Chronic Hypoxemia in a Child: Thinking Outside the Box

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Background: Chronic hypoxemia is generally attributed to primary cardiac or pulmonary entities. **Case characteristics**: A 9-year-old boy presenting with cyanosis, clubbing and hypoxemia, without icterus or hepatosplenomegaly. Cardiovascular and respiratory system examinations were normal. **Outcome**: He was diagnosed as type IB Abernethy malformation, a rare cause of hepatopulmonary syndrome. **Message**: Pediatricians should consider hepatopulmonary syndrome in the differential diagnosis of chronic hypoxemia, even in the absence of jaundice or hepatosplenomegaly.

Keywords: *Abernethy malformation, Central cyanosis, Clubbing, Hepatopulmonary syndrome.*

Central cyanosis and clubbing is a child is mostly due to congenital cyanotic heart disease or chronic pulmonary disease. When cardiac and respiratory system examination is normal, it presents a diagnostic challenge. We report an unusual cause of chronic hypoxemia in a child that required extensive work-up before a diagnosis could be made.

CASE REPORT

A 9-year-old boy, resident of an orphanage, was brought with history suggestive of upper respiratory infection. There was no history of chronic cough, prolonged fever, breathlessness, palpitations, chest pain, edema, jaundice, hematemesisor syncope. There was no history of respiratory distress, cyanosis or hospitalization in the past. There was no history of any drug intake or bleeding manifestations. He was developmentally normal.

On examination, he was afebrile with a respiratory rate of 18/min and pulse rate of 96/min. He had marked central cyanosis and pan-digital clubbing. There was no icterus, lymphadenopathy, pallor or edema. He weighed 19.3 kg (3rd centile for age) with height 123 cm (between 3rd and 15th centile). Respiratory and cardiovascular system examination was normal. There was no hepatomegaly or splenomegaly. The oxygen saturation (SpO_2) was 84% in the upper limbs that improved to 94% with oxygen. The chest *X*-ray showed plethoric lung fields; electrocardiogram and echocardiogram were normal.

Due to the absence of an intracardiac shunt or pulmonary hypertension to explain the cyanosis, we investigated him for an extracardiac shunt by injecting agitated saline intravenously. The microbubbles appeared on the left side of the heart after the fourth cardiac cycle, demonstrating an extracardiac shunt. Pulmonary computed tomography (CT) angiography was performed which did not reveal any extracardiac pulmonary shunt or pulmonary arteriovenous malformation. There was also no evidence of chronic lung disease or bronchiectasis on the CT scan. The partial pressure of oxygen (PaO_2) was 41.1 mm Hg which improved to 46.5 mm Hg with oxygen. The change in SpO_2 with position was not significant. The arterial alveolar oxygen gradient ($AaDO_2$) was elevated (88.2 mm Hg).

The liver function tests and prothrombin time were normal. The packed cell volume was 45% (Hemoglobin 14 g/dL). Ultrasonography of abdomen (including Doppler) showed a normal sized liver with no focal lesions, nodularity or biliary dilatation. Right and left branches of portal vein were atretic and replaced by an echogenic strand of tissue. The main portal vein was patent and shown to drain into the inferior vena cava (IVC). CT portovenography done to delineate the vessels showed the formation of main portal vein (MPV) by confluence of the superior mesenteric vein (SMV) and the splenic vein (SV) which had an end to side communication with the IVC without branching into the liver. ***Fig.* 1** shows the abnormal communication between the portal vein and inferior vena cava.

Based on these observations, the child was diagnosed to have hepatopulmonary syndrome (HPS) due to Abernethy malformation (type 1B). The caregivers of the child did not consent for liver biopsy, and opted to get him discharged without further treatment.

DISCUSSION

Abernethy malformation (type IB) with HPS is a rare cause of hypoxemia which could pose a diagnostic challenge.

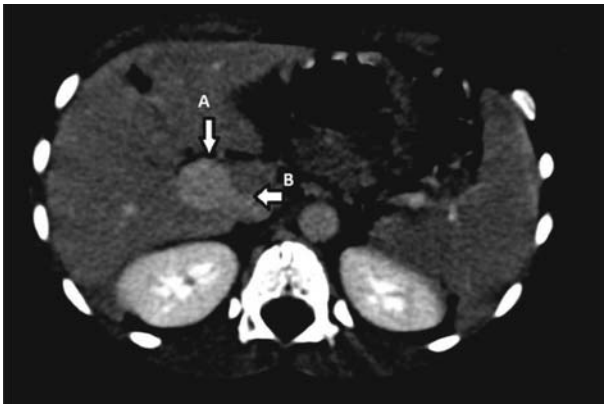

FIG. 1 *Axial section of contrast enhanced CT study showing abnormal communication between the portal vein (arrow A) and inferior vena cava (arrow B).*

The most common causes of chronic central cyanosis and clubbing in children were ruled out in our patient by appropriate investigations. Pulmonary arteriovenous malformations (PAVM) and abnormal hemoglobin are uncommon causes of cyanosis. PAVM in the pediatric age group may be a result of hereditary hemorrhagic telangiectasia or HPS where it is referred to as intrapulmonary vascular dilatation [1].

HPS in children is commonly a result of cirrhosis [2]. Extrahepatic portal venous obstruction (EHPVO), which is a non-cirrhotic disease, is a very rare cause of HPS [3].The development of HPS in EHPVO is a classic example of how redirection of blood flow away from the liver by collateral flow leads to absence of hepatic handling of unknown circulating factors in portal blood and thereby HPS. The case under discussion is an extension of the same concept where the uncommon Abernethy malformation leads to HPS by virtue of shunting blood away from the liver [4-6].

Abernethy malformation is a rare cause of hepatopulmonary syndrome [7], characterized by congenital extrahepatic portosystemic shunts (CEPS) [8]. They have been classified as type 1 when the intrahepatic portal branches are absent and the portal blood completely empties into the systemic vessels, either directly through the SMV and SV (type 1A) or after forming the portal vein (type 1B) as in our case [9]. In type 1, SpO_2 improves with inhaled oxygen as in our case. In type 2 CEPS, the intrahepatic portal branches are patent and the portosystemic communication is a side- to- side communication between the portal vein and the systemic circuit [9]. With the easy availability of ultrasound, the diagnosis of CEPS is often incidental. CEPS may also manifest with growth restriction, focal nodular hyperplasia, malignant transformation of the hepatic nodules, hepatic encephalopathy, HPS or brain abscess. The management of CEPS is surgical. Liver transplantation (including auxiliary partial orthotopic liver transplantation) may be the only option in some cases [10].

To conclude, this case demonstrates the serial analysis and work-up of an unusual cause of central cyanosis and clubbing due to type IB Abernethy malformation with HPS. HPS should be considered in the differential diagnosis of central clubbing and cyanosis of non-cardiopulmonary origin.

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