

Maria Stella Vari

List of Publications by Year in descending order

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Version: 2024-02-01

25
papers

408
citations

840776

11
h-index

794594

19
g-index

25
all docs

25
docs citations

25
times ranked

724
citing authors

#	ARTICLE	IF	CITATIONS
1	Confirmation of mutations in <i>PROSC</i> as a novel cause of vitamin B6-dependent epilepsy. <i>Journal of Medical Genetics</i> , 2017, 54, 809-814.	3.2	66
2	Management of genetic epilepsies: From empirical treatment to precision medicine. <i>Pharmacological Research</i> , 2016, 107, 426-429.	7.1	52
3	Three de novo DDX3X variants associated with distinctive brain developmental abnormalities and brain tumor in intellectually disabled females. <i>European Journal of Human Genetics</i> , 2019, 27, 1254-1259.	2.8	41
4	Biochemical phenotyping unravels novel metabolic abnormalities and potential biomarkers associated with treatment of GLUT1 deficiency with ketogenic diet. <i>PLoS ONE</i> , 2017, 12, e0184022.	2.5	26
5	Genotype-phenotype correlations in patients with de novo <i>KCNQ2</i> pathogenic variants. <i>Neurology: Genetics</i> , 2020, 6, e528.	1.9	24
6	Epilepsy Course and Developmental Trajectories in <i>STXBP1</i> -DEE. <i>Neurology: Genetics</i> , 2022, 8, .	1.9	24
7	New Trends and Most Promising Therapeutic Strategies for Epilepsy Treatment. <i>Frontiers in Neurology</i> , 2021, 12, 753753.	2.4	23
8	Novel <i>AMPD2</i> mutation in pontocerebellar hypoplasia, dysmorphism, and teeth abnormalities. <i>Neurology: Genetics</i> , 2017, 3, e179.	1.9	22
9	A novel homozygous MFN2 mutation associated with severe and atypical CMT2 phenotype. <i>European Journal of Paediatric Neurology</i> , 2018, 22, 563-567.	1.6	21
10	Pyridoxine-dependent epilepsies: an observational study on clinical, diagnostic, therapeutic and prognostic features in a pediatric cohort. <i>Metabolic Brain Disease</i> , 2018, 33, 261-269.	2.9	18
11	A further contribution to the delineation of epileptic phenotype in PACS2-related syndrome. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2020, 79, 53-55.	2.0	15
12	Clinical spectrum and genotype-phenotype correlations in PRRT2 Italian patients. <i>European Journal of Paediatric Neurology</i> , 2020, 28, 193-197.	1.6	14
13	An Open Retrospective Study of a Standardized Cannabidiol Based-Oil in Treatment-Resistant Epilepsy. <i>Cannabis and Cannabinoid Research</i> , 2020, , .	2.9	12
14	Complex Neurological Phenotype Associated with a De Novo DHDDS Mutation in a Boy with Intellectual Disability, Refractory Epilepsy, and Movement Disorder. <i>Journal of Pediatric Genetics</i> , 2021, 10, 236-238.	0.7	10
15	Expanding Phenotype of Poirierâ€“Bienvenu Syndrome: New Evidence from an Italian Multicentric Cohort of Patients. <i>Genes</i> , 2022, 13, 276.	2.4	10
16	Epileptic Encephalopathy, Myoclonusâ€“Dystonia, and Premature Pubarche in Siblings with a Novel C-Terminal Truncating Mutation in ATRX Gene. <i>Neuropediatrics</i> , 2019, 50, 327-331.	0.6	6
17	Electroclinical features of MEF2C haploinsufficiency-related epilepsy: A multicenter European study. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2021, 88, 60-72.	2.0	6
18	De novo 12q22.q23.3 duplication associated with temporal lobe epilepsy. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2017, 50, 80-82.	2.0	4

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19	Abnormal circadian rhythm in patients with GRIN1-related developmental epileptic encephalopathy. <i>European Journal of Paediatric Neurology</i> , 2019, 23, 657-661.	1.6	4
20	Atypical Presentation of Aromatic L-Amino Acid Decarboxylase Deficiency with Developmental Epileptic Encephalopathy. <i>Journal of Pediatric Epilepsy</i> , 2021, 10, 124-127.	0.2	4
21	Todd Paralysis in Rolandic Epilepsy. <i>Pediatric Neurology Briefs</i> , 2015, 29, 50.	0.2	4
22	Multiorgan mitochondrial dysfunction is not a main feature of MFN2 mutations (Reply to: CMT2 due to Tj ETQq0 0 0 rgBT /Overlock 10 Tf Paediatric Neurology, 2018, 22, 892-893.	1.6	1
23	Pelizaeusâ€“Merzbacher Disease due to PLP1 Frameshift Mutation in a Female with Nonrandom Skewed X-Chromosome Inactivation. <i>Neuropediatrics</i> , 2019, 50, 268-270.	0.6	1
24	Erratum to â€œDe novo 12q22.q23.3 duplication associated with temporal lobe epilepsyâ€•[Seizure 57 (2018) 63â€“65]. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2018, 57, R1.	2.0	0
25	De novo 12q22.q23.3 duplication associated with temporal lobe epilepsy. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2018, 57, 63-65.	2.0	0