PFIC (Progressive Familial Intrahepatic Cholestasis) Type 3

PFIC-3 is caused by mutations in the MDR3 gene, a flippase that moves phospholipid to the outer leaflet of cell membranes permitting biliary exctretion. Without functional MDR3, bile is deficient in phosphatidylcholine and abnormally caustic. This damages hepatocytes and bile ducts. The abnormal bile also is prone to form gallstones.

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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