

Claudia Carducci

List of Publications by Year in descending order

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Version: 2024-02-01

48
papers

1,574
citations

430874

18
h-index

330143

37
g-index

48
all docs

48
docs citations

48
times ranked

2066
citing authors

#	ARTICLE	IF	CITATIONS
1	The diagnostic challenge of mild citrulline elevation at newborn screening. <i>Molecular Genetics and Metabolism</i> , 2022, 135, 327-332.	1.1	7
2	Engineering new metabolic pathways in isolated cells for the degradation of guanidinoacetic acid and simultaneous production of creatine. <i>Molecular Therapy - Methods and Clinical Development</i> , 2022, 25, 26-40.	4.1	1
3	3-Methylglutaconic Aciduria Type I Due to AUH Defect: The Case Report of a Diagnostic Odyssey and a Review of the Literature. <i>International Journal of Molecular Sciences</i> , 2022, 23, 4422.	4.1	2
4	Creatine Levels in Patients with Phenylketonuria and Mild Hyperphenylalaninemia: A Pilot Study. <i>Life</i> , 2021, 11, 425.	2.4	1
5	Intellectual Disability and Brain Creatine Deficit: Phenotyping of the Genetic Mouse Model for GAMT Deficiency. <i>Genes</i> , 2021, 12, 1201.	2.4	4
6	Simultaneous determination of 5-hydroxytryptophan and 3-O-methyldopa in dried blood spot by UPLC-MS/MS: A useful tool for the diagnosis of L-amino acid decarboxylase deficiency. <i>Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences</i> , 2021, 1185, 122999.	2.3	2
7	Cognitive Outcomes and Relationships with Phenylalanine in Phenylketonuria: A Comparison between Italian and English Adult Samples. <i>Nutrients</i> , 2020, 12, 3033.	4.1	7
8	Long-term clinical outcome of 6-pyruvoyl-tetrahydropterin synthase-deficient patients. <i>Molecular Genetics and Metabolism</i> , 2020, 131, 155-162.	1.1	11
9	Executive functioning, adaptive skills, emotional and behavioral profile: A comparison between autism spectrum disorder and phenylketonuria. <i>Molecular Genetics and Metabolism Reports</i> , 2020, 23, 100577.	1.1	3
10	Co-occurring WARS2 and CHRNA6 mutations in a child with a severe form of infantile parkinsonism. <i>Parkinsonism and Related Disorders</i> , 2020, 72, 75-79.	2.2	16
11	Clinical characterization of tremor in patients with phenylketonuria. <i>Molecular Genetics and Metabolism</i> , 2019, 128, 53-56.	1.1	9
12	Newborn screening for homocystinurias: Recent recommendations versus current practice. <i>Journal of Inherited Metabolic Disease</i> , 2019, 42, 128-139.	3.6	37
13	Adult cognitive outcomes in phenylketonuria: explaining causes of variability beyond average Phe levels. <i>Orphanet Journal of Rare Diseases</i> , 2019, 14, 273.	2.7	30
14	<i><sc>AP</sc>1S2</i> truncating variant in a patient with severe neurodevelopmental disorder and cerebral folate deficiency. <i>Acta Paediatrica, International Journal of Paediatrics</i> , 2019, 108, 564-565.	1.5	2
15	Successful Pregnancy in a Patient with Amino Acid Decarboxylase Deficiency: Therapeutic Management and Clinical Outcome. <i>Movement Disorders Clinical Practice</i> , 2018, 5, 446-447.	1.5	7
16	A new therapy prevents intellectual disability in mouse with phenylketonuria. <i>Molecular Genetics and Metabolism</i> , 2018, 124, 39-49.	1.1	18
17	Molecular Analysis of PKU-Associated PAH Mutations: A Fast and Simple Genotyping Test. <i>Methods and Protocols</i> , 2018, 1, 30.	2.0	3
18	Development of a new UPLC-ESI-MS/MS method for the determination of biopterin and neopterin in dried blood spot. <i>Clinica Chimica Acta</i> , 2017, 466, 145-151.	1.1	8

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19	Altered tetrahydrobiopterin metabolism in patients with phenylalanine hydroxylase deficiency. <i>European Journal of Pediatrics</i> , 2017, 176, 917-924.	2.7	1
20	Predictability and inconsistencies in the cognitive outcome of early treated PKU patients. <i>Journal of Inherited Metabolic Disease</i> , 2017, 40, 793-799.	3.6	18
21	Psychiatric disorders in adolescent and young adult patients with phenylketonuria. <i>Molecular Genetics and Metabolism</i> , 2016, 117, 12-18.	1.1	32
22	The Spectrum of Early Movement Disorders in Congenital Defects of Biogenic Amine Metabolism. <i>Journal of Pediatric Neurology</i> , 2015, 13, 213-224.	0.2	2
23	Neurocognitive and neuroimaging outcome of early treated young adult PKU patients: A longitudinal study. <i>Molecular Genetics and Metabolism</i> , 2015, 115, 84-90.	1.1	58
24	Urine sepiapterin excretion as a new diagnostic marker for sepiapterin reductase deficiency. <i>Molecular Genetics and Metabolism</i> , 2015, 115, 157-160.	1.1	18
25	The outcome of white matter abnormalities in early treated phenylketonuric patients: A retrospective longitudinal long-term study. <i>Molecular Genetics and Metabolism</i> , 2015, 116, 171-177.	1.1	27
26	Granulocytic Myeloid Derived Suppressor Cells Expansion during Active Pulmonary Tuberculosis Is Associated with High Nitric Oxide Plasma Level. <i>PLoS ONE</i> , 2015, 10, e0123772.	2.5	67
27	Age-Related Psychophysiological Vulnerability to Phenylalanine in Phenylketonuria. <i>Frontiers in Pediatrics</i> , 2014, 2, 57.	1.9	9
28	Report of Two Never Treated Adult Sisters with Aromatic L-Amino Acid Decarboxylase Deficiency: A Portrait of the Natural History of the Disease or an Expanding Phenotype?. <i>JIMD Reports</i> , 2014, 15, 39-45.	1.5	29
29	Dopamine transporter deficiency syndrome: phenotypic spectrum from infancy to adulthood. <i>Brain</i> , 2014, 137, 1107-1119.	7.6	265
30	Erythrocyte-mediated delivery of phenylalanine ammonia lyase for the treatment of phenylketonuria in BTBR-Pahenu2 mice. <i>Journal of Controlled Release</i> , 2014, 194, 37-44.	9.9	45
31	Screening for dopa-responsive dystonia in patients with scans without evidence of dopaminergic deficiency (SWEDD). <i>Journal of Neurology</i> , 2014, 261, 2204-2208.	3.6	12
32	A new case of malonic aciduria with a presymptomatic diagnosis and an early treatment. <i>Brain and Development</i> , 2013, 35, 675-680.	1.1	15
33	Very early pattern of movement disorders in sepiapterin reductase deficiency. <i>Neurology</i> , 2013, 81, 2141-2142.	1.1	19
34	Behavioral and Neurochemical Characterization of New Mouse Model of Hyperphenylalaninemia. <i>PLoS ONE</i> , 2013, 8, e84697.	2.5	17
35	A New Tyrosine Hydroxylase Genotype Associated With Early-Onset Severe Encephalopathy. <i>Journal of Child Neurology</i> , 2012, 27, 523-525.	1.4	11
36	Urinary Neopterin and Phenylalanine Loading Test as Tools for the Biochemical Diagnosis of Segawa Disease. <i>JIMD Reports</i> , 2012, 7, 67-75.	1.5	7

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37	Enhanced interpretation of newborn screening results without analyte cutoff values. <i>Genetics in Medicine</i> , 2012, 14, 648-655.	2.4	117
38	In vitro study of uptake and synthesis of creatine and its precursors by cerebellar granule cells and astrocytes suggests some hypotheses on the physiopathology of the inherited disorders of creatine metabolism. <i>BMC Neuroscience</i> , 2012, 13, 41.	1.9	17
39	Clinical validation of cutoff target ranges in newborn screening of metabolic disorders by tandem mass spectrometry: A worldwide collaborative project. <i>Genetics in Medicine</i> , 2011, 13, 230-254.	2.4	308
40	Renal transplant in methylmalonic acidemia: could it be the best option?. <i>Pediatric Nephrology</i> , 2007, 22, 1209-1214.	1.7	43
41	Quantitative determination of guanidinoacetate and creatine in dried blood spot by flow injection analysis-electrospray tandem mass spectrometry. <i>Clinica Chimica Acta</i> , 2006, 364, 180-187.	1.1	61
42	A mutation on exon 6 of guanidinoacetate methyltransferase (GAMT) gene supports a different function for isoform a and b of GAMT enzyme. <i>Molecular Genetics and Metabolism</i> , 2006, 87, 88-90.	1.1	13
43	Guanidinoacetate and Creatine plus Creatinine Assessment in Physiologic Fluids: An Effective Diagnostic Tool for the Biochemical Diagnosis of Arginine:Glycine Amidinotransferase and Guanidinoacetate Methyltransferase Deficiencies. <i>Clinical Chemistry</i> , 2002, 48, 1772-1778.	3.2	57
44	Automated high-performance liquid chromatographic method for the determination of guanidinoacetic acid in dried blood spots: a tool for early diagnosis of guanidinoacetate methyltransferase deficiency. <i>Biomedical Applications</i> , 2001, 755, 343-348.	1.7	37
45	Two New Severe Mutations Causing Guanidinoacetate Methyltransferase Deficiency. <i>Molecular Genetics and Metabolism</i> , 2000, 71, 633-638.	1.1	30
46	Automated high-performance liquid chromatographic method for the determination of homocysteine in plasma samples. <i>Journal of Chromatography A</i> , 1999, 846, 93-100.	3.7	32
47	Neuropsychological and Neuroradiological (MRI) Variations During Phenylalanine Load: Protective Effect of Valine, Leucine, and Isoleucine Supplementation. <i>Journal of Child Neurology</i> , 1997, 12, 338-340.	1.4	5
48	Automated method for the measurement of amino acids in urine by high-performance liquid chromatography. <i>Journal of Chromatography A</i> , 1996, 729, 173-180.	3.7	34