Doug M Turnbull

List of Publications by Year
in descending order

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309 papers	25,818 citations	5876 81 h-index	8138 148 g-index
318 all docs	318 docs citations	318 times ranked	22702 citing authors

#	Article	IF	Citations
1	Reanalysis and revision of the Cambridge reference sequence for human mitochondrial DNA. Nature Genetics, 1999, 23, 147-147.	9.4	2,800
2	Mitochondrial DNA mutations in human disease. Nature Reviews Genetics, 2005, 6, 389-402.	7.7	1,530
3	Ageing and Parkinson's disease: Why is advancing age the biggest risk factor?. Ageing Research Reviews, 2014, 14, 19-30.	5.0	681
4	Mammalian mitochondrial genetics: heredity, heteroplasmy and disease. Trends in Genetics, 1997, 13, 450-455.	2.9	415
5	The Epidemiology of Leber Hereditary Optic Neuropathy in the North East of England. American Journal of Human Genetics, 2003, 72, 333-339.	2.6	404
6	Mitochondrial changes within axons in multiple sclerosis. Brain, 2009, 132, 1161-1174.	3.7	393
7	Multi-system neurological disease is common in patients with OPA1 mutations. Brain, 2010, 133, 771-786.	3.7	385
8	The epidemiology of pathogenic mitochondrial DNA mutations. Annals of Neurology, 2000, 48, 188-193.	2.8	381
9	What causes mitochondrial DNA deletions in human cells?. Nature Genetics, 2008, 40, 275-279.	9.4	334
10	The genetics and pathology of mitochondrial disease. Journal of Pathology, 2017, 241, 236-250.	2.1	329
11	Molecular pathology of MELAS and MERRF. The relationship between mutation load and clinical phenotypes. Brain, 1997, 120, 1713-1721.	3.7	319
12	Mitochondrial DNA deletions and neurodegeneration in multiple sclerosis. Annals of Neurology, 2011, 69, 481-492.	2.8	306
13	Mitochondrial enzyme activity in amyotrophic lateral sclerosis: Implications for the role of mitochondria in neuronal cell death. Annals of Neurology, 1999, 46, 787-790.	2.8	292
14	ETFDH mutations as a major cause of riboflavin-responsive multiple acyl-CoA dehydrogenation deficiency. Brain, 2007, 130, 2045-2054.	3.7	292
15	Random Intracellular Drift Explains the Clonal Expansion of Mitochondrial DNA Mutations with Age. American Journal of Human Genetics, 2001, 68, 802-806.	2.6	289
16	Role of mitochondrial DNA mutations in human aging: Implications for the central nervous system and muscle. Annals of Neurology, 1998, 43, 217-223.	2.8	280
17	Mitochondrial DNA mutations are established in human colonic stem cells, and mutated clones expand by crypt fission. Proceedings of the National Academy of Sciences of the United States of America, 2006, 103, 714-719.	3.3	269
18	Selective inhibition of mutant human mitochondrial DNA replication in vitro by peptide nucleic acids. Nature Genetics, 1997, 15, 212-215.	9.4	252

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19	Mutations causing mitochondrial disease: What is new and what challenges remain?. Science, 2015, 349, 1494-1499.	6.0	251
20	Autosomal recessive mitochondrial ataxic syndrome due to mitochondrial polymerase Î³ mutations. Neurology, 2005, 64, 1204-1208.	1.5	246
21	Mitochondrial Diseases: Hope for the Future. Cell, 2020, 181, 168-188.	13.5	243
22	An Evaluation of the Measurement of the Activities of Complexes I-IV in the Respiratory Chain of Human Skeletal Muscle Mitochondria. Biochemical Medicine and Metabolic Biology, 1994, 51, 35-42.	0.7	241
23	Mitochondrial DNA and disease. Journal of Pathology, 2012, 226, 274-286.	2.1	239
24	Clinical features, diagnosis and management of maternally inherited diabetes and deafness (MIDD) associated with the 3243A>G mitochondrial point mutation. Diabetic Medicine, 2008, 25, 383-399.	1.2	229
25	The inheritance of mitochondrial DNA heteroplasmy: random drift, selection or both?. Trends in Genetics, 2000, 16, 500-505.	2.9	227
26	Recent Advances in Mitochondrial Disease. Annual Review of Genomics and Human Genetics, 2017, 18, 257-275.	2.5	217
27	mt <scp>DNA</scp> heteroplasmy level and copy number indicate disease burden in m.3243A>G mitochondrial disease. EMBO Molecular Medicine, 2018, 10, .	3.3	199
28	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
29	A partial deficiency of cytochrome c oxidase in chronic progressive external ophthalmoplegia. Journal of the Neurological Sciences, 1983, 60, 31-53.	0.3	189
30	Endurance training and detraining in mitochondrial myopathies due to single large-scale mtDNA deletions. Brain, 2006, 129, 3391-3401.	3.7	189
31	Mitochondrial <scp>DNA</scp> Depletion in Respiratory Chainâ€“Deficient <scp>P</scp>arkinson Disease Neurons. Annals of Neurology, 2016, 79, 366-378.	2.8	189
32	Fatty acid oxidation is required for the respiration and proliferation of malignant glioma cells. Neuro-Oncology, 2017, 19, 43-54.	0.6	189
33	Cardiac involvement in mitochondrial DNA disease: clinical spectrum, diagnosis, and management. European Heart Journal, 2012, 33, 3023-3033.	1.0	182
34	Comparative Genomics and the Evolution of Human Mitochondrial DNA: Assessing the Effects of Selection. American Journal of Human Genetics, 2004, 74, 229-238.	2.6	177
35	Peptide nucleic acid delivery to human mitochondria. Gene Therapy, 1999, 6, 1919-1928.	2.3	176
36	Mitochondrial Diseaseâ€”Its Impact, Etiology, and Pathology. Current Topics in Developmental Biology, 2007, 77, 113-155.	1.0	169

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37	The mitochondrial ND6 gene is a hot spot for mutations that cause Leber's hereditary optic neuropathy. Brain, 2001, 124, 209-218.	3.7	167
38	The Spectrum of Mitochondrial Ultrastructural Defects in Mitochondrial Myopathy. Scientific Reports, 2016, 6, 30610.	1.6	165
39	Mitochondrial enzyme-deficient hippocampal neurons and choroidal cells in AD. Neurology, 2001, 57, 260-264.	1.5	164
40	Presentation and clinical investigation of mitochondrial respiratory chain disease. A study of 51 patients. Brain, 1995, 118, 339-357.	3.7	162
41	Mutations of the mitochondrial ND1 gene as a cause of MELAS. Journal of Medical Genetics, 2004, 41, 784-789.	1.5	161
42	Mitochondrial Dysfunction in Parkinsonâ€™s Diseaseâ€”Cause or Consequence?. Biology, 2019, 8, 38.	1.3	153
43	The determination of complete human mitochondrial DNA sequences in single cells: implications for the study of somatic mitochondrial DNA point mutations. Nucleic Acids Research, 2001, 29, 74e-74.	6.5	152
44	Random Genetic Drift Determines the Level of Mutant mtDNA in Human Primary Oocytes. American Journal of Human Genetics, 2001, 68, 533-536.	2.6	147
45	Mutations of ANT1 , Twinkle , and POLG1 in sporadic progressive external ophthalmoplegia (PEO). Neurology, 2003, 60, 1354-1356.	1.5	147
46	The distributions of mitochondria and sodium channels reflect the specific energy requirements and conduction properties of the human optic nerve head. British Journal of Ophthalmology, 2004, 88, 286-290.	2.1	147
47	Resistance training in patients with single, large-scale deletions of mitochondrial DNA. Brain, 2008, 131, 2832-2840.	3.7	147
48	A new mtDNA mutation showing accumulation with time and restriction to skeletal muscle. American Journal of Human Genetics, 1997, 60, 373-80.	2.6	144
49	Aggregated Î±-synuclein and complex I deficiency: exploration of their relationship in differentiated neurons. Cell Death and Disease, 2015, 6, e1820-e1820.	2.7	139
50	Reversal of a mitochondrial DNA defect in human skeletal muscle. Nature Genetics, 1997, 16, 222-224.	9.4	137
51	The spectrum of hearing loss due to mitochondrial DNA defects. Brain, 2000, 123, 82-92.	3.7	132
52	Increased mitochondrial content in remyelinated axons: implications for multiple sclerosis. Brain, 2011, 134, 1901-1913.	3.7	131
53	Mitochondrial DNA Mutations in Disease, Aging, and Neurodegeneration. Annals of the New York Academy of Sciences, 2008, 1147, 21-29.	1.8	129
54	Molecular neuropathology of MELAS: level of heteroplasmy in individual neurones and evidence of extensive vascular involvement. Neuropathology and Applied Neurobiology, 2006, 32, 359-373.	1.8	127

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55	Effects of physical activity and age on mitochondrial function. QJM - Monthly Journal of the Association of Physicians, 1996, 89, 251-258.	0.2	125
56	Cytochrome c oxidase deficient cells accumulate in the hippocampus and choroid plexus with age. Neurobiology of Aging, 2001, 22, 265-272.	1.5	125
57	Mitochondrial disease in adults: A scale to monitor progression and treatment. Neurology, 2006, 66, 1932-1934.	1.5	125
58	Abnormal RNA processing associated with a novel tRNA mutation in mitochondrial DNA. A potential disease mechanism. Journal of Biological Chemistry, 1993, 268, 19559-64.	1.6	121
59	Analysis of European mtDNAs for Recombination. American Journal of Human Genetics, 2001, 68, 145-153.	2.6	116
60	Quantitative 3D Mapping of the Human Skeletal Muscle Mitochondrial Network. Cell Reports, 2019, 26, 996-1009.e4.	2.9	116
61	Mitochondrial DNA and disease. Lancet, The, 1999, 354, S17-S21.	6.3	114
62	Formation of mitochondrialâ€derived vesicles is an active and physiologically relevant mitochondrial quality control process in the cardiac system. Journal of Physiology, 2016, 594, 5343-5362.	1.3	113
63	Mechanism of neurodegeneration of neurons with mitochondrial DNA mutations. Brain, 2010, 133, 797-807.	3.7	108
64	Cell-permeable succinate prodrugs bypass mitochondrial complex I deficiency. Nature Communications, 2016, 7, 12317.	5.8	106
65	The ageing neuromuscular system and sarcopenia: a mitochondrial perspective. Journal of Physiology, 2016, 594, 4499-4512.	1.3	105
66	A novel immunofluorescent assay to investigate oxidative phosphorylation deficiency in mitochondrial myopathy: understanding mechanisms and improving diagnosis. Scientific Reports, 2015, 5, 15037.	1.6	104
67	Disease progression in patients with single, large-scale mitochondrial DNA deletions. Brain, 2014, 137, 323-334.	3.7	103
68	Sequence variation in mitochondrial complex I genes: mutation or polymorphism?. Journal of Medical Genetics, 2005, 43, 175-179.	1.5	102
69	Topoisomerase 3Î± Is Required for Decatenation and Segregation of Human mtDNA. Molecular Cell, 2018, 69, 9-23.e6.	4.5	102
70	Phenotypic heterogeneity in m.3243A>G mitochondrial disease: The role of nuclear factors. Annals of Clinical and Translational Neurology, 2018, 5, 333-345.	1.7	102
71	Mitochondrial disease in adults: recent advances and future promise. Lancet Neurology, The, 2021, 20, 573-584.	4.9	96
72	URINE HETEROPLASMY IS THE BEST PREDICTOR OF CLINICAL OUTCOME IN THE m.3243A>G mtDNA MUTATION. Neurology, 2009, 72, 568-569.	1.5	95

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73	Mitochondrial disease: genetics and management. Journal of Neurology, 2016, 263, 179-191.	1.8	95
74	Mitochondrial Nanotunnels. Trends in Cell Biology, 2017, 27, 787-799.	3.6	95
75	Transmission of mitochondrial DNA disorders: possibilities for the future. Lancet, The, 2006, 368, 87-89.	6.3	93
76	MELAS ASSOCIATED WITH MUTATIONS IN THE POLG1 GENE. Neurology, 2007, 68, 1741-1742.	1.5	92
77	Cerebellar Ataxia in Patients With Mitochondrial DNA Disease. Journal of Neuropathology and Experimental Neurology, 2012, 71, 148-161.	0.9	91
78	Clinical features, investigation, and management of patients with defects of mitochondrial DNA. Journal of Neurology, Neurosurgery and Psychiatry, 1997, 63, 559-563.	0.9	90
79	Point Mutations of the mtDNA Control Region in Normal and Neurodegenerative Human Brains. American Journal of Human Genetics, 2001, 68, 529-532.	2.6	90
80	Linking the Metabolic State and Mitochondrial DNA in Chronic Disease, Health, and Aging. Diabetes, 2013, 62, 672-678.	0.3	90
81	Mitochondrial DNA mutations in the pathogenesis of human disease. Trends in Molecular Medicine, 2000, 6, 425-432.	2.6	89
82	The rise and rise of mitochondrial DNA mutations. Open Biology, 2020, 10, 200061.	1.5	89
83	The phenotypic spectrum of neutral lipid storage myopathy due to mutations in the PNPLA2 gene. Journal of Neurology, 2011, 258, 1987-1997.	1.8	87
84	Mitochondrial Donation â€" How Many Women Could Benefit?. New England Journal of Medicine, 2015, 372, 885-887.	13.9	87
85	The clinical, histochemical, and molecular spectrum of PEO1 (Twinkle)-linked adPEO. Neurology, 2010, 74, 1619-1626.	1.5	84
86	Diagnosis and Treatment of Mitochondrial Myopathies. Neurotherapeutics, 2018, 15, 943-953.	2.1	84
87	Mitochondrial DNA diseaseâ€"molecular insights and potential routes to a cure. Experimental Cell Research, 2014, 325, 38-43.	1.2	81
88	Variation in the Calpain-10 Gene Affects Blood Glucose Levels in the British Population. Diabetes, 2002, 51, 247-250.	0.3	79
89	Mitochondrial DNA deletions cause the biochemical defect observed in Alzheimer's disease. Neurobiology of Aging, 2012, 33, 2210-2214.	1.5	79
90	Mitochondrial dysfunction plays a key role in progressive axonal loss in Multiple Sclerosis. Medical Hypotheses, 2005, 64, 669-677.	0.8	78

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91	Batteries not included: diagnosis and management of mitochondrial disease. Journal of Internal Medicine, 2009, 265, 210-228.	2.7	78
92	Homoplasmy, heteroplasmy, and mitochondrial dystonia. Neurology, 2007, 69, 911-916.	1.5	76
93	The ageing mitochondrial genome. Nucleic Acids Research, 2007, 35, 7399-7405.	6.5	76
94	The Impact of Pathogenic Mitochondrial DNA Mutations on Substantia Nigra Neurons. Journal of Neuroscience, 2013, 33, 10790-10801.	1.7	75
95	Age-related decline in mitochondrial DNA copy number in isolated human pancreatic islets. Diabetologia, 2008, 51, 1440-1443.	2.9	74
96	Mitochondrial and inflammatory changes in sporadic inclusion body myositis. Neuropathology and Applied Neurobiology, 2015, 41, 288-303.	1.8	73
97	Dopaminergic midbrain neurons are the prime target for mitochondrial DNA deletions. Journal of Neurology, 2008, 255, 1231-1235.	1.8	72
98	Mitochondrial DNA disease: new options for prevention. Human Molecular Genetics, 2011, 20, R168-R174.	1.4	72
99	Somatic mitochondrial DNA mutations in adult-onset leukaemia. Leukemia, 2003, 17, 2487-2491.	3.3	70
100	Clinical, Genetic, and Radiological Features of Extrapyramidal Movement Disorders in Mitochondrial Disease. JAMA Neurology, 2016, 73, 668.	4.5	69
101	Age-associated mitochondrial DNA mutations cause metabolic remodeling that contributes to accelerated intestinal tumorigenesis. Nature Cancer, 2020, 1, 976-989.	5.7	69
102	Heteroplasmic ratio of the A3243G mitochondrial DNA mutation in single pancreatic beta cells. Diabetologia, 2003, 46, 296-299.	2.9	67
103	Consensus-based statements for the management of mitochondrial stroke-like episodes. Wellcome Open Research, 2019, 4, 201.	0.9	66
104	Prevalence and progression of diabetes in mitochondrial disease. Diabetologia, 2007, 50, 2085-2089.	2.9	64
105	ANO5Gene Analysis in a Large Cohort of Patients with Anoctaminopathy: Confirmation of Male Prevalence and High Occurrence of the Common Exon 5 Gene Mutation. Human Mutation, 2013, 34, 1111-1118.	1.1	64
106	Multipotent Basal Stem Cells, Maintained in Localized Proximal Niches, Support Directed Long-Ranging Epithelial Flows in Human Prostates. Cell Reports, 2017, 20, 1609-1622.	2.9	64
107	A Novel Mitochondrial DNA Point Mutation in the tRNAIleGene: Studies in a Patient Presenting with Chronic Progressive External Ophthalmoplegia and Multiple Sclerosis. Biochemical and Biophysical Research Communications, 1998, 243, 47-51.	1.0	62
108	Ablation of Cellular Prion Protein Expression Affects Mitochondrial Numbers and Morphology. Biochemical and Biophysical Research Communications, 2002, 291, 372-377.	1.0	62

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109	Transmitochondrial embryonic stem cells containing pathogenic mtDNA mutations are compromised in neuronal differentiation. Cell Proliferation, 2009, 42, 413-424.	2.4	62
110	Comparison of Mitochondrial Mutation Spectra in Ageing Human Colonic Epithelium and Disease: Absence of Evidence for Purifying Selection in Somatic Mitochondrial DNA Point Mutations. PLoS Genetics, 2012, 8, e1003082.	1.5	61
111	Sensory neuronopathy in patients harbouring recessive polymerase Î³ mutations. Brain, 2012, 135, 62-71.	3.7	60
112	RRM2B mutations are frequent in familial PEO with multiple mtDNA deletions. Neurology, 2011, 76, 2032-2034.	1.5	59
113	International Workshop:. Neuromuscular Disorders, 2017, 27, 1126-1137.	0.3	58
114	Mitochondrial dysfunction impairs osteogenesis, increases osteoclast activity, and accelerates age related bone loss. Scientific Reports, 2020, 10, 11643.	1.6	58
115	Mitochondrial DNA deletion in "identical" twin brothers. Journal of Medical Genetics, 2004, 41, 19e-19.	1.5	57
116	A national perspective on prenatal testing for mitochondrial disease. European Journal of Human Genetics, 2014, 22, 1255-1259.	1.4	57
117	Maternally inherited diabetes and deafness: prevalence in a hospital diabetic population. , 1997, 14, 457-460.		56
118	Ageâ€associated mitochondrial DNA mutations lead to small but significant changes in cell proliferation and apoptosis in human colonic crypts. Aging Cell, 2010, 9, 96-99.	3.0	56
119	Cytochromec oxidase activity in single muscle fibers: Assay techniques and diagnostic applications. Annals of Neurology, 1993, 33, 28-35.	2.8	55
120	Mitochondrial diabetes: investigation and identification of a novel mutation. Diabetes, 1998, 47, 1800-1802.	0.3	55
121	Progress in mitochondrial replacement therapies. Nature Reviews Molecular Cell Biology, 2018, 19, 71-72.	16.1	53
122	Accurate Measurement of Mitochondrial DNA Deletion Level and Copy Number Differences in Human Skeletal Muscle. PLoS ONE, 2014, 9, e114462.	1.1	53
123	Immunocytochemical studies of cytochrome oxidase subunits in skeletal muscle of patients with partial cytochrome oxidase deficiencies. Journal of the Neurological Sciences, 1988, 87, 75-90.	0.3	52
124	Defects in multiple complexes of the respiratory chain are present in ageing human colonic crypts. Experimental Gerontology, 2010, 45, 573-579.	1.2	52
125	Impaired mitochondrial function abolishes gamma oscillations in the hippocampus through an effect on fast-spiking interneurons. Brain, 2011, 134, e180-e180.	3.7	52
126	Therapeutic Manipulation of mtDNA Heteroplasmy: A Shifting Perspective. Trends in Molecular Medicine, 2020, 26, 698-709.	3.5	52

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127	A scale to monitor progression and treatment of mitochondrial disease in children. Neuromuscular Disorders, 2006, 16, 814-820.	0.3	51
128	Pathogenic mitochondrial tRNA mutations - Which mutations are inherited and why?. Human Mutation, 2009, 30, E984-E992.	1.1	49
129	Research into Policy: A Brief History of Mitochondrial Donation. Stem Cells, 2016, 34, 265-267.	1.4	49
130	Mitochondrial DNA haplogroup distribution within Leber hereditary optic neuropathy pedigrees. Journal of Medical Genetics, 2004, 41, e41-e41.	1.5	48
131	Ultrasensitive deletion detection links mitochondrial DNA replication, disease, and aging. Genome Biology, 2020, 21, 248.	3.8	48
132	MT-ND5 Mutation Exhibits Highly Variable Neurological Manifestations at Low Mutant Load. EBioMedicine, 2018, 30, 86-93.	2.7	47
133	Subcellular origin of mitochondrial DNA deletions in human skeletal muscle. Annals of Neurology, 2018, 84, 289-301.	2.8	47
134	A novel mitochondrial tRNA phenylalanine mutation presenting with acute rhabdomyolysis. Annals of Neurology, 1997, 41, 408-410.	2.8	46
135	What is influencing the phenotype of the common homozygous polymerase-Î³ mutation p.Ala467Thr?. Brain, 2012, 135, 3614-3626.	3.7	46
136	Mitochondrial DNA mutations and ageing. Biochimica Et Biophysica Acta - General Subjects, 2009, 1790, 1015-1020.	1.1	45
137	Microangiopathy in the cerebellum of patients with mitochondrial DNA disease. Brain, 2012, 135, 1736-1750.	3.7	44
138	The role of astrocytes in seizure generation: insights from a novel in vitro seizure model based on mitochondrial dysfunction. Brain, 2019, 142, 391-411.	3.7	44
139	Defining the Importance of Mitochondrial Gene Defects in Maternally Inherited Diabetes by Sequencing the Entire Mitochondrial Genome. Diabetes, 2002, 51, 2317-2320.	0.3	43
140	Alphaâ€synuclein pathology and Parkinsonism associated with POLG1 mutations and multiple mitochondrial DNA deletions. Neuropathology and Applied Neurobiology, 2009, 35, 120-124.	1.8	43
141	Mitochondrial DNA mutations affect calcium handling in differentiated neurons. Brain, 2010, 133, 787-796.	3.7	43
142	Relationship Between Mitochondria and Î±-Synuclein. Archives of Neurology, 2012, 69, 385.	4.9	43
143	Development of passive CLARITY and immunofluorescent labelling of multiple proteins in human cerebellum: understanding mechanisms of neurodegeneration in mitochondrial disease. Scientific Reports, 2016, 6, 26013.	1.6	43
144	Extensive respiratory chain defects in inhibitory interneurones in patients with mitochondrial disease. Neuropathology and Applied Neurobiology, 2016, 42, 180-193.	1.8	43

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145	Review: Central nervous system involvement in mitochondrial disease. Neuropathology and Applied Neurobiology, 2017, 43, 102-118.	1.8	42
146	Pathological mechanisms underlying single largeâ€scale mitochondrial <scp>DNA</scp> deletions. Annals of Neurology, 2018, 83, 115-130.	2.8	42
147	Mitochondrial function in muscle from elderly athletes. Annals of Neurology, 1997, 41, 114-116.	2.8	41
148	Mitochondrial DNA Mutations in Aging. Progress in Molecular Biology and Translational Science, 2014, 127, 29-62.	0.9	41
149	The urinary proteome and metabonome differ from normal in adults with mitochondrial disease. Kidney International, 2015, 87, 610-622.	2.6	41
150	Neuropathological and Histochemical Changes in a Multiple Mitochondrial DNA Deletion Disorder. Journal of Neuropathology and Experimental Neurology, 2000, 59, 621-627.	0.9	40
151	Pseudoâ€obstruction, stroke, and mitochondrial dysfunction: A lethal combination. Annals of Neurology, 2016, 80, 686-692.	2.8	40
152	Increase in mitochondrial density within axons and supporting cells in response to demyelination in the Plp1 mouse model. Journal of Neuroscience Research, 2009, 87, 452-459.	1.3	39
153	Mitochondrial disease in pregnancy: a systematic review. Obstetric Medicine, 2011, 4, 90-94.	0.5	39
154	Cytochrome c oxidase-intermediate fibres: Importance in understanding the pathogenesis and treatment of mitochondrial myopathy. Neuromuscular Disorders, 2012, 22, 690-698.	0.3	39
155	Analysis of fatty acid oxidation intermediates in cultured fibroblasts to detect mitochondrial oxidation disorders. Clinical Chemistry, 1994, 40, 2267-2275.	1.5	38
156	Human Insulin Receptor Substrate-1: Variant Sequences in Familial Non-insulin-dependent Diabetes Mellitus. Diabetic Medicine, 1996, 13, 133-138.	1.2	38
157	Novel POLG1 mutations associated with neuromuscular and liver phenotypes in adults and children. Journal of Medical Genetics, 2008, 46, 209-214.	1.5	38
158	Mitochondrial changes within axons in multiple sclerosis. Current Opinion in Neurology, 2012, 25, 221-230.	1.8	38
159	Clonally expanded mitochondrial DNA deletions within the choroid plexus in multiple sclerosis. Acta Neuropathologica, 2012, 124, 209-220.	3.9	38
160	The mitochondrial brain: From mitochondrial genome to neurodegeneration. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2010, 1802, 111-121.	1.8	37
161	Complex mitochondrial DNA rearrangements in individual cells from patients with sporadic inclusion body myositis. Nucleic Acids Research, 2016, 44, 5313-5329.	6.5	37
162	Quantification of mitochondrial DNA mutation load. Aging Cell, 2009, 8, 566-572.	3.0	36

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163	Causes of Death in Adults with Mitochondrial Disease. JIMD Reports, 2015, 26, 103-113.	0.7	36
164	Dissecting the mechanisms underlying the accumulation of mitochondrial DNA deletions in human skeletal muscle. Human Molecular Genetics, 2014, 23, 4612-4620.	1.4	34
165	So doctor, what exactly is wrong with my muscles? Glutaric aciduria type II presenting in a teenager. Neuromuscular Disorders, 2006, 16, 269-273.	0.3	33
166	Mitochondrial DNA deletions in muscle satellite cells: implications for therapies. Human Molecular Genetics, 2013, 22, 4739-4747.	1.4	33
167	Respiratory chain deficiency in aged spinal motor neurons. Neurobiology of Aging, 2014, 35, 2230-2238.	1.5	33
168	Similar patterns of clonally expanded somatic mtDNA mutations in the colon of heterozygous mtDNA mutator mice and ageing humans. Mechanisms of Ageing and Development, 2014, 139, 22-30.	2.2	33
169	Pathogenic variants in MTâ€ATP6: A United Kingdomâ€“based mitochondrial disease cohort study. Annals of Neurology, 2019, 86, 310-315.	2.8	33
170	Investigation of mitochondrial biogenesis defects in single substantia nigra neurons using post-mortem human tissues. Neurobiology of Disease, 2020, 134, 104631.	2.1	33
171	Mitochondrial dysfunction in myofibrillar myopathy. Neuromuscular Disorders, 2016, 26, 691-701.	0.3	32
172	Assessment of mitochondrial respiratory chain enzymes in cells and tissues. Methods in Cell Biology, 2020, 155, 121-156.	0.5	32
173	Mitochondrial enteropathy: the primary pathology may not be within the gastrointestinal tract. Gut, 2001, 48, 121-124.	6.1	31
174	Investigation of mitochondrial function in hereditary spastic paraparesis. NeuroReport, 2003, 14, 485-488.	0.6	30
175	OPA1 IN MULTIPLE MITOCHONDRIAL DNA DELETION DISORDERS. Neurology, 2008, 71, 1829-1831.	1.5	30
176	Detection of cytochrome c oxidase activity and mitochondrial proteins in single cells. Journal of Neuroscience Methods, 2009, 184, 310-319.	1.3	30
177	Triplex real-time PCRâ€“an improved method to detect a wide spectrum of mitochondrial DNA deletions in single cells. Scientific Reports, 2015, 5, 9906.	1.6	30
178	Preferential amplification of a human mitochondrial DNA deletion in vitro and in vivo. Scientific Reports, 2018, 8, 1799.	1.6	30
179	Mitochondrial donation: from test tube to clinic. Lancet, The, 2018, 392, 1191-1192.	6.3	30
180	POLG1, C10ORF2, and ANT1 mutations are uncommon in sporadic progressive external ophthalmoplegia with multiple mitochondrial DNA deletions. Neurology, 2006, 66, 1439-1441.	1.5	28

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181	Mitochondrial Mutations. Neuroscientist, 2011, 17, 645-658.	2.6	28
182	Quantitative quadruple-label immunofluorescence of mitochondrial and cytoplasmic proteins in single neurons from human midbrain tissue. Journal of Neuroscience Methods, 2014, 232, 143-149.	1.3	28
183	Dysferlin mutations and mitochondrial dysfunction. Neuromuscular Disorders, 2016, 26, 782-788.	0.3	28
184	The low abundance of clonally expanded mitochondrial DNA point mutations in aged substantia nigra neurons. Aging Cell, 2009, 8, 496-498.	3.0	26
185	Catastrophic presentation of mitochondrial disease due to a mutation in the tRNA ^{His} gene. Neurology, 2004, 62, 1420-1423.	1.5	25
186	Loss of Myelin-Associated Glycoprotein in Kearns-Sayre Syndrome. Archives of Neurology, 2012, 69, 490.	4.9	25
187	Scientific and Ethical Issues in Mitochondrial Donation. New Bioethics, 2018, 24, 57-73.	0.5	25
188	Mitochondrial Donation â€" Which Women Could Benefit?. New England Journal of Medicine, 2019, 380, 1971-1972.	13.9	25
189	Forecasting stroke-like episodes and outcomes in mitochondrial disease. Brain, 2022, 145, 542-554.	3.7	25
190	Mitochondrial myopathy with skeletal muscle cytochrome oxidase deficiency. Journal of Inherited Metabolic Disease, 1982, 5, 27-28.	1.7	24
191	Frequency of rare mitochondrial DNA mutations in patients with suspected Leber's hereditary optic neuropathy. Journal of Medical Genetics, 2003, 40, 85e-85.	1.5	24
192	Pathogenic mtDNA mutations causing mitochondrial myopathy: The need for muscle biopsy. Neurology: Genetics, 2016, 2, e82.	0.9	24
193	Mitochondrial Mutations: Newly Discovered Players in Neuronal Degeneration. Neuroscientist, 2011, 17, 645-658.	2.6	24
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