

# Marzia Pasquali

## List of Publications by Year in descending order

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Version: 2024-02-01

24  
papers

1,060  
citations

759233

12  
h-index

642732

23  
g-index

24  
all docs

24  
docs citations

24  
times ranked

2060  
citing authors

#	ARTICLE	IF	CITATIONS
1	Prospective identification by neonatal screening of patients with guanidinoacetate methyltransferase deficiency. <i>Molecular Genetics and Metabolism</i> , 2021, 134, 60-64.	1.1	7
2	Creatine metabolism in patients with urea cycle disorders. <i>Molecular Genetics and Metabolism Reports</i> , 2021, 29, 100791.	1.1	8
3	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. <i>Clinica Chimica Acta</i> , 2021, 523, 285-289.	1.1	2
4	Towards a Newborn Screening Common Data Model: The Utah Newborn Screening Data Model. <i>International Journal of Neonatal Screening</i> , 2021, 7, 70.	3.2	4
5	Clinical and biochemical outcomes of patients with medium-chain acyl-CoA dehydrogenase deficiency. <i>Molecular Genetics and Metabolism</i> , 2020, 129, 13-19.	1.1	21
6	Retrospective analysis of 19 patients with 6-Pyruvoyl Tetrahydropterin Synthase Deficiency: Prolactin levels inversely correlate with growth. <i>Molecular Genetics and Metabolism</i> , 2020, 131, 380-389.	1.1	7
7	Clinical and biochemical outcome of patients with very long-chain acyl-CoA dehydrogenase deficiency. <i>Molecular Genetics and Metabolism</i> , 2019, 127, 64-73.	1.1	24
8	Intrathecal enzyme replacement for Hurler syndrome: biomarker association with neurocognitive outcomes. <i>Genetics in Medicine</i> , 2019, 21, 2552-2560.	2.4	25
9	Unusual Metabolites in a Patient with Isovaleric Acidemia. <i>Clinical Chemistry</i> , 2019, 65, 595-597.	3.2	7
10	Biochemical changes and clinical outcomes in 34 patients with classic galactosemia. <i>Journal of Inherited Metabolic Disease</i> , 2018, 41, 197-208.	3.6	19
11	CAP/ACMG proficiency testing for biochemical genetics laboratories: a summary of performance. <i>Genetics in Medicine</i> , 2018, 20, 83-90.	2.4	7
12	Galactose-1-Phosphate Uridyltransferase Activities in Different Genotypes: A Retrospective Analysis of 927 Samples. <i>Journal of Applied Laboratory Medicine</i> , 2018, 3, 222-230.	1.3	3
13	Effect of genotype on galactose-1-phosphate in classic galactosemia patients. <i>Molecular Genetics and Metabolism</i> , 2018, 125, 258-265.	1.1	7
14	Global Analysis of Plasma Lipids Identifies Liver-Derived Acylcarnitines as a Fuel Source for Brown Fat Thermogenesis. <i>Cell Metabolism</i> , 2017, 26, 509-522.e6.	16.2	185
15	Anaplerotic therapy in propionic acidemia. <i>Molecular Genetics and Metabolism</i> , 2017, 122, 51-59.	1.1	28
16	Elevated cerebral spinal fluid biomarkers in children with mucopolysaccharidosis I-H. <i>Scientific Reports</i> , 2016, 6, 38305.	3.3	25
17	Diagnosis, Treatment, and Clinical Outcome of Patients with Mitochondrial Trifunctional Protein/Long-Chain 3-Hydroxy Acyl-CoA Dehydrogenase Deficiency. <i>JIMD Reports</i> , 2016, 31, 63-71.	1.5	25
18	Acylglycine Analysis by Ultra-Performance Liquid Chromatography-Tandem Mass Spectrometry (UPLC-MS/MS). <i>Current Protocols in Human Genetics</i> , 2016, 91, 17.25.1-17.25.12.	3.5	6

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19	Effect of dietary lysine restriction and arginine supplementation in two patients with pyridoxine-dependent epilepsy. <i>Molecular Genetics and Metabolism</i> , 2016, 118, 167-172.	1.1	32
20	A novel method for simultaneous quantification of alpha-amino adipic semialdehyde/piperidine-6-carboxylate and pipercolic acid in plasma and urine. <i>Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences</i> , 2016, 1017-1018, 145-152.	2.3	20
21	Carnitine transport and fatty acid oxidation. <i>Biochimica Et Biophysica Acta - Molecular Cell Research</i> , 2016, 1863, 2422-2435.	4.1	556
22	Feasibility of newborn screening for guanidinoacetate methyltransferase (GAMT) deficiency. <i>Journal of Inherited Metabolic Disease</i> , 2014, 37, 231-236.	3.6	37
23	Metabolic Lysosomal Enzyme Probes. <i>FASEB Journal</i> , 2013, 27, 576.1.	0.5	0
24	Serum Acylcarnitines and Vitamin B12 Deficiency. <i>Clinical Chemistry</i> , 2002, 48, 1126-1128.	3.2	5