147
64	Genomic variation landscape of the human gut microbiome. <i>Nature</i> , 2013, 493, 45-50.	13.7	783
65	Calibration of Multiple In Silico Tools for Predicting Pathogenicity of Mismatch Repair Gene Missense Substitutions. <i>Human Mutation</i> , 2013, 34, 255-265.	1.1	80
66	Impact of deleterious passenger mutations on cancer progression. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2013, 110, 2910-2915.	3.3	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. <i>American Journal of Human Genetics</i> , 2013, 92, 15-27.	2.6	83
68	Mutational heterogeneity in cancer and the search for new cancer-associated genes. <i>Nature</i> , 2013, 499, 214-218.	13.7	4,761
69	Sequencing studies in human genetics: design and interpretation. <i>Nature Reviews Genetics</i> , 2013, 14, 460-470.	7.7	236
70	Balancing Selection on a Regulatory Region Exhibiting Ancient Variation That Predates Human-Neandertal Divergence. <i>PLoS Genetics</i> , 2013, 9, e1003404.	1.5	26
71	Deleterious Alleles in the Human Genome Are on Average Younger Than Neutral Alleles of the Same Frequency. <i>PLoS Genetics</i> , 2013, 9, e1003301.	1.5	63
72	Gain-of-function mutations in the mechanically activated ion channel PIEZO2 cause a subtype of Distal Arthrogyriposis. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2013, 110, 4667-4672.	3.3	193

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73	Inferring causality and functional significance of human coding DNA variants. <i>Human Molecular Genetics</i> , 2012, 21, R10-R17.	1.4	47
74	The mystery of missing heritability: Genetic interactions create phantom heritability. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2012, 109, 1193-1198.	3.3	1,322
75	Differential Relationship of DNA Replication Timing to Different Forms of Human Mutation and Variation. <i>American Journal of Human Genetics</i> , 2012, 91, 1033-1040.	2.6	220
76	Systematic Localization of Common Disease-Associated Variation in Regulatory DNA. <i>Science</i> , 2012, 337, 1190-1195.	6.0	3,129
77	Patterns and rates of exonic de novo mutations in autism spectrum disorders. <i>Nature</i> , 2012, 485, 242-245.	13.7	1,597
78	The accessible chromatin landscape of the human genome. <i>Nature</i> , 2012, 489, 75-82.	13.7	2,434
79	The interface of protein structure, protein biophysics, and molecular evolution. <i>Protein Science</i> , 2012, 21, 769-785.	3.1	188
80	Evolution and Functional Impact of Rare Coding Variation from Deep Sequencing of Human Exomes. <i>Science</i> , 2012, 337, 64-69.	6.0	1,535
81	Mutation mapping and identification by whole-genome sequencing. <i>Genome Research</i> , 2012, 22, 1541-1548.	2.4	126
82	Extremely low-coverage sequencing and imputation increases power for genome-wide association studies. <i>Nature Genetics</i> , 2012, 44, 631-635.	9.4	239
83	Exome sequencing and the genetic basis of complex traits. <i>Nature Genetics</i> , 2012, 44, 623-630.	9.4	340
84	Computational and statistical approaches to analyzing variants identified by exome sequencing. <i>Genome Biology</i> , 2011, 12, 227.	13.9	116
85	Genome sequencing reveals insights into physiology and longevity of the naked mole rat. <i>Nature</i> , 2011, 479, 223-227.	13.7	517
86	Development and Validation of a Computational Method for Assessment of Missense Variants in Hypertrophic Cardiomyopathy. <i>American Journal of Human Genetics</i> , 2011, 88, 183-192.	2.6	73
87	Progressive Multifocal Leukoencephalopathy (PML) Development Is Associated With Mutations in JC Virus Capsid Protein VP1 That Change Its Receptor Specificity. <i>Journal of Infectious Diseases</i> , 2011, 204, 103-114.	1.9	135
88	Pooled Association Tests for Rare Variants in Exon-Resequencing Studies. <i>American Journal of Human Genetics</i> , 2010, 86, 832-838.	2.6	715
89	Human allelic variation: perspective from protein function, structure, and evolution. <i>Current Opinion in Structural Biology</i> , 2010, 20, 342-350.	2.6	63
90	A method and server for predicting damaging missense mutations. <i>Nature Methods</i> , 2010, 7, 248-249.	9.0	11,491

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91	Multiplex padlock targeted sequencing reveals human hypermutable CpG variations. <i>Genome Research</i> , 2009, 19, 1606-1615.	2.4	62
92	Power of deep, all-exon resequencing for discovery of human trait genes. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2009, 106, 3871-3876.	3.3	147
93	Adaptive Mutations in the JC Virus Protein Capsid Are Associated with Progressive Multifocal Leukoencephalopathy (PML). <i>PLoS Genetics</i> , 2009, 5, e1000368.	1.5	114
94	Triplet repeat length bias and variation in the human transcriptome. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2009, 106, 17095-17100.	3.3	33
95	Human mutation rate associated with DNA replication timing. <i>Nature Genetics</i> , 2009, 41, 393-395.	9.4	371
96	SNP2RFLP: a computational tool to facilitate genetic mapping using benchtop analysis of SNPs. <i>Mammalian Genome</i> , 2008, 19, 687-690.	1.0	8
97	Protein identification pipeline for the homology-driven proteomics. <i>Journal of Proteomics</i> , 2008, 71, 346-356.	1.2	76
98	Large-Scale Identification and Evolution Indexing of Tyrosine Phosphorylation Sites from Murine Brain. <i>Journal of Proteome Research</i> , 2008, 7, 311-318.	1.8	153
99	Proportionally more deleterious genetic variation in European than in African populations. <i>Nature</i> , 2008, 451, 994-997.	13.7	365
100	Hypermutable Non-Synonymous Sites Are under Stronger Negative Selection. <i>PLoS Genetics</i> , 2008, 4, e1000281.	1.5	32
101	Shifting Paradigm of Association Studies: Value of Rare Single-Nucleotide Polymorphisms. <i>American Journal of Human Genetics</i> , 2008, 82, 100-112.	2.6	292
102	Separating the Wheat from the Chaff: Unbiased Filtering of Background Tandem Mass Spectra Improves Protein Identification. <i>Journal of Proteome Research</i> , 2008, 7, 3382-3395.	1.8	37
103	Assessing the Evolutionary Impact of Amino Acid Mutations in the Human Genome. <i>PLoS Genetics</i> , 2008, 4, e1000083.	1.5	586
104	Analysis of Sequence Conservation at Nucleotide Resolution. <i>PLoS Computational Biology</i> , 2007, 3, e254.	1.5	65
105	Widely distributed noncoding purifying selection in the human genome. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2007, 104, 12410-12415.	3.3	84
106	Medical Sequencing at the Extremes of Human Body Mass. <i>American Journal of Human Genetics</i> , 2007, 80, 779-791.	2.6	199
107	Most Rare Missense Alleles Are Deleterious in Humans: Implications for Complex Disease and Association Studies. <i>American Journal of Human Genetics</i> , 2007, 80, 727-739.	2.6	547
108	Common Single-Nucleotide Polymorphisms Act in Concert to Affect Plasma Levels of High-Density Lipoprotein Cholesterol. <i>American Journal of Human Genetics</i> , 2007, 81, 1298-1303.	2.6	38

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109	Sequence similarity-driven proteomics in organisms with unknown genomes by LC-MS/MS and automated <i>de novo</i> sequencing. <i>Proteomics</i> , 2007, 7, 2318-2329.	1.3	98
110	Identification and analysis of functional elements in 1% of the human genome by the ENCODE pilot project. <i>Nature</i> , 2007, 447, 799-816.	13.7	4,709
111	A universal trend of amino acid gain and loss in protein evolution. <i>Nature</i> , 2005, 433, 633-638.	13.7	282
112	A limited role for balancing selection. <i>Trends in Genetics</i> , 2005, 21, 30-32.	2.9	103
113	Error-tolerant EST database searches by tandem mass spectrometry and multiTag software. <i>Proteomics</i> , 2005, 5, 4118-4122.	1.3	20
114	Small fitness effect of mutations in highly conserved non-coding regions. <i>Human Molecular Genetics</i> , 2005, 14, 2221-2229.	1.4	74
115	Evolutionary constraints in conserved nongenic sequences of mammals. <i>Genome Research</i> , 2005, 15, 1373-1378.	2.4	50
116	Indel-Based Evolutionary Distance and Mouse-Human Divergence. <i>Genome Research</i> , 2004, 14, 1610-1616.	2.4	48
117	The Power and the Limitations of Cross-Species Protein Identification by Mass Spectrometry-driven Sequence Similarity Searches. <i>Molecular and Cellular Proteomics</i> , 2004, 3, 238-249.	2.5	143
118	Positive selection at sites of multiple amino acid replacements since rat-mouse divergence. <i>Nature</i> , 2004, 429, 558-562.	13.7	70
119	From analysis of protein structural alignments toward a novel approach to align protein sequences. <i>Proteins: Structure, Function and Bioinformatics</i> , 2004, 54, 569-582.	1.5	16
120	Homology-based functional proteomics by mass spectrometry: Application to the <i>Xenopus</i> microtubule-associated proteome. <i>Proteomics</i> , 2004, 4, 2707-2721.	1.3	67
121	Session Introduction. , 2004, , .		1
122	Metabolites: a helping hand for pathway evolution?. <i>Trends in Biochemical Sciences</i> , 2003, 28, 336-341.	3.7	133
123	Increase of functional diversity by alternative splicing. <i>Trends in Genetics</i> , 2003, 19, 124-128.	2.9	208
124	MultiTag: A Multiple Error-Tolerant Sequence Tag Search for the Sequence-Similarity Identification of Proteins by Mass Spectrometry. <i>Analytical Chemistry</i> , 2003, 75, 1307-1315.	3.2	118
125	A Model for Statistical Significance of Local Similarities in Structure. <i>Journal of Molecular Biology</i> , 2003, 326, 1307-1316.	2.0	161
126	Impact of selection, mutation rate and genetic drift on human genetic variation. <i>Human Molecular Genetics</i> , 2003, 12, 3325-3330.	1.4	32

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127	Human non-synonymous SNPs: server and survey. <i>Nucleic Acids Research</i> , 2002, 30, 3894-3900.	6.5	1,995
128	Dobzhansky-Muller incompatibilities in protein evolution. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2002, 99, 14878-14883.	3.3	281
129	Charting the Proteomes of Organisms with Unsequenced Genomes by MALDI-Quadrupole Time-of-Flight Mass Spectrometry and BLAST Homology Searching. <i>Analytical Chemistry</i> , 2001, 73, 1917-1926.	3.2	569
130	Individual variation in protein-coding sequences of human genome. <i>Advances in Protein Chemistry</i> , 2000, 54, 409-437.	4.4	11
131	Towards a structural basis of human non-synonymous single nucleotide polymorphisms. <i>Trends in Genetics</i> , 2000, 16, 198-200.	2.9	333
132	SNP frequencies in human genes. <i>Trends in Genetics</i> , 2000, 16, 335-337.	2.9	78
133	Prediction of nonsynonymous single nucleotide polymorphisms in human disease-associated genes. <i>Journal of Molecular Medicine</i> , 1999, 77, 754-760.	1.7	42
134	Homology-based fold predictions for <i>Mycoplasma genitalium</i> proteins 1 Edited by G. Von Heijne. <i>Journal of Molecular Biology</i> , 1998, 280, 323-326.	2.0	112