Albert R La Spada

List of Publications by Year
in descending order

84 papers	16,800 citations	76031 42 h-index	64407 83 g-index
137 all docs	137 docs citations	137 times ranked	31284 citing authors

#	Article	IF	Citations
1	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.	0.6	13
2	Respiratory dysfunction in a mouse model of spinocerebellar ataxia type 7. DMM Disease Models and Mechanisms, 2021, 14, .	1.2	7
3	Gene therapy with AR isoform 2 rescues spinal and bulbar muscular atrophy phenotype by modulating AR transcriptional activity. Science Advances, 2021, 7, .	4.7	20
4	In vivo assessment of neurodegeneration in Spinocerebellar Ataxia type 7. NeuroImage: Clinical, 2021, 29, 102561.	1.4	4
5	Altered H3 histone acetylation impairs high-fidelity DNA repair to promote cerebellar degeneration in spinocerebellar ataxia type 7. Cell Reports, 2021, 37, 110062.	2.9	5
6	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.	2.4	5
7	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.	1.1	20
8	Nicotinamide Pathway-Dependent Sirt1 Activation Restores Calcium Homeostasis to Achieve Neuroprotection in Spinocerebellar Ataxia Type 7. Neuron, 2020, 105, 630-644.e9.	3.8	63
9	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.	1.7	12
10	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.	1.4	6
11	Harmony Lost: Cellâ€“Cell Communication at the Neuromuscular Junction in Motor Neuron Disease. Trends in Neurosciences, 2020, 43, 709-724.	4.2	17
12	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, .	1.2	8
13	Autophagy gene haploinsufficiency drives chromosome instability, increases migration, and promotes early ovarian tumors. PLoS Genetics, 2020, 16, e1008558.	1.5	39
14	Reduced C9ORF72 function exacerbates gain of toxicity from ALS/FTD-causing repeat expansion in C9orf72. Nature Neuroscience, 2020, 23, 615-624.	7.1	157
15	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.	2.1	135
16	Low-Cost Gait Analysis for Behavioral Phenotyping of Mouse Models of Neuromuscular Disease. Journal of Visualized Experiments, 2019, , .	0.2	19
17	Differential effects of various genetic mouse models of the mechanistic target of rapamycin complex I inhibition on heart failure. GeroScience, 2019, 41, 847-860.	2.1	10
18	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.	2.9	49

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19	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.	1.6	24
20	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.	2.4	14
21	CCP1 promotes mitochondrial fusion and motility to prevent Purkinje cell neuron loss in pcd mice. Journal of Cell Biology, 2019, 218, 206-219.	2.3	25
22	Overriding FUS autoregulation in mice triggers gain-of-toxic dysfunctions in RNA metabolism and autophagy-lysosome axis. ELife, 2019, 8, .	2.8	65
23	Mutant huntingtin impairs PNKP and ATXN3, disrupting DNA repair and transcription. ELife, 2019, 8, .	2.8	83
24	MAP4K3 mediates amino acid-dependent regulation of autophagy via phosphorylation of TFEB. Nature Communications, 2018, 9, 942.	5.8	80
25	Selective modulation of the androgen receptor AF2 domain rescues degeneration in spinal bulbar muscular atrophy. Nature Medicine, 2018, 24, 427-437.	15.2	35
26	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.	0.8	15
27	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.	1.0	67
28	Fasting Activates Fatty Acid Oxidation to Enhance Intestinal Stem Cell Function during Homeostasis and Aging. Cell Stem Cell, 2018, 22, 769-778.e4.	5.2	266
29	Therapy development in Huntington disease: From current strategies to emerging opportunities. American Journal of Medical Genetics, Part A, 2018, 176, 842-861.	0.7	75
30	Converging pathways in neurodegeneration, from genetics to mechanisms. Nature Neuroscience, 2018, 21, 1300-1309.	7.1	325
31	Antisense oligonucleotides targeting mutant Ataxin-7 restore visual function in a mouse model of spinocerebellar ataxia type 7. Science Translational Medicine, 2018, 10, .	5.8	63
32	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.	1.3	27
33	Senataxin mutations elicit motor neuron degeneration phenotypes and yield TDP-43 mislocalization in ALS4 mice and human patients. Acta Neuropathologica, 2018, 136, 425-443.	3.9	43
34	Mitochondrial dysfunction in iPSC-derived neurons of subjects with chronic mountain sickness. Journal of Applied Physiology, 2018, 125, 832-840.	1.2	5
35	The replicative lifespanâ€extending deletion of SGF73 results in altered ribosomal gene expression in yeast. Aging Cell, 2017, 16, 785-796.	3.0	14
36	Transcriptional regulation of core autophagy and lysosomal genes by the androgen receptor promotes prostate cancer progression. Autophagy, 2017, 13, 506-521.	4.3	88

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37	PPARÎ´ activation by bexarotene promotes neuroprotection by restoring bioenergetic and quality control homeostasis. Science Translational Medicine, 2017, 9, .	5.8	54
38	Adenylyl cyclase activating polypeptide reduces phosphorylation and toxicity of the polyglutamine-expanded androgen receptor in spinobulbar muscular atrophy. Science Translational Medicine, 2016, 8, 370ra181.	5.8	37
39	Increased 4E-BP1 Expression Protects against Diet-Induced Obesity and Insulin Resistance in Male Mice. Cell Reports, 2016, 16, 1903-1914.	2.9	52
40	Guidelines for the use and interpretation of assays for monitoring autophagy (3rd edition). Autophagy, 2016, 12, 1-222.	4.3	4,701
41	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.	15.2	88
42	Ataxin-3, DNA Damage Repair, and SCA3 Cerebellar Degeneration: On the Path to Parsimony?. PLoS Genetics, 2015, 11, e1004937.	1.5	6
43	Autophagy in polyglutamine disease: Imposing order on disorder or contributing to the chaos?. Molecular and Cellular Neurosciences, 2015, 66, 53-61.	1.0	47
44	Proteolytic cleavage of ataxin-7 promotes SCA7 retinal degeneration and neurological dysfunction. Human Molecular Genetics, 2015, 24, 3908-3917.	1.4	22
45	Nemo-like kinase is a novel regulator of spinal and bulbar muscular atrophy. ELife, 2015, 4, e08493.	2.8	16
46	Unwinding the role of senataxin in neurodegeneration. Discovery Medicine, 2015, 19, 127-36.	0.5	19
47	Identification of the SCA21 disease gene: remaining challenges and promising opportunities. Brain, 2014, 137, 2626-2628.	3.7	1
48	Nonallele Specific Silencing of Ataxin-7 Improves Disease Phenotypes in a Mouse Model of SCA7. Molecular Therapy, 2014, 22, 1635-1642.	3.7	51
49	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
50	The many faces of autophagy dysfunction in Huntington's disease: from mechanism to therapy. Drug Discovery Today, 2014, 19, 963-971.	3.2	112
51	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.	3.8	150
52	Disease modifying effect of adiponectin in model of Î± â€synucleinopathies. Annals of Clinical and Translational Neurology, 2014, 1, 479-489.	1.7	25
53	Endoplasmic reticulum stress in spinal and bulbar muscular atrophy: a potential target for therapy. Brain, 2014, 137, 1894-1906.	3.7	31
54	Polyglutamine-expanded androgen receptor interferes with TFEB to elicit autophagy defects in SBMA. Nature Neuroscience, 2014, 17, 1180-1189.	7.1	142

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55	Something wicked this way comes: huntingtin. Nature Neuroscience, 2014, 17, 1014-1015.	7.1	3
56	The SAGA Histone Deubiquitinase Module Controls Yeast Replicative Lifespan via Sir2 Interaction. Cell Reports, 2014, 8, 477-486.	2.9	62
57	Let-7 Coordinately Suppresses Components of the Amino Acid Sensing Pathway to Repress mTORC1 and Induce Autophagy. Cell Metabolism, 2014, 20, 626-638.	7.2	67
58	Peripheral Androgen Receptor Gene Suppression Rescues Disease in Mouse Models of Spinal and Bulbar Muscular Atrophy. Cell Reports, 2014, 7, 774-784.	2.9	148
59	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.	2.5	104
60	A functional deficiency of TERA/VCP/p97 contributes to impaired DNA repair in multiple polyglutamine diseases. Nature Communications, 2013, 4, 1816.	5.8	60
61	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.	1.4	42
62	PGC-1Î± Rescues Huntingtonâ€™s Disease Proteotoxicity by Preventing Oxidative Stress and Promoting TFEB Function. Science Translational Medicine, 2012, 4, 142ra97.	5.8	376
63	The expanding world of stem cell modeling of Huntington's disease: creating tools with a promising future. Genome Medicine, 2012, 4, 68.	3.6	3
64	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.	2.8	106
65	PPARGC1A/PGC-1Î±, TFEB and enhanced proteostasis in Huntington disease. Autophagy, 2012, 8, 1845-1847.	4.3	44
66	Guidelines for the use and interpretation of assays for monitoring autophagy. Autophagy, 2012, 8, 445-544.	4.3	3,122
67	Convergent Transcription at the Ataxinâ€7 Locus Produces dsRNA Fragments that are Processed by Dicerâ€1. FASEB Journal, 2012, 26, 747.4.	0.2	0
68	Mitochondrial autophagy in neural function, neurodegenerative disease, neuron cell death, and aging. Neurobiology of Disease, 2011, 43, 46-51.	2.1	119
69	A Simple Composite Phenotype Scoring System for Evaluating Mouse Models of Cerebellar Ataxia. Journal of Visualized Experiments, 2010, , .	0.2	253
70	Repeat expansion disease: progress and puzzles in disease pathogenesis. Nature Reviews Genetics, 2010, 11, 247-258.	7.7	425
71	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.	3.8	40
72	Polyglutamine-Expanded Androgen Receptor Truncation Fragments Activate a Bax-Dependent Apoptotic Cascade Mediated by DP5/Hrk. Journal of Neuroscience, 2009, 29, 1987-1997.	1.7	56

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73	Nutrient Deprivation Induces Neuronal Autophagy and Implicates Reduced Insulin Signaling in Neuroprotective Autophagy Activation. Journal of Biological Chemistry, 2009, 284, 2363-2373.	1.6	107
74	Autophagy activation and enhanced mitophagy characterize the Purkinje cells of pcd mice prior to neuronal death. Molecular Brain, 2009, 2, 24.	1.3	95
75	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.	0.7	36
76	Targeting protein aggregation in neurodegeneration â€“ lessons from polyglutamine disorders. Expert Opinion on Therapeutic Targets, 2006, 10, 505-513.	1.5	21
77	Thermoregulatory and metabolic defects in Huntington's disease transgenic mice implicate PGC-1Î± in Huntington's disease neurodegeneration. Cell Metabolism, 2006, 4, 349-362.	7.2	519
78	Efficient recombination-based methods for bacterial artificial chromosome fusion and mutagenesis. Gene, 2006, 371, 136-143.	1.0	24
79	The Purkinje cell degeneration 5J mutation is a single amino acid insertion that destabilizes Nna1 protein. Mammalian Genome, 2006, 17, 103-110.	1.0	35
80	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.	3.3	215
81	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.	1.4	82
82	Purkinje cell degeneration (pcd) Phenotypes Caused by Mutations in the Axotomy-Induced Gene, Nna1. Science, 2002, 295, 1904-1906.	6.0	217
83	Polyglutamine-Expanded Ataxin-7 Antagonizes CRX Function and Induces Cone-Rod Dystrophy in a Mouse Model of SCA7. Neuron, 2001, 31, 913-927.	3.8	244
84	Androgen receptor gene mutations in X-linked spinal and bulbar muscular atrophy. Nature, 1991, 352, 77-79.	13.7	2,710