Alex Mackenzie

List of Publications by Year
in descending order

42 papers	3,491 citations	516710 16 h-index	315739 38 g-index
42 all docs	42 docs citations	42 times ranked	3599 citing authors

#	Article	IF	Citations
1	Suppression of apoptosis in mammalian cells by NAIP and a related family of IAP genes. Nature, 1996, 379, 349-353.	27.8	982
2	The gene for neuronal apoptosis inhibitory protein is partially deleted in individuals with spinal muscular atrophy. Cell, 1995, 80, 167-178.	28.9	969
3	Mutations in GPC3, a glypican gene, cause the Simpson-Golabi-Behmel overgrowth syndrome. Nature Genetics, 1996, 12, 241-247.	21.4	732
4	DNM1L-related mitochondrial fission defect presenting as refractory epilepsy. European Journal of Human Genetics, 2016, 24, 1084-1088.	2.8	113
5	Pyridoxine-Dependent Epilepsy in Zebrafish Caused by Aldh7a1 Deficiency. Genetics, 2017, 207, 1501-1518.	2.9	81
6	NAIP protects the nigrostriatal dopamine pathway in an intrastriatal 6â€ŌHDA rat model of Parkinson's disease. European Journal of Neuroscience, 2001, 14, 391-400.	2.6	72
7	Celecoxib increases SMN and survival in a severe spinal muscular atrophy mouse model via p38 pathway activation. Human Molecular Genetics, 2013, 22, 3415-3424.	2.9	52
8	Neuronal apoptosis inhibitory protein, NAIP, is an inhibitor of procaspase-9. International Journal of Biochemistry and Cell Biology, 2010, 42, 958-964.	2.8	48
9	Mouse lysine catabolism to aminoadipate occurs primarily through the saccharopine pathway; implications for pyridoxine dependent epilepsy (PDE). Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2017, 1863, 121-128.	3.8	43
10	Molecular Analysis of Cystinosis: Probable Irish Origin of the Most Common French Canadian Mutation. European Journal of Human Genetics, 1999, 7, 671-678.	2.8	35
11	A Review of the Current Impact of Inhibitors of Apoptosis Proteins and Their Repression in Cancer. Cancers, 2022, 14, 1671.	3.7	32
12	Inhibitor of apoptosis proteins, NAIP, cIAP1 and cIAP2 expression during macrophage differentiation and M1/M2 polarization. PLoS ONE, 2018, 13, e0193643.	2.5	27
13	Current knowledge for pyridoxine-dependent epilepsy: a 2016 update. Expert Review of Endocrinology and Metabolism, 2017, 12, 5-20.	2.4	26
14	Neuronal apoptosis inhibitory protein (NAIP) localizes to the cytokinetic machinery during cell division. Scientific Reports, 2017, 7, 39981.	3.3	23
15	Direct health-care costs for children diagnosed with genetic diseases are significantly higher than for children with other chronic diseases. Genetics in Medicine, 2019, 21, 1049-1057.	2.4	20
16	Newborn Screening for Spinal Muscular Atrophy: Ontario Testing and Follow-up Recommendations. Canadian Journal of Neurological Sciences, 2021, 48, 504-511.	0.5	18
17	Genetic linkage analysis of Canadian spinal muscular atrophy kindreds using flanking microsatellite 5q13 polymorphisms. Human Genetics, 1993, 90, 501-4.	3.8	16
18	SMA genes: deleted and duplicated. Nature Genetics, 1995, 9, 112-113.	21.4	16

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19	DLG4-related synaptopathy: a new rare brain disorder. Genetics in Medicine, 2021, 23, 888-899.	2.4	16
20	Core Outcome Sets for Medium-Chain Acyl-CoA Dehydrogenase Deficiency and Phenylketonuria. Pediatrics, 2021, 148, .	2.1	16
21	High-throughput DNA Extraction and Genotyping of 3dpf Zebrafish Larvae by Fin Clipping. Journal of Visualized Experiments, 2018, , .	0.3	15
22	Outcomes in pediatric studies of medium-chain acyl-coA dehydrogenase (MCAD) deficiency and phenylketonuria (PKU): a review. Orphanet Journal of Rare Diseases, 2020, 15, 12.	2.7	15
23	Sense in Antisense Therapy for Spinal Muscular Atrophy. New England Journal of Medicine, 2012, 366, 761-763.	27.0	14
24	Mining the transcriptome for rare disease therapies: a comparison of the efficiencies of two data mining approaches and a targeted cell-based drug screen. Npj Genomic Medicine, 2017, 2, 14.	3.8	14
25	VPAC2 receptor agonist BAY 55-9837 increases SMN protein levels and moderates disease phenotype in severe spinal muscular atrophy mouse models. Orphanet Journal of Rare Diseases, 2014, 9, 4.	2.7	13
26	cDNA cloning and the 5'genomic organization of Naip2, a candidate gene for murine Legionella resistance. Mammalian Genome, 1999, 10, 761-763.	2.2	11
27	The Canadian Neuromuscular Disease Registry: Connecting patients to national and international research opportunities. Paediatrics and Child Health, 2018, 23, 20-26.	0.6	11
28	Transcriptomic RNAseq drug screen in cerebrocortical cultures: toward novel neurogenetic disease therapies. Human Molecular Genetics, 2018, 27, 3206-3217.	2.9	11
29	Establishing core outcome sets for phenylketonuria (PKU) and medium-chain Acyl-CoA dehydrogenase (MCAD) deficiency in children: study protocol for systematic reviews and Delphi surveys. Trials, 2017, 18, 603.	1.6	9
30	Sodium Channel Inhibitors Reduce DMPK mRNA and Protein. Clinical and Translational Science, 2015, 8, 298-304.	3.1	8
31	Differential regulation of autophagy by STAU1 in alveolar rhabdomyosarcoma and nonâ€transformed skeletal muscle cells. Cellular Oncology (Dordrecht), 2021, 44, 851-870.	4.4	7
32	Ontario Newborn Screening for Spinal Muscular Atrophy: The First Year. Canadian Journal of Neurological Sciences, 2022, 49, 821-823.	0.5	7
33	A National Spinal Muscular Atrophy Registry for Real-World Evidence. Canadian Journal of Neurological Sciences, 2020, 47, 810-815.	0.5	6
34	Human Growth Hormone Increases SMN Expression and Survival in Severe Spinal Muscular Atrophy Mouse Model. Journal of Neuromuscular Diseases, 2014, 1, 65-74.	2.6	5
35	High-throughput kinome-RNAi screen identifies protein kinase R activator (PACT) as a novel genetic modifier of CUG foci integrity in myotonic dystrophy type 1 (DM1). PLoS ONE, 2021, 16, e0256276.	2.5	3
36	Human Growth Hormone Increases SMN Expression and Survival in Severe Spinal Muscular Atrophy Mouse Model. Journal of Neuromuscular Diseases, 2014, 1, 65-74.	2.6	3

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37	Pharmacologic normalization of pathogenic dosage underlying genetic diseases: an overview of the literature and path forward. Emerging Topics in Life Sciences, 2019, 3, 53-62.	2.6	1
38	Separating the Wheat from the Chaff: The Use of Upstream Regulator Analysis to Identify True Differential Expression of Single Genes within Transcriptomic Datasets. International Journal of Molecular Sciences, 2021, 22, 6295.	4.1	1
39	The Search for Pompe Patients in Canada: Assessing Feasibility of a National Disease Registry to Facilitate Research. Journal of Neuromuscular Diseases, 2015, 2, S45-S46.	2.6	0
40	The determination of purine crystal structures: an overlooked prequel to the discovery of the double helix. Genome, 2019, 62, 43-44.	2.0	0
41	NAIP expression increases in a rat model of liver mass restoration. Journal of Molecular Histology, 2021, 52, 113-123.	2.2	0
42	The Search for Pompe Patients in Canada: Assessing Feasibility of a National Disease Registry to Facilitate Research. Journal of Neuromuscular Diseases, 2015, 2, S45-S46.	2.6	0