

E Martina Bebin

List of Publications by Year in descending order

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Version: 2024-02-01

41
papers

2,032
citations

331259

21
h-index

301761

39
g-index

41
all docs

41
docs citations

41
times ranked

3563
citing authors

#	ARTICLE	IF	CITATIONS
1	Disruption of MeCP2â€“TCF20 complex underlies distinct neurodevelopmental disorders. Proceedings of the National Academy of Sciences of the United States of America, 2022, 119, .	3.3	15
2	Inherited variants in CHD3 show variable expressivity in Snijders Blok-Campeau syndrome. Genetics in Medicine, 2022, 24, 1283-1296.	1.1	9
3	Tuber Locations Associated with Infantile Spasms Map to a Common Brain Network. Annals of Neurology, 2021, 89, 726-739.	2.8	24
4	Long-read genome sequencing for the molecular diagnosis of neurodevelopmental disorders. Human Genetics and Genomics Advances, 2021, 2, 100023.	1.0	20
5	Long-term safety and efficacy of highly purified cannabidiol for treatment refractory epilepsy. Epilepsy and Behavior, 2021, 117, 107862.	0.9	27
6	Variants in the degron of AFF3 are associated with intellectual disability, mesomelic dysplasia, horseshoe kidney, and epileptic encephalopathy. American Journal of Human Genetics, 2021, 108, 857-873.	2.6	19
7	Pharmacogenetic Predictors of Cannabidiol Response and Tolerability in Treatmentâ€™Resistant Epilepsy. Clinical Pharmacology and Therapeutics, 2021, 110, 1368-1380.	2.3	22
8	Updated International Tuberous Sclerosis Complex Diagnostic Criteria and Surveillance and Management Recommendations. Pediatric Neurology, 2021, 123, 50-66.	1.0	230
9	Epilepsy Is Heterogeneous in Early-Life Tuberous Sclerosis Complex. Pediatric Neurology, 2021, 123, 1-9.	1.0	5
10	Profile of Autism Spectrum Disorder in Tuberous Sclerosis Complex: Results from a Longitudinal, Prospective, Multisite Study. Annals of Neurology, 2021, 90, 874-886.	2.8	13
11	EEG Spectral Features in Sleep of Autism Spectrum Disorders in Children with Tuberous Sclerosis Complex. Journal of Autism and Developmental Disorders, 2020, 50, 916-923.	1.7	2
12	Cognitive function and adaptive skills after a one-year trial of cannabidiol (CBD) in a pediatric sample with treatment-resistant epilepsy. Epilepsy and Behavior, 2020, 111, 107299.	0.9	24
13	Cannabidiol normalizes resting-state functional connectivity in treatment-resistant epilepsy. Epilepsy and Behavior, 2020, 112, 107297.	0.9	17
14	Effects of highly purified cannabidiol (CBD) on fMRI of working memory in treatment-resistant epilepsy. Epilepsy and Behavior, 2020, 112, 107358.	0.9	10
15	Fibulin-5 mutation featuring Charcot-Marie-Tooth disease, joint hyperlaxity, and scoliosis. Neurology: Genetics, 2020, 6, e476.	0.9	0
16	Epilepsy and Electroencephalographic Abnormalities in SATB2-Associated Syndrome. Pediatric Neurology, 2020, 112, 94-100.	1.0	10
17	Pilot Study of Neurodevelopmental Impact of Early Epilepsy Surgery in Tuberous Sclerosis Complex. Pediatric Neurology, 2020, 109, 39-46.	1.0	23
18	Reproducibility of Structural and Diffusion Tensor Imaging in the TACERN Multi-Center Study. Frontiers in Integrative Neuroscience, 2019, 13, 24.	1.0	32

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19	Drug-drug interactions with cannabidiol (CBD) appear to have no effect on treatment response in an open-label Expanded Access Program. <i>Epilepsy and Behavior</i> , 2019, 98, 201-206.	0.9	34
20	Cognitive functioning following long-term cannabidiol use in adults with treatment-resistant epilepsy. <i>Epilepsy and Behavior</i> , 2019, 97, 105-110.	0.9	34
21	fMRI study of cannabidiol-induced changes in attention control in treatment-resistant epilepsy. <i>Epilepsy and Behavior</i> , 2019, 96, 114-121.	0.9	30
22	Quality of life in adults enrolled in an open-label study of cannabidiol (CBD) for treatment-resistant epilepsy. <i>Epilepsy and Behavior</i> , 2019, 95, 10-17.	0.9	29
23	Higher cannabidiol plasma levels are associated with better seizure response following treatment with a pharmaceutical grade cannabidiol. <i>Epilepsy and Behavior</i> , 2019, 95, 131-136.	0.9	35
24	Deleterious Variation in BRSK2 Associates with a Neurodevelopmental Disorder. <i>American Journal of Human Genetics</i> , 2019, 104, 701-708.	2.6	19
25	Early white matter development is abnormal in tuberous sclerosis complex patients who develop autism spectrum disorder. <i>Journal of Neurodevelopmental Disorders</i> , 2019, 11, 36.	1.5	32
26	Visual and semi-automatic non-invasive detection of interictal fast ripples: A potential biomarker of epilepsy in children with tuberous sclerosis complex. <i>Clinical Neurophysiology</i> , 2018, 129, 1458-1466.	0.7	46
27	Natural history and genotype-phenotype correlations in 72 individuals with <i>SATB2</i> -associated syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 925-935.	0.7	57
28	Cover Image, Volume 176A, Number 4, April 2018. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, .	0.7	0
29	<i>NBEA</i> : Developmental disease gene with early generalized epilepsy phenotypes. <i>Annals of Neurology</i> , 2018, 84, 788-795.	2.8	44
30	High vigabatrin dosage is associated with lower risk of infantile spasms relapse among children with tuberous sclerosis complex. <i>Epilepsy Research</i> , 2018, 148, 1-7.	0.8	25
31	Social correlates of health status, quality of life, and mood states in patients treated with cannabidiol for epilepsy. <i>Epilepsy and Behavior</i> , 2017, 70, 364-369.	0.9	15
32	Mutations in EBF3 Disturb Transcriptional Profiles and Cause Intellectual Disability, Ataxia, and Facial Dysmorphism. <i>American Journal of Human Genetics</i> , 2017, 100, 117-127.	2.6	62
33	The use of cannabidiol for seizure management in patients with brain tumor-related epilepsy. <i>Neurocase</i> , 2017, 23, 287-291.	0.2	20
34	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. <i>American Journal of Human Genetics</i> , 2017, 101, 664-685.	2.6	337
35	Interactions between cannabidiol and commonly used antiepileptic drugs. <i>Epilepsia</i> , 2017, 58, 1586-1592.	2.6	267
36	Presentation and Diagnosis of Tuberous Sclerosis Complex in Infants. <i>Pediatrics</i> , 2017, 140, .	1.0	90

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37	Eliciting preferences on secondary findings: the Preferences Instrument for Genomic Secondary Results. <i>Genetics in Medicine</i> , 2017, 19, 337-344.	1.1	36
38	MYT1L mutations cause intellectual disability and variable obesity by dysregulating gene expression and development of the neuroendocrine hypothalamus. <i>PLoS Genetics</i> , 2017, 13, e1006957.	1.5	60
39	De novo <i>FGF12</i> mutation in 2 patients with neonatal-onset epilepsy. <i>Neurology: Genetics</i> , 2016, 2, e120.	0.9	29
40	Clinical Electroencephalographic Biomarker for Impending Epilepsy in Asymptomatic Tuberous Sclerosis Complex Infants. <i>Pediatric Neurology</i> , 2016, 54, 29-34.	1.0	93
41	Cannabis, cannabidiol, and epilepsy – From receptors to clinical response. <i>Epilepsy and Behavior</i> , 2014, 41, 277-282.	0.9	136