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A Case of Abernethy Malformation Type I Combined with Hepatoblastoma

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Dear Editor,

A 2-year-old male was incidentally found to have a hepatic mass during a health examination. Physical examination revealed the absence of the left auricle and no external auditory canal. Bilateral facial asymmetry was observed, with the left side being smaller. Laboratory tests indicated elevated levels of AFP and PIVKA-II, along with abnormal liver function. Echocardiography indicated a patent foramen ovale and CT imaging revealed a mass in the left lobe of the liver with multiple intrahepatic nodules. Intraoperative exploration revealed a tumor-like slightly enlarged portal vein

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trunk, which directly joined the inferior vena cava anterior to the pancreas, with an absence of the intrahepatic portal vein, leading to a diagnosis of Abernethy malformation Type I. Pathological examination supported the diagnosis of a GPC3 (positive) and GS6 (diffusely positive) expressing hepatoblastoma. The patient had no symptoms during the 24-month follow-up and had good quality of life.

DISCUSSIONS:

Abernethy malformation, also called congenital extrahepatic portosystemic shunt, is a rare condition in which the portosystemic blood drains into systemic circulation, bypassing the liver.

Abernethy malformation is classified into two types: type I and type II. Type I refers to the total aplasia of intrahepatic portal venous branches with complete extrahepatic shunting of portovenous blood into systemic veins. Type II Abernethy refers to hypoplastic intrahepatic portal venous branches with partial extrahepatic shunting of portovenous blood into a systemic vein. (1) Single or multiple hepatic nodules are encountered in approximately 40% of the patients with CEPS and consist of FNH, NRH, adenomas, hemangiomas, HCC and hepatoblastoma. They occur more frequently in type 1 than in type 2 CEPS (50% vs 22%, respectively) (2)

This article describes an extremely rare case of Abernethy malformation Type I combined with hepatoblastoma. Furthermore, the medical history was characterized by several other abnormalities including gross facial asymmetry and cardiac defects. Thus, a diagnosis of Goldenhar syndrome in the setting of Abernethy type I was made, which was difficult to diagnose preoperatively. Imaging techniques or venography can typically provide a diagnosis, but in some cases, only a liver biopsy can demonstrate the absence of venous structures within the portal space.(2)

Abernethy syndrome is an underdiagnosed condition whose incidence is increasing because of improvements in liver imaging, and clinical manifestations are variable(3).Therefore, clinicians may consider Abernethy malformation when encountering patients with hepatoblastoma combined with craniofacial and cardiac defects, particularly when imaging studies do not reveal a normal portal vein. This consideration is crucial for selecting an appropriate treatment plan and improving patient survival rates.

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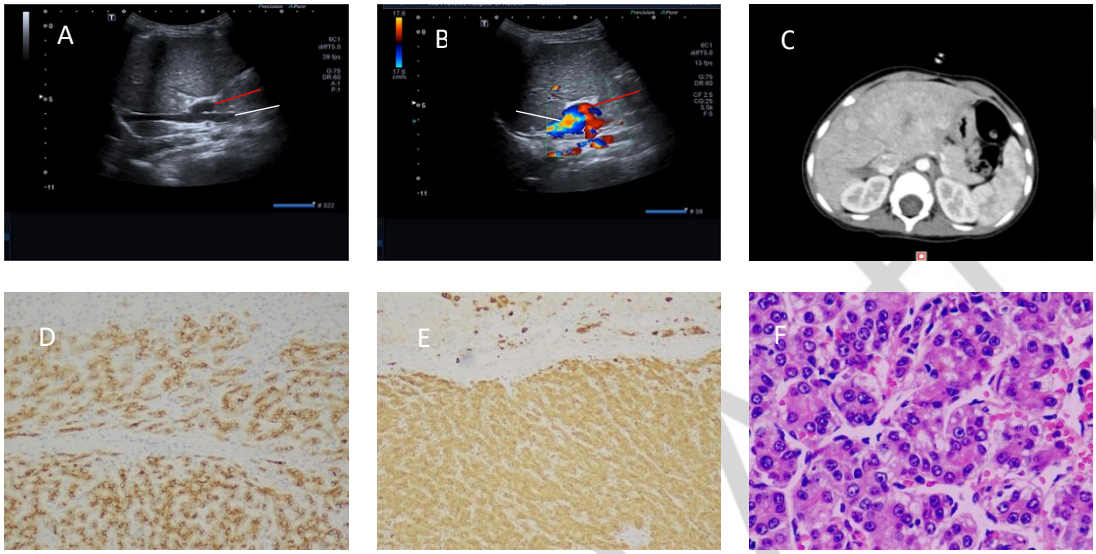

Fig. A 1.2-year-old male with Abernethy Malformation Type I combined with Hepatoblastoma. A. Postoperative abdominal ultrasound showed that the right branch of the portal vein was not visible, and the portal vein lumen showed localized tumor-like dilation（red arrow）, communicating with the inferior vena cava（white arrow） B. Postoperative abdominal color ultrasound C.CT-enhanced arterial phase D. Microscopic examination revealed the tumor to be positive for GPC3 (HE, X10) E. Microscopic examination revealed the tumor to be diffusely positive for GS6 (HE, X10).F. Tumor fragment (immunohistochemistry, X40).

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