History
Toddler with hyperbilirubinemia.

Diagnosis
Caroli Disease

Discussion
Caroli disease is an autosomal recessive disorder that is characterized by improper formation of the hepatic biliary system during embryogenesis. Under normal circumstances a complex process of cellular proliferation and apoptosis results in the generation of the biliary tree throughout the liver. This process begins at the hilum of the liver with formation of large bile ducts and progresses outwards towards the periphery of the liver, gradually forming the smaller hepatic bile ductules. However, in the case of Caroli’s disease this process is disrupted leading to inflammation and ultimately dilation of the large intrahepatic bile ducts. This failure to form the biliary system can lead to fibrosis and cirrhosis of the liver, portal hypertension, cholestasis, and cholangitis. In terms of symptomology, patients often present with right upper quadrant pain and fever. More severe symptoms can include jaundice and other signs associated with liver failure or sepsis. Patients with Caroli's disease may also present with cholangiocarcinoma.

Imaging modalities used to diagnose Caroli’s disease include: ultrasound, endoscopic retrograde cholangiopancreatography, and magnetic resonance cholangiography. The key diagnostic feature of Caroli’s disease involves abnormalities of the intrahepatic ducts. It can be further classified based on segmental to diffuse biliary dilation, fusiform to saccular dilation, and isolated intrahepatic involvement to intra and extrahepatic involvement of this system. Other findings on imaging include irregularities of the walls of the bile ducts, strictures, stones, and periductal fibrosis. Dilation of the extrahepatic duct system (common hepatic and common bile ducts) may also occur.

The differential diagnosis when addressing a patient who potentially has Caroli’s disease includes: Primary sclerosing cholangitis, recurrent pyogenic cholangitis, polycystic liver disease, choledochal cysts, biliary papillomatosis, and obstructive biliary dilation. Although many of these diseases have similar presentation in regards to dilation of the duct system, the main factor that differentiates Caroli’s is the presence of saccular dilation of the intrahepatic biliary ducts.

Findings
US-Saccular dilation of peripheral intrahepatic bile ducts, increased periportal and parenchymal echogenicity, and smooth contour

Reference

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