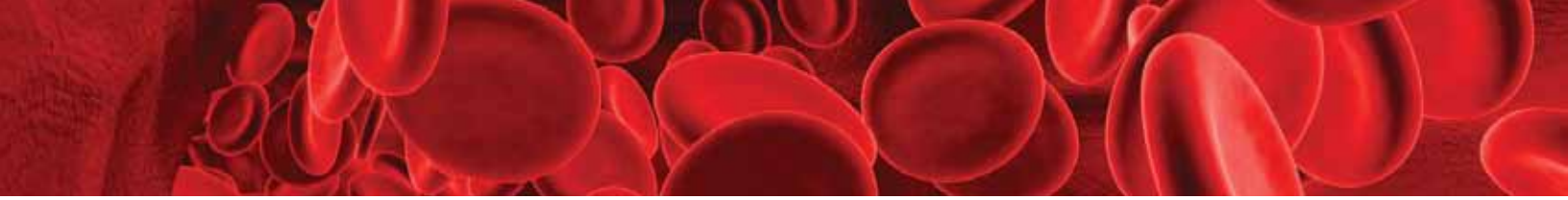

Hereditary Hemorrhagic Telangiectasia
Foundation International, Inc.

OSLER - WEBER - RENDU SYNDROME

HHT QUICK FACTS FOR HEALTH CARE PROFESSIONALS

- Hereditary Hemorrhagic Telangiectasia (also known as Osler-Weber-Rendu Syndrome) is a multi-system vascular dysplasia. It is uncommon, but not rare. Approximately 1.4 million people worldwide have HHT.
- Telangiectases and arteriovenous malformations (AVMs) are the characteristic lesions.
- Location of lesions and severity of symptoms is highly variable and is significantly under diagnosed in affected individuals.
- Most commonly affected organs are the nose, lungs, GI tract, brain and liver.
- HHT is an autosomal dominant genetic disorder. Denovo mutations are rare. A targeted family history shows almost all cases to be familial.
- Defects in at least 3 different genes can cause HHT.
- 90-95% of individuals with HHT will develop epistaxis by adulthood, but severity varies from infrequent and minor to daily and severe.
- 90-95% develop at least a few small telangiectasia on the face and/or hands by middle age.
- 20-25% develop significant gastric or intestinal bleeding, but rarely before 50 unless affected with juvenile polyposis in conjunction with HHT.
- 40% have lung AVMs. These pose significant risk.
- 5-20% have at least one brain AVM. These can be present at any time of life, even at birth, and can cause serious complications. Routine screening is recommended in North American centers.
- Liver AVM are relatively common, but only approximately 10% cause symptoms.
- The severity of epistaxis or skin telangiectases does not correlate with the likelihood of having brain or lung AVMs.

Lung AVMs –The following precautions are recommended for all HHT patients with lung AVMs (treated and untreated), as well as those not yet screened by an HHT Center of Excellence

- **Antibiotic Prophylaxis** is recommended for dental and other procedures that can cause bacteria in the blood.
- **IV Filter** - An IV air filter (bubble trap) should be used when a patient has an intravenous line. This is to prevent any large air bubble from entering the bloodstream, going through a lung AVM, and then causing a temporary stroke. This is most effectively done by using a filter in the IV line as close to the patient as possible. A 0.22 micron filter is best if available, but a blood filter is also acceptable (about 260 microns) and will stop all large air bubbles. During a blood transfusion, a standard blood filter is all that is needed. Please note that filters often cannot be used for IV contrast injections like you might get for CT or MRI scans.

Brain AVMs - Untreated brain AVMs are a common cause of hemorrhagic stroke in HHT families.

Epistaxis Severity Scoring Tool (ESS) - This is a way to monitor the severity of a patients nosebleeds and their response to treatment. This is a simple score that is calculated automatically when the patient answers 6 simple questions about their nosebleeds. The ESS tool can be accessed through the HHT Foundation website, www.hht.org.

Molecular Genetic Testing for HHT is now available in a handful of labs worldwide. Genetic testing is best coordinated by a medical geneticist or genetic counselor. Testing an individual suspected to have HHT, without first testing a clinically affected family member is rarely informative.

Diagnosis and treatment for HHT has changed significantly in the last decade. Recommended screening and treatment (if, when, and how) is dramatically different for pulmonary vs. cerebral vs. hepatic AVMs. The HHT Foundation recommends that people with HHT be assessed at least once at a specialized HHT Center.

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PHYSICIAN INFORMATION

International Guidelines for the Diagnosis and Management of Hereditary Hemorrhagic Telangiectasia

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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

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The HHT Foundation International, Inc. was formed to aid and support families with the genetic disorder Heriditary Hemorrhagic Telangiectasia (Osler-Weber-rendu Syndrome).