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