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	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
2	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
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	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
6	A treatable new cause of chorea: Betaâ€ketothiolase deficiency. Movement Disorders, 2013, 28, 1054-1056.	3.9	25
7	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
8	Biallelic MADD variants cause a phenotypic spectrum ranging from developmental delay to a multisystem disorder. Brain, 2020, 143, 2437-2453.	7.6	21
9	The QuÃ©bec NTBC Study. Advances in Experimental Medicine and Biology, 2017, 959, 187-195.	1.6	15
10	Hyperphosphatasia with mental retardation syndrome, expanded phenotype of PIGL related disorders. Molecular Genetics and Metabolism Reports, 2018, 15, 46-49.	1.1	13
11	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
12	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
13	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
14	Timing of therapy and neurodevelopmental outcomes in 18 families with pyridoxine-dependent epilepsy. Molecular Genetics and Metabolism, 2022, 135, 350-356.	1.1	11
15	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
16	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
17	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
18	French-Canadian families from Saguenay-Lac-Saint-Jean: a new founder population for APECED. Endocrine, 2021, , 1.	2.3	1