Asymptomatic child with multiple benign liver lesions and absence of portal vein - abernethy malformation

An 11 year old girl was incidentally diagnosed with congenital extrahepatic portosystemic shunt. She has a factor VII deficiency. Surgical repair of a congenital atrial septal defect type II was performed after birth.

Imaging Findings:

We report the case of an 11 year old asymptomatic girl with factor VII deficiency. Surgical repair of a congenital cardiac defect (atrial septal defect type II) was performed after birth. Routine ultrasound two years before presentation at our department revealed only multiple liver lesions. US findings obtained 6, 12 and 24 months later were indeterminate with regard to lesion characterization, number, and progression. Histology demonstrated focal nodular hyperplasia (FNH). Development was normal for age and laboratory tests demonstrated moderately increased transaminase levels, which were constant over time. US follow-up demonstrated further progression of the focal hepatic lesions after 48 months. A MRI of the liver confirmed multiple focal liver lesions (Figure 1). According to the knowledge of the pathological result the liver lesions were classified as atypical FNH. Atypical features of FNH can consist of lesion heterogeneity, hypointensity in the portal venous phase, the absence of central scar in lesion greater than 3cm, or scar hypointensity on T2-weighted images [1]. Dynamic T1-weighted images in this case demonstrated two hitherto unknown portosystemic shunts secondary to absence of the intrahepatic portal vein (Figure 2).

Discussion:

The constellation of symptoms - portosystemic shunt, absence of intrahepatic portal vein, benign liver lesions, and congenital cardiac defect – is consistent with type I Abernethy malformation (AM). Most of 34 reported cases of AMs are diagnosed in juveniles. An association with factor VII deficiency has not been reported before. AM is a rare congenital vascular anomaly [2] characterized by congenital extrahepatic portosystemic shunting. The malformation is based on an embryonal developmental disorder of the portal vein between 4-10 weeks of gestation. The physiological portal vein develops from the right and left vena omphalomesenterica. The anatomical portal vein results from a regression of caudal fraction of right vena omphalomesenterica and cranial fraction of left vena omphalomesenterica. Hypoplastic or aplastic portal vein with an embryonal portosystemic shunt is the direct consequence by an increased regression tendency of both veins. Two types of AM are classified. Type I malformation is characterized by an absent of portal vein and complete end-to-side anastomosis. In type I two subtypes were graduate: type Ia with separate drainage of the superior mesenteric and splenic veins into the inferior vena cava, and type Ib both veins joining to pseudoform of extrahepatic portal vein which drains into the inferior vena cava. The congenital portosystemic side-to-side shunt by a presence of portal vein is presented in type II [3].
No associated anomalies were described in type II malformation. Type I predominantly occurs in females and is associated with other congenital anomalies (e.g. congenital heart disease, biliary atresia, malrotation, duodenal atresia, annular pancreas, polyps, situs inversus, urogenital anomalies, skeletal anomalies) and other congenital syndrome complexes, such as Dubin-Johnson syndrome (hereditary hyperbilirubinemia) and Goldenhar syndrome (oculoauriculo-vertebral dysplasia) (4,5). None of the cases of AM reported in the literature had an associated factor VII deficiency. Benign focal liver lesions (FNH, adenoma, nodular regenerative hyperplasia) occur in both types [6] and may undergo malignant transformation, like hepatocellular carcinoma or hepatoblastoma. Therefore, long-term follow-up and 6 months monitoring are recommended for patients with AM associated benign focal liver lesions [4,7]. The condition may present with developmental disorders secondary to hepatic encephalopathy and portal hypertension [8]. Liver transplant is the only treatment in type I disease, while type II can be managed by surgical or interventional shunt occlusion [9]. This case report shows that contrast enhanced MRI of the liver is actually the best diagnostic method for detection of portosystemic shunt in AM. The child and the parents are opposite a transplantation. Follow-up is necessary to assess the spontaneous development of this malformation.

**Differential Diagnosis List:** Abernethy malformation type I

**Final Diagnosis:** Abernethy malformation type I

**References:**


Description: Portal venous VIBE-sequences (GRE; TR/TE:3.35/1.35; SL:3mm; flip angle: 12°; matrix: 158x256) show absence of the intrahepatic portal vein and two portosystemic shunts (S1/S2) (communication between the venous confluence (C) and the inferior vena cava (VC)). The splenic vein (SV) and mesenteric vein drain directly via the shunt. Origin:
Figure 2

Description: A and B are T2 weighted images (tse; TR/TE: 4754/76; SL: 5mm; flip angle: 150°; matrix 168x384).
The contrast-enhanced images C and D (VIBE; GRE; TR/TE:3.35/1.35; SL:3mm; flip angle: 12°; matrix: 158x256) allow reliable diagnosis of multiple focal liver lesions (*). Histologically, the lesions were multiple FNHs. Origin: