Appendix 1. Routine Screening Protocols for All Patients at Toronto HHT Centre

All patients assessed for suspected hereditary hemorrhagic telangiectasia (HHT) at the Toronto HHT Centre are screened for pulmonary arteriovenous malformations (AVMs) (contrast echocardiography, followed by CT thorax in positive cases) and treated preventatively, as per international HHT Guidelines.[1] All patients are also screened for cerebral AVMs with cerebral magnetic resonance imaging (MRI), and treated preventatively based on consultation with the Toronto HHT Centre neurovascular expert team. If patients have an iron deficiency anemia out of keeping with the severity or frequency of their epistaxis, or if they have melenic stools or blood in their stools, screening for gastrointestinal (GI) telangiectases by upper GI endoscopy is performed. Patients are considered to have HHT-related GI bleeding if they have both iron-deficiency anemia *and* GI telangiectases.

Patients are not routinely screened for spinal AVMs or liver vascular malformations (VMs). When patients are assessed pre-pregnancy, they are offered a prepregnancy lumbo-sacral MRI to screen for spinal AVM. Patients are investigated for liver VMs (mesenteric Doppler ultrasound) if they have symptoms of liver VMs, hepatic bruit, or abnormal liver function tests.

Reference

1. Faughnan ME, Palda VA, Garcia-Tsao G, Geisthoff UW, McDonald J, Proctor DD, et al. International Guidelines for the Diagnosis and Management of Hereditary Hemorrhagic Telangiectasia. J Med Genet 2009; Jun 29.

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The authors provided this information as a supplement to their article.

