Ana M Gaspar

List of Publications by Year
in descending order

14 papers	173 citations	1305906 8 h-index	1255698 13 g-index
14 all docs	14 docs citations	14 times ranked	329 citing authors

#	Article	IF	Citations
1	Role of RNA in Molecular Diagnosis of MADD Patients. Biomedicines, 2021, 9, 507.	1.4	4
2	Phenylketonuria in Portugal: Genotypeâ€“phenotype correlations using molecular, biochemical, and haplotypic analyses. Molecular Genetics & Genomic Medicine, 2021, 9, e1559.	0.6	4
3	Targeted next generation sequencing identifies novel pathogenic variants and provides molecular diagnoses in a cohort of pediatric and adult patients with unexplained mitochondrial dysfunction. Mitochondrion, 2019, 47, 309-317.	1.6	8
4	Follow-up of fatty acid Î²-oxidation disorders in expanded newborn screening era. European Journal of Pediatrics, 2019, 178, 387-394.	1.3	19
5	Molecular and Clinical Investigations on Portuguese Patients with Multiple acyl-CoA Dehydrogenase Deficiency. Current Molecular Medicine, 2019, 19, 487-493.	0.6	6
6	Data supporting the co-expression of PDHA1 gene and of its paralogue PDHA2 in somatic cells of a family. Data in Brief, 2016, 9, 68-77.	0.5	0
7	Complex genetic findings in a female patient with pyruvate dehydrogenase complex deficiency: Null mutations in the PDHX gene associated with unusual expression of the testis-specific PDHA2 gene in her somatic cells. Gene, 2016, 591, 417-424.	1.0	5
8	Functional correction by antisense therapy of a splicing mutation in the GALT gene. European Journal of Human Genetics, 2015, 23, 500-506.	1.4	15
9	A frequent splicing mutation and novel missense mutations color the updated mutational spectrum of classic galactosemia in Portugal. Journal of Inherited Metabolic Disease, 2014, 37, 43-52.	1.7	14
10	Trimethylaminuria (fish odor syndrome): Genotype characterization among Portuguese patients. Gene, 2013, 527, 366-370.	1.0	16
11	Phenylalanine hydroxylase deficiency: Molecular epidemiology and predictable BH4-responsiveness in South Portugal PKU patients. Molecular Genetics and Metabolism, 2011, 104, S86-S92.	0.5	18
12	Incidence of maple syrup urine disease in Portugal. Molecular Genetics and Metabolism, 2010, 100, 385-387.	0.5	18
13	Pyruvate dehydrogenase deficiency: identification of a novel mutation in the PDHA1 gene which responds to amino acid supplementation. European Journal of Pediatrics, 2009, 168, 17-22.	1.3	19
14	Molecular and structural analyses of maple syrup urine disease and identification of a founder mutation in a Portuguese Gypsy community. Molecular Genetics and Metabolism, 2008, 94, 148-156.	0.5	27