Arteriovenous Fistula and Portal Hypertension in a Child with Down Syndrome

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The normal pressure in the portal vein is low due to the minimal vascular resistance in the hepatic sinusoids. Increased resistance to blood flow can occur at three levels: a) pre-sinusoidal (e.g., portal vein thrombosis), b) sinusoidal (e.g., liver cirrhosis), and c) post-sinusoidal (e.g., Budd-Chiari syndrome). Portal hypertension also may arise from increased blood flow (e.g., massive splenomegaly or arteriovenous fistulas), but this is controlled by the low outflow resistance of the normal liver. Thus, hepatic arteriovenous fistula is a rare cause of portal hypertension and gastrointestinal bleeding.

Hepatic vascular malformation usually occurs secondary to trauma [1], percutaneous interventions, neoplasms, and cirrhosis. It may also be congenital (e.g., Osler-Weber-Rendu syndrome), most likely due to AVF. It may be asymptomatic or present with features of portal hypertension, such as variceal bleeding, ascites, splenomegaly, diarrhea or heart failure.

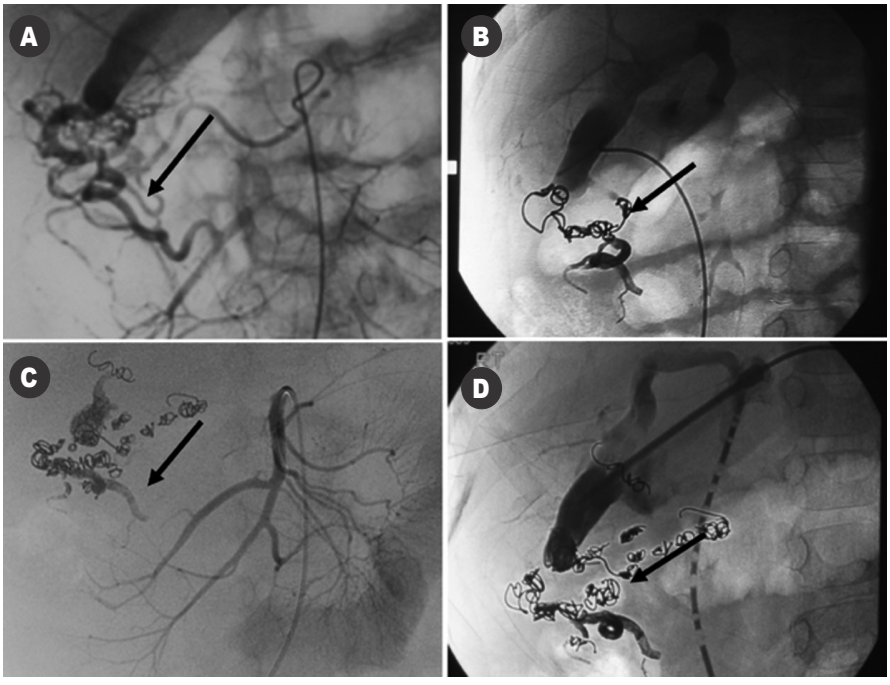
We describe a child with trisomy 21 (Down syndrome) and AVF associated with extrahepatic portal hypertension and gastrointestinal bleeding. Treatment consisted of percutaneous transvascular embolization of the AVF.

Patient Description

A 3 year 9 month old boy with Down syndrome and hypothyroidism was referred to our facility for evaluation of his candidacy for liver transplantation. His medical history was significant for severe portal hypertension and episodes of recurrent gastrointestinal bleeding that required blood transfusions and upper gastrointestinal variceal sclerotherapy.

At physical examination, the patient was found to have Down-characteristic dysmorphic facies, hepatomegaly, significant splenomegaly, and increased abdominal venous markings. No abdominal murmur was heard, and no thrill was palpated. Laboratory studies showed a total bilirubin level of 1.7 mg/dl (mainly indirect), hemoglobin 8.3 g/dl, and platelet count 40 K/μl. Other blood biochemical parameters (including liver function tests) and arterial blood gasses and coagulation profile were within normal range.

Abdominal and Doppler sonography showed splenomegaly and hepatomegaly, with the liver exhibiting a non-homogeous echo-texture, suspicious for cirrhotic liver. There were no focal lesions. A waveform signal of arterial flow was demonstrated near the liver, compatible with pre-hepatic AVF. Liver biopsy exhibited mild fibrosis



Transfemoral arteriography demonstrating the arteriovenous fistula (arrow) between the superior mesenteric artery – through the pancreaticoduodenal vessels – and the portal vein, before **[A]** and after **[B]** the procedures. Transhepatic catheterization demonstrating the portal vein and the arteriovenous fistula (arrow) during the first procedure **[C]** and after the procedures **[D]**.

AVF = arteriovenous fistula

and inflammation and bile duct hyperplasia, but no evidence of cirrhosis.

Angiography performed for further evaluation demonstrated an arteriovenous fistula between the superior mesenteric artery – through the pancreaticoduodenal vessels – and the portal vein [Figure]. Treatment consisted of percutaneous transhepatic embolization of the venous side of the AVF using coils (COOK Group, Inc., Bloomington, IN, USA) and biological glue (MTI Onyx System, Micro Therapeutics, Inc., Irvine, CA). However, the procedure was followed by the emergence of melena, with persistent splenomegaly and thrombocytopenia. Findings on repeated angiography were similar to the preoperative picture. Therefore, during the next 5 months, we performed three further angiographic procedures. These procedures, using a transfemoral coil embolization approach of the arterial side of the AVF and a transhepatic approach with biological glue insertion of the venous side of the AVF [Figure], were performed with the addition of activated factor VII administration, thus enhancing the probability of definite closure of the AVF.

Following these embolizations, the gastrointestinal bleeding subsided and the hemoglobin and platelet levels stabilized. The patient was discharged in a hemodynamically stable condition, and during the subsequent months of follow-up gastroscopy demonstrated no esophageal or gastric varices. There was no gastrointestinal bleeding and no other significant symptoms or signs.

Comment

Hepatic AVF is a rare but treatable cause of portal hypertension and gastrointestinal bleeding. Treatment of an AVF is directed toward its closure [2] or redirecting the blood flow. Closure may be performed by occluding the feeding artery, the draining vein, the AVF itself, or any combination thereof. Surgical options include vascular ligation or resection, and portocaval shunting. However, the significant morbidity and mortality associated with surgical treatment has made endovascular occlusion the mainstay of treatment. It has been found to be effective and safe, and is also applicable for hardly accessible vascular malformations, which was the case in our patient.

The association of AVF with extrahepatic portal hypertension is rare, and these findings are even rarer in trisomy 21. To the best of our knowledge, only one other case of vascular malformation as a cause of extrahepatic portal hypertension and gastrointestinal bleeding has been reported in a child with trisomy 21 [3]. These authors resected the malformation via laparotomy, and during the year after the operation gastrointestinal endoscopy showed no existence of esophageal varices, and the patient was doing well without bleeding. Interestingly, there are two reports of children with trisomy 21 and a portosystemic venous shunt [4,5]. Although portosystemic shunts are a completely different pathogenetic condition, these cases are further examples of the possible association of vascular malformation with trisomy 21.

In summary, we describe a child with trisomy 21 who presented with symptomatic AVF associated with upper gastrointestinal bleeding, splenomegaly and thrombocytopenia. In the only other case of vascular malformation-induced portal hypertension and upper gastrointestinal bleeding in a child with trisomy 21 the malformation was resected via laparotomy [3]. By contrast, we used less invasive angiographic embolizations, with a good outcome.

References

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