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List of Publications by Year
in descending order

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Version: 2024-02-01

26 papers	983 citations	840119 11 h-index	610482 24 g-index
28 all docs	28 docs citations	28 times ranked	2539 citing authors

#	Article	IF	Citations
1	De novo variants in ATP2B1 lead to neurodevelopmental delay. American Journal of Human Genetics, 2022, 109, 944-952.	2.6	11
2	Genome-wide sequencing and the clinical diagnosis of genetic disease: The CAUSES study. Human Genetics and Genomics Advances, 2022, 3, 100108.	1.0	7
3	Development of minimally invasive 13C-glucose breath test to examine different exogenous carbohydrate sources in patients with glycogen storage disease type Ia. Molecular Genetics and Metabolism Reports, 2022, 31, 100880.	0.4	2
4	Hyperleucinosis during infections in maple syrup urine disease post liver transplantation. Molecular Genetics and Metabolism Reports, 2021, 27, 100763.	0.4	3
5	C2orf69 mutations disrupt mitochondrial function and cause a multisystem human disorder with recurring autoinflammation. Journal of Clinical Investigation, 2021, 131, .	3.9	13
6	NANS-CDG: Delineation of the Genetic, Biochemical, and Clinical Spectrum. Frontiers in Neurology, 2021, 12, 668640.	1.1	11
7	Development of Minimally Invasive 13C-Glucose Breath Test to Examine Different Dietary Therapies in Patients with Glycogen Storage Disorders. Current Developments in Nutrition, 2020, 4, nzaa055_034.	0.1	0
8	Clinical and biochemical improvement with galactose supplementation in SLC35A2-CDG. Genetics in Medicine, 2020, 22, 1102-1107.	1.1	42
9	Evaluation of the quality of clinical data collection for a pan-Canadian cohort of children affected by inherited metabolic diseases: lessons learned from the Canadian Inherited Metabolic Diseases Research Network. Orphanet Journal of Rare Diseases, 2020, 15, 89.	1.2	11
10	Vitreous Changes in Gaucher Disease Type 3. Ophthalmology, 2020, 127, 813.	2.5	0
11	Diagnostic Yield and Treatment Impact of Targeted Exome Sequencing in Early-Onset Epilepsy. Frontiers in Neurology, 2019, 10, 434.	1.1	70
12	Clinical, neuroradiological, and biochemical features of SLC35A2â€CDG patients. Journal of Inherited Metabolic Disease, 2019, 42, 553-564.	1.7	32
13	Atypical cerebral palsy: genomics analysis enables precision medicine. Genetics in Medicine, 2019, 21, 1621-1628.	1.1	47
14	The genotypic and phenotypic spectrum of MTO1 deficiency. Molecular Genetics and Metabolism, 2018, 123, 28-42.	0.5	24
15	The Indicator Amino Acid Oxidation Method with the Use of L-[1-13C]Leucine Suggests a Higher than Currently Recommended Protein Requirement in Children with Phenylketonuria. Journal of Nutrition, 2017, 147, 211-217.	1.3	5
16	Case Report. Journal of Child Neurology, 2017, 32, 403-407.	0.7	5
17	Patient care standards for primary mitochondrial disease: a consensus statement from the Mitochondrial Medicine Society. Genetics in Medicine, 2017, 19, 1380-1397.	1.1	173
18	Response to Newman et al.. Genetics in Medicine, 2017, 19, 1380-1380.	1.1	3

#	Article	IF	Citations
19	Loss-of-Function and Gain-of-Function Mutations in KCNQ5 Cause Intellectual Disability or Epileptic Encephalopathy. American Journal of Human Genetics, 2017, 101, 65-74.	2.6	99
20	Exome Sequencing and the Management of Neurometabolic Disorders. New England Journal of Medicine, 2016, 374, 2246-2255.	13.9	254
21	Recessive <scp>ITPA</scp> mutations cause an early infantile encephalopathy. Annals of Neurology, 2015, 78, 649-658.	2.8	45
22	Individualized long-term outcomes in blood phenylalanine concentrations and dietary phenylalanine tolerance in 11 patients with primary phenylalanine hydroxylase (PAH) deficiency treated with Sapropterin-dihydrochloride. Molecular Genetics and Metabolism, 2015, 114, 409-414.	0.5	5
23	Expansion of the QARS deficiency phenotype with report of a family with isolated supratentorial brain abnormalities. Neurogenetics, 2015, 16, 145-149.	0.7	11
24	Mitochondrial Carbonic Anhydrase VA Deficiency Resulting from CA5A Alterations Presents with Hyperammonemia in Early Childhood. American Journal of Human Genetics, 2014, 94, 453-461.	2.6	82
25	Recurrent subacute post-viral onset of ataxia associated with a PRF1 mutation. European Journal of Human Genetics, 2013, 21, 1232-1239.	1.4	19
26	A variant of unknown significance in the GLA gene causing diagnostic uncertainty in a young female with isolated hypertrophic cardiomyopathy. Gene, 2012, 497, 320-322.	1.0	6