

Albert R La Spada

List of Publications by Year in descending order

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Version: 2024-02-01

84
papers

16,800
citations

66315

42
h-index

56687

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). <i>Autophagy</i> , 2016, 12, 1-222.	4.3	4,701
2	Guidelines for the use and interpretation of assays for monitoring autophagy. <i>Autophagy</i> , 2012, 8, 445-544.	4.3	3,122
3	Androgen receptor gene mutations in X-linked spinal and bulbar muscular atrophy. <i>Nature</i> , 1991, 352, 77-79.	13.7	2,710
4	Thermoregulatory and metabolic defects in Huntington's disease transgenic mice implicate PGC-1 β in Huntington's disease neurodegeneration. <i>Cell Metabolism</i> , 2006, 4, 349-362.	7.2	519
5	Repeat expansion disease: progress and puzzles in disease pathogenesis. <i>Nature Reviews Genetics</i> , 2010, 11, 247-258.	7.7	425
6	PGC-1 β Rescues Huntington's Disease Proteotoxicity by Preventing Oxidative Stress and Promoting TFEB Function. <i>Science Translational Medicine</i> , 2012, 4, 142ra97.	5.8	376
7	Converging pathways in neurodegeneration, from genetics to mechanisms. <i>Nature Neuroscience</i> , 2018, 21, 1300-1309.	7.1	325
8	Fasting Activates Fatty Acid Oxidation to Enhance Intestinal Stem Cell Function during Homeostasis and Aging. <i>Cell Stem Cell</i> , 2018, 22, 769-778.e4.	5.2	266
9	A Simple Composite Phenotype Scoring System for Evaluating Mouse Models of Cerebellar Ataxia. <i>Journal of Visualized Experiments</i> , 2010, , .	0.2	253
10	Polyglutamine-Expanded Ataxin-7 Antagonizes CRX Function and Induces Cone-Rod Dystrophy in a Mouse Model of SCA7. <i>Neuron</i> , 2001, 31, 913-927.	3.8	244
11	Purkinje cell degeneration (pcd) Phenotypes Caused by Mutations in the Axotomy-Induced Gene, <i>Nna1</i> . <i>Science</i> , 2002, 295, 1904-1906.	6.0	217
12	Polyglutamine-expanded ataxin-7 inhibits STAGA histone acetyltransferase activity to produce retinal degeneration. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2005, 102, 8472-8477.	3.3	215
13	Reduced C9ORF72 function exacerbates gain of toxicity from ALS/FTD-causing repeat expansion in C9orf72. <i>Nature Neuroscience</i> , 2020, 23, 615-624.	7.1	157
14	Muscle Expression of Mutant Androgen Receptor Accounts for Systemic and Motor Neuron Disease Phenotypes in Spinal and Bulbar Muscular Atrophy. <i>Neuron</i> , 2014, 82, 295-307.	3.8	150
15	Peripheral Androgen Receptor Gene Suppression Rescues Disease in Mouse Models of Spinal and Bulbar Muscular Atrophy. <i>Cell Reports</i> , 2014, 7, 774-784.	2.9	148
16	Polyglutamine-expanded androgen receptor interferes with TFEB to elicit autophagy defects in SBMA. <i>Nature Neuroscience</i> , 2014, 17, 1180-1189.	7.1	142
17	TFEB dysregulation as a driver of autophagy dysfunction in neurodegenerative disease: Molecular mechanisms, cellular processes, and emerging therapeutic opportunities. <i>Neurobiology of Disease</i> , 2019, 122, 83-93.	2.1	135
18	Mitochondrial autophagy in neural function, neurodegenerative disease, neuron cell death, and aging. <i>Neurobiology of Disease</i> , 2011, 43, 46-51.	2.1	119

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19	The many faces of autophagy dysfunction in Huntington's disease: from mechanism to therapy. <i>Drug Discovery Today</i> , 2014, 19, 963-971.	3.2	112
20	Nutrient Deprivation Induces Neuronal Autophagy and Implicates Reduced Insulin Signaling in Neuroprotective Autophagy Activation. <i>Journal of Biological Chemistry</i> , 2009, 284, 2363-2373.	1.6	107
21	PGC-1 β at the intersection of bioenergetics regulation and neuron function: From Huntington's disease to Parkinson's disease and beyond. <i>Progress in Neurobiology</i> , 2012, 97, 142-151.	2.8	106
22	S-Nitrosylation of Dynamin-Related Protein 1 Mediates Mutant Huntingtin-Induced Mitochondrial Fragmentation and Neuronal Injury in Huntington's Disease. <i>Antioxidants and Redox Signaling</i> , 2013, 19, 1173-1184.	2.5	104
23	Autophagy activation and enhanced mitophagy characterize the Purkinje cells of pcd mice prior to neuronal death. <i>Molecular Brain</i> , 2009, 2, 24.	1.3	95
24	PPAR δ is repressed in Huntington's disease, is required for normal neuronal function and can be targeted therapeutically. <i>Nature Medicine</i> , 2016, 22, 37-45.	15.2	88
25	Transcriptional regulation of core autophagy and lysosomal genes by the androgen receptor promotes prostate cancer progression. <i>Autophagy</i> , 2017, 13, 506-521.	4.3	88
26	Mutant huntingtin impairs PNKP and ATXN3, disrupting DNA repair and transcription. <i>ELife</i> , 2019, 8, .	2.8	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. <i>Human Molecular Genetics</i> , 2003, 13, 53-67.	1.4	82
28	MAP4K3 mediates amino acid-dependent regulation of autophagy via phosphorylation of TFEB. <i>Nature Communications</i> , 2018, 9, 942.	5.8	80
29	Therapy development in Huntington disease: From current strategies to emerging opportunities. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 842-861.	0.7	75
30	Let-7 Coordinately Suppresses Components of the Amino Acid Sensing Pathway to Repress mTORC1 and Induce Autophagy. <i>Cell Metabolism</i> , 2014, 20, 626-638.	7.2	67
31	The CAG β polyglutamine repeat diseases: a clinical, molecular, genetic, and pathophysiologic nosology. <i>Handbook of Clinical Neurology</i> / Edited By P J Vinken and G W Bruyn, 2018, 147, 143-170.	1.0	67
32	Overriding FUS autoregulation in mice triggers gain-of-toxic dysfunctions in RNA metabolism and autophagy-lysosome axis. <i>ELife</i> , 2019, 8, .	2.8	65
33	Antisense oligonucleotides targeting mutant Ataxin-7 restore visual function in a mouse model of spinocerebellar ataxia type 7. <i>Science Translational Medicine</i> , 2018, 10, .	5.8	63
34	Nicotinamide Pathway-Dependent Sirt1 Activation Restores Calcium Homeostasis to Achieve Neuroprotection in Spinocerebellar Ataxia Type 7. <i>Neuron</i> , 2020, 105, 630-644.e9.	3.8	63
35	The SAGA Histone Deubiquitinase Module Controls Yeast Replicative Lifespan via Sir2 Interaction. <i>Cell Reports</i> , 2014, 8, 477-486.	2.9	62
36	A functional deficiency of TERA/VCP/p97 contributes to impaired DNA repair in multiple polyglutamine diseases. <i>Nature Communications</i> , 2013, 4, 1816.	5.8	60

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37	Polyglutamine-Expanded Androgen Receptor Truncation Fragments Activate a Bax-Dependent Apoptotic Cascade Mediated by DP5/Hrk. <i>Journal of Neuroscience</i> , 2009, 29, 1987-1997.	1.7	56
38	PPAR γ activation by bexarotene promotes neuroprotection by restoring bioenergetic and quality control homeostasis. <i>Science Translational Medicine</i> , 2017, 9, .	5.8	54
39	Increased 4E-BP1 Expression Protects against Diet-Induced Obesity and Insulin Resistance in Male Mice. <i>Cell Reports</i> , 2016, 16, 1903-1914.	2.9	52
40	Nonallele Specific Silencing of Ataxin-7 Improves Disease Phenotypes in a Mouse Model of SCA7. <i>Molecular Therapy</i> , 2014, 22, 1635-1642.	3.7	51
41	Metabolic and Organelle Morphology Defects in Mice and Human Patients Define Spinocerebellar Ataxia Type 7 as a Mitochondrial Disease. <i>Cell Reports</i> , 2019, 26, 1189-1202.e6.	2.9	49
42	Autophagy in polyglutamine disease: Imposing order on disorder or contributing to the chaos?. <i>Molecular and Cellular Neurosciences</i> , 2015, 66, 53-61.	1.0	47
43	PPARGC1A/PGC-1 β , TFEB and enhanced proteostasis in Huntington disease. <i>Autophagy</i> , 2012, 8, 1845-1847.	4.3	44
44	Senataxin mutations elicit motor neuron degeneration phenotypes and yield TDP-43 mislocalization in ALS4 mice and human patients. <i>Acta Neuropathologica</i> , 2018, 136, 425-443.	3.9	43
45	Reduction of mutant ataxin-7 expression restores motor function and prevents cerebellar synaptic reorganization in a conditional mouse model of SCA7. <i>Human Molecular Genetics</i> , 2013, 22, 890-903.	1.4	42
46	Mitochondrial Dysfunction in NnaD Mutant Flies and Purkinje Cell Degeneration Mice Reveals a Role for Nna Proteins in Neuronal Bioenergetics. <i>Neuron</i> , 2010, 66, 835-847.	3.8	40
47	Autophagy gene haploinsufficiency drives chromosome instability, increases migration, and promotes early ovarian tumors. <i>PLoS Genetics</i> , 2020, 16, e1008558.	1.5	39
48	Adenylyl cyclase activating polypeptide reduces phosphorylation and toxicity of the polyglutamine-expanded androgen receptor in spinobulbar muscular atrophy. <i>Science Translational Medicine</i> , 2016, 8, 370ra181.	5.8	37
49	The zinc-binding domain of Nna1 is required to prevent retinal photoreceptor loss and cerebellar ataxia in Purkinje cell degeneration (pcd) mice. <i>Vision Research</i> , 2008, 48, 1999-2005.	0.7	36
50	The Purkinje cell degeneration 5J mutation is a single amino acid insertion that destabilizes Nna1 protein. <i>Mammalian Genome</i> , 2006, 17, 103-110.	1.0	35
51	Selective modulation of the androgen receptor AF2 domain rescues degeneration in spinal bulbar muscular atrophy. <i>Nature Medicine</i> , 2018, 24, 427-437.	15.2	35
52	Endoplasmic reticulum stress in spinal and bulbar muscular atrophy: a potential target for therapy. <i>Brain</i> , 2014, 137, 1894-1906.	3.7	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. <i>Advances in Neurobiology</i> , 2018, 20, 265-281.	1.3	27
54	Disease modifying effect of adiponectin in model of α -synucleinopathies. <i>Annals of Clinical and Translational Neurology</i> , 2014, 1, 479-489.	1.7	25

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55	CCP1 promotes mitochondrial fusion and motility to prevent Purkinje cell neuron loss in <i>pcd</i> mice. <i>Journal of Cell Biology</i> , 2019, 218, 206-219.	2.3	25
56	Efficient recombination-based methods for bacterial artificial chromosome fusion and mutagenesis. <i>Gene</i> , 2006, 371, 136-143.	1.0	24
57	Gene expression analysis reveals early dysregulation of disease pathways and links Chmp7 to pathogenesis of spinal and bulbar muscular atrophy. <i>Scientific Reports</i> , 2019, 9, 3539.	1.6	24
58	Proteolytic cleavage of ataxin-7 promotes SCA7 retinal degeneration and neurological dysfunction. <i>Human Molecular Genetics</i> , 2015, 24, 3908-3917.	1.4	22
59	Targeting protein aggregation in neurodegeneration – lessons from polyglutamine disorders. <i>Expert Opinion on Therapeutic Targets</i> , 2006, 10, 505-513.	1.5	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). <i>Genetics in Medicine</i> , 2020, 22, 681-685.	1.1	20
61	Gene therapy with AR isoform 2 rescues spinal and bulbar muscular atrophy phenotype by modulating AR transcriptional activity. <i>Science Advances</i> , 2021, 7, .	4.7	20
62	Low-Cost Gait Analysis for Behavioral Phenotyping of Mouse Models of Neuromuscular Disease. <i>Journal of Visualized Experiments</i> , 2019, , .	0.2	19
63	Unwinding the role of senataxin in neurodegeneration. <i>Discovery Medicine</i> , 2015, 19, 127-36.	0.5	19
64	Harmony Lost: Cell-Cell Communication at the Neuromuscular Junction in Motor Neuron Disease. <i>Trends in Neurosciences</i> , 2020, 43, 709-724.	4.2	17
65	Nemo-like kinase is a novel regulator of spinal and bulbar muscular atrophy. <i>ELife</i> , 2015, 4, e08493.	2.8	16
66	X-Linked Spinal and Bulbar Muscular Atrophy: From Clinical Genetic Features and Molecular Pathology to Mechanisms Underlying Disease Toxicity. <i>Advances in Experimental Medicine and Biology</i> , 2018, 1049, 103-133.	0.8	15
67	The replicative lifespan-extending deletion of <i>SGF73</i> results in altered ribosomal gene expression in yeast. <i>Aging Cell</i> , 2017, 16, 785-796.	3.0	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. <i>Acta Neuropathologica Communications</i> , 2019, 7, 27.	2.4	14
69	SUMOylated Senataxin functions in genome stability, RNA degradation, and stress granule disassembly, and is linked with inherited ataxia and motor neuron disease. <i>Molecular Genetics & Genomic Medicine</i> , 2021, 9, e1745.	0.6	13
70	4E-BP1 Protects Neurons from Misfolded Protein Stress and Parkinson's Disease Toxicity by Inducing the Mitochondrial Unfolded Protein Response. <i>Journal of Neuroscience</i> , 2020, 40, 8734-8745.	1.7	12
71	Differential effects of various genetic mouse models of the mechanistic target of rapamycin complex I inhibition on heart failure. <i>GeroScience</i> , 2019, 41, 847-860.	2.1	10
72	Deterioration of muscle force and contractile characteristics are early pathological events in spinal and bulbar muscular atrophy mice. <i>DMM Disease Models and Mechanisms</i> , 2020, 13, .	1.2	8

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73	Respiratory dysfunction in a mouse model of spinocerebellar ataxia type 7. <i>DMM Disease Models and Mechanisms</i> , 2021, 14, .	1.2	7
74	Ataxin-3, DNA Damage Repair, and SCA3 Cerebellar Degeneration: On the Path to Parsimony?. <i>PLoS Genetics</i> , 2015, 11, e1004937.	1.5	6
75	Tight expression regulation of senataxin, linked to motor neuron disease and ataxia, is required to avert cell-cycle block and nucleolus disassembly. <i>Heliyon</i> , 2020, 6, e04165.	1.4	6
76	Mitochondrial dysfunction in iPSC-derived neurons of subjects with chronic mountain sickness. <i>Journal of Applied Physiology</i> , 2018, 125, 832-840.	1.2	5
77	Altered H3 histone acetylation impairs high-fidelity DNA repair to promote cerebellar degeneration in spinocerebellar ataxia type 7. <i>Cell Reports</i> , 2021, 37, 110062.	2.9	5
78	De novo pathogenic variant in SETX causes a rapidly progressive neurodegenerative disorder of early childhood-onset with severe axonal polyneuropathy. <i>Acta Neuropathologica Communications</i> , 2021, 9, 194.	2.4	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. <i>Rare Diseases (Austin, Tex)</i> , 2014, 2, e962402.	1.8	4
80	In vivo assessment of neurodegeneration in Spinocerebellar Ataxia type 7. <i>NeuroImage: Clinical</i> , 2021, 29, 102561.	1.4	4
81	The expanding world of stem cell modeling of Huntington's disease: creating tools with a promising future. <i>Genome Medicine</i> , 2012, 4, 68.	3.6	3
82	Something wicked this way comes: huntingtin. <i>Nature Neuroscience</i> , 2014, 17, 1014-1015.	7.1	3
83	Identification of the SCA21 disease gene: remaining challenges and promising opportunities. <i>Brain</i> , 2014, 137, 2626-2628.	3.7	1
84	Convergent Transcription at the Ataxin-7 Locus Produces dsRNA Fragments that are Processed by Dicer-1. <i>FASEB Journal</i> , 2012, 26, 747.4.	0.2	0