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	Ontario Newborn Screening for Spinal Muscular Atrophy: The First Year. Canadian Journal of Neurological Sciences, 2022, 49, 821-823.	0.5	7
2	A Review of the Current Impact of Inhibitors of Apoptosis Proteins and Their Repression in Cancer. Cancers, 2022, 14, 1671.	3.7	32
3	Newborn Screening for Spinal Muscular Atrophy: Ontario Testing and Follow-up Recommendations. Canadian Journal of Neurological Sciences, 2021, 48, 504-511.	0.5	18
4	NAIP expression increases in a rat model of liver mass restoration. Journal of Molecular Histology, 2021, 52, 113-123.	2.2	0
5	DLG4-related synaptopathy: a new rare brain disorder. Genetics in Medicine, 2021, 23, 888-899.	2.4	16
6	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
7	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
8	Core Outcome Sets for Medium-Chain Acyl-CoA Dehydrogenase Deficiency and Phenylketonuria. Pediatrics, 2021, 148, .	2.1	16
9	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
10	A National Spinal Muscular Atrophy Registry for Real-World Evidence. Canadian Journal of Neurological Sciences, 2020, 47, 810-815.	0.5	6
11	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
12	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
13	The determination of purine crystal structures: an overlooked prequel to the discovery of the double helix. Genome, 2019, 62, 43-44.	2.0	0
14	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
15	The Canadian Neuromuscular Disease Registry: Connecting patients to national and international research opportunities. Paediatrics and Child Health, 2018, 23, 20-26.	0.6	11
16	High-throughput DNA Extraction and Genotyping of 3dpf Zebrafish Larvae by Fin Clipping. Journal of Visualized Experiments, 2018, , .	0.3	15
17	Transcriptomic RNAseq drug screen in cerebrocortical cultures: toward novel neurogenetic disease therapies. Human Molecular Genetics, 2018, 27, 3206-3217.	2.9	11
18	Inhibitor of apoptosis proteins, NAIP, cIAP1 and cIAP2 expression during macrophage differentiation and M1/M2 polarization. PLoS ONE, 2018, 13, e0193643.	2.5	27

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19	Neuronal apoptosis inhibitory protein (NAIP) localizes to the cytokinetic machinery during cell division. Scientific Reports, 2017, 7, 39981.	3.3	23
20	Current knowledge for pyridoxine-dependent epilepsy: a 2016 update. Expert Review of Endocrinology and Metabolism, 2017, 12, 5-20.	2.4	26
21	Pyridoxine-Dependent Epilepsy in Zebrafish Caused by Aldh7a1 Deficiency. Genetics, 2017, 207, 1501-1518.	2.9	81
22	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
23	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
24	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
25	DNM1L-related mitochondrial fission defect presenting as refractory epilepsy. European Journal of Human Genetics, 2016, 24, 1084-1088.	2.8	113
26	Sodium Channel Inhibitors Reduce DMPK mRNA and Protein. Clinical and Translational Science, 2015, 8, 298-304.	3.1	8
27	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
28	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
29	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
30	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
31	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
32	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
33	Sense in Antisense Therapy for Spinal Muscular Atrophy. New England Journal of Medicine, 2012, 366, 761-763.	27.0	14
34	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
35	NAIP protects the nigrostriatal dopamine pathway in an intrastriatal 6â€OHDA rat model of Parkinson's disease. European Journal of Neuroscience, 2001, 14, 391-400.	2.6	72
36	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

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37	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
38	Mutations in GPC3, a glypican gene, cause the Simpson-Golabi-Behmel overgrowth syndrome. Nature Genetics, 1996, 12, 241-247.	21.4	732
39	Suppression of apoptosis in mammalian cells by NAIP and a related family of IAP genes. Nature, 1996, 379, 349-353.	27.8	982
40	SMA genes: deleted and duplicated. Nature Genetics, 1995, 9, 112-113.	21.4	16
41	The gene for neuronal apoptosis inhibitory protein is partially deleted in individuals with spinal muscular atrophy. Cell, 1995, 80, 167-178.	28.9	969
42	Genetic linkage analysis of Canadian spinal muscular atrophy kindreds using flanking microsatellite 5q13 polymorphisms. Human Genetics, 1993, 90, 501-4.	3.8	16