

Hereditary Haemorrhagic Telangiectasia (HHT, Osler Weber Rendu syndrome)

What is Hereditary Haemorrhagic Telangiectasia or HHT?

HHT is a uncommon inherited (genetic) disorder which is characterised by abnormal connections between arteries and veins – these are called “vascular malformations” or AVMs. .

What causes HHT?

HHT is a genetic disorder – a change in the genes causes HHT. We inherit half of our genes from each of our parents. When a gene change occurs, there is the potential that this can cause problems with health. HHT occurs when there is a change in the genes that control the development of blood vessels.

HHT is an autosomal dominant disorder – this means you only need one copy of the abnormal gene (from one parent) to develop HHT putting you at risk of developing AVMs.

What are AVM's?

AVMs occur when an artery develops an abnormal connection straight to a vein. Normally blood travels from the heart in arteries and then gets to body organs and tissues by dividing into very small blood vessels called capillaries. The capillaries then drain into veins which carry the blood back to the heart. An AVM occurs when the blood travels straight from the arteries to the vein.

What are the problems with AVM's?

The abnormal connection between the artery and the vein is often very fragile and can rupture or burst leading to bleeding into the tissues. The type of symptoms that a ruptured AVM causes will depend on where the AVM occurs in the body. AVMs can occur in the skin (called telangiectasia), in the lining of the nose (can cause severe blood noses), in the brain (cause strokes) and in the lungs (cause bleeding into the lungs). Less commonly, AVMs can occur in the liver and the 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 examination to see if patients have AVMs and by specialised radiology or imaging studies to see if AVMs are present in organs in the body. There is also specialised genetic test that can be performed to identify the gene changes that are responsible for HHT.

The type of specialised radiology tests for each of the different types of AVMs is listed below.

Site of AVM	Screen procedure	Notes
1. Brain AVM	MRI	Approximately 25% of patients with HHT will develop brain AVMs. Performed at least once. This may be deferred until a child can tolerate MRI without general anaesthetic.
2. Lung AVM	Contrast (Bubble) ECHO	Occur in 15 – 50% of adult patients Need to repeat every 5 years
3. Liver AVM	Ultrasound Abdomen	Liver AVMs occur in up to 78% of patients but only a small percentage of patients will develop symptoms.
4. Gastro-intestinal AVM	Serum ferritin or endoscopy	May occur in majority of patients with HHT but smaller percentage develop symptoms related to bleeding Serum ferritin should be assessed on an annual basis.

I have HHT and blood noses – what should I do?

The management of blood noses (epistaxis) in patients with HHT can be difficult. Epistaxis is very common in HHT with approximately 50% of patients with HHT experiencing epistaxis as a child / adolescent. Epistaxis is common in HHT due to the presence of abnormally fragile vessels in lining of the nose. A number of treatments are available including keeping these blood vessels moist (e.g. humidification or applying a lubricant), minor surgical procedures to remove the blood vessels (e.g. cauterisation) or even applying a skin graft to the area to stop the bleeding (septodermoplasty). It is important that the doctors dealing with the blood noses are familiar with HHT since some procedures can lead to more bleeding.

Genetic Diagnosis

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

Resources used to produce this information sheet.

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.

Govani FS, Shovlin CL. Hereditary haemorrhagic telangiectasia: a clinical and scientific review. *European journal of human genetics : EJHG*. 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 **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