Doug M Turnbull

List of Publications by Citations

302 papers	20,480 citations	73 h-index	135 g-index
318 ext. papers	23,210 ext. citations	8.9 avg, IF	6.81 L-index

#	Paper	IF	Citations
302	Reanalysis and revision of the Cambridge reference sequence for human mitochondrial DNA. *Nature Genetics*, **1999**, 23, 147	36.3	2349
301	Mitochondrial DNA mutations in human disease. *Nature Reviews Genetics*, **2005**, 6, 389-402	30.1	1243
300	Ageing and Parkinson's disease: why is advancing age the biggest risk factor?. *Ageing Research Reviews*, **2014**, 14, 19-30	12	487
299	Mammalian mitochondrial genetics: heredity, heteroplasmy and disease. *Trends in Genetics*, **1997**, 13, 450-5	8.5	356
298	The epidemiology of pathogenic mitochondrial DNA mutations. *Annals of Neurology*, **2000**, 48, 188-193	9.4	352
297	Mitochondrial changes within axons in multiple sclerosis. *Brain*, **2009**, 132, 1161-74	11.2	338
296	The epidemiology of Leber hereditary optic neuropathy in the North East of England. *American Journal of Human Genetics*, **2003**, 72, 333-9	11	329
295	Multi-system neurological disease is common in patients with OPA1 mutations. *Brain*, **2010**, 133, 771-86	11.2	314
294	What causes mitochondrial DNA deletions in human cells?. *Nature Genetics*, **2008**, 40, 275-9	36.3	302
293	Mitochondrial enzyme activity in amyotrophic lateral sclerosis: implications for the role of mitochondria in neuronal cell death. *Annals of Neurology*, **1999**, 46, 787-90	9.4	264
292	Role of mitochondrial DNA mutations in human aging: implications for the central nervous system and muscle. *Annals of Neurology*, **1998**, 43, 217-23	9.4	256
291	Mitochondrial DNA deletions and neurodegeneration in multiple sclerosis. *Annals of Neurology*, **2011**, 69, 481-92	9.4	255
290	Molecular pathology of MELAS and MERRF. The relationship between mutation load and clinical phenotypes. *Brain*, **1997**, 120 (Pt 10), 1713-21	11.2	251
289	Random intracellular drift explains the clonal expansion of mitochondrial DNA mutations with age. *American Journal of Human Genetics*, **2001**, 68, 802-6	11	251
288	ETFDH mutations as a major cause of riboflavin-responsive multiple acyl-CoA dehydrogenation deficiency. *Brain*, **2007**, 130, 2045-54	11.2	238
287	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, 714-9	11.5	234
286	Autosomal recessive mitochondrial ataxic syndrome due to mitochondrial polymerase gamma mutations. *Neurology*, **2005**, 64, 1204-8	6.5	224

285	Selective inhibition of mutant human mitochondrial DNA replication in vitro by peptide nucleic acids. *Nature Genetics*, **1997**, 15, 212-5	36.3	223
284	The genetics and pathology of mitochondrial disease. *Journal of Pathology*, **2017**, 241, 236-250	9.4	222
283	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		210
282	Mutations causing mitochondrial disease: What is new and what challenges remain?. *Science*, **2015**, 349, 1494-9	33.3	204
281	The inheritance of mitochondrial DNA heteroplasmy: random drift, selection or both?. *Trends in Genetics*, **2000**, 16, 500-5	8.5	198
280	Mitochondrial DNA and disease. *Journal of Pathology*, **2012**, 226, 274-86	9.4	195
279	A partial deficiency of cytochrome c oxidase in chronic progressive external ophthalmoplegia. *Journal of the Neurological Sciences*, **1983**, 60, 31-53	3.2	179
278	Clinical features, diagnosis and management of maternally inherited diabetes and deafness (MIDD) associated with the 3243A>G mitochondrial point mutation. *Diabetic Medicine*, **2008**, 25, 383-99	3.5	178
277	Endurance training and detraining in mitochondrial myopathies due to single large-scale mtDNA deletions. *Brain*, **2006**, 129, 3391-401	11.2	156
276	Peptide nucleic acid delivery to human mitochondria. *Gene Therapy*, **1999**, 6, 1919-28	4	156
275	Comparative genomics and the evolution of human mitochondrial DNA: assessing the effects of selection. *American Journal of Human Genetics*, **2004**, 74, 229-38	11	155
274	The mitochondrial ND6 gene is a hot spot for mutations that cause Leber's hereditary optic neuropathy. *Brain*, **2001**, 124, 209-18	11.2	153
273	Recent Advances in Mitochondrial Disease. *Annual Review of Genomics and Human Genetics*, **2017**, 18, 257-275	9.7	149
272	Presentation and clinical investigation of mitochondrial respiratory chain disease. A study of 51 patients. *Brain*, **1995**, 118 (Pt 2), 339-57	11.2	146
271	Mitochondrial disease--its impact, etiology, and pathology. *Current Topics in Developmental Biology*, **2007**, 77, 113-55	5.3	145
270	Mitochondrial enzyme-deficient hippocampal neurons and choroidal cells in AD. *Neurology*, **2001**, 57, 260-4	6.5	145
269	Mutations of ANT1, Twinkle, and POLG1 in sporadic progressive external ophthalmoplegia (PEO). *Neurology*, **2003**, 60, 1354-6	6.5	141
268	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, 936-8	5.5	138

267	Cardiac involvement in mitochondrial DNA disease: clinical spectrum, diagnosis, and management. *European Heart Journal*, **2012**, 33, 3023-33	9.5	138
266	A new mtDNA mutation showing accumulation with time and restriction to skeletal muscle. *American Journal of Human Genetics*, **1997**, 60, 373-80	11	136
265	Mutations of the mitochondrial ND1 gene as a cause of MELAS. *Journal of Medical Genetics*, **2004**, 41, 784-9	5.8	135
264	Mitochondrial DNA Depletion in Respiratory Chain-Deficient Parkinson Disease Neurons. *Annals of Neurology*, **2016**, 79, 366-78	9.4	131
263	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, E74-4	20.1	130
262	Reversal of a mitochondrial DNA defect in human skeletal muscle. *Nature Genetics*, **1997**, 16, 222-4	36.3	129
261	Resistance training in patients with single, large-scale deletions of mitochondrial DNA. *Brain*, **2008**, 131, 2832-40	11.2	124
260	Fatty acid oxidation is required for the respiration and proliferation of malignant glioma cells. *Neuro-Oncology*, **2017**, 19, 43-54	1	123
259	Random genetic drift determines the level of mutant mtDNA in human primary oocytes. *American Journal of Human Genetics*, **2001**, 68, 533-6	11	121
258	Cytochrome c oxidase deficient cells accumulate in the hippocampus and choroid plexus with age. *Neurobiology of Aging*, **2001**, 22, 265-72	5.6	120
257	Abnormal RNA processing associated with a novel tRNA mutation in mitochondrial DNA. A potential disease mechanism. *Journal of Biological Chemistry*, **1993**, 268, 19559-64	5.4	117
256	mtDNA heteroplasmy level and copy number indicate disease burden in m.3243A>G mitochondrial disease. *EMBO Molecular Medicine*, **2018**, 10,	12	115
255	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, 286-90	5.5	115
254	Mitochondrial DNA mutations in disease, aging, and neurodegeneration. *Annals of the New York Academy of Sciences*, **2008**, 1147, 21-9	6.5	114
253	Molecular neuropathology of MELAS: level of heteroplasmy in individual neurones and evidence of extensive vascular involvement. *Neuropathology and Applied Neurobiology*, **2006**, 32, 359-73	5.2	114
252	Increased mitochondrial content in remyelinated axons: implications for multiple sclerosis. *Brain*, **2011**, 134, 1901-13	11.2	110
251	The spectrum of hearing loss due to mitochondrial DNA defects. *Brain*, **2000**, 123 (Pt 1), 82-92	11.2	108
250	Effects of physical activity and age on mitochondrial function. *QJM - Monthly Journal of the Association of Physicians*, **1996**, 89, 251-8	2.7	107

249	Mitochondrial disease in adults: a scale to monitor progression and treatment. *Neurology*, **2006**, 66, 1932-4	6.5	106
248	Aggregated Esynuclein and complex I deficiency: exploration of their relationship in differentiated neurons. *Cell Death and Disease*, **2015**, 6, e1820	9.8	104
247	Analysis of European mtDNAs for recombination. *American Journal of Human Genetics*, **2001**, 68, 145-153	11	102
246	The Spectrum of Mitochondrial Ultrastructural Defects in Mitochondrial Myopathy. *Scientific Reports*, **2016**, 6, 30610	4.9	99
245	Mitochondrial Diseases: Hope for the Future. *Cell*, **2020**, 181, 168-188	56.2	97
244	Mitochondrial DNA and disease. *Lancet, The*, **1999**, 354 Suppl 1, SI17-21	40	97
243	Mechanism of neurodegeneration of neurons with mitochondrial DNA mutations. *Brain*, **2010**, 133, 797-807	11.2	91
242	Sequence variation in mitochondrial complex I genes: mutation or polymorphism?. *Journal of Medical Genetics*, **2006**, 43, 175-9	5.8	90
241	Transmission of mitochondrial DNA disorders: possibilities for the future. *Lancet, The*, **2006**, 368, 87-9	40	88
240	Mitochondrial Dysfunction in Parkinson's Disease-Cause or Consequence?. *Biology*, **2019**, 8,	4.9	85
239	Point mutations of the mtDNA control region in normal and neurodegenerative human brains. *American Journal of Human Genetics*, **2001**, 68, 529-32	11	82
238	Disease progression in patients with single, large-scale mitochondrial DNA deletions. *Brain*, **2014**, 137, 323-34	11.2	81
237	Urine heteroplasmy is the best predictor of clinical outcome in the m.3243A>G mtDNA mutation. *Neurology*, **2009**, 72, 568-9	6.5	81
236	MELAS associated with mutations in the POLG1 gene. *Neurology*, **2007**, 68, 1741-2	6.5	79
235	A novel immunofluorescent assay to investigate oxidative phosphorylation deficiency in mitochondrial myopathy: understanding mechanisms and improving diagnosis. *Scientific Reports*, **2015**, 5, 15037	4.9	78
234	The phenotypic spectrum of neutral lipid storage myopathy due to mutations in the PNPLA2 gene. *Journal of Neurology*, **2011**, 258, 1987-97	5.5	77
233	Formation of mitochondrial-derived vesicles is an active and physiologically relevant mitochondrial quality control process in the cardiac system. *Journal of Physiology*, **2016**, 594, 5343-62	3.9	76
232	Cell-permeable succinate prodrugs bypass mitochondrial complex I deficiency. *Nature Communications*, **2016**, 7, 12317	17.4	74

231	The clinical, histochemical, and molecular spectrum of PEO1 (Twinkle)-linked adPEO. *Neurology*, **2010**, 74, 1619-26	6.5	73
230	Cerebellar ataxia in patients with mitochondrial DNA disease: a molecular clinicopathological study. *Journal of Neuropathology and Experimental Neurology*, **2012**, 71, 148-61	3.1	71
229	Mitochondrial dysfunction plays a key role in progressive axonal loss in Multiple Sclerosis. *Medical Hypotheses*, **2005**, 64, 669-77	3.8	71
228	Variation in the calpain-10 gene affects blood glucose levels in the British population. *Diabetes*, **2002**, 51, 247-50	0.9	71
227	Mitochondrial donation--how many women could benefit?. *New England Journal of Medicine*, **2015**, 372, 885-887	59.2	69
226	Linking the metabolic state and mitochondrial DNA in chronic disease, health, and aging. *Diabetes*, **2013**, 62, 672-8	0.9	69
225	Dopaminergic midbrain neurons are the prime target for mitochondrial DNA deletions. *Journal of Neurology*, **2008**, 255, 1231-5	5.5	68
224	Homoplasmy, heteroplasmy, and mitochondrial dystonia. *Neurology*, **2007**, 69, 911-6	6.5	68
223	The ageing mitochondrial genome. *Nucleic Acids Research*, **2007**, 35, 7399-405	20.1	68
222	Mitochondrial DNA disease-molecular insights and potential routes to a cure. *Experimental Cell Research*, **2014**, 325, 38-43	4.2	67
221	Mitochondrial DNA mutations in the pathogenesis of human disease. *Trends in Molecular Medicine*, **2000**, 6, 425-32		67
220	The ageing neuromuscular system and sarcopenia: a mitochondrial perspective. *Journal of Physiology*, **2016**, 594, 4499-512	3.9	66
219	Mitochondrial DNA disease: new options for prevention. *Human Molecular Genetics*, **2011**, 20, R168-74	5.6	63
218	Somatic mitochondrial DNA mutations in adult-onset leukaemia. *Leukemia*, **2003**, 17, 2487-91	10.7	63
217	Batteries not included: diagnosis and management of mitochondrial disease. *Journal of Internal Medicine*, **2009**, 265, 210-28	10.8	62
216	Topoisomerase 3β Is Required for Decatenation and Segregation of Human mtDNA. *Molecular Cell*, **2018**, 69, 9-23.e6	17.6	61
215	The impact of pathogenic mitochondrial DNA mutations on substantia nigra neurons. *Journal of Neuroscience*, **2013**, 33, 10790-801	6.6	61
214	Mitochondrial DNA deletions cause the biochemical defect observed in Alzheimer's disease. *Neurobiology of Aging*, **2012**, 33, 2210-4	5.6	61

213	Age-related decline in mitochondrial DNA copy number in isolated human pancreatic islets. *Diabetologia*, **2008**, 51, 1440-3	10.3	61
212	A novel mitochondrial DNA point mutation in the tRNA(Ile) gene: studies in a patient presenting with chronic progressive external ophthalmoplegia and multiple sclerosis. *Biochemical and Biophysical Research Communications*, **1998**, 243, 47-51	3.4	60
211	Transmitochondrial embryonic stem cells containing pathogenic mtDNA mutations are compromised in neuronal differentiation. *Cell Proliferation*, **2009**, 42, 413-24	7.9	58
210	Heteroplasmic ratio of the A3243G mitochondrial DNA mutation in single pancreatic beta cells. *Diabetologia*, **2003**, 46, 296-9	10.3	57
209	Mitochondrial Nanotunnels. *Trends in Cell Biology*, **2017**, 27, 787-799	18.3	56
208	Phenotypic heterogeneity in m.3243A>G mitochondrial disease: The role of nuclear factors. *Annals of Clinical and Translational Neurology*, **2018**, 5, 333-345	5.3	54
207	RRM2B mutations are frequent in familial PEO with multiple mtDNA deletions. *Neurology*, **2011**, 76, 2037-4	8.3	54
206	Sensory neuronopathy in patients harbouring recessive polymerase mutations. *Brain*, **2012**, 135, 62-71	11.2	53
205	Cytochrome c oxidase activity in single muscle fibers: assay techniques and diagnostic applications. *Annals of Neurology*, **1993**, 33, 28-35	9.4	53
204	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	6	52
203	ANO5 gene 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, 1111-8	4.7	51
202	Quantitative 3D Mapping of the Human Skeletal Muscle Mitochondrial Network. *Cell Reports*, **2019**, 26, 996-1009.e4	10.6	50
201	Multipotent Basal Stem Cells, Maintained in Localized Proximal Niches, Support Directed Long-Ranging Epithelial Flows in Human Prostates. *Cell Reports*, **2017**, 20, 1609-1622	10.6	50
200	Mitochondrial and inflammatory changes in sporadic inclusion body myositis. *Neuropathology and Applied Neurobiology*, **2015**, 41, 288-303	5.2	49
199	Mitochondrial DNA deletion in "identical" twin brothers. *Journal of Medical Genetics*, **2004**, 41, e19	5.8	49
198	Ablation of cellular prion protein expression affects mitochondrial numbers and morphology. *Biochemical and Biophysical Research Communications*, **2002**, 291, 372-7	3.4	49
197	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	3.2	49
196	Mitochondrial disease: genetics and management. *Journal of Neurology*, **2016**, 263, 179-91	5.5	48

195	A national perspective on prenatal testing for mitochondrial disease. *European Journal of Human Genetics*, **2014**, 22, 1255-9	5.3	48
194	Age-associated mitochondrial DNA mutations lead to small but significant changes in cell proliferation and apoptosis in human colonic crypts. *Aging Cell*, **2010**, 9, 96-9	9.9	47
193	Prevalence and progression of diabetes in mitochondrial disease. *Diabetologia*, **2007**, 50, 2085-9	10.3	47
192	Mitochondrial diabetes: investigation and identification of a novel mutation. *Diabetes*, **1998**, 47, 1800-2	0.9	47
191	A scale to monitor progression and treatment of mitochondrial disease in children. *Neuromuscular Disorders*, **2006**, 16, 814-20	2.9	46
190	Clinical, Genetic, and Radiological Features of Extrapyramidal Movement Disorders in Mitochondrial Disease. *JAMA Neurology*, **2016**, 73, 668-74	17.2	46
189	Diagnosis and Treatment of Mitochondrial Myopathies. *Neurotherapeutics*, **2018**, 15, 943-953	6.4	46
188	A novel mitochondrial tRNA phenylalanine mutation presenting with acute rhabdomyolysis. *Annals of Neurology*, **1997**, 41, 408-10	9.4	44
187	Mitochondrial DNA mutations and ageing. *Biochimica Et Biophysica Acta - General Subjects*, **2009**, 1790, 1015-20	4	43
186	Defects in multiple complexes of the respiratory chain are present in ageing human colonic crypts. *Experimental Gerontology*, **2010**, 45, 573-9	4.5	43
185	Progress in mitochondrial replacement therapies. *Nature Reviews Molecular Cell Biology*, **2018**, 19, 71-72	48.7	42
184	International Workshop:: Outcome measures and clinical trial readiness in primary mitochondrial myopathies in children and adults. Consensus recommendations. 16-18 November 2016, Rome, Italy. *Neuromuscular Disorders*, **2017**, 27, 1126-1137	2.9	42
183	Mitochondrial DNA mutations affect calcium handling in differentiated neurons. *Brain*, **2010**, 133, 787-96	11.2	42
182	Pathogenic mitochondrial tRNA mutations--which mutations are inherited and why?. *Human Mutation*, **2009**, 30, E984-92	4.7	42
181	Maternally inherited diabetes and deafness: prevalence in a hospital diabetic population. *Diabetic Medicine*, **1997**, 14, 457-60	3.5	41
180	Accurate measurement of mitochondrial DNA deletion level and copy number differences in human skeletal muscle. *PLoS ONE*, **2014**, 9, e114462	3.7	40
179	Alpha-synuclein pathology and Parkinsonism associated with POLG1 mutations and multiple mitochondrial DNA deletions. *Neuropathology and Applied Neurobiology*, **2009**, 35, 120-4	5.2	39
178	What is influencing the phenotype of the common homozygous polymerase-□mutation p.Ala467Thr?. *Brain*, **2012**, 135, 3614-26	11.2	39

177	Relationship between mitochondria and Esynuclein: a study of single substantia nigra neurons. *Archives of Neurology*, **2012**, 69, 385-93		38
176	Consensus-based statements for the management of mitochondrial stroke-like episodes. *Wellcome Open Research*, **2019**, 4, 201	4.8	38
175	Mitochondrial function in muscle from elderly athletes. *Annals of Neurology*, **1997**, 41, 114-6	9.4	36
174	Mitochondrial DNA haplogroup distribution within Leber hereditary optic neuropathy pedigrees. *Journal of Medical Genetics*, **2004**, 41, e41	5.8	36
173	Neuropathological and histochemical changes in a multiple mitochondrial DNA deletion disorder. *Journal of Neuropathology and Experimental Neurology*, **2000**, 59, 621-7	3.1	36
172	Increase in mitochondrial density within axons and supporting cells in response to demyelination in the Plp1 mouse model. *Journal of Neuroscience Research*, **2009**, 87, 452-9	4.4	35
171	Impaired mitochondrial function abolishes gamma oscillations in the hippocampus through an effect on fast-spiking interneurons. *Brain*, **2011**, 134, e180; author reply e181	11.2	35
170	Microangiopathy in the cerebellum of patients with mitochondrial DNA disease. *Brain*, **2012**, 135, 1736-50	11.2	35
169	Novel POLG1 mutations associated with neuromuscular and liver phenotypes in adults and children. *Journal of Medical Genetics*, **2009**, 46, 209-14	5.8	35
168	Defining the importance of mitochondrial gene defects in maternally inherited diabetes by sequencing the entire mitochondrial genome. *Diabetes*, **2002**, 51, 2317-20	0.9	35
167	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	4.9	34
166	Quantification of mitochondrial DNA mutation load. *Aging Cell*, **2009**, 8, 566-72	9.9	34
165	The rise and rise of mitochondrial DNA mutations. *Open Biology*, **2020**, 10, 200061	7	33
164	MT-ND5 Mutation Exhibits Highly Variable Neurological Manifestations at Low Mutant Load. *EBioMedicine*, **2018**, 30, 86-93	8.8	33
163	Analysis of fatty acid oxidation intermediates in cultured fibroblasts to detect mitochondrial oxidation disorders. *Clinical Chemistry*, **1994**, 40, 2267-2275	5.5	33
162	Extensive respiratory chain defects in inhibitory interneurones in patients with mitochondrial disease. *Neuropathology and Applied Neurobiology*, **2016**, 42, 180-93	5.2	32
161	Mitochondrial DNA mutations in aging. *Progress in Molecular Biology and Translational Science*, **2014**, 127, 29-62	4	30
160	So doctor, what exactly is wrong with my muscles? Glutaric aciduria type II presenting in a teenager. *Neuromuscular Disorders*, **2006**, 16, 269-73	2.9	30

159	Clonally expanded mitochondrial DNA deletions within the choroid plexus in multiple sclerosis. *Acta Neuropathologica*, **2012**, 124, 209-20	14.3	29
158	Respiratory chain deficiency in aged spinal motor neurons. *Neurobiology of Aging*, **2014**, 35, 2230-8	5.6	28
157	Cytochrome c oxidase-intermediate fibres: importance in understanding the pathogenesis and treatment of mitochondrial myopathy. *Neuromuscular Disorders*, **2012**, 22, 690-8	2.9	28
156	Mitochondrial disease in pregnancy: a systematic review. *Obstetric Medicine*, **2011**, 4, 90-4	1.5	28
155	Mitochondrial changes within axons in multiple sclerosis: an update. *Current Opinion in Neurology*, **2012**, 25, 221-30	7.1	28
154	OPA1 in multiple mitochondrial DNA deletion disorders. *Neurology*, **2008**, 71, 1829-31	6.5	28
153	Complex mitochondrial DNA rearrangements in individual cells from patients with sporadic inclusion body myositis. *Nucleic Acids Research*, **2016**, 44, 5313-29	20.1	28
152	Pseudo-obstruction, stroke, and mitochondrial dysfunction: A lethal combination. *Annals of Neurology*, **2016**, 80, 686-692	9.4	27
151	The mitochondrial brain: From mitochondrial genome to neurodegeneration. *Biochimica Et Biophysica Acta - Molecular Basis of Disease*, **2010**, 1802, 111-21	6.9	27
150	POLG1, C10ORF2, and ANT1 mutations are uncommon in sporadic progressive external ophthalmoplegia with multiple mitochondrial DNA deletions. *Neurology*, **2006**, 66, 1439-41	6.5	27
149	Review: Central nervous system involvement in mitochondrial disease. *Neuropathology and Applied Neurobiology*, **2017**, 43, 102-118	5.2	26
148	The urinary proteome and metabonome differ from normal in adults with mitochondrial disease. *Kidney International*, **2015**, 87, 610-22	9.9	26
147	Human insulin receptor substrate-1: variant sequences in familial non-insulin-dependent diabetes mellitus. *Diabetic Medicine*, **1996**, 13, 133-8	3.5	26
146	Dissecting the mechanisms underlying the accumulation of mitochondrial DNA deletions in human skeletal muscle. *Human Molecular Genetics*, **2014**, 23, 4612-20	5.6	25
145	Investigation of mitochondrial function in hereditary spastic paraparesis. *NeuroReport*, **2003**, 14, 485-8	1.7	25
144	The role of astrocytes in seizure generation: insights from a novel in vitro seizure model based on mitochondrial dysfunction. *Brain*, **2019**, 142, 391-411	11.2	24
143	Subcellular origin of mitochondrial DNA deletions in human skeletal muscle. *Annals of Neurology*, **2018**, 84, 289-301	9.4	24
142	The low abundance of clonally expanded mitochondrial DNA point mutations in aged substantia nigra neurons. *Aging Cell*, **2009**, 8, 496-8	9.9	24

141	Detection of cytochrome c oxidase activity and mitochondrial proteins in single cells. *Journal of Neuroscience Methods*, **2009**, 184, 310-9	3	24
140	Age-associated mitochondrial DNA mutations cause metabolic remodelling that contributes to accelerated intestinal tumorigenesis. *Nature Cancer*, **2020**, 1, 976-989	15.4	24
139	Quantitative quadruple-label immunofluorescence of mitochondrial and cytoplasmic proteins in single neurons from human midbrain tissue. *Journal of Neuroscience Methods*, **2014**, 232, 143-9	3	23
138	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	5.6	23
137	Triplex real-time PCR--an improved method to detect a wide spectrum of mitochondrial DNA deletions in single cells. *Scientific Reports*, **2015**, 5, 9906	4.9	23
136	Mitochondrial DNA deletions in muscle satellite cells: implications for therapies. *Human Molecular Genetics*, **2013**, 22, 4739-47	5.6	23
135	Loss of myelin-associated glycoprotein in kearns-sayre syndrome. *Archives of Neurology*, **2012**, 69, 490-9		23
134	Mitochondrial myopathy with skeletal muscle cytochrome oxidase deficiency. *Journal of Inherited Metabolic Disease*, **1982**, 5, 27-28	5.4	23
133	The comparative efficacy of antiepileptic drugs for partial and tonic-clonic seizures. *Journal of Neurology, Neurosurgery and Psychiatry*, **1985**, 48, 1073-7	5.5	22
132	Dysferlin mutations and mitochondrial dysfunction. *Neuromuscular Disorders*, **2016**, 26, 782-788	2.9	22
131	Mitochondrial dysfunction in myofibrillar myopathy. *Neuromuscular Disorders*, **2016**, 26, 691-701	2.9	22
130	Therapeutic Manipulation of mtDNA Heteroplasmy: A Shifting Perspective. *Trends in Molecular Medicine*, **2020**, 26, 698-709	11.5	22
129	Pathological mechanisms underlying single large-scale mitochondrial DNA deletions. *Annals of Neurology*, **2018**, 83, 115-130	9.4	21
128	Causes of Death in Adults with Mitochondrial Disease. *JIMD Reports*, **2016**, 26, 103-13	1.9	21
127	Catastrophic presentation of mitochondrial disease due to a mutation in the tRNA(His) gene. *Neurology*, **2004**, 62, 1420-3	6.5	21
126	Frequency of rare mitochondrial DNA mutations in patients with suspected Leber's hereditary optic neuropathy. *Journal of Medical Genetics*, **2003**, 40, e85	5.8	21
125	Mitochondrial dysfunction impairs osteogenesis, increases osteoclast activity, and accelerates age related bone loss. *Scientific Reports*, **2020**, 10, 11643	4.9	21
124	Mitochondrial disease in adults: recent advances and future promise. *Lancet Neurology, The*, **2021**, 20, 573-584	24.1	21

123	Pathogenic variants in MT-ATP6: A United Kingdom-based mitochondrial disease cohort study. *Annals of Neurology*, **2019**, 86, 310-315	9.4	20
122	Late-onset respiratory failure due to TK2 mutations causing multiple mtDNA deletions. *Neurology*, **2013**, 81, 2051-3	6.5	20
121	Gastrointestinal tract involvement associated with the 3243A>G mitochondrial DNA mutation. *Neurology*, **2008**, 70, 1290-2	6.5	20
120	Mitochondrial enteropathy: the primary pathology may not be within the gastrointestinal tract. *Gut* , **2001**, 48, 121-4	19.2	20
119	Investigation of mitochondrial biogenesis defects in single substantia nigra neurons using post-mortem human tissues. *Neurobiology of Disease*, **2020**, 134, 104631	7.5	20
118	Mitochondrial donation: from test tube to clinic. *Lancet, The*, **2018**, 392, 1191-1192	40	20
117	Mitochondrial Donation - Clearing the Final Regulatory Hurdle in the United Kingdom. *New England Journal of Medicine*, **2017**, 376, 171-173	59.2	19
116	Potential compounds for the treatment of mitochondrial disease. *British Medical Bulletin*, **2015**, 116, 5-18	5.4	19
115	Preferential amplification of a human mitochondrial DNA deletion in vitro and in vivo. *Scientific Reports*, **2018**, 8, 1799	4.9	19
114	Novel MTND1 mutations cause isolated exercise intolerance, complex I deficiency and increased assembly factor expression. *Clinical Science*, **2015**, 128, 895-904	6.5	18
113	Scientific and Ethical Issues in Mitochondrial Donation. *New Bioethics*, **2018**, 24, 57-73	1	18
112	Relationship between insulin sensitivity and insulin receptor substrate-1 mutations in non-diabetic relatives of NIDDM families. *Diabetic Medicine*, **1996**, 13, 341-5	3.5	18
111	Investigating complex I deficiency in Purkinje cells and synapses in patients with mitochondrial disease. *Neuropathology and Applied Neurobiology*, **2016**, 42, 477-92	5.2	18
110	Clinical progression of mitochondrial myopathy is associated with the random accumulation of cytochrome c oxidase negative skeletal muscle fibres. *Journal of the Neurological Sciences*, **2003**, 211, 63-6	3.2	16
109	Pathogenic mtDNA mutations causing mitochondrial myopathy: The need for muscle biopsy. *Neurology: Genetics*, **2016**, 2, e82	3.8	16
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