Eric Vallabh Minikel

List of Publications by Year in descending order

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#	Article	IF	CITATIONS
1	The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 2020, 581, 434-443.	27.8	6,140
2	Quantifying prion disease penetrance using large population control cohorts. Science Translational Medicine, 2016, 8, 322ra9.	12.4	289
3	Characterization of the Prion Protein Binding Properties of Antisense Oligonucleotides. Biomolecules, 2020, 10, 1.	4.0	186
4	Evaluating drug targets through human loss-of-function genetic variation. Nature, 2020, 581, 459-464.	27.8	115
5	Antisense oligonucleotides extend survival of prion-infected mice. JCI Insight, 2019, 4, .	5.0	80
6	Age at onset in genetic prion disease and the design of preventive clinical trials. Neurology, 2019, 93, e125-e134.	1.1	73
7	Prion protein lowering is a disease-modifying therapy across prion disease stages, strains and endpoints. Nucleic Acids Research, 2020, 48, 10615-10631.	14.5	69
8	Towards a treatment for genetic prion disease: trials and biomarkers. Lancet Neurology, The, 2020, 19, 361-368.	10.2	60
9	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.	7.1	41
10	Ascertainment Bias Causes False Signal of Anticipation in Genetic Prion Disease. American Journal of Human Genetics, 2014, 95, 371-382.	6.2	40
11	Cerebrospinal fluid and plasma biomarkers in individuals at risk for genetic prion disease. BMC Medicine, 2020, 18, 140.	5.5	34
12	Domain-specific Quantification of Prion Protein in Cerebrospinal Fluid by Targeted Mass Spectrometry. Molecular and Cellular Proteomics, 2019, 18, 2388-2400.	3.8	22
13	Publicly Available Data Provide Evidence against NR1H3 R415Q Causing Multiple Sclerosis. Neuron, 2016, 92, 336-338.	8.1	21
14	Multimodal small-molecule screening for human prion protein binders. Journal of Biological Chemistry, 2020, 295, 13516-13531.	3.4	14
15	Regional variability and genotypic and pharmacodynamic effects on PrP concentration in the CNS. JCI Insight, 2022, 7, .	5.0	11
16	Autoantibodies against the prion protein in individuals with <i>PRNP</i> mutations. Neurology, 2020, 95, e2028-e2037.	1.1	7
17	Novel quaternary structures of the human prion protein globular domain. Biochimie, 2021, 191, 118-125.	2.6	4
18	Implications of new genetic risk factors in prion disease. Nature Reviews Neurology, 2021, 17, 5-6.	10.1	1