Ilona Krey

List of Publications by Year in descending order

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