Liver Vascular Malformations in HHT & Recommendations for Management and Treatment

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Liver vascular malformations (VMs) in the liver are common in HHT. However, they cause symptoms in only a minority of patients later in adult life. The liver receives blood from two blood vessels, the hepatic artery and the portal vein, and blood exits the liver through the hepatic vein. VMs result in 3 types of abnormal blood flow patterns (short circuits or “shunts”): from hepatic artery to hepatic vein, from portal vein to hepatic vein and from hepatic artery to portal vein. The most common is hepatic artery to hepatic vein shunt.

What are the symptoms of liver VMs?

Liver VMs can cause a wide range of symptoms depending on the type of shunt and the degree of shunting. These symptoms can be grouped into “syndromes”, the three major ones being heart failure, portal hypertension and biliary ischemia.

Heart failure symptoms include fatigue, shortness of breath and swelling of the legs. They occur because blood passes rapidly through the “shunts” in the liver and immediately back to the heart, instead of going through smaller blood vessels where the blood normally slows down. This strains the heart and can lead to a type of heart failure called high output heart failure.

Portal hypertension symptoms include fluid in the abdomen (ascites) and enlarged blood vessels (varices) in the esophagus that can bleed. They occur because pressure in the portal vein increases (portal hypertension) when blood passes through “shunts” from the hepatic artery into the portal vein. Portal hypertension can also occur when the irregular blood flow in the liver causes a condition called “nodular regenerative hyperplasia” in which the liver becomes lumpy, blocking the flow from the portal vein into the liver.

Biliary ischemia causes abdominal pain and, in more severe cases, it may cause jaundice (yellow coloration of the eyes and skin) and fever. Liver blood tests are abnormal and ultrasound or CAT scans show bile duct abnormalities or cysts in the liver. Shunting of blood through liver VMs decreases blood supply to the bile ducts (tubes carrying bile from the liver to the gut) and because they lack oxygen (ischemia), they become damaged.

Abdominal pain can also occur because of blood that is shunted through the liver and “steals” blood away from the intestines (mesenteric ischemia).

Another symptom of liver VMs that usually occurs in association with some of the above syndromes is encephalopathy (confusion). It occurs because toxic substances like ammonia are shunted away from the liver (portal vein to hepatic vein shunt) and go to the brain causing mental changes that are usually reversible.

Can these symptoms be caused by other diseases?

There are many causes for the syndromes and symptoms listed above. Thus, it is important to work with your physician and with an HHT Treatment Center when being evaluated for these problems.

Should all individuals with HHT have their liver checked for VMs?

There is no reason to screen for liver VMs in patients who do not have symptoms because there is no current therapy that will prevent symptoms from occurring. Imaging tests of the liver are usually performed when symptoms that could be due to liver VMs arise. Abdominal ultrasound (including a technique called Doppler) or computed tomography (CT) are the most commonly used tests.

Are there tests that should not be done when liver VMs are present?

Some tests, such as ERCP (endoscopic retrograde cholangio-pancreatography) and liver biopsy, can be dangerous in individuals with liver VMs and should be avoided.

What is standard treatment for symptoms due to liver VMs?

If liver VMs are not causing symptoms, they do not require treatment.

For heart failure, treatment usually involves a low salt diet, diuretics to increase salt and water excretion, iron to correct anemia and specialized heart medications to correct heart rhythm irregularities. A heart catheterization test, using a small tube inserted into a vein and advanced into the heart, is usually performed to monitor therapy.

For portal hypertension, treatment usually involves medications (beta-blockers) to reduce portal pressure or putting rubber bands around varices to prevent bleeding.

For biliary ischemia, treatment involves analgesics for pain, frequent small meals and, in the case of infection, antibiotics. Sometimes when biliary cysts get infected they may need to be drained through a tube placed through the skin.

For encephalopathy, treatment involves the use of medications that will lower ammonia levels in the gut.

What is treatment for patients who do not respond to standard treatment?

Liver transplantation has been successful in patients with liver VMs that do not respond to standard therapy. The best candidates and the timing for liver transplant have not yet been well defined.

Embolization of liver VMs (selective occlusion of blood vessels by introducing artificial emboli) has been successful to control heart failure in some patients, but this procedure is risky and can cause liver failure and death.

Finally, medications that inhibit the formation of blood vessels (anti-angiogenic drugs), which are used for cancer treatment, have been used in a few patients with liver VMs with apparent good results. However, more information is needed regarding their efficacy and safety before they can be recommended.

If you or someone in your family has liver VMs with significant symptoms, we urge immediate consultation with an HHT center before any decision is made regarding invasive diagnostic tests or treatment.

Key points

- Liver VMs are common in HHT but only a few patients develop symptoms related to them
- Screening studies to investigate the presence of liver VMs in patients with HHT are not recommended
- In patients with symptoms suggestive of liver VMs, abdominal ultrasound or CAT scan are used to establish their presence
- Symptoms are mostly related to three syndromes: heart failure, portal hypertension and biliary ischemia
- Initial treatment is directed at the symptoms (or syndromes that lead to them)
- In patients who do not respond favorably to medical therapy, liver transplant should be considered