Ermelinda Santos Silva

List of Publications by Year
in descending order

29 papers	555 citations	840776 11 h-index	642732 23 g-index
31 all docs	31 docs citations	31 times ranked	843 citing authors

#	Article	IF	Citations
1	Mutations in NOTCH1 Cause Adams-Oliver Syndrome. American Journal of Human Genetics, 2014, 95, 275-284.	6.2	150
2	Successful medical treatment of severely decompensated Wilson disease. Journal of Pediatrics, 1996, 128, 285-287.	1.8	82
3	Demonstration of McCune-Albright mutations in the liver of children with high Î³GT progressive cholestasis. Journal of Hepatology, 2000, 32, 154-158.	3.7	49
4	ORTHOTOPIC LIVER TRANSPLANTATION FOR CRIGLER-NAJJAR TYPE I DISEASE IN SIX CHILDREN. Transplantation, 1995, 60, 1095-1098.	1.0	45
5	Analysis of the UDP-glucuronosyltransferase gene in Portuguese patients with a clinical diagnosis of Gilbert and Criglerâ€“Najjar syndromes. Blood Cells, Molecules, and Diseases, 2006, 36, 91-97.	1.4	28
6	Neonatal cholestasis: an uncommon presentation of hyperargininemia. Journal of Inherited Metabolic Disease, 2010, 33, 503-506.	3.6	22
7	A case of hepatopulmonary syndrome solved by mycophenolate mofetil (an inhibitor of angiogenesis) Tj ETQq1 1 0.784314 rgBT /Ove	3.7	21
8	European lipodystrophy registry: background and structure. Orphanet Journal of Rare Diseases, 2020, 15, 17.	2.7	21
9	Liver transplantation in a case of argininaemia. Journal of Inherited Metabolic Disease, 2001, 24, 885-887.	3.6	16
10	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.	1.5	16
11	Metabolic liver diseases presenting with neonatal cholestasis: at the crossroad between old and new paradigms. European Journal of Pediatrics, 2019, 178, 515-523.	2.7	12
12	Lethal dilated cardiomyopathy due to long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency. Journal of Inherited Metabolic Disease, 1996, 19, 373-374.	3.6	10
13	Insights Into Pediatric Autoimmune Gastritis. Journal of Pediatric Gastroenterology and Nutrition, 2019, 68, e99-e104.	1.8	9
14	Childhood Fructoholism and Fructoholic Liver Disease. Hepatology Communications, 2019, 3, 44-51.	4.3	9
15	Clinical practices among healthcare professionals concerning neonatal jaundice and pale stools. European Journal of Pediatrics, 2017, 176, 361-369.	2.7	8
16	Symmetrical Enchondromatosis of the Hands and Feet in Two Sisters. Journal of Pediatric Orthopaedics Part B, 1997, 6, 15-19.	0.6	7
17	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.	1.9	7
18	Hyperinsulinaemic Hypoglycaemia and Polycystic Kidney Disease â€“ A Rare Case Concerning PMM2 Gene Pleiotropy. European Endocrinology, 2020, 16, 66.	1.5	7

#	Article	IF	Citations
19	Elevation of gamma-glutamyl transferase in adult: Should we think about progressive familiar intrahepatic cholestasis?. Digestive and Liver Disease, 2016, 48, 203-205.	0.9	6
20	Chronic Inflammatory Demyelinating Polyneuropathy Associated With Autoimmune Hepatitis. Pediatric Neurology, 2014, 51, e13-e14.	2.1	5
21	Adamsâ€“Oliver syndrome and portal hypertension: Fortuitous association or common mechanism?. American Journal of Medical Genetics, Part A, 2012, 158A, 648-651.	1.2	4
22	Fatty Liver and Autoimmune Hepatitis: Two Forms of Liver Involvement in Lipodystrophies. GE Portuguese Journal of Gastroenterology, 2019, 26, 362-369.	0.8	4
23	The Stool Color Card as a Screening Tool for Biliary Atresia in the Digital Version of the Portuguese Child and Youth Health Booklet. Acta Medica Portuguesa, 2021, 34, 632-633.	0.4	4
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.	0.8	3
25	Neonatal cholestasis: development of a diagnostic decision algorithm from multivariate predictive models. European Journal of Pediatrics, 2021, 180, 1477-1486.	2.7	2
26	Alpha-1-Antitrypsin Deficiency Presenting as Neonatal Cholestasis: Predictors of Outcome and Effect of Ursodeoxycholic Acid. Journal of Liver, 2015, 04, .	0.3	2
27	DoenÃ§a hepÃ¡tica autoimune na crianÃ§a e no adolescente - dificuldades no diagnÃ³stico. GE Jornal PortuguÃªs De Gastrenterologia, 2012, 19, 229-240.	0.0	1
28	A New Mutation Causing Progressive Familiar Intrahepatic Cholestasis Type 3 in Association with Autoimmune Hepatitis. European Journal of Case Reports in Internal Medicine, 2017, 4, 000537.	0.4	1
29	Incidental Liver Lesions in children: a practical and evidence-based approach. Clinics and Research in Hepatology and Gastroenterology, 2022, , 101904.	1.5	0