Charalampos Tzoulis

List of Publications by Year
in descending order

86	3,012	172386	182361
papers	citations	29 h-index	51 g-index

98	98	98	4208
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	The spectrum of clinical disease caused by the A467T and W748S POLG mutations: a study of 26 cases. Brain, 2006, 129, 1685-1692.	3.7	337
2	Defective mitochondrial DNA homeostasis in the substantia nigra in Parkinson disease. Nature Communications, 2016, 7, 13548.	5.8	197
3	A multicenter study on Leigh syndrome: disease course and predictors of survival. Orphanet Journal of Rare Diseases, 2014, 9, 52.	1.2	182
4	POLG1 mutations cause a syndromic epilepsy with occipital lobe predilection. Brain, 2008, 131, 818-828.	3.7	176
5	The NADPARK study: A randomized phase I trial of nicotinamide riboside supplementation in Parkinsonâ€™s disease. Cell Metabolism, 2022, 34, 396-407.e6.	7.2	111
6	MRI characterisation of adult onset alpha-methylacyl-coA racemase deficiency diagnosed by exome sequencing. Orphanet Journal of Rare Diseases, 2013, 8, 1.	1.2	102
7	Severe nigrostriatal degeneration without clinical parkinsonism in patients with polymerase gamma mutations. Brain, 2013, 136, 2393-2404.	3.7	90
8	Glitazone use associated with reduced risk of Parkinson's disease. Movement Disorders, 2017, 32, 1594-1599.	2.2	90
9	Neuronal complex I deficiency occurs throughout the Parkinsonâ€™s disease brain, but is not associated with neurodegeneration or mitochondrial DNA damage. Acta Neuropathologica, 2018, 135, 409-425.	3.9	89
10	Molecular pathogenesis of polymerase gammaâ€“related neurodegeneration. Annals of Neurology, 2014, 76, 66-81.	2.8	77
11	Localized cerebral energy failure in DNA polymerase gamma-associated encephalopathy syndromes. Brain, 2010, 133, 1428-1437.	3.7	70
12	Practical guidance for CD management involving treatment of botulinum toxin: a consensus statement. Journal of Neurology, 2015, 262, 2201-2213.	1.8	59
13	Phenotype-genotype correlations in Leigh syndrome: new insights from a multicentre study of 96 patients. Journal of Medical Genetics, 2018, 55, 21-27.	1.5	54
14	Rare genetic variation in mitochondrial pathways influences the risk for Parkinson's disease. Movement Disorders, 2018, 33, 1591-1600.	2.2	51
15	Genome-wide histone acetylation analysis reveals altered transcriptional regulation in the Parkinsonâ€™s disease brain. Molecular Neurodegeneration, 2021, 16, 31.	4.4	51
16	The angiogenic switch leads to a metabolic shift in human glioblastoma. Neuro-Oncology, 2017, 19, now175.	0.6	50
17	Serial Diffusion Imaging in a Case of Mitochondrial Encephalomyopathy, Lactic Acidosis, and Stroke-Like Episodes. Stroke, 2009, 40, e15-7.	1.0	49
18	Spastic Paraplegia Type 7 Is Associated with Multiple Mitochondrial DNA Deletions. PLoS ONE, 2014, 9, e86340.	1.1	49

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19	Ultradeep mapping of neuronal mitochondrial deletions in Parkinson's disease. Neurobiology of Aging, 2018, 63, 120-127.	1.5	47
20	Increased levels of cell-free mitochondrial DNA in the cerebrospinal fluid of patients with multiple sclerosis. Mitochondrion, 2017, 34, 32-35.	1.6	46
21	The clinical spectrum and natural history of early-onset diseases due to DNA polymerase gamma mutations. Genetics in Medicine, 2017, 19, 1217-1225.	1.1	45
22	<scp>GBA</scp> and <scp>APOE</scp> Impact Cognitive Decline in Parkinson's Disease: A 10â€Year Populationâ€Based Study. Movement Disorders, 2022, 37, 1016-1027.	2.2	45
23	<scp>ADCK</scp>3 mutations with epilepsy, strokeâ€like episodes and ataxia: a <scp>POLG</scp> mimic?. European Journal of Neurology, 2016, 23, 1188-1194.	1.7	42
24	Common gene expression signatures in Parkinsonâ€™s disease are driven by changes in cell composition. Acta Neuropathologica Communications, 2020, 8, 55.	2.4	38
25	Diseaseâ€specific phenotypes in <scp>iPSC</scp> â€derived neural stem cells with <scp>POLG</scp> mutations. EMBO Molecular Medicine, 2020, 12, e12146.	3.3	38
26	Acute mitochondrial encephalopathy reflects neuronal energy failure irrespective of which genome the genetic defect affects. Brain, 2012, 135, 3627-3634.	3.7	36
27	Betaâ€propeller proteinâ€associated neurodegeneration: a case report and review of the literature. Clinical Case Reports (discontinued), 2018, 6, 353-362.	0.2	34
28	Excellent response of intramedullary Erdheim-Chester disease to vemurafenib: a case report. BMC Research Notes, 2015, 8, 171.	0.6	32
29	Mitochondrial DNA homeostasis is essential for nigrostriatal integrity. Mitochondrion, 2016, 28, 33-37.	1.6	32
30	Dopaminergic and Opioid Pathways Associated with Impulse Control Disorders in Parkinsonâ€™s Disease. Frontiers in Neurology, 2018, 9, 109.	1.1	32
31	Differences in RNA processing underlie the tissue specific phenotype of ISCU myopathy. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2010, 1802, 539-544.	1.8	31
32	Targeting NAD+ in translational research to relieve diseases and conditions of metabolic stress and ageing. Mechanisms of Ageing and Development, 2020, 186, 111208.	2.2	31
33	Understanding the Epilepsy in POLG Related Disease. International Journal of Molecular Sciences, 2017, 18, 1845.	1.8	30
34	Mitochondrial DNA depletion in progressive external ophthalmoplegia caused byPOLG1mutations. Acta Neurologica Scandinavica, 2009, 120, 38-41.	1.0	29
35	Leukoencephalopathy with brainstem and spinal cord involvement caused by a novel mutation in the DARS2 gene. Journal of Neurology, 2012, 259, 292-296.	1.8	29
36	In vitro characterization of six STUB1 variants in spinocerebellar ataxia 16 reveals altered structural properties for the encoded CHIP proteins. Bioscience Reports, 2017, 37, .	1.1	27

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37	Base excision repair causes age-dependent accumulation of single-stranded DNA breaks that contribute to Parkinson disease pathology. Cell Reports, 2021, 36, 109668.	2.9	26
38	A nationwide study of the incidence, prevalence and mortality of Parkinsonâ€™s disease in the Norwegian population. Npj Parkinson's Disease, 2022, 8, 19.	2.5	25
39	Hereditary spastic paraplegia caused by the novel mutation 1047insC in the SPG7 gene. Journal of Neurology, 2008, 255, 1142-1144.	1.8	23
40	Number of CAG repeats in POLG1 and its association with Parkinson disease in the Norwegian population. Mitochondrion, 2012, 12, 640-643.	1.6	23
41	Association of glucocerebrosidase polymorphisms and mutations with dementia in incident Parkinson's disease. Alzheimer's and Dementia, 2018, 14, 1293-1301.	0.4	23
42	Progressive striatal necrosis associated with anti-NMDA receptor antibodies. BMC Neurology, 2013, 13, 55.	0.8	19
43	Novel SACS Mutations Identified by Whole Exome Sequencing in a Norwegian Family with Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay. PLoS ONE, 2013, 8, e66145.	1.1	19
44	Novel SLC19A3 Promoter Deletion and Allelic Silencing in Biotin-Thiamine-Responsive Basal Ganglia Encephalopathy. PLoS ONE, 2016, 11, e0149055.	1.1	18
45	PNKP Mutations Identified by Whole-Exome Sequencing in a Norwegian Patient with Sporadic Ataxia and Edema. Cerebellum, 2017, 16, 272-275.	1.4	17
46	LIVE@Home.Pathâ€”innovating the clinical pathway for home-dwelling people with dementia and their caregivers: study protocol for a mixed-method, stepped-wedge, randomized controlled trial. Trials, 2020, 21, 510.	0.7	16
47	Differential transcript usage in the Parkinsonâ€™s disease brain. PLoS Genetics, 2020, 16, e1009182.	1.5	15
48	HTRA2 p.G399S in Parkinson disease, essential tremor, and tremulous cervical dystonia. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, E2268.	3.3	14
49	Mitochondrial respiratory chain deficiency correlates with the severity of neuropathology in sporadic Creutzfeldt-Jakob disease. Acta Neuropathologica Communications, 2020, 8, 50.	2.4	14
50	No evidence of ischemia in stroke-like lesions of mitochondrial POLG encephalopathy. Mitochondrion, 2017, 32, 10-15.	1.6	13
51	Management of dystonia in Europe: a survey of the European network for the study of the dystonia syndromes. European Journal of Neurology, 2016, 23, 772-779.	1.7	12
52	Simvastatin is associated with decreased risk of <scp>P</scp>arkinson disease. Annals of Neurology, 2017, 81, 329-330.	2.8	12
53	Neuropsychological performance in patients with POLG1 mutations and the syndrome of mitochondrial spinocerebellar ataxia and epilepsy. Epilepsy and Behavior, 2009, 16, 172-174.	0.9	11
54	Meta-analysis of whole-exome sequencing data from two independent cohorts finds no evidence for rare variant enrichment in Parkinson disease associated loci. PLoS ONE, 2020, 15, e0239824.	1.1	11

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55	Using urine to diagnose largeâ€scale mtDNA deletions in adult patients. Annals of Clinical and Translational Neurology, 2020, 7, 1318-1326.	1.7	11
56	Mitochondrial Respiratory Chain Dysfunctionâ€"A Hallmark Pathology of Idiopathic Parkinsonâ€™s Disease?. Frontiers in Cell and Developmental Biology, 2022, 10, 874596.	1.8	11
57	Familial aggregation of Parkinson's disease may affect progression of motor symptoms and dementia. Movement Disorders, 2017, 32, 241-245.	2.2	10
58	Mitochondrial DNA depletion in sporadic inclusion body myositis. Neuromuscular Disorders, 2019, 29, 242-246.	0.3	10
59	MELAS ASSOCIATED WITH MUTATIONS IN THE POLG1 GENE. Neurology, 2008, 70, 1054-1055.	1.5	9
60	Subcellular Parkinsonâ€™s Disease-Specific Alpha-Synuclein Species Show Altered Behavior in Neurodegeneration. Molecular Neurobiology, 2017, 54, 7639-7655.	1.9	9
61	Alzheimer disease associated variants in SORL1 accelerate dementia development in Parkinson disease. Neuroscience Letters, 2018, 674, 123-126.	1.0	9
62	Erdheimâ€"Chester disease presenting with an intramedullary spinal cord lesion. Journal of Neurology, 2012, 259, 2240-2242.	1.8	8
63	The presence of anaemia negatively influences survival in patients with POLG disease. Journal of Inherited Metabolic Disease, 2017, 40, 861-866.	1.7	8
64	3,3â€²-Diaminobenzidine staining interferes with PCR-based DNA analysis. Scientific Reports, 2018, 8, 1272.	1.6	8
65	The impact of common genetic variants in cognitive decline in the first seven years of Parkinsonâ€™s disease: A longitudinal observational study. Neuroscience Letters, 2021, 764, 136243.	1.0	8
66	Poly-ADP-ribose assisted protein localization resolves that DJ-1, but not LRRK2 or Î±-synuclein, is localized to the mitochondrial matrix. PLoS ONE, 2019, 14, e0219909.	1.1	7
67	NSAID use is not associated with Parkinsonâ€™s disease incidence: A Norwegian Prescription Database study. PLoS ONE, 2021, 16, e0256602.	1.1	7
68	Myoclonus-dystonia and epilepsy in a family with a novel epsilon-sarcoglycan mutation. Journal of Neurology, 2014, 261, 358-362.	1.8	6
69	Early Forms of Î±-Synuclein Pathology Are Associated with Neuronal Complex I Deficiency in the Substantia Nigra of Individuals with Parkinsonâ€™s Disease. Biomolecules, 2022, 12, 747.	1.8	6
70	En mann i 50-Ã¥rene med hÃ¸yt ferritinnivÃ¥ og Ã¸kende kognitiv svikt. Tidsskrift for Den Norske Laegeforening, 2015, 135, 1369-1372.	0.2	5
71	Distinct Mitochondrial Remodeling During Mesoderm Differentiation in a Human-Based Stem Cell Model. Frontiers in Cell and Developmental Biology, 2021, 9, 744777.	1.8	5
72	Ultra-deep whole genome bisulfite sequencing reveals a single methylation hotspot in human brain mitochondrial DNA. Epigenetics, 2022, 17, 906-921.	1.3	5

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73	No evidence for rare TRAP1 mutations influencing the risk of idiopathic Parkinsonâ€™s disease. Brain, 2018, 141, e16-e16.	3.7	4
74	Movement disorders in mitochondrial disease: a clinicopathological correlation. Current Opinion in Neurology, 2018, 31, 472-483.	1.8	4
75	Nigrostriatal denervationsineparkinsonism: Table 1. Brain, 2016, 139, e25-e25.	3.7	3
76	Real-World Dosing of OnabotulinumtoxinA and IncobotulinumtoxinA for Cervical Dystonia and Blepharospasm: Results from TRUDOSE and TRUDOSE II. Toxins, 2021, 13, 488.	1.5	2
77	Chip Protein U-Box Domain Truncation Affects Purkinje Neuron Morphology and Leads to Behavioral Changes in Zebrafish. Frontiers in Molecular Neuroscience, 2021, 14, 723912.	1.4	0
78	En kvinne i 70-Ã¥rene med langvarige gangvansker. Tidsskrift for Den Norske Laegeforening, 2015, 135, 1753-1755.	0.2	0
79	Differential transcript usage in the Parkinsonâ€™s disease brain. , 2020, 16, e1009182.		0
80	Differential transcript usage in the Parkinsonâ€™s disease brain. , 2020, 16, e1009182.		0
81	Differential transcript usage in the Parkinsonâ€™s disease brain. , 2020, 16, e1009182.		0
82	Differential transcript usage in the Parkinsonâ€™s disease brain. , 2020, 16, e1009182.		0
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