Patrick F Chinnery

List of Publications by Year
in descending order

171 papers	23,545 citations	12330 69 h-index	8167 148 g-index
173 all docs	173 docs citations	173 times ranked	22780 citing authors

#	Article	IF	Citations
1	High diagnostic rate of trio exome sequencing in consanguineous families with neurogenetic diseases. Brain, 2022, 145, 1507-1518.	7.6	14
2	Heteroplasmic mitochondrial DNA variants in cardiovascular diseases. PLoS Genetics, 2022, 18, e1010068.	3.5	19
3	Extreme heterogeneity of human mitochondrial DNA from organelles to populations. Nature Reviews Genetics, 2021, 22, 106-118.	16.3	139
4	[11C]PK11195-PET Brain Imaging of the Mitochondrial Translocator Protein in Mitochondrial Disease. Neurology, 2021, 96, e2761-e2773.	1.1	7
5	Oxygen tension modulates the mitochondrial genetic bottleneck and influences the segregation of a heteroplasmic mtDNA variant in vitro. Communications Biology, 2021, 4, 584.	4.4	7
6	An atlas of mitochondrial DNA genotypeâ€"phenotype associations in the UK Biobank. Nature Genetics, 2021, 53, 982-993.	21.4	72
7	MitoPhen database: a human phenotype ontology-based approach to identify mitochondrial DNA diseases. Nucleic Acids Research, 2021, 49, 9686-9695.	14.5	14
8	Mitochondrial DNA variants modulate N-formylmethionine, proteostasis and risk of late-onset human diseases. Nature Medicine, 2021, 27, 1564-1575.	30.7	40
9	Cell reprogramming shapes the mitochondrial DNA landscape. Nature Communications, 2021, 12, 5241.	12.8	21
10	Shortening the diagnostic odysseyâ€"the impact of whole genome sequencing in the NHS. BMJ, The, 2021, 375, n2683.	6.0	2
11	Use of whole genome sequencing to determine genetic basis of suspected mitochondrial disorders: cohort study. BMJ, The, 2021, 375, e066288.	6.0	42
12	Single-molecule mitochondrial DNA sequencing shows no evidence of CpG methylation in human cells and tissues. Nucleic Acids Research, 2021, 49, 12757-12768.	14.5	18
13	Mitochondrial DNA heteroplasmy is modulated during oocyte development propagating mutation transmission. Science Advances, 2021, 7, eabi5657.	10.3	22
14	Mitochondrial heteroplasmy beyond the oocyte bottleneck. Seminars in Cell and Developmental Biology, 2020, 97, 156-166.	5.0	57
15	Visualizing, quantifying, and manipulating mitochondrial DNA in vivo. Journal of Biological Chemistry, 2020, 295, 17588-17601.	3.4	14
16	Mitochondrial Diseases: A Diagnostic Revolution. Trends in Genetics, 2020, 36, 702-717.	6.7	73
17	Heredity and segregation of mtDNA. , 2020, , 87-107.		0
18	Recurrent horizontal transfer identifies mitochondrial positive selection in a transmissible cancer. Nature Communications, 2020, 11, 3059.	12.8	18

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19	Nuclear-mitochondrial DNA segments resemble paternally inherited mitochondrial DNA in humans. Nature Communications, 2020, 11, 1740.	12.8	75
20	Chronic pain is common in mitochondrial disease. Neuromuscular Disorders, 2020, 30, 413-419.	0.6	26
21	Cracking the enigma of mitochondrial-DNA variants and cancer. Nature Metabolism, 2020, 2, 221-222.	11.9	0
22	Metabolic effects of bezafibrate in mitochondrial disease. EMBO Molecular Medicine, 2020, 12, e11589.	6.9	45
23	Mitochondrial disorders due to mutations in the nuclear genome. , 2020, , 415-425.		0
24	Assessing mitochondrial heteroplasmy using next generation sequencing: A note of caution. Mitochondrion, 2019, 46, 302-306.	3.4	56
25	De-fusing mitochondria defuses the mtDNA time-bomb. Cell Research, 2019, 29, 781-782.	12.0	5
26	Diagnosis of â€˜possibleâ€™ mitochondrial disease: an existential crisis. Journal of Medical Genetics, 2019, 56, 123-130.	3.2	42
27	Pathogenic variants in MTâ€ATP6: A United Kingdomâ€“based mitochondrial disease cohort study. Annals of Neurology, 2019, 86, 310-315.	5.3	33
28	Germline selection shapes human mitochondrial DNA diversity. Science, 2019, 364, .	12.6	178
29	Mitochondria in neuroinflammation â€“ Multiple sclerosis (MS), leber hereditary optic neuropathy (LHON) and LHON-MS. Neuroscience Letters, 2019, 710, 132932.	2.1	52
30	Mitochondrial Cardiovascular Diseases. , 2018, , 239-258.		1
31	Mitochondrial DNA Heteroplasmy and Purifying Selection in the Mammalian Female Germ Line. Development Growth and Differentiation, 2018, 60, 21-32.	1.5	49
32	Segregation of mitochondrial DNA heteroplasmy through a developmental genetic bottleneck in human embryos. Nature Cell Biology, 2018, 20, 144-151.	10.3	182
33	Oldies but Goldies mtDNA Population Variants and Neurodegenerative Diseases. Frontiers in Neuroscience, 2018, 12, 682.	2.8	50
34	First-line genomic diagnosis of mitochondrial disorders. Nature Reviews Genetics, 2018, 19, 399-400.	16.3	49
35	Mutation-specific effects in germline transmission of pathogenic mtDNA variants. Human Reproduction, 2018, 33, 1331-1341.	0.9	36
36	The mitochondrial DNA genetic bottleneck: inheritance and beyond. Essays in Biochemistry, 2018, 62, 225-234.	4.7	85

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37	Oxygen in mitochondrial disease: can there be too much of a good thing?. Journal of Inherited Metabolic Disease, 2018, 41, 761-763.	3.6	11
38	The Human Phenotype Ontology in 2017. Nucleic Acids Research, 2017, 45, D865-D876.	14.5	699
39	Mitochondrial DNA point mutations and relative copy number in 1363 disease and control human brains. Acta Neuropathologica Communications, 2017, 5, 13.	5.2	83
40	Genetic heterogeneity of motor neuropathies. Neurology, 2017, 88, 1226-1234.	1.1	81
41	Monitoring clinical progression with mitochondrial disease biomarkers. Brain, 2017, 140, 2530-2540.	7.6	44
42	Mutations in mitochondrial DNA causing tubulointerstitial kidney disease. PLoS Genetics, 2017, 13, e1006620.	3.5	52
43	Background sequence characteristics influence the occurrence and severity of disease-causing mtDNA mutations. PLoS Genetics, 2017, 13, e1007126.	3.5	49
44	MSeqDR: A Centralized Knowledge Repository and Bioinformatics Web Resource to Facilitate Genomic Investigations in Mitochondrial Disease. Human Mutation, 2016, 37, 540-548.	2.5	42
45	A multiple sclerosisâ€like disorder in patients with OPA1 mutations. Annals of Clinical and Translational Neurology, 2016, 3, 723-729.	3.7	27
46	Mitochondrial diseases. Nature Reviews Disease Primers, 2016, 2, 16080.	30.5	1,001
47	Both mitochondrial DNA and mitonuclear gene mutations cause hearing loss through cochlear dysfunction. Brain, 2016, 139, e33-e33.	7.6	15
48	Reply: Parsing the differences in affected with LHON: genetic versus environmental triggers of disease conversion. Brain, 2016, 139, e18-e18.	7.6	13
49	Mitochondrial DNA sequence characteristics modulate the size of the genetic bottleneck. Human Molecular Genetics, 2016, 25, 1031-1041.	2.9	53
50	Mitochondrial disease in adults: what's old and what's new?. EMBO Molecular Medicine, 2015, 7, 1503-1512.	6.9	110
51	Adult Onset Leigh Syndrome in the Intensive Care Setting: A Novel Presentation of a C12orf65 Related Mitochondrial Disease. Journal of Neuromuscular Diseases, 2015, 2, 409-419.	2.6	22
52	Mitochondrial dysfunction in aging: Much progress but many unresolved questions. Biochimica Et Biophysica Acta - Bioenergetics, 2015, 1847, 1347-1353.	1.0	267
53	Prevalence of nuclear and mitochondrial <scp>DNA</scp> mutations related to adult mitochondrial disease. Annals of Neurology, 2015, 77, 753-759.	5.3	706
54	A Unique Gene Regulatory Network Resets the Human Germline Epigenome for Development. Cell, 2015, 161, 1453-1467.	28.9	556

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55	Mitochondrial DNA mutations in neurodegeneration. Biochimica Et Biophysica Acta - Bioenergetics, 2015, 1847, 1401-1411.	1.0	120
56	Clonal Expansion of Secondary Mitochondrial DNA Deletions Associated With Spinocerebellar Ataxia Type 28. JAMA Neurology, 2015, 72, 106.	9.0	41
57	Mitochondrial Donation â€" How Many Women Could Benefit?. New England Journal of Medicine, 2015, 372, 885-887.	27.0	87
58	Deep Resequencing of Mitochondrial DNA. Methods in Molecular Biology, 2015, 1264, 59-66.	0.9	9
59	Mitochondrial disease: mimics and chameleons. Practical Neurology, 2015, 15, 424-435.	1.1	14
60	Extreme-Depth Re-sequencing of Mitochondrial DNA Finds No Evidence of Paternal Transmission in Humans. PLoS Genetics, 2015, 11, e1005040.	3.5	62
61	The dynamics of mitochondrial DNA heteroplasmy: implications for human health and disease. Nature Reviews Genetics, 2015, 16, 530-542.	16.3	679
62	Prevalence of neurogenetic disorders in the North of England. Neurology, 2015, 85, 1195-1201.	1.1	26
63	Disturbed mitochondrial dynamics and neurodegenerative disorders. Nature Reviews Neurology, 2015, 11, 11-24.	10.1	533
64	Single-Cell Analysis of Mitochondrial DNA. Methods in Molecular Biology, 2015, 1264, 67-76.	0.9	6
65	Origins and functional consequences of somatic mitochondrial DNA mutations in human cancer. ELife, 2014, 3, .	6.0	318
66	Accurate mitochondrial DNA sequencing using off-target reads provides a single test to identify pathogenic point mutations. Genetics in Medicine, 2014, 16, 962-971.	2.4	64
67	The Challenges of Mitochondrial Replacement. PLoS Genetics, 2014, 10, e1004315.	3.5	61
68	Recent Mitochondrial DNA Mutations Increase the Risk of Developing Common Late-Onset Human Diseases. PLoS Genetics, 2014, 10, e1004369.	3.5	131
69	Use of Whole-Exome Sequencing to Determine the Genetic Basis of Multiple Mitochondrial Respiratory Chain Complex Deficiencies. JAMA - Journal of the American Medical Association, 2014, 312, 68.	7.4	304
70	Mutations in the SPG7 gene cause chronic progressive external ophthalmoplegia through disordered mitochondrial DNA maintenance. Brain, 2014, 137, 1323-1336.	7.6	151
71	Mitochondrial DNA mutations in ageing and disease: implications for HIV?. Antiviral Therapy, 2014, 20, 109-120.	1.0	12
72	Age-Related Mitochondrial DNA Depletion and the Impact on Pancreatic Beta Cell Function. PLoS ONE, 2014, 9, e115433.	2.5	44

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73	Dominant Optic Atrophy: Novel OPA1 Mutations and Revised Prevalence Estimates. Ophthalmology, 2013, 120, 1712-1712.e1.	5.2	57
74	Early neuropsychiatry features in neuroferritinopathy. Movement Disorders, 2013, 28, 1310-1313.	3.9	11
75	Hereditary mtDNA Heteroplasmy: A Baseline for Aging?. Cell Metabolism, 2013, 18, 463-464.	16.2	44
76	Neuroferritinopathy. International Review of Neurobiology, 2013, 110, 91-123.	2.0	24
77	Diagnosis and treatment of mitochondrial myopathies. Annals of Medicine, 2013, 45, 4-16.	3.8	144
78	Clinical features of MS associated with Leber hereditary optic neuropathy mtDNA mutations. Neurology, 2013, 81, 2073-2081.	1.1	100
79	Defects in Mitochondrial Dynamics and Mitochondrial DNA Instability. , 2013, , 141-161.		1
80	Universal heteroplasmy of human mitochondrial DNA. Human Molecular Genetics, 2013, 22, 384-390.	2.9	344
81	Frailty and mortality are not influenced by mitochondrial DNA haplotypes in the very old. Neurobiology of Aging, 2013, 34, 2889.e1-2889.e4.	3.1	12
82	Preventing the transmission of pathogenic mitochondrial DNA mutations: can we achieve long-term benefits from germ-line gene transfer?. Human Reproduction, 2013, 28, 554-559.	0.9	31
83	Concentric hypertrophic remodelling and subendocardial dysfunction in mitochondrial DNA point mutation carriersâ€. European Heart Journal Cardiovascular Imaging, 2013, 14, 650-658.	1.2	30
84	Two-stage association study and meta-analysis of mitochondrial DNA variants in Parkinson disease. Neurology, 2013, 80, 2042-2048.	1.1	129
85	Visual and psychological morbidity among patients with autosomal dominant optic atrophy. Acta Ophthalmologica, 2013, 91, e413-4.	1.1	10
86	Adult-onset spinocerebellar ataxia syndromes due toMTATP6mutations. Journal of Neurology, Neurosurgery and Psychiatry, 2012, 83, 883-886.	1.9	42
87	OPA1 mutations induce mtDNA proliferation in leukocytes of patients with dominant optic atrophy. Neurology, 2012, 79, 1515-1517.	1.1	11
88	Dysfunctional mitochondrial maintenance: what breaks the circle of life?. Brain, 2012, 135, 9-11.	7.6	21
89	MFN2 mutations cause compensatory mitochondrial DNA proliferation. Brain, 2012, 135, e219-e219.	7.6	41
90	Spastic paraplegia gene 7 in patients with spasticity and/or optic neuropathy. Brain, 2012, 135, 2980-2993.	7.6	148

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91	Neuroferritinopathy: Update on Clinical Features and Pathogenesis. Current Drug Targets, 2012, 13, 1200-1203.	2.1	15
92	Variation in germline mtDNA heteroplasmy is determined prenatally but modified during subsequent transmission. Nature Genetics, 2012, 44, 1282-1285.	21.4	128
93	Epigenetics, epidemiology and mitochondrial DNA diseases. International Journal of Epidemiology, 2012, 41, 177-187.	1.9	146
94	A new phenotype of brain iron accumulation with dystonia, optic atrophy, and peripheral neuropathy. Movement Disorders, 2012, 27, 789-793.	3.9	41
95	Neuroferritinopathy: a new inborn error of iron metabolism. Neurogenetics, 2012, 13, 93-96.	1.4	51
96	Current Concepts and Controversies in Neurodegeneration with Brain Iron Accumulation. Seminars in Pediatric Neurology, 2012, 19, 51-56.	2.0	23
97	Non-Random mtDNA Segregation Patterns Indicate a Metastable Heteroplasmic Segregation Unit in m.3243A>G Cybrid Cells. PLoS ONE, 2012, 7, e52080.	2.5	21
98	A randomized placebo-controlled trial of idebenone in Leberâ€™s hereditary optic neuropathy. Brain, 2011, 134, 2677-2686.	7.6	461
99	Mitochondrial aging is accelerated by anti-retroviral therapy through the clonal expansion of mtDNA mutations. Nature Genetics, 2011, 43, 806-810.	21.4	201
100	The implications of mitochondrial DNA copy number regulation during embryogenesis. Mitochondrion, 2011, 11, 686-692.	3.4	68
101	Neurodegeneration with brain iron accumulation. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2011, 100, 161-172.	1.8	33
102	Mitochondrial optic neuropathies â€“ Disease mechanisms and therapeutic strategies. Progress in Retinal and Eye Research, 2011, 30, 81-114.	15.5	514
103	Nuclear factors involved in mitochondrial translation cause a subgroup of combined respiratory chain deficiency. Brain, 2011, 134, 183-195.	7.6	66
104	OPA1 mutations impair mitochondrial function in both pure and complicated dominant optic atrophy. Brain, 2011, 134, e164-e164.	7.6	34
105	OPA1 links human mitochondrial genome maintenance to mtDNA replication and distribution. Genome Research, 2011, 21, 12-20.	5.5	207
106	A founder mutation in Anoctamin 5 is a major cause of limb girdle muscular dystrophy. Brain, 2011, 134, 171-182.	7.6	254
107	Mitochondrial myopathies: developments in treatment. Current Opinion in Neurology, 2010, 23, 459-465.	3.6	38
108	Previous Estimates of Mitochondrial DNA Mutation Level Variance Did Not Account for Sampling Error: Comparing the mtDNA Genetic Bottleneck in Mice and Humans. American Journal of Human Genetics, 2010, 86, 540-550.	6.2	37

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109	Mitochondrial DNA haplogroups and risk of transient ischaemic attack and ischaemic stroke: a genetic association study. Lancet Neurology, The, 2010, 9, 498-503.	10.2	88
110	Dramatic response of facial stereotype/tic to tetrabenazine in the first reported cases of neuroferritinopathy in the United States. Movement Disorders, 2010, 25, 2470-2472.	3.9	26
111	Pronuclear transfer in human embryos to prevent transmission of mitochondrial DNA disease. Nature, 2010, 465, 82-85.	27.8	421
112	OPA1 mutations cause cytochrome c oxidase deficiency due to loss of wild-type mtDNA molecules. Human Molecular Genetics, 2010, 19, 3043-3052.	2.9	95
113	The Prevalence and Natural History of Dominant Optic Atrophy Due to OPA1 Mutations. Ophthalmology, 2010, 117, 1538-1546.e1.	5.2	162
114	Prevalence of genetic muscle disease in Northern England: in-depth analysis of a muscle clinic population. Brain, 2009, 132, 3175-3186.	7.6	414
115	Clinical features and natural history of neuroferritinopathy caused by the 458dupA FTL mutation. Brain, 2009, 132, e109-e109.	7.6	58
116	Secondary mtDNA Defects Do Not Cause Optic Nerve Dysfunction in a Mouse Model of Dominant Optic Atrophy. , 2009, 50, 4561.		20
117	Quality of Life in Patients with Leber Hereditary Optic Neuropathy. , 2009, 50, 3112.		87
118	Locating the stem cell niche and tracing hepatocyte lineages in human liver. Hepatology, 2009, 49, 1655-1663.	7.3	135
119	A Methodological Approach to Tracing Cell Lineage in Human Epithelial Tissues. Stem Cells, 2009, 27, 1410-1420.	3.2	72
120	Subtle neurological and metabolic abnormalities in an Opa1 mouse model of autosomal dominant optic atrophy. Experimental Neurology, 2009, 220, 404-409.	4.1	44
121	Prevalence of mitochondrial DNA disease in adults. Annals of Neurology, 2008, 63, 35-39.	5.3	540
122	Selection against Pathogenic mtDNA Mutations in a Stem Cell Population Leads to the Loss of the 3243Aâ†'G Mutation in Blood. American Journal of Human Genetics, 2008, 82, 333-343.	6.2	112
123	Pathogenic Mitochondrial DNA Mutations Are Common in the General Population. American Journal of Human Genetics, 2008, 83, 254-260.	6.2	534
124	The Distribution of Mitochondrial DNA Heteroplasmy Due to Random Genetic Drift. American Journal of Human Genetics, 2008, 83, 582-593.	6.2	72
125	A reduction of mitochondrial DNA molecules during embryogenesis explains the rapid segregation of genotypes. Nature Genetics, 2008, 40, 249-254.	21.4	438
126	What causes mitochondrial DNA deletions in human cells?. Nature Genetics, 2008, 40, 275-279.	21.4	334

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127	155th ENMC workshop: Polymerase gamma and disorders of mitochondrial DNA synthesis, 21â€“23 September 2007, Naarden, The Netherlands. Neuromuscular Disorders, 2008, 18, 259-267.	0.6	65
128	Mutation of OPA1 causes dominant optic atrophy with external ophthalmoplegia, ataxia, deafness and multiple mitochondrial DNA deletions: a novel disorder of mtDNA maintenance. Brain, 2008, 131, 329-337.	7.6	381
129	Normal Levels of Wild-Type Mitochondrial DNA Maintain Cytochrome c Oxidase Activity for Two Pathogenic Mitochondrial DNA Mutations but Not for m.3243Aâ†'G. American Journal of Human Genetics, 2007, 81, 189-195.	6.2	85
130	Clinical Expression of Leber Hereditary Optic Neuropathy Is Affected by the Mitochondrial DNAâ€"Haplogroup Background. American Journal of Human Genetics, 2007, 81, 228-233.	6.2	331
131	Depletion of mitochondrial DNA in leucocytes harbouring the 3243A->G mtDNA mutation. Journal of Medical Genetics, 2006, 44, 69-74.	3.2	72
132	Mitochondrial DNA Mutations in Individuals Occupationally Exposed to Ionizing Radiation. Radiation Research, 2006, 165, 202-207.	1.5	18
133	The Power to Detect Disease Associations with Mitochondrial DNA Haplogroups. American Journal of Human Genetics, 2006, 78, 713-720.	6.2	100
134	Neuroferritinopathy. Seminars in Pediatric Neurology, 2006, 13, 176-181.	2.0	42
135	Progressive depletion of mtDNA in mitochondrial myopathy. Neurology, 2006, 67, 502-504.	1.1	10
136	Clinical features and natural history of neuroferritinopathy caused by the FTL1 460InsA mutation. Brain, 2006, 130, 110-119.	7.6	178
137	Mitochondrial DNA polymerase-Î³ and human disease. Human Molecular Genetics, 2006, 15, R244-R252.	2.9	192
138	Mitochondrial DNA in Homo Sapiens. , 2006, , 3-15.		2
139	Spectrum of movement disorders in neuroferritinopathy. Movement Disorders, 2005, 20, 95-99.	3.9	45
140	Mitochondrial DNA and survival after sepsis: a prospective study. Lancet, The, 2005, 366, 2118-2121.	13.7	162
141	Minimum prevalence of spinocerebellar ataxia 17 in the north east of England. Journal of the Neurological Sciences, 2005, 239, 105-109.	0.6	35
142	Noninvasive diagnosis of the 3243A>G mitochondrial DNA mutation using urinary epithelial cells. European Journal of Human Genetics, 2004, 12, 778-781.	2.8	107
143	Familial myopathy: New insights into the T14709C mitochondrial tRNA mutation. Annals of Neurology, 2004, 55, 478-484.	5.3	71
144	A novel sporadic mutation in cytochrome c oxidase subunit II as a cause of rhabdomyolysis. Neuromuscular Disorders, 2004, 14, 162-166.	0.6	58

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145	Risk of developing a mitochondrial DNA deletion disorder. Lancet, The, 2004, 364, 592-596.	13.7	201
146	Changes in the human mitochondrial genome after treatment of malignant disease. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2003, 525, 19-27.	1.0	48
147	Late-onset mitochondrial disorder with electromyographic evidence of myotonia. Muscle and Nerve, 2003, 28, 757-759.	2.2	4
148	Genotypes from patients indicate no paternal mitochondrial DNA contribution. Annals of Neurology, 2003, 54, 521-524.	5.3	76
149	A Model of the Nuclear Control of Mitochondrial DNA Replication. Journal of Theoretical Biology, 2003, 221, 565-583.	1.7	48
150	A compositional segmentation of the human mitochondrial genome is related to heterogeneities in the guanine mutation rate. Nucleic Acids Research, 2003, 31, 6043-6052.	14.5	19
151	Mitochondrial DNA mutations in human colonic crypt stem cells. Journal of Clinical Investigation, 2003, 112, 1351-1360.	8.2	454
152	Can mitochondrial DNA mutations cause sperm dysfunction?. Molecular Human Reproduction, 2002, 8, 719-721.	2.8	138
153	Detection and quantification of mitochondrial DNA deletions in individual cells by real-time PCR. Nucleic Acids Research, 2002, 30, 68e-68.	14.5	279
154	Neuroferritinopathy: A Window on the Role of Iron in Neurodegeneration. Blood Cells, Molecules, and Diseases, 2002, 29, 522-531.	1.4	67
155	Accumulation of mitochondrial DNA mutations in ageing, cancer, and mitochondrial disease: is there a common mechanism?. Lancet, The, 2002, 360, 1323-1325.	13.7	203
156	Inheritance of mitochondrial disorders. Mitochondrion, 2002, 2, 149-155.	3.4	23
157	Leber hereditary optic neuropathy: Does heteroplasmy influence the inheritance and expression of the G11778A mitochondrial DNA mutation?. American Journal of Medical Genetics Part A, 2001, 98, 235-243.	2.4	130
158	Mutation in the gene encoding ferritin light polypeptide causes dominant adult-onset basal ganglia disease. Nature Genetics, 2001, 28, 350-354.	21.4	533
159	Mitochondrial DNA mutations in the pathogenesis of human disease. Trends in Molecular Medicine, 2000, 6, 425-432.	2.6	89
160	The inheritance of mitochondrial DNA heteroplasmy: random drift, selection or both?. Trends in Genetics, 2000, 16, 500-505.	6.7	227
161	The spectrum of hearing loss due to mitochondrial DNA defects. Brain, 2000, 123, 82-92.	7.6	132
162	In-vitro genetic modification of mitochondrial function. Human Reproduction, 2000, 15, 79-85.	0.9	23

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163	Analysis of Mitochondrial DNA Mutations: Point Mutations. , 2000, 38, 265-277.		1
164	Pathogenic mitochondrial DNA mutations and human reproduction. Human Fertility, 1999, 2, 133-137.	1.7	3
165	Reanalysis and revision of the Cambridge reference sequence for human mitochondrial DNA. Nature Genetics, 1999, 23, 147-147.	21.4	2,800
166	Relaxed Replication of mtDNA: A Model with Implications for the Expression of Disease. American Journal of Human Genetics, 1999, 64, 1158-1165.	6.2	167
167	Genetic Counseling and Prenatal Diagnosis for mtDNA Disease. American Journal of Human Genetics, 1998, 63, 1908-1910.	6.2	25
168	Selective inhibition of mutant human mitochondrial DNA replication in vitro by peptide nucleic acids. Nature Genetics, 1997, 15, 212-215.	21.4	252
169	Mammalian mitochondrial genetics: heredity, heteroplasmy and disease. Trends in Genetics, 1997, 13, 450-455.	6.7	415
170	Mitochondrial myopathies. , 0, , 363-389.		1
171	Laboratory Investigation of Mitochondrial Diseases. , 0, , 39-51.		1