Dennis Bartholomew

List of Publications by Year
in descending order

13	556	933447 10	1125743 13
papers	citations	h-index	g-index

13	13	13	1240
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Inherited and de novo variants extend the etiology of TAOK1-associated neurodevelopmental disorder. Journal of Physical Education and Sports Management, 2022, , mcs.a006180.	1.2	6
2	Pathogenic variants in SMARCA5 , a chromatin remodeler, cause a range of syndromic neurodevelopmental features. Science Advances, 2021, 7, .	10.3	17
3	Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor Î² Signaling. Biological Psychiatry, 2020, 87, 100-112.	1.3	42
4	Long-term safety and efficacy of glycerol phenylbutyrate for the management of urea cycle disorder patients. Molecular Genetics and Metabolism, 2019, 127, 336-345.	1.1	10
5	Diagnostic Utility of Whole Exome Sequencing in the Neuromuscular Clinic. Neuropediatrics, 2019, 50, 096-102.	0.6	28
6	Three additional patients with EED-associated overgrowth: potential mutation hotspots identified?. Journal of Human Genetics, 2019, 64, 561-572.	2.3	16
7	Phenylalanine and tyrosine measurements across gestation by tandem mass spectrometer on dried blood spot cards from normal pregnant women. Genetics in Medicine, 2019, 21, 1821-1826.	2.4	7
8	Clinically severe CACNA1A alleles affect synaptic function and neurodegeneration differentially. PLoS Genetics, 2017, 13, e1006905.	3.5	80
9	Clinical course of sly syndrome (mucopolysaccharidosis type VII). Journal of Medical Genetics, 2016, 53, 403-418.	3.2	133
10	Self-reported treatment-associated symptoms among patients with urea cycle disorders participating in glycerol phenylbutyrate clinical trials. Molecular Genetics and Metabolism, 2015, 116, 29-34.	1.1	12
11	Blood ammonia and glutamine as predictors of hyperammonemic crises in patients with urea cycle disorder. Genetics in Medicine, 2015, 17, 561-568.	2.4	30
12	Mutations in PURA Cause Profound Neonatal Hypotonia, Seizures, and Encephalopathy in 5q31.3 Microdeletion Syndrome. American Journal of Human Genetics, 2014, 95, 579-583.	6.2	92
13	Ammonia control and neurocognitive outcome among urea cycle disorder patients treated with glycerol phenylbutyrate. Hepatology, 2013, 57, 2171-2179.	7.3	83