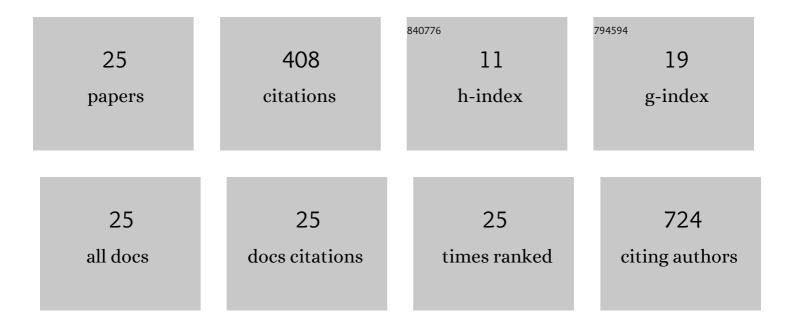
## Maria Stella Vari

List of Publications by Year in descending order

Source: https://exaly.com/author-pdf/5584876/publications.pdf Version: 2024-02-01



#	Article	IF	CITATIONS
1	Confirmation of mutations in <i>PROSC</i> as a novel cause of vitamin B <sub><sub>6</sub></sub> -dependent epilepsy. Journal of Medical Genetics, 2017, 54, 809-814.	3.2	66
2	Management of genetic epilepsies: From empirical treatment to precision medicine. Pharmacological Research, 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. European Journal of Human Genetics, 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. PLoS ONE, 2017, 12, e0184022.	2.5	26
5	Genotype-phenotype correlations in patients with de novo <i>KCNQ2</i> pathogenic variants. Neurology: Genetics, 2020, 6, e528.	1.9	24
6	Epilepsy Course and Developmental Trajectories in <i>STXBP1</i> -DEE. Neurology: Genetics, 2022, 8, .	1.9	24
7	New Trends and Most Promising Therapeutic Strategies for Epilepsy Treatment. Frontiers in Neurology, 2021, 12, 753753.	2.4	23
8	Novel <i>AMPD2</i> mutation in pontocerebellar hypoplasia, dysmorphisms, and teeth abnormalities. Neurology: Genetics, 2017, 3, e179.	1.9	22
9	A novel homozygous MFN2 mutation associated with severe and atypical CMT2 phenotype. European Journal of Paediatric Neurology, 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. Metabolic Brain Disease, 2018, 33, 261-269.	2.9	18
11	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
12	Clinical spectrum and genotype-phenotype correlations in PRRT2 Italian patients. European Journal of Paediatric Neurology, 2020, 28, 193-197.	1.6	14
13	An Open Retrospective Study of a Standardized Cannabidiol Based-Oil in Treatment-Resistant Epilepsy. Cannabis and Cannabinoid Research, 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. Journal of Pediatric Genetics, 2021, 10, 236-238.	0.7	10
15	Expanding Phenotype of Poirier–Bienvenu Syndrome: New Evidence from an Italian Multicentrical Cohort of Patients. Genes, 2022, 13, 276.	2.4	10
16	Epileptic Encephalopathy, Myoclonus–Dystonia, and Premature Pubarche in Siblings with a Novel C-Terminal Truncating Mutation in ATRX Gene. Neuropediatrics, 2019, 50, 327-331.	0.6	6
17	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
18	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

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