

Ilona Krey

List of Publications by Year in descending order

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Version: 2024-02-01

13
papers

669
citations

933447

10
h-index

996975

15
g-index

16
all docs

16
docs citations

16
times ranked

1290
citing authors

#	ARTICLE	IF	CITATIONS
1	Genotype-phenotype correlations in <i>SCN8A</i> -related disorders reveal prognostic and therapeutic implications. <i>Brain</i> , 2022, 145, 2991-3009.	7.6	69
2	Prenatal phenotype of PNKP-related primary microcephaly associated with variants affecting both the FHA and phosphatase domain. <i>European Journal of Human Genetics</i> , 2022, 30, 101-110.	2.8	3
3	L-Serine Treatment is Associated with Improvements in Behavior, EEG, and Seizure Frequency in Individuals with GRIN-Related Disorders Due to Null Variants. <i>Neurotherapeutics</i> , 2022, 19, 334-341.	4.4	21
4	The genetic landscape of intellectual disability and epilepsy in adults and the elderly: a systematic genetic work-up of 150 individuals. <i>Genetics in Medicine</i> , 2021, 23, 1492-1497.	2.4	31
5	Sub-genic intolerance, ClinVar, and the epilepsies: A whole-exome sequencing study of 29,165 individuals. <i>American Journal of Human Genetics</i> , 2021, 108, 965-982.	6.2	35
6	Clinical and therapeutic significance of genetic variation in the GRIN gene family encoding NMDARs. <i>Neuropharmacology</i> , 2021, 199, 108805.	4.1	25
7	The Angelman Syndrome Online Registry – A multilingual approach to support global research. <i>European Journal of Medical Genetics</i> , 2021, 64, 104349.	1.3	1
8	Genotype-phenotype correlation on 45 individuals with West syndrome. <i>European Journal of Paediatric Neurology</i> , 2020, 25, 134-138.	1.6	23
9	The landscape of epilepsy-related GATOR1 variants. <i>Genetics in Medicine</i> , 2019, 21, 398-408.	2.4	137
10	Ultra-Rare Genetic Variation in the Epilepsies: A Whole-Exome Sequencing Study of 17,606 Individuals. <i>American Journal of Human Genetics</i> , 2019, 105, 267-282.	6.2	237
11	Biallelic inherited <i>SCN8A</i> variants, a rare cause of <i>SCN8A</i> -related developmental and epileptic encephalopathy. <i>Epilepsia</i> , 2019, 60, 2277-2285.	5.1	18
12	NfL is a biomarker for adult-onset leukoencephalopathy with axonal spheroids and pigmented glia. <i>Neurology</i> , 2018, 91, 755-757.	1.1	11
13	A recurrent mutation in <i>KCNA2</i> as a novel cause of hereditary spastic paraplegia and ataxia. <i>Annals of Neurology</i> , 2016, 80, .	5.3	49