

# Emmanuel Jacquemin

## List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

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	Targeted-Capture Next-Generation Sequencing in Diagnosis Approach of Pediatric Cholestasis. <i>Diagnostics</i> , <b>2022</b> , 12, 1169	3.8	0
69	In vitro functional rescue by ivacaftor of an ABCB11 variant involved in PFIC2 and intrahepatic cholestasis of pregnancy. <i>Orphanet Journal of Rare Diseases</i> , <b>2021</b> , 16, 484	4.2	2
68	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. <i>Clinical Chemistry</i> , <b>2021</b> ,	5.5	1
67	Efficacy and safety of maralixibat treatment in patients with Alagille syndrome and cholestatic pruritus (ICONIC): a randomised phase 2 study. <i>Lancet, The</i> , <b>2021</b> , 398, 1581-1592	40	17
66	Effect of CFTR correctors on the traffic and the function of intracellularly retained ABCB4 variants. <i>Liver International</i> , <b>2021</b> , 41, 1344-1357	7.9	2
65	Integrated Genomic Analysis Identifies Driver Genes and Cisplatin-Resistant Progenitor Phenotype in Pediatric Liver Cancer. <i>Cancer Discovery</i> , <b>2021</b> , 11, 2524-2543	24.4	4
64	Seronegative Autoimmune Hepatitis-associated Severe Aplastic Anemia: Looking for the Best Treatment. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , <b>2021</b> , 73, e48	2.8	
63	Pediatric Wilson's Disease: Phenotypic, Genetic Characterization and Outcome of 182 Children in France. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , <b>2021</b> , 73, e80-e86	2.8	1
62	Ursodeoxycholic acid therapy throughout pregnancy in women affected with chronic cholestasis of childhood: No evidence for teratogenicity. <i>Clinics and Research in Hepatology and Gastroenterology</i> , <b>2021</b> , 45, 101472	2.4	
61	Long term results of liver transplantation for alpha-1 antitrypsin deficiency. <i>Digestive and Liver Disease</i> , <b>2021</b> , 53, 606-611	3.3	7
60	Adenosine kinase deficiency: Three new cases and diagnostic value of hypermethioninemia. <i>Molecular Genetics and Metabolism</i> , <b>2021</b> , 132, 38-43	3.7	3
59	Long-term results of pediatric liver transplantation for autoimmune liver disease. <i>Clinics and Research in Hepatology and Gastroenterology</i> , <b>2021</b> , 45, 101537	2.4	0
58	Molecular Regulation of Canalicular ABC Transporters. <i>International Journal of Molecular Sciences</i> , <b>2021</b> , 22,	6.3	4
57	Impact of Genotype, Serum Bile Acids, and Surgical Biliary Diversion on Native Liver Survival in FIC1 Deficiency. <i>Hepatology</i> , <b>2021</b> , 74, 892-906	11.2	6
56	Long-term Follow-up of a Randomized Trial of Tacrolimus or Cyclosporine A Microemulsion in Children Post Liver Transplantation. <i>Transplantation Direct</i> , <b>2021</b> , 7, e765	2.3	1
55	Glycerol Phenylbutyrate Therapy in Progressive Familial Intrahepatic Cholestasis Type 2. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , <b>2020</b> , 70, e139-e140	2.8	
54	Functional rescue of an ABCB11 mutant by ivacaftor: A new targeted pharmacotherapy approach in bile salt export pump deficiency. <i>Liver International</i> , <b>2020</b> , 40, 1917-1925	7.9	17

53	Genotype correlates with the natural history of severe bile salt export pump deficiency. <i>Journal of Hepatology</i> , <b>2020</b> , 73, 84-93	13.4	22
52	Combined Lung and Liver Transplantation for Short Telomere Syndrome. <i>Liver Transplantation</i> , <b>2020</b> , 26, 840-844	4.5	6
51	pCLIF-SOFA is a reliable outcome prognostication score of critically ill children with cirrhosis: an ESPNIC multicentre study. <i>Annals of Intensive Care</i> , <b>2020</b> , 10, 137	8.9	1
50	Alagille Syndrome and Chronic Arthritis: An International Case Series. <i>Journal of Pediatrics</i> , <b>2020</b> , 218, 228-230.e1	3.6	3
49	Case report: progressive familial intrahepatic cholestasis type 3 with compound heterozygous ABCB4 variants diagnosed 15 years after liver transplantation. <i>BMC Medical Genetics</i> , <b>2020</b> , 21, 238	2.1	3
48	Diagnostic protocol of neonatal and infantile cholestasis: can it be improved?. <i>Journal of Pediatrics</i> , <b>2020</b> , 216, 247	3.6	2
47	Inherited IL-18BP deficiency in human fulminant viral hepatitis. <i>Journal of Experimental Medicine</i> , <b>2019</b> , 216, 1777-1790	16.6	42
46	The efficacy of surgical shunts to treat severe portal hypertension after a Kasai procedure for biliary atresia. <i>Journal of Pediatric Surgery</i> , <b>2019</b> , 54, 531-536	2.6	4
45	Immunosuppressive therapy for indeterminate acute hepatitis or pediatric acute liver failure. <i>Journal of Pediatrics</i> , <b>2019</b> , 214, 243-244	3.6	
44	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. <i>Transplantation</i> , <b>2019</b> , 103, 1903-1915	1.8	33
43	Management of Biliary Atresia in France 1986 to 2015: Long-term Results. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , <b>2019</b> , 69, 416-424	2.8	25
42	Liver disease related to alpha1-antitrypsin deficiency in French children: The DEFI-ALPHA cohort. <i>Liver International</i> , <b>2019</b> , 39, 1136-1146	7.9	19
41	mTOR inhibitors in pediatric liver transplant recipients. <i>Clinics and Research in Hepatology and Gastroenterology</i> , <b>2019</b> , 43, 403-409	2.4	8
40	Longterm Risk of Solid Organ De Novo Malignancies After Liver Transplantation: A French National Study on 11,226 Patients. <i>Liver Transplantation</i> , <b>2018</b> , 24, 1425-1436	4.5	31
39	Mutations in the novel gene FOPV are associated with familial autosomal dominant and non-familial obliterative portal venopathy. <i>Liver International</i> , <b>2018</b> , 38, 358-364	7.9	14
38	Cholic acid for primary bile acid synthesis defects: a life-saving therapy allowing a favorable outcome in adulthood. <i>Orphanet Journal of Rare Diseases</i> , <b>2018</b> , 13, 190	4.2	21
37	Attempt to Determine the Prevalence of Two Inborn Errors of Primary Bile Acid Synthesis: Results of a European Survey. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , <b>2017</b> , 64, 864-868	2.8	14
36	Bile Acid Synthesis Disorders in Arabs: A 10-year Screening Study. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , <b>2017</b> , 65, 613-620	2.8	11

35	Cholic Acid to Treat HSD3B7 and AKR1D1 Deficiencies. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , <b>2017</b> , 65, e134	2.8	2
34	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). <i>Hepatology</i> , <b>2017</b> , 65, 560-570	11.2	29
33	MYO5B mutations cause cholestasis with normal serum gamma-glutamyl transferase activity in children without microvillous inclusion disease. <i>Hepatology</i> , <b>2017</b> , 65, 164-173	11.2	84
32	Cholestasis Reveals Severe Cortisol Deficiency in Neonatal Pituitary Stalk Interruption Syndrome. <i>PLoS ONE</i> , <b>2016</b> , 11, e0147750	3.7	10
31	DCDC2 Mutations Cause Neonatal Sclerosing Cholangitis. <i>Human Mutation</i> , <b>2016</b> , 37, 1025-9	4.7	32
30	A functional classification of ABCB4 variations causing progressive familial intrahepatic cholestasis type 3. <i>Hepatology</i> , <b>2016</b> , 63, 1620-31	11.2	53
29	Rapid and reliable diagnosis of Wilson disease using X-ray fluorescence. <i>Journal of Pathology: Clinical Research</i> , <b>2016</b> , 2, 175-86	5.3	15
28	Targeted pharmacotherapy in progressive familial intrahepatic cholestasis type 2: Evidence for improvement of cholestasis with 4-phenylbutyrate. <i>Hepatology</i> , <b>2015</b> , 62, 558-66	11.2	75
27	Haploinsufficiency of the NOTCH1 Receptor as a Cause of Adams-Oliver Syndrome With Variable Cardiac Anomalies. <i>Circulation: Cardiovascular Genetics</i> , <b>2015</b> , 8, 572-581		65
26	Long term results of liver transplantation for Wilson's disease: experience in France. <i>Journal of Hepatology</i> , <b>2014</b> , 60, 579-89	13.4	78
25	Liver transcript analysis reveals aberrant splicing due to silent and intronic variations in the ABCB11 gene. <i>Molecular Genetics and Metabolism</i> , <b>2014</b> , 113, 225-9	3.7	10
24	Obliterative portal venopathy: a study of 48 children. <i>Journal of Pediatrics</i> , <b>2014</b> , 165, 190-193.e2	3.6	33
23	Clinical utility gene card for: progressive familial intrahepatic cholestasis type 1. <i>European Journal of Human Genetics</i> , <b>2014</b> , 22,	5.3	4
22	Clinical utility gene card for: progressive familial intrahepatic cholestasis type 2. <i>European Journal of Human Genetics</i> , <b>2014</b> , 22,	5.3	2
21	Clinical utility gene card for: progressive familial intrahepatic cholestasis type 3. <i>European Journal of Human Genetics</i> , <b>2014</b> , 22,	5.3	5
20	MYO5B and bile salt export pump contribute to cholestatic liver disorder in microvillous inclusion disease. <i>Hepatology</i> , <b>2014</b> , 60, 301-10	11.2	80
19	Arthrogyriposis, renal dysfunction, and cholestasis syndrome caused by VIPAR mutation. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , <b>2014</b> , 58, e29-32	2.8	8
18	Progressive familial intrahepatic cholestasis. <i>Clinics and Research in Hepatology and Gastroenterology</i> , <b>2012</b> , 36 Suppl 1, S26-35	2.4	152

17	Successful mutation-specific chaperone therapy with 4-phenylbutyrate in a child with progressive familial intrahepatic cholestasis type 2. <i>Journal of Hepatology</i> , <b>2012</b> , 57, 695-8	13.4	71
16	Primary bile acid therapy during pregnancy in patients with 3β-hydroxy-Δ <sup>5</sup> -C27-steroid oxidoreductase deficiency. <i>Pediatrics International</i> , <b>2011</b> , 53, 792	1.2	
15	The spectrum of liver diseases related to ABCB4 gene mutations: pathophysiology and clinical aspects. <i>Seminars in Liver Disease</i> , <b>2010</b> , 30, 134-46	7.3	175
14	Differences in presentation and progression between severe FIC1 and BSEP deficiencies. <i>Journal of Hepatology</i> , <b>2010</b> , 53, 170-8	13.4	145
13	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. <i>Hepatology</i> , <b>2010</b> , 51, 1645-55	11.2	188
12	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. <i>Hepatology</i> , <b>2009</b> , 49, 1218-27	11.2	47
11	Oral cholic acid for hereditary defects of primary bile acid synthesis: a safe and effective long-term therapy. <i>Gastroenterology</i> , <b>2009</b> , 137, 1310-1320.e1-3	13.3	74
10	Progressive familial intrahepatic cholestasis. <i>Orphanet Journal of Rare Diseases</i> , <b>2009</b> , 4, 1	4.2	229
9	Albumin liver dialysis as pregnancy-saving procedure in cholestatic liver disease and intractable pruritus. <i>World Journal of Gastroenterology</i> , <b>2008</b> , 14, 6572-4	5.6	12
8	Altered hepatobiliary gene expressions in PFIC1: ATP8B1 gene defect is associated with CFTR downregulation. <i>Hepatology</i> , <b>2006</b> , 43, 1125-34	11.2	58
7	Adams-Oliver syndrome and hepatoportal sclerosis: occasional association or common mechanism?. <i>American Journal of Medical Genetics, Part A</i> , <b>2005</b> , 135, 186-9	2.5	41
6	SRD5B1 (AKR1D1) gene analysis in delta(4)-3-oxosteroid 5beta-reductase deficiency: evidence for primary genetic defect. <i>Journal of Hepatology</i> , <b>2004</b> , 40, 716-8	13.4	51
5	Molecular genetics of 3beta-hydroxy-Delta <sup>5</sup> -C27-steroid oxidoreductase deficiency in 16 patients with loss of bile acid synthesis and liver disease. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2003</b> , 88, 1833-41	5.6	83
4	The wide spectrum of multidrug resistance 3 deficiency: from neonatal cholestasis to cirrhosis of adulthood. <i>Gastroenterology</i> , <b>2001</b> , 120, 1448-58	13.3	394
3	Progressive familial intrahepatic cholestasis. Genetic basis and treatment. <i>Clinics in Liver Disease</i> , <b>2000</b> , 4, 753-63	4.6	67
2	Progressive familial intrahepatic cholestasis. <i>Journal of Gastroenterology and Hepatology (Australia)</i> , <b>1999</b> , 14, 594-9	4	36
1	Heterozygous non-sense mutation of the MDR3 gene in familial intrahepatic cholestasis of pregnancy. <i>Lancet, The</i> , <b>1999</b> , 353, 210-1	4.0	297