Maria Stella Vari

List of Publications by Year
in descending order

25	408	840776 11	794594 19
papers	citations	h-index	g-index

25	25	25	724
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Expanding Phenotype of Poirierâ€“Bienvenu Syndrome: New Evidence from an Italian Multicentrical Cohort of Patients. Genes, 2022, 13, 276.	2.4	10
2	Epilepsy Course and Developmental Trajectories in STXBP1 -DEE. Neurology: Genetics, 2022, 8, .	1.9	24
3	Complex Neurological Phenotype Associated with a De Novo DHDDS Mutation in a Boy with Intellectual Disability, Refractory Epilepsy, and Movement Disorder. Journal of Pediatric Genetics, 2021, 10, 236-238.	0.7	10
4	Atypical Presentation of Aromatic L-Amino Acid Decarboxylase Deficiency with Developmental Epileptic Encephalopathy. Journal of Pediatric Epilepsy, 2021, 10, 124-127.	0.2	4
5	Electroclinical features of MEF2C haploinsufficiency-related epilepsy: A multicenter European study. Seizure: the Journal of the British Epilepsy Association, 2021, 88, 60-72.	2.0	6
6	New Trends and Most Promising Therapeutic Strategies for Epilepsy Treatment. Frontiers in Neurology, 2021, 12, 753753.	2.4	23
7	An Open Retrospective Study of a Standardized Cannabidiol Based-Oil in Treatment-Resistant Epilepsy. Cannabis and Cannabinoid Research, 2020, , .	2.9	12
8	Clinical spectrum and genotype-phenotype correlations in PRRT2 Italian patients. European Journal of Paediatric Neurology, 2020, 28, 193-197.	1.6	14
9	A further contribution to the delineation of epileptic phenotype in PACS2-related syndrome. Seizure: the Journal of the British Epilepsy Association, 2020, 79, 53-55.	2.0	15
10	Genotype-phenotype correlations in patients with de novo KCNQ2 pathogenic variants. Neurology: Genetics, 2020, 6, e528.	1.9	24
11	Epileptic Encephalopathy, Myoclonusâ€“Dystonia, and Premature Pubarche in Siblings with a Novel C-Terminal Truncating Mutation in ATRX Gene. Neuropediatrics, 2019, 50, 327-331.	0.6	6
12	Abnormal circadian rhythm in patients with GRIN1-related developmental epileptic encephalopathy. European Journal of Paediatric Neurology, 2019, 23, 657-661.	1.6	4
13	Pelizaeusâ€“Merzbacher Disease due to PLP1 Frameshift Mutation in a Female with Nonrandom Skewed X-Chromosome Inactivation. Neuropediatrics, 2019, 50, 268-270.	0.6	1
14	Three de novo DDX3X variants associated with distinctive brain developmental abnormalities and brain tumor in intellectually disabled females. European Journal of Human Genetics, 2019, 27, 1254-1259.	2.8	41
15	A novel homozygous MFN2 mutation associated with severe and atypical CMT2 phenotype. European Journal of Paediatric Neurology, 2018, 22, 563-567.	1.6	21
16	Erratum to â€œDe novo 12q22.q23.3 duplication associated with temporal lobe epilepsyâ€•[Seizure 57 (2018) 63â€“65]. Seizure: the Journal of the British Epilepsy Association, 2018, 57, R1.	2.0	0
17	Multiorgan mitochondrial dysfunction is not a main feature of MFN2 mutations (Reply to: CMT2 due) Tj ETQq1 1 0.784314 rgBT /Ove Paediatric Neurology, 2018, 22, 892-893.	1.6	1
18	De novo 12q22.q23.3 duplication associated with temporal lobe epilepsy. Seizure: the Journal of the British Epilepsy Association, 2018, 57, 63-65.	2.0	0

#	Article	IF	Citations
19	Pyridoxine-dependent epilepsies: an observational study on clinical, diagnostic, therapeutic and prognostic features in a pediatric cohort. Metabolic Brain Disease, 2018, 33, 261-269.	2.9	18
20	Confirmation of mutations inPROSCas a novel cause of vitamin B_₆-dependent epilepsy. Journal of Medical Genetics, 2017, 54, 809-814.	3.2	66
21	De novo 12q22.q23.3 duplication associated with temporal lobe epilepsy. Seizure: the Journal of the British Epilepsy Association, 2017, 50, 80-82.	2.0	4
22	Novel AMPD2 mutation in pontocerebellar hypoplasia, dysmorphisms, and teeth abnormalities. Neurology: Genetics, 2017, 3, e179.	1.9	22
23	Biochemical phenotyping unravels novel metabolic abnormalities and potential biomarkers associated with treatment of GLUT1 deficiency with ketogenic diet. PLoS ONE, 2017, 12, e0184022.	2.5	26
24	Management of genetic epilepsies: From empirical treatment to precision medicine. Pharmacological Research, 2016, 107, 426-429.	7.1	52
25	Todd Paralysis in Rolandic Epilepsy. Pediatric Neurology Briefs, 2015, 29, 50.	0.2	4