# Richard J Thompson

List of Publications by Year
in descending order



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|---|---|---|---|
| 88 papers | 6,350 citations | 109137 35 h-index | 66788 78 g-index |
| 93 all docs | 93 docs citations | 93 times ranked | 3915 citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | A gene encoding a liver-specific ABC transporter is mutated in progressive familial intrahepatic cholestasis. Nature Genetics, 1998, 20, 233-238. | 9.4 | 968 |
| 2 | Hepatocanalicular bile salt export pump deficiency in patients with progressive familial intrahepatic cholestasis. Gastroenterology, 1999, 117, 1370-1379. | 0.6 | 423 |
| 3 | Hepatocellular carcinoma in ten children under five years of age with bile salt export pump deficiency. Hepatology, 2006, 44, 478-486. | 3.6 | 345 |
| 4 | Severe Bile Salt Export Pump Deficiency: 82 Different ABCB11 Mutations in 109 Families. Gastroenterology, 2008, 134, 1203-1214.e8. | 0.6 | 331 |
| 5 | Mutations in VPS33B, encoding a regulator of SNARE-dependent membrane fusion, cause arthrogryposisâ€“renal dysfunctionâ€“cholestasis (ARC) syndrome. Nature Genetics, 2004, 36, 400-404. | 9.4 | 313 |
| 6 | The human bile salt export pump: Characterization of substrate specificity and identification of inhibitors. Gastroenterology, 2002, 123, 1649-1658. | 0.6 | 308 |
| 7 | Characterization of mutations inATP8B1associated with hereditary cholestasis. Hepatology, 2004, 40, 27-38. | 3.6 | 263 |
| 8 | Mutations in TJP2 cause progressive cholestatic liver disease. Nature Genetics, 2014, 46, 326-328. | 9.4 | 244 |
| 9 | A novel spliceâ€“site mutation in the Î³ subunit of the epithelial sodium channel gene in three pseudohypoaldosteronism type 1 families. Nature Genetics, 1996, 13, 248-250. | 9.4 | 228 |
| 10 | Identification of a Locus for Progressive Familial Intrahepatic Cholestasis PFIC2on Chromosome 2q24. American Journal of Human Genetics, 1997, 61, 630-633. | 2.6 | 190 |
| 11 | Progressive familial intrahepatic cholestasis, type 1, is associated with decreased farnesoid X receptor activity. Gastroenterology, 2004, 126, 756-764. | 0.6 | 185 |
| 12 | Differences in presentation and progression between severe FIC1 and BSEP deficiencies. Journal of Hepatology, 2010, 53, 170-178. | 1.8 | 182 |
| 13 | Contribution of variant alleles of ABCB11 to susceptibility to intrahepatic cholestasis of pregnancy. Gut, 2009, 58, 537-544. | 6.1 | 179 |
| 14 | Missense mutations and single nucleotide polymorphisms in ABCB11 impair bile salt export pump processing and function or disrupt pre-messenger RNA splicing. Hepatology, 2009, 49, 553-567. | 3.6 | 147 |
| 15 | Mutations in Bile Salt Export Pump (ABCB11) in Two Children with Progressive Familial Intrahepatic Cholestasis and Cholangiocarcinoma. Journal of Pediatrics, 2007, 150, 556-559. | 0.9 | 129 |
| 16 | Progressive Familial Intrahepatic Cholestasis. Clinics in Liver Disease, 2018, 22, 657-669. | 1.0 | 124 |
| 17 | BSEP: Function and Role in Progressive Familial Intrahepatic Cholestasis. Seminars in Liver Disease, 2001, 21, 545-550. | 1.8 | 117 |
| 18 | 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 |

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|---|---|---|---|
| 19 | Sequencing of FIC1, BSEP and MDR3 in a large cohort of patients with cholestasis revealed a high number of different genetic variants. Journal of Hepatology, 2017, 67, 1253-1264. | 1.8 | 97 |
| 20 | Localisation of Pseudohypoaldosteronism Genes to Chromosome 16p12.2â€“13.11 and 12p13.1-Pter by Homozygosity Mapping. Human Molecular Genetics, 1996, 5, 293-299. | 1.4 | 73 |
| 21 | Genetics of liver disease: From pathophysiology to clinical practice. Journal of Hepatology, 2015, 62, S6-S14. | 1.8 | 73 |
| 22 | Mutations in DCDC2 (doublecortin domain containing protein 2) in neonatal sclerosing cholangitis. Journal of Hepatology, 2016, 65, 1179-1187. | 1.8 | 65 |
| 23 | Hepatocellular carcinoma associated with tightâ€junction protein 2 deficiency. Hepatology, 2015, 62, 1914-1916. | 3.6 | 63 |
| 24 | Genotype correlates with the natural history of severe bile salt export pump deficiency. Journal of Hepatology, 2020, 73, 84-93. | 1.8 | 61 |
| 25 | Recurrent low gamma-glutamyl Transpeptidase cholestasis following liver transplantation for bile salt export pump (BSEP) disease (posttransplant recurrent BSEP disease). Liver Transplantation, 2010, 16, 856-863. | 1.3 | 60 |
| 26 | Identification of Polycystic Kidney Disease 1 Like 1 Gene Variants in Children With Biliary Atresia Splenic Malformation Syndrome. Hepatology, 2019, 70, 899-910. | 3.6 | 58 |
| 27 | Mutations inTJP2, encoding zona occludens 2, and liver disease. Tissue Barriers, 2015, 3, e1026537. | 1.6 | 55 |
| 28 | Outcomes of surgical management of familial intrahepatic cholestasis 1 and bile salt export protein deficiencies. Hepatology Communications, 2018, 2, 515-528. | 2.0 | 54 |
| 29 | Odevixibat treatment in progressive familial intrahepatic cholestasis: a randomised, placebo-controlled, phase 3 trial. The Lancet Gastroenterology and Hepatology, 2022, 7, 830-842. | 3.7 | 54 |
| 30 | VPS33B mutation with ichthyosis, cholestasis, and renal dysfunction but without arthrogryposis: Incomplete ARC syndrome phenotype. Journal of Pediatrics, 2006, 148, 269-271. | 0.9 | 50 |
| 31 | Massive gene amplification drives paediatric hepatocellular carcinoma caused by bile salt export pump deficiency. Nature Communications, 2014, 5, 3850. | 5.8 | 49 |
| 32 | Novel Mutations Responsible for Autosomal Recessive Multisystem Pseudohypoaldosteronism and Sequence Variants in Epithelial Sodium Channel Î±-, Î²-, and Î³-Subunit Genes. Journal of Clinical Endocrinology and Metabolism, 2002, 87, 3344-3350. | 1.8 | 48 |
| 33 | Genetic Defects in Hepatocanalicular Transport. Seminars in Liver Disease, 2000, Volume 20, 365-372. | 1.8 | 43 |
| 34 | Effects of odevixibat on pruritus and bile acids in children with cholestatic liver disease: Phase 2 study. Clinics and Research in Hepatology and Gastroenterology, 2021, 45, 101751. | 0.7 | 40 |
| 35 | Congenital bile duct anomalies (biliary atresia) and chromosome 22 aneuploidy. Journal of Pediatric Surgery, 2008, 43, 1736-1740. | 0.8 | 38 |
| 36 | Enriched conditioning expands the regenerative ability of sensory neurons after spinal cord injury via neuronal intrinsic redox signaling. Nature Communications, 2020, 11, 6425. | 5.8 | 37 |

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|---|---|---|---|
| 37 | Effect of ischemic preconditioning on the genomic response to reperfusion injury in deceased donor liver transplantation. Liver Transplantation, 2009, 15, 1750-1765. | 1.3 | 36 |
| 38 | Heterogeneity of the egammadeltabeta-thalassaemias: characterization of three novel English deletions. British Journal of Haematology, 2005, 128, 722-729. | 1.2 | 35 |
| 39 | Multiple Î²-catenin mutations in hepatocellular lesions arising in Abernethy malformation. Human Pathology, 2016, 53, 153-158. | 1.1 | 32 |
| 40 | Lack of hepatocellular CD10 along bile canaliculi is physiologic in early childhood and persistent in Alagille syndrome. Laboratory Investigation, 2007, 87, 1138-1148. | 1.7 | 31 |
| 41 | Gene Structure of the Human Amiloride-Sensitive Epithelial Sodium Channel Beta Subunit. Biochemical and Biophysical Research Communications, 1998, 252, 208-213. | 1.0 | 29 |
| 42 | Maralixibat for the treatment of PFIC: Longâ€term, IBAT inhibition in an openâ€label, Phase 2 study. Hepatology Communications, 2022, 6, 2379-2390. | 2.0 | 26 |
| 43 | Bile composition in Alagille Syndrome and PFIC patients having Partial External Biliary Diversion. BMC Gastroenterology, 2008, 8, 47. | 0.8 | 25 |
| 44 | Impact of Genotype, Serum Bile Acids, and Surgical Biliary Diversion on Native Liver Survival in FIC1 Deficiency. Hepatology, 2021, 74, 892-906. | 3.6 | 25 |
| 45 | Review article: liver disease in adults with variants in the cholestasisâ€related genes ABCB11, ABCB4 and ATP8B1. Alimentary Pharmacology and Therapeutics, 2020, 52, 1628-1639. | 1.9 | 25 |
| 46 | Histopathology of livers in patients with congenital portosystemic shunts (Abernethy malformation): a case series of 22 patients. Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin, 2019, 474, 47-57. | 1.4 | 24 |
| 47 | Systematic Review and Metaâ€analysis. Journal of Pediatric Gastroenterology and Nutrition, 2020, 71, 176-183. | 0.9 | 21 |
| 48 | Exocrine Pancreatic Function in Children with Progressive Familial Intrahepatic Cholestasis Type 2. Journal of Pediatric Gastroenterology and Nutrition, 2006, 42, 416-418. | 0.9 | 20 |
| 49 | Use of a Comprehensive 66-Gene Cholestasis Sequencing Panel in 2171 Cholestatic Infants, Children, and Young Adults. Journal of Pediatric Gastroenterology and Nutrition, 2021, 72, 654-660. | 0.9 | 20 |
| 50 | Inherited disorders of transport in the liver. Current Opinion in Genetics and Development, 2000, 10, 310-313. | 1.5 | 19 |
| 51 | Cholestasis Due to USP53 Deficiency. Journal of Pediatric Gastroenterology and Nutrition, 2021, 72, 667-673. | 0.9 | 18 |
| 52 | Mutations in Myosin 5B in Children With Earlyâ€onset Cholestasis. Journal of Pediatric Gastroenterology and Nutrition, 2020, 71, 184-188. | 0.9 | 16 |
| 53 | Impact of longâ€term administration of maralixibat on children with cholestasis secondary to Alagille syndrome. Hepatology Communications, 2022, 6, 1922-1933. | 2.0 | 12 |
| 54 | Cholestatic and Metabolic Liver Diseases: Working Group Report of the Second World Congress of Pediatric Gastroenterology, Hepatology, and Nutrition. Journal of Pediatric Gastroenterology and Nutrition, 2004, 39, S611-S615. | 0.9 | 11 |

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|---|---|---|---|
| 55 | Reduced Hepatocellular Expression of Canalicular Transport Proteins in Infants with Neonatal Cholestasis and Congenital Hypopituitarism. Journal of Pediatrics, 2018, 200, 181-187. | 0.9 | 10 |
| 56 | Cholestasis: Current Issues and Plan for the Future. Journal of Pediatric Gastroenterology and Nutrition, 2008, 47, 220-224. | 0.9 | 9 |
| 57 | Longitudinal Outcomes in Young Patients with Alpha-1-Antitrypsin Deficiency with Native Liver Reveal that Neonatal Cholestasis is a Poor Predictor of Future Portal Hypertension. Journal of Pediatrics, 2020, 227, 81-86.e4. | 0.9 | 9 |
| 58 | Genetic and metabolic liver disease. , 2012, , 157-259. | | 8 |
| 59 | Biliary transporter gene mutations in severe intrahepatic cholestasis of pregnancy: Diagnostic and management implications. Journal of Gastroenterology and Hepatology (Australia), 2019, 34, 425-435. | 1.4 | 8 |
| 60 | Mutation Analysis and Disease Features at Presentation in a Multiâ€Center Cohort of Children With Monogenic Cholestasis. Journal of Pediatric Gastroenterology and Nutrition, 2021, 73, 169-177. | 0.9 | 8 |
| 61 | Liver Disease and Risk of Hepatocellular Carcinoma in Children With Mutations in TALDO1. Hepatology Communications, 2022, 6, 473-479. | 2.0 | 8 |
| 62 | Progressive familial intrahepatic cholestasis â€" farnesoid X receptor deficiency due to NR1H4 mutation: A case report. World Journal of Clinical Cases, 2021, 9, 3631-3636. | 0.3 | 7 |
| 63 | Study of Acute Liver Failure in Children Using Next Generation Sequencing Technology. Journal of Pediatrics, 2021, 236, 124-130. | 0.9 | 7 |
| 64 | Microvilli as markers of disordered apical-membrane trafficking and assembly: Bowel and liver. Hepatology, 2014, 60, 34-36. | 3.6 | 6 |
| 65 | Antiâ€CD20 Monoclonal Antibody Therapy in Functional Bile Salt Export Pump Deficiency After Liver Transplantation. Journal of Pediatric Gastroenterology and Nutrition, 2015, 60, e50-3. | 0.9 | 6 |
| 66 | Pigmented well-differentiated hepatocellular neoplasm with Î²-catenin mutation. Hepatobiliary and Pancreatic Diseases International, 2015, 14, 660-664. | 0.6 | 5 |
| 67 | Massive ascites and the heterozygous alpha 1 antitrypsin (Î±<sub>1</sub><scp>AT</scp>) living related donor liver in the homozygous child. Pediatric Transplantation, 2018, 22, e13075. | 0.5 | 5 |
| 68 | Portal venous deprivation in patients with portosystemic shunts and its effect on liver tumors. Journal of Pediatric Surgery, 2020, 55, 651-654. | 0.8 | 5 |
| 69 | Human iPSC-derived hepatocyte system models cholestasis with tight junction protein 2 deficiency. JHEP Reports, 2022, 4, 100446. | 2.6 | 5 |
| 70 | Complications of congenital portosystemic shunts: liver tumors are affected by shunt severity, but pulmonary and neurocognitive associations are not. Hepatology International, 2022, 16, 918-925. | 1.9 | 5 |
| 71 | Treating genetic disease: Expanding the options. Hepatology, 2015, 62, 349-351. | 3.6 | 4 |
| 72 | ATP8B1 Gene Expression Is Driven by a Housekeeping-Like Promoter Independent of Bile Acids and Farnesoid X Receptor. PLoS ONE, 2012, 7, e51650. | 1.1 | 4 |

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|---|---|---|---|
| 73 | Sequencing of transporter genes in cholestasis: We are still learning. Journal of Hepatology, 2017, 67, 1132-1133. | 1.8 | 3 |
| 74 | Donor transmitted mutation of the ABCB11 gene and ensuing intrahepatic cholestasis of pregnancy in a liver transplant recipient. Liver Transplantation, 2017, 23, 1229-1232. | 1.3 | 2 |
| 75 | Pediatric liver disease. Current Opinion in Gastroenterology, 1999, 15, 249-252. | 1.0 | 2 |
| 76 | Pediatric liver disease. Current Opinion in Gastroenterology, 2000, 16, 271-274. | 1.0 | 1 |
| 77 | Healy's law and the pediatric liver allograft. Liver Transplantation, 2008, 14, 1557-1558. | 1.3 | 1 |
| 78 | Challenges in understanding the consequences of variants in ABCB4 gene. Journal of Hepatology, 2021, 74, 242-243. | 1.8 | 1 |
| 79 | The Influence of Donor and Recipient Complement C3 Polymorphisms on Liver Transplant Outcome. International Journal of Hepatology, 2021, 2021, 1-14. | 0.4 | 1 |
| 80 | Defects in the enterohepatic circulation of bile acids. Journal of Hepatology, 2000, 32, 5-6. | 1.8 | 0 |
| 81 | 879 CHOLANGIOPATHY AND HCC IN MDR3 DEFICIENCY. Journal of Hepatology, 2008, 48, S330. | 1.8 | 0 |
| 82 | SÃndromes colestÃ¡ticos familiares. Annales NestlÃ© (Ed EspaÃ±ola), 2008, 66, 121-126. | 0.1 | 0 |
| 83 | Familial Cholestatic Syndromes. Annales Nestle, 2008, 66, 121-126. | 0.1 | 0 |
| 84 | Syndromes cholestatiques familiaux. Annales Nestle [Ed Francaise], 2008, 66, 121-126. | 0.0 | 0 |
| 85 | Familial Intrahepatic Cholestasis. , 2016, , 663-670. | | 0 |
| 86 | Reply to: â€œDoublecortin domain containing protein 2 (DCDC2) genetic variants in primary sclerosing cholangitisâ€• Journal of Hepatology, 2017, 67, 652-653. | 1.8 | 0 |
| 87 | Severe bile salt export pump (BSEP) deficiency: mutations, immunohistochemically assessed BSEP expression, and malignancy risk. , 2009, , 206-216. | | 0 |
| 88 | The role of ABCB11 (BSEP) variation in susceptibility to intrahepatic cholestasis of pregnancy. , 2009, , 46-50. | | 0 |