Laura Vilarinho

List of Publications by Citations

50 papers	1,320 citations	17 h-index	35 g-index
52 ext. papers	1,596 ext. citations	3.6 avg, IF	3.84 L-index

#	Paper	IF	Citations
50	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, 230-54	8.1	250
49	Guidelines for diagnosis and management of the cobalamin-related remethylation disorders cblC, cblD, cblE, cblF, cblG, cblJ and MTHFR deficiency. *Journal of Inherited Metabolic Disease*, **2017**, 40, 21-48	5.4	126
48	Clinical presentation and outcome in a series of 88 patients with the cblC defect. *Journal of Inherited Metabolic Disease*, **2014**, 37, 831-40	5.4	98
47	Enhanced interpretation of newborn screening results without analyte cutoff values. *Genetics in Medicine*, **2012**, 14, 648-55	8.1	93
46	Four years of expanded newborn screening in Portugal with tandem mass spectrometry. *Journal of Inherited Metabolic Disease*, **2010**, 33 Suppl 3, S133-8	5.4	77
45	Spectrum of MMACHC mutations in Italian and Portuguese patients with combined methylmalonic aciduria and homocystinuria, cblC type. *Molecular Genetics and Metabolism*, **2008**, 93, 475-80	3.7	73
44	Maternally inherited deafness associated with a T1095C mutation in the mDNA. *European Journal of Human Genetics*, **2001**, 9, 147-9	5.3	60
43	Syndromes associated with mitochondrial DNA depletion. *Italian Journal of Pediatrics*, **2014**, 40, 34	3.2	37
42	cblE type of homocystinuria due to methionine synthase reductase deficiency: functional correction by minigene expression. *Human Mutation*, **2005**, 25, 239-47	4.7	35
41	Novel TTC19 mutation in a family with severe psychiatric manifestations and complex III deficiency. *Neurogenetics*, **2013**, 14, 153-60	3	34
40	Neonatal Screening in Europe Revisited: An ISNS Perspective on the Current State and Developments Since 2010. *International Journal of Neonatal Screening*, **2021**, 7,	2.6	34
39	Age related reference values for urine creatine and guanidinoacetic acid concentration in children and adolescents by gas chromatography-mass spectrometry. *Clinica Chimica Acta*, **2004**, 348, 155-61	6.2	33
38	Outcome of three cases of untreated maternal glutaric aciduria type I. *European Journal of Pediatrics*, **2008**, 167, 569-73	4.1	27
37	About the "Pathological" role of the mtDNA T3308C mutationellipsis. *American Journal of Human Genetics*, **1999**, 65, 1457-9	11	26
36	Molecular basis of Leigh syndrome: a current look. *Orphanet Journal of Rare Diseases*, **2020**, 15, 31	4.2	25
35	Newborn screening for homocystinurias: Recent recommendations versus current practice. *Journal of Inherited Metabolic Disease*, **2019**, 42, 128-139	5.4	24
34	Short-chain 3-hydroxyacyl-CoA dehydrogenase deficiency: the clinical relevance of an early diagnosis and report of four new cases. *Journal of Inherited Metabolic Disease*, **2011**, 34, 835-42	5.4	23

33	Pediatric mitochondrial respiratory chain disorders in the Centro region of Portugal. *Pediatric Neurology*, **2009**, 40, 351-6	2.9	17
32	The mitochondrial DNA A3243G mutation in Portugal: clinical and molecular studies in 5 families. *Journal of the Neurological Sciences*, **1999**, 163, 168-74	3.2	17
31	Neonatal cholestasis: an uncommon presentation of hyperargininemia. *Journal of Inherited Metabolic Disease*, **2010**, 33 Suppl 3, S503-6	5.4	16
30	Trimethylaminuria (fish odor syndrome): genotype characterization among Portuguese patients. *Gene*, **2013**, 527, 366-70	3.8	14
29	Mitochondrial DNA analysis in ocular myopathy. Observations in 29 Portuguese patients. *European Neurology*, **1998**, 39, 148-53	2.1	14
28	Identification of a novel R21X mutation in the liver-type arginase gene (ARG1) in four Portuguese patients with argininemia. *Human Mutation*, **1999**, 14, 355-6	4.7	14
27	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, 1284-92	6.9	12
26	Follow-up of fatty acid β-oxidation disorders in expanded newborn screening era. *European Journal of Pediatrics*, **2019**, 178, 387-394	4.1	12
25	A novel SUCLA2 mutation in a Portuguese child associated with "mild" methylmalonic aciduria. *Journal of Child Neurology*, **2015**, 30, 228-32	2.5	11
24	Mitochondria proteome profiling: a comparative analysis between gel- and gel-free approaches. *Talanta*, **2013**, 115, 277-83	6.2	11
23	3-Methylcrotonyl-CoA carboxylase deficiency: Mutational spectrum derived from comprehensive newborn screening. *Gene*, **2016**, 594, 203-210	3.8	10
22	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, 71-4	1.6	10
21	Infantile-onset disorders of mitochondrial replication and protein synthesis. *Journal of Child Neurology*, **2011**, 26, 866-75	2.5	9
20	Molecular investigation of pediatric portuguese patients with sensorineural hearing loss. *Genetics Research International*, **2011**, 2011, 587602	0	9
19	Cystic Fibrosis Newborn Screening in Portugal: PAP Value in Populations with Stringent Rules for Genetic Studies. *International Journal of Neonatal Screening*, **2018**, 4, 22	2.6	9
18	Rhabdomyolysis as a presenting manifestation of very long-chain acyl-coenzyme a dehydrogenase deficiency. *Clinics and Practice*, **2013**, 3, e22	2.4	8
17	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	4.1	7
16	Clinical, biochemical, molecular, and histological features of 65 Portuguese patients with mitochondrial disorders. *Muscle and Nerve*, **2017**, 56, 868-872	3.4	5

15	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	4.9	5
14	Identification of novel L2HGDH gene mutations and update of the pathological spectrum. *Journal of Human Genetics*, **2010**, 55, 55-8	4.3	5
13	Clinical and molecular studies in three Portuguese mtDNA T8993G families. *Pediatric Neurology*, **2000**, 22, 29-32	2.9	4
12	TYROSINEMIA TYPE III: A CASE REPORT OF SIBLINGS AND LITERATURE REVIEW. *Revista Paulista De Pediatria*, **2020**, 38, e2018158	1.2	4
11	Molecular and Clinical Investigations on Portuguese Patients with Multiple acyl-CoA Dehydrogenase Deficiency. *Current Molecular Medicine*, **2019**, 19, 487-493	2.5	4
10	NPC1 silent variant induces skipping of exon 11 (p.V562V) and unfolded protein response was found in a specific Niemann-Pick type C patient. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1451	2.3	4
9	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, 277-80	3.7	3
8	Role of RNA in Molecular Diagnosis of MADD Patients. *Biomedicines*, **2021**, 9,	4.8	2
7	Atypical adult-onset methylmalonic acidemia and homocystinuria presenting as hemolytic uremic syndrome. *CEN Case Reports*, **2018**, 7, 73-76	1	1
6	<i>PAH</i> mutational spectrum: still expanding. *Open Journal of Genetics*, **2011**, 01, 9-12	0.2	1
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	3.2	1
4	PCR in the Analysis of Clinical Samples: Prenatal and Postnatal Diagnosis of Inborn Errors of Metabolism. *Methods in Molecular Biology*, **2017**, 1620, 213-224	1.4	0
3	Parkinsonism and iron deposition in two adult patients with L-2-hydroxiglutaric aciduria. *Parkinsonism and Related Disorders*, **2021**, 86, 45-47	3.6	0
2	Phenylketonuria in Portugal: Genotype-phenotype correlations using molecular, biochemical, and haplotypic analyses. *Molecular Genetics & Genomic Medicine*, **2021**, 9, e1559	2.3	0
1	Reply. *Muscle and Nerve*, **2017**, 56, E49	3.4	