

Matthias Zielonka

List of Publications by Year in descending order

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Version: 2024-02-01

28
papers

443
citations

687363

13
h-index

752698

20
g-index

29
all docs

29
docs citations

29
times ranked

493
citing authors

#	ARTICLE	IF	CITATIONS
1	Quantitative clinical characteristics of 53 patients with MPS VII: a cross-sectional analysis. <i>Genetics in Medicine</i> , 2017, 19, 983-988.	2.4	42
2	Impact of Diagnosis and Therapy on Cognitive Function in Urea Cycle Disorders. <i>Annals of Neurology</i> , 2019, 86, 116-128.	5.3	42
3	Clinical characteristics of 248 patients with Krabbe disease: quantitative natural history modeling based on published cases. <i>Genetics in Medicine</i> , 2019, 21, 2208-2215.	2.4	33
4	Ultra-orphan lysosomal storage diseases: A cross-sectional quantitative analysis of the natural history of alpha-mannosidosis. <i>Journal of Inherited Metabolic Disease</i> , 2019, 42, 975-983.	3.6	26
5	Early prediction of phenotypic severity in Citrullinemia Type 1. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 1858-1871.	3.7	26
6	A cross-sectional quantitative analysis of the natural history of Farber disease: an ultra-orphan condition with rheumatologic and neurological cardinal disease features. <i>Genetics in Medicine</i> , 2018, 20, 524-530.	2.4	24
7	Cognitive and Behavioral Consequences of Pediatric Delirium: A Pilot Study*. <i>Pediatric Critical Care Medicine</i> , 2018, 19, e531-e537.	0.5	22
8	Severe Acute Subdural Hemorrhage in a Patient With Glutaric Aciduria Type I After Minor Head Trauma. <i>Journal of Child Neurology</i> , 2015, 30, 1065-1069.	1.4	21
9	Cystathionine β -synthase deficiency in the <sc>EaHOD registry</sc> I: pyridoxine responsiveness as a determinant of biochemical and clinical phenotype at diagnosis. <i>Journal of Inherited Metabolic Disease</i> , 2021, 44, 677-692.	3.6	20
10	Pharmacologic rescue of hyperammonemia-induced toxicity in zebrafish by inhibition of ornithine aminotransferase. <i>PLoS ONE</i> , 2018, 13, e0203707.	2.5	19
11	FDA orphan drug designations for lysosomal storage disorders – a cross-sectional analysis. <i>PLoS ONE</i> , 2020, 15, e0230898.	2.5	17
12	Bioenergetic dysfunction in a zebrafish model of acute hyperammonemic decompensation. <i>Experimental Neurology</i> , 2019, 314, 91-99.	4.1	16
13	Quantitative retrospective natural history modeling for orphan drug development. <i>Journal of Inherited Metabolic Disease</i> , 2021, 44, 99-109.	3.6	16
14	Dopamine-Responsive Growth-Hormone Deficiency and Central Hypothyroidism in Sepiapterin Reductase Deficiency. <i>JIMD Reports</i> , 2015, 24, 109-113.	1.5	15
15	A cross-sectional quantitative analysis of the natural history of free sialic acid storage disease – an ultra-orphan multisystemic lysosomal storage disorder. <i>Genetics in Medicine</i> , 2019, 21, 347-352.	2.4	14
16	From genotype to phenotype: Early prediction of disease severity in argininosuccinic aciduria. <i>Human Mutation</i> , 2020, 41, 946-960.	2.5	14
17	Severity-adjusted evaluation of newborn screening on the metabolic disease course in individuals with cytosolic urea cycle disorders. <i>Molecular Genetics and Metabolism</i> , 2020, 131, 390-397.	1.1	14
18	QDPR homologues in <i>Danio rerio</i> regulate melanin synthesis, early gliogenesis, and glutamine homeostasis. <i>PLoS ONE</i> , 2019, 14, e0215162.	2.5	12

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19	Chronic hyperammonemia causes a hypoglutamatergic and hyperGABAergic metabolic state associated with neurobehavioral abnormalities in zebrafish larvae. <i>Experimental Neurology</i> , 2020, 331, 113330.	4.1	12
20	Disasters in Germany and France: An Analysis of the Emergency Events Database From a Pediatric Perspective. <i>Disaster Medicine and Public Health Preparedness</i> , 2019, 13, 958-965.	1.3	11
21	Long-term effects of medical management on growth and weight in individuals with urea cycle disorders. <i>Scientific Reports</i> , 2020, 10, 11948.	3.3	11
22	Fatal outcome after heart surgery in PMM2-CDG due to a rare homozygous gene variant with double effects. <i>Molecular Genetics and Metabolism Reports</i> , 2020, 25, 100673.	1.1	5
23	Postauthorization safety study of betaine anhydrous. <i>Journal of Inherited Metabolic Disease</i> , 2022, 45, 719-733.	3.6	5
24	Unmet Needs of Parents of Children with Urea Cycle Disorders. <i>Children</i> , 2022, 9, 712.	1.5	4
25	FDA orphan drug designations for lysosomal storage disorders – a cross-sectional analysis. , 2020, 15, e0230898.		0
26	FDA orphan drug designations for lysosomal storage disorders – a cross-sectional analysis. , 2020, 15, e0230898.		0
27	FDA orphan drug designations for lysosomal storage disorders – a cross-sectional analysis. , 2020, 15, e0230898.		0
28	FDA orphan drug designations for lysosomal storage disorders – a cross-sectional analysis. , 2020, 15, e0230898.		0