Liver Involvement in Autosomal Recessive Polycystic Kidney Disease

M. Alexandra Bramlage MD[1], Jessica Sang MD, MPH[2], Jason Lewis MD[2]
1.Atrium Health Department of Internal Medicine 2.Atrium Health Department of Gastroenterology and Hepatology

Introduction

- Autosomal recessive polycystic kidney disease (ARPKD) is an uncommon genetic disorder, which is seen in approximately 1 in 20,000 children.[1]
- Because this disease is so rare, the associated pathologies of ARPKD can often be overlooked.
- It is important to know that liver involvement is invariably present in all cases of ARPKD and the following case highlights this association.[2]

Objectives

- Appreciate that congenital hepatic fibrosis has a known association with ARPKD.
- Recognize that EUS is an innovative modality for measuring portal pressure gradient.

Case Report

Presentation

- A 36-year-old female with ARPKD status post renal transplant on mycophenolate and tacrolimus for immunosuppression presented to the hospital with melena and hematemesis.
- Her hemoglobin was 7.2 g/dL on admission; her baseline hemoglobin was 12.5 g/dL.

Workup

- EGD was performed and grade 2 esophageal varices were identified and banded.
- These findings prompted workup for portal hypertension (PH) etiology because the patient had no known history of liver disease.
- She had a repeat EGD one month later to evaluate for eradication of varices. Endoscopic ultrasound (EUS) was also performed at that time for liver biopsy and portal pressure gradient (PPG) measurements. The calculated EUS-guided portal pressure gradient was 18.3mmHg.

Endoscopy

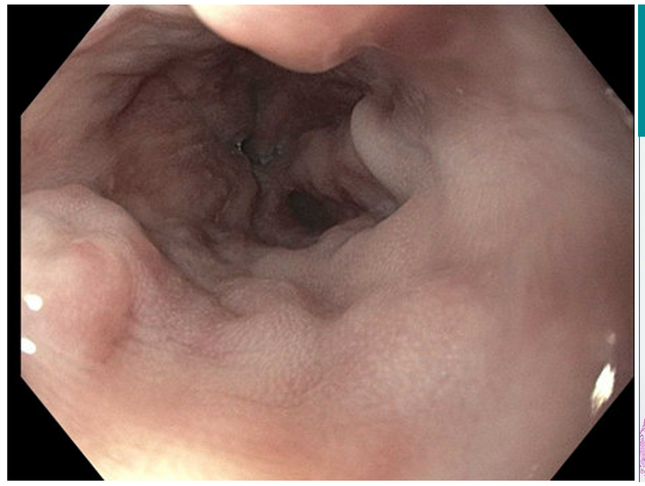

Image 1. Esophageal varices

Pathology

Multiple ductal plate malformations- ectatic bile ducts embedded in fibrous stroma H&E 10x

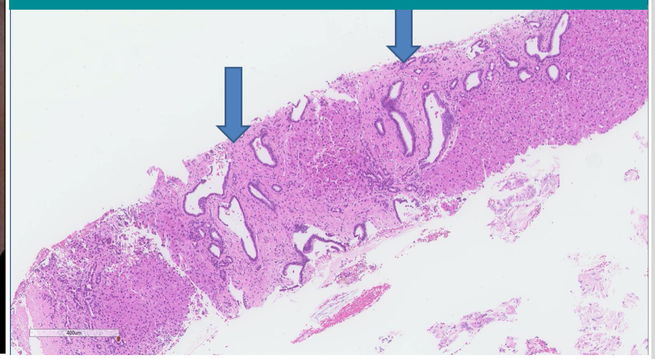

Image 2. Ductal plate malformation

EUS

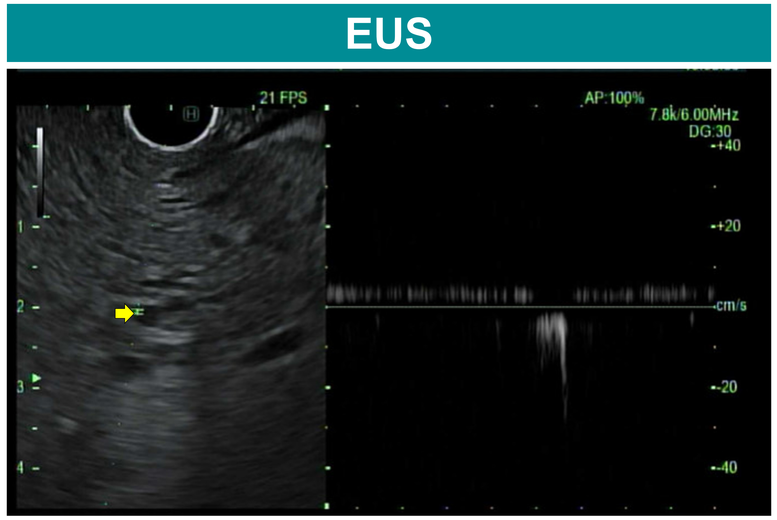

Image 3. Doppler with portal waveform. Note that liver has honeycomb pattern and heterogenous echotexture.

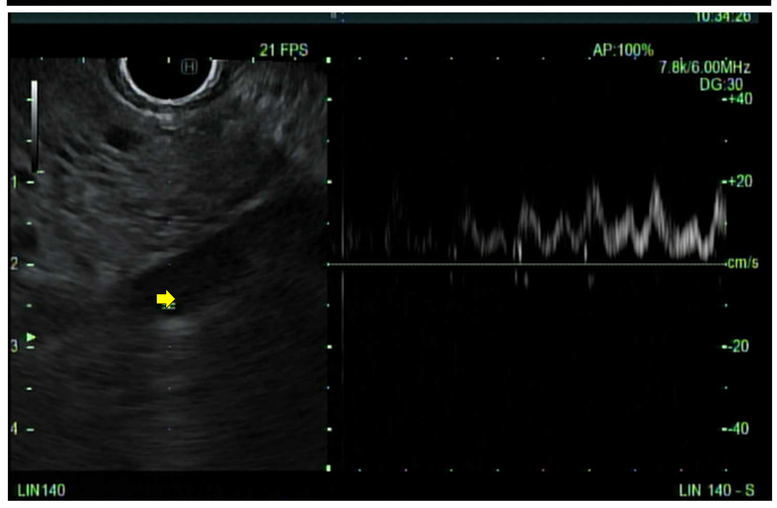

Image 4. Doppler with waveform of hepatic vein.

Findings

- The sonographic findings of the liver revealed a diffuse abnormal echotexture which was characterized by a heterogenous appearance.
- Liver biopsy revealed a benign hepatic parenchyma with multiple ductal plate malformations. This finding is consistent with congenital hepatic fibrosis which has a known association with ARPKD.

Discussion

- This young patient's presentation with esophageal varices was rather perplexing considering she had no known history of liver disease, alcohol dependence, or known risk factors for PH. The key to identifying the cause of her PH was her history of ARPKD.
- The novel method of EUS-guided PPG measurement was used to evaluate PH severity and provided helpful information without the need of an additional procedure.
- Liver biopsy confirmed the diagnostic suspicion. Approximately 10% of ARPKD patients ultimately require liver transplantation.[3]

References

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