Stephanie Efthymiou

List of Publications by Year
in descending order

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74	2,038	361413	302126
papers	citations	20 h-index	39 g-index

81	81	81	3337
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Clinicoâ€radiological features, molecular spectrum, and identification of prognostic factors in developmental and epileptic encephalopathy due to inosine triphosphate pyrophosphatase (ITPase) deficiency. Human Mutation, 2022, 43, 403-419.	2.5	9
2	Bi-allelic variants inCHKAcause a neurodevelopmental disorder with epilepsy and microcephaly. Brain, 2022, 145, 1916-1923.	7.6	3
3	Mutations inTAF8cause a neurodegenerative disorder. Brain, 2022, 145, 3022-3034.	7.6	3
4	Inhibition of G-protein signalling in cardiac dysfunction of intellectual developmental disorder with cardiac arrhythmia (IDDCA) syndrome. Journal of Medical Genetics, 2021, 58, 815-831.	3.2	3
5	Novel variants broaden the phenotypic spectrum of PLEKHG5 â€associated neuropathies. European Journal of Neurology, 2021, 28, 1344-1355.	3.3	4
6	Brownâ€'Vialettoâ€'Van Laere and Fazioâ€'Londe syndromes: SLC52A3 mutations with puzzling phenotypes and inheritance. European Journal of Neurology, 2021, 28, 945-954.	3.3	5
7	Expanding the phenotype of PIGSâ€associated early onset epileptic developmental encephalopathy. Epilepsia, 2021, 62, e35-e41.	5.1	11
8	Expanding the Spectrum of <scp>AP5Z1â€</scp>Related Hereditary Spastic Paraplegia (<scp>HSPâ€SPG48</scp>): A Multicenter Study on a Rare Disease. Movement Disorders, 2021, 36, 1034-1038.	3.9	9
9	Allelic and phenotypic heterogeneity in Junctophillin-3 related neurodevelopmental and movement disorders. European Journal of Human Genetics, 2021, 29, 1027-1031.	2.8	5
10	Tay-Sachs Disease: Two Novel Rare HEXA Mutations from Pakistan and Morocco. Klinische Padiatrie, 2021, 233, 226-230.	0.6	4
11	Homozygous SCN1B variants causing early infantile epileptic encephalopathy 52 affect voltageâ€gated sodium channel function. Epilepsia, 2021, 62, e82-e87.	5.1	9
12	De novo mutation in SLC25A22 gene: expansion of the clinical and electroencephalographic phenotype. Journal of Neurogenetics, 2021, 35, 67-73.	1.4	3
13	Two novel biâ€allelic <scp>KDELR2</scp> missense variants cause osteogenesis imperfecta with neurodevelopmental features. American Journal of Medical Genetics, Part A, 2021, 185, 2241-2249.	1.2	7
14	Clinical, neuroimaging, and molecular spectrum of TECPR2 â€associated hereditary sensory and autonomic neuropathy with intellectual disability. Human Mutation, 2021, 42, 762-776.	2.5	18
15	Biallelic and monoallelic variants in PLXNA1 are implicated in a novel neurodevelopmental disorder with variable cerebral and eye anomalies. Genetics in Medicine, 2021, 23, 1715-1725.	2.4	22
16	Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia. Brain, 2021, 144, 1422-1434.	7.6	22
17	RFC1 expansions are a common cause of idiopathic sensory neuropathy. Brain, 2021, 144, 1542-1550.	7.6	63
18	Pathogenic variants in PIDD1 lead to an autosomal recessive neurodevelopmental disorder with pachygyria and psychiatric features. European Journal of Human Genetics, 2021, 29, 1226-1234.	2.8	8

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19	Novel ALDH3A2 mutations in structural and functional domains of FALDH causing diverse clinical phenotypes in SjÃ¶grenâ€“Larsson syndrome patients. Human Mutation, 2021, 42, 1015-1029.	2.5	0
20	Bi-allelic premature truncating variants in LTBP1 cause cutis laxa syndrome. American Journal of Human Genetics, 2021, 108, 1095-1114.	6.2	19
21	A novel variant in the DSE gene leads to Ehlersâ€“Danlos musculocontractural type 2 in a Pakistani family. Congenital Anomalies (discontinued), 2021, 61, 177-182.	0.6	3
22	Biallelic variants in PCDHGC4 cause a novel neurodevelopmental syndrome with progressive microcephaly, seizures, and joint anomalies. Genetics in Medicine, 2021, 23, 2138-2149.	2.4	11
23	Coding and noncoding variants in EBF3 are involved in HADDS and simplex autism. Human Genomics, 2021, 15, 44.	2.9	16
24	Genomeâ€Wide Association Study Identifies Risk Loci for Cluster Headache. Annals of Neurology, 2021, 90, 193-202.	5.3	31
25	Delineating the molecular and phenotypic spectrum of the SETD1B-related syndrome. Genetics in Medicine, 2021, 23, 2122-2137.	2.4	16
26	Prominent and Regressive Brain Developmental Disorders Associated with Nance-Horan Syndrome. Brain Sciences, 2021, 11, 1150.	2.3	10
27	Î±â€Synuclein (<scp>SNCA</scp>) <scp>A30G</scp> Mutation as a Cause of a Complex Phenotype Without Parkinsonism. Movement Disorders, 2021, 36, 2209-2212.	3.9	1
28	Early-onset phenotype of bi-allelic GRN mutations. Brain, 2021, 144, e22-e22.	7.6	5
29	Myoclonic status epilepticus and cerebellar hypoplasia associated with a novel variant in the GRIA3 gene. Neurogenetics, 2021, , 1.	1.4	2
30	Bi-allelic variants in OGDHL cause a neurodevelopmental spectrum disease featuring epilepsy, hearing loss, visual impairment, and ataxia. American Journal of Human Genetics, 2021, 108, 2368-2384.	6.2	12
31	A rare PANK2 deletion in the first north African patient affected with pantothenate kinase associated neurodegeneration. Journal of the Neurological Sciences, 2020, 410, 116639.	0.6	3
32	Identification of common genetic markers of paroxysmal neurological disorders using a network analysis approach. Neurological Sciences, 2020, 41, 851-857.	1.9	3
33	Loss of UGP2 in brain leads to a severe epileptic encephalopathy, emphasizing that bi-allelic isoform-specific start-loss mutations of essential genes can cause genetic diseases. Acta Neuropathologica, 2020, 139, 415-442.	7.7	38
34	Neuronal intranuclear inclusion disease is genetically heterogeneous. Annals of Clinical and Translational Neurology, 2020, 7, 1716-1725.	3.7	38
35	Early-infantile onset epilepsy and developmental delay caused by bi-allelic GAD1 variants. Brain, 2020, 143, 2388-2397.	7.6	28
36	De Novo and Bi-allelic Pathogenic Variants in NARS1 Cause Neurodevelopmental Delay Due to Toxic Gain-of-Function and Partial Loss-of-Function Effects. American Journal of Human Genetics, 2020, 107, 311-324.	6.2	32

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37	RFC1 expansions can mimic hereditary sensory neuropathy with cough and SjÃ¶gren syndrome. Brain, 2020, 143, e82-e82.	7.6	25
38	A homozygous GDAP2 loss-of-function variant in a patient with adult-onset cerebellar ataxia. Brain, 2020, 143, e49-e49.	7.6	5
39	MYORG -related disease is associated with central pontine calcifications and atypical parkinsonism. Neurology: Genetics, 2020, 6, e399.	1.9	13
40	Bi-allelic JAM2 Variants Lead to Early-Onset Recessive Primary Familial Brain Calcification. American Journal of Human Genetics, 2020, 106, 412-421.	6.2	47
41	Novel variants underlying autosomal recessive intellectual disability in Pakistani consanguineous families. BMC Medical Genetics, 2020, 21, 59.	2.1	3
42	Cerebellar ataxia, neuropathy, vestibular areflexia syndrome due to RFC1 repeat expansion. Brain, 2020, 143, 480-490.	7.6	140
43	Rare novel CYP2U1 and ZFYVE26 variants identified in two Pakistani families with spastic paraplegia. Journal of the Neurological Sciences, 2020, 411, 116669.	0.6	12
44	Bi-allelic Variants in the GPI Transamidase Subunit PIGK Cause a Neurodevelopmental Syndrome with Hypotonia, Cerebellar Atrophy, and Epilepsy. American Journal of Human Genetics, 2020, 106, 484-495.	6.2	22
45	Novel likely disease-causing CLN5 variants identified in Pakistani patients with neuronal ceroid lipofuscinosis. Journal of the Neurological Sciences, 2020, 414, 116826.	0.6	3
46	The genetics of intellectual disability: advancing technology and gene editing. F1000Research, 2020, 9, 22.	1.6	56
47	Age-dependent epileptic encephalopathy associated with an unusual co-occurrence of ZEB2 and SCN1A variants. , 2020, 22, 111-115.		2
48	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	12.8	150
49	Homozygous Missense Variants in NTNG2, Encoding a Presynaptic Netrin-G2 Adhesion Protein, Lead to a Distinct Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 105, 1048-1056.	6.2	30
50	Genotype-Phenotype Correlations in Charcot-Marie-Tooth Disease Due to MTMR2 Mutations and Implications in Membrane Trafficking. Frontiers in Neuroscience, 2019, 13, 974.	2.8	7
51	Biallelic mutations in neurofascin cause neurodevelopmental impairment and peripheral demyelination. Brain, 2019, 142, 2948-2964.	7.6	43
52	Continuum of phenotypes in hereditary motor and sensory neuropathy with proximal predominance and Charcotâ€“Marieâ€“Tooth patients with TFG mutation. American Journal of Medical Genetics, Part A, 2019, 179, 1507-1515.	1.2	7
53	Mutations in ACTL6B Cause Neurodevelopmental Deficits and Epilepsy and Lead to Loss of Dendrites in Human Neurons. American Journal of Human Genetics, 2019, 104, 815-834.	6.2	59
54	Biallelic expansion of an intronic repeat in RFC1 is a common cause of late-onset ataxia. Nature Genetics, 2019, 51, 649-658.	21.4	338

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55	Mutations in the Neuronal Vesicular SNARE VAMP2 Affect Synaptic Membrane Fusion and Impair Human Neurodevelopment. American Journal of Human Genetics, 2019, 104, 721-730.	6.2	88
56	A de novo truncating mutation in ASXL1 associated with segmental overgrowth. Journal of Genetics, 2019, 98, 1.	0.7	5
57	Pioneers and Emerging Pediatric Neurologists and Epileptologists in the World. Journal of Pediatric Epilepsy, 2019, 08, 001-001.	0.2	0
58	Biotin-Thiamine Responsive Encephalopathy: Report of an Egyptian Family with a Novel SLC19A3 Mutation and Review of the Literature. Journal of Pediatric Genetics, 2019, 08, 100-108.	0.7	7
59	Genotypeâ€phenotype correlations, dystonia and disease progression in spinocerebellar ataxia type 14. Movement Disorders, 2018, 33, 1119-1129.	3.9	26
60	Mutations in XRCC1 cause cerebellar ataxia and peripheral neuropathy. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 1230-1232.	1.9	18
61	A homozygous lossâ€ofâ€function mutation in PDE2A associated to earlyâ€onset hereditary chorea. Movement Disorders, 2018, 33, 482-488.	3.9	52
62	A loss-of-function homozygous mutation in DDX59 implicates a conserved DEAD-box RNA helicase in nervous system development and function. Human Mutation, 2018, 39, 187-192.	2.5	44
63	Electroclinical history of a five-year-old girl with GRIN1-related early-onset epileptic encephalopathy: a video-case study. Epileptic Disorders, 2018, 20, 423-427.	1.3	7
64	A Review of Copy Number Variants in Inherited Neuropathies. Current Genomics, 2018, 19, 412-419.	1.6	19
65	PDE10A and ADCY5 mutations linked to molecular and microstructural basal ganglia pathology. Movement Disorders, 2018, 33, 1961-1965.	3.9	38
66	A novel SLC1A4 homozygous mutation causing congenital microcephaly, epileptic encephalopathy and spastic tetraparesis: a video-EEG and tractography â€“ case study. Journal of Neurogenetics, 2018, 32, 316-321.	1.4	12
67	Paroxysmal Movement Disorder and Epilepsy Caused by a De Novo Truncating Mutation in KAT6A. Journal of Pediatric Genetics, 2018, 07, 114-116.	0.7	10
68	Biallelic Mutations in ADPRHL2, Encoding ADP-Ribosylhydrolase 3, Lead to a Degenerative Pediatric Stress-Induced Epileptic Ataxia Syndrome. American Journal of Human Genetics, 2018, 103, 431-439.	6.2	62
69	Homozygous mutations in <scp>VAMP</scp>1 cause a presynaptic congenital myasthenic syndrome. Annals of Neurology, 2017, 81, 597-603.	5.3	48
70	Mutations in NKX6-2 Cause Progressive Spastic Ataxia and Hypomyelination. American Journal of Human Genetics, 2017, 100, 969-977.	6.2	38
71	The phenotypic and molecular spectrum of PEHO syndrome and PEHO-like disorders. Brain, 2017, 140, e49-e49.	7.6	33
72	Analysis of the prion protein gene in multiple system atrophy. Neurobiology of Aging, 2017, 49, 216.e15-216.e18.	3.1	6

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73	PRUNE is crucial for normal brain development and mutated in microcephaly with neurodevelopmental impairment. Brain, 2017, 140, 940-952.	7.6	62
74	Genotype-phenotype correlations and expansion of the molecular spectrum of AP4M1-related hereditary spastic paraplegia. Orphanet Journal of Rare Diseases, 2017, 12, 172.	2.7	17