GUESS THE DIAGNOSIS

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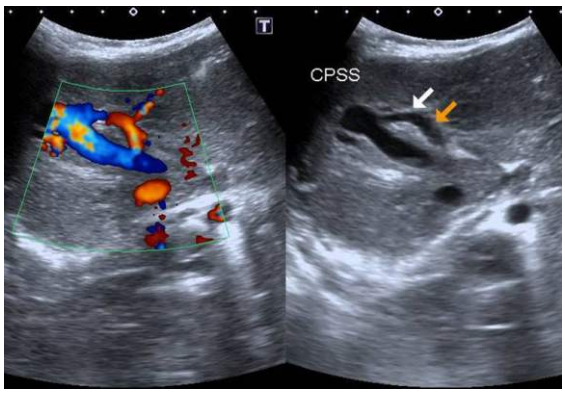
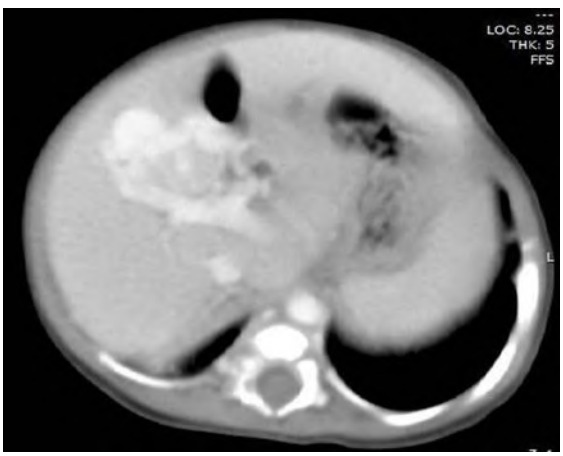
65 days old male infant, born full term small-for-gestational age (birth weight = 2.1 kg) of a non-consanguineous marriage, presented with conjugated hyperbilirubinemia since Day 15 of life. He had history of symptomatic hypoglycaemia within first hour of life which lasted for 2 days. His development was appropriate for age. Examination revealed a 3 cm soft palpable liver and grade III ejection systolic murmur at pulmonary area. Stools were well pigmented. Laboratory tests revealed conjugated hyperbilirubinemia (total bilirubin 4.1 mg/dL, direct fraction 2.4 mg/dL) and elevated transaminases (AST/ALT 439/154 IU/L), fasting hypoglycaemia (blood glucose <40 mg/dL after 4 hours fast) with positive urine non-glucose reducing substances. Ammonia level was 217 microgm/dL and alpha-fetoprotein level was 192,000 ng/mL. Galactose-1-phosphatase uridyl transferase (GALT) assay was normal. Echocardiography showed ostium secundum atrial septal defect (5 mm). His ultrasonography of abdomen (Picture 1) showed something characteristic for which he was taken up for computerized tomography (Picture 2). What is his diagnosis and how can we manage him?

Pictures 1 (A and B) and 2





Answer: **Congenital portosystemic shunt (CPSS).**

CPSS is a rare vascular anomaly of the liver with an incidence of 1:30,000 live births. This defect happens due to abnormal development of the liver vasculature. It has been postulated that incomplete involution of vitelline venous system during the development of hepatic sinusoids results in shunt formation. Around one-sixth of CPSS patients may have multiple anomalies, most commonly involving cardiovascular systems including atrial or ventricular septal defects, coarctation of aorta, tetrology of Fallot, patent ductus arteriosus, splenic artery aneurysm, coronary artery fistula or cutaneous hemangiomas. Other abnormalities seen are polysplenia syndrome, anomalies involving renal and biliary system, and genetic syndromes (trisomy 21, Leopard, Osler-Weber-Rendu). Patients with CPSS show wide spectrum of symptoms from incidental detection (22%) on colour-doppler to severe complications. Altered fetal hepatic perfusion leads to intrauterine growth retardation, neonatal cholestasis (9%), hypoglycemia and hypergalactosemia. Later presentations include unexplained neurocognitive dysfunction, behavioural issues and minimal hepatic encephalopathy due to elevated ammonia. One-fourth of these individuals develop hepatopulmonary syndrome or portopulmonary hypertension secondary to excessive vasoactive mediators bypassing the liver. Moreover due to abnormal vascular supply, tumours develop in 24% of such livers – focal nodular hyperplasia, nodular regenerative hyperplasia, adenomas and hepatocellular carcinoma [1].

Diagnosis is based on colour-Doppler which demonstrates abnormal communication between portal (PV) and hepatic veins (HV) or persistent ductus venosus. Intrahepatic PV branches may be non-visible or hypoplastic showing hyperechoic bands surrounded by hypoechoic stripes. PV flow may be slow or minimal. CPSS is classified as extrahepatic with complete (type I) or partial (type II) absence of intrahepatic PV flow; or intrahepatic (type 1 – right PV joins vena cava; type 2 – localized shunt from PV to HV in one lobe; type 3 – aneurysmal communication between PV and HV; type 4 – multiple communications between PV and HV; and type 5 – patent ductus venosus [2]. The infant in Pictures 1 and 2 has type 2 intrahepatic CPSS with dilated middle HV (solid arrows in Pictures 1B and 2) communicating with a branch of PV (arrows, Picture 1B). Main PV and vena cava is shown with black arrows and triangle, respectively (Picture 2). Treatment is based on shunt size and fraction, location, age, symptom severity and presence of tumours. All symptomatic shunts, asymptomatic extrahepatic shunts or asymptomatic large intrahepatic shunts need early intervention. Contrarily, small intrahepatic shunts can be left alone till 1 year of age to allow for spontaneous closure. Interventional closure can be performed either percutaneously using Amplatzer devices or coils, or surgically in one or two stages. Balloon occlusion test is performed prior to surgical closure to check PV pressure and tolerance of bowel. With the advancement in interventional radiology techniques, liver transplantation is rarely required for CPSS [1, 2].

References:

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2. Sokollik C, Bandsma RH, Gana JC, van den Heuvel M, Ling SC. Congenital portosystemic shunt: characterization of a multisystem disease. J Pediatr Gastroenterol Nutr. 2013 Jun;56(6):675-81. Review.