Simon As Pope

List of Publications by Year
in descending order

17 papers	1,196 citations	687363 13 h-index	940533 16 g-index
18 all docs	18 docs citations	18 times ranked	2397 citing authors

#	Article	IF	Citations
1	Mutations in the histone methyltransferase gene KMT2B cause complex early-onset dystonia. Nature Genetics, 2017, 49, 223-237.	21.4	186
2	Dysregulation of glucose metabolism is an early event in sporadic Parkinson's disease. Neurobiology of Aging, 2014, 35, 1111-1115.	3.1	174
3	Loss ofPLA2G6leads to elevated mitochondrial lipid peroxidation and mitochondrial dysfunction. Brain, 2015, 138, 1801-1816.	7.6	143
4	Oxidative stress and mitochondrial dysfunction in neurodegeneration; cardiolipin a critical target?. Biochimica Et Biophysica Acta - Bioenergetics, 2008, 1777, 794-799.	1.0	129
5	Missense dopamine transporter mutations associate with adult parkinsonism and ADHD. Journal of Clinical Investigation, 2014, 124, 3107-3120.	8.2	129
6	Consensus guideline for the diagnosis and treatment of tetrahydrobiopterin (BH4) deficiencies. Orphanet Journal of Rare Diseases, 2020, 15, 126.	2.7	85
7	Cerebral folate deficiency: Analytical tests and differential diagnosis. Journal of Inherited Metabolic Disease, 2019, 42, 655-672.	3.6	69
8	Clinical, pathological and functional characterization of riboflavin-responsive neuropathy. Brain, 2017, 140, 2820-2837.	7.6	64
9	Glial cells are functionally impaired in juvenile neuronal ceroid lipofuscinosis and detrimental to neurons. Acta Neuropathologica Communications, 2017, 5, 74.	5.2	57
10	PDXK mutations cause polyneuropathy responsive to pyridoxal 5â€²â€phosphate supplementation. Annals of Neurology, 2019, 86, 225-240.	5.3	54
11	Gene therapy restores dopamine transporter expression and ameliorates pathology in iPSC and mouse models of infantile parkinsonism. Science Translational Medicine, 2021, 13, .	12.4	25
12	Analysis of human cerebrospinal fluid monoamines and their cofactors by HPLC. Nature Protocols, 2017, 12, 2359-2366.	12.0	23
13	<scp>DNAJC6</scp> Mutations Disrupt Dopamine Homeostasis in Juvenile <scp>Parkinsonismâ€Dystonia</scp>. Movement Disorders, 2020, 35, 1357-1368.	3.9	22
14	Aromatic <scp>l</scp>-amino acid decarboxylase deficiency: a patient-derived neuronal model for precision therapies. Brain, 2021, 144, 2443-2456.	7.6	16
15	Utility of Whole Blood Thiamine Pyrophosphate Evaluation in TPK1-Related Diseases. Journal of Clinical Medicine, 2019, 8, 991.	2.4	13
16	Role of Intramuscular Levofolinate Administration in the Treatment of Hereditary Folate Malabsorption: Report of Three Cases. JIMD Reports, 2017, 39, 7-12.	1.5	7
17	Cerebrospinal fluid folate, ascorbate, and tetrahydrobiopterin deficiency in superficial siderosis: A new potential mechanism of neurological dysfunction?. Journal of the Neurological Sciences, 2020, 414, 116856.	0.6	0