

Shamil R Sunyaev

List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

145 papers	43,401 citations	66 h-index	161 g-index
161 ext. papers	51,345 ext. citations	17.9 avg, IF	6.83 L-index

#	Paper	IF	Citations
145	StrVCTVRE: A supervised learning method to predict the pathogenicity of human genome structural variants.. <i>American Journal of Human Genetics</i> , 2022 ,	11	4
144	Genome-wide analysis of somatic noncoding mutation patterns in cancer.. <i>Science</i> , 2022 , 376, eabg5601	33.3	2
143	Maintenance of Complex Trait Variation: Classic Theory and Modern Data. <i>Frontiers in Genetics</i> , 2021 , 12, 763363	4.5	0
142	Shared associations identify causal relationships between gene expression and immune cell phenotypes. <i>Communications Biology</i> , 2021 , 4, 279	6.7	0
141	Purifying selection on noncoding deletions of human regulatory loci detected using their cellular pleiotropy. <i>Genome Research</i> , 2021 , 31, 935-946	9.7	1
140	Replicate sequencing libraries are important for quantification of allelic imbalance. <i>Nature Communications</i> , 2021 , 12, 3370	17.4	0
139	The origin of human mutation in light of genomic data. <i>Nature Reviews Genetics</i> , 2021 , 22, 672-686	30.1	2
138	Polygenic adaptation of rosette growth in <i>Arabidopsis thaliana</i> . <i>PLoS Genetics</i> , 2021 , 17, e1008748	6	4
137	Population-specific causal disease effect sizes in functionally important regions impacted by selection. <i>Nature Communications</i> , 2021 , 12, 1098	17.4	16
136	Commonalities across computational workflows for uncovering explanatory variants in undiagnosed cases. <i>Genetics in Medicine</i> , 2021 , 23, 1075-1085	8.1	2
135	Population sequencing data reveal a compendium of mutational processes in the human germ line. <i>Science</i> , 2021 , 373, 1030-1035	33.3	7
134	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	8.3	6
133	Non-parametric Polygenic Risk Prediction via Partitioned GWAS Summary Statistics. <i>American Journal of Human Genetics</i> , 2020 , 107, 46-59	11	16
132	Identification of cancer driver genes based on nucleotide context. <i>Nature Genetics</i> , 2020 , 52, 208-218	36.3	64
131	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	36.3	12
130	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	36.3	33
129	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	8.3	11

128	GWAS for quantitative resistance phenotypes in <i>Mycobacterium tuberculosis</i> reveals resistance genes and regulatory regions. <i>Nature Communications</i> , 2019 , 10, 2128	17.4	52
127	Associations of variants in the hexokinase 1 and interleukin 18 receptor regions with oxyhemoglobin saturation during sleep. <i>PLoS Genetics</i> , 2019 , 15, e1007739	6	14
126	Polygenic adaptation on height is overestimated due to uncorrected stratification in genome-wide association studies. <i>ELife</i> , 2019 , 8,	8.9	166
125	Applicability of the Mutation-Selection Balance Model to Population Genetics of Heterozygous Protein-Truncating Variants in Humans. <i>Molecular Biology and Evolution</i> , 2019 , 36, 1701-1710	8.3	9
124	Mutations in RABL3 alter KRAS prenylation and are associated with hereditary pancreatic cancer. <i>Nature Genetics</i> , 2019 , 51, 1308-1314	36.3	31
123	Quantification of frequency-dependent genetic architectures in 25 UK Biobank traits reveals action of negative selection. <i>Nature Communications</i> , 2019 , 10, 790	17.4	55
122	Reply to R Selective effects of heterozygous protein-truncating variantsR <i>Nature Genetics</i> , 2019 , 51, 3-4	36.3	1
121	Admixture mapping identifies novel loci for obstructive sleep apnea in Hispanic/Latino Americans. <i>Human Molecular Genetics</i> , 2019 , 28, 675-687	5.6	20
120	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	36.3	17
119	A literature review at genome scale: improving clinical variant assessment. <i>Genetics in Medicine</i> , 2018 , 20, 936-941	8.1	1
118	Multiethnic Meta-Analysis Identifies RAI1 as a Possible Obstructive Sleep Apnea-related Quantitative Trait Locus in Men. <i>American Journal of Respiratory Cell and Molecular Biology</i> , 2018 , 58, 391-401	5.7	37
117	An integrated clinical program and crowdsourcing strategy for genomic sequencing and Mendelian disease gene discovery. <i>Npj Genomic Medicine</i> , 2018 , 3, 21	6.2	15
116	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	36.3	72
115	PINES: phenotype-informed tissue weighting improves prediction of pathogenic noncoding variants. <i>Genome Biology</i> , 2018 , 19, 173	18.3	12
114	Automated typing of red blood cell and platelet antigens: a whole-genome sequencing study. <i>Lancet Haematology</i> , 2018 , 5, e241-e251	14.6	35
113	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	36.3	124
112	Negative selection in humans and fruit flies involves synergistic epistasis. <i>Science</i> , 2017 , 356, 539-542	33.3	53
111	Estimating the selective effects of heterozygous protein-truncating variants from human exome data. <i>Nature Genetics</i> , 2017 , 49, 806-810	36.3	84

110	Identifying DNase I hypersensitive sites as driver distal regulatory elements in breast cancer. <i>Nature Communications</i> , 2017 , 8, 436	17.4	14
109	Lessons from the CAGI-4 Hopkins clinical panel challenge. <i>Human Mutation</i> , 2017 , 38, 1155-1168	4.7	4
108	Bayesian inference of negative and positive selection in human cancers. <i>Nature Genetics</i> , 2017 , 49, 1785-1788	36.3	46
107	Variants in angiopoietin-2 (ANGPT2) contribute to variation in nocturnal oxyhaemoglobin saturation level. <i>Human Molecular Genetics</i> , 2016 , 25, 5244-5253	5.6	15
106	Genes with monoallelic expression contribute disproportionately to genetic diversity in humans. <i>Nature Genetics</i> , 2016 , 48, 231-237	36.3	47
105	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	10.2	70
104	An argument for early genomic sequencing in atypical cases: a WISP3 variant leads to diagnosis of progressive pseudorheumatoid arthropathy of childhood. <i>Rheumatology</i> , 2016 , 55, 586-9	3.9	4
103	Clinical Sequencing Exploratory Research Consortium: Accelerating Evidence-Based Practice of Genomic Medicine. <i>American Journal of Human Genetics</i> , 2016 , 98, 1051-1066	11	107
102	Excess of Deleterious Mutations around HLA Genes Reveals Evolutionary Cost of Balancing Selection. <i>Molecular Biology and Evolution</i> , 2016 , 33, 2555-64	8.3	29
101	Increasing Generality and Power of Rare-Variant Tests by Utilizing Extended Pedigrees. <i>American Journal of Human Genetics</i> , 2016 , 99, 846-859	11	13
100	Identification of cis-suppression of human disease mutations by comparative genomics. <i>Nature</i> , 2015 , 524, 225-9	50.4	84
99	Widespread macromolecular interaction perturbations in human genetic disorders. <i>Cell</i> , 2015 , 161, 647-660	56.2	343
98	Genome-wide patterns and properties of de novo mutations in humans. <i>Nature Genetics</i> , 2015 , 47, 822-826	36.3	267
97	Widespread non-additive and interaction effects within HLA loci modulate the risk of autoimmune diseases. <i>Nature Genetics</i> , 2015 , 47, 1085-90	36.3	112
96	Exome sequencing identifies rare LDLR and APOA5 alleles conferring risk for myocardial infarction. <i>Nature</i> , 2015 , 518, 102-6	50.4	463
95	Inherited CHST11/MIR3922 deletion is associated with a novel recessive syndrome presenting with skeletal malformation and malignant lymphoproliferative disease. <i>Molecular Genetics & Genomic Medicine</i> , 2015 , 3, 413-23	2.3	8
94	APOBEC-Induced Cancer Mutations Are Uniquely Enriched in Early-Replicating, Gene-Dense, and Active Chromatin Regions. <i>Cell Reports</i> , 2015 , 13, 1103-1109	10.6	57
93	Dominance of Deleterious Alleles Controls the Response to a Population Bottleneck. <i>PLoS Genetics</i> , 2015 , 11, e1005436	6	53

92	Leveraging Distant Relatedness to Quantify Human Mutation and Gene-Conversion Rates. <i>American Journal of Human Genetics</i> , 2015 , 97, 775-89	11	56
91	Integrative analysis of 111 reference human epigenomes. <i>Nature</i> , 2015 , 518, 317-30	50.4	3849
90	Cell-of-origin chromatin organization shapes the mutational landscape of cancer. <i>Nature</i> , 2015 , 518, 360-364	50.4	344
89	No evidence that selection has been less effective at removing deleterious mutations in Europeans than in Africans. <i>Nature Genetics</i> , 2015 , 47, 126-31	36.3	129
88	Reduced local mutation density in regulatory DNA of cancer genomes is linked to DNA repair. <i>Nature Biotechnology</i> , 2014 , 32, 71-5	44.5	92
87	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	11.5	437
86	Evaluating empirical bounds on complex disease genetic architecture. <i>Nature Genetics</i> , 2013 , 45, 1418-27	36.3	120
85	Genomic variation landscape of the human gut microbiome. <i>Nature</i> , 2013 , 493, 45-50	50.4	571
84	Calibration of multiple in silico tools for predicting pathogenicity of mismatch repair gene missense substitutions. <i>Human Mutation</i> , 2013 , 34, 255-65	4.7	70
83	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, 2910-5	11.5	210
82	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	11	72
81	Mutational heterogeneity in cancer and the search for new cancer-associated genes. <i>Nature</i> , 2013 , 499, 214-218	50.4	3616
80	Sequencing studies in human genetics: design and interpretation. <i>Nature Reviews Genetics</i> , 2013 , 14, 460-70	30.1	200
79	Balancing selection on a regulatory region exhibiting ancient variation that predates human-neandertal divergence. <i>PLoS Genetics</i> , 2013 , 9, e1003404	6	21
78	Deleterious alleles in the human genome are on average younger than neutral alleles of the same frequency. <i>PLoS Genetics</i> , 2013 , 9, e1003301	6	49
77	Gain-of-function mutations in the mechanically activated ion channel PIEZO2 cause a subtype of Distal Arthrogryposis. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2013 , 110, 4667-72	11.5	151
76	Differential relationship of DNA replication timing to different forms of human mutation and variation. <i>American Journal of Human Genetics</i> , 2012 , 91, 1033-40	11	172
75	Systematic localization of common disease-associated variation in regulatory DNA. <i>Science</i> , 2012 , 337, 1190-5	33.3	2262

74	Patterns and rates of exonic de novo mutations in autism spectrum disorders. <i>Nature</i> , 2012 , 485, 242-5	50.4	1300
73	The accessible chromatin landscape of the human genome. <i>Nature</i> , 2012 , 489, 75-82	50.4	1900
72	The interface of protein structure, protein biophysics, and molecular evolution. <i>Protein Science</i> , 2012 , 21, 769-85	6.3	136
71	Evolution and functional impact of rare coding variation from deep sequencing of human exomes. <i>Science</i> , 2012 , 337, 64-9	33.3	1280
70	Mutation mapping and identification by whole-genome sequencing. <i>Genome Research</i> , 2012 , 22, 1541-8	9.7	101
69	Extremely low-coverage sequencing and imputation increases power for genome-wide association studies. <i>Nature Genetics</i> , 2012 , 44, 631-5	36.3	184
68	Exome sequencing and the genetic basis of complex traits. <i>Nature Genetics</i> , 2012 , 44, 623-30	36.3	303
67	Inferring causality and functional significance of human coding DNA variants. <i>Human Molecular Genetics</i> , 2012 , 21, R10-7	5.6	46
66	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, 1193-8	11.5	1064
65	Computational and statistical approaches to analyzing variants identified by exome sequencing. <i>Genome Biology</i> , 2011 , 12, 227	18.3	104
64	Genome sequencing reveals insights into physiology and longevity of the naked mole rat. <i>Nature</i> , 2011 , 479, 223-7	50.4	410
63	Development and validation of a computational method for assessment of missense variants in hypertrophic cardiomyopathy. <i>American Journal of Human Genetics</i> , 2011 , 88, 183-92	11	60
62	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, 103-14	7	113
61	A method and server for predicting damaging missense mutations. <i>Nature Methods</i> , 2010 , 7, 248-9	21.6	9235
60	Pooled association tests for rare variants in exon-resequencing studies. <i>American Journal of Human Genetics</i> , 2010 , 86, 832-8	11	615
59	Human allelic variation: perspective from protein function, structure, and evolution. <i>Current Opinion in Structural Biology</i> , 2010 , 20, 342-50	8.1	56
58	Multiplex padlock targeted sequencing reveals human hypermutable CpG variations. <i>Genome Research</i> , 2009 , 19, 1606-15	9.7	59
57	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, 3871-6	11.5	139

56	Adaptive mutations in the JC virus protein capsid are associated with progressive multifocal leukoencephalopathy (PML). <i>PLoS Genetics</i> , 2009 , 5, e1000368	6	99
55	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, 17095-100	11.5	27
54	Human mutation rate associated with DNA replication timing. <i>Nature Genetics</i> , 2009 , 41, 393-5	36.3	288
53	Proportionally more deleterious genetic variation in European than in African populations. <i>Nature</i> , 2008 , 451, 994-7	50.4	299
52	Hypermutable non-synonymous sites are under stronger negative selection. <i>PLoS Genetics</i> , 2008 , 4, e1000281	10.2	26
51	Shifting paradigm of association studies: value of rare single-nucleotide polymorphisms. <i>American Journal of Human Genetics</i> , 2008 , 82, 100-12	11	255
50	Separating the wheat from the chaff: unbiased filtering of background tandem mass spectra improves protein identification. <i>Journal of Proteome Research</i> , 2008 , 7, 3382-95	5.6	35
49	Assessing the evolutionary impact of amino acid mutations in the human genome. <i>PLoS Genetics</i> , 2008 , 4, e1000083	6	473
48	SNP2RFLP: a computational tool to facilitate genetic mapping using benchtop analysis of SNPs. <i>Mammalian Genome</i> , 2008 , 19, 687-90	3.2	8
47	Protein identification pipeline for the homology-driven proteomics. <i>Journal of Proteomics</i> , 2008 , 71, 346-56	3.9	70
46	Large-scale identification and evolution indexing of tyrosine phosphorylation sites from murine brain. <i>Journal of Proteome Research</i> , 2008 , 7, 311-8	5.6	139
45	Sequence similarity-driven proteomics in organisms with unknown genomes by LC-MS/MS and automated de novo sequencing. <i>Proteomics</i> , 2007 , 7, 2318-29	4.8	93
44	Identification and analysis of functional elements in 1% of the human genome by the ENCODE pilot project. <i>Nature</i> , 2007 , 447, 799-816	50.4	4121
43	Analysis of sequence conservation at nucleotide resolution. <i>PLoS Computational Biology</i> , 2007 , 3, e254	5	55
42	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, 12410-5	11.5	75
41	Medical sequencing at the extremes of human body mass. <i>American Journal of Human Genetics</i> , 2007 , 80, 779-91	11	180
40	Most rare missense alleles are deleterious in humans: implications for complex disease and association studies. <i>American Journal of Human Genetics</i> , 2007 , 80, 727-39	11	461
39	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, 1298-303	11	32

38 Understanding the Functional Importance of Human Single Nucleotide Polymorphisms **2006**, 126-132

37 A universal trend of amino acid gain and loss in protein evolution. *Nature*, **2005**, 433, 633-8 50.4 212

36 A limited role for balancing selection. *Trends in Genetics*, **2005**, 21, 30-2 8.5 92

35 Error-tolerant EST database searches by tandem mass spectrometry and multiTag software. *Proteomics*, **2005**, 5, 4118-22 4.8 19

34 Small fitness effect of mutations in highly conserved non-coding regions. *Human Molecular Genetics*, **2005**, 14, 2221-9 5.6 67

33 Evolutionary constraints in conserved nongenic sequences of mammals. *Genome Research*, **2005**, 15, 1373-8 9.7 44

32 Indel-based evolutionary distance and mouse-human divergence. *Genome Research*, **2004**, 14, 1610-6 9.7 42

31 The power and the limitations of cross-species protein identification by mass spectrometry-driven sequence similarity searches. *Molecular and Cellular Proteomics*, **2004**, 3, 238-49 7.6 133

30 Positive selection at sites of multiple amino acid replacements since rat-mouse divergence. *Nature*, **2004**, 429, 558-62 50.4 58

29 From analysis of protein structural alignments toward a novel approach to align protein sequences. *Proteins: Structure, Function and Bioinformatics*, **2004**, 54, 569-82 4.2 13

28 Homology-based functional proteomics by mass spectrometry: application to the Xenopus microtubule-associated proteome. *Proteomics*, **2004**, 4, 2707-21 4.8 64

27 Impact of selection, mutation rate and genetic drift on human genetic variation. *Human Molecular Genetics*, **2003**, 12, 3325-30 5.6 23

26 Metabolites: a helping hand for pathway evolution?. *Trends in Biochemical Sciences*, **2003**, 28, 336-41 10.3 117

25 Increase of functional diversity by alternative splicing. *Trends in Genetics*, **2003**, 19, 124-8 8.5 171

24 MultiTag: multiple error-tolerant sequence tag search for the sequence-similarity identification of proteins by mass spectrometry. *Analytical Chemistry*, **2003**, 75, 1307-15 7.8 106

23 A model for statistical significance of local similarities in structure. *Journal of Molecular Biology*, **2003**, 326, 1307-16 6.5 139

22 Human non-synonymous SNPs: server and survey. *Nucleic Acids Research*, **2002**, 30, 3894-900 20.1 1767

21 Dobzhansky-Muller incompatibilities in protein evolution. *Proceedings of the National Academy of Sciences of the United States of America*, **2002**, 99, 14878-83 11.5 229

20	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, 1917-26	7.8	530
19	Individual variation in protein-coding sequences of human genome. <i>Advances in Protein Chemistry</i> , 2000 , 54, 409-37		10
18	Towards a structural basis of human non-synonymous single nucleotide polymorphisms. <i>Trends in Genetics</i> , 2000 , 16, 198-200	8.5	293
17	SNP frequencies in human genes an excess of rare alleles and differing modes of selection. <i>Trends in Genetics</i> , 2000 , 16, 335-7	8.5	72
16	Prediction of nonsynonymous single nucleotide polymorphisms in human disease-associated genes. <i>Journal of Molecular Medicine</i> , 1999 , 77, 754-60	5.5	38
15	Homology-based fold predictions for <i>Mycoplasma genitalium</i> proteins. <i>Journal of Molecular Biology</i> , 1998 , 280, 323-6	6.5	100
14	Low-frequency variant functional architectures reveal strength of negative selection across coding and non-coding annotations		1
13	Excess of Deleterious Mutations around HLA Genes Reveals Evolutionary Cost of Balancing Selection		1
12	Estimating the Selective Effect of Heterozygous Protein Truncating Variants from Human Exome Data		1
11	Quantification of frequency-dependent genetic architectures and action of negative selection in 25 UK Biobank traits		18
10	Population sequencing data reveal a compendium of mutational processes in human germline		4
9	Unexpected variability of allelic imbalance estimates from RNA sequencing		3
8	StrVCTVRE: A supervised learning method to predict the pathogenicity of human genome structural variants		5
7	Signals of polygenic adaptation on height have been overestimated due to uncorrected population structure in genome-wide association studies		19
6	Non-parametric polygenic risk prediction using partitioned GWAS summary statistics		3
5	Fine-scale haplotype structure reveals strong signatures of positive selection in a recombining bacterial pathogen		1
4	Maintenance of adaptive dynamics and no detectable load in a range-edge out-crossing plant population		2
3	Population-specific causal disease effect sizes in functionally important regions impacted by selection		4

2	Leveraging pleiotropy to discover and interpret GWAS results for sleep-associated traits	1
1	The missing link between genetic association and regulatory function	1