

MY CHILD HAS HHT: SCREENING GUIDELINES



HHT is a genetic disease, meaning it runs in families and each child born has a 50% chance of inheriting the genetic change. Having helped our own children to manage HHT, we understand the conflicting emotions that can accompany a child's diagnosis. There may be fear, anger, sadness, and even guilt, but it is important to remember that the best thing you can do for your child is to have him or her screened and properly treated.

FACTSHEET **FS**

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WHY DOES MY CHILD NEED A SCREENING?

For children, genetic testing is the only way to know if the gene has been inherited. For those children that test negative, then you will be assured that they do not have HHT and neither will their children and children's children. If a child has inherited the gene, then the next step is to screen the child, as it is vital to understand if internal organs are involved.

The results from this screening allow for a treatment and management; thereby, preventing unanticipated events.

Cure HHT offers multiple helpful resources, including Centers of Excellence all over the country, and we encourage you to contact us - we will always help in any way we can.



The Cornerstone of
the HHT Community

HHT Foundation International Inc.
P.O. Box 329 / Monkton / MD 21111



If someone in your family has tested positive for a gene mutation with HHT, all children should be tested against the identified gene. Even if your child does not have nosebleeds or the red spots on the skin known as skin telangiectasia, they could still be at risk with HHT impacting major organs such as the brain and lungs.

** If you plan on not having a screening through an HHT Center of Excellence, all children with a brain MRI showing an AVM or an abnormal echo bubble test should have a consult with an HHT Center's medical professional. **

What Should I Expect When Scheduling A Screening?

An HHT Screening should be performed at an HHT Center of Excellence. For a complete list and contact information, please visit the HHT Website.

Note: If you decide NOT to visit an HHT Center of Excellence but still would like to be screened, please have your pediatrician contact the **Cure HHT office at 410-357-9932 to receive all the necessary information for the protocols for screening.

What Is Involved With The HHT Screening?

- Physical examination
- Pulse oximetry to determine if the blood oxygen levels are lower than normal
- MRI (with and without contrast) to determine any blood vessel abnormalities in the brain
- Echo Bubble cardiogram to determine if lung AVM's are present
- CT scan of the lungs to determine the size of lung AVM's if the echo bubble test is positive.
- Interview with the Medical Director to discuss test results and proposed treatment plan (estimated 60-90 minutes)

The entire screening process will be completed within a day, if possible. If additional family members are traveling and will be receiving screening, additional time will be required

What Does The HHT Screening Tell You?

The primary reason to screen a child for HHT is to see if she/he has the genetic disease. If the screening is positive, a nurse and physician will discuss treatment plans, management, and monitoring of HHT in the future.

Will There Be More Tests?

Each case is different, but the initial results from your child's HHT screening may lead to additional tests. Consult with the HHT Center of Excellence to know if you need to schedule any follow up appointments specific to the results.

What Should I Do After The Screening?

You need to keep a record of all results from the screening and contact your pediatrician to schedule a follow up appointment. Bring along the results as well as any information you received about HHT from the HHT Center of Excellence. Your pediatrician may wish to consult with the HHT Center Director so that they can coordinate care.

Monitor your child's health and contact your pediatrician or HHT Center if any of the following occur:

- Decreased exercise tolerance
- Shortness of breath
- Increased frequency and severity in nosebleeds
- Coughing up blood
- Change in neurological status or behavior

It is important to pay attention to your child's abilities during athletic activity. Shortness of breath with basic fitness tasks or sports could be due to low oxygen levels that are common with Lung AVMs.

Tips For Discussing HHT With Your Child's Nurse or Pediatrician

When you make the visit to your child's pediatrician and/or discuss HHT with your child's nurse, bring along the My Child has HHT: Screening Guidelines fact sheet so that the caregiver is aware of what to look out for with HHT.

It is essential to keep track of any prescribed treatments or medications. They may need to be included on a school medical form or an individualized health care form, which requires a doctor's signature.



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