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Unexplained chronic desaturation in children: should we look beyond the chest?

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Introduction:

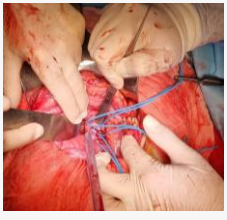
- Hypoxemia is defined as a deficiency of oxygen in the blood, most commonly caused by pulmonary or cardiac disease, or anemia. We report our cases of patients with chronic hypoxemia secondary to congenital extrahepatic portosystemic shunt, also known as Abernethy syndrome.
- This study aimed to investigate congenital extrahepatic portosystemic shunt as an underlying cause of chronic hypoxemia.

Methods:

- We conducted a retrospective study of patients diagnosed with congenital extrahepatic portosystemic shunt, between 2016 and 2025, in our department.

Results:

- All three patients were female, with a mean age of 3 years.
- Clinical presentation was similar across cases, marked by cyanosis of the lips and fingers, digital clubbing, and exercise intolerance. One patient required continuous oxygen therapy.
- The first case involved an Abernethy malformation type Ib, in which the superior mesenteric vein and splenic vein joined to form a short extrahepatic portal vein draining into the inferior vena cava (IVC).
- The second case also showed a type Ib malformation, with a portosystemic shunt between the splenomesenteric trunk and the left renal vein, with no visible portal trunk.
- The third case had a type Ib malformation with a communication between the right superior renal vein and the splenic vein via an extrahepatic shunt.
- The portal trunk and IVC were patent.
- All three patients underwent surgical treatment consisting of ligation of the shunt.



- Postoperative outcomes were uneventful.
- The patient who had been oxygen-dependent was successfully weaned off oxygen therapy one month after surgery.
- Anticoagulant therapy was administered intravenously for one month, followed by oral anticoagulation.

Conclusion:

- AII in children over 5 years is no longer rare, representing 9.6% of 1169 AII cases in this study.
- Contrary to prior reports associating AII with PLP in older children, 92.86% of cases were idiopathic.