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List of Publications by Year
in descending order

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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	Forecasting stroke-like episodes and outcomes in mitochondrial disease. Brain, 2022, 145, 542-554.	3.7	25
2	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
3	COVID-19â€“Related Outcomes in Primary Mitochondrial Diseases. Neurology, 2022, 98, 576-582.	1.5	7
4	Moving towards clinical trials for mitochondrial diseases. Journal of Inherited Metabolic Disease, 2021, 44, 22-41.	1.7	45
5	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
6	Applying genomic and transcriptomic advances to mitochondrial medicine. Nature Reviews Neurology, 2021, 17, 215-230.	4.9	32
7	Long-term Safety and Efficacy of Mexiletine in Myotonic Dystrophy Types 1 and 2. Neurology: Clinical Practice, 2021, 11, e682-e685.	0.8	4
8	Uniparental isodisomy of chromosome 2 causing MRPL44-related multisystem mitochondrial disease. Molecular Biology Reports, 2021, 48, 2093-2104.	1.0	1
9	Cardiolipin, Mitochondria, and Neurological Disease. Trends in Endocrinology and Metabolism, 2021, 32, 224-237.	3.1	113
10	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
11	Use of Twitter in Neurology: Boon or Bane?. Journal of Medical Internet Research, 2021, 23, e25229.	2.1	7
12	Mitochondrial DNA variants in genomic data: diagnostic uplifts and predictive implications. Nature Reviews Genetics, 2021, 22, 547-548.	7.7	3
13	Iterative Reanalysis of Hypertrophic Cardiomyopathy Exome Data Reveals Causative Pathogenic Mitochondrial DNA Variants. Circulation Genomic and Precision Medicine, 2021, 14, e003388.	1.6	2
14	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
15	Introducing The International Centre for Genomic Medicine in Neuromuscular Diseases (ICGNMD). Journal of the Neurological Sciences, 2021, 429, 117765.	0.3	0
16	Mitochondrial Strokes: Diagnostic Challenges and Chameleons. Genes, 2021, 12, 1643.	1.0	7
17	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
18	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

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19	Use of whole genome sequencing to determine genetic basis of suspected mitochondrial disorders: cohort study. BMJ, The, 2021, 375, e066288.	3.0	42
20	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
21	Inflammation causes remodeling of mitochondrial cytochrome c oxidase mediated by the bifunctional gene C15orf48. Science Advances, 2021, 7, eabl5182.	4.7	29
22	Reducing Intrathecal Baclofen Related Infections: Service Evaluation and Best Practice Guidelines. Neuromodulation, 2020, 23, 991-995.	0.4	8
23	Mitochondrial DNA-related diseases associated with single large-scale deletions and point mutations. , 2020, , 353-374.		0
24	Delayed diagnoses of mitochondrial cytopathies in patients presenting with end stage kidney disease: two case reports. BMC Nephrology, 2020, 21, 361.	0.8	0
25	Novel Biallelic NSUN3 Variants Cause Early-Onset Mitochondrial Encephalomyopathy and Seizures. Journal of Molecular Neuroscience, 2020, 70, 1962-1965.	1.1	24
26	CSF lactate. Practical Neurology, 2020, 20, 320-323.	0.5	18
27	Primary mitochondrial diseases increase susceptibility to bipolar affective disorder. Journal of Neurology, Neurosurgery and Psychiatry, 2020, 91, 892-894.	0.9	17
28	Chronic pain is common in mitochondrial disease. Neuromuscular Disorders, 2020, 30, 413-419.	0.3	26
29	Multisystem mitochondrial disease caused by a rare m.10038G>A mitochondrial tRNA ^{Gly} (MT-TG) variant. Neurology: Genetics, 2020, 6, e413.	0.9	2
30	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
31	Expanding the molecular and phenotypic spectrum of truncating MT-ATP6 mutations. Neurology: Genetics, 2020, 6, e381.	0.9	21
32	Metabolic shift underlies recovery in reversible infantile respiratory chain deficiency. EMBO Journal, 2020, 39, e105364.	3.5	26
33	Utility of Whole Blood Thiamine Pyrophosphate Evaluation in TPK1-Related Diseases. Journal of Clinical Medicine, 2019, 8, 991.	1.0	13
34	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
35	Differential phenotypic expression of a novel PDHA1 mutation in a female monozygotic twin pair. Human Genetics, 2019, 138, 1313-1322.	1.8	12
36	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

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37	Pathogenic variants in MTâ€ATP6: A United Kingdomâ€“based mitochondrial disease cohort study. Annals of Neurology, 2019, 86, 310-315.	2.8	33
38	MRPS25 mutations impair mitochondrial translation and cause encephalomyopathy. Human Molecular Genetics, 2019, 28, 2711-2719.	1.4	33
39	Autosomal dominant optic atrophy and cataract â€œplusâ€•phenotype including axonal neuropathy. Neurology: Genetics, 2019, 5, e322.	0.9	12
40	Urogenital symptoms in mitochondrial disease: overlooked and undertreated. European Journal of Neurology, 2019, 26, 1111-1120.	1.7	7
41	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
42	Vestibular dysfunction: a frequent problem for adults with mitochondrial disease. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, 838-841.	0.9	11
43	Consensus-based statements for the management of mitochondrial stroke-like episodes. Wellcome Open Research, 2019, 4, 201.	0.9	66
44	MT-ND5 Mutation Exhibits Highly Variable Neurological Manifestations at Low Mutant Load. EBioMedicine, 2018, 30, 86-93.	2.7	47
45	NDUFA4 (Renamed COXFA4) Is a Cytochrome-c Oxidase Subunit. Trends in Endocrinology and Metabolism, 2018, 29, 452-454.	3.1	44
46	Clinicopathologic and molecular spectrum of RNASEH1 -related mitochondrial disease. Neurology: Genetics, 2017, 3, e149.	0.9	19
47	When the Wind Comes Back. , 2017, , 207-209.		0
48	When the Wind Does Not Come Back. , 2017, , 211-213.		0
49	A Clinical, Neuropathological and Genetic Study of Homozygous A467T POLG-Related Mitochondrial Disease. PLoS ONE, 2016, 11, e0145500.	1.1	36
50	CAV3 mutations causing exercise intolerance, myalgia and rhabdomyolysis: Expanding the phenotypic spectrum of caveolinopathies. Neuromuscular Disorders, 2016, 26, 504-510.	0.3	38
51	Mitochondrial Extrapyramidal Syndromes. JAMA Neurology, 2016, 73, 630.	4.5	3
52	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
53	Effects of ketosis in mitochondrial myopathy: potential benefits of a mitotoxic diet. EMBO Molecular Medicine, 2016, 8, 1231-1233.	3.3	4
54	Interventions for dysphagia in long-term, progressive muscle disease. The Cochrane Library, 2016, 2016, CD004303.	1.5	27

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55	Extra-ocular muscle MRI in genetically-defined mitochondrial disease. European Radiology, 2016, 26, 130-137.	2.3	24
56	Rhabdomyolysis: a genetic perspective. Orphanet Journal of Rare Diseases, 2015, 10, 51.	1.2	101
57	Glycogen storage disease type XV: A case report. Neuromuscular Disorders, 2015, 25, S221.	0.3	1
58	The urinary proteome and metabonome differ from normal in adults with mitochondrial disease. Kidney International, 2015, 87, 610-622.	2.6	41
59	Whole-genome sequencing and the clinician: a tale of two cities. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 1012-1015.	0.9	2
60	Novel C12orf65 mutations in patients with axonal neuropathy and optic atrophy. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 486-492.	0.9	35
61	Peripheral neuropathy predicts nuclear gene defect in patients with mitochondrial ophthalmoplegia. Brain, 2014, 137, 3200-3212.	3.7	45
62	Mitochondrial myopathies in adults and children. Current Opinion in Neurology, 2014, 27, 576-582.	1.8	32
63	NDUFA4 Mutations Underlie Dysfunction of a Cytochrome c Oxidase Subunit Linked to Human Neurological Disease. Cell Reports, 2013, 3, 1795-1805.	2.9	104
64	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
65	RRM2B-Related Mitochondrial Disease. , 2013, , 171-182.		3
66	COX10Mutations Resulting in Complex Multisystem Mitochondrial Disease That Remains Stable Into Adulthood. JAMA Neurology, 2013, 70, 1556-61.	4.5	27
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	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
69	Muscle MRI in Bethlem myopathy. BMJ Case Reports, 2013, 2013, bcr2013008596-bcr2013008596.	0.2	6
70	Adults with RRM2B-related mitochondrial disease have distinct clinical and molecular characteristics. Brain, 2012, 135, 3392-3403.	3.7	70
71	Genetic dysfunction of MT-ATP6 causes axonal Charcot-Marie-Tooth disease. Neurology, 2012, 79, 1145-1154.	1.5	97
72	Single deletions in mitochondrial DNA â€“ Molecular mechanisms and disease phenotypes in clinical practice. Neuromuscular Disorders, 2012, 22, 577-586.	0.3	62

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73	Toward a mtDNA locus-specific mutation database using the LOVD platform. Human Mutation, 2012, 33, 1352-1358.	1.1	8
74	A dizzy and disorientated DJ. Practical Neurology, 2011, 11, 252-255.	0.5	0
75	Kearns-Sayre syndrome caused by defective R1/p53R2 assembly. Journal of Medical Genetics, 2011, 48, 610-617.	1.5	49
76	Cardiac manifestations of mitochondrial disorders: reply. European Journal of Heart Failure, 2010, 12, 637-638.	2.9	0
77	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
78	Copper deficiency: an unusual case of myelopathy with neuropathy. Annals of Clinical Biochemistry, 2008, 45, 434-435.	0.8	7
79	Neurology â€“ the Peripheral Nervous System â€“ Muscle and Nerve. , 0, , 84-94.		0