Emmanuel Gonzales

List of Publications by Year
in descending order

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51 papers	2,079 citations	346980 22 h-index	274796 44 g-index
51 all docs	51 docs citations	51 times ranked	2026 citing authors

#	Article	IF	Citations
1	Galaxy Is a Suitable Bioinformatics Platform for the Molecular Diagnosis of Human Genetic Disorders Using High-Throughput Sequencing Data Analysis: Five Years of Experience in a Clinical Laboratory. Clinical Chemistry, 2022, 68, 313-321.	1.5	7
2	Spontaneous evolution patterns of focal congenital hepatic hemangiomas: a case series of 25 patients. Pediatric Radiology, 2022, 52, 1048-1060.	1.1	4
3	Targeted-Capture Next-Generation Sequencing in Diagnosis Approach of Pediatric Cholestasis. Diagnostics, 2022, 12, 1169.	1.3	5
4	Modern therapeutic approaches to liver-related disorders. Journal of Hepatology, 2022, 76, 1392-1409.	1.8	22
5	Odevixibat treatment in progressive familial intrahepatic cholestasis: a randomised, placebo-controlled, phase 3 trial. The Lancet Gastroenterology and Hepatology, 2022, 7, 830-842.	3.7	54
6	Long term results of liver transplantation for alpha-1 antitrypsin deficiency. Digestive and Liver Disease, 2021, 53, 606-611.	0.4	17
7	Pharmacological Premature Termination Codon Readthrough of ABCB11 in Bile Salt Export Pump Deficiency: An In Vitro Study. Hepatology, 2021, 73, 1449-1463.	3.6	12
8	Cardiovascular disorders in patients with congenital portosystemic shunts: 23 years of experience in a tertiary referral centre. Archives of Cardiovascular Diseases, 2021, 114, 221-231.	0.7	11
9	Adenosine kinase deficiency: Three new cases and diagnostic value of hypermethioninemia. Molecular Genetics and Metabolism, 2021, 132, 38-43.	0.5	8
10	Long-term results of pediatric liver transplantation for autoimmune liver disease. Clinics and Research in Hepatology and Gastroenterology, 2021, 45, 101537.	0.7	6
11	Molecular Regulation of Canalicular ABC Transporters. International Journal of Molecular Sciences, 2021, 22, 2113.	1.8	13
12	Effect of CFTR correctors on the traffic and the function of intracellularly retained ABCB4 variants. Liver International, 2021, 41, 1344-1357.	1.9	4
13	Integrated Genomic Analysis Identifies Driver Genes and Cisplatin-Resistant Progenitor Phenotype in Pediatric Liver Cancer. Cancer Discovery, 2021, 11, 2524-2543.	7.7	41
14	RAB10 Interacts with ABCB4 and Regulates Its Intracellular Traffic. International Journal of Molecular Sciences, 2021, 22, 7087.	1.8	3
15	Defining the natural history of rare genetic liver diseases: Lessons learned from the NAPPED initiative. European Journal of Medical Genetics, 2021, 64, 104245.	0.7	5
16	Impact of Genotype, Serum Bile Acids, and Surgical Biliary Diversion on Native Liver Survival in FIC1 Deficiency. Hepatology, 2021, 74, 892-906.	3.6	25
17	Effects of odevixibat on pruritus and bile acids in children with cholestatic liver disease: Phase 2 study. Clinics and Research in Hepatology and Gastroenterology, 2021, 45, 101751.	0.7	40
18	Efficacy and safety of maralixibat treatment in patients with Alagille syndrome and cholestatic pruritus (ICONIC): a randomised phase 2 study. Lancet, The, 2021, 398, 1581-1592.	6.3	77

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19	In vitro functional rescue by ivacaftor of an ABCB11 variant involved in PFIC2 and intrahepatic cholestasis of pregnancy. Orphanet Journal of Rare Diseases, 2021, 16, 484.	1.2	3
20	Alagille Syndrome and Chronic Arthritis: An International Case Series. Journal of Pediatrics, 2020, 218, 228-230.e1.	0.9	5
21	Defining clinical subgroups and genotypeâ€“phenotype correlations in NBAS-associated disease across 110 patients. Genetics in Medicine, 2020, 22, 610-621.	1.1	46
22	Endoscopic Retrograde Cholangiopancreatography in Infants. Journal of Pediatric Gastroenterology and Nutrition, 2020, 71, e54-e58.	0.9	4
23	Glycerol Phenylbutyrate Therapy in Progressive Familial Intrahepatic Cholestasis Type 2. Journal of Pediatric Gastroenterology and Nutrition, 2020, 70, e139-e140.	0.9	1
24	Functional rescue of an ABCB11 mutant by ivacaftor: A new targeted pharmacotherapy approach in bile salt export pump deficiency. Liver International, 2020, 40, 1917-1925.	1.9	25
25	Genotype correlates with the natural history of severe bile salt export pump deficiency. Journal of Hepatology, 2020, 73, 84-93.	1.8	61
26	Congenital portosystemic shunts. Clinics and Research in Hepatology and Gastroenterology, 2020, 44, 452-459.	0.7	10
27	Successful Treatment with Rituximab and Immunoadsorption for an Auto-Antibody Induced Bile Salt Export Pump Deficiency in a Liver Transplanted Patient. Pediatric Gastroenterology, Hepatology and Nutrition, 2020, 23, 174.	0.4	4
28	Congenital Portosystemic Shunts. Journal of Pediatric Gastroenterology and Nutrition, 2019, 68, 615-622.	0.9	37
29	mTOR inhibitors in pediatric liver transplant recipients. Clinics and Research in Hepatology and Gastroenterology, 2019, 43, 403-409.	0.7	11
30	Loss-of-Function Mutations in UNC45A Cause a Syndrome Associating Cholestasis, Diarrhea, Impaired Hearing, and Bone Fragility. American Journal of Human Genetics, 2018, 102, 364-374.	2.6	40
31	Congenital portosystemic shunts: diagnosis and treatment. Abdominal Radiology, 2018, 43, 2023-2036.	1.0	55
32	Cholic acid for primary bile acid synthesis defects: a life-saving therapy allowing a favorable outcome in adulthood. Orphanet Journal of Rare Diseases, 2018, 13, 190.	1.2	34
33	Cholic Acid to Treat HSD3B7 and AKR1D1 Deficiencies. Journal of Pediatric Gastroenterology and Nutrition, 2017, 65, e134.	0.9	3
34	MYO5B mutations cause cholestasis with normal serum gammaâ€glutamyl transferase activity in children without microvillous inclusion disease. Hepatology, 2017, 65, 164-173.	3.6	117
35	DCDC2Mutations Cause Neonatal Sclerosing Cholangitis. Human Mutation, 2016, 37, 1025-1029.	1.1	56
36	Long-term successful liverâ€“kidney transplantation in a child with atypical hemolytic uremic syndrome caused by homozygous factor H deficiency. Pediatric Nephrology, 2016, 31, 2375-2378.	0.9	3

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37	Cholestasis Reveals Severe Cortisol Deficiency in Neonatal Pituitary Stalk Interruption Syndrome. PLoS ONE, 2016, 11, e0147750.	1.1	15
38	Targeted pharmacotherapy in progressive familial intrahepatic cholestasis type 2: Evidence for improvement of cholestasis with 4â€phenylbutyrate. Hepatology, 2015, 62, 558-566.	3.6	90
39	Clinical utility gene card for: Progressive familial intrahepatic cholestasis type 1. European Journal of Human Genetics, 2014, 22, 572-572.	1.4	7
40	Clinical utility gene card for: Progressive familial intrahepatic cholestasis type 2. European Journal of Human Genetics, 2014, 22, 572-572.	1.4	4
41	Clinical utility gene card for: Progressive familial intrahepatic cholestasis type 3. European Journal of Human Genetics, 2014, 22, 572-572.	1.4	5
42	Liver transcript analysis reveals aberrant splicing due to silent and intronic variations in the ABCB11 gene. Molecular Genetics and Metabolism, 2014, 113, 225-229.	0.5	12
43	Successful mutation-specific chaperone therapy with 4-phenylbutyrate in a child with progressive familial intrahepatic cholestasis type 2. Journal of Hepatology, 2012, 57, 695-698.	1.8	81
44	Primary bile acid therapy during pregnancy in patients with 3Î²â€hydroxyâ€Î””⁵â€₂₇â€steroid oxidoreductase deficiency. Pediatrics International, 2011, 53, 792-792.	0.2	0
45	ATP8B1 and ABCB11 analysis in 62 children with normal gamma-glutamyl transferase progressive familial intrahepatic cholestasis (PFIC): Phenotypic differences between PFIC1 and PFIC2 and natural history. Hepatology, 2010, 51, 1645-1655.	3.6	236
46	The Spectrum of Liver Diseases Related toABCB4Gene Mutations: Pathophysiology and Clinical Aspects. Seminars in Liver Disease, 2010, 30, 134-146.	1.8	212
47	Mutation specific drug therapy for progressive familial or benign recurrent intrahepatic cholestasis: A new tool in a near future?. Journal of Hepatology, 2010, 53, 385-387.	1.8	16
48	Relapsing features of bile salt export pump deficiency after liver transplantation in two patients with progressive familial intrahepatic cholestasis type 2. Journal of Hepatology, 2010, 53, 981-986.	1.8	72
49	Oral Cholic Acid for Hereditary Defects of Primary Bile Acid Synthesis: A Safe and Effective Long-term Therapy. Gastroenterology, 2009, 137, 1310-1320.e3.	0.6	91
50	Progressive familial intrahepatic cholestasis. Orphanet Journal of Rare Diseases, 2009, 4, 1.	1.2	308
51	Liver diseases related to MDR3 (ABCB4) gene deficiency. Frontiers in Bioscience - Landmark, 2009, Volume, 4242.	3.0	61