

Patient information on Hereditary Haemorrhagic Telangiectasia (HHT)

Who is this information for?

This information is for patients, families and carers with hereditary haemorrhagic telangiectasia (HHT).

What is hereditary haemorrhagic telangiectasia (HHT)?

- HHT is a genetic (inherited) disease, also known as Osler-Weber-Rendu syndrome, in which abnormal blood vessels (telangiectasia) may develop.
- It can cause some people considerable distress while others escape with less severe problems.
- For the majority of patients, nosebleeds are the main problem.
- Some people with HHT may develop abnormal blood vessels in their lungs (pulmonary arteriovenous malformations (PAVMs)) or brain (cerebral AVMs (CAVMs)). These may cause complications so screening tests are recommended. If AVMs are found then treatment options will be discussed with you.
- In about 20% of cases, bleeding from the lining of the stomach or intestines may occur (usually later in life).

How do I know if I have HHT?

- HHT does not usually cause problems in early life.
- The first signs of HHT in a child may be that nosebleeds are more frequent and heavier than those of other children; this may not happen until after the age of 10.
- Half of patients with HHT have frequent nosebleeds by the age of 16, and over 90% have regular nose bleeds by the age of 30.
- Telangiectasia may appear as red spots on the face, lips, tongue and finger tips.

Will I need any screening tests?

- **PAVM** screening is important for anyone with HHT – it is either done with a contrast transthoracic echocardiogram (CTTE, “bubble test”) or a CT scan of the chest. Screening in adults with HHT is recommended every **5** years. In children, after initial screening, screening should not occur again until after puberty.
- Prior to pregnancy, it is recommended that PAVM screening should occur as some women with PAVMs have complications during pregnancy (*refer to HHT and Pregnancy patient information sheet*).
- **CAVM** screening is usually done once as an adult (and sometimes as a child), with an MRI scan of the brain.

- Screening for stomach or intestinal lesions, with endoscopy, is usually only done if you have symptoms of bleeding.
- If you have PAVMs, even if they have been treated, you will be advised to take antibiotics prior to any dental treatment or surgery.

How does HHT affect the nose and what management options are available?

- Patients with HHT often have frequent and severe nosebleeds (*refer to Epistaxis patient information sheet*).
- Medical and surgical management options are available - these depend on the patient and the severity of the problem.

What are the medical management options available for nosebleeds in HHT?

- Topical treatment: applying **Vaseline** or antibiotic creams into the front of the nose can be helpful.
- Hormone treatment: **oestrogen** and **progesterone** have both been used in the past to reduce the frequency and severity of nosebleeds. High doses are required and side effects were often not acceptable.
- **Tamoxifen** is a newer hormonal therapy with reduced side effects and improved symptom control (*refer to HHT & Tamoxifen patient information sheet*).
- **Bevacizumab (Avastin®)** is an antiangiogenic drug that reduces abnormal blood vessel formation. It has been used for severe complications of HHT rather than nosebleeds, but is starting to be looked at as a possible treatment for nosebleeds by injecting or spraying it into the nose, and rarely by administering it into the blood-stream (intravenous).

What are the surgical options available for nosebleeds in HHT?

- **Cautery:** while this is used successfully for “normal” nosebleeds, it is not recommended in HHT as it has low success rates and repeated use can cause a septal perforation (hole in the septum of the nose).
- **Laser treatment:** aims to reduce the frequency and severity of nosebleeds by sealing off the individual telangiectasias within the nose. It is mainly useful for patients with mild to moderate disease but is not always successful. It requires a general anaesthetic, but can also be performed under local anaesthetic for minor disease. Repeat treatment is often needed every few months. Lasers which are absorbed preferentially by blood vessels are often employed – *e.g. pKTP, Blue-light Laser*.
- **Septodermoplasty:** telangiectasias are most frequent at the front of the nose, particularly on the nasal septum. It is possible to remove part of the lining over the septum and replace it with a skin graft from the thigh. The worst side is usually operated on first, and the other side can be done 3 months later if wished. Again, it only aims to reduce the frequency and severity of bleeding but is often helpful when laser does not work very well. The length of benefit varies from patient to patient. Over time, nasal bleeding may return as the graft shrinks and the telangiectasia recur around it; laser treatment can help this.
- **Closure of the nose (Young’s procedure):** this is the only treatment that can stop nosebleeds completely. Breathing through the nose can traumatise the fragile lining of the

nose; if air no longer enters the nose, the bleeding will stop. Closing the nose is done by creating little skin flaps inside the nose, which are not visible from the outside. Patients can safely breathe through their mouth and do not require the nose to breathe. If a nosebleed did occur when the nose is closed, blood would run into the mouth but this is very rare and usually suggests a little hole has opened up again at the front of the nose. The operation is usually done under a short general anaesthetic.

What is the risk of passing HHT onto my children?

- HHT is inherited in an autosomal dominant manner – this means that if one parent has HHT, any child has a 50:50 chance of inheriting the condition.
- Many of the genetic mutations can now be identified, meaning that family members including children can be screened using a blood test.
- This should be done at a specialist HHT centre – in Melbourne, Australia this is at **the Genetics department** at the **Royal Melbourne Hospital**.

Concerns or questions?

You can contact your ENT Specialist at the Melbourne ENT Group (MEG):

- Phone: 1300- 952-808
- Email: admin@melbentgroup.com.au
- Website: www.melbentgroup.com.au



Your GP is also the best contact for ongoing care and concerns.

Further information

Further information about HHT can be obtained from the Australian HHT Alliance website www.hht.org.au or from the Cure HHT (previously the HHT Foundation) website www.hht.org

www.hht.org.au



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