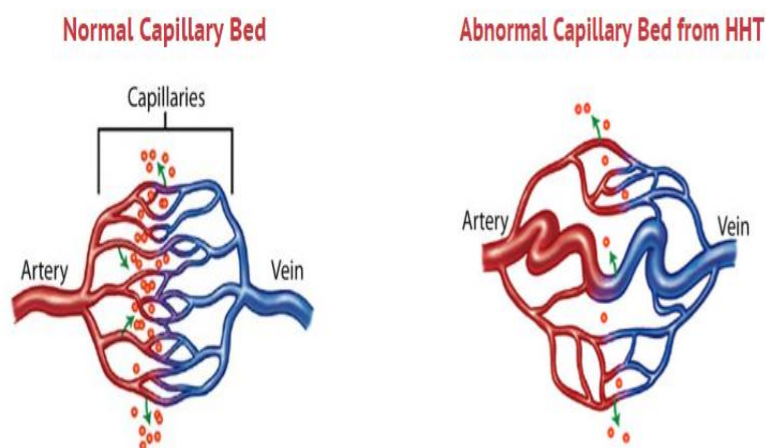


Hereditary Haemorrhagic Telangiectasia (HHT)

Information for patients

Hereditary Haemorrhagic Telangiectasia (HHT) is a hereditary disorder that is characterised by abnormal blood vessel formation. A person with HHT has a tendency to form blood vessels that lack normal capillaries between an artery and vein.



What Causes HHT?

HHT is caused by a change (called a mutation) in one of several HHT associated genes.

How is it inherited?

HHT is an autosomal dominant genetic disorder. This means that if a parent has HHT, no matter their gender, each of their children will have a 50% chance of having HHT.

Nearly everyone with HHT will have inherited the disorder from a parent who also has HHT.

It affects 1 in 5,000 people resulting in around 1,000 people in Scotland having the condition.

Signs and Symptoms

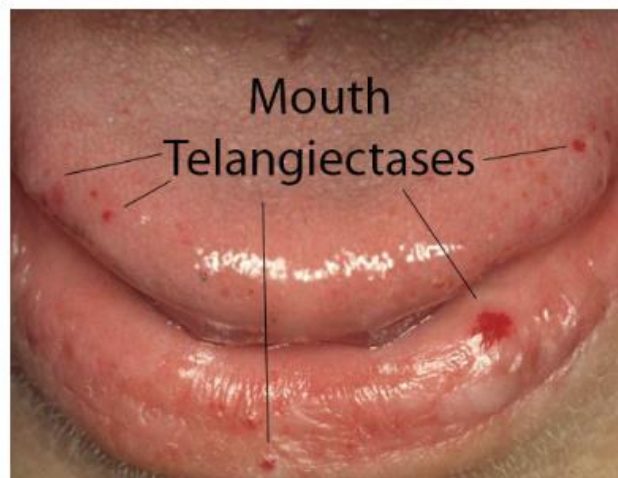
There are many times that a person with HHT may appear to have no symptoms at all.

Patients may show almost no signs of the disease up until their fourth or fifth decade of life. HHT affects everyone differently and will even affect members in the same family very differently.

The most common symptom that HHT patients experience is nosebleeds. It can however affect many different parts of the body. The two types of vascular abnormalities that characterise HHT are telangiectasia and arteriovenous malformations (AVMs).

Telangiectasia

Telangiectasias are small vascular malformations that appear as small red spots on the surface of the skin (especially the hands, face and mouth), the lining of the nose and the lining of the gut.



Nose

Most people with HHT have telangiectasia on the inside of their nose, which cause nosebleeds (epistaxis) when ruptured.

- Recurring nosebleeds affect about 90% of people.
- They can vary from a social nuisance to severe, sometimes causing lots of blood loss and even multiple blood transfusions.

Gastrointestinal

- 80% of HHT patients have telangiectasia in the stomach/ intestines.
- 20% of these will have a bleed.

Patients are only offered endoscopies if they are having bleeds.

Arteriovenous malformations (AVMs)

Arteriovenous malformations (AVMs) are large vascular malformations that can occur in the liver, lungs, brain, and sometimes the spine. Unlike telangiectasia, AVMs cannot be seen without proper imaging procedures.

Lung AVMs

40 – 50% of people with HHT have lung AVMs (also known as pulmonary AVMs, or PAVMs). Lung AVMs can lead to serious complications, such as stroke or brain abscesses, so it is important to be screened.

Brain (Cerebral) AVMs

20% of patients have brain AVMs. The evidence around screening is controversial and therefore each unit may vary in their protocol. In Dundee we tend to offer screening to all patients after a discussion of the pros and cons.

Liver (Hepatic) AVMs

40% of patients have liver AVMs with only 8% being symptomatic. We tend not to screen routinely unless patients have symptoms (increasing shortness of breath, swelling, abdominal pain, jaundice).

Testing

A person can be diagnosed with HHT through a clinical examination, genetic testing, or both.

- **A positive genetic test would confirm that someone does have HHT.**
- **A negative test does not rule out HHT**, 15% of people with HHT have mutations in genes that are not yet identified HHT genes.

Genetic Testing

DNA (genetic material carried in cells) is extracted from blood or saliva and analysed to identify a change (mutation) in one of the HHT genes.

It is important to get tested to allow for the correct screening and advice regarding other family members who may be at risk of having the disease.

Screening

Lung AVMs

If a pregnant patient has not had a recent evaluation for PAVM, it is very important for them to do so as soon as pregnancy is recognised.

AVMs larger than a certain size should be treated to prevent complications. If an AVM has been diagnosed or a patient is awaiting screening then antibiotics prior to most surgical procedures including dental work and colonoscopies is recommended.

- **Until PAVMs are excluded by testing, a person with known or suspected HHT should take antibiotics before all dental cleanings, dental work and other invasive medical procedures.**

Contrast Echocardiogram (bubble echo) or CTPA are the recommended tests for PAVM. Further testing may be needed.

Cerebral Screening

The evidence that active treatment is needed is debated but we discuss and offer screening if the patients request. This is often in the form of an MRI scan.

Children

It is important that children who may have the disease are assessed by a trained professional with experience in dealing with HHT. The time and extent of the screening programme is debated in children and this should be discussed with your doctor.

Nosebleed Treatments

90% of individuals affected by HHT will develop chronic nosebleeds. [The Epistaxis Severity Score \(ESS\)](#) is a useful tool for tracking nosebleeds to help determine whether treatment is necessary.

Keeping the nose humidified is important and topical treatments such as Vaseline and ointments given by your doctor can be useful.

Medical treatments can also include oral medication such as tranexamic acid and tamoxifen.

Coagulation therapy (either chemical or with electrical diathermy) or laser treatment makes the affected blood vessels shrink causing vessel destruction so that they are less likely to bleed. This is a quick and effective way to stop nosebleeds, although the procedure usually needs to be repeated periodically.

Further surgery is typically only considered when nosebleeds are so severe that a patient's quality of life is affected and when other therapies have failed. These will be discussed fully with you should this be necessary.

AVM Treatments

Not all AVMs require treatment. If your doctor decides that you need to treat your AVMs, there are several treatment options available.

The recommended treatment for AVMs depends almost entirely on the size of the malformation and its location in the body.

Iron Deficiency

People with HHT who suffer from bleeding should be routinely screened for iron deficiency. It is important that iron deficiency be treated aggressively, by replacing the iron stores in the body. There are multiple methods of replacing iron stores.

Dietary

- Consume iron with a source of vitamin C to increase absorption. Many fruits, vegetables, and juices are good sources of vitamin C, including citrus fruits and juices, cantaloupe, strawberries, broccoli, peppers and tomatoes.
- Meat has iron and also increases the absorption of iron from plants, for example, beef in chilli will help you absorb the iron in kidney beans.

Oral Replacement

Iron stores can be replaced by taking iron by mouth in the form of iron supplements. In individuals with severe iron deficiency anaemia or frequent/severe bleeding, oral iron may not be enough to restore iron stores. In these cases, it is important that treatment with intravenous iron be considered.

HHT Patient Advice

Nosebleed Advice:

Nosebleed (epistaxis) is the most common symptom of HHT patients. It is important to be aware of things to avoid increasing this risk and also some first aid advice as well as what to do if the bleeding doesn't stop.

If you are having regular nose bleeds try to avoid:

- Nose blowing
- Avoid anything you may be allergic to
- If you are getting increasing frequency or severity of bleeds it may help limiting the below food and drinks as they can increase the risk of bleeding:
 - Hot drinks
 - Alcohol
 - Spicy foods
- Avoid heavy lifting
- Avoid straining, this includes on the toilet. If you are constipated, consider taking a laxative to help or discuss with your GP.

First aid instructions:

- Sit up straight with your head forward; try to avoid swallowing any blood.
- Pinch your nose just below the bones in the middle of the nose and hold for 10 – 15 minutes.
- If the bleeding doesn't stop you need to seek medical advice at your local accident and emergency.
- If possible we want to avoid packing the nose with a non-absorbable pack however in an emergency these may need to be used.
- If there are any issues when in acute care they can contact the ENT registrar on call at Ninewells Hospital on bleep 4496.
- If your bleeding has stopped but you feel the frequency and severity are such that you need to be seen by ENT, contact us via the ENT secretaries (01382 496571) or Ward 26 on 01382 633286

Dental/Procedure Prophylaxis

In 2008 NICE produced guidance regarding prevention of heart valve infections after certain "dirty procedures".

This has caused some confusion with antibiotic prophylaxis regarding HHT.

The main reason for giving patients with HHT antibiotic prophylaxis is that if they have a PAVM they are 500 times more likely to get a brain abscess.

Basically, there are two categories of HHT patients with regards to needing antibiotics prior to any “dirty procedures”, like dental work.

1. **Do not need** prophylactic (preventive) antibiotics if your lungs have been screened and you do not have any PAVMs.
2. **Need** prophylactic antibiotics if you have PAVMs **or** you aren't sure if you have PAVMs. If you haven't been screened within 5 years then we would recommend antibiotics.

Antibiotic of Choice

- Co-amoxiclav 625mg TDS for 7 days, if allergic then Clindamycin.

Pregnancy and HHT

The vast majority of the time patients who are pregnant with HHT will progress through pregnancy and labour without a complication. However there are small but significant and potentially life threatening risks to be aware of and pregnant patients need to be counselled appropriately regarding these risks.

Pregnant patients are no more likely to bleed excessively at or after delivery than those in the general population, as the uterus is not a typical place for an AVM to occur. There appears to be a risk of PAVM bleeding during labour, but this hasn't been shown to be the case with cerebral AVMs.

Shovlin et al published the largest study looking at these risks:

Based on this study the recommendations for management of pregnant patients with HHT:

- All patients with HHT considering pregnancy should be screened for pulmonary AVMs.
- If pulmonary AVMs are found, those patients should be treated prior to conception.
- If pregnant and found to have asymptomatic pulmonary AVMs, they should not be treated during pregnancy.
- All pregnant patients should have an MRI of the spine to rule out spinal AVM, this would enable regional anaesthesia (such as an epidural) (No case report of HHT and spinal complication, but there have been reports of other spinal AVM and paraplegia).
- If a family history of stroke or central nervous system symptoms, an MRI of the head is recommended.
- If any pregnant patient with HHT starts to cough up blood or sudden shortness of breath, then urgent hospitalisation is encouraged.
- For all pregnant patients with HHT, antibiotic prophylaxis during delivery is recommended.
- Avoidance of a prolonged stage of labour in HHT patients in whom a brain AVM has not been excluded is advised.

Developed by ENT Registrar and reviewed by ENT Surgeons

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