Hemangioma – A pointer to Abernethy syndrome?

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ABSTRACT

Diffuse pulmonary arteriovenous malformations or pulmonary arterial hypertension (PAH) may result from congenital portosystemic venous shunts. Hemangioma as a physical sign of congenital portosystemic shunts (like Abernethy syndrome) has not been described. I report two children (one with severe cyanosis from pulmonary arteriovenous malformations and the other with severe PAH) with cutaneous hemangioma and Abernethy syndrome. Hemangioma may be a clinical pointer to portosystemic shunts even in the absence of obvious liver disease.

Keywords: Abernethy syndrome, hemangioma, pulmonary arterial hypertension

CASE 1

A 9-year-old girl presented with worsening cyanosis and breathlessness for the last 3 years. On examination, she was severely cyanosed and clubbed [Figure 1a]. The vitals were normal. The clinical examination was otherwise unremarkable with normal second heart sound. There was a hemangioma on the left ear [Figure 1b]. There was no family history of similar skin lesions. The chest X-ray and electrocardiogram (ECG) were within normal limits. Her hemoglobin was 16 gm%, and oximetry showed 78% saturation. The liver and kidney function tests were normal. On echocardiography, structurally normal heart was seen. However, a contrast echo showed opacification of the left atrium after 3–4 beats and was diagnostic of pulmonary arteriovenous fistulae (PAVF). The chest computed tomography (CT) showed no parenchymal abnormality and a CT angiogram showed the presence of portosystemic shunt (extrahepatic portosystemic shunt or Abernethy syndrome). The portal vein was draining into the left renal vein [Figure 2]. The intrahepatic portal radicles were not seen.

CASE 2

An 8-year-old girl evaluated for worsening of breathlessness for the last 1 year. She was a known case of pulmonary hypertension diagnosed from 3 years of age and was receiving sildenafil for the same for the last 1 year. The patient had undergone a thoracotomy with a wrong diagnosis of aortopulmonary window at 3 years of age elsewhere. She had nephrotic syndrome 2 years earlier and a kidney biopsy showed membranous glomerulonephritis. The patient had previously received steroids for nephrotic syndrome. On examination, her vital status was normal, and there was no pedal edema. A loud pulmonic component of second heart sound was heard on auscultation, but there were no other abnormal findings. Similar to the first patient, she also had a hemangioma over the left ear [Figure 3]. There was no family history of similar illness or skin lesions. Her chest X-ray showed mildly dilated pulmonary artery. The ECG was normal. The echocardiogram showed no other congenital heart disease and normal right and left ventricle function. There was mild tricuspid regurgitation with an estimated pulmonary artery pressure of 70 mmHg.

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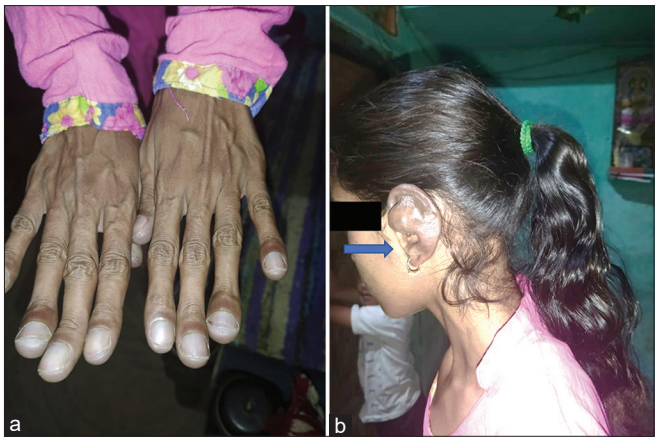

Figure 1: Patient 1 with cyanosis and clubbing (panel a) from diffuse pulmonary arteriovenous fistula and hemangioma (arrow) on the left ear (panel b)

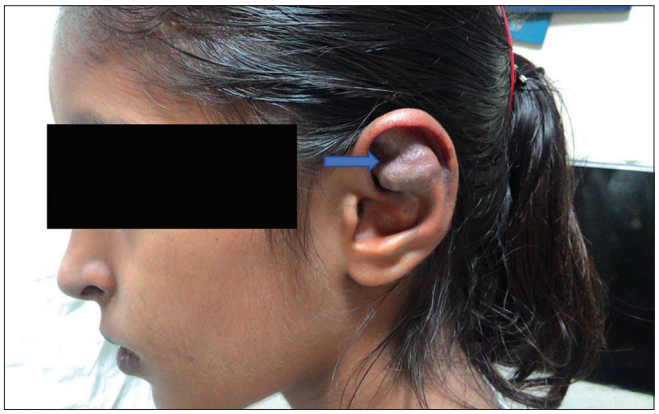

Figure 3: Patient 2-hemangioma on the left ear (arrow)

The CT angiogram showed the portal vein communicating to the inferior vena cava with no intrahepatic portal radicles (Abernethy syndrome) [Figure 4].

DISCUSSION

Portosystemic venous shunt that results in diversion of portal venous blood to the systemic circulation may cause PAVF or pulmonary arterial hypertension (PAH).[1] Portosystemic shunts may be intra- or extrahepatic (Abernethy syndrome) and are being increasingly recognized. The purpose of this communication is to highlight that hemangioma may be a clinical pointer to an underlying portosystemic shunt as seen in both of these cases. Persistent hemangioma in childhood is rare,[2] and fortuitous occurrence of Abernethy malformation with unrelated hemangioma, although possible, seems less likely. Whether such an association represents a distinct syndrome is not known. In fact, such angioma has previously been seen in portosystemic shunt patient including an infant,[3,4] but has not been emphasized. It is possible that more such reports can follow once the condition is brought into the

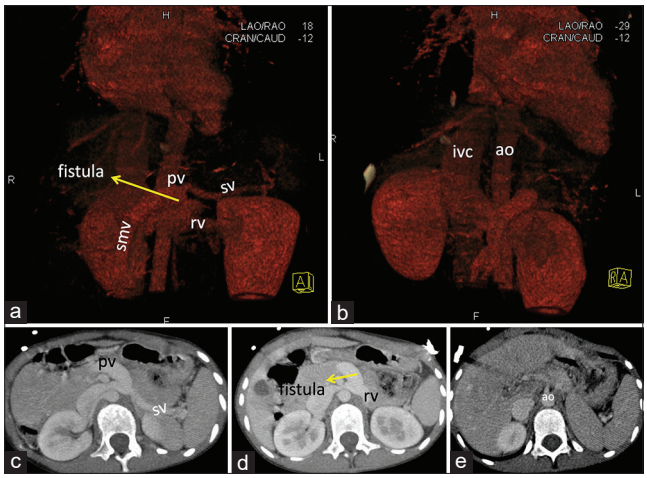

Figure 2: Reconstructed volume rendered technique images (panel a and b) and contrast enhanced computed tomography images in axial section at the level of splenoportal axis (panel c-e) showing fistulous communication between portal vein and left renal vein. No evidence of intrahepatic portal venous radicles seen. SV: Splenic vein, Ao: Aorta, SMV: Superior mesenteric vein, IVC: Inferior vena cava

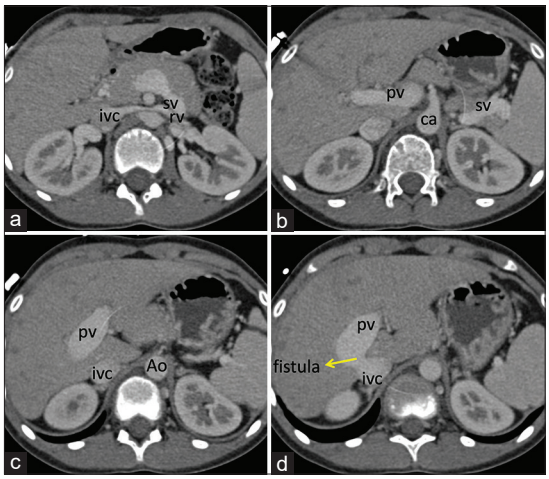

Figure 4: Contrast-enhanced computed tomography images in axial section at the level of splenoportal axis showing end to side fistulous communication (arrow) between portal vein and inferior vena cava. No evidence of intrahepatic portal venous radicles seen. RV: Left renal vein, SV: Splenic vein, CA: Coeliac artery, Ao: Aorta

focus. Angiomatous lesions in patients with liver disease or in hereditary hemorrhagic telangiectasis may occur and might coexist with PAVF or PAH, but their association with Abernethy malformation has not been recognized. Both these patients did not have significant liver disease at the presentation. Whether these angiomas could result from subtle liver dysfunction is speculative; even so, the recognition of the association may be useful. Since hepatopulmonary syndrome or PAH due to portosystemic shunts could be treatable,[5,6] it is obviously important to identify such patients. Children with these shunts may also present with liver disease, encephalopathy, or may be asymptomatic. Sometimes, these malformations exist with congenital heart diseases such as patent ductus

arteriosus, heterotaxy syndromes, and others,[1] and thereby the recognition may be even more elusive. Hence, the presence of cutaneous hemangioma may suggest an underlying portosystemic shunt in appropriate context. The utility of this finding needs more studies.

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Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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