

Kaitlin E Samocha

List of Publications by Year in descending order

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Version: 2024-02-01

46
papers

31,589
citations

126708

33
h-index

197535

49
g-index

71
all docs

71
docs citations

71
times ranked

50659
citing authors

#	ARTICLE	IF	CITATIONS
1	Reduced reproductive success is associated with selective constraint on human genes. <i>Nature</i> , 2022, 603, 858-863.	13.7	29
2	Non-coding region variants upstream of MEF2C cause severe developmental disorder through three distinct loss-of-function mechanisms. <i>American Journal of Human Genetics</i> , 2021, 108, 1083-1094.	2.6	42
3	Addendum: The mutational constraint spectrum quantified from variation in 141,456 humans. <i>Nature</i> , 2021, 597, E3-E4.	13.7	45
4	The contribution of X-linked coding variation to severe developmental disorders. <i>Nature Communications</i> , 2021, 12, 627.	5.8	33
5	Exome sequencing in schizophrenia-affected parent-offspring trios reveals risk conferred by protein-coding de novo mutations. <i>Nature Neuroscience</i> , 2020, 23, 185-193.	7.1	125
6	Evidence for 28 genetic disorders discovered by combining healthcare and research data. <i>Nature</i> , 2020, 586, 757-762.	13.7	343
7	Evaluating drug targets through human loss-of-function genetic variation. <i>Nature</i> , 2020, 581, 459-464.	13.7	115
8	The mutational constraint spectrum quantified from variation in 141,456 humans. <i>Nature</i> , 2020, 581, 434-443.	13.7	6,140
9	A structural variation reference for medical and population genetics. <i>Nature</i> , 2020, 581, 444-451.	13.7	614
10	Transcript expression-aware annotation improves rare variant interpretation. <i>Nature</i> , 2020, 581, 452-458.	13.7	142
11	Gene family information facilitates variant interpretation and identification of disease-associated genes in neurodevelopmental disorders. <i>Genome Medicine</i> , 2020, 12, 28.	3.6	42
12	Large-Scale Exome Sequencing Study Implicates Both Developmental and Functional Changes in the Neurobiology of Autism. <i>Cell</i> , 2020, 180, 568-584.e23.	13.5	1,422
13	Contribution of retrotransposition to developmental disorders. <i>Nature Communications</i> , 2019, 10, 4630.	5.8	43
14	Reply to "Selective effects of heterozygous protein-truncating variants". <i>Nature Genetics</i> , 2019, 51, 3-4.	9.4	6
15	Base-specific mutational intolerance near splice sites clarifies the role of nonessential splice nucleotides. <i>Genome Research</i> , 2018, 28, 968-974.	2.4	41
16	SMCHD1 mutations associated with a rare muscular dystrophy can also cause isolated arhinia and Bosma arhinia microphthalmia syndrome. <i>Nature Genetics</i> , 2017, 49, 238-248.	9.4	131
17	The ExAC browser: displaying reference data information from over 60 000 exomes. <i>Nucleic Acids Research</i> , 2017, 45, D840-D845.	6.5	587
18	Refining the role of de novo protein-truncating variants in neurodevelopmental disorders by using population reference samples. <i>Nature Genetics</i> , 2017, 49, 504-510.	9.4	298

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19	Human knockouts and phenotypic analysis in a cohort with a high rate of consanguinity. <i>Nature</i> , 2017, 544, 235-239.	13.7	292
20	Polygenic transmission disequilibrium confirms that common and rare variation act additively to create risk for autism spectrum disorders. <i>Nature Genetics</i> , 2017, 49, 978-985.	9.4	401
21	De Novo Coding Variants Are Strongly Associated with Tourette Disorder. <i>Neuron</i> , 2017, 94, 486-499.e9.	3.8	155
22	Estimating the selective effects of heterozygous protein-truncating variants from human exome data. <i>Nature Genetics</i> , 2017, 49, 806-810.	9.4	157
23	A framework for the detection of de novo mutations in family-based sequencing data. <i>European Journal of Human Genetics</i> , 2017, 25, 227-233.	1.4	29
24	Analysis of protein-coding genetic variation in 60,706 humans. <i>Nature</i> , 2016, 536, 285-291.	13.7	9,051
25	Patterns of genic intolerance of rare copy number variation in 59,898 human exomes. <i>Nature Genetics</i> , 2016, 48, 1107-1111.	9.4	167
26	High-throughput discovery of novel developmental phenotypes. <i>Nature</i> , 2016, 537, 508-514.	13.7	1,001
27	Quantifying unobserved protein-coding variants in human populations provides a roadmap for large-scale sequencing projects. <i>Nature Communications</i> , 2016, 7, 13293.	5.8	35
28	Quantifying prion disease penetrance using large population control cohorts. <i>Science Translational Medicine</i> , 2016, 8, 322ra9.	5.8	289
29	Genetic Effect of Chemotherapy Exposure in Children of Testicular Cancer Survivors. <i>Clinical Cancer Research</i> , 2016, 22, 2183-2189.	3.2	15
30	Genetic risk for autism spectrum disorders and neuropsychiatric variation in the general population. <i>Nature Genetics</i> , 2016, 48, 552-555.	9.4	326
31	Network Analysis of Genome-Wide Selective Constraint Reveals a Gene Network Active in Early Fetal Brain Intolerant of Mutation. <i>PLoS Genetics</i> , 2016, 12, e1006121.	1.5	24
32	Interpreting <i>de novo</i> Variation in Human Disease Using denovolyzeR. <i>Current Protocols in Human Genetics</i> , 2015, 87, 7.25.1-7.25.15.	3.5	84
33	De novo mutations in congenital heart disease with neurodevelopmental and other congenital anomalies. <i>Science</i> , 2015, 350, 1262-1266.	6.0	646
34	The Evaluation of Tools Used to Predict the Impact of Missense Variants Is Hindered by Two Types of Circularity. <i>Human Mutation</i> , 2015, 36, 513-523.	1.1	283
35	Insights into Autism Spectrum Disorder Genomic Architecture and Biology from 71 Risk Loci. <i>Neuron</i> , 2015, 87, 1215-1233.	3.8	1,219
36	A respiratory chain controlled signal transduction cascade in the mitochondrial intermembrane space mediates hydrogen peroxide signaling. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2015, 112, E5679-88.	3.3	58

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37	A recurrent de novo mutation in KCNC1 causes progressive myoclonus epilepsy. <i>Nature Genetics</i> , 2015, 47, 39-46.	9.4	245
38	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
39	Synaptic, transcriptional and chromatin genes disrupted in autism. <i>Nature</i> , 2014, 515, 209-215.	13.7	2,254
40	A framework for the interpretation of de novo mutation in human disease. <i>Nature Genetics</i> , 2014, 46, 944-950.	9.4	943
41	Autism spectrum disorder severity reflects the average contribution of de novo and familial influences. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2014, 111, 15161-15165.	3.3	125
42	Analysis of Rare, Exonic Variation amongst Subjects with Autism Spectrum Disorders and Population Controls. <i>PLoS Genetics</i> , 2013, 9, e1003443.	1.5	133
43	Patterns and rates of exonic de novo mutations in autism spectrum disorders. <i>Nature</i> , 2012, 485, 242-245.	13.7	1,597
44	Fine mapping of QTL for prepulse inhibition in LG/J and SM/J mice using F ₂ and advanced intercross lines. <i>Genes, Brain and Behavior</i> , 2010, 9, 759-767.	1.1	34
45	Genome-Wide Association Studies and the Problem of Relatedness Among Advanced Intercross Lines and Other Highly Recombinant Populations. <i>Genetics</i> , 2010, 185, 1033-1044.	1.2	99
46	Replication of long-bone length QTL in the F9-F10 LG,SM advanced intercross. <i>Mammalian Genome</i> , 2009, 20, 224-235.	1.0	32