Joseph D Symonds

List of Publications by Year
in descending order

20	968	687363 13	752698 20
papers	citations	h-index	g-index

23	23	23	1400
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Development and Validation of a Prediction Model for Early Diagnosis of SCN1A -Related Epilepsies. Neurology, 2022, 98, .	1.1	24
2	Gene variant effects across sodium channelopathies predict function and guide precision therapy. Brain, 2022, 145, 4275-4286.	7.6	43
3	The gain of function SCN1A disorder spectrum: novel epilepsy phenotypes and therapeutic implications. Brain, 2022, 145, 3816-3831.	7.6	43
4	SLITRK2 variants associated with neurodevelopmental disorders impair excitatory synaptic function and cognition in mice. Nature Communications, 2022, 13, .	12.8	6
5	Early childhood epilepsies: epidemiology, classification, aetiology, and socio-economic determinants. Brain, 2021, 144, 2879-2891.	7.6	64
6	CHD2 epilepsy: epigenetics and the quest for precision medicine. Developmental Medicine and Child Neurology, 2020, 62, 549-550.	2.1	1
7	Epilepsy and developmental disorders: Next generation sequencing in the clinic. European Journal of Paediatric Neurology, 2020, 24, 15-23.	1.6	98
8	SCN1A variants from bench to bedsideâ€"improved clinical prediction from functional characterization. Human Mutation, 2020, 41, 363-374.	2.5	37
9	Neuronal antibody prevalence in children with seizures under 3 years. Neurology, 2020, 95, e1590-e1598.	1.1	9
10	Infantile spasms: Etiology, lead time and treatment response in a resource limited setting. Epilepsy and Behavior Reports, 2020, 14, 100397.	1.0	14
11	Biological concepts in human sodium channel epilepsies and their relevance in clinical practice. Epilepsia, 2020, 61, 387-399.	5.1	65
12	Incidence and phenotypes of childhood-onset genetic epilepsies: a prospective population-based national cohort. Brain, 2019, 142, 2303-2318.	7.6	248
13	SLC35A2â€CDG: Functional characterization, expanded molecular, clinical, and biochemical phenotypes of 30 unreported Individuals. Human Mutation, 2019, 40, 908-925.	2.5	39
14	Further expansion of the genetic GABA-A-opathies. European Journal of Paediatric Neurology, 2018, 22, 344.	1.6	0
15	Genetics update: Monogenetics, polygene disorders and the quest for modifying genes. Neuropharmacology, 2018, 132, 3-19.	4.1	13
16	De novo mutations in MED13, a component of the Mediator complex, are associated with a novel neurodevelopmental disorder. Human Genetics, 2018, 137, 375-388.	3.8	46
17	Heart rate variability in epilepsy: A potential biomarker of <scp>sudden unexpected death in epilepsy</scp> risk. Epilepsia, 2018, 59, 1372-1380.	5.1	105
18	Heterozygous truncation mutations of the <scp>SMC</scp>1A gene cause a severe early onset epilepsy with cluster seizures in females: Detailed phenotyping of 10 new cases. Epilepsia, 2017, 58, 565-575.	5.1	35

#	Article	IF	Citations
19	Advances in epilepsy gene discovery and implications for epilepsy diagnosis and treatment. Current Opinion in Neurology, 2017, 30, 193-199.	3.6	72
20	Does measurement technique explain the mismatch between European head size and WHO charts?. Archives of Disease in Childhood, 2017, 102, 639-643.	1.9	3