A S Knisely

List of Publications by Year
in descending order

31 papers	2,013 citations	535685 17 h-index	511568 30 g-index
32 all docs	32 docs citations	32 times ranked	2515 citing authors

#	Article	IF	Citations
1	Pediatric Wilson disease presenting as acute liver failure: Prognostic indices. World Journal of Clinical Cases, 2021, 9, 3273-3286.	0.3	7
2	TJP2 hepatobiliary disorders: Novel variants and clinical diversity. Human Mutation, 2020, 41, 502-511.	1.1	25
3	Lowâ€GGT intrahepatic cholestasis associated with biallelic USP53 variants: Clinical, histological and ultrastructural characterization. Liver International, 2020, 40, 1142-1150.	1.9	34
4	Severe Deoxyguanosine Kinase Deficiency in Austria. Journal of Pediatric Gastroenterology and Nutrition, 2019, 68, e1-e6.	0.9	13
5	Beyond an Obvious Cause of Cholestasis in a Toddler: Compound Heterozygosity for ABCB11 Mutations. Pediatrics, 2019, 143, .	1.0	8
6	Defects in myosin VB are associated with a spectrum of previously undiagnosed low Î³â€glutamyltransferase cholestasis. Hepatology, 2017, 65, 1655-1669.	3.6	107
7	An expanded role for heterozygous mutations of ABCB4, ABCB11, ATP8B1, ABCC2 and TJP2 in intrahepatic cholestasis of pregnancy. Scientific Reports, 2017, 7, 11823.	1.6	98
8	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.	0.5	2
9	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.	3.6	45
10	Mutations in DCDC2 (doublecortin domain containing protein 2) in neonatal sclerosing cholangitis. Journal of Hepatology, 2016, 65, 1179-1187.	1.8	65
11	Mutations in the nuclear bile acid receptor FXR cause progressive familial intrahepatic cholestasis. Nature Communications, 2016, 7, 10713.	5.8	227
12	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.	2.6	73
13	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.	0.8	6
14	Hepatocellular carcinoma associated with tightâ€junction protein 2 deficiency. Hepatology, 2015, 62, 1914-1916.	3.6	63
15	ATP8B1-mediated spatial organization of Cdc42 signaling maintains singularity during enterocyte polarization. Journal of Cell Biology, 2015, 210, 1055-1063.	2.3	17
16	Vasotropic light-chain amyloidosis and ischaemic cholangiopathy. BMJ Case Reports, 2015, 2015, bcr2015210883.	0.2	1
17	Massive gene amplification drives paediatric hepatocellular carcinoma caused by bile salt export pump deficiency. Nature Communications, 2014, 5, 3850.	5.8	49
18	The lipid flippase heterodimer ATP8B1â€“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.	1.8	33

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19	Bile salt export pump expression: Can immunohistochemistry in isolation mislead?. Hepatology, 2014, 59, 2056-2056.	3.6	0
20	Mutations in TJP2 cause progressive cholestatic liver disease. Nature Genetics, 2014, 46, 326-328.	9.4	244
21	Patent ductus venosus and acute liver failure in the neonate: Consider neonatal hemochromatosis with liver scarring. Liver Transplantation, 2014, 20, 124-124.	1.3	5
22	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.	0.4	14
23	Mutation detection in cholestatic patients using microarray resequencing of ATP8B1 and ABCB11. F1000Research, 2013, 2, 32.	0.8	6
24	Morphologic Findings in Progressive Familial Intrahepatic Cholestasis 2 (PFIC2). American Journal of Surgical Pathology, 2011, 35, 687-696.	2.1	58
25	An inborn error of bile salt transport with features mimicking abusive head trauma. Child Abuse and Neglect, 2010, 34, 472-476.	1.3	10
26	Trafficking and Transporter Disorders in Pediatric Cholestasis. Clinics in Liver Disease, 2010, 14, 619-633.	1.0	17
27	Hepatocellular carcinoma in ten children under five years of age with bile salt export pump deficiency. Hepatology, 2006, 44, 478-486.	3.6	345
28	Progressive Familial Intrahepatic Cholestasis: An Update. Pediatric and Developmental Pathology, 2004, 7, 309-314.	0.5	17
29	Biliary tract malformations. American Journal of Medical Genetics Part A, 2003, 122A, 343-350.	2.4	20
30	Complex inheritance of familial hypercholanemia with associated mutations in TJP2 and BAAT. Nature Genetics, 2003, 34, 91-96.	9.4	302
31	Neonatal hemochromatosis. Gastroenterology Clinics of North America, 2003, 32, 877-889.	1.0	101