Ana M Gaspar

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/9086559/publications.pdf

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14	173	1163117 8	1125743 13
papers	citations	h-index	g-index

14	14	14	314
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	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.	1.1	27
2	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.	2.7	19
3	Follow-up of fatty acid Î²-oxidation disorders in expanded newborn screening era. European Journal of Pediatrics, 2019, 178, 387-394.	2.7	19
4	Incidence of maple syrup urine disease in Portugal. Molecular Genetics and Metabolism, 2010, 100, 385-387.	1.1	18
5	Phenylalanine hydroxylase deficiency: Molecular epidemiology and predictable BH4-responsiveness in South Portugal PKU patients. Molecular Genetics and Metabolism, 2011, 104, S86-S92.	1.1	18
6	Trimethylaminuria (fish odor syndrome): Genotype characterization among Portuguese patients. Gene, 2013, 527, 366-370.	2.2	16
7	Functional correction by antisense therapy of a splicing mutation in the GALT gene. European Journal of Human Genetics, 2015, 23, 500-506.	2.8	15
8	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.	3.6	14
9	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.	3.4	8
10	Molecular and Clinical Investigations on Portuguese Patients with Multiple acyl-CoA Dehydrogenase Deficiency. Current Molecular Medicine, 2019, 19, 487-493.	1.3	6
11	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.	2.2	5
12	Role of RNA in Molecular Diagnosis of MADD Patients. Biomedicines, 2021, 9, 507.	3.2	4
13	Phenylketonuria in Portugal: Genotypeâ€“phenotype correlations using molecular, biochemical, and haplotypic analyses. Molecular Genetics & Genomic Medicine, 2021, 9, e1559.	1.2	4
14	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.	1.0	0