PHYSICIAN INFORMATION

Diagnosis and Management of Brain AVMs

AVMs, short for arteriovenous malformations, are abnormal vessels with direct artery to vein connections. At least 10% of people with HHT have brain AVMs. People are often unaware that they have cerebral AVMs until they develop a life-threatening complication, such as stroke or seizure. With the right screening and treatment, these life-threatening complications can be prevented, however, there is no single 'standard treatment' that can be recommended for all brain AVMs in HHT at the current time.

Please review the checklist below with your physician to make sure your HHT is being treated properly.

HHT GUIDELINES RECOMMENDATION	COMPLETED
Screen adult patients with possible or definite HHT for cerebral VMs*. Level of Evidence: III GRADE Strength of Recommendation: Weak Expert Agreement: 77%	❑ YES ❑ NO
Use MRI for cerebral VM screening in adults with possible or definite HHT, using a protocol with and without contrast administration and using sequences that detect blood products, to maximize sensitivity. Level of evidence: III GRADE1 Strength of recommendation: Weak Expert Agreement: 100%	❑ YES ❑ NO
Screen children with possible or definite HHT for cerebral VMs in the first 6 months of life (or at time of diagnosis) with an unenhanced MRI, and refer all patients with an MRI positive for these lesions to a center with neurovascular expertise for consideration of invasive testing and further management. Level of evidence: III GRADE Strength of recommendation: Weak Expert Agreement: 64%	❑ YES ❑ NO
Adults presenting with an acute hemorrhage secondary to a cerebral VM should be considered for definitive treatment in a center with neurovascular expertise. Level of evidence: III GRADE Strength of recommendation: Strong Expert Agreement: 94%	❑ YES ❑ NO
All other adults diagnosed with cerebral VMs should be referred to a center with neurovascular expertise to be considered for invasive testing and individualized management. Level of evidence: II GRADE Strength of Recommendation: Strong Expert Agreement: 100%	❑ YES ❑ NO
Pregnant women with suspected or confirmed HHT harboring an asymptomatic cerebral AVM during pregnancy should have definitive treatment of their cerebral AVM deferred until after delivery of their fetus. The delivery of the fetus should follow obstetrical principles. Level of evidence: III GRADE Strength of Recommendation: Weak Expert Agreement:80%	❑ YES ❑ NO

*The term cerebral vascular malformations (VM's) includes AVM's and other similar vascular lesions in the brain

1 Schunemann HJ, Jaeschke R, Cook DJ, Bria WF, El-Solh AA, Ernst A, Fahy BF,Gould MK, Horan KL, Krishnan JA, Manthous CA, Maurer JR, McNicholas WT, Oxman AD, Rubenfeld G, Turino GM, Guyatt G. An official ATS statement: grading the quality of evidence and strength of recommendations in ATS guidelines and recommendations. Am J Respir Crit Care Med 2006: 174(5): 605-614.

International Guidelines for the Diagnosis and Management of Hereditary Hemorrhagic Telangiectasia

J Med Genet. 2009 June 29. [Epub ahead of print]

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Abstract

HHT is an autosomal dominant disease with an estimated prevalence of at least 1/5000 which can frequently be complicated by the presence of clinically significant arteriovenous malformations in the brain, lung, gastrointestinal tract and liver. HHT is under-diagnosed and families may be unaware of the available screening and treatment, leading to unnecessary stroke and life-threatening hemorrhage in children and adults. The goal of this international HHT guidelines process was to develop evidence-informed consensus guidelines regarding the diagnosis of HHT and the prevention of HHT-related complications and treatment of symptomatic disease. The overall guidelines process was developed using the AGREE framework, using a systematic search strategy and literature retrieval with incorporation of expert evidence in a structured consensus process where published literature was lacking. The Guidelines Working Group included experts (clinical and genetic) from eleven countries, in all aspects of HHT, guidelines methodologists, health care workers, health care administrators, HHT clinic staff, medical trainees, patient advocacy representatives and patients with HHT. The Working Group determined clinically relevant questions during the pre-conference process. The literature search was conducted using the OVID MEDLINE database, from 1966 to October 2006. The Working Group subsequently convened at the Guidelines Conference to partake in a structured consensus process using the evidence tables generated from the systematic searches. The outcome of the conference was the generation of 33 recommendations for the diagnosis and management of HHT, with at least 80% agreement amongst the expert panel for 30 of the 33 recommendations.

Open Access Link to International HHT Clinical Guidelines:
http://jmg.bmj.com/content/early/2009/06/29/jmg.2009.069013.long