Simon J R Heales

List of Publications by Year
in descending order

77 papers	6,387 citations	101384 36 h-index	74018 75 g-index
83 all docs	83 docs citations	83 times ranked	8478 citing authors

#	Article	IF	Citations
1	Association between mitochondrial dysfunction and severity and outcome of septic shock. Lancet, The, 2002, 360, 219-223.	6.3	1,360
2	Glucocerebrosidase deficiency in substantia nigra of parkinson disease brains. Annals of Neurology, 2012, 72, 455-463.	2.8	473
3	Nitric Oxideâ€Mediated Mitochondrial Damage in the Brain: Mechanisms and Implications for Neurodegenerative Diseases. Journal of Neurochemistry, 1997, 68, 2227-2240.	2.1	458
4	Effect of Peroxynitrite on the Mitochondrial Respiratory Chain: Differential Susceptibility of Neurones and Astrocytes in Primary Culture. Journal of Neurochemistry, 1995, 64, 1965-1972.	2.1	446
5	Mechanisms of action for the medium-chain triglyceride ketogenic diet in neurological and metabolic disorders. Lancet Neurology, The, 2018, 17, 84-93.	4.9	296
6	The monoamine neurotransmitter disorders: an expanding range of neurological syndromes. Lancet Neurology, The, 2011, 10, 721-733.	4.9	290
7	The ketogenic diet component decanoic acid increases mitochondrial citrate synthase and complex I activity in neuronal cells. Journal of Neurochemistry, 2014, 129, 426-433.	2.1	153
8	Nitric oxide-induced mitochondrial dysfunction: implications for neurodegeneration. Free Radical Biology and Medicine, 2003, 34, 287-303.	1.3	152
9	Oxidative stress and mitochondrial dysfunction in neurodegeneration; cardiolipin a critical target?. Biochimica Et Biophysica Acta - Bioenergetics, 2008, 1777, 794-799.	0.5	129
10	Identification of novel CSF biomarkers for neurodegeneration and their validation by a high-throughput multiplexed targeted proteomic assay. Molecular Neurodegeneration, 2015, 10, 64.	4.4	121
11	Determination of Coenzyme Q10 Status in Blood Mononuclear Cells, Skeletal Muscle, and Plasma by HPLC with Di-Propoxy-Coenzyme Q10 as an Internal Standard. Clinical Chemistry, 2005, 51, 2380-2382.	1.5	119
12	Mutations in PROSC Disrupt Cellular Pyridoxal Phosphate Homeostasis and Cause Vitamin-B6-Dependent Epilepsy. American Journal of Human Genetics, 2016, 99, 1325-1337.	2.6	118
13	Glycine decarboxylase deficiency causes neural tube defects and features of non-ketotic hyperglycinemia in mice. Nature Communications, 2015, 6, 6388.	5.8	116
14	Astrocyte-Derived Nitric Oxide Causes Both Reversible and Irreversible Damage to the Neuronal Mitochondrial Respiratory Chain. Journal of Neurochemistry, 2002, 75, 694-700.	2.1	115
15	Neonatal presentation of coenzyme Q10 deficiency. Journal of Pediatrics, 2001, 139, 456-458.	0.9	112
16	Peroxynitrite and Brain Mitochondria: Evidence for Increased Proton Leak. Journal of Neurochemistry, 1998, 70, 2195-2202.	2.1	110
17	Mitochondrial dysfunction associated with neuronal death following status epilepticus in rat. Epilepsy Research, 2002, 48, 157-168.	0.8	96
18	Neurodegeneration or Neuroprotection: The Pivotal Role of Astrocytes. Neurochemical Research, 2004, 29, 513-519.	1.6	93

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19	Mitochondrial dysfunction is an important cause of neurological deficits in an inflammatory model of multiple sclerosis. Scientific Reports, 2016, 6, 33249.	1.6	89
20	CNS Langerhans cell histiocytosis: Common hematopoietic origin for LCHâ€associated neurodegeneration and mass lesions. Cancer, 2018, 124, 2607-2620.	2.0	73
21	Impairment of brain mitochondrial function by reactive nitrogen species: the role of glutathione in dictating susceptibility. Neurochemistry International, 2002, 40, 469-474.	1.9	71
22	Cerebral folate deficiency: Analytical tests and differential diagnosis. Journal of Inherited Metabolic Disease, 2019, 42, 655-672.	1.7	69
23	Clinical, pathological and functional characterization of riboflavin-responsive neuropathy. Brain, 2017, 140, 2820-2837.	3.7	64
24	The Identification of New Biomarkers for Identifying and Monitoring Kidney Disease and Their Translation into a Rapid Mass Spectrometry-Based Test: Evidence of Presymptomatic Kidney Disease in Pediatric Fabry and Type-I Diabetic Patients. Journal of Proteome Research, 2013, 12, 2013-2021.	1.8	63
25	Induction of mitochondrial oxidative stress in astrocytes by nitric oxide precedes disruption of energy metabolism. Journal of Neurochemistry, 2005, 95, 388-395.	2.1	57
26	Drug-Induced Mitochondrial Toxicity. Drug Safety, 2016, 39, 661-674.	1.4	56
27	Pretreatment of Astrocytes with Interferonâ€Î±/Î² Prevents Neuronal Mitochondrial Respiratory Chain Damage. Journal of Neurochemistry, 1998, 70, 432-434.	2.1	54
28	PDXK mutations cause polyneuropathy responsive to pyridoxal 5â€²â€phosphate supplementation. Annals of Neurology, 2019, 86, 225-240.	2.8	54
29	Oxidative Stress: Mechanistic Insights into Inherited Mitochondrial Disorders and Parkinsonâ€™s Disease. Journal of Clinical Medicine, 2017, 6, 100.	1.0	52
30	Inhibition of monocyte luminol-dependent chemiluminescence by tetrahydrobiopterin, and the free radical oxidation of tetrahydrobiopterin, dihydrobiopterin and dihydroneopterin. Cell Biochemistry and Function, 1988, 6, 191-195.	1.4	46
31	Paroxysmal dystonic choreoathetosis: Clinical features and investigation of pathophysiology in a large family. Movement Disorders, 2000, 15, 648-657.	2.2	46
32	The pleiotropic effects of decanoic acid treatment on mitochondrial function in fibroblasts from patients with complex I deficient Leigh syndrome. Journal of Inherited Metabolic Disease, 2016, 39, 415-426.	1.7	44
33	Neuronal decanoic acid oxidation is markedly lower than that of octanoic acid: A mechanistic insight into the mediumâ€chain triglyceride ketogenic diet. Epilepsia, 2017, 58, 1423-1429.	2.6	43
34	Glutathione depletion is accompanied by increased neuronal nitric oxide synthase activity. Neurochemical Research, 1996, 21, 35-39.	1.6	40
35	Secondary neurotransmitter deficiencies in epilepsy caused by voltage-gated sodium channelopathies: A potential treatment target?. Molecular Genetics and Metabolism, 2016, 117, 42-48.	0.5	40
36	Neurochemical Effects Following Peripheral Administration of Tetrahydropterin Derivatives to the hphâ€1 Mouse. Journal of Neurochemistry, 1996, 66, 1150-1156.	2.1	39

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37	Can folic acid have a role in mitochondrial disorders?. Drug Discovery Today, 2015, 20, 1349-1354.	3.2	38
38	Biochemical Diagnosis of Coenzyme Q₁₀ Deficiency. Molecular Syndromology, 2014, 5, 147-155.	0.3	35
39	Argininosuccinic aciduria fosters neuronal nitrosative stress reversed by Asl gene transfer. Nature Communications, 2018, 9, 3505.	5.8	34
40	Defining the Newborn Blood Spot Screening Reference Interval for TSH: Impact of Ethnicity. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 3445-3449.	1.8	29
41	Increased glucocerebrosidase (GBA) 2 activity in GBA1 deficient mice brains and in Gaucher leucocytes. Journal of Inherited Metabolic Disease, 2013, 36, 869-872.	1.7	28
42	Gene therapy restores dopamine transporter expression and ameliorates pathology in iPSC and mouse models of infantile parkinsonism. Science Translational Medicine, 2021, 13, .	5.8	25
43	An LCâ€“MS/MS-Based Method for the Quantification of Pyridox(am)ine 5â€²-Phosphate Oxidase Activity in Dried Blood Spots from Patients with Epilepsy. Analytical Chemistry, 2017, 89, 8892-8900.	3.2	24
44	Proposed Therapeutic Range of Treosulfan in Reduced Toxicity Pediatric Allogeneic Hematopoietic Stem Cell Transplant Conditioning: Results From a Prospective Trial. Clinical Pharmacology and Therapeutics, 2020, 108, 264-273.	2.3	22
45	CoQ10 Deficient Endothelial Cell Culture Model for the Investigation of CoQ10 Bloodâ€“Brain Barrier Transport. Journal of Clinical Medicine, 2020, 9, 3236.	1.0	22
46	<scp>DNAJC6</scp> Mutations Disrupt Dopamine Homeostasis in Juvenile <scp>Parkinsonismâ€Dystonia</scp>. Movement Disorders, 2020, 35, 1357-1368.	2.2	22
47	Atypical parkinsonism with apraxia and supranuclear gaze abnormalities in type 1 Gaucher disease. Expanding the spectrum: Case report and literature review. Movement Disorders, 2010, 25, 1506-1509.	2.2	21
48	Dopamine but not l-dopa stimulates neural glutathione metabolism. Potential implications for Parkinsonâ€™s and other dopamine deficiency states. Neurochemistry International, 2013, 62, 684-694.	1.9	21
49	Blood Mononuclear Cell Mitochondrial Respiratory Chain Complex IV Activity is Decreased in Multiple Sclerosis Patients: Effects of Î²-Interferon Treatment. Journal of Clinical Medicine, 2018, 7, 36.	1.0	21
50	The Effect of Cellular Coenzyme Q10 Deficiency on Lysosomal Acidification. Journal of Clinical Medicine, 2020, 9, 1923.	1.0	21
51	Reversible parkinsonism following heroin pyrolysate inhalation is associated with tetrahydrobiopterin deficiency. Movement Disorders, 2004, 19, 1248-1251.	2.2	20
52	Proteomic Discovery and Development of a Multiplexed Targeted MRM-LC-MS/MS Assay for Urine Biomarkers of Extracellular Matrix Disruption in Mucopolysaccharidoses I, II, and VI. Analytical Chemistry, 2015, 87, 12238-12244.	3.2	20
53	Levels of 5-methyltetrahydrofolate and ascorbic acid in cerebrospinal fluid are correlated: Implications for the accelerated degradation of folate by reactive oxygen species. Neurochemistry International, 2013, 63, 750-755.	1.9	17
54	Ketogenic diet in the treatment of epilepsy in children under the age of 2 years: study protocol for a randomised controlled trial. Trials, 2017, 18, 195.	0.7	17

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55	K.Vita: a feasibility study of a blend of medium chain triglycerides to manage drug-resistant epilepsy. Brain Communications, 2021, 3, fcab160.	1.5	17
56	Mitochondrial Disease: A Historical, Biochemical, and London Perspective. Neurochemical Research, 2004, 29, 483-491.	1.6	16
57	Aromatic <scp>l</scp>-amino acid decarboxylase deficiency: a patient-derived neuronal model for precision therapies. Brain, 2021, 144, 2443-2456.	3.7	16
58	Depletion of glutathione does not affect electron transport chain complex activity in brain mitochondria: Implications for Parkinson disease and postmortem studies. Free Radical Biology and Medicine, 2011, 50, 899-902.	1.3	15
59	The novel R347g pathogenic mutation of aromatic amino acid decarboxylase provides additional molecular insights into enzyme catalysis and deficiency. Biochimica Et Biophysica Acta - Proteins and Proteomics, 2016, 1864, 676-682.	1.1	15
60	Inhibition of neuronal mitochondrial complex I or lysosomal glucocerebrosidase is associated with increased dopamine and serotonin turnover. Neurochemistry International, 2017, 109, 94-100.	1.9	14
61	Assessment of Mitochondrial Dysfunction in Experimental Autoimmune Encephalomyelitis (EAE) Models of Multiple Sclerosis. International Journal of Molecular Sciences, 2019, 20, 4975.	1.8	14
62	An examination of biochemical parameters and their association with response to ketogenic dietary therapies. Epilepsia, 2017, 58, 893-900.	2.6	13
63	Utility of Whole Blood Thiamine Pyrophosphate Evaluation in TPK1-Related Diseases. Journal of Clinical Medicine, 2019, 8, 991.	1.0	13
64	Diagnosing Mitochondrial Disorders Remains Challenging in the Omics Era. Neurology: Genetics, 2021, 7, e597.	0.9	13
65	Biochemical characterization of proliferative and differentiated SH-SY5Y cell line as a model for Parkinson's disease. Neurochemistry International, 2021, 145, 105009.	1.9	11
66	Coenzyme Q10 and Pyridoxal Phosphate Deficiency Is a Common Feature in Mucopolysaccharidosis Type III. JIMD Reports, 2015, 25, 1-7.	0.7	8
67	Protective effects of medium chain triglyceride diet in a mouse model of Dravet syndrome. Epilepsia, 2021, 62, 3131-3142.	2.6	6
68	Special Issue in Honour of Professor Juan Bolanos. Neurochemical Research, 2021, 46, 1-2.	1.6	5
69	53 Astrocytic mitochondrial respiratory chain damage: effect on cellular ATP levels. Biochemical Society Transactions, 1998, 26, S346-S346.	1.6	4
70	Inhibition of Mitochondrial Complex I Impairs Release of Î±-Galactosidase by Jurkat Cells. International Journal of Molecular Sciences, 2019, 20, 4349.	1.8	2
71	Earwax: A potentially useful medium to identify inborn errors of metabolism?. JIMD Reports, 2020, 52, 72-78.	0.7	2
72	Effect of astrocytic nitric oxide production on neuronal mitochondrial activity. Biochemical Society Transactions, 1997, 25, 405S-405S.	1.6	1

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73	pH AND BRAIN FUNCTION.. Brain, 1999, 122, 1794-1796.	3.7	1
74	Cerebellar and Midbrain Lysosomal Enzyme Deficiency in Isolated Dystonia. Movement Disorders, 2022, 37, 875-877.	2.2	1
75	52 Astrocytic mitochondrial respiratory chain complex I activity and cellular GSH status. Biochemical Society Transactions, 1998, 26, S345-S345.	1.6	0
76	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.	0.3	0
77	Differential activity of glucocerebrosidase in neurons and astrocytes; implications for evaluating tissue homogenates derived from Parkinsonâ€˜s disease brains. Journal of the Neurological Sciences, 2020, 418, 117097.	0.3	0