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	Cerebellar and Midbrain Lysosomal Enzyme Deficiency in Isolated Dystonia. Movement Disorders, 2022, 37, 875-877.	2.2	1
2	Special Issue in Honour of Professor Juan Bolanos. Neurochemical Research, 2021, 46, 1-2.	1.6	5
3	Aromatic <scp>l</scp>-amino acid decarboxylase deficiency: a patient-derived neuronal model for precision therapies. Brain, 2021, 144, 2443-2456.	3.7	16
4	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
5	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
6	Diagnosing Mitochondrial Disorders Remains Challenging in the Omics Era. Neurology: Genetics, 2021, 7, e597.	0.9	13
7	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
8	Protective effects of medium chain triglyceride diet in a mouse model of Dravet syndrome. Epilepsia, 2021, 62, 3131-3142.	2.6	6
9	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
10	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
11	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
12	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
13	<scp>DNAJC6</scp> Mutations Disrupt Dopamine Homeostasis in Juvenile <scp>Parkinsonismâ€Dystonia</scp>. Movement Disorders, 2020, 35, 1357-1368.	2.2	22
14	The Effect of Cellular Coenzyme Q10 Deficiency on Lysosomal Acidification. Journal of Clinical Medicine, 2020, 9, 1923.	1.0	21
15	Earwax: A potentially useful medium to identify inborn errors of metabolism?. JIMD Reports, 2020, 52, 72-78.	0.7	2
16	Utility of Whole Blood Thiamine Pyrophosphate Evaluation in TPK1-Related Diseases. Journal of Clinical Medicine, 2019, 8, 991.	1.0	13
17	Assessment of Mitochondrial Dysfunction in Experimental Autoimmune Encephalomyelitis (EAE) Models of Multiple Sclerosis. International Journal of Molecular Sciences, 2019, 20, 4975.	1.8	14
18	Inhibition of Mitochondrial Complex I Impairs Release of Î±-Galactosidase by Jurkat Cells. International Journal of Molecular Sciences, 2019, 20, 4349.	1.8	2

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19	PDXK mutations cause polyneuropathy responsive to pyridoxal 5â€²â€phosphate supplementation. Annals of Neurology, 2019, 86, 225-240.	2.8	54
20	Cerebral folate deficiency: Analytical tests and differential diagnosis. Journal of Inherited Metabolic Disease, 2019, 42, 655-672.	1.7	69
21	CNS Langerhans cell histiocytosis: Common hematopoietic origin for LCHâ€associated neurodegeneration and mass lesions. Cancer, 2018, 124, 2607-2620.	2.0	73
22	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
23	Argininosuccinic aciduria fosters neuronal nitrosative stress reversed by Asl gene transfer. Nature Communications, 2018, 9, 3505.	5.8	34
24	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
25	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
26	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
27	An examination of biochemical parameters and their association with response to ketogenic dietary therapies. Epilepsia, 2017, 58, 893-900.	2.6	13
28	Clinical, pathological and functional characterization of riboflavin-responsive neuropathy. Brain, 2017, 140, 2820-2837.	3.7	64
29	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
30	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
31	Oxidative Stress: Mechanistic Insights into Inherited Mitochondrial Disorders and Parkinsonâ€™s Disease. Journal of Clinical Medicine, 2017, 6, 100.	1.0	52
32	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
33	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
34	Mitochondrial dysfunction is an important cause of neurological deficits in an inflammatory model of multiple sclerosis. Scientific Reports, 2016, 6, 33249.	1.6	89
35	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
36	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

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37	Drug-Induced Mitochondrial Toxicity. Drug Safety, 2016, 39, 661-674.	1.4	56
38	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
39	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
40	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
41	Glycine decarboxylase deficiency causes neural tube defects and features of non-ketotic hyperglycinemia in mice. Nature Communications, 2015, 6, 6388.	5.8	116
42	Can folic acid have a role in mitochondrial disorders?. Drug Discovery Today, 2015, 20, 1349-1354.	3.2	38
43	Coenzyme Q10 and Pyridoxal Phosphate Deficiency Is a Common Feature in Mucopolysaccharidosis Type III. JIMD Reports, 2015, 25, 1-7.	0.7	8
44	Biochemical Diagnosis of Coenzyme Q₁₀ Deficiency. Molecular Syndromology, 2014, 5, 147-155.	0.3	35
45	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
46	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
47	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
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	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
50	Glucocerebrosidase deficiency in substantia nigra of parkinson disease brains. Annals of Neurology, 2012, 72, 455-463.	2.8	473
51	The monoamine neurotransmitter disorders: an expanding range of neurological syndromes. Lancet Neurology, The, 2011, 10, 721-733.	4.9	290
52	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
53	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
54	Oxidative stress and mitochondrial dysfunction in neurodegeneration; cardiolipin a critical target?. Biochimica Et Biophysica Acta - Bioenergetics, 2008, 1777, 794-799.	0.5	129

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55	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
56	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
57	Mitochondrial Disease: A Historical, Biochemical, and London Perspective. Neurochemical Research, 2004, 29, 483-491.	1.6	16
58	Neurodegeneration or Neuroprotection: The Pivotal Role of Astrocytes. Neurochemical Research, 2004, 29, 513-519.	1.6	93
59	Reversible parkinsonism following heroin pyrolysate inhalation is associated with tetrahydrobiopterin deficiency. Movement Disorders, 2004, 19, 1248-1251.	2.2	20
60	Nitric oxide-induced mitochondrial dysfunction: implications for neurodegeneration. Free Radical Biology and Medicine, 2003, 34, 287-303.	1.3	152
61	Association between mitochondrial dysfunction and severity and outcome of septic shock. Lancet, The, 2002, 360, 219-223.	6.3	1,360
62	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
63	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
64	Mitochondrial dysfunction associated with neuronal death following status epilepticus in rat. Epilepsy Research, 2002, 48, 157-168.	0.8	96
65	Neonatal presentation of coenzyme Q10 deficiency. Journal of Pediatrics, 2001, 139, 456-458.	0.9	112
66	Paroxysmal dystonic choreoathetosis: Clinical features and investigation of pathophysiology in a large family. Movement Disorders, 2000, 15, 648-657.	2.2	46
67	pH AND BRAIN FUNCTION.. Brain, 1999, 122, 1794-1796.	3.7	1
68	52 Astrocytic mitochondrial respiratory chain complex I activity and cellular GSH status. Biochemical Society Transactions, 1998, 26, S345-S345.	1.6	0
69	53 Astrocytic mitochondrial respiratory chain damage: effect on cellular ATP levels. Biochemical Society Transactions, 1998, 26, S346-S346.	1.6	4
70	Pretreatment of Astrocytes with Interferonâ€Î±/Î² Prevents Neuronal Mitochondrial Respiratory Chain Damage. Journal of Neurochemistry, 1998, 70, 432-434.	2.1	54
71	Peroxynitrite and Brain Mitochondria: Evidence for Increased Proton Leak. Journal of Neurochemistry, 1998, 70, 2195-2202.	2.1	110
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	Nitric Oxideâ€Mediated Mitochondrial Damage in the Brain: Mechanisms and Implications for Neurodegenerative Diseases. Journal of Neurochemistry, 1997, 68, 2227-2240.	2.1	458
74	Glutathione depletion is accompanied by increased neuronal nitric oxide synthase activity. Neurochemical Research, 1996, 21, 35-39.	1.6	40
75	Neurochemical Effects Following Peripheral Administration of Tetrahydropterin Derivatives to the hphâ€1 Mouse. Journal of Neurochemistry, 1996, 66, 1150-1156.	2.1	39
76	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
77	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