Laura Vilarinho

List of Publications by Year
in descending order

50 papers	1,818 citations	448610 19 h-index	312153 41 g-index
52 all docs	52 docs citations	52 times ranked	2544 citing authors

#	Article	IF	Citations
1	Neonatal Screening in Europe Revisited: An ISNS Perspective on the Current State and Developments Since 2010. International Journal of Neonatal Screening, 2021, 7, 15.	1.2	118
2	Parkinsonism and iron deposition in two adult patients with L-2-hydroxiglutaric aciduria. Parkinsonism and Related Disorders, 2021, 86, 45-47.	1.1	1
3	Role of RNA in Molecular Diagnosis of MADD Patients. Biomedicines, 2021, 9, 507.	1.4	4
4	Phenylketonuria in Portugal: Genotypeâ€“phenotype correlations using molecular, biochemical, and haplotypic analyses. Molecular Genetics & Genomic Medicine, 2021, 9, e1559.	0.6	4
5	Impact of iodine supplementation during preconception, pregnancy and lactation on maternal thyroid homeostasis and offspring psychomotor development: protocol of the IodineMinho prospective study. BMC Pregnancy and Childbirth, 2020, 20, 693.	0.9	7
6	NPC1 silent variant induces skipping of exon 11 (p.V562V) and unfolded protein response was found in a specific Niemannâ€Pick type C patient. Molecular Genetics & Genomic Medicine, 2020, 8, e1451.	0.6	10
7	Molecular basis of Leigh syndrome: a current look. Orphanet Journal of Rare Diseases, 2020, 15, 31.	1.2	62
8	TYROSINEMIA TYPE III: A CASE REPORT OF SIBLINGS AND LITERATURE REVIEW. Revista Paulista De Pediatria, 2020, 38, e2018158.	0.4	12
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.	1.6	8
10	Newborn screening for homocystinurias: Recent recommendations versus current practice. Journal of Inherited Metabolic Disease, 2019, 42, 128-139.	1.7	37
11	Follow-up of fatty acid Î²-oxidation disorders in expanded newborn screening era. European Journal of Pediatrics, 2019, 178, 387-394.	1.3	19
12	Diagnosis, management, and follow-up of mitochondrial disorders in childhood: a personalized medicine in the new era of genome sequence. European Journal of Pediatrics, 2019, 178, 21-32.	1.3	9
13	Molecular and Clinical Investigations on Portuguese Patients with Multiple acyl-CoA Dehydrogenase Deficiency. Current Molecular Medicine, 2019, 19, 487-493.	0.6	6
14	Atypical adult-onset methylmalonic acidemia and homocystinuria presenting as hemolytic uremic syndrome. CEN Case Reports, 2018, 7, 73-76.	0.5	5
15	Cystic Fibrosis Newborn Screening in Portugal: PAP Value in Populations with Stringent Rules for Genetic Studies. International Journal of Neonatal Screening, 2018, 4, 22.	1.2	16
16	Clinical, biochemical, molecular, and histological features of 65 Portuguese patients with mitochondrial disorders. Muscle and Nerve, 2017, 56, 868-872.	1.0	6
17	Reply. Muscle and Nerve, 2017, 56, E49.	1.0	0
18	Guidelines for diagnosis and management of the cobalaminâ€related remethylation disorders cblC, cblD, cblE, cblF, cblG, cblJ and MTHFR deficiency. Journal of Inherited Metabolic Disease, 2017, 40, 21-48.	1.7	206

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19	PCR in the Analysis of Clinical Samples: Prenatal and Postnatal Diagnosis of Inborn Errors of Metabolism. Methods in Molecular Biology, 2017, 1620, 213-224.	0.4	2
20	3-Methylcrotonyl-CoA carboxylase deficiency: Mutational spectrum derived from comprehensive newborn screening. Gene, 2016, 594, 203-210.	1.0	20
21	A Novel SUCLA2 Mutation in a Portuguese Child Associated With â€œMildâ€•Methylmalonic Aciduria. Journal of Child Neurology, 2015, 30, 228-232.	0.7	13
22	Clinical presentation and outcome in a series of 88 patients with the cblC defect. Journal of Inherited Metabolic Disease, 2014, 37, 831-840.	1.7	133
23	Syndromes associated with mitochondrial DNA depletion. Italian Journal of Pediatrics, 2014, 40, 34.	1.0	45
24	Trimethylaminuria (fish odor syndrome): Genotype characterization among Portuguese patients. Gene, 2013, 527, 366-370.	1.0	16
25	Mitochondria proteome profiling: A comparative analysis between gel- and gel-free approaches. Talanta, 2013, 115, 277-283.	2.9	12
26	Novel TTC19 mutation in a family with severe psychiatric manifestations and complex III deficiency. Neurogenetics, 2013, 14, 153-160.	0.7	42
27	Rhabdomyolysis as a Presenting Manifestation of Very Long-Chain Acyl-Coenzyme a Dehydrogenase Deficiency. Clinics and Practice, 2013, 3, 58-60.	0.6	14
28	Enhanced interpretation of newborn screening results without analyte cutoff values. Genetics in Medicine, 2012, 14, 648-655.	1.1	117
29	Diagnosis of a patient with a kinetic variant of medium and short-chain 3-hydroxyacyl-CoA dehydrogenase deficiency by newborn screening. Molecular Genetics and Metabolism, 2012, 106, 277-280.	0.5	4
30	Mutations at the flavin binding site of ETF:QO yield a MADD-like severe phenotype in Drosophila. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2012, 1822, 1284-1292.	1.8	14
31	Clinical validation of cutoff target ranges in newborn screening of metabolic disorders by tandem mass spectrometry: A worldwide collaborative project. Genetics in Medicine, 2011, 13, 230-254.	1.1	308
32	Shortâ€chain 3â€hydroxyacylâ€CoA dehydrogenase deficiency: the clinical relevance of an early diagnosis and report of four new cases. Journal of Inherited Metabolic Disease, 2011, 34, 835-842.	1.7	28
33	Infantile-Onset Disorders of Mitochondrial Replication and Protein Synthesis. Journal of Child Neurology, 2011, 26, 866-875.	0.7	10
34	Molecular Investigation of Pediatric Portuguese Patients with Sensorineural Hearing Loss. Genetics Research International, 2011, 2011, 1-5.	2.0	9
35	<i>PAH</i> mutational spectrum: still expanding. Open Journal of Genetics, 2011, 01, 9-12.	0.1	1
36	Cardiomyopathy and Kidney Disease in a Patient with Maternally Inherited Diabetes and Deafness Caused by the 3243A>G Mutation of Mitochondrial DNA. Cardiology, 2010, 115, 71-74.	0.6	10

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37	Four years of expanded newborn screening in Portugal with tandem mass spectrometry. Journal of Inherited Metabolic Disease, 2010, 33, 133-138.	1.7	92
38	Neonatal cholestasis: an uncommon presentation of hyperargininemia. Journal of Inherited Metabolic Disease, 2010, 33, 503-506.	1.7	22
39	Identification of novel L2HGDH gene mutations and update of the pathological spectrum. Journal of Human Genetics, 2010, 55, 55-58.	1.1	6
40	Pediatric Mitochondrial Respiratory Chain Disorders in the Centro Region of Portugal. Pediatric Neurology, 2009, 40, 351-356.	1.0	20
41	Outcome of three cases of untreated maternal glutaric aciduria type I. European Journal of Pediatrics, 2008, 167, 569-573.	1.3	29
42	Spectrum of MMACHC mutations in Italian and Portuguese patients with combined methylmalonic aciduria and homocystinuria, cblC type. Molecular Genetics and Metabolism, 2008, 93, 475-480.	0.5	80
43	cblEType of homocystinuria due to methionine synthase reductase deficiency: Functional correction by minigene expression. Human Mutation, 2005, 25, 239-247.	1.1	44
44	Age related reference values for urine creatine and guanidinoacetic acid concentration in children and adolescents by gas chromatographyâ€“mass spectrometry. Clinica Chimica Acta, 2004, 348, 155-161.	0.5	37
45	Maternally inherited deafness associated with a T1095C mutation in the mDNA. European Journal of Human Genetics, 2001, 9, 147-149.	1.4	68
46	Clinical and molecular studies in three portuguese mtdna t8993g families. Pediatric Neurology, 2000, 22, 29-32.	1.0	5
47	Identification of a novel R21X mutation in the liver-type arginase gene (ARG1) in four Portuguese patients with argininemia. , 1999, 14, 355-356.		15
48	About the â€œPathologicalâ€•Role of the mtDNA T3308C Mutationâ€¦. American Journal of Human Genetics, 1999, 65, 1457-1459.	2.6	30
49	The mitochondrial DNA A3243G mutation in Portugal: clinical and molecular studies in 5 families. Journal of the Neurological Sciences, 1999, 163, 168-174.	0.3	20
50	Mitochondrial DNA Analysis in Ocular Myopathy. European Neurology, 1998, 39, 148-153.	0.6	14