Marzia Pasquali

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/9200538/publications.pdf

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24	1,060	759233 12	642732 23
papers	citations	h-index	g-index

24	24	24	2060
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Carnitine transport and fatty acid oxidation. Biochimica Et Biophysica Acta - Molecular Cell Research, 2016, 1863, 2422-2435.	4.1	556
2	Global Analysis of Plasma Lipids Identifies Liver-Derived Acylcarnitines as a Fuel Source for Brown Fat Thermogenesis. Cell Metabolism, 2017, 26, 509-522.e6.	16.2	185
3	Feasibility of newborn screening for guanidinoacetate methyltransferase (GAMT) deficiency. Journal of Inherited Metabolic Disease, 2014, 37, 231-236.	3.6	37
4	Effect of dietary lysine restriction and arginine supplementation in two patients with pyridoxine-dependent epilepsy. Molecular Genetics and Metabolism, 2016, 118, 167-172.	1.1	32
5	Anaplerotic therapy in propionic acidemia. Molecular Genetics and Metabolism, 2017, 122, 51-59.	1.1	28
6	Elevated cerebral spinal fluid biomarkers in children with mucopolysaccharidosis I-H. Scientific Reports, 2016, 6, 38305.	3.3	25
7	Diagnosis, Treatment, and Clinical Outcome of Patients with Mitochondrial Trifunctional Protein/Long-Chain 3-Hydroxy Acyl-CoA Dehydrogenase Deficiency. JIMD Reports, 2016, 31, 63-71.	1.5	25
8	Intrathecal enzyme replacement for Hurler syndrome: biomarker association with neurocognitive outcomes. Genetics in Medicine, 2019, 21, 2552-2560.	2.4	25
9	Clinical and biochemical outcome of patients with very long-chain acyl-CoA dehydrogenase deficiency. Molecular Genetics and Metabolism, 2019, 127, 64-73.	1.1	24
10	Clinical and biochemical outcomes of patients with medium-chain acyl-CoA dehydrogenase deficiency. Molecular Genetics and Metabolism, 2020, 129, 13-19.	1.1	21
11	A novel method for simultaneous quantification of alpha-aminoadipic semialdehyde/piperideine-6-carboxylate and pipecolic acid in plasma and urine. Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences, 2016, 1017-1018, 145-152.	2.3	20
12	Biochemical changes and clinical outcomes in 34 patients with classic galactosemia. Journal of Inherited Metabolic Disease, 2018, 41, 197-208.	3.6	19
13	Creatine metabolism in patients with urea cycle disorders. Molecular Genetics and Metabolism Reports, 2021, 29, 100791.	1.1	8
14	CAP/ACMG proficiency testing for biochemical genetics laboratories: a summary of performance. Genetics in Medicine, 2018, 20, 83-90.	2.4	7
15	Effect of genotype on galactose-1-phosphate in classic galactosemia patients. Molecular Genetics and Metabolism, 2018, 125, 258-265.	1.1	7
16	Unusual Metabolites in a Patient with Isovaleric Acidemia. Clinical Chemistry, 2019, 65, 595-597.	3.2	7
17	Retrospective analysis of 19 patients with 6-Pyruvoyl Tetrahydropterin Synthase Deficiency: Prolactin levels inversely correlate with growth. Molecular Genetics and Metabolism, 2020, 131, 380-389.	1.1	7
18	Prospective identification by neonatal screening of patients with guanidinoacetate methyltransferase deficiency. Molecular Genetics and Metabolism, 2021, 134, 60-64.	1.1	7

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19	Acylglycine Analysis by Ultraâ€Performance Liquid Chromatographyâ€Tandem Mass Spectrometry (UPLCâ€MS/MS). Current Protocols in Human Genetics, 2016, 91, 17.25.1-17.25.12.	3.5	6
20	Serum Acylcarnitines and Vitamin B12 Deficiency. Clinical Chemistry, 2002, 48, 1126-1128.	3.2	5
21	Towards a Newborn Screening Common Data Model: The Utah Newborn Screening Data Model. International Journal of Neonatal Screening, 2021, 7, 70.	3.2	4
22	Galactose-1-Phosphate Uridyltransferase Activities in Different Genotypes: A Retrospective Analysis of 927 Samples. journal of applied laboratory medicine, The, 2018, 3, 222-230.	1.3	3
23	Quantitative analysis of urine acylglycines by ultra-performance liquid chromatography-tandem mass spectrometry (UPLC-MS/MS): Reference intervals and disease specific patterns in individuals with organic acidemias and fatty acid oxidation disorders. Clinica Chimica Acta, 2021, 523, 285-289.	1.1	2
24	Metabolic Lysosomal Enzyme Probes. FASEB Journal, 2013, 27, 576.1.	0.5	0