Abernethy Malformation : A Rare Cause of Hypoxemia

Archana Deshpande[1], Sharath Adiga M[2], Ameya Deshpande[3]

ABSTRACT

Abernathy malformation is a rare congenital disorder which may present like a congenital heart disease. Though rare, suspicion of this condition must be kept in mind in a patient who presents similar to a congenital heart disease but with a normal 2D echocardiogram. Simple routine procedures like CT angiography of abdomen and contrast 2D echocardiography can help in arriving at the diagnosis.

Introduction :

Abernethy malformation, also known as Congenital extrahepatic portosystemic shunt (CEPS) is a condition in which portal blood is shunted partially or completely into the systemic circulation via an abnormal communication of the portal system with the systemic circulation[1]. It was diagnosed by John Abernethy in 1793 at the autopsy of a 10 month old baby[2]. Until now, more than 300 cases have been reported with a literature review, and most patients were female and less than 18 years old. The clinical manifestations of Abernethy malformation are highly variable and can be divided into 3 types : a) asymptomatic, b) symptoms due to the abnormal liver development such as hepatic encephalopathy or multiple liver nodules / tumors, and c) shunt related symptoms such as pulmonary hypertension or hepatopulmonary syndrome (HPS). HPS is characterized by the presence of dyspnea and hypoxia in patients with liver diseases.[3]

2 subtypes are defined :

- Type 1 - End to end shunt with congenital absence of intrahepatic portal vein
- Type 2 - Side to side shunt

  Type 1 is further subdivided into 2 types

  - ✍ Type 1a - SMV and splenic vein directly drain into IVC
  - ✍ Type 1b - SMV and splenic vein join to form a short course of portal vein before draining to IVC[1]
- Blood from spleen and intestine drains into the inferior vena cava through a shunt bypassing the liver, thereby causing an alteration in the metabolism of pulmonary vasoactive substances. It leads to pulmonary vasodilation, diffusion-perfusion defects and eventually arterial hypoxemia. This syndrome is considered as one of the rare cause of the hepato-pulmonary syndrome[3]

[1]Associate Professor, [2]Senior Resident, [3]Junior Resident,
Department of Medicine, Government Medical College, Nagpur

Address for Correspondence -
Dr. Archana Deshpande
E-mail : arcsandeshpande@rediffmail.com

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Case Report :

16 year old girl presented with increased yellowish discolouration of eyes since 15 days. No history of fever, vomiting, altered sensorium or bleeding tendency. Past history of brain abscess 5 years back which was drained and treated completely. On examination the patient was thin built with a height of 150 cms and weight 30 kgs giving a BMI 13.33 kg/m2. Pulse was 90 per minute and BP 100/60 mmHg. Respiratory rate- 18/min, no dyspnea and no flaps were present. Cyanosis over tongue and nails was present. JVP was not raised and no edema feet or puffy face. Deep icterus present. Grade 3 clubbing in bilateral upper limb and lower limb present. Patients oxygen saturation levels were 75% in supine position and 68% in sitting position. Systemic examination was within normal limits.

Investigation :

- CBC-TLC-5700, Hb-17.6gm%, Platelet-6.69 lakh/cumm
- LFT-total protein 6.2 gm, total bilirubin-12.3 gm (direct-5.5; indirect-6.8) ALP-285, SGOT-111, SGPT-40

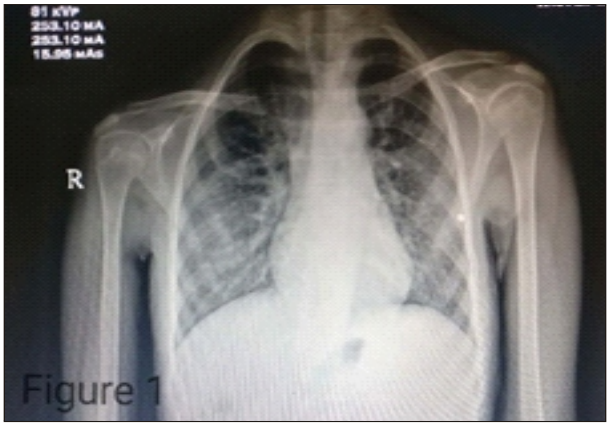


Figure 1 : **Chest X-Ray of the patient**

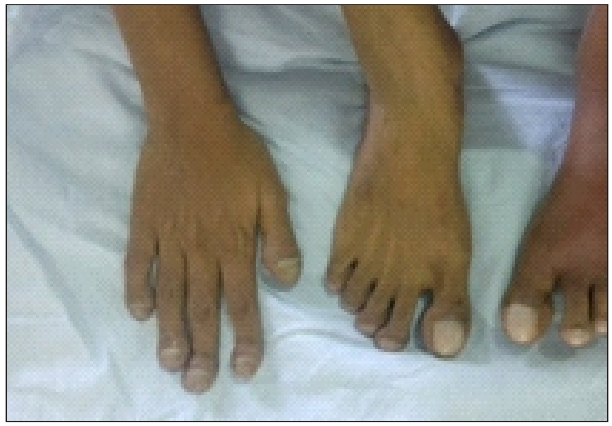


Figure 2 : **Grade 3 clubbing of upper limb and lower limb**

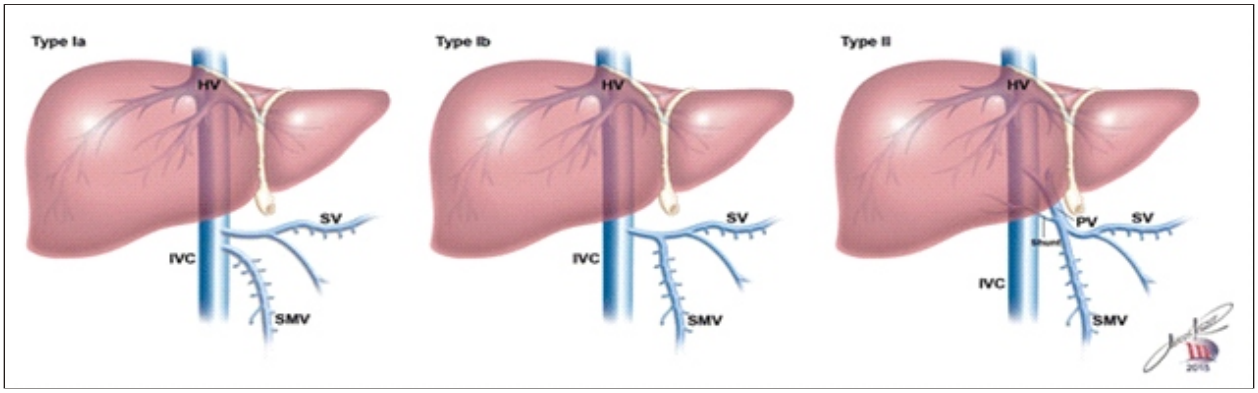


Figure 3 : **Diagram showing types of Abernethy Malformation**

- KFT-blood urea - 34 mg/dl, serum creatinine-1.1 mg/dl
- Serum sodium-135 meq/lit, serum potassium-3.6 meq/lit
- ABG-pO2-35, pCO2-25, pH-7.45, HCO3-18.8
- Based on history the patient was suspected to have congenital cyanotic heart disease and was evaluated further.
- ECG - No P wave abnormality or signs of ventricular hypertrophy.
- Chest X-ray- Within normal limits.
- CT-PA - No evidence of Pulmonary A-V malformation
- CT Abdomen - Portal vein directly opening into inferior vena cava with non-visualisation of intrahepatic portion of portal vein. This condition is known as abernethy malformation type 1b.

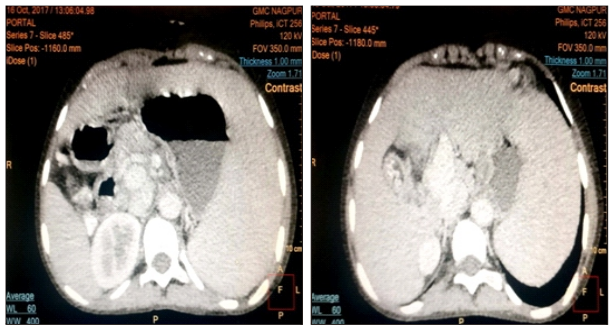
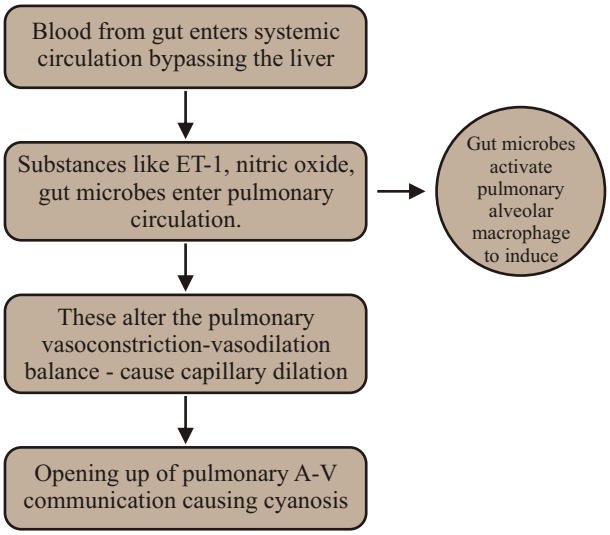


Figure 4 : **Cross section showing IVC and PV just before and after they join**

- Contrast 2-D echo - Showed air bubbles in left atria after 4-5 heart beats signifying pulmonary A-V communication.

Discussion :[2-4]



Abernethy malformation can be associated with : [1,5]

- GI abnormalities like - polysplenia, biliary and duodenal atresia, choledochal cyst, suprahepatic gall bladder
- CVS abnormalities like : ASD, patent foramen ovale, VSD, PDA, TOF and dextrocardia.
- Abnormalities of genito-urinary system.

- Early recognition of porto-systemic shunt is important as it increases the risk of hepatic neoplasm like benign focal nodular hyperplasia, hepatocellular adenoma and degenerative nodules.[5]
- Treatment options :
  - In type 1 : limited to liver transplantation
  - In type 2 : shunt can be occluded either surgically or by per-cutaneous trans-catheter coil placement[3]

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