Albert R La Spada

List of Publications by Year
in descending order

84 papers	16,800 citations	66343 42 h-index	56724 83 g-index
137 all docs	137 docs citations	137 times ranked	28827 citing authors

#	Article	IF	Citations
1	Guidelines for the use and interpretation of assays for monitoring autophagy (3rd edition). Autophagy, 2016, 12, 1-222.	9.1	4,701
2	Guidelines for the use and interpretation of assays for monitoring autophagy. Autophagy, 2012, 8, 445-544.	9.1	3,122
3	Androgen receptor gene mutations in X-linked spinal and bulbar muscular atrophy. Nature, 1991, 352, 77-79.	27.8	2,710
4	Thermoregulatory and metabolic defects in Huntington's disease transgenic mice implicate PGC-1Î± in Huntington's disease neurodegeneration. Cell Metabolism, 2006, 4, 349-362.	16.2	519
5	Repeat expansion disease: progress and puzzles in disease pathogenesis. Nature Reviews Genetics, 2010, 11, 247-258.	16.3	425
6	PGC-1Î± Rescues Huntingtonâ€™s Disease Proteotoxicity by Preventing Oxidative Stress and Promoting TFEB Function. Science Translational Medicine, 2012, 4, 142ra97.	12.4	376
7	Converging pathways in neurodegeneration, from genetics to mechanisms. Nature Neuroscience, 2018, 21, 1300-1309.	14.8	325
8	Fasting Activates Fatty Acid Oxidation to Enhance Intestinal Stem Cell Function during Homeostasis and Aging. Cell Stem Cell, 2018, 22, 769-778.e4.	11.1	266
9	A Simple Composite Phenotype Scoring System for Evaluating Mouse Models of Cerebellar Ataxia. Journal of Visualized Experiments, 2010, , .	0.3	253
10	Polyglutamine-Expanded Ataxin-7 Antagonizes CRX Function and Induces Cone-Rod Dystrophy in a Mouse Model of SCA7. Neuron, 2001, 31, 913-927.	8.1	244
11	Purkinje cell degeneration (pcd) Phenotypes Caused by Mutations in the Axotomy-Induced Gene, Nna1. Science, 2002, 295, 1904-1906.	12.6	217
12	Polyglutamine-expanded ataxin-7 inhibits STAGA histone acetyltransferase activity to produce retinal degeneration. Proceedings of the National Academy of Sciences of the United States of America, 2005, 102, 8472-8477.	7.1	215
13	Reduced C9ORF72 function exacerbates gain of toxicity from ALS/FTD-causing repeat expansion in C9orf72. Nature Neuroscience, 2020, 23, 615-624.	14.8	157
14	Muscle Expression of Mutant Androgen Receptor Accounts for Systemic and Motor Neuron Disease Phenotypes in Spinal and Bulbar Muscular Atrophy. Neuron, 2014, 82, 295-307.	8.1	150
15	Peripheral Androgen Receptor Gene Suppression Rescues Disease in Mouse Models of Spinal and Bulbar Muscular Atrophy. Cell Reports, 2014, 7, 774-784.	6.4	148
16	Polyglutamine-expanded androgen receptor interferes with TFEB to elicit autophagy defects in SBMA. Nature Neuroscience, 2014, 17, 1180-1189.	14.8	142
17	TFEB dysregulation as a driver of autophagy dysfunction in neurodegenerative disease: Molecular mechanisms, cellular processes, and emerging therapeutic opportunities. Neurobiology of Disease, 2019, 122, 83-93.	4.4	135
18	Mitochondrial autophagy in neural function, neurodegenerative disease, neuron cell death, and aging. Neurobiology of Disease, 2011, 43, 46-51.	4.4	119

#	Article	IF	Citations
19	The many faces of autophagy dysfunction in Huntington's disease: from mechanism to therapy. Drug Discovery Today, 2014, 19, 963-971.	6.4	112
20	Nutrient Deprivation Induces Neuronal Autophagy and Implicates Reduced Insulin Signaling in Neuroprotective Autophagy Activation. Journal of Biological Chemistry, 2009, 284, 2363-2373.	3.4	107
21	PGC-1Î± at the intersection of bioenergetics regulation and neuron function: From Huntington's disease to Parkinson's disease and beyond. Progress in Neurobiology, 2012, 97, 142-151.	5.7	106
22	S-Nitrosylation of Dynamin-Related Protein 1 Mediates Mutant Huntingtin-Induced Mitochondrial Fragmentation and Neuronal Injury in Huntington's Disease. Antioxidants and Redox Signaling, 2013, 19, 1173-1184.	5.4	104
23	Autophagy activation and enhanced mitophagy characterize the Purkinje cells of pcd mice prior to neuronal death. Molecular Brain, 2009, 2, 24.	2.6	95
24	PPAR-Î´ is repressed in Huntington's disease, is required for normal neuronal function and can be targeted therapeutically. Nature Medicine, 2016, 22, 37-45.	30.7	88
25	Transcriptional regulation of core autophagy and lysosomal genes by the androgen receptor promotes prostate cancer progression. Autophagy, 2017, 13, 506-521.	9.1	88
26	Mutant huntingtin impairs PNKP and ATXN3, disrupting DNA repair and transcription. ELife, 2019, 8, .	6.0	83
27	Interference of Crx-dependent transcription by ataxin-7 involves interaction between the glutamine regions and requires the ataxin-7 carboxy-terminal region for nuclear localization. Human Molecular Genetics, 2003, 13, 53-67.	2.9	82
28	MAP4K3 mediates amino acid-dependent regulation of autophagy via phosphorylation of TFEB. Nature Communications, 2018, 9, 942.	12.8	80
29	Therapy development in Huntington disease: From current strategies to emerging opportunities. American Journal of Medical Genetics, Part A, 2018, 176, 842-861.	1.2	75
30	Let-7 Coordinately Suppresses Components of the Amino Acid Sensing Pathway to Repress mTORC1 and Induce Autophagy. Cell Metabolism, 2014, 20, 626-638.	16.2	67
31	The CAGâ€“polyglutamine repeat diseases: a clinical, molecular, genetic, and pathophysiologic nosology. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2018, 147, 143-170.	1.8	67
32	Overriding FUS autoregulation in mice triggers gain-of-toxic dysfunctions in RNA metabolism and autophagy-lysosome axis. ELife, 2019, 8, .	6.0	65
33	Antisense oligonucleotides targeting mutant Ataxin-7 restore visual function in a mouse model of spinocerebellar ataxia type 7. Science Translational Medicine, 2018, 10, .	12.4	63
34	Nicotinamide Pathway-Dependent Sirt1 Activation Restores Calcium Homeostasis to Achieve Neuroprotection in Spinocerebellar Ataxia Type 7. Neuron, 2020, 105, 630-644.e9.	8.1	63
35	The SAGA Histone Deubiquitinase Module Controls Yeast Replicative Lifespan via Sir2 Interaction. Cell Reports, 2014, 8, 477-486.	6.4	62
36	A functional deficiency of TERA/VCP/p97 contributes to impaired DNA repair in multiple polyglutamine diseases. Nature Communications, 2013, 4, 1816.	12.8	60

#	Article	IF	Citations
37	Polyglutamine-Expanded Androgen Receptor Truncation Fragments Activate a Bax-Dependent Apoptotic Cascade Mediated by DP5/Hrk. Journal of Neuroscience, 2009, 29, 1987-1997.	3.6	56
38	PPARÎ´ activation by bexarotene promotes neuroprotection by restoring bioenergetic and quality control homeostasis. Science Translational Medicine, 2017, 9, .	12.4	54
39	Increased 4E-BP1 Expression Protects against Diet-Induced Obesity and Insulin Resistance in Male Mice. Cell Reports, 2016, 16, 1903-1914.	6.4	52
40	Nonallele Specific Silencing of Ataxin-7 Improves Disease Phenotypes in a Mouse Model of SCA7. Molecular Therapy, 2014, 22, 1635-1642.	8.2	51
41	Metabolic and Organelle Morphology Defects in Mice and Human Patients Define Spinocerebellar Ataxia Type 7 as a Mitochondrial Disease. Cell Reports, 2019, 26, 1189-1202.e6.	6.4	49
42	Autophagy in polyglutamine disease: Imposing order on disorder or contributing to the chaos?. Molecular and Cellular Neurosciences, 2015, 66, 53-61.	2.2	47
43	PPARGC1A/PGC-1Î±, TFEB and enhanced proteostasis in Huntington disease. Autophagy, 2012, 8, 1845-1847.	9.1	44
44	Senataxin mutations elicit motor neuron degeneration phenotypes and yield TDP-43 mislocalization in ALS4 mice and human patients. Acta Neuropathologica, 2018, 136, 425-443.	7.7	43
45	Reduction of mutant ataxin-7 expression restores motor function and prevents cerebellar synaptic reorganization in a conditional mouse model of SCA7. Human Molecular Genetics, 2013, 22, 890-903.	2.9	42
46	Mitochondrial Dysfunction in NnaD Mutant Flies and Purkinje Cell Degeneration Mice Reveals a Role for Nna Proteins in Neuronal Bioenergetics. Neuron, 2010, 66, 835-847.	8.1	40
47	Autophagy gene haploinsufficiency drives chromosome instability, increases migration, and promotes early ovarian tumors. PLoS Genetics, 2020, 16, e1008558.	3.5	39
48	Adenylyl cyclase activating polypeptide reduces phosphorylation and toxicity of the polyglutamine-expanded androgen receptor in spinobulbar muscular atrophy. Science Translational Medicine, 2016, 8, 370ra181.	12.4	37
49	The zinc-binding domain of Nna1 is required to prevent retinal photoreceptor loss and cerebellar ataxia in Purkinje cell degeneration (pcd) mice. Vision Research, 2008, 48, 1999-2005.	1.4	36
50	The Purkinje cell degeneration 5J mutation is a single amino acid insertion that destabilizes Nna1 protein. Mammalian Genome, 2006, 17, 103-110.	2.2	35
51	Selective modulation of the androgen receptor AF2 domain rescues degeneration in spinal bulbar muscular atrophy. Nature Medicine, 2018, 24, 427-437.	30.7	35
52	Endoplasmic reticulum stress in spinal and bulbar muscular atrophy: a potential target for therapy. Brain, 2014, 137, 1894-1906.	7.6	31
53	Senataxin, A Novel Helicase at the Interface of RNA Transcriptome Regulation and Neurobiology: From Normal Function to Pathological Roles in Motor Neuron Disease and Cerebellar Degeneration. Advances in Neurobiology, 2018, 20, 265-281.	1.8	27
54	Disease modifying effect of adiponectin in model of Î± â€synucleinopathies. Annals of Clinical and Translational Neurology, 2014, 1, 479-489.	3.7	25

#	Article	IF	Citations
55	CCP1 promotes mitochondrial fusion and motility to prevent Purkinje cell neuron loss in pcd mice. Journal of Cell Biology, 2019, 218, 206-219.	5.2	25
56	Efficient recombination-based methods for bacterial artificial chromosome fusion and mutagenesis. Gene, 2006, 371, 136-143.	2.2	24
57	Gene expression analysis reveals early dysregulation of disease pathways and links Chmp7 to pathogenesis of spinal and bulbar muscular atrophy. Scientific Reports, 2019, 9, 3539.	3.3	24
58	Proteolytic cleavage of ataxin-7 promotes SCA7 retinal degeneration and neurological dysfunction. Human Molecular Genetics, 2015, 24, 3908-3917.	2.9	22
59	Targeting protein aggregation in neurodegeneration â€“ lessons from polyglutamine disorders. Expert Opinion on Therapeutic Targets, 2006, 10, 505-513.	3.4	21
60	Points to consider: is there evidence to support BRCA1/2 and other inherited breast cancer genetic testing for all breast cancer patients? A statement of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2020, 22, 681-685.	2.4	20
61	Gene therapy with AR isoform 2 rescues spinal and bulbar muscular atrophy phenotype by modulating AR transcriptional activity. Science Advances, 2021, 7, .	10.3	20
62	Low-Cost Gait Analysis for Behavioral Phenotyping of Mouse Models of Neuromuscular Disease. Journal of Visualized Experiments, 2019, , .	0.3	19
63	Unwinding the role of senataxin in neurodegeneration. Discovery Medicine, 2015, 19, 127-36.	0.5	19
64	Harmony Lost: Cellâ€“Cell Communication at the Neuromuscular Junction in Motor Neuron Disease. Trends in Neurosciences, 2020, 43, 709-724.	8.6	17
65	Nemo-like kinase is a novel regulator of spinal and bulbar muscular atrophy. ELife, 2015, 4, e08493.	6.0	16
66	X-Linked Spinal and Bulbar Muscular Atrophy: From Clinical Genetic Features and Molecular Pathology to Mechanisms Underlying Disease Toxicity. Advances in Experimental Medicine and Biology, 2018, 1049, 103-133.	1.6	15
67	The replicative lifespanâ€extending deletion of SGF73 results in altered ribosomal gene expression in yeast. Aging Cell, 2017, 16, 785-796.	6.7	14
68	Astroglial-targeted expression of the fragile X CGG repeat premutation in mice yields RAN translation, motor deficits and possible evidence for cell-to-cell propagation of FXTAS pathology. Acta Neuropathologica Communications, 2019, 7, 27.	5.2	14
69	SUMOylated Senataxin functions in genome stability, RNA degradation, and stress granule disassembly, and is linked with inherited ataxia and motor neuron disease. Molecular Genetics & Genomic Medicine, 2021, 9, e1745.	1.2	13
70	4E-BP1 Protects Neurons from Misfolded Protein Stress and Parkinson's Disease Toxicity by Inducing the Mitochondrial Unfolded Protein Response. Journal of Neuroscience, 2020, 40, 8734-8745.	3.6	12
71	Differential effects of various genetic mouse models of the mechanistic target of rapamycin complex I inhibition on heart failure. GeroScience, 2019, 41, 847-860.	4.6	10
72	Deterioration of muscle force and contractile characteristics are early pathological events in spinal and bulbar muscular atrophy mice. DMM Disease Models and Mechanisms, 2020, 13, .	2.4	8

#	Article	IF	Citations
73	Respiratory dysfunction in a mouse model of spinocerebellar ataxia type 7. DMM Disease Models and Mechanisms, 2021, 14, .	2.4	7
74	Ataxin-3, DNA Damage Repair, and SCA3 Cerebellar Degeneration: On the Path to Parsimony?. PLoS Genetics, 2015, 11, e1004937.	3.5	6
75	Tight expression regulation of senataxin, linked to motor neuron disease and ataxia, is required to avert cell-cycle block and nucleolus disassembly. Heliyon, 2020, 6, e04165.	3.2	6
76	Mitochondrial dysfunction in iPSC-derived neurons of subjects with chronic mountain sickness. Journal of Applied Physiology, 2018, 125, 832-840.	2.5	5
77	Altered H3 histone acetylation impairs high-fidelity DNA repair to promote cerebellar degeneration in spinocerebellar ataxia type 7. Cell Reports, 2021, 37, 110062.	6.4	5
78	De novo pathogenic variant in SETX causes a rapidly progressive neurodegenerative disorder of early childhood-onset with severe axonal polyneuropathy. Acta Neuropathologica Communications, 2021, 9, 194.	5.2	5
79	Motor neuron degeneration in spinal and Bulbar Muscular Atrophy is a skeletal muscle-driven process: Relevance to therapy development and implications for related motor neuron diseases. Rare Diseases (Austin, Tex), 2014, 2, e962402.	1.8	4
80	In vivo assessment of neurodegeneration in Spinocerebellar Ataxia type 7. NeuroImage: Clinical, 2021, 29, 102561.	2.7	4
81	The expanding world of stem cell modeling of Huntington's disease: creating tools with a promising future. Genome Medicine, 2012, 4, 68.	8.2	3
82	Something wicked this way comes: huntingtin. Nature Neuroscience, 2014, 17, 1014-1015.	14.8	3
83	Identification of the SCA21 disease gene: remaining challenges and promising opportunities. Brain, 2014, 137, 2626-2628.	7.6	1
84	Convergent Transcription at the Ataxinâ€7 Locus Produces dsRNA Fragments that are Processed by Dicerâ€1. FASEB Journal, 2012, 26, 747.4.	0.5	0