Case Report

Abernethy malformation: A rare presentation in the elderly

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ABSTRACT

Abernethy malformation or congenital extrahepatic portosystemic shunt is a rare clinical entity, and its initial manifestation in adults is still rarer. Here, we report the case of a 68-year-old male who presented with encephalopathy and got admitted to the ICU. All the regular radiologic and metabolic workups for encephalopathy were negative except for high ammonia. Further, evaluation with contrast-enhanced computed tomography of abdomen showed hypoplastic portal vein with a large portacaval shunt, suggestive of type 2 Abernethy malformation. The encephalopathy associated with hyperammonemia improved with anti-encephalopathy measures.

Key words: *Abernethy malformation, Hepatic encephalopathy, Hyperammonemia*

Abernethy malformation is a rare clinical condition and also termed as congenital extrahepatic portosystemic shunt (CEPS). Approximately <200 cases of CEPS were reported worldwide [1]. Shunting of portal venous blood to systemic venous system occurs due to the vascular developmental abnormalities. It was John Abernethy who described this condition in a 10-month-old child for the first time in 1793 [1].

Two types of CEPS have been described depending on communication between the portal vein and inferior vena cava (IVC). Intrahepatic portal venous supply also plays role in classification as described by Morgen and Superina. Type 1: Complete drainage from portal venous system to IVC and an intrahepatic portal vein is absent. It is common in females and associated with cardiac, viscera, and skeletal anomalies. Type 2: Partial drainage from portal venous system to IVC by side-to-side anastomosis. The portal vein is usually hypoplastic in this situation. There was no association with anomalies [2-5].

CASE REPORT

A 68-year-old gentleman with a background history of hypertension for the past 20 years and transient ischemic attack 8 years back, presented to the department with complaints of irrelevant speech and altered sensorium for 3 days. There was no history of alcohol abuse or any drug intake which could lead to altered sensorium. Physical examination revealed pulse rate of 92/min, blood pressure of 150/100 mm-Hg, and respiratory rate of 16/min. The patient was afebrile and was maintaining saturation on room air. He was drowsy and arousable with no focal neurological deficits.

Liver function testing revealed total bilirubin level of 2 mg/dl (normal 0.10–1.20 mg/dl), direct bilirubin of 0.3 mg/dl (normal 0–0.4 mg/dl), aspartate transaminase of 47 IU/L (normal 5–37 IU/L), alanine transaminase of 22 IU/L (normal 10–50 IU/L), and alkaline phosphatase of 151 IU/L (normal 45–135 IU/L). Total protein was 6.7 g/dl (normal 6–8.2 g/dl), and serum albumin was 3.3 g/dl (3.5–5.2 g/dl). Hematological studies revealed hemoglobin - 12 g/dl, prothrombin time - 12.1 s, international normalized ratio - 1.1, and activated partial thromboplastin time - 31.2 s. Sodium was 134 mmol/L (normal 135–145 mmol/L), ionized calcium 4.5 mg/dl (normal 4.1–5.2 mg/dl), and magnesium 1.8 mg/dl (normal 1.7–2.4 mg/dl). Magnetic resonance imaging (MRI) of the brain with angiogram revealed basilar artery narrowing and no evidence of infarct. Cerebrospinal fluid analysis ruled out the possibility of neuroinfection. Thyroid-stimulating hormone was 1.420 micIU/mL (0.340–4.250). Serum cortisol was normal, and serum ammonia was elevated 276.0 mcg/dl (27.0–102.0 mcg/dl). Serological markers for hepatitis B and C were negative. Serum alpha-fetoprotein was normal. An ultrasound abdomen did not reveal any evidence of chronic liver disease.

The patient was initiated on anti-encephalopathy measures (lactulose to target 3–4 loose stools per day and rifaximin 400 mg thrice daily), and over 48 h, his sensorium improved. Repeat serum ammonia was 147 mcg/dl. On the day 4 of hospital stay in view of drowsiness, repeat ammonia was done, which was 210 mcg/dl. The vascular anomaly of splanchnic venous system and urea cycle disorders ware considered in view of hyperammonemia. Contrast-enhanced computed tomography of the abdomen showed small calibered (5 mm) but patent portal vein. Dilated venous collaterals from the superior mesenteric vein, draining into the left renal

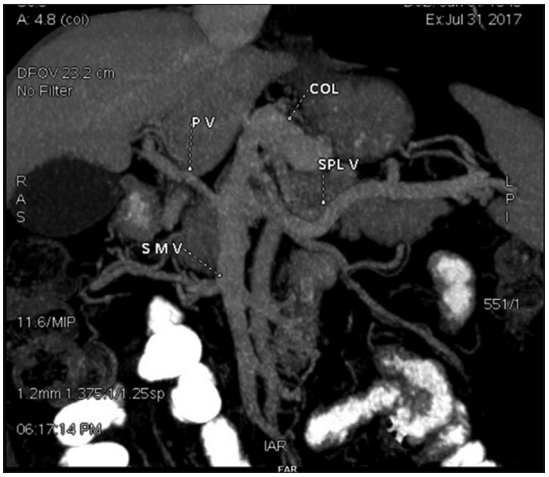

Figure 1: Dilated venous collateral from the superior mesenteric vein, draining into left renal vein, and inferior vena cava suggestive of type 2 Abernethy malformation

vein, and IVC were suggestive of type 2 Abernethy malformation (Fig. 1). The family was counseled regarding the need for surgical or endovascular interventions. However, the family refused consent to undergo surgical or endovascular intervention. He was discharged on day 5 and did not come for further follow up.

DISCUSSION

An extrahepatic portosystemic shunt was first described in 1982 in a 67-year-old lady. She had no cirrhosis. The shunt was noted between superior mesenteric vein and IVC, and around 15 similar cases have been reported till 2015 [6]. In a pediatric case series, nearly 316 case reports were available until 2013. Type 1 is more common in children. Abernethy detection is incidental in neonatal age group during the workup of cardiovascular, skeletal, and visceral anomalies. The right-sided heart and the right aortic arch were described by Abernethy in his first report [3]. Biliary and duodenal atresia, choledochal cyst, intrahepatic gallbladder, and polysplenia are the associated important visceral anomalies. Genitourinary abnormalities can also be observed among type 2 patients [3,5].

Adults may or may not have clinical manifestation depending on the shunt ratio between the portal and systemic circulation and a ratio of >60% usually associated with hepatic encephalopathy. Advancing age may also have bearing on clinical manifestation of encephalopathy [4], and 15% cases can have only encephalopathy at presentation [5]. Abernethy malformations can have associated with focal nodular hyperplasia and hepatic adenomas. Hepatocellular carcinoma has also been described [3,5]. Diagnosis can be made using Doppler USG, computed tomography, or MRI. Portal venography may be superior in identifying pressure gradient as well as portal venous anatomy.

Type 1 shunts without clinical manifestations need regular follow-up with imaging and blood investigations. Imaging plays important role in diagnosis as patients with type 1 shunts can present with varied manifestations in childhood involving liver (nodular lesions, adenoma, hepatocellular carcinoma, and cholestasis), central nervous system (irritability, behavioral disorder, dyslexia, EEG abnormalities, and epilepsy), pulmonary hypertension, metabolic (hyperammonemia, hypoglycemia, and hyperinsulinemia), and multiorgan involvements. Type II shunts present predominantly as hepatic encephalopathy and liver dysfunction, with pulmonary hypertension as a secondary complication.

Shunt occlusion can be dangerous as intestinal, and splenic venous drainage is dependent on shunt [7]. Liver transplant remains the ultimate option in case of fulminant hepatic failure. Type 2 malformations can be tackled surgically. Endovascular intervention using percutaneous transcatheter coiling remains the other treatment option [8]. Anti-encephalopathy measures such as lactulose can be used for clinical improvement [6]. Our patient showed clinical improvement with anti-encephalopathy measures. Surgical or endovascular intervention was not performed.

CONCLUSION

An adult patient with hepatic encephalopathy in the absence of cirrhosis and preceding risk factors needs to be evaluated for Abernethy malformation or large spontaneous portosystemic shunts.

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