Daniela Buhas

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/7031600/publications.pdf

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18	507	840776 11	794594 19
papers	citations	h-index	g-index

19	19	19	1434
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Timing of therapy and neurodevelopmental outcomes in 18 families with pyridoxine-dependent epilepsy. Molecular Genetics and Metabolism, 2022, 135, 350-356.	1.1	11
2	Life-threatening viral disease in a novel form of autosomal recessive IFNAR2 deficiency in the Arctic. Journal of Experimental Medicine, 2022, 219, .	8.5	33
3	Consensus guidelines for the diagnosis and management of pyridoxineâ€dependent epilepsy due to Î±â€aminoadipic semialdehyde dehydrogenase deficiency. Journal of Inherited Metabolic Disease, 2021, 44, 178-192.	3.6	47
4	Enhanced cGAS-STINGâ€“dependent interferon signaling associated with mutations in ATAD3A. Journal of Experimental Medicine, 2021, 218, .	8.5	43
5	French-Canadian families from Saguenay-Lac-Saint-Jean: a new founder population for APECED. Endocrine, 2021, , 1.	2.3	1
6	Monocarboxylate transporter-1 deficiency results in severe metabolic acidosis with ketogenic diet in early onset absence epilepsy: Case report. Seizure: the Journal of the British Epilepsy Association, 2020, 74, 31-32.	2.0	1
7	Biallelic MADD variants cause a phenotypic spectrum ranging from developmental delay to a multisystem disorder. Brain, 2020, 143, 2437-2453.	7.6	21
8	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.	2.7	11
9	HSD10 mitochondrial disease: p.Leu122Val variant, mild clinical phenotype, and founder effect in Frenchâ€Canadian patients from Quebec. Molecular Genetics & Genomic Medicine, 2019, 7, e1000.	1.2	8
10	Proteomic investigations of human HERC2 mutants: Insights into the pathobiology of a neurodevelopmental disorder. Biochemical and Biophysical Research Communications, 2019, 512, 421-427.	2.1	13
11	Combined malonic and methylmalonic aciduria due to ACSF3 mutations: Benign clinical course in an unselected cohort. Journal of Inherited Metabolic Disease, 2019, 42, 107-116.	3.6	23
12	Combined malonic and methylmalonic aciduria due to ACSF3 mutations: benign clinical course in an unselected cohort. Journal of Inherited Metabolic Disease, 2019, 42, 107.	3.6	4
13	Hyperphosphatasia with mental retardation syndrome, expanded phenotype of PIGL related disorders. Molecular Genetics and Metabolism Reports, 2018, 15, 46-49.	1.1	13
14	The QuÃ©bec NTBC Study. Advances in Experimental Medicine and Biology, 2017, 959, 187-195.	1.6	15
15	RMND1-Related Leukoencephalopathy With Temporal Lobe Cysts and Hearing Lossâ€”Another Mendelian Mimicker of Congenital Cytomegalovirus Infection. Pediatric Neurology, 2017, 66, 59-62.	2.1	12
16	Recurrent De Novo and Biallelic Variation of ATAD3A , Encoding a Mitochondrial Membrane Protein, Results in Distinct Neurological Syndromes. American Journal of Human Genetics, 2016, 99, 831-845.	6.2	146
17	Succinateâ€CoA ligase deficiency due to mutations in SUCLA2 and SUCLG1: phenotype and genotype correlations in 71 patients. Journal of Inherited Metabolic Disease, 2016, 39, 243-252.	3.6	79
18	A treatable new cause of chorea: Betaâ€ketothiolase deficiency. Movement Disorders, 2013, 28, 1054-1056.	3.9	25