Matthias Zielonka

List of Publications by Year in descending order

Source: https://exaly.com/author-pdf/2228487/publications.pdf

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28	443	687363 13	752698 20
papers	citations	h-index	g-index

29	29	29	493
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Postauthorization safety study of betaine anhydrous. Journal of Inherited Metabolic Disease, 2022, 45, 719-733.	3.6	5
2	Unmet Needs of Parents of Children with Urea Cycle Disorders. Children, 2022, 9, 712.	1.5	4
3	Cystathionine Î²â€synthase deficiency in the <scp>Eâ€HOD registryâ€part</scp> I: pyridoxine responsiveness as a determinant of biochemical and clinical phenotype at diagnosis. Journal of Inherited Metabolic Disease, 2021, 44, 677-692.	3.6	20
4	Quantitative retrospective natural history modeling for orphan drug development. Journal of Inherited Metabolic Disease, 2021, 44, 99-109.	3.6	16
5	Long-term effects of medical management on growth and weight in individuals with urea cycle disorders. Scientific Reports, 2020, 10, 11948.	3.3	11
6	Fatal outcome after heart surgery in PMM2-CDG due to a rare homozygous gene variant with double effects. Molecular Genetics and Metabolism Reports, 2020, 25, 100673.	1.1	5
7	From genotype to phenotype: Early prediction of disease severity in argininosuccinic aciduria. Human Mutation, 2020, 41, 946-960.	2.5	14
8	Chronic hyperammonemia causes a hypoglutamatergic and hyperGABAergic metabolic state associated with neurobehavioral abnormalities in zebrafish larvae. Experimental Neurology, 2020, 331, 113330.	4.1	12
9	FDA orphan drug designations for lysosomal storage disorders â€“ a cross-sectional analysis. PLoS ONE, 2020, 15, e0230898.	2.5	17
10	Severity-adjusted evaluation of newborn screening on the metabolic disease course in individuals with cytosolic urea cycle disorders. Molecular Genetics and Metabolism, 2020, 131, 390-397.	1.1	14
11	FDA orphan drug designations for lysosomal storage disorders â€“ a cross-sectional analysis. , 2020, 15, e0230898.		0
12	FDA orphan drug designations for lysosomal storage disorders â€“ a cross-sectional analysis. , 2020, 15, e0230898.		0
13	FDA orphan drug designations for lysosomal storage disorders â€“ a cross-sectional analysis. , 2020, 15, e0230898.		0
14	FDA orphan drug designations for lysosomal storage disorders â€“ a cross-sectional analysis. , 2020, 15, e0230898.		0
15	A cross-sectional quantitative analysis of the natural history of free sialic acid storage diseaseâ€”an ultra-orphan multisystemic lysosomal storage disorder. Genetics in Medicine, 2019, 21, 347-352.	2.4	14
16	Ultraâ€orphan lysosomal storage diseases: A crossâ€sectional quantitative analysis of the natural history of alphaâ€mannosidosis. Journal of Inherited Metabolic Disease, 2019, 42, 975-983.	3.6	26
17	Disasters in Germany and France: An Analysis of the Emergency Events Database From a Pediatric Perspective. Disaster Medicine and Public Health Preparedness, 2019, 13, 958-965.	1.3	11
18	Early prediction of phenotypic severity in Citrullinemia Type 1. Annals of Clinical and Translational Neurology, 2019, 6, 1858-1871.	3.7	26

#	Article	IF	Citations
19	Impact of Diagnosis and Therapy on Cognitive Function in Urea Cycle Disorders. Annals of Neurology, 2019, 86, 116-128.	5.3	42
20	QDPR homologues in Danio rerio regulate melanin synthesis, early gliogenesis, and glutamine homeostasis. PLoS ONE, 2019, 14, e0215162.	2.5	12
21	Clinical characteristics of 248 patients with Krabbe disease: quantitative natural history modeling based on published cases. Genetics in Medicine, 2019, 21, 2208-2215.	2.4	33
22	Bioenergetic dysfunction in a zebrafish model of acute hyperammonemic decompensation. Experimental Neurology, 2019, 314, 91-99.	4.1	16
23	A cross-sectional quantitative analysis of the natural history of Farber disease: an ultra-orphan condition with rheumatologic and neurological cardinal disease features. Genetics in Medicine, 2018, 20, 524-530.	2.4	24
24	Cognitive and Behavioral Consequences of Pediatric Delirium: A Pilot Study*. Pediatric Critical Care Medicine, 2018, 19, e531-e537.	0.5	22
25	Pharmacologic rescue of hyperammonemia-induced toxicity in zebrafish by inhibition of ornithine aminotransferase. PLoS ONE, 2018, 13, e0203707.	2.5	19
26	Quantitative clinical characteristics of 53 patients with MPS VII: a cross-sectional analysis. Genetics in Medicine, 2017, 19, 983-988.	2.4	42
27	Severe Acute Subdural Hemorrhage in a Patient With Glutaric Aciduria Type I After Minor Head Trauma. Journal of Child Neurology, 2015, 30, 1065-1069.	1.4	21
28	Dopamine-Responsive Growth-Hormone Deficiency and Central Hypothyroidism in Sepiapterin Reductase Deficiency. JIMD Reports, 2015, 24, 109-113.	1.5	15