Case report

Abernethy Syndrome with Left Renal Artery Stenosis: Our first experience

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Received Date: 26 June 2023
Accepted Date: 18 July 2023
Published Date: 28 Sep 2023
DOI: https://doi.org/10.3329/jrpmc.v8i2.69389

Abstract
Abernethy syndrome is defined as the congenital diversion of portal blood away from the liver either end-to-side shunt or, side-to-side shunt. Here we are reporting a case of a 15-year-old boy presented with recurrent encephalopathy and uncontrolled hypertension. Persistent rise in blood ammonia level was noted with normal other biochemical parameters. Ultrasound and Doppler study of the abdomen revealed drainage of the portal vein into the inferior vena cava and a small size of the left kidney. Renal artery angiogram and hepatic angiogram were performed, which confirmed the diagnosis of Type 1B Abernethy syndrome with left renal artery stenosis. After counseling the patient and his parents and taking consent, he has been treated with Left renal artery angioplasty, antihypertensive along supportive treatment. The treatment of Abernethy syndrome depends on its type, associated congenital anomalies, and complications. If possible closer of thes hunt is the mainstay of treatment but in case of type 1 malformation with the absence of the possibility to close the shunt may require a liver transplant.

Keywords: Abernethy syndrome, Renal artery stenosis, Ultrasound, Hepatic angiogram, Liver transplant

Introduction:

The Abernethy malformation was first reported by John Abernethy in 1793.[1] It is a rare congenital malformation of the portal venous system. It can be divided into two types (types 1 & 2). In Abernethy malformation type 1, all the portal venous blood is drained into the inferior vena cava and there is no intrahepatic portal vein [Figure-1]. In type 2, the portal vein is partially drained into the inferior vena cava via side-by-side anastomosis [Figure-1].[2] Type 1 malformation further divides those in which splenic and superior mesenteric veins drain separately into a systemic vein (type 1a) and those in which the two veins form a common trunk that drains into the systemic circulation (type 1B) [Figure-1].[3] Patients may suffer from some complications, including hepatic encephalopathy, hepatopulmonary syndrome, portopulmonary hypertension, and even hepatocellular carcinomas.[4] Imaging has an important role in the diagnosis and follow-up of this malformation.[5] In this article, we describe a patient diagnosed with type 1B Abernethy malformation.

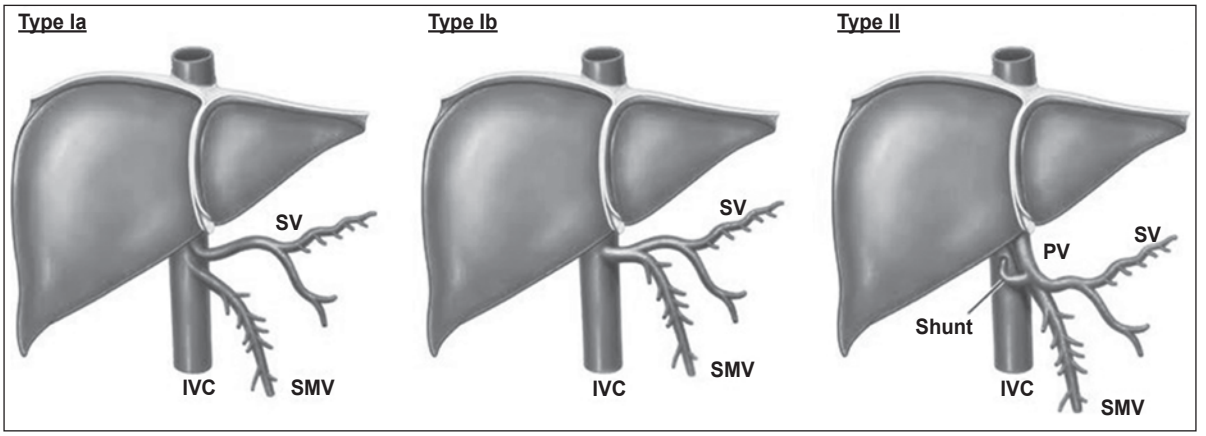

Figure-1: Different types of Abernethy Syndrome

Case presentation:

A 15-year-old boy was admitted to the medicine department of Rangpur Medical College & Hospital for the evaluation of uncontrolled hypertension for 6 years and recurrent encephalopathy. At the age of 10 months in 2009, his parents consulted with a Pediatrician, and he was diagnosed as a case of pneumonia and heart failure & managed with antibiotics, nebulization, and diuretics. His symptoms improved with this treatment. Up to 2 years of his age, he took heart failure medication. Age (02-08) years, he complains of anorexia, frequent vomiting, and feeling sick. Sometimes, he stopped eating for a few days due to vomiting and anorexia. His mother noticed that his intellectual development not growing properly. Treated with IV fluid, ORS, Vitamin, and antiemetic. His parents consulted with many physicians but yet not establish any diagnosis. Symptomatic treatment was given. At the age of 09 years in 2017, he was admitted into the Gastroenterology Department of Rangpur Medical College & Hospital with complaints of an altered level of consciousness for 12 hours. Diagnosed as Encephalopathy and under evaluation. All available investigation was done, but etiology was not identified. Improved after 3 days with supportive treatment. At the age of 10 years in 2018, he consulted with a Physician for headache and dizziness. Blood pressure was 170/100 mmHg recorded. He was diagnosed as a case of Hypertension & managed with Olmesartan 10mg OD but his blood pressure was not well controlled. At the age of 12 years in 2020, he consulted with the Medicine unit in Narayana Health City, India for his uncontrolled hypertension & diagnosed with Hypertension due to Left Renal Artery stenosis. Along with some incidental congenital abnormality of the portal vein (portal vein forming aneurysmal dilatation at the end and drain into IVC). Left Renal artery angioplasty was done. All of his biochemical and hematological reports were normal except the Blood ammonia level, which was persistently raised. Discharged with Tab Aspirin 75mg+clopidogrel 75mg OD, Tab. Amlodipine 5mg + Olmesartan 20MG OD. After 2 months of getting the above treatment, he developed melena and an altered level of consciousness. At that time he managed in the ICU of Rangpur Medical College & Hospital with blood transfusion & supportive treatment after diagnosis of Asprin-induced antral erosion and Encephalopathy due to Abernethy Syndrome. Advice to stop Aspirin. He had a history of 3 episodes of encephalopathy. Recovered after supportive management without any residual neurological deficit. He is nondiabetic, with no history of jaundice, hypoglycemia, electrolyte imbalance, fever, convulsion, or abnormal movement. On query, he gave h/o occasional tremor, dyspnea on exertion, and easy fatigability. He was born as a preterm low birth weight baby. His school performance is poor. History of 5 unit blood transfusion during his illness. Parental consanguinity absent. On examination, he is cooperative, BMI – 19kg/m2, Anaemia, jaundice, cyanosis, clubbing, edema – absent, pulse – 80 bpm, blood pressure – 140/90 mm Hg without a postural drop, bedside heat coagulation test is negative. Fundoscopy – Grade 2 hypertensive retinopathy. Other systems are quite normal. 0n investigations, Hb – 12.4 gm/dl, MCV

– 88fl, Platelet -80,000, ESR – 08 mm in 1st hour, Blood Film – Anisopoikilocytosis, thrombocytopenia, Urinalysis – Normal, Blood Sugar – 5.4 mmol/L, Chestxray PA view – Normal, ECHO-Trivial TR& concentric LVH, PT-15.8 sec, S.Albumin – 3.6 gm/dl, ALT – 30 U/L, AST-27 U/L, ALP- 127 U/L, ANA-Negative, Plasma Ammonia – 71 micromol/L (10-30), HBsAg– Negative, Anti HBc total– positive, HBV DNA – not detected, Anti HBsAg – positive(>250mlU/ml), Anti-HCV- negative, USG of W/A- Normal, Ceruloplamin level-normal, CT scan of Brain – Normal, DTPA renogram- 25% left kidney (eGFR-26.26ml/min), 75% right kidney (eGFR-80.48 ml/min), Fibroscan of Liver – 5 kPa which is within normal limit. Perform CT abdomen and CT hepatic angiogram- observation was evidence of fusiform aneurysmal dilatation of the distal end of the extrahepatic main portal vein with the absence of intrahepatic portal segment. There is an abnormal side-to-side shunt between extrahepatic PV and IVC & patent left renal stent, CT chest-Features of mild PAH, Upper GI Endoscopy – Mild congestive gastropathy. Our final diagnosis is Abernethy syndrome 1B with portopulmonary hypertension and secondary Hypertension due to Left renal artery stenosis with post-angioplasty status. On-going treatments are Amlodipin 5mg OD, Bisoprolol 5 mg BD, Rifaximin 550 mg BD, Cholestyramine 4g OD, and Lactulose BD when needed.

Discussion:

Abernethy malformation is a rare congenital communication between the portal vein and systemic circulation. It has female dominance and is usually diagnosed during childhood.[6] Some patients remain asymptomatic throughout their life. When symptoms do occur, the clinical manifestations are mainly associated with the hepatic shunt and associated congenital malformations. Metabolic and vasoactive substances bypass the liver through portosystemic collaterals and thus cause portopulmonary hypertension, hyperammonemia, and encephalopathy.[7] Nodular liver lesions are seen in up to half of patients: these include benign focal nodular hyperplasia, hepatocellular adenoma, and regenerative nodules and may be explained by the absence of portal flow and compensatory increased hepatic arterial blood flow.[8] Although most of these lesions are benign, there are several case reports in the literature describing the coexistence of hepatocellular carcinoma and hepatoblastoma. Therefore, long-term follow-up and monitoring are recommended.[9] Congenital hepatic shunts can also present with metabolic dysregulation, such as hypoglycemia, due to metabolic alterations in the liver.[10] Despite the existence of a direct connection between the portal and systemic venous systems, hyperammonemia is found in only 26% of patients with the Abernethy malformation and only 15% of all patients experience hepatic encephalopathy.[11] Hyperammonemia can be present without encephalopathy, especially in younger patients; clinical encephalopathy is more common at older ages. Possible explanations for this phenomenon are an age-dependent increase in sensitivity to deleterious metabolites and the impact of the extent of shunting, determined by the portal/systemic shunt ratio of more than 60% may predict the age of onset of encephalopathy.[12] Some congenital anomalies are associated with congenital heart disease, polysplenia, renovascular anomaly, biliary atresia, annular pancreas, situs inversus, and duodenal atresia.[13] Diagnosis is usually confirmed by imaging techniques like ultrasonogram, CT scan, MRI, and CT angiogram.[4] Treatment may vary from surgical correction of the shunt to even liver transplan tation.[14]

In this case report, our patient is male and he was diagnosed with Abernethy syndrome 1B in his childhood. He presented with Encephalopathy and recurrent hyperammonemia from his early childhood. He subsequently developed portopulmonary hypertension. Our patient has left renal artery stenosis as an association. No other complications are noted, but we are advised for meticulous follow-up in the future and advice for liver transplantation if severe Encephalopathy and complications develop.

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