A S Knisely

List of Publications by Year in descending order

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#	Article	IF	CITATIONS
1	Hepatocellular carcinoma in ten children under five years of age with bile salt export pump deficiency. Hepatology, 2006, 44, 478-486.	7.3	345
2	Complex inheritance of familial hypercholanemia with associated mutations in TJP2 and BAAT. Nature Genetics, 2003, 34, 91-96.	21.4	302
3	Mutations in TJP2 cause progressive cholestatic liver disease. Nature Genetics, 2014, 46, 326-328.	21.4	244
4	Mutations in the nuclear bile acid receptor FXR cause progressive familial intrahepatic cholestasis. Nature Communications, 2016, 7, 10713.	12.8	227
5	Defects in myosin VB are associated with a spectrum of previously undiagnosed low γâ€glutamyltransferase cholestasis. Hepatology, 2017, 65, 1655-1669.	7.3	107
6	Neonatal hemochromatosis. Gastroenterology Clinics of North America, 2003, 32, 877-889.	2.2	101
7	An expanded role for heterozygous mutations of ABCB4, ABCB11, ATP8B1, ABCC2 and TJP2 in intrahepatic cholestasis of pregnancy. Scientific Reports, 2017, 7, 11823.	3.3	98
8	Biallelic IARS Mutations Cause Growth Retardation with Prenatal Onset, Intellectual Disability, Muscular Hypotonia, and Infantile Hepatopathy. American Journal of Human Genetics, 2016, 99, 414-422.	6.2	73
9	Mutations in DCDC2 (doublecortin domain containing protein 2) in neonatal sclerosing cholangitis. Journal of Hepatology, 2016, 65, 1179-1187.	3.7	65
10	Hepatocellular carcinoma associated with tightâ€junction protein 2 deficiency. Hepatology, 2015, 62, 1914-1916.	7.3	63
11	Morphologic Findings in Progressive Familial Intrahepatic Cholestasis 2 (PFIC2). American Journal of Surgical Pathology, 2011, 35, 687-696.	3.7	58
12	Massive gene amplification drives paediatric hepatocellular carcinoma caused by bile salt export pump deficiency. Nature Communications, 2014, 5, 3850.	12.8	49
13	Bile salt export pumpâ€reactive antibodies form a polyclonal, multiâ€inhibitory response in antibodyâ€induced bile salt export pump deficiency. Hepatology, 2016, 63, 524-537.	7.3	45
14	Lowâ€GGT intrahepatic cholestasis associated with biallelic <i>USP53</i> variants: Clinical, histological and ultrastructural characterization. Liver International, 2020, 40, 1142-1150.	3.9	34
15	The lipid flippase heterodimer ATP8B1–CDC50A is essential for surface expression of the apical sodium-dependent bile acid transporter (SLC10A2/ASBT) in intestinal Caco-2 cells. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2014, 1842, 2378-2386.	3.8	33
16	<i>TJP2</i> hepatobiliary disorders: Novel variants and clinical diversity. Human Mutation, 2020, 41, 502-511.	2.5	25
17	Biliary tract malformations. American Journal of Medical Genetics Part A, 2003, 122A, 343-350.	2.4	20
18	Progressive Familial Intrahepatic Cholestasis: An Update. Pediatric and Developmental Pathology, 2004, 7, 309-314.	1.0	17

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19	Trafficking and Transporter Disorders in Pediatric Cholestasis. Clinics in Liver Disease, 2010, 14, 619-633.	2.1	17
20	ATP8B1-mediated spatial organization of Cdc42 signaling maintains singularity during enterocyte polarization. Journal of Cell Biology, 2015, 210, 1055-1063.	5.2	17
21	Polymorphisms in ABCB11 and ATP8B1 Associated with Development of Severe Intrahepatic Cholestasis in Hodgkin's Lymphoma. Journal of Clinical and Experimental Hepatology, 2013, 3, 159-161.	0.9	14
22	Severe Deoxyguanosine Kinase Deficiency in Austria. Journal of Pediatric Gastroenterology and Nutrition, 2019, 68, e1-e6.	1.8	13
23	An inborn error of bile salt transport with features mimicking abusive head trauma. Child Abuse and Neglect, 2010, 34, 472-476.	2.6	10
24	Beyond an Obvious Cause of Cholestasis in a Toddler: Compound Heterozygosity for <i>ABCB11</i> Mutations. Pediatrics, 2019, 143, .	2.1	8
25	Pediatric Wilson disease presenting as acute liver failure: Prognostic indices. World Journal of Clinical Cases, 2021, 9, 3273-3286.	0.8	7
26	Fibrinogen storage disease in a Chinese boy with de novo fibrinogen Aguadilla mutation: Incomplete response to carbamazepine and ursodeoxycholic acid. BMC Gastroenterology, 2016, 16, 92.	2.0	6
27	Mutation detection in cholestatic patients using microarray resequencing of ATP8B1 and ABCB11. F1000Research, 2013, 2, 32.	1.6	6
28	Patent ductus venosus and acute liver failure in the neonate: Consider neonatal hemochromatosis with liver scarring. Liver Transplantation, 2014, 20, 124-124.	2.4	5
29	Intrahepatic cholestasis in two omani siblings associated with a novel homozygous ATP8B1 mutation, c.379C>G (p.L127V). Saudi Journal of Gastroenterology, 2017, 23, 303.	1.1	2
30	Vasotropic light-chain amyloidosis and ischaemic cholangiopathy. BMJ Case Reports, 2015, 2015, bcr2015210883.	0.5	1
31	Bile salt export pump expression: Can immunohistochemistry in isolation mislead?. Hepatology, 2014, 59, 2056-2056.	7.3	0