Claudia Carducci

List of Publications by Year
in descending order

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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	Clinical validation of cutoff target ranges in newborn screening of metabolic disorders by tandem mass spectrometry: A worldwide collaborative project. Genetics in Medicine, 2011, 13, 230-254.	2.4	308
2	Dopamine transporter deficiency syndrome: phenotypic spectrum from infancy to adulthood. Brain, 2014, 137, 1107-1119.	7.6	265
3	Enhanced interpretation of newborn screening results without analyte cutoff values. Genetics in Medicine, 2012, 14, 648-655.	2.4	117
4	Granulocytic Myeloid Derived Suppressor Cells Expansion during Active Pulmonary Tuberculosis Is Associated with High Nitric Oxide Plasma Level. PLoS ONE, 2015, 10, e0123772.	2.5	67
5	Quantitative determination of guanidinoacetate and creatine in dried blood spot by flow injection analysis-electrospray tandem mass spectrometry. Clinica Chimica Acta, 2006, 364, 180-187.	1.1	61
6	Neurocognitive and neuroimaging outcome of early treated young adult PKU patients: A longitudinal study. Molecular Genetics and Metabolism, 2015, 115, 84-90.	1.1	58
7	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. Clinical Chemistry, 2002, 48, 1772-1778.	3.2	57
8	Erythrocyte-mediated delivery of phenylalanine ammonia lyase for the treatment of phenylketonuria in BTBR-Pahenu2 mice. Journal of Controlled Release, 2014, 194, 37-44.	9.9	45
9	Renal transplant in methylmalonic acidemia: could it be the best option?. Pediatric Nephrology, 2007, 22, 1209-1214.	1.7	43
10	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. Biomedical Applications, 2001, 755, 343-348.	1.7	37
11	Newborn screening for homocystinurias: Recent recommendations versus current practice. Journal of Inherited Metabolic Disease, 2019, 42, 128-139.	3.6	37
12	Automated method for the measurement of amino acids in urine by high-performance liquid chromatography. Journal of Chromatography A, 1996, 729, 173-180.	3.7	34
13	Automated high-performance liquid chromatographic method for the determination of homocysteine in plasma samples. Journal of Chromatography A, 1999, 846, 93-100.	3.7	32
14	Psychiatric disorders in adolescent and young adult patients with phenylketonuria. Molecular Genetics and Metabolism, 2016, 117, 12-18.	1.1	32
15	Two New Severe Mutations Causing Guanidinoacetate Methyltransferase Deficiency. Molecular Genetics and Metabolism, 2000, 71, 633-638.	1.1	30
16	Adult cognitive outcomes in phenylketonuria: explaining causes of variability beyond average Phe levels. Orphanet Journal of Rare Diseases, 2019, 14, 273.	2.7	30
17	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?. JIMD Reports, 2014, 15, 39-45.	1.5	29
18	The outcome of white matter abnormalities in early treated phenylketonuric patients: A retrospective longitudinal long-term study. Molecular Genetics and Metabolism, 2015, 116, 171-177.	1.1	27

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19	Very early pattern of movement disorders in sepiapterin reductase deficiency. Neurology, 2013, 81, 2141-2142.	1.1	19
20	Urine sepiapterin excretion as a new diagnostic marker for sepiapterin reductase deficiency. Molecular Genetics and Metabolism, 2015, 115, 157-160.	1.1	18
21	Predictability and inconsistencies in the cognitive outcome of early treated PKU patients. Journal of Inherited Metabolic Disease, 2017, 40, 793-799.	3.6	18
22	A new therapy prevents intellectual disability in mouse with phenylketonuria. Molecular Genetics and Metabolism, 2018, 124, 39-49.	1.1	18
23	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. BMC Neuroscience, 2012, 13, 41.	1.9	17
24	Behavioral and Neurochemical Characterization of New Mouse Model of Hyperphenylalaninemia. PLoS ONE, 2013, 8, e84697.	2.5	17
25	Co-occurring WARS2 and CHRNA6 mutations in a child with a severe form of infantile parkinsonism. Parkinsonism and Related Disorders, 2020, 72, 75-79.	2.2	16
26	A new case of malonic aciduria with a presymptomatic diagnosis and an early treatment. Brain and Development, 2013, 35, 675-680.	1.1	15
27	A mutation on exon 6 of guanidinoacetate methyltransferase (GAMT) gene supports a different function for isoform a and b of GAMT enzyme. Molecular Genetics and Metabolism, 2006, 87, 88-90.	1.1	13
28	Screening for dopa-responsive dystonia in patients with scans without evidence of dopaminergic deficiency (SWEDD). Journal of Neurology, 2014, 261, 2204-2208.	3.6	12
29	A New Tyrosine Hydroxylase Genotype Associated With Early-Onset Severe Encephalopathy. Journal of Child Neurology, 2012, 27, 523-525.	1.4	11
30	Long-term clinical outcome of 6-pyruvoyl-tetrahydropterin synthase-deficient patients. Molecular Genetics and Metabolism, 2020, 131, 155-162.	1.1	11
31	Age-Related Psychophysiological Vulnerability to Phenylalanine in Phenylketonuria. Frontiers in Pediatrics, 2014, 2, 57.	1.9	9
32	Clinical characterization of tremor in patients with phenylketonuria. Molecular Genetics and Metabolism, 2019, 128, 53-56.	1.1	9
33	Development of a new UPLC-ESI-MS/MS method for the determination of biopterin and neopterin in dried blood spot. Clinica Chimica Acta, 2017, 466, 145-151.	1.1	8
34	Urinary Neopterin and Phenylalanine Loading Test as Tools for the Biochemical Diagnosis of Segawa Disease. JIMD Reports, 2012, 7, 67-75.	1.5	7
35	Successful Pregnancy in a Patient with Lâ€Amino Acid Decarboxylase Deficiency: Therapeutic Management and Clinical Outcome. Movement Disorders Clinical Practice, 2018, 5, 446-447.	1.5	7
36	Cognitive Outcomes and Relationships with Phenylalanine in Phenylketonuria: A Comparison between Italian and English Adult Samples. Nutrients, 2020, 12, 3033.	4.1	7

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37	The diagnostic challenge of mild citrulline elevation at newborn screening. Molecular Genetics and Metabolism, 2022, 135, 327-332.	1.1	7
38	Neuropsychological and Neuroradiological (MRI) Variations During Phenylalanine Load: Protective Effect of Valine, Leucine, and Isoleucine Supplementation. Journal of Child Neurology, 1997, 12, 338-340.	1.4	5
39	Intellectual Disability and Brain Creatine Deficit: Phenotyping of the Genetic Mouse Model for GAMT Deficiency. Genes, 2021, 12, 1201.	2.4	4
40	Molecular Analysis of PKU-Associated PAH Mutations: A Fast and Simple Genotyping Test. Methods and Protocols, 2018, 1, 30.	2.0	3
41	Executive functioning, adaptive skills, emotional and behavioral profile: A comparison between autism spectrum disorder and phenylketonuria. Molecular Genetics and Metabolism Reports, 2020, 23, 100577.	1.1	3
42	The Spectrum of Early Movement Disorders in Congenital Defects of Biogenic Amine Metabolism. Journal of Pediatric Neurology, 2015, 13, 213-224.	0.2	2
43	<scp>AP</scp>1S2â€truncating variant in a patient with severe neurodevelopmental disorder and cerebral folate deficiency. Acta Paediatrica, International Journal of Paediatrics, 2019, 108, 564-565.	1.5	2
44	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. Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences, 2021, 1185, 122999.	2.3	2
45	3-Methylglutaconic Aciduria Type I Due to AUH Defect: The Case Report of a Diagnostic Odyssey and a Review of the Literature. International Journal of Molecular Sciences, 2022, 23, 4422.	4.1	2
46	Altered tetrahydrobiopterin metabolism in patients with phenylalanine hydroxylase deficiency. European Journal of Pediatrics, 2017, 176, 917-924.	2.7	1
47	Creatine Levels in Patients with Phenylketonuria and Mild Hyperphenylalaninemia: A Pilot Study. Life, 2021, 11, 425.	2.4	1
48	Engineering new metabolic pathways in isolated cells for the degradation of guanidinoacetic acid and simultaneous production of creatine. Molecular Therapy - Methods and Clinical Development, 2022, 25, 26-40.	4.1	1