

Eric Minikel

List of Publications by Year in descending order

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Version: 2024-02-01

23
papers

7,848
citations

471509

17
h-index

580821

25
g-index

41
all docs

41
docs citations

41
times ranked

18623
citing authors

#	ARTICLE	IF	CITATIONS
1	The mutational constraint spectrum quantified from variation in 141,456 humans. <i>Nature</i> , 2020, 581, 434-443.	27.8	6,140
2	Using high-resolution variant frequencies to empower clinical genome interpretation. <i>Genetics in Medicine</i> , 2017, 19, 1151-1158.	2.4	355
3	Quantifying prion disease penetrance using large population control cohorts. <i>Science Translational Medicine</i> , 2016, 8, 322ra9.	12.4	289
4	Characterization of the Prion Protein Binding Properties of Antisense Oligonucleotides. <i>Biomolecules</i> , 2020, 10, 1.	4.0	186
5	Evaluating drug targets through human loss-of-function genetic variation. <i>Nature</i> , 2020, 581, 459-464.	27.8	115
6	Strictly co-isogenic C57BL/6J- <i>Prnp^{0/0}</i> mice: A rigorous resource for prion science. <i>Journal of Experimental Medicine</i> , 2016, 213, 313-327.	8.5	98
7	Antisense oligonucleotides extend survival of prion-infected mice. <i>JCI Insight</i> , 2019, 4, .	5.0	80
8	Age at onset in genetic prion disease and the design of preventive clinical trials. <i>Neurology</i> , 2019, 93, e125-e134.	1.1	73
9	Prion protein lowering is a disease-modifying therapy across prion disease stages, strains and endpoints. <i>Nucleic Acids Research</i> , 2020, 48, 10615-10631.	14.5	69
10	Towards a treatment for genetic prion disease: trials and biomarkers. <i>Lancet Neurology</i> , The, 2020, 19, 361-368.	10.2	60
11	Addendum: The mutational constraint spectrum quantified from variation in 141,456 humans. <i>Nature</i> , 2021, 597, E3-E4.	27.8	45
12	Prion protein quantification in human cerebrospinal fluid as a tool for prion disease drug development. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2019, 116, 7793-7798.	7.1	41
13	Ascertainment Bias Causes False Signal of Anticipation in Genetic Prion Disease. <i>American Journal of Human Genetics</i> , 2014, 95, 371-382.	6.2	40
14	Measuring per Mile Risk for Pay-As-You-Drive Automobile Insurance. <i>Transportation Research Record</i> , 2012, 2297, 97-103.	1.9	36
15	Cerebrospinal fluid and plasma biomarkers in individuals at risk for genetic prion disease. <i>BMC Medicine</i> , 2020, 18, 140.	5.5	34
16	Domain-specific Quantification of Prion Protein in Cerebrospinal Fluid by Targeted Mass Spectrometry. <i>Molecular and Cellular Proteomics</i> , 2019, 18, 2388-2400.	3.8	22
17	Publicly Available Data Provide Evidence against NR1H3 R415Q Causing Multiple Sclerosis. <i>Neuron</i> , 2016, 92, 336-338.	8.1	21
18	Using High-Resolution Variant Frequencies Empowers Clinical Genome Interpretation and Enables Investigation of Genetic Architecture. <i>American Journal of Human Genetics</i> , 2019, 104, 187-190.	6.2	15

#	ARTICLE	IF	CITATIONS
19	Multimodal small-molecule screening for human prion protein binders. Journal of Biological Chemistry, 2020, 295, 13516-13531.	3.4	14
20	Regional variability and genotypic and pharmacodynamic effects on PrP concentration in the CNS. JCI Insight, 2022, 7, .	5.0	11
21	Autoantibodies against the prion protein in individuals with <i>PRNP</i> mutations. Neurology, 2020, 95, e2028-e2037.	1.1	7
22	Novel quaternary structures of the human prion protein globular domain. Biochimie, 2021, 191, 118-125.	2.6	4
23	Implications of new genetic risk factors in prion disease. Nature Reviews Neurology, 2021, 17, 5-6.	10.1	1