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Case Study

TYPE 1B ABERNETHY MALFORMATION IN AN ADOLESCENT MALE: A CASE REPORT

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ABSTRACT

Abernethy malformation or congenital extrahepatic portosystemic shunt is a rare genetic disorder characterised by bypass of portal venous blood into the systemic circulation. This rare vascular anomaly is classified into two types based on the pattern of vascular shunting if left untreated, it can lead to severe complications. We report the case of 17 y old male patient admitted in CCU with complaints of shortness of breath and chest pain for 1 w with history of chest fullness and breathlessness. On evaluation, Lab parameters revealed pancytopenia and altered liver profile. CECT of abdomen showed multiple collaterals venous channels with large splenorenal shunt consistent with portal hypertension these findings confirmed a diagnosis of type 1b Abernethy malformation. Patient was managed conservatively with symptomatic medical treatment, no surgical or interventional radiological procedure was performed, after few days patient cardiac parameters were stable with no significant improvement in overall disease condition, discharged with follow up for further management.

Keywords: Abernethy malformation, Congenital portosystemic shunt, Splenorenal shunt, Type 1b CEPS

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INTRODUCTION

Abernethy malformation, also known as congenital extrahepatic portosystemic shunt is a rare anomaly is characterised by partial or complete diversions of the portal blood into the systemic venous circulation, this congenital anomaly is mainly observed in childhood and is often associated with neonatal cholestasis or congenital heart disease [1, 2].

Patients may develop hepatic encephalopathy, pulmonary hypertension or hepatocellular carcinoma (HCC) however, the actual incidence of such complications is unknown [3, 4].

Abernethy malformation is of two types type 1 and type 2 depends on shunting in the blood vessels type 1 includes complete diversion of portal blood. Type 1a characterised by lack of portal veins and presence of patent ductus venosus, superior mesenteric vein (SMV) and splenic vein (SV) drain separately into a systemic vein [5].

Type 1b characterised by presence of multiple collaterals between mesenteric and systemic veins allow portal circulation bypass. Type 2 characterised by a side-to-side connection (shunt) exists between a patent portal vein and systemic vein (e. g., IVC) allowing small amount of portal blood to reach the liver [6, 7].

CT and MRI play the major role for diagnosing this disease early diagnosis is very important if left untreated it can causes hepatocellular carcinoma, severe pulmonary hypertension [8].

CASE REPORT

A 17 y old male patient admitted in cardiac ward in cardiac care unit A of Geetanjali Medical College and Hospital, Udaipur Rajasthan on 15 October 2024 with chief complaint of shortness of breath and chest pain since 7 d. Patient was asymptomatic 7 d before then he developed symptoms of shortness of breath (SOB) associated with chest pain occasionally which got aggravated during lying position and feeling burning in chest sensation after eating, patient has history of feeling of chest fullness and breathlessness after running. During admission the patient present with the increased blood pressure of 148/94 mmHg and tachycardia heart rate (HR) was 108 bpm indicating primarily patient has cardiac issues.

On physical examination it revealed patient with pallor positive on face with afebrile condition patient was conscious and oriented. During admission the oxygen saturation level was found to be 86% which primarily indicates to pulmonary issues associated with heart.

Lab investigations on day 1 of admission revealed patient had pancytopenia with a Hb level of 6.6 g/dl, low TLC of 3.42 and platelets of 90 cells/µl. On next day patient condition became more worsened as Hb levels and platelets levels were low. To manage this condition fresh frozen plasma (FFP) was transfused. Liver profile showed increased serum bilirubin levels and low total protein and lower albumin levels and kidney profile revealed low serum creatinine levels (0.48 mg/dl) and low ferritin levels supporting with pancytopenia and hepatopulmonary syndrome. These incidental lab findings support the fact that patient has hepatopulmonary syndrome associated with cardiac problems.

The unexplained lab abnormalities prompt an ultrasonography (USG) of whole abdomen and portal venous doppler was performed on day 2 and 3 of admission revealed portal vein being replaced by multiple collateral venous channels in peripancreatic, splenorenal and hepato-gastric and moderate splenomegaly from these findings final note made of portal cavernoma likely to be extrahepatic portal vein obstruction (EHPVO). ECG findings on day of admission revealed left ventricular hypertrophy (LVH), 2D-ECHO findings on first day revealed decreased LV ejection fraction of 25-30% with severe mitral regurgitation (MR).

To further characterize the vascular anomaly seen on USG, a contrast enhanced computed tomography (CECT) abdomen is performed. This reveals the underlying anatomy of severe portal venous stenosis with large splenorenal shunt and changes of portal hypertension leading to the diagnosis of Type 1b Abernethy Malformation. The cardiac findings of LVH and MR can then be contextualised as potential long-term complications of the portosystemic shunt due to hyperdynamic circulation or associated congenital defects.

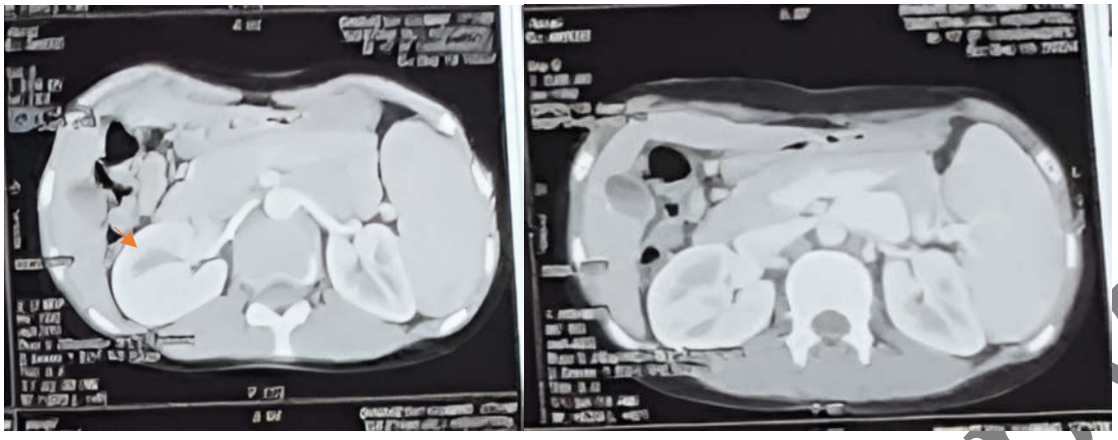

Fig. 1: Axial contrast-enhanced CT image in portal venous phase. A large splenorenal shunt (arrow) is seen. Additional findings include moderate splenomegaly and liver morphological changes with and enlarged left lobe suggestive of chronic liver disease and portal hypertension

The patient was treated symptomatically, prescribed with multivitamin injection for pancytopenia, ramipril 5 mg once a day for hypertension, spironolactone and torsemide for LVH and oedema due to heart failure and digoxin 0.25 mg once a day for tachycardia and heart failure due to low ejection fraction for further management ceftriaxone 1000 mg two times a day for treating pulmonary and preventing hospital acquired infection. No modification in therapy were done after few days patients vitals were stable and ejection fraction was 50% ejection fraction was managed properly by digoxin and other medication but no significant improvement was observed in patients type 1b Abernethy malformation condition. Patient was discharged after 10 d told to follow up after 5 d for further management.

DISCUSSION

Abernethy malformation also known as a congenital extra hepatic portosystemic shunt is uncommon anatomical vascular anomaly. Individuals with this condition may present with symptoms of abdominal pain, abnormal CBC profile, abnormal liver function tests, hepatopulmonary syndrome, pulmonary hypertension or portosystemic encephalopathy precise identification of the shunt and portal venous system and effective management of complication is vital in these patients it can majorly affect adolescents and children similar to the study by Claesen E *et al.* [5] and Agarwal A *et al.* [6]. Abernethy malformation is of two types type 1 and type 2 characterised by type of shunting in patient respectively type 1a characterised by lack of portal vein and presence of abnormal systemic circulation type 1b characterised by multiple collaterals connections between the blood vessels type 2 has portal vein but venous flow is redirected to vena cava directly with aberrant connection similar to the studies by Baiges A *et al.* and Claesen E *et al.* [3, 5] routine examination are important and useful for the diagnosis of the condition CT and MRI scan should do for proper clear reveal of the disease early diagnosis is necessary if left untreated it may cause severe liver injury of hepatocellular carcinoma associated pulmonary fibrosis.

Abernethy malformation can be associated with cardiac anomalies or congenital heart disease, left ventricular hypertrophy (LVH) literature suggest that it mainly associated with septal defects and tetralogy of Fallot according to the study by May Y *et al.* [12]. However, in this study the cardiac anomalies were managed by symptomatic treatment which showed good improvement in this case its observed patient has LVH with decreased ejection fraction (25-30%) with severe mitral regurgitation (MR). The portosystemic shunt can lead to reduced hepatic clearance of vasoactive substances, potentially contributing to hypoxia and in some cases cyanosis and clubbing in this case similar to the study by Agarwal R *et al.* [10] the treatment modalities for the disease depends upon the patients condition in severe cases transcatheter closure of the congenital portosystemic shunt is option is chosen to improve the condition according to the study by Lin Y *et al.* [11]. However, in this case patient is being managed with symptomatic treatment because severity of disease condition, patients age and the need for involvement of multidisciplinary planning of hepatology, interventional radiology, surgery etc. hence, symptomatic treatment was chosen to manage patient symptoms and taking follow up.

CONCLUSION

This case highlights case of congenital portosystemic shunt like Abernethy malformation in young patient with cardiac complications, pancytopenia and abnormal liver profile screening by CECT is crucial for diagnosis. Management is complex and requires multidisciplinary approach with treatment strategies ranging from conservative medical management to interventional shunt closure depending upon clinical presentation. In this case patient was managed with symptomatic treatment only. No surgical interventions were used as patient condition was severe and multidisciplinary approach was needed. We can take a message home is that Abernethy malformation should be diagnosed at early stages to prevent complications like hepatocellular carcinoma or pulmonary fibrosis.

AI USE STATEMENT

No artificial intelligence (AI) or AI – assisted tools were used in the writing, data analysis or creation of fig. for this manuscript

AUTHORS CONTRIBUTIONS

Dr. AS is responsible for case conceptualization and case understanding, Dr. AS responsible for literature review and initial manuscript preparation, Dr. MSR were responsible for reviewing the manuscript and Dr. MSR were responsible for final review and approval of manuscript.

CONFLICT OF INTERESTS

Authors declare no conflict of interest between them

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