MY CHILD'S HHT CARE CHECKLIST

USING THE HHT GUIDELINES

The HHT Pediatric Care Guidelines are detailed on the next pages

Date: _____

Child's Name: _____

Please check all that apply

☐ MY CHILD HAS A PARENT WITH HHT AND WE DON'T KNOW IF MY CHILD IS AFFECTED.

- ☐ Talk to my child's doctor about genetic testing for my child.

☐ MY CHILD HAS HHT OR MAY HAVE HHT (THE HHT DIAGNOSIS HAS NOT BEEN RULED OUT).

- ☐ Ask the doctor for screening for lung arteriovenous malformations (AVMs).
- ☐ Ask the doctor for screening (usually MRI) for brain vascular malformations (VMs).

☐ MY CHILD HAS LUNG (PULMONARY) AVMs.

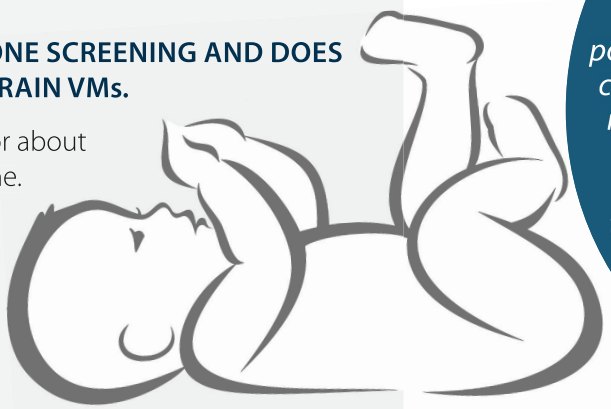
- ☐ Consider preventative treatment (embolization) for large AVMs or if my child has low oxygen levels.
- ☐ Talk to my child's dentist and other health care professionals about the need for lung AVM precautions, lifelong.
- ☐ Plan regular follow-up for the lung AVMs, whether or not they are treated.

☐ MY CHILD HAS BRAIN VMs.

- ☐ Ask for a referral to an expert multi-disciplinary neurovascular team for assessment of my child's risk of bleeding from the VM(s) and for consideration for treatment.

☐ MY CHILD HAS UNDERGONE SCREENING AND DOES NOT HAVE LUNG AVMs OR BRAIN VMs.

- ☐ Talk to my child's doctor about re-screening in 5 years' time.



INTERNATIONAL HHT GUIDELINES

WHAT ARE THE HHT GUIDELINES AND WHY ARE THEY IMPORTANT?

- The HHT Guidelines are recommendations for care based on evidence and expertise from HHT experts from around the world.
- The HHT Guidelines help ensure that people living with HHT get the best care possible.

WHAT IS MY ROLE AS SOMEONE LIVING WITH HHT?

- Be aware of the Guidelines. Share them with your care team. Ideally you should be seen at an HHT Center of Excellence or your care team may want to consult with one.
- Read up on your condition and know what care is available for HHT.
- **Prepare ahead of time for your appointments**: Bring your HHT Care Checklists and a family member or friend. They can help you communicate your questions and priorities, as well as act as a second set of ears. Share your experiences, worries and priorities to help your care team better understand your needs and provide individualized care.

PEDIATRIC CARE

While some manifestations of HHT, such as telangiectasia and epistaxis, are age dependent and may be absent in young children with HHT, potentially serious and even life-threatening complications of pulmonary 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.

HHT GUIDELINES RECOMMENDATIONS

PEDIATRIC CARE

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The expert panel recommends:

E1 THAT DIAGNOSTIC GENETIC TESTING BE OFFERED FOR ASYMPTOMATIC CHILDREN OF A PARENT WITH HHT.

Clinical Considerations: An affected family member should be tested first to determine the causative mutation, prior to testing an asymptomatic child who does not meet the clinical diagnostic criteria for HHT (Curaçao criteria). The benefits of testing, alternatives, pros and cons should be discussed with children or – as appropriate – their parents.

E2 SCREENING FOR PULMONARY AVMs IN ASYMPTOMATIC CHILDREN WITH HHT OR AT RISK FOR HHT AT THE TIME OF PRESENTATION / DIAGNOSIS.

Clinical Considerations: Screening may be performed with either chest X-ray and pulse oximetry OR transthoracic contrast echocardiography (TTCE). Screening with CT is not recommended, though CT chest remains the confirmatory diagnostic test when screening tests are positive.

E3 THAT LARGE PULMONARY AVMs AND PULMONARY AVMs ASSOCIATED WITH REDUCED OXYGEN SATURATION BE TREATED IN CHILDREN TO AVOID SERIOUS COMPLICATIONS.

Clinical Considerations: Pulmonary AVMs with feeding arteries ≥3 mm diameter are suitable for embolotherapy. Follow-up is indicated, to detect recanalization and reperfusion of treated AVMs and growth of small untreated AVMs. Specific protocols vary among centers (CT, oximetry or TTCE), as do intervals.

E4 REPEATING PULMONARY AVM SCREENING IN ASYMPTOMATIC CHILDREN WITH HHT OR AT RISK FOR HHT; TYPICALLY AT 5 YEAR INTERVALS.

Clinical Considerations: Typically, negative screening is repeated every 5 years. In children with indeterminate or borderline screening results, either based on imaging or oximetry, screening should be repeated sooner.

E5 SCREENING FOR BRAIN VM IN ASYMPTOMATIC CHILDREN WITH HHT, OR AT RISK FOR HHT, AT THE TIME OF PRESENTATION / DIAGNOSIS.

Clinical Considerations: First-line screening is MRI (contrast enhanced more sensitive) to identify brain VM and determine subtype and risk factors for hemorrhage. This typically requires sedation or anesthesia in young children. The decision to treat versus observe is based on risk of treatment versus risk of hemorrhage. As such, the decision to screen the child should be a shared decision among clinicians, caregivers and the child (where possible). There are important differences in clinical practice across countries: from screening asymptomatic children with MRI in infancy, to no routine screening of asymptomatic children for brain VM. Patient representatives felt strongly that children should be screened for brain VMs citing anecdotal evidence of disastrous outcomes in unscreened patients.

E6 THAT BRAIN VMS WITH HIGH RISK FEATURES BE TREATED.

Clinical Considerations: Given the need to balance natural history risk with treatment risk, children with HHT who have brain VM should be referred to a center with multidisciplinary expertise in neurovascular disease management. Treated brain VMs require close follow-up; the follow-up for small (untreated) brain VMs is not well defined.