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Original Research Paper

Radio-Diagnosis

ABERNETHY MALFORMATION PRESENTING WITH LIVER MASS : A RARE PRESENTATION OF A RARE ENTITY

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ABSTRACT Abernethy malformation is a very rare congenital extrahepatic porto-systemic shunt (CEPS) that develops between portomesenteric vasculature and systemic veins (IVC / left renal vein / left iliac vein / left hepatic vein) and thus bypassing the liver and resulting in deranged metabolism of various metabolites. They have clinical features varying from that of hypergalactosemia to hyperbilirubinemia to hyperammonemia and pulmonary veins congestion, hepatopulmonary syndrome, hepatic encephalopathy. Another presentation in the spectrum is patient presenting with a hepatic mass lesion, which form as a liver's response to absent/reduced portal flow leading to regenerative nodular hyperplasia. These nodules with time can progress into hepatic tumors like adenoma ,focal nodular hyperplasia(FNH), hepatoblastoma and even hepatocellular carcinoma(HCC). Ultrasound (USG) is a useful tool for screening of congenital anomalies associated with the liver. Diagnosis of abernethy malformation requires a high index of suspicion and can easily be overlooked if not carefully evaluated. In the majority of cases, the complications and other secondary features often lead to diagnosis of abernethy malformation. Also,we should emphasize the search for an underlying abernethy malformation on the background of hepatic mass(as in our case). We report a case of abernethy malformation presenting with liver mass on screening USG and confirmed on contrast enhanced computed tomography (CECT).

KEYWORDS : Abernethy malformation, Congenital extrahepatic porto-systemic shunt, hepatic mass, focal nodular hyperplasia

INTRODUCTION

Abernethy malformation is a very rare congenital vascular malformation defined by diversion of portal blood away from the liver. Portal vein (PV) is formed between 5 to 10 weeks of intrauterine gestation by selective involvement of peri intestinal vitelline venous loop. Intra hepatic Inferior Vena Cava (IVC) develops as an amalgamation of several embryological venous channels with Common hepatic vein. Excessive involution between 5 to 10 weeks of intrauterine gestation leads to complete absence of PV which results in CEPS with abnormal mesentrocaval connection.

We present a case of a 6.5 year female child who presented with liver mass and diagnosed as type 1b Abernethy malformation, presenting with liver mass (FNH).

CASE REPORT

A 6.5 year old female child was referred to our center for biopsy of a liver mass which was detected incidentally in a screening USG advised in routine workup of her UTI. On retrospective evaluation of her previous reports, we found that AFP, bilirubin and INR levels were normal. On the abdomen USG, we found a well defined iso to hypoechoic rounded mass lesion in the right lobe of the liver and the background liver was non cirrhotic . On careful evaluation, a suspicious communication of the portal vein to IVC was noted. CECT abdomen was advised for further characterization of our USG findings.

Abdominal CECT was then performed to evaluate this suspicious vascular communication and to characterize the mass lesion seen on USG.

CT images demonstrated formation of Portal vein (PV) by confluence of Splenic Vein (SV) and Superior Mesenteric Vein (SMV) at the neck of pancreas . PV was thereafter seen to directly communicate with IVC and the portal radicles were absent; suggestive of type 1b Abernethy malformation. (Figure 1).

A well defined mass lesion was noted in the right lobe of the liver , measuring 62x55 mm, with bright homogeneous contrast enhancement on the arterial phase which persisted on delayed phases. The central scar of the lesion remained low-attenuation on early phases and demonstrated mild enhancement on delayed image . The imaging findings were suggestive of FNH (Figure 2).

Patient was advised liver biopsy to confirm the diagnosis of type 1b Abernethy malformation and was counseled for future follow ups and need for liver transplantation.

DISCUSSION

John Abernethy first reported congenital absence of portal vein and congenital mesenteric-caval shunt in 1793.[1] Abernethy malformation is a congenital extrahepatic portosystemic shunt that develops between portomesenteric vasculature and systemic veins (IVC / left renal vein / left iliac vein / left hepatic vein) and thus bypassing the liver and resulting in deranged metabolism of various metabolites .The patients thus present with clinical features varying from that of hypergalactosemia to hyperbilirubinemia to hyperammonemia and pulmonary veins congestion, hepatopulmonary syndrome, hepatic encephalopathy.

Another presentation in the spectrum is patient presenting with a hepatic mass lesion , which form as a liver's response to

absent/reduced portal flow leading to regenerative nodular hyperplasia.These nodules with time can progress into hepatic tumors like Adenoma , FNH, Hepatoblastoma and even Hepatocellular carcinoma (HCC).[2] The majority of these lesions were characterized as benign , such as FNH (36.73%). Other reported lesions included nodular regenerative hyperplasia (16.33%), hepatoblastoma (4.08%), hepatic adenoma (10.20%), hepatocellular carcinoma (26.53%) and cirrhosis (6.12%).[3]

Morgan and Superina classification (Figure 3) of CEPS divided it into type 1 and type 2 based on presence/absence of intrahepatic PV branches .

Type 1: Complete diversion of portal blood into the inferior vena cava with congenital absence of the portal vein .
1a: Superior mesenteric vein and splenic vein do not join to form a confluence
1b: Superior mesenteric vein and splenic vein join to form a confluence
Type 2: Intact portal vein (may be hypoplastic) with some portal flow diversion to the inferior vena cava through side-to-side extrahepatic communication .

Early diagnosis and treatment are important to prevent complications and thus we need to keep in the back of mind that Abernethy malformation can be seen on the background of a liver mass .

The planning of treatment is dependent on the type of shunt as classified by Morgan and Superina[4] and needs to be tailored to the individual patient in accordance with preoperative evaluation.[5] Usually in patients with CEPS type I, occlusion of shunt is not performed, as it is the only drainage route for the mesenteric venous blood. But recently published experiences by several authors[5-6] point out that many patients with CEPS type I malformations might have small portal vein radicals which cannot be seen on ultrasonography but could be visualized on shunt angiography. The balloon occlusion test of the fistula can also be done.[5-6] This test helps to decide on a single stage or a two-staged shunt closure procedure. A two-step procedure allows the portal branches to enlarge slowly and can avoid acute severe portal hypertension.[5-6] Extremely hypoplastic or undetectable portal veins will require banding of the fistula before closure.[6] Shunt closure results in restoration of intrahepatic portal flow in most patients.[6] Clinical improvement in the form of regression of benign liver masses, and regression or stabilization of pulmonary, cardiac, neurological, and renal complications is seen in patients post shunt.[6] CEPS type I patients also need clinical, biochemical and imaging follow-up[6]; as is done in our patient. For patients with CEPS type I developing complications like encephalopathy or neoplasms till recently liver transplant was the only treatment option[5,7-9], but transplant should be reserved for exceptionally complex anatomy where closure of the shunt is not possible.

Patients with CEPS type II with hepatic encephalopathy can benefit by early shunt occlusion surgery.[5-6] Reconstruction of the portal vein should be done early to avoid mesenteric venous congestion.[9] Shunt surgery when possible is the treatment of choice for CEPS type I and type II.

CONCLUSION

Ultrasound is a useful tool for screening of congenital anomalies associated with the liver. Diagnosis of Abernethy malformation requires a high index of suspicion and can easily be overlooked if not carefully evaluated. In the majority of cases, the complications and other secondary features often lead to diagnosis of Abernethy malformation. Also, we should emphasize the search for an underlying Abernethy malformation on the background of hepatic mass(as in our case).Prognosis of the patients with congenital portosystemic shunts depends on the site of the shunt as determined by Morgan and Superina classification, the associated congenital anomalies and the extent of liver disease. Many patients will benefit from shunt surgery. The extent of associated abnormalities should not deter pediatricians from referring patients for treatment. A long-term follow-up is indicated for all asymptomatic patients of Abernethy malformation.

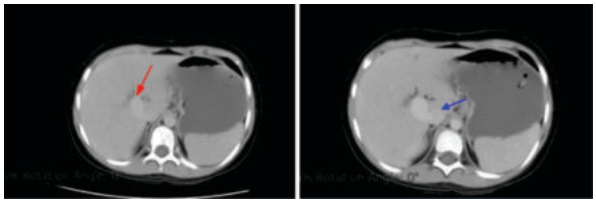

Figure 1a **Figure 1b**

Figure 1; Figure 1a-axial C+arterial phase, Figure 1b-axial C+portal venous phase: The axial CECT images demonstrate dilated and anomalous portal vein (red arrow) draining into the IVC (blue arrow) and absent portal radicles in the liver parenchyma.

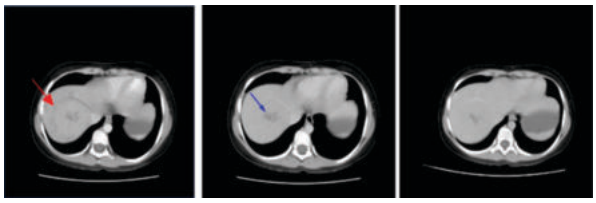

Figure 2a **Figure 2b** **Figure 2c**

Figure 2 ; Figure 2a-axial C+arterial phase, Figure 2b-axial C+portal venous phase, Figure 2c-axial C+delayed phase : The axial CECT images demonstrate a well defined rounded mass lesion in the right lobe of the liver (red arrow) with bright homogeneous contrast enhancement on the arterial phase which persisted on delayed phases. The central scar (blue arrow) of the lesion remained low-attenuation on early phases and demonstrated mild enhancement on delayed image. No CT features of cirrhosis are noted in the background liver .

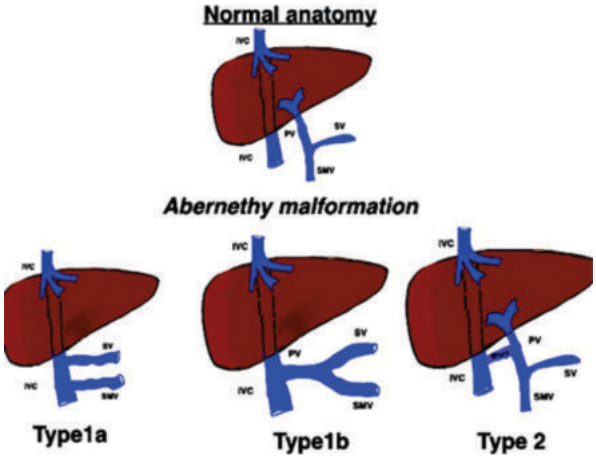

Figure 3

Conflict of Interest- None declared

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