Henrike O Heyne

List of Publications by Year in descending order

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777949 1051228 1,746 16 13 16 citations h-index g-index papers 22 22 22 3446 docs citations times ranked citing authors all docs

#	Article	IF	CITATIONS
1	Rare coding variants in ten genes confer substantial risk for schizophrenia. Nature, 2022, 604, 509-516.	13.7	326
2	<i>STXBP1</i> encephalopathy. Neurology, 2016, 86, 954-962.	1.5	264
3	Ultra-Rare Genetic Variation in the Epilepsies: A Whole-Exome Sequencing Study of 17,606 Individuals. American Journal of Human Genetics, 2019, 105, 267-282.	2.6	237
4	De novo variants in neurodevelopmental disorders with epilepsy. Nature Genetics, 2018, 50, 1048-1053.	9.4	230
5	Predicting functional effects of missense variants in voltage-gated sodium and calcium channels. Science Translational Medicine, 2020, 12, .	5.8	84
6	Comprehensive characterization of amino acid positions in protein structures reveals molecular effect of missense variants. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 28201-28211.	3.3	68
7	Paternal-age-related de novo mutations and risk for five disorders. Nature Communications, 2019, 10, 3043.	5.8	63
8	Identification of pathogenic variant enriched regions across genes and gene families. Genome Research, 2020, 30, 62-71.	2.4	47
9	Targeted gene sequencing in 6994 individuals with neurodevelopmental disorder with epilepsy. Genetics in Medicine, 2019, 21, 2496-2503.	1.1	45
10	The origin and legacy of the Etruscans through a 2000-year archeogenomic time transect. Science Advances, 2021, 7, eabi7673.	4.7	44
11	Gene variant effects across sodium channelopathies predict function and guide precision therapy. Brain, 2022, 145, 4275-4286.	3.7	43
12	Sub-genic intolerance, ClinVar, and the epilepsies: A whole-exome sequencing study of 29,165 individuals. American Journal of Human Genetics, 2021, 108, 965-982.	2.6	35
13	<i>KCNT1</i> -related epilepsies and epileptic encephalopathies: phenotypic and mutational spectrum. Brain, 2021, 144, 3635-3650.	3.7	34
14	Analysing an allelic series of rare missense variants of <i>CACNA1I</i> in a Swedish schizophrenia cohort. Brain, 2022, 145, 1839-1853.	3.7	18
15	MISCAST: MIssense variant to protein StruCture Analysis web SuiTe. Nucleic Acids Research, 2020, 48, W132-W139.	6.5	14
16	A novel compound heterozygous leptin receptor mutation causes more severe obesity than in Lepr mice. Journal of Lipid Research, 2021, 62, 100105.	2.0	5