

LIVER VASCULAR MALFORMATIONS & HHT



COMPANION FACTSHEET TO
MY HHT CARE CHECKLISTS

SIGNS AND SYMPTOMS

CHRONIC LIVER PAIN
ELEVATED LIVER FUNCTION TESTS
(LFTS)
YELLOWING OF THE SKIN AND EYES
SHORTNESS OF BREATH
FATIGUE
LOSS OF APPETITE
DECREASED EXERCISE TOLERANCE
SWOLLEN LEGS AND FEET
CHEST PAIN

FACTSHEET
FS

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SOME IMPORTANT FACTS TO REMEMBER ABOUT HHT ARE:

Liver VMs occur in approximately 75% of HHT patients.

Liver VMs are typically more severe in patients with ACVRL1 mutation (HHT2).

Screening for liver VMs should be offered to adults with definite or suspected HHT.

Liver VMs are most often asymptomatic but can result in heart failure and other complications.

Echocardiogram is recommended at the time of liver VM diagnosis.

Patients with symptomatic liver VMs should be managed by an expert team at an HHT Center of Excellence, with at least annual follow-up.

Liver biopsy should be avoided in patients with definite or suspected VMs.

Hepatic artery embolization should be avoided in patients with liver VMs.

MRI and CT scan findings of Liver VMs are often confused with cirrhosis when viewed by physicians who may not have expertise in HHT.



The Cornerstone of
the HHT Community

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Liver vascular malformations (VMs) are abnormal vascular connections in the liver. They occur in approximately 75% of **HHT (Hereditary Hemorrhagic Telangiectasia)** patients, more commonly in women and often presenting in the 5th decade. The clinical presentation is typically more severe in patients with **ACVRL1 mutation (HHT2)**. Liver VMs in HHT typically present as **diffuse small lesions (telangiectasias)** throughout the liver, and rarely as discrete large **arteriovenous malformations (AVMs)**. Clinicians should offer screening for liver VMs and be aware of possible symptoms or complications and prognostic factors. First-line management depends on symptoms.

HERE ARE SOME THINGS TO DISCUSS WITH YOUR PHYSICIAN:

About diagnostic imaging for liver VMs.

If you have been previously diagnosed with and/or treated for liver VMs.

If you have any of the listed signs and symptoms.

Getting an expert opinion at a HHT Center of Excellence.

Getting diagnostic imaging for liver VMs, using specialized Doppler ultrasound, CT or MRI with special contrast (dye) protocols.

Getting an echocardiogram to look for cardiac effects of liver VMs.

HOW IT IS DIAGNOSED

- > Clinical history, physical examination and bloodwork (LFTs).
- > **Doppler ultrasound:** The imaging of choice for screening. This test uses sound waves to produce a picture of the organs in the abdomen. No radiation is used during this study.
- > **CT (computed tomography) scan:** May also be performed to evaluate for liver VMs. This a high-resolution X-ray of the abdomen. An IV will need to be started for contrast (X-ray dye) to be given.
- > **Magnetic resonance imaging (MRI):** May also be performed to evaluate for liver VMs. This test utilizes strong magnetic fields to form images of the body. No radiation is used during this study. An IV will need to be started for contrast (dye) to be given. The scanner resembles a large tube and the patient is required to lie still during the actual MRI scanning. If the patient has claustrophobia, the doctor may prescribe an oral medication to take prior to the MRI.
- > **Echocardiogram:** Used to assess for cardiac effects of liver VMs. This test uses sound waves (ultrasound) to determine how the heart muscle and valves are working. No radiation is used during this study. It is recommended at the time of liver VM diagnosis.
- > The type of imaging should be performed based on the risk/benefit balance, local expertise, and availability/cost
- > These tests will be most informative when performed in a center with HHT expertise, in the context of a clinical assessment at an HHT Center of Excellence.

TREATMENT

- > No treatment recommended for asymptomatic liver VMs.
- > Treatment is reserved only for patients with complications and/or symptomatic liver VMs.
- > Patients with heart failure and pulmonary hypertension should be co-managed by an HHT Center of Excellence and an HHT cardiologist or a pulmonary hypertension specialty clinic.
- > **Bevacizumab:** An IV medication used to treat a number of types of cancers. Although not a chemotherapy drug, it slows the growth of blood vessels and has been shown to help patients with severe liver VMs and heart failure who have failed other types of medical management.
- > **Liver transplant:** Considered for patients with symptomatic liver VMs, specifically those with refractory heart failure, biliary ischemia, or complicated portal hypertension.

AFFILIATED ISSUES

- > Heart failure
- > Pulmonary hypertension
- > Biliary ischemia
- > Cirrhosis
- > Portal hypertension



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