Carolyn M Sue

List of Publications by Year
in descending order

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113 papers	6,555 citations	61984 43 h-index	69250 77 g-index
115 all docs	115 docs citations	115 times ranked	11381 citing authors

#	Article	IF	Citations
1	The impact of device-assisted therapies on the gut microbiome in Parkinsonâ€™s disease. Journal of Neurology, 2022, 269, 780-795.	3.6	19
2	Patient care standards for primary mitochondrial disease in Australia: an Australian adaptation of the Mitochondrial Medicine Society recommendations. Internal Medicine Journal, 2022, 52, 110-120.	0.8	3
3	Mitochondrial donation: is Australia ready?. Medical Journal of Australia, 2022, 216, 118-121.	1.7	3
4	Standardized practices for RNA diagnostics using clinically accessible specimens reclassifies 75% of putative splicing variants. Genetics in Medicine, 2022, 24, 130-145.	2.4	45
5	Strong Predictive Algorithm of Pathogenesis-Based Biomarkers Improves Parkinsonâ€™s Disease Diagnosis. Molecular Neurobiology, 2022, 59, 1476-1485.	4.0	3
6	Decompensation of cardiorespiratory function and emergence of anemia during pregnancy in a case of mitochondrial myopathy, lactic acidosis, and sideroblastic anemia 2 with compound heterozygous <scp>YARS2</scp> pathogenic variants. American Journal of Medical Genetics, Part A, 2022, 188, 2226-2230.	1.2	4
7	Nutritional Intake and Gut Microbiome Composition Predict Parkinsonâ€™s Disease. Frontiers in Aging Neuroscience, 2022, 14, .	3.4	10
8	The Gut Microbiome in Parkinsonâ€™s Disease: A Longitudinal Study of the Impacts on Disease Progression and the Use of Device-Assisted Therapies. Frontiers in Aging Neuroscience, 2022, 14, .	3.4	15
9	Use of Whole-Genome Sequencing for Mitochondrial Disease Diagnosis. Neurology, 2022, 99, .	1.1	33
10	Viewpoint on Milestones for Fellowship Training in Movement Disorders. Movement Disorders, 2022, 37, 1605-1609.	3.9	2
11	Increased Added Sugar Consumption Is Common in Parkinson's Disease. Frontiers in Nutrition, 2021, 8, 628845.	3.7	23
12	Dystonia Responsive to Dopamine: POLG Mutations Should Be Considered If Sensory Neuropathy Is Present. Journal of Movement Disorders, 2021, 14, 157-160.	1.3	6
13	Mitochondrial disease in adults: recent advances and future promise. Lancet Neurology, The, 2021, 20, 573-584.	10.2	96
14	PARK Genes Link Mitochondrial Dysfunction and Alpha-Synuclein Pathology in Sporadic Parkinsonâ€™s Disease. Frontiers in Cell and Developmental Biology, 2021, 9, 612476.	3.7	32
15	037â€...The gut microbiome in Parkinsonâ€™s disease: longitudinal insights into disease progression and the use of device-assisted therapies. , 2021, , .		0
16	Single cell morphology distinguishes genotype and drug effect in Hereditary Spastic Paraplegia. Scientific Reports, 2021, 11, 16635.	3.3	10
17	091â€...The impact of device-assisted therapy initiation on the gut microbiome in Parkinsonâ€™s disease. , 2021, , .		0
18	015â€...Gut microbiota and nutritional profiles of Parkinsonâ€™s disease patients. , 2021, , .		0

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19	Health-Related Quality of Life for Parkinsonâ€™s Disease Patients and Their Caregivers. Journal of Movement Disorders, 2021, 14, 42-52.	1.3	22
20	Cognitive Influences in Parkinson's Disease Patients and Their Caregivers: Perspectives From an Australian Cohort. Frontiers in Neurology, 2021, 12, 673816.	2.4	3
21	LRRK2 mutations impair depolarization-induced mitophagy through inhibition of mitochondrial accumulation of RAB10. Autophagy, 2020, 16, 203-222.	9.1	124
22	Parkinsonâ€™s disease and the gastrointestinal microbiome. Journal of Neurology, 2020, 267, 2507-2523.	3.6	119
23	Depression in Parkinson's disease: Perspectives from an Australian cohort. Journal of Affective Disorders, 2020, 277, 1038-1044.	4.1	26
24	Mitochondrial Function in Hereditary Spastic Paraplegia: Deficits in SPG7 but Not SPAST Patient-Derived Stem Cells. Frontiers in Neuroscience, 2020, 14, 820.	2.8	17
25	Longâ€Term Followâ€Up and Evolution of ADCY5 â€” From a Ballistic to Dystonic Phenotype. Movement Disorders Clinical Practice, 2020, 7, 985-986.	1.5	4
26	Lossâ€ofâ€Function Variants in <scp>HOPS</scp> Complex Genes <scp>VPS16</scp> and <scp>VPS41</scp> Cause Early Onset Dystonia Associated with Lysosomal Abnormalities. Annals of Neurology, 2020, 88, 867-877.	5.3	70
27	The diagnostic utility of genome sequencing in a pediatric cohort with suspected mitochondrial disease. Genetics in Medicine, 2020, 22, 1254-1261.	2.4	59
28	Serum FGF-21, GDF-15, and blood mtDNA copy number are not biomarkers of Parkinson disease. Neurology: Clinical Practice, 2020, 10, 40-46.	1.6	23
29	Oxidative Stress-Induced Axon Fragmentation Is a Consequence of Reduced Axonal Transport in Hereditary Spastic Paraplegia SPAST Patient Neurons. Frontiers in Neuroscience, 2020, 14, 401.	2.8	23
30	Antibody-Free Targeted Proteomics Assay for Absolute Measurement of Î±-Tubulin Acetylation. Analytical Chemistry, 2020, 92, 11204-11212.	6.5	2
31	Gastrointestinal dysfunction in Parkinsonâ€™s disease. Journal of Neurology, 2020, 267, 1377-1388.	3.6	48
32	Rehabilitation for ataxia study: protocol for a randomised controlled trial of an outpatient and supported home-based physiotherapy programme for people with hereditary cerebellar ataxia. BMJ Open, 2020, 10, e040230.	1.9	14
33	High Degree of Genetic Heterogeneity for Hereditary Cerebellar Ataxias in Australia. Cerebellum, 2019, 18, 137-146.	2.5	21
34	Motor Evoked Potentials in Hereditary Spastic Paraplegiaâ€”A Systematic Review. Frontiers in Neurology, 2019, 10, 967.	2.4	12
35	Whole genome sequencing for the genetic diagnosis of heterogenous dystonia phenotypes. Parkinsonism and Related Disorders, 2019, 69, 111-118.	2.2	44
36	The gut microbiota: A novel therapeutic target in Parkinson's disease?. Parkinsonism and Related Disorders, 2019, 66, 265-266.	2.2	30

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37	Diagnosis of â€˜possibleâ€™ mitochondrial disease: an existential crisis. Journal of Medical Genetics, 2019, 56, 123-130.	3.2	42
38	A Novel Homozygous Mutation in the FUCA1 Gene Highlighting Fucosidosis as a Cause of Dystonia: Case Report and Literature Review. Neuropediatrics, 2019, 50, 248-252.	0.6	10
39	Increased Diagnostic Yield of Spastic Paraplegia with or Without Cerebellar Ataxia Through Whole-Genome Sequencing. Cerebellum, 2019, 18, 781-790.	2.5	28
40	Genetic mimics of cerebral palsy. Movement Disorders, 2019, 34, 625-636.	3.9	76
41	Hereditary sensory and autonomic neuropathy type IC accompanied by upper motor neuron abnormalities and type II juxtafoveal retinal telangiectasias. Journal of the Peripheral Nervous System, 2019, 24, 224-229.	3.1	5
42	125â€…Characterising sleep and fatigue in patients with primary mitochondrial disease. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, A40.3-A41.	1.9	0
43	024â€…Resistance exercises with blood flow restriction in patients with sporadic inclusion body myositis. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, A9.1-A9.	1.9	1
44	046â€…A critical review of biomarkers for hereditary spastic paraplegia. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, A15.3-A16.	1.9	0
45	New insights into the complex role of mitochondria in Parkinsonâ€™s disease. Progress in Neurobiology, 2019, 177, 73-93.	5.7	268
46	How Do I Manage Patients With the Levodopa/Carbidopa Intestinal Gel?. Movement Disorders Clinical Practice, 2019, 6, 181-181.	1.5	1
47	Mitochondrial Dysfunction in Parkinsonâ€™s Disease: New Mechanistic Insights and Therapeutic Perspectives. Current Neurology and Neuroscience Reports, 2018, 18, 21.	4.2	401
48	Single Heterozygous ATP13A2 Mutations Cause Cellular Dysfunction Associated with Parkinson's Disease. Movement Disorders, 2018, 33, 852-854.	3.9	4
49	Movement disorders in mitochondrial disease. Journal of Neurology, 2018, 265, 1230-1240.	3.6	41
50	Levodopaâ€carbidopa intestinal gel: â€˜dismantling the road blocks of a journeyâ€™. Internal Medicine Journal, 2018, 48, 472-474.	0.8	5
51	Maximizing benefits of the levodopa/carbidopa intestinal gel: Systematic considerations, challenging convention and individualizing approaches. Basal Ganglia, 2018, 14, 58-60.	0.3	1
52	Motor protein binding and mitochondrial transport are altered by pathogenic TUBB4A variants. Human Mutation, 2018, 39, 1901-1915.	2.5	17
53	Patient-Derived Stem Cell Models in SPAST HSP: Disease Modelling and Drug Discovery. Brain Sciences, 2018, 8, 142.	2.3	12
54	Expanding the spectrum of PEX16 mutations and novel insights into disease mechanisms. Molecular Genetics and Metabolism Reports, 2018, 16, 46-51.	1.1	21

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55	Mitochondrial diseases. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2018, 147, 125-141.	1.8	30
56	TIA1 variant drives myodegeneration in multisystem proteinopathy with SQSTM1 mutations. Journal of Clinical Investigation, 2018, 128, 1164-1177.	8.2	75
57	Nix restores mitophagy and mitochondrial function to protect against PINK1/Parkin-related Parkinsonâ€™s disease. Scientific Reports, 2017, 7, 44373.	3.3	152
58	Practical approaches to commencing deviceâ€assisted therapies for Parkinson disease in Australia. Internal Medicine Journal, 2017, 47, 1107-1113.	0.8	14
59	A SLC39A8 variant causes manganese deficiency, and glycosylation and mitochondrial disorders. Journal of Inherited Metabolic Disease, 2017, 40, 261-269.	3.6	101
60	Patient care standards for primary mitochondrial disease: a consensus statement from the Mitochondrial Medicine Society. Genetics in Medicine, 2017, 19, 1380-1397.	2.4	173
61	Response to Newman et al.. Genetics in Medicine, 2017, 19, 1380-1380.	2.4	3
62	EPG5-Related Vici Syndrome: A Primary Defect of Autophagic Regulation with an Emerging Phenotype Overlapping with Mitochondrial Disorders. JIMD Reports, 2017, 42, 19-29.	1.5	7
63	Hereditary Parkinsonism-Associated Genetic Variations in PARK9 Locus Lead to Functional Impairment of ATPase Type 13A2. Current Protein and Peptide Science, 2017, 18, 725-732.	1.4	8
64	Role of microRNAs in the Regulation of Î±-Synuclein Expression: A Systematic Review. Frontiers in Molecular Neuroscience, 2016, 9, 128.	2.9	38
65	<scp>N</scp>omenclature of genetic movement disorders: <scp>R</scp>ecommendations of the international <scp>P</scp>arkinson and movement disorder society task force. Movement Disorders, 2016, 31, 436-457.	3.9	228
66	Loss of ATP13A2 impairs glycolytic function in Kufor-Rakeb syndrome patient-derived cell models. Parkinsonism and Related Disorders, 2016, 27, 67-73.	2.2	20
67	A comparison of current serum biomarkers as diagnostic indicators of mitochondrial diseases. Neurology, 2016, 86, 2010-2015.	1.1	89
68	Defining the genetic basis of early onset hereditary spastic paraplegia using whole genome sequencing. Neurogenetics, 2016, 17, 265-270.	1.4	32
69	Mechanism of impaired microtubule-dependent peroxisome trafficking and oxidative stress in SPAST-mutated cells from patients with Hereditary Spastic Paraplegia. Scientific Reports, 2016, 6, 27004.	3.3	49
70	Reply letter to Jinnah â€œLocus pocusâ€•and Albanese â€œComplex dystonia is not a category in the new 2013 consensus classificationâ€• Necessary evolution, no magic!. Movement Disorders, 2016, 31, 1760-1762.	3.9	1
71	Activation of Â-Glucocerebrosidase Reduces Pathological Â-Synuclein and Restores Lysosomal Function in Parkinson's Patient Midbrain Neurons. Journal of Neuroscience, 2016, 36, 7693-7706.	3.6	220
72	Thioredoxin interacting protein (TXNIP) regulates tubular autophagy and mitophagy in diabetic nephropathy through the mTOR signaling pathway. Scientific Reports, 2016, 6, 29196.	3.3	106

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73	Functional hyperspectral imaging captures subtle details of cell metabolism in olfactory neurosphere cells, disease-specific models of neurodegenerative disorders. Biochimica Et Biophysica Acta - Molecular Cell Research, 2016, 1863, 56-63.	4.1	48
74	Mutations in HSPB8 causing a new phenotype of distal myopathy and motor neuropathy. Neurology, 2016, 86, 391-398.	1.1	107
75	Olfactory impairment in older adults is associated with poorer diet quality over 5Âyears. European Journal of Nutrition, 2016, 55, 1081-1087.	3.9	51
76	Dietary intakes of fats, fish and nuts and olfactory impairment in older adults. British Journal of Nutrition, 2015, 114, 240-247.	2.3	15
77	An Update on the Hereditary Spastic Paraplegias: New Genes and New Disease Models. Movement Disorders Clinical Practice, 2015, 2, 213-223.	1.5	25
78	Mutations in TUBB4A and spastic paraplegia. Movement Disorders, 2015, 30, 1857-1858.	3.9	4
79	Systematic review of cardiac electrical disease in Kearnsâ€“Sayre syndrome and mitochondrial cytopathy. International Journal of Cardiology, 2015, 181, 303-310.	1.7	81
80	Mutation in mitochondrial ribosomal protein S7 (MRPS7) causes congenital sensorineural deafness, progressive hepatic and renal failure and lactic acidemia. Human Molecular Genetics, 2015, 24, 2297-2307.	2.9	64
81	A novel quantitative assay of mitophagy: Combining high content fluorescence microscopy and mitochondrial DNA load to quantify mitophagy and identify novel pharmacological tools against pathogenic heteroplasmic mtDNA. Pharmacological Research, 2015, 100, 24-35.	7.1	47
82	The role of ATP13A2 in Parkinson's disease: Clinical phenotypes and molecular mechanisms. Movement Disorders, 2015, 30, 770-779.	3.9	144
83	Expanding the phenotype of GMPPB mutations. Brain, 2015, 138, 836-844.	7.6	54
84	Allogeneic haematopoietic stem cell transplantation for mitochondrial neurogastrointestinal encephalomyopathy. Brain, 2015, 138, 2847-2858.	7.6	128
85	SQSTM1 splice site mutation in distal myopathy with rimmed vacuoles. Neurology, 2015, 85, 665-674.	1.1	74
86	Use of Whole-Exome Sequencing for Diagnosis of Limb-Girdle Muscular Dystrophy. JAMA Neurology, 2015, 72, 1424.	9.0	164
87	LARS2 Variants Associated with Hydrops, Lactic Acidosis, Sideroblastic Anemia, and Multisystem Failure. JIMD Reports, 2015, 28, 49-57.	1.5	48
88	Parkin western blotting is useful for identification of patients with Parkin-related Parkinson's disease: FigureÂ1. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 1436-1437.	1.9	1
89	Parkinson's disease-associated human ATP13A2 (PARK9) deficiency causes zinc dyshomeostasis and mitochondrial dysfunction. Human Molecular Genetics, 2014, 23, 2802-2815.	2.9	136
90	Low dose tubulin-binding drugs rescue peroxisome trafficking deficit in patient-derived stem cells in Hereditary Spastic Paraplegia. Biology Open, 2014, 3, 494-502.	1.2	47

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91	Mutations inGNAL. JAMA Neurology, 2014, 71, 490.	9.0	70
92	The broadening spectrum of mitochondrial disease: Shifts in the diagnostic paradigm. Biochimica Et Biophysica Acta - General Subjects, 2014, 1840, 1360-1367.	2.4	48
93	Parkinson's disease-linked human PARK9/ATP13A2 maintains zinc homeostasis and promotes Î±-Synuclein externalization via exosomes. Human Molecular Genetics, 2014, 23, 2816-2833.	2.9	205
94	The deubiquitinase USP15 antagonizes Parkin-mediated mitochondrial ubiquitination and mitophagy. Human Molecular Genetics, 2014, 23, 5227-5242.	2.9	264
95	Targeted next generation sequencing in SPAST-negative hereditary spastic paraplegia. Journal of Neurology, 2013, 260, 2516-2522.	3.6	49
96	A patient-derived stem cell model of hereditary spastic paraplegia with SPAST mutations. DMM Disease Models and Mechanisms, 2013, 6, 489-502.	2.4	55
97	Fibroblast growth factor 21 is a sensitive biomarker of mitochondrial disease. Neurology, 2013, 81, 1819-1826.	1.1	85
98	Frequency of the D620N Mutation in VPS35 in Parkinson Disease. Archives of Neurology, 2012, 69, 1360.	4.5	76
99	ATP13A2 mutations impair mitochondrial function in fibroblasts from patients with Kufor-Rakeb syndrome. Neurobiology of Aging, 2012, 33, 1843.e1-1843.e7.	3.1	106
100	Two Faces of the Same Coin: Benign Familial Infantile Seizures and Paroxysmal Kinesigenic Dyskinesia Caused by <emph type="ital">PRRT2</emph> Mutations. Archives of Neurology, 2012, 69, 668.	4.5	28
101	The phenotypic spectrum of dystonia in Mohrâ€“Tranebjaerg syndrome. Movement Disorders, 2012, 27, 1034-1040.	3.9	22
102	Pathogenic effects of novel mutations in the Pâ€type ATPase ATP13A2 (PARK9) causing Kuforâ€Rakeb syndrome, a form of earlyâ€onset parkinsonism. Human Mutation, 2011, 32, 956-964.	2.5	105
103	The Genetics of Mitochondrial Disease. Seminars in Neurology, 2011, 31, 519-530.	1.4	32
104	Mutant Parkin Impairs Mitochondrial Function and Morphology in Human Fibroblasts. PLoS ONE, 2010, 5, e12962.	2.5	140
105	Mitochondrial disease: recognising more than just the tip of the iceberg. Medical Journal of Australia, 2010, 193, 195-196.	1.7	9
106	Disease-specific, neurosphere-derived cells as models for brain disorders. DMM Disease Models and Mechanisms, 2010, 3, 785-798.	2.4	175
107	Mitochondrial DNA haplogroups J and K are not protective for Parkinson's disease in the Australian community. Movement Disorders, 2009, 24, 290-292.	3.9	23
108	Prevalence of Mitochondrial 1555Aâ†'G Mutation in Adults of European Descent. New England Journal of Medicine, 2009, 360, 642-644.	27.0	115

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109	Mitochondrial DNA disease prevalence: Still underrecognized?. Annals of Neurology, 2008, 64, 471-471.	5.3	7
110	Population prevalence of the MELAS A3243G mutation. Mitochondrion, 2007, 7, 230-233.	3.4	248
111	Aerobic exercise and muscle metabolism in patients with mitochondrial myopathy. Muscle and Nerve, 2006, 33, 524-531.	2.2	62
112	Identical Mitochondrial DNA Deletion in a Woman with Ocular Myopathy and in Her Son with Pearson Syndrome. American Journal of Human Genetics, 2002, 71, 679-683.	6.2	76
113	Pigmentary retinopathy associated with the mitochondrial DNA 3243 point mutation. Neurology, 1997, 49, 1013-1017.	1.1	56