Title: ATP8B1 Deficiency *GeneReview* Histologic Findings
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Histologic Findings Associated with ATP8B1 Deficiency

Severe ATP8B1 deficiency. The characteristic histopathologic features at presentation include bland intracanalicular cholestasis in the setting of a relatively preserved lobular architecture with "tidy"-appearing and sometimes small hepatocytes. Bile ducts are sometimes small and inconspicuous and may appear hypoplastic; however, paucity of interlobular bile ducts is not seen.

There is no significant lobular disarray or marked giant cell transformation of hepatocytes (which is more typical of BSEP deficiency). There is no ductal or significant ductular reaction (which is associated with biliary obstruction including biliary atresia).

In infancy, fibrosis is usually limited to the portal tracts. There usually is progression of fibrosis with portal to portal bridging, pericellular and pericentral fibrosis that can be quite marked with the appearance of cirrhosis [Morotti & Saxena 2018].

Although the pathognomonic pathologic feature on transmission electron microscopy, is the classic coarsely granular "Byler" bile, treatment with ursodeoxycholic acid may alter this finding.

Mild ATP8B1 deficiency. Findings during an episode of cholestasis resemble those at presentation in severe ATP8B1 deficiency.

Literature Cited

Morotti RA, Saxena R. Intrahepatic cholestasis: molecular physiology of bile formation and secretion. In: Saxena, R, ed. *Practical Hepatic Pathology: A Diagnostic Approach*. 2 ed. Amsterdam: Elsevier, Inc; 2018:chap 29A.