Kaitlin E Samocha

List of Publications by Year
in descending order

46 papers	31,589 citations	126907 33 h-index	197818 49 g-index
71 all docs	71 docs citations	71 times ranked	50659 citing authors

#	Article	IF	Citations
1	Analysis of protein-coding genetic variation in 60,706 humans. Nature, 2016, 536, 285-291.	27.8	9,051
2	The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 2020, 581, 434-443.	27.8	6,140
3	Synaptic, transcriptional and chromatin genes disrupted in autism. Nature, 2014, 515, 209-215.	27.8	2,254
4	Patterns and rates of exonic de novo mutations in autism spectrum disorders. Nature, 2012, 485, 242-245.	27.8	1,597
5	Large-Scale Exome Sequencing Study Implicates Both Developmental and Functional Changes in the Neurobiology of Autism. Cell, 2020, 180, 568-584.e23.	28.9	1,422
6	Insights into Autism Spectrum Disorder Genomic Architecture and Biology from 71 Risk Loci. Neuron, 2015, 87, 1215-1233.	8.1	1,219
7	High-throughput discovery of novel developmental phenotypes. Nature, 2016, 537, 508-514.	27.8	1,001
8	A framework for the interpretation of de novo mutation in human disease. Nature Genetics, 2014, 46, 944-950.	21.4	943
9	De novo mutations in congenital heart disease with neurodevelopmental and other congenital anomalies. Science, 2015, 350, 1262-1266.	12.6	646
10	A structural variation reference for medical and population genetics. Nature, 2020, 581, 444-451.	27.8	614
11	The ExAC browser: displaying reference data information from over 60 000 exomes. Nucleic Acids Research, 2017, 45, D840-D845.	14.5	587
12	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
13	Polygenic transmission disequilibrium confirms that common and rare variation act additively to create risk for autism spectrum disorders. Nature Genetics, 2017, 49, 978-985.	21.4	401
14	Evidence for 28 genetic disorders discovered by combining healthcare and research data. Nature, 2020, 586, 757-762.	27.8	343
15	Genetic risk for autism spectrum disorders and neuropsychiatric variation in the general population. Nature Genetics, 2016, 48, 552-555.	21.4	326
16	Refining the role of de novo protein-truncating variants in neurodevelopmental disorders by using population reference samples. Nature Genetics, 2017, 49, 504-510.	21.4	298
17	Human knockouts and phenotypic analysis in a cohort with a high rate of consanguinity. Nature, 2017, 544, 235-239.	27.8	292
18	Quantifying prion disease penetrance using large population control cohorts. Science Translational Medicine, 2016, 8, 322ra9.	12.4	289

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19	The Evaluation of Tools Used to Predict the Impact of Missense Variants Is Hindered by Two Types of Circularity. Human Mutation, 2015, 36, 513-523.	2.5	283
20	A recurrent de novo mutation in KCNC1 causes progressive myoclonus epilepsy. Nature Genetics, 2015, 47, 39-46.	21.4	245
21	Patterns of genic intolerance of rare copy number variation in 59,898 human exomes. Nature Genetics, 2016, 48, 1107-1111.	21.4	167
22	Estimating the selective effects of heterozygous protein-truncating variants from human exome data. Nature Genetics, 2017, 49, 806-810.	21.4	157
23	De Novo Coding Variants Are Strongly Associated with Tourette Disorder. Neuron, 2017, 94, 486-499.e9.	8.1	155
24	Transcript expression-aware annotation improves rare variant interpretation. Nature, 2020, 581, 452-458.	27.8	142
25	Analysis of Rare, Exonic Variation amongst Subjects with Autism Spectrum Disorders and Population Controls. PLoS Genetics, 2013, 9, e1003443.	3.5	133
26	SMCHD1 mutations associated with a rare muscular dystrophy can also cause isolated arhinia and Bosma arhinia microphthalmia syndrome. Nature Genetics, 2017, 49, 238-248.	21.4	131
27	Autism spectrum disorder severity reflects the average contribution of de novo and familial influences. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 15161-15165.	7.1	125
28	Exome sequencing in schizophrenia-affected parentâ€“offspring trios reveals risk conferred by protein-coding de novo mutations. Nature Neuroscience, 2020, 23, 185-193.	14.8	125
29	Evaluating drug targets through human loss-of-function genetic variation. Nature, 2020, 581, 459-464.	27.8	115
30	Genome-Wide Association Studies and the Problem of Relatedness Among Advanced Intercross Lines and Other Highly Recombinant Populations. Genetics, 2010, 185, 1033-1044.	2.9	99
31	Interpreting de novo Variation in Human Disease Using denovolyzeR. Current Protocols in Human Genetics, 2015, 87, 7.25.1-7.25.15.	3.5	84
32	A respiratory chain controlled signal transduction cascade in the mitochondrial intermembrane space mediates hydrogen peroxide signaling. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, E5679-88.	7.1	58
33	Addendum: The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 2021, 597, E3-E4.	27.8	45
34	Contribution of retrotransposition to developmental disorders. Nature Communications, 2019, 10, 4630.	12.8	43
35	Gene family information facilitates variant interpretation and identification of disease-associated genes in neurodevelopmental disorders. Genome Medicine, 2020, 12, 28.	8.2	42
36	Non-coding region variants upstream of MEF2C cause severe developmental disorder through three distinct loss-of-function mechanisms. American Journal of Human Genetics, 2021, 108, 1083-1094.	6.2	42

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37	Base-specific mutational intolerance near splice sites clarifies the role of nonessential splice nucleotides. Genome Research, 2018, 28, 968-974.	5.5	41
38	Quantifying unobserved protein-coding variants in human populations provides a roadmap for large-scale sequencing projects. Nature Communications, 2016, 7, 13293.	12.8	35
39	Fine mapping of QTL for prepulse inhibition in LG/J and SM/J mice using F₂ and advanced intercross lines. Genes, Brain and Behavior, 2010, 9, 759-767.	2.2	34
40	The contribution of X-linked coding variation to severe developmental disorders. Nature Communications, 2021, 12, 627.	12.8	33
41	Replication of long-bone length QTL in the F9-F10 LG,SM advanced intercross. Mammalian Genome, 2009, 20, 224-235.	2.2	32
42	A framework for the detection of de novo mutations in family-based sequencing data. European Journal of Human Genetics, 2017, 25, 227-233.	2.8	29
43	Reduced reproductive success is associated with selective constraint on human genes. Nature, 2022, 603, 858-863.	27.8	29
44	Network Analysis of Genome-Wide Selective Constraint Reveals a Gene Network Active in Early Fetal Brain Intolerant of Mutation. PLoS Genetics, 2016, 12, e1006121.	3.5	24
45	Genetic Effect of Chemotherapy Exposure in Children of Testicular Cancer Survivors. Clinical Cancer Research, 2016, 22, 2183-2189.	7.0	15
46	Reply to â€˜Selective effects of heterozygous protein-truncating variantsâ€™. Nature Genetics, 2019, 51, 3-4.	21.4	6