PFIC (Progressive Familial Intrahepatic Cholestasis) Type 2

PFIC-2 is caused by mutations in the gene that codes for the bile salt export pump, or BSEP. Hepatic excretion of bile acids is primarily controlled by BSEP, so severe cholestasis is common in patients with BSEP disease. As BSEP is expressed only in the liver, extrahepatic disease in PFIC-2 is secondary rather than primary.

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

http://PFIC.org A PFIC resource for parents

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