New perspectives in cystic fibrosis associated liver diseases in children

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Cystic fibrosis (CF) is a multisystem genetic disorder caused by a defect in the cystic fibrosis transmembrane conductance regulator (CFTR) gene, encoding the transporter protein responsible for chloride flux across the apical membrane of the epithelial cell. Significant liver disease is an unusual presentation in CF population with the incidence of 4-7%; however an emerging cause of mortality as a result of advanced care in pulmonary, nutritional and lung transplantation care. The cause of CFLD is unknown. The presentation of CFLD varies widely in the CF population with most liver disease occurring at or before puberty. Although dual-pass liver biopsy increases diagnostic yield with the presence of fibrosis, the distribution of fibrotic liver lesions in CFLD is focal and yet the procedure is rather invasive. Currently there are no known risk factors predicting the development of CFLD. The development of non-invasive fibrosis markers has progressed rapidly. Such markers include hyaluronic acid, metalloproteinases, tissue Inhibitors of matrix metalloproteinases (TIMPS), transforming growth factor (TGF-β1), etc. These are not routine laboratory investigations and tend to be expensive. In addition, they may be elevated secondary to fibrosis in extrahepatic organs such as the lungs. Many simple and inexpensive methods such as APR, FIB-4, liver ultrasound have been used as a non-invasive biochemical marker. Furthermore, in CF the role of these tests to reflect CFLD is unclear but may help for early detection of a child with CFLD. Novel therapies exist in CF children. It is still unknown if any therapy can prevent the development and progression of liver disease in CF children.

Speaker Biography

Wikrom Karnsakul is an Assistant Professor of Pediatrics at the Johns Hopkins University School of Medicine. His clinical interests are in the care of pediatric liver diseases, and general gastrointestinal diseases. He received his Medical degree in 1992 from Mahidol University School of Medicine in Bangkok, Thailand. He completed his Residency in Pediatrics at Advocate Hope Children's Hospital, University of Illinois at Chicago in 1998 and did a fellowship in Pediatric Gastroenterology, Hepatology and Nutrition at Texas Children's Hospital, Baylor College of Medicine in Houston, Texas. He completed his Post-doctoral research training at USDA/ARS Children's Nutrition Research Center, Baylor College of Medicine. He joined the faculty at Johns Hopkins University School of Medicine in 2008. His research interests center on the understanding and treatment of chronic liver disease, ascites, cholestasis, viral hepatitis, metabolic liver diseases and living related liver transplantation. He has a particular focus on hepatitis E infection. He is also involved in NIH-funded multicenter research studies including the Cholestatic Liver Disease Consortium and Cystic Fibrosis Related Liver Disease Project.

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