

An Infant with Abernethy Malformation Associated with Heterotaxy and Pulmonary Hypertension

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Abernethy malformation (AM) is a rare congenital anomaly in which the splanchnic blood bypasses the liver and drains directly into the systemic veins. It was first described by Abernethy in 1793 and has two types. Type I is characterized by a complete portosystemic shunt and an absence of a portal vein (consists of superior mesenteric and splenic veins), while Type II exhibits partial portosystemic shunt and a hypoplastic portal vein. Type I is further subclassified into subtype a and b based on the absence or presence of a patent connection between the superior mesenteric vein and the splenic vein.

Pulmonary hypertension (PH) is an extremely rare complication of AM, but it has a substantial impact on survival and requires consistent treatment both before and after surgery.^[1]

An 8-month-old girl was referred to our hospital for recurrent respiratory tract infections and tachypnea that were refractory to treatment. Physical examination was unremarkable except for a tachypnea and slight cyanosis. The O₂ saturation was 88% in room air. Laboratory test showed slightly elevated alanine aminotransferase (ALT) and aspartate transaminase (AST; ALT 86 U/L, AST 187 U/L). Blood cell counting indicated thrombocytopenia. Serologic markers were negative for respiratory viruses, hepatitis B, and C virus. The girl was G¹P¹ and had no significant familial history.

The chest X-ray showed mild cardiomegaly and increased pulmonary vascularity with bulged pulmonary conus. Echocardiography revealed a 10 mm atrial septal defect and a significantly dilated right ventricle with a D-shaped left ventricle [Figure 1a]. The estimated mean pulmonary

arterial pressure, which was calculated using the maximal velocity of regurgitation through the pulmonary valve, was 49 mmHg [Figure 1b]. Chest and abdominal computed tomography (CT) were performed for further exploration. There was no sign of pulmonary arteriovenous malformation. However, abdominal CT angiography demonstrated the absence of a portal vein. The splenic vein and superior mesenteric vein joined and drained directly into the hemiazygos vein without passing through the liver [Figure 1c and 1d]. Flow through the hemiazygos vein ultimately entered into the superior vena cava through the azygos vein [Figure 1e]. Furthermore, the hepatic veins connected directly to the right atrium. The inferior vena cava was absent [Figure 1e]. Blood flow through the bilateral iliac, renal, and inferior mesenteric veins entered into the azygos vein and superior vena cava through the hemiazygos vein. In addition, left-sided polysplenia, abnormal symmetric bronchial branching pattern, and bilateral left atrial appendages were detected. There was no evidence of liver nodules or encephalopathy. A primary diagnosis was made based on these findings as AM Type Ib with severe PH, heterotaxy syndrome of left-sided polysplenia, and an atrial septal defect.

In AM patients, the portal venous blood most commonly drained into the inferior vena cava and occasionally into the

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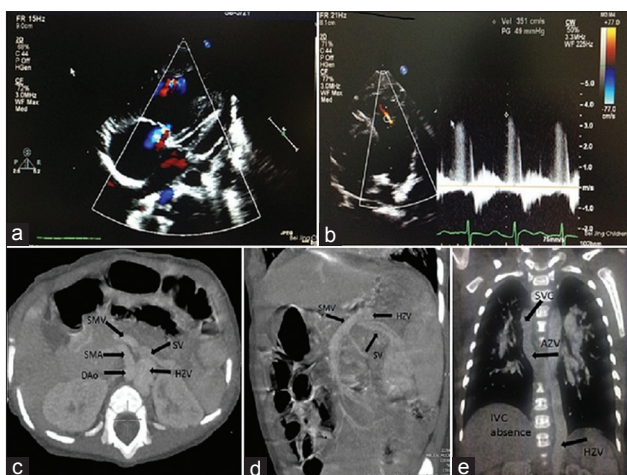


Figure 1: Echocardiography and computed tomography. (a) Echocardiography showed the right ventricle and right atrium dilation; (b) velocity of pulmonary regurgitation was about 3.51 m/s; (c) the dilated hemiazygos vein on the left side of descending aorta. The superior mesenteric vein and splenic vein drains into a confluence, enters the hemiazygos vein; (d) the sagittal plane of the confluence of superior mesenteric vein and splenic vein drains into the hemiazygos vein; (e) hemiazygos vein drains into the superior vena cava via the azygos vein.

renal veins, iliac veins, azygos veins, or right atrium.^[2] In this case, the splenic vein and superior mesenteric vein joined together, drained into the hemiazygos vein, and ultimately entered the superior vena cava through the azygos vein. Furthermore, the patient further experienced complications of left-sided polysplenia and an interrupted inferior vena cava. Until now, there have been limited reports in the literature.

Complications of AM include the presence of liver nodules, hepatopulmonary syndrome, portopulmonary hypertension, and encephalopathy.^[3] Among these complications, portopulmonary hypertension has a substantial impact on survival and requires chronic treatment.^[1]

Histological examination during the autopsy of children with AM showed pulmonary artery pathology, including muscular hypertrophy of large- and medium-sized pulmonary arterial branches, severe stenosis of the distal pulmonary arterioles, microthrombotic occlusion of small arteries, and necrotizing arteritis.^[4,5] These putative mechanisms can result from several factors, including circulating vasoactive mediators, blood volume overload, and hyperdynamic circulation. Among these, the effects of vasoactive mediators are often the most studied by researchers due to their correlation with medical treatment. This is because vasoactive substances, including serotonin (5-hydroxytryptamine), histamine, estrogen, and endotoxin, are produced in the splanchnic

bed and bypass normal hepatic metabolism, thereby allowing them to exert a vasoconstrictive effect on the small pulmonary arteries.

Treatment of AM consists mostly of portosystemic shunt occlusion and liver transplantation and is tailored to the type. Mild PH might be alleviated after the operation. However, patients with moderate-to-severe PH exhibit an unsatisfactory survival rate. It is necessary to provide medical treatment continuously, even before and after the operation, such as endothelin receptor antagonists (bosentan) or phosphodiesterase 5 inhibitors (sildenafil).

In summary, AM is a rare, but important, cause of PH, and must be considered during the diagnostic work-up of PH in children, particularly when it is associated with heterotaxy syndrome. Closure of the portosystemic shunt or liver transplantation may help decrease PH for some patients. Furthermore, anti-PH drugs also play a vital role in producing satisfactory long-term results.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient/guardian consent forms. The patient/guardian understand that their names and initials will not be published in the journal 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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