Esmeralda G Martins

List of Publications by Year
in descending order

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47 papers	875 citations	643344 15 h-index	591227 27 g-index
48 all docs	48 docs citations	48 times ranked	1325 citing authors

#	Article	IF	Citations
1	Dyslipidemia Diagnosis and Treatment: Risk Stratification in Children and Adolescents. Journal of Nutrition and Metabolism, 2022, 2022, 1-10.	0.7	13
2	Glutaric Aciduria Type 2 Presenting in Adult Life With Hypoglycemia and Encephalopathic Hyperammonemia. Journal of Medical Cases, 2022, 13, 56-60.	0.4	2
3	Long-term efficacy and safety of vestronidase alfa enzyme replacement therapy in pediatric subjects < 5 years with mucopolysaccharidosis VII. Molecular Genetics and Metabolism, 2022, 136, 28-37.	0.5	4
4	Congenital Disorders of Glycosylation in Portugalâ€"Two Decades of Experience. Journal of Pediatrics, 2021, 231, 148-156.	0.9	9
5	Implementing a Transition Program from Paediatric to Adult Services in Phenylketonuria: Results after Two Years of Follow-Up with an Adult Team. Nutrients, 2021, 13, 799.	1.7	10
6	Continuous use of glycomacropeptide in the nutritional management of patients with phenylketonuria: a clinical perspective. Orphanet Journal of Rare Diseases, 2021, 16, 84.	1.2	11
7	SLC35A2-CDG: Novel variant and review. Molecular Genetics and Metabolism Reports, 2021, 26, 100717.	0.4	15
8	Quantitative analysis of the natural history of prolidase deficiency: description of 17 families and systematic review of published cases. Genetics in Medicine, 2021, 23, 1604-1615.	1.1	10
9	Utility of Gene Panels for the Diagnosis of Inborn Errors of Metabolism in a Metabolic Reference Center. Genes, 2021, 12, 1262.	1.0	6
10	Metabolic Control of Patients with Phenylketonuria in a Portuguese Metabolic Centre Comparing Three Different Recommendations. Nutrients, 2021, 13, 3118.	1.7	14
11	Phenylketonuria in Portugal: Genotypeâ€"phenotype correlations using molecular, biochemical, and haplotypic analyses. Molecular Genetics & Genomic Medicine, 2021, 9, e1559.	0.6	4
12	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
13	Assessing Lysosomal Disorders in the NGS Era: Identification of Novel Rare Variants. International Journal of Molecular Sciences, 2020, 21, 6355.	1.8	8
14	Neonatal Cholestasis Over Time: Changes in Epidemiology and Outcome in a Cohort of 154 Patients From a Portuguese Tertiary Center. Frontiers in Pediatrics, 2020, 8, 351.	0.9	7
15	The long-term safety and efficacy of vestronidase alfa, rhGUS enzyme replacement therapy, in subjects with mucopolysaccharidosis VII. Molecular Genetics and Metabolism, 2020, 129, 219-227.	0.5	19
16	Consensus guideline for the diagnosis and management of mannose phosphate isomeraseâ€congenital disorder of glycosylation. Journal of Inherited Metabolic Disease, 2020, 43, 671-693.	1.7	40
17	TYROSINEMIA TYPE III: A CASE REPORT OF SIBLINGS AND LITERATURE REVIEW. Revista Paulista De Pediatria, 2020, 38, e2018158.	0.4	12
18	Ironâ€sulfur cluster ISD11 deficiency (LYRM4 gene) presenting as cardiorespiratory arrest and 3â€methylglutaconic aciduria. JIMD Reports, 2019, 49, 11-16.	0.7	6

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19	Lipid Antigen Presentation by CD1b and CD1d in Lysosomal Storage Disease Patients. Frontiers in Immunology, 2019, 10, 1264.	2.2	10
20	The European Phenylketonuria Guidelines and the challenges on management practices in Portugal. Journal of Pediatric Endocrinology and Metabolism, 2019, 32, 623-629.	0.4	6
21	Over Restriction of Dietary Protein Allowance: The Importance of Ongoing Reassessment of Natural Protein Tolerance in Phenylketonuria. Nutrients, 2019, 11, 995.	1.7	13
22	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
23	Phenotype, treatment practice and outcome in the cobalaminâ€dependent remethylation disorders and MTHFR deficiency: Data from the Eâ€HOD registry. Journal of Inherited Metabolic Disease, 2019, 42, 333-352.	1.7	53
24	Fatty Liver Caused by Glycogen Storage Disease Type IX: A Small Series of Cases in Children. GE Portuguese Journal of Gastroenterology, 2019, 26, 430-437.	0.3	3
25	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
26	Molecular and Clinical Investigations on Portuguese Patients with Multiple acyl-CoA Dehydrogenase Deficiency. Current Molecular Medicine, 2019, 19, 487-493.	0.6	6
27	Early onset lysosomal acid lipase deficiency presenting as secondary hemophagocytic lymphohistiocytosis: Two infants treated with sebelipase alfa. Clinics and Research in Hepatology and Gastroenterology, 2018, 42, e77-e82.	0.7	16
28	Metabolic Control in Patients With Phenylketonuria Pre- and Post-Sapropterin Loading Test. FIRE Forum for International Research in Education, 2018, 6, 232640981878889.	0.7	2
29	Clinical practices among healthcare professionals concerning neonatal jaundice and pale stools. European Journal of Pediatrics, 2017, 176, 361-369.	1.3	8
30	Symmetric asymptomatic reticular lesions of the skin. Journal of Paediatrics and Child Health, 2017, 53, 1024-1024.	0.4	0
31	D-Bifunctional Protein Deficiency: A Cause of Neonatal Onset Seizures and Hypotonia. Pediatric Neurology, 2015, 52, 539-543.	1.0	12
32	Infantile Refsum Disease: Influence of Dietary Treatment on Plasma Phytanic Acid Levels. JIMD Reports, 2015, 26, 53-60.	0.7	7
33	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
34	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
35	Newborn screening for medium-chain acyl-CoA dehydrogenase deficiency: regional experience and high incidence of carnitine deficiency. Orphanet Journal of Rare Diseases, 2013, 8, 102.	1.2	18
36	Trimethylaminuria (fish odor syndrome): Genotype characterization among Portuguese patients. Gene, 2013, 527, 366-370.	1.0	16

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37	MAN1B1 Deficiency: An Unexpected CDG-II. PLoS Genetics, 2013, 9, e1003989.	1.5	63
38	Relative frequency of known causes of multiple mtDNA deletions: Two novel POLG mutations. Neuromuscular Disorders, 2011, 21, 483-488.	0.3	16
39	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
40	Incidence of maple syrup urine disease in Portugal. Molecular Genetics and Metabolism, 2010, 100, 385-387.	0.5	18
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	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
44	Maple syrup disease presenting as paroxysmal dystonia. Annals of Neurology, 2004, 56, 749-750.	2.8	16
45	The Correlation of Genotype and Phenotype in Portuguese Hyperphenylalaninemic Patients. Molecular Genetics and Metabolism, 2000, 69, 195-203.	0.5	14
46	Identification of a novel R21X mutation in the liver-type arginase gene (ARG1) in four Portuguese patients with argininemia. , 1999, 14, 355-356.		15
47	Sialuria in a Portuguese Girl: Clinical, Biochemical, and Molecular Characteristics. Molecular Genetics and Metabolism, 1999, 67, 131-137.	0.5	20