

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	Mitochondrial Dysfunction in Parkinson's Disease: New Mechanistic Insights and Therapeutic Perspectives. <i>Current Neurology and Neuroscience Reports</i> , 2018, 18, 21.	4.2	401
2	New insights into the complex role of mitochondria in Parkinson's disease. <i>Progress in Neurobiology</i> , 2019, 177, 73-93.	5.7	268
3	The deubiquitinase USP15 antagonizes Parkin-mediated mitochondrial ubiquitination and mitophagy. <i>Human Molecular Genetics</i> , 2014, 23, 5227-5242.	2.9	264
4	Population prevalence of the MELAS A3243G mutation. <i>Mitochondrion</i> , 2007, 7, 230-233.	3.4	248
5	Nomenclature of genetic movement disorders: Recommendations of the international Parkinson and movement disorder society task force. <i>Movement Disorders</i> , 2016, 31, 436-457.	3.9	228
6	Activation of β -Glucocerebrosidase Reduces Pathological α -Synuclein and Restores Lysosomal Function in Parkinson's Patient Midbrain Neurons. <i>Journal of Neuroscience</i> , 2016, 36, 7693-7706.	3.6	220
7	Parkinson's disease-linked human PARK9/ATP13A2 maintains zinc homeostasis and promotes α -Synuclein externalization via exosomes. <i>Human Molecular Genetics</i> , 2014, 23, 2816-2833.	2.9	205
8	Disease-specific, neurosphere-derived cells as models for brain disorders. <i>DMM Disease Models and Mechanisms</i> , 2010, 3, 785-798.	2.4	175
9	Patient care standards for primary mitochondrial disease: a consensus statement from the Mitochondrial Medicine Society. <i>Genetics in Medicine</i> , 2017, 19, 1380-1397.	2.4	173
10	Use of Whole-Exome Sequencing for Diagnosis of Limb-Girdle Muscular Dystrophy. <i>JAMA Neurology</i> , 2015, 72, 1424.	9.0	164
11	Nix restores mitophagy and mitochondrial function to protect against PINK1/Parkin-related Parkinson's disease. <i>Scientific Reports</i> , 2017, 7, 44373.	3.3	152
12	The role of ATP13A2 in Parkinson's disease: Clinical phenotypes and molecular mechanisms. <i>Movement Disorders</i> , 2015, 30, 770-779.	3.9	144
13	Mutant Parkin Impairs Mitochondrial Function and Morphology in Human Fibroblasts. <i>PLoS ONE</i> , 2010, 5, e12962.	2.5	140
14	Parkinson's disease-associated human ATP13A2 (PARK9) deficiency causes zinc dyshomeostasis and mitochondrial dysfunction. <i>Human Molecular Genetics</i> , 2014, 23, 2802-2815.	2.9	136
15	Allogeneic haematopoietic stem cell transplantation for mitochondrial neurogastrointestinal encephalomyopathy. <i>Brain</i> , 2015, 138, 2847-2858.	7.6	128
16	<i>LRRK2</i> mutations impair depolarization-induced mitophagy through inhibition of mitochondrial accumulation of RAB10. <i>Autophagy</i> , 2020, 16, 203-222.	9.1	124
17	Parkinson's disease and the gastrointestinal microbiome. <i>Journal of Neurology</i> , 2020, 267, 2507-2523.	3.6	119
18	Prevalence of Mitochondrial 1555A>G Mutation in Adults of European Descent. <i>New England Journal of Medicine</i> , 2009, 360, 642-644.	27.0	115

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19	Mutations in <i>HSPB8</i> causing a new phenotype of distal myopathy and motor neuropathy. <i>Neurology</i> , 2016, 86, 391-398.	1.1	107
20	ATP13A2 mutations impair mitochondrial function in fibroblasts from patients with Kufor-Rakeb syndrome. <i>Neurobiology of Aging</i> , 2012, 33, 1843.e1-1843.e7.	3.1	106
21	Thioredoxin interacting protein (TXNIP) regulates tubular autophagy and mitophagy in diabetic nephropathy through the mTOR signaling pathway. <i>Scientific Reports</i> , 2016, 6, 29196.	3.3	106
22	Pathogenic effects of novel mutations in the P-type ATPase <i>ATP13A2</i> (<i>PARK9</i>) causing Kufor-Rakeb syndrome, a form of early-onset parkinsonism. <i>Human Mutation</i> , 2011, 32, 956-964.	2.5	105
23	A <i>SLC39A8</i> variant causes manganese deficiency, and glycosylation and mitochondrial disorders. <i>Journal of Inherited Metabolic Disease</i> , 2017, 40, 261-269.	3.6	101
24	Mitochondrial disease in adults: recent advances and future promise. <i>Lancet Neurology</i> , The, 2021, 20, 573-584.	10.2	96
25	A comparison of current serum biomarkers as diagnostic indicators of mitochondrial diseases. <i>Neurology</i> , 2016, 86, 2010-2015.	1.1	89
26	Fibroblast growth factor 21 is a sensitive biomarker of mitochondrial disease. <i>Neurology</i> , 2013, 81, 1819-1826.	1.1	85
27	Systematic review of cardiac electrical disease in Kearns-Sayre syndrome and mitochondrial cytopathy. <i>International Journal of Cardiology</i> , 2015, 181, 303-310.	1.7	81
28	Identical Mitochondrial DNA Deletion in a Woman with Ocular Myopathy and in Her Son with Pearson Syndrome. <i>American Journal of Human Genetics</i> , 2002, 71, 679-683.	6.2	76
29	Frequency of the D620N Mutation in VPS35 in Parkinson Disease. <i>Archives of Neurology</i> , 2012, 69, 1360.	4.5	76
30	Genetic mimics of cerebral palsy. <i>Movement Disorders</i> , 2019, 34, 625-636.	3.9	76
31	TIA1 variant drives myodegeneration in multisystem proteinopathy with SQSTM1 mutations. <i>Journal of Clinical Investigation</i> , 2018, 128, 1164-1177.	8.2	75
32	<i>SQSTM1</i> splice site mutation in distal myopathy with rimmed vacuoles. <i>Neurology</i> , 2015, 85, 665-674.	1.1	74
33	Mutations in <i>GNAL</i> . <i>JAMA Neurology</i> , 2014, 71, 490.	9.0	70
34	Loss of Function Variants in <i>HOPS</i> Complex Genes <i>VPS16</i> and <i>VPS41</i> Cause Early Onset Dystonia Associated with Lysosomal Abnormalities. <i>Annals of Neurology</i> , 2020, 88, 867-877.	5.3	70
35	Mutation in mitochondrial ribosomal protein S7 (<i>MRPS7</i>) causes congenital sensorineural deafness, progressive hepatic and renal failure and lactic acidemia. <i>Human Molecular Genetics</i> , 2015, 24, 2297-2307.	2.9	64
36	Aerobic exercise and muscle metabolism in patients with mitochondrial myopathy. <i>Muscle and Nerve</i> , 2006, 33, 524-531.	2.2	62

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37	The diagnostic utility of genome sequencing in a pediatric cohort with suspected mitochondrial disease. <i>Genetics in Medicine</i> , 2020, 22, 1254-1261.	2.4	59
38	Pigmentary retinopathy associated with the mitochondrial DNA 3243 point mutation. <i>Neurology</i> , 1997, 49, 1013-1017.	1.1	56
39	A patient-derived stem cell model of hereditary spastic paraplegia with <i>SPAST</i> mutations. <i>DMM Disease Models and Mechanisms</i> , 2013, 6, 489-502.	2.4	55
40	Expanding the phenotype of GMPPB mutations. <i>Brain</i> , 2015, 138, 836-844.	7.6	54
41	Olfactory impairment in older adults is associated with poorer diet quality over 5 years. <i>European Journal of Nutrition</i> , 2016, 55, 1081-1087.	3.9	51
42	Targeted next generation sequencing in <i>SPAST</i> -negative hereditary spastic paraplegia. <i>Journal of Neurology</i> , 2013, 260, 2516-2522.	3.6	49
43	Mechanism of impaired microtubule-dependent peroxisome trafficking and oxidative stress in <i>SPAST</i> -mutated cells from patients with Hereditary Spastic Paraplegia. <i>Scientific Reports</i> , 2016, 6, 27004.	3.3	49
44	The broadening spectrum of mitochondrial disease: Shifts in the diagnostic paradigm. <i>Biochimica Et Biophysica Acta - General Subjects</i> , 2014, 1840, 1360-1367.	2.4	48
45	<i>LARS2</i> Variants Associated with Hydrops, Lactic Acidosis, Sideroblastic Anemia, and Multisystem Failure. <i>JIMD Reports</i> , 2015, 28, 49-57.	1.5	48
46	Functional hyperspectral imaging captures subtle details of cell metabolism in olfactory neurosphere cells, disease-specific models of neurodegenerative disorders. <i>Biochimica Et Biophysica Acta - Molecular Cell Research</i> , 2016, 1863, 56-63.	4.1	48
47	Gastrointestinal dysfunction in Parkinson's disease. <i>Journal of Neurology</i> , 2020, 267, 1377-1388.	3.6	48
48	Low dose tubulin-binding drugs rescue peroxisome trafficking deficit in patient-derived stem cells in Hereditary Spastic Paraplegia. <i>Biology Open</i> , 2014, 3, 494-502.	1.2	47
49	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. <i>Pharmacological Research</i> , 2015, 100, 24-35.	7.1	47
50	Standardized practices for RNA diagnostics using clinically accessible specimens reclassifies 75% of putative splicing variants. <i>Genetics in Medicine</i> , 2022, 24, 130-145.	2.4	45
51	Whole genome sequencing for the genetic diagnosis of heterogenous dystonia phenotypes. <i>Parkinsonism and Related Disorders</i> , 2019, 69, 111-118.	2.2	44
52	Diagnosis of "possible" mitochondrial disease: an existential crisis. <i>Journal of Medical Genetics</i> , 2019, 56, 123-130.	3.2	42
53	Movement disorders in mitochondrial disease. <i>Journal of Neurology</i> , 2018, 265, 1230-1240.	3.6	41
54	Role of microRNAs in the Regulation of α -Synuclein Expression: A Systematic Review. <i>Frontiers in Molecular Neuroscience</i> , 2016, 9, 128.	2.9	38

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55	Use of Whole-Genome Sequencing for Mitochondrial Disease Diagnosis. <i>Neurology</i> , 2022, 99, .	1.1	33
56	Defining the genetic basis of early onset hereditary spastic paraplegia using whole genome sequencing. <i>Neurogenetics</i> , 2016, 17, 265-270.	1.4	32
57	PARK Genes Link Mitochondrial Dysfunction and Alpha-Synuclein Pathology in Sporadic Parkinson's Disease. <i>Frontiers in Cell and Developmental Biology</i> , 2021, 9, 612476.	3.7	32
58	The Genetics of Mitochondrial Disease. <i>Seminars in Neurology</i> , 2011, 31, 519-530.	1.4	32
59	Mitochondrial diseases. <i>Handbook of Clinical Neurology</i> / Edited By P J Vinken and G W Bruyn, 2018, 147, 125-141.	1.8	30
60	The gut microbiota: A novel therapeutic target in Parkinson's disease?. <i>Parkinsonism and Related Disorders</i> , 2019, 66, 265-266.	2.2	30
61	Two Faces of the Same Coin: Benign Familial Infantile Seizures and Paroxysmal Kinesigenic Dyskinesia Caused by <i>PRRT2</i> Mutations. <i>Archives of Neurology</i> , 2012, 69, 668.	4.5	28
62	Increased Diagnostic Yield of Spastic Paraplegia with or Without Cerebellar Ataxia Through Whole-Genome Sequencing. <i>Cerebellum</i> , 2019, 18, 781-790.	2.5	28
63	Depression in Parkinson's disease: Perspectives from an Australian cohort. <i>Journal of Affective Disorders</i> , 2020, 277, 1038-1044.	4.1	26
64	An Update on the Hereditary Spastic Paraplegias: New Genes and New Disease Models. <i>Movement Disorders Clinical Practice</i> , 2015, 2, 213-223.	1.5	25
65	Mitochondrial DNA haplogroups J and K are not protective for Parkinson's disease in the Australian community. <i>Movement Disorders</i> , 2009, 24, 290-292.	3.9	23
66	Serum FGF-21, GDF-15, and blood mtDNA copy number are not biomarkers of Parkinson disease. <i>Neurology: Clinical Practice</i> , 2020, 10, 40-46.	1.6	23
67	Oxidative Stress-Induced Axon Fragmentation Is a Consequence of Reduced Axonal Transport in Hereditary Spastic Paraplegia SPAST Patient Neurons. <i>Frontiers in Neuroscience</i> , 2020, 14, 401.	2.8	23
68	Increased Added Sugar Consumption Is Common in Parkinson's Disease. <i>Frontiers in Nutrition</i> , 2021, 8, 628845.	3.7	23
69	The phenotypic spectrum of dystonia in Mohr-Tranebjaerg syndrome. <i>Movement Disorders</i> , 2012, 27, 1034-1040.	3.9	22
70	Health-Related Quality of Life for Parkinson's Disease Patients and Their Caregivers. <i>Journal of Movement Disorders</i> , 2021, 14, 42-52.	1.3	22
71	Expanding the spectrum of PEX16 mutations and novel insights into disease mechanisms. <i>Molecular Genetics and Metabolism Reports</i> , 2018, 16, 46-51.	1.1	21
72	High Degree of Genetic Heterogeneity for Hereditary Cerebellar Ataxias in Australia. <i>Cerebellum</i> , 2019, 18, 137-146.	2.5	21

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73	Loss of ATP13A2 impairs glycolytic function in Kufor-Rakeb syndrome patient-derived cell models. <i>Parkinsonism and Related Disorders</i> , 2016, 27, 67-73.	2.2	20
74	The impact of device-assisted therapies on the gut microbiome in Parkinson's disease. <i>Journal of Neurology</i> , 2022, 269, 780-795.	3.6	19
75	Motor protein binding and mitochondrial transport are altered by pathogenic TUBB4A variants. <i>Human Mutation</i> , 2018, 39, 1901-1915.	2.5	17
76	Mitochondrial Function in Hereditary Spastic Paraplegia: Deficits in SPG7 but Not SPAST Patient-Derived Stem Cells. <i>Frontiers in Neuroscience</i> , 2020, 14, 820.	2.8	17
77	Dietary intakes of fats, fish and nuts and olfactory impairment in older adults. <i>British Journal of Nutrition</i> , 2015, 114, 240-247.	2.3	15
78	The Gut Microbiome in Parkinson's Disease: A Longitudinal Study of the Impacts on Disease Progression and the Use of Device-Assisted Therapies. <i>Frontiers in Aging Neuroscience</i> , 2022, 14, .	3.4	15
79	Practical approaches to commencing device-assisted therapies for Parkinson disease in Australia. <i>Internal Medicine Journal</i> , 2017, 47, 1107-1113.	0.8	14
80	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. <i>BMJ Open</i> , 2020, 10, e040230.	1.9	14
81	Patient-Derived Stem Cell Models in SPAST HSP: Disease Modelling and Drug Discovery. <i>Brain Sciences</i> , 2018, 8, 142.	2.3	12
82	Motor Evoked Potentials in Hereditary Spastic Paraplegia—A Systematic Review. <i>Frontiers in Neurology</i> , 2019, 10, 967.	2.4	12
83	A Novel Homozygous Mutation in the FUCA1 Gene Highlighting Fucosidosis as a Cause of Dystonia: Case Report and Literature Review. <i>Neuropediatrics</i> , 2019, 50, 248-252.	0.6	10
84	Single cell morphology distinguishes genotype and drug effect in Hereditary Spastic Paraplegia. <i>Scientific Reports</i> , 2021, 11, 16635.	3.3	10
85	Nutritional Intake and Gut Microbiome Composition Predict Parkinson's Disease. <i>Frontiers in Aging Neuroscience</i> , 2022, 14, .	3.4	10
86	Mitochondrial disease: recognising more than just the tip of the iceberg. <i>Medical Journal of Australia</i> , 2010, 193, 195-196.	1.7	9
87	Hereditary Parkinsonism-Associated Genetic Variations in PARK9 Locus Lead to Functional Impairment of ATPase Type 13A2. <i>Current Protein and Peptide Science</i> , 2017, 18, 725-732.	1.4	8
88	Mitochondrial DNA disease prevalence: Still underrecognized?. <i>Annals of Neurology</i> , 2008, 64, 471-471.	5.3	7
89	EPG5-Related Vici Syndrome: A Primary Defect of Autophagic Regulation with an Emerging Phenotype Overlapping with Mitochondrial Disorders. <i>JIMD Reports</i> , 2017, 42, 19-29.	1.5	7
90	Dystonia Responsive to Dopamine: POLG Mutations Should Be Considered If Sensory Neuropathy Is Present. <i>Journal of Movement Disorders</i> , 2021, 14, 157-160.	1.3	6

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91	Levodopa-carbidopa intestinal gel: "dismantling the road blocks of a journey"™. Internal Medicine Journal, 2018, 48, 472-474.	0.8	5
92	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
93	Mutations in <i>TUBB4A</i> and spastic paraplegia. Movement Disorders, 2015, 30, 1857-1858.	3.9	4
94	Single Heterozygous <i>ATP13A2</i> Mutations Cause Cellular Dysfunction Associated with Parkinson's Disease. Movement Disorders, 2018, 33, 852-854.	3.9	4
95	Long-Term Follow-Up and Evolution of ADCY5 " From a Ballistic to Dystonic Phenotype. Movement Disorders Clinical Practice, 2020, 7, 985-986.	1.5	4
96	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 <i>YARS2</i> pathogenic variants. American Journal of Medical Genetics, Part A, 2022, 188, 2226-2230.	1.2	4
97	Response to Newman et al.. Genetics in Medicine, 2017, 19, 1380-1380.	2.4	3
98	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
99	Mitochondrial donation: is Australia ready?. Medical Journal of Australia, 2022, 216, 118-121.	1.7	3
100	Cognitive Influences in Parkinson's Disease Patients and Their Caregivers: Perspectives From an Australian Cohort. Frontiers in Neurology, 2021, 12, 673816.	2.4	3
101	Strong Predictive Algorithm of Pathogenesis-Based Biomarkers Improves Parkinson's Disease Diagnosis. Molecular Neurobiology, 2022, 59, 1476-1485.	4.0	3
102	Antibody-Free Targeted Proteomics Assay for Absolute Measurement of α -Tubulin Acetylation. Analytical Chemistry, 2020, 92, 11204-11212.	6.5	2
103	Viewpoint on Milestones for Fellowship Training in Movement Disorders. Movement Disorders, 2022, 37, 1605-1609.	3.9	2
104	Parkin western blotting is useful for identification of patients with Parkin-related Parkinson's disease: Figure 1. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 1436-1437.	1.9	1
105	Reply letter to Jinnah "Locus pocus" and Albanese "Complex dystonia is not a category in the new 2013 consensus classification": Necessary evolution, no magic!. Movement Disorders, 2016, 31, 1760-1762.	3.9	1
106	Maximizing benefits of the levodopa/carbidopa intestinal gel: Systematic considerations, challenging convention and individualizing approaches. Basal Ganglia, 2018, 14, 58-60.	0.3	1
107	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
108	How Do I Manage Patients With the Levodopa/Carbidopa Intestinal Gel?. Movement Disorders Clinical Practice, 2019, 6, 181-181.	1.5	1

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109	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
110	046â€¦A critical review of biomarkers for hereditary spastic paraplegia. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, A15.3-A16.	1.9	0
111	037â€¦The gut microbiome in Parkinsonâ€™s disease: longitudinal insights into disease progression and the use of device-assisted therapies. , 2021, , .		0
112	091â€¦The impact of device-assisted therapy initiation on the gut microbiome in Parkinsonâ€™s disease. , 2021, , .		0
113	015â€¦Gut microbiota and nutritional profiles of Parkinsonâ€™s disease patients. , 2021, , .		0