Emmanuel Jacquemin

List of Publications by Citations

70 papers	3,029 citations	28 h-index	55 g-index
71 ext. papers	3,611 ext. citations	6.3 avg, IF	4.98 L-index

#	Paper	IF	Citations
70	The wide spectrum of multidrug resistance 3 deficiency: from neonatal cholestasis to cirrhosis of adulthood. *Gastroenterology*, **2001**, 120, 1448-58	13.3	394
69	Heterozygous non-sense mutation of the MDR3 gene in familial intrahepatic cholestasis of pregnancy. *Lancet, The*, **1999**, 353, 210-1	40	297
68	Progressive familial intrahepatic cholestasis. *Orphanet Journal of Rare Diseases*, **2009**, 4, 1	4.2	229
67	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, 1645-55	11.2	188
66	The spectrum of liver diseases related to ABCB4 gene mutations: pathophysiology and clinical aspects. *Seminars in Liver Disease*, **2010**, 30, 134-46	7.3	175
65	Progressive familial intrahepatic cholestasis. *Clinics and Research in Hepatology and Gastroenterology*, **2012**, 36 Suppl 1, S26-35	2.4	152
64	Differences in presentation and progression between severe FIC1 and BSEP deficiencies. *Journal of Hepatology*, **2010**, 53, 170-8	13.4	145
63	MYO5B mutations cause cholestasis with normal serum gamma-glutamyl transferase activity in children without microvillous inclusion disease. *Hepatology*, **2017**, 65, 164-173	11.2	84
62	Molecular genetics of 3beta-hydroxy-Delta5-C27-steroid oxidoreductase deficiency in 16 patients with loss of bile acid synthesis and liver disease. *Journal of Clinical Endocrinology and Metabolism*, **2003**, 88, 1833-41	5.6	83
61	MYO5B and bile salt export pump contribute to cholestatic liver disorder in microvillous inclusion disease. *Hepatology*, **2014**, 60, 301-10	11.2	80
60	Long term results of liver transplantation for Wilson's disease: experience in France. *Journal of Hepatology*, **2014**, 60, 579-89	13.4	78
59	Targeted pharmacotherapy in progressive familial intrahepatic cholestasis type 2: Evidence for improvement of cholestasis with 4-phenylbutyrate. *Hepatology*, **2015**, 62, 558-66	11.2	75
58	Oral cholic acid for hereditary defects of primary bile acid synthesis: a safe and effective long-term therapy. *Gastroenterology*, **2009**, 137, 1310-1320.e1-3	13.3	74
57	Successful mutation-specific chaperone therapy with 4-phenylbutyrate in a child with progressive familial intrahepatic cholestasis type 2. *Journal of Hepatology*, **2012**, 57, 695-8	13.4	71
56	Progressive familial intrahepatic cholestasis. Genetic basis and treatment. *Clinics in Liver Disease*, **2000**, 4, 753-63	4.6	67
55	Haploinsufficiency of the NOTCH1 Receptor as a Cause of Adams-Oliver Syndrome With Variable Cardiac Anomalies. *Circulation: Cardiovascular Genetics*, **2015**, 8, 572-581		65
54	Altered hepatobiliary gene expressions in PFIC1: ATP8B1 gene defect is associated with CFTR downregulation. *Hepatology*, **2006**, 43, 1125-34	11.2	58

53	A functional classification of ABCB4 variations causing progressive familial intrahepatic cholestasis type 3. *Hepatology*, **2016**, 63, 1620-31	11.2	53
52	SRD5B1 (AKR1D1) gene analysis in delta(4)-3-oxosteroid 5beta-reductase deficiency: evidence for primary genetic defect. *Journal of Hepatology*, **2004**, 40, 716-8	13.4	51
51	A missense mutation in ABCB4 gene involved in progressive familial intrahepatic cholestasis type 3 leads to a folding defect that can be rescued by low temperature. *Hepatology*, **2009**, 49, 1218-27	11.2	47
50	Inherited IL-18BP deficiency in human fulminant viral hepatitis. *Journal of Experimental Medicine*, **2019**, 216, 1777-1790	16.6	42
49	Adams-Oliver syndrome and hepatoportal sclerosis: occasional association or common mechanism?. *American Journal of Medical Genetics, Part A*, **2005**, 135, 186-9	2.5	41
48	Progressive familial intrahepatic cholestasis. *Journal of Gastroenterology and Hepatology (Australia)*, **1999**, 14, 594-9	4	36
47	Obliterative portal venopathy: a study of 48 children. *Journal of Pediatrics*, **2014**, 165, 190-193.e2	3.6	33
46	Phase I/II Trial of Liver-derived Mesenchymal Stem Cells in Pediatric Liver-based Metabolic Disorders: A Prospective, Open Label, Multicenter, Partially Randomized, Safety Study of One Cycle of Heterologous Human Adult Liver-derived Progenitor Cells (HepaStem) in Urea Cycle Disorders and Crigler-Najjar Syndrome Patients. *Transplantation*, **2019**, 103, 1903-1915	1.8	33
45	DCDC2 Mutations Cause Neonatal Sclerosing Cholangitis. *Human Mutation*, **2016**, 37, 1025-9	4.7	32
44	Longterm Risk of Solid Organ De Novo Malignancies After Liver Transplantation: A French National Study on 11,226 Patients. *Liver Transplantation*, **2018**, 24, 1425-1436	4.5	31
43	Functional defect of variants in the adenosine triphosphate-binding sites of ABCB4 and their rescue by the cystic fibrosis transmembrane conductance regulator potentiator, ivacaftor (VX-770). *Hepatology*, **2017**, 65, 560-570	11.2	29
42	Management of Biliary Atresia in France 1986 to 2015: Long-term Results. *Journal of Pediatric Gastroenterology and Nutrition*, **2019**, 69, 416-424	2.8	25
41	Genotype correlates with the natural history of severe bile salt export pump deficiency. *Journal of Hepatology*, **2020**, 73, 84-93	13.4	22
40	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	4.2	21
39	Liver disease related to alpha1-antitrypsin deficiency in French children: The DEFI-ALPHA cohort. *Liver International*, **2019**, 39, 1136-1146	7.9	19
38	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	7.9	17
37	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	40	17
36	Rapid and reliable diagnosis of Wilson disease using X-ray fluorescence. *Journal of Pathology: Clinical Research*, **2016**, 2, 175-86	5.3	15

35	Attempt to Determine the Prevalence of Two Inborn Errors of Primary Bile Acid Synthesis: Results of a European Survey. *Journal of Pediatric Gastroenterology and Nutrition*, **2017**, 64, 864-868	2.8	14
34	Mutations in the novel gene FOPV are associated with familial autosomal dominant and non-familial obliterative portal venopathy. *Liver International*, **2018**, 38, 358-364	7.9	14
33	Albumin liver dialysis as pregnancy-saving procedure in cholestatic liver disease and intractable pruritus. *World Journal of Gastroenterology*, **2008**, 14, 6572-4	5.6	12
32	Bile Acid Synthesis Disorders in Arabs: A 10-year Screening Study. *Journal of Pediatric Gastroenterology and Nutrition*, **2017**, 65, 613-620	2.8	11
31	Liver transcript analysis reveals aberrant splicing due to silent and intronic variations in the ABCB11 gene. *Molecular Genetics and Metabolism*, **2014**, 113, 225-9	3.7	10
30	Cholestasis Reveals Severe Cortisol Deficiency in Neonatal Pituitary Stalk Interruption Syndrome. *PLoS ONE*, **2016**, 11, e0147750	3.7	10
29	Arthrogryposis, renal dysfunction, and cholestasis syndrome caused by VIPAR mutation. *Journal of Pediatric Gastroenterology and Nutrition*, **2014**, 58, e29-32	2.8	8
28	mTOR inhibitors in pediatric liver transplant recipients. *Clinics and Research in Hepatology and Gastroenterology*, **2019**, 43, 403-409	2.4	8
27	Long term results of liver transplantation for alpha-1 antitrypsin deficiency. *Digestive and Liver Disease*, **2021**, 53, 606-611	3.3	7
26	Combined Lung and Liver Transplantation for Short Telomere Syndrome. *Liver Transplantation*, **2020**, 26, 840-844	4.5	6
25	Impact of Genotype, Serum Bile Acids, and Surgical Biliary Diversion on Native Liver Survival in FIC1 Deficiency. *Hepatology*, **2021**, 74, 892-906	11.2	6
24	Clinical utility gene card for: progressive familial intrahepatic cholestasis type 3. *European Journal of Human Genetics*, **2014**, 22,	5.3	5
23	The efficacy of surgical shunts to treat severe portal hypertension after a Kasai procedure for biliary atresia. *Journal of Pediatric Surgery*, **2019**, 54, 531-536	2.6	4
22	Clinical utility gene card for: progressive familial intrahepatic cholestasis type 1. *European Journal of Human Genetics*, **2014**, 22,	5.3	4
21	Integrated Genomic Analysis Identifies Driver Genes and Cisplatin-Resistant Progenitor Phenotype in Pediatric Liver Cancer. *Cancer Discovery*, **2021**, 11, 2524-2543	24.4	4
20	Molecular Regulation of Canalicular ABC Transporters. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	4
19	Alagille Syndrome and Chronic Arthritis: An International Case Series. *Journal of Pediatrics*, **2020**, 218, 228-230.e1	3.6	3
18	Case report: progressive familial intrahepatic cholestasis type 3 with compound heterozygous ABCB4 variants diagnosed 15 years after liver transplantation. *BMC Medical Genetics*, **2020**, 21, 238	2.1	3

17	Adenosine kinase deficiency: Three new cases and diagnostic value of hypermethioninemia. *Molecular Genetics and Metabolism*, **2021**, 132, 38-43	3.7	3
16	Cholic Acid to Treat HSD3B7 and AKR1D1 Deficiencies. *Journal of Pediatric Gastroenterology and Nutrition*, **2017**, 65, e134	2.8	2
15	Clinical utility gene card for: progressive familial intrahepatic cholestasis type 2. *European Journal of Human Genetics*, **2014**, 22,	5.3	2
14	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	4.2	2
13	Effect of CFTR correctors on the traffic and the function of intracellularly retained ABCB4 variants. *Liver International*, **2021**, 41, 1344-1357	7.9	2
12	Diagnostic protocol of neonatal and infantile cholestasis: can it be improved?. *Journal of Pediatrics*, **2020**, 216, 247	3.6	2
11	pCLIF-SOFA is a reliable outcome prognostication score of critically ill children with cirrhosis: an ESPNIC multicentre study. *Annals of Intensive Care*, **2020**, 10, 137	8.9	1
10	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*, **2021**,	5.5	1
9	Pediatric Wilson's Disease: Phenotypic, Genetic Characterization and Outcome of 182 Children in France. *Journal of Pediatric Gastroenterology and Nutrition*, **2021**, 73, e80-e86	2.8	1
8	Long-term Follow-up of a Randomized Trial of Tacrolimus or Cyclosporine A Microemulsion in Children Post Liver Transplantation. *Transplantation Direct*, **2021**, 7, e765	2.3	1
7	Long-term results of pediatric liver transplantation for autoimmune liver disease. *Clinics and Research in Hepatology and Gastroenterology*, **2021**, 45, 101537	2.4	0
6	Targeted-Capture Next-Generation Sequencing in Diagnosis Approach of Pediatric Cholestasis. *Diagnostics*, **2022**, 12, 1169	3.8	0
5	Glycerol Phenylbutyrate Therapy in Progressive Familial Intrahepatic Cholestasis Type 2. *Journal of Pediatric Gastroenterology and Nutrition*, **2020**, 70, e139-e140	2.8	
4	Immunosuppressive therapy for indeterminate acute hepatitis or pediatric acute liver failure. *Journal of Pediatrics*, **2019**, 214, 243-244	3.6	
3	Primary bile acid therapy during pregnancy in patients with 3□hydroxy-□ -C27 -steroid oxidoreductase deficiency. *Pediatrics International*, **2011**, 53, 792	1.2	
2	Seronegative Autoimmune Hepatitis-associated Severe Aplastic Anemia: Looking for the Best Treatment. *Journal of Pediatric Gastroenterology and Nutrition*, **2021**, 73, e48	2.8	
1	Ursodeoxycholic acid therapy throughout pregnancy in women affected with chronic cholestasis of childhood: No evidence for teratogenicity. *Clinics and Research in Hepatology and Gastroenterology*, **2021**, 45, 101472	2.4	