Christos Proukakis

List of Publications by Year
in descending order

61 papers	4,853 citations	147801 31 h-index	168389 53 g-index
67 all docs	67 docs citations	67 times ranked	6216 citing authors

#	Article	IF	Citations
1	Hereditary spastic paraplegia: clinical features and pathogenetic mechanisms. Lancet Neurology, The, 2008, 7, 1127-1138.	10.2	481
2	A novel Î±-synuclein missense mutation in Parkinson disease. Neurology, 2013, 80, 1062-1064.	1.1	396
3	Î±-Synucleinopathy associated with G51D SNCA mutation: a link between Parkinsonâ€™s disease and multiple system atrophy?. Acta Neuropathologica, 2013, 125, 753-769.	7.7	369
4	Infantile-onset symptomatic epilepsy syndrome caused by a homozygous loss-of-function mutation of GM3 synthase. Nature Genetics, 2004, 36, 1225-1229.	21.4	359
5	Heterozygous missense mutations in BSCL2 are associated with distal hereditary motor neuropathy and Silver syndrome. Nature Genetics, 2004, 36, 271-276.	21.4	349
6	SPG20 is mutated in Troyer syndrome, an hereditary spastic paraplegia. Nature Genetics, 2002, 31, 347-348.	21.4	240
7	Evolution of Prodromal Clinical Markers of Parkinson Disease in aGBAMutationâ€“Positive Cohort. JAMA Neurology, 2015, 72, 201.	9.0	180
8	Mutation of FA2H underlies a complicated form of hereditary spastic paraplegia (SPG35). Human Mutation, 2010, 31, E1251-E1260.	2.5	174
9	Genetic and phenotypic characterization of complex hereditary spastic paraplegia. Brain, 2016, 139, 1904-1918.	7.6	170
10	Maspardin Is Mutated in Mast Syndrome, a Complicated Form of Hereditary Spastic Paraplegia Associated with Dementia. American Journal of Human Genetics, 2003, 73, 1147-1156.	6.2	158
11	The identification of a conserved domain in both spartin and spastin, mutated in hereditary spastic paraplegia. Genomics, 2003, 81, 437-441.	2.9	128
12	Is the Transportation Highway the Right Road for Hereditary Spastic Paraplegia?. American Journal of Human Genetics, 2002, 71, 1009-1016.	6.2	119
13	Mutations in B4GALNT1 (GM2 synthase) underlie a new disorder of ganglioside biosynthesis. Brain, 2013, 136, 3618-3624.	7.6	115
14	Hyposmia and cognitive impairment in Gaucher disease patients and carriers. Movement Disorders, 2012, 27, 526-532.	3.9	108
15	Extended phenotypic spectrum of KIF5A mutations. Neurology, 2014, 83, 612-619.	1.1	92
16	Distinct clinical and neuropathological features of G51D SNCA mutation cases compared with SNCA duplication and H50Q mutation. Molecular Neurodegeneration, 2015, 10, 41.	10.8	90
17	Visual short-term memory deficits associated with GBA mutation and Parkinsonâ€™s disease. Brain, 2014, 137, 2303-2311.	7.6	77
18	Defective Mitochondrial mRNA Maturation Is Associated with Spastic Ataxia. American Journal of Human Genetics, 2010, 87, 655-660.	6.2	76

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19	Review: Somatic mutations in neurodegeneration. Neuropathology and Applied Neurobiology, 2018, 44, 267-285.	3.2	75
20	Spastin and microtubules: Functions in health and disease. Journal of Neuroscience Research, 2007, 85, 2778-2782.	2.9	70
21	The H50Q Mutation Induces a 10-fold Decrease in the Solubility of Î±-Synuclein. Journal of Biological Chemistry, 2015, 290, 2395-2404.	3.4	65
22	Evaluation of the detection of GBA missense mutations and other variants using the Oxford Nanopore MinION. Molecular Genetics & Genomic Medicine, 2019, 7, e564.	1.2	65
23	Somatic copy number gains of Î±-synuclein (SNCA) in Parkinsonâ€™s disease and multiple system atrophy brains. Brain, 2018, 141, 2419-2431.	7.6	63
24	A 6.4 Mb Duplication of the Î±-Synuclein Locus Causing Frontotemporal Dementia and Parkinsonism. JAMA Neurology, 2014, 71, 1162.	9.0	60
25	Selective vulnerability in Î±-synucleinopathies. Acta Neuropathologica, 2019, 138, 681-704.	7.7	58
26	Human spastin has multiple microtubule-related functions. Journal of Neurochemistry, 2005, 95, 1411-1420.	3.9	54
27	Somatic alphaâ€synuclein mutations in Parkinson's disease: Hypothesis and preliminary data. Movement Disorders, 2013, 28, 705-712.	3.9	53
28	Troyer syndrome revisited. Journal of Neurology, 2004, 251, 1105-10.	3.6	42
29	A large family with hereditary spastic paraparesis due to a frame shift mutation of the spastin (SPG4) gene: association with multiple sclerosis in two affected siblings and epilepsy in other affected family members. Journal of Neurology, Neurosurgery and Psychiatry, 2001, 71, 788-791.	1.9	41
30	Î±-Synuclein mutations cluster around a putative protein loop. Neuroscience Letters, 2013, 546, 67-70.	2.1	36
31	Investigation of somatic CNVs in brains of synucleinopathy cases using targeted SNCA analysis and single cell sequencing. Acta Neuropathologica Communications, 2019, 7, 219.	5.2	35
32	Two Craniosynostotic Syndrome Loci, Crouzon and Jackson-Weiss, Map to Chromosome 10q23-q26. Genomics, 1994, 22, 418-424.	2.9	33
33	Evolution and clustering of prodromal parkinsonian features in GBA1 carriers. Movement Disorders, 2019, 34, 1365-1373.	3.9	33
34	Somatic mutations in neurodegeneration: An update. Neurobiology of Disease, 2020, 144, 105021.	4.4	32
35	Screening of patients with hereditary spastic paraplegia reveals seven novel mutations in theSPG4 (Spastin) gene. Human Mutation, 2003, 21, 170-170.	2.5	31
36	Detection of novel mutations and review of published data suggests that hereditary spastic paraplegia caused by spastin (SPAST) mutations is found more often in males. Journal of the Neurological Sciences, 2011, 306, 62-65.	0.6	31

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37	Î±-Synuclein structural features inhibit harmful polyunsaturated fatty acid oxidation, suggesting roles in neuroprotection. Journal of Biological Chemistry, 2017, 292, 6927-6937.	3.4	31
38	DNA isolation protocol effects on nuclear DNA analysis by microarrays, droplet digital PCR, and whole genome sequencing, and on mitochondrial DNA copy number estimation. PLoS ONE, 2017, 12, e0180467.	2.5	27
39	No association with common Caucasian genotypes in exons 8, 13 and 14 of the human cytoplasmic dynein heavy chain gene (DNCHC1) and familial motor neuron disorders. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders: Official Publication of the World Federation of Neurology, Research Group on Motor Neuron Diseases, 2003, 4, 150-157.	1.2	26
40	Analysis of Parkinson's disease brainâ€“derived DNA for alphaâ€synuclein coding somatic mutations. Movement Disorders, 2014, 29, 1060-1064.	3.9	22
41	Four novel SPG3A/atlastin mutations identified in autosomal dominant hereditary spastic paraplegia kindreds with intraâ€familial variability in age of onset and complex phenotype. Clinical Genetics, 2009, 75, 485-489.	2.0	21
42	Complex mosaic structural variations in human fetal brains. Genome Research, 2020, 30, 1695-1704.	5.5	21
43	Three novel spastin (SPG4) mutations in families with autosomal dominant hereditary spastic paraplegia. Journal of the Neurological Sciences, 2002, 201, 65-69.	0.6	16
44	A clinical, genetic and candidate gene study of Silver syndrome, a complicated form of hereditary spastic paraplegia. Journal of Neurology, 2004, 251, 1068-74.	3.6	11
45	Copy number variation of LINGO1 in familial dystonic tremor. Neurology: Genetics, 2019, 5, e307.	1.9	8
46	Investigation of Somatic Mutations in Human Brains Targeting Genes Associated With Parkinson's Disease. Frontiers in Neurology, 2020, 11, 570424.	2.4	8
47	A crowdsourced set of curated structural variants for the human genome. PLoS Computational Biology, 2020, 16, e1007933.	3.2	6
48	A large Indian family with rearrangement of chromosome 4p16 and 3p26.3 and divergent clinical presentations. BMC Medical Genetics, 2015, 16, 104.	2.1	5
49	Intronic Haplotypes in the <scp>GBA</scp> Gene Do Not Predict Age at Diagnosis of Parkinson's Disease. Movement Disorders, 2021, 36, 1456-1460.	3.9	5
50	Genetics of <scp>P</scp>arkinson's disease: alphaâ€synuclein and other insights from <scp>G</scp>reece. European Journal of Neurology, 2014, 21, 946-947.	3.3	3
51	The Interaction of Genetic Mutations in PARK2 and FA2H Causes a Novel Phenotype in a Case of Childhood-Onset Movement Disorder. Frontiers in Neurology, 2019, 10, 555.	2.4	3
52	<scp>CAG</scp> Somatic Instability in a Huntington Disease Expansion Carrier Presenting with a Progressive Supranuclear Palsyâ€like Phenotype. Movement Disorders, 2022, 37, 1555-1557.	3.9	3
53	AN AUDIT OF INPATIENT PARKINSON'S DISEASE MANAGEMENT AT BARNET AND CHASE FARM HOSPITALS. Journal of Neurology, Neurosurgery and Psychiatry, 2013, 84, e2.177-e2.	1.9	1
54	Combined Fluorescent In Situ Hybridization (FISH) and Immunofluorescence for the Targeted Detection of Somatic Copy Number Variants in Synucleinopathies. Neuromethods, 2022, , 229-243.	0.3	1

#	Article	IF	Citations
55	Exit, pursued by a bear. Lancet, The, 2008, 372, 262.	13.7	0
56	073â€…Hereditary spastic paraplegia caused by spastin (SPAST, SPG4) mutations is found more often in males: report of novel mutations from one centre, and review of published literature. Journal of Neurology, Neurosurgery and Psychiatry, 2012, 83, e1.19-e1.	1.9	0
57	A PROSPECTIVE AUDIT OF PRESCRIBING IN PARKINSON'S DISEASE. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, e4.153-e4.	1.9	0
58	A crowdsourced set of curated structural variants for the human genome. , 2020, 16, e1007933.		0
59	A crowdsourced set of curated structural variants for the human genome. , 2020, 16, e1007933.		0
60	A crowdsourced set of curated structural variants for the human genome. , 2020, 16, e1007933.		0
61	A crowdsourced set of curated structural variants for the human genome. , 2020, 16, e1007933.		0