Grainne Gorman

List of Publications by Year
in descending order

110 papers	6,707 citations	81900 39 h-index	69250 77 g-index
112 all docs	112 docs citations	112 times ranked	7851 citing authors

#	Article	IF	Citations
1	Mitochondrial diseases. Nature Reviews Disease Primers, 2016, 2, 16080.	30.5	1,001
2	Prevalence of nuclear and mitochondrial <scp>DNA</scp> mutations related to adult mitochondrial disease. Annals of Neurology, 2015, 77, 753-759.	5.3	706
3	Multi-system neurological disease is common in patients with OPA1 mutations. Brain, 2010, 133, 771-786.	7.6	385
4	100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care â€" Preliminary Report. New England Journal of Medicine, 2021, 385, 1868-1880.	27.0	352
5	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
6	Mitochondrial Diseases: Hope for the Future. Cell, 2020, 181, 168-188.	28.9	243
7	mt <scp>DNA</scp> heteroplasmy level and copy number indicate disease burden in m.3243A>G mitochondrial disease. EMBO Molecular Medicine, 2018, 10, .	6.9	199
8	The Spectrum of Mitochondrial Ultrastructural Defects in Mitochondrial Myopathy. Scientific Reports, 2016, 6, 30610.	3.3	165
9	Mutations in the SPG7 gene cause chronic progressive external ophthalmoplegia through disordered mitochondrial DNA maintenance. Brain, 2014, 137, 1323-1336.	7.6	151
10	Disease progression in patients with single, large-scale mitochondrial DNA deletions. Brain, 2014, 137, 323-334.	7.6	103
11	Topoisomerase 3Î± Is Required for Decatenation and Segregation of Human mtDNA. Molecular Cell, 2018, 69, 9-23.e6.	9.7	102
12	Phenotypic heterogeneity in m.3243A>G mitochondrial disease: The role of nuclear factors. Annals of Clinical and Translational Neurology, 2018, 5, 333-345.	3.7	102
13	Adult-onset cerebellar ataxia due to mutations inCABC1/ADCK3. Journal of Neurology, Neurosurgery and Psychiatry, 2012, 83, 174-178.	1.9	99
14	Mitochondrial disease in adults: recent advances and future promise. Lancet Neurology, The, 2021, 20, 573-584.	10.2	96
15	Cognitive behavioural therapy with optional graded exercise therapy in patients with severe fatigue with myotonic dystrophy type 1: a multicentre, single-blind, randomised trial. Lancet Neurology, The, 2018, 17, 671-680.	10.2	95
16	The phenotypic spectrum of neutral lipid storage myopathy due to mutations in the PNPLA2 gene. Journal of Neurology, 2011, 258, 1987-1997.	3.6	87
17	Mitochondrial Donation â€" How Many Women Could Benefit?. New England Journal of Medicine, 2015, 372, 885-887.	27.0	87
18	The clinical, histochemical, and molecular spectrum of PEO1 (Twinkle)-linked adPEO. Neurology, 2010, 74, 1619-1626.	1.1	84

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19	Retrospective natural history of thymidine kinase 2 deficiency. Journal of Medical Genetics, 2018, 55, 515-521.	3.2	73
20	Genetic determinants of disease severity in the myotonic dystrophy type 1 OPTIMISTIC cohort. Neurology, 2019, 93, e995-e1009.	1.1	71
21	Adults with RRM2B-related mitochondrial disease have distinct clinical and molecular characteristics. Brain, 2012, 135, 3392-3403.	7.6	70
22	Clinical, Genetic, and Radiological Features of Extrapyramidal Movement Disorders in Mitochondrial Disease. JAMA Neurology, 2016, 73, 668.	9.0	69
23	Perceived fatigue is highly prevalent and debilitating in patients with mitochondrial disease. Neuromuscular Disorders, 2015, 25, 563-566.	0.6	67
24	Consensus-based statements for the management of mitochondrial stroke-like episodes. Wellcome Open Research, 2019, 4, 201.	1.8	66
25	Mutations in TOP3A Cause a Bloom Syndrome-like Disorder. American Journal of Human Genetics, 2018, 103, 221-231.	6.2	65
26	ANO5Gene 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, 1111-1118.	2.5	64
27	Epilepsy in adults with mitochondrial disease: A cohort study. Annals of Neurology, 2015, 78, 949-957.	5.3	62
28	RRM2B mutations are frequent in familial PEO with multiple mtDNA deletions. Neurology, 2011, 76, 2032-2034.	1.1	59
29	Novel reproductive technologies to prevent mitochondrial disease. Human Reproduction Update, 2017, 23, 501-519.	10.8	59
30	International Workshop:. Neuromuscular Disorders, 2017, 27, 1126-1137.	0.6	58
31	Solid organ transplantation in primary mitochondrial disease: Proceed with caution. Molecular Genetics and Metabolism, 2016, 118, 178-184.	1.1	55
32	Sudden adult death syndrome in m.3243A>G-related mitochondrial disease: an unrecognized clinical entity in young, asymptomatic adults. European Heart Journal, 2016, 37, 2552-2559.	2.2	53
33	Ultrasensitive deletion detection links mitochondrial DNA replication, disease, and aging. Genome Biology, 2020, 21, 248.	8.8	48
34	MT-ND5 Mutation Exhibits Highly Variable Neurological Manifestations at Low Mutant Load. EBioMedicine, 2018, 30, 86-93.	6.1	47
35	Defining cardiac adaptations and safety of endurance training in patients with m.3243A>G-related mitochondrial disease. International Journal of Cardiology, 2013, 168, 3599-3608.	1.7	43
36	Extensive respiratory chain defects in inhibitory interneurones in patients with mitochondrial disease. Neuropathology and Applied Neurobiology, 2016, 42, 180-193.	3.2	43

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37	Review: Central nervous system involvement in mitochondrial disease. Neuropathology and Applied Neurobiology, 2017, 43, 102-118.	3.2	42
38	Pathological mechanisms underlying single largeâ€scale mitochondrial <scp>DNA</scp> deletions. Annals of Neurology, 2018, 83, 115-130.	5.3	42
39	Safety of drug use in patients with a primary mitochondrial disease: An international Delphiâ€based consensus. Journal of Inherited Metabolic Disease, 2020, 43, 800-818.	3.6	42
40	Clonal Expansion of Secondary Mitochondrial DNA Deletions Associated With Spinocerebellar Ataxia Type 28. JAMA Neurology, 2015, 72, 106.	9.0	41
41	The urinary proteome and metabonome differ from normal in adults with mitochondrial disease. Kidney International, 2015, 87, 610-622.	5.2	41
42	Nutritional interventions in primary mitochondrial disorders: Developing an evidence base. Molecular Genetics and Metabolism, 2016, 119, 187-206.	1.1	41
43	Clinical Features, Molecular Heterogeneity, and Prognostic Implications in YARS2-Related Mitochondrial Myopathy. JAMA Neurology, 2017, 74, 686.	9.0	41
44	Pseudoâ€obstruction, stroke, and mitochondrial dysfunction: A lethal combination. Annals of Neurology, 2016, 80, 686-692.	5.3	40
45	Habitual Physical Activity in Mitochondrial Disease. PLoS ONE, 2011, 6, e22294.	2.5	37
46	Vertigo and vestibular abnormalities in spinocerebellar ataxia type 6. Journal of Neurology, 2009, 256, 78-82.	3.6	34
47	Cardiomyopathy is common in patients with the mitochondrial DNA m.3243A>G mutation and correlates with mutation load. Neuromuscular Disorders, 2012, 22, 592-596.	0.6	34
48	How Can We Treat Mitochondrial Encephalomyopathies? Approaches to Therapy. Neurotherapeutics, 2008, 5, 558-568.	4.4	33
49	Mitochondrial DNA deletions in muscle satellite cells: implications for therapies. Human Molecular Genetics, 2013, 22, 4739-4747.	2.9	33
50	Pathogenic variants in MTâ€ATP6: A United Kingdomâ€“based mitochondrial disease cohort study. Annals of Neurology, 2019, 86, 310-315.	5.3	33
51	Concentric hypertrophic remodelling and subendocardial dysfunction in mitochondrial DNA point mutation carriersâ€. European Heart Journal Cardiovascular Imaging, 2013, 14, 650-658.	1.2	30
52	Mitochondrial donation: from test tube to clinic. Lancet, The, 2018, 392, 1191-1192.	13.7	30
53	Measuring Habitual Physical Activity inÂNeuromuscular Disorders: A Systematic Review. Journal of Neuromuscular Diseases, 2017, 4, 25-52.	2.6	28
54	Extraocular Muscle Atrophy and Central Nervous System Involvement in Chronic Progressive External Ophthalmoplegia. PLoS ONE, 2013, 8, e75048.	2.5	27

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55	Distal weakness with respiratory insufficiency caused by the m.8344A>G â€œMERRFâ€•mutation. Neuromuscular Disorders, 2014, 24, 533-536.	0.6	26
56	Scientific and Ethical Issues in Mitochondrial Donation. New Bioethics, 2018, 24, 57-73.	1.1	25
57	Mitochondrial Donation â€” Which Women Could Benefit?. New England Journal of Medicine, 2019, 380, 1971-1972.	27.0	25
58	Forecasting stroke-like episodes and outcomes in mitochondrial disease. Brain, 2022, 145, 542-554.	7.6	25
59	Pathogenic mtDNA mutations causing mitochondrial myopathy: The need for muscle biopsy. Neurology: Genetics, 2016, 2, e82.	1.9	24
60	Late-onset respiratory failure due to TK2 mutations causing multiple mtDNA deletions. Neurology, 2013, 81, 2051-2053.	1.1	23
61	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
62	Novel MTND1 mutations cause isolated exercise intolerance, complex I deficiency and increased assembly factor expression. Clinical Science, 2015, 128, 895-904.	4.3	21
63	Disease burden of myotonic dystrophy type 1. Journal of Neurology, 2019, 266, 998-1006.	3.6	21
64	POLRMT mutations impair mitochondrial transcription causing neurological disease. Nature Communications, 2021, 12, 1135.	12.8	21
65	Adult-onset Mendelian PEO Associated with Mitochondrial Disease. Journal of Neuromuscular Diseases, 2014, 1, 119-133.	2.6	19
66	Pathophysiology of exercise intolerance in chronic diseases: the role of diminished cardiac performance in mitochondrial and heart failure patients. Open Heart, 2017, 4, e000632.	2.3	19
67	Instability of the mitochondrial alanyl-tRNA synthetase underlies fatal infantile-onset cardiomyopathy. Human Molecular Genetics, 2019, 28, 258-268.	2.9	19
68	Height as a Clinical Biomarker of Disease Burden in Adult Mitochondrial Disease. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 2057-2066.	3.6	19
69	Decreased male reproductive success in association with mitochondrial dysfunction. European Journal of Human Genetics, 2017, 25, 1162-1164.	2.8	18
70	Leigh syndrome caused by mutations in <scp>MTFMT</scp> is associated with a better prognosis. Annals of Clinical and Translational Neurology, 2019, 6, 515-524.	3.7	17
71	Systematic review of cognitive deficits in adult mitochondrial disease. European Journal of Neurology, 2020, 27, 3-17.	3.3	17
72	Natural History of Leigh Syndrome: A Study of Disease Burden and Progression. Annals of Neurology, 2022, 91, 117-130.	5.3	17

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73	Both mitochondrial DNA and mitonuclear gene mutations cause hearing loss through cochlear dysfunction. Brain, 2016, 139, e33-e33.	7.6	15
74	Analyzing walking speeds with ankle and wrist worn accelerometers in a cohort with myotonic dystrophy. Disability and Rehabilitation, 2019, 41, 2972-2978.	1.8	13
75	Initial development and validation of a mitochondrial disease quality of life scale. Neuromuscular Disorders, 2013, 23, 324-329.	0.6	11
76	Orthostatic intolerance is common in chronic disease â€" A clinical cohort study. International Journal of Cardiology, 2014, 174, 861-863.	1.7	11
77	Discrete gait characteristics are associated with m.3243A>G and m.8344A>G variants of mitochondrial disease and its pathological consequences. Journal of Neurology, 2014, 261, 73-82.	3.6	11
78	Lewy body pathology is more prevalent in older individuals with mitochondrial disease than controls. Acta Neuropathologica, 2020, 139, 219-221.	7.7	11
79	Skeletal muscle mitochondrial oxidative phosphorylation function in idiopathic pulmonary arterial hypertension: in vivo and in vitro study. Pulmonary Circulation, 2018, 8, 1-5.	1.7	10
80	Cognitive deficits in adult m.3243A>Gâ€•and m.8344A>Gâ€•related mitochondrial disease: importance of correcting for baseline intellectual ability. Annals of Clinical and Translational Neurology, 2019, 6, 826-836.	3.7	10
81	Neuromuscular Junction Abnormalities in Mitochondrial Disease. Neurology: Clinical Practice, 2021, 11, 97-104.	1.6	10
82	Mitochondrial DNA abnormalities in ophthalmological disease. Saudi Journal of Ophthalmology, 2011, 25, 395-404.	0.3	9
83	Adult-onset Mendelian PEO Associated with Mitochondrial Disease. Journal of Neuromuscular Diseases, 2014, 1, 119-133.	2.6	9
84	Rapid identification of human muscle disease with fibre optic Raman spectroscopy. Analyst, The, 2022, 147, 2533-2540.	3.5	9
85	Preliminary Evaluation of Clinician Rated Outcome Measures in Mitochondrial Disease. Journal of Neuromuscular Diseases, 2015, 2, 151-155.	2.6	8
86	Identification of a novel heterozygous guanosine monophosphate reductase (GMPR) variant in a patient with a lateâ€•onset disorder of mitochondrial DNA maintenance. Clinical Genetics, 2020, 97, 276-286.	2.0	7
87	Activities of daily living in myotonic dystrophy type 1. Acta Neurologica Scandinavica, 2020, 141, 380-387.	2.1	7
88	COVID-19â€"Related Outcomes in Primary Mitochondrial Diseases. Neurology, 2022, 98, 576-582.	1.1	7
89	Diagnostic investigations of patients with chronic progressive external ophthalmoplegia. British Journal of Ophthalmology, 2012, 96, 1536.2-1536.	3.9	6
90	miR-223-3p and miR-24-3p as novel serum-based biomarkers for myotonic dystrophy type 1. Molecular Therapy - Methods and Clinical Development, 2021, 23, 169-183.	4.1	6

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91	RRM1 variants cause a mitochondrial DNA maintenance disorder via impaired de novo nucleotide synthesis. Journal of Clinical Investigation, 2022, 132, .	8.2	6
92	The diagnosis of posterior reversible encephalopathy syndrome. Lancet Neurology, The, 2015, 14, 1073.	10.2	5
93	Sideroblastic anemia with myopathy secondary to novel, pathogenic missense variants in the YARS2 gene. Haematologica, 2018, 103, e564-e566.	3.5	5
94	The application of Raman spectroscopy to the diagnosis of mitochondrial muscle disease: A preliminary comparison between fibre optic probe and microscope formats. Journal of Raman Spectroscopy, 2022, 53, 172-181.	2.5	5
95	Multifocal demyelinating motor neuropathy and hamartoma syndrome associated with a de novo PTEN mutation. Neurology, 2018, 90, e1842-e1848.	1.1	4
96	A study protocol for quantifying patient preferences in neuromuscular disorders: a case study of the IMI PREFER Project. Wellcome Open Research, 2020, 5, 253.	1.8	4
97	Older mothers are not at risk of having grandchildren with sporadic mtDNA deletions. Genetics in Medicine, 2010, 12, 313-314.	2.4	3
98	RRM2B-Related Mitochondrial Disease. , 2013, , 171-182.		3
99	Adultâ€•onset myoclonus ataxia associated with the mitochondrial m.8993<scp>T</scp>><scp>C</scp> â€œ<scp>NARP</scp>â€•mutation. Movement Disorders, 2015, 30, 1432-1433.	3.9	3
100	Change over time in ability to perform activities of daily living in myotonic dystrophy type 1. Journal of Neurology, 2020, 267, 3235-3242.	3.6	3
101	Risk of cardiac manifestations in adult mitochondrial disease caused by nuclear genetic defects. Open Heart, 2021, 8, e001510.	2.3	3
102	Pathogenic SLC25A26 variants impair SAH transport activity causing mitochondrial disease. Human Molecular Genetics, 2022, 31, 2049-2062.	2.9	3
103	Childhood presentation of â€œadultâ€•polyglucosan body disease. Annals of Neurology, 2013, 73, 317-318.	5.3	2
104	Comment on â€œA severe linezolidâ€•induced rhabdomyolysis and lactic acidosis in Leigh syndromeâ€• Journal of Inherited Metabolic Disease, 2021, 44, 6-7.	3.6	2
105	Opening One's Eyes to Mosaicism in Progressive External Ophthalmoplegia. Neurology: Genetics, 2017, 3, e202.	1.9	1
106	Circulating small RNA signatures differentiate accurately the subtypes of muscular dystrophies: small-RNA next-generation sequencing analytics and functional insights. RNA Biology, 2022, 19, 507-518.	3.1	1
107	Clinical Reasoning: An unusual case of papilledema after orthotopic liver transplantation. Neurology, 2009, 73, e25-9.	1.1	0
108	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.	1.9	0

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109	Reply. Annals of Neurology, 2016, 80, 314-314.	5.3	0
110	Generating hand dysaesthesiae: the "GHD phenomenon" - straight to the diagnosis. BMJ Case Reports, 2009, 2009, bcr0220091544-bcr0220091544.	0.5	0