Joanna L Elson

List of Publications by Year
in descending order

77 papers	3,653 citations	186265 28 h-index	138484 58 g-index
81 all docs	81 docs citations	81 times ranked	4470 citing authors

#	Article	IF	Citations
1	Investigation of the correlation between mildly deleterious mtDNA Variations and the clinical progression of multiple sclerosis. Multiple Sclerosis and Related Disorders, 2021, 53, 103055.	2.0	3
2	A broad comparative genomics approach to understanding the pathogenicity of Complex I mutations. Scientific Reports, 2021, 11, 19578.	3.3	5
3	Heterologous Inferential Analysis (HIA) and Other Emerging Concepts: In Understanding Mitochondrial Variation In Pathogenesis: There is no More Low-Hanging Fruit. Methods in Molecular Biology, 2021, 2277, 203-245.	0.9	4
4	Single-Cell Approaches for Studying the Role of Mitochondrial DNA in Neurodegenerative Disease. Methods in Molecular Biology, 2021, 2277, 299-329.	0.9	1
5	Substrate utilisation of cultured skeletal muscle cells in patients with CFS. Scientific Reports, 2020, 10, 18232.	3.3	8
6	Evolutionary dissection of mtDNA hg H: a susceptibility factor for hypertrophic cardiomyopathy. Mitochondrial DNA Part A: DNA Mapping, Sequencing, and Analysis, 2020, 31, 238-244.	0.7	3
7	Design and evaluation of bi-functional iron chelators for protection of dopaminergic neurons from toxicants. Archives of Toxicology, 2020, 94, 3105-3123.	4.2	24
8	Novel 1-hydroxypyridin-2-one metal chelators prevent and rescue ubiquitin proteasomal-related neuronal injury in an in vitro model of Parkinsonâ€™s disease. Archives of Toxicology, 2020, 94, 813-831.	4.2	8
9	DREADD Activation of Pedunculopontine Cholinergic Neurons Reverses Motor Deficits and Restores Striatal Dopamine Signaling in Parkinsonian Rats. Neurotherapeutics, 2020, 17, 1120-1141.	4.4	18
10	The effect of myalgic encephalomyelitis/chronic fatigue syndrome (ME/CFS) severity on cellular bioenergetic function. PLoS ONE, 2020, 15, e0231136.	2.5	24
11	Mitochondrial DNA mutations induce mitochondrial biogenesis and increase the tumorigenic potential of Hodgkin and Reedâ€“Sternberg cells. Carcinogenesis, 2020, 41, 1735-1745.	2.8	10
12	A new approach to find biomarkers in chronic fatigue syndrome/myalgic encephalomyelitis (CFS/ME) by single-cell Raman micro-spectroscopy. Analyst, The, 2019, 144, 913-920.	3.5	16
13	Assessing cellular energy dysfunction in CFS/ME using a commercially available laboratory test. Scientific Reports, 2019, 9, 11464.	3.3	9
14	Altered motor, anxiety-related and attentional task performance at baseline associate with multiple gene copies of the vesicular acetylcholine transporter and related protein overexpression in ChAT::Cre+ rats. Brain Structure and Function, 2019, 224, 3095-3116.	2.3	8
15	Implementing a new variant load model to investigate the role of mtDNA in oxidative stress and inflammation in a bi-ethnic cohort: the SABPA study. Mitochondrial DNA Part A: DNA Mapping, Sequencing, and Analysis, 2019, 30, 440-447.	0.7	3
16	The unresolved role of mitochondrial DNA in Parkinson's disease: An overview of published studies, their limitations, and future prospects. Neurochemistry International, 2019, 129, 104495.	3.8	19
17	Panel-Based Nuclear and Mitochondrial Next-Generation Sequencing Outcomes of an Ethnically Diverse Pediatric Patient Cohort with Mitochondrial Disease. Journal of Molecular Diagnostics, 2019, 21, 503-513.	2.8	12
18	The role of mitochondria in ME/CFS: a perspective. Fatigue: Biomedicine, Health and Behavior, 2019, 7, 52-58.	1.9	6

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19	MtDNA population variation in Myalgic encephalomyelitis/Chronic fatigue syndrome in two populations: a study of mildly deleterious variants. Scientific Reports, 2019, 9, 2914.	3.3	21
20	What can a comparative genomics approach tell us about the pathogenicity of mtDNA mutations in human populations?. Evolutionary Applications, 2019, 12, 1912-1930.	3.1	5
21	Mitochondrial complex activity in permeabilised cells of chronic fatigue syndrome patients using two cell types. PeerJ, 2019, 7, e6500.	2.0	24
22	Temporal-Spatial Profiling of Pedunculopontine Galanin-Cholinergic Neurons in the Lactacystin Rat Model of Parkinsonâ€™s Disease. Neurotoxicity Research, 2018, 34, 16-31.	2.7	6
23	Mitochondrial DNA copy number is not associated with fatigue status in Primary SjÃ¶grenâ€™s Syndrome. Fatigue: Biomedicine, Health and Behavior, 2018, 6, 123-131.	1.9	2
24	Haplogroup Context is Less Important in the Penetrance of Mitochondrial DNA Complex I Mutations Compared to mt-tRNA Mutations. Journal of Molecular Evolution, 2018, 86, 395-403.	1.8	10
25	New mtDNA Association Model, MutPred Variant Load, Suggests Individuals With Multiple Mildly Deleterious mtDNA Variants Are More Likely to Suffer From Atherosclerosis. Frontiers in Genetics, 2018, 9, 702.	2.3	12
26	The aetiology of cardiovascular disease: a role for mitochondrial DNA?. Cardiovascular Journal of Africa, 2018, 29, 122-132.	0.4	14
27	MutPred mutational load analysis shows mildly deleterious mitochondrial DNA variants are not more prevalent in Alzheimer's patients, but may be under-represented in healthy older individuals. Mitochondrion, 2017, 34, 141-146.	3.4	18
28	Clinically proven mtDNA mutations are not common in those with chronic fatigue syndrome. BMC Medical Genetics, 2017, 18, 29.	2.1	15
29	Using MutPred derived mtDNA load scores to evaluate mtDNA variation in hypertension and diabetes in a two-population cohort: The SABPA study. Journal of Genetics and Genomics, 2017, 44, 139-149.	3.9	20
30	Mitochondrial DNA changes in pedunculopontine cholinergic neurons in Parkinson disease. Annals of Neurology, 2017, 82, 1016-1021.	5.3	45
31	Cellular bioenergetics is impaired in patients with chronic fatigue syndrome. PLoS ONE, 2017, 12, e0186802.	2.5	106
32	Mitochondrial DNA sequence context in the penetrance of mitochondrial t-RNA mutations: A study across multiple lineages with diagnostic implications. PLoS ONE, 2017, 12, e0187862.	2.5	18
33	Could we offer mitochondrial donation or similar assisted reproductive technology to South African patients with mitochondrial DNA disease?. South African Medical Journal, 2016, 106, 234.	0.6	4
34	Pedunculopontine cell loss and protein aggregation direct microglia activation in parkinsonian rats. Brain Structure and Function, 2016, 221, 2319-2341.	2.3	17
35	Pharmacogenetic stimulation of cholinergic pedunculopontine neurons reverses motor deficits in a rat model of Parkinsonâ€™s disease. Molecular Neurodegeneration, 2015, 10, 47.	10.8	41
36	Deep-brain stimulation associates with improved microvascular integrity in the subthalamic nucleus in Parkinson's disease. Neurobiology of Disease, 2015, 74, 392-405.	4.4	86

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37	Perceived fatigue is highly prevalent and debilitating in patients with mitochondrial disease. Neuromuscular Disorders, 2015, 25, 563-566.	0.6	67
38	Mitochondrial DNA as a Risk Factor for False Positives in Case-Control Association Studies. Journal of Genetics and Genomics, 2015, 42, 169-172.	3.9	30
39	The presence of highly disruptive 16S rRNA mutations in clinical samples indicates a wider role for mutations of the mitochondrial ribosome in human disease. Mitochondrion, 2015, 25, 17-27.	3.4	29
40	An animal model mimicking pedunculopontine nucleus cholinergic degeneration in Parkinsonâ€™s disease. Brain Structure and Function, 2015, 220, 479-500.	2.3	49
41	Heterologous Inferential Analysis (HIA) as a Method to Understand the Role of Mitochondrial rRNA Mutations in Pathogenesis. Methods in Molecular Biology, 2015, 1264, 369-383.	0.9	10
42	Clonal Expansion of Early to Mid-Life Mitochondrial DNA Point Mutations Drives Mitochondrial Dysfunction during Human Ageing. PLoS Genetics, 2014, 10, e1004620.	3.5	115
43	The role of the mitochondrial ribosome in human disease: searching for mutations in 12S mitochondrial rRNA with high disruptive potential. Human Molecular Genetics, 2014, 23, 949-967.	2.9	35
44	Mitochondrial Abnormality Associates with Type-Specific Neuronal Loss and Cell Morphology Changes in the Pedunculopontine Nucleus in Parkinson Disease. American Journal of Pathology, 2013, 183, 1826-1840.	3.8	53
45	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.	3.5	61
46	Characterization of mtDNA variation in a cohort of South African paediatric patients with mitochondrial disease. European Journal of Human Genetics, 2012, 20, 650-656.	2.8	30
47	Raising Doubts about the Pathogenicity of Mitochondrial DNA Mutation m.3308T>C in Left Ventricular Hypertraveculation/Noncompaction. Cardiology, 2012, 122, 113-115.	1.4	6
48	Relationship Between Mitochondria and Î±-Synuclein. Archives of Neurology, 2012, 69, 385.	4.5	43
49	A proposed consensus panel of organisms for determining evolutionary conservation of mt-tRNA point mutations. Mitochondrion, 2012, 12, 533-538.	3.4	28
50	Aggregation, impaired degradation and immunization targeting of amyloid-beta dimers in Alzheimerâ€™s disease: a stochastic modelling approach. Molecular Neurodegeneration, 2012, 7, 32.	10.8	25
51	Toward a mtDNA locus-specific mutation database using the LOVD platform. Human Mutation, 2012, 33, 1352-1358.	2.5	8
52	Common mtDNA Polymorphisms and Neurodegenerative Disorders. , 2012, , 63-78.		0
53	Habitual Physical Activity in Mitochondrial Disease. PLoS ONE, 2011, 6, e22294.	2.5	37
54	Call for participation in the neurogenetics consortium within the Human Variome Project. Neurogenetics, 2011, 12, 169-173.	1.4	5

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55	A comparative analysis approach to determining the pathogenicity of mitochondrial tRNA mutations. Human Mutation, 2011, 32, 1319-1325.	2.5	159
56	Mitochondrial DNA disease: new options for prevention. Human Molecular Genetics, 2011, 20, R168-R174.	2.9	72
57	Mitochondrial tRNA mutations and disease. Wiley Interdisciplinary Reviews RNA, 2010, 1, 304-324.	6.4	145
58	Older mothers are not at risk of having grandchildren with sporadic mtDNA deletions. Genetics in Medicine, 2010, 12, 313-314.	2.4	3
59	Pathogenic mitochondrial tRNA mutations - Which mutations are inherited and why?. Human Mutation, 2009, 30, E984-E992.	2.5	49
60	The low abundance of clonally expanded mitochondrial DNA point mutations in aged substantia nigra neurons. Aging Cell, 2009, 8, 496-498.	6.7	26
61	Purifying selection of mtDNA and its implications for understanding evolution and mitochondrial disease. Nature Reviews Genetics, 2008, 9, 657-662.	16.3	155
62	Nature of Mitochondrial DNA Deletions in Substantia Nigra Neurons. American Journal of Human Genetics, 2008, 82, 228-235.	6.2	123
63	Does mitochondrial DNA predispose to neuromyelitis optica (Devic's disease)?. Brain, 2008, 131, e93-e93.	7.6	17
64	Strong Purifying Selection in Transmission of Mammalian Mitochondrial DNA. PLoS Biology, 2008, 6, e10.	5.6	425
65	Relative Rates of Evolution in the Coding and Control Regions of African mtDNAs. Molecular Biology and Evolution, 2007, 24, 2213-2221.	8.9	33
66	Testing the adaptive selection of human mtDNA haplogroups: an experimental bioenergetics approach. Biochemical Journal, 2007, 404, e3-5.	3.7	10
67	Associating Mitochondrial DNA Variation with Complex Traits. American Journal of Human Genetics, 2007, 80, 378-382.	6.2	16
68	Mitochondrial DNA haplogroups and amyotrophic lateral sclerosis. Neurogenetics, 2007, 8, 65-67.	1.4	13
69	Does the mitochondrial genome play a role in the etiology of Alzheimerâ€™s disease?. Human Genetics, 2006, 119, 241-254.	3.8	102
70	Mitochondrial DNA clonality in the dock: can surveillance swing the case?. Trends in Genetics, 2006, 22, 603-607.	6.7	56
71	mtDNA mutations and common neurodegenerative disorders. Trends in Genetics, 2005, 21, 583-586.	6.7	78
72	Mitochondrial DNA and survival after sepsis: a prospective study. Lancet, The, 2005, 366, 2118-2121.	13.7	162

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73	African Haplogroup L mtDNA Sequences Show Violations of Clock-like Evolution. Molecular Biology and Evolution, 2004, 21, 1843-1854.	8.9	43
74	Assigning pathogenicity to mitochondrial tRNA mutations: when â€˜definitely maybeâ€™ is not good enough. Trends in Genetics, 2004, 20, 591-596.	6.7	159
75	Reduced-Median-Network Analysis of Complete Mitochondrial DNA Coding-Region Sequences for the Major African, Asian, and European Haplogroups. American Journal of Human Genetics, 2002, 70, 1152-1171.	6.2	482
76	The length of cytochrome c oxidase-negative segments in muscle fibres in patients with mtDNA myopathy. Neuromuscular Disorders, 2002, 12, 858-864.	0.6	20
77	Mitochondrial DNA variation in Parkinsonâ€™s disease: Analysis of â€œout-of-placeâ€•population variants as a risk factor. Frontiers in Aging Neuroscience, 0, 14, .	3.4	7