PFIC (Progressive Familial Intrahepatic Cholestasis) Type 1

PFIC-1 is characterized by a mutation in FIC-1, a gene predicted to encode a P-type ATPase that may be involved in phospholipid translocation. It was previously identified as a clinical entity known as Byler's disease. Patients with PFIC may have severe cholestasis manifest by intense pruritus, fat malabsorption and fat soluble vitamin deficiencies. PFIC-1 may be associated with extrahepatic manifestations, especially after liver transplantation, since the FIC-1 gene is expressed in many tissues other than the liver.

For Additional information:
Cholestatic Liver Disease Consortium (CLiC)
http://rarediseasesnetwork.epi.usf.edu/clic/learnmore/pfic.htm

http://PFIC.org A resource for parents

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