Gavin Hudson

List of Publications by Year in descending order

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#	Article	IF	CITATIONS
1	Phenotypic spectrum associated with mutations of the mitochondrial polymerase gene. Brain, 2006, 129, 1674-1684.	3.7	397
2	Multi-system neurological disease is common in patients with OPA1 mutations. Brain, 2010, 133, 771-786.	3.7	385
3	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.	3.7	381
4	Characterizing mild cognitive impairment in incident Parkinson disease. Neurology, 2014, 82, 308-316.	1.5	359
5	Whole-genome sequencing of patients with rare diseases in a national health system. Nature, 2020, 583, 96-102.	13.7	338
6	Inherited mitochondrial optic neuropathies. Journal of Medical Genetics, 2008, 46, 145-158.	1.5	336
7	Loss of VPS13C Function in Autosomal-Recessive Parkinsonism Causes Mitochondrial Dysfunction and Increases PINK1/Parkin-Dependent Mitophagy. American Journal of Human Genetics, 2016, 98, 500-513.	2.6	333
8	Clinical Expression of Leber Hereditary Optic Neuropathy Is Affected by the Mitochondrial DNA–Haplogroup Background. American Journal of Human Genetics, 2007, 81, 228-233.	2.6	331
9	Excessive burden of lysosomal storage disorder gene variants in Parkinson's disease. Brain, 2017, 140, 3191-3203.	3.7	323
10	Mitochondrial genetics. British Medical Bulletin, 2013, 106, 135-159.	2.7	275
11	A founder mutation in Anoctamin 5 is a major cause of limb girdle muscular dystrophy. Brain, 2011, 134, 171-182.	3.7	254
12	Efficient mitochondrial biogenesis drives incomplete penetrance in Leber's hereditary optic neuropathy. Brain, 2014, 137, 335-353.	3.7	229
13	Mutant POLG2 Disrupts DNA Polymerase γ Subunits and Causes Progressive External Ophthalmoplegia. American Journal of Human Genetics, 2006, 78, 1026-1034.	2.6	215
14	Dissection of the genetics of Parkinson's disease identifies an additional association 5' of SNCA and multiple associated haplotypes at 17q21. Human Molecular Genetics, 2011, 20, 345-353.	1.4	202
15	Mitochondrial DNA polymerase-γ and human disease. Human Molecular Genetics, 2006, 15, R244-R252.	1.4	192
16	Segregation of mitochondrial DNA heteroplasmy through a developmental genetic bottleneck in human embryos. Nature Cell Biology, 2018, 20, 144-151.	4.6	182
17	Identification of an X-Chromosomal Locus and Haplotype Modulating the Phenotype of a Mitochondrial DNA Disorder. American Journal of Human Genetics, 2005, 77, 1086-1091.	2.6	181
18	Reduced mitochondrial DNA copy number is a biomarker of Parkinson's disease. Neurobiology of Aging, 2016, 38, 216.e7-216.e10.	1.5	178

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19	Germline selection shapes human mitochondrial DNA diversity. Science, 2019, 364, .	6.0	178
20	The Prevalence and Natural History of Dominant Optic Atrophy Due to OPA1 Mutations. Ophthalmology, 2010, 117, 1538-1546.e1.	2.5	162
21	Epigenetics, epidemiology and mitochondrial DNA diseases. International Journal of Epidemiology, 2012, 41, 177-187.	0.9	146
22	Recent Mitochondrial DNA Mutations Increase the Risk of Developing Common Late-Onset Human Diseases. PLoS Genetics, 2014, 10, e1004369.	1.5	131
23	Two-stage association study and meta-analysis of mitochondrial DNA variants in Parkinson disease. Neurology, 2013, 80, 2042-2048.	1.5	129
24	A pathway-based analysis provides additional support for an immune-related genetic susceptibility to Parkinson's disease. Human Molecular Genetics, 2013, 22, 1039-1049.	1.4	122
25	Molecular basis of infantile reversible cytochrome c oxidase deficiency myopathy. Brain, 2009, 132, 3165-3174.	3.7	112
26	Reduced cerebrospinal fluid mitochondrial DNA is a biomarker for earlyâ€stage Parkinson's disease. Annals of Neurology, 2015, 78, 1000-1004.	2.8	106
27	Associations of Mitochondrial and Nuclear Mitochondrial Variants and Genes with Seven Metabolic Traits. American Journal of Human Genetics, 2019, 104, 112-138.	2.6	106

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37	Genetic and pathological links between Parkinson's disease and the lysosomal disorder Sanfilippo syndrome. Movement Disorders, 2012, 27, 312-315.	2.2	56
38	Assessing mitochondrial heteroplasmy using next generation sequencing: A note of caution. Mitochondrion, 2019, 46, 302-306.	1.6	56
39	Circulating cell-free mitochondrial DNA levels in Parkinson's disease are influenced by treatment. Molecular Neurodegeneration, 2020, 15, 10.	4.4	56
40	OPA1 increases the risk of normal but not high tension glaucoma. Journal of Medical Genetics, 2010, 47, 120-125.	1.5	53
41	Mitochondrial DNA sequence characteristics modulate the size of the genetic bottleneck. Human Molecular Genetics, 2016, 25, 1031-1041.	1.4	53
42	Mutations in mitochondrial DNA causing tubulointerstitial kidney disease. PLoS Genetics, 2017, 13, e1006620.	1.5	52
43	Singleâ€cell expression profiling of dopaminergic neurons combined with association analysis identifies pyridoxal kinase as Parkinson's disease gene. Annals of Neurology, 2009, 66, 792-798.	2.8	49
44	Background sequence characteristics influence the occurrence and severity of disease-causing mtDNA mutations. PLoS Genetics, 2017, 13, e1007126.	1.5	49
45	What is influencing the phenotype of the common homozygous polymerase-Î ³ mutation p.Ala467Thr?. Brain, 2012, 135, 3614-3626.	3.7	46
46	Mitochondrial DNA and traumatic brain injury. Annals of Neurology, 2014, 75, 186-195.	2.8	46
47	Mitochondrial DNA changes in pedunculopontine cholinergic neurons in Parkinson disease. Annals of Neurology, 2017, 82, 1016-1021.	2.8	45
48	<scp><i>GBA</i></scp> and <scp><i>APOE</i></scp> Impact Cognitive Decline in Parkinson's Disease: A 10‥ear Populationâ€Based Study. Movement Disorders, 2022, 37, 1016-1027.	2.2	45
49	Novel mutations in the TK2 gene associated with fatal mitochondrial DNA depletion myopathy. Neuromuscular Disorders, 2008, 18, 557-560.	0.3	42
50	POLG1 Mutations Manifesting as Autosomal Recessive Axonal Charcot-Marie-Tooth Disease. Archives of Neurology, 2008, 65, 133-6.	4.9	42
51	A new phenotype of brain iron accumulation with dystonia, optic atrophy, and peripheral neuropathy. Movement Disorders, 2012, 27, 789-793.	2.2	41
52	Late-onset ptosis and myopathy in a patient with a heterozygous insertion in POLG2. Journal of Neurology, 2010, 257, 1517-1523.	1.8	39
53	No evidence of an association between mitochondrial DNA variants and osteoarthritis in 7393 cases and 5122 controls. Annals of the Rheumatic Diseases, 2013, 72, 136-139.	0.5	39
54	Novel POLG1 mutations associated with neuromuscular and liver phenotypes in adults and children. Journal of Medical Genetics, 2008, 46, 209-214.	1.5	38

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55	De Novo Truncating Mutations in WASF1 Cause Intellectual Disability with Seizures. American Journal of Human Genetics, 2018, 103, 144-153.	2.6	36
56	Exome sequencing in dementia with Lewy bodies. Translational Psychiatry, 2016, 6, e728-e728.	2.4	35
57	The genetics of mitochondrial disease: dissecting mitochondrial pathology using multiâ€omic pipelines. Journal of Pathology, 2021, 254, 430-442.	2.1	33
58	No association between common <i>POLG1</i> variants and sporadic idiopathic Parkinson's disease. Movement Disorders, 2009, 24, 1092-1094.	2.2	32
59	Cell-free mitochondrial DNA in progressive multiple sclerosis. Mitochondrion, 2019, 46, 307-312.	1.6	32
60	No evidence of substantia nigra telomere shortening in Parkinson's disease. Neurobiology of Aging, 2011, 32, 2107.e3-2107.e5.	1.5	29
61	Reversion after replacement of mitochondrial DNA. Nature, 2019, 574, E8-E11.	13.7	27
62	A genome-wide association study of mitochondrial DNA copy number in two population-based cohorts. Human Genomics, 2019, 13, 6.	1.4	25
63	Mitochondrial DNA Damage and Brain Aging in Human Immunodeficiency Virus. Clinical Infectious Diseases, 2021, 73, e466-e473.	2.9	23
64	Raised Intraocular Pressure as a Potential Risk Factor for Visual Loss in Leber Hereditary Optic Neuropathy. PLoS ONE, 2013, 8, e63446.	1.1	23
65	X-Inactivation patterns in females harboring mtDNA mutations that cause Leber hereditary optic neuropathy. Molecular Vision, 2007, 13, 2339-43.	1.1	21
66	Heteroplasmic mitochondrial DNA variants in cardiovascular diseases. PLoS Genetics, 2022, 18, e1010068.	1.5	19
67	Does mitochondrial DNA predispose to neuromyelitis optica (Devic's disease)?. Brain, 2008, 131, e93-e93.	3.7	17
68	Leber hereditary optic neuropathy. Expert Opinion on Medical Diagnostics, 2008, 2, 789-799.	1.6	17
69	Human liver stem cells originate from the canals of hering. Hepatology, 2009, 50, 992-993.	3.6	17
70	Maternal germline factors associated with aneuploid pregnancy loss: a systematic review. Human Reproduction Update, 2021, 27, 866-884.	5.2	17
71	Imaging mass cytometry reveals generalised deficiency in OXPHOS complexes in Parkinson's disease. Npj Parkinson's Disease, 2021, 7, 39.	2.5	17
72	The A467T and W748S POLG substitutions are a rare cause of adult-onset ataxia in Europe. Brain, 2006, 130, E69-E69.	3.7	14

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73	Post-mortem ventricular cerebrospinal fluid cell-free-mtDNA in neurodegenerative disease. Scientific Reports, 2020, 10, 15253.	1.6	14
74	Astrocytic Changes in Mitochondrial Oxidative Phosphorylation Protein Levels in Parkinson's Disease. Movement Disorders, 2022, 37, 302-314.	2.2	14
75	Reply: Parsing the differences in affected with LHON: genetic versus environmental triggers of disease conversion. Brain, 2016, 139, e18-e18.	3.7	13
76	Leber Hereditary Optic Neuropathy Presenting in a 75-Year-Old Man. Journal of Neuro-Ophthalmology, 2008, 28, 155.	0.4	12
77	Frailty and mortality are not influenced by mitochondrial DNA haplotypes in the very old. Neurobiology of Aging, 2013, 34, 2889.e1-2889.e4.	1.5	12
78	Variation in complement protein C1q is not a major contributor to cognitive impairment in Parkinson's disease. Neuroscience Letters, 2015, 594, 66-69.	1.0	11
79	Neuropathological and biochemical investigation of Hereditary Ferritinopathy cases with ferritin light chain mutation: Prominent protein aggregation in the absence of major mitochondrial or oxidative stress. Neuropathology and Applied Neurobiology, 2021, 47, 26-42.	1.8	7
80	Genetic variation of CHRNA4 does not modulate attention in Parkinson's disease. Neuroscience Letters, 2010, 479, 123-125.	1.0	6
81	Unique mitochondrial DNA in highly inbred feral cattle. Mitochondrion, 2012, 12, 438-440.	1.6	6
82	Interactions between nuclear and mitochondrial SNPs and Parkinson's disease risk. Mitochondrion, 2022, 63, 85-88.	1.6	6
83	A subcellular cookie cutter for spatial genomics in human tissue. Analytical and Bioanalytical Chemistry, 2022, 414, 5483-5492.	1.9	6
84	Variation in MAPT is not a contributing factor to the incomplete penetrance in LHON. Mitochondrion, 2011, 11, 620-622.	1.6	4
85	Mitochondrial DNA does not contribute to the heritability of non-alcoholic fatty liver disease. Mitochondrion, 2011, 11, 234-235.	1.6	4
86	The Ageing Brain, Mitochondria and Neurodegeneration. , 2016, , 59-80.		4
87	The Human Coronavirus Receptor <scp>ANPEP</scp> (<scp>CD13</scp>) Is Overexpressed in Parkinson's Disease. Movement Disorders, 2020, 35, 2134-2136.	2.2	4
88	Mitochondrial isolation: when size matters. Wellcome Open Research, 2020, 5, 226.	0.9	4
89	Genetic variation in the methylenetetrahydrofolate reductase gene, MTHFR, does not alter the risk of visual failure in Leber's hereditary optic neuropathy. Molecular Vision, 2009, 15, 870-5.	1.1	4
90	Variation in OPA1 does not explain the incomplete penetrance of Leber hereditary optic neuropathy. Molecular Vision, 2010, 16, 2760-4.	1.1	4

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91	Reduced mitochondrial DNA is not a biomarker of depression in Parkinson's disease. Movement Disorders, 2016, 31, 1923-1924.	2.2	3
92	The Isolation and Deep Sequencing of Mitochondrial DNA. Methods in Molecular Biology, 2021, 2277, 433-447.	0.4	3
93	Mitochondrial isolation: when size matters. Wellcome Open Research, 2020, 5, 226.	0.9	3
94	Mitochondrial Haplogroup and the Risk of Acute Kidney Injury Following Cardiac Bypass Surgery. Scientific Reports, 2019, 9, 2279.	1.6	2
95	A critical analysis of the combined usage of protein localization prediction methods: Increasing the number of independent data sets can reduce the accuracy of predicted mitochondrial localization. Mitochondrion, 2011, 11, 444-449.	1.6	1
96	Genetic variations within the OPA1 gene are not associated with neuromyelitis optica. Multiple Sclerosis Journal, 2012, 18, 240-243.	1.4	1
97	Inherited mtDNA variations are not strong risk factors in human prion disease. Neurobiology of Aging, 2015, 36, 2908.e1-2908.e3.	1.5	Ο