melbourne haematology

fact sheet

Hereditary Haemorrhagic Telangiectasia (HHT)

Osler Weber Rendu syndrome

What is HHT?

HHT is an uncommon inherited (genetic) disorder characterised by abnormal connections between arteries and veins — called “vascular malformations” or AVMs.

What causes HHT?

HHT is a genetic disorder caused by a change in the genes that control the development of blood vessels. We inherit half our genes from each parent; when a gene change occurs, there is potential for health problems to result.

HHT is an autosomal dominant disorder — meaning you only need one copy of the abnormal gene (from one parent) to be at risk of developing AVMs.

What are AVMs?

AVMs occur when an artery develops an abnormal direct connection to a vein. Normally, blood travels from the heart through arteries, then into very small vessels called capillaries, before draining into veins that return blood to the heart. An AVM occurs when blood travels straight from an artery to a vein, bypassing the capillaries.

What are the problems with AVMs?

The abnormal connection between artery and vein is often fragile and can rupture, leading to bleeding into the tissues. Symptoms depend on where in the body the AVM occurs. AVMs can occur in the skin (called telangiectasia), the lining of the nose (causing severe nosebleeds), the brain (causing strokes), and the lungs (causing bleeding into the lungs). Less commonly, AVMs occur in the liver and gastrointestinal tract.

How is HHT and AVMs diagnosed?

HHT can be diagnosed by taking a careful family history to identify relatives who may have had HHT, by close clinical examination for AVMs, and by specialised radiology or imaging studies to check for AVMs in body organs. A specialised genetic test can also identify the gene changes responsible for HHT.

The recommended screening approach for each type of AVM is summarised below.

Site of AVM	Screening Procedure	Notes
Brain AVM	MRI	Approximately 25% of patients with HHT will develop brain AVMs. Performed at least once; may be deferred until a child can tolerate MRI without general anaesthetic.
Lung AVM	Contrast (bubble) echo	Occurs in 15–50% of adult patients. Needs to be repeated every 5 years.
Liver AVM	Abdominal ultrasound	Liver AVMs occur in up to 78% of patients, but only a small percentage develop symptoms.
Gastrointestinal AVM	Serum ferritin or endoscopy	May occur in the majority of patients with HHT, though a smaller percentage develop bleeding

		symptoms. Serum ferritin should be assessed annually.

I have HHT and nosebleeds — what should I do?

Managing nosebleeds (epistaxis) in patients with HHT can be difficult. Epistaxis is very common in HHT, with approximately 50% of patients experiencing it as a child or adolescent, due to abnormally fragile vessels in the nasal lining. Treatments include keeping the vessels moist (humidification or lubricant), minor surgical procedures to remove the vessels (e.g. cauterisation), or applying a skin graft to the area (septodermoplasty). It's important that the doctors managing nosebleeds are familiar with HHT, since some procedures can worsen bleeding.

Genetic diagnosis

The aim of genetic diagnosis is to identify the specific gene change in the family. This involves sequencing the ENG/HHT1 and ACVRL1/HHT2 genes. There are no common mutations — each family typically has a “private” HHT mutation. Approximately 75% of patients with clinical HHT will have a mutation identified through sophisticated gene testing. Gene testing can be expensive; it's best to discuss this with your doctor.

References

1. Faughnan ME, Palda VA, Garcia-Tsao G, et al. International guidelines for the diagnosis and management of hereditary haemorrhagic telangiectasia. Journal of Medical Genetics. Feb 2011;48(2):73-87.

2. Govani FS, Shovlin CL. Hereditary haemorrhagic telangiectasia: a clinical and scientific review. European Journal of Human Genetics. Jul 2009;17(7):860-871.

Further Questions?

The information presented in this fact sheet is intended as a general guide only.

Patients should seek further advice and information about **Hereditary Haemorrhagic Telangiectasia (HHT)** and their individual condition from their treating haematologist or doctor.

For additional information about blood disorders and their treatment, or to contact one of our specialist haematologists, visit the Melbourne Haematology website:

www.melbournehaematology.com.au