

CHILDREN & HHT



COMPANION FACTSHEET TO
MY HHT CARE CHECKLISTS

SOME IMPORTANT FACTS TO REMEMBER ABOUT HHT ARE:

Life-threatening complications of lung AVMs and brain VMs can occur at any age.

The clinical diagnostic criteria (Curacao Criteria) for HHT are less reliable for young children as many symptoms do not present until late childhood or adulthood.

Screening should be performed at the time of diagnosis/presentation.

Screening can be performed in newborns.

Genetic testing can identify asymptomatic HHT or rule-out HHT in children of HHT families with a known mutation.



The Cornerstone of
the HHT Community

FACTSHEET
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HHT (Hereditary Hemorrhagic Telangiectasia)

is autosomal dominant genetic disease (it does not skip a generation), and each **child** born has a 50% chance of inheriting the genetic change. While some manifestations of HHT, such as **telangiectasia** and **epistaxis (nosebleeds)**, are age dependent and may be absent in young children with HHT, potentially serious and even life-threatening complications of **pulmonary (lung) arteriovenous malformations (AVMs)** and **brain vascular malformations (VMs)** can occur at any age. The pediatric HHT guidelines focus therefore on screening and management of pulmonary AVMs and brain VMs in children.

HERE ARE SOME THINGS TO DISCUSS WITH YOUR CHILD'S PHYSICIAN:

Genetic testing for your child.

Screening for pulmonary AVMs for your child.

Screening for brain VMs for your child.

If your child has pulmonary AVMs or brain VMs.

If your child has nosebleeds.

Getting screening and/or treatment at an HHT Center of Excellence.

Talk to your child's dentist and other health care professionals about the need for pulmonary AVM precautions, lifelong.

RECOMMENDATIONS

- > Genetic testing should be offered for **asymptomatic children** of a parent with HHT. The affected family member should be tested first to determine the mutation, prior to testing an asymptomatic child.
- > Screening for **brain VMs** should be performed in **asymptomatic children** with HHT or at risk for HHT at the time of diagnosis/presentation.
- > Children with HHT who have **brain VMs** should be referred to a center with multidisciplinary expertise in neurovascular disease management.
- > Screening for **pulmonary AVMs** should be performed in **asymptomatic children** with HHT or at risk for HHT at the time of diagnosis/presentation and should be repeated every 5 years if negative.

TREATMENT

- > Large **pulmonary AVMs** and those associated with reduced oxygen saturation should be treated in children to avoid complications.
- > **Brain VMs** with high-risk features should be treated in children and treated brain VMs require close follow-up.

SCREENING STUDIES

- > Screening for **pulmonary AVMs** in children may be performed with either chest X-ray and pulse oximetry OR contrast echocardiography.
- > **Magnetic Resonance Imaging (MRI):** The recommended test for identifying **brain VMs**. This 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. This typically requires **sedation** or anesthesia in young children.
- > **Contrast echocardiography (echo bubble study):** The recommended study for initial screening of pulmonary AVMs in adults and in some cases children. This test uses sound waves (ultrasound) to determine if injected saline bubbles can get through the lung circulation and be seen back in the heart, on the left side. This is called a **shunt**. An **IV** will need to be started for saline bubbles to be given. No radiation is used during this study.
- > **CT (computed tomography) scan:** If the echo bubble study is positive, diagnosis should be confirmed with a CT scan. This is a high-resolution X-ray of the lungs. If contrast (X-ray dye) is used, an **IV** will need to be started.



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