Doug M Turnbull

List of Publications by Year in Descending Order

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	A subcellular cookie cutter for spatial genomics in human tissue.. *Analytical and Bioanalytical Chemistry*, **2022**, 1	4.4	1
301	Mitochondrial complex I subunit deficiency promotes pancreatic β-cell proliferation.. *Molecular Metabolism*, **2022**, 101489	8.8	
300	Detecting respiratory chain defects in osteoblasts from osteoarthritic patients using imaging mass cytometry.. *Bone*, **2022**, 158, 116371	4.7	0
299	Automated quantitative high-throughput multiplex immunofluorescence pipeline to evaluate OXPHOS defects in formalin-fixed human prostate tissue.. *Scientific Reports*, **2022**, 12, 6660	4.9	0
298	Endocrine manifestations and new developments in mitochondrial disease. *Endocrine Reviews*, **2021**,	27.2	1
297	Natural History of Leigh Syndrome: A Study of Disease Burden and Progression. *Annals of Neurology*, **2021**,	9.4	3
296	Risk of cardiac manifestations in adult mitochondrial disease caused by nuclear genetic defects. *Open Heart*, **2021**, 8, e001510	3	1
295	Imaging mass cytometry reveals generalised deficiency in OXPHOS complexes in Parkinson's disease. *Npj Parkinson's Disease*, **2021**, 7, 39	9.7	8
294	Mitochondrial DNA disorders: from pathogenic variants to preventing transmission. *Human Molecular Genetics*, **2021**, 30, R245-R253	5.6	3
293	Mitochondrial disease in adults: recent advances and future promise. *Lancet Neurology, The*, **2021**, 20, 573-584	24.1	21
292	Neuromuscular Junction Abnormalities in Mitochondrial Disease: An Observational Cohort Study. *Neurology: Clinical Practice*, **2021**, 11, 97-104	1.7	3
291	Forecasting stroke-like episodes and outcomes in mitochondrial disease.. *Brain*, **2021**,	11.2	2
290	Age-associated mitochondrial complex I deficiency is linked to increased stem cell proliferation rates in the mouse colon. *Aging Cell*, **2021**, 20, e13321	9.9	5
289	The Role of Mitochondria-Linked Fatty-Acid Uptake-Driven Adipogenesis in Graves Orbitopathy. *Endocrinology*, **2021**, 162,	4.8	1
288	Mutation-Independent Allele-Specific Editing by CRISPR-Cas9, a Novel Approach to Treat Autosomal Dominant Disease. *Molecular Therapy*, **2020**, 28, 1846-1857	11.7	4
287	The rise and rise of mitochondrial DNA mutations. *Open Biology*, **2020**, 10, 200061	7	33
286	Mitochondrial Diseases: Hope for the Future. *Cell*, **2020**, 181, 168-188	56.2	97

285	The feasibility of muscle mitochondrial respiratory chain phenotyping across the cognitive spectrum in Parkinson's disease. *Experimental Gerontology*, **2020**, 138, 110997	4.5	0
284	Mitochondrial morphology and function: two for the price of one!. *Journal of Microscopy*, **2020**, 278, 89-106	1.9	4
283	Measuring the effects of exercise in neuromuscular disorders: a systematic review and meta-analyses. *Wellcome Open Research*, **2020**, 5, 84	4.8	11
282	Mitochondrial isolation: when size matters. *Wellcome Open Research*, **2020**, 5, 226	4.8	1
281	Mitochondrial isolation: when size matters. *Wellcome Open Research*, **2020**, 5, 226	4.8	2
280	Mitochondrial donation - hope for families with mitochondrial DNA disease. *Emerging Topics in Life Sciences*, **2020**, 4, 151-154	3.5	4
279	Investigation of mitochondrial biogenesis defects in single substantia nigra neurons using post-mortem human tissues. *Neurobiology of Disease*, **2020**, 134, 104631	7.5	20
278	Assessment of mitochondrial respiratory chain enzymes in cells and tissues. *Methods in Cell Biology*, **2020**, 155, 121-156	1.8	14
277	Lewy body pathology is more prevalent in older individuals with mitochondrial disease than controls. *Acta Neuropathologica*, **2020**, 139, 219-221	14.3	7
276	Complex I reductions in the nucleus basalis of Meynert in Lewy body dementia: the role of Lewy bodies. *Acta Neuropathologica Communications*, **2020**, 8, 103	7.3	2
275	Mitochondrial dysfunction impairs osteogenesis, increases osteoclast activity, and accelerates age related bone loss. *Scientific Reports*, **2020**, 10, 11643	4.9	21
274	Lower urinary tract dysfunction in adult patients with mitochondrial disease. *Neurourology and Urodynamics*, **2020**, 39, 2253-2263	2.3	4
273	Distinctive Features of Orbital Adipose Tissue (OAT) in Graves'Orbitopathy. *International Journal of Molecular Sciences*, **2020**, 21,	6.3	3
272	Decoding mitochondrial heterogeneity in single muscle fibres by imaging mass cytometry. *Scientific Reports*, **2020**, 10, 15336	4.9	9
271	Age-associated mitochondrial DNA mutations cause metabolic remodelling that contributes to accelerated intestinal tumorigenesis. *Nature Cancer*, **2020**, 1, 976-989	15.4	24
270	Ultrasensitive deletion detection links mitochondrial DNA replication, disease, and aging. *Genome Biology*, **2020**, 21, 248	18.3	14
269	Exercise Training and Neurodegeneration in Mitochondrial Disorders: Insights From the Harlequin Mouse. *Frontiers in Physiology*, **2020**, 11, 594223	4.6	1
268	Systematic review of cognitive deficits in adult mitochondrial disease. *European Journal of Neurology*, **2020**, 27, 3-17	6	10

267	Therapeutic Manipulation of mtDNA Heteroplasmy: A Shifting Perspective. *Trends in Molecular Medicine*, **2020**, 26, 698-709	11.5	22
266	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
265	Quantitative 3D Mapping of the Human Skeletal Muscle Mitochondrial Network. *Cell Reports*, **2019**, 26, 996-1009.e4	10.6	50
264	Cognitive deficits in adult m.3243A>G- and m.8344A>G-related mitochondrial disease: importance of correcting for baseline intellectual ability. *Annals of Clinical and Translational Neurology*, **2019**, 6, 826-836	5.3	6
263	Understanding mitochondrial DNA maintenance disorders at the single muscle fibre level. *Nucleic Acids Research*, **2019**, 47, 7430-7443	20.1	9
262	Pathogenic variants in MT-ATP6: A United Kingdom-based mitochondrial disease cohort study. *Annals of Neurology*, **2019**, 86, 310-315	9.4	20
261	Mitochondrial Neurodegenerative Disorders I: Parkinsonism and Cognitive Deficits **2019**, 223-239		
260	Reproductive Options for Women with Mitochondrial Disease **2019**, 371-382		2
259	Mitochondrial Dysfunction in Parkinson's Disease-Cause or Consequence?. *Biology*, **2019**, 8,	4.9	85
258	Mitochondrial Donation - Which Women Could Benefit?. *New England Journal of Medicine*, **2019**, 380, 1971-1972	59.2	14
257	Mitochondrial Medicine: A Historical Point of View **2019**, 1-18		
256	Height as a Clinical Biomarker of Disease Burden in Adult Mitochondrial Disease. *Journal of Clinical Endocrinology and Metabolism*, **2019**, 104, 2057-2066	5.6	12
255	Effects of obesity and weight loss on mitochondrial structure and function and implications for colorectal cancer risk. *Proceedings of the Nutrition Society*, **2019**, 78, 426-437	2.9	9
254	Dissecting the neuronal vulnerability underpinning Alpers' syndrome: a clinical and neuropathological study. *Brain Pathology*, **2019**, 29, 97-113	6	7
253	Design and baseline characteristics of the Biomarkers Of Risk In Colorectal Cancer (BORICC) Follow-Up study: A 12+ years follow-up. *Nutrition and Health*, **2019**, 25, 231-238	2.1	1
252	Consensus-based statements for the management of mitochondrial stroke-like episodes. *Wellcome Open Research*, **2019**, 4, 201	4.8	38
251	A case-comparison study of pregnant women with mitochondrial disease - what to expect?. *BJOG: an International Journal of Obstetrics and Gynaecology*, **2019**, 126, 1380-1389	3.7	12
250	MT-ND5 Mutation Exhibits Highly Variable Neurological Manifestations at Low Mutant Load. *EBioMedicine*, **2018**, 30, 86-93	8.8	33

249	The adjunctive application of transcranial direct current stimulation in the management of de novo refractory epilepsia partialis continua in adolescent-onset -related mitochondrial disease. *Epilepsia Open*, **2018**, 3, 103-108	4	13
248	Preferential amplification of a human mitochondrial DNA deletion in vitro and in vivo. *Scientific Reports*, **2018**, 8, 1799	4.9	19
247	Progress in mitochondrial replacement therapies. *Nature Reviews Molecular Cell Biology*, **2018**, 19, 71-72	48.7	42
246	Pathological mechanisms underlying single large-scale mitochondrial DNA deletions. *Annals of Neurology*, **2018**, 83, 115-130	9.4	21
245	Topoisomerase 3 Is Required for Decatenation and Segregation of Human mtDNA. *Molecular Cell*, **2018**, 69, 9-23.e6	17.6	61
244	Predominant Asymmetrical Stem Cell Fate Outcome Limits the Rate of Niche Succession in Human Colonic Crypts. *EBioMedicine*, **2018**, 31, 166-173	8.8	14
243	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
242	Scientific and Ethical Issues in Mitochondrial Donation. *New Bioethics*, **2018**, 24, 57-73	1	18
241	Impact of age on the association between cardiac high-energy phosphate metabolism and cardiac power in women. *Heart*, **2018**, 104, 111-118	5.1	9
240	Roles of Mitochondrial DNA Mutations in Stem Cell Ageing. *Genes*, **2018**, 9,	4.2	13
239	Subcellular origin of mitochondrial DNA deletions in human skeletal muscle. *Annals of Neurology*, **2018**, 84, 289-301	9.4	24
238	mtDNA heteroplasmy level and copy number indicate disease burden in m.3243A>G mitochondrial disease. *EMBO Molecular Medicine*, **2018**, 10,	12	115
237	Inherited pathogenic mitochondrial DNA mutations and gastrointestinal stem cell populations. *Journal of Pathology*, **2018**, 246, 427-432	9.4	9
236	Diagnosis and Treatment of Mitochondrial Myopathies. *Neurotherapeutics*, **2018**, 15, 943-953	6.4	46
235	Mitochondrial donation: from test tube to clinic. *Lancet, The*, **2018**, 392, 1191-1192	40	20
234	Mitochondrial respiratory chain function and content are preserved in the skeletal muscle of active very old men and women. *Experimental Gerontology*, **2018**, 113, 80-85	4.5	10
233	Skeletal muscle mitochondrial oxidative phosphorylation function in idiopathic pulmonary arterial hypertension: in vivo and in vitro study. *Pulmonary Circulation*, **2018**, 8, 2045894018768290	2.7	7
232	Review: Central nervous system involvement in mitochondrial disease. *Neuropathology and Applied Neurobiology*, **2017**, 43, 102-118	5.2	26

231	Fatty acid oxidation is required for the respiration and proliferation of malignant glioma cells. *Neuro-Oncology*, **2017**, 19, 43-54	1	123
230	Recent Advances in Mitochondrial Disease. *Annual Review of Genomics and Human Genetics*, **2017**, 18, 257-275	9.7	149
229	Association of mitochondrial respiratory chain deficiency in older men with muscle mass and physical performance: findings from the Hertfordshire Sarcopenia Study. *Lancet, The*, **2017**, 389, S87	40	2
228	Mitochondrial Donation - Clearing the Final Regulatory Hurdle in the United Kingdom. *New England Journal of Medicine*, **2017**, 376, 171-173	59.2	19
227	Mitochondrial Nanotunnels. *Trends in Cell Biology*, **2017**, 27, 787-799	18.3	56
226	Decreased male reproductive success in association with mitochondrial dysfunction. *European Journal of Human Genetics*, **2017**, 25, 1162-1164	5.3	12
225	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
224	Pathophysiology of exercise intolerance in chronic diseases: the role of diminished cardiac performance in mitochondrial and heart failure patients. *Open Heart*, **2017**, 4, e000632	3	13
223	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
222	Novel variants associated with late-onset de novo status epilepticus and progressive ataxia. *Neurology: Genetics*, **2017**, 3, e181	3.8	1
221	Using a quantitative quadruple immunofluorescent assay to diagnose isolated mitochondrial Complex I deficiency. *Scientific Reports*, **2017**, 7, 15676	4.9	14
220	The genetics and pathology of mitochondrial disease. *Journal of Pathology*, **2017**, 241, 236-250	9.4	222
219	Mitochondrial disease: genetics and management. *Journal of Neurology*, **2016**, 263, 179-91	5.5	48
218	Extensive respiratory chain defects in inhibitory interneurones in patients with mitochondrial disease. *Neuropathology and Applied Neurobiology*, **2016**, 42, 180-93	5.2	32
217	The Spectrum of Mitochondrial Ultrastructural Defects in Mitochondrial Myopathy. *Scientific Reports*, **2016**, 6, 30610	4.9	99
216	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
215	Pseudo-obstruction, stroke, and mitochondrial dysfunction: A lethal combination. *Annals of Neurology*, **2016**, 80, 686-692	9.4	27
214	Expanding Our Understanding of mtDNA Deletions. *Cell Metabolism*, **2016**, 24, 3-4	24.6	12

213	Cell-permeable succinate prodrugs bypass mitochondrial complex I deficiency. *Nature Communications*, **2016**, 7, 12317	17.4	74
212	Unique quadruple immunofluorescence assay demonstrates mitochondrial respiratory chain dysfunction in osteoblasts of aged and PolgA(-/-) mice. *Scientific Reports*, **2016**, 6, 31907	4.9	8
211	Mitochondria, the Synapse, and Neurodegeneration **2016**, 219-239		1
210	Localization of MRP-1 to the outer mitochondrial membrane by the chaperone protein HSP90 *FASEB Journal*, **2016**, 30, 1712-23	0.9	8
209	Causes of Death in Adults with Mitochondrial Disease. *JIMD Reports*, **2016**, 26, 103-13	1.9	21
208	The ageing neuromuscular system and sarcopenia: a mitochondrial perspective. *Journal of Physiology*, **2016**, 594, 4499-512	3.9	66
207	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
206	Applying the Airbrakes: Treating Mitochondrial Disease with Hypoxia. *Molecular Cell*, **2016**, 62, 5-6	17.6	4
205	Complex mitochondrial DNA rearrangements in individual cells from patients with sporadic inclusion body myositis. *Nucleic Acids Research*, **2016**, 44, 5313-29	20.1	28
204	Clinical, Genetic, and Radiological Features of Extrapyramidal Movement Disorders in Mitochondrial Disease. *JAMA Neurology*, **2016**, 73, 668-74	17.2	46
203	Dysferlin mutations and mitochondrial dysfunction. *Neuromuscular Disorders*, **2016**, 26, 782-788	2.9	22
202	Pathogenic mtDNA mutations causing mitochondrial myopathy: The need for muscle biopsy. *Neurology: Genetics*, **2016**, 2, e82	3.8	16
201	Spectrum of Movement Disorders in Mitochondrial Disorders-Reply. *JAMA Neurology*, **2016**, 73, 1254-1255	17.2	
200	Mitochondrial dysfunction in myofibrillar myopathy. *Neuromuscular Disorders*, **2016**, 26, 691-701	2.9	22
199	Mitochondrial DNA Depletion in Respiratory Chain-Deficient Parkinson Disease Neurons. *Annals of Neurology*, **2016**, 79, 366-78	9.4	131
198	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
197	Novel MTND1 mutations cause isolated exercise intolerance, complex I deficiency and increased assembly factor expression. *Clinical Science*, **2015**, 128, 895-904	6.5	18
196	Mutations causing mitochondrial disease: What is new and what challenges remain?. *Science*, **2015**, 349, 1494-9	33.3	204

195	Mitochondrial and inflammatory changes in sporadic inclusion body myositis. *Neuropathology and Applied Neurobiology*, **2015**, 41, 288-303	5.2	49
194	A CLINICAL AUDIT OF ACUTE MANAGEMENT OF STROKE-LIKE EPISODES FROM A NATIONAL MITOCHONDRIAL DISEASE CENTRE. *Journal of Neurology, Neurosurgery and Psychiatry*, **2015**, 86, e4.161-e4	5.5	
193	The diagnosis of posterior reversible encephalopathy syndrome. *Lancet Neurology, The*, **2015**, 14, 1073	24.1	4
192	Aggregated □synuclein and complex I deficiency: exploration of their relationship in differentiated neurons. *Cell Death and Disease*, **2015**, 6, e1820	9.8	104
191	Potential compounds for the treatment of mitochondrial disease. *British Medical Bulletin*, **2015**, 116, 5-18	5.4	19
190	The urinary proteome and metabonome differ from normal in adults with mitochondrial disease. *Kidney International*, **2015**, 87, 610-22	9.9	26
189	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
188	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
187	Adult-onset myoclonus ataxia associated with the mitochondrial m.8993T>C "NARP" mutation. *Movement Disorders*, **2015**, 30, 1432-3	7	2
186	Mitochondrial donation--how many women could benefit?. *New England Journal of Medicine*, **2015**, 372, 885-887	59.2	69
185	Ageing and Parkinson□s disease: why is advancing age the biggest risk factor?. *Ageing Research Reviews*, **2014**, 14, 19-30	12	487
184	Mitochondrial DNA disease-molecular insights and potential routes to a cure. *Experimental Cell Research*, **2014**, 325, 38-43	4.2	67
183	Disease progression in patients with single, large-scale mitochondrial DNA deletions. *Brain*, **2014**, 137, 323-34	11.2	81
182	Mitochondrial DNA mutations in aging. *Progress in Molecular Biology and Translational Science*, **2014** , 127, 29-62	4	30
181	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
180	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
179	A national perspective on prenatal testing for mitochondrial disease. *European Journal of Human Genetics*, **2014**, 22, 1255-9	5.3	48
178	Dissecting the mechanisms underlying the accumulation of mitochondrial DNA deletions in human skeletal muscle. *Human Molecular Genetics*, **2014**, 23, 4612-20	5.6	25

177	Respiratory chain deficiency in aged spinal motor neurons. *Neurobiology of Aging*, **2014**, 35, 2230-8	5.6	28
176	Accurate measurement of mitochondrial DNA deletion level and copy number differences in human skeletal muscle. *PLoS ONE*, **2014**, 9, e114462	3.7	40
175	The impact of pathogenic mitochondrial DNA mutations on substantia nigra neurons. *Journal of Neuroscience*, **2013**, 33, 10790-801	6.6	61
174	Mitochondrial DNA deletions and depletion within paraspinal muscles. *Neuropathology and Applied Neurobiology*, **2013**, 39, 377-89	5.2	14
173	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
172	Initial development and validation of a mitochondrial disease quality of life scale. *Neuromuscular Disorders*, **2013**, 23, 324-9	2.9	8
171	Linking the metabolic state and mitochondrial DNA in chronic disease, health, and aging. *Diabetes*, **2013**, 62, 672-8	0.9	69
170	Monitoring mitochondrial dynamics and complex I dysfunction in neurons: implications for Parkinson's disease. *Biochemical Society Transactions*, **2013**, 41, 1618-24	5.1	11
169	Late-onset respiratory failure due to TK2 mutations causing multiple mtDNA deletions. *Neurology*, **2013**, 81, 2051-3	6.5	20
168	Mitochondrial DNA deletions in muscle satellite cells: implications for therapies. *Human Molecular Genetics*, **2013**, 22, 4739-47	5.6	23
167	No excess of mitochondrial DNA deletions within muscle in progressive multiple sclerosis. *Multiple Sclerosis Journal*, **2013**, 19, 1858-66	5	11
166	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
165	Neuro-Sweet's disease. *Practical Neurology*, **2012**, 12, 126-30	2.4	8
164	Mitochondrial DNA deletions cause the biochemical defect observed in Alzheimer's disease. *Neurobiology of Aging*, **2012**, 33, 2210-4	5.6	61
163	Cytochrome c oxidase-intermediate fibres: importance in understanding the pathogenesis and treatment of mitochondrial myopathy. *Neuromuscular Disorders*, **2012**, 22, 690-8	2.9	28
162	Mitochondrial DNA and disease. *Journal of Pathology*, **2012**, 226, 274-86	9.4	195
161	Clonally expanded mitochondrial DNA deletions within the choroid plexus in multiple sclerosis. *Acta Neuropathologica*, **2012**, 124, 209-20	14.3	29
160	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

159	What is influencing the phenotype of the common homozygous polymerase-γ mutation p.Ala467Thr?. *Brain*, **2012**, 135, 3614-26	11.2	39
158	Relationship between mitochondria and α-synuclein: a study of single substantia nigra neurons. *Archives of Neurology*, **2012**, 69, 385-93		38
157	Microangiopathy in the cerebellum of patients with mitochondrial DNA disease. *Brain*, **2012**, 135, 1736-50	11.2	35
156	Mitochondrial changes within axons in multiple sclerosis: an update. *Current Opinion in Neurology*, **2012**, 25, 221-30	7.1	28
155	Sensory neuronopathy in patients harbouring recessive polymerase γ mutations. *Brain*, **2012**, 135, 62-71	11.2	53
154	Cardiac involvement in mitochondrial DNA disease: clinical spectrum, diagnosis, and management. *European Heart Journal*, **2012**, 33, 3023-33	9.5	138
153	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
152	Management of patients with dental disease and mitochondrial disorders. *Dental Update*, **2012**, 39, 654-5	0.3	0
151	Loss of myelin-associated glycoprotein in kearns-sayre syndrome. *Archives of Neurology*, **2012**, 69, 490-9		23
150	Hyperventilation during the EEG is safe in mitochondrial disease. *Clinical Neurophysiology*, **2011**, 122, 1270-1	4.3	1
149	A new biomarker for mitochondrial disease. *Lancet Neurology, The*, **2011**, 10, 777-8	24.1	4
148	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
147	Mitochondrial DNA deletions and neurodegeneration in multiple sclerosis. *Annals of Neurology*, **2011**, 69, 481-92	9.4	255
146	Increased mitochondrial content in remyelinated axons: implications for multiple sclerosis. *Brain*, **2011**, 134, 1901-13	11.2	110
145	Mitochondrial Mutations: Newly Discovered Players in Neuronal Degeneration. *Neuroscientist*, **2011** , 17, 645-658	7.6	12
144	Mitochondrial disease in pregnancy: a systematic review. *Obstetric Medicine*, **2011**, 4, 90-4	1.5	28
143	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
142	Mitochondrial DNA disease: new options for prevention. *Human Molecular Genetics*, **2011**, 20, R168-74	5.6	63

141	RRM2B mutations are frequent in familial PEO with multiple mtDNA deletions. *Neurology*, **2011**, 76, 2038-4	6.9	54
140	Mitochondrial mutations: newly discovered players in neuronal degeneration. *Neuroscientist*, **2011**, 17, 645-58	7.6	7
139	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
138	Mitochondrial DNA mutations affect calcium handling in differentiated neurons. *Brain*, **2010**, 133, 787-96	11.2	42
137	The clinical, histochemical, and molecular spectrum of PEO1 (Twinkle)-linked adPEO. *Neurology*, **2010**, 74, 1619-26	6.5	73
136	Mechanism of neurodegeneration of neurons with mitochondrial DNA mutations. *Brain*, **2010**, 133, 797-807	11.2	91
135	The mitochondrial brain: From mitochondrial genome to neurodegeneration. *Biochimica Et Biophysica Acta - Molecular Basis of Disease*, **2010**, 1802, 111-21	6.9	27
134	Multi-system neurological disease is common in patients with OPA1 mutations. *Brain*, **2010**, 133, 771-86	11.2	314
133	Defects in multiple complexes of the respiratory chain are present in ageing human colonic crypts. *Experimental Gerontology*, **2010**, 45, 573-9	4.5	43
132	Mitochondrial DNA and genetic disease. *Essays in Biochemistry*, **2010**, 47, 139-51	7.6	13
131	Modelling mitochondrial DNA mutations in bacterial cytochrome c oxidase: link to colon cancer?. *Proceedings of the National Academy of Sciences of the United States of America*, **2009**, 106, E57	11.5	1
130	Pathogenic mitochondrial tRNA mutations--which mutations are inherited and why?. *Human Mutation*, **2009**, 30, E984-92	4.7	42
129	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
128	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
127	Transmitochondrial embryonic stem cells containing pathogenic mtDNA mutations are compromised in neuronal differentiation. *Cell Proliferation*, **2009**, 42, 413-24	7.9	58
126	Batteries not included: diagnosis and management of mitochondrial disease. *Journal of Internal Medicine*, **2009**, 265, 210-28	10.8	62
125	A diagnostic tattoo. *Clinical Genetics*, **2009**, 75, 37-8	4	4
124	The low abundance of clonally expanded mitochondrial DNA point mutations in aged substantia nigra neurons. *Aging Cell*, **2009**, 8, 496-8	9.9	24

123	Quantification of mitochondrial DNA mutation load. *Aging Cell*, **2009**, 8, 566-72	9.9	34
122	Detection of cytochrome c oxidase activity and mitochondrial proteins in single cells. *Journal of Neuroscience Methods*, **2009**, 184, 310-9	3	24
121	Urine heteroplasmy is the best predictor of clinical outcome in the m.3243A>G mtDNA mutation. *Neurology*, **2009**, 72, 568-9	6.5	81
120	Mitochondrial DNA mutations and ageing. *Biochimica Et Biophysica Acta - General Subjects*, **2009**, 1790, 1015-20	4	43
119	Mitochondrial changes within axons in multiple sclerosis. *Brain*, **2009**, 132, 1161-74	11.2	338
118	Novel POLG1 mutations associated with neuromuscular and liver phenotypes in adults and children. *Journal of Medical Genetics*, **2009**, 46, 209-14	5.8	35
117	What causes mitochondrial DNA deletions in human cells?. *Nature Genetics*, **2008**, 40, 275-9	36.3	302
116	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
115	Resistance training in patients with single, large-scale deletions of mitochondrial DNA. *Brain*, **2008**, 131, 2832-40	11.2	124
114	Melas associated with mutations in the polg1 gene. *Neurology*, **2008**, 70, 1054; author reply 1054-5	6.5	8
113	OPA1 in multiple mitochondrial DNA deletion disorders. *Neurology*, **2008**, 71, 1829-31	6.5	28
112	Gastrointestinal tract involvement associated with the 3243A>G mitochondrial DNA mutation. *Neurology*, **2008**, 70, 1290-2	6.5	20
111	Age-related decline in mitochondrial DNA copy number in isolated human pancreatic islets. *Diabetologia*, **2008**, 51, 1440-3	10.3	61
110	Dopaminergic midbrain neurons are the prime target for mitochondrial DNA deletions. *Journal of Neurology*, **2008**, 255, 1231-5	5.5	68
109	Mitochondrial DNA mutations in disease, aging, and neurodegeneration. *Annals of the New York Academy of Sciences*, **2008**, 1147, 21-9	6.5	114
108	Mitochondrial disease--its impact, etiology, and pathology. *Current Topics in Developmental Biology*, **2007**, 77, 113-55	5.3	145
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