Jack A Kosmicki

List of Publications by Year
in descending order

28	23,667	218677	477307
papers	citations	26 h-index	29 g-index

45	45	45	43313
all docs	docs citations	times ranked	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	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
4	Predicting Splicing from Primary Sequence with Deep Learning. Cell, 2019, 176, 535-548.e24.	28.9	1,305
5	A framework for the interpretation of de novo mutation in human disease. Nature Genetics, 2014, 46, 944-950.	21.4	943
6	Computationally efficient whole-genome regression for quantitative and binary traits. Nature Genetics, 2021, 53, 1097-1103.	21.4	457
7	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
8	Exome sequencing and analysis of 454,787 UK Biobank participants. Nature, 2021, 599, 628-634.	27.8	377
9	Genetic risk for autism spectrum disorders and neuropsychiatric variation in the general population. Nature Genetics, 2016, 48, 552-555.	21.4	326
10	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
11	Predicting the clinical impact of human mutation with deep neural networks. Nature Genetics, 2018, 50, 1161-1170.	21.4	288
12	De novo variants in neurodevelopmental disorders with epilepsy. Nature Genetics, 2018, 50, 1048-1053.	21.4	230
13	Whole-genome sequencing reveals host factors underlying critical COVID-19. Nature, 2022, 607, 97-103.	27.8	174
14	Autism spectrum disorder and attention deficit hyperactivity disorder have a similar burden of rare protein-truncating variants. Nature Neuroscience, 2019, 22, 1961-1965.	14.8	148
15	Transcript expression-aware annotation improves rare variant interpretation. Nature, 2020, 581, 452-458.	27.8	142
16	Sequencing of 640,000 exomes identifies GPR75 variants associated with protection from obesity. Science, 2021, 373, .	12.6	130
17	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
18	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

#	Article	IF	Citations
19	Genome-wide analysis provides genetic evidence that ACE2 influences COVID-19 risk and yields risk scores associated with severe disease. Nature Genetics, 2022, 54, 382-392.	21.4	97
20	Pan-ancestry exome-wide association analyses of COVID-19 outcomes in 586,157 individuals. American Journal of Human Genetics, 2021, 108, 1350-1355.	6.2	72
21	Paternal-age-related de novo mutations and risk for five disorders. Nature Communications, 2019, 10, 3043.	12.8	63
22	Loss of heterozygosity of essential genes represents a widespread class of potential cancer vulnerabilities. Nature Communications, 2020, 11, 2517.	12.8	60
23	A Potential Contributory Role for Ciliary Dysfunction in the 16p11.2 600 kb BP4-BP5 Pathology. American Journal of Human Genetics, 2015, 96, 784-796.	6.2	53
24	Addendum: The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 2021, 597, E3-E4.	27.8	45
25	Gene family information facilitates variant interpretation and identification of disease-associated genes in neurodevelopmental disorders. Genome Medicine, 2020, 12, 28.	8.2	42
26	Discovery of rare variants for complex phenotypes. Human Genetics, 2016, 135, 625-634.	3.8	40
27	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
28	ERAP1, ERAP2, and Two Copies of HLA-Aw19 Alleles Increase the Risk for Birdshot Chorioretinopathy in HLA-A29 Carriers. , 2021, 62, 3.		14