Isabel A Rivera

List of Publications by Year
in descending order

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46	1,544	471371 17	302012 39
papers	citations	h-index	g-index

47	47	47	2163
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Galactokinase deficiency: lessons from the GalNet registry. Genetics in Medicine, 2021, 23, 202-210.	1.1	14
2	Structural and functional impact of clinically relevant E1Î± variants causing pyruvate dehydrogenase complex deficiency. Biochimie, 2021, 183, 78-88.	1.3	10
3	Dihydrolipoamide dehydrogenase, pyruvate oxidation, and acetylation-dependent mechanisms intersecting drug iatrogenesis. Cellular and Molecular Life Sciences, 2021, 78, 7451-7468.	2.4	8
4	Pyruvate dehydrogenase complex deficiency: updating the clinical, metabolic and mutational landscapes in a cohort of Portuguese patients. Orphanet Journal of Rare Diseases, 2020, 15, 298.	1.2	25
5	Darier disease: first molecular study of a Portuguese family. Heliyon, 2019, 5, e02520.	1.4	2
6	The natural history of classic galactosemia: lessons from the GalNet registry. Orphanet Journal of Rare Diseases, 2019, 14, 86.	1.2	84
7	Homocysteine Metabolism in Children and Adolescents: Influence of Age on Plasma Biomarkers and Correspondent Genotype Interactions. Nutrients, 2019, 11, 646.	1.7	18
8	Molecular basis and clinical presentation of classic galactosemia in a Croatian population. Journal of Pediatric Endocrinology and Metabolism, 2018, 31, 71-75.	0.4	4
9	Sweet and sour: an update on classic galactosemia. Journal of Inherited Metabolic Disease, 2017, 40, 325-342.	1.7	92
10	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
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.	1.0	5
12	The spectrum of pyruvate oxidation defects in the diagnosis of mitochondrial disorders. Journal of Inherited Metabolic Disease, 2015, 38, 391-403.	1.7	44
13	Arginine Functionally Improves Clinically Relevant Human Galactose-1-Phosphate Uridylyltransferase (GALT) Variants Expressed in a Prokaryotic Model. JIMD Reports, 2015, 23, 1-6.	0.7	17
14	Small aminothiol compounds improve the function of Arg to Cys variant proteins: effect on the human cystathionine Î²-synthase p.R336C. Human Molecular Genetics, 2015, 24, 7339-7348.	1.4	8
15	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
16	Insights into the Regulatory Domain of Cystathionine Beta-Synthase: Characterization of Six Variant Proteins. Human Mutation, 2014, 35, 1195-1202.	1.1	15
17	Functional and structural impact of the most prevalent missense mutations in classic galactosemia. Molecular Genetics & Genomic Medicine, 2014, 2, 484-496.	0.6	31
18	Retrospective study of the mediumâ€chain acylâ€<scp>CoA</scp> dehydrogenase deficiency in Portugal. Clinical Genetics, 2014, 85, 555-561.	1.0	11

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19	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
20	Reduced response of Cystathionine Betaâ€Synthase (CBS) to Sâ€Adenosylmethionine (SAM): Identification and functional analysis of CBS gene mutations in Homocystinuria patients. Journal of Inherited Metabolic Disease, 2014, 37, 245-254.	1.7	21
21	Protein Arginine Methylation Is More Prone to Inhibition by S-Adenosylhomocysteine than DNA Methylation in Vascular Endothelial Cells. PLoS ONE, 2013, 8, e55483.	1.1	19
22	Asymmetric dimethylarginine in adults with cystathionine Î²-synthase deficiency. Atherosclerosis, 2012, 222, 509-511.	0.4	11
23	Pyruvate dehydrogenase complex: mRNA and protein expression patterns of E1Î± subunit genes in human spermatogenesis. Gene, 2012, 506, 173-178.	1.0	8
24	Demethylation of the Coding Region Triggers the Activation of the Human Testis-Specific PDHA2 Gene in Somatic Tissues. PLoS ONE, 2012, 7, e38076.	1.1	12
25	Cellular hypomethylation is associated with impaired nitric oxide production by cultured human endothelial cells. Amino Acids, 2012, 42, 1903-1911.	1.2	17
26	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
27	Chromatin-Modifying Agents Increase Transcription of CYP46A1, a Key Player in Brain Cholesterol Elimination. Journal of Alzheimer's Disease, 2011, 22, 1209-1221.	1.2	15
28	The TCN2 776C>G polymorphism correlates with vitamin B12 cellular delivery in healthy adult populations. Clinical Biochemistry, 2010, 43, 645-649.	0.8	26
29	Global DNA methylation: comparison of enzymatic- and non-enzymatic-based methods. Clinical Chemistry and Laboratory Medicine, 2010, 48, 1793-1798.	1.4	13
30	Human testis-specific PDHA2 gene: Methylation status of a CpG island in the open reading frame correlates with transcriptional activity. Molecular Genetics and Metabolism, 2010, 99, 425-430.	0.5	11
31	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
32	Molecular genetic analysis of the cystathionine Î²-synthase gene in Portuguese homocystinuria patients: three novel mutations. Clinical Genetics, 2008, 60, 161-163.	1.0	3
33	Association of A313â€ƒG polymorphism (GSTP1*B) in theglutathione-S-transferase P1gene with sporadic Parkinson's disease. European Journal of Neurology, 2007, 14, 156-161.	1.7	35
34	Homocysteine metabolism, hyperhomocysteinaemia and vascular disease: An overview. Journal of Inherited Metabolic Disease, 2006, 29, 3-20.	1.7	254
35	Mutational spectrum of classical galactosaemia in Spain and Portugal. Journal of Inherited Metabolic Disease, 2006, 29, 739-742.	1.7	20
36	Intracellular S-adenosylhomocysteine increased levels are associated with DNA hypomethylation in HUVEC. Journal of Molecular Medicine, 2005, 83, 831-836.	1.7	79

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37	Increased Homocysteine and S-Adenosylhomocysteine Concentrations and DNA Hypomethylation in Vascular Disease. Clinical Chemistry, 2003, 49, 1292-1296.	1.5	365
38	5,10â€Methylenetetrahydrofolate reductase 677Câ†'T and 1298Aâ†'C mutations are genetic determinants of elevated homocysteine. QJM - Monthly Journal of the Association of Physicians, 2003, 96, 297-303.	0.2	93
39	Prokaryotic expression analysis of I269L and R270K mutations of the phenylalanine hydroxylase gene. Gene Function & Disease, 2001, 2, 46-50.	0.3	2
40	The V388M Mutation Results in a Kinetic Variant Form of Phenylalanine Hydroxylase. Molecular Genetics and Metabolism, 2000, 69, 204-212.	0.5	38
41	The Correlation of Genotype and Phenotype in Portuguese Hyperphenylalaninemic Patients. Molecular Genetics and Metabolism, 2000, 69, 195-203.	0.5	14
42	Mutation Analysis of the GCDH Gene in Italian and Portuguese Patients with Glutaric Aciduria Type I. Molecular Genetics and Metabolism, 2000, 71, 535-537.	0.5	20
43	Relative frequency of IVS10nt546 mutation in a Portuguese phenylketonuric population. Human Mutation, 1997, 9, 272-273.	1.1	2
44	Mutation Analysis of phenylketonuria in South and Central Portugal: Prevalence of V388M mutation. Human Mutation, 1995, 6, 192-194.	1.1	9
45	A Single HPLC System for the Evaluation of Purine and Pyrimidine Metabolites in Body Fluids. Advances in Experimental Medicine and Biology, 1991, 309B, 11-14.	0.8	0
46	Deciphering Protein Arginine Methylation in Mammals. , 0, , .		3