Stephanie Efthymiou

List of Publications by Year
in descending order

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	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
2	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	12.8	150
3	Cerebellar ataxia, neuropathy, vestibular areflexia syndrome due to RFC1 repeat expansion. Brain, 2020, 143, 480-490.	7.6	140
4	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
5	RFC1 expansions are a common cause of idiopathic sensory neuropathy. Brain, 2021, 144, 1542-1550.	7.6	63
6	PRUNE is crucial for normal brain development and mutated in microcephaly with neurodevelopmental impairment. Brain, 2017, 140, 940-952.	7.6	62
7	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
8	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
9	The genetics of intellectual disability: advancing technology and gene editing. F1000Research, 2020, 9, 22.	1.6	56
10	A homozygous lossâ€ofâ€function mutation in PDE2A associated to earlyâ€onset hereditary chorea. Movement Disorders, 2018, 33, 482-488.	3.9	52
11	Homozygous mutations in <scp>VAMP</scp>1 cause a presynaptic congenital myasthenic syndrome. Annals of Neurology, 2017, 81, 597-603.	5.3	48
12	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
13	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
14	Biallelic mutations in neurofascin cause neurodevelopmental impairment and peripheral demyelination. Brain, 2019, 142, 2948-2964.	7.6	43
15	Mutations in NKX6-2 Cause Progressive Spastic Ataxia and Hypomyelination. American Journal of Human Genetics, 2017, 100, 969-977.	6.2	38
16	PDE10A and ADCY5 mutations linked to molecular and microstructural basal ganglia pathology. Movement Disorders, 2018, 33, 1961-1965.	3.9	38
17	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
18	Neuronal intranuclear inclusion disease is genetically heterogeneous. Annals of Clinical and Translational Neurology, 2020, 7, 1716-1725.	3.7	38

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19	The phenotypic and molecular spectrum of PEHO syndrome and PEHO-like disorders. Brain, 2017, 140, e49-e49.	7.6	33
20	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
21	Genomeâ€Wide Association Study Identifies Risk Loci for Cluster Headache. Annals of Neurology, 2021, 90, 193-202.	5.3	31
22	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
23	Early-infantile onset epilepsy and developmental delay caused by bi-allelic GAD1 variants. Brain, 2020, 143, 2388-2397.	7.6	28
24	Genotypeâ€phenotype correlations, dystonia and disease progression in spinocerebellar ataxia type 14. Movement Disorders, 2018, 33, 1119-1129.	3.9	26
25	RFC1 expansions can mimic hereditary sensory neuropathy with cough and SjÃ¶gren syndrome. Brain, 2020, 143, e82-e82.	7.6	25
26	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
27	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
28	Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia. Brain, 2021, 144, 1422-1434.	7.6	22
29	A Review of Copy Number Variants in Inherited Neuropathies. Current Genomics, 2018, 19, 412-419.	1.6	19
30	Bi-allelic premature truncating variants in LTBP1 cause cutis laxa syndrome. American Journal of Human Genetics, 2021, 108, 1095-1114.	6.2	19
31	Mutations in XRCC1 cause cerebellar ataxia and peripheral neuropathy. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 1230-1232.	1.9	18
32	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
33	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
34	Coding and noncoding variants in EBF3 are involved in HADDS and simplex autism. Human Genomics, 2021, 15, 44.	2.9	16
35	Delineating the molecular and phenotypic spectrum of the SETD1B-related syndrome. Genetics in Medicine, 2021, 23, 2122-2137.	2.4	16
36	MYORG -related disease is associated with central pontine calcifications and atypical parkinsonism. Neurology: Genetics, 2020, 6, e399.	1.9	13

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37	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
38	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
39	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
40	Expanding the phenotype of PIGSâ€associated early onset epileptic developmental encephalopathy. Epilepsia, 2021, 62, e35-e41.	5.1	11
41	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
42	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
43	Prominent and Regressive Brain Developmental Disorders Associated with Nance-Horan Syndrome. Brain Sciences, 2021, 11, 1150.	2.3	10
44	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
45	Homozygous SCN1B variants causing early infantile epileptic encephalopathy 52 affect voltageâ€gated sodium channel function. Epilepsia, 2021, 62, e82-e87.	5.1	9
46	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
47	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
48	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
49	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
50	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
51	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
52	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
53	Analysis of the prion protein gene in multiple system atrophy. Neurobiology of Aging, 2017, 49, 216.e15-216.e18.	3.1	6
54	A de novo truncating mutation in ASXL1 associated with segmental overgrowth. Journal of Genetics, 2019, 98, 1.	0.7	5

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55	A homozygous GDAP2 loss-of-function variant in a patient with adult-onset cerebellar ataxia. Brain, 2020, 143, e49-e49.	7.6	5
56	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
57	Allelic and phenotypic heterogeneity in Junctophillin-3 related neurodevelopmental and movement disorders. European Journal of Human Genetics, 2021, 29, 1027-1031.	2.8	5
58	Early-onset phenotype of bi-allelic GRN mutations. Brain, 2021, 144, e22-e22.	7.6	5
59	Novel variants broaden the phenotypic spectrum of PLEKHG5 â€associated neuropathies. European Journal of Neurology, 2021, 28, 1344-1355.	3.3	4
60	Tay-Sachs Disease: Two Novel Rare HEXA Mutations from Pakistan and Morocco. Klinische Padiatrie, 2021, 233, 226-230.	0.6	4
61	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
62	Identification of common genetic markers of paroxysmal neurological disorders using a network analysis approach. Neurological Sciences, 2020, 41, 851-857.	1.9	3
63	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
64	Novel variants underlying autosomal recessive intellectual disability in Pakistani consanguineous families. BMC Medical Genetics, 2020, 21, 59.	2.1	3
65	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
66	De novo mutation in SLC25A22 gene: expansion of the clinical and electroencephalographic phenotype. Journal of Neurogenetics, 2021, 35, 67-73.	1.4	3
67	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
68	Bi-allelic variants inCHKAcause a neurodevelopmental disorder with epilepsy and microcephaly. Brain, 2022, 145, 1916-1923.	7.6	3
69	Mutations inTAF8cause a neurodegenerative disorder. Brain, 2022, 145, 3022-3034.	7.6	3
70	Age-dependent epileptic encephalopathy associated with an unusual co-occurrence of ZEB2 and SCN1A variants. , 2020, 22, 111-115.		2
71	Myoclonic status epilepticus and cerebellar hypoplasia associated with a novel variant in the GRIA3 gene. Neurogenetics, 2021, , 1.	1.4	2
72	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

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73	Pioneers and Emerging Pediatric Neurologists and Epileptologists in the World. Journal of Pediatric Epilepsy, 2019, 08, 001-001.	0.2	0
74	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