E Martina Bebin

List of Publications by Year
in descending order

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41 papers	2,032 citations	331670 21 h-index	302126 39 g-index
41 all docs	41 docs citations	41 times ranked	3563 citing authors

#	Article	IF	Citations
1	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	6.2	337
2	Interactions between cannabidiol and commonly used antiepileptic drugs. Epilepsia, 2017, 58, 1586-1592.	5.1	267
3	Updated International Tuberous Sclerosis Complex Diagnostic Criteria and Surveillance and Management Recommendations. Pediatric Neurology, 2021, 123, 50-66.	2.1	230
4	Cannabis, cannabidiol, and epilepsy â€" From receptors to clinical response. Epilepsy and Behavior, 2014, 41, 277-282.	1.7	136
5	Clinical Electroencephalographic Biomarker for Impending Epilepsy in Asymptomatic Tuberous Sclerosis Complex Infants. Pediatric Neurology, 2016, 54, 29-34.	2.1	93
6	Presentation and Diagnosis of Tuberous Sclerosis Complex in Infants. Pediatrics, 2017, 140, .	2.1	90
7	Mutations in EBF3 Disturb Transcriptional Profiles and Cause Intellectual Disability, Ataxia, and Facial Dysmorphism. American Journal of Human Genetics, 2017, 100, 117-127.	6.2	62
8	MYT1L mutations cause intellectual disability and variable obesity by dysregulating gene expression and development of the neuroendocrine hypothalamus. PLoS Genetics, 2017, 13, e1006957.	3.5	60
9	Natural history and genotypeâ€phenotype correlations in 72 individuals with SATB2â€associated syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 925-935.	1.2	57
10	Visual and semi-automatic non-invasive detection of interictal fast ripples: A potential biomarker of epilepsy in children with tuberous sclerosis complex. Clinical Neurophysiology, 2018, 129, 1458-1466.	1.5	46
11	NBEA: Developmental disease gene with early generalized epilepsy phenotypes. Annals of Neurology, 2018, 84, 788-795.	5.3	44
12	Eliciting preferences on secondary findings: the Preferences Instrument for Genomic Secondary Results. Genetics in Medicine, 2017, 19, 337-344.	2.4	36
13	Higher cannabidiol plasma levels are associated with better seizure response following treatment with a pharmaceutical grade cannabidiol. Epilepsy and Behavior, 2019, 95, 131-136.	1.7	35
14	Drugâ€"drug interactions with cannabidiol (CBD) appear to have no effect on treatment response in an open-label Expanded Access Program. Epilepsy and Behavior, 2019, 98, 201-206.	1.7	34
15	Cognitive functioning following long-term cannabidiol use in adults with treatment-resistant epilepsy. Epilepsy and Behavior, 2019, 97, 105-110.	1.7	34
16	Reproducibility of Structural and Diffusion Tensor Imaging in the TACERN Multi-Center Study. Frontiers in Integrative Neuroscience, 2019, 13, 24.	2.1	32
17	Early white matter development is abnormal in tuberous sclerosis complex patients who develop autism spectrum disorder. Journal of Neurodevelopmental Disorders, 2019, 11, 36.	3.1	32
18	fMRI study of cannabidiol-induced changes in attention control in treatment-resistant epilepsy. Epilepsy and Behavior, 2019, 96, 114-121.	1.7	30

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19	De novo FGF12 mutation in 2 patients with neonatal-onset epilepsy. Neurology: Genetics, 2016, 2, e120.	1.9	29
20	Quality of life in adults enrolled in an open-label study of cannabidiol (CBD) for treatment-resistant epilepsy. Epilepsy and Behavior, 2019, 95, 10-17.	1.7	29
21	Long-term safety and efficacy of highly purified cannabidiol for treatment refractory epilepsy. Epilepsy and Behavior, 2021, 117, 107862.	1.7	27
22	High vigabatrin dosage is associated with lower risk of infantile spasms relapse among children with tuberous sclerosis complex. Epilepsy Research, 2018, 148, 1-7.	1.6	25
23	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.	1.7	24
24	Tuber Locations Associated with Infantile Spasms Map to a Common Brain Network. Annals of Neurology, 2021, 89, 726-739.	5.3	24
25	Pilot Study of Neurodevelopmental Impact of Early Epilepsy Surgery in Tuberous Sclerosis Complex. Pediatric Neurology, 2020, 109, 39-46.	2.1	23
26	Pharmacogenetic Predictors of Cannabidiol Response and Tolerability in Treatmentâ€Resistant Epilepsy. Clinical Pharmacology and Therapeutics, 2021, 110, 1368-1380.	4.7	22
27	The use of cannabidiol for seizure management in patients with brain tumor-related epilepsy. Neurocase, 2017, 23, 287-291.	0.6	20
28	Long-read genome sequencing for the molecular diagnosis of neurodevelopmental disorders. Human Genetics and Genomics Advances, 2021, 2, 100023.	1.7	20
29	Deleterious Variation in BRSK2 Associates with a Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 104, 701-708.	6.2	19
30	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.	6.2	19
31	Cannabidiol normalizes resting-state functional connectivity in treatment-resistant epilepsy. Epilepsy and Behavior, 2020, 112, 107297.	1.7	17
32	Social correlates of health status, quality of life, and mood states in patients treated with cannabidiol for epilepsy. Epilepsy and Behavior, 2017, 70, 364-369.	1.7	15
33	Disruption of MeCP2â€“TCF20 complex underlies distinct neurodevelopmental disorders. Proceedings of the National Academy of Sciences of the United States of America, 2022, 119, .	7.1	15
34	Profile of Autism Spectrum Disorder in Tuberous Sclerosis Complex: Results from a Longitudinal, Prospective, Multisite Study. Annals of Neurology, 2021, 90, 874-886.	5.3	13
35	Effects of highly purified cannabidiol (CBD) on fMRI of working memory in treatment-resistant epilepsy. Epilepsy and Behavior, 2020, 112, 107358.	1.7	10
36	Epilepsy and Electroencephalographic Abnormalities in SATB2-Associated Syndrome. Pediatric Neurology, 2020, 112, 94-100.	2.1	10

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37	Inherited variants in CHD3 show variable expressivity in Snijders Blok-Campeau syndrome. Genetics in Medicine, 2022, 24, 1283-1296.	2.4	9
38	Epilepsy Is Heterogeneous in Early-Life Tuberous Sclerosis Complex. Pediatric Neurology, 2021, 123, 1-9.	2.1	5
39	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.	2.7	2
40	Cover Image, Volume 176A, Number 4, April 2018. American Journal of Medical Genetics, Part A, 2018, 176, .	1.2	0
41	Fibulin-5 mutation featuring Charcot-Marie-Tooth disease, joint hyperlaxity, and scoliosis. Neurology: Genetics, 2020, 6, e476.	1.9	0