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List of Publications by Year
in descending order

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39 papers	2,910 citations	304743 22 h-index	330143 37 g-index
40 all docs	40 docs citations	40 times ranked	3672 citing authors

#	Article	IF	Citations
1	Comprehensive In Silico Analysis of Retrotransposon Insertions within the Survival Motor Neuron Genes Involved in Spinal Muscular Atrophy. Biology, 2022, 11, 824.	2.8	1
2	Detection of SMN1 to SMN2 gene conversion events and partial SMN1 gene deletions using array digital PCR. Neurogenetics, 2021, 22, 53-64.	1.4	14
3	Genomic Variability in the Survival Motor Neuron Genes (SMN1 and SMN2): Implications for Spinal Muscular Atrophy Phenotype and Therapeutics Development. International Journal of Molecular Sciences, 2021, 22, 7896.	4.1	35
4	Development and validation of a 4-color multiplexing spinal muscular atrophy (SMA) genotyping assay on a novel integrated digital PCR instrument. Scientific Reports, 2020, 10, 19892.	3.3	17
5	Spinal muscular atrophy diagnosis and carrier screening from genome sequencing data. Genetics in Medicine, 2020, 22, 945-953.	2.4	78
6	Using Systems Biology and Mathematical Modeling Approaches in the Discovery of Therapeutic Targets for Spinal Muscular Atrophy. Advances in Neurobiology, 2018, 21, 267-281.	1.8	0
7	Establishing a reference dataset for the authentication of spinal muscular atrophy cell lines using STR profiling and digital PCR. Neuromuscular Disorders, 2017, 27, 439-446.	0.6	15
8	The effects of C5-substituted 2,4-diaminoquinazolines on selected transcript expression in spinal muscular atrophy cells. PLoS ONE, 2017, 12, e0180657.	2.5	4
9	Effect of the Butyrate Prodrug Pivaloyloxymethyl Butyrate (AN9) on a Mouse Model for Spinal Muscular Atrophy. Journal of Neuromuscular Diseases, 2016, 3, 511-515.	2.6	5
10	Copy Number Variations in the Survival Motor Neuron Genes: Implications for Spinal Muscular Atrophy and Other Neurodegenerative Diseases. Frontiers in Molecular Biosciences, 2016, 3, 7.	3.5	124
11	Identification of early gene expression changes in primary cultured neurons treated with topoisomerase I poisons. Biochemical and Biophysical Research Communications, 2016, 479, 319-324.	2.1	4
12	Applicability of digital PCR to the investigation of pediatric-onset genetic disorders. Biomolecular Detection and Quantification, 2016, 10, 9-14.	7.0	17
13	Protective effects of butyrate-based compounds on a mouse model for spinal muscular atrophy. Experimental Neurology, 2016, 279, 13-26.	4.1	25
14	SMN1 and SMN2 copy numbers in cell lines derived from patients with spinal muscular atrophy as measured by array digital PCR. Molecular Genetics & Genomic Medicine, 2015, 3, 248-257.	1.2	56
15	The effect of the DcpS inhibitor D156844 on the protective action of follistatin in mice with spinal muscular atrophy. Neuromuscular Disorders, 2015, 25, 699-705.	0.6	12
16	Transcriptome Profiling of Spinal Muscular Atrophy Motor Neurons Derived from Mouse Embryonic Stem Cells. PLoS ONE, 2014, 9, e106818.	2.5	37
17	Systems Biology Investigation of cAMP Modulation to Increase SMN Levels for the Treatment of Spinal Muscular Atrophy. PLoS ONE, 2014, 9, e115473.	2.5	14
18	The effect of diet on the protective action of D156844 observed in spinal muscular atrophy mice. Experimental Neurology, 2014, 256, 1-6.	4.1	15

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19	NF-ÃŽÂºBÃ¢â‚¬â„¢mediated Pax7 dysregulation in the muscle microenvironment promotes cancer cachexia. Journal of Clinical Investigation, 2013, 123, 4821-4835.	8.2	293
20	Trans-Splicing, More Than Meets the Eye: Multifaceted Therapeutics for Spinal Muscular Atrophy. Human Gene Therapy, 2011, 22, 121-125.	2.7	1
21	Effects of 2,4-diaminoquinazoline derivatives on SMN expression and phenotype in a mouse model for spinal muscular atrophy. Human Molecular Genetics, 2010, 19, 454-467.	2.9	110
22	Effect of diet on the survival and phenotype of a mouse model for spinal muscular atrophy. Biochemical and Biophysical Research Communications, 2010, 391, 835-840.	2.1	52
23	Detection of human survival motor neuron (SMN) protein in mice containing the SMN2 transgene: Applicability to preclinical therapy development for spinal muscular atrophy. Journal of Neuroscience Methods, 2008, 175, 36-43.	2.5	16
24	Synthesis and Biological Evaluation of Novel 2,4-Diaminoquinazoline Derivatives as SMN2 Promoter Activators for the Potential Treatment of Spinal Muscular Atrophy. Journal of Medicinal Chemistry, 2008, 51, 449-469.	6.4	88
25	Protein phosphatase 1 binds to the RNA recognition motif of several splicing factors and regulates alternative pre-mRNA processing. Human Molecular Genetics, 2008, 17, 52-70.	2.9	76
26	Let all DNA vote. Neurology, 2008, 70, 662-663.	1.1	3
27	Translational Control of Glial Glutamate Transporter EAAT2 Expression. Journal of Biological Chemistry, 2007, 282, 1727-1737.	3.4	75
28	Ribonucleoprotein Assembly Defects Correlate with Spinal Muscular Atrophy Severity and Preferentially Affect a Subset of Spliceosomal snRNPs. PLoS ONE, 2007, 2, e921.	2.5	266
29	A novel method for oral delivery of drug compounds to the neonatal SMNÎ”7 mouse model of spinal muscular atrophy. Journal of Neuroscience Methods, 2007, 161, 285-290.	2.5	37
30	Abnormal motor phenotype in the SMNÎ”7 mouse model of spinal muscular atrophy. Neurobiology of Disease, 2007, 27, 207-219.	4.4	96
31	Dystrophin glycoprotein complex dysfunction: A regulatory link between muscular dystrophy and cancer cachexia. Cancer Cell, 2005, 8, 421-432.	16.8	260
32	SMNÎ”7, the major product of the centromeric survival motor neuron (SMN2) gene, extends survival in mice with spinal muscular atrophy and associates with full-length SMN. Human Molecular Genetics, 2005, 14, 845-857.	2.9	550
33	Association of Excitatory Amino Acid Transporters, Especially EAAT2, with Cholesterol-rich Lipid Raft Microdomains. Journal of Biological Chemistry, 2004, 279, 34388-34396.	3.4	146
34	Perspectives on models of spinal muscular atrophy for drug discovery. Drug Discovery Today: Disease Models, 2004, 1, 151-156.	1.2	13
35	Methyl-Î²-cyclodextrin but not retinoic acid reduces EAAT3-mediated glutamate uptake and increases GTRAP3-18 expression. Journal of Neurochemistry, 2003, 84, 891-894.	3.9	30
36	Increased expression of the glial glutamate transporter EAAT2 modulates excitotoxicity and delays the onset but not the outcome of ALS in mice. Human Molecular Genetics, 2003, 12, 2519-2532.	2.9	235

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37	Human Glioma Cells and Undifferentiated Primary Astrocytes That Express Aberrant EAAT2 mRNA Inhibit Normal EAAT2 Protein Expression and Prevent Cell Death. Molecular and Cellular Neurosciences, 2002, 21, 546-560.	2.2	39
38	Molecular cloning, gene structure, expression profile and functional characterization of the mouse glutamate transporter (EAAT3) interacting protein GTRAP3â€“18. Gene, 2002, 292, 81-90.	2.2	51
39	Effects of inhibitors of SLC9A-type sodium-proton exchangers on Survival Motor Neuron2 (SMN2) mRNA splicing and expression. Molecular Pharmacology, 0, , MOLPHARM-AR-2022-000529.	2.3	0