Caroline Anne Vance

List of Publications by Year
in descending order

52	10,776	126708 33	189595 50
papers	citations	h-index	g-index

55	55	55	10155
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Mutations in FUS, an RNA Processing Protein, Cause Familial Amyotrophic Lateral Sclerosis Type 6. Science, 2009, 323, 1208-1211.	6.0	2,295
2	TDP-43 Mutations in Familial and Sporadic Amyotrophic Lateral Sclerosis. Science, 2008, 319, 1668-1672.	6.0	2,268
3	Exome sequencing in amyotrophic lateral sclerosis identifies risk genes and pathways. Science, 2015, 347, 1436-1441.	6.0	823
4	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. Neuron, 2018, 97, 1268-1283.e6.	3.8	517
5	Hexanucleotide Repeats in ALS/FTD Form Length-Dependent RNA Foci, Sequester RNA Binding Proteins, and Are Neurotoxic. Cell Reports, 2013, 5, 1178-1186.	2.9	419
6	Novel Mutations in TARDBP (TDP-43) in Patients with Familial Amyotrophic Lateral Sclerosis. PLoS Genetics, 2008, 4, e1000193.	1.5	393
7	Familial amyotrophic lateral sclerosis with frontotemporal dementia is linked to a locus on chromosome 9p13.2â€“21.3. Brain, 2006, 129, 868-876.	3.7	363
8	Exome-wide Rare Variant Analysis Identifies TUBA4A Mutations Associated with Familial ALS. Neuron, 2014, 84, 324-331.	3.8	308
9	Familial amyotrophic lateral sclerosis is associated with a mutation in D-amino acid oxidase. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 7556-7561.	3.3	229
10	Overexpression of human wild-type FUS causes progressive motor neuron degeneration in an age- and dose-dependent fashion. Acta Neuropathologica, 2013, 125, 273-288.	3.9	225
11	NEK1 variants confer susceptibility to amyotrophic lateral sclerosis. Nature Genetics, 2016, 48, 1037-1042.	9.4	218
12	Differential roles of the ubiquitin proteasome system (UPS) and autophagy in the clearance of soluble and aggregated TDP-43 species. Journal of Cell Science, 2014, 127, 1263-78.	1.2	216
13	Chromosome 9p21 in sporadic amyotrophic lateral sclerosis in the UK and seven other countries: a genome-wide association study. Lancet Neurology, The, 2010, 9, 986-994.	4.9	205
14	The C9ORF72 expansion mutation is a common cause of ALS+/â˜FTD in Europe and has a single founder. European Journal of Human Genetics, 2013, 21, 102-108.	1.4	201
15	ALS mutant FUS disrupts nuclear localization and sequesters wild-type FUS within cytoplasmic stress granules. Human Molecular Genetics, 2013, 22, 2676-2688.	1.4	199
16	CCNF mutations in amyotrophic lateral sclerosis and frontotemporal dementia. Nature Communications, 2016, 7, 11253.	5.8	174
17	Mutational analysis reveals the FUS homolog TAF15 as a candidate gene for familial amyotrophic lateral sclerosis. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2011, 156, 285-290.	1.1	148
18	The heat shock response plays an important role in TDP-43 clearance: evidence for dysfunction in amyotrophic lateral sclerosis. Brain, 2016, 139, 1417-1432.	3.7	131

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19	Mutations in the vesicular trafficking protein annexin A11 are associated with amyotrophic lateral sclerosis. Science Translational Medicine, 2017, 9, .	5.8	129
20	A genome-wide association meta-analysis identifies a novel locus at 17q11.2 associated with sporadic amyotrophic lateral sclerosis. Human Molecular Genetics, 2014, 23, 2220-2231.	1.4	123
21	An MND/ALS phenotype associated with C9orf72 repeat expansion: Abundant p62â€positive, TDPâ€43â€negative inclusions in cerebral cortex, hippocampus and cerebellum but without associated cognitive decline. Neuropathology, 2012, 32, 505-514.	0.7	110
22	Novel mutations support a role for Profilin 1 in the pathogenesis of ALS. Neurobiology of Aging, 2015, 36, 1602.e17-1602.e27.	1.5	87
23	Amyotrophic lateral sclerosis-like superoxide dismutase 1 proteinopathy is associated with neuronal loss in Parkinsonâ€™s disease brain. Acta Neuropathologica, 2017, 134, 113-127.	3.9	78
24	Two Families with Familial Amyotrophic Lateral Sclerosis Are Linked to a Novel Locus on Chromosome 16q. American Journal of Human Genetics, 2003, 73, 390-396.	2.6	76
25	Mitochondrial abnormalities and disruption of the neuromuscular junction precede the clinical phenotype and motor neuron loss in hFUSWT transgenic mice. Human Molecular Genetics, 2018, 27, 463-474.	1.4	74
26	Wild type human TDP-43 potentiates ALS-linked mutant TDP-43 driven progressive motor and cortical neuron degeneration with pathological features of ALS. Acta Neuropathologica Communications, 2015, 3, 36.	2.4	73
27	Optineurin inclusions occur in a minority of TDP-43 positive ALS and FTLD-TDP cases and are rarely observed in other neurodegenerative disorders. Acta Neuropathologica, 2011, 121, 519-527.	3.9	70
28	Age of onset of amyotrophic lateral sclerosis is modulated by a locus on 1p34.1. Neurobiology of Aging, 2013, 34, 357.e7-357.e19.	1.5	69
29	Non-nuclear Pool of Splicing Factor SFPQ Regulates Axonal Transcripts Required for Normal Motor Development. Neuron, 2017, 94, 322-336.e5.	3.8	61
30	ALS-associated missense and nonsense TBK1 mutations can both cause loss of kinase function. Neurobiology of Aging, 2018, 71, 266.e1-266.e10.	1.5	59
31	Pathogenic Huntingtin Repeat Expansions in Patients with Frontotemporal Dementia and Amyotrophic Lateral Sclerosis. Neuron, 2021, 109, 448-460.e4.	3.8	56
32	Association of Variants in the SPTLC1 Gene With Juvenile Amyotrophic Lateral Sclerosis. JAMA Neurology, 2021, 78, 1236.	4.5	46
33	Granule Localization of Glutaminase in Human Neutrophils and the Consequence of Glutamine Utilization for Neutrophil Activity. Journal of Biological Chemistry, 2004, 279, 13305-13310.	1.6	44
34	Transportin 1 colocalization with Fused in Sarcoma (FUS) inclusions is not characteristic for amyotrophic lateral sclerosisâ€FUS confirming disrupted nuclear import of mutant FUS and distinguishing it from frontotemporal lobar degeneration with FUS inclusions. Neuropathology and Applied Neurobiology, 2013, 39, 553-561.	1.8	27
35	Altered SOD1 maturation and post-translational modification in amyotrophic lateral sclerosis spinal cord. Brain, 2022, 145, 3108-3130.	3.7	25
36	ALS-FUS pathology revisited: singleton FUS mutations and an unusual case with both a FUS and TARDBP mutation. Acta Neuropathologica Communications, 2015, 3, 62.	2.4	22

#	Article	IF	Citations
37	Four novel SPG3A/atlastin mutations identified in autosomal dominant hereditary spastic paraplegia kindreds with intraâ€familial variability in age of onset and complex phenotype. Clinical Genetics, 2009, 75, 485-489.	1.0	21
38	The CHCHD10 P34S variant is not associated with ALS in a UK cohort of familial and sporadic patients. Neurobiology of Aging, 2015, 36, 2908.e17-2908.e18.	1.5	19
39	Screening for OPTN mutations in a cohort of British amyotrophic lateral sclerosis patients. Neurobiology of Aging, 2012, 33, 2948.e15-2948.e17.	1.5	18
40	C9ORF72 and UBQLN2 mutations are causes of amyotrophic lateral sclerosis in New Zealand: a genetic and pathologic study using banked human brain tissue. Neurobiology of Aging, 2017, 49, 214.e1-214.e5.	1.5	18
41	Antisense oligonucleotide therapies for Amyotrophic Lateral Sclerosis: Existing and emerging targets. International Journal of Biochemistry and Cell Biology, 2019, 110, 149-153.	1.2	18
42	Mutation analysis of VCP in British familial and sporadic amyotrophic lateral sclerosis patients. Neurobiology of Aging, 2012, 33, 2721.e1-2721.e2.	1.5	16
43	Striking phenotypic variation in a family with the P506S UBQLN2 mutation including amyotrophic lateral sclerosis, spastic paraplegia, and frontotemporal dementia. Neurobiology of Aging, 2019, 73, 229.e5-229.e9.	1.5	16
44	Autosomal dominant inheritance of rapidly progressive amyotrophic lateral sclerosis due to a truncation mutation in the fused in sarcoma (FUS) gene. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2014, 15, 557-562.	1.1	15
45	Identification of a novel interaction of FUS and syntaphilin may explain synaptic and mitochondrial abnormalities caused by ALS mutations. Scientific Reports, 2021, 11, 13613.	1.6	15
46	Review: Modelling the pathology and behaviour of frontotemporal dementia. Neuropathology and Applied Neurobiology, 2019, 45, 58-80.	1.8	13
47	Genetic analysis of amyotrophic lateral sclerosis in the Slovenian population. Neurobiology of Aging, 2015, 36, 1601.e17-1601.e20.	1.5	10
48	CHMP2B mutations are not a common cause of familial or sporadic amyotrophic lateral sclerosis. Journal of Neurology, Neurosurgery and Psychiatry, 2008, 79, 849-850.	0.9	7
49	Genome-Wide Analyses Identify KIF5A as a Novel ALS Gene. SSRN Electronic Journal, 0, , .	0.4	4
50	A recessive S174X mutation in Optineurin causes amyotrophic lateral sclerosis through a loss of function via allele-specific nonsense-mediated decay. Neurobiology of Aging, 2021, 106, 1-6.	1.5	3
51	Amyotrophic lateral sclerosis and other disorders of the lower motor neuron. , 0, , 136-147.		0
52	Expanded G4C2 repeats linked to C9ORF72ALS and FTD form length-dependent RNA foci, sequester RNA binding proteins and are neurotoxic. Molecular Neurodegeneration, 2013, 8, .	4.4	0