Eric Minikel

List of Publications by Year
in descending order

23 papers	7,848 citations	535685 17 h-index	651938 25 g-index
41 all docs	41 docs citations	41 times ranked	20031 citing authors

#	Article	IF	Citations
1	Regional variability and genotypic and pharmacodynamic effects on PrP concentration in the CNS. JCI Insight, 2022, 7, .	2.3	11
2	Implications of new genetic risk factors in prion disease. Nature Reviews Neurology, 2021, 17, 5-6.	4.9	1
3	Addendum: The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 2021, 597, E3-E4.	13.7	45
4	Novel quaternary structures of the human prion protein globular domain. Biochimie, 2021, 191, 118-125.	1.3	4
5	Characterization of the Prion Protein Binding Properties of Antisense Oligonucleotides. Biomolecules, 2020, 10, 1.	1.8	186
6	Prion protein lowering is a disease-modifying therapy across prion disease stages, strains and endpoints. Nucleic Acids Research, 2020, 48, 10615-10631.	6.5	69
7	Multimodal small-molecule screening for human prion protein binders. Journal of Biological Chemistry, 2020, 295, 13516-13531.	1.6	14
8	Evaluating drug targets through human loss-of-function genetic variation. Nature, 2020, 581, 459-464.	13.7	115
9	The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 2020, 581, 434-443.	13.7	6,140
10	Cerebrospinal fluid and plasma biomarkers in individuals at risk for genetic prion disease. BMC Medicine, 2020, 18, 140.	2.3	34
11	Towards a treatment for genetic prion disease: trials and biomarkers. Lancet Neurology, The, 2020, 19, 361-368.	4.9	60
12	Autoantibodies against the prion protein in individuals with PRNP mutations. Neurology, 2020, 95, e2028-e2037.	1.5	7
13	Age at onset in genetic prion disease and the design of preventive clinical trials. Neurology, 2019, 93, e125-e134.	1.5	73
14	Prion protein quantification in human cerebrospinal fluid as a tool for prion disease drug development. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 7793-7798.	3.3	41
15	Domain-specific Quantification of Prion Protein in Cerebrospinal Fluid by Targeted Mass Spectrometry. Molecular and Cellular Proteomics, 2019, 18, 2388-2400.	2.5	22
16	Using High-Resolution Variant Frequencies Empowers Clinical Genome Interpretation and Enables Investigation of Genetic Architecture. American Journal of Human Genetics, 2019, 104, 187-190.	2.6	15
17	Antisense oligonucleotides extend survival of prion-infected mice. JCI Insight, 2019, 4, .	2.3	80
18	Using high-resolution variant frequencies to empower clinical genome interpretation. Genetics in Medicine, 2017, 19, 1151-1158.	1.1	355

#	Article	IF	Citations
19	Strictly co-isogenic C57BL/6J-Prnpâ^'/â^' mice: A rigorous resource for prion science. Journal of Experimental Medicine, 2016, 213, 313-327.	4.2	98
20	Publicly Available Data Provide Evidence against NR1H3 R415Q Causing Multiple Sclerosis. Neuron, 2016, 92, 336-338.	3.8	21
21	Quantifying prion disease penetrance using large population control cohorts. Science Translational Medicine, 2016, 8, 322ra9.	5.8	289
22	Ascertainment Bias Causes False Signal of Anticipation in Genetic Prion Disease. American Journal of Human Genetics, 2014, 95, 371-382.	2.6	40
23	Measuring per Mile Risk for Pay-As-You-Drive Automobile Insurance. Transportation Research Record, 2012, 2297, 97-103.	1.0	36