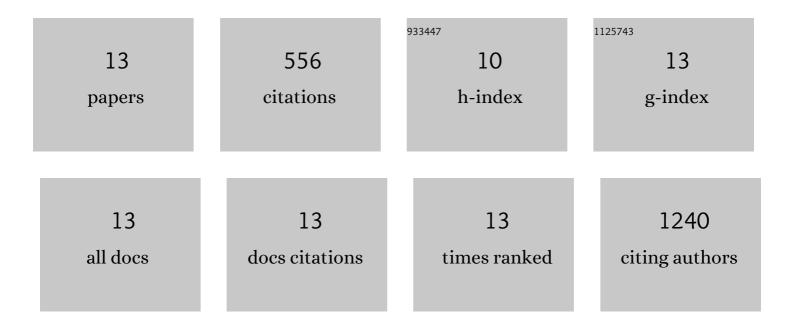
Dennis Bartholomew

List of Publications by Year in descending order

Source: https://exaly.com/author-pdf/4297111/publications.pdf Version: 2024-02-01



| # | Article | IF | CITATIONS |
|----|---|------|-----------|
| 1 | Clinical course of sly syndrome (mucopolysaccharidosis type VII). Journal of Medical Genetics, 2016, 53, 403-418. | 3.2 | 133 |
| 2 | 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 |
| 3 | Ammonia control and neurocognitive outcome among urea cycle disorder patients treated with glycerol phenylbutyrate. Hepatology, 2013, 57, 2171-2179. | 7.3 | 83 |
| 4 | Clinically severe CACNA1A alleles affect synaptic function and neurodegeneration differentially. PLoS Genetics, 2017, 13, e1006905. | 3.5 | 80 |
| 5 | Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor β Signaling. Biological Psychiatry, 2020, 87, 100-112. | 1.3 | 42 |
| 6 | 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 |
| 7 | Diagnostic Utility of Whole Exome Sequencing in the Neuromuscular Clinic. Neuropediatrics, 2019, 50, 096-102. | 0.6 | 28 |
| 8 | Pathogenic variants in <i>SMARCA5</i> , a chromatin remodeler, cause a range of syndromic neurodevelopmental features. Science Advances, 2021, 7, . | 10.3 | 17 |
| 9 | Three additional patients with EED-associated overgrowth: potential mutation hotspots identified?. Journal of Human Genetics, 2019, 64, 561-572. | 2.3 | 16 |
| 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 | 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 |
| 12 | 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 |
| 13 | 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 |