Spraul Anne

List of Publications by Year in Descending Order

20 papers	802 citations	10 h-index	20 g-index
20 ext. papers	930 ext. citations	4.4 avg, IF	3.68 L-index

#	Paper	IF	Citations
20	Adenosine kinase deficiency: Three new cases and diagnostic value of hypermethioninemia. *Molecular Genetics and Metabolism*, **2021**, 132, 38-43	3.7	3
19	Deep phenotyping of MARS1 (interstitial lung and liver disease) and LARS1 (infantile liver failure syndrome 1) recessive multisystemic disease using Human Phenotype Ontology annotation: Overlap and differences. Case report and review of literature. *European Journal of Medical Genetics*, **2021**, 64, 104334	2.6	1
18	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
17	A Novel CFC1 Mutation in a Family With Heterotaxy and Biliary Atresia Splenic Malformation Syndromes. *Journal of Pediatric Gastroenterology and Nutrition*, **2020**, 70, e24-e25	2.8	1
16	Genotype correlates with the natural history of severe bile salt export pump deficiency. *Journal of Hepatology*, **2020**, 73, 84-93	13.4	22
15	Bile Acid Synthesis Disorders in Arabs: A 10-year Screening Study. *Journal of Pediatric Gastroenterology and Nutrition*, **2017**, 65, 613-620	2.8	11
14	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
13	Cholestasis Reveals Severe Cortisol Deficiency in Neonatal Pituitary Stalk Interruption Syndrome. *PLoS ONE*, **2016**, 11, e0147750	3.7	10
12	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
11	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
10	Clinical utility gene card for: progressive familial intrahepatic cholestasis type 1. *European Journal of Human Genetics*, **2014**, 22,	5.3	4
9	Clinical utility gene card for: progressive familial intrahepatic cholestasis type 2. *European Journal of Human Genetics*, **2014**, 22,	5.3	2
8	Clinical utility gene card for: progressive familial intrahepatic cholestasis type 3. *European Journal of Human Genetics*, **2014**, 22,	5.3	5
7	Secondary Mitochondrial Respiratory Chain Defect Can Delay Accurate PFIC2 Diagnosis. *JIMD Reports*, **2014**, 14, 17-21	1.9	7
6	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
5	NR1H4 analysis in patients with progressive familial intrahepatic cholestasis, drug-induced cholestasis or intrahepatic cholestasis of pregnancy unrelated to ATP8B1, ABCB11 and ABCB4 mutations. *Clinics and Research in Hepatology and Gastroenterology*, **2012**, 36, 569-73	2.4	17
4	Simple and fast quantification of nitisone (NTBC) using liquid chromatography-tandem mass spectrometry method in plasma of tyrosinemia type 1 patients. *Journal of Chromatographic Science*, **2012**, 50, 446-9	1.4	10

3	Liver glycogen storage diseases due to phosphorylase system deficiencies: diagnosis thanks to non invasive blood enzymatic and molecular studies. *Molecular Genetics and Metabolism*, **2011**, 104, 137-43	3.7	35
2	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
1	Progressive familial intrahepatic cholestasis. *Orphanet Journal of Rare Diseases*, **2009**, 4, 1	4.2	229