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CASE REPORT | LIVER

# Congenital Absence of Portal Vein Presenting With Hematemesis in a Young Patient

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## ABSTRACT

Upper gastrointestinal bleeding is caused by multiple different etiologies, with one being portal hypertension. Identifying the underlying cause of portal hypertension is extremely important in determining further management. This is a rare case of a patient who had developed portal hypertension secondary to a congenital absence of a portal vein with no other medical history. This case highlights a rare cause of portal hypertension and the need for prompt medical management.

**KEYWORDS:** portal hypertension; bleed; varices

## INTRODUCTION

Upper gastrointestinal (GI) bleeds have multiple different etiologies and can vary depending on age and clinical presentation. The most common causes in school-aged children include esophagitis, gastritis, esophageal varices, peptic ulcer disease, and Mallory-Weiss tears.[1] Esophageal varices tend to account for 6%–20% of upper GI bleeds in children, most commonly due to portal hypertension, with prehepatic etiologies as the number one contributor to portal hypertension in pediatrics.[1,2] The literature has focused on portal vein thrombosis as a known prehepatic cause of portal hypertension; however, congenital absence of a portal vein (CAPV) is extremely rare and typically associated with other congenital anomalies.[3] Thus, we report a rare case of a pediatric patient who was found to have absence of a normal portal vein in an otherwise healthy child.

## CASE REPORT

Our patient is a 7-year-old girl born term with no medical history who presented with 1 episode of hematemesis. The patient recently immigrated from Jamaica where she had an episode of hematemesis 1 year ago. Laboratory results showed normal liver enzymes, slightly elevated coagulation factors with international normalized ratio of 1.33, prothrombin time of 14 seconds, and hemoglobin of 6.5 g/dL. She received 10 mL/kg blood transfusion, sandostatin bolus, and pantoprazole. Ultrasound of the abdomen showed a dilated vein along the lesser curvature of the stomach, with limited view of the main portal vein. She required additional blood transfusions and, ultimately, was taken to the operating room for esophagogastroduodenoscopy (EGD). EGD was significant for Grade 4 esophageal varices with red signs present, no active bleeding, and 3 bands were placed around the dilated vessels. There was also evidence of small gastric varices. Extensive liver workup was negative for autoimmune and infectious causes. Magnetic resonance imaging/magnetic resonance angiography showed splenomegaly, gallbladder wall thickening, moderate ascites, gastric varices along the lesser curvature, and absence of a normal portal vein with a venous structure demonstrating significant tapering and branching into at least 2 small vessels shortly after the confluence of the superior mesenteric and splenic veins Figures 1–5. In addition, a venous structure arising from the superior mesenteric vein coursed toward the hepatic hilum. Hemoglobin remained stable, and she was discharged to follow-up for interval EGD with banding. Ultimately, thrombosis workup revealed protein C and S deficiency.

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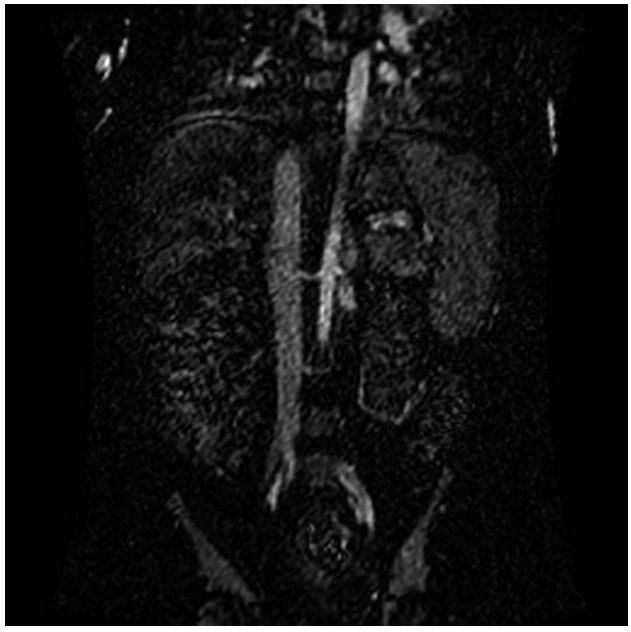

**Figure 1.** Normal aorta and inferior vena cava with absent portal vein.

## DISCUSSION

This patient did not have evidence of a thrombus on imaging, as this is a more common cause of hematemesis in this pediatric age group, but instead presented with absence of a portal vein. CAPV is extremely rare with an incidence rate of 1 in every 30,000 newborns, in which the mesenteric venous blood flow drains directly into the systemic circulation.[4] The portal vein develops embryologically between the 4th and 10th weeks. CAPV may result from an embryologic insult, which causes defect of the cardiovascular system, and complicated cardiogenesis could be affected by the insult or the systemic diversion of portal venous flow.[5] Morgan and Superina (1994) proposed a classification of portosystemic anomalies into the following: Type I (liver are not perfused with portal blood because of complete shunt) and type II (liver perfused with portal blood in the presence of a partial shunt).[3] When the portal vein is absent, toxic metabolites such as ammonia and bile acids collected from the GI tract had to bypass the liver directly draining into the systemic circulation, thus may initiate hepatic encephalopathy.[5]

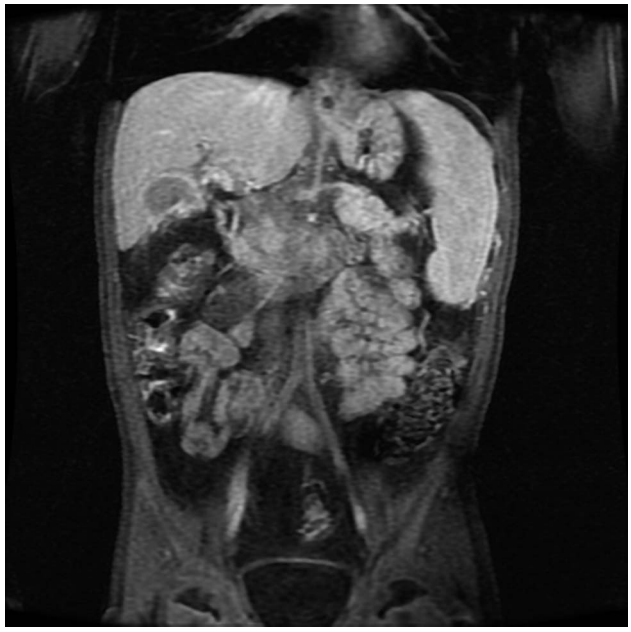

**Figure 2.** Postcontrast view of the abdomen showcasing collateral vessels in the porta hepatis where the main portal vein should be.

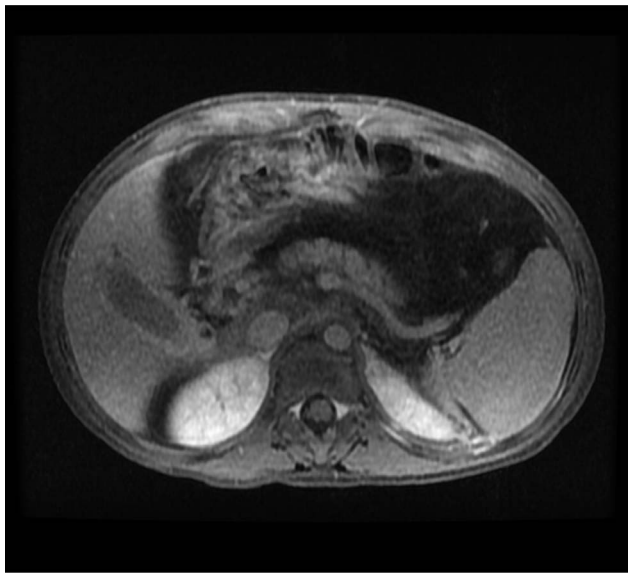

**Figure 3.** Splenic vein communication with the superior mesenteric vein, which tapers off above this level.

The first account of CAPV was given by a London surgeon, John Abernethy in 1793.[3] Of published case reports, 30% of patients are identified before the age of 5 years, although diagnosis can

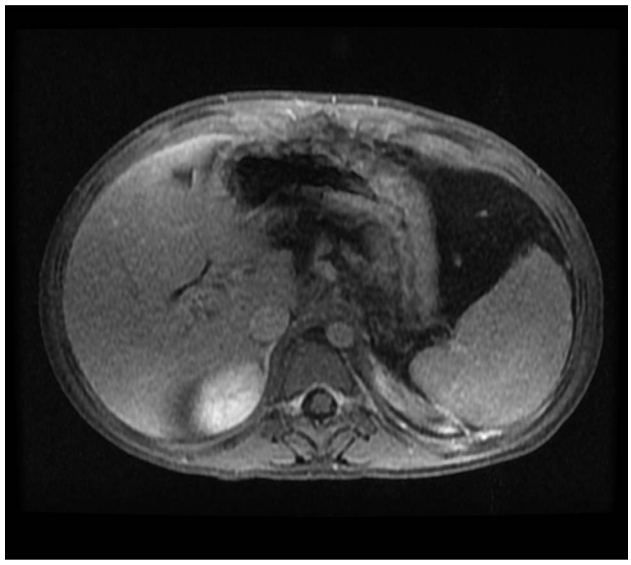

**Figure 4.** Postcontrast view of superior mesenteric vein. No communication with main portal vein.

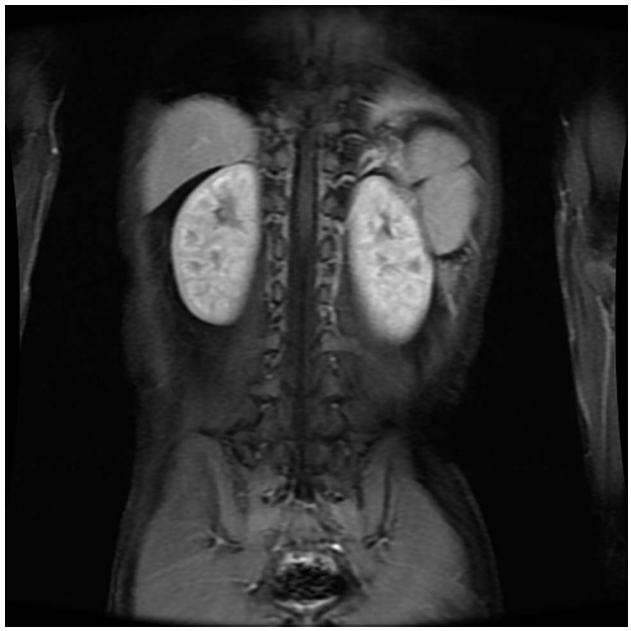

**Figure 5.** Showcasing large collaterals above the left kidney.

range between the neonatal period and adolescence. Notably, however, more than 22% of patients additionally had congenital heart disease, with others having associated anomalies of spleen, urinary and male genital tract, and brain and skeletal abnormalities.[3] Medical management is extremely important in preventing significant morbidity in this population. Complications can range anywhere from worsening liver function, portal hypertension, hepatic encephalopathy, pulmonary hypertension, GI bleeding from varices, increased risk of liver tumors, and further systemic symptoms from bypassing of blood from the intestines and spleen directly into systemic circulation without processing from the liver.[5]

Management should be stressed on a case-by-case basis, depending on the type or anatomy of the disease, as well as the symptoms and the clinical conditions of the patient.[5] Our patient had no additional anomalies or significant medical history before presentation with an episode of emesis. Thus, it highlights a rare presentation of CAPV and the need for prompt investigation to prevent further complications.

## DISCLOSURES

Author contributions: L. Fawaz and N. Al-Ansari contributed to writing and editing the manuscript. J. Davis provided radiological images with explanations for publication. L. Fawaz is the article guarantor.

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All attempts have been exhausted in trying to contact the patient, next of kin, and/or parent/guardian for informed consent to publish their information, but consent could not be obtained. Additionally, the patient is no longer living in the country.

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