Robert D S Pitceathly

List of Publications by Year
in descending order

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Version: 2024-02-01

79 papers	2,131 citations	218381 26 h-index	253896 43 g-index
85 all docs	85 docs citations	85 times ranked	2958 citing authors

#	Article	IF	Citations
1	The UK MRC Mitochondrial Disease Patient Cohort Study: clinical phenotypes associated with the m.3243A>G mutation–implications for diagnosis and management. Journal of Neurology, Neurosurgery and Psychiatry, 2013, 84, 936-938.	0.9	193
2	Corneal confocal microscopy: A novel means to detect nerve fibre damage in idiopathic small fibre neuropathy. Experimental Neurology, 2010, 223, 245-250.	2.0	166
3	Cardiolipin, Mitochondria, and Neurological Disease. Trends in Endocrinology and Metabolism, 2021, 32, 224-237.	3.1	113
4	NDUFA4 Mutations Underlie Dysfunction of a Cytochrome c Oxidase Subunit Linked to Human Neurological Disease. Cell Reports, 2013, 3, 1795-1805.	2.9	104
5	Rhabdomyolysis: a genetic perspective. Orphanet Journal of Rare Diseases, 2015, 10, 51.	1.2	101
6	Genetic dysfunction of MT-ATP6 causes axonal Charcot-Marie-Tooth disease. Neurology, 2012, 79, 1145-1154.	1.5	97
7	Adults with RRM2B-related mitochondrial disease have distinct clinical and molecular characteristics. Brain, 2012, 135, 3392-3403.	3.7	70
8	Consensus-based statements for the management of mitochondrial stroke-like episodes. Wellcome Open Research, 2019, 4, 201.	0.9	66
9	Single deletions in mitochondrial DNA â€“ Molecular mechanisms and disease phenotypes in clinical practice. Neuromuscular Disorders, 2012, 22, 577-586.	0.3	62
10	Kearns-Sayre syndrome caused by defective R1/p53R2 assembly. Journal of Medical Genetics, 2011, 48, 610-617.	1.5	49
11	Pathological ribonuclease H1 causes R-loop depletion and aberrant DNA segregation in mitochondria. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, E4276-85.	3.3	48
12	MT-ND5 Mutation Exhibits Highly Variable Neurological Manifestations at Low Mutant Load. EBioMedicine, 2018, 30, 86-93.	2.7	47
13	Peripheral neuropathy predicts nuclear gene defect in patients with mitochondrial ophthalmoplegia. Brain, 2014, 137, 3200-3212.	3.7	45
14	Moving towards clinical trials for mitochondrial diseases. Journal of Inherited Metabolic Disease, 2021, 44, 22-41.	1.7	45
15	NDUFA4 (Renamed COXFA4) Is a Cytochrome-c Oxidase Subunit. Trends in Endocrinology and Metabolism, 2018, 29, 452-454.	3.1	44
16	Safety of drug use in patients with a primary mitochondrial disease: An international Delphiâ€based consensus. Journal of Inherited Metabolic Disease, 2020, 43, 800-818.	1.7	42
17	Use of whole genome sequencing to determine genetic basis of suspected mitochondrial disorders: cohort study. BMJ, The, 2021, 375, e066288.	3.0	42
18	The urinary proteome and metabonome differ from normal in adults with mitochondrial disease. Kidney International, 2015, 87, 610-622.	2.6	41

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19	CAV3 mutations causing exercise intolerance, myalgia and rhabdomyolysis: Expanding the phenotypic spectrum of caveolinopathies. Neuromuscular Disorders, 2016, 26, 504-510.	0.3	38
20	A Clinical, Neuropathological and Genetic Study of Homozygous A467T POLG-Related Mitochondrial Disease. PLoS ONE, 2016, 11, e0145500.	1.1	36
21	Novel C12orf65 mutations in patients with axonal neuropathy and optic atrophy. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 486-492.	0.9	35
22	Pathogenic variants in MTâ€ATP6: A United Kingdomâ€“based mitochondrial disease cohort study. Annals of Neurology, 2019, 86, 310-315.	2.8	33
23	MRPS25 mutations impair mitochondrial translation and cause encephalomyopathy. Human Molecular Genetics, 2019, 28, 2711-2719.	1.4	33
24	Mitochondrial myopathies in adults and children. Current Opinion in Neurology, 2014, 27, 576-582.	1.8	32
25	Applying genomic and transcriptomic advances to mitochondrial medicine. Nature Reviews Neurology, 2021, 17, 215-230.	4.9	32
26	Inflammation causes remodeling of mitochondrial cytochrome c oxidase mediated by the bifunctional gene C15orf48. Science Advances, 2021, 7, eabl5182.	4.7	29
27	COX10Mutations Resulting in Complex Multisystem Mitochondrial Disease That Remains Stable Into Adulthood. JAMA Neurology, 2013, 70, 1556-61.	4.5	27
28	Interventions for dysphagia in long-term, progressive muscle disease. The Cochrane Library, 2016, 2016, CD004303.	1.5	27
29	Chronic pain is common in mitochondrial disease. Neuromuscular Disorders, 2020, 30, 413-419.	0.3	26
30	Metabolic shift underlies recovery in reversible infantile respiratory chain deficiency. EMBO Journal, 2020, 39, e105364.	3.5	26
31	Forecasting stroke-like episodes and outcomes in mitochondrial disease. Brain, 2022, 145, 542-554.	3.7	25
32	Extra-ocular muscle MRI in genetically-defined mitochondrial disease. European Radiology, 2016, 26, 130-137.	2.3	24
33	Novel Biallelic NSUN3 Variants Cause Early-Onset Mitochondrial Encephalomyopathy and Seizures. Journal of Molecular Neuroscience, 2020, 70, 1962-1965.	1.1	24
34	Expanding the molecular and phenotypic spectrum of truncating MT-ATP6 mutations. Neurology: Genetics, 2020, 6, e381.	0.9	21
35	Drug repurposing in neurological diseases: an integrated approach to reduce trial and error. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, 1270-1275.	0.9	20
36	Clinicopathologic and molecular spectrum of RNASEH1 -related mitochondrial disease. Neurology: Genetics, 2017, 3, e149.	0.9	19

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37	Constitutive activation of the PI3K-Akt-mTORC1 pathway sustains the m.3243â€‰Aâ€‰>â€‰G mtDNA mutation. Nature Communications, 2021, 12, 6409.	5.8	19
38	CSF lactate. Practical Neurology, 2020, 20, 320-323.	0.5	18
39	Primary mitochondrial diseases increase susceptibility to bipolar affective disorder. Journal of Neurology, Neurosurgery and Psychiatry, 2020, 91, 892-894.	0.9	17
40	Distal myopathy with cachexia: an unrecognised phenotype caused by dominantly-inherited mitochondrial polymerase Î³ mutations. Journal of Neurology, Neurosurgery and Psychiatry, 2013, 84, 107-110.	0.9	16
41	Utility of Whole Blood Thiamine Pyrophosphate Evaluation in TPK1-Related Diseases. Journal of Clinical Medicine, 2019, 8, 991.	1.0	13
42	Differential phenotypic expression of a novel PDHA1 mutation in a female monozygotic twin pair. Human Genetics, 2019, 138, 1313-1322.	1.8	12
43	Autosomal dominant optic atrophy and cataract â€œplusâ€•phenotype including axonal neuropathy. Neurology: Genetics, 2019, 5, e322.	0.9	12
44	Mitochondrial <scp>D</scp><scp>NA</scp> Analysis from Exome Sequencing Data Improves Diagnostic Yield in Neurological Diseases. Annals of Neurology, 2021, 89, 1240-1247.	2.8	12
45	2-Deoxy-D-glucose couples mitochondrial DNA replication with mitochondrial fitness and promotes the selection of wild-type over mutant mitochondrial DNA. Nature Communications, 2021, 12, 6997.	5.8	12
46	Vestibular dysfunction: a frequent problem for adults with mitochondrial disease. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, 838-841.	0.9	11
47	Toward a mtDNA locus-specific mutation database using the LOVD platform. Human Mutation, 2012, 33, 1352-1358.	1.1	8
48	Homozygous R627W mutations in POLG cause mitochondrial DNA depletion leading to encephalopathy, seizures and stroke-like episodes. Mitochondrion, 2019, 48, 78-83.	1.6	8
49	Adult-onset Leigh syndrome linked to the novel stop codon mutation m.6579G>A in MT-CO1. Mitochondrion, 2019, 47, 294-297.	1.6	8
50	Reducing Intrathecal Baclofen Related Infections: Service Evaluation and Best Practice Guidelines. Neuromodulation, 2020, 23, 991-995.	0.4	8
51	Copper deficiency: an unusual case of myelopathy with neuropathy. Annals of Clinical Biochemistry, 2008, 45, 434-435.	0.8	7
52	Urogenital symptoms in mitochondrial disease: overlooked and undertreated. European Journal of Neurology, 2019, 26, 1111-1120.	1.7	7
53	Use of Twitter in Neurology: Boon or Bane?. Journal of Medical Internet Research, 2021, 23, e25229.	2.1	7
54	Mitochondrial Strokes: Diagnostic Challenges and Chameleons. Genes, 2021, 12, 1643.	1.0	7

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55	COVID-19â€“Related Outcomes in Primary Mitochondrial Diseases. Neurology, 2022, 98, 576-582.	1.5	7
56	Muscle MRI in Bethlem myopathy. BMJ Case Reports, 2013, 2013, bcr2013008596-bcr2013008596.	0.2	6
57	Effects of ketosis in mitochondrial myopathy: potential benefits of a mitotoxic diet. EMBO Molecular Medicine, 2016, 8, 1231-1233.	3.3	4
58	Long-term Safety and Efficacy of Mexiletine in Myotonic Dystrophy Types 1 and 2. Neurology: Clinical Practice, 2021, 11, e682-e685.	0.8	4
59	RRM2B-Related Mitochondrial Disease. , 2013, , 171-182.		3
60	Mitochondrial Extrapyramidal Syndromes. JAMA Neurology, 2016, 73, 630.	4.5	3
61	Mitochondrial DNA variants in genomic data: diagnostic uplifts and predictive implications. Nature Reviews Genetics, 2021, 22, 547-548.	7.7	3
62	Whole-genome sequencing and the clinician: a tale of two cities. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 1012-1015.	0.9	2
63	Multisystem mitochondrial disease caused by a rare m.10038G>A mitochondrial tRNA ^{Gly} (MT-TG) variant. Neurology: Genetics, 2020, 6, e413.	0.9	2
64	Comment on â€œA severe linezolidâ€induced rhabdomyolysis and lactic acidosis in Leigh syndromeâ€. Journal of Inherited Metabolic Disease, 2021, 44, 6-7.	1.7	2
65	Iterative Reanalysis of Hypertrophic Cardiomyopathy Exome Data Reveals Causative Pathogenic Mitochondrial DNA Variants. Circulation Genomic and Precision Medicine, 2021, 14, e003388.	1.6	2
66	Self-reported postural symptoms predict vestibular dysfunction and falls in patients with multi-sensory impairment. Journal of Neurology, 2022, 269, 2788-2791.	1.8	2
67	THE MEDICAL RESEARCH COUNCIL NEUROMUSCULAR CENTRE FOR TRANSLATIONAL RESEARCH MITOCHONDRIAL DISEASE PATIENT COHORT STUDY UK: FROM CONCEPTUALISATION TO UTILISATION. Journal of Neurology, Neurosurgery and Psychiatry, 2013, 84, e2.188-e2.	0.9	1
68	Glycogen storage disease type XV: A case report. Neuromuscular Disorders, 2015, 25, S221.	0.3	1
69	Uniparental isodisomy of chromosome 2 causing MRPL44-related multisystem mitochondrial disease. Molecular Biology Reports, 2021, 48, 2093-2104.	1.0	1
70	Cardiac manifestations of mitochondrial disorders: reply. European Journal of Heart Failure, 2010, 12, 637-638.	2.9	0
71	A dizzy and disorientated DJ. Practical Neurology, 2011, 11, 252-255.	0.5	0
72	Neurology â€“ the Peripheral Nervous System â€“ Muscle and Nerve. , 0, , 84-94.		0

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73	Mitochondrial DNA-related diseases associated with single large-scale deletions and point mutations. , 2020, , 353-374.		0
74	Delayed diagnoses of mitochondrial cytopathies in patients presenting with end stage kidney disease: two case reports. BMC Nephrology, 2020, 21, 361.	0.8	0
75	Mitochondrial disease and COVID-19: An international cohort study confirms risks and long-term outcomes. Journal of the Neurological Sciences, 2021, 429, 119358.	0.3	0
76	Introducing The International Centre for Genomic Medicine in Neuromuscular Diseases (ICGNMD). Journal of the Neurological Sciences, 2021, 429, 117765.	0.3	0
77	Self-reported postural symptoms predict vestibular dysfunction and falls in mitochondrial disease patients with multi-sensory impairment. Journal of the Neurological Sciences, 2021, 429, 117858.	0.3	0
78	When the Wind Comes Back. , 2017, , 207-209.		0
79	When the Wind Does Not Come Back. , 2017, , 211-213.		0