Agnes Lumi Nishimura

List of Publications by Year
in descending order

41 papers	7,246 citations	218381 26 h-index	288905 40 g-index
42 all docs	42 docs citations	42 times ranked	8371 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	Characterizing the RNA targets and position-dependent splicing regulation by TDP-43. Nature Neuroscience, 2011, 14, 452-458.	7.1	956
3	A Mutation in the Vesicle-Trafficking Protein VAPB Causes Late-Onset Spinal Muscular Atrophy and Amyotrophic Lateral Sclerosis. American Journal of Human Genetics, 2004, 75, 822-831.	2.6	854
4	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
5	Mutant induced pluripotent stem cell lines recapitulate aspects of TDP-43 proteinopathies and reveal cell-specific vulnerability. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, 5803-5808.	3.3	308
6	Astrocyte pathology and the absence of non-cell autonomy in an induced pluripotent stem cell model of TDP-43 proteinopathy. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 4697-4702.	3.3	301
7	FUS-SMN Protein Interactions Link the Motor Neuron Diseases ALS and SMA. Cell Reports, 2012, 2, 799-806.	2.9	229
8	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
9	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
10	Nuclear import impairment causes cytoplasmic trans-activation response DNA-binding protein accumulation and is associated with frontotemporal lobar degeneration. Brain, 2010, 133, 1763-1771.	3.7	165
11	C9ORF72 repeat expansion causes vulnerability of motor neurons to Ca2+-permeable AMPA receptor-mediated excitotoxicity. Nature Communications, 2018, 9, 347.	5.8	151
12	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
13	Downregulation of MicroRNA-9 in iPSC-Derived Neurons of FTD/ALS Patients with TDP-43 Mutations. PLoS ONE, 2013, 8, e76055.	1.1	117
14	Sexually dimorphic effect of the Val66Met polymorphism of BDNF on susceptibility to Alzheimer's disease: New data and metaâ€analysis. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2010, 153B, 235-242.	1.1	89
15	A common founder for amyotrophic lateral sclerosis type 8 (ALS8) in the Brazilian population. Human Genetics, 2005, 118, 499-500.	1.8	85
16	Rett Syndrome in a Boy with a 47,XXY Karyotype Confirmed by a Rare Mutation in the MECP2 Gene. Neuropediatrics, 2001, 32, 162-164.	0.3	84
17	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
18	C9orf72 poly GA RAN-translated protein plays a key role in amyotrophic lateral sclerosis via aggregation and toxicity. Human Molecular Genetics, 2017, 26, 4765-4777.	1.4	64

#	Article	IF	Citations
19	A novel locus for late onset amyotrophic lateral sclerosis/motor neurone disease variant at 20q13. Journal of Medical Genetics, 2004, 41, 315-320.	1.5	61
20	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
21	Spastic paraplegia, optic atrophy, and neuropathy is linked to chromosome 11q13. Annals of Neurology, 2005, 57, 730-737.	2.8	53
22	The p.P56S mutation in the VAPB gene is not due to a single founder: the first European case. Clinical Genetics, 2010, 77, 302-303.	1.0	48
23	Allele-Specific Knockdown of ALS-Associated Mutant TDP-43 in Neural Stem Cells Derived from Induced Pluripotent Stem Cells. PLoS ONE, 2014, 9, e91269.	1.1	39
24	Analysis of IL-1Î±, IL-1Î², and IL-RA Polymorphisms in Dysthymia. Journal of Molecular Neuroscience, 2004, 22, 251-256.	1.1	36
25	Analysis of the serotonin transporter polymorphism (5-HTTLPR) in Brazilian patients affected by dysthymia, major depression and bipolar disorder. Molecular Psychiatry, 2000, 5, 348-349.	4.1	30
26	Lack of Association Between the Brain-Derived Neurotrophin Factor (C-270T) Polymorphism and Late-Onset Alzheimer's Disease (LOAD) in Brazilian Patients. Journal of Molecular Neuroscience, 2004, 22, 257-260.	1.1	30
27	Association of MAO A polymorphism and alcoholism in Brazilian females. Psychiatric Genetics, 2005, 15, 141-144.	0.6	27
28	A mutation in human VAP-Bâ€“MSP domain, present in ALS patients, affects the interaction with other cellular proteins. Protein Expression and Purification, 2007, 55, 139-146.	0.6	24
29	Monoamine Oxidase A Polymorphism in Brazilian Patients: Risk Factor for Late-Onset Alzheimer's Disease?. Journal of Molecular Neuroscience, 2005, 27, 213-218.	1.1	20
30	The Use of Stem Cells to Model Amyotrophic Lateral Sclerosis and Frontotemporal Dementia: From Basic Research to Regenerative Medicine. Stem Cells International, 2016, 2016, 1-9.	1.2	16
31	The Genetics of Alzheimerâ€™s Disease in Brazil: 10 Years of Analysis in a Unique Population. Journal of Molecular Neuroscience, 2009, 37, 74-79.	1.1	12
32	The human serotonin transporter gene explains why some populations are more optimistic?. Molecular Psychiatry, 2009, 14, 828-828.	4.1	10
33	Analysis of the disease risk locus DXS1047 polymorphism in Brazilian Alzheimer patients. Molecular Psychiatry, 2000, 5, 563-566.	4.1	9
34	Synaptopathy Mechanisms in ALS Caused by C9orf72 Repeat Expansion. Frontiers in Cellular Neuroscience, 2021, 15, 660693.	1.8	9
35	Comment on â€œDrug Screening for ALS Using Patient-Specific Induced Pluripotent Stem Cellsâ€• Science Translational Medicine, 2013, 5, 188le2.	5.8	7
36	ALS-linked FUS mutants affect the localization of U7 snRNP and replication-dependent histone gene expression in human cells. Scientific Reports, 2021, 11, 11868.	1.6	7

#	Article	IF	Citations
37	No evidence of association between the D10S1423 locus and Alzheimer disease in Brazilian patients. Journal of Neural Transmission, 2001, 108, 305-310.	1.4	6
38	Generation of six induced pluripotent stem cell lines from patients with amyotrophic lateral sclerosis with associated genetic mutations in either FUS or ANXA11. Stem Cell Research, 2021, 52, 102246.	0.3	3
39	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
40	iPS Cells and Spinocerebellar Ataxia. Pancreatic Islet Biology, 2015, , 45-61.	0.1	1
41	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